Recent Advances in Autism Research as Reflected in DSM-5 Criteria for Autism Spectrum Disorder

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Abstract
This article provides a selective review of advances in scientific knowledge about autism spectrum disorder (ASD), using DSM-5 (Diagnostic and Statistical Manual of Mental Disorders, fifth edition) diagnostic criteria as a framework for the discussion. We review literature that prompted changes to the organization of ASD symptoms and diagnostic subtypes in DSM-IV, and we examine the rationale for new DSM-5 specifiers, modifiers, and severity ratings as well as the introduction of the diagnosis of social (pragmatic) communication disorder. Our goal is to summarize and critically consider the contribution of clinical psychology research, along with that of other disciplines, to the current conceptualization of ASD.
INTRODUCTION

We are entering an interesting period in the history of autism spectrum disorders (ASDs): Mental health services are moving closer to parity with traditional medicine, the numbers of children with ASD is increasing dramatically every few years, and investigators are hoping to see long-term benefits of more broadly disseminated and implemented interventions and opportunities. The almost daily mention in the media of ASD has resulted in greater demand by both policy makers and funders for evidence to support treatment approaches used in health care (and to a lesser extent in the schools) as well as justification for time spent in diagnosis and family support. In addition, the recent introduction of new criteria for ASD in the fifth edition of the Diagnostic and Statistical Manual of Mental Health Disorders (DSM-5; Am. Psychiatr. Assoc. 2013) has stimulated much discussion in the mental health community about the nature and needs of individuals on the autism spectrum. This article provides a selective review of advances in scientific knowledge about ASD, using the new diagnostic criteria as a framework for the discussion. Our goal is to summarize and critically consider the contribution of clinical psychology research, along with that of other disciplines, to the current conceptualization of ASD.

As most mental health researchers and providers know, the DSM is the product of efforts supported by the American Psychiatric Association (APA). Clinical researchers in different fields are nominated within various levels of the APA organization to revise the DSM and are vetted in terms of conflicts of interests, particularly related to relationships with pharmaceutical companies. Autism [eventually renamed autism spectrum disorders in DSM-5 (Am. Psychiatr. Assoc. 2013)] was part of the mandate of the committee on neurodevelopmental disorders, along with intellectual disabilities, speech and communication disorders, and learning disabilities. The committee’s chair was a pediatrician, Dr. Susan Swedo; other members of the committee included five child psychiatrists and five psychologists (two neuropsychologists, one clinical and developmental psychologist, one developmental psychologist, and one clinical psychologist). The committee held weekly conference calls for about two years as well as semiannual face-to-face meetings with members of other overlapping committees [such as those that worked on attention-deficit/hyperactivity disorder (ADHD) and fetal alcohol syndrome] and a yearly meeting; “homework” included data analyses, perusing the website contributions, and writing draft parts of the text. In the end, the products were the ASD criteria, background text, a framework for considering severity, and some representative cases.
Two issues central to diagnostic decisions about ASD are how ASD-related symptoms are affected by (a) age and developmental levels and (b) context and opportunities to learn or practice behaviors. These are recurring themes in child and developmental psychology more generally, and they are particularly relevant for creating schemas to identify and describe individuals with ASD in a way that leads us to useful treatment recommendations. These issues also influenced the decisions made during the DSM-5 ASD deliberations. It was hoped that by having behavioral criteria that allowed greater consideration of developmental and contextual factors, diagnostic accuracy would be enhanced and the diagnostic process could lead more directly to the development of treatment plans and the measurement of treatment outcomes.

Prior to discussion of specific criteria, a question raised was whether the new criteria should respond in some way to continued findings of increasing prevalence of autism and related disorders. The prevalence of what were formerly called pervasive developmental disorders (PDDs) increased from 1 in 110 in 2006 to 1 in 88 in 2008 to 1 in 68 in 2010 (CDC 2007, 2009, 2014). Some recent studies have suggested that the prevalence may be even higher if children with no known diagnosis are considered (Kim et al. 2011, Yeagin-Allsopp et al. 2003). Given that the majority of the increase in prevalence reported by the Centers for Disease Control and Prevention is accounted for by children with average or above average intelligence, not by those with intellectual disabilities, it has been suggested that the increase in the number of children reported to have ASD is due to the broadening of diagnostic criteria (King & Bearman 2009), first in DSM-III-R (Am. Psychiatr. Assoc. 1987) and then in DSM-IV (Am. Psychiatr. Assoc. 1994), particularly for the subtype of pervasive developmental disorders—otherwise specified (PDD-NOS). In addition, the specificity of autism diagnostic instruments, especially those based on caregiver reports, is substantially reduced in samples of children with other known psychiatric or genetic disorders (Charman et al. 2007, DiGuiseppi et al. 2010, Hus et al. 2013), which could lead to artificially inflated numbers of children with ASD in some studies. However, recent research also suggests that females with ASD may be underidentified (see Frazier et al. 2014, Giarelli et al. 2010), and there are continued disparities in the number of children identified with ASD across race, ethnicity, states, and even neighborhoods within cities, with children of color and from less advantaged backgrounds being diagnosed less often and later than other children (Mandell et al. 2009, Mandell & Palmer 2005). Thus, it is expected that prevalence numbers will increase as disparities in diagnosis decrease. Ultimately, the DSM-5 committee decided not to intentionally tighten the criteria in order to decrease prevalence, even though this has been claimed (McPartland et al. 2012). Instead, the committee decided that a primary goal of DSM-5 was to provide ASD criteria that as accurately as possible reflected behaviors that clinicians were using to diagnose ASD across the life span and across the full range of cognitive and language functioning.

Since the proposed criteria were published, a number of groups have attempted to use existing data to directly address the question of how DSM-5 criteria might change the numbers of children and adults receiving ASD diagnoses under DSM-IV (Kulage et al. 2014). The difficulty with this approach is that existing datasets do not necessarily include information about symptoms that are newly relevant in DSM-5 (e.g., sensory interests/aversions), nor do they include the additions specifically aimed at more accurately describing younger children, more intellectually able children and adults, females, and adolescents and adults. Thus, with data that are limited by information that was in DSM-IV, it is not at all surprising that sensitivity would decrease somewhat if attempting to map DSM-IV items onto DSM-5 criteria. In two larger samples that included more comprehensive symptom-level data, sensitivity did not differ substantially between DSM-IV and DSM-5 (Frazier et al. 2012, Huerta et al. 2012).
ELIMINATION OF DIAGNOSTIC SUBTYPES WITHIN AUTISM SPECTRUM DISORDER

Probably the most significant change arising from DSM-5 is the consolidation of the various subtypes of PDD into a single diagnosis of ASD. This change came about in response to a large literature that found that diagnoses of the different subtypes (particularly Asperger’s disorder and PDD-NOS) were not reliable across clinicians (Lord et al. 2011) or time (Lord et al. 2006). Although the concept of Asperger’s disorder has been valuable in calling attention to the fact that significant basic social deficits can occur in individuals without intellectual or language delays, differentiating Asperger’s from autism without language delay has been very difficult to do without circular research designs (e.g., defining individuals with Asperger’s disorder as having higher verbal skills than nonverbal skills and then finding that an autism group has nonverbal skills that are higher than those of the Asperger’s group). Several studies have found that people diagnosed with Asperger’s disorder cannot be clearly differentiated from those with high-functioning autism, likely in part because the majority of people diagnosed with Asperger’s disorder actually met DSM-IV autism criteria (Bennett et al. 2008, Kamp-Becker et al. 2010, South et al. 2005, Woodbury-Smith et al. 2005). Furthermore, an analysis of data from a large multisite study found that clinician diagnoses of any of the ASD subtypes (i.e., Asperger’s, PDD-NOS, autism) were more related to the site where the clinician worked than to any characteristic of the child being diagnosed, except algorithm scores on the Autism Diagnostic Observation Schedule (ADOS) (indicating severity) and sometimes verbal IQ (Lord et al. 2011). Results suggested that each of 12 different sites appeared to use a different constellation of behaviors, including age and IQ, to differentiate autism from PDD-NOS and Asperger’s disorder. Thus, clinicians did differentiate among the subtypes, but each clinic differentiated in a different way, rendering the subtypes meaningless across locations, except as a general index of severity.

NUMBER OF DEFINING DOMAINS

Another, less controversial, change to ASD criteria that also arose directly from research was the shift from three domains [social deficits, communication deficits, and restricted repetitive behaviors (RRBs)] to two (social-communication impairments and RRBs). This decision was based on repeated findings that differentiations between the DSM-IV communication and social criteria were arbitrary. Analyses of large datasets from DSM-IV-based instruments, such as the Autism Diagnostic Interview-Revised (ADI-R) (Rutter et al. 2003) and the ADOS (Lord et al. 1999) showed that most items from the DSM-IV social and communication domains loaded onto a single factor that included verbal and nonverbal communication and social behaviors (e.g., Constantino et al. 2004, Frazier et al. 2008, Georgiades et al. 2013, Gotham et al. 2007, Lecavalier et al. 2006, Mandy et al. 2012). Items differed more by level of analysis—for example, the DSM-IV item “marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction” describes specific behaviors by an individual, whereas “a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people” describes a construct that could in fact be operationalized by lack of gaze, facial expression, and gesture. Positive features of abnormal language use, such as stereotyped language, delayed echolalia, and verbal rituals tended to load with repetitive behaviors rather than with social or communication abnormalities. Although behaviors listed as repetitive are less strongly related to each other than to social and communication deficits in ASD, they do form a constellation of behaviors that is distinct from social communication, perhaps because these behaviors that are not generally seen in typical development beyond infancy and early
preschool years. Social-communication deficits are generally defined in terms of the absence or diminution of typical behaviors rather than the presence of abnormal behaviors, although there are some exceptions to this (e.g., see Wing & Gould 1978). In fact, in some analyses, “odd” social communication behaviors, such as the use of someone’s body as a tool for communication or unusual intonation, load with RRBs (Gotham et al. 2007), although this is not addressed in DSM-5.

CHANGES WITHIN DOMAINS OF AUTISM SPECTRUM DISORDER

Changes were made from DSM-IV to DSM-5 both in the constructs that defined each ASD domain and also in the number of criteria that had to be met for a diagnosis. The general purpose was to use a smaller number of broader principles to define DSM-5 autism in a way that allowed an individual clinician to describe how ASD was reflected in an individual patient—male or female, young or old, and across cultures. With broader principles, which included deficits in (a) social-emotional reciprocity; (b) nonverbal communicative behaviors used for social interaction; and (c) developing, maintaining, and understanding relationships and adjusting to suit various social contexts, the commonalities across individuals with ASD of different developmental levels, cultures, and genders could be better reflected. This avoided the multiple combinations of possibilities for arriving at an autism diagnosis under DSM-IV (Carey 2012) and better exemplified the similarities that do exist within the syndrome while recognizing the diverse ways that these core deficits can be manifested. Thus, to receive a diagnosis of ASD, an individual must currently exhibit (or be reported to have previously exhibited) difficulties in each of these three areas of social communication. The areas are sufficiently broad so that examples can be drawn from each of them for a person of any age or language level and across cultures, where differences exist in whether various behaviors are considered natural or inappropriate (e.g., standing closely to others, expecting eye contact). Unlike those of DSM-IV, the principles within the DSM-5 social communication domain are not listed as a finite set of criteria but rather as broad principles that a clinician, parent, or self-advocate can exemplify in many ways.

The use of a smaller number of broader principles also leaves room for changes across development within the ASD domains, a phenomenon that is very well documented in research (e.g., Charman et al. 2005, Richler et al. 2010, Shattuck et al. 2007). This different strategy for defining criteria also avoids moving an individual from one subtype of PDD to another across development (for example, moving from an early diagnosis of PDD-NOS to autism as more symptoms develop and the diagnosis becomes more clear, then moving to Asperger’s disorder as language emerges). Diagnostic accuracy can also be enhanced by not focusing on specific symptoms that are only relevant to specific age groups or certain types of individuals with ASD. For example, not responding to one’s name and not following another person’s pointing gesture point are strong predictors of autism in toddlers and younger preschool children but not in older children or adults (Gotham et al. 2007, 2008). Not having close friends is associated with autism in school-age children and young adults but is not a developmentally appropriate factor for very young children, and it becomes a more complicated factor in adults as it becomes more difficult to disentangle issues related to social motivation and social preference from social ability (Bishop & Seltzer 2012). Thus, having broader principles allows the clinician (and advocates for an individual) to provide developmentally appropriate examples.

Well into the process of creating the DSM-5 definition of ASD, it was proposed that criteria should be able to be met either by current examples or by history. Initially, analyses of large existing datasets supported this strategy for RRBs because a substantial proportion of individuals with ASD (especially those without intellectual disabilities) exhibited RRBs as young children but not as adolescents or adults (Eshensen et al. 2009, Shattuck et al. 2007). However, researchers
working with adults with ASD, as well as self-advocates, proposed that the same strategy would be appropriate for social communication deficits, which show significant abatement over time for some individuals with ASD (Anderson et al. 2014, Shattuck et al. 2007). Importantly, to receive a diagnosis of ASD, the individual must still show impairment in current functioning (even if the specific criteria are met by history). Nevertheless, an obvious drawback to allowing criteria to be met either currently or by history is that specificity could be greatly reduced in some groups (e.g., individuals with severe language delays or intellectual disabilities without ASD). The data from older children and adolescents suggested that by far the greatest majority of those with DSM-IV diagnoses of any PDD met DSM-5 social communication criteria currently (Huerta et al. 2012), suggesting that perhaps the historical option was not necessary. However, far fewer data are available for adults than children, and the committee elected to value sensitivity over specificity, a trend that extends to DSM-5 generally and that has been criticized (Frances 2013).

For the most part, DSM-IV criteria can be easily mapped onto the DSM-5 domains and principles, although, as we discuss below, the converse is not true. The one deliberate exception to the inclusion of all DSM-IV criteria within the DSM-5 domains is that delayed language acquisition and the failure to use verbal language is now no longer considered part of ASD criteria. This change was based on a number of different pieces of information. First is the recognition that the diagnosis of ASD has expanded to include individuals who are not language delayed. Although many children with ASD do show delays early on, the proportion of older children and adolescents with ASD who do not have delays in the structural aspects of language has increased steadily, in part because the definition of ASD has broadened and in part, one hopes, because of increasing access to better education and early intervention (see Lord & Bishop 2010). Second, mild early language delays are quite common in many conditions and are not specific to autism (Robins et al. 2014, Wetherby et al. 2002); severe language delays are less common in older children and adults and quite handicapping but also are not entirely specific to autism (Tureck et al. 2013). Thus, it was felt more appropriate for language delay and disorder to be coded independently as separate diagnoses rather than included as criteria for ASD. Overall, the goal was to encourage clinicians to pay closer attention to a patient’s language ability before beginning to consider ASD as a possible diagnosis, with the idea that this would require better understanding of an individual’s strengths and weaknesses and result in more appropriate treatments.

Within the domain of RRBs, DSM-5 also included a number of changes in organization and constructs. As in the social communication domain (with the exception of language delay), items from DSM-IV are all subsumed into the new principles in DSM-5. Four principles are described: (a) stereotyped or repetitive motor movements, use of objects, or speech; (b) insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior; (c) highly restricted, fixated interests that are abnormal in intensity or focus; and (d) hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment. Though extensive data exist about the presence of each of the defined RRBs in children with autism (Cuccaro et al. 2003, Honey et al. 2012, Mandy et al. 2012, Szatmari et al. 2006), the groupings themselves were not empirically derived. Factor analyses of repetitive behavior items generally support the presence of two types of repetitive behavior: repetitive sensory motor, which includes most items from principles a and d except stereotyped speech; and insistence on sameness, which is represented by principle b (see Bishop et al. 2013). Some evidence also suggests the presence of a third separate grouping of behavior, fixated interests (Lam et al. 2008), but most studies have found that interests that are unusual in intensity load with insistence on sameness, and interests that are unusual in content load with repetitive sensory motor behaviors (Bishop et al. 2013).

In addition, behaviors related to what seem to be differences in sensory perception, including sensory aversions and negative reactions (e.g., to certain foods, specific sounds) as well as sensory...
seeking (e.g., peripheral vision, smelling things), have been added to DSM-5. Though not previously recognized as part of the official criteria in DSM-IV, these behaviors have been described in many individuals with ASD (Baranek et al. 2013, Lane et al. 2010) and are an important part of personal accounts by self-advocates (Grandin 2008). These sensory-related behaviors now constitute a fourth subdomain within RRBs.

In DSM-IV, an individual needed to have only a single example of an RRB to meet diagnostic criteria for autism or Asperger’s disorder, and no RRB was needed for to meet PDD-NOS criteria. However, when datasets were examined, almost all individuals with any ASD (i.e., PDD-NOS, Asperger’s, or autism) had behaviors from at least one RRB subdomain, and when history was taken into account, the vast majority had behaviors from at least two or even three of the different DSM-5 subdomains (Huerta et al. 2012). Having a history of behaviors in only one of the four subdomains was not specific to autism. What is interesting is that some of the repetitive behaviors in ASD are very striking, particularly when they occur in combination (for example, a child who uses peripheral vision and stands in an unusual posture while spinning himself or while spinning the wheels of an overturned bike), but others are behaviors that occur in significant minorities of children at any given time (such as hand-flapping in toddlers, aversion to tags in clothing, or restricted interests in video games or Disney movies). This indicates that a balance exists between having sufficiently liberal criteria to include a behavior in ASD that interferes with daily life and requiring enough of these behaviors to differentiate ASD from other disorders and from typical development. We hope that future, more detailed research across development and in large samples will compare people with ASD to other clinical populations to provide further insight into what behaviors best predict and differentiate ASD. In addition, theoretical constructs from neurobiological studies of brain function and of genetic syndromes may provide important clarifications about the contribution of RRBs to ASD.

DELETION OF AGE-OF-ONSET CRITERIA

Much has been learned about the development of behaviors associated with autism in the early years from studies of infant siblings of children with autism (e.g., Ozonoff et al. 2011b, Zwaigenbaum et al. 2005) and from studies in which children suspected of autism in the very early years have been followed (Ellis Weismer et al. 2010, Guthrie et al. 2013, Lord et al. 2012). Findings about subtle differences in development that are evident in children with ASD before parents suspect the disorder or before clinicians can reliably diagnose it have led to a much more careful use of the terms “age of onset” and “age of recognition” (Volkmar et al. 2004).

