

Gender Identity and Preferences in Children with Variations in Sex Development

<https://orcid.org/0000-0003-0040-6396> Canice E. Crerand¹,
<https://orcid.org/0000-0000-0000-0000> Natalie M. Gallagher²,
<https://orcid.org/0000-0000-0000-0000> Margaret P. Adam^{3,4},
<https://orcid.org/0000-0000-0000-0000> Maria G. Vogiatzi⁵,
<https://orcid.org/0000-0000-0000-0000> Elizabeth McCauley^{3,4},
<https://orcid.org/0000-0000-0000-0000> Jennifer Hansen-Moore¹,
<https://orcid.org/0000-0000-0000-0000> Margaret Shnorhavorian^{3,4},
<https://orcid.org/0000-0000-0000-0000> Patricia Y. Fechner^{3,4},
<https://orcid.org/0000-0000-0000-0000> Anne-Marie E. Amies Oelschlager^{3,4},
<https://orcid.org/0000-0000-0000-0000> Justin A. Indyk¹,
<https://orcid.org/0000-0000-0000-0000> V. Rama Jayanthi¹,
<https://orcid.org/0000-0000-0000-0000> Hailey M. Umbaugh¹,
<https://orcid.org/0000-0000-0000-0000> Rachel Horton²,
<https://orcid.org/0000-0000-0000-0000> Shira Kahn-Samuelson²,
<https://orcid.org/0000-0000-0000-0000> Grace Raber²,
<https://orcid.org/0000-0000-0000-0000> Madeline McClinchie¹,
<https://orcid.org/0000-0000-0000-0000> Kristina R. Olson²

¹Nationwide Children's Hospital, Columbus, Ohio, United States

²Princeton University, Princeton, New Jersey, United States

³University of Washington, Seattle, Washington, United States

⁴Seattle Children's Hospital, Seattle, Washington, United States

⁵Children's Hospital of Philadelphia, Philadelphia Pennsylvania, United States

Corresponding author: Canice E. Crerand, Nationwide Children's Hospital, Columbus, Ohio, United States

E-mail: canice.crerand@nationwidechildrens.org

Received: 02.06.2025

Accepted: 06.10.2025

Epub: 29.11.2025

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 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 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 compared their gender development to that of a comparison 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 development¹ (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 (Davis, 2015). We use the term ‘variations in sex development’ and specific diagnosis names (Lin-Su et al., 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 variations in sex development range from 1.7 in 100 births (2) to 1.8 in 10,000 births (3) depending on the breadth of one’s definition and inclusion or exclusion of particular.

Parents of youth with VSDs often come to healthcare providers with questions about the likely course of their children’s gender development. The current research aims 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) (4). 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 (5-7).

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, (8, 9)). This decision can also have implications for potential surgical or endocrine interventions, a controversial and debated issue in the context of VSDs (10).

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 Congenital Adrenal Hyperplasia (CAH)² who are initially raised as girls sometimes show higher rates of gender dysphoria (11, 12) or identification as boys (13-15) than comparison groups, but other studies find no differences in gender identity (16, 17).² 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 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 (17-19). Other studies have shown a parallel ‘masculinity bias’ amongst girls with CAH in other domains: masculine-typed games (20), masculine playmates (21), and masculine-type clothing/dress-up (17). 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 (22-24). In contrast, as recently as 2011, estimates suggested numbers closer to 0.3% of the adult population identified as transgender (25).

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.

Materials and Methods

Participants and Procedures

Participants aged 3-12 with variations in sex development were recruited from 3 children’s hospitals: [Exact names withheld for anonymous review: Location 1] ($n = 44$), [Location 2] ($n = 22$), and [Location 3] ($n = 12$) 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 listed 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 1] and [University 2]. 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 5 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.

Child Measures

Categorical Gender Identity (5)

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 (26)

In line with more recent conceptualizations of gender identity as a non-discrete aspect of human experience (27-29), 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 (30)

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 2 children of the same gender, not used to

compute scores; pairs within a trial were matched for race: 6 pairs included White children, 1 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 & Clothing Preference (5, 30)

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 5 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 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 (31)

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 (32-39). Not every participant answered every question. In tables, we display non-response (for categorical measures) or total Ns (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,³ those with and without VSDs differed significantly on only two of seven measures.³ 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 (though note that neither effect is statistically significant if a Bonferroni correction for the 7 tests is applied). The two groups did not differ on their clothing preference, peer preference, similarity to boys, similarity to girls, or likelihood of picking “boy” in response to the categorical gender identity question ($ps > 0.15$). 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 hold even if a Bonferroni correction (for 7 tests) is 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 ($ps > 0.2$). 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 ($ps > 0.2$). 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 (7). 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 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 ($\chi^2(df = 1) = 26.889$, $p < 0.001$, $fe = 0.61$, one-sided 95% CI [0.42, 1]). The large 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. Because of 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 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 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 – 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 (16,18,21). 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 (e.g., boys with hypospadias showed more masculine behavior than a comparison group; (40)). Given that we examined 7 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 seems to be considerable similarity in gender development across children with and without VSDs. Notably, we observed that while girls and boys tended to show gender-stereotypic responses on average, 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 expansive, 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(41).

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 (17, 18, 21).

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 utilized 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 – children 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 (5), 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.

Conclusions

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. 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](#) (41). [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](#) (42).

[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; (11, 14, 16)). 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 (10). 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.

Supplementary Material

This supplement includes (1) additional methodological details, (2) an additional table for the comparison amongst CAH girls and girls without CAH, (3) additional information about the toy and clothing measure (including piloting details), and (4) information about a parental measure of children's gendered behavior and associated results.

Additional Methodological Details

Families were told about the possibility to complete the study before, after, or between their visits with medical care providers at one of the following types of clinics: differences in sex development (DSD) clinic, pediatric endocrinology clinic, CAH clinic, or Klinefelter clinic. To be included a child must have been judged by medical providers as having a VSD, the child had to be between 3 and 12 years of age and have enough English comprehension and production to complete the study. Children with severe intellectual impairments who would be unable to answer these questions and children who did not speak English were excluded from recruitment. Nearly all participants completed the research session during or immediately after a clinic visit, but one did so on Zoom later.

There are two pairs of siblings among the 78 participants with VSDs and one pair of siblings among the community comparison participants; these six individuals are treated independently for these analyses.

Participation for community controls occurred either in person at a research laboratory ($n = 39$) or online through Zoom ($n = 39$). Both university databases exist for the recruitment of child development research participants. Families often sign up to be included in the database when the children are born or later at a community event. They are told that researchers studying a range of topics recruit from these databases (e.g., language development, prosocial behavior, infant cognition). Parents were told they could make decisions later about which studies they would or would not participate in. All parents of these community participants were told that this was a study about gender development and agreed to have their child participate.

Assent involved hearing a short script describing the study (using different, developmentally appropriate language for 3–8-year-olds vs. 9–12-year-olds) indicating that participants could choose to participate or not, skip questions, and stop participation at any time. The child answered questions one-on-one with a researcher on a tablet or laptop, while parents completed a questionnaire on paper or on a tablet (typically in another room; two parents did not complete the questionnaires).

Toy and Clothing Preference Measures – Further information

There were different stimuli for children aged 3–7 years old and for children aged 8–12 years old because children at these ages play with different toys and wear different outfits. In addition, thirty-two youth (16 youth with VSDs and their 16 community comparison youth) completed these tasks with the stimuli from existing publications (5,30) that had originally been pilot-tested in 2013–2014. One hundred and twenty-four youth (62 youth with VSDs and their 62 community comparison youth) completed these tasks with the same structure, but with updated toy and clothing stimuli (updated as described below). We treat these two sets of stimuli as equivalent for analytic purposes; results hold when analyzing the results of only the 124 youth receiving the second set of stimuli.

Pilot testing of new items. Pilot testing of the exact stimuli used as part of the revised toy and clothing measure involved the following steps.

First, children between the ages of 3 and 12 (who were not part of the main study) were shown outfits or toys thought by study staff to be appropriate for their age and they were asked “Who would play with this toy?” or “Who would wear this outfit?”. Each child saw 25 clothing and 25 toy options, selected from a larger set of 100 total items of each type. They responded to each of the items with one of 5 selections: only boys, mostly boys, both boys and girls, mostly girls, and only girls. These were converted to scores from 1 (only boys) to 5 (only girls). The average score was then computed for each item. Using these scores, we created 4 sets of 5 toys that were considered strongly feminine, moderately feminine, neutral, moderately masculine, and strongly masculine.

In the second round of piloting, a new group of 21 7–11 year old children (also participants who were not in the main study) saw a set of 5 items that included one each of each rating (e.g., strongly feminine, moderately feminine, etc.) and were asked to select one item from each set to place into each of five categories: many more girls than boys wear/play with the item, slightly more girls than boys wear, play with the item, boys and girls equally wear/play with the item, slightly more boys than girls wear/play with the item, many more boys than girls wear/play with the item. Children were asked to answer based on what most kids would think. We used this second round of piloting to confirm that the selections from the first part of piloting were valid in representing these categories to a new group of participants. Further, we wanted to ensure that the sets of 5 items we placed together into a set did not somehow impact the relative perception of their masculinity/femininity. We found that children sorted the items as expected based on the initial ratings and therefore used these new item sets for the remainder of the study participants. We did not ask younger children to complete this secondary part of the piloting process because it seemed too challenging, but we did ask the 7–11 year olds to rate both the younger and older stimuli, assuming their opinions might be closer to those of younger children than adult raters (though research staff also confirmed that the sets seemed face valid as well).

Parent-Reported Youth Gender Identity and Experience

While not the focus of the primary analyses for the current paper, parents completed a measure of their perception of children's gender identity and expression. We provide that information here so as not to distract from the focus on children's own gendered behavior and preferences in the main paper.

We used two similar questionnaires asking parents about their children's gender-typed preferences, behaviors, femininity/masculinity, interests, and feelings about their body, etc. Each item was scored from 0–1 where 1 indicates either the response most stereotypically aligned with the gender in which the child was raised, or the child's highest possible satisfaction with their gender and body (0 represents the response least stereotypically aligned with the child's gender-of-raising, or the child's lowest possible satisfaction with their gender and body). Responses indicating that the child does not participate in an activity of a particular kind are omitted. We use the average score across items and treat the two scales as equivalent for research purposes ($\alpha_{RCGIE-GS} = 0.85$; $\alpha_{GIQ} = 0.66$).

Most parents answered the Recalled Childhood Gender Identity and Experience-Gender Spectrum-Parent Version (RCGIE-GS), a 32-item measure (46) adapted from (47) (we further adapted this measure to ask parents about their youth in the present tense; 62 parents of youth with a VSD were invited to complete this measure). A smaller number of parents completed an 18-item version of the parent-report Gender Identity Questionnaire for Children (GIQ; (48); we adapted this measure so all parents answered two boy-specific and two girl-specific items; 16 parents of children with VSDs were invited to complete this measure). We switched from (48) to (46) because it uses more contemporary language; however, questionnaire version was matched between youth with VSDs and their community control youth.

Results involving the RCGIE-GIS/GIQ

Parents of youth with VSDs and community comparison parents did not differ in their ratings of their child's gender typicality on the RCGIE-GS/GIQ ($t(145.31) = 0.764$, $p = 0.446$, $d = 0.124$; $N_{\text{VSD}} = 75$, $M_{\text{VSD}} = 0.75$, $SD_{\text{VSD}} = 0.11$; $N_{\text{Community}} = 77$, $M_{\text{Community}} = 0.76$, $SD_{\text{Community}} = 0.09$). To evaluate the relationship between parent and child report, we reverse-scored the child-report composite for children raised as boys, so that higher scores represented greater gender typicality in child behavior in the experimental tasks. Parent reports of children's gendered behavior on the RCGIE/GIQ were positively correlated with the gender-typicality composite ($r(150) = 0.51$, $p < 0.001$).

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Gender-Of-Raising	Diagnosis	Sample Size	Categorization
Boy	Hypospadias	6	Severe hypospadias
	Hypospadias with 87% 46,XY and 13% sex chromosome aneuploidy consisting of XXY	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	
	17 α -hydroxylase deficiency (46,XY)	2	Other VSD
	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,YYY	1	
	Ovotesticular DSD	1	
	Ovotesticular DSD secondary to chimerism	1	
	Vanishing testes syndrome	1	
Girl	46,XX male	1	46,XX CAH
	21-hydroxylase deficiency	18	
	17 α -hydroxylase deficiency (46,XX)	7	
	11-beta-hydroxylase deficiency	3	46,XX nonsyndromic VSD
	46,XX DSD due to a heterozygous pathogenic variant of <i>WT1</i> (gain-of-function)	1	
	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	
	CAIS	2	46,XY nonsyndromic VSD
	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	

Note. Categorization applies to presentation of data in Figure 2.

		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%
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%

Table 3. Within-Group Means and Standard Deviations for Continuous Measures of Gendered Self-Report										
	Raised-as Boys					Raised-as Girls				
	Children with VSD		Community Comparison		t-test	Children with VSD		Community Comparison		t-test
Measure	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)	$t(86.99) = 1.059$ $p = 0.293$ $d = 0.224$
Note. Each measure ranges from 0 (most stereotypically associated with boys) to 1 (most stereotypically associated with girls).										

Table 4. Categorical Gender Self-Report						
	Raised-as Boys			Raised-as Girls		
Response	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%	

Table S1. Within-Group Means for Measures of Gendered Self-Report between CAH Girls and Comparison Participants.					
Measure	Children with VSD		Community Comparison		Raised-as Girls
	N	M (SD)	N	M (SD)	t-test
Toy Preference	28	0.44 (0.31)	28	0.75 (0.21)	$t(48.22) = 4.314$ $p < 0.001$ $d = 1.153$
Clothing Preference	28	0.76 (0.20)	28	0.82 (0.19)	$t(52.76) = 1.176$ $p = 0.245$ $d = 0.314$
Peer Preference	28	0.78 (0.20)	28	0.76 (0.22)	$t(53.09) = -0.506$ $p = 0.615$ $d = -0.135$
Continuous Gender Identity	28	0.92 (0.14)	28	0.89 (0.18)	$t(51.71) = -0.687$ $p = 0.495$ $d = -0.184$
Similarity to Boys	22	0.65 (0.27)	27	0.74 (0.24)	$t(42.45) = 1.327$ $p = 0.192$ $d = 0.383$
Similarity to Girls	22	0.69 (0.23)	28	0.68 (0.31)	$t(47.76) = -0.215$ $p = 0.831$ $d = -0.060$
Note. Each measure ranges from 0 (most stereotypically associated with boys) to 1 (most stereotypically associated with girls).					

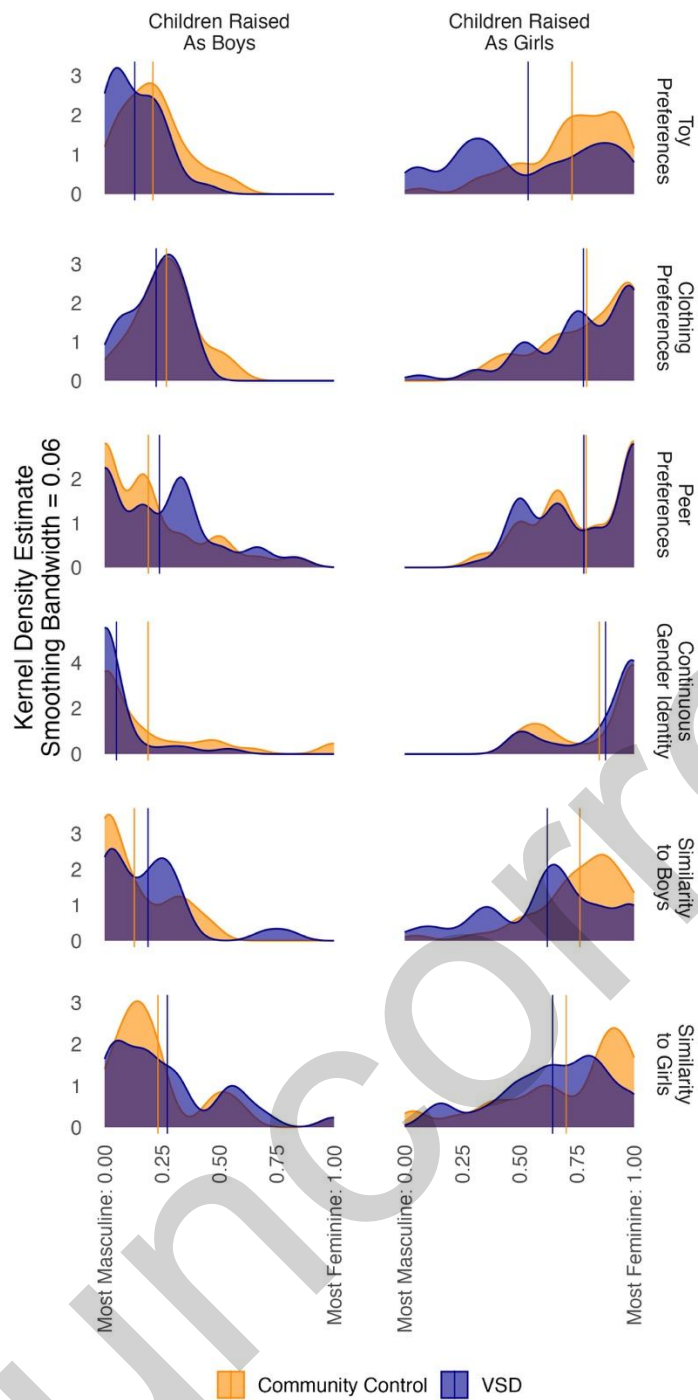


Figure 1. Distributions of Responses on the Primary Gender Measures
Figure 1 Legend. Each measure ranges from 0 (most masculine) to 1 (most feminine). Mean values for each group are indicated by vertical lines.

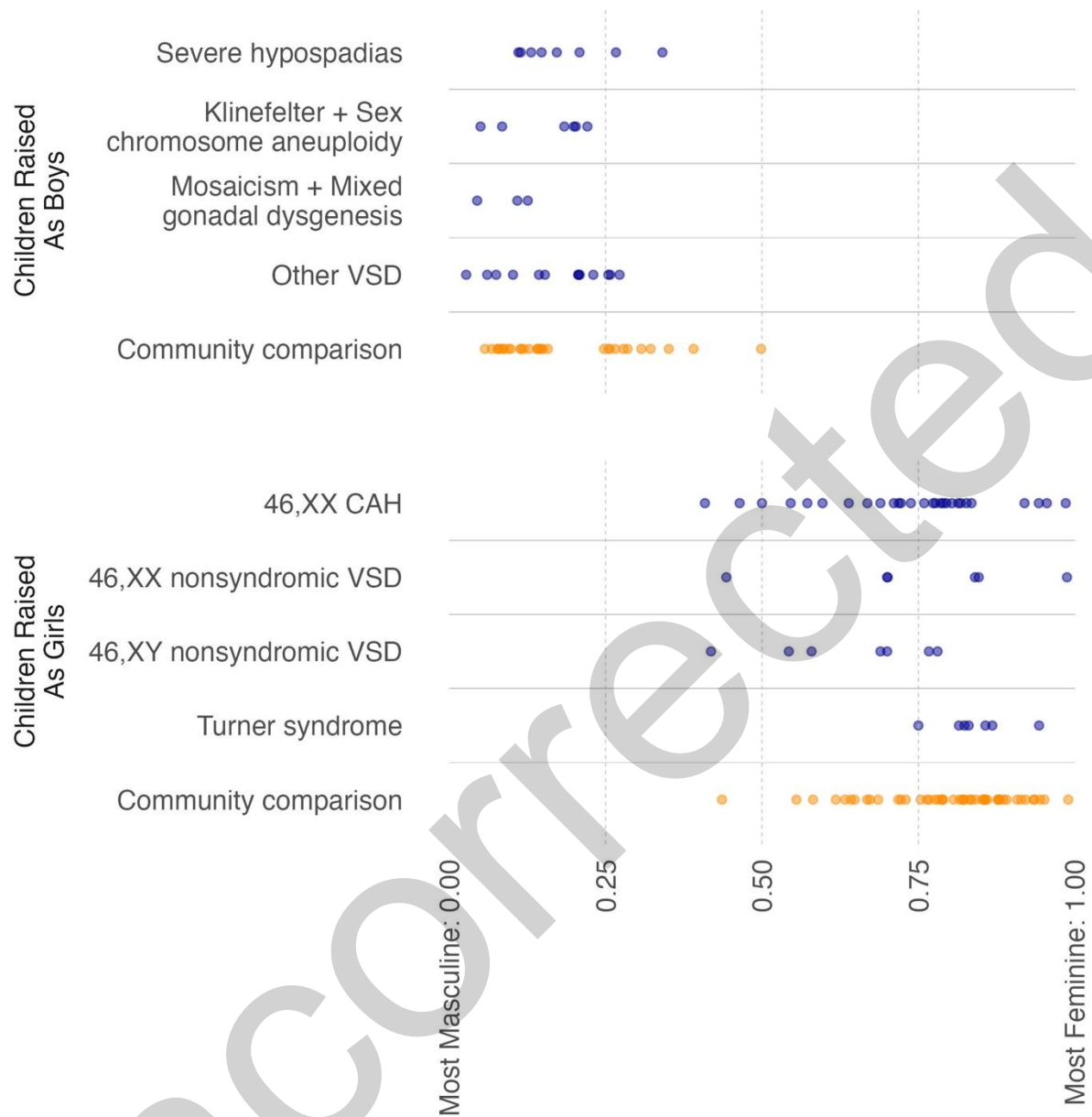


Figure 2. Composite Gendered Self-Report Within Diagnostic Subgroups
Figure 2 Legend. The vertical dashed lines show 0.25, 0.5, and 0.75 as reference points. Composite ranges from 0 (most masculine) to 1 (most feminine).