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Update on diagnostic classification in autism

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Abstract

Purpose of review—In the lead up to and following the recent publication of the DSM-5, the diagnostic construct of autism has received intense scrutiny.

Recent findings—This article briefly reviews the history of the diagnosis of autism, the changes that have occurred in the diagnosis over time, and the rationale for change. The most significant changes being introduced with the DSM-5 are highlighted, as well as some of the concerns that will be a focus of attention with respect to the potential impacts going forward.

Summary—The categorical divisions that characterized the pervasive developmental disorders are now collapsed into a single entity, autism spectrum disorder. The final DSM-5 criteria have yet to be formally compared prospectively against prior criteria, but early indications suggest that the boundaries around the pervasive developmental disorders have not been substantially altered.

Keywords

Asperger's disorder; autism spectrum disorder; DSM-5; pervasive developmental disorder

INTRODUCTION

Dennis Cantwell, MD, was an icon in child and adolescent psychiatry who served on the original DSM-III task force. Among the widely quoted statements he often made regarding the development and application of psychiatric diagnoses was his observation that 'kids don't read the DSM'. The statement is a wonderful distillation of the experience of clinician scientists who have endeavored, as Mercier suggested over a century ago, to 'draw up an elaborate scheme of classes, orders, and genera' into which mental disorders might be classified (p. 284). 'Cases will always occur partaking pretty equally of the nature of two adjoining groups, and other cases will occur which exhibit at one time the features of one group, and at another time those of another' [1].

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Conflicts of interest

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Against the seeming backdrop of difficulty, defining disorders in psychiatry in general and improving the approach for autism in particular has long been recognized as an important path, and should include not only understanding underlying disease processes, but diagnostic prognosis and effective treatment [2,3■]. The field continues to make progress in this regard, and the past year has been of particular significance with respect to diagnostic classification in autism.

REVIEW OF AUTISM CRITERIA OVER TIME

Although there are historical clinical descriptions of individuals who would now be diagnosed with autism, Kanner [4] was the first to formally introduce the syndrome in his seminal paper entitled ‘Autistic Disturbances of Affective Contact’. The core elements of the phenotype that Kanner highlighted included a profound lack of affective contact with others, an anxiously obsessive desire for the preservations of sameness, a monotonous repetition of verbal and motor behavior, a fascination for objects, and mutism or language that did not seem intended to serve the purpose of interpersonal communication. Kanner also highlighted the lack of obvious congenital abnormalities common in children with comparable intellectual disabilities and the wide scatter of cognitive and motor abilities of the children whom he had seen.

At about the same time, Asperger [5] described a similar constellation of symptoms in a sample of children from his clinic. Asperger’s cases had social communication difficulties that were particularly impairing in light of their general intellectual ability, had circumscribed interests, and ‘conspicuously lacked common sense’. Unusual sensory responses were also common. Interestingly, Asperger’s impression was that the syndrome was not particularly rare, reporting that he had seen upwards of 200 such children in his clinic.

A few years later, Cappon [6] observed that the field was wrestling with whether autism truly existed as a discreet entity, or whether it was more properly viewed as a manifestation of psychosis or schizophrenia in childhood. Cappon summarized the diagnostic criteria for autism at the time, including impairments in social relation, preference for objects, pronomial reversal, hyperactivity, low frustration tolerance, concrete thinking, echolalia, marked repetition of words and activity, pain insensitivity, and wide scatter in intellectual abilities.

Many of these features were captured with the introduction of autistic disorder in the official diagnostic nomenclature in Diagnostic and Statistical Manual (DSM)-III [7]. Core features specified onset prior to 30 months of age, pervasive lack of responsiveness to others, gross deficits in language development, peculiar speech patterns (including pronomial reversal and echolalia), and bizarre responses to the environment, including resistance to change and fascination with objects. In order to create a clear boundary between autism and schizophrenia, an explicit trumping rule was inserted in DSM-III such that autism could not occur in the presence of delusions, hallucinations, loosening of associations, or incoherence. Childhood-onset pervasive developmental disorder (PDD) and atypical PDD were also introduced in DSM-III. In 1987, DSM-III-R [8] expanded the menu of symptoms supporting

the diagnosis of autistic disorder, and formally separated core features into three domains of impairment in reciprocal social interaction, communication, and restricted or repetitive behaviors.

The next significant change occurred in 1994 [9,10] with the introduction of the categorical diagnoses of Asperger's disorder, childhood disintegrative disorder, Rett's disorder, and PDD not otherwise specified (PDD-NOS). At the time, there was considerable debate about whether evidence supported these changes; particularly whether so-called high-functioning autism (e.g. autism without intellectual disability) and Asperger's disorder actually represented different entities. Miller and Ozonoff [11] observed, for example, that Asperger's original cases would not meet the criteria for Asperger's disorder. Providing an impetus, in part, for these changes were the significant advances that were being made during the 1980s and 1990s in identifying the core, early emerging characteristics of autism. Pioneering work identifying impairments in affective reciprocity, joint attention, social orienting, imitation and theory of mind underscored the key challenges in social communication.

DSM-5

In the years since the introduction of the DSM-IV criteria, advances in genetics coupled with a significant increase in the prevalence of PDDs laid the foundation for the changes that were incorporated in DSM-5 [12■■]. These changes included a change in name and dissolution of categories such that autistic disorder, Asperger's, childhood disintegrative disorder, and PDD-NOS were all collapsed into autism spectrum disorder. The latter term recognizes that, over the past decade, 90% of publications in the field use 'autism spectrum disorder' instead of PDD.

The elimination of Asperger's disorder in the DSM-5 has been understandably controversial, with reports in the literature providing both evidence in support and evidence against the removal of the categorical subtype. For example, Tsai and Ghazziuddin [13] reviewed 125 studies relating to more than 90 clinical variables, comparing Asperger's disorder with autistic disorder and PDD-NOS. These authors argue that, because some studies suggest that there are quantitative and qualitative differences between, for example, low-functioning autistic disorder and Asperger's disorder (as well as other between-group comparisons), these conditions do not lie on a continuum and should be treated as separate disorders. These and other authors have also raised the concern that future research will be hampered by the dissolution of the categorical diagnoses within the pervasive developmental disorders [14] and may make epidemiological and other comparisons invalid [15]. Tsai [16] predicts a comeback of Asperger's disorder within a decade or two.

On the contrary, Sharma *et al.* [17■■] reviewed 69 studies published over two decades on Asperger's syndrome and concluded that, because of the overlapping signs and symptoms relative to autistic disorder, the DSM-IV criteria were insufficient and invalid. A multisite study including over 2000 individuals all carefully diagnosed with gold standard diagnostic instruments by Lord *et al.* [18■■,19] suggested that the diagnosis of Asperger's disorder was more significantly influenced by where one was evaluated than by the symptoms with which

one presented. Additionally, review of the literature does not suggest that there are either differential responses to treatment or differences in cause when Asperger's disorder and autistic disorder are compared [20].

The significant advances that have occurred in the past decade with respect to the identification of genes and copy number variants associated with autism underscore the failure to identify differences in cause between the behaviourally defined subtypes. These studies have universally demonstrated that the risks that these genetic abnormalities confer are to autism spectrum disorders and not to the specific categorical subtypes of the DSM-IV. For example, in a study of autism spectrum disorders in 63 males with fragile X syndrome [21], roughly 30% met criteria for autistic disorder and another 30% for the diagnosis of PDD-NOS. Further, in this and other populations of individuals followed over time, the same individual may transition from a diagnosis of autistic disorder or Asperger's disorder to a diagnosis of PDD-NOS. While there may be some value in using different categorical terms as a proxy for severity, these fluid boundaries between categories are more consistent with a spectrum construct than discrete, independent diagnoses with different causes.

Other changes introduced with the DSM-5 included the consolidation of three core symptom domains into two, with language pulled out as an independent specifier. Thus, social communication and social interaction became one core domain, and restricted or repetitive behaviors or interests became the other. Additionally, sensory hypo or hypersensitivity was added to the latter group of symptoms. Mandy *et al.* [22] used confirmatory factor analysis, on a large heterogeneous sample, to test the construct validity of this proposed DSM-5 symptom model. These analyses supported the merging of social and communication domains into one domain, with a second domain of repetitive behavior, speech, use of language, and sensory difficulties. They also found support for the conceptualization of sensory abnormalities as an aspect of restricted, repetitive behaviors or interests. Similarly, Guthrie *et al.* [23] examined the factor structure of autism symptoms in toddlers aged 12–30 months, comparing DSM-5, DSM-IV, and a one-factor and an alternative three-factor model proposed by van Lang *et al.* [24]. Guthrie *et al.* [23] found that the DSM-5 two-factor model is superior to all other models in classifying autism symptoms, as measured by the Autism Diagnostic Observation Schedule – Toddler Module (ADOS-T) with 18–30-month-olds. Consistency of these results with other studies addressing the question of factor structure of autism symptoms in older children and adults suggests that this is similar throughout development.

EARLY TESTING OF THE DSM-5

In the run up to the formal changes to the diagnosis of autism spectrum disorder (ASD) in DSM-5, a number of studies approached the important question of how proposed changes might alter the boundaries around the PDD spectrum – who would be diagnostically included or excluded going forward. Using a variety of different strategies, a number of groups came to varied conclusions. For example, Mattila *et al.* [25] used a screened epidemiological sample of 82 individuals, out of 5848 eight-year-olds, and compared DSM-IV criteria with DSM-5. They found DSM-5 to be less sensitive than DSM-IV; however, the study referred to an early draft version of DSM-5 that was later updated, and the study did

not examine PDD-NOS. McPartland *et al.* [26■] applied the proposed DSM-5 ASD criteria to a large sample of participants who were evaluated on DSM-III and DSM-IV criteria. Using a checklist of 61 items from the original evaluation, re-analysis based on their DSM-5 algorithm suggested that it was less sensitive to autism in cognitively able participants; specificity was reported at 60.6% and sensitivity at 94.9%. Similarly, Taheri and Perry [27] conducted a retrospective chart review on a sample of 131 children. Only 63% of their DSM-IV children with PDD met draft DSM-5 ASD criteria and only 17% of the DSM-IV PDD-NOS sample retained a diagnosis. The group meeting DSM-5 criteria generally had a lower intelligence quotient (IQ), lower level of adaptive skills, and greater autism severity. Matson *et al.* [28] also conducted a set of studies on toddlers, children, adolescents, and adults in order to compare DSM-IV and DSM-5 draft criteria for the diagnosis of ASD; they found a lower proportion of diagnosis in all age groups with DSM-5. In contrast, Huerta *et al.* [29] reported relatively high specificity (95%) and sensitivity (91%) with the suggested DSM-5 criteria in their retrospective data analysis on a large database including 4453 children and adolescents with DSM-IV PDD using just parent data [Autism Diagnostic Interview-Revised (ADI-R)]. Sensitivity was adequate (estimated at 53%) also in subgroups such as PDD-NOS, autistic disorder, girls and children under the age of 4, and was an improvement over DSM-IV. Young and Rodi [30] examined the validity of proposed DSM-5 criteria using data from newly referred individuals, as opposed to relying on archival data, thus avoiding many of the limitations of previous studies. DSM-5 demonstrated high specificity (1.0), but poor sensitivity (0.57) relative to DSM-IV as a gold standard. Those diagnosed with PDD-NOS and Asperger's according to DSM-IV were most likely to fall short of DSM-5 criteria thresholds, and most of these individuals excluded from DSM-5 diagnosis failed to meet all three social communication criteria. A 'by history' criterion that was added to the final DSM-5 was not evaluated in this study.

IMPORTANT NEW CHANGES IN THE DSM-5

Three additional and very significant changes to the ASD criteria in DSM-5 address concerns about diagnostic conversion and diagnostic independence. First is the inclusion of 'by history' in the assessment of diagnostic criteria. As noted above, unfortunately this criterion was not part of early draft criteria that were circulated for review, and many studies comparing the impact of the transition from DSM-IV to DSM-5 would likely have yielded different results had the full criteria set been utilized. Second, the insertion of a 'grandfather' clause in the DSM-5, such that individuals with established diagnoses of PDD should simply be given the diagnosis of ASD, was not part of draft criteria. A final and similarly important change to the criteria for autism relates to the elimination of 'trumping rules' that had previously prevented the co-diagnosis of attention deficit hyperactivity disorder or of schizophrenia in the setting of autism.

Lastly, a new disorder, social communication disorder, was added outside the autism spectrum to provide a diagnostic cover for individuals with significant difficulties with social communication, but who have no history of repetitive or restricted behaviors. Some have suggested that individuals who previously may have been given the diagnosis of PDD-NOS may be shifted into this diagnosis (and thus inappropriately off the spectrum), and the argument has also been made that social communication deficits are the singular defining

feature of the autism (or social communication development) spectrum [31]. On the contrary, one could equally assert that individuals with social communication or social interaction deficits (e.g. the individual who impulsively interrupts, speaks too loudly, violates personal space, etc.) in the absence of any restricted or repetitive behavior history should not have been given a diagnosis of PDD-NOS in the first place – for example, that PDD-NOS was only a default because of the absence of a better way to capture a deficit in social communication pragmatics.

CONCLUSION

The changes to the diagnostic criteria for autism, now autism spectrum disorder, have obviously been received with mixed reviews. Some leaders in the field have raised important cautions and reservations (e.g. we will be failing to identify children who require services). Others have welcomed the changes (e.g. without evidence for treatment approaches specific to subtypes or causative differences, the overarching term more accurately reflects the state of science). As with any change, but particularly given the fact that prior criteria have been constant for nearly two decades, it will be important to assess the consequences going forward [32]. Studies are no doubt underway to assess the real impact on prevalence and on the service systems. At the same time, genetic and other studies have begun to challenge the very existence of the boundaries between psychiatric disorders such as autism or schizophrenia, as similar genetic mechanisms have been found to be related to these different, phenotypically quite distinct, diagnostic entities [33]. As Tsai and Ghaziuddin [13] recently characterized the DSM-5 approach as one that was ‘forward into the past’, the same may well be said for psychiatric diagnoses more broadly. Even as the ink was drying on the DSM-5 there were calls to revisit the entire system in favor of an approach that is organized around dimensions of neurobiology and observable behavior [34■■]. At the end of the day, the field still wrestles with the fact that ‘children do not read the DSM’.

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KEY POINTS

- DSM-5 introduces a controversial shift away from discrete and independent categorical entities within the pervasive developmental disorders spectrum to a single construct of autism spectrum disorder.
- The diagnostic boundaries around the newly constituted autism spectrum have not been clearly delineated relative to prior criteria as no prospective studies using the final DSM-5 criteria have been performed.
- The process of delineating psychiatric disorders by behavioral manifestations will continue to be challenged by the complexity and heterogeneity of children (and adults) who ‘do not read the DSM’.