Title: Characteristics Associated with Autism Spectrum Disorder Risk in Individuals with Down Syndrome

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Introduction: The presence of autism spectrum disorder (ASD) symptoms in individuals with neurogenetic syndromes, such as Down syndrome (DS), is of great debate in the literature because it is difficult to distinguish behaviors associated with ASD from those associated with the genetic syndrome itself or with intellectual disability more broadly. It is important to identify individuals who display particular patterns of symptoms so that these individuals can be provided with appropriate treatment options. The emerging literature on ASD in DS suggests a prevalence rate of approximately 7-18% (DiGuiseppi et al., 2010; Kent et al., 1999; Lowenthal et al., 2007) and that greater severity of ASD symptoms is associated with lower adaptive functioning and more severe intellectual disability (Reilly, 2009). These findings are mostly based on small to moderate sample sizes that have mainly relied on medical or educational reports to determine level of intellectual disability. The current study seeks to fill these gaps by examining the characteristics associated with ASD risk through direct assessment of cognitive ability and parent report of ASD symptoms and adaptive and maladaptive behaviors in a large sample of individuals with DS from a national network of sites comprising the Down Syndrome Cognition Project (DSCP).

Method: A total of 207 individuals with DS ages 6-25 years from the larger DSCP database met inclusion criteria for this study (i.e., completed the in-person assessment and whose parent completed the Social Communication Questionnaire [SCQ]). We used the Kaufman Brief Intelligence Test, 2nd edition (K-BIT2) to measure IQ (level of cognitive impairment) and the Nisonger Child Behavior Rating Form (NCBRF) and the Scales of Independent Behavior-Revised (SIB-R) to measure maladaptive and adaptive behaviors across several subscales (parent report). We used the SCQ to screen for the presence of ASD risk. Using the recommended SCQ clinical cutoff score of 15 (Rutter et al., 2003), we dichotomized our sample into “high risk” (HR) and “low risk” (LR) groups. We compared the two groups on age, sex, IQ, and adaptive (SIB-R; NCBRF) and maladaptive (NCBRF) behaviors.

Results: Twenty-six individuals with DS met criteria for “high risk” (HR) of ASD on the SCQ, representing 12.6% of our sample. The HR (n = 26) and LR (n = 181) groups did not differ by age (HR: 8.00–21.50, M = 13.84, SD = 3.57; LR: 6.00–25.00, M = 13.66, SD = 5.07; p = .87). The groups did, however, differ by sex (HR: 69.2% male, LR: 48.6% male; Chi-square = 3.87, p = .05). Because a large number of participants scored at the floor level on the KBIT-2 (IQ = 40), we dichotomized this variable (at floor vs. above floor) to compare participant groups. There was a significant difference between the two risk groups in the number of participants scoring at floor (HR: 65.4%, LR: 28.7%; Chi-square = 13.75, p = .001). Finally, there was also a significant difference between groups in SIB-R adaptive behavior (HR: Mean standard score = 32.46, SD = 23.62; LR: Mean standard score = 59.04, SD = 24.08; p < .001) as well as NCBRF adaptive behavior (HR: Mean Z-score = -.61, SD = .84; LR: Mean Z-score = .09, SD = .90; p < .001) and maladaptive behavior (HR: Mean Z-score = .55, SD = .86; LR: Mean Z-score = -.08, SD = .63; p = .001). A specific profile of maladaptive behavior was observed such that certain NCBRF subscales differentiated the two participant groups: Conduct Problems, Hyperactive, Self-Isolated/Ritualistic, & Self-Injury/Stereotypic. We also explored correlations of sample characteristics with ASD risk as a continuous variable; results paralleled those of the between-group (dichotomous) findings.

Discussion: Using a large national sample of individuals with DS to gain a broader perspective on the features associated with ASD risk, we found that 12.6% screened at elevated risk for ASD on the SCQ, which fits with rates reported by previous studies. We also found that those in the “high risk” sample were more likely to be male, have lower IQs, lower adaptive behavior, and more maladaptive behavior. Our findings support and expand the current literature by including a direct assessment of cognitive abilities and a more nuanced profile of maladaptive behavior in a very large, nation-wide sample. These data provide insight into how ASD symptoms may manifest in individuals with DS. Our findings, therefore, inform clinicians and researchers who are working to develop additional measures or modified algorithms to more accurately and efficiently screen for, and identify, ASD in neurogenetic syndromes such as DS (Glennon et al., 2017), ultimately leading to more expedient treatment options to support optimal development.
References/Citations: