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**Repetitive interests, behaviours and activities in
autism: their relationship to social-communication
impairments, and to cognitive inflexibility.**

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Overview

Autism is currently conceptualized as a unitary syndrome in which social-communication impairments and repetitive interests, behaviours and activities (RIBAs) are assumed to co-occur more often than would be expected by chance. It is argued that this assumption is not supported by the literature, and that the association between RIBAs and social-communication impairments is likely to be moderate or minimal. This suggests that autism may be a disorder of multiple underlying impairments, with particular susceptibilities being associated with specific autistic traits, rather than with the syndrome as a whole. The study reported in part two of this thesis was designed according to this possibility, with an association being sought between an executive function (set-shifting) and an element of RIBA ('Insistence on Sameness'). In a sample of 46 young people with an autism spectrum disorder and an IQ in the normal range there was evidence of a moderate association between these two variables, with those showing greater cognitive inflexibility having a stronger tendency towards insistence on sameness. The 'multiple underlying impairments' model of autism was supported by the fact that, in this sample, there was no association between social-communication impairment and RIBAs, or between cognitive inflexibility and social communication impairment. Future research in this area should be guided by the possibility that social-communication impairments and RIBAs are independent domains.

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Part One: Literature Review

**What is the association between the social-
communication element of the autism
syndrome and repetitive interests,
behaviours and activities?**

Abstract

Autism is currently conceptualised as a unitary syndrome, in which social-communication impairments are found alongside repetitive interests, behaviours and activities (RIBAs). This relies upon the validity of the assumption that social-communication impairments and RIBAs co-occur at an above chance level as a result of sharing underlying causes. In the current review it is argued that the evidence for this assumption is scarce: the very great majority of RIBA research has not been intended for or suited to its examination. In fact only three studies are fit to address directly the question of the relationship between social-communication impairment and RIBA, and these contradict each other. In consequence, further relevant evidence was sought in the behavioural and molecular genetic literature. This approach suggested that the correlation between social-communication impairments and RIBAs has been exaggerated in the current consensus about the autism syndrome, and that these aspects of autism may well share largely independent underlying causes. Some clinical and research implications are discussed.

Introduction

'Autism' is a label that describes a collection of phenomena, namely impairments in social interaction and communication, along with repetitive interests, behaviours and activities (American Psychiatric Association, 2000; World Health Organisation, 1992). It is also a working hypothesis that behind this collection of behaviours lies a 'disease entity' (Rutter, 1978). This raises the question of whether or not the correct collection of symptoms has been agreed upon by which to detect the underlying autism disease entity. How valid is the autism syndrome as it is currently defined?

In 1988, Michael Rutter and Eric Schopler, two eminent pioneers in the field of autism research, stated that 'there is no doubt that autism constitutes a valid and meaningful psychiatric syndrome; indeed the evidence on its validity is stronger than for any other psychiatric condition in childhood' (Rutter & Schopler, 1988, p. 427). More recently, the Handbook of Autism and Pervasive Developmental Disorders (Third Edition) endorsed this statement, adding that 'clinicians and researchers have achieved consensus on the validity of autism as a diagnostic category, and the many features central to its definition' (Volkmar & Klin, 2005, p.5).

Nevertheless, a number of different methodologies continue to be used to assess the validity of the autism syndrome. The resultant findings have not always given cause for the sort of optimism expressed by Rutter, Schopler and others. For example, factor analytic studies using data collected from both clinical (Constantino et al., 2004; Van Lang et al., 2006) and community (Austin, 2005) samples have suggested that social and communication impairments do not load onto different factors, and so probably do not represent distinct elements of a triad of impairments.

Recently, researchers have begun to consider another aspect of syndrome validity, by asking questions about the extent and nature of the association between, on the one hand, the social-communication aspects of the syndrome, and on the other, repetitive interests, behaviours and activities (RIBAs; Charman & Swettenham, 2001; Ronald et al., 2006; Ronald, Happe & Plomin, 2005). Previously it had been assumed that the curious collection of RIBAs described in the diagnostic manuals formed part of the autism syndrome cluster, and were in some (as yet unknown) way related to social and communication impairments.

It is the purpose of this review to examine this assumption by investigating the relationship between social-communication impairment and RIBAs. This will involve consideration of the extent to which these sorts of behaviours co-occur, and of whether they are likely to share common causes.

A note on terminology

As a synonym for RIBAs I have borrowed the term *nonsocial behaviours* from Ronald and colleagues. This conveys that two elements of the triad of autistic impairments (social impairments and communication impairments) are much more obviously related to social life than is the third (RIBAs).

In this review the terms Pervasive Developmental Disorder (PDD) and Autism Spectrum Disorder (ASD) are held to be synonymous (Charman, 2003). Both describe a set of disorders, of which autism is the prototype. Other PDDs or ASDs are diagnosed when full criteria for autism are not met, but when significant autistic difficulties are still held to be present. For example Asperger's Syndrome is

diagnosed when no developmental delay has occurred, and yet there are difficulties in social-communication and RIBAs. Atypical Autism and its DSM-IV equivalent Pervasive Developmental Disorder - Not Otherwise Specified are other ASD / PDD labels and will be discussed in the next section of this review.

Autism, 'Atypical Autism' and Pervasive Developmental Disorder - Not Otherwise Specified

There are people who have profound qualitative social and communication impairments, as well as RIBAs – which is to say that some people (around 20 per 10000 according to recent estimates; see Fombonne (2003) for review of epidemiological literature) meet the current criteria for a diagnosis of autism or asperger's syndrome. Numerous studies have documented a range of RIBAs in samples comprising people with autism. In these studies almost everyone with social and communication impairments, regardless of age or intellectual level, has shown at least some of the nonsocial elements of the syndrome (see Turner (1999) for a comprehensive and authoritative review of this literature).

It is crucial to appreciate that these studies are not methodologically suitable for addressing the question of whether there is an association between social-communication impairments and RIBAs. This is because they only include participants who meet criteria for autism, which ensures that everybody in the sample has RIBAs as well as social-communication difficulties. Thus it is unclear from the majority of research on RIBAs in autism to what extent the frequently described association of social-communication impairment and RIBAs is a real phenomenon, or the artefact of a selection bias in these studies.

It is not widely recognised in the autism literature that there are some children who have extreme autistic social-communication difficulties, in the absence of clinically significant levels of RIBAs. The diagnostic manuals can accommodate such individuals, with their inclusion in the heterogeneous categories of 'Pervasive Developmental Disorder – Not otherwise specified' (PDD-NOS; DSM-IV) or 'Atypical Autism' (ICD-10). Such children are familiar to clinicians involved with diagnosing autism. Despite this, they rarely appear in the research literature, and when they do it is often in a fleeting and incidental manner. At the time of writing there is only one peer-reviewed paper in which these individuals have been the main focus of investigation (Walker et al., 2004). Usually their presence is mentioned in passing, or can be inferred from information presented about the sample.

For example, in an investigation of 'social handicaps' in 63 children with a PDD, Tanguay, Robertson and Derrick (1998) noticed that of the 17 children diagnosed PDD-NOS 'almost all ... had marked abnormalities in the first two DSM-IV autism domains [i.e. social and communication impairment], but not in the third [i.e. RIBAs]' (p. 274).

A similar finding was reported by Walker and colleagues (2004) in the one paper specifically aimed at describing this phenomenon. Again a significant sub-group (n = 11 out of 21) of individuals with PDD-NOS were identified as being above threshold on the Autism Diagnostic Interview-Revised (ADI-R; Lord, Rutter & Le Couteur, 1994) for social and communication impairments, without abnormal levels of RIBAs. Walker and colleagues suggested that these children be given a specific label of 'atypical autism'.

In summary, it is certain that there are some individuals who have both the social-communication and nonsocial behaviours of the autism syndrome. There are also people who have profound social-communication impairments without clinically significant levels of RIBAs. Although clinically meaningful, these facts alone tell us little about the size of the association between social-communication and nonsocial autistic traits. In fact they would be compatible both with a near perfect correlation, or with a correlation that was very close to zero. Nevertheless, the apparent presence of what Walker called 'atypical autism' may represent an anomaly within the dominant paradigm (Kuhn, 1962) of our current conceptualisation of autism as a social-communication and nonsocial syndrome, and serves as an intriguing pointer towards further investigation of this matter.

Research Strategy

The type of study that could directly address this issue of syndrome analysis would involve using independent measures of social-communication and nonsocial autistic behaviour in a sample that has not been selected to meet full criteria for autism.

Surprisingly perhaps, considering the certainty with which it is generally asserted that RIBAs belong in the autism syndrome, there are only three such studies, two of which were conducted on the same sample by the same research team. The first was carried out during the 1970s (Wing & Gould, 1979), with the other two taking place some three decades later (Ronald et al., 2006; Ronald et al., 2005). As will be described below, these studies contradict each other, and do not in themselves allow for any firm conclusions. In consequence, I have attempted to draw on other types of autism literature in order to explore further the question in hand. This has involved reviewing the following types of literature on the phenomenology and aetiology of

autism: (1) historical information about the validation of the syndrome; (2) studies employing statistical methods such as Factor Analysis and Structural Equation Modelling to characterise clinical and non-clinical populations; and (3) quantitative and molecular genetic studies.

The scope of this review partly reflects a conceptual disagreement with one method used in past attempts at addressing the question in hand. The validity of the syndrome with respect to RIBAs has previously been considered in terms of their 'universality' and 'specificity' (Charman & Swettenham, 2001; Rutter, 1978). This approach to syndrome analysis assesses the validity of a particular symptom or sign according to whether it is very common amongst people thought to have the disorder (universality) and rather rare amongst people who do not (specificity). The argument goes that if almost all people with autism have RIBAs, and almost nobody without autism has them, then they are a valid component of the syndrome. I accept the value of insisting on universality, but I reject the importance of specificity in this matter. An analogy will serve to illustrate this. A sore throat is universal in tonsillitis, but not specific: people with tonsillitis always have a sore throat, but so do people with lots of other conditions. This lack of specificity has no bearing on the question of whether infected tonsils cause a sore throat, and does not mean that it is irrelevant that a person has a sore throat when trying to identify a case of tonsillitis. In consequence, I have not investigated the specificity of RIBAs by considering whether it is possible to have them in the absence of social-communication difficulties.

A brief history of autism

It has already been stated that the current diagnostic criteria for autism depend upon the validity of an assumption that social-communication and nonsocial autistic deficits cluster together, co-occurring more often than would be expected by chance. Between 1943, when autism was first conceptualised in something like its current form, and 2000, when the diagnostic criteria that we have today were published, this assumption was only put to the test once. It is important to recognise that the association between social-communication impairment and RIBAs, which is enshrined in the diagnostic manuals, is not built upon sound empirical foundations.

There is no definitive account of the process by which the current autism diagnostic consensus was reached, and some of the brief, fragmentary accounts that do exist sometimes contradict each other (e.g. Szatmari, 2001; Waterhouse, Wing, Spitzer & Siegel, 1992). It is possible however to identify three particularly influential pieces of work that gave rise to the notion that autism is a syndrome involving nonsocial as well social deficits. First is Kanner's (1946; Eisenberg & Kanner, 1956) work, in which he made the first description of infantile autism. Secondly is a literature review conducted by Rutter (1978), in which the so-called 'Rutter Criteria' were set out. These were highly influential on DSM III's (American Psychiatric Association, 1980) account of the syndrome (Volkmar & Klin, 2005). Thirdly is Lorna Wing and Judith Gould's (1979) classic Camberwell Study which influenced the design of DSM-III-R (American Psychiatric Association, 1987).

Although the preceding literature does include reports of children who would today be described as having autism (e.g. De Sanctis' (1906,1908) 'dementia precosissima

catatonica' or Earl's (1934) 'primitive catatonic psychosis of idiocy'; both cited in Rutter & Schopler, 1988, p.408), it was Kanner (1943) who first described the syndrome in its current form. In his initial paper on eleven children, he reported an apparent cluster of features that included an inability to form relationships, delay in speech, failure to use speech communicatively, pronominal reversal, normal physical appearance, repetitive behaviours and an obsessive insistence on sameness. Subsequently, with larger samples, he made his description more parsimonious, characterising the syndrome as having two core features: "extreme aloneness" and "preoccupation with the preservation of sameness" (Eisenberg & Kanner, 1956). Thus it was Kanner who first proposed the association between social-communication and nonsocial impairments as part of an autism syndrome. For the purposes of this review, this apparent association will be called the '*Kanner RIBA hypothesis*'.

Kanner's methodology was inductive, in that he observed similarities between particular patients that he encountered, and used these to construct a hypothetical syndrome, namely autism. Such methodology is productive in yielding hypotheses, and has been the origin of several of the most influential ideas in clinical psychology and psychiatry, such as Psychoanalysis and Cognitive Behavioural Therapy. However, on its own this approach does not constitute 'science', at least in the Popperian (1959) sense. Rather, inductive techniques should be viewed as the beginning of a process of validation, rather than the end, used to generate testable ('falsifiable') hypotheses. Accordingly, whilst Kanner was able to demonstrate that there are individuals who have both social-communication and nonsocial deficits together, his work does not speak to the question of whether there is an association

between these two types of difficulty. There are individuals who are tall and have fair hair, yet it does not follow from this that above average height and blondness co-occur more often than would be expected by chance, nor that both share an underlying cause.

The Rutter Criteria (1978) were, in effect, an endorsement of Kanner's original description of autism and, like Kanner's description, they were not substantiated by appropriate data. Rutter's method of syndrome analysis was to use the literature available to him to identify which putative autism symptoms were both specific and universal to the syndrome. In this manner, he addressed several controversies that were alive at the time (e.g. autism and its relationship to schizophrenia, the importance of age of onset, the incidence of learning disabilities amongst autistic people), and posited the triad of impairments which, in slightly modified form, are still used today. What is notable is that the empirical literature available at that time did not address the Kanner RIBA hypothesis, already three decades old by this point.

The first study to do this was carried out by Wing and Gould in Camberwell during the 1970s, with a specific aim of assessing 'the prevalence and distribution of the three types of abnormalities, *and whether they tended to occur together*' (Wing & Gould, 1979, p.13; italics mine). To this end, Wing and Gould sought children with either social impairments, or communication impairments, or repetitive behaviours, or 'severe mental retardation' within a cohort of 35 000 people under 15 years of age in the London borough of Camberwell. They did this by screening any children 'known to the local health, education, or social services for reasons of physical or mental handicap or behaviour disturbance' (p.13). Eventually they selected 132

individuals, who were then the subject of a thorough structured interview and direct observation.

Using the resultant behavioural data, sample members were classified as being either 'sociable, severely retarded' (n = 58) or 'socially impaired' (n = 72). Crucially, all of the latter group showed some repetitive behaviour, whilst the equivalent statistic for the sociable group was 38%. This difference was statistically significant. The authors concluded that their study 'showed a marked tendency for these problems [i.e. social impairment, abnormalities of language and repetitive stereotyped behaviour] to occur together.' This was the first time that the Kanner RIBA hypothesis had been put to the test, and the resultant data failed to falsify it.

However, the population from which Wing and Gould sampled limits the generalisability of their findings. They drew participants only from amongst people with a learning disability – it is notable that not one of their participants was in mainstream education. Thus, it is only possible to argue from their data that people with autistic social difficulties always have nonsocial difficulties *if they also have a severe learning difficulty*. This criticism of the study has become more pertinent with each passing year, as estimates of the proportion of people with social communication difficulties who have normal range IQs has been repeatedly revised in the upwards direction (e.g. Chakrabarti & Fombonne, 2005).

A second issue impacting on generalisability concerns comorbidity, symptom severity and missed cases. Wing and Gould identified their participants from amongst people already known to services by the age of 15. Thus they were likely to

attain a sample of individuals with particularly severe difficulties, and perhaps also raised levels of comorbidity. This could potentially have inflated the association they found between social-communication impairments and RIBAs. It is notable that the number of 'socially impaired' children they identified gave them a prevalence rate for ASDs of 21.2 per 10 000. A recent gold-standard epidemiological study (Baird et al., 2006) discovered a much higher prevalence for all ASDs of 116 per 10 000. This would suggest, assuming that the incidence of PDDs has not increased in the last 30 years (see Baron Cohen, 2006; Charman, 2003; and Fombonne, 2003 for discussion of this interesting question), that within their cohort of 35 000, around 330 children who would meet current criteria for an ASD were missed by Wing and Gould. How strong was the association between social-communication and nonsocial impairment in this group? We do not know.

In summary, during the development of the current conceptualisation of the autism syndrome, only once was the notion that social-communication and nonsocial impairments cluster together subjected to direct scientific investigation. This study failed to falsify the Kanner RIBA hypothesis. However, it lacked generalisability in relation to current ideas about the characteristics of people with autism.

Further attempts to address directly the Kanner RIBA hypothesis

Given that the hypothesised association between social-communication impairment and nonsocial behaviours in autism was only supported by one study, which inevitably had its limitations, it is perhaps surprising that another three decades passed before this question was once more addressed directly in an empirical study.

Ronald and colleagues published two reports (Ronald et al., 2006; Ronald et al., 2005) that appear to contradict Wing and Gould's findings, suggesting only a minimal to moderate phenomenological and aetiological association between autistic social-communication impairments and nonsocial behaviours in a community (non-clinical) sample. In their first study they gave a questionnaire to parents and teachers of over 3000 twins, who were part of the Twins Early Development Study, a cohort of twins born in the UK between 1994 and 1996. This questionnaire had 10 items designed to measure 'social impairments' (S; these covered social and communication aspects of autism) and 6 items addressing 'nonsocial behaviours relevant to autism' (NS). The results were striking: the correlation between these two scales in the parent data (n = 3996) was 0.29, and was .15 in the teacher data (n = 3090). This limited correlation of S and NS scores was also found in participants at the extreme high end of the distribution. Thus, according to teacher report, of those scoring in the top 5% for S, only 14% also scored in the top 5% for NS. Conversely, the high NS group (top 5%) had a mean S score that was only slightly above the sample mean. Parent report results showed a similar pattern: of those children scoring in the top 5% for either NS or S, only 15% scored in the top 5% of both scales.

This study, which used a twin sample, was designed to address the question of the heritability and shared genetic causes of social and nonsocial traits. It was found that heritabilities for both traits were high (62 to 76%). Crucially, the genetic correlation was low (.07 to .4), leading the authors to predict that 'over half of the genes found to be associated with quantitative variation in social behaviours will not be found to be associated with nonsocial behaviours associated with autism' (p.454).

These data would seem to offer a strong challenge to the notion that social and nonsocial impairments cluster together and share an aetiology. However, there are two features of the study that might temper such a pronouncement. The first concerns the nature of the sample, and the second concerns the psychometric properties of the instruments used.

In this piece of research a community sample was used in which autism was rare. Is it inappropriate that such a sample be used to learn about the relationship between autistic traits? Do the findings contrast with Wing and Gould's work simply because different populations were studied? It would be reasonable to dismiss the criticisms implicit in these questions if autism represented the extreme end of the distribution of traits that exist within the general population (dimensional approach). By contrast, if autism can be shown to be categorically distinct from variations within the normal range on social-communication and nonsocial traits (categorical approach), then it would be meaningless for Ronald and colleagues to have used data from a community sample to make pronouncements about autism.

Current evidence points towards the validity of the dimensional approach. The fact that relatives of autistic people have sub-threshold autistic traits (Bolton et al., 1994; Piven, Palmer, Jacobi, Childress, & Arndt, 1997) was an early pointer in this direction, implying that the genetic liability for the disorder was also associated with milder, non-psychopathological features. That these traits are found in the general population (not just in relatives of autistic people) has been lent credibility by a study measuring autistic traits in a large community sample (Constantino & Todd, 2003), which found a continuous distribution, with no discontinuity between normality and

psychopathology, as would have been evidenced by a bi-modal distribution. This finding has since been replicated in a Scandinavian community sample (Posserud, Lundervold & Gillberg, 2006). Therefore, on current evidence, it seems reasonable that Ronald and colleagues used a community rather than a clinical sample. However, as will be discussed below, the categorical versus dimensional debate in autism has not been conclusively settled.

Criticisms about the instruments used in the study are not so easily dismissed. As Ronald and colleagues acknowledge, there are problems with the questionnaires they used to measure autistic traits. Psychometric theory suggests that a measure should be assessed in terms of both reliability and validity (e.g. Streiner & Norman, 1995). In this case reliability was poor. Internal consistency was fairly low, with Cronbach's alphas of .53 and .56 for parent social and nonsocial scales, and .76 and .56 for teacher social and nonsocial scales. Test-retest reliability of the NS scale also appears to be inadequate ($r = .50, n=35$). It is therefore difficult to know to what extent the low correlations between the S and NS scales were due to high measurement error. It should be pointed out in defence of the study that high heritabilities were found for S and NS, which implies reliability (although not validity) for the instrument.

The validity of the scales is questionable, and not just because of their sub-optimal reliability. Before the study no formal testing was done to assess construct or criterion validity – only face validity was considered prior to data collection, with items being designed to correspond to specific DSM-IV criteria. This lack of validity testing raises the possibility that the scales measure something other than autistic traits. Data collected during the study allowed for the formal testing of this

possibility, since a proportion of the sample was said by their parents to have been given an ASD diagnosis. This diagnostic information was available for 37 individuals with an ASD. If the instruments used by Ronald were measuring autistic traits, we would expect most if not all of these to score highly on both the S and NS scales. In fact only 11 scored high S and NS, 15 scored high S but not high NS and 1 scored high NS but not high S. 10 did not score in the top 5% for either scale. This suggests that the scales used in this study had poor construct validity, and may not be measuring autistic traits.

The same group conducted a similar study shortly afterwards, using the same sample. Perhaps mindful of psychometric difficulties with their initial study, they used a different instrument to collect behavioural data: the Childhood Asperger Syndrome Test (CAST; Scott, Baron-Cohen, Bolton & Brayne, 2002). The findings broadly replicated the first study, although with slightly higher associations between social-communication and nonsocial traits. Thus, correlations between the elements of the triad were moderate: Communication Impairment and Social Impairment = .34; Communication Impairment and Repetitive Behaviour = .38; Social Impairment and Repetitive Behaviour = .23. Once again heritabilities for autistic traits were high (.78 to .81), and genetic influences on these were, for the most part, thought not to be shared.

There is something compelling about the fact that Ronald and colleagues replicated their initial findings, albeit on the same sample. However, certain psychometric difficulties persisted with this second study. Whilst a validated instrument was used, it was employed in a manner for which it had not been designed, and for which it had

not been validated. The CAST is a screening tool, which gives a single score for severity of autism symptoms as a whole. In this study items were selected, on the basis of face validity, that were thought to measure each element of the autism triad. These were then summed to give three scores, one each for an element of the autism triad. For these sub-scales, internal consistency was fairly low (alphas ranged between 0.48 and 0.64). Construct validity for the CAST as a whole was better, but still moderate: 75% (38 of 51) of children said by their parents to have an ASD scored above cut off on the CAST, and the equivalent statistic for asperger's syndrome was 55% (11 of 20). These psychometric difficulties partly reflect the originality of Ronald's and her colleagues' work, since instruments to measure individual autistic traits in the general population are still in need of further development.

Therefore, the examination of the three studies designed to directly address the Kanner RIBA hypothesis is not conclusive. In fact the Wing and Gould (1979) and Ronald (2006; 2005) studies flatly contradict each other. Perhaps their differences partly reflect the different samples (clinical versus community). However, such an explanation does not fit with the convincing notion that autistic traits are continuously distributed in the general population. It is not possible to resolve their contradictory findings through other considerations of methodology, as both sets of studies have their methodological problems, as well as their merits.

Thus we will turn to other types of study concerned with the behaviours and causes of autism. Whilst these were not designed specifically to address the Kanner RIBA hypothesis, it will be argued that they can shed some light on this issue. These will

be reviewed in respect to specific hypotheses, pertaining to the question of the association between the social-communication and nonsocial elements of the autism triad.

Factor Analytic Studies

One way in which the phenomenology of autism can be investigated is through the use of factor analytic techniques. These are used to detect underlying structures, ('factors' or 'components'), in a dataset by seeing which items tend to co-vary. A distinction can be made between exploratory and confirmatory analyses, with the latter being more likely to yield valid results, since they involve the testing of a model, rather than the data-driven generation of new models.

Factor analysis can be informative as to whether the social-communication and nonsocial elements of the autism syndrome co-vary (Constantino et al., 2004; Stella, Mundy & Tuchman, 1999). If they do, we would not expect them to show up as different factors, since individuals who score highly on social items would also be expected to score highly on nonsocial items and vice versa. If on the other hand the social and nonsocial elements of the triad are not closely correlated, we would expect to see distinct nonsocial and social-communication factors.

The outcome of a factor analysis depends upon the items that are used, and the characteristics of the sample from which data were collected. In addition, sample size in relation to number of items entered is an important consideration, with respect to power to detect different factors. Accordingly, the following criteria were used to select suitable studies: (1) Items should be entered into the analysis that measure the

full range of possible autistic behaviours. For example an analysis conducted using only social and communication items (e.g. Tanguay, Robertson & Derrick, 1997) would not make it possible for a 'nonsocial' factor to emerge, and so would not enable the assessment of the Kanner RIBA hypothesis; (2) Participants should be from across the autism spectrum, as attempts to attain too homogenous a sample (e.g. only individuals with strictly defined classical autism) may create an artefactual covariance for items that in a more loosely defined group would load onto different factors; (3) Only studies using data from well validated instruments should be used, to maximise validity and generalisability of results; (4) Sample size should be sufficient to justify the number of items entered into the analysis (Field, 1999).

Seven studies meeting these criteria were identified. Four of these used data from the ADI or ADI-R (Constantino et al., 2004; Georgiades et al., 2007; Tadevosyan-Leyfer et al., 2003; Van Lang et al., 2006), and one each used the Gilliam Rating Scale (Lecavalier, 2005), the Autism Behaviour Checklist (Miranda-Linne & Melin, 2002), the Autism Quotient (Austin, 2005) and the Social Responsiveness Scale (Constantino et al., 2004). Only one study (Van Lang et al., 2006) used confirmatory (Structural Equation Modelling) rather than exploratory analysis.

One notable feature of this literature is the lack of consistency between studies in terms of the specific factors yielded by principle components analysis. This partly reflects the fact that no two studies were the same in design. For example, even amongst those that used the ADI or ADI-R there was variability in terms of age of sample, sample composition, and specific items entered into the analysis.

Nevertheless, taken as a whole, there are two patterns that emerge from this collection of studies.

Firstly, in all analyses at least one factor was derived which related to some aspect of social-communication impairment. These were variously called 'social skills' (Austin et al., 2005); 'social impairment' (LeCavalier, 2005); 'non-responsive behaviour' (Miranda-Linne & Melin, 2002); 'social intent' (Tadevoysan-Leyfer et al., 2003); 'social-communication' (Georgiades et al., 2007; Van Lang et al., 2005); and 'social responsiveness' (Constantino et al., 2004).

Secondly, in all but one of the studies, there was at least one factor that comprised an aspect of nonsocial autistic impairment. These were 'patterns / detail' (Austin et al., 2005); 'Repetitive and Stereotyped Behaviour' (LeCavalier et al., 2005); 'Stereotyped Behaviour' (Miranda et al., 2002), 'compulsions' (Tadevoysan-Leyfer et al., 2003); 'inflexible language and behaviour' and 'repetitive sensory and motor behaviour' (Georgiades et al., 2007); and 'stereotyped language and behaviour' (Van Lang et al., 2005).

In the Constantino study 5 factors emerged (one large one accounting for 35% of the variance) which were theoretically indistinct from each other, so that social-communication and nonsocial items did not load onto different factors. This finding, contrary to six of seven other studies reported here, may be explicable in terms of the nature of the sample, which included ASD and non-ASD clinical participants. Any study mixing ASD and non-ASD groups runs the risk of missing factors within the clinical data. This is because between groups variance on items that discriminate

ASD from non-ASD can mask differential item variance within the clinical group (Stella et al., 1999).

In summary, whilst the factor analytic studies reviewed are discordant in terms of the specific factor structures they each propose, there are similarities. All studies yielded at least one social-communication factor. All but one also yielded at least one distinct nonsocial factor. The one study that used confirmatory, rather than exploratory factor analysis (Van Lang et al., 2006), showed this pattern of distinct social-communication and nonsocial factors. The one study that did not (Constantino et al., 2004) had problems with its design which may have reduced power to detect a factor including RIBAs. Thus the factor analytic literature lends support to the notion that social-communication and nonsocial elements of the syndrome are separate dimensions. It is worth noting that all but one study used clinical populations, some members of which were selected for having both social-communication impairments and RIBAs. This could have naturally worked against the discovery of distinct social-communication and nonsocial factors, by exaggerating the relationship between them. In light of this, the above findings are especially challenging to the Kanner RIBA hypothesis.

Research into the genetics of autism

When Kanner (1943) first described autism, he reasoned from the early age of onset that it had a neuropathological origin (Folstein & Rosen-Sheidley, 2001). However, in accordance to the dominant paradigm of the day, and in response to certain observable facts (autistic children did not have dysmorphic features or histories of serious birth trauma, as might be expected in people with inherited neurological

impairments, and their parents were often seen to be socially reticent) an account of autism's aetiology emerged in which environmental, relational causes were given prominence. The most (in)famous of these accounts was the 'refrigerator mother' hypothesis, popularised by Bruno Bettelheim (1967).

Subsequent observation of the high familiarity of autism (Rutter, 1968) led to the suggestion that genes may be important in the aetiology of autism. This idea has received emphatic confirmation in the three twin studies that have since been conducted to test this hypothesis (Folstein & Rutter, 1977; Steffenburg et al., 1989; Bailey et al., 1995). Across these studies, the average rate of concordance for autism between identical (monozygotic - MZ) twins was 70%, whilst the equivalent statistic for non-identical (dizygotic - DZ) twins was 0%. This yields a heritability estimate of around 90% for the populations studied. Even when combined, the numbers of twin pairs studied are not large (n=66, 36 MZ pairs and 30 DZ pairs), and the usual reservations about the twin studies apply here (see Chapter 3 of Rutter, 2006). However, the sheer magnitude of the effects found in these twin studies strongly imply that there is a significant genetic component to the aetiology of autism.

In response to this fact, and in concordance with the interest in genetics within the broader scientific community, much research has been carried out in the last two decades aimed at uncovering the genetic mechanisms involved in the development of autism (for recent reviews see Folstein & Rosen-Sheidley, 2001; Bacchelli & Maestrini, 2006; Klauck, 2006). It is notable that this sizeable body of work has yielded few substantive, replicable findings. The possibility that samples of people with autism participating in these studies are actually rather heterogeneous, despite

all sharing a diagnosis, has been used to explain this. This in turn has led to a search for ways of making more genetically homogenous groups upon which to perform genome scans. In effect, this has involved increased scrutiny of the autism syndrome, in terms of whether it really represents a unitary disorder, or a collection of independent impairments, each with different genetic underpinnings.

Such work has implications for the question with which the present review is concerned, and the genetic literature can usefully be interrogated on the question of whether the social-communication and nonsocial elements of the autism syndrome represent the same underlying abnormality. This can be approached both by considering the ways in which these phenomena do or do not co-occur, and the likelihood that they share the same genetic underpinnings.

In this section, I will review three types of genetic research with respect to the Kanner RIBA Hypothesis: (1) Family studies designed to trace patterns of familiarity for autism and autistic traits; (2) Quantitative genetic studies, designed to estimate the size of the genetic influence on autism and autistic traits; (3) Molecular genetic studies, aimed at discovering the actual genes associated with the development of autism.

It will be argued that these all lend support to the findings of Ronald and colleagues, that social-communication impairments and RIBAs are not strongly related, phenomenologically or aetiologically.

The Broader Autism Phenotype

The finding of Folstein and Rutter (1977) that many of the non-autistic co-twins in their twin study, whilst not meeting the criteria for autism, showed autistic-like personality traits sparked a renewed interest in the non-autistic relatives of autistic people. This interest resulted in a pair of large, parallel studies: the Maudsley Hospital Family Study (MHFS; e.g. Bolton et al. 1994) and the Johns Hopkins Study (JHS; e.g. Piven, Palmer, Jacobi, Childress & Arndt, 1997). Taken together, these reveal that the parents and siblings of autistic people have an increased tendency to show social reticence, communication difficulties and an insistence on sameness. These conceptually correspond to the autism triad, but are milder and came to be known as the Broader Autism Phenotype (BAP). It has been proposed that these are manifestations of the genetic liability to autism (Folstein & Rutter, 1988), and that these characteristics are at the mild end of a dimension, the extreme of which represents the clinical syndrome of autism.

For the purposes of the current review, it is helpful to ask of the BAP literature the following question: in relatives who display the BAP, do social-communication and nonsocial characteristics co-occur at a high rate? If the Kanner RIBA hypothesis is correct (and assuming that the BAP has the same genetic underpinnings as clinical autism), we would expect to find such an association, even in the milder, non-clinical form of the disorder. By contrast, if the association has been exaggerated, and is really an illusion maintained by the selection bias of studies of autism, we would expect to see low correlations between social and nonsocial difficulties in the parents and siblings of autistic people.

Although peer-reviewed papers on this subject have not offered explicit data on the correlations between social-communication and nonsocial BAP traits, they do present a number of facts that offer support for the latter of these two possibilities. Both BAP studies found that communication and social impairments were more common in relatives of autistic children than were RIBAs. In the MHFS (Bolton et al., 1994) the proportion of first-degree relatives with communication difficulties was 8.7%; with social difficulties was 8.7%; and with stereotyped behaviours was 5.7%. In the JHS (Piven et al., 1997) the estimates of the incidence of BAP traits tended to be higher, but a similar pattern emerged. For example, amongst fathers of autistic children, 57% showed social deficits, whilst only 26% showed stereotyped behaviours. These findings necessitate the presence of individuals with social difficulties, but without RIBAs. A further analysis of the MFHS (Pickles et al., 2000), which incorporated additional data collected from families of very low-functioning (IQ < 30) people with autism, offered more direct evidence on this dissociation of the triad within the BAP, showing that amongst the relatives of autistic children 6% were impaired in one aspect of the triad, 1.1% were impaired in two domains, and only 0.5% were impaired in all three domains.

Thus it would seem that the majority of affected relatives of people with autism show isolated autistic traits, rather than a mild form of the disorder incorporating slight impairments in all three domains. This offers evidence against the Kanner RIBA hypothesis, since it demonstrates a phenomenological dissociation of these traits and suggests their separate aetiologies.

Quantitative Genetics

Quantitative genetic studies aim to estimate the relative strength of genetic and environmental influences on variations of particular traits within a population. They represent 'natural experiments' in which environmental and genetic influences that would normally go together are separated out. Most commonly this is done using twin and adoptee designs (Simonoff, 2003). In relation to autism, such studies involve the measurement of autistic traits, and estimation of their shared environmental and genetic influences. Six of these studies are relevant to this review since they have sought to estimate the heritability of RIBAs, and so can be used to consider the extent to which these share an aetiology with the social-communication aspects of the autism syndrome. Two of these studies have already been described in some detail (Ronald et al., 2006; Ronald et al., 2005). It will be argued that, taken as a whole, the remaining four studies support the findings of Ronald and colleagues, thus working against the Kanner RIBA hypothesis.

Silverman and colleagues (2002) found evidence that the severity of an autistic person's RIBAs were largely unrelated to their level of social-communication impairment. In a large sample (n=457) made up of the members of 212 multiply affected sibships (i.e. collections of siblings in which two, three or even four children had autism) they looked at familiarity patterns of autistic traits, as measured by the ADI-R. They discovered that variability amongst family members was significantly lower than variability between families for measures of the autism triad, and took this as evidence that these traits are heritable. More importantly for the Kanner RIBA hypothesis, they found little overlap between sibships that scored highly for one aspect of the triad, and those that scored highly on another. For example, knowing

that a pair of siblings scored particularly highly in terms of social impairments was not helpful for estimating whether they would score highly for RIBA. The authors concluded that “the level of these clinical features are largely independent of each other and may have separate underpinnings” (p.70).

Such a finding is supported by a subsequent study from the same group (Kolevzom, Smith, Schmeidler, Buxbaum & Silverman, 2004), also concerned with comparing levels of intrafamilial and interfamilial trait variability to estimate familiarity. In a small sample of 15 pairs of identical twins and one set of quadruplets (n = 34) no positive associations were found within families for any of the autism symptom domains, supporting the idea that these are largely independent of each other, and may have different underlying genetic causalities.

However, two other studies offer a contrasting picture in relation to the Kanner RIBA hypothesis. A cluster analysis (Spiker, Lotspeich, Dimiceli, Myers, & Risch, 2002) involving 351 autistic siblings from 171 multiplex families found no evidence for distinct behaviourally defined sub-groups within their sample. This means that they did not empirically derive a group of children with social-communication impairments, but without RIBAs. Instead they derived three clusters that existed along a continuously defined dimension of symptom severity, which was heritable. In each of their groups, people had social-communication and non-social difficulties. Such a finding would suggest a co-occurrence of the social-communication and RIBA elements of the syndrome. However, a consideration of the study’s inclusion criteria greatly diminish its value in considering the Kanner RIBA hypothesis. Only those with “a research diagnosis of autism” (p. 131) were included, which meant that

only individuals with both social-communication and nonsocial impairments were entered into the analysis. This would exclude anyone with a dissociation between these types of autistic behaviour. The fact that none were found is testament to the researchers' rigour in applying their selection criteria.

The second study to suggest shared aetiology of RIBA and social-communication impairments (Sung et al., 2005) is harder to dismiss. 694 people from 201 multiplex autism families were included, and five domains of autistic traits were measured, including 'range of interest/flexibility'. Complex and esoteric analyses were performed, to adjust for the method of participant ascertainment; to estimate heritabilities; and to estimate shared environmental and genetic influences on these traits. These suggested a high (.92) genetic correlation between 'social motivation' and 'interest/inflexibility'. One point to make is that neither of these measures correspond exactly to the autism phenotype. Autism is a disorder of social skill, not social motivation (as is evidenced by Wing and Gould's (1979) "active but odd" category), and RIBAs include behaviours beyond a restricted range of interests and inflexibility.

Overall, the quantitative genetic literature reviewed here contradicts the Kanner RIBA hypothesis, with the exception of the study by Sung and colleagues.

Molecular Genetics

The two molecular genetic studies relevant to this review are in accordance with the quantitative genetics literature, since they both suggest that RIBAs are partly caused by genes that are not associated with social-communication impairment. Before these

studies are reviewed, it will be necessary to describe some features of molecular genetic research in general, and to describe how this research has been used to investigate the genetic causes of autism.

Autism molecular genetic studies, which aim to find specific genes for the disorder, fall into one of two categories: Linkage studies and Association studies. The research relevant to this review is of the former type. Such designs generally require families in which relatives (usually siblings) both suffer from autism. Their genomes are then scanned, being sampled at particular points, to see if there is co-inheritance between particular sequences of DNA and the trait being studied, in this case autism. If a sequence of DNA is close to a gene that causes the disorder or trait being studied, it is assumed that, within families, it will appear more often in affected individuals than in non-affected individuals. This relationship is probabilistic, due to recombination during meiosis. The statistic used to describe the likelihood that a sampled sequence of DNA is linked to a relevant gene is called the LOD (log odds) score. It is conventional to interpret a LOD score in excess of 3 as showing significant linkage, which means that such scores suggest the likely presence of a gene relevant to the behaviour being investigated (Simonoff, 2003). LOD scores of less than -2 are thought to exclude linkage and LOD scores in excess of 1 are taken to suggest possible linkage (Rutter, 2006, p.157).

Much money and time has been spent trying to investigate the genes that increase a person's risk of being autistic: since 1997 the results of 10 independent genome scans of this nature have been published (reviewed by Baccheli & Maestrini, 2006). This enterprise has not yet borne much fruit, since new studies have tended to

suggest new possible loci for pertinent genes, rather than to confirm previous findings. To date, some evidence of linkage (LOD score > 1) has been reported on every single chromosome except 12, 14, 20, 21 and Y. There is some agreement between studies, with a region on the long arm of chromosome 7 being the most replicated finding to date. Other regions of interest lie on Chromosome 2q (4 studies) and 17q (2 studies). However, even in these replications, there is not agreement as to the exact location of the region of interest on the chromosome, and there remains a majority of analyses that have not pointed to any of these regions as holding genes for autism.

One response to this has been to seek ever more behaviourally homogenous samples, with the assumption that this will equate to more genetically homogeneous samples in which genetic effects will be easier to detect. Molecular geneticists have therefore looked for subgroups of autistic people who are particularly similar according to a particular trait, in the hope that they will share particular autism genes relevant to that trait. Given the apparent high heritability and possible aetiological independence of the nonsocial element of the triad (Cuccarro et al., 2003; Hollander, King, Delaney, Smith, & Silverman, 2003; Ronald et al., 2006; Silverman et al., 2002) it is not surprising that these sorts of behaviours have been used to define subgroups in linkage analyses.

This fact offers us an opportunity to test the Kanner RIBA hypothesis according to the following logic: if RIBAs and social-communication impairments share their genetic underpinnings, we would not expect the selection of a high RIBA subgroup from amongst an autistic sample to offer any particular advantage in increasing the

frequency of particular autism trait genes. Thus, if attempts to subgroup according to RIBA score do not yield higher LOD scores, this will offer evidence for the Kanner RIBA hypothesis. However, if RIBAs and social-communication impairments have separate genetic underpinnings we would expect RIBA genes to be found at a high frequency in a subgroup chosen for their high levels of RIBA. This should be reflected in higher LOD scores for loci thought to be relevant to RIBAs. Such findings would contradict the Kanner RIBA hypothesis.

Shao and colleagues (2003) employed a novel technique called Ordered Subset Analysis (OSA) to promote sample homogeneity with respect to nonsocial autistic impairment in a linkage analysis. They used an empirically derived aspect of RIBA which they called 'insistence on sameness' (IS), after Kanner (Cuccaro et al., 2003). This involved summing a sub-set of the ADI-R items designed to measure RIBA. These were concerned with insistence on sameness and compulsions. OSA is designed to identify an homogeneous subset of families that contribute to linkage at a given locus. The researchers did not conduct a genome-wide search, but chose instead to focus their attention on chromosome 15q11 – 13. There were sound reasons for choosing this particular candidate region: linkage, association and cytogenetic evidence had all suggested the possible presence of autism genes there, and there was some indication that RIBA genes in particular might lie in this region.

Using OSA a group of participants homogenous in terms of their IS were identified, and this increased linkage at a locus within the region of interest from a LOD score of 1.45 to a LOD score of 4.71.

The following year, the results were published of a linkage analysis carried out on a sub-set of pairs of relatives (n=115) both of whom had an ASD and particularly high levels of obsessive-compulsive behaviours (Buxbaum et al., 2004). This trait has much in common with IS, being measured using several of the same ADI-R items. Once again, it was found that increasing sample homogeneity according to a nonsocial trait (i.e. creating a sub-sample of people who scored highly on obsessive compulsive behaviours), increased linkage at particular locus.

It is notable that this study did not find linkage at 15q11-13. This could be for a number of reasons. Perhaps the Shao result was a false positive. Alternatively, it could be that the slight difference in the way nonsocial autism traits were conceptualised was important, or that the Buxbaum study lacked the power to detect an effect on 15q11-13. This is not impossible given its sample size (n=115) and the relatively crude, categorical approach it took to creating an homogenous sub-set of participants for linkage analysis.

Regardless of discordant findings about the specific loci of genes associated with RIBA in autism, both studies support the notion that the nonsocial behaviours of autism are aetiologically distinct from the social impairments. This accords with the majority of the quantitative genetic literature, discussed above, that has investigated the independence of the different elements of the autism triad.

Conclusions

The consensus that autism is a unitary disorder, the components of which co-occur because of a shared aetiology, was based on scant evidence. In particular the

assumption that social-communication impairments cluster together with RIBAs in autism has received insufficient scrutiny. Instead it has been perpetuated over the decades by research designs inadequate to the task of detecting any possible dissociation. A pair of recent studies have strongly challenged the Kanner RIBA hypothesis. These are important and timely, but had methodological features that may have led to an under-estimation of the true association between social-communication and nonsocial impairments in their sample. Nevertheless, further evidence from genetic and factor analytic studies has lent support to the notion that the correlation between social-communication and RIBA aspects of autism has been exaggerated in the current consensus about the syndrome.

On the question of the Kanner RIBA hypothesis there is a spectrum of possible positions that can be taken. At one end is the 'strong traditionalist position', which states that RIBAs are virtually universal in people with autistic social-communication difficulties. At the other end is a 'strong revisionist position', which argues that there is no meaningful association between RIBAs and social-communication impairment, and that evidence for such a correlation is illusory, based on a sampling strategy that serves only to confirm what researchers already believed to be true. On the basis of the above evidence, where along this spectrum should the circumspect yet open-minded scholar place herself?

Happé, Ronald and Plomin (2006), in a 'perspective' piece in *Nature Neuroscience*, take a position towards the 'strong revisionist' end of the spectrum, arguing for a 'behavioural fractionation' (p. 1218) of the autism triad. Accordingly they suggest that researchers give up their search for causes of the syndrome as a whole, and

instead concentrate their efforts on understanding the development of its parsed elements. Such a stance has been largely supported by the current literature review. It should also be added that other strands of evidence, not covered here for reasons of space, have also tended to support this account of the social and nonsocial association in autism. For example, the absence of a convincing cognitive or neuropsychological account of autism which can encompass the full social and nonsocial range of autistic symptomology would fail to contradict the notion that these types of behaviour may have partially distinct aetiologies.

However, I would like to offer three recommendations against a move too far towards the 'strong revisionist' end of the spectrum. The first relates to the dimensionality of autism; the second addresses not the size of the association between social-communication and RIBA traits, but its meaning; and the third concerns construct validity of nonsocial autistic traits.

Firstly, whilst it is a growing consensus amongst researchers that autism is a dimensional rather than a categorical disorder, this debate has not been resolved conclusively. For instance, just because there is probably an overlap between aetiological processes implicated in normal range and pathological autistic traits does not mean there will be no specific genetic effects on profound social-communication impairment. Such a situation is thought to exist vis-à-vis IQ, with the genetic and environmental effects that influence variation within the normal range not being relevant to many cases of severe intellectual impairment (Volkmar & Dykens, 2002). If it were the case that there are at least some qualitative distinctions between autistic and non-autistic populations, this would diminish the relevance of the Ronald studies

to the question of autism syndrome analysis, and would weaken a strong revisionist stance.

Secondly, although the current literature suggests only a moderate correlation between social-communication and nonsocial behaviours, this is not the same as there being no association at all, and we should not throw the baby out with the bath water. Ronald and colleagues suggest that this correlation lies somewhere in the region of 0.3. Leaving aside methodological concerns, it is the case that a correlation in psychology of this magnitude is often taken to be highly meaningful. For example, correlations between goal conflicts and ill health (Emmons & King, 1988), trait positive affectivity and job satisfaction (Agho, Mueller & Price, cited in Warr, 1996) and use of primitive defence mechanisms and anti-social personality traits (Chabrol & Leichsenring, 2006) have all been reported to be in this range. That RIBAs are not universal in people with social communication disorders should not obscure the fact that these two sets of behaviours are moderately, and perhaps meaningfully, related.

This is mysterious: why should two sets of behaviours that are so conceptually distinct co-occur? What underlying mechanisms (genetic, cognitive, environmental) do they share? Is one a risk factor for the other? Attempts at answering these sorts of question will surely be fruitful in the quest for an understanding of autism, and social cognition more generally. Thus, whilst I acknowledge the value of research designs that measure different aspects of autistic symptomatology rather than confounding these with a single measure of 'autism severity', it is important not to lose sight of the fact that social and nonsocial deficits are related, even in community samples.

It is likely that a lack of construct validity has contributed to the failure to test the Kanner RIBA hypothesis convincingly. A recent development in the autism literature has been to parse RIBAs, reflecting the possibility that this diverse collection of behaviours do not all share the same aetiology. Thus three independent factor analyses using ADI and ADI-R data from clinical samples (Cuccaro et al., 2003; Richler, Bishop, Kleinke, & Lord, 2007; and Szatmari et al., 2006) all yielded an effectively identical two-factor model of RIBAs: 'Insistence on Sameness' and 'Repetitive Sensory Motor Actions'. In two of these studies designed to assess patterns of inheritance (Cuccaro et al., 2003; Szatmari et al., 2006), these two factors were independently heritable, and seemed to have differential relationships to developmental level. All but one of the studies described in this review (Shao et al., 2005) did not measure these two constructs separately. In fact, in the RIBA literature there is great variation in quite how 'nonsocial' behaviours are conceptualised and measured. Some studies measure a wide range of RIBAs (e.g. Bolton et al., 1994) whilst others have focused on a specific aspect of RIBA, such as 'obsessive-compulsive' behaviours (Buxbaum et al., 2004). It is possible that there is a 'true' autistic component of RIBA, and that there are other RIBAs that have more to do with developmental level. If this is the case, any study that failed to take account of this would risk underestimating the association between socio-communication impairment and autistic nonsocial impairments.

The ideal test of the Kanner RIBA hypothesis would use well validated measures that measured each element of the autistic triad, and were able to distinguish within RIBAs between 'Insistence on Sameness' and 'Repetitive Sensory Motor Behaviours'. It would use a large community sample which included people with

autism. Such a design will help us to better understand the nature and causes of social-communication difficulties by telling us the extent to which social impairments co-occur with nonsocial traits, such as obsessionality or insistence on sameness.

Clinical implications

The probability that autism as currently described in DSM-IV and ICD-10 is not unitary has implications for the way in which people with social-communication impairments and/or particularly high levels of RIBAs are labelled. Currently people who suffer severe social-communication difficulties without having RIBAs tend to be labelled with the terms PDD-NOS or atypical autism. Such 'catch-all' terms lack precision, as they are applied in response to a variety of symptom profiles. In this sense they fail in one of the key purposes of a diagnosis (Kendell & Jablensky, 2003), since they communicate with limited precision about the people to whom they have been attached. They are confusing and unsatisfactory to health-care professionals and service users alike.

The following (mutually exclusive) pair of changes to diagnostic convention would help improve matters: (1) the creation of a new category, such as 'atypical autism', containing only people with social-communication problems but without RIBAs; or (2) the removal of the restriction that RIBAs be present for an autism diagnosis to be made.

In order to justify either of these options, we would need evidence on the question of whether 'atypically autistic' children (i.e. who have social-communication

difficulties without RIBAs) are qualitatively distinct from classically autistic children, in terms of the difficulties they face, the help they need and their prognoses (Kendell & Jablensky, 2003). This information is not currently available, although there is some evidence for a statistical difference in terms of social (Tanguay et al., 1997) and functional (Walker et al., 2005) impairment between these two groups. Whether this statistically significant difference is clinically significant is another question. The fact remains that the 'atypically autistic' children in both these studies were highly impaired socially, and probably need psychological, social and economic support at similar levels to children with classical autism.

Research Implications

These follow from the idea that it is 'time to give up on a single explanation for autism' (Happé et al., 2006). Previously, cognitive theories of autism have tended to be criticised for the fact that they can only account for one aspect of the syndrome. For example, the 'Theory of Mind' (Baron-Cohen et al., 1985) account of autism has been criticised for failing to explain nonsocial aspects of the disorder (e.g. Turner, 1997). If it is the case that the social-communication and nonsocial behaviours of autism are only moderately or minimally related, this becomes a less significant criticism.

The traditional design of studies aimed at finding correlates of autistic behaviour in order to advance theory as to causes of these behaviours will have to change.

Previously, a typical design would involve comparing a group of autistic people with a control group of non-autistic people on a measure thought to be relevant to autism.

Given that different autistic traits appear to have different causes, results of such

studies are hard to interpret. If the autistic group perform worse than typically developing controls on, for example, the Wisconsin Card Sorting Test (Ozonoff & Jensen, 1999), does this relate to their autistic behaviours per se, or just their RIBAs, or just their social-communication difficulties? Standard case-control studies do not allow for an answer to such a question, as they confound different autistic traits.

Bishop and Norbury (2005) propose one solution to this, by using comparison groups that differ according to only one autistic trait. Thus they compared a fully autistic group who experienced difficulties in all three areas of the triad (social, communication and RIBAs), with a group who only had impairments in the social-communication domain. This enabled them to interpret group differences as being related to RIBAs.

The difficulty with such a design is that it can be hard to recruit sufficiently large numbers of people with the required symptom profile. However, multiple regression could also be used to examine the relationship between a particular variable (e.g. theory of mind) and a particular trait (e.g. social impairment) whilst other autistic traits are controlled for through their entry into the model as predictors. This would allow for the independence of autistic traits without setting such challenges for recruitment.

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Part Two: Empirical Paper

What is the relationship between cognitive flexibility and insistence on sameness behaviour in autism?

Abstract

This study aimed to investigate whether cognitive inflexibility was associated with one specific element of autistic behaviour, insistence on sameness (IS), but not with the other elements of the syndrome. Cognitive inflexibility was measured using three tests of set-shifting (The Wisconsin Card Sort Test, the California Trail Making Test and the Intradimensional/Extradimensional Shift Task) in a sample of 46 young people with an autism spectrum disorder and a verbal IQ in the normal range. IS was measured using a parent-report questionnaire, the Repetitive Behaviour Scale – Revised, and social-communication impairment was quantified using the 3Di, a comprehensive, semi-structured parent interview. No correlation was found between IS and social-communication impairments. One set-shifting measure (trail making) showed a moderate correlation with IS in the direction predicted when a one-tailed test was used. There was no relationship between this measure of set-shifting and social-communication impairment. These findings suggest that cognitive inflexibility is associated with greater IS in children with an ASD, but not with social-communication impairments. They also offer support for the idea that the social-communication and repetitive behaviour components of the autism syndrome may represent separate dimensions with distinct underlying susceptibilities.

Introduction

Autistic Spectrum Disorders (ASDs), of which childhood autism can be considered the prototype, are currently characterised by the following triad of impairments: (1) Qualitative impairments in social interaction, (2) Qualitative impairments in social communication and (3) A restricted repertoire of interests, behaviours and activities (Charman, 2003). Implicit in this definition of the syndrome are two related assumptions: firstly that these three types of behavioural impairment co-occur more often than would be expected by chance; and secondly that this occurs because they share a common underlying abnormality.

It is likely that these assumptions are not sound. Clinicians and researchers have noticed that there are some individuals who have severe social and communication impairments, without showing repetitive interests, behaviours and activities (RIBAs; Tanguay, Robertson & Derrick, 1997; Walker et al., 2005). More compellingly, a pair of studies measuring autistic symptomatology in a large ($n > 3000$) community twin sample have found only a moderate correlation between the 'social' (i.e. social and communication impairments) and 'nonsocial' (i.e. RIBA) elements of autism (Ronald et al., 2006; Ronald, Happe & Plomin, 2005). These same studies also found minimal shared genetic overlap between the three autism domains. Such findings suggest that autistic behaviours often occur independently, without the presence of other elements of the syndrome. It is likely that the numerous studies describing a co-occurrence of social and nonsocial autistic impairments (see Turner, 1999) reflect a sampling effect rather than a real phenomenon. This is because they sample only individuals with a full autism diagnosis, and so exclude individuals who have some but not all of the autism triad of

impairments. Evidence for a dissociation of behavioural elements of the syndrome raises the possibility that autism may not be explicable by one underlying abnormality, and may in fact be ‘a disorder of multiple underlying impairments’ (Goodman, 1989).

This has implications for the designs of studies seeking to discover the underlying abnormalities associated with autistic behaviours (Bishop & Norbury, 2005a).

Traditional case-control studies comparing matched groups (autism versus typically developing or learning disabled) confound diverse elements of autistic symptoms. For example, if an autism group (selected to have the triad of impairments) scores worse on the perseverative responses measure of the Wisconsin Card Sort Test task (often taken as a measure of set-shifting) than a well matched non-autistic control group (Ozonoff & McEvoy, 1994), this does not tell us whether difficulties with set-shifting are correlated with all three elements of the triad, or just two or just one. Furthermore, it is far from clear whether elements of the triad, which have been treated as unitary, are in fact heterogeneous. It may therefore be possible that only one aspect of an element from the triad correlates with set-shifting. Accordingly, studies seeking to understand better the underlying abnormalities in autism should be designed to accommodate the possibility that particular abnormalities correlate with particular types of autistic *behaviour*, rather than autism per se.

The current study is concerned with the least researched and understood category of autistic behaviours from the triad of impairments: RIBAs. This curious collection of features comprising the nonsocial aspects of autism, as described in DSM-IV-TR (American Psychiatric Association, 2000) and ICD-10 (World Health Organisation,

1992) appears to be heterogeneous. For example it includes such diverse behaviours as being obsessively interested in one subject, and making repeated finger-flapping movements. Recently, empirical evidence has been published to support this impression that the RIBA construct is not unitary. Two factor analytic studies (Cuccaro et al., 2003; Szatmari et al., 2005), carried out by independent research groups using data from separate samples of autistic children, have yielded strikingly similar results, showing that RIBA itself may comprise two distinct components: 'Insistence on Sameness' (IS) and 'Repetitive Sensory and Motor Behaviours' (RSMBs).¹ There are three reasons to suggest IS is the more specifically autistic trait of these two, with RSMB being a general reflection of a learning disability or developmental delay. Firstly, in both studies, RSMBs were negatively correlated to a measure of cognitive development, whereas IS did not have a relationship with developmental level. Secondly, both research groups found IS to be heritable unlike RSMBs, and ASDs have a strong genetic component (Folstein & Rutter, 1977). Thirdly, IS behaviours are rare in individuals with a learning difficulty, where as RSMBs are not (Turner, 1999). Given these facts, it is plausible that, when researching correlates of the nonsocial communication element of the triad, it will be fruitful to focus on IS, rather than on the heterogeneous collection of RIBAs.

One common approach to trying to understand the cause (or causes) of autism has been to look for correlations between particular cognitive abilities and autistic behaviour. The hope is that such correlations can suggest cognitive causal mechanisms for the

¹ It should be noted that 'Insistence on Sameness' is a slightly misleading term, as the factors derived by both groups included items measuring compulsions, as well as those measuring insistence on sameness. However the insistence on sameness (IS) term will be used here, for the sake of brevity, and to reflect this construct's origins in the factor analytic work of Cuccaro, Szatmari and their colleagues.

development of autistic behaviour. This approach has yielded a number of accounts of autism, the most influential of which focus on 'Theory of Mind' (Baron-Cohen, Leslie & Frith, 1986), 'Weak Central Coherence' (Frith, 1989) and 'Executive Function' (Russell, 1997). It is the third of these that is pursued in this study.

As long ago as 1978, Antonio Damasio noted certain similarities in behaviour between autistic individuals and people who had sustained damage to their frontal lobes (Damasio & Maurer, 1978). Since that time, a number of researchers have taken up the idea that the range of abilities associated with the frontal lobes, and particularly prefrontal cortex, could be the key to understanding the behaviours seen in autism. This has come to be known as the 'Executive Function' theory of autism (see Russell, 1997). Executive function has been described as 'the ability to maintain an appropriate problem-solving set for the attainment of a future goal' (Pennington & Ozonoff, 1996, p. 54). It draws upon a diverse collection of interrelating abilities, including working memory, inhibition, generativity, planning, maintaining set and set-shifting. A consensus has emerged that autistic people have a particular executive function profile, showing unimpaired working memory and response inhibition along with deficits in planning, cognitive flexibility (set-shifting) and generativity (e.g. Ozonoff & Jensen, 1999). It should be noted that this consensus relates to autism as a whole, and does not extend to our understanding of the relationships between executive functions and specific autistic symptoms.

One of the advantages that the executive function theory of autism has over its leading rivals (Theory of Mind and Weak Central Coherence) is that it is conceptually able to

explain RIBAs. This fact has been remarked upon by executive function researchers (e.g. Turner, 1997; Hill, 2004). Surprisingly, there is little empirical evidence published in peer-reviewed journals that would allow us to evaluate this intuitively valid idea, and the little research that has been done does not add up to a consistent account of the relationship between RIBAs and executive function. Turner (1997) has argued that RIBA results from deficits in generativity and inhibition. However, the data she uses to back up her arguments have not been published. Recently Bishop and Norbury (2005a, 2005b) tested these ideas and failed to replicate Turner's unpublished findings. Lopez, Lincoln, Ozonoff and Lai (2005) correlated various executive functions with scores of RIBA in a sample of 17 adults with autism who had normal-range IQs. They reported that scores for planning and generativity did not correlate with levels of RIBA, concluding that a pattern of intact working memory and inhibition along with deficits in cognitive fluency best predicted variations in RIBA. However these findings are hard to interpret, as they are founded on analysis that is inappropriate given the small sample, and so may reflect type one and type two errors. Furthermore Lopez et al. (2005) did not control for level of social and communication impairment in their analyses, and so this makes it harder still to interpret their findings on the question of this executive function profile being specifically associated with RIBAs, rather than with autistic symptoms more generally. Also, the measure of cognitive flexibility used is probably invalid, as it includes a score that fails to control for, amongst other things, processing speed, knowledge of alphabet, ability to count and motor control.

Thus, whilst the idea that executive function strengths and difficulties underlie RIBAs (including IS) has face validity, there is currently little empirical evidence in its favour.

The relevant literature allows for no certain pronouncements on the relationship between executive function and IS. It does, however, suggest which executive functions might be investigated as IS correlates. The two executive function skills commonly found not to be impaired in people with autism (working memory and response inhibition) are unlikely to correlate with IS, whereas the three executive functions skills that are impaired in autism (planning, cognitive flexibility and generativity) are all possible candidates. Of these, planning (Lopez et al., 2005) and generativity (Bishop & Norbury, 2005a) have both been shown not to correlate with measures of RIBA. That leaves cognitive flexibility, which has been found to correlate with RIBA as a whole amongst adults with autism (Lopez et al., 2005). This, therefore, seems the most likely place to start when looking for a correlate with IS.

Cognitive flexibility has been conceptualised in the autism executive function literature in terms of the ability to shift from one cognitive set to another (Pennington & Ozonoff, 1996). When attempting to measure set-shifting it is important to acknowledge the difficulty in identifying tasks that reflect specific executive abilities, rather than clusters of interrelated cognitive processes. This is known as the ‘task impurity problem’ (e.g. Miyake et al., 2000): since executive functions necessarily draw on a range of other cognitive processes, a low score on a test will not necessarily reflect an executive weakness. Furthermore, it is clear that some distinct elements of executive function cannot really exist independently of other executive functions. For example, set-shifting involves the disengagement from one irrelevant task set, and the engagement with another relevant task set (Miyake et al., 2000). It is hard to conceptualise such an operation without involving inhibition and working memory, and possibly generativity

too. Thus the interrelatedness of executive processes makes their individual measurement difficult.

Accordingly, for the current study I decided to use three tasks that have been considered to measure set-shifting yet have varied surface characteristics, in order to maximize the chance of obtaining a valid measure of cognitive flexibility. The classic test of executive function, the Wisconsin Card Sorting Test (WCST), was selected as it has been widely used as a measure of set-shifting in a variety of populations (see Pennington & Ozonoff, 1996). As this measure was used in the only other study similar in design to this one (Lopez et al., 2005), its use was also selected to allow comparison between the studies' findings. The WCST was supplemented with the Intradimensional/Extradimensional Shift Task (ID/ED). This task manipulates variables that are not controlled for in the WCST, and so distinguishes between those who can and cannot make an 'extradimensional' set shift (see methods section for explanation of this task). Thirdly, the California Trail Making Test was selected. This had also been used by Lopez and colleagues, and like the WCST had the advantage of allowing comparison of results. Furthermore, it has been proposed as a valid measure of set-shifting and yet has very different face characteristics from the other two tasks. The California Trail Making Test is well designed to control for the various cognitive processes involved in its completion, and so was thought to suffer less from the task impurity problem than the other two tasks. The WCST and trail-making task yield dimensional set-shifting scores, whereas the ID/ED yields a categorical score for set-shifting.

In summary the following three ideas are especially relevant to the design of this study: first, autism may be a disorder of multiple underlying impairments in which distinct impairments underlie different aspects of the syndrome; second, the RIBA construct may lack validity, and needs to be parsed, with ‘Insistence on Sameness’ being a likely component of the RIBA construct particularly relevant to autism; finally, the ability to shift sets is a potential underlying abnormality for IS.

On the basis of these, the following predictions were made for a sample of high-functioning children with an ASD: (1) A dimensional measure of set-shifting will correlate with IS, even when levels of social-communication impairment are controlled for, and poorer set-shifting will be associated with higher levels of IS; (2) The categorical ID/ED measure will correlate with levels of IS, with those able to make the extradimensional shift showing the lower levels of IS.

Methods

Design

A correlational design was used to examine covariance between measures of set-shifting and a questionnaire measure of IS. In order to test the first hypothesis a hierarchical multiple regression was used, with a measure of IS entered as the outcome variable. A measure of social-communication impairment was entered in the first block and a measure of set-shifting was entered in a second block. To test the second hypothesis a point-biserial correlation coefficient was derived, using a binary extradimensional shift variable (shift made versus shift not made) and a continuous IS variable.

Participants

Participants comprised 46 young people who had attended a Social Communication Disorders Clinic (SCDC) at a Tier IV hospital. The following inclusion criteria were applied: (1) Formal diagnosis of an ASD according to IDC-10 criteria, given by the multi-disciplinary team at the SCDC, using information from parents, school and direct observation; (2) Age between 8 and 16 years; (3) Verbal IQ of 70 or above. It was decided to include individuals with the full range of ASD diagnoses, rather than only those with narrowly defined classical autism, so as to maximise the chance of studying individuals with low as well as high levels of IS. This reflects arguments made in the introduction to this paper that social-communication and IS may be distinct dimensions, and that studies should avoid sampling biases that exclude individuals who experience one type of impairment but not the other. Those under the age of 8 were excluded from the study, as there are no norms for the Delis Kaplan Trail Making for this young age group. Adults were excluded from the study to promote sample homogeneity, in case this group showed distinct relationships between social-communication, IS and cognitive flexibility. The IQ criterion for inclusion was chosen to ensure that participants would have the cognitive ability required to complete the tests of cognitive flexibility, and to limit the effect of IQ as a confounding variable in analyses.

Table 1 describes the sample according to diagnostic category. The gender ratio is approximately 9 to 1 in favour of males, as would be expected in a high-functioning ASD group (Gilmour, Hill, Place & Skuse, 2004).

Initially, 86 individuals meeting inclusion criteria were identified and invited to take part in the study. Of these, 50 agreed to participate. Of these, 4 were not included for geographical reasons (i.e. they lived too far away). This gives a response rate of 54 per cent. Clinical data on autistic symptomatology, age and gender were available for individuals who did not participate in the study. T-tests revealed that these non-participants were similar to participants with respect to level of social-communication impairment and age. Although the non-participant group (7 out of 40) appeared to have a higher proportion of females than the participant group (4 out of 46), this apparent trend was not significant (Chi-square (1,N = 86) = 1.58, $p = .21$). On a scale measuring RIBAs, ranging from 0 to 12, the participants (mean = 4.86, SD = 2.29) scored higher than the non-participants (3.66, SD = 2.51; $t(77) = 2.21$, $p < 0.05$).

Table 1 – Participant Characteristics

	Autism	Asperger's Syndrome	Atypical Autism	Total
Age range (years)	8.1 to 16.3	8.0 to 15.0	9.9 to 15.7	8.0 to 16.3
Age Mean (SD)	11.8 (2.5)	11.7 (2.0)	12.4 (1.8)	11.8 (2.2)
Males	19 (91%)	16 (94%)	7 (88%)	42 (91%)
Right-handed	20 (95%)	14 (82%)	8 (100%)	42 (91%)
VIQ range	70 to 128	71 to 138	70 to 116	70 to 138
VIQ Mean (SD)	92.2 (15.4)	104.9 (18.9)	90.3 (16.9)	96.5 (17.9)
PIQ range	69 to 127	77 to 126	53 to 125	53 to 127
PIQ Mean (SD)	96.2 (17.3)	103.8 (15.4)	83.6 (24)	96.7 (18.5)
FIQ range	70 to 131	78 to 127	63 to 121	63 to 131
FIQ Mean (SD)	93.4 (15.8)	104.8 (15.6)	86.3 (17.2)	96.3 (17.2)
Total Number	21	17	8	46

Measures

Social-communication impairment

Social and communication impairments were characterised and quantified using the Dimensional, Developmental and Diagnostic Interview (3Di; Skuse et al., 2004). This computerised, semi-structured interview is administered by trained clinicians, and takes between one and two hours to complete. It includes 122 items that contribute to an ASD algorithm, which outputs scores for social impairment, communication impairment and restricted interests, behaviours and activities. In practice, the social and communication impairments are highly correlated in clinical samples, and can be summed to generate a single score of social-communication impairment (Skuse et al., in press).

Psychometrically, the instrument appears to be sound. Test-retest and interrater reliabilities are in the good to excellent range (Intraclass Correlations all exceed .86). Criterion validity was assessed through comparison to the Autism Diagnostic Interview – Revised (Lord, Rutter & LeCouteur, 1994), and agreement on threshold was found to be good for social (86%) and communication (100%) domains of the autism triad.

Repetitive Behaviour

The Repetitive Behaviour Scale – Revised (RBS-R; Bodfish, Symons, Parker & Lewis, 2000) was used to measure repetitive behaviours (see Appendix 3). This questionnaire comprises 43 items, grouped into six subscales (Stereotyped Behaviour, Self-injurious Behaviour, Compulsive Behaviour, Sameness Behaviour, Ritualistic Behaviour and Restricted Behaviour) and as such provides a quantitative measure of the full range of repetitive behaviours as defined currently in the diagnostic manuals. The questionnaire

is designed to be completed by parents, who are asked to rate 43 distinct aspects of repetitive behaviour on a scale from zero to three, on which zero means the behaviour does not occur, and three means the behaviour occurs and is a severe problem for their child.

The 'Compulsive Behaviour' and 'Sameness Behaviour' sub-scales were summed to create a variable corresponding to the 'Insistence on Sameness' construct described above. Amongst participants in this study, this 19-item scale had excellent internal reliability (Cronbach's alpha = 0.9). In common with the construct described by Cucarro et al. (2003) and Szatmari et al. (2006), it did not correlate with age ($r(46) = .19, p = .21$), verbal IQ ($r(46) = -.19, p = .2$), performance IQ ($r(46) = -.18, p = .24$) or full-scale IQ ($r(46) = -.21, p = .17$).

Intelligence

The Wechsler Abbreviated Scale of Intelligence (WASI; Wechsler, 1999) is a measure of intelligence, consisting of four subtests: Vocabulary, Block Design, Matrix Reasoning and Similarities. These are similar in format to their equivalents in the Wechsler Adult Intelligence Scale III (Wechsler, 1998) and Wechsler Intelligence Scale for Children III (Wechsler, 1991). They were selected for their high loadings on general intelligence ($r > .7$) and because they cover both the verbal and performance domains of ability. The test is suitable for people aged between 6 and 89 years and takes around 30 minutes to administer. It was standardised on a large ($n = 2245$) sample in the USA, and yields scores for Verbal, Performance and Full-Scale IQ (Wechsler, 1999).

Cognitive Flexibility

Three tests were administered to measure cognitive flexibility: the Delis-Kaplan Executive Function System (D-KEFS) Trail Making Test (Delis, Kaplan & Kramer, 2001); the Wisconsin Card Sorting Test – 64 Card Version (Kongs, Thompson, Iverson & Heaton, 2000); the Cambridge Neuropsychological Test Automated Battery's Interdimensional/Extradimensional (ID/ED) shift task (Robbins et al., 1994).

The D-KEFS Trail Making Test has five conditions. The primary cognitive flexibility task is a number-letter switching condition, similar to the Part B of traditional Trail Making tasks (Lezak, 1995) in which participants are asked to connect a sequence of circles, alternating between those containing a number and those containing a letter. The D-KEFS test contains four other conditions: visual scanning (in which every three on the page has to be crossed out); number sequencing (joining a sequence of numbers, one to 16); letter sequencing (joining a sequence of letters, A to P); and motor speed (tracing along a dotted line between blank circles as quickly as possible). These components are included to control for levels of skills required for successful completion of the number-letter sequencing task, such as the ability to count to 16, penmanship, processing speed and so on. For each component of the test, completion time is the primary outcome measure.

This trail making test has been standardised on a large ($n = 1750$) representative USA sample of people aged between 8 and 89 years. The set-shifting score is derived by subtracting a standardised score reflecting time to complete both number and letter

sequencing from a standardised score for the number-letter switching condition completion time. Forthwith, this will be referred to as the 'trail making shifting score'.

The Wisconsin Card Sorting Test – 64 Card Version (WCST-64) is an abbreviated version of the classic task of executive function. It requires participants to match each of 64 cards to one of four key cards, according to one of three dimensions (shape, colour, number of shapes). Each time a card is placed by a key card, the examiner states whether or not this is a correct match. Initially the participant is required to work out the rule by which successful matching can be accomplished. Once it is clear that this has been achieved (10 successive correct answers) the rule is changed, unbeknownst to the participant. This changing of the sorting rule after ten correct matches continues throughout the test, and the participant is never told explicitly that rule changes are happening. Individuals differ in the extent to which they persist with an old rule to guide their card sorting, despite feedback that they are now matching cards incorrectly. Thus, the test generates, amongst other scores, a measure of 'Perseverative Errors', which is the number of incorrect matches, made according to an out of date rule. This is thought to be a measure of cognitive flexibility. The WCST-64 has been standardised for use with children and young people on a USA sample of 452 young people aged between six and 17 years old, so raw scores can be converted into age-normed standardised scores, as well as T-scores and percentiles.

Several studies described in the test manual have shown the WCST-64 to generate scores closely related to those yielded by the longer (128-card) version of the test, in

clinical (Smith-Seemiller, Franzen & Bowers, 1997), non-clinical (Heaton & Thompson, 1992) and child (Sillanpaa et al., 1993) populations.

The Intradimensional/Extradimensional Shift subtest (ID/ED) is part of the Cambridge Neuropsychological Test Automated Battery, a set of exercises that are administered using a touch screen computer. It is a descendant of the WCST, designed to identify more precisely the nature of an individual's set-shifting difficulties. Participants are presented with distinct pairs of stimuli that both have two attributes: their shape, and a line pattern. At each presentation they are asked to select one stimulus, and are given feedback by the computer as to whether they chose the right one. In this way they learn a rule that can be applied to ensure they pick the correct stimulus each time. At first this rule always concerns the stimulus shape, so that the line pattern is irrelevant.

After six consecutive correct choices the rule is changed. The first six times this happens, shape continues to be the salient component of the stimuli. Thus, whilst a shift is required to a new rule, this is 'intradimensional', with the dimension in this case being shape. For the seventh rule change however, line pattern becomes the salient dimension: the participant is required to make an 'extradimensional' shift. The ID/ED measure used in this study is the binary outcome of whether or not the extradimensional shift was made.

Procedure

Ethical permission for the study was obtained from the Great Ormond Street and Institute of Child Health Research Ethics Committee (see Appendix 1), and the project was registered with the University College London Data Protection Officer.

Individuals meeting eligibility criteria were identified from a database of young people who had been assessed for an ASD at a Tier IV children's hospital. Letters were sent to the parents of those identified as being eligible for the study, inviting them and their child to participate. These letters contained separate information sheets for children and adults, child and adult consent forms (these documents are presented in Appendix 2), and a copy of the RBS-R. Those wishing to participate were asked to return the completed questionnaire and consent forms in a stamped, addressed envelope provided. Informed consent was required from both parents and children. When telephone numbers were available, those who did not reply to the mail out were followed up to see if they wished to join the study.

With those who did decide to participate, an appointment was arranged for the principal investigator (WM) to visit their home to carry out cognitive testing. In all cases the three tests of cognitive flexibility were administered according to standard procedures described in their user manuals. The ID/ED was always the final test, as its instructions forewarn participants of a rule change, and it was thought that this could influence performance on the WCST, which is to be administered with participants not anticipating a rule change. The order in which the trail making test and WCST were administered was alternated with each testing session. IQ testing was only carried out when a WASI had not previously been completed with the child.

Results

Performance on tests of cognitive flexibility

Age-standardised scores on the WCST and trail making test are displayed in Table 2.

This is not a case-comparison study, and so with no control group, confident statements on the level of participants' set-shifting ability cannot be made. Nevertheless, standardised tests were used, and it appears that as a group, the participants performed relatively well on these tests of cognitive flexibility. Mean scores for WCST perseverative errors and for the trail making shifting score are both close to the standardised means for these tests: 100 (SD = 10) and 10 (SD = 3) respectively. It should be noted that the range of scores for each measure is high. Further analysis revealed that there were larger than would be expected proportions of individuals in this sample who performed poorly on the tests. In a normally distributed sample, we would expect roughly 16 percent of individuals to get a standard score one standard deviation below the mean, and roughly two percent to score two standard deviations below the mean. As can be seen in table 2, greater numbers than this performed at this low level.

Every participant was able to make the intradimensional shift on the ID/ED, but only a third (15 of 46) successfully made the extradimensional shift.

Relationships between tests of cognitive flexibility

The three cognitive tests were selected in order to measure the same construct, namely set-shifting. The data suggest that they did not do this. WCST perseverative errors standard score and the trail making shifting standard score were uncorrelated ($r(44) = .04, p = .8$). Thus there was no tendency for those performing poorly (one SD below the

age-standardised mean) on one of these tests to also struggle with the other (chi-squared (1,N=44) = .65, $p = .4$).

Performance on the ID/ED (i.e. whether or not the extradimensional shift was achieved) was related to perseverative errors on the WCST ($r_{pb}(46) = .40$, $p < .01$) but not performance on the trail making measure of shifting ($r_{pb}(44) = .06$, $p = .68$).

Relationships between behavioural measures

There was no significant correlation between IS and social-communication impairment as measured by the 3Di ($r(46) = .20$, $p = .19$). Similarly, an RBS-R summary score, reflecting the full range of RIBAs did not co-vary with the measure of social-communication impairment ($r(46) = .21$, $p = .16$). The measure of IS administered during testing correlated with a crude measure of RIBA collected at the time of clinical assessment ($r(46) = .41$, $p < .01$).

Effects of IQ and age

Table 3 shows the relationships between IQ, age, measures of autistic symptomatology and the measures of cognitive flexibility. Surprisingly, there was a significant relationship between age and WCST perseverative error score, despite the fact that the latter is standardised according to age. This measure also correlated strongly and significantly with verbal, performance and full-scale IQ. The trail making shifting score did not correlate with age, verbal IQ or full scale IQ, but did have a positive relationship with performance IQ.

Table 2 – Performance on measures of cognitive flexibility

	N	Standardised Mean (SD)	Mean (SD)	Range	Percentage 1 SD below standardised mean	Percentage 2 SD below standardised mean
WCST Conceptual Level Response ^a	46	100 (10)	95.8 (15.1)	65 to 136	35	11
WCST Perseverative Errors	46	100 (10)	95.3 (22.7)	55 to 145	46	26
Trail Making Number-Letter Switching ^b	44 ^c	10 (3)	7.7 (3.9)	1 to 14	39	21
Trail Making shifting score	44 ^c	10 (3)	9.4 (3.2)	4 to 19	27	7

^a This is a measure of overall performance on the WCST

^b This is a measure of time taken to complete number-letter switching task

^c Two participants did not complete the trail making test due to motivational factors

Whether or not a participant made the extradimensional shift was related to performance IQ, full-scale IQ and age.

IS score showed no significant correlations with age or any measure of IQ, although its relationship to age did approach significance ($p = .1$). The 3Di measure of social-communication impairment did not correlate with any of the IQ measures, or with age.

Relationships between cognitive measures and measures of autistic symptoms

As can be seen in Table 3, WCST perseverative errors standard score did not correlate with the IS score ($p = .41$), or with the 3Di measure of social-communication impairment ($p = .93$). The correlation between the trail making shifting score and the IS score approached significance using a two-tailed test ($p = .07$). Given that the correlation was in the direction predicted, a one-tailed test was performed, and this did reach significance ($p = .04$). There was no relationship between trail making set-shifting and social-communication impairment ($p = .66$).

Table 3 – Relationships between age, IQ, autistic symptomatology and measures of set-shifting

	PIQ	FSIQ	Age	WCST Perseverative Errors	Trail Making Shifting Score	ID/ED	Insistence on Sameness	Social- communication Impairment
VIQ	.52**	.87**	.02	.50**	-.11	.26	-.19	-.11
PIQ	-	.86**	-.06	.53**	.32*	.50**	-.18	.04
FSIQ	-	-	-.04	.56**	.09	.41**	-.21	-.08
Age	-	-	-	.28*	-.09	.37*	.024	-.25
WCST	-	-	-	-	-.02	.40**	-.12	.01
Perseverative Errors	-	-	-	-	-	-	-	-
Trail Making Shifting Score	-	-	-	-	-	.06	-.27	.07
ID/ED	-	-	-	-	-	-	.13	-.08
Insistence on Sameness	-	-	-	-	-	-	-	.20

* p < 0.05, **P < 0.01

A hierarchical regression was performed to test whether there was a relationship between cognitive flexibility and IS, even when other autistic symptoms were controlled for. IS was entered as the outcome variable, with social-communication impairment being entered in the first block, and trail making shifting score entered in the second. As can be seen in Table 4, the first model was not significantly better at predicting the outcome variable than using the mean IS score. The second model, in which trail making set-shifting was entered, was not significantly better than the first at predicting IS, although the R^2 improvement effected by the addition of this predictor did near significance.

Table – 5 Multiple regression analysis with set-shifting as a predictor of IS whilst controlling for social-communication impairments.

Model	Predictor	R^2 of Model	R^2 Change	F Change	Standardised Beta	T
1	Social-communication	.04	.04	1.77 (p = .19)	.2	1.32 (p=.19)
2	Social-communication Trail Making Shifting Score	.12	.08	3.91 (p = .06)	.22 -.29	1.51 -1.98 (p=.06)

To test the second hypothesis that performance on the ability to make the extradimensional shift on the ID/ED would be related to levels of IS, a point-biserial correlation was used. There was no correlation between these variables ($r_{pb}(46) = .13, p = .40$). Exploratory analysis was carried out to see if performance on the ID/ED correlated with other aspects of autistic symptomatology. There was no relationship with overall social-communication impairment ($r_{pb}(46) = -.08, p = .61$).

Discussion

In a sample of children and young people with an ASD and a normal range verbal IQ we found a broad range of set-shifting abilities with some participants showing seriously impaired performance on these tests, and others demonstrating exceptional ability.

Scores reflecting both RIBAs as a whole and IS more specifically did not correlate with scores for social-communication impairment, supporting the idea that autism is not a unitary construct. Two measures of set-shifting showed no relationship with any autistic symptoms. The third measure does appear to correlate with IS, but not with social-communication impairment. Those who did well on the trail making number-letter switching task had a slight tendency to show less IS.

Interpretation of findings

Before these results can be discussed further the issue of measurement of set-shifting in this study must be addressed. The three measures of set-shifting did not all covary, and had different relationships to the measure of IS. Scores on the WCST and ID/ED, both of which tasks are historically and conceptually related, did correlate. The trail making set-shifting measure had no relationship to either of these. This raises questions about what it was that these tests were actually measuring in this sample, and so complicates interpretation of findings in relation to the hypothesis that difficulty set-shifting would be associated with higher levels of IS, but not greater social-communication impairment.

One way to approach this difficulty is through consideration of test validity in relation to the 'task impurity problem'. According to surface characteristics, the WCST would seem to suffer from greater task impurity than the trail making test: it lacks a condition

in which a range of cognitive processes are controlled for; it involves a conceptual generation component as the participant has to generate a new rule, rather than merely remember what the next letter or number is; it involves the dynamic integration of verbal feedback; and it is not timed, so it allows participants to mask set-shifting difficulties using conceptual thinking. There is empirical evidence that supports this notion that the WCST is a complex task that relies on abilities other than set-shifting. In the current study, WCST scores were highly intercorrelated ($r > .65$) suggesting that the perseverative errors score reflects something more general than just set-shifting, that is also tapped by the conceptual level response and non-perseverative errors score. This impression is lent further credence by the fact that the WCST perseverative error score, unlike the trail making shifting score, correlated with all three measures of IQ in this study. It should be noted that the way IQ was measured in this study (vocabulary, similarities, block design and matrix reasoning sub-tests) had no great set-shifting component. Factor analytic studies on children (Fisher & DeLuca, as cited in Royall et al., 2002, p.391), normal adults (Miyake et al., 2000) and people diagnosed with psychiatric illnesses (Mirsky, 1996) have shown that performance on the task relies on distinct cognitive skills, not just set-shifting. In neuroimaging studies performance of this task is associated with activation of extensive areas of the frontal regions, bilaterally (Royall et al., 2002), which is again suggestive of task impurity.

There is not equivalent evidence on the validity of the less widely used ID/ED, but it can be argued that it suffers from many of the same criticisms as the WCST, as its outcome measure correlated with that of the WCST in this study, and it is similar in form and

concept. Furthermore it had a similar relationship to IQ and age as the WCTS in this sample.

Thus there is an argument, based on face validity, data from the present study, and external findings on test validity, for excluding the WCST, and possibly also the ID/ED, as a measure of set-shifting in this sample. What is the evidence that the trail making test functioned as a test of set-shifting? Clearly the task contains a set-shifting component, and the fact that the test is timed strengthens its case for sensitivity, since set-shifting, especially when not initiated by an external cue, has a temporal cost (Miyake et al., 2000). Furthermore, the use of a subtracted standardised score reflecting performance on the letter and number sequencing tasks controls for motor coordination, visual scanning and other perceptual factors, ability to count and decline alphabet (i.e. to generate the new relevant task set to which to switch), ability to respond to task instructions, motivational factors and age. Nevertheless, this test is not free of the task impurity problem. In performing number-letter switching there is an inhibition component and a strong working memory operation that is not controlled for. Therefore it must be acknowledged that there are ways of doing badly on this test that are not purely related to set-shifting. In part this reflects the nature of set-shifting, which is a complex process relying on various executive functions. This raises the question of whether a test that controlled for working memory function and inhibition would be a meaningful measure of set-shifting, since the ability to shift sets is so dependent on these particular executive functions.

Whilst the finding that set-shifting difficulties are related to higher levels of Insistence on Sameness has intuitive meaning, it should be treated cautiously. In this study three measures of set-shifting were used, but two were later excluded from analyses. Whilst there are strong arguments that their exclusion as valid measures of set-shifting was justified, it must be acknowledged that the selection of one test from three, after data had been analysed, does create an inflated risk of type one error. Furthermore, the correlation between IS and the chosen measure of set-shifting was small to moderate ($r = -.27$) and was significant at the one-tailed but not the two-tailed level. Using multiple regression to control for levels of social-communication impairment, set-shifting was not found to significantly predict IS, but the effect described did approach significance, with the chance of it being real and not a false positive estimated at 94%.

It is worth noting that the discovery of an association between these two variables accords with the findings of Lopez and colleagues (2005) in the only other study of comparable design to this one. They reported a much larger effect ($r = .68$) in their small sample of high-functioning adults. This may reflect the different ages of participants in the two studies. However, a different methodological factor is probably more important, namely the different measurements used, and their relationships to IQ. Lopez and colleagues had a composite cognitive flexibility score, incorporating WCST perseverative errors and a standardised score for time taken to complete the number-letter switching score of the D-KEFS Trail Making test. It is hard to understand why they chose this score from the trail making task, thus denying themselves the opportunity to control for the range of cognitive abilities involved in number and letter sequencing. It is possible that their trail making measure reflects processing speed,

ability to count and decline the alphabet, motor control, motivation and so on. I would expect their measure of cognitive flexibility to correlate highly with IQ, and this does seem to be the case: their measure of cognitive flexibility correlated with verbal IQ ($r = -.81$), performance IQ ($r = -.81$) and full-scale IQ ($r = -.87$). They correlated this cognitive flexibility measure with a score reflecting RIBAs in general, rather than IS. This RIBA score was also highly correlated with IQ (VIQ, $r = -.45$; PIQ, $r = -.40$; FIQ, $r = -.46$). This raises the possibility that the measure of cognitive flexibility used in the study was in part a measure of IQ, and this makes it hard to interpret their findings in relation to cognitive flexibility and RIBAs. This could partly explain the different effect size found in these two studies.

In this study, no significant correlation was found between a measure of IS and a measure of social and communication impairment. This is an interesting finding in relation to the work described above (Ronald et al., 2005), in which a minimal association was reported between ‘social’ (i.e. social-communication impairments) and ‘nonsocial’ (i.e. RIBAs) autistic behaviours. That work was with a community sample, and the current finding is, to my knowledge, the first time that such a dissociation has been reported in a clinical ASD sample. It is particularly noteworthy since it exists despite the fact that the inclusion criteria of this study (ASD diagnosis, meaning that many participants were included because they have the full range of autistic impairments) would work against the detection of this dissociation.

However, two methodological issues are relevant as they potentially impact on the veracity of this non-correlation. It should be noted that these issues are also of relevance

to the other important non-correlation in this study, namely the lack of association between trail-making set-shifting and social-communication impairment.

The first issue concerns the interval between the collection of data on social-communication impairment and IS. The former came from a clinical assessment, whilst the latter was gathered during testing, and the mean interval between these two events was 30 months. Could it be that during this period the levels and nature of autistic behaviour changed sufficiently to undermine a genuine correlation? This question was put to the test, with exploratory analysis in which participants were split into two groups according to the time elapsed between their clinical assessment and participation in the current study. The group with less time elapsed ($r(23) = .08, p = .74$) did not show a stronger relationship between social-communication impairment and IS than the group with more time elapsed ($r(23) = .29, p = .19$). This can be understood in relation to the fact that autism is a lifelong disorder, the severity of which is stable over time (Charman 2003). The way in which autistic traits are measured in the study, and more generally using gold standard instruments like the ADI-R, involves a large proportion of questions about infancy and childhood (as well as items on current behaviour) the answers to which remain stable over time. This is reflected in the lack of correlation between 3Di scores and age reported in this sample. The stability of 3Di scores over time in this sample is further suggested by the fact that measure of IS correlates ($r(46) = .41, p < .01$) with a crude measure of RIBA recorded at the time of the clinical assessment.

The second methodological issue is to do with power. The analysis of IS and social-communication had an n of 46 and the analysis of IS and set-shifting had an n of 44. Consequentially, the study was powered to detect, using correlation, large effects ($r =$

>.49; Cohen, 1988) with a probability of avoiding type two error in the region of 95%, when the probability of a type one error is held at 5%. However, the sample size does not offer acceptable power to detect medium effects: it provides only a 50% chance of avoiding a type two error when $r = 0.3$. Thus on the question of whether there is a large ($r < 0.49$) correlation between IS and social-communication impairment, or a large correlation between IS and set-shifting, the study is able to offer a confident 'no'. However, if the effects we are looking for are medium, or small, it is important to acknowledge that the non-correlations reported here may be false negatives.

This power issue is also of relevance to the findings on the relationship between set-shifting and IS, which approach significance but do not reach it. This tells us that the relationship, if it does exist, is likely to be moderate or small, rather than large.

It is notable that a wide range of executive function abilities were shown by the people with ASD in this study. On the two tests that had been normed on typically developing children and young people, scores ranged from the severely impaired to the exceptional range. Many participants did not have impaired executive functions, as measured by these widely used executive function tests. This is damaging to the executive function theory of autism, as it suggests a lack of universality for these deficits. In this study, it was possible to do well on the WCST which, as has been discussed, draws upon a range of executive functions (generativity, working memory, inhibition as well as set-shifting), and yet to have autism or asperger's syndrome. One way of understanding these data is that there may be subtypes not reflected by these crude diagnostic categories, and that that executive function difficulties could play a role in the development of some but not

all cases of an ASD. The data presented here do not allow for an evaluation of this speculation.

Limitations of the study

Some limitations of this study have already been discussed, such as those to do with power and measurement of set-shifting. Two further issues remain to be considered: the measurement of the IS construct and generalisability.

The specific IS construct described by Cucarro and Szatmari and their colleagues (2003; 2006) has only recently been identified, and there is little precedent for its measurement using any instrument other than the ADI-R (Lord et al., 1994). In this study a scale was constructed from a validated parent-report instrument, which incorporated items thought to map onto IS. These were chosen according to face validity. Validity was then further assessed by checking that the scale related to IQ and age in a similar way to the ADI-R IS scales. Whilst there is nothing to suggest that the IS scale used is not a valid measure of the IS construct, it must be acknowledged that its criterion validity has not yet been assessed. The scale does appear to have internal validity, since its internal consistency was excellent for this sample ($\alpha > .9$).

The participants in this study appear to be representative of those diagnosed with an ASD at the Tier IV hospital they attended, with respect to age and social-communication impairment. However, they may have experienced more severe RIBAs. This difference perhaps reflects a response bias, with young people with more severe RIBAs apparently being more likely to take part in a study investigating these. The sample described here

is not representative of the population of people with an ASD as a whole. As a group they are higher functioning than the ASD population, and had fewer females. This suggests that the findings reported here relate to high-functioning ASDs, but that further research needs to be done to see if they apply to lower functioning groups.

Conclusions

This study sought to investigate the relationship between set-shifting and IS in a group of children and young people with an ASD and verbal IQs in the normal range. There was a significant, small to moderate correlation between set-shifting and IS in the direction predicted when a one-tailed test was used, but this was not significant with a two-tailed test. There was no correlation between set-shifting and social-communication impairment. Furthermore, in this sample, no correlation was found between IS and social-communication impairment.

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Part 3: Critical Appraisal

Critical Appraisal

The study described above had two main findings. Firstly, in accordance with the argument made in my literature review, no correlation was found between social-communication impairments and Insistence on Sameness (IS). Secondly, there was a correlation (significant in the direction predicted at the one-tailed level) between set-shifting ability and IS, but not between set-shifting and social-communication impairment.

I will use this 'critical appraisal' section of my thesis to consider how the design of the study impacts on interpretation of these findings, and briefly to sketch out some implications of these findings for future research. I will also offer some reflections on the experience of carrying out this project.

Strengths and weaknesses of the study

Several limitations of the way in which I investigated the relationship between cognitive flexibility and IS have been described in part two of this thesis. These included the issue of statistical power; the potential lack of generalisability of findings from this high-functioning sample to the broader population of people with an autism spectrum disorder (ASD); the use of a non-standard scale to measure IS and the questions about validity raised by this; the difficulties of measuring cognitive flexibility unconfounded by other cognitive and motivational factors (the 'task impurity problem'); and the interval between measurement of social-cognition and of the other variables in this study. I will now consider two further issues relevant to the validity of the findings of my study: (1) the use of questionnaire data alone to measure my outcome variable (i.e. IS); (2) the fact that those who consented to join the study had higher levels of repetitive behaviours than those who did not opt in.

Questionnaire data

The Repetitive Behaviour Scale – Revised (RBS-R; Bodfish & Lewis, 2000) accords with several of the principles of good questionnaire design (Coolican, 2004). Its items are neither overly complex, nor written in technical language and they offer concrete examples of the behaviours they seek to measure. The questionnaire offers clear descriptions of the categories of repetitive interests, behaviours and activities (RIBAs) being considered. Furthermore, taken together, the items are comprehensive, covering the full range of RIBAs found amongst people with autism. These merits are reflected in the fact that the RBS-R has good internal consistency, adequate interrater reliability, as well as strong face and discriminant validity (Bodfish & Lewis, 2000; Lam & Aman, 2007).

Nevertheless, the RBS-R suffers from a problem inherent in all questionnaires: they reflect the opinion of the person that completed them. It is an obvious point, but one worth making, that the outcome variable in my study was not ‘level of IS’, but rather ‘level of IS as described by one of the participants’ parents’. Clearly this introduces error. One important question is whether this error was systematic. Was there a tendency for parents as a group to exaggerate or minimise the levels of IS of their children? I have no empirical data with which to answer this question, although anecdotal evidence suggests there was not. Some parents gave me the impression that, because their child had been labelled as having an ASD, they were particularly sensitive to noticing IS in their child, and consequently may have inflated that child’s IS score somewhat. By contrast, other parents seemed to have become so accustomed to (and skilled at) structuring family life around their child’s routines and

compulsions that they tended not to notice IS in their child, and so underestimated the extent of this type of behaviour when completing the questionnaire.

One way to diminish this subjectivity problem would have been to seek multiple perspectives on each participant's level of IS. To this end, future studies could supplement parental questionnaire data with teacher questionnaire data, participant self-report data and a direct observational measure. This latter source of measurement could have been made using the Repetitive and Stereotyped Behaviour component of the Autism Diagnostic Observation Schedule – Generic (ADOS-G; Lord et al., 2001), or a variant of the structured play situation designed by Pierce and Courschene (2001) to measure exploration in children with autism. It should be noted that these additional data sources would also have their problems. For example, the authors of the ADOS-G do not include its measurement of RIBAs in the instrument's diagnostic algorithm, as they found it to be uncorrelated to parent report data. They attributed this to the infrequency of some RIBAs, which makes them difficult to measure accurately in the limited time window of an ADOS session (Lord et al., 2001). Furthermore, the ADOS-G is designed to measure RIBA, with IS being only a sub-component of this construct. In consequence, only a handful of ADOS-G items would be relevant to the outcome variable measured in this study.

Regardless of the difficulties inherent in different types of measurement of IS, it would still have been useful to have data from these various perspectives to get a sense of the validity of my IS measure. If scores from parents, teachers, participants and direct observation had been inter-correlated, these could have been summed to create an overall IS outcome variable that would have a stronger claim on validity

than the one used in the current study. If scores had tended not to covary, this in itself would have been interesting data, perhaps reflecting situational factors impacting on levels of observable IS behaviours.

Differences in RIBA between participants and those not opting in

It is reported in the ‘participants’ section of the empirical paper that those who took part in my study had, as a group, higher levels of RIBA than those who were invited to participate but did not opt in. This must be an important fact, pertaining as it does to a collection of behaviours which include IS.

It is interesting to reflect on why this may be the case. I speculate that this sampling bias arises in a large part from the diagnoses given to people who attend the Tier IV service from which participants were drawn, and the implications this had for whether or not they stayed in touch with researchers at the clinic. Individuals with social-communication impairments, but without high levels of RIBAs would have received a diagnosis of ‘Atypical Autism’. They would subsequently not have been asked to take part in research projects based at the clinic, as so much ASD research insists that participants have a full autism or asperger’s diagnosis. Thus they would be more likely to lose touch with the clinic and the researchers attached to it. It is notable that several of the people who did not opt in had moved house and never received my letters. In this study it was rare (n=4) to actually speak to an individual and for them not to opt in, which suggests that much non-participation arose because people did not receive their invitations to join the study. By contrast, people with an autism or asperger’s diagnosis, through continued contact with researchers at the clinic, were more easily contactable, and so more likely to take part in my study.

What are the possible implications of this sampling bias on the findings of my study?

The finding that IS and social-communication were not correlated in my sample is made more convincing by this bias. If this non-correlation exists in a sample particularly rich in RIBAs, we can be confident that it would exist in a sample that included a greater proportion of people with no RIBAs but with high levels of social-communication impairment. The impact of the sampling bias on the other finding of the study (i.e. the moderate correlation between IS and set-shifting) is harder to assess.

Strengths of the study

The main strength of this study arises from the influence on its design of a new and convincing framework for thinking about ASDs. In both the literature review and empirical paper, I presented evidence for a dissociation between the social-communication and nonsocial parts of the autism triad. I have also described the implications that such a dissociation has for the design of studies seeking to find the neurocognitive and genetic underpinnings of autistic symptomatology. The outcome of the current study, in which set-shifting was related to one set of autistic symptoms but not another, confirms the value of this approach. Had I used a traditional case-control design (autism versus non-autism), I would have confounded social-communication impairment and RIBAs: if a difference between the two groups in terms of set-shifting had emerged, it would have been unclear whether this reflected differences in social-communication, differences in RIBAs, or both.

Another strength of the study stems from use of recent developments in the conceptualisation of autistic behaviours. The specific construct of IS was measured,

rather than RIBA as a whole. As described in the introduction, there is evidence that this is the part of RIBA that is distinct from developmental level, and so is more likely to be a construct of interest in autism research, as opposed to research into learning difficulties.

Research implications of findings

Finding one: moderate correlation between set-shifting and IS

Whilst a moderate correlation was found between set-shifting and IS in this study in the direction expected (poorer set-shifting associated with higher IS), it is worth noting that set-shifting was only predictive of eight percent of the variance in IS. Thus, 92% of variance is left to be explained. Some of this will be explicable as measurement error, as has been discussed above. However, it is very likely that there are other more meaningful predictors that could be used to build a better model of IS. Below are some ideas, based on the autism literature and on personal experience gained during this project, as to what other influences on IS might be:

Other executive functions

The possibility remains that executive functions other than set-shifting might be of relevance here. As was described in the introduction to the empirical paper, generativity and planning deficits are found in groups of autistic people (e.g. Ozonoff & Jensen, 1999), and so are potential candidates to be investigated as predictors of IS. However both generativity (Bishop & Norbury, 2005) and planning (Lopez, Lincoln, Ozonoff & Lai, 2005) have been shown not to be correlated to RIBA. Despite this, there are two reasons not to discount these abilities on the basis of these studies. Firstly, neither study was powered to detect medium effects, and so these

findings may be type II errors. Even a well-powered study has a 20% chance of missing a real effect. Secondly, both studies measured RIBAs rather than the narrower construct of IS. It would seem to be worth pursuing the possibility that these executive functions are implicated in IS, using sample sizes adequate to the task of detecting medium or even small effects.

Lopez and colleagues (2005) suggest a further possibility with respect to an executive function account of RIBA: that it may be the pattern of relative strengths and difficulties that is important. They speculate that a combination of strong working memory, along with impaired set-shifting and inhibition is particularly conducive to high levels of RIBAs. This is an interesting and sophisticated approach to the study of the cognitive correlates of autistic symptomatology, and could be promising if pursued in relation to IS. This should be done using a sample big enough to provide data for a multiple regression with four predictors, namely social-communication impairment, plus the three executive functions.

Other cognitive characteristics of people with autism

Of the cognitive theories of autism (aside from the executive function account), the one that suggests possible predictors of IS is the 'Weak Central Coherence' (Frith, 1989) account. This suggests that people with autism tend to focus on the local rather than global features of their environment, meaning that they can have an eye for insignificant detail at the expense of being able spontaneously to grasp 'the bigger picture'. Such a theory appears to have explanatory value with regard to some RIBAs, particularly circumscribed interests. It also possibly relates to insistence on sameness, since a being very aware of small details could partially explain resistance

to alterations in even the minor features of the environment (Turner, 1997).

Furthermore a difficulty in creating global representations of the environment could well lead to an insistence that the environment be kept constant, to minimise confusion, uncertainty and anxiety.

Thus, it would be interesting to see if performance on tests thought to measure central coherence (e.g. the embedded figures task) predict variation in levels of IS.

Situational and emotional factors

When I spoke to parents of participants in this study, it was common for them to describe their own understandings of their child's tendency for an insistence on sameness. One particularly widely held belief was that IS was a way of trying to reduce anxiety in a world that was confusing and daunting. The idea of IS as a response to anxiety was for example evoked by one parent who had taken her son out of secondary school because he had been badly bullied and was deeply unhappy there. She noticed that his need for sameness had reduced since he began home schooling, along with a general improvement in his quality of life and mood. This is an interesting parallel to some accounts of Obsessive Compulsive Disorder (e.g. Salkovskis, Forrester, Richards & Morrison, 1999), in which compulsions and routines are conceptualised as a response to anxiety.

I think that this is an idea worth taking seriously. It is supported by the testimony of some people with autism (e.g. Grandin & Johnson, 2005). Furthermore, there is some empirical evidence with respect to RIBAs (as opposed to IS specifically) that repetition can be used by individuals with autism to modulate their emotions in

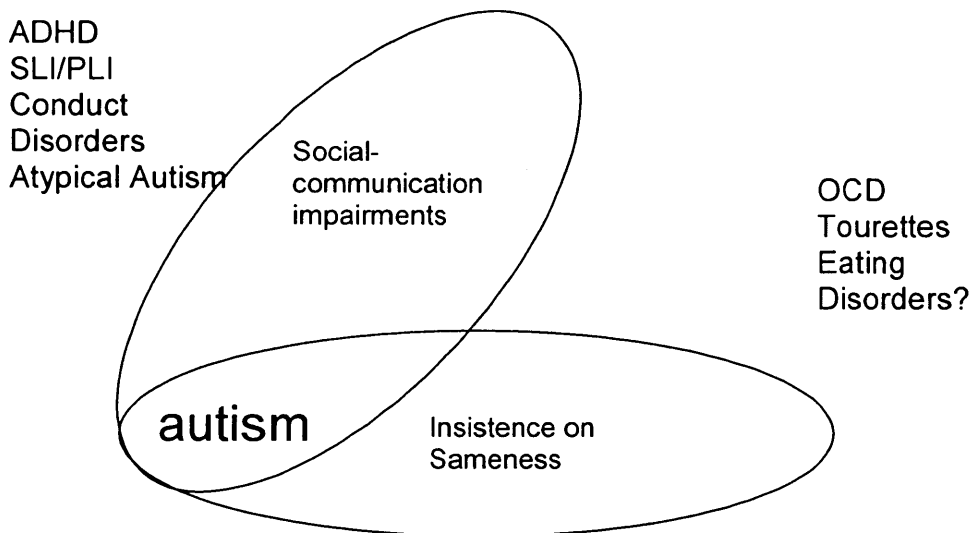
difficult situations (Hutt & Hutt, 1965; Colman, Frankel, Ritvo & Freeman, 1976; both cited in Turner, 1999, p. 841). Clark and Rutter (1981) reported that in highly structured situations autistic children showed fewer RIBAs. Perhaps this reflects the children in this study feeling themselves to be in a predictable environment, and so not needing to impose so many rigid rules of their own.

Thus it is worth having a model of IS that incorporates situational and emotional factors, as well as cognitive, predispositional factors. Perhaps, for any given person at a particular point in time, the level and intensity of their IS behaviours reflects both the predisposition of that individual to this sort of behaviour, and the situation in which they find themselves. By investigating cognitive abilities, this study has concentrated on the predisposition part of this model. It would be an interesting area for further study to incorporate emotional and situational measures when seeking to predict variation in IS.

Finding two: no correlation between IS and social-communication difficulties

Taken together, the literature review and empirical paper presented in this thesis suggest a correlation between IS and social-communication impairment in autistic and non-autistic samples that is at most moderate, and possibly non-existent. This provides us with a working hypothesis that social-communication difficulties and IS are separate dimensions that can exist separately of each other. This is depicted in figure 1. This model has implications for future research. Firstly, it suggests the value of epidemiological investigation of the distribution and prevalence of these

Figure 1 – Two factor model of autism



dimensions in the general population. As described in the literature review, some attempts at this have already been made (Ronald et al., 2006; Ronald et al., 2005). However the validity of the measures used is questionable, and large-scale surveys that convincingly measure autistic social-communication impairment and IS are needed.

If these did indeed identify individuals who had one type of autistic trait but not the other, it would be very interesting to follow these people up. Do they suffer from psychopathologies at a higher rate than others, and if so are particular psychopathologies associated with particular autistic traits? For example, are people who are high in IS, but who do not have social communication problems, more prone to OCD, Tourettes or even eating disorders? Are people higher in social-communication impairment, without IS, prone to conduct disorder when exposed to other risk factors? In fact, Gilmour, Hill, Place and Skuse (2004) have presented

evidence that there is a link between one aspect of social-communication impairment (pragmatic language deficits) and conduct disorder. It would be interesting to know whether these children had other autistic social-communication impairments, and whether or not they had high levels of IS. Work aimed at the investigation of autistic traits in non-autistic populations has the potential to identify risk factors for a range of developmental outcomes. Perhaps we might find that being high in IS, without social-communication impairment, could be an advantage under some circumstances (Baron-Cohen, 2003).

The epidemiological and follow-up studies described would be large and expensive. However, there are ways to investigate autistic traits as risk factors for psychopathology with much smaller samples. Groups could be compared (e.g. OCD versus Depression versus community control) to look for differences in social-communication and IS scores. The advantage of this work is that it would both reflect and serve to evaluate the model of autistic traits described in figure 1. For example, it would be useful to know, if children with OCD have high trait IS, whether they also have social-communication impairments.

Experience of carrying out the research

When carrying out quantitative research into a psychological phenomenon, there is an implicit aspiration that the end result will be that things are made more simple. In this case, I hoped to account partly for the fairly broad collection of motivations, behaviours, beliefs and emotions that comprise IS in terms of a single aspect of neuropsychological functioning. What I found striking is that this reductionist intent, that is particularly prominent during the desk-bound parts of the project (planning,

analysis and writing up), is so thoroughly undermined by the actual experience of being with young people with autism during the data collection phase.

Some children that I met were very particular about their food always being the same each day, on the same plate, eaten at the same chair, with the same cutlery, and yet would really look forward to going to visit new places. Others were the opposite, dramatically influencing their family's holiday plans and general freedom to travel with their fear of new places, whilst blithely eating whatever was put in front of them. Quite a few participants were obsessed with a role-playing game that involved waging wars in a quasi-medieval, virtual world ('Warhammer'). Others were preoccupied by making music, or horse-riding, or transport systems, or cricket. Also, some people in the study who had set-shifting difficulties were not particularly inflexible at all according to their parents. It can be tempting to try to explain away this diversity when seeking to construct a coherent narrative in the language of 'proper science'. However I found that the experience of collecting data, as well as clinical work, was a check against this tendency.

Furthermore, the experience of being with participants and their families helps to raise questions that I did not previously know were important, and that are not marked as such in the academic literature. For example, the effect of IS on quality of life for people with autism and their families is, in my anecdotal experience, large and often detrimental. Some of the people and families I met seemed to be as disabled by IS as they were by social-communication impairments. This is not an issue that has received much attention in the literature, and yet it is probably more

important to service users than the sorts of things that are the addressed in the most high profile research into autism, such as genetic and neuropsychological factors.

Another research question raised by my experiences of carrying out the research related to developing ways of intervening with respect to IS. I noticed that some parents were better able to manage and live with their child's IS than were others. This must be worth investigating, as such research could yield the sort of insights that would be of immediate, practical use to families and young people with an autism spectrum disorder.

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Appendix 1: Ethical approval letter



Institute of Child Health/Great Ormond Street Hospital Research Ethics Committee

The Institute of Child Health
30 Guilford Street
London
WC1N 1EH

05 June 2006

Mr William P. L. Mandy
Trainee Clinical Psychologist
Camden and Islington Mental Health and Social Care Trust
St Pancras Hospital
4 St Pancras Way
London NW1 0PE

Dear Mr Mandy

Full title of study: A cognitive and behavioural conceptualisation of a group of children with Atypical Autism.

REC reference number: 06/Q0508/33

Thank you for your letter received on 26 May 2006, responding to the Committee's request for further information on the above research and submitting revised documentation.

The further information has been considered on behalf of the Committee by the Chairman.

Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation as revised.

Ethical review of research sites

The Committee has designated this study as exempt from site-specific assessment (SSA). There is no requirement for other Local Research Ethics Committees to be informed or for site-specific assessment to be carried out at each site.

Conditions of approval

The favourable opinion is given provided that you comply with the conditions set out in the attached document. You are advised to study the conditions carefully.

Approved documents

The final list of documents reviewed and approved by the Committee is as follows:

<i>Document</i>	<i>Version</i>	<i>Date</i>
Application		14 March 2006
Investigator CV	William P L Mandy	13 March 2006
Protocol	1	10 March 2006
Covering Letter		15 March 2006
Peer Review	Dr Stephen Butler	15 February 2006
Compensation Arrangements	UCL Employer's Liability Insurance	01 August 2005
Questionnaire	Childhood Routines Inventory Adapted from David G Evans	
Questionnaire	Repetitive Behaviour Scale - Revised (RBS-R)	

Letter of invitation to participant	1, to Parents	13 March 2006
Letter of invitation to participant	2, to parents	13 March 2006
Participant Information Sheet	1, Parent/ Guardian	13 March 2006 superseded
Participant Information Sheet	2, Parent/Guardian	13 March 2006 superseded
Participant Information Sheet	3, Young People	13 March 2006 superseded
Participant Information Sheet	4, Young People	13 March 2006 superseded
Participant Information Sheet	4a, Young People	
Participant Information Sheet	3a, Young People	
Participant Information Sheet	2a, Parent/ Guardian	
Participant Information Sheet	1a, Parent/ Guardian	24 May 2006
Participant Consent Form	1, Adult	13 March 2006
Participant Consent Form	2, Adult	13 March 2006
Participant Consent Form	3, Young Persons	13 March 2006
Participant Consent Form	4, Young Persons	13 March 2006
Response to Request for Further Information		
Supervisor CV	Dr Jane D Gilmour	Received 26 May 2006

Research governance approval

You should arrange for the R&D department at all relevant NHS care organisations to be notified that the research will be taking place, and provide a copy of the REC application, the protocol and this letter.

All researchers and research collaborators who will be participating in the research must obtain final research governance approval before commencing any research procedures. Where a substantive contract is not held with the care organisation, it may be necessary for an honorary contract to be issued before approval for the research can be given.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

06/Q0508/33	Please quote this number on all correspondence
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With the Committee's best wishes for the success of this project

Yours sincerely

Enclosures: Standard approval conditions, SL-AC2 for studies other than Clinical Trials of Investigational Medicinal Products

Copy to: Great Ormond Street Hospital/ Institute of Child Health R&D Office

SF1 list of approved sites

Appendix 2: Participant information sheets and consent forms

Institute of Child Health

and Great Ormond Street Hospital for Children NHS Trust

UNIVERSITY COLLEGE LONDON



The
child
first
and
always

18th August 2006

Dear Parents of

30 Guilford Street
London

WC1N 1EH

Tel. No: 020 7831 0975

Fax. No: 020 7831 7050

I am writing to ask if you and [name of child] would like to take part in a research project about Autism Spectrum Disorders that we are running at Great Ormond Street Hospital. The reason I am writing to you is that in the last couple of years, Owen came to Professor David Skuse's clinic at Great Ormond Street Hospital, so we know that he is just the sort of person we are looking to involve in our research. Professor Skuse is supervising this research project.

I have included two information sheets – one for you and one for your child – that explain what the study is about and what it would be like to take part. These are to help you make up your minds about whether you would like to take part. I suggest you read your information sheet first before giving Owen his copy. I would like to stress that you do not have to take part in this study, and whatever decision you make about this will not affect any care your child receives in future.

If you and your child read the information sheets and both decide that you are interested in taking part in the study, I would ask you to fill in the consent forms that I have enclosed with this letter. I would also ask you to fill in the two questionnaires I have sent you. Once you have done this, I would be grateful if you could put them in the stamped, addressed envelope provided, and post them back to me.

Please do not hesitate to get in touch if you or Owen have any questions. You can email me at w.mandy@ucl.ac.uk, or telephone me on 07974 227810.

Yours sincerely,

Will Mandy
Clinical Psychologist in training.



THE QUEEN'S
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2000

Behavioural and Brain Sciences Unit
Neurosciences and Mental Health Directorate

Head of Unit: Professor David Skuse
Professor Peter Hobson Dr Tony Charman
Unit Administrator: Marta Salamonowicz



Habits and interests of young people with and without an Autism Spectrum Disorder

You are being asked to take part in a research project. Please read this sheet, as it tells you why we are doing this project, and what you would have to do if you do decide to take part.

We know you sometimes have worries about getting along with other people. You might also have routines and habits that are very important to you. Sometimes people with these sorts of worries have something called autism, or a similar autistic difficulty. It is OK to have autism. We know lots of children who have autism who are getting along well.

What is research?

Research is a way of finding out new things about the world and the people who live in it. This project aims to find out about habits and interests that some young people have.

Why have I been asked to take part?

In the last couple of years you have been to see the team at Great Ormond Street Hospital. Because we have met you before, we know that you are just the sort of person we want to talk to for our research. We are hoping to meet around 60 children who have been to Great Ormond Street.

Do I have to take part?

No - it is up to you whether you take part or not. We have sent your parent some information about the project. Perhaps you should talk with them about taking part. There is no hurry - you should take your time to decide what you would like to do.

If you decide to take part, and then change your mind later that's OK. You don't have to tell us why you wanted to stop. Whatever you decide about taking part, it won't change anything that happens to you in hospital.

What will I do if I take part in the research?

If you do want to take part, here's what will happen:

Step 1 - We will ask your parent about how you are getting along.

Step 2 - Next, one of our team from Great Ormond Street Hospital would arrange a time to come to your home, to do some puzzles with you. One of these involves using a computer, and another involves drawing with a pencil. It will take about an hour to do the puzzles.

What would be good about taking part?

We cannot give you anything for taking part, but most young people enjoy these puzzles. Also, the things we learn from the research could be useful and may help other children in future.

What if something goes wrong?

We do not expect anything will go wrong, but if it does we will talk to your parents or guardian about what to do.

What will happen to the results of the project?

We hope to write a report so that other people can learn from our research. Your name will not appear in the report.

Thank you for helping us. If you have any questions or worries about the study you can telephone or email the person who is in charge of the project. His name is Will Mandy. His telephone number is and his email is

Great Ormond Street and Institute of Child Health Local Research Ethics Committee has reviewed this project.

Habits and interests of young people with and without an autism spectrum disorder

You and your child are being asked to take part in a research project about autism spectrum disorders (ASDs). Before you decide whether or not to take part, it is important for you to understand why the research is being done and what it involves. Please take time to read the following information carefully.

Please do get in touch if you have any questions. Take your time to decide whether or not you wish to take part.

What is the purpose of the study?

People with ASDs are known to have a number of social difficulties. Some people with ASDs also have other types of difficulties which are not social - for example, some people with an ASD can repeat actions over and over again.

We do not know if these two types of difficulties (social and non-social) are always linked in ASD. This study aims to find out more about this.

The study is being carried out as part of a doctoral training in Clinical Psychology being undertaken by the principal researcher.

Why has my child been chosen to take part in the study?

Your child was assessed at Professor David Skuse's Social Communication Disorders Clinic, at Great Ormond Street Hospital for Children. As a result of this, we know that your child would be just the sort of person we are looking for to take part in the study. We hope to involve around 60 children who have previously been seen at the Social Communication Disorders Clinic.

Do we have to take part?

No - it is up to you and your child to decide whether or not to take part. If you do decide to take part, you are both free to pull out of the research at any time, without giving a reason. Your decisions about taking part will have no effect on the care your child will receive in future.

What will happen to my child and me if we agree to take part?

There are two stages in the study. If you and your child decide that you would like to take part, we will ask for the following:

Step 1: We would ask you to fill in two questionnaires about how your child is getting along. It takes about 20 minutes to complete the two questionnaires. Once they are completed, we would ask you to return them to us in a stamped addressed envelope.

Step 2: Next, one of our researchers from Great Ormond Street Hospital would get in touch to arrange a visit to your home at a time convenient to you and your child. During this meeting we would do some games and puzzles with your child, which will take less than an hour to complete.

What are the possible risks and benefits of taking part?

Although there are no direct benefits involved in taking part, in our experience children usually enjoy doing these sorts of puzzles. The puzzles have been designed especially for children, and one of them involves using a computer. By helping us understand more about ASD, the study will help professionals dealing with it in the future. We are not able to give individual feedback, but on finishing the study, we will send you a general summary of our findings.

What happens to the information collected?

All information collected during the study will be kept strictly confidential. Instead of using your name, we use a code to label the information you give us. A list of names and their codes will be kept separately and securely, so that only the principle researcher can access it. Completed questionnaires will be kept in a locked, secure cabinet. The study will be written up and published as a research paper, but the individuals who took part will not be identifiable from this.

What if something goes wrong?

We do not expect any problems, but we are obliged to tell you the following: If something goes wrong there are no special compensation arrangements available. In the event of any negligence, you may have grounds for legal action but you may have to pay for it. Regardless of

this, if you do have any complaints or worries about the study, the usual National Health Service complaints mechanisms would be available to you.

Ethical Review

Great Ormond Street and Institute of Child Health Local Research Ethics Committee has reviewed this project.

Want to find out more?

If anything written above is unclear to you, or if you or your child would like to find out more, please do not hesitate to get in touch with the project's principal researcher, Will Mandy. You can call him on

Date: 13/03/2006
Version: 3

YOUNG PERSON'S CONSENT FORM

Habits and interests of young people with and without an Autism Spectrum Disorder

Participant ID Number:

Project ID: A1

		Please tick if you agree
1	I have read and understood the information sheet dated 13/03/2006 (version 3) and have asked any questions I wanted to.	
2	I have had enough time to decide if I want to take part in the project.	
3	I understand that I only need to take part if I want to and that I am free to stop doing the project at any time, without having to give a reason.	
4	I understand that the person doing the research (Will Mandy) may look at my hospital notes if they need to. This is OK if my Parent or Guardian lets them.	
5	I agree to take part in this project	

Name of participant	Date	Signature

Comments or concerns during the study

If you have any comments or concerns you should discuss these with the principle investigator **Will Mandy** – 07974 227810. If you wish to go further and complain about any aspect of the way you have been approached or treated during the course of the study, you should write or get in touch with the Complains Manager at Great Ormond Street Hospital. Please Quote the GOSH project number at the top of this form.

Date: 13/03/2006

Version: 1

ADULT CONSENT FORM

Habits and interests of young people with and without an Autism Spectrum Disorder

Participant ID Number:

Project ID: A1

		Please tick if you agree
1	I confirm that I have read and understood the information sheet dated xxx (version y) for the above study and have had the opportunity to ask questions.	
2	I confirm that I have had sufficient time to consider whether or not I want my child and me to be included in the study.	
3	I understand that my child's and my participation is voluntary and that we are free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.	
4	I understand that information collected when my child was assessed at Great Ormond Street Hospital may be looked at by Will Mandy. I give permission for this.	
5	I agree for my child and me to take part in the study	

Please give a telephone number in the box below so that one of our researchers can contact you to arrange an appointment at a convenient time.

Tel:

Name of participant	Date	Signature

Comments or concerns during the study

If you have any comments or concerns you should discuss these with the chief investigator **Will Mandy – 07974 227810**. If you wish to go further and complain about any aspect of the way you have been approached or treated during the course of the study, you should write or get in touch with the Complains Manager at Great Ormond Street Hospital. Please Quote the GOSH project number at the top of this form.

Appendix 3: The Repetitive Behaviour Scale - Revised

REPETITIVE BEHAVIOUR SCALE - Revised (RBS-R)

StudyID#: _____

Date of Birth: ___/___/___

Gender: female male

Today's Date: ___/___/___

Completed by (please circle): Mother Father Other (please specify) _____

Instructions:

Please rate your child's behaviour by reading each of the items listed and then choosing the score that best describes how much of a problem the item is for your child. Be sure to read and score all items listed.

Consider each item in Two Ways:

- 1) Make your ratings based on your **current** observations and interactions with the person. Use the definitions in the box given below to score each item:

0 = behaviour does not occur
 1 = behaviour occurs and is a mild problem
 2 = behaviour occurs and is a moderate problem
 3 = behaviour occurs and is a severe problem

- 2) Make another rating based on whether you have **ever** observed this behaviour in your child in the past, using the following definitions to score each item:

0 = behaviour was never observed in the past
 1 = behaviour was observed in the past

When deciding on a score for each item, consider: (a) how frequently the behaviour occurs (e.g. weekly versus hourly), (b) how difficult it is to interrupt the behaviour (e.g. can be easily redirected versus becomes distressed if interrupted) and (c) how much the behaviour interferes with ongoing events (e.g. easy to ignore versus very disruptive).

I. Stereotyped Behaviour Subscale

(DEFINITION: apparently purposeless movements or actions that are repeated in a similar manner)

1	WHOLE BODY (Body rocking, Body swaying)	Current: 0 1 2 3	Ever: 0 1
2	HEAD (Rolls head, Nods head, Turns head)	Current: 0 1 2 3	Ever: 0 1
3	HAND/FINGER (Flaps hands, Wiggles or flicks fingers, Claps hands, Waves or shakes hand or arm)	Current: 0 1 2 3	Ever: 0 1
4	LOCOMOTION (Turns in circles, Whirls, Jumps, Bounces)	Current: 0 1 2 3	Ever: 0 1
5	OBJECT USAGE (Spins or twirls objects, Twiddles or slaps or throws objects, Lets objects fall out of hands)	Current: 0 1 2 3	Ever: 0 1
6	SENSORY (Covers eyes, Looks closely or gazes at hands or objects, Covers ears, Smells or sniffs items, Rubs surfaces)	Current: 0 1 2 3	Ever: 0 1

Current:

0 = behaviour does not occur

1 = behaviour occurs and is a mild problem

2 = behaviour occurs and is a moderate problem

3 = behaviour occurs and is a severe problem

Ever:

0 = behaviour was never observed in the past

1 = behaviour was observed in the past

II. Self-Injurious Behaviour Subscale

(DEFINITION: movement or actions that have the potential to cause redness, bruising, or other injury to the body, and that are repeated in a similar manner)

7	HITS SELF WITH BODY PART (Hits or slaps head, face, or other body area)	Current:	0	1	2	3
		Ever:	0	1		
8	HITS SELF AGAINST SURFACE OR OBJECT (Hits or bangs head or other body part on table, floor or other surface)	Current:	0	1	2	3
		Ever:	0	1		
9	HITS SELF WITH OBJECT (Hits or bangs head or other body area with objects)	Current:	0	1	2	3
		Ever:	0	1		
10	BITES SELF (Bites hand, wrist, arm, lips or tongue)	Current:	0	1	2	3
		Ever:	0	1		
11	PULLS (Pulls hair or skin)	Current:	0	1	2	3
		Ever:	0	1		
12	RUBS OR SCRATCHES SELF (Rubs or scratches marks on arms, leg, face or torso)	Current:	0	1	2	3
		Ever:	0	1		
13	INSERTS FINGER OR OBJECT (Eye-poking, Ear-poking)	Current:	0	1	2	3
		Ever:	0	1		
14	SKIN PICKING (Picks at skin on face, hands, arms, legs or torso)	Current:	0	1	2	3
		Ever:	0	1		

III. Compulsive Behaviour Subscale

(DEFINITION: behaviour that is repeated and is performed according to a rule, or involves things being done "just so")

15	ARRANGING / ORDERING (Arranges certain objects in a particular pattern or place; Need for things to be even or symmetrical)	Current:	0	1	2	3
		Ever:	0	1		
16	COMPLETENESS (Must have doors opened or closed; Takes all items out of a container or area)	Current:	0	1	2	3
		Ever:	0	1		
17	WASHING / CLEANING (Excessively cleans certain body parts; Picks at lint or loose threads)	Current:	0	1	2	3
		Ever:	0	1		
18	CHECKING (Repeatedly checks doors, windows, drawers, appliances, clocks, locks, etc.)	Current:	0	1	2	3
		Ever:	0	1		
19	COUNTING (Counts items or objects; Counts to a certain number or in a certain way)	Current:	0	1	2	3
		Ever:	0	1		
20	HOARDING/SAVING (Collects, hoards or hides specific items)	Current:	0	1	2	3
		Ever:	0	1		
21	REPEATING (Need to repeat routine events; In / out door, up / down from chair, clothing on/off)	Current:	0	1	2	3
		Ever:	0	1		

22	TOUCH / TAP (Need to touch, tap, or rub items, surfaces, or people)	Current:	0	1	2	3
		Ever:	0	1		

Current:

- 0 = behaviour does not occur
- 1 = behaviour occurs and is a mild problem
- 2 = behaviour occurs and is a moderate problem
- 3 = behaviour occurs and is a severe problem

Ever:

- 0 = behaviour was never observed in the past
- 1 = behaviour was observed in the past

IV. Ritualistic Behaviour Subscale

(DEFINITION: performing activities of daily living in a similar manner)

23	EATING / MEALTIME (Strongly prefers/insists on eating/ drinking only certain things; Eats or drinks items in a set order; Insists that meal related items are arranged in a certain way)	Current:	0	1	2	3
		Ever:	0	1		
24	SLEEPING / BEDTIME (Insists on certain pre-bedtime routines; Arranges items in room "just so" prior to bedtime; Insists that certain items be present with him/her during sleep; Insists that another person be present prior to or during sleep)	Current:	0	1	2	3
		Ever:	0	1		
25	SELF-CARE - BATHROOM AND DRESSING (Insists on specific order of activities or tasks related to using the bathroom, to washing, showering, bathing or dressing; Arranges items in a certain way in the bathroom or insists that bathroom items not be moved; Insists on wearing certain clothing items)	Current:	0	1	2	3
		Ever:	0	1		
26	TRAVEL / TRANSPORTATION (Insists on taking certain routes/paths; Must sit in specific location in vehicles; Insists that certain items be present during travel, e.g., toy or material; Insists on seeing or touching certain things or places during travel such as a sign or store)	Current:	0	1	2	3
		Ever:	0	1		
27	PLAY / LEISURE (Insists on certain play activities; Follows a rigid routine during play / leisure; Insists that certain items be present/available during play/leisure; Insists that other persons do certain things during play)	Current:	0	1	2	3
		Ever:	0	1		
28	COMMUNICATION / SOCIAL INTERACTIONS (Repeats same topic(s) during social interactions; Repetitive questioning; Insists on certain topics of conversation; Insists that others say certain things or respond in certain ways during interactions)	Current:	0	1	2	3
		Ever:	0	1		

V. Sameness Behaviour Subscale

(DEFINITION: resistance to change, insisting that things stay the same)

29	Insists that things remain in the same place(s) (e.g. toys, supplies, furniture, pictures, etc.)	Current:	0	1	2	3
		Ever:	0	1		
30	Objects to visiting new places	Current:	0	1	2	3
		Ever:	0	1		
31	Becomes upset if interrupted in what he/she is doing	Current:	0	1	2	3
		Ever:	0	1		
32	Insists on walking in a particular pattern (e.g., straight line)	Current:	0	1	2	3
		Ever:	0	1		
33	Insists on sitting at the same place	Current:	0	1	2	3
		Ever:	0	1		

34	Dislikes changes in appearance or behaviour of the people around him/her	Current:	0	1	2	3
		Ever:	0	1		

Current:

- 0 = behaviour does not occur
- 1 = behaviour occurs and is a mild problem
- 2 = behaviour occurs and is a moderate problem
- 3 = behaviour occurs and is a severe problem

Ever:

- 0 = behaviour was never observed in the past
- 1 = behaviour was observed in the past

V. Sameness Behaviour Subscale cont...

(DEFINITION: resistance to change, insisting that things stay the same)

35	Insists on using a particular door	Current:	0	1	2	3
		Ever:	0	1		
36	Likes the same CD, tape, record or piece of music played continually; Likes same movie / video or part of movie / video	Current:	0	1	2	3
		Ever:	0	1		
37	Resists changing activities; Difficulty with transitions	Current:	0	1	2	3
		Ever:	0	1		
38	Insists on same routine, household, school or work schedule everyday	Current:	0	1	2	3
		Ever:	0	1		
39	Insists that specific things take place at specific times	Current:	0	1	2	3
		Ever:	0	1		

VI. Restricted Behaviour Subscale

(DEFINITION: Limited range of focus, interest or activity)

40	Fascination, preoccupation with one subject or activity (e.g., trains, computers, weather, dinosaurs)	Current:	0	1	2	3
		Ever:	0	1		
41	Strongly attached to one specific object	Current:	0	1	2	3
		Ever:	0	1		
42	Preoccupation with part(s) of object rather than the whole object (e.g., buttons on clothes, wheels on toy cars)	Current:	0	1	2	3
		Ever:	0	1		
43	Fascination, preoccupation with movement / things that move (e.g., fans, clocks)	Current:	0	1	2	3
		Ever:	0	1		

Thank you for completing this questionnaire!