

ISSUES RELATED TO IDENTIFICATION OF CHILDREN WITH AUTISM SPECTRUM DISORDERS (ASDs): INSIGHTS FROM DSM-5

Abstract: The article examines issues related to identification of children with autism spectrum disorders (ASDs). The focus is on the Diagnostic Criteria in DSM-5 diagnostic features, associated features supporting diagnosis, and prevalence are discussed.

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INTRODUCTION

The American Psychiatric Association's *Diagnostic and Statistical Manual of Mental Disorders* (DSM) is a classification of mental disorders with associated criteria designed to facilitate more reliable diagnoses of these disorders. Autism spectrum disorder is a new DSM-5 name that reflects a scientific consensus that four previously separate disorders are actually a single condition with different levels of symptom severity in two core domains. It has become a standard reference for clinical practice in the mental health field. DSM has been used by clinicians and researchers from different orientations (biological, psychodynamic, cognitive, behavioural, interpersonal, family /systems), all of whom strive for a common language to communicate the essential characteristics of mental disorders presented by their patients.

The criteria are concise and explicit and intended to facilitate an objective assessment of symptom presentations in a variety of clinical settings—inpatient, outpatient, partial hospital, consultation- liaison, clinical, private practice, and primary care—as well in general community epidemiological studies of mental disorders. DSM-5 is also a tool for collecting and communicating accurate public health statistics on mental disorder morbidity and mortality rates (American Psychiatric Association 2013).

This manual introduces neurodevelopmental disorders, among which is autism spectrum disorder which is characterized by persistent deficits in social communication and social interaction across multiple contexts (deficits in social-emotional reciprocity, non-verbal communication and developing relationships), as well as restricted, repetitive patterns of behaviour, interests, or activities (American Psychiatric Association 2013).

The manual changed subsuming several PDD subtypes into a single diagnosis of autism spectrum disorder in the DSM-5 that reflected a wide-spread consensus that autism is best considered as existing on a spectrum with variable manifestations across lifespan, gender, and intellectual level and/or language.

SUBTYPES SUBSUMED UNDER A SINGLE DIAGNOSIS

In the DSM-5 (American Psychiatric Association 2013) four PDD subcategories specified in DSM-IV of autistic disorder, Asperger's disorder (syndrome), childhood disintegrative disorder (CDD), and pervasive developmental disorder not otherwise specified (PDD-NOS) are now subsumed into the one broad category of autism spectrum disorder. The subtype terms will no longer be used diagnostically under the DSM-5.

The PDD subtype Rett's Disorder (syndrome) is excluded from the new ASD category. This is because the DSM focuses on disorders that can be defined behaviourally, without a molecular or biological test (Kurita 2011). Rett's syndrome is a single gene neurological disorder in which those with the condition may go through a phase of social impairment, language regression and repetitive motor mannerisms resembling autism (Rutter and Uher 2012).

Terminology has also changed. The name pervasive developmental disorder (PDD) has now been changed to Autism Spectrum Disorder (ASD). The term “mental retardation” has been replaced with the term “intellectual disabilities”.

AUTISM SPECTRUM DISORDERS (ASDs), PDD-NOS AND ASPERGER SYNDROME

DSM-5 folds Asperger syndrome and PDD-NOS into the category of autism spectrum disorder. The aim is to produce a clearer and simpler diagnostic system with improved recognition and diagnosis for those on the autism spectrum across all ages and ability levels. The main objective of the revision to the DSM relating to ASD was to increase the specificity of diagnosis, that is, make it easier to identify ASD as distinguished from other non-autistic disorders and to increase the stability of diagnosis over time (Skuse 2012). Having a single diagnostic entity avoids the problem of an individual receiving serial or sometimes concurrent, diagnoses of PDD-NOS, autism and Asperger syndrome, depending on the clinician they see or maturation. Numerous studies have reported little qualitative difference between autism disorder and Asperger Disorder subtypes (Broadstock 2014). Happé (2011) suggests that the DSM-5 changes reflect that it is a time to

reintegrate Asperger disorder with the rest of the spectrum and to demand the same level of respect and lack of stigma for individuals across the full range of manifestations of the spectrum.

Removal of Asperger syndrome and PDD-NOS as distinct disorder classifications from the DSM reflects multiple concerns regarding the reliability and application of these diagnoses when applying their DSM-IV diagnostic criteria. These include that: in clinical field trials, ASD experts frequently make different diagnoses based on the same presenting symptoms in the same individual; the boundary between Asperger syndrome and autism is not clear on a population basis; and in up to half of patient diagnoses of autism, Asperger's Disorder and PDD NOS are not stable within the same individual over time (Broadstock 2014).

DIAGNOSTIC CRITERIA IN DSM-5

To be diagnosed with ASD, a person needs to fulfil the following criteria (American Psychiatric Association 2013):

1. Persistent deficits in social communication and interaction across multiple contexts, as demonstrated by all of the following:
 1. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and inability to have normal back-and-forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions.
 2. Deficits in nonverbal communicative behaviours used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
 3. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behaviour to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absence of interest in peers.
 4. (These criteria can be currently occurring or have occurred in the patient's past. Examples are illustrative, not exhaustive.)
 2. Restricted, repetitive patterns of behaviour, interests, or activities, as manifested by at least two of the following:
 1. Stereotyped or repetitive motor movements, use of objects, or speech (e.g., repetitive hand flapping, lining up toys or flipping objects, delayed or immediate parroting of others' speech, idiosyncratic phrases).
 2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behaviour (e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food every day).
 3. Highly restricted, fixated interests that are abnormal in intensity or focus (e.g., a child who is extremely attached to a spoon, an adult who spends hours rewriting specific phrases).
 4. Extremely exaggerated or dulled reactions to sensations or unusual interest in sensory aspects of the environment (e.g., apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement).
 5. (These criteria can be currently occurring or have occurred in the patient's past. Examples are illustrative, not exhaustive.)
 3. Symptoms must be present in the early developmental period. Though, symptoms may not become fully apparent until social demands exceed limited capacities. Symptoms may also be masked by learned strategies in later life.
 4. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning.
 5. These disturbances are not better explained by intellectual disability (intellectual developmental disorder) or global developmental delay. Intellectual disability and autism spectrum disorder frequently co-occur. Social communication should be below what is expected for general developmental level, in order to make comorbid diagnoses of autism spectrum disorder and intellectual disability.
- In DSM-5, sensory abnormalities are newly included as a core, diagnostic symptom of the restricted, repetitive patterns of behaviour (RRB) domain. This change is based on empirical

research finding that this symptom has sensitivity and specificity as a diagnostic criterion for ASD, and factor analytic evidence that places it in the RRB symptom dimension. This improves the relevance of the criteria to younger children with ASD, because sensory issues are common concerns in this population (Broadstock 2014).

SYMPTOM ONSET

The time of diagnosis may be much later than the time of actual onset of a disorder. The DSM-5 acknowledges that ASD may not manifest itself fully in infancy because of difficulty in identifying early signs, poor parental recall, and minimal social demands made of children in the early years. As requirements for social abilities increase with age, social impairments become more apparent (Lauritsen 2013). For these reasons, the requirement for symptom onset before 3 years has been changed to “the early developmental period” (Broadstock 2014).

PREVALENCE

In recent years, reported frequencies for autism spectrum disorder across U.S. and non- U.S. countries have approached 1% of the population, with similar estimates in child and adult samples. It remains unclear whether higher rates reflect an expansion of the diagnostic criteria of DSM-IV to include subthreshold cases, increased awareness, differences in study methodology, or a true increase in the frequency of autism spectrum disorder (Abdallah and Eissa Saad 2014).

RISK AND PROGNOSTIC FACTORS

The best-established prognostic factors for individual outcome within autism spectrum disorder are presence or absence of associated intellectual disability and language impairment (e.g., functional language by age 5 years is a good prognostic sign) and additional mental health problems. Epilepsy, as a comorbid diagnosis, is associated with greater intellectual disability and lower verbal ability.

Environmental. A variety of nonspecific risk factors, such as advanced parental age, low birth weight, or fetal exposure to valproate, may contribute to risk of autism spectrum disorder.

Genetic and physiological. Heritability estimates for autism spectrum disorder have ranged from 37% to higher than 90%, based on twin concordance rates. Currently, as many as 15% of cases of autism spectrum disorder appear to be associated with a known genetic mutation, with different de novo copy number variants or de novo mutations in specific genes associated with the disorder in different families. However, even when an autism spectrum disorder is associated with a known genetic mutation, it does not appear to be fully penetrant. Risk for the remainder of cases appears to be polygenic, with perhaps hundreds of genetic loci making relatively small contributions (Abdallah and Eissa Saad 2014).

COMORBIDITY

Autism spectrum disorder is frequently associated with intellectual impairment and structural language disorder (i.e., an inability to comprehend and construct sentences with proper grammar), which should be noted under the relevant specifiers when applicable. Many individuals with autism spectrum disorder have psychiatric symptoms that do not form part of the diagnostic criteria for the disorder (about 70% of individuals with autism spectrum disorder may have one comorbid mental disorder, and 40% may have two or more comorbid mental disorders). When criteria for both ADHD and autism spectrum disorder are met, both diagnoses should be given. This same principle applies to concurrent diagnoses of autism spectrum disorder and developmental coordination disorder, anxiety disorders, depressive disorders, and other comorbid diagnoses. Among individuals who are nonverbal or have language deficits, observable signs such as changes in sleep or eating and increases in challenging behaviour should trigger an evaluation for anxiety or depression. Specific learning difficulties (literacy and numeracy) are common, as is developmental coordination disorder.

Medical conditions commonly associated with autism spectrum disorder should be noted under the "associated with a known medical/genetic or environmental/acquired condition" specifier. Such medical conditions include epilepsy, sleep problems, and constipation. Avoidant-restrictive food intake disorder is a fairly frequent presenting feature of autism spectrum disorder, and extreme

and narrow food preferences may persist (Abdallah, Eissa Saad 2014).

CONCLUSION

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that is characterized by sustained social impairments in reciprocal social communication and interactions; and repetitive behaviours, interests, or activities. These essential markers of autism spectrum disorder present in early childhood and limit everyday functioning.

The Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5) is the 2013 update to the American Psychiatric Association's classification and diagnostic tool. DSM is commonly used to guide the diagnosis of autism (American Psychiatric Association (APA 2000). The fifth edition of the manual, DSM-5, has substantially revised the criteria for the disorder (APA 2013). The new criteria will include individuals previously diagnosed with autistic disorder (AD), Asperger disorder, childhood disintegrative disorder, and pervasive developmental disorder-not otherwise specified (PDD-NOS) into a new diagnosis called autism spectrum disorder (ASD).

In DSM-5, sensory abnormalities are newly included as a core, diagnostic symptom of the restricted, repetitive patterns of behaviour (RRB) domain. This change is based on empirical research finding that this symptom has sensitivity and specificity as a diagnostic criterion for ASD, and factor analytic evidence that places it in the RRB symptom dimension. This improves the relevance of the criteria to younger children with ASD, because sensory issues are common concerns in this population.

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