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REVIEW

Autism spectrum disorders

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Abstract The revision of the diagnostic criteria for ASD has been widely anticipated and is expected to be an important contribution to the refinement of the definition of ASD. In the upcoming DSM-5, several changes have been made compared to the previous diagnostic criteria. They include no emphasis on language delay and age of onset except that ASD is defined as a neurodevelopmental disorder with symptoms in early childhood although the disorder may first be diagnosed later in life. The three areas of impairments in ASD are reduced to two areas, namely a social-communication domain and a behavioral domain including fixated interests and repetitive behaviors. In addition, the clinical presentation of ASD in the individual is described in more detail in terms of clinical specifiers. In addition to reporting these changes in the classification, the major international guidelines are introduced and a brief description of good clinical practice of assessment and the overall principles of intervention is provided.

Keywords Autism spectrum disorder · Autism · Diagnosis · DSM · ICD

Introduction

When the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) will be published in May 2013, a period of 14 years will have passed since the previous edition of the various diagnostic criteria of mental disorders. It has been custom to redefine the criteria with certain time intervals because a redefinition of the diagnostic criteria of mental disorders has been shown to be necessary to adapt common practice of diagnosing to the changes in society with respect to cultural and sociologic factors.

Several refinements in the new diagnostic criteria have been suggested based on the limitations of the previous criteria, e.g. related to validity of diagnoses. Due to the large numbers of diagnoses in the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV) [1] and the International Classification of Diseases, tenth edition (ICD-10) [2], the validity is assumed to be insufficient. Another disadvantage is the large number of "not otherwise specified" (NOS) diagnoses. In addition, the changing practice in recent years with an increase of diagnosing various comorbidities contributes to queries into the validity of diagnoses.

Autism spectrum disorders in DSM-5

Autism spectrum disorders (ASD) is a term that is being gradually more and more used to describe the diagnoses which in ICD-10 and DSM-IV were included under the term of pervasive developmental disorders and in DSM-5 ASD will include the diagnoses that were previously classified as autistic disorder, Asperger's disorder, childhood disintegrative disorder, and pervasive neurodevelopmental disorder not otherwise specified. As the name indicates, ASDs consist of a group of developmental disorders with symptoms that are seen on a continuum ranging from mild to severe expression. ASD must be present from infancy or early childhood, but in the new DSM-5 the age criteria for the diagnosis will not be further specified and it

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is recognized that the deficits of ASD may not be detected until later in life because of minimal social demands and because the deficits may be compensated for by parents or caregivers earlier in life. The proposed revision of ASD in the DSM-5 is shown in Table 1.

One of the goals when introducing the new diagnostic criteria is that validity of the criteria for ASD should be the same meaning that no more cases should be diagnosed with the new criteria than with the old ones. However, one of the limitations of the new diagnoses introduced in DSM-5 is that they are only sparsely validated and especially the reliability of different diagnoses across countries and cultural backgrounds and between clinicians is not fully investigated. This statement is partly supported by several studies showing that the validity of the different ASD diagnoses is low, whereas the development of ASD has been shown to be reliably distinguished from typical development. A study by Lord et al. [3] has shown that the definition of autistic disorder and Asperger's disorder may differ between sites of assessment for ASD. In earlier studies, autistic disorder, Asperger's disorder, and PDD-NOS could not be distinguished from each other [4, 5] which is supported by a recent review [6] concluding that there is an overlap between the criteria for Asperger's syndrome and autistic disorder, and that there is an inconsistent distinction between the different ASDs. Existing studies used different diagnostic criteria, both from the ICD and DSM system and even other modified clinical criteria. The distinction between the different ASDs is suggested to be dependent on the severity of the disorder, the language level, and the presence of learning disability/mental retardation.

Onset of ASD

ASD is a neurodevelopmental disorder with onset early in life. The onset of ASD has been studied intensively [7–9] and it is generally accepted that the time of an ASD diagnosis may be much later than the time of onset of the disorder. In addition, the time at which the parents acknowledge that their child exhibits early signs of abnormal development or symptoms of ASD until a diagnosis is applied to the child may be of long duration. On the other hand, detecting the specific early signs of ASD has been found to be hard and no specific signs have yet been identified [10]. This discussion has had the impact on the new DSM-criteria that the time of onset of symptoms before the age of 3 years has been removed from the diagnostic criteria because the time of recognition of ASD symptoms is only arbitrary.

In addition, at the other end of the age span the deletion of the age criteria is more in line with the fact that some individuals with ASD are first diagnosed as adolescents or adults. The diagnosis of ASD later than early childhood may be explained by the recognition of symptoms only when the requirements for social abilities increase with age.

Table 1 Proposed revision of the Diagnostic Statistical Manual (DSM) for Autism Spectrum Disorders (DSM-5)

Autism Spectrum Disorder
Must meet criteria A, B, C, and D:
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.

Language delay

The presence of language delay for applying a diagnosis of autistic disorder has been suggested not to be included among the new criteria for ASD because it is well established that language delay is not specific to ASD as it is present both among children with ASD and children without ASD. Furthermore, several children with ASD develop fluent speech as they grow up even though they had language delay when they were younger.

The development of language is, however, crucial with respect to outcome because severe language delay in early life appears to be a predictor for poor outcome. Furthermore, the choice of treatment may be influenced by the language level of the person with ASD and the clinical presentation of ASD is dependent on the language level of the individual with ASD [11].

Language disorder versus Asperger's disorder

One of the other groups of neurodevelopmental disorders in the DSM-5 consists of communication disorders including a subgroup of Social Communication Disorders. It has been debated whether some persons with Asperger's disorder will be misclassified in this diagnostic group instead of the ASD group meaning that not all individuals with Asperger's disorder according to DSM-IV will be registered in the group of ASD according to DSM-5. This proposal again is not in accordance with one of the goals of the new diagnostic criteria saying that all individuals with ASD should remain in this diagnostic group.

Another important consequence of the DSM-5 diagnostic criteria of communication disorders is that when a person with e.g. Asperger's disorder is suffering from other deficits of communication like a language or speech disorder, he/she besides ASD should be diagnosed with this communication disorder. One exception is the Social Communication Disorder which cannot be applied when the person is suffering from ASD as it is required in the criteria of DSM-5 that ASD is ruled out to be diagnosed with a Social Communication Disorder.

Domains

Based on the population-based studies and twin studies of ASD [12, 13], it has been shown that the impairment in social interaction and communication is part of the same domain. This has resulted in the three areas of impairment being reduced to two areas, namely a social-communication domain and a behavioral domain including fixated interests and repetitive behaviors. Furthermore, the two new domains have been more specifically described for different developmental age groups and the individual's clinical presentation is more extensively described in terms of clinical specifiers like severity of symptoms, and level of language development. In addition, it is suggested that associated features like the presence of intellectual disability and co-morbidity are described.

Disintegrative disorder

The validity of disintegrative disorder has been debated in recent years, and it is suggested to include this disorder into ASD. In this debate, the focus has been on the problems with defining developmental regression and the time of onset of regression in addition to the problem of defining whether delayed development was present before regression. Furthermore, it has been discussed whether changes or symptoms appear gradually or suddenly. Therefore, the DSM-5 suggests removing the category of childhood disintegrative disorder. This decision is in contrast to what is currently suggested in the criteria of the ICD-11 where attempts have been made to define childhood disintegrative disorder.

Concluding comment

A major attempt of the DSM-5 is to make the diagnostic criteria of ASD more clear which may also increase the validity of included diagnoses if they are well defined seen from a clinical and evidence-based point of view. Certainly, the diagnostic criteria of ASD in the DSM-5 will become simpler in the areas of communication and social difficulties because these areas are difficult to disentangle from each other in daily life as well as in a clinical setting during assessment. In addition, the deletion of the age criterion for language delay will serve most likely as a refinement of the diagnostic criteria and it is assumed not to increase the number of individuals included in the group of ASD. Most probably, these individuals were included in the unspecific diagnostic groups of ASD in the previous diagnostic criteria.

One of the most debated changes in the revision of DSM-5 is the removal of Asperger's syndrome. This suggestion is, however, based on the evidence from studies showing that no clear difference could be found between Asperger's syndrome and e.g. autistic disorder with respect to different measures of outcome. However, from a clinical point of view it is also expected to be easier in DSM-5 to apply a diagnosis of ASD especially in those cases where the collection of data on the early developmental history is difficult due to poor parental memory about these issues.

Assessment guidelines

The need for clinical guidelines is manifold. One of the purposes is to standardise the diagnostic process and

clinical management as these processes may be very different between countries and even within countries. Moreover, different health services may be involved in the assessment depending on the age of the individual suspected to be diagnosed with ASD. As a consequence of these differences, the parents through parental autistic societies have advocated for guidelines to be developed.

Various national guidelines have been performed in the USA, the UK, Scotland, New Zealand, but in clinical settings, guidelines have also been performed in many countries like e.g. the Nordic countries. Some of the recent and most well-known clinical guidelines are shown in Table 2.

