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

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Introduction: Research suggests that individuals with Down syndrome (DS) show an increased prevalence of ASD, and that many children with DS who are not diagnosed with ASD still exhibit elevated ASD symptoms. Past research demonstrates that children with DS show elevated scores on measures of autism symptoms (Channell et al., 2015), often exhibit obsessive-compulsive behaviors that are salient enough to raise questions about an ASD diagnosis (Kent et al., 1999), and demonstrate a high frequency of restricted/repetitive behaviors (Evans & Gray, 2000). This presentation aims to investigate the presence of autism symptoms in individuals with DS in order to better inform screening and diagnosis of ASD in this population.

Method: Primary caregivers of children aged 6-18 years with either a DS (n=76) or ASD (n=37) diagnosis completed an online survey that included demographic questions, an autism screening measure (the Social Communication Questionnaire; SCQ), a dimensional measure of autism symptoms (the Autism Spectrum Rating Scales; ASRS), and a measure of adaptive behavior. Children with DS were recruited via Down Syndrome Connect, and children with ASD were recruited via targeted Qualtrics survey panels and a local research registry. All data collection is complete.

Result: A total of five of the seventy-six included respondents in the DS group (6.6%) indicated that their child had a comorbid ASD diagnosis, and seventeen out of the seventy-six individuals (22.4%) in the DS group scored above the cut off score of 15 on the Social Communication Questionnaire ("screen positive"), suggesting potential concern for ASD. Additionally, on the ASRS, children with DS had overall scores, as well as scores across the three subscales (social communication, unusual behaviors, and self-regulation), that were significantly higher than the typical population mean score of 50. To investigate the specific symptom areas driving elevations in ASD symptoms for the subset of children with DS showing substantial ASD-related symptoms, we split the DS group into a screen positive (n=17) and screen negative group (n=59) based on SCQ scores. On the ASRS, both the screen positive and screen negative DS group had scores across all subscales of the ASRS significantly higher than the mean score of 50. However, screen-positive children with DS showed levels of social communication problems and unusual behaviors that were similar to those of children in the ASD comparison group, but significantly higher compared to screen-negative children with DS. Screen-positive children with DS were similar to screen-negative children in the area of self-regulation problems (with the ASD comparison group showing significantly more self-regulation problems that either DS group). A more fine-grained analysis of the 8 treatment scales of the ASRS (peer socialization, adult socialization, social emotional reciprocity, atypical language, stereotypy, behavioral rigidity, sensory sensitivity, and attention) suggested that behaviors relating to social emotional reciprocity and sensory sensitivities may best discriminate children with DS who are demonstrating significant ASD-related symptoms.

Discussion: Consistent with previous research, the percentage of individuals with DS who entered the study with a comorbid diagnosis of autism, as well as the percentage of those who screened positive for ASD concerns, suggests that children with DS may be at increased risk for ASD. The overall elevation of ASRS scores in children with DS suggest that elevations in ASD symptoms are present to some extent in many individuals with Down syndrome. However, more detailed examination of screen-positive vs. screen-negative subgroups suggests that a subset of children with DS have substantial symptoms of autism above and beyond the elevations seen across individuals with DS. Our results suggest that questions related to social emotional reciprocity and sensory sensitivities appear to discriminate children with DS who screened positive from those who screened negative particularly well. This suggests that future research should examine these symptom areas in more detail to determine whether they may be particularly useful for ASD screening and diagnosis in children with DS.

References/Citations: