Diagnosis and management of ASD in children and adolescents

Zoë Ellison-Wright MBBS, MSc, MRC Psych, Chrissy Boardman BM, MRC Psych, DCh

As part of our series on managing neurological and psychiatric conditions in children and adolescents, Dr Ellison-Wright and Dr Boardman discuss autism spectrum disorders (ASD).

Autism spectrum disorders (ASDs) affect around 1-1.5% of school age children. They are more common in boys than girls in a ratio of 5:1. They are developmental conditions involving a triad of impairments in (i) social interaction, (ii) social communication and (iii) restricted interests or repetitive behaviours. The ASDs include childhood autism, Asperger syndrome, atypical autism and other pervasive developmental disorders with autistic features.

Introduction
Over the last 50 years there have been great advances in the recognition and understanding of ASDs. Improved recognition has been achieved by the use of structured diagnostic assessment interviews, the formation of autism teams, better training of health staff and greater public awareness.

ASDs can affect individuals in many different ways. Some children (around 50%) have intellectual disability (IQ < 70) whereas others are of normal or above-average intelligence. In an Icelandic birth cohort, children with ASDs included 28% with autism (28% IQ > 70), 18% with Asperger syndrome (100% IQ > 70) and 54% with other ASD diagnoses (54% IQ > 70).

Early and accurate diagnosis is important in ensuring children are offered the optimum support and advice, enabling them to achieve the best quality of life.

A. Abnormal or impaired development is evident before the age of three years in at least one of the following areas:
   i. receptive or expressive language as used in social communication
   ii. development of selective social attachments or of reciprocal social interaction
   iii. functional or symbolic play.

B. A total of at least six symptoms from the following symptom clusters with at least two from (a) and at least one from each of (b) and (c):
   a. Qualitative abnormalities in reciprocal social interactions: eg failure adequately to use eye-to-eye gaze; lack of peer relationships that involve a mutual sharing of interests; lack of modulation of behaviour according to social context.
   b. Qualitative abnormalities in communication: eg relative failure to initiate or sustain conversational interchange; stereotyped and repetitive use of language; lack of varied spontaneous make-believe play.
   c. Restricted, repetitive, and stereotyped patterns of behaviour, interests, and activities: eg preoccupation with restricted patterns of interest; compulsive adherence to specific rituals; repetitive motor mannerisms that involve either hand or finger flapping or twisting, or complex whole body movements; preoccupations with non-functional elements of objects (eg their odour, the feel of their surface).

C. The clinical picture is not attributable to the other varieties of pervasive developmental disorder.

Diagnostic criteria for Asperger syndrome
The above criteria also apply except there is no general delay or retardation in language or cognitive development. There may, however, be other communication difficulties.

Box 1. Diagnostic criteria for childhood autism
The difficulties caused by mild ASDs may be subtle and cause behavioural problems involving conduct or attention, or anxious, obsessive or emotional symptoms. Mild ASDs may be missed in: girls; children with other psychiatric disorders; children with an intellectual disability; children who appear mature in their use of language, and children who demonstrate good eye contact or show affection (when they have learned to respond to others).

Diagnosis
ASDs are characterised in 1CD-10 by abnormal development before the age of three years and a triad of impairments (Box 1). ICD-10 refers to pervasive developmental disorders, which includes childhood autism and Asperger syndrome, atypical autism and...
some other pervasive developmental disorders. DSM-V now has a single diagnosis of autistic spectrum disorders. Asperger syndrome is usually diagnosed later than autism because children with Asperger do not have delay in language or cognitive development.

Causes
ASDs have a strong genetic basis. The heritability of ASDs in the population is around 90%. The rate of ASDs is about 25 times higher in siblings of affected children than in the general population. Most ASDs probably result from the combination of common variant (moderate risk) gene alleles or from a few rare (high risk) alleles but the detailed genetics has not yet been established.

In a small proportion of children (5%) a single gene disorder or chromosomal disorder is responsible, eg Fragile X syndrome. These cases can be identified by genetic testing.

Various risk factors have been identified, eg a sibling with autism, birth defects including cerebral palsy, gestational age less than 35 weeks, maternal use of valproate in pregnancy, chromosomal disorders such as Down’s syndrome, genetic disorders such as fragile X, muscular dystrophy, neurofibromatosis and tuberous sclerosis.

Assessment
GPs and healthcare staff should be able to identify the essential features of ASDs so they can refer patients for more specialist assessment. It is important to maintain a high level of vigilance for ASDs, otherwise more subtle cases may be missed. The Scottish Intercollegiate Guidelines Network (SIGN) provides a useful list of ‘clinical clues’ for possible ASD in pre-school, school-age and adolescent children (Box 2).

In pre-school children:
- delay or absence of spoken language
- looks through people
- not responsive to other people’s feelings
- lack of pretend play or social play
- unable to share pleasure
- does not point out objects to another person
- lack of gaze monitoring
- unusual or repetitive hand and finger mannerisms
- unusual reactions to sensory stimuli

In school-age children:
- persistent echolalia
- reference to self as ‘you’, ‘she’ or ‘he’ beyond three years
- unusual vocabulary for child’s age
- tendency to talk freely only about specific topics
- inability to join in play of other children (may manifest as disruptive behaviour)
- easily overwhelmed by social and other stimulation
- extreme reactions to invasion of personal space
- inability to cope with change

In adolescent children:
- socially ‘naïve’, not as independent as peers
- speech peculiarities
- difficulty making and maintaining peer friendships
- preference for highly specific, narrow interests or hobbies, or may enjoy collecting, numbering or listing
- strong preferences for familiar routines
- problems using imagination

Box 2. Clinical clues for ASDs (adapted from SIGN, 2007)

The Checklist for Autism in Toddlers (CHAT) was designed to identify 18-month old children at risk of ASD. Although the sensitivity is too low for population screening it can be used as an adjunctive tool to identify ‘clinical clues’ in younger children.

Autism team
Each area should have a local autism team which usually includes a paediatrician / child psychiatrist, psychologist and a speech and language therapist. The team provides a single point of access for referrals, and carries out ASD assessments, provides feedback and organises or refers on to other services.

History
It is important to clarify concerns of parents, and if appropriate, depending on their age and language skills, the child. The history includes the child’s development (enquiring about diagnostic criteria at various developmental stages), prenatal, perinatal, family history, past and current health conditions. It is also important to obtain information from the child’s school via a questionnaire or discussion.

A standardised assessment tool should be used to improve reliability of diagnosis. The main tools are:

i. the autism diagnostic interview-revised (ADI-R) with the autism diagnostic observation schedule (ADOS-G)

ii. the diagnostic interview for social and communication disorders (DISCO)

iii. the developmental, dimensional and diagnostic interview (3di)
Examination

Interaction with and observation of the child forms the basis of the examination. A standardised assessment tool can be used to supplement this information (eg ADOS-G). A mental state examination, especially in adolescents, is important to consider differential diagnoses of psychosis, obsessive compulsive disorder and emerging borderline personality disorder.

Physical examination may reveal skin stigmata of neurofibromatosis or tuberous sclerosis (using a Wood’s lamp), signs of injury, eg self-harm or child maltreatment, congenital anomalies or dysmorphic features, eg macrocephaly, microcephaly.10

Investigations

In some cases genetic testing or an EEG may be indicated. A paediatrician or paediatric neurologist should be consulted if the child is older than three years with regression in language, or is of any age with regression in motor skills.10 This is to consider differential diagnoses of acquired epileptic dysphasia (Landau Kleffner syndrome), Rett syndrome or other neurodegenerative conditions such as mitochondrialopathies.11

Differential diagnoses

A multi-axial framework can be useful in considering the differential diagnoses (and co-morbidities) of ASDs:

- **Axis 1**: Clinical psychiatric syndromes, eg attention deficit hyperactivity disorder (ADHD), mood disorder, anxiety disorders, obsessive-compulsive disorder, attachment disorder, oppositional defiant disorder, conduct disorder, psychosis, selective mutism, borderline personality disorder
- **Axis 2**: Specific disorders of psychological development, eg specific language delay, developmental coordination disorder
- **Axis 3**: Intellectual level
- **Axis 4**: Medical conditions, eg hearing impairment, visual impairment
- **Axis 5**: Psychosocial problems, eg maltreatment.

ASDs are frequently (around 70%) associated with comorbid disorders: ADHD, tics or Tourette syndrome, anxiety disorders, mood disorders, self-injurious behaviour, anorexia nervosa, selective eating problems, sleep difficulties, enuresis, encopresis, coordination problems, specific learning difficulties, intellectual disability10 and medical conditions such as epilepsy (7%).5

Formulation

Following the assessment, the autism team should meet to agree a formulation including diagnosis, cause, co-morbidity, strengths, resiliencies and management plan. The team provides feedback to parents and child including a written report.

Management

A local autism care pathway ensures consistent support across health, education and social care services. The autism team should ensure each child with ASD is allocated a key worker to co-ordinate their care and develop a care plan.17

Parent education

Psycho-education for families includes advice on optimising the home environment, supporting siblings, managing sensory sensitivities and challenging behaviour.

Written information should be available regarding local and national support organisations (Box 3), education sessions, ASD specific parenting programmes, opportunities to meet other families with experience of ASDs, welfare rights, social support and leisure activities.

Families should be given information about having a carer’s assessment and about respite care.

