2020 Gatlinburg Conference Poster Submission

Title: The Impact of the *FMR1* Premutation Phenotype on Mother-Child Synchrony in Fragile X Syndrome

Authors: Carly Moser, Laura Hahn, Ph.D., Jessica Klusek, Ph.D., CCC-SLP

Introduction: The quality of the mother-child interaction plays a key role in child development and is particularly relevant for children and adolescents with developmental disabilities, including fragile X syndrome (FXS), a disorder characterized by intellectual disability and behavioral features such as autism symptoms and hyperactivity. The parenting style of mothers of children with FXS is associated with child language and cognitive outcomes (Sterling, Warren, Brady, & Fleming, 2013; Wheeler, Hatton, Reichardt, & Bailey, 2007). The study of mother-child interaction quality in adolescence and adulthood in FXS is important, as parents of children with disabilities are likely to provide more intensive care for their children into adulthood (Seltzer, Floyd, Song, Greenberg, & Hong, 2011). While the importance of mother-child interaction quality in FXS has been examined during the childhood period, few studies have examined these interactions during adolescence. The influence of maternal characteristics on interaction quality is also poorly understood. FXS is a family condition with inheritance patterns affecting multiple family members, where mothers of children with FXS are genetic carriers of the *FMR1* premutation. The *FMR1* premutation can be associated with its own phenotypic consequences which include elevated risk for mental health disorders, executive function deficits, and pragmatic language difficulties (Cornish et al., 2005; Farzin et al., 2006; Hunter, Rohr, & Sherman, 2010; Losh et al., 2012). The present study aims to examine how the phenotypic features of the *FMR1* premutation affect the mother-child interaction of adolescents with FXS.

Method: Participants included 30 mothers with the *FMR1* premutation and their biological sons with full mutation FXS. Dyadic interaction (i.e., mother-child synchrony) was measured as an indicator of maternal-child interaction quality. Mother-child synchrony was coded from videotaped interactions of the dyad during two unstructured interactive play tasks, with each task lasting 10 minutes each (a puzzle and a box of toys) using the Anchor Points for Observational Rating of Mother-Adolescent Synchrony (Criss, Shaw, & Ingoldsby, 2003) coding scheme. One-minute segments were rated for synchrony quality and averaged across the total time (20 minutes), yielding an overall synchrony rating for the dyad (i.e. 0-9). Maternal pragmatic language was measured using the Pragmatic Rating Scale (PRS; Landa et al., 1992). The PRS is coded for various pragmatic violations from a 20-minute conversational sample with an interview about their life history. Maternal mental health characteristics were measured using the Beck Depression Inventory II (BDI-II; Beck, Steer, & Carbin, 1988) and the Beck Anxiety Inventory (BAI; Beck, Epstein, Brown, & Steer, 1988). Maternal executive function was indexed using the Hayling Sentence Completion Test (Burgess & Shallice, 1997). Child variables include IQ indexed by the Leiter International Performance Scale-Revised Brief IQ (Roid & Miller, 1997), autism severity measured by the Autism Diagnostic Observation Schedule-Second Edition (Lord et al., 2012), and severity of child problem behavior measured by the Child Behavior Checklist (Achenbach & Rescorla, 2001).

Results: Descriptively, the average rating for mother-child synchrony was 3.58 and scores ranged from 0 to 5. Overall, mothers and their children with FXS were rated well below the highest possible synchrony score of 9. Multiple regressions were performed to examine maternal pragmatics, depression symptoms, anxiety symptoms, and executive function as predictors of mother-child synchrony. Child IQ, autism severity, and severity of child problem behavior were added to each regression model as covariates. A significant curvilinear association was detected between maternal pragmatic language and mother-child synchrony, such that the overall model testing the combined influence of maternal pragmatics, child IQ, , autism severity and severity of child problem behavior on mother-child synchrony was significant ($F(5,24)=6.48$ $p=.006$, $R^2 = .57$). Maternal pragmatics accounted for significant variance of mother-child synchrony ($p=.<.001$, $\eta 2p = .38$). All other maternal characteristics were not significant predictors after controlling for child characteristics ($p's>.402$).

Discussion: Maternal pragmatic language skills were closely tied to the synchrony of mother-child interactions, such that the subset of mothers who showed elevated pragmatic difficulties also exhibited the most asynchronous interactions with their child. These findings highlight the need for a family-centered intervention approach in those with FXS that targets the needs of

all family members. Understanding specific maternal characteristics that influence the quality of the mother-child interaction (i.e. maternal pragmatic language skills) can help inform targeted family-centered intervention efforts.

References:

Achenbach, T. M., & Rescorla, L. (2001). *Manual for the ASEBA school-age forms & profiles: An integrated system of multi-informant assessment*. Aseba Burlington, VT:

Beck, A.T., Epstein, N., Brown, G., & Steer, R. A. (1988). *An inventory for measuring clinical anxiety: Psychometric properties. J*.

Beck, A. T., Steer, R. A., & Carbin, M. G. (1988). Psychometric properties of the Beck Depression Inventory: Twenty-five years of evaluation. *Clinical Psychology Review*, *8*(1), 77–100. https://doi.org/10.1016/0272-7358(88)90050-5

Burgess, P., & Shallice, T. (1997). Hayling Sentence Completion Test. *Suffolk: Thames Valley Test Company.*

Cornish, K., Kogan, C., Turk, J., Manly, T., James, N., Mills, A., & Dalton, A. (2005). The emerging fragile X premutation phenotype: Evidence from the domain of social cognition. *Brain and Cognition*, *57*(1), 53–60. https://doi.org/10.1016/j.bandc.2004.08.020

Criss, M. M., Shaw, D. S., & Ingoldsby, E. M. (2003). Mother-son positive synchrony in middle childhood: Relation to antisocial behavior. *Social Development*, *12*(3), 379–400. https://doi.org/10.1111/1467-9507.00239

Farzin, F., Perry, H., Hessl, D., Loesch, D., Cohen, J., Bacalman, S., … Hagerman, R. (2006). Autism Spectrum Disorders and Attention-Deficit/Hyperactivity Disorder in Boys with the Fragile X Premutation. *Journal of Developmental & Behavioral Pediatrics*, *27*(2), S137.

Hunter, J. E., Rohr, J. K., & Sherman, S. L. (2010). Co-occurring diagnoses among FMR1 premutation allele carriers. *Clinical Genetics*, *77*(4), 374–381. https://doi.org/10.1111/j.1399-0004.2009.01317.x

Landa, R., Piven, J., Wzorek, M. M., Gayle, J. O., Chase, G. A., & Folstein, S. E. (1992). Social language use in parents of autistic individuals. *Psychological Medicine*, *22*(1), 245–254. https://doi.org/10.1017/S0033291700032918

Lord, C., Rutter, M., DiLavore, P., Risi, S., Gotham, K., & Bishop, S. (2012). Autism diagnostic observation schedule–2nd edition (ADOS-2). *Los Angeles, CA: Western Psychological Corporation*.

Losh, M., Klusek, J., Martin, G. E., Sideris, J., Parlier, M., & Piven, J. (2012). Defining genetically meaningful language and personality traits in relatives of individuals with fragile X syndrome and relatives of individuals with autism. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, *159B*(6), 660–668. https://doi.org/10.1002/ajmg.b.32070

Roid, G. H., & Miller, L. J. (1997). Leiter international performance scale-revised (Leiter-R). *Wood Dale, IL: Stoelting*.

Seltzer, M. M., Floyd, F. J., Song, J., Greenberg, J. S., & Hong, J. (2011). Midlife and Aging Parents of Adults with Intellectual and Developmental Disabilities: Impacts of Lifelong Parenting. *American Journal on Intellectual and Developmental Disabilities*, *116*(6), 479–499. https://doi.org/10.1352/1944-7558-116.6.479

Sterling, A. M., Warren, S. F., Brady, N., & Fleming, K. (2013). Influences on Maternal Responsivity in Mothers of Children With Fragile X Syndrome. *American Journal on Intellectual and Developmental Disabilities*, *118*(4), 310–326. https://doi.org/10.1352/1944-7558-188.4.310

Wheeler, A., Hatton, D., Reichardt, A., & Bailey, D. (2007). Correlates of maternal behaviours in mothers of children with fragile X syndrome. *Journal of Intellectual Disability Research : JIDR*, *51*(Pt. 6), 447–62. https://doi.org/10.1111/j.1365-2788.2006.00896.x