Toward specifying pervasive developmental disorder-not otherwise specified. Mandy W, Charman T, Gilmour J, Skuse D. Autism Res. 2011 Feb 4. doi: 10.1002/aur.178. [Epub ahead of print] PMID: 21298812

Towards specifying Pervasive Developmental Disorder – Not Otherwise Specified.

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Running title: PDD-NOS

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Towards Specifying Pervasive Developmental Disorder – Not Otherwise Specified

LAY ABSTRACT

Pervasive developmental disorder – not otherwise specified (PDD-NOS) is the most common and least satisfactory of the PDD diagnoses. It is not clearly defined in the diagnostic manuals, limiting the consistency with which it is used by researchers and clinicians. This in turn limits the amount that we have learnt about people with PDD-NOS. In a sample of 256 young people (mean age = 9.1 years) we aimed to implement a clear, transparent definition of PDD-NOS, and then to describe those receiving this diagnosis (n=66), investigating whether they differed from people with autistic disorder (n=97) and Asperger's disorder (n=93). Groups were compared on measures of core PDD symptomatology, associated autistic features, and intelligence. Contrary to the assumption that PDD-NOS is heterogeneous, almost all (97%) of those with PDD-NOS had one distinct symptom pattern, namely impairments in social communication, without significant repetitive and stereotyped behaviours (RSB). Compared to autistic disorder and Asperger's disorder, they had comparably severe but more circumscribed social communication difficulties, with fewer non-social features of autism, such as sensory, feeding and visuo-spatial problems. These individuals appear to have a distinct variant of autism that does not merely sit at the less severe end of the same continuum of symptoms. The current draft guidelines for DSM-V, which insist on the presence of RSBs for any PDD diagnosis, would exclude such people from the autistic spectrum.

Towards Specifying Pervasive Developmental Disorder – Not Otherwise Specified

ABSTRACT

Pervasive developmental disorder – not otherwise specified (PDD-NOS) is the most common and least satisfactory of the PDD diagnoses. It is not formally operationalised, which limits its reliability and has hampered attempts to assess its validity. We aimed firstly to improve the reliability and replicability of PDD-NOS by operationalising its DSM-IV-TR description and secondly to test its validity through comparison with autistic disorder and Asperger's disorder. In sample of 256 young people (mean age = 9.1 years) we used Developmental, Diagnostic and Dimensional (3Di) algorithmic analysis to classify DSM-IV-TR autistic disorder (n=97), Asperger's disorder (n=93) and PDD-NOS (n=66). Groups were compared on independent measures of core PDD symptomatology, associated autistic features, and intelligence. Contrary to the assumption that PDD-NOS is heterogeneous, almost all (97%) of those with PDD-NOS had one distinct symptom pattern, namely impairments in social communication, without significant repetitive and stereotyped behaviours (RSB). Compared to autistic disorder and Asperger's disorder, they had comparably severe but more circumscribed social communication difficulties, with fewer non-social features of autism, such as sensory, feeding and visio-spatial problems. These individuals appear to have a distinct variant of autism that does not merely sit at the less severe end of the same continuum of symptoms. The current draft guidelines for DSM-V, which mandate the presence of RSBs for any PDD diagnosis, would exclude such people from the autistic spectrum.

Key words: Pervasive Developmental Disorder – Not Otherwise Specified (PDD-NOS); Autistic Disorder; Asperger's Disorder; Autism Spectrum Disorder; Diagnostic and Statistical Manual (DSM).

INTRODUCTION

The pervasive developmental disorders (PDDs) are currently characterised by a triad of behavioural features, namely: (a) impairments in social interaction; (b) impairments in communication; and (c) repetitive and stereotyped behaviour (RSB) (American Psychiatric Association [APA], 2000). As defined in DSM-IV-TR, the main PDDs are autistic disorder (AD), Asperger's disorder (AsD) and pervasive developmental disorder – not otherwise specified (PDD-NOS). The latter is a residual category, designed to encompass people with clinically significant autistic difficulties who do not meet criteria for any other PDD. In recent epidemiological studies PDD-NOS has been shown to be the most common PDD, with approximately double the prevalence of autistic disorder (Baird et al., 2006; Chakrabarti & Fombonne, 2005). Despite this the current draft of DSM-V proposes the abolition of PDD-NOS, suggesting that it be subsumed by a broader 'autism spectrum disorder' category. The current study aims to further our understanding of the consequences and value of this proposal by describing the characteristics of individuals meeting current PDD-NOS criteria.

Despite being more common than AD or AsD, PDD-NOS is the most problematic of the PDD diagnoses and is best considered a 'work in progress' (Towbin, 2005). This both reflects and is perpetuated by confusions about its definition. Unlike AD and AsD, PDD-NOS is not formally operationalised in DSM-IV-TR. Instead it is described within a single paragraph of text, prescribed for people who do not meet criteria for a specific PDD but who have *'a severe and persistent impairment in the development of reciprocal social interaction associated with impairment in either verbal or nonverbal communication skills or with the presence of stereotyped behaviour, interests and activities'* (APA, 2000, p.84).

Furthermore, even this broad definition has been subject to substantive change in consecutive editions of the DSM. In DSM-III (APA, 1980) and DSM-III-R (APA, 1987) residual categories were described, 'Atypical PDD' and 'PDD-NOS' respectively, which were both implicitly identical to the current DSM-IV-TR defined disorder. However a wording change was made at a late stage of the production of DSM-IV (APA, 1994) which meant that impairment in just one area of the autism triad was sufficient for a PDD-NOS diagnosis. Inevitably this made the diagnosis over-inclusive (Volkmar,

Shaffer, & First, 2000), and PDD-NOS criteria had to be revised for DSM-IV-TR in line with the earlier DSM-III-R definition of the disorder.

These imprecisions and uncertainties have implications for the value of the PDD-NOS diagnosis. Unlike for AD and AsD, interrater agreement on PDD-NOS diagnosis is low (K = .18), even amongst expert clinicians working on the same study (Mahoney et al., 1998). This lack of reliability hampers attempts to discover more about the characteristics of individuals qualifying for a diagnosis of PDD-NOS, which in turn limits the category's validity and utility (Kendell & Jablensky, 2003).

A search of the literature using PUBMED identified 27 studies in which an explicit attempt had been made to describe PDD-NOS, in relation to other PDDs, other neurodevelopmental disorders or to typical development. Of these, 11 used the over-inclusive DSM-IV criteria to define their PDD-NOS group (Buitelaar, Van der Gaag, Klin, & Volkmar, 1999; Chakrabarti & Fombonne, 2001; Gadow, DeVincent, Pomeroy, & Azizian, 2004; Gadow, DeVincent, Pomeroy, & Azizian, 2005; Jensen, Larrieu, & Mack, 1997; Koyama & Kurita, 2008; Luteijn et al., 2000; Njardvik, Matson, & Cherry, 1999; Roeyers, Keymeulen, & Buysse, 1998; Serra, Loth, van Geert, Hurkens, & Minderaa, 2002; Serra et al., 2003). Only five used DSM-IV-TR (Matson, Dempsey, & Fodstad, 2009; Scheirs & Timmers, 2009) or DSM-III-R (Allen et al., 2001; Pearson et al., 2006; Serra, Minderaa, van Geert, & Jackson, 1995) criteria, whilst four used idiosyncratic criteria not found in the DSMs (de Bruin, Verheij, & Ferdinand, 2006; de Bruin, Verheij, Wiegman, & Ferdinand, 2006; Lord et al., 2006; Verte et al., 2006). The remaining seven used a mix of criteria, thus including people who met one of several definitions of PDD-NOS and its ICD-10 (World Health Organisation, 1993) near equivalent atypical autism (Koyama, Tachimori, Osada, & Kurita, 2006; Matson, Wilkins, Smith, & Ancona, 2008; Mayes, Volkmar, Hooks, & Cicchetti, 1993; Paul et al., 2004; Serra, Minderaa, van Geert, & Jackson, 1999; Takeda, Koyama, & Kurita, 2007; Walker et al., 2004). In contrast to studies of AD or AsD, diagnoses were never made using operationalised definitions according to scores on well-validated autism assessment tools. Instead, clinician diagnoses were used which limits transparency of case identification, making it unlikely that identical PDD-NOS criteria were being replicated across studies. Furthermore some studies distinguished PDD-NOS from AD without attempting to identify AsD (e.g. Allen et al. 2001;Paul et al., 2004), raising the possibility that some individuals meeting current criteria

for AsD may have been included in their PDD-NOS and AD groups. Given these varied definitions of PDD-NOS and unstandardised methods of case definition, it is unsurprising that few consistent findings have emerged from the PDD-NOS literature.

PDD-NOS and core autistic symptomatology

Whilst it is clear that people with PDD-NOS have milder autistic symptoms than those with autism, as would be expected given the disorders' definitions, there is no agreement in the literature about the precise symptom constellation of PDD-NOS. Several studies (e.g. Lord et al., 2006; Verte et al., 2006) have supported the statement of Buitelaar and colleagues (1999) that PDD-NOS is 'basically a lesser variant of autism' (p.41), characterised by milder impairments in each area of the autistic triad. By contrast, others have suggested that PDD-NOS has a distinct symptom pattern, in which some impairments are similar to those found in autism and others are significantly milder. There is no consensus on what this symptom pattern is. For example, it has been reported that that PDD-NOS is best discriminated from AD by lower levels of RSB (Walker et al., 2004) with the two disorders showing similar levels of social communication impairment. Yet other researchers have observed contrasting symptom patterns, reporting no differences between PDD-NOS and AD in terms of RSB, instead finding that those with PDD-NOS in their sample had milder social (Gadow et al., 2004; Mayes et al., 1993) or communication (Paul et al., 2004) impairments.

IQ and social cognition

Findings on cognition and PDD-NOS are similarly mixed. Walker and colleagues (2004) found that their PDD-NOS (n= 21) sample had higher IQ than those with autism (n=216) but lower IQ than their Asperger's disorder group (n = 23). This phenomenon was only partially observed in a UK epidemeological sample, where IQ differences were found between PDD-NOS and autism (PDD-NOS>AD), but not between PDD-NOS and Asperger's disorder (Chakrabarti & Fombonne, 2001). De Bruin, Verheij and Ferdinand (2006) report a contradictory finding, with significant VIQ differences between PDD-NOS and Asperger's syndrome (PDD-NOS<AsD), but not PDD-NOS and autism.

There is a suggestion that, at the group level, people with PDD-NOS show the characteristic autistic profile on the Wechsler Intelligence Scales for Children of superior block design and inferior

comprehension performance (Koyama et al., 2008; Koyama et al., 2006). Similarly, people with PDD-NOS tend to show autistic-like difficulties with various aspects of social cognition compared to typically developing controls (Serra et al., 2003; Serra et al., 2002; Serra et al., 1995; Serra et al., 1999). It is not clear however whether these difficulties differ in type or severity from those found in AD or AsD.

Associated features, functional adaption and comorbidity

We know of no studies describing in PDD-NOS the sorts of non-core symptoms often associated with autism, such as motor difficulties, sleep disturbance, eating problems and sensory sensitivities. Paul and colleagues (Paul et al., 2004) used the Vineland Adaptive Behavior Scales to assess whether level and nature of adaptive functioning could distinguish PDD-NOS from autism and found specific deficits in communication, but not socialisation or daily living skills.

High rates of psychiatric comorbidity have been described in a large (n=92) sample of Dutch children with PDD-NOS (de Bruin, Ferdinand, Meester, de Nijs, & Verheij, 2007), but these findings are hard to interpret as no control group was used. One study has suggested that those with DSM-IV PDD-NOS are more likely than children with autistic disorder to be diagnosed with oppositional defiant disorder, and that they experience higher rates of generalised anxiety disorder than those with Asperger's syndrome (Gadow et al., 2004). However, these findings should be treated with some caution as they have not been replicated elsewhere (Simonoff et al., 2008; Verte et al., 2006; Weisbrot, Gadow, DeVincent, & Pomeroy, 2005).

In summary, whilst we know that people with PDD-NOS experience milder core autistic symptoms than those with AD, the current literature tells us little of certainty about their profile of core and associated autistic symptomatology, level of intellectual abilities, and comorbid difficulties. Largely this arises from the lack of reliability of the PDD-NOS diagnosis. Thus little is known about the validity and clinical utility of this most common of the PDDs.

The aim of this study is to characterise a group of individuals diagnosed according to a clearly operationalised DSM-IV-TR definition of PDD-NOS. In doing so we aim to contribute to the debate

about the proposal in the DSM-V draft criteria to abolish PDD-NOS. Diagnosis will be assigned using a well-validated PDD assessment instrument, the Developmental, Dimensional and Diagnostic Interview (3Di), in order to promote the reliability, transparency and replicability of diagnosis. To explore the validity of PDD-NOS we offer a comprehensive description of the disorder within a large clinical sample, in relation to AD and AsD, using measures independent to those used to assign diagnosis. Groups were compared according to: (1) core diagnostic symptoms of autism; (2) intelligence; (3) associated features of autism, including auditory sensitivity, motor development, feeding problems and sleep difficulties.

METHODS

Participants

Participants (n=256) were consecutive referrals receiving a comprehensive PDD assessment at a specialist service in London, UK between June 2000 and July 2009. Data were collected by experienced psychiatrists and clinical psychologists as part of a clinical assessment for a high-functioning PDD. As is shown in Table 1, diagnostic groups did not differ according to age or gender.

Ethical approval for the current study was granted after review by the Great Ormond Street Hospital Research Ethics Committee.

[Table 1 here]

Materials

The Developmental, Dimensional and Diagnostic Interview (3Di) All participants were assessed using the 3Di (Santosh et al., 2009; Skuse et al., 2004). This validated, semi-structured, parent-report interview uses a computerised algorithm to combine responses to 122 items, generating dimensional scores expressing degree of impairment in each domain of the autistic triad. This algorithm is designed to generate scores that are equivalent to those derived from the Autism Diagnostic Interview-Revised (ADI-R) with ranges and thresholds for abnormality identical to those of the ADI-R diagnostic algorithm (Lord, Rutter & LeCouteur, 1994). Like the ADI-R, the 3Di uses ICD-10 and DSM-IV-TR diagnostic guidelines for pervasive developmental disorders. Interrater and test-retest reliability of the

3Di are high, yielding intraclass correlation coefficients above 0.86. Agreement between the 3Di and the ADI-R for threshold scores that comprise the ADI-R algorithm is high: 86% for reciprocal social interaction, 100% for communication and 76% for repetitive and stereotyped behaviours. Agreement on caseness with clinician diagnosis is also excellent (positive predictive power =.93, negative predictive power = .91).

In addition to its ADI-R equivalent PDD algorithm, the 3Di also includes the following scales for assessing the types of difficulty commonly associated with PDDs: Fine Motor Impairment, comprising six items (e.g. 'Is he able to tie his shoelaces without help?), Cronbach's $\alpha = .83$; Visio-Spatial Impairment, comprising five items (e.g. 'Has [name] ever had difficulties turning a key the right way to get through a door?'), $\alpha = .82$; Gross Motor Impairment, comprising 3 items (e.g. 'How good is he at kicking a ball that is not moving?'), $\alpha = .72$; Auditory Sensitivity, comprising seven items (e.g. 'Does [name] sometimes put his hands over his ears in response to ordinary sounds?'), $\alpha = .83$; Feeding Difficulties, comprising three items (e.g. 'Does [name] have problems with food that needs chewing?'), $\alpha = .60$; Sleep Problems, comprising 11 items (e.g. 'Does [name] complain of waking up and not being able to get back to sleep?'), $\alpha = .71$. Motor and visio-spatial scale items are scored using a four-point likert scale from 'has no problem' to 'cannot'. Items for the other scales are scored present or absent.

The Autism Diagnostic Observation Schedule (ADOS) This semi-structured observational tool is designed for the assessment of PDDs, and measures reciprocal social interaction and communication impairment, as well as imagination and RSBs (Lord et al., 2000). The standard ADOS algorithm provides scores for reciprocal social interaction and social communication, with higher scores reflecting greater severity and/or frequency of impairment in these areas. Observations of RSBs are also recorded. In the current study the ADOS was scored from video-recordings, administered by psychologists at masters level and above, supervised by research reliable clinical psychologists.

Measures of intelligence. Data on IQ were collected as part of a routine clinical assessment, and as such a variety of measures were used. Instruments included the British Picture Vocabulary Scale (Dunn, Dunn, Whetton C., & Pintillie, 1982), the Wechsler Abbreviated Scale of Intelligence (Wechsler, 1999) and the Wechsler Intelligence Scale for Children – Third (Wechsler, Golombok, & Rust S, 1991) and

Fourth (Wechsler, 2003) editions. Summary variables were computed from these scores, for verbal and performance IQ, standardized to have a mean of 100 and a standard deviation of 15. In the current study verbal and performance IQ data were available for 67% and 58% of the total sample respectively. This does not reflect any tendency to conduct IQ testing depending upon a particular child's presenting difficulties, but rather resulted from changes in clinic practice, with comprehensive psychometric assessment becoming a routine element of assessment after the sampling period of the current study had begun. There were no differences between those with and without IQ data in terms of diagnosis received or degree of autistic symptomatology (all Ps > .36).

Procedure

Participants were classified using the 3Di's PDD algorithm (Skuse et al., 2004). Given the difficulties of differentiating Asperger's disorder (AsD) and autistic disorder (AD) using DSM criteria (Miller & Ozonoff, 1997; Szatmari et al., 2000), Szatmari guidelines (Szatmari, 2000) were used to distinguish these PDDs, according to whether or not a delay in the onset of language was observed. Accordingly, for a diagnosis of AD scores above the standard 3Di cut-points in reciprocal social interaction, communication and RSBs were required, as well as delayed development of onset of single word (>24 months) or phrase speech (>36 months). AsD was diagnosed in the presence of above-threshold 3Di scores for reciprocal social interaction, communication and RSBs, without the delayed development of either single-word or phrase speech.

DSM-IV-TR suggests that PDD-NOS should be diagnosed in the presence of either of two specific, mutually exclusive symptom profiles: (1) impairment in reciprocal social interaction plus impairment in RSBs; or (2) impairment in reciprocal social interaction plus impairment in communication. We call the first of these PDD Social Impairment and Repetitive Behaviour type (PDD SR) and the second PDD Social and Communication Impairment Type (PDD SC). In the current study PDD SR was diagnosed when an individual scored above 3Di threshold for reciprocal social interaction impairment and repetitive behaviour, with a below-threshold score for communication impairment. PDD SC was diagnosed when there were above-threshold 3Di scores for reciprocal social interaction and communication impairment, and a below-threshold score for repetitive and stereotyped behaviour.

Data analysis

When independent variables were normally distributed, group differences were assessed using one-way ANOVAs. When significant, these were followed by Bonferroni corrected post hoc analyses to control for type I error. When the assumption of normality was violated, non-parametric Kruskal-Wallis tests were used, followed with post hoc Mann-Whitney tests comparing PDD-NOS to AD and AsD. Effect sizes were calculated using Cohen's d. To examine the relative frequency of repetitive behaviours within diagnostic groups relative risks were calculated, expressing the chance of a behaviour being present in AD and AsD compared to PDD-NOS. Ninety-five percent confidence intervals were calculated for all effect sizes and relative risks.

RESULTS

Diagnostic features of PDD-NOS

In the current study there were two symptom patterns on the 3Di's diagnostic algorithm which warranted a PDD-NOS diagnosis. The very great majority of the PDD-NOS sample (64 of 66; 97%) had PDD Social and Communication Impairment Type (PDD SC), showing clinically significant impairments in reciprocal social interaction and communication, without high levels of RSB. Only two had PDD Social Impairment and Repetitive Behaviour Type (PDD SR). Both of these had very marginally sub-threshold communication impairments, scoring within less than a quarter of a point of the 3Di's threshold for abnormality on the communication scale. By contrast, as is shown in table 2, the PDD-NOS group mean (1.77, SD = 1.18) for RSB was comfortably below the threshold for abnormality of 3 on the 3Di's RSB scale. 3Di triadic scores, standardised as a percentage of the scale maximum, are depicted in Figure 1.

[Table 2 here]

[Figure 1 here]

Core autistic symptomatology

Of the total sample, 20 (7.8%) were assessed with Module 1, 36 (14.1%) with Module 2, 155 (60.1%) with Module 3 and 45 (17.6%) with Module 4. For analyses combining data from different ADOS modules, all algorithm scale scores were standardised as percentages of the scale maximum.

ADOS scores for social communication, reciprocal social interaction and RSB, presented in Table 2, were all non-normally distributed (all Kolmogorov-Smirnov $p<.01$), and so non-parametric analyses were used. Kruskal-Wallis tests revealed that the clinical groups did not differ significantly according to the ADOS measures of reciprocal social interaction ($\chi2(2)=2.77$, $p=.25$) or Social Communication ($\chi2(2)=3.08$, $p=.22$). By contrast the groups did differ according to repetitive, stereotyped behaviour ($\chi2(3)=8.04$, $p=.018$), with the PDD-NOS group having less RSB than the AD group ($U=2018$, $z=2.74$, $p=.006$). Differences between PDD-NOS and AsD for RSB were not significant ($U=1791$, $z=-1.18$, $p=.24$). Similarly, AsD and AD did not differ on ADOS measures of RSB ($U=3839$, $z=-1.57$, $p=.12$).

Repetitive and Stereotyped Behaviours

Table 3 shows the frequency with which clinically significant repetitive and stereotyped behaviours were reported in each of the three diagnostic groups. Each RSB was rarer in PDD-NOS than in AD or AsD. In particular, sensory interests (e.g. inspecting objects from unusual angles, enjoyment of spinning objects, having infant-like sensory interests) were rare amongst those with PDD-NOS in absolute and relative terms. Routinised behaviour was infrequently reported amongst the PDD-NOS group compared to AD and AsD. Also, the tendency 'endlessly and exactly' to repeat words was much rarer in PDD-NOS. By contrast, focused interests were reported for nearly half of the PDD-NOS participants, although these were still significantly less common than in AD or AsD.

[Table 3 here]

Comparison of associated features of autism

Table 4 shows mean group scores on 3Di scales for a range of associated features of autism. Data were not normally distributed (all Kolmogorov-Smirnov $p<.001$) due to positive skew, so Kruskal-Wallis tests were used, followed by post hoc Mann-Whitney analyses. For gross motor and fine motor impairment and sleep problems there were no group differences. By contrast, on measures of visio-spatial impairment, auditory sensitivity, and feeding difficulties the PDD-NOS group were less impaired that the AD (all $ps<.018$) and AsD groups (all $ps<.045$), with effect sizes in the moderate to large range (Cohen's d .33 to .60). AD and AsD did not differ on these measures of associated autistic features (all $ps>.14$).

[Table 4 here]

Verbal IQs (VIQ) were available for 171 individuals, 67% of the sample. Performance IQ data were available for 148 individuals, 58% of the sample. Group VIQ and PIQ means and standard deviations are shown in Table 1. One-way ANOVA revealed the presence of significant VIQ differences according to group ($f(2,168)=11.74$, $p<.001$). Post hoc Bonferroni tests revealed that the PDD-NOS group did not differ from the AD group ($p=.29$) on VIQ. The AsD group had a higher VIQ than the PDD-NOS ($p=.024$) and AD ($p<.001$) groups. There were also group differences for PIQ

(f(2,145)=5.23, p=.006), although post hoc analyses showed that PDD-NOS did not differ from AD (p>.99) or AsD (p=.06) on this measure of non-verbal intelligence. The AsD group had higher PIQ than those with AD (p=.008). As is shown in Table 1, the proportion of participants scoring below 70 for VIQ ($\chi 2(2)=4.95$, p=.08) or for PIQ ($\chi 2(2)=1.78$, p=.41) did not differ according to group in this sample.

DISCUSSION

Pervasive developmental disorder – not otherwise specified is a residual category designed to encompass individuals with serious autistic difficulties who do not meet full criteria for a specified PDD. As such it is generally assumed to be heterogeneous and lacking in validity. To test these assumptions, DSM-IV-TR guidelines were operationalised with two mutually exclusive symptom profiles on the 3Di warranting a PDD-NOS diagnosis: (a) social impairment and repetitive and stereotyped behaviour without communication impairments (PDD SR) and (b) social and communication impairment without repetitive stereotyped behaviour (PDD SC). PDD SR was exceptionally rare, with the very great majority (97%) of PDD-NOS cases fulfilling criteria for PDD SC. This parent-reported symptom profile was also observed in independent, observational (ADOS) data. Thus, counter to the assumption that PDD-NOS is heterogeneous, almost all PDD-NOS cases in the current study had a clearly identifiable symptom pattern of social communication impairment without abnormal levels of repetitive stereotyped behaviour (RSB). These individuals had a more circumscribed social communication disorder than those with AD or AsD, since they had severe social and communication impairments, with minimal or reduced levels of the non-social aspects of the autism syndrome, such as RSB and diverse associated autistic features. In particular, the participants with PDD-NOS showed fewer sensory interests and less routinised, repetitive behaviour than those with other PDDs.

Individuals with PDD SC have been described in the literature before, albeit in relation to small numbers of cases diagnosed according to the over-inclusive DSM-IV PDD-NOS definition. Walker and colleagues (2004) described 11 children with PDD SC, comprising 50% of their clinician diagnosed PDD-NOS sample. Tanguay, Robertson and Derrick (1998) noticed that of their sample of 17 children diagnosed with PDD-NOS ‘almost all...had marked abnormalities in the first two DSM-IV autism

domains [i.e. reciprocal social interaction and communication] but not in the third [repetitive and stereotyped behaviours]' (p.274). Our findings confirm that this phenomenon persists in a large sample of PDD-NOS when both parent-report and direct observational methods are used, and stricter DSM-IV-TR criteria are applied.

There is an ongoing debate about whether PDD-NOS is simply a 'less severe form of autism' or if at least some PDD-NOS cases have a distinct symptom pattern (Buitelaar et al., 1999). According to parent report (3Di), comparison of AD and PDD-NOS on RSB yielded an effect size that was at least double the effect sizes obtained from comparison of these groups on social and communication impairment. A similarly discrepant ratio of effect sizes was observed in the direct observational (ADOS) data. This suggests that individuals with PDD-NOS do not merely have a milder version of the symptom constellation seen in AD, but instead show a related but different pattern of difficulties. This is especially evident in figure one, and implies that PDD SC is a distinct variant of autism that does not merely sit at the less severe end of the same continuum of symptoms.

A related question concerns the validity of the PDD-NOS diagnosis. In psychiatric nosology, *'a diagnostic concept is assumed to have validity to the extent that the defining features of the disorder contain information not contained in the definition of the disorder'* (Spitzer, 2001, p.353). Due to the issues of reliability discussed in the introduction to this paper, little is known about the validity of PDD-NOS. In the current study, measures of autistic associated features of are relevance to discussions of PDD-NOS' validity, as they are independent of the behaviours used to decide diagnosis. We observed that, as a group, participants with PDD-NOS differed from those with AD and AsD in terms of a range of associated features of autism. They were less likely to be abnormally sensitive to sound and had fewer eating problems. They were also less impaired on measures of visio-spatial abilities. These are initial signs that PDD-NOS has validity as a diagnostic category, although further work will be required to test this fully. In particular it will be valuable to examine the extent to which PDD-NOS has meaningfully different aetiology, prognosis and treatment needs compared to the other PDDs.

Fractionation of the autistic triad

There is currently debate about the unity of the autistic triad (e.g. Happé & Ronald, 2008; Mandy & Skuse, 2008), in which a range of opinions can be described. At the 'strong revisionist' end of this spectrum, on the basis of behavioural genetic findings from community samples, researchers have argued that each element of the autistic triad is largely phenomenologically and aetiologically independent (Happé , Ronald, & Plomin, 2006). At the opposite, 'strong traditionalist' end of the spectrum are researchers who maintain that the autism syndrome, with its triad of impairments, is unitary (Constantino & Todd, 2003). In between these two extremes, albeit nearer the revisionist position, are those who propose partial fractionation of the triad, arguing for one domain of impairment that includes the social and communication elements of the autistic triad along with at least one other domain of impairment (Mandy & Skuse, 2008).

Our data do not support the strong revisionist position, since we found an association between social and communication impairment in our PDD-NOS sample: individuals who had impairments in reciprocal social interaction almost always had accompanying impairments in communication, even though such a combination was not necessary to receive this diagnosis. Our findings also argue against the strong traditionalist position, since we describe a large group of individuals who have only part of the autistic triad. This offers support for a multiple underlying impairments (Goodman, 1989) model of PDDs, in which different underlying abnormalities relate to different behavioural symptom clusters. Accordingly we hypothesise that the social communication impairments seen in autism will share underlying causes, and that these will be at least partially independent of the causes of RSBs. In addition we hypothesise that, compared to AD, individuals with PDD SC will have less extensive abnormalities at the genetic, neuroanatomical and neuropsychological level, reflecting their more circumscribed pattern of symptoms.

Limitations

We present data on a large PDD-NOS group, defined according to clearly operationalised, replicable diagnostic criteria. This group was characterised in diverse terms independent of the measures used to assign diagnosis. However the following limitations should be considered when generalising the reported findings. Firstly, the great majority of the sample had IQ outside of the intellectual disability range. Whilst most people on the autism spectrum have IQs in the normal range (Chakrabarti &

Fombonne, 2005), it is important to acknowledge that there may be other PDD-NOS presentations amongst people with intellectual disability. Secondly, we describe a clinic-referred sample that allows us to observe the phenomena but not the prevalence of PDD-SC relative to other PDDs. It will be important to see how common PDD-SC is in epidemiological studies using whole population screening in which individuals receive comprehensive assessment if they present with just part of the autism syndrome. This will avoid a systematic bias against individuals with PDD-SC, and would allow for the characterisation of any other 'partial triad' presentations that may exist (Happé & Ronald, 2008).

Implications for DSM-V

The draft diagnostic criteria for DSM-V (APA, 2010) propose several significant changes to how autism spectrum disorders are conceptualised. They suggest that the DSM-IV-TR categories of AD, AsD and PDD-NOS be subsumed by a single 'Autism Spectrum Disorder' (ASD) diagnosis and propose that the autistic triad be replaced by an autistic dyad, comprising social-communication deficits and RSBs. According to the draft DSM-V criteria, an ASD diagnosis would only be made in the presence of clinically significant deficits in both domains of this dyad. This necessitates the presence of at least two of the following three types of RSB: (1) stereotyped motor behaviours, or unusual sensory behaviours; (2) excessive adherence to routines and ritualised patterns of behaviour; (3) restricted, fixated interests.

Whilst the nosological value of AsD is not the focus of this paper, our data do allow us to make some comment on the validity of AsD as a disorder distinct from high-functioning autism. The AsD participants we described were, as a group, somewhat more intelligent than those with AD. Despite this, on measures of core and associated autistic features there were no significant differences between AsD and AD. This finding is in accordance with much of the relevant literature (e.g. Frith, 2004; Sanders, 2009), and suggests that AsD, at least as defined in the current study, is a variant of autism occurring in higher-functioning individuals, and not a distinct disorder. As such our data support the suggestion that AsD and AD be amalgamated in DSM-V. In addition, our findings are supportive of the DSM-V plan to merge social and communication domains of autistic impairment. In our PDD-NOS group we observed only 2 out of 66 individuals who had social impairments but not communication impairments,

and even these had significant difficulties with communication that did not quite meet 3Di threshold for impairment.

However our data are more equivocal with respect to the proposal that both social-communication impairment and RSB be present for any ASD diagnosis. Despite their severe and impairing difficulties with reciprocal social interaction and communication, most of the children classified as PDD SC in the current study would not meet proposed DSM-V ASD criteria, due to a lack of unusual sensory interests and the absence of stereotyped, routinised patterns of behaviour. Given that a quarter of our clinical PDD sample met PDD SC criteria, and the even higher prevalence of PDD-NOS relative to AD and AsD in epidemiological samples (e.g. Baird et al., 2006), this suggests that current proposals for DSM-V ASD would cause a significant narrowing of the autism spectrum.

Such reform would bring both advantages and costs. It would reduce the chances of ASD being an over-inclusive diagnosis, making it less likely that mildly elevated social-communication difficulties will be unhelpfully labelled as pathological. It may also yield more homogenous groups for the purposes of research. However a tightening of ASD diagnostic criteria will exclude some individuals with very severe social-communication difficulties from receiving the benefits associated with diagnosis, such as funding for clinical and educational support. In addition, without a diagnostic label, individuals with PDD SC are less likely to be the focus of research, despite the potential insights their particular pattern of difficulties offers to those wishing to better understand the phenomena and aetiology of autism.

Acknowledgement: The authors would like to thank the children and families who participated in data collection. They also thank their anonymous reviewers, whose comments improved the paper.

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TABLES

Table 1 – Sample Characteristics by Diagnosis

	PDD-NOS	**Autistic Disorder**	**Asperger's Disorder**	**P**	**Group differences**
	N=66	**N=97**	**N=93**		
Age in years (mean; SD)	9.05(3.51)	9.43 (3.50)	8.82 (3.81)	P=.50	-
Proportion male	79%	85%	83%	P=.26	-
Verbal IQ (mean;SD)[1]	93.34 (16.14)	87.60 (15.98)	102.55 (19.70)	P<.001	**AsD>PDD-NOS,AD**
Proportion VIQ above 70	91%	86%	97%	P =.08	-
Performance IQ[2] **(Mean;SD)**	92.78 (15.78)	91.45 (18.07)	101.62 (17.85)	P=.006	**AsD>PDD-NOS,AD**
Proportion PIQ above 70	94%	90%	96%	P=.41	-

[1]Verbal IQ (VIQ) available for n=171, 67% of sample

[2] Performance IQ (PIQ) available for n=148, 58% of sample

Table 2 – Core autistic symptomatology by PDD diagnosis

(higher scores and positive effects denote greater impairment; effect sizes expressed as Cohen's D relative to PDD-NOS)

	PDD-NOS	**Autistic Disorder**		**Asperger's Disorder**		**P**	**Group differences**
	N=66	**N=97**		**N=93**			
	Mean (SD)	**Mean (SD)**	**Effect size (95% confidence interval)**	**Mean (SD)**	**Effect size (95% confidence interval)**		
3Di ADI-R equivalent algorithm							
Reciprocal Social Interaction	14.30 (2.73)	16.54 (3.08)	+.77*** (.44 to 1.08)	16.31 (3.62)	+.63*** (.29 to .94)	P<.001	PDD-NOS<AD, AsD
Communication	12.37 (2.77)	15.47 (3.32)	+.1.01*** (.67 to 1.33)	14.42 (3.59)	+.64*** (.30 to .95)	P<.001	PDD-NOS<AD, AsD
Repetitive and Stereotyped Behaviour	1.77 (1.18)	5.67 (1.82)	+2.54*** (2.03 to 2.85)	5.98 (1.96)	+2.60*** (2.08 to 2.92)	P<.001	PDD-NOS<AD, AsD
ADOS[1]							

Reciprocal Social Interaction	35.10 (22.40)	41.08 (21.60)	+0.27 (-.04 to .59)	34.72 (21.00)	+0.02 (-.34 to .30)	P=.09	-
Social Communication	23.59 (17.90)	29.06 (18.00)	+0.19 (-.01 to .62)	24.17 (19.08)	+0.03 (-.29 to .35)	P=.06	-
Repetitive and Stereotyped Behaviour	15.25 (19.55)	25.71 (24.16)	+0.48** (.15 to .78)	20.03 (21.60)	+0.23 (-.09 to .55)	P=0.01	PDD-NOS<AD

*p<.05, ** p<.01,***p<.001

[1] ADOS scores expressed as percentage of scale maximum to allow comparison across modules.

Table 3 – Parent reported (3Di) repetitive and stereotyped behaviours by diagnosis.

The relative risk expresses the probability within a diagnostic category, relative to PDD-NOS, of a behavior being present at a clinically significant level.

	PDD-NOS	**AD**		**AsD**	
	N = 66	**N = 97**		**N = 93**	
	% in clinical range	% in clinical range	Relative risk (95% confidence interval)	% in clinical range	Relative risk (95% confidence interval)
Studies objects from unusual angles	6	39***	6.07 (1.95 to 18.92)	34***	5.37 (1.72 to 16.78)
Strong interest in numbers	6	21*	3.23 (1.03 to 14.08)	37***	5.79 (1.86 to 18.02)
Endlessly and exactly repeats words	7	51***	7.58 (2.88 to 19.93)	59***	8.90 (3.40 to 23.29)
Interest in spinning things	9	34**	3.97 (1.47 to 10.74)	36**	4.18 (1.56 to 11.23)

Precise yet odd routines	9	64***	7.03 (3.23 to 15.30)	70***	7.69 (3.54 to 16.68)
Hand or finger mannerisms	11	50***	4.67 (2.25 to 9.66)	51***	4.77 (2.30 to 9.88)
Complex mannerisms	11	41***	3.89 (1.86 to 8.15)	30**	2.83 (1.32 to 6.11)
Infant-like sensory interests	14	43***	3.18 (1.66 to 6.07)	54***	3.94 (2.09 to 7.45)
Uses sophisticated, unusual words	16	23	1.45 (.73 to 2.86)	38**	2.42 (1.29 to 4.53)
Odd preoccupation	20	55***	2.77 (1.65 to 4.66)	55***	2.78 (1.65 to 4.69)
Oddly formal play	21	64***	3.01 (1.85 to 4.91)	54***	2.53 (1.53 to 4.19)
Large store of factual information	27	36	1.34 (.82 to 2.18)	51**	1.92 (1.22 to 3.03)

Replays sections of DVD or video	30	60**	2.02 (1.25 to 3.27)	59**	1.98 (1.22 to 3.19)
One over-riding interest	44	73***	1.66 (1.23 to 2.24)	74***	1.69 (1.25 to 2.27)
Exaggerated interest in one area	45	85***	1.91 (1.37 to 2.66)	75***	1.71 (1.22 to 2.42)

*p<.05, **p<.01, ***p<.001

Table 4 – Associated features of autism by diagnosis

(higher scores and positive effects denote greater impairment; effect sizes expressed as Cohen's D relative to PDD-NOS)

	PDD-NOS	**Autistic Disorder**		**Asperger's Disorder**		**P**	**Group differences**
	N=66	**N=97**		**N=93**			
	Mean (SD)	**Mean (SD)**	**Effect size (95% confidence interval)**	**Mean (SD)**	**Effect size (95% confidence interval)**		
Fine Motor Impairment	7.70 (4.05)	8.99 (4.46)	+0.27 (-.07 to .61)	9.29 (5.26)	+0.34 (-.02 to .69)	P=.09	-
Visio-spatial Impairment	3.36 (3.32)	4.93 (3.66)	+0.45** (.11 to .78)	4.81 (4.12)	+0.39* (.03 to .74)	P=.03	PDD-NOS<AD,AsD
Gross Motor Impairment	2.32 (2.15)	2.48 (2.01)	+0.08 (-.26 to .41)	2.71 (2.09)	+0.18 (-.17 to .54)	P=.46	-
Auditory Sensitivity	1.77 (1.61)	2.41 (1.54)	+0.41* (.07 to .76)	2.72 (1.53)	+0.60** (.25 to .97)	P=.002	PDD-NOS<AD,AsD
Feeding Difficulties	1.54	2.18	+.36*	2.37	+.44**	P=.008	PDD-NOS<AD,AsD

	(1.83)	(1.77)	(.01 to .70)	(1.91)	(.09 to .80)		
Sleep Problems	6.38 (3.55)	7.09 (3.89)	+.19 (-.15 to .53)	7.70 (4.58)	+.32 (-.04 to .67)	P=.30	

*p<.05, ** p<.01,***p<.001