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# **Parents of children with Asperger Syndrome: What is the cognitive phenotype?**

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## **Abstract**

Two cognitive anomalies have been found in autism: a superiority on the Embedded Figures Task, and a deficit in 'theory of mind'. Using adult level versions of these tasks, the present study investigated if parents of children with Asperger Syndrome might show a mild variant of these anomalies, as might be predicted from a genetic hypothesis. Significant differences were found on both measures. Parents were significantly faster than controls on the Embedded Figures Task, and slightly but significantly less accurate at interpreting photographs of the eye-region of the face in terms of mental states. The results are discussed in terms of the broader cognitive phenotype of Asperger Syndrome, and in terms of their implications for cognitive neuroscientific theories of the condition.

Autism is diagnosed when a child fails to show normal social and communicative development, in combination with restricted development of imagination, and the presence of unusually repetitive behaviour. Asperger Syndrome (AS) is diagnosed in a similar way, but whereas in autism there is also a period of language delay, in AS language development proceeds on time<sup>1</sup>. In autism there is often (but not always) associated mental handicap (ie: below average IQ), whereas in AS, IQ is usually in the normal range. Since the social, communicative, and imaginative abnormalities are common to both, it is nowadays thought that both disorders lie on the same *continuum*, with AS closer to the normal end (Wing, 1988).

A major breakthrough in understanding autism and AS occurred with the discovery of *genetic* factors in their aetiology. This can be traced back to the landmark study by Folstein and Rutter (1977) which identified that the concordance rate for autism was far higher amongst monozygotic (MZ) twins than dizygotic (DZ) twins. For MZ twins, the autism concordance rate was 36%, whilst for DZ twins, the rate was zero. Later twin studies confirmed this MZ-DZ difference, to an even more marked degree. Thus, Bailey, Le Couteur, Gottesman, Bolton, Simonoff, Yuzda, and Rutter (1995), using a combined sample from the previous study and new families, found the MZ autism concordance was 60%, whilst the DZ autism concordance rate again was zero. The DZ rate is likely to be similar to the sib-risk rate for autism (2-3%), which itself is significantly above

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<sup>1</sup> When establishing if language is delayed or not, only simple lexical and syntactic development are usually considered. ICD-10 defines language delay as not using single words by 2 years old, or phrase speech by 3. Phrase speech is anything more than single words (eg: two-word utterances). Pragmatics is not considered, which in our view is a mistake. If pragmatics was considered, then children with AS would in all likelihood be classified as showing language delay, or atypical language development.

general population rates. All of this evidence is consistent with the idea of genetic factors causing autism (Bolton and Rutter, 1990).

The seeds of the genetic theory go even further back: Leo Kanner (1943) in the first description of autism noted that the parents of children with autism appeared to share some of their children's psychological characteristics. His description was interpreted, regrettably, by some to mean that the parenting style might be causing the child's autism. Hans Asperger (1944), who described the related condition that now bears his name, was more explicit in suggesting that the parent-child similarities reflected genetic factors. Indeed, he suggested the syndrome was inherited from the fathers.

A range of other evidence also suggests that both autism and Asperger Syndrome (AS) is of genetic origin. First, in classic autism, the sex ratio is 4:1 (m:f) (Wing, 1976; Rutter, 1978). In high-functioning autism or AS, the sex ratio is even more biased towards males. Wing (1981) suggests it is 9:1 (m:f). Such sex differences are unlikely to reflect differences in socialization: they are more likely to reflect neurodevelopmental differences between the two sexes, ultimately with a genetic basis. Secondly, families with a child with AS show an increased rate of AS profiles amongst other relatives (Gillberg, 1991). Thirdly, there is an increased rate of other developmental disorders, such as language delay and dyslexia, amongst the siblings of children with autism, relative to population baseline levels (Folstein and Rutter, 1988).

*The cognitive phenotype*

This latter finding suggests that the phenotype for autism or AS may be broader than the diagnostic symptoms for the two conditions. Rutter and others (Bolton et al, 1994; Bailey et al, 1995; Piven et al, 1994) refer to this as the “extended phenotype” or the “broader phenotype”. (Here we shall use these two terms synonymously). Their suggestion is that relatives of people with autism or AS may not have the condition itself, but may nevertheless have a “lesser variant” of it, as a result of carrying or expressing some of the genes for it. However, there is as yet no consensus on the nature of the extended phenotype. In this article, we test for the presence of a cognitive phenotype in parents of children with AS.

Other studies have suggested that the broader phenotype includes both differences in social functioning (Wolff, Narayan, and Moys, 1988; Piven et al, 1994), pragmatics (Landa, Folstein, and Isaacs, 1991; Landa et al, 1992), OCD phenomena (Bolton et al, 1994), and/or a history of early language or reading problems (August, Stewart, and Tsai, 1981; Minton et al, 1982; Baird and August, 1985). As Gillberg (1989) and Piven et al (1994) suggested, first degree relatives seem to show a mild version of the same deficits as their affected children. DeLong and Dwyer (1988) went further in finding a high rate of AS among the parents of children with autism.

In the study reported here, we test the notion that social functioning may be abnormal in first degree relatives of people with AS. We take advantage of previous cognitive studies of autism to predict that the cognitive phenotype will involve a deficit in social cognition,

specifically in ‘mindreading’ or ‘theory of mind’. This is a more focused hypothesis than simply suggesting that there are problems in ‘social functioning’. In addition, and again on the basis of previous cognitive studies, we predict a relative superiority in the identification of ‘embedded figures’. These strong, specific predictions are explained next.

### *Cognition in autism and AS*

I. Mindreading: A range of studies show that children with autism, in the borderline average IQ range, are impaired in the development of a theory of mind, or in what can also be called ‘mindreading’. For shorthand, they are said to suffer from ‘mindblindness’ (Baron-Cohen, 1990, 1995). Mindreading is defined as the ability to think about mental states (one’s own or another person’s), and reason about behaviour in terms of underlying mental states. Mental states include the full range of intentional states (beliefs, desires, intentions, knowledge, pretence, imagination, etc.,). Mindreading is the major way in which the normal child or adult makes sense of or predicts social behaviour (Dennett, 1978).

Many children with autism fail first-order belief tests (inferring someone’s beliefs), tasks which are normally passed by children by 4 years of age (Baron-Cohen, Leslie and Frith, 1985). Some children with autism can pass at this first-order level, yet fail at the next level of complexity, second-order belief tests (Baron-Cohen, 1989). In second-order tests, the subject has to reason that “John thinks that Mary thinks x”. Such reasoning is within

the capability of a normal 5-6 year old child. Almost all children with autism or AS fail such tasks, although some with higher IQ may pass even these (Ozonoff, Pennington, and Rogers, 1991). This reflects ceiling effects due to the tests being at the 5-6 year old level. For example, when such subjects are tested at a 9 year old level, deficits are again revealed (O’Riordan, Baron-Cohen, Jones, Stone, and Plaisted, 1996). In the study reported below, we employ an adult test of mindreading<sup>2</sup>.

ii. Embedded Figures: A second cognitive anomaly in autism and AS is their relative superiority in finding embedded figures. This was first reported by Shah and Frith (1983), using the Children’s Embedded Figures Task (CEFT). They found that such children were more accurate in finding the part (a simple target shape) within the whole (a complex design). Frith (1989) suggests that such a skill may be a sign of “weak central coherence”. By this she means that in the normal brain there is a drive towards Gestalt (whole object) perception, at the expense of detailed information processing in order to establish global meaning. In the brain in autism or AS, Frith’s theory holds that the drive for Gestalt perception or meaning is less strong. This is held to lead to an increased focus on *parts* of objects.

Ozonoff et al (1991) failed to replicate the finding of superior performance on the CEFT, but did find that this aspect of cognition was unimpaired in both subjects with autism and AS. Neither study collected response time (RT) data, but in a recent study Jolliffe and

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<sup>2</sup> Reviews of the evidence for mindblindness in autism can be found in Baron-Cohen, Tager-Flusberg, and Cohen (1993), and Baron-Cohen (1995).



Baron-Cohen (in press) have found that people with autism or AS are actually *quicker* at identifying parts within wholes. This latter study tested high-functioning adults with autism or AS, and used the adult Embedded Figures Test (EFT) (Witkin, Oltman, Raskin, and Karp, 1971). In the study reported below we also used the adult EFT to test the prediction that parents of children with AS are also quicker on the EFT.

## **The Experiment**

### *Subjects*

2 groups of subjects were tested:

(1) The first group comprised 30 parents of children with AS. (Henceforth, this is the AS-Parent Group). All subjects in this group had a child who met established criteria for AS or autism (ICD-10, 1994), as diagnosed by an experienced psychiatrist in the University of Cambridge. They were recruited via the Cambridge AS Association. Their age range was 30-70 years ( $x = 44.36$ ,  $sd = 10.61$ ). They comprised 15 couples (15 mothers and 15 fathers). They were of mixed socioeconomic status (SES). They were all of normal intelligence as assessed by the NART ( $x = 120.3$ ,  $sd = 10.6$ ).

(2) The second group comprised 30 parents of children without AS or autism. They were aged 25-65 years old. (Henceforth, this is referred to as the Control Group). Their mean age was 42.3 years ( $sd = 13.6$ ), which was not significantly different from the mean age of the AS-Parent Group. As with the AS-Parent Group, they were of mixed SES. They

comprised 15 couples (15 mothers and 15 fathers). On the NART, their IQ again was in the normal range ( $x = 122.6$ ,  $sd = 11.4$ ), and did not differ significantly from that of the AS-Parent Group ( $t$  test,  $p > 0.05$ ). Finally, their educational level, as assessed by number of years in education, was comparable to the AS-Parent Group.

## *Method*

Each subject was given one of two cognitive tests, the Reading the Mind in the Eyes Test, and the Embedded Figures Test. These are described next.

### *a) The Reading the Mind in the Eyes Test (Eyes Test)*

This test was first used by Baron-Cohen, Jolliffe, Mortimore, Robertson (1996), with normal adults and adults with autism/AS. It assesses adult level theory of mind abilities. It comprises photographs of the eye region of 25 different faces (male and female). The faces were taken from magazine photos. All faces were standardized to one size (6 x 4 ins), with the same region of the face selected for each photo - from midway along the nose, to just above the eye-brow. See Figure 1 for examples. Each picture is shown for 3 seconds, and a forced choice between two mental state terms is then presented, and the Experimenter says to the subject "Which word best describes what this person is feeling or thinking?". Subjects are given 5 seconds to respond. (Figure 1 also shows the relevant mental state terms for the example photos). Baron-Cohen et al (1996) found that normal female adults were slightly but significantly better than normal male adults, with both sexes performing well above chance on this test. In addition, they found that adults with autism/AS were significantly worse on this test, relative to controls. The maximum score on this test is 25.

insert Figure 1 here

b) *The Embedded Figures Test (EFT)*

This is a published test (Witkin et al, 1971). Just Set A was used. In this test, the subject is first given a practice trial, in which it is explained that the subject must find the simple target shape within the complex design. The complex design is presented for 15 seconds and the subject is then invited to describe it, to ensure they are attending to it. This is then turned over, to show the simple target shape for 10 seconds. The card is then turned back, thus re-presenting the complex design, and the subject is given a maximum of 3 minutes (180 secs) to locate the simple shape within the complex design. (The subject can turn back to look at the simple shape as often as they like). The task thus entails spatial analysis of a visual design into constituent segments. The subject is instructed to go as quickly as possible, and performance is timed. There are 12 items in the complete test. Witkin et al (1971) and other studies consistently find a male superiority on the EFT, in terms of response time.

## **Results**

Results are displayed in Table 1. All group differences were tested using independent t tests, 1 tailed to test for predicted differences, with a significance level set at  $p < 0.01$  to

correct for multiple comparisons. On the Eyes Test, control males were significantly worse than control females ( $p < 0.0027$ ), replicating the earlier finding by Baron-Cohen et al (1996). AS-Fathers were significantly worse than AS-Mothers ( $p < 0.014$ ). Critically, when comparing the AS parents with their sex matched controls, AS-Fathers were significantly worse than control males ( $p < 0.004$ ), and AS-Mothers were significantly worse than control females ( $p < 0.0001$ ). On the EFT, the opposite sex difference was found. Control males were significantly faster than control females ( $p < 0.01$ ), replicating previous studies (Witkin et al, 1971). AS-Fathers were also faster than AS-mothers ( $p < 0.039$ ), and the AS-Parents were significantly faster than their sex matched controls (AS-Fathers x Control Males,  $p < 0.01$ ; AS-Mothers x Control Females,  $p < 0.005$ ).

**Table 1: Mean scores (with standard deviations in brackets) on the Embedded Figures Task (EFT) and the Eyes Test.**

<b>GROUP</b>	<b>n</b>	<b>EFT x (sd)</b>	<b>EYES x (sd)</b>
<b>CONTROLS</b>	30		
<b>male</b>	15	46.2 (20.5)	19.5 (2.6)
<b>female</b>	15	66.7 (36.7)	22.1 (2.0)
<b>PARENTS</b>	30		
<b>male</b>	15	32.8 (17.7)	17.3 (1.6)
<b>female</b>	15	48.6 (31.8)	18.9 (2.1)

## Discussion

The present study finds that the cognitive phenotype of AS, as predicted, involves two aspects. First, mothers and fathers of children with AS are faster on the Embedded Figures Test (EFT), relative to sex-matched controls. This superior performance mirrors the ‘islet of ability’ found in autism (Shah and Frith, 1983; Jolliffe and Baron-Cohen, in press). Secondly, mothers and fathers are slightly impaired on a mindreading test, the Reading the Mind in the Eyes Test. Again, this deficit is relative to sex-matched controls. This subtle deficit mirrors the ‘mindblindness’ found in autism (Baron-Cohen et al, 1996; Baron-Cohen, Campbell, Karmiloff-Smith, et al, 1995; Baron-Cohen, Leslie, and Frith, 1985; Baron-Cohen, 1995).

Therefore, the first conclusion that can be drawn from this study is that, as suggested by others (Piven et al, 1994), first-degree relatives of AS show a milder (lesser) variant of the *cognitive* profile of autism. As one father of a child with autism put it to us, when he was describing his child, “His mind is just like mine, but writ large”.<sup>3</sup> The mindreading deficit found in the parent group in this study is likely to be broader than a deficit in reading mental states in the eyes (tested here); rather, it is likely to be due to the same factor that causes the lack of tact found in some studies (Piven et al, 1994), and the pragmatics deficits found in others (Landa et al, 1992), since tact and pragmatics are just

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<sup>3</sup> The example this father gave was that he (the father) was interested in meteorology, and therefore tried to catch the weather forecast on television after the evening news. His son had taken this interest to obsessional lengths. He watched every weather forecast on television, and listened to them on the radio, all

two examples of mindreading ability at work.

The second conclusion to be drawn from this study is that first-degree relatives of AS show a slightly magnified form of the ‘male’ profile on these tests. Normal males tend to be slightly but significantly faster on EFT, and slightly but significantly less accurate on the Eyes Test, relative to normal females. Elsewhere we have argued that this may reflect autism and AS being extreme forms of the ‘male brain’ (Baron-Cohen and Hammer, in press). The ‘male brain’ can be thought of as a particular cognitive profile, more often found in males, but not restricted to them. It is defined as showing a significant discrepancy between social (SOC) and spatial (SP) skills (a  $SOC < SP$  discrepancy). In the present study, the SOC skill measured was a mindreading test, and the SP skill assessed was EFT. Relevant to this is the fact that most people with autism and AS are male (Wing, 1976; Rutter, 1978).

#### *Implications for cognitive neuroscientific theories*

The results from this study reveal an inverse relationship between EFT and mindreading skills. Why should this relationship exist? Frith (1989) suggests it is because mindreading requires strong central coherence, whilst EFT requires weak central coherence. In a later paper, Frith and Happe (1994) argue against this position, suggesting that because one can find subjects with autism who pass second-order theory of mind tests but who still exhibit weak central coherence, the two domains must be independent of one another.

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day and all night. Apparently, such forecasts are broadcast every 15 minutes throughout the day, and

This rejection of Frith's (1989) position may however be premature. In the Frith and Happe study, the dissociation between mindreading and weak central coherence may have occurred because tasks were not matched for complexity. The present study suggests that when an adult mindreading test is employed, weak central coherence (as expressed by superiority on EFT) may go hand-in-hand with impaired mindreading. It is therefore still possible that there is an important (inverse) relationship between these two domains.

Do the present results challenge earlier views about the modularity of mindreading (Leslie, 1991; Baron-Cohen, 1994, 1995)? A relationship between mindreading and EFT could occur without implying that mindreading is not modular. These two systems could simply be independently, coincidentally affected in people with autism or their relatives. The present results cannot therefore address the modularity debate.

In terms of genetic theories of autism and AS however, the finding of superior performance in the parents of children with AS might account for why genes for autism should persist in the gene pool. From the present study one can speculate that in its mild form (as expressed in the parents), the genes responsible for the social (mindreading) deficit also provide a cognitive advantage - in this case, in the rapid analysis of information into its constituent parts. Clarifying the adaptive value of such a cognitive phenotype will be important for future research.

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continue during the night on some channels. His son therefore hardly ever slept.



In terms of the brain basis of such a cognitive phenotype, there are only clues at present. First, according to recent neuroimaging studies (using both SPECT and PET), mindreading may involve prefrontal cortical areas, such as orbito-frontal cortex (Baron-Cohen, Ring, Moriarty, et al, 1994) and/or medial frontal cortex (Fletcher, Happe, Frith, Baker, Dolan, Frackowiak, & Frith, C, (1995); Goel, Grafman, Sadato, and Hallett, 1995). Secondly, Baron-Cohen and Ring (1994) and Brothers (1990) hypothesize that decoding mental states from the eye-region of the face may involve the superior temporal sulcus, given findings from single cell recording studies with non-human primates (Perrett et al, 1985) and given evidence from neuropsychological studies of human patients with prosopagnosia who have deficits in gaze-detection (Campell et al, 1990). Third, they speculate that the amygdala may also be part of a neural circuit for mindreading, given its role in emotion perception (Damasio, 1995) and in gaze perception (Brothers, 1990). Mindreading may therefore involve a number of different brain regions. More work needs to be done to identify this in normal individuals, as well as in understanding the deficit in mindreading in autism and AS.

Regarding the brain basis of superior performance on the Embedded Figures Test, there is currently only a clue to guide us. Lamb and Robertson's work (discussed in Rafal and Robertson, 1996) shows that patients with right sided lesions in the temporal-parietal junction are more likely to see local features in the Navon task (ie: when presented with a large letter H made up of little letter S's, they will say they saw an S). In contrast, patients with a left sided lesion in the temporal-parietal junction are more likely to report the opposite - to see the global level and not the local. (In the example used, they will

report seeing an H). This suggests the right temporal-parietal junction is involved in global processing, and that patients with autism (or their relatives) may have abnormalities in this domain. It will be important for future work to relate the findings from the cognitive level more closely to the neural level.

### *Examples of EFT in real life, in autism and AS*

The EFT is a laboratory task. Are there any examples from real life to which this might correspond? Our first example of superior EFT-style processing is from an adult with autism (and of normal intelligence) whose childhood obsessions included shredding a tiny piece of paper into hundreds of even tinier pieces. The pieces were so small that noone else noticed them, but for her she reported they were very important. If one piece was missing, she would become anxious and upset. When she was alone in her room, she fiddled with the paint on her bedroom wall for hours, inspecting the tiny pieces of dust in the cracks in the paint. When she went to the pub, she watched the shapes in the fruit machine for hours, monitoring their position as customers played new games, describing herself as “sucked into the detail of how the machine worked”<sup>4</sup>.

Consider another set of examples: We know of several families where the father and son share strong interests in train-timetables and train routes. Such cases comprise families where the content of the close-focus interest coincide in the parent and child. We also

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<sup>4</sup> A second example was given earlier, in discussing the father and son who were both fascinated by meteorological reports. (See Footnote 4). In each case, it is the details of the activity that become preoccupying. This mirrors the EFT.

know of examples where the interests of the parent and son do not coincide, but both still reflect the 'EFT style'. Thus, we met a father who is a carpenter (with all of the precision and eye-for-detail that this presumes, and who also describes himself as "a loner") whose child with AS spends all of his time drawing rows of houses with their street numbers marked on. As one might expect, such drawings show the houses on both sides of the street, with odd numbers on one side and even numbers on the other, and absorb the child in an almost endless activity (putting in the numbers on the houses for every street he draws). Our final example is of a father who is a taxonomist, making ever more fine-grained categorization (of plants), whilst his son with AS has similarly narrow interests (in African frog species). These interests are examples of becoming locked into detail, seeing "each tree as well as the wood". In contrast, the normal brain, according to Gestalt psychologists, focus on the wood more than the trees.

The present results, and these anecdotes, suggest that autism and AS may constitute a particular cognitive style, rather than an impairment, as Frith and Happe (1994) discuss. The style seems to involve deficits in mindreading in the presence of superior processing of local information. Whilst some progress has been made in characterizing the cognitive phenotype of people with autism and AS, and their parents, there is a need for neuroimaging studies to clarify the brain basis of this cognitive phenotype.

## Figure Legends

Figure 1: Examples from the Eyes Test (reproduced from Baron-Cohen et al, 1996, with permission)<sup>5</sup>.

Fig 1a: Sad vs Happy

Fig 1b: Reflective vs Unreflective

Fig 1c: Dominant vs Submissive

Fig 1d: Sympathetic vs Unsympathetic

(Correct word is the first of each pair. In the test, correct word is randomized with respect to position).

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<sup>5</sup> These pictures were originally parts of larger photographs from magazines. Unfortunately their sources are no longer recognizable to us.

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