Introduction

- Disorders of sex development (DSD) may present as ambiguous genitalia in the newborn.
- It is a very stressful time for parents.  
- 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.  

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.  
- 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 perineal hypospadas (1), ambiguous genitalia (4), micropenis (1).
- For six patients born at the tertiary centre, the parameter ‘Time to transfer to DSSD 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 to 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.

Discussion

- It is a very stressful time for parents.
- The long-term impact on the child’s physical, emotional and sexual development are paramount.
- Expert multi-disciplinary team input is essential to expedite diagnosis and gender determination, and manage any significant health issues.
- Management guidelines are currently based on expert clinicians' opinions.  

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

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

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