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When to address form and when to address function: Timing of surgical reconstruction for a patient with 46 XY DSD

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ABSTRACT

Differences of sexual development (DSD) refers to congenital conditions characterized by discordant appearances of external genitalia with respect to sex chromosomes. We present a case of a 46 XY DSD adolescent with bilateral undescended testes and severe scrotolabial anomalies who was lost to follow-up for several years who recently presented with "recurrent UTIs." Although the patient desired immediate reconstruction to void while standing, shared-decision making was used to first address his bilateral cryptorchidism, with plans to delay other reconstruction until the patient is older. Pediatric patients with DSD have complicated medical and surgical problems and require a collaborative multidisciplinary team.

1. Introduction

Differences of sex development (DSD; formerly Disorders of sex development) can result from perturbances in sexual differentiation, a highly organized and complex process in a developing fetus.[1] Abnormalities in the sex-determining region Y gene on the Y chromosome can result in the lack of differentiation of the bipotential gonad into testes and subsequent absence of testosterone production. Disrupted hormonal production or activity can result in disorders of phenotype despite a normal karyotype. Over the years, care management for individuals who have a DSD have radically changed based on input from multidisciplinary groups. Despite the presence of evidence-based clinical guidelines, care for DSD patients is not always linear and thus both continuity of care and informed consent are paramount for the overall success of patients. For sensitive or irreversible procedures, it is recommended the intervention be postponed until the patient is old enough to be actively involved in the decision-making process whenever possible. This allows for shared decision-making to promote the patient's welfare and facilitate a more informed decision-making process.[2] In this article, we aim to describe our experience with an adolescent patient with DSD who had significant gaps in his care.

2. Case presentation

A 14-year-old patient who self-identifies as male presented to the pediatric urology clinic with a chief complaint of recurrent UTIs. The patient was seen on day 3 of life by a pediatric endocrinologist for "ambiguous genitalia." Physical exam demonstrated thick labia majora, a phallic structure length measurement of 0.8 cm (normal 4.6 ± 1.8 mm).[3] The urethra was noted on the underside of the phallic structure and no gonads were palpated. Pelvic ultrasound failed to reveal a uterus or ovaries but showed possible gonads in the inguinal canals (Fig. 1). Initial workup revealed 17-hydroxyprogesterone of 90 ng/dL (normal), androstenedione of 1140 ng/dL (high), total testosterone of 8 ng/dL (normal), 17-hydroxypregneolone of 95 ng/dL (normal), normal electrolytes and glucose levels, and karyotype revealed 46 XY. The patient was diagnosed with ambiguous genitalia with undescended gonads and probable defect of testosterone synthesis. Prenatal history was notable for alcohol use during the first 5 months of pregnancy. The patient had 2 siblings and 5 half-siblings without similar diseases in the family. The mother was 39 years old at the time and decided to raise the patient as a girl.

Due to social issues, the patient was lost to follow-up and never had additional interventions. Care was re-established with a recent in-office exam that revealed an anxious young self-identifying male patient with facial hair. Limited genital exam revealed Tanner stage III development with severe chordee. An exam under anesthesia was recommended and revealed severe chordee >90°, penile length 3 cm × 1 cm, a perineal urethral, and bilateral testicular-like structures in the inguinal canals (Fig. 2). Cystoscopy revealed urethral length of 4 cm with normal ejaculatory ducts. Vaginoscopy revealed a blind-ending pouch without a cervix. The patient's goal was to stand when voiding due to anxiety

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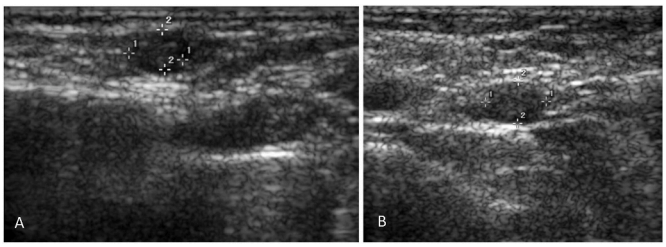

Fig. 1. A, Right sagittal groin ultrasound demonstrating 0.8 × 0.6 × 0.7 cm ovoid mass in high inguinal area. **B**, Left sagittal groin ultrasound demonstrating 1.2 × 0.3 × 0.8 cm ovoid mass in high inguinal area.

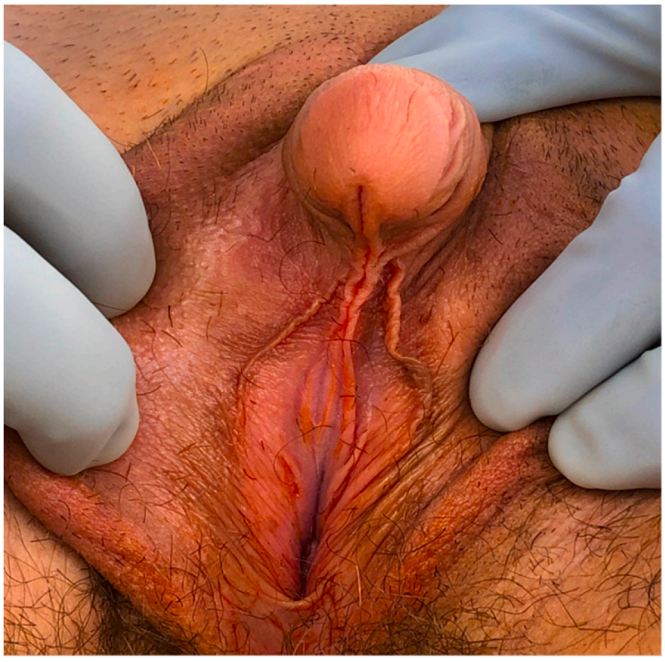

Fig. 2. Intraoperative photo demonstrating severe chordee, proximal meatal location, labial fusion at the penoscrotal junction, poorly developed scrotum with paucity of ventral penile skin.

about peers' perception of him. Weighing the pros and cons of reconstruction and other psychosocial factors, a shared decision was made with orchiopexy planned as the initial step to maximize fertility potential and allow for easier palpation of testicular cancer instead of prophylactic orchiectomy. Timing of phallic and urethral reconstruction was delayed until the child is older after penile growth concludes. The patient was also concurrently and regularly seen by a therapist, psychiatrist, and endocrinologist for psychological and medical care.

3. Discussion

Medical care for patients with DSD is complex and requires a multidisciplinary approach.[4] Counseling for parents of children with DSD is critical as caretakers face challenges regarding the child's complex medical condition, stigma-related concerns, anatomical differences, uncertainty about the child's gender stability, fertility potentials, and future disruptions in day-to-day function. There is no one-size-fits-all approach for patients with DSD, and some interventions may be more time-sensitive than others. An individualized care plan should be developed with the multidisciplinary team and with the patient and their caregivers. In the present case, the mother decided to raise the child in the female gender role. The pediatrician repeatedly made efforts to connect the patient and his caretakers to pediatric urology to remove testicular tissue and prevent characteristics of male sexual development. However, the lack of follow-up and several substantial changes in the patient's social circumstances led to a large gap in his care.

Only after a long delay did the patient resume medical care and focus was brought to the patient's goals of care. Correspondence with pediatric psychiatry revealed that the patient self-identifies as male. Assessment of gender identity and gender-related contentedness are particularly difficult and sensitive topics for individuals with DSD. Conversations regarding gender identity with DSD patients should be initiated early on in their care to align the individualized care plan with the patient's identity and goals.

In the present case, it was critical that the patient's native testes were brought down into an orthotopic location to optimize future fertility and allow for more facile palpation of testicular cancer (which this patient is at higher risk of developing given the advanced age with an undescended testes). Although the patient's desire to void in a standing position is important, such an intervention may necessitate major, irreversible reconstructive surgery with associated risks and thus should be guided by a multidisciplinary approach with shared decision-making. It is important for the clinician to follow detailed guidance in clinical review and data collection for DSD patients and ensure patients or parents of minors are thoroughly informed and give consent for any intervention.

4. Conclusion

Pediatric patients with DSD have complicated medical and surgical problems and require a collaborative multidisciplinary care team. An individualized plan should begin early in childhood and proceed through adolescence with as much involvement with the patient as possible based on their cognitive development. Continuity of care and informed consent are especially important factors in DSD care management for optimizing care.

Ethics approval and consent to participate

IRB for this case report was approved by our institution, Johns Hopkins University.

Verbal and written consent to participate was obtained from legal guardian.

Consent for publication

Written informed consent was obtained from the patient's legal guardian for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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Authors' contributions

1. Oscar Li BS- Oscar is our medical student who is interested in urology. He is the primary author on the initial draft.

2. Andrew Gabrielson MD- Dr. Gabrielson is one of the urology residents, who contributed to the Discussion section of the manuscript.

Ming-Hsien Wang MD, FAAP- Associate Professor of urology and a pediatric urologist. She is the primary physician for this patient. Her contribution includes the initial conceptual design, obtaining consent, and manuscript finalization.

Declaration of competing interest

None.

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