Adolescent girls with disorders of sex development: A needs analysis of transitional care

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KEYWORDS
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Abstract
Objective: To collect information on clinical concerns relating to adolescent girls with disorders of sex development (DSD) during the process of transition from paediatric-to-adult clinical services.

Subjects and methods: This was a prospective audit of the clinical indications for referral and ongoing clinical needs for all girls aged 12–20 years seen in a specialist DSD clinic over a 6-month period. Clinical needs were classified according to level of urgency using a simple ‘traffic light’ classification: green for low, amber for moderate, and red for high.

Results: Fifty girls were seen during the study period and all were referred from paediatric services. Patients may have had one or more indication(s) for referral to the adult clinic and these were: urology/gynaecology (70%), endocrinology (42%) and psychology (14%). The most common indication for on-going clinical input was psychology, with 46% of patients requiring monitoring and intervention. Of the 14 patients (28%) classified red suggesting they had an urgent clinical need, psychology was a major factor in all but one patient.

Conclusion: Clinicians working with adolescents with DSD need to develop a co-ordinated programme for transitional care that recognises the importance of psychological input within the multi-disciplinary team.

Introduction
As more and more young people with serious and chronic medical conditions survive into adulthood, there has been an expansion of publications documenting the barriers to smooth transition from paediatric-to-adult care. These publications include general [1,2] as well as disease-specific reports on topics such as cystic fibrosis [3], haemoglobinopathy [4], congenital cardiac disease [5] and palliative care [6]. Some of the key issues identified by clinicians working with adolescents across a range of clinical specialties [7] are autonomy, information coordination and appropriate professional support, but with an increasing emphasis upon their own personal resources to
successfully access the healthcare system as independent young adults.

Transition from paediatric-to-adult care is a key indicator of service quality, and poor transition has been implicated in non-compliance with follow up [8]. Although problems of transitional care are often discussed thoughtfully, few reports offer evidence to inform service development and resource allocation. From the available literature, transitional care is perhaps most advanced in rheumatology, where planned programmes have led to significant improvement in quality-of-life indicators [9,10], although examples of good practice are emerging from other specialty groups [11].

Particular care in managing transition is needed for the group of conditions known as disorders of sex development’ (DSD) [12]. Medical management of DSD has been controversial for 50 years. Whilst there is now a consensus statement on how best to care for infants born with a DSD diagnosis and their families [12], the needs of patients on approaching adolescence and adulthood have not been articulated in any detail. So, despite clinicians’ commitment to improve patient experiences, there is little information to help them to design transitional care.

In general, adolescence is a time of rapid physical and psychological change [13]. For individuals and close others affected by a combination of atypical sex anatomy, infertility, steroidal deficiencies, atypical karyotype and heredity, there are significant additional social and emotional challenges. Unmanaged negative impact on identity, self evaluation, emotional well being and behavioural responses [8] may rebound on compliance. Physical health risks relating to non-compliance with investigations and treatments include osteoporosis and malignancy. Psychological risks include unexpressed distress arising from gender insecurity and fear of negative evaluation [14,15]. Where difficulties persist in family and friendship networks, the adolescent may be at risk of social isolation leading to episodes of depressed mood and/or behavioural disturbance.

This report is a first methodical attempt to collect information on clinical concerns relating to adolescent and young adult women with DSDs. The aim was to clarify what may be appropriate initial multi-disciplinary clinical responses for paediatric-to-adult care transition in the field of DSD.

**Method**

We performed a prospective audit of all adolescent girls and young women (aged 12–20 years) seen in the adolescent DSD service at our centre within a 6-month period. The sample included newly referred patients and those already under follow up. The audit protocol was developed by the multi-disciplinary team of physicians, psychologists and surgeons working in our affiliated paediatric and adult DSD services.

Information was collected on which clinician had referred the patient to the adult service and at what age. The reason for the initial clinical referral was taken from the transition referral letter and the clinical notes from the first consultation. The key clinical requirements at the time of the audit were recorded by the clinician completing the audit protocol and classified into three main areas:

- endocrine management (e.g. induction of puberty, oestrogen therapy, bone density monitoring);
- gynaecological and/or urological requirements (e.g. examination under anaesthetic, vaginal dilation, reconstructive surgery);
- psychological support for managing emotional and social effects (e.g. concerns about disclosing diagnostic and treatment information, investigations and treatments, and relationships and sexual intimacy).

Clinicians were asked to classify the urgency of the current clinical need using a ‘traffic lights system’ as follows:

- green — low level of professional concern (e.g. on appropriate HRT, regular monitoring taking place, no current indication for surgery);
- amber — potential concern (e.g. examination under anaesthetic pending, possible surgery, may require psychological support);
- red — urgent clinical input required (e.g. non-compliance with clinical advice/treatment/reviews, suspicion of tumour in gonads, menstrual obstruction, significant psychological distress/risk).

Red entries were discussed at the weekly DSD multi-disciplinary meeting. All entries were amalgamated on an MS Excel spreadsheet.

**Results**

Fifty adolescent girls with a mean age of 17.8 years (range 13–20 years) were seen by the DSD service in the 6-month audit period (October 2009–March 2010). All were referred for transition from paediatric services and the mean age at the first referral for transition was 16.3 years (range 12–20 years). The diagnoses were: complete or partial androgen insensitivity syndrome (n = 17), congenital adrenal hyperplasia (CAH) (n = 16), XY gonadal dysgenesis (n = 9), Mayer–Rokitansky–Küster–Hauser syndrome (n = 2), Turner’s XY mosaic (n = 2), cloacal extrophy (n = 2), ano-rectal anomaly with vaginal agenesis (n = 1), unknown diagnosis under investigation (n = 1).

The main clinical reasons for the first referral are given in Table 1. The clinical needs as assessed by the clinician at time of audit and the traffic light status are given in Table 2. Fifteen patients (30%) were given green status meaning that there were no outstanding clinical issues and that they were compliant with routine monitoring. Fourteen patients (28%) were given red status. Of these, 11 patients had psychological issues only, and one had endocrine issues only (poor metabolic control in pregnancy). Two had psychological issues affecting compliance with CAH medication leading to poor endocrine control.

**Discussion**

Where there is a chronic medical condition, the young person is expected to gradually assume responsibility for their own health care. Tasks such as arranging and attending appointments, interacting with clinicians,
processing the meaning of the diagnosis, and retaining and implementing treatment information are gradually transferred from the parent(s) to the adolescent. For young people with DSDs, there may begin a lifelong process of coming to terms with having an atypical sex anatomy and perhaps karyotype, infertility and dependence on exogenous hormones. Many of the young people will want to begin to actively negotiate social and intimate relationships, in spite of potential barriers. For their clinicians, the challenge of identifying and meeting clinical needs may intensify in the transitional years. Many clinicians will want to rethink care protocols and resources to advance quality care, often in a context of rigid organizational constraints. The successes and difficulties at both sides of the counter are as yet unknown.

Clinical management of DSD has had an inglorious past, and many lessons have been learned from adult patients who have struggled throughout childhood and young adulthood in a world of secrecy and misinformation and non-consensual treatments [16,17]. Modernized clinical guidelines [18] emphasise multi-disciplinary input, professional and parental collaboration, stepped disclosure tailored to the developmental stage of the child [19], and patient autonomy. Full disclosure of diagnosis with psychological support is recommended [20] and this often takes place in adolescence. If genital reconstruction is required to permit sexual intercourse, this is often performed during adolescence [21], and education and support are integral to these psychologically and/or physically invasive interventions.

This audit has documented broad clinical concerns and needs. Surgical examinations and interventions are episodic. Endocrine treatment is often ongoing and many adolescents struggle with optimal metabolic control. Our information has also highlighted the overwhelming need for dedicated psychological expertise. The largest number of initial indications for referral were for gynaecology and/or urology assessment (70%) followed by endocrinology (42%). Only seven of the original referrals were for psychological issues and these were always associated with another clinical need. Yet, in terms of on-going clinical input, the greatest workload is likely to be in the form of psychological input, with 46% of patients currently awaiting psychological review and intervention.

<table>
<thead>
<tr>
<th>Table 1</th>
<th>Indications for initial referral (patient may have more than one).</th>
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<tbody>
<tr>
<td>Gynaecology/Urology</td>
<td>Endocrinology</td>
</tr>
<tr>
<td><em>(n = 35)</em></td>
<td><em>(n = 21)</em></td>
</tr>
<tr>
<td>Examination under anaesthetic 3</td>
<td>On-going CAH management <em>(n = 16)</em></td>
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<tr>
<td>Vaginoplasty 4</td>
<td>Oestrogen replacement <em>(n = 5)</em></td>
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<tr>
<td>Incontinence 1</td>
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<td>Gonadectomy 11</td>
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<td>Cliteromegaly 2</td>
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<td>Breast augment 1</td>
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<td>Vaginal dilation 11</td>
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<td>General review 2</td>
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<tr>
<th>Table 2</th>
<th>On-going clinical needs at time of audit and traffic light status (patient may have more than one).</th>
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<tr>
<td>Gynaecology Urology</td>
<td>Endocrinology</td>
</tr>
<tr>
<td><em>(n = 14)</em></td>
<td><em>(n = 19)</em></td>
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<tr>
<td>Clinical need</td>
<td>Examination under anaesthetic 1</td>
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<td>Vaginoplasty 6</td>
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<tr>
<td>Dilation 4</td>
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<td>Clitoral reduction 1</td>
<td></td>
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<tr>
<td>Breast augment 1</td>
<td>Difficult CAH or oestrogen replacement</td>
</tr>
<tr>
<td>Gonadectomy 1</td>
<td></td>
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<tr>
<td>Traffic light status</td>
<td>Amber 14 Red 0</td>
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In acute hospital contexts, psychological needs are often poorly met. One of the reasons is that psychological services tend to be located in the community, whilst expert medical services are usually national centres of excellence which may be geographically distant. Generic community-based psychological services may have limited knowledge — if any — about DSDs. In our experience, the practitioners are often reluctant or lack confidence and expertise to offer DSD patients the highly specialized input developed in multi-disciplinary DSD centres. Even if they are willing, community-based psychological care would be separated from rather than integrated into a multi-disciplinary team, with no obvious communication pathways between providers. If acute medicine is to take on conditions with long-term psychological sequelae like DSDs, then the question of how to situate and resource expert psychological interventions must be formally resolved. A lack of investment may prove to be a false economy in the long run, as an effective transition service may offset many of its own costs by reducing loss of appointments and minimizing crises and complications.

The co-ordinated transitional care programme for adolescents with chronic rheumatological disease [3] can be a useful starting point for DSD. This programme has been demonstrated to significantly improve health-related quality-of-life, knowledge of disease and satisfaction with the clinical care. The central tenet of the programme is development of an individual transition plan for each child to reflect the development stage reached. Age-appropriate information resources and development of a departmental transition policy template are also important. Development of an effective transition programme will not happen without organizational commitment and financial investment. A funding model for this work would need to be developed.

The success of the rheumatology programme is in part attributed to the appointment of a coordinator. In DSD, this audit suggests that such a coordinator would require advanced psychological skills to: 1) assess and formulate the on-going developmental psychological needs of adolescents with DSDs and sometimes their families, especially where psychological concerns arise prior to transitioning; 2) co-ordinate links with specialist and local services and liaise with these services to support transitioning based on needs; 3) engage the families/carer system and provide support to foster increased patient autonomy in the young person. Administrative back up is essential to monitor transactions and to ensure vulnerable adolescent patients do not become lost to follow up.

Conclusion

Transition services for most chronic medical conditions are haphazard and the field of DSD is no exception. This prospective audit highlights a broad range of clinical concerns relating to young women with DSD. Clinicians need to work towards developing a co-ordinated programme for transitional care for growing children with DSD, with strong emphasis on developing cost-effective specialist psychological interventions, including those that target peer support amongst service users, and on monitoring uptake and impact on patient experience and compliance.

Conflict of interest/funding

None.

References
