

A Nationwide Study of the Prevalence & Initial Management of Atypical Genitalia and Delayed Sex Assignment in the Newborn

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Introduction

- The prevalence of atypical genitalia and the time taken to assign sex in such cases remains unclear.
- Provision of optimum healthcare during this period requires a clear understanding of the occurrence of atypical genitalia.

Aims

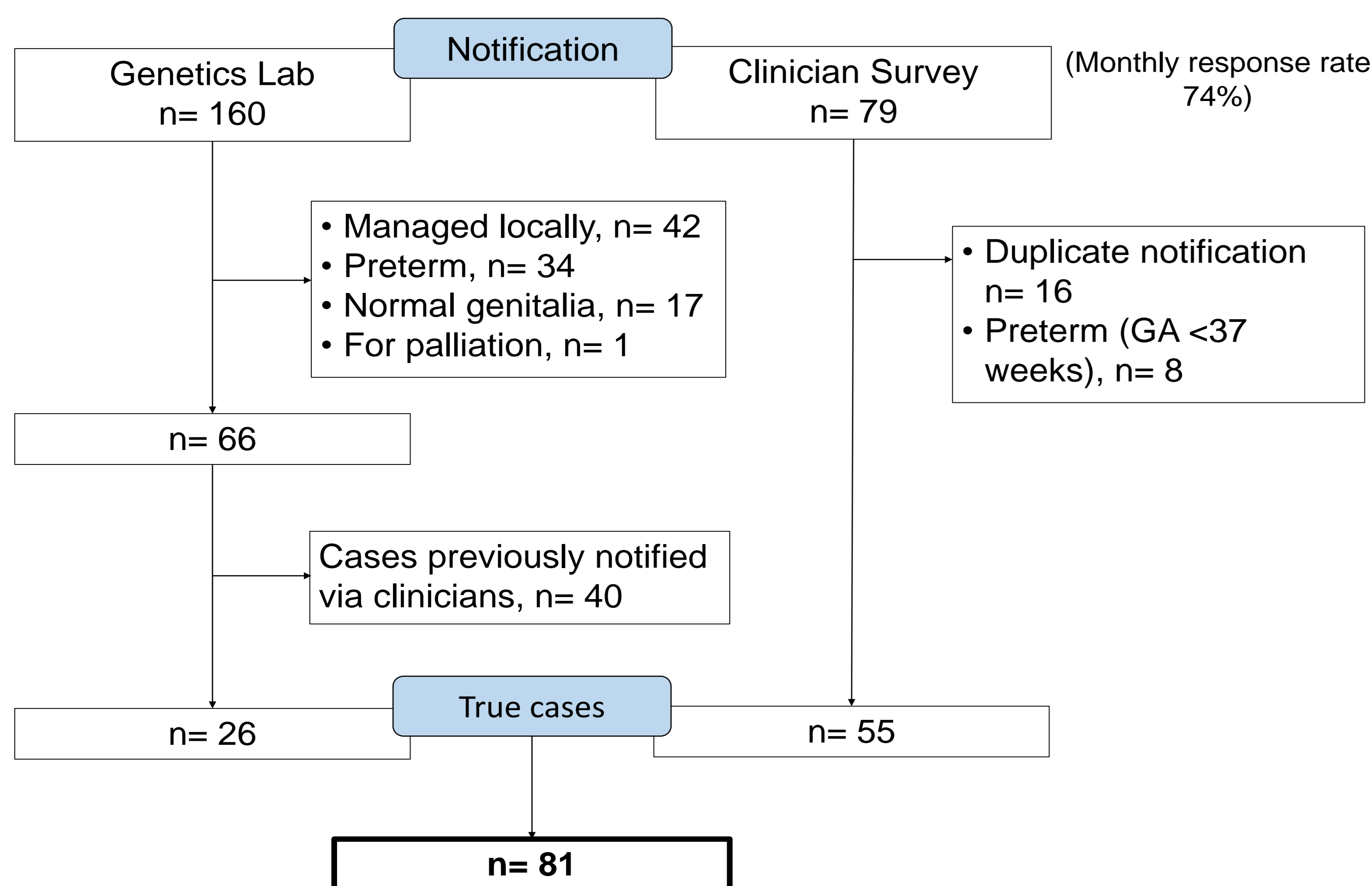
- To determine the prevalence of atypical genitalia in term newborns requiring specialist input.
- To determine the time taken to assign sex in cases of atypical genitalia.

Methods

- Prospective, electronic monthly survey of clinicians within two managed clinical networks in Scotland between 2013-2018.
- Notification from clinicians for term neonates (GA ≥37 weeks), followed up to age 3 months and requiring specialist input for atypical genitalia at <4 weeks of age.
- Cross-verification of notification through 4 regional genetics labs, using karyotype as a surrogate marker with request codes 'genitalia' or 'sex' to identify additional newborns with atypical genitalia.
- Positive notifications followed up by a questionnaire sent to clinicians enquiring about patient's care.

Results

Case notification

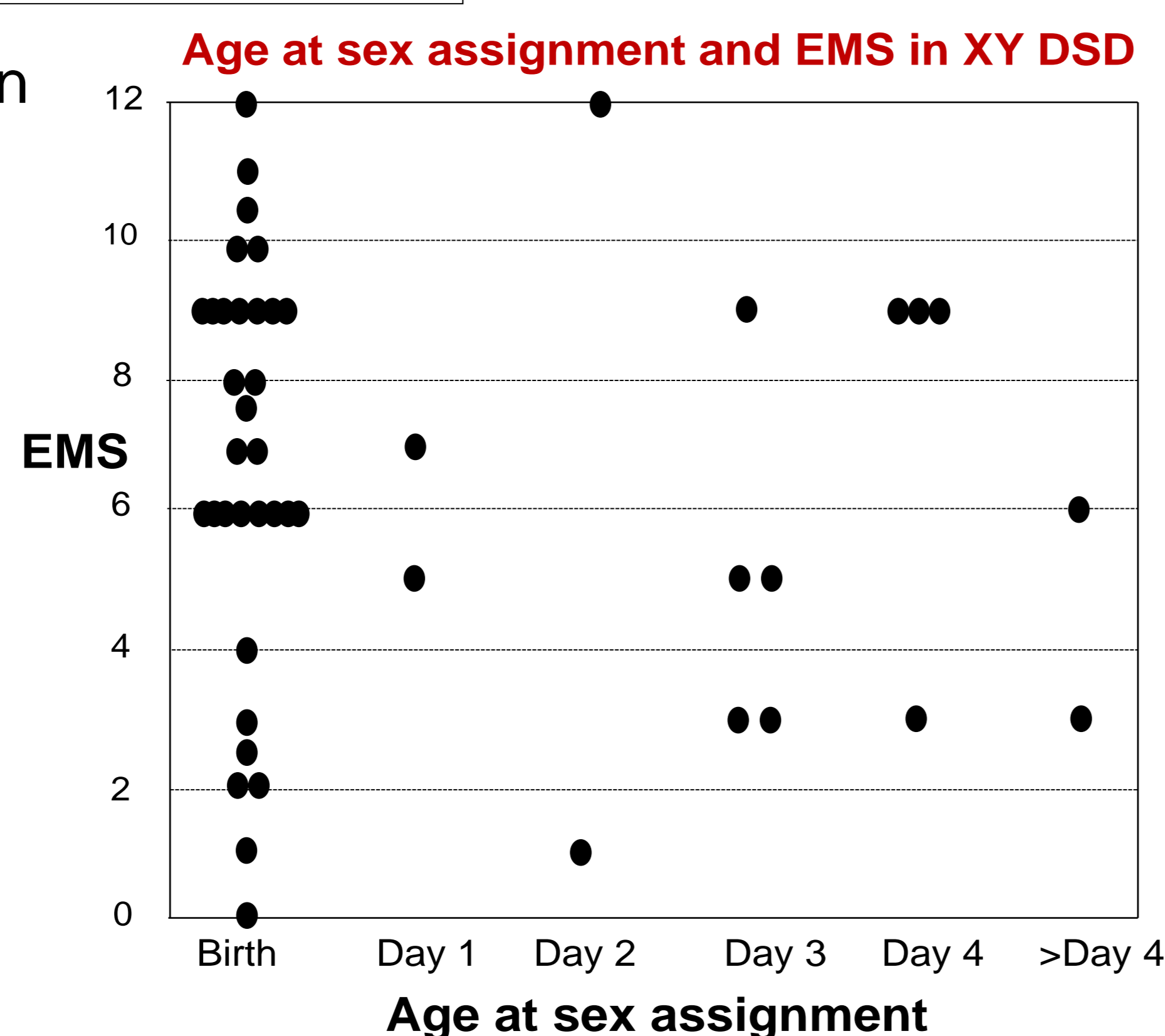


→ Birth prevalence of 1 in 3,378 term births

(calculated using annual live birth rate of 54,000 births in Scotland over the duration of survey as the denominator)

Age at sex assignment and EMS

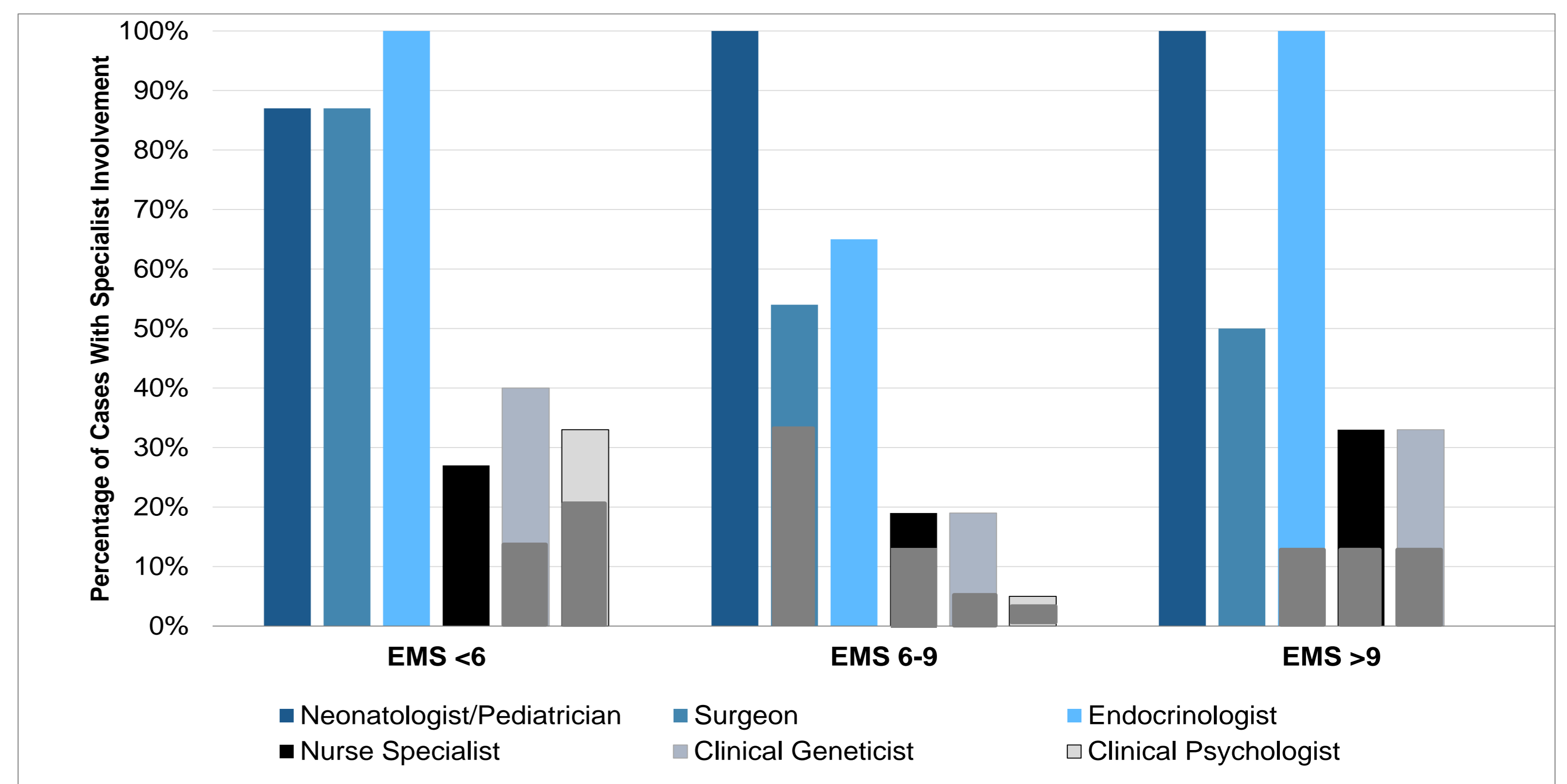
- 49 (64%) presented within 24 hrs of birth
- Age at presentation - birth to 28 days
- 51 (66%) cases assigned sex at birth
- Age at sex assignment - birth to 14 days



Results

- Prevalence of a case of atypical genitalia where sex assignment delayed beyond birth ~ 1 in 10,524 births.
- No significant difference in median (range) EMS between the group assigned sex on Day 3 or later compared with those assigned sex at an earlier age: 5 (3, 9) versus 7.3 (0, 12), p=0.19.

Involvement of MDT in patient care



*Grey section of each bar represents the proportion of infants who had delayed sex assignment after birth

- In infants with XY DSD, the median number of health professionals encountered by those cases with an EMS <6, EMS 6-9 and EMS >9 was 4, 2 and 3 respectively.
- The median EMS of the infants who met a psychologist and who did not meet a psychologist was 3 (2, 9) and 7.8 (0, 12), respectively (p=0.06).

Investigations performed in the XY infant

| | Percentage of cases (n, 59) | Median age at test in days (range) | Median EMS (range) |
|------------------------------|-----------------------------|------------------------------------|--------------------|
| Urea & electrolytes | 80% | 2 (1, 5) | 7 (1, 12) |
| Testosterone | 69% | 4 (1, 86) | 6 (0, 12) |
| Ultrasound scan of pelvis | 75% | 3 (1, 58) | 6 (0, 12) |
| Anti-Müllerian hormone | 59% | 3.5 (1, 30) | 6 (0, 10.5) |
| Androstenedione | 54% | 4 (1, 86) | 6 (2, 12) |
| Urinary steroid analysis | 37% | 5 (1, 46) | 6 (1.5, 12) |
| Cortisol | 39% | 3 (2, 11) | 6 (3, 9) |
| Glucose | 32% | 1 (1, 3) | 6 (1, 12) |
| Luteinizing hormone | 29% | 3 (1, 77) | 5 (0, 9) |
| Follicle stimulating hormone | 29% | 3 (1, 77) | 5 (0, 9) |
| Dihydrotestosterone | 17% | 5 (3, 86) | 6 (2, 9) |
| HCG stimulation | 12% | 33 (1, 89) | 6 (3, 10) |
| ACTH stimulation | 15% | 6.5 (3, 27) | 6 (1.5, 9) |

Conclusions

- Atypical genitalia requiring specialist input within the first month of life is rare in term newborns.
- In one third of cases, sex assignment was delayed beyond birth.
- Delayed sex assignment is not necessarily related to EMS.
- MDT input, especially psychology, is more likely to occur in those with a lower EMS and delayed sex assignment.
- This study provides further benchmarks for comparing and improving the delivery of care for DSD.