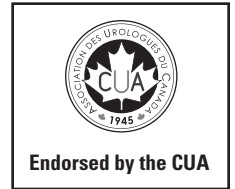


Pediatric Urologists of Canada (PUC) 2021 position statement: Differences of sex development (AKA disorders of sex development)



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Introduction

The term “disorders of sex development” (DSD) was first proposed in a consensus statement in 2006. Since then, the subject of DSD has grown increasingly contentious for a number of reasons, including the related heterogeneous spectrum of congenital conditions encompassed within DSD, and the perceptions around need, timing, and consenting for genital surgery in children. Even the term DSD itself has received criticism; modifications, such as “differences” or “variations” in sexual development, have been proposed.

Today, society appreciates issues of gender and sex differently than those of prior generations. Moreover, lack of high-quality evidence, the constant evolution in overall treatment philosophies, the increasing role of patient advocacy, and the importance of ethical informed consent add nuance to the management of the DSD population worldwide.

Recently, the pendulum has swung in many institutions with respect to surgical interventions. Indeed, in some jurisdictions around the world, legislation has been enacted or is being considered that prohibits genital surgical procedures unless informed consent can be provided by DSD patients themselves rather than their substitute decision-makers (i.e., parents).

Position statement

1. Nomenclature and inclusion under the DSD spectrum

- Shared decision-making when treating patients with DSD begins with the nomenclature. Patients and

families have voiced concerns about the use of terms such as “disorders” and “intersex.”

- There is evidence that patients identify more with their specific diagnosis (e.g., complete androgen insensitivity, mixed gonadal dysgenesis, congenital adrenal hyperplasia [CAH]) rather than with a blanket, general term; moreover, the CAH population has specifically expressed reluctance to inclusion under the DSD label.

Recommendations

- Early in the interaction with patients and families with a diagnosis currently included under the DSD umbrella, providers should actively seek out family preference with regards to how to address them and their diagnoses. We favor “differences in sex development” as the general term, replacing the medicalizing term, “disorders.” We also suggest that consideration be given to referring to DSD patients by their specific diagnosis.
- Research directed at establishing what constitutes DSD with a patient-centered lens should be encouraged and prioritized. This will avoid confusion due to the heterogeneity of conditions grouped under one blanket term. CAH is a cautionary example where some patients later in life may not see themselves as belonging to the large heterogeneous DSD group. Other examples might be patients with isolated hypospadias or isolated bilateral undescended testes.

2. Multidisciplinary care, sex assignment, and disclosure

- Multidisciplinary care** by pediatric endocrinology, pediatric urology, pediatric and adolescent gynecology, genetics, social work, and mental health/psychology/psychiatry is recommended for patients with DSD. Mental health and social work professionals provide

critical expertise and family support in the context of evolving patient psychosocial and psychosexual development.

- b) **Lack of disclosure** about diagnosis, sex assignment or re-assignment decisions, and past interventions performed are specifically the most cited reasons for resentment directed at family members and health-care providers by DSD patients.

Recommendations

- Patients with DSD and related diagnoses should be managed by multidisciplinary teams (MDT) with an interest and experience in the management of such patients. Such management includes regular followup and assessment of objective outcomes over time.
- Sex assignment at birth is neither a medical nor social “emergency,” but does require a MDT approach to achieve as timely as possible elucidation of available data to orient parents and families to the most likely diagnosis underlying their child’s presentation. Education and support of families and the child must be ongoing and grounded in **shared decision-making** and must avoid any fixed or physician-led approach.
- Pediatric urologists should strongly:
 - **Recognize** that DSD patients and families have been harmed in the past, mostly due to incomplete or total lack of information and disclosure.
 - **Advocate** for: i) multidisciplinary care, including systematic and ongoing mental health services; ii) as the child ages, graded and ultimately full patient disclosure of diagnosis; iii) open discussions with caregivers and patients about management options, including observation or medical and/or surgical interventions.

3. Surgical treatment

- a) Surgery in patients with DSD may include diagnostic procedures, surgery to biopsy or remove gonads harboring a malignancy or with malignant potential, surgery to correct inadequate urinary or vaginal drainage, and surgery to prevent kidney damage, as well as gender-affirming surgery or genital reconstruction.

- b) There is a lack of high-quality evidence about the **impact of timing of genital surgery and outcomes of genital surgery in adult DSD patients**. Long-term functional and patient-reported outcomes remain a major and critical gap in knowledge.
- c) In recent years, discussion about timing of genital surgery has been largely centered on informed consent, and lack thereof when performed in young children.

Recommendations

- Discussions about surgical intervention for patients with DSD, especially genital surgery, should take place using a shared decision-making model in the multidisciplinary setting between families, patients, and all team members. Recommendations for major interventions (or not) should be documented as a MDT recommendation and not a standalone surgeon recommendation.
- We support **complete disclosure** to patients and families surrounding the controversies, potential complications, and knowledge gaps around DSD surgery, based on the limited existing published outcomes.
- Data is insufficient to dictate proper timing and extent of any surgical intervention for all DSD patients and must be accrued long-term. We support the creation of a national DSD database given the low volume of cases and ongoing interaction among centers of excellence monitored by the Pediatric Urologists of Canada (PUC). As more information about long-term outcomes of (early and late) surgical interventions becomes available, it should be shared with patients and families during followup.
- Focus on **collaboration** between families, advocacy groups, and the physician, surgeon, and allied health-care professional members of the MDT, to improve shared decision-making and to develop decision-supporting tools for the treatment of DSD.

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