Title: A Comparison of Developmental Functioning in Infants and Toddlers with Genetic Disorders

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Introduction: Individuals with genetic disorders often demonstrate impairments across multiple areas of functioning. Some widely studied genetic disorders include Angelman syndrome, trisomy X syndrome (i.e., Triple X), Prader Willi syndrome, Williams syndrome, and Di George syndrome. While these disorders have separate genetic bases and distinct cognitive and behavioral profiles, some share symptoms such as impaired intellectual abilities, language delays, and/or motor deficits (Collet et al., 2009; Holm et al., 1993; Otter, Schrander-Stumpel, & Curfs, 2009; Williams et al., 2006). However, few studies have compared functioning in individuals with different genetic disorders. The current study aimed to investigate overall developmental functioning as well as specific developmental domains across genetic disorders in infants and toddlers.

Method: The sample of participants was extracted from a database of 2,459 infants and toddlers from EarlySteps, the State of Louisiana’s Early Intervention System. The Battelle Developmental Inventory, Second Edition (BDI-2) was used to measure developmental functioning, and the Baby and Infant Screen for Children with aUtIsm Traits—Part 1 (BISCUIT- Part 1) was used to gather demographic and relevant medical data. Individuals with Angelman syndrome (n = 10), Triple X syndrome (n = 12), Williams syndrome (n = 10), Prader Willi syndrome (n = 10), and Di George syndrome (n = 15) were included in the analyses, for a total sample size of 57 participants.

Results: An ANOVA was conducted to examine the relationship between genetic disorder group and developmental functioning. Significant differences were found in overall developmental functioning between groups [F(4, 52) = 8.43, p < .000, partial η² = .234]. Results from post hoc Scheffé tests revealed significant group differences between the Angelman and Prader Willi (p = .002), Angelman and Di George (p = .004), Triple X and Prader Willi (p = .006), and Trisomy X and Di George’s groups (p = .011). A MANOVA was conducted to further investigate group differences across five developmental domains (i.e., Adaptive, Personal-Social, Communication, Motor, and Cognitive). Using Pillai’s trace, there was a significant effect of group on domains, V = .936, F(20,204) = 3.12, p < .000. There were also significant differences across all five domains (p < .001). The Angelman syndrome group had the lowest developmental functioning for the person-social (M = 67.40, SD = 8.78), communication (M = 57.60, SD = 8.22), and motor subscales (M = 56.40, SD = 8.22). Both the Triple X (M = 61.17, SD = 11.34) and Angelman syndrome (M = 61.00, SD = 7.79) groups had the lowest adaptive functioning, and the Triple X group had the lowest cognitive functioning (M = 61.42, SD = 7.53). Further, the Prader Willi group had the highest functioning in the person-social (M = 89.09, SD = 15.55), personal-social (M = 91.60, SD = 15.07), communication (M = 78.10, SD = .71), and cognitive domains (M = 82.60, SD = 12.52), although the Di George group had the highest motor functioning (M = 86.13, SD = 19.21).

Discussion: Individuals in the Angelman syndrome group were found to have the lowest overall developmental functioning, while the Prader Willi syndrome group had the highest. These findings provide preliminary evidence of potential distinctions in functioning in young children with genetic disorders. Additional investigation of similarities and discrepancies between disorders with a genetic basis may serve to further inform treatment.

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