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Gender Identity and Preferences in Children with Variations in Sex Development

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ABSTRACT

Objective: To assess gender-typed preferences and gender identity in children with and without variations in sex developments (VSDs).

Methods: In this cross-sectional study, 78 children with VSDs (ages 3-12; mean age=7 years; 55% White, non-Hispanic) recruited through specialty clinics in the United States and 78 children without VSDs (ages 3-13; mean age=7 years; 55% White, non-Hispanic) recruited through university-based community databases completed assessments of gender-typed toy, clothing and peer preferences, continuous and categorical measures of gender identity, and perceived similarity to boys and to girls.

Results: Generally, children with and without VSDs did not differ in their gender development on 5 of 7 measures for each gender group. Children raised as girls who had VSDs had more masculine toy preferences, $t(84.89)=3.421$; $p=0.001$; $d=0.698$, and viewed themselves as more

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similar to boys, $t(67.43)=2.994$; $p=0.004$; $d=0.648$, than comparison children raised as girls. Boys with VSDs selected more masculine toys [$t(55.17)=2.413$; $p=0.019$; $d=0.623$], and responded in a more-masculine way on the continuous gender identity measure [$t(38.40)=2.364$; $p=0.023$; $d=0.621$], than did boys in the community comparison sample, though these effects, unlike the effects amongst girls, were not robust against corrections for multiple comparisons.

Conclusion: During early and mid-childhood, VSDs were not strongly associated with differences in gender development. Future longitudinal research on the gender development of youth with VSDs is necessary, particularly as they mature into adolescence.

Keywords: Disorders of sex development, gender identity, intersex

What is already known on this topic?

Gender development and relatedly, gender identity, are complex issues in the context of variations in sex development (VSD). Children with VSD appear to experience higher gender identity discordance or gender dysphoria relative to the general population. However, few studies have studied gender development in children with VSD using contemporary measures and appropriate comparison groups.

What this study adds?

This study of 78 children with VSD aged between 3 and 12 years compared their gender development to that of a comparison matched sample of youth without VSD. Few differences between groups were observed, and youth in both groups reported a range of both gender stereotypic and diverse identities and preferences. Results have implications for VSD care.

Introduction

Variations in sex development (VSDs) (1) include genital or reproductive differences (e.g., “atypical” genitalia), less common genetic karyotypes (e.g., 47,XXY, 45,X), and/or a combination of features (e.g., vulva and testes) that are either discordant with an individual’s sex chromosomes (1) or less common in the general population. [Terminology is a complex issue in the context of variations in sex development, including historic use of stigmatizing terms to describe these variations, with controversies continuing today about use of terms such as “differences” or “disorders” of sex development or “intersex” across healthcare providers, patients, and families [Davies (2), 2015]. We use the term ‘variations in sex development’ and specific diagnosis names [Lin-Su et al. (3), 2015; e.g., congenital adrenal hyperplasia (CAH)], to avoid terms more often associated with these controversies. We report data including and separating data for females with CAH to reflect stakeholder preferences. Exact estimates of the frequency of VSDs range from 1.7 in 100 births (4) to 1.8 in 10,000 births (5) depending on the breadth of one’s definition and inclusion or exclusion of particular conditions.

Parents of youth with VSDs often come to healthcare providers with questions about the likely course of their children’s gender development. The aim of the current research was to provide contemporary answers about what gender development looks like in children with VSDs as compared to their peers without VSDs.

Today, “gender development” is understood to encompass many distinct aspects of a person’s experience including their gender

identity (i.e., whether they think of their gender category as a boy, girl, nonbinary, etc), gender presentation (i.e., whether they prefer clothing that is culturally-stereotyped as masculine, feminine, or androgynous), and gender role (i.e., for children, preferred play style or type) (6). Empirical data from children without (known) VSDs supports the notion that while, for example, gender identity is often correlated with gender-typed preferences, these do not always closely correspond with one another in stereotypical ways and can be fully dissociable in some children (7,8,9).

Due to the presence of less common variations in genitalia and/or the potential for discordances between physical characteristics and sex chromosomes, healthcare providers and parents of children with VSDs sometimes grapple with whether to initially raise a child with a VSD as a boy, a girl, or in a more gender neutral or open way [e.g., as a theybie, (2,10)]. This decision can also have implications for potential surgical or endocrine interventions, a controversial and debated issue in the context of VSDs (11).

Studies of individuals with VSDs suggest that some may experience more uncertainty about their gender identity or might experience more gender identity change than people without VSDs. For example, children with CAH who are initially raised as girls sometimes show higher rates of gender dysphoria (12,13) or identification as boys (14,15,16) than comparison groups, but other studies find no differences in gender identity (17,18). [When we use the term CAH for children raised as girls, we include 46,XX individuals with 21-hydroxylase deficiency,

17 α -hydroxylase, or 11 β -hydroxylase. The two 46,XY youth with CAH, both raised as boys, have 17 α -hydroxylase].

Research on other aspects of gender development in children with VSDs is more limited. The literature that exists in this area has primarily focused on children with CAH who are raised as girls. The most robust effect reported in this literature suggests that, compared to girls without CAH, girls with CAH often show stronger preferences for toys that are culturally stereotyped as masculine (18,19, 20). Other studies have shown a parallel “masculinity bias” amongst girls with CAH in other domains: masculine-typed games (21), masculine playmates (22), and masculine-type clothing/dress-up (18). Much less is known about gender-typed preferences in children with other VSDs as systematic analyses and the use of a wide range of measures of gender expression and/or gender roles are rarer in this literature.

A further limitation of past work on gender development in children with VSDs is that much of it was completed 20 or more years ago. In the intervening time, societal ideas about gender have been shifting. Setting aside VSDs, more youth today are identifying as genders that differ from the ones they are assumed to have at birth. Large-scale and representative surveys and polls suggest that anywhere from 1.2 to 5% of adolescents and young adults identify as transgender and/or nonbinary (23,24,25). In contrast, as recently as 2011, estimates suggested numbers closer to 0.3% of the adult population identified as transgender (26).

In the present work, our aim was to investigate gender development, adopting measures of a diverse range of distinct aspects of gender, in a group of youth with VSDs and compare them to a group of youth without VSDs. Though we did not officially preregister a hypothesis, the study was designed to ask if children with VSDs showed more gender nonconformity than children without VSDs.

Methods

Participants and Procedures

Participants aged 3-12 years with variations in sex development were recruited from 3 children’s hospitals: Seattle Children’s Hospital (n=44); The Children’s Hospital of Philadelphia (n=22); and Nationwide Children’s Hospital (n=12) between October 7, 2019, and June 20, 2023 ($M_{age}=7.14$ years, $SD_{age}=2.85$ years). See Supplemental Material for additional recruitment details. The list of diagnoses of participants with VSDs is shown in Table 1. Demographics of both participant groups are included in Table 2.

Community comparison participants were recruited through one of two university databases of families who signed up to participate in child development research at the University of Washington and Princeton University. Each participant with a VSD was matched with a child raised as the same gender who

was within five months of age at the time of testing ($M_{age}=7.18$ years, $SD_{age}=2.91$ years).

This research was approved through the IRBs of five testing locations. At least one parent had to provide informed consent, the child had to provide verbal assent, and the child had to complete the study assessment to be included in this analysis. This study approved by the Institutional Review Board of Princeton University (approval no.: 12940, date: 25.07.2020).

Child Measures

Categorical Gender Identity (7)

Children were asked if they were “a boy,” “a girl,” or “something else.” Youth who selected “something else” then chose between “both,” “neither,” “it changes over time,” and “I don’t know.”

Continuous Gender Identity (27)

In line with more recent conceptualizations of gender identity as a non-discrete aspect of human experience (28,29,30), children were given a non-categorical measure of gender identity. Children were shown a line on which they could indicate their gender from one end, described and labeled as “boy/man” (0) and the other end described and labeled as “girl/woman” (1), with a label in the middle indicating “in the middle means you feel like a mix of both”. Children were told that “Some people feel they are a boy, some people feel they are a girl, and some people feel they are somewhere in between a boy and a girl. On the line below, move the slider to the place you think best shows how you feel on the inside.” The slider appeared at the center of the slider, and they could move it in either direction.

Peer Preference (31)

Children saw eight trials, each featuring the photographs of two children. Six of the trials included one child who looked stereotypically like a girl and one child who looked stereotypically like a boy (the two remaining trials were filler trials featuring two children of the same gender, not used to compute scores; pairs within a trial were matched for race: six pairs included White children, one pair each were Black and Asian). Children were asked whom they would rather be friends with. Responses are reported as the proportion of times children selected the girl.

Toy and Clothing Preference (7,31)

Participants completed four trials selecting toys (“Which toy would you like to play with the most?”) and four trials selecting clothing (“Which outfit would you like to wear?”). On each trial there were five possible selections presented in a random array. The items on each trial were selected based on pilot testing. Within each trial (i.e., within one set of five items), there was one item that the pilot (child) participants rated as especially masculine, one was seen as moderately masculine, one was

Table 1. Participant VSD variations and the gender assigned at birth

Gender-of-raising	Diagnosis	Sample size	Categorization
Boy	Hypospadias	6	Severe hypospadias
	Hypospadias with 87% 46,XY and 13% sex chromosome aneuploidy consisting of XYYY	1	
	Hypospadias, bifid scrotum, bilateral undescended testes	1	
	Klinefelter syndrome	4	Klinefelter syndrome + sex chromosome aneuploidy
	48,XXXY[4]/49,XXXXY[26]	1	
	Klinefelter syndrome + 21-hydroxylase deficiency	1	
	45,X/46,XY mosaicism with mixed gonadal dysgenesis	2	Mosaicism + mixed gonadal dysgenesis
	45,X/46,XY mosaicism	1	Other VSD
	17 α -hydroxylase deficiency (46,XY)	2	
	Undescended testes	2	
	46,XY 5-alpha reductase deficiency	1	
	46,XY gonadal dysgenesis	1	
	46,XY partial gonadal dysgenesis due to a pathogenic variant in <i>MAP3K1</i>	1	
	Hypogonadotropic hypogonadism	1	
	47,XYY	1	
	Ovotesticular DSD	1	
	Ovotesticular DSD secondary to chimerism	1	
	Vanishing testes syndrome	1	
	46,XX male	1	
Girl	21-hydroxylase deficiency	18	46,XX CAH
	17 α -hydroxylase deficiency (46,XX)	7	
	11-beta-hydroxylase deficiency	3	
	46,XX DSD due to a heterozygous pathogenic variant of <i>WT1</i> (gain-of-function)	1	46,XX nonsyndromic VSD
	46,XX female with UG (urogenital) sinus and posterior labial fusion	1	
	46,XX female with a UG, posterior fusion, duplicated vagina/cervix	1	
	46,XX female with clitoromegaly, posterior fusion, and UG sinus	1	
	46,XX isolated clitoromegaly	1	
	MRKH	1	46,XY nonsyndromic VSD
	CAIS	2	
	17 beta-HSD deficiency	1	
	46,XY 5-alpha reductase deficiency	1	
	46,XY DSD	1	
	46,XY partial gonadal dysgenesis with a urogenital sinus	1	
	PAIS	1	
	Turner syndrome	4	Turner syndrome
	Turner syndrome with mosaicism	3	

Categorization applies to presentation of data in Figure 2

VSD: variations in sex development; DSD: differences of sex development; MAP3K1: mitogen-activated protein kinase kinase kinase 1; CAH: congenital adrenal hyperplasia; WT1: Wilms tumor 1; UG: urogenital; MRKH: Mayer-Rokitansky-Küster-Hauser syndrome; CAIS: complete androgen insensitivity syndrome; HSD: hydroxysteroid dehydrogenase; PAIS: partial androgen insensitivity syndrome

Table 2. Parent-reported participant demographics, including reports of gender/sex history, current gender, and current pronouns			
		Children with variation n=78	Community comparison n=78
Gender-of-raising	Boy	38.46%	38.46%
	Girl	61.54%	61.54%
Race and ethnicity	White, non-hispanic	55.13%	62.82%
	White, hispanic	6.41%	6.41%
	Asian, non-hispanic	8.97%	5.13%
	Black, non-hispanic	3.85%	2.56%
	Middle Eastern, non-hispanic	1.28%	0.00%
	Native American/Alaska native, non-hispanic	1.28%	0.00%
	Multiple race/ethnicity	12.82%	19.23%
	Incomplete or no response	10.26%	3.85%
Education of participating parent	Some schooling	1.28%	0.00%
	High school diploma	15.38%	1.28%
	Some college	28.21%	7.69%
	Bachelor's degree	21.79%	25.64%
	Advanced degree (MA, MD, PhD, etc)	28.21%	64.10%
	Non-response	5.13%	1.28%
Household income	Under \$25,000/year	11.54%	0.00%
	\$25,001-\$50,000/year	12.82%	1.28%
	\$50,001-\$75,000/year	8.97%	8.97%
	\$75,001-\$125,000/year	21.79%	20.51%
	Over \$125,001/year	39.74%	65.38%
	Non-response	5.13%	3.85%
Child's relationship to participating parent	Biological child	84.62%	98.72%
	Adopted child	8.97%	0.00%
	Grandchild	1.28%	0.00%
	Non-response	5.13%	1.28%
Informed of VSD before birth	No	74.36%	97.44%
	Yes	17.95%	1.28%
	Unknown (not birth parent)	5.13%	0.00%
	No response	2.56%	1.28%
Reported sex before child's birth	Binary aligned with gender in which child was raised	74.36%	84.62%
	Binary not-aligned with gender in which child was raised	2.56%	0.00%
	Was not told	10.26%	14.10%
	Unknown (not birth parent)	5.13%	0.00%
	Other	5.13%	0.00%
	Non-response	2.56%	1.28%
Sex on birth certificate	Binary aligned with gender in which child was raised	94.87%	98.72%
	Binary not-aligned with gender in which child was raised	2.56%	0.00%
	Other	0.00%	0.00%
	Non-response	2.56%	1.28%

Table 2. Continued		Children with variation n=78	Community comparison n=78
Child's gender	Binary aligned with gender in which child was raised	94.87%	97.44%
	Binary not-aligned with gender in which child was raised	0.00%	0.00%
	Both	1.28%	1.28%
	Neither	1.28%	0.00%
	Other	0.00%	0.00%
	Non-response	2.56%	1.28%
Child's pronouns	Binary aligned with gender in which child was raised	93.59%	97.44%
	Binary not-aligned with gender in which child was raised	0.00%	0.00%
	They/them	2.56%	0.00%
	Mixed	1.28%	1.28%
	Non-response	2.56%	1.28%

VSD: variations in sex development; MA: Master of Arts; MD: Doctor of Medicine; PhD: Doctor of Philosophy

gender neutral, one was moderately feminine, and one was especially feminine. Participants' selections on each trial were initially coded as 1 (selecting most masculine) to 5 (selecting most feminine). Their scores on the four trials of each type were averaged ($\alpha_{\text{toys}}=0.88$; $\alpha_{\text{clothes}}=0.87$) and then recoded to match the other measures such that the overall score ranged from 0 (most masculine) to 1 (most feminine). See Supplementary Material for more details on this task's piloting process and the variation in stimuli seen across participants.

Similarity to Boys and Girls (32)

Children were asked five questions about how similar they are to boys and five questions about how similar they are to girls (e.g., "How similar are you to boys/girls?", "How much do you look like boys/girls?", "How much do you act like boys/girls?"). Children indicated their responses on a 5-point pictorial scale coded from 0 (very different) to 5 (very similar). To compute overall scores, we calculated the average score of all of the boy items and an average score of all of the girl items ($\alpha_{\text{boy}}=0.89$, $\alpha_{\text{girl}}=0.88$). We then reverse-scored the boy items, to have high scores across all gendered measures represent similarity/identity with girl/feminine.

Parent Measures

Parents were asked: (a) whether they were told that their child had a variation in sex development before birth; (b) what sex they were told their child would have before birth; and (c) what sex was listed on their child's birth certificate. Parents were further asked about (d) their child's current gender and (e) which pronouns their child used in everyday life (he/him, she/her, they/them, other; parents also had an opportunity to provide further clarification).

Parents reported demographic information (see Table 1) and completed a measure of their children's gender identity and expression that are reported in the Supplementary Material.

Statistical Analyses

All analyses were conducted in R (R Foundation for Statistical Computing, Vienna, Austria), using the tidyverse, rstatix, psych, knitr, kableExtra, and gridExtra packages (33,34,35,36,37,38,39,40). Not every participant answered every question. In tables, we display non-response (for categorical measures) or total n (for continuous measures). When aggregating across items and measures (e.g., calculating similarity to girls across the five questions, calculating the gendered composite), non-response is omitted from the calculation. Data cannot be shared publicly due to privacy concerns; many participants have rare medical conditions and would be identifiable with minimal or no demographic information provided. Data will be shared when requested by researchers with approval of the authors' IRB and requester IRB with agreement to approved privacy protections.

Results

Children's Gender-related Responses

Overall children with and without VSDs did not show many significant differences in their gender development. Among children raised as boys, those with and without VSDs differed significantly on only two of seven measures. Throughout this paper we refer to participants in our study with the descriptor "raised as boys" and "raised as girls". We use this term to indicate that this was the gender they were raised as throughout most of

their childhood. A few participants may be better characterized as having another gender at the time of testing (see Table 2), however results are reported by socialized gender because: (1) there are not enough nonbinary youth for separate analysis; and (2) past literature typically reports results by socialized gender, allowing for comparison across current and past results.

Boys with VSDs selected more masculine toys [$t(55.17)=2.413$; $p=0.019$; $d=0.623$], and responded in a more-masculine way on the continuous gender identity measure [$t(38.40)=2.364$; $p=0.023$; $d=0.621$], than did boys in the community comparison sample, although it should be noted that neither effect was statistically significant if a Bonferroni correction for the seven tests was applied. The two groups did not differ in their clothing preference, peer preference, similarity to boys, similarity to girls, or likelihood of picking “boy” in response to the categorical gender identity question (all $p>0.15$). The distributions of these responses can be seen in Figure 1. Tables 3 and 4 contain summaries of the distributions and statistical comparisons.

Among children raised as girls, those with and without VSDs differed significantly on only two of seven measures, and these differences held even when a Bonferroni correction for seven tests was applied. Girls with VSDs had more masculine toy preferences [$t(82.06)=3.538$; $p=0.001$; $d=0.722$], and felt more similar to boys [$t(73.30)=2.822$; $p=0.006$; $d=0.609$], than community comparison girls. The two groups did not differ on the continuous gender identity measure, clothing preference, peer preference, similarity to girls, or likelihood of picking “girl” in response to the categorical gender identity question (all $p>0.2$). Tables 3 and 4 contain summaries of the distributions and statistical comparisons.

The only specific VSD diagnosis for which we had a large enough group for separate analysis was girls with CAH. We compared this group to 28 age-and-gender matched community comparison girls. As with the overall comparison, girls with CAH had significantly more masculine toy preferences than the community comparison girls [$t(48.22)=4.314$, $p<0.001$, $d=1.153$]; the two groups did not differ significantly in clothing preference, peer preference, continuous gender identity, similarity to girls, or categorical gender identity (all $p>0.2$). Unlike the overall comparison, the difference for similarity to boys was not significant [$t(42.45)=1.327$, $p=0.192$, $d=0.383$]. Full results comparing girls with CAH to community comparison girls can be seen in the Supplementary Material (Table S1).

To show the general results by diagnosis type, we computed a composite of all gender-relevant child measures. To include the gender identity category measure, we treat “girl” as 1, “boy” as 0, and all other answers (“both,” “neither,” “it changes,” “I don’t know”) as 0.5. While these measures were selected because they represent a range of gender development constructs (e.g.,

identity, gender-typed preferences) that can be dissociable for individual children, past work suggests that these measures are often correlated (9). The seven measures formed a reliable composite ($\alpha=0.94$) in which 0 represents the responses most stereotypically associated with boys and 1 represents the responses most stereotypically associated with girls. In Figure 2, we show the level of this composite variable for participants

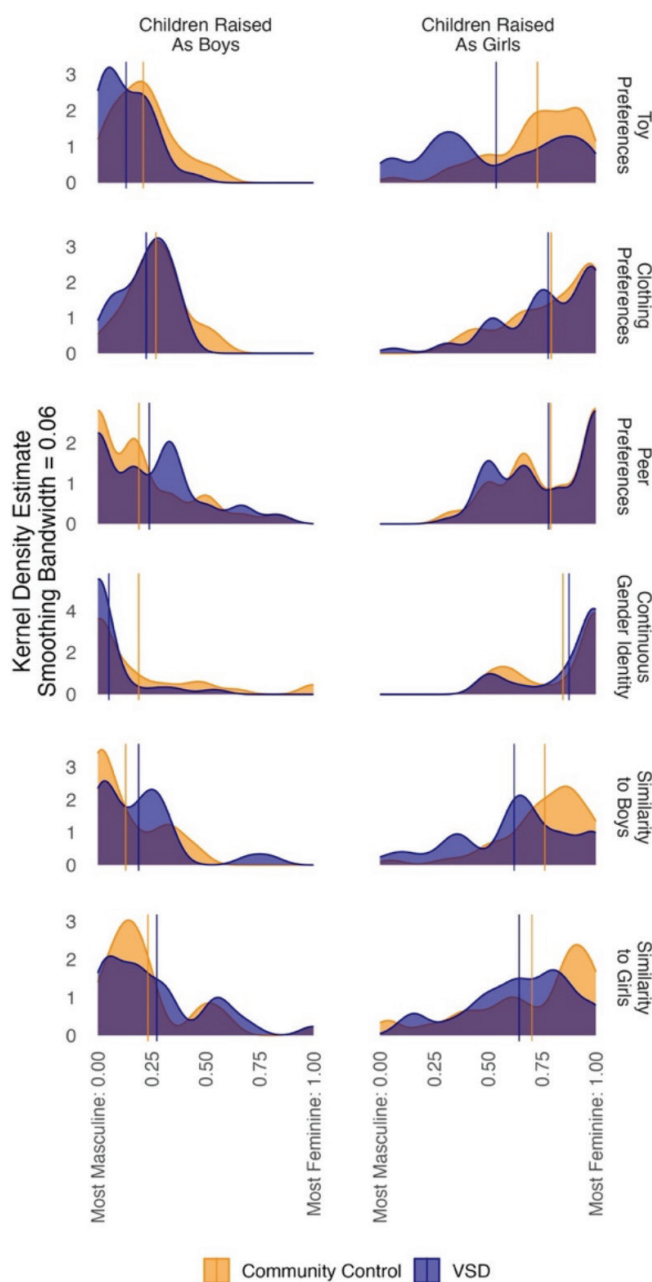


Figure 1. Distributions of responses on the primary gender measures. Each measure ranges from 0 (most masculine) to 1 (most feminine). Mean values for each group are indicated by vertical lines
VSD: variations in sex development

within each diagnostic category, including community comparison participants as their own subgroup. We present this data for exploratory purposes, rather than formal analyses, given the heterogeneity within groups and the small sample sizes.

Parent Report of Child History, Current Gender, and Current Pronouns

Among parents of children with VSDs, a chi-square test comparing those who said yes (17.95%) or no (74.36%) showed that parents were more likely not to have been told about their child's VSD prior to the child's birth [χ^2 (df=1)=26.889, $p<0.001$, $fei=0.61$, one-sided 95% confidence interval (0.42,1)]. The majority of parents in both groups reported that: (a) they were told their child's natal sex before their child's birth; (b) the natal sex reported on their child's birth certificate aligns with the gender in which the child

was raised; (c) their child's current gender is the same as the gender in which the child was raised and (d) their child's current pronouns reflect the gender in which the child was raised. Due to the infrequency of non-majority responses on these measures, we did not conduct statistical comparisons between the parents of children with VSDs and community comparison parents. Full responses can be seen in Table 2.

The results from the parent-reported gender measure converge with observations from the children themselves; full details are described in the Supplementary Material.

Discussion

Overall, we observed very few differences between a sample of children with a range of VSDs and children from a community

Table 3. Within-group means and standard deviations for continuous measures of gendered self-report

Measure	Raised-as boys					Raised-as girls				
	Children with VSD		Community comparison		t-test	Children with VSD		Community comparison		t-test
	N	M (SD)	N	M (SD)		N	M (SD)	N	M (SD)	
Toy preference	30	0.13 (0.11)	30	0.21 (0.14)	$t(55.17)=2.413$ $p=0.019$ $d=0.623$	48	0.54 (0.31)	48	0.73 (0.21)	$t(82.06)=3.538$ $p=0.001$ $d=0.722$
Clothing preference	30	0.22 (0.11)	30	0.27 (0.13)	$t(56.49)=1.420$ $p=0.161$ $d=0.367$	48	0.78 (0.22)	48	0.79 (0.20)	$t(93.07)=0.351$ $p=0.727$ $d=0.072$
Peer preference	30	0.24 (0.23)	29	0.19 (0.23)	$t(56.99)=-0.810$ $p=0.442$ $d=-0.211$	48	0.78 (0.22)	47	0.79 (0.21)	$t(92.99)=0.233$ $p=0.816$ $d=0.048$
Continuous gender identity	29	0.05 (0.13)	29	0.19 (0.29)	$t(38.40)=2.364$ $p=0.023$ $d=0.621$	48	0.88 (0.18)	48	0.85 (0.20)	$t(93.41)=-0.706$ $p=0.482$ $d=-0.144$
Similarity to boys	28	0.19 (0.20)	29	0.13 (0.15)	$t(50.90)=-1.282$ $p=0.206$ $d=-0.340$	41	0.62 (0.27)	47	0.76 (0.20)	$t(73.30)=2.822$ $p=0.006$ $d=0.609$
Similarity to girls	28	0.27 (0.25)	28	0.23 (0.23)	$t(53.41)=-0.636$ $p=0.527$ $d=-0.170$	41	0.64 (0.24)	48	0.70 (0.28)	$t(86.99)=1.059$ $p=0.293$ $d=0.224$

Each measure ranges from 0 (most stereotypically associated with boys) to 1 (most stereotypically associated with girls)
VSD: variations in sex development; N: sample size; M: mean; SD: standard deviation

Table 4. Categorical gender self-report

Response	Raised-as boys			Raised-as girls		
	Children with VSD	Community comparison	Chi-square	Children with VSD	Community comparison	Chi-square
Boy	93.33%	90.00%	χ^2 (df=1)<0.01 $p>0.9$ Cohen's $w<0.001$	2.08%	0.00%	χ^2 (df=1)<0.01 $p>0.9$ Cohen's $w<0.001$
Girl	0.00%	3.33%		85.42%	87.50%	
Both	0.00%	0.00%		2.08%	2.08%	
Neither	3.33%	0.00%		0.00%	0.00%	
It changes over time	0.00%	3.33%		4.17%	2.08%	
I don't know	3.33%	0.00%		4.17%	8.33%	
Non-response	0.00%	3.33%		2.08%	0.00%	

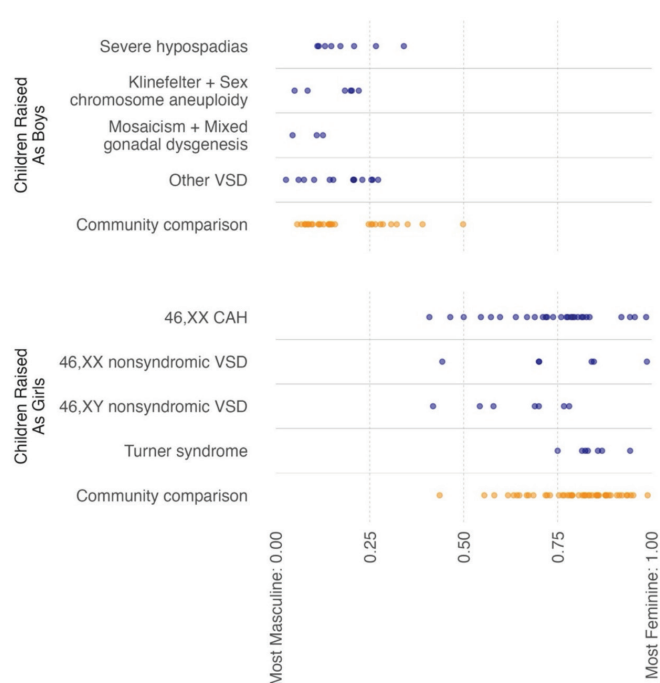


Figure 2. Composite gendered self-report within diagnostic subgroups. The vertical dashed lines show 0.25, 0.5, and 0.75 as reference points. Composite ranges from 0 (most masculine) to 1 (most feminine)
VSD: variations in sex development; CAH: congenital adrenal hyperplasia

sample in their gender development. Both groups showed patterns common in the literature for children who were raised as members of their gender group. We saw this pattern, that is a lack of significant difference between groups, across measures assessing distinct aspects of gender development, including gender identity (the categorical measure) and gender-typed preferences (e.g., clothing, peers).

A few differences emerged. Children raised as girls who had VSDs demonstrated more masculine toy preferences and indicated that they were more similar to boys on the similarity measure than the community sample of girls. The preference for more masculine toys amongst girls with VSDs, particularly those diagnosed with CAH, is consistent with past research (17,19,22). Boys with VSDs also had more masculine toy preferences than their matched community sample, and more masculine gender identities as indicated on a continuous measure of identity. These differences observed in the boys with VSDs are less well-documented, though some studies have noted similar findings, for example in a study of boys with hypospadias, the patients showed more masculine behavior than a comparison group (41). Given that we examined seven different dependent variables, and that a Bonferroni correction for that number of

tests would not yield significant differences amongst the boys, and given less past literature documenting these effects, we urge some caution in interpreting them. Future research is needed to better understand this association with VSDs and masculine preferences/identity. If the results continue to replicate, including in larger samples, we would have greater confidence in these results.

In general, the primary take-away message appears to be considerable similarity in gender development across children with and without VSDs. Notably, we observed that while girls and boys tended to generally show gender-stereotypic responses, there was a wide range, such that some girls provided strongly feminine responses, and others provided more gender-neutral responses. We observed this wide range in youth with and without VSDs.

As broader cultural understandings of gender become more widely accepted, it will be interesting to ask whether more youth with VSDs adopt these more expansive terms for their own genders, and if they do so at higher rates than other children. Adolescence is a time in which gender may be especially salient. We are eager to examine what happens as these youth enter adolescence, a time when more of these changes in identity seem to be emerging in broader society. It is important to track children's gender development over time and to recruit samples of adolescents to further understand cultural impacts on gender development amongst contemporary youth with VSDs. Anecdotally, at some of our clinics we have begun to meet some youth who are, for example, opting to use they/them pronouns or identify as nonbinary. Whether this is at higher rates than amongst their peers is currently unknown.

Many children in our sample were first identified as having VSDs within the first year of life. Therefore, it is difficult to determine the causes of the couple of small differences we observed between children with VSDs and the community comparison sample. For most of these youth, their caretakers were aware of their VSD-related differences, making a strict separation of biological and social contributors to gender development difficult to discern. Further, this idea of being able to separate biology and social experience is overly simplistic, as gender development is affected by multiple interacting influences including biological, socio-cultural, and individual factors (42).

Notably, toy preferences for youth with VSDs were different relative to the comparison group, with children raised as girls who had VSDs demonstrating more masculine toy preferences. Of note, our VSD sample had a large number of girls with CAH. Prior research has observed more masculine toy preferences among girls with CAH which is often attributed to early (prenatal) androgen exposure that occurs in the context of CAH (18,19,22).

While gender development and identity in the context of VSD is complex, there are strategies and approaches that clinicians can use and model for families which can support healthy development for children with VSD, including open communication about their condition, asking about how the child feels about who they are, and creating a supportive and accepting environment for discussion of gender and related exploration. Further, clinicians can also offer resources (e.g., connections to family support organizations, allies) and support when caregivers and other important individuals in the child's life encounter difficulties accepting or understanding complexities around gender identity (43).

Some VSDs are associated with higher risks for gender dysphoria [e.g., 5-alpha-reductase-2 deficiency; (44)], while other diagnoses are associated with little gender identity change or distress [e.g., Klinefelter syndrome; (45)], or have variable rates of gender diversity and/or identity change [e.g., CAH; (12,15,17)]. Healthcare providers and parents of children with VSDs sometimes grapple with decisions about whether to initially raise a child with a VSD as a boy, a girl, or in a more gender neutral or open way and whether the selected gender will align with the child's later gender identity. Deciding whether to initially raise a child as a boy, girl, or another gender can also have implications for potential surgical interventions to remove gonads and/or modify the child's genitalia, a controversial and debated issue in the context of VSDs (11).

While our data suggest that overall, children with VSDs are similar to their peers on indices of gender development, VSD care providers can normalize the existence of gender variations across all children and offer education and guidance about appropriate support and resources. Further, our findings also highlight the importance of interdisciplinary, coordinated care for children with VSDs that incorporates psychologists and/or other psychosocial providers who can tailor education about gender development in the context of an individual child's VSD and offer strategies to support healthy psychosocial functioning and adaptation (42). For example, clinicians can educate parents and children over time about gender identity and its relationship to their VSD diagnosis and to other important factors, such as socio-cultural and individual factors which can affect gender development (46).

Study Limitations

Our study has several strengths, including rigorous assessment of gender development in children with VSDs and inclusion of an age and sex-matched comparison group. The study also used more updated measures of gender, including a more continuous and less discrete assessment of gender identity. Nonetheless, our findings are potentially limited by the cross-sectional design, heterogeneity of the VSDs sample, and modest sample size. Our

study included a large age range from 3 to 12 years of age. While previous work has suggested that there are few developmental shifts on these types of measures in this age range (7), one still may wonder about that possibility in a VSD sample. The sample size was too small, especially within a single diagnostic category, to examine age differences. These are important topics for future work.

Our comparison group, while matched on age and gender, was not well-matched on parental education or socioeconomic status. Our most common finding, that the two groups did not differ on measures of gender, reduces this concern to some extent, but better matched groups would be useful in the future. Our findings underscore the need to evaluate gender development in youth with VSDs using larger samples and longitudinal designs that span the transition to adolescence and/or adulthood to further understand changes over time.

Conclusion

In this multi-site study of children with VSDs and an age-matched comparison group, our primary findings were similarities between groups with respect to gender development. We did observe some differences in toy preferences among youth with VSDs relative to the comparison group. Our findings have implications for clinical care, as these data may inform anticipatory guidance for families of children born with VSDs regarding what to expect about youth's gender during childhood. Additional longitudinal work is needed to better understand how gender development changes over time in children with DSD as they navigate adolescence and adulthood, as gender may be especially salient during these stages of development.

Ethics

Ethics Committee Approval: This study approved by the Institutional Review Board of Princeton University (approval no.: 12940, date: 25.07.2020).

Informed Consent: At least one parent had to provide informed consent, the child had to provide verbal assent, and the child had to complete the study assessment to be included in this analysis.

Footnotes

Authorship Contributions

Concept: Kristina R. Olson, Design: Kristina R. Olson, Data Collection or Processing: Canice E. Crerand, Margaret P. Adam, Maria G. Vogiatzi, Elizabeth McCauley, Jennifer Hansen-Moore, Margaret Shnorhavorian, Patricia Y. Fechner, Anne-Marie E. Amies Oelschlager, Justin A. Indyk, V. Rama Jayanthi, Hailey M. Umbaugh, Shira Kahn-Samuels, Grace Raber, Madeline McClinchie, Kristina R. Olson, Analysis or Interpretation: Canice E. Crerand, Natalie M. Gallagher, Kristina R. Olson, Literature Search: Canice E. Crerand, Jennifer Hansen-Moore, Kristina R. Olson, Writing: Canice E. Crerand, Natalie M. Gallagher, Margaret P. Adam, Maria G. Vogiatzi, Elizabeth McCauley, Jennifer Hansen-Moore,

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Conflict of Interest: Co-author Dr. Fechner is performing industry sponsored clinical research studies using new medications in the treatment of congenital adrenal hyperplasia with Diurnal, Neurocrine Bioscience and Spruce Bioscience. She serves as a consultant for Neurocrine Biosciences. Maria Vogiatzi is performing industry sponsored clinical research trials for new treatments in CAH with Spruce and Neurocrine Biosciences, Adrenas therapeutics and Crinetics. She serves as a consultant for Spruce Bioscience, Crinetics and Eton Pharmaceuticals. The other authors report no conflicts of interests.

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References

1. Cools M, Nordenström A, Robeva R, Hall J, Westerveld P, Flück C, Köhler B, Berra M, Springer A, Schweizer K, Pasterski V, COST Action BM1303 working group 1. Caring for individuals with a difference of sex development DSD: a Consensus Statement. *Nat Rev Endocrinol*. 2018;14:415-429. Epub 2018 May 16
2. Davies M. Raising Theybies: Navigating within a Gendered World. *SSRN Journal*. 2020.
3. Lin-Su K, Lekarev O, Poppas DP, Vogiatzi MG. Congenital adrenal hyperplasia patient perception of "disorders of sex development" nomenclature. *Int J Pediatr Endocrinol*. 2015;2015:1-7.
4. Conway GS. Differences in sex development DSD and related conditions: mechanisms, prevalences and changing practice. *Int J Impot Res*. 2023;35:46-50. Epub 2022 Sep 8
5. Sax L. How common is intersex? A response to Anne Fausto-Sterling. *J Sex Res*. 2002;39:174-178.
6. Tate CC, Youssef CP, Bettergarcia JN. Integrating the study of transgender spectrum and cisgender experiences of self-categorization from a personality perspective. *Rev Gen Psychol*. 2014;18:302-312.
7. Gülgöz S, Glazier JJ, Enright EA, Alonso DJ, Durwood LJ, Fast AA, Lowe R, Ji C, Heer J, Martin CL, Olson KR. Similarity in transgender and cisgender children's gender development. *Proc Natl Acad Sci U S A*. 2019;116:24480-24485. Epub 2019 Nov 18
8. Martin CL, Dinella LM. Congruence between gender stereotypes and activity preference in self-identified tomboys and non-tomboys. *Arch Sex Behav*. 2012;41:599-610.
9. Rae JR, Gülgöz S, Durwood L, DeMeules M, Lowe R, Lindquist G, Olson KR. Predicting early-childhood gender transitions. *Psychol Sci*. 2019;30:669-681. Epub 2019 Mar 29
10. Morris A. It's a Theyby! [Internet]. New York NY: The New Yorker; 2018 [updated 2018; cited 2025 May 27]. Available from: <https://www.thecut.com/2018/04/theybies-gender-creative-parenting.html>
11. Lee PA, Mazur T, Houk CP. DSD/intersex: historical context and current perspectives. *J Pediatr Endocrinol Metab*. 2023;36:234-241. Epub 2023 Jan 12
12. de Jesus LE, Costa EC, Dekermacher S. Gender dysphoria and XX congenital adrenal hyperplasia: how frequent is it? Is male-sex rearing a good idea? *J Pediatr Surg*. 2019;54:2421-2427. Epub 2019 Feb 22
13. Dessens AB, Slijper FM, Drop SL. Gender dysphoria and gender change in chromosomal females with congenital adrenal hyperplasia. *Arch Sex Behav*. 2005;34:389-397.
14. Berenbaum SA, Bailey JM. Effects on gender identity of prenatal androgens and genital appearance: evidence from girls with congenital adrenal hyperplasia. *J Clin Endocrinol Metab*. 2003;88:1102-1106.
15. Chowdhury TK, Laila K, Hutson JM, Banu T. Male gender identity in children with 46,XX DSD with congenital adrenal hyperplasia after delayed presentation in mid-childhood. *J Pediatr Surg*. 2015;50:2060-2062.
16. Pasterski V, Zucker KJ, Hindmarsh PC, Hughes IA, Acerini C, Spencer D, Neufeld S, Hines M. 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*. 2015;44:1363-1375. Epub 2014 Sep 20
17. Endendijk JJ, Beltz AM, McHale SM, Bryk K, Berenbaum SA. Linking prenatal androgens to gender-related attitudes, identity, and activities: evidence from girls with congenital adrenal hyperplasia. *Arch Sex Behav*. 2016;45:1807-1815. Epub 2016 Mar 3
18. Meyer-Bahlburg HF, Dolezal C, Baker SW, Carlson AD, Obeid JS, New MI. Prenatal androgenization affects gender-related behavior but not gender identity in 5-12-year-old girls with congenital adrenal hyperplasia. *Arch Sex Behav*. 2004;33:97-104.
19. Berenbaum SA, Hines M. Early androgens are related to childhood sex-typed toy preferences. *Psychol Sci*. 1992;3:203-206.
20. Nordenström A, Servin A, Bohlin G, Larsson A, Wedell A. Sex-typed toy play behavior correlates with the degree of prenatal androgen exposure assessed by CYP21 genotype in girls with congenital adrenal hyperplasia. *J Clin Endocrinol Metab*. 2002;87:5119-5124.
21. Berenbaum SA, Snyder E. Early hormonal influences on childhood sex-typed activity and playmate preferences: Implications for the development of sexual orientation. *Dev Psychol*. 1995;31:31-42.
22. 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*. 2011;59:549-555.
23. Brown A. About 5% of young adults in the US say their gender is different from their sex assigned at birth. *Pew Research Center* [Internet]. 2022 Jun 7 [cited 2025 May 27]. Available from: <https://www.pewresearch.org/short-reads/2022/06/07/about-5-of-young-adults-in-the-u-s-say-their-gender-is-different-from-their-sex-assigned-at-birth/>
24. Herman JL, Flores AR, O'Neill KK. How many adults and youth identify as transgender in the United States? [Internet]. Los Angeles CA: The Williams Institute, UCLA School of Law; 2022 [updated 2022; cited 2025 May 27]. Available from: <https://williamsinstitute.law.ucla.edu/wp-content/uploads/Trans-Pop-Update-Jun-2022.pdf>
25. Rider GN, McMorris BJ, Gower AL, Coleman E, Eisenberg ME. Health and care utilization of transgender and gender nonconforming youth: a population-based study. *Pediatrics*. 2018;141:e20171683.
26. Gates GJ. How many people are lesbian, gay, bisexual, and transgender? [Internet]. Los Angeles CA: The Williams Institute, UCLA School of Law; 2011 [updated 2011; cited 2025 May 27]. Available from: <https://williamsinstitute.law.ucla.edu/wp-content/uploads/How-Many-People-LGBT-Apr-2011.pdf>
27. Gülgöz S, Edwards DL, Olson KR. Between a boy and a girl: measuring gender identity on a continuum. *Soc Dev*. 2022;31:916-929.

28. Brenøe AA, Heursen L, Ranehill E, Weber RA. Continuous gender identity and economics. In: American Economic Association. 2022. p. 573-577.
29. Moore JK, Thomas CS, van Hall HW, Strauss P, Saunders LA, Harry M, Mahfouda S, Lawrence SJ, Zepf FD, Lin A. The Perth Gender Picture PGP: Young people's feedback about acceptability and usefulness of a new pictorial and narrative approach to gender identity assessment and exploration. *Int J Transgend Health*. 2021;22:337-348.
30. Reilly D. Gender can be a continuous variable, not just a categorical one: comment on Hyde, Bigler, Joel, Tate, and van Anders 2019. *Am Psychol*. 2019;74:840-841.
31. Fast AA, Olson KR. Gender development in transgender preschool children. *Child Dev*. 2018;89:620-637.
32. Martin CL, Andrews NC, England DE, Zosuls K, Ruble DN. A dual identity approach for conceptualizing and measuring children's gender identity. *Child Dev*. 2017;88:167-182. Epub 2016 Jun 1
33. Auguie B, Antonov A, Auguie MB. Package gridExtra: Miscellaneous functions for "grid" graphics. R Package Version. 2017 Sep 9;9.
34. Kassambara A. rstatix: Pipe-friendly framework for basic statistical tests. CRAN: contributed Packages. 2019 May 27.
35. Revelle W. Psych: Procedures for Psychological, Psychometric, and Personality Research. CRAN: Contributed Packages. 2007.
36. RStudio Team. RStudio: integrated development environment for R [Internet]. Posit Software; 2023. Available from: <http://www.posit.co/>
37. R Core Team. R: A language and environment for statistical computing [Internet]. Vienna AUT: R Foundation for Statistical Computing; 2023 [cited 2025 May 27]. Available from: <https://www.R-project.org/>
38. Wickham H, Averick M, Bryan J, Chang W, McGowan L, François R, Grolemond G, Hayes A, Henry L, Hester J, Kuhn M, Pedersen T, Miller E, Bache S, Müller K, Ooms J, Robinson D, Seidel D, Spinu V, Takahashi K, Vaughan D, Wilke D, Woo K, Yutani H. Welcome to the Tidyverse. *JOSS*. 2019;4:1686.
39. Xie Y. knitr: A general-purpose Tool for dynamic report generation in R. R. 2021.
40. Zhu H. kableExtra: Construct complex table with 'kable' and pipe syntax. 2021.
41. Sandberg DE, Meyer-Bahlburg HF, Yager TJ, Hensle TW, Levitt SB, Kogan SJ, Reda EF. Gender development in boys born with hypospadias. *Psychoneuroendocrinology*. 1995;20:693-709.
42. Sandberg DE, Gardner M. Differences/disorders of sex development: medical conditions at the intersection of sex and gender. *Annu Rev Clin Psychol*. 2022;18:201-231.
43. Kimball A, Rutter MM, Kavanaugh G, Fishler K. Exploring caregiver experiences of communicating with children about differences of sex development DSD. *Patient Educ Couns*. 2025;130:108470. Epub 2024 Dec 12
44. Cohen-Kettenis PT. Gender change in 46,XY persons with 5alpha-reductase-2 deficiency and 17beta-hydroxysteroid dehydrogenase-3 deficiency. *Arch Sex Behav*. 2005;34:399-410.
45. Kreukels BPC, Köhler B, Nordenström A, Roehle R, Thyen U, Bouvattier C, de Vries ALC, Cohen-Kettenis PT, dsd-LIFE group. Gender dysphoria and gender change in disorders of sex development/intersex conditions: results from the dsd-LIFE study. *J Sex Med*. 2018;15:777-785. Epub 2018 Mar 30
46. Hines M. Human gender development. *Neurosci Biobehav Rev*. 2020;118:89-96.
47. Veale JF. Factorial validity and invariance assessment of a short version of the recalled childhood gender identity/role questionnaire. *Arch Sex Behav*. 2016;45:537-550.
48. Zucker KJ, Mitchell JN, Bradley SJ, Tkachuk J, Cantor JM, Allin SM. The recalled childhood gender identity/gender role questionnaire: psychometric properties. *Sex Roles*. 2006;54:469-483.
49. Johnson LL, Bradley SJ, Birkenfeld-Adams AS, Kuksis MA, Maing DM, Mitchell JN, Zucker KJ. A parent-report gender identity questionnaire for children. *Arch Sex Behav*. 2004;33:105-116.