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Characterizing Early Psychosocial Functioning of Parents of Children with Moderate to Severe Genital Ambiguity due to Disorders of Sex Development

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Abstract

Purpose—We examined the psychosocial characteristics of parents of children with disorders of sex development at early presentation to a disorders of sex development clinic. Parental anxiety, depression, quality of life, illness uncertainty and posttraumatic stress symptoms were assessed. Additionally we evaluated the relationship of assigned child gender to parental outcomes.

Materials and Methods—A total of 51 parents of children with ambiguous or atypical genitalia were recruited from 7 centers specializing in treatment of disorders of sex development. At initial assessment no child had undergone genitoplasty. Parents completed the Cosmetic Appearance Rating Scale, Beck Anxiety Inventory, Beck Depression Inventory, SF-36, Parent Perception of Uncertainty Scale and Impact of Event Scale-Revised.

Results—A large percentage of parents (54.5%) were dissatisfied with the genital appearance of their child, and a small but significant percentage reported symptoms of anxiety, depression, diminished quality of life, uncertainty and posttraumatic stress. Few gender differences emerged.

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Study received review board approval of participating institutions.

Conclusions—Although many parents function well, a subset experience significant psychological distress around the time of diagnosis of a disorder of sex development in their child. Early screening to assess the need for psychosocial interventions is warranted.

Keywords

anxiety; depression; disorders of sex development; quality of life; stress disorders; post-traumatic

Disorders of sex development are congenital medical conditions in which there is discordance between chromosomal, phenotypic and gonadal sex.[1] Although disease occurrence is low, somatic/phenotypic and psychological effects are significant for affected children and their parents. Compared to current reference data for health related quality of life, children report decreased health related quality of life across several areas, including overall health related quality of life, whereas parents report lower levels of emotional well-being.[2] Additionally children with congenital adrenal hyperplasia and those with other disorders of sex development diagnoses are vulnerable to development of anxiety disorders, depression and attention-deficit/hyperactivity disorder.[3,4]

During the last few decades techniques for identifying and diagnosing DSD have improved. With these medical advances research has begun to focus on the psychosocial adjustment of parents of children with DSD.[1,5] Preliminary research has shown that parents of children with DSD are at increased risk for psychological distress.[6,7] Furthermore, research indicates that parents raising boys with DSD experience higher rates of depression compared to those raising girls.[6] Much remains unknown about the psychological effects on parents of an infant with atypical genitalia. However, previous research demonstrates that decisions about surgery evoke distress and greater stress immediately after the diagnosis of a disorder of sex development in the child.[8]

Although previous research has identified negative psychosocial outcomes in parents of children with DSD, these series are limited by study design, inclusion of children across developmental levels and retrospective assessment postoperatively. To our knowledge, no study has examined psychosocial functioning of parents during early presentation to a DSD clinic, when decisions regarding genitoplasty are often being considered. Thus, we aimed to assess parental anxiety, depression, QOL, illness uncertainty and PTSS within the first 2 years of birth of the affected child and before genitoplasty to understand the psychosocial characteristics of parents of patients recently born with atypical genitalia due to DSD.

No specific hypotheses were made in terms of expected levels of parental adjustment, given the lack of research among parents of newly diagnosed children. However, similar to parents of children with other chronic or acute illnesses, we expected increased mean scores above existing norms for the clinical measures. We examined gender effects on parental adjustment, hypothesizing that parents of boys would report increased psychological distress compared to parents of girls. The current analysis is part of an ongoing prospective study of parents of children born with DSD from 7 pediatric hospitals.

MATERIALS AND METHODS

Participants

Participants were 51 parents of children with moderate to severe genital ambiguity due to DSD (51 of 59 surveyed, 86.4% enrollment rate), including atypical external genitalia, recruited within 2 years of birth and before the child underwent genitoplasty. Patient age was 2 years or younger at enrollment. Families were excluded from the study if the child had profound central nervous system malformations that would make future investigation of gender impractical or if the child had other organ system malformations. Atypical external genitalia were defined as Prader rating 3 to 5 in a genetic female or Quigley rating 3 to 6 in a child with a 46,XY or 45,XO, 46,XY chromosomal complement.

Diagnoses are outlined in table 1. Families of children seen at DSD clinics across the United States were asked to participate in the study. The supplementary table (http://jurology.com/) contains recruitment site information. The demographic form consisted of parent reported child gender of rearing, child age and date of birth, diagnosis of disorder of sex development, parent gender, marital status, race/ethnicity, parent date of birth and income.

The Cosmetic Appearance Rating Scale is a single item self-reported Likert-type scale, where parents rate their satisfaction with the cosmetic appearance of the genitals of their child before genitoplasty. Answers range from 1 (satisfied) to 4 (dissatisfied). Higher scores indicate greater dissatisfaction with appearance.

BAI is a 21-item self-reported multiple-choice measure of anxiety symptoms. Participants were asked to indicate the degree to which they were bothered by symptoms of anxiety. Items are rated on a scale of 0 to 3, with higher scores representing greater levels of anxiety. According to norms, total scores from 0 to 9 indicate minimal anxiety, 10 to 16 mild anxiety, 17 to 29 moderate anxiety and 30 to 63 severe anxiety. Cronbach alpha for the current sample was 0.93.

BDI-II is a 21-item self-reported multiple-choice measure of depressive symptomatology. Item responses range from 0 to 3. Higher scores reflect higher levels of depressive symptoms. Total scores from 0 to 13 are considered to be in the minimal range, while scores of 14 to 19 indicate mild depression, 20 to 28 moderate depression and 29 to 63 severe depression. Cronbach alpha for the current sample was 0.96.

SF-36 is a 36-item self-reported measure used to assess health related quality of life. Responses are presented on a 5-item Likert scale, with higher scores indicating better health related QOL. SF-36 is composed of 8 subscales. Subscale scores are combined to calculate 2 summary scores assessing physical and mental health. A score of 42 on the mental health summary indicates clinically significant psychiatric symptoms. Cronbach alpha for the current sample was 0.96.

PPUS is a 31-item parent reported measure that assesses the level of illness related uncertainty the parent experiences.[9] Item responses are rated on a 5-point Likert scale ranging from “strongly agree” to “strongly disagree.” Higher scores on the PPUS indicate greater levels of uncertainty. Cronbach alpha for the current sample was 0.93.

IES-R is a 22-item parent reported measure used to assess PTSS.[10] Responses for the IES-R are presented on a Likert scale, ranging from “not at all” to “extremely.” Higher scores indicate increased PTSS. A total score of 33 or higher is the clinical cutoff for the IES-R.[7] Cronbach alpha for the current sample was 0.95.

Procedures

Review board approval was obtained from participating institutions. Parents were approached during regularly scheduled clinic appointments. Parents were eligible to enroll in the study regardless of their decision to proceed with genitoplasty. However, the baseline data presented were collected preoperatively for all families. Families will be compensated $50 after completion of the 5-year study.

Statistical Analyses

Analyses were conducted using SPSS®, version 22. Data were examined for missing information. We first described scores for each measure in the context of existing criteria for severity and, when possible, compared parent mean scores to existing norms. Bivariate correlations were computed to examine relationships between demographic variables (eg race/ethnicity, parent age, child age, surgery, yearly income), and all dependent variables (eg BAI, BDI-II, SF-36 subscales and summary scales, PPUS, IES-R). Independent samples t-tests and analyses of covariance were conducted to determine differences in scores of anxiety, depression, health related QOL, IU and PTSS based on parent gender and child gender of rearing.

RESULTS

Parent and child demographics are presented in tables 1 and 2. Bivariate correlations were examined between demographic variables and outcome scores. Correlations are outlined in table 3. Correlating variables were controlled for in statistical analyses. Total scores with means and standard deviations for all study measures are outlined in table 4.

Parent Report of Cosmetic Appearance of Child External Genitalia

Examination of Cosmetic Appearance Rating Scale scores revealed a mean ± SD score of 2.53 ± 0.97, where 10 parents (19.6%) reported being satisfied with the appearance of the genitalia in their child, 11 (21.6%) were somewhat satisfied, 21 (41.2%) were somewhat dissatisfied and 6 (11.8%) were dissatisfied. Data for 3 patients (5.9%) were missing.

Further examination of the data showed almost identical scores for how mothers (mean ± SD 2.50 ± 0.99) and fathers (2.45 ± 0.96) rated the cosmetic appearance of genitalia in their child. Parent ratings of infants being raised as boys (mean ± SD 2.56 ± 0.96), girls (2.38 ± 1.02) or uncertain (2.50 ± 0.71) were also similar. Finally, examining ratings of mothers and fathers of boys again indicated similar mean ± SD scores of 2.69 ± 0.95 and 2.42 ± 1.00, respectively.

Parent Anxiety Symptoms

Mean ± SD score on the BAI was 8.06 ± 9.78. A total of 31 parents (60.8%) reported symptoms in the minimal range, 5 (9.8%) in the mild range, 4 (7.8%) in the moderate range and 4 (7.8%) in the severe range of anxious symptoms. Data were missing for 7 parents (13.7%). Thus, 13 parents of children with a new diagnosis (25.4%) experienced mild to severe symptoms of anxiety, with 8 (15.6%) falling in the moderate to severe range. According to norms, mean BAI total scores for this sample were lower than would be expected compared to a community sample of adults.[11]

ANCOVA was completed to determine if there was a relationship between child gender of rearing and BAI total scores when controlling for parent age and annual income. Results were nonsignificant ($p > 0.05$), and thus gender of rearing of the child did not impact parent reported anxiety.

Parent Depressive Symptoms

Mean ± SD score on the BDI-II was 8.78 ± 11.30. A total of 32 participants (62.7%) reported depressive symptoms in the minimal range, 3 (5.9%) in the mild range, 3 (5.9%) in the moderate range and 3 (5.9%) in the severe range. Data were missing for 10 parents (19.6%). Thus, approximately 18% of parents experienced a clinically significant level of depressive symptoms. The majority of participants experienced depressive symptomatology similar to that in the typical population, while a small subset of parents experienced increased levels of depressive symptomatology.

ANOVA was completed to determine the relationship between child gender of rearing and BDI-II total scores. Results were nonsignificant ($p > 0.05$), indicating that child gender of rearing did not influence depressive symptoms among parents of children with DSD.

Quality of Life Outcomes

Mean ± SD scores were 54.80 ± 11.09 on the SF-36 physical health summary and 45.33 ± 13.06 on the mental health summary. Examination of cutoffs for the mental health summary score demonstrated that more than 25% of parents reported experiencing a significant negative impact on QOL related to their mental health. Additionally QOL subscales were consistently below expected norms for 25 to 34-year-old individuals from a community sample.[12]

ANCOVA was completed to determine the relationship between child gender of rearing and SF-36 physical health subscale scores when controlling for child age. Results were nonsignificant ($p > 0.05$), indicating that child gender of rearing did not influence QOL related to physical health among parents. Another ANCOVA was completed to determine if there was a relationship between child gender of rearing and SF-36 mental health subscale scores when controlling for parent gender and yearly income. These results were also nonsignificant ($p > 0.05$).

Parental Illness Uncertainty

Examination of PPUS revealed a mean ± SD total score of 64.78 ± 19.01. Therefore, parents of children with DSD scored slightly lower than parents of children with cancer or cystic fibrosis.[13]

ANOVA was completed to determine the relationship between child gender of rearing and PPUS total scores. A significant relationship between parent reported child gender of rearing and IU scores was found ($F[2.42] = 3.352$, $p = 0.045$). Parents of boys and parents who were unsure of the gender of their child had greater uncertainty (mean ± SD 71.90 ± 17.92 and 72.00 ± 0.0, respectively), compared to parents of girls (57.96 ± 18.60).

Parent Posttraumatic Stress Symptoms

Mean ± SD total score on IES-R was 18.73 ± 18.16. Eight parents (15.7%) reported PTSS in the elevated range. According to previous research, parents of children with a cancer diagnosis report similar mean scores on this measure,[14] suggesting that these populations are comparable.

ANCOVA was completed to determine if there was a relationship between child gender of rearing and IES-R total scores, controlling for marital status. Results were nonsignificant ($p > 0.05$), suggesting no relationship between child gender of rearing and PTSS.

DISCUSSION

We aimed to characterize the early psychosocial functioning of parents of children with DSD before genitoplasty. Overall a subset of parents experienced distress following diagnosis of a disorder of sex development in their child. Approximately 53% of parents were dissatisfied with the genital appearance of their child. There were no differences in how mothers and fathers rated the appearance of their child.

Although many parents of children with DSD did not evidence clinically increased symptoms of depression or anxiety, a considerable proportion (18% to 25%) experienced depressive and anxious symptomology. Furthermore, QOL mental health scores among parents were lower than among community samples. Additionally QOL physical functioning scores were only slightly higher than scores for parents of children with cancer.[15] Therefore, a subgroup of parents are experiencing a significant impact on physical and mental QOL.

Parent scores of IU were comparable to parents of children with other chronic illnesses, such as type I diabetes mellitus.[16] Additionally a subset of parents reported increased scores of PTSS, which is a concern from a clinical perspective. The only other study to examine PTSS in parents of children with DSD found rates of PTSS similar to those of parents of children with cancer.[7]

Parents of boys reported more uncertainty than parents of girls. This uncertainty may be due to the fact that the etiologies underlying DSD for boys in the present study are more heterogeneous and rare. Thus, less is known about the natural history of these types of DSD and the likelihood of future offspring being similarly affected. Although differences were

found between parents of boys and girls related to IU, inconsistent with previous studies no other psychosocial outcomes differed for parents of boys and girls.[6] However, this sample of parents differed from previous samples as data were collected during the early visit of the family to the clinic and preoperatively.

These findings should be interpreted in light of some limitations. First, the study design was cross-sectional, and thus causal relationships cannot be established. As this study continues, longitudinal data will become available to assess the trajectory of adjustment through time. No true comparison group was used, and it is impossible to know whether the symptomatology reported is directly due to having a child with DSD. Our sample was largely married (72.5%) and white (60.8%), limiting generalizability to single parents or parents from other minority groups. Results should be interpreted in light of the fact that the sample was composed of parent dyads, and, therefore, dependency between parents should be considered. Finally, it could not be guaranteed that parents did not discuss their responses with each other, thus creating problems with dependency of their answers.

Strengths of this study include that parents were recruited from DSD clinics across the United States. In addition, there were a great number of fathers represented in this sample. Therefore, results are more generalizable than previous investigations. Finally, participants were affected by a wide range of DSD diagnoses, adding to the generalizability of the results.

CONCLUSIONS

A subset of parents of children diagnosed with DSD experience significant psychological distress at diagnosis. Parents need psychological support, especially parents of boys and those who feel uncertain about the gender of their child. Although each family is different, by understanding and addressing parent psychosocial problems health care providers optimize child outcomes. Incorporating family based interventions and psychological supports into regular care for families of children with DSD could act to attenuate psychological symptomatology in this population.[17]

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Abbreviations and Acronyms

BAI®	Beck Anxiety Inventory®
BDI®-II	Beck Depression Inventory-II®
DSD	disorders of sex development
IES-R	Impact of Event Scale-Revised
IU	illness uncertainty
PPUS	Parent Perception of Uncertainty Scale

PTSS	posttraumatic stress symptoms
QOL	quality of life
SF-36®	Short Form (36) Health Survey

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Table 1

Parent demographics

% Rearing Gender*	
M	48.1
F	46.3
Unsure	3.7
Missing	1.9
Mean ± SD age (mos)	9.70 ± 6.45
% Karyotype:	
46,XX	44.8
46,XY	41.4
45,XO/46,XY	13.8
% Diagnosis:	
Congenital adrenal hyperplasia	44.8
Unclassified	24.1
Mixed gonadal dysgenesis	13.8
Partial androgen insensitivity syndrome	6.9
Unknown	6.9
5-Alpha reductase deficiency	3.4

*Per parent report.

Table 2

Parent demographics

% Gender:	
M	45.1
F	52.9
Missing	2.0
Mean ± SD age (yrs)	31.16 ± 5.90
% Marital status:	
Married *	72.5
Single, never married	5.9
Living with partner *	5.9
Single, divorced	3.9
Missing	11.8
% Race/ethnicity:	
White	60.8
Hispanic	15.7
Black	3.9
Asian-American	7.8
Other	3.9
Multiracial	2.0
Missing	5.9
% Yearly household income: †	
Less than $20,000	17.2
$20,000—$39,000	24.1
$40,000—$59,000	13.8
$60,000—$79,000	6.9
$80,000—$99,000	10.3
$100,000 or Greater	24.1
Missing	3.5

* Partner is biological parent of child with disorder of sex development.

† Based on 29 households.

Table 3

Correlations between key study variables

	1	2	3	4	5	6	7	8	9	10	11	12	13
Child age	–												
Child gender of rearing	−0.20	–											
Parent age	0.02	0.11	–										
Parent gender	−0.05	0.05	0.26	–									
Parent race/ethnicity	0.01	0.23	−0.16	0.02	–								
Parent marital status	−0.12	−0.21	0.32*	−0.04	−0.19	–							
Yearly income	−0.31*	0.13	0.41*	0.03	−0.08	0.29	–						
BAI	0.16	0.06	−0.33*	−0.31*	0.03	−0.40*	−0.34*	–					
BDI-II	0.13	−0.03	−0.02	−0.27	0.08	−0.18	−0.17	0.70†	–				
SF-36 physical health	−0.32	0.02	−0.24	0.16	−0.07	−0.33	0.12	−0.01	−0.62†	–			
SF-36 mental health	−0.17	0.02	0.19	0.42*	−0.16	0.32	0.43†	−0.75†	−0.83†	0.32*	–		
PPUS	0.17	0.35*	0.10	−0.09	0.17	−0.25	−0.10	0.46†	0.48†	−0.21	−0.56†	–	
IES-R	−0.01	0.29	0.01	−0.23	0.08	−0.50†	−0.19	0.67†	0.52†	−0.08	−0.72†	0.56†	–

*p <0.05.

†p <0.01.

Table 4

Key study variables

	Mean ± SD	Norms
Cosmetic Appearance Rating Scale	2.5 ± 1.0	Not applicable
BAI	8.1 ± 9.8	11.54 ± 10.3[11]
BDI-II	8.8 ± 11.3	23.16 ± 9.5
SF-36:		
Physical functioning	90.3 ± 22.5	94.5 ± 13.5[12]
Role physical	84.5 ± 24.5	87.5 ± 29.8[12]
Bodily pain	81.2 ± 22.3	86.7 ± 22.6[12]
General health	74.8 ± 22.0	79.4 ± 17.9[12]
Vitality	55.4 ± 23.5	66.8 ± 18.4[12]
Social functioning	78.8 ± 26.3	91.1 ± 16.9[12]
Role emotional	80.2 ± 29.7	90.3 ± 26.2[12]
Mental health	53.1 ± 34.6	77.2 ± 16.2[12]
Physical health summary	54.8 ± 11.1	50.0 ± 10.0
Mental health summary	45.3 ± 13.1	50.0 ± 10.0
PPUS	64.8 ± 19.0	70.3 ± 14.5[13]
IES-R	18.7 ± 18.2	18.5 ± 16.1[14]