Detecting Cognitive Endophenotypes for Autism Using a General Population Twin Family Sample

R.A. Hoekstra1,2, M. Bartels2, G.F. Estourgie-van Burk2, S. Baron-Cohen1, D.I. Boomsma2

1Autism Research Centre, Section of Developmental Psychiatry, University of Cambridge, Cambridge, UK;
2Department of Biological Psychology, VU University, Amsterdam, The Netherlands

Contact: rah58@medschl.cam.ac.uk

Background

Autism spectrum conditions (ASC) show an uneven cognitive profile:
• Relative peak performance on Block Design1,2 and Embedded Figures3.
• Poor performance on cognitive flexibility4, language2 and social cognition5.

Studies of the Broader Autism Phenotype show that a similar uneven cognitive profile may also characterize first-degree relatives, suggesting familial influences6.

Recent studies indicate that autistic traits are continuously distributed in the general population2 and that individual differences in these traits show substantial heritability6.

Methods I

Participants
18-year-old twin pairs (n = 197 pairs) and their siblings (n = 96; mean age 18.29 years, SD = 2.11) , who are all registered in the Netherlands Twin Register.

Methods II

Objectives
I. To explore the association between autistic traits and performance on cognitive tests in a general population sample.
II. To examine whether this association is due to shared genetic or environmental influences.

A genetic link between certain cognitive abilities and autistic traits would point to these being promising endophenotypes for autism.

Participants
18-year-old twin pairs (n = 197 pairs) and their siblings (n = 96; mean age 18.29 years, SD = 2.11) , who are all registered in the Netherlands Twin Register.

Results I

Poor performance on the Wechsler Vocabulary subtest (VOC) and Semantic Verbal Fluency (SVF) predicted SOC difficulties ($\chi^2 = 7.52$, df = 1, $p < .01$ and $\chi^2 = 6.35$, df = 1, $p = .01$).

Performance on the Block Design (BD) and Information (INF) subtests of the Wechsler Intelligence scale both predicted ATT ($\chi^2 = 5.41$, df = 1, $p = .02$ and $\chi^2 = 6.25$, df = 1, $p = .01$).

Descriptives

<table>
<thead>
<tr>
<th>N</th>
<th>Mean</th>
<th>SD</th>
<th>Sex effect</th>
<th>Age effect</th>
</tr>
</thead>
<tbody>
<tr>
<td>VOC</td>
<td>453</td>
<td>10.18</td>
<td>2.71</td>
<td>ns</td>
</tr>
<tr>
<td>SVF</td>
<td>453</td>
<td>10.05</td>
<td>2.79</td>
<td>p &lt; .001</td>
</tr>
</tbody>
</table>

Correlations and cross correlations are higher in MZ twins than in 1st degree relatives.

Conclusions

General population autistic traits covary with the same cognitive strengths and weaknesses as observed in clinical ASC.

The relationship between autistic traits and the uneven cognitive profile is partly genetic, suggesting these could be used as endophenotypes for autism.

References


Funding
Netherlands Organisation for Scientific Research (NWO Rubicon; 575-25-006; SPI 56-464-14192).