Examining the broader autism phenotype in the context of genetic etiology

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Broader autism phenotype (BAP):

- Subclinical traits associated with ASD symptoms and social impairment (Bolton et al., 1994)
- BAP features are reduced in simplex (1 child with ASD) relative to multiplex (2+ children with ASD) parents (Gerdts et al., 2013)
- Disparity across BAP measures → need convergence across multiple informants and BAP traits (Davidson et al., 2014)

Objective: To examine the extent by which parental BAP is associated with children's genetic etiology and children's social skills.

Hypothesis: More elevated BAP symptoms in parents who also have the ASD-risk genetic event that their child inherited.

Child has **Inherited** CNV or SNV > Child has ***de novo*** CNV or SNV or Child has **No Event** (CNV or SNV)

Method:

- Local participants from Simons Simplex Collection (SSC, *n* = 163) and a genetics-first study (TIGER, *n* = 54)
  - o Excluded children with a combination of *de novo* and inherited events (*n* = 32) and children without an ASD diagnosis (*n* = 29)
- Child and parental copy number variant (CNV) and single nucleotide variants (SNV) were confirmed:
  - o Previously (SSC; Sanders et al., 2015; 2012) or
  - o Via local sequencing and genetic testing report
- <u>BAP Measures</u>:
  - o Broader Phenotype Autism Symptom Scale (BPASS; Dawson et al., 2007)
  - o Broader Autism Phenotype Questionnaire (BAPQ; Hurley et al., 2017)
  - o Social Responsiveness Scale – II (SRS-2; Constantino & Gruber, 2012)

Subclinical ASD traits in parents **do not differ** between parents whose child did or did not inherit a ASD-risk genetic event.

There was **agreement** between **self-, other-, and clinician**-reported BAP.

Clinician measured parental BAP was broadly related to **children's adaptive skills.**

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	de novo	Inherited	No known event	Group difference [§]
N Individuals **N families**	28 17	26 15	14 7	
Female:Male	16:12	13:13	7:7	χ^2=.34, *p* = .85
Age Mean (SD) **Range**	45.7 (11.2) 33-80	43.3 (6.0) 31-56	45.3 (5.3) 37-56	*F*(2,62) = .51, *p*=.60
FSIQ Mean (SD) **Range**	128.8 (44) 88-250	113.9 (12.5) 98-134	117.9 (12.4) 104-142	*F*(2,62) = 1.7, *p*=.18
BPASS Total Mean (SD)	1.78 (.3)	1.83 (.4)	1.89 (.4)	*F*(2,66) = .46, *p*=.63
• Social Motivation	2.36 (.9)	2.31 (.7)	2.50 (.9)	*F*(2,66) = .42, *p*=.79
• Expressiveness	1.28 (.3)	1.33 (.4)	1.34 (.3)	*F*(2,66) = .20, *p*=.82
• Conversational skills	1.27 (.4)	1.29 (.5)	1.32 (.3)	*F*(2,66) = .08, *p*=.92
• Range of interests/Flexibility	2.2 (.6)	2.4 (6)	2.38 (.5)	*F*(2,66) = .96, *p*=.39
BAPQ Raw	95.9 (27.5)	96.5 (19.2)	100.1 (22.3)	*F*(2,62) = .15, *p*=.86
SRS-2 Total T	44.4 (8.5)	46.9 (8.1)	49.3 (9.5)	*F*(2,66) = 1.65, *p*=.20

[§] One-WAY ANOVAs conducted using R 3.3.1 aov() with Tukey correction for multiple comparisons