Title: Comparing Social, Communicative, and Repetitive Behaviors in Children with Down Syndrome and Idiopathic Autism Spectrum Disorder Matched on Verbal Mental Ability Level

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Introduction: Autism spectrum disorder (ASD) is characterized by impairments in social interaction and communication skills (SOC-COM) as well as a restricted or repetitive range of behavior and interests (RBI). Although these symptoms often occur together, research suggests that independent genetic factors may account for each component, and therefore, these symptoms may be separable [1]. Youth with genetic disorders that have elevated rates of ASD symptoms offer an opportunity to examine SOC-COM and RBI skills within the context of a disorder with a known etiology. In particular, children with Down Syndrome (DS), although known for their social strengths [2], have an elevated risk for meeting the criteria for ASD compared to typically developing children [3,4]. Past studies have shown individuals with DS and comorbid ASD have a unique symptom profile from those with idiopathic ASD on the Social Communication Questionnaire [5]. The current study aims to investigate the nature of ASD symptom profiles as measured by the Autism Diagnostic Interview-Revised (ADI-R) in children with DS (with and without comorbid ASD) and compare this profile to that found for youth with idiopathic ASD matched carefully on verbal mental age.

Method: One-hundred fourteen children participated (diagnostic groups: Idiopathic ASD=76, DS=27, DS+ASD=11; mean chronological age = 4.50; 36 females). Participants with DS were a subset of individuals enrolled in studies at the University of Colorado School of Medicine who were carefully evaluated for ASD using expert clinical opinion. See [4] for more details. Participants with idiopathic ASD (i-ASD) were drawn from the National Database on Autism Research and were carefully matched to the DS participants (using a 2:1 ratio) based on verbal mental age (average of the Mullen Expressive and Receptive Language scale scores), sex, and chronological age. In order to compare the profile of scores on the SOC, COM, and RBI domains of the ADI-R, algorithm items were drawn from each of these domains. We utilized the algorithm for participants 2 years - 3 years, 11 months to identify items for each domain. We only evaluated items for the communication domain that were targeted for nonverbal participants, as all participants had ratings on these items. Scores on each of the domains were then averaged (i.e., average domain scores for SOC, COM, and RBI reflected the mean rating, 0-2, on relevant items).

To evaluate whether SOC-COM and RBI profiles varied as a function of diagnostic group, a 2 (domain: mean SOC/COM, and RBI score) x 3 (diagnostic group) repeated measures ANOVA was completed. This analysis was followed by a 3x3 repeated measures ANOVA with one within-subjects factor (domains: SOC, COM, RBI) and one between-subjects factor (diagnostic group), analogous to the ASD symptom triad model from the DSM-IV.

Results: The results of the 2x3 repeated measures ANOVA revealed a main effect of group (F[2,111]=93.81, p<.01), such that both the i-ASD and DS+ASD groups had significantly higher scores than the DS group overall as expected. This main effect was qualified by a significant group x domain interaction (F[2, 111]=3.83, p<.05), such that the ASD group differed on SOC-COM and RBI (SOC-COM more impaired RBI); p < 01) while both DS groups’ scores did not differ. In addition, the DS only group had lower SOC-COM and RBI scores than the two ASD groups; however, these groups did not differ from one another on SOC-COM or RBI.

The results of the 3x3 repeated measures ANOVA revealed a somewhat more nuanced pattern of findings. Again the DS group was less impaired than the two ASD groups overall (F[2,111]=100.58, p<.01) However, the group x domain interaction (F[4,222]=4.10, p<.01) identified differences in the DS+ASD and i-ASD profiles. First, the i-ASD group was significantly more impaired on the SOC domain than the DS+ASD group (p<.01), but scores on the COM and RBI domains did not differ. Second, the DS+ASD and i-ASD groups presented with a different pattern of scores on the SOC, COM, and RBI domains. Specifically, for the i-
ASD group, COM scores were the most impaired, followed by the SOC domain, followed by the RBI domain (all ps<.001). While the DS+ASD group’s overall pattern of scores was similar to the i-ASD group, only the COM domain was significantly more impaired than the SOC and RBI domains (ps<.05). Lastly, unlike the ASD groups, the DS group presented with no differences among the SOC, COM and RBI domains.

Discussion: Our findings illuminate the unique symptom profiles for children with DS and comorbid ASD compared to those with idiopathic ASD. Specifically, these data demonstrate that children with DS and comorbid ASD’s pattern of behavior more closely resembles those with i-ASD than those with only DS. However, social reciprocity skills appear to be less impaired in those with DS+ASD than those with i-ASD. This pattern of findings was only revealed when the SOC and COM domains from the ADI-R were considered separately, consistent with the DSM-IV symptom clustering. Thus, it appears that the dyad model from the DSM-5 may not be as informative when studying youth with ASD and comorbid genetic disorders. In conclusion, these findings illustrate the need for a finer grained approach to examining ASD symptom profiles for different groups of youth presenting with the disorder. This study is a first step in examining the DS+ASD symptom profile in greater depth using the ADI-R. Future research will be conducted to examine the ability of specific ADI-R items to distinguish between the DS, DS+ASD, and i-ASD groups using graph analytic procedures.

References/Citations: