Initial care of babies born with ambiguous genitalia: a service evaluation

Wijeyaratne L [1], Nicoll N [2], Alderson J [2], Burren CP [2], Barton J [2], Crowne EC [2]

[1] University of Bristol, Medical Sciences Building, University Walk, Bristol BS8 1TD
[2] Bristol Royal Hospital for Children, Upper Maudlin St, Bristol BS2 8BJ, United Kingdom

Introduction

- Disorders of sex development (DSD) may present as ambiguous genitalia in the newborn.
- It is a very stressful time for parents. [1,2]
- Expert multi-disciplinary team input is essential to expedite diagnosis and gender determination, and manage any significant health issues.
- The long-term impact on the child's physical, emotional and sexual development are paramount.
- Management guidelines are currently based on expert clinicians' opinions. [3]

Aims

- To assess the initial management of babies presenting with ambiguous genitalia in a tertiary centre for DSD.
- To identify areas for improvement in this centre's provision of holistic initial care for neonatal DSD.
- To recognise opportunities to contribute to local/national evidence-based guidelines.

Methods

- A retrospective analysis of patient notes was carried out to assess initial care for DSD.
- 18 consecutive newborns with DSD were referred to a tertiary centre between January 2012-June 2014.
- The care pathway recommended by the regional DSD Team is presented in **Figure 1**. [10]
- The following parameters from the DSD team care pathway were used to judge the standard of care: time to refer, transfer, assess, test the karyotype, hormone profile, 17-OHP and urinary steroid profile (USP), and time to determine gender and diagnosis.
- All healthcare providers involved, and all documentation of management and communication with parents, were noted.

Results

- Two patients were initially diagnosed in separate centres, and three case notes could not be traced. Thirteen patients were included in the final analysis **(Table 1)**.
- The presentations of DSD were: bilateral impalpable testes (7), bilateral impalpable testes with penile hypospadias (1), ambiguous genitalia (4), micropenis (1).
- For six patients born at the tertiary centre, the parameter 'Time to transfer to DSD team' was inapplicable.
- Babies who presented with ambiguous genitalia were assessed in <1 day and the median time to send a sample for karyotyping was 1 day (<1-3).
- For babies presenting with bilateral impalpable testes, time for referral, and therefore assessment and karyotyping were delayed.
- 5 babies had multiple congenital anomalies **(Figure 2)**. Time to referral, assessment, sending the sample for and receiving the result of the karyotype and determining gender were all increased. Time to transfer was not used because 3 patients were born at the tertiary centre.
- Assessments and investigations were delayed for babies born on a Friday or a weekend.
- The DSD nurse specialist and clinical psychologist were involved only in the care of 3 patients diagnosed with CAH.
- Communication with parents was documented in all cases.
- Additional diagnostic issues are noted in **Figure 3**.

Figure 1. Initial care pathway for neonates presenting with ambiguous genitalia

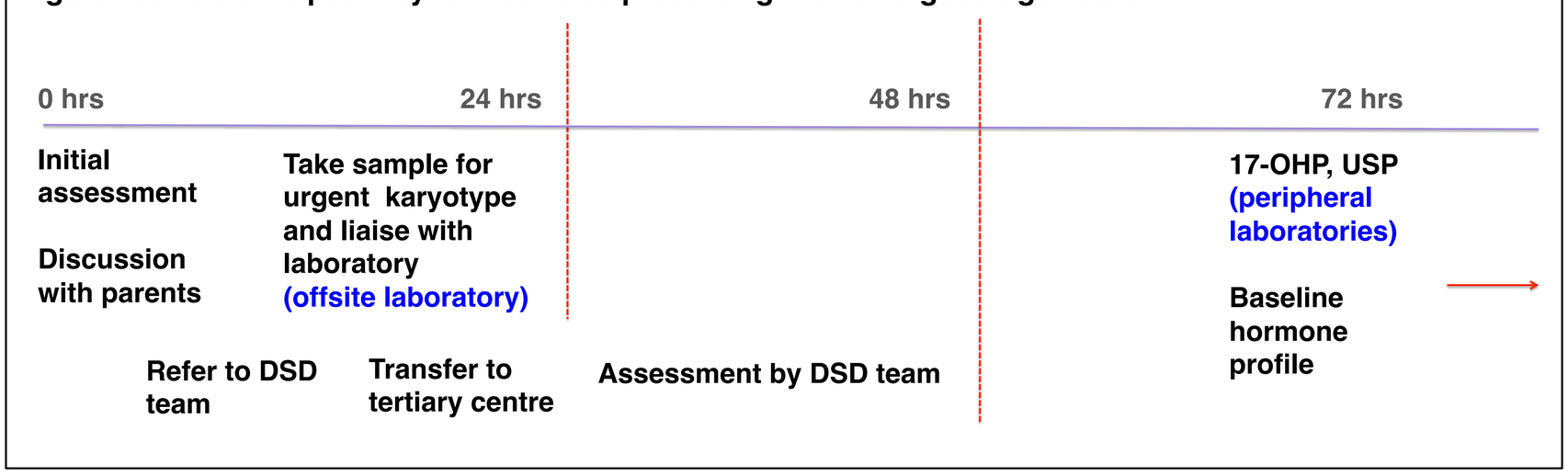


Table 1. Median (range) number of days to investigate DSD in overall group (n=13)

Parameter	Median number of days	Range
Time to refer to DSD team	<1-1	<1-3
Time to transfer to tertiary centre (7 patients)	1	<1-4
Time for DSD assessment	<1-1	<1- 4
Time to send sample for karyotype	1	<1-6
Time to receive karyotype result	3	1-8
Time to test hormone profile	4	3-8
Time to receive hormone results	1	<1-3
Time to test for 17-OHP	4-5	3-8
Time to receive 17-OHP result	10	2-19
Time to test urinary steroid profile	8	3 days-6 weeks
Time to receive urinary steroid profile	3 weeks	2 days – 2 months
Time to determine gender	5	2-12
Time to determine diagnosis	5	2-12

(17-hydroxyprogesterone=(17-OHP)).

Figure 2. Median number of days (range) taken to investigate babies with DSD only vs. babies with DSD and multiple congenital abnormalities

