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52nd Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, 2025

12–14 November 2025, Sheffield

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CME Training Day Sessions

CME Symposium 1**CME1.1****Recent advances in the management of pituitary adenomas**

Marta Korbonits

Barts, London, United Kingdom

There is increasing recognition of pituitary adenomas, mostly due to increased imaging, a trend seen both in adults and children. Recently, the first guideline has been published on paediatric pituitary adenomas outlining diagnostic pathways and management. Key messages include emphasis on early diagnosis, care in centres of excellence in pituitary adenomas, the importance of discussion with 'adult' endocrinologist colleagues, genetic testing, novel imaging modalities, novel radiotherapy options, indications and issues with access to medications. For paediatric prolactinomas, the most common tumour type, the key discussion points are the importance of medical treatment, considering long-term side effects, genetics and ruling out alternative causes of hyperprolactinaemia (drug, dopamine pathway abnormality). For GH excess, early diagnosis can be hindered by differentiation from constitutional tall stature and other pathological causes of tall stature. Access to medication (GH receptor antagonist, 2nd generation somatostatin analogue) and prevention of long-term debilitation consequences are key. Genetics plays the largest role in this type of paediatric pituitary adenoma and family history as well as associated other manifestations are key for the correct diagnosis. Follow up of gene carrier children with a disease associated with low penetrance has the advantage of picking up disease early; however, it has its own challenges generating anxiety and screening fatigue. In paediatric Cushing's disease, high quality imaging (dynamic, volumetric), potentially including novel methods, access to experienced pituitary surgeon and rarely genetic aspects (germline MEN1, somatic USP8) are current hot topics. Small non-functioning lesions are often stable, but we suggest following them and handover to adult services for potential discharge later. Non-functioning macroadenomas in children usually need to be operated eventually if show growth or hormonal/visual complications. TSHomas are rare, can occur both in micro and macroadenomas, and thyroid hormone resistance represents the main differential diagnostic problem. The nationwide/international monthly case presentation meeting Hypothalamic Pituitary Axis Tumours (HPAT) meetings is extremely successful providing expert opinion and management support for these often rare and complex conditions and we strongly encourage all colleagues to present paediatric pituitary adenoma cases here.

DOI: 10.1530/endoabs.111.CME1.1

CME1.2**An approach to endocrine late effects**

Hoong-Wei Gan

Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom

Childhood cancer survivorship is increasing year on year, having more than doubled over the last 50 years, with a continually accruing cohort of currently >40000 people in the UK alone having survived a paediatric cancer. Despite mainstream media's common portrayal of "beating cancer", many patients in fact face long-term chronic morbidity, including >50% having at least one endocrinopathy. Whilst these are often mislabelled as "late effects", many of these evolve from diagnosis. The presumption that endocrine dysfunction is easily treatable is debatable, particularly in the context of the hypothalamic syndrome. Newer evolving immunological and molecular therapies also bring new challenges with the need for monitoring of off-target endocrine side effects. As such, the paediatric endocrinologist should be involved as an integral part of the oncology MDT from diagnosis, and transition to appropriate adult endocrinology services for lifelong follow-up is eventually needed for many of these patients.

DOI: 10.1530/endoabs.111.CME1.2

CME Symposium 2**CME2.1**

Abstract Unavailable

DOI: 10.1530/endoabs.111.CME2.1

CME2.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.CME2.2

CME Symposium 3**CME3.1**

Abstract Unavailable

DOI: 10.1530/endoabs.111.CME3.1

CME3.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.CME3.2

CME Symposium 4**CME4.1**

Abstract Unavailable

DOI: 10.1530/endoabs.111.CME4.1

CME4.2**Talking to children and young people about sex development and gender identity**

Julie Alderson

Bristol Royal Hospital for Children, Bristol, United Kingdom

When a child becomes aware that their body is different or not as expected they will make some sense or personal understanding of it. Good psychological adaptation is being able to make sense of oneself as a valuable person, of interest to others, likely to contribute and belong. In adolescence they might ask who am I, what can I do, how do people see me, and what is my life for? In Difference in Sex Development services we try to support our patients to understand their body, know their mind and appreciate that mind and body are not separate. This presentation uses child development theories to help guide conversations with children, and how to understand what they might feel and say as they develop cognitively.

DOI: 10.1530/endoabs.111.CME4.2

Plenary

PL1

Navigating the role of paediatric endocrinology in young people's gender services

Talat Mushtaq

Leeds Teaching Hospitals, Leeds, United Kingdom

This presentation will outline the changing nature of young people's gender services in the UK and consider how the medical role within these services may develop in the future. It will reflect on evolving service models, the emerging research framework, and the ongoing uncertainties surrounding best practice.

DOI: 10.1530/endoabs.111.PL1

Symposia

CEW Symposium**CS1.1****Current and future pharmacological therapy in weight management**

Neil Wright

Sheffield Children's Hospital, Sheffield, United Kingdom

The talk will cover the historical context of medications used to support weight loss discussing their side effect profile and effectiveness. The majority of the talk will focus on the current GLP-1 medications discussing their roll, clinical effectiveness, side effects and the funding context. It will also discuss the pipeline of future options currently undergoing clinical trials.

DOI: 10.1530/endoabs.111.CS1.1

CS1.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.CS1.2

CS1.3

Abstract Unavailable

DOI: 10.1530/endoabs.111.CS1.3

Endocrine Symposium 1**ES1.1****Screening 100,000 newborns using whole genome sequencing: the generation study**

David Bick, Meekai To, Amanda Pichini, Lindsay Ratan, Christine Cavanagh, Ciara Leckie, Harriet Etheredge, Mathilde Leblond, Liz Gardner, Dalia Kasperaviciute, Joanna Ziff, Alice Tuff-Lacey, Ellen Thomas & Richard Scott
Genomics England, London, United Kingdom

Worldwide, research programs are examining the clinical utility of whole genome sequencing (WGS) as an adjunct to current newborn screening for treatable genetic disorders (<https://www.iconseq.org/>). Genomics England in partnership with National Health Service England (NHSE) have undertaken a Research Ethics Committee approved research study, the Generation Study (GS), to understand the utility of WGS to diagnose rare but treatable conditions by sequencing 100,000 unselected newborns. Genes and their associated conditions for 922 gene-condition pairs (GCPs) were assessed against 4 principles (<https://www.genomicsengland.co.uk/initiatives/newborns>). GCPs fulfilling the principles were reviewed by paediatric specialists and NHSE. Women were recruited during pregnancy at sites across England. Cord blood was the source of DNA for the study. When cord blood was missed, a heel prick was taken. Core laboratories performed DNA extraction then sequencing using the NovaSeq 6000 Sequencing System. WGS data was analysed with a Genomics England purpose-built pipeline. To maximize positive predictive value, only pathogenic and likely pathogenic variants were prioritized. Variants prioritised by the pipeline were reviewed by a laboratory clinical scientist. 'Condition suspected' results were passed to a specialist with experience treating the condition to decide whether to contact parents. The specialist seeing the child arranged confirmatory tests and treatment as appropriate. The 'condition suspected' cases were confirmed with non-molecular confirmatory tests, which exists for nearly all selected conditions. 488 gene-condition pairs, found in 462 genes resulting in 208 conditions are included in the programme. There are 37 recruitment sites accepting participants. 18,349 families have enrolled. 12,286 newborn genomes have been sequenced and analysed. 460 cases have been prioritized by the pipeline for clinical scientist review. 46 cases representing 33 different disorders were returned to a specialist for review and decision whether to report to parents. Evaluation of the GS will include assessing the process and acceptability of newborn sequencing as well as clinical utility and an economic analysis. The Genomics England GS will provide critical experience and extensive evaluation to help define the risks and benefits of genomics as an adjunct to newborn screening in a national program.

DOI: 10.1530/endoabs.111.ES1.1

ES1.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.ES1.2

ES1.3

Abstract Unavailable

DOI: 10.1530/endoabs.111.ES1.3

Endocrine Symposium 2 (Nurses/Endocrine Professionals Session)**ES2.1**

Abstract Unavailable

DOI: 10.1530/endoabs.111.ES2.1

ES2.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.ES2.2

ES2.3**Hypothalamic damage - grieving survival**

Benj Hemingway

Parent, Saviese, Switzerland

For a parent, there is no worse fear than losing a child. But what if you were told that you weren't going to lose your child physically but they will be changed for the rest of their lives, their obesity would make them unrecognisable physically and their personality, character and identity would be gone. Your suffering and grief would have no limit, no rest and no escape. To watch your child suffer everyday and have no solution, no answer and no means to bring her back. When Ivy was diagnosed with a craniopharyngioma at the age of 4, she started a journey that would result in exactly this, that would leave her hating herself and her body. It would also leave her siblings and parents broken and hanging together by a thread. Worst of all it leaves you asking the worst of questions 'why has her survival left us with grief instead of joy?' Today our family are on a quest for solutions which took us to make drastic and life changing steps. In 2024 we moved to my wife's native Switzerland where we are pursuing the drug Setmelantide through Rythm pharmaceuticals. Our hope, is that one day we will bring Ivy home again.

DOI: 10.1530/endoabs.111.ES2.3

Endocrine Symposium 3**ES3.1****Artificial intelligence: reimagining growth and pituitary management**

Paul Dimitri

University of Sheffield Sheffield, United Kingdom

Artificial Intelligence (AI) is reshaping the landscape of paediatric endocrinology, offering novel computational tools to decode the multifactorial complexity of growth and pituitary disorders. These conditions often present with subtle,

overlapping phenotypes and require longitudinal data interpretation, an area where AI excels. By leveraging large-scale datasets encompassing growth trajectories, biochemical markers, neuroimaging, and genetic profiles, machine learning algorithms are now capable of identifying diagnostic signatures with a level of precision that surpasses traditional clinical heuristics. Predictive analytics are increasingly informing therapeutic decisions, particularly in growth hormone therapy. Algorithms trained on real-world treatment data can forecast response trajectories, identify likely non-responders, and support dynamic dose titration, thereby enhancing both efficacy and safety. Machine learning algorithms trained on multimodal datasets including hormonal profiles, MRI imaging, growth velocity data, and genetic markers, are capable of identifying diagnostic signatures of conditions such as growth hormone deficiency, hypopituitarism, and craniopharyngioma-related dysfunction with high sensitivity and specificity. These models can detect subtle deviations in pituitary morphology and function that may precede overt clinical symptoms, enabling earlier recognition and stratification. Overall, as federated learning and synthetic data generation mature, AI systems are poised to overcome data scarcity and heterogeneity. AI-driven insights are facilitating a shift toward precision endocrinology, where care is tailored to individual biological and contextual profiles. However, the integration of AI into paediatric practice brings challenges. Data heterogeneity, limited paediatric-specific training sets, and algorithmic bias pose risks to generalisability and equity. Rigorous validation, transparent model governance, and inclusive data sourcing are essential to mitigate these concerns. Moreover, embedding AI into clinical workflows demands interoperability, clinician trust, and multi-disciplinary collaboration. Looking ahead, innovations such as federated learning, AI-assisted imaging, generative AI, quantum AI-fusion, synthetic data generation, and AI-enabled registries hold promise for advancing research and care in rare endocrine conditions and improving population-level surveillance. The convergence of clinical expertise and computational intelligence is poised to redefine diagnostic paradigms, therapeutic strategies, and long-term outcomes in paediatric growth and pituitary disorders, heralding a new era of data-driven, child-centred endocrine care.

DOI: 10.1530/endoabs.111.ES3.1

ES3.2

Looking to the future - an oncologist's view of managing late effects

Anna Jenkins

Sheffield Children's Hospital, Sheffield, United Kingdom

Approximately 85% of children, adolescents, and young adults (CAYA) diagnosed with cancer survive long term with an estimated 45,000 long term survivors of childhood cancer living in the UK today. It is well recognised that survival comes at a cost, with 60 - 90% of survivors experiencing at least one long term health problem (late effect). Endocrine late effects are common, occurring in approximately 50% of survivors, but the list of potential health problems is extensive and leads to significant increased morbidity and mortality in this population. The long-term consequences of recent developments in the treatment and supportive care of CAYA cancer are still to be established, but they are likely to impact on future health of these patients, including increasing the risk of endocrine problems. Managing the health needs of this ever-increasing population as they age places significant demands on services. As well as focusing on the current physical and mental health needs of survivors, we should be researching and developing ways to encourage healthy behaviours that have the potential to modify long term health risks and improve health in the future for this population.

DOI: 10.1530/endoabs.111.ES3.2

ES3.3

Abstract Unavailable

DOI: 10.1530/endoabs.111.ES3.3

Diabetes Symposium 1

DS1.1

Abstract Unavailable

DOI: 10.1530/endoabs.111.DS1.1

DS1.2

Genetic screening for type 1 diabetes and primary prevention studies from GPADD

Loredana Marcovecchio

University of Cambridge, Cambridge, United Kingdom

The Global Platform for the Prevention of Autoimmune Diabetes (GPPAD) was established in 2015 to enable population-scale genetic screening in infancy and to conduct trials of interventions aimed at preventing β -cell autoimmunity and type 1 diabetes (T1D). The GPPAD screening programme offers genetic risk testing to newborns or infants in several European countries, including Germany, Poland, Belgium, Sweden, and the United Kingdom. A genetic risk score (GRS), derived from approximately 46 single-nucleotide polymorphisms (SNPs) encompassing HLA-DR/DQ haplotypes and non-HLA risk loci, together with information on first-degree family history of T1D, is used to stratify risk. Infants whose GRS and family background confer a >10% risk of developing multiple β -cell autoantibodies by age 6 years are identified as high-risk and invited to participate in primary prevention trials.

To date, three main trials have been conducted within GPPAD:

- POINT (Primary Oral Insulin Trial), which investigates oral insulin tolerance induction and is due to report results soon;
- SINT1A (Supplementation with *Bifidobacterium infantis* for Primary Prevention of Type 1 Diabetes), where recruitment is complete and follow-up is ongoing; and
- AVAnT1A (Anti-viral Action against Type 1 Diabetes Autoimmunity), currently open to recruitment, in which the intervention consists of COVID-19 vaccination (three doses starting at 6 months of age) to assess whether antiviral protection reduces the incidence of islet autoimmunity.

Follow-up across all GPPAD trials includes longitudinal monitoring for islet autoantibodies and progression to clinical T1D. GPPAD thus provides a unique infrastructure for large-scale genetic screening and early-life intervention studies aimed at the primary prevention of T1D

DOI: 10.1530/endoabs.111.DS1.2

DS1.3

Abstract Unavailable

DOI: 10.1530/endoabs.111.DS1.3

Diabetes Symposium 2

DS2.1

The GIRFT programme for children and young adults' diabetes

Dita Aswani

Consultant Paediatrician, Sheffield Children's NHS Foundation Trust, Sheffield, United Kingdom. NHSE GIRFT Clinical Advisor for CYA Diabetes. NPDA dataset and methodology group, RCPCH

This session will present key findings and prevalent themes from the GIRFT programme for Children and Young Adults' Diabetes services, following completion of all 42 ICB review meetings across England: insights into system-level architecture, examples of good practice, and common challenges. The session will also outline overarching recommendations that will inform the upcoming GIRFT national report, with a focus on key opportunities for improvement that are achievable within current service realities and in alignment with the 10 year health plan for England.

DOI: 10.1530/endoabs.111.DS2.1

DS2.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.DS2.2

DS2.3**Grand challenge****Simon Heller**

University of Sheffield, Sheffield, United Kingdom

I will describe how Diabetes UK and Breakthrough T1D have worked with the Steve Morgan Foundation to distribute £50m of research funding to accelerate the progress to cure in type 1 diabetes. Drawing on the generosity of the global diabetes scientific community who have provided advice and guidance, the funding has been distributed to 3 areas, causes of T1D and prevention, cell therapy as curative therapy and novel insulins. The first awards were distributed to 3 early career researchers who are addressing the first two areas and further funding has been provided to other groups in the UK who are examining key steps which will improve the ability of immune therapies to prevent the onset of diabetes at high risk, a clear pathway to cure. The GC funding has allowed us to work with NIHR to ensure that the UK is an attractive option for both pharma and academics to place their clinical trials of stem cell therapy. We are now planning to set up a clinical trial network involving the NIHR CRFs and BRCs to facilitate both rapid recruitment and high quality delivery across the UK. Both approaches identify key pathways to a cure, although this is still some years away. The final tranche of funding has been provided to address the major challenge of finding novel insulins which are fit for therapeutic use. We have awarded preliminary funding to international academics who are proposing exciting new innovative approaches to solving this longstanding challenge. Since most scientists who are conducting research in this area work outside Europe, with Steve and Sally Morgan's approval, funding has been awarded to do labs in the USA, China and Australia. There is no doubt that this unprecedented and generous funding has sparked global interest and accelerated the path to a cure for those with T1D. The next challenge is to ensure that the scientific activity which has been generated can be built on, ensuring that progress in these key areas continues, accelerates

and those who have been supported can use their work as a springboard to leverage substantial funding from other sources.

DOI: 10.1530/endoabs.111.DS2.3

Diabetes Symposium 3**DS3.1**

Abstract Unavailable

DOI: 10.1530/endoabs.111.DS3.1

DS3.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.DS3.2

DS3.3

Abstract Unavailable

DOI: 10.1530/endoabs.111.DS3.3

How Do I?

HDI1.1

Abstract Unavailable

DOI: 10.1530/endoabs.111.HDI1.1

HDI1.2

Abstract Unavailable

DOI: 10.1530/endoabs.111.HDI1.2

Debate

D1.1**Use of growth hormone in idiopathic short stature (For)**Justin Davies

University of Southampton, Southampton, United Kingdom

Idiopathic Short Stature (ISS) is a term given to children who are born with a normal birth weight and length who have short stature with no identifiable cause. ISS defined as a height standard deviation score (SDS) ≥ 2 below the corresponding mean height for age, sex, and population, and no identifiable pathological cause following evaluation. It affects an estimated 2.5% of the general population and is challenging to diagnose and manage. In 2003, the Food and Drug Administration (FDA) approved the use of recombinant human growth hormone (hGH) for children with ISS with a height of 2.25 SDS below the mean ($\sim < 1.2$ nd height centile). hGH therapy was subsequently approved for the treatment of ISS in US, Canada, and Latin America, but not in the European Union or the UK. More than two decades later, use of hGH therapy for ISS remains a subject of ongoing discussion as ISS is not universally accepted as an indication for hGH treatment. This debate will highlight the issues in favour (Prof Justin Davies) and against (Prof Helen Storr) the use of hGH for ISS with the aim of informing and promoting discussions of the use of hGH for growth management.

DOI: 10.1530/endoabs.111.D1.1

D1.2**Debate: use of GH in idiopathic short stature (ISS). professors helen storr and Justin Davies**Helen Storr

Queen Mary University London and Barts Health NHS Trust, London, United Kingdom

Idiopathic Short Stature (ISS) is a term given to children who are born with a normal birth weight and length who have short stature with no identifiable cause. ISS defined as a height standard deviation score (SDS) ≥ 2 below the corresponding mean height for age, sex, and population, and no identifiable pathological cause following evaluation. It affects an estimated 2.5% of the general population and is challenging to diagnose and manage. In 2003, the Food and Drug Administration (FDA) approved the use of recombinant human growth hormone (hGH) for children with ISS with a height of 2.25 SDS below the mean ($\sim < 1.2$ nd height centile). hGH therapy was subsequently approved for the treatment of ISS in US, Canada, and Latin America, but not in the European Union or the UK. More than two decades later, use of hGH therapy for ISS remains a subject of ongoing discussion as ISS is not universally accepted as an indication for hGH treatment. This debate will highlight the issues in favour (Prof Justin Davies) and against (Prof Helen Storr) the use of hGH for ISS with the aim of informing and promoting discussions of the use of hGH for growth management.

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Personal Practice Session

PPS1

Abstract Unavailable

DOI: 10.1530/endoabs.111.PPS1

Oral Communications

CME Case Presentations 1

OC1.1

“Hidden in plain sight”: a mini-series of diagnostic difficulties in paediatric cushing’s disease

Aikaterini Perogiannaki^{1,2}, James Yong^{1,3}, Talat Mushtaq^{1,3}, Nikolaos Kyriakakis^{1,3} & Caroline Steele^{1,3}

¹Leeds Teaching Hospitals NHS Trust, Leeds, United Kingdom; ²Barts Health NHS Trust, London, United Kingdom; ³University of Leeds, Leeds, United Kingdom

Background

Paediatric Cushing’s Disease (CD) is rare, and symptoms are variable, resulting in delayed diagnosis, prolonged morbidity, and psychological impact.

Case Series

We present three adolescents with CD highlighting challenges in diagnosis.

- **Case 1:** A 13-year-old girl with coeliac disease was reviewed for stalled puberty, possible hirsutism and poor growth despite higher weight. Turner syndrome was excluded by karyotype but raised DHEAS (15.8umol/l) noted. Three significantly elevated urinary cortisols and unsuppressed cortisols on overnight- (ONDST) and 48hour-dexamethasone suppression confirmed CD. Pituitary MRI was initially inconclusive, but inferior petrosal sinus sampling (IPSS) suggested right-sided ACTH production. Treatment with metyrapone began whilst clarification of lateralisation occurred, before transsphenoidal resection. Despite initial fall in ACTH/cortisol there is evidence of relapse and metyrapone treatment has re-started, with PET imaging pending.
- **Case 2:** A 17-year-old male with obesity developed hyperglycaemia with raised HbA1c (resolved with lifestyle changes) alongside COVID pneumonitis (aged 15 years), hypertension, and metabolic-associated steatotic liver disease (MASLD). Weight gain initially coincided with increased snacking, but persisted despite lifestyle changes. Examination revealed acanthosis nigricans and round facies. Further investigations due to persistent symptoms found raised urinary cortisol and failure to suppress cortisol on ONDST. Pituitary MRI identified right sided microadenoma. Retrospective questioning revealed stalled growth (age 13); examination revealed 2ml testes and small penis. MEN1 was suspected (but excluded) (hyperparathyroidism and raised glucagon). Imaging excluded ectopic ACTH and pancreatic tumour. Transsphenoidal surgical resection 9 months after diagnosis found ACTH-secreting adenoma (histologically) and was curative, with subsequent resolution of symptoms.
- **Case 3:** A 14-year-old boy presented, initially in private practice as not fulfilling the criteria for Complications of Excess Weight (CEW) clinic, with short stature, central obesity, facial plethora, increased body hair and no striae. Elevated urinary cortisol and unsuppressed cortisols on ONDS and 48hour-dexamethasone suppression confirmed CD. MRI showed a 4.5 mm left-sided microadenoma and he recently had an IPSS.

Conclusion

A high index of suspicion is essential to diagnose Cushing’s disease (CD) in adolescents, as the underlying symptoms vary. Early endocrine referral and coordinated diagnostic pathways are critical to reduce delay in investigation, reducing long-term complications, and improving patient outcomes.

DOI: 10.1530/endoabs.111.OC1.1

OC1.2

A rare case of papillary thyroid carcinoma in a child with a CDC73 mutation

Munibah Bashir¹, James Blackburn¹, Charlotte Jarvis¹, Suzanne Armitage², Neil Houghton³, Samantha Goh⁴, Francesca McDowell⁵ & Renuka Ramakrishnan¹

¹Department of Paediatric Endocrinology, Alder Hey Children’s Hospital, Liverpool, United Kingdom; ²Department of Biochemistry, Alder Hey Children’s Hospital, Liverpool, United Kingdom; ³Department of Endocrine Surgery, Liverpool University Foundation Trust, Liverpool, United Kingdom; ⁴Ear, Nose & Throat Department, Alder Hey Children’s Hospital, Liverpool, United Kingdom; ⁵Department of Paediatric Histopathology, Alder Hey Children’s Hospital, Liverpool, United Kingdom

Background

Thyroid cancers are less common in children than in adults, with papillary thyroid carcinoma (PTC) being the most frequent type. Although papillary thyroid carcinoma (PTC) has been reported in adults with CDC73 mutations, there are no previous reports of this occurring in children.

Case

An asymptomatic 15-year-old male under regular monitoring for unilateral PUJ obstruction was found to have persistent hypercalcaemia and primary

hyperparathyroidism. Biochemistry showed: adjusted calcium of 3.08 mmol/l (2.15–2.74), PTH of 26.4 pmol/l (1.1–6.9), and an increased urine calcium: creatinine ratio of 1.13 mmol/ mmol Cr (<0.60). USS neck and Tc-99m sestamibi scan localized the parathyroid adenoma to the right neck. Familial hyperparathyroidism gene panel reported a heterozygous CDC73 likely pathogenic variant. A jaw radiograph was normal. Family genetics didn’t report this mutation in first-degree relatives. Normal calcium levels were achieved with cinacalcet. He underwent a right-sided neck exploration for parathyroid adenoma excision. Due to central neck lymphadenopathy and his genetic risk, a right hemithyroidectomy and central neck dissection was also performed. Histology confirmed a right parathyroid adenoma with metastatic PTC in the lymph nodes. The patient subsequently underwent completion of thyroidectomy, followed by radioiodine therapy. Postoperatively, he recovered well and is currently on levothyroxine and oral calcium supplements.

Discussion

Papillary thyroid carcinoma accounts for 90% of childhood thyroid cancers. Commonly associated gene mutations include RET, BRAF, RAS, and NTRK; less common gene mutations include DICER1, TP53, and PTEN. CDC73 mutations are usually autosomal dominant but can also arise de novo. CDC73-related disorders include HPT-JT syndrome, familial isolated hyperparathyroidism (FIHP), and parathyroid carcinoma. Presentation often occurs in late adolescence or early adulthood with a single parathyroid adenoma. This condition predisposes to tumours in various sites, including the jaw bones, kidneys, uterus, and ovaries. Our case offers a rare and valuable insight into the potential role of CDC73 mutations in paediatric thyroid cancer. Although there are no established national guidelines for the management of patients with CDC73 mutations, studies recommend offering genetic testing to parents and first-degree relatives and regular biochemical and radiological screening to the affected members.

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CME Case Presentations 2

OC2.1

Hyperparathyroidism, pseudohypoparathyroidism (PHP) or pseudopseudohypoparathyroidism (PPHP) – solving the GNAS riddle

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Background

Pseudopseudohypoparathyroidism is a rare genetic disorder caused by heterozygous paternally inherited inactivating mutations in the *GNAS* gene, leading to the skeletal phenotype of Albright Hereditary Osteodystrophy (AHO), but without the hormone resistance. Our PPHP case had normal stature and shortened metacarpals on presentation along with an elevated parathyroid hormone (PTH) thus sharing features with PHP1a/c and PHP1b.

Case Presentation

An 11-year-old female presented to the maxillofacial team with a peripheral ossifying fibroma of her hard palate which required excision. There was no personal or family history of bone disorders or fractures. Dietary history revealed minimal dairy intake. On examination, she had no dysmorphic features, puberty was established, she had short 3rd–5th metacarpals and partial syndactyly of the 2nd and 3rd toes. Her height was on the 25th centile, but with a recent trajectory indicating a final height below the 0.4th centile. Baseline bloods indicated elevated PTH levels of 34.9 pmol/l (1.3–9.3), low Vitamin D (23 nmol/l) and normal calcium, phosphate, ALP and thyroid function. LH 5.1 IU/l, FSH 8.9 IU/l and oestradiol 270 pmol/l. She was commenced on vitamin D treatment and an increase in dietary calcium was advised. The bone age was advanced by two years and showed short and broad 3rd to 5th metacarpals. A skeletal survey confirmed brachydactyly, cone-shaped epiphyses, osteopenia, square iliac bones, and mild vertebral endplate changes. The diagnosis was suggestive of pseudohypoparathyroidism. Genetic testing confirmed a *GNAS*-related disorder consistent with pseudopseudohypoparathyroidism.

Conclusion

At presentation, this girl exhibited features overlapping with both AHO and PHP1b. However, the elevated PTH was due to secondary hyperparathyroidism caused by co-existing Vitamin D deficiency and low dietary calcium, rather than true PTH resistance. The normal stature was due to well established puberty with the final height expected to be very short. This case highlights the importance of careful clinical, pubertal, biochemical and dietary assessment when considering the differential diagnosis of *GNAS*-related disorders. Genetic confirmation is key as it facilitates appropriate counselling and family planning. Given the complex imprinting pattern of the *GNAS* gene, her offspring are at risk of inheriting PHP1a.

DOI: 10.1530/endoabs.111.OC2.1

OC2.2**A rare case of heterozygous INSR variant presenting as hyperandrogenism in adolescence**Laurentya Olga¹, Binu Anand¹ & Ajay Thankamony²¹West Suffolk NHS Foundation Trust, Bury St Edmunds, United Kingdom;²Cambridge University Hospitals, Cambridge, United Kingdom**Background**

Monogenic insulin resistance syndromes are rare but important differential diagnoses in adolescents presenting with features of hyperandrogenism, particularly when obesity is absent. We present a case highlighting diagnostic challenges and the role of genetic testing in confirming a type A insulin resistance. Case presentation

A 12-year-old female presented with progressive hirsutism, acne, and deepening of the voice. Her BMI was within the normal range (16.9 kg/m², 23rd centile), and her height was on the 88th centile. Initial laboratory investigations revealed elevated serum testosterone (2.4 nmol/l; reference <1.8), markedly raised C-peptide (7070 pmol/l; reference 174–960), and fasting insulin (554 pmol/l; reference 0–80), with adiponectin levels ranging between 8.3–9.9 µg/mL. Pelvic ultrasound demonstrated prepubertal uterine features without evidence of polycystic ovarian morphology. Over time, her hyperandrogenic features progressed, and she remained amenorrhoeic. Family history included maternal polycystic ovary syndrome (PCOS) diagnosed at age 20 and a half-sister with mild hirsutism. A comprehensive metabolic and endocrine work-up excluded adrenal pathology and lipodystrophy. Genetic testing identified a heterozygous, maternally inherited INSR likely pathogenic variant (c.3392C>G; p.Pro1131Arg), consistent with type A insulin resistance syndrome.

Discussion

Heterozygous INSR variants can present with variable phenotypic expression, ranging from asymptomatic carriers to adolescents with pronounced hyperandrogenism and insulin resistance. Unlike more severe biallelic INSR mutations, heterozygous variants are associated with milder metabolic derangements but have important implications for reproductive health, including an increased risk of gestational diabetes.

Conclusion

This case underscores the importance of considering monogenic insulin resistance in lean adolescents with hyperandrogenism and highlights how genetic diagnosis informs counselling and long-term management. Awareness of these rare presentations is crucial for early recognition and appropriate follow-up.

Keywords

Type A insulin resistance, INSR mutation, hyperandrogenism, adolescence, monogenic diabetes

DOI: 10.1530/endoabs.111.OC2.2

CME Case Presentations 3**OC3.1****Teenage kicks: the complexity of an adolescent living with diabetes, subcutaneous insulin resistance syndrome (SIRS) or a case of induced illness?**

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A 16-year-old female with type 1 diabetes presented with ketotic hyperglycaemia and had a 13-week admission with presumed Subcutaneous Insulin Resistance Syndrome (SIRS). She was on a hybrid closed-loop system (Omnipod 5 & DEXCOM G6), using Fiasp. She has multiple dermatological diagnoses - hyperhidrosis, ichthyosis, keratosis pilaris and spontaneous surgical emphysema. She started on a sliding scale of intravenous insulin, adjusted according to her (limited) response to various subcutaneous insulins tried. The following were unsuccessful regimes: Tresiba 40 units alongside the Omnipod 5mm angled cannula with Fiasp, Novorapid, Actrapid, Humalog, Lyumjev and Apidra. Her TDD (total daily dose), reached ~500 units via her pump. Longer needles and soft sets were tried. Corrections were administered via pen alongside these regimes (using both 8mm and 12mm needles). She was changed from Tresiba (100units/ml) to Toujeo (300units/ml) to protect her sites. This was increased to 264units. Humalog and Lyumjev were tried via a Medtronic 780G pump with a 10mm steel needle. A Humalog-Heparin mix was tried before pump treatment was discontinued. She moved to HumulinR(U500) via pens alongside increasing doses of Toujeo. There was no response at a TDD of 1440 units (1200 units HumulinR and 240 units Toujeo). Her initial insulin antibodies, run in the Surrey laboratory, were reported as 'positive'. 10 weeks into admission concerns were raised regarding issues with her intravenous insulin. Multiple intravenous lines appeared to have been tampered with. She refused to see clinical psychology. She was referred to the National Severe Insulin Resistance Service, Cambridge. Her

insulin IgG antibody was 3 mg/l (0-5 mg/l) via Cambridge laboratory and was reported as negative. Subcutaneous Actrapid was resumed via pens; with staff strictly administering all injections. There was an immediate response, the patient was weaned off intravenous insulin within 36 hours. Confounders in this case included the reported delivery of insulin through the pump (visible on the pump settings), her dermatological diagnoses, level of supervision and reported desire for discharge. Furthermore, it was charted injections were being 'administered' by nursing staff when in fact the patient was being observed self-administering. This case provides significant learning on the complexities of managing adolescent diabetics.

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OC3.2**A difficult diagnosis of B-all presenting with hypercalcaemia and multiple fractures**Jennifer McKechnie¹, Thomas Savage², Shahzya Chaudhury³, Avril Mason¹ & Sze Choong Wong¹

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Introduction

Acute lymphoblastic leukaemia (ALL) can present with hypercalcaemia in infancy. We describe a 10-month-old boy with B-ALL who presented with hypercalcaemia and multiple fractures, including vertebral fractures (VF), where diagnosis required multiple bone marrow biopsies. This case also raises important questions about the duration of bisphosphonate therapy and the role of routine spine imaging in ALL.

Case

The infant presented with a one-month history of vomiting, fever, lethargy, and reduced oral intake. There were no signs of bruising, night sweats, or organomegaly. Blood tests revealed marked hypercalcaemia (adjusted calcium 4.81 mmol/l), undetectable parathyroid hormone (PTH), normal 25-hydroxyvitamin D, and evidence of acute kidney injury. Full blood count showed mild normocytic anaemia; the blood film was unremarkable. Imaging revealed generalised osteopenia, multiple VF, anterior rib, and long bone fractures. Dense bones were not observed, making osteopetrosis unlikely. Whole-body MRI showed no additional pathology but suggested an infiltrative process. Diagnosis of B-ALL was confirmed after three bone marrow biopsies over six weeks. Hypercalcaemia was managed with two courses of IV pamidronate. Given ongoing corticosteroid and chemotherapy use, regular IV zoledronate was initiated for bone protection. Spine radiographs at 10 months post-diagnosis showed evidence of vertebral reconstitution, with stable appearances on subsequent imaging.

Discussion

This case highlights the diagnostic complexity of ALL in infants, where obtaining adequate marrow samples can be technically challenging. Diagnosis must be confirmed before commencing chemotherapy to allow appropriate monitoring of treatment response. It also raises key considerations in the management of VF in ALL. Studies suggest 16% of children have VF at diagnosis with routine spine imaging and 25% may develop new VF during treatment. While many children with ALL and VF may undergo spontaneous vertebral reshaping without bisphosphonates, the optimal duration of treatment in symptomatic cases remains uncertain. The role of routine spinal imaging at diagnosis and with follow-up, particularly in asymptomatic patients, also warrants further investigation. We propose an age-stratified monitoring and management approach, informed by current evidence, to guide bisphosphonate use and spine imaging in paediatric ALL.

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CME Case Presentations 4**OC4.1****Diagnostic and phenotypic challenges in a rare case of 46, XX differences in sex development (DSD) with a heterozygous RSP01 variant**Sushil Yewale¹, Louise Apperley¹, Jaynee Chauhan², Harriet Corbett³, Urmi Das¹, Zoe Edwards², Rachel Hart², Fiona McAndrew³, Cara Williams³ & Julie Park¹

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Introduction

We present a case of 46,XX sex-determining region Y (*SRY*)-negative difference in sex development (DSD) where a heterozygous paternally inherited pathogenic *RSPO1* gene variant has been identified. Further work is needed to uncover a second variant.

Case presentation

A term infant, born to non-consanguineous parents, presented at birth with atypical genitalia. There was no history of maternal androgen exposure or family history of DSD/infertility. Examination revealed an enlarged genital tubercle (2 cm), perineal opening at its base, mild labioscrotal rugosity, and no palpable gonads; a mobile left inguinal lump was noted (external genitalia score 1.5/12). Ultrasound showed an anteverted uterus (33x11x8mm), an echogenic structure (7x11x5mm) consistent with a testis in left inguinal canal, with associated hernia. Standard evaluations for congenital adrenal hyperplasia were normal. Quantitative fluorescent PCR and peripheral blood karyotype confirmed 46,XX *SRY*-negative pattern. Hormonal assessment (day 9) revealed LH 11.4 IU.L⁻¹, FSH 15.8 IU.L⁻¹, testosterone 1.4 nmol.L⁻¹ and oestradiol <100 pmol.L⁻¹. Laparoscopic hernia repair allowed visualisation of healthy urethra, uterus, and vagina. Gonadal biopsy confirmed testicular tissue (46,XX karyotype). Human chorionic gonadotropin stimulation test showed robust testosterone response (basal/stimulated): DHEAS 0.8/<0.4 µmol.L⁻¹ (NR 1.6–7.8), androstenedione 1.9/3.6 nmol.L⁻¹ (2.0–5.4), testosterone 4.4/8.4 nmol.L⁻¹, dihydrotestosterone 1.2/2.09 nmol.L⁻¹ (0.32–1.64). Genetic analysis identified a paternally inherited heterozygous pathogenic *RSPO1* variant. Female sex assignment was agreed through multidisciplinary team discussions with the family. Follow-up ultrasound at 3 months showed bilateral gonadal structures (2.2x0.9x1.3 cm and 1.1x0.7x0.8 cm) with cysts, and previously biopsied testis. Eye examination was normal, and no skin changes. Further genetic analysis is underway to identify a second *RSPO1* variant.

Discussion

Pathogenic biallelic *RSPO1* variants typically cause an extremely rare autosomal recessive 46,XX DSD associated with palmoplantar keratoderma and predisposition to squamous cell carcinoma; thus attribution of this infant's phenotype to a single heterozygous *RSPO1* variant would be premature. The presence of histologically confirmed testicular tissue in a 46,XX context, bilateral evolving gonadal structures, and robust Leydig responsiveness suggest broader gonadal developmental complexity, potentially within the ovotesticular/complex DSD spectrum. Our case underscores the value of staged phenotyping, longitudinal imaging, and multidisciplinary counselling when genotype-phenotype correlation is incomplete.

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Endocrine Oral Communications 1

OC5.1

A novel polygenic risk prediction tool: improving the diagnosis of short stature

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Introduction

Short stature (SS), defined as height more than 2 standard deviation scores (SDS) below the mean, affects ~2% of UK children and accounts for ~50% of referrals to paediatric endocrine clinics. Over 80% of children with SS remain undiagnosed, resulting in repeated investigations, delayed management, and significant anxiety for families and clinicians. This highlights the urgent need for advanced genetic diagnostics to enable early diagnosis, personalised management, and improved clinical outcomes. Height is a highly heritable trait (80-90%). Recent trans-ancestry genome-wide association studies have identified over 12,000 common variants accounting for ~40% of height variance. SS may be due to a rare monogenic disorder or a high burden of common variants associated with short height. Current diagnostic strategies target monogenic causes but cannot distinguish these from polygenic ones. We hypothesised that a polygenic prediction model could help discriminate monogenic from polygenic SS and improve diagnostic pathways.

Methods

We analysed a sub-cohort ($n = 161$) from our Genetic Research Analysing Short Patients (GRASP) study, including monogenic diagnoses ($n = 81$), variants of uncertain significance (VUS) ($n = 23$), and undiagnosed ($n = 57$) individuals. SNP array genotyping was performed, and polygenic scores for height were calculated using >12,000 trans-ancestry GWAS variants. These scores were regressed against measured height SDS using simulated reference data ($n = 10,000$). Deviation from genetically predicted height was defined as a residual > 2 and Mahalanobis $P < 0.001$.

Results

Eighty individuals deviated significantly from their polygenic height prediction suggesting a likely monogenic cause. Of these, 48 had confirmed monogenic diagnoses, 11 had putative monogenic causes (VUS), and 21 were undiagnosed. The greatest deviations were found in the most severe SS individuals with known pathogenic variants in *GHR* (Laron syndrome), *CCDC8* (3M syndrome), and *BLM* (Bloom syndrome), supporting the model's ability to discriminate 'true' monogenic SS.

Conclusion

Our approach may prioritise cases for advanced sequencing, reduce unnecessary investigation in polygenic cases, and support earlier, equitable precision care in SS. Genome-wide SNP testing is also cost-effective (£30-40 per patient) compared to ~£2,000 for comprehensive NHS genetic testing. Integration of polygenic scores into diagnostics represents a clinically actionable advancement in paediatric endocrine genomics and contribute to broader implementation in precision medicine.

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OC5.2

Pilot data from the I-HH registry: improving management in hypogonadotropic hypogonadism

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Background

Hypogonadotropic hypogonadism (HH) is a rare endocrine disorder, either congenital due to pathogenic variant(s) in over 60 known genes or acquired, usually secondary to a brain tumour. The condition has significant implications for growth and psychosocial wellbeing in both sexes, and fertility in men. Despite growing awareness, timely diagnosis remains a challenge, and management approaches vary, particularly with the evolving role of gonadotropin therapy in male puberty. The International Hypogonadotropic Hypogonadism (I-HH) Registry, hosted on the SDMregistries platform, was established to support standardised data collection and facilitate international collaboration to evaluate treatment strategies and clinical outcomes.

Methods

The first comprehensive data extraction from the I-HH Registry was completed in April 2025, capturing baseline data from 96 individuals (59 males, 37 females) across 15 clinical centres in 13 countries. Participants ranged from 10 to 34.5 years of age. Pubertal induction was initiated in 49 patients (34 males, 15 females), at a median age of 14 years (IQR 12–17; range 11–19). An additional female received treatment for pubertal completion. Among males, 13 (22%) reported olfactory deficits—8 with anosmia and 5 with hyposmia; 3 females (9%) had anosmia. Micropenis was documented in 15 males (25%) and undescended testes in 17 (29%), with 14 (24%) presenting both. Genetic analyses were available for 32 patients, revealing pathogenic variants in HH-associated genes including *ANOS1*, *FGFR1*, *PROKR2*, *CHD7*, *SOX10*, *GNRHR*, and *PROPL1*. Medication data from 34 males showed that 16 were treated with gonadotropins, across four centres in three countries. Seven females underwent fertility treatment; no males had completed fertility therapy at the time of data collection. Spermatogenesis was assessed in four males with sperm counts ranging from 14.4 to 59.9 × 10⁶/ml in those post-gonadotropin therapy.

Conclusion

This early dataset from the I-HH Registry highlights the feasibility of coordinated, prospective data collection for HH. While current data are primarily cross-sectional, the registry establishes a framework for evaluating the impact of clinical management and long-term outcomes from longitudinal data. Future directions include incorporation of growth metrics such as peak height velocity using SITAR growth curve modelling.

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OC5.3

Assessment of fetal growth trajectories and relationships with early infant adiposity at a single site within the UK pregnancies better eating and activity trial (UPBEAT)

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Introduction

Cardiometabolic disease risk is related to small (SGA) or large-for-gestational-age (LGA). Previously, we demonstrated suboptimal fetal growth without SGA is related to adverse risk (BSPED 2022, OC6.2). The multi-site UK Pregnancies Better Eating and Activity Trial (UPBEAT, $n = 1,555$) had randomised pregnant women with obesity ($\text{BMI} \geq 30 \text{ kg/m}^2$) into a pregnancy lifestyle intervention. UPBEAT infants provide an ideal cohort to examine excessive fetal growth and infant health markers.

Aims

1) Define fetal weight trajectories ($\Delta\text{fetalwt}$) within the Manchester UPBEAT participants. 2) Assess relationships between $\Delta\text{fetalwt}$ and postnatal adiposity markers at six months.

Methods

Retrospective data from 20-week scans were used to calculate estimated fetal weight (EFW) and centiles. Fetal weight trajectory was calculated as WHO birthweight centile minus WHO EFW centile, divided by the number of days in between. Using available data from the 6-month UPBEAT postnatal study, Pearson's and Spearman's rank tests were performed in R version 4.5, to assess correlations between $\Delta\text{fetalwt}$ and 6-month weight, BMI, abdominal circumference (AC) and biceps, triceps and sum of skinfold thicknesses (SDS).

Results

Median maternal booking BMI and gestational weight gain were 35 kg/m^2 (range 30 to 60) and 7 kg (-1 to 23). 35/122 infants (29%) had participated postnatally, comprising 57% males, 72% appropriate-for-gestational-age, 14% LGA (>90th centile) and 14% SGA (<10th centile). 80% were White, 9% South Asian and 6% Black, and half were breastfed. 25/35 (71%) had a positive $\Delta\text{fetalwt}$, with no difference between UPBEAT control (mean 0.08 (SD 0.3)) and intervention (mean 0.07 (0.3)) groups. $\Delta\text{fetalwt}$ correlated with 6-month AC SDS ($r = 0.37$, $P = 0.046$), and a trend towards significance was observed with 6m weight SDS ($r = 0.32$, $P = 0.072$), but not with 6m BMI SDS nor any other 6m measures. When LGA-born infants were excluded, $\Delta\text{fetalwt}$ correlated with 6m AC SDS ($r = 0.42$, $P = 0.031$, $n = 30$) alone. A trend was observed when LGA and SGA-born were excluded ($r = 0.39$, $P = 0.067$, $n = 25$).

Conclusion

In this Manchester subset of UPBEAT participants, greater $\Delta\text{fetalwt}$ was associated with larger AC SDS, even in the absence of LGA. Including data from other UPBEAT sites could help establish relationships between excessive fetal weight gain and postnatal adiposity markers in infants born appropriate-for-gestational-age.

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OC5.4

Disruption of the Wnt-antagonist APC in the pituitary stem cells is a driver of adamantinomatous craniopharyngioma

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Background

Adamantinomatous craniopharyngiomas (aPCs) are complex intracranial neoplasms that arise in the sellar or parasellar region affecting the endocrine system leading to severe comorbidities. Activating mutations, resulting in degradation resistant from of β -catenin are the main driver for many of these neoplasms. However, underlying genetic drivers for a number of tumours remains unknown.

Aims

To determine if disruption the Wnt inhibitor, adenomatous polyposis coli (APC), can lead to aCP tumorigenesis independent of β -catenin mutations.

Methods

To demonstrate that APC can be a driver of aCPs independent of β -catenin, two novel transgenic murine lines were created. Using the pituitary specific Cre driver, *Prop1:Cre*, the large final coding exon of *Apc* was deleted, in the other model a hypomorph allele was created. Detailed phenotypic assessment was performed using immunofluorescence, in-situ hybridisation and mRNA sequencing of tumour initiating cells.

Results

Phenotyping of both models shows that *Apc* can drive aCP tumour formation. These tumours present with all histological and molecular hallmarks of aCPs. The hypomorph allele for *Apc* leads to postnatal tumour development, indicating that disruption of *Apc* function alone, is sufficient to drive aCP formation independent of β -catenin mutations. This model also developed a phenotype of hypothalamic damage, with morbid obesity developing in early adulthood. Transcriptomic analyses of early tumour-initiating cells reveal early clusters of accumulating β -catenin undergo senescence, which differs molecularly to β -catenin positive tumours.

Discussion

Our results show that disruption of *Apc* leads to the development of aCPs independent of β -catenin. The two novel genetic models highlight the phenotypic variability aCPs and form a basis for further study of this heterogeneity. The onset of tumours in the postnatal period and the development of hypothalamic obesity, provides an opportunity to develop therapeutic agents to target tumour development and hypothalamic sequelae of the tumour. Together our findings reinforce the need for genetic testing for mutations in *APC* in patient with aCP, and the increased surveillance of patients with APC-pathogenic syndromes for the development of aCPs as part of their phenotypic spectrum.

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OC5.5

Comparison of electronic growth chart software reveals clinically relevant variability hindering growth assessment: a UK study

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Background

Introduction of electronic patient records has prompted electronic growth chart software (EGCS) use for growth assessment. EGCS is recommended but there is no standardisation in clinical practice. Accuracy and consistency between EGCS are unknown.

Aim

To assess accuracy of UK EGCS packages including approaches to gestational age (GA) correction.

Methods

Of 82 participating centres, 60 (73%) used EGCS. 14 selected centres enabled investigation of 10 EGCS. For each EGCS, two centres completed standardised testing (clinical vignettes with "dummy data"), generating SDSs to analyse clinical growth management decisions. The responses informed a second-stage protocol to analyse observed discrepancies in birthweight, GA correction, height, head circumference (HC) and body mass index (BMI) SDS. Coefficient of Variation (CV) quantified consistency between centres: <5% good, 5-30% moderate and >30% poor. The proportion of EGCS-generated SDSs matching RCPCH-Application Programming Interface (API) EGCS-generated SDSs (gold standard) defined the accuracy.

Results

10 EGCS were analysed (8 commercial, 2 in-house). Data were available for 4/10 EGCS packages from two centres and 6/10 from one. 14 centres completed the first proforma, of which 13 (including one RCPCH-API user) completed the second. For birthweight < -2SDS calculation, we observed high variability: CV 101% at 27-weeks; 7/12 RCPCH concordant), 49% at 33-weeks (6/12) and 24% at 37-weeks (6/12). GA correction methods varied widely with only 50% correcting for GA in infants born between 36-42 weeks. RCPCH-API corrected up to and beyond 18 years. For height calculations < -2 SDS at 11m, 23m and 36m for a child born at 27-weeks' gestation, CVs were 60% (3/12), 64% (3/12) and 79% (0/12), respectively. Inconsistencies were observed at 33-weeks' (22% (3/12), 9% (3/12) and 14% (1/12)) and 37-weeks' gestation (8% (1/12), 6% (2/12) and 92% (1/12)). HC variability was greatest at +1-2SDS (CV 29-33%). For BMI SDS > 3.5, consistency was poor (33% (10/13); range 1.3-5.2 SDS).

Conclusion

Clear inconsistencies and lack of standardisation between EGCS growth assessments exist. This impedes short stature, small-for-gestational-age and severe obesity identification, hinders clinical decision-making for referral, investigation and treatment and introduces geographical inequalities. We highlight an urgent need to develop comprehensive national standards/a position statement for EGCS packages. DOI: 10.1530/endoabs.111.OC5.5

OC5.6

Practice patterns and treatment outcomes of growth-promoting therapies in CAH: insights from real-world data

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Background

Children with CAH are at risk of reduced adult height due to early bone maturation. Adjunct Growth promoting therapies aim to optimize height outcomes by delaying skeletal maturation and pubertal progression. This study evaluates real-world practice patterns and the impact of such therapies on growth trajectories in children with CAH.

Methods

We conducted a retrospective analysis of children with CAH due to 21 hydroxylase deficiency, who received adjunct growth-promoting therapy. Data from 19 countries was collected through the I-CAH registry.

Results

Of 162 children (92 males, 69 females), median age at diagnosis was 0.20 years (range 0.04-3.55). Aromatase inhibitors (AIs), GnRH analogues, growth hormone (GH), cyproterone acetate, and spironolactone were used in diverse combinations. Letrozole was the most commonly used AI (68%, $n = 36$), predominantly at a dose of 2.5 mg daily (97.2%), with a median treatment duration of 2.98 years (IQR 1.84-4.6). Letrozole monotherapy significantly improved both bone age (BA)-adjusted height SDS ($\Delta 0.81$, $P < 0.0001$) and height SDS relative to mid-parental height (MPH) ($\Delta 0.85$, $P = 0.0002$). Combined use of Letrozole with GnRH analogues also demonstrated significant gains (BA-adjusted height SDS $\Delta 1.27$, $P = 0.0078$; vs MPH $\Delta 1.27$, $P = 0.0312$). The positive effect of AIs correlated with duration of therapy in monotherapy use ($r = 0.70$, $P = 0.0006$). No significant changes in glucocorticoid dose or 17-OHP levels were observed with AI therapy. Cyproterone acetate ($n = 37$) was initiated at a median age of 6.4 years, with a median dose of 50 mg/day (IQR 50-75) over 3.56 years. GH ($n = 34$) and GnRH analogues showed variable results; GnRH monotherapy led to significant improvement in height SDS ($\Delta 0.576$, $P = 0.0036$). GH, whether alone or in combination, showed limited efficacy. Notably, a combination of cyproterone, GH, and GnRH resulted in a significant improvement (Δ height SDS 2.23, $P = 0.037$).

Conclusion

There is marked heterogeneity in global clinical practice regarding growth-promoting therapies in CAH. AIs and GnRH analogues, particularly in combination, appear most effective. These findings underscore the need for standardized international guidelines and long-term safety evaluation.

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OC5.7

A multi-centre study of adrenal recovery in children tapering corticosteroids (ARCTiC study)

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Background

Prolonged and high dose glucocorticoids can result in steroid induced adrenal insufficiency (SIAI). While guidance exists regarding who is at risk of SIAI, the evidence based remains limited. Similarly, evidence-based guidance for weaning children off steroids safely but in a timely manner are lacking.

Aims

Identify factors influencing SIAI and recovery in children treated with prolonged glucocorticoids.

Methods

A retrospective cohort study of paediatric patients who underwent their first Short Synacthen Test (SST) for suspected SIAI between 2018 and 2023 at three tertiary UK centres (Sheffield, Nottingham, London). Patient demographics, steroid formulation, dose and SST results were collected. Patients were categorised as 'sufficient', 'insufficient' or 'borderline' based on their first SST outcome. A physiological hydrocortisone-equivalent dose was defined as 10 mg/m²/day. SPSS and ANCOVA were used for multiple and logistical regressions.

Results

Among 146 patients who underwent 200 tests, 102 (51%) were 'sufficient', 75 'insufficient' and 23 'borderline'. The average wean to a physiological steroid dose was 78 and 112 days for the sufficient and insufficient groups respectively. Patients had a higher median peak cortisol on SST when weaned on prednisolone compared to hydrocortisone (461.1 vs 373.4 nmol/l, $P = 0.0012$). The management of steroid doses following a failed SST (whether a dose was continued or changed) did not significantly affect likelihood of success of the following SST ($P = 0.324$). Independent variables including age, sex, steroid formulation, treatment duration, total steroid duration, days since physiological dose reached, average dose 14 days prior to SST, IV methylprednisolone had no statistically significant effect on the likelihood of passing SST or on cortisol measurements. However, the sufficient group had a significantly higher mean age at steroid initiation and lower mean daily HC dose compared to the insufficient group ($P = 0.003$).

Discussion

An adequate response to the first SST is statistically significantly associated with a shorter steroid wean, weaning on prednisolone rather than hydrocortisone, and lower steroid dose at SST. Younger age and higher steroid dose are risk factors for SIAI. Further research is needed to clarify others factors influencing SIAI and to guide paediatric steroid weaning strategies.

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OC5.8

The effect of letrozole on bone age maturation and estimated adult height in patients with congenital adrenal hyperplasia

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Background

Attenuated adult height secondary to premature skeletal maturation is a potential complication of congenital adrenal hyperplasia (CAH) due to CYP21A2 mutation. Aromatase inhibitors such as letrozole may mitigate these effects by suppressing oestrogen production at the growth plate and slowing skeletal maturation. This study evaluated the safety and effectiveness of letrozole in improving adult height in male CAH patients with advanced bone age.

Methods

This retrospective case series included eight male patients with classic CAH and advanced bone age, who were treated with letrozole for a minimum of two years. Bone age, estimated adult height (EAH), EAH Z-score, hydrocortisone dosage and bone health index (BHI) were assessed at baseline and after two years of treatment. Statistical analyses were performed to determine the significance of changes in these parameters.

Results

Letrozole treatment was associated with a significant reduction in bone age Z-score from 2.89 at baseline to 1.52 at two years ($P = 0.0097$) and a corresponding improvement in EAH Z-score -0.84 to 0.08 ($P = 0.0023$). The mean EAH increased by approximately 7 cm at two years of treatment ($P = 0.0047$). There was no significant difference in mean BHI between baseline and 2 years of letrozole treatment ($P = 0.93$).

Conclusion

Letrozole may have a role in the management of male patients with CAH and advanced bone age, potentially improving adult height outcomes. Prospective

controlled studies are needed to validate these findings and further evaluate the long-term safety and efficacy of aromatase inhibitors in this population.

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Endocrine Oral Communications 2

OC6.1

Longitudinal evaluation of bone health index for assessment of bone health in duchenne muscular dystrophy

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Background

Duchenne muscular dystrophy (DMD) is a progressive neuromuscular disorder associated with osteoporosis and increased fracture risk. Dual-energy X-ray absorptiometry (DXA) is the gold standard for assessing bone density but can be limited in DMD by scoliosis and positioning challenges. The Bone Health Index (BHI), derived from automated analysis of hand radiographs using BoneXpert, may offer a practical alternative by estimating cortical geometry from the metacarpals. To date, no longitudinal data on BHI in DMD have been reported.

Methods

We conducted a retrospective longitudinal study of 34 boys with DMD who underwent same-day DXA scans and left-hand radiographs at two timepoints. DXA outcomes included lumbar spine bone mineral apparent density (LS-BMAD) Z-scores and total body less head bone mineral content adjusted for bone area (TBLH-BMC) Z-scores. Results are reported as median (range); $P < 0.05$ was considered significant.

Results

At baseline, BHI Z-scores correlated positively with TBLH-BMC ($r = 0.65$, $P < 0.0001$) and LS-BMAD Z-scores ($r = 0.46$, $P < 0.05$). At baseline, median age was 10.0 years (4.6 to 17.2); 79% were ambulant. All were on glucocorticoids (33 on daily regimens), with 12% on bisphosphonates. At follow-up (median age 13.6 years), ambulation declined to 47%, all remained on glucocorticoids (32 daily), and 56% were on bisphosphonates. Median BHI Z-scores declined from -1.5 (-3.6 to +0.9) to -2.3 (-5.5 to +0.6) ($P < 0.001$), and TBLH-BMC Z-scores decreased from -1.5 (-2.3 to +1.4) to -1.8 (-3.2 to -0.8) ($P < 0.001$). In contrast, LS-BMAD Z-scores increased from -1.3 (-3.0 to +2.7) to -0.8 (-3.5 to +4.3) ($P = 0.038$), driven by improvement in the subgroup ($n = 16$) who commenced bisphosphonates post-baseline ($P = 0.002$). No change was seen in those already on or never on bisphosphonates.

Conclusion

BHI Z-scores deteriorated over time in boys with DMD, mirroring changes in TBLH-BMC Z-scores and reflecting disease progression and loss of ambulation. Unlike LS-BMAD, BHI did not improve with bisphosphonate therapy, but may be useful for tracking bone health prior to treatment initiation. Further research is needed to evaluate the utility of BHI Z-scores to predict fractures in DMD.

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OC6.2

Retrospective review of remission and relapse in paediatric graves' disease

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Introduction

Graves' disease (GD) is relatively rare in children. Medical therapy with carbimazole remains the mainstay of management. While longer treatment duration has been suggested to increase remission rates, evidence in the paediatric population remains limited. Our aim was to evaluate remission and relapse rates in paediatric GD and to identify factors influencing treatment outcomes.

Methods

We identified children with GD managed in our paediatric endocrine department between October 2022 and February 2025 and retrospectively reviewed their

records. Those treated with carbimazole for ≥ 18 months were included. Data were analysed to assess variables affecting disease remission (discontinuation of carbimazole) and relapse (need to recommence carbimazole).

Results

Ninety children (69% female) were included, with a mean (range) age at diagnosis of 10.7 (2.9–16.2) years. Mean (range) follow-up duration was 4.9 (2–14.1) years. Mean (range) carbimazole treatment duration was 4.3 (1.7–14.1) years; all were managed with dose-titration regime. Twenty-eight children (31.1%) achieved remission after a mean treatment duration of 3.9 years (range 1.7–9.8). Duration of carbimazole treatment did not correlate with remission rates: remission rates for treatment ≤ 3 years, 3–5 years, and > 5 years were 38.7%, 29.4% and 24% respectively ($P = 0.446$). Among those who achieved remission, 53.6% (15/28) experienced relapse after a mean of 12.8 months (range: 5 months–2.8 years). Duration of carbimazole treatment did not predict risk of relapse ($P = 0.325$). Eighteen percent (16/90) had definitive treatment (thyroidectomy [13/16]; radioactive iodine [3/16]) either due to non-remission after mean 5.3 years [13/16]; or relapse after remission [3/16]. No significant differences were observed between remission and non-remission groups regarding age, sex, ethnicity, or TSH receptor antibody levels at presentation. Free T3 at presentation was higher in patients who did not achieve remission compared to those who did (29.1 vs. 16.1 pmol/L, $P = 0.035$) and in those who relapsed after remission compared to those who did not (25.6 vs. 14.8 pmol/L, $P = 0.022$).

Conclusion

Prolonged carbimazole therapy was not associated with higher remission rates. Relapse was common amongst those who initially achieved remission. In this cohort, higher free T3 at diagnosis was associated with lower remission rates and higher relapse risk, suggesting prognostic significance of the degree of T3 elevation.

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OC6.3

Evaluating fibroscan as a screening tool for metabolic dysfunction-associated steatotic liver disease in children and young people with obesity

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Introduction

Transient elastography (TE), commonly known as Fibroscan, is a quick, non-invasive technique that employs pulse-echo ultrasound to evaluate liver fibrosis. While its effectiveness is well-established in adults, limited research has explored its application in children. The controlled attenuation parameter (CAP), a component of TE, identifies hepatic steatosis $> 10\%$ and grades its severity in adults. This study aimed to assess the feasibility and clinical utility of TE-CAP in identifying metabolic dysfunction-associated steatotic liver disease (MASLD: altered liver enzymes, fatty liver in ultrasound & metabolic risk factor) among paediatric patients with obesity at our centre.

Methods

Fibroscan was performed over a 6-month period in our outpatient clinics. Controlled Attenuation Parameter (CAP, in dB/m) was used to assess liver fat content, while Liver Stiffness Measurement (LSM, in kPa) was used to evaluate fibrosis.

Results

111 (53 male) patients, mean age 11.7 years (8.3–16.7) patients, mean BMI SDS 3.6 ± 0.6 underwent the fibroscan. Average scan time 8.60 minutes. BMI SDS was significantly higher in patients with steatosis (3.6 ± 0.6) v/s those without (3.2 ± 0.9 , $P = 0.047$). LSM values were higher in the MASLD patients (7.01 ± 2.6 v/s 6.15 ± 2.2 kPa, $P = 0.065$), suggesting an increasing trend towards liver stiffness. Patients with MASLD exhibited significantly higher CAP scores (285.5 ± 46.5) compared to those without MASLD (265.3 ± 54.8 ; $p = 0.048$), reflecting increased hepatic fat accumulation. Weight SDS ($r = 0.314$, $P = 0.001$) and BMI SDS ($r = 0.236$, $P = 0.013$) showed significant and positive correlation with CAP. Similarly, LSM demonstrated positive correlations with weight SDS ($r = 0.322$, $P = 0.001$) and BMI SDS ($r = 0.187$, $P = 0.049$), suggesting a positive relationship between elevated anthropometric indices and increased liver stiffness. The scan was well tolerated by children with neurodevelopmental and learning difficulty concerns.

Conclusion

CAP emerged as a promising marker of hepatic steatosis in children with obesity. Fibroscan proved to be a practical, reliable tool for screening steatosis and fibrosis in paediatric patients with obesity. Large scale studies are required to define the relationship between liver biomarkers and fibroscan measurements as potential predictors of MASLD in children with obesity.

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OC6.4

Systematic reassessment of children with congenital hypothyroidism in north west england through development of a multi-centre clinical network of paediatricians

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Background

Some children diagnosed with Congenital Hypothyroidism (CHT) have a transient form of the condition and do not require lifelong levothyroxine. It is currently recommended that children without a definitive diagnosis of permanent CHT should be re-evaluated at age 3 years. Studies investigating transient CHT either have high exclusion rates from drop-out in follow-up, or are small single centre studies. In Manchester, after initial assessment at the referral centre (Royal Manchester Children's Hospital), subsequent follow-up is at the local hospital managed by a clinical network of paediatricians linked with the referral centre.

Methods

All children diagnosed with CHT following newborn screening (NBS) in Manchester were followed up with an annual survey to the responsible paediatrician. At the age of 3 years, children with gland in-situ, levothyroxine dose < 50 $\mu\text{g/day}$ and TSH below the top of the reference interval were recommended for re-evaluation.

Results

Over a 4-year study period (April 2015 to March 2019), 217,696 babies were screened for CHT and 183 children received a CHT suspected result from NBS (1 in 1190). After initial assessment, 159 children were started on levothyroxine (1 in 1369). Seven children were excluded over the study period because they moved out of the area or died; one child was lost to follow-up. Of the 152 children included, 68 were deemed suitable for re-evaluation as per pre-determined criteria. Following re-testing, 50 (74% fitting eligible criteria, 33% for whole group) eligible children had a successful trial off levothyroxine and did not require further outpatient review. However, 18 re-commenced medication and continued long-term outpatient review.

Conclusion

A clinical network approach to systematic follow-up of longer term CHT outcomes enabled longitudinal data collection in $> 95\%$ of a cohort referred to the NBS programme in Manchester with finding of a significant proportion (33%) with transient CHT, optimising compliance with national guidelines. Roll out of a similar system across the UK could be facilitated by digital implementations like that used for sickle cell disease newborn outcomes.

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OC6.5

Boys with klinefelter syndrome referred to paediatric endocrine clinics have a much higher range of complexities than those in the community

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Introduction

Most of the information about childhood Klinefelter syndrome (KS) comes from newborn-screening follow-up programmes. A recent study of developmental progress in antenatally-identified KS boys has shown little difference from the reference population. Therefore, how do those boys referred to paediatric clinics differ from those in the community? How should care be planned in endocrine clinics?

Patients and Methods

A multidisciplinary clinic was established in 2009 for KS boys and extended with a joint adult transition service in 2015. 133 antenatal and postnatal referrals were received. The clinic database was analysed from 2009–2024 to examine the referral demographics and requirements for medical and other interventions.

Results

In 133 boys, the karyotypes were: 47,XXY ($n = 110$, 82.7%); 48,XXXY ($n = 2$, 1.5%); 49,XXXXY, ($n = 9$, 6.8%); 48,XXYY ($n = 10$, 7.5%); mosaic 46,XY/47,XXY ($n = 1$, 1%); 47,XXYY ($n = 1$, 1%). In the 47,XXY boys ($n = 110$), [excluding the 21 (15.8%) diagnosed antenatally], the indication for karyotyping was: learning, speech or developmental delay 72 (65.5%); ambiguous genitalia 7 (6.4%); pubertal delay 2 (1.8%); dysmorphic features 8 (7.3%). In these XXY boys, 78 (71%) had learning difficulties, an Education, Health and Care Plan (EHCP) being required in 51 (46.4%). 57 (51.8%) had speech delay, 34 (31%) receiving therapy. Whereas in those diagnosed-antenatally ($n = 21$), only 4 (19%) had learning difficulties, 3 (14.3%) requiring an EHCP, and only 4 (19%) had speech delay needing therapy. Almost all the boys with complex karyotypes had speech and learning delay. Testosterone replacement therapy was required in 57.1% ($n = 75$), indications being:

micropenis 8.3% ($n = 11$), gynaecomastia 21.1% ($n = 28$), pubertal boost 15% ($n = 20$), low bone mineral density 6.0% ($n = 8$), and low testosterone levels 6.0% ($n = 8$). 53.4% ($n = 71$) had reached adult height, median 180 cm (IQR: 11.9), range 152.5-200 cm. The median follow-up was 4.8 years (IQR: 5.5) range 0.31-12.4 years. In 58% ($n = 77$), transition to adult endocrinology had been completed.

Conclusions

The range of additional complex needs of boys with KS and variants referred to paediatric endocrine clinics differs markedly from those of unrecognised KS boys in the community and those antenatally-diagnosed. This highlights the need for establishing both specialist and transition clinics with multidisciplinary input.

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Diabetes Oral Communications 1 OC7.1

Revealing metabolic clues through the use of continuous glucose monitoring

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Introduction

Assessing metabolic complications in children and young people (CYP) with obesity can be challenging. In paediatric practice, standardised tests and clear cut-offs for diagnosing insulin resistance remain limited. A Homeostatic Model Assessment for Insulin Resistance (HOMA-IR) cut-off > 3.42 has been recommended as identifying those at risk of cardiometabolic risk factors. Total insulin levels (TIL) $> 300\mu\text{IU/ml}$ during a five-sample oral glucose tolerance test (OGTT) suggest hyperinsulinism. A 60-minute glucose level $> 155\text{ mg/dl}$ during an OGTT is a predictor of diabetes risk. The aim of our study was to assess the role of continuous glucose monitoring (CGM) in identifying metabolic risk compared to traditional methods.

Methods

31 patients (16 F) with an average age of 13.8 years (9.41-16.74) and mean BMI SDS of $+3.49 (\pm 0.45)$ were involved in a pilot study looking at glycaemic control in childhood obesity. The participants attended two visits for a five-sample OGTT and CGM insertion. The TIL, HOMA-IR and 60-minute glucose values were compared to average blood glucose (BG) and percentage time in range (TIR) on CGM.

Results

The average HOMA-IR ($n = 59$) was 6.4 ± 3.2 . 89.8% of the patients had a value of > 3.42 . When compared to CGM data, HOMA-IR was positively correlated with average BG ($r = 0.08$; $P = 0.53$) and negatively correlated with TIR ($r = -0.06$; $P = 0.66$). TIL were calculated ($n = 37$) with a mean of $785.4\mu\text{IU/ml} (\pm 315.7\text{SD})$. 97.3% of the results were $> 300\mu\text{IU/ml}$. The TIL was negatively correlated with average BG ($r = -0.13$; $P = 0.45$), but unexpectedly positively correlated with TIR ($r = 0.16$; $P = 0.34$). The mean 60-minute glucose ($n = 42$) was $121.3\text{ mg/dl} (\pm 24.7\text{SD})$. Interestingly, only 9.5% of the participants had a level $> 155\text{ mg/dl}$. Analysis revealed a positive correlation with average BG ($r = 0.29$; $P = 0.07$) and negative correlation with TIR ($r = -0.15$; $P = 0.36$). The TIR was significantly correlated with BG ($r = -0.84$; $P < .001$) on all three data sets.

Conclusion

These findings highlight the potential of CGM as a valuable tool for assessing glycometabolic status and risk factors in CYP with obesity. Traditional methods often require fasting, hospital admission, and multiple blood tests. CGM offers a less invasive alternative that may provide comprehensive metabolic information. While further research with larger cohorts is needed, these initial results are encouraging for the evaluation of metabolic complications.

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OC7.2

A novel capillary-adjusted home oral glucose tolerance test: a practical and reliable alternative to the gold standard venous method in childhood early-stage type 1 diabetes

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Objectives

The gold standard oral glucose tolerance test (OGTT) is used to stage and monitor disease progression in children and young people (CYP) with early-stage type 1 diabetes (T1D), but is poorly tolerated, contributing to high dropout rates during follow-up. We assessed a novel capillary OGTT device (cOGTT), which transforms capillary glucose measures to a venous equivalent, as an accurate and feasible alternative in CYP.

Method

We undertook two studies:

1. The cOGTT was assessed for a) agreement to venous glucose, using Bland-Altman, and b) its ability to classify standard glucose thresholds at fasting (5.6 mmol/l, 7.0 mmol/l), and 120-minutes (7.8 mmol/l, 11.1 mmol/l) during an in-hospital OGTT, (1.75g/kg glucose load, maximum 75g), in 31 CYP across the spectrum of glycaemia (normoglycaemia ($n = 19$), dysglycaemia ($n = 6$), hyperglycaemia ($n = 6$)), (mean age 12.2 years [SD 2.9], 48% male).
2. Assessment of the feasibility and acceptability of the cOGTT in the home environment in 41 children with early-stage T1D. Acceptability was assessed by an age-appropriate questionnaire.

Results

Capillary glucose showed good agreement with venous glucose over the diagnostic range (3.5–15 mmol/l), with a small negative bias (-0.1 mmol/l , -2.47 to 2.31 mmol/l ; absolute mean, 95% limits of agreement). Capillary glucose was highly correlated to venous glucose (Spearman's $\text{Rho} = 0.83$, $P < 0.0001$). At 0 minutes, cOGTT had 100% (1/1) sensitivity/ 96% (24/25) specificity to detect venous glucose of $\geq 5.6\text{ mmol/l}$, and 75% (3/4) sensitivity/ 100% (26/26) specificity to detect venous glucose of $\geq 7.0\text{ mmol/l}$. At 120 minutes, cOGTT had 50% (2/4) sensitivity/ 94% (17/18) specificity to detect venous glucose of $\geq 7.8\text{ mmol/l}$, and 80% (4/5) sensitivity/ 100% (22/22) specificity to detect venous glucose of $\geq 11.1\text{ mmol/l}$. Of those that responded ($n = 30$), 83% (25/30) chose the cOGTT device as their preferred method for future glycaemic assessment. Ninety-percent (37/41) of cOGTT were successfully completed in the home setting. Ten-percent (4/41) of cOGTT were unsuccessful, 7.5% (3/41) due to device failures, 2.5% (1/41) due to participant issues. Ninety-eight percent (40/41) found completing the test at home acceptable.

Conclusions

The cOGTT device is a highly specific and acceptable alternative to venous sampling. The cOGTT offers a practical alternative to the standard in-hospital OGTT with the potential to improve adherence to testing.

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OC7.3

A quality improvement innovation to improve the early diagnosis of type 1 diabetes in children and young people in wales: has it made an impact?

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Introduction

In Wales, Children and Young People (CYP) are most likely to present with symptoms of Type 1 Diabetes (T1D) to primary care. Delayed diagnosis increases risk of developing life-threatening diabetic ketoacidosis (DKA). The incidence of DKA at diagnosis remains unchanged. Quality Improvement (QI) initiatives were introduced in Cardiff and Vale University Health Board (CAVUHB) in 2018 to address this. Collaborative work with primary care identified barriers to facilitate change. Initiatives included a referral pathway, sustained teaching programs and reflective learning promoted via feedback.

Objectives

This study aims to evaluate incidence of T1D and DKA in CYP across Wales over 6 years and study the success of QI initiatives to facilitate early diagnosis of T1D and reduce incidence of DKA at diagnosis.

Methods

Data analysis from the All-Wales Brecon Paediatric Diabetes Registry of all newly diagnosed CYP with T1D. Data was organised by health board, number of new T1D, age, DKA severity from pre-pandemic (1/4/18-31/3/20), pandemic (1/4/20-31/03/22) to post-pandemic (1/1/23-31/12/24). Analysis of CAVUHB data to study the incidence of delayed diagnosis, delayed presentation and feedback to primary care.

Results

Across Wales there was a 22% increase in new T1D diagnoses during the pandemic. Overall DKA incidence increased from 31% to 35% during the pandemic and post pandemic it was 33.6%. CAVUHB has the lowest mean DKA incidence over the 6 years (29%) compared to rest of Wales. There has been no increase in rate of DKA at diagnosis seen in CAVUHB over the 6 years. In CAVUHB there was improvement from pre-pandemic rate of delayed diagnosis from 17.5% to 6.7% during the pandemic and 7.9% post pandemic. The rest were delayed presentations. Post-pandemic, 81% of CYP were diagnosed promptly by capillary blood glucose tests in primary care.

Conclusions

Over the last years, incidence of DKA at diagnosis remained high. CAVUHB demonstrated a lower incidence and improvement in prompt diagnosis following the QI initiatives. Delayed presentations are a concern. Further data analysis, feedback, training and renewed public awareness campaigns across all health boards in Wales is planned.

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OC7.4

Fast-tracking success: early hybrid closed loop initiation dramatically improves hba1c in the first year of care in CYP with type 1 diabetes

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Introduction

Achieving HbA1c 48 mmol/mol or below is recommended within the first year of care (FYOC), to promote metabolic memory and reduce long-term complication risk. NICE now advocates managing type 1 diabetes (T1D) in children and young people (CYP) with hybrid closed loop (HCL) technology.

Methods

The Leeds CYP Diabetes team have embraced HCL technology with consideration moving earlier in their FYOC pathway since 2023. The current pathway aims for HCL initiation within 4 weeks of diagnosis. CYP diagnosed with T1D have been prospectively followed over the past 3 years with HbA1c collected at 3 monthly intervals within the FYOC and 6 monthly thereafter. Data is included for patients diagnosed longer than 3 months. Time to HCL initiation has also been monitored.

Results

130 CYP have been diagnosed with T1D between June 2022-June 2025. 44 are currently in their FYOC. 87.7% are currently using HCL technology with 90.9% of those in their FYOC. Mean HbA1c for the current FYOC cohort has significantly reduced compared to the previous 2 years at each timepoint (table 1). This correlates with a reduction in mean time to HCL initiation (table 2). Mean time to HCL initiation has further reduced to 2.9 weeks over the past 6 months.

Conclusion

Earlier HCL technology use has positively impacted mean HbA1c supporting the emerging evidence for HCL use within the FYOC to promote metabolic memory.

Table 1

Time Since Diagnosis	Mean HbA1c (mmol/mol)							
	Current	3m	6m	9m	12m	18m	24m	30m
3-12m (n = 37)	47.3 (n = 37)	47.3 (n = 35)	50.9 (n = 22)	50.2 (n = 13)	49.7 (n = 6)			
12-24m (n = 47)	53.1 (n = 47)	53.2 (n = 46)	53.5 (n = 44)	55.8 (n = 46)	53.8 (n = 44)	53.9 (n = 25)		
24-36m (n = 39)	55.2 (n = 38)	51.3 (n = 33)	52.6 (n = 34)	56.7 (n = 36)	58.6 (n = 37)	54.6 (n = 36)	56.7 (n = 34)	56.1 (n = 23)

Table 2

Time Since Diagnosis	Mean Time to Pump Start (weeks)
0-12m	7.8
12-24m	30.6
24-36m	64.3

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OC7.5

Efficacy and safety of a tubeless AID system compared with MDI in children and adolescents with T1D in the RADIANT study

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Background and Aims

Despite advances in technology, many children and adolescents with type 1 diabetes (T1D) continue to use multiple daily injections (MDI), with glycaemic outcomes remaining at suboptimal levels. This analysis evaluated outcomes in paediatric participants with T1D from the RADIANT study, a randomized controlled trial comparing the Omnipod® 5 Automated Insulin Delivery (AID) System with MDI and CGM therapy.

Methods

Children and adults aged 4-70y with T1D ≥1y and screening HbA1c 58-97 mmol/mol currently using MDI with a FreeStyle Libre 2 CGM (study CGM) for ≥3 months were enrolled. Participants completed 14 days of data collection with MDI+CGM, then were randomly assigned 2:1 to intervention (Omnipod 5 + CGM) or continued with MDI+CGM for 13 weeks (control). Secondary and exploratory endpoints included glycaemic and clinical outcomes in paediatric participants.

Results

A total of 109 paediatric participants (median age: 12y; 43% female) were randomly assigned to intervention (n = 73) or control (n = 36). After 13 weeks, HbA1c improvements were observed with AID compared with control (adjusted mean difference: -9 mmol/mol, P < 0.0001), with a higher proportion of participants achieving HbA1c <53 mmol/mol at study end with AID, P < 0.01. Similarly, time in range (3.9-10.0 mmol/l) was greater with AID, with an adjusted mean difference (95% CI) of 20.5% (17.5, 23.4; P < 0.0001), corresponding to +4.9 hours per day in range. There were no significant differences in time <3.0 mmol/l and <3.9 mmol/l with AID compared with control (median 0.44% compared with 0.18% and 2.92% compared with 3.31%, respectively, both P>0.05). No episodes of severe hypoglycaemia or diabetic ketoacidosis occurred in either treatment group.

Conclusions

These results demonstrate the efficacy and safety of direct transition from MDI + CGM therapy to the Omnipod 5 System in children and adolescents with T1D not meeting glycaemic targets.

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Endocrine Oral Communications 3

OC8.1

The use of medtech in empowering families within a complications of excess weight clinic: the evira® app

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Introduction

The West Midlands Complications of Excess Weight (CEW) clinic is an NHS England-funded pilot aimed at improving the health, well-being and quality of life of children and young people (CYP) living with weight-related health complications. A MedTech device (Evira®), consisting of a smartphone App and an electronic weight measurement scale, became available to our service in 2024 as a weight management intervention. It involves patients taking daily weight measurements, which are presented as a trend within the App to monitor individual progress. Instant messaging within the App enables frequent communication with families and the CEW team to encourage and reflect on progress together.

Method

The technology was discussed with all eligible patients in the CEW service and offered at every face-to-face contact. Patients and families were educated on the technology prior to commencing the intervention. All members of the CEW team were trained in monitoring the patient's progress remotely and providing regular messaging to patients.

Results

Since May 2024, a total of 96 patients have started the intervention. For patients that have used Evira for 6 months ($n = 44$), there has been a mean average change of BMI z-score of -0.13 ($SD \pm 0.03$) compared to baseline. 23 patients paused or stopped using Evira within the first 6 months of treatment, mainly due to a lack of engagement or difficulty in sustaining changes. We have not encountered any significant technological barriers in our patient cohort.

Case Study

A 4-year-old child in our service was initially struggling to implement lifestyle changes. After introducing Evira, the CEW team sent encouraging messages and the patients BMI z-score reduced by 0.68 over 7 months.

Conclusion

The use of Evira MedTech empowers some patients within the CEW service to observe their progress on their weight loss journey and implement changes according to the direction of their trend. It also allows members of the CEW team to support families between clinic appointments and at a point in time when they require assistance. Enabling this frequent contact with the CEW team is particularly useful for families living far away from the hospital.

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OC8.2

Genotype-endocrine phenotype associations in a large cohort of children with NF1

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Introduction

NF1 is an autosomal dominant condition affecting 1 in 2500 births and can be associated with short stature and macrocephaly. Although genotype-phenotype associations have been studied for other phenotypes of NF1, endocrine phenotypes have never been previously explored.

Methods

Retrospective data analysis was conducted in children with NF1 known to a quaternary centre. Data on demographics, pubertal status, height, weight, body mass index (BMI) and head circumference (HC) was recorded, with the latter measurements converted into standard deviation scores (SDS). Data on mutation type was collected and grouped accordingly: frameshift, nonsense, missense, intronic, miscellaneous intragenic and whole gene deletions.

Results

201 patients (97 girls, mean age 10.0 years) were included. Table 1 demonstrates the mean height, weight, BMI and OFC SDS by mutation type, with no significant difference generally between groups on ANCOVA analysis, except with BMI SDS. Independent T-test comparing the whole gene deletion to other types of mutations demonstrated significantly taller stature ($P = 0.027$), greater weight (P

$= 0.020$) and greater BMI ($P = 0.022$). Pubertal issues (precocious or delayed) were only observed in those with frameshift (10.2%), nonsense (8.2%) or intronic (7.1%) mutations.

Conclusion

Similar growth phenotypes are observed across mutation types, with the exception of whole gene deletions, due to a gene within the microdeletion whose haploinsufficiency may cause overgrowth, which has been suggested previously. The variability in BMI SDS observed may be due to genuine but non-significant differences in height and weight SDS between other groups, which warrants further work with larger groups.

Table 1

		Frameshift	Nonsense	Missense	Intronic	Misc intragenic	Whole gene deletion	p-value
Height SDS	Mean	-0.8	-0.8	-0.8	-0.7	-0.8	-0.2	0.57
	SD	1.3	1.1	1.4	1.4	1.6	1.02	
Weight SDS	Mean	-2.0	-0.2	-0.6	-0.1	0.5	0.8	0.13
	SD	2.3	1.4	1.4	1.2	1.2	1.5	
BMI SDS	Mean	0.2	0.3	0.0	0.4	1.2	1.04	0.045
	SD	1.2	1.5	1.2	1.2	0.9	1.3	
HC SDS	Mean	0.7	1.3	0.8	1.3	1.1	0.7	0.45
	SD	1.3	1.7	1.7	1.3	0.7	1.5	

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OC8.3

Quantification of 11-oxyandrogen biomarkers from dried blood spots using tandem mass spectrometry

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Introduction

The 11-oxygenated androgens are elevated in congenital adrenal hyperplasia (CAH). These steroid biomarkers can be quantified by LC-MS/MS, which has high specificity and allows multi-analyte analysis from a single sample. Dried blood spots (DBS) are minimally invasive and easy to collect, transport and store. They are widely used for monitoring CAH disease control and represent a useful matrix for steroid analysis. Volumetric micro-sampling devices are increasingly available - they collect a fixed volume of blood for analysis and are reported to be more accurate than traditional dried blood spots.

Aim

This study aimed to develop and validate a multiplex LC-MS/MS assay for seven analytes (cortisol, 17-OHP, androstenedione, testosterone, 11-ketotestosterone (11-KT), 11-hydroxyandrostenedione (11-OHA4) and progesterone) from dried blood spots. In addition, we compared W-903 filter paper to the volumetric Capitainer®B10 device.

Method

We developed a LC-MS/MS assay using commercially available standards. The extraction procedure, liquid chromatography method and ionisation conditions were optimised to improve sensitivity. Data were analysed in Skyline and validation metrics calculated in R (version 4.5.1). Standard curves were created over the quantitation range of the assay, to assess linearity, the limits of detection (LOD) and quantitation (LOQ) and the matrix effect. Quality control samples were analysed to determine the accuracy and precision of the assay.

Results

A linear relationship ($R^2 \geq 0.99$) was observed at a concentration range of 0 – 215 ng/ml or 0 – 500 ng/ml depending on the analyte and matrix (Capitainer® or W-903 filter paper). The matrix effect was $> 100\%$ for all analytes. LODs ranged between 0.06-0.47 ng/ml (Capitainer®) and 0.05 – 0.53 ng/ml (W-903). LOQs ranged between 0.18-1.1 ng/ml (Capitainer®) and 0.12 – 1.35 ng/ml (W-903). Intra-batch and inter-batch precision and accuracy coefficients of variation (CVs) were $\leq 20\%$ for analytes, except 11-OHA4 using W-903 filter paper.

Discussion

We report a multiplex LC-MS/MS method specifically designed for measuring relevant biomarkers, including 11-KT and 11-OHA4, from paediatric CAH patients using DBS samples. The assay achieved a clinically useful LOQ using both Capitainer® and W-903 filter paper. The method is accurate and precise for all analytes using the Capitainer® device. Further studies involving patient samples are planned.

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OC8.4

The IMPACT survey in the UK: treatment patterns in osteogenesis imperfecta

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Background and aims

The treatment landscape of osteogenesis imperfecta (OI)—a rare hereditary connective tissue disorder—is not fully understood. This study aimed to capture real-world insights on pharmacologic interventions in people with OI living in the United Kingdom.

Methods

The IMPACT Survey was developed by the OI Federation Europe, the OI Foundation and an international steering committee of experts to explore the clinical, humanistic and economic impact of OI. The survey was fielded online from July–September 2021 and open to individuals with OI, caregivers and other close relatives. This descriptive analysis presents findings from British adults with OI and caregiver-reported data on children aged ≤ 18 years.

Results

Data on treatment patterns were collected from 144 adults (mean age 46.9 years; 73.6% female) and 27 children by proxy (mean age 5.9 years; 33.3% female) with OI. Most respondents reported their OI as Type I (47.2% adults; 33.3% children) or rated OI severity as moderate (42.4% adults; 44.4% children), though distribution across types and severity was spread. Many individuals were taking, or had previously taken, vitamin D or calcium supplementation (adults 92.3% and 65.3%; children 96.3% and 44.4%, respectively). Current or prior bisphosphonate use was reported by 66.7% adults and 66.7% children. Of those currently taking bisphosphonates (38 [26.4%] adults; 18 [66.7%] children), most were receiving zoledronate (36.8% adults; 50.0% children). Some adults were receiving risedronate (21.1%), alendronate (18.4%), pamidronate (13.2%) or ibandronate (2.6%). Some children were receiving pamidronate (44.4%) or risedronate (5.6%). Current or prior use of other pharmacologic treatments included: muscle relaxants (15.9% adults; 14.8% children), parathyroid hormone (9.1% adults), oestrogen (7.0% adults), and antibodies (e.g., denosumab, romosozumab, setrusumab; 4.2% adults). Pain medication was regularly used by 61.8% of adults and 33.3% children, with 43.8% adults using it daily.

Conclusion

The IMPACT Survey provides unique patient-reported perspectives on the British OI treatment landscape highlighting use of a wide-range of pharmacologies. Data indicate prevalent prescription of bisphosphonates in children and adults and emphasise the importance and need of pain management in individuals of all ages.

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OC8.5

Evaluating the predictive utility of basal cortisol for synacthen test outcomes in neonates: a retrospective cohort study

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Background

Adrenal insufficiency in neonates presents a diagnostic challenge due to non-specific clinical signs, physiological immaturity of the hypothalamic-pituitary-adrenal axis, and lack of established gestation-specific reference intervals or cutoff values for random unstimulated cortisol levels. Unstimulated (basal) cortisol is frequently measured in neonates as a preliminary screen for adrenal insufficiency. However, its discriminatory value in predicting adrenal response on Short Synacthen Testing (SST) remains poorly established.

Objective

To evaluate the discriminatory performance of unstimulated serum cortisol in predicting SST outcomes in a neonatal cohort.

Methods

This was a retrospective cohort study which included neonates admitted to a tertiary neonatal intensive care unit over a five-year period (2020–2024) who underwent both basal cortisol testing and SST. SST “pass” was defined as peak

serum cortisol ≥ 450 nmol/l at both 30 and 60 minutes post-ACTH stimulation. Receiver Operating Characteristic (ROC) curve analysis was performed to assess discriminatory performance, and optimal thresholds derived using the Youden Index.

Results

A total of 47 neonates were included, with a mean gestational age of 32.8 ± 0.8 weeks and a mean birth weight of 1846 ± 166 g. Sixty-six percent were male, and the cohort included 36.2% small-for-gestational-age (SGA), 59.6% appropriate-for-gestational-age (AGA), and 4.3% large-for-gestational-age (LGA) infants. The most common clinical indication for cortisol testing was hypoglycaemia (42.3%), followed by hyperbilirubinaemia (17.9%), clinical suspicion of adrenal insufficiency (16.7%), differences in sex development (6.8%), concerns regarding pituitary abnormalities (6.4%), and other causes (9.8%). The area under the ROC curve was 0.640, indicating limited discriminatory ability of basal cortisol to predict an adequate cortisol response following ACTH stimulation. A cortisol threshold of 183.5 nmol/l achieved high specificity (90%) but low sensitivity (33%), with a Youden Index of 0.233.

Conclusion

Basal cortisol demonstrated limited utility in predicting adrenal response to ACTH stimulation in our neonatal cohort, with poor sensitivity. These findings underscore the inadequacy of relying on basal cortisol alone in the evaluation of neonatal adrenal function. Future research should focus on establishing gestation-specific reference intervals for cortisol to improve the interpretation of basal values and support more accurate and timely diagnosis of adrenal insufficiency in this vulnerable population.

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Diabetes Oral Communications 2

OC9.1

Developing a national schools training platform transforming diabetes awareness in schools across the uk to meet the needs highlighted in the ‘The INSCHOOL’ project

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Background & Objectives

Children and Young People (CYP) living with diabetes often face significant challenges managing their condition in school. Evidence shows that CYP with diabetes attain GCSE results that are lower than peers without the condition (Diabetes Care, 2022;45:2852–2861). Additionally, stigma and challenges managing diabetes were highlighted in The INSCHOOL project (J Adolesc. 2024 Feb;96(2):337-349). Previous diabetes training materials, including the schools e-learning module from Breakthrough T1D and the National Children and Young People's Diabetes Network, were over 8 years old and lacked content on newer technologies and the emotional impact of living with diabetes.

Methods

To address these gaps, DigiBete, the National Children and Young People's Diabetes Network, Breakthrough T1D, and the Together Type 1 Programme at Diabetes UK collaborated with families, teachers, and healthcare professionals to co-design a refreshed training platform: www.diabetesinschools.org. The updated resource places young people's and families' lived experiences at the centre of learning and support and is underpinned by The INSCHOOL project Development activities included:

- Eight task-and-finish groups ($n = 8$) with families and healthcare professionals to evaluate and redesign the previous module
- Three days ($n = 3$) of filming to capture authentic lived experiences
- Three one-hour reflective workshops ($n = 3$) with healthcare professionals to review new materials
- Secondary analysis of the INSCHOOL data

Results

Since its launch nine months ago, the platform has seen strong engagement and positive feedback:

- 61,000 users visited the website
- 23,256 people registered on courses
- 80% assessment pass rate, with an average score of 91.62%
- 92% of users surveyed reported feeling more confident in supporting diabetes care in schools

Conclusions

This national platform highlights the need for consistent, standardised diabetes education in schools. Despite pressures, schools are keen to engage when training is accessible and clearly benefits students' wellbeing, helping them feel safe and

enabling them to thrive academically, socially, and emotionally. Diabetes teams are also reporting reduced time and cost burdens, supported by a flipped learning approach that enriches personalised care training when working with schools.

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OC9.2

Integrated, patient-centred, holistic transition model improves physical and psychosocial outcomes of young adults (16-25) with diabetes - transition safe and sound (TraSS), an NHS England pilot

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Background

Transition from paediatric to adult services represents a high-risk period for young adults (YA) with diabetes, often leading to disengagement and poor outcomes. Transition Safe and Sound (TraSS) is an NHS England-funded pilot designed to bridge this gap through a failsafe, multidisciplinary model.

Aim

To evaluate the clinical, psychosocial, and service-level impact of the TraSS model for YA aged 16 to 25 years.

Method

The pilot implemented a team including a failsafe officer, transition outreach specialist nurse, youth worker, dietician, psychologist and project manager. Integrated pathways were created (between paediatric and three adult services) to identify and support YA with recurrent non-attendance, high HbA1c, or classified as 'unable to contact' (UtC). Referrals were triaged and patients underwent assessment by youth worker and/or transition outreach nurse and targeted further support provided. Validated standardised screening tools were used for psychological screening. Contact data, engagement, and outcomes were analysed.

Results

211 patients were referred to the TraSS model. The primary reasons for referral included 35% for diabetes education, 21% for psychology support, 21% for youth worker support, 18% for high HbA1c support, 11% were uncontactable, and 11% were non-attenders. Following assessment, 45% of contacts were provided by youth worker, 33% by Diabetes Specialist Nurse, 12% by multiple members, 10% by failsafe officer, psychologist, or dietician. Of 24 patients on the UtC pathway, 83% were successfully contacted and 58% engaged with adult care. Psychological screening ($n = 81$) showed 65% reported moderate to severe diabetes distress; 28% reported severe distress and 12% showed disordered eating risk (including insulin omission). 36 peer group events involving 73 YA were delivered, including "Cook & Count" and psychosocial drop-in, with positive YA feedback. HbA1c levels ($n = 76$ paired samples) demonstrated a median reduction of 8.4 mmol/mol (and 5.8 mmol/mol, excluding newly diagnosed patients).

Conclusion

The TraSS model successfully supports young adults with diabetes, leading to improvements in both clinical outcomes (e.g. HbA1c) and psychosocial wellbeing. An integrated service enhanced continuity of care. Holistic, tailored, patient-centred support combined with psychological screening unveiled high levels of unmet emotional and psychosocial needs, underscoring the essential role of multi-disciplinary care during transition.

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OC9.3

Reducing 'was not brought' rates in paediatric diabetes clinic: empowering patient choice to improve service delivery

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Introduction

National guidance recommends each paediatric diabetes patient is offered four multidisciplinary team (MDT) clinic appointments annually. At University Hospital Southampton (UHS), we identified a higher "was not brought" (WNB) rate compared to other centres in our regional network. In response, we initiated a quality improvement project to reduce WNB rates.

Aim

To evaluate the impact of a patient consultation, and subsequent service improvement, on paediatric diabetes clinic attendance at UHS.

Method

In November 2024, we conducted a telephone survey with 27 families (10% of our cohort). Families with the highest WNB and cancellation rates were prioritised for inclusion. The consultation identified our fixed booking system as a key barrier to attendance. In response, from January 2025 we introduced a flexible booking system, allowing patients to choose the date and time of their next appointment. We conducted a retrospective service evaluation collecting clinic data, over two seven-month periods: before (1st December 2023–30th June 2024) and after (1st December 2024–30th June 2025) the intervention.

Results

Data were collected for 1,908 clinic bookings, and their associated outcomes. A logistic regression model was used to analyse the data.

- WNB appointments decreased by 50.6% (from 83 to 41)
- Patients were 2.3 times more likely to miss their appointment pre-intervention (OR = 2.26, 95% CI: 1.52–3.34, $P < 0.001$)
- Patients were 7.7 times more likely to book their own appointment post-intervention (OR = 7.67, 95% CI: 4.60–12.79, $P < 0.001$)
- The number of patients seen increased by 11.4% (from 516 to 575)
- The number of offered appointments remained stable (from 599 to 616)

Conclusion

Engaging families through consultation and implementing a flexible booking system led to a significant reduction in missed appointments and an increase in patient-initiated bookings. The number of patients seen increased, without expanding clinic capacity, demonstrating better utilisation of existing resources. These findings support the value of patient autonomy as a driver of service efficiency in paediatric diabetes care. Ongoing monitoring is needed to assess the sustainability of this change.

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OC9.4

Quantifying the effect of screening for childhood type 1 diabetes on HbA1c, DKA and hospitalisation at diagnosis: a systematic review and meta-analysis

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Aims

Screening for islet autoantibodies identifies individuals with early-stage type 1 diabetes (T1D) who will progress to stage 3, enabling staging, monitoring, support and education to be offered. This systematic review and meta-analysis aims to quantify the effect of screening for T1D on HbA1c, rate of DKA and hospitalisation at diagnosis.

Methods

Data were sourced from CINAHL, MEDLINE, Cochrane Central, Embase, PsychINFO, Web of Science. To be included, studies had to report outcomes of a screened and non-screened group at diagnosis of T1D and include children up to the age of 18. Random effects meta-analyses quantified the mean difference in glycated haemoglobin (HbA1c) and risk ratio (RR) for DKA. Where there was insufficient data (hospitalisation data), a narrative synthesis was undertaken. A risk of bias assessment was performed using the ROBINS-I tool. CRD42023394661

Results

In this study, 8353 studies were identified and 497 underwent full text review. Nine studies ($n = 29181$ children and young people diagnosed with T1D (screened = 730, non-screened = 28451)) were included comparing the difference in at least one of HbA1c, DKA, and hospitalisation between a screened and unscreened population. The mean (SD) age was 7.2 (2.6) years at screening. Seven studies reported data for HbA1c, six for DKA and three for hospitalisation. Screening for T1D was associated with a mean reduction in HbA1c at diagnosis of -31.8 mmol/mol [95%CI: -40.5; -23.1] ($P < .05$), a 34.6% change. The screened population were less likely to develop DKA at diagnosis, RR 0.18 [95%CI: 0.04; 0.44] ($P < .05$), than the non-screened population. Screening was associated with an absolute risk reduction in DKA at diagnosis of 23.9%. There was insufficient data for a meta-analysis of hospitalisation data, however in the studies retrieved ($n = 3$) screening was associated with reduced hospitalisation at diagnosis or a reduced mean stay in hospital.

Conclusions

Screening for T1D is associated with a 35% reduction in HbA1c and a risk reduction of 24% for diagnosis in DKA. Improvement in clinical outcomes at diagnosis has important benefits for children with type 1 diabetes, reducing the need for hospitalisation at diagnosis, and the psychological and physiological morbidity associated with DKA. Our study quantifies the benefits of screening.

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OC9.5

Mind the gap: ongoing inequalities in glycaemic levels in young people living with type 1 diabetes across england and wales

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Background

Utilising data from the National Paediatric Diabetes Audit (NPDA) for England and Wales, inequalities in glycaemic levels in children and young people (CYP) living with Type 1 Diabetes (T1D) across different ethnic and socio-economic groups were first highlighted in 2016. Almost ten years on, we wanted to know if this difference persists.

Methods

Analysis of 27,919 CYP with T1D from the 2022-2023 NPDA. Multivariable linear regression was used to assess any association between HbA1c and Socio-economic status (SES) or ethnicity, adjusting for age, gender, diabetes duration and pump use.

Findings

CYP from ethnic minority groups continue to have significantly higher mean HbA1c levels compared to white CYP with the largest difference in black children (6.8 mmol/mol, [0.6%], 95% CI 5.7, 7.9 mmol/mol). Lower SES remains significantly associated with higher HbA1c levels with the largest difference between the least deprived and most deprived (6.9 mmol/mol; [0.6%], 95% CI: 6.4, 7.6) (see Table 1) Additionally, CYP from ethnic minority groups (Black 34.2% vs White 49.56%, $P < .0001$) and those living in more deprived neighbourhoods (Most Deprived 41% vs Least deprived 54% vs $P = 0.0001$) were less likely to use insulin pumps.

Conclusions

Ethnicity and SES remained significant predictors of HbA1c level. Black CYP continue to have the highest glycaemic levels, and lower SES remains associated with poorer HbA1c outcomes. Substantial inequalities persist in the use of insulin pump therapy highlighting the ongoing need for targeted interventions to improve equity in diabetes care and outcomes.

Table 1

	White	Black	Least Deprived	Most Deprived
HbA1c mmol/mol	63.1 mmol/mol	69.9 mmol/mol	60 mmol/mol	66.9 mmol/mol
(SD)	(15.8)	(18.0)	(13.7)	(17.7)

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OC9.6

Analysing the impact of socioeconomic deprivation and ethnicity on the development of vascular complications in young people with type 1 diabetes – a regional perspective

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Background

Type 1 Diabetes is associated with high rates of morbidity and mortality due to diabetes-related vascular complications. A key modifiable risk factor is glycaemic control. Despite an improvement in HbA1c levels in young people in the UK, complication rates remain unchanged.

Aim

To explore whether an association exists between ethnicity and/or socio-economic status and vascular complications in young people with Type 1 Diabetes

Methods

Retrospective case note review from 1st April 2021 – 31st March 2024 for young people aged 12- 30 years with a diagnosis of Type 1 Diabetes, of ≥ 3 years duration in 2 large centres in the West Midlands, UK. Data collected included demographics, date of diagnosis, HbA1c, type of insulin regimen and development of vascular complications as per NICE guidance. Data was analysed using binomial generalised linear models. The null model adjusted for duration of diagnosis (years) and HbA1c (mmol/mol), and the selection variables of interest were ethnicity and index of multiple deprivation (IMD) quintile.

Results

Data was analysed for 425 patients; 52% of white ethnicity and 51% from IMD quintile 1. Retinopathy was the most observed complication (18%). HbA1c levels increased with the duration of diabetes diagnosis. The probability of developing any vascular complication increased with the duration of diabetes diagnosis [odds 1.16 for every year increase in duration (95% CI: 1.10, 1.22)], and HbA1c levels [odds 1.01 (95% CI: 1.00, 1.02) for every 1 mmol/mol increase in HbA1c]. There was a significant reduction in the development of diabetes-related complications between the most deprived IMD quintile, and the second most deprived quintile (-0.56, (95% CI -1.04); P 0.02). Ethnicity had no impact on the development of complications.

Discussion and Conclusion

The population analysed are from the most deprived socio-economic groups and more ethnically diverse compared to UK census data. Our study shows that increasing socio-economic deprivation increases the risk of developing vascular complications independent of duration of diagnosis and HbA1c. Further detailed evaluation of the factors contributing to this finding as well as a personalised approach to caring for the most deprived patients is essential to improve the outcomes for this cohort.

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OC9.7

A phase ii, international, randomised, double-blind, efficacy and safety trial of sodium valproate in paediatric and adult patients with wolfram syndrome

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Objectives

Wolfram syndrome (WFS1-Spectrum Disorder) is a rare (1:500,000) monogenic form of childhood onset diabetes mellitus and optic atrophy. There are no therapies to slow or stop disease progression. Sodium valproate prevents cell death in Wolfram neuronal models. We aimed to investigate whether valproate can slow disease progression in this condition.

Methods

We conducted a pivotal, randomised, placebo-controlled, multi-centre, double-blind trial of sodium valproate in patients with Wolfram syndrome defined by diabetes mellitus and/or optic atrophy diagnosed under 16 years; pathogenic variants in the WFS1 gene; and visual acuity assessed as a LogMAR score of 1.6 or better on an EDTRS chart. Patients were randomly assigned 2:1 to either treatment with sodium valproate or matched placebo, and followed up for progression of visual acuity loss for 36 months. Secondary outcomes included safety, and effects on glycaemic control.

Results

A total of 63 participants (median age 18 years, range 8-61 years; 29/63 (46% under 16 years) underwent randomisation, 42 to the valproate group and 21 to the placebo group. At baseline, the placebo treated group vs valproate treated group was younger (median age 15yrs (range 11-21yrs) vs 19 yrs (range 14-26yrs)), had

shorter duration of optic atrophy (median 5.9yrs (3.5-8.7yrs) vs 6.6yrs (3.9-10.9yrs)) and lower HbA1c (median 6.95% (6.4-7.96% vs 7.5% (6.9-8.3%)). There was not a significant effect of valproate on visual acuity (progression 0.0431 LogMar/yr both groups, $P = 0.66$) or HbA1c (-2 mmol/mol vs +1 mmol/mol), insulin dose adjusted A1c, or total insulin consumption per kg body weight between the groups. There was an excess of hypoglycaemic events in the valproate treated group (11/42(26%) vs 4/21(19%); $P < 0.05$).

Conclusions

This is the first multi-centre randomised controlled trial of a treatment for Wolfram syndrome. We did not observe an effect of valproate on the primary outcome or on glycaemic control. However we cannot exclude the possibility that this is due to the differences between the groups at baseline. Further analysis is ongoing to supplement the placebo group with an external cohort to make the groups comparable. The protocol was approved by the National Research Ethics Service (18/WS/0020). Trial registration no. NCT03717909.

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Endocrine Oral Communications 4

OC10.1

Application of multi-omic approaches to investigate novel LZTR1 gene variants and identify therapeutic targets implicated in the pathogenesis of Noonan syndrome (NS)

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Introduction

Noonan syndrome (NS) is caused by genetic variations which alter the RAS-MAPK pathway leading to hyperactivation of signalling. However, the pathogenesis of multisystem disease including short stature, is poorly understood. NS-affiliated missense *LZTR1* variants have been associated with defective ubiquitination of Ras leading to increased Ras substrate availability. We hypothesised that Ras dysregulation may attenuate p53 signalling and investigated the role of *LZTR1* in this pathway.

Methods

Two patients with short stature and NS underwent whole exome sequencing revealing the presence of dominantly inherited *LZTR1* variants. Both single nucleotide substitutions were generated by mutagenesis of an N-terminal MYC tagged-*LZTR1*-cDNA. Constructs were expressed in mammalian cells and lysates prepared for phosphoproteomics. Analysis of transcriptomic data was conducted using Ingenuity Pathway-Analysis. Significant phospho-peptides, protein-protein interactions and pathways of interest were probed using immunoblotting, immunofluorescence, nanoluciferase assays and *in-silico* modelling.

Results

Both heterozygous *LZTR1* variants were shown *in-vitro* to be thermodynamically stable and associated with elevated pan-Ras levels. Phosphoproteomics revealed upregulation of the histone acetyltransferase inhibitor, NUC2L, in both variants. This finding, consistent upon immunoblotting and immunofluorescence, was associated with impaired p53 acetylation. Effectors of the DNA damage response (DDR), ATM and CHK1 appeared enhanced in both *LZTR1* variants whilst two major substrates representative of their kinase activity, Rad50 and Adducin (ADD1/2), were preferentially phosphorylated, via residues Serine 635 and Serine 713/726 respectively. Despite apparent activation of the DDR and diminished p53 activity, levels of LC3 and phosphorylated-p70-S6-kinase were increased. *In-silico* structure modelling and nanoluciferase assays suggested that *LZTR1* interacts with NUC2L, an interaction previously unknown and disrupted in both *LZTR1* variants.

Conclusion

NUC2L and p53 form a complex which dictates p53 activation. We demonstrate a novel interaction between NUC2L and *LZTR1* and hypothesise that *LZTR1* modulates activity of this complex. NUC2L upregulation leads to p53-mediated transcription inhibition. *LZTR1* mutations potentiate NUC2L activity leading to reduced apoptosis and compensatory increases in autophagy that perpetuate chronic DNA damage gaining insights into NS pathogenesis. Currently, no inhibitors of NUC2L exist, but this complex represents a therapeutic target that warrants exploration. Future work will ascertain the role of NUC2L in other genetic causes of NS.

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OC10.2

Iatrogenic vitamin D toxicity within the paediatric population: an acute surge in national cases

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Introduction

Vitamin D supplementation has become increasingly widespread in the UK, increasing the risk of vitamin D toxicity, particularly among infants. While vitamin D is essential for calcium regulation and bone development, excessive intake can result in hypercalcaemia. Although vitamin D toxicity remains rare, an acute national surge prompted a review of cases. Relaxed regulatory standards for over-the-counter food supplements, a lack of clear treatment guidelines, and growing use of high-dose vitamin D for both supplementation and therapeutic purposes may lie behind this.

Methods

We present a case series of 12 children who developed vitamin D toxicity and were admitted to secondary and tertiary care centers across the UK between December 2024 - March 2025. In all cases, toxicity was attributed to formulation errors in prescribed vitamin D preparations.

Results

Age of presentation ranged from 2 months - 12 years, with 33% of patients ≤ 1 year. The most common presenting symptoms were vomiting (50%), lethargy (42%), altered bowel habits (33%), and poor feeding with weight loss (33%). The mean (SD) adjusted calcium on admission was 3.51 mmol/l (± 0.48). Mean (SD) serum 25(OH)D levels were 1281 nmol/l (± 462), although inter-assay variability across laboratories may limit the reliability of this measurement. All patients had suppressed parathyroid hormone (PTH) levels, confirming vitamin D-induced hypercalcaemia. Treatment was promptly initiated in all cases. Eleven of twelve patients (92%) received intravenous hyper-hydration. Seven (55%) received pamidronate infusions, with three requiring repeat doses. One patient received furosemide due to deteriorating renal function, and another was treated with tapering prednisolone. Seven (58%) patients required strict adherence to a low calcium diet. Two patients (18%) developed nephrocalcinosis, and four (33%) experienced acute kidney injury (AKI). All patients were receiving Vitamin D treatment prior to admission, with 92% prescribed a defective batch of Vitamin D3, which was subsequently withdrawn by the manufacturer.

Conclusion

This case series highlights Vitamin D toxicity as a serious but preventable cause of hypercalcaemia in children. Stricter regulation of food supplement formulations and the development of national treatment guidelines are urgently needed to minimise the risk of iatrogenic harm in this vulnerable population.

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OC10.3

The united kingdom adult northstar network consensus recommendations of transition of osteoporosis care and management of osteoporosis in adults with duchenne muscular dystrophy

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Background and objectives

Osteoporosis is a well-recognised complication in Duchenne muscular dystrophy (DMD). While international clinical guidance exist for paediatric management, there is currently no guidance for osteoporosis care during transition to adulthood and for long-term management of therapies initiated in childhood.

Methods

In 2023, a UK expert working group was convened to develop national consensus recommendations for managing osteoporosis in adults with DMD within the adult NorthStar Network. The group included 13 adult and 5 paediatric bone specialists, 3 adult neuromuscular clinicians (2 with paediatric experience), a clinician-scientist/densitometrist, and 3 patient representatives. Systematic and scoping reviews were conducted on DMD-specific osteoporosis therapies, DXA-based fracture risk prediction in DMD and adult glucocorticoid-induced osteoporosis guidelines. A survey of paediatric clinicians, distributed via patient organisations, explored transition-related bone care. A focus group with adults with DMD captured patient perspectives. Four virtual meetings of the expert panel were held. A core team developed the clinical guidance statements and consensus was reached using a modified Delphi process via online voting.

Results

Thirteen clinical guidance statements were developed following three Delphi voting rounds, with 80% agreement required. Key recommendations include:

- All adults with DMD should undergo fracture risk assessment, including DXA and vertebral fracture (VF) evaluation, where this would influence clinical decision-making.
- VF reassessment and DXA is recommended every two years for those on glucocorticoids and should be individualised in others, including those with spinal instrumentation.
- For individuals on bisphosphonates for > 10 years who have completed puberty, treatment discontinuation should be considered at transition based on risk factors (e.g., glucocorticoid use, existing or worsening VF, or recent low-trauma long bone fracture).
- If therapy is discontinued, re-evaluation is advised at two years or earlier if a low-trauma fracture occurs or bone density declines significantly.

This expert consensus guidance was endorsed by the clinical committee of BSPED in 2024.

Conclusion

This UK-wide expert consensus provides the first national guidance for managing osteoporosis in adults with DMD. The recommendations address key transitional care gaps and support a structured, risk-based approach to lifelong bone health in this population.

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OC10.4

Pubertal induction in girls with turner syndrome: updated retrospective data from the international TS registry

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Background

Current guidelines for girls with gonadal dysgenesis due to Turner Syndrome (TS) recommend initiating estrogen therapy at 11-12 years of age, using 'natural' 17- β estradiol. However, there is scant evidence regarding the optimal modality of induction, particularly concerning whether oral or transdermal routes are more effective or acceptable.

Aim

To retrospectively evaluate differences in puberty induction approaches and outcomes using anonymised data extracted from the I-TS module of <https://sdmregistries.org/>.

Methods & Patients

50 centers from 21 countries have responded to our invitations to enter anonymised retrospective data on pubertal induction in their patients on the I-TS platform (reference number I-TS/202311_AG).

Results

Clinical and laboratory data of 834 girls with TS and unique ID were available for descriptive analysis. Karyotype distributions were 40.0% (45,X); 12% (45,X/46,XX); 11% (45,X/46,XY); 8% (45,X/46,XiXq); 3% (45,X/47,XXX); 3% (45,X/46,XrX); 23% (other karyotypes). The mean/median age at initial contact with the center was 8.27 years. Data related to pharmacology were available for 307 (37%) patients in 16 countries. Data concerning the initiation of pharmacological intervention for puberty and growth induction are presented in the table. Oral and transdermal natural estradiol were utilized by 72 (34%) and 79 (37.3%) patients, respectively. The synthetic preparation ethinylestradiol was used in 14 (6.6%) patients.

Conclusion

For paediatric rare disease, collection of anonymised retrospective multicentre data enables analysis of large international patient cohorts. Here, we use these data to describe different treatment protocols used in Turner syndrome. Natural oestradiol continues to be the primary choice for puberty induction in most centres, although initiation was delayed compared to current standards. However, reporting of progesterone usage was low. The collection of retrospective data is now complete and the final/updated analysis will be shared at BSPED. The final results will help to inform the prospective study of pubertal induction in TS which commences in Autumn 2025.

Medicine	Number	Median age at start [years]	Interquartile range
Growth hormone	264	9.3	5.1-11.5
Oxandrolone	8	13.0	11.9-14.1
Estrogen(s)-all	276	13.3	12.2-14.7
Progesterone	51	15.2	14.5-16.3

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OC10.5

Assessing the scale of the teenage obesity epidemic in northern ireland: an audit of prevalence and practice in the tertiary paediatric hospital

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The PHA's Children's Health in Northern Ireland 2023/24 report highlighted that Northern Ireland 'NI' has the highest level of obesity in Primary 1 children in the UK. Using the International Obesity Task Force Classification, they cited that 21% of Primary 1 children and 27% of Year 8 children were measured as overweight or obese in 2023/4. Despite these figures there is no paediatric obesity service in NI. This audit assesses the prevalence and management of obesity in children and young people (CYP) aged 12 years and over, who were booked to attend general paediatric outpatient clinics over a 6-month period (June 24-November 24) in the tertiary children's hospital in NI. 509 patients were included in the audit. $n = 273$, 53.6% were female and $n = 236$, 46.4% were male. 359 of these patients had their weight and height recorded. $n = 87$, 24.2% (95%CI 20.3-28.7) had a BMI > 95th centile. $n = 66$, 18.4% (95%CI 14.8-22.3) had a BMI > 98th centile and $n = 40$, 11.1% (95%CI 7.8-14.8) had a BMI > 99.6th centile. $n = 33$, 9.2% (95%CI 6.1-12.5) had a BMI > 30. Of those with a BMI > 98th centile, in 33% of the consultations obesity was addressed (95%CI 23.1-45.4); only 4 of these CYP were referred due to weight. Diet was addressed in 39% (95%CI 28.5-51.5) of the consultations, activity levels in 36% (95%CI 25.8-48.4) and sleep in 17% (95%CI 9.4-27.6). 56% (95%CI 44.1-67.4) had their blood pressure recorded, 30% (95%CI 20.5-42.3) were screened for diabetes, 53% had their LFTs checked (95%CI 41.2-64.6) and 23% had an ultrasound of their liver done (95%CI 14.2-34.3) with 73% of the scans performed showing fatty infiltrates (95%CI 47.6-89.5). 17% had their lipids checked (95%CI 9.4-27.6), 52% had their TFTs checked (95%CI 39.7-63.2). Of those with a blood pressure recorded, in 30% (95%CI 17.4-45.9) it was > 95th centile for sex, age and height. 65.08% (95%CI 52.8-76.0) of these CYP with a BMI > 98th centile were in the 1st quartile deprivation index. This audit highlights the urgent need for a paediatric obesity service in NI, there is not the capacity within busy general paediatrics clinics for obesity and its implications to be addressed in full.

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OC10.6

"Real world" experience" of treatment with Wegovy (semaglutide) and Saxenda (liraglutide) in a paediatric population living with severe obesity

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The Glucagon like one peptides (GLP-1's) **Wegovy** (semaglutide) and **Saxenda** (Liraglutide) are licensed for the treatment of severe obesity in young people aged 12-17 years. We report our "**real-world experience**" of treating a large cohort of 98 young people with GLP-1's under the umbrella of our complications of excess weight clinic (CEW clinic). All young people and their families were offered a minimum of three months dietetic and lifestyle support from a multidisciplinary team comprising nurse specialists, dietician, family support workers and psychologist. Those who struggled to manage their weight with diet and lifestyle changes were offered support with a GLP-1. Families continued to receive lifestyle support and were reviewed 4 weekly in clinic. Ninety-eight young people (48 male & 50 female) received treatment - 21 individuals initially received Saxenda and subsequently Wegovy, whilst 77 received Wegovy alone. The mean age was 14.1 years (range 11-17 years). Mean weight before treatment was 118.8 kg, BMI 41.6 kg/m² and mean BMI standard deviation score (SDS) +3.7. The change in weight, BMI and BMI SDS at 3, 6, 9 & 12 months are shown in the table. Nine patients stopped medication within 6 months as a result of noncompliance or failure to attend appointments. Two stopped because of nausea and one because of a deterioration in mental health. Nausea that subsequently resolved was commonly reported. There were no serious adverse events such as pancreatitis. A reduction in BMI SDS of -0.25 is widely recognised as the threshold to improve physical health. The reduction in weight of 22.0 kg and in BMI SDS of -0.74 noted at 12 months is likely to result in clinically meaningful improvements in complications such as hypertension, MASLD and diabetes risk. These data and data on quality of life are currently being analysed.

Table 1

	Mean Change in Weight (kg)	Mean Change in BMI (kg/m ²)	Mean Change in BMI SDS
3 months ($n = 98$)	-3.3	-1.5	-0.11
6 months ($n = 54$)	-12.8	-4.8	-0.51
9 months ($n = 43$)	-14.9	-6.3	-0.61
12 months ($n = 33$)	-22.0	-7.7	-0.74

Conclusion

Support with GLP-1 s alongside lifestyle intervention results is significant and clinically relevant weight loss.

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OC10.7

Citizens advice sheffield (CAS) and complications from excess weight (CEW) innovative integration pilot, recovering finances of over £29k for 31 families in initial 6 months

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Introduction

Severe obesity is unevenly distributed. By the end of primary school, severe obesity is four times as prevalent in the most deprived areas¹. Children from ethnic minority backgrounds are over-represented in CEW clinics compared to Office for National Statistics Data² distribution. Ethnicity and deprivation do not operate in silo. They compound and deepen vulnerability, worsening health outcomes³. For many families, immediate tasks of daily survival supersede prevention or action towards improving long term health. Significant CEW MDT time is spent assisting with basic needs of shelter and security causing barriers for progression with weight management.

Methods

Sensitive enquiry of socioeconomic barriers is part of holistic CEW assessments to understand family factors and context affecting resilience or compromising ability to follow lifestyle management plans. From December 2024, a CAS advisor joined the Sheffield CEW MDT for a 12-month pilot. This integration enables direct and consented referral to the CAS Advisor who contacts families to arrange confidential expert help with various legal, financial and housing challenges. This is relational care - support from someone who knows your story, offered by someone you already trust. Hands on digital assistance, using interpreters as required, ensures no one is excluded from care. With permission the advisor reports back and can directly request additional gateway information from MDT to promptly unlock access.

Results and Conclusion

In just 6 months, 31 families disclosing a single issue to the CEW MDT were referred to the CAS advisor. Proactive and expert enquiry by CAS then identified a total of 222 issues including benefits, debt, housing, utilities, tax, travel, education, asylum, and untapped charitable support such as food banks. The advisor worked alongside families to navigate overwhelming systemic complexities resulting in a total of £29 514 of recovered access to rightful financial support. When embedded in a CEW service model, CAS can address Maslow's hierarchy of needs to create the conditions for motivation, engagement and improved well-being, increasing the effectiveness of CEW services⁴.

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OC10.8

Behind the glucose dip: a retrospective review of paediatric ketotic hypoglycaemia and evolving management approaches

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Introduction

Ketotic hypoglycemia (KH), a common cause of hypoglycaemia in young children, is typically a diagnosis of exclusion. Given the increase in referrals of recurrent KH to paediatric endocrine and metabolic services, we aimed to examine the clinical outcomes of KH referrals in a large children's hospital in the UK.

Methods

Data were collected retrospectively for all patients with KH known to either the paediatric endocrinology or metabolic departments between 2019 and 2024.

Results

Over a 6-year period, a total of 165 children (53% female) with KH were reviewed by tertiary services. Of these, 126 children (76%) were reviewed by the metabolic team, and 39 (24%) by the endocrine team. Median age at referral was 3 years (range: 9 months - 15 years) whilst the reported age of symptom onset ranged from 3 days to 9 years. Neurodevelopmental disorders (13.3%) was a common comorbidity with KH. However, the presence of comorbidities was not associated with an increased risk of recurrence (odds ratio: 0.98 [0.41-2.36]). In 44 children with recurrent hypoglycaemia, gene panel testing identified two children with metabolic diagnoses (phosphoglucomutase 1 deficiency and dihydropyrimidine dehydrogenase deficiency), while 5 children had variants of unknown significance in the glycogen storage disorder IX and monocarboxylate transporter 1 genes. Hypoglycaemia resolution occurred in 83% of endocrine patients by 8 years of age and 95% of metabolic patients by 7 years of age. Short-term continuous glucose monitoring (CGM) was used in 25/39 (endocrine) and 28/126 (metabolic) patients to demonstrate normoglycaemia, providing reassurance prior to discharge. Endocrine patients remained under follow-up for a mean duration of 16 months (range: 1 - 78 months), compared to 48 months (range: 0 - 177 months) for metabolic patients, reflecting significant workload volumes for both services.

Conclusion

The study highlights the clinical heterogeneity and favourable outcomes of ketotic hypoglycaemia in a large cohort of patients referred to tertiary services. Although infrequent, it is important to investigate inborn errors in metabolism in those with persistent and recurrent ketotic hypoglycaemia.

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Poster Presentations

Adrenal 1

P1

Cortisol responses to glucagon stimulation tests in children with short stature

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Introduction

The clinical utility of cortisol response to the Glucagon Stimulation Test (GST) in children remains unclear, with conflicting evidence. Concerned about potential overuse of the Synacthen Stimulation Test (SST), we evaluated cortisol levels during GST and assessed the impact of clinical factors such as age, sex, pubertal status, and BMI.

Methods

A retrospective chart review was conducted on children who underwent GST for short stature between January 2024 and February 2025 at Evelina Children's Hospital, London. Demographic and anthropometric data were collected. Cortisol and GH responses to GST, and SST results where available, were analysed using non-parametric tests.

Results

105 children underwent GST during the study period. 13 (12.3%) had suboptimal cortisol responses; 12 (92.3%) passed subsequent SST, and 1 on steroids had SST deferred. Cortisol peaked at 120' in 8 (7.5%), 150' in 26 (24.7%), and 180' in 67 (63.8%); mean peak was 592 nmol/l. Among those with suboptimal cortisol, mean age was 14.5 years, 12 (92.3%) were male, 9 (69.2%) pubertal, and 5 (38.4%) overweight/obese. Five also had suboptimal GH responses, and one failed a second growth hormone stimulation test with arginine. Younger children had significantly higher cortisol levels; higher BMI ($p < 0.001$) was associated with lower cortisol. SST peaks were consistently higher than GST peaks. (Figure-1)

Conclusion

Our findings support that although both GST and SST are viable tools for assessing adrenal reserve in children, SST consistently produces higher peak cortisol responses than GST. Additionally, GST results are influenced by age and BMI, with older and overweight children demonstrating lower cortisol, potentially leading to false positives. In contrast, SST gives a more robust, reliable assessment with well established cut offs. Therefore, while GST can serve as an alternative, SST remains the preferred method for accurate pediatric adrenal evaluation.

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P2

Neonatal hypothermia and adrenal insufficiency: a case series of two presentations

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Introduction

Adrenal insufficiency (AI) in neonates is rare but potentially life-threatening. Early diagnosis is challenging due to nonspecific presentations. We describe two neonates who presented within one month with hypothermia and were diagnosed with AI, highlighting variable presentations, extensive work-up, and the need for early endocrine consideration.

Case 1

A male infant, born at 34+2 weeks via emergency Caesarean for suspicious CTG, had an uneventful early course. On day 7, he presented with hypothermia (32.5°C), lethargy, and apnoeas. Sepsis was suspected; CRP, blood and CSF cultures, glucose, and electrolytes were normal. At day 14, he re-presented with hypothermia (33.7°C); cortisol was low (74 nmol/l), and a short synacthen test (SST) showed inadequate response, confirming AI. MRI brain showed an absent posterior pituitary bright spot but no structural abnormality; MRI adrenal glands were normal. He improved with steroids. At 6 weeks, he presented with vomiting and hypertension, initially attributed to steroids, but was diagnosed with pyloric stenosis requiring surgery. On follow-up for persistent hypertension, repeat SST was normal; steroids were discontinued, suggesting transient secondary AI, likely due to hypothalamic-pituitary axis immaturity.

Case 2

A female infant born at 35+5 weeks had antenatal concerns of macrocephaly and short long-bones. She was treated postnatally for jaundice and suspected omphalitis

(cultures negative), then discharged on oral antibiotics. At 2 weeks, she presented with hypothermia (29.9°C), bilious vomiting, constipation, apnoea, and bradycardia requiring resuscitation. Blood glucose was 2.6 mmol/l; CRP was <1, cultures (negative). CT suggested white matter changes; MRI showed poor gyri development and bilateral conatal periventricular cysts. Adrenal ultrasound revealed hypoplastic adrenal glands. She failed SST. An extensive endocrine, metabolic, and genetic work-up (including Zellweger screen) was negative. She improved with steroid therapy and remains under endocrine follow-up.

Conclusion

These cases show various presentations of neonatal AI, from recurrent hypothermia to acute collapse, initially mimicking sepsis. Case 1 highlights transient secondary AI, with resolution by 3 months; Case 2 demonstrates primary AI with adrenal and CNS involvement. Persistent hypothermia without infection should prompt endocrine investigation. Early input and follow-up are key to diagnosis and management.

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P3

Evaluating the indications for measuring cortisol in neonates, assessing frequency of short synacthen tests (SSTs) and identifying SST challenges and variations in newcastle, UK

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Background

SSTs are performed in neonates for various indications and are important in identifying babies at risk of adrenal insufficiency who may require exogenous corticosteroids.

Objectives

To identify common indications for measuring cortisol in neonates, to determine which babies proceeded to have a SST, and to identify challenges and variations in how SSTs were performed.

Methods

We retrospectively evaluated all 55 babies in our NICU who had a cortisol level measured between January 2021-December 2022. We identified which babies proceeded to have a SST and reviewed patient notes to identify the indication for SST and any challenges and variations in performing the test.

Results

The average age when cortisol was first measured was 14.5 days with an average corrected gestation of 34+4 (range 22+6 – < 42+5). Common indications for measuring cortisol were persistent hypoglycaemia (24/55), conjugated hyperbilirubinemia (8/55), cerebral/spinal abnormalities (8/55) and electrolyte abnormalities (5/55), with some having >1 indication. 20/55 proceeded to undergo a SST, with 19/20 cases documented as discussed with paediatric endocrine. There was significant variation in how SSTs were performed. Cortisol was measured either once or twice post synacthen and at varying time points anywhere between 20-115 minutes. It was not always clear whether this variation was secondary to endocrine advice or other factors. In 5/20 cases at least one sample was not processed due to insufficient volume. Cortisol response was higher at 60 minutes compared to 30 minutes but declined after 105 minutes.

Conclusion

SSTs are conducted in our unit for clear indications and generally discussed with paediatric endocrinology. However, there is significant variation as to when cortisol is measured post synacthen and a risk of insufficient sampling. Measuring cortisol at 60 minutes vs. 30 minutes is more likely to exclude adrenal insufficiency, and this has been observed in multiple previous studies. While a single measurement at 60 minutes would reduce pain and blood loss in neonates, it is important to consider the impact delays or insufficient sampling may have on results when relying on a single measurement and we therefore recommend measuring cortisol at 30 and 60 minutes post synacthen.

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P4

Non-invasive biochemical monitoring for congenital adrenal hyperplasia (CAH) using 24-hour urinary steroid profiles

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Background

Monitoring disease control in congenital adrenal hyperplasia (CAH) using random serum 17-hydroxyprogesterone (17OHP) measurements is invasive and may not reflect overall daily adrenal steroid production. Urinary steroid profiling offers a non-invasive alternative that may better reflect cumulative steroid output. This study aimed to evaluate whether urinary steroid metabolites, quantified using a novel approach of liquid chromatography–high resolution mass-spectrometry (LC-HRMS), could predict serum 17OHP and identify the most informative urinary biomarkers of biochemical control.

Methods

Twenty-four-hour urine samples were collected from 92 children with CAH, due to 21-hydroxylase deficiency, across 13 UK centres and analysed at the University of Edinburgh using LC-HRMS. A panel of 15 urinary metabolites was measured, including tetrahydro-glucocorticoids, cortol/cortolone derivatives, androgen and 11-oxygenated androgen metabolites, and pregnane metabolites. Paired serum and salivary samples were collected post-morning glucocorticoid dose and analysed for 17OHP, cortisol, and cortisone using LC-MS. Missing data were addressed using multiple imputation using Amelia II package in R, applying expectation-maximisation with bootstrapping. Multivariate linear regression was used to evaluate model performance (R^2).

Results

The cohort (49 female, 43 male) ranged from 8 to 18 years, with 95% receiving oral hydrocortisone 2–4 times daily. Mean daily hydrocortisone equivalent was 18.7 mg (SD 6.7), or 13.5 mg/m² (SD 3.8). A multivariable model including 15 urinary steroid metabolites and glucocorticoid dose explained 54.9% of the variance in serum 17OHP ($R^2 = 0.55$, $P < 0.001$). Salivary cortisol/cortisone, sex, age, and body surface area did not significantly improve model performance ($p > 0.05$). Two urinary metabolites were independently associated with serum 17OHP: androsterone ($P = 0.033$) and pregnanetriol ($P = 0.005$). Pearson's correlation confirmed these associations: pregnanetriol showed a strong positive correlation with serum 17OHP ($R^2 = 0.63$, $P < 0.001$), and androsterone showed a moderate positive correlation ($R^2 = 0.46$, $P < 0.001$). The two metabolites were moderately correlated with each other ($R^2 = 0.65$), suggesting overlapping but distinct contributions.

Conclusions

Urinary steroid profiling, combined with glucocorticoid dose, predicts serum 17OHP with good accuracy. Androsterone and pregnanetriol emerged as key urinary biomarkers reflecting adrenal androgen and progesterone metabolism, supporting their potential utility in non-invasive disease monitoring.

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P5

Parent perspectives to develop a symptom-tracker app to optimise home management of childhood adrenal insufficiency

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Background

Intercurrent illnesses may precipitate life-threatening adrenal crises in children with adrenal insufficiency (AI). Incidence of these may be reduced by effective home management in response to early symptoms of illness; however, such symptoms are not well characterized and often individual. We showed recently that symptom-tracking is popular in families living with childhood AI. In this study we present parent/carer perspectives from the Manchester Adrenal Clinic to guide development of a web-based AI symptom-tracker app.

Methods

We convened a 90-minute online focus group workshop with 12 parents/carers of children aged 3–13 years with AI due to Congenital Adrenal Hyperplasia (CAH). After semi-structured walkthroughs of a low-fidelity prototype, an open question and answer (Q&A) session captured facilitators' notes and audio transcripts for thematic analysis.

Results

Thematic analysis revealed five key areas of participant enthusiasm: 1. Structured, yet flexible, symptom taxonomy: Pre-defined visual menus with icon-based 1–5 severity ratings, plus an optional free-text box for unique observations. 2. Integrated dose logging: Real-time entry of hydrocortisone doses, missed doses, and emergency injections with automated alerts for late or missed doses. 3. Multi-user role access: Graded permissions allowing school staff and secondary caregivers to submit symptoms. 4. Mood and emotional-health tracking: A simple well-being slider alongside free-text notes to capture AI-related stress and mood changes. 5. Automated trend reports & data sharing: One-click generation of clinician-friendly summaries (email or print), helping families recall symptom and dosing patterns between infrequent clinic visits and streamline communication with care teams.

Conclusion

Parent/carer feedback strongly supported the need for an AI symptom-tracker app combining structured symptom logging, real-time dose management, role-based access, mood monitoring, and automated report sharing. These features may enhance home management, facilitate timely interventions, and improve communication with clinical teams to improve the care of childhood AI. Next steps involve iterative prototyping and a pilot usability study to evaluate clinical impact.

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P6

Analysis of real-world data on the care provision of children with congenital adrenal hyperplasia in the United Kingdom and republic of Ireland

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Background

To explore the current practice of CAH management in children, we launched a 5-year-project in 03/2022, collecting annually longitudinal data from the I-CAH Registry on the SDMregistries platform on patients under 18 with 21-hydroxylase deficiency (21OHD) from the UK and Republic of Ireland.

Methods

By April 2025, 1291 visits from 182 patients under 18 with 21OHD (100 females), from 15 centres had been recorded in the I-CAH registry since 01/01/2016. Using regression strategies in the *RStudio*, we analysed information related to medication, biomarkers and height standard deviation scores (SDS) for age and sex (WHO reference). We expressed glucocorticoid (GC) doses as hydrocortisone (HC) equivalent per body surface area.

Results

Most patients (95%) were treated with hydrocortisone given three or four times daily, the remaining 8 patients using prednisolone. The mean daily GC dose was 12.5 ($\pm$ 3.3 SD) HC-equivalent mg/m²/day, mean dose per patient ranging from 4.7 ($\pm$ 0.06) to 22.3 ($\pm$ 0.4) mg/m²/day. There were broad variations in mean GC doses between centres, fluctuating between 9.8 and 16.8 mg/m²/day. The GC dose increased with age by 0.2 mg/m²/day per year, depending upon centre, and varied with sex, doses used in boys being lower by 1.1 mg/m²/day. There was large variation in the timing of the GC doses, with six centres using late doses between 22:00-24:00. Total daily doses of fludrocortisone ranged between 25 and 350 mg/day, with significant variability between centres ($R^2=0.02$, $P < 0.01$). 17-hydroxyprogesterone was reported high in 42% patient visits and low in 14%; androstenedione was high in 25% patient visits and low in 20%. Height SDS fluctuated with age, starting from -0.5 in infancy increasing to 1.2 at 10 years and then decreasing to -1.0 at 17.5 years.

Conclusions

Through active engagement with the collaborating centres, we have significantly increased the number of patients consented and recorded in the I-CAH registry. There is evidence of significant variation between centres in the treatment strategies used for CAH. Further analysis linking different approaches to health outcomes will garner further insights towards improving treatment and help develop strategies for benchmarking care for CAH within the UK and the Republic of Ireland.

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P7

Establishing the current practice of the use of genetic testing for patients with congenital adrenal hyperplasia (CAH)

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Background

There are no clear national or international guidelines for genetic testing for diagnosing 21 Hydroxylase Deficiency (21OHD) CAH (CAH). This study aimed to assess the current international practice of genetic testing in CAH.

Methods

We collected and analysed data on patients diagnosed with 21OHD CAH, using the SDMregistries core module data, including date of birth, first presentation and diagnosis, and the investigations on which the diagnosis was based. We also distributed questionnaires to the registry centre leads, exploring site- and clinician- specific approach towards the diagnosis of 21OHD.

Results

There were 437 patients (279 females) from 18 countries (53 centres) included in the study. The proportion of patients diagnosed with 21OHD that had genetic tests varied among countries and centres between 0 and 100%, with a median of 82(IQR 43-99)%. Genetic testing was more frequent in females compared to males (76% vs 56%, Chi square $P < 0.01$). The time elapsed between presentation and diagnosis was not influenced by sex or the use of genetic testing, with a median of 4.3 (IQR 1.2-53.6) weeks. The survey was answered by 64 clinicians from 26 countries. While 22% indicated that they used genetic testing in all patients suspected of 21OHD, the majority (62%) use it only if hormonal tests are abnormal or inconclusive. Direct sequencing was used by 64% centres, multiplex ligation-dependent probe by 52%, and targeted mutation analysis by 16%. The turnaround time for genetic results varied between 1 and 52 weeks. In 62.5% centres, genetic testing was funded via the public health system, other sources being health insurance, research funding or self-funding. The majority (98%) of clinicians indicated that the main benefit of genetic testing is supporting family counselling, although 28% considered it was also helpful in guiding hormone replacement.

Conclusion

There is wide variability in the use of genetic testing for the diagnosis of 21OHD across countries and centres. Genetic testing is primarily used to clarify the diagnosis when hormonal tests are inconclusive and to support family counselling. Our data does not indicate that genetic testing influences significantly the time of the diagnosis or of treatment initiation.

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P8

Care provision for patients with congenital adrenal hyperplasia: a scoring review to address gaps in research and patient care

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Background

Congenital adrenal hyperplasia (CAH) is a rare inherited disorder, usually due to 21-hydroxylase deficiency, causing reduced cortisol and aldosterone production with excess adrenal androgens. Although corticosteroid replacement and new treatments have improved outcomes, people with CAH still experience serious long-term complications. Managing CAH remains complex, with challenges in optimising treatment, preventing adrenal crises, and enhancing quality of life.

Objective

This study aimed to identify ongoing gaps in research and practice and set priorities for better lifelong care. We systematically reviewed literature published since 2001 to map the scope and depth of existing research on CAH.

Methods

Following Arksey and O'Malley's scoping review framework, we systematically searched multiple databases, including MEDLINE, EMBASE, and CINAHL, for studies published from 2001 onwards that address CAH-related complications, treatment approaches, and patient outcomes. The search strategy included the MeSH term "Congenital Adrenal Hyperplasia" and its variations, covering both children and adults. We excluded case reports, review articles, and molecular studies. The Rayyan platform was used to record, screen, and organise the articles. Two independent reviewers screened the articles, with any disagreements resolved by a third reviewer.

Results

The initial search identified 3,016 articles, of which 426 met the inclusion criteria. Full texts were available for 312 articles. To provide more categorised data, we initially grouped the articles into eleven preliminary themes, which were refined to seven final themes, ultimately yielding 259 articles. To link our findings more effectively to patient outcomes, the themes were organised around major patient complications, overall quality of life, and complexities in care. The final thematic distribution included 56 articles on cardiovascular health, 57 on cognitive and mental health, 78 on metabolic outcomes, 21 on adrenal crisis and hospital attendance, 73 on growth, 35 on quality of life, and 143 on care provision. Overall, yielding 118 retrospective studies.

Conclusion

This review provides a comprehensive map of current evidence, clarifies where knowledge gaps remain, and offers practical directions for future research and clinical practice. Improving CAH care requires ongoing multidisciplinary efforts to develop more personalised, safe, and effective management strategies that address the full spectrum of patient needs across the lifespan.

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P9

17 α -Hydroxylase deficiency: from a rare presentation in infancy to transition to adult care

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Introduction

17 α -hydroxylase deficiency (17OHD) is a rare autosomal recessive disorder caused by biallelic mutations in the *CYP17A1* gene, accounting for approximately 1% of cases of congenital adrenal hyperplasia. It commonly presents with primary amenorrhoea, cryptorchidism, and atypical genitalia, while adrenal crisis is a rare occurrence. Here, we describe a case of 17OHD presenting at 10 months with hypoglycaemia.

Case presentation

A 10-month-old girl of consanguineous parents presented to the Emergency Department with diarrhoea, vomiting, and dehydration, caused by rotavirus infection. She had normal female external genitalia with a gonad palpable in the right inguinal canal. Investigations revealed hypoglycaemia (1.7 mmol/l) with

hypokalaemia (3.1 mmol/l), with low serum cortisol (106 nmol/l). A standard Synacthen test showed a peak cortisol of 53 nmol/l and raised ACTH (175 ng/l). The workup for adrenal insufficiency revealed low androgen and cortisol metabolites, along with elevated corticosterone metabolites in the urinary steroid profile (USP), confirming 17OHD. The patient was started on hydrocortisone, with good clinical progress. There were no further episodes of hypoglycaemia or adrenal crises, and renin normalised. The karyotype was 46 XY. The patient was reared as female and underwent cystovaginostomy and bilateral laparoscopic gonadectomy at 2.5 years. She was started on 17 β oestradiol patches at 11 years to support secondary sexual characteristics. The patient had a gynaecological assessment to determine the need for vaginal dilatation. She is currently a teenager with a final height consistent with her parental target, ready for transition to adult services. Her sisters were screened by checking their karyotype and USP to exclude a similar diagnosis. Genetic testing is planned to determine their carrier status.

Conclusion

Patients with 17OHD are often diagnosed during adolescence following investigations for hypertension or delayed puberty. Our case had an atypical presentation with infantile hypoglycaemia. Management consisted of hydrocortisone replacement with monitoring of blood pressure, renin, and aldosterone, and hormone replacement therapy at the expected age of puberty. This case highlights the implications of this diagnosis for family members and the importance of involvement of the multidisciplinary team to support gender identity decisions, gonadectomy timing, hormone replacement, and psychosocial adjustment.

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P10

Cortisol response to hypoglycaemia in infants with congenital hyperinsulinism: a single centre retrospective cohort 2015-2025

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Background

In congenital hyperinsulinism (CHI), inappropriate, excessive insulin secretion leads to recurrent severe hypoglycaemia with risk of seizures and neurodevelopmental injury. Cortisol is a key counter regulatory hormone, yet published data suggests blunting of the cortisol response in hyperinsulinemic hypoglycaemia.

Aim & Methods

We retrospectively reviewed infants <6 months with CHI managed at Alder Hey Children's Hospital (2015–2025). Inclusion required paired glucose (<3 mmol/l), and cortisol sampled at the time of hypoglycaemia; demographic and biochemical data plus short Synacthen test (SST) results were extracted. A normal response was a peak cortisol of >500 nmol/l.

Results

Eighty three infants were screened; fifty met inclusion criteria (complete glucose and cortisol dataset). (30 male; mean birthweight 2.82 kg; mean gestation 36 w; 14/50 maternal diabetes; 50 diagnosed with CHI - 7 persistent, 43 transient). Mean cortisol during hypoglycaemia was 369 nmol/l. Distribution: <100 nmol/l ($n = 12$), 100–150 nmol/l ($n = 7$), >150 nmol/l ($n = 31$; mean 497 nmol/l). SST data (subset): 14/18 (78%) showed normal adrenal reserve on initial Synacthen testing; failures in 4 infants (<100:3/8; >150:1/7; 100–150:0/3). These patients received hydrocortisone therapy, which was discontinued in every case, as no infant had evidence of persistent adrenal insufficiency by 21 months. Among infants with cortisol <150 nmol/l ($n = 19$), 6 had birthweight <2.5 kg, 6 were premature, and 7 had maternal diabetes, suggesting vulnerability of an immature HPA axis and/or low cortisol binding proteins in early life.

Conclusions

Infants with CHI commonly exhibit a suboptimal cortisol response during spontaneous hypoglycaemia, yet most have intact adrenal capacity on stimulation testing with ACTH and recover if initially insufficient. SST should be reassessed for those commenced on hydrocortisone, as it frequently normalises on repeat testing. Apparent adrenal dysfunction may be developmentally mediated and was transient in this cohort. Our findings highlight the need for further research into the maturation of the hypothalamic-pituitary-adrenal axis in neonates, including how it may be influenced in the context of hyperinsulinism.

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Bone

P11

Bone mineral density and vertebral fractures in teenage and young adult patients with acute lymphoblastic leukaemia: a report from the british osteonecrosis study

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Introduction

The British Osteonecrosis Study (BONES) is the first multicentre prospective study assessing bone health and vertebral fractures in patients aged 10-24 in the UK undergoing treatment for acute lymphoblastic leukaemia (ALL) or lymphoblastic lymphoma (LBL).

Methods

61 patients were recruited from 3 tertiary centres in the UK. Dual-energy x-ray absorptiometry (DXA) scans with vertebral fracture analysis were performed within 4 weeks of diagnosis and annually for 3 years. Subjective pain assessments were performed at the same timepoints.

Results

Bone mineral density (assessing total body less head (TBLH) significantly reduced after 2-years, compared to baseline (estimate = -0.964, 95% CI [-1.357, -0.572]), with greatest decrease occurring within the first year. Vertebral fracture prevalence was 4.9%, with 2 further patients experiencing incident vertebral fractures. All vertebral fractures occurred in male patients, 75% of whom were British Asian. Back pain was not a predictor of low BMD or vertebral fractures.

Discussion

We report a lower vertebral fracture prevalence in patients aged 10-24 with ALL than has been previously reported in a cohort of younger patients. Male British Asian patients appeared to be at higher risk of vertebral fractures in this study. BMD and pain were not predictors of vertebral fractures.

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P12

¹⁸Fluorocholine PET/CT scan in the investigation of a patient with persistent hypercalcaemia

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Background

Paediatric primary hyperparathyroidism (PHP) caused by adenoma of the parathyroid gland is rare. The investigation to localise parathyroid adenomas using imaging modalities currently available in children can be challenging, due to the small dimensions of functional parathyroid adenomas.

Case

An 8-year-old male had an incidental finding of hypercalcaemia. Adjusted calcium was raised 3.59 mmol/l (2.15 – 2.74 mmol/l) with normal phosphate 0.89 mmol/l (0.81 – 1.61 0.89 mmol/l), PTH was elevated 13.1 pmol/l (1.1 – 6.9 pmol/l). Urine calcium: creatinine ratio was 1.27 mm/mm Cr (0 – 0.6 mm/mm Cr). The patient was managed with intravenous fluids, furosemide and pamidronate. After an initial brief response, he re-presented due to persisting hypercalcaemia. Cinacalcet was introduced and doses were gradually titrated to maintain calcium within normal range. Conventional imaging modalities, neck ultrasound followed by planar scintigraphy and delayed SPECT/CT, were insufficient to localise a parathyroid adenoma. Additional neck MRI was performed for increased uptake in

the left submandibular region on SPECT/CT, but was also inconclusive. Following this, a ¹⁸Fluorocholine PET/CT scan was performed, which showed a functional lesion in the left lower parathyroid gland. This facilitated a targeted surgical excision of the parathyroid adenoma, with the support of intra-operative PTH monitoring. Operative findings accurately mirrored those of the imaging. Post-operative PTH normalised within one day and subsequently all medications were discontinued.

Discussion

This case highlights the role of ¹⁸Fluorocholine PET/CT in the investigation of patients with suspected parathyroid adenoma. Recent studies have highlighted the sensitivity and specificity of ¹⁸Fluorocholine PET/CT imaging in identifying parathyroid adenomas, compared to other imaging modalities. Other advantages include low radiation and short scan time. The challenges of ¹⁸Fluorocholine PET/CT include limited availability of the tracer and that often the test is required to be performed in an adult care setting, which requires adaptation of dosing and patient care protocols. With more frequent use and therefore experience, and given such sensitivity and specificity, ¹⁸Fluorocholine PET/CT may become a preferred option for the investigation of patients with PHP in future.

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P13

Beyond the numbers: rare cause of persistent vitamin d deficiency in a child

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Introduction

The group-specific component (GC) gene, located on chromosome 4q12-q13, encodes the vitamin D binding protein (DBP). It is a highly polymorphic protein and 80-90% of vitamin D (VD) metabolites bind to a single site of DBP. DBP deficiency is a recessive condition, which causes low VD levels and features of rickets.

Case Report

A 17.5-year-old female, originally from Pakistan, was referred for persistent VD deficiency despite treatment. She previously had rickets. At 11.3 years, she presented with abnormal biochemistry (Table1). Already on calcium supplements, she commenced VD 20,000units twice weekly for 10 weeks, then 800units daily. Her bone profile improved, but VD remained undetectable. She was re-referred at 16.8 years with bone pain, headaches and undetectable VD levels. She was treated with 25,000units daily for 8 days, then 800 units daily. With no improvement (Table2), intramuscular VD (300,000units) was given at age 17, with continued oral maintenance dosing. Again, the VD level remained undetectable, but bone biochemistry normalised. The 1,25 dihydroxy-VD was also undetectable. This raised the rare possibility of DBP deficiency and levels were found to be undetectable. Genetic analysis is pending. Further VD level monitoring would be uninterpretable, but optimising calcium and VD intake improved her symptoms.

Conclusion

This case highlights the importance of considering DBP deficiency in low VD levels despite treatment and improved bone biochemistry. Delay in diagnosis led to unnecessary high doses of VD supplementation and blood tests. The patient responded to supplementation both clinically and biochemically. In such patients, regular VD supplementation is necessary, and the response should be monitored clinically with bone biochemistry, rather than VD levels.

Table 1 Biochemistry

Age (years) Test (NR)	11.1	11.3	11.4	11.8
Corrected Calcium (mmol/l; 2.15-2.74)	1.60	2.06	2.18	2.24
Phosphate (mmol/l; 0.97-1.94)	0.82	1.24	1.13	1.28
Alkaline Phosphatase (iu/l; 203-1151)	2039	1472	868	230
Total VD (nmol/l; <50)	<5	<5	9	<10
Parathyroid Hormone (pmol/l; 1.1-6.9)	X	X	35.1	25.9

Table 2 Biochemistry

Age (years) Test (NR)	16.8	17
Corrected Calcium (mmol/l; 2.25-2.74)	2.28	2.32
Phosphate (mmol/l; 0.74-1.94)	0.78	1.05
Alkaline Phosphatase (iu/l; 38-123)	85	97
Total VD (nmol/l; >50)	<9	<9
Parathyroid Hormone (pmol/l; 1.1-6.9)	9.4	7.1
1,25 Dihydroxy-VD (pmol/l; 108-246)	<12	<12

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P14**Impact of body mass index on vertebral fracture risk and bone mineral density parameters in children with osteogenesis imperfecta**Sameera Hannah Auckburally¹, Mohamed Zulficar Mughal², Raja Padidela^{1,3} & Amish Chinoy¹¹Royal Manchester Children's Hospital, Manchester, United Kingdom; ²Al Jalila Children's Hospital, Dubai, UAE; ³University of Manchester, Manchester, United Kingdom**Introduction**

Osteogenesis imperfecta (OI) is a rare genetic disorder characterised by reduced bone mineral density (BMD) and increased susceptibility to fractures. In the general population, higher BMI is positively associated with BMD and may lower fracture risk, but this relationship remains underexplored in children with OI. This study investigated associations between standardised BMI (BMI SDS), the number of vertebral fractures, and BMD in a paediatric OI cohort.

Methods

Data were analysed from children with OI managed at a tertiary UK centre. Data included BMI, vertebral fracture count from dual-energy X-ray absorptiometry (DXA) or spinal radiographs, and DXA-derived BMD measures prior to bisphosphonate therapy. BMI SDS was calculated using UK population reference data. Overweight and obesity were classified as BMI SDS of ≥ 1.33 and ≥ 2 respectively. Statistical analyses assessed associations between BMI SDS, vertebral fractures, and bone density measures.

Results

Eighty-six patients (56.5% female, mean age 10.6 years) with confirmed OI were included in the data analysis. Mean BMI SDS was higher than the general population (0.427; 95% CI 0.230–0.624, $P = 0.001$), with 25.6% in the overweight/obesity categories. Fifty-seven percent had at least one vertebral fracture. There was no association between mean BMI SDS and number of vertebral fractures ($\rho = 0.096$, $P = 0.378$). There was no association between having a BMI in the overweight/obesity range and the presence of at least one vertebral fracture ($\chi^2 = 0.962$, $P = 0.326$). BMI SDS showed no significant associations with most site-specific bone parameters, including lumbar spine bone mineral apparent density (BMAD) and pQCT measures. However, BMI SDS correlated positively with lean mass adjusted for height ($\rho = 0.483$, $P < 0.001$), whole-body BMD and bone mineral content when adjusted for age and height respectively ($\rho = 0.606$, $P < 0.001$; $\rho = 0.311$, $P < 0.05$).

Conclusion

BMI SDS was higher in this paediatric OI cohort compared to the general population. BMI SDS does not appear to increase vertebral fracture risk in children with OI, but is positively associated with lean mass adjusted for height and whole-body less head BMD, possibly due to an impact on muscle mass and therefore bone health. The findings highlight the complex relationship between body composition, bone health and fracture risk in children with OI.

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P15**Long-term outcomes of intravenous zoledronate on DXA bone mineral content in duchenne muscular dystrophy**A McLean^{1,2}, J Dunne³, I Horrocks³, S Joseph³, S Shepherd⁴ & SC Wong^{2,4}¹School of Medicine, University of Glasgow, Glasgow, United Kingdom;²Bone, Endocrine, Nutrition Research Group in Glasgow, Human Nutrition, University of Glasgow, Glasgow, United Kingdom;³Department of Paediatric Neurology, Royal Hospital for Children, Glasgow, United Kingdom;⁴Department of Paediatric Endocrinology, Royal Hospital for Children, Glasgow, United Kingdom**Introduction**

Intravenous bisphosphonates are the standard of care for treating skeletal complications in Duchenne muscular dystrophy (DMD), typically initiated following vertebral fracture. Short-term benefits in bone mineral density/content have been demonstrated in research studies including clinical trials. Current international guidelines recommend continued use until completion of linear growth; however, long-term efficacy data in DMD remain limited.

Aim

To evaluate changes in dual-energy X-ray absorptiometry (DXA) outcomes following long-term intravenous bisphosphonate therapy in boys with DMD.

Methods

This retrospective study included boys with DMD treated with intravenous zoledronate between 2014 and 2020 who remained on therapy for over two years. DXA data were collected at four time points: (a) first available scan, (b) pre-

treatment baseline, (c) 20–24 months post-initiation, and (d) maximum follow-up. Continuous data are presented as mean (SD); p-values < 0.05 were considered statistically significant.

Results

Fourteen boys (mean age at initiation: 10.8 years, SD 2.6) received zoledronate after vertebral fracture diagnosis. All were on corticosteroids (13/14 daily); 8/14 (57%) were ambulant at initiation, and 1/14 (7%) was on testosterone. At maximum follow-up (mean age 15.5 years, SD 2.9), 4/14 (29%) remained ambulant, and 4/14 were on testosterone. Despite expected age-related increases, total body less head (TBLH) bone area and BMC did not significantly change ($P = 0.62$ and $P = 0.75$, respectively). TBLH BMC-for-bone-area SDS remained stable across all time points ($P = 0.05$). In contrast, lumbar spine (LS) bone area and BMC significantly increased over time, with LS BMC-for-bone-area SDS improving from -0.05 (SD 1.1) at first scan to 0.85 (SD 2.0) at maximum follow-up ($P = 0.02$ vs first scan; $P = 0.03$ vs baseline). No atypical femoral fractures or cases of osteonecrosis of the jaw were reported.

Conclusion

Long-term zoledronate therapy was associated with improved spine bone mineral content, but not total body bone mineral content, highlighting the underlying impact of progressive myopathy. These findings underscore the need to investigate osteoanabolic therapies in DMD to better support skeletal health.

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P16**Hypercalcaemia as a sentinel sign of immunodeficiency: lessons from a case of infantile pneumocystis jirovecii pneumonia**

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PTH-independent hypercalcaemia in infancy has a broad differential and poses a diagnostic challenge. Causes include vitamin D intoxication, idiopathic infantile hypercalcaemia (often due to CYP24A1 or SLC34A1 mutations), Williams-Beuren syndrome, subcutaneous fat necrosis, malignancy, and granulomatous diseases such as sarcoidosis or infections like tuberculosis. While *Pneumocystis jirovecii* pneumonia (PJP) is not classically associated with hypercalcaemia, case reports have described mild hypercalcaemia in immunosuppressed solid organ transplant recipients, particularly kidney and liver, leading to a diagnosis of PJP. We report an infant presenting with PTH-independent hypercalcaemia as the initial endocrine issue that led to a diagnosis of *Pneumocystis jirovecii* pneumonia secondary to an underlying primary immunodeficiency. A 5-month-old male presented with a two-week history of respiratory distress on a 2-month history of faltering growth. Investigations revealed persistent hypercalcaemia (adjusted calcium up to 3.33 mmol/l), suppressed PTH (0.3 pmol/l), and an inappropriately normal 1,25-dihydroxyvitamin D (209 pmol/l, reference range 77–471 pmol/l), suggesting a PTH-independent process. Vitamin D toxicity, genetic syndromes, and malignancy were excluded. Given an unsuppressed 1,25-vitamin D, granulomatous disease was considered, and PCR of bronchoalveolar lavage confirmed PJP. Immunological studies showed low IgG and IgA, elevated IgM, vaccine failure, and reduced CD40 ligand expression – consistent with X-linked hyper-IgM syndrome. Initial management of hypercalcaemia included intravenous fluid therapy and dietary calcium restriction. These measures were ineffective until targeted treatment for PJP, comprising intravenous cotrimoxazole and corticosteroids, was initiated. This led to clinical improvement and normalisation of serum calcium levels, permitting the reintroduction of standard feeding. The patient was commenced on regular immunoglobulin replacement therapy and is currently being assessed for haematopoietic stem cell transplantation.

Discussion

Our case is notable for unexplained hypercalcaemia as an initial feature of PJP. The proposed mechanism parallels other granulomatous diseases, where activated pulmonary macrophages and monocytes in inflammatory granulomas express extrarenal 1 α -hydroxylase, driving increased 1,25-dihydroxyvitamin D production and PTH-independent hypercalcaemia.

Take-home message

Endocrinologists should consider opportunistic infections like *Pneumocystis jirovecii* in the differential diagnosis of PTH-independent hypercalcaemia, particularly in the context of known or suspected primary or secondary immunodeficiency.

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P17

Bone health in children with spinal muscular atrophy – a pilot study from a uk centre

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Background

Recently introduced disease modifying treatments have dramatically changed the clinical course of spinal muscular atrophy (SMA) and improved survival. Children are now encountering more long-term complications. Bone fragility is a poorly understood complication and whether disease modifying drugs affect bone health (and fragility) is yet to be determined. Our aim was to describe bone health in children with SMA under the care of a tertiary UK children's hospital.

Method

A retrospective case notes review was conducted of all paediatric patients with SMA at our tertiary centre in 2025. Clinical data on fracture frequency, bone mineral density (BMD), bone biochemistry and vitamin D supplementation were collected.

Results

Twenty-nine children with SMA (Type 1 = 16, Type 2 = 8, Type 3 = 4 and pre-symptomatic = 1) were included. Median age was 7 years (range 2-15). All children were receiving disease modifying drugs (11 onasemnogene abeparvovec (OA), 3 nusinersen and 16 risdiplam). Fifteen children had switched from one drug to another (7 nusinersen to OA and 9 nusinersen to risdiplam). Of children aged 5-15 years, 12/24 underwent DXA scans. All had low BMD on at least one scan. Mean total body less head (TBLH) Z-score was -3.09 (SD +/- 0.49) and lumbar spine Z-score was -2.42 (SD +/- 1.14). Mean BMD Z-scores were lower in children with Type 1 than those with Type 2 and Type 3. Four children (Type 1 = 3 and Type 2 = 1) had a combined total of five fractures, four femoral and one vertebral, all caused by low trauma mechanisms. 97% of children received vitamin D supplementation and mean vitamin D levels was 64nmol/l (SD +/- 20). Two children who sustained fractures received bisphosphonate treatment for bone fragility.

Conclusion

Our pilot data showed a high prevalence of low BMD in children with SMA who underwent DXA scans. Moreover, although uncommon, fractures sustained were caused by minimal trauma. Vertebral fractures were however rare. A multi-centre retrospective study (SMA-ABC) is now underway to further contribute to the understanding of bone health in this population and inform future management plans.

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P18

Severe vitamin D deficiency in early infancy mimicking metabolic and genetic disorders: a case report

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Background

Vitamin D deficiency is recognised as a global public health concern, with a prevalence of 20–25% in the general population. In infants, intrauterine deficiency can significantly impact growth, immune function, and neurodevelopmental outcomes.

Case presentation

6-week-old infant presented to hospital with poor weight gain and recurrent vomiting. Born at 39+6 weeks, with a birth weight of 2.54 kg. Mother had anaemia during pregnancy, requiring blood transfusion. Initial investigations revealed hypomagnesemia, which was corrected. Abdominal ultrasound and brain MRI were normal, in echocardiography patent foramen ovale, atrial septal defect for cardiology follow-up were reported. Dysmorphic features included low-set ears and a triangular face were noted and baby was referred to geneticists. At 8 weeks of age, patient was re-admitted due to persistent failure to thrive, with static weight and episodes of vomiting. Laboratory tests showed severe hypocalcaemia, hypomagnesemia, hypophosphatemia, markedly elevated parathyroid hormone (PTH), high alkaline phosphatase, and undetectable 25OH-vitamin D levels with low urine Ca/Cr ratio. Baby was admitted to HDU where required continuous intravenous calcium infusion for 23 days, magnesium correction and phosphate supplement along with oral cholecalciferol. Biochemical parameters were not improving with high calcium requirement, persistently elevated PTH and

undetectable vitamin D levels and after 10 days alfacalcidol was added and intramuscular ergocalciferol was given due to concerns of malabsorption. After 3 weeks of treatment 25OHD3 was detectable with decrease in PTH, intravenous calcium was weaned and calcium levels remained stable on oral supplement. Due to static weight special formula was prescribed. Post discharge, baby was gaining weight and laboratory results improved. During inpatient admission many specialities were involved including critical care, gastroenterology, metabolic team, dietetic team, biochemistry and genetic team. Genetic testing including R14 panel have not detected any pathological variants.

Conclusion

Intrauterine vitamin D deficiency can significantly impair fetal growth and mimic rare metabolic or genetic disorders. Early identification and prevention through routine supplementation during pregnancy along with neonatal supplementation, are crucial to optimise infant growth and development. This case highlights the importance of prioritising vitamin D sufficiency as a public health strategy beginning in pregnancy and continuing through early childhood.

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Diabetes 1

P19

Focussing on engagement provides an opportunity for equity of outcomes: no deterioration in glycaemic outcomes in young people aged 15 to 25 years

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Introduction

Adolescence and young adulthood have traditionally been associated with significant deteriorations in glycaemic outcomes, and lower engagement in young people (YP) with type 1 diabetes (T1D).

Objectives

In our city paediatric and adult diabetes teams have held combined transition clinics for 16 to 25 year (y) olds for over a decade. We have prioritised engagement alongside biomedical outcomes. In this study we evaluate glycaemic outcomes across the lifespan.

Methods

Data up to the end of December 2024, of 256 children attending a paediatric hospital, and 2,882 adults attending the corresponding adult hospital were analysed, and latest mean $\pm$ SD and median HbA1cs calculated.

Results

The mean age (y) $\pm$ SD, and duration of T1D (y) $\pm$ SD of children 0-14 y (n = 180) was 10.1 $\pm$ 3.4 and 4.4 $\pm$ 3.3, in YP 15-25 y (n = 486) 20.7 $\pm$ 3.0 and 10.2 $\pm$ 5.6, and in adults $\geq$ 26 y (n = 2472) 50.5 $\pm$ 15.6 and 25.6 $\pm$ 15.7. Engagement was high in all 3 groups, with 99.9%, 95.5% and 92.8% of children, YP and adults respectively having an HbA1c recorded in 2024. Children were more likely to be on HCL, 85.6%, and more YP were on HCL than adults, 56.9% versus 20.1%, P < 0.05. In children aged $\leq$ 14 y, the HbA1c mean $\pm$ SD was 54.2 $\pm$ 8.6, compared to 62.0 $\pm$ 18.5 in YP aged 15 to 25 y, and 64.1 $\pm$ 16.5 in adults aged $\geq$ 26 y. The corresponding medians for children, YP and adults were 53, 57 and 61 respectively. HbA1c outcomes are stable until the age of 12, then increased, before plateauing from the age of 15 to 26.

Conclusions

Engaging young people with T1D in specialist diabetes care throughout transition is of paramount importance. By designing a service according to their needs, they know they are always welcome, attendance rates are high and the service can offer the latest technological advances. We have reduced the magnitude and the number of years over which glycaemic deterioration is usually observed in adolescence and young adulthood, meaning more young people can expect to live with much lower risks of microvascular complications.

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P20

Reducing DNA rates and improving outcomes in a young person diabetes clinic: a quality improvement initiative

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Background

Young people with diabetes often experience a drop in clinic attendance during the transition from paediatric to adult services. This group is particularly vulnerable to disengagement, resulting in poor glycaemic control and long-term complications. In 2021, a young adult diabetes clinic was established at University Hospital Lewisham to address this need. However, initial data revealed a high mean DNA (Did Not Attend) rate of 48.05%. The service was notably under-resourced, lacking a psychologist, youth worker, or dietitian.

Aim

To reduce the DNA rate by at least 50% (target $\leq 24\%$) and assess the impact on glycaemic outcomes (HbA1c levels).

Methods

This quality improvement (QI) project implemented low-cost, person-centred interventions:

- Tailored appointment reminders: letters, texts, and calls at 7 and 2 days pre-appointment
- Telephone consultations for non-attenders
- Joint appointments with the transition CNS, Diabetes nurse, and consultant
- Non-judgemental, supportive clinic environment focused on the individual, not just diabetes
- DNA letters written directly to patients in a personalised and caring tone

Results

Over a 3-year period (2021–2024), the mean DNA rate was reduced from 48.05% to **10.9%**, exceeding the original target. In parallel, patients who attended at least two consecutive appointments showed a **median HbA1c reduction of 13 mmol/mol ($n = 80$)**, reflecting improved diabetes engagement and management. These improvements occurred without additional staffing or funding, utilising existing team resources more effectively. In addition, the HbA1c reduction was greatest in people who had HbA1c of > 64 mmol/mol during transition from paediatrics, with a mean reduction of 12.6 mmol/mol by Wilcoxon test ($P < .002$).

Conclusion

In the context of significant resource limitations, meaningful reductions in DNA rates and improvements in HbA1c can be achieved through personalised, relationship-focused approaches. By reorienting clinic culture to be more youth-centred and supportive, services can better engage this at-risk group.

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P21

Socioeconomic disparities and their effect on hybrid closed-loop adherence and glycaemic outcomes in youth with type 1 diabetes

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Background

The UK National Paediatrics Diabetes Audit (NPDA) 2025 recently identified notable disparities in health outcomes and increasing inequality in access to diabetes technologies among children and young people (CYP) living in the most versus the least deprived areas.

Aim

This study aimed to assess the impact of social deprivation on adherence to hybrid closed-loop therapy and glycaemic outcomes in CYP with Type 1 diabetes (T1D).

Methods

We carried out a retrospective analysis using continuous glucose monitoring (CGM) metrics alongside socioeconomic data derived from the English Index of Multiple Deprivation (IMD). The IMD is a composite index encompassing seven domains—income, employment, education, skills and training, health and disability, crime, barriers to housing and services, and living environment—used to classify areas from most deprived (rank 1) to least deprived (rank 32,844). Variables analysed included diabetes duration, age, average CGM adherence, CGM-derived metrics, and mean HbA1c at 12 months. Correlation analyses and regression modelling were used to explore associations.

Results

There were significant inverse correlations between deprivation measures and glycaemic outcomes. A higher IMD rank (indicating less deprivation) was significantly associated with lower HbA1c at 12 months ($r = -0.26$, $P = 0.007$). Comparable negative correlations were found in IMD sub-domains such as income ($r = -0.226$, $P = 0.01$) and education and skills ($r = -0.294$, $P = 0.002$), suggesting that improved socioeconomic status was linked to better glycaemic control. In regression modelling, IMD rank emerged as a significant independent predictor of mean HbA1c at 12 months ($P = 0.009$).

Conclusion

This study demonstrated that glycaemic outcomes were notably poorer in CYP from the most deprived areas, even when treated with hybrid closed-loop therapy.

Education and employment levels within households were key determinants of successful glycaemic management in CYP with T1D. These findings are consistent with adult data indicating that socioeconomic deprivation contributes to adverse health outcomes. The study highlights the substantial influence of deprivation on the effectiveness of diabetes technologies and supports the need for targeted strategies and policy reform to improve outcomes for disadvantaged CYP.

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P22

Improving asymptomatic screening rates for type 2 diabetes in at-risk children: a quality improvement project in a district general hospital

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Background

Type 2 diabetes in children is an aggressive condition associated with complications including neuropathy, nephropathy, cardiovascular disease, and early mortality. National guidelines, including those from ACDC (endorsed by BSPED), recommend asymptomatic screening in children with a BMI > 85 th centile plus another risk factor, such as family history or high-risk ethnicity. Failure to identify at-risk children delays timely intervention. A baseline audit in November 2024 showed that only 1 in 10 eligible children were being screened at our District General Hospital. A pre-project survey also revealed limited awareness among paediatric doctors of the current screening criteria.

Aim

To increase the rate of asymptomatic Type 2 diabetes screening in eligible children attending general paediatric outpatient clinics to at least 5 in 10.

Methods

The primary outcome was the proportion of eligible children who had an HbA1c test recorded on Cerner following clinic attendance. Children with haemoglobinopathies or a recent HbA1c result were excluded.

Multiple interventions were tested using PDSA cycles. These included:

- Creation and dissemination of an infographic outlining screening criteria (via email, WhatsApp, and posters in clinic rooms)
- Development of a patient information leaflet to support clinician counselling
- A departmental teaching session on screening guidelines
- Individual discussions with consultants to encourage engagement

Results

Screening rates improved from 1 in 10 to nearly 4 in 10 by February 2025. A re-audit four months later confirmed this improvement was sustained. However, no statistically significant shifts were seen on run chart analysis, likely due to variability in clinician decision-making; screening was often omitted when bloods were not otherwise required or when eligibility was borderline.

Conclusion

This project demonstrated that simple, targeted interventions can substantially increase awareness and implementation of Type 2 diabetes screening guidelines in a paediatric outpatient setting. Further strategies may be needed to embed screening into routine practice, particularly for borderline cases or when no other investigations are planned.

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P23

Using set basal rates for “sick day rules” in hybrid closed-loop therapy could result in insufficient insulin delivery

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Background

In acute illness, hybrid closed-loop (HCL) therapy may not cope with illness-associated hyperglycaemia. Temporary increased basal rates in manual mode ($+30\%$ and $+50\%$) can be utilised. However, most pumps deliver basal insulin independently of the set basal rate and may automatically deliver significantly more than the set rate. Reverting to manual mode may lead to lower insulin delivery, even with temporary increased basal rates.

Aims

To review how quickly automated (delivered) basal rates increase in paediatric patients receiving HCL therapy and to predict how outdated set basal rates could affect insulin delivery when using sick day rules.

Methods

Review of clinic documentation and insulin pump data was conducted on a 3-monthly basis up to 12 months for paediatric patients with T1DM receiving HCL therapy in April 2024 at Barnsley Hospital. Data collection was completed at the end of August 2025; 100% of patients had concluded data collection.

Results

86 patients were included; 67% used an Omnipod 5, MiniMed 780G, or CamAPS system. In this group, automated rates increased by an average of 4.6 units over 12 months. In our clinic, set basal rates increased by 4.1 units over the same period. If set basal rates were not updated, by 12 months patients would receive on average only 70% of their usual basal insulin if moved to manual mode. In manual mode with +30% and +50% temporary basal rates, 55% and 45% of patients respectively would receive less insulin than automated mode would have delivered. Sub-analysis by age and HCL system was also undertaken. Automated basal rates increased by 5.1 units on the t:slim X2 pump and set basal rates by 2.4 units. This pump uses set basal rates for automated insulin delivery so was excluded from the temporary basal rate analysis.

Discussion

Patients who use temporarily increased manual basal rates for sickness risk receiving less insulin than they would have in automated mode. There is a clinical risk of diabetic ketoacidosis if teams use this strategy without robust mechanisms to update basal rates. Sick day rules guidance should account for the clinical risk of outdated set basal rates.

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P24

Patient and carer reported perspectives on continuous glucose monitoring in paediatric diabetes: device accuracy in comparison to gold standard

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Background

Type 1 Diabetes Mellitus (T1DM) is associated with significantly increased morbidity/mortality due to chronic hyperglycaemia complications and hypoglycaemic events. T1DM affects many paediatric patients (Prevalence 229.3/100,000, incidence 31.1/100,000 in ages 0-15years, peak incidence 10-14). T1DM also places a burden on parents/families, (30% reduce work hours, 11% leave employment, and 64% report disrupted sleep). Long term outcomes depend on glycaemic control and monitoring. Finger prick testing (FPT) is gold standard, but technological advances have allowed a shift in monitoring toward continuous glucose monitoring devices (CGM). Patients using CGM achieve lower HbA1c, reducing eventual complications of chronic hyperglycaemia. Device accuracy is therefore vital to successful T1DM management.

Methods

Bedford Paediatric Diabetes clinic undertook a study recording paediatric T1DM patient/carers' impressions of their CGM device through a questionnaire assessing device accuracy compared to FPT and identifying barriers to CGM use (MCQ/free text questions).

Results

Of 98 survey respondents, 49% reported ≥ 1 mmol discrepancy between CGM and FPT when device reported normal capillary blood glucose (CBG) (4-10 mmol/l). This discrepancy was higher when the CGM reading was outside of this range (hypo/hyperglycaemic) with 65.9% patients reporting ≥ 1 mmol discrepancy (statistically significant $P = 0.05$), and 71% of these patients reporting ≥ 1.5 mmol ($P = 0.01$) discrepancy. Within normal CBG, 62% of Libre 2 users reported ≥ 1 mmol discrepancy vs 42% of Dexcom users. This discrepancy increased when CGM reported CBG $< 4 / > 10$ mmol/l, with 73% of Libre 2 users reporting ≥ 1 mmol discrepancy vs 48% of Dexcom users and 64% of Libre 2 users reporting > 1.5 mmol discrepancy vs 24% of Dexcom users. Differences in device accuracy were statistically significant in all comparisons ($P = 0.01$). Patient reported barriers to CGM use were: device accuracy (22.4%), skin reactions (20%) and appearance/body image concerns (17%).

Conclusion

CGM is less accurate than gold standard (FPT), with the discrepancy being worse at extremes of CBG. There is notable differences in accuracy between Dexcom and Libre, with Dexcom clearly perceived as more accurate. This study highlights patient perspectives of efficacy of CGM. Further studies are needed to assess quantitative impact on diabetes burden to ensure NHS recommendations for devices maximally improve diabetic control and quality of life.

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P25

Prevalence of neurodevelopmental disorders (ASD/ADHD) in CYP with type 1 diabetes mellitus

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Introduction

Type 1 diabetes (T1DM) is characterized by symptoms caused by elevated blood glucose due to insulin deficiency caused by pancreatic β -cells loss. There may be a potential link between T1DM and neurodevelopmental disorders in children and adolescents (CYP), needing more studies to understand the underlying pathogenesis. Neurodevelopmental disorders, such as attention-deficit/ hyperactivity disorder (ADHD), autism spectrum disorder (ASD) are characterised by learning difficulties, attention, memory and social communication problems. These impairments can undermine patients' abilities to execute complex tasks compromising glycaemic control, increasing risk of complications. The prevalence of ASD in the UK is 1-2% and ADHD is 3-4%. Our aim was to find out prevalence of ASD/ADHD in CYP with T1DM and to compare glycaemic control among patients with and without ASD/ADHD.

Methods

Data was collected to find out CYP with T1DM and ASD/ADHD under Royal Derby Hospital, average HbA1c (lowest, highest and latest), type of treatment (MDI/Pump), presence of associations and complications, age, gender, duration of Diabetes.

Results

Total Patients with T1DM were 271. In Patients with T1DM and either ASD, ADHD or both (21, 7.7% of 271), with ASD were 14 (5.2% of 271), ADHD 3 (1.1% of 271), ASD and ADHD 04 (1.4% of 271), males 9 (42.9%), females 12 (57.1%), mean age 13.3 yrs, mean diabetes duration 6 yrs, median HbA1c mmol/mol 66.3. Patients on MDI therapy 1 (4.8%), insulin pump therapy 20 (95.2%), patients with hypothyroidism 02 (9.5%), Coeliac disease 02 (9.5%) Nephropathy 01 (4.8%). In patients with T1DM without neurodevelopmental diagnosis (250) males were 129 (51.6%), females 121 (48.4%), mean Age 12.8 yrs, mean diabetes duration 5yrs 5months, median HbA1c 65.3 mmol/mol. Patients using MDI 28 (11.2%), insulin pump therapy 222 (88.8%), hypothyroidism 05 (2%), Coeliac disease 11 (4.4%), Nephropathy 03 (1.2%), Hypertension 01 (0.4%).

Discussion

Our data showing increased Prevalence of ASD/ADHD in CYP with T1DM highlights the importance of early screening for neurodevelopmental disorders in children with Type 1 DM. Higher HbA1c in CYP with T1DM and neurodevelopmental disorders (ASD/ADHD) favours considering enhanced standards for management in patients with T1DM having neurodevelopmental disorders.

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P26

Clinical characteristics and epidemiological patterns of initial presentation of diabetes mellitus in children and young people: a single-centre study in Sri Lanka

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Background

The majority of children and young people (CYP) with Type 1 and Type 2 diabetes mellitus (DM) present with diabetic ketoacidosis (DKA) at diagnosis. Presentation in DKA has been associated with younger age, socio-economic deprivation, ethnic background, and absence of a family history of DM.

Aim

To describe the clinical and epidemiological characteristics of children presenting with newly diagnosed DM at one of the largest tertiary referral centres in Sri Lanka.

Methodology

A retrospective cohort study was conducted using case notes of all children with new-onset DM registered between January and December 2022 at the Lady Ridgeway Hospital for Children, Colombo, Sri Lanka.

Results

Fifty-six children (19 males, 37 females) with newly diagnosed DM were included. The mean age at presentation was 9.75 years (range 1.5–14.75years). Fourteen children (25%) presented in DKA: mild-5 (8.9%), moderate-3 (5.4%), and severe- 6 (10.7%). The median duration of symptoms was 14 days (range 2 days-1year). The most common symptoms at presentation were polyuria (94.5%), polydipsia (92.7%), and weight loss (92.7%). Only two children presented with impaired consciousness. Acanthosis nigricans was noted in 11

children. Children whose fathers had completed tertiary education experienced a significantly shorter mean symptom duration before admission compared to those whose fathers had only primary education (9.67 vs. 46 days, $P = 0.02$). Although not statistically significant, a similar trend was observed with maternal education (14 vs. 39 days for tertiary vs. primary/ordinary level education). Age, ethnicity, family history of DM, parents' education level and distance from the local hospital were not significantly associated with the risk of DKA at presentation.

Conclusion

The incidence and severity of DKA in this cohort are comparable to data from developed countries, with most cases being mild to moderate. Parental education—particularly paternal tertiary education resulted in earlier healthcare seeking behaviour and significantly shorter duration of symptoms before diagnosis.

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P27

Hybrid closed-loop treatment technology implementation and evaluation on glycaemic outcomes in children and young people living with type 1 diabetes

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Background

NICE Technology appraisal guidance (TA 943) published December 2023 recommended Hybrid-closed loop (HCL) pumps as treatment options for all Children and Young People (CYP) with Type 1 Diabetes. The NHSE outlined an implementation strategy highlighting children and young people (CYP) as a target group to roll-out this therapy. We sought to evaluate the delivery of HCL systems over 1-year period to CYP in our paediatric diabetes unit (PDU)

Methods

We conducted a retrospective study looking at CYP commenced on HCL pumps between 01/10/2023 – 30/09/2024. Health outcomes analysed include median HbA1c and average glucose time in range (TIR). Data was also collected on patient ages, deprivation status (IMD1-5, 1 – most deprived, 5 – least deprived).

Results

45 patients were identified during this period. 7 patients excluded due to leaving or transfer of care to young adult team following transition. 38 patients were included and belonged to age 3-18 years. The median age was 14 years. 11 patients (29%) were from IMD-1 most deprived group. 76.3% (29 patients) showed improvement in HbA1c post-HCL start, 21.1% (8 patients) showed no change or worsening HbA1c and 2.6% (1 patient) maintained good HbA1c. Proportion of CYP achieving NICE HbA1c target 48 mmol/mol increased from 7.9% to 23.7% post-HCL ($P < .031$). Those with HbA1c 80 mmol/mol or higher reduced 18.4% to 2.6% post-HCL. Median HbA1c decreased 61 mmol/mol to 54 mmol/mol ($P < .001$). Median glucose TIR increased from 51% to 65%, (1 patient excluded due to lack of pre-HCL TIR data). 8 CYP had no change or worsening in HbA1c and of them 4 (50%) were from most deprived quintile IMD-1 and 4(50%) were 16-18 age group.

Discussion

Most of the CYP improved their HbA1c and increased time in range following HCL therapy, with median HbA1c decreasing significantly. More CYP achieved NICE treatment target. Half of the patients showing no improvement were from most deprived quintile and were in 16-18 yrs age. CYP in transition and most deprived requires close monitoring despite advanced treatment technology. Limitations in our study is small sample size. Further research is needed to understand deprivation and age influencing outcomes in CYP on HCL technology.

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P28

Characterising type 2 diabetes in a multidisciplinary clinic: findings from a single-centre cohort

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Background

Adolescent type 2 diabetes mellitus (T2DM) is a complex condition with increasing prevalence and rapidly changing management strategies. We characterise patients and treatment approaches in our multidisciplinary paediatric T2DM clinic.

Method

Data was collected retrospectively using electronic patients records for all current patients with T2DM.

Results

Ten patients with T2DM were identified; 80% female, mean age at diagnosis 13.0 years (± 1.9 SD) years. Six were White British, two South Asian and two Black African. Mean follow-up was 2.8 years (± 2.0 SD). Five patients were diagnosed through screening for HbA1c in the complications of excess weight clinic. Two presented with osmotic symptoms and were initially managed as type 1 diabetes mellitus. Two were diagnosed through investigations for other conditions, with one patient having vaginal candidiasis and another autoimmune encephalitis. One patient had a severe perineal infection at diagnosis, presenting in sepsis with diabetic ketoacidosis. 40% had a diagnostic oral glucose tolerance test; average fasting blood glucose 4.7 mmol/l (± 0.5 SD) and 9.9 mmol/l (± 3.1 SD) at 120 minutes. C-Peptide was measured in 80%; mean value 1875pmol/l (± 1553.2 SD). 80% had diabetes autoantibodies measured: negative in all patients except one with concurrent GAD autoimmune encephalitis managed with high dose steroids. Mean HbA1c at diagnosis was 80.4 mmol/mol (± 36.8 SD) and 57.3 mmol/mol (± 21.4 SD) during follow-up. All patients were obese at diagnosis: mean BMI 31.4 kg/m² (± 4.6 SD) (BMI SDS +2.91 (± 0.54 SD)), and 31.2 kg/m² (± 5.1 SD) (BMI SDS +2.46 (± 1.07 SD)) during follow-up. 70% had a strong family history of T2DM. Patients had access to a specialist dietitian, clinical psychologist and paediatric diabetes nurse specialist at medical appointments. 50% were receiving monotherapy (80% metformin; 20% sodium-glucose cotransporter 2 (SGLT-2) inhibitor) and 50% a combination of medications (one metformin/SGLT-2 inhibitor; two metformin/insulin and two metformin/SGLT-2 inhibitor/glucagon-like peptide 1 (GLP-1) agonist). 40% had hepatic steatosis; one hypertension, one bilateral proliferative retinopathy from diagnosis and nephropathy and one is being investigated for obstructive sleep apnoea.

Conclusions

Our cohort is predominantly female and White British, with half detected through screening. Metformin is still widely prescribed but newer therapies are being adopted. With early-onset of complications in adolescence, further clinical trials are urgently needed to identify optimal management strategies.

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Diabetes 2

P29

Improvement in insulin sensitivity and glycaemic control in a type 1 diabetes patient following initiation of a GLP-1 receptor agonist for excess weight management

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Background

Patients with type 1 diabetes mellitus (T1DM) who have high insulin requirements may have concurrent insulin resistance, often associated with obesity or features of metabolic syndrome.

Case Report

We present case of a 17-year-old patient with long-standing T1DM and persistently high total daily insulin requirements, suboptimal glycaemic control, and features of insulin resistance, including elevated BMI. The patient was diagnosed with T1DM at the age of 7 and initially managed with multiple daily insulin injections. Insulin requirements were consistently high (>1 unit/kg/day), and between ages 11–12, ranged from 1.3 to 1.7 units/kg/day. HbA1c values fluctuated between 59–88 mmol/mol, and target glycaemic control was not achieved. An insulin pump was initiated at age 12, and metformin was later added to improve insulin sensitivity. At age 14.5 years, a closed-loop system (Control-IQ) was introduced, resulting in a modest improvement in HbA1c and time-in-range (TIR: 44–56%), but insulin requirements remained high. The patient's BMI continued to increase, and they experienced symptoms of diabetes burnout, for which psychological support was offered. The patient had ongoing input from dietitians and continued metformin. Insulin requirements remained between 1.5–1.7 units/kg/day with high HbA1c. At the age 16.5 years, with a BMI z-score (3.02), dyslipidaemia, and elevated HbA1c, semaglutide was initiated for weight management. The dose was titrated from 0.25 mg to 1 mg weekly. The patient reported reduced insulin requirements. Patient's HbA1c and lipid profile improved and a 5% weight loss within 4 months was noticed. After 9 months, HbA1c had improved to 60 mmol/mol, weight had decreased by 9% (BMI z-score 2.56), total daily insulin dose was reduced to 1.05 units/kg/day, and TIR improved to 61%. Despite irregular bolusing, patient reported that diabetes management felt easier and their overall well-being had improved.

Summary

Although not licensed for use in T1DM, glucagon-like peptide-1 receptor agonists may offer significant benefits in selected patients by improving glycaemic control, reducing insulin requirements, and supporting weight and metabolic management.

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P30**Impact of hybrid closed loop (HCL) systems on children and young people with type 1 diabetes**

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Introduction

Hybrid closed-loop (HCL) insulin pump systems are increasingly utilised in managing Type 1 Diabetes Mellitus (T1D) in Children and Young People (CYP). These systems aim to optimise glycaemic control by improving Time in Range (TIR), reducing HbA1c, and easing the burden of daily diabetes management.

Aim

This retrospective cohort study aimed to evaluate the impact of HCL insulin pump systems on glycaemic outcomes in CYP with T1D by assessing changes in TIR, HbA1c, and Time Below Range (TBR) before and after HCL initiation and comparing outcomes across different HCL pump types.

Methods

CYP with T1D from three paediatric diabetes centres within Barts Health NHS Trust (Royal London, Whipps Cross, and Newham Hospitals) who had transitioned to HCL systems since January 2022 and used them for at least 12 months were included. Data were collected retrospectively from clinical records, the Twinkle paediatric diabetes management system, and insulin pump downloads. Outcomes were compared before and at 2 weeks, 3 months, and 12 months post-HCL initiation. Patients with less than 12 months of HCL use or incomplete data were excluded.

Results

A total of 200 CYP were included (mean age 13.5 ± 3.9 years; 108 male, 92 female). Devices used were: Medtronic 670G ($n = 10$), Medtronic 780G ($n = 26$), Tandem T-Slim Control IQ ($n = 21$), Ypsomed CamAPS FX ($n = 12$), and Omnipod 5 ($n = 131$). The average duration of HCL use was 1.47 ± 0.5 years. HbA1c improved significantly from 65.3 mmol/mol pre-HCL to 57.2 mmol/mol at 3 months ($p < 0.001$) with sustained improvement at 12 months (56.4 mmol/mol, $p < 0.001$). TIR improved from 37.4% to 52.3% at 2 weeks ($P = 0.004$), 53.6% at 3 months, and 53.2% at 12 months ($p < 0.001$). TBR increased transiently at 2 weeks ($2.6\% \pm 2.46$, $P = 0.10$) compared to baseline ($1.9\% \pm 2.66$) but returned to baseline by 12 months ($1.92\% \pm 2.0$, $P = 0.40$). No significant differences between pump types ($P = 0.45$).

Conclusion

HCL systems significantly improved glycaemic control in CYP with T1DM. The results highlight the effectiveness of these systems in paediatric diabetes management and support the broad use of HCL in CYP with T1DM.

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P31**Symptomatic hypophosphatemia in a teenage girl with severe diabetic ketoacidosis**

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Diabetic ketoacidosis (DKA) is a medical emergency that is associated with hyperglycaemia, metabolic acidosis, and electrolyte imbalances. Hypophosphatemia commonly occurs during DKA treatment but is typically mild and self-limiting. Current BSPED guidelines do not recommend routine phosphate replacement unless symptoms of severe deficiency develop. We present a case of severe symptomatic hypophosphatemia in an adolescent with DKA, highlighting the clinical impact and need for early recognition.

Case Summary

A 14-year-old previously healthy girl presented to paediatric ED in severe DKA with a pH of 6.83 and glucose of 37 mmol/L. She was tachycardic and drowsy (GCS 14/15) on arrival and managed as per BSPED guidelines. Her initial serum phosphate level was normal at 1.24 mmol/L (0.9-1.8 mmol/L). Despite ongoing management, over the next 24 hours, she developed worsening acidosis, acute kidney injury, and myocardial dysfunction, confirmed on echocardiography. By 16 hours post-admission, phosphate had fallen to <0.1 mmol/L, prompting oral replacement with phosphate Sandoz, however this was not tolerated due to ongoing nausea and dysphagia. Antiemetic therapy with ondansetron, cyclizine and pantoprazole was trialled but was ineffective. By day four, she remained on IV insulin with her

symptoms persisting and additionally developed confusion, hallucinations and extreme weakness which were attributed to very low phosphate levels. Given ongoing symptoms and persistent hypophosphatemia, she was commenced on intravenous phosphate (Phosphate Polyfusor). By day six, serum phosphate had normalised, and her clinical condition had improved.

Discussion

Phosphate is lost during DKA due to osmotic diuresis and serum phosphate is often low during the recovery phase. Severe hypophosphatemia can lead to neurological symptoms, myocardial dysfunction, ileus, and myopathy. This case demonstrates that severe hypophosphatemia can cause significant morbidity in DKA and may not respond adequately to oral replacement alone. However, we considered whether her symptoms had been aggravated by the severity of dehydration at presentation.

Conclusion

This case highlights the need for phosphate monitoring in severe DKA and increased awareness of clinical signs of phosphate depletion. Current UK paediatric guidelines recommend replacement for symptomatic cases, however the lack of paediatric specific thresholds for treatment highlights the need for further research.

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P32**Type 2 diabetes in children and adolescents: presentation, treatment, and outcomes from a single-centre audit**

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Aim

Review of the demographics, clinical presentation, management and monitoring of complications in children and young people with type 2 diabetes.

Methods

A retrospective review of medical records was conducted for patients with type 2 diabetes under the care of the Paediatric Diabetes Service at Oxford University Hospitals.

Results

At the time of review, 13 patients (76% female, mean age of 16.2 ± 2 years) were under care of the service. The mean age at diagnosis was 13.7 ± 2.45 years. 38% patients were Asian, followed by White British (30%). A family history of type 2 diabetes was present in both parents for 5 patients and in one parent for 4 patients; for the rest of patients it was unknown. At diagnosis, 30% of patients presented with classical symptoms (polyuria and polydipsia), while 40% presented with nonspecific symptoms such as fatigue or were diagnosed during screening for excess weight by elevated HbA1c. All patients tested negative for diabetes-related antibodies. The mean HbA1c at diagnosis was 81 ± 39 mmol/mol (median 66). Initial treatment included metformin in 70% of patients. Basal insulin was required from diagnosis in 38%, and 3 patients commenced on basal-bolus therapy with metformin added once antibody results were available. At the most recent review, 76% of patients remained on metformin; half of these were also prescribed empagliflozin. Two patients were treated with a GLP-1 receptor agonist. Complications included hypertension (38%), hypercholesterolemia (30%), metabolic dysfunction-associated steatotic liver disease in 46%, and polycystic ovary syndrome in 30% of female patients. Two patients had a diagnosis of autism spectrum disorder, 2 anxiety, and 1 was on child protection plan. Review of patient records revealed that not all individuals had full screening for excess weight and diabetes-related complications in first year post diagnosis. At 6 months post-diagnosis (data available for 11 patients), the mean HbA1c was 55 ± 21 mmol/mol. The most recent HbA1c (10 patients) was 66 ± 10 mmol/mol.

Summary

The most recent median HbA1c remained above the therapeutic target. To improve care pathways we have updated the local guideline and developed proforma for the first appointment and annual reviews.

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P33**Changing needs, user improvement suggestions and satisfaction with hybrid closed-loop insulin pump therapy**

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Aim

This study aimed to evaluate the changing needs, improvement suggestions and user satisfaction with hybrid closed loop (HCL) insulin pump therapy, focusing on its impact on daily life and diabetes management in a real-world setting.

Methods

A structured questionnaire was developed using Microsoft Forms and distributed initially via Digibete platform. Families were approached in outpatient clinic (OPD) and given the option to participate in this anonymised feedback. The survey collected information on the duration of diabetes, length of HCL pump use, and the perceived impact on glycaemic control and quality of life, including anxiety related to diabetes care, hypoglycaemia, and sleep. Participants were also asked about the Diabetes updates they would like and were invited to provide free-text comments on suggestions for improvement.

Results

Of the 29 families approached in OPD, 27 completed the questionnaire. Ninety-six percent reported that the system met their expectations, while 4% felt it did so to some extent. Patients ranged in age from 6 to 16 years and had used HCL therapy for 5 to 18 months. Eighty-nine percent reported improved time-in-range for blood glucose levels, and all respondents mentioned improvements in glycaemic control during meals, exercise and overnight and recommend HCL to others. All participants reported reduced fear of hyperglycaemia, hypoglycaemia, and diabetic ketoacidosis. Seventy-eight percent felt safer using HCL and 63% reported reduced anxiety and improved sleep. Users reported that the system was beneficial when mealtime insulin was missed. Positive descriptive terms such as “game changer” and “magic wand” were used to describe their experience. Desired updates included guidance on pump failure, reinforcing the difference of managing parameters like hypoglycaemia to overcome previous habits, skin care, travel with pump and sick day management using HCL. Users suggested improvements included full automation, technical prompts to return to Auto Mode, wider infusion site options, and simplified mealtime adjustments without bolus.

Conclusion

This evaluation demonstrates high levels of user satisfaction with HCL insulin pump therapy, with significant perceived improvements in glycaemic control, safety, and quality of life. The findings also highlight evolving user needs with change in technology and areas for future technological development.

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P34

Optimising paediatric DKA management from emergency department to ward: integrating human factors through visual aids and competency tools

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Aim

To evaluate the management of paediatric diabetic ketoacidosis (DKA) across the Emergency Department (ED) and Paediatric Ward, focusing on adherence to locally adapted BSPED guidelines and identifying and addressing the factors contributing to deviations.

Methods

A retrospective audit was conducted on all Paediatric DKA presentations to the ED between January and March 2025. Management was assessed against key BSPED DKA guideline components: diagnosis, clinical observations, fluid and insulin administration, investigations, and transition to subcutaneous (SC) insulin. Deviations were analysed, and contributing factors identified. In response, an action prompting aid and medical and nursing staff DKA competency tools were developed.

Results

Six patients presented with DKA during the study period, most being new diagnoses; one had known type 1 diabetes. Three patients were diagnosed to have severe DKA, and three mild to moderate DKA. Two of them attended the primary care physician in the preceding week. Diagnosis and initiation of treatment were timely in all cases. Initial fluid boluses were appropriate, though in one case with shock, the bolus was incorrectly subtracted from total fluid calculations. Intravenous insulin was initiated promptly in all but two cases, where delays were due to the lack of a second functioning cannula. Monitoring met standards in severe cases but was delayed in mild to moderate DKA. Transition to dextrose-saline fluids and SC was appropriate in all but one case, where a delay occurred during ED-to-ward transfer. An action prompting visual flowchart, designed to support real-time nursing decision-making, received excellent feedback and integrated into the ED app. Competency tools for both medical and nursing staff were well received, with training now aligned to these standards.

Conclusion

DKA management aligned well with BSPED guidelines for most patients. Deviations such as delayed insulin initiation, fluid miscalculations, and transition delays, were primarily linked to human factors. The introduction of visual aids and competency tools have been positively received and are expected to improve compliance, safety, and outcomes in our Paediatric DKA care.

Reference

1. **BSPED Guideline for the Management of Children and Young People under the age of 18 years with Diabetic Ketoacidosis – 2021.** Available: <https://www.bsped.org.uk/clinical-resources/bsped-dka-guidelines/>

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P35

Integrating ultralong-acting insulin with HCL pumps in high-risk diabetes management: bridging the safety concern

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Background

Hybrid closed loop (HCL) insulin pump systems have demonstrated improvements in glycaemic control, time in range (TIR), frequency of hypoglycaemia and quality of sleep for children and young people when using HCL for 6 months². NICE recommends considering HCL for children with type 1 diabetes¹. However, initiating HCL therapy in high-risk patients, particularly those with poor glycaemic control and inconsistent adherence to treatment, raises concerns about the potential for diabetic ketoacidosis (DKA). To mitigate this risk, we incorporated ultralong-acting insulin (Insulin Degludec) during the transition phase to HCL therapy.

Aim

To assess the impact of combining ultralong-acting insulin with HCL insulin pump therapy on glycaemic outcomes, body mass index (BMI), and quality of life in high-risk individuals with type 1 diabetes.

Methods

Patients with persistently elevated HbA1c and suspected disguised non-compliance were initiated on HCL insulin pump therapy. As a safety measure, 30% of their basal insulin requirement was provided as insulin Degludec (Tresiba) to reduce the risk of DKA during the transition. Initial glucose targets were conservatively set at 8.3 mmol/l to avoid rapid glycaemic shifts and potential retinopathy. Outcomes assessed included changes in HbA1c, TIR, time above range (TAR), Body mass index (BMI), and patient-reported quality of life.

Results

Five patients were initiated on this combined regimen. Four of them continued HCL therapy successfully. Their Mean HbA1c decreased from 92.25 pre-HCL pump start to 67 mmol/mol on HCL, with the mean TIR improving from 19.5% to 53.75%. The mean TAR, particularly time spent in the very high blood glucose range, significantly reduced. No episodes of DKA or clinically significant hypoglycaemia were reported. All participants noted improved quality of life and engaged better with diabetes care. BMI trends showed improvement in the underweight patients and maintenance or reduction of BMI in the children who were overweight.

Conclusion

The adjunctive use of ultralong-acting insulin during HCL pump initiation appears to be a safe and effective strategy for high-risk patients. It supports improved glycaemic control, enhances patient engagement and seems to reduce the risk of acute complications without compromising safety.

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P36

Closing the loop on high HbA1c in paediatric type 1 diabetes with hybrid closed loop insulin pump therapy

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Aim

To evaluate the impact of Hybrid closed loop (HCL) insulin pump therapy on children and young people with type 1 diabetes who had high HbA1c levels of ≥ 69 mmol/mol, prior to initiation of HCL therapy and to identify factors associated with suboptimal response to HCL therapy.

Methods

Our analysis focused on children with type 1 diabetes who had an HbA1c level of ≥ 69 mmol/mol at the time of initiation on Hybrid Closed Loop (HCL) insulin

pump therapy. Outcomes assessed included changes in HbA1c, TIR, time above range (TAR), and the occurrence of significant hypoglycaemia or diabetic ketoacidosis (DKA). For patients who did not show improvement, potential contributing factors were explored.

Results

A total of 32 patients with baseline HbA1c >69 mmol/mol were included. The mean HbA1c decreased from 71.4 mmol/mol at initiation of HCL to 56.2 mmol/mol at 3 months and stabilised around 60 mmol/mol at both 6 and 9 months. By 9 months, only 7.2% of patients had a HbA1c above 68 mmol/mol. Common factors associated with lack of sustained improvement included missed meal boluses and failure to return to Auto Mode after switching to Manual Mode. The mean TIR improved from 34.9% at baseline to 58.3%, with this improvement of TIR >50% maintained over 12 months. Time above range decreased from a mean of 65% to 37.8%, with a notable reduction in the time spent in the very high range (from 37.6% to 11.3%). No significant hypoglycaemic events were reported during the study period. Two patients were admitted with mild DKA due to pump failure.

Conclusion

HCL insulin pump therapy significantly improved glycaemic control in children and young people with initially high HbA1c levels, in our cohort of patients. Improvements in HbA1c, TIR, and TAR were sustained over time, with no major safety concerns observed. Identifying and addressing behavioural patterns such as missed boluses and not reverting back to automode, may further enhance outcomes.

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P37

Prevalence and risk factors for diabetic retinopathy in children with type 1 diabetes: an update from a single centre

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Introduction

Diabetic retinopathy (DR) is a leading cause of vision loss in the UK and poor glycaemic control (HbA1c) is a key modifiable risk factor. Although sight-threatening retinopathy is rare in childhood, it is well-established that early changes of retinopathy can be seen in children and young people (CYP). From National Paediatric Diabetes Audit longitudinal data, HbA1c has reduced significantly over the last decade, but CYP retinopathy rates have remained relatively static. We demonstrated, in 2008, that the prevalence of background DR was 19.5% with a median HbA1c of 73 mmol/mol (1). This study aimed to explore if DR prevalence had changed and associations with clinical and/or socio-demographic factors.

Methods

Retrospective review of electronic health records of all CYP with Type 1 Diabetes (T1D) eligible for DR screening (>=12 years) between 2022-2024. All values are median unless stated otherwise.

Results

84 eligible children were offered screening. Of these, 16 (19%) did not attend their offered screening appointment. In those who attended screening, DR was detected in 6/68 (8.9%). All had background retinopathy (R1) with no maculopathy. Table 1 summarises key findings. Patients with DR showed no significant difference in HbA1c ($P = 0.1$), duration of diabetes ($P = 0.8$), ethnicity ($P = 0.3$) or socio-economic status ($P = 0.4$). Notably, R1 retinopathy does not automatically progress. In both cohorts no DR progression was seen across 2 audit years.

Conclusions

DR prevalence has reduced by over 50% over 17 years in our centre. Although there appeared to be differences in the DR cohort for HbA1c and duration of diabetes this was not found to be significant, probably due to the small number in the DR group. Further work needs to be done to explore these initial data.

Table 1

	2022-2024 Diabetes duration (years, IQR)	HbA1c (mmol/mol, IQR)		2008-2010 (1) Diabetes duration (years, range)	HbA1c (mmol/mol, range)
DR (n = 6)	5.3 (1.7-8.9)	68.5 (49-73)	DR (n = 30)	7.7 (0.6-13.7)	76 (55-130)
No DR (n = 62)	3.0 (1.7-5)	56 (39-101)	No DR (n = 119)	5 (0.2-12.5)	70 (38-120)

Reference

1. Dhillon *et al* (2016). Natural history of retinopathy in children and young people with type 1 diabetes. *Eye*, 30(7), pp.987-991.

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P38

Review of paediatric admissions of known diabetics in the belfast trust february 2023 - january 2025

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Introduction

We undertook a review of all known paediatric diabetic admissions in the Belfast Trust during a two year period between February 2023 and January 2025. We reviewed number of admissions, reason for admission, age, length of stay, HbA1c and need for Paediatric Intensive Care. We wanted to ascertain the proportion of known diabetics that were admitted, how many were admitted with diabetic ketoacidosis (DKA) and what demand this had on our workforce.

Methodology

A previous list of patients collected by our Paediatric Diabetic Specialist Nurses was used. Missing data was identified and collected using electronic records and paper notes. Using these methods we were able to acquire majority of all the data we needed.

Results

Over a two year period we found 35 admissions for 18 different patients. 6 patients were admitted multiple times, 4 of which were over 12 yrs of age. One patient was admitted a total of 7 times over a 2 year period. 51% of admissions were due to DKA. Our longest stay was 82 days for investigation of Subcutaneous Insulin Resistance Syndrome. The median length of stay was 2 days. Of those admitted with DKA, only 28% required Paediatric Intensive Care. Mean length of stay for DKA was 2.7 days, range 1 – 10 days. The majority of our DKA admissions were over 12 yrs of age and had a higher HbA1c compared other admissions. Admission rate for our known diabetics is 17.5 per year. Our workforce calculations showed that 9% of our current case load of patients were admitted per year which equated to 42.5 hours of our workforce time per year.

Conclusion

9% of our current caseload per year of known paediatric diabetic patients get admitted. Half of admissions are due to DKA, majority are over 12 years old and have high HbA1c. We use a traffic light system for a management of high HbA1c and recently started over 12 years diabetic pump clinic. We are considering further age banded and high HbA1c clinics to reduce rate of DKA admission and work force time.

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Diabetes 3

P39

Evaluating the effectiveness of local network perioperative diabetes guidelines in great ormond street hospital (GOSH)

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Background

There are >35,000 children and young people (CYP) living with diabetes in the UK (NPDA 2024). The options to support management of diabetes is evolving and can make the surgical management of CYP a challenge. The North Central London (NCL) network developed standardized perioperative guidelines for managing Diabetes during surgery.

Aim

To assess adherence to local perioperative diabetes guidelines for patients with Diabetes undergoing surgery at GOSH.

Method

In 2022 GOSH introduced a perioperative plan field into the electronic health record to be completed at pre-assessment or on admission to GOSH. We retrospectively reviewed records from patients with a diabetes perioperative plan documented between 2022 and 2025. We assessed whether management aligned with guidelines and identified reasons for deviations.

Results

Of 92 patients with a perioperative plan, 86 (93.5%) underwent surgery. Preoperative HbA1c was documented in 54.7% of cases. Minor surgery (<90min) was performed in 53.5%, major surgery (>90min) in 45.3%, with one case (1.2%) undocumented. Of the patients admitted on CSII, 36.9% had the surgical procedure in 'manual mode', 34.7% pump mode not stated, 28.2% on sliding scale. Of the patients admitted on MDI one required sliding scale. There were 11.6% intraoperative complications of

which 60% were related to diabetes (hypoglycaemia). Guideline deviations were observed in 44.2% of cases:

- 28.9% lacked documentation regarding pump delivery mode
- 23.7% incomplete intraoperative BG monitoring
- 7.9% lacked prescription of diabetes medication
- 7.9% retained sensors intraoperatively
- 28.9% other

Postoperative complications occurred in 19.8% of cases, with 52.9% related to hypoglycaemia/hyperglycaemia. Discharge was delayed in 15.1%, with 61.5% of delays due to diabetes.

Conclusion

Perioperative plans are consistently created by the Diabetes CNS team, yet documentation gaps and deviations from protocol persist. Improved multidisciplinary communication, adherence to guidelines, and documentation—particularly around pump/sensor management and BG monitoring—are essential to optimize perioperative diabetes care.

Reference NPDA

1. Reference diabetes surgical guidelines

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P40

A novel non-autoimmune insulin-deficient diabetes subtype in a young child

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Background

Autoantibody-negative type 1 diabetes (also named atypical diabetes, idiopathic diabetes, type 1b diabetes or “ketosis-prone” diabetes) is a heterogeneous and understudied group. There is growing evidence for a subgroup of diabetes patients in Sub-Saharan African populations who are autoantibody negative with low type 1 genetic risk scoring and higher endogenous insulin production but without features of type 2 diabetes.

Case

6-year-old boy of Somali ethnicity presented with a 4-week history of polyuria, polydipsia and weight loss of 4.5 kg. He had an initial blood glucose of 24 mmol/l. He was ketotic (5.1 mmol/l) but not in diabetic ketoacidosis. HbA1c at diagnosis 109 mmol/mol; BMI was 16 with no acanthosis nigricans. There was a family history of diabetes (paternal aunt with type 2 diabetes and paternal second cousin diagnosed with type 1 diabetes at 18 months). He was started on a multiple daily injection regime of insulin. Insulin was stopped completely at 5 months after diagnosis, due to recurrent hypoglycaemia. He remained off insulin for 16 months and currently has a very low but increasing insulin requirement of 0.1 unit/kg/d 4 years after diagnosis. He was triple antibody negative (to glutamic acid decarboxylase (GAD), insulinoma-associated antigen 2 (IA-2), and zinc transporter 8 (ZnT8)) at diagnosis and on two further occasions. Urinary C-peptide creatinine ratio (UCPCR) at one year after diagnosis was 0.58 nmol/mmol. Blood C-peptide level 4 years after diagnosis <94pmol/l. Exome sequencing of all known monogenic diabetes genes was negative.

Conclusion

We report a paediatric patient of Somali ethnicity with an atypical diabetes who was ketotic at presentation, without autoimmunity and with prolonged low insulin requirement. We highlight the importance of being aware of a potentially new subgroup of children and young people with diabetes of Sub-Saharan African ancestry in the UK; further work will highlight the pathophysiology and treatment strategies for this group of patients.

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P41

Improving care for young people with type 2 diabetes in an ethnically diverse and socioeconomically deprived area of London: a local audit and pathway redesign

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Type 2 diabetes (T2DM) in children and young people (CYP) is an emerging challenge within paediatric diabetes services. We present findings from a local audit and describe the development of a new care pathway addressing the complexity within this population. Our diabetes service, based in a socially deprived and culturally diverse borough of London, currently cares for (8/171)

T2DM patients. Five were from South-Asian backgrounds, 2 African/African-Caribbean and 1 Caucasian ethnicity. Six were from low socioeconomic groups. Four were male. The mean age of diagnosis was 12.6 years. Patients were seen in a predominantly T1DM clinic. The cohort is further characterised by a strong family history of diabetes ($n = 5$), high prevalence of severe obesity ($n = 7$), neurodiversity ($n = 2$), and mental health issues ($n = 2$). There was also poor engagement with care ($n = 2$), poor school attendance ($n = 1$), home schooling ($n = 1$) and one child living under special guardianship. Most patients received key annual NPDA care processes. Retinal screening had lowest completion rate ($n = 1$). Co-morbidities identified included: acanthosis nigricans ($n = 5$) hypertension ($n = 1$) high cholesterol ($n = 3$) raised albumin: creatinine ratio ($n = 1$) NAFLD on liver ultrasound ($n = 3$) features of OSA ($n = 1$) All had been offered CGM and 6 were using this technology. Treatment regimes varied: Four were prescribed Metformin. Five were taking Insulin. One patient was taking no treatment. One patient was prescribed Dulaglutide. Several had reported side-effects with treatments resulting in non-compliance or deviation from first line therapies. None had reduced weight by 5% at 3 months or 10% at 1 year. In response, we co-developed a new localised care pathway prioritising holistic, personalised, and family-centred care delivered through a strengthened multidisciplinary team (MDT) approach. Key elements include structured education, standardisation of care processes and medication, culturally sensitive communication (e.g. Ramadan considerations) and signposting to local physical activity and weight management services. Re-audit data is being collected and will be available before November 2025. The aim is to improve engagement and use of local resources. This work highlights the level of complexity of T2DM and highlights the importance of context-specific models of care. A localised pathway is intended to improve outcomes for this vulnerable group.

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P42

Bridging faith and care: ramadan fasting micro-teaching for muslim children and young people with type 1 diabetes

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Introduction

Fasting during the holy month of Ramadan is one of the five pillars of Islam. It requires abstaining from all food and drink from dawn until dusk. While exemptions are granted to those with medical conditions such as diabetes, many Muslim children and young people (CYP) with diabetes still choose to fast. Without appropriate guidance and awareness of both medical considerations and religious rulings, fasting can be challenging. We collected evidence from Islamic literature to support Muslim CYP with type 1 diabetes in deciding about fasting and also to provide some guidance about fasting while on hybrid closed-loop systems (HCL) or Multiple daily injections (MDI). Micro-teaching in the clinic waiting area is a local initiative that has been implemented for years as part of our diabetes team's quality improvement. This was seen as a good opportunity to provide education and counselling about fasting before the start of Ramadan.

Methods

Islamic literature was collated to provide evidence on key rulings regarding fasting and insulin treatment. A review of the literature on fasting Ramadan in children and young people with diabetes was also conducted to determine best practices. Proposed guidance on blood sugar cut-offs to break the fast was compiled and presented to the wider Diabetes MDT for feedback.

Results

Guidance from the Holy Quran and Islamic resources were reviewed to provide clarity on three key areas:

1. Religious allowances for the sick to abstain from fasting
2. The Islamic view on adherence to medical advice
3. Confirmation that insulin administration and blood glucose monitoring do not invalidate the fast Cut-off values for when to break the fast if fasting was started, as well as further management regarding hypoglycaemia and hyperglycaemia, were added to create a resource about fasting as micro-teaching slides. A leaflet was also designed for take-home messages.

Further Work

CYP who fasted last Ramadan and their families will be surveyed about their perceptions of the slides and leaflets, and asked to participate in a pre- and post-quiz on the slides to assess the improvement in their knowledge.

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P43

Assessing the impact of hybrid closed-loop systems on sleep in children with type 1 diabetes and their families at a tertiary diabetes centre in LondonLee Fernandes¹, Karen Logan² & Sudeshna Ghosh²¹Faculty of Medicine, Imperial College London, London, United Kingdom;²Department of Paediatrics, St. Mary's Hospital, London, United Kingdom**Introduction**

Hybrid closed-loop (HCL) systems have been shown to improve glycaemic control and treatment satisfaction in children and young people with Type 1 Diabetes Mellitus (T1DM), including increased time-in-range and reduced hypoglycaemic events. While HCL may reduce nocturnal glycaemic variability and improve sleep, few have explored this relationship qualitatively to understand its real-life impact. We evaluated the impact of HCL systems on sleep in paediatric patients with T1DM at Imperial College Healthcare Trust (ICHT).

Methods

In May 2024, a qualitative survey was conducted over a three-week period at ICHT paediatric diabetes annual review clinics. Participation was voluntary and based on convenience sampling during routine clinic visits. Eligible participants included children aged 18 or under with a HCL system, and parents/guardians of children in this group. The survey included open- and closed-ended questions, exploring sleep and factors affecting sleep quality.

Results

Sixteen responses were collected as part of a small, exploratory quality improvement project and conducted across two diabetes clinics - one paediatric and one adolescent - at ICHT. Participants included 9 parents/guardians and 7 children aged 4 to 18 years, with 2 children choosing not to participate. On a scale from 1 ("Very Bad") to 5 ("Very Good"), children rated their average sleep quality at 2.96, while parents rated theirs lower at 2.30. Both groups reported frequent sleep disturbances. Among children, disrupted sleep was commonly attributed to symptoms of hyperglycaemia or hypoglycaemia, restlessness, and urinary urgency. Parents most frequently cited anxiety and the demands of night-time caregiving. Seven parents described how disturbed sleep affected their child's functioning the following day. They reported consequences such as grogginess, migraines, low energy, and difficulty waking in the morning. While all parents reported consistent night-time routines involving blood glucose checks, five of the seven children reported no structured bedtime routine.

Discussion

Despite advances in closed-loop insulin delivery, sleep disturbance remains a significant challenge for children with T1DM and their families. Sleep quality should be specifically explored at clinic review. A larger study is needed to gain further understanding of the factors contributing to sleep disturbance, so that strategies can be developed to improve sleep.

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P44

The adaptation of an eating disorder prevention intervention to a wales diabetes context: application of the ADAPT guidance to clinical context

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¹Cwm Taf Morgannwg University Health Board, Bridgend, United Kingdom; ²Cardiff Metropolitan University, Cardiff, United Kingdom;³Expert by experience, Wales, United Kingdom; ⁴Expert by Occupation, Wales, United Kingdom**Introduction**

Individuals with Type 1 Diabetes (T1D) face a significantly higher risk of eating disorders, with prevalence estimates of up to 10% for clinical eating disorders and 40% for disordered eating behaviours. Despite these risks, paediatric diabetes care remains predominantly focused on prevention of physical complications, with limited integration of psychological risk reduction especially around eating disorders. Early, structured prevention strategies that incorporate psychological and educational support are necessary to address this gap.

Aim

This adaptation work applies the MRC/NIHR ADAPT framework to modify an evidence-based eating disorder prevention intervention for a Welsh population of children and young people living with T1D.

Methods

The adaptation process followed the ADAPT framework, incorporating six stakeholder workshops. The research team engaged 12 healthcare professionals (experts by occupation, EbO) and 7 individuals with T1D (experts by experience, EbE), supported by the NHS Executive Diabetes Transitional Care Coordinator. Stakeholders reviewed existing interventions, identifying the Diabetes Body Project and the recommended modifications to enhance relevance for a Welsh population.

Results

Adaptations to Diabetes Body Project EbO and EbE stakeholder workshops

Table 1

	Diabetes Body Project	Diabetes Body Project Cymru
Age of intervention Population	Aged 14-35 Females with Type 1 Diabetes	Aged 11-13 All young people with Type 1 Diabetes
Delivery	Experiencing some level of body image concerns	Embedded within routine paediatric diabetes care
Target group	Young People	Young people with supporting information for parents and training for paediatric diabetes team
Underpinning theory	Cognitive dissonance with Cognitive Behaviour Therapy (CBT)	Acceptance Commitment Therapy (ACT) with focus on increasing Psychological Flexibility through building skills around Cognitive Defusion and Values Based Action.
To Reduce	Thin ideal	Perfect ideal (including body image and diabetes)
Inclusion of Diabetes Structure Outcome measures	Introduced later in the manual 6 x 1 hour sessions online EDDI, HbA1c, TIR Demographics, DEBQ-RS, BDI, PANAS, IBIS-R, PAID, DEPS-R, Health Care Utilisation, T1DAL	Included throughout 4 x 1.5 hours face to face HbA1c, TIR Demographics, Parent Reported DEPS-R, SEEDS, CompACT-Y, PAID-T

Discussion

Further stakeholder sessions will refine outcome measures and intervention logic models. Adaptations subject to approval from the Diabetes Body Project team.

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P45

From fluctuation to foundation: a case of ABCC8 neonatal diabetes underscoring the role of genetic diagnosis in personalized managementMarwa Bebars Mohamed^{1,2} & Heather Mitchell¹¹West Hertfordshire Teaching Hospital, Watford, United Kingdom;²Princess Alexandra Hospital, Harlow, United Kingdom

Neonatal diabetes mellitus (NDM) is a rare monogenic disorder (incidence ~1:100,000). Mutations in KCNJ11 and ABCC8 genes are common causes. Timely genetic diagnosis is crucial to differentiate NDM from Type 1 diabetes, predict clinical course, and guide management, often enabling transition from insulin to oral sulfonylurea therapy. We present a male infant, admitted at three weeks of age with vomiting and incidental hyperglycaemia (16.6 mmol/l). A paternal diabetes history was noted. Initial management involved observation and home monitoring. Despite the high initial reading, subsequent glucose levels fluctuated (5.6-13.9 mmol/l), creating an ambiguous clinical picture. This case highlights a critical diagnostic challenge: NDM can present with fluctuating hyperglycaemia not immediately warranting aggressive intervention. It underscores the necessity of maintaining a high index of suspicion and pursuing genetic testing even with ambiguous presentations. Genetic testing serves as a definitive tool when clinical presentation is unclear, providing clarity for immediate management and long-term prognosis. The paternal diabetes history, initially coincidental, proved a crucial clue supporting genetic testing. Investigations showed negative Type 1 diabetes autoantibodies and an HbA1c of 41.5 mmol/mol. Genetic testing confirmed a heterozygous pathogenic missense variant in the ABCC8 gene, diagnostic of transient neonatal diabetes. This definitive diagnosis allowed a targeted management plan focused on monitoring. The family was counselled on the condition's transient nature, high remission likelihood, and significant relapse risk in adolescence/adulthood. A clear protocol for initiating sulfonylurea therapy upon relapse was established. The infant is currently thriving under regular follow-up. This case demonstrates that NDM can present with subtle, fluctuating glycaemic patterns, posing a diagnostic pitfall. It champions early genetic testing as the definitive tool to reveal the underlying

aetiology. This approach is paramount for accurate diagnosis, predicting disease trajectory, and implementing personalized, effective long-term management.

Key Learning Points:

- NDM may present with variable glucose levels that don't consistently meet diabetic thresholds.
- Family history of diabetes should prompt consideration of monogenic diabetes.
- Early genetic testing is crucial for definitive diagnosis and management planning.
- Sulfonylurea therapy is the preferred treatment for K-ATP channel mutations.

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P46

Evaluating inequalities in access to diabetes related technology at nottingham university hospitals (NUH)

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Background

The National Paediatric Diabetes Audit (NPDA) continues to identify inequalities in diabetes care. Children and young people with type 1 diabetes mellitus (T1D) from deprived areas are likely to have a higher HbA1c and a lower rate of access to diabetes technologies. We aimed to assess the access to technology of our clinic population from deprived areas compared to the overall clinic and national NPDA data.

Method

Index of Multiple Deprivation (IMD) deciles were derived, based on recorded postcode, for each patient with T1D under the care of Nottingham Children's Hospital. As of September 2024, the use of technology (continuous glucose monitoring, pump/hybrid-closed loop), average HbA1c and demographic data were ascertained for patients from IMD decile group 1 (IMD1).

Results

Within IMD1, there were 62 patients, of which 33 were male (53.2%). The average HbA1c was 56.8 mmol/mol, similar to 56.4 mmol/mol in the clinic as a whole. Although most patients in this cohort identified as white, patients identifying as other ethnic backgrounds were over-represented. All 62 patients had been offered pump/HCL therapy. Of the 20 patients not on a pump/HCL, 8 were awaiting to start and 4 had recently attended a pump showcase. The average HbA1c was 56.5 mmol/mol for those on a pump and 57.7 mmol/mol for those not. 5 of 8 patients who had declined pump/HCL therapy were white, male and older adolescents. 2 had been admitted to the ward previously for stabilisation.

Conclusions

Nationally, patients in IMD1 are at risk of reduced access to diabetes technology, which can result in compounding social, economic, and health disparities. Local access rates (68%) are comparable to NPDA 2023-24 rates for the two most deprived deciles (66.8%), and their outcomes are similar to our overall clinic outcomes. In contrast to the national NPDA data, the average HbA1c was similar for those on a pump and those not on a pump within IMD1. Older male patients are at risk of continuing to be disadvantaged and require particular targeted interventions to optimise uptake of technology.

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P47

Family perception of the process and challenges for their child/young person returning to school following a diagnosis of type 1 diabetes mellitus

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Background

The return to school signifies an important milestone in returning to usual routine for children and young people with a new diagnosis of Type 1 Diabetes Mellitus (T1DM). We aimed to identify the challenges faced by families to further guide how we facilitate this process.

Methods

We conducted a retrospective audit and service evaluation of the family perception of the return to school process for 48 children and young people, diagnosed with T1DM between January 2023-December 2024 at the Royal London Children's Hospital. Data was collected from the Clinical Records System, Twinkle and by service evaluation questionnaire sent to the families identified.

Results

Service evaluation responses were received from 22 families (46%). Return to school in full time capacity was achieved in under 2 weeks (24%), between 2-4 weeks (32%), 4-8 weeks (32%) and >8 weeks (14%) from hospital discharge, with a part time return to nursery/school occurring earlier for some. There was no significant difference in age of child at diagnosis in association with time to return. School care planning meetings and in person school training were facilitated by the diabetes team, with 87% families clearly reporting an individual health care plan in place prior to school return. Return to school delays were perceived by families in relation to school readiness (55%); with families reporting school staff appeared very confident (27%), somewhat confident (59%) and not confident (14%). 41 % of families had opportunity for school staff to shadow their management of their child. Parent readiness (32%) and child/young person readiness (18%) and the young person's physical health (9%) were also cited in relation to school return delays. Families reported 41% of their children were concerned about being different to their peers, 36% were worried about school, 36% had difficulties managing their glucoses, 27% reported disturbed sleep with 32% reporting no problems with the above. Clear communication and support from all professionals were deemed most helpful.

