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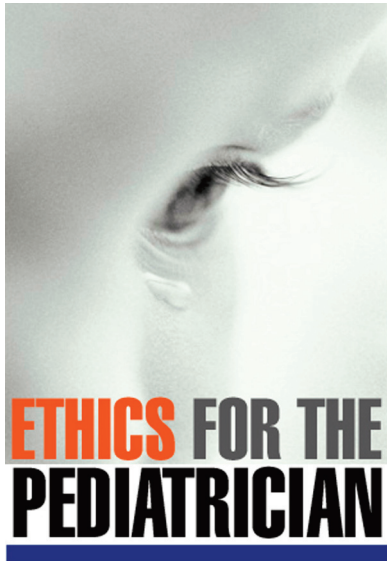
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Disorders of Sex Development: Optimizing Care

Katrina Karkazis, PhD, MPH,* Wilma C. Rossi, MD[†]

Introduction

Disorders of sex development (DSD) are “congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical.” (1) Standard practice from the 1950s to the 1990s had been to identify the cause, assess intervention possibilities, and determine the optimal gender assignment for the child. Surgery usually was performed to make the infant’s genitals more typical for the sex of rearing and to remove gonads discordant with the sex of rearing. Traditional practice also tended toward paternalistic decision-making and incomplete disclosure to parents and affected individuals about the condition and its treatment.

Beginning in the 1990s, adults cared for as children voiced concerns about their care, citing problems with sexual function due to surgical complications, feelings of being on medical display, lifelong struggles with depression and shame, incomplete information about their condition, and insufficient attention by medical caregivers to quality of life issues. Based on patients’ criticisms of care, medical experts and patient advocates convened to revise the model of care for DSD, resulting in the “Consensus Statement on the Management of Intersex Disorders.” (1) The Consensus Statement reflects the current standard of care for DSD and is used as the basis for the following discussion, highlighting

important shifts from previous treatment paradigms.

Optimizing Care

Children born with DSD need a specialized approach to facilitate the highest quality of decision-making required to achieve the best possible care.

Team Approach

Care for children who have DSD requires an experienced multidisciplinary team comprised of a range of pediatric subspecialists (eg, endocrinology, urology/surgery, psychology/psychiatry, social work, nursing, bioethics, and child life). (1)(2) The team also should include the child’s pediatrician (or other primary medical practitioner), who can provide a whole-child/whole-family perspective. Moreover, input from specialists such as reproductive endocrinologists and gynecologists who provide care to adults who have DSD can be critical to understanding long-term sequelae of pediatric treatments. The team should include parents and the child, when appropriate, as partners in discussions regarding the child’s care. Involvement of parents early in the process can help them understand the range of perspectives, help practitioners understand the family’s values and concerns, and create transparency. Open discussion and the valuing of multiple perspectives support the parents’ and team members’ participation in discussions and problem-solving.

Psychosocial Support

Research on parents’ experiences at the birth of their children born with DSD, when gender assignment and

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surgical decisions initially are broached, shows that they typically experience shock, confusion, fear, guilt, anger, sadness, anxiety, shame, and alienation and may grieve the loss of their anticipated “perfect child.” (3) Team members must take great care in relaying information to the parents. Such communication includes reassuring parents that their feelings are normal, emphasizing that the condition is only one aspect of the child, stressing other positive aspects of the newborn, and ensuring that decisions are not made before parents’ emotional responses have been explored. Psychosocial support can come from many sources, including psychologists, social workers, other parents, and support groups.

Shared Decision-making

Based on the principles of beneficence and nonmaleficence, the care of patients must be designed to promote their welfare and cause no harm, respectively. Because many treatments designed to help patients also carry serious risks, the salient ethical issue is whether the benefits of a particular treatment outweigh the burdens for that patient. In DSD, there are few long-term outcome studies of general health and well-being, and results of existing studies are conflicting. Although everyone agrees that the “best interest of the child” is the gold standard for determining care and treatments are considered to be in patients’ best interests when the benefits of treatment outweigh the risks and burdens, the lack of robust outcomes evidence makes it difficult to determine what is best for any one patient. Therefore, it is important to attend to the process of how decisions about care are made.

Numerous organizations have encouraged doctors to adopt a shared decision-making (SDM) approach for complex or difficult health-care

decisions. SDM is especially useful when there is no clear “best-choice” treatment and the ranking of treatment options depends heavily on the decision-maker’s values. Although SDM cannot provide a “right” answer that is empirically defensible, an SDM process can facilitate the increased sharing of information essential for making health-care decisions. The process of thoroughly examining alternatives and encouraging transparency and questioning involved in SDM can help to ensure that the best interests of the child and family are served, patient care and the doctor-patient relationship are improved, satisfaction with the decision-making process is increased for both physician and parents, and decisional conflict and regret are minimized. (4)

Assent and Consent

SDM can help caregivers meet legal and ethical standards for informed consent (or in pediatrics, informed permission and child assent, as appropriate), which generally require physicians to provide all information that a reasonable parent (and child when appropriate) would find important in making an informed decision and to inform parents fully of all options available regardless of the clinician’s biases or preferences. (4) The American Academy of Pediatrics has endorsed the concept of assent for pediatric patients. The child should participate in decisions about his or her treatment at a level appropriate to his or her developmental capacity, and the child’s preferences should be elicited and taken seriously. Overriding these preferences must be explained to the child. (5)

Gender Assignment

Gender assignment entails consideration of the child’s diagnosis, hormonal functioning, internal genitalia, appearance of external genitalia,

potential for fertility, and psychosocial and cultural factors. Caregivers should consult available evidence regarding the likelihood of a female or male gender identity, given the diagnosis. For some diagnoses (eg, complete androgen insensitivity syndrome [CAIS]), gender assignment is straightforward because evidence shows that virtually all women who have this condition identify as female, but for other conditions, gender assignment is more complex and uncertain because data are lacking or inconclusive and gender identity development is a complex interplay of physiologic and sociocultural influences that are not well understood. For more complex cases of gender assignment, caregivers may wish to consult with a specialist in gender identity development for patients born with DSD.

Children who have DSD have significantly higher rates of gender transition than the general population. Thus, despite the best efforts and intentions, the child may not grow up to identify with the gender assigned at birth. As the child matures, the appropriateness of the gender of rearing must be determined. If the child expresses gender dysphoria, it is important to consult with a specialist in gender identity development for patients who have DSD to determine if it is appropriate to consider gender change. Some physicians and parents may feel that gender-atypical behavior is a sign of incorrect gender assignment. Such interpretation of behavior often is not the case, but because such assessments are complex, it is important to consider these issues in conjunction with someone well-versed in child and gender identity development. The child’s ability to contribute to this discussion progresses over childhood. The possibility of gender change later in life is an important consideration when mak-

ing decisions regarding irreversible genital and gonadal surgery in children.

Genital Surgery

The timing, necessity, benefits, and risks of elective genital surgery for children born with DSD are intensely debated. (There is no dispute about the necessity of operations that address imminent threats to the infant's health, such as the creation of a urinary opening.) Although parents and clinicians may wish to attend to genital atypicality as quickly as possible once the child has been assigned a gender (and surgical possibilities may inform gender assignment decisions), gender assignment is a social and legal process that may proceed irrespective of medical or surgical intervention.

Decisions about genitoplasty are made challenging by the dearth of clinical evidence regarding their long-term psychological and physical outcomes. Although parents and clinicians alike have turned to surgical interventions aimed at eliminating atypical sexual characteristics based on the belief that doing so will ease the child's adjustment and relieve parental distress, the Consensus Statement notes that evidence to support this belief is lacking.

In the absence of robust evidence, practitioners are prone to make treatment recommendations based on their personal values, anecdotal evidence, and "gut feeling." (4) Therefore, it is important to explore the physician's bases for recommendations and the parents' values, which can help in weighing the pros and cons of various treatments. These values may include protecting the child from embarrassment or stigma, avoiding unnecessary interventions, keeping surgical and gender options open for the future, ensuring the best physical outcome, ensuring the best

psychological outcome, supporting the child's healthy sexual and gender identity development, enabling the child to have a healthy sex life in adulthood, and preserving fertility. Understanding how the parents weigh these different values should make it easier for the team to outline treatment options so as to address realistically parents' concerns and the child's near- and long-term well-being. (4)

Gonadal Surgery

The preservation of fertility in children born with DSD is important both ethically and legally. Gonadectomy sometimes is contemplated for children who have DSD and who would be fertile, and reproductive technologies increasingly are allowing previously infertile people to become biologic parents. Parents generally cannot consent to elective sterilization of a minor without approval by a court. However, there are no court decisions regarding how these laws apply to cases of DSD. Until the law is settled, legal consultation is advised before any nonemergent removal of the gonads. (6)

In some DSD, including androgen insensitivity, gonadectomy sometimes is recommended due to the risk of malignancy or to prevent the emergence of secondary sex characteristics inconsistent with gender assignment. (7)(8) If the diagnosis is made in infancy or childhood (ie, prepubertally), a decision must be made as to whether gonadectomy should be performed at the time of diagnosis or delayed until after puberty. (7)(8) The Consensus Statement allows for either immediate or delayed orchidectomy in CAIS because the risk of tumor formation is low (<5%) and malignancies never have been seen before age 14 years. In CAIS, deferring surgery to allow the individual to be involved in the

decision is critical. Moreover, deferring gonadectomy until after pubertal development avoids the need for estrogen induction of secondary characteristics. In addition, such patients potentially may be fertile through reproductive technologies such as sperm aspiration. However, for disorders associated with the possibility of prepubertal onset and higher risk of malignancy (eg, partial androgen insensitivity and gonadal dysgenesis), early gonadectomy is advisable.

Disclosure and Education

Just as parents have a right to complete medical information about their child's diagnosis, children have both an ethical and legal right (at age 18 years) to full disclosure about their karyotype, gonadal status, treatment, and prospects for future fertility. It can be devastating for patients to find out information accidentally about the diagnosis or treatment of DSD that never was disclosed to them. Disclosure and education about the diagnosis is a collaborative process that should begin early in childhood and proceed through adolescence. Information should be offered in a developmentally appropriate manner. For example, gender differences can be explained to a young child, genital anatomy and its variations to an older child, and discussion of diagnosis and personal sexual development to the adolescent. Infertility often is a difficult issue for adolescents born with DSD to discover. For infertile patients, other means of having a family should be discussed. Allowing the child to express his or her understanding of the condition at each encounter provides an excellent means of continuing to educate and inform. It is optimal for parents to take the lead in the ongoing process of disclo-

sure with the support of the DSD team.

Conclusion

Children born with DSD have complicated medical problems and require intensive psychosocial support. Medical knowledge in this area is limited and often conflicting. It is critical for the clinician to be open and honest in approaching patients and parents. SDM is recommended to determine the treatment that is in the best interests of the child. Children should be allowed to participate in their care as much as possible, based on their cognitive functioning. The care of patients born with DSD should focus on ultimate patient satisfaction with both medical and psychosocial health and quality of life.

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Suggested Reading

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