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Article

Capturing Gender Diversity in Twin Research

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Abstract

Most twin registries have not systematically collected the data required to determine gender identity, which has limited opportunities to evaluate potential familial contributors to gender diversity. This study addresses this gap by analyzing responses to gender identity questions introduced in Twins Research Australia's 2023 survey. Among 4475 respondents (mean age 52.2 years, $SD = 15.3$), 36 (0.8%) indicated a transgender or gender diverse identity, which is consistent with population-based estimates of gender diversity internationally. Gender diversity co-occurred in 2/19 monozygotic pairs and 0/8 dizygotic pairs, giving rise to tetrachoric correlations of 0.62 (95% CI [0.33, 0.87]) and 0.00 (95% CI [0.00, 0.88]), respectively. These results broadly align with previous concordance estimates from twin studies that were specifically focused on gender identity. Although limited by a small sample size, these findings demonstrate the feasibility and utility of systematically collecting gender identity data through routine twin registry surveys.

Keywords: Twin research; Transgender; Gender diversity; Twin registries; Gender identity

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'Gender diverse' is a term that describes people whose gender differs from what was assigned to them at birth (Coleman et al., 2022; Thorne et al., 2019; see Text Box 1 for definitions of relevant terms). Gender diverse people have become increasingly visible in recent years, prompting growing societal awareness and debate. There are some published data suggesting a genetic contribution to gender diversity (Polderman et al., 2018), but the relative contribution of genetic and environmental factors remains unclear.

Twin registries have long provided a powerful avenue for exploring the relative contributions of genetic and environmental factors to a range of complex human traits. However, twin registries have seldom contributed insights into gender diversity, mostly because details about sex and gender are not systematically separated, making it impossible to accurately identify transgender participants.

Several non-registry-based twin studies have provided heritability estimates for gender diversity (Bailey et al., 2000; Burri et al., 2011; Diamond, 2013; Heylens et al., 2012; Karamanis et al., 2022; Polderman et al., 2018; Sasaki et al., 2016). Most have findings suggesting a genetic influence; however, they all have methodological limitations. For instance, ascertainment bias in published summaries of prior case reports reflecting past preferential reporting of concordant twins (Diamond, 2013; Heylens et al., 2012). A gender-specific issue is the use of childhood gender-related behavior (Sasaki et al., 2016) or previous access to gender-affirming interventions (Karamanis et al., 2022) as imprecise proxy surrogates for gender identity.

National recommendations for collecting basic demographic information now include comprehensive gender data, achieved by a simple two-step approach that allows the self-reporting of both birth-assigned sex and gender identity (Australian Bureau of Statistics, 2020). Incorporating this into twin registries should be relatively straightforward. If this were to happen, some of the biases in past twin studies of gender diversity would be largely overcome. For example, self-reported gender identity would overcome the imprecision of proxy indicators and general twin surveys would be less prone to ascertainment bias. In this way, twin registry-based studies implementing the now recommended two-step collection of gender data are enabled to support transgender health research, particularly to advance knowledge of genetic and environmental influences.

Responding to the above considerations, Twins Research Australia (TRA), a national twin research institute headquartered at the University of Melbourne (Murphy et al., 2019), introduced the two-step approach to their 2023 survey. We describe here the data obtained from these questions and demonstrate some of their potential utility.

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Text box 1. Terminology*

Term	Definition (adapted from American Psychological Association, 2015)
Assigned male/ female at birth	Sex assigned at birth due to appearance of external genitalia.
Cisgender	Individuals who have a congruent sex assigned at birth and gender identity; a person who is not gender diverse.
Gender diverse	Umbrella term that describes people whose gender differs from what was presumed/assigned at birth. This includes transgender individuals as well as those with a non-binary identity.
Nonbinary	Individual whose gender is not exclusively male/ man or female/woman. This includes those who identify as genderqueer or gender fluid.
Transgender	Like 'gender diverse', this is an umbrella term that describes people whose gender differs from what was assigned at birth. However, 'transgender' can be more restrictive than 'gender diverse'; for example, some individuals who identify as non-binary do not identify as transgender, which can align with more binary notions of gender.
Transgender man	Individuals assigned female at birth, who have a male gender identity.
Transgender woman	Individuals assigned male at birth, who have a female gender identity.

Note: * Terminology in this area continues to evolve. For example, in the past, the terms such as 'transexual', 'male-to-female (MTF)', and 'female-to-male (FTM)' were commonly used but these are no longer considered acceptable.

Methods

Participants were recruited through the 'Twins Health Behaviours and Screening Questionnaire (HBQ)', which comprised general questions about healthcare service usage and screening behaviour for the national cancer screening programs (colorectal, breast and cervical cancer). The 2023 survey incorporated for the first time questions on gender identity, which comprised the two-step approach sourced from the Australian Bureau of Statistics (2020).

1. At birth you were recorded as:
Male
Female
Another term (please specify) — free text box available.
Prefer not to answer

2. How do you describe your gender?
Man or male
Woman or female
Non-binary
I use a different term (please specify) — free text box available.
Prefer not to answer

Administered by Twins Research Australia, the HBQ survey utilized the TRA database of almost 75,000 twins to recruit participants, with 16,380 (8190 twin pairs) aged 18–88 years invited to participate. TRA members were initially contacted via email with an invitation to participate, with nonresponders sent follow-up email (or SMS) reminders after 7 days. Zygosity data were obtained from twins' self-reported TRA records, as determined during their initial registration process when individuals are given a range of responses to choose from (DNA testing, blood tests, opposite-sex status, or the Peas-In-A-Pod questionnaire).

Table 1. Summary of responses for gender identity and sex assigned at birth

	Sex assigned at birth				
Gender identity	Male	Female	Other	Prefer not to say	Total
Male	1015	15	0	0	1030
Female	4	3417	0	0	3421
Nonbinary	2	10	1	0	13
Free text term	1	4	0	0	5
Prefer not to answer	3	2	0	1	6
Total	1025	3448	1	1	4475

Responses were reported as prevalence estimates and concordance proportions. The subsequent analysis included the Gibbs chi-square test to evaluate the null hypothesis that concordance proportions are equal between MZ and DZ twin pairs, tetrachoric correlations, and relevant 95% confidence intervals (CI), and utilized the Sib-pair genetic analysis package (Duffy, 2020).

The Twins Research Australia study was approved by The University of Melbourne's Office of Research Ethics and Integrity (Ref: 2023-26857-42474-4).

Results

Responses were received from 4475 of those invited (27.3%). They comprised 1862 paired and 751 unpaired responses. The mean age of responders was 52.2 years (*SD* 15.3), with 77.1% birth-assigned females and 22.9% birth-assigned males.

Among the 4475 respondents, all but six (99.9%) provided a response to the gender identity questions. Of the remaining 4469, 36 (0.8%, 95% CI [0.5, 1.1]) provided responses indicating gender diversity (15 transgender men [0.34%], 4 transgender women [0.09%], 12 nonbinary individuals [0.27%] and 5 entering their own free text descriptor of gender [0.11%]) (Table 1).

Gender identity data from both co-twins were obtained for 1862 pairs, among which 27 (1.45%) included at least one gender diverse twin. Among the responding pairs, 1334 (71.6%) were monozygous, 522 (28.0%) were dizygous, 6 (0.3%) unknown; 331 (17.8%) were assigned male-male, 1365 (73.3%) female-female, 165 (8.9%) male-female, and 1 (0.05%) unknown.

Among the 27 pairs with at least one gender diverse twin, the age distribution was younger than for the overall cohort (mean 40.8 years; SD 16.4). 19 were monozygous (2 concordant for gender diversity) and 8 were dizygous (all discordant). Three (11.1%) were assigned male-male; 19 (70.4%) female-female, among whom 2 were concordant for gender diversity, and 5 (18.5%) male-female (Table 2). The Gibbs chi-square test yielded a likelihood ratio test statistic of 1.47, with an associated p value of .225.

Discussion

In this study, we implemented for the first time a two-step approach for collecting gender identity data from our 2023 national twin registry survey. Feasibility was demonstrated by the high completion rate for the two added questions (99.9%). The approach taken could be more accurate, efficient, and cost-effective than a survey specific to gender identity, as it simply requires the questions to be nested in existing and/or planned survey questionnaires, obviating any need to survey specifically for gender diversity. Looking ahead, this report also demonstrates the

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Table 2. Concordance proportions for gender diversity, separated by zygosity and birth assigned sex

Zygosity	Concordant pairs (*n*)	Discordant pairs (*n*)	Proportion	Tetrachoric correlation (95% CI)
MZ ($n = 19$)	2	17	2/19 (0.11)	0.62 (0.33, 0.87)
DZ ($n = 8$)	0	8	0/8 (0.00)	0.00 (0.00, 0.88)

simplicity and utility of incorporating a standardised two-step process for collecting gender identity data in other twin registry surveys.

Data on gender diversity from twin registries can advance understanding of the relative contributions of genetic and environmental determinants of gender diversity. In our study, among twins where at least one identified as trans, concordance was observed in 2 of 19 monozygous and 0 of 8 dizygous pairs. Although slightly lower than most reported concordance rates in the literature (21.0−39.1% in monozygotic twins and 0.0−11.5% in dizygotic twins (Diamond, 2013; Heylens et al., 2012; Polderman et al., 2018; Sasaki et al., 2016), the data align with the general trend of higher rates of concordance in MZ twins when compared to DZ twins. The tetrachoric correlations show a moderate to strong positive association in MZ twins (0.62, 95% CI [0.33, 0.87]). However, in DZ twins, the 95% CI is too wide (0.00, 0.88) to draw meaningful conclusions from the correlation value (0.00). Interestingly, although our study is underpowered ($p = .23$), our results do not align with recent findings from Karamanis et al. (2022), who observed a higher concordance rate in opposite-sex twins (37%) compared to same-sex twins (0%).

Although our small sample size limits the statistical power of these data, its methodological approach offers two key advantages over previous studies that have investigated gender diversity in twins. Twin registry-based ascertainment of twin pairs (with at least one gender diverse twin) is an important methodological advance on many of the earlier reports, as it helps reduce the aforementioned biases. Looking ahead, in the event that other twin registries internationally may be able to incorporate this two-step approach for gendered data collection, it will create future opportunities for collaborative data pooling with resultant gains in statistical power.

Our use of a population-based cohort has also yielded an independent population-based estimate of the frequency of gender diversity among adults in Australia, which is only now incorporating questions about gender identity into the national census. The recent *Standards of Care for the Health of Transgender and Gender Diverse People* (Version 8, 2022) report population estimates ranging between 0.3% to 4.5% (Coleman et al., 2022). The population estimate of 0.8% (95% CI [0.5, 1.1]) calculated in our study falls within the lower end of this range, which is consistent with other reports indicating that higher estimates of gender diversity are typically observed in younger cohorts (Coleman et al., 2022; Eisenberg et al., 2017; Johns et al., 2019; Marino et al., 2024; Strauss et al., 2020). In this regard, it is likely relevant to note that our cohort was relatively old, with a mean age of 52.2 years. Interestingly, our estimate of 0.8% is directly comparable to the recent estimate made by the Australian Bureau of Statistics in their report *Estimates and characteristics of LGBTI+ populations in Australia*, which found that approximately 0.9% of Australian adults are gender diverse (Australian Bureau of Statistics, 2022). In this way, our findings also demonstrate the utility of including gender-related questions in twin registries to generate population estimates of gender diversity. In time, the inclusion of gender-related questions will likely also yield additional valuable health data, particularly for following health outcomes among gender diverse people.

Data availability statement. The datasets used and/or analysed during the current study available from the corresponding author on reasonable request.

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Competing interests. The authors declare that they have no conflict of interest.

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