For a child to have DSM-IV autism, he or she had to have onset of some kind of symptom (including delayed language) related to autism prior to age 3; otherwise, a child who met autism criteria in every other way would receive a diagnosis of PDD-NOS or, if the child’s language and cognitive development were not delayed, a diagnosis of Asperger’s disorder. However, research in the past 20 years has provided important information that has a bearing on decisions for DSM-IV about age of onset as a criterion for ASD. First, numerous studies have indicated that some, although not all, children who are later diagnosed as having ASD have delays in motor development, social responsiveness, or language development in the early part of the second year of life, if not before (Landa & Garrett-Mayer 2006, Messinger et al. 2013, Mitchell et al. 2006, Zwaigenbaum et al. 2005). Although the presence of these difficulties has emerged in several studies, findings have differed in each study, so more research is needed. Second, neurobiological studies of both pathology and genetics suggest that the mechanisms behind autism are in place during fetal development (Stoner et al. 2014), so from a biological standpoint, onset may be before birth. Third, although deficits and delays in children who will develop autism can be documented...
on a group level, diagnosing individual children with ASD prior to age 15–18 months is very difficult (Luyster et al. 2009, Zwaigenbaum et al. 2005). Thus, providing a date of actual onset is rarely possible. Furthermore, many factors, both internal and external to a child, affect when he or she first receives a diagnosis. These include ethnicity and race, access to appropriate services, bilingual status and region of the country, as well as whether the child has delays besides those that characterize autism, such as in motor coordination or cognitive development (e.g., Mandell et al. 2009, Shattuck et al. 2009).

Given the relationships among the age of diagnosis or recognition and sociocultural and developmental factors, discriminating between autism and other subtypes of PDD on the basis of age of onset does not seem fruitful. In addition, several studies have shown that simple definitions of language delays (such as no meaningful spoken words by age 24 months or no spontaneous, non-repetitive two- or three-word sentences by age 33 or 36 months) do not predict different symptom patterns (i.e., Asperger’s disorder versus high-functioning autism) when current language skill is controlled (Eisenmajer et al. 1998). In addition, studies of the accuracy of parent reports of language milestones, earlier social skills, and children’s loss of social and language skills suggest that many factors (including age at reporting and birth order) affect these data (Hus et al. 2011, Jones et al. 2014, Ozonoff et al. 2011a). Consequently, differentiating children or adults with ASD by age of first words or phrases, particularly as a marker of onset, is less meaningful than a description of a concurrent language delay.

Regression is another factor related to patterns of onset that was considered in differentiating PDD subtypes under DSM-IV. A recent meta-analysis suggested a mean prevalence of parent-reported regression of 32% (Barger et al. 2013), but rates vary depending on how stringent a definition is employed (Hansen et al. 2008, Stefanatos 2008). For example, recent studies suggest that for many children with ASD, regression may take the form of a subtle plateauing, loss of engagement, or failure to acquire more sophisticated social-communication skills as expected rather than an identifiable loss of actual skills (e.g., Brian et al. 2014, Ozonoff et al. 2010).

In DSM-IV, an attempt was made to acknowledge that the loss of skills across systems (for example, including loss of language, motor, and adaptive skills as well as social intent) following a period of clearly typical development up to at least two years of age was different from the more common phenomenon of increased social withdrawal and/or decreased use of vocal communication or gestures occurring in the second year of life. This acknowledgment took the form of a separate diagnosis, within PDD, called childhood disintegrative disorder (CDD). The intent was to discriminate CDD from the more commonly described regressions occurring in younger children who developed ASD (Rosman & Bergia 2013). However, given the extreme rarity of this level of multisystem loss of skills occurring in a child whose development had been truly normal up to that point, CDD was removed as a separate diagnosis from DSM-5. With improvements in neurological and genetic testing, children with degenerative conditions or rare forms of epilepsy, who in earlier years might have received diagnoses of CDD (Volkmar & Rutter 1995), now receive more specific diagnoses (e.g., Rett syndrome) or specifiers as necessary.

The end result of these research findings was the recommendation that different patterns of onset and recognition should be acknowledged in making a DSM-5 diagnosis of ASD but should not be used to differentiate subtypes, at least at this point. As larger samples of children with ASD become available, the goal is to obtain markers of different patterns of development for use in future studies.

**NEUROBIOLOGICAL SPECIFIERS**

Over the past 50 years, ASD has gradually become accepted as a neurobiological disorder. In the past 10 to 15 years, extensive genetics research has been conducted, with continued neuroimaging
studies and, most recently, molecular biology and animal models. Major advances have occurred within basic science but to date have had little practical value for individuals with ASD. Attempts to group mental disorders by biological substrates have been made (Hyman 2010, Karalunas et al. 2014), but more research is required to establish meaningful links to behaviors, developmental course, or treatment (Sisti et al. 2013). For DSM-5, one goal was to disentangle some of the genetically determined DSM-IV diagnoses, such as Rett syndrome and fragile X syndrome, from ASD and to be very clear that the diagnosis of ASDs is a purely behavioral description of a constellation of symptoms. The concept of “specifiers,” somewhat like what was formerly Axis III for the World Health Organization’s ICD-10 (International Statistical Classification of Diseases and Related Health Problems, tenth revision) and the APA’s DSM-IV, allows the separate demarcation of genetic or medical conditions that may accompany the behavioral diagnosis. Thus, an individual can have ASD and fragile X syndrome, or ASD and Rett syndrome, or ASD and Down syndrome. As hundreds of genetic risk factors are identified (Murdoch & State 2013, Ronemus et al. 2014), each can be considered a specifier associated with the behavioral diagnosis of ASD. The assumption is that the same specifiers can also accompany other developmental and/or psychiatric diagnoses (e.g., not all individuals with fragile X have ASD).

Specifiers can also include other risk factors or biological conditions besides genetic variants, such as epilepsy or very low birth weight or congenital anomalies. Factors such as extreme pre-maturity may function as risk factors for autism or, as in the case of epilepsy, may themselves be increased in cases of ASD. Thus, the relationship between behavioral characteristics and biological specifiers is neither unidirectional nor unidimensional but does provide a mechanism to denote the co-occurrence of biological conditions with ASD. In some ways, working from the top down rather than the bottom up complements an approach like the National Institute of Health’s Research Domain Criteria, which attempts to delineate discrete biologically based behavioral dimensions by working up to behavior from biological substrates (Hyman 2010, Insel et al. 2010). DSM-5 ASD starts with the behavior but leaves room for clinicians to identify concomitant biological conditions through specifiers. To date, only a relatively small number of potential specifiers are associated with distinct behavioral profiles within (or outside of) ASD, but it is hoped that with greater knowledge, these specifiers will provide information beyond describing their own identification (Rutter 2011). For example, individuals—even those with comorbid ASD—who have Down syndrome have a particular physical and cognitive profile (Warner et al. 2014), as do individuals with ASD, Rett syndrome, and fragile X syndrome (McDuffie et al. 2014). In contrast, it has been more difficult to identify specific behavioral phenotypes in association with other genetic patterns or copy number variants (e.g., 16p11.12 deletion), perhaps because of their rarity and consequently limited sample sizes (e.g., Hanson et al. 2014).

MODIFIERS

In accordance with the beginnings of a more dimensional approach to psychopathology and developmental disorders, modifiers—meaning other behavioral diagnoses that reflect comorbidity—can now accompany a diagnosis of ASD. This shift reflects a reorganization of what previously existed in DSM-IV and currently exists in ICD-10 as multiaxial diagnoses. For ASD, diagnoses of intellectual disability and language disorder are most relevant. In addition, it is newly permissible under DSM-5 to diagnose ADHD concurrently with ASD.

The associations between ASD and both intellectual disability and language disorders are well documented. The proportion of individuals with ASD who have either intellectual disability or significantly delayed language has changed dramatically in the past 20 years, probably due to broadened definitions of ASD, earlier and more effective interventions, and greater awareness of
milder cases (Zahorodny et al. 2014). Nevertheless, language skills and IQ (nonverbal at early ages; verbal at later ages) are the strongest predictors of outcome both in naturalistic studies (Anderson et al. 2014) and in response to treatment (Beglinger & Smith 2005, Howlin et al. 2004, Sallows & Graupner 2005).

It has long been known that symptoms of hyperactivity and inattention are common in children with ASD. In DSM-IV, there was a concern that children with ASD would be misdiagnosed as having ADHD because of inattention that resulted directly from ASD symptoms, such as lack of social engagement and repetitive motor mannerisms. However, the balance between avoiding misdiagnoses and recognizing the effect that ADHD symptoms have on a child with ASD has shifted because a high frequency of ADHD in children with autism has been documented (Leitner 2014, Simonoff et al. 2008). In addition, it has become increasingly apparent that, at least for older children with ASD, the most effective treatments are those that properly address comorbidities. Effective treatments for ADHD exist, and ethically it would be wrong to fail to consider these treatments for children with ASD and ADHD. Thus, ADHD can now be included as a modifier, along with depression, anxiety disorders, and other psychiatric diagnoses. Like ADHD, these other diagnoses are now recognized to occur commonly in individuals with ASD, including lifetime estimates for depression as high as 25% (Leyfer et al. 2006) and anxiety as high as 35% (van Steensel et al. 2011). Comorbidities are particularly common among higher-functioning adolescents and adults. Recent research has even suggested that suicidality may be increased among some adolescents with ASD, which further highlights the need to attend very carefully to psychiatric symptoms that may co-occur with ASD (Mayes et al. 2013).

SEVERITY METRICS

Though it is widely recognized that substantial variability exists in symptom severity among individuals with ASD, it can be very difficult to separate ASD-related severity from IQ and language level (e.g., see Weitlauf et al. 2014). IQ and language level are closely tied to overall functioning and outcome in ASD (Anderson et al. 2014, Howlin et al. 2013), so for practical purposes, knowing an individual’s IQ and functional language ability usually serves as a reasonable proxy for how severely affected the individual is. On the other hand, even within narrower bands of ability (e.g., considering only individuals with IQ under 70 or only individuals with single-word speech) there is wide variability in ASD symptom severity. The ability to denote these varying degrees of severity is important for clinical purposes, such as when describing similarities among or differences between individuals with ASD, discussing prognosis, or measuring within-person change. A severity metric is also of great value for research aimed at understanding developmental trajectories, measuring response to treatment, or investigating etiology, including studies that compare symptom profiles between children with known genetic causes of ASD and idiopathic ASD (McDuffie et al. 2014, Thurman et al. 2014).

To address the need for an ASD severity metric, standardized scores have been developed for the ADOS-2 (Gotham et al. 2009, Hus et al. 2013, 2014). The ADOS-2 was designed as a diagnostic measure and not a measure of severity or change, but because it is widely used by clinicians and researchers, the hope was that standardized scores could serve as an immediately accessible means of capturing the severity of ASD-related deficits (as exhibited in the context of the ADOS-2 assessment). These scores have little relation to age or IQ (Kanne et al. 2011, Shumway et al. 2012), and some evidence suggests that they may be useful for identifying different trajectories of ASD severity (Gotham et al. 2012). Still, despite showing promise as a way to measure severity beyond the influence of age and IQ, these standardized scores are only standardized within each ADOS-2 module, meaning that children of similar language levels are compared to each other.
Thus, the fact that a child of a particular age (e.g., age 10) receives module 1 of the ADOS-2 (which is designed for children with single-word speech or who are preverbal) as opposed to module 5 (which is designed for children and adolescents with fluent speech) may be as important in denoting his/her functional level as is his/her actual severity score on the instrument. More research is needed to determine the utility of the ADOS-2 standardized scores as a meaningful measure of ASD severity. In addition, the development of instruments that can measure changes in ASD symptom domains outside of the ADOS context is an important research priority.

In response to a general requirement of DSM-5 for all disorders, DSM-5 ASD criteria include specifiers about severity. For DSM-5, “severity” has been operationalized as the degree of support required. A clinician indicates whether “support,” “substantial support,” or “very substantial support” is required for social communication impairments and separately for restricted and repetitive behaviors. These severity indicators are new in DSM-5 and were not tested prior to the publication of the new criteria. Thus, it is not yet clear how the indicators will function in practice. Of particular concern is that it is not specified what methods clinicians should employ to determine the level of severity. Weitlauf et al. (2014) used retrospective data analysis to compare three levels of severity across the ADOS comparison score, cognitive ability, and adaptive functioning and found significant discrepancies. Concordance was highest for individuals classified as “severe” across all three measures, whereas agreement between the measures was much lower for individuals scoring in the mild or moderate ranges. The authors suggest that because of how the severity indicators are worded in DSM-5 (i.e., “level of support”), and in the absence of explicit guidelines about how to apply these indicators, clinicians may rely more on information about cognitive and adaptive functioning than on measures of actual ASD symptom severity (Weitlauf et al. 2014).

SOCIAL (PRAGMATIC) COMMUNICATION DISORDER

The change in diagnostic criteria to require repetitive behaviors for a diagnosis of ASD has brought about a concern that children who were formerly diagnosed with PDD-NOS might not receive any diagnosis under DSM-5 (see Kulage et al. 2014). An investigation of large datasets that included multiple questions about behavior, such as the ADI-R or the Social Responsiveness Scale (Constantino & Gruber 2005), indicated that few children with any former PDD diagnosis (including PDD-NOS) would fail to meet ASD criteria (Frazier et al. 2012, Huerta et al. 2012). Studies have also shown that most children with PDD-NOS diagnoses have a history of more than one repetitive behavior (Lord et al. 2006, 2011). Studies that used more limited datasets, such as the DSM-IV checklist or the ADDM reviews of existing reports (Maenner et al. 2014, McPartland et al. 2012), found less evidence of repetitive behaviors in children with PDD-NOS, which suggests that it is very important that clinicians collect adequate information about these behaviors.

