

Diagnosing Autism Spectrum Disorder

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Abstract

Autism Spectrum Disorder (ASD), a lifelong neurodevelopmental disorder characterised by qualitative impairments in social communication together with patterns of restricted, repetitive and often stereotyped interests and behaviours, affects approximately 1% of the population. The revised DSM 5 criteria, published in 2013, now use ASD as a single diagnostic category. Clinical presentations will vary between individuals and across chronological and developmental age and so possible signs of ASD must be interpreted with reference to the “normal” range of skills expected for a given age and in the context of an individual’s broader developmental profile. Where there are concerns about possible ASD, a referral should be made for an ASD diagnostic assessment. In addition to determining whether an individual reaches the required criteria for an ASD diagnosis and considering relevant differential diagnoses, a multidisciplinary ASD diagnostic assessment should provide an individual skills-and-needs-based profile and identify co-morbidity. The multidisciplinary team’s diagnostic formulation and individualised recommendations about future support and interventions (informed by the skills-and-needs profile), should be shared with the family/carers, and where appropriate the individual undergoing assessment. With consent, the diagnostic report can also be shared with relevant education, social care and health care services.

Introduction

Autism Spectrum Disorder (ASD) is a lifelong neurodevelopmental disorder characterised by qualitative impairments in social communication together with patterns of restricted, repetitive and often stereotyped interests and behaviours. Previously considered to be a rare disorder, it is now estimated that ASD affects approximately 1% of the population (1), leading to increasing pressure on community child health, child and adolescent mental health (CAMHS) and adult mental health services to provide pathways for ASD diagnosis.

ASD across the lifespan

Clinical presentations will vary between individuals and across chronological and developmental age. Indeed there are no pathognomonic features of ASD. Some signs and symptoms may be identified in early childhood and remain relatively constant, others may vary over time or only become apparent as relevant developmental milestones are encountered and/or social communication and educational/occupational expectations increase. For these reasons it is important that any possible ASD features are interpreted with reference to the “normal” range of skills expected for age and within the context of an individual’s broader developmental profile.

Age at presentation will impact upon the process of assessment, the professional team undertaking the diagnostic assessment, the range of differential diagnoses considered and the types of interventions or supports recommended for future care.

After briefly mentioning diagnostic criteria, relevant national policy and clinical guidelines, this paper

will consider clinical presentations of ASD with reference to different age groups.

Diagnostic Criteria

The diagnostic criteria remain the same regardless of the age at which an individual presents.

The fourth edition of the Diagnostic and Statistical Manual (DSM IV) (2) and the 10th edition of International Classification of Diseases (ICD-10) (3) provide a range of diagnostic sub-groupings in an attempt to reflect clinical heterogeneity (e.g. autism, Asperger's Syndrome, Pervasive Developmental Disorder Not Otherwise Specified). Following concerns about the validity of these sub-groups and the reliability with which the different terms were used (4) the recently published DSM-5 (5) uses ASD as the single diagnostic category.

The revised criteria (figure 1) contain two behavioural domains - social communication and repetitive stereotyped behaviours (including a new sub-domain of unusual hyper-reactivity or hypo-reactivity to sensory input) – rather than the previous three (qualitative impairment in social interaction, qualitative impairment in communication and restrictive, repetitive and stereotyped patterns of behaviour, interests and activities), with each behavioural domain containing several sub-domains. To qualify for an ASD diagnosis there must be evidence of symptoms in all three social communication sub-domains and two of the four repetitive stereotyped behaviour sub-domains. Reflecting recent research, which has highlighted high rates of additional mental health needs and other co-occurring problems in those with ASD (6), DSM-5 now includes provision for clinicians to indicate co-morbidity, a practice that both ICD 10 and DSM IV discouraged. The 11th edition of the International Classification of Diseases is likely to be published in 2015.

National Policy and Guidelines

A number of ASD clinical guidelines and best practice documents have been published internationally (7-14). Recently published evidence-based National Institute for Health and Care Excellence (NICE) guidelines and Quality Standards include recommendations about ASD assessment for children, young people and adults (11-15). For further details please see paper on NICE guidelines.

Signs and symptoms of ASD

Pre-school children

From birth to school age, children negotiate a wide range of different developmental challenges and milestones. Particular patterns of delayed development or unusual features may raise concerns about ASD (11).

Most children with ASD will not receive a diagnosis until they are 4-5 years old, despite many parents first expressing concerns about their child's development at 12-18 months (16, 17) and accurate diagnosis being possible for some children from at least 2 years onwards. In addition to parental concern, health visitors and, where applicable, nursery staff may be well placed to identify the presence of unusual or delayed development and behaviour including difficulties with social communication. A number of screening tools have been developed in an attempt to improve early identification of ASD, although universal screening for ASD is not currently recommended in the UK (please see Early Screening paper for further details).

When considering possible social communication deficits associated with a diagnosis of ASD in young children, early features reported in infants include a delayed or absent response to their name being called, problems initiating and maintaining joint attention and limited use of social greeting (smiles) and/or facial expressions. Parents also commonly report language delay or an apparent loss of previously acquired speech. The pattern of loss of speech is seen uncommonly in speech and language disorders, whereas for a quarter to a third of young children with ASD, parents

report a history of loss or regression of language and less frequently loss of other skills (18). Other communication abnormalities may include odd or flat intonation, frequent repetition of words and phrases or a reduced use of language in order to communicate. Some commonly reported social impairments include problems with social interest in other people (especially other children), clumsy or disruptive attempts to interact and reduced use of gestures and eye contact to communicate. Young children with ASD may prefer to play alone and may struggle understanding social imitative and imaginative play even when initiated by parents or other familiar adults. Repetitive, stereotyped interests and behaviours such as motor mannerisms (e.g. hand flapping), unusual play (e.g. ordering toys by colour or size), intense or unusual interests that interfere with other activity, difficulties managing changes to daily schedule or surrounding environment and unusual reactions to sensory stimuli may also prompt professionals, and indeed parents, to think about ASD.

School-aged children

The start of formal education brings new challenges, many of which may be particularly difficult for children with ASD - spending time outside the family with unfamiliar children and adults, following explicit and implicit (socially expected) rules, change to existing routines. Many young children with ASD will struggle to understand the increasing social communication demands of learning environments. These challenges, combined with observations across settings from the range of adults that the young child comes into contact with, together with the process of monitoring early years outcomes (19) often highlight a pattern of emerging difficulties. In contrast some children, perhaps as a consequence of their own abilities and relative strengths, may negotiate some or all of these hurdles without raising concerns and so not receive a diagnosis until they are much older (17).

In addition to the signs and symptoms seen in the pre-school period, children with ASD might begin to stand out in other ways. Alongside speech delay, abnormal intonation and a reduced quantity, speech may be increasingly repetitive, containing stereotyped phrases or frequent references to particular areas of interest. Children may use their speech to broadcast information rather than engaging in a two-way reciprocal conversation. They may make rude or inappropriate statements and fail to moderate their behaviour to fit different situations – for example talking loudly in the library or inappropriately to a member of staff – unaware of the social rules that peers have absorbed. A tendency to interpret things in a concrete manner and from their own point of view may manifest itself in difficulties understanding sarcasm, humour or the intentions of others and/or an insistence that all “rules” are obeyed. Similarly instructions may be followed precisely without understanding of metaphorical meaning – for example reaching down to sort out socks when told to “pull your socks up”.

For these older children with ASD, not noticing or misinterpreting subtleties of communication (facial expressions, gestures, body language), difficulties putting themselves in someone else’s shoes and a lack of social understanding may contribute to confusion about the actions and reactions of others. In turn if a child with ASD does not use non-verbal methods of communication, peers, family members and teaching staff may be less able to pick up how they are feeling and moderate their social approaches and responses accordingly.

Adolescence and Transition to Young Adulthood

For some individuals who have progressed through early education without raising concerns, underlying deficits may only become apparent with the increasingly complex social and developmental requirements of secondary school, adolescence or adulthood.

For those young people with unrecognised ASD, the increasing independence of adolescence combined with a lack of social understanding may lead to “risky” behaviour. Such previously “unseen” vulnerabilities may prompt teachers, parents and other professionals to consider ASD, whilst the young person themselves may become increasingly aware that they are in some way “different” from their peers. Mental health and other neurodevelopmental (e.g. ADHD) co-morbidities are common and may be the presenting problem for a proportion of young people with previously undetected

social communication difficulties. A recent UK population study reported that 70% of young people with ASD (aged 10-14) met criteria for one co-morbid disorder and 41% for two or more (6).

The transition from adolescence into young adulthood presents further developmental targets - moving into employment or adult-based education, accessing adult health services, living independently – that can be challenging for regularly developing young people but are often particularly difficult for individuals with ASD. For those individuals reaching adulthood without a diagnosis, problems negotiating these transitions may indicate previously unrecognised deficits and the need for a diagnostic assessment.

Finally, for adults and indeed older adults who may present for an ASD assessment following difficulties in later life (failed personal relationships, not progressing in the workplace, loss of parents, inability to manage change in living arrangements), obtaining a developmental history may present practical challenges. In addition to the length of time that has passed since the early developmental period, sometimes there may not be a suitable informant, as may also be the case for looked-after or adopted children.

The adult autism strategy requires that staff working in health and social care are able to recognise the signs and symptoms of ASD in adults. With reference to screening the recently published adult-orientated NICE guidelines (13) did not find sufficient evidence to support the routine use of any screening instrument in primary care (and other settings) to improve case identification and recognition of adults with autism.

Diagnostic assessment

Where there are concerns about possible ASD, a referral should be made for an ASD diagnostic assessment and a skills-and-needs-based assessment. A diagnostic assessment should take into account the concerns of the family/carers and, where appropriate, the individual undergoing assessment, make use of existing information and combine clinical assessments across a variety of settings (figure 2).

In addition to determining whether the developmental history and presenting problems reach diagnostic criteria for ASD, and considering relevant differential diagnoses (figure 3), a multidisciplinary ASD diagnostic assessment should provide an individual skills-and-needs-based profile (figure 4) and identify co-morbidity. Professionals undertaking assessments should have experience of ASD across the ability range. Individual clinical presentations will dictate the need for supplementary assessments (e.g. cognitive, speech and language, motor co-ordination or sensory assessments) and the range of professional disciplines involved in the assessment process. Decisions about biomedical investigations such as neuroimaging, genetic testing and electroencephalography are similarly guided by individual presentation and should only be undertaken if clinically indicated.

Unfortunately at present there is little consistency and wide variation in provision across the UK. Most pre-school children are assessed by the local child health team and school-aged children by CAMHS teams but there is less consistency for adults with adult mental health services or specialist service providers or, in some areas, as yet no services with the established skills to provide ASD diagnostic pathways for adults, defined as from 18 years.

Diagnostic Instruments

There are a number of diagnostic instruments, some of which were originally developed for research, that can provide a standardised structure for components of an ASD assessment. The Autism Diagnostic Interview – Revised (ADI-R) (20), the Diagnostic Interview for Social and Communication Disorders (DISCO) (21) and the Developmental, Dimensional and Diagnostic Interview (3di) (22) are semi-structured interviews which provide a framework for a developmental history. All three require specific training. The DISCO and to a lesser degree the 3di also include information focussing on other

possible co-occurring problems. The Autism Diagnostic Observation Schedule (ADOS) (23) is a semi-structured standardised play, activity and conversational assessment that allows the trained examiner to observe behaviours relevant to ASD. These diagnostic instruments provide a useful structure for gathering information and examples of behaviour but are not to be used in isolation and should be used alongside other assessments to inform a comprehensive clinical formulation.

Outcomes of assessment

At the end of the assessment the multidisciplinary team's diagnostic formulation should be shared with family members, carers and, where appropriate, the individual undergoing assessment, before being brought together in a diagnostic report. Whilst the term ASD may be a helpful way of highlighting the types of difficulties a child, young person or adult experiences, in itself it is unlikely to be of practical help (given the wide variation seen between individuals on the autism spectrum). The detailed description provided by a skills-and-needs-profile will help a family and, with increasing age, the individual undergoing assessment, understand what a diagnosis means for them and inform individualised recommendations for future interventions and support.

With consent, the diagnostic report and recommendations can be shared with relevant education, social care and health services. The assessment team should also offer a follow-up appointment and should also signpost to local community-based resources (especially those tailored towards ASD) and additional financial supports that may be available. Consider a second opinion if there are disagreements or uncertainty around diagnosis.

Figure 1:

DSM-5 Autism Spectrum Disorder Diagnostic Criteria (5)

1. Persistent deficits in social communication and social interaction across multiple contexts
 - Deficits in social-emotional reciprocity
 - Deficits in nonverbal communicative behaviours used for social interaction
 - Deficits in developing, maintaining and understanding relationships
2. Restricted, repetitive patterns of behaviour, interests or activities
 - Stereotyped or repetitive motor movements, use of objects or speech
 - Insistence on sameness, inflexible adherence to routines or ritualised patterns of verbal or non-verbal behaviour
 - Highly restricted, fixated interests that are abnormal in intensity or focus
 - Hyper or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment
3. Symptoms must be present in the early developmental period
4. Symptoms must cause clinically significant impairment
5. Disturbances are not better explained by intellectual disability

Figure 2:

Core Components of ASD Diagnostic Assessment

- Detailed developmental history to establish the features of early development and current functioning necessary for the diagnosis of ASD as outlined by internationally agreed diagnostic criteria.
- Medical history, including details of any prenatal or postnatal complications.
- Family history to identify any family members with ASD, other neurodevelopmental disorders (e.g. dyslexia, attention deficit hyperactivity disorder, developmental co-ordination disorder) or mental health diagnoses which may indicate a degree of genetic vulnerability.
- Physical examination to identify dysmorphic features (e.g. fragile X), stigmata that may indicate an underlying disorder (e.g. tuberous sclerosis), to exclude differential diagnoses (e.g. Rett syndrome) and to assess general developmental level.
- Direct observations of the individual undergoing assessment, ideally in more than one context.

Figure 3:

Differential Diagnosis for ASD (11)

Neurodevelopmental disorders

- Specific language delay
- Global developmental delay
- Developmental co-ordination disorder

Mental and behavioural disorders

- Attention deficit hyperactivity disorder
- Mood disorder
- Anxiety disorder
- Attachment disorder
- Oppositional defiant disorder
- Conduct disorder
- Obsessive compulsive disorder
- Psychosis

Conditions with developmental regression

- Rett syndrome
- Epileptic encephalopathy

Figure 4:

Areas to consider in skills and needs based profile (11)

- Intellectual ability and learning style
- Academic skills
- Speech and language communication
- Fine and gross motor skills
- Adaptive behaviour – including self-help skills
- Mental and emotional health – including self-esteem
- Physical health and nutrition
- Sensory sensitivities
- Behaviour likely to affect day to day functioning and social participation
- Socialisation skills

GP Comment.

What have I learned from this paper?

1. Autism was previously considered to be a rare disorder but ASD is now thought to affect about 1% of the population. This means that both diagnosis and provision of resources constitute significant challenges.
2. The new diagnostic criteria have just two domains: social communication difficulties and repetitive stereotyped behaviours.
3. Although diagnosis is possible from 2 years onwards, most children are not diagnosed with ASD until they are at least 4 or 5 years old, sometimes much older.
4. Issues that may cause concern and raise suspicion of the diagnosis include delayed or absent response to name call, problems initiating and maintaining joint attention, limited use of smiles and facial expression, language delay or loss of language skills, odd intonation of speech, frequent repetition of words or phrases, difficulty with social communication as indicated by reduced gesture and eye contact, the tendency to play alone and repetitive, stereotyped interests, including unusual play, and sometimes motor mannerisms such as hand-flapping. If I notice these characteristics, I shall have a low threshold for referring for specialist opinion to determine whether the individual has ASD.
5. Because ASD can create major problems with regard to personal relationships, education and employment, early diagnosis and provision of appropriate services is of major importance.

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