

Disorders of Sex Development/Intersex: Gaps in Psychosocial Care for Children

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GAPS IN DISORDERS AND/OR DIFFERENCES OF SEX DEVELOPMENT PSYCHOSOCIAL CARE

Access to psychosocial care is currently inadequate for patients and families affected by a disorder of sex development (DSD; some individuals prefer the terms “intersex” or “differences of sex development,” and some do not identify with any of the labels). In 1 recent study of parents of infants with a DSD, only half of those parents who identified a need for emotional support received what they perceived to be adequate psychological care.¹ This is troubling in that a case can be made that the primary challenges faced by parents of children newly diagnosed with a DSD are informational and psychosocial, not medical or surgical. Even in the case of life-threatening congenital adrenal hyperplasia, the most common cause of a 46,XX DSD, clinical management pathways are well established and driven by clinical practice guidelines not dissimilar from, for example, type 1 diabetes. In a DSD, however, families grapple with a variety of challenges beyond understanding complex medical and/or genetic information, including stigma-related concerns, coping with anatomical differences (whether or not surgery has occurred), surgical complications, lack of clarity regarding gonadal tumor risk, uncertainty about the stability of the child’s gender identity, fertility potential, and the disruptions and burdens associated with repeated medical visits. Detection of a DSD at birth is commonly a time of particularly acute distress; parents report high levels of emotional upheaval and cognitive confusion that often occurs concomitant with medical and surgical decision-making.

There are close to 80 single-gene etiologies for isolated or syndromic DSDs.² Major categories include genes affecting sex determination (eg, gonadal dysgenesis, ovotesticular DSD) and those including sex differentiation (eg, sex hormone synthesis, receptors). Among the most commonly recognized DSDs are classic congenital adrenal hyperplasia, complete or partial androgen insensitivity syndrome, and 5- α reductase deficiency. In the aggregate, estimates of the incidence of DSDs range from 1:100³ to ~1:4500 to 5000 live births.^{4,5} In response to intersex advocacy and invigorated interest and advances in genetic discovery, surgical procedures, and clinical care for people affected by these



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medical conditions, a consensus conference was convened in 2005 by the Lawson Wilkins Pediatric Endocrine Society (currently the Pediatric Endocrine Society) and the European Society for Pediatric Endocrinology. The conference included 50 international experts in the field, along with representatives from the patient advocacy community. The 2006 Consensus Statement on Management of Intersex Disorders (hereafter referred to as the Consensus Statement) was used to introduce a new diagnostic nomenclature under the umbrella term “disorders of sex development”, defined as “congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical.”⁶ The Consensus Statement established principles to guide optimal care. Prominent among these was the principle of comprehensive and integrated multidisciplinary care, including involvement of psychosocial specialists. However, despite these recommendations, the reality is that essential behavioral health services are far from standard, with 1 survey of services offered by DSD teams suggesting that approximately a third of families lack routine access to psychological services.⁷ Barriers include the scarcity of behavioral health providers with specialized training and experience, inadequate prioritizing of the psychosocial component of care in many institutions, and financial models that disincentivize the extra time required to coordinate integrated care.

ADVOCACY EFFORTS TO CHANGE SURGICAL PRACTICES AND THE ROLE FOR PSYCHOSOCIAL CARE

The inadequacies in availability of psychological services was recently highlighted in an October 2017 UK media report in which the author stated the Care Quality Commission of the National Health Service was inquiring whether the Great

Ormond Street Hospital for Children was meeting standards of care for patients with intersex conditions.⁸ The author of a British Broadcasting Corporation report claimed the hospital performed genital surgery without first providing psychological support to families, considered a breach of “national standards and guidelines of care.” In the report, it was noted that this deficiency was compounded by not all cases being discussed at multidisciplinary DSD team meetings and a flawed informed consent process.⁹ The Great Ormond Street Hospital for Children labeled the accusations as unsubstantiated.¹⁰

This story would unlikely have been reported even a relatively few years ago. The author of the piece presumes that psychosocial care is integral to multidisciplinary DSD care, and that without it, genital surgery should not be performed in minors. The timing of the report coincides with increased efforts by some patient advocacy and both national and international human rights organizations calling for a moratorium on gonadal and genital surgeries.^{11,12} Surgical delay would represent a sea change in clinical practice for many infants with a DSD.

Since the early 1990s, intersex advocates have challenged the scientific and ethical basis for performing early gonadal and genital surgery to align the sex anatomy with the child’s gender of rearing. They view surgery as a misguided effort used to address parental anxieties because it does not address what they perceive as the primary factors driving early surgery, that of anxiety, shame, and an accompanying desire to maintain secrecy about the child’s sex anatomy. As an alternative to surgery, advocates call for a robust patient- and family-centered approach to care in which psychological services are essential.

In the current debate, demands for a moratorium on all early elective gonadal or genital surgery,

together with legal actions already taken in the United States,¹³ have overshadowed the accompanying call for expert psychosocial services. Regardless of one’s ethical and legal viewpoints on these surgeries and regardless of the choices that are available to families now or in the future, psychosocial interventions designed to promote open and developmentally appropriate information sharing with the child are vital given documented harm known to follow from secrecy.¹⁴ In addition, the extended family, close family friends, and other caregivers must be educated about DSDs so that they are disabused of misconceptions that exert negative influences on the family and the child’s psychosocial development. For those families who chose early surgery for their infant, management of expectations related to psychosocial sequelae and functional and surgical outcomes is indicated. For those families choosing to defer surgery, problem-solving for successful navigation of psychosocial situations may optimize both child and parent positive acceptance of genital difference. If access to surgical options are curtailed, then access to psychological expertise becomes increasingly crucial. For those families who desire surgery, a legal withdrawal of the surgical option may lead to increased distress, feelings of shame, and steps taken to maintain secrecy about their child’s genital difference. Given the importance of family adjustment for child emotional-behavioral health, psychosocial interventions targeting parent perceptions of self-efficacy in managing anticipated challenges for them as parents or for their child will be critical.

Thorough assessment of parent concerns, family values and preferences, and the introduction of psychosocial approaches as either alternatives or adjuncts to elective surgical approaches are in line with agreed upon principals of

optimal shared decision-making.¹⁵ Psychological interventions used to promote acceptance of differences are a largely unexplored option and may help clarify for whom deferred surgery is most appropriate. Although evidence-based psychosocial interventions have not specifically been tested in families affected by a DSD, cognitive-behavioral and problem-solving psychosocial interventions have demonstrable efficacy in improving the psychosocial functioning of patients and families in other pediatric conditions.

DSD PSYCHOSOCIAL CARE MOVING FORWARD

To accommodate effective psychosocial interventions in DSD, system-level changes are required. Current care models frequently do not offer adequate financial support for complex multidisciplinary care, and traditional fee-for-service payment systems may not support the provision of behavioral health within a subspecialty clinic.¹⁶ Alternate models, such as bundled care, need to be developed and tested. Recognition of the value of psychosocial interventions by DSD teams and health regulatory systems will need to move the conversation away from “what hospitals can afford” to what is an essential level

of care; integrated behavioral health is the recommended standard for numerous subspecialty populations (eg, solid organ transplant, oncology), and national credentialing of centers of excellence may depend on involvement of behavioral health. This is the model recommended in the Consensus Statement, and it has received broad conceptual acceptance. To increase availability of expert psychosocial care, health psychologist training must be used to highlight how expected competencies for the field can be applied to DSD psychosocial concerns. In addition, mentorship and special interest groups within professional societies can be used to accelerate the dissemination of clinical expertise (eg, the newly established DSD Special Interest Group of the Society of Pediatric Psychology, Division 54 of the American Psychological Association).

The work of patient advocates has improved many aspects of clinical management, including the tacit endorsement of openness with patients and parents about all aspects of the child’s condition and its implications. There is a similar acceptance of shared decision-making as an element of patient and family centeredness. Intersex advocacy groups are driving increased accountability of

teams serving patients with a DSD to provide effective psychosocial services that go beyond tokenistic “hand holding” to pacify patients and families in emotional crises. The imperative to provide comprehensive biopsychosocial DSD care is heightened within a changing current of clinical care. Action must be quickly taken to fill gaps in the availability of high-quality psychosocial services within multidisciplinary DSD health care teams. Psychological assessment and intervention have always been important for patients with a DSD and their families; courts and regulatory boards may soon be intervening to insure that standards of care are executed. Perhaps such threats of legal action are necessary to catalyze efforts to actualize the broadly accepted principles promoted in the Consensus Statement.

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ABBREVIATION

DSD: disorder and/or difference of sex development

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