Nevertheless, continued concern from some professionals about children who have impairing social deficits that affect communication without concomitant repetitive behaviors ultimately resulted in the proposal of a new diagnosis for DSM-5: social (pragmatic) communication disorder (SCD). This disorder is not considered an ASD but instead is grouped with communication disorders. It requires that ASD be ruled out and that the symptoms are not accounted for by intellectual disability or general language delay. The focus is on social communication deficits defined in very similar ways as those in ASD. Whether children or adults receiving diagnoses of SCD can be reliably distinguished from those with ASD or other disorders when cognitive level, structural language skills, and source of information are controlled is an open question (Norbury 2014). There is a concern that this diagnosis will be incorrectly used in lieu of ASD diagnoses because it sounds less severe and requires less information to document (Lord & Jones 2012). However,
without the SCD diagnosis or something like it, there may be a population of individuals with significant social communication deficits who might not meet criteria for ASD because of lack of RRBs and who might therefore be excluded from receiving services (Gibson et al. 2013). Also, it is unclear how schools will serve children with SCD and how insurance companies will treat this diagnosis. In the absence of large-scale field trials for DSM-5, these are questions that will have to be examined now that DSM-5 is in use.

### SUMMARY AND CONCLUSIONS

DSM-5 ASD criteria represent an attempt to respond to recent developments across multiple areas of ASD research, ranging from clinical research findings about the structure of ASD symptoms and differential diagnosis at different points in development to insights from basic scientists about the association between ASD and identifiable genetic or neurobiological abnormalities. Researchers have identified a wide range of risk factors for ASD (e.g., older maternal and paternal age, rare genetic variants), but it is clear that no single path leads to the development of this disorder. Rather, there may be hundreds of etiologies underlying the manifestation of ASD. DSM-5 directly acknowledges this by allowing ASD to be diagnosed in cases where a specific etiology is suspected as well as in cases with no identified biological association. Thus, despite having known neurobiological roots, DSM-5 presents ASD as a behavioral diagnosis that should be used to describe a particular constellation of social-communication impairments and RRBs that might occur in the context of a variety of biologically based conditions. Furthermore, other behavioral dimensions not specifically accounted for by these social-communication impairments and RRBs, such as cognitive or language impairment, inattention/hyperactivity, or depressive or anxiety symptoms, can be indicated alongside the ASD diagnosis.

Significant changes to established systems are never without controversy. Some investigators have proposed that DSM-5 will result in decreased prevalence of ASD, particularly among individuals with higher cognitive and language abilities and/or those with milder symptoms (e.g., McPartland et al. 2012), whereas others suggest that nearly all those who received a DSM-IV diagnosis of PDD would receive an ASD diagnosis in DSM-5 (e.g., Huerta et al. 2012). Moreover, analyses that include both ASD and non-ASD participants highlight the poor specificity that is achieved when only symptom counts (from DSM-IV or DSM-5) are considered (e.g., Huerta et al. 2012). This poor specificity underscores the need to be thoughtful about how written criteria are applied to patients. Clinicians and researchers must be careful about how they obtain and interpret information relevant to an ASD diagnosis. Of particular importance is a recognition that context—in space (e.g., at home versus at school versus in the clinic), time (e.g., during toddlerhood versus in the adult years), and development (e.g., in the presence of intellectual disability or language impairment versus normal IQ or language ability)—is paramount when assessing social-communication and repetitive behaviors. Obtaining information from multiple sources is also critical in making a valid and reliable diagnosis of ASD (e.g., Risi et al. 2006). Thus, as was the case under DSM-IV and other previous editions of the DSM, the onus remains on clinicians to collect high-quality information about behaviors relevant to a diagnosis of ASD in order to determine (a) whether an ASD diagnosis is warranted and (b) what other specifiers, modifiers, and/or comorbid diagnoses are appropriate given the individual’s specific symptom profile. This second step is necessary to ensure that a diagnosis, as the immediate end result of a comprehensive diagnostic evaluation, contains sufficient information to provide a preliminary roadmap for individualized intervention that can then be adapted based on the priorities of the individual and/or the family (see Lord & Bishop 2010).
SUMMARY POINTS

1. DSM-5 criteria for autism spectrum disorder eliminate the PDD subtypes that were present in DSM-IV and introduce significant changes to the organization of ASD symptoms, although conceptualizations about which symptoms are core to the disorder remain largely unchanged.

2. Specific domain criteria were written in a way that directly acknowledges the heterogeneity of ASD symptom presentation among individuals of different ages, language levels, and cognitive abilities.

3. Specifiers and modifiers further acknowledge the substantial heterogeneity among individuals with ASD while also clearly recognizing ASD as a behavioral diagnosis with multiple etiologies.

4. Retrospective analyses of large ASD data repositories indicate that the majority of individuals with DSM-IV PDD will continue to meet criteria for ASD under DSM-5, but prospective studies are needed to understand how diagnoses of ASD and social (pragmatic) communication disorder, as well as new ASD specifiers, modifiers, and severity ratings, will function in practice.

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Catherine Lord receives royalties from sales of the Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS). Catherine Lord and Somer Bishop receive royalties from sales of the Autism Diagnostic Observation Schedule, 2nd edition (ADOS-2). Both authors donate all proceeds related to their own clinical or research activities to charity.

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