Endocrine and metabolic profiles in adults with Prader-Willi syndrome

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OBJECTIVES

Prader-Willi syndrome (PWS) is a genetic syndrome usually diagnosed in childhood.
Its reported prevalence ranges from 1 in 8000 to 1 in 45000 with geographical variation.
Clinical manifestations include obesity, hyperphagia, short stature, incomplete sexual development, and cognitive disabilities.
The majority of published data regarding PWS comes from paediatric populations.

METHODS

This is a retrospective observational study of adult patients with PWS performed in an Irish tertiary referral centre. We collected anthropometric measurements, hormonal data, results of genetic testing, and clinical data regarding co-morbid conditions, psychological and social circumstances.

RESULTS

Twenty-two adult patients (15 women) with a diagnosis of PWS were identified.
The median age was 24.5 years. Median height and BMI was 154.3 cm and 41.7 kg/m^2, respectively.
Eighteen of patients were assessed for growth hormone deficiency (GHD), 15 using the insulin tolerance test. Sixteen (88.2% of the patients tested) had severe GHD. Fifteen received GH therapy. Height velocity was 4 cm in the first 6 months and 9 cm in the first year of therapy.
Fifty percent of patients had evidence of obstructive sleep apnoea; 90.9% had spinal scoliosis; 81.8% had hypogonadism; 50% of those who had a DXA scan had osteoporosis (n=5).
Fifty percent of patients had abnormal blood pressure, 22% had lipid abnormalities and 18% had an abnormal HbA1c. 21/22 had learning disabilities and 50% had associated psychiatric diagnoses.
There was no difference in BMI, height, lipids, fasting glucose between GH treated and untreated patients.

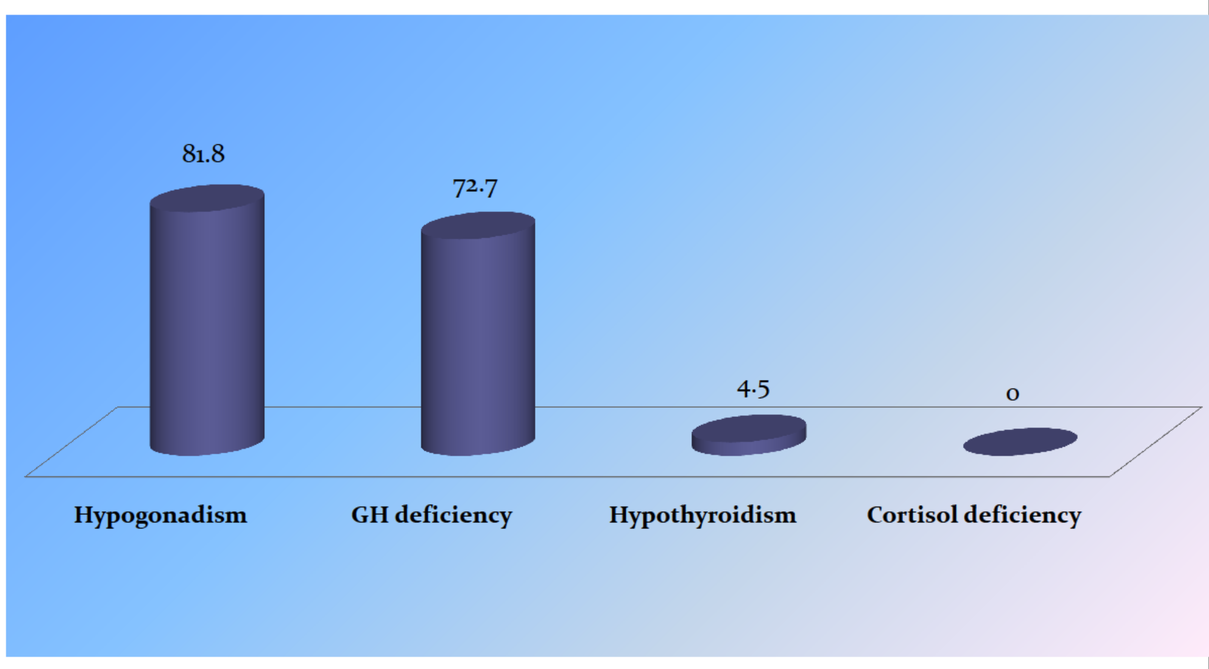

Fig.1 Endocrine profiles in adults with Prader-Willi syndrome

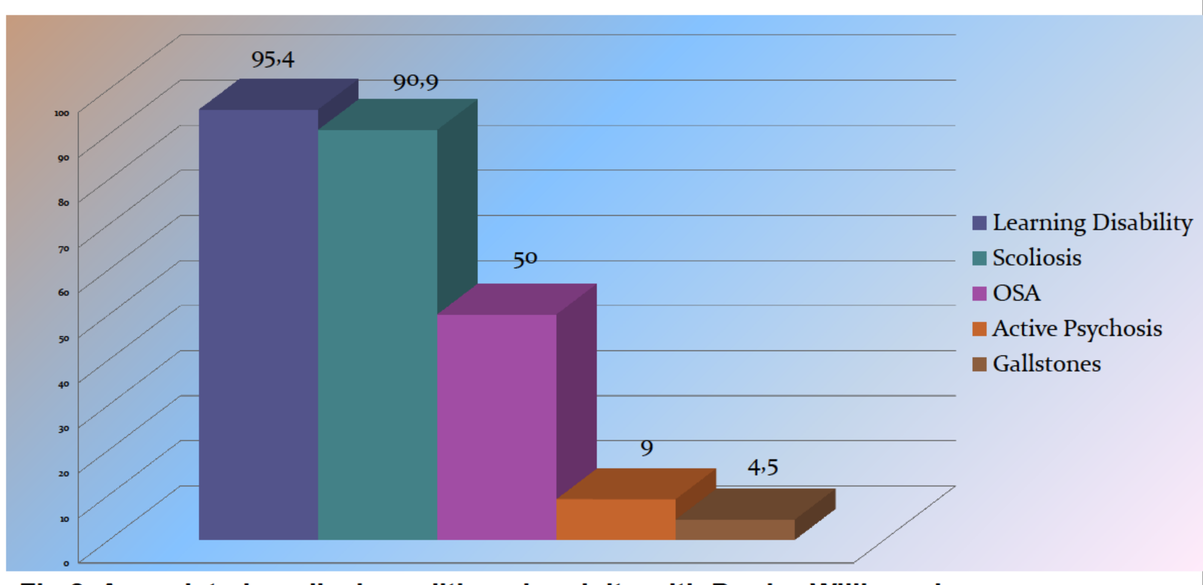

Fig 2. Associated medical conditions in adults with Prader-Willi syndrome

CONCLUSIONS

Adult patients with PWS have multiple endocrine abnormalities and require careful follow up and management. Early diagnosis and management of endocrine manifestations will potentially improve health and developmental outcomes in adulthood.

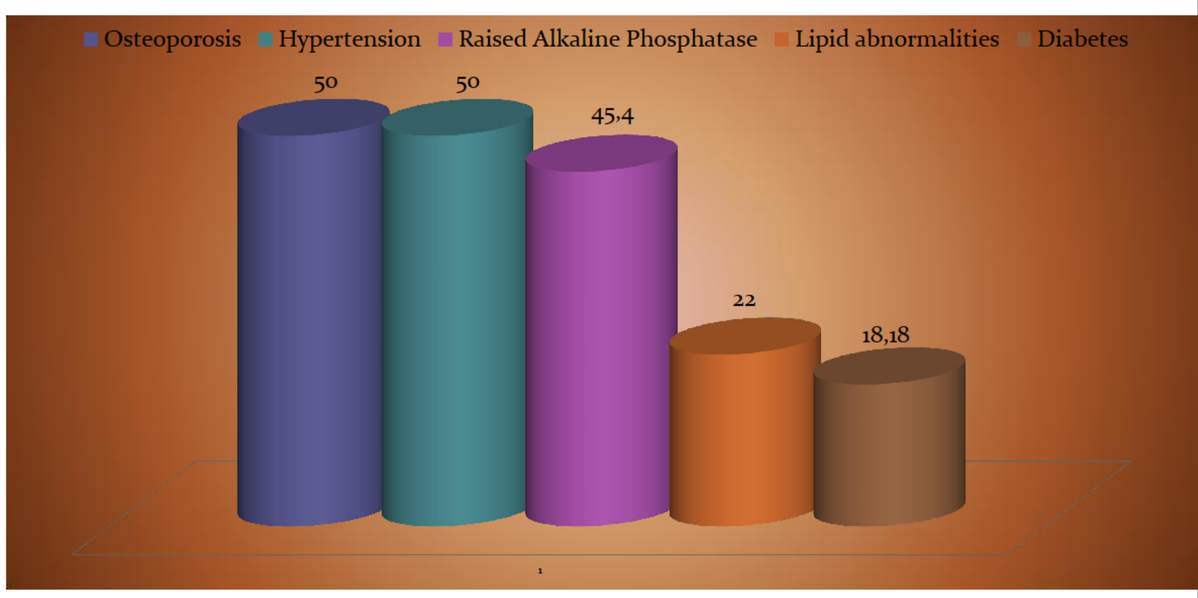

Fig. 3 Metabolic profiles in adults with Prader-Willi syndrome

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