Title: What Might Explain Social Impairments in Children with Chromosome 22q11.2 Deletion Syndrome?

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Introduction: Autism spectrum disorders (ASD) are frequently reported in children with chromosome 22q11.2 deletion syndrome (22q) (Vorstman et al., 2006; Antshel et al., 2007; Fine et al., 2005), although no studies have used gold standard evaluations to diagnose ASD. It is unclear if the social impairments in 22q are better explained by underlying cognitive challenges. This study investigates whether specific cognitive abilities are related to the social impairments in 22q that frequently lead to ASD diagnoses.

Method: We performed a retrospective analysis of relevant data from 114 children, collected as part of a study designed to study the neurocognitive bases of spatiotemporal impairments in children ages 7-14 with 22q. The Social Communication Questionnaire (SCQ; Rutter et al., 2003), a screening tool for autism, was used to quantify social impairments. These were related to subtest scores from the Wechsler Intelligence Test for Children, 4th edition (WISC-IV; Wechsler 2003) to test our hypothesis that conceptual and linguistic delays contribute strongly to social impairments. Pearson’s correlation coefficient was calculated to determine the strength of relationship between variables.

Results: Mean age was 11.2±2.5 years. 50% were male. SCQ score correlated negatively with WISC-IV Verbal Comprehension Index (VCI; r=0.2, p=0.03) and WISC-IV VCI subscales Vocabulary (r=0.27, p=0.004) and Comprehension (r=0.23, p=0.02), but not Similarities (r=0.09, p=0.33). However, Perceptual Reasoning (r=0.07, p=0.5) and Processing Speed (r=0.1, p=0.27) composites were not related to SCQ scores.

Discussion: In children with 22q, social impairments (as measured by the SCQ) were related to communication and language abilities but not other WISC-IV domains. This suggests a specific role for cognitive abilities underlying communication in partially explaining the level of social functioning in children with 22q. Future study should compare a broad array of cognitive domains in 22q and idiopathic ASD to determine whether distinct intermediate phenotypes explain observable behavior and thus provide different targets for treatment.

References/Citations: