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Improving the recognition of autism in children and adults

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What are the symptoms and signs of autism?

How should diagnosis be confirmed?

What are the management approaches?

AUTISM IS A SET OF NEURODEVELOPMENTAL CONDITIONS CHARACTERISED BY difficulties in social communication, alongside unusually narrow interests and strongly repetitive behaviour.[1] Autism occurs in about 1% of people, which may be as many as 605,000 individuals in the UK. Autism has a male:female ratio of 3:1.

Autism covers a wide spectrum across the dimensions of social communication, repetitive and stereotyped behaviours as well as other non-clinical and cognitive features. Autistic people have a strong preference for predictability that can also manifest as a difficulty in adjusting to unexpected change. The autistic learning or cognitive style tends towards a preference for detail rather than seeing the bigger picture, a preference for factual and well specified precision rather than ambiguity, and a systematic, step by step logic rather than making big leaps. Sensory hypersensitivity is also very common, as are difficulties with executive function.

Individuals with autism can function well in certain environments, where there are fewer demands to multitask and factual information and pattern recognition are required. However, they may not function well in other kinds of environments, particularly highly social environments, or situations characterised by rapid and unpredictable change. In this sense it is all about the fit between the person and their environment, and the disability can be exacerbated or reduced depending on the environmental fit or environmental adjustments and modifications. Autism is best understood as a form of 'neurodiversity' i.e. where the brain is wired differently.

Individuals with autism may have one or more co-occurring conditions such as anxiety, depression, language delay, learning difficulties, gastrointestinal pain, self-injury, epilepsy, and attention deficit hyperactivity disorder (ADHD). These may be secondary to the autism (e.g. poor mental health may be the result of lack of support and/or negative life events such as bullying at school), while others may reflect co-occurring partly genetic conditions.

Identification of autism can be difficult because of its heterogeneity. The condition may be diagnosed in early childhood, or more commonly later in childhood or adolescence or even in adulthood (by which time the individual may have had a host of negative life experiences leading to worse mental health. High rates of suicidality in autistic people have been reported.[2] This highlights the need for »

appropriate service planning and support to reduce risk in this group. NICE guidelines aim to improve recognition of autism in both childhood and adulthood.[3,4]

CAUSAL FACTORS

Autism has high heritability,[5] with estimates of 40-50% of the variance in risk for autism being due to genetic factors. This has been established through twin studies, in monozygotic twins both individuals are more likely to be affected where one of the twins has autism than is the case with dizygotic twins. Having an older sibling with autism increases the likelihood of autism in the next sibling by 20-32.2%.[6,7,8,9,10,11] Around 20% of parents of children with autism also show the broader autism phenotype, with a high number of autistic traits.[12,13,14] The genetics of autism suggest it is polygenic and includes both rare genetic mutations and combinations of specific common genetic variants that we all carry.

Environmental and other factors that increase the likelihood of autism in the child include: advanced paternal or maternal reproductive age,[15,16,17] polycystic ovary syndrome in the mother,[18,19] pre-eclampsia,[20] maternal infection during pregnancy,[21] maternal gestational diabetes,[22,23] high maternal BMI at age 18, low neonatal birthweight/gestational age, low neonatal Apgar score,[24] maternal use of sodium valproate in pregnancy,[25] and having an intellectual disability.[26]

NICE (CG 128) recommends that, when considering referral to the autism team, account should be taken of factors associated with increased prevalence of autism. GPs need to consider the family history and to take a detailed obstetric history when considering the possibility of autism.[3]

SYMPTOMS

Autism symptoms generally manifest in early childhood yet many individuals experience delays in diagnosis and accessing specialist services. Signs and symptoms of autism in young children include: deficits or delays in the emergence of joint attention (e.g. looking at what a parent is looking at, or pointing at objects to share interest) and pretend play, taking an atypical perspective, reduced reciprocal affective behaviour, decreased response to hearing one's own name, reduced imitation, delayed verbal and nonverbal communication, motor delay, unusually repetitive behaviours, atypical visuomotor exploration, inflexibility in disengaging visual attention, and extreme variation in temperament.

However, symptom presentations vary immensely. In older children, signs of autism may include 'talking at' others rather than a dialogue; long-standing difficulties in reciprocal social communication and interaction; reduced or absent understanding of friendship; social isolation and apparent preference for being alone; poorly integrated gestures, facial expressions, body orientation, and eye contact, highly repetitive behaviours or rituals that negatively affect daily activities; over- or under-reaction to sensory stimuli, for example textures, sounds, smells. They may have an unusual profile of skills and deficits (for example, social or motor coordination skills being poorly developed, while particular areas of knowledge, reading or vocabulary skills are advanced for chronological or mental age).

'Autism symptoms generally manifest in early childhood yet many individuals experience delays in diagnosis'

Older people presenting with possible autism may have previously camouflaged their autism or have been living in a supportive environment, making identification difficult especially in articulate individuals. Autistic girls and women may be missed or experience late diagnosis because of camouflaging their autism (perhaps because of greater social expectations on females to be sociable and communicative). Women may have been misdiagnosed with other mental health conditions, such as personality disorders or eating disorders,[27,28] anxiety and depression.[29,30]

SCREENING TOOLS

The GP's role is vital in timely and accurate recognition of autistic traits in patients. GPs need to be able to identify patients who require a specialist diagnostic assessment and make appropriate referrals. However, in a survey 80% of GPs stated that they required guidance to identify people who may have autism.[31] Parents may raise concerns about their child by 18 months but there is frequently a significant delay in arriving at a diagnosis. Delays in referring patients for a specialist diagnostic assessment can result in late diagnosis and delays in receiving appropriate intervention, help and support.

GPs may be the first point of contact for parents who may be concerned that their child has autism, as well as for adults who feel that they themselves may be autistic. Brief tools are useful for GPs to identify red flags to alert them to consider a referral for a diagnostic assessment.

The Autism Spectrum Quotient (AQ)[32] was first developed to measure autistic traits in adults. The AQ has excellent sensitivity, specificity and positive predictive value (all > 0.80) in a clinically referred sample.[33] There are now also child and adolescent versions of the AQ,[34,35] along with an early developmental version called the Quantitative Checklist for Autism in Toddlers (Q-CHAT).[36]

Short versions have been developed and tested in large samples identifying the ten items that best differentiated people with and without autism,[37] independently validated by Booth and colleagues.[38] The AQ-10 is recommended in the NICE guideline (CG142) on autism in adults.[4] These tools have the potential to help GPs make decisions in real clinic time about whether to make a referral for an assessment. Although a positive score may support a decision to refer a patient a negative score does not rule out autism. However, NICE also recommends that patients should be referred if autism is suspected based on clinical judgement.

To date, they have not been widely implemented and there is a need for more research to evaluate their use prospectively in primary care since most of the research comes from population or clinical studies where patients have already been referred.

Box 1 (opposite) contains the Q-CHAT-10, to use with toddlers, box 2 (p14) the AQ-Child-10 for children age 4 to 11 years old, box 3 (p14) the AQ-Adol-10 for adolescents aged 12-15 years, and Box 4 (p15) the AQ-Adult-10 for those aged 16 years and older. The first three are parent reported, and the last one is self reported.

NICE recommends that assessment should be considered if an adult shows one or more of: persistent difficulties in social interaction, persistent difficulties in social communication, and stereotypic (rigid and repetitive) behaviours, resistance to change or restricted interests and at least one of the following: problems in obtaining or »

Box 1

Q-CHAT-10 Quantitative Checklist for Autism in Toddlers

A quick referral guide for parents to complete about their toddler (18 – 24 months) with concerns about autism.

For each item, please circle the response which best applies to your child:

		A	B	C	D	E
1	Does your child look at you when you call his/her name?	Always	Usually	Sometimes	Rarely	Never
2	How easy is it for you to get eye contact with your child?	Very easy	Quite easy	Quite difficult	Very difficult	Impossible
3	Does your child point to indicate that s/he wants something? (e.g. a toy that is out of reach)	Many times a day	A few times a day	A few times a week	Less than once a week	Never
4	Does your child point to share interest with you? (e.g. pointing at an interesting sight)	Many times a day	A few times a day	A few times a week	Less than once a week	Never
5	Does your child pretend? (e.g. care for dolls, talk on a toy phone)	Many times a day	A few times a day	A few times a week	Less than once a week	Never
6	Does your child follow where you're looking?	Many times a day	A few times a day	A few times a week	Less than once a week	Never
7	If you or someone else in the family is visibly upset, does your child show signs of wanting to comfort them? (e.g. stroking hair, hugging them)	Always	Usually	Sometimes	Rarely	Never
8	Would you describe your child's first words as:	Very typical	Quite typical	Slightly unusual	Very unusual	My child doesn't speak
9	Does your child use simple gestures? (e.g. wave goodbye)	Many times a day	A few times a day	A few times a week	Less than once a week	Never
10	Does your child stare at nothing with no apparent purpose?	Many times a day	A few times a day	A few times a week	Less than once a week	Never

SCORING: For questions 1-9: if you circle an answer in columns C, D or E, score 1 point per question. For question 10: if you circle an answer in columns A, B or C, score 1 point. Add points together for all ten questions. If your child scores more than 3 out of 10, the health professional may consider referring your child for a multi-disciplinary assessment.

Key reference: Allison C, Auyeung B, and Baron-Cohen S, (2012) *Journal of the American Academy of Child and Adolescent Psychiatry* 51(2):202-12.

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AQ-10 (Child Version)

Autism Spectrum Quotient (AQ)

Box 2

A quick referral guide for parents to complete about a child aged 4-11 years with suspected autism who does not have a learning disability.

Please tick one option per question only:		**Definitely Agree**	**Slightly Agree**	**Slightly Disagree**	**Definitely Disagree**
1	S/he often notices small sounds when others do not				
2	S/he usually concentrates more on the whole picture, rather than the small details				
3	In a social group, s/he can easily keep track of several different people's conversations				
4	S/he finds it easy to go back and forth between different activities				
5	S/he doesn't know how to keep a conversation going with his/her peers				
6	S/he is good at social chit-chat				
7	When s/he is read a story, s/he finds it difficult to work out the character's intentions or feelings				
8	When s/he was in preschool, s/he used to enjoy playing games involving pretending with other children				
9	S/he finds it easy to work out what someone is thinking or feeling just by looking at their face				
10	S/he finds it hard to make new friends				

SCORING: Only 1 point can be scored for each question. *Score 1 point for Definitely or Slightly Agree on each of items 1, 5, 7 and 10. Score 1 point for Definitely or Slightly Disagree on each of items 2, 3, 4, 6, 8 and 9.* