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Investigating the structure of the restricted, repetitive behaviours and interests domain of autism

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Background: The Restricted, Repetitive Behaviours and Interests (RRBIs) are represented in the DSM-IV and measured by the Autism Diagnostic Interview-Revised (ADI-R) as one of the three homogeneous symptom categories of Pervasive Developmental Disorders. Although this conceptualisation is well accepted in the field, the grouping of symptoms is based primarily on clinical judgment rather than on empirical evidence. **Methods:** The objective of this study was to examine the factor structure of the RRBI domain of autism. Eleven items from this domain of the ADI-R were used in a Principal Components Analysis (PCA). Our sample consisted of 339 individuals with a Best Estimate diagnosis of Pervasive Developmental Disorder (PDD). **Results:** Findings indicate that the RRBI domain is composed of two distinct factors or dimensions*: Insistence on Sameness* (IS) and *Repetitive Sensory and Motor Behaviours* (RSMB). RSMB is negatively correlated with adaptive skills; that is, lower functioning individuals tend to have higher levels of repetitive sensory and motor behaviours. On the other hand, IS is positively correlated with autistic symptoms in the communication and language domain. Further analyses suggest moderate familial aggregation among affected sibling pairs within the IS but not the RSMB factor. **Conclusions:** These results provide evidence for the heterogeneity of the RRBI domain of the ADI-R in terms of both clinical presentation and other correlates. In addition, the IS factor seems to be under familial (presumably genetic) control, while RSMB appears to simply reflect variation in developmental level. **Keywords:** Pervasive developmental disorders, autism, restricted repetitive behaviours and interests, principal components analysis, structure of autism domains, genetic studies. **Abbreviations:** RRBIs: Restricted Repetitive Behaviours and Interests; AD: Autistic Disorder; ADI-R: Autism Diagnostic Interview-Revised; IS: Insistence on Sameness; RSMB: Repetitive Sensory and Motor Behaviours.

Since Kanner's (1943) original description of autism, important developments have taken place within the field. Autistic Disorder (AD) is now the most widely studied disorder among a larger spectrum of disorders called the 'Pervasive Developmental Disorders' (PDDs), which also includes Asperger syndrome, Atypical Autism (PDD-NOS), Rett's disorder, and Childhood Disintegrative Disorder (APA, 1994). A greater appreciation of the clinical manifestations of these spectrum disorders is now possible, though the validity of these distinctions is unclear. All the PDD subtypes as defined in DSM-IV share common impairments in social reciprocity, verbal and non-verbal communication, and a preference for repetitive stereotyped behaviours, interests and activities (APA, 1994). Although this tripartite conceptualisation is well accepted in the field, the grouping of symptoms is based primarily on clinical judgment rather than empirical evidence.

Only a few studies have examined the structure of the social and communication domains (Tanguay, Robertson, & Derrick, 1998; Robertson, Tanguay, L'Ecuyer, Sims, & Waltrip, 1999), or the specific cluster of behaviours usually described as 'Restricted and Repetitive Behaviours and Interests' (RRBIs) (Tadevosyan-Layfer et al., 2003; Shao et al., 2003; Cuccaro et al., 2003). Militerni, Bravacio, Falco, Fico, and Palermo (2002) note that, even though RRBIs are represented in the DSM-IV and measured by the ADI-R as a homogeneous category of behaviour, the individual items are in fact quite heterogeneous in terms of their phenomenology. Therefore, it is important to see if it is possible to decompose the RRBI domain of autism into simpler, more homogeneous, dimensions, or whether it is a single dimension as conceptualised in DSM-IV. One way to accomplish this is to do a factor analysis to see if all the behaviours subsumed under this construct represent a single underlying dimension or whether the items represent multiple domains.

We are aware of only two factor analytic studies specifically on the RRBI domain of autism. The Duke group (Shao et al., 2003; Cuccaro et al., 2003) conducted factor analyses of the ADI-R protocols on overlapping samples of 221 and 207 independent individuals diagnosed with Autistic Disorder using samples of both simplex and multiplex families (families with more than one affected child). Results

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from their analyses indicate that the Restricted, Repetitive Behaviours and Interests domain of the ADI-R can be broken down into two factors. Factor 1, labelled 'Repetitive Sensory and Motor Behaviours and Interests' (RSMB), consists of ADI-R items such as 'Hand and finger mannerisms', 'Repetitive use of objects or parts of objects', 'Unusual sensory interests', 'Other complex mannerisms or stereotyped body movements', and 'Rocking'. Factor 2, labelled 'Insistence on Sameness' (IS), or 'Resistance to Change' (RC), consists of ADI-R items such as 'Difficulties with minor changes in personal routine and environment', 'Resistance to trivial changes in environment', and 'Compulsions/rituals'.

Tadevosyan-Leyfer et al. (2003) also conducted a principal component analysis of a larger pool of 98 variables from all three domains of the ADI-R. Their results indicated that the ADI-R can be described using six clusters (factors) of variables. One of these factors labelled 'Compulsions' includes items 'Unusual preoccupations', 'Resistance to trivial changes in environment', 'Compulsions/rituals', and 'Unusual attachment to objects'. This factor is similar to the IS factor identified by Shao et al. (2003) and Cuccaro et al. (2003). At the same time, Tadevosyan-Leyfer et al. (2003) do not report a distinct factor that resembles the RSMB factor reported by the Duke group.

Based on the existing literature therefore, it is unclear whether the RRBI domain of autism represents 1 or 2 domains. The main objective of our study was to help resolve this issue. A second aim was to examine the association between the empirically derived factors and other variables such as age, gender, IQ, other autistic symptoms, and level of functioning (LOF) that might explain variation in these underlying domains. Finally we wished to investigate any potential familial correlations between sibling pairs on empirically derived factors. This study is the first to investigate the association between empirically derived factors within the RRBI domain and other variables associated with the PDDs.

Methods

Sample

Our sample consisted of 339 individuals with a Best Estimate Committee diagnosis of PDD (see section below for details) who are participating in a study aimed at the identification of genetic causes of autism. Participants were recruited from across Ontario, through physicians and other health care professionals who referred children to this regional diagnostic centre and through parent support groups. Additional participants were recruited across Canada from various sources, including child and family centres and other university centres. Children with identifiable neurological or chromosomal conditions of any type were excluded. For this study, our selected sample consisted of 270 males and 69 females (mean age = 100.79 months, SD = 66.13 months), for a total of 339 individuals with PDD. There were no significant gender differences in age or developmental level. The mean Leiter IQ score was 65.7 (SD = 28.7) and the average age at ADI-R interview was 100.8 months (SD = 66.20). The distribution of subtype diagnoses was autism 74.6%; Asperger syndrome 14.2%, and atypical autism (or PDDNOS) 11.2%.

Best Estimate Committee

All participants were initially assessed using the Autism Diagnostic Interview–Revised (ADI-R) and either the original Autism Diagnostic Observation Schedule (ADOS), or the newer generic version (ADOS-G). A clinician (PS) gave the child an initial 'primary' clinical diagnosis based on this information. Further information was collected from the Vineland Adaptive Behaviour Scales (VABS), the Autism Behaviour Checklist (ABC), and the Arthur Adaptation of the Leiter International Performance Scale (Leiter). Raw data from the ADI-R and ADOS, as well as standard scores from the VABS, Leiter, ABC, and any previous clinical notes (including language, psychological, paediatric/psychiatric, occupational therapy, and school records), were forwarded to three raters considered 'experts' in the diagnosis of autism. These experts had an average of 20 years' experience in the diagnosis and assessment of autism. All information regarding previous diagnosis and any identifying data were removed, thus ensuring that raters were blind to any previous diagnoses, family history, or whether the child came from a family with other potential cases of PDD. Each rater made a diagnosis independently using DSM-IV criteria. Cases for whom there was disagreement regarding the diagnosis were discussed until a consensus diagnosis was reached. Further information regarding the consensus best estimate procedure and its reliability and validity is given in Mahoney et al. (1998).

Since the ADI-R was not constructed to diagnose ASD subtypes, this differentiation had to be made by the Best Estimate Committee using data obtained from the ADI-R, ADOS, and clinical records. Autism was diagnosed according to DSM IV criteria (that is, severe impairments in qualitative social interaction, deficits in verbal and nonverbal communication, and a pattern of restrictive, repetitive behaviours). Asperger disorder was diagnosed in children without clinically significant cognitive and language delay but who still demonstrated the triad of autistic impairments outlined above. The cognitive-language exclusionary criteria were operationally defined as 'an IQ above 70' and 'speaking spontaneously in phrases with a verb by 36 months of age'. According to the DSM-IV, there is no requirement that children with autism have speech delay. In fact, in the DSM-IV, this is only one of 4 possible symptoms in the communication domain. Thus, children with AS or Autism may meet the ADI-R algorithm for autism based on the presence of any of the other three characteristics. In view of the difficulties encountered in strictly applying the DSM-IV criteria for Asperger disorder (Szatmari, Archer, Fisman, Streiner, & Wilson, 1995; Miller & Ozonoff, 1997), if an individual met the criteria for both autism and Asperger disorder, a diagnosis of the latter was given. A review of the cases diagnosed with

Asperger disorder by best estimate showed that all but two of the children also met criteria for autism based on the ADI-R algorithm alone. A diagnosis of Atypical Autism was given when the individual demonstrated fewer than the six total behaviours needed to receive a DSM-IV diagnosis of autism, when the child missed the threshold in one or more of the three domains, or when age of onset was after three years of age.

Measures

Autism Diagnostic Interview Revised (ADI-R). The Revised version of the ADI (Lord, Rutter, & Le Couteur, 1994), the most widely used research measure for the diagnosis of AD, is a standardised semi-structured interview consisting of three major domains: 1) social interaction, 2) nonverbal and/or verbal communication, and 3) restricted, repetitive behaviours and interests. A cut-off point for each of the three domains provides a reliable diagnostic algorithm that accurately discriminates AD from other developmental disorders. The ADI-R is scored using 'current' (within 3 months prior to the interview) and 'ever' (throughout the individual's life) ratings.

Autism Diagnostic Observation Schedule (ADOS). The ADOS (Lord et al., 1989, 2000) is a semi-structured direct assessment of communication, social interaction and play or imaginative use of materials, for individuals suspected of having autism or other pervasive developmental disorders (PDDs). The ADOS consists of four modules, each of which is appropriate for children and adults of differing language levels, ranging from nonverbal to verbally fluent.

Vineland Adaptive Behaviour Scales (VABS). The VABS (Sparrow, Balla, & Cicchetti, 1984) is an interview-based measure, completed with the primary caregiver of the target individual, that assesses adaptive behaviour in the three domains of communication (receptive, expressive, and written), daily living skills (personal, domestic, and community), and socialisation (interpersonal relations, play and leisure time, and coping skills). Scores from domains and sub-domains permit the comparison of specific profiles of adaptive behaviours across different groups.

Leiter International Performance Sales (Levine, 1986). The Leiter is a standard, nonverbal measure of problem solving and learning ability. The Leiter does not require verbal instructions or responses, which makes it appropriate for in individuals with autism, particularly those with low linguistic levels (Tidmarsh & Volkmar, 2003).

Statistical analysis

Based on our objective of clarifying the factor structure of the RRBI domain, 11 items from the RRBI domain of the ADI-R were selected for analyses (see Table 1). Scores based on the diagnostic algorithm of the ADI-R (scores of 3 recoded as 2) were used for statistical analyses. As noted earlier, the ADI-R can be scored using 'current' (within 3 months prior to the interview) or 'ever' (throughout the individual's life) ratings. We report analyses using both current and ever scores. This way, we will be able to compare our results across both time periods and provide stronger support for our findings.

Principal Components Analysis (PCA) is a statistical technique that transforms an original set of variables into a smaller set of uncorrelated variables, often referred to as factors or components that represent most of the information of the original variables (Dunteman, 1989). ADI-R protocols of 339 individuals with a diagnosis of PDD from Simplex, Multiplex, and other (adopted and twins) families were used for the PCA analysis. Note that two preliminary PCAs using only probands or only the other affected siblings (i.e., the non-probands) yielded a similar factor structure. We report results from two PCA analyses, one for 'current' and one for 'ever' scores, using ADI-R protocols of 339

Table 1 ADI-R items (current and ever scores) selected for analyses ($N = 339$, 0 = normal, 2 = abnormal)

	Current scores				Ever scores			
	Frequency in % (range: 0–2)				Frequency in % (range: 0–2)			
ADI-R items from RRBI domain	0	1	2	Mean (Std. Dev.)	0	1	2	Mean (Std. Dev.)
# 70 – Circumscribed interests	44.8	12.1	43.1	.98 (.93)	44.0	8.3	47.8	1.04 (.95)
# 71 – Unusual preoccupations	48.7	11.2	40.1	.91 (.94)	39.8	9.4	50.7	1.11 (.94)
# 72 – Repetitive use of objects or parts of objects	38.6	16.2	45.1	1.06 (.91)	25.1	12.4	62.5	1.37 (.86)
# 73 – Difficulties with minor changes in personal routine and environment	34.2	24.2	41.6	1.07 (.89)	30.7	14.7	54.6	1.24 (.89)
# 74 – Resistance to trivial changes in environment	77.9	11.5	10.6	.33 (.66)	74.3	10.0	15.6	.41 (.75)
# 75 – Compulsions/rituals	60.8	12.4	26.8	.66 (.87)	54.3	10.6	35.1	.81 (.93)
# 76 – Unusual attachment to objects	60.8	14.7	24.5	.64 (.85)	51.6	14.5	33.9	.82 (.90)
# 77 – Unusual sensory interests	30.7	27.4	41.9	1.11 (.85)	26.0	20.9	53.1	1.27 (.85)
# 81 – Hand and finger mannerisms	43.4	15.9	40.7	.97 (9.2)	36.0	10.3	53.7	1.12 (.93)
# 84 – Other complex mannerisms or stereotyped body movements	46.0	19.2	34.8	.89 (.89)	40.7	15.6	43.7	1.03 (.92)
# 85 – Rocking	79.4	8.3	12.4	.33 (.69)	73.5	7.4	19.2	.46 (.79)

individuals with PDD. Two, three, and four-factor solutions were examined. Varimax rotation (Kaiser, 1958) was then used to achieve a simpler structure while maintaining independence between the rotated factors. To evaluate the extent to which the obtained factors were correlated with other variables, we calculated factor scores (Armitage & Colton, 1998) on each of the obtained factors for every child.

To investigate any potential familial relationships, we calculated Intraclass Correlations (ICCs) between sibling pairs on the empirically derived factors. For comparison purposes, the ICC for the original total RRBI domain of the ADI-R was also calculated. For this analysis, only Multiplex families with two or more siblings diagnosed with PDD were included ($n = 237$, excluding the 6 monozygotic twin siblings).

We examined the association between the empirically derived factors and other variables of interest (including adaptive level as measured by VABS) using correlation coefficients. More specifically, Pearson correlations were calculated between factor scores and age at ADI-R interview, Leiter-R IQs, VABS communication, daily living, social skills, and adaptive behaviour composite scores, and ADI-R social domain and communication domain total scores. This analysis was conducted using the total sample but also with males and females separately. Linear regression analyses were used to assess the overall variance in factor scores accounted for by differences in cognitive, adaptive function and ADI-R scores on the two emerged factors. Variables that had a significant association with the factor scores in the univariate correlations estimated above were used for these analyses. Finally, we compared means on the emerged factor scores based on gender and Best Estimate diagnosis (Autism vs. Asperger vs. Atypical).

Results

Table 1 presents descriptive statistics for the individual items (current and ever scores) of the ADI-R used in our analyses. The majority of ratings are either '0' (symptom not present) or '2' (which signifies abnormality) both for current and ever scores. Despite the differences in samples (size, PDD subtypes etc.) the means and standard deviations presented here ($N = 339$ children with PDD) appear to be in line with those presented by Lord et al., 1994 in the original ADI-R paper ($N = 25$ children with AD).

Eleven individual items from the RRBI domain of the ADI-R were used in the factor analysis. By using item factor loadings with absolute value >.40 across both sets of PCA (current and ever scores), we concluded that the Restricted, Repetitive Behaviours and Interests domain of the ADI-R can be best described using two factors: 1) 'Insistence on Sameness' (IS), and 2) 'Repetitive Sensory and Motor Behaviours' (RSMB). For both sets of analyses, although four Eigenvalues were larger than 1.0, the differences between the 2-factor and the 3- or 4-factor solutions were minimal. The third and fourth factors consisted of only 2 and 1 items respectively and little added variance was explained. The selection of the number of factors to extract was also based on conceptual interpretability of the emerged factors. As a result, the 2-factor solution was selected, with 9 out of 11 items loading on the two factors. One item had a loading greater than .40 on both factors. Specifically, item # 70 'Circumscribed Interests' had a loading of .50 (current) and .42 (ever) on the IS factor and a loading of −.38 (current) and −.41 on the RSMB. As a result, this item was dropped from both factors to maintain factors that are independent and uncorrelated with each other.

The two extracted components accounted for 36% (current) and 33% (ever) of the variance. Table 2 presents the factor loadings on IS and RSMB for individuals with a Best Estimate diagnosis of PDD (total $N = 339$).

Table 2 Factor loadings on Insistence on Sameness (IS) and Repetitive Sensory and Motor Behaviours and Interests (RSMB) using 'ever' and 'current' scores ($N = 339$)

	Factor loadings			
ADI-R items from RRBI domain	Factor 1 'IS' *current* scores	Factor 2 'RSMB' *current* scores	Factor 1 'IS' *ever* scores	Factor 2 'RSMB' *ever* scores
73: Difficulties with minor changes in personal routine and environment	**.75**	.08	**.73**	.03
74: Resistance to trivial changes in environment	**.65**	.19	**.66**	.09
75: Compulsions/rituals	**.57**	.05	**.59**	−.01
77: Unusual sensory interests	−.06	**.68**	.00	**.67**
81: Hand and finger mannerisms	−.15	**.64**	−.05	**.64**
85: Rocking	.11	**.52**	.27	**.46**
72: Repetitive use of objects	.17	**.58**	.23	**.45**
84: Complex mannerisms	.13	**.48**	.08	**.41**
70: Circumscribed interests*	.50	−.38	.42	−.41
71: Unusual preoccupations	.22	.38	.36	.24
76: Unusual attachment to objects	.35	.31	.32	.13

Note: These two components accounted for 36% for current scores 33% for ever scores of the variance. *Item 70 loads highly (but differentially) on both factors. As a result, this item was dropped from both factors to maintain factors that are independent and uncorrelated with each other.

Table 3 Zero-order correlations between IS and RSMB factor scores ('current' and 'ever') and Age at ADI-R interview, Leiter IQ, VABS communication skills domain, VABS daily living skills domain, VABS social skills domain, VABS adaptive behaviour composite, ADI-R social domain, ADI-R nonverbal communication, ADI-R verbal communication and ADI-R total communication domain

	Age at ADI-R interview	LEITER IQ score	VABS commun. skills	VABS daily living skills	VABS social skills	VABS adaptive behaviour composite	ADI-R social domain total	ADI-R commun. domain total
IS current	**.12***	**.14***	.10	.01	−.02	.02	.05	**.16****
IS ever	**.23****	.11	**.11***	.00	−.01	.02	.04	**.24****
Males	**.22****	.12	.08	−.01	−.02	.00	.06	**.27****
Females	**.29***	.05	.18	.10	−.02	.08	−.01	.10
RSMB current	**−.21****	**−.35****	**−.41****	**−.42****	**−.36****	**−.41****	**.24****	−.09
RSMB ever	−.08	**−.34****	**−.34****	**−.40****	**−.34****	**−.38****	**.24****	.01
Males	−.08	**−.30****	**−.31****	**−.39****	**−.32****	**−.36****	**.22****	.05
Females	−.08	**−.44****	**−.47****	**−.38****	**−.40****	**−.45****	**.32****	−.07

**Correlation is significant at the .01 level (2-tailed).
*Correlation is significant at the .05 level (2-tailed).

The IS factor includes items that measure resistance to change in personal routine, resistance to changes in environment, and compulsions and rituals. The RSMB factor loads on sensory and repetitive motor behaviours that might reflect self-stimulatory behaviours.

Table 3 presents the zero-order correlations between IS and RSMB (current and ever scores) and the other variables of interest. In general, the higher the IS, the older the child, the better the language abilities and the higher the autistic symptoms in the ADI-R communication domain. On the other hand, the higher the RSMB, the lower the functioning level and the higher the autistic symptoms of social reciprocity of the child. The correlation between the two factors (ever scores) and other variables of interest described above was consistent across gender (see Table 3).

Further analyses were conducted using 'ever' factor scores only, since the use of current scores yielded essentially identical results.

To examine the extent to which the correlated variables predict individual scores on the two emerged factors (IS and RSMB), we used linear regression analyses. For the first regression, the IS factor was the Dependent Variable (DV) and the Independent Variables (IVs) included age (at ADI-R interview), VABS communication, and ADI-R communication total scores (the variables with a significant correlation noted above). These were entered as a block. Results indicated that the model significantly predicted the IS factor ($F_{(4,285)} = 9.04$, $p < .001$), but only accounted for a small amount of the explained variance with an adjusted R^2 value of .11. The only variables that were independently associated with the IS factor score were the VABS communication score ($t_{(285)} = 2.26$, $p < .05$), and ADI-R Communication total score ($t_{(285)} = 2.65$, $p < .01$). For the second regression the RSMB factor score was the DV and the IVs included Leiter IQ score, VABS communication, daily living and social skills scores, and ADI-R social total score, which again were entered as a block. The second model significantly predicted the RSMB factor ($F_{(6,283)} = 10.17$, $p < .001$). Once more, the low value of the adjusted R^2 equal to .17 accounted for only a small amount of the explained variance. Three of the variables were independently associated with the RSMB factor; Leiter IQ score ($t_{(283)} = -2.73$, $p < .01$), VABS daily living score ($t_{(283)} = -3.11$, $p < .01$), and ADI-R social score ($t_{(283)} = 2.23$, $p < .05$). Finally, in order to examine the surprising association between IS and the ADI-R communication domain, we conducted linear regression analysis with IS as the DV and the four sub-domains (C1, C4, C2V, C3V) of the ADI-R communication domain as the IVs. This model significantly predicted the IS factor ($F_{(4,334)} = 8.02$, $p < .001$). Even though the amount of explained variance was small ($R^2 = .09$), the independent association ($t_{(334)} = 2.5$, $p < .05$) of sub-domain C3V 'Stereotyped, repetitive or idiosyncratic speech' is worth noting.

To test for potential familial relationships between sibling pairs we calculated Intraclass Correlations (ICCs) for IS, RSMB, and the original total RRBI domain of the ADI-R. Results indicate a significant ICC only for the IS factor. Specifically, the intraclass correlation for IS was .27 ($p < .01$), and for RSMB .00 ($p > .10$). At the same time, the ICC for the original total RRBI domain of the ADI-R was .16 (not shown; $p = .03$). For comparison purposes we also present ICCs of other studies (see Table 4).

Results from *t*-tests indicated no significant differences on either of the two factor scores between males and females (not shown; $p > .05$ for all). An ANOVA was conducted to compare mean scores on derived factors for Best Estimate diagnosis (Autism vs. Atypical vs. Asperger). Post-hoc tests (Tukey HSD) indicated that individuals with Autism scored significantly higher on the RSMB factor than individuals with Asperger or Atypical, who did not differ. At the same time, individuals with Asperger scored significantly higher on the IS factor than individuals

Table 4 A comparison across existing studies of Intraclass Correlations (ICCs) for sibling pairs with PDD from multiplex families based on sub-domain and factor scores from the RRBI domain of the ADI-R

	Intraclass correlations (ICCs) for sibling pairs				
	Sub-domains from ADI-R		Empirically derived factors from ADI-R		
	Spiker et al. (2002)	Silverman et al. (2002)	Tadevosyan-Leyfer et al. (2003)	Cuccaro et al. (2003)	Current study
Original sub-domains from ADI-R RRBI domain					
D1: Preoccupation/circumscribed interests	.10	.30			
D2: Compulsive adherence to routines rituals	.06	.31			
D3: Stereotyped and repetitive motor mannerisms	−.06				
D4: Preoccupations with part of objects or non-functional elements of materials	.06				
Empirically derived factors from ADI-R RRBI domain					
Insistence on Sameness				.26	**.27**
Repetitive Sensory and Motor Behaviours				.12	**.00**
Compulsions			.24		

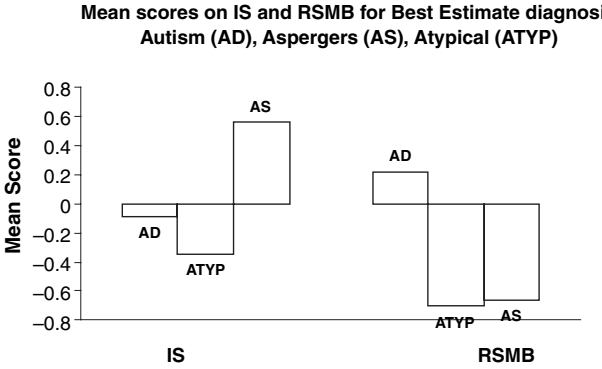

Figure 1 Graphic representation of mean scores on IS and RSMB based on Best Estimate Diagnosis (Autism vs. Atypical vs. Asperger)

with Autism or Atypical, who again did not differ (see Figure 1).

Discussion

Although the presence of Restricted, Repetitive Behaviours and Interests (RRBIs) is one of the three categorical criteria necessary for a PDD diagnosis, few studies have empirically examined this domain, or considered variables that might account for individual differences on this construct. One hypothesis is that the behavioural symptoms grouped in the RRBI domain are heterogeneous and represent more than one construct (Turner, 1999). The main objective of this study was to investigate the factor structure of the ADI-R RRBI domain. To examine this, 11 items (current and ever scores) from this domain were used in a Principal Components Analysis (PCA).

Our findings indicate that the RRBI domain is in fact composed of two underlying factors or dimensions: *Insistence on Sameness* (IS) and *Repetitive Sensory and Motor Behaviours* (RSMB). The results are remarkably similar, if not identical, to those obtained by Shao et al. (2003) and Cuccaro et al. (2003). Not only were the same two factors identified in their studies, but also most of the actual items on each factor and the amount of variance explained by the factors were similar. Specifically, the RSMB factor, composed of ADI-R items 77, 81, 85, 72, and 84, is identical across all three studies (Shao et al. 2003; Cuccaro et al., 2003; current results). At the same time, the three ADI-R items (73, 74, and 75) that make up the Insistence on Sameness (or Resistance to Change) factor identified by Shao et al. (2003) and Cuccaro et al. (2003) are the same three items on the IS factor derived from our analysis. In our study, one additional ADI-R item, item 70, loads highly (but differentially) on both factors. As a result, this item was dropped from both factors to maintain factors that are independent and uncorrelated with each other.

Taken together, these results provide evidence for the heterogeneity of the RRBI domain of the ADI-R. The amount of variance explained in both our study and those of the Duke group is small, however, suggesting that the RRBI domain either includes constructs other than IS and RSMB or else contains considerable measurement error. The behavioural items tend to be difficult to rate; in particular, it can be difficult to decide whether to categorise behaviours as sensory, ritualistic, preoccupations or some combination of each. The question that remains to be answered is whether there are other unidentified constructs in this domain.

In addition to being identified as two separate dimensions of RRBI, IS and RSMB appear to be differentially associated with other variables, adding to the validity of the distinction. The RSMB dimension is negatively correlated with level of adaptive functioning. The more developmentally delayed the child, the more repetitive sensory and motor behaviour observed. Unlike the RSMB factor, the IS factor seems to be related to autistic symptoms in the ADI-R communication domain. Specifically, the higher (more atypical) the ADI-R communication ratings, the more insistence on sameness behaviour is

observed. This may indicate a common substrate between rigidity in behaviour (indexed by IS) and lack of flexibility in communicative skills, as seen in delayed echolalia, repetitive speech, repetitive questions, and verbal rituals. This possibility is supported by the significant independent association between IS and sub-domain C3V (Stereotyped, repetitive or idiosyncratic speech) of the ADI-R Communication domain (see Results section). Again, however, the amount of variance explained in both models is small, suggesting that we have not totally captured variables that account for individual differences in these two factors and that more items are needed to supplement and define the construct. We also emphasise that there are inherent conceptual and measurement problems in studying developmental phenomena. It remains possible, for example, that RSMB and IS are different manifestations of the same or similar underlying processes, with one (RSMB) representing a developmentally less mature but related phenomenon.

Table 4 also presents a comparison across all existing studies that reported Intraclass Correlations (ICCs) for sibling pairs with PDD from multiplex families based on sub-domain and factor scores from the RRBI domain of the ADI/ADI-R. There is clear evidence for familial aggregation of the IS construct or related dimensions but not of the RSMB across studies (Cuccaro et al., 2003; current results). Note further that one study using the IS construct found positive linkage signals on chromosome 15q (Shao et al., 2002).

Possible explanations for the moderate ICCs might be that the there is too much measurement error and/or there is not enough variation to show familial aggregation. Consideration should also be given to the fact that the ADI-R was developed to distinguish autism from Mental Retardation (MR), not to measure severity of symptoms. Therefore, it is unclear whether ADI-R items can be used in a quantitative manner to measure variation along a continuum of severity. Notably, most RRBI items on the ADI-R in our sample were coded as either 0 or 2, essentially absent or present (Table 1), yielding very little quantification of these behaviours. Arguably, the factor scores reflect the breadth of symptoms as opposed to the severity of symptoms.

Clinical and research implications

Our findings, together with those of others (Shao et al., 2003; Cuccaro et al., 2003), provide some support for an empirical reorganisation of the autism RRBI domain into two constructs. We recognise that the two constructs require further study, with a focus on clarifying relevant items, perhaps adding new items and improving measurement approaches to allow quantification of behaviours. These caveats notwithstanding, our ability to decompose the heterogeneous phenotype represented in the RRBI domain might have both clinical and research implications. One possibility is that the use of this construct for diagnosis needs to be reconsidered. If we use the Best Estimate diagnosis (see Methods section) as an external criterion for diagnosing PDDs, our findings indicate that individuals with Autism score significantly higher on the RSMB factor than individuals with Asperger or Atypical. At the same time, individuals with Asperger score significantly higher on the IS factor than individuals with Autism or Atypical (see Figure 1). In general, individuals with Atypical score low on both dimensions. Also, if it is true that the IS domain is more closely related to a lack of flexibility and that the RSMB is more closely related to repetitive behaviour in lower functioning individuals, it may not be appropriate to give equal weight to both in diagnostic algorithms. IS should perhaps carry greater weight in the diagnosis of AS and RSMB in children with autism. Low scores on both would indicate atypical autism. In addition, there are grounds for assuming that RSMB may be useful in differentiating autism plus mental handicap from mental handicap alone. The IS items, in contrast, may have more sensitivity and specificity for high functioning autism and its differentiation from Specific Language Impairment and other learning disabilities such as ADHD. Finally, we have reported (Walker et al., 2004) that in terms of diagnosis, atypical autism (PDDNOS) presents a more homogeneous clinical picture if it focuses on the absence of IS in higher functioning ASD individuals rather than symptoms of RSMB in the overall clinical picture.

Compelling evidence of two distinct dimensions within the RRBI domain could be used in linkage studies aimed at identifying etiologic causes of autism. Our findings demonstrate that the use of the original total RRBI domain of the ADI-R in familiality analysis can be less specific than the use of IS and RSMB. More specifically, the ICC for the original RRBI total domain (ICC = .16) is approximately the average of the ICCs for the two new dimensions, IS (ICC = .27) and RSMB (ICC = .00). If one wanted to focus on phenotypes for genetic studies, then a focus on IS is warranted since only that dimension appears to be under familial control.

Limitations

One major limitation of this study is the reliance on a parent-report measure, notably the ADI-R, to examine RRBI. Even though RRBIs are usually quite intense and easy to observe, a closer and more detailed assessment is needed in order to understand their exact nature (Turner, 1999). Future studies on the issue should make use of questionnaires specifically designed to assess the RRBI domain (e.g., Bodfish, Symons, Parker, & Lewis's RBS-R questionnaire, 2000), and/or methods involving direct observation of the child's behaviour over an extended period. The

whole issue of measurement error in the assessment of these behaviours deserves careful attention. We also note that the ADI-R is not the best way to measure severity of symptoms in AD since it was originally developed to distinguish autism from simple developmental delay. Therefore, it is possible that the same ADI-R items might measure different constructs depending on the level of functioning of the individual being assessed. Another limitation of this study is a potential response-bias problem regarding the ADI-R assessment. As noted earlier, ADI-R algorithm scores range from '0' (symptom not present) to '2' (autistic-like abnormal behaviour). By taking a closer look at Table 1, we can conclude that there appears to be a systematic lower endorsement of the score '1' (representing less frequent/severe autistic-like behaviour), when compared to scores '0' and '2'. This more general issue regarding the scoring of the ADI-R may reflect a bimodal distribution rather than a linear one. Future examinations of the RRBI domain should make use of quantitative measures of both frequency and severity. A final limitation of the current study has to do with the results of the principal components analysis. Specifically, item 70, 'Circumscribed Interests', had a loading of an absolute value greater than .40 (cut-off) on both factors. This might indicate a potential 'link' between the two factors that has something to do with the circumscribed interests of children with ASDs. Even though we decided to exclude this item to maintain the independence of the two factors, future factor analytic studies of the autism RRB domain should look more closely at child behaviour related to circumscribed interests to determine its relation to IS and RSMB.

Taken together, these results provide evidence for the heterogeneity of the RRBI domain of the ADI-R. More importantly, the IS factor seems to be under familial (presumably genetic) control, while RSMB appears simply to reflect variation in developmental level in autism. At the same time, the two distinct dimensions could be informative for clinicians when making a clinical diagnosis. The empirical reorganisation of the autism RRBI domain suggested by our findings may have important implications for attempts to decompose the autism phenotype and improve our understanding of the factors that account for the enormous variation in symptom severity and level of functioning seen in the ASDs.

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