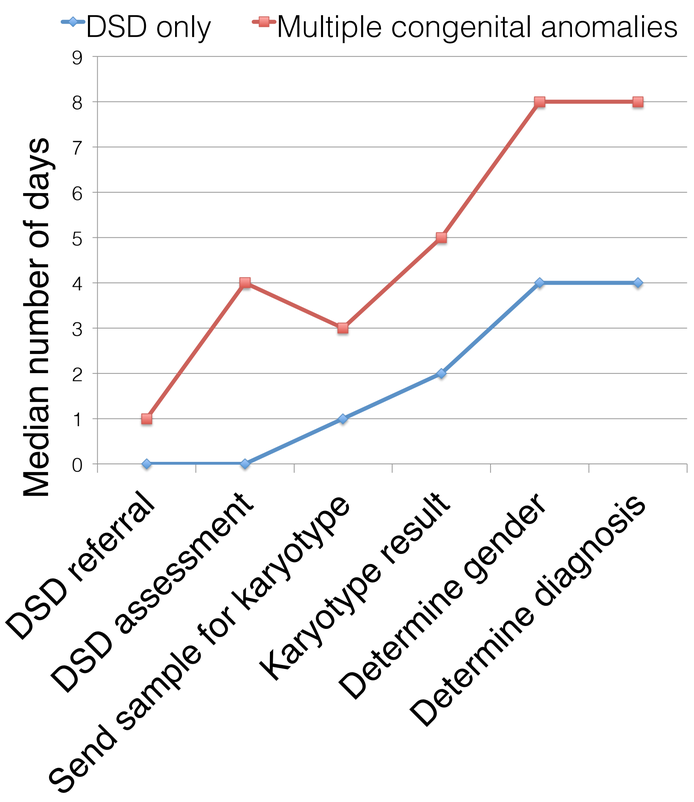
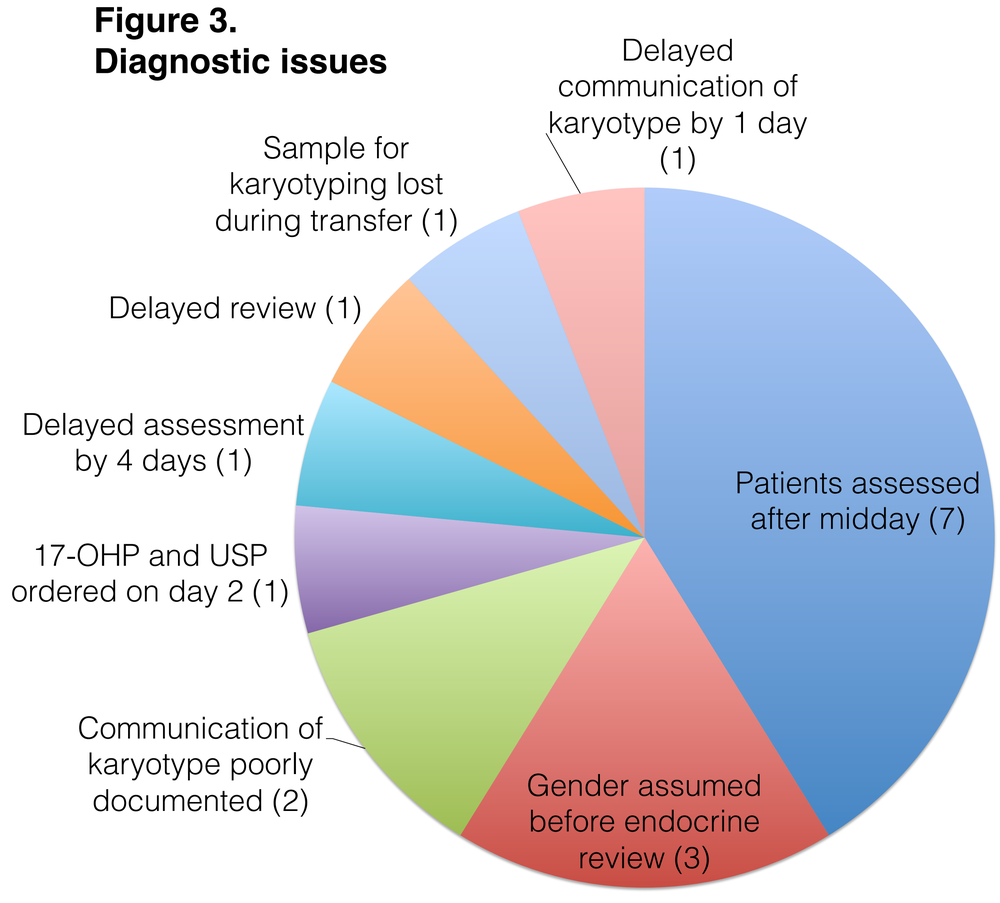


Figure 3. Diagnostic issues



Conclusions

- Times for referral, transfer and initial assessment by the DSD team were reasonable, given the regional geography and need for transfers to the centre.
- Reasons for delayed referral for babies born with bilateral impalpable testes should be investigated and rectified.
- There is room for improvement in the time taken to test samples for a karyotype, hormone profile and 17-OHP.
- Efforts should be made to expedite referral and management for babies born with multiple congenital anomalies.
- The same standard of care should be ensured throughout the week
- A DSD Clinical Nurse Specialist and Clinical Psychologist should be available for all babies presenting with DSD.

References

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