The Boundaries of the Cognitive Phenotype of Autism: Social Cognition and Central Coherence in Young People with Autistic Traits and their First Degree Relatives.

Catherine Best

PhD

The University of Edinburgh

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Part 2

Abstract

Autism is a behaviourally defined disorder. The impairments in social communication and repetitive behaviours are individually non-specific. The disorder has indistinct boundaries both with other psychiatric disorders and with normal personality types.
At the cognitive level, groups of people with autistic disorder can be differentiated from people without the disorder by their ability to reason about beliefs and knowledge (Theory of Mind) and by tests of visual disembedding (central coherence).
This study examined whether young people with some of the behavioural features of autism but not necessarily a diagnosis, would show this distinctive cognitive profile. In a sample of 60 young people with additional learning support needs, we found that those with high levels of autistic traits (n=40) showed the same cognitive profile as has been found in people diagnosed with autistic disorder. This supports the view that autism is an extreme on a continuum of cognitive traits.
Given the highly heritable nature of autism, we hypothesised that the parents of the young people with autistic traits will also display these cognitive features. The results indicated that there was no difference between the groups of parents on an advanced test of social cognition. Parents of people with high autistic traits were more resistant to one of the visual illusions and saw fewer reversals of an ambiguous figure when IQ was statistically controlled. These results in a sample with a low genetic load suggest ambiguous figures will be important in delineating the broader cognitive phenotype of autism.

Part 1

Introduction

How autism is currently defined?

The diagnosis of the syndrome known as autism was introduced in the mid twentieth century on a descriptive basis. The syndrome continues to be defined based on a set of behaviours which when present together are indicative of a relatively predictable course. At the time of the introduction, there was no biological marker and this continues to be the situation. Operational definitions were introduced into psychiatric classifications from the 1970s because of the demonstrable lack of reliability of purely descriptive definitions. The most widely used definitions are those of the American Psychiatric Association's Diagnostic and Statistical Manual (DSM-IV; American Psychiatric Association, 1994) and the World Health Organisation's International Classification of Diseases (ICD-10; World Health Organisation, 1992).

Volkmar and Pauls (2003) summarise the features and sample behaviours required for a diagnosis of autism according to the DSM-IV and ICD-10:

1. Qualitative impairment in social interaction
    a. Impaired eye gaze
    b. Lack of social reciprocity
    c. Poor or absent joint attention
    d. Limited or absent peer relationships

2. Qualitative impairment in communication
    a. No language (no alternative compensatory means used to communicate)
    b. If language spoken, involves echolalia, difficulties in pragmatic language

c. Lack of appropriate imaginative play

3. Restricted pattern of behaviour, interests
    a. Abnormal preoccupations, interests and activities
    b. Difficulties with change
    c. Stereotyped mannerisms

4. Onset before 3 years old

There is an algorithm for making a diagnosis of autism, which sets out how many features need to be present in each domain.
There are also standardised tools to support clinicians and researchers in making relatively objective and reliable judgments about diagnosis. Widely used examples are the Autism Diagnostic Observation Schedule (ADOS-G; Lord, Risi et al. 2000) and the Autism Diagnostic Interview Revised (ADI-R; Lord, Rutter et al. 1994).

In the Diagnostic and Statistical Manual of the American Psychiatric Association, (DSM-IV, APA 1994) autistic disorder is included under the umbrella term Pervasive Developmental Disorders (PDDs). PDDs are 'severe and pervasive impairments in several areas of development: reciprocal social interaction skills, communication skills, or the presence of stereotyped behaviour, interests and activities' (DSM-IV, APA 1994). In addition to autistic disorder, the PDDs include:

- Asperger disorder,
- childhood disintegrative disorder,
- Rett syndrome, and
- Pervasive developmental disorder not otherwise specified (PDD-NOS).

PDD-NOS is used where there are not enough features of autism to meet the full diagnostic criteria.

There have been many revisions of classification since autistic disorder was first identified. We will now set out how the current criteria have developed since their inception.

Historical introduction

Psychiatrist Leo Kanner was the first to describe autistic disorder, in 1943 (Kanner 1943). He termed the disorder as 'early infantile autism'. A year later, paediatrician Hans Asperger published a similar series of cases (Asperger 1944). He termed his cases 'autistic psychopathy'. In using the term 'psychopathy' Asperger sought to indicate a long-standing personality trait and was not suggesting an association with sociopathy. The term 'autism', chosen by both men, was not a new term. Bleuler (1911) had used 'autism' to describe the self-absorption characteristic of schizophrenia. Kanner and Asperger did not use 'autism' in this sense. Autism as originally described by Kanner is a fundamental aloneness due to an innate inability to relate to other human beings. Kanner described 'the outstanding, "pathognomic", fundamental disorder [in autism as] the children's inability to relate themselves in the ordinary way to people and situations from the beginning of life'. Prior to Kanner's description, such children would have been regarded as intellectually disabled or as suffering from a childhood onset psychosis.

Both Kanner and Asperger were born in Austria: however, at the time they were writing about autism, Kanner was working in America, while Asperger was working in Austria. Asperger's article was published in German during the Second World War and was largely disregarded in the English speaking world until the 1980s. In 1981, Lorna Wing published an article examining his original cases and other similar case histories. Wing's article opened up an area of fruitful debate about the relationship between these two disorders.

In ‘Autism and Asperger Syndrome’ edited by Uta Frith, she describes the common features of the two descriptions as poverty of social interaction and failure of communication, stereotyped behaviour, isolated special interests, outstanding skills, resistance to change, clear separation from childhood schizophrenia, attractive appearance, and the similarities between parents’ and children’s behaviour (Frith 1991). The differences present between the first papers were that Kanner focused on the deficits in the children’s language abilities and described echolalia, pronoun reversal and difficulties generalising word meanings, while Asperger described his children as having preserved, even sophisticated, language but with the disturbed pragmatics characterised by lack of humour and pedantry. Kanner believed that only these children’s relation to people was disturbed and their relation to objects was superior, whereas Asperger described disturbed relation to people and objects.

Autism and Asperger’s syndrome.

There is ongoing debate about whether the two groups of children described by Kanner and Asperger are part of a single diagnostic entity and whether a meaningful distinction can be made. Van Krevelen (1971) thought that Kanner’s autism and Asperger disorder were two distinct disorders. He described Asperger disorder as an unchanging personality trait and autism as progressing with development.

In the DSM-IV (APA 1994), the most widely used diagnostic system in the UK, Asperger disorder and autistic disorder are both diagnosed based on qualitative impairments in social interaction and a restricted pattern of behaviour and interests. The differences are that in Asperger disorder there is no significant general delay in language acquisition or impairment of general cognitive abilities. In autistic disorder, the onset must be before the age of three years. These criteria are still contested and some practitioners argue that normal early language development cannot precede the communicative abnormalities described for Asperger disorder in DSM IV so the

diagnosis is logically impossible (Mayes, Calhoun et al. 2001). Other alternative criteria for Asperger disorder have been suggested e.g. Ehlers and Gillberg (1993) and Gillberg and Gillberg (1989) . For example, Asperger disorder has been differentially linked to general clumsiness and poor co-ordination (Gillberg 1989) but this is not currently part of the diagnostic criteria.

If the two diagnostic groups, autistic disorder and Asperger disorder as defined by DSM IV were to show a similar prognosis then this is evidence against their dissociation. Under the current definitions, there is some evidence that outcomes are similar for the two disorders. For example, Howlin (2003) examined the outcomes at 18 years of a group of adults with high functioning autism compared to a group with Asperger disorder. Howlin found that their social outcome measures or current Autism Diagnostic Interview (ADI) scores could not differentiate the groups. At follow-up, both had similar levels of language problems. This is despite the groups being differentially diagnosed based on the autism group's early language delay.

The behaviours resulting from autism are not static: behavioural symptoms change with development. For example in a small child, a deficit in imagination may be expressed as a lack of social imitative behaviour such as pretending to vacuum. In adolescence, the same psychological deficit is expressed as an interest in collecting information such as memorizing bus timetables for a whole city. This may be to the extent that a child with classical Kanner's autism develops into a teenager with Asperger disorder (Szatmari, Bryson et al. (2000) for overlap between developmental trajectories). This concurs with the school of thought that sees autistic disorder and Asperger disorder as part of a single spectrum of disorders that are grades or variations of the same condition. This belief is supported by the fact that there is co-occurrence of the disorders within families, e.g. Gillberg and Cederlund (2005), suggesting a common genetic root.

Some of the early proponents of the idea of a single group are Wolff and Barlow (1979) who described a range of function between autistic, schizoid and normal children.

Wing and Gould (1979) defined the core triad of impairments in autism as deficits in socialisation, communication and imagination. In the Camberwell study (Wing and Gould 1979), a large-scale study of children with mental retardation, they found that impairments in these three domains cluster together. All the children found to have deficits in social interaction had deficits in the other two areas, proving that they are not a random association of traits. The possible range of deficits across the three areas of impairment is the source of variability in presentation of the disorder. This supports the concept of autism as a spectrum of conditions that range in their severity and may well form a continuum with the rest of the population.

The concept of a continuum has received support from a recent study, which examined the extent to which these three symptom domains (social, communicative and imaginative: taking repetitive behaviour to be equivalent to impaired imagination) are phenotypically independent. Constantino, Gruber et al. (2004) cluster analysed the data (ADI-r and SRS[1]) from 226 children with and without PDD. They found evidence of 'a single, continuously distributed' underlying factor. This is evidence that people with autism and the rest of the population are on a continuum of severity of autistic traits and to some extent, this can be measured uni-dimensionally.

Reflecting the possibly indistinct boundaries between the conditions, the term Autistic spectrum disorder (ASD) refers to autistic disorder, Asperger disorder and PDD-NOS. This usually excludes Rett disorder, due to its distinct etiology, and childhood disintegrative disorder, due to its distinct developmental course.

[1] Social Responsiveness Scale, formerly known as the Social Reciprocity Scale (Constantino, J. N. (2002). The Social Responsiveness Scale. Los Angeles, Western Psychological Services.). A 65 item questionnaire that yields a single continuously distributed factor, used to measure grades of social behaviour in typically developing samples.

Conclusions

Currently there is no biological marker for ASD; the disorder is diagnosed on behavioural features. There are standard criteria for the diagnosis of autism and Asperger disorder. Strict diagnostic criteria that are used consistently are essential for answering questions about epidemiology and etiology. These criteria have been developed based on clinical data. However, it is still a matter of debate whether autism, Asperger disorder and PDD-NOS are different manifestations of the same disorder or whether they are distinct conditions. If it could be shown that there is a common underlying substrate for all the ASDs, then this would support the concept of the autistic spectrum.

We will now examine the available evidence on causes of Autistic Spectrum Disorders.

What is the cause?

Kanner initially recognised the innate nature of autistic disorder. He later rejected this hypothesis, but subsequently vacillated between viewing it as a psychogenic disorder and a hereditary condition. The view of autism as a psychogenic disorder persisted for the next three decades: 'refrigerator mothers' (Bettelheim 1967), who were seen as cold, intellectualising and distant from their children, were blamed for their child's disorder. During the 1970s, new techniques available for investigating the causes of psychopathology led to the re-evaluation of this conclusion. 30% of children with ASD develop seizure disorders (Rutter 1970; Gillberg 1984; Olsson, Steffenberg et al. 1988). It is usually either in early infancy or adolescence that these seizure disorders become evident in autism.
Facts such as this high rate of seizure disorder and the association of autism with intellectual disability (rates thought to be about 75% at that time) suggested neurological involvement.

Folstein and Rutter published the first twin study of autism in 1977. Twin studies separate the effects of shared environment from genetic effects. Both monozygotic and dizygotic twins can be expected to share many of the same environmental influences (diet, exposure to toxins, parenting practices) but monozygotic twins have identical genetic material, whereas dizygotic twins share the same amount of genetic material as typical siblings. It follows that if monozygotic twins show higher concordance for a disorder than dizygotic twins then that disorder is heritable. If concordance between monozygotic twins is less than 100%, then that indicates environmental influences also play a part. In Folstein and Rutter's study there was found to be higher concordance for the disorder in monozygotic than dizygotic twins. This indicates a heritable disorder. Estimates of the heritability of autism range up to 90% (Bailey, Le Couteur et al. 1995).

Autism is established as a neurodevelopmental disorder. It is believed to be disorder of the central nervous system due to its association with epilepsy and intellectual disability. It is characterised as a developmental disorder due to its insidious onset and the changing nature of symptoms through time. The precise nature of the pathology is not known. It is possible that the damage to the CNS is part of a more generalised disorder, e.g. impairment of immunological responses which would affect all bodily systems. The evidence for these assertions, local CNS pathology versus a more general disorder, is presented below:

1. Central Nervous System

a. Post-mortem Studies

Methods of directly examining the structure and function of the brain in life are very much restricted. Post-mortem studies were for a long time the only method of studying the brain in psychiatric disorders. Post-mortem studies of autism are difficult as premature death is not common in this disorder, there are high levels of comorbidity with other disorders, also there is limited control over post mortem interval and the presence of other confounds. Nevertheless, one of the most reliable findings in the biological study of autism is that children with autism have greater post-mortem brain weights than average (Bauman and Kemper 1985). It has been suggested that this may be a feature of the disorder in childhood and brain size may not be significantly increased by the time the children reach adulthood (Kemper and Bauman 1998).

In a living person, brain size may be indirectly assessed by measuring head circumference. This allows larger samples to be ascertained. A limitation is that head circumference only reflects brain size in childhood and not adulthood. Courchesne and colleagues (2003) compared longitudinal records of head circumference in a group of infants who were later diagnosed with ASD, to normative data. They found that the children with ASD had smaller head circumferences than average just after birth. On

average, the children with ASD then had an excessive increase in head circumference between birth and 6-14 months. This accelerated rate of growth was greater for autistic disorder than PDD-NOS. Only 6% of the healthy controls showed the same pattern. These findings were based on a sample of only 48 cases, for which full data were available for a minority, so this finding requires confirmation.

Post-mortem studies have found that there are no malformations of brain structures obvious to the naked eye in the brains of people with autism. This has lead researchers to examine the brain for changes at the cellular level. Cellular structure can also only be examined in post-mortem studies. Visible differences have been found between the cellular structure of both the cerebellum and the amygdala of subjects with autism compared to control subjects. Bailey and colleagues (1998) found reduced Purkinje cell numbers in the cerebella of their adult post-mortem subjects. Bauman and Kemper (2003) found that small and tightly packed neurons in the medially placed nuclei of the amygdale are a common feature of the brains of people with autism.

Again, at the cellular level, but this time in widespread areas of the brain there have been reported changes in minicolumnar organisation. Minicolumns are functional 'modules' of 80-100 interconnected afferent and efferent neurones; they are the smallest unit of vertical functional organisation in the cortex. Casanova et al. (2002) report smaller, more numerous minicolumns but without any concomitant increase in cell density. The main structural changes are in the neuropil region of the minicolumns that contain the inhibitory GABAnergic projections. Casanova and colleagues have speculated that the reduction in these inhibitory influences may be responsible for the increased incidence of epilepsy in autism. These findings still require verification by other research groups.

b. Structural Imaging

Since the 1970s new technologies for examining the live human brain have been developed. Structural Magnetic Resonance Imaging (MRI) is a method of examining the brain that can give information about overall brain size, ventricle size, relative size of particular brain structures and the distribution of white and grey matter. It does not give information about cellular structure. This method has been used to examine the brains of people with autism. Structural MRI has supported the post-mortem findings of increased brain volumes in childhood (Aylward, Minshew et al. 2002).

There are currently conflicting results about whether brain enlargement is due to an increase in white or grey matter. For example, Waiter, Williams et al. (2004) found increased volumes of grey matter, Herbert, Ziegler et al. (2003) increased volumes of white matter in autism. It is probable that the age group studied is critical in determining these results. In her review of this area, Lainhart (2006) suggests that the developmental trajectories of grey and white matter growth may be altered in autism. She outlines the evidence for developmental progression as follows. Both white and grey matter are increased in early childhood (Courchesne, Karns et al. 2001). White matter contributes most to the increase in brain size at this time (Courchesne, Karns et al. 2001). As brain-size normalises in later childhood, there remains an increased level of white matter but decreased grey (Herbert, Ziegler et al. 2003; Herbert, Ziegler et al. 2004). In adulthood, there is increased grey matter and decreased white matter ((Waiter, Williams et al. 2004). These volumetric changes in grey and white matter parcellation are not uniform across the brain but are regionally differentiated. The early increase in white matter is mainly in the frontal cortices (Carper, Moses et al. 2002). Even when there are normal overall levels of white or grey matter there can be regional deficits. This developmental trajectory, proposed by Lainhart (2006), is largely based on single studies in each of the age groups. Within age groups, there are contradictory results from different research groups. Nevertheless, there is evidence that the abnormalities are

relative to developmental level. Large longitudinal studies are needed to clarify this area.

Structural MRI has been used to measure the size of specific brain structures in autism. Courchesne and colleagues (1988) have reported reduced size of cerebellar vermal lobules VI and VII in autism. The study has been criticised for failing to control for IQ or overall brain size in subjects. Other research groups have not replicated these findings regarding the cerebellum. Increased amygdala volumes have been reported (Howard, Cowell et al. 2000) but there have also been contradictory reports (Haznedar, Buchsbaum et al. 2000).
An anatomical finding that has largely been unchallenged is that of diminished callosal subregions (Egaas, Courchesne et al. 1995). Whether this decrease in size is due to less myelination or to a reduction in neural projection, is not known.

The caveats mentioned above on the study of the cerebellum apply to much of the imaging research that has been done in autism to date. These studies of the CNS in autism generally have small sample sizes, for example, Haznedar et al. had 17 cases and 17 controls in their study. Many also lack appropriate control groups, for example, Egaas, Courchesne and Saitoh (1995) used healthy controls matched on age and sex. They did not use a developmentally delayed control group. Control groups need to be careful matched: conflating variables include age, sex, IQ, socioeconomic group, medication status and handedness, presence versus absence of psychopathology and presence of co morbid conditions.

Another approach to studying brain function in autism is through indirect measures of neurotransmission. The main neurotransmitters involved in higher functions are noradrenalin, serotonin, dopamine and GABA. All these neurotransmitters have been investigated in most psychiatric disorders and autism is not an exception. Abnormalities in serotonin metabolism are some of the most consistent findings in autism. For example, whole blood serotonin levels are raised peripherally in autism (Schain and

Freedman 1961). Positron Emission Tomography studies have found evidence of abnormal whole brain serotonin synthesis capacity in children with autism (Chandana, Behen et al. 2005). Serotonin not only functions in the adult brain as a neurotransmitter but also has a developmental role regulating neuronal maturation and migration. Gillberg and Coleman (2000) note that there have been findings of both abnormally raised and abnormally low serotonin levels in a wide range of other disorders, for example Huntington disease and motor neurone disease.

Research on brain structure and function in autism is still at an early stage. Studies with larger samples, well-matched controls and longitudinal design are required in order to develop these findings further.

2. General disorders

a. Immune function

There is evidence of disturbed immune function in autism. One aspect of this is disturbed autoimmunity. For example, higher levels of CNS auto antibodies are found in the sera of individuals with autism, compared to controls. There is evidence of increased levels of autoimmune disease (for example, type 1 diabetes, adult rheumatoid arthritis and hypothyroidism) in the families of people with autism compared to families of children with autoimmune disorders and children without a diagnosis (Comi, Zimmerman et al. 1999; Sweeten, Bowyer et al. 2003). Sweeten et al. (2003) found that levels of autoimmune disease were higher for probands with autistic disorder and Asperger disorder families, but not PDD-NOS.

Other evidence of immune dysfunction points to a disturbed cytokine response to infection. There is evidence of these disturbed immune functions in autism both centrally and peripherally. Higher levels of nitric oxide related to levels of a cytokine (interferon-gamma) involved in its production, have been detected in the plasma of

children with autism (Sweeten, Posey et al. 2004). Nitric oxide is involved in regulating the immune system. Centrally, activation of microglia and astroglia have been detected (Vargas, Nascimbene et al. 2005). Sweeten et al. (2003) also found raised neopterin levels and high monocyte counts in individuals with autism.

A possible trigger for an abnormal immune response has been suggested to be the measles mumps and rubella vaccine. The recent controversy about a link between autism and the MMR vaccine began with an early report published in the Lancet in 1998 by Wakefield (Wakefield, Murch et al. 1998). He made a link between autism and gastro immune dysregulation on the basis of the incidence of gastric complaints and 'macroscopically observed lymphonodule hyperplasia' and 'macroscopically determined enterocolitis' in children with autism. The study concerned 12 children who had a regressive disorder and who also suffered from chronic enterocolitis. In 8 of these children, the parents or GPs linked the onset of symptoms to the MMR vaccine. The resulting attention to this finding has caused anxiety among parents and a decline in the uptake for MMR vaccination. No supporting evidence has been found for a link between the MMR vaccine and the increasing prevalence of autism at the population level. The evidence against Wakefield's conclusions has been drawn from much larger scale studies examining the incidence of autism relative to the vaccination coverage over time (Kaye, del Mar Melero-Montes et al. 2001) or relationships between age of symptom onset and vaccination in cases (Taylor, Miller et al. 1999). The Institute of Medicine and the Medical Research Council have both reviewed the existing evidence and concluded that there is no support for a link between the MMR vaccine and autism. It may not be possible to detect such environmental factors at the population level. In a disorder with possibly multiple etiologies, these factors may only affect a small subgroup of the ASD population because of their specific kind of genetic predisposition. There is ongoing research into bowel disorders in regressive autism. However, these studies need to include appropriate control groups of children with developmental disorders distinct from autism, so that the case for a specific link to autism can be tested.

There is still research going on into a connection between immune responses to the measles virus and autism: Singh and Jensen (2003) conducted a serologic study of the measles, mumps and rubella viruses in 88 cases of autism with 32 controls and 14 siblings. They found that levels of viral antibody to the measles virus were raised in autism and concluded that there is a hyperimmune response to the measles virus in autism. It is important to note (as by Courchesne et al. 2003) that the neuroanatomical abnormalities discussed above in the first section would have begun prenatally and that the evidence cited for accelerated brain growth is in the first year of life thus preceding exposure to the measles virus by some time. If there is a link between measles virus or immune response and autism, it is against a background of preexisting neurodevelopmental abnormalities.

b. Metabolic disorders

Poling, Frye et al. (2006) suggest that deficits of oxidative phosphorylation may be a putative cause of some cases of autism.
There have been other suggestions of metabolic disorders underlying autism but none have a compelling level of evidence as yet.

Conclusions

It can be seen from this discussion that although there are some promising areas of research there is no firm evidence for a specific pathophysiological mechanism in autism.

At a more fundamental level than the pathophysiological processes underlying autism, is the question of the balance between environmental and genetic influences on the disorder. Elaboration of the nature of either genetic or environmental influences could give clues as to the pathology of autism.

1. Environmental influences

a. Toxins

It has been suggested that the carrier base for vaccine, thimerosal, which contains mercury, could be a possible cause for autism. A population study in Denmark comparing those who had received a thimerosal-containing vaccine with those who had not did not show evidence of a relationship between thimerosal and autism (Hviid, Stellfield et al. 2003). Producers of the vaccines have already reduced the levels of thimerosal in response to concerns so all studies are retrospective.
Prenatal exposure to valproic acid and thalidomide had also been shown to cause autism in a small number of cases (Christianson, Chesler et al. 1994; Stromland, Nordin et al. 1994).

b. Maternal prenatal infections

Dassa Takei et al (1995) studied the relationship between maternal influenza infection during pregnancy and autism. This large-scale investigation revealed no connection between maternal infection and a subsequent diagnosis of autism.
There have been cases noted of children with congenital rubella and autism (Fombonne, du Mazaubrun et al. 1997; Barton and Volkmar 1998). The last rubella epidemic in 1964 was studied by Chess, Korn et al. (1971). They found that rubella infection produced temporary autistic symptoms and many of their cases had recovered by age 7.
Yamashita, Fujimoto et al. (2003)found that out of 7 children found to have symptomatic congenital cytomegalovirus infection between 1988 and 1995, 2 went on to develop typical autistic disorder. In the 1970s, a similar connection between latent viral infection and schizophrenia was advanced (e.g. Fisman 1975).

c. Post natal infections

Other infectious diseases that have been associated with autism are herpes simplex encephalitis and cytomegalovirus. Peterson and Torrey (1976) performed a study of 78 subjects with autism matched to the same number of controls. They found that 25% of the subjects with autism were positive for the HSV type one antibodies compared to 13% of controls.

There have been descriptions of previously normal adult individuals developing 'autism' after herpes encephalitis (DeLong, Beau et al. 1981; Gillberg 1986; Gillberg 1991).

The negative findings of large-scale studies such as that by Dassa et al. (1995) make it unlikely that maternal infections or even postnatal infections are a probable large-scale cause of autism in the population. However, the link between autism and immunity has not yet been fully clarified.

d. Birth complications

There is an increased incidence of birth complications in autism but this is interpreted as a consequence rather than a cause of the autistic disorder. Bolton, MacDonald et al. (1994) found that in verbal autistic children the severity of autistic symptoms and incidence of birth complications were proportional to the family genetic loading for autism. Glasson, Bower et al. (2004) found that the pattern of perinatal insults observed in probands with autism was also shared with their unaffected siblings suggesting that a common genetic liability to birth complications. Both these findings suggest that the genetic liability to autism includes a predisposition to birth complications.

Conclusions

Although certain environmental factors can increase the risk of autism (e.g. valproic acid) there are no environmental factors that will produce autism in every case of exposure. Many circumstances that produce brain damage will produce a subset of people with autism. The interaction with genetic predisposition, nature and timing of the brain damage and other variables will influence which individuals develop autism. The precise mechanisms by which this occurs have still to be established.

2. Genetic influences

Twin studies have shown that monozygotic twins with the same genotype are more likely to be dually affected with autism than dizygotic twins who do not have exactly the same genetic material. When the broader autistic phenotype is included then the rates of concordance are even higher (Bailey, Le Couteur et al. 1995). The concordance for the broader phenotype in monozygotic twins is 70-90% and in dizygotic twins 0-10% (Folstein and Rosen-Sheidley 2001).

The siblings of probands with autism have an increased risk of the disorder (2-8%) compared to the rest of the population (~0.6%) (Chakrabarti and Fombonne 2001). Pickles, Starr et al. (2000) used a family history method with the relatives of 149 probands with autism. They found a relative risk of having a close family member with ASD traits is 3.88 for those with autism compared to those with Down's syndrome. This is support for the idea that autism is inherited in a continuum of traits including the disorder itself as an extreme.

There has been research investigating which particular genes may be involved. Given the preponderance of male cases of autism it was suspected that a recessive X linked gene may be responsible. However, simple Mendelian inheritance has been rejected in favour of the theory that more than one, probably 5-10, interacting genes may be

involved. Chromosomes 2, 7, 16 and 17 have been implicated (Medical Research Council 2001). Genome scans and linkage studies have identified many possible genes; for example, Ramoz, Reichert et al. (2004) have found linkage between the gene for mitochondrial aspartate/glutamate carrier and autism. The gene is on 2q. There are no linkage findings that have been replicated independently.
Chromosomal rearrangements and specific gene mutations that are associated with autism have been identified. For example, duplication of chromosome 15q11-13 (Baker, Piven et al. 1994) and mutations in the X linked genes neuroligin-3 (NLGN-3) and NLGN-4 can cause autism (Jamain, Quach et al. 2003; Zoghbi 2003). These known genetic causes are not implicated in the vast majority of cases of autism but they may provide clues as to causal mechanisms.

One of the Pervasive Developmental Disorders, Rett syndrome, has an identified genetic cause (Williamson and Christodoulou 2006). It has a highly characteristic developmental course. Months of normal development are followed by regression with the loss of motor control accompanied by stereotypic hand-wringing movements and complete loss of social and communicative behaviour. Mental retardation is usually present. Unlike autism, children with Rett syndrome are then largely stable in their presentation and show less subsequent development in their abilities. The genetic cause of Rett syndrome has been identified as mutation of the gene coding for MECP2. This gene is found on the X chromosome. MECP2 is a protein that controls the expression of other genes. It was thought that Rett syndrome only affected females but the mutation has also been identified in males where it produces much more severe effects. The most severe expression in males causes a mental retardation syndrome and early death. Now that the genetic cause of Rett syndrome has been identified the activity of the protein MECP2 can be studied. Study of the pathways that this protein is involved in may suggest other possible mechanisms that could have similar behavioural sequelae.

It has been suggested for both Rett syndrome and cases of autism where there is a period of normal development preceding the onset of symptoms, that they may both be

disorders of synaptic modulation or maintenance (Zoghbi 2003). This is based on the level of cortical development attained at the onset of symptoms and on the premise that molecules such as NLGN3 and 4, which have been implicated in autism, are cell adhesion molecules with a role in both pre and postsynaptic differentiation.

3. Other genetic disorders

A number of genetic disorders share features with autism. Thanks to genetic testing, differentiating between these disorders and autism is relatively straightforward. The disorders outlined below are very rare. All these disorders are associated with intellectual disability. It is not suggested that the generality of people with autism suffer from a minor form of these or a similar disorder. The particular interest of the association of these rare disorders and autism is the fact that they have a known, demonstrable biological basis. Examination of the way the biological basis of these disorders is associated with autism may cast light on the pathophysiology of the generality of autism cases.

a. Fragile X

Fragile X syndrome is caused by an excessive repetition of a 3-nucleotide sequence (CGG). Affected individuals show problems in relating to other people and have difficulty regulating attention, arousal and activity levels. It has been estimated that 15-25% of those with fragile X also have autism (McCabe, de la Cruz et al. 1999). People with fragile X can recognise facial expressions and emotions in others (Turk and Cornish 1998). Males with Fragile X are much more severely affected than females. In all those who have the disorder the degree of impairment depends on the length and amount of methylation of the repeat sequence. Over 200 repeats produce the full disorder. There is considerable interest in the finding that individuals with intermediate mutations of 50-200 repeats, a premutation, have more subtle deficits (Farzin, Perry et al. 2006).

b. Tuberous Sclerosis

This is an autosomal dominant genetic disorder associated with mutations in the TSC1 on 9q or TSC2 on 16p (Cheadle, Reeve et al. 2000). It results in the growth of hamartomatous lesions on multiple organs. In the brain, these are known as tubers. There is a high concordance of ASD and tuberous sclerosis, with 25% of people with tuberous sclerosis estimated to have autism (Baker, Piven et al. 1998; Smalley 1998). The prevalence of tuberous sclerosis in autism is about 1% (Harrison and Bolton 1997). It has been shown that the positioning of the tubers on the brain affects whether autism occurs. For example, tubers on the temporal lobes have been associated with ASD (Bolton and Griffith 1997). Humphrey and colleagues (2004) described two monozygotic twins with very different outcome, one with mild intellectual disability plus autism, the other with much less impaired functioning. The one who received a diagnosis of autism had much larger but fewer tubers and the other a greater number of smaller tubers.

Epilepsy is an extremely common feature of tuberous sclerosis affecting 80-90% of cases (Ess 2006). Tubers are often foci for epileptic activity. Bolton et al. (2002) found that a diagnosis of autism and tuberous sclerosis was associated with epileptiform discharges in the temporal lobe that began in early infancy. This was a stronger predictor of autism than the presence of temporal tubers alone indicating that epileptic discharges may have a role in the neuropathology of autism in this disorder.

c. Untreated Phenylketonuria (PKU)

Phenylketonuria is an autosomal recessive disease that affects the metabolism of the amino acid phenylalanine. When this disorder is untreated, it results in a build up of phenylalanine in the body. When levels of this amino acid reach a certain threshold in the extra-cellular fluid then damage to brain tissue occurs. In particular, it produces damage to the myelin that surrounds nerve cells, the loss of neurons themselves and the

loss of connections between nerve cells. There is an established link between untreated PKU and autism (Baieli, Pavone et al. 2003). In their study, Baieli, Pavone et al. (2003) found that out of 62 cases of early-identified treated PKU there were no cases of autism. Out of 35 cases of classic PKU that were untreated, all had intellectual disability and two boys had autistic disorder. This rate of 5.7% in this group is much higher than in the general population. Routine neonatal testing now identifies PKU so untreated cases are extremely rare.

d. Williams Syndrome

Another genetic disorder that has social consequences is Williams syndrome. It is caused by a microdeletion on chromosome 7s (7q11.23). Affected individuals are described as socially disinhibited, but have few friends. They have deficits in spatial cognition (Bellugi, Lichtenberger et al. 2000). They perform well however on language and face processing tasks (Bellugi, Bihrle et al. 1990). They are also described as having inattention, over-activity, ADHD, impulsivity and anxiety. Williams syndrome does not produce autism; in fact it has been suggested as the anti-syndrome, with the precocious sociability of these children the opposite of the pattern found in autism, although this has now been discounted. Nevertheless the study of genetic disorders affecting social behaviour can be informative to research on autism.

Muhle et al. (2004) in their review of the genetics of autism conclude that diagnosable medical conditions, cytogenetic abnormalities and single gene defects account for <10% of cases of autism. The other 90% have to be accounted for by the interaction of multiple genes and environmental factors. The identification of these genes may depend on refining further the phenotypes analysed in these genetic studies.

The findings from the twin and family studies cited above give support for a heritable component to autism, estimates of heritability range up to 90%. No specific genetic loci have yet been identified and independently replicated as being causally related to the

disorder. The reason for the lack of conclusive findings even in very large genetic studies may be due to heterogeneity in the population. It supports the notion that there may be etiologically distinct subpopulations. Researchers are currently looking at ways of defining subpopulations to look for genetic linkage among a more restricted range of autism phenotypes. Possible candidates are presence or absence of language, presence or absence of mental retardation, regressive subtypes, screening for co-morbid disorders and the use of cognitive phenotypes.

Conclusions

In summary, autism is a complex genetic disorder that in combination with environmental factors affects the development of the brain. Evidence is growing for some kind of immune dysregulation in autism but further research is needed to determine whether this is primary to the disorder or merely a secondary effect of disordered brain development.

Meanwhile research is starting to make progress in describing brain development in autism, outlining differences in brain development at the structural and cellular levels. Although there is much hope for future progress in these areas, research is at an early stage and cannot yet describe specific pathological process/es for autism.

This leaves the diagnostic criteria as the only means of defining cases. Given this lack of biological criteria, the disorder of autism unavoidably has fuzzy boundaries and there is considerable overlap with other disorders. The diagnostic differentiation of autism from other disorders will now be described.

Autism and other disorders – diagnostic differentiation

In all types of psychopathology diagnosed in childhood, there are high rates of comorbidity. Young people with ASD often have features of Attention Deficit Hyperactivity Disorder, Obsessive Compulsive Disorder, sleep disorders and or eating disorders (Brereton, Tonge et al. 2006). Adolescents with ASD may be particularly vulnerable to depression and anxiety (Bradley and Bolton 2006). Where a young person meets the criteria for multiple disorders, it is the symptoms with most prominence at the time of evaluation that determine what diagnosis is given. There are also constraints in the diagnostic systems over which disorders are exclusionary. For example, the DSM-IV (APA, 1994) system does not permit ADHD and autistic disorder to be diagnosed in one individual. It is implied that some symptoms of hyperactivity or lack of attention are part of the PDD and so do not necessitate a separate diagnosis. This is an example of the symptoms of one disorder being a core part of another disorder. The same is true of autism and sleep disorders; sleep disorders are so prevalent amongst children with ASD that they are often considered part of the ASD. Richdale (1999) reports estimates of the prevalence of sleep disorders in autistic disorder to be between 44 and 83 percent. Williams and colleagues (2004) found that over 40% of respondents to their survey of parents of children with PDD had children with problems getting to sleep and with patterns of restless sleep.

A slightly different example is that of Obsessive Compulsive Disorder and autism. It has been suggested that although young people with ASD show repetitive ritualistic behaviour this is not as ego-dystonic as the behaviour in Obsessive Compulsive Disorder (Baron-Cohen 1990). In OCD, the person dislikes doing the action but performs it for fear of the consequences of not doing it, albeit in a superstitious sense. In ASD, repetitive behaviours can be recognised as unusual by the doers but not as unpleasant or intrusive in themselves. This is an example of the need to go beyond behavioural definitions to look at the underlying cognition in order to distinguish types of psychopathology.

Prior to the 1940s, children with symptoms of autism would often have been diagnosed with a childhood psychosis. Although now it is accepted that autism and schizophrenia have distinct developmental courses, there is a behavioural overlap between the negative symptoms of schizophrenia and autism. Konstantareas and Hewitt (2001) examined this diagnostic overlap. They used the Structured Clinical interview (SCID), the schedule for positive symptoms (SAPS), the scale for negative symptoms (SANS) of schizophrenia, the childhood autism rating scale (CARS) and the DSM-IIIR. None of the people with schizophrenia met criteria for autism but half of those with autism met criteria for schizophrenia with negative symptoms.

Schizoid personality disorder in childhood

Wolff (1995) identifies a substantial overlap between children who are 'loners' in childhood, described by her as schizoid, and the children described by Asperger in his original paper. The children she describes are not as impaired as children with autism. They are socially isolated, lack empathic concern for others while being personally very sensitive, have an extremely active fantasy life, can be introverted or extroverted, have special interests and often have specific developmental delays. The outcome for these children is more positive than for the generality of children with autism, some being particularly gifted.

The cluster A personality disorders of adulthood in DSM-IV - schizoid, schizotypal and paranoid - are those that are associated with schizophrenia, that is they have been found to be more common in people who go on to develop schizophrenia and in their relatives. The relationship is not obligatory however; the majority of people with these personality types do not become ill. The schizoid disorder of childhood occurs when these same personality features are displayed in childhood. Of the children studied by Wolff, a minority did go on to develop schizophrenia (4%) which is elevated, compared to under 1% of the general population developing schizophrenia by the age of follow up. Wolff

also found that IQ was a protective factor for children with schizoid personalities and that those with higher IQs were less likely to become ill. However, the relationship between IQ and the prodromal states is not clear-cut and a depression of IQ may be part of the schizophrenic illness prodrome.

Semantic pragmatic disorder

Semantic pragmatic disorder is a specific language impairment that also has an overlap with Asperger disorder. As the label suggests, children with semantic-pragmatic disorder have preserved syntax and phonetics but have difficulty using language as a communicative tool. Asperger commented on the same characteristic in the children he described in 1944. In semantic-pragmatic disorder however, the deficits are limited to communication and do not affect more general social interaction. Studies examining the usefulness of the ADI and ADOS to discriminate ASD from specific language impairment have shown very little misclassification when the tools are used by experienced clinicians (Noterdaeme, Kurz et al. 1999; Mildenberger, Sitter et al. 2001). In addition to overlaps with other psychiatric disorders, there are also overlaps in symptomatology with sensory disorders.

Congenitally blind children

Brown and colleagues examined the behaviour of congenitally blind children for autistic-like features (Brown, Hobson et al. 1997). This was assessed by teacher report and systematic observations. They found autistic-like features were more common in the blind children, compared with sighted controls. They also compared 9 blind children with IQs of less than 70 with 9 group-matched participants with autism and found a 'substantial overlap in clinical presentation, despite subtle differences in clinical impression'.

Another area where there is considerable overlap is between autism and intellectual disability.

Autism and intellectual disability

Kanner originally stated that the children he described were of normal intelligence but that their autistic difficulties prevented them from functioning adaptively. Later researchers established that the intellectual deficits shown by children with autism were stable and not distinguishable from the problems shown by children with intellectual disability alone. It was then accepted that many children with autism were intellectually impaired. Until recently, it was estimated that around 75% of cases of autistic disorder there was also intellectual disability (DSM-IV, APA1994). Due to the wider definitions of the disorder currently in use, estimates are now that around 25-40% of cases of ASD also have IQs below 70 (Baird, Charman et al. 2000; Chakrabarti and Fombonne 2001). These children will have general developmental delay on all aspects of functioning but their social and communication skills will lag behind other areas of development.

The prevalence of PDD is much higher in people with intellectual disability than in the generality of the population. For the population as a whole, the MRC Review of autism research: epidemiology and causes (2001) concluded that the prevalence for autistic spectrum disorders is approximately 60 and narrowly defined autism 10-30, per 10000 children under 8. Prevalence in the adult population is not known. Among people with mild intellectual disability, prevalence estimates for PDD vary, for example from 1.8% (Wing 1981) to 18% (Kraijer 1997). The second rate found by Kraijer is much higher, as this sample included people who had been institutionalised. It might be expected that such a population would have a much greater degree of co-morbid problems. Among people with severe intellectual disability, the rate rises sharply with estimates of 48.4% (Bouras, Holt et al. 1999), 48.5% (Gillberg 1986; Gillberg, Persson et al. 1986), 40.8% (Kraijer 1997) and 41.5% (La Malfa, Lassi et al. 2004). It is clear that there exists a strong relationship between PDDs and intellectual disability. However, the reasons for this are far from clear.

The relationship between the various aspects of intelligence appears to be different in autism compared with the general population. In ASD there is often found to be a highly uneven profile of subtest scores on the standard IQ tests (Asarnow, Tanguay et al. 1987). This indicates a looser association between the various facets of intelligence in people with autistic spectrum disorders than is found in typically developing individuals. In typically developing individuals, performance across the subtests is uniform (Jensen 2000).

Some researchers have characterised Asperger disorder as a type of specific learning disability. Klin and colleagues compared the neuropsychological profiles of Asperger subjects and high functioning participants with autism (Klin, Volkmar et al. 1995). They found that the profile for Asperger disorder converged with that for non-verbal learning disabilities. Non-verbal learning disabilities include deficits in visual-perceptual-organisational psychomotor coordination and complex tactile perceptual skills (Harnadek and Rourke 1994). This indicates that even where there is normal overall intelligence people with Asperger's often show a distinctive profile on standard tests of intelligence.

The phenomenon of savant abilities supports this view of a different relationship between 'intelligences' in ASD.

Savant abilities.

Savant abilities are specific talents very much out of line with general level of functioning.

Hermelin and O'Connor have studied savant abilities in individuals with intellectual disability. About 10% of people with autism are estimated to have a savant ability (Rimland 1978). Hermelin and O'Connor examined the domains in which savant abilities occur and mechanisms that may underlie these islets of superior ability. In her recent book, Bright splinters of the mind (2001), Hermelin outlines current research findings in this field.

Hermelin gives an example of the case of a man with mental retardation who learnt many foreign languages. It was found that he used vocabulary as the building blocks for his competence but often transposed this lexical knowledge onto English grammar. Calendar calculators are savants who when given a date, even 30 years in the future, can tell you what day of the week it will fall on. Hermelin attributes this ability to a strong memory for dates and extraction of the regularities underlying the calendar. For example, one of the regularities is the 28 year rule, i.e. the calendar repeats itself every 28 years. There are also correspondences between months, so that the day-date relationships in March are the same as in November. Through constant exposure to their interest, savants extract an implicit knowledge of the sequential daily and weekly structure of the calendar.

The common themes underlying savant abilities appear to be the capacity to extract regularities from a system over high levels of exposure to it and an above average long term memory.

Savant abilities are a challenge to general theories of intelligence. Theories of intelligence are divided between those that propose a single underlying factor, such as Spearman's 'g' (Spearman 1904), as measured by intelligence tests and those that propose a multiple intelligences model such as Gardner (Gardner 1983).

A comprehensive theory of intelligence would have to explain both the fact that although some people are good at maths and bad at spelling, knowing something about a person's performance in one cognitive domain usually allows you to predict something about their performance in others. The factor analysis of intelligence tests' batteries has been shown to yield a single underlying factor: proponents of the alternative view that there are multiple dissociated intelligences have been unable to create tests that are not correlated with each other to some extent. There is also evidence that this unitary intelligence has an underlying biological basis that is heritable (Neisser, Boodoo et al. 1996). Alternatively, a theory would also have to cope with the aforementioned savant abilities that argue for some level of separation between some types of intelligence and overall processing capacity. In addition, in cases of focal brain injury where cognitive

capabilities become dissociated, it is clear that there is some independence between domains of intelligence.

Anderson's minimum cognitive architecture is a theory of intelligence that also has a developmental perspective (Anderson 2001). Anderson argues that standard intelligence tests measure knowledge and that there are two routes by which this knowledge is created. The first is by thought. Thought can be in the verbal domain or the visuospatial domain and it is through the action of thought that knowledge is created. A central processor accomplishes this action of 'Thought'. Differences in the efficiency of the processor are the source of individual differences in IQ. The other route to knowledge is through dedicated modules. Modules govern capacities such as syntactic parsing, Theory of Mind and 3-D perception. These modules are encapsulated, automatic and become mature through development. They produce knowledge directly and are not dependent on the central processor. The rates at which these different modules come online are the source of developmental differences in intelligence. Differences in intellectual capacities *between* age groups are due to structural differences in the architecture of intelligence due to the maturation of modules. In particular, the executive functions are modules that mature through development. This model therefore suggests that differences between individuals *within* age groups are due to the efficiency of the basic processor. This model goes some way towards accommodating both a general intelligence and domain specific intelligences.

There have been attempts to isolate a basic processing factor. Inspection time tasks are those that attempt to tap a basic speed of processing constraint that may underlie the correlation between different tests of intelligence across domains. In inspection time tasks, the presentation of stimulus items is very brief and followed by a masking stimulus (Nettelbeck and Lally 1976). Testing aims to find the minimum presentation time necessary for the subject to make an elementary judgement about a stimulus. The correlation between inspection time measures and IQ in adults is around 0.5 (Nettelbeck 1987; Kranzler and Jensen 1989).

Due to the uneven cognitive profile on standard intelligence tests and the presence of savant abilities in a significant proportion of people with autism, the nature of intelligence in autism is more mysterious even than in the rest of the population. Anderson, O'Connor and Hermelin examined the inspection time for an autistic savant prime number calculator (Anderson, O'Connor et al. 1998). His inspection time was at odds with his low IQ but more in keeping with his calculating abilities. Scheuffgen, Happé, Anderson, and Frith (2000) found that a group of children with autism (IQ 1 standard deviation below average) had lower inspection times than an IQ matched control group (Scheuffgen, Happé et al. 2000). The authors suggest that, in autism, damage to a particular module has implications for IQ but not for the central processor.

It has been found that the prevalence of autism increases with decreasing IQ levels. Wing found prevalence for autism of 1 per 10000 for subjects with normal IQs to 82.2% for those with IQs of less than 20 (Wing 1981). Many studies of autism focus on people of normal intellectual functioning in order to rule out confounds due to the presence of intellectual disability. However, given the distinctive nature of the relationship between autism and intellectual disability, it may be intrinsic to understanding the disorder.

We have not yet defined what is meant by intellectual disability. When trying to define what 'low' or 'normal' intelligence is in order to examine its relationship to psychopathology, it is necessary to operationalise it. This often coincides with definitions used for service delivery as this is how potential participants will be identified. Large scale studies of mental illness and intelligence have often used diagnosis of intellectual disability as a way of operationalising lower intelligence

Definitions of Intellectual Disability

Firstly, it is necessary to say something about terminology. Efforts to find terms that are not offensive to describe people with intellectual disability have resulted in frequent changes in terminology.

Currently, in America the term 'mental retardation' is used to describe people with intellectual disability. The Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision (DSM-IV-TR: American Psychiatric Association 2000) defines mental retardation as follows:

- Significantly subaverage intellectual functioning - An intelligence quotient (IQ) of approximately 70 or below
- Concurrent deficits or impairments in adaptive functioning in at least 2 of the following areas: communication, self-care, home living, social/interpersonal skills, use of community resources, self-direction, functional academic skills, work, leisure, health, and safety
- Onset before age 18 years

In Britain, the term 'learning difficulties' or 'intellectual disability' is used. In America the term 'learning disability' refers to specific difficulties such as dyslexia rather than more global impairments in functioning.
The ICD-10 classifies levels of intellectual disability: an IQ of 50-70 is mild, 35-49 moderate, 20-34 severe and less than 20 profound.

The History of the Distinction between Mental Illness and Mental Retardation.

The task of trying to understand the relationship between psychiatric disorders and mental retardation has a long history. Miller (1995) and Linaker and Nitter (1990) have described the history of the distinction between mental illness and mental retardation:
In the very early nineteenth century eminent physicians such as Pinel (1801)thought that mental retardation was a medical, psychiatric disorder of reason and either acquired or congenital. This implies that mental retardation is an illness and is not differentiated from other forms of mental illness.
Esquirol, in 1828, distinguished between mental retardation and psychiatric disorders, and decided they were separate entities (Hayman 1939). Georget, writing in the first half of the nineteenth century, also agreed with this view. This is a departure from the 'disease' concept and views mental retardation as failure of development.

There were others at this time, for example Phelps, who held that mental illness and mental retardation were part of a continuum that could not be entirely separated.
Prior to the 1840s, people with mental retardation were admitted to the same asylums as those with mental illness. From the 1840s onwards, separate institutions were introduced in both Europe and America, although the emphasis was on treating children.
Tregold (1908) thought that mental disorders of the same kind as seen in the normal population were found in the mentally retarded, but the latter group were much more frequently afflicted. Kraepelin held an intermediate position and suggested that some cases of dementia praecox developed as a result of mental retardation and that some cases of mental retardation resulted from an early onset psychosis.
Pritchard also distinguished mental retardation from mental illness and linked mental retardation to normal continuum. Sollier (1901) developed the idea of comparing level of development in mental retardation to ages of the normal child.
Binet and Simon in the early 20th century worked on the first quantitative measure of level of intelligence for the French government. These became the first IQ tests. They

were developed for use in education to identify those in need of extra support but have since been extended for use with adults.

People with mental retardation can be identified by a combination of low IQ score and impaired adaptive functioning. Mental retardation is always present from birth. Mental illness on the other hand appears against a background of normal early development. Despite this clear separation in the course of mental illness versus mental retardation there is evidence of a link between them. Before looking at the link between the two, we need to see how mental illness is diagnosed in the presence of mental retardation.

Diagnostic Issues in Mental Retardation

Differentiating mental retardation and mental illness is an on-going issue. Diagnostic overshadowing is a problem described by Reiss (1982) where, in the presence of intellectual disability, psychiatric problems maybe overlooked. There is a tendency for potential symptoms of mental illness to be grouped together as behavioural problems and for these to be seen as secondary to the effects of intellectual disability. For this reason, it is suspected that psychiatric disturbances are very much under-diagnosed in the intellectually disabled population. The DSM is a series of behavioural criteria and where these behaviours are present; there are grounds for diagnosis to be made, whether or not these behaviours occur in someone with intellectual disability. It is necessary however for there to be adapted tools for use with intellectually disabled populations that have been validated within this group. There are also difficulties when the range of observed behaviours is extremely low and there is an absence of verbal communication as, for instance, the presence or absence of delusions cannot be established in these cases. Psychiatric diagnosis is largely based on the patient's subjective description of their inner mental experiences. Behavioural observation is secondary and in many cases there may be little disturbed behaviour evident even in the presence of severe illness: i.e. phenomena believed to be characteristic of specific disorders, e.g. thought disorder in schizophrenia, require advanced verbal ability to be expressed.

Prevalence of Psychopathology in Mental Retardation

In a study of the prevalence of co-morbidity for psychiatric disorders in people with mental retardation attending community based day programs in Chicago, the overall rate of co-morbidity was found to be 39% (Reiss 1990). This was estimated using two step methodology where subjects were first screened and then a sub-sample evaluated by a clinical psychologist. The authors conclude that the rates of serious mental illness were not high, but that people with mental retardation were more susceptible to personality problems such as hypersensitivity to rejection/criticism, excessive dependency and social inadequacy. Only 11.7% of the 205 subjects had a psychiatric diagnosis in their case files which suggested that mental illness was under-diagnosed in this group. Social functioning appeared to be a particular problem affecting 45.8% of the subjects based on the screening measure. In 8.9% of the subjects, this was found to be to such an extent that social inadequacy was a 'major problem' for these people. The Reiss Screen's definition of social inadequacy is 'has difficulty relating to peers in appropriate or satisfying ways'. The examples listed on the Reiss screen are 'has no friends, lacks appropriate social skills, insensitive to the feelings of other people'.

A population-based study conducted in Norway, Stromme and Diseth, sought referral of all children suspected of having intellectual disabilities (Stromme and Diseth 2000). They thoroughly investigated all these cases both psychometrically and clinically and found that out of 30037 children born in one county between 1980 and 1985, 178 had mental retardation, and of these 8% had PDDs.

The levels of psychopathology found in these studies are much higher than in the general population. This indicates that the separation between mental illness and mental retardation is not clear-cut and, for some reason, people with mental retardation have an increased liability to mental illness. This appears particularly to be the case with problems with social functioning. Understanding the cause of this liability depends on how mental retardation is construed.

Developmental issues in Mental Retardation

Another modern correlate of the argument about the relationship between mental illness and mental retardation is found in the mental retardation literature. There are two main strands.

1. One group or two groups debate (also known as developmental-difference debate).
    a. Difference theory suggests that all mental retardation is due to organic dysfunction. This position predicts that there will be specific deficits in cognitive functioning in people with mental retardation and they will have atypical cognitive development. ('One Group' hypothesis)
    b. Developmental theory is that the above position only applies to those whose retardation is due to an organic impairment. The rest are known as having cultural-familial retardation and are those occupying the lower portion of the normal distribution of intelligence. This predicts that these people will follow the same sequence of developmental milestones as the rest of the population but at a slower rate and their final level of functioning will be lower than average. ('Two Group' hypothesis Zigler 1967; Zigler 1969).
2. Similar structure and similar sequence hypotheses.
    a. Similar sequence hypothesis: related to the above debate is that about whether people with mental retardation proceed through the same developmental stages in the same order, as in the typically developing population.
    b. Similar structure hypothesis: whether the cognitive structures underlying these achievements are the same as in the typically developing population.

Most research has confirmed the similar sequence hypothesis concerning Piagetian sensorimotor development in intellectual disability(e.g.Morgan, Cutrer et al. 1989). This

picture may change as genetic studies uncover the specific etiologies of mental retardation.

Regarding the relationship of mental retardation with psychopathology, if the similar sequence and structure theories are correct, then the fact of having mental retardation should not predispose a person to mental illness other than by the extra stress caused by lower adaptive functioning and social stigma. Alternatively, if it is not correct it may be that the different way that cognition develops in those with mental retardation may produce a direct liability to some kinds of mental illness.

Conclusions

We have examined the overlap between autism and a number of disorders and these are shown in diagram 1.

Diagram 1 Autism and Other Disorders

TS- tuberous sclerosis
NF- neurofibromatosis
ID – intellectual disability
PDD – pervasive developmental disorder

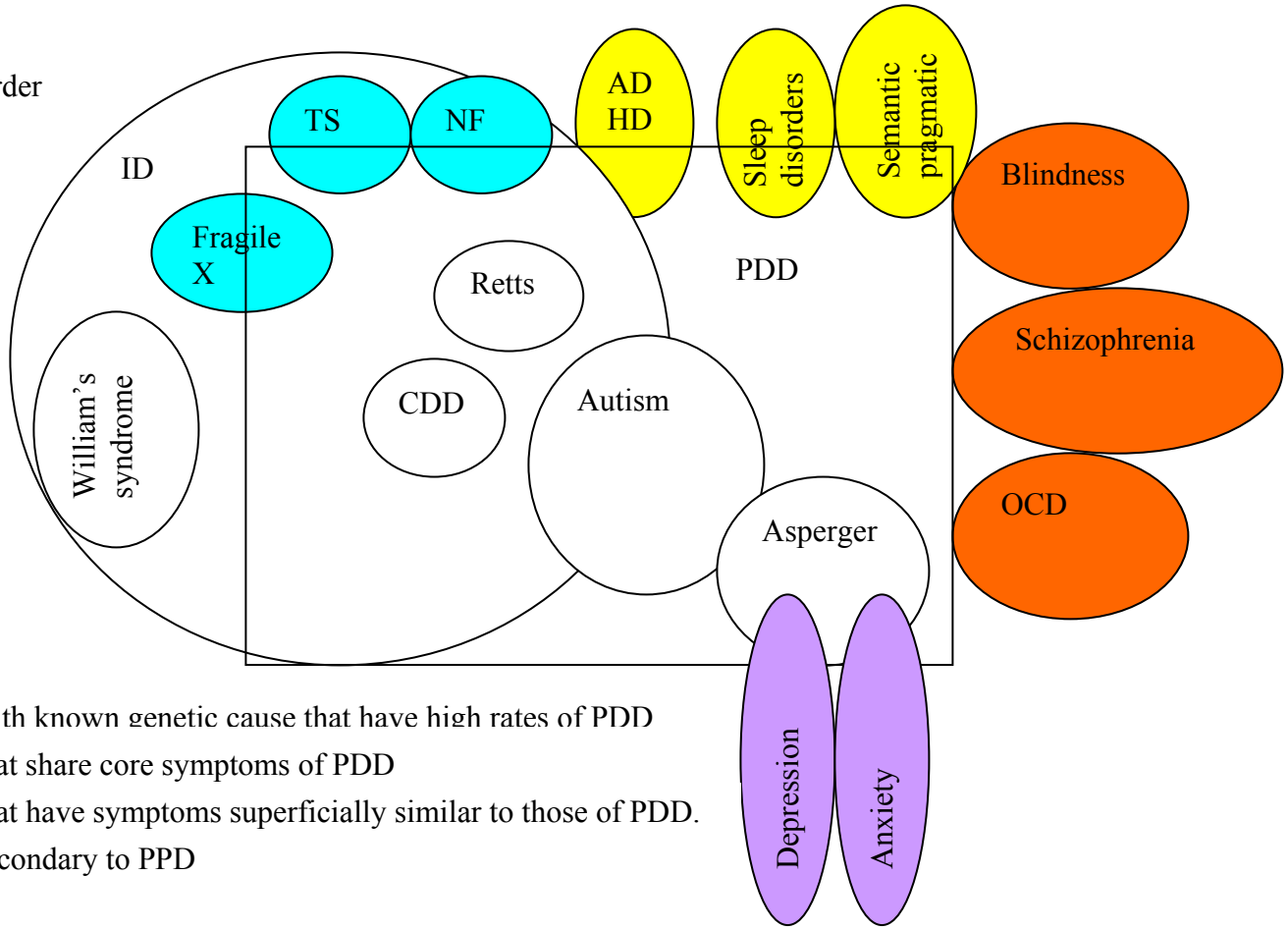


Diagram 1 shows that autism has many commonalities with other disorders. The diagram is not intended to convey the proportion of overlap between disorders, but merely to indicate where an overlap exists. Not all overlapping conditions have been shown.

The relationships between disorders are of four kinds:

1. Those with a common etiological mechanism.
2. Those with common core characteristics and
3. Those with superficial similarities.
4. Disorders secondary to PDD.

The genetic disorders with a common mechanism account for very few of the total number of cases of autism.

In the disorders that have common core symptoms (for example semantic pragmatic disorder and ASD) differentiation is based on extent rather than kind. In ASD there are social and language problems whereas in semantic-pragmatic disorder there are only pragmatic language difficulties.

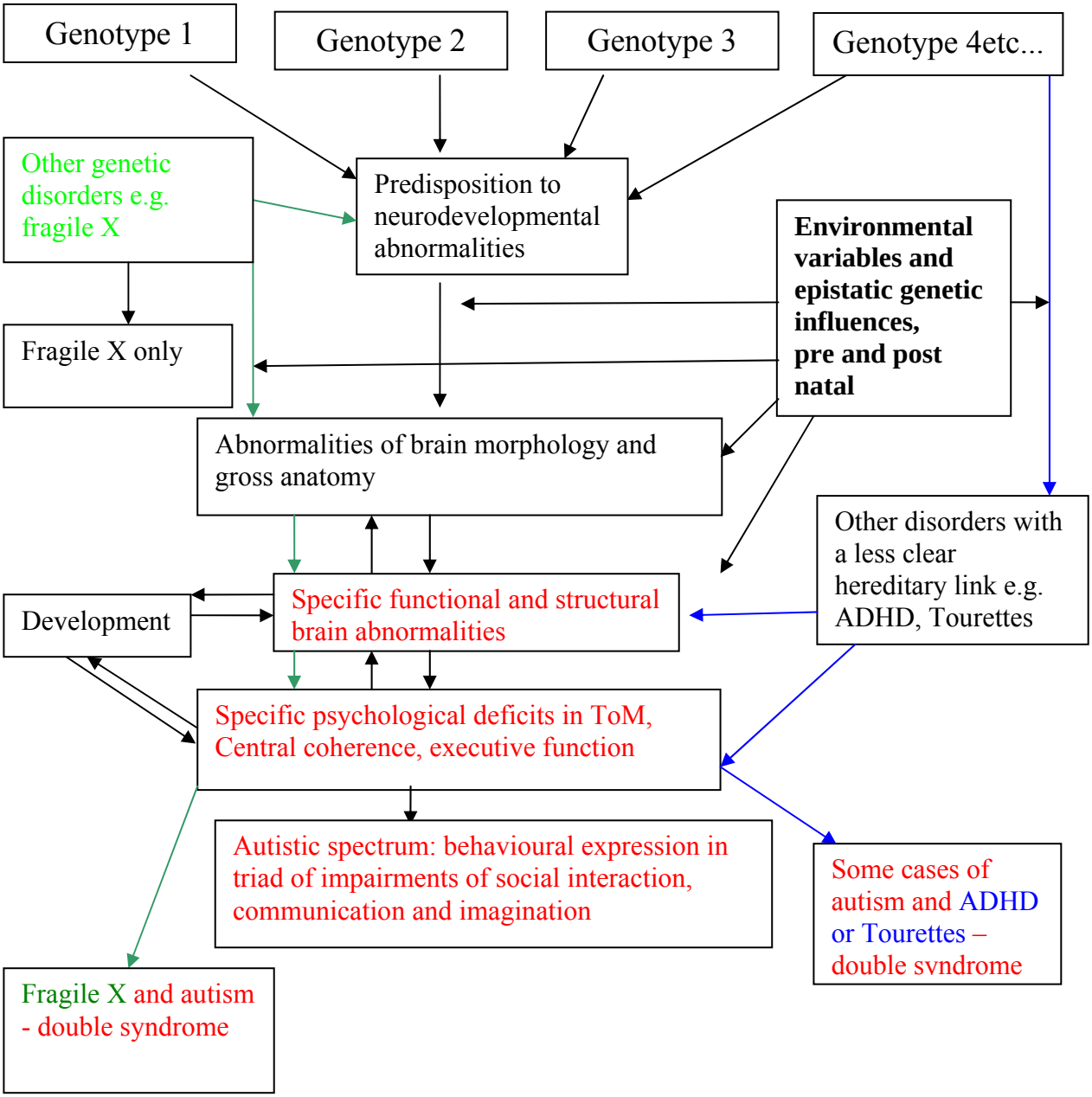
Autism is postulated to be part of a spectrum of disorders that are grades or variations of a single disorder which has highly variable manifestations. This spectrum of conditions also has indistinct boundaries with a number of other disorders as just outlined. Despite the overlaps between conditions the reliability and validity of the core definition of autistic disorder is good (Rutter 1989). The disorder can thus be defined at the behavioural level but not at the physiological level. It is possible that a clear distinction between disorders could be made based on the underlying cognitive characteristics. For example, Baron-Cohen (1990) has suggested that the compulsions of Obsessive Compulsive Disorder may be more ego-dystonic than in ASD. In schizophrenia, social problems and withdrawal may be due to acquired difficulties understanding other minds and/or monitoring own thoughts and behaviour. In autism the difficulty understanding

other minds is innate and present earlier in development. Superficially similar symptoms can be distinguished at another level of explanation, that of cognitive psychology. This may allow differentiation of these difficulties in a way not possible at the level of individual behaviours.

The psychological level of explanation may also help to determine whether the spectrum concept of autism is correct. Some disagree with the spectrum concept, for example, Gillberg and Coleman (2000) argue that there are multiple disease entities of autism and that autism is a collection of syndromes that will ultimately be disentangled. To determine which interpretation is correct spectrum or collection of syndromes the task is to determine where the final common pathway begins that leads to the triad of impairments in social interaction, communication and imagination. Then it can be determined whether they are a single or multiple disease entity.
The fact that a single biological marker for autism has yet to be found and the theory that multiple interacting genes mediate the disorder would argue that the disorder is etiologically heterogeneous. The spectrum concept of autism implies a single underlying factor common to all the various manifestations of autism. The heterogeneity of presentation would be due to environmental and personal variables. However, at what point in the pathway that leads to the expression of autism there is a single causal factor that is common to all forms is a moot point.

The diagram below shows a possible location for final common pathway in red.

Diagram 2. Pathways from Multiple Etiologies to the Triad of Impairments



It is likely that the final common pathway specific to autism involves discrete and limited changes to brain structure and function. As the core symptoms of autism can be present with average or above average IQ, gross and diffuse brain damage underlying the core symptoms is unlikely.

The link between the neurobiological level and the psychological level is reciprocal in line with the dynamic systems approach to neurodevelopmental disorders (Bishop 1997; Plunkett, Karmiloff-Smith et al. 1997) which shows that initial abnormal neurological development can produce abnormal psychology which has secondary effects on later brain development.

The explanation of autism will ultimately be at the level of interaction between brain development and function and there will probably be multiple combinations of genetic and environmental etiologies that can result in these core disturbances in brain systems which produce autistic behaviour. Psychology has a key role in directing the attempts to map brain behaviour relationships. The goal will be a 'vertical integration' (Churchland 1986) of symptoms and neurobiology.

If a primary psychological deficit for autism can be determined then this can direct attempts to locate the neural mechanism. Psychological theory is also informed by new discoveries about the brain in this reciprocal process.

Psychological Theories of Autism

The primary psychological deficit in autism must meet the criteria of universality. This means that it must be present in all cases. If there are cases of people with autism who do not have the deficit then it cannot be necessary for the disorder. The other criterion is uniqueness - that it is not present in other disorders. This means that the deficit must be sufficient to cause the disorder and thus must cause the disorder wherever it is present. The difficulty with ASD is its changing presentation over development. Is it necessary that a certain capacity should be completely absent? Is it sufficient that a certain capacity is out of step with other areas of development and can therefore have specific behavioural consequences? It is also a significant question whether a deficit at a certain point in development can be causative: it can be if it means certain opportunities for learning are then missed as this could have long reaching developmental consequences even if later compensation is made.

If cognitive markers of autism that meet the criteria for universality and specificity can be found then it supports the concept of autism as a single entity. To isolate cognitive characteristics that are specific and unique to autism necessitates the use of particular research methods.

Research methods

Hermelin and O'Connor did the first pioneering work systematically comparing the cognitive profile of people with autism with matched developmentally delayed control groups (e.g.Hermelin and O'Connor 1964). This was a major methodological development as it meant that the effects of general developmental delay were controlled for and any significant differences between groups were due to the autistic disorder alone. In this way, the specific pattern of psychological strengths and weaknesses could be investigated. Most other work at the psychological level has continued in this

tradition. In an ideal design both developmental level, that is, mental age and chronological age are controlled with two separate individually matched control groups.

We will now examine the evidence for the main theories of the cognitive basis of autism under the headings of social cognition, Weak Central Coherence and executive function.

Social cognition

Theory of Mind

'By Theory of Mind we mean being able to infer the full range of mental states (beliefs, desires, intentions, imagination, emotions etc.) that cause action. In brief, to be able to reflect on the contents of one's own and other's minds.'(Baron-Cohen, Tager-Flusberg et al. 2000). Theory of Mind is part of common folk psychology, the body of informal psychological knowledge that people use in their everyday lives to reason about the causes of other people's behaviour. Philosophers call it 'belief-desire psychology'. When thinking about other's actions for example 'I wonder why Jean did not say 'Hi!' when I saw her in Tesco's?' then we automatically turn to concepts such as belief, knowledge, attitudes, and emotions to produce possible explanations such as:

1. may be she is still angry with me for forgetting her birthday,
2. maybe she did not see me,
3. maybe she is preoccupied as she is moving house this week.

The term 'Theory of Mind' first came into the psychological literature in a paper by Premack and Woodruff in 1978. They had tried to teach chimpanzees to practice deceptive pointing. Deception is used as a measure of Theory of Mind as it requires reasoning about beliefs in order to manipulate behaviour. They were successful with two out of four animals. They argued that the chimpanzee's ability to predict what a human actor will do to achieve certain goals implies it has a Theory of Mind. The problem with these experiments, particularly in animals, is that it is hard to prove that

the animals are using a reasoning process and not acting on previously learnt behaviours based on environmental cues.

Philosophers Dennett and Pylyshyn argued that the only way to show that reasoning about behaviour is predicated on belief, is to show reasoning based on a false belief: reasoning about a true belief is conflated with reality and with the subject's own belief (Dennett 1978; Pylyshyn 1978). Wimmer and Perner studied the development of Theory of Mind in normally developing 3-5 year old children using such a false belief paradigm (Wimmer and Perner 1983). They found that children of four could not reason about a false belief but by five most children were able to do so.

In 1985, Baron-Cohen, Leslie and Frith published the paper 'Does the autistic child have a 'Theory of Mind'?' This paper described the application of the false belief paradigm to children with autism . This involved the Sally-Anne test, a false belief test slightly altered from the story about Maxi used by Wimmer and Perner In this test, one of a pair of dolls, Sally, places her marble in a box. When she has gone away the other doll, Anne, moves the marble to a basket. When the Sally doll is brought back, the child is asked, 'Where will Sally look for her marble?' Children of three years old will respond that Sally will look in the marble's current location, whereas older children of 4 to 5 years will say that she will look where she left it, thus showing evidence of understanding her false belief. Children with autism were found to have significantly more difficulty with this task than mental age matched controls. Out of 14 children with Down's syndrome (mean IQ 64), 86% passed. Out of 20 children with autism (mean IQ 82) 80% failed. The finding of deficits in Theory of Mind in children with autism relative to mental age matched controls is a robust one. Yirmiya and colleagues have confirmed this in a meta-analysis (Yirmiya, Erel et al. 1998). Happé also reviewed a number of studies of Theory of Mind in autism and found that the average mental age children with autism achieve the false belief task is 9 compared to 4-5 for normal children (Happé 1995).

Not all children with autism fail however. What about those who pass? Higher functioning people with autism sometimes can also go on to pass second order false belief tasks. These tasks require reasoning of the form 'John thinks that Mary thinks that…' Although a small minority of high-functioning people go on to pass these tests it has been suggested that they may use alternative methods rather than the more intuitive and possible innately determined mechanisms used by younger controls. This is supported by the fact that those people with autism who go on to pass these tests still experience considerable difficulty in day-to-day social interaction.

There has been a large amount of research developing and extending these findings regarding Theory of Mind in a number of directions. There has been an interaction between research with typically developing children and participants with autism. The following list gives a brief overview of the many ways in which the concept of theory of mind has been explored.

1. Facilitating performance on false belief tasks such that normal children as young as three can pass these tasks under certain conditions
    a. Decreasing the salience of the real location of the object by putting it out of sight
    b. Changes to the questions asked e.g. 'Where will Sally *first* look for her ball?' as opposed to 'Where will Sally look for her ball?'
    c. Reducing the verbal skills required - a 'pointing only' response, for example.

2. Carefully constructed control tasks
    a. False photograph task

3. Variation in modes of presentation
    a. Acted out with models
    b. Story book presentation

c. Video
d. Real life actors

4. Developing other tasks requiring false belief reasoning
    a. Appearance reality tasks
    b. Own false belief tasks
    c. Deception/sabotage
    d. Level II perspective taking
    e. Droodle task (Taylor 1988)
    f. Increased reliability of results when using more than one false belief task

5. Extending studies of Theory of Mind to other age groups.
    a. Second order false belief tasks - more advanced tests of false belief reasoning passed usually by 8 years old (in participants with autism (Baron-Cohen 1989).
    b. Precursors to Theory of Mind in infants
        i. Imitation
        ii. Joint attention
        iii. Social referencing

    c. Precursors to Theory of Mind in pre-schoolers
        i. Pretence
        ii. False beliefs about intentions (Moses and Flavell 1990) and values
        iii. Vocabulary about perception and emotion
        iv. Understanding that desires lead to actions
        v. Seeing leads to knowing
        vi. Intended vs. unintended actions

d. Theory of Mind in adults
    i. Hinting task
    ii. Happé's strange stories
    iii. Irony/sarcasm
    iv. Visual jokes
    v. Mind in the eyes, Mind in the Voice
    vi. Theory of Mind in older adults. Happé and colleagues found that healthy older adults perform better than young adults on Theory of Mind stories (Happé, Winner and Brownell 1998).

6. Relationship of experimental Theory of Mind skills to real life social skills
    a. correlation with social skills as measured by Vineland Adaptive Behaviour scales
    b. presence and sophistication of pretend play

7. Theory of Mind and language development
    a. Syntax – mastery of embedded complements necessary for Theory of Mind
    b. semantics - relationship between using mentalistic terms in real life and experimental Theory of Mind competence
    c. pragmatics
    d. conversational maxims
    e. correlation with verbal IQ
    f. relevance theory

8. Theory of Mind and cognitive complexity

9. Theory of Mind and executive function

10. Theory of Mind in other groups

a. Schizophrenia
b. Deaf and blind
c. Psychopathy and other personality disorders
d. Conduct disorder
e. Right hemisphere / frontal focal brain injury
f. Tourette's syndrome

11. Neuroimaging studies of Theory of Mind

12. Theory of Mind skills in the relatives of children with autism

13. Longitudinal studies of the development of Theory of Mind

14. Cross cultural studies of the development of Theory of Mind

15. Teaching Theory of Mind
    a. to children with autism via photograph in the head technique
    b. to children under three.

16. Theoretical models of the development of Theory of Mind and of autism
    a. Leslie – m-representations
    b. Baron-Cohen – deficient folk psychology
    c. Theories about the development of Theory of Mind range from
        i. 'Theory' theory – that children *construct* a Theory of Mind as an empirical theory that is then revised and updated throughout development. This necessitates both the capacity to develop theories and possibly an innate starting theory (Gopnik, 1993).
        ii. Simulation theory (Harris 1991; Carruthers and Smith 1996)
        iii. Maturational accounts
        iv. Strong modularised accounts (ToMM) (Leslie 1987)

Factors Influencing the Development of Theory of Mind

1. Early Social Development – Precursors

In autism, there is an absence of early imitative behaviours. Typically developing infants are able to imitate facial expressions from the first few hours after birth (Meltzoff and Moore 1977). Rogers and colleagues have shown that toddlers with autism are deficient in their ability to imitate facial expressions and actions on objects (Rogers, Hepburn et al. 2003). Rogers and Pennington proposed that this imitation deficit might be primary in autism (Rogers and Pennington 1991). Early imitative behaviours may provide a foundation for the development of Theory of Mind (Meltzoff and Decety 2003). The early correspondences that map others' behaviour onto one's own would provide a foundation for understanding that other people are the same as oneself. They suggest that this provides the basis for a simulative generation of a Theory of Mind.

There are differences in social attention in young children with autism. Swettenham and colleagues studied spontaneous switches of attention during free play (Swettenham, Baron-Cohen et al. 1998). They found that children with autism spent more time looking at objects and less time looking at people: they also switched attention less frequently between the two. Children with autism also do not engage in joint attention behaviours, such as drawing adults' attention to items of interest. Joint attention typically develops between the ages of six and twelve months: it involves sharing attention with others e.g. by pointing.

Charman has suggested that joint attention deficits are core to autism. In a longitudinal study of infants with autism and PDD between 20 and 42 months, he found that their joint attention abilities were positively related to language outcomes at 42 months and

negatively related to social difficulties (Charman 2002). Particular types of joint attention behaviours were found to have this predictive relationship to later development – such as declarative behaviours and triadic gaze shifting. Imperative joint attention behaviours which have a more functional use such as 'I want a drink' were not found to be predictive of later social behaviours. These predictive relationships were restricted to the social and communicative aspects of autism and did not predict the presence or level of stereotyped behaviours.

It has been suggested that gaze following is necessary for the development of joint attention. Gaze following has been found to be deficient in autism through observation studies conducted in naturalistic settings (Leekam, Lopez et al. 2000). Swettenham et al. (2003) have shown that deficits or delays in following eye direction are not due to perceptual or attention deficits, since reflexive responding to eye gaze is intact in autism (Swettenham, Condie et al. 2003; Kylliainen and Hietanen 2004).

2. Intellectual functioning

There is considerable evidence that Theory of Mind ability increases with development. It is a matter of debate whether this reflects solely the maturation of a dedicated module or also reflects increases in general information-processing capacity. Andrews et al (2003) sought to determine how much of the age related variance in Theory of Mind performance was explained by the capacity to manage relations of increasing complexity (Andrews, Halford et al. 2003). Relational complexity theory predicts that most tasks can be decomposed into the number of underlying relationships between arguments. With increasing age, children develop the ability to handle relations with increasing numbers of arguments.

Andrews et al. (2003) propose that Theory of Mind tests require the capacity to handle ternary relations, that is, relations with three arguments. These can be expressed as (cue, setting condition, response) or (where will Sally look?, false belief, original location) vs.

(where will Sally look?, reality, new location). The argument is that children under three can only process two relations so that there can be no influence of setting condition. Andrews et al. (2003) found that much of the variance in false belief task performance attributable to age was subsumed by performance on equivalent tasks (ternary relations - tasks such as transitive inference). This demonstrated that there are information-processing constraints on the performance of false belief tasks. However, not all of the age-related variance was predicted in this way and the variance in task performance unrelated to age needs still to be explained by social cognitive mechanisms independent of information processing capacity.

There is a relationship between false belief task performance and verbal IQ. This also can be interpreted as an information-processing capacity constraint on social cognitive development. Alternatively, this has been reframed and researchers have asked whether a degree of language competence is necessary to develop a Theory of Mind (de Villiers 2000). This could be through both the exposure and use of mental and emotional linguistic terms or more generally, through the exposure to social stimuli and socialisation that language provides. Alternatively, it could be that language provides the cognitive tools necessary for false belief reasoning. It has been argued that mastery of embedded complements does provide such a tool. Embedded complements are syntactical structures such as 'John thinks that the cat is dead' or 'Dad said that the cat is dead'. They have the property that the complement can be false and the sentence can be true. The cat does not have to be dead for Dad to have really said 'the cat is dead'. Tager-Flusberg argues that these syntactic structures provide children with the cognitive tools for false belief reasoning (Tager-Flusberg 2000). To test this, they have examined the relationship between the use of these forms in expressive language and performance on false belief tasks: a close relationship between the two was found.

3. Theory of Mind and Self Awareness

We have a powerful sense that our awareness of our own mental states is immediate and direct, much as our perceptions of the external world are immediate and direct. This is in contrast to our awareness of other people's mental states, which must be inferred. In fact, the problem of whether anyone else has mental states akin to our own is an ancient philosophical problem known as the ontological question.

The primacy of perception of our own mental states has been challenged in many ways. For a start, one can be mistaken about one's own mental states as one can about perception. Memory for own motives has been shown to be closer to a reconstructive explanation for behaviour than coherent introspection (Nisbett and Wilson 1977). In addition, phenomenologically trying to determine what one feels about another person from a mix of dislike, jealousy, irritation and admiration, often does not feel like direct perception. These challenges have been confirmed from within the developmental literature and Theory of Mind research. It has been found that far from passing 'own false belief' tasks before 'other's false belief' tasks, the two skills are achieved simultaneously in development. This supports the idea that we use the same method for reasoning about our own beliefs as we do for those of other people, neither being direct and unmediated.

Happé (2003) has suggested that as there is good evidence that the mechanism for reasoning about other's mental states is deficient in autism, then people with autism should also have deficits in their self-awareness. This has some experimental support. For example, in a 'Seeing leading to knowing' task, Kazak, Collis and Lewis found no superiority in 'own knowledge' over 'other knowledge' in the participants with autism (Kazak, Collis et al. 1997). Happé has suggested that a deficit in self-awareness would have implication for the monitoring of actions and maintenance of goal directed behaviours - areas in which people with autism have problems known as executive function deficits.

Other Approaches to Understanding Social Deficits in Autism

1. The Social Attribution Task

Heider and Simmel developed a silent cartoon animation in which geometric shapes moved around (Heider and Simmel 1944). They showed this cartoon to female college students and found that almost all the subjects gave social meaning to the movement of the shapes saying that they were chasing, hitting each other etc. The subjects also derived psychologically-based personality features from the shapes' movements such as 'that one is a bully'.

Klin used Heider and Simmel's animation in a sample of 20 subjects with autism, 20 with Asperger's syndrome and 20 typically developing adolescents and adults (Klin 2000). The subjects were asked to provide a description of what happened in the cartoon. The clinical groups both identified fewer social elements of the 'plot'. This task measures an inclination to 'psychologise' to use elements of folk psychology to explain behaviour even in non-human systems. Typically developing humans over-extend the application of folk psychology, such as the tendency to anthropomorphise members of the animal kingdom and computers. It may be that participants with autism lack this tendency due to their impaired ability to use metarepresentation and thus Theory of Mind. Alternatively, it may be that people with autism lack the initial drive to reflect about social motivation from a very early age, which in turn inhibits the development of metarepresentational abilities and a Theory of Mind. Further research on the development of precursors to Theory of Mind skills may resolve this.

2. Deficits of personal relatedness.

Hobson (1993) rejects the cognitive orientation of 'Theory of Mind' explanations of autism. Hobson has described the cardinal feature of autism as a deficient capacity for and experience of 'personal relatedness'. He states that the normal child directly perceives social signals and responds to them as such without the need to conceptualise. "What she requires is the capacities to register emotional meanings and to respond to those meanings as having reference to the world 'out there'." This has similarities to ecological psychology, where the input of top down processes is seen as minimal and complex information is conceived as being out there in the environment available for direct perception. The primary deficit would be a deficit in empathic emotional engagement with others: that is, a deficit in 'innately determined perceptual-affective sensibilities towards the bodily appearances and behaviours of others' (Hobson 1991).

Hobson has tested this theory through the measurement of the salience of emotional expression to people with autism. Weeks and Hobson conducted an experiment where 15 participants with autism and 15 controls performed a sorting task. They were required to sort pictures of people who varied in the following respects: sex, age, facial expression of emotion and type of hat. To establish a hierarchy of salience, they had to match 16 new people to two photos. All fifteen of the non-autistic children sorted by facial expression eventually, while only 6 out of 15 children with autism did so (Weeks and Hobson 1987).

Hobson, Ouston and Lee (1988) found that participants with autism performing a matching task with photos of faces performed just as well when the photos were presented upside down as they did when they were the right way up, unlike controls. They also performed a cross-modality matching task where subjects had to match voices

to faces by emotion. The control tasks were matching pictures of water e.g. a stream, with the sound it made or matching people running or walking with the sound they would make. Participants with autism were impaired only in the emotion face/voice matching condition relative to controls. However when the autistic and non-autistic subjects were matched on the BPVS test of verbal ability they were not significantly different in their performance on the emotion recognition tasks (Hobson, Ouston et al. 1988).

3. Emotion recognition

Other researchers have also examined whether the social deficits in autism are due to a difficulty in recognizing other's emotions. Gepner, Deruelle and Grynfeltt (2001) showed video clips of a person's face showing an emotion in three conditions, still, dynamic and strobe. They displayed non-emotional vowel sounds as a control condition. The children were then asked to match what they had seen to photographs. There were no significant differences in performance between emotional versus non-emotional conditions. However, slow dynamic presentation was found to facilitate facial expression recognition in children with autism (Gepner, Deruelle et al. 2001).

Boucher, Lewis and Collis (2000) matched children with autism to children with specific language impairment (SLI) based on language ability. On vocal affect naming and vocal facial affect matching tasks, children with autism were superior to those with SLI. A typically developing control group matched to the participants with autism on TROG[2] mental age were superior to the participants with autism on the affect matching task (Boucher, Lewis et al. 2000). Thus, there is no conclusive evidence that children with autism have a deficit in affect recognition that is independent of language deficits.

4. Empathy

[2] Test of Reception of Grammar (TROG) a test of language comprehension (Bishop 1983).

Empathy has two components, the affective and the cognitive. The cognitive aspect is closely related to Theory of Mind; this is also known as cold social cognition and involves reasoning about people's epistemic mental states, such as beliefs and knowledge. The affective component is an automatic response to another person's emotions, also known as hot social cognition. This is closer to the personal relatedness described by Hobson. Baron-Cohen and Wheelwright (2004) have designed a questionnaire measure for empathy known as the Empathy Quotient including 60 questions. They employed this measure with 90 adults with Asperger disorder or High Functioning autism and a matched group of 90 controls. The participants with autism had a significantly lower mean score than controls. They also employed the questionnaire on a sample of 71 normal males and 126 normal females. The males had a significantly lower mean score than females (Baron-Cohen and Wheelwright 2004). The authors suggest that autism can be characterised as a disorder of empathy.

5. Autism and gender

More boys than girls are diagnosed with autistic disorder. The ratio of boys to girls is higher at higher levels of functioning. The ratio of male: female is about 4:1 in the broader definition of PDD (Volkmar, Szatmari et al. 1993) : in the higher functioning cases, estimates of the male: female ratio go up to 10:1 (Lord, Schopler et al. 1982). Males are over-represented in the learning-disabled population as a whole. Gissler et al. (1999) in their follow up study of the Finnish 1987 birth cohort found that boys had a 43% higher cumulative incidence of intellectual disability (Gissler, Jarvelin et al. 1999). Murphy et al. (1988) found that the ratio of boys with learning disabilities to girls was 1.4:1(Murphy, Boyle et al. 1988).

To explain the further elevated incidence of autism in boys, Simon Baron-Cohen has put forward a theory that autism is an extreme form of the male brain (Baron-Cohen 2002). He argues that the male brain is inclined towards systematising, which means

working out the rules that govern formal, rule-bound, closed systems. Systematising involves the observation of the regularities of the inputs and outputs from a system and the generation of explanatory rules to predict these regularities. This is an inductive process where exceptions provoke a revision of the rule. The female brain is inclined to empathising. This involves the behaviours described earlier under Theory of Mind and social cognition. The theory is that autism is an extreme version of the male brain and that the empathising component is hypodeveloped.

In conclusion, there is strong evidence that people with autism have deficits in social cognition. This evidence is strongest for a deficit in understanding of other people's knowledge and beliefs as assessed by the false belief tasks.

The Neurobiology of Social Cognition

Gallagher and Frith (2003) have proposed a model of the mentalising system based on a review of imaging studies. It includes:

1. anterior paracingulate – key region for mentalising
2. superior temporal sulcus – intentionality and biological motion
3. temporal poles – personal semantic and episodic memory

They do not include the amygdala or the orbitofrontal cortex in the mentalising system but acknowledge that they may have a role in social cognition (Gallagher and Frith 2003).

FMRI and PET studies of subjects with autism during mentalising tasks have revealed reduced brain activation in the following areas (from (Di Martino and Castellanos 2003)):

1. ventromedial prefrontal cortex
2. temporoparietal junction (includes STS and STG)
3. amygdala

4. and periamygdaloid cortex

There is corresponding increase in activation in visual and auditory areas. Thus, there is evidence that people with autism show reduced activation during mentalising tasks in many of the brain areas that are hypothesised to perform social cognitive functions in the typically developing brain.

The amygdala is reported above as being less activated in participants with autism during mentalising tasks. There is also evidence from lesion studies that this structure is important for mentalising. Fine, Lumsden, and Blair (2001) report on a patient with early left amygdala damage who by adulthood had been diagnosed with both Asperger disorder and schizophrenia. He failed two second-order Theory of Mind tasks but was not impaired on executive function tasks. This is evidence for the necessity of the amygdala in the development of Theory of Mind abilities and/or the performance of Theory of Mind tests (Fine, Lumsden et al. 2001).

Stone et al. (2003) examined the performance of two subjects with acquired bilateral amygdala lesions on two advance tests of Theory of Mind, the Mind in the Eyes task and the Recognition of Faux Pas tasks. They were both significantly impaired on the Faux Pas task and one was significantly impaired on the Reading the Mind in the Eyes task indicating that the amygdala's role in Theory of Mind is not just developmental but is part of 'on-line' reasoning about mental states (Stone, Baron-Cohen et al. 2003).

Single cell recording in monkeys has shown cells that respond to certain types of motion. It has been speculated that these areas may subserve the more primitive social cognition seen in monkeys. The cells identified and their responses are:

1. Superior Temporal Sulcus (upper bank) – biological motion. Cells have been found in adjacent areas that respond to particular directions of eye gaze (Perrett, Hietanen et al. 1992). It also contains cells that respond to the sights and sounds generated by others but not by the self.

2. Superior Temporal Sulcus (lower bank) - observation of movement in goal-directed actions (Perrett, Harries et al. 1989).
3. Lateral inferior frontal regions - specific actions irrespective of the agent 'mirror cells' (Di Pellegrino, Fadiga et al. 1992).
4. Posterior anterior cingulate – cells activated before onset of self-initiated movement (Tsujimoto, Shimazu et al. 2006).

The mirror cells in particular have attracted a lot of attention as they respond to certain movements regardless of who is performing them. For example, a cell may respond to monkey seeing another monkey raise its right arm and the same cell will also respond to the monkey itself raising its right arm. Analogues of mirror neurons have been found in the human brain: mirror cells may be the basis of early social imitative behaviour seen in neonates (Meltzoff and Decety 2003). It has also been suggested that mirror cells or similar cells could be a method of internalising the social behaviour of others and then using a simulation method to solve social cognitive problems (Williams, Whiten et al. 2001). It is noted however, that monkeys in whom these cells have been identified most precisely display no imitative behaviour themselves but the cells may provide the basis for the evolution of this behaviour in their descendants. It has been proposed that mirror neurons provide the basis for the development of both language and advanced social cognition through their provision of an innately wired correspondence between self and other. The language connection is proposed, as it is known that the sight of someone's lips moving as one hears a sound, influences what is heard (the McGurk effect Munhall, Gribble et al. 1996). This is evidence of a role for motor representation in language comprehension. The site at which the mirror neurones have been identified in humans is very close to the language areas. The mirror neuron analogue in humans has been identified using transcranial magnetic stimulation (Fadiga, Fogassi et al. 1995). Observing someone moving their fingers selectively lowered the threshold for stimulation of these same distal muscles in the observer thus demonstrating an intimate connection between observing action and performing the same action oneself.

Face and emotion recognition in the brain

In the normal brain, there is a distinct brain region that has been implicated in the processing of faces, known as the fusiform face area. Schultz, Gauthier et al. (2000) found that during a face recognition task, subjects with autism showed significantly lower levels of activity in the fusiform gyrus and higher levels of activity in the inferior temporal gyri than controls. This was the case only in the face discrimination condition and not when subjects were asked to discriminate objects. Supporting this finding, Critchley et al. (2000) found that high functioning people with autistic disorder activated different brain areas compared to controls when consciously and unconsciously processing facial expressions. Again, the controls had significantly more activity in the right fusiform cortex during both conditions. The subjects with autistic disorder had greater activity than the controls in the left superior temporal gyrus and left peristriate visual cortex across all conditions. During unconscious processing of facial expression (determining gender), subjects with autistic disorder show less activity in the left cerebellum and amygdalohippocampal region. In the explicit processing condition, controls showed more activation in the left middle temporal gyrus. It has been suggested that these results may not be specific to face recognition and thus social cognition but may instead represent an effect of expertise. That is, people with autism may attend less to faces and thus develop less expertise in viewing them. The fusiform area may be an area for processing visual information the participant has had a high level of exposure to. Further research is needed to resolve this.

Wicker, Perrett, Baron-Cohen and Decety (2003) showed 10 normal subjects videos of the eye section of an actor's face in four conditions. The conditions were

1. direct gaze and emotion
2. averted gaze and emotion

3. direct gaze control
4. averted gaze control

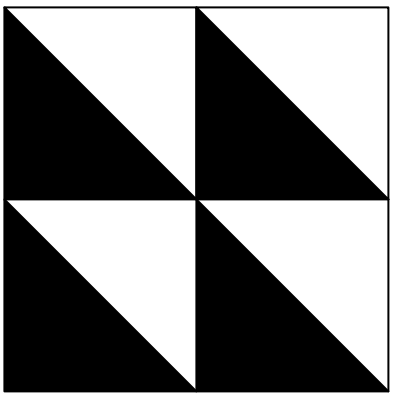
The subjects were asked to attribute hostile or friendly intent, or to judge gaze direction. The subjects performed these tasks during PET scanning. The results showed that activity in the anterior STG was specific to emotional expressions directed towards the self. Zibovicius and colleagues found hypoperfusion in this area in children with autism (Zibovicius, Barthelemy et al. 1998).

In summary, imaging studies have provided some support for functional differences between people with autism and typically developing controls in how they process social stimuli.

Weak Central Coherence

Weak Central Coherence is the tendency to process information in isolation from its context. The theory of Weak Central Coherence is based not on the deficits shown by those with autism but on their particular strengths. It has been shown that people with autism perform faster and more accurately than mental age matched controls on the embedded figures and block design tests. In the Embedded Figures Test (EFT) a simple shape, like a house or a tent shape has to be found in a more complex shape. Children with autism are more accurate on the EFT than developmentally delayed controls (Shah and Frith 1983; Mottron, Burack et al. 2003). People with High-Functioning autism and adults with Asperger's syndrome perform faster than other adults on the EFT (Jolliffe and Baron-Cohen 1997). However, Brian and Bryson (1996) found no difference between the performance of high functioning adolescents with autism/PDD and younger typically developing controls(Brian and Bryson 1996). This may have been due to the inclusion of subjects with PDD rather than a strict diagnosis of autism or Asperger disorder (these subjects were excluded for one analysis).

The block design test is a subtest of the standard intelligence test batteries. The block design test involves re-arranging patterned three-dimensional cubes to match a simple design.
For example:



Shah and Frith (1993) found that the performance of young typically developing and learning disabled children is enhanced by pre-segmenting the figures. The segmentation did not help the people with autism. The authors suggest that those with autism already perceive the design in a fragmented way explaining their superior performance.

Children with autism have been found to be less susceptible to visual illusions than controls (Happé 1996). However, Ropar and Mitchell (1999) did not replicate this finding. Ropar and Mitchell used a method whereby subjects had to adjust the size of the illusory elements on a computer screen. There were no significant differences between the clinical and control groups in their overall susceptibility to the illusions. In a second part of their experiment, where their method more closely resembled that used by Happé, there is evidence of a trend in their data.

Diagram 3 Percentage of subjects who succumbed to each illusion (subjects who failed the control task excluded. Ropar and Mitchell 1999).

	Muller-Lyer	Titchner	Ponzo	Hat
Autism	95	75	25	15
MLD	100	84.6	30.8	23.1

There is a trend for a smaller proportion of the participants with autism to succumb to the illusions than the participants with mild learning disability.
Weak Central Coherence has also been studied in the verbal domain. Frith and Snowling (1983) found that participants with autism were less able to use context to disambiguate the pronunciation of homographs than were controls - such as ‘To take the dog for a walk they needed to find his lead’ and ‘The sky was the colour of lead’.

The theory of Weak Central Coherence (Frith 2003) explains these findings by suggesting that people with autism are more inclined to process information in discrete units as opposed to integrating elements into a larger whole or Gestalt. Thus in the case of the visual illusions, they perceive the dimensions of the stimulus distinct from its

illusion generating context. In the case of the homograph pronunciation task, they do not process the meaning of the whole sentence and use this to disambiguate the homograph. Shah and Frith describe it as 'less capture by meaning'. Frith describes the drive for central coherence as being the drive for meaning, to draw together separate pieces of information to form a coherent whole. People with strong central coherence would be those able to see the 'whole picture' whereas those with Weak Central Coherence would be good at tasks requiring attention to detail such as data entry.

The theory of Weak Central Coherence has provoked a large volume of research to determine the nature and extent of this cognitive style. Jolliffe and Baron-Cohen (1999) examined local coherence in linguistic processing among young adults with autism. Local coherence is defined as within the capacity of short- term memory; that is up to three sentences. They found that participants with autism were less accurate and slower at identifying an appropriate bridging inference between two sentences. They also found that participants with autism were less able to use context to disambiguate an ambiguous sentence. Jolliffe and Baron-Cohen (2000) conducted experiments investigating whether subjects with autism could rearrange sentences to form coherent stories. In the temporal condition, the sentences could be arranged by temporal cues; in the other - the 'coherence' condition - the subjects had to arrange the sentences with reference to the meaning of the story. Subjects with autism were unimpaired in the temporal condition but made more errors and were slower than normal controls in the coherence condition. They also examined subject's ability to make global inferences from a story. Subjects with autism were found to be less able to make inferences about the meaning of the whole story than to answer questions about particular character's desires.

Mottron and Belleville (1993) studied a man with autism who had savant drawing abilities. Savant abilities are isolated talents found in individuals who are functioning at a lower than average level in other domains. The man, E.C., produced better drawings than trained draughtsmen who were used as comparisons. They found that this savant artist showed a bias towards local processing in his drawing method. He started with

small details and added to them rather than sketching out the overall shape first. He also showed a lack of global preference on hierarchical figures such as the Navon test. The Navon test is a large letter made up of small letters. The subjects are asked 'is there an M?' or 'is there an H?'

Most adults have interference from the global level on the local level on this test. That is, they are faster to detect the global H than the local M. There have been mixed results using the Navon test as a measure of global/local processing in autism (e.g.Mottron, Burack et al. 2003, found no effects) . This is because it is very sensitive to variations in test design -for example, factors such as duration of presentation and the exact instructions given to subjects influence results. Participants with autism perform in the same way as controls on this task when orientated to the global level by the task instructions.

As well as in the visual and verbal domains, Weak Central Coherence has been investigated in the auditory domain. Until recently no evidence had been found of auditory Weak Central Coherence. However Foxton et al. (2003) have reconsidered the nature of the local and global stimuli in the auditory domain. In previous studies (Heaton, Pring et al. 1999; Mottron, Peretz et al. 2000) local features were taken as the absolute pitch values and the global features as the pitch-direction change patterns. These studies found no impairment at the global level of perception, that is in perceiving pitch direction changes, for participants with autism. In the study by Foxton et al. the global level is reconceived as a combination of absolute pitch, pitch direction changes

and time values which combine to be more than the sum of the parts. They found that participants with autism experienced less interference from the combined effects of changes in local pitch and timing changes than controls.

Reduced generalisation

Plaisted has further investigated the Weak Central Coherence theory at the visual-perceptual level. She hypothesised that in a detection task, Weak Central Coherence would predict that participants with autism would be slower than controls when the target required integration of two features for its detection (a conjunctive target). For example, they would be slower to find a big red T among small red Ts and big green Ts than if the target differed from the distracters by only one feature. She found that the opposite was the case: in fact, the participants with autism were faster at this task than developmentally matched controls (Plaisted, O'Riordan et al. 1998a). This is interpreted as evidence against the Weak Central Coherence theory. Plaisted has suggested that the performance of participants with autism can be explained by reduced generalisation and enhanced feature perception, where the novel features of a stimulus are processed more fully than those held in common between stimuli. Plaisted tested this theory by examining the ability of participants with autism to extract categories using a prototype recognition task. Examples of two categories were presented until the subjects could reliably categorise them and then either another example or the prototypical example was presented. Findings in adult populations have shown that the prototype is easier to categorise in this situation. Participants with autism did not find the prototype easier to identify. Plaisted interprets this as evidence that the features held in common by stimuli are not processed as fully in autism. Plaisted views the operation of Weak Central Coherence effects to be at the perceptual level, because in autism perception of features is enhanced but this does not affect the integration of features at the representational level.

Many of the mixed results in the field of central coherence can be attributed to the fact that the level at which ‘central’ coherence acts is still to be delineated. This is

particularly clear in the case of auditory central coherence outlined above. Is there more than one level at which central coherence operates and if so, is it a unitary function? The most robust findings regarding Weak Central Coherence are

1. those from the block design and embedded figures tests and
2. those involving higher level linguistic processing (homograph test).

People with autism are able to dissect complex figures more naturally than developmentally matched controls and they have difficulty connecting together the meanings of words and phrases to create global meaning. Intuitively it would seem that these two phenomena differ in their level of operation. That is, the detection of shapes is perceptual and automatic, whereas the understanding of text is a higher level process that requires conscious awareness for its operation. Visual illusions and Gestalt phenomena show the influence of top down input on what is experienced as direct perception. These findings would be attributable to the operation of higher cortical areas imposing 'meaning' on lower level input. It may be that these top down influences are the same mechanisms that 'chunk' text into meaningful units. It could be argued that the operation of central coherence is a metarepresentational mechanism. That is, multiple units of new information are transformed into a representation of 'gist'. This mechanism is predisposed to using existing forms which connect associatively to other existing representations. 'Capture by meaning' occurs when suitable preexisting forms are identified, which are then substituted for the raw data. This has the benefit of economy but the raw data is lost. This explains why controls find recalling the gist of meaningful word strings easier while participants with autism retain the surface form possibly at the expense of meaning via associative connections. The Gestalt principles operate on the formation of these metarepresentations, which enables inferences to be made across incomplete examples of the same type.

A problem with this approach is that if it extends too far then the argument entails that children with autism should not be able to learn. All learning even at the most basic level requires some generalisation across circumstances and hence a representation of the gist of those situations that require a certain class of response. Children with autism

plainly are capable of learning. The limits of the operation of Weak Central Coherence still have to be defined.

Another issue is the operation of attention or even intent. There is some evidence that it is not the *ability* to process detail versus global qualities but the *tendency* that is altered in autism. That is, that people with autism can abstract meaning but their tendency is not to do so unless explicitly directed (Happé and Frith 2006).

We have defined coherence as a transformation of the raw data. It uses established forms to represent gist but also encompasses connections to other related information. For example in the block design test, information about angles, line length and colour contrast may be replaced with 'black triangle'. Individual pieces of information are put together in a way that changes their nature; it is not merely additive but transformative. From this perspective we would challenge the use of conjunctive targets as examples of coherence. The combination of the features of 'big T' and 'red' does not translate to global meaning. This is indeed conjunction rather than integration and the product is not more than the sum of the parts. Central coherence must add something new or rather be a substitution of a more global representation for the underlying raw data. The global representation has richer associative connections than the raw components.

This conceptualization of central coherence, based on Frith's account, would lead us to expect that the operation of central coherence would require a degree of mental flexibility and that this may be a constraint on the operation of central coherence in autism. That is, typical global central coherence may require the capacity to switch between alternative representations of a stimulus from constituent elements to unified whole.

In conclusion, there is strong evidence for people with autism having advantage on visuospatial tasks where focus on detail aids performance, such as the block design test, in the unsegmented condition. This can be explained by the theory of Weak Central

Coherence as an enhanced ability to resist substituting a global form for awareness of the constituent parts. This may be because switching or substituting between representations is more difficult for participants with autism due to lack of mental flexibility.

Evidence for Weak Central Coherence in autism from studies of the brain

Ring and colleagues examined the brain regions involved when participants with autism performed the Embedded Figures Tasks compared to those involved when controls performed the task. They found that participants with autism had less extensive task related activations. The participants with autism relied on the ventral occipitotemporal regions to complete the task whereas the control group had more activation in prefrontal areas. The authors suggest that the different pattern of activation suggests that the participants with autism are using a different strategy to complete the task, relying more on visual object recognition systems, whereas the controls rely more on spatial working memory (Ring, Baron-Cohen et al. 1999).

A methodological issue for imaging research based on psychological theory is that in order to show a brain behaviour relationship specific to a given deficit, it is necessary to have shown that some brain areas function normally in autism. It is not sufficient to show that, for example, faces are processed in a different brain area from typically developing controls, without showing that objects are processed in the same area as controls. This is the case, as when at rest people with autism show different brain states to controls, i.e. in the absence of a task there are differences between groups in brain activation (Kennedy, Redcay et al. 2006). If all brain areas show physiological abnormalities regardless of a relationship to the cognitive deficits in autism, then this does not elucidate the brain behavioural relationships involved.

Executive Function

Executive functions are those that govern flexible goal-directed behaviour. These are functions such as inhibiting pre-potent responses, set-shifting, planning and working memory. The executive functions were first identified through the pattern of deficits shown by patients with frontal lobe damage (Luria 1966). The functions have been defined through the tests on which frontal lobe patients exhibit specific performance deficits. These are tests such as the Wisconsin Card Sorting Test (set shifting and inhibition), the Tower of Hanoi (planning), and verbal fluency (generativity, inhibition).

It has been found that there are functional and anatomical dissociations between the components of executive function. It is possible that various aspects of executive function may be differentially impaired in different psychiatric disorders.
Children with autism have been found to be impaired in their:

1. Planning abilities - The Tower of Hanoi (Ozonoff, Pennington et al. 1991; Hughes, Russell et al. 1994; Ozonoff and McEvoy 1994; Ozonoff and Jensen 1999).
2. Inhibition of prepotent responses – 'detour reaching task' (Hughes and Russell 1993) not impaired on the Stroop task (Ozonoff and Jensen 1999), perseverate more than matched controls on the windows and detour reaching tasks.
3. Mental flexibility – Winconsin card sorting task (Shu, Lung et al. 2001)

Ozonoff and Strayer (2001) found that working memory is intact in autism. They suggest that previous findings of impairment in this area are due to the tasks used e.g. the Tower of Hanoi which may rely on competition between subgoals rather than working memory.
In her review of the literature, Hill (2004) concludes that the specific deficits in autism are in planning and mental flexibility.

Executive theories are about how behaviour is moderated in relation to higher order goals. This gives particular scope for executive theories of autism to explain the repetitive movements and restricted interests found in autism. Turner (1997) has investigated this connection. She has developed a taxonomy of repetitive behaviour (Turner 1995) that includes 11 types. She has also developed a Repetitive Behaviours Interview for use with carers that assesses the level of repetitive behaviour in each class. Using the interview with participants with autism and with a control group, she found that there were higher levels of repetitive behaviour in the participants with autism than the controls, at all levels of functioning.

Turner has examined whether certain types of executive function relate to certain classes of repetitive behaviour. She has found a degree of specificity between the executive deficit and type of repetitive behaviour reported. For these purposes, the eleven classes of behaviour were combined to four. She found that Recurrent (same response) Perseveration was related to repetitive movements and circumscribed interests. Stuck-in-set (same rule) Perseverations were related to repetitive use of language and circumscribed interests, and deficits in generativity (thinking of new things) were related to sameness behaviour and circumscribed interests.
Turner has also examined the relationship between repetitive behaviours and Theory of Mind competence: she found that 'passers' of Theory of Mind tasks were no different in their level of repetitive behaviour than 'failers' on Theory of Mind tasks. This was also the case for performance on tests of central coherence.

Russell suggests that the deficit in autism is one of intention monitoring. He believes that children with autism fail to monitor their own intentions due to their executive deficits. This was tested using an error correction task first used by Frith and Done (1989) in a study of schizophrenia. The task was presented on a computer screen: subjects had to respond to a target by firing at it. This response had to be fast but once fired the object moved slowly and there was time to correct errors before the missile reached its target. There were two conditions: one where the subjects could see the

missile's trajectory and the other where this could not be seen and the subject had to rely on their memory of which controls they had pressed. In people with schizophrenia, they found that participants had difficulty making internal corrections in the condition where they could not see the missile's progress but not in the other condition. Participants with autism in a slightly modified task used by Russell and Jarrold (1998) corrected significantly fewer errors under both conditions than controls. Also in Phillips et al's study of intention and outcome, they found that children with autism were less able than controls to remember their intended actions. Russell goes on to say that this action monitoring impairment has implications for the development of self- awareness and the verbal regulation of action, that would explain poor performance on tests of Theory of Mind without recourse to a specific social cognitive deficit. A later study of intention monitoring (Russell and Hill 2001) did not find a deficit in the participants with autism.

Studies of executive function in very young children with autism have not found evidence of a deficit. Griffith et al. (1999) found no executive function deficits in young children of 4 years old with autism. The tasks used have to be adapted for younger children. Tests such as the spatial reversal task were used. In this test, a penny is hidden behind one of two pots without the child seeing. The child then has to guess which pot the penny is behind. Unknown to the child the penny is placed behind the same pot each time. This continues until the child realises this and chooses the correct pot four times in a row. Critically the pot the penny is hidden behind then changes. This is designed to identify perseveration to the previously rewarded place. If the child keeps going back to the original pot over a number of attempts then this is an indicator of more immature executive functioning. Other tasks that are used in younger children are 'A not B' and object retrieval. It has been commented, for example Pennington et al (1997), that these tasks require further validation as tests of frontal or executive functioning, since they are based on the adult tests, tests on infants under a year old and tests used in animals and have not had extensive validation in children with frontal lobe damage. The frontal lobes are the last area of the cortex to mature, not reaching full maturity until

adolescence. It is possible that the immature frontal lobes do not subsume the same functions as in the adult case.

In summary, although there have been widespread findings of executive function deficits in autism, they have been inconsistent as to the nature of the deficit, i.e. flexibility, working memory, set shifting. The strongest evidence for executive deficits in autism is in the domains of flexibility and planning.

Executive function deficits are not specific to autism. Executive deficits have been found in many childhood disorders including ADHD, Tourette's syndrome, and conduct disorder. A general deficit in executive function would predict impairments through many areas of functioning rather than the specific patterns of strengths and weaknesses seen in autism. Ozonoff (1997) discusses the discriminant validity problem and reviews evidence for specific patterns of executive deficits across a number of disorders. She finds:

1. ADHD – deficits in inhibition but preserved flexibility
2. Schizophrenia –deficits in planning and inhibition but a substantial overlap with deficits found in autism and mood disorders.
3. Tourette's syndrome – inconsistent findings of executive deficits – any impairments maybe due to co-morbidity
4. Obsessive Compulsive Disorder –inconsistent findings of deficits in executive functions.

A difficulty in this area of research is that many of the commonly used tests of executive function measure a number of different abilities. For example, the Wisconsin Card Sorting test is a complex task. Recently, tests that are more specific have been devised that measure precise aspects of executive function. These have yet to be used extensively in autism research. Added to this, the lack of evidence for executive function

deficits in young children with autism undermines the case for executive function deficits being primary to the disorder.

Executive function and the brain

Damasio and Maurer (1978)proposed a link between autism and the mesolimbic cortex. They made this link based on neurological signs present in people with autism such as dystonias, dyskinesias, gait disturbance and other signs. This connection was strengthened by the evidence that people with autism are impaired on many of the same tasks that people with focal prefrontal cortex lesions were impaired upon. Robbins (1997) commented that it is a major strength of the mesolimbic theory that it can explain neurological signs and stereotypic behaviours. Stereotyped behaviours are frequently associated with malfunctions of the striatum, in particular from hyperactivation of the doperminergic projection. Thus, both executive deficits and restrictive and repetitive behaviours can be explained within one neural system.

Imaging studies have been conducted to attempt to localise the various executive functions in typically developing individuals. Fassbender et al. (2004) found that successful inhibition required the ventral prefrontal cortex, the left dorsal prefrontal cortex and the right inferior parietal cortex. The rostral anterior cingulate is involved in error detection and the pre-Supplementary Motor Area in conflict monitoring.

In the only study of brain activation during executive function tasks in autism Schmitz et al. (2006) found that successful completion of executive function tasks by participants with autism entailed higher levels of frontal brain activation than in controls. The functional differences in brain activation between participants with autism and controls were paralleled by anatomical differences at the same locations.

Conclusions

None of the above theories (Theory of Mind, Weak Central Coherence or executive function) alone has sufficient explanatory power to be a sufficient causal explanation for autistic disorder in all its various manifestations. Theory of Mind cannot fully explain the non-social aspects of autism particularly sensory hypersensitivity and 'insistence on sameness'. The Weak Central Coherence theory struggles to explain the lack of social motivation shown by young children with autism.
Executive function theories currently lack specificity to autism, executive problems being present in a number of disorders.

However, the Theory of Mind and Weak Central Coherence theory have sufficient evidence to be considered established cognitive characteristics of autism. The Theory of Mind deficit has been shown to be universal in that there has not been a case where a participant with autism has been able to pass a first order false belief task at the usual age. Theory of Mind deficits are specific in that although other disorders show Theory of Mind deficits it is not as such a profound level as found in autism; for example, people with schizophrenia show second order false belief difficulties not first order.

Weak Central Coherence cannot be argued to be universal in autism. It is estimated that about a quarter of people with autism show a peak of performance on tasks such as the block design or embedded figures task (Siegel, Minshew et al. 1996). On the homograph pronunciation task, a small proportion of people with autism make errors. However, it may be that these tasks are not subtle enough to tap the information processing bias in these people with autism. It is noteworthy that only one study has failed to find superior performance at a group level on visual disembedding tasks and this study included people with a diagnosis of PDD rather than autism (Brian and Bryson 1996).

These theories are not exclusive in nature. They may all coexist as part of the psychopathology of autism. It has been proposed, that these competencies, Weak Central Coherence, Theory of Mind and executive function, may interact in development. Deficits in Weak Central Coherence may produce a deficit in Theory of Mind or vice versa. However, research has shown that the abilities of participants with autism in these areas are independent. That is, participants with autism who pass false belief tasks may still show Weak Central Coherence (Happé 1994) . It may be that the tests are not matched on difficulty or are again not sensitive enough to detect the variation.

Ozonoff, Pennington and Rogers (1991)studied the relationship between executive function deficits and Theory of Mind in individuals with autism. They found that when full scale IQ score was partialled out in the participants with autism, executive function and Theory of Mind were significantly correlated. In the control group, these functions were not correlated. It may be that Theory of Mind ability, a constraint not present in the rest of the population may limit development of executive capacities in autism. They also found that executive deficits were more pervasive in the participants with autism than Theory of Mind deficits. This however may be due to the level at which these abilities were tested. The participants with autism may have had more subtle Theory of Mind deficits that were not detected by the first and second order Theory of Mind tests used.

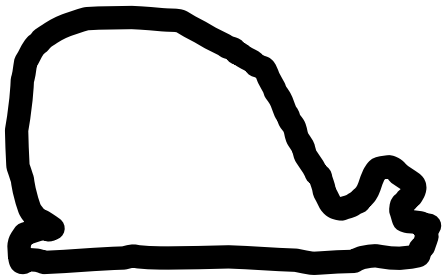
The link between joint attention, a precursor for Theory of Mind, and executive function in development has been investigated. It has been found that deficits in joint attention predict later mental flexibility for participants with autism but not for controls (Griffith, Pennington et al. 1999) . Joint attention abilities appear to be particularly related to executive function tasks that are thought to be subserved by the ventromedial prefrontal cortex such as delayed matching to sample (Dawson and al. 1998).

These two results both indicate that Theory of Mind skills constrain the development of executive function in people with autism. Alternatively, it may be that there is a third variable necessary for both Theory of Mind and executive function.

A tantalising possibility for psychological research in autism would be to find a task that would tap a cognitive mechanism that links performance in all three areas. That is, Theory of Mind, central coherence and executive function.

Ambiguous figures

Ambiguous figures are figures that can be perceived in two mutually exclusive ways without any alteration in their physical form. For example, the figure shown below can be seen as either a snail or a whale.



Most adults when looking at such a figure for any length of time, and when aware that it has more than one interpretation, find that it spontaneously reverses. That is, they see both interpretations of the figure sequentially and are not aware of consciously choosing to change perception of the figure but that this occurs both passively and irresistibly. Gopnik and Rosati (2001) in a study of normally developing 3 to 5 year old children found that 3 to 4 year old children almost never spontaneously reversed a figure during an inspection period even when they were informed of both interpretations of the figure. However, they found that older children of 5 did spontaneously reverse and that this was related to ability to pass a false belief test. Ropar, Mitchell and Ackroyd (2003) investigated the ability of children with autism to reverse ambiguous figures. They found that children with autism were able to perceive both versions of an ambiguous figure. Ropar, Mitchell and Ackroyd (2003) did not measure the number of reversals within a given time period.

Reversals of ambiguous figures exhibit the characteristic of 'sequential stochastic independence' (Taylor and Aldridge 1974) , i.e. periods of dominance by one interpretation are not related to the period of time it was previously suppressed. Reversal frequency can be influenced by the following variables (Leopold and Logothetis 1999):

1. intent – subjects can speed up or slow down reversal at will
2. semantic content – the meaningfulness of an interpretation will influence reversal rate.
3. gestalt properties of a stimulus
4. personality variables, intelligence and mood disorders
5. pharmacological agents such as caffeine or sodium amytal

Ambiguity is referred to in the central coherence literature in that children with autism are less able to use context to disambiguate verbal stimuli. In research on Theory of Mind, children with autism have also been shown to be less able to appreciate that people may interpret ambiguous stimuli differently to themselves (Droodle test Taylor 1988). It has also been found that the ability to understand that someone else could perceive the stimuli differently is related to the ability to perceive the two alternative forms of an ambiguous representation oneself. So the ability to hold two mutually exclusive interpretations of single stimuli is related to Theory of Mind competence. Shamay-Tsoory et al. (2004) found that cognitive flexibility in a sample of patients with prefrontal brain damage was correlated with cognitive empathy but not the affective dimension of empathy. They measured cognitive flexibility with the Wisconsin Card Sorting test, with the alternate uses test and design fluency tests.
The ability to see the two alternative forms of an ambiguous figure spontaneously reversing would seem also to be a test of cognitive flexibility, an executive function. This may be a link between the three main explanations.

Theory of Mind – the capacity to maintain multiple representations of the same reality and switch between them according to whose point of view you are considering.

Weak Central Coherence – the use of knowledge/context to select from mutually exclusive interpretations, but to hold both interpretations while this is taking place
Executive function – mental flexibility.
Ambiguous stimuli can be termed in each of these ways. The capacity to spontaneously reverse may index core switching function between representations. This switching mechanism may be 'sticky' in autism.

Ambiguous figures and the brain

There is very little research on the neurobiological basis of ambiguity. In research on perceptual ambiguity the focus has been on determining whether the instability in perception is triggered by low or high level brain areas. There is one theory that neural satiation is responsible for reversals. That is, it is a low level neural phenomenon that does not involve higher cognitive processes. Others think that as intent and other personality variables can affect reversals then higher level processing must be involved.

Perceptual ambiguity

In typically developing subjects, Lumer, Friston and Rees (1998) found that perceptual transitions during a period of binocular rivalry were related to activity in the fusiform gyrus. This activity was limited to the extrastriate not the striate cortex. They found that frontoparietal regions implicated in attention orienting were involved in transitions. This supports the role of higher-level attention mechanisms in instability. Kleinschmidt et al. (1998) also found increased activity in the fusiform area. They also found that the thalamus and striate cortex were deactivated during instability. A possible factor not mentioned in the discussion of these findings is that they both involved processing stimuli involving faces: other groups have found activity in the fusiform gyrus is correlated with the perception of the face in Rubin's face vase illusion e.g. Hasson et al. (2001). It may be that the brain areas specialised for faces are responsible for

maintaining these versions. Thus, brain areas involved in recognition as well as attention areas are implicated in perceptual reversals.
Ricci and Blundo (1990) found that patients with frontal lobe lesions were less able to perceive both versions of an ambiguous stimulus than patients with posterior lesions. All these results indicate that reversals of ambiguous figures are not dependent on low-level neural satiation but to higher level recognition and attention mechanisms.

Pettigrew and Miller (1998) have used binocular rivalry alternations as a proxy for inter-hemispheric switching. They found that alternations in percepts were significantly slower in subjects with bipolar disorder than in controls. The interest of this study is the use of an ambiguous stimulus as a marker for a clinical condition. Autism has also been linked to cerebral lateralisation (Happé, Brownell et al. 1999).

Conceptual ambiguity

Nomura et al. (2003)have examined the processing of ambiguously expressed emotion in humans. They presented subjects with faces that portrayed either ambiguous emotion (positive vs. negative) or ambiguous gender. Subjects had to make judgements about these faces while undergoing fMRI scanning. They found that the main effect of ambiguity was activation in the anterior cingulate and the left inferior frontal gyrus. The areas particularly involved in the processing of emotionally ambiguous stimuli were the anterior cingulate, the dorsal part of the medial frontal gyrus and the right inferior frontal gyrus. Structural equation modelling demonstrated functional connectivity between these areas.
Chan et al (2004) studied brain activation in response to lexically ambiguous versus concrete terms. Chan and colleagues found that words with a precise meaning in a word generation task activated the inferior prefrontal and mid-superior temporal areas. Lexically ambiguous words however activated the left dorsal-lateral frontal areas, the anterior cingulate, and the right inferior parietal lobe.

These studies suggest that conceptual ambiguity activates brain areas involved in response competition. These areas are close to those implicated social cognition and complex language processing.

In conclusion, there is some evidence from imaging studies that ambiguity activates specific brain areas and that the frontal cortex is implicated confirming that it is not a low-level neural phenomenon but involves attention and response competition mechanisms. There is no research on brain activation on viewing ambiguous stimuli in autism.

Conclusions

Cognitive psychology has produced strong evidence that in two areas of functioning, Theory of Mind (false belief task) and Weak Central Coherence (block design task) people with autism show consistent group differences compared to matched controls. The preceding discussion shows that neurobiological investigation is starting to produce interesting results about how these skills are manifest in the brain, at least in the area of mentalising skills, but these investigations are still in a very early stage. It is not yet possible to say whether any area of the brain is consistently hypoactive or hyperactive in autism, or to say whether this is causally related to behaviour. This leaves us with the cognitive tests as the most consistent markers of autism outside the behavioural diagnostic criteria.

There is considerable heterogeneity in the way people with autistic spectrum disorders may present: with or without language; with or without intellectual impairment; onset before or after three years; but nevertheless we are asked to accept that they have the same or closely related disorders. There is also heterogeneity in the level of impairment across the triad of impairments. For some people with ASD, the restricted interests may be the most prominent factor; they may have intense preoccupations that dominate their waking hours and are done to the exclusion of all else. These same people may be socially curious and attempt to initiate interaction but in an inappropriate manner. For others their main feature may be a complete lack of interest in other people; they may push past people who get in their way, and all interactions are purely instrumental. These people may have some stereotypic movements and a higher degree of intellectual impairment. Not only is there a high level of variability within the autistic spectrum but it also has blurred boundaries with many other disorders. It is also suggested that these traits are present in the whole population only differing by degree i.e. a continuum of ability. The supportive evidence for autism existing on a continuum of severity that

extends beyond current diagnostic boundaries would be if autistic traits were shown to have a common psychological basis independent of diagnostic status.
From these conclusions, we would argue that one of the best methods available to test the limits of the continuum concept of ASD is to examine how widespread these cognitive characteristics of autism are. The question we will address is:

> **'Do the cognitive characteristics of Theory of Mind deficits and Weak Central Coherence extend beyond people diagnosed with autism to be found in people with high levels of autistic traits who do not meet criteria for the disorder?'**

Given the evidence suggesting that ambiguous figures may represent a link between Theory of Mind and mental flexibility, we also wish to investigate the perception of ambiguous figures in relation to Theory of Mind and Weak Central Coherence.
To answer these questions, we required a suitable subject group. 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

At Edinburgh University Division of Psychiatry, there is currently running a research project known as the Edinburgh Co-morbidity Study. The full title is the Cognitive Impairment in Schizophrenia – a clinical imaging and genetic study of co-morbidity. The Co-morbidity study is funded by the MRC and the Principal Investigator is Prof. Eve C Johnstone.

Background to the Co-morbidity Study

The point prevalence for schizophrenia in the mildly learning disabled is 3% as opposed to 1% in the rest of the population (Turner 1989). There has been little investigation into the reason for the association between low IQ and schizophrenia. An exception to this is studies by Doody et al. (1998) and Sanderson et al. (1999) conducted at Edinburgh. The study by Sanderson et al. (1999) involved taking structural magnetic resonance images on age- and sex-matched patients with learning disability (n=20), schizophrenia (n=25) and both disorders (n=23). They found that the brains of the co-morbid patients more closely resembled the brains of the patients with schizophrenia than the patients with learning disability only. The authors suggest that the enhanced risk for schizophrenia among people with learning disability is because the depressed IQ is part of the schizophrenic illness. The co-morbid illness appears to be a more severe form of schizophrenia that is more highly heritable. There may therefore be people who are considered to have a learning disability who in fact have a treatable mental illness.

Another study, the Edinburgh High Risk study (Johnstone, Ebmeier et al. 2005), followed young people of normal IQ at elevated familial risk for schizophrenia over a period of ten years. At 18-month intervals, they were assessed on measures of general IQ, attention, motor speed, executive function, verbal learning and memory. They were also given MRI scans. They completed behavioural measures including the Child

Behaviour Checklist (Achenbach 1991) and the Structured Interview for Schizotypy (Kendler, Lieberman et al. 1989). It was found that baseline performance on certain of these measures could distinguish the high risk participants who became ill from those who did not (Miller, Byrne et al. 2002a; Miller, Byrne et al. 2002b; Johnstone, Ebmeier et al. 2005). It was also found that prior to becoming ill, participants showed a drop in IQ (Johnstone, Cosway et al. 2002; Johnstone, Lawrie et al. 2002) and also there were characteristic brain changes associated with the onset of illness (Lawrie, Whalley et al. 2002; Job, Whalley et al. 2005).

Method

The Edinburgh Co-morbidity study employs the variables that were found to be useful in predicting subsequent illness in the High Risk sample (Miller, Byrne et al. 2002). The participants in the Co-morbidity study are people who are at slightly enhanced risk of mental health problems due to intellectual disability. The study will determine whether among this sample there are young people whose learning needs are in fact due to the early stages of a severe form of schizophrenic illness. It will also determine whether the same variables as predicted schizophrenia in the Edinburgh High Risk study are useful in predicting who will become ill in this population.

The sampling frame was young people between he 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. Currently in Scotland, young people can receive additional learning support for the following reasons:

- learning environment: difficulty accessing the curriculum
- family circumstances
- disability and health
- social and emotional factors

It was therefore expected that the young people recruited to take part would be heterogeneous for type of learning problem, co-morbidity for psychiatric and developmental disorders and the range of social and emotional difficulties. Schools and

colleges were asked to approach young people who had an estimated IQ in the range of 50-80 points.

As discussed above in the section on diagnostic differentiation, there is some overlap between the symptoms of ASD and the negative symptoms of schizophrenia. It is also known that the risk for autism is increased in learning disability. Therefore, in addition to the measures that were found to be useful in determining liability to schizophrenic illness for the High Risk study, participants in the Co-morbidity study were also screened with the Social Communication Questionnaire (SCQ). The Social Communication Questionnaire (SCQ, previously known as the Autism Screening Questionnaire, (Berument, Rutter et al. 1999) is completed by the parent and reflects upon current behaviour and behaviours seen in the child between the ages of 4 and 5 years old. The SCQ is based on the Autism Diagnostic Interview algorithm and scores indicate whether the person is in the autism, PDD or non-autistic range. It is a 40 item scale. A score over 22 indicates the young person is probably in the autism category, a score between 15 and 22 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 PDDs from other diagnoses has a specificity of 0.75 and a sensitivity of 0.85. The SCQ is better at distinguishing PDD from non-PDD diagnoses than at distinguishing between autism and other PDDs (22 cut off autism vs. PDD specificity 0.6 and sensitivity 0.75).

The Co-morbidity study has four stages.

Stage 1 – completion of two questionnaires by parents about their son/daughter. The questionnaires are the Child Behaviour Checklist (CBCL) (Achenbach 1991) and the Social Communication Questionnaire. The parent completes these questionnaires with a researcher in their own home.

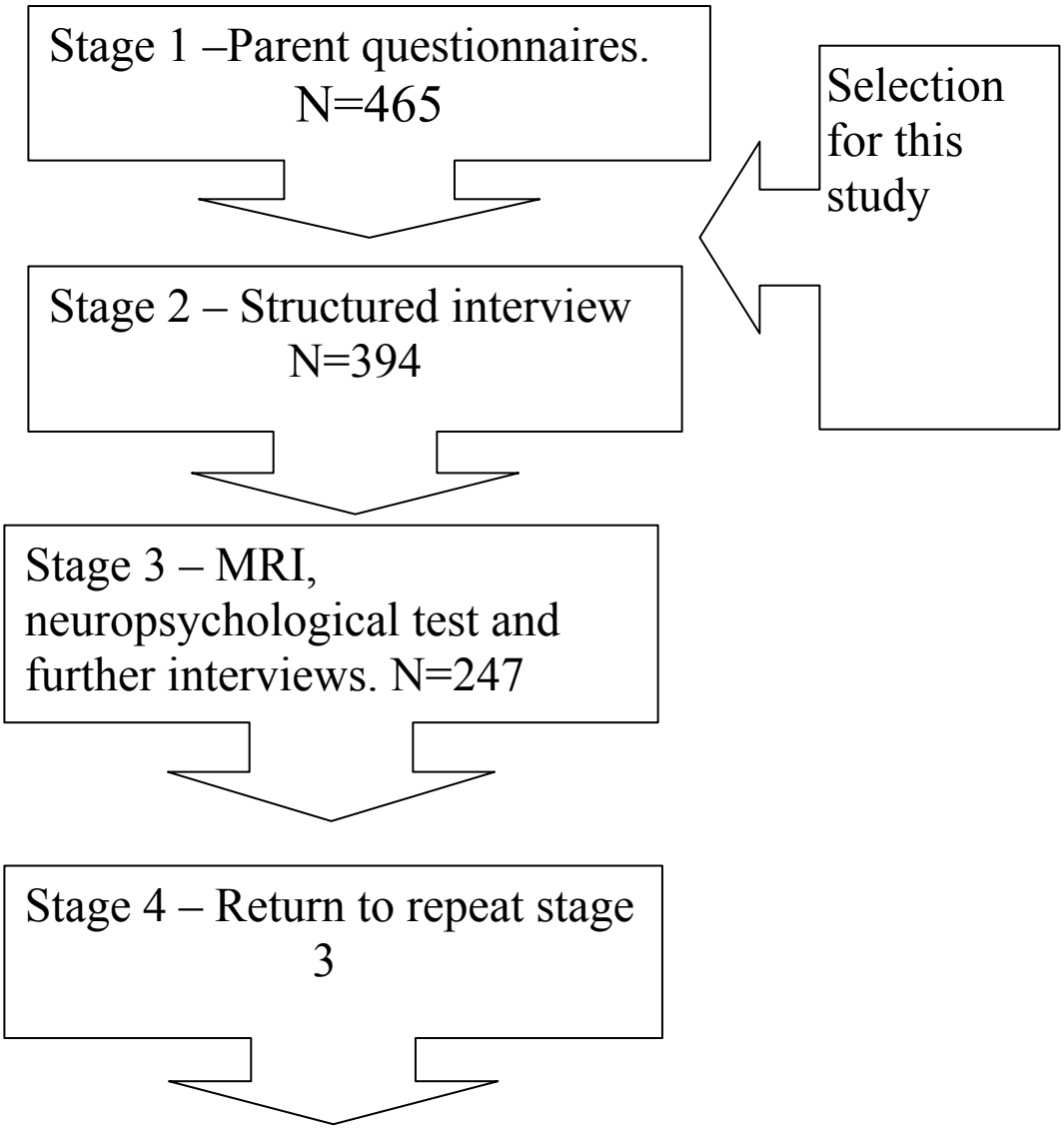
Stage 2 – Structured Interview for Schizotypy (SIS) (Kendler, Lieberman et al. 1989). Psychiatric Interview with young person at their school.

Based on their scores on the CBCL and the SIS, a subset of participants is selected for stage 3.

Stage three – Young person visits Royal Edinburgh Hospital. Full scale IQ, Rivermead behavioural memory test, Behavioural Assessment of Dysexecutive Syndrome assessments. MRI scan, dermatoglyphics and blood taken.

Stage four - One year later young person returns to repeat stage 3.

Diagram 4 Protocol for Edinburgh Co-morbidity Study



When the first 200 participants' carers had completed the SCQ, the findings were analysed and it was found that 37% of the participants were over the threshold for probable PDD.

This is considerably higher than most estimates of the prevalence of ASD in the general population. For example, Scott et al. (2002) found the prevalence of ASD to be 57 per 10 000 children aged 5-11 in Cambridgeshire. Breaking the prevalence down, Scott et al. found that in mainstream schools, the prevalence was 33/10 000 and in special schools 12.5%. Variation in prevalence estimates can result from differences in methods of case ascertainment, the age group studied, the inclusion of people with special educational needs and the range of conditions included (PDD vs. ASD). As can be seen from Scott et al.'s results, the prevalence is much higher is special schools.

The MRC review of autism research: epidemiology and causes (2001) concluded that the prevalence for autistic spectrum disorders is approximately 60, and narrowly defined autism 10-30, per 10000 children under 8. Prevalence in the adult population is not known. The rate of 37% PDD found in the Co-morbidity sample is far higher than this and is closer to the level usually found in severely intellectually impaired populations (Kraijer 1997; La Malfa et al. 2004). It is substantially higher than the 12.5% found in special schools (Scott et al, 2002). There are a number of possible reasons for this:

1. false positives – problem with the SCQ
2. carers over-reporting problems
3. a genuinely high level of unrecognised problems in this group. In a sample with mild intellectual disability plus a high level of co-morbid problems, this may substantially elevate the prevalence of ASD.

There has been speculation about whether there has been an increase in the prevalence of autistic spectrum disorders in the population at large. There have been reports of an 'iceberg' of undiagnosed cases of autism (Hansard debates, 2002). However, the increase in prevalence in routinely collected data over the last two decades can mainly be attributed to:

1. changing diagnostic boundaries and increased appreciation of the broader range of social-communication impairments that are contained within the autistic spectrum,
2. increased awareness among the lay public and professionals, leading to increased referral for assessment
3. increased specialist service provision leading to increased diagnosis of the condition.

The rate of 37% for PDDs in this sample was so much higher than expected that it merited further investigation. Were these participants really on the autistic spectrum? Due to the considerable overlap between autism and intellectual disability, other psychiatric disorders and certain genetic disorders, we would hypothesise that despite the increasing level of awareness among both parents and professionals about ASDs, there are still significant numbers of young people who, despite having high levels of autistic traits, are not diagnosed as autistic. This may be due to diagnostic overshadowing either by intellectual disability or by other psychiatric problems. Young people with social and emotional problems may also have autistic features but not meet criteria for diagnosis. It may be that although in this population there are a high number of young people who have sufficient autistic traits to have a positive result on the SCQ these difficulties are not truly autistic in nature and are better described by other diagnoses or none at all. Whether all autistic traits are in fact 'true' autism is a question about the assumption of an underlying continuum of autistic features. Are all autistic traits caused by a common psychological mechanism?

We know that the triad of impairments cluster together at a rate greater than would be likely by chance. This would indicate that the traits do not vary completely independently. This has received support from the study by Constantino et al (2004) who found evidence of 'a single, continuously distributed' underlying factor. The concept of a continuum has not yet been fully validated however.

Taking, for example the social deficits found in autism, it has been well established by Frith, Baron-Cohen, Leslie and others that people with autism have specific deficits in reasoning about other minds. These deficits are present in the face of average or even superior ability to reason about physical matters given developmental level. Using the fine cuts method, these cognitive deficits in autism have been isolated as a specific and universal delay in acquiring a Theory of Mind. (Other disorders e.g. schizophrenia have Theory of Mind deficits but not at the first order level). There is no doubt that in the typical population as a whole there is a gradation in social ability. Some people are more skilful in handling social situations than others; some are more interested in other people's mental lives than others. Social skill in a typical adult will be over-determined: determinants will include learned skills, emotional responsiveness, motivation and many others. Thus although there is undoubtedly a continuum of social ability, it is hard to ascertain whether there is a continuum in the established underlying cognitive constraint on social skill in autism, that is, in Theory of Mind ability.

Constantino and Todd (2000) have used the Social Reciprocity Scale to examine social abilities in relatives of people with ASD and the general population and have found evidence of both a continuum in the general population and genetic transmission of these traits. Although this measure has good psychometric properties and is thus well fitted to answer questions about the continuum of social ability, it does not specifically address the core deficit in autism as has been established, that is in Theory of Mind per se. What we seek to do here is to start to test whether the assumption of a continuum is legitimate. We are asking whether the cognitive deficit in Theory of Mind and the presence of Weak Central Coherence occurs as a function of autistic 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.

Kunihira et al. (2006) have examined performance of typical Japanese adults on Theory of Mind and central coherence tasks to see if they co-vary with scores on the Autistic Spectrum Quotient (AQ, Baron-Cohen, Wheelwright et al. 2001c). They found they did not. This may be due to the comparatively low level of autistic traits in this group and inadequate statistical power to detect small effect sizes.

We expected that among the young people participating in the Co-morbidity study we would find more people with a high level of autistic traits than in typical population samples but that our participants would be more heterogeneous for type of learning problems and the presence or absence of social and psychiatric problems. Some of these young people would have a diagnosis of an ASD but the majority would not. If those with very high SCQ scores really are part of the autistic spectrum one would expect them to show the characteristic cognitive traits associated with ASD, i.e. poor Theory of Mind and Weak Central Coherence. This study was devised in order to test whether this was the case.

Selection of measures

We know that in this sample, there will be a small number of young people with a diagnosis of an ASD but most will not. This makes it very important that we select tests that have a high level of evidence that people with autism perform significantly differently on them than people without autism. So that

1. a null result cannot be attributable to the tests not being sensitive to differences in cognition between people with and without autism
2. tests with large effect sizes are more likely to detect more subtle differences in people who have only autistic traits not the full disorder (if these exist).

The other consideration in selecting the tests is that they should be of appropriate difficulty for the participants, avoiding floor or ceiling effects. In addition to

consideration of general level of intellectual functioning, we know that a significant proportion of participants in the Co-morbidity study have other problems that may affect test performance, for example ADHD or dyslexia. Many have problems with concentration and motivation. The tests also had to be acceptable to the participants. For these reasons, we chose to measure Theory of Mind using the first and second order false belief tests. These tests were chosen because of the very high degree of evidence for people with autism being impaired on these measures. The appropriate level of difficulty was based on an estimate of the expected level of functioning of the participants. We already had data on the full scale IQ of participants in the Co-morbidity study but this did not allow direct prediction of what their Theory of Mind competence might be. Therefore, the tests were first used in a feasibility phase of the project in order to examine their suitability before extending their use to the full sample.

We chose to measure central coherence 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 receive.

In addition to these two core tests, which have a strong weight of evidence to support their use, we also used two tests that are more exploratory in nature. We used visual illusions as a second central coherence measure. The evidence for this as a suitable measure of central coherence is more mixed, but we have the advantage of being able to compare performance on the visual illusions with performance on the block design task. The selection of the visual illusions was also made because they involve simple perceptual judgments that should be quick to perform and acceptable to participants. We expected that performance on the visual illusions would be unrelated to IQ.
The other more exploratory test used was an ambiguous figure. We selected the cat/swan as used by Ropar, Mitchell, and Ackroyd (2003). This is a simple line drawing. As stated by Ropar et al. it has the advantage that under the two alternative interpretations, cat and swan, the head is at opposite ends of the figure. This can be used

to check that participants have really perceived the two identities of the stimulus by asking them to point to the head. We decided to take the number of spontaneous reversals seen in one minute as the dependent measure. As described earlier in the section, there is some evidence that being able to see reversals of the ambiguous figure is related to Theory of Mind competence, particularly second order Theory of Mind competence. Also reversals of the ambiguous figure may be a measure of cognitive flexibility. It was also expected that performance on this test would be unrelated to IQ.

It was also important to have some measure of our participant's IQ since if the groups are not well matched for intellectual ability, this might present an alternative explanation for any differences found between the groups' performance on the measures. We decided not to measure full scale IQ as this takes over one hour per participant. Instead, we used two subtests of the appropriate intelligence test (WISC or WAIS). 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.

In addition, for a majority of our subjects who have also completed stage three of the Co-morbidity project, 41 of our sixty subjects, we will be able to test how well the two subtests correlate with full scale IQ in this sample.

To conclude, the present study seeks to investigate autistic spectrum disorders in a sample of young people with additional learning support needs. These young people will not *necessarily* have IQs below 70 or have been diagnosed as having intellectual disability. However, the majority will be of below average IQ and it is expected that the range will be around 50-80 IQ points.

We aim to examine the relationship of autistic features to performance on tests of Theory of Mind, central coherence, ambiguous figures and IQ. The main research question is whether an undiagnosed non-clinic-derived population will show the same cognitive strengths and weaknesses in relation to their autistic traits as have been found in traditional autism research. That is, will people with high scores on the autism screening measure (SCQ≥15) also have more problems with Theory of Mind tasks? Will they also be unfacilitated by pre-segmentation on the block design task?
We will also examine the relationship between measures.

Specific Hypotheses

1. The participants with SCQ scores ≥15, the 'PDD' groups, will show superior performance on the Block Design test relative to the SCQ<15 'non-PDD'group.
2. The performance of the non-PDD group will be facilitated in the segmentated condition of the block design test. The 'PDD' groups' performance will not be facilitated by this manipulation.
3. A larger proportion of the 'PDD' groups will resist the visual illusions than the non-PDD group.
4. A larger proportion of the 'PDD' groups will fail the first and second order false belief tests than the non-PDD group.
5. The 'PDD' groups will report fewer spontaneous reversals of the ambiguous figures during a 60 second inspection period than the members of the other groups.

Method

This study was completed in two phases. The initial phase was a feasibility project which employed half the final sample (n=29). The feasibility project showed that the protocol was successful so a further 31 participants were recruited and tested in precisely the same manner. The method was unchanged between stages so the method and results are reported once for the complete sample (n=60).

Participants

All the participants for this study are drawn from those taking part in the Edinburgh Co-morbidity study, also known as Cognitive Impairment in Schizophrenia – a clinical imaging and genetic study of co-morbidity. The Co-morbidity study is funded by the Medical Research Council and the Principal Investigator is Prof. Eve C Johnstone. The young people participating in the Edinburgh Co-morbidity study have been drawn from the population of young people in Scotland with additional learning support needs. All the young people participating in the Co-morbidity study are between the ages of 13 and 22.

The young people taking part in the Co-morbidity study were approached through education services. The teachers at participating schools or colleges identified young people receiving additional learning support. The young people were given a letter inviting them to take part.
In total 465 young people and their parents or guardians gave informed consent.

The parents of the young people taking part in the Co-morbidity study were asked to complete two questionnaire measures about their child, the Social Communication Questionnaire (Berument, Rutter et al. 1999) and the Child Behaviour Checklist

(Achenbach 1991). The parents completed these questionnaires with a researcher in their own home.

Selection of participants for this study

Participants to be approached for this study were chosen based on their scores on the Social Communication Questionnaire completed by their parents as part of the Co-morbidity study described above. The Social and Communication Questionnaire (SCQ, previously known as the Autism Screening Questionnaire, (Berument et al. 1999) is completed by the parent and reflects upon current behaviour and behaviours seen in the child between the ages of 4 and 5 years old. The SCQ is based on the Autism Diagnostic Interview algorithm and scores indicate whether the person is in the autism, Pervasive Developmental Disorder (PDD) or non-autistic range. A score over 22 indicates the young person is probably in the autism category, a score between 15 and 22 indicates probable PDD and a score below 15 indicates that the participant is probably not on the autistic spectrum.

For this project 60 young people were identified by SCQ score. They were matched for age and sex.

Group 1: SCQ scores over 22 – autism n=20,

Group 2: SCQ scores 15-22 – PDD n=20,

Group 3: SCQ scores below 15 n=20.

Researchers on the Co-morbidity project performed the selection of participants. This was for two reasons

1. To preserve confidentiality until participants had given consent to take part in further research and

2. To ensure that the researcher conducting testing would be blind to the group membership of participants.

Participants in group 1 with the highest SCQ scores (≥22) were recruited first as this group comprised about 14% of the total Co-morbidity participants.

Approach to participants

Participants were approached to take part in this study by researchers on the Co-morbidity project with whom they were already familiar. If they expressed any interest in taking part then they were sent an information sheet and consent form. Once a completed consent form was returned, they were considered recruited to this study. Eighty-two letters were sent out to participants in the Co-morbidity study who had expressed willingness to consider taking part in further research. Of these 82, 62 young people returned completed consent forms. Two of these young people were then not available for testing, leaving 60 participants.

Participant characteristics

Each group contained 15 males and 5 females.
The mean age of the sample was 16 years and 6 months. The range was 13 years and 9 months to 22 years and 4 months.

Diagram 5 Participant Characteristics

	Mean (SD)		
	Group 1- autism	Group 2- PDD	Group 3- non-autistic
N	20	20	20
M:F	15:5	15:5	15:5
Age years; months	16; 6 (2; 0)	16;8 (1;10)	16;3 (1; 7)

Experimental tasks and stimuli.

Participants chose where they would prefer to be tested. For the majority this was in their home (50). A few preferred to be tested at their school (5) or at the university (4) or their place of work (1). Testing took between an hour to an hour and a half, depending on the needs of the participants. Participants were able to take a break if required.

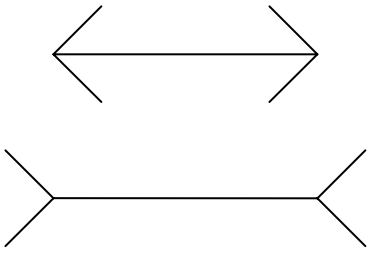
Every effort was made to ensure that the researcher was blind to which group each participant belonged. On two occasions however, the participant or their family inadvertently disclosed that the participant had a diagnosis of an ASD making membership of group 1 probable, but in the majority of cases the researcher was unaware to which group participants belonged.

The participants were asked to complete the following tests.

1. Ponzo, Hat and Muller-Lyer illusions

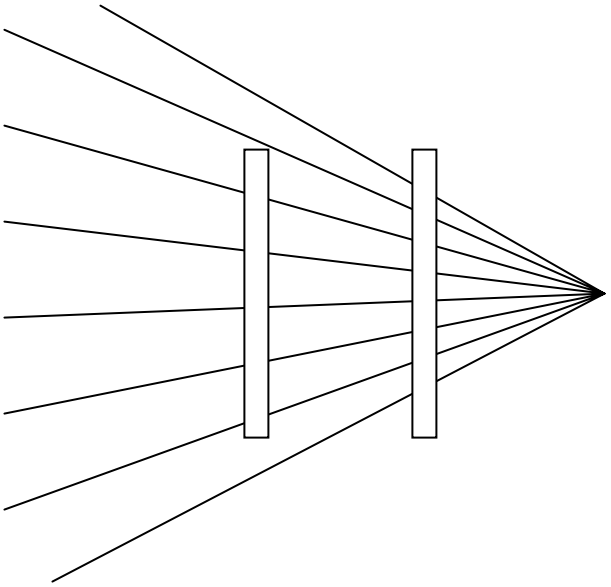
The illusions used were

a) the Muller-Lyer illusion:



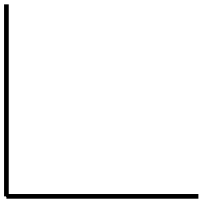
The inward pointing arrowheads at the end of the bottom horizontal line make it look longer than the upper horizontal line with the outward pointing arrowheads.

b) the Ponzo illusion



The lines give an illusion of perspective. The right hand block looks bigger than the left hand block as it is closer to the vanishing point.

c) and the Hat illusion



This is also known as the vertical-horizontal illusion. The vertical line appears longer than the horizontal line.

The illusions were presented on laminated paper. They were printed in black on a white background. The dimensions of the stimuli were

Ponzo: 10 cms by 8 cms

Muller Lyer: 17 cms by 12 cms

Hat: 6 cms by 6 cms.

For the Muller Lyer illusion (a) shown above, participants were asked whether they thought the horizontal lines were the same or different lengths. The researcher pointed out the critical lines so the participant could not misunderstand which lines were referred to. If the participants turned the paper or attempted to measure using their fingers they were asked to give their first impression based on what they saw when looking at the lines.

If the participant did not respond, they were asked 'Is one line bigger than the other or are they both the same size?' If the participant responded that they were different lengths then they were asked which one was longer. Once a response was recorded then the next stimuli was presented.

The illusions were all presented in the same manner.

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 tested on all three illusions and the control stimuli in a fixed order of presentation. This was in order that no participant saw the control before its illusion stimulus. The order of presentation was:

1. Hat control
2. Ponzo illusion
3. Hat illusion
4. Muller Lyer illusion
5. Ponzo control
6. Muller Lyer control

Hypothesis: Fewer group 1 & 2 (PDD, SCQ≥15) participants will succumb the illusions than group 3 (non-PDD).

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 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 cms. The stimulus was given to the participant to hold in their hand at a comfortable viewing distance. The procedure follows that used by Gopnik and Rosati (2001). The participant was asked what they thought the picture represented. If they said only a single interpretation (cat or swan) then they were asked if the picture could also be something else and asked to look at the figure again. If they did not see the other alternative (cat or swan) then the researcher pointed out the other version to them. If they still could not see both options then this was recorded and no further questions asked. If they could see both versions, the researcher explained that when viewing the figure some people find that the figure reverses, that is, it changes from looking like a swan to a cat or vice versa. The researcher also stated that other people do not see this happen. The participant was then asked to inspect the picture for 1 minute and tell the researcher if the figure reversed. That is, whether their perception of the figure changed from a swan to a cat or from a cat to a swan while they continuously viewed it. If there was no spontaneous response from the participant during inspection then the researcher prompted every 20 seconds to ask what the stimulus looked like to them now. The number of reversals reported in a 60s inspection period was recorded.
Hypothesis: Participants in groups 1 & 2 will see fewer reversals of the ambiguous figure than participants in group 3.

3. First order false belief test

The first order false belief task used was a location change task (Baron-Cohen, Leslie, and Frith, 1985). The participant was shown two model people and told their names, Sally and Tom. The researcher then acted out a short story using the two model people and two boxes. The story began with Sally and Tom playing with a ball. When it was time for Sally and Tom to go home, Sally put her ball away in a box. Then she went home. Once she had gone, Tom decided to play a trick on her. In her absence, Tom moved the ball to another hiding place. The next day Sally came back to get her ball. When the model Sally returned, the participant was asked where she would look for the ball, 'Where will Sally first look for her ball?' This is to test whether the participant will reason based on Sally's false belief in order to predict where the model Sally will look for the ball. If the participant gets this question correct then they are asked 'and where is the ball really?' and 'where was the ball to start with?' to check that they have understood the story and were attending to all of it.

Hypothesis: fewer participants in groups 1 & 2 will get the first order false belief question correct than in group 3.

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).

Two children, Sally and Tom are playing in a park. It is a hot day and there is an ice-cream van. The two children go over to buy an ice cream. Then Tom remembers that he has left his money at home. He is upset. The ice cream seller

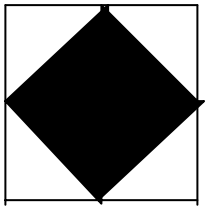
sees he is upset. She says, 'Don't worry; I am going to be here all day. When you go home for lunch you can get your money and get an ice cream this afternoon.' Tom cheers up and he and Sally go off and play in the park again. It is soon lunchtime and Tom goes home for his lunch. His model figure is placed into one of the houses. Sally stays in the park. The ice cream seller tells Sally that she has changed her mind. There is no one left in the park so she is going to drive her ice cream van round to the church to try to sell some ice cream there. Sally hears this but Tom does not because he is at home. Then Sally goes home. The ice cream seller drives her van round the village on the way to the church. On the way, she passes Tom's house as he is just coming out. The ice cream seller stops her van to speak to him. 'I have changed my mind about staying in the park' she says. 'I am going to drive over to the church. Come and see me there if you want an ice cream'. 'OK' says Tom, 'I will'. The participant is asked 'Does Sally know that Tom has spoken to the ice cream lady?' If the participant does not pass this question then the story is repeated and the question asked again.

Tom then goes out. The model of Tom is then taken out of sight of the participant. Sally is shown walking round to Tom's house. She knocks at the door. Tom's mother opens the door. Sally asks her if Tom is in. 'No you have just missed him' says his mother. 'He has gone out to buy an ice cream'. Then the participant is asked 'Where will Sally think that Tom has gone to buy an ice cream?' (second order false belief question). If they answer this question correctly, they are then asked 'why' (the justification question), 'and where has he really gone?' (reality question). This is to check that the participant has understood the story and is not responding with a guess or based on incorrect reasoning.

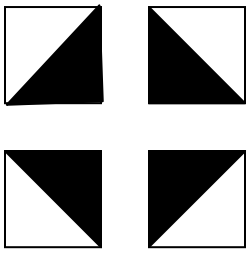
Hypothesis: Fewer participants in groups 1 & 2 will get the second order false belief question correct than in group 3.

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 the one shown below:



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 below:



There was no time limit. The time taken to complete each design was recorded.
Hypothesis: Participants in groups 1 & 2 will show a smaller difference between their times in the segmented and unsegmented conditions, that is, they will gain less benefit from the segmentation of the designs than participants in the other group.

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 1998).

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

Hypothesis: there will be no difference between the mean scaled scores of the three groups on these tests.

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 us to compare 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 – Young People

The total sample consisted of 60 young people. The participants were divided into groups based on their SCQ score. The groups were SCQ≥22 the 'autistic' group, SCQ 15-22- the 'PDD' group and SCQ<15 – the non-autistic group.

There were 15 females and 45 males in the sample. The groups were matched on gender: with a ratio of three males to one female in each group.

The mean age of the participants was 16 years and 6 months. The range was 13 years and 9 months to 22 years and 4 months.

Diagram 6 Participant characteristics

	Mean (SD)		
	Group 1- 'autism'	Group 2 – 'PDD'	Group 3 – non-autistic
N	20	20	20
M:F	15:5	15:5	15:5
Age (months)	198.3 (24.4)	200.7 (21.9)	195.1 (18.7)
SCQ range	23-40	15-22	0-14

The groups are very well matched on age. A one-way ANOVA gives F=0.332 p=0.72 showing there is no difference between groups in the mean age of participants.

Socio-Economic Status

Using the ACORN postcode classification method, we were able to classify all but one of our subjects. The table below shows how our participants fit into this classification system.

Diagram 7 Participants Socioeconomic Status by Group

ACORN classification	Group1 Autism	Group 2 PDD	Group 3 Non-autistic	Total	UK pop.
44-56 Hard pressed	11 55%	10 50%	13 65%	34 56%	22.4%
37-43 Moderate means	4 20%	5 25%	3 15%	12 20%	14.5%
24-36 Comfortably off	2 10%	1 5%	1 5%	4 6.67%	26.6%
13-23 Urban prosperity	2 10%	1 5%	2 10%	5 8.33%	10.7%
01-12 Wealthy achievers	1 5%	2 10%	1 5%	4 6.67%	26.6%
Missing	0	1 5%	0	1 1.67%	0.3%

The groups are well matched on socio-economic status. Kruskal-Wallis test of ACORN classification by group gives χ^2=0.88 df=2 Exact significance p=0.65. The relatively economically deprived neighbourhoods are over- represented in our sample compared to the total UK population.

IQ Matching Between Groups

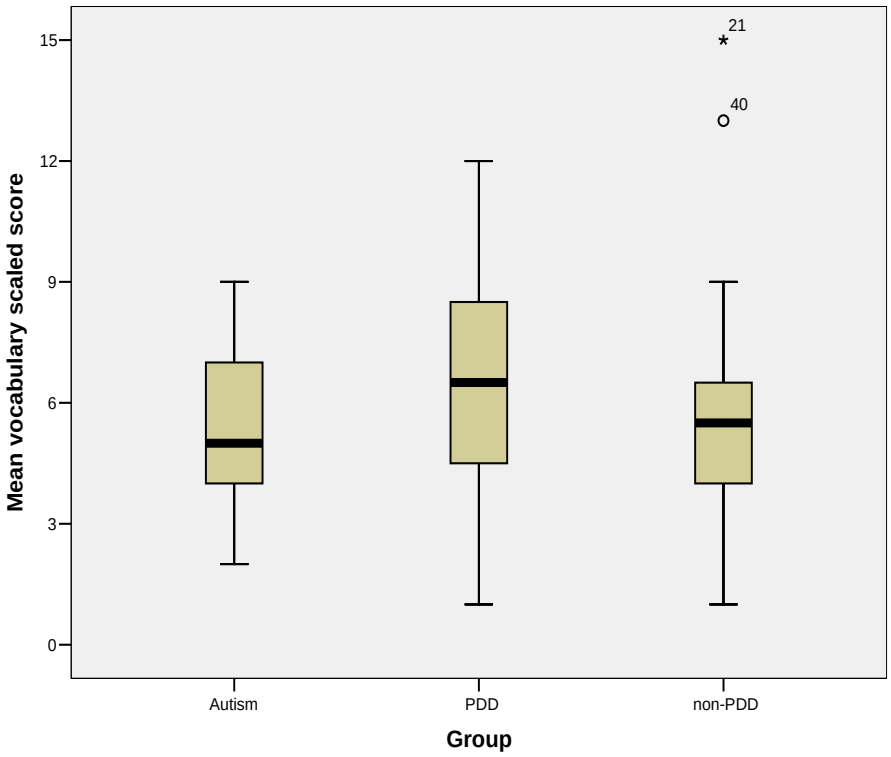
There are no significant differences between the groups on the two measures of IQ used, digit span and vocabulary scaled score. **In one-way ANOVA digit span scaled score factored by group F=0.75 p=0.48 and vocabulary scaled score factored by group F=0.96 p=0.39.** These high p values indicate there is no difference between groups on digit span scaled score or vocabulary scaled score.

Diagram 8 IQ subtest scores by group

	Mean (SD)		
	Group 1	Group 2	Group 3
N	20	20	20
Digit span scaled	5.70 (2.70)	6.60 (2.96)	6.15 (3.03)
Vocabulary scaled	5.20 (2.04)	6.50 (3.19)	5.80 (3.50)

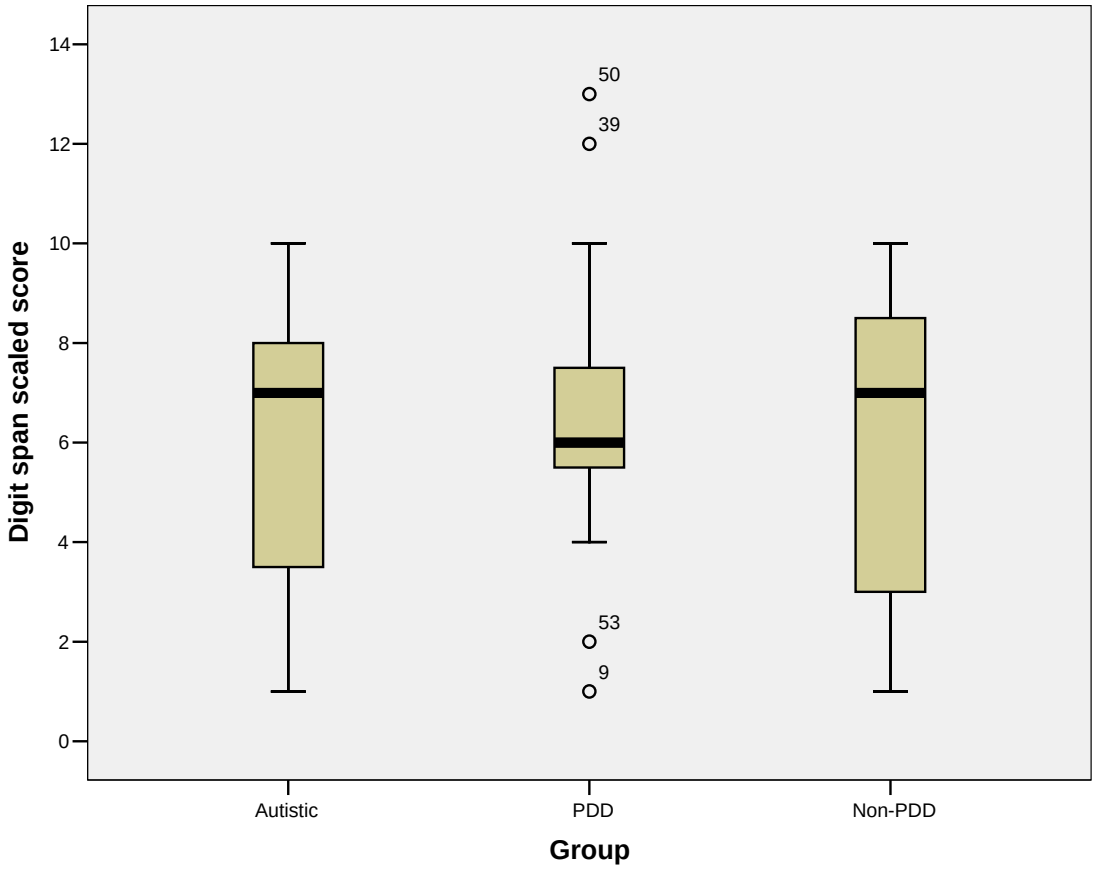
The vocabulary scores and digit span scores are reported as scaled scores, which have an approximate mean of 10 and a standard deviation of three. Our participants are on average one standard deviation below the general population mean on these measures.

Diagram 9 Box Plot of Vocabulary Scaled Score by Group



The box plot shows that the three groups are well matched for vocabulary scaled score. The non-PDD group has one outlier and one extreme value.

Diagram 10 Box Plot of Digit Span Scaled Score by Group



The box plot shows that the three groups are well matched for digit span scaled score. The PDD group has four outliers.

Comparison of the mean scaled scores on the digit span and vocabulary subtests of the WISC/WAIS between groups indicates that there are no significant differences between them in terms of intellectual functioning.

Forty-one of our participants have also taken part in stage three of the Edinburgh Co-morbidity study. For these participants we have their full scale IQ. It is therefore possible to check how good an estimate the subtest scaled scores used here are for the full scale IQ of participants. For vocabulary scaled score and full scale IQ Pearson's r=0.74, performance IQ r=0.48, verbal IQ r=0.81 n=40. For digit span scaled score and

full scale IQ Pearson's r=0.32, performance IQ r=0.25 verbal IQ r=0.31 n=41. It can be seen that vocabulary scaled score provides the best estimate for full scale IQ and verbal IQ in our participants.

The vocabulary scaled scores and full-scale IQ scores are highly correlated. We can assume that as the participants are well matched on vocabulary scaled score they are likely to be well matched on full scale IQ.

In summary, the three groups are very closely matched in terms of age, gender, socioeconomic status and IQ. This means that any differences in group performance on the target measures cannot be attributed to differences in age, social advantage or intellectual functioning between groups.

We will now examine the performance of the groups on the target measures of Theory of Mind, central coherence and cognitive flexibility. We hypothesise that the autistic and PDD groups will differ from the non-PDD group on these measures. Specifically that the non-PDD group will show better Theory of Mind, receive more advantage from the segmentation of the block designs, be more likely to see the visual illusions and more reversals of the ambiguous figure than the PDD groups.

Theory of Mind

The research evidence as outlined in the introduction supports the theory that people with autism show universal delay in acquiring Theory of Mind. Therefore, we hypothesise that the participants in groups 1 & 2 with high levels of autistic traits will be less likely to pass the false belief tests than the participants in group 3.

The table below shows the number of participants in each group who passed none, one or both of the Theory of Mind test.

Score of 0 - failed both first and second order Theory of Mind tests

Score of 1 – passed only first order

Score of 2 – passed first and second order

Diagram 11 Theory of Mind Score by Group

ToM score	Group 1 autistic	Group 2 PDD	Group 3 other	Total
0	3 (75%)	0 (0%)	1 (25%)	4 (100%)
1	7 (25.9%)	14 (51.9%)	6 (22.2%)	27 (100%)
2	10 (34.5%)	6 (20.7%)	13 (44.8%)	29 (100%)
Total	20	20	20	60

It can be seen from the table that most of the participants who failed all the Theory of Mind questions are in group 1 the autism group. Most of the participants who only passed the first order question are in group 2 and the majority who passed all questions are in group 3 the non-PDD group.

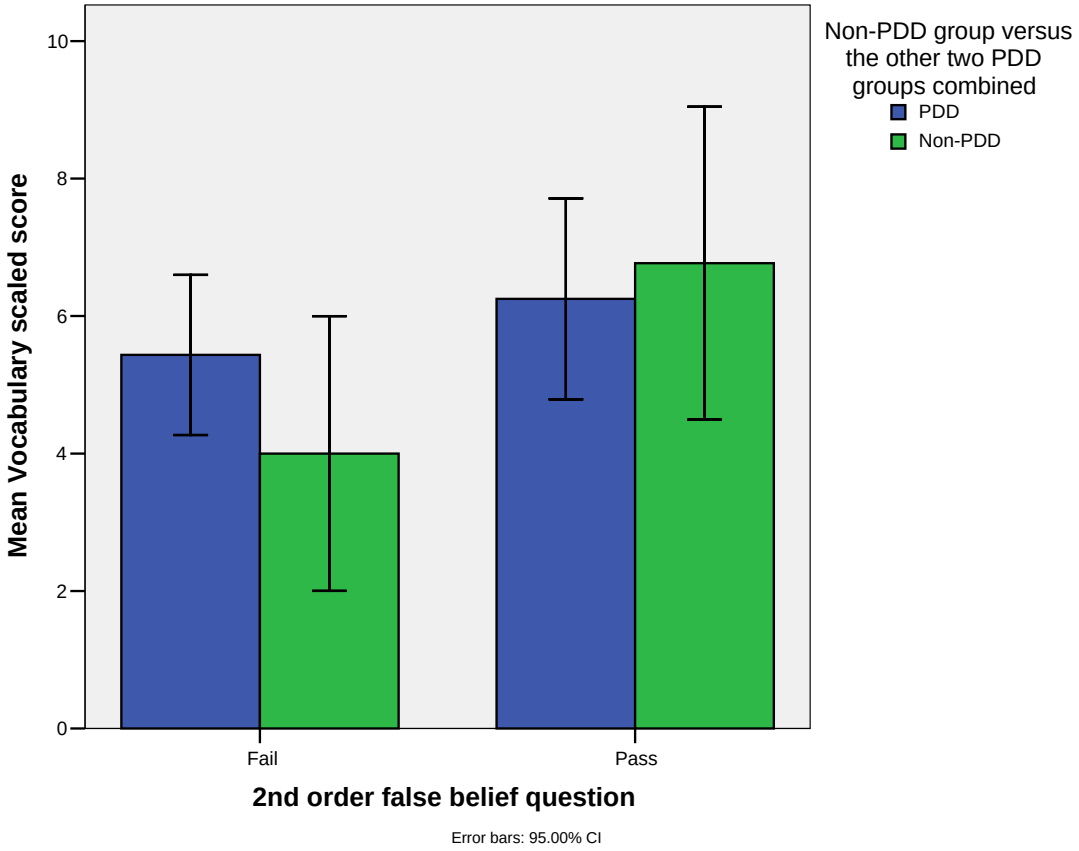
The Kruskal-Wallis test of Theory of Mind by group gives χ^2=3.44, df=2 p=0.18.

Given that our participants cover a range of ability levels it is possible that the 1st and 2nd order Theory of Mind tests are not a sensitive enough measure for the most able participants in groups 1&2. That is, the most able participants in the PDD and autism groups will pass both these tests using general verbal reasoning ability rather than intuitive Theory of Mind skills.

If this is the case in our sample, the participants who pass the 2nd order Theory of Mind test in the groups 1 & 2 (autism and PDD) will have greater verbal intelligence scores than those in the non-PDD group that pass.

To examine this, we plotted mean vocabulary scaled score for those who passed the 2nd order false belief task and those who failed. The participants were divided into 'PDD and autism' and 'non-PDD'.

Diagram 12 Mean vocabulary scaled score of those who pass and those who fail the 2nd order false belief test by SCQ status.



The graph shows the mean vocabulary scaled score of those who failed and those who passed the 2nd order false belief test. The graph shows that, generally, those who pass the false belief test have higher vocabulary scores than those who fail. However, this is much more pronounced for the non-PDD group. In the non-PDD group, those who fail the false belief task have a vocabulary score of around 4 and those who pass over 6. In the PDD and autism groups combined, the mean vocabulary score is around is 6 for those who pass and about 5.5 for those who fail. This indicates that it is not that the 'PDD and autism' participants have higher vocabulary scores to pass the false belief test, those who pass the false belief task are about the same as those in the non-PDD group. The distinction is between those that fail. It is more likely that a person in the PDD and

autism group with a high vocabulary score will fail the false belief test than a person with high IQ in the non-PDD group. In subjects with vocabulary-scaled scores of 6 or more, in the non-PDD group 0 out of 5 fail the false belief question and in the PDD and autism group 9 out of 16 fail. This difference in proportions is significant - Fisher's Exact test (2 sided) p=0.045.

To test whether this was a specific effect of verbal ability or general intelligence, we compared the average time to complete an unsegmented design in the block design task among those who fail the false belief task. In the non-PDD group, those who fail the false belief task have an average time of 56.27s. Those in the autism and PDD groups who fail the false belief task, have a much lower average time of 28.34s, they are much better at the block design test. This difference is statistically significant using an independent samples t test, t_{29}=-2.31, p=0.028. Among those who pass the false belief test there is no difference between 'autism and PDD' and 'non-PDD' participants. This supports the conclusion that in the autism and PDD group participants are more likely to fail the second order false belief question despite a relatively high IQ.

Two subjects from group 2, the PDD group, failed the justification question for the second order false belief test. However, these two participants had low vocabulary scaled scores and therefore their exclusion does not affect the outcome of the comparisons based on IQ.

Block design test

It has been found that people with autism have an islet of ability on the block design subtest of the WISC/WAIS. Shah and Frith (1983) found that this was due to an enhanced ability to perceive the design in a fragmented way. Control groups find constructing pre-segmented designs much easier than constructing whole designs, but the performance of people with autism is much the same on segmented and unsegmented designs. This supports the conclusion that autistic participants already have access to

fragmented perception of the designs due to the inherent nature of their perceptual system.

We therefore hypothesised that the participants with high levels of autistic traits (in groups 1& 2) would perform well on the unsegmented version of the block designs and their performance would improve only a little when given the segmented version of the block designs. However, the participants in group 3 would be more facilitated by the segmented condition.

We found that over half of the participants were unable to complete all of the eight designs in the unsegmented condition of the block design test. The table below shows the number of designs completed by the participants.

Diagram 13 Number of unsegmented designs completed by group

		Group			Total
		1-Autism	2-PDD	3-non-autistic	
No. of unsegmented block designs completed Total=8	1	1	0	4	5
	2	1	2	0	3
	3	1	1	0	2
	5	1	2	0	3
	6	2	3	4	9
	7	5	5	4	14
	8	9	7	8	24
Total		20	20	20	60

This means that it cannot be suggested that the autism or PDD groups are at ceiling on this task. This has been put forward as an explanation for a lack of facilitation in the segmented condition for people with autism. That is, that they are already performing at an optimal level so their performance cannot be improved by the segmenting of the design. It is clear that this is not the case for our participants.

As can be seen from the table above, more than half of members of each group did not complete all the designs. The number of designs completed did not differ between groups. Kruskal-Wallis test χ^2=0.46 df=2 Exact significance p=0.80.
A small number of participants were also not able to complete all the designs in the segmented condition. The number of participants who completed all nine designs in each group is shown in the table below.

Diagram 14 Number of segmented designs completed by group.

	Group		
	1-autistic	2-PDD	3-non-autistic
Did not complete all	3	2	5
Completed all designs	17	18	15
	20	20	20

The number of segmented designs completed also did not differ between groups. Kruskal-Wallis test χ^2=1.72 df=2 Exact significance p=0.42.
This is evidence against universal weak central coherence in the PDD participants. In all groups more designs were completed under the segmented than the unsegmented condition. Participants in both the PDD and non-PDD groups were facilitated by the segmentation of the designs and most of the participants completed all the designs in the segmented condition.

The difference in average time to complete a design between the segmented and unsegmented conditions of the block design test (BDDIFF) was calculated. This calculation only included the designs participants were able to complete successfully.

Diagram 15 Difference in mean time to complete a design between segmented and unsegmented condition (BDDIFF) by group.

Group	Mean BDDIFF (seconds)	Std deviation (seconds)
1 autism	11.93	10.34
2 PDD	17.14	22.72
3 non-autistic	21.63	22.45

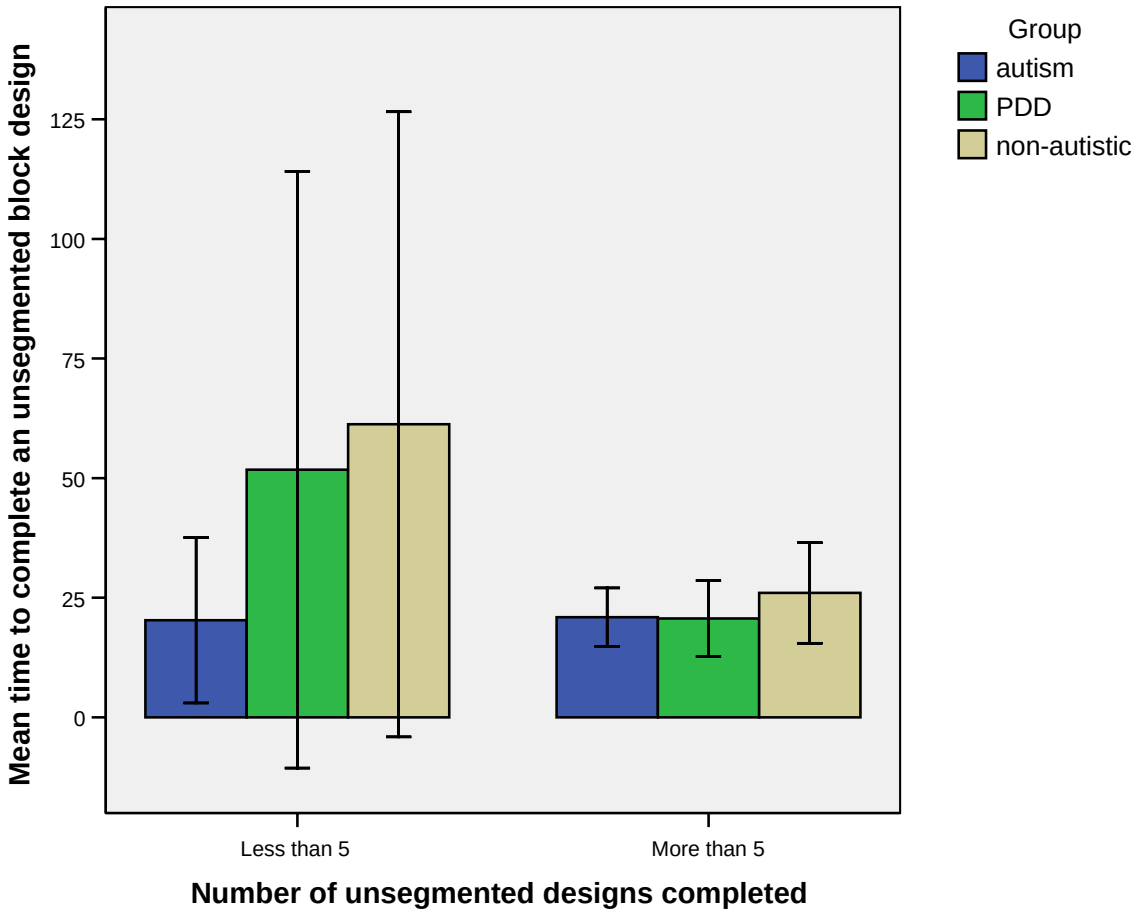
It is clear from the table that participants in group 1 gained less from the segmentation of the designs than participants in the other two groups. A one-way ANOVA shows the mean difference between the time taken to complete a design in the unsegmented and the segmented conditions was not statistically different for the three groups. When the autism group is compared to the other two groups combined, an independent samples t test gives a t_{58}= 1.76, **p=0.083**, which approaches significance. The means clearly differ between group1 and the other two groups but the large variances in the scores of groups 2 and 3 mean this does not reach statistical significance.

The difference between the autism group and the other two groups combined in average time taken to complete a design in the unsegmented condition also approaches significance. In an independent samples t test t_{56}=-1.9, p=0.059.

On the designs they were able to complete, the participants in group 1 were on average faster than the participants in the other two groups. However, about half of the participants in group 1 were unable to complete all the unsegmented designs as were participants in groups 2 and 3. Does this mean that there is a subset of participants in group 1 with exceptional performance who were responsible for the lower BDDIFF in this group? This subset with weak central coherence who are extremely good at the block design test may find doing all the designs very easy and cannot be facilitated by segmentation as they are already at ceiling and cannot do any better. This exceptional group may be the source of the difference in mean times between the PDD and non-PDD groups. To clarify whether this is the case, we will plot the average time to complete a

design in the unsegmented condition by group. Participants are divided into those who made more or less than five designs so that we can see whether the differences are more pronounced for those who were good at this task compared to those who were not.

Diagram 16 Mean times to complete a design by those completing less than or more than five designs.



The error bars show the 95% confidence interval.

In contradiction to what was expected, this graph shows that among participants who completed less than five designs, the autism (SCQ≥22) group were much faster than the other participants were. Among the subjects that could complete all the designs, there is very little difference between groups. For subjects in groups 2 and 3 (SCQ<22), the mean time taken to complete a design in the unsegmented condition is significantly

different for those completing less than 5 designs compared to those who complete more than five designs. Independent samples t test t_{38}=3.4 p=0.002. For the autism group this is not the case t_{18}=-0.099, p=0.92. The difference is not due to a small group of participants in group 1 who have exceptional visuospatial skills and thus complete all the designs. The differences between groups are even *greater* for the *less* able participants. It should be noted however that the absolute numbers in each group who completed less than five designs was small, about three or four per group. There was a strong trend across all levels of ability for the group 1 'autism' participants to be faster but this was enhanced in the group comparisons by the very large differences in the participants unable to complete five designs.
Possible explanations for these results are outlined in the discussion.

Visual illusions

Our hypothesis is that subjects in the PDD groups 1 & 2 will be more resistant to the visual illusions than the subjects in the non-PDD group. The number of participants in each group who succumbed to the Hat illusion is shown in the table below.

Diagram 17 Number of participants succumbing to the Hat illusion

Hat illusion	Group 1 autistic		Group 2 PDD		Group 3 non-autistic	
	All	Excluded	**All**	Excluded	**All**	Excluded
Not seen	**6**	3	**8**	6	**10**	7
Illusion seen	**14**	7	**12**	8	**10**	7
Total	**20**	10	**20**	14	**20**	14

The table shows the number of participants in each group who saw the illusion. This is shown with all participants included (All) and with the subjects who failed the Hat control task excluded (Excluded). The difference between groups in the numbers of subjects succumbing to the illusion is **not significant**. In fact, the trend is in the opposite direction to that hypothesised. The result remains non-significant whether the participants who failed the control task are included or excluded from the analysis.

It is notable that a large proportion of subjects (40%) did not see the illusion across all groups possibly suggesting that the illusory effect was relatively weak. In addition, a large proportion of participants failed the control task for the illusion.

The number of participants in each group who succumbed to the Muller-Lyer illusion is shown in the table below.

Diagram 18 Number of participants succumbing to the Muller Lyer Illusion

M-L illusion	Group 1 autistic		Group 2 PDD		Group 3 non-autistic	
	All	Excluded	**All**	Excluded	**All**	Excluded
Not seen	**5**	5	**5**	4	**2**	2
Illusion seen	**15**	11	**15**	11	**18**	14
Total	**20**	16	**20**	15	**20**	16

The difference between groups in the numbers of subjects succumbing to the illusion is **not significant**. The trend is however in the direction hypothesised. 80% of participants saw this illusion suggesting it was relatively persuasive. The results are non-significant whether the participants who failed the control task are included or excluded.

The number of participants in each group who succumbed to the Ponzo illusion is shown in the table below.

Diagram 19 Number of participants succumbing to the Ponzo illusion

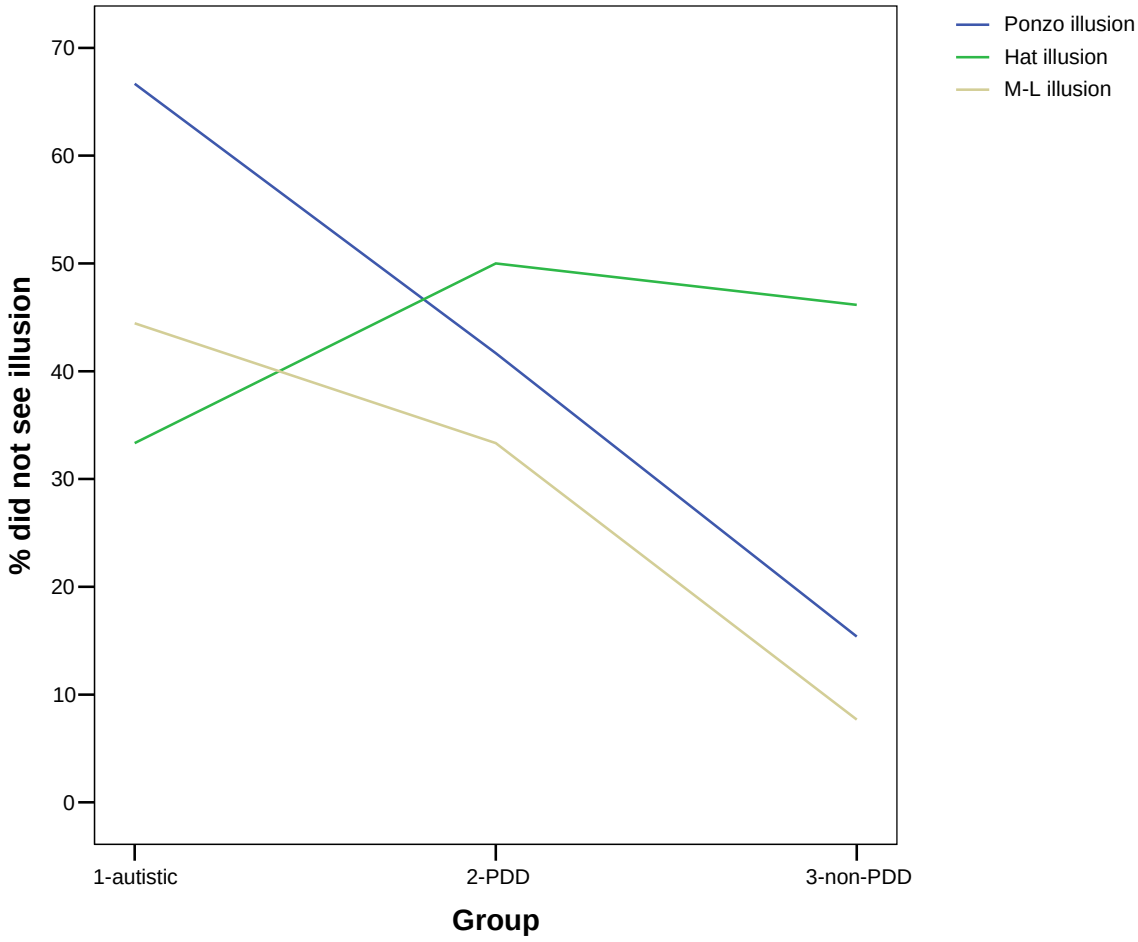
Ponzo illusion	Group 1 autistic		Group 2 PDD		Group 3 non-autistic	
	All	Excluded	All	Excluded	All	Excluded
Not seen	8	8	6	6	2	2
Illusion seen	12	10	14	10	18	17
Total	20	18	20	16	20	19

73% of participants saw this illusion.

Seven participants failed the control test for the Ponzo illusion. When these participants are removed from the analysis and the non-autistic group compared to the PDD groups combined, in a cross tabulation the Fisher's Exact test, two tailed, gives a significance of p=0.029. When the subjects that failed the control task are included then Fisher's Exact test gives a two tailed significance of p=0.062.

The graph below shows the percentage of subjects from each group who saw each of the illusions when the subjects who failed any of the control tasks are excluded (n=34).

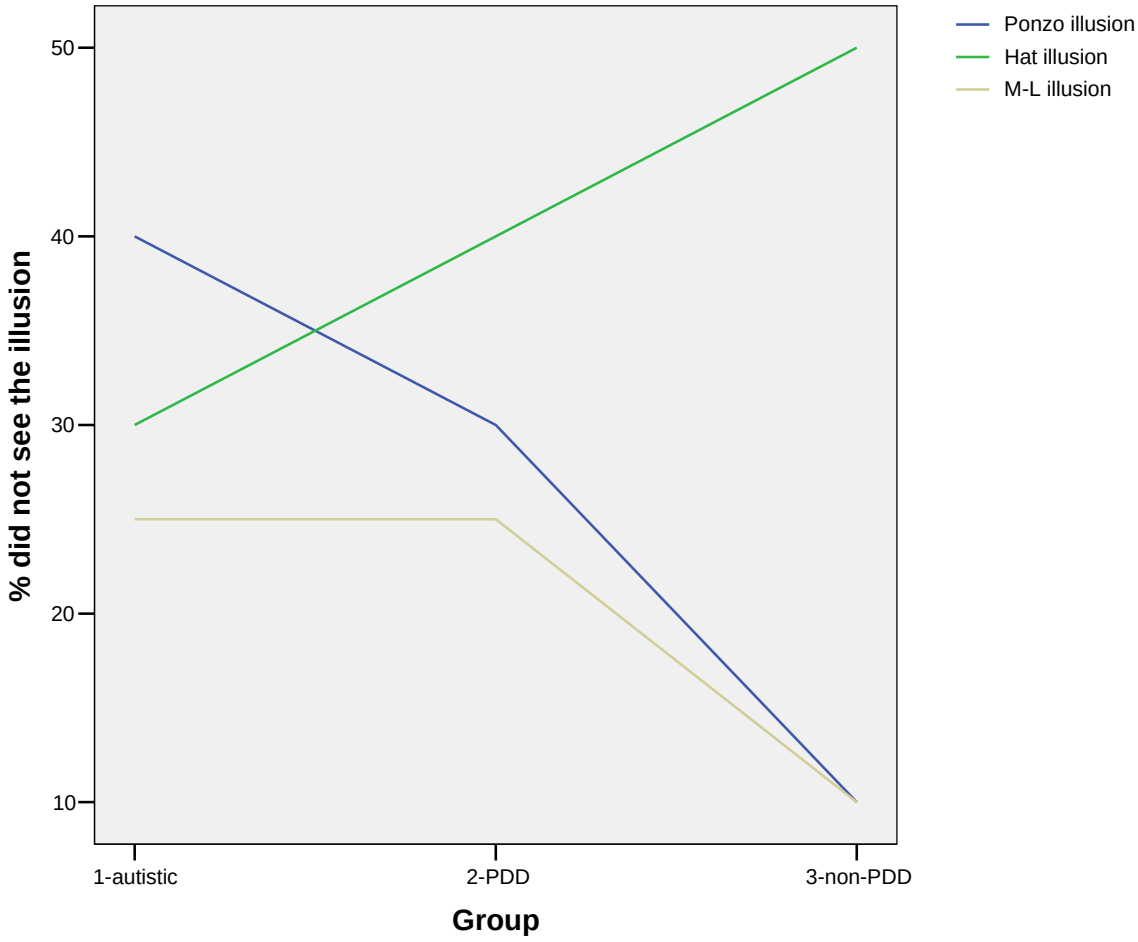
Diagram 20 Percentage of participants succumbing to the illusions by group (with participants who failed the control tasks excluded) illustrating trend in the data.



The graph shows that in group 3 non-PDD a very small percentage did not see the Ponzo and Muller Lyer illusions. Percentage seeing the Hat illusion is largely unaffected by group.

The graph below shows the percentage of subjects from each group seeing the illusions with all subjects included.

Diagram 21 Percentage of participants succumbing to the illusions by group



It can be seen from the graph that the pattern of results is very similar for the Muller Lyer and Ponzo illusions. There is a strong trend for a lower proportion of participants with high SCQs to succumb to these illusions than low SCQs, as hypothesised. The trend for the Hat illusion is in the opposite direction.

Ambiguous figures

We hypothesise that participants in group 1 will be less likely to see reversals of the ambiguous figure. That is, they will be less likely to see the figure switch between a cat and a swan under continuous viewing.

Version seen initially

The spontaneous response of participants when first asked what they thought the ambiguous figure represented was recorded. 30 of the subjects saw the cat initially, nine a mouse/rat, ten a swan and seven both versions, one said elephant, one dinosaur, one Loch Ness monster and one did not know.

There was no difference between the groups in the number of participants able to see both versions of the ambiguous figure spontaneously. Seven such participants said that the figure was a cat and a swan: one from group 1 four from group 2 and two from group 3.

Three participants gave highly idiosyncratic responses (Loch Ness monster, dinosaur and elephant) these were two participants from group 3 and one from group 1.

Both versions of the figure

When informed of the ambiguous nature of the stimulus, that it could have two distinct interpretations, eight subjects failed to see both versions of the ambiguous figure. This was after the researcher indicated the two different versions. Five of these participants were from group 2 and three from group 3.

Reversals of the ambiguous figure

The number of reversals of the ambiguous figure seen in one minute was recorded. That is the number of times a participant reported that the phenomenological experience

changed from that of a cat to a swan while they continuously viewed the figure. Subjects who did not see both versions of the figure (n=8) were coded as seeing no reversals. Additionally, two subjects did not complete this test, as they did not view the figure continuously for 1 minute. The number of reversals of the figure in a 1-minute period by group is shown below.

Diagram 22 Number of reversals of the cat/swan seen by Group

	No. reversals/min							Total
	0	1	2	3	4	5	6	
Group 1	10	5	2	2	1	0	0	20
Group 2	16	0	0	4	0	0	0	20
Group 3	7	0	4	2	0	3	2	18
Total	33	5	6	8	1	3	2	58

Kruskal-Wallis test for the cross tabulation of group by number of reversals seen gives χ^2=5.97, df=2 Exact significance p=0.01. Further examination shows that group 3 is different from the other two groups who are not significantly different from each other. Participants from group 3 saw more reversals of the ambiguous figure.

In summary, our results strongly support the hypothesis that participants with high levels of autistic traits will show significant differences in their performance on certain cognitive measures compared to young people who do not have these traits. On most of our measures, it is the PDD threshold (SCQ≥15) on the SCQ that is the best cut-off for detecting these differences in cognition. For example, we have shown that young people who score above 15 on the SCQ are more likely to fail a second order false belief question despite having a relatively high general level of intelligence compared to young people with SCQ below 15. We have shown that young people with an SCQ score of over 15 see significantly fewer reversals of an ambiguous figure than young people with

an SCQ below 15. This indicates that those with SCQ over 15 have a lower level of cognitive flexibility. This result is highly statistically significant p=0.01.

We have also found support for young people with autistic traits having some resistance to visual illusions. We found this on one of the illusions. Young people with SCQ≥15 who passed the control test, were less likely to succumb to the Ponzo illusion.

On the block design test we found that the optimal cut off was SCQ≥22. Young people who scored over 22 on the SCQ were faster at the unsegmented block designs and gained less benefit from segmentation of the designs than the other participants. These results approached statistical significance at p=0.059 and p=0.083 respectively.

These results strongly support our original hypothesis that there would be differences in cognition in our sample as a function of SCQ status.

Now we proceed to multivariate analysis in order to:

a) Determine whether any interactions among these variables or with the WAIS/WISC subtest scores will increase our ability to distinguish between SCQ groups.
b) Indicate whether these variables will be additive in their contribution to predicting SCQ status. In other words, whether they all contribute independent information that is useful in modeling SCQ status or whether one or two variables can account for almost all of the variance alone.

Multivariate analysis

A forward stepwise binary logistic regression was performed. It was possible to create an equation that predicted membership of the non-PDD group versus the other two PDD groups combined. The number of subjects included was 58, excluding the 2 subjects who did not complete the ambiguous figures task. 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 and, as would be expected from the earlier analyses, number of reversals of the ambiguous figure entered first (chi-squared=9.01, p=0.003) Ponzo illusion (categorical) entered next

(chi-squared=5.27, p=0.02). It also included whether the second order false belief question was answered correctly (categorical) (chi-squared =4.27, p=0.04). The Hosmer and Lemeshow test gives a Chi square value of 7.32 for the full model, p= 0.40 indicating the model is an adequate fit to the data. The coefficient for Ponzo illusion (not being seen) was B=-2.18, SE=1.00 p=0.03, odds ratio =0.11. For the number of reversals B=0.56, SE=0.22 p=0.01 odds ratio =1.75 and for failing the second order false belief question B=-1.41, SE=0.71 p=0.05 odds ratio =0.24.

The number of reversals per minute produces the greatest increase in odds of being in the non-PDD group per increase in the number of reversals seen. For the Ponzo illusions and the 2nd order false belief question, failing the false belief question and not seeing the illusion produce a small but significant decrease in the odds of being in the non-PDD group.

BDDIFF narrowly missed entry to the model (p in 0.05 p out 0.1, iterations 20, cut 0.5), the p value for its score being 0.055. The above model correctly categorises 77.6% of cases. A model containing only a constant categorises 69% of cases correctly.

No other variables or interactions between variables were useful in creating predictive models including those including vocabulary scaled score and digit span scaled score.

Small adjustments to the model make little difference to the findings. For example, excluding the two cases that failed the second order false belief question means that BDDIFF then just makes criteria to enter the model. Giving the two cases that did not complete the ambiguous figures a score of zero so that all 60 cases are included also means that BDDIFF meets criteria. In neither of these adjustments is the overall shape of the model changed.

These results indicate that taken together these cognitive measures are useful in discriminating PDD from non-PDD groups. They contribute additively to discriminating the SCQ status of participants.

This provides some evidence that there is a specific profile for those in our sample who fall into the autistic spectrum (by SCQ) compared to those that do not. This provides support for the notion of a continuum in the underlying cognitive characteristics of autistic spectrum disorders that extends beyond strict diagnostic boundaries.

We know that our sample contains some young people who do have a diagnosis of an ASD, as our sample was drawn completely without reference to diagnostic status. It is possible therefore that the reported results are driven by the presence of those who do have a diagnosis of an ASD. For this reason, we will analyze the results by known diagnostic status. The diagnosis is as reported to us by the parent completing the SCQ.

Analysis by diagnostic status

Diagram 23 Diagnostic status by group.

	Group 1	Group 2	Group 3	Total
No autism diagnosis	12	13	20	45
Autism diagnosis	8	7	0	15

Subjects were included as having an autism diagnosis if their parents reported 'autism' or 'Asperger's syndrome'. One subject in group one was reported has having 'semantic pragmatic tendencies' and one subject in group two as having 'semantic pragmatic disorder'. These two subjects are in the 'no autism diagnosis' category. One subject in group 3 was also described as having 'autistic tendencies' and this person is also in the 'no diagnosis' category.

IQ

Diagram 24 Vocabulary and digit span scaled score by diagnosis

	Vocabulary scaled score, mean (SD)	Digit span scaled score, mean (SD)
No autism diagnosis	5.56 (3.06)	5.89 (2.58)
Autism diagnosis	6.67 (2.61)	6.93 (3.62)

An independent samples t-test shows that there is not a significant difference between the vocabulary scaled score and the digit span scaled score of the subjects reported to have a diagnosis of autism and those who do not. For vocabulary scaled score t_{58}=-1.26, p=0.21. For digit span scaled score t_{58}=-1.22p=0.23.
As there are fewer subjects with a diagnosis than are in the 'autism' group by SCQ, the power to detect real differences between groups in these comparisons is low.

Theory of Mind

First order false belief

Of the four subjects who failed the first order false belief task, only one had a diagnosis of autism. Of the other three, one had semantic pragmatic disorder, one had Williams syndrome and one no diagnosis.

Second order false belief

Among the subjects with no autism diagnosis, twenty-four out of forty-five (53%) passed the second order false belief test. Five out of fifteen with an autism diagnosis passed the second order false belief test (33.3%). (65% and 40% by SCQ).

Ponzo illusion

Of those with an autism diagnosis nine out of fifteen (60%) saw the illusion. Of those without a diagnosis of autism, thirty-five out of forty-five (78%) saw the illusion. (65% and 95% by SCQ)

Reversals of the ambiguous figure

The mean number of reversals of the ambiguous figure seen by subjects with a diagnosis of autism was 0.93 (s.d. 1.39). The mean number seen by subjects without a diagnosis was 1.35 (s.d. 1.86). This is not statistically significant. (Means of 0.78 and 2.05 by SCQ)

Block Design

The mean difference between times for the segmented and unsegmented block designs was 18.59 (s.d. 21.64) for the subjects with no diagnosis and 11.85 (s.d. 9.38) for those with a diagnosis of autism. (Means of 19.39s and 11.93s by SCQ)

It can be seen that when the subjects are divided by the presence of an autism diagnosis the pattern of results is the same. The difference between the groups does not seem to be greater and none is statistically significant. This indicates that our results are not due to a small number of participants with autism diagnoses who have extreme scores.

In fact the pattern of results is such that the subjects with a diagnosis perform in a slightly more 'autistic' profile (the exception being the reversal of the ambiguous figure) although not enough to cause the main results. However, the SCQ based non-PDD group performs in a more 'non-autistic' profile than the non-diagnosis group. That is, of the non-PDD profile: more likely to see the Ponzo illusion, more reversals of the ambiguous figure and more likely to pass the false belief task and a higher BDDIFF. This suggests that the diagnosis is more specific for this profile but the SCQ is more sensitive.

Relationship between variables

The correlation matrix below shows the correlations between the metric variables for the entire sample. The upper values are the correlation coefficients and the lower vales of each cell the p values.

Diagram 25 Correlation matrix

	SCQ score	Reversals/ min	Vocab scaled	Digit span scaled	BDDIFF
SCQ score		-0.26* 0.05	0.01 0.92	0.02 0.89	-0.22 0.09
Reversals/ min			0.19 0.17	<0.01 1.00	-0.20 0.13
Vocab scaled				0.40** <0.01	-0.52** <0.01
Digit span scaled					-0.24 0.07

The only tests with a significant relationship to SCQ score are the reversals of the ambiguous figure as would be expected from the results of the group analysis. Also as

expected the two intelligence subtests are highly correlated and they are correlated with the BDDIFF score.

Theory of Mind and Reversals of the Ambiguous Figure

The table below shows the number of reversals seen by participants who passed or failed the 2nd order false belief test.

Diagram 26 Number of reversals seen by false belief task score

Number of reversals seen	2nd order false belief failed	2nd order false belief passed
6	0	2
5	1	2
4	0	1
3	3	5
2	4	2
1	3	2
0	18	15
Missing	2	0
Total	31	29

On average subjects who failed the second order false belief test saw slightly fewer reversals of the ambiguous figure (mean 0.86 reversals s.d. 1.33) than those who passed (mean 1.62 reversals s.d. 2.04). In an independent samples t test t_{56}=-1.68 this is not significant p=0.1.

With the two subjects who failed the control tests removed from the analysis, t_{54}=-1.49, p= 0.14.

Central Coherence and Reversals of the Ambiguous Figure

BDDIFF and number of reversals of the ambiguous figure are not significantly correlated. Pearson's correlation r=-0.2, p=0.13.

Comparing those who saw at least one reversal of the ambiguous figure with those who did not see any, the subjects who did not see any reversals of the ambiguous figure (n=33) had a mean BDDIFF of 16.61s (s.d. 19.69) and those who did see at least one reversal (n=25) had a mean BDDIFF of 13.59s (s.d. 14.61). This would indicate that there is no relationship between seeing reversals of the ambiguous figure and this measure of central coherence.

No relationship was found between seeing the Ponzo illusion and reversals of the ambiguous figure with or without participants who failed the control question included. The mean number of reversals for those who did not see the Ponzo illusions =1.31, s.d. =1.74 n=16. For those who did see the Ponzo illusion the mean number of reversals is 1.21 s.d. =1.77 n=42.

These results and those from the correlation matrix support the conclusion that there is relatively little relationship between the independent variables in the sample as a whole.

Theory of Mind and Central Coherence

For the whole sample the non-parametric correlation between Theory of Mind and central coherence (as measured by the difference between conditions on the block design test (BDDIFF)) is not statistically significant Kendall's tau b=-0.14, p=0.18. Although it is not significant this negative correlation represents a trend for young people who passed the Theory of Mind tests to have low BDDIFF times, i.e. they had weak central coherence. Good Theory of Mind goes with weak central coherence.

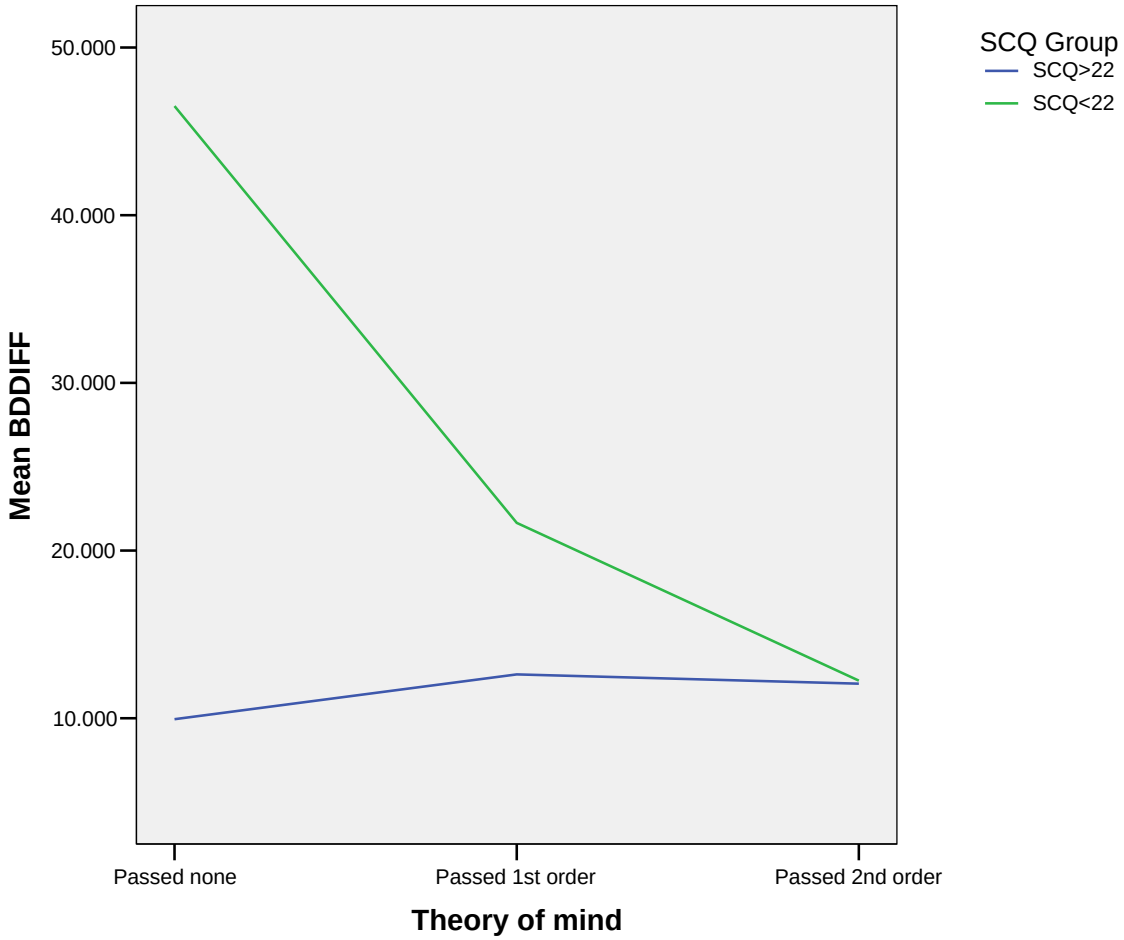
In our sample, the average time to complete a design in the segmented condition differs between those who pass or fail the second order false belief question. The mean time for those who pass (n=29) is 19.71 s and for those who fail (n=29) the mean time is 34.65s

this difference is significant t_{58}=2.46 p=0.02. This supports the conclusion that weak central coherence is associated with good Theory of Mind in this population.

Both Theory of Mind and BDDIFF have associations with general intelligence in our sample. The vocabulary scaled score of those who passed the false belief test (mean=6.48) is higher than those who do not (mean=5.1) p=0.07. BDDIFF is correlated with vocabulary scaled score p<0.01. The negative relationship between Theory of Mind and central coherence (good Theory of Mind being associated with weak central coherence) can be attributed to the effect of general intelligence. Those with high vocabulary scaled scores are both more likely to have low BDDIFF and to pass the Theory of Mind test.

It is then more striking that this relationship is broken in some participants with high autistic traits. We have found that this group is both more likely to fail the false belief task given IQ level and is more likely to have weak central coherence. This is illustrated in the graph below which shows the mean block design scores for people with high SCQ scores ≥22 and low SCQ scores <22 by how many of the false belief questions they answered correctly. The relationship between Theory of Mind and central coherence is very different for the autistic and non-autistic groups. It can be seen that for the high SCQ group BDDIFF does not vary at all by Theory of Mind score but for the low SCQ group it does markedly.

Diagram 27 Mean BDDIFF score by false belief task score by group.

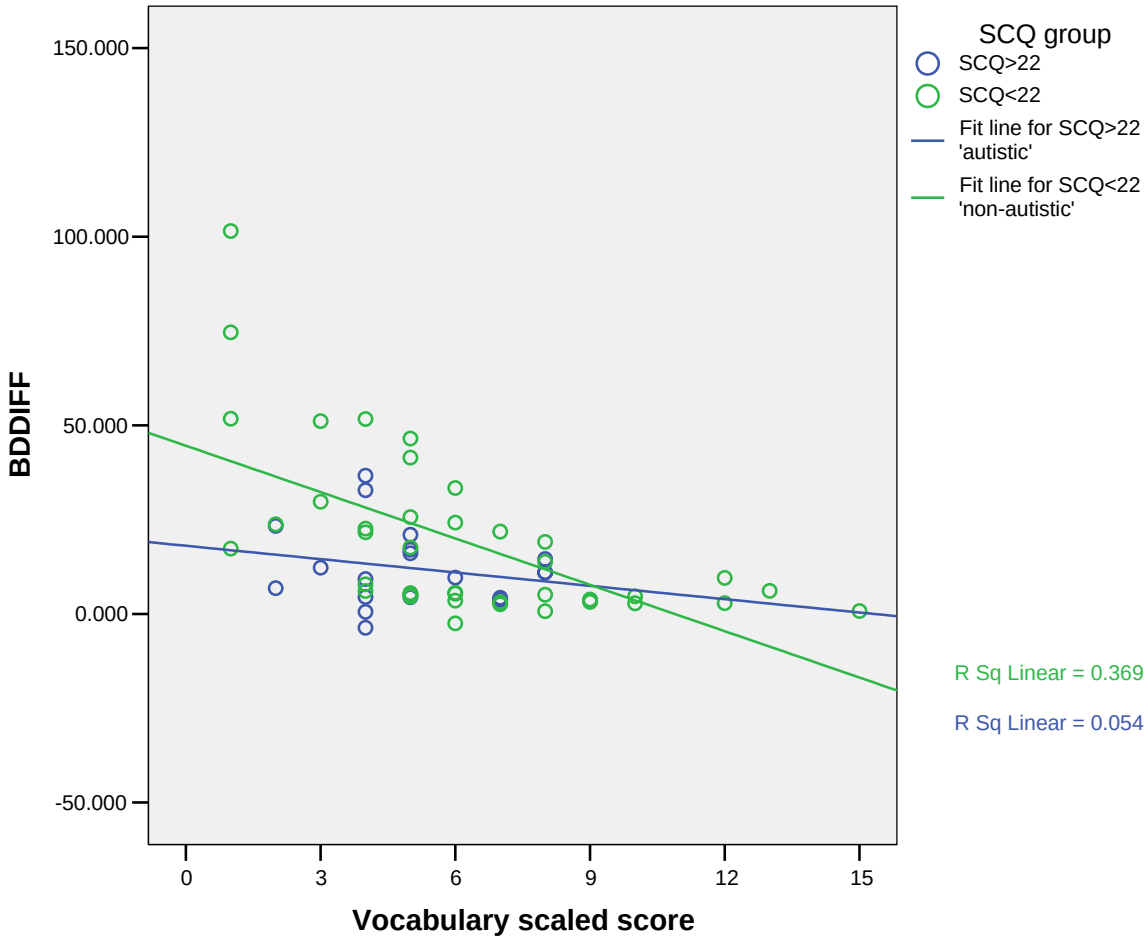


In the PDD groups there is no correlation between BDDIFF and Theory of Mind Kendall's tau b=-0.06 p=0.66 in the non-PDD group there is a large negative correlation Kendall's tau b=-0.38, $p<0.05$.

This mirrors the results for Theory of Mind where we found that in the non-PDD group those who failed the Theory of Mind task had low vocabulary and block design scores compared to those who passed. In the PDD (high SCQ) groups the vocabulary and block design tests scores were almost identical between those who failed and those who passed the Theory of Mind tests. In the PDD groups, there is dissociation between general intelligence and Theory of Mind ability.

In the young people with very high SCQ scores ≥22 there is also a dissociation between BDDIFF score and general ability. This is shown in the scatter plot below.

Diagram 28 BDDIFF by Vocabulary Scaled Score



In the young people with SCQ≥22 vocabulary scaled score accounts for very little of the variance in BDDIFF R^2 =0.05 where as in young people with SCQ<22 it accounts for a much lager amount of variance R^2 =0.37. Vocabulary scaled score accounts for 37% of the variance in difference between block design conditions in young people with low levels of autistic traits and 5 % in young people with high autistic traits. This could be attributed to the high SCQ group being at ceiling on this task but the fact that only 9 of the 20 participants with SCQ≥22 could complete all 8 unsegmented designs argues against a ceiling effect.

In conclusion, we have shown that a number of variables are sensitive to differences in cognition between young people with autistic traits (SCQ≥15) and those without. In the young people with lowest autistic traits, the relationship between performances on these measures is mediated by general intellectual ability. In the group with the highest autistic traits, this is not the case and there is a much lower correlation between measures.

Discussion

Our results clearly show that young people with high levels of autistic traits can be differentiated from those without these traits at a group level by their performance on cognitive measures, namely the ambiguous figure, the Ponzo illusion and the second order false belief test. These measures clearly separated the PDD group (SCQ≥15) from the non-PDD group (SCQ<15).

The reason the difference was significant at the PDD cut-off on the SCQ (15) as opposed to the autism cut-off (22) is that the sensitivity and specificity are greater. (The 15 cut off on the SCQ has sensitivity of 0.85, specificity 0.75 for PDD versus non-PDD diagnoses. The 22-cut off has sensitivity 0.75 and specificity 0.60 (Berument et al. 1999). The sensitivity being the proportion of true positives detected and the specificity the proportion of true negatives rejected).

The exception is the block design test where the clearest differences in our results were between the SCQ≥22 and SCQ<22 groups as we shall later discuss.

Ambiguous figures

The test on which we have the most statistically significant result is the ambiguous figure. We found that the PDD group (SCQ≥15) saw significantly fewer reversals of the ambiguous figure than the non-PDD group.

Ambiguous figures have only recently been used in research on autism. They have transferred from research on typically developing children investigating the relationship between perception of ambiguous figures and Theory of Mind development. The table below shows the research findings on the perception of ambiguity and Theory of Mind in typically developing children.

Diagram 29 Research on Ambiguous Figures and Theory of Mind

Authors	Participants	Materials	Measures	Results
Gopnik and Rosati (2001)	29 TD 3.5-4 year olds (41-59 months, M=49.7 months)	Rabbit/ duck, Man/mouse, OR Vase/faces	Spontaneous reversals	Never reversed spontaneously-recognized could have 2 interpretations after explanation .
			Informed reversals	28% reversed. 80% passed false belief task. 17 passed fb but did not reverse only 2 showed opposite pattern. ($p<0.05$)
			Unexpected contents task	
	11 TD 4 year olds and 17 TD 5 year olds.	Rabbit/ duck, Man/mouse, OR Vase/faces	Informed reversal	Age effects. Strong correlation between informed reversals and performance on Droodle task.
			Droodle task	
	Subjects in this study excluded if they did not pass control tasks		Control task Holographic sticker and normal sticker.	Children will report physical changes.
Mitroff (1998)	TD 5-9 years		Spontaneous reversals	Children who spontaneously reversed were more likely to pass the 2nd order Theory of Mind task.
			2nd order Theory of Mind	

TD –Typically developing.

Gopnik and Rosati (2001) in their study of typically developing children found that children of 3-4 years old never spontaneously reversed. That is, when presented with an ambiguous figure they never recognised both interpretations of the figure without prompting. All of the children eventually recognised that the figures could have another

interpretation but even under this informed condition, the majority did not see the figure change from one interpretation to the other when they viewed it continuously for one minute. That is, they did not experience alternate percepts (reversals) as they viewed the figure.

The participants in Gopnik and Rosati's study were also tested with an unexpected contents task, which is a first order false belief task. The overwhelming trend was for children to be able to pass the false belief task and not see reversals rather than being able to see the reversals and not passing the false belief task. This would indicate that the cognitive skills underlying false belief task reasoning are also necessary for perceiving the reversal of the ambiguous figure. However, performance on the false belief task was almost at ceiling. A second part to this experiment examined performance of 4 and 5 year old children on ambiguous figure and the Droodle task. The Droodle task is a test of whether children understand that people can interpret ambiguous stimuli in different ways. A line drawing of a flower on a piece of paper is covered with another sheet of paper that has a circular hole cut in it. Only a small proportion of the drawing of the flower shows through. The child is asked what they think the picture is of. The child will guess incorrectly as the lines showing are not suggestive of a flower. The top piece of paper is then removed and the flower is revealed. The child recognises it as a flower. The top paper is replaced and the experimenter asks the child 'if (classmate) comes in here and I show him this picture what will he think it is?' If the child correctly states that the classmate will guess erroneously, the child passes this test. Gopnik and Rosati (2001) found a strong correlation between informed reversal and performance on the Droodle task.

Gopnik and Rosati (2001) also included control tasks to ascertain whether the children would report the absence or presence of reversals in the presence of real physical changes. The control task consisted of two types of stickers. One was a sticker that did not change when you looked at it: the other was a sticker that did show changes in colour when viewed continuously. They found that the children did report both the presence and absence of real physical changes accurately.

Mitroff (Mitroff 1998; cited in Gopnik, Capps and Meltzoff, 2000) found that many 5-9 year old typically developing children identify both versions of an ambiguous figure without extensive prompting. The children that could do this were more likely to pass higher order Theory of Mind tests than those that did not.

Gopnik (1993) explains this link between Theory of Mind success and identifying both versions of an ambiguous figure through a 'relation between the ability to infer the mental states of others and the ability to have certain kinds of related first-person phenomenological experiences, such as the experience of reversal'.

Two studies have examined reversals of ambiguous figures in children with autism. These studies are shown in Diagram 30 below.

Diagram 30 Research on ambiguous figure perception in autism.

Author	Participants	Controls	Materials	Measure	
Ropar, Mitchell and Ackroyd (2003)	22 A CA 12;7 VMA 7;8	25 MLD CA 12;7 VMA 6;11	Cat/swan or Snail/whale	Spontaneous reversals	A=MLD spontaneous reversals. A>MLD failed false belief task.
Sobel, Capps and Gopnik (2005)	25 ASD (9 A, 16AS) CA 10.74 VIQ 104.64 FSIQ 101.76	22 TD CA 10.30 VIQ 108.00 FSIQ 108.68	Duck/rabbit AND Man/mouse AND Vases/face	Spontaneous and informed reversals	ASD<TD spontaneous reversals ASD=TD informed reversals
			2 nd order false belief Strange Stories	Theory of Mind	ASD +ve correlation spontaneous revs and Strange Stories and inverse relationship to informed reversals.

A - Autistic disorder, MLD - mild learning disability
AS – Asperger disorder
CA - chronological age, VMA – verbal mental age
VIQ - verbal IQ, FSIQ – full scale IQ.

Ropar, Mitchell and Ackroyd (2003) examined whether children with autism would be able to identify the two alternate interpretations of an ambiguous figure and whether this ability would be related to their Theory of Mind performance. 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. Sixteen participants in each group were able to identify both versions of the figure in this way. The autistic participants performed much more poorly on the false belief task however. There are two points to be made about this study. The first is that

1. what is being studied here is not the phenomenological experience of reversal as described by Gopnik et al. in their explanation above. In the Ropar et al. study, the participants could achieve criterion by forgetting what they had originally perceived the object to represent and recognizing it as something else. This is different
    a. from spontaneously relinquishing one interpretation and recognizing another as happens with spontaneous reversals, that is when subjects realize for themselves that the figure has two interpretations, without prompting, or
    b. from, while knowing that there are two interpretations of the stimulus, seeing them flip between interpretations as in informed reversals.

2. the participants covered a large range of verbal mental age. The 22 autism participants had verbal mental ages between 4;3 and 19;6. It is known that there are developmental changes in these capacities. Under the age of 5 years, typically developing children do not offer alternative interpretations of ambiguous figures and it may be that a more homogenous older sample would be a stronger test of this hypothesis.

Sobel, Capps and Gopnik (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 the ASD group, there was a positive correlation between spontaneous reversals and Strange Stories score. There was

an inverse relationship between informed reversals and Strange Story score in this group.

In conclusion, the evidence is:

1. There is a developmental progression in these abilities in typical development. No alternate versions are acknowledged by children of 4 and under. After 5 years old, the frequency of informed and spontaneous reversals increases with age.
2. There is a relationship between the ability to reverse and Theory of Mind ability in typical development.
3. In autism there is no evidence for reduced ability to identify both versions of an ambiguous figure but there is evidence that high functioning people are less likely than controls to spontaneously reverse the figures.

In our sample, we did not find any difference between the PDD and non-PDD groups in the tendency to spontaneously reverse. That is, there was no difference between groups in the proportion of participants who noticed for themselves that the stimulus could be interpreted in two ways. This is in contradiction to the results of Sobel, Capps and Gopnik (2005) who found that their younger, higher functioning participants with ASD were less likely to spontaneously identify both versions of the figure. This difference in findings may be due to chronological age differences between the samples, as we know that these abilities undergo developmental changes.

We found no difference in the proportion of participants in each group that could appreciate that the figure could have two interpretations. After being informed that the stimulus could have two interpretations and the researcher telling the participant what these two interpretations were, eight of our subjects did not think that the stimulus could be both a cat and a swan. Five were PDD and three non-PDD participants. This result is in agreement with that of Ropar, Mitchell and Ackroyd (2003) who found autistic participants were as able to acknowledge both versions of a figure with prompting.

Our results indicate that when informed of the ambiguous nature of a stimulus people with autistic traits see fewer reversals of the ambiguous figure. Our PDD group saw significantly fewer informed reversals of the ambiguous figure than the non-PDD group. This differs from the findings of Sobel, Capps and Gopnik (2005) who found no difference between groups in informed reversals. They did however find that their ASD group was more likely to see no reversals at all than their control group. There are slight methodological differences between the studies. Sobel, Capps and Gopnik (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. We however asked subjects to report any reversals seen and only prompted when subjects did not see any reversals. Also, Sobel, Capps and Gopnik's (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.

The young people in our study who passed the second order false belief task on average saw twice as many reversals of the ambiguous figure than those who failed. This result does not reach statistical significance. Although our study was not designed to test these questions, there is an indication in our data that PDD status has a much stronger association with the number of reversals seen than Theory of Mind ability. The difference in mean number of reversals seen is much greater when participants are split between the PDD and non-PDD groups than when they are split between those who pass the second order Theory of Mind test and those who fail. There remains the possibility that the Theory of Mind tests we used lack the sensitivity to detect the full range of Theory of Mind ability in our sample. Had we used an advanced test of Theory of Mind, such as the Strange Stories, in addition to the first and second order tests then the power to detect a relationship between Theory of Mind and reversals of the ambiguous figure (should one exist) would have been greater.

Deficits in Theory of Mind are a well-established feature of ASD and we and others have demonstrated that experience of ambiguous figures is also different in ASD. As a possible theoretical explanation, we suggest that the ability to maintain two alternate

versions of reality is a common ability necessary for both tasks. We have also suggested in the Introduction that the flexibility indexed by the reversals of the ambiguous figure may be related to the characteristic deficits in autism. We suggest that the deficit is not just the ability to recognize that something may have more than one identity but also to be able to manipulate this knowledge flexibly. We would suggest that the typically developing human brain trials various interpretations of ambiguous stimuli in order to get the best solution. The best solution for a perceptual stimulus will fit with Gestalt laws of simplicity etc and conceptually solutions will be favoured that have personal meaning and are appropriate for a given context. Also, where it is necessary to switch perspectives rapidly, for example in ongoing social interaction, it is necessary to maintain awareness of both one's own perspective and that of one's conversational partners. We hypothesize that finding contextual solutions and maintaining dual representations of reality require cognitive abilities indexed by the reversal of ambiguous figures. These require alternation between alternative representations of a single reality over time and/or require maintenance of alternative solutions for a given stimulus in order to select the best response from them. We suggest that people with autism are not able to do this, particularly with reference to selecting from options based on contextual information and that this is due to an impairment in the ability to maintain multiple representations of reality while awaiting the application of contextual information to make a decision.

This characterization of the deficit in autism is distinct from more general theories of executive function deficits in autism. We are not suggesting that all areas of mental flexibility are impaired but specifically those that require a single 'thing' to have more than one identity and to manipulate these versions flexibly. We suggest that, for typically developing people, maintaining two mutually exclusive possibilities for a period of time while contextual or associative information can be applied to choose between them is a part of typical cognition.

We are proposing that autistic people are not able to maintain two mutually exclusive possibilities and are thus not able to use semantic information to make choices and

decide on strategies. We note that associative paired learning and recognition are unimpaired in autism (Williams, Goldstein et al. 2006).

For people with autism, the preferred mode of dealing with environmental information is sequentially and in a piecemeal fashion and they will adopt this mode unless task demands explicitly direct otherwise. When presented with the ambiguous figure, although able to see both versions, they will perseverate on one. We found some support for this hypothesis in that the group with autistic traits did see fewer reversals of the ambiguous figure. This does not of course confirm the role for reversals as an index of the maintenance of alternatives awaiting choices. What would be required to show this would be to demonstrate a relationship between the number of reversals seen and the ability to respond flexibly in a context-sensitive manner in the absence of explicit direction as to what the relevant criteria are. That is, in an open choice response task such as the homograph test without direction as to the nature of homographic words.

There is another possible explanation for these results and this also presents an alternative testable hypothesis. One feature of the results on the ambiguous figures task was the large proportion of subjects who saw no reversals of the figure. Thirty-three out of 58 participants who completed this test saw no reversals of the figure. Also, phenomenologically, the reversals of the cat/swan are much less marked than those of, for example, the Necker cube where participants sometimes gasp or exclaim when they see it 'flip'. There were no such indications that our participants were really seeing reversals in the case of the cat/swan. As participants were also prompted at twenty-second intervals about whether they had seen any reversals, it may be that the number of reversals is in fact a measure of

1. how suggestible the participant is, or
2. how inclined they are to try to please the researcher and see what they are supposed to.

Both these interpretations, but particularly the second, would mean that the ambiguous figure could be characterized as an advanced test of Theory of Mind. That is, the ambiguous figure is detecting differences between the subjects in how they respond to

the task demands and, more importantly, the researcher - rather than in how they perceive the stimulus.

It is known that participants can influence the rate of reversals at will. So we are not suggesting that participants were not reporting what they saw but that the desire to see what was expected was stronger for some participants than others and this was related to their level of autistic traits.

It would be possible to determine whether this explanation of the results is correct. One possible method, possibly the simplest, is to manipulate task instructions. For example, for one group (split into high and low autistic trait participants), instruct them that a large proportion of people do not see the figure reverse. They are told that they should view the figure for one minute and report if they see a reversal. For the other group, instruct them that the figure will reverse when viewed continuously and prompt every 15 seconds. We would expect:

	Group	Outcome
Positive bias task instruction	High autistic traits	Few reversals
	Low autistic traits	More reversals
Negative bias task instructions	High autistic traits	Few reversals
	Low autistic traits	Few reversals

A crossover design would be possible were two different figures used.

Another way to resolve this would be to make psychophysical measurements. Kornmeier and Bach (2006) discuss the ERP components of reversals and the Reversal Positivity that occurs 130ms after stimulus onset. This component is not found in exogenously induced reversals so would be an indicator of reversals occurring in the participant's perception of the figure. This would not control for the influence of intent on reversals. That is, subjects can speed up or slow down reversals at will. It would mean that the researcher would not need to prompt the participants to make verbal responses and therefore would not indirectly encourage the reporting of reversals. It is

also known that reversals are associated with eye movements (Gale and Findlay 1983) so eye-movement tracking would be an indicator of reversals.

From our results, we can see that the young people who passed the second order false belief task saw on average twice as many reversals of the ambiguous figure as those who failed. Although this result is not statistically significant it does provide some support for a relationship between Theory of Mind and the ambiguous figure. However, it does not distinguish between the explanations given above. Either

1. both the ambiguous figure and the Theory of Mind task require the maintenance and manipulation of multiple representations of reality. Both tasks require the appreciation that something can have simultaneously more than one meaning or
2. those participants who have superior social reasoning abilities were more swayed by the interpersonal factors and the task demands of the ambiguous figures task and were thus more suggestible to seeing reversals of the figure.

We have shown that people with high levels of autistic traits (SCQ≥15) see significantly fewer reversals of the ambiguous figure. Further research is needed to clarify, which if either, of the above explanations for these results is the case.

Visual illusions

The visual illusions shown to participants in this study were the Muller-Lyer, the Ponzo and the Hat illusions. The method followed that of Happé (1996) though using two-dimensional stimuli only. The Ponzo illusion gave a statistically significant result. That is, significantly fewer young people with high SCQ (≥15) succumbed to the Ponzo illusion than did young people with low SCQ when participants who failed the control task were excluded.

The results for the other two illusions were not significant. This result leaves some questions:

Why did only one of the illusions show differences between the groups? In Happé's (1996) study, she found that subjects with autism were more resistant to the Ponzo illusion than MLD controls but that they were not more resistant to the Muller-Lyer illusion which is what we have also found. Happé did not use the Hat illusion but did use the Kanisza, Titchner, Hering and Poggendorf illusions, all of which she found the autistic subjects less likely to succumb to. It may thus be that the selection of illusions is critical.

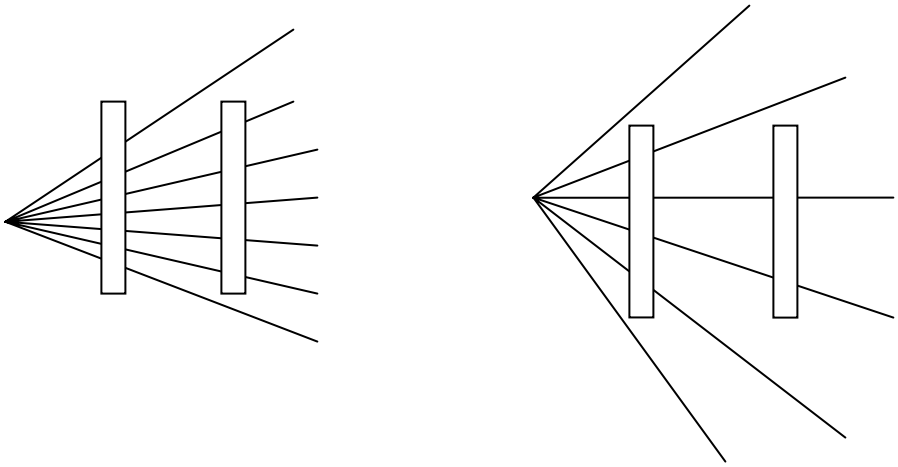
In Ropar and Mitchell's (1999) study, where subjects had to actively match the dimensions of the illusory stimulus on a computer screen, they found that autistic subjects were more susceptible to the Muller-Lyer illusion than controls. In this study they found that subjects with autism and Asperger disorder systematically distorted their estimations of illusory size as much as controls on the Titchner circles illusion, the Muller-Lyer, and the Ponzo illusion. Subjects with autism did not show any difference between estimates for the Hat illusion and control conditions. Although there were effects of condition for all the illusions, in that most subjects differed in their estimates between the control and illusion conditions, the absolute magnitude of the differences in mean size for some illusions was very small. For example, on the Titchner circles illusion, the actual diameter of the circle was 31 pixels, in the control condition the autistic group's mean estimate was 32 pixels, and in the illusion condition just over 29

pixels. The task was designed so that one key press adjusted the line length by two pixels. The same was true for the Ponzo illusion where the difference in average estimates between the control and illusion conditions is around 2 pixels, a single key press and about the size of two full stops. For the other two illusions, the differences in estimates between conditions were much larger. In one of these, the Muller Lyer illusion, there was clear susceptibility of the autism subjects to the illusion; but in the case of the Hat illusion there was not. Ropar and Mitchell's meticulous study provides evidence that the majority of subjects with autism are susceptible to illusions. It does not exclude the possibility that a proportion of individuals with autism are resistant to certain illusions.

Although all the illusions we have used can be described as illusions of perspective it is possible that within any one illusion there are multiple mechanisms operating to create the illusion and that between illusions the relative mix of these mechanisms is different. It may be the people with ASD are only immune to certain of these mechanisms and not others. Alternatively, as suggested by Happé, it may be even simpler than this. The Ponzo illusion is less integrated than the other two illusions we used, that is, the Hat and the Muller-Lyer illusions. In our presentation, the Ponzo figure must be separated into figure and ground in order for the illusion to be perceived. It may be that, as suggested by Happé, the Muller-Lyer is a more unitary figure and less likely to be perceived in a fragmented way.

Alternatively it may be a matter of the strength of the illusion. In our study and those of Happé (1996) and Ropar and Mitchell (1999), the Muller-Lyer was the most persuasive of the illusions used. We found that 80% of subjects saw this illusion. Happé found 86% of her subjects succumbed and Ropar and Mitchell found 91% susceptible. It may be that people with ASD are more able to resist weak illusions but still succumb to very powerful illusory effects. In Happé's study, significant differences were found on illusions where between 52 % and 76% of control subjects succumbed. Ropar and Mitchell found a result on the Hat illusions where on the verbal form of the task less than 25% of subjects succumbed.

In further studies, a wider range of illusions should be used. The illusion strength should be varied within each illusion. If the strength of the illusions was varied for example



thresholds for the perception of each of the illusions could be computed separately for autism and control groups that would provide a sensitive test of susceptibility to illusions.

Another possible interpretation was suggested by the fact that some of the subjects said that they were aware that the line looked longer but they knew that it was actually the same length. It may be that subjects with autism are more likely to answer the question literally in this situation. This could be due to the effect of some subjects having already had some experience with illusions. They then know that the stimuli have a deceptive nature so answer contrary to what they perceive. It is also possible that some subjects directly perceive their dual nature without previous experience of illusions. Some subjects said that that it looked longer but they knew it was the same length. This then introduces a situation were there is a single reality with two possible interpretations - a single reality with two identities, actual length and perceived length. It is just these situations, we have argued, that autistic people deal with differently. In the absence of the ability to maintain both realities, one will be chosen instantly and it is more likely to

be the more concrete i.e. actual length. Thus we would argue this is why autistic people are less likely to perceive illusion. The operation of this effect would be restricted to these situations where the subject becomes aware of the illusory influence distinct from the actual line length. This would be rather rare and would explain why immunity to illusions is not universal in autism.

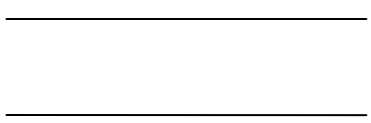
The control tasks for the illusion stimuli were the same lines but without the illusion-generating context. The correct answer for all the control stimuli was that the lines were the same length.

It is of concern that a large proportion of the participants failed the control tasks. For the Hat control, 22 out of 60 (37%) failed; for the Ponzo control 7 out of 60 (12%) failed and for the Muller-Lyer 13 out of 60 failed (22%). Ropar and Mitchell (1999) give data for how many subjects failed any of their 12 control tests and these were 4 out of 17 (24%) MLD, 3 out of 18 (17%) Asperger's, 10 out of 29 (34%) autism and 12 out of 35 (34%) with typical development. This is in the same order as the numbers that failed the control tasks in our study. In Happé's study, it is not reported how many subjects failed the control tasks as they were excluded prior to testing with the illusions, so her sample was more selected than that used by Ropar and Mitchell or ourselves which may be a reason for her clearer results.

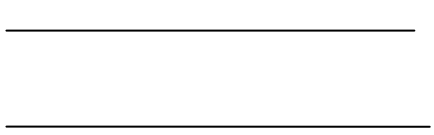
In future research, an improved design would be to include control tasks that are not the same size in order to test participants' sensitivity to physical differences in length, as opposed to illusory ones. Ropar and Mitchell (1999) used such control tasks in their study and demonstrated that subjects were more likely to report physical than illusory differences in length. However, as noted above, they had relatively high proportions of subjects failing their control tasks. In this population, it arguable whether it is appropriate to exclude subjects on the basis of errors on the control tasks since in our sample a large proportion of the subjects who failed the control tasks were nevertheless able to experience the illusions. The control tasks are included in order to check that participants are inspecting the stimuli carefully and are able to understand the questions they are being asked. The results from our subjects who failed the control tasks indicate

that they were not responding randomly, that is, that they were inspecting the stimuli. We assert this because the subjects who failed the control tasks and answered 'different' to the illusion stimuli overwhelmingly said the difference was in the direction of the illusion rather than the other way. In the case of the Muller-Lyer control 13 participants failed the control task and all of these succumbed to the illusion. The same was true for the Ponzo control task all who failed it saw the illusion. In the case of the Hat control task 22 failed, 15 of whom saw the illusion, 6 did not and correctly recognized that the two lines were the same length and only one person of 22 said that the lines were different in the opposite direction to the illusion. If the participants who failed the illusions were not looking at the stimuli then we would expect half to answer in the opposite direction to the illusory effect. A more plausible explanation is that the participants are seeing very slight differences in the control stimuli where there are none, a type of illusion in itself.

For example, are these lines the same size?



and are these lines the same size?



The first two lines are the same length and the second two are not. The best way to resolve this would be to do as was suggested for the illusions, to present lines of variable length and to compute individual thresholds for the perception of real and illusory

differences. It would be hypothesized that participants with autism would have comparable thresholds to controls for real differences but have higher thresholds for illusory differences.

It would be useful to ask participants whether they were aware of the illusory nature of the stimulus. That is, that it looked as if the lines were different lengths but they were actually the same length. We would predict that no participants with autism would report this. We would however expect participants with autism to be sensitive to the wording of the test question as suggested by Scott, Brosman, and Wheelwright (submitted, quoted in Happe and Frith 2006). That is, whether they are asked 'do these lines look the same length?' versus 'are these lines the same length?' We would expect that when the wording refers to what the lines look like, autistic participants would succumb to the same number of illusions as controls: but with the wording 'are these lines the same length?' then autistic subjects would succumb to fewer illusions. This is because we believe that autistic subjects can be directed to the level of interpretation of a stimulus that is appropriate. With highly unambiguous instructions, autistic participants will respond in the same manner as typically developing controls. However, when responses can be affected by the controls' access to a dual representation of a stimulus or state of reality, then autistic subjects will differ from controls.

Block design test

Differences were found between the participants with high SCQ≥22 and the participants with SCQ<22 on the block design test. These were on the average time to complete a design in the segmented condition, $p=0.059$ and on the difference between segmented and unsegmented conditions (BDDIFF) $p=0.083$. There was no difference in the number of designs completed between groups.

There was a tendency for the high SCQ group to be faster under all conditions but the difference between groups was greatest at the lowest levels of ability. Among the participants who completed fewer than five of the designs in the segmented condition there was a significant difference between the high SCQ and low SCQ participants in

how fast they completed the designs they did manage. There are two possible interpretations:

1. that the high SCQ group was less motivated and only completed designs that they found easy. This would in any case indicate that their underlying competence was in fact higher than that of the other group. The two groups completed equal numbers of designs under both conditions. If the high SCQ group members only completed a few designs due to low motivation and they still performed at the same level as an IQ matched control group then this indicates their underlying competence was in fact greater than the matched group.

2. that this is again a consequence of the inability to have multiple representations of a single stimulus. That is, when a complex figure is perceived, it always has the potential for multiple interpretations; among these it can be composed into a Gestalt or its constituent elements. Again we have a stimulus with multiple interpretations. We suggest that in most individuals these multiple versions are instantaneously available on viewing the figure and an almost instant decision is made as to the most meaningful/ context-appropriate/ useful choice. The other is then lost and can only be effortfully regained in most instances. In rare cases where there are two equally relevant options they are both maintained, as in the case of ambiguous figures or alternative homograph meanings, until the context can be disambiguated. In the case of the block designs, we suggest that most people have instant access to the whole and its constituents but the constituents are almost immediately lost in favour of the more meaningful whole shape. We would suggest that this is the source of the normal population variation in central coherence – how strongly consistently and immediately the choice is made for the whole versus the parts. Some people with a more piecemeal information-processing bias may chose the parts level as more useful. Caron et al (2006) examined performance on a modified block design task in high functioning autistic people with a block design peak, typically developing people with a block design peak, high functioning people with autism without a visuospatial peak and typically developing controls without a visuospatial peak. The variation of the block design task used manipulated the level of perceptual coherence of the designs. They found that increasing perceptual coherence

had almost no effect on the time taken for the HFA peak group to complete the designs and the typically developing controls with a BD peak performed in almost the same way. However, the typically developing group was strongly affected by the increased coherence of the designs. This supports the conclusion that for typically developing participants the coherence of the design is incrementally detrimental to performance. This is where we diverge from most current conceptualizations of the operations of Weak Central Coherence. We wish to propose that, instead of being an extreme on a continuum of normal population variation in preference for parts versus wholes, in autism the preference is random. That is, whether the parts or the whole is chosen is initially on a random basis.

The difficulty is how to define what a 'part' is and what a 'whole'. This has been exemplified with the Navon figure. Do parts and wholes distinguish on physical size? We suggest that for most people the distinction is on meaningfulness: but in autism when there are two possible interpretations the choice is made arbitrarily and the two are never maintained for context or meaning-driven choice to be made. There is a random mechanism that could go for the whole or the part but once a decision is made, there is no flexibility.

Recognition memory is intact in autism so if the level is externally determined participants with autism can find things they are asked to look for, for example, in the Embedded Figures Test where people with autism often display superior performance. This is an alternative explanation for good performance on the block design test: we suggest that typically developing people prefer meaningful shapes, so they will perceive these in the designs to the detriment of their performance. Typically developing participants then have to break these down in order to construct the designs out of blocks. However, for people with autism, whether the whole or part is perceived is random. Therefore, for a proportion of the designs they will do very well. In more able subjects, this may trigger a strategy in that they will then try to recognize the constituent shapes (triangles and squares) in future designs and use this method to achieve high performance. Other less able participants will find some designs very easy but others more difficult, depending on what interpretation they become locked in on for each

design. Siegel et al. (1996) estimated that 22-38% of people with autism have a relative peak on the block design test. That is, they have a comparatively high level of performance on the block design task in comparison to the other subtests of the intelligence scales. This would fit well with the theory that a proportion, but not all people with autism will develop a highly adaptive strategy for dealing with these types of test.

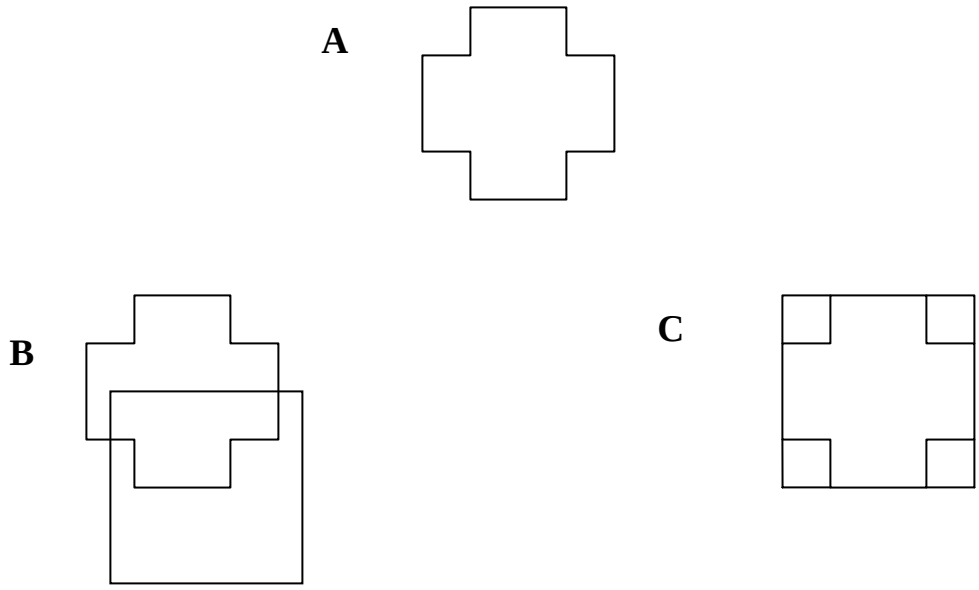
There is evidence of a developmental progression that would fit with our idea of some people with autism coming to use low-level perception as a strategy. Joseph et al.(2002) found a greater visual spatial peak in autistic children aged 8 years 11 months than in those aged 5 years and 5 months.

The predictions entailed by this explanation are that at higher ability levels, people with autism will be better or the same as IQ-matched controls at the block design test. At lower ability levels however, when the participants are not able to complete all designs, participants with autism will do very well on a subset of the designs and not on the rest. It should be noted that the block design test not only requires the ability to deconstruct the design into its constituent elements, but also requires participants to construct the design again from these elements. We predict that the designs that the autism participants do well at, will be a random subset. The constructive element may mean that these overlap with the designs that controls find easy.

Developmental perspective

Ghent (1956) found that young children find searching for embedded figures much more difficult than overlapping figures. For example if asked to find the figure A in figure B or C young children will find it much easier to locate the shape in B where lines intersect at points rather than C where lines of the square and cross are

co-extensive.



Children become more adept at the embedded figures as they get older. Ghent ascribes very young children's difficulty with the embedded figure task to a difficulty in perceiving lines as belonging to more than one shape. This explanation suggested to us the interpretation given above for the performance of children with autism on similar tests. This may seem perverse, in that people with autism excel at these tasks, whereas young children find them very difficult. However, the suggestion is that people with autism approach the task completely differently to typically developing controls. They use only object recognition strategies and locate the cross very easily.

Typically developing adults would see all possible versions of the figure instantly but then select the best figure conforming to Gestalt principles and the other would be lost. The Gestalt would have to be overcome to locate the embedded figure. In people with autism, they would not see all versions but only one possible interpretation and if this was the embedded figure then they would achieve very fast times. We also suggest that if they do not perceive the Gestalt image then they have less to overcome to use object recognition strategies to locate the figure.

We suggest that the difference would be brought out by an open-ended task where a complex figure was shown to participants and then participants were asked to show all shapes they could see. We would predict the autism participants would see fewer shapes and they would identify no shapes with mutual contours. That is, they would not use the same lines twice.

Relationship to other theories

The explanation we have outlined has different predictions to the theory of Weak Central Coherence. Weak Central Coherence predicts that those within the autistic spectrum who display Weak Central Coherence will use detail-focussed processing across a range of tasks. Our predictions differ for individuals with autism who are high and low functioning. We predict that participants functioning at low levels will display a high level of variability across tasks even within the visuospatial domain. This is because where there is the potential for multiple interpretations of a given stimulus then the way they interpret it is randomly determined. In any one task, a participant may have a learned strategy but across tasks in different cognitive domains, this will not be the case. In higher functioning individuals, we predict there is greater probability for compensatory strategies that extend across tasks. In these cases there will be individuals who have either generated strategies that they generalise by associative chaining across tasks or possibly through compensatory development of their basic perceptual functions: these individuals will have enhanced ability on tasks where perceiving the constituent elements of a stimulus is advantageous. The main prediction of our theory is that we believe that it is unlikely these strategies or compensated processes will generalise across domains. That is, it is unlikely that one individual will develop strong perceptual skills and this would affect their ability to chunk text for example. Therefore, we would predict that even in high functioning individuals there would be little relation between performance on the block design task and the homograph task once general IQ is controlled. We believe that both impairment on the homograph task and superior functioning on the block design and embedded figures tasks have common underlying

determinants - that is the inability to perceive two mutually exclusive interpretations of any one stimulus. This will lead to performance on these tasks differing on a group level from non-autistic samples. It will mean that a substantial proportion of autistic subjects will perceive the elemental version of the block design and thus construct it more easily and a proportion of the autistic subjects will choose the incorrect version of the homograph but these will not necessarily be the same members of the autistic sample and may well not be.

The main challenge for Weak Central Coherence and other theories of visuospatial cognition in autism is to explain enhanced visual search abilities (Plaisted, O'Riordan et al. 1998b) and the presence of normal global precedence in many cases (Ozonoff, Strayer et al. 1994; Mottron, Burack et al. 1999; Plaisted, Swettenham et al. 1999). We think the best explanation for enhanced visual search abilities is as compensatory development of the perceptual system in the absence of the typical top-down influences (an example of Paradoxical Functional Facilitation, Kapur 1996 as suggested by Mottron et al, 2006). We propose that object or feature recognition abilities undergo such hyperdevelopment in autism. This is very similar to Plaisted's (Plaisted, O'Riordan et al. 1998a; Plaisted 2001) theory of enhanced discrimination and reduced generalisation in autism. She proposes that the distinguishing features of an object are processed more fully than the features it has in common with other objects. This results in superior visual search abilities, particularly conjunctive visual search. Categorisation and the formation of prototypes are impaired because every item is perceived as unique rather than as part of a class, that shares similarities along a number of dimensions. This has a degree of overlap with our interpretation, as it entails a person with autism not being able to perceive an object as both itself and a member of a class i.e. not an orange and a fruit, via perceptual experience of oranges and fruit. However, the mechanisms proposed do differ; we do not predict any special status for the unique attributes of a stimulus and would not expect them to be more fully processed than attributes held in common. We would expect enhanced visual search abilities due to autistic subjects' specialisation in object recognition and would expect them to perform prototype recognition tasks based on visual similarity to exemplars, rather than based on the

creation of an internal prototype to match with. The two conceptions, enhanced discrimination, and ours, could be tested by comparing the memory of autistic subjects for parts of objects that were unique versus those held in common. If unique features were processed more fully in autism, then one would expect autistic subjects to have better free recall of these features than controls.

Mottron (Mottron et al. 2006) also proposed Enhanced Perceptual Functioning in autism. He suggests that all aspects of perceptual function are hyperdeveloped in autism, the levels of perception that are performed by the earliest visual areas V1 being the most superior. He proposes this as the primary cause of autistic deficits. The primacy of lower order perceptual processes in autistic cognition creates an imbalance between lower order and higher order processes in cognition under EPF. The 'optional property of higher-order intervention in lower-order operations' (Mottron et al 2006) is responsible for differences between autistic subjects and controls across tasks and produces much more variable effects than in neurotypicals. Happé and Frith (2006) in their update of the Weak Central Coherence theory state that global abilities are probably intact in autism and possibly low-level abilities are enhanced, rather than the earlier conception of impaired global abilities. They also suggest that instead of an obligatory reliance on local processing, people with autism have access to both but prefer local processing in open-ended tasks. We think that there is not a local preference per se but as in Mottron's (Mottron and Belleville 1993) previous description, an absence of perceptual hierarchy in autism. That is, whichever level of processing is achieved is initially arbitrary on a new task (but may later lead to strategies) in autistic participants. In comparison, in the general population a global preference is the norm. Thus, a local preference is apparent in autism but only relative to the general population's systematic preference for globally more meaningful interpretations.

The absence of global precedence in autism has been investigated using the Navon figure. We think that the results from these hierarchical figures are too contradictory to pose a threat to the Weak Central Coherence theory or any other theory at present. On the hierarchical figures task, it seems to be important whether the task is selective or divided attention. When the task is divided attention, then the subject has to respond to

the target whether it appears at the global or local level: when the task is selective attention then the subject knows whether they have to attend to the global or local level. It is easy to see why both autism and control groups should show a global advantage on selective attention tasks. They are being directed to the appropriate level for processing and the global level has a larger retinal size, so both groups would find it easier to perceive (e.g. Ozonoff, Strayer et al. 1994). Both our theory and the updated formulation of Weak Central Coherence would predict this result. The finding the autistic participants experience local interference in selective attention tasks (Rinehart, Bradshaw et al. 2000; Rinehart, Bradshaw et al. 2001) fits well with the Theory of Weak Central Coherence, which proposes a local processing bias in autism. That is, autism participants sometimes respond to the local rather than the global level indicated by task instructions. This finding could be interpreted as a lack of flexibility in the autism group (Rinehart et al 2001) when responding to changing task demands or that they are using a different strategy for the task. Our interpretation would suggest that the participants with autism are using a different strategy. They are using object recognition strategies to search the whole array for the target object on both levels. Typically developing subjects are probably using a verbally mediated strategy for example 'Is the big letter an S or an H'. This strategy is less likely to have interference from the local level.

In divided attention tasks, the findings are very variable. There have been findings of global advantage in autism (Mottron et al 1999) and of no global advantage (Plaisted et al 1999). There have also been inconsistent findings of global advantage in controls (no advantage controls in Mottron et al 1999, advantage in Plaisted et al. 1999).

As stated in the introduction, the Navon figure is very sensitive to variations in presentation - for example, duration of presentation, visual angle subtended, divided or selective attention, same or different targets used at global and local level. It is not even clear that it is a good example of a stimulus that is more than the sum of its parts and if it is so then this is only in the divided attention condition. In the selective attention condition, subjects are merely sequentially attending to a large and small stimulus - there is no need for subjects to register them as the same stimulus. There is also the possibility for executive effects of perseveration in clinical groups when subjects are

asked to switched from a previously rewarded area of attention to a new one (as suggested by Rinehart et al 2001), or to respond to a new target when the old one is present. This could be an alternative explanation for the findings of local interference in autism samples. That is, they have more trouble disengaging from the previously targeted level.

Research should now focus on the level of correlation between detailed focussed information processing across cognitive domains in autism and typical development.

Theory of Mind

We found that most of the people who failed the first order Theory of Mind test were in group 1, with the highest SCQ scores. Most people who failed the second order Theory of Mind test were in the SCQ range 15-22 and most people who passed all the Theory of Mind tests were in the lowest SCQ range under 15. However, in the Group 1 we did have a large number of participants who passed all the Theory of Mind tests. This meant that the relationship between Theory of Mind and SCQ score was not strictly linear. This may be because the first and second order Theory of Mind tests were not advanced enough tests of social cognition to detect more subtle Theory of Mind deficits in the more able participants in group 1. We found that of the participants with higher IQs, they were more likely to fail the second order false belief question if their SCQ was 15 or over than if it was under 15. The participants with SCQs under 15 failed the 2nd order question because of low verbal ability: the participants with SCQs over 15 failed the 2nd order question because of low verbal ability or poor Theory of Mind.

There were cases in our data of people with high SCQ scores and low vocabulary scaled scores who nevertheless passed the second order Theory of Mind test. This data was double-checked for accuracy and it was found that some young people fell into this profile. For one person whose recorded responses were checked in detail, it appeared that although he scored very low on the vocabulary test, this was due to highly idiosyncratic responses on this test. On the vocabulary test, participants are asked to

explain what a target word means. That is, they have to offer definitions for progressively more difficult words. This participant was very amenable to testing and tried hard to complete the task. However, his responses were close to word association rather than definitions and were tangential in their connection to the target word. Although the participant was obviously highly articulate, as the association were often sophisticated, but they were not accurate definitions of the given words and could not be scored as such. An improved method would be to give participants the full version of the standardised intelligence tests, as these would give a much better estimate of participant's IQ.

Despite the suggestion that some of the participants in group 1 may have passed the 2nd order false belief question while still having more subtle social cognitive deficits relative to their peers, we still find that the 2nd order false belief question was appropriate for the participant group. Exactly half of the participants passed and only two failed the control questions. The balance between having an extremely well established test of Theory of Mind versus a newer test with a wider range of possible scores was in this case in favour of a more established test. In future work, now that it is established that the SCQ can identify young people with Theory of Mind difficulties, this can be extended using advanced tests and/or naturalistic of Theory of Mind.

Relationship between Theory of Mind and central coherence.

Happé (1994; 1997) has found that Weak Central Coherence is independent of Theory of Mind skills in autistic individuals. She found that all her autistic sample had good performance on the block design test whether or not they passed Theory of Mind tests. She also found the same with visual illusions (Happé 1996) and the disambiguation of homographs (Happé, 1997).

Other studies that have examined the relationship in autistic and non-autistic samples have produced mixed results. Jarrold, Butler, Cottington and Jimenez (2000) found that in neurotypical adults (n=60), typically developing 5 year olds (n=24) and children with

autism (n=17), Weak Central Coherence was associated with poor Theory of Mind skills. For the typically developing children and the children with autism this was only the case when verbal mental age and chronological age were partialled out. However, Pellicano, Maybery and Durkin (2005) found that in a sample of 76 typically developing 4 and 5 year olds, central coherence was only weakly correlated with Theory of Mind and that this was eliminated when age, verbal and non-verbal ability were partialled out. They used a number of central coherence measures and found that these were not strongly inter-correlated and in fact formed two factors, one of which was related to constructional abilities. The other factor loaded more heavily on the tasks that involve disembedding a figure from its context. The construction factor correlated with Theory of Mind before partialling for age and verbal mental age and non-verbal mental age: the other factor showed no relationship at all with Theory of Mind. This is in contrast to the Jarrold, Butler, Cottington and Jimenez (2000) study where they used the Embedded Figures Test and the block design test and found a relationship between these measures and Theory of Mind. The sample of typical children used in the Pellicano, Maybery and Durkin (2005) study was much larger, 76 versus 24, 5year olds. Another difference between the studies is that Jarrold, Butler, Cottington and Jimenez (2000) partialled for age and verbal mental age and Pellicano, Maybery and Durkin (2005) partialled for age, verbal mental age and non-verbal mental age.

Lawson et al. 2004 used the Social Stories Questionnaire and the Physical Prediction Questionnaire in typically developing adults and adults with Asperger disorder. In the whole sample, they found no correlation between scores on the two measures.

Morgan, Maybery and Durkin (2003) found that in a sample of very young children both with and without ASD EFT performance was related to joint attention but that when diagnostic status was controlled for the relationship between the central coherence measure and the joint attention Theory of Mind measure was no longer present. They found that these measures independently contributed to the prediction of diagnostic status.

In our study, we have found no evidence of a relationship between Theory of Mind ability and central coherence other than the effects of general level of intelligence on the

measures. Using non-parametric test we found a weak negative correlation between Theory of Mind score (0-3) and BDDIFF the difference between average times for the two block design test conditions Kendall's tau b=-0.14, p=0.18. This weak negative correlation indicates that participants with high Theory of Mind scores were likely to have low BDDIFF scores. It is likely that this is an effect of general IQ, because across all participants the ones with higher IQ were more likely to pass the Theory of Mind and have low BDDIFF scores. As stated in the results, the difference between the non-parametric correlation for the PDD group and non-PDD groups is extremely large. In the PDD group there is no correlation between BDDIFF and Theory of Mind, whereas in the non-PDD group there is a strong negative correlation. From our results, it seems that the relationship between BDDIFF and Theory of Mind in the non-PDD group is driven by individual differences in processing capacity. In the PDD group, the relationship between Theory of Mind and processing capacity is broken resulting in a low level of correlation between measures.

It is possible that in our sample, the variation in intellectual ability that ranges from very impaired to slightly above average is much greater than would be found in typically developing groups. A more homogenous sample would be required to test the relationship between these variables in typically developing non-PDD participants. The use of multiple measures of Theory of Mind and central coherence would also further develop the research mentioned above.

Relationship between variables

We found relatively little evidence of relationships between any of the variables apart from what could be attributed to the effects of general intellectual functioning.

The multivariate analysis also supports this with the variables all making a significant independent contribution to the prediction of PDD status.

If as stated earlier, the fundamental disorder in autism is due to the inability to maintain multiple representations of a single reality and that this has consequences for the use of context to make choices then we would predict little relationship between variables. The

version of reality (real versus illusory length on the illusions, reality versus false belief on the Theory of Mind tasks, gestalt versus elements on the block design test, cat or swan on the ambiguous figure) processed on each task will vary arbitrarily for the PDD group leading to little relationship between variables.

Conclusions

We have shown that there are clear differences in the performance of young people with high levels of autistic traits and low levels of autistic traits on a number of cognitive measures known to be part of the cognitive phenotype of autism. The measures we used were 1st and 2nd order Theory of Mind, informed reversals of an ambiguous figure, visual illusions and the block design test in segmented and unsegmented conditions. All these tests were able to differentiate between these age, IQ and socioeconomic status matched groups that differed only on level of autistic traits present by parental report. There was however little relationship between the variables themselves leading to the conclusion that they independently predict aspects of a cognitive phenotype of autism. Our explanation of this is that the measures are all separate indicators of a representational inflexibility in PDD. That is, the representational level selected (global/local) is arbitrary in PDD as opposed to preferentially global and subject to contextual effects in typically developing participants. The idiosyncratic approach to tasks produced by this cognitive trait in PDD can sometimes enhance performance relative to controls, for example on the block design test.

There is some evidence that relatives of people with autism sometimes also show superior performance on the same measures as people with autism. The evidence for this 'broader phenotype' will now be described.

Part 2

Introduction

The Broader Autism Phenotype

The broader autism phenotype is the pattern of subclinical impairment of social reciprocity, communication and/or imagination observed in the close relatives of people with autism (Bolton, MacDonald et al. 1994) . An overview of the research in this field is shown in Diagram 31 overleaf.

Diagram 31 Research on the Broader Autism Phenotype

Coloured fonts indicate the contribution of the larger research groups. Other research groups that have contributed more than one study to this review have the same first author to their research publications. This summary of the research in this field is not exhaustive.

Authors	Relation	ASD	Control	Construct	Measure	Finding
Cox et al. (1975)	P	19 Infantile Autism (Rutter 1971 criteria)	23 DLD	Subclinical triad and psychiatric disorder	Interview and Leyton Obsessional Inventory	n.s. expressed warmth, emotional responsiveness, sociability, obessionality and mental health.
Folstein and Rutter (1977)	Monozygotic Twins	Autism	None	General cognitive	Twin study	Unaffected monozygotic twins showed MR, reading problems and speech problems
August et al. (1981)	S	41 Autism (Rutter 1971 def.) (71 S)	15 DS (38 S)	General cognitive	FHM	A>DS MR, reading and speech problems.
Wolff et al (1988)	P	21 A probands (35 P)	21 MR probands (39 P)	Subclinical triad	Blind interview	Autism parents lacked emotional reciprocity and had more restricted interests A>MR schizoid traits.
Freeman et al. (1989)	P, S	62 Autism (122P & 153 S)	Published norms*	General cognitive	WPPSI, WISC-R, WAIS, WRAT-R	No cognitive disorder in relatives

Smalley and Asarnow (1990)	S	9 A (15 P & 10 S)	9 TD (12 P & 10 S)	Cognitive theories	Benton test of facial recognition, Emotion matching and labeling tasks, vocabulary, comprehension and digit span subtests, test of line orientation.	n.s. S-A>S-TD line orientation
Landa, Folstein and Isaacs (1991)	P	29 Autism (41 P)	13 DS (13 P)	Subclinical triad	Spontaneous narrative	A<DS spontaneous narratives
Piven et al. (1991)	P	42 Autism (81 P)	18 DS (34 P)	Psychiatric	Subject version of Modified SADS-L	A>DS anxiety disorders
Landa et al. (1992)	P	28 A (43 P)	10 DS (10 P)	Subclinical triad	Pragmatic rating scale	A>DS pragmatic abnormalities
Gillberg, Gillberg, Steffenberg (1992)	P, S	35 A (68 P & 33 S)	42 DAMP (34 P & 65 S), 41 TD (102 P& 75 S)	General cognitive and psychiatric	FHM & subject interview	Am>DAMP & TD schizoaffective DAMP>A&TD general cognitive
Szatmari et al. (1993)	P, S	33 A, 12 atypical autism, 7AS (97 P & 84 S)	13 DS, low birth weight (54 P & 46 S)	Subclinical triad, general cognitive & cognitive theories	FHI, subtests of WISC-R/WAIS, WCST, WRAT emotion recognition	n.s.
Ozonoff et al. (1993)	S	14 A , 4 PDD (18 S)	18 LD (18 S)	Cognitive theories	Subtests of WISC-R/ WAIS, WCST, Tower of Hanoi, Theory of Mind tasks	No deficit on ToM. A<LD tower of Hanoi

Bolton et al. (1994)	FH	99 Autism probands (195 P, 137 S0	36 DS probands (72 P & 64 S)	Subclinical triad	Family History	A>DS communication and social deficits and stereotyped behaviours.
Piven et al (1994)	P	48 Autism (87 P)	20 DS (38 P)	Subclinical triad	M-PAS	A>DS social deficits- aloof, tactless and undemonstrative.
Bailey et al. (1995)	Twins	Autism	n/a	Subclinical triad and General cognitive	Twin study	92% concordance for social or cognitive abnormality in monozygotic twins and 10% concordance in dyzygotic twins.
Szatmari et al. (1995)	Parents and collateral relatives	PDD (103 P & 987 2nd & 3rd degree)	20 DS (66 P & 684 2nd & 3rd degree)	Subclinical triad	FHM	n.s.
Leboyer et al. (1995)	P, S	26 A females (48 P,31 S)	26 DS (41 P, 37 S)	Cognitive theories	Verbal and visuospatial tasks	Brothers of A <verbal skills than controls
Plumet et al. (1995)1995	P, S	26 A (47 P 31S)	26 DS (44P, 29 S)	Cognitive theories	Battery of tests for verbal abilities	A-P=DS-P A-S<DS-S
Smalley et al (1995)	P, S	44 A (62 P, 34 S)	21 Tuberous sclerosis or seizure disorder (26 P, 19 S)	psychiatric	K-SADS-E & SADS-LA	A>controls for major depression and social phobia
Piven et al. (1997)	Antecedents and collateral Family members	Multiple incidence autism (25 families= 25 M, 23 F))	30 DS families (30M, 30 F)	Subclinical triad	Family History Interview	A>DS on social deficits, and restricted interests and rigidity. Not communication
Piven et al. (1997)	P	Multiple incidence autism (25	30 DS families (30M, 30 F)	Subclinical triad	MPAS-R, PRS, FI	A>DS social impairments Aloof, hypersensitive, anxious and rigid.

		families (48 P)				
Boutin et al. (1997)	P, S	46 A, 3 PDD (95 P, 61 S)	18 MR (33 P, 22 S)	General cognitive	FHM	n.s.
Fombonne et al (1997)	P, S,	99A	36 DS	General Cognitive	Standardized IQ tests and tests of reading and spelling	n.s.
Piven and Palmer (1997)	P	25 Multiple incidence autism (47P)	30 DS (53P)	Cognitive theories	WAIS-R subtests, Woodcock Johnson tests of achievement, Rapid Automatised Naming tests, Tower of Hanoi.	A<DS 4 ring ToH planning, performance IQ, A>DS time to complete passage comprehension and automatised naming.
Hughes et al. (1997)	P	Autism (40 P)	LD (40 P), TD (36 P)	Cognitive theories	Working memory, planning, and ID/ED shift Spatial span	A<LD & TD working memory, planning set shifting. Spatial span =n.s.
Baron-Cohen and Hammer (1997)	P	AS (15 mothers, 15 fathers)	TD (15 mothers, 15 fathers)	Cognitive theories	Embedded figures test and Eyes.	AS m > TD m on EFT. AS f > TD f on EFT. AS m < TD m on Eyes AS f < TD f on Eyes
Baron-Cohen et al (1997)	P	919 Autism	40 Tourette's S 464 DS 98 DLD 125 TD	Cognitive theories	Parental occupation. Grandparent occupation – for autism and Tourette's syndrome families only.	Engineers were over represented as occupations of the parents and grandparents of people with autism.
Bolton et al (1998)	FH	99 Autism	36 DS families	Psychiatric	FHM & informant version of SADS-L	A>DS motor tics, OCD and affective disorder.

Folstein et al. (1999)	P, S	90 Autism	40 DS	General cognitive	WAIS-R, Word attack test, Pragmatic rating scale	A-P<DS-P on WAIS subtests and Word Attack Test A-P>DS-P early language problems
Piven and Palmer (1999)	Parents and FH	25 Multiple incidence autism families	30 DS families	Psychiatric	SADS-LA-R, FHI, MPA-R, PRS, FI.	A>DS major depression & social phobia in parents and wider family. Not related to presence of broader phenotype.
Hughes et al. (1999)	S	Autism (31 S)	DD (32 S) and TD (32 S)	Cognitive theories	ID/ED shift, Tower of London, Spatial working memory test and spatial span test.	A<DD & TD attention shifting and planning. A>DD & TD spatial and verbal span.
Pickles et al. (2000)	1st & 2nd degree relatives, aunts and uncles.	149 autism families	36 DS families	Subclinical triad	FHI	A>DS subclinical triad in extended family
Murphy et al. (2000)	Parents and adult siblings.	99 autism (195 P, 97S)	36 DS (72 P, 52 S)	Subclinical triad psychiatric	M-PAS, FHI;	A>DS anxious, impulsive, aloof, shy, over-sensitive, irritable and eccentric. 3 broad groups of traits: withdrawn, difficult and tense.
Happé, Briskman and Frith (2001)	P, S	5Autism/17AS probands	15 Dyslexia, 10 TD	Cognitive theories	BD, EFT, visual illusions, sentence completion	A fathers> DS & TD fathers on BD, EFT, visual illusions, and sentence completion
Bolte and Poustka (2003)	P, S	35 A probands (82 P& 20S)	21 SZ (35P & 11S) 22 TD	Cognitive theories	Emotion recognition	n.s.
Pilowsky, T., N. Yirmiya, et al. (2003)	S	27 autism	23 DLD 22 TD	Subclinical triad	CELF	n.s.

Dorris et al. (2004)	S	27 AS (17 families)	27 TD	Cognitive theories	Eyes – C BPVS-II	AS-S<TD-S Eyes-C
Ghaziuddin (2005)	FH	58 AS	39 HFA	Psychiatric	Family History Interview	AS >HFA relatives with depression, schizophrenia and BAP.
Bishop et al. (2006)	S	29 Autism & 13 PDDNOS	46 TD	Subclinical triad	CCC-2	A& PDDNOS<TD syntax subscale only
Bolte and Poustka (2006)	P	62 ASD	36 EOS, 30 MR	Cognitive theories	Embedded figures test, Block design test, Tower of Hanoi, Wisconsin card sorting, Trial making	ASD>EOS&MR on Embedded Figures test
Bolte, Knecht and Poustka (2006)	P	87 Autism simplex, 38 autism multiplex.	37 OCD, 34 EOS, and 27 MR	Psychiatric	Personality Style and Disorder Inventory, Symptom Checklist-90-revised.	A & MR>EOS reserved/ schizoid, Self-critical/insecure, critical/negativistic, loyal/dependant, and quiet/depressive scales of PSSI.

M-PAS - Modified Personality Assessment Schedule
MPA-R – Modified Personality Assessment Schedule-Revised - semi-structured instrument designed to assess 8 personality characteristics hypothesized to contribute to the broad autism phenotype.
SADS-LA-R – Schedule for Affective Disorders and Schizophrenia – Lifetime version modified for the Study of Anxiety Disorders, Revised.
PRS – Pragmatic rating scale assesses pragmatic language (19 items) and speech (6 items) during interview.
FI – Friendship Interview – semi-structured interview that assesses the number and quality of a person's friendships.
Together the MPA-R, the PRS and the FI can be combined to provide an index of the broader autism phenotype (Piven and Palmer 1999) .
EFT – Embedded Figures Test
Eyes – Mind in the Eyes task – test of social cognition (Baron-Cohen, Jolliffe et al. 1997). C – children's version .
BPVS II – British Picture Vocabulary Scale.

Evidence for the subclinical triad of impairments

Kanner (1943) observed the similarities between the social problems of the children he described as autistic and the subtle social deficits of their parents. He described the parents as aloof, cold and emotionally unresponsive although he emphasised their intelligence. He also described the parent's language as pedantic. It was not until about thirty years after Kanner's description of the children and their parents that researchers started to approach these parental characteristics in a scientific manner (e.g. Cox et al. 1975). Up until this point, the characteristics of the parents were viewed as a putative cause of their children's problems (Bettelheim, 1967). The re-conceptualisation of autism as a polygenetic neurodevelopmental disorder revealed the value of parental characteristics as an indicator of a common genetic mechanism.

Many of the first studies of parental characteristics consisted of clinical interviews. The interviews were designed to elicit evidence of poor social adjustment from the parent's life history and from their interpersonal manner (e.g. Wolff et al. 1988). Wolff et al. (1998) found that a higher proportion of the parents of autistic children had schizoid personalities than parents of children with mixed developmental difficulties. That is, more of the parents of people with autism were solitary, lacked empathy, were personally sensitive, were single-minded in pursuit of special interests and had unusual modes of communication. This description of schizoid traits largely converges with conceptions of the broader autism phenotype.

Information about greater numbers of relatives can be gained using family history methods (e.g. Baird and August 1985; DeLong and Dwyer 1988) . Family history methods are interviews with an informant family member to assess the presence of impairment in other family members not directly seen. Two research groups, those of Professor Rutter at the Institute of Psychiatry in London and Dr. Folstein at the John Hopkins University jointly developed the Family History Interview for Developmental

Disorders of Cognition and Social Functioning (Bolton et al. 1994) to measure the broader phenotype in relatives. These two research groups, in Britain and America have employed this measure in families with autistic member(s) and control families with a member with Down syndrome to investigate the broader autism phenotype. Their research is shown in green (Britain) and orange (America) in Diagram 31.

The Family History Interview probes for the presence of subclinical impairment in communication and social function and for the presence of stereotyped behaviours. The dimensions of social impairment included in the FHI are 'lack of affection, social dysfunction, impaired friendships, impaired social play, odd behaviour, and impaired conversation' and in communication 'language delay, significant reading retardation, articulation disorder and spelling difficulties'(Bailey, Palferman et al. 1998). Stereotyped behaviours include circumscribed interests, rigidity, and perfectionism. The family history method has found evidence of subclinical impairment in the social and communication domains in the immediate and extended families of people with autism in comparison to families of people with Down syndrome (Bolton et al. 1994, Piven et al. 1997, Pickles et al. 2000). There are studies using similar methods where no deficits have been found (Szatmari et al. 1993, and Szatmari et al, 1995). This may be because these samples included probands that had a PDD diagnosis as opposed to autistic disorder.

The family history method has the advantages that:

1. it allows data to be collected on large numbers of relatives
2. includes data on relatives who have not necessarily had children and this may be important if severe phenotypes affect the ability to form long term relationships and raise a family (Bailey et al. 1998)

and its disadvantages are that:

1. The quality of evidence is not uniform, close family members may be reported accurately but more distant relatives may be less well known by the informant
2. As awareness of the concept of the broader phenotype grows, it may be more difficult to obtain unbiased information on the traits of family members.

The disadvantages of the family history method have led to the increased use of methods of direct assessment with family members. One semi-structured interview designed to assess the broader phenotype in relatives is the Modified Personality Assessment Schedule (M-PAS, Piven et al. 1994). The personality characteristics included in the M-PAS – Revised (Piven et al. 1997) are:

1. Conscientious: dependable, steadfast, striving, single-minded (i.e. goal-directed)
2. Rigidity: little interest in and/or difficulty adjusting to change (i.e. new situation, ideas, or altered routines).
3. Aloof: lack of interest or enjoyment from being with people
4. Undemonstrative: restricted range (verbal and non-verbal) affective expression.
5. Anxious: nervousness or anxiety, not amounting to an anxiety state or phobic disorder.
6. Hypersensitive to criticism: excessive distress at comments or behaviour of others that are felt to be critical or insensitive.
7. Unresponsive: a lack of responsiveness to the emotional cues of others.
8. Untactful: behaviour that puts others off, upsets or irritates them, or may even lead to their suffering.

Studies using these methods have found that parents of people with autism are more likely to be rated as aloof, hypersensitive to criticism, anxious and rigid (Piven et al. 1997).

In the communication domain, the Pragmatic Rating Scale has been used to assess the social use of language (Landa et al. 1992). Evidence of language problems in the relatives of autistic probands has been found (Folstein et al. 1999, Landa et al. 1991) Folstein et al. found that parents of children with autism performed more poorly on the Word attack test (reading novel words) and had a greater incidence of early language delay. This raises the question of whether a distinction should be made between the

kind of pragmatic communication deficits observed by Kanner, such as pedantry, and evidence of deficits that could be described as specific learning problems. 'Spelling difficulties' included by Bolton et al (1994) in their Family History Interview for example, are specific learning problems. Bailey et al. (1998) suggests that 'the heterotypic continuities between language difficulties (Bishop 1992) raise the possibility that an increased risk for reading and spelling difficulties might derive from their association with language impairment'. That is that a higher incidence of reading and spelling difficulties in families may be due to a higher rate of lower level language problems. The evidence for more general cognitive deficits such as mental retardation and reading and spelling difficulties has also been examined in the families of people with autism.

General cognitive deficits

The first twin study of autism by Folstein and Rutter (1977) found evidence of increased concordance for autism between monozygotic twins compared to dizygotic twins. When they included Mental Retardation, speech and reading difficulties as part of the broader phenotype of autism then the concordance increased in both the monozygotic and dizygotic twins. Subsequently there have been mixed findings about whether the broader phenotype includes general cognitive deficits. August et al. (1981) found cognitive problems in siblings of autistic probands. Other studies have failed to find evidence of any cognitive deficit in the relatives of people with autism e.g. Freeman et al. (1989), Szatmari et al (1993), Szatmari et al (1995). It is thought that once the confound of proband mental retardation has been eliminated then there is not a link between autism and general intellectual impairment (Baird and August, 1985; Bailey et al. 1998).

In summary, family history and direct personality assessment measures have found evidence of subclinical features of autism in the relatives of people with autistic disorder. The most common feature is rigidity that is detected in up to 50% of parents (Piven et al. 1997). Having few friends and being aloof are also features of this broader

phenotype. There is evidence of pragmatic difficulties in the language of some parents of people with autism. Motor stereotypies are not generally present in relatives. The personality traits found in family members are common in the general population and lack specificity to autism. They could also be related to certain psychiatric disorders, for example rigidity could be associated with anxiety disorder.

Psychiatric disorders

Bolton et al (1998) studied 99 families of autistic and 36 families of Down syndrome probands. They found that relatives of the autistic people were more likely to have Obsessive Compulsive Disorder (OCD) and affective disorder. The family members with OCD were more likely to have the broader phenotype of subtle social and communication difficulties but the family members with affective problems were not. Piven and Palmer (1999) compared the rates of psychiatric disorder in families with multiple incidence of autism to the rates in families with one member with Down syndrome. The families of people with autism had higher rates of major depression and social phobia than the Down syndrome comparison group. This was not associated with the broader autism phenotype.

In a study that did not measure the subclinical triad, Ghaziuddin (2005) examined the family histories of people with Asperger syndrome and people with high functioning autism. He found that people with Asperger syndrome had a greater incidence of depression and schizophrenia in their family histories.

It would appear that affective disorders are more common in the families of people with autism but that this is not associated with personality traits of the broader phenotype. Family members are not depressed because they are resistant to change or are socially isolated. The liability to affective disorder is inherited separately. In the case of OCD, however these symptoms are associated with the broader phenotype.

In this area of research, it is important that the control groups are an adequate match for the stress caused by raising a child with autism. There is evidence that different types of

developmental disabilities place different levels of burden on the family and that having an autistic family member is particularly difficult (Bouma and Schweitzer 1990; Dumas, Wolf et al. 1991) . An indication that the raised incidence of depression among family members is not attributable to carer burden is that the first episode of depression usually occurred prior to the birth of the child with autism. Smalley et al. (1995) found that 64% of parents with major depression experienced their first episode of illness prior to the birth of their autistic child.

In their meta-analysis of research in this area, Yirmiya and Shaked (2005) found that the selection of a comparison group was critical to whether parents of people with autism had higher rates of psychiatric disorders. If the comparison group was families with a member with Down syndrome then parents of a person with autism did have higher levels of psychiatric disorder but compared to parents of a psychiatric control group they did not.

Cognitive theories of autism.

It is only comparatively recently that research has focussed on whether some of the cognitive theories of autism can be applied to relatives of those with the disorder. That is, whether the subtle deficits in social, communicative and imaginative behaviour present in relatives have the same psychological cause as the deficits in people with autistic disorder. The main cognitive theories: theory of mind, weak central coherence and executive function, have been examined.

1. Executive function.

Deficits in executive functions have been found in parents and siblings (Hughes et al. 1997-parents, Hughes et al. 1999-siblings) of people with autistic disorder. Planning deficits have been demonstrated on tasks such as the Tower of Hanoi (Piven and Palmer 1997, Ozonoff et al 1993). Deficits have also been found on tasks measuring spatial working memory (Hughes et al. 1997). The results are more mixed for set shifting deficits. There have been findings of no deficits in siblings (Ozonoff et al. 1993) and no

deficits in relatives of people with PDD (Szatmari et al. 1993) on the Wisconsin Card Sorting test. Hughes et al. (1997) did find deficits in attentional set shifting on the ID/ED shift paradigm.

2. Theory of Mind

Deficits have been found in social cognition in relatives of people with autism using the Mind in the Eyes task (Baron-Cohen et al. 1997-parents, Dorris et al. (2004)-siblings)

3. Weak central coherence

In contrast parents, especially fathers, have been found to have a relative strength on tasks that require fragmentation of a Gestalt, such as the embedded figures task (Baron-Cohen and Hammer, 1997a; Happé, Briskman and Frith, 2001; Bolte and Poustka, 2006). Other tasks that also favour low level visual processing such as the block design test in segmented and unsegmented conditions have yielded positive results (Happé, Briskman and Frith, 2001) with fathers of people with autism showing superior performance in the unsegmented condition but not the segmented condition. Other studies that have used the block design task as part of a general battery of IQ tests have yielded non-significant results (Smalley and Asarnow, 1990; Fombonne et al. 1997; Piven and Palmer, 1997).

Happé, Briskman and Frith (2001) found that fathers of people with autism were less susceptible to visual illusions and more likely to perform poorly on a sentence completion task. These two tasks are measures of weak central coherence in very different information processing domains. In the second part to this study, they found that these detail-focused information-processing styles were related to real life skills and preferences as ascertained by questionnaire measure (Briskman, Happé and Frith, 2001). Baron-Cohen et al. (1997) found that engineering was disproportionately common as an occupation of parents and grandparents of people with autism. The authors interpret this as reflecting the parents' preferred detail-focused information processing style.

Thus, there is some evidence for the existence of Theory of Mind deficits, executive dysfunction and weak central coherence in relatives of people with autism compared to

control groups. The number of studies in this area is however still small and these findings need verification.

The participants involved in part 1 of this study and their families provided an opportunity to test whether evidence of the broader phenotype could be found in the relatives of people with high levels of autistic traits.

General methodological considerations

1. Control groups

a. Choice of control groups.

Most studies have used families with a member with Down syndrome as a control group. This may not adequately control for the stress associated with raising a child with autism. This is particularly the case where studies compare multiple incidence autism families to single incidence Down syndrome. Other control groups that have been used include mixed developmental disorders, mental retardation, low birth weight, dyslexia, and typically developing controls. Another issue is that many of these control groups including Down syndrome are not associated with a genetic risk to family members. Recently, control groups of families with members with Early Onset Schizophrenia have been used to test the specificity of any cognitive phenotype to autism as opposed to other complex genetic disorders. In this study, we are fortunate to have a control group drawn from the same population as the target group. That is, we are planning to compare the parents of young people high levels of autistic traits to the parents of young people with low levels of autistic traits. Level of autistic traits was determined after participant ascertainment. Although it is still possible that there is a differential level of stress associated with raising a child with additional learning support needs plus autistic traits compared to a young person with only additional learning needs, the fact we are using tests of cognitive traits makes it unlikely this could explain any differences between group. It is not clear why the stress of raising a child with a high level of autistic traits

should make a parent immune to visual illusions whereas it is quite possible that it could make a parent socially withdrawn (Happé et al. 2001).

b. Matching control groups.

Control groups can be matched on the characteristics of the proband, family members or both. Variables that groups are commonly matched upon include for the probands: age, sex, level of functioning and in parents: IQ, socio economic status, and years of education. In this study, the target and comparison groups will be matched on proband characteristics. The probands and their controls are matched individually on age and sex and at a group level on IQ. This may lead to high variability in the characteristics of the parents but by using all the available parents of the young people who have already taken part in our study we maximise the available sample.

2. Proband diagnosis.

Early studies e.g. Cox et al. (1975) used Rutter's criteria for diagnosing autism. Studies that are more recent use ICD or DSM criteria. In early studies before routine screening for Fragile X syndrome, Fragile X cases may also have been included in the sample. Now variation is more likely to be introduced by the acknowledged inclusion of Asperger syndrome or PDD-NOS probands. In our study, we are not reliant on probands having a particular diagnosis, as we are interested in autistic traits and their expression in relatives.

Selection of Measures

It was planned that we should test the parents of the young people who participated in our earlier study on a battery of tests as similar as possible to those completed by the young people.

The visual illusions and ambiguous figure were used in exactly the same format as for the young people. This was possible as these tasks are based on simple perceptual judgements not expected to be influenced by IQ. An extra ambiguous figure was used in the parent group, the Necker cube. It was thought that as the parents would not have any attention or concentration problems then the number of figures used could be increased to see whether the results would be consistent between different types of ambiguous figure.

As the parents were expected to be of average intellectual functioning it was thought that the first and second order false belief tests would be unsuitable for this group. Every parent could be expected to pass these tests. As an alternative, we decided to use Happé's Strange Stories in a version recommended by the author for this purpose. The Strange Stories are an advanced Theory of Mind test. The version we used consisted of eight social stories, eight physical stories and eight jumbled stories. The social stories were taken from twenty-four developed by Happé (1994). These stories are short passages that describe common social situations where for example, a white lie, double bluff or emotional blackmail is used. They were developed to provide a more complex but naturalistic extension to the false belief tasks. They would detect subtle Theory of Mind deficits in high-functioning autistic people. Happé (1994) showed that able autistic people who could pass second order false belief tasks were still impaired relative to controls on the Strange Stories task. She also demonstrated that scores on the Strange Stories were related to false belief competence supporting their validity as a measure of Theory of Mind. The set of stories as used here are adapted from those used in Happé (1994). The set of Strange Stories used comprises eight as opposed to 24 social stories and the physical stories have been equated with the social stories in terms of difficulty. In this format they have been used in a number of different studies, for example Fletcher

et al (1995) used these materials in a functional imaging study to identify brain areas that are activated during the mentalising stories opposed to physical and jumbled passages in typically developing adults. Happé, Brownell and Winner (1999) compared the performance of patients with right hemisphere damage (RHD) to normal elderly controls on the mental and physical stories and found that the RHD patients were impaired on the mentalising stories. Happé et al. (1998) (1998) used these stories to compare Theory of Mind ability between older and younger adults. Kaland et al. (2005) have replicated findings that the Strange Stories task (as in Happé (1994)) is sensitive to impairments in social cognition in children and adolescents with Asperger syndrome. Thus, there is considerable evidence that these stories are sensitive to impairment in Theory of Mind.

To estimate the IQ of participants we used the National Adult Reading Test (Nelson 1982) and the Quick Test (Ammons and Ammons 1962; revised Mortimer and Bowen 1999). This is because these tests are quick to administer and give a good estimate of the IQ of participants. The National Adult Reading Test is a standardised test of intelligence based on the ability to read irregularly spelled words. Its main use has been in estimating premorbid IQ in elderly patients with dementia because reading ability is largely resistant to cognitive decline. It has been standardised on large samples of normal adults so for our purposes provides a fast measure of intellectual ability without the need for full IQ testing.

The Quick Test has been used to estimate current intellectual functioning in schizophrenic patients and in other groups. The participant is given a sheet on which there are four pictures. The pictures are of couples dancing, a cruet set on a tabletop, a racing track with cars and a police officer directing traffic. The researcher reads out a list of fifty words and the participant has to point to which of the pictures each word refers. The advantages of the Quick Test are that it is easy to administer and more readily acceptable to participants than the full IQ test. The Quick test has been used in a number of different populations. For example, Simon (1995) found that in low functioning criminal defendants the correlation between the Quick test and WAIS-R was 0.66. Gessler, Cutting and Frith (1989) found that in a group of nine patients with schizophrenia the Quick Test correlated with WAIS-R 0.91. Mortimer and Bowen

updated the original Quick Test developed by Ammons and Ammons (1962) in 1999. They made the pictures clearer and changed some of the test words from American English to an appropriate UK synonym. Mortimer and Bowen found that the revised Quick Test slightly over-estimated WAIS-R IQ, as did the original version. The Quick IQ test revised is reported to correlate r=0.72 with the WAIS-R IQ score in schizophrenic patients (Mortimer and Bowen 1999). The Quick test will therefore give us a useful estimate of IQ from which to determine whether the parent groups are well matched for general intellectual ability.

The parents completed the block design test in segmented and unsegmented conditions, as did the young people. Six extra designs of nine, as opposed to four, blocks were added to the test to make it more challenging. This meant that the parents completed fourteen designs in the unsegmented condition and fifteen in the segmented condition.

We wished to determine whether parent so of young people with high levels of autistic traits would show the same profile of performance as the young people. Therefore our hypotheses were that the parents of young people with SCQ≥15 would

1. succumb to fewer of the illusions, particularly the Ponzo illusion
2. see fewer reversals of the ambiguous figures
3. receive less benefit from the segmentation of the block designs
4. make fewer correct justifications on the social Strange Stories
5. perform at the same level on the physical stories, jumbled stories, Quick IQ test and NART IQ test as participants of young people with SCQ<15.

The young people were well matched for IQ between groups. It may well be the case that the parent groups are less well matched. If this is the case then multivariate analysis will be used. IQ can then be co-varied to statistically control for the effect of IQ differences between groups.

Method – Parents

Participants

The participants are parents of the young people involved in the first part of this study.

Recruitment of participants

It was anticipated that fathers would be more difficult to recruit than mothers would. This was firstly, because the biological father was living at home in only approximately fifty percent of cases. Secondly, the mother had generally been the main point of contact for the young people's involvement with the study. For this reason, families with the father living at home and families for whom the father was the main point of contact were approached first.

The researcher had already had contact with all the families of the young people participating in this study during the course of their recruitment and testing.

The researcher phoned the parents of the young people. Parents were asked if they themselves would be willing to take part in further research. If they expressed interest then the parents were sent an information pack in the post. When a completed consent form was returned, parents were considered recruited to the study. Thirty-eight parents agreed to take part in the study: twelve fathers and twenty-six mothers.

The researcher was blind to the SCQ score of the young people, therefore parents were recruited to this study without knowledge of their offspring's SCQ score. The researcher did however know which parents belonged to the same group without knowledge of whether the groups were high or low SCQ. This was in order that the recruitment was not highly biased towards a particular group.

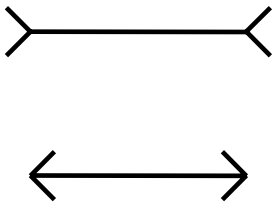
Testing

Testing took place in either the participant's own homes (n=35) or at the University (n=3). Testing took longer for the parents than for the young people due to the inclusion of more challenging stimuli. The maximum was 2 hours and the minimum 1 hour.

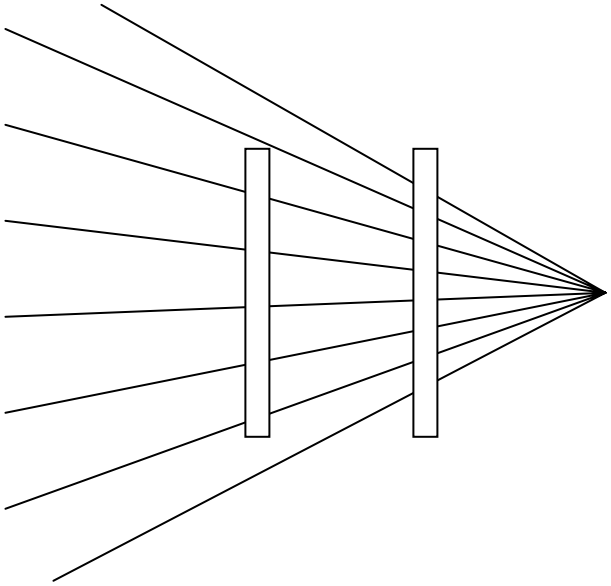
Experimental Tasks and Stimuli

1. Visual illusions

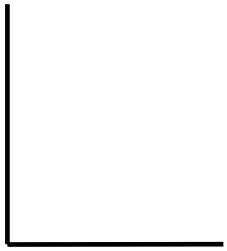
The visual illusions used were the same as for the young people. That is, the Muller Lyer,



The Ponzo illusion



and the Hat illusion.



The same materials, illusions and control stimuli, were used with the parents as with the young people. The parents were given the illusions printed in black and white on laminated paper. The parents were asked to view the stimuli at a comfortable distance. They were then asked whether the lines were the same length or different lengths. The critical lines were pointed out to the participants by the researcher. The order of the question (same / different) was counterbalanced between participants. If the parent

responded that the lines were of different lengths then the researcher asked which line was longer. The response was recorded.
The illusions were presented in the same fixed order as for the young people.

2. Ambiguous figures

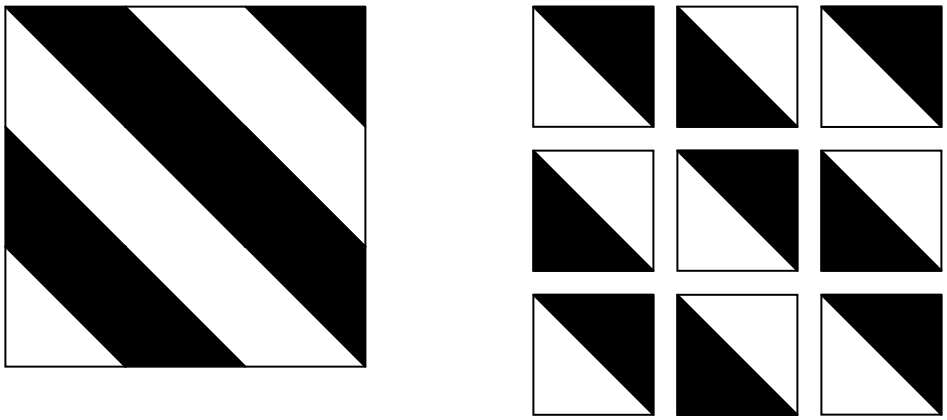
The cat/swan was presented in the same format as for the young people.
First, it was ascertained that the participant could see both versions of the figure. If they could not see both versions of the figure then the researcher pointed out the second interpretation. They were then asked to view the figure for one minute and report any reversals. A reversal was defined as a phenomenological change in perception of the figure from being cat to a swan or vice versa. If the participant made no response then they were prompted at 20 second intervals as to what version of the ambiguous figure they could see.
A second ambiguous figure was also used with the parents. This was the Necker cube. This stimulus was printed on unlaminated paper with the two versions of the cube presented in disambiguated form
below.

Parents were shown the Necker cube at the top of the page and its two possible interpretations at the bottom of the page. The researcher explained how if the lowest line of the Necker cube was at the 'front' of the cube then the lower left hand version would be seen. If the lowest line of the Necker cube was the 'back' of the cube then the

lower right hand version would be seen. Parents were asked to view the upper cube for one minute. They were asked to report if they saw any reversals of the cube during this time. Again they were prompted at 20 second intervals if they did not report any reversals.

3. Block design test

The parents were given the same four-block designs as the young people. They were also given an additional set of more complex designs to make sure the materials were sufficiently challenging. These designs used nine rather than four blocks. Six extra designs were presented. They were presented in unsegmented and segmented form. An example of a nine block design is shown:



The unsegmented designs were presented first and then the segmented designs. This was the order for all participants to avoid suggesting the strategy of segmentation. The time taken to complete each design was recorded. No time limit was given.

4. Strange Stories

The Strange Stories were presented in booklet form. There was one booklet for each condition: social, physical or jumbled stories. The participant was given the choice of reading the stories silently to themselves or of having the stories read out to them by the researcher. Thirteen of the thirty-eight participants elected to have the story read out to them. The order of presentation of the story types, social, physical or jumbled, was counterbalanced between participants.

It was explained to the participants that they could read the passage or have it read out to them as many times as they liked. When the participant was happy that they had understood the passage then they either turned the page over and read the question on the next page or indicated they wanted the question read to them. Once the question had been read they could not return to the story.

In the case of the social and physical stories the question essentially asked 'Why?' That is, the question required an explanation of what had happened in the story. In the jumbled stories, the question probed factual recall of one of the sentences contained in the passage. This required a yes or no response from the participant.

The time taken for the participant to study each story was recorded. Their response to each question was written down in full.

The responses to the social and physical stories could be scored as 0 for an incorrect explanation, 1 for partially correct or 2 for fully correct. Scoring was done with reference to the sample responses provided by the author. Scores for the social and physical stories could range from 0 to 16. Responses to the jumbled passages were yes or no and could be scored as correct or incorrect. Responses could range from 0 to 8. Scores for the jumbled passages were doubled so they could be compared directly to the scores on the social and physical stories.

5. The Quick Test

The participant was given a sheet of paper with four pictures printed on it. The pictures were of couples dancing, a cruet set on a table, a racing track with cars and a policeman directing traffic. The researcher read out a list of fifty words and the participant had to point to which picture best fitted the word. The words started simply e.g. belt (on the policeman) and became more difficult e.g. conviviality (the dancing).
Participants were encouraged not to guess at words that they did not recognise as they had a one in four probability of success by chance alone.
The number of correct pictures chosen was recorded.

6. The National Adult Reading Test

The participant was given a sheet of A4 paper with the words of the NART printed on it in two columns. The participant was asked to read the words aloud. The number of words correctly read was recorded.

Participants were given encouragement during testing but no specific feedback on correct or incorrect answers. The order of presentation of the tests was counterbalanced between participants.

Results – Parents

Participants

The table below shows the number of parents who participated in this study. The parents were split into two groups. Parents of young people with SCQ scores of 15 or over (high autistic traits) and parents of young people with SCQ scores under 15 (low autistic traits).[3]

Diagram 32 Parent Participants

Parents of	Mothers	Fathers	Total
Group 1 (SCQ≥15)	13	8	21
Group 2 (SCQ<15)	13	4	17
Total	26	12	38

Our main hypothesis is that parents of young people with high autistic traits (group 1) will show a distinctive profile on the cognitive measures compared to the parents in group 2. The profile is that found in the young people with high autistic traits: superior performance on the block design test, resistance to visual illusions, fewer reversals of the ambiguous figure and poorer performance on tests of social cognition relative to physical reasoning. It is not suggested that the parents will be clinically impaired but that a subtle profile may be present that includes some areas of superior functioning.

Before testing this hypothesis, we will determine whether the parental groups differ on any potentially confounding variables such as socio economic status or general level of intelligence.

Socio-economic Status

[3] A mother in group 1 completed only a limited subset of the tests. Therefore, for most results reported here the total number of participants is 37 unless otherwise stated.
Group 1 parents also included a mother who was not biologically related to the young person in the study, a stepparent. This mother was included in group 2, in the table above and for the other analyses, as her biological children were not on the autistic spectrum

Parents were recruited on a family, as opposed to an individual, basis. In every family where a father took part, the mother did also. For this reason, the socioeconomic distribution of the participants is shown separately for fathers and mothers.
The table below shows the socio-economic status of the mothers that took part.

Diagram 33 Socio-Economic Status of Mothers

ACORN classification	Group 1	Group 2	Total	UK pop.
44-56 Hard pressed	5 38%	7 54%	12 46%	22.4%
37-43 Moderate means	4 31%	2 15%	6 23%	14.5%
24-36 Comfortably off	1 8%	1 8%	2 8%	26.6%
13-23 Urban prosperity	2 15%	2 15%	4 15%	10.7%
01-12 Wealthy achievers	1 8%	1 8%	2 8%	26.6%

The Kruskal Wallis test for ACORN classification by group gives χ^2 =1.12 Exact significance, p=0.59. This shows that there is no relationship between group and ACORN classification. The groups are well matched on socio economic status. The lower socio- economic groups are slightly over-represented in this sample compared to the UK population. The table below shows the socio economic status of the fathers that took part.

Diagram 34 Socio-Economic Status of Fathers.

ACORN classification	Group 1	Group 2	Total	UK pop
44-56 Hard pressed	3 37.5%	2 50%	5 42%	22.4%
37-43 Moderate means	3 37.5%	0 0%	3 25%	14.5%
24-36 Comfortably off	0 0%	1 25%	1 8%	26.6%
13-23 Urban prosperity	1 12.5%	1 25%	2 17%	10.7%
01-12 Wealthy achievers	1 12.5%	0 0%	1 8%	26.6%

The Kruskal Wallis test for ACORN classification by group gives χ^2 =0.07 Exact significance, p=0.98. This again shows that the fathers in each group are well matched for socioeconomic status.

IQ Matching

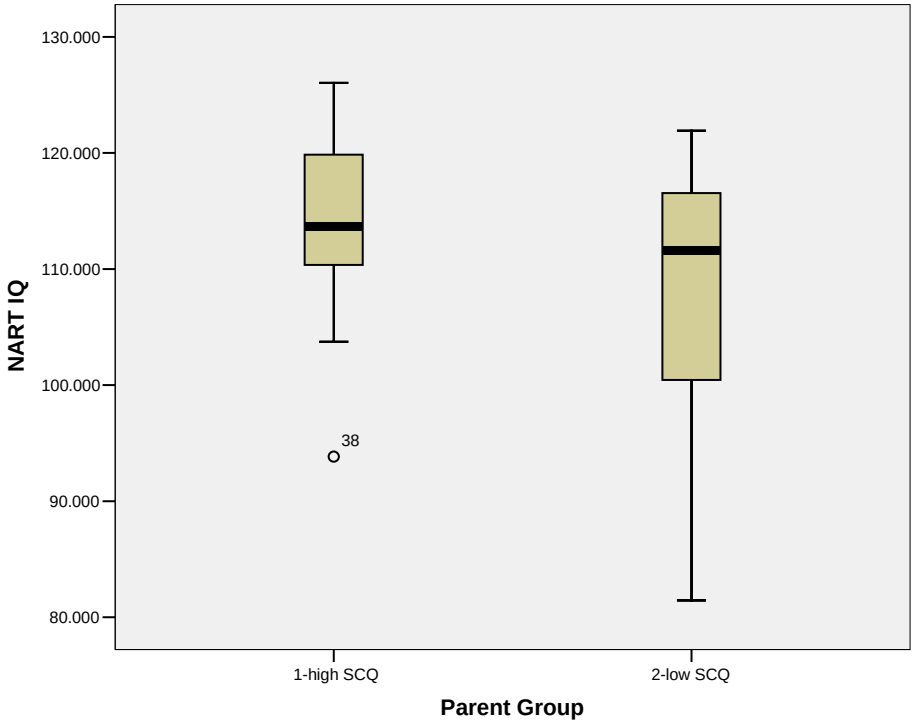
The parents completed the National Adult Reading Test and the Quick test revised. These measures were included to give a general measure of intelligence. This is so that we could establish that any differences found between groups were due to differences in their children's level of autistic traits as opposed to a difference in mean IQ.

Diagram 35 IQ Matching of Parent Groups

	N	Mean	Standard deviation
NART IQ Group 1 parents	19	114.53	7.81
NART IQ Group 2 parents	17	107.83	11.34
Quick IQ Group 1 parents	20	102.95	12.26
Quick IQ Group 2 parent	17	103.06	11.20

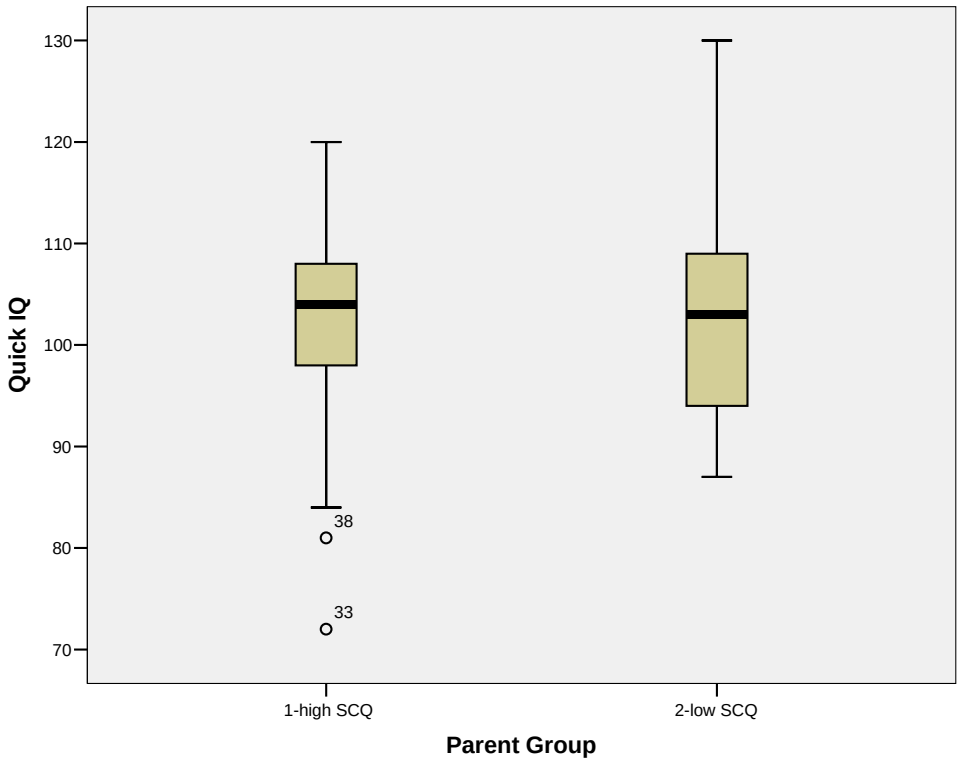
The subject numbers for the two measures differ as one subject in group 1 was not able to complete the NART. An independent samples t test shows there is no significant difference between the two groups on Quick IQ, t_{35}=-0.028, p=0.98.
An independent samples t test shows that the two groups have significantly different mean NART IQ scores, t_{34}=2.08, p=0.05.

Diagram 36 Box plot of NART IQ by Group.



Although the median values of the two groups are very similar, there is a longer 'tail' in the parents of young people with low SCQ scores. This indicates that a few low scores are distorting the mean. There is also one outlier below the median in group 1.

Diagram 37 Boxplot of Quick IQ scores by Group.



The groups are very well matched for Quick IQ. There are two outliers in group 1, one of whom falls into the borderline IQ range.

If the case with missing NART IQ data is also excluded for the Quick IQ analysis there remains no difference between the groups. Therefore the missing case from the NART IQ data does not explain why there is a difference between groups in NART IQ but not Quick IQ. We can conclude that the groups are discrepant for high-level verbal skills but are more closely matched on performance IQ.

To statistically control for any differences in verbal abilities between groups in addition to making bivariate analysis of the results, multivariate analysis will be used including NART IQ as a covariate.

We will now examine the performance of the two groups of parents on the target variables.

Target Measures

Strange stories

n=37

Our hypothesis is that parents of young people with high levels of autistic traits will perform more poorly than the other parents on the social stories. There will be no difference between groups on the physical stories or the jumbled passages.

The table below shows the mean scores obtained by parents in group1 (parents of high SCQ young people) and parents in group 2 (low SCQ) on the mentalising stories, the physical stories and the jumbled passages.

Diagram 38 Mean Strange Stories Scores

	Group 1 (Parents of high SCQ) n=20	Group 2 (Parents of low SCQ) n=17
Mentalising stories (max=16)	13.05 (1.91)	12.29 (2.54)
Physical stories (max=16)	11.85 (2.80)	11.65 (3.57)
Jumbled passages (max=16)	10.80 (3.14)	11.06 (2.93)

There were no statistically significant differences between the mean scores for groups 1 & 2 on the mentalising, physical or jumbled passages. An independent samples t test for the social stories $t_{35}=1.03$, $p=0.31$, for the physical stories $t_{35}=0.19$, $p=0.85$ for the jumbled stories $t_{35}=-0.26$, $p=0.80$. This result does not support the hypothesis that parents in group 1 would have lower scores on the social cognitive task. The null hypothesis that there is no difference between groups in social cognitive ability is retained.

Block design test

n=38

Our hypothesis is that parents of young people with high levels of autistic traits will show a smaller difference between mean time to complete a design in the segmented and unsegmented conditions of the block design test than the other parents. This is because they are hypothesised to have weaker central coherence and will be unaffected by the coherence of the unsegmented designs.

As with the young people, we found that not all of our participants were able to complete every design in the unsegmented condition.

The table below shows the number of designs completed in the unsegmented condition.

Diagram 39 Number of Unsegmented Designs Completed

		Group 1 SCQ≥15	Group 2 SCQ<15
Number of unsegmented designs completed Max.=14	9	3	1
	12	1	1
	13	5	3
	14	12	12
Total		21	17

As can be seen from the table only just over half of the participants (63%) were able to complete all the designs. Approximately the same proportion of parents from group1 and group 2 completed all the designs. Kruskal Wallis test gives χ^2=0.79, Exact significance p=0.40.

In the segmented condition, only one father from group 1 was not able to complete all the designs. Thus, almost half of the participants from both groups benefited from the segmentation of the designs in that they were able to complete more of the designs in the segmented than the unsegmented condition.

Next we will examine the difference between the mean time to complete a design in the unsegmented and segmented condition by group. The amount of facilitation given by the segmentation of the designs was no different for the parents of those with SCQ≥15

and SCQ<15. That is the difference between conditions was the same for both parent groups.

There are no differences between the groups for benefit gained from the segmentation of the designs. Mean difference (BDDIFF) for group 1 =15.04, for group 2= 15.20. Using an independent samples t test t_{35}=-0.06, p=0.95.

In the young people, the group who had the least benefit from the segmentation of the design were those with SCQ≥22. We re-examine the data to see whether the parents of young people with SCQ over 22 would gain less benefit from the segmentation of the designs than the other parents.

Diagram 40 Difference in mean time to complete a design between the unsegmented and segmented conditions by group (SCQ 22 cut off).

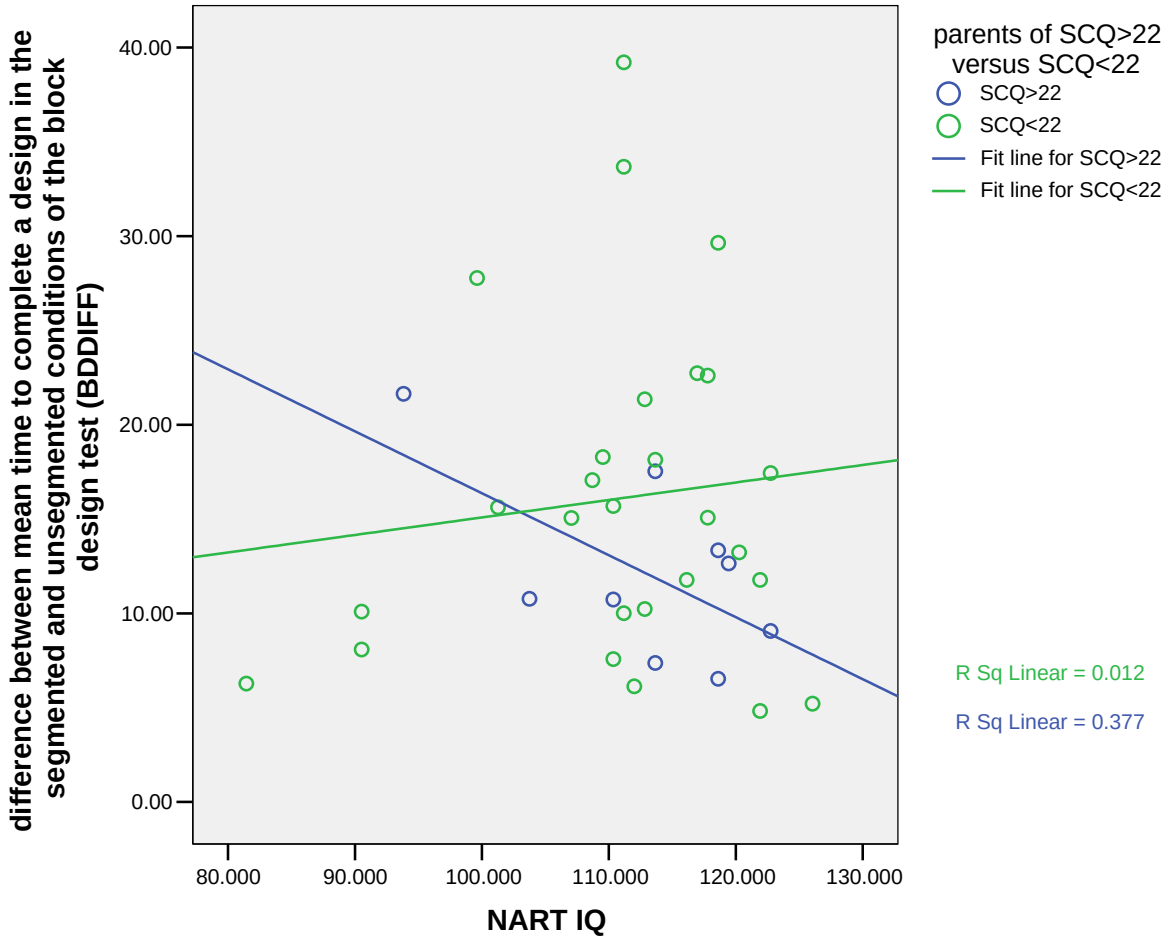
	Parents of SCQ≥22 (n=10)	Parents of SCQ<22 (n=27)
Difference between block design conditions/ secs	12.49 (4.68)	16.09 (8.82)

The parents of the young people with SCQs over 22, derived less benefit from the segmentation of the designs. This difference was not statistically significant. An independent samples t test t_{35}=-1.22, p=0.23.

The groups are differentiated by the fact that in the parents of young people with high SCQs the time difference between conditions (BDDIFF) was highly correlated with NART IQ (r=-0.61, p=0.08) whereas in the parents of young people with low SCQ scores it was not (r=0.11 p=0.58).

The scatter plot below demonstrates this.

Diagram 41 Scatter plot of BDDIFF by NART IQ.



Visual illusions

n=36

Our hypothesis was that parents of young people with high levels of autistic traits would be less likely to succumb to visual illusions than the other parents. The tables below show the number of parents in each group who saw each of our three illusions. Results are shown with participants who failed the control task for each illusion included 'All' or excluded 'Excluded'.

Diagram 42 Number of participants who saw the Muller-Lyer illusion by Group

	Group 1		Group 2	
	All	Excluded	All	Excluded
Saw illusion	14	11	15	14
Did not see illusion	5	5	2	2
Total	19	16	17	16

Fisher's Exact test, All p=0.41, Excluded p=0.39.

Diagram 43 Number of participants who saw the Ponzo illusion by Group

	Group 1		Group 2	
	All	Excluded	All	Excluded
Saw illusion	15	15	16	16
Did not see illusion	4	3	1	1
	19	18	17	17

Fisher's Exact test All p=0.34, Excluded p=0.60.

Diagram 44 Number of participants who saw the Hat illusion by Group

	Group 1		Group 2	
Saw illusion	17	12	15	12
Did not see illusion	2	2	2	2
	19	14	17	14

Fisher's Exact test All p=1.00 Excluded p=1.00.

The proportion of participants who succumbed to each of the illusions did not differ between groups. This was the case whether the participants who failed the control tasks were included or excluded.

Using logistic regression analysis forward stepwise method, we found that the interaction term Ponzo illusion*Quick IQ met criteria (0.05 in, 0.1 out, cut 0.5) to be

entered into a model to distinguish parents with children with very high SCQ≥22 from the other parents. This interaction term alone allowed the creation of a robust model. Change in the -2loglikelihood for the full model versus constant only chi squared =5.47, df=1, p=0.02. The Cox and Snell R squared =0.14. For the variable Ponzo illusion*Quick IQ B=0.02, SE=0.01, p=0.03 odds ratio=1.02. An odds ratio greater than 1 means that increasing values of the variable produce increased odds of being in group 2 (low SCQ in children) so a high IQ parent who sees the illusion is likely to be a group 2 parent.

Similar results are obtained if a hierarchical method is used and NARTIQ or Quick IQ are entered on step 1 and the Ponzo illusion entered on the next step. Ponzo illusion is a more significant predictor with Quick IQ than NART IQ. When Quick IQ is entered in step 1 the change in -2 log likelihood has chi squared =4.41 p=0.04. B=0.087 s.e.=0.046 p=0.03 odds ratio=1.102. Ponzo illusions as a categorical variable is entered on step 2 the change in the -2loglikelihood has a chi squared value of 4.66 p=0.03. B=-2.35 s.e.= 1.12 p=0.04 odds ratio=0.096. As Quick IQ increases the probability of being in group 2 does as well. Not seeing the Ponzo illusion increases the probability of being in group1.

Reversals of the ambiguous figures

Our hypothesis was that parents of young people with high levels of autistic traits would see fewer reversals of the ambiguous figures than parents of young people with low levels of autistic traits. The table below shows the number of reversals of the cat/swan seen by parents during a one minute inspection period. It also shows the proportion of parents in each group who saw no reversals of the cat/ swan.

Diagram 45 Mean number of reversals of the cat / swan seen by group (SD).

	Group 1	Group 2
N	21	16
Cat / swan	1.57 (3.40)	2.50 (3.67)
% seeing no reversals	61.9%	50%

There are no significant differences between the groups in the mean number of reversals of the cat/swan seen. In an independent samples t test t_{35}=-0.80 p=0.43. Fisher's Exact test of the cross tabulation of group and whether or not any reversals of the cat/swan were seen gives p=0.52. This shows there were no differences between groups in the proportion of parents seeing reversals of the figure.

In logistic regression analysis when NART IQ and a dichotomous variable 'any reversals of the cat swan seen yes/no' were entered created a model that predicted whether parents were of very high SCQ ≥22 or low SCQ<22 young people. Change in the -2loglikelihood between the full model and the constant only model=6.26 df=2 p=0.04. For the variable 'any reversals of the cat swan' (1) B=2.30 s.e.=1.14 odds ratio=9.99. For the variable NARTIQ B=-0.03 s.e.=0.05 p=0.45 odds ratio=0.97. An odds ratio greater than 1 means that increasing values of the variable produce increased odds of being in group 2 (low SCQ in children) so parents who see reversals of the cat/swan are more likely to be in group 2 (low SCQ) whereas a parent with a high NART IQ is more likely to be in group 1.

'Any reversals of the cat/swan' is a more significant predictor with NART IQ as a covariate than with Quick IQ.

The table below shows the number of reversals of the Necker cube seen by parents during a one minute inspection period. The table also shows the proportion of parents seeing no reversals of the Necker cube.

Diagram 46 Mean number of reversals of the Necker cube seen by group.

	Group 1	Group 2
N	19	12
Necker cube	11.37 (10.0)	12.58 (14.6)
% seeing no reversals	21%	16.7%

There are no significant differences between the groups in the mean number of reversals seen. In an independent samples t-test t_{29}=-0.28 p=0.79. Fisher's Exact test of the cross

tabulation of group and whether or not any reversals of the Necker cube were seen gives p=1.0. This again indicates no differences between groups in the proportion of parents who saw no reversals of the Necker cube.

Multivariate analysis does not show any relationship between IQ, seeing the Necker cube reverse and SCQ of children.

In summary, the variables 'Ponzo illusion' and 'any reversals of the cat/swan' when combined with Quick or NART IQ differentiate between parents of young people who have high levels of autistic traits (SCQ≥22) and parents of those who do not. The significant differences we have found are between parents of young people with the highest levels of autistic traits SCQ≥22 and the other parents.

Other research in this area has shown significant differences between mothers and fathers of people with autism versus controls, but only when mothers and fathers are considered separately. We will now examine whether our data also follows this pattern. Following from our findings so far, we will look for differences between mothers of young people SCQ≥22 and mothers of young people SCQ<22, and fathers of young people with SCQ≥22 and fathers of young people SCQ<22.

Results for mothers

Mothers of young people in group 1 (SCQ≥22, autism group) were significantly different from mothers of other young people in our study on the reversals of the cat/swan ambiguous figure. Mothers of young people in group 1 (n= 6) saw a mean of 0.33 reversals and mothers of the other young people (n=20) saw an average of 2.65 reversals. Using an independent samples t test, this difference in means was significant $t_{24} = -2.70$ p=0.01.

This result is highly significant and indicates a clear difference between the mothers in the two groups. Mothers in the high SCQ group saw much fewer reversals of the ambiguous figure on average than mothers of young people with low SCQ scores.

Results for Fathers

Fathers of young people in group 1 (SCQ≥22, autism group) did not differ from the fathers of the young people in the other groups on any of the measures.

Diagnosis

As with the results for the young people, it is necessary to eliminate the possibility that the results we have in our sample are due to the small proportion of parents who have a child with a diagnosis of autism or Asperger syndrome. Eight of the thirty-eight parents who participated reported that their child had a diagnosis of autism or Asperger syndrome (referring to six young people).

The parents of young people with a diagnosis of autism or Asperger syndrome performed significantly differently to the other parents on two of our measures. We found that contrary to what would be hypothesised, the parents of the people with a diagnosis of autism or Asperger syndrome performed better on the social stories than the other parents. Using an independent samples t-test $t_{35} = -1.94$ and this result approached statistical significance $p=0.06$: the parents did not differ on their performance on the physical stories $t_{35} = -0.05$ $p=0.62$. The other measure was the Muller Lyer illusion where, after removing the participants who failed the Muller Lyer control task from the analysis, the parents of people with autism or Asperger syndrome were less likely to succumb to the illusion $p=0.057$.

On all other measures, the parents of young people with a diagnosis of autism or Asperger syndrome did not differ from the other parents nor did they perform in a more extreme profile then the other parents in group1.

Relationship between parent and child scores

1. Fathers

    a. Block design test

We will examine whether there is any evidence in our data of a correlation between parent and child scores on the block design test.
For fathers and their children, the correlation between the mean time to complete an unsegmented block design for the child and father: Pearsons' correlation r= -0.08, p=0.81, n=12. This indicates there is no relation between the performance of children and their fathers on this measure.
The correlation between the child BDDIFF and the father BDDIFF=-0.06, p=0.86 N=12.

    b. IQ

There is also no statistically significant relationship between the young people's measures of intelligence and that of their fathers.

Diagram 47 Correlations between father and child IQ measures.

		Father NART IQ	Father Quick IQ
Child vocabulary scaled score	Correlation	0.43	0.48
	p value	0.19	0.11
	N	11	12
Child digit span scaled score	Correlation	-0.002	0.07
	p value	1.00	0.83
	N	12	12

There is a greater relationship between the child's vocabulary score and the paternal measure of IQ but this does not reach statistical significance. This may be due to the small sample as the correlation coefficients for vocabulary score and paternal NART and Quick IQ are moderate in size.

c. Illusions

If all participants that failed a control task are excluded, then there are 7 father and child pairs remaining.

Diagram 48 Fathers and their children succumbing to the Ponzo illusion

Ponzo illusion		Fathers	
		Did not see	Saw illusion
Young people	Did not see	2	0
	Saw illusion	0	5

The relationship between whether parent and child saw the illusions is perfect. That is, in every case where the child saw the illusion the father did so too and vice versa.
Fisher's Exact test for this cross tabulation gives a p value of 0.048 (2 sided)
This is not the case for the other two illusions.

Diagram 49 Fathers and their children succumbing to the Hat illusion

Hat illusion		Fathers	
		Did not see	Saw illusion
Young people	Did not see	1	3
	Saw illusion	1	2

Fisher's Exact test p value=1.00

Diagram 50 Fathers and their children succumbing to the Muller-Lyer illusion

Muller-Lyer illusion		Fathers	
		Did not see	Saw illusion
Young people	Did not see	2	1
	Saw illusion	1	3

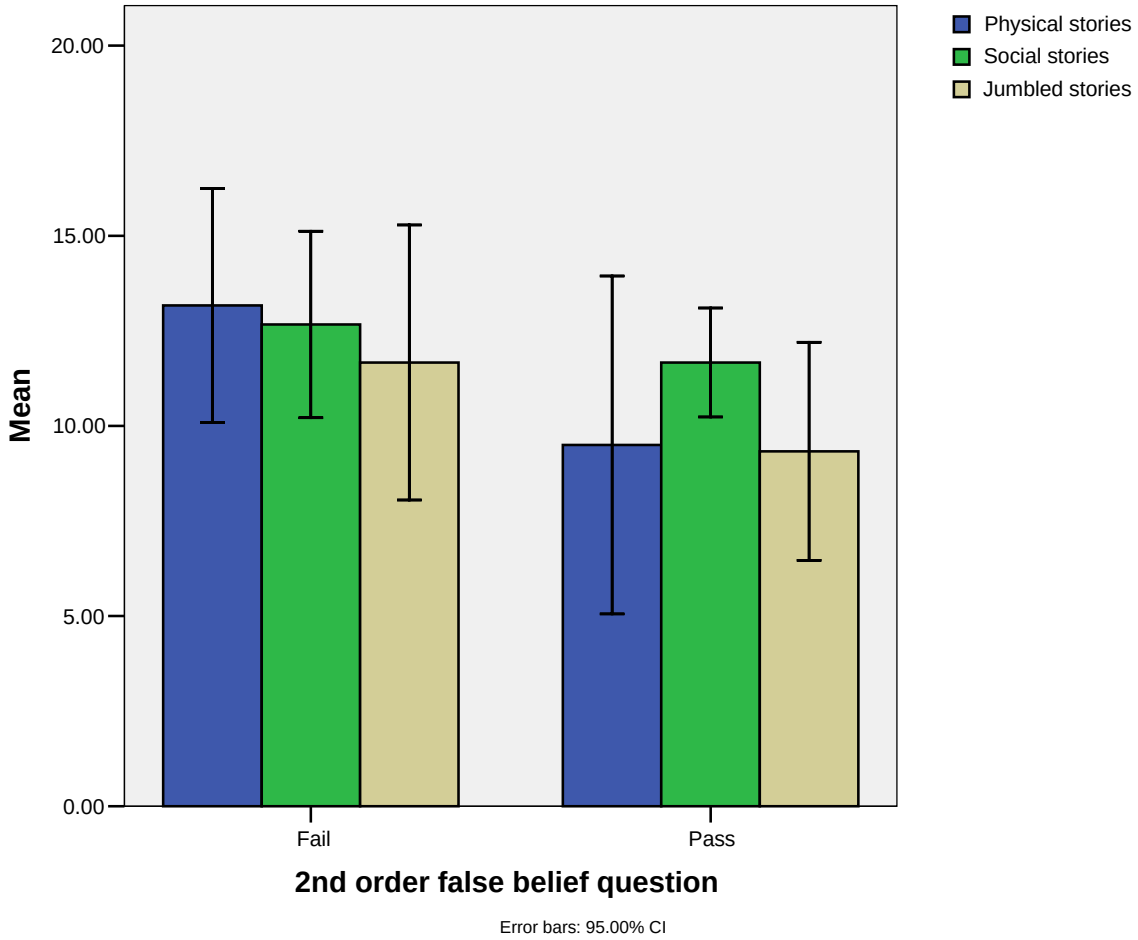
Fisher's Exact test p value =0.49

With all cases included none of the cross tabulations are significant.

d. Theory of Mind

A one way ANOVA of father's score on the social stories factored by child's Theory of Mind score revealed no relationship between the two measures. For the social stories F=1.85, p=0.21. For the physical stories F=3.25, p=0.09 and for the jumbled passages F=2.36 p=0.15.

Diagram 51 Father's Mean Strange Story Score by Child False Belief Score.



The graph shows that although there are no statistically significant differences there is a trend for the fathers of those who pass the false belief question to do better on the social than the physical stories and the opposite trend in the fathers of those who fail the first order false belief question. This trend is not statistically significant.

e. Ambiguous figures

There is no correlation between the number of reversals per minute seen by the child and the number of reversals of the cat/swan seen by the father: Pearson's correlation coefficient =0.16 p=0.65 n=11. Correlation between child reversals on cat/swan and

father's reversals on the Necker cube: Pearson's correlation coefficient r=-0.19, p=0.59 n=10.

2. Mothers

We next examine whether there is any relationship between the performance of mothers and their child on these measures.

a. Block design test

The correlation between mother BDDIFF and child BDDIFF is 0.28 p=0.17 n=25. There is no relationship between the difference in block design conditions between mother and child.
The correlation between the number of designs completed in the unsegmented condition between mother and child is 0.37 p=0.07, n=26. This approaches significance.

b. IQ

Diagram 52 Correlations Between Mother and Child IQ Measures

		Mother NART IQ	Mother Quick IQ
Child vocabulary scaled score	Correlation	0.24	0.28
	p value	0.27	0.19
	N	24	24
Child digit span scaled score	Correlation	-0.11	0.09
	p value	0.62	0.69
	N	25	25

Again, there is not a significant relationship between child IQ and mother IQ measures. However, the correlations for vocabulary score are moderate in size.

c. Illusions

There was no relationship between mother and child in susceptibility to the illusions. This was the case for the individual illusions and the illusions combined. It was also the case whether or not the participants who failed the control tasks were included.

d. Ambiguous figures

There were no significant relationships between the number of reversals per minute of the cat/swan seen by the young people and their mothers. Pearson's r= -0.10, p=0.64, n=26.
Nor with the reversals of the cat/swan seen by the child and the reversals of the Necker cube seen by the mothers: Pearson's r=0.2, p=0.28, n=21.

e. Theory of Mind

There is no relationship between the Strange Stories scores of mothers and the false belief task performance of their child. Comparing the mean scores of the mothers of young people who passed and failed the false belief question by ANOVA, for the mentalising stories F=0.07, p=0.80, for the physical stories F=0.95, p=0.34 and for the jumbled passages F=1.11 p=0.30.

In summary, there is little evidence for a relationship between parent and child scores on these measures. The exception is the relationship between fathers and their children resisting the Ponzo illusions. All other correlations are explained by a general effect of IQ. These relationships based on IQ approach significance for the father's IQ scores and the child's vocabulary scaled score, and in the mothers for the number of unsegmented designs completed by mother and child.

Relationship Between Variables

In the parents as a whole resisting the Ponzo illusion was associated with not seeing any reversals of the cat swan.

Diagram 53 Relationship Between Seeing Reversals of the Cat/Swan and the Ponzo Illusion

	Saw Ponzo illusion	Did not see illusion
Saw reversals of cat/swan	16	0
Did not see reversals	14	5

Fisher's Exact test p=0.049

Parents who saw no reversals of the cat/swan had significantly lower BDDIFF scores (Mean BDDIFF=13.07s, s.d.=7.31 n=20) than parents who did not see reversals(mean BDDIFF=18.32s, s.d.=7.93) independent samples t test t_{34}=-2.06, p=0.047.

There was no relationship between seeing reversals of the cat/swan and Strange Story score.

Correlation matrices for the metric variables are shown separately for mothers and fathers.

1. Fathers

Diagram 54 Correlation Matrix - Fathers

	NART IQ	Quick IQ	Physical stories	Social stories	Reversals cat/swan	Reversals Necker	BDDIFF	Child SCQ
NART IQ	1	0.73* 0.01 11	0.62* 0.04 11	0.04 0.90 11	0.05 0.90 10	0.33 0.39 9	-0.18 0.58 11	0.55 0.08 11
Quick IQ	0.73* 0.01 11	1	0.70* 0.01 12	0.63* 0.03 12	-0.67* 0.03 11	0.33 0.36 10	-0.20 0.53 12	0.02 0.94 12
Physical stories	0.62* 0.04 11	0.70* 0.01 12	1	0.31 0.33 12	-0.27 0.43 11	-0.13 0.72 10	-0.37 0.24 12	0.28 0.38 12
Social stories	0.04 0.90 10	0.63* 0.03 12	0.31 0.33 12	1	-0.35 0.29 11	0.14 0.70 10	<-0.01 1.00 12	<0.01 0.99 12
Revs cat/ swan	0.05 0.90 10	-0.67* 0.03 11	-0.27 0.43 11	-0.35 0.29 11	1	-0.19 0.59 10	0.17 0.62 11	0.30 0.37 11
Revs Necker	0.33 0.39 9	0.33 0.36 10	-0.13 0.72 10	0.14 0.70 10	-0.19 0.59 10	1	-0.23 0.53 10	0.52 0.13 10
BDDIFF	-0.18 0.59 11	-0.20 0.53 12	-0.37 0.24 12	-0.01 1.00 12	0.17 0.62 11	-0.23 0.53 10	1	-0.11 0.73 12
Child	0.55	0.02	0.28	<0.01	0.30	0.52	-0.11	1

SCQ	0.08 11	0.94 12	0.38 12	0.99 12	0.37 11	0.13 10	0.73 12	

The two measures of IQ, NART IQ and Quick IQ are highly correlated. This general factor of intelligence seems to be the source of an association with the physical stories score, which is correlated with both NART IQ and Quick IQ. The social story score is only correlated with Quick IQ. BDDIFF is not correlated with IQ.
Quick IQ is correlated with reversals of the cat/swan. It appears that fathers with lower IQs saw more reversals of the figure and vice versa.

Illusions and IQ

Fathers who failed at least one control task for the visual illusions had a mean IQ of 79.5 (n=2) and those who passed had a mean IQ of 103.6 (n=10). This difference in means was statistically significant in an independent samples t test t_{10}=-3.45, p=0.01. Comparisons of the mean IQ of those who did and did not succumb to the illusions revealed no differences.

2. Mothers

Diagram 55 Correlation Matrix - Mothers

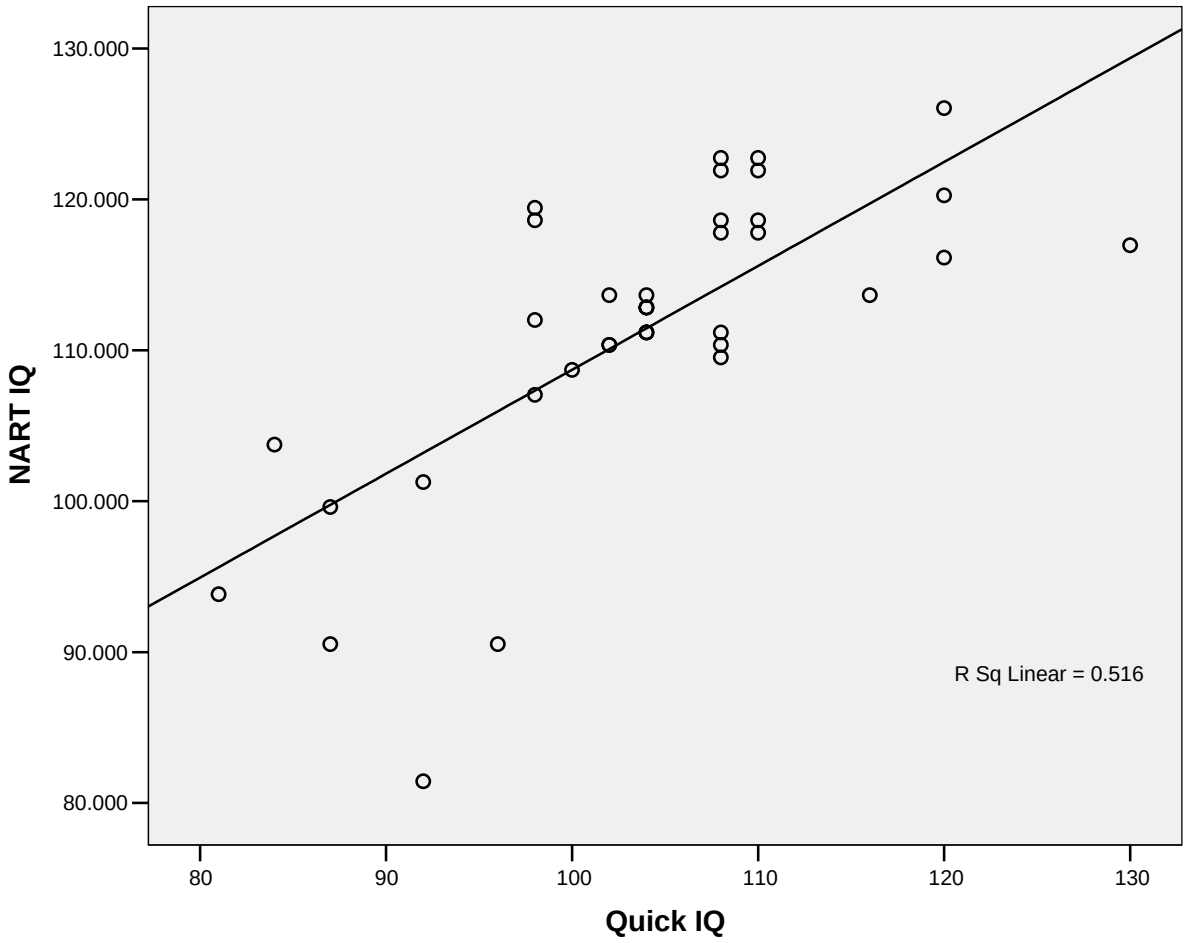
	NART IQ	Quick IQ	Physical stories	Social stories	Revs cat/swan	Revs Necker	BDDIFF	Child SCQ
NART IQ	1	0.75* <0.01 25	0.45* 0.03 25	0.11 0.62 25	-0.01 0.97 25	0.02 0.95 20	0.18 0.39 25	0.16 0.44 25
Quick IQ	0.75* <0.01 25	1	0.43* 0.03 25	0.06 0.77 25	0.28 0.17 25	-0.11 0.64 20	0.13 0.53 25	-0.06 0.76 25
Physical stories	0.45* 0.03 25	0.43* 0.03 25	1	0.33 0.11 25	0.26 0.21 26	0.13 0.57 21	-0.17 0.42 25	-0.13 0.53 26
Social stories	0.11 0.62 25	0.06 0.77 25	0.33 0.11 25	1	0.37 0.07 25	-0.30 0.21 20	-0.14 0.51 25	0.22 0.28 25
Revs cat/ swan	-0.01 0.97 25	0.28 0.17 25	0.26 0.21 26	0.37 0.07 25	1	-0.21 0.37 21	0.17 0.43 25	-0.35 0.08 26
Revs Necker	0.02 0.95 20	-0.11 0.64 20	0.13 0.57 21	-0.30 0.21 26	-0.21 0.37 21	1	0.05 0.85 20	-0.14 0.56 21
BDDIFF	0.18 0.39 25	0.13 0.53 25	-0.17 0.42 25	-0.14 0.51 25	0.17 0.43 25	0.05 0.85 20	1	-0.27 0.19 25
Child SCQ	0.16 0.44 25	-0.06 0.76 25	-0.13 0.53 26	0.22 0.28 25	-0.35 0.08 26	-0.14 0.56 21	-0.27 0.19 25	1

In the mothers, the only strong relationships are between the measures of IQ and the physical stories score. There is a relationship between the number of reversals of the cat/swan seen and social story score which approaches significance p=0.07. No such relationship was present for the fathers.

Relationship Between Measures of IQ in the Whole Sample

As would be expected, the Quick IQ and NART IQ scored were highly correlated. Pearson's r=0.72. The scatter plot below shows the relationship between the two variables.

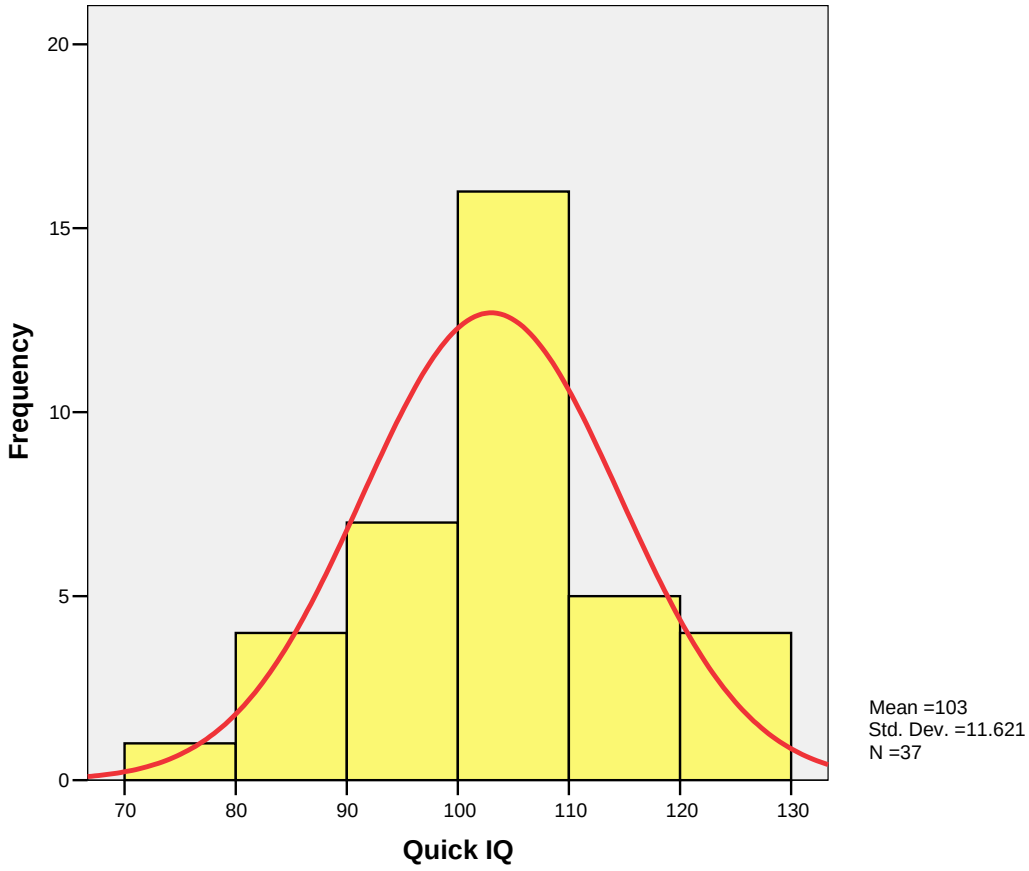
Diagram 56 Scatter Plot of NART IQ with Quick IQ



There would seem to be a tendency for the two measures to have less agreement at the lower IQ ranges and to agree better at higher values. In fact, a quadratic equation is a marginally improved model for the relationship over the linear, the r squared quadratic being 0.54.

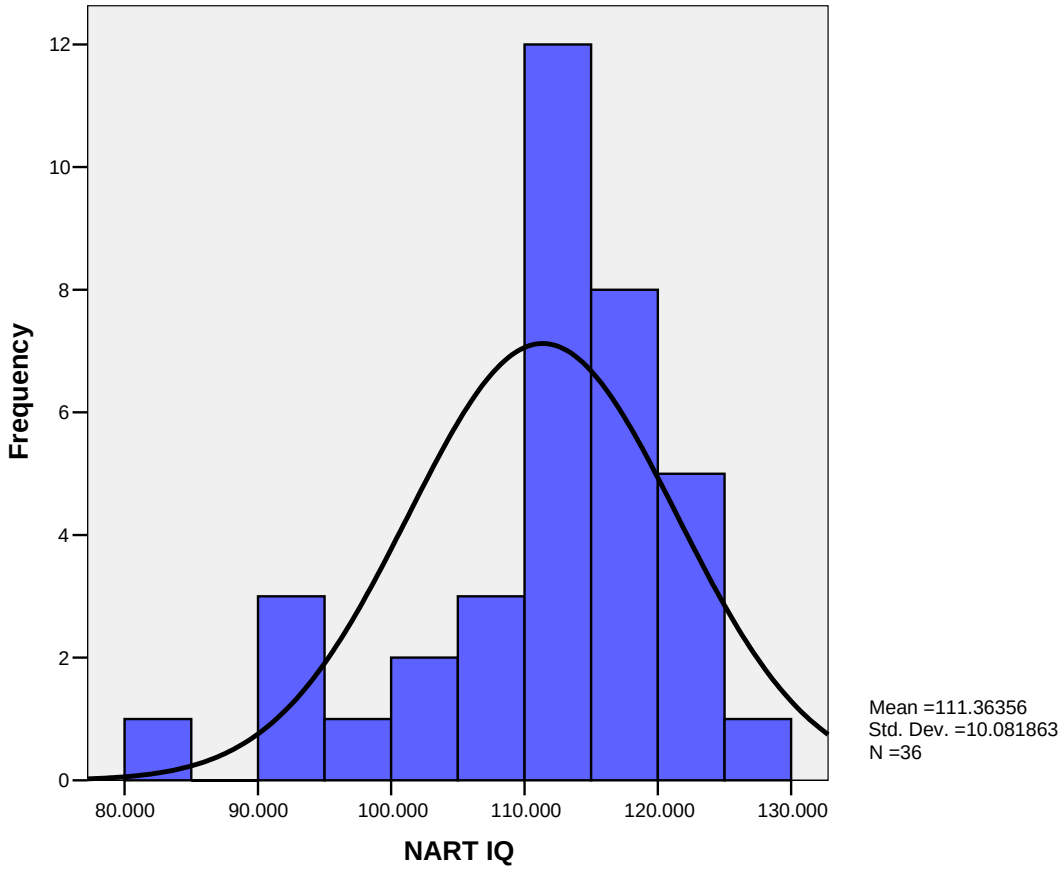
Examination of the distribution of the two variables in this sample shows that Quick IQ is normally distributed

Diagram 57 Histogram of Quick IQ in Whole Sample



whereas the distribution of NART IQ scores is negatively skewed.

Diagram 58 Histogram of NART IQ in Whole Sample



This means that NART IQ has more extreme low values that reduce the mean score relative to the median score.

The mean Quick IQ in our sample is 103.86 and the mean NART IQ 111.36. The mean difference between the IQ measures in our sample is 7.5 points s.d.=7.74. There was a trend for fathers to have greater differences mean difference for fathers =9.01 s.d.=8.97. Mean difference for mothers=6.84 s.d.=7.23.

What is unexpected is that whether the parents are split at the 15 or the 22 cut off on the SCQ, parents of higher SCQ children have much greater discrepancies between their NART IQ and Quick IQ estimates than the other parents. Parents of high SCQ have much higher NART IQ than Quick IQ estimates.

Diagram 59 Mean Difference Between IQ Estimates by Group.

	Parents of SCQ≥22 (n=9)	Parents of SCQ<22 (n=27)
NART IQ-Quick IQ	12.63 (7.48)	5.79 (7.16)

Anova gives F=6.02, p=0.02.

Conclusions

We found that parents of young people with SCQs greater than 22 were less likely to succumb to the Ponzo illusion and less likely to see any reversals of the cat/swan ambiguous figure than other parents once IQ had been statistically controlled. We found no evidence of social cognitive deficits in parents of young people with autistic traits. There was a trend for parents of people with high SCQs to gain less benefit from the segmentation of the block designs but this was not statistically significant. In parents of those with high SCQ scores the difference in times between block design conditions was correlated with IQ where as in the other parents it was not. In parents of high SCQ people NART IQ accounted for about 38% of the variance in block design difference. In parents of people with low SCQs NART IQ accounted for 1% of the variance in block design difference.

We found that fathers and their children share the tendency to resist the Ponzo illusion. Fathers who succumbed to the illusion had children who did likewise and vice versa. On none other of the measures was there any relationship between parent and child score except for an effect of general intellectual ability.

Discussion - Parents

Two of the measures used are sensitive to differences in cognition between parents of young people with high levels of autistic traits and the parents of those with low. These measures are:

1. Ponzo illusion when Quick IQ is controlled: and
2. whether any reversals of the cat/swan are seen when NART IQ is controlled.

There were no significant differences between the parents of people with high autistic traits and the other parents on any of the other measures. We will discuss the results for each of the measures used, with reference to our original hypotheses and other research in this area.

Strange Stories

The mean scores for the social stories were slightly higher than the physical stories across the whole sample. There were no differences between the mean scores of the two parent groups on the mentalising/social stories, physical stories or jumbled passages. On neither the mentalising stories, the physical stories nor the jumbled passages were the participants at ceiling. On the social stories, four out of 37 parents scored the maximum of 16. On the physical stories, 2 parents scored the maximum of 16. We must therefore conclude that there was no evidence of even very subtle social cognitive deficits in the parents of the young people with high levels of autistic traits.
Our sample is particularly unusual because it is parents of young people identified by their level of autistic traits as opposed to a diagnosis of the disorder. Thus, we would expect any effects to be smaller than found in other research on the broader phenotype

and any negative findings could be the result of the dilution of the phenotype in our sample. This is one explanation for our negative results.

The other explanation is, of course, that deficits in Theory of Mind do not form part of the broader cognitive phenotype. In other research in this area, there have been findings of no deficits on measures of social cognition in relatives of people diagnosed with ASD. For example, Ozonoff et al. (1993) found no differences between the performance of siblings of children with autistic disorder or PDD and the siblings of children with learning disability on tests of Theory of Mind. The tests used, second order false belief and two perspective-taking tasks, may not have been ideal for this subject group, which had a small sample size and large variation in participants' ages. A very recent study by Shaked, Gamliel and Yirmiya (2006) which examined the Theory of Mind abilities of four-year-old siblings of people with autism also found no Theory of Mind deficits in the siblings.

Where research has found significant differences in social cognitive ability between the relatives of people with autism and control groups, it has been on The Mind in the Eyes task. Baron-Cohen and Hammer (1997) found that parents of people with Asperger disorder performed more poorly than parents of controls, on the Mind in the Eyes task. Dorris et al. (2004) also found that siblings of people with Asperger disorder performed more poorly on the children's version of this task than the siblings of typically developing children. This difference is isolated to identifying emotion from the eye region of the face, as in tests of facial recognition, emotion-matching and emotion labelling tasks no differences have been found between relatives of people with autism and controls (Smalley and Asarnow, 1990; Szatmari et al., 1993). Thus, it seems that if there are any differences between the relatives of people with ASD and controls in social cognition these are very subtle and can only be detected with some advanced tests. It should be noted that the two studies that found differences on the Eyes test used probands with Asperger disorder. Matching control groups is less complicated for Asperger disorder, as by definition probands have no clinically significant language delay or intellectual disability and in both the above studies, they were matched with a

typically developing control group that may be more homogenous than relatives of people with other disorders.

Based on our results, we cannot reject the null hypothesis that there is no difference in social cognition between the parents of people with high levels of autistic traits and those with low.

It cannot be taken as confirmation of our hypotheses, but may be useful for further development of the materials to examine performance on individual stories.

Among the social stories, the parents of young people with high levels of autistic traits performed significantly differently to the parents of the other young people on one of the stories. This was:

> Simon is a big liar. Simon's brother Jim knows this; he knows that Simon never tells the truth! Now yesterday Simon stole Jim's ping-pong paddle, and Jim knows Simon has hidden it somewhere, though he can't find it. He's very cross. So he finds Simon and he says, "Where is my ping-pong paddle? You must have hidden it either in the cupboard or under the bed, because I've looked everywhere else. Where is it, in the cupboard or under your bed?" Simon tells him the paddle is under his bed.

The question associated with this story is 'Why will Jim look in the cupboard for the paddle?'

The parents of children with high levels of autistic traits in fact did significantly better on this question than the other parents. Participants could get full points (2) on this question by referring to the fact that Simon is a liar. This is the first line of the story and most participants echoed this as an answer. 'Because Simon is a liar' was a common response. 28 out of 37 parents used the word liar or a close approximation (e.g., he never tells the truth) in their answer. All the parents in the group 1 (high SCQ offspring) scored 2 on this question. It could be queried whether this is a good measure of Theory of Mind when the correct answer can be directly taken from the information given.

The physical story that all the participants found most difficult was the story:

> Mrs. Simpson, the librarian, receives a special book which she has to catalogue and find an appropriate place for. She has to decide which section to file it under. The library is very big, and has different sections on many different subjects. The new book is about plants and their medical uses, and is heavily illustrated. However, Mrs. Simpson does not put it on the shelf with the rest of the books on botany. Neither does she put it with the books on medicine. Instead, she carefully takes it into a separate room. In this room all the books are kept in special cases, and the temperature is kept constant.
>
> The question was 'Why did she do this?'

Nine out of the total 37 parents scored zero on this question. This question was possibly reliant upon the amount of experience the participants had with libraries and books. Also, the reference in this question is rather oblique and it requires a greater degree of inference than some of the other stories in order to find the correct answer. Both of these factors may have contributed to the lower score of the group 1 parents. In the group 1 parents, with high SCQ offspring, 7 out of 20 scored zero and in the other group only 2 out of 17 scored zero.

However for the following question the parents of the young people with a high level of autistic traits did much better than the other participants:

> Old Mrs. Robinson is very frail. One day she slips on her icy doorstep and falls on her side. She gets up right away, although she feels quite bruised and shaken. The next day her leg feels very stiff and she can scarcely walk. She makes her way to the doctors. As soon as the doctor hears about the fall, and sees her swollen side, he says, "Go immediately to the hospital". At the hospital, they take an X-ray.
>
> The question was 'Why did they take an X ray?

This question seemed to be more within the general experience of the participants. Parents of young people with high autistic traits did much better on this question than those of young people without autistic traits.

Among the Strange Stories as a whole, the physical stories score was highly correlated with both the Quick and NART IQ measures. This is support for the use of the physical stories as a measure of general ability and as a baseline against which to compare the social story performance. Social story score was not correlated with the IQ measures.

There was no relationship between parent and child performance on the social cognitive measures. This supports the conclusion that social cognitive abilities are not highly heritable in this population. However, it must be noted that the false belief questions are both dichotomous variables and hence the scores have low variance, which limits the power to detect relationships between the false belief scores and Strange Stories scores should they exist. It should also be noted, however, that failing the second order false belief test in our sample of young people, who are all over 13 years of age, indicates a high degree of impairment of Theory of Mind abilities. If this is not associated with any impairment of Theory of Mind abilities in the parents, as we have found, then it does support the conclusion that Theory of Mind abilities are not highly heritable.

Visual illusions

In general, the parents were more susceptible to the illusions than the young people. On the Hat illusion, only 11% of parents resisted the illusion whereas 40% of the young people resisted the illusion. On the Muller-Lyer illusion, 17% of parents resisted and 20% of the young people, which is roughly equivalent. On the Ponzo illusion, 14% of parents resisted and 27% of the young people. For all three illusions, a larger proportion of the young people did not see the illusions than the parents. This was clearly so for the Hat and Ponzo illusions but the difference between parents and young people was smaller for the Muller-Lyer illusion.

Research in this area suggests that these illusions have different developmental trajectories. Susceptibility to the Muller-Lyer illusion is thought to decrease with age (Comalli 1965) . In our subjects this was not the case, the parents being slightly more

susceptible than the young people. Susceptibility to the Ponzo illusion is thought to increase with age and then level off in adulthood (Leibowitz and Gwozdecki 1967) . This is in line with our results. More of the parents succumbed to this illusion than the young people. Susceptibility to the Hat illusion increases with age toward adulthood (Hanley and Zerbolio 1965) and this is again what we have found.

One of the illusions, the Ponzo illusion, was a significant variable in a logistic regression equation to predict group membership separating parents of people with high autistic traits from parents of people with low autistic traits. It only reached significance as a predictor once Quick IQ or NART IQ had been entered into the equation. As previously discussed, there is a developmental progression in susceptibility to illusions. (Leibowitz and Gwozdecki 1967) found that susceptibility to illusions was a function of mental age in adults with intellectual disability and that overall susceptibility was greater in this group than in the general population. It does suggest, perhaps counterintuitively, that susceptibility will be a function of IQ.

It is significant that the only illusion on which we can find any difference between the performances of the two groups of parents is the same illusion that was significant in discriminating the young people with high autistic traits. This supports the conclusion that, firstly, there are different mechanisms operating across the illusions and people with high levels of autistic traits and their relatives will only be immune to certain illusions. This may be due to the illusory strength of the stimulus, or its perceptual coherence or the mechanism by which the illusion works e.g. size constancy. Secondly, that these effects will be consistent across samples once IQ is controlled.

On the illusion control tasks, a smaller proportion of the parents failed the control tasks than the young people. On the control task for the Muller Lyer illusion, 11% of parents failed and 22% of young people. That is, 11% of the parents said that the control lines for the Muller-Lyer illusion were different in length when in fact they were the same length. For the Ponzo illusion, 3% of parents and 12% of young people failed the control task. For the Hat illusion, 22% of parents failed the control task and 37% of young people.

It is notable that among the fathers, those who failed one illusion control task (n=2) had significantly lower IQs than those who did not fail any. It is not clear why IQ should affect performance on the illusion control task. The wording of the control question was very simple. The participants were asked if the two lines were the same or different lengths. Nevertheless, those fathers that answered 'different' had substantially lower IQs than those who answered correctly. This may be because people with low IQs are more susceptible to the very subtle illusions that may operate even on the control tasks as suggested in the earlier discussion. Alternatively, it may have been a misunderstanding of the task. It does suggest that for either of these reasons, these simple length judgments may be less reliable in people with low IQ scores and this may be why the Quick IQ was a necessary variable in the logistic regression equation.

Ambiguous Figures

The parents who participated viewed two ambiguous figures. The cat/ swan, as used with the young people and the Necker cube. There was no difference in the mean number of reversals of the ambiguous figures seen by the two groups. However, when 'whether or not any reversals of the cat/swan were seen' as a dichotomous variable and NART IQ were both included in a logistic regression equation this significantly improved prediction of group membership over a constant only model. Also, when mothers and fathers were considered separately there was a significant difference between the mean number of reversals of the cat/swan seen by the mothers of young people with high levels of autistic traits compared to the other mothers. Mothers of young people with high levels of autistic traits saw fewer reversals of the cat/swan. This parallels the finding in the young people that those with high levels of autistic traits saw fewer reversals of the cat/swan. This was only the case for the cat/swan ambiguous figure. There were no differences between the parent groups in the number of reversal of the Necker cube seen. The correlation between the number of reversals of the cat/swan and of the Necker cube by each participant was actually negative $r=-0.20$, $p=0.27$. This may be because, although they are both reversible figures, they are

different types. The cat/swan ambiguous figure requires a representational change. The stimulus can be either a cat or a swan, each with their separate properties and associations. To see the reversals the subject has to perceive the figure to be a cat and then to be a swan. The Necker cube remains a cube under both interpretations. The aspect of the cube merely switches. This may be why subjects are more startled by the reversals of the Necker cube. It appears to flip or move because it is perceived as the same cube that moves to a different presentation. The mind identifies the cube as a constant therefore, movement is perceived. For the cat/swan, the subject does not perceive movement but does perceive that the stimulus has two distinct identities. Some subjects were aware that which version of the cat/swan they perceived, depended on where they fixated. It may be that because the cat/swan is 'harder work' it may be more sensitive to subject's response to task demands. Alternatively, it may be that the representational nature of the cat/swan figure means it taps individual variability in representational flexibility and this distinguishes our groups.

The pattern of association among the variables was different for mothers and fathers. In fathers, the number of reversals of the cat swan was significantly negatively correlated with IQ. Fathers with higher IQs saw fewer reversals of the figure. Fathers did show a relationship between social story score and IQ. Mothers did not show a relationship between social story score and IQ and neither was this relationship present within the parents as a whole. However, in mothers, although there was no association between social story score and IQ, but there was a relationship between social story score and the number of reversals of the cat/swan seen, $r=0.37$ $p=0.07$. This may then support the idea that the numbers of reversals of the cat/swan seen is an advanced test of Theory of Mind, in that it taps a participant's willingness to identify and comply with the aims of testing. However, it could also be an indicator of a common representational mechanism that is required both for the appreciation of multifaceted social interactions and for perceiving representationally ambiguous figures.

The fact that fathers have a greater correlation between social story score and IQ may indicate that they use a more sequential, systematic approach to solving these problems

and have less access to intuitive mechanisms for their resolution. In mothers, solving these problems may have greater reliance on representational flexibility.

As we found in the young people, a large proportion of the parents saw no reversals of the ambiguous figures at all. 62% of the parents of young people with high SCQ scores saw no reversals of the cat/swan and 50% of the parents of young people with low SCQ scores saw no reversals.

The proportion of parents seeing no reversals of the Necker cube was smaller for both groups. 21% of parents in group 1 (high SCQ) saw no reversals and 17% of parents in group 2.

Block design test

The parents of young people with high levels of autistic traits (SCQ≥22) gained less benefit from the segmentation of the block designs (mean difference between conditions = 12.49s) than the parents of young people with a low level of autistic traits (mean difference between conditions = 16.09s). This difference in means was not statistically significant.

As with the young people, we found that across high and low SCQ groups a large proportion (37%) of the parents were unable to complete all the designs in the unsegmented condition. Only one parent was not able to complete all the designs in the segmented condition. This suggests that a large proportion of parents in both groups benefited from the segmentation of the designs.

A number of studies have found non-significant results using the block design test with relatives of people with autistic disorder, for example, Smalley and Asarnow (1990), Szatmari et al. (1993), Fombonne et al. (1997) and Piven and Palmer (1997). In Smalley and Asarnow's study, they compared the parents (n=15) and siblings (n=9) of 9 nine males with autism to the parents (n=12) and siblings (n=9) of 9 controls. The probands were matched to the controls using the block design subtest so this may have reduced

variability in the relatives. Both siblings and parents of the people with autism were better at the block design test than the relatives of controls, but this was not statistically significant. All these studies used the block design test as in the standard intelligence tests, not in segmented and unsegmented versions as we have here. This makes these analyses vulnerable to the effects of imperfect IQ-matching between parents. The advantage of using the block design test in segmented and unsegmented form is that it serves as its own control for general visuospatial/constructional ability. The effect of the cohesiveness of the whole design can be isolated by subtracting the time taken to complete the design when the coherence of the whole is disrupted, that is in the segmented condition. This is confirmed by our finding that in the parents as a whole the difference in performance between the two block design conditions was not correlated with either of the measures of IQ. Happé et al (2001) used the block design test in segmented and unsegmented forms in parents of people with autism and found that the fathers of people with autism were faster than the fathers of typically developing and dyslexic boys. Our study had fewer fathers than that of Happé et al. (2001), which may have resulted in low power to detect small effect sizes in the fathers alone.

We did not find a statistically significant difference in the benefit the groups gained from the segmentation of the designs. We did find that in the parents of the young people with high SCQs the BDDIFF (difference between block design conditions) was highly correlated with IQ whereas in the rest of the sample it was not.

Our theory, based on the results obtained from the young people, is that young people with autistic traits cannot maintain two interpretations of a single stimulus and/or have rigidity in switching between interpretations (e.g. from a cat to a swan). We have suggested that this trait could influence performance in a number of cognitive domains including on the block design test. We have stated that this rigidity in switching between interpretations will mean that when viewing a complex geometric pattern (with 2+ possible interpretations) subjects with this trait will fixate on one interpretation. If this happens to be at the local level this will lead to success on the block design task, if the global level is chosen these subjects will be unable to switch to a segmented version and will be unable to complete the task. Early in development, the level of processing

fixated on would be arbitrary for each design. We predict that some high functioning people with this trait would establish a strategy where they looked for local constituent elements using object recognition strategies and achieved a high level of performance on this task so that at later stages the level would be no longer arbitrary.

In young people with no autistic traits, they will show the typical global precedence. The designs they succeed upon will be those with a low level of coherence. Their success on more coherent designs will depend upon their level of cognitive flexibility to seek alternatives to the Gestalt figure. In the young people as a whole, there was a trend for those who saw more reversals to have lower BDDIFF scores.

In the parents as a whole, this theory would predict that parents of young people with high SCQ scores may be more likely to also show this rigidity in switching between interpretations. They would be more likely to have rigid perception and to fixate on one level. On the block design test we found that in parents of young people with high autistic traits, BDDIFF was correlated with IQ whereas in the other parents it was not. This may be because in parents with more rigid perception success on this task will be associated with finding a strategy of object recognition for constituent elements. As we have suggested before this will be a function of IQ. People that are more able will find these strategies where less high functioning individuals will not. This would possibly explain the correlation between IQ and BDDIFF in this group.

Our theory would predict that in the parents as a whole high reversals would be associated with success on this task. However this is not the case. The only significant difference is that parents who saw no reversals of the cat/swan had significantly smaller differences between block design conditions than those who saw reversals. This is the reverse of the trend in the young people.

A possible explanation would be based on the fact that the number of reversals seen is significantly different between young people with high levels of autistic traits and those with low. Autistic traits were not measured directly in the parents. It may be that the broader phenotype in parents is more closely associated with not seeing reversals than with having a child with autistic traits. The most common element of the broader phenotype in relatives is rigidity operationalised as a lack of interest in seeking change.

Piven, Palmer, Landa et al. (1997) found this was present in 50% of parents of people with autism. We are asserting that a degree of rigidity is present in everyone with autism or the broader autism phenotype. Thus, it could be that reversals are a better measure of the broader phenotype than child autistic traits in this group. We have found that those parents who saw no reversals of the ambiguous figure have lower BDDIFF scores. They are also less likely to see the Ponzo illusion. Mothers who saw no reversals of the cat/swan have significantly lower social stories scores. We have discerned this same pattern in young people with high autistic traits who also see significantly fewer reversals of the ambiguous figure.

There are two points that need further exploration:

1. In the young people, it is the number of reversals per minute that differs between high and low SCQ groups. In the parents, it is whether any reversals were seen or not. This may be a maturational effect whereby the optimal measure is different in young adults with intellectual disabilities than in unaffected adults.

2. In the parents there is greater association between variables than in the young people. When the parents are divided into those who saw reversals and those who did not then these groups score significantly differently on many of the other measures. This is not the case in the young people where the number of reversals seen has little relationship with the other variables. Nevertheless, we would argue that the cognitive traits indexed by reversals of the figure are of particular significance in the etiology of autistic traits. We would suggest that these traits are normally distributed in the general population and that the slight rigidity of perception evidenced in the unaffected parents is very common in the normal population. As can be seen from the block design test this is an advantageous trait. It may confer the same benefits as have been suggested for people with Weak Central Coherence in that people with this trait will tend to adopt local processing strategies and favour detailed focused sequential information processing, which are advantageous characteristics in science and engineering. However in contrast to the predictions of Weak Central Coherence that says that local processing is the preferred mode we suggest that it is an adopted mode secondary either to an inability to maintain two versions of a stimulus or to rigidity in switching between

versions. Initially, early in development the level of processing is arbitrary in these cases. We have suggested that in higher functioning individuals they may later adopt a strategy of looking for the local features.

It may be that a particularly severe manifestation of this rigidity at the extreme of the normal continuum is enough to affect early development and produce the behavioural and academic problems experienced by the young people. Alternatively, it may be that some young people have a 'double hit' and it is these rigid traits plus other neurodevelopmental abnormalities in combination that produce autistic traits. Which ever of these explanations is the case we suggest that our results strongly support the use of ambiguous figures in further exploring cognition in autistic spectrum disorders and in the broader phenotype.

Conclusions

Even in the relatives of young people who have autistic traits and not necessarily a diagnosis of an ASD, it is possible to detect subtle parts of the broader autism phenotype. We have found clear differences between parents of young people with SCQ scores over 22 and the other parents. Parents of people with high levels of autistic traits see fewer reversals of an ambiguous figure when IQ is statistically controlled. We also found that the Ponzo illusion was useful in distinguishing the groups, in both the young people and their parents when IQ was controlled, suggesting there is something in particular about the presentation of this illusion here that can detect elements of this phenotype. We would expect that the genetic loading in our sample would be low given the fact that the young people do not all have the full expression of the disorder. Given this, it is remarkable that we have some clear indications of distinctions between the parent groups. We suggest that these distinctions stem from a perceptual rigidity that is common and normally distributed in the population but more prevalent in the relatives of people with ASD. High levels of this trait in the normal population cause superior block

design performance, resistance to some visual illusions and in females, lower performance on social cognitive measures.

Diagnosis

As with the results for the young people, it is possible that the differences between the parent groups are due to a small number of probands who have a diagnosis of autism or Asperger disorder. We divided the parents into those with a child with a diagnosis of autism and those without. We then examined their performance on the measures to see if parents of children with a diagnosis were responsible for the differences found between high and low SCQ groups.

The only significant difference was that the parents of young people with a diagnosis had *higher* social story scores than the other parents. The Muller-Lyer illusion approached significance with parents of people with a diagnosis of autism or Asperger disorder less likely to succumb to this illusion. The parents of those with a diagnosis of autism or Asperger disorder do not have more extreme scores than the other parents of young people with high SCQ scores.

Relationship between tests of IQ.

The scores on the two IQ tests used, the NART (Nelson and Willison 1991) and Quick test revised (Mortimer and Bowen 1999) were, as would be expected, highly correlated. The correlation between the two scores r=0.72.

Five other studies have used both the NART IQ and Quick test revised to measure IQ in participants including unaffected controls. Four of these studies used the NART IQ and the Quick test in its original format. In all four studies the NART produced a higher IQ estimate than the Quick test in the healthy control groups (Frith, Leary et al. 1991; Warwick, Doody et al. 1999; McIntosh, Forrester et al. 2001; Badcock, Michie et al. 2002). The single study we are aware of that has used both the NART IQ and the Quick

test revised Kondel et al (2003) found the difference in healthy controls to be 3.53 (95%CI 0.87-6.2).

It can be seen from the studies outlined above that NART IQ consistently over estimates IQ relative to Quick IQ in control participants. The difference between mean NART and Quick IQ scores in these studies being a minimum of 3.5 points for healthy controls. Our study is therefore not untypical in finding a discrepancy between the Quick IQ revised and NART IQ estimates. The mean difference between the IQ estimates in our group of unaffected parents is 7.5 points.

We found also that parents of young people with high SCQ scores had significantly greater discrepancies between their NART and Quick IQ estimates than the other parents. A verbal-performance IQ discrepancy has been suggested to be part of the broader phenotype of autism. Research in this area has produced contradictory results. For example in line with our results Fombonne et al. (1997) found higher verbal abilities in parents and siblings of people with autism than controls. However, Piven and Palmer (1997) found decreased performance IQ in multiplex autism families relative to controls. Bailey et al. (1998) in their review of the literature conclude that these differences in findings may be due to ascertainment differences between studies.

Relationship between parent and child scores.

In general, there was very little evidence of a relationship between parent and child scores on any of our target measures. The exception to this being susceptibility to the Ponzo illusion in fathers and offspring. In this case the relationship was striking. For every father who resisted the illusion, his child also did so and vice versa. This would suggest a high degree of heritability for susceptibility to this illusion. However, we did not find any relationship on the difference between block design tests conditions (BDDIFF) in parents and children. Between mothers and their offspring, the correlation of the number of designs completed in the segmented condition approached significance. We have found that the number of designs completed did not relate to the difference between conditions. We conclude that this correlation between the number of designs

completed by mothers and their offspring is due to an effect of general intelligence rather than central coherence. Although not statistically significant, there were moderate correlations between the IQ measures for the parent and the vocabulary subscale scores of their offspring.

Relationship between variables

Briskman, Happé and Frith (2001) studied the real-life skills and preferences in the parents of boys with autism. They found that parents' self- report measures of social and non-social skills and preferences were correlated in the parents of people with autism, but not in the parents of the typically developing or dyslexia control groups. In our sample, where we directly measured ability in these domains in parents we found no relationship between measures of Theory of Mind and either of our measures of Weak Central Coherence. A recent study by Ronald et al (2006) supports this finding as they found that extreme traits in the triad of impairments were highly heritable but showed relatively little phenotypic and genetic overlap and that in the general population social impairments, communicative impairments and repetitive behaviours were only loosely related.

Final Conclusions

These studies have shown that the psychological characteristics of people with autism extend beyond the narrow definition of the disorder. People who show behavioural signs associated with autistic disorder, but who have not necessarily received a diagnosis of the disorder, can be differentiated from young people who do not show autistic-like behaviours on the same psychological measures that are sensitive to diagnosed autism. This indicates a spectrum of autistic conditions that vary by degree rather than kind, at least at the psychological level of explanation.

The parents of the young people with high levels of autistic traits show some of the same cognitive characteristics as the young people. In particular, they show resistance to the same illusion as the young people. The parents of the young people with high levels of autistic traits also saw fewer reversals of one of the ambiguous figures when IQ was statistically controlled. The same ambiguous figure that young people with high levels of autistic traits were less likely to see reverse.

There were no correlations between the performance of parent and child on the majority of these measures suggesting that although these measures may tap elements of the broader cognitive phenotype of autism they are inherited via a complex genetic mechanism not directly transmitted from parent to child. This lack of correlation between parent and child scores also argues against these being environmental effects: either from parent to child or child to parent. The exception to this is the relationship between fathers and offspring for succumbing to the Ponzo illusion where there may be a direct mechanism.

Our explanation for these results hinges on the ambiguous figure. This measures the ability to see a representationally bistable figure alternate from one interpretation to another. We suggest that this taps the ability to map two identities onto a single object and to manipulate these flexibly. We suggest that this feature can be found in many of the tasks that people with autism find most difficult. These are tasks where a stimulus has more than one possible interpretation and it is necessary to use knowledge or experience to choose between the interpretations.

In parents, we do not suggest that the ability to identify an object as having more than one identity is absent merely that there is hallmark rigidity in the manipulation of alternate representations. This may result in the tendency to perseverate on one interpretation of an ambiguous figure and to favour detail-focused information processing in open-ended tasks. This trait will be present only in a minority of parents of young people with autistic traits. This rigidity produces a tendency not to see reversals of the ambiguous figure, to resist the Ponzo illusion and means that a successful strategy on the block design test is more reliant on general intellectual ability than in other adults.

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Appendix

Block designs

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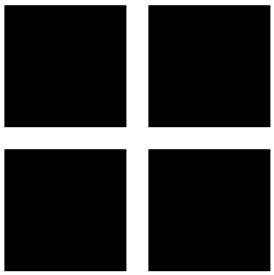

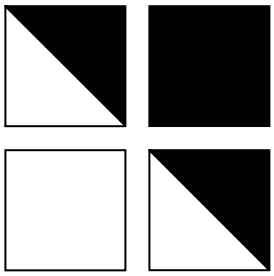

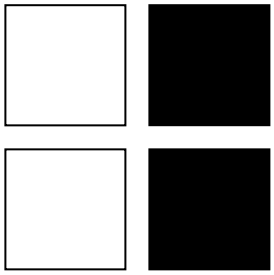

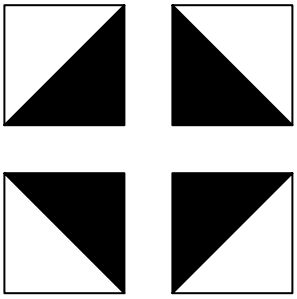

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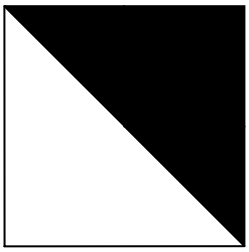
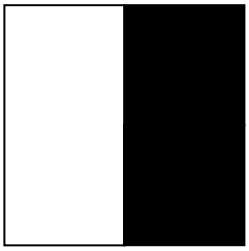
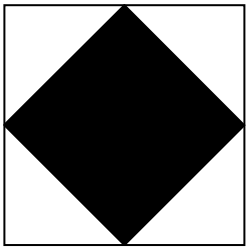
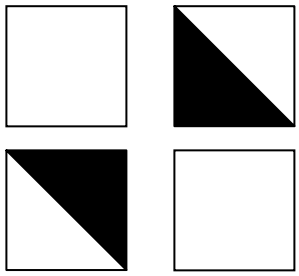
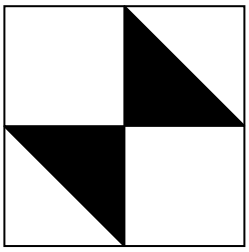
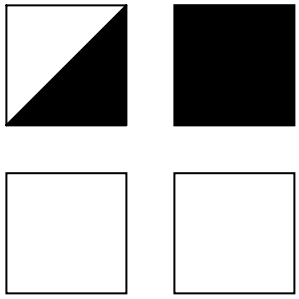


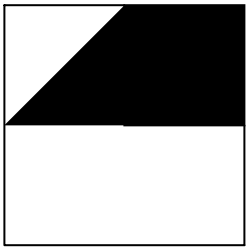


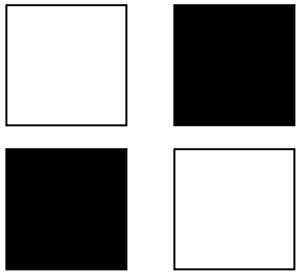


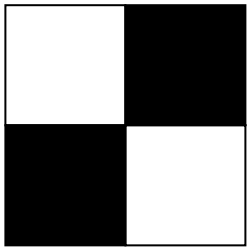


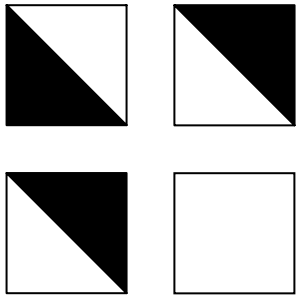


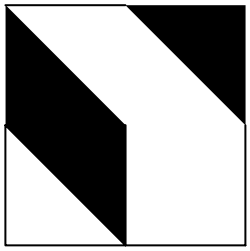


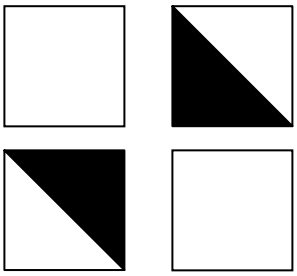


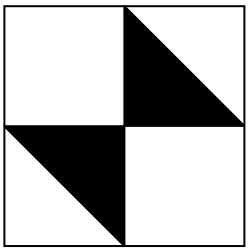




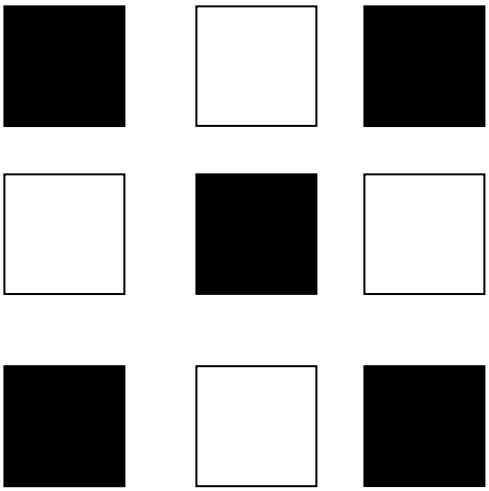


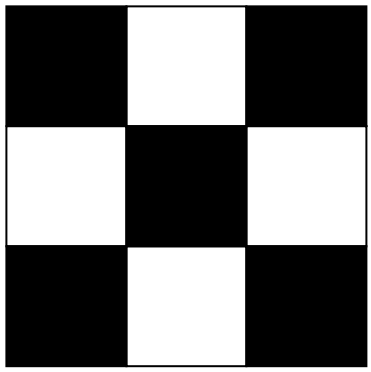


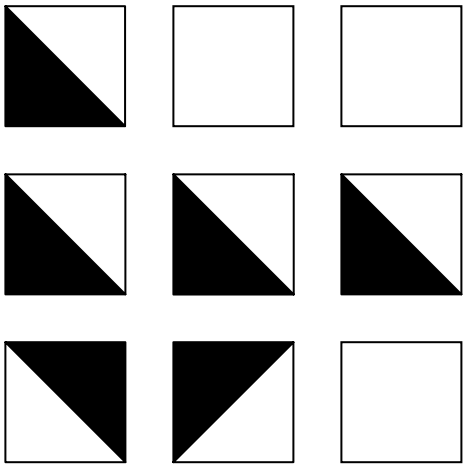


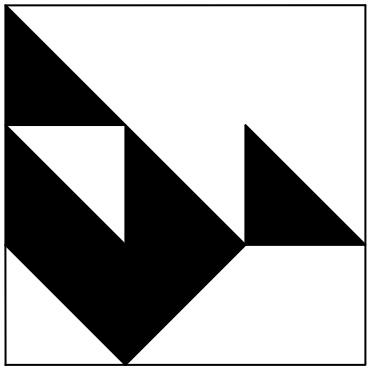


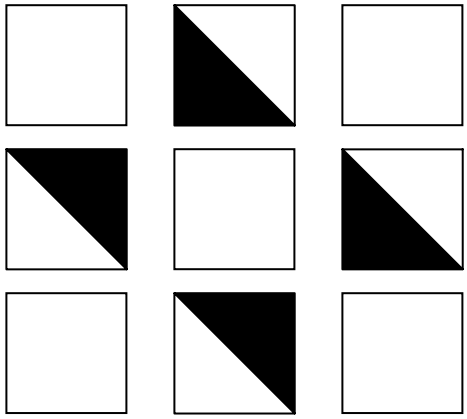
Nine Block designs for parents

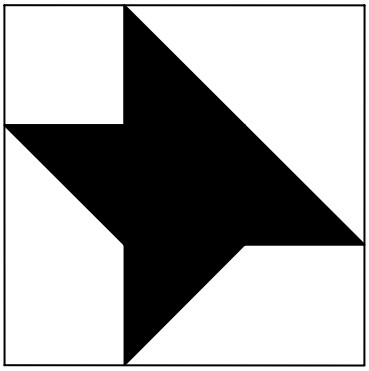


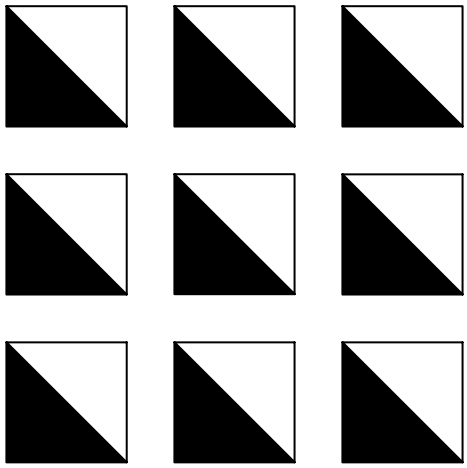


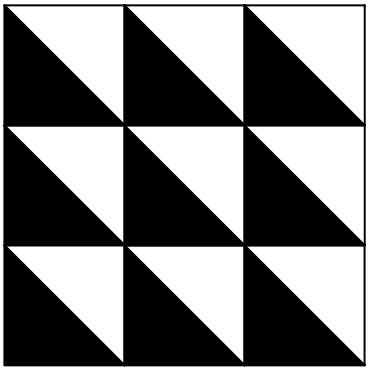


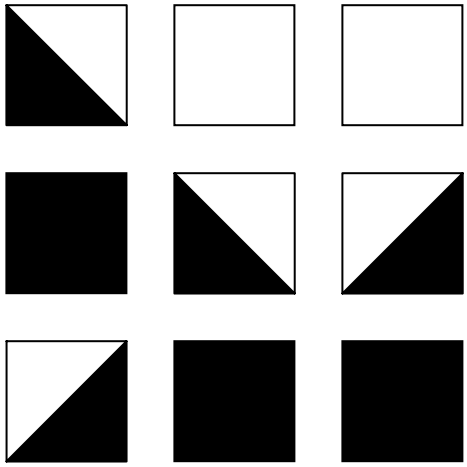


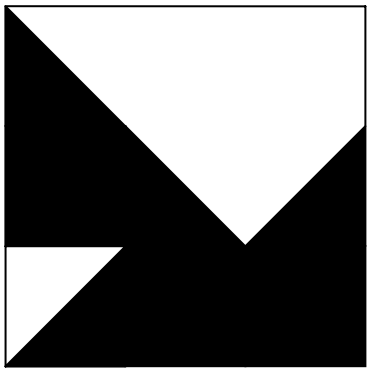


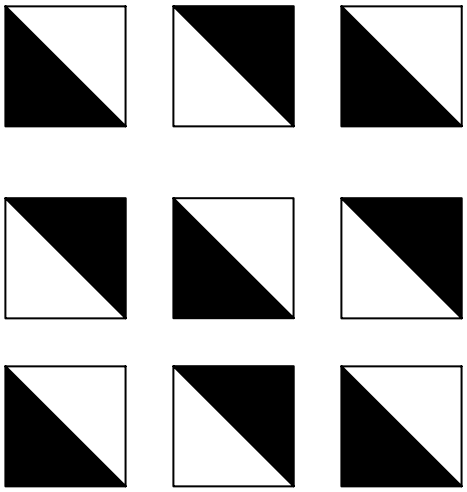


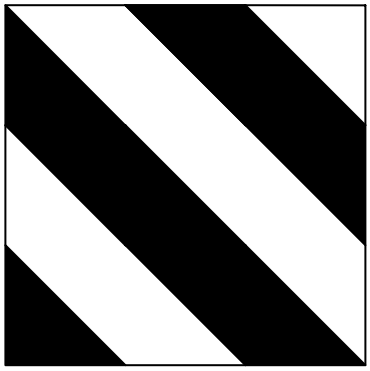












Cat / Swan Ambiguous Figure