Title: Are Filled Pauses Related to ASD Symptomatology in Adolescents and Young Adult Males with Fragile X Syndrome?

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Introduction: Fragile X syndrome (FXS) is caused by a mutation in the FMR1 gene. In addition to the presence of intellectual disability, the phenotype of FXS includes a variety of behavioral features, with those akin to the symptoms associated with a diagnosis of autism spectrum disorder (ASD) among the most prominent. In fact, with as many as 60% of males with FXS receive a comorbid diagnosis of ASD, and FXS is the leading single-gene cause of ASD. However, it is important to note that there has been considerable variability in estimates of the ASD comorbidity prevalence across studies. Moreover, there is uncertainty within the field as to whether the behavioral features shared between the FXS and ASD phenotypes reflect similar underlying mechanisms or would benefit from similar approaches to treatment (Abbeduto et al., 2014). More research is needed to resolve this uncertainty. Within the literature on idiopathic ASD, there is a growing body of work demonstrating that individuals with ASD are less likely than typical peers or peers with specific language impairments to use “um” as a filled pause in conversation, suggesting that the use of “um” relative to other types of filled pauses (e.g., “uh”) may be diagnostic (e.g. Irvine et al., 2016; McGregor & Hadden, 2018). To date, it is not known whether the use of “um” fillers is similarly related to ASD symptomatology in individuals with FXS. The information gained from this line of investigation has the potential to inform our understanding of the mechanisms underlying ASD symptomatology in FXS. In the present study, we considered the relations between ASD symptomatology and the use of “um” and “uh” fillers to signal a pause in male adolescents and young adults with FXS.

Method: Participants were 49 males with FXS between the ages of 15 to 23 (M CA = 18.2), with a mean nonverbal IQ of 38.9. All participants were able to produce at least two-word phrases to communicate. 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. Using these criteria, 14 individuals were classified as having FXS only and 35 were classified as having FXS+ASD. The ADOS-2 calibrated comparison score (CSS) was also used to provide a continuous metric of ASD symptom severity. Use of the filled pauses “um” and “uh” was assessed from a semi-structured 10-minute conversation with a trained examiner; in analyses, we considered frequency of use. Any immediate repetition of “um” or “uh” in a sequence (e.g. “um” or “uh uh”) was excluded from frequency counts. Additional participant features considered in relation to the use of the filled pauses were nonverbal cognitive ability, language ability, general anxiety, social avoidance/social anxiety, hyperactivity, phonological memory, and Fragile X Mental Retardation Protein (FMRP) as measured in peripheral blood.

Results: Participants with FXS+ASD and participants with FXS-only did not differ in their frequency of use of either “um” or “uh”, but a significant negative relationship was observed between our continuous metric of ASD severity and both “um” (r = -.32) and “uh” (r = -.36) across the entire FXS group. We also found that use of “um” was significantly associated with language ability (r = .35) and social avoidance/social anxiety (r = -.49). No other participant features were correlated with the production of “uh”. Finally, ASD severity, language ability, and social avoidance/social anxiety accounted for 15.6% of the variance in “um” use, with language emerging as the only significant unique contributor in the model (β = 1.69, p = .02).

Discussion: A failure to use “um” may have important consequences for conversation, including disrupting turn taking between participants in the conversation. The failure of individuals with idiopathic ASD to use this type of filled pauses is thus thought to reflect an impairment in social communication. The present results suggest that “um” is not uniquely diagnostic of ASD in FXS and its use may reflect language ability rather than social-communication skill. More generally, these findings add to the growing literature about the validity of the ASD categorical diagnosis in FXS.

References/Citations: