

# The Boundaries of the Cognitive Phenotype of Autism: Theory of Mind, Central Coherence and Ambiguous Figure Perception in Young People with Autistic Traits

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**Abstract** Theory of Mind, Weak Central Coherence and executive dysfunction, were investigated as a function of behavioural markers of autism. This was irrespective of the presence or absence of a diagnosis of an autistic spectrum disorder. Sixty young people completed the Social Communication Questionnaire (SCQ), false belief tests, the block design test, viewed visual illusions and an ambiguous figure. A logistic regression was performed and it was found that Theory of Mind, central coherence and ambiguous figure variables significantly contributed to prediction of behavioural markers of autism. These findings provide support for the continuum hypothesis of autism. That is, mild autistic behavioural traits are distributed through the population and these behavioural traits may have the same underlying cognitive determinants as autistic disorder.

**Keywords** Autistic spectrum · Continuum · Theory of Mind · Central coherence · Ambiguous figures

Autism is a heritable disorder that affects the pre and postnatal development of the brain (Medical Research Council 2001). Although the biological basis of the

disorder is firmly established, the precise mechanisms by which it develops are not. In the absence of biological markers, autistic disorder remains behaviourally defined. People with autism necessarily have deficits in socialisation, communication and repetitive/restricted patterns of behaviour (American Psychiatric Association 2000). Kanner's initial descriptive definition of autistic disorder was based on two core features 'extreme autistic aloneness' and 'insistence on sameness' (Kanner 1943). There is now increasing recognition that there is an autistic spectrum (Wing 1988). That is, that the expression of autism can vary across a number of dimensions. Psychological research has focused on identifying a single universal and specific psychological cause for autism that can unite this distinctive constellation of behaviours. There are three main candidates namely, Theory of Mind (Baron-Cohen 1995), Weak Central Coherence (Happé and Frith 2006), and executive dysfunction (Russell 1997). These theories are based on the fact that people with autism perform differently to matched groups of neurotypical people on certain measures (e.g., false belief, block design test and planning tasks respectively).

The relationship between these psychological variables and behaviour, in autism and the general population, is relevant to the debate about whether autism is an extreme on a continuum of autistic traits or is a qualitatively distinct pathological state that has determinants that are not present in the general population. There is growing evidence that autistic behavioural traits can be measured quantitatively in the general population and that these traits form a normal distribution (single distribution Baron-Cohen et al. 2001; fractionated triad Happé et al. 2006). It is not established however, whether these behavioural traits have the same underlying cognitive substrate in the general population as in the diagnosed disorder.

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The established cognitive characteristics of autism present an opportunity to test the continuum hypothesis of autism outside diagnostic boundaries. The question is whether the cognitive deficits in Theory of Mind, the presence of Weak Central Coherence and executive dysfunction occur as a function of autistic behavioural traits or solely as a function of diagnosis. That is, whether people in whom the symptoms of autism are severe and form the most prominent difficulty, are qualitatively different in their underlying cognition to people who fall outside the diagnostic boundaries due to the presence of other conditions or less severe symptoms. Should it be shown that these cognitive characteristics extend beyond the boundaries of the disorder then they could be used as alternative phenotypes in behavioural genetic and brain imaging studies.

One study has examined this issue. Kunihiro and colleagues (2006) tested typical Japanese adults on Theory of Mind and central coherence tasks to see if they co-varied with scores on the Autistic Spectrum Quotient (AQ, Baron-Cohen et al. 2001). They found they did not. This may be due to the comparatively low level of autistic traits in this group and small sample size.

In this study our sample is drawn from young people in Scotland age 13 to 22, who receive additional support for learning. It was hypothesised that although pupils can receive additional support with learning for a wide range of reasons, this sample would nevertheless include a higher proportion of people with autistic behavioural traits than in the general population.

## Methods

### Participants

Participants for this research were drawn from an on-going research project in the Division of Psychiatry at the University of Edinburgh, the Edinburgh Co-morbidity study. The participants in the Edinburgh Co-morbidity study are people who are at slightly enhanced risk of mental health problems due to additional learning support needs. The Edinburgh Co-morbidity study employs the variables that were found to be useful in predicting subsequent illness in the Edinburgh High Risk study of schizophrenia (Miller et al. 2002). The Co-morbidity study will determine whether there are young people whose learning needs are in fact due to the early stages of a severe form of schizophrenic illness (Johnstone et al. 2007).

The sampling frame for the Co-morbidity study consisted of young people between the ages of 13 and 22 receiving additional support for learning in Scotland. Young people were identified and recruited to the Co-morbidity study through their schools or colleges.

As part of the protocol for the Edinburgh Co-morbidity study, the young people were screened with the Social Communication Questionnaire (SCQ). The SCQ (Berument et al. 1999) is completed by the parent or carer. The SCQ is based on the Autism Diagnostic Interview algorithm. A score over 15 indicates probable Pervasive Developmental Disorder and a score below 15 indicates that the participant is probably not on the autistic spectrum. The cut-off of 15 for differentiating Pervasive Developmental Disorders from other diagnoses has a specificity of 0.75 and a sensitivity of 0.85.

Participants were selected from those who had completed the SCQ as part of the Edinburgh Co-morbidity study. Sixty participants were recruited. There were 34 participants with SCQs greater than or equal to 15 and 26 participants with SCQ scores less than 15.

### Selection of Measures

Theory of Mind was measured using the first and second order false belief tests. These tests were chosen because of the very high level of evidence that people with autism perform differently to control groups on these measures (Yirmiya et al. 1998)

Central coherence was measured using the block design test in the segmented and unsegmented conditions. Again, there is strong evidence that people with autism show strength on the block design test and receive less benefit from the segmentation of the designs than the control subjects (Shah and Frith 1993).

A second central coherence measure was used, visual illusions (Happé 1996, but see Ropar and Mitchell 1999, 2001). Including a second measure of central coherence provides the opportunity to compare performance on the two measures and verify whether they are indeed measuring a common cognitive domain. Visual illusions were also included because they involve simple perceptual judgments. It was therefore expected that performance on the visual illusions would be unrelated to IQ.

The final test used was an ambiguous figure. Sobel and colleagues (Sobel et al. 2005) found that children with autism were less likely to reverse an ambiguous figure than control participants. Reversals of the ambiguous figure can be construed as a measure of cognitive flexibility. It was expected that performance on this test would also be unrelated to IQ.

The participants also completed two subtests of the appropriate intelligence test (WISC-III, Wechsler 1992; or WAIS-III, Wechsler 1999). The subtests chosen were the vocabulary subtest and the digit span subtest. These were chosen primarily as a control for the Theory of Mind task as it was thought that verbal ability as measured by

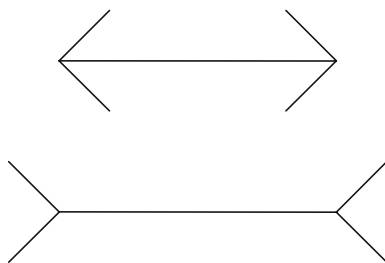
vocabulary score and short-term memory, as measured by the digit span task are the variables most likely to affect performance on this task. The block design task is its own control as the independent variable is the difference between conditions. The other two tasks, visual illusions and ambiguous figures, are simple perceptual judgments not anticipated to depend on general intelligence.

## Materials

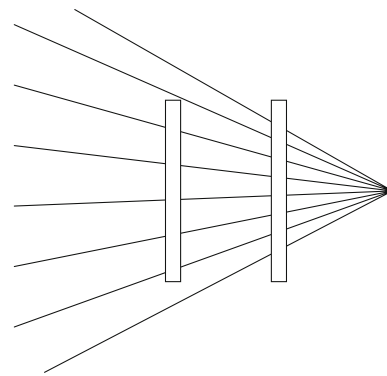
Testing took between three quarters of an hour to an hour, depending on the needs of the participants. Participants were able to take a break if required.

The researcher was blind to the SCQ score and the diagnosis of participants. The participants were asked to complete the following tests.

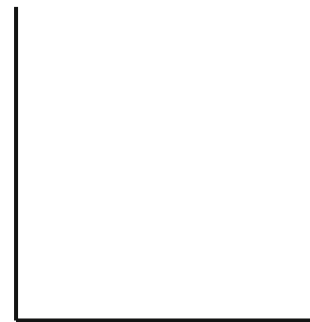
1. Visual illusions. The illusions used were the Muller-Lyer, the Ponzo and the Hat illusions (Figs. 1–3). The illusions were presented on laminated paper. They were printed in black on a white background. The dimensions of the stimuli were: Ponzo 10 cm by 8 cm, Muller-Lyer 17 cm by 12 cm, Hat 6 cm by 6 cm. Each illusion had its own control stimulus, which consisted of lines of the same dimensions as the illusion but without the illusion-generating context. This was to test the accuracy of participant's length judgments and motivation. Participants were shown all three illusions and the control stimuli in a fixed order of presentation.
2. Ambiguous figures (cat/swan). The participants were shown an ambiguous figure that had two possible interpretations. This figure was the cat/swan as used by Ropar, Mitchell et al. (2003). The figure was presented in black on a white background on laminated paper. The dimensions of the stimulus were 12 by 11 cm. The procedure followed that used by Gopnik and Rosati (2001). The number of informed reversals seen in a 1 min exposure was recorded.
3. First order false belief test. The first order false belief task used was a location change task (Baron-Cohen et al. 1985). The researcher acted out a short story



**Fig. 1** The Muller-Lyer illusion

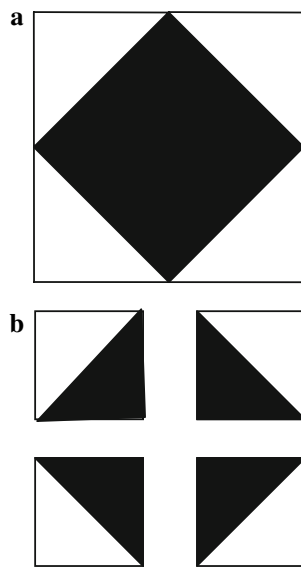


**Fig. 2** The Ponzo illusion



**Fig. 3** The Hat illusion

- using two model people and two boxes. The researcher then asked the false belief question and two control questions.
4. Second order false belief test. The second order false belief task was another scenario, this time acted out in a model village. The village contained a park with a roundabout and see saw, two houses, a tree, an ice cream van and a church. The church was situated at the other end of the village to the park. To complete this task more sophisticated reasoning was required in the form of 'John thinks that Mary thinks that...'. The format was based on Baron-Cohen (1989).
  5. Block design subtest in segmented/unsegmented conditions. The participants were given 4 blocks. The blocks had two white sides, two black sides and two black and white sides. After being given one practice design, they were asked to copy 8 patterns similar to that shown in Fig. 4a. The participants had to make the top faces of the blocks into the pattern shown. The designs were presented one at a time and the time taken to complete each design was recorded. Then the participants were given nine more patterns to copy. This time the designs were segmented as shown in Fig. 4b. There was no time limit. The time taken to complete each design was recorded.



**Fig. 4** (a) An unsegmented design. (b) A segmented design (after Shah and Frith 1993)

6. Digit span and vocabulary tests. Participants were given the digit span and vocabulary subtests of the appropriate Wechsler intelligence test. Participants of 16 or under were tested on the Wechsler Intelligence Scales for Children, third edition (WISC-III Wechsler 1992) and participants older than 16 on the Wechsler Adult Intelligence Scales, third edition (WAIS-III Wechsler 1999).

The raw scores on these tests were transformed into scaled scores to give an estimate of the level of intellectual functioning of the participants.

The order of presentation of the tests was the same for all participants.

**Socio-economic Status**

The socio-economic status of our participants was estimated using the ACORN postcode classification system (CACI 2003). The ACORN system is a geo-demographic classification system used in marketing. It allows comparison of the relative socio-economic status of our participants based on their postcode. Further details of the classification system can be found at <http://www.caci.co.uk/acorn/>

**Ethical Approval**

The Multi Centre Research Ethics Committee for Scotland gave a favourable ethical opinion regarding this research. NHS Lothian —Primary Care Organisation’s Research and

Development Committee Board also gave local agreement to the research proposal. The researcher held an Honorary Contract with the Lothian Primary Care NHS Trust for the duration of this project.

**Results**

The total sample consisted of 60 young people. There were 45 males and 15 females. Fifty six percent of the participants lived in areas defined by ACORN as ‘hard pressed’ compared with 22.4% of the total UK population.

Table 1 shows the characteristics of the participants and their performance on the interval measures.

Table 2 shows their performance on the categorical measures.

A forward stepwise binary logistic regression was performed. It was possible to create an equation that significantly improved prediction of whether participants had an SCQ score of 15 or over, compared to a constant only model. The number of participants included was 60. Thirty-four had SCQ scores of 15 or over and 26 had SCQ scores of less than 15. Predictors were entered based on the most significant score statistic with a p of 0.05 or less and were removed if the p of the –2 log likelihood test was greater than 0.10. The final model contained a constant. The block design difference score entered the model first (chi-squared = 5.19, *p* = 0.02) number of reversals of the ambiguous figure entered the model next (chi-squared = 6.75, *p* = 0.01). The only other variable that met criteria was the second order false belief question as a categorical variable (chi-squared = 4.39, *p* = 0.04). The Hosmer and Lemeshow test gives a Chi square value of 8.20 for the full model, *p* = 0.41 indicating the model is an adequate fit to the data. The coefficient for the block design difference

**Table 1** Participant characteristics

	Mean	Standard deviation	Range
Age (years:months)	16:6	1:9	13:9–22:4
Vocabulary scaled score	5.83	2.98	1–15
Digit span scaled score	6.15	2.88	1–13
Block design	16.90	19.47	–3.97–101.50
No. of revs/min	1.20	1.74	0–6

**Table 2** Performance on categorical measures

	Percentage of participants
Passing 1st order false belief	93.3
Passing 2nd order false belief	48.3
Succumbing to all illusions	41.7

score was  $B = 0.06$ ,  $SE = 0.02$ ,  $p = 0.01$ , odds ratio = 1.06. For the number of reversals  $B = 0.40$ ,  $SE = 0.18$ ,  $p = 0.03$  odds ratio = 1.49 and for failing the second order false belief question  $B = -1.31$ ,  $SE = 0.65$ ,  $p = 0.04$  odds ratio = 0.27. Increasing block design difference scores produce the greatest increase in odds of not having behaviours characteristic of autism. For the number of reversals and the 2nd order false belief question, passing the false belief question and seeing more reversals produce a small but significant increase in the odds of not having behaviours characteristic of autism. The above model correctly categorises 72% of cases. A model containing only a constant categorises 55.9% of cases correctly.

The variables digit span scaled score, vocabulary span scaled score, age in months, gender and ACORN category all failed to meet criteria to enter the model. No interaction terms increased the performance of the model. The ‘number of illusions seen’ by participants also made no contribution to the model. There was also no relationship between the visual illusions and block design test score of the participants indicating that they are not measuring a common cognitive domain. The relationship between the two measures is shown in the graph below (Fig. 5). The error bars indicate the 95% confidence interval.

In summary, these results indicate that the cognitive measures 2nd or false belief, block design and ambiguous figure reversals in the model are useful in discriminating young people with behaviours characteristic of autism from those who do not. They contribute additively to discriminating the SCQ status of participants.

Eighteen of the participants in this study informed us they had a diagnosis of autism, Asperger disorder, semantic pragmatic disorder or had been told they had

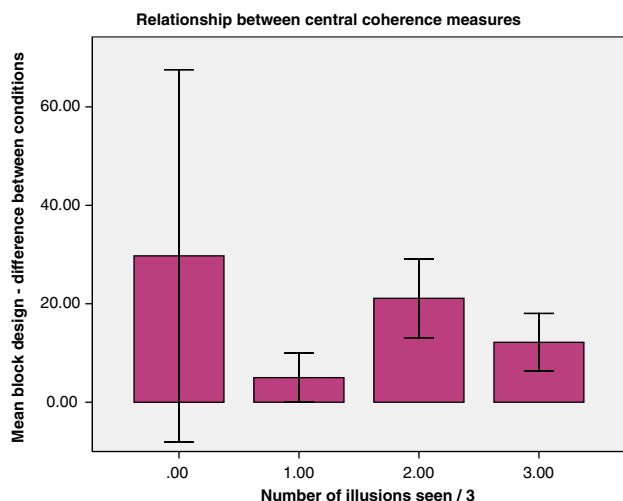


Fig. 5 Graph showing relationship between the two measures of central coherence

Table 3 Diagnoses of participants

	Dyslexia	ADHD	Autistic disorder/AS/semantic pragmatic disorder	Epilepsy	Cerebral palsy	Dyspraxia	Intellectual impairment	Other physiological disorder	No diagnosis	Total
One diagnosis only	6	2	14	2	3	1	2	4	16	50
Additional Diagnoses		2	2	1		1	1			3
Dyslexia										
ADHD										
Autistic disorder/AS/semantic pragmatic disorder										
Epilepsy								1		1

‘autistic tendencies’ before they joined the study. They diagnoses reported by participants are shown in Table 3.

A logistic regression equation was performed on this data. It was possible to create an equation that significantly improved accuracy of prediction of whether participants had a diagnosis related to autism or not, over a constant only model. The number of subjects included was 60. Predictors were entered based on the most significant score statistic with a  $p$  of 0.05 or less and were removed if the  $p$  of the  $-2$  log likelihood test was greater than 0.10. The final model contained a constant. The second order false belief question as a categorical variable entered the model first (chi-squared = 4.48,  $p = 0.03$ ) and then the block design difference score (chi-squared = 5.23,  $p = 0.02$ ). The Hosmer and Lemeshow test gives a Chi square value of 7.60 for the full model,  $p = 0.47$  indicating the model is an adequate fit to the data. The coefficient for failing the second order false belief question was  $B = 1.59$ ,  $SE = 0.65$ ,  $p = 0.01$ , odds ratio = 4.91. For the block design difference score  $B = -0.04$ ,  $SE = 0.02$ ,  $p = 0.058$  odds ratio = 0.96. Digit span scaled score, vocabulary span scaled score, age in months, gender and ACORN category all failed to meet criteria to enter the model. No interaction terms increased the performance of the model.

A constant only model correctly classifies 70% of the cases and the full model 71.7%. This model is not as strong the first model based on SCQ score. Examination of the predicted group membership reveals that all of the participants predicted not to have a diagnosis of ASD in the second model are also predicted not to have behaviours characteristic of autism in the first model. The second model misclassifies many of the remaining participants. We would suggest that this is because there are many participants in this group who have no diagnosis of autism but have high levels of related cognitive and behavioural features.

It can be seen from Table 3 that some of the participants reported diagnoses of both ADHD and autistic disorder. To disambiguate these reported diagnoses, all of the participants in this study were given the Autism Diagnostic Observation Schedule (ADOS-G, Lord et al. 2000) by a psychiatrist specifically trained in its application. Only four of our sixty participants met criteria for autistic disorder. An additional three met criteria for an Autistic Spectrum Disorder.

Forty-two of our participants have also taken part in stage three of the Edinburgh

Co-morbidity study. For these participants we have a measure of their full scale IQ. It is therefore possible to check how good an estimate the subtest scaled scores are for the full scale IQ of participants. The correlation coefficients are shown in Table 4.

It can be seen that vocabulary scaled score provides the best estimate for full scale IQ and verbal IQ in our

**Table 4** Pearson’s correlation coefficients between subtests scores and full scale IQ tests

	Full scale IQ	Verbal IQ	Performance IQ
Digit span scaled score ( $n = 42$ )	0.31	0.31	0.24
Vocabulary scaled score ( $n = 42$ )	0.75	0.82	0.48

participants. The vocabulary scaled scores and full-scale IQ scores are highly correlated. The fact that vocabulary scaled score does not contribute significantly to either of the above models shows that general intelligence is not a mediating factor in whether behaviours characteristic of autism are present or not.

These results are support for the hypothesis that the psychological characteristics of people with autism extend beyond diagnostic boundaries and will be found in many of those with the behavioural features of autism.

## Discussion

This study has shown that young people with high levels of autistic behavioural traits share the cognitive characteristics of people with a diagnosis of autism. That is, they are more likely to have poor Theory of Mind but to show superior performance on tests of visual disembedding. Young people with high levels of autistic traits also display lower levels of cognitive flexibility as measured by reversals of the ambiguous figure.

This supports the hypothesis that autism is an extreme on a continuum of traits that are also present in the general population. These traits are behavioural and are underpinned by a cluster of psychological characteristics that are also distributed through the population. This study supports the view that genetic and neuroimaging studies should study phenotypes based on Theory of Mind ability, central coherence and mental flexibility as an alternative to strict definitions of autistic disorder.

The cognitive profile revealed is different to what one would expect were these behaviours related to general intellectual impairment. One would expect good Theory of Mind performance and good visual disembedding to be associated with high IQ. In fact, there was poor Theory of Mind in the presence of good visual disembedding in the profile that predicted behaviours characteristic of autism, which suggests that IQ differences do not explain our findings.

The core measures of the 2nd order false belief test and the block design test measure of central coherence have the strongest evidence of sensitivity to the cognitive

characteristics of autism and these have been shown to make a significant contribution to the prediction of behavioural autistic traits. The other measure of central coherence, the visual illusions, did not contribute to prediction of behavioural traits. The lack of a relationship between succumbing to the illusions and score on the block design test suggests that they may not be measuring a unitary function.

The other more exploratory measure used was the ambiguous figure. This measure contributed significantly to the prediction of the presence of autistic behavioural traits

The ambiguous figure was used here as a measure of mental flexibility. However, ambiguous figures have been used in developmental research to investigate the perception of ambiguous figures in relation to the acquisition of Theory of Mind. Following on from this work in typically developing children, two studies have investigated the perception of ambiguous figures in autism. In the first, Ropar et al. (2003) examined whether children with autism would be able to identify the two alternate interpretations of an ambiguous figure. They found that the children with autism were just as able as controls with mild intellectual impairment to see the alternative interpretations of a figure with prompting.

In the second, Sobel et al. (2005) found that high-functioning children with ASD were less likely to reverse ambiguous figures spontaneously, that is before being informed that they might reverse, than a control group. They were also more likely only to acknowledge a single interpretation of the stimuli.

In contrast to the results of Sobel et al. (2005) our results indicate that people with autistic traits differ only on perception of *informed* reversals. That is, when they know the figure has two interpretations and that it might reverse they are less likely to see reversals of the ambiguous figure. There are slight methodological differences between the studies. Ropar et al. (2003) did not measure informed reversals only perception of both interpretations of the figure. Sobel et al. (2005) used three different ambiguous stimuli and asked participants three times during the course of viewing each stimulus if it had changed. So the number of reversals seen could only range between 1 and 3 for each figure. In this study however, subjects were asked to report all reversals seen and only prompted when subjects did not see any reversals. In addition, Sobel et al. (2005) sample was chronologically younger than ours which may be significant when there is evidence for a developmental progression in the probability of seeing informed reversals. It may be that differences in perception of ambiguous figures between people with autistic traits and without have different expression depending on developmental level of participants.

There is growing evidence that ambiguous figures may be important in the study of perception in autism. Further research is required to determine whether they are measuring mental flexibility as an executive function as suggested here. This interpretation is supported by the relationship between Dimensional Change Card Sort performance and ambiguous figure perception (Bialystok and Shapero 2005). Alternatively, they could be measuring a developing capacity for managing multiple representations of objects as suggested by the Theory of Mind literature. Further research is required to investigate this area.

In summary, these results suggest that ambiguous figure perception, Theory of Mind and central coherence all contribute to a cognitive phenotype underpinning behaviours characteristic of autism and that this cognitive phenotype extends beyond the boundaries of the current diagnostic criteria.

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