Facial Identity Recognition and the Broader Autism Phenotype

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Broader Autism Phenotype (BAP)

Expression of 'autistic-like' traits in non-autistic family members, in a milder, but qualitatively similar form. Folstein & Rutter, 1977; Bolton, MacDonald, Pickles & Rios, 1994

Parents of ASD children described as "serious minded", "mildly obsessive", "perfectionist", and "with an intense interest in abstract ideas".

Kanner, 1943.



Why study the BAP?

Wide variability *within* ASD. Different characteristics are likely to have different causes.

 Studying the BAP provides insight into which characteristics of ASD *might* have a genetic basis.
Guide the search for genes?

- Help to establish meaningful subtypes?

- Opportunity to study the clustering of autistic features in a non-autistic population.



Previous research: BAP characteristics

- Increased rates of stereotyped behaviours
- Social and communication deficits

Piven, Palmer, Jacobi, Childress & Arndt, 1997; Piven, Palmer Landa, Santangelo, Jacobi & Childress, 1997.

Broader Autism Phenotype Questionnaire

-Traits stronger in males than females?

-Happe, Briskman & Frith, 2001; Hughes, Leboyer, Bouvard, 1997.

- Not all relatives of ASD individuals show BAP traits



Face Recognition



Evidence that face recognition skills run in families in:

- Typical population (Wilmer et al. 2010; Zhu et al., 2010)
- Congenital prosopagnosia (Duchaine et al, 2007; Schmalzl et al. 2008)



Heterogeneity of facial identity recognition in ASD

• Some children perform at age-appropriate levels but others are severely impaired



Do familial factors contribute to ASD individuals' level of face recognition skill?

Three questions:

1) Is face recognition impaired in the BAP?

2) Is face recognition ability associated with other common BAP traits?

Experiment 1

3) Is face recognition ability in parent associated with face recognition ability in the proband?

Experiment 2



Experiment 1

Question 1: Is face recognition impaired in the BAP?

33 parents of ASD children completed a standardized test of memory for faces.

Cambridge Face Memory Test (CFMT). Duchaine & Nakayama, 2006.



Internal reliability: $\alpha = 0.89$ (Bowles et al., 2009).



Experiment 1 Results of CFMT

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ASD parents scored significantly below average:

$$t(32) = -2.89, p < 0.01.$$

Mothers: t (18) = -1.76, p = 0.10Fathers: t (13) = -2.31, p = 0.04

Variability within parent group





Variability within parent group

Unsurprising... ASD is a highly heterogeneous condition.

Question 2: Is face recognition ability associated with other common BAP traits?



Broader Autism Phenotype Questionnaire (BAPQ)

Hurley, Losh, Parlier, Reznick & Piven, 2007

Three personality traits thought to be more common in the BAP than in the typical population:

Aloof personality: lack of interest in or enjoyment of social interaction. Pragmatic language impairment: deficits in the social aspects of language. Rigid personality:little interest in change, or difficulty adjusting to change.

BAPQ: 36 item self-rating questionnaire, on a scale of 1 - 6.

Adolphs et al (2008): aloof ASD parents were worse at facial *emotion* recognition than non-aloof ASD parents.



Experiment 1

Broader Autism Phenotype Questionnaire (BAPQ)

Results:

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- Fathers significantly above mean score of typical adults on **aloof personality** and **pragmatic language**.
- Mothers scored within normal range.
- -Wide range of scores.

No associations between BAPQ subscales and CFMT standardized scores.



Question 3: Is face recognition ability in the BAP related to face recognition ability in the proband?

Experiment 2:

20 ASD children and 33 parents (19 mothers, 14 fathers) completed equivalent tasks of facial identity recognition *and* a control task.



Experiment 2 Method

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Children: 2-alternative forced choice (AFC) matching task.

Adults: 4-AFC matching task.

Control task: shoe matching

Used scores from typically developing samples to standardize scores according to performance on control task.





Experiment 2

Is the face recognition ability of the parent and the proband associated *after* controlling for cognitive ability?



Conclusions

1) Is face recognition impaired in the BAP?

✓ Heterogeneous face recognition in ASD parents (as a group, impaired on face memory but not on face matching).

2) Is face recognition ability associated with other common BAP traits?

▶ No evidence, according to the BAPQ.

3) Is face recognition ability in parent associated with face recognition ability in the proband?

 Results suggest an association between proband and mothers' ability to recognise faces, that is not attributable to general cognitive ability.



Outstanding Questions

- 'Familial factors'...
 - Genetic?
 - Environmental?

- Relationship between 'familial factors' and other risk factors in ASD? e.g.

- early experience with faces
- interest in social stimuli



Final comment on methodology

We all know ASD is a heterogeneous condition...

therefore it is extremely important to pay attention to this variability, and not simply analyse group means.

When investigating the BAP, it is important to measure the equivalent skill level in the proband.



Acknowledgements

Thanks to Jon Brock, Romina Palermo, Mike Burton & David White.

I am particularly grateful to all my participants, and their families and schools.



Southside Montessori School



Autism Spectrum Australia



Macquarie University Special Education Centre

C. E. Wilson, J. Brock, A. M. Burton & R. Palermo. In press. Facial identity recognition in the broader autism phenotype. *PloS ONE.*

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	Mothers	Fathers	Control mean	Suggested cut-off
	(N = 19)	(N = 14)	(females, $N = 32$; males, $N = 32$)	for presence of BAP traits
Total	2.48 (0.59)	3.19 (0.66) **	2.74 (0.55)	3.15
	1.42 – 3.61	2.06 - 4.42		
Aloof	2.48 (0.81)	3.30 (0.86) **	2.75 (0.78)	3.25
	1.17 – 4.23	2.17 - 5.33		
	2.21 (0.60)	2.88 (0.73) **	2.45(0.51)	2.75
Pragmatic	1.17 – 3.58	1.67 - 2.27	2.45 (0.51)	2.75
D 1 11	2.67 (0.81)	3.39 (0.76) *		
Rigid	1.67 – 4.33	2.17 - 4.33	3.02 (0.55)	3.5

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