Title: Symptoms of Autism Spectrum Disorder in Individuals with Down Syndrome

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Introduction: With a prevalence rate of approximately 1 in 700 live births, Down syndrome (DS) is the most common genetic cause of intellectual disability. Children with DS are often described as being highly sociable, being affectionate, and getting along well with others. Indeed, research on the behavioral phenotype of DS demonstrates relative strengths in many aspects of social communication and social-emotional functioning. Despite these areas of strength, there is a growing body of work identifying co-occurring social-cognitive, social-emotional, social-communicative challenges associated with DS (e.g., Channell et al., 2015). In fact, recent studies have found that the prevalence of autism spectrum disorder (ASD) is higher in individuals with DS than it is in the general population, although there is considerable variability in estimates of ASD prevalence across studies. Much remains to be understood, however, as regards the presence and profile of ASD symptomatology in individuals with DS. This is an important area of investigation as both subclinical and clinical levels of ASD symptoms are likely to negatively impact success in daily life for individuals with DS. In the present study, we sought to (1) describe the prevalence of ASD in DS using the Autism Diagnostic Observation Schedule, 2nd edition (ADOS-2), a semi-structured observational measure of ASD symptomatology using a DSM-5 based approach; (2) describe the ASD symptoms that differentiate individuals with DS who are classified as having ASD from those classified as having DS only; and (3) consider the influences of an individual’s characteristics (chronological age, nonverbal IQ, and verbal IQ) on ASD symptom severity.

Method: Participants were 34 males and 29 females with DS between the ages of 6 and 23 years (M CA = 14.94, SD = 5.46). The mean nonverbal IQ score for our sample was 49.30 and the mean verbal IQ score for our sample was 39.89, when using deviation IQ scores from the Stanford Binet-5 to adjust for floor effects (Sansone et al., 2014). All participants were able to produce at least two-word phrases. Both the Autism Diagnostic Observation Schedule-2 (ADOS-2; Lord et al., 2010) and Autism Diagnostic Interview-Revised (ADI-R; Rutter et al., 2003) were administered, with adherence to Risi et al. (2006) caseness criteria to determine ASD diagnostic status. The ADOS-2 Calibrated Comparison Score, an indicator of overall ASD severity level, as well as both Social Affective (SA) and Restricted and Repetitive Behavior (RRB) domain severity scores (Hus et al., 2014) were also used. In the present study, 4, 37, and 22 participants received Module 1, Module 2, and Module 3, respectively. The deviation IQ standard score for the Stanford Binet-5 Nonverbal and Verbal domains were also considered in analyses (see Sansone et al., 2014 for procedures).

Result: Approximately 30% of our sample met overall ADOS-2 criteria for an ASD diagnosis. Severity scores on the SA and the RRB domains fell into the concern range for 39.68% and 30.16% of our sample, respectively. When considered as a function of module, we observed variation in the proportion of participants classified as having ASD using the ADOS-2 (Module 1: 25% classified as ASD, Module 2: 40.5% classified as ASD, and Module 3: 13.6% classified as ASD). At the item-level, scores on the items assessing “eye contact”, “facial expressions”, “showing”, “quality of social overtures”, and “hand and finger and other complex mannerisms” were most discriminating of individuals with DS classified as having or not having ASD on the ADOS-2 Module 2. For the ADOS-2 Module 3, scores on items assessing “quality of social overtures”, “quality of social response”, “overall quality of rapport”, and “excessive interest in unusual of highly specific topics/objects or repetitive behaviors” were most discriminating of the two subgroups. Although severity scores on both the SA domain and the RRB domain were significantly associated with overall CCS scores, the association between severity scores on the two domains was not significant (r = .26, p = .24). Finally, we considered the relations between ASD severity scores and CA, nonverbal IQ, and verbal IQ. Nonverbal IQ was negatively correlated with SA severity scores (r = -.44, p = .04). Verbal IQ was negatively correlated with both overall CSS scores (r = -.57, p = .01) and SA severity scores (r = -.59 p = .01). No significant associations were observed between the participant characteristics considered and RRB severity scores. Finally, none of the ASD severity scores varied with CA.

Discussion: When using the ADOS-2, we found that approximately 30% of our sample met criteria for an ASD diagnosis. Individuals with DS who used phrase speech up to but not including fluent speech (Module 2) were most at risk for meeting criteria for an ASD diagnosis; however, we had very few participants who used no speech up to simple phrases (Module 1) on the ADOS-2. We found that multiple items, mostly in the SA domain, differentiated children with DS+ASD from those with DS only. Finally, developmental characteristics, particularly language skills, were significantly associated with both overall ASD severity and SA symptom severity in our sample. Theoretical and clinical implications will be discussed.
References/Citations:


