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Methodological Issues for Psychological Evaluation across the Lifespan of Individuals with a Difference/Disorder of Sex Development

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Abstract
The aim of the current report is to provide guidance relevant to psychological evaluation for healthcare providers and researchers working in the field of disorders of sexual development (DSD). In doing so, we give careful consideration to methodological issues and limitations that may influence the utility of investigations. For example, rarity and heterogeneity of DSD conditions restrict sample sizes when conducting evaluations aimed at establishing condition-specific psychological outcomes. At the same time, the potential for stigmatization by virtue of conducting psychological evaluations is particularly high given the fundamental contribution of sex and gender to one's sense of self and integrity. This article will provide basic theory for psychological evaluation as well as give a review of specific measures that can be employed for clinical purposes depending on a variety of parameters, including life stage of the patient and goal(s) of the evaluation. Care providers and service users may benefit from guidance in coping with the difficulties inherent in having and/or treating DSD. The potential for identification with the patient with DSD is higher than in other domains of medicine because sexual and gender identities are fundamental to all humans and are continually evolving from a sociological perspective.

Differences/disorders of sex development (DSD; also called "intersex conditions") refers to a group of conditions in which chromosomal, gonadal, or anatomical sex is atypical. Classification is based on genotype (46,XY DSD, 46,XX DSD, or sex chromosome DSD) with further distinctions in terms of gonadal development and androgen synthesis, action, or excess [Pasterski et al., 2010a]. The incidence depends on the condition, with a range of 1:300–1:15,000 [Hiort et al., 2014]. In some cases, especially those diagnosed at birth, sex/gender may be ambiguous, requiring decisions about gender allocation [Houk et al., 2006; Balsamo et al., 2016] and possibly surgical intervention [Mouriquand et al., 2016]. In cases diagnosed later in childhood or adolescence, considerations centre on psychosexual development and assimilation of implications of the diagnosis/condition. Having a DSD

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can heavily impact an individual's health status and/or psychological well-being in each life stage [Cohen-Kettenis, 2010; D'Alberton et al., 2015]. Though scientific understanding of the pathophysiology underlying DSD has greatly increased in recent decades, mostly from studies of mammalian sex development [Eggers et al., 2014] and genetic underlying mechanisms [Ono and Harley, 2013], conclusive knowledge about clinical outcomes is lagging [Schober et al., 2012; Roen and Pasterski, 2014].

Historically, many healthcare providers have taken a paternalistic view of disclosure of DSD to parents and patients [Morris, 1953; Karkazis, 2008]. This perspective was based on the belief that some information may be too difficult to understand/assimilate and the parent(s) or patient would suffer less for not knowing. Unfortunately, many patients would have ultimately discovered withheld information at some point, usually shrouded in some degree of secrecy [Brinkmann et al., 2007]. Considering DSD within the context of secrecy has been noted to induce feelings of shame, which could be integrated into the sense of self, potentially impairing self-esteem and/or a healthy psychosexual development. Fortunately, disclosure practices and communication about DSD have improved greatly, with a renewed focus on helping patients assimilate full knowledge about their condition and to cope with sometimes difficult consequences [D'Alberton, 2010; Liao et al., 2010].

Approaches to Psychological Evaluation in DSD

There are 3 primary functions of psychological evaluation in DSD: (1) for conducting research aimed at elucidating mechanisms underlying normative sex development [Hines, 2011], which may be applied to understanding development in the context of DSD; (2) for conducting research aimed at charting clinical outcomes for specific conditions or specific features of one or more conditions [Strandqvist et al., 2014]; or (3) for assessment of psychological functioning for clinical purposes in management of the individual patient [Cohen-Kettenis, 2010]. Methodology may differ depending on the goal(s) of the evaluation, and the goal(s) of the evaluation may impose limitations to its usefulness.

Methodological limitations of psychological evaluation in DSD may be considered in the approaches described above. Even when the goal is individual assessment, for example, to identify a particular patient's strengths and weaknesses and/or level(s) of psychological and emotional functioning, much of the interpretation of results will depend on previously published studies of norms or group-specific outcomes. In this regard, methodological limits of primary concern pertain to studies of groups of patients and include (1) small sample sizes in individual studies, which may impair statistical analysis and/or generalizability [Button et al., 2013]; (2) selection bias in individual studies, which may impair generalizability to groups as a whole whose characteristics are not well represented by the sample [Hernán et al., 2004]; (3) reliance on retrospective reports, which introduces error in terms of possible false-negative or false-positive accounting of events [Hardt and Rutter, 2004]; and (4) lack of standardized assessment across replication studies and/or across studies of different groups [Roen and Pasterski, 2014]. As with all psychological investigations, attention to goals and methodological limitations is critical for psychological evaluation in DSD.

Psychological Evaluation in Normative Research

Theoretical studies aimed at elucidating mechanisms underlying normative gender-related development have often included individuals with DSD conducted in academic centres linked to hospitals with clinics providing specialist care [Pasterski et al., 2007, 2011; Hines et al., 2016]. Alterations in hormone exposure in different critical periods, for example, offer insights into the development of (potentially) hormone-dependent sequelae, such as changes in gender-related behaviour or sexual functioning. Such studies are critical as experimental manipulation of biological parameters could not otherwise be ethically studied in humans. Nevertheless, the underlying assumptions and research aims are not always directly clinically relevant, and individuals with DSD included in normative studies may feel exploited or that they are being used as "guinea pigs." Such impressions among patient groups have led to reluctance to participate in any form of research. Compounded by the rarity of specific conditions, the result is small sample sizes, which represents the primary methodological limitation in normative research involving participants with DSD. The problem of retrospective bias is less of an issue in theoretical research. Such studies often evaluate performance on a task or current preferences/attitudes in comparison to unaffected control groups. Furthermore, research aimed at informing theory often employs assessment tools that have been standardized on larger populations [Pasterski et al., 2015].

Selection bias and small samples notwithstanding, some normative studies have bridged the gap between elucidating mechanisms of development and providing

results which may directly inform clinical care. For example, 2 studies including children and adults with congenital adrenal hyperplasia have shown reduced short term memory [Browne et al., 2015; Collaer et al., 2016], with the working hypothesis that long-term treatment with glucocorticoids may impair hippocampal function. The implications here are obvious. Even so, the effort should always be made to consider the human side of research participants and to be mindful of their vulnerabilities and needs.

Psychological Evaluation in Charting Outcomes

The extant literature involving patients with DSD includes a large body of reports aimed at assessing clinical outcomes as a function of various parameters [Dessens et al., 2005; Cohen-Kettenis, 2010; D'Alberton et al., 2015]. Some studies report a range of findings from evaluation of outcomes in a single condition [Meyer-Bahlburg et al., 2006], whereas others have centred on singular features common to several conditions [Meyer-Bahlburg, 2005]. In both cases, clinical relevance depends, in part, on stages of development across the life-span. As an example, outcomes may be reported for assimilation of (assigned) gender identity in early to middle childhood [Pasterski et al., 2015], while other studies report outcomes for sexual functioning in adulthood, relevant to clinical interventions such as surgical correction of genital anomalies [Callens et al., 2012a]. Though such studies of clinical outcomes conducted by a primary care team may include most or all cases presenting in a particular centre, the methodological limitation of small samples remains, given the rarity of conditions and specificity to developmental stages. In an effort to increase sample sizes, many studies have included wide age ranges, spanning life stages. Many participants will be asked to recall past experiences, which introduces the potential for retrospective bias, though some studies have found methods for confirming recalled reports [Rieger et al., 2008], and others report retrospective findings that are consistent with prospective reports and are useful in developing predictive models [Callens et al., 2016].

Further potential methodological complications relate specifically to the evaluation of physical, psychological, and emotional sequelae of surgical intervention for genital malformations [Lucas-Herald et al., 2016; Mouriquand et al., 2016]. Although such studies often employ standardized measures for current functioning, conclusions may not apply to clinical practice going forward. This is because surgical approaches and techniques have changed radically in recent decades [Callens et al., 2012a], and it can be difficult to ascertain details of interventions performed up to 20 or 30 years earlier, making it near impossible to draw meaningful conclusions. By the time the extent of sexual functioning, for example, can be fully appreciated, it is likely that the surgical approach related to the outcome will be obsolete. Furthermore, the heterogeneity of conditions and centre-specific treatment approaches makes it difficult to provide appropriate comparison groups that would be needed to establish genuine, independent, factors affecting psychological outcomes. Because randomized clinical trials are not possible, it is incumbent upon clinical researchers to employ rigorous methodology, paying particular attention to the relevant use of specific measures – for example, those utilizing patient-/parent- versus examiner-dependent judgements about outcome [Nokoff et al., 2017]. As technology improves and interventions (potentially) become standardized, there should be less variance across time. Also, attention to record keeping or inclusion in prospective studies could reduce the problem of attrition in longitudinal studies and lead to greater applicability of outcomes to current and future clinical management, including benefits and harms of specific medical procedures.

Psychological Evaluation of Individuals for Clinical Management

Psychological evaluation of individuals diagnosed with DSD is usually implemented on a case-by-case basis as a function of identified needs and as a part of individual clinical management. This approach utilizes instruments that assess general psychological and emotional functioning/well-being as well as those that target functioning specific to aspects of DSD, such as sexual functioning [Callens et al., 2012b], gender-related development, and health-related quality of life [Alpern et al., 2017]. Clinical instruments with widely published norms as well as those with published outcomes in studies of DSD may be applied. In this case, methodological considerations are those relevant to the specific instruments and to outcome studies, as mentioned above. However, further methodological considerations pertain to the approach and goal(s) of individual evaluations, including attention to a patient's particular needs and the patient-healthcare professional relationship. For example, a study looking at stakeholder experiences of interprofessional teamwork in DSD revealed that consistency among team members and review with professionals known to the family was described as very helpful in coping with clinical aspects of DSD [Sanders et al., 2017].

Addressing Methodological Limitations

Given the implications for clinical management in DSD, overcoming methodological limitations in studies of outcomes has been an area of focus. Toward that end, international collaborators have come together to systematically address shortfalls in outcome research. One such collaboration has been established within the framework of "DSDnet" (http://www.dsdnet.eu), a network granted by the EU-funded program European Cooperation in Science and Technology (COST). From the outset, key goals have been to establish a consensus on best practices for standardized treatment, assessment, and longitudinal follow-up of individuals with DSD across the lifespan as well as to provide specialist training for all subspecialties within multidisciplinary teams (MDTs). This includes specialist training for psychologists, with an emphasis on the key role psychology plays not only in clinical case management but in coordination of MDT activities and guidance for team members regarding psychological impact of patient-care provider relationships.

Crucial to the process of facilitating such a large-scale, multi-centre assessment of outcomes is the establishment of a centralised DSD registry. A key feature of the DSDnet programme is the I-DSD Registry. MDTs can incorporate basic patient data, with predetermined parameters agreed upon in consensus meetings of a multidisciplinary group of healthcare providers, service users, and patient advocates. The proposal reflects special attention to phenotypical aspects, associated morbidities, mental health, and gender-related development in DSD. Life stage specific parameters have been identified at ages 1 month, 4 and 8 years, prepuberty, and between ages 18–25 years, 25–40 years, 40–60 years, and 60–75 years. When implemented, the "phenotyping tool" will allow practitioners from participating centres across Europe to collate information in a standardized and systematic fashion. The registry will also allow specialists to identify large samples of individuals with specific diagnoses for further study, with the possibility for inclusion in studies using more in-depth protocols. If successful, limitations such as small sample size, selection bias, retrospective reporting bias, and lack of standardized assessment should be greatly reduced.

Qualitative and Quantitative Evaluation in DSD

DSD represent a constellation of conditions unique in all of medicine. This is because sex development is fundamental to identity, social acceptance, and individual integrity. At the same time, however, many aspects of sex development border on the social taboo. For this reason, much of what we know about the experiences of patients and families living with DSD is comparatively recent, and there has been a strong focus on exploratory investigations. This qualitative approach is widely used in social sciences [Lichtman, 2013] and allows researchers some degrees of freedom or openness when taking in information. Whereas quantitative methods use systematic empirical investigation of observable phenomena and statistical analysis of large datasets, qualitative investigations are more loosely structured and often report on single case studies or small samples. In the case of DSD, this approach has been useful in terms of understanding the unique experiences of patients and their families [Liao et al., 2014; Lundberg et al., 2016]. It has been argued that in order to deliver patient-centred care, information about how patients experience, how they make sense of their lives, and how they understand their condition is important [Lundberg et al., 2016].

Psychological Evaluation for Clinical Management: Guidance across Life Stages

It is widely accepted, and has been formally issued in a Consensus Statement [Lee et al., 2006], that DSD healthcare should be holistic and conducted within MDTs of sub-specialists, including endocrinology, urology, psychology, specialist nursing, social work, genetics, and medical ethics. A close collaboration between team members is essential for holistic care, from communication of the diagnosis to longer term follow-up investigations. The patient-centred approach also requires a considered attention to individual patient and family needs. While guidance for clinical care for individual diagnoses have been established [Speiser et al., 2010; Hughes et al., 2012], MDTs may also be guided by needs and concerns communicated by patients and families.

Nomenclature and Communication

As part of the Consensus Statement on Clinical Management of Intersex Disorders [Houk et al., 2006], a new nomenclature was established, reflecting the growing sentiment that previous terminology, e.g., "pseudohermaphrodite," was insensitive or pejorative. The umbrella term "intersex" was replaced with "disorders of sex development." In parallel, and stemming from greater under-

standing of genetic underlying mechanisms, the classification system was changed to reflect known mechanisms and eliminate a system organized around the appearance of the genitalia. The new system is organized around sex chromosomes and subsumed mechanisms of dysfunction, e.g., androgen synthesis errors, which more clearly incorporates conditions where there is no genital ambiguity, such as androgen insensitivity syndrome [Pasterski et al., 2010a]. Nevertheless, the term "disorders of sex development" remains unacceptable to some who prefer "differences" or "divergence" as less stigmatizing or medicalized. In a more sociologically based movement, a subset of individuals have re-appropriated the term "intersex." However, this seems to represent an identity, rather than any specific condition(s) and may or may not include individuals with diagnoses that fall under the umbrella formerly called intersex.

Though the term DSD proposed in the Consensus Statement appears to have been widely adopted by clinicians and researchers [Pasterski et al., 2010b], some patient (advocacy) groups remain dissatisfied. Practically speaking, the issue of agreed upon language is yet unresolved, in part due to the methodological limitation of participant self-selection in the few studies/statements published on the topic. Those who accept the DSD terminology may not be part of groups speaking out. In addition, it is likely that not all healthcare professionals working in this domain use the umbrella term "DSD" and so many participants may not have encountered it. Interestingly, a study including families of 121 patients in Turkey have suggested the preference for Latin disease terms that avoid inclusion of the word "sex" [Tiryaki et al., 2017]. Such findings might ultimately contribute to resolving other tensions around the use of the umbrella term (which includes the word "sex") for individuals with diagnoses such as Turner or Klinefelter syndrome, who do not see their conditions as related to sex development. The continued controversy, including the re-appropriation of the term "intersex," reflects sensitivities within the domain of sex development, making it clear that simply changing the nomenclature is far from sufficient when considering language used with patients and families.

With respect to communication among members of the MDT itself, attention should be paid to conveying information to patients and families in a consistent and comprehensible manner. Toward this end, each team member should have some basic knowledge within others' domains and should be kept informed, to the extent possible, of the state of disclosure/discussion that has taken place. Inconsistent or confusing information can cause distress, especially in early stages of the diagnostic process. As an example, a study assessing traumatic stress in parents of children diagnosed with DSD found that those parents who experienced confusion about the diagnosis experienced greater levels of distress [Pasterski et al., 2014]. More specifically, they found that parents whose child was "undiagnosed," in the sense that the genetic mutation underlying a presentation of androgen synthesis impairment was not determined, experienced the highest levels of traumatic stress. Even though the clinical diagnosis and the treatment protocol had been established, communication of the circumstance left them feeling less in control or that they could not move to the next phase of coping. In such cases, clearer communication that the clinical diagnosis determines management and that the mutation may be less relevant could reduce psychological discomfort. Consider the following example: most patients and physicians will accept a diagnosis of influenza without ever knowing the specific virus causing the infection. The clinical diagnosis can be made and treatments are the same, regardless. Though DSD is clearly more serious than influenza, such conceptualizations may be useful for parents of children with "undiagnosed" XY DSD. Goals may be restructured to include best treatment options, adherence, and future monitoring of outcomes, rather than extensive testing to find the mutation. The use of a term that does not include "undiagnosed" could also be useful.

Finally, the process of communication between MDT members and with patients and families may provide the secondary function of conveying information about gender-related beliefs and norms, whether or not it is intended. Given the wide range of gender experiences of patients with DSD and their families, it is critical that judgement about gender (non)conformity is minimized to avoid stigmatization. Attention to one's own assumptions about a (non)binary gender framework may be helpful in providing a holistic support for patients and families and for reducing (potential) bias in data collection as part of psychological assessment.

Objectives of Psychological Evaluation

In considering the course for psychological/psychosocial/psychosexual evaluation of individuals (and families) with DSD, attention should be given to the objective, the evaluation itself. In all stages of clinical care, there is the possibility for unintended iatrogenic effects, in this case caused by stigmatization [Hughes et al., 2007; Meyer-

Bahlburg, 2010]. While the patient-centred clinical approach aims to investigate parameters of well-being relevant to long term quality of life and to provide the patient with insight/feedback regarding their unique experience, unsolicited investigations can have unwanted effects [Crissman et al., 2011].

Ideally, patients and families with DSD should be fully supported by the MDT, with the provision of psychological support as a standard of care. However, even though it seems that most primary care centres include this subspecialty as part of the MDT [Pasterski et al., 2010b; Kyriakou et al., 2016], one study of practices across Europe found that the majority of families were not taking up the offer of psychological support [Pasterski et al., 2010b]. One interpretation of this finding is that patients and families may associate psychological support with mental illness and may have found the prospect stigmatizing. Perhaps a greater effort toward establishing good rapport from the outset and normalizing participation of the psychological support staff (i.e., psychologists, psychiatrists, and social workers) could open the channels of communication, allowing for patients and families to reach out without feeling stigmatized. Additional barriers to engaging with psychological support services may include distance from the centre of expertise or financial burdens associated with taking up the support. In either case, careful consideration should be given to objectives and benefits of psychologically based evaluations as well as to potential hidden barriers before concluding that any individual/family should be repeatedly invited or altogether dismissed from the potential engagement.

Domains of Psychological Evaluation in DSD

Like other types of chronic disease, a holistic care model for DSD requires attention to all areas of functioning, as the complexities of chronic disease have implications for psychological, psychosocial, psychosexual, emotional, and cognitive functioning. With respect to DSD, some of these specific domains, which can be subsumed under the heading of psychological evaluation, overlap with those relevant to general functioning/well-being, though some carry particular importance. Table 1 gives a selection of standardized tests, both qualitative and quantitative, that have been used with patients and in studies of DSD. Categories are broken down in terms of areas of functioning/well-being as well as appropriateness at different life stages. In the following section we highlight stage-dependent considerations and present case studies to illustrate the diversity of presentation and conditions relevant to psychological evaluation. Note that the five case studies presented are for illustrative purposes only. Though these cases represent genuine clinical experience, they do not represent the full range of concerns faced by patients and families living with DSD, nor does any individual case represent all cases of its type.

Evaluation at Different Life Stages

Parents of the Newborn or Young Child with DSD

Psychological evaluation in the case of a newborn or infant diagnosed with DSD pertains primarily to parents. Many studies have documented the distress associated with learning of a DSD diagnosis [De Silva et al., 2014; Pasterski et al., 2014; Suorsa et al., 2015]. Of particular relevance in these cases is emotional status/functioning, levels of (traumatic) stress, parent-child bonding, and ability to cope effectively with the challenges inherent in caring for their child in the early stages. Secondary assessment may be made to identify areas of need (e.g., social support). Clear and consistent communication of information related to diagnosis and decisions about clinical management is essential and should be followed up to ascertain the parents' understanding of relevant information.

Making decisions about gender assignment and/or surgical interventions can be a source of acute distress in parents, and the process of psychological evaluation may be a point of entry for providing much needed additional clinical support [see Mouriquand et al., 2016 for review of gender assignment and related surgical intervention]. For example, though certain procedures are medically necessary for survival or optimal health outcomes for the infant (e.g., correction of the midline at the point of the genitalia in cloacal exstrophy or orchiopexy), the medical necessity of certain other procedures is debated. Likewise, guidance for gender assignment is not conclusive in all cases of DSD. The potential for violating the child's right to bodily integrity in the case of a nonnecessary surgical procedure, or to self-determination of gender identity/role, may be a source of intense distress and guilt for parents. In conducting general evaluations, clinicians and researchers should aim to exercise utmost sensitivity where parents find themselves in such difficult positions. The need for added guidance by the MDT in the decision-making process may be flagged as a part of the process.

Table 1. Standardized tests used in studies of DSD

Domain	Measures	Life stage	Reference
Quality of life	Health-related Quality of Life (HQoL)	parents; adult patients	[Malouf et al., 2010]
	World Health Organization Quality of Life Assessment (WHOQoL-BREF)	parents; adult patients	[D'Alberton et al., 2015; Leithner et al., 2015]
	Questionnaire for Adult's Health-Related Quality of Life (TAAQoL)	parents; adult patients	[Callens et al., 2014]
Psychological functioning/ psychopathology	Achenbach Adult Self-Report (ASR; for psychopathology)	parents; adult patients	[Achenbach, 1997; Callens et al., 2014]
	Achenbach Youth Self-Report (YSR; for psychopathology)	adolescent patients	[Ivanova et al., 2007]
	Beck Anxiety Inventory (BAI)	parents; adolescent/adult patients	[Beck et al., 1988; Suorsa et al., 2015]
	Beck Depression Inventory-II (BDI-II)	parents; adolescent/adult patients	[Beck et al., 1996]
	Brief Symptom Inventory (BSI)	parents; adolescent/adult patients	[Derogatis and Spencer, 1993]
	Centre Epidemiological Studies Depression Scale (CES-D)	parents; adolescent/adult patients	[De Silva et al., 2014]
	Child Behavior Checklist (CBCL)	child patients/siblings	[Achenbach, 1993; deVries et al., 2016]
	Child Depression Inventory (CDI)	child patients/siblings	[Helsel and Matson, 1984; Poomthavorn et al., 2009]
	Impact of Events Scale – Revised (IESR)	parents; adolescent/adult patients	[Creamer et al., 2003; Pasterski et al., 2014]
	State-Trait Anxiety Inventory (STAI)	parents; adolescent/adult patients	[Spielberger, 2010]
	Strengths and Difficulties Questionnaire (SDQ)	child/adolescent patients/siblings	[Goodman, 1997; Poomthavorn et al., 2009]
	Symptom Checklist-90-Revised (SCL-90-R)	parents; adolescent/adult patients	[Derogatis, 1992; Fliegner et al., 2014]
Personality and identity	Gender Identity Interview for Children (GIIC)	child/adolescent patients	[Zucker et al., 1993; Pasterski et al., 2015]
	Gender Identity Questionnaire (GIQ)	child/adolescent patients	[Cohen-Kettenis et al., 2006; Pasterski et al., 2015]
	Gender Role Questionnaire (GRQ)	child/adolescent patients	[Zucker et al., 2006; Nermoen et al., 2010]
	Junior Dutch Personality Questionnaire	child/adolescent patients	[Scholte and De Bruyn, 2001; Nermoen et al., 2010]
	Personal Attributes Questionnaire (PAQ)	parents; adolescent/adult patients	[Helmreich et al., 1981; Schützmann et al., 2009]
Sexuality and sexual functioning	Female Sexual Distress Scale-Revised (FSDS-R)	adolescent/adult patients	[Derogatis et al., 2008; Callens et al., 2012b]
	Female Sexual Function Index (FSFU)	adolescent/adult patients	[Rosen et al., 2000; Callens et al., 2012b]
	Male Sexual Health Questionnaire (MSHQ)	adolescent/adult patients	[Rosen et al., 2004; Wolfe-Christensen et al., 2012]
	Romantic and Sexual Interest Questionnaire	adolescent/adult patients	[Menke et al., 2010]
	The Sexual Awareness Questionnaire (SAS)	adolescent/adult patients	[Snell et al., 1991]
Body image (experience)	Body Experience Questionnaire	adolescent/adult patients	[Schweizer et al., 2016]
	Body Image Scale (BIS)	adolescent/adult patients	[Callens et al., 2014]
	Body Parts Satisfaction Scale-Revised (BPSS-R)	adolescent/adult patients	[Petrie et al., 2002; Ros et al., 2013]
Social support	Maternal Social Support Scale (MSSS)	mothers	[Feldman et al., 2000]
	Parenting Stress Index Short Form (PSI/SF)	parents	[Abidin, 1990; De Silva et al., 2014]
	Scale of Perceived Social Support (SPSP)	parents; adolescent/adult patients	[Zimet et al., 1990]
Life satisfaction	Rosenberg Self-Esteem Scale	parents; adolescent/adult patients	[Rosenberg, 1965; Callens et al., 2014]
	Satisfaction with Life Scale (SLS)	parents; adolescent/adult patients	[Diener et al., 1985]

Case Study

Greg's parents were referred to our centre when the baby was less than 1 month old and had atypical external genitalia. The infant presented with severe hypospadias, bifid scrotum, a palpable inguinal testis on the right side, and no palpable gonad on the left. Ultrasound and voiding cystourethrogram revealed a vaginal canal that joined the perineal urogenital sinus. Residual müllerian structures were characterized by an apparent small emiutero on the left with an adjoining tube. A left gonad could not be visualized. Genetic testing revealed a diagnosis of chromosomal mosaicism with a 45,X/46,XY karyotype. Some interviews were conducted by the pediatric endocrinologist and psychologist to assess the child's condition. The parents were given the opportunity to ask any questions they had about the diagnosis, and the basic pros and cons regarding sex of rearing were explained, though they were also told that further surgical and genetics input would be required to complete the diagnostic process.

Greg's parents were then directed to the pediatric endocrinologist and psychologist, depending on the aspects of care they wanted to discuss. Despite the presence of a complex problem, the emotional atmosphere surrounding the parents was relaxed and no further psychological assessment was conducted at the time. It was agreed that should they feel the need for additional psychological support/investigation later in life, they would do so as part of the continuation of clinical care. After this series of interviews, the parents decided to register their newborn as male.

The Patient in Childhood (4–12 Years Old)

For the patient in early to mid-childhood, psychological evaluation should be considered for the developing child as well as the parents. In the case of a new diagnosis in childhood, considerations for assimilation of the diagnostic information by the parents are the same as for those whose child was diagnosed at birth. While parents may experience shock at learning the diagnosis, they may be slightly reassured having experienced a period of relatively good health with their child. In this case, they may feel less unsure than when a child is diagnosed in moments/days after birth. Special attention to emotional and cognitive development of the child and to family dynamics, such as parent-child attachments is important. The child may be monitored through discussion with the parents for any signs of gender incongruity or distress in the assigned gender. For the child approaching or at the start of puberty, attention to the developing sense of sexual identity may be warranted.

Case Study

Julia's parents arrived in consultation when Julia was nearly 4 years old. Present in the consultation were the pediatric endocrinologist and the psychologist, who were the first to meet the parents and who spoke on behalf of other members of the MDT who were subsequently involved. In the first session, they talked about the clinical history of their daughter and their various presentations at other health care institutions throughout the country. Their primary concern was atypical genitalia, discovered a couple of years after birth.

Julia was brought up as a female and named after her maternal grandmother who her mother had been very fond of. However, when Julia was nearly 4 years old the parents were told that something was wrong with her, which left them, especially the mother, in a state of great despair. They struggled to decide what was best for Julia and worried about how she would accept her "problem" and how they could manage to explain it to their relatives and friends.

Over the course of several meetings, the psychologist would meet the parents while Julia played with a trainee, except for once when she remained in the room with her parents. Julia displayed difficulties in coping with issues that were too complex for a 4-year-old girl. At the thought of being "male or female" she stiffened, ground her teeth and gave a sort of howl, banging her head with her hands, as if she wanted to express the unbearable concrete form of her bodily sensations that could not be transformed into thoughts. Medical evaluations pointed to a male reassignment, and the parents became more and more familiar with the idea that Julia could actually be a boy and consequently experimented in dressing "her" as a "him" to see how they and Julia would feel. "All of a sudden he changed. He does not seem to be the child I brought up for 4 years. He plays football, jumps, and runs. He is free now, but we have been torturing him for 4 years" was the mother's report. She added, "The last 15 days have been the happiest of our lives. We did not feel forced anymore to keep secrets, even from our closest friends, and we found a great understanding. Now we feel free to let him go to the loo with someone else without accompanying him, fearing that someone could realize that there was something wrong."

Julian's fitting in to his new male identity was confirmed when he met the MDT. Three years after meeting "Julia," the teams could not see any reason not to support the parents' choice. They seemed to have made the choice competently, considering pros and cons, and involved their son in the decision as far as possible. Now Julian is

attending the first year of a primary school, and he feels and behaves as a typical little boy. When he asked questions, Julian's parents replied in the best way they could within their knowledge. He clearly needed to be informed about tests and any medical or surgical interventions. He often asked his parents, "Are you lying to me?" They never lied to him and involved him in every decision they made. The family continued to meet the psychologist from time to time, and Julian had the opportunity to ask any questions that came to his mind.

In the most recent meeting with Julian, who was at the threshold of puberty, he seemed contented with his life and things were developing in a positive fashion. He did not have any questions for the psychologist. However, in the course of the meeting, he spoke about the magical characteristics of his home town, including a buried civilization discovered beneath the current construction. He made the striking distinction between "grottos" hidden below ground, over which the new city had been rebuilt. The way in which the family's relationship with the team was developing gave hope that he would have reliable support when the time comes to explore the "grottos" of his personal history.

Adolescence (13–18 Years Old)

Adolescence can be a difficult time for anyone, and added stress of living or being newly diagnosed with a DSD can have detrimental effects. By this time, MDT attention to the patient's pubertal development will be important along with aspects of the co-evolution of gender and sexual identity. Health-related quality of life may be an area of concern and, as with previous stages, psychological support should remain available to parents [Bennecke et al., 2015]. For young patients faced with the possibility of surgical interventions at/after puberty, additional psychological support and/or assessment may be useful as part of the decision-making process.

Case Study

The first time Erika visited the endocrinologist, she was also invited to meet with the psychologist at a later date. She was 17 years old, and her life had been unremarkable until it was noticed that she had not yet entered menarche and showed no breast development. The diagnosis of complete androgen insensitivity syndrome (CAIS) hit her like a ton of bricks, and she fell into a wordless pain that rendered her virtually silent throughout the psychological interviews. There was visible anguish of such intensity that the psychologist feared she would throw herself out the window when she took a break to use the bathroom. The psychologist stood near the door and was relieved when she came out alive. Erika did not return to see the psychologist, though she continued contact with the endocrinologist and later with the gynaecologist for hormone replacement therapy. Erika's parents remained in contact with the psychologist. They seemed overwhelmed by grief, having noticed deterioration in their daughter's mood, and they asked for advice. They followed the advice to contact a support group, and this seemed to help a great deal. With time, they became close with their daughter, whose sadness seemed to slowly lift. She gradually managed to cope with her new reality to integrate it in her sense of self.

Adulthood (18–50 Years Old)

At this stage of life, psychological evaluation may be considered as part of a bio-psycho-somatic approach to reinforcing a patient's experience of wholeness. In adulthood, patients may be more open to psychological evaluation and support. Psychological and emotional well-being remains important, as does health-related quality of life. Special attention should be given to concerns about romantic relationships and establishing a family of one's own. Doubts about one's gender-related role(s) in family and society may arise and attention to social support systems could be key.

Case Study

Paola presented with ambiguous genitalia at birth, and a 46,XY karyotype was established in the diagnostic process. She was reared as male, but suffered a great deal of dysphoria about her gender. She has since chosen gender reassignment to female and seemed to be happy in herself and with her life as a woman. In psychological interviews, however, she again questioned her gender identity, considering her DSD to be the root cause of loss of desire and marital problems. Further enquiry revealed a fear of having to care for her unwell husband. It seemed for her that the end of the illusion of remaining a young, married couple forever was destabilizing. The discussion centred on the issue of growing old together and evitability of changing sexuality. The fear of not being ready to adapt as her role as a woman made her feel deficient. She attributed perceived inadequacies in the context of not being a "real" or genetic woman. Working with the psychologist, she was able to put her DSD diagnosis in context as a "part of her" but not "who she is," which allowed her to move to a point of considering the normality of her fears all along.

Later Life (50+ Years Old)

Little is known about the very long-term outcomes for patients with DSD, however, considerations for psychological evaluation and support should be similar to many of those in earlier adulthood. Attention to health-related quality of life may be of particular importance given implications for effects of long-term hormone replacement therapy in some patients. Familial relationships and social support continue to be a source of interest throughout adulthood. As in earlier adulthood, concepts of gendered aspects of growing older should be considered.

Case Study

Sara, 63, diagnosed with CAIS, is a woman successful in her career, but who has lived her life alone. She has always felt different from her family and suspected that they never expected anything from her. Having felt alone for most of her life, Sara became closed within herself, feeling a sense of shame and a real fear of rejection. In this state, she never considered the possibility of authentic relationships. Only in her work had she ever felt satisfied with herself. Recently, however, a man has entered her life, and the fear of revealing her true "identity" prompted her to flee. The diagnosis had become a wall behind which she could hide her fears about trust in relationships. Over time, however, Sara decided to face her fear and to trust a partner. Finding that she could be accepted in totality, including her diagnosis of CAIS, allowed her to feel an authenticity that she believed she could not achieve. Though the resolution came late in life, she was thankful for having achieved the insight and self-acceptance that came from being wholly accepted by others.

Conclusion

The DSD umbrella encompasses a heterogeneous array of rare and complex conditions. This combination of characteristics contributes to methodological challenges with respect to psychological and other evaluations. Further, specific domains of evaluation, e.g., theoretical studies of normative development or outcome studies meant to inform clinical management, are met with greater or fewer challenges depending on the goals of the evaluation. Historically, primary hurdles have included small sample sizes due to rarity of individual conditions, sample bias due to participant self-selection, reliance on retrospective reporting secondary to small samples and rarity, and lack of standardized assessment across reports of evaluations. Interpretations of results of individual evaluations for clinical management are also limited as a result of the limitations affecting research reports. Thankfully, clinicians and researchers are now able to create large-scale, multi-centre collaborations that may overcome some of these methodological limitations. These and other future studies should aim to employ widely standardized assessments. Reliable findings may then be directly or indirectly applied to clinical care across the life span for individual patients and their families living with DSD.

References

Abidin RR: Parenting Stress Index-Short Form (Pediatric Psychology Press, Charlottesville 1990).

Achenbach TM: Empirically Based Taxonomy: How to Use Syndromes and Profile Types Derived From the CBCL/4–18, TRF, and YSR (Department of Psychiatry, University of Vermont, Burlington 1993).

Achenbach TM: Manual for the Young Adult Self-Report and Young Adult Behavior Checklist (Department of Psychiatry, University of Vermont, Burlington 1997).

Alpern AN, Gardner M, Kogan B, Sandberg DE, Quittner AL: Development of health-related quality of life instruments for young children with disorders of sex development (DSD) and their parents. J Pediatr Psychol 42:544–558 (2017).

Balsamo A, Ghirri P, Bertelloni S, Scaramuzzo RT, D'Alberton F, et al: Disorders of Sexual Development in Newborns, in Neonatology: A Practical Approach to Neonatal Diseases, pp 1–24 (Springer, New York 2016).

Beck AT, Epstein N, Brown G, Steer RA: An inventory for measuring clinical anxiety: psychometric properties. J Consult Clin Psychol 56:893-897 (1988).

Beck AT, Steer RA, Brown G: Beck depression inventory-II. San Antonio 78:490–498. (1996).

Bennecke E, Werner-Rosen K, Thyen U, Kleinemeier E, Lux A, et al: Subjective need for psychological support (psysupp) in parents of children and adolescents with disorders of sex development (DSD). Eur J Pediatr 10:1287–1297 (2015).

Brinkmann L, Schuetzmann K, Richter-Appelt H: Gender assignment and medical history of individuals with different forms of intersexuality: evaluation of medical records and the patients' perspective. J Sex Med 4:964–980 (2007).

Browne WV, Hindmarsh PC, Pasterski V, Hughes IA, Acerini CL, et al: Working memory performance is reduced in children with congenital adrenal hyperplasia. Horm Behav 67:83–88 (2015).

Button KS, Ioannidis JP, Mokrysz C, Nosek BA, Flint J, et al: Power failure: why small sample size undermines the reliability of neuroscience. Nat Rev Neurosci 5:365–376 (2013).

Callens N, van der Zwan YG, Drop SL, Cools M, Beerendonk CM, et al: Do surgical interventions influence psychosexual and cosmetic outcomes in women with disorders of sex development? ISRN Endocrinol 2012:276742 (2012a).

Callens N, De Cuypere G, Wolffenbuttel KP, Beerendonk C, van der Zwan YG, et al: Long-term psychosexual and anatomical outcome after vaginal dilation or vaginoplasty: a comparative study. J Sex Med 7:1842–1851 (2012b).
Callens N, Weyers S, Monstrey S, Stockman S, Van Hoorde B, et al: Vaginal dilation treatment in women with vaginal hypoplasia: a prospective one-year follow-up study. Am J Obstet Gynecol 211:228.e1–228.e12 (2014).
Callens N, van Kuyk M, van Kuppenveld JH, Drop SL, Cohen-Kettenis PT, et al: Recalled and current gender role behavior, gender identity and sexual orientation in adults with disorders/differences of sex development. Horm Behav 86:8–20 (2016).
Cohen-Kettenis P: Psychosocial and psychosexual aspects of disorders of sex development. Best Pract Res Clin Endocrinol Metab 2:325–334 (2010).
Cohen-Kettenis PT, Wallien M, Johnson LL, Owen-Anderson AF, Bradley SJ, Zucker KJ: A parent-report gender identity questionnaire for children: a cross-national, cross-clinic comparative analysis. Clin Child Psychol Psychiatry 3:397–405 (2006).
Collaer ML, Hindmarsh PC, Pasterski V, Fane BA, Hines M: Reduced short term memory in congenital adrenal hyperplasia (CAH) and its relationship to spatial and quantitative performance. Psychoneuroendocrinology 64: 164–173 (2016).
Creamer M, Bell R, Failla S: Psychometric properties of the impact of event scale -revised. Behav Res Ther 12:1489–1496 (2003).
Crissman HP, Warner L, Gardner M, Carr M, Schast A, et al: Children with disorders of sex development: A qualitative study of early parental experience. Int J Pediatr Endocrinol 2011:10 (2011).
D'Alberton F: Disclosing disorders of sex development and opening the doors. Sex Dev 4–5: 304–309 (2010).
D'Alberton F, Assante MT, Foresti M, Balsamo A, Bertelloni S, et al: Quality of life and psychological adjustment of women living with 46,XY differences of sex development. J Sex Med 6:1440–1449 (2015).
De Silva K, de Zoysa P, Dilanka W, Dissanayake B: Psychological impact on parents of children with congenital adrenal hyperplasia: a study from Sri Lanka. J Pediatr Endocrinol Metab 5–6: 475–478 (2014).
Derogatis LR: SCL-90-R: Administration, Scoring & Procedures Manual-II for the R (evised) Version and Other Instruments of the Psychopathology Rating Scale Series (Clinical Psychometric Research, Baltimore 1992).
Derogatis LR, Spencer P: Brief Symptom Inventory: BSI (Pearson, Upper Saddle River 1993).
Derogatis L, Clayton A, Lewis-D'Agostino D, Wunderlich G, Fu Y: Validation of the female sexual distress scale-revised for assessing distress in women with hypoactive sexual desire disorder. J Sex Med 2:357–364 (2008).
Dessens AB, Slijper FM, Drop SL: Gender dysphoria and gender change in chromosomal females with congenital adrenal hyperplasia. Arch Sex Behav 4:389–397 (2005).
de Vries AL, Steensma TD, Cohen-Kettenis PT, VanderLaan DP, Zucker KJ: Poor peer relations predict parent-and self-reported behavioral and emotional problems of adolescents with gender dysphoria: a cross-national, cross-clinic comparative analysis. Eur Child Adolesc Psychiatry 6:579–588 (2016).
Diener E, Emmons RA, Larsen RJ, Griffin S: The satisfaction with life scale. J Pers Assess 1:71–75 (1985).
Eggers S, Ohnesorg T, Sinclair A: Genetic regulation of mammalian gonad development. Nat Rev Endocrinol 11:673–683 (2014).
Feldman PJ, Dunkel-Schetter C, Sandman CA, Wadhwa PD: Maternal social support predicts birth weight and fetal growth in human pregnancy. Psychosom Med 5:715–725 (2000).
Fliegner M, Krupp K, Brunner F, Rall K, Brucker SY, et al: Sexual life and sexual wellness in individuals with complete androgen insensitivity syndrome (CAIS) and Mayer-Rokitansky-Küster-Hauser syndrome (MRKHS). J Sex Med 3:729–742 (2014).
Goodman R: The strengths and difficulties questionnaire: a research note. J Child Psychol Psychiatry 5:581–586 (1997).
Hardt J, Rutter M: Validity of adult retrospective reports of adverse childhood experiences: review of the evidence. J Child Psychol Psychiatry 2:260–273 (2004).
Helmreich RL, Spence JT, Wilhelm JA: A psychometric analysis of the personal attributes questionnaire. Sex Roles 11:1097–1108 (1981).
Helsel WJ, Matson JL: The assessment of depression in children: the internal structure of the child depression inventory (CDI). Behav Res Therapy 3:289–298 (1984).
Hernán MA, Hernández-Díaz S, Robins JM: A structural approach to selection bias. Epidemiology 5:615–625 (2004).
Hines M: Gender development and the human brain. Annu Rev Neurosci 34:69–88 (2011).
Hines M, Pasterski V, Spencer D, Neufeld S, Patalay P, et al: Prenatal androgen exposure alters girls' responses to information indicating gender-appropriate behaviour. Philos Trans R Soc Lond B Biol Sci 371:20150125 (2016).
Hiort O, Birnbaum W, Marshall L, Wünsch L, Werner R, et al: Management of disorders of sex development. Nat Rev Endocrinol 9:520–529 (2014).
Houk CP, Hughes IA, Ahmed SF, Lee PA: Summary of consensus statement on intersex disorders and their management. International intersex consensus conference. Pediatrics 2: 753–757 (2006).
Hughes IA, Nihoul-Fékété C, Thomas B, Cohen-Kettenis P: Consequences of the ESPE/LWPES guidelines for diagnosis and treatment of disorders of sex development. Best Pract Res Clin Endocrinol Metab 3:351–365 (2007).
Hughes IA, Davies JD, Bunch TI, Pasterski V, Mastroyannoloulou K, MacDougall J: Androgen insensitivity syndrome. Lancet 380:1419–1428 (2012).
Ivanova MY, Achenbach TM, Rescorla LA, Dumenci L, Almqvist F, et al: The generalizability of the youth self-report syndrome structure in 23 societies. J Consult Clin Psychology 5:729 (2007).
Kyriakou A, Dessens A, Bryce J, Iotova V, Juul A, et al: Current models of care for disorders of sex development – results from an international survey of specialist centres. J Rare Dis 11:155 (2016).
Karkazis K: Fixing Sex: Intersex, Medical Authority, and Lived Experience (Duke University Press, Durham 2008).
Lee PA, Houk CP, Ahmed SF, Hughes IA: Consensus statement on management of intersex disorders. International consensus conference on intersex. Pediatrics 2:e488–e500 (2006).
Leithner K, Naderer A, Hartung D, Abrahamowicz C, Alexopoulos J, et al: Sexual and psychosocial functioning in women with MRKHS after neovaginoplasty according to Wharton-Sheares-George: a case control study. PLoS One 4:e0124604 (2015).
Liao LM, Green H, Creighton S, Crouch N, Conway G: Service users' experiences of obtaining and giving information about disorders of sex development. BJOG 2:193–199 (2010).
Liao LM, Baker E, Boyle M, Woodhouse C, Creighton S: Experiences of surgical continence management approaches for cloacal anomalies: a qualitative analysis based on 6 women. J Pediatr Adolesc Gynecol 5:266–270 (2014).
Lichtman M: Qualitative Research for the Social Sciences (Sage Publications, Los Angeles 2013).
Lucas-Herald A, Bertelloni S, Juul A, Bryce J, Jiang J, et al: The long-term outcome of boys with partial androgen insensitivity syndrome and a mutation in the androgen receptor gene. J Clin Endocrinol Metab 11:3959–3967 (2016).
Lundberg T, Roen K, Hirschberg AL, Frisén L: "It's part of me, not all of me": Young women's experiences of receiving a diagnosis related to diverse sex development. J Pediatr Adolesc Gynecol 4:338–343 (2016).
Malouf MA, Inman AG, Carr AG, Franco J, Brooks LM: Health-related quality of life, mental health and psychotherapeutic considerations for women diagnosed with a disorder of sexual development: Congenital adrenal hyperplasia. Int J Pediatr Endocrinol 1: 253465 (2010).
Menke LA, Sas TC, Visser M, Kreukels BP, Stijnen T, et al: The effect of the weak androgen oxandrolone on psychological and behavioral characteristics in growth hormone-treated girls with Turner syndrome. Horm Behav 3: 297–305 (2010).

Meyer-Bahlburg HF: Gender identity outcome in female-raised 46,XY persons with penile agenesis, cloacal exstrophy of the bladder, or penile ablation. Arch Sex Behav 34:423–438 (2005).
Meyer-Bahlburg HF: From mental disorder to iatrogenic hypogonadism: dilemmas in conceptualizing gender identity variants as psychiatric conditions. Arch Sex Behav39:461–476 (2010).
Meyer-Bahlburg HF, Dolezal C, Baker SW, Ehrhardt AA, New MI: Gender development in women with congenital adrenal hyperplasia as a function of disorder severity. Arch Sex Behav 35:667–684 (2006).
Morris JM: The syndrome of testicular feminization in male pseudohermaphrodites. Am J Obstet Gynecol 6:1192–1211 (1953).
Mouriquand PD, Gorduza DB, Gay CL, Meyer-Bahlburg HF, Baker L, et al: Surgery in disorders of sex development (DSD) with a gender issue: If (why), when, and how? J Pediatr Urol 3:139–149 (2016).
Nokoff NJ, Palmer B, Mullins AJ, Aston CE, Austin P, et al: Prospective assessment of cosmesis before and after genital surgery. J Pediatr Urol 13:28.e1–28.e6 (2017).
Nermoen I, Husebye ES, Svartberg J, Løvås K: Subjective health status in men and women with congenital adrenal hyperplasia: a population-based survey in Norway. Eur J Endocrinol 3:453–459 (2010).
Ono M, Harley VR: Disorders of sex development: new genes, new concepts. Nat Rev Endocrinol 2:79–91 (2013).
Pasterski V, Hindmarsh P, Geffner M, Brook C, Brain C, Hines M: Increased aggression and activity level in 3- to 11-year-old girls with congenital adrenal hyperplasia (CAH). Horm Behav 3:368–374 (2007).
Pasterski V, Prentice P, Hughes IA: Consequences of the Chicago consensus on disorders of sex development (DSD): current practices in Europe. Arch Dis Child 8:618–623 (2010a).
Pasterski V, Prentice P, Hughes IA: Impact of the consensus statement and the new DSD classification system. Best Pract Res Clin Endocrinol Metab 2:187–195 (2010b).
Pasterski V, Geffner ME, Brain C, Hindmarsh P, Brook C, Hines M: Prenatal hormones and childhood sex segregation: playmate and play style preferences in girls with congenital adrenal hyperplasia. Horm Behav 4:549–555 (2011).
Pasterski V, Mastroyannopoulou K, Wright D, Zucker KJ, Hughes IA: Predictors of posttraumatic stress in parents of children diagnosed with a disorder of sex development. Arch Sex Behav 2:369–375 (2014).
Pasterski V, Zucker KJ, Hindmarsh PC, Hughes IA, Acerini C, et al: Increased cross-gender identification independent of gender role behavior in girls with congenital adrenal hyperplasia: results from a standardized assessment of 4-to 11-year-old children. Arch Sex Behav 5:1363–1375 (2015).
Petrie TA, Tripp MM, Harvey P: Factorial and construct validity of the body parts satisfaction scale-revised: an examination of minority and nonminority women. Psychology of Women Quarterly 3:213–221 (2002).
Poomthavorn P, Stargatt R, Zacharin M: Psychosexual and psychosocial functions of anorchid young adults. J Clin Endocrinol Metab 7: 2502–2505 (2009).
Rieger G, Linsenmeier JA, Gygax L, Bailey JM: Sexual orientation and childhood gender nonconformity: evidence from home videos. Dev Psychol 1:46 (2008).
Roen K, Pasterski V: Psychological research and intersex/DSD: recent developments and future directions. Psychol Sex 1:102–116 (2014).
Ros C, Alobid I, Balasch J, Mullol J, Castelo-Branco C: Turner's syndrome and other forms of congenital hypogonadism impair quality of life and sexual function. Am J Obstet Gynecol 208:484 (2013).
Rosen CB, Heiman J, Leiblum S, Meston C, Shabsigh R, et al: The female sexual function index (FSFI): a multidimensional self-report instrument for the assessment of female sexual function. J Sex Marital Ther 2:191–208 (2000).
Rosen RC, Catania J, Pollack L, Althof S, O'Leary M, Seftel AD: Male sexual health questionnaire (MSHQ): scale development and psychometric validation. Urology 4:777–782 (2004).
Rosenberg M: Society and the Adolescent Self-Image (Princeton University Press Princeton 1965).
Sanders C, Edwards Z, Keegan K: Exploring stakeholder experiences of interprofessional teamwork in sex development outpatient clinics. J Interprof Care 31:376–385 (2017).
Schober J, Nordenström A, Hoebeke P, Lee P, Houk C, et al: Disorders of sex development: summaries of long-term outcome studies. J Pediatr Urol 6:616–623 (2012).
Scholte RH, De Bruyn EE: The revised junior Eysenck personality questionnaire (JEPQ-R): Dutch replications of the full-length, short, and abbreviated forms. Personal Individ Differ 4:615–625 (2001).
Schützmann K, Brinkmann L, Schacht M, Richter-Appelt H: Psychological distress, self-harming behavior, and suicidal tendencies in adults with disorders of sex development. Arch Sex Behav 38:16–33 (2009).
Schweizer K, Brunner F, Gedrose B, Handford C, Richter-Appelt H: Coping with diverse sex development: treatment experiences and psychosocial support during childhood and adolescence and adult well-being. J Pediatr Psychol 42:504–519 (2016).
Snell WE, Fisher TD, Miller RS: Development of the sexual awareness questionnaire: components, reliability, and validity. Ann Sex Res 1: 65–92 (1991).
Speiser PW, Azziz R, Baskin LS, Ghizzoni L, Hensle TW, et al: Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: an Endocrine Society clinical practice guideline. J Clin Endocrinol Metab 95:4133–4160 (2010).
Spielberger CD: State-Trait Anxiety Inventory (Wiley Online Library 2010).
Strandqvist A, Falhammar H, Lichtenstein P, Hirschberg A, Wedell A, et al: Suboptimal psychosocial outcomes in patients with congenital adrenal hyperplasia: epidemiological studies in a nonbiased national cohort in Sweden. J Clin Endocrinol Metab 4:1425–1432 (2014).
Suorsa KI, Mullins AJ, Tackett AP, Reyes KJS, Austin P, et al: Characterizing early psychosocial functioning of parents of children with moderate to severe genital ambiguity due to disorders of sex development. J Urol 6:1737–1742 (2015).
Tiryaki S, Tekin A, Yağmur İ, Özen S, Özbaran B, et al: Parental view on the terminology of disorders of sex development. J Clin Res Pediatr Endocrinol 9:1 (2017).
Wolfe-Christensen C, Fedele DA, Kirk K, Phillips TM, Mazur T, et al: Degree of external genital malformation at birth in children with a disorder of sex development and subsequent caregiver distress. J Urol 4:1596–1600 (2012).
Zimet GD, Powell SS, Farley GK, Werkman S, Berkoff KA: Psychometric characteristics of the multidimensional scale of perceived social support. J Pers Assess 55:610–617 (1990).
Zucker KJ, Bradley SJ, Sullivan CBL, Kuksis M, Birkenfeld-Adams A, Mitchell JN: A gender identity interview for children. J Pers Assess 61:443–456 (1993).
Zucker KJ, Mitchell JN, Bradley SJ, Tkachuk J, Cantor JM, Allin SM: The recalled childhood gender identity/gender role questionnaire: psychometric properties. Sex Roles 54:469–483 (2006).