

# 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 (Gerds 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.



**Method:**

- Local participants from Simons Simplex Collection (SSC, *n* = 163) and a genetics-first study (TIGER, *n* = 54)
  - 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:
  - Previously (SSC; Sanders et al., 2015; 2012) or
  - Via local sequencing and genetic testing report
- **BAP Measures:**
  - Broader Phenotype Autism Symptom Scale (BPASS; Dawson et al., 2007)
  - Broader Autism Phenotype Questionnaire (BAPQ; Hurley et al., 2017)
  - 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.

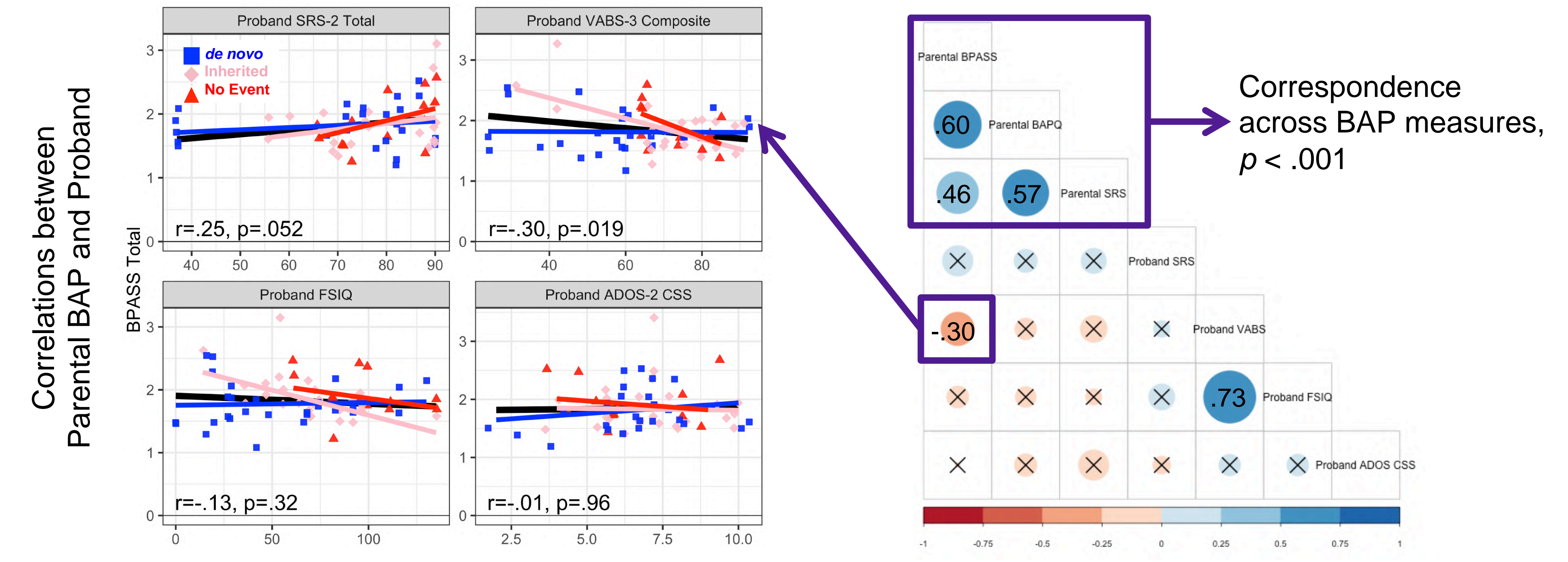
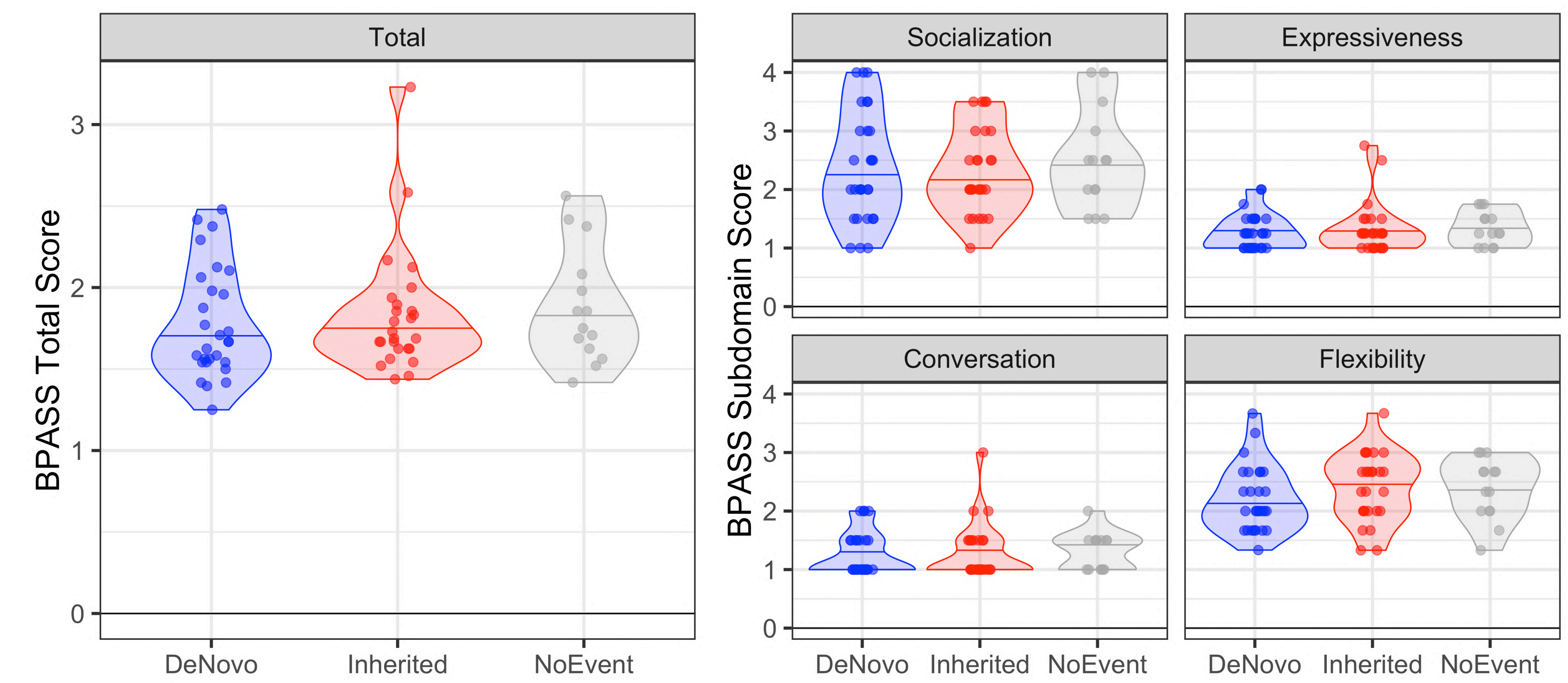
Clinician measured parental BAP was broadly related to children's adaptive skills.

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	<i>de novo</i>	Inherited	No known event	Group difference §
<b>N Individuals</b>	28	26	14	
<b>N families</b>	17	15	7	
<b>Female:Male</b>	16:12	13:13	7:7	$\chi^2=.34, p=.85$
<b>Age Mean (SD)</b>	45.7 (11.2)	43.3 (6.0)	45.3 (5.3)	$F(2,62) = .51, p=.60$
<b>Range</b>	33-80	31-56	37-56	
<b>FSIQ Mean (SD)</b>	128.8 (44)	113.9 (12.5)	117.9 (12.4)	$F(2,62) = 1.7, p=.18$
<b>Range</b>	88-250	98-134	104-142	
<b>BPASS Total Mean (SD)</b>	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)	$F(2,66) = .96, p=.39$
<b>BAPQ Raw</b>	95.9 (27.5)	96.5 (19.2)	100.1 (22.3)	$F(2,62) = .15, p=.86$
<b>SRS-2 Total T</b>	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



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