**Title:** Correlations between Proband and Paternal IQ and SRS Scores In De Novo 16p11.2 Deletion: A Dimensional Perspective

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**Introduction:** 16p11.2 is a genetic variation that confers increased risk for Autism Spectrum Disorder and comorbid intellectual disability. Previous research on de novo 16p11.2 deletion has found intact correlations between parent and proband IQ and Social Responsiveness Scale (SRS) scores, as well as other dimensions (Moreno-De-Luca, Evans & Ledbetter 2015). From this, and similar findings from other familial studies, researchers speculate that specific genetic variations do not necessarily precipitate abnormal functioning directly, but “shift” scores on these dimensions downwards from the family’s start point (Finucane, Challman, Martin & Ledbetter 2016). In other words, for families with on average higher IQ scores, this shift may not move probands into the clinical range, merely into the average or low average range. Conversely, probands with less protective genetic backgrounds will fall more often into clinical ranges (Finucane et al. 2016). This perspective dovetails well with the increasing emphasis on a dimensional approach to psychopathology (Insel & Cuthbert 2010; Finucane et al. 2016).

**Method:** Using the SFARI data set, the present study expands upon this research by examining correlations between proband IQ scores and paternal and maternal IQ scores in de novo 16p11.2 deletion independently. Specifically, correlations between probands and their mothers and fathers are examined separately, rather than using a bi-parental mean. It is important to note these findings and discussions are exploratory in nature, and that samples used in this analysis are relatively small (N=~30-55). Results should be replicated and interpreted with caution.

**Results:** This yields the surprising finding that maternal IQ scores are not significantly correlated with proband scores, while paternal correlations are robust. A similar pattern exists for SRS scores. These finding are particularly surprising given that maternal scores, especially for IQ, tend to correlate even more strongly than paternal IQ in typical populations. The present poster will explore this relationship in several ways. First, maternal and paternal correlations with probands will be compared to those of siblings. If this relationship is related to 16p11.2 deletion, sibling maternal correlations on these dimensions will be intact. Paternal and maternal correlations with probands and unaffected siblings will also be explored on other dimensions, including verbal and nonverbal IQ, BMI, and the Purdue Pegboard task.

**Discussion:** Broader implications of this surprising relationship will be discussed, such as how this finding builds upon previous research supporting and emerging dimensional take on the impact of genetic variations on outcomes, and the potentially protective influence of paternal IQ and SRS scores.

**References/Citations:**