Challenges faced by children and adolescents with Differences in Sex Development (DSD) managed at a tertiary care specialized university center in Sri Lanka.

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INTRODUCTION

- Children and adolescents with DSD face different medical and psychosocial challenges at different ages in their lives.[1]
- There is a paucity of data from patients from relatively low resource settings[2], where difficulties faced and their management vary from high resource health care settings.

AIM

To describe the **underlying condition, presentation, challenges and their management** in a cohort of children and adolescents with DSD.

METHOD

Ethics approval (EC– 18-092) was obtained from the Ethics Review Committee, Faculty of Medicine, University of Colombo. Socio-demographic and clinical data of all consenting patients with DSDs were collected to a digital database between 2018- 2020 and analysed using descriptive statistics.

RESULTS

- Among 111 individuals with DSD (mean age 12.5), 26 (23%) were children below 10 years of age and 85 (77%) were adolescents (aged 10- 22 years).
- Predominant condition was 46 XY DSD. (**Figure 1.**)
- CAH (n=41,37%) was the commonest underlying condition followed by Turner's syndrome (n=14,13%).
- Gender of rearing was female in 63 (57 %) and male in 48 (43%)

Table 1. Initial presenting features of patients

Presentation	n (%)
Ambiguous genitalia	64 (58%)
Lump at vulva	3 (3%)
Hypospadias and cryptorchidism	11 (10%)
Features of CAH	10 (9%)
Dysmorphic features	11 (10%)
Precocious puberty	1 (1%)

Figure 1. Distribution according to type of DSD

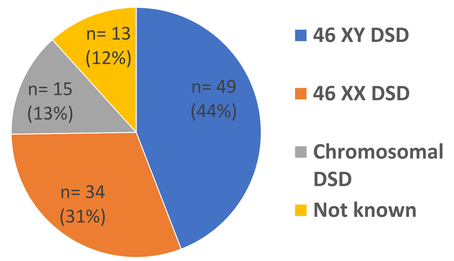

Table 2. Challenges identified in patients with DSD

Birth and childhood	n (%)
Ambiguous genitalia at birth	64 (58%)
Changes to registered gender at birth needed	9 (8%)
Genital surgery Feminizing genitoplasty Hypospadias+/- orchidopexy	58 (52%) 3 (3%) 20 (18%)
Exact genetic diagnosis unavailable	108 (97%)
Living outside the province of center's location	56 (50%)
Adolescence	
Puberty incongruent with gender of rearing	3 (3%)
Gynaecomastia	5 (5%)
Pubertal induction	21 (19%)
Pubertal suppression	10 (9%)

CONCLUSIONS

- Whilst the patients had **relatively good access to hormone replacement therapy, genital surgery and multi-disciplinary care; the lack of genetic facilities, legal support and absence of nearby centers** remain prevalent issues hindering optimum patient outcome.
- Higher percentage of patients in adolescent age group depicts the necessity of structured transitional pathways to adult health care services.

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