Degree of External Genital Malformation at Birth in Children with a Disorder of Sex Development and Subsequent Caregiver Distress

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Abbreviations and Acronyms

BAI = Beck Anxiety Inventory

BDI-2 = Beck Depression Inventory, 2nd edition

CAH = congenital adrenal hyperplasia

DSD = disorders of sex development

Study received institutional review board approval.

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Purpose: We determined whether the degree of genital malformation at birth in children with a disorder of sex development is related to subsequent caregiver distress, specifically symptoms of depression and anxiety.

Materials and Methods: A total of 66 caregivers of children with disorders of sex development were recruited from 3 centers that specialize in disorders of sex development medicine. The caregivers completed the Beck Depression Inventory, 2nd Edition and the Beck Anxiety Inventory. The child's Prader score at birth was determined by the child's treating pediatric endocrinologist and/or pediatric urologist at each institution.

Results: Results from the current study revealed that for caregivers of male children, under masculinization of the child's genitals at birth was significantly related to higher levels of subsequent caregiver depression. In contrast, over masculinization of the genitals of female children at birth was unrelated to caregiver depression or anxiety.

Conclusions: These findings suggest that caregivers of male children with disorders of sex development may be at increased risk for psychological distress and could benefit from family based psychosocial interventions.

Key Words: disorders of sex development, genitalia, parent-child relations, mental health

Disorders of sex development are a class of congenital conditions in which discrepancy exists among chromosomal, gonadal and phenotypic sex.[1] For some parents of newborns affected by DSD, decisions about sex of rearing can be difficult, especially when malformations of the external genitalia exist at birth. Although it is uncommon for children with DSD to reject their sex of rearing,[2] this rejection happens more often in children reared female than male.[3] Although specific statistics on the prevalence of external genital malformations among newborns are difficult to obtain, it is estimated that minor abnormalities such as glanular hypospadias occur as frequently as 1 in 300 births.[4] More severe malformations associated with difficulty in assigning sex to newborns with DSD are thought to be less frequent and have been estimated at 1 in 5,000 births.[5] The majority of

0022-5347/12/1884-1596/0
THE JOURNAL OF UROLOGY®

http://dx.doi.org/10.1016/j.juro.2012.02.040
Vol. 188, 1596-1600, October 2012
Printed in U.S.A.

children with 46,XX DSD are affected by CAH due to 21-hydroxylase deficiency. In contrast, children with 46,XY DSD including external genital malformations only receive a definitive clinical diagnosis 50% of the time.[6]

While some evidence exists that people born with genital malformations resulting from DSD have an increased risk of psychological distress and impaired sexual quality of life,[7–9] little is known about the impact of a child's DSD on the mental health of the parents.[10] We previously reported that female and male caregivers were differentially affected by decisions concerning whether to have their child undergo surgery in an attempt to treat atypical genital development. Specifically mothers reported more parenting stress than fathers if their child did not undergo genital surgery. Furthermore, fathers reported more parenting stress and were more likely use an overprotective parenting style than mothers if their child underwent genital surgery before age 12 months.[11] Finally, our group reported that female caregivers of boys born with genital malformations viewed their children as significantly more vulnerable than female caregivers of girls born with atypical genitalia.[12] Although both of these investigations examined the effects of caring for a child with DSD on caregiver mental health and parenting characteristics, neither study investigated whether the child's degree of genital malformation at birth related to caregiver distress.

The extant literature on pediatric illness across multiple health conditions documents that a subset of parents of children with a chronic medical condition are indeed at increased risk for psychological distress, including depression and anxiety. For example, increased levels of depression and/or anxiety have been identified in parents of children with juvenile fibromyalgia syndrome,[13] asthma,[14] congenital cardiac malformations,[15] cerebral palsy[16] and cancer,[17] among others. Additionally, some studies reveal that parental levels of depression and anxiety are negatively affected by the increasing severity of a child's illness.[14]

In this study we determine whether caregiver levels of distress, specifically anxious and depressive symptoms, relate to the degree of malformation of the child's genitals at birth. We hypothesize that greater malformation of a child's external genitalia at birth will be associated with higher levels of depressive and anxious symptoms among caregivers. Specifically we hypothesize that for caregivers of newborns with a 46,XX chromosomal complement and reared female, higher Prader scores (ie indicating more over masculinization of the external genitalia) at birth will be associated with higher levels of caregiver depressive and anxious symptoms. Conversely we hypothesize that for caregivers of newborns with a 46,XY chromosomal complement and reared male, lower Prader scores (ie indicating more under masculinization) at birth will be associated with greater caregiver reported anxious and depressive symptoms.

MATERIALS AND METHODS

Participants

Participants for the current study include caregivers of children with DSD who had concordance between their genetic sex and sex of rearing. Diagnoses included 46,XX DSD due to 21-hydroxylase deficiency or transposition of the SRY gene reared female, and 46,XY DSD due to androgen biosynthetic defects, androgen insensitivity or unknown causes reared male (tables 1 and 2). Primary caregivers of children with DSD were invited to participate if the child was diagnosed with DSD at least 6 months before recruitment. The 6-month cutoff was chosen to insure that caregivers of children newly diagnosed with DSD were not included in the study as they are expected to exhibit increased levels of depressive and anxious symptoms. Other exclusion criteria were that the child's genetic sex and sex of rearing were discordant, and that the child had significant developmental delay or cognitive impairment. Inclusion criteria were that the caregiver was 18 years old or older, was able to provide informed consent and was able to read English at an 8th grade reading level. In families with 2 caregivers, both were invited to participate. However, for the current study only 1 caregiver per child was used. A random number generator was used to randomly select the included caregiver for each child. Families served by DSD specialists at the University of Oklahoma Children's Hospital, Johns Hopkins Hospital, and the Women's and Children's Hospital of Buffalo were invited to participate. In addition, other parents could participate if they initiated contact with the senior author.

Demographics Questionnaire

As part of a larger study of caregiver adjustment to having a child with DSD, participants completed several subsets of measures. Caregivers completed a questionnaire that assessed age and race, child age, number of children and caregivers in the home, marital status, household income and whether the caregiver had ever received psychological counseling.

Table 1. *Demographic characteristics of caregiver participants*

% Male gender	36
Median age (range)	34.96 (18–49)
% Married	71.7
% Ethnicity:	
Caucasian	74.6
African-American	7.5
Native American	7.5
Hispanic	4.5
Other	3.0
% Annual household income:	
Less than \$20,000	15.0
\$20,000–\$40,000	18.0
\$40,000–\$60,000	24.4
Greater than \$60,000	38.8
Not reported	3.0