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ADVANCES IN PEDIATRICS

Using Shared Decision-Making Tools to Improve Care for Patients with Disorders of Sex Development

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Keywords

- Disorder of sex development • DSD • Shared decision making
- Complete androgen insensitivity syndrome • CAIS
- Congenital adrenal hyperplasia • CAH
- Mayer Rokitansky Kuster Hauser syndrome

Key points

- Disorders of sex development (DSD) are complex conditions and require a different and comprehensive approach to assure adequate education for the patient and caregivers.
- Patient/family values must be considered when discussing sensitive issues regarding the body and sexual activity, regardless of patient age.
- Controversies in the care of these patients should be shared with the age-appropriate patients and/or caregivers in an up-to-date and unbiased manner.
- Informed consent involves a complete understanding of the expected outcomes. All questions must be answered thoroughly before irreversible surgery, especially regarding gonads or other reproductive organs.

INTRODUCTION

Disorders or differences of sex development (DSD) is a broad term that encompasses a wide range of diagnoses affecting infants, children, and adolescents [1]. Patients with DSD can have genetic abnormalities, anatomic differences, or a combination of the 2 (Table 1). The families of DSD patients are faced with

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Table 1
Types of disorders of sexual differentiation

46,XX DSD	46,XY DSD	Gonadal ambiguities or absence	Anatomic/ developmental anomalies
CAH 21-hydroxylase def. 11-hydroxylase def. 3β-hydroxysteroid def. Aromatase def.	Androgen insensitivity syndrome (AIS) Partial AIS Complete AIS Insufficient testosterone production P450scc defect P450c11 defect P450c17 defect P450c21 defect Inability to convert testosterone to dihydrotestosterone 3β-hydroxysteroid dehydrogenase 17-ketosteroid reductase def. 5α-reductase def.	Mixed gonadal dysgenesis (45,X/46,XY) Pure gonadal dysgenesis (46,XX or 46,XY) Ovotesticular DSD (46,XX or 46,XY)	Cloacal abnormalities Bladder exstrophy Caudal regression syndrome VACTERL syndrome Persistent Mullerian duct syndrome MRKH/vaginal agenesis Distal vaginal agenesis Persistent urogenital sinus

Abbreviation: def, deficiency.

important decisions regarding medical, surgical, and psychological care. There is active research focused on improving the long-term outcomes for these patients and their families. In the setting of that research, the concept of shared decision making has played an increasingly important role [2,3]. The barriers that the patient and family can face include value systems that conflict with those of the providers, a lack of understanding of these complex diagnoses, an ever-changing environment with regards to the controversies and evolution of care, and more basic concerns, such as lack of access to care due to cost or distance.

In this overview, the authors discuss the concept of shared decision-making tools for patients with DSD and their family members. They highlight the controversies in care that led to the development of these checklists, and the intended results regarding optimization of informed consent and expected outcomes.

MANAGEMENT GOALS

History of the care of children with disorders of sex development

A major shift in the care of these complex patients has happened in the last decade as a result of collaboration between medical providers and patient advocates. A Consensus Statement on Management of Intersex Disorders by