Diagnosing autism/autism spectrum disorders

Kirsty Yates

Ann Le Couteur

Abstract

Awareness of autism spectrum disorder (ASD) within public and medical domains has increased. Demand on services is high as children and young people are being presented for earlier advice, assessment and diagnosis. To maximise detection and minimise harm it is essential for all clinicians working with children including primary care teams, allied healthcare professionals, educational and social care staff to have a sound knowledge of the presentation and assessment of autism spectrum disorders (ASD) and an understanding of the co-morbidities. Whilst routes of entry for referrals can vary due to a diversity of presentation and local service provision, there are standards in the recognition, referral and diagnosis of autism. Early identification is advantageous in order to maximize the child's potential, provide appropriate support and targeted intervention for ASD and co-occurring conditions with the aim of improving outcomes. This review addresses the diagnosis of ASD and provides an assessment framework for professionals who encounter a child with a suspected autism spectrum disorder.

Keywords autism; autism spectrum disorder; co-morbidity; diagnosis; multidisciplinary assessment

What's new?

- Changes to the DSM International diagnostic classification criteria were introduced in 2013. Autism Spectrum Disorder is the only diagnostic category. ICD-11 is expected in 2018 and likely to be similar.
- A new diagnosis of Social (pragmatic) communication disorder has been introduced in DSM-5 to identify those individuals with persistent deficits in social communication and social interaction in the absence of restricted, repetitive patterns of behaviour, interests and activities. The clinical and research utility of this new diagnosis is to be ascertained.
- NICE Guidance on the support and management of Autism in under 19's was published in 2013. Both NC128 & 170 are currently under systematic review to consider the need for updating.
- There is continued emphasis on transition planning to young adulthood, supported by the Children and Families Act 2014, Adult autism Statutory Guidance 2015 and the Autism Act 2009. NICE have recently published guidance on behaviours that challenge in the learning disability population and transition in young people using health or social care services.

What is autism/autism spectrum disorder?

ASD is a lifelong neurodevelopmental disorder. The term autism refers to the prototypical condition described in 1943 by Leo Kanner, also known as core autism. However, it is well recognized that there is a spectrum of presentation and a broader autism phenotype with less severe and more subtle behavioural features that may only manifest after a change in environmental demand. Autism spectrum disorder has been characterized by qualitative behavioural abnormalities in communication, reciprocal social interaction together with patterns of repetitive, restricted and stereotyped interests and activities. These deficits are pervasive, persistent, usually present in early childhood and likely to lead to impairments in functioning across different settings.

Kirsty Yates MBBS MRCPCH is a Consultant Paediatrician, Chester Le Street Community Hospital, Durham, UK. Conflict of interest: none declared.

Ann Le Couteur BSc MBBS MRCPsych FRCPsych FRCPCH is Professor of Child and Adolescent Psychiatry, Institute of Health and Society, Newcastle University, Sir James Spence Institute, Royal Victoria Infirmary, Newcastle upon Tyne, UK. Conflict of interest: ALC is one of the authors of the ADI-R and a member of the Clinical Guideline Group for the ASD NICE guidelines.

There are currently two international classification systems for diagnosing ASD. In 2013 the American Psychiatric Association revised the Diagnostic Statistical Manual (DSM-5). Changes included the introduction of Autism Spectrum Disorder as a single diagnosis and the removal of the diagnostic subgroupings (autism, Asperger's syndrome, atypical autism); combining the qualitative impairments of social communication and social interaction into one diagnostic domain and expanding the Restrictive, Repetitive Behaviours and Interests domain (see below in section on Symptoms and Signs) to include stereotyped and repetitive speech, hypo- and hyper-reactivity to sensory input and unusual sensory interests.

DSM-5 recommends the use of a range of specifiers highlighting the importance of addressing the individual's profile of strengths and needs. These include severity specifiers that may be used to describe current symptomology for each of the ASD domains with the recognition that severity may vary with time and environmental context so should not be used to determine eligibility for and provision of services. Specifiers also include whether there is intellectual disability, language impairment, other associated disorders or comorbidities e.g. medical, genetic, mental or behavioural. The current World Health Organisation (ICD-10) criteria are based on the original triad of impairments, though ICD-11 update is expected in 2018 and likely to be similar to DSM-5.

Epidemiology

ASD is not rare. The National Institute of Health and Clinical Excellence (NICE) states the diagnosis is queried in approximately 3% of the child population and epidemiological studies suggest prevalence rates of at least 1 in 100. Broadening of diagnostic criteria and improved case recognition are likely to have contributed to the increase in diagnostic rates reported

since the 1990s. There are no high quality robust studies confirming a rise in the true prevalence. The condition is three to four times more common in boys, with a male preponderance rising in the high functioning group. NICE guidelines recognise that in clinical practice girls may be under-diagnosed. It has been suggested that high functioning females may be better at masking their difficulties through imitation and observation of social actions and better verbal skills.

What causes autism spectrum disorders?

ASD is accepted to be a neurodevelopmental condition with a biological basis. The heterogeneity of affected individuals and genetic complexity has undoubtedly contributed to the daunting task of identifying the cause(s) of ASD. Continuing research has not identified a clear aetiology, but evidence suggests that it has a complex genetic basis with strong heritability (60% concordance reported in twin studies). Recurrence rates for siblings have been reported between 3 and 10% with up to 18.7% when the broader autism spectrum is considered.

Advances in molecular genetics have identified genetic variations e.g. 'rare causal' copy number variants and single gene polymorphisms which are significant or 'causal' in approximately 10% of people diagnosed with ASD. *De novo* events may be implicated in simplex families, whereas multiplex families (when more than one family member is affected by ASD) may pass a specific genetic variation through the generations which increases the risk of ASD. It is possible that several genes of small effect may act through an epigenetic mechanism and environmental factors influence phenotypic expression. In 10–15% of cases ASD is associated with a known medical condition. Consistently recognized genetic conditions include tuberous sclerosis (TS) and fragile X. Studies have shown that between 1% and 3% of children with autism have TS and similar percentages have fragile X. Other associations and a list of additional medical risk factors are shown in Box 1.

Risk factors for autism spectrum disorder
- Sibling with ASD
- Parental schizophrenia-like-psychosis or affective disorder
- Maternal sodium valproate use during pregnancy
- Gestational age less than 35 weeks
- Intellectual disability
- Birth defects associated with central nervous system including cerebral palsy
- Down syndrome
- Fragile X
- Tuberous sclerosis

Other medical conditions associated with ASD
- Neurofibromatosis
- Phenylketonuria (untreated)
- Fetal alcohol syndrome
- Smith–Lemli–Opitz syndrome
- CHARGE syndrome
- Duchenne muscular dystrophy
- Congenital rubella
- Iron-deficiency anaemia

Box 1 Risk factors and medical conditions associated with autism spectrum disorder

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Clinical research has demonstrated differences in trajectories of head growth in children with ASD. Macrocephaly is a recognized feature of ASD in 20–30% of cases though must be interpreted in the context of parental head circumferences. Studies have shown that as a group, head circumference accelerates during the first 2 years of life, with deceleration possibly occurring in later childhood since average head circumference has been reported in adolescence and adulthood. Although there have been conflicting views around the relevance and cause of these changes, they are reported to happen prior to the onset of clinical symptoms and may be a useful clinical indicator. Gene mutations in *PTEN* (Phosphatase and Tensin Homolog) have been found in children with ASD and macrocephaly with case series reporting a yield of 5% in those with head circumferences greater than 98th percentile.

Research continues to study neurobiological differences in ASD considering variation in neurotransmitters, volumetric and functioning differences of various regions within the brain, but the relevance to clinical practice of most identified abnormalities has not been established.

Various environmental factors have been reported in the literature. Risk factors are shown in Box 1 and include prematurity less than 35 weeks gestation, prenatal maternal valproate use and congenital rubella. The controversy of links between the MMR vaccine and ASD are unfounded.

Making a diagnosis

ASD is a heterogeneous condition with no single pathognomonic feature or specific diagnostic test. Diagnosis can be challenging as affected individuals display variation in the degree of behavioural severity, language and intellectual abilities. Moreover, their behavioural profiles are likely to change with age and co-occurring problems and co-morbidities are common. DSM-5 recognises that symptoms in the early developmental period may not manifest until capabilities are exceeded by social demands. Similarly, there is recognition that for some adolescents, repetitive behavioural manifestations are reduced through developmental progress or intervention so criterion can be met based on history. For a diagnosis of ASD under ICD-10, abnormal or impaired development should be present by the age of 3 years.

Many parents express concerns as early as 15–18 months of age, but despite increased awareness and guidance, average age at diagnosis remains at 4–5 years. This is possibly due to a combination of factors that include variability of assessment pathways, demand on services, lack of recognition of subtle difficulties at a young age, presence of additional diagnoses and inclusion of school age individuals who may only present at an older age when their difficulties may become more overt as they are unable to manage increasingly challenging academic and/or social expectations. Studies have shown that diagnosis of ASD at 2 years of age is possible and stable over time, although it is less reliable for the broader autism spectrum.

Symptoms and signs of autism spectrum disorders

Social communication
Difficulties and delay in social interaction are often the earliest features in ASDs, but they can be subtle and easily missed. Absence of joint attention (i.e. failure to show interest, share a