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Identifying and Counting Individuals with Differences of Sex Development Conditions in Population Health Research

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Abstract

Purpose: The study purpose was to examine opinions about a single-item assessment of differences of sex development (DSD) to be used in research.
Methods: An online survey was conducted with a convenience sample of 111 adults who self-identified as intersex or having a DSD diagnosis. Participants read and provided feedback on the proposed single-item assessment.
Results: The item received general endorsement to represent a population that is often not identified in research; however, participants provided suggestions for improvement.
Conclusion: This study represents a first step toward identifying people with DSD conditions in surveys to better understand their needs.

Keywords: differences of sex development, health, intersex, survey research

Introduction

"Differences of sex development" (DSD) represent a group of over 20 congenital conditions involving the development of gonadal, chromosomal, or sexual anatomical characteristics that do not conform to the traditional female-male sex binary.[1] DSD is an umbrella term which includes those with intersex conditions and does not necessarily signify ambiguity in genital appearance or uncertainty about gender of rearing.[2,3] Intersex and other DSD conditions are distinct from gender identity and sexual orientation/identity.[1] No known population studies collect information on individuals with DSD conditions. Although some people state their gender identity as intersex when given a write-in option on surveys, there is a dearth of information from general community surveys or research using nonclinic-based samples on the subjective experiences of health among people with DSD conditions. Due to the lack of population data and variable definitions of DSD conditions, the estimated incidence of individuals with DSD conditions varies across studies (e.g., from as low as one case of androgen insensitivity syndrome out of 20,400 over seven years in Denmark[4] to an estimated population incidence of 2% for individuals with nondimorphic chromosomal, gonadal, hormonal, or genital characteristics[5]). Lack of data from population studies also limits the identification of general health and psychosocial concerns among individuals with DSD conditions.

Advocacy groups have played a critical role in describing some of the psychological challenges and physical traumas experienced by people with DSD conditions due to stigma and problems experienced in medical care (e.g., irreversible genital surgeries and gonadectomies to reinforce gender assignment and repeated genital examinations).[6] Advocacy groups have urged the medical community to ask critical questions about appropriate standards of care for surgeries related to DSD conditions.[6] Advocacy groups have also brought attention to relevant questions such as whether and when surgeries are medically necessary versus elective, which types of surgeries are considered medically necessary, at what age should surgeries be done, who has the right to consent to these procedures, and related ethical issues.[1] However, standards of care have been difficult to achieve, in part, due to the heterogeneity of DSD and dearth of useful and rigorous studies with people with DSD conditions.[1]

Although there has been increasing interest in the health and well-being of people with DSD conditions, as well as

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some consideration of their inclusion in research with sexual and gender minority communities, scientists and scholars often exclude these individuals from research efforts.[7,8] This exclusion may be due, in part, to challenges in assessing, using self-report methods, whether people reliably report having a DSD (including intersex conditions) as opposed to identifying as intersex (i.e., report intersex as their gender identity in the absence of a confirmed DSD). In 2014, the Gender Identity in U.S. Surveillance (GenIUSS) group convened by The Williams Institute identified three major issues that have hindered the inclusion of people with DSD conditions in research efforts aimed at advancing the health of sexual and gender minority communities.[8] First, some may not endorse the term ''intersex'' as a type of identity *per se* (e.g., akin to gender identity or sexual orientation identity), but instead may consider their DSD condition to be a diagnosis without carrying implications for personal identity. Second, ''intersex'' is sometimes used as an identity among people who do not have DSD conditions. Third, with few exceptions, ''intersex'' is not an option on birth certificates.[8] In addition, intersex conditions are a subset of DSD, and experts disagree on how to define the subset.[3]

In consideration of these issues, the GenIUSS group recommended a single item to identify people who may have a DSD condition, which can be used in population surveys and community research studies outside the context of clinic-based samples.[8] However, researchers have yet to obtain input on the recommended item from these community members. As such, the purpose of this study was to gather input on the recommended item, as well as suggestions for improvement, from community members with a diagnosed DSD condition or who identify as intersex. We were especially interested in the perspectives of a nonclinic-based sample, as this item could be particularly useful in research involving community samples or in population surveys. This study represents an initial step in examining community perspectives on this single-item measure, which, if implemented broadly in population and community research, could improve knowledge about the size, demographic characteristics, and general health and psychosocial concerns of individuals with a diagnosed DSD condition or who identify as intersex.