The guideline by the American Academy of Pediatrics [14] describes evaluations to be taken by pediatricians at very early steps like the first preventive care visit. Interestingly, the guideline states that ideally the assessment including the diagnosis should be made by a team of child specialists with expertise in ASDs. However, due to a lack of availability of specialist teams in each region or due to long waiting lists, in some communities the assessment might be made by pediatric subspecialists or child psychologists. A major goal of this guideline is the surveillance and identification of autism spectrum disorders at an early age. However, the guideline also describes the comprehensive assessment needed to apply a diagnosis of autism and it provides a guideline to management of children with ASD [15].

The NAP-C [16] is a national plan which was developed in the UK after widespread variation in clinical diagnostic and management services for children with autism was found in 2001. A multidisciplinary, multi-agency approach to assessment and diagnosis was recommended in the NAP-C and well-established and well-used diagnostic tests and assessments were suggested to be included in the assessment. In a study [17] assessing the impact of the NAP-C on clinical practice, it was concluded overall that an improvement in diagnostic services for children with ASD was seen but differences between diagnostic sites still existed.

The Scottish guideline by SIGN [18] focuses on the clinical assessment and management/treatment including outcome and the target population is children and young people with ASD. Like the NICE guideline, it systematically evaluates evidence for the methods used. On the contrary, the NICE guideline [19] does not include population screening and surveillance although it lists the signs and symptoms that should prompt professionals to make a referral for specialist assessment. The NICE guideline focuses on the different components of diagnostic assessment as described below and a comprehensive review of evidence for clinical practice of assessment and management is provided. One of the areas specifically described in this guideline is reassessment and review of diagnosis.

Like the other guidelines, the New Zealand Autism Spectrum Disorders guideline [20] covers both diagnosis and initial assessment of ASD in addition to support, treatment, and management of ASD. In contrast to the other guidelines, it includes a section about professional learning and development.

Diagnostic assessment after referral

In general, it is recommended that the diagnostic evaluation of ASD includes information from different sources dependent on the age of the person. Mandatory for the evaluation is an interview with the caregivers to obtain information about the early history of development to verify whether the ASD symptoms had been present from infancy or early childhood. The interview with the caregivers also aims to collect information about the medical history including information about pregnancy and birth in addition to associated medical problems/symptoms and disorders.

Table 2 Examples of clinical guidelines for ASD

Author	Name	Publication year
National Autistic Society: Ann Le Couteur, Chair, Core Working Group	National Autism Plan for Children (NAPC)	2003
Scottish Intercollegiate Guidelines Network (SIGN)	Assessment, diagnosis and clinical interventions for children and young people with autism spectrum disorders: a national clinical guideline	2007
The American Academy of Pediatrics	a. Identification and evaluation of children with autism spectrum disorders	2007
	b. Management of children with autism spectrum disorders	2007
Ministries of Health and Education	New Zealand: autism spectrum disorders guideline	2008
Guideline development group under National Collaborating Centre for Women's and Children's Health	National Institute for Health and Clinical Excellence (NICE) guideline	2011

Another mandatory source of information is the observation of the individual with ASD both in the living environment and as part of a psychiatric evaluation in the assessment centre. Structured observations of the individual suspected to suffer from ASD including the ADOS (Autism Diagnostic Observation Schedule) [21] are widely used throughout at least Europe and North America. In addition, the diagnostic evaluation has to include a detailed medical and neurological examination aiming at ruling out any associated medical disorder like tuberous sclerosis, the Fragile X syndrome, other genetic disorders, and any sensory deficits. The third major element of the diagnostic evaluation is the psychological evaluation of the cognitive level of the individual with ASD. Psychological testing may also serve the purpose of searching for specific neuropsychological deficits. Finally, it is also important to search for any co-morbid psychiatric disorders as they may need to be treated to maintain the psychological well-being of the person.

Intervention

Overall, the goal of intervention is to optimise the functional independence of the individual with ASD by minimising the core ASD features. Another important goal of intervention is to facilitate development and learning including promoting the social skills of the individual. Another goal of the intervention is to reduce restricted interests and stereotypic behavior and eliminate maladaptive behavior. No medical treatment, however, can eliminate ASD symptoms but medical treatment may be necessary to reduce symptoms from comorbid psychiatric disorders.

In principle, the developmental level of the person with ASD should be determined to adapt the demands from the people around the person with ASD and to adjust the environment to potential sensory problems of the ASD person. In addition, the quality of life and well-being of the person with ASD is dependent on support from the family and for that reason education and support of ASD families need to have priority in the plan for intervention.

Concluding comment

The guidelines are in general very thorough but the guidelines where evidence-based practice is systematically presented are recommendable. The NICE guideline is the most recent and is therefore recommended as a work of reference for assessment and management of autism. The guideline from New Zealand, however, is also very extensive and very usable due to its clear presentation of evidence. More locally based guidelines are most likely more useful for the specific country because they take into account the local conditions under which assessment is performed.

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