Editorial Perspective: Autism Spectrum Disorders in DSM-5 – an historical perspective and the need for change

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The newest revision of the Diagnostic and Statistical Manual of Mental Disorders (DSM), the DSM-5 (http://www.dsm5.org), will be published in just a few months and anticipation is high. The changes to the Pervasive Developmental Disorders (PDD) criteria are likely to be among the most extensive of the revisions to the manual, as well as among the most contested. Worries about loss of diagnosis and service eligibility are common, especially among parents. Some have felt that the proposed changes are confusing, even capricious, or poorly justified (e.g., Ghaziuddin, 2011). The rationale for combining the individual PDD diagnoses into a single category of Autism Spectrum Disorder (ASD), when we hear so much about the heterogeneity of autism, has been questioned. Rather than referring to “autism” as a unitary condition, many researchers now refer to “autisms” in the plural (Boucher, 2011) so the DSM-5 proposal to lump rather than split can seem perplexing.

But in fact the proposed changes have a clear empirical basis, resulting from extensive literature reviews and secondary data analyses undertaken by the DSM workgroups. In the course of preparing a Virtual Issue of papers previously published in JCPP (which can be found at http://bit.ly/MxjWjj), a comprehensive search of the Wiley-Blackwell database turned up 535 papers on PDD/ASD from the period between the first edition of JCPP in 1961 to today. Of these, 158 papers were specifically on diagnostic or classification issues. In the Virtual Issue, I selected a dozen papers from JCPP that traced the changing conceptualizations of the PDDs over the last two decades. As I hope the present editorial and the Virtual Issue make clear, the changes are anything but capricious or arbitrary.

To ground this within the appropriate context, I begin with a list (not exhaustive) of the proposed changes to the criteria:

- Change in name from Pervasive Developmental Disorder to Autism Spectrum Disorder
- Creation of a single diagnosis, Autism Spectrum Disorder, rather than a category containing five individual diagnoses
  - Autistic Disorder, Asperger Disorder, Childhood Disintegrative Disorder, and PDD Not Otherwise Specified (PDDNOS) are subsumed under one label: Autism Spectrum Disorder
  - Rett Disorder is eliminated now that its molecular basis is known (the DSM focuses on disorders without a molecular or biological test, that must instead be defined behaviorally)
- Three symptom domains (social, communication, and repetitive behavior) become two (social-communication and repetitive behaviors)
- Number of symptoms streamlined from 12 to 7 by merging criteria that were overlapping or described similar behaviors (e.g., limited social-emotional reciprocity, limited sharing of interests, and reduced back-and-forth conversation are combined into one reciprocity symptom) and eliminating symptoms that are not specific to ASD (e.g., delayed development of language)
- Provision of “severity” criteria to better capture the spectrum nature of the disorder and the inter-individual variations that differ less in quality than in quantity (e.g., intensity and duration of symptoms, degree of impairment, and distress they cause)
- Development of a new Social Communication Disorder category (outside the autism spectrum) to provide diagnostic coverage to children who present with only social-communication problems and do not display the repetitive and stereotyped behaviors of ASD

My thoughts on the 12 individual papers featured in the Virtual Issue can be found in the VI Editorial (http://bit.ly/MxjWjj). Each paper represented a particular perspective in our evolving understanding of the classification of ASDs and each contributed significantly to the conceptual development of the proposed DSM-5 criteria. What particularly struck me in re-reading that group of papers, as well as other recent literature, is that many of the ideas being applied in DSM-5 have actually been around for quite a while. One of the most significant alterations is the change in name of the category. While this may appear to be a radical modification, in fact, calls for revision have been around for decades. The Pervasive Developmental Disorder label has been described as both misleading, in obscuring the specificity of the deficits, and unhelpful, in obscuring the relationship with autism. Over two decades ago, Happé and Frith1 (1991) suggested that the

1Italicised names and dates indicate an article cited in the online Virtual Issue: these are available for immediate access at http://onlinelibrary.wiley.com/journal/10.1111/(ISSN)1469-7610/homepage/autism.htm
terminology “autism spectrum disorders” be used instead. While these words of wisdom were ahead of their time and were not applied to DSM-IV, the term rapidly came into wide usage by professionals and the public alike (Williams et al., 2008). The term will now be explicitly adopted by DSM-5, over 20 years later, and will finally have a set of defined criteria associated with it, so that, going forward, there can be the consistency in usage that is necessary for the best clinical care.

Another area in need of modification, based on similar issues of consistency of usage (or lack thereof), are the subtypes of PDD. When DSM-IV was published in 1994, it included, for the first time, the diagnosis of Asperger Disorder. Articles on this topic quickly began to appear in the literature and dozens of studies were published examining the relationship between Asperger Disorder and autism. While a few found different phenotypic profiles (e.g., Klin et al., 1995), the majority of these studies failed to find empirical differences between the two PDD subtypes (summarized in Frith, 2004; Macintosh & Disananyake, 2004). To the extent that differences between subtypes have been found, they were quantitative (e.g., differing in degree of impairment, severity of symptoms, or level of cognitive function) rather than qualitative (Prior et al., 1998).

All revisions of the DSM have been undertaken to improve accuracy of classification and the current revision is no exception. There is ample evidence of inaccuracy in the use of the PDD subtypes. Multiple studies demonstrated that most children with an Asperger-like clinical presentation actually met DSM-IV criteria for Autistic Disorder (Gilchrist et al., 2001; Williams et al., 2008). A striking lack of consistency in how the labels were used has been reported in both North America (Lord et al., 2011) and Australia (Williams et al., 2008). A large multisite study demonstrated the highly varied approaches to classification that are taken by even experienced evaluators (Lord et al., 2011). In this important study, conducted across multiple sites with substantial diagnostic expertise, participant characteristics (IQ, autism severity) did not differ across sites, yet rates of specific PDD diagnoses were highly variable. There was significant agreement across sites on specific scores on standardized instruments, but significant variation across sites in the weighting and interpretation of this information to arrive at a clinical diagnosis. Sites agreed on the dimensional aspects of the phenomena; what they did not agree on was the categories. The strongest predictor of diagnosis was what site made it, rather than any characteristic of the child. This is a clear sign that the PDD subtypes were just not working.

So, although the subsuming of Asperger Disorder within an Autism Spectrum Disorder diagnosis has been controversial, these papers collectively illustrate the empirical basis upon which the decision was made and should dispel some of the mystery surrounding the rationale. While it might seem paradoxical at first glance, the “lumping” approach taken by the new criteria may also address inequities that exist in many parts of the world, where individuals with Asperger Disorder (and often PDDNOS as well) are ineligible for government-based services. From my perspective as a practicing clinician, if these conditions cannot be validly distinguished empirically and the labels are used inconsistently, then it is logically questionable (as well as patently unfair) to deny services to some while providing a full range of interventions to others. Creating a single diagnostic entity will also, as summarized by Lord and Jones (2012), “avoid the current phenomenon of the same individual receiving serial or sometimes even concurrent diagnoses of PDDNOS, autism, and Asperger Syndrome, depending on the knowledge and biases of their diagnostician.”

The DSM had hoped to “carve nature at its joints” by defining a finite set of non-overlapping categories. It is now clear that the boundaries between DSM categories are not nearly as tidy as envisioned and hoped. This, in fact, has been evident for some time, as described by Caron and Rutter (1991) and Cantwell (1996). Summarizing literature from the decades preceding DSM-IV, they demonstrated clearly that diagnoses co-occur at very high rates in psychiatric patients. Comorbidity may arise in a number of ways that are artifactual, such as artificial subdivision of syndromes, overlapping criteria between disorders, and one disorder being an early manifestation of another disorder (Caron & Rutter, 1991). More recent papers, post-DSM-IV, have fully supported these findings. So the revisions undertaken in the fourth edition did not solve the problems and thus we are facing them again, this time head on, through the DSM-5’s dimensional approach to defining psychopathology. While this will help address issues of overlap and subdivision of syndromes, it is worth reminding ourselves that there will always be the need for categories. A “categorical v. dimensional” argument presents a false dichotomy since “dimensions can always be made into categories by defining thresholds” (Lord & Jones 2012, p. 492). This is exactly what the DSM-5 will ultimately do. Psychopathology will be defined by dimensional ratings of symptom counts, level of impairment, severity, and need for support, but cutoffs and criteria will be applied to boil this down to categories that are needed for clinical care. As stated succinctly by Pine (2011, p. 1221), “even if risk ultimately is shown only to manifest continuously, decisions on treatment are made categorically. ...Clinicians regularly must answer a series of categorical questions, even if they are posed in a dimensional landscape. Is the level of suffering or impairment in this particular child sufficiently extreme to warrant treatment?” The DSM-5 balances the dimensional and categorical frameworks better than any previous version.
One of the chief concerns about the proposed revisions is whether some children who met DSM-IV criteria will no longer meet DSM-5 criteria. This is a significant and legitimate concern for all of us, parents, clinicians, and researchers alike. Indeed, the instructions given all the DSM working groups was, first and foremost, “do no harm”, asking explicitly about each change, “how will this affect patients?” (Kuhl et al., 2011; Swedo et al., 2012). In the case of ASD, this concern has played out particularly in the new requirement that both social-communication and repetitive behavior symptoms be present in order for a diagnosis to be made. Previously, a diagnosis of PDDNOS did not require repetitive behavior symptoms. Mandy et al. (2011) demonstrated that this is a rather large group of children (indeed, in their sample, 97% of the PDDNOS group) who might suddenly be excluded from services.

A new diagnosis, called Social Communication Disorder, is also being proposed by the DSM-5 Neurodevelopmental Disorders Working Group. It is intended to provide a label for children with significant social and communication difficulties who do not exhibit the repetitive behaviors of ASD. At first glance, this addition may appear to be an improvement on the DSM-IV, as it may help identify a group of children not previously recognized in the DSM, those with significant pragmatic language disorders, who often show co-occurring social delays (Bishop & Norbury, 2002; Reisinger et al., 2011). This may also provide a new diagnostic “home” for the children with PDDNOS who do not meet DSM-5 ASD criteria because of lack of stereotyped behaviors (Mandy et al., 2011). Several critical questions have been raised, however, about the validity of the new Social Communication Disorder diagnosis (Skuse, 2012; Tanguay, 2011): essentially, what is the evidence that this condition is any different (e.g., etiology, symptom profile, management) from ASD? I would second these concerns, which are highly reminiscent of those posed about Asperger Disorder when it was first included in DSM-IV. It seems logically and internally inconsistent for the DSM-5, so pioneering in its dimensional approach, to introduce a separate category that is so qualitatively similar to another condition. Let’s not take two steps forward and one step back.

As we await the results of the field trials later this year, and the final version of the criteria available in May 2013, the literature reminds us of what molded this revision, what lessons were learned, and what issues remain yet to be resolved. It is clear that the changes, on the surface so radical, arise from empirical and conceptual forces at work for decades.

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[Articles with italicized author names and dates above are not referenced below but are all cited in a recent JCPP Virtual Issue and available for immediate access at http://bit.ly/MxjWjj]


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