

# Phenotypic Overlap Between Core Diagnostic Features and Emotional/Behavioral Problems in Preschool Children with Autism Spectrum Disorder

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**Abstract** This study examined the phenotypic overlap between core diagnostic features and emotional/behavioral problems in a sample of 335 preschool children with autism spectrum disorder (ASD). Results from principal component analysis (2 components; 49.70% variance explained) suggested substantial phenotypic overlap between core diagnostic features and emotional/behavioral problems. Component I, Emotional Behavioral Repetitive Problems, was independent of the children's intellectual, adaptive functioning, and structural language abilities. Component II, Social Communication Deficits, was negatively related to the children's intellectual, adaptive

functioning, and structural language abilities. Both components were positively related to parental stress. This exploratory study contributes to our understanding of the ASD phenotype and provides further support for including emotional/behavioral problems as part of the clinical characterization of children with ASD.

**Keywords** Autism spectrum disorder · Comorbidity · Emotional/behavioral problems · Phenotype · Principal component analysis

## Introduction

Autism Spectrum Disorders (ASDs; also known as Pervasive Developmental Disorders), are defined in the 4th edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV) as a group of neurodevelopmental disorders characterized by symptoms of social and communication impairment and by the presence of repetitive, restricted, stereotyped behaviours. In the DSM-IV, each of these three distinct categories includes a set of symptoms rated on binary criteria (yes/no) to qualify for a diagnosis (APA 1994, 2000). The current DSM-IV classification distinguishes among three ASD subtypes, namely Autistic Disorder (AD), Asperger's Disorder (AS), and Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS). Many investigators now believe that this conceptualization of ASD, which is based primarily on clinical expertise rather than empirical evidence, is limited in capturing the variability and complexity of clinical presentations of the disorder (Frazier et al. 2008; Georgiades et al. 2007). As a result, the *DSM 5 Neurodevelopmental Disorders Work Group* is proposing the use of symptom *dimensions* in addition to symptom *categories* for the classification of ASD (APA 2010).

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Over the past decade, a better understanding of the basic structure of the core ASD clinical phenotype has emerged from a number of factor analytic studies (Bölte and Poustka 2001; Frazier et al. 2008; Georgiades et al. 2007; Boomsma et al. 2008; Gotham et al. 2008; Robertson et al. 1999; Snow et al. 2009; Szatmari et al. 2002; Tadevosyan-Leyfer et al. 2003; Tanguay et al. 1998; Van Lang et al. 2006; Kamp-Becker et al. 2009). Despite their methodological differences, most studies share at least two major findings: (a) the largest amount of variance in the core ASD clinical phenotype reflects symptoms and/or behaviors related to a single combined social-communication domain; and (b) a smaller amount of additional variance is accounted for by at least one other domain reflecting repetitive, restricted, stereotyped behaviors. Whereas most researchers agree that these two domains are essential to the core ASD phenotypic structure, the optimal number and most importantly the domain content areas needed to best describe the clinical presentation of the disorder warrant further investigation.

Although the aforementioned studies are informative in many ways, they are constrained by at least two methodological limitations. First, the use of a single ASD symptom-based instrument (e.g., the Autism Diagnostic Interview-Revised or Social Responsiveness Scale) may not fully capture the wide range of symptoms and/or behaviors that affect the daily life and development of children with ASD. Second, analyses that include participants across a wide age range (i.e., children, youth, and adults) do not take into account the potential variability in phenotypic structure across developmental stages.

To further our understanding of the associations among symptom and behavior domains in young children with ASD, we examined the underlying structure of a more comprehensive clinical phenotype that, in addition to the core diagnostic ASD features, includes information related to other emotional and behavioral (i.e., internalizing and externalizing) problems observed in a representative sample of newly diagnosed preschool children with ASD. The inclusion of emotional/behavioral problems in our investigation was based on several important reasons drawn from the ASD literature. First, the topic of “comorbidity” between other child disorders and ASD has attracted increasing attention in recent years. Specifically, a number of studies have documented the presence of symptoms/behaviours that are part of other disorders such as attention deficit hyperactivity disorder (ADHD), oppositional defiant disorder (ODD), anxiety and mood disorders in individuals with ASD (e.g., Bauminger et al. 2010; Brereton et al. 2006; Gadow et al. 2004; Witwer and Lecavalier 2008). According to Pandolfi et al. (2009), it is often difficult to distinguish such co-occurring problem behaviors from the core diagnostic features of ASD. Second, these other

emotional/behavioral problems are reported to increase the stress and reduce the quality of life experienced by families of children with ASD (Bauminger et al. 2010; Estes et al. 2009) so understanding the mechanism by which such “comorbidities” arise is important. Third, little is known about the early manifestations of these emotional/behavioral problems in preschool children with ASD, as opposed to their presentation later in childhood or adolescence (Bauminger et al. 2010; Pandolfi et al. 2009).

As noted by Brereton et al. (2006), children and youth with ASD experience high levels of emotional/behavioral problems beyond their core autism symptoms. If there is significant phenotypic overlap (i.e., shared variance) between ASD symptoms and these other emotional/behavioral problems, then use of the term “comorbidity” may be somewhat misleading, at least when describing very young children with ASD. Although the term “comorbidity” was introduced by Feinstein (1970) to describe the simultaneous existence of two or more *distinct* medical disorders in the same individual, it is often used to refer to a greater-than-chance co-occurrence of two or more psychiatric disorders or a specific constellation of symptoms. Thus, it is unclear whether the concomitant observation of these sets of symptoms reflects *distinct clinical entities* or *multiple manifestations of a single clinical entity* (Maj 2005). This important question for the ASD field is deserving of careful empirical investigation.

In a recent study of the association between autistic-like and internalizing traits in a community sample of typically developing twins, Hallett et al. (2009) concluded that there is moderate phenotypic overlap between the two sets of traits. Based on these findings from the general population, Hallett et al. (2009) hypothesized that these traits might be more strongly associated in individuals with clinical levels of autistic symptoms and noted that this question should be examined in clinical samples. To the best of our knowledge, the current study is the first to explore the underlying structural association (and thus potential phenotypic overlap) between core diagnostic features and other emotional/behavioral problems in such a clinical sample of preschool children diagnosed with ASD.

## Methods

### Participants and Procedure

The sample consisted of 335 newly-diagnosed preschool children with ASD participating in the *Pathways in ASD*, a Canadian longitudinal study examining the developmental trajectories of children with ASD (see Table 1 for sample demographic information). With a mean age of 39.8 months (SD = 9.0 months), children met criteria for a clinical

**Table 1** Demographic information for sample ( $N = 335$ )

Characteristic	<i>N</i>	%	Mean	SD
<i>Sex</i>				
Male	284	84.8		
Female	51	15.2		
Child's age at diagnosis (months)			39.8	9.0
<i>Child's age group</i>				
2-year-olds	123	36.7		
3-year-olds	143	42.7		
4-year-olds	69	20.6		
<i>Ethnicity</i>				
Caucasian	240	71.6		
Other	95	28.4		

diagnosis of ASD according to the DSM-IV arrived at by a multi-disciplinary team with expertise in the diagnosis. In addition all cases also met criteria for ASD according to both the Autism Diagnostic Observation Schedule (ADOS; Lord et al. 2002) and the Autism Diagnostic Interview-Revised (ADI-R; Rutter et al. 2003) using the Risi et al. (2006) criteria. To ensure the independence of observations, only one child per family was recruited to the study. The study was approved by the local Research Ethics Boards at each site. Families willing to participate went through an informed consent session prior to joining the study.

Instruments (see Table 2)

*Autism Diagnostic Interview-Revised (ADI-R; Rutter et al. 2003)*

The ADI-R is a standardized semi-structured interview used in the differential diagnosis of ASD. It is designed to be used with a parent or caregiver who is familiar with the developmental history and current behavior of individuals over the age of 2 years. The ADI-R consists of three major domains: (a) language and communication, (b) reciprocal social interaction, and (c) restricted, repetitive, and stereotyped behaviors and interests. A cut-off point for each of the three domains provides a reliable diagnostic algorithm shown to be accurate in differentiating autism from other developmental disorders (Rutter et al. 2003).

*Child Behavior Checklist (CBCL 1.5–5; Achenbach and Rescorla 2000)*

The CBCL/1.5–5 is a well-validated measure of externalizing and internalizing behavior problems in typically-developing preschool children as well as children with ASD (Achenbach and Rescorla 2000; Pandolfi et al. 2009). The CBCL obtains parent/caregiver ratings of 99 problem items that are empirically clustered into 6 domains:

**Table 2** Descriptive statistics for sample ( $N = 335$ )

Instrument scale/subscale scores	Mean	SD	Min.	Max.
ADI-R Social domain score	17.2	5.1	3	29
ADI-R Communication domain nonverbal score	11.9	3.3	0	21
ADI-R Behaviours domain score	5.0	2.2	0	12
RBS-R Stereotyped behavior mean score	5.0	3.3	0	15
RBS-R Self-injurious behavior mean score	2.1	2.9	0	19
RBS-R Compulsive behavior mean score	4.0	3.6	0	18
RBS-R Ritualistic behavior mean score	3.6	3.5	0	18
RBS-R Sameness behavior mean score	5.8	5.6	0	29
RBS-R Restricted behavior mean score	3.9	3.1	0	12
MP-R Developmental index standard score	57.2	25.4	10	136
VABS II Adaptive behavior composite score	73.0	10.1	52	101
PLS-4 Total language standard score	65.3	18.9	50	136

*ADI-R* Autism Diagnostic Interview-Revised, *RBS-R* Repetitive Behavior Scales-Revised, *MP-R* Merrill Palmer-Revised Scales of Development Developmental Index standard score, *VABS II* Vineland Adaptive Behavior Scales Second edition Adaptive Behavior Composite score, *PLS-4* Preschool Language Scale, 4th edition Total Language standard score

Emotionally Reactive, Anxious/Depressed, Somatic Complaints, Withdrawn, Attention Problems and Aggressive Behavior. The CBCL provides raw total scores as well as *T*-scores based on a normative sample of children aged 1.5–5 years. A *T*-score  $\geq 70$  indicates that the child is within “clinical range”, that is above the 98th percentile (Achenbach and Rescorla 2000).

*Repetitive Behavior Scale-Revised (RBS-R; Bodfish et al. 1999, 2000)*

This is an empirically-derived clinical rating scale for measuring the presence and severity of a variety of forms of restricted, repetitive behaviors. The RBS-R is designed to provide a quantitative, continuous measure of the spectrum of repetitive behaviors. It comprises 43 items distributed across six conceptually-derived subscales: Stereotyped Behavior, Self-injurious Behavior, Compulsive Behavior, Routine Behavior, Sameness Behavior and Restricted Behavior. This scale is completed by parent/caregiver informants. Although the RBS-R was designed for somewhat older children, the 6-factor structure was recently validated in our sample with subscale internal consistency ( $\alpha$ ) ranging from .71 to .88 (see Mirenda et al. 2010).

*Vineland Adaptive Behavior Scales, Second Edition (VABS II; Sparrow et al. 2005)*

The VABS II assesses child adaptive behavior in the communication, socialization, daily living skills and motor

domains, and expresses overall functioning in the Adaptive Behavior Composite (ABC) score. The VABS II is administered to a parent or caregiver using a semi-structured interview format.

*Merrill-Palmer-Revised Scales of Development (M-P-R; Roid and Sampers 2004)*

This is an individually-administered measure of intellectual ability that is appropriate for children aged 2–78 months. The Developmental Index standard score comprises cognitive, receptive language and fine motor scales.

*Preschool Language Scale, 4th Edition (PLS-4; Zimmerman et al. 2002)*

This is an individually administered test of receptive and expressive structural language ability designed for children from birth through 6 years. This is a well-researched instrument with good reliability, validity, and utility (Zimmerman et al. 2002). The Total Language standard score was used in the analysis.

*Parenting Stress Index–Short Form (PSI/SF; Abidin 1995)*

This is a parent self-report questionnaire designed to identify potentially stressful parent–child dyadic relationship systems. The instrument yields a Total Stress Score (used in the current study), plus scale scores for components that reflect child and parent characteristics and their interactions that pinpoint sources of stress within the family.

#### Data Analyses

Means and standard deviations of the total raw scores and *T*-scores were calculated to describe the levels/distributions of emotional/behavioral symptoms in the current sample. The percentages of children scoring within the “clinical range” (i.e., *T*-score  $\geq 70$ ;  $\geq 98$ th percentile; Achenbach and Rescorla 2000) on the six CBCL domains were also calculated. To illustrate the differences between the ASD sample and the normative sample of similar age range, effect sizes were computed based on the ratio of the difference of *T*-scores from the two samples and the standard deviation of the normative sample (Achenbach and Rescorla 2000).

To investigate the structure of the ASD phenotype, 15 domain scores – 3 domain total scores from the ADI-R, 6 subscale *T*-scores from the CBCL, and 6 total mean scores from the RBS-R—were used as indicators in principal component analysis (see Table 4 for a list of indicators). Data were available on these indicators for all children in

our sample regardless of their verbal ability. Specifically, the communication sub-domain “B(V): Verbal Total” of the ADI-R that applied only to verbal children (i.e., is language dependent) was excluded from the analysis so that no imputation of the “verbal-only” items was required. Instead, the domain “B(NV): Nonverbal Total” was included as an indicator of nonverbal communication impairment for all children. Principal Component Analysis (PCA) with a Varimax Rotation and Kaiser Normalization was conducted using the 15 indicators. The PCA extraction method was selected over Exploratory Factor Analysis (EFA) because it is intended to reduce data while keeping as much information from the original set as possible (Norris and Lecavalier 2010). Orthogonal (i.e., Varimax) rotation was selected over oblique (i.e., non-orthogonal) rotation because we were interested in deriving components that are distinct from each other. The selected extraction and rotation methods fit well with our long-term analytic plans to use empirically derived components to model the developmental trajectories of children on relatively distinct constructs. To ensure there were no differences in the metrics of the indicators from the four instruments, the analysis was conducted using both original scores and *Z*-scores (i.e., standardized scores). Components were constructed by selecting loadings over .40 (Tabachnick and Fidell 2001).

To examine the potential associations between the derived components and other variables of interest, we calculated partial correlations between the component scores and children’s intellectual abilities (indexed by MP-R), adaptive functioning (indexed by VABS II), and structural language abilities (indexed by PLS-4), as well as parental stress (indexed by PSI-SF). This analysis was conducted while controlling for the effect of children’s ages.

#### Results

Table 3 shows the means and standard deviations of the total raw scores and *T*-scores of the six CBCL problem behavior subscales for the ASD and normative samples. This table also depicts the percentage of children from the ASD sample who scored within the “clinical range” on these domains. Specifically, 39.2% of children with ASD scored in the “clinical range” on the *Withdrawn* subscale, whereas 11.7 and 7.2% scored above the clinical cut-off point on the *Attention Problems* and *Emotionally Reactive* subscales, respectively. The effect sizes for the six subscales ranged from 0.1 to 2.7. Children with ASD scored especially higher than the normative sample on *Withdrawn* (effect size 2.7), *Attention Problems* (effect size 1.1) and *Emotionally Reactive* (effect size 0.8).

**Table 3** Mean and standard deviations of total raw scores and *T*-scores, and percentage of children scoring in the clinical range in the current ASD sample and the CBCL 1.5–5 normative sample (Achenbach and Rescorla 2000)

	CBCL Normative sample ( <i>N</i> = 700)				Pathways ASD sample ( <i>N</i> = 335)					Effect size <sup>b</sup>
	Total raw scores		<i>T</i> -scores		Total raw scores		<i>T</i> -scores			
	Mean	SD	Mean	SD	Mean	SD	Mean	SD	% Within clinical range <sup>a</sup>	
Emotionally reactive	2.4	2.2	54.0	5.7	4.0	3.3	58.3	9.0	7.2	0.8
Anxious/depressed	2.9	2.3	54.2	5.7	3.1	2.6	55.0	6.7	1.5	0.1
Somatic complaints	1.8	1.9	54.0	5.8	2.6	2.4	56.3	7.3	5.4	0.4
Withdrawn	1.5	1.7	54.1	5.8	6.0	2.9	69.6	9.4	39.2	2.7
Attention problems	2.5	1.9	54.1	5.6	4.5	2.3	60.1	8.3	11.7	1.1
Aggressive behavior	10.4	6.9	54.2	6.0	13.2	7.3	56.9	8.6	7.8	0.4

<sup>a</sup> *T*-score ≥ 70 (98th percentile; Achenbach and Rescorla 2000)

<sup>b</sup> Effect sizes were computed based on the ratio of the difference of *T*-scores from the two samples and the standard deviation of the normative sample

PCA results using original indicator scores were no different from those employing standardized scores. Therefore, original scores were used in all subsequent analyses. The following considerations were used to select the most appropriate component solution: (a) Eigenvalues; (b) scree plots; (c) percentage of variance explained; (d) minimum number of item cross-loadings; and (e) clinical interpretability of the derived components. Three Eigenvalues were larger than 1.0; thus a 3-component solution was first examined then rejected because of multiple cross-loadings on two of the components and lack of conceptual interpretability. Results indicated that the structure of the ASD phenotype in this preschool sample was optimally modeled by two distinct components (Table 4). The 2-component solution explained 49.70% of the variance and consisted of the following components: Component I, labeled Emotional Behavioral Repetitive Problems (EBRP), included the ADI-R repetitive behaviours indicator, all the RBS-R indicators, as well as the CBCL indicators with the exception of the Withdrawn subscale. Component II, labeled Social Communication Deficits (SCD), included the ADI-R indicators of social reciprocity and nonverbal communication impairment as well as the CBCL Withdrawn subscale. It should be noted that two indicators, the CBCL Withdrawn and Attention subscales, loaded relatively high on both components. Finally, as expected due to the orthogonal rotation method, the two components were independent of each other ( $p > .05$ ).

The partial correlations between the two derived components and other variables of interest, controlling for the effect of child age (i.e., 2- vs. 3- vs. 4-year-olds), are reported in Table 5. The EBRP component was not significantly correlated with children’s intellectual, adaptive functioning or structural language abilities. In contrast, the

**Table 4** Two-component solution for the structure of the phenotype in a sample of 335 preschool children with Autism Spectrum Disorder

Indicators	Component	
	EBRP	SCD
ADI-R Social domain score	0.03	<b>0.84</b>
ADI-R Communication domain nonverbal score	−0.02	<b>0.69</b>
ADI-R Behaviours domain score	<b>0.42</b>	0.22
RBS-R Stereotyped behavior mean score	<b>0.63</b>	0.27
RBS-R Self-injurious behavior mean score	<b>0.56</b>	0.18
RBS-R Compulsive behavior mean score	<b>0.75</b>	0.05
RBS-R Ritualistic behavior mean score	<b>0.76</b>	0.02
RBS-R Sameness behavior mean score	<b>0.82</b>	0.08
RBS-R Restricted behavior mean score	<b>0.72</b>	0.14
CBCL Emotionally reactive <i>T</i> -score	<b>0.75</b>	0.18
CBCL Anxious/depressed <i>T</i> -score	<b>0.63</b>	0.00
CBCL Somatic complaints <i>T</i> -score	<b>0.69</b>	0.09
CBCL Withdrawn <i>T</i> -score	0.38	<b>0.60</b>
CBCL Attention problems <i>T</i> -score	<b>0.45</b>	0.37
CBCL Aggressive behavior <i>T</i> -score	<b>0.71</b>	0.15

Variance explained: 49.70%; Extraction Method: Principal Components; Rotation Method: Varimax with Kaiser Normalization. *EBRP* Emotional Behavioral Repetitive Problems, *SCD* Social Communication Deficits. Item loadings over .40 are in bold font

SCD component was negatively related to intellectual, adaptive functioning and structural language abilities. Both components were positively related to parental stress.

**Discussion**

The present study explored the phenotypic overlap (i.e., structural associations) between core diagnostic features



**Table 5** Partial correlations between EBRP and SCD component scores and other variables of interest, controlling for the effect of age (2- vs. 3- vs. 4-year-olds)

	Intellectual ability (M-P-R)	Adaptive functioning (VABS II)	Structural language (PLS-4)	Parental stress (PSI-SF)
EBRP component score	-.01	-.11	.05	.50**
SCD component score	-.18**	-.35**	-.17*	.38**

*EBRP* Emotional Behavioral Repetitive Problems, *SCD* Social Communication Deficits, *MP-R* Merrill Palmer-Revised Scales of Developmental Index standard score, *VABS II* Vineland Adaptive Behavior Scales Second edition Adaptive Behavior Composite score, *PLS-4*: Preschool Language Scale, 4th edition Total Language standard score, *PSI-SF* Parenting Stress Index—Short Form Total Stress score

\*  $p < .05$ ; \*\*  $p < .01$

and other emotional/behavioral problems in preschool children with ASD. At least three of the study findings warrant discussion and further investigation.

First, even at this young age, children with ASD present with increased levels of emotional/behavioral symptoms. Not surprisingly, children with ASD have elevated scores on the *Withdrawn*, *Attention Problems*, and *Emotionally Reactive* domains compared to the norms. This finding is in line with those from previous studies (Garon et al. 2009; Sikora et al. 2008). The presence of elevated scores on the CBCL at this young age highlights the need to identify and treat these challenging behaviours early, given evidence of the stability and intractability of these problems (e.g., Richler et al. 2010). Previous research suggests that one of the best ways to support families with children with ASD is to address these problems early by teaching parents how to manage them (e.g., Koegel and Koegel 2006).

Second, the underlying structure of this expanded ASD clinical phenotype can be described using two independent components/domains, Emotional Behavioral Repetitive Problems (EBRP) and Social Communication Deficits (SCD). The SCD component identified in this study is similar in content to the social-communication component reported in numerous previous studies (Kamp-Becker et al. 2009; Snow et al. 2009), including a study by our group (Georgiades et al. 2007). This robust finding of a joint social-communication domain, which appears to hold even in this very young more age-homogeneous sample, has already been incorporated in the proposed revisions of the ASD section of the DSM 5 (APA 2010). The EBRP component identified in the present study includes content from the fixated interests and repetitive behaviours (FIRB) component proposed in the revisions of the DSM 5 committee (APA 2010). However, our EBRP component extends the traditional construct of repetitive behaviors by including other problem behaviours frequently seen in children with ASD such as anxiety, emotional reactivity, somatic complaints, attention problems and aggression. It is worth noting that the CBCL Withdrawn and Attention subscales loaded relatively high on both components. Interestingly, these are the same two indicators on which

children with ASD have significantly elevated scores compared to the norms (see Table 3). Thus, one could hypothesize that these core deficits known to be elevated in children with ASD (Dawson et al. 2004) represent a common underlying theme between the two main phenotypic domains of ASD.

These findings suggest that, at least in very young children with ASD, emotional/behavioral problems should not be described as “comorbid” distinct clinical entities in relation to ASD symptoms. Rather, they can be conceptualized as multiple manifestations of a single clinical entity (Maj 2005), in this case ASD. However, it is difficult to determine from cross-sectional data whether emotional/behavioral problems are truly “comorbid” or “part of” the ASD phenotype. This is especially true within this young age range, when these problems may represent relatively isolated symptoms (commonly seen in young children) rather than full-blown disorders. Accumulating longitudinal data from our study will allow us to examine whether these problems indeed co-vary over time with FIRB and/or SCD, that is, whether they influence one another or have independent trajectories. Substantial co-variation between these domains over time would raise the possibility of a common set of risk factors. Evidence for such a possibility would challenge the widely held assumption that comorbid emotional/behavioral problems represent distinct content domains over and above the core features of the ASD clinical phenotype.

Third, there are differential associations between the two phenotypic components and other variables of interest. Specifically, the SCD component is inversely associated with children’s abilities in the areas of intellect, structural language and adaptive functioning. This finding is in line with those reported in previous studies (Georgiades et al. 2007; Snow et al. 2009) and indicates that in children with ASD lower intellectual, language, and adaptive functioning abilities tend to accompany more social communication deficits. On the other hand, EBRPs appear to be independent of these variables. In general, these differential associations might be due to different etiological mechanisms responsible for each of the two ASD domains. If that is the

case, this finding would provide support to the idea that no single unitary account can explain both social and non-social features of autism (Happé and Ronald 2008). Finally, our finding that both components are associated with parental stress is noteworthy and speaks to the importance of early interventions targeting both core areas of children's symptoms, with the potential of reducing parental stress. As noted by Bauminger et al. (2010), the documented link between parental stress and children's emotional/behavioral problems is crucial and can inform intervention and support programs for parents of children with ASD.

Several limitations call for careful interpretation of the study findings. First, we acknowledge that the indicators used in the initial component analysis do not represent the full scope of the ASD-related phenotype. For example, it could be argued that adaptive functioning, language and intellectual ability are constructs that should be included as “parts of” rather than as “correlates” of the ASD phenotype. We believe that this is a possibility worth investigating further. However, for conceptual as well as practical reasons, we focused our current component analysis on symptoms and problem behaviors rather than on standardized assessments of abilities. Second, our study design (i.e., ASD sample) does not allow us to test whether the two domains (i.e., EBRP & SCD) distinguish ASD from other disorders (e.g., ADHD). Third, since our current data are limited to one time-point, we are unable to examine the temporal stability of the structural association between SCD and EBRP. Fourth, the cross-sectional nature of the data does not allow for “cause-and-effect” examinations regarding SCD and EBRP and the other variables (intellectual and structural language ability, adaptive functioning and parental stress). Fifth, data used in the component analysis were collected using parent report (i.e., ADI-R, CBCL & RBS-R), albeit with different methods (i.e., interview and rating scales), and thus are subject to biases associated with using a single informant. Although ADOS data were available, we chose not to include these in the principal component analysis due to the complex issue created by the administration of different, metrically incompatible modules in our sample. Furthermore, the ADOS calibrated severity metric created by Gotham et al. (2009) would not be appropriate because it is a “total” score and does not allow for the investigation of the structural associations between different domains of ASD symptoms (i.e., SCD & FIRB), a main focus of this paper.

In conclusion, this exploratory study demonstrates that there is substantial phenotypic overlap between core diagnostic features and emotional/behavioral problems in young children with ASD. Study findings add to the existing ASD literature by emphasizing the importance of assessing general emotional/behavioral problems in

conjunction with the core diagnostic symptoms in preschool children with ASD (Bauminger et al. 2010; Duarte et al. 2003; Sikora et al. 2008). Our findings are also consistent with the proposed DSM 5 revisions that recommend the use of two domains (SCD and FIRB) to characterize children with ASD (APA 2010). Furthermore, these findings raise new questions about the potential mechanisms underlying the relationship between repetitive behaviors and emotional/behavioral problems in ASD. Therefore, it is important for future research to examine the developmental course and structural associations of these emotional/behavioral problems in relation to core ASD symptoms. Such an approach would be useful in illuminating the complex and, in our view, under-examined issue of “comorbidity” in individuals with ASD.

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**Conflicts of interest** The authors report no conflicts of interest.

## References

- Abidin, R. (1995). *Parenting stress index* (3rd ed.). Odessa, Florida: Psychological Assessment Resources.
- Achenbach, T. M., & Rescorla, L. A. (2000). *Manual for ASEBA preschool forms & profiles*. Burlington, VT: University of Vermont, Research Center for Children, Youth, & Families.
- American Psychiatric Association. (1994). *Diagnostic and statistical manual of mental disorders, 4th edition (DSM-IV)* (4th ed.). Washington, DC: American Psychiatric Association.
- American Psychiatric Association. (2000). *Diagnostic and statistical manual of mental disorders (4th ed., text revision)*. Washington, DC: American Psychiatric Association.
- American Psychiatric Association. (2010). Report of the DSM 5 Neurodevelopmental Disorders Work Group. <http://www.dsm5.org>. Accessed July 18, 2010.
- Bauminger, N., Solomon, M., & Rogers, S. J. (2010). Externalizing and internalizing behaviors in ASD. *Autism Research*, 26, 101–112.
- Bodfish, J. W., Symons, F., & Lewis, M. (1999). *The repetitive behavior scale: Test manual*. Morganton: Western Carolina Center Research Reports.

- Bodfish, J. W., Symons, F. J., Parker, D. E., & Lewis, M. H. (2000). Varieties in repetitive behavior in autism. *Journal of Autism and Developmental Disorders*, 30, 237–243.
- Bölte, S., & Poustka, F. (2001). Factor structure of the autism diagnostic interview-revised (ADI-R): A study of dimensional versus categorical classification of autistic disorders. *Zeitschrift für Kinder- und Jugendpsychiatrie und Psychotherapie*, 29, 221–229.
- Boomsma, A., Van Lang, N. D., De Jonge, M. V., De Bildt, A. A., Van Engeland, H., & Minderaa, R. B. (2008). A new symptom model for autism cross-validated in an independent sample. *Journal of Child Psychology and Psychiatry*, 49, 809–816.
- Brereton, A., Tonge, B., & Einfeld, S. (2006). Psychopathology in children and adolescents with autism compared to young people with intellectual disability. *Journal of Autism and Developmental Disorders*, 36, 863–870.
- Dawson, G., Toth, K., Abbott, R., Osterling, J., Munson, J., & Estes, A. (2004). Early social attention impairments in autism: Social orienting, joint attention, and attention to distress. *Developmental Psychology*, 40, 271–283.
- Duarte, C. S., Bordin, I. A. S., de Oliveria, A., & Bird, H. (2003). The CBCL and the identification of children with autism and related conditions in Brazil: Pilot findings. *Journal of Autism and Developmental Disorders*, 33(6), 703–707.
- Estes, A., Munson, J., Dawson, G., Koehler, E., Zhou, X., & Abbott, R. (2009). Parenting stress and psychological functioning among mothers of preschool children with autism and developmental delay. *Autism*, 13, 375–387.
- Feinstein, A. R. (1970). The pre-therapeutic classification of comorbidity in chronic disease. *Journal of Chronic Disease*, 23, 455–468.
- Frazier, T. W., Youngstrom, E. A., Kubu, C. S., Sinclair, L., & Rezaei, A. (2008). Exploratory and confirmatory factor analysis of the autism diagnostic interview-revised. *Journal of Autism and Developmental Disorders*, 38, 474–480.
- Gadow, K., DeVincent, C., Pomeroy, J., & Azizian, A. (2004). Psychiatric symptoms in preschool children with PDD and clinic and comparison samples. *Journal of Autism and Developmental Disorders*, 34, 379–393.
- Garon, N., Bryson, S., Zwaigenbaum, L., Smith, I., Brian, J., Roberts, W., et al. (2009). Temperament and its relationship to autistic symptoms in a high-risk infant sib cohort. *Journal of Abnormal Child Psychology*, 37, 59–78.
- Georgiades, S., Szatmari, P., Zwaigenbaum, L., Duku, E., Bryson, S., Roberts, W., et al. (2007). Structure of the autism symptom phenotype: A proposed multidimensional model. *Journal of the American Academy of Child and Adolescent Psychiatry*, 46, 188–196.
- Gotham, K., Pickles, A., & Lord, C. (2009). Standardizing ADOS scores for a measure of severity in autism spectrum disorders. *Journal of Autism and Developmental Disorders*, 39, 693–705.
- Gotham, K., Risi, S., Dawson, G., Tager-Flusberg, H., Joseph, R., Carter, A., et al. (2008). A replication of the autism diagnostic observation schedule (ADOS) revised algorithms. *Journal of the American Academy of Child and Adolescent Psychiatry*, 47, 642–651.
- Hallett, V., Ronald, A., & Happé, F. (2009). Investigating the association between autistic-like and internalizing traits in a community-based twin sample. *Journal of the American Academy of Child and Adolescent Psychiatry*, 48, 618–627.
- Happé, F., & Ronald, A. (2008). 'Fractionable autism triad': A review of evidence from behavioural, genetic, cognitive and neural research. *Neuropsychology Review*, 18, 287–304.
- Kamp-Becker, I., Ghahreman, M., Smidt, J., & Remschmidt, H. (2009). Dimensional structure of the autism phenotype: Relations between early development and current presentation. *Journal of Autism and Developmental Disorders*, 39, 557–571.
- Koegel, R. L., & Koegel, L. K. (2006). *Pivotal response treatments for autism*. Baltimore, MD: Paul H. Brookes.
- Lord, C., Rutter, M., DiLavore, P. C., & Risi, S. (2002). *ADOS: The autism diagnostic observation schedule*. Los Angeles, CA: Western Psychological Services.
- Maj, M. (2005). Psychiatric comorbidity: an artifact of current diagnostic systems? *British Journal of Psychiatry*, 186, 182–184.
- Mirenda, P., Smith, I., Vaillancourt, T., Georgiades, S., Duku, E., et al. (2010). Validating the repetitive behavior scale-revised in young children with autism spectrum disorder. *Journal of Autism and Developmental Disorders* (Epub ahead of print).
- Norris, M., & Lecavalier, L. (2010). Evaluating the use of exploratory factor analysis in developmental disability psychological research. *Journal of Autism and Developmental Disorders*, 40, 8–20.
- Pandolfi, V., Magyar, C. I., & Dill, C. A. (2009). Confirmatory factor analysis of the child behavior checklist 1.5–5 in a sample of children with autism spectrum disorders. *Journal of Autism and Developmental Disorders*, 39, 986–995.
- Richler, J., Huerta, M., Bishop, S. L., & Lord, C. (2010). Developmental trajectories of restricted and repetitive behaviors and interests in children with autism spectrum disorders. *Developmental Psychopathology*, 22, 55–69.
- Risi, S., Lord, C., Gotham, K., Corsello, C., Chrysler, C., Szatmari, P., et al. (2006). Combining information from multiple sources in the diagnosis of autism spectrum disorders. *Journal of the American Academy of Child and Adolescent Psychiatry*, 45, 1094–1103.
- Robertson, J., Tanguay, P., L'Ecuyer, S., Sims, A., & Waltrip, C. (1999). Domains of social communication handicap in autism spectrum disorder. *Journal of the American Academy of Child & Adolescent Psychiatry*, 38, 738–745.
- Roid, G., & Sampers, J. (2004). *Merrill-Palmer-revised scales of development*. Wood Dale, IL: Stoelting Co.
- Rutter, M., LeCouteur, A., & Lord, C. (2003). *ADI-R: The autism diagnostic interview-revised*. Los Angeles, CA: Western Psychological Services.
- Sikora, D. M., Hall, T. A., Hartley, S. L., Gerrard-Morris, A. E., & Cagle, S. (2008). Does parent report of behavior differ across ADOS-G classifications: Analysis of scores from the CBCL and GARS. *Journal of Autism and Developmental Disorders*, 38, 440–448.
- Snow, A. V., Lecavalier, L. A., & Houts, C. (2009). The structure of the autism diagnostic interview-revised: Diagnostic and phenotypic implications. *Journal of Child Psychology and Psychiatry*, 50, 734–742.
- Sparrow, S. S., Cicchetti, D. V., & Balla, D. A. (2005). *Vineland adaptive behavior scales: Second edition (Vineland II) survey interview form/caregiver rating form*. Livonia, MN: Pearson Assessments.
- Szatmari, P., Merette, C., Bryson, S., Thivierge, J., Roy, M.-A., Cayer, M., et al. (2002). Quantifying dimensions in autism: A factor analytic study. *Journal of the American Academy of Child and Adolescent Psychiatry*, 41, 467–474.
- Tabachnick, B. G., & Fidell, L. S. (2001). *Using multivariate statistics* (4th ed.). New York, NY: Harper Collins College Publishers.
- Tadevosyan-Leyfer, O., Dowd, M., Mankoski, R., Winklisht, B., Putnam, S., McGrath, L., et al. (2003). A principal components analysis of the autism diagnostic interview-revised. *Journal of the American Academy of Child and Adolescent Psychiatry*, 42, 864–872.
- Tanguay, P. E., Robertson, J., & Derrick, A. (1998). A dimensional classification of autism spectrum disorder by social communication domains. *Journal of the American Academy of Child and Adolescent Psychiatry*, 37, 271–277.



- Van Lang, N., Boomsma, A., Sytema, S., Bildt, A., Kraijer, D., Ketelaars, C., et al. (2006). Structural equation analysis of a hypothesised symptom model in the autism spectrum. *Journal of Child Psychology and Psychiatry*, *47*, 37–44.
- Witwer, A. N., & Lecavalier, L. (2008). Examining the validity of autism spectrum disorder subtypes. *Journal of Autism and Developmental Disorders*, *38*, 1611–1624.
- Zimmerman, I., Steiner, V., & Pond, R. E. (2002). *Preschool language scale, fourth edition (PLS-4)*. San Antonio, TX: The Psychological Corporation.