Parenting programmes

Further support can be offered through well validated parenting programmes which can provide a range of ‘tips’ for expert parenting. These programmes include Earlybirds for parents of preschool children, Earlybird Plus for parents of children age 4-8 years, Cygnets for parents of children aged 7–16 years and Incredible Years Parenting Course adapted for ASD. This latter programme is based on social learning theory and supports the parents in forming relationships with their children. All these programmes encourage parents to use play-based strategies to increase joint attention, engagement and reciprocal communication in the child, strategies to manage challenging behaviour and advice for the parents on looking after themselves.

Promoting learning

Supporting the child’s education is a crucial factor in management. To allow learning to happen, the initial challenge is to help the child to contain their arousal level by using clear explanations, routines, visual techniques and sensory integration skills. The learning
environment can be enhanced by providing visual supports such as words, pictures or symbols that are meaningful for the child, adjusting the amount of personal space given to the child and considering individual sensory sensitivities to noise levels, for example.  

A central objective of management is to encourage the child to gain skills, such as social skills, which they find difficult. Carers and professionals need to recognise that the child may find generalising difficult and therefore has to relearn the same skill when in a new situation. Structured social environments, such as special interest clubs, allows the practising of social skills in an easier setting than more informal groups such as youth clubs.

**Psychosocial Interventions**

Children should be offered specific social-communication interventions for the core features of autism to expand a child’s communication, interactive play and social routines. These should be delivered by a trained professional and take into account the child’s developmental level, language and communication skills.

Therapist modelling and video interaction feedback can help to increase the parents’ understanding and responsiveness to the child’s patterns of communication.

The following interventions are not suitable for managing core features of ASDs: antipsychotics; antidepressants; anticonvulsants, or exclusion diets (such as gluten- or casein-free diets).  

**Managing challenging behaviour**

Children with ASDs often exhibit challenging behaviour which can be driven by anxiety, frustration, sensory overload or anxious ruminations. This is compounded by the child having difficulty recognising an emotion in themselves and thinking that others can ‘mind read’ their thoughts.

These behaviours may be reduced by anticipating transition problems (eg changing schools), treating co-existing disorders, providing extra support for the family, or adjustments to the child’s environment, eg increasing structure and minimising unpredictability.

If specialist assessment is needed it should determine whether there is a triggering factor for the behaviour. If no triggering mental or physical disorder is identified, it should be managed using a psychosocial intervention. This involves a functional assessment to identify triggers, the patterns of behaviour, the consequences and reinforcing factors. This is followed by a psychologically-based intervention to reduce the challenging behaviour and reinforce alternative positive behaviours. This can be carried out by parents especially with support from a specialist parenting programme or by teachers.

Antipsychotic medication (eg risperidone) should be used only when psychosocial interventions are ineffective or could not be delivered because of the severity of the behaviour. It should be prescribed using the minimum effective dose and monitored by a paediatrician or psychiatrist. Benefits and side-effects should be reviewed after three to four weeks and regularly after that. It should be discontinued if there is no good response at six weeks. Local guidelines often suggest that it should not be used for longer than four months without a period off the medication because of the risk of longer term side-effects such as weight gain.

**Managing co-morbid disorders**

If a comorbid mental health problem is diagnosed, this should be treated by CAMHS following NICE guidelines for the specific disorder.

Anxiety disorders are frequently comorbid with ASDs. Individual cognitive behavioural therapy can be used if the child has sufficient language skills and if it is adjusted to the needs of children with autism. The child’s difficulty in generalising to situations outside the clinic room may be helped by including a parent in the sessions who can then reinforce learning in real-life situations.

Selective serotonin re-uptake inhibitors may be useful in treating depression or anxiety if psychosocial strategies alone are ineffective.

Sleep problems are best managed by behavioural techniques and a sleep plan. Medication (eg melatonin) should be used only if sleep problems persist and have a negative impact, and then should be used in conjunction with behavioural techniques and regularly reviewed.

**Outcomes**

The prognosis of ASDs depends on the individual and is influenced by the support and education during childhood. A study which investigated childhood ASD severity over 8–12 years identified four groups: a persistent high severity group (46%), a persistent moderately severe group (38%), a worsening group that tended to increase in ASD severity over time (9%), and an improving group that decreased in ASD severity over time (7%).

Outcome is better for those with an IQ above 50. A group of children diagnosed with autism at age seven years with mean IQ above 50 were followed up for two decades. A minority of adults achieved relatively high levels of independence, but most remained very dependent on their families.
or other support services. Stereotyped behaviours or interests frequently persisted into adulthood. In some occupations, ASD traits may be advantageous and scores on adult autism questionnaires are higher in some disciplines, e.g. mathematicians and scientists.20

Dr Ellison-Wright is a Consultant in Child and Adolescent Psychiatry at Blandford Hospital, Dorset HealthCare University NHS Foundation Trust, and Dr Boardman is an Associate Specialist in Child Psychiatry at Dorset County Hospital, Dorset HealthCare University NHS Foundation Trust.

Declaration of interests
No conflicts of interest were declared.

References