Methods

Participants and procedures

This study involved a community-based participatory research partnership between University researchers and interACT, an advocacy organization that promotes the rights of children born with DSD conditions or born with intersex traits. The overarching goal of this partnership was to identify the needs of people with DSD conditions or who identify as intersex even in the absence of a DSD condition. Between December 2016 and April 2017, we conducted an online anonymous survey with adults who identified as having a DSD condition or identified as intersex. Individuals were eligible for this study if they: (1) were 18 years of age or older, (2) identified as intersex or as having a diagnosis of DSD, and (3) consented to participate in the study.

Participants were recruited using targeted sampling through outreach by Facebook advertisements and posts to community intersex and DSD forums. The recruitment post stated: ''Seeking people who identify as intersex or have a diagnosis of DSD to complete a brief 10–12 minute anonymous survey.'' In total, 201 potential participants clicked on the survey link. The first screen of the survey link provided a description of the study and a screener question that assessed age followed by a screen providing informed consent information and the option for consenting to participate. Of those who clicked on the survey link, 5 respondents were under the age of 18, and 14 respondents did not indicate their age, rendering them ineligible to continue. Thirteen additional respondents did not consent to participate. Of those who consented, 55 respondents completed very little or no information and were subsequently excluded from the analysis. Of the 114 individuals remaining, 3 did not identify as having a DSD condition or identify as intersex and were excluded, which resulted in a sample of 111 participants who met the inclusion criteria.

Participation involved an Internet-administered survey which took $\sim$20–30 minutes. Participants were asked demographic questions about age, sex assigned at birth, gender identity (open-ended item), race/ethnicity, education, and income levels, and whether they lived in a rural, urban, or suburban location. Participants were also asked about their DSD condition in an open-ended format. Participants read and responded to the single-item measure developed by the GenIUSS group[8] which asked: ''Have you ever been diagnosed by a medical doctor with an intersex condition or a 'Difference of Sex Development (DSD)' or were you born with (or developed naturally in puberty) genitals, reproductive organs, and/or chromosomal patterns that do not fit standard definitions of male or female?'' (response options were *Yes*, *No*, or *I don't know*). Then they completed open-ended questions asking for their opinions about the single-item measure and suggestions for improving assessment of individuals with DSD conditions in survey research. Respondents were not compensated, but received information about community resources and websites related to DSD after completing the survey. The Institutional Review Board of Brown University approved all research protocols.

Analyses

First, we conducted univariate analyses to characterize the study sample. Next, we used a framework analysis for the open-ended responses, which is particularly well suited to studies that attempt to answer a focused set of questions. Consistent with the steps outlined in framework analysis, the second and third authors began by familiarizing themselves with the responses. Second, the second and third authors devised and refined a thematic framework for coding by reading the data, identifying the themes that emerged, and writing analytic memos about those themes. During a series of meetings, the second, third, and senior authors read and re-read the data, discussed themes with the other authors, and wrote analytic memos about those themes. The second and third authors indexed the data, identifying specific sections, which corresponded with our themes. All analyses were double coded, and reliability was assessed using Cohen's kappa ($\kappa = 0.92$). Discrepancies between the second and third authors' codes were resolved with the senior author.

Results

Participants ranged in age from 19 to 74 ($M = 37.05$, $SD = 14.04$). The sample identified predominantly as White

(83.8%, $n=93$) with 9.9% ($n=11$) identifying as Hispanic/Latinx, 3.6% ($n=4$) identifying as Black/African American, 1.8% ($n=2$) identifying as multiracial, and 0.9% ($n=1$) identifying as Asian. The sample was relatively well educated, with 36.9% reporting some graduate school or a graduate degree, 27% reporting a Bachelor's degree, and 35.1% reporting less than a Bachelor's degree. With regard to income, 25.2% of participants reported an income of less than $20,000 per year, 27.9% reported an income between $20,000 and $49,999, 19.8% reported an income between $50,000 and $79,999, 19.8% reported an income of $80,000 or more, and 7.3% of the sample did not respond to this question. The majority of the sample resided in an urban (43.0%) or suburban (37.6%) community, with 11.8% reporting residing in a rural community; however, 7.6% of the sample did not respond to this question.

Nearly three-quarters of the sample (72.1%) reported being assigned female sex at birth. There was substantial variability in how participants described their gender identity. In response to the open-ended gender identity question, 36.0% identified as female, 9.9% identified as male, 32.4% identified as non-binary/gender fluid, 18.0% identified as intersex (without reference to male or female gender categories), and 3.6% identified as some other gender identity. In total, 63.1% of respondents had different gender identities than the sex assigned to them at birth; only 36.9% had the same gender identity as their sex assigned at birth.

Almost all participants (96.4%) reported having a diagnosed DSD condition; an additional 2.7% reported not having an ''official'' diagnosis but described intersex-like characteristics, and 0.9% reported not knowing if they have a diagnosis but described intersex-like characteristics. Regarding diagnosis, 41.5% of the sample reported having a form of Androgen Insensitivity Syndrome—Complete Androgen Insensitivity Syndrome (17.1%), Partial Androgen Insensitivity Syndrome (15.3%), or didn't specify (9.1%). Other diagnoses reported included, in order of incidence, the following: Klinefelter syndrome (8.1%), Swyer syndrome (8.1%), Gonadal Dysgenesis (7.2%), Congenital Adrenal Hyperplasia (6.3%), Mayer–Rokitansky–Küster–Hauser syndrome (3.6%), Chimerism (3.6%), Cloacal Exstrophy (1.8%), Hypospadias (1.8%), Mosaicism (1.8%), and Bladder Exstrophy (0.9%). For 14.4% of respondents, their specific diagnosis was unclear, and 1 respondent (0.9%) did not answer this question.

The majority ($n=80$, 72%) of participants felt that the single item recommended by the GenIUSS group to capture people with DSD conditions was important and straightforward, and they wanted the item to be included in survey research.

I think it's an important question. We need to be heard.

I think it is a great way to establish what the intersex community is comprised of, and brings awareness of natural born non-binary existence.

I like it. Intersex folks are frequently an excluded demographic by default simply because researchers don't provide the opportunity for us to make ourselves and our bodies known.

Although participants generally understood the importance of including this item in survey research, some also felt that the item was too medicalizing and may not include people who have not had access to care to receive a diagnosis.

This question strikes me as indicative of the general academic and medical understanding among people sympathetic to intersex people. While it is clearly well meaning, the definition of 'intersex' seems a little narrow, and the language is medicalizing and a little uncomfortable; but I understand its importance.

I think it is definitely helpful but also assumes the participants have had access to care or procedures necessary to make a diagnosis.

I think many if not most people born intersex are shoved into one category through all medical history. I think it would be helpful to add that you yourself are questioning your sexual and reproductive organs being that of a binary so there is room for those without medical support to have a voice.

When asked for recommendations to improve this measure, participants suggested several potential alternatives. Five participants suggested having a list of different conditions. Several participants suggested that items included in surveys that assess an individuals' sex assigned at birth could include a third response option (e.g., male, female, and intersex). One participant suggested that the third option on the item be ''undetermined.''

Instead of having two questions, just have one that asks what sex a person was assigned at birth and three options.

Although the word ''disorder'' was not used in the single-item measure, many voiced their general aversion to the word ''disorder'' that constitutes or is implicit in the DSD acronym (the survey item used the expression ''Difference of Sex Development'') One participant suggested that researchers refrain from using the term DSD.

A little intro that talks about language in a way that won't turn off people who hate to see 'DSD' and won't even fill out survey if they see it (at same time in a way that 'intersex' doesn't scare off another group of patients).

Discussion

Greater efforts are needed to identify and count individuals with DSD conditions and who identify as intersex in population health research and to distinguish those who identify as intersex in the absence of a documented DSD. The overwhelming majority (96.4%) of our sample reported having a diagnosed DSD condition, whereas only 18% endorsed intersex as a gender identity. Consistent with historical gender assignment practices, 72% of the sample reported being assigned female sex at birth.[1] The extent to which these statistics reflect our recruitment methods remains to be determined. Advocacy groups have called for the need to develop standards of care in surgical procedures related to DSD conditions as noted in the recent Human Rights Watch report.[6] Our findings generally support the use of a single-item research tool developed by the GenIUSS group that can be included in survey research to begin to advance these efforts.[8]

Participants unanimously wanted an item that allowed them to be included in survey research and generally endorsed the single-item GenIUSS assessment.[8] Participants described the importance of using items in representative national surveys, such as the one developed by the GenIUSS group, as a step toward better characterizing the sociodemographic and health characteristics of people with DSD conditions and/or who identify as intersex outside the context of clinical research. In addition, research using Medicare and Medicaid databases has the potential to compare demographic and health characteristics of people with and without DSD conditions, although these databases include primarily older citizens, the poor, and the young. To our knowledge, no such research involving people with DSD conditions using Medicare or Medicaid databases has been conducted.

Recommendations included expanding the measure to assess DSD conditions. Participants' feedback on the measure highlighted the distinction between using the terminology ''DSD'' versus ''intersex'' and noted that some individuals will not respond to an item if they see the acronym ''DSD'' given the historical use of the term ''disorder.'' Participants also recommended the use of clear language and explanation of terminology when introducing the assessment item to survey respondents—clarifying that DSD refers to ''differences'' of sex development, not ''disorders.'' In addition, people with a DSD condition may or may not identify as intersex, and some individuals identify as intersex in the absence of a DSD condition. Therefore, these categories do not necessarily align. Furthermore, the health needs of individuals with DSD conditions differ depending on the nature of their particular DSD. For example, depending on the particular DSD condition and/or treatment history, individuals with DSD conditions may have an ongoing need for particular medical treatments, such as hormone replacement and cancer screenings.[9] Thus, additional research is warranted to develop measures that are responsive to the diverse needs of individuals with DSD conditions, as well as those who identify as intersex in the absence of a DSD condition.

Limitations

There are limitations to the study, including the nonprobability convenience sample of predominantly White and well-educated adults. Our recruitment strategy also relied on recruiting participants from online community forums who may not be representative of individuals with DSD conditions who are included in studies using clinic samples. Thus, future research using more rigorous recruitment methods is warranted to obtain opinions from a more representative sample of individuals with DSD conditions. The Internet-based recruitment strategy was useful in accessing this difficult-to-reach and diverse nonclinical community sample of people with DSD conditions and/or who identify as intersex, but the sample might be biased based on the specific forms of Internet outreach (e.g., at intersex forums). Future research is warranted using more rigorous recruitment strategies (e.g., chain-referral sampling targeting multiple recruitment venues[10]) to obtain the opinion of individuals who do not incorporate a DSD condition or intersex into their identity. This study also relied on self-report data, which may be subject to social desirability bias and inaccurate responses.

Conclusion

A more inclusive field of sexual and gender minority health relies on having better data on the prevalence, characteristics, and health concerns of people with DSD conditions and those who identify as intersex with or without a medically verified condition. This study represents a first step in promoting the use of the item developed by the GenIUSS group to identify individuals diagnosed with an intersex or DSD condition in survey research. Our study findings suggest that multiple items may be necessary to accurately capture the diversity of individuals with DSD conditions and/or who identify as intersex and to disaggregate this diverse sample into subgroups for meaningful analyses. In addition to the measure developed by the GenIUSS group, researchers could include an item that lists specific diagnoses with a write-in option, as well as check boxes for particular aspects of anatomy, surgical history, and medications, which have the potential to distinguish individuals with a DSD condition and those who identify as intersex. Use of single or multi-item measures in population surveys and in studies related to individuals with DSD conditions and those who identify as intersex can advance knowledge about the health and well-being of these groups, including advancing efforts toward developing standards of care for surgical procedures with these communities. Future research efforts using more rigorous recruitment strategies are warranted to validate the item put forth by the GenIUSS group and to develop more nuanced measures to assess the diversity of individuals with DSD conditions and/or who identify as intersex, who are not currently counted in survey research.

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Author Disclosure Statement

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