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Introduction to Special Issue

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Introduction to the Special Section: Disorders of Sex Development

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In 2005, an international group of health-care experts, researchers, and patient advocates convened in Chicago to formulate a consensus on the clinical care of persons born with intersex conditions. The *Consensus Statement on Management of Intersex Disorders*, published in 2006, recommended eliminating confusing and potentially stigmatizing terms such as "intersex," "pseudohermaphroditism," "hermaphroditism," and "sex reversal" to refer to these conditions. The conference summary (hereafter referred to as the Consensus Statement)—adopted by the American Academy of Pediatrics as a policy statement—incorporated all variations in somatic sex development under the umbrella term Disorders of Sex Development (DSD), defined as "congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical" (Lee, Houk, Ahmed, Hughes, & in collaboration with the participants in the International Consensus Conference on Intersex organized by the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, 2006). In the majority of the cases, DSD are detected at birth owing to atypical genital appearance; in other cases, the condition only becomes apparent at the time of puberty (e.g., failure to menstruate in girls with a 46,XY karyotype and Complete Androgen Insensitivity syndrome, CAIS). In both early- and late-diagnosed cases, one or more aspects of sex determination or differentiation is/are atypical. "Sex determination" refers to differentiation of the bipotential gonad into either testis or ovary and "sex differentiation" involves formation of the internal reproductive anatomy and external genitalia under the influence of peptide hormones and sex steroids synthesized by a normal functioning gonad (Achermann & Hughes, 2011).

DSD are classified based on the number and visual appearance of the karyotype: *Sex chromosome DSD* encompasses differences in the typical number of sex chromosomes (e.g., 45,X—Turner syndrome and its variants; 47,XXY—Klinefelter syndrome and its variants), or when a person has more than one cell population, that is, mosaicism (e.g., 45,X/46,XY—mixed gonadal dysgenesis); *46,XY DSD* are the consequence of disorders of testis development, disorders of androgen synthesis or action, and other conditions. 46,XY DSD can present as subtle variations in genital development, such as in uncomplicated distal hypospadias (i.e., in which the urinary opening appears just below the glans, rather than at the tip of the penis), to more significant atypicality, such as in CAIS in which a person with XY chromosomes is completely insensitive to the action of androgens, resulting in female-typical external genitalia. Finally, *46,XX DSD* can result from disorders of ovary development, disorders of androgen synthesis, or other conditions, including exposure to exogenous androgens during sensitive periods of sex development. Congenital adrenal hyperplasia (CAH), a treatable but potentially life-threatening condition, is the most common cause of atypical genitalia in 46,XX newborns. Variations in genes responsible for adrenal gland steroid synthesis result in insufficient cortisol and aldosterone (i.e., salt-retaining hormone), but with associated excess androgen production

responsible for masculinization of the genitalia in females.

An inclusive definition of DSD, including Turner and Klinefelter syndromes and hypospadias, has an estimated occurrence of approximately 1 in 100 live births (Arboleda, Sandberg, & Vilain, 2014). Additional details about DSD classification and principles of clinical management can be found in the Consensus Statement (Lee et al., 2006) and in its recent update (Lee et al., 2016). Achermann and Hughes (2011) provide a comprehensive review of the underlying genetics and pathophysiology of DSD.

The birth of a child with DSD, and attendant uncertainty about the child's gender and future psychosocial and psychosexual development, is recognized as being extraordinarily stressful for families (Duguid et al., 2007; Pasterski, Mastroyannopoulou, Wright, Zucker, & Hughes, 2014; Wisniewski & Sandberg, 2015). Potential stressors include the parents' need to gather complex medical information, make decisions about gender assignment, medical or surgical interventions, cope with medical treatments and the possibility of multiple operations, and handle strains on social support because of the attendant perceptions of stigma associated with DSD. These challenges are compounded for families in which DSD are a consequence of a chronic and life-threatening medical condition (e.g., CAH) for which the patient and family have the additional burden of life-long medication management. Stress for patients and families also potentially derives from controversy surrounding clinical management strategies in DSD (Lee et al., 2016, p. 176). For health-care providers, atypical genitalia or discordance between genotype and phenotype (internal reproductive structures and external genitalia) can be the most challenging aspect of an already complex medical condition.

A catalyst for this special section was a research workshop—*Growing up with DSD: Critical Developmental Issues for Children and Families Affected by Disorders of Sex Development*—convened by the National Institute of Child Health and Human Development (NICHD) and affiliated National Institutes of Health offices in Bethesda, MD, in March 2014. Clinical and research experts from a wide range of disciplines were brought together, along with representation from an "umbrella" patient advocacy organization, to generate multidisciplinary discussions serving to inform the NICHD about research priorities in the area of DSD. The topics covered included (a) knowledge of DSD etiologies as well as current models of clinical care for children and adolescents with DSD and their families; (b) knowledge gaps and research needs required to better inform all aspects of care for affected individuals and families; and (c) specific research questions to advance the field. The proceedings were published in a special issue of the journal *Hormone and Metabolic Research* (Freund, Taymans, Wehr, & Stratakis, 2015).

In the 40 years since publication of the first issue of the *Journal of Pediatric Psychology*, there appears to have been no more than six papers published with a focus on DSD: four primary research reports on the topic of psychosocial outcomes associated with hypospadias and its clinical management (Mureau, Slijper, Slob, & Verhulst, 1997; Sandberg et al., 2001; Sandberg, Meyer-Bahlburg, Aranoff, Sconzo, & Hensle, 1989; Sung, Han, Chung, Lee, & Cho, 2014); a systematic review of outcomes in hypospadias (Schönbucher, Weber, & Landolt, 2008); and one case report of a woman with "male pseudohermaphroditism" (i.e., 46,XY DSD) (Quattrin, Aronica, & Mazur, 1990). The primary research all used quantitative research methods. This first special section in the *Journal of Pediatric Psychology* dedicated to the topic of DSD—including five papers, with two using qualitative research methods—comes close to doubling the number of publications.

The five papers comprising this special section are diverse in focus and methodologies. The authors are from Germany, Norway, the United Kingdom, and the United States. The topics range from parent and patient education about CAH (Lundberg, Lindstrom, Roen, & Hegarty, 2016), to recalled experiences in the home and health-care environments of adults with diverse DSD (Meyer-Bahlburg, Khuri, Reyes-Portillo, & New, 2016; Schweizer, Brunner, Gedrose, Handford, & Richter-Appelt, 2016), to a review of studies examining procedures adopted in genital examinations and strategies designed to mitigate potential negative psychological consequences (Tishelman, Shumer, & Nahata, 2016). Finally, the section includes a paper describing the development of parent-proxy and parent self-report measures that focus on issues specific to, and shared by, young patients with DSD and their families, that are not otherwise captured by generic health-related quality of life (HRQoL) measures (Alpern, Gardner, Kogan, Sandberg, & Quittner, 2016).

Factors Potentially Mediating or Moderating the Relationships Between DSD and Quality-of-Life Outcomes

Cheryl Chase (aka Bo Laurent), the founder of the (now defunct) Intersex Society of North America, identified "stigma and emotional trauma" (and not "gender") as the primary challenges of intersexuality (Chase, 2003). It is only in recent years, however, that the role of stigma in DSD, and how it relates to clinical care and well-being, has been taken up as a focus of formal investigation. Anticipated or experienced stigma is recognized as a barrier to health-care delivery and quality of life in general and, in particular for people with DSD and their families (Earnshaw & Quinn,

2012; Rolston, Gardner, Vilain, & Sandberg, 2015; Sanders, Carter, & Goodacre, 2012). A theoretical perspective that assesses the existence of health-related stigma and brings together both individual and social dimensions of this complex phenomenon is lacking. A supportable theory would aid researchers and clinicians in development of a comprehensive measurement tool for stigma, and inform design, monitoring, and evaluation of anti-stigma interventions, in particular in the healthcare setting (Deacon, 2006).

Autobiographical accounts by adults affected by DSD recollecting how, as children and adolescents, they have experienced some medical procedures as stigmatizing, have not always stimulated systematic investigation—perhaps, paradoxically, because the medical treatment of DSD has been motivated by an interest in reducing stigma (Baratz & Karkazis, 2015; Dreger, 1998; Mouriquand et al., 2016; Preves, 2003). In children with CAH, specialists monitor the adequacy of glucocorticoid replacement therapy in suppressing excess endogenous androgen production via laboratory testing, physical examinations, and, in some circumstances, medical photography. When receiving care in academic medical settings, some adults recall having felt obliged to allow learners to view their genitalia because of the rarity of their condition and the perceived responsibility in educating health-care providers about CAH (Engberg, Möller, Hagenfeldt, Nordenskjöld, & Frisén, 2016). Despite providers' intentions to promote feelings of normality and reduce psychological distress during follow-up examinations, the scrutiny associated with genital examinations, teaching demonstrations, or medical photography (Creighton, Alderson, Brown, & Minto, 2002), for some, fortified feelings of being abnormal (Engberg et al., 2016).

The report by Meyer-Bahlburg et al. (2016) is a retrospective qualitative study of adult women (18–51 years) with CAH focusing on the contexts and factors that contributed to and amplified the perceived influence of childhood and adolescent genital examinations on experiences of stigma. A total of 63 women participated in comprehensive (8–10 hr) evaluations (interviews and questionnaires) assessing long-term outcomes. While explicitly avoiding a focus on "stigma" or "differentness," the open-ended format of the interviews was worded to facilitate reports of stigma-related experiences and associated feelings. Most of the women had undergone genital surgery, many more than one. Approximately one-quarter of the participating women reported genital exams in childhood and adolescence as adverse events contributing to an undesired sense of differentness.

The narrative data the authors provide are compelling and useful for sensitizing health-care providers to the importance of adequate preparation and cooperation of the patient before genital exams. Despite the fact that genital exams should be a part of each well-child visit (American Academy of Pediatrics & Committee on Practice Ambulatory Medicine, 2011; Bright Futures/American Academy of Pediatrics, 2016), they are often omitted. Because of this, the experience for patients with DSD can be particularly salient and anxiety-provoking. While repeated genital examinations of the child with atypical genitalia may be necessary for a variety of reasons, this manuscript importantly highlights that this procedure requires compassion, sensitivity, and discretion, and should not be undertaken without careful consideration of the child's understanding and reactions that may contribute to internalized stigma. The question that providers need to ask themselves before performing a genital examination seems to be "will my findings ultimately influence my management?" (Quigley, 2016).

Among the questions unanswered by this study is how the group of women who reported stigma in medical settings differs from the majority who did not report such experiences. Given the wide age range of participants and the only relatively recent admonition in the broadly disseminated Consensus Statement that genital examinations and medical photography can be experienced as shaming (Lee et al., 2006), age cohort effects may be evident. Although not a substitute for better controlled investigations, this article highlights that experienced, anticipated, and internalized stigma in medical settings may be important factors mediating the relationship between atypical genitalia and negative psychosocial outcomes in populations with DSD.

In an effort to identify additional factors that potentially modulate outcomes in DSD, Schweizer and colleagues (2016) considered the quality of parental care and social support. In recent decades, findings from studies of psychological adjustment in individuals with DSD have influenced the development of psychosocial services within the interdisciplinary team approach to clinical management (Pasterski, Prentice, & Hughes, 2010). At the heart of this component of improved patient care lies recognition of the complexity of DSD as well as the life-long implications secondary to early treatment-related decisions. While decisions about an early irreversible surgery have been challenged (Feder, 2014), the need for psychological and emotional support remains, regardless (Liao, Wood, & Creighton, 2015). Further, the need for support of parents in their own decision-making has come to the fore (Siminoff & Sandberg, 2015). In their report, Schweizer et al. (2016) recognize the role parents play as "first agents" in nurturing a child with DSD. They have also considered the role other sources of support may play in psychosocial outcomes. To this end, the authors used an exploratory approach to assess interrelationships between medical, familial, and

social childhood experiences, as they relate to outcomes in adulthood.

In a retrospective, cross-sectional questionnaire survey of 69 men and women (aged 16–60 years) with diverse DSD recruited from "medical doctors (e.g., gynecologists, endocrinologists), the main German intersex support groups, a website informing about the study, and publicity material distributed at scientific conferences" (recruitment details described in Schönbucher et al., 2012), Schweizer and colleagues found evidence of treatment-related experiences that predicted worse psychological outcomes. For example, repeated genital surgeries were associated with reduced satisfaction with physical appearance in later life. However, the authors also contributed to our understanding of "protective" factors, which warrant attention in adopting a holistic care approach. That is, they found that reports of positive parenting experiences were related to reduced psychological distress and improved self-perceptions of attractiveness. Furthermore, they showed that having a childhood "confidant" also improved self-perceptions of attractiveness, reduced body-related insecurities, and reduced prevalence of suicidal thoughts. In sum, while Schweizer et al. (2016) corroborated the long-held notion that medical interventions must be considered with the utmost caution and care, they were able to point to contextual or background factors that potentially attenuate difficulties associated with DSD.

Education Beyond the Facts

Educating parents about details of their child's medical condition and strategies to facilitate adaptive coping and self-management is a core principle of patient- and family-centered pediatric care. Yet, there potentially remain gaps in the matching of the information provided and parents' (and, when older, the patient's) understanding of how to apply the knowledge learned. In the study by Lundberg and colleagues (2016), 20 parents of 22 children (16 daughters and 6 sons, 1–20 years) with CAH from the United Kingdom and Sweden were interviewed about experiences related to CAH. Interview transcripts were summarized generating themes along a timeline corresponding with their prominence in the child's development. Following the definitions of Pols (2013), this article focuses on the "different kinds of knowing" about a medical condition in the context of three themes that emerged from the interviews: *Making Sense of the Situation*, *Attending to Medical Needs*, and *Building Independence*. The first two forms of "knowing"—knowing "what" and "how"—will perhaps be the most familiar to health-care providers because they deal with the facts about the medical condition and the actions required to ensure the child's physical health, that is, what is CAH and what medications are required and when they are to be administered. But even here, there is the nuance of parents knowing how to openly talk about CAH with their child; parents can struggle mightily with discussing the topic of atypical genital appearance with daughters, irrespective of whether genital surgery has been performed. In the case of girls with a urogenital sinus (i.e., common channel for the urethra and vagina instead of them being normally separate), vaginal dilation (with or without surgery) will frequently be needed during adolescence or young adulthood among those wishing to experience sexual intercourse (Auchus & Quint, 2015). Similarly, preparing the youth to share details of their condition with select others is critical, as failing to do so with even the closest of friends may augment feelings of shame. Although the focus of this study is CAH, the lessons learned translate well to other DSD. An outstanding paper by a parent of a daughter with Partial Androgen Insensitivity syndrome captures precisely the varied forms of knowledge that parents of children with DSD need, even if they do not directly request it (Magritte, 2012). The behavioral health member of the DSD interdisciplinary health-care team is optimally positioned—by virtue of training and availability for extended clinical interactions—to champion the topic of such deep "knowing" by both parents and patients.

Mitigating Potential Harm

As clearly illustrated in the paper by Meyer-Bahlburg et al. (2016), and as noted in several other sources (Creighton, Alderson, Brown, & Minto, 2002; Crissman et al., 2011; Lossie & Green, 2015; Money & Lamacz, 1987), the genital examination can be experienced as an adverse and stigmatizing event with long-term negative consequences for the relationship between the patient and their health-care providers. The Consensus Statement notes that "repeated examination of the genitalia, including medical photography, may be experienced as deeply shaming" (p. e493), but provides limited guidance regarding how negative consequences of these procedures can be mitigated or avoided. Systematic research on the procedures used and emotional reactions to genital examinations or medical photography that can be used to inform best practices in DSD care is lacking. In this issue, Tishelman and colleagues (2016) provide suggestions for provider behavior, based on a selective review of studies of youth undergoing genital examination related to investigations of child sex abuse (CSA). This literature illustrates the importance of adequate preparation of the child and the caregiver for the procedures. The value of providing the caregiver with guidance on how to communicate with their child about the purpose for, and description of, the

impending procedure is unquestionably also relevant to DSD. Increased child knowledge about the exam is associated with reduced distress during and following CSA exams, as are procedures such as distraction and, when possible, offering the child control over aspects of the exam. Characteristics of the provider (e.g., "perceived kindness"), use of participant modeling films and psychoeducational videotapes providing information regarding the procedure and coping strategies have all been shown to reduce anxiety. In general, the literature reviewed established that distress related to genital exams is not inevitable and, when observed, is related to potentially modifiable factors. Attesting to the transferability of the strategies shown to be helpful in reducing distress stemming from CSA genital exams, the authors point out that many of these same procedures are recommended by the American Heart Association to prepare children and adolescents for invasive cardiac procedures (LeRoy et al., 2003). Tishelman et al. (2016) conclude with several practical recommendations, which can immediately be applied to the DSD clinic setting.

Measurement of HRQoL in DSD

A consensus has emerged that disease/category-specific HRQoL measures are more sensitive to smaller differences and smaller changes over time than generic measures and are viewed as more relevant by respondents (Matza, Swensen, Flood, Secnik, & Leidy, 2004; Quittner, Cruz, Modi, & Marciel, 2009; U. S. Food and Drug Administration, 2009). In the final paper of this special section, Alpern and colleagues (2016) describe the development of parent-proxy and parent self-report HRQoL measures targeting children $\leq$6 years with DSD. Although DSD is an umbrella term for multiple discrete conditions and syndromes, they share physical, clinical management, and social and emotional sequelae. Following as closely as possible the guidance of the Food and Drug Administration's regulatory guidance on the development of patient-reported outcomes (PROs) (U. S. Food and Drug Administration, 2009), as well as the *Journal of Pediatric Psychology* "author checklist" for measure development and validation manuscripts (Holmbeck & Devine, 2009), the Quality of Life DSD Proxy (QOL-DSD-Proxy) and Parent Report (QOL-DSD-Parent) meet stringent standards for methods construction. Because young children are unable to report for themselves, proxy reports have become essential in the assessment of function and adaptation in the early years (Quittner, Cejas, & Blackwell, 2013). Accordingly, the QOL-DSD-Proxy (2–6 years) uses parents (or other primary caregivers) as informants. The QOL-DSD-Parent assesses parents' HRQoL in relation to their child with DSD (birth–6 years). The importance of tapping the effects of the child's DSD and his/her clinical management on the parents stems from the prominent role that caregivers take in early clinical decisions, some of which are elective, but irreversible (i.e., next-generation genetic sequencing with its associated difficulties in interpeting results or in reporting nonrelated "incidental" findings, genital surgery, or removal of the gonads). The QOL-DSD-Parent provides a means for assessing parent/family status in multiple domains, many of which may be pertinent to early decision-making and in the tracking of reactions to decisions over time.

These measures promise to enhance health care by identifying children's and parents' immediate informational, emotional, and social needs; alerting clinicians to problems that arise; and evaluating benefits of new medical or psychosocial interventions.

Recommendations for Future Research

Sampling Strategies

Attention to the source of participants in DSD studies needs to be aggressively addressed in future studies. Not infrequently, participants are recruited through patient support organizations (for example, as in the Schweizer et al. paper in this issue) without consideration that data from these self-selected samples may substantially vary in their experiences from those systematically recruited through medical centers (e.g., Zucker, 2002). Patient participation rates in peer support networks have been reported to be $<$10% and little is known about determinants of interest and engagement (Baratz, Sharp, & Sandberg, 2014).

Optimal Recall Period for HRQoL Measures in DSD

What constitutes a suitable recall period for PROs is debatable and hinges on several factors, including the purpose and intended use of the measure, the characteristics of the condition under study, the intervention being evaluated, and informant ability to accurately recall the information requested (Stull, Leidy, Parasuraman, & Chassany, 2009). Clinical experience suggests that the challenges associated with DSD can be episodic rather than uniform; a short recall period may underestimate symptom burden. On the other hand, longer recall intervals can result in increased errors, making it more difficult to demonstrate differences between treatment groups in a clinical trial. Depending on the purpose of the assessment, examining different aspects of experiences associated with DSD may require different recall periods.

Decision-Making for Surgery

In considering the many challenges facing the parents/caregivers of a child diagnosed with DSD,

decision-making with respect to surgical intervention is perhaps the most difficult. The decision itself is paradoxical in that it often pertains to the future status of an individual that cannot be known until later in life (e.g., gender identity, sexual orientation, psychosexual functioning, and the person's sense of authenticity [Pachankis, 2007; Quinn & Chaudoir, 2009]). Historically, decisions about surgical intervention have centered on "sex-assignment" of the child, with obvious far-reaching implications when assignment implied irreversible surgical intervention. More recently, however, the carefully considered distinction of "social" gender assignment at birth has allowed for a deferment of nonessential surgical intervention(s) until the child has reached the age of autonomy. Nevertheless, a categorization of all surgeries as elective may cause distress for parents where particular interventions are genuinely medically necessary. In addition, guidance and criticism on the issue has come from a diversity of nonmedical stakeholders, adding to the already complex process. For example, blanket opinions on urogenital surgical intervention have been issued by medical ethicists (Earp, 2015), social anthropologists (Fortier, 2016), and political bodies such as the United Nations (Méndez, 2013), with domain-bound perspectives that may not appreciate condition-specific complexities. While these perspectives may have broad implications, they should build on specific evidence-based guidance. Given the heterogeneous presentation of DSD and rarity of individual conditions, there is, to date, no definitive consensus regarding indication, timing, procedure, or evaluation of outcome that spans the category DSD (Mouriquand, Caldamone, Malone, Frank, & Hoebeke, 2014; Mouriquand et al., 2016).

Despite a growing body of evidence on technological advancements in surgical procedures and functional and cosmetic outcomes, and suggestions that the frequency of genital surgery may be declining (Michala, Liao, Wood, Conway, & Creighton, 2014), many studies assessing functional outcomes of early genital surgery are based on outdated surgical techniques in older participants and/or small samples of unoperated participants. With respect to the latter point, much of the argument against surgical intervention is predicated on "counterfactual thinking" (Kahneman & Tversky, 1982). That is, considering negative outcomes only, as they relate to surgery, without allowing for potential distress and/or dysfunction that may result from withholding DSD-related surgical interventions. For example, a recent study of 91 women with DSD (64% having received surgery) found that two-thirds of these women were at risk of developing a sexual dysfunction regardless of whether they had surgery (Callens et al., 2012). They concluded that surgery did not improve or harm this aspect of functioning; however, future studies considering these and other aspects of overall well-being in patients (and their parents) may shed additional light on the motivations for, and impact of, such decisions. Finally, and as mentioned earlier, further potential confounds in existing reports of outcome include selection bias of study participants (e.g., self-selection) and potential researcher bias, depending on beliefs about urogenital surgery, which may shape the approach to answering research questions. There remains a critical need for prospective, condition-specific studies using larger and more representative samples adopting rigorous protocols of evaluation. To prevent the possibility of particular stakeholder groups dismissing out of hand results of future studies, the design of such investigations would ideally involve the input of all stakeholder groups invested in enhancing health and quality-of-life outcomes in people with DSD (Vayena et al., 2016).

Final Thoughts

The Director of the National Institute on Minority Health and Health Disparities recently announced that sexual and gender minorities (SGMs) are now designated as health disparity populations for NIH research. The term SGM encompasses "lesbian, gay, bisexual, and transgender populations, as well as those whose sexual orientation, gender identity and expressions, or reproductive development (read: DSD) varies from traditional, societal, cultural, or physiological norms" (National Institute on Minority Health and Health Disparities (NIMHD) & Director's Office, October 6, 2016). It is hoped that with its new designation as a "disparity population for research," DSD will attract the attention of the pediatric psychology community to fill gaps in our knowledge of factors influencing developmental trajectories and outcomes. To date, the topic of DSD has been examined by a relatively small community of social scientists. Perhaps this stems from perceptions that DSD are extraordinarily rare or that their psychosocial consequences are so different from other congenital conditions such that pediatric psychologists have come to believe that studying DSD would be impractical or require extensive additional training. While it is true that discrete DSD syndromes are rare, they are prevalent in the aggregate (Arboleda et al., 2014). There is also a strong argument to be made for adopting a "noncategorical" approach to the study of DSD that takes into account the unique aspects of the child's medical condition, while also considering the total life experience of the child, family, and broader social context (Sandberg & Mazur, 2014). One example of the commonality that DSD share with other conditions concerns the risks versus benefits of educating the child or adolescent with DSD about particular details of their condition

(e.g., sex chromosomes discordant with gender of rearing and identity; Sisk, Bluebond-Langner, Wiener, Mack, & Wolfe, 2016). The Consensus Statement noted the proven value for positive psychological adjustment of openness as learned from different conditions and circumstances, including pediatric HIV (American Academy of Pediatrics, 1999). The argument has been made that lessons relevant to DSD clinical research and health-care delivery can potentially be learned from studies of peer relations in physically typical children (Bukowski, McCauley, & Mazur, 2015) and from children with other congenital conditions (Holmbeck & Aspinall, 2015). Progress in DSD research will benefit greatly when investigators from other areas of child development (typical and atypical) and other social scientists direct their methodologies and research strategies toward the issues confronted by these patients and their families.

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References

Achermann, J., & Hughes, I. (2011). Disorders of sex development. In S Melmed, K Polonsky, P Larsen, & H Kronenberg (Eds.), *Williams textbook of endocrinology* (12th ed., pp. 868–934). Philadelphia, PA: W B Saunders Co

Alpern, A. N., Gardner, M., Kogan, B., Sandberg, D. E., & Quittner, A. L. (2016). Development of health-related quality of life instruments for young children with disorders of sex development DSD) and their parents. *Journal of Pediatric Psychology*, *42*, 544–558. doi:10.1093/jpepsy/jsw022

American Academy of Pediatrics. (1999). Committee on pediatric AIDS. Disclosure of illness status to children and adolescents with HIV infection. *Pediatrics*, *103*, 164–166.

American Academy of Pediatrics & Committee on Practice Ambulatory Medicine. (2011). Use of chaperones during the physical examination of the pediatric patient. *Pediatrics*, *127*, 991–993. doi:10.1542/peds.2011-0322

Arboleda, V. A., Sandberg, D. E., & Vilain, E. (2014). DSDs: Genetics, underlying pathologies and psychosexual differentiation. *Nature Reviews. Endocrinology*, *10*, 603–615. doi:10.1038/nrendo.2014.130

Auchus, R. J., & Quint, E. H. (2015). Adolescents with disorders of sex development (DSD)–lost in transition?. *Hormone and Metabolic Research*, *47*, 367–374. doi:10.1055/s-0035-1545303

Baratz, A., & Karkazis, K. (2015). *Cris de coeur* and the moral imperative to listen to and learn from intersex people. *Narrative Inquiry in Bioethics*, *5*, 127–132. doi:https://doi.org/10.1353/nib.2015.0030

Baratz, A. B., Sharp, M. K., & Sandberg, D. E. (2014). Disorders of sex development peer support. In O. Hiort & S. Ahmed (Eds.), *Understanding differences and disorders of sex development (DSD). Endocrine development* (pp. 99–112). Basel: Karger

Bright Futures/American Academy of Pediatrics. (2016). Recommendations for preventive pediatric health care. Retrieved from https://www.aap.org/en-us/Documents/periodicity_schedule.pdf

Bukowski, W. M., McCauley, E., & Mazur, T. (2015). Disorders of sex development (DSD): Peer relations and psychosocial well-being. *Hormone and Metabolic Research*, *47*, 357–360. doi:10.1055/s-0035-1549884

Callens, N., van der Zwan, Y., Drop, S., Cools, M., Beerendonk, C., Wolffenbuttel, K., & Dessens, A. (2012). Do surgical interventions influence psychosexual and cosmetic outcomes in women with disorders of sex development? *ISRN Endocrinology*, *2012*, 276742. doi: doi:10.5402/2012/276742

Chase, C. (2003). What is the agenda of the intersex patient advocacy movement?. *Endocrinologist*, *13*, 240–242. doi:10.1097/01.ten.0000081687.21823.d4

Creighton, S., Alderson, J., Brown, S., & Minto, C. L. (2002). Medical photography: Ethics, consent and the intersex patient. *BJU International*, *89*, 67–71. doi:10.1046/j.1464-410X.2002.02558.x

Crissman, H. P., Warner, L., Gardner, M., Carr, M., Schast, A., Quittner, A. L. … Sandberg, D. E. (2011). Children with disorders of sex development: A qualitative study of early parental experience. *International Journal of Pediatric Endocrinology*, *2011*, 10. doi:10.1186/1687-9856-2011-10

Deacon, H. (2006). Towards a sustainable theory of health-related stigma: Lessons from the HIV/AIDS literature. *Journal of Community & Applied Social Psychology*, *16*, 418–425. doi:10.1002/casp.900

Dreger, A. (1998). 'Ambiguous sex'—Or ambivalent medicine? Ethical issues in the treatment of intersexuality. *Hastings Center Report*, *28*, 24–35. doi:10.2307/3528648

Duguid, A., Morrison, S., Robertson, A., Chalmers, J., Youngson, G., & Ahmed, S. F.; the Scottish Genital Anomaly Network. (2007). The psychological impact of genital anomalies on the parents of affected children. *Acta Pædiatrica*, *96*, 348–352. doi:10.1111/j.1651-2227.2006.00112.x

Earnshaw, V. A., & Quinn, D. M. (2012). The impact of stigma in healthcare on people living with chronic illnesses. *Journal of Health Psychology*, *17*, 157–168. doi:10.1177/1359105311414952

Earp, B. D. (2015). Female genital mutilation and male circumcision: Toward an autonomy-based ethical framework. *Medicolegal and Bioethics*, *5*, 89–104.

Engberg, H., Möller, A., Hagenfeldt, K., Nordenskjöld, A., & Frisén, L. (2016). The experience of women living with congenital adrenal hyperplasia: Impact of the condition and the care given. *Clinical Endocrinology*, *85*, 21–28. doi:10.1111/cen.13054

Feder, E. K. (2014). *Making sense of intersex: Changing ethical perspectives in biomedicine*. Bloomington, IN: Indiana University Press.

Fortier, C. (2016). Inscribing trans and intersex people in the dominant binary categories of gender. *eTropic: Electronic Journal of Studies in the Tropics*, *13*, 36–43.

Freund, L. S., Taymans, S., Wehr, E., & Stratakis, C. A. (2015). Developmental focus on disorders of sex development: Special issue overview. *Hormone and Metabolic Research*, *47*, 309–311. doi:10.1055/s-0035-1549880

Holmbeck, G. N., & Aspinall, C. L. (2015). Disorders of sex development: Lessons to be learned from studies of spina bifida and craniofacial conditions. *Hormone and Metabolic Research*, *47*, 380–386. doi:10.1055/s-0035-1545273

Holmbeck, G. N., & Devine, K. A. (2009). Editorial: An author's checklist for measure development and validation manuscripts. *Journal of Pediatric Psychology*, *34*, 691–696. doi:10.1093/jpepsy/jsp046

Kahneman, D., & Tversky, A. (1982). The simulation heuristic. In D. Kahneman, P. Slovic, & A. Tversky (Eds.), *Judegment under uncertainty: Heuristics and biases* (pp. 201–208). New York, NY: Cambridge University Press.

Lee, P. A., Houk, C. P., Ahmed, S. F., & Hughes, I. A.; in collaboration with the participants in the International Consensus Conference on Intersex organized by the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology. (2006). Consensus statement on management of intersex disorders. *Pediatrics*, *118*, e488–e500. doi:10.1542/peds.2006-0738

Lee, P. A., Nordenstrom, A., Houk, C. P., Ahmed, S. F., Auchus, R., Baratz, A. ... Vilain, E.; Witchel SGlobal DSD Update Consortium. (2016). Global disorders of sex development update since 2006: Perceptions, approach and care. *Hormone Research in Paediatrics*, *85*, 158–180. doi:10.1159/000442975

LeRoy, S., Elixson, E. M., O'brien, P., Tong, E., Turpin, S., & Uzark, K. (2003). Recommendations for preparing children and adolescents for invasive cardiac procedures. A statement from the American Heart Association Pediatric Nursing Subcommittee of the Council on Cardiovascular Nursing in collaboration with the Council on Cardiovascular Diseases of the Young. *Circulation*, *108*, 2550–2564. doi:10.1161/01.cir.0000100561.76609.64

Liao, L. M., Wood, D., & Creighton, S. M. (2015). Parental choice on normalising cosmetic genital surgery. *British Medical Journal*, *351*, h5124. doi:10.1136/bmj.h5124

Lossie, A. C., & Green, J. (2015). Building trust: The history and ongoing relationships amongst DSD clinicians, researchers, and patient advocacy groups. *Hormone and Metabolic Research*, *47*, 344–350. doi:10.1055/s-0035-1548793

Lundberg, T., Lindstrom, A., Roen, K., & Hegarty, P. (2016). From knowing nothing to knowing what, how and now: Parents' experiences of caring for their children with congenital adrenal hyperplasia. *Journal of Pediatric Psychology*, *42*, 520–529. doi:10.1093/jpepsy/jsw001

Magritte, E. (2012). Working together in placing the long term interests of the child at the heart of the DSD evaluation. *Journal of Pediatric Urology*, *8*, 571–575. doi:10.1016/j.jpurol.2012.07.011

Matza, L. S., Swensen, A. R., Flood, E. M., Secnik, K., & Leidy, N. K. (2004). Assessment of health-related quality of life in children: A review of conceptual, methodological, and regulatory issues. *Value Health*, *7*, 79–92. doi:10.1111/j.1524-4733.2004.71273.x

Méndez, J. E. (2013). Report of the special rapporteur on torture and other cruel, inhuman or degrading treatment or punishment. Human Rights Council, Twenty-eighth session. Agenda item, 3. Retrieved from http://www.ohchr.org/Documents/HRBodies/HRCouncil/RegularSession/Session22/A.HRC.22.53_English.pdf

Meyer-Bahlburg, H. F. L., Khuri, J., Reyes-Portillo, J., & New, M. I. (2016). Stigma in medical settings as reported retrospectively by women with congenital adrenal hyperplasia (CAH) for their childhood and adolescence. *Journal of Pediatric Psychology*, *42*, 496–503. doi:10.1093/jpepsy/jsw034

Michala, L., Liao, L. M., Wood, D., Conway, G. S., & Creighton, S. M. (2014). Practice changes in childhood surgery for ambiguous genitalia? *Journal of Pediatric Urology*, *10*, 934–939. doi:10.1016/j.jpurol.2014.01.030

Money, J., & Lamacz, M. (1987). Genital examination and exposure experienced as nosocomial sexual abuse in childhood. *Journal of Nervous and Mental Disease*, *175*, 713–721.

Mouriquand, P., Caldamone, A., Malone, P., Frank, J. D., & Hoebeke, P. (2014). The ESPU/SPU standpoint on the surgical management of disorders of sex development (DSD). *Journal of Pediatric Urology*, *10*, 8–10. doi:10.1016/j.jpurol.2013.10.023

Mouriquand, P. D., Gorduza, D. B., Gay, C. L., Meyer-Bahlburg, H. F., Baker, L., Baskin, L. S. ... Lee, P. (2016). Surgery in disorders of sex development (DSD) with a gender issue: If (why), when, and how? *Journal of Pediatric Urology*, *12*, 139–149. doi:10.1016/j.jpurol.2016.04.001

Mureau, M. A., Slijper, F. M., Slob, A. K., & Verhulst, F. C. (1997). Psychosocial functioning of children, adolescents, and adults following hypospadias surgery: A comparative study. *Journal of Pediatric Psychology*, *22*, 371–387.

National Institute on Minority Health and Health Disparities (NIMHD) & Director's Office. (October 6, 2016). Director's message. Sexual and gender minorities formally designated as a health disparity population for research purposes. Retrieved from https://www.nimhd.nih.gov/about/directors-corner/message.html

Pachankis, J. E. (2007). The psychological implications of concealing a stigma: A cognitive-affective-behavioral model. *Psychological Bulletin*, *133*, 328–345.

Pasterski, V., Mastroyannopoulou, K., Wright, D., Zucker, J., & Hughes, I. (2014). Predictors of posttraumatic stress in parents of children diagnosed with a disorder of sex development. *Archives of Sexual Behavior*, *43*, 369–375. doi:10.1007/s10508-013-0196-8

Pasterski, V., Prentice, P., & Hughes, I. (2010). Consequences of the Chicago consensus on disorders of sex development (DSD): Current practices in europe. *Archives of Disease in Childhood*, *95*, 618–623. doi:10.1136/adc.2009.163840

Pols, J. (2013). Knowing patients: Turning patient knowledge into science. *Science, Technology and Human Values*, *39*, 73–97. doi:10.1177/0162243913504306

Preves, S. E. (2003). *Intersex and identity: The contested self*. New Brunswick, NJ: Rutgers University.

Quattrin, T., Aronica, S., & Mazur, T. (1990). Management of male pseudohermaphroditism: A case report spanning twenty-one years. *Journal of Pediatric Psychology*, *15*, 699–709.

Quigley, C. (2016). Genital examination: When and how? (what I learned from Jodie) Retrieved from http://dsdfamilies.org/mednote/genital_examination.php

Quinn, D. M., & Chaudoir, S. R. (2009). Living with a concealable stigmatized identity: The impact of anticipated stigma, centrality, salience, and cultural stigma on psychological distress and health. *Journal of Personality and Social Psychology*, *97*, 634–651. doi:10.1037/a0015815

Quittner, A. L., Cejas, I., & Blackwell, L. S. (2013). Advances in the measurement and utilization of health-related quality of life instruments. In W. T. O'Donohue, L. T. Benuto, & L. Woodward Tolle (Eds.), *Handbook of adolescent health psychology* (pp. 153–163). New York, NY: Springer.

Quittner, A. L., Cruz, I., Modi, A. C., & Marciel, K. K. (2009). Health-related quality of life instruments for adolescents with chronic diseases In W. T. O'Donohue & L. W. Tolle (Eds.), *Behavioral approaches to chronic disease in adolescence* (pp. 311–327). New York, NY: Springer

Rolston, A. M., Gardner, M., Vilain, E., & Sandberg, D. E. (2015). Parental reports of stigma associated with child's disorder of sex development. *International Journal of Endocrinology*, *2015*, 15. doi:10.1155/2015/980121

Sandberg, D. E., & Mazur, T. (2014). A noncategorical approach to the psychosocial care of persons with DSD and their families. In B. P. C. Kreukels, T. D. Steensma, & A. L. C. de Vries (Eds.), *Gender dysphoria and disorders of sex development* (pp. 93–114). US: Springer.

Sandberg, D. E., Meyer-Bahlburg, H. F., Hensle, T. W., Levitt, S. B., Kogan, S. J., & Reda, E. F. (2001). Psychosocial adaptation of middle childhood boys with hypospadias after genital surgery. *Journal of Pediatric Psychology*, *26*, 465–475. doi:10.1093/jpepsy/26.8.465

Sandberg, D. E., Meyer-Bahlburg, H. F. L., Aranoff, G. S., Sconzo, J. M., & Hensle, T. W. (1989). Boys with hypospadias: A survey of behavioral difficulties. *Journal of Pediatric Psychology*, *14*, 491–514. doi:10.1093/jpepsy/14.4.491

Sanders, C., Carter, B., & Goodacre, L. (2012). Parents need to protect: Influences, risks and tensions for parents of prepubertal children born with ambiguous genitalia. *Journal of Clinical Nursing*, *21*, 3315–3323. doi:10.1111/j.1365-2702.2012.04109.x

Schönbucher, V. B., Weber, D. M., & Landolt, M. A. (2008). Psychosocial adjustment, health-related quality of life, and psychosexual development of boys with hypospadias: A systematic review. *Journal of Pediatric Psychology*, *33*, 520–535. doi:10.1093/jpepsy/jsm098

Schönbucher, V., Schweizer, K., Rustige, L., Schützmann, K., Brunner, F., & Richter-Appelt, H. (2012). Sexual quality of life of individuals with 46,XY disorders of sex development. *The Journal of Sexual Medicine*, *9*, 3154–3170. doi:10.1111/j.1743-6109.2009.01639.x

Schweizer, K., Brunner, F., Gedrose, B., Handford, C., & Richter-Appelt, H. (2016). Coping with diverse sex development: Treatment experiences and psychosocial support during childhood and adolescence and adult well-being. *Journal of Pediatric Psychology*, *42*, 504–519. doi:10.1093/jpepsy/jsw058

Siminoff, L. A., & Sandberg, D. E. (2015). Promoting shared decision making in disorders of sex development (DSD): Decision aids and support tools. *Hormone and Metabolic Research*, *47*, 335–339. doi:10.1055/s-0035-1545302

Sisk, B. A., Bluebond-Langner, M., Wiener, L., Mack, J., & Wolfe, J. (2016). Prognostic Disclosures to Children: A Historical Perspective. *Pediatrics*, *138*(3). Published Online (date) August 25, 2016 doi: 10.1542/peds.2016-1278

Stull, D. E., Leidy, N. K., Parasuraman, B., & Chassany, O. (2009). Optimal recall periods for patient-reported outcomes: Challenges and potential solutions. *Current Medical Research and Opinion*, *25*, 929–942. doi:10.1185/03007990902774765

Sung, J. Y., Han, S. W., Chung, K. M., Lee, H., & Cho, S. H. (2014). Investigation of gender role behaviors in boys with hypospadias: Comparative study with unaffected boys and girls. *Journal of Pediatric Psychology*, *39*, 1061–1069. doi:10.1093/jpepsy/jsu055

Tishelman, A. C., Shumer, D. E., & Nahata, L. (2016). Disorders of sex development: Pediatric psychology and the genital exam. *Journal of Pediatric Psychology*, *42*, 530–543. doi:10.1093/jpepsy/jsw015

U. S. Food and Drug Administration. (2009). Guidance for industry: Patient-reported outcome measures: Use in medical product development of support labeling claims. Retrieved from http://www.fda.gov/downloads/Drugs/GuidanceComplianceRegulatoryInformation/Guidances/UCM193282.pdf.

Vayena, E., Brownsword, R., Edwards, S. J., Greshake, B., Kahn, J. P., Ladher, N. ... Tasioulas, J. (2016). Research led by participants: A new social contract for a new kind of research. *Journal of Medical Ethics*, *42*, 216–219. doi:10.1136/medethics-2015-102663

Wisniewski, A. B., & Sandberg, D. E. (2015). Parenting children with disorders of sex development (DSD): A developmental perspective beyond gender. *Hormone and Metabolic Research*, *47*, 375–379. doi:10.1055/s-0034-1398561

Zucker, K. J. (2002). Re: Sexual behaviors, sexual orientation and gender identity in adult intersexuals: A pilot study. *Journal of Urology*, *168*, 1507–1508; author reply 1508. doi:10.1097/01.ju.0000027943.36766.11