DSM-5 and Proposed Changes to the Diagnosis of Autism

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When the American Psychiatric Association releases the *Diagnostic and Statistical Manual of Mental Disorders,* fifth edition (*DSM-5*) next month, children currently diagnosed with autism may lose access to services. Modifications to the proposed criteria have been suggested to address concerns of sensitivity.

IMPLICATIONS OF DSM-5

The American Psychiatric Association's stated goals for the changes to its diagnostic criteria for autism spectrum disorder (ASD) in *DSM-5* are to "accurately and completely identify" individuals with autism by production of reliable and valid diagnostic criteria[1] in order to offer a clearer, simpler, more reliable diagnostic scheme and recognize the "essential shared features of the autism spectrum."[2] This means that, for the clinician, it should be easier to diagnose ASD than trying to distinguish between high-functioning autism (HFA) and Asperger's disorder. In addition, the vague diagnosis of pervasive developmental disorder not otherwise specified (PDD-NOS) has been replaced, in part, by social communication disorder (SCD) (see Table 1, page 162).

Questions have already been raised about the appearance of a new diagnosis, SCD, that describes children with difficulties in the pragmatics of verbal and nonverbal communication, leading to impaired social function. This disorder excludes an autism diagnosis, and thus rules out the presence of restrictive, repetitive behaviors (RRBs);[3] however, SCD seems to resemble mild autism or PDD-NOS.[4,5] Questions remain about how the criteria for SCD will be implemented, and whether children who meet criteria for SCD will meet eligibility for medical and educational services.[6]

Overall, the changes should result in more reliable and valid diagnoses, with estimates of individual severity and associated conditions. It will remain important for clinicians to take a thoughtful and complete history and combine information from multiple sources with clinical observation to diagnose children with ASD with improved specificity.

For clinicians and families, the singular question is whether the new criteria will change access to treatment and/or educational services. Studies have noted that individuals with a diagnosis of Asperger's disorder or PDD-NOS, those with higher IQ, and female patients may be at most risk.[4] This could impact whether children with an established *DSM-IV* diagnosis of Asperger's disorder or PDD-NOS would need

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TABLE 1.

Comparison of Autism Criteria in *DSM-IV* and *DSM-5*

DSM-IV	*DSM-5*
A. (1) a. marked impairment in the use of multiple nonverbal aspects of social interaction	A2
A. (1) b. failure to develop appropriate peer relationships	A3
A. (1) c. lack of spontaneous sharing with other people	A1
A. (1) d. lack of social or emotional reciprocity	A1
A. (2) a. delay or lack of spoken language	?
A. (2) b. impaired ability to initiate or sustain a conversation	A1
A. (2) c. stereotypical and repetitive language	B1
A. (2) d. lack of make-believe or imitative play	A3
A. (3) a. stereotypical and restricted patterns of interest	B3
A. (3) b. inflexible adherence to rituals or routines	B2
A. (3) c. stereotypical and repetitive motor movements	B1
A. (3) d. persistent preoccupations with parts of objects	B3
None	B4
B. delays in at least one of three areas with onset prior to age 3 years of social interaction, language used as (1) social communication, (2) symbolic or (3) imaginative play, or (4) hyper-or hypo-reactivity to sensory input	A1 and A3 with onset in early childhood

re-evaluation per the *DSM-5* criteria. The question then becomes whether insurance companies and educational systems continue to accept these diagnoses for coverage.

Improved ability to identify a more homogenous group of ASD should help those researching the condition by improving the ability to detect etiologies, endophenotypes, genetic markers, and by strengthening treatment and outcome data. Yet, questions remain about how the proposed changes will affect the compatibility of future research with prior research and how the criteria will mesh with International Classification of Diseases, eleventh revision (ICD-11) standards.[7]

HISTORY OF AUTISM IN THE DSM

Although autism was first described by Kanner in 1935,[8] it was not considered a formal psychiatric diagnosis until the release of *DSM-III* in 1980.[9] In this system, to be diagnosed with autism, individuals had to meet all diagnostic criteria (monothetic) and the categories described a condition similar to classical autism.[4,9]

In the 1987 *DSM-III-R* version, a subset with a range of criteria (polythetic) could be applied during diagnosis, leading to a more heterogeneous diagnostic group[10] *DSM-IV*, published in 1994 (see Table 2, page 163), has continued the polythetic approach.[11] To meet the diagnostic criteria, patients must meet a minimum of six behavioral criteria subsets: two from social impairment; one from communication; and one from RRBs. Also, the onset of the condition must be prior to age 3 years.

Introduction of Asperger's Disorder

The *DSM-IV* requires that for an Asperger's disorder diagnosis, the patient meet a minimum of three criteria: two from social impairment and one from RRB. Asperger's disorder differs from autism in that there can be no communication impairment or delay in language, and no age of onset by 3 years. Also, there can be no delay in cognitive development or nonsocial adaptive behavior. *DSM-IV* criteria for PDD-NOS are for subthreshold presentations of autistic symptoms featuring impairment in social or communicative functioning or RRB (see Table 2, page 163).

Concerns about the DSM-IV and ASD

Questions about how the *DSM-IV* addresses ASD led to revisions in the *DSM-5*. First, the number of possible symptom combinations in the *DSM-IV* has been estimated to be 2,027, with inherent heterogeneity of the diagnostic group.[4] Concerns exist also with its reliability and validity. In a recent multisite study of 2,102 probands, the best-estimate clinical diagnoses were compared with those from standardized diagnostic instruments.[12]

Experts differed on how they interpreted *DSM-IV* criteria, even though the reliability of the data from the standardized interviews was good. Patterns of diagnosis were identifiable according to regional sites, with factors such as verbal IQ and language level influencing the process.

Also, research has not identified meaningful differences between *DSM-IV-text revision* (published in 2000) subtypes of ASD controlled for IQ and language.[13] In particular, Asperger's disorder is not thought to be distinct from HFA with little support of *DSM-IV* distinction.[2,14,15]

Diagnostic Challenges of Asperger's Disorder

Asperger's disorder is difficult to diagnose using *DSM-IV* criteria because accurately measuring language delays retrospectively is a challenge, as is clinically distinguishing these patients from those with autism.[2] In addition, Asperger's disorder diagnoses have been shown to be unreliable between expert clinicians.[12] It is also important to note children with autism who met language milestones before the age of 3 years may have the same adult outcome as those children with autism who did not.[16]

The *DSM-IV* criteria also have been faulted with how well they diagnose autism in children younger than 5 years, adolescents, females, and ethnic minority groups.[1] These concerns with the limitations of *DSM-IV* have been raised over the last 20 years by researchers in the area of diagnosis and classification of autism spectrum disorders and have prompted the development of the criteria found in *DSM-5*.

TABLE 2.

Diagnostic Criteria for Autistic Disorder from *DSM-IV*

A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):

(1) Qualitative impairment in social interaction, as manifested by at least two of the following:

(a) marked impairments in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body posture, and gestures to regulate social interaction;

(b) failure to develop peer relationships appropriate to developmental level;

(c) a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people, (eg, by a lack of showing, bringing, or pointing out objects of interest to other people); and

(d) lack of social or emotional reciprocity (note: in the description, it gives the following as examples: not actively participating in simple social play or games, preferring solitary activities, or involving others in activities only as tools or "mechanical" aids).

(2) Qualitative impairments in communication as manifested by at least one of the following:

(a) delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime);

(b) in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others;

(c) stereotyped and repetitive use of language or idiosyncratic language;

(d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level.

(3) Restricted repetitive and stereotyped patterns of behavior, interests and activities, as manifested by at least two of the following:

(a) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus;

(b) apparently inflexible adherence to specific, nonfunctional routines or rituals;

(c) stereotyped and repetitive motor mannerisms (eg, hand or finger flapping or twisting, or complex whole-body movements);

(d) persistent preoccupation with parts of objects.

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.

C. The disturbance is not better accounted for by Rett's disorder or childhood disintegrative disorder.

Source: American Psychiatric Association[11]

CHANGES IN THE DSM-5

In the *DSM-5*, autism, Asperger's disorder, and PDD-NOS will be combined into a single category of ASD[6] and supplemented with a dimensional aspect for assessing the level of dysfunction. This is important because social communication function appears to be distributed in a continuous fashion across the general population.[17,18]

In particular, the domains for social and communication problems have been combined into one set of deficits, labeled, "social communication and interactive problems." The set of symptoms for restricted, repetitive interests remains, but unusual sensory behaviors have been added to their diagnostic set.

The *DSM-5*'s approach to social communication symptoms is monothetic, requiring that individuals meet all criteria from the social-communicative set; for RRB, the *DSM-5* is polythetic, requiring that two of four symptoms be present. In all, five of seven symptoms must be present in the *DSM-5*, compared with six of 12 symptoms required by the *DSM-IV* (see Table 3, page 164).

The deficits in communication and social behaviors were combined into one domain because the Autism Work Group of the *DSM-5* Committee believed the two represent a similar impairment.[5] Because a delay in language is not believed to be unique or universal in ASD, this criterion is eliminated altogether.

The requirement for two symptom sets for repetitive behaviors and fixated interests by history or direct observation is thought to increase the stability of the diagnosis of ASD over time. The criteria now include symptoms for abnormal sensory behaviors. This improves the relevance of the criteria to younger children with ASD, because sensory issues are common concerns in this population.

The development of the *DSM-5* has been based on literature review, expert consultations, work group discussions, and secondary analysis of data sets. As noted, making the diagnosis should be

easier for the pediatrician since the criteria are designed to promote more agreement between clinicians.

EVIDENCE FOR AND AGAINST DSM-5 CHANGES

Several studies have been conducted to determine how *DSM-5* changes will affect the ASD population. Mattila et al[19] compared *DSM-IV* criteria with early draft criteria for the *DSM-5* in 82 individuals derived from an epidemiological sample of 5,484 8-year-olds. The *DSM-5* group was less sensitive for HFA (IQ > 70) and Asperger's disorder, but the group was an earlier version later updated by the Neurodevelopmental Disorders Workgroup. For patients with HFA, 73% were identified by the *DSM-5* but none of 11 subjects with Asperger's disorder were identified using the *DSM-5* criteria.

The study's authors suggested five modifications to relax the *DSM-5* criteria and create a "mild" version of autism compared with the "strict version" identified by the *DSM-5*. After the authors modified the *DSM-5* criteria, 96% of overall subjects were identified. No information is offered on effects of specificity.[19]

Supporting evidence was provided by Mandy et al[20] who reported that the *DSM-5* offered improved construct validity over *DSM-IV-TR* by improving the criteria language and by the inclusion of hyper- and hyposensory abnormalities as part of the symptoms cluster.

In a large analysis of siblings by Frazier et al[21] from the Interactive Autism Network, 8,911 siblings were found to have ASD; 5,863 did not. Compared with the *DSM-IV TR*, the proposed criteria show greater specificity to reduce false-positive diagnoses but slightly lower sensitivity so more false-negative diagnoses may result, especially with females.

Frazier et al[21] proposed relaxing the *DSM-5* criteria by requiring one less symptom criteria of SCI or RRB to increase sensitivity by 11% to 12%. This may be very pertinent for those diagnosed with Asperger's disorder or those youth whose early life symptoms are not easily obtained. Overall, the study's superior specificity validated the *DSM-5*.[21]

However, different conclusions were found by McPartland et al[4] in a sample of 933 subjects from a previous *DSM-IV* field trial of which 657 were diagnosed with ASD. This group was evaluated with proposed *DSM-5* criteria and sensitivity and specificity were measured. In this group, 60.6% of ASD cases meet revised *DSM-5* criteria for ASD with a specificity of 94.9%. Those with IQ > 70 and Asperger's disorder were less likely to be diagnosed according to the *DSM-5*. For example, the sensitivity of the *DSM-5* criteria to diagnose ASD in *DSM-IV* cases of Asperger's disorder was only 25%. They concluded the new criteria could have detrimental clinical and research effect. Others have questioned the validity of their findings given the historical data and methods.[1]

Matson and colleagues[22,23] have published several studies comparing the *DSM-IV* with the *DSM-5* and conclude the proposed changes will lead to 30% to 45% of children, adolescents, and adults classified with ASD per *DSM-IV-TR* to not meet criteria for ASD with *DSM-5*.

The most recent study to date estimated how many children diagnosed with PDD or non-PDD using the *DSM-IV-TR* will no longer meet the necessary criteria for ASD under *DSM-5*. Using data from 4,453 children with a clinical PDD diagnosis,

TABLE 3.

DSM-5 Proposed Criteria for Autism Spectrum Disorders

A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, and manifest by all three of the following:

1. Deficits in social-emotional reciprocity; ranging from abnormal social approach and failure of normal back and forth conversation through reduced sharing of interests, emotions, and affect and response to total lack of initiation of social interaction.

2. Deficits in nonverbal communicative behaviors used for social interaction; ranging from poorly integrated verbal and nonverbal communication, through abnormalities in eye contact and body language, or deficits in understanding and use of nonverbal communication, to total lack of facial expression or gestures.

3. Deficits in developing and maintaining relationships, appropriate to developmental level (beyond those with caregivers); ranging from difficulties adjusting behavior to suit different social contexts through difficulties in sharing imaginative play and in making friends to an apparent absence of interest in people.

B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least two of the following:

1. Stereotyped or repetitive speech, motor movements, or use of objects; (such as simple motor stereotypies, echolalia, repetitive use of objects, or idiosyncratic phrases).

2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change; (such as motoric rituals, insistence on same route or food, repetitive questioning or extreme distress at small changes).

3. Highly restricted, fixated interests that are abnormal in intensity or focus; (such as strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).

4. Hyper-or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment; (such as apparent indifference to pain/heat/cold, adverse response to specific sounds or textures, excessive smelling or touching of objects, fascination with lights or spinning objects).

C. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)

D. Symptoms together limit and impair everyday functioning.

Source: American Psychiatric Association[5]

TABLE 4.

Studies Comparing Diagnosis of ASD in *DSM-IV* and *DSM-5*

Study / year	Number of Subjects	Type of Sample	Instruments Used	Results	Limitations
Mattila et al[19] / 2011	82	Screened epidemiological sample diagnosed with *DSM-IV* criteria	ADI-R, ADOS, early *DSM-5* criteria	*DSM-5* was less sensitive than *DSM-IV* for ASD, (0.46)	Early *DSM-5* criteria, prevalence for PDD-NOS was not examined
Mandy et al[20] / 2012	708	Consecutive referrals to an autism specialty clinic	3Di	*DSM-5* model was superior to *DSM-IV*	Higher functioning sample, 3Di is a *DSM-IV* derived tool
Frazier et al[21] / 2012	14,744 siblings (8,911 with autism)	Family-selected internet registry	Mapped caregiver rated SRS and SCQ to *DSM-5* criteria	*DSM-5* had lower sensitivity, (0.81 vs. 0.95) but greater specificity, (0.97 vs. 0.86) than *DSM-IV*	Early *DSM-5* criteria, self-selected sample, reliance on caregiver reports only
McPartland et al[4] / 2012	933 (657 diagnosed with ASD)	Multicenter *DSM-IV* field trial database, with clear reliability data	Algorithm of items from *DSM-IV* mapped to match *DSM-5* criteria	60.6% of cases with ASD met *DSM-5* criteria with a specificity of 94.9%	Included only 48 *DSM-IV* subjects with Asperger's disorder, modified a historical data set to new criteria
Matson et al[22,23] / 2012	2,721 toddlers aged 17-36 months	EarlySteps participants	Clinical judgment using diagnostic algorithms	52.2% of toddlers were diagnosed with ASD by *DSM-5*	Single author review of evaluations based on *DSM-IV*
Gibbs et al[13] / 2012	132 youth	Referred to tertiary autism clinic for initial evaluation	ADOS, ADI-R	76.5% of participants were diagnosed with ASD by *DSM-5*	ADOS and ADI-R are *DSM-IV* based tools
Taheri and Perry[33] / 2012	131 children aged 2-12 years	Retrospective file review	CARS, *DSM-IV* checklist	62.6% of total sample met diagnosis of ASD by *DSM-5*	No Asperger's disorder patients, *DSM-5* criteria were evaluated by checklist
Huerta et al[24] / 2012	5,143 subjects, 4453 had PDD	Data sets from family genetics study, university and autism center databases	ADI-R and ADOS matched to *DSM-IV* and *DSM-5* criteria. Included parent report and/or direct observation	*DSM-5* identified 91% of children with PDD diagnoses. Overall specificity was low, (0.53) but improved over *DSM-IV*	More severe clinical sample, retrospective data analysis

3Di = Developmental, Dimensional, and Diagnostic Interview; ADI-R = Autism Diagnostic Interview, ADOS = Autism Diagnostic Observation Schedule; ASD = autism spectrum disorder; CARS = Childhood Autism Rating Scale; NOS = not otherwise specified; PDD = pervasive developmental disorder; Revised; SCQ = Social Communication Questionnaire; SRS = Social Responsiveness Scale.

and from 690 children with a non-PDD diagnosis, data from the Autism Diagnostic Interview, Revised (ADI-R) and Autism Diagnostic Observation Schedule (ADOS) was matched to the *DSM-5* criteria. In this study, it was found that most children who received a diagnosis of one of the PDDs under the *DSM-IV* would receive the diagnosis of ASD under the *DSM-5*.[23]

The overall sensitivity of the two were similar: the *DSM-5* criteria identified 91% of children with a clinical *DSM-IV* diagnosis of PDD, with no change based on gender or IQ. The specificity was improved in the *DSM-5*, especially when impairment in social reciprocity and nonverbal behavior was required in both the parent report and the clinical observation. Children who did not meet the *DSM-5* criteria did not demonstrate required impairments in social and communication functioning.[24] To date, data from studies on the effectiveness of ASD diagnostic criteria have been retrospective. The most stringent have used diagnostic instruments based on the *DSM-IV*; however, the two studies with the largest sample size show the highest levels of sensitivity for *DSM-5* (see Table 4).

CALLS FOR MODIFICATION OF DSM-5

Modifications to the *DSM-5* criteria already have been suggested to address these concerns of sensitivity. In addition to the proposals to reduce the criteria for social communication and interaction from three to two,[4,19,21,23,24] there have also been proposals to change the number of RRB criteria from two to one.[13,23] Another theory is that relaxing the onset criteria may improve the ability to detect early social interaction problems, thus improving sensitivity.[19,25] These changes would likely increase sensitivity while maintaining acceptable specificity.[4,21]

Work on the *DSM-5* continues, and as it nears publication more accurate estimates of sensitivity and specificity will be measured as criteria will be compared in vivo against *DSM-IV* criteria and the "gold standard" of expert diagnosis. Community and clinical populations will be assayed to provide current measures of sensitivity and specificity.[1]

Because it places primary focus on social communication problems, continued interest will explore the boundaries of normal and abnormal social communication. This is relevant because autistic traits appear to have some continuous dimension within a population.[18] The field will need suitable instruments to measure social communication skills and determine distribution in the population, assign cutoff points, operationalize mild and moderate impairment, and measure adaptive function.[6]

CONCLUSION

Philosophical questions, such as whether autism is a single, continuous entity marked by impaired social interaction, will still exist after publication of the *DSM-5*. Some will point out the broad distribution of autistic symptoms in the population[26] and elevated rates in twins and siblings.[18,27] Conversely, there is the question of whether autism is a diverse collection of heterogeneous conditions with a shared group of symptoms. Then there are those who see autism as a collection of different syndromes and who point out that latent or emerging symptoms differentiate ASD from non-ASD children,[28] a point of view supported by data on trajectory of brain development.[29] The contrast between the two groups is impetus for improved understanding of autism. The *DSM-5* fits squarely in the middle of these discussions.[30]

The *DSM-5* is the next evolutionary step for the diagnosis of autism based on the empirical input of the last 20 years of nosology and epidemiology.[31,32] As with all diagnostic systems, it is a work in progress and is the best attempt so far to describe autism as we understand it today. ■

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