Title: Longitudinal Social Communication Comparisons in Preschool Age Children with Down Syndrome and Fragile X Syndrome

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Introduction: Individuals with neurogenetic syndromes, such as fragile X syndrome (FXS) and Down syndrome (DS) are at elevated risk for autism spectrum disorder, with reported rates of 60-75%1,2 and 7-18%3 respectively. However, it can often be difficult to disentangle if social communication impairments seen in these populations are attributable to intellectual disability or specific to a co-morbid diagnosis of autism spectrum disorder (ASD). Additionally, the longitudinal unfolding of autism symptomatology in these populations may be unique and specific to the genetic conditions, rather than what is seen in non-syndromic ASD. Given that the co-morbidity of ASD in these neurogenetic syndromes results in more significant impairments across a range of domains4,4, accurate diagnoses are imperative to inform targeted treatments.

Method: Participants were seen as part of a larger longitudinal study on the early developmental features of ASD in children with FXS, with comprehensive diagnostic visits occurring annually between 2-5 years. As part of the parent study, all participants were administered the ADOS-2 and the Mullen Scales of Early Learning, at each time point. A clinical best estimate (CBD) diagnosis procedure was conducted at each annual visit. The CBE procedure was led by a licensed psychologist and research reliable staff who reviewed developmental data, ASD data, and behavioral data including watching a video clip of the participants’ assessments. Participants with DS are part of an initial pilot sample collected as a comparison group. Video clips from the ADOS-2 administrations were used to create two 6-minute segments, and the Brief Observation of Social Communication Change (BOSCC) was applied to the video clips. Total scores on the BOSCC range from 0-75, with higher scores indicating more severe autism symptomatology. This study presents 2 aims; (1) to examine the prevalence of ASD in preschool aged children with Down Syndrome (n=13, 11 males, 2 females), and (2) to compare trajectories of BOSCC scores between participants with FXS and DS. For aim 2, the participants in aim 1 are matched to 13 children with FXS on sex, intellectual ability and their ASD diagnostic outcome. Currently, 4 participants in each group (total n = 8) have BOSCC scores at 2 time points to report. All ADOS’s have been collected and all BOSCC data will be coded and available for analysis by the time of the conference (n=26: 13 DS and 13 FXS).

Results: Prevalence estimates indicate that 15% (n=2) of the participants with DS met diagnostic criteria for ASD. Trajectories of BOSCC total scores were compared between participants with FXS and DS, based on their ASD diagnostic status. Results suggest that participants with ASD, regardless of neurogenetic syndrome (FXS or DS) have higher initial BOSCC scores at 24 months and generally increasing trajectories. In comparison, participants without ASD have lower initial BOSCC scores and generally stable trajectories, or slight decreases in symptoms over time. Additionally, within the ASD group, participants with FXS appear to have higher initial scores and more steeply increasing trajectories. See Figure 1.

Discussion: Prevalence estimates in the sample are consistent with what has been found in previous work, providing additional evidence for the increased prevalence of ASD in DS. This is the first sample of preschool age children with DS to be assessed longitudinally for ASD. Although preliminary, these results suggest that trajectories of ASD symptom development in preschool age children with FXS and DS may differ, with those with FXS evidencing steeper increases in symptom impairment between 2 and 3 years old. Additionally, these findings highlight that the trajectory of symptoms across development may be a critical feature to consider when examining cross-syndrome comparisons of ASD in young children. These findings highlight the need for ongoing monitoring of symptoms throughout the preschool and early school age years.

References/Citations: