In this review, we briefly summarize much of the existing literature on gender-related concerns and autism spectrum disorders (ASD†), drawing attention to critical shortcomings in our current understanding and potential clinical implications. Some authors have concluded that gender identity disorder (GID), or gender dysphoria (GD), is more common in individuals with ASD, providing a range of potential explanations. However, existing literature is quantitatively limited, and our capacity to draw conclusions is further complicated by conceptual challenges regarding how gender identity is best understood. Discourses that emphasize gender as a component of identity formation are gaining prominence and seem particularly salient when applied to ASD. Individuals with ASD should enjoy equal rights with regard to treatment for gender dysphoria. Clinicians may be able to assist individuals in understanding this aspect of their identity by broadening the social frame and facilitating an exploration of gender roles.

INTRODUCTION

An increasing number of reports describe gender-related concerns in individuals with autism spectrum disorder (ASD). Abelson [1] drew attention to the specific potential for gender issues in individuals with ASD, and subsequently, a number of case reports and at least one systematic study have appeared in the literature [2-9]. Perhaps the earliest case report [2] was of two male individuals with ASD who presented with “feminine gender-stereotyped preoccupations,” and subsequent work has cited this case report as an early example of comorbid gender identity disorder (GID) and ASD [9]. A subsequent case report appeared in 1997 [3] and drew attention to the hypothesis that impairment in social interaction could contribute to atypical gender identity formation.

The report of two cases by Mukaddes [3] was notable for identifying patterns of transgender identity and behavior in two boys seen at the same clinic for several years, although even in this study the individuals were not followed-up beyond a young age (7 and 10 years) — a critical shortcoming given the low rate of persistent transgender identification following puberty [10]. It was not until 2005 that a case report described an adult with Asperger syndrome (AS) and “GID according to DSM-IV (Diagnostic and Statistical Manual) criteria” [8]. The authors go on to introduce the hypothesis that GID developed as a “sequel to AS,” highlighting the extreme-male brain hypothesis and the patient adopting “male emotional and cognitive traits.” Perhaps most recent was the report of two cases of individuals with both ASD and GD according to DSM-5 criteria [11]. In this paper, the authors highlight specific aspects of ASD, including theory of mind deficits and cognitive style as being potential influences on the process of gender identity formation.

De Vries et al. describe a carefully conducted study looking at the rates of autism and AS in a sample of 204 children and adolescents referred to a gender identity clinic and report a 7.8 percent (n = 16) incidence of ASD [9]. The authors reasonably conclude that the incidence of ASD appears to be higher in “gender dysphoric” individuals than in the general population. This conclusion is particularly apt as it acknowledges some of the limitations of the study findings. Pertinently, of the participants who were thought to meet criteria for ASD, only one of the children (out of seven) had persistent GID, and the adolescent group was heterogeneous with five (out of nine) participants meeting criteria for GID and the remainder for Gender Identity Disorder Not Otherwise Specified (GID-NOS) or transvestic fetishism. In concluding that ASD is more common in “gender dysphoric”
individuals, the authors were drawing attention to the fact that ASD is more common in individuals with a broad range of gender-related concerns or questions. It is important to note the authors were not referring to the DSM-5 construct of gender dysphoria (GD), which is a narrower construct. Reflecting this finding, the authors make the recommendation that future studies focus on “dimensional measures and specific cognitive or neuropsychological profiles of individuals with co-occurring gender dysphoria and ASD.” This issue is somewhat complicated by the markedly different view of the “broader” autism phenotype and the rather more restricted view of “autism spectrum” disorders in DSM-5 [12].

CONCEPTUAL CHALLENGES

An obvious challenge to our understanding of gender in ASD is the small number of existing reports and studies. However, there are a number of additional challenges. The first arises from the reliance on categorical diagnosis. Gender-related concerns are currently codified in the DSM-5 under the diagnosis of “gender dysphoria,” reflecting a change from prior editions of the DSM-IV that regarded transgender patients as having “gender identity disorder” [13]. This change was significant for a number of reasons: It counters the notion that being transgender is a mental illness, while retaining the capacity for clinicians to provide a label for those who may suffer psychiatric distress owing to gender-related concerns (and may wish to receive support from insurance to undergo relevant treatment) [14]. Despite continued critiques of the inclusion of “gender dysphoria” in the DSM-5, many regard the changes as a significant improvement — notably symbolized in a statement from the chairs of the World Professional Association for Transgender Health [15].

However, this new definition represents something of a challenge for certain types of research efforts. Given that the definition relies upon the presence of distress as being a necessary criterion for the label, many individuals with gender-related questions would not meet criteria. It is likely that a significant number of individuals who, in some ways, identify with a different label to their biological gender will not be captured by this definition. The extent to which this represents a problem is unclear. It may be that only those who have significant distress related to their gender warrant investigative and clinical attention. On the other hand, even individuals without distress may benefit from interventions that help them better understand their own gender narratives — perhaps particularly in the case of individuals with ASD.

It may be helpful to conceptualize the process of gender identity formation as a normal developmental process through which all children determine “their placement in a gender group” [16]. It is known that an individual’s ability to distinguish one’s own gender generally occurs by age 3 in neurotypical individuals [16]. Awareness of “gendered” toys, roles, appearances, and forms of play similarly begin between 2 and 2.5 years of age. Further differentiation of gender identity is felt to progress in stages, with an understanding of gender stability (the concept that an individual’s gender remains stable over time) and gender consistency (one’s gender is “a fixed and immutable characteristic not altered by superficial transformations in appearance or activities”) generally progressing stepwise between the ages of 3 and 7 years old [16]. The variations in cognitive and social development between individuals complicate simple efforts to extrapolate results from same age and typically developing samples. Comparison to children with developmental problems may not totally address this issue. Furthermore, it may be difficult to control for cultural factors that play a role in gender identity formation.

The idea that gender-related concerns may represent a developmental process is further substantiated by studies of children with gender dysphoria followed through adolescence. In a study of pre-pubertal male and female children with gender dysphoria followed-up approximately 10 years later, only 27 percent of children with gender dysphoria remained gender dysphoric at follow-up [10]. Of those individuals who no longer expressed gender dysphoria at follow-up, a significant portion (all female and half the male participants) expressed a non-heterosexual sexual orientation [9]. Thus, gender concerns in neurotypical children prior to puberty may represent a developmental process related to both gender and sexuality for many individuals. This is further substantiated by data showing it is common for gender identity to evolve up to the onset of puberty, with most children who initially had questions about their gender assuming the identity of their natal gender by the end of adolescence [17]. Similar results emerged in another study looking at a population of 25 girls (average age of 8 years) with GID followed-up post-adolescence [18]. Notably, the authors suggest that these findings point to a plasticity in gender identity differentiation early in development that narrows through adolescence. As such, children with GID prior to adolescence are more multipotential with regard to gender identity and become more differentiated with regard to the presence of gender dysphoria through adolescence [18]. Putting this in the context of ASD, it seems reasonable that individuals with ASD, who are known to display differing patterns of social development when compared with neurotypical individuals, may similarly come to progress through gender development along differing timelines or frameworks, complicating the issue of how best to categorize gender concerns in children and adolescents with ASD.

CLINICAL CONSIDERATIONS

Existing literature contains a variety of recommendations about the clinical implications of a potential co-occurrence of ASD and gender-related concerns. Some authors draw attention to the importance of considering the
influence of the underlying neurodevelopmental disorder when assessing a patient with gender related concerns [3]. De Vries et al. [9] suggest that there is a need to determine whether gender dysphoria represents a “general feeling of being ‘different’ or a ‘core’ cross-gender identity.”

Individuals with ASD have the same rights as other individuals to appropriate assessment, diagnosis, and treatment of gender-related concerns. The challenge that exists surrounds being attentive to the particular concerns that may influence this presentation in ASD individuals; the goal should be to facilitate improved understanding and patient satisfaction and not to increase the number of barriers to appropriate treatment. Although a significant amount of additional study is required, some tentative recommendations are possible.

First, it may be helpful to be attentive to the development of gender identity formation in individuals with ASD from an early age. This facilitates the role of the clinician as an ally in helping the individual explore his or her own gender narrative, rather than as a gatekeeper for medical intervention. This approach is supported by some early findings that the development of gender identity is correlated with social skills, communication skills, and mental age and therefore potentially delayed in individuals with ASD [1]. The role of a clinician (or, indeed, parent) in this context would be to facilitate expansion of the social frame and facilitate identity formation in the broadest sense. In addition, education about sexuality and specialized sex education may be of particular value, with an emphasis on techniques that are effective in the context of social skills training [14,15]. Facilitating an exploration of sexuality seems especially pertinent given recent findings that most children with gender-related concerns eventually identify with their natal gender following puberty and frequently adopt homosexual or bisexual identities [19]. It is possible that individuals with ASD may experience similar trajectories in their gender narratives, but potentially follow a different timeline than normally developing individuals owing to reduced social interaction and fewer opportunities to explore their sexual identity.

CONCLUSIONS AND OUTLOOK

Existing literature makes the significant contribution of drawing our attention to the presence of gender-related concerns in individuals with ASD. However, it is unclear whether the most fruitful way to conceptualize this issue is in terms of comorbidity. A more complex approach that attempts to understand gender in developmental terms is potentially more salient for both research and clinical purposes. Our current understanding about the unique social development of individuals with ASD, which may impact the process of gender identity formation, underlines the need for such an approach. Future work should attempt to explore the development of gender identity in a longitudinal fashion, in order to improve our understanding of the relationship between gender and ASD. Such an understanding could improve patient well-being by allowing clinicians to better guide and advise individuals with ASD who seek assistance for gender-related concerns.

REFERENCES