Exploring social profiles of individuals with 16p11.2 deletion and duplication

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INTRODUCTION

- Copy number variation (CNV) of the 16p11.2 chromosomal region has been associated with a wide range of neurodevelopmental outcomes.
- Approximately 19% of pediatric duplication carriers and 26% of pediatric deletion carriers meet criteria for a diagnosis of autism spectrum disorder, and a majority of carriers exhibit some autistic features (Green Snyder et al., 2016; Hanson et al., 2015).
- Few studies have investigated specific patterns of social strengths and challenges in 16p11.2 CNV carriers. Mouse model studies of 16p11.2 deletion have reported a pattern of intact social approach with atypical social interactions (Yang et al., 2015).
- Recently, clinical phenotyping of a 16p11.2 duplication cohort has similarly revealed a possible social profile of intact social motivation in the presence of impacted social cognition (Green Snyder et al., 2016).
- Investigation of social behaviors in a large cohort of 16p11.2 duplication and deletion carriers may further validate this social profile.

METHOD

Participant data from 102 children with 16p11.2 deletions and 41 children with 16p11.2 duplications were extracted from the Simons VIP Phase 2 data (Simons VIP Consortium, 2012).

A repeated measures ANOVA was used to test whether mean *T*-scores differed on theoretically derived subscales of the Social Responsiveness Scale, Second Edition (SRS-2; Constantino & Gruber, 2012) within the 16p11.2 duplication and deletion groups.

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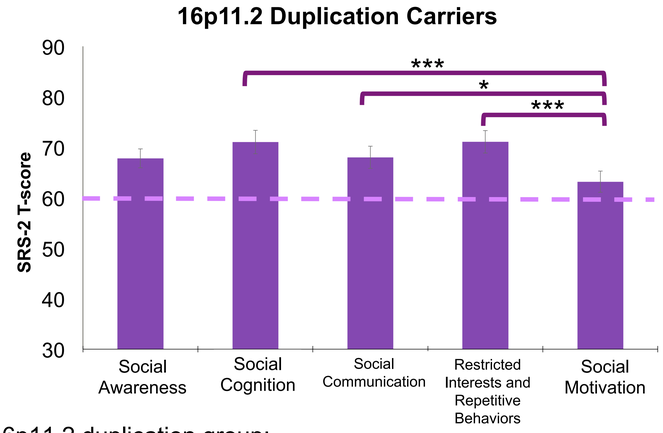
Social motivation represents a relative social strength for both 16p11.2 duplication and deletion carriers.

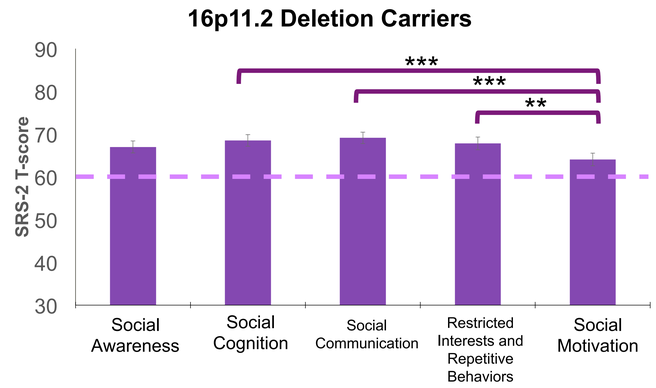
Social profiles identified in 16p11.2 CNV carriers are consistent with mouse model findings of intact social approach paired with atypical social interactions.

RESULTS



16p11.2 duplication group:

- Significant main effect of SRS-2 subscale, *Greenhouse-Geisser Adjusted* $F(3.10, 124.07) = 9.55$, $p < 0.001$, *partial* $\omega^2 = 0.17$.
- Follow-up paired *t*-tests among SRS-2 subscales using Dunn-Sidak adjustment revealed that the Social Motivation subscale had a lower mean *T*-score, suggestive of better developed social motivation abilities, compared to compared to Social Cognition (adjusted $p < 0.001$), Social Communication (adjusted $p = 0.050$), and RRB (adjusted $p < 0.001$) subscales.



16p11.2 deletion group:

- Significant main effect of SRS-2 subscale, *Greenhouse-Geisser Adjusted* $F(3.18, 321.56) = 8.10$, $p < 0.001$, *partial* $\omega^2 = 0.06$.
- Follow-up paired *t*-tests among SRS-2 subscales using Dunn-Sidak adjustment revealed that the Social Motivation subscale had a lower mean *T*-score compared to Social Cognition (adjusted $p < 0.001$), Social Communication (adjusted $p < 0.001$), and RRB (adjusted $p = 0.013$) subscales.