



Diagnosing autism spectrum disorders in primary care

Allely CS, Wilson P. Diagnosing autism spectrum disorders in primary care.

Practitioner 2011;255 (1745):27-30

Dr Clare S Allely
MA (Hons) MRes PhD
Research Assistant

Dr Philip Wilson
DPhil MRCPCH FRCGP
GP and Senior Lecturer

Institute of Health and Wellbeing, University of Glasgow, RHSC Yorkhill, Glasgow



Practitioner
Medical Publishing Ltd

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AUTHOR
Dr Clare S Allely
MA (Hons) MRes PhD
Research Assistant

Dr Philip Wilson
DPhil MRCPCH FRCGP
GP and Senior Lecturer

Institute of Health and
Wellbeing, University of
Glasgow, RHSC Yorkhill,
Glasgow



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What are the diagnostic criteria for ASD?

How should children be assessed?

Which patients should be referred?



AUTISM IS A DISORDER OF SOCIAL COMMUNICATION, FIRST DESCRIBED BY KANNER IN 1943¹ AS A

triad of impaired social interaction, communication, and imagination, associated with a rigid, repetitive pattern of behaviour. More recently the core deficit has been described as a lack of social instinct.^{2,3}

Autism, like most psychiatric disorders, covers a spectrum of severity from severely disabling classic autism to

milder forms of Asperger's syndrome which border on normality. The term autism spectrum disorder (ASD) includes autism, atypical autism and Asperger's syndrome.

Some parents consult their GP worried that their child might have autism. Others may attend with a range of concerns that might point to the condition such as problems with hearing, vision, hypersensitivity to sensory stimuli, behaviour management, language impairment or repetitive behaviour.

Autism conditions often co-exist with neurological disorders (particularly those including epilepsy), and more than 300 syndromes which include autism have been described.⁴

The new NICE guidelines, launched in September 2011,⁵ cover children, from birth up to 19 years, on the autism spectrum and build on the guidance published by SIGN in 2007.⁶

ASD was once believed to be relatively rare but is now thought to occur in about 1% of children.^{7,8} There



Table 1

Risk factors for ASD⁵

- ASD in a sibling
- birth defects associated with CNS malformation and/or dysfunction
- gestational age <35 weeks
- parental schizophrenia-like psychosis or affective disorder
- maternal use of sodium valproate in pregnancy
- intellectual disability
- neonatal encephalopathy or epileptic encephalopathy, including infantile spasms
- chromosomal disorders such as Down's syndrome
- genetic disorders such as fragile X syndrome; muscular dystrophy; neurofibromatosis and tuberous sclerosis

is certainly increasing demand for diagnostic services for children and young people. Whether the apparent increasing prevalence⁹ signifies an epidemic or an epidemic of discovery¹⁰ is debatable.

DIAGNOSTIC CRITERIA

The two primary diagnostic classification systems currently in use are the ICD-10¹¹ and the DSM-IV.¹² The ICD-10 is the most commonly used ASD classification system in the UK and forms the basis for the SIGN guideline.⁶

The DSM-IV, published by the American Psychiatric Association in 1994, defines autistic disorder by the presence of deficits or unusual behaviours within the following three domains together with onset before the age of three:

- reciprocal social interaction
- communication
- restricted, repetitive interests and behaviours

For the revised fifth edition of the DSM, there are proposed changes in the diagnosis of ASD.¹³ The revised criteria include only two symptom domains (social-communication and fixated, repetitive interests), the elimination of ASD subtypes, and the categorisation of individual differences in terms of dimensions of severity in the two domains.¹⁴ Some authorities consider that the proposed changes undervalue the importance of deficits in social imagination, and may lead to underdiagnosis in infancy, adulthood and in girls and women.¹⁵

RISK FACTORS

There is a substantial genetic basis with strong heritability.^{16,17} Approximately 70% of individuals with ASD also meet diagnostic criteria for at least one other (often unrecognised) mental and behavioural disorder.¹⁸

Factors associated with an increased prevalence of ASD are listed in table 1, left. Sleep problems in children and young people with ASD are also reported more frequently by parents.^{19–23}

SIGN states that GPs should be aware of the importance of considering comorbid problems in children and young people with ASD.⁶

REFERRAL

The NICE guidelines recommend the provision of an autism team in each area with a core membership consisting of: a paediatrician and/or child and adolescent psychiatrist, a speech and language therapist and a clinical and/or educational psychologist. In the absence of such a team, GP referrals would normally be to a paediatrician or a child or adolescent psychiatrist. The decision to refer should be made on the basis of signs or symptoms. However, there is also a need to consider the range, number, severity, duration, pervasiveness and impact of these signs and symptoms.

Particular attention should be paid to the degree of parental concern. With consent from parents or carers, and if appropriate the patient, information should be gathered from schools or other agencies. Before diagnosis, children may be labelled as 'naughty' and may also be underachieving, misunderstood and unsupported. They may also experience anxiety and distress about attending school or may even be excluded from school. Caution must, however, be exercised since the contribution of information from teachers and schools towards accurate diagnosis has not been thoroughly tested.⁶

With consent, sharing the profile or diagnostic assessment with professionals in education (for example through a school visit by a member of the autism team) can provide important information which contributes to the design of the individual education plan and needs-based management plan.⁵ By informing the school, additional support for learning (ASL) can also be implemented. Provision of this support is the responsibility of educational services.

Referral decisions should also take into consideration the presence of risk factors, see table 1, above. In cases where the signs and symptoms are not

sufficient to warrant an immediate referral, NICE recommends that a healthcare professional should consider keeping a close eye on the individual as they mature since signs and symptoms may change during this time. If the parent, carer or healthcare professional remains concerned, then a referral needs to be reconsidered.

The NICE guidelines also state that children younger than three years should be referred to an autism team if there is regression in language or social skills. However, these teams are not universally accessible throughout the UK. In children older than three with regression in language or motor skills, referral should be to a paediatrician or paediatric neurologist in the first instance.

Irrespective of any earlier assessments, re-referral for further ASD assessment should be considered at any age.⁶ ASD should not be ruled out because of: good eye contact; smiling and showing affection to family members; reported pretend play or apparently normal language development.⁵

EARLY RECOGNITION

The evidence that early intervention in ASD is better than later intervention in terms of developmental outcomes is relatively weak, and parents rather than children may be the principal beneficiaries. Nevertheless, early assessment for health problems and developmental disorders is likely to be desirable.⁶ Early identification of, and intervention for, developmental problems in general can improve outcomes as well as alleviating parental anxiety.²⁴ The evidence for developmental benefits of early intervention in ASD is largely based on observational studies.^{25,26}

Recognition and diagnosis may lead to the provision of autism-specific support to families, financial benefits and sometimes more appropriate education, which can lead to better outcomes for the individual.⁵

It should always be borne in mind that an incorrect diagnosis of ASD, and sometimes even a correct diagnosis, in the absence of significant functional impairment, can carry a risk of harm to families. Early detection research can nevertheless allow new insights into the developmental mechanisms underlying risk of ASD, produce new treatment strategies aimed at restoring developmental trajectories and reduce or prevent long-term disability.²⁷

key points

SELECTED BY

Dr Peter Saul

GP, Wrexham and associate GP Dean for North Wales

Autism is a disorder of social communication, originally described as a triad of impaired social interaction, communication, and imagination, associated with a rigid, repetitive pattern of behaviour. More recently the core deficit has been described as a lack of social instinct. The term autism spectrum disorder (ASD) includes autism, atypical autism and Asperger's syndrome. ASD was once believed to be relatively rare but is now thought to occur in about 1% of children.

The two primary diagnostic classification systems

currently in use are the ICD-10 and the DSM-IV. The ICD-10 is the most commonly used ASD classification system in the UK and forms the basis for the SIGN guideline. According to the NICE guidelines, diagnostic assessment should start within three months of referral. Information from all sources (i.e. medical history, parental concerns, clinical judgement) should be used to diagnose ASD based on ICD-10 or DSM-IV criteria. It is not advisable to rely on any one autism-specific diagnostic tool alone to diagnose ASD.

Diagnosis is based primarily on the recognition and interpretation of behavioural symptoms. The Checklist for Autism in Toddlers (CHAT) or the modified version (M-CHAT) are simple checklists that can be used for the assessment of young children in primary care by GPs or health visitors when ASD is suspected. They can identify clinical features indicative of increased risk but should not be used to rule out ASD. The M-CHAT is a promising instrument for the early detection of ASD, but it fails to recognise 15% of children with the condition.

There is a substantial genetic basis with strong heritability.

Approximately 70% of individuals with ASD also meet diagnostic criteria for at least one other mental and behavioural disorder. GPs should be aware of the importance of considering comorbid problems in children and young people with ASD.

The decision to refer should be made on the basis of signs

or symptoms. Children younger than three years should be referred to an autism team if there is regression in language or social skills. In children older than three, referral should be to a paediatrician or paediatric neurologist. Irrespective of any earlier assessments, re-referral for further ASD assessment should be considered at any age. ASD should not be ruled out because of: good eye contact; smiling and showing affection to family members; reported pretend play or apparently normal language development.

The majority of intensive behavioural programmes for ASD

are based on the principles of behaviour modification using applied behavioural analysis.

DIAGNOSTIC ASSESSMENT

There is no specific diagnostic test for ASD. Diagnosis is based primarily on the recognition and interpretation of behavioural symptoms.²⁸ According to the NICE guidelines, diagnostic assessment should start within three months of referral. Information from all sources (i.e. medical history, parental concerns, clinical judgement) should be used to diagnose ASD based on ICD-10 or DSM-IV criteria. It is not advisable to rely on any one autism-specific diagnostic tool alone to diagnose ASD.

Based on physical examination, clinical judgement and the child or young person's profile, genetic tests may be performed if there are specific dysmorphic features, congenital anomalies and/or evidence of intellectual disability. EEG may be useful if epilepsy is suspected.⁵ Parents should have an opportunity to discuss the risk of ASD occurring in siblings and future children.

The minimum age at which ASD can be reliably diagnosed remains uncertain. There is a wealth of evidence concerning the manifestations of ASD between the ages of one and three years.^{29,30} In children under two years typical ASD behaviours may not, however, always be evident. Some of the very early symptoms of autism include deficits in reciprocal social interaction and communication, repetitive behaviours, deficits in imitation, spontaneous and pretend play, and joint attention.³¹⁻³³ The possibility of diagnosis should not be ruled out in the absence of typical ASD behaviours.³⁴

Accuracy of assessment tools

There is a multiplicity of structured autism diagnostic assessment tools see box 1, above. Assessments of intellectual ability and/or receptive and expressive language also complement these assessment tools.⁵

The combination of ADI/ADI-R and ADOS is accurate in diagnosing autism in preschool children and children with an intellectual disability.

Overall, evidence supporting the use of autism-specific diagnostic tools, either individually or in combination, is poor, so the clinical benefits of using these tools remain uncertain. The NICE guidelines nevertheless acknowledged that both an autism-specific semi-structured interview and observation are effective in implementing a structured means of collating information to aid diagnostic assessment.

The Checklist for Autism in Toddlers (CHAT) or the modified version

Box 1

Assessment tools

Structured autism diagnostic assessment tools

- Autism Diagnostic Interview and Autism Diagnostic Interview – Revised (ADI/ADI-R)
- Developmental, Dimensional and Diagnostic Interview (3di)
- Diagnostic Interview for Social and Communication Disorders (DISCO)
- Autism Diagnostic Observation Schedule (ADOS)

Checklists

- Checklist for Autism in Toddlers (CHAT)
- Modified version of CHAT (M-CHAT)

(M-CHAT) are simple checklists that can be used for the assessment of young children in primary care by GPs or health visitors when ASD is suspected. They can identify clinical features indicative of increased risk but should not be used to rule out ASD.⁶ The M-CHAT is a promising instrument for the early detection of ASD.³⁵ It fails to recognise 15% of children with the condition.³⁶

INTERVENTIONS

A comprehensive evaluation of speech and language and communication skills of all children and young people with ASD should be carried out in order to inform intervention. An individual's level of comprehension may be at a lower developmental level than that suggested by their expressive language skills. There is often a range of further impairments in intellectual, neuropsychological and adaptive skills and so occupational therapy and physiotherapy assessments should be considered where relevant.⁶

The majority of intensive behavioural programmes for ASD are based on the principles of behaviour modification using applied behavioural analysis (ABA). These programmes can be very intensive and expensive, usually involving 20 to 40 hours of intervention per week for preschool children. The Lovaas programme^{37,38} is the best known ABA. Further interventions are designed to support social communication and interaction directly, e.g. visual timetabling, operationalising through short stories or the use of speech bubbles or cartoons. Published evidence for these interventions is generally weak.^{39,40}

»

SPECIAL REPORT

AUTISM SPECTRUM DISORDERS

Various pharmacological interventions are discussed in the SIGN guidelines.

● Risperidone can be useful for short-term treatment of significant aggression, tantrums or self-injury in children with ASD, but weight should be monitored regularly.

● Methylphenidate may be considered for treatment of attention difficulties or hyperactivity.

● Melatonin may also be considered for treatment of sleep problems which have persisted despite behavioural interventions.

● A single trial found statistically significant but relatively small clinical benefit from fluoxetine on repetitive behaviours in children and young people with ASD. Side effects were similar to placebo.⁴¹

● Naltrexone and secretin appear ineffective and are not recommended.

A thorough Cochrane systematic review looked at biomedical and nutritional interventions and was unable to identify an evidence base for or against casein and gluten exclusion diets.⁴² Gastrointestinal symptoms, which are common in children and young people with ASD, should be managed in the same way as in children and young people without ASD.

CONCLUSION

When making a referral decision in a suspected case of ASD, a broad range of issues need to be considered. These include parental concerns, the impact of symptoms on the family, educational factors, medical conditions with an increased risk of ASD, and the likelihood of an alternative diagnosis.

Account needs to be taken of the severity and duration of the signs and/or symptoms and the degree to which these are manifest in various settings (i.e. home and school).

GPs can help support the child and family before and after diagnosis is confirmed. Clear information on support available locally should be provided, and opportunities given for the family to ask questions. Contact information for local and national support organisations is likely to be of particular value, as well as information about organisations that can provide advice on welfare benefits, educational support, social care and preparing for the future. Children and their parents should routinely receive written information, perhaps including copies of the letters sent to the various professionals who have been asked to assess their child.

Finally, encouragement should be given to children, young people and their parents to continue to learn about ASD.

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Useful information

NICE CG128: Autism: Recognition, referral and diagnosis of children and young people on the autism spectrum
www.nice.org.uk

SIGN 98: Assessment, diagnosis and clinical interventions for children and young people with autism spectrum disorders
www.sign.ac.uk

Web-based learning resource for primary care practitioners
www.nes.scot.nhs.uk/asd

The National Autistic Society
www.autism.org.uk

'Transition: getting it right for young people'
www.dh.gov.uk

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