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Interdisciplinary Care in Disorders/Differences of Sex Development (DSD): The Psychosocial Component of the DSD – Translational Research Network

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Abstract

Scientific discovery and clinical management strategies for Disorders/Differences of Sex Development (DSD) have advanced in recent years. The 2006 Consensus Statement on Management of Intersex Disorders stated that a mental health component to care is integral to promote positive adaptation, yet the parameters of this element have not been described. The objective of this paper is three-fold: to describe the psychosocial screening protocol adopted by the clinical centers of the DSD-Translational Research Network; to summarize psychosocial data collected at one of the 10 network sites; and to suggest how systematic behavioral health screenings can be employed to tailor care in DSD that results in better health and quality of life outcomes. Steps taken in developing the largely “noncategorical” screening protocol are described. These preliminary findings suggest that DSD, as one category of pediatric chronic conditions, is not associated with marked disturbances of psychosocial adaptation, either for the family or the child; however, screening frequently uncovered “risk factors” for individual families or patients that can potentially be addressed in the context of ongoing clinical care.

Administration of the DSD-TRN psychosocial screening protocol was demonstrated to be feasible in the context of interdisciplinary team care and was acceptable to families on a longitudinal basis. The ultimate value of systematic screening will be demonstrated through a tailoring of psychosocial, medical and surgical services, based on this information that enhances the quality of patient and family-centered care and subsequent outcomes.

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Keywords

disorders of sex development; intersex; psychosocial factors; psychosocial assessment; registry; standardization; case management

INTRODUCTION

Disorders (or Differences) of Sex Development (DSD) are defined as “congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical” [Lee et al., 2006] and are subcategorized as Sex Chromosome DSD, 46,XY DSD, and 46,XX DSD. Conditions subsumed under the DSD umbrella are diverse; they can, but do not necessarily imply ambiguity in genital appearance or uncertainty about gender of rearing [Achermann and Hughes 2011; Ahmed et al., 2016; Lee et al., 2006], yet all are thought to be associated with significant psychosocial burdens [Sandberg et al., 2012].

The majority of children born with DSD are identified in the newborn period due to visible genital differences and/or health concerns (e.g., salt-wasting crises in classic congenital adrenal hyperplasia, CAH); however, detection can occur later when expected features of pubertal development (e.g., menstruation in girls) do not progress on time or include changes characteristic of the opposite sex (gynecomastia in boys or hirsutism in girls). In adulthood, DSD may first present as fertility problems [Van Batavia and Kolon 2016].

Aspects of clinical management in DSD have been in a state of flux, triggered in large part by reports of adults having experienced dissatisfying outcomes [Chase 1998; Feder 2014; Karkazis 2008; Preves 2003]. Features of DSD and associated clinical interventions are thought to represent significant risk factors for parental emotional distress and maladaptive psychosocial and psychosexual development of the affected child. Parents have reported levels of post-traumatic stress syndrome comparable to those reported by caregivers of children diagnosed with cancer [Pasterski et al., 2014]. The perceived need for psychosocial services reported by parents of children with a DSD is high and unrelated to genital appearance [Bennecke et al., 2015]. The literature on the influences of the parent-child relationship on the child’s development of emotional and behavioral self-regulation is rich with examples of how parental emotional states and parenting styles can profoundly influence the child’s developmental trajectory [Grusec and Hastings 2015; Thompson 2015]. Similarly, it is known that a chronic medical condition in childhood and adolescence can exert negative influences on the child’s emotional and psychosocial development, in particular in the area of peer relations, if countervailing buffers – such as parenting styles that promote positive adaptation – are not in place [Crnic and Neece 2015].

Stressors that come to fore when a child is found to have a DSD vary and can present challenges to shared decision-making with healthcare providers because of the understandable tendency to want to quickly arrive at decisions believed to deliver remedy and relief from a stressful situation [Wisniewski and Sandberg 2015]. Examples of stressors faced by parents of children with DSD, and when older, patients themselves, include: the presence and degree of atypical genitalia, decisions regarding gender of rearing (and the possibility that gender reassignment may follow), discordance between sex chromosomes

and gender of rearing, ongoing controversies regarding the risks and benefits of early genital surgery, and anticipated stigma and accompanying shame and secrecy [Boyle et al., 2005; Creighton et al., 2001; Crouch et al., 2004; Dreger 1998; Sandberg and Mazur 2014].

Improved access to psychosocial services within and outside the medical setting has been proposed since the mid-1990s to help affected individuals and families manage ongoing challenges and enhance psychosocial adaptation and well-being [Dreger 1999]. The 2006 Consensus Statement on Management of Intersex (hereafter referred to as the Consensus Statement) and related guideline documents [Ahmed et al., 2016; Consortium on the Management of Disorders of Sex Development 2006; Lee et al., 2016] also called for an integrated, interdisciplinary healthcare team that includes qualified mental health providers who can help families understand and address early emotional reactions, explore present and future worries, adjust to the period of uncertainty during the diagnostic process, and facilitate the process of shared decision-making for themselves or their child. These efforts are in line with: (i) the definition of health not merely being an absence of disease or a reduced mortality and morbidity, but also a “state of mental and well-being” [World Health Organization 1946]; and (ii) the notion that good outcomes transcend strictly medical or surgical ones and include achieving positive psychological and social adaptation for the patient and family [Lee et al., 2006; Lee et al., 2016].

Despite the assumed benefits of involving behavioral health specialists and a significant proportion of parents indicating a high need for psychological support [Bennecke et al., 2015], there is a clear mismatch between these factors and availability of behavioral health service and their integration within the model of care. A recent international survey of DSD clinical services showed that only a minority of centers (41%) included a mental health provider [Kyriakou et al., 2016]. An earlier European survey indicated that child psychologists or child psychiatrists were regularly available to deliver psychological services at only 17% and 11% of centers, respectively [Pasterski et al., 2010].

The importance of a behavioral health component in the management of DSD is not about pathologizing individuals affected by DSD; rather, it stems from recognizing that clinical decisions are frequently driven by considerations of promoting positive psychological adaptation and well-being instead of exclusively addressing a specific medical health need. In comparison to knowledge about hormonal factors involved in the process of psychosexual differentiation (i.e., gender identity, gender role and sexual orientation) in DSD, we face major gaps in our understanding of other factors likely contributing to specific and global adaptation in people with DSD [Stout et al., 2010]. This literature has generally neglected theory or research strategies derived from developmental psychology or pediatric psychology that considers the interaction of biological and environmental factors on child developmental trajectories [Pickles and Hill 2006; Rose et al., 2004; Wallander et al., 2003].

A number of potential barriers coalesce to reduce the likelihood that expert psychological services are available to patients and families in the context of interdisciplinary team care. Provider expertise in DSD is typically located only at select academic hospitals: geographic distance and health insurance coverage restrictions (especially in the US) can limit access and contribute to fragmented care. An additional obstacle presents as a lack of accepted

protocols for routine and longitudinal assessment of patient and family psychosocial adaptation. In contrast to the assessment of gender, for which protocols for DSD have been suggested [Meyer-Bahlburg 2011], there are no suggestions for routine surveillance of other key domains of patient and family adaptation. Such a protocol would assess risk and resilience factors within the family as well as establish a schedule for evaluating the developmental, behavioral/emotional, social, and educational status of the child in the context of ongoing care delivered by the DSD team. Child and adolescent assessments would also encompass self-perceptions of domain-specific competencies or adequacies, including body image, and a multidimensional assessment of the child's gender identity.

Because the discrete syndromes falling under the DSD umbrella are individually rare, research into the psychosocial sequelae of these conditions is hampered by the limited numbers of patients treated at any individual site. Multi-site treatment networks and patient registries have been shown to accelerate the path to discovery and improved healthcare (for example, as in the case of cystic fibrosis [Stevens and Marshall 2014]) and the same strategy can be extended to the psychosocial aspects of the medical condition provided that a standardized assessment protocol is in place. At present, four major international DSD initiatives are underway (for an overview, see [Sandberg et al., 2015]). The U.S. NIH-sponsored DSD Translational Research Network (DSD-TRN) (https://dsdtrn.genetics.ucla.edu/) seeks to improve clinical care through: (i) standardizing clinical management practices – including psychosocial assessment; and (ii) collecting systematic data on patient characteristics, interventions and outcomes to inform emerging best practices.

The objective of this paper is to describe the standardized psychosocial screening protocol adopted by DSD-TRN clinical sites and to present data on patient and family psychosocial adaptation and needs at one site.

METHODS

Measures

Noncategorical framework—Progress in developing more effective medical treatments for specific pediatric conditions has driven a disease-specific ("categorical") approach to care [Stein 2011]. In the case of DSD, this approach has contributed to a better understanding of genetic factors involved and progress in genital surgical techniques. The categorical approach applied to DSD has also been associated with an emphasis on the process of psychosexual differentiation (i.e., the development of gender identity, gender role, and sexual orientation and behavior) [Hines 2011; Meyer-Bahlburg 2008]. This focus follows from research – largely in non-human animals – elucidating the influence of sex hormones, during steroid-sensitive periods of brain development, on the expression of reproductive and non-reproductive sex-dimorphic behaviors [Hines 2011; Stout et al., 2010].

A "noncategorical" approach factors both the specific aspects of the medical condition alongside the total life experience of the child, family, and broader cultural and social context. This perspective guides theory development and clinical care by taking into account a growing body of evidence that successful developmental trajectories of persons with

chronic medical conditions are influenced as much by the psychosocial environment, support resources, and organization of healthcare delivery as by the specific nature of the person's medical condition [Sandberg and Mazur 2014; Stein 2011; Stein and Jessop 1982]. This framework holds the potential to systematically account for variability in psychosocial outcomes and the translation of clinical interventions, proven effective in the treatment of other chronic conditions, to the challenges faced by those affected by DSD.

Measure selection and administration—The process of developing a standardized psychosocial assessment protocol was led by pediatric behavioral health specialists and site clinical coordinators comprising the DSD-TRN Psychosocial Workgroup, in partnership with representatives from DSD patient advocacy and support organizations (the DSD-TRN Advocacy Advisory Network) and Accord Alliance, a nonprofit convener of stakeholders interested in promoting comprehensive and integrated approaches to care in DSD. Because there are no evidence-based guidelines for the psychological/psychosocial evaluation of DSD-affected patients and families, this working group started with a blank slate except for adopting a noncategorical framework.

The initial goal was to outline the parameters of a psychosocial evaluation of the patient and family, including recommendations for the timing of longitudinal evaluations. The purpose of the assessment is to proactively monitor patient and family psychosocial adaptation (as opposed to the more common model of mental health consultation upon request of medical specialists detecting problems) and therapeutic intervention, as indicated [Drotar 1995]. In addition to the clinical interview at the time of scheduled clinic visits, questionnaires were selected as the means of systematically screening domains of individual and family adaptation that allow for a developmentally-sensitive approach to psychosocial assessment and ongoing monitoring of understanding and adaptation of affected individuals and family members. Instruments were selected only if there was an expectation that they could deliver immediately actionable information to DSD team providers at the clinical sites about the individual patient and family. Selected parent/caregiver- and patient-report questionnaires focus on five domains: (i) family resiliency, risk, and social support (Psychosocial Assessment Tool, PAT [Kazak et al., 2015; Pai et al., 2008]; Patient Health Questionnaire-4, PHQ-4, a measure of depression and anxiety [Kroenke et al., 2009]; Support and Resources Assessment [Sandberg and Gardner 2011c]); (ii) patient and family understanding of the medical condition (Knowledge of Condition [Sandberg and Gardner 2011a; Sandberg and Gardner 2011b]); (iii) parent-proxy and patient self-report of social competencies and behavioral/emotional problems (Child Behavior Checklist and Youth Self-Report; [Achenbach 2009]); (iv) patient self-concept (Self-Perception Profile [Harter 1985; Harter 1988]; Body Image Scale adapted from [Lindgren and Pauly 1975]), and (v) patient gender development (Multidimensional Gender Identity Scale [Egan and Perry 2001]) (see Table 1).

The questionnaires administered to patients and families at regular intervals, coupled with face-to-face clinical interviews, provide the clinician with the capacity to tailor any intervention to the needs of the person and their family. Scoring of these questionnaires can be used to triage services to patients/families requiring a higher intensity of services and delivering routine psychoeducational counseling to patients/families showing positive

adaptive coping. Interventions extend beyond DSD-specific psychoeducational counseling to include all the issues encountered by families with a child with special healthcare needs.

The questionnaires comprising the psychosocial screening protocol are integrated into usual care at DSD-TRN sites, although fidelity to the protocol varies across sites. Because there is no precedent for conducting routine and standardized evaluations at these sites, considerable effort has been directed toward creating a process experienced by patients, families and their healthcare providers as patient and family-centered. During the initial contact with the DSD team, the behavioral health provider – or, in the absence of such a provider, the team coordinator or member designated to focus on psychosocial issues – explains to the family the holistic approach followed, which emphasizes positive psychosocial adaptation. Communicating to the family that the psychosocial component is integral to the model of care is a responsibility of the entire interdisciplinary DSD team [Lee et al., 2016; Sandberg and Mazur 2014]. The behavioral health provider further explains that the psychosocial component of care is delivered by educating the parents and child about the potential implications of the medical condition for daily functioning (i.e., anticipatory guidance). The objectives of the psychosocial component of care are achieved through iterative evaluations and counseling.

Data Analysis Plan

Implementation of the standardized psychosocial assessment protocol remains fairly new for many of the 10 member sites in the DSD-TRN. For this reason, data from one of the original sites that incorporated the full protocol provide the basis for the current data analysis. The standardized schedule for administration calls for repeated assessments at either one- or two-year intervals; however, to serve as a benchmark, data from only the first administration of any measure are included in the current analyses. Descriptive statistics are provided for all quantitative measures. As questionnaires are administered at different ages (Table 1), patient age at completion is reported separately for each measure. Because of the qualitative nature of the information collected using the Knowledge of Condition questionnaire, those data are not summarized here.

RESULTS

One hundred one parents/caregivers (n = 49 biological mothers, 35 biological fathers, 5 adoptive mothers, 4 adoptive fathers, 1 stepmother, 3 legal guardians, and 4 other caregivers) of 64 consecutively-referred patients, aged newborn through 18 years, completed at least one component of the standardized psychosocial assessment. No family refused to complete the assessment although some took forms home to complete, but failed to return them by mail. The time required to complete protocol ranged between 10 to 35 minutes (depending upon the specific questionnaires completed at a particular visit). Patient diagnoses fell into all three diagnostic categories outlined in the Consensus Statement: 8 (12.5%) with sex chromosome DSD, 38 (59.4%) with a 46,XY DSD, and 18 (28.1%) with a 46,XX DSD (Table 2).

Family and Caregiver Risk and Resilience

Psychosocial Assessment Tool (PAT)—The PAT was administered to 64 parents and caregivers to complete on behalf of the patient and their family. On average, patients were 8.0 ± 6.1 years old. Results showed variability in total PAT score (range = 0 – 3.3), with the mean score (1.1 ± 0.7) falling in the "targeted" range; i.e., reporting acute distress with risk factors present (Table 3). Approximately half (n = 31, 48.4%) of families fell in the universal (i.e., reporting many supportive resources and relatively low psychosocial risk) range of risk for psychosocial problems, 26 (40.6%) fell in the targeted range, and 7 (n=10.9%) fell in the clinical range (i.e., reporting few supportive resources and multiple areas of difficulty; persistent and/or escalating distress). Mean subscale scores also demonstrated variability with the majority (67.2% to 100%) of scores not rising to the threshold (i.e., 0.5) at which a targeted intervention would be recommended to address difficulties.

Patient Health Questionnaire-4 (PHQ-4)—The PHQ-4 was administered to 92 parents and caregivers of 53 patients aged newborn through 18 years (mean = 8.4 ± 5.8 yrs). Parental responses represented the full range of possible Total (mean = 1.6 ± 2.4), Depression (mean = 0.6 ± 1.2) and Anxiety (mean = 1.0 ± 1.3) scale scores (Table 3). The majority of caregivers scored in the non-clinical range for the total and subscale scores.

Support and Resources Assessment (SRA)—The SRA was administered to 101 parents of 61 patients aged newborn to 18 years (mean = 8.1 ± 5.9 yrs). With regard to sharing information about their child's medical condition with others, responses ranged from no parents reporting they would withhold information from their spouse/partner to a maximum of 38 of 76 (50%) parents reporting they would not inform their coworkers (Table 4). With regard to their plans about seeking additional information and/or support, parents' responses varied from none reporting that they would not seek out information or support from their child's doctors to a maximum of 53 of 98 (54.1%) reporting they would not want to seek out information or support from friends.

Patient Psychosocial Adaptation

Self-Perception Profile (SPP)—The SPP includes two versions, one for children (SPP-ch) and one for adolescents (SPP-ad). The SPP-ad includes three scales that do not appear in the SPP-ch: Job Competence, Close Friendships and Romantic Appeal. The mean age of patients administered the SPP-ch was 10.9 (±1.5) years and the SPP-ad was 15.6 (±1.5) years. Reported mean Global Self-Worth and domain-specific competency scores fell in the "positive" range (i.e., ≥ 2.5; the respondent indicates more positive than negative self-statements) with one exception, Romantic Appeal, which fell in the "mildly low" (2.0 to 2.4) range (Table 5). The most highly rated competencies/qualities were Behavioral Conduct, Social Acceptance, and Job Competence.

Body Image Survey (BIS)—The BIS, comprising 26 items focusing on discrete body parts, was administered to 24 patients, beginning at age 10 for girls and age 12 for boys (mean 14.2 ± 2.3 yrs). Each item is rated on a 5-point scale (1 = "very happy," 3 = "neutral," 5 = "very unhappy") and the items are grouped into 3 subscales: Primary Sex Characteristics (e.g., private parts, facial hair, body hair, and breasts/chest); Secondary Sex-Related

Characteristics (e.g., hips, figure/body shape, muscles); and Hormonally Unresponsive Characteristics (e.g., nose, shoulders, chin, hands, eyebrows). The median subscale scores for the sample indicated the least happiness for Primary Sex Characteristics (2.17 where 2 = "happy" and 3 = "neutral"). The range of responses was also the largest for Primary Sex Characteristics (1.0 to 4.5) in comparison to the other scales (1.0 to 3.0) (Table 6).

At the level of individual items, patients rated being happiest (i.e., rated as either "very happy" or "happy") with their "face," "calves," and "nose" (n = 21, 91.3%) and unhappiest (i.e., rated as either "neutral," "unhappy," or "very unhappy") with their "private parts" (n = 10; 41.7%), "breasts" (n = 8, 33.3%), and "voice" (n = 7, 29.2%). If the patient reported feeling "neutral" (3) to "very unhappy" (5) with a particular body characteristic, they were asked whether or not they would choose to medically or surgically change it. With regard to "private parts," of the 10 patients falling into this group, 5 wished for appearance-altering interventions, 3 were not in favor of intervention, and 2 left this follow-up question blank. Of the 5 requesting appearance change to their genitals, 4 were 46,XY: 2 were reared and living as girls and are requesting additional feminizing surgery; 1 was originally gender assigned as a girl, but with gender dysphoria developing in adolescence, is requesting masculinizing surgery; and 1 was reared as a girl, but a decision was made at age 4 to reassign the child as a boy and the now pre-adolescent boy desires masculinizing surgery. The fifth case is a girl with CAH requesting feminizing clitoroplasty.

Multidimensional Gender Identity Scale (MGIS)—The self-reported MGIS comprises three scales: Gender Typicality, Gender Contentedness and Felt Pressure (to conform to gender-stereotypic behavior). Scale scores represent the mean of items rated on a 4-point scale. Higher scores on Gender Typicality and Gender Contentedness reflect perceptions of higher gender typicality and contentedness, respectively, whereas higher scores on the Felt Pressure scale reflects patient perceptions of greater felt pressure for gender conformity from adults or peers. The MGIS was administered to 25 patients aged 8 to 18 years (13.3 ± 2.9 yrs). Mean scores on the Gender Typicality, Gender Contentedness, and Felt Pressure scales were 2.4 ±0.7, 3.6 ±0.6, and 1.7 ±0.6, respectively. Scores above the scale mid-point (2.5) were interpreted as reflecting higher gender typicality and contentedness, whereas mean scores greater than 1.5 on the felt pressure scale was considered worthy of further clinical discussion. Applying these cutoffs, fewer than half (n = 11, 44.0%) reported behaving in a manner typical of their gender, yet a strong majority (n=24, 96.0%) reported feeling content with their gender assignment. Finally, the majority also reported feeling varying degrees of pressure (i.e., > 1.5 on 4-point scale) to behave in a manner consistent with their gender of rearing (n = 16, 64.0%).

Child Behavior Checklist (CBCL) and Youth Self Report (YSR)—The CBCL was administered to 70 parents of patients 1.5 to 18 years old (mean = 9.3 ± 4.7 yrs). The YSR was administered to 24 patients 11 to 18 years old (mean = 13.5 ± 2.4 yrs). On average, both parent- and patient-reported scores on the social competency and behavior problem scales fell in the "non-clinical" range; however, individual patients' scale scores fell in the "borderline clinical" or "clinical" ranges (Table 7).

DISCUSSION

Regular screenings for developmental or emotional/behavioral problems in the general pediatric-aged population are increasingly called for by national and international health organizations [Bright Futures/American Academy of Pediatrics 2016; World Health Organization Europe 2008]. The benefits of regular psychosocial screenings for assessment and the tailoring of clinical care based on findings are obviously no less important in the case of children with chronic medical conditions, including those born with DSD. Accordingly, the DSD-TRN developed a standardized psychosocial screening protocol for patients and families attending member clinics. The measures comprising the screening protocol were selected because of their collective breadth of coverage, strong psychometric properties, and/or expectation of their sensitivity to issues of particular relevance to patients born with DSD and their caregivers. The screening questionnaires augment the clinical interview by systematically probing for parent and self-reported details of adaptation in several domains. Our experience to date has demonstrated the acceptability to families of these screenings and their feasibility for integration within the context of an interdisciplinary DSD clinic.

These psychosocial data, from consecutively-referred patients and families seen at one member site of the DSD-TRN, point toward a number of cross-cutting (i.e., noncategorical) as well as DSD-specific (i.e., categorical) aspects of family and patient psychosocial adaptation. For example, although caregiver PHQ-4 anxiety and depression symptom scores for the combined group fell below the clinical cutoff, the scores for 16%, 5% and 2% of caregivers fell in the “mild,” “moderate,” and “severe” symptom ranges, respectively. Emotional distress is one factor recognized to bias healthcare decision making [Lipstein et al., 2012]. Clearly, knowledge of parents’ emotional state at the time of making irreversible decisions (e.g., removal of the gonads or genital surgery) could be very useful to providers invested in shared decision-making with parents of infants or young children [Siminoff and Sandberg 2015; Wisniewski and Sandberg 2015]. Similarly, an extensive scientific literature attests to the value of social support in maintaining and enhancing psychological well-being, both directly and as a stress buffer [Lakey and Orehek 2011; Thoits 2011]. Accordingly, the SRA finding that a sizable minority of parents in our cohort have not (and will not) share information about their child with relatives (27%) or close friends (20%) is potentially worrisome.

These findings are in line with other studies suggesting that anxiety and depression symptoms decrease with time after a chronic condition diagnosis [Wolfe-Christensen et al., 2017], but can potentially remain elevated for a prolonged period in a subset of parents choosing not to seek support from relatives or close friends in order to withhold details of their child’s medical condition. Although the decision to withhold information from sources of support can be motivated by a desire to protect the child’s privacy and to prevent stigmatization, it may be done at a cost to parents of poorer psychosocial adaptation [Crissman et al., 2011].

Use of the SRA also revealed that the only source of additional information and support universally sought by parents in our cohort was their child’s doctors. This observation

should serve as a reminder to DSD healthcare providers to encourage and refer families to local and national support groups or arrange for tailored, individualized peer support. Peer support aims to enhance knowledge and confidence, reduce stress and distress, and promote the reciprocal sharing of experiences [Baratz et al., 2014]. Overcoming barriers to engagement with peer support (originating with either providers or caregivers) should be considered a high priority insofar as the benefits are non-overlapping with professional services and are considered a component of comprehensive and integrated interdisciplinary patient- and family-centered care [Baratz et al., 2014; Lee et al., 2006; Lee et al., 2016].

As observed from scores on the SPP, the children and adolescents with a variety of DSD report a wide range of self-perceptions regarding domain-specific competencies and qualities. Although mean scale scores indicated that these patients, in general, were making slightly more positive than negative self-statements, a significant minority rated themselves more negatively (i.e., < 2.0 on the 4-point scale) regarding Scholastic Competence (15%), Social Acceptance (12%), Athletic Competence (29%) and (for those 13 years or older) Romantic Appeal (18%).

Continuing with measures assessing generic (noncategorical) aspects of psychosocial adaptation, parent reports of their children's social competencies and behavior problems using the CBCL indicated the majority fell into the nonclinical range for instrument norms; yet a substantial minority received scores in the borderline or clinical range. It is noteworthy that the youths' parallel self-reports on the YSR reflected a more positive appraisal of psychosocial adaptation than that of their parents. There are multiple ways of interpreting cross-informant agreement and disagreement, but there is a consensus that each source of information (parent, child, teacher, or others) provides unique and meaningful variance to our understanding of the child's adaptation.

As noted earlier, a noncategorical approach to psychosocial assessment does not exclude examining the effects of specific features of the person's medical condition, for example, sex-typical body features. The SPP includes a scale labeled Physical Appearance, but the wording of items does not specify body parts affected in some DSD, for example, genitalia or breasts. For this reason, the psychosocial protocol included the BIS. This measure revealed that some patients were unhappy with physical characteristics associated with their DSD which, in the context of ongoing clinical care, can trigger a discussion between providers and patients. Clinically, it is easier to have a discussion with a pre-teen or teen about a sensitive topic on which they have already expressed their thoughts and wishes for change through the questionnaire.

Scores on the MGIS for the total sample suggests self-perceptions of moderate gender typicality associated with overwhelming gender contentedness, i.e., behaving in a manner typical (or atypical) of one's gender is not synonymous with the degree of contentedness with one's gender. As noted in the Consensus Statement, atypical gender-role behavior in an individual with a DSD is not an indication of incorrect gender assignment. Nevertheless, we found that the majority of patients (64.0%) reported feeling some degree of pressure to behave in a manner consistent with their gender of rearing. In a recent mixed methods survey employing a convenience sample of 272 people (16–87 yrs) with "intersex

variations" largely (approximately 80% from Australia), 44% reported receiving "counselling/training/pressure from institutional practitioners (doctors, psychologists etc.) on gendered behaviour; and 43% from parents" [Jones et al., 2016].

The MGIS findings regarding gender development in youth with DSD should not be considered more than very preliminary because of the small sample derived from a single site, yet they raise the same questions of interpretation as the scores on the other measures in this screening protocol: how different or similar are these scores from the population of youth without a DSD? And do the same scores need to be interpreted differently for different populations? Comparison of the MGIS scale scores for the current sample appear comparable to those obtained from a public school sample [Egan and Perry 2001; Yunger et al., 2004]. But do comparable scores have the same meaning in the two populations? Consider the following examples from the Gender Typicality (GT) and Gender Contentedness (GC) scales (from the girls' version of the form):

GT	Some girls feel they are <u>different</u> from other boys.		**BUT**	Other girls feel they are <u>similar</u> to other boys.	
	Very true for me	Sort of true for me		Sort of true for me	Very true for me

GC	Some girls are happy that they were born a girl.		**BUT**	Other girls are not happy they were born a girl.	
	Very true for me	Sort of true for me		Sort of true for me	Very true for me

It is easy to appreciate how responses to individual items, such as these, might provoke different questions depending upon whether the form is being completed by a child with a DSD versus a child from the general population born physically typical. The challenge in the DSD Clinic is to utilize these screening measures as a means to flexibly guide discussions with the patient and family, while simultaneously taking into account that degrees of gender atypicality, gender discontent and felt pressure for gender conformity are normative phenomena [Perry and Pauletti 2011; Sandberg and Meyer-Bahlburg 1994; Sandberg et al., 1993; Yunger et al., 2004].

Overall, the scale scores on the various questionnaires comprising the psychosocial screening battery presented here do not suggest that this group (of parents and their children) is necessarily exhibiting more problems of psychosocial adaptation, self-image/concept, or gender development, than families or children unaffected by a DSD. Although a proportion of the sample (depending upon measure) received scores falling in the range labeled as clinical or otherwise worthy of further investigation, this would be the finding in any sample given what is known about the statistical distribution of psychological traits [Pickles and Hill 2006; Rescorla et al., 2012]. Moreover, the proximal "cause" of extreme (low or high) scores in individual cases cannot be discerned with any certainty despite what may appear to be obvious connections, e.g., unhappiness with genital appearance in a child with a DSD. Despite these limitations, such screening tools – qualified by questionnaire sensitivity and specificity – are useful in identifying individual patients or families either at risk for or already exhibiting problems of psychosocial adaptation. All families receiving care for the

child's DSD will profit from enhanced knowledge and understanding of the condition and its implications, and those families with problems predating or those exhibiting difficulties coinciding with DSD ascertainment, will require additional tailored services regardless of whether the group, as a whole, function any differently from those in the general population.

Gaps and next steps

Systematic and regular screening of psychosocial adaptation in comprehensive interdisciplinary healthcare team settings is a new and growing area for research with multiple challenges (Table 8). Important questions for future research include: what is the relationship between screening, access to and differential pathways of care based on findings from psychosocial questionnaires and clinical interviews? What is the optimal interval for screening and should the interval vary based on domain assessed? How should surveillance of psychosocial adaptation be integrated within the delivery of DSD care more generally? For example, it is common that the interval between clinic visits will be relatively short following initial detection and diagnostic workup of the DSD. In the case of CAH, the interval between visits (commonly 3 months; [Speiser et al., 2010]) provides ample opportunity to embed a psychosocial component into the model of care. By contrast, a child diagnosed with a DSD not requiring regular treatment for metabolic control, and who is beyond the period of monitoring the outcome of early surgery, if indicated, may not be brought back to clinic until the issue of pubertal onset and progression become the next focus of medical care. This practice could result in the family not being seen at all by members of the DSD team between the ages of 5 to 10 years (i.e., middle childhood), a period of substantial developmental importance, in particular with respect to peer relations [Bukowski et al., 2015].

Conclusions

The preliminary data summarized in this report attest to the feasibility and acceptability of incorporating a systematic psychosocial screening protocol in the context of ongoing interdisciplinary care for DSD. Scale scores on the various measures did not exhibit either ceiling or floor effects in this sample making them suitable for differentiating the adaptation of individuals within this population. The use of psychosocial screening tools is not intended to replace in-depth interviews or be understood as a substitute for evidence-based interventions, but is the first step in a process of behavioral healthcare delivery.

The clinical care (and research) agenda in DSD needs to be broadened, in part to respond to justifiable criticism by former patients and advocates that poor psychosocial outcomes may be less a function of atypical prenatal sex hormone exposure or genital surgical procedures than the consequence of a complex interaction among multiple experiences (e.g., medical decision making, experience of DSD as stigmatizing and associated with feelings of secrecy and shame, parental mental health or marital problems that predate the child's birth, financial hardships attendant with a chronic medical condition, etc.) that modulate the outcomes within and across developmental stages [Wisniewski and Sandberg 2015].

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Table 1

Administration schedule for standardized psychosocial assessment.

Measure	Assesses	Focus	Informant	Administration Schedule		
				Patient Age (yrs)	Initial	Follow-Up
Psychosocial Assessment Tool[a]	Psychosocial risk factors: Family Structure & Resources; Family Social Support; Family Problems; Parent Stress Reactions; Family Beliefs; Child Problems; Sibling Problems	Family	Parent	All	First visit	Annual
Support and Resource Assessment[b]	Degree to which social support systems and educational resources are accessed	Parent	Parent	All	First visit	Annual
Patient Health Questionnaire-4[c]	Depression and anxiety screen	Parent	Parent	All	First visit	Annual
Knowledge of Condition[d]						
Caregiver Report	Knowledge of diagnosis and understanding of cause; parental impression of child's understanding	Parent Patient	Parent	All	≥6 mos after first visit	Annual
Self-Report		Patient	Patient	≥10		
Child Behavior Checklist[e]	Competencies: Activities, Social, School; Behavior problems: Internalizing, Externalizing, Total	Patient	Parent	1.5 – 18	First visit	Annual
Self-Perception Profile[f]	Global self-worth; Domain-specific self-concept (eg, social & job competence)	Patient	Patient	8 – 18	First visit	2 years
Body Image Scale[g]	Satisfaction with primary- and secondary- sex-related characteristics and hormonally unresponsive body features	Patient	Patient	Girls ≥10 Boys ≥12	≥6 mos after first visit	2 years
Multidimensional Gender Identity Scale[h]	Gender typicality; experience of gender identity discontent; pressure to conform to gender stereotypes	Patient	Patient	8 – 18	≥6 mos after first visit	2 years
Youth Self-Report[e]	Competencies: Activities, Social, School; Behavior problems: Internalizing, Externalizing, Total	Patient	Patient	11 – 18	First visit	Annual

[a] PAT [Kazak et al., 2015; Pai et al., 2008]

[b] SRA [Sandberg and Gardner 2011c]

[c] PHQ-4 [Kroenke et al., 2009]

[d] KOC-CR & KOC-SR[Sandberg and Gardner 2011a; Sandberg and Gardner 2011b]

[e] CBCL & YSR [Achenbach 2009]

[f] SPP [Harter 1985; Harter 1988]

[g] BIS, adapted from [Lindgren and Pauly 1975]

[h] MGIS [Egan and Perry 2001]

Table 2

Patient diagnoses.

Sex Chromosome DSD			
Reared as a Boy (n=3)	**n**	**Reared as a Girl (n=5)**	**n**
Klinefelter syndrome	1	Turner syndrome and variants	4
47,XYY with proximal hypospadias, penoscrotal transposition, and undescended testes	1	Mixed gonadal dysgenesis	1
Mixed gonadal dysgenesis	1		
46,XY DSD			
Reared as a Boy (n=18)	**n**	**Reared as a Girl** (n=20)	**n**
Pure gonadal dysgenesis	1	46,XY gonadal dysgenesis - *MAP3K1*	7
Partial androgen insensitivity syndrome	3	Complete androgen insensitivity syndrome	4
17β-hydroxysteroid dehydrogenase deficiency	1	17β-hydroxysteroid dehydrogenase deficiency	2
5α-reductase type 2 deficiency	1	5α-reductase type 2 deficiency	1
Cloacal/Bladder exstrophy	2	17α-hydroxylase/17,20-lyase deficiency	1
22q 11.2 deletion syndrome	1	WAGR 11p13 deletion syndrome	1
Persistent Müllerian duct syndrome	1	Atypical genitalia with unknown genetic etiology	4
Atypical genitalia of unknown genetic etiology	8		
46,XX DSD			
Reared as a Boy (n=1)	**n**	**Reared as a Girl** (n=17)	**n**
46,XX *SRY+*	1	CAH (11β-hydroxylase deficiency)	1
		CAH (21-hydroxylase deficiency)	3
		CAH, nos	8
		Cloacal anomalies	2
		VACTERL association	2
		Atypical genitalia of unknown genetic etiology	1
Total Reared as Boys	**22**	**Total Reared as Girls**	**42**

Table 3

Family and caregiver risk and resilience: PAT and PHQ-4.

Scale	n	Scale Scores			Risk Category* (%)		
		Mean	SD	Range			
PAT					**Universal**	**Targeted**	**Clinical**
Structure/Resources	64	0.15	0.18	0 – 0.75	89.1	10.9	n/a
Social Support	63	0.08	0.17	0 – 0.75	92.1	7.9	n/a
Patient Problems	64	0.32	0.26	0 – 0.87	67.2	32.8	n/a
Sibling Problems	40	0.15	0.17	0 – 0.67	95.0	5.0	n/a
Caregiver Problems	64	0.22	0.22	0 – 1.00	85.9	14.1	n/a
Stress Reaction	64	0.04	0.10	0 – 0.33	100	0	n/a
Caregiver Beliefs	64	0.17	0.17	0 – 0.57	93.8	6.3	n/a
Total PAT Score	64	1.08	0.71	0 – 3.29	48.4	40.6	10.9
PHQ-4					**Non-Clinical**	**Clinical**	
Depression	92	0.64	1.17	0 – 6	93.5	6.5	
Anxiety	92	0.95	1.34	0 – 6	88.0	12.0	
					None	**Mild**	**Moderate**	**Severe**
Total PHQ-4 Score	92	1.59	2.35	0 – 12	76.1	16.3	5.4	2.2

*Risk category designations differ between subscale and total scale scores; e.g., on the PAT, the “clinical” range is only calculated for the total score; not subscale scores

Table 4

Parental support and resources: Support and Resources Assessment (SRA).

	Endorsed item		
	N*	n	%
I have not & will not tell _____ about my child's condition:			
My coworkers	76	38	50.0
My child's daycare provider	29	13	44.8
My other relatives	93	25	26.9
My close friends	92	18	19.6
My other children	80	12	15.0
My spouse/partner's parents	82	7	8.5
My parents	94	5	5.3
My spouse/partner	97	0	0
I have not & will not seek out additional information and support by:			
Talking to my friends	98	53	54.1
Talking with adults who have a urogenital condition	95	38	40.0
Talking with other parents of children who have a similar condition	95	23	24.2
Going to a library or read printed material	99	22	22.2
Talking to my family	98	20	20.4
Visiting a support, resource, or advocacy organization's website	95	13	13.7
Talking to my child's doctors	101	0	0

* the N for responses to selected items varies due to applicability (e.g., family does not use daycare services) or missing data

Table 5

Patient self-concept: Self-Perception Profile (SPP).

Scale	n	Scale Scores			Category (%)		
		Mean	SD	Range	Positive*	Mildly Low	Low
Scholastic Competence	33	2.71	0.78	1.0 – 4.0	57.6	27.3	15.2
Social Acceptance	33	2.95	0.84	1.0 – 4.0	72.7	15.2	12.1
Athletic Competence	33	2.58	0.85	1.0 – 4.0	70.8	0	29.2
Physical Appearance	33	2.94	0.74	1.2 – 4.0	66.7	30.3	3.0
Job Competence	18	2.95	0.56	1.8 – 4.0	88.9	5.6	5.6
Romantic Appeal	17	2.42	0.61	1.8 – 4.0	35.3	47.1	17.6
Behavior Conduct	33	2.97	0.69	1.6–4.0	81.8	9.1	9.1
Close Friendships	18	2.89	0.79	1.2 – 4.0	66.7	27.8	5.6
Global Self-Worth	33	3.16	0.65	1.3 – 4.0	81.8	15.2	3.0

*Positive = making more positive than negative self-statements.

Table 6

Patient body image: Body Image Scale (BIS).

BIS Scale Scores	n	Median	Range
Overall Body Image	24	1.75	1.0 – 3.0
Primary Sex Characteristics	24	2.17	1.0 – 4.5
Secondary Sex Characteristics	24	1.62	1.0 – 3.0
Hormonally Unresponsive Characteristics	24	1.73	1.0 – 3.0

		Neutral, Unhappy, or Very Unhappy		Would Medically or Surgically Change		
Body Characteristic	**N**	**n**	**%**	**Yes (n)**	**% of Total N**	**% of Neutral, Unhappy, or Very Unhappy**
Private Parts	24	10	41.7%	5	20.8%	50.0%
Breasts	24	8	33.3%	5	20.8%	62.5%
Voice	24	7	29.2%	4	16.7%	57.1%
Weight	23	6	26.1%	4	17.4%	66.7%
Height	24	6	25.0%	2	16.7%	66.7%
Hair	24	6	25.0%	1	8.3%	33.3%
Facial Hair	24	6	25.0%	4	8.3%	33.3%
Arms	24	6	25.0%	2	4.2%	16.7%
Adam’s Apple	22	5	22.7%	2	0.0%	0.0%
Figure/Body Shape	24	5	20.8%	0	16.7%	80.0%
Appearance	24	5	20.8%	4	12.5%	60.0%
Hips	24	5	20.8%	3	8.3%	40.0%
Buttocks	24	5	20.8%	2	8.3%	40.0%
Body Hair	24	5	20.8%	2	8.3%	40.0%
Muscles	24	4	16.7%	2	12.5%	75.0%
Hands	24	4	16.7%	2	8.3%	50.0%
Thighs	24	4	16.7%	3	8.3%	50.0%
Biceps	24	4	16.7%	2	8.3%	50.0%
Shoulders	24	4	16.7%	1	4.2%	25.0%
Chin	24	4	16.7%	1	4.2%	25.0%
Eyebrows	24	3	12.5%	1	4.2%	33.3%
Waist	24	3	12.5%	1	4.2%	33.3%

		Neutral, Unhappy, or Very Unhappy			Would Medically or Surgically Change		
Body Characteristic	N	n	%	Yes (n)	% of Total N	% of Neutral, Unhappy, or Very Unhappy	
Feet	24	3	12.5%	1	4.2%	33.3%	
Nose	24	3	12.5%	0	0.0%	0.0%	
Calves	24	3	12.5%	0	0.0%	0.0%	
Face	24	3	12.5%	0	0.0%	0.0%	

Table 7

Social competencies and behavior problems: Child Behavior Checklist (CBCL) and Youth Self-Report (YSR) (T-scores).

Scale	n	Scale Scores			Category (%)		
		Mean	SD	Range	Non-Clinical	Borderline	Clinical
Parent-Report: CBCL							
Total Competencies	48	38.04	9.78	19 – 58	58.3	22.9	18.8
Activities	51	42.65	8.97	26 – 64	80.4	7.8	11.8
Social	49	40.47	10.01	20 – 62	77.6	10.2	12.2
School	51	41.39	9.63	24 – 55	68.6	19.6	11.8
Total Behavior Problems	70	55.06	13.76	26 – 76	57.1	12.9	30.0
Internalizing	70	55.21	12.86	29 – 80	55.7	10.0	34.3
Externalizing	70	53.00	12.69	28 – 83	68.6	8.6	22.9
Patient-Report: YSR							
Total Competencies	24	39.92	8.54	28 – 55	75.0	8.3	16.7
Activities	24	40.29	7.85	27 – 52	70.8	12.5	16.7
Social	24	43.50	9.86	29 – 60	66.7	29.2	4.2
Total Behavior Problems	24	53.29	8.70	38 – 70	75.0	16.7	8.3
Internalizing	24	52.71	10.78	27 – 69	70.8	12.5	16.7
Externalizing	24	50.17	8.60	29 – 63	83.3	16.7	0

Table 8

Challenges to routine psychosocial screening.

Challenge	Strategies for Addressing Challenge
Stigma	Assure acceptability of screening Make screening standard for all patients/families Provide education on comprehensive care Document patient and family satisfaction with screening
Unrecognized need	Discuss how screening can improve care/outcomes Demonstrate how screening aids care management Address discipline-specific concerns
Time	Determine actual time necessary to screen Create option for online administration at home Integrate screening into routine clinical processes Schedule appointments to accommodate time
Limited resources	Create algorithms for triaging high needs patients/families with existing resources Use aggregate data to argue for expanded psychosocial resources
Impact on workflow	Establish responsibility for questionnaire administration, scoring, and feedback to the family and the clinical team Create alerts for high-risk responses or profiles to prompt immediate attention Identify team member responsible for reviewing and coordinating response Integrate screening findings within electronic health record
Sustainability of screening	Continue to train providers to implement Monitor pathways of care Review utility of individual measures through assessment of their psychometric properties in the DSD population with ongoing refinement Utilize existing billing codes for “psychological testing” or engage insurers through contracting