Title: Examining Cognitive and Behavioral Development in Infants and Toddlers with Turner Syndrome

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Introduction: Turner syndrome (TS) is a non-inherited genetic syndrome present in females identified by partial or entire loss of the second X chromosome with an estimated prevalence of 1 in 2,500 births (Kinger & Wintergerst, 2015; Saenger, 1996). In addition to medical risks including growth delays and reproductive and cardiac problems, neuropsychological and behavioral difficulties exist. Girls with TS tend to have cognitive profiles that include relatively stronger verbal abilities as compared nonverbal skills, along with challenges in executive functioning (Grayholt et al., 2017). Behaviorally, differences in social awareness and functioning may be observed similar to those with autism spectrum disorder, along with anxiety and attention-deficit/hyperactivity symptomology (Hong, Dunkin, & Reiss, 2011). Current research focuses primarily on older females with a limited examination of the cognitive and social-behavioral functioning of infants and toddlers with TS. To address this gap, the current study was designed to examine the core cognitive/development, temperament, and adaptive behavior for infants with TS.

Method: The sample was comprised of 31 females with TS (23 with complete X monosomy, 8 mosaic), 53 typically developing females, and 54 typically developing males all approximately one year of age. Participants were administered the Mullen Scales of Early Learning (Mullen) to capture early verbal, nonverbal, and motor abilities. Parents completed the Carey Temperament Scales (CTS) are caregiver report questionnaires used to measure how often a child behaves in a specific way and the Vineland Adaptive Behavior Scales, Second Edition (Vineland-II) to examine adaptive skills.

Results: For infants with TS, overall mean Mullen scores were primarily within the average range for 12-month-old infants. On the Early Learning Composite approximately 22% scored at least one standard deviation below the mean. Similar to the Mullen, most of the parent ratings of adaptive behavior were within the expected range. Approximately 7% to 11% scored in the at-risk range on Communication, Daily Living and Socialization Skills, respectively, with 19% scoring in the at-risk range on the overall Adaptive Behavior Composite. In contrast, over 42% of infants received ratings that placed them in the at-risk range on the Motor Skills Scale. Caregiver ratings of temperament for the overall sample were within the average range at 12 months of age. Few group differences were noted at 12 months of age on the Mullen. Visual Reception, Receptive Language, Expressive Language, Fine Motor, and the Early Learning Composite for the infants with TS were indistinguishable from the typical male and female groups. The only Mullen scale that evidenced a significant group difference was Gross Motor, where the infants with TS scored significantly lower than the typical males, F(2, 125) = 4.66, p < .001.

Discussion: This study represents the first to examine the neurodevelopmental status of infants with TS using standardized measures of development, temperament, and adaptive behavior. Current findings indicate that infants with TS are primarily within the average range in adaptive behavior, temperament, and overall developmental functioning at 12-months. Given that many girls with TS experience a variety of cognitive and social-behavioral challenges as they move into the school-age years, it is no surprise that about 23% of the sample performed at least one standard deviation below the mean in their overall development, with higher rates of risk noted on receptive language, expressive language, and gross motor functions. Findings suggest that if difficulties are present, they may evolve over time, or perhaps, remain undetected clinical measures our at these early ages. Overall, there remains a need for further efforts around early identification and access to intervention services in this population that can be bolstered by continuing investigations of this developmental period for infants and toddlers with TS.
References/Citations:


