"Gender Dysphoria" and Autism Spectrum Disorder: Is the Link Real?

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The topic of gender identity in children and adolescents has gained widespread attention in Western society during the past decade, from news media to legislative agendas and popular television. Simultaneously, there have been more referrals to pediatric gender clinics, an increase in the number of pediatric gender clinics, and a surge in research related to pediatric gender identity (Figure 1). More recently, growing attention has been paid to a putative relation between gender dysphoria (GD) and autism spectrum disorder (ASD). This concept has become particularly popular in the lay press.[1] Some individuals have gone so far as to suggest that transgender identity is a result of underlying psychopathology, with ASD being one example.[2] These conclusions are not supported by extant research, and practicing child and adolescent psychiatrists should be aware of the literature on this topic and its limitations.

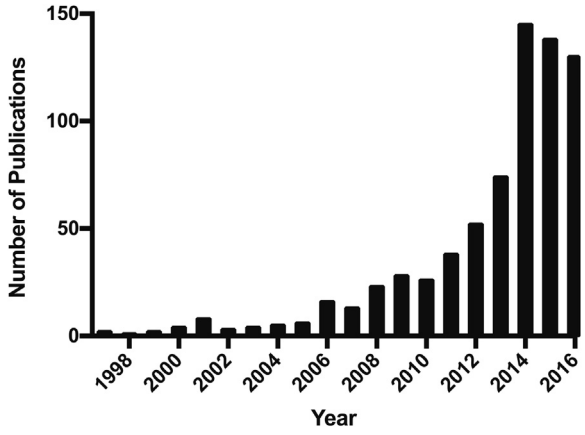
Proponents of the link between ASD and GD have pointed to studies that suggest that gender variance is present in 5% to 7% of youth with ASD[3] compared with approximately 1% in the general population. However, these studies have used a definition of "gender variance" that does not indicate GD or transgender identity. Conversely, some have argued that ASD is over-represented in those with GD,[3] although these studies have used symptomatology scales that might not be specific to ASD. This has led to a number of theories linking these 2 phenomena, although none are supported by concrete evidence.

The 2 groups of studies contain significant limitations. The first such group examines the prevalence of gender variance among youth with ASD. Three such studies have been published.[3] None examined gender identity in these youth. Rather, all 3 used a single item from the Child Behavior Checklist (CBCL) to examine the child's gender variance. Item 110 on the CBCL asks whether a child "wishes to be the opposite sex," and the item is scored on a Likert scale with the options "never," "sometimes," and "often." These studies defined a gender variant group as a composite of those who qualify as sometimes or often. Although neurotypical individuals with gender identity disorder (GID; now GD) have been found to be more likely to endorse these items than those without these diagnoses,[4] the measure is not specific to these diagnoses. It would be incorrect to suggest that these individuals are transgender or experience GD, because the question makes no attempt to assess core gender identification. Many of these individuals could in fact be cisgender, with mere transient desires to be the opposite gender, perhaps owing to gender-atypical interests (a boy who enjoys ballet, for instance, might "sometimes wish to be a girl" in an abstract way that is not tied to a core gender identity). This item is particularly problematic for youth with ASD who are prone to rigid thinking. An adolescent birth-assigned male with ASD who enjoys knitting, for instance, might label this activity as "feminine" and thus report that he "sometimes" wishes to be the opposite gender because he sometimes wishes to knit. This adolescent could very well identify as male and have no desire for social transition, hormonal interventions, or other interventions often associated with transgender identity. We urge future investigators to conduct comprehensive interviews with youth who screen positive with the CBCL to better understand the nature of their cross-gender interests. To meet *DSM-5* criteria for GD, one must have incongruence between one's gender assigned at birth and one's gender identity, in addition to clinically significant distress or impairment. At this time, these studies do not establish an over-representation of GD among those with ASD.

The second set of studies takes a converse approach by examining the rates of ASD symptoms in youth referred to gender identity clinics. A number of case reports have shown that youth with ASD might indeed experience gender-related concerns that might require gender-affirmative treatment.[3] Subsequent research has attempted to establish an over-representation of ASD in youth with GD by relying on the use of ASD screening instruments (such as the Social Responsiveness Scale and Autism Quotient) to explore this co-occurrence.[3] These findings have been helpful in establishing that youth and adults with GD might experience social impairments, but they do not establish that these patients have ASD. It is increasingly understood that such screening instruments can be nonspecific for ASD, with youth suffering from other emotional or behavioral problems having higher scores in the absence of ASD.[5] Higher scores would be expected in youth with GD, because this population is known to have high rates of internalizing psychopathology.[6] True ASD is a biological brain-based disorder present from the earliest periods of development, with established genetic risk factors, empirical treatment paradigms, and cognitive-neuroscientific findings.[7] By contrast, social impairments could be the result of a number of diverse processes. When these social deficits are the result of reversible or modifiable processes (such as poor peer relationships and environmental stigma commonly seen in transgender youth), these symptoms can disappear over time, as vividly illustrated in studies of emotionally deprived youth.[8] This underscores the importance of diagnostic approaches to ASD that rely on longitudinal information from multiple informants, a robust developmental assessment, and consideration of reversible factors that could be contributing to patterns of behavior. In addition, it highlights that the prognosis for many youth with social deficits and GD could be promising, because they might have no intrinsic social cognitive deficit.

Notably, 1 study explored the link between the *DSM-IV* diagnosis of GID and ASD in a clinical sample of children and adolescents using the more comprehensive Diagnostic Interview for Social and Communication Disorders (DISCO), a semistructured 2- to 4-hour interview.[9] This study identified 1 child (of 52 with GID) who also met criteria for ASD according to the DISCO. Of the 77 adolescents with GID, 5 met criteria for ASD according to the DISCO (6.5%). The remaining

FIGURE 1 PubMed indexed publications from 1997 to 2016 using the search term "transgender youth."



youth who screened positive for ASD carried a diagnosis of GID not otherwise specified and thus cannot be considered to necessarily identify as transgender. Because there was no non-GID control group, there are several alternative explanations that do not involve an etiologic relation between GID and ASD, including higher rates of ASD in clinical populations generally, a type 1 error secondary to a small sample, and youth with ASD being more likely to seek gender-affirmative care. Further, it is notable that the rate of ASD for children in the sample was in keeping with population norms. Because ASD is generally considered to be present from the earliest developmental periods, the hypothesis that ASD is over-represented in those with GID would predict higher rates in children and adolescents. In contrast, the hypothesis that social deficits in adolescents might be related to non-ASD psychosocial challenges associated with GID is more consistent with these findings, particularly because depression risk has been shown to increase significantly as transgender youth progress from childhood into adolescence.[10]

In conclusion, current research has not established an over-representation of GD in those with ASD or the converse. Existing studies have provided an important first step in highlighting that youth with ASD can have a range of gender-related experiences and indeed might present with GD; however, further research is needed to understand whether there is a true relation between these 2 phenomena. We note that regardless of whether an over-representation exists, by chance, there will be a group of individuals with ASD and true GD or transgender identity. In line with clinical guidelines,[11] these youth should be considered for similar gender-affirmative interventions as neurotypical individuals.[12] In addition, we posit that youth with cross-gender identification, because of a high prevalence of minority stress, poor peer relationships, and familial non-acceptance, might score higher on measures of social impairment owing to environmental factors related to social stress. This could represent a reversible condition, because these patients do not have the intrinsic social cognitive deficits that underlie ASD. In addition, all research in the area has been associational. There have been no data to suggest that GD stems from ASD itself. Nonetheless, the research in this area to date has created considerable interest in understanding the gender experiences of youth with ASD and could provide the impetus to conduct the necessary work to truly understand the gender narratives of youth with ASD and how they can best be supported.

See Supplement 1 (available online) for a version of this Translations article that features an expanded list of references for further reading on this topic.

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SUPPLEMENT 1: FULLY ANNOTATED ARTICLE

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identification, owing to a high prevalence of minority stress, poor peer relationships, and familial non-acceptance, might score higher on measures of social impairment owing to environmental factors related to social deprivation. This could represent a reversible condition, because these patients do not have the intrinsic social cognitive deficits that underlie ASD. In addition, all research in the area has been associational. There have been no data to suggest that GD stems from ASD itself. Nonetheless, the research in this area to date has created considerable interest in understanding the gender experiences of youth with ASD and could provide the impetus to conduct the necessary work to truly understand the gender narratives of youth with ASD and how they can best be supported.

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