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Autism Spectrum Disorder and the Diagnostic and Statistical Manual of Mental Disorders—Fifth Edition (DSM-5): The Experience of 10 Years

Annio Posar[1,2], Paola Visconti[1]

[1]IRCCS Istituto delle Scienze Neurologiche di Bologna, UOSI Disturbi dello Spettro Autistico, Bologna, Italy
[2]Department of Biomedical and Neuromotor Sciences, University of Bologna, Bologna, Italy

Autism spectrum disorder (ASD) is a usually lifelong condition characterized by impairments in social communication skills as well as restricted and repetitive patterns of behavior, the diagnosis of which is still based on clinical criteria.[1,2] In this paper, we report and discuss the lights and the shadows of the Diagnostic and Statistical Manual of Mental Disorders—Fifth Edition (DSM-5) as regards the diagnosis of ASD.[1]

In 2013, DSM-5 introduced a number of important changes concerning ASD, some of which have obtained a broad consensus in the scientific world.[1] Among them, we can mention having unified the criteria relating to the qualitative impairments in social interaction and in communication which were separate in the previous version of the manual (DSM-IV).[3] This change has been implemented since it was rightly considered that alterations of social interaction inevitably lead to alterations of communication. Furthermore, in the DSM-5 the presence of sensory abnormalities was correctly valued among the diagnostic criteria for ASD in the context of "restricted, repetitive patterns of behavior." Finally, DSM-5 included ASD in the chapter on neurodevelopmental disorders along with other conditions such as intellectual disability, attention-deficit/hyperactivity disorder (ADHD), and communication disorders, rightly emphasizing the aspects that tend to unify these conditions, including the frequent comorbidity existing among them.[1]

But 10 years after the release of the DSM-5, clinical experience also suggests the usefulness of a number of improvements to the section of the DSM-5 dedicated to ASD. First of all, the clinical description obtained from the DSM-5 diagnostic criteria does not fully illustrate the extreme heterogeneity of this condition, which appears to be profoundly influenced by frequent neurological and psychiatric comorbidities.[4] Consequently, the same ASD diagnostic label is given to quite different clinical conditions, leading the spectrum of autism to include cases ranging, for example, from severe epileptic encephalopathies associated with autistic behavior and profound intellectual disability to high or very high functioning cases.[5] Between these 2 extremes, there is a great variety of possible situations. Diagnostic and Statistical Manual of Mental Disorders—Fifth Edition completely eliminated the subdivision of 5 diagnostic categories of pervasive developmental disorders (PDDs) included in the DSM-IV, i.e., Asperger's disorder, autistic disorder (the classic Kannerian autism), Rett's disorder, childhood disintegrative disorder, and pervasive developmental disorder not otherwise specified.[1,3] First, in DSM-5 Rett's disorder was excluded as the gene involved in its etiopathogenesis (i.e. *MECP2* in most cases) was found.[6] In this regard, are we sure that having found a definite etiology justifies the exclusion of this disorder from the ASDs? If it has to be so, then why not exclude also all other cases of ASD with a known etiology? It is recognized, in fact, that in a non-negligible minority of cases, ASD is caused by a well-defined genetic condition (e.g. mutations of the *SHANK3* gene). Yet these cases, pertaining to the so-called syndromic autism,[7] have not been excluded from the diagnosis of ASD according to DSM-5, and in the future, they might be described by combining the diagnosis of ASD with a known genetic etiology (e.g. *SHANK3* mutation-induced ASD). Furthermore, it should be

Corresponding author:
Annio Posar
✉ annio.posar@unibo.it
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emphasized that not all cases of Rett's disorder are related to the *MECP2* gene: some of them are caused by mutations in the *CDKL5* gene, others by mutations in the *FOXG1* gene, while for some other rare cases, the etiology is still unknown.[6] In DSM-5, the other 4 categories of PDDs included in the DSM-IV[3] were all fused into ASD, as the DSM-5 authors deemed that there were not enough elements to distinguish them and that ASD is characterized by a continuum of symptoms. According to DSM-5, there is only one possible diagnosis for an individual with autistic behavior, which is precisely ASD. However, 3 levels of severity were instituted in DSM-5: level 1 ("requiring support"), level 2 ("requiring substantial support"), and level 3 ("requiring very substantial support"), each accompanied by a description of the relevant clinical features.[1] But the clinical characterization based on this description is too poor, mainly due to the great heterogeneity of the possible associated neurological (e.g., epilepsy and insomnia), psychiatric (e.g., intellectual disability and ADHD), and even medical (e.g., gastrointestinal disorders) conditions.[4] The real impairment of the adaptive skills of individuals with ASD cannot be described by the simple attribution to the clinical picture of 1 of the 3 levels of severity[1] and the situation is made even more complicated by the possible comorbidity with an intellectual disability. In this case, according to the DSM-5, how can the severity of the intellectual disability be classified? For this purpose, the DSM-5 has removed the quantification of the intelligence quotient (system that was used in DSM-IV)[3] and has offered a series of qualitative criteria relating to the ability of the individual to adapt to the environment, on the basis of which the intellectual disability is subdivided into mild, moderate, severe, and profound.[1] These criteria are very useful for the evaluation of an individual with intellectual disability and without ASD. But how can these criteria, as they are, be applied to individuals with ASD who, by definition, have significant deficits in adaptive skills? Evidently, this could be confusing, also because the possible presence of an intellectual disability represents notoriously one of the most important elements in defining the degree of impairment of the individual with ASD and also has repercussions on the long-term prognosis.[8] In an individual with ASD and intellectual disability, it is very complicated, and perhaps all in all useless to distinguish between the clinical features that are related to ASD and those that are related to intellectual disability.

In conclusion, a subdivision of individuals with ASD into more clinically homogeneous subgroups would be appropriate, setting aside the oversimplified subdivision into 3 groups according to the level of support as proposed by the DSM-5.[1] We believe this is necessary also in consideration of the impressive increase in the last few decades of the prevalence of ASD, which according to the most recent studies in the USA has reached 2.76% at the age of 8 years.[9] In our opinion, this different approach, motivated by the enormous phenotypic heterogeneity that characterizes ASD as it is conceived today according to the DSM-5, should be followed for both healthcare and research purposes.

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