The autism spectrum: definitions, assessment and diagnoses

This article summarizes current research related to autism spectrum disorders. Current epidemiological trends, theories about aetiology, and relevant issues in assessment and diagnosis of autism spectrum disorders are discussed.

Autism is a heterogeneous syndrome defined by a constellation of difficulties in social interaction and communication and by repetitive behaviours and interests. Because there is not yet a genetic or physiological marker for autism, the diagnosis is based purely on behaviour and developmental history. Below, a summary is provided of recent research about definitions of autism and epidemiological findings. Issues concerning diagnostic assessment are also addressed.

Definitions of autism and autism spectrum disorders

In recent years, the conceptualization of autism has changed considerably from that of primarily non-verbal, severely learning disabled children with very obvious handicaps to a much broader spectrum of disabilities that includes highly specific social-cognitive deficits in individuals with otherwise relatively normal non-verbal skills. Although there have been a number of attempts to separate autism into independent components, the association between social and communication deficits and repetitive behaviours and/or restricted interests has remained strong. This is true not only in individuals with autism spectrum disorders, but also in siblings and other family members of these individuals (Bailey et al, 1995, 1996), and even within the general population (Constantino and Todd, 2003). Thus, there is evidence that there is something unique about the association between social and communication deficits and restricted and repetitive behaviours and interests, which merits consideration of autism as a syndrome. Nevertheless, the relationship between social and communication deficits and repetitive behaviours manifests itself in many different ways depending on the age and intellectual level of the individual.

There is a great deal of evidence that autism is a neurobiological disorder, although it is likely that autism is not one disorder with a single aetiology, but rather a collection of disorders with several different aetiologies. (Freitag, 2007) Because of the suspected neurobiological heterogeneity of autism spectrum disorders, autism spectrum disorders may be more similar to conditions such as cerebral palsy or intellectual disabilities, which can occur through multiple pathways, than it is to fragile X or Rett syndrome, which entail specific biological mechanisms.

Autism can be reliably diagnosed in children as young as 2 years of age, although diagnoses are less stable in children under 3 years than at any other time in development (Lord et al, 2006). There is better diagnostic agreement among clinicians and among standardized measures for more narrowly defined autism, which is often associated with significant language comprehension and other more general cognitive delays, than for milder difficulties. However, even less severely affected individuals with more broadly defined autistic symptoms, such as individuals with Asperger's syndrome or pervasive developmental disorder – not otherwise specified, most often remain affected by their disability throughout their lives (Howlin and Goode, 1998). Because pervasive developmental disorders, the term for the category describing these conditions in *Diagnostic and Statistical Manual of Mental Disorders* (4th edn) (DSM-IV) (American Psychiatric Association, 1994) and the *ICD-10 Classification of Mental and Behavioral Disorders: Clinical Descriptions and Diagnostic Guidelines* (ICD-10) (World Health Organization, 1992), is not easily interpretable, the term autism spectrum disorders has come to be adopted by advocacy groups, researchers, and clinicians.

Language delay is often the first reason why parents of children with autism seek help. In many ways, associated language delay is the most limiting aspect of autism spectrum disorders, in that in most cultures a child or adult who cannot speak fluently has very restricted opportunities for independence. In the majority of cases, severe language delays in autism spectrum disorders include significant problems in language comprehension and so differ from more common expressive language and/or speech impairments. These more severe language delays involving comprehension are usually, although not always, associated with additional, milder delays in

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non-verbal problem solving. However, in the last few years, the proportion of children identified in epidemiological studies with autism who have severe cognitive delays has decreased significantly as more children with milder difficulties have been identified (Chakrabarti and Fombonne, 2001).

Three domains of autism

One of the consequences of identifying many more children with autism spectrum disorders without severe language impairments is that language characteristics are being seen as an integral, but no longer defining, aspect of autism. There is a movement towards reorganizing the three domains of autism in ICD-10 and DSM-IV (social reciprocity, communication, and restricted/repetitive behaviours) to merge social aspects of communication (e.g. non-verbal communication such as gestures and eye contact, as well as conversation and socially-directed chat) into a more general social deficit category (Lecavalier et al, 2006).

In addition, there have been proposals to divide the third defining domain of autism, repetitive behaviours/restricted interests, into two sub-domains (Cuccaro et al, 2003). Sensory interests, repetitive use of objects (e.g. spinning objects, lining toys up) and specific types of hand and body mannerisms constitute a different kind of behaviour in trajectories across development and in associations with intellectual disabilities than more compulsive behaviours (e.g. needing all doors shut), rituals and insistence on certain routines (Cuccaro et al, 2003). Repetitive sensory motor behaviours are more common, affecting more than two-thirds of individuals with autism spectrum disorders across a range of age and ability levels. These behaviours also appear to be associated with intellectual disability, particularly after preschool. Behaviours in the insistence on sameness category are less common, tend to be more prevalent in older children, and occur relatively independently of language level and intellectual disability (Bishop et al, 2006). These findings are likely to affect new diagnostic frameworks as things move on from DSM-IV and ICD-10.

Asperger's syndrome

Asperger's syndrome has become an increasingly popular diagnosis among clinicians, parents and individuals. One of the greatest difficulties in using this term is that it has different meanings for almost everyone who uses it (Klin et al, 2005). The definition of Asperger's syndrome in ICD-10 and DSM-IV, which requires excluding individuals who meet criteria for autism, results in such a small number of cases that many investigators and clinicians have simply created their own diagnostic criteria (Klin et al, 2005). Most commonly, Asperger's syndrome is used to describe individuals with social difficulties and some form of repetitive or restricted interest or behaviour associated with autism, who have fluent language and no intellectual disability. However, definitions of Asperger's syndrome vary widely among different clinicians and researchers.

The number of individuals who do not meet either autism or Asperger's syndrome criteria, but who still have related, impairing conditions (e.g. pervasive developmental disorder – not otherwise specified, atypical autism) is greater than either other condition. Establishing diagnostic guidelines for these individuals beyond saying they have pervasive developmental disorder – not otherwise specified, or 'not quite autism', will be an important goal for those working on new diagnostic frameworks. Persons with atypical autism or pervasive developmental disorder – not otherwise specified have a more variable prognosis than individuals with autism, but in most cases require substantial educational, social and psychiatric services that are appropriate to their individual needs, even if they fall outside of the more clearly defined criteria for autism and the more popularized conception of Asperger's syndrome (Lord et al, 2006).

Prevalence

Epidemiological studies began to show an increased prevalence of autism in the mid-1980s with the publication of DSM-III-R. In the last 10 years, prevalence studies have included children with more broadly defined autism spectrum disorders, and have shown continuing increases in numbers, which are likely to be associated mainly with changing definitions and increased public awareness of autism spectrum disorders (Fombonne, 2005). The most recent figures reported have indicated prevalence rates of autism spectrum disorders of about 1 in 166 births. Given the sex ratio of approximately 4 males to 1 female, this translates to about 1 in 103 males and 1 in 415 females.

In addition, at least in the USA, large increases in autism have paralleled smaller decreases in educational classifications of mental retardation, although these shifts do not account completely for the increased prevalence of autism (Shattuck, 2006). Furthermore, estimates of children receiving educational services for autism spectrum disorders are still below estimates of prevalence from population studies that used multiple methods of ascertainment, implying that numbers of children with autism spectrum disorders will continue to rise over the next few years.

Diagnosis and assessment

Standardized diagnostic instruments, such as the autism diagnostic interview revised (ADI–R) (Le Couteur et al, 2003) and the autism diagnostic observational schedule (ADOS) (Lord et al, 1999), have had a significant effect on research by creating uniform standards for participants. This has made possible the comparison of samples across studies, as well as the merging of samples within analyses. Larger samples are particularly important to

neurobiological studies because of the likely aetiological heterogeneity of the disorder.

Assessment

Assessments of individuals suspected of having autism spectrum disorders must include attention to the individual's language level, including receptive and expressive skills, and a separate measure of non-verbal intellectual ability. It is important to measure receptive and expressive language abilities separately; relative deficits in receptive language (compared to expressive language) have been found to differentiate children with fragile X or tuberous sclerosis without autism spectrum disorders from those with comorbid autism spectrum disorders (Rogers et al, 2001). In addition, separate verbal or language tests and non-verbal tests are also necessary, because the majority of individuals with autism have performance scores that exceed verbal scores by more than a standard deviation. Consequently, a single full-scale IQ score is not particularly meaningful except in more able individuals for whom verbal and performance scores may be more similar.

Although language level and non-verbal ability are not part of the autism spectrum disorders diagnosis, this information is crucial for interpreting results from diagnostic instruments. On the ADI-R, children with non-verbal mental ages below 18 months almost uniformly receive scores within the range of autism, regardless of their clinical diagnoses (Risi et al, 2006). The ADI-R may nevertheless be useful in describing behaviours of profoundly impaired individuals, but the diagnostic thresholds are not meaningful. Similarly, on the ADOS, children with non-verbal mental ages under 15 months almost always exceed diagnostic thresholds for autism spectrum disorders diagnoses, regardless of clinical diagnosis, again suggesting that, while the ADOS may be useful with this group for descriptive and baseline analyses, the algorithms are not sufficiently specific with very young and/or very delayed children. To address this issue, a new module of the ADOS is being developed specifically for toddlers. The trajectories of very young children between 12 and 24 months on the toddler ADOS may also be useful in measuring progress.

As instruments such as the ADI-R and ADOS have been more extensively studied, evidence has accumulated that they measure somewhat different concepts. This is not surprising given that the ADI-R is based on parent report of lifetime behaviours, and the ADOS is based on the observations of a skilled clinician within a brief window of time. Research has indicated that, when used together, the sensitivity and specificity of the ADI-R and ADOS for autism is very high, as measured by the gold standard of a best estimate consensus diagnosis (Risi et al, 2006; Bishop et al, 2007). Use of the ADOS and the social communication questionnaire produced almost as high sensitivity and specificity as the ADI-R in predicting best estimate diagnoses of autism and autism spectrum disorders (Corsello et al, 2007). The same may be true of other combinations of instruments involving both parent report and clinician observation.

The predictive validity of the ADI-R and ADOS for autism in children from as young as the age of 2 years up to 9 years of age is also very high (Lord et al, 2006). However, clinical judgment adds significantly to this predictability, particularly for children who fall within the range of pervasive developmental disorder – not otherwise specified or atypical autism. How a clinician weighs information from multiple sources (e.g. parent report and direct observation) may depend on who collected the information. If a clinician is collecting the information directly, he or she will tend to weight the ADOS more heavily. On the other hand, if a clinician is basing a decision on written scores of tests administered by someone else, he or she will tend to place more weight on the ADI-R (Risi et al, 2006). The point, which also seems relevant to instruments other than the ADI-R and ADOS, is that information from parents, observations of a skilled examiner, and the judgment of an experienced clinician, each provide separate contributions to a valid diagnosis of autism.

Pervasive development disorder – not otherwise specified

Diagnoses of pervasive developmental disorder – not otherwise specified or atypical autism are more problematic. In relatively high functioning individuals, differential diagnosis between autism spectrum disorders and other disorders, such as attention deficit-hyperactivity disorder (ADHD), can be especially difficult (Reiersen et al, 2007). The standardized instruments are useful in terms of providing metrics that allow for comparisons across studies. Yet when thresholds are set low enough to include more than about 70% of children receiving autism spectrum disorders clinical diagnoses (not including autism), the specificity of the instruments falls to about 70–75% (Risi et al, 2006).

New algorithms for the ADOS have improved specificity and sensitivity. They still have less predictive power for verbally fluent, older youngsters than would be ideal (Gotham et al, 2007), although probably yield better agreement about non-autism autism spectrum disorders diagnoses than one would find across independent clinicians. Using DSM-IV criteria in a literal way for autism spectrum disorders diagnoses elicited more disagreement among experienced clinicians than just asking for their global diagnoses (Volkmar et al, 1994). This highlights the importance of referring children suspected of having autism spectrum disorders to trained clinicians who have enough experience in making these diagnoses that they do not need to rely on verbatim DSM-IV criteria when making diagnostic decisions about autism spectrum disorders.

Conclusions

The field has made significant advancements over the past several years in developing and refining assessment and diagnostic practices for autism spectrum disorders. Questions still remain as to the optimal organization of impairments included in the diagnostic criteria for autism spectrum disorders, as well as how best to categorize children with milder and/or less clear forms of autism spectrum disorders (e.g. pervasive developmental disorder – not otherwise specified). **BJHM**

Conflict of interest: Dr C Lord receives royalties from the ADI-R and ADOS. However, as part of her conflict agreement with the University of Michigan, all profits from Michigan research are donated to charity.

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KEY POINTS

- The three domain definition of autism may be more accurately represented as deficits in social skills and communication, as well as the presence of repetitive sensory motor and/or insistence on sameness behaviours.
- Measurement of language skills and non-verbal problem solving is crucial to interpreting diagnostic information about children and adults suspected of having autism spectrum disorders and for developing treatment plans. However, delays in language or non-verbal problem-solving abilities are not diagnostic in themselves.
- The prevalence of autism spectrum disorders is increasing as more children are identified and as definitions broaden, with some evidence of diagnostic substitution, at least in the USA. Numbers of children with educational categories of autism spectrum disorders are likely to increase further, given that multi-method full population studies have shown even greater prevalence than studies using educational classifications.
- Diagnoses of autism and autism spectrum disorders can be made reliably in children as young as 2 years of age, although diagnoses become significantly more stable at the age of 3 years.
- Diagnoses using standardized diagnostic instruments and clinical judgment yield the most valid prognoses. A combination of parent report, structured observation by a trained examiner, and a clinician who considers all available information yielded the most accurate predictions of diagnoses from 2–9 years of age.
- Diagnoses of autism in children and adults are more reliable than diagnoses of non-autism autism spectrum disorders. At this point, there are unavoidable trade-offs between sensitivity and specificity for both clinical diagnoses and standard diagnostic instruments.