

Classification of autism and related conditions: progress, challenges, and opportunities

Fred R. Volkmar MD; Brian Reichow, PhD; James McPartland, PhD



Since Kanner's classic description of the syndrome of early infantile autism in 1943, conceptions of the disorder have evolved while retaining important continuity with what Kanner viewed as the hallmarks of the condition—social impairment (autism) and difficulties in dealing with change in the nonsocial world (insistence on sameness). This paper reviews the history of this evolution and the important potential advantages and disadvantages of changes being contemplated for DSM-5. The convergence of diagnostic approach in DSM-IV and ICD-10 provided a shared system that fostered a tremendous body of research. The changes proposed in DSM-5 may impact both research comparability and service eligibility.

© 2012, LLS SAS

Dialogues Clin Neurosci. 2012;14:229-237.

Keywords: *autism; diagnosis; DSM-5*

Author affiliations: Child Study Center; Yale University School of Medicine, New Haven, Connecticut, USA

Address for correspondence: Fred R. Volkmar, Child Study Center, Yale University School of Medicine, PO Box 207900 (230 South Frontage Rd), New Haven, CT 06520-7900, USA
(e-mail: fred.volkmar@yale.edu)

Introduction

Since Kanner's classic account of the syndrome of "early infantile autism," considerable progress has been made in our understanding of this and related conditions.^{1,2} Although it is likely that cases of autism had been seen well before Kanner's report, eg, reports of so-called "feral" children, it was Kanner's report that caught professional attention.^{3,4} As noted in the remainder of this paper, over the nearly 70 years that followed Kanner's report there have been many clarifications regarding aspects of autism and approaches to diagnosis. This paper summarizes some of the historic and enduring tensions and the current controversies regarding the proposed new *Diagnostic and Statistical Manual of Mental Disorders (DSM)-5* diagnostic approach. A review of the development of diagnostic concepts, a brief review of the tensions between categorical vs dimensional diagnostic approaches, and distinctions between research and clinical utility are helpful in framing the relevant issues and setting the stage for a discussion of current issues. The development of diagnostic concepts, the role of dimensional approaches to diagnosis, and the background for the current (*DSM-IV/ICD-10*) definition of autism will be reviewed. Issues involved in the potential changes to and divergence of the official *DSM* and *ICD* definitions will then be summarized.

Approaches to diagnosis: a brief review

Both categorical and dimensional approaches to diagnosis have been utilized, although in actual clinical practice diagnostic approaches (for many reasons) tend to be

State of the art

ideographic, ie, encompassing all the complexities of the specific individual.⁵ Categorical approaches have tended to dominate in official classification schemes, but are not incompatible with dimensional ones, eg, selection of an arbitrary “cutoff point” for hypertension or intellectual disability.

Categorical approaches have become much more sophisticated in recent years—notably with the advent of the research diagnostic criteria (RDC) adopted with *DSM-III* (which first officially recognized autism in 1980).^{6,7} Categorical systems have great value for record-keeping and statistical purposes but face some intrinsic challenges, eg, the problem of setting a specific diagnostic threshold while recognizing “subthreshold” forms of conditions, dealing with co-occurring conditions (comorbidity), and addressing developmental change, as well as the enduring tension between narrow vs broader definitions. The latter reflects, in part, an intended use for research or more general clinical approaches.

The official approaches applied in *DSM-IV* and *ICD-10* exemplify this difference, with *DSM-IV* being intended for both clinical and research use, while *ICD-10* provides two different guides for these two purposes. Similarly, *ICD-10*, in general, discourages comorbidity while this is more acceptable in *DSM-IV* (see ref 4 for a discussion). The tensions between narrow vs broad definitions have important implications for service planning, as well as for research. For the latter purpose a very specific definition is often the goal while for purposes of service provision a broader diagnostic concept may be more appropriate. The latter is particularly an issue in the US where labels like autism may provide specific rights to service from schools and other services. As discussed subsequently, other issues arise given advances in science, eg, with the identification of genetic and other pathophysiological mechanisms.

Dimensional approaches to diagnosis

Dimensional approaches offer some considerable advantages, with instruments often having had extensive periods of development and well known psychometric properties, ie, of reliability. These are exemplified in the use of standard tests of intelligence, adaptive behavior, or communication. For disorders like autism where highly unusual behaviors or developmental features are sampled there can be special problems for developing and using dimensional assessments. However, a considerable

body of work now exists on their use both for purposes of screening and diagnosis.^{8,9} And now some of these approaches have been used to “crosswalk” back to categorical ones.

In autism and related conditions dimensional approaches have taken various forms. For example, instruments designed to assess normative development, eg, of intelligence, communication, motor development, and adaptive behavior are widely used.^{10,11} Such instruments provide information that can be used both to monitor progress to refine interventions and may also inform issues of diagnosis. As noted, subsequent differences in psychological profiles may mark different expressions of the autism phenotype, eg, with individuals with Asperger’s disorder exhibiting rather different profiles compared with those with classical autism.¹² One of these instruments, the Vineland Adaptive Behavior Scales, has also been used as a screening tool and had considerable utility in discriminating individuals with and without autism.¹³

Other instruments focus on behaviors or features more specific to autism, eg, specific symptoms, behavioral ratings, or historical information. These can be provided based on direct assessment of the individual, parent or teacher report, or both. Some of these instruments are designed for screening and others for diagnostic purposes.^{8,14} Challenges for instruments of this type relate, as do categorical approaches, to the balance of sensitivity and specificity, as well as much more complex problems of sampling, instrument design, and so forth. Issues of how intense, frequent, and disabling symptoms are become important as do aspects of informant bias. For some of the best of these instruments the training requirements and length of administration time may limit use in actual practice (again highlighting the tension between research and clinical use of classification systems).

Historical development of diagnostic concepts

Infantile autism

Kanner’s clinical description of children with “autistic disturbances of affective contact” has proven enduring.¹ He was a careful phenomenologist in the days before the importance of an a theoretical approach was emphasized.¹⁵ His description was also carefully grounded in available child development research, eg, he emphasized how normal infants exhibit marked interest in social

interaction from early in life. Kanner suggested that the condition he described was inborn, and that the children he had seen exhibited a curious lack of interest in the social environment combined with an increased interest in the nonsocial environment. The latter phenomenon he referred to as “insistence on sameness” or “resistance to change,” emphasizing an overengagement of the child with trivial changes in the nonsocial world. His use of Bleuler’s term “autism” was intended to describe the lack of connection to others (in contrast to Bleuler’s use of the word in describing highly idiosyncratic and self-centered thought processes). Kanner also noted marked problems in communication, with either a total absence of spoken language or with highly unusual language marked by features such as pronoun reversal, echolalia, and difficulties using social language. Unfortunately some aspects of his report misled investigators. His use of the term “autism” raised confusion with schizophrenia and, given the broad views of schizophrenia, fostered the assumption that autism was a form of schizophrenia.² Kanner also noted that the parents of his initial cases were remarkably successful, leading to the idea that autism was a phenomenon associated with social class. This led, unfortunately, to an entire school of thought focused on parental pathogenesis of autism in the 1950s. Kanner did also not initially recognize how frequently autism was associated with intellectual disability; in retrospect this is not surprising, given the importance of a fundamental social orientation for learning what is and isn’t important in the nonsocial environment, and the frequent scattered IQ profile observed in autism.

Asperger’s disorder

The inclusion of this condition, first described by Hans Asperger in 1944, was one of the sources of greatest controversy in *DSM-IV* and *ICD-10*.⁵⁻¹⁶ Although debate continues regarding the best approach for defining Asperger’s disorder, official recognition in the *DSM-IV* and *ICD-10* has resulted in a dramatic increase in research (from approximately 75 peer-reviewed publications between 1944 and 1994 to greater than 1000 in the 20 years since). In this condition, early language development seems normal but marked social difficulties (of the type seen in autism) develop, particularly with peers, and come to attention somewhat later in life than in autism. Circumscribed interests are marked, and are a source of disability. A body of work has now asso-

ciated Asperger’s disorder with a specific learning profile (that of Nonverbal Learning Disability).¹⁷ In contrast to autism, better-preserved language abilities offer an important route for intervention (note that communication is often significantly impaired and a focus of treatment). The *DSM-IV* definition of Asperger’s disorder has been rightly criticized, and the difficulties likely reflect, in part, the understandable ambivalence about including new disorders in *DSM*.^{18,19}

Pervasive Developmental Disorder-Not Otherwise Specified/atypical autism

Both the *DSM-IV* and *ICD-10* include these subthreshold Pervasive Developmental Disorder (PDD) diagnoses with very slight differences in description. These diagnoses are used when symptoms do not meet specific criteria for a PDD, but there are major social difficulties and problems in either restricted behaviors or communication of the type seen in autism. In clinical practice this diagnosis is easily, and frequently, made, perhaps reflecting, at least in part, the potential complex and multifaceted genetics of autism when it is more strictly defined; indeed these cases likely represent some aspects of a broader “autism spectrum.”²⁰ These conditions account for the largest proportion of the cases of “PDD” or “Autism Spectrum Disorder” (ASD).²¹

Childhood disintegrative disorder

This condition, sometimes termed Heller’s syndrome (after the man who first described it in 1908) or disintegrative psychosis, is characterized by a prolonged period of normal development (typically 3 or 4 years) followed by a dramatic developmental deterioration in multiple areas and development of a fairly classic autistic presentation.²² Recovery is usually limited. Although this was at first thought to be a childhood dementia, development stabilizes at a lower level but no further deterioration occurs. The main reasons for including this condition in *DSM-IV* and *ICD-10* included its unusual clinical presentation, poor outcome, and, potentially, some specific neuropathological process etiologically.²²

Rett’s disorder

Described by Rett in 1966, this is a condition essentially confined to females (males presumably die before

State of the art

birth).²³ Very early development is normal, but then deteriorates with a striking clinical pattern including some social unresponsiveness (in the preschool years), motor and respiratory problems, seizures, and profound developmental delay. Rett originally thought this might be a form of autism, and it was included in the PDD category in *DSM-IV* and *ICD-10*, although important differences between Rett's disorder and other PDDs were acknowledged.²⁴ Subsequently, a specific genetic etiology has been determined.²⁵ As a consequence, Rett's disorder is anticipated to be removed from the *DSM-5*. As similar advances in genetics make it likely that a range of conditions of childhood onset (and for that matter adult onset) will have very identifiable genetic components, taxonomies of psychiatric conditions may be significantly reduced.⁴

It should be noted that other concepts have been proposed but have not endured or, in other instances, diagnostic categories have persisted with some relationship to autism and related conditions. Mahler's concept of symbiotic psychosis²⁶ is now of only historic interest, as is her theoretical notion of a normal "autistic phase" of infant development. In contrast, Rank's notion of atypical development²⁷ prefigured, in some respects, the concept of atypical autism/PDD-NOS. Similarly the concept of schizoid disorder elaborated by Wolff²⁸ has some potential overlap with Asperger's disorder.

Evolution of autism as a diagnostic concept – from Kanner to *DSM-IV*

In the 20 to 25 years after Kanner's paper appeared, several lines of work clarified that autism was a neurobiological disorder (eg, with about 20% of cases exhibiting seizures),²⁹ was very strongly genetic with high concordance rates in identical twins,³⁰ and was distinctive from childhood schizophrenia.^{31,32} In the late 1970s, various attempts were made to provide more operational guidelines for autism^{33,34} and the condition was officially recognized for the first time in *DSM-III*.⁶ Inclusion of infantile autism as an explicitly defined category was a major accomplishment.

Unfortunately the *DSM-III* definition proved overly narrow (indeed focusing on the "infantile" form of the disorder), was "monothetic" (ie, every single feature/criterion had to be present) and thus was overly stringent. In *DSM-III* developmental change was dealt with by including a category for "residual" infantile autism.⁵ This

problem was addressed in the revision of *DSM-III* that appeared in 1987.³⁵ The *DSM-III-R* definition was polythetic, with combinations of multiple criteria in the three traditional areas of disturbance (social, communication, behavior) with highly detailed criteria (some of which included examples). This definition owed a considerable intellectual debt to Lorna Wing's work, focused on a broader spectrum concept of autism and related conditions.³⁶ A field trial was conducted but proved problematic in some respects.³⁷⁻³⁹ It appeared that the criteria provided favored overdiagnosis of autism in more cognitively impaired individuals (where high rates of stereotyped behaviors are frequent) and a relative underdiagnosis in more cognitively able groups. Another potential problem included the potential for major differences with the changes to be made in *ICD-10* then scheduled to appear at about the same time as the new *DSM-IV*. Given the concern that for autism, two competing diagnostic approaches would impact research some consideration was made for a joint effort to derive a diagnostic approach suitable for both publications.

In the diagnosis of autism spectrum disorders (ASDs), both *DSM-IV* and *ICD-10* adopt an explicit categorical approach, and although the systems differ in some respects for autism the definitions are virtually identical based on the results of a large international field trial.⁴⁰ The field trial included 21 sites with over 100 raters providing information on nearly 1000 cases who were included in the field trial if autism was being considered in the differential diagnosis. The sample exhibited a range of ages (from young children to adults), levels of functioning (from those who had severe cognitive impairments to gifted individuals), and symptom severity. Based on a series of preliminary data reanalysis it was agreed that the system developed for autism should aim to have a reasonable balance of sensitivity and specificity across the IQ and age ranges and a convergence, at least for autism and related conditions, between *DSM-IV* and *ICD-10* if at all possible. The final definition for autistic disorder (childhood autism) included 12 criteria grouped into 3 categories (social, communication-play, and restricted interests and behaviors) with a minimum requirement of a total of 6 criteria, 2 of which had to be social (highlighting the strength of social dysfunction being the best predictor of diagnosis of autism), 1 of which had to be communicative, and 1 of which needing to be behavioral with the remaining two capable of coming from any domain (over 2000

combinations of criteria can meet this threshold). Interrater reliability of individual criteria was generally good to excellent, as was agreement of PDD versus non-PDD diagnosis. The field trial also provided sufficient data for the inclusion of several disorders “new” to *DSM-IV* and/or to *ICD-10*. These conditions included Asperger’s disorder, Rett’s disorder, Childhood Disintegrative Disorder, and “subthreshold” PDD (PDD-NOS) as well as autistic disorder/childhood autism.

From *DSM-IV* to *DSM-5*

Before considering the impact of potential changes in *DSM-5*, it is important to consider the strengths and weaknesses of the *DSM-IV* approach. Nomenclature changes should be carefully and thoughtfully made based on data that has accumulated and should also include a reasonable sense of conservatism (ie, unneeded change complicates research and clinical activities). In this regard it is important to recall that unlike *ICD-10* (and presumably *ICD-11*), *DSM-5* will encompass both clinical and research use. It is also relevant that the convergence of *DSM-IV* and *ICD-10* with regard to disorders and definitions within the broader PDD category has facilitated research. Indeed, in the year before *DSM-IV* appeared there were about 350 peer-reviewed scientific publications on autism; in 2011 there were well over 2000. Having the same system in the US and the rest of the world has also fostered cross-national collaboration and the growing emergence of autism programs around the world. With an increased awareness has come more support for intervention at progressively younger ages, and what appears to be an emerging pattern of increasingly positive outcome.^{41,42} The consensus on diagnostic approach has also facilitated research in numerous areas but the advances in the genetics of autism have been particularly striking with a number of potential genes now identified.⁴³ The attempt in *DSM-IV* and *ICD-10* to have a system that balanced sensitivity and specificity across the IQ range also had an important impact in fostering increased awareness of severe social disabilities in more cognitively able individuals. With this great awareness has come increased services for this population as well as for adults.

What are some of the possible limitations? The *DSM-IV* field trial was large and international in nature and included a wide range of individuals (over age, levels of cognitive ability, and so forth). Given limitations of

funding and time it was not an epidemiological sample. Rather it was meant to be informative of both clinical and research utility in a range of cases and of countries and settings. Cases did include young children and adults, although, 20 years ago, the tremendous increase in infants with possible autism could not be foreseen.⁴⁴ Work since *DSM-IV* appeared suggests that before age 3 the diagnosis is less stable than after this age.⁴⁵ Often children who go on to have autism by age 3 have the social and communication features of the disorder before that time, but the “restricted interests” criteria are slower to develop, in their most robust form being preceded by odd but not diagnostic sensory interests.⁴⁴ For most younger children with a question of autism, the issue is typically a move from autistic disorder to PDD-NOS or vice versa. For Asperger’s disorder, a number of concerns have been raised about the stringency (or lack thereof) of the *DSM-IV* diagnostic criteria.^{46,47} Due to dissatisfaction with the original description, the entire written narrative was replaced in *DSM-IV-TR*, but no changes were made to the wording of actual diagnostic criteria.⁴⁸ Several different approaches to the diagnosis of this condition are in common use, with more stringent criteria sets more likely to yield differences in neuropsychological profiles, family history, and comorbid psychiatric conditions in probands.⁴⁹⁻⁵¹ For Childhood Disintegrative Disorder, concerns have centered around issues such as the reliability of parental report of regression and the issue of whether or not the condition is sufficiently distinctive to merit inclusion in its own right (rather than as part of the a broader autism “spectrum”).⁵²⁻⁵⁵ Part of the source of this disagreement relates to the nature of methods used to assess regression. In a study from our center about 20% of a large sample of parents with children with autism reported regression, but only a small fraction of these could clearly be shown to have had such a regression.⁵⁶ Somewhat surprisingly for Rett’s disorder the discovery of an etiological gene has raised another problem—should single-gene disorders be considered “psychiatric”? For the broader PDD-NOS group the increased awareness of the many different genes potentially contributing to autism has increased interest in seeing PDD-NOS as one part of the broader autism continuum or spectrum (the broader autism phenotype).^{43,57} Unfortunately this *DSM-IV* subgroup has been the least well studied, and is undoubtedly the most heterogeneous.²⁰

State of the art

The fifth edition of the *DSM* is scheduled for publication in May 2013. The diagnostic criteria and taxonomic structure of the Pervasive Developmental Disorders are expected to change in several regards.⁵⁸ First, the current diagnostic subcategories are replaced by a single broad category of ASD would replace the term PDD. Second, the current three symptom domains (ie, social, communication, and atypical behaviors) would be reduced to two. Currently distinct domains representing social interaction and communicative behavior would be remapped into a single domain (Social/Communicative Deficits). A second domain, “restricted repetitive behaviors” (RRB), would incorporate sensory symptoms (ie, over- or under-responsiveness to sensory stimuli or atypical interest in sensory information). Third, the diagnostic threshold would require endorsement of all symptom clusters in the Social/Communicative domain, instead of a longer list of individual symptoms from which an individual must meet a subset. Fourth, to meet criteria for an ASD, two of four symptom clusters in the RRB domain must be endorsed, an increase from the current possibility of meeting criteria for PDD-NOS in the absence of symptoms in the RRB domain. Fifth, the revised diagnostic system adds a universal onset criterion (ie, symptoms present in “early childhood” though they may not “become fully manifest until social demands exceed limited capacities”) that was previously included only for Autistic Disorder.

In addition to these changes to the ASDs, a distinct, novel diagnosis, Social Communication Disorder (SCD), is proposed for inclusion in *DSM-5*.⁵⁸ This disorder, resembling current PDD-NOS without RBBS, would rule out individuals meeting criteria for ASD. It would be defined by pragmatic difficulties and marked problems in the use of verbal and nonverbal communication in naturalistic social contexts. For a diagnosis of SCD, these difficulties would have to impair interpersonal relationships and social comprehension, and would not be explained by more basic language difficulties (ie, deficits in sentence structure, grammar, or general cognitive ability). Deficits in social communication would also need to be evaluated as significantly negatively influencing communication, social involvement, academic achievement, or occupational performance.

Because these proposed changes alter the symptom profile required to meet diagnostic threshold for an ASD, it is possible that the population of individuals meeting criteria for ASD could change according to the new crite-

ria. Several studies have examined this possibility by contrasting *DSM-IV-TR* criteria and proposed *DSM-5* criteria in clinic and research samples. Our own research group re-examined the large data set collected as part of the *DSM-IV* field trial.⁵⁹ We evaluated sensitivity and specificity by creating an algorithm mapping symptom checklists collected during the field trial onto proposed *DSM-5* diagnostic criteria. The algorithm suggested that 60.6% of individuals clinically diagnosed with an ASD met revised diagnostic criteria; 94.9% of individuals without a clinical diagnosis were accurately excluded from the spectrum. Sensitivity varied by diagnostic subgroup such that individuals with milder forms of autism (Asperger’s Disorder = .25; PDD-NOS = .28) were less likely to meet criteria than individuals with classic autism (Autistic Disorder = .76). Individuals with cognitive impairment (IQ <70 = .70) were more likely to meet criteria than individuals with normative intellectual abilities (IQ ≥70 = .46). Similar results were obtained by Mattila and colleagues in a smaller study that relied on a previous version of the proposed criteria.⁶⁰ This study indicated that, overall, 46% of individuals with ASD met the *DSM-5* diagnostic threshold. Smaller numbers of individuals with average IQ (36%) or Asperger’s Syndrome (0%) met revised diagnostic criteria. Worley and Matson demonstrated that individuals meeting proposed *DSM-5* criteria tended to have more severe impairments than individuals meeting *DSM-IV-TR* criteria,⁶¹ a pattern replicated by Matson et al, who also found⁶² that 36.5% of individuals in a sample of developmentally disabled adults with ASD failed to meet proposed criteria, and also found⁶³ that 47.8% of toddlers meeting *DSM-IV-TR* ASD criteria did not meet *DSM-5* criteria. A large study using a national registry found that up to 12% of individuals with ASD might fail to meet *DSM-5* criteria, an effect most pronounced among females.⁶³ The prevailing trend among published studies to date is that revised criteria offer greater specificity (ie, decrease in false-positive diagnoses) but reduced sensitivity (ie, increased failure to detect true positive diagnoses). These studies all have notable limitations, including reliance on older datasets, use of outdated versions of proposed *DSM-5* criteria, or exclusive reliance on clinician observation or parent report. Most importantly, none of these studies compared diagnostic rubrics in a prospective manner, concurrently evaluating children on both criteria sets using the actual *DSM-5* criteria set (in lieu of an algorithm approximating the concepts but

employing distinct language); only with this information can any true change in prevalence associated with alteration in diagnostic rubric be estimated.

Summary

Tremendous advances in autism research and clinical practice have occurred in the nearly 70 years following Kanner's seminal publication. During this time the concept of autism, more strictly defined, has expanded to include a "spectrum" of conditions, all marked by problems in social interaction. Advances in our understanding of the impact of autism on learning and children's development have been translated into innovative and increasingly effective intervention strategies. As a group, even in more strictly defined cases, outcome appears to be substantially improving, with more adults able to achieve lives of independence and self-sufficiency; however, these individuals often continue to need support. Ongoing research continues to inform understanding, including prospective and longitudinal studies elucidating development from birth forward.

These factors must be considered as we consider a novel approach to diagnosis in *DSM-5*. Elements of the approach advocated in *DSM-5*, such as the name change to Autism Spectrum Disorder, the inclusion of a metric of impairment, and the remapping of three to two symptom domains, are important changes that will improve correspondence between *DSM* criteria and current research. On the other hand, the rationale for moving to two symptom domains (justified apparently by factor analytic work) raises some practical issues as it gives substantially less flexibility to clinicians working with the system. In the *DSM-IV* field trial data a series of factor

analyses were conducted and, depending on what constraints were used, resulted in two, three, and five factor solutions. Another problem, to some extent throughout *DSM-5*, is the great reliance on dimensional measures developed for diagnosis (which often started with *DSM-IV* criteria). While these instruments often have wonderful research behind them, they are used in research contexts and often require substantive, sometimes very substantive, training. It remains unclear how well this approach will fare in a "dual use" manual—ie, where clinicians with no previous experience are expected to use the items/criteria with little or no training. At the time of this writing (June, 2012) detailed research on the *DSM-5* field trials had yet to appear but other studies, using a range of methods focused on the proposed *DSM-5* criteria suggest that the new system could also result in significant changes in diagnostic practice, reducing the proportion of high-functioning individuals who meet *DSM* criteria and paradoxically rendering "autism spectrum disorder" similar to "Kanner's autism." Although extensive empirical work on the justification has yet to appear the rationale for these changes remains to be elaborated. Papers on this issue are continuing to appear on aspects of *DSM-5* in general⁶⁴ as well as autism in particular.⁵⁹⁻⁶⁷

One of the likely byproducts of the proposed changes in *DSM-5* is a lack of convergence with *ICD-11*. Over the time since *DSM-IV* and *ICD-10* appeared, the convergence of diagnostic approaches has stimulated a tremendous amount of research. A result of the proposed changes, at least as they are presently constituted in *DSM-5*, could mean that eventually three different diagnostic methods will be in frequent use—the current one (*DSM-IV/ICD-10*), the new *DSM-5*, and eventually *ICD-11*. □

REFERENCES

1. Kanner L. Autistic disturbances of affective contact. *Nervous Child*. 1943;2:217-250.
2. Volkmar FR, State M, Klin A. Autism and autism spectrum disorders: diagnostic issues for the coming decade. *J Child Psychol Psychiatry*. 2009;50:108-115.
3. Wolff S. The history of autism. *Eur Child Adolesc Psychiatry*. 2004;13:201-208.
4. Rutter M. Research review: child psychiatric diagnosis and classification: concepts, findings, challenges and potential. *J Child Psychol Psychiatry Allied Discipl*. 2011;52:647-660.
5. Volkmar FR, Klin A. Issues in the classification of autism and related conditions. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005:5-41.
6. American Psychiatric Association. *Diagnostic and Statistical Manual of Mental Disorders*. 3rd ed. Washington, DC: American Psychiatric Association; 1980.
7. Spitzer RL, Endicott JE, Robbins E. Research Diagnostic Criteria. *Arch Gen Psychiatry*. 1978;35:773-782.
8. Coonrod EE, Stone WL. Screening for autism in young children. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005.
9. Lord C, Corsello C. Diagnostic instruments in autism spectrum disorders. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005.
10. Klin A, Saulnier C, Tsatsanakis K, Volkmar FR. Clinical evaluation in autism spectrum disorders: psychological assessment within a transdisciplinary framework. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005:772-798.

State of the art

Clasificación del autismo y patologías relacionadas: progresos, desafíos y oportunidades

Desde la clásica descripción de Kanner en 1943 sobre el síndrome de autismo infantil precoz, las distintas concepciones del trastorno han evolucionado manteniendo una importante continuidad con lo que él visualizó como lo central de la patología: deterioro social (autismo) y dificultades para enfrentar los cambios en el mundo no social (apego a la rutina). Este artículo revisa la historia de esta evolución y las importantes ventajas y desventajas potenciales de los cambios que están siendo contemplados para el DSM-5. La convergencia de la aproximación diagnóstica del DSM-IV y la CIE-10 proporcionó un sistema compartido que fomentó gran investigación. Los cambios propuestos en el DSM-5 pueden impactar tanto en la comparación de las investigaciones como en la disponibilidad de asistencia.

Classification de l'autisme et des pathologies qui y sont liées : progrès, défis et perspectives

Depuis la description classique de Kanner du syndrome de l'autisme infantile précoce en 1943, les conceptions sur le trouble ont évolué tout en maintenant une importante continuité avec ce que Kanner considérait comme les caractéristiques de la maladie, un handicap social (autisme) et des difficultés à s'adapter au changement dans le monde non social (attachement à la routine). Cet article analyse l'histoire de cette évolution et les avantages et inconvénients potentiels importants des changements à envisager pour le DSM-V. La convergence de l'approche diagnostique dans le DSM-IV et l'ICD-10 a fourni un système partagé qui a permis d'élaborer un corpus de recherche considérable. Les changements proposés dans le DSM-V peuvent influencer à la fois sur la comparabilité de la recherche et la disponibilité des services.

11. Tager-Flusberg H, Paul HR, Lord C. Language and communication in autism. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005:335-364.

12. Klin A, Volkmar FR, Sparrow SS, Cicchetti DV, Rourke BP. Validity and neuropsychological characterization of Asperger syndrome: Convergence with nonverbal learning disabilities syndrome. *J Child Psychol Psychiatry Allied Discip.* 1995;36:1127-1140.

13. Volkmar FR, Sparrow SS, Cicchetti DV, Paul R, Cohen DJ. Social deficits in autism: an operational approach using the Vineland Adaptive Behavior Scales. *J Am Acad Child Adolesc Psychiatry.* 1987;26:156-161.

14. Lord C, Corsello C. Diagnostic Instruments in autistic spectrum disorders. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Vol 2: Assessment, interventions, and policy. Hoboken, NJ: Wiley; 2005:730-771.

15. Rutter M. Autism: its recognition, early diagnosis, and service implications. *J Dev Behav Pediatrics.* 2006;27(suppl):S54-S58.

16. Asperger H. Die "autistischen Psychopathen" im Kindersalter. *Archiv für psychiatrie und Nervenkrankheiten.* 1944;117:76-136.

17. Rourke, BP, Tsatsanis KD. Nonverbal learning disabilities and Asperger syndrome. In: Klin A, Volkmar FR, eds. *Asperger Syndrome*. New York, NY: The Guilford Press; 2000:231-253.

18. Miller JN, Ozonoff S. Did Asperger's cases have Asperger disorder? A research note. *J Child Psychol Psychiatry Allied Discip.* 1997;38:247-251.

19. Woodbury-Smith M, Klin A, Volkmar F. Asperger's Syndrome: a comparison of clinical diagnoses and those made according to the ICD-10 and DSM-IV. *J Autism Dev Disord.* 2005;35:235-240.

20. Towbin, KE. Pervasive developmental disorder not otherwise specified. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005:165-200.

21. Fombonne E. Epidemiology of autistic disorder and other pervasive developmental disorders. *J Clin Psychiatry.* 2005;66 Suppl 10:3-8.

22. Volkmar FR, Koenig K, State M. Childhood disintegrative disorder. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders*. Hoboken, NJ: Wiley; 2005:70-78.

23. Rett A. Über ein eigenartiges hirntophisches Syndrom bei hyperammonie im Kindersalter. *Wein Medizinische Wochenschrift.* 1966;118:723-726.

24. Rutter M. Debate and argument: there are connections between brain and mind and it is important that Rett syndrome be classified somewhere [comment]. *J Child Psychol Psychiatry Allied Discip.* 1994;35:379-381.

25. Zoghbi H. Genetic aspects of Rett syndrome. *J Child Neurology.* 1988;3(suppl):S76-S78.

26. Mahler MS. On early infantile psychosis. The symbiotic and autistic syndromes. *J Am Acad Child Psychiatry.* 1965;4:554-568.

27. Rank B. Adaptation of the psychoanalytic technique for the treatment of young children with atypical development. *Am J Orthopsychiatry.* 1949;19:130-139.

28. Wolff S. Schizoid disorders of childhood and adolescence. In: Last CG, Hersen M, eds. *Handbook of Child Psychiatric Diagnosis*. (Wiley series on personality processes) Oxford, UK: Wiley & Sons; 1989:209-232.

29. Volkmar FR, Nelson DS. Seizure disorders in autism. *J Am Acad Child Adolesc Psychiatry.* 1990;29:127-9.

30. Folstein SE. Genetic aspects of infantile autism. *Ann Rev Med.* 1985;36:415-419.

31. Rutter M. Childhood schizophrenia reconsidered. *J Autism Child Schizophrenia.* 1972;2:315-337.

32. Kolvin I. Studies in the childhood psychoses. I. Diagnostic criteria and classification. *Br J Psychiatry.* 1971;118:381-384.

33. Ritvo ER, Freeman BJ. Current research on the syndrome of autism: introduction. National Society for Autistic Children definition of the syndrome of autism. *J Pediatr Psychol.* 1977;1977;2:142-145.

34. Rutter M. Diagnosis and definition of childhood autism. *J Autism Child Schizophrenia.* 1978;8:139-161.

35. American Psychiatric Association. *Diagnostic and Statistical Manual of Mental Disorders*. 3rd ed, revised. Washington, DC: American Psychiatric Association; 1987.

36. Wing L. Language, social, and cognitive impairments in autism and severe mental retardation. *J Autism Dev Disord.* 1981;11:31-44.

37. Spitzer RL, Siegel B. The DSM-III-R field trial of pervasive developmental disorders. *J Am Acad Child Adolesc Psychiatry.* 1990;29:855-862.

38. Volkmar, FR, Cicchetti DV, Bregman D, Cohen DJ. Three diagnostic systems for autism: DSM-III, DSM-III-R, and ICD-10. *J Autism Dev Disord.* 1992;22:483-492.
39. Hertzog ME, Snow ME, New E, Shapiro T. DSM-III and DSM-III-R diagnosis of autism and pervasive developmental disorder in nursery school children. *J Am Acad Child Adolesc Psychiatry.* 1990;29:123-126.
40. Volkmar FR, Klin A, Siegel B, et al. Field trial for autistic disorder in DSM-IV. *Am J Psychiatry.* 1994;151:1361-1367.
41. Howlin P. Outcomes in autism spectrum disorders. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders.* Hoboken, NJ: Wiley; 2005:201-222.
42. National Research Council. *Educating Young Children with Autism.* Washington, DC: National Academy Press; 2001:307.
43. State MW, Levitt P. The conundrums of understanding genetic risks for autism spectrum disorders. *Nat Neurosci.* 2011;14:1499-1506.
44. Chawarska, K, Klin A, Volkmar F, eds. *Autism Spectrum Disorders in Infants and Toddlers: Diagnosis, Assessment, and Treatment.* New York, NY: Guilford Press; 2008.
45. Lord C. Follow-up of two-year-olds referred for possible autism. *J Child Psychol Psychiatry.* 1996;36:1065-1076.
46. Woodbury-Smith M, Klin A, Volkmar F. Asperger's syndrome: a comparison of clinical diagnoses and those made. *J Autism Dev Disord.* 2005;35:235-240.
47. Miller, JN, Ozonoff S. Did Asperger's cases have Asperger's disorder: a research note. *J Child Psychol Psychiatry.* 1997;38:247-251.
48. American Psychiatric Association. *Diagnostic and Statistical Manual of Mental Disorders.* 4th ed, Text Revision. Washington, DC: American Psychiatric Association; 2000.
49. Klin A, Pauls D, Schultz R, Volkmar PR. Three diagnostic approaches to Asperger syndrome: implications for research. *J Autism Devel Disord.* 2005;35:221-234.
50. Klin, A, McPartland J, Volkmar FR. Asperger Syndrome. In: Volkmar FR, Paul R, Klin A, Cohen DJ, eds. *Handbook of Autism and Pervasive Developmental Disorders.* Hoboken, NJ: Wiley; 2005:88-125.
51. Lincoln A, Courchesne E, Allen M, et al. Neurobiology of Asperger Syndrome: Seven case studies and quantitative magnetic resonance imaging findings. In: Schopler E, Mesibov GB, Kunc LJ, eds. *Asperger Syndrome or High Functioning Autism?* New York, NY: Plenum; 1998:145-166.
52. Hansen, RL, Ozonoff S, Krakiowak P, et al. Regression in autism: prevalence and associated factors in the CHARGE Study. *Ambul Pediatr.* 2008;8:25-31.
53. Jones, LA, Campbell JM. Clinical characteristics associated with language regression for children with autism spectrum disorders. *J Autism Dev Disord.* 2010;40:54-62.
54. Luyster R, Richter J, Risi S, et al. Early Regression in social communication in autism spectrum disorders: a CPEA Study. *Devel Neuropsychol.* 2005;27:311-336.
55. Rogers SJ. Developmental regression in autism spectrum disorders. *Ment Retard Dev Disabil Res Rev.* 2004;10:139-143.
56. Sipersein R, Volkmar F. Brief report: parental reporting of regression in children with pervasive developmental disorders. *J Autism Devel Disord.* 2004;34:731-734.
57. Piven J. The broad autism phenotype: a complementary strategy for molecular genetic studies of autism. *Am J Med Genet.* 2001;105:34-35.
58. American Psychiatric Association. Proposed draft revisions to DSM disorders and criteria. Arlington, VA: American Psychiatric Association; 2012; [cited 2012, April 12]; Available at: <http://www.dsm5.org/Pages/Default.aspx>.
59. McPartland JC, Reichow B, Volkmar FR. Sensitivity and specificity of proposed DSM-5 diagnostic criteria for autism spectrum disorder. *J Am Acad Child Adolesc Psychiatry.* 2012;51:368-383.
60. Mattila ML, Kielinen M, Linna SL, et al. Autism spectrum disorders according to DSM-IV-TR and comparison with DSM-5 draft criteria: an epidemiological study. *J Am Acad Child Adolesc Psychiatry.* 2011;50:583-592.
61. Worley JA, Matson JL. Comparing symptoms of autism spectrum disorders using the current DSM-IV-TR diagnostic criteria and the proposed DSM-V diagnostic criteria. *Res Autism Spectr Disord.* 2012;6:965-790.
62. Matson J, Belva B, Horovitz M, Kozlowski A, Bamburg J. Comparing symptoms of autism spectrum disorders in a developmentally disabled adult population using the current DSM-IV-TR diagnostic criteria and the proposed DSM-5 diagnostic criteria. *J Dev Phys Disabil.* In press.
63. Matson J, Kozlowski A, Hattier M, Horovitz M, Sipes M. DSM-IV versus DSM-5 diagnostic criteria for toddlers with autism. *Dev Neurorehab.* 2012;15:185-190.
64. Frazier TW, Youngstrom EA, Speer L, et al. Validation of proposed DSM-5 criteria for Autism Spectrum Disorder. *J Am Acad Child Adolesc Psychiatry.* 2012;51:28-40.
65. Jones KD. A critique of the DSM-5 field trials. *J Nerv Ment Dis.* 2023;200:517-519.
66. Gibbs V, Aldridge F, Chandler F, et al. Brief report: an exploratory study comparing diagnostic outcomes for autism spectrum disorders under DSM-IV-TR with the proposed DSM-5 revision. *J Autism Devel Dis.* 2012;42:1750-1756.
67. Taheri A, Perry A. Exploring the proposed DSM-5 criteria in a clinical sample. *J Autism Devel Dis.* 2012;42:1810-1817.