Conclusion

Close working between the families, school and diabetes teams is critical to facilitate a smooth school return and family responses provide important insights to help further plan the process.

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P48

Short term impact of a paediatric diabetes camp on glycaemic control- a service evaluation

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Introduction

The University Hospital Dorset paediatric diabetes service runs a diabetes camp for children and young people (CYP) with type 1 diabetes (T1D). Peer support, sharing experiences and reducing isolation of living with diabetes are among the primary goals of the camp. CYP with T1D face specific challenges in maintaining glycaemic control and developing self-management skills. Diabetes camps provide supervised social settings that could promote effective self-management of T1D. We aim to evaluate the short-term impact of diabetes camp on glycaemic control of children with T1D.

Method

Data was collected retrospectively as part of the routine service evaluation using continuous glucose monitoring data from 37 individuals with a diagnosis of T1D between the ages of 8 and 16. Change in Time in Range (TIR) two weeks prior and 2 weeks from the start of 3-day residential camp was the primary outcome. Time Above Range (TAR), Time Below Range (TBR), and average glucose levels were also collected. Paired t-tests were utilized for inferential analysis, while histograms and Q-Q plots were used to examine the data for normality in Microsoft excel.

Results

The mean TIR before and after the camp was 54% and 53.13%, respectively. A paired t-test was used, and the resulting p-value was 0.53. This implies that TIR did not significantly change before or after. However, the post-camp TIR increased for 19 individuals and decreased for 16. For two participants, the post-camp TIR stayed constant.

Discussion

These results suggest that although diabetes camps may offer peer support and psychosocial benefits, in this case there were no short-term improvement in glycaemic control. This diabetes camp did not include any structured diabetes aspect. Further short and long-term data on wellbeing and diabetes distress in relation to diabetes camps in children with T1D would allow further delineation of their potential benefits.

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Miscellaneous/Other 1

P49

Hypercalcaemia: a rare cause to considerAndrew Tester¹, M.Guftar Shaikh¹ & Lana McMillan²¹Royal Hospital for Children, Glasgow, United Kingdom; ²University Hospital Wishaw, Lanarkshire, United Kingdom

Introduction

Hypercalcaemia is uncommon in children; it may appear with classic signs/symptoms or be identified incidentally during investigations for other reasons. Causes of hypercalcaemia are diverse and include malignancy. If left untreated, hypercalcaemia causes significant morbidity and mortality. Rarely, hypercalcaemia is due to primary hyperparathyroidism, of which the main cause is a parathyroid adenoma. This should be considered in the differential of paediatric hypercalcaemia, especially in adolescence. Early diagnosis and treatment of a parathyroid adenoma removes the significant morbidity burden.

Presentation

A 15 year old girl presented to her GP with vague symptoms of lethargy, muscle aches, palpitations and general malaise. Blood results indicated hypercalcaemia (Ca²⁺ 3.07 mmol/l), raised PTH (10.8pmol/l) and high 1,25 Vitamin D (205pmol/l), suggestive of hyperparathyroidism. She was initially treated with hyperhydration of intravenous fluids and later pamidronate. Nuclear medicine imaging identified a rounded focus of uptake in the left inferior parathyroid gland, suggestive of a parathyroid adenoma. Renal ultrasound scan identified no nephrocalcinosis. She underwent excision of the left inferior parathyroid gland; histopathology of the lesion identified a 400 mg, encapsulated nodule with clear borders on the tissue specimen. Three months after excision of this lesion, her bone profile was entirely normal. Genetic bloods were negative for the hyperparathyroidism/familial hypercalcaemia hypocalciuric panel.

Discussion

Primary hyperparathyroidism is a rare but a significant cause of morbidity in children, more so than in the adult population. The commonest cause of primary hyperparathyroidism in children is a parathyroid adenoma. These are usually sporadic and most often occur singularly, occasionally occurring as part of a genetic syndrome, such as multiple endocrine neoplasia (MEN). The biochemical features seen are an inappropriately high PTH in the context of hypercalcaemia. Once a parathyroid adenoma is located by scintigraphy, parathyroidectomy should be considered, usually resulting in resolution of biochemical features and symptomatology. Parathyroid adenoma should be considered in children, especially beyond the first decade of life, with hypercalcaemia and high PTH and investigated accordingly.

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P50

A single centre experience in paediatric cases of pheochromocytomas and paragangliomasDhanya Soodhana^{1,2}, Alina Oprea¹, Louise Izatt³ & Christina Wei¹¹Evelina Children's Hospital, London, United Kingdom; ²Aster MIMS, Kozhikode, India; ³Guy's and St. Thomas' Hospital, London, United Kingdom

Background

Phaeochromocytomas and paragangliomas (PPGL) are rare neuroendocrine tumours arising from the adrenal medulla or extra-adrenal sympathetic and parasympathetic paraganglia. Paediatric cases account for only 10–20% of all detected cases of PPGL. Approximately 70–80% of PPGL in children are caused by an inherited pathogenic variant in one of at least 25 tumour susceptibility genes. Thus, germline genetic testing has high priority in the diagnostic work-up and guides personalized diagnostics, management, therapeutic and surveillance strategies in children and adolescents with PPGL.

Methods

Medical records were retrospectively analysed for paediatric patients followed in the paediatric neuroendocrine clinic at the Evelina Children's Hospital, London between 2013 and 2025.

Results

We describe a case series of 6 paediatric patients (5/6 male) diagnosed with PCC ($n = 4$) and PPGL ($n = 2$) at median age of 10.74 (09.6–16.4) years, through histopathological confirmation. Routine PPGL surveillance detected 2/6 with known PPGL genetic susceptibility. The majority had symptomatic presentation: 3/6 hypertension, 1/6 transient weakness of the tongue and hearing loss. Histopathology report incidentally confirmed the diagnosis in 1/6 who underwent

nephrectomy for unilateral kidney dysplasia due to chronic hypertension without undergoing endocrine work-up. 4/6 had elevated plasma normetadrenaline levels and 1/6 had elevated plasma methoxytyramine levels. 4/6 had unilateral non-malignant PCC; 1/6 developed a second PCC in the other adrenal gland 1.6 years after the first. 5/6 had germline mutations (3/6 SDHB; 2/6 VHL). 1/6 had somatic VHL mutation on tumour analysis. 2/6 were diagnosed as a part of the cascade screening and in 1/6 the screening of the relatives led to the mother being diagnosed with the same mutation.

Conclusion

In paediatric patients with confirmed hereditary PPGL, lifelong follow-up is essential to screen for both metastatic disease and metachronous tumours or syndrome-related pathologies. The long-term surveillance protocols should be tailored to the specific pathogenic variant. Cascade genetic testing in the wider family is required, as 4 of these paediatric cases were the first PPGL presentation in their family. In all cases the pathogenic variant was confirmed as being inherited from a parent. The penetrance of PPGL in non-index cases is gene-dependent.

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P51

A national survey of paediatric turner syndrome services in the united kingdom: current practice and variability in careJames M. Law¹, Nadia Amin², Elspeth C. Ferguson³, Jan Idkowiak⁴, Sasha R. Howard⁵, Harshini Katugampola⁶ & Nils Krone⁷¹Nottingham University Hospitals NHS Trust, Nottingham, United Kingdom; ²Leeds Teaching Hospitals NHS Trust, Leeds, United Kingdom;³Sheffield Children's NHS Foundation Trust, Sheffield, United Kingdom;⁴Birmingham Women & Childrens NHS Foundation Trust, Birmingham, United Kingdom; ⁵Queen Mary, University of London and Barts Health NHS Trust, London, United Kingdom; ⁶Great Ormond Street Hospital, London, United Kingdom; ⁷University of Sheffield, Sheffield, United Kingdom

Background

Turner syndrome (TS) is a complex condition requiring lifelong multidisciplinary care. International guidelines recommend structured clinic models and coordinated transition, yet service delivery across the UK has not previously been evaluated at a national level.

Objectives

To assess current UK paediatric TS service provision, including variation in clinic configuration, multidisciplinary team (MDT) composition, adherence to consensus guidelines, and use of clinical pathways and registries.

Methods

An electronic survey was distributed to all UK tertiary paediatric endocrine centres and completed between June 2023 and February 2024. The survey captured data on consultant staffing, patient numbers, clinic structure (dedicated vs general endocrine), MDT membership, follow-up frequency by age group, local versus regional service differences, transition pathways, and awareness and use of the International TS Consensus Guidelines, the i-TS Registry, and Turner Syndrome Support Society resources.

Results

Twenty centres responded. Six of 20 had a dedicated TS clinic; these tended to have more patients and greater consultant capacity. Most MDTs were largely limited to paediatric endocrinologists and clinical nurse specialists, with infrequent access to psychology, gynaecology, and dietetics. Centres without dedicated clinics often cited limited staffing or low patient numbers. Outreach patients living away from the tertiary centre may be seen less often and with reduced specialist input. Most centres saw patients twice a year but there was variation, with some increasing frequency during late childhood and adolescence. Transition models varied: 9 centres had a TS-specific pathway, 9 used a general endocrine pathway and 2 reported no transition pathway. All centres were aware of the international TS Consensus Guidelines and routinely informed families of the Turner Syndrome Support Society. Most centres were aware of the i-TS Registry: 5 were already participating and 11 have plans to.

Conclusion

This national survey highlights the variation in paediatric TS care across the UK. Smaller centres are less likely to have dedicated clinics and more limited consultant and MDT resources. Transition services are inconsistent, and registry participation is limited. While awareness of best practice is widespread, further support is needed to ensure equitable care and improve service alignment with international standards.

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P52

Taming the cortisol storm: a paediatric case series on cushing's syndrome diagnostic challenges and managementHabab Easa¹, Nida Aslam² & Taffy Makaya³¹Paediatric Registrar, Oxford University Hospital, Oxford, United Kingdom; ²Clinical Observer in Paediatric Endocrinology, Oxford University Hospital, Oxford, United Kingdom; ³Paediatric Diabetes and Endocrinology Consultant, Oxford University Hospital, Oxford, United Kingdom

Introduction

We present a case series of 3 patients managed in our centre over the last 2 years, with endogenous Cushing syndrome (CS) from three distinct causes. This series provides a great learning opportunity for a rare childhood phenomenon.

Case 1

A 10-year-old girl presented with a history of significant weight gain and osmotic symptoms. Examination confirmed cushingoid features and hypertension (HTN). Tests confirmed diabetes, elevated 24-hour urinary cortisol while the ACTH suppressed on Dexamethasone suppression test (DST), consistent with ACTH-independent CS. Imaging showed bulky adrenal glands without focal lesions, and genetic testing confirmed PRKAR1A mutation consistent with primary pigmented nodular adrenocortical disease (PPNAD). She was treated with metyrapone and anticoagulants, followed by bilateral adrenalectomy. Echocardiography excluded cardiac myxomas. Postoperatively, she has lost 20 kg of weight, and HTN and diabetes have resolved. She is stable on hydrocortisone (HC) and fludrocortisone (FCN) therapy.

Case 2

A 14-year-old girl presented with growth and puberty arrest and increasing fatigue. Clinically, she looked Cushingoid. Biochemistry showed elevated urine cortisol and a lack of ACTH suppression on DST, consistent with ACTH-dependent Cushing's disease. MRI pituitary was inconclusive. She underwent inferior petrosal sinus sampling (IPSS) for localisation, which was also inconclusive. Management included metyrapone and anticoagulation, followed by exploratory pituitary surgery, which identified a lesion which was removed. She is now stable on HC replacement.

Case 3

A 15-year-old male presented with malaise, anxiety, and rapid weight gain. Examination revealed HTN, hyperglycaemia, and an abdominal mass. Tests showed elevated ACTH and cortisol levels. Imaging revealed widespread metastatic lesions, consistent with ACTH-producing neuroendocrine tumour, likely originating from the right lung. He was treated with chemotherapy and metyrapone; insulin and antihypertensives were briefly required. Denosumab was used for the metastatic bone lesions. Unfortunately, his condition progressed, and he sadly passed away one year later.

Conclusion

Childhood CS is rare. Weight gain and growth retardation are hallmark features of CS. Evaluation typically includes 24-hour urinary cortisol. DST is considered the gold standard for distinguishing between ACTH-dependent and independent CS. Whole-body imaging helps identify potential ACTH-secreting neuroendocrine tumours. Management is guided by the underlying aetiology, with metyrapone a beneficial adjuvant therapy.

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P53

Uptake and patient perception of tumour surveillance in a paediatric population with SDHx variants in a single-centre family clinicEmma Alden^{1,2}, Elizabeth Nash², Emily Connolly², Lee Martin², Eugene Lim³, Scott Akker³ & Rathi Prasad²¹Queen Mary University of London, London, United Kingdom; ²Royal London Children's Hospital, Barts Health NHS Trust, London, United Kingdom; ³St Bartholomew's Hospital, Barts Health NHS Trust, London, United Kingdom

Background

Phaeochromocytomas and paragangliomas have an annual incidence of 0.4-2 cases per million children. Inherited pathogenic variants in tumour suppressor genes are implicated in 70-80% of cases. Lifelong screening is recommended for asymptomatic variant carriers from childhood/early adulthood. We aimed to evaluate uptake of paediatric surveillance at Barts Health NHS Trust and gain insights into family perception of screening.

Methods

A retrospective audit analysed uptake and outcomes of annual surveillance offered to the paediatric cohort at Barts Health NHS Trust from January 2019-December 2024. A service evaluation questionnaire was sent to individuals aged 10-20 years and their parents.

Results

The audit comprised a population of 55 young people aged 5-19 years (from 32 families). Of these, 87% had heterozygous *SDHx* variants. This cohort demonstrated transmission ratio distortion for *SDHx* variants, with a transmission ratio of 0.76 (95% confidence interval: 0.64-0.85), higher than the expected 0.5. Annually, an average of 89% attended clinical reviews (face-to-face/telephone), 79% had metanephrine testing, and 89% underwent imaging. Two young people with *SDHx* variants had lesions identified on first screening. One was diagnosed with a retroperitoneal paraganglioma at 8.7 years, and the other with a phaeochromocytoma at 16.9 years. Both were managed with alpha-blocked resection, with no recurrences/metastases on six-monthly surveillance thereafter. Over this period, 27 of 202 scans conducted showed unrelated incidental findings (22 individuals), with 10 individuals undergoing further investigations and 1 requiring treatment. Service evaluation responses were received from 12 families (37.5%). Young people showed good awareness of the purpose of screening. They reported low levels of associated health anxiety (mean score 2.3/10). Parents reported higher levels of anxiety about their child's health (mean score 5.3/10). Parents rated their child's worry regarding different screening components higher than the young people scored themselves. Overall, satisfaction with screening was high for young people (mean 7.1/10) and parents (mean 8.3/10). Areas identified for development include the provision of clearer explanatory materials, reducing the interval between screening and results appointments, and increasing psychological support.

Conclusion

Within our cohort, annual screening was well-tolerated and demonstrated high compliance. Findings also emphasised the value of early surveillance.

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P54

To induce or not to induce – that is the question. delayed puberty in a male with anorexia and ARFID – a case report and literature reviewCatriona McKay¹, Noina Abid¹, Linda Agnew² & Katherine Murtagh¹¹Royal Belfast Hospital for Sick Children, Belfast, United Kingdom;²Belfast Health and Social Care Trust, Belfast, United Kingdom

A 14 year and 11 month old male, with anorexia nervosa, ARFID and a background of autism, rectal prolapse and bilateral scrotal fixation, presented to endocrine clinic with delayed puberty. His auxology, staging and bloods are shown (Table 1&2). His investigations demonstrate he was clinically and biochemically pre-pubertal with delayed bone age. Due to an absence of evidence for pubertal induction with testosterone in anorexic males, priority focused on weight restoration. He was admitted for medical stabilisation prior to transfer to a mental health unit. He has spent over 6 months there and approaches weight restoration. There are no physical signs of puberty. It is hoped that if he maintains his weight these will ensue. This case is felt to be unusual - it is absence rather than arrest of puberty due to enduring anorexia and ARFID. ARFID is increasingly prevalent and is relevant to endocrinologists due to its effects on growth and puberty. Consideration was given to evidence for the use of testosterone in this cohort, which is limited, this has been identified as an area for further research.

1. Auxology & pubertal staging

Date and location	Age (Years and months)	Height (Centimetres)	Weight (Kilograms)	BMI (kilograms per metres squared)	Weight for Height (% of ideal BMI)	Pubertal staging
First medical admission due to low weight 03/07/20	10y7m	144	26.8	12.9	76.3	Not done
Endocrine clinic 26/11/24	14y11m	147	27.3	12.6	65.3	G1P1A1 TV < 3mls
Medical admission 13/01/25	15y1m	147	26.7	12.4	64.2	G1P1A1 TV < 3mls
Discharge to IP MH facility 18/02/25	15y2m	147	29.8	13.8	71.5	G1P1A1 TV < 3mls
Latest measurements 07/07/25	15y7m	150	42.4	18.8	94.9	Staging pending Nil changes reported by patient/ staff

1. Laboratory and imaging results

Date	Test (units)	Reference range	Result
26/11/24	IGF-1 (nmol/l)	15.7 - 65.6	2.7
01/05/25			20.7
26/11/24	LH (IU/l)	1.7 - 8.6	<0.3
01/05/25			1.7
26/11/24	FSH (IU/l)	1.5 - 12.4	<0.3
01/05/25			3.6
26/11/24	Testosterone (nmol/l)	Tanner stage 2 <15	0.4
01/05/25			<0.2
26/11/24	GH (ng/ml)	N/a	3.2
26/11/24	Greulich & Pyle Bone Age	Within two standard deviations of chronological age considered normal.	12.4 years - 2.22SD < mean for age

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P55**Hypoparathyroidism as first manifestation of kearns-sayre syndrome (KSS)**

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Background

Kearns-Sayre syndrome is a rare mitochondrial disorder which can manifest as numerous endocrinological abnormalities. The case we are presenting shows the importance of consideration of such in children presenting with primary hypoparathyroidism with other multi-system involvement.

Case presentation

An 8 year old boy presented with a longstanding history of lethargy, weakness, poor appetite and growth concerns. He was seen in paediatric clinic at the age of 6 in view of faltering growth with normal investigations at the time. On presentation his height was 116 cm (1st centile), weight 18 kg (0.12 centile) with mid-parental height at 60th centile. He had bilateral ptosis, strabismus, and injected conjunctiva. Neurological examination was significant for ataxia and dysmetria. Investigations revealed high lactate, hypocalcaemia, hypomagnesaemia and hypokalaemia with normal glucose and infection markers. Further investigations showed low PTH which confirmed the diagnosis of primary hypoparathyroidism, as well as renal tubulopathy. He had normal TFT's, Hb1AC, random cortisol and aldosterone levels. Ophthalmology review was significant for pigmented retinopathy and external ophthalmoplegia. He was found to have moderate sensorineural hearing loss. He had normal metabolic and autoimmune screen, but raised protein and lactate on CSF. MRI brain was suggestive of metabolic encephalopathy. Mitochondrial disorder was suspected and genetics test showed large scale mtDNA deletion which had confirmed the diagnosis of KSS. His management involved regular follow up in endocrinology, cardiology and renal clinics, with OT/physio input, and regular monitoring of PTH, TFT and adrenal axis, as well as supplementation with calcium, vitamin D, magnesium and co-enzyme-10.

Conclusion

KSS is a rare mitochondrial multisystem disorder defined by the triad of onset before age 20, pigmentary retinopathy and progressive external ophthalmoplegia with at least one of the following: cardiac conduction block, high CSF protein levels, or cerebellar ataxia. Other features include endocrine involvement, hearing loss, dementia, and myopathy. Endocrine involvement includes short stature, GH deficiency, DM, hypothyroidism, adrenal insufficiency or primary hypoparathyroidism as presented here. Management is supportive with hormonal replacement therapy as required, cardiac pacemaker in case of conduction abnormalities, corrective surgery for eye abnormalities, cochlear implants and co-enzyme 10 (ubiquinone). Prognosis is guarded.

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P56**Immunotherapy related neurotoxicity presenting as late-onset hypophysitis in an adolescent female with B-cell acute lymphoblastic leukaemia**Sushil Yewale¹, Ramya Gokul¹, David Cullingford¹, Poonam Dharmaraj¹, Mohammed Didi², Christopher Howell², Julie Park¹, Renuka Ramakrishnan¹ & Joanne Blair¹

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Introduction

We present a rare case of late-onset hypophysitis in a 16-year-old female with B-cell acute lymphoblastic leukaemia (ALL), which developed following treatment with Blinatumomab, an immune checkpoint inhibitor.

Case presentation

A 16-years-8-month-old girl diagnosed with B-cell ALL at 14-years-9-months was referred for polyuria exceeding 4 ml.kg⁻¹.h⁻¹. Initial lumbar puncture confirmed no central nervous system involvement. Her chemotherapy regimen included dexamethasone, peg-asparaginase, methotrexate, mercaptopurine, vincristine, imatinib, and two cycles of intravenous blinatumomab. Her induction chemotherapy was complicated by steroid-induced diabetes, febrile neutropenia with septic shock, perineal abscess requiring debridement and sigmoid colostomy, multiple transfusions, and acute-on-chronic pancreatitis necessitating drainage, stenting, and enzyme supplementation. Upon referral, her height was 164.1 cm (+0.2 SDS), weight 44.2 kg (-1.23 SDS), and BMI 16.4 kg.m⁻² (-1.94 SDS), reflecting significant weight loss since diagnosis. Investigations revealed arginine vasopressin deficiency with hyponatremia (sodium 149 mmol.L⁻¹) (NR 135-145), elevated plasma osmolality (299 mosm.kg⁻¹) (275-296), decreased urine osmolality (76 mosm.kg⁻¹) (200-800), low co-peptin (3.6 pmol.L⁻¹) (>5), and partial response to DDAVP during water deprivation test. Initially, remainder hypothalamic-pituitary axis screening was unremarkable: TSH 0.48 mu.L⁻¹ (0.3-3.8), free T4 10.3 pmol.L⁻¹ (9-19), cortisol 472 nmol.L⁻¹ (140-500). Neuroimaging revealed absent posterior pituitary bright spot and thickened pituitary stalk; no mass lesion. After commencing oral desmopressin, she developed persistent hypotension, hyponatremia (sodium 132 mmol.L⁻¹) (135-145), lethargy, and vomiting over the next month. A subsequent lumbar puncture remained normal. Repeat endocrine evaluation indicated secondary adrenal insufficiency (09.00 cortisol 98 nmol.L⁻¹) (140-500); (ACTH <1.1 pmol.L⁻¹) (2-11), central hypothyroidism (free T4 6.6 pmol.L⁻¹, inappropriate normal TSH 0.68 mu.L⁻¹), low LH (<0.1 IU.L⁻¹) (3-8) and FSH (0.2 IU.L⁻¹) (2-6), raised prolactin (1717 mU.L⁻¹) (0-500), and low IGF-1 (5.1 pmol.L⁻¹) (24.7-55.8), consistent with panhypopituitarism, presenting 19 months post-blinatumomab. Workup for granulomatous hypophysitis and Langerhans histiocytosis was negative. She showed prompt clinical response to hydrocortisone and levothyroxine and remains under close follow-up.

Discussion

Hypophysitis secondary to immune checkpoint inhibitors is well described with ipilimumab and nivolumab but not reported with blinatumomab. Likely driven by T-cell-mediated pituitary destruction and complement activation, it is typically irreversible and steroid-unresponsive. With other causes excluded, blinatumomab-induced hypophysitis was considered causative.

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P57**Review of nottingham university hospitals' clinical practice for the screening of endocrinopathies in children receiving blood transfusions for sickle cell disease/rare anaemias**

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Introduction

Endocrine issues in Sickle Cell Disease (SCD): dysglycaemia, growth failure, hypogonadism, thyroid, and adrenal insufficiency (1) are often worsened by iron overload from transfusions (2). Data on their prevalence in children are limited, and screening practices vary (3,4). Improving screening can lead to early detection and treatment. We aimed to audit adherence to guidance in our unit and assess dysglycaemia rates in transfused CYP with SCD.

Methods

We reviewed electronic records from October 2021 to October 2024, following 2021 BSH guidelines (4). Recommendations included pubertal assessment from age 10; annual OGTT from puberty or age 10 if family history; yearly vitamin D, cortisol, thyroid, and pubertal assessment; biannual height/weight; and biennial DEXA scans.

Inclusion

Diagnosed with SCD or rare anaemia, on regular transfusions, aged at diagnosis and treatment start.

Results

Our cohort included 8 patients; median age at diagnosis was 10 years, treatment started at 20 years, with a median treatment duration of 10 years. Gender was evenly split; ethnicity included Pakistani (n,2 (25%)), Black Caribbean (n,3 (38%)), Black African (n,2 (25%)), and others (n,1 (12%)). All received chelation. Screening adherence varied: vitamin D (100%), thyroid tests (60-100%), OGTT improved from 25% to over 60% over the years, while height measurements were lowest (25-50%). adherence was for height measurements, with compliance rates

between 25% and 50%. Data for OGTT: Median age at first OGTT was 15 years (range 12–24) with Median follow-up duration was 5.5 years (range 2–9). The median number of OGTTs per patient was 3.5 (range 2–7). During follow-up, 2 patients showed impaired fasting glucose, which resolved later. One patient initially had impaired glucose tolerance, which also resolved.

Conclusion

Screening adherence was inconsistent, particularly for height. Dysglycaemia was rare and transient. A larger multicentre study is needed to better understand endocrine issues in SCD.

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Obesity 1

P58

Social prescribing and complications of excess weight (CEW) clinics
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In England, Tier 3 weight management services known as Complications of Excess Weight (CEW) clinics have been formed to support the high numbers of children living with severe obesity and facing significant physical and mental health complications. Social prescribing helps connect families to local community services which can provide additional, non-medical support. This could include referral to physical activity groups and to information services, such as Citizen's Advice. Whilst social prescribing is commonly used in adult services it has not yet been formally used in children's weight management. Based on research showing that families accessing CEW are living in the most deprived areas of the country and experience complex needs, including low self-esteem, confidence and social isolation, social prescribing may be a helpful way to provide additional holistic support to patients and their families. This research used a realist approach, which aims to understand how social prescribing may (or may not) work in CEW clinics. 20 interviews were conducted with staff from a CEW clinic, linked community-based services and CEW patients to explore theories of how social prescribing may work in this setting. Social prescribing was noted to be helpful in providing peer support opportunities for CEW families. Referral to information services was reported to help address wider social barriers, meaning that clinical staff could focus on weight and comorbidity management. CEW clinicians commonly referred young people to physical activity groups but faced challenges in finding accessible activities. Mapping work demonstrated that of 933 community-based services local to one CEW clinic, only 19 met the specific needs of children living with severe obesity. This highlights a need for more consistent investment towards community services to offer more accessible, sustainable opportunities for children with complex health and social needs. Engagement with social prescribing was supported by a family support worker in the CEW team, who met with families through home visits and spent time building trust with patients. This person-centred approach was reported to be important for improving families' mental health and quality of life outcomes; however future research may be needed to explore the long-term impacts of this.

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P59

"I brush my teeth once a month": a service evaluation of dental team led oral health advice alongside complications from excess weight (CEW) clinics

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Introduction

Oral health (OH) and type 2 diabetes (T2D) are key areas in CORE20PLUS5. Sugar consumption is a common risk factor for dental decay, obesity and T2D. Complications from Excess Weight (CEW) teams have observed many children and young people (CYP) are living in the most deprived quintiles, are high risk for decay, do not access dental care, and live with neurodiversity. A collaboration between CEW and dental teams is facilitating access to urgent/routine dental care

via a bespoke pathway and provision of OH advice with a one stop shop. A service evaluation was undertaken to explore anecdotal findings and inform service development.

Methods

A questionnaire was developed to explore CYP's oral hygiene and dietary practices, access to dental care, and dental problems. An OH team supported one CEW clinic per month for 12 months. Families received tailored OH advice from the OH team after their CEW appointment and were invited to complete the questionnaire. A dental student outreach programme was implemented concurrently, to support these sessions, and student learning about the interplay of oral and systemic disease and social determinants of health.

Results

Thirty-three questionnaires were completed between January – December 2024. Two CYP (6%) had never visited a dental team and seven (21%) reported current dental problems. Only 17 CYP (52%) reported brushing teeth twice daily and six (18%) never brushing or brushing less than once a week. Chocolate and sweets ($n = 16$; 49%), water ($n = 16$; 48%) and squash ($n = 14$; 42%) were the most frequently consumed snacks and beverages. Twenty-two families (67%) reported what they had learned, which included upskilling brushing technique, importance of fluoride toothpaste and awareness of sugar content in food/drinks and impact on oral health. Students attending reported positive learning experiences.

Conclusion

Findings suggest CYP attending CEW are at high risk of dental disease and families were receptive to OH advice. The frequency of OH support is increasing and resources to support OH for CYP with neurodiversity are under development. This inter-professional collaboration aligns with the NHS 10 Year Plan by facilitating access to dental care and supporting CYP with both their weight and OH.

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P60

Glucagon-like peptide-1 receptor agonists in paediatric hypothalamic obesity-single center experience

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Background

Hypothalamic obesity (HO) is a rare but severe complication arising from hypothalamic injury, characterised by dysregulation of neuroendocrine circuits governing energy homeostasis, satiety, and metabolic rate. It is refractory to conventional lifestyle and pharmacological interventions, particularly in paediatric cohorts, for whom no regulatory-approved pharmacotherapy exists. Recent studies have suggested that glucagon-like peptide-1 receptor agonists (GLP-1RAs) may modulate central appetite pathways and facilitate weight management; however, empirical evidence in paediatric HO remains limited.

Objective

To describe a single-centre observational experience utilising subcutaneous Semaglutide in four paediatric patients with syndromic or acquired forms of hypothalamic obesity, assessing anthropometric responses over a 12-month treatment period.

Methods

Four female patients (ages 12.7–18.9 years) with clinically and radiologically confirmed HO were commenced on Semaglutide therapy, titrated from 0.25–0.5 mg to a maximum of 2.4 mg weekly over 3–9 months. Aetiologies included septo-optic dysplasia, ROBO1-associated pituitary stalk interruption syndrome, non-metastatic intracranial germinoma, and Moebius syndrome with structural hypothalamic involvement. Longitudinal changes in body mass index (BMI) and BMI standard deviation score (SDS) were recorded at baseline, 6, and 12 months.

Results

The cohort's mean baseline BMI was $50.3 \pm 8.03 \text{ kg/m}^2$, with a mean BMI SDS of 4.21 ± 0.5 . Two patients demonstrated clinically meaningful reductions in BMI and BMI SDS (maximum decrease: 16.8% weight loss with 0.62 BMI SDS reduction at 12 months). One patient experienced a relative attenuation of pre-treatment weight velocity, while another exhibited weight gain initially, followed by stabilisation. Individual variability in response was observed across both dosing trajectories and phenotypic context. No participants experienced severe adverse effects or treatment discontinuation.

Conclusion

This real-world series provides preliminary evidence that Semaglutide may facilitate BMI reduction or stabilisation in paediatric patients with HO, a notoriously treatment-resistant condition. Observed heterogeneity in response

underscores the need for mechanistic studies investigating differential drug efficacy in relation to hypothalamic integrity, genetic background, and neuroendocrine phenotype. Further prospective randomised controlled trials are warranted to establish efficacy, durability, and safety of GLP-1RAs in the paediatric HO population.

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P61

Survey on impact of childhood obesity on child health services in Wessex

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Background

One in 3 children in England leave primary school living with overweight/ obesity and this is associated with increased risk of a number of comorbidities. We conducted a survey of health professionals treating children in secondary or tertiary care within the Wessex Deanery to better understand the impact of managing children living with obesity on their services.

Method

An electronic survey was emailed to general paediatric consultants, tertiary paediatric specialty consultant, community paediatric consultants, anaesthetists, surgeons and specialist nurses working in 9 different hospitals in Wessex asking for anonymised feedback regarding the impact of childhood obesity on care provided in their service. Thematic analysis of free text answers was conducted by a consultant clinic psychologist.

Results

We received 102 responses; 72.3% from consultants/ specialty doctors, 18.8% from specialty nurses and 8.9% from other professional groups. Responders were from 9 different hospitals. Only 50.5% of responders said they routinely discuss weight/ signpost to resources if children noted to be living with obesity present to their service. The most commonly given reason for this was lack of confidence/ knowledge on how to do so (56.9%) followed by concern regarding causing offence/ damaging patient relationship (43.1%). 46.7% of general paediatricians who responded said they did not have any service to assess or treat children living with obesity as recommended by NICE guidance. 100% of the general paediatricians stated current provision did not meet local need. Thematic analysis of free text responses looking at healthcare professionals perspective on the impact of obesity in paediatric patients identified key themes including clinical and procedural challenges, psychosocial and mental health effects, and need for increased time and resources. The need to address social determinants of obesity and the need for multidisciplinary support was also highlighted.

Discussion

Childhood obesity has a significant impact on child health services, often making care more complex or time consuming. Additional training needs to be offered to clinicians looking after children to overcome gaps in knowledge or confidence addressing this issue. Commissioning of additional services to manage children living with additional weight is also urgently needed.

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P62

Efficacy of tirzepatide in an adolescent with alström syndrome

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Alström syndrome (ALMS) is a rare, autosomal recessive condition caused by variants in the *ALMS1* gene. It is a multisystem, progressive condition characterised by vision (retinal dystrophy) and hearing loss, obesity, insulin

resistance, type 2 diabetes mellitus, cardiomyopathy and renal dysfunction requiring specialist multidisciplinary management. Treatment of obesity in ALMS is challenging, with limited efficacy of glucagon-like peptide (GLP-1) receptor agonists and inconclusive results for the MC4R agonist, Setmelanotide. We present a case of an adolescent living with ALMS who has shown a successful response to Tirzepatide, a dual agonist for GLP-1 and Gastric Inhibitory Polypeptide (GIP) receptors. This male patient was born at term weighing 4.4 kg and developed early-onset, hyperphagic obesity. He was diagnosed with retinal dystrophy aged one year and moved to the United Kingdom aged 9 years, whilst being referred to a weight management service at 11 years. At that time his weight was 86.6 kg, height 162.5 cm, BMI 32.9 kg/m² (BMI-SDS +3.32). He was hypertensive, with evidence of metabolic dysfunction associated steatotic liver disease (MASLD), and significant visual impairment due to retinal dystrophy and was diagnosed with ALMS at 12 years of age. Despite behavioural and dietetic input from the multi-professional weight management team, he continued to gain weight. He initially commenced daily GLP-1 agonist therapy (Liraglutide) at 14 years of age but experienced further weight gain. Nine months later he was switched to Semaglutide, a weekly GLP-1 receptor agonist, for seven months, but weight gain persisted with no reported appetite change. Aged 17 years he was commenced on Tirzepatide (GLP-1 and GIP dual agonist) off-licence due to his obesity associated complications, with weight 122.7 kg, BMI 37.2 kg/m² (BMI SDS +3.77). Following, 6 months of treatment (5 mg weekly), he has lost 13.7 kg (11.2% weight loss, BMI-SDS reduction 0.84) with a corresponding drop in glycated haemoglobin (HbA1c) from 45 mmol/mol (NR <42) to 31 mmol/mol. The medication has been well tolerated with the patient reporting considerable reduction in appetite. With negligible reported benefits from Semaglutide and Setmelanotide in Alström's syndrome, these early weight and appetite changes suggest possible benefits from dual agonist treatment which warrants further investigation in this condition.

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P63

Teen cooking for health (TECH): nutrition workshops for adolescents living with obesity

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Obesity in adolescence is associated with high rates of neurodiversity, lower socioeconomic status, and poor health-related quality of life, and as a result engagement with health-related activities for this population may be challenging. We identified few practical cooking sessions locally catering for young people (YP) living with obesity. Therefore, we developed a series of free nutrition workshops called Teen Cooking for Health (TECH) incorporating feedback from YP under the care of our complications of excess weight (CEW) service. 111 YP (≥ 12 years) from our service were asked to complete questionnaires indicating if they would be interested in nutrition workshops, and what topics and format they would like for the sessions. 27 YP responded with 85% stating they were 'interested' in attending, and 15% 'maybe interested'. Popular choices of topics included snacks (78%), dinner ideas (70%), quick meals (61%), meal planning (52%), lunch ideas (48%), cooking on a budget (48%) and food shopping tips (35%) with a preference for a mix of practical cooking, YP only sessions and ingredients to take home. Three free workshops (six attendees each) have been filled, with each session to comprise of a mix of short talks, practical YP cooking sessions accommodating food and sensory preferences, information sessions for parents and recipes/ingredients to take away. Workshops are led by a nutritionist with expertise in child nutrition, supported by clinicians from the CEW service, with hospital charity funding. Support with transport costs is being provided where needed. The first session had very positive feedback with 100% (5 YP and 5 parents) reporting that they enjoyed the workshop, found it useful and relevant, felt it catered for their needs, and felt comfortable in the group. 90% stated they would 'definitely', and 10% 'maybe', like to attend similar workshops in the future. YP engaged positively in the group and reported they particularly liked the practical aspects, and parents highlighted the value in having the opportunity to talk with other parents. These TECH workshops have had a positive response and support the future use of group sessions to engage and provide relevant nutrition information for YP living with obesity.

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P64

National evaluation of 39 complications of excess weight (CEW) clinics: exploring how innovation in design has been determined by individual and population health inequities

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Background

Metabolic complications affecting adults are experienced by many CYP due to increasing prevalence of obesity from an early age. An NHS England pilot has funded specialist multi-disciplinary team (MDT) paediatric weight management services, to deliver holistic person and family centred care to CYP living with severe and complex obesity. Support is tailored to family and socioeconomic context, at individual and local population level. MDT interventions aim to improve BMI and prevent, identify early, treat and reverse associated medical and psychosocial complications. This study analyses physical and mental health outcome measures, and evaluates operational creativity through different service models, MDT composition and relational approach.

Methods

The evaluation will understand: 1) How CEW services vary 2) Experiences of CYP and families attending clinics 3) Which modes and components of support have the greatest impact or value for CYP 4) How clinics tailor care to protected characteristics for inclusion, equity, respect, cultural sensitivity, and to counter discrimination. 5) Whether CEW services are cost effective.

Results

There is rich variety in delivery models and MDT composition including non-clinical roles such as citizens advice and family support. Weight loss medications (GLP-1s) are not commissioned at all clinics. Beyond medical and psychological treatment there is support spanning home, school and community, and to safeguard against risk of harm. Data for over 5000 patients has been analysed. Ethnic minority representation and deprivation are greater than baseline population figures for UK CYP. 24% had autism spectrum disorder. Of patients assessed 30% had liver disease and 21% had sleep apnoea. Emerging data of BMI SDS reduction from first to last appointment supports metabolic improvement of clinical significance.

Conclusions

CEW clinics are serving CYP who are typically underrepresented in health services, tailoring care to meet global needs of CYP living with obesity. Initial data suggests that the most deprived have greater improvements in BMI. Clinics directly support NHS Long Term priorities of prevention, equity, CYP mental health, and use of GLP-1 medicines with wraparound care. A national CEW service specification and quality standards framework to support future commissioning and roll out of specialist CYP obesity services will be developed.

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P65

The complexity of patients referred to a complications of excessive weight service

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Background

The Complications of Excess Weight (CEW) Service, is a tier three weight management service for children and young people (CYP), providing a holistic approach for those with, or at risk of complications due to excess weight. We aimed to better understand the health and social care needs of patients referred to a CEW service over the initial two years of operation.

Methods

Data was collected at baseline (first appointment) as part of routine clinical care. This included age, ethnicity, education status and social care involvement. Health complications related to obesity were also documented. Body mass index standard deviation score (BMISDS) was calculated at baseline and at 6-month intervals. A BMISDS reduction of 0.25 was considered clinically significant. Reasons for discharge were recorded.

Results

Over the two years, 408 CYP had access to the service. The majority were of secondary school age (182 (44.6%)). 142(34.8%) were discharged during the two years. At baseline, 58(15.7%) had obstructive sleep apnoea, with 20 (5.4%) needing ventilatory support, 74(20%), had suspected obstructive sleep apnoea, 3 were pre-school age. 8 (2.2%) patients had type 2 diabetes mellitus and 57(15.4%) had metabolic dysfunction-associated steatotic liver disease (MASLD). The frequency of complications increased with age. 85(22.97%) patients at baseline, were either currently or previously receiving support from Social Care. 5(1.4%) patients had diagnosed depression and 9(2.4%) anxiety, with a further 24(6.5%) having suspected/ self-reported anxiety. School attendance data was available for 142 patients; 7(4.9%) were not in any education or employment. Of those discharged; 17(12.2%) had achieved clinically significant weight loss, 11 (7.9%) completed 2 years in the service without a clinically significant BMI reduction, 33(23.7%) did not engage and 38(26.7%) never attended a first appointment.

Discussion

The CEW service sees CYP with considerable medical and social complexity. Engagement with the service is not optimal. Consideration should be given to how the service is portrayed to CYP and their families at referral and how better engagement can be encouraged and supported to improve outcomes.

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Pituitary and Growth**P66****After the last inch: rethinking growth hormone therapy beyond final height**

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Introduction

In adolescents with growth hormone deficiency (GHD), continuation of GH therapy beyond final height is often necessary to address persistent metabolic abnormalities. Two adolescents with congenital pan/hypopituitarism who required initiation of adult-dose GH therapy due to symptomatic hypoglycaemia will be presented.

Case 1

A patient presented at age 4 with growth failure. Height was -3.41 SDS, weight -2.49 SDS, BMI -0.2 SDS, target height 1.41 SDS, and bone age 1.5 years. TSH was 2.72 mU/L, free T4 8.4 pmol/L, ACTH 8.1 pmol/L, cortisol 339 nmol/L. Levothyroxine sodium replacement therapy was initiated for central hypothyroidism. Despite achieving euthyroidism, growth velocity remained low (3.4 cm/year); IGF-1 <3.9 nmol/L (-3.36 SDS), GH stimulation tests showed peak levels of 1.75 and 3.26 µg/L. GH therapy was initiated at age 5. Genetic analysis revealed a homozygous PROP1 gene deletion. At age 12, GH was discontinued upon reaching the 25th percentile for adult height. At 15 years, routine labs revealed hypoglycaemia; retesting showed a peak GH response of 0.11 µg/L, IGF-1 was <-2 SDS. Off-label adult-dose GH therapy was restarted, the hypoglycaemia improved.

Case 2

Referred in the neonatal period for hypoglycemia, GH levels were undetectable (<0.05 µg/L) during and after the episodes. Investigations revealed central hypothyroidism and low prolactin; adrenal insufficiency was excluded. GH therapy was initiated for neonatal hypoglycemia secondary to GHD. At age 11, she was diagnosed with type 1 diabetes mellitus, confirmed by diabetes-related autoantibodies. The final height was 156 cm. The patient had irregular follow-up visits, experienced recurrent, treatment-resistant hypoglycemia, despite very low insulin requirements (<0.05 U/kg/day). GH retesting revealed a peak <0.03 µg/L. Genetic analysis identified a homozygous POU1F1 mutation and adult-dose GH therapy was reinitiated, resulting in resolution of the hypoglycaemia.

Conclusion

In GHD patients who have achieved final height, the continued physiological roles of GH—such as maintaining glucose homeostasis, lipid metabolism, bone density, and quality of life—must be considered. Structured transition protocols should be developed to guide therapy into adulthood, paediatric endocrinologists should be enabled to prescribe adult dose GH therapy when clinically indicated.

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P67**Height velocity and final height in a cohort of children born small for gestational age and treated with growth hormone**Honor Hewitt¹, Charlotte Merson¹ & Talat Mushtaq²¹University of Leeds, Leeds, United Kingdom; ²Leeds Children's Hospital, Leeds, United Kingdom**Background**

Children born small for gestational age (SGA), aged four years or older, who do not undergo a catch-up growth are eligible for growth hormone (GH) treatment. NICE criteria indicate height velocity (HV) should increase by 50% or more from baseline in first year of treatment for treatment to be continued. We reviewed a cohort of GH treated SGA children to ascertain change in growth parameters.

Methods

Baseline, one year, and final height, weight and BMI from children treated with GH was collected. The data was analysed to assess change in HV and height SDS and then furthermore if they had met the recommended criteria for the continuation of treatment.

Results

17 children had longitudinal data available until final height (8 male; 9 female). The mean age at the start and end of treatment was 7.11 years $\pm$ 2.52 and 13.37 years $\pm$ 2.91 respectively, with treatment lasting for a mean of 6.27 years (range: 3.90 - 9.30). The pre-treatment, one year and final height SDS were, -3.10 $\pm$ 0.98, -1.72 $\pm$ 1.46 and -1.85 $\pm$ 1.22 respectively. The mean pre-treatment and one year height velocities were 4.86 cm/year $\pm$ 1.79 and 9.6 cm/year $\pm$ 2.90; (range 6.06 - 12.76) 4 patients had no improvement between starting height (-2.86 SDS) and final height SDS (-3.15 SDS). Of these, 2 also had insufficient HV in the first year to have potentially justified ongoing therapy. The HV in these two individuals was 0.11 cm/year (2%) and 1.14 cm/year (20%)

Conclusion

GH treatment results in an improvement in HV and final height in our cohort of SGA children. The first-year improvement in HV in some children is less than the current recommendations. Continuation criteria could include a pre-calculated HV and target height to be achieved after one year of treatment.

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P68**Beware the possibility of false normal results when testing for growth hormone deficiency**Charlotte Merson¹, Honor Hewitt¹ & Talat Mushtaq²¹University of Leeds, Leeds, United Kingdom; ²Leeds Children's Hospital, Leeds, United Kingdom**Background**

To minimise the risk of false-positive Growth Hormone (GH) test results, UK guidelines recommend performing two GH stimulation tests to diagnose GH deficiency. In contrast, the possibility of false-negative results is rarely discussed. We present cases in two male children who had normal GH levels on stimulation testing, yet their auxological and clinical features were consistent with GH deficiency. Both were commenced on GH treatment with positive clinical and growth responses.

Results

Case 1 presented at 4.1 years of age with a height of -3.2 SDS. The baseline IGF-I was 6.4 nmol/l (2.9 - 27). Pituitary function tests were normal with a peak GH of 7.6 mg/l. A metabolic and growth failure in early childhood gene panel were negative. Given persistent clinical suspicion of GH deficiency he had a trial of GH. The HV improved to 9.6 cm a year (height -1.27 SDS). After 4.8 years of treatment his height is -0.34 SDS. The MRI scan of his head was normal. Case 2 presented at 2.1 years of age with a height of -3.9 SDS. Pituitary function testing was normal with a peak GH of 8.2 mg/l. The IGF-1 remained undetectable before and after an IGF-1 generation test, thus primary IGF-1 deficiency was suspected. The options of a trial of GH or recombinant IGF-1 were discussed. GH was considered more practical initial therapy and the HV improved to 16.7 cm/year and height increased by 1.6 SDS in a year. An MRI showed an abnormal pituitary and absent pituitary stalk.

Conclusion

These two cases highlight the importance of prioritising clinical judgement when evaluating children with short stature. Despite normal GH levels on testing, both boys demonstrated significant catch-up growth following treatment. These cases highlight the potential for false-negative results in GH testing. An MRI of the pituitary region should be considered as part of the diagnostic workup if clinical suspicion remains. The boys' positive growth responses provide biological confirmation of GH responsiveness, supporting the use of a therapeutic trial of GH in selected cases.

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P69**BMI trajectories in idiopathic central precocious puberty during GnRHa therapy: who is most at risk?**Tugce Kandemir^{1,2}, Sarah Abdelmageed¹, Renuka Dias^{1,3}, Ruth Krone¹, Zainaba Mohamed¹, Melanie Kershaw¹, Jan Idkowiak¹, Ruchi Nadar¹ & Chamila Balagamage¹¹Birmingham Women's and Children's NHS Foundation Trust, Birmingham, United Kingdom; ²Istanbul University Istanbul Faculty of Medicine, Department of Paediatric Endocrinology, Istanbul, Turkey; ³Department of Applied Health Sciences, University of Birmingham, Birmingham, United Kingdom**Background**

Gonadotropin-releasing hormone analogue (GnRHa) therapy is the standard treatment for central precocious puberty (CPP). While weight gain during treatment is a recognised concern, its pattern, magnitude, and contributing factors remain incompletely understood.

Aim

To evaluate changes in BMI standard deviation score (SDS) during GnRHa therapy in children with idiopathic CPP and identify factors associated with increased weight gain.

Methods

This retrospective study included 73 children (66 girls, 90.4%) with idiopathic CPP treated with GnRHa. BMI SDS was recorded at baseline, after one year, and at end of treatment. BMI categories were defined as: underweight (≤ -2 SDS), normal weight (> -2 to ≤ 1 SDS), overweight (> 1 to ≤ 2 SDS), and obese (> 2 SDS), based on WHO reference standards. Associations between BMI SDS change and baseline BMI, treatment duration, age at initiation, ethnicity, and socioeconomic status were evaluated.

Results

The median age at treatment initiation was 8.39 years (IQR: 7.7-8.9), and the median treatment duration was 1.89 years (IQR: 1.2-2.58). Ethnicity distribution was 36.9% White, 32.8% Asian, 12.3% Black. Most participants (67.1%) were from the lowest three IMD deciles. BMI SDS increased significantly from baseline to one year ($+0.22$, $P < 0.0001$) and to end of treatment ($+0.32$, $P = 0.0002$), with gain correlated with treatment duration ($r = 0.3083$, $P = 0.0080$). Significant increases in BMI SDS were observed in children with both normal and elevated baseline BMI. Among those with a normal baseline BMI, the median BMI SDS increased from 0.39 to 0.75 ($P = 0.0065$). Following treatment, children, overweight/obese at baseline exhibited significantly higher BMI SDS compared to their normal/underweight counterparts (2.27 vs. 0.81, $P < 0.0001$). No significant associations were found with ethnicity ($P > 0.99$), deprivation ($P > 0.99$), or age group (< 7 vs ≥ 7 years, $P = 0.0693$), although younger age showed a trend toward greater gain.

Conclusion

BMI SDS increases early in GnRHa therapy and tends to continue throughout treatment. These findings highlight the need for early and sustained lifestyle interventions, including nutritional counselling, physical activity, and behavioural support, particularly for children with excess weight at treatment initiation.

Keywords

Central precocious puberty, BMI SDS, GnRH analogue therapy, Weight gain

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P70**Inconsistent availability and functionality of electronic growth charts across the United Kingdom**Rebecca Moon^{1,2}, Reena Perchard^{3,4}, Nadine D'Silva¹, Helen L Storr^{5,6} & Justin Davies^{1,7}¹Regional Centre for Paediatric Endocrinology, Southampton Children's Hospital, University Hospital Southampton NHS Foundation Trust, Southampton, United Kingdom; ²MRC Lifecourse Epidemiology Centre, University of Southampton, Southampton, United Kingdom; ³Division of Departmental Biology & Medicine, Faculty of Biology, Medicine & Health, University of Manchester, Manchester, United Kingdom; ⁴Paediatric Endocrinology, Royal Manchester Children's Hospital, Manchester University NHS Foundation Trust, Manchester, United Kingdom; ⁵Centre for Endocrinology, William Harvey Research Institute, Queen Mary University London, Barts and the London School of Medicine, London, United Kingdom; ⁶Paediatric Endocrinology, Barts Health NHS Trust, London, United Kingdom; ⁷Faculty of Medicine, University of Southampton, Southampton, United Kingdom

Background

Growth charting, incorporating the use of recommended reference data, is a fundamental aspect of paediatric care. Electronic growth charting software can record and display anthropometric data and generate standard deviation scores (SDS) corrected for age and sex to inform clinical decision-making. We assessed the availability of electronic growth charts, reference data used, and functionality of software across the United Kingdom.

Methods

Paediatric endocrinologists and subspecialty trainees were invited via e-mail and the BSPED newsletter to (1) complete an online survey assessing growth chart availability and brand and functionality of available electronic growth chart packages and (2) disseminate the survey to secondary care paediatricians within their regional network. Where possible, commercial software developers were contacted to determine product specifications.

Results

One hundred responses were received between June and November 2024 from 82 different hospitals, including all 22 specialist paediatric endocrinology centres. 72% of hospitals had access to electronic growth charts. These were available in all paediatric endocrine centres, compared with 62% of secondary care hospitals ($P < .001$). For only 10% of users, historical measurements had been transferred onto electronic charts when these were introduced. Twenty-eight different software were in use: 16 locally developed and 12 commercially available products. Various growth reference data were reported to be used (UK-WHO 69%, British 1990 10%, WHO child growth standards 4%, unknown 15%). Respondents using the same software often reported using different reference data. Software functionality varied between and within software. For example, overall, 71% of respondents were able to generate SDS, but for one product only 93% of users reported having this function. Sixty-nine percent reported their software can generate a mid-parental height, but only 50% of users of one software stated this function was available. BMI and occipitofrontal circumference (OFC) chart were available to 85% and 90% of respondents, respectively, but the maximum age for OFC charts ranged from 42 weeks to 18 years.

Conclusions

Electronic growth charts are not universally available. Where available, reported reference data and software functionality was inconsistent between and within software. This variability could impact clinical decision-making and access to investigation, treatment and referral to appropriate services.

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P71

Growth hormone stimulation testing in a tertiary paediatric hospital: a review of practice

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Background

Growth hormone deficiency (GHD) is an uncommon yet significant cause of short stature in children. There is no gold standard test for diagnosis of GHD, however growth hormone stimulation testing (GHST) remains a key diagnostic tool. Current international consensus recommends that failure to reach a peak GH of $> / = 7 \text{ ng/ml}$ in two separate GHSTs is needed to confirm GHD in order to limit false positive results.

Aims

This study aimed to review the use of GHSTs in children with suspected GHD in a tertiary paediatric endocrinology unit and investigate the rate of false positive results of a single GHST based on subsequent clinical diagnosis of GHD.

Methods

This was a retrospective observational study of children who underwent GHST between January 2022 and December 2023 at Children's Health Ireland (CHI), Temple Street. Data were retrieved from the endocrine nurse database, clinic letters, radiology, and laboratory systems. Age, sex, medical history, auxological measurements, IGF-I, IGF-BP3, GHST results and bone age determination were recorded. Peak GH values on GHSTs and subsequent diagnosis or exclusion of GHD were recorded for each child and the false positive rate of a single GHST was calculated. The sensitivity and specificity of IGF-I and IGF-BP3 as diagnostic markers were calculated.

Results

Out of 73 children (75% male) tested, 46% were diagnosed with GHD following one ($n = 12$) or two ($n = 22$) GHSTs. Eleven of the 12 children diagnosed following one test had a very poor GH response ($< 3 \text{ ng/ml}$) and a known structural pituitary anomaly. The false positive rate of a single GHST was 33% using a threshold of $< 7 \text{ ng/ml}$ and 23% using a threshold of $< 5.5 \text{ ng/ml}$. The majority of false positive results occurred in peripubertal boys. IGF-I was found to

have a sensitivity of 54% and specificity of 47%, while IGF-BP3 demonstrated high specificity of 100% but very low sensitivity of 12.5%.

Conclusion

This study highlights the importance of clinical judgement in the diagnosis of GHD. Due to high false positive rates of a single GHST, two tests should be conducted where possible, in particular for suspected Idiopathic-isolated GHD. Sex hormone priming should be considered for all peripubertal children.

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P72

The association between parental height and child stature: influencing socioeconomic factors in Turkish primary school children living in urban area

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Aim

Each individual has a genetic basis with a specific growth potential. Prenatal and postnatal factors influence growth by modulating genetic potential. Environmental factors such as socioeconomic factors, including maternal and paternal education, also play a role in physical growth. The aim of study was to investigate whether the height of healthy children in this age group aligns with their genetic height potential, and to assess how socioeconomic status (SES) influences these factors.

Participants and Methods

This is a prospective study conducted in Manisa province between September 2017 and June 2018. Nine hundreds seventy pre-pubertal children aged 7-8 years in the second and/or third grades of primary school and their parents were included in the study. A survey was administered to determine the SES of the children in the study, and survey responses were obtained from the parents. Height and weight of children and their parents were measured. Anthropometric data were evaluated with percentile and standard deviation score (SDS). Mid Parental height (MPH) was calculated by adding 6.5 cm to MPH for boys or subtracting 6.5 cm for girls.

Results

The mean age of the children was 7.3 ± 0.6 years. According to socioeconomic status, 28.0% (273) of children were classified as high, 39.8% (386) as moderate, and 32.1% (311) as low. Children with low SES had significantly lower height SDS than those with high SES ($P < .001$). BMI SDS was similar among SES groups. Maternal height, paternal height and MPH differed significantly between low and high SES groups ($P = 0.020$, $P = 0.003$, and $P < 0.01$, respectively). MPH SDS and percentile were significantly lower in children with low SES than in children with middle and high SES. A moderate positive significant correlation was found between children's height SDS and maternal and paternal heights, and target height SDS ($r: 0.281$, $r: 0.295$, $r: 0.252$, respectively, $P < 0.001$).

Conclusions

Socioeconomic inequalities influence parents' heights, which are reflected in their children's heights during primary school. Such a difference is not observed among children's body weights. For individuals in this age group, current height SDS may partially reflect final height SDS.

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P73

Final height in seven children treated with recombinant IGF-1 for severe primary IGF-1 deficiency

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Introduction

Growth hormone (GH) insensitivity, also known as severe primary IGF-1 deficiency (SPIGFD) is a rare condition that causes significant growth impairment in children. The classical form of SPIGFD is Laron syndrome, where a genetic defect of the GH receptor gene leads to GH resistance with high GH and low or undetectable IGF-1 levels. Twice daily injections of recombinant IGF-1 (rhIGF-1) therapy are used for the treatment of SPIGFD (height < -3 SD, serum IGF-1 < 2.5 th centile and normal GH).

Aim

To report final height in seven children with SPIGFD due to a confirmed GH receptor mutation (Laron syndrome) who were treated with rhIGF-1 (Mecasermin)

Methods

A retrospective review of all patients diagnosed with SPIGFD who received rhIGF-1 treatment

Results

Seven patients (5 male, 2 female) received treatment with rhIGF-1, at mean age of initiation of 9.1 years (range: 6.7–15.1 years) for a mean duration of 8.51 years (range: 1.5 to 12 years). The mean baseline height at the initiation treatment was -4.75 SD with a mean final height of -3.6 SD, reflecting a mean gain of 1.1 SDS. Final heights ranged from -5.44 to -2.24 SD. Height velocity increased from a baseline of 3.19 cm/year to 6.23 cm/year in the first year and 3.66 cm/year in the second year of treatment. The mean BMI increased from -1.2 SD to -0.2 SD. All children had lipohypertrophy at the injection sites. Two had lymphoid hyperplasia and snoring, one experienced hypoglycaemia. One child who started treatment at age 15.1 years had a reduction in height SDS probably reflecting that he was reaching final height prior to initiation of treatment.

Conclusion

This single-centre study, representing one of the largest reported cohorts of children with Laron syndrome in the UK, demonstrates that rhIGF-1 therapy enhances height velocity, with an increase in height velocity in the first year. There is a modest gain in final height for most patients. Suboptimal height gains may be associated with late initiation of therapy and issues with treatment adherence.

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P74

Looking beyond the thyroid - a clinical case

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An 11-year-old boy was referred to the paediatric department due to abnormal thyroid function tests (TFTs), prompted by symptoms of fatigue, poor appetite and constipation. His medical and family history otherwise unremarkable. Upon systemic examination; there were no evidence of hypothyroidism, such as hyperpigmentation, and he was pre-pubertal. However, it was noted his growth had slowed, with his weight and height dropping from the 75th percentile at the age of three, to between the 9th and 25th percentiles.

Biochemical analysis revealed the following results:

- TSH: 1.89 mIU/l (normal range: 0.60 - 4.84)
- T4: 7.5 pmol/l (normal range: 12.5 - 21.5)
- T3: 4.2 pmol/l (normal range: 3.88 - 8.02)

Anti-TPO antibodies were negative, cortisol levels were low at 60 nmol/l (normal range: 166 - 507 nmol/l) and ACTH 14 ng/l (normal range: 7.2-63.3 7-10am (ng/l)). Given the concerns regarding a potential central cause for these abnormalities, a low-dose synacthen test was administered, revealing a suboptimal response that suggested partial central cortisol deficiency. His IGF-1 level was 8.6 nmol/l (normal range: 12.3 - 51.4), indicating hypopituitarism. He was subsequently started on hydrocortisone, followed by levothyroxine. An MRI head and pituitary revealed an Adamantinomatous Craniopharyngioma (AC), which was exerting pressure on the optic chiasm, cerebral arteries and inferior frontal lobes. AC's are uncommon, representing 1.2-4% of all childhood intracranial tumours. The patient showed atypical features, lacking signs of increased intracranial pressure, headaches, or visual impairment. His subtle symptoms could have delayed diagnosis. Thankfully, he did not experience visual disturbance but had regular ophthalmological assessments to monitor this. Following neuroimaging, the patient was referred to Great Ormond Street Hospital (GOSH) for a trans-sphenoidal resection. Unfortunately, ten days postoperatively, he was readmitted due to vomiting. An MRI conducted at that time revealed early refilling of the residual pituitary cystic tumour, accompanied by haemorrhagic products. While craniopharyngiomas are known to recur, this typically occurs months to years after surgery. This case emphasises the crucial need to assess cortisol levels in any instance of hypothyroidism and highlights the necessity of considering a central cause when multiple endocrinopathies are present despite minimal clinical features.

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P75

GloBE-reg: an international registry platform for long-term evaluation of growth hormone therapy in children and adults

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Introduction

The Global Registry for Novel Therapies in Rare Bone & Endocrine Conditions (GloBE-Reg, <https://globe-reg.net/>) was launched in 2022 to support studies evaluating the effectiveness and long-term safety of specific therapies. Due to gaps in knowledge about new formulations and indications, its initial focus has been on recombinant human growth hormone (rhGH).

Methods

GloBE-Reg is structured in three dataset layers: (1) internationally agreed core elements for any rare condition; (2) therapy- and diagnosis-specific selection; and (3) a minimum dataset (MDS) tailored to the specific therapy and condition. The MDS collects information on diagnosis, treatment, clinician- and patient-reported outcomes, and adverse events and is developed with input from short-life international expert working groups.

Results

Since its launch, 32 centres from 20 countries in 4 continents have enrolled 3,580 (M:F, 2,155:1,425) patients with a median age of 12.9 years (range 0.3, 67.1), of whom 3,180 (89%) are currently under 18 years of age. Among these, 1,979 (55%) were on daily rhGH, 1,572 (44%) on long-acting rhGH. In 19 cases, rhGH therapy had not been initiated while 10 cases had discontinued the treatment. Fifteen different brands of rhGH were in use across these centres for eight indications, while long-acting rhGH was prescribed for seven indications. The most common indication was short stature due to growth hormone deficiency (60%), small for gestational age (SGA) (13%), idiopathic short stature (10%), Turner syndrome (7%), Prader-Willi syndrome (PWS) (3%), and Noonan syndrome (NS) (3%). Additionally, 61 (2%) patients were receiving rhGH for conditions that were not a recognised indication. Of the 3,580 cases, 1,166 (33%) were also included in other disease registries and 1,156 (32%) of these were entered by three centres into GloBE-Reg through its bulk upload facility. Modules containing Childhood GHD, Adulthood GHD, and NS MDS are now fully operational and are currently supporting two studies led by industry and four studies led by investigators; other modules that are currently under development include PWS and SGA.

Conclusion

The GloBE-Reg project has shown that a low-cost, drug, indication and region-agnostic platform can be widely accepted and can support several long-term safety and effectiveness studies.

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Adrenal 2

P76

Phenotypic heterogeneity and clinical implications of paediatric pheochromocytoma: two case narratives

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Introduction

Pheochromocytoma (PCC) constitutes a rare but clinically significant catecholamine-secreting neoplasm of chromaffin cell origin, predominantly arising within the adrenal medulla. Its incidence in the paediatric population is exceedingly low (0.2–0.3 cases per million children), yet it bears substantial morbidity due to hypertensive crises and catecholamine-induced cardiomyopathy. This report delineates two phenotypically divergent paediatric cases, underscoring the diagnostic complexity, variable clinical trajectories, and therapeutic strategies necessitated by this neuroendocrine tumour.

Case presentation

The first case involves a 6-year-old male presenting acutely with fever, dyspnoea, and paroxysmal tachyarrhythmia. Cardiological evaluation revealed dilated cardiomyopathy with a severely depressed ejection fraction (26%). Biochemical profiling demonstrated markedly elevated plasma and urinary normetanephrines and vanillylmandelic acid. Cross-sectional imaging identified a right-sided adrenal mass, and subsequent laparoscopic adrenalectomy confirmed PCC with a high Pheochromocytoma of the Adrenal gland Scaled Score (PASS = 9), suggestive of aggressive biological behaviour. Loss of succinate dehydrogenase complex subunit B (SDHB) immunoreactivity and a pathogenic variant in the Von Hippel-Lindau (VHL) gene were consistent with syndromic PCC. The patient remains under oncological surveillance three years postoperatively. Conversely, the second case describes a 13-year-old male with a pre-existing bicuspid aortic valve who presented with headache, dizziness, and sustained

hypertension. Biochemical indices revealed substantial catecholaminergic excess; Contrast MRI delineated a 4 cm right adrenal tumour. Despite initial intolerance to phenoxybenzamine, he achieved haemodynamic stability with doxazosin prior to successful laparoscopic resection. Histopathology confirmed PCC with loss of SDHB staining and a moderate PASS score (4), predicting risk of recurrence. Genetic screening was negative for common susceptibility loci, and the patient has entered routine follow-up.

Conclusion

These cases exemplify the phenotypic breadth and genetic underpinnings of PCC in childhood. They reinforce the imperative for heightened clinical vigilance in hypertensive children, especially in the context of cardiac dysfunction or ambiguous symptomatology. Prompt biochemical and radiologic evaluation, judicious preoperative alpha-adrenergic blockade, and precise surgical intervention remain cornerstones of optimal paediatric PCC management.

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P77

Assessing effects of low dose prophylactic hydrocortisone on hypothalamic–pituitary–adrenal axis in preterm infants: systematic review

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Background

Prophylactic low dose hydrocortisone in extremely preterm neonates (<28 weeks) is increasingly used to reduce bronchopulmonary dysplasia (BPD), yet its impact on the developing hypothalamic–pituitary–adrenal (HPA) axis is uncertain.

Methods

A systematic review was registered with PROSPERO (CRD42025640878). PubMed, Scopus, Cochrane Library and Google Scholar were searched from inception to April 2025 using terms related to prematurity, hydrocortisone, cortisol and HPA axis. Two reviewers independently screened titles, abstracts and full texts; a third independent reviewer resolved decisions when consensus was not reached. The primary outcome was biochemical or clinical evidence of adrenal insufficiency after hydrocortisone therapy use in the ex-preterm (<37 weeks) neonate.

Results

Of 993 records, 821 remained after duplicate removal. 36 full text articles were assessed. 23 studies were excluded for evaluating dexamethasone (n = 17) or inhaled corticosteroids (n = 5); 11 lacked HPA outcomes. Three studies remained. Two did not directly assess adrenal insufficiency (Romijn 2023, Takayanagi 2015). The single eligible manuscript (Rosano 2024) measured random plasma cortisol concentrations after hydrocortisone therapy to determine the utility of cortisol in the assessment of adrenal insufficiency, however dynamic testing was not performed on all patients and therefore diagnosis of patients was not uniform.

Conclusion

No contemporary study rigorously evaluates the short or long term effect of prophylactic low dose hydrocortisone on adrenal function in preterm infants. Given the expanding use of prophylactic hydrocortisone to prevent BPD, high quality prospective trials incorporating appropriate assessment of the HPA axis are required.

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P78

Urinary steroid metabolomics, body composition and metabolic risk in children with premature adrenarche

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Background

Premature adrenarche (PA) is a common presentation in pre-pubertal children and mostly occurs in girls before their 8th birthday. There is controversy about whether children with PA are at higher risk of developing metabolic complications or may progress to developing Polycystic Ovary Syndrome (PCOS) in later life.

Aim

We aim to characterise a phenotypic, hormonal and biochemical metabolic risk signature in cohorts of children with PA.

Design and Methods

Single-centre case-control study in children with PA and PCOS, matched to healthy volunteers. We assessed auxology, (height, weight, BMI [z-score], waist-to-hip/length ratio, dual-energy X-ray-absorptiometry), fasting hormonal (DHEAS, androstenedione, testosterone, SHBG, prolactin, IGF-1), 24-hour urinary steroid profiling, and standard metabolic risk markers (lipid profile, HbA1c, glucose).

Results

68 PA children (58 girls, 10 boys; age: 7.7 years, range 4.4-9.9) and 30 healthy controls (18 girls, 10 boys; age: 7.7 years, range 4.5-10.3) were included; precocious puberty, and congenital adrenal hyperplasia were excluded. Weight, BMI and BMI z-score were significantly higher in PA compared to controls. Androgens were also significantly higher in PA: DHEAS (median 2.87 [IQR 2.01-3.38] vs 1.08 nmol/l [0.7-1.73]; $P < 0.001$), androstenedione (1.2 [0.85-1.6] vs 0.75 [0.58-1.43] nmol/l; $P = 0.023$) and testosterone (0.3 [0.2-0.3] vs 0.15 [0.1-0.225] nmol/l; $P = 0.0015$). We also observed significantly lower fasting HDL cholesterol (1.50 [1.4-1.6] vs 1.40 [1.2-1.5] mU/l; $P = 0.033$) and SHBG (61 [45.25-90.75] vs 88 [62-119] nmol/l; $P = 0.0219$). Urinary androgen metabolites An, Et, DHEA, and SPT are higher in PA compared to controls. IGF1 was higher in PA compared to controls (30.1 [26-35.9] vs 17.7 [13.9-24.4] nmol/l; $P < 0.0001$). There was a mild association between PT and fasting glucose in PA. We did not observe any significant differences between PA and controls for HbA1c, triglycerides, total/ LDL cholesterol and prolactin.

Summary and Conclusion

Children with PA are heavier and taller, they have higher IGF1 adrenal androgen production. Recruitment is ongoing, and more in-depth biochemical assessment, such as fasting insulin/ HOMA-IR (B) and multi-steroid profiling, may help to identify metabolic risk parameters.

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P79

A rare case of adrenal insufficiency associated with allgrove syndrome

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Introduction

Allgrove syndrome (AS) or 3A syndrome (AAA) is characterized by a triad of adrenocorticotrophic hormone (ACTH) resistant adrenal insufficiency, alacrimia, and achalasia along with progressive neurological impairment with or without mild mental retardation. Sometimes known as 4A syndrome, if it also involves autonomic dysfunction. The molecular basis for Allgrove syndrome appears to be an autosomal recessive pattern of inheritance. Having only two clinical features also strongly suggests Allgrove syndrome.

Case

Here we present a case of a male child with first-degree consanguineous parents who presented to the healthcare setting several times due to vomiting and poor weight gain. Mum reported vomiting most of the feeds, and vomiting has been getting worse since starting weaning food. A trial of feeds thickener, domperidone and omeprazole had little effect. Child's weight dropped to below 0.4th centile. The mother also mentioned observing that he cries without tears. An ophthalmological exam was arranged, which was reported as normal. An upper GI contrast study was done that showed oesophageal achalasia. A detailed workup showed low early-morning cortisol levels. An SST confirmed the diagnosis of adrenal insufficiency. He was started on oral hydrocortisone for and was referred

to a Paediatric surgeon. Genetic testing could not be performed due to financial constraints. He had balloon dilatation and myotomy done for achalasia. He had mild mental retardation and developmental delay.

Discussion

The clinical presentation AAA syndrome depends on the patient's age. Ophthalmological abnormalities are usually present at birth, whereas adrenal and gastrointestinal abnormalities typically appear after the first six months to the first decade of life. Many of the patients with AAA syndrome are diagnosed in childhood, but its presentation varies. Alacrimia is usually the first sign present at birth. Prompt treatment is necessary for adrenal insufficiency. Prompt surgical referral to treat Achalasia is important. Allgrove's syndrome may be an underdiagnosed disorder, and a high index of suspicion is needed when patients present with such complex symptoms at variable stages, i.e. failure to thrive, dysphagia, crying and alacrimia.

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P80

Adrenal complications in paediatric patients living with spinal muscular atrophy – a single centre retrospective study

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Background

Recently introduced disease modifying treatments have dramatically changed the clinical course of SMA and improved survival. As a consequence, children are experiencing more complications secondary both to the disease and the new treatments. All infants and children who receive the gene therapy onasemnogene abeparvovec (OA) require a minimum of 28 days of high dose steroids to counteract common side effects, putting them at risk of steroid-induced adrenal insufficiency (SIAI) and adrenal crisis. This study aims to describe adrenal complications in the UK paediatric SMA population treated with OA and high dose steroids.

Methods

A retrospective case notes review was conducted on all paediatric patients living with SMA who received OA managed at our tertiary centre. Data on steroid duration and doses, adrenal function testing and the frequency of SIAI were collected. The risk factors for SIAI in this population were explored.

Results

Eleven children received OA and adjuvant prednisolone, 10 of whom had SMA type 1 and one was presymptomatic. Mean age at infusion was 2.6 years (range 0.5-7.3). All children were started on 1 mg/kg of prednisolone for a minimum of one month as per the OA treatment protocol and six patients (54%) required an increase to 2 mg/kg due to infusion related side effects. One child required three doses of methylprednisolone. The average duration of supra-physiological steroids was 126 days (range 52-210). The mean duration of steroid weaning was 80 days (range 19-182). Four patients (36%) developed SIAI, among whom the maximum time to full adrenal recovery was 29 months (range 6-29). There was no difference in weight and age at infusion or duration of supra-physiological steroids between children with and without SIAI.

Conclusion

A significant proportion of our cohort developed SIAI. No potential risk factors for SIAI were identified, however the sample size was small. There was wide variation in steroid weaning duration. Currently there are no specific steroid weaning guidelines for this population. We are conducting a large multi-centre UK study (SMA-ABC) to further examine adrenal complications in SMA and inform future guideline development.

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P81

Real-world stability of salivary cortisol and cortisone samples

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Background

Salivary cortisol (SC) and cortisone (SCn) samples are stable under a range of laboratory conditions, although high temperatures lead to faster degradation of samples and decreased stability. Sample stability in 'real world' environments is required to assess the impact of exposure to elements that may not be examined in a controlled laboratory environment.

Aim

To report stability of SC and SCn in samples returned to the laboratory by post and car journeys.

Methods

Three sets of samples collected from adult volunteers: One control sample stored in the laboratory at -80°C, one posted by second-class Royal Mail, and one transported by 30-minute car journey. SPSS was used for data analysis; Difference between measurements was tested by paired T-test for normally distributed data, Wilcoxon test for non-normally distributed data. Samples collected during the cooler Winter months and hot Spring months were also examined separately by Pearson's (r) for normally distributed data and Spearman's correlation (p) for non-normally distributed data.

Results

n = 18, contributed 66 samples (22 per group)

Conclusion

There was no significant difference between control samples and those transported by car or post. Conversely, correlation analysis indicated that samples returned to the laboratory during warm weather may deteriorate during transport. We recommend that samples be frozen in household freezer before returning samples by car or first-class post, in the first postal collection of the day, Monday – Thursday only.

Table 1 Median (range) SC and SCn concentrations in control, car and postal samples

	SC Control (nmol/l)	SC post (nmol/l)	SC car (nmol/l)	SCn Control (nmol/l)	SCn post (nmol/l)	SCn car (nmol/l)
All Samples	5.0(1.1-9.3)	5.0(0.4-7.0)	4.0(1.0-7.0)	25.0(10.5-43.8)	*22.0(0.3-35.1)	25.0(9.6-35.1)
Winter Samples	5.0(2.0-9.3)	4.0(2.7-6.7)	5.0(3.1-6.9)	24.0(16.6-36.8)	21.0(18.7-35.1)	22.0(16.2-30.2)
Spring Samples	5.0(1.1-8.3)	5.0(0.4-7.0)	4.0(1.0-7.0)	28.0(10.5-43.8)	23.0(0.3-34.7)	27.0(9.6-35.1)

* P < 0.05

Table 2 SC and SCn correlation of car and postal samples with control samples.

	SC post p = 515*	SC car r = 0.534*	SCn post r = 0.575*	SCn car r = 0.341
All Samples				
Winter Samples (n = 8)	r = 879*	r = 0.758*	r = 0.935*	r = 0.781*
Spring Samples (n = 14)	p = 0.336	r = 0.444	p = 0.336	r = 0.444

* P < 0.05

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P82

Iatrogenic adrenal suppression in children: how 'stressed' should paediatricians be?

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Introduction

Steroid treatment is beneficial and important for many conditions in paediatrics. However, paediatricians are forced to balance the benefits with a number of side effects, including dysglycaemia, weight gain and adrenal suppression. Early diagnosis and treatment of adrenal suppression is important for morbidity and mortality due to the risk of an adrenal crisis, most commonly after gastrointestinal infections.

Aims

Our primary aim was to identify the incidence of patients with steroid-induced adrenal suppression at our local DGH, from endocrinologist referrals, due to concern of prolonged or high-dose steroid use. We hoped to describe the characteristics of patients with adrenal suppression and establish a management pathway.

Results

We had a total of 39 patients referred due to prolonged or high-dose steroid use. 13 (33%) were female and the most common primary diagnosis was respiratory (30). 3 (7.7%) of our patients were suppressed, with asthma, IBD and nephrotic syndrome. Two patients had been managed on long-term oral steroids (prednisolone/budesonide), whilst one required multiple oral courses, in addition to high-dose inhaled steroids (Seretide 1000 mg/day). All 3 children were started on hydrocortisone and given sick day rules as per BSPED guidelines.

Discussion

Whilst we know the risk of adrenal suppression in adults on steroids, there is little data in children and no clear evidence on cumulative steroid dose thresholds for

development of adrenal suppression. Our suppressed patients had all been treated with oral steroids over the previous year, with one using high-dose inhaled steroids. The steroid doses were above the screening threshold of our tertiary guideline. Most children on inhaled steroids were not suppressed, despite being above the guideline threshold. Studies have shown that inhaled corticosteroids have the potential to cause adrenal suppression in paediatrics, although our results show this to be at a high level, combined with additional oral steroids. Further research on thresholds will help guide clinicians on when to screen for adrenal suppression, together with research on the reliability of salivary cortisol measurements, to prevent noxious tests in our paediatric cohort. In the meantime, it is pertinent to continue to educate families and clinicians on adrenal crisis to safeguard our patients.

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P83

National survey of current UK practice for the assessment of neonatal adrenal function

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Introduction

Adrenal function testing in neonates is undertaken for multiple indications however permanent neonatal adrenal insufficiency (AI) is rare. Accurate assessment of neonatal adrenal status is challenging due to changes in the adrenal gland perinatally, non-specific clinical signs, and a paucity of normative data.

Objective

Describe current UK practice regarding investigation and management of neonates considered at risk of AI.

Methods

A 46-question survey evaluating approach to: unstimulated and stimulated (SST) cortisol testing, result interpretation and management was sent to paediatric residents and consultants practising endocrinology at tertiary and secondary care centres throughout the UK.

Results

Responses were received from 35 centres, 18/22 tertiary and 17 secondary care centres. 88% of respondents were consultants (31/35). There was consensus on indications for neonatal cortisol testing with the majority having guidelines recommending testing in hypoglycaemia (33/35), ambiguous genitalia (27/35) & conjugated hyperbilirubinaemia (20/35). Only 9/35 of centres currently have a specific guideline relating to neonatal adrenal assessment, results interpretation or management. Most centres (31/35) do not use neonatal specific laboratory cut-offs for cortisol. Moreover, the laboratory cut-offs for cortisol sufficiency were significantly higher (median = 295nmol/l, range 100-500) than the level clinicians consider highly suspicious of AI (median = 100nmol/l range 50-420). Despite this discrepancy in biochemical and clinical thresholds the majority advised proceeding to an SST (26/35) following a laboratory reported abnormal cortisol. The SST testing method reported was generally uniform - standard dose and no recommended specific time of day (28/35). Approach to interpretation and management of abnormal SSTs varied. 14/35 employ a borderline category for peak cortisol cut off e.g. sick day steroids then retest prior to discharge. For failed SSTs the management and retesting also varied: individualised patient approach (11/35), start replacement hydrocortisone (14/35). Reported retesting timeframes ranged from 4 weeks to 6 months, at term or on discharge.

Conclusion

There is wide variation across the UK in approach to neonates at risk of AI, likely reflecting lack of a robust evidence-base to support safe but pragmatic testing, result interpretation and management of cortisol levels in neonates.

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P84

Predictive value of early morning cortisol levels in diagnosing adrenal insufficiency: a retrospective audit

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Background

Adrenal insufficiency (AI) is a potentially life-threatening condition if not identified and treated promptly. While the short Synacthen test (SST) remains the gold standard for diagnosis, it is not always available. Early morning cortisol (EMC) measurement is widely used as a practical screening tool; however, cut-off values vary across centres.

Aim

To assess the diagnostic accuracy of EMC levels in predicting AI using SST as the reference standard and to evaluate whether SSTs can be avoided in clear-cut cases.

Methods

This retrospective audit was conducted at Oxford Children's Hospital and reviewed 134 records of patients who had EMC measurement between 2017 and 2023. Patients aged > 6 months and < 17 years, who had an early morning cortisol measurement taken within 30 days prior to the SST were included into analysis. An adequate adrenal response was defined as a peak cortisol ≥ 430 nmol/l at 30 minutes post-Synacthen. Collected data included demographics, clinical presentation, history of steroid exposure, EMC levels, and SST outcomes. Receiver operating characteristic (ROC) curve analysis was performed to assess the predictive value of EMC.

Results

A total of 42 patients (mean age 9.5 ± 5 years) were included. Adrenal insufficiency was diagnosed in 5 patients (11.9%), while 37 had normal SST responses. Presenting symptoms included hypoglycaemia (12%), recurrent vomiting (10%), and fatigue or lethargy (38%). Long-term steroid exposure was identified in 7% of the cohort. The median early morning cortisol level was 159.0 nmol/l in patients diagnosed with AI and 196.0 nmol/l in those without. ROC analysis yielded an area under the curve (AUC) of 0.76, indicating fair discriminatory ability of EMC. A cortisol threshold of < 100 nmol/l strongly suggested AI, while levels > 450 nmol/l reliably excluded it. Intermediate values were inconclusive and required confirmatory SST.

Conclusion

Early morning cortisol measurement demonstrates acceptable predictive value for diagnosing adrenal insufficiency. Thresholds < 100 nmol/l (confirming AI) and > 450 nmol/l (ruling out AI) can provide practical clinical guidance. This audit supports the role of early morning cortisol as an effective screening tool, potentially reducing the need for SST in clear-cut cases.

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Diabetes 4

P85

Disaster response and diabetes: rebuilding support systems for children and young people with type 1 diabetes post-earthquake in myanmar 2025

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Objectives

To evaluate the impact and operational insights of a rapid, multi-layered emergency response led by Action4Diabetes (A4D) to ensure uninterrupted access to insulin, blood glucose (BG) monitoring kits, and psychological support for children and young people (CYP) with type 1 diabetes (T1D) following the 2025 Myanmar earthquake.

Background

On the 28 March 2025, central Myanmar was struck by two powerful earthquakes (7.7; 6.4 magnitude), causing over 5,000 fatalities, including one A4D-supported T1D child. The disaster affected about 17 million, with critical disruptions in healthcare, electricity, and communications. CYP with T1D were severely impacted as widespread power outages destroyed cold-chain insulin storage and telecommunications failed.

Methods

A4D, a UK-registered charity supporting more than 1,000 T1D CYP in Southeast Asia, launched a three-tier emergency assessment: (1) reviewed deliverables and operational status of partner hospitals, (2) evaluated patient safety, insulin availability and care access from digitally health records, and (3) coordinated with local distributors for emergency stock and logistics to supply and deliver. Priority was given to patients with less than one week's insulin supply. Aid packages were distributed and services temporarily resumed at Kandaw Nadi Hospital within the week. Once connectivity was restored, a UK-based psychologist provided online psychological training to Myanmar healthcare staff.

Results

Within the week, 72 of 101 affected CYP with T1D were contacted. By week three, 44 CYP had received insulin, 65 aid packages were delivered, and 8 home

visits were made despite major infrastructure challenges. Pre-disaster, supplies were routinely delivered via cargo shipments; however, due to damage to roads, bridges and infrastructure, A4D shifted to smaller-scale logistics, using mini-vans and small passenger vehicles for approximately 3 weeks to maintain deliveries and supplies.

Conclusion

In the aftermath of the earthquake, the destruction caused significant challenges in the timely procurement, distribution and delivery of essential diabetes management supplies, directly impacting CYP with T1D who rely on uninterrupted access to these critical resources. The rapid response highlights the critical need for digital health records, adaptable communication, decentralised supply chains, strong local coordination, and integrated psychological support to ensure continuity of T1D care during large-scale emergencies.

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P86

Barriers and facilitators in transitioning to multiple daily insulin in a resource limited setting: a qualitative study of laotian children and young people with T1D

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Introduction

In many low income countries in Southeast Asia, twice daily pre-mixed insulin regimen has been the conventional insulin therapy initiated for children and young people (CYP) with type 1 (T1D) diabetes. In Laos, universal health coverage for diabetes does not include insulin provision and blood glucose testing kits. We aimed to explore, This study aims to explore the views and perceptions of CYP with T1D in Laos on how transitioning from twice daily insulin regimen to a MDI regimen affected their diabetes management and quality of life.

Methods

Data were collected from the medical records of, and through individual semi-structured face to face interviews with, CYP supported by the Action4Diabetes program in Laos. Participants were recruited as they switched from a BD to MDI regimen. Quantitative data were stratified into HbA1c 6 and 12 months before and after the switch, and male vs. female sex; associations were examined using t-tests. Qualitative data were analysed using Gibbs's framework.

Results

Overall, 24 youth (62.5% female) from across nine provinces transitioned to an MDI regimen. In the 6 and 12-month periods prior to the switch, mean $\pm$ SD HbA1cs were 8.8 ± 2.3 and $8.3 \pm 2.2\%$ respectively. In the 6 and 12-month periods following the switch, HbA1c had improved to 7.6 ± 2.7 and $7.7 \pm 2.1\%$, respectively. No differences were observed between males and female sexes. Interviews were conducted with 15 CYP (73.3% female). Mean ages at T1D diagnosis was 10.6 years and at the switch time were 14.3 years (range 4-24) respectively. Describing how transitioning to an MDI regimen could and did affect their T1D management and quality of life, three themes emerged: pragmatism; empowerment and agency; and foundations of success. Prior to transitioning, lack of confidence in carbohydrate counting and injections during school-time were raised as barriers. However, the switch was viewed positively, with cited benefits including increased food flexibility, and improved glucose stability (with less hypoglycaemia), and sense of well-being.

Conclusions

This study provides valuable insights that will guide future work in supporting the switch for youth with T1D from BD to MDI regimens.

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P87

ADNAT- a digital health assessment app for enhancing diabetes care and wellbeing in children and young people

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Background

The Adolescent Diabetes Needs Assessment Tool (ADNAT) is a validated, evidence-based digital health assessment tool designed to support children and young people (CYP) with Type 1 Diabetes (T1D) to manage their condition and engage in shared decision-making. ADNAT has been developed into a mobile and tablet accessible app. The app supports healthcare professionals to provide targeted support across domains to improve mental health, wellbeing, nutrition and medicine self-management.

Methods

A multi-method evaluation conducted at an NHS site explored the acceptability and perceptions of CYP, parents and professionals of implementing the app into clinical practice.

Results

CYP reported that despite the app having 'lots of questions', it was 'quick and easy' to use as they could 'dip in and out' and didn't have to do it all in 'one go'. CYP also described how ADNAT improved their knowledge and helped them reflect on their diabetes management. Parents felt the ADNAT app helped their child gain control of their consultation and complete the assessment in a less rushed and more meaningful way. Professionals reported that whilst implementing ADNAT had required the paediatric diabetes team to adapt some systems, it had prompted a more targeted consultation based on a young person's need.

Conclusion

ADNAT aims to replace traditional paper-based assessments, such as PedsQL, enabling CYP to complete assessments remotely via familiar technology before clinic appointments, thereby improving accessibility, the quality of consultations and saving time for both CYP and clinicians. To support the implementation and long-term sustainability of ADNAT, a dedicated website with user guidance and training materials for both professionals and CYP is under development. Following successful evaluation, ADNAT can be scaled beyond the initial NHS Trust to support national adoption in paediatric diabetes services.

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P88

Evaluating inequalities in the use of diabetes-related technology in children and young people from a black, asian and minority ethnic background in nottingham children's hospital

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The National Paediatric Diabetes Audit (NPDA) has identified inequalities in diabetes care associated with ethnicity, including use of diabetes-related technology. Children with type 1 diabetes mellitus (T1DM) from a Black, Asian, and Minority Ethnic (BAME) background are less likely to use routine continuous glucose monitoring (CGM), insulin pumps and hybrid-closed loop (HCL) systems (NPDA 2019-2023). HCL use is associated with enhanced glycaemic control and quality of life for patients and their carers (Ng *et al*, 2024). Using the Diamond electronic patient record, we identified 104 patients with T1DM under the care of Nottingham Children's Hospital who identify as a non-white ethnicity. Records were reviewed (November 2024 – March 2025) to collect data on CGM, pump, and HCL use. Reasons for non-use and data on clinical outcomes such as HbA1c and time in range over a 3-month period was also collected. Eighty-six patients (82%) were on pumps/HCL compared to 66% overall clinic uptake, 55% national uptake (NPDA 2023-24) and 58% regional uptake (NPDA Unit Level Report East Midlands 2023-24). Of those not on a pump, all were offered HCL therapy: 2 were awaiting pump starts, 5 recently attended a showcase, 4 had booked a showcase, and 2 were previously on a pump and decided to revert to injections. Eleven patients were not on a pump and had not attended/planned to attend a showcase. Reasons given included: concerns about sport, social stigma, disliking the sensation of being attached to a pump and, anxiety. These patients had a similar HbA1C (54.4 mmol/mol) and TIR (61.8%) to those on HCL (54.5 mmol/mol and 61.9% respectively). Older male teenagers were overrepresented in those declining pumps/HCL. Allowing for small numbers in individual groups, there was no ethnic group markedly less likely to decline pump/HCL use. HCL is now considered standard care and, reassuringly, ethnicity does not appear to be associated with reduced diabetes technology use in the NUH cohort. However, male teenagers are at risk of being disadvantaged and require specific consideration. Nevertheless, barriers to use remain, and are complex and multifactorial. To reduce disparities, care teams must continue to ensure that care is culturally, socially, and ethnically relevant.

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P89

A regional 12-month project to evaluate diabetic ketoacidosis (DKA) presentations at diagnosis of type 1 diabetes

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Background

Reduction in DKA at diagnosis of type 1 diabetes (T1D) is part of Aim 1, National CYP Diabetes Network (NCYPDN) Delivery Plan, with recommendations to have a system in place to understand DKA rates. 2. A regional audit in 2022/23 on DKA at diagnosis identified a higher incidence than previously reported, with prior HCP contact in a significant proportion.

Aim

A regional review of children and young people (CYP) presenting in DKA at diagnosis of T1D to understand rates and trends. To raise awareness and respond where NICE NG18 is not appropriately followed.

Method

Prospective data collection from April 2024 for 12 months. 18 units submitted data using a questionnaire based on Aim 1 recommendations. Data was uploaded to a real-time dashboard. The Network provided comprehensive reports for each unit and combined Network analysis.

Results

113 CYP presented in DKA, with 43% in severe DKA. 59% sought prior medical advice, with recognition or response to symptoms and access to GP review identified as modifiable factors. Lack of family awareness of symptoms was highlighted in 42.6%. Comparison with the NPDA Dashboard at 9m suggested under-reporting of DKA at diagnosis (31.6% Network Vs 13.7% NPDA).

Conclusions

Rates of DKA were higher than previously reported, with modifiable factors delaying diagnosis identified. Continuous data review enabled early identification of trends and real-time actions to be implemented, including education sessions within primary care and recirculating updated Network Primary Care Referral Guideline. Data collection was limited to meet objectives of understanding rates of DKA and delays, but comparative data on those not in DKA at diagnosis may have allowed more robust analysis of risk factors.

1. DKA prevention at diagnosis NCYPDN 2. A regional audit of diabetic ketoacidosis (DKA) presentations at diagnosis of type 1 diabetes (T1DM) and DKA management | BSPED2024 3. Overview | Diabetes (type 1 and type 2) in children and young people: diagnosis and management | Guidance | NICE

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reduction in HbA1c was attendance to the 6 week PDSN follow up, which reduced HbA1c by 19.19%. The second largest factor affecting reduction in HbA1c was commencing an insulin pump which saw a 18.65% reduction in HbA1c. The reduction in CYP already on insulin pumps was less pronounced with an 11.6% reduction in HbA1c.

Discussion

Implementation of a 'High HbA1c pathway' for CYP with T1DM can have a positive effect in reducing high HbA1c and subsequent complications. The greatest factor affecting reduction in HbA1c was attendance to nurse-led clinics which demonstrates the importance of patient and family engagement in healthcare outcomes. Given starting insulin pump therapy was the second largest factor affecting reduction in HbA1c, a further study is underway to determine whether the improvement in HbA1c upon starting insulin pump therapy is sustained. As a result of this study the median HbA1c for all CYP with T1DM at FPH reduced from 61 mmol/mol (2023/2024) to 56 mmol/mol which places FPH in the top 5 performing units in London and the Southeast.

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P91

Diabetes mellitus characteristics in an international trial cohort with wolfram syndrome

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This study reports baseline diabetes mellitus (DM) characteristics and predictors of DM progression in 63 participants with genetically confirmed Wolfram syndrome enrolled in the TREATWOLFRAM international clinical trial (ClinGov NCT03717909). Wolfram syndrome is a rare, monogenic, neurodegenerative and DM disorder typically diagnosed in childhood or adolescence. Participants were recruited from the UK, France, Poland, and Spain between 2019 and 2021. Beta cell function at baseline was assessed by haemoglobin and insulin dose-adjusted A1c (HbA1c and IDAA1c), fasting C-peptide, and fasting C-peptide corrected for fasting blood glucose. These were then compared according to age, sex, time since DM diagnosis, and *WFS1* mutation type. At baseline, 97% had DM, with a median duration of 10.6 years (range 1 day to 51.0 years). Median HbA1c was 7.5% (range 5% to 12.4%); median IDAA1c was 10.8 (range 5.0 to 16.7); median fasting C-peptide was 86 pmol/l (range 31.5 pmol/l to 472.5 pmol/l); and median glucose-adjusted fasting C-peptide was 7.8 (range 2.9 to 52.5). Fasting C-peptide levels were inversely correlated with time since DM diagnosis ($P = 0.03$, $n = 14$), declining most rapidly in the first 9.75 years according to piece-wise regression. Fasting C-peptide levels were typically below 80 pmol/l (indicating absolute insulin requirement) after 9.30 years. Glucose-adjusted fasting C-peptide showed a similar negative correlation with time since DM diagnosis ($P = 0.01$, $n = 14$). Results suggested higher C-peptide levels in males (median 207.5 pmol/l, range 65.5 to 472.5 pmol/l, $n = 6$) than females (median 61.75 pmol/l, range 31.5 to 98 pmol/l, $n = 8$). No associations were found between beta-cell function markers (C-peptide, IDAA1c, HbA1c) and age, sex, or type of *WFS1* mutation. There was a significantly younger age of DM

P90

Reduction of high HbA1c in children and young people with type 1 diabetes mellitus through implementation of a 'high HbA1c pathway'

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Introduction

This project investigated how to improve the HbA1c of Children and Young People (CYP) with Type 1 Diabetes Mellitus (T1DM) with a high HbA1c at Frimley Park Hospital (FPH).

Methods

CYP with a HbA1c ≥ 69 mmol/mol were placed on the 'High HbA1c Pathway' which involves their keyworker being informed; more frequent contact; involvement of a dietician; discussion topics and paediatric diabetes specialist nurse (PDSN) follow up in 6 weeks.

Results

76 CYP were recorded to have a HbA1c ≥ 69 mmol/mol between September 2023 and October 2024. 13 CYP were excluded due to recent T1DM diagnosis or they had transitioned to adult services. 63 CYP were included in the study. At the start the average HbA1c of the CYP was 80.22 mmol/mol; average age was 13 years and 33% were on insulin pump therapy. At the end of the study 40 CYP had a HbA1c < 69 mmol/mol and average HbA1c reduced to 69.05 mmol/mol. Average reduction in HbA1c was 12.64%. The greatest factor affecting the

onset for individuals with higher frequencies of nonsense/frameshift mutations ($P = 0.002$). The study concludes that C-peptide levels decline over time in individuals with Wolfram syndrome, falling to $<80\text{pmol/l}$ after 9.30 years. Results showed some difference in C-peptide between males and females. There is little influence from age or genetic mutation class. These findings are important for clinical management and for evaluating new treatments targeting diabetes progression in Wolfram syndrome.

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P92

Audit of national first year of care (FYOC) pathway for children and young people (CYP) with a new diagnosis of type 1 diabetes in England
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Introduction

The first year following diagnosis of Type 1 diabetes is a crucial period for children and young people (CYP), establishing the foundation for long-term glycaemic control and self-management. Early optimisation of glucose control is associated with reduced risk of diabetes-related complications in later life.

Objectives

To assess the impact of a standardised First Year of Care (FYOC) pathway on clinical and process outcomes in CYP newly diagnosed with Type 1 diabetes.

Methods

This retrospective audit included CYP with new-onset Type 1 diabetes across six NHS Trusts. Data were collected for two periods: pre-FYOC (April 2021–March 2022) and post-FYOC (April 2023–March 2024). Outcomes included proportion with HbA1c $<48\text{ mmol/mol}$ at 4–6 and 9–12 months, time in range (TIR, 4–10 mmol/l) $>70\%$ at 6–12 months, psychology review within 2–6 weeks, and completion of Level 3 carbohydrate counting. Qualitative data was collected from each organisation.

Results

There were 247 CYP in the pre-FYOC group and 198 post-FYOC. At 6 months, 36% of the post-FYOC group had HbA1c $<48\text{ mmol/mol}$ vs 27% pre-FYOC ($P = 0.009$). At 12 months, 41% of the post-FYOC group had TIR $>70\%$ vs 32% pre-FYOC ($P < 0.001$). By 12 months, 60% of CYP in the post-FYOC group were using hybrid closed loop (HCL) systems. HCL use was highest in the 0–4 year group (73%) but HbA1c $<48\text{ mmol/mol}$ was lower in this group (20%) compared to 42% in 15–19 year olds. Qualitative data highlighted challenges in timely psychology access and identifying CYP in their first year when attending general clinic.

Conclusions

The FYOC pathway improved clinical and care process outcomes in newly diagnosed CYP. NICE TA943, implemented in December 2023, facilitated HCL access nationally; the FYOC pathway supported high uptake within the first year. Successful implementation requires clearly defined local processes for each CYP and a well-resourced multidisciplinary team, including psychology input.

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P93

The impact of proactive introduction of hybrid closed loop pump therapy on glycaemic control and reduction in inequity of tech uptake

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Introduction

Hybrid closed loop pumps (HCL) incorporate insulin pumps (IP) with continuous glucose monitoring (CGM). We reviewed the glycaemic control of patients followed up at Evelina with Type 1 Diabetes(T1D) on HCL pumps between January 2021 and December 2024.

Method

As per standard care, patients were reviewed in clinic every 3 months. We collected HbA1c and Time in range data before starting HCL and at 3months, 6months and 1 year after initiating on HCL. We also reviewed the ethnicity distribution and deprivation index among HCL users. We compared the present data with our 2023-2024 National Paediatric Diabetes Audit (NPDA) data.

Results

There were 88 patients on HCL, of which 46(52.27%) were males. Median age was 12.29 years (range 2.08- 19.38). Before starting HCL, 35 were on standard insulin pumps (39.77%) and 53 (60.22%) were on basal bolus insulin regimen. Median duration of HCL use was 1.45 years. Mean HbA1c (mmol/mol) improved significantly from 69.77 ± 18.5 before HCL to 55.86 ± 6.49 after one year ($P 0.0008$). TIR (%) also increased significantly, from 46.49 ± 18.19 to 61.55 ± 8.71 after one year ($P 0.0005$). The proportion of patients using HCL increased from 37% (2023-2024 NPDA) to 79.3%. Black African patients made up the largest ethnic group (23.2%) in our diabetes population, with 22.8% using HCL, when compared to 9.9% in the previous year. Among patients in the lower deprivation deciles (2nd & 3rd), 83.3% and 75% were using HCL compared to 40% and 44.3%. (2023-2024 NPDA)

Conclusion

There has been a significant improvement in glycaemic control since switching to Hybrid close loop system- both in terms of HbA1c and Time in range. Additionally, proactive use of HCL has improved equity of uptake both in terms of ethnicity and deprivation index.

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P94

Smart solutions for advancing diabetes education to schools

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Aim and Objectives

To enhance and increase the capacity for diabetes knowledge in schools through the organization of a medical needs conference and the development of a series of educational video materials, thereby reducing the need for individual school visits by the Diabetes MDT.

Method

Teachers responsible for medical needs were contacted directly through the Local Authority (LA) Special Educational Needs and Disability Coordinators (SENDCO) forums and invited to a conference held in the Atrium of the Royal Free Hospital. The content included carbohydrate counting, correction dosing, the use of glucose monitors, insulin pump management, and the treatment and awareness of hypoglycaemic events. This was followed in the afternoon by hands-on demonstrations as part of a carousel of activities and a plenary session where questions could be answered by a specialist panel. Additionally, a set of videos was developed and promoted to schools, along with invitations to online drop-in sessions for further questions and to check understanding. This allowed school staff to grasp key concepts of diabetes management at their own pace and reduced travel time for the Diabetes MDT.

Results

The conference successfully trained 43 school staff, significantly increasing the number of individuals with knowledge about diabetes within schools. Feedback indicated that participants felt more confident in their ability to assist students with diabetes and found the additional contact with the Diabetes MDT beneficial. Furthermore, the video forum served as a valuable option for discussion, with schools reporting increased participation and understanding of diabetes.

Conclusion

A group-based approach to diabetes education for schools was an effective way to educate and inform a large number of schools simultaneously. By hosting a conference and providing access to video resources, we efficiently educated a larger audience, thereby enhancing the overall support system for students with diabetes. This model not only saves time and resources but also fosters a collaborative environment among schools and provides a forum to discuss best practices and shared experiences.

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Diabetes 5

P95

A patient with mauriac syndrome - a rare complication of poor compliance with T1DM management

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Introduction

Mauriac Syndrome is a rare complication of Type 1 Diabetes Mellitus (T1DM), associated with poor glycaemic control. Typical features include hepatomegaly, due to hepatic glycogenosis, hypercholesterolaemia, delayed growth and puberty and cushingoid features. Improvement in glycaemic control is associated with subsequent improvement in symptoms.

Case Report

A 16-year-old girl with T1DM was admitted with DKA and noted to have hepatomegaly and deranged LFTs (AST > 3000 U/l and ALT > 1000 U/l). She has a background of coeliac disease, hypothyroidism, learning difficulties and a complex social background. She had poor compliance with her diabetes management (HbA1c over 100 mmol/mol for two years (highest 170 mmol/mol)). Multi-professional support had not resulted in any improvement in her glycaemic control. Viral and autoimmune hepatitis screens were negative, and a liver ultrasound demonstrated a globally enlarged liver with mildly coarsened echotexture, normal echogenicity and contour and no focal lesions. Her cholesterol was raised at 11.9 mmol/l; her height had decreased from the 2nd to the 0.4th centile. A clinical diagnosis was made of Mauriac syndrome. She had a brief period of improvement in her diabetes management associated with a decrease in HbA1c to 91 mmol/mol, but this was not sustained and she had further admissions with severe DKA.

Conclusion

This case demonstrates how poor glycaemic control can lead to hepatomegaly, deranged LFTs, high cholesterol and delay in growth. A previous case report implicated a mutation in PHKG2, the catalytic subunit of the enzyme glycogen phosphorylase kinase (PhK), suggesting that poor glycaemic control may not be the only responsible factor for the development of Mauriac syndrome. The effect of chronic insulinisation, on the growth hormone-IGF-1 axis has been implicated in the pathogenesis of the growth failure. Although the more severe forms of this syndrome are rare in developed countries, the presence of hepatomegaly, subtle degrees of growth failure or the blunting of the pubertal growth spurt should alert clinicians to potential development of this rare complication.

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P96

Highlighting differences in management between paediatric and adult services for 16-19 year olds with type 1 diabetes at guys and st thomas foundation trust

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Purpose

It is well-known that adolescence is a time when glycaemic management can worsen and transitioning into adult services can compound the issue. There is currently no approved standardised transition model making practise across different hospitals variable. Our aim was to review the differences in management and outcomes of patients with T1DM aged 16-19yrs between those cared for by the paediatric and the young adult service at GSTT. Patients in this age group are vulnerable, some hospital trusts insist that patients > 16 years of age are admitted onto adult wards. GSTT is one such trust and whilst there is some crossover between paediatric and adult diabetes teams the staffing ratios and care pathways are significantly different.

Methods

Data for patients with T1DM (aged 16-19 years) for the 2024-25 NPDA submission was collected by the paediatric and adult teams for patients under their care. The data from each service was collated separately prior to combining the data for NPDA submission. Outcomes for each service were reviewed and compared.

Results

The data showed suboptimal care and glycaemic levels in both groups. 53 patients were included (18 under paediatrics services and 35 under young adult service). The table below shows differences between care processes and outcomes for these patients:

	Patients under paediatrics (n 18)	Patients under adults (n 35)
Completion of all 7 care processes	40%	3.2%
Eye screening	66.7%	29%
Insulin pump	66.7%	31.4%
Offered additional dietetic appointment	60%	29%
Average appointments/year	4.7	3.0
Average contacts/year	25	4
Median HbA1c	70.9	72

Conclusion

The paediatric diabetes team have a higher staff to patient ratio including a dedicated allocated key worker for each patient facilitating additional contacts between clinic visits. The results amplify the importance of establishing a standardised transition process for the completion of care processes and patient pathways. There is a clear need for joint working between paediatric and adult teams to enable improved outcomes for these patients across the board.

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P97

Real-world performance of the omnipod® 5 automated insulin delivery (AID) system in children and adolescents with type 1 diabetes in the united kingdom

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Background and Aims

The Omnipod® 5 AID System, which allows for personalized therapy through customisable glucose targets, is CE marked for use in individuals ages two years and older with type 1 diabetes and is commercially available in some European countries. This study aimed to evaluate real-world performance of the system in paediatric users in the United Kingdom (UK).

Methods

A retrospective analysis of continuous glucose monitoring (CGM) and insulin data from Omnipod 5 users with T1D aged 2 to <18 years in the UK whose guardian provided consent and had ≥90 days of data with sufficient CGM data (≥75% of days with ≥220 readings) available in the cloud-based data management system was conducted.

Results

Data from 14,386 users in the UK were available at the time of analysis. Preliminary results demonstrated a median time in target range (TIR; 3.9-10.0 mmol/l) of 66% (n = 7,808), 65% (n = 4,313), and 63% (n = 2,265) with use of the 6.1 mmol/l, 6.7 mmol/l, and 7.2-8.3 mmol/l targets, respectively. Time below range (TBR; <3.9 mmol/l) was low (median ≤ 1.90%) across glucose targets. Use of the lowest target (used by 54% of all users) was associated with the highest TIR with some age-related variability (2-5y: 67% [n = 263]; 6-12y: 67% [n = 3,310]; 13-17y: 64% [n = 4,235]), minimal TBR, and a high percentage of time spent in Automated Mode (median 95% across all age groups).

Conclusions

These real-world results in > 14,300 Omnipod 5 paediatric users with T1D in the UK demonstrate the substantial glycaemic benefits of AID use. Additionally, these findings highlight the importance of target glucose selection and setting adjustments to optimise outcomes for children and adolescents using the Omnipod 5 System.

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P98

Adjunctive glucose-lowering agents in children with type 1 diabetes: a systematic review and meta-analysis

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Background

Adjunctive non-insulin glucose-lowering agents may offer additional glycaemic benefits in children with established type 1 diabetes (T1D), but evidence supporting their routine use remains limited. This systematic review and meta-analysis aimed to assess their efficacy and safety in paediatric T1D management.

Methods

We searched Medline, Embase, CENTRAL, ClinicalTrials.gov, and WHO ICTRP for English-language randomised control trials from 1946 to 12 March 2024 (PROSPERO: CRD42024512564). Eligible studies included children (≤ 18 years) with T1D duration > 3 months, comparing adjunctive non-insulin glucose-lowering agents with usual care, placebo, or another glucose-lowering agent, with ≥ 12 weeks of treatment. Key efficacy outcomes included changes in HbA1c, total daily insulin dose (TDD), and safety outcomes included rates of hypoglycaemia and diabetic ketoacidosis (DKA). Data were pooled using a random-effects model.

Results

From 5384 records, 31 studies ($n = 1510$) met inclusion criteria. Meta-analysis showed a significant reduction in HbA1c (%) at 3 months with adjunctive metformin (Mean Difference [MD] -0.31, 95% CI -0.61 to -0.02), in studies with a mean age 12.8–16.9 years, receiving 1000–2000 mg/day. Single study results showed omega-3 fatty acids (MD = -0.3, 95% CI = -0.52 to -0.08), and carnosine (MD -0.90, 95% CI -1.72 to -0.08) also reduced HbA1c (%) at 3 months. Significantly reduced TDD (unit/kg/day) was seen with metformin at both 3 months (MD = -0.12, 95% CI = -0.18 to -0.06, $I^2 = 41\%$) and 6 months (MD = -0.15, 95% CI = -0.23 to -0.06, $I^2 = 42\%$). Sulphonylureas (MD = -0.21, 95% CI = -0.39 to -0.03), omega-3 fatty acids (MD = -0.10, 95% CI = -0.15, -0.05), and thiamine (MD = -0.14, 95% CI = -0.27, -0.01) also reduced TDD. No significant increase in adverse events, hypoglycaemia, or DKA was observed. However, 68% of studies had some concern or high risk of bias (Cochrane RoB2).

Conclusion

Certain adjunctive agents, such as metformin, may reduce HbA1c (%) by up to -0.31% and TDD by up to -0.21 unit/kg/day in children with T1D without increasing adverse events. However, these findings may not be considered significant for clinical practice and are limited by study quality and heterogeneity, such as missing outcome data. Further high-quality trials are needed, particularly evaluating promising newer agents, such as GLP-1 receptor agonists.

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P99

Semaglutide as an add-on treatment to optimise glycaemic control in children and young people with type 1 diabetes (smile T1D trial) development and protocol

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Background

Despite advances in insulin therapy and diabetes technology, only 12.8% of children and young people with type 1 diabetes (CYPD) in England and Wales achieved the NICE HbA1c target (≤ 48 mmol/mol) in 2022/2023. GLP-1 receptor agonists, such as liraglutide and semaglutide, have demonstrated glucose-lowering effects in adults with type 1 diabetes and are approved for use in children with obesity and type 2 diabetes.

Methods

The Smile T1D trial is a multi-centre, randomised, open-label, parallel-group, superiority trial designed to evaluate the glucose-lowering efficacy and safety of once-weekly semaglutide as an adjunct to insulin in CYPD with suboptimal glycaemic control. Eligible participants (aged 10–24 years) with type 1 diabetes (≥ 12 months), HbA1c between 58–86 mmol/mol, insulin dose ≥ 0.5 units/kg/day on any regimen, and using a continuous glucose monitor, will be randomised (3:2) to semaglutide plus insulin or insulin alone for 26 weeks, across 7 NHS sites, using a treat-to-range approach. Key exclusions include low BMI, severe hypoglycaemia, recent DKA, pancreatitis, renal impairment, proliferative retinopathy, thyroid cancer risk, gastroparesis, pregnancy, and DIY closed-loop systems. The primary outcome is change in HbA1c at 26 weeks. Other outcomes include HbA1c at earlier timepoints, time-in-range, insulin dose, BMI-SDS,

diabetes-related patient-reported outcome measures, beta cell function, and safety outcomes including hypoglycaemia, DKA and growth. A sample size of 240 will provide 90% power to detect a 5.45 mmol/mol HbA1c difference, accounting for 10% attrition, using a mixed-effects linear regression model. Analysis will follow the intention-to-treat principle.

Results

A trial-specific PPI group involving CYPD was established to review trial documents. Global semaglutide shortages since 2022 delayed approvals. Regulatory bodies (REC and MHRA) required key protocol amendments, including a 5-week post-discontinuation follow-up, monthly pregnancy testing with contraception review, an additional pregnancy information leaflet, and enhanced participant education on pancreatitis, thyroid tumour risks, and renal function-based age-specific exclusion criteria.

Conclusion

The Smile T1D trial is expected to start recruitment at end of 2025. It will provide critical evidence on the role of semaglutide in improving glycaemic control and quality of life in CYPD, potentially informing future treatment guidelines.

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P100

Improving the transition service and transfer process in Leeds

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Introduction

There is a long-established transition service in Leeds with MDT including Youth Workers and Psychologists working across Transition and Young Adult (YA) clinics. However, we have only recently been able to employ a YA DSN and have YA doctor and nurse attendance at four Joint Transition clinics per month. We want to improve the service in consultation with the young people (YP).

Aims

The goal was to standardise and improve process of transition using feedback from young people. Paediatric and adult services worked together as one transition team with monthly meetings, 18+ case management and Seamless Diabetes Transition training. This allowed development of a standardised process with individualised care provided to reduce inequalities for all in our caseload.

Methods

Regular team meetings focused on standardising care, including improving language around transition and team education sessions. Feedback was obtained from YP about the transition process (patients both pre & post transfer) using a questionnaire and youth forum. Transition documentation was updated. Participation in Seamless Diabetes Transition training supported the team to work together to navigate challenges.

Results

Questionnaire feedback was generally positive but highlighted areas for improvement including lack of flexibility with clinic appointments and challenges with clinic attendance. Youth forum qualitative feedback included improving visual impact of transition documentation, structure of YA clinic and suggestions re peer support opportunities. The transition process has now been standardised with clear expectations for both staff and patients, outlined in the transition pathway documents. The focused transition quarter has been successful with a team approach to Ready Steady Go (burden on staff eased, no longer tick box exercise). We have developed QR code education resources facilitating increased use of age-appropriate Digibete resources.

Discussion

Progress has been challenging at times, with clinical priorities taking precedence. However the process to date has supported progress, improving cohesion across the CYP and YA teams to enhance the transition experience of all patients. Moving forward we plan to compare quantitative data (HbA1c, DNA rates) from 2024 to now, to implement suggested changes including evening clinics and peer support opportunities, while continuing to collaborate with YP attending the service.

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P101

Culturally tailored digital resources for children and young adults with type 2 diabetes to promote safe medication use on young type 2 website and appDita Aswani¹, Nabil Boulos², James Yong³, Emma Bell³, Hannah Smith⁴, Maddie Julian⁵ & Heather Clark⁶¹Sheffield Children's Hospital, Sheffield, United Kingdom. ²Southampton Children's Hospital, Southampton, United Kingdom. ³Leeds Teaching Hospital Trust, Leeds, United Kingdom. ⁴University Hospital Bristol and Weston, Bristol, United Kingdom. ⁵DigiBete, Leeds, United Kingdom. ⁶Birmingham Women and Childrens, Leeds, United Kingdom**Introduction**

Childhood obesity is increasingly linked to early-onset Type 2 diabetes (T2D) in children and young adults (CYA), as shown by data from the National Paediatric Diabetes Audit (NPDA) and the National Diabetes Audit (NDA). Building on the success of the Type 2 App, DigiBete partnered with members of T2D National Working Group, National Children Young People's Diabetes Network to co-develop accessible medication resources. These support the safe use of commonly prescribed medications like Metformin and GLP-1 receptor agonists (GLP-1RA), helping users understand how their medication works, how to take it, benefits, side effects, contraindications, safety measures (including contraception), and when to contact healthcare professionals.

Methods

The platform's first version was co-designed over nine months and launched in 2023. A six-month extension in 2025 focused on expanding treatment content. Contributions came from:

1. A clinical oversight group, including a specialist pharmacist, to ensure medical accuracy and cultural relevance.
2. A multicultural user group representing the diverse CYA population living with obesity and/or T2D.
3. DigiBete's clinical safety team.

Resources were reviewed and endorsed by the Leeds Children's Diabetes Team and the T2D National Working Group.

Results

New sections were added to the platform's Treatment area for Metformin, GLP-1RA, and SGLT2 inhibitors. For each medication there is an animated explainer video and a visually engaging, easy-read PDF. Materials feature real individuals from diverse backgrounds and are fully compliant with WCAG AA accessibility standards. These resources enhance understanding and support treatment continuity by reinforcing previously advised care. These developments will feature on the app for the reassurance of instant off-line functionality.

Conclusion

The updated DigiBete platform is freely accessible to users and healthcare teams across England. Instructional animations—particularly for GLP-1RA use have greatly improved user engagement and understanding. The inclusion of the ReachDeck accessibility toolbar and multilingual translation enhances usability to support equity. Feedback via youngtype2.org reflects strong user engagement and points to the potential for developing a broader digital resource. The versatile content is also beneficial for Complications of Excess Weight (CEW) services, where similar medications are used in children without T2D, supporting education on related health issues.

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P102

Actual automode use offers superior discrimination of HbA1c outcomes compared to relative automode use in children with type 1 diabetes using AID systemsProteek Sen¹, John Pemberton¹, Zainaba Mohamed¹, Melanie Kershaw¹, Leslie Drummond¹, Renuka Dias^{1,2}, Suma Uday^{1,3}, Vrinda Saraff¹, Ruchi Nadar¹, Chamila Balagamage¹, Louise Collins¹ & Ruth Krone¹¹Birmingham Children's Hospital, Birmingham, United Kingdom. ²University of Birmingham, Department of Applied Health Research, Birmingham, United Kingdom. ³Department of Metabolism and Systems Science, University of Birmingham, Birmingham, United Kingdom**Background and Aims**

Automated Insulin Delivery (AID) systems have transformed glycaemic management in children and young people (CYP) with type 1 diabetes (T1D). Metrics such as percent sensor wear time (Sensor%) and percentage automated mode during sensor use (PAM%), may misrepresent actual automode use. This study compared Sensor%, PAM%, and actual automode percentage over a defined period (AAM%) to assess which best predicts HbA1c 6 and 12 months after AID initiation.

Materials and Methods

This retrospective cohort study included 170 CYP with T1D initiated on AID therapy between 2021 and 2024 at a UK tertiary centre. HbA1c values at 6 and 12

months were paired with corresponding 90-day CGM/pump use data. Sensor% and PAM% were taken directly from clinical reporting tools. AAM% was derived by multiplying Sensor% by PAM% and dividing by 100, i.e., (Sensor% × PAM%) / 100. Repeated-measures mixed-effects models assessed the relationship between each usage metric and HbA1c. To explore clinical utility, the proportion of individuals achieving HbA1c thresholds (≤ 48 , ≤ 52 , ≤ 58 mmol/mol) was calculated across use bins ($< 80\%$, $80-89\%$, $90-94\%$, $\geq 95\%$).

Results

Across the 340 observations, mean HbA1c was 55.3 mmol/mol (SD = 9.1), and mean age was 11.3 years (SD = 3.3). All three metrics significantly predicted HbA1c ($P < 0.001$). All three metrics explained ~80% of HbA1c variance, however, AAM% demonstrated the lowest Bayesian Information Criterion (BIC), indicating superior model parsimony (see Table). AAM% showed the clearest gradient across all HbA1c cut offs, for example, the proportion achieving HbA1c ≤ 58 mmol/mol rose from 38% at $< 80\%$ use to 83% at $\geq 95\%$ use, a greater differential than seen with Sensor% (44% to 75%) or PAM% (41% to 79%), further supporting AAM%'s stronger discriminatory power.

Conclusion

AAM% integration into clinical reporting tools may enhance patient stratification and inform AID optimisation strategies.

Table 1 Predictive strength of AID system use metrics for HbA1c: Model coefficients and fit statistics

Predictor	Beta Coefficient	95% CI Lower	95% CI Upper	Marginal R ²	Conditional R ²	BIC
Sensor%	-0.3	-0.41	-0.18	0.792	0.665	2362
PAM%	-0.34	-0.42	-0.25	0.812	0.696	2334
AAM%	-0.28	-0.35	-0.21	0.796	0.679	2330

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P103

Is advanced paediatric diabetes technology accessible to all? – a quality improvement project at the children's hospital for walesShreyaa Parasuram¹, Ambika Shetty², Rachel Harris², Alicia Girling², Erin James² & Azza Idris²¹Cardiff University School of Medicine, Cardiff, United Kingdom. ²The Noah's Ark Children's Hospital for Wales, Department of Paediatric Diabetes and Endocrinology, Cardiff, United Kingdom**Background**

Health inequalities persist in Paediatric type 1 diabetes care. The latest NPDA showed Cardiff and Vale had the lowest uptake of advanced technologies such as Hybrid Closed Loop (HCL) systems among children and young people (CYP), with 30% from deprived areas and 20% ethnic minorities.

Objectives

This project explores disparities in access to diabetes technology amongst those who are socioeconomically deprived and/or from ethnic minorities in Cardiff, identifies key barriers, and proposes QI-based interventions.

Methods

Data on CYP not using HCLs, including ethnicity and postcodes, was extracted from the database. Socioeconomic levels were stratified using deprivation indices from the Office for National Statistics. HbA1c levels were compared across these groups to assess correlations. Email surveys assessed family awareness and barriers to HCLs, with handouts and follow-up calls addressing digital literacy. MDT surveys identified perceived barriers, potential solutions, and were thematically analysed.

Results

HbA1c levels varied significantly by deprivation, with the lowest mean (64.77, $n = 30$) in the 0-13.4% range (least deprived) and the highest (86.58, $n = 12$) in the 26-56% range (most deprived). However, a weak correlation ($r = 0.29$) suggests ethnicity may play a larger role. HbA1c differences were seen across ethnicities: Arab patients had the highest mean (99), followed by mixed (91) and black (75), whilst Asian (50.5) and white (69.7) CYP had the lowest. Family surveys showed 55.6% were unfamiliar with HCLs, but 89% were open to using them once they were informed. 33% cited lack of knowledge and 16% mentioned lifestyle compatibility as concerns. Peer support groups were suggested. All MDT members observed uptake disparities, citing language, travel costs, and phone requirements as the main causes. Proposed solutions included translated materials, more MDT time, targeted group sessions, and financial aid for phones and travel.

Conclusions

Despite strong interest in HCLs, barriers such as financial and access constraints, along with a lack of awareness, hinder uptake. Tailored support and education including translated materials, poverty proofing training for the MDT, and peer support are planned to ensure that CYP living in poverty or ethnic minorities are not excluded from accessing life improving diabetes technology.

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P104**The seamless diabetes transition (SDT) programme across yorkshire and humber: enhancing transition care for young people with diabetes**Fiona Campbell¹, Tricia Woodhead¹, Emma Savage¹ & Maddie Julian²¹Leeds Teaching Hospitals, Leeds, United Kingdom. ²Digibete, Leeds, United Kingdom**Objectives**

To evaluate the effectiveness of the Seamless Diabetes Transition (SDT) Programme in supporting paediatric and adult diabetes teams across Yorkshire and Humber (UK) to deliver high-quality, person-centred transition care for young people aged 13–25 with diabetes. The programme encouraged joint working between paediatric and adult teams, with each site identifying a transition team champion to lead the project locally.

Methods:

This nine-month quality improvement programme involved multidisciplinary diabetes teams from 14 NHS trusts. It used a blended learning approach, including e-learning modules (powered by DigiBete), three virtual half-day workshops, one face-to-face event, and regular coaching and team champion calls, with support from the SDT master quality improvement trainer. Teams applied improvement techniques such as Plan-Do-Study-Act (PDSA) cycles to test and embed changes. Progress, collaboration and team support was monitored through data sharing, poster submissions, and shared learning events. Thematic analysis was applied to qualitative data, including service-user feedback and staff reflections.

Results

Key interventions included:

- Development of “transition passports”
- A motivational conversation tool
- High HbA1c care pathways
- Meet-and-greet sessions with adult teams
- Earlier initiation of transition education during paediatric care.

The programme fostered improved collaboration between paediatric and adult teams. Teams reported greater clarity around transition roles and responsibilities. The e-learning component helped establish a shared understanding of transition care, with a 91% module completion rate across participating sites. Teams identified increased confidence in delivering person-centred transition care. Challenges included initial disconnect between services, differences in team structures, and variable baseline approaches to transition.

Conclusions:

The SDT Programme provided a structured, collaborative framework for enhancing transition care across the region. It supported earlier preparation, improved inter-team communication, and empowered teams to adapt care to local and individual needs. The programme demonstrated clear potential for national scalability and long-term sustainability in delivering equitable, person-centred transition care for young people with diabetes.

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Diabetes 6**P105****Development of a national first year of care (FYOC) pathway for children and young people (CYP) with T2 diabetes (T2D) to improve care and outcomes**Nivedita Aswani¹, Heather Clark², Katie Bareford³, Elizabeth Procter⁴,Adele Swart⁵, Jennifer Simpson¹, Victoria Timmis¹ & Claire Semple⁶¹Sheffield Children's Foundation Trust, Sheffield, United Kingdom. ²West Midlands CYPDN Network Manager, Birmingham, United Kingdom.³Diabetes UK, London, United Kingdom. ⁴Nottingham University Hospitals NHS Foundation Trust, Nottingham, United Kingdom. ⁵Lewisham and Greenwich NHS Trust, Lewisham, United Kingdom. ⁶University Hospitals Bristol and Weston NHS FT, Bristol, United Kingdom

There is a National drive to improve care and outcomes for CYP with T2D in recognition of the aggressive nature of early onset T2D¹ and an 88% increase in numbers receiving care in paediatric services from 2019/20 to 2023/24². The NHSE Diabetes Programme, Core20Plus5 and National CYP Diabetes Network (NCYPDN) priorities are in alignment and have sought to strengthen healthcare professional competencies in managing T2D, bring parity of care with T1D, and reduce health inequalities. A task and finish group from NCYPDN T2D produced a FYOC Pathway to assist Paediatric Diabetes Units (PDUs) to implement holistic support for CYP in line with ACDC³ and NICE⁴ guidance. This comprehensive framework promotes a mindset shift utilising admission at diagnosis to convey severity of T2D in CYP to families, complete timely screening, understand social

context and vulnerability factors, formulate a culturally tailored plan and identify risk to signpost family members. A frequent outpatient contact schedule is proposed, adaptable to local resources and circumstances. Intensive support in the initial stages is key with focus on dietetic and psychology input to encourage remission and engagement. The Pathway includes supporting evidence and rationale. It links to toolkits on obesity, use of appropriate language and understanding barriers. Development was informed by the NCYPDN T1 FYOC Pathway, existing T2 guidelines developed by other PDUs during 2022-24, and Sheffield Children's Hospital pilot FYOC T2 Pathway and 2 year audit results. NCYPDN members were consulted on key components via a T2 Working Group Survey, and a separate live Study Day poll (over 350 delegates). Respondents endorsed the value of admission at diagnosis and CGM for education. The Pathway is deliberately aspirational as a catalyst for quality improvement given poor essential care process completion and outcomes, and to be a lever with commissioners. It will be reviewed in 2027 incorporating learning from the NHSE Complication from Excess Weight pilots, PDU rollout and other T2D service development initiatives.

1. Long-Term Complications in Youth-Onset Type 2 Diabetes. TODAY study Group. N Engl J Med 2021;385:416-426.

2. NPDA T2 diabetes Spotlight report 2023/24.

3. ACDC T2 guidelines 2023.

4. NICE NG18

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P106**STID - sport and T1D: a pilot clinic**Edel Magee¹, Paul McMullan² & Sarinda Millar¹¹SHSCT, Newry, United Kingdom; ²SEHSCT, Belfast, United Kingdom**Aim**

T1D mainly affects children and young people (CYP), however adults can develop it as well. NICE, (2023) recommend that CYP living with T1D should have access to four clinic appointments a year with their multidisciplinary team (MDT). Due to advanced technology and the subsequent data from this it can be challenging to fully address areas which are most important to the young person at clinic. The aim of this STID clinic pilot was to obtain baseline information on those living with T1D and participating in sports at club level in the Southern Health & Social Care Trust (SHSCT), Northern Ireland (NI) and offer individualised advice.

Methods

Participants 24 CYP were invited to attend, from March 2023 – August 2024. The clinic was held every 3 months, data was captured in a longitudinal approach. CYP were screened prior to attending to meet the inclusion criteria, attending regular diabetes clinic appointments and participating in physical activity at club level.

Procedure Referrals were accepted from the extended diabetes MDT in the SHSCT. An appointment was scheduled as one of their four regular appointments, to avoid appointment burden associated with a long term condition. The appointments were face-face or virtual (telephone or video encounter). A proforma was used to obtain baseline information to inform the advice provided.

Results

The sports played by the CYP were, gaelic football, soccer, rugby, hockey, swimming, and climbing, with the most common sport played by both female and male populations was gaelic football, 60% and 64% respectively.

Conclusions

Due to the ever growing abundance of available data with the technological advances, it is important to remember that the everyday challenges of T1D are not eradicated. Therefore, this pilot has demonstrated the need for individualising sports treatment plans specific to the CYP interests.

Table 1 Characteristics of CYP (n = 19)

Characteristics	Female (n = 5)	Male (n = 14)
Age (years)	13.2	15.6
Multiple daily injections	2	6
CSII	3	8
Blood glucose monitoring		
Blood glucose meter		1
CGM	5	13

NOTE: Age is expressed as mean, CSII = Continuous subcutaneous insulin infusion, CGM = Continuous glucose monitor

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P107**Improving care for children and young people (CYP) with type 2 diabetes: a guideline evaluation**

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Background

Children and young people (CYP) with youth-onset type 2 diabetes mellitus (T2DM) are at high risk of developing early complications, leading to increased morbidity and mortality during their most productive years. Due to limited paediatric-specific research, national guidelines aim to standardise care, though implementation and outcomes remain variable. We evaluated local adherence to the paediatric T2DM guideline and assessed changes in clinical outcomes before and after implementation.

Methods

15 CYP with T2DM were included in a retrospective audit covering two time periods: pre-guideline (April 2023–March 2024) and post-guideline implementation (April 2024–March 2025). Data were extracted from electronic records (Patient Centre, LEPRES, EMIS) and laboratory systems (ICE). Clinical outcomes, guideline adherence, and comorbidity prevalence were compared using Microsoft Excel for analysis.

Results

All patients diagnosed after January 2024 were admitted as per guideline recommendations, except one due to mental health concerns. HbA1c monitoring improved from 20% (2/10) pre-guideline to 53.3% (8/15) post-guideline. Of the 8 patients with regular HbA1c monitoring, 5 showed a reduction in mean HbA1c. Patients diagnosed after guideline implementation had more sustained improvements in glycaemic control. Two patients with HbA1c >69.4 mmol/mol were commenced on combination therapy. All patients received glucose monitoring equipment and were offered multidisciplinary team (MDT) input at diagnosis and annually; 87% (13/15) engaged. Annual psychological screening was completed in 13/15 patients, with 5 receiving targeted psychological support. Weight targets were not achieved in any patient; however, 47% (7/15) achieved a BMI reduction (mean -0.84), and 47% had a Z-score reduction (mean -0.14). Comorbidities included non-alcoholic fatty liver disease (53%) and dyslipidaemia (47%). Two patients developed new complications during the post-guideline period despite having no complications at diagnosis.

Conclusions

Implementation of a structured paediatric T2DM guideline resulted in improved monitoring and glycaemic outcomes. Positive trends in BMI and psychological engagement were observed. These findings support the ongoing use of standardised pathways and MDT input to enhance outcomes in CYP with T2DM, though challenges in weight management and complication prevention remain.

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P108**Psychosocial factors are associated with poor glycaemic control in a type 1 diabetes mellitus (T1DM) paediatric cohort**Amelia Prentice¹ & Evagelia Paraskevopoulou²¹City St George's, University of London, London, United Kingdom; ²St George's University Hospitals NHS Foundations Trust, London, United Kingdom**Background & Aims**

While national audits show improving glycaemic control in paediatric T1DM many children, particularly those from socially vulnerable backgrounds, still fail to meet recommended targets. This project aimed to identify psychosocial factors associated with poor glycaemic control and reduced healthcare engagement.

Methodology

A retrospective review was conducted of 60 paediatric T1DM patients in a south London tertiary centre. Patients were assessed using multiple variables including age, gender, HbA1c, BMI, hospital admissions, missed appointments, safeguarding involvement, interpreter need, and psychological support referrals.

Results

58% of boys and 63% of girls had HbA1c levels above the national target, with a cohort average of 67 mmol/mol, exceeding the 60 mmol/mol reported by the 2022/23 NPDA audit. Children referred to social services experienced the highest rates of hospital admissions and DNAs, followed by those requiring interpreter or psychological support. These findings suggest that both external (social and

language barriers) and internal (mental health) factors contribute to reduced engagement and poorer outcomes. Moreover, children from Black, Asian and Minority Ethnic (BAME) backgrounds had higher admission rates and missed appointments than White British peers, echoing national trends in ethnic healthcare disparities.

Conclusion

This evaluation highlights the complex interplay between psychosocial vulnerability and poor glycaemic outcomes in paediatric T1DM. Social instability, mental health needs, language barriers, and ethnicity appear to significantly affect disease management and healthcare engagement. These findings reinforce the importance of early holistic, culturally sensitive care and ensuring there are integrated psychosocial services in diabetes teams. They also raise awareness to diabetes teams of the importance of addressing health inequalities to minority ethnic groups and those from areas of social deprivation. Reports have shown the role of family support and youth workers in this patient population is vital in bridging these gaps and disparities. Ensuring this is addressed before transition may reduce complications, DNAs, and long-term strain on adult diabetes services. Future work will include statistical analysis and expanding the sample size to further validate these findings and create statistical inference.

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P109**Improving out-of-hours troubleshooting in paediatric type 1 diabetes: a quality improvement initiative**Nway Nway Htun Kyi¹, Grace Muinde², Su Sandar², Simon Fountain-Polley² & Swe Lynn²¹Health Education and Improvement Wales, Cardiff, United Kingdom;²Hywel Dda University Health Board, Carmarthen, United Kingdom**Introduction**

Type 1 diabetes (T1D) is an increasingly prevalent long-term condition among children and young people (CYP), affecting 32,809 individuals in England and Wales (NPDA 2023–24). As diabetes technologies evolve, providing safe and consistent out-of-hours (OOH) advice requires specific expertise—particularly when the specialist diabetes team is unavailable. The Association of Children's Diabetes Clinicians (ACDC) recommends that OOH advice be delivered by trained staff using standardised protocols, with regular service evaluation. Locally, families reported variability in the OOH advice quality, reflecting inconsistent knowledge among the acute paediatric team.

Aim

To improve the knowledge and confidence of the on-call paediatric team in delivering safe, consistent OOH advice to families of CYP with T1D.

Method

A baseline survey was conducted among resident paediatric doctors and senior nurses at a district general hospital to assess current practices and confidence in managing seven common OOH T1D scenarios. These included hypoglycaemia, hyperglycaemia, and intercurrent illness in CYP using multiple daily injections (MDI), insulin pumps, or hybrid closed-loop systems (HCLS), as well as ketone interpretation. Confidence was rated on a 5-point Likert scale.

Results

Twenty-one staff completed the pre-intervention survey. Most preferred locally agreed printed algorithms, training sessions, and digital resources. Confidence scores ranged from 2.43 to 3.43, with the lowest reported in managing hyperglycaemia on pumps/HCLS (2.43) and hypoglycaemia on pumps (2.76). In response, seven scenario-specific troubleshooting algorithms were developed by the paediatric diabetes team. Laminated copies were placed in key clinical areas, with digital versions accessible via mobile devices. A local diabetes website is in development. An interactive regional study day with workshops and case discussions is planned for paediatric trainees across Wales. A post-implementation survey was conducted following the rollout of both paper and digital formats, with ongoing Plan-Do-Study-Act (PDSA) cycles. Post-intervention, confidence improved across all domains, particularly in managing insulin pump-related issues and ketone interpretation.

Conclusion

Structured, accessible algorithms enhanced staff confidence in managing OOH T1D scenarios. This initiative supports safer, more consistent care—particularly for complex technology-assisted cases—with further improvements expected through training and digital resources. The team is happy to share the algorithms at a national level.

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P110**Audit of a multidisciplinary annual review clinic for children and young people with diabetes mellitus**

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Background

NICE guidance recommends that children and young people (CYP) with type 1 diabetes (T1D) have structured, annual reviews (ARs), including checks of glycaemic control and growth, and screening for comorbidities and complications. At Cambridge University Hospitals the paediatric diabetes AR clinic for CYP aged < 16 years runs weekly, supported by a multidisciplinary team (MDT) of doctors and diabetes educators, with input from dietitians and clinical psychologists as required. In 2023, a quality improvement project was initiated to optimise adherence to screening guidelines, standardise the MDT approach, and improve CYP/carers and staff satisfaction with the clinic.

Objectives

This audit aimed to evaluate and improve: adherence to NICE guideline NG18; compliance with departmental standards, including timing of reviews; MDT member satisfaction; and CYP and family understanding of, and satisfaction with, the AR clinic.

Methods

Changes introduced in 2023 included: standardising clinic administration; introducing templates and checklists to cover all recommended AR assessments and improve consultation structure; involving clinical psychologists in the clinic; increasing staff awareness of dietetic referral criteria; and providing written information to CYP and families. Data sources comprised the National Paediatric Diabetes Audit, retrospective reviews of CYP's medical notes, an online staff survey, and a CYP/carer survey.

Results

Between 2023 and 2025, significant improvements were seen in the proportion of CYP having foot assessments (78% to 93%), lipid measurements (78% to 91%), psychology questionnaire (PI-ED) completion (74% to 100%), and smoking status documentation (73% to 99%). However, recording of retinal screening results worsened (87% to 66%) and measurement of albumin-creatinine ratio was persistently low (76-77%). In 2025 there was higher staff satisfaction regarding clinic structure, appointment length, understanding of MDT roles, and awareness of referral pathways for dietetic and psychological services. Data from the CYP/carer survey are awaited.

Conclusion

This local audit demonstrates that targeted quality improvement strategies - particularly the use of templates/checklists, enhanced MDT collaboration, and standardised processes - can improve adherence to national and departmental standards, staff satisfaction, and clinical assessments in a paediatric diabetes AR clinic. Ongoing evaluation, including CYP/carer feedback, will be key to guiding further improvements and ensuring long-term sustainability.

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P111**Enhancing HbA1c outcomes through integrated collaboration within and between diabetes MDTs and families**

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Aim and Objectives

As a Diabetes MDT, we believe the characteristics of a functional team include shared goals, clearly defined roles and responsibilities, open and effective communication, mutual respect and trust, collaborative problem-solving, flexibility, shared accountability, a focus on outcomes, and, very importantly, a joy in work. Here we outline approaches designed to foster these characteristics within our MDT and in our relationships with families, as a means of improving HbA1c outcomes.

Method

To achieve these objectives, we cultivated a culture of collaboration, within the MDT and with families, through social events and strengthened interpersonal relationships.

Social initiatives included:

- Regular Pizza Parties
- An after-school scriptwriting course run by *Read for Good*
- A targeted HbA1c intervention for teenagers
- Promotion of the YES Programme
- Funded attendance to a family diabetes camp

Internally, we focused on being warm and approachable, with each other and with families, demonstrating empathy, building alliances, and advocating for children and families in matters of social care and education. We actively uphold the principles of the *Language Matters* initiative, striving to remain friendly, accessible and person-centred.

We also prioritised:

- Acting on feedback from newly diagnosed young people at discharge
- Delivering weekly best practice training to non-specialist nursing staff
- Holding regular MDT lunches to strengthen internal cohesion
- Respecting the wishes and preferences of families, regardless of presentation
- Comprehensive MDT education

Results

Despite serving a population with one of the highest deprivation indices, NPDA data shows our setting achieves some of the lowest HbA1c outcomes nationally. Families appreciated opportunities to connect with others facing similar challenges, sharing experiences and strategies for diabetes management. The informal nature of our sessions helped normalise diabetes discussions and provided a space for incidental clinical advice from diabetes consultants, dietitians, psychologists, and specialist nurses in a non-clinical setting.

Conclusion

Strengthening therapeutic relationships is essential to effective diabetes care in children and young people. Our initiatives created a supportive network that not only contributed to improved HbA1c outcomes but also enhanced the overall experience of diabetes management for families and the MDT. Trust, empathy, and open communication proved vital in fostering a collaborative, family-centred approach to care.

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P112**How is paediatric type 1 diabetes treatment and glucose control impacted by safeguarding concerns?**

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Background

Approximately 10% of children and young people (CYP) referred to child protection services for medical neglect in America have Type 1 Diabetes (T1DM). Little published evidence exists comparing glucose control in CYP with safeguarding concerns to those without, therefore guidance on management is lacking.

Aims

To assess glucose control in CYP with T1DM and safeguarding concerns.

Methods

Data from electronic records of paediatric patients with T1DM at a large London hospital between Dec 2021 and 2022 were analysed. HbA1c readings, diabetes-related admissions, insulin administration and glucose monitoring methods, appointments and postcodes were recorded. Those with concerns were categorised into Child Protection Plan (CPP), Child in Need, Early Help and 'local concern', allowing further comparison. The same cohort was analysed in June 2024 after changes in hybrid closed-loop system eligibility.

Results

Of 155 active patients, 18 had documented safeguarding concerns (11.6%). This group had significantly higher HbA1c readings (10.0% vs 8.3% [$P < .001$]) and significantly more admissions and incidents of diabetic ketoacidosis. They had more out-of-clinic contact attempts and higher outpatient non-attendance rates. Children on CPPs had the highest HbA1c results. 6% (1/18) of the safeguarding group had an insulin pump with continuous glucose monitoring (CGM) compared to 27% (38/142) of the remaining patients. Those with insulin pumps and/or CGM had significantly lower HbA1c readings. 1 more patient in the safeguarding group had a hybrid closed-loop system following changes in eligibility.

Conclusions

CYP with T1DM and safeguarding concerns have significantly worse glucose control and reduced access to insulin pumps and CGM. These patients require frequent contact attempts with families, schools, GPs and social workers to facilitate engagement. All units should have a register of those with concerns to be discussed regularly within the multidisciplinary team. Individual management plans and out-of-hospital appointments may enable better engagement. These CYP should be enabled to use hybrid closed-loop systems safely. Successful

management strategies should be shared between centres and safeguarding data shared with the national paediatric diabetes audit. A separate medical neglect category for child safeguarding, as in other countries, may enhance support from social services with targeted interventions for those with chronic illnesses.

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P113

Improving care for children at risk of steroid-induced hyperglycaemia: integrating proactive monitoring into routine hospital care

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Background

Steroid-induced hyperglycaemia (SIH) is a recognised but under-monitored complication in children receiving high-dose corticosteroids for oncological, nephrological, or immunosuppressive indications. Delayed recognition can result in significant metabolic deterioration, prolonged admission, and missed opportunities for early intervention, particularly in patients without pre-existing diabetes.

Aim

To identify system-level barriers to early detection and management of SIH in paediatric inpatients and design a practical pathway to embed proactive glucose monitoring into routine hospital care.

Methods

A retrospective review was conducted between November 2024 and February 2025 at Leeds Children's Hospital. Seven cases of SIH (defined as ≥ 14 mmol/l post-steroid initiation) were identified in children under the care of oncology, nephrology, and hepatology teams. Audit data were triangulated with multi-disciplinary feedback to develop a targeted quality improvement framework.

Conclusion

SIH in paediatrics is frequently under-recognised outside of diabetes-specialist settings. This project highlights a scalable, MDT-driven approach to improve early identification, family communication, and escalation of care. A re-audit is planned to assess the impact of implemented interventions. Wider adoption of proactive SIH monitoring can enhance patient safety and align care with paediatric endocrinology best practice.

Table : Key Barriers & Proposed Interventions in SIH Care:

Identified Barrier	Intervention
Limited awareness of SIH risk and monitoring protocols among non-diabetes teams	MDT education, ward posters, CGM teaching sessions
No standardised local standard operating procedure (SOP) for SIH detection & referral	Development of local SIH SOP with glucose thresholds
Inadequate family awareness and engagement	Tailored SIH leaflets + diabetes specialised nurse (DSN) involvement at steroid initiation
Delayed diabetes team escalation in borderline cases	Proposed EHR-based automatic referral triggers (e.g. > 14 mmol/l) for at risk patients.

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P114

Factors contributing to late diagnosis of patients with type 1 diabetes mellitus: a retrospective audit at bedford hospital

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Background

Late diagnosis of type 1 diabetes mellitus (T1DM) in children can lead to diabetic ketoacidosis (DKA), a potentially life-threatening complication. Early identification and intervention are essential for improved outcomes.

Aims

To identify the key factors contributing to delayed diagnosis and DKA presentation in children diagnosed with T1DM at Bedford Hospital between 2021 and 2024.

Methods

A retrospective audit of 84 children (aged 1–15 years) diagnosed with T1DM was conducted. Demographic data, GP visit patterns, initial symptoms, investigations, family history, and clinical presentation at diagnosis were analyzed.

Results

- 38.1% had no GP visit prior to diagnosis; 53.6% had one G.P visit.
- Only 14.3% underwent both blood and urine testing at GP level.

- 3.3% had G.P visit with symptoms but unfortunately the symptoms were missed, and no investigations done and all of them presented in DKA
- Common presenting symptoms included polyuria (6%), polydipsia (4.8%), and weight loss (3.6%).
- At diagnosis, 63.1% had ketones (>1.5 mmol/l), and 53.6% had pH <7.34 .
- HbA1c >120 mmol/mol was observed in 33.3% of cases.
- 44% required IV insulin therapy; 79.8% were hospitalized for 1–3 days.
- 89.3% had no complications; 2.4% required ITU admission.

Conclusion

A significant proportion of children presented with DKA due to missed or delayed diagnosis. Lack of GP visits, minimal investigations, and limited symptom recognition contributed to late identification.

Learning Points:

- Increased GP awareness and early glucose/urine testing are crucial.
- Family education can lead to earlier presentation.
- Standardized pathways for early recognition are needed.

Future Plans:

- Develop local guidelines for prompt testing in symptomatic children.
- Implement educational initiatives for primary care providers.
- Re-audit in 12 months post-intervention to assess impact.

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Gonadal, DSD and Reproduction

P115

Blood chimerism in asymptomatic co twins: two paediatric cases highlighting genotype-phenotype discordance

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Background

Chimerism, the coexistence of two genetically distinct cell lines within one individual, is likely under-recognised in paediatric endocrinology because low-level donor lines may be blood limited and clinically silent. Discordant genotype and phenotype, especially in twins sharing a placenta, should prompt multi-tissue testing.

Case 1:

We present monochorionic monozygotic IVF twin sisters, now aged 12 years. Twin 1 had antenatal cystic hygroma (resolved), neonatal bilateral pedal lymphoedema and strawberry naevus. Peripheral blood karyotype showed mosaic Turner syndrome [TS] (XX/XO). She has mild TS stigmata (dysmorphic features, conductive hearing loss, visual impairment), autoimmune hypothyroidism (levothyroxine 25 mg), and short stature treated with growth hormone from 3y11m when height -3.07 SDS; now -1.44 SDS. Gonadotropins confirm primary ovarian insufficiency: LH 15 IU/l, FSH 96.6 IU/l, oestradiol <100 pmol/l, AMH <0.1 pmol/l. She is due to commence pubertal induction with oestradiol. Her co twin's neonatal blood also reported mosaic TS. This was performed as her twin was affected. However, Twin 2 does not have clinical features; her height SDS is -0.42 without growth hormone treatment; she has normal thyroid function; AMH 26.2 pmol/l (ref 3–46.6), LH <0.1 IU/l, FSH 2.3 IU/l, oestradiol <100 pmol/l. In view of the difference in phenotype compared to her genotype, buccal swabs were performed for both twins. Buccal swabs for Quantitative Fluorescence-PCR have confirmed XO in twin 1 and XX in twin 2. **Case 2:** A 16-month-old reared female was referred to the DSD service following an incidental blood XX/XY finding (indication unknown). Examination showed normal female external genitalia. Buccal swab demonstrated predominantly XX cells with low-level XY, thought to be secondary to lymphocytes within her saliva. Pregnancy was monochorionic diamniotic; male co-twin sadly died in utero at 18 weeks gestation.

Conclusion

We describe two children in whom a blood-restricted chimeric line derived from a co-twin likely explains genotype-phenotype discordance (mosaic TS in an asymptomatic twin; XX/XY blood line in a phenotypic girl). Shared placentation and early twin-to-twin haematogenous exchange are plausible mechanisms. Consider chimerism and multi-site sampling when cytogenetic results do not match phenotype.

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P116**Anthropometric benchmarks of care in congenital adrenal hyperplasia (ABC-CAH): initial results from an international, multi-centre registry benchmarking exercise**Joseph McElvaney^{1,2}, Salma R Ali^{1,2}, Malika Alimussina^{1,2}, Justin H Davies^{3,4}, Aneta Gawlik-Starzyk⁵ & ABC-CAH Study Group⁶¹Developmental Endocrinology Research Group, Royal Hospital for Children, University of Glasgow, Glasgow, United Kingdom; ²Office for Rare Conditions Registries, University of Glasgow, Glasgow, United Kingdom; ³Paediatric Endocrinology, Regional Centre for Paediatric Endocrinology, Southampton, United Kingdom; ⁴Faculty of Medicine, University of Southampton, Southampton, United Kingdom; ⁵Department of Pediatrics and Pediatric Endocrinology, Medical University of Silesia, Katowice, Poland; ⁶I-CAH Registry, Glasgow, United Kingdom**Background**

Impaired final adult height and excess body weight associated with 21-hydroxylase deficiency congenital adrenal hyperplasia (21-OHD CAH) have been linked to unfavourable quality of life outcomes. This study aims to establish international anthropometry benchmarks for 21-OHD children using real-world data existing in the I-CAH registry.

Methods

All 21-OHD cases in the I-CAH registry with height and weight data collected from birth to 18.5yrs were included. One clinic visit per year of life was selected for each case by proximity to birth date and country specific reference data were used when available to calculate standard deviation scores (SDS). For this initial analysis, multifactorial analysis was undertaken describing medians and ranges by year of life, sex, growth stage.

Results

19,322 longitudinal visits between 1969 – 2025 from 1,405 21-OHD cases across 26 countries in 5 continents were extracted and of these 17,480 visits from 1,324 cases were included with a median number of visits per case of 10 (1, 66). From this, 6,175 single visits per year of life from 1021 cases were selected for preliminary analysis with a median of 8 visits per case (1, 18) at a median age of 7yrs (1, 18). Median height SDS was -0.9 (-4.0, 3.6) in the 1-3yrs age band (n, 714), 0.4 (-3.6, 4.0) in the 7-9yrs age band (n, 524) and -0.9 (-3.9, 2.0) in the 16-18yrs age band (n, 235). Median BMI SDS for 4-6yrs (n, 605), 7-9yrs (n, 525) and 16-18yrs (n, 235) was 0.6 (-3.5, 3.9), 0.8 (-3.1, 3.9) and 0.6 (-3.1, 3.8) respectively.

Conclusions

I-CAH is a rich source of real-world anthropometry data and initial analysis shows that patterns of growth in this large international cohort are similar to other reports of smaller number of cases. Further analysis will explore temporal and geographical variation and establish anthropometric benchmarks that can lead to the generation of centre-specific reports.

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P117**Puberty induction with gonadotropin treatment in males with hypogonadotropic hypogonadism: the ping study**Rachel Varughese¹, Elli Enayat¹, Annastazia Learoyd¹, Amrita Ahluwalia¹, Channa N Jayasena², Angeliki Bogosian³, Neil Smith⁴, Mehul Dattani⁵ & Sasha Howard¹¹Queen Mary University London, London, United Kingdom; ²Imperial College London, London, United Kingdom; ³City University, London, United Kingdom; ⁴Imperial College London, London, United Kingdom; ⁵UCL Great Ormond Street Institute of Child Health, London, United Kingdom**Background**

Males with congenital or acquired hypogonadotropic hypogonadism (HH) or other central disorders of the hypothalamic-pituitary-gonadal (HPG) axis experience absent or delayed puberty due to gonadotropin deficiency (GD). This condition disrupts key developmental windows, including mini-puberty and adolescent puberty, resulting in underdeveloped testes, impaired fertility potential, and significant psychosocial burden. Standard testosterone therapy, while effective for virilisation, does not stimulate testicular growth or spermatogenesis, making it insufficient for long-term reproductive outcomes.

The PinG Study

The PinG study is an NIHR-funded, UK multicentre, open-label, randomised controlled trial to evaluate efficacy of gonadotropin regimens for pubertal induction in males aged 12–35 years with GD. Participants are stratified into partial (maximal testes volume $\geq 4\text{mL}$) or severe ($<4\text{mL}$) disease. All arms receive gonadotropins, and treatment duration ranges from 18 to 24 months, with follow-up continuing until the conclusion of therapy. PinG aims to determine

whether combination gonadotropin therapy offers superior outcomes to hCG alone in partial GD, and whether rFSH pre-treatment benefits those with severe GD. This trial builds on findings from our meta-analysis of over 100 studies, which confirmed the efficacy of gonadotropins in promoting testicular development and spermatogenesis in young males. The primary outcome is the proportion of participants achieving spermatogenesis. Secondary outcome measures include hormonal biomarkers, testicular volume, time to spermatogenesis and maximum sperm concentration, quality of life and pathogenic genetic variant identification via whole genome sequencing. The trial design reflects current clinical practice but adds structured monitoring and broader outcome assessments. A nested qualitative study will be conducted to deepen understanding of the lived experience of GD. Participants will take part in two semi-structured interviews: one before starting treatment to explore the impact of delayed puberty on self-esteem, relationships, diagnosis, and expectations; and one after treatment to explore individual's perceptions of change in physical and psychological wellbeing. This component aims to capture patient priorities and inform patient-centred care. Study has initial approvals: NIHR CPMS 58941.

Conclusion

By informing optimal pubertal induction strategies, PinG has potential to improve quality of life and also reproductive outcomes for males with GD and contribute to the standardisation of gonadotropin-based pubertal induction in adolescence.

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P118**An audit of clinical practice for girls with turner syndrome in nhs lothian, scotland**Julia Hill¹, Tarini Chetty² & Kathryn Cox²¹University of Edinburgh, Edinburgh, United Kingdom; ²Royal Hospital for Children & Young People, Edinburgh, United Kingdom**Introduction**

Turner syndrome (TS) is a chromosomal disorder affecting females caused by complete or partial deletion of an X chromosome. TS affects multiple systems through all stages of life requiring multidisciplinary care. Our objective was to evaluate the current management of TS in Lothian (Scotland) against consensus clinical practice guidelines.

Methods

Individuals with TS under the care of Paediatric Endocrinology in Lothian were identified. Data were obtained retrospectively from electronic patient records.

Results

Our cohort consisted of 29 patients; median age 13.99y. 10 (34.5%) were diagnosed antenatally. The median age of postnatal diagnosis was 3y (SD 4.17y). Karyotypes for the cohort were 14 mosaic, 12 45XO, 2 Ring-X, 1 isochromosome. 28 patients attend a dedicated TS clinic; 1 patient is seen in a general endocrine clinic. Of 19 patients of pubertal age, 7 (36.8%) achieved spontaneous menarche, of which one patient presented with POI at the age of 13.4y. No individuals with 45XO karyotype achieved spontaneous menarche. 12 individuals (63.2%) required pubertal induction, all receiving transdermal oestrogen, at a median age of 11.44y (SD 0.5y). 24 (82.8%) received growth hormone with median treatment starting age 6.08y (SD 3.2y). The mean height SDS for the cohort was -1.32 (SD 0.81). Median height SDS for patients not treated with GH was 0.203. 25 (86%) have had a documented blood pressure measurement in the last 12 months. All individuals had cardiac imaging. Normal imaging in 16 of 29 (55.2%). 13 (44.8%) had some form of congenital heart disease: 5 (17.2%) bicuspid aortic valve. In the last 12 months, 24 (82.8%) have had their TFTs checked, 22 (75.9%) HbA1c and 21 (72.4%) IGF1. In the last 24m, 11 (37.9%) have had their anti-TTG checked.

Conclusion

Management of individuals with Turner Syndrome in Lothian displayed good adherence to international guidelines, particularly in the use of hormone replacement and monitoring for comorbidities. Variability in age of diagnosis and treatment initiation suggests opportunities for earlier identification and intervention. We intend to develop a checklist to ensure care aligns with the 2023 International Clinical Practice Guidelines.

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P119**A service evaluation of the current management of children and adolescents with klinefelter syndrome across south and west wales**
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Introduction

Klinefelter syndrome (KS) is the most common chromosomal abnormality in males, typically presenting with hypogonadism, tall stature, and learning or behavioural difficulties. Despite affecting 1 in 600 males, KS remains significantly underdiagnosed due to its variable presentation, particularly in milder forms such as mosaicism. Diagnosis requires genetic testing, and management primarily involves testosterone replacement therapy (TRT) and fertility support. Early diagnosis and holistic care addressing both physical and neurodevelopmental symptoms are crucial for improving long-term outcomes.

Aims

- To investigate the diagnosis and management of KS including clinical, genetic, and biochemical tools, timing of diagnosis, and current practices in South and West Wales.
- To evaluate hormone monitoring and treatment strategies.

Methods

A retrospective review of 46 patients with KS was conducted using departmental databases. Clinical data, endocrine parameters, TRT initiation, and fertility discussions were extracted from records and analysed using Microsoft Excel.

Results

Of the 46 patients reviewed, 28% were diagnosed antenatally, primarily via amniocentesis. Most postnatal diagnoses occurred before age 5 due to learning, developmental, or behavioural difficulties. Only 11% were diagnosed during adolescence, despite this being a typical time for KS presentation. 56% of patients received TRT, typically starting at a mean age of 15.6 years. Biochemical hypogonadism was the most common indication, with most patients showing elevated FSH/LH and low testosterone levels. 10 patients were started on TRT for a combination of clinical and biochemical hypogonadism, while 2 patients were started on TRT for only clinical hypogonadism. Fertility discussions were documented in 67% of cases, but only 25% were referred to fertility services, highlighting a gap in care pathways.

Conclusions

KS is often underdiagnosed due to its varied presentation; early diagnosis, timely TRT, and further research are vital to improve outcomes and support, particularly around fertility and long-term care. Early KS care can be provided by increasing clinical awareness and routine genetic testing in boys with developmental concerns. Standardised, multidisciplinary care pathways should guide hormone monitoring, timely initiation of TRT, and structured transitions to adult care. Psychosocial support, fertility counselling, and patient education are essential.

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P120

Beyond the obvious: WT1 mutation in a newborn with ambiguous genitalia

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Background

Disorders of sex development (DSDs), or atypical genitalia in newborns, are heterogeneous in origin and require early, ongoing involvement of a specialist multidisciplinary team (MDT) to ensure timely diagnosis and management.

Case Summary

We present the case of a term infant born with perineal hypospadias and bilateral cryptorchidism. Initial investigations revealed a 46, XY karyotype, normal testosterone (8.04 nmol/l), and undetectable 17-hydroxyprogesterone (<1.6 nmol/l), thereby reducing the likelihood of any underlying endocrine abnormalities. Parents were counselled that genital surgery would be required in stages, but no other major concerns were expected. Over the following months, he developed persistent vomiting, faltering growth, and periorbital puffiness in the morning. These symptoms were initially attributed to gastro oesophageal reflux, cow's milk protein allergy, and nasolacrimal duct obstruction. At 5 months, he presented with generalised oedema and anuria and further investigations revealed end-stage kidney disease. Genetic testing identified a heterozygous pathogenic mutation in the *WT1* gene, confirming a *WT1*-related disorder and congenital nephrotic syndrome. Management involved the commencement of peritoneal dialysis, plans for bilateral nephrectomy and renal transplantation, alongside surgical correction of hypospadias and cryptorchidism. Surveillance for Wilms tumour was also initiated in line with protocols for *WT1*-related conditions. The diagnosis was made only after clinical features suggestive of renal failure became

evident. In retrospect, no urine dipstick test was performed at birth, and a raised creatinine was "attributed to maternal levels". There was also a delay in communicating the genetic result, which had been requested earlier.

Discussion

This case reinforces the importance of considering broader differential diagnosis in newborns with atypical genitalia with 46,XY DSD. A thorough and systematic evaluation of infants with atypical genitalia is essential to avoid missed diagnoses that may have significant consequences. Early genetic assessment plays a key role in guiding appropriate management.

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P121

Outcomes of surgery in females with congenital adrenal hyperplasia - a scoping review of the literature

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Background

Surgery in women with Congenital Adrenal Hyperplasia (CAH) is one of the most challenging and controversial aspects of the management, having complex ethical implications and a significant impact on patients' well-being. Our aim was to obtain an overview of the published evidence on the outcomes of surgery in women with CAH, to identify unmet needs in research and clinical care.

Methods

A systematic search of PubMed and Ovid MEDLINE was conducted for studies published between 1994 - 2024. Search terms combined "females" with "CAH", "feminising genital surgeries", and "health outcomes". Search results were stored using the Rayyan platform and screened by two independent reviewers; disagreements were resolved through discussions and by a third reviewer. A thematic analysis of the selected articles was conducted.

Results

The search yielded 394 articles, of which 55 were included (10 multicentre and 45 single-centre; 13 prospective and 42 retrospective). The most frequently reported outcomes were long-term (40 studies, 73%) and surgical outcomes (34 studies, 56%). Reintervention-related outcomes included the need for further surgery (25 studies, 45%) and ongoing dilatation (12 studies, 22%). Four studies examined delivery outcomes and infant mortality post-surgery. Commonly reported surgical complications included incontinence (21 studies, 38%), urinary tract infections (UTIs) (17 studies, 30%), ongoing pain (10 studies, 18%), and fistulas (11 studies, 20%). Regarding UTIs, only 13 studies reported increased prevalence, with reduced clinical severity within the CAH cohort. Long-term outcomes frequently addressed quality of life (QoL) (13 studies) and sexual function (19 studies), with four of the QoL studies additionally exploring the quality of relationships. Findings regarding QoL were heterogeneous: six studies reported no statistically significant difference compared to control groups, while six suggested an association between surgery and reduced QoL. Overall, the lack of control groups in several of the studies limited the validity and generalisability of findings.

Conclusion

Our scoping review found that published evidence on the outcomes of surgery in women with CAH is limited and heterogeneous. There is a need for more robust evidence from multicenter, larger cohort studies that combine quantitative and qualitative methods to help develop benchmarking tools and improve the quality of care.

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P122

Isolated premature menarche: is there an increasing trend- a case series

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Introduction

Menarche is a late pubertal event that occurs at a mean age of 12.4 years and follows on an average of 2.5 years after the onset of thelarche. Some studies suggest an increase in precocious puberty (including early menarche) during the COVID-19 pandemic, potentially due to lifestyle changes and stress. However, this is an area of ongoing research, and the exact mechanisms are not fully understood. There is no published literature to suggest a similar increase in the incidence of isolated premature menarche.

Methods

We audited the clinical and laboratory data on girls referred to our service for early menarche. There were 11 referrals from primary care from Oct 2019 to April 2025 with an age range of 6.27-9.19 years. 1 patient is still being investigated and hence excluded from analysis.

Results and outcomes

The Tanner stage was B2, with advanced bone age in 3 girls and B1 in 7. The uterine length was pre-pubertal in 9 and borderline at 3.5 cm in 1 girl. Thyroid functions were checked in 8 girls and were normal. 4 girls were overweight based on BMI and 4 showed evidence of increased height velocity. 3 girls had recurrent vaginal bleed and had an MRI scan of brain (normal results), but only 1 required treatment with GnRH analogue. 8 girls have been discharged from the service as there were no ongoing concerns and 1 is still under follow-up.

Conclusions

We have noticed a sudden increase in referrals for isolated menarche since 2020. Although increased exposure to endocrine-disrupting chemicals through diet or environment could be contributing to this trend, the exact cause of this remains unknown at present.

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P123

Complete gonadal dysgenesis in a 46, XY individual associated with a pathogenic MAP3K1 variant: a case report and review of management

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Introduction

Variants in the MAP3K1 gene are increasingly recognised as a cause of 46, XY disorders of sexual development (DSD), particularly in individuals with phenotypically female presentation. We report the case of a 15-year-old female who presented with delayed puberty and was ultimately diagnosed with 46, XY DSD due to a gain-of-function variant in MAP3K1.

Case

A 15-year-old girl was referred to the paediatric endocrine service for evaluation of absent pubertal development. There were no concerns regarding her sexual differentiation at birth. Her medical history is significant for recurrent urinary tract infections, chronic kidney disease secondary to reflux nephropathy and bilateral renal scarring. Examination findings: Weight - 25th percentile, height 75th percentile. Blood pressure - normal at 117/71 mmHg. Systemic examination - unremarkable.

Results

FSH (150 IU/l) and LH (61.6 IU/l) were markedly elevated indicating hypergonadotropic hypogonadism, consistent with primary gonadal failure. AMH was undetectable (<1 pmol/l), and both testosterone (0.3 nmol/l) and oestradiol (20 pmol/l) were low, supporting complete gonadal dysgenesis. Pelvic MRI showed a 2 cm hypoplastic uterus with vaginal tissue but no visible gonads. Karyotype was 46, XY with a balanced translocation t (1;4) (p13.3; q31.21), and genetic testing identified a heterozygous likely pathogenic *MAP3K1* variant (c.1016G>A; p. Arg339Gln).

Management

She has been started on pubertal induction with transdermal oestradiol. She is awaiting laparoscopic cystoscopy and vaginoscopy. Gonadal localisation and possible gonadectomy will be performed concurrently.

Discussion

MAP3K1 encodes a serine/threonine kinase that plays a critical regulatory role in the MAPK signalling pathway, influencing the balance between pro-testis and pro-ovary pathways during gonadal differentiation. The gain-of-function MAP3K1 variants can lead to upregulation of β -catenin and downregulation of SOX9 expression, thereby disrupting testicular development and favouring ovarian-like differentiation in 46, XY individuals. In individuals with complete gonadal dysgenesis, the risk of developing gonadal malignancies—particularly gonadoblastoma and dysgerminoma—is significantly elevated due to the presence of dysgenetic gonads. Consequently, prophylactic bilateral gonadectomy is recommended.

Conclusion

This case underscores the need to consider 46, XY DSD in adolescents with absent puberty, with *MAP3K1* variants as an emerging cause disrupting testicular development via MAPK pathway dysregulation.

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Miscellaneous/Other 2

P124

Improving CEW service awareness and obesity management in a district general hospital (DGH): an educational and quality improvement initiative

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Background

Childhood obesity continues to rise in the UK, with one in four children affected, and higher rates in the most deprived areas. The Complications of Excess Weight (CEW) service is aimed at supporting children and young people (CYP) and families in identifying and managing any complications associated with obesity. They provide specialist MDT support, but awareness of referral pathways, criteria, and local responsibilities is limited among DGH teams. Following a CEW service project presented at BSPED, I organised a teaching session in my department, which informed the development of a departmental guideline to support ongoing learning and improve service delivery for this cohort of patients.

Aim

To improve awareness of the CEW service, referral criteria, and baseline management in a DGH setting, and to support sustained practice change through a local guideline.

Methods

A pre-teaching survey was used to assess the team's knowledge of the CEW service, confidence in identifying obesity-related complications, and familiarity with referral criteria. I then delivered a structured teaching session covering:

- Childhood obesity classification
 - Indications and criteria for CEW service referral
 - Relevant baseline investigations
 - Social determinants, including ethnicity, food deserts, and areas of deprivation
- A post-teaching quiz assessed knowledge gained, and a local guideline was developed afterwards.

Results

Fifteen staff responded to the pre-teaching survey; 53% were unaware of the CEW service, and only 6/15 correctly identified referral criteria. Confidence in recognising complications averaged 6.3/10.

Post-teaching results ($n = 10$) showed:

- 100% correctly identified childhood obesity classification
- 80% knew the correct age criteria for referral
- 60% identified the correct BMI threshold
- 30% correctly selected all baseline investigations

Feedback was positive, and several team members requested more information, prompting the development of a local guideline outlining referral criteria, baseline assessments, and supportive care needed.

Conclusion

A single teaching session significantly improved the team's awareness of CEW services and obesity management. The introduction of a departmental guideline supports sustained learning and practice. Continued education is needed, especially around baseline investigations and signposting patients to community-based supports available. Locally tailored guidelines can help bridge the gap between tertiary and general paediatric care.

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P125

Swinging variability in diazoxide responsiveness in congenital hyperinsulinism due to pathogenic *HNF4A* variants: the role of continuous glucose monitoring

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Background

Loss-of-function variants in the hepatocyte nuclear factor 4 alpha (*HNF4A*) gene cause diazoxide-responsive congenital hyperinsulinism (CHI) which progresses to maturity-onset diabetes of the young (MODY). While the switch from hypoglycaemia to hyperglycaemia is well-documented, neonatal swinging in diazoxide responsiveness and glycaemic fluctuations, identified by continuous glucose monitoring (CGM), is not well-known.

Objectives

To illustrate the swinging variability in glycaemic response to diazoxide using CGM in *HNF4A*-CHI.

Methods

Clinical data were retrospectively collected from two children with neonatal CHI. CGM (Dexcom G7®) data were tracked up to 11 weeks for episodes of hypoglycaemia (<3.5 mmol/l) and hyperglycaemia (<10 mmol/l). **Cases** **Patient A**, male, presented at Day 2 of life with CHI resulting from a paternal heterozygous *HNF4A* frameshift variant (c.577del, p.(Asp193Thrfs*31)). During the first two weeks, he responded to diazoxide (3 mg/kg/d) and intravenous glucagon (10 mg/kg/h). Diazoxide was increased to 10 mg/kg/d to facilitate cessation of glucagon. CGM-identified hyperglycaemia was noted (12.1-16.7 mmol/l) on Day 23, which persisted for three days, prompting discontinuation of diazoxide. Subsequent CGM-identified hypoglycaemia on Day 26 (2.3-3.3 mmol/l) resulted in reintroduction of diazoxide (3.3 mg/kg/d) and glucagon (10 mg/kg/d), with diazoxide being titrated up to 10 mg/kg/day alongside a weaning dose of glucagon. A second episode of hyperglycaemia to diazoxide (13.9-21.8 mmol/l) was noted on Day 39. Following discontinuation, diazoxide was recommenced on Day 41 and increased to 12 mg/kg/d based on CGM response. **Patient B**, female, presented at Day 1 of life with CHI resulting from an antenatally diagnosed paternal *HNF4A* nonsense variant (c.48C>A, p.(Tyr16Ter)). She was initially unresponsive to diazoxide (15 mg/kg/day) and relied on intravenous glucagon (12.5 mg/kg/d) to maintain normal glycaemic profile on CGM from Day 8 to 60. Hypoglycaemia (2.4-3.0 mmol/l) was observed on Day 61; diazoxide (7 mg/kg/d) was reintroduced and titrated up to 10.7 mg/kg/day, while glucagon was weaned. Hyperglycaemia to diazoxide (12.9-26.5 mmol/l) was noted on Day 75, leading to medication discontinuation. On Day 78, following hypoglycaemia, diazoxide was restarted and escalated to 12.5 mg/kg/day to maintain CGM euglycaemia.

Conclusion

HNF4A-CHI may show fluctuating diazoxide responsiveness and a swinging glycaemic phenotype for which high frequency monitoring by CGM is useful for diazoxide dose adjustments.

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P126

Digital poverty – barriers to care: a departmental experience

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Background

Digital innovation at Great Ormond Street Children's Hospital (GOSH) has led to an unprecedented amount of support for families from direct messaging systems to electronic access in real time to results & letters. This unfortunately can result in a widening gap in the care provided to families who are digitally 'rich' in terms of access to devices and the ability to utilise these supports compared with those who face socio-economic and language barriers.

Aims and Objectives

We set out to ascertain the nature and extent of these barriers families face when accessing digital services. Alongside this, we describe the measures our trust has taken to support families.

Methods

We carried out a mixed-methods study with a retrospective analysis of all patients under the care of the endocrinology department from 18/04/2019 – 01/07/2025, by analysing our electronic healthcare record using the 'PICTURE' data platform.

Results

There were 10,913 patients under the care of our team during the study period, with two subgroups for congenital hyperinsulinism (CHI) (*n* = 901) and congenital hypothyroidism (CHT) (*n* = 807). Of the general endocrine cohort, 39% were white British with 72% self-reporting English as their first language, with 74 other primary languages identified. Although only 8.88% of our CHI and 9.12% of our CHT cohorts attended their first visit via telemedicine, 55% & 60% respectively, avail of follow up telemedicine visits. Using IMD (Index of multiple

deprivation) deciles, we have identified that 44% of our CHT cohort, are especially vulnerable, in the bottom 30% of this range. 90% of our endocrinology patients are signed up to our 'MyGOSH' app, a digital innovation which allows families to access specific parts of their electronic record. Our department has incorporated a digital poverty screening tool to be used by our central booking office team to identify the needs of families.

Conclusions

Our results demonstrate that there are identifiable cohorts with ongoing barriers to accessing our digital support services. In collaboration with the health inequalities board, our trust offers SIM cards and donated devices. Furthermore, our inpatient 'MyGOSH' app – 'MyGOSH Bedside' will allow families equitable access using bedside screens during their inpatient stays.

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P127

Diagnosis and management of congenital hyperinsulinism at a tertiary neonatal unit: a service review

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Introduction

Congenital hyperinsulinism (CHI) is a rare and important cause of neonatal hypoglycaemia characterised by hypoglycaemia secondary to inappropriate insulin secretion. Fluctuating glycaemic trends in early neonatal life and time-sensitive biochemical testing present clinical challenges. Management of CHI is complicated, requiring communication between multiple specialist teams. Early detection of hypoglycaemia prevents neurodevelopmental sequelae.

Objective

To evaluate CHI diagnosis & management in a level 3 neonatal unit, and identify practice patterns/knowledge gaps among specialists in line with 2023 UK Collaborative Consensus.

Methods

All neonates diagnosed with hyperinsulinism between March 2020 and March 2024 at Luton NICU were identified retrospectively using *Badgernet* database. Data included patient demographics, gestational age, birth weight, ethnicity, maternal and family history, hypoglycaemia screening tests, medical interventions (glucose bolus and medications), nutritional sources, glucose infusion rate (GIR), day of life at diagnosis, genetics tests requested, glucose monitoring, dietetic reviews and neurodevelopmental follow-up.

Results

We identified 30 neonates with CHI (gestational age 27 + 6 weeks - 40 + 6 weeks), average weight was 2.69 kg (0.62 kg - 5.19 kg). The majority were full term (19/30), and boys (20/30). Insulin level was sent for 29/30 infants; average insulin level was 16.2mIU/l. First line investigations including growth hormone (0/30), lactate (24/30), and c-peptide (16/30) were frequently missed. 14/30 received 20% dextrose via central line with mean GIR 13.7 mg/kg/min (6.9-17.7 mg/kg/min). 18/30 babies were commenced on medication (diazoxide & chlorothiazide) after appropriate cardiac monitoring. One infant had a genetic cause for CHI and received early input from a CHI quaternary centre.

Conclusion

CHI is often overlooked; mean age at diagnosis was 11.7 days with average of 5 hypo-episodes/neonate recorded. Testing for C-peptide was missed in 53.3%, free fatty acids (FFA) in 40% and ketones in 36%. Importance of FFA/ketones testing should be reiterated to aid faster diagnosis, as insulin may be undetectable (20%), and levels may not correlate with hypoglycaemic severity. Amino acids (57.6%) and urine organic acids (64.7%) needed repetition due to prematurity or insufficient sampling, wasting time and resources. Developmental follow-up is crucial, as four infants (18.1%) were detected with developmental delay. Only 16.6% of infants were exclusively breastfed on discharge, highlighting need for better breastfeeding support.

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P128

Growth in children with Septo-optic dysplasia (SOD) treated with GnRH agonist therapy (GnRHa)

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Introduction

Septo-optic dysplasia (SOD) is characterised by a classical triad of optic nerve hypoplasia, pituitary hormone abnormalities and midline brain defects. Growth in children with SOD can be affected by multiple factors such as endocrinopathies, associated genetic and chronic medical conditions. Children with SOD can also present with precocious or delayed puberty.

Aim

We aimed to review data on growth, endocrinopathies and MRI findings in children with SOD in whom GnRHs were used.

Methods

We accessed electronic medical records of children with SOD in whom GnRHs (Triptorelin, Gonapeptyl or decapeptyl) was used. We collected data on their height, BMI, endocrinopathies and MRI findings.

Results

21 children (8 M, 13 F) met the inclusion criteria. Girls were commenced on puberty blockers at a median age of 8.6 years whereas boys were started on puberty blockers at a median age of 10.6 years. Puberty blockers were stopped at around the same time in both (13 years for boys and 12.98 years in girls). The median height SDS of boys improved significantly from -0.57 SDS to 2.01 SDS at the last visit. However, the height SDS worsened after the use of puberty blockers amongst girls (-0.3 VS -1.5 at last visit). No significant change in BMI was noted amongst both the sexes. Puberty blockers were stopped in 18 patients and all of them underwent spontaneous puberty except one girl who required sex steroids. Six males (75%) and eight females (61 %) presented with multiple pituitary endocrinopathies. Similarly, 50% ($n = 4/8$) of males and 53% ($n = 7/13$) of females had a small anterior pituitary gland on MRI scan.

Conclusion

Puberty blockers were used in girls more frequently and for longer duration than boys possibly because the incidence of precocious puberty in girls is higher than in boys in SOD. Height prospects were better in boys than in girls treated with puberty blockers.

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P129

Active management of endocrine waiting lists by clinical triage

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Introduction

Paediatric endocrinology typically has high follow-up rates in out-patients from the first appointment because of the nature of the conditions seen. When there are clinical capacity problems, large follow-up waiting lists can easily develop.

Methods

124 outstanding endocrine follow-up appointments in a district general hospital endocrine service with sickness and capacity problems were triaged to review the need, urgency and method of follow-up. Using a pre-populated spread-sheet, an experienced paediatric endocrinologist reviewed the electronic notes, recorded the diagnosis, decisions made when last seen, relevant investigations and decided whether to discharge with or without a letter or offer a face-to-face or telephone appointment. The reasons for discharge were recorded. Most of the diagnoses related to growth, puberty, weight or thyroid issues. The time and cost / patient were calculated.

Results

Triage of the 124 patients took about 4.5 minutes / patient, equivalent to about 10 hours of clinical time. Because of the delay, 54.8% were over 16 years old at the time of triage. The cost of triage was £10.73 / patient and a total cost of £1330. 98 [79%] could be discharged with no further action required. 9 [7.3%] were offered face-to-face appointments. 19 [15.3%] were offered telephone follow-up which were accepted by 10. 1 urgent appointment was required.

Outcome

Our experience is that an apparently large waiting list problem can be managed in an efficient and cost effective way by clinical triage. This outcome data may not apply to other specialties or outside paediatrics. This is a good use of resources when there are clinical capacity problems and should become a routine tool in waiting list management. Waiting list are dynamic, not static tools and require active management.

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P130

A rare case of polyurea in infancy

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Introduction

Congenital nephrogenic diabetes insipidus (CNDI) is a rare genetic disorder characterized by the kidneys' inability to respond to arginine vasopressin (AVP), resulting in profound polyuria and risk of hypernatremic dehydration in early infancy. Approximately 90% of cases are X-linked, caused by mutations in the AVPR2 gene, while a minority arise from autosomal mutations in the aquaporin-2 (AQP2) gene. Early diagnosis is critical, as untreated CNDI may result in severe dehydration, failure to thrive, and neurodevelopmental impairment. Despite typical laboratory findings of dilute polyuria and hypernatremia, distinguishing CNDI from central diabetes insipidus can be challenging, especially in the neonatal period. Management focuses on minimizing urinary water loss, correcting electrolyte disturbances, and ensuring adequate growth and development.

Case

We report a 4-month-old boy presenting with polyuria (6.5 mL/kg/h), poor weight gain (4.8 kg, <3rd centile), and length 64 cm. There was no significant family history. Laboratory evaluation revealed hypernatremia (serum sodium 152 mmol/L), low urine osmolality (149 mosm/kg), and urine specific gravity of 1.001. An initial trial of oral desmopressin produced no change in urine output or osmolality. Given the early onset, laboratory findings, and lack of desmopressin response, CNDI was suspected. Water deprivation testing was avoided due to the patient's age and risk of dehydration. Genetic analysis identified a pathogenic hemizygous mutation in the AVPR2 gene. Management comprised hydrochlorothiazide therapy (2 mg/kg/day) resulting in normalization of serum sodium and a reduction in urine output to 1.9 mL/kg/h. However, the child developed hypokalaemia, prompting the addition of low-dose amiloride.

Discussion

CNDI is a diagnostic and therapeutic challenge in infants, with genetic analysis playing a pivotal role in definitive diagnosis. Early recognition is essential to prevent irreversible sequelae of chronic dehydration. The treatment involves a combination of thiazide diuretics, potassium-sparing diuretics, and dietary modification to minimize urinary losses and promote growth. In our case, prompt genetic confirmation allowed for early targeted therapy, leading to normalization of electrolytes, improved growth trajectory, and favourable outcome. Long-term monitoring for growth, neurological development, and renal function remains imperative given the risks of chronic kidney disease.

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P131

Hyperinsulinism in a child with an insulin gene variant. causative or co-INS-idence?

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Introduction

The human insulin gene (*INS*) encodes pre-proinsulin, which is processed into the peptide hormone, insulin. Pathogenic *INS* variants classically lead to permanent neonatal diabetes (PNDM) with a few rare reports describing hyperinsulinaemia and insulin-resistant diabetes in adults. To our knowledge, *INS* variants have not been described to cause hyperinsulinaemic hypoglycaemia. We report a neonate with a pathogenic *INS* variant that is predicted to cause PNDM, but who instead presented with congenital hyperinsulinaemic hypoglycaemia.

Case Report

A term female infant (birth weight [BW] 4.5 kg) was admitted at 48-hours with poor feeding, lethargy, and hypothermia (32.9°C). A hypoglycaemia screen with a plasma glucose 2.6 mmol/L showed insulin 149pmol/L, C peptide 884pmol/L, 3 hydroxybutyrate <100 µmol/L and free fatty acids <275 µmol/L confirming hyperinsulinaemia hypoglycaemia. She required a glucose infusion rate of 9.8 mg/kg/min and a glucagon infusion to maintain normoglycaemia. Mother had a normal antenatal oral glucose tolerance test. Father and paternal grandmother have diabetes mellitus. They were diagnosed at 5 and 3 years, respectively, with

BW of 3.02 kg and 4.55 kg. Diazoxide (15 mg/kg/day) and Chlorothiazide (10 mg/kg/day) are required, with ongoing evidence of occasional hypoglycaemia down to 3.4 mmol/l at 9-weeks. Hyperglycaemia episodes have resolved with feed adjustments. Trio next generation sequencing performed within the national *Generation Study*, a broad research-based neonatal genomic screening programme, identified a heterozygous variant in the *INS* gene (c.188-31G>A, p.?) which has been confirmed. This variation is a well-established cause of PNDM. Routine diagnostic screening of the known congenital hyperinsulinism genes did not identify a second pathogenic variant.

Conclusion

This case highlights a novel observation between hyperinsulinaemic hypoglycaemia and a pathogenic *INS* variant. The mechanism is not fully understood, so a causative relationship cannot be confirmed. The variant was identified via an antenatal research study and the result became available following hospitalisation. Further research is required to establish whether this *INS* variant is causal, but these findings may potentially broaden the spectrum of *INS*-related disease to include hyperinsulinism. This would require further investigation including the identification of additional cases with a bi-phasic phenotype to establish causality. Our patient will be closely monitored as she is high risk of developing diabetes.

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P132

Challenging investigations in the diagnostic workup: a clinical perspective

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Introduction

Secondary hyperparathyroidism is commonly seen in adults, but less often reported in paediatric patients, with chronic kidney disease, which leads to tertiary hyperparathyroidism following a renal transplant. It is thought to be secondary to parathyroid hyperplasia or downregulation of the calcium-sensing or vitamin D receptors. Increased age, phosphate and parathyroid hormone (PTH) levels pre-transplant are risk factors for persistent hyperparathyroidism. This leads to hypercalcaemia, hypophosphatemia, reduced graft function and growth concerns and therefore needs appropriate investigation and management.

Case Report

A 10-year-old boy with a background of a renal transplant secondary to bilateral renal dysplasia, autism spectrum disorder (ASD) and nasogastric feeding was admitted with hypercalcaemia due to hyperparathyroidism (Table1). The patient was treated with hyperhydration and low calcium diet. Neck ultrasound showed no parathyroid masses, and renal ultrasound showed no nephrocalcinosis. On stopping intravenous fluids and restarting standard diet his calcium increased again. Cinacalcet was commenced and increased in increments (Table1). The possibility of a parathyroid adenoma needed investigating. Magnetic Resonance Imaging and a Sestamibi scan were considered but were not ideal imaging modalities due to lower sensitivities and long imaging time under general anaesthetic (GA) due to his neurodivergence. An 18F Choline Positron Emission Tomography-Computed Tomography (PET-CT) was performed under GA. The scan clearly highlighted bilateral parathyroid hyperplasia. A multidisciplinary team have advised removal of all four parathyroid glands with auto transplantation.

Conclusion

18F Choline PET-CT is a specialised scan with high sensitivity in detecting parathyroid adenomas. It is only available in our local adult hospital, which was challenging to organise in our patient, but very successful in identifying the lesions. It is important to diagnose tertiary hyperparathyroidism in these patients by using high sensitivity modalities such as Choline PET-CT and initiate appropriate management to optimise bone biochemistry, graft function, bone health and growth.

Table 1: Biochemistry trend and medication changes

	Corrected Calcium (2.15-2.74) mmol/l	PTH (1.1-6.9) pmol/l	Phosphate (0.97-1.94) mmol/l	Vitamin D (>50nmol/l)	Urea (2.3-6.4) mmol/l	Creatinine (0.60) mmol/l	u.Ca/cre-ate (0-0.60) mm/mm Cr
At diagnosis	3.04	16.4	1.05	91	6.6	68	1.08
On Cinacalcet	2.36	13.5	1.19	98	7.6	76	x

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Miscellaneous/Other 3

P133

From prematurity and adrenal insufficiency to a syndromic diagnosis: a rare case of MIRAGE syndrome

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Introduction

MIRAGE syndrome is a rare disorder characterized by Myelodysplasia, Infections, Restriction of growth, Adrenal hypoplasia, Genital phenotypes and Enteropathy, caused by a mutation in the *SAMD9* gene. The genital phenotypes are variable, ranging from hypospadias to complete female phenotype in 46,XY patients, while 46, XX patients may have dysgenetic ovaries. Mortality is high, with a median survival of 3 years; death is typically due to recurrent infections.

Case presentation

We report the case of a 1-month-old female preterm infant (born at 27 weeks' gestation; birth weight 640 g; -1.77 SDS) who presented critically unwell with hypotension resistant to inotropic support but responsive to hydrocortisone, and recurrent episodes of hyponatremia. The clinical picture was also complicated by respiratory distress syndrome, sepsis, thrombocytopenia and conjugated hyperbilirubinemia. Initial blood tests revealed low cortisol (<0.3 nmol/l), TSH (1.4 mIU/l) and FT4 (7.2 pmol/l). Replacement therapy with hydrocortisone and levothyroxine was started. Further assessment of the pituitary function showed high ACTH (92 ng/l) while IGF1, LH and FSH were normal. The adrenal insufficiency work-up showed high renin (> 23.7 nmol/l/h) and low aldosterone (< 50 pmol/l), DHEAS (< 0.4 umol/l), 17-OHP (< 0.3 nmol/l) and androstenedione (< 0.7 nmol/l). Abdominal ultrasound documented absent adrenal tissue bilaterally, suggesting congenital adrenal hypoplasia. Fludrocortisone was also commenced. Further examination raised concerns of genital ambiguity (prominent clitoris and large labia majora). Investigations revealed a 46,XY karyotype, high inhibin B (346 pg/ml) and AMH (701.3 pmol/l), along with low testosterone (0.3 nmol/l) and 5DHT (< 0.25 nmol/l). Pelvic ultrasound showed absent uterine and ovarian tissue, with gonads located bilaterally in the groins. Genetic investigation identified a heterozygous pathogenic *SAMD9* variant (c.2920G>A p.(Glu974Lys)), consistent with MIRAGE syndrome. Despite intensive care, the infant passed away at 2 months of age.

Conclusion

MIRAGE syndrome should be suspected in infants with adrenal insufficiency and multisystem involvement, particularly when the clinical course is different from that typically expected for a sick infant. Diagnosis may be challenging due to the overlapping features with complications of prematurity.

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P134

Perseverance in looking beyond the primary diagnosis; Addison's disease and dent disease - an unlikely combination

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Background

Addison's disease is an autoimmune condition which affects approximately 1 in 14,000 people in the UK. It is caused by destruction of the adrenal cortex leading to insufficient production of cortisol and aldosterone. Dent disease is caused by a defect in a voltage gated chloride ion channel due to a mutation in *CLCN5* gene. It is characterised by renal tubular dysfunction, including proteinuria, hypercalciuria, nephrocalcinosis, and nephrolithiasis. Dent disease is considered a rare disease with prevalence estimated between 1 in 400,000 and 1 in 1,000,000.

Clinical Case

4-year-old male presented to hospital whilst on holiday in Turkey with a 3-day history of vomiting and lethargy. He was found to be in circulatory shock with hypoglycaemia, hyponatraemia and hypotension. The working diagnosis was a suspected underlying adrenal insufficiency; he was incidentally found to have bilateral nephrocalcinosis on renal ultrasound scan during his admission. Following discharge from PICU he was admitted to his local hospital. A short synacthen test confirmed adrenal insufficiency and adrenal antibodies were

positive confirming the diagnosis of Addison's Disease. He was commenced on hydrocortisone and fludrocortisone as well as sodium supplements for hyponatraemia. 6 months after his initial presentation the patient was admitted with lethargy and fever and was found to be COVID positive. Concerns were raised during this admission due to worsening renal function, hypokalaemia, proteinuria and his renin being significantly elevated (1,928.0 mU/l). He had normal renal function prior to his initial presentation. There were significant concerns from the Endocrine Team that despite initiating treatment for Addison's, the patient required increasing sodium supplements, and a renal pathology or unifying genetic diagnosis was suspected in addition to his diagnosis of Addison's. Further renal evaluation showed Glomerular Filtration Rate 54 suggestive of CKD stage 3, this indicated significant renal deterioration over 1 year. Genetics confirmed a secondary diagnosis of Dent's Disease, 19 months after his initial presentation with Addisonian crisis and nephrocalcinosis.

Conclusion

This case highlights the importance of perseverance in looking for additional diagnoses when the initial one does not encompass the entirety of the complex presentation.

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P135

Development and implementation of a CNS-led pathway to discontinue diazoxide at home in infants with transient hyperinsulinism

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Background

Transient Hyperinsulinism (HI) often resolves within the first year of life, but many affected infants require temporary treatment with diazoxide, a medication associated with side effects including pulmonary hypertension, fluid retention, and hypertrichosis. Historically, discontinuation of diazoxide required hospital admission for glucose monitoring and fasting tolerance testing, typically scheduled between 6–12 months of age. During the COVID-19 pandemic, restrictions delayed inpatient admissions, potentially prolonging unnecessary exposure to diazoxide therapy.

Objective

To reduce delays in re-evaluating the need for diazoxide therapy and unwarranted exposure to associated risks, we developed a new CNS-led pathway allowing parents/carers to safely stop diazoxide at home under structured guidance and support from the HI specialist nursing team.

Methods

The pathway included clear education for parents/carers, regular telephone check-ins, and ongoing home blood glucose monitoring. Once off diazoxide, infants were admitted for a single overnight fasting tolerance test, rather than the previous 48-hour inpatient stay with a 24-hour glucose profile. A service satisfaction survey was conducted in 2023 to evaluate family experience with the pathway.

Results

Implementation of the pathway led to a significant reduction in time on diazoxide therapy, with the average age of discontinuation reduced to 3 months. Hospital admission duration was halved, reducing burden on families and improving inpatient efficiency and financial benefit. Feedback from 30 families showed 100% recalled discussions about the pathway, with 72% strongly agreeing and 24% agreeing they felt confident stopping diazoxide at home. Parents appreciated shorter admissions, fewer medication side effects, and improved feeding flexibility.

Conclusion

This CNS-led pathway enabled earlier discontinuation of diazoxide, reducing exposure to side effects and improving quality of life for infants and families. It also led to cost savings through reduced bed days and medication use. Positive parental feedback supports continued use of the pathway.

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P136

Transition of patients with congenital hyperinsulinism from proglycem® diazoxide liquid to diazoxide tablets

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Background

Diazoxide is the only oral treatment used in the management of Congenital Hyperinsulinism (CHI). While the liquid formulation (Proglycem®) is commonly prescribed, it poses challenges due to high cost and limited availability in primary and secondary care. Most patients under our care receive prescriptions directly from our service creating a dependency on tertiary services. We aimed to transition eligible patients (aged ≥7 years) from the liquid to tablet formulation, improving access, reducing costs, and supporting independent prescribing in primary care.

Methods

We identified eligible patients currently receiving diazoxide liquid. A multi-disciplinary team (MDT) developed a standardized dose conversion guide and educational materials for patients and families to facilitate the transition. Additional support from play specialists and psychology was offered where appropriate. We worked with pharmacy to secure a stable supply of diazoxide tablets and created a contingency plan for unlicensed capsule use.

Results

19 eligible patients were identified on retrospective review of our patient cohort. To date, 10 have successfully transitioned to tablets/capsules. Patient and caregiver education materials, including tablet-swallowing training, were developed and implemented. Early findings show maintained glycaemic control, no adverse events, and improved prescribing efficiency. Projected cost savings are estimated at ≥75%. This project is ongoing, with positive feedback from families and improved prescribing sustainability.

Conclusion

The structured transition from diazoxide liquid to tablet formulation demonstrates a safe, effective, and sustainable approach for managing CHI in older children. It reduces tertiary centre dependency, empowers families, supports GP prescribing, and achieves significant cost savings without compromising patient care.

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P137

Use of continuous glucose monitoring (CGM) to aid weaning of medication in children with hyperinsulinism

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Background

Many children with Hyperinsulinism are on long term medications to control their blood glucose levels (BGL). Prior to CGMs, a patient would be admitted for two-nights to assess efficacy of treatment plan, often not reflecting the child's daily routine. With CGMs we can frequently assess this and make changes to remotely. Including weaning of medications and in some cases stopping medications.

Method

Capillary testing gives a small snapshot of a child's BGL's. Prior to clinic appointments we remotely assess CGM data to decide potential treatment changes. We can identify when patients have been stable on medications and advise when medications can be weaned. We can identify when patients are unstable on their current regime and make changes, accordingly, often preventing the need for repeat admissions. A CGM is frequently used for a short time when weaning medications reassuring professionals, patients and carers it is safe to wean medications whilst being in their home environment.

Results

Routine annual fasting test and hospital admissions to adjust treatment are no longer standard practice unless clinically indicated. The length of stay has decreased from 2 nights to 1 night, reducing pressure on hospital bed capacity and staffing resources. The implementation of CGM into patient management has enhanced patient safety, optimised blood glucose control and improved quality of life of the patients and their families. Patients can remain at home in their normal environment whilst making medications/feed adjustments with minimal disruption to their daily life and education.

Conclusion

Patients and their carers are often apprehensive to start weaning a long-term medication and CGM offers reassurance that they won't miss any hypoglycaemic episodes with capillary testing. The technology offers alerts and trends, which are particularly useful for our cohort of patients' safety. Positive parent/ carer feedback and reduction of hospital admission are evidence of the benefits. While CGM can be expensive, when reducing hospital admissions, it saves the trust money and enable to patients to live without regular hospital intervention.

Additionally, this work is in line with the new NHS 10-year plan shifting working from hospital to community and embracing digital technology.

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P138

Raising awareness of hyperinsulinism: empowering healthcare professionals in the early detection of a rare disease

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Background

Hyperinsulinism (HI) is a rare, life-threatening condition affecting 1 in 28,000 babies each year. It results from inappropriate, unregulated insulin secretion from pancreatic beta cells, leading to hypoglycaemia. High insulin levels prevent the utilisation of alternative fuels essential for the brain; hence hypoglycaemia can lead to seizures, developmental delay, visual impairment and even death.

Objective

As experienced Clinical Nurse Specialists (CNSs) at a Highly Specialised Congenital Hyperinsulinism Centre, our aim was to raise awareness of HI. This was achieved by sharing knowledge to improve early recognition, diagnosis and to prevent adverse outcomes.

Methods

We identified key health care professionals who would have the biggest impact on patient outcomes. This target group included bedside midwives, neonatal nurses / doctors, health visitors and general paediatricians. It was vital that the take home messages were impactful and could be implemented. We worked with the Royal College of Midwives and together the HI CNSs from the two UK Highly Specialised Centres delivered a webinar presentation. To reach other health care professionals we held a hybrid conference inviting National and International HI experts. A nursing article is awaiting publication. Lastly, we have worked with the UK and International HI parent groups to produce resources that are shared online.

Results

Raising awareness of HI led to much discussion and interest. The numbers of professionals attending the webinar and conference reflected this, with comments such as "I will be far more cautious with low glucose on post-natal wards or in A&E". There is an ongoing demand for further teaching sessions and future publications.

Conclusion

This interdisciplinary approach to raising awareness of HI has demonstrated the clear need for ongoing education. Early recognition and intervention are critical in preventing irreversible brain injury in affected infants. Preventing harm in even a single case validates the importance for us to continue with this work of raising awareness of HI.

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P139

Implementation of a digital perioperative planning interface in electronic patient records: a clinical audit in high-risk paediatric patients

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Background

Effective perioperative care in high-risk patients depends on timely access to clear and actionable management plans. However, critical instructions are often within unstructured or non-standardised clinical documentation, limiting visibility. This can adversely affect patient care and outcomes. Integrating a clinician-facing

perioperative planning interface into the electronic patient record (EPR) provides a scalable solution to improve information access, streamline workflows, and enhance safety for paediatric patients with complex comorbidities.

Objective

To design and audit the implementation, usability, and uptake of a digital interface for embedding perioperative plans in paediatric patients with complex medical conditions.

Methods

We conducted a 12-month retrospective audit (1 July 2023 – 1 July 2024) of a digital perioperative planning interface integrated into the EPR at a tertiary children's hospital. The interface enabled clinicians to document structured plans using pre-configured "speed buttons" for high-risk conditions such as adrenal insufficiency, diabetes, and hypoglycaemia. Plans were designed for real-time visibility within the EPR storyboard, where hovering over the relevant section displayed the plan for convenient review. Usage was tracked by number of plans entered, categorised by subspecialty. Qualitative feedback from nurses and doctors was collected to explore usability and perceived impact on clinical workflow.

Results

A total of 1019 structured perioperative plans were documented, with highest usage in adrenal (44.7%) and hypoglycaemia (23.3%) pathways, followed by diabetes, metabolic, and other conditions ($p < 0.001$). The digital interface enabled timely access to perioperative instructions, improving clarity and visibility. Themes of feedback reported by end-users included enhanced workflow efficiency, patient safety, and interdisciplinary communication. While uptake was high in endocrine-related pathways, usage was lower in other specialties, highlighting opportunities for broader engagement and system-wide optimisation.

Conclusion

Future implementation studies should evaluate whether structured perioperative planning interfaces within the EPR translate into measurable improvements in patient outcomes and care quality. Identifying barriers to adoption will require an iterative, user-centred approach, incorporating methods such as design sprints and co-design workshops. Broad stakeholder engagement will be essential to ensuring sustained uptake and functionality. Priorities for further development include interface customisation, visual design enhancements, and seamless information flow across trusts.

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P140

Severe hypercalcaemia presenting with acute abdominal pain: learning from two cases of primary hyperparathyroidism

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Background

Primary hyperparathyroidism (PHPT) is rare in children, with an estimated incidence of 2–5 per 100,000. Hypercalcaemia presents with non-specific symptoms, often resulting in delayed diagnosis. Children typically manifest more severe biochemical and clinical features than adults, including nephrocalcinosis and marked skeletal involvement at presentation.

Aim

To describe the presentation, clinical features, management, and outcomes of two paediatric cases of PHPT.

Clinical cases

Both cases presented to the emergency department with a short history of acute abdominal pain. Blood tests revealed marked hypercalcaemia and elevated

Table 1. Clinical and biochemical case comparison

Parameter	Case 1	Case 2
Sex, Age	Female, 12 years	Female, 14 years
Presenting complaints	48 hours of abdominal pain, polyuria, polydipsia, lethargy	6 hours of abdominal pain
Medical history	Constipation, learning difficulties	Chronic headache, knee pain
Adjusted Calcium (2.25–2.69 mmol/l)	3.44	3.94
Parathyroid Hormone (1.95–8.49 pmol/l)	23.11	79.18
25OH Vitamin D (>50 nmol/l)	36.0	30.2
Urinary Calcium 24h (2.50–7.50 mmol/24h)	-	14.54
Serum Creatinine (35–65 µmol/l)	39	91
Parathyroid ultrasound	Right superior node enlarged (11 × 5 × 7 mm)	Left superior node enlarged (22 × 6 × 12 mm)
Acute management	Hyperhydration, furosemide	Hyperhydration, pamidronate
Parathyroidectomy timing	Day 4	Day 10
Post-operative adjusted calcium (mmol/l)	2.16	1.84
Post-operative course	Uncomplicated	Hungry bone syndrome

parathyroid hormone levels (Table 1). Ultrasound imaging identified solitary parathyroid enlargement in both cases. Case 1 received furosemide and hyperhydration with good effect, whereas Case 2 had concurrent acute kidney injury and hyperhydration-resistant hypercalcaemia. She therefore received pamidronate reducing serum calcium to 3.10 mmol/l. Both underwent parathyroidectomy with histological confirmation of adenoma. Post-operatively, Case 1 achieved normocalcaemia. Case 2 developed symptomatic hypocalcaemia with a fall in PTH levels (1.17 pmol/l), managed with oral calcium supplementation. Case 2's chronic knee pain and headaches resolved post-operatively. Multiple endocrine neoplasia gene panels were negative.

Conclusion

PHPT, though rare in children, often presents with acute abdominal pain. Early recognition, multidisciplinary care, and definitive parathyroidectomy are essential to prevent long-term complications. Clinicians should remain vigilant for hungry bone syndrome post-operatively. These cases prompted the development of a local paediatric hypercalcaemia guideline.

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Obesity 2

P141

Inpatient stay to improve and sustain weight outcomes as part of holistic weight management strategy in Tier 3 children's obesity service

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Introduction

The Complications of Excess Weight (CEW) service is an NHS-England pilot for children and young persons (CYP) living with severe childhood obesity, aiming to reduce associated complications. Thirty centres in England deliver care using a multidisciplinary team (MDT) approach. Alongside the usual MDT input, an inpatient admission is offered to those with difficulty achieving weight stabilisation. This provides an intense period of management for the patient and family, can demonstrate change is feasible and informs safeguarding. The families receive healthy eating advice, exercise information, and are shown examples of age-appropriate meals and portion sizes. Special consideration is given to family circumstances and additional needs of the child/family. Patients are admitted for 10-12 days opposite the nursing station in a designated ward. An individualised package supports the inpatient stay. The parent and CYP sign a contract that checks understanding of the process and outlines expectations.

Methods

Data were collected for all patients who had had an inpatient stay between 1st May 2024 and 30th June 2025 from their electronic patient record, including age, gender, duration of stay, weight/BMI change during admission and weight change following admission with median (range) calculated.

Results

Six children, median age 10.7 (range 6.0-12.2) years, completed an inpatient stay. Median length of stay was 11 (9-12) days during which time there was 2.3 kg (1.3-2.8) weight loss (2.6% (1-4.3)). Patients continued to lose further weight, data available, for up to 3 months after admission (0.35-3 kg). Two patients then gaining weight compared to discharge between 4-6 months, two below discharge weight and two data not yet available.

Conclusion

All patients lost weight during their admission allowing families to see this was possible. Difficulties were identified around availability of healthy choices on hospital menus, poor documentation of diet, prioritizing elective admissions, parental agreement for admission and MDT capacity to provide intense input. Weight loss during the admission is not sustained in the long-term demonstrating the difficulty in enabling persistent behaviour change.

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P142

Weight trajectory in children and young people from referral to first appointment in a Tier 3 weight management clinic- impact of age, gender and IMD

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The Complications of Excess Weight (CEW) service is a Tier 3 NHS England programme providing specialised care for children and adolescents with obesity. Understanding weight trajectories prior to first appointments is essential for contextualising patient progress within CEW. This retrospective study analysed 176 children and young people referred to the Nottingham CEW service between January 2021 and April 2024, focusing on growth patterns before specialist intervention. Patient demographics, obesity-related comorbidities, and weight velocity from referral to first appointment were evaluated alongside socio-economic deprivation indices. The cohort had a median age of 13.3 years, with 55% male and 48% from the most deprived quintile. At referral, 22% presented with one or more obesity-related complications, predominantly concentrated within the lower deprivation deciles (82% IMD 1-4). Weight velocity between referral and first appointment was categorised as weight gain (>1 kg/year), weight loss (<-1 kg/year), or weight maintenance (-1 to +1 kg/year). Seventy-nine percent gained weight with a median of 10.69 kg/year (IQR 8.32 kg/year), 16% lost weight with a median of -8.25 kg/year (IQR 23.24 kg/year), and 5% maintained stable weight with a median of 0.02 kg/year (IQR 0.74 kg/year). Females comprised 64% of the weight-loss group, contrasting with a male majority in the overall cohort. Chi-square analysis found no significant association between sex and weight loss when including those who maintained weight ($P = 0.076$), but a significant association was observed when comparing only those that gained or lost weight ($P = 0.023$), with males more likely to gain weight. Furthermore, patients aged 15-18 were significantly more likely to lose weight than age groups 0-5, 6-11 and 12-14 ($P = 0.014$). No significant relationship was found between deprivation status and weight change. Further analysis of the subset who lost weight between referral and first appointment showed considerable variability in their weight gain in the year prior to referral, ranging from -0.16 to 32.89 kg/year (median 4.69 kg/year). This suggests referral may serve as a critical intervention point in altering weight trajectories in a subset of patients, particularly older females.

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P143

Clinical outcomes of GLP-1 receptor agonist therapy in paediatric patients in the Thames valley: a retrospective audit

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Introduction

The use of glucagon-like peptide-1 receptor agonists (GLP-1 RAs) in children and adolescents has emerged as a promising therapeutic strategy for managing obesity and type 2 diabetes mellitus.

Methods

Data were collected retrospectively from patients' records from Complication of Excess Weight Clinic (CEW), Paediatric Endocrine and Paediatric Diabetes Clinics at Oxford University Hospitals.

Results

Medical records of 28 patients (mean age 15.9 ± 1.6 years, 50% females) were analysed. Most patients were treated for primary obesity and five for secondary obesity to hypothalamic dysfunction (3-craniopharyngioma, 1-neurofibromatosis and 1-Langerhans cell histiocytosis). At the time of analysis, the mean treatment duration was 11.6 ± 6 months. The mean initial BMI z-score was 3.83 ± 0.6, which decreased to 3.54 ± 0.7. In those with hypothalamic obesity, initial mean weight was 100.6 ± 14 kg, reducing to 94 ± 12 kg at 3 months and 92 ± 12 kg after an average 13 months of treatment (8.5% total weight loss). Among patients with primary obesity, the initial mean weight was 113.2 ± 16 kg, decreasing to 104 ± 19 kg at 6 months and 101.3 ± 19 kg after 11.1 ± 5 months, with a total weight loss of 10.6%. The median semaglutide dose at analysis was 1.7 mg/week. In patients without diabetes, mean HbA1c decreased from 38.7 to 33.5 mmol/mol. In all patients with prediabetes, HbA1c normalized, indicating reversal of prediabetes. Liver function tests also improved during treatment in 30% of patients and in 1 patient severe sleep apnoea requiring BiPAP has reversed. Two patients had difficulties with compliance, resulting in initial weight loss followed by regain. Both are currently undergoing dose re-escalation. One patient discontinued treatment due to nausea and vomiting. Overall, 10% of the cohort reported nausea, and one patient reported possible sleep disturbances.

Summary

Our preliminary data suggest that GLP-1RAs is an effective and well-tolerated treatment option for children and adolescents with obesity, including those with complex aetiologies such as hypothalamic obesity. Improvements in BMI, HbA1c, liver function with greater weight loss among those with primary obesity

were observed. These findings support the role of GLP-1 RAs as part of a multidisciplinary approach to paediatric obesity management.

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P144

What are the components and outcomes of multidisciplinary weight management interventions for children with obesity? a systematic review

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Multidisciplinary weight management interventions are recommended for children with obesity due to its complex nature. However, there is wide variation between different multidisciplinary interventions in terms of healthcare professionals involved and their mode of delivery. It is important to explore what the main components are in these multidisciplinary interventions and the theories underpinning them to understand how they work. Similarly, it is important to explore what outcome measures are collected and if the interventions are effective at improving these outcomes. Previous systematic reviews have often limited the focus to one or two outcomes, exclude children with comorbidities or do not comment on underpinning theory.

Methods

Systematic searches of electronic databases (Embase, Medline, Ovid, Web of Science, Psycinfo and Cinahl) were conducted. Studies that evaluated a multidisciplinary intervention (delivered by a minimum of 2 healthcare professionals) for children with obesity were included.

Results

One hundred and sixteen studies were included. The studies included over 34,000 participants and were conducted in 27 countries (4 in UK). Intervention length ranged from 2 months to 36 months. Only 28 out of 116 studies were underpinned by theory and cognitive behavioural theory was most used. The average number of healthcare professionals involved was 4 yet only 55 of out 116 interventions included a psychology professional. Furthermore, only 17 studies measured any psychological outcomes (such as quality of life). The most common primary outcome was body mass index (BMI) which was used in 80 out of 116 studies (69%) and significant changes to BMI were found in 64 studies (80 %).

Conclusion

Of the large number of multidisciplinary weight management interventions that have been conducted, most (69%) focus on BMI as the primary outcome and only 14% included psychosocial measures despite 47% including a psychology professional as part of the intervention. Future interventions should aim to include more psychosocial measures as the primary outcome.

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P145

Does social care involvement in children and adolescents living with obesity influence weight loss and BMI?

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Introduction

Childhood and adolescent obesity are a rising global problem. Overnutrition leading to obesity may contribute to concerns regarding potential neglect prompting a social care (SC) referral. Approximately 18% of the 549 children and young people (CYP) managed by our weight management (WM) service have some form of SC involvement. We undertook an audit to determine whether SC involvement had a positive effect in CYP severely overweight under our service.

Methods

We identified 15 children in whom neglect was a concern and where their weight was a major, but typically not the only, contributory factor. Data were collected on weight and BMI at start of SC involvement, then at 6 and 12 months. Growth Analyser was used to calculate BMI SDS.

Results

The mean age when first referred to WM was 8.4yrs (60% female). Three CYP had ongoing SC input. The mean age of SC involvement when under the WM

service was 10.1yrs ($n = 15$) with a median BMI SDS of 4.2 (range 3.5-5.4), and a median BMI of 39.5 (Range 29.3 – 54.9) kg/m². After 6 months ($n = 14$), median BMI SDS was 3.9 (3.7-5.1), median BMI was 40.8 (28.5-57.5) kg/m². 43% demonstrated a decrease in BMI, with a mean decrease in BMI for this cohort of -2.6 kg/m². At 1 year ($n = 15$) median BMI SDS was 3.9 (3.3-5.2) and median BMI was 37 (27.2-55) kg/m². 40% demonstrated a decrease in BMI from the point of SC involvement with a mean decrease in BMI for this cohort of -4.5 kg/m². Table 1 below demonstrates mean values. Whilst receiving SC support ($n = 15$), 20% of our CYP were on Metformin, 20% on Orlistat and 13% on Semaglutide.

Discussion

Our data suggest that a year is needed to fully realise the benefit of SC involvement on BMI. Not all individuals lost weight. It was difficult to determine whether the magnitude of weight loss (BMI SDS -0.3) differed from our wider clinic population.

	Mean BMI(kg/m ²)	Change in Mean BMI(kg/m ²)	Mean SDS	Change in Mean SDS
Baseline ($n=15$)	39.5		4.3	
6 months after ($n=14$)	39.9	-0.4	4.1	-0.2
1 year after ($n=15$)	38.9	-0.6	4.0	-0.3

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P146

Group based weight management programmes in young people living with excess weight, do they work? an initial review

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Introduction

Young people (YP) living with excess weight engage poorly with group-based weight management programmes despite them being delivered holistically and offering valuable peer support.

Objective

To evaluate a structured group programme for YP in Complications from Excess Weight (CEW) service.

Methods

3, 90 min single-sex group sessions were delivered by a health and wellbeing practitioner (HWP) twice weekly for 8 weeks. Community venues facilitated attendance and reduced costs: £717 per group. Interactive delivery techniques were used, icebreaker activities, group discussions and practical activity demonstrations. Motivation, sleep, stress, anxiety, nutrition, and physical activity were discussed using psycho-education techniques and SMART goal setting to facilitate change. Participants completed pre- and post-intervention, WHO-5 Wellbeing Index, a five-item questionnaire assessing mood, energy, and interest over the past two weeks. Scale 0 to 25 higher scores show better wellbeing. They also completed an Exercise Confidence Scale, measuring self-efficacy related to physical activity.

Results

12 CYP (6 female; mean age 15 yrs (13–17) mean BMI Z score 3.6, enrolled out of 50 eligible YP (23 female; mean age 17 yrs (13-19) Z score 3.6. BMI remained static in both groups. 10 YP completed all/some the sessions. WHO-5 scores improved in 90%, increasing +1 to +6 points; one declined (-1 point). The two partial completers improved by +3 and +4 points. These changes exceeded the minimal clinically important difference (MCID) in previously published data (Topp *et al* 2015). Exercise confidence increased in all YP completing the groups. All completers finished 12 months in CEW, compared to 24% of YP not participating in groups.

Discussion

This pilot shows that a structured, group-based intervention is deliverable in this challenging population, improves wellbeing and physical activity confidence in YP living with excess weight. A single HWP with small groups is a holistic low-cost model, novel within CEW. These findings support the value of HWP group work, and warrant further evaluation of the impact on BMI and future scalability.

Reference

Topp, C. W., Østergaard, S. D., Søndergaard, S., & Bech, P. (2015). WHO-5 Well-Being Index: a systematic review of the literature. *Psychotherapy and psychosomatics*, 84(3), 167–176. <https://doi.org/10.1159/000376585>

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P147**OGTT testing in paediatric obesity: can fasting glucose and HbA1c guide us?**David J Cullingford^{1,2}, Munibah Bashir¹, Joanne C Blair^{1,3} & Senthil Senniappan¹¹Department of Endocrinology, Alder Hey Children's Hospital, Liverpool, United Kingdom; ²University of Western Australia, Perth, Australia;³Honorary Professor, Faculty of Health and Life Sciences, University of Liverpool, Liverpool, United Kingdom**Objective**

Whilst oral glucose tolerance tests (OGTT) are routinely used to assess for diabetes mellitus and impaired glucose tolerance (IGT) in paediatric obesity, they are costly, time consuming, can be unpleasant and have poor reproducibility. Some studies have suggested lower fasting plasma glucose (FPG) and HbA1c thresholds for predicting diabetes/IGT risk. We assessed the FPG and HbA1c thresholds with the greatest sensitivity for detecting IGT on OGTT.

Method

Retrospective analysis of children undergoing OGTT at a single paediatric tertiary institution from 2020-2025. Patient age, body mass index (BMI) standard deviation score (SDS), ethnicity, FPG, 2-hour glucose and HbA1c were recorded. IGT was defined as 7.8-11 mmol/l and hyperglycaemia as ≥ 11.1 mmol/l. Incremental changes in FPG and HbA1c cut-offs for screening were assessed for corresponding sensitivity and specificity for detecting an abnormal OGTT result.

Results

OGTT's from 307 patients, (150 female, mean age 12.9 +/- 3.2 years) were paired with HbA1c, with 11.2% having IGT or hyperglycaemia. In participants under 10-years-old, 1/56 had hyperglycaemia, with an HbA1c of 54 mmol/mol (diabetes range). For those over 10-years-old, the highest sensitivity threshold was HbA1c ≥ 35 mmol/mol or FPG ≥ 5 mmol/l, giving 100% sensitivity (specificity 35.6%) for those aged 10-13 years, and 84.2% sensitivity (specificity 40%) ≥ 14 years. All cases of hyperglycaemia, and all but 3 cases of IGT screened positive (Table 1). BMI SDS and ethnicity were not significantly different between those with and without IGT.

Conclusion

Routine screening with OGTT has a low yield in those under 10-years-old. IGT is uncommon in those without FPG < 5 mmol and HbA1c < 35 mmol/mol, particularly in those under 14-years-old. Reserving OGTT for those with FPG/HbA1c above these thresholds or strong clinical suspicion of diabetes could be considered, reducing testing by approximately one third. Prospective studies to validate these thresholds are recommended.

Table 1: OGTT results

	Negative screen			Positive screen			p
	Normal	IGT	Hyper-gly-caemia	Normal	IGT	Hyper-gly-caemia	
≥ 10 years (n=251)	83	3	0	136	22	7	< 0.001
10-13 years (n=117)	37	0	0	67	10	3	0.009
≥ 14 years (n=134)	46	3	0	69	12	4	0.042

P-value reflects difference in frequency of abnormal OGTT based on positive or negative screens (chi-square analysis).

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Thyroid**P148****Congenital hypothyroidism with thyroid dysmorphogenesis- a case report**

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Introduction

Worldwide incidence of Congenital Hypothyroidism is 1:3000 to 1:4000 births. 80-90% of cases of Congenital Hypothyroidism are related to defects in thyroid gland development. 10- 20% related to thyroid hormone biosynthesis due to mutations in genes involved in thyroid hormone synthesis with autosomal recessive inheritance pattern. The steps in thyroid hormone synthesis include iodine trapping, synthesis and secretion of thyroglobulin, oxidation of iodide, organification of iodides, coupling reaction and storage and secretion. Enzyme defect in any of these steps can lead to dysmorphogenesis, with implications of genetic testing in Congenital Hypothyroidism.

Case

16-day old baby referred from Newborn Screening lab with abnormal Guthrie test. TSH on day 5- 13 mIU/l Day 12- 10.7mIU/l. Term baby girl with normal clinical examination and gaining weight. Parents from Pakistan, non-consanguineous marriage. Mother takes Thyroxine for Autoimmune Hypothyroidism, which was diagnosed 5 years ago with previous pregnancy. Thyroid function test done on day 16- TSH 10.85mIU/l (0.38-5.33) T4 14 pmol/l (11.5-22.7). Patient was not started on treatment at this stage. Repeat blood test after 2 weeks showed TSH normalised at 4.43 mIU/l. Working diagnosis- Transient Hypothyroidism. Patient had a normal clinical examination, including development and normal thyroid ultrasound scan. Thyroid function monitored every few weeks. TSH normalised quickly, but due to persistently borderline low T4, both parents consented for genetic blood testing. The patient is heterozygous for a pathogenic *duox2* variant. All *duox2* gene variants limit the ability of the enzyme dual oxidase 2 to generate hydrogen peroxide. Without hydrogen peroxide, thyroid production is disrupted. The patient is a carrier of Thyroid Dysmorphogenesis type 6.

Conclusion

There is a low threshold to do genetic testing in Congenital Hypothyroidism, especially with Transient Hypothyroidism. Identification of asymptomatic mutation carriers has prognostic value in differentiating from transient to permanent hypothyroidism and monitoring for associated features such as hearing loss in Pendred's syndrome. Thyroid Dysmorphogenesis should be suspected in foetal/neonatal goitre, normal thyroid scan but needing thyroxine replacement, enlarged thyroid gland with low thyroglobulin or blunted/absent uptake in scintigraphy in normal located gland.

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P149**Time for standardised universal reference range for thyroid function test (TFTs): A study of impact of inter-laboratory variation in TFT results on clinical decision making**Hannah Bidwell¹ & Sunil Goyal²¹Cardiff University, Cardiff, United Kingdom; ²Aneurin Bevan University Health Board, Newport, United Kingdom**Background**

There is no universal reference range for thyroid function tests (TFTs), and laboratories adopt different ranges based on their assay method. However, discrepancies between assay methods used across different health boards can lead to variability in TFT interpretation, potentially impacting clinical decision-making. This is not only affecting patient care, but also incurs an additional cost.

Aim

To evaluate the frequency and nature of discrepancies in TFT results between two different inter-laboratory assay methods, and whether these differences have led to a change in clinical management.

Method

A retrospective review of thyroid function data from a large university health board database was conducted. TFTs originally analysed using a particular assay method and subsequently re-evaluated using a different method at an adjacent university health board were identified. Discrepancies in results and any changes in clinical management were recorded. A chi-squared test was used to assess statistical significance.

Results

64 TFT results were referred for re-evaluation. 34% showed differing outcomes between the two assay methods. 100% of these cases resulted in a change to the clinical management plan. 14 patients were hyperthyroid and 8 were hypothyroid under the initial assay method, but all were reclassified as euthyroid using the different assay method at the adjacent university hospital, avoiding unnecessary medication changes. The association between discrepant results and altered management was statistically significant with a p-value < 0.001 . Analysis of the re-evaluated values showed that discrepant cases are usually due to a result that is only marginally outside of the reference range. For TSH, values up to 0.37 below or 1.63 above the reference range were likely to be reclassified as normal when re-evaluated. For FT4, all raised results below 21 pmol/l were reclassified as normal.

Conclusion

Assay variability has a significant impact on the interpretation of TFTs and consequent clinical management in paediatric patients. Re-evaluation delays patient care and has cost implications. Borderline results are particularly susceptible; hence we recommend standardisation of TFT reference ranges irrespective of assay method.

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P150**Case report: an autonomously functioning thyroid nodule in a 13-year-old**

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Introduction

Thyroid nodules are less common in children than adults, however they have an increased risk of being malignant compared to adults. Follicular adenomas are the most common benign thyroid lesion in children. They present twice as frequently in girls, and the average age at presentation is 12.5 years. Case reports predominantly document cold benign nodules in children. We present the case of a solitary thyroid nodule due to hyperfunctioning follicular adenoma in a 13-year-old girl.

Case

A 13-year-old girl was referred with an incidental finding of a left-sided thyroid swelling. There were no symptoms of thyroid dysfunction. She had no significant past medical history or family history of thyroid disorders or malignancy. On review, she was clinically euthyroid, however she had a smooth, firm, non-tender, solitary, left-sided thyroid mass measuring 5x4 cm which was mobile on swallowing. No lymphadenopathy was evident. Biochemical assessment showed

evidence of mild hyperthyroidism with a suppressed TSH 0.02 mIU/l(0.5-4.88), raised FT3 9.6 pmol/l(3.9-7.70) and a normal FT4 14.6 pmol/l(11.2-21.3). Thyroid peroxidase (TPO) antibodies and TSH receptor antibodies (TRAB) were negative. An ultrasound scan revealed a large nodule filling the entire left lobe of her thyroid, with a volume of 26ml. It contained cystic spaces separated by soft septae and peripheral solid components. The right lobe appeared normal. Per technetate thyroid scan showed heterogenous and intense uptake of iodine-123 within the left lobe of thyroid with faint homogenous uptake in the right lobe suggestive of a hyperfunctioning left-sided thyroid nodule. The patient underwent a left hemithyroidectomy. Histology found a cystic nodule measuring 50mm suggestive of a follicular adenoma, with complete excision. The surrounding thyroid tissue was unremarkable. Post operatively, she had transient clinical and biochemical hypothyroidism which resolved without treatment over 2 months.

Conclusion

Autonomously functioning hot nodules in children are rare. Although often benign, they can, on occasion, be malignant. It is not always possible to make this differentiation on ultrasound. Surgical management can definitively treat thyroid dysfunction in these cases and avoid the need for long-term medication whilst also allowing a definitive histological diagnosis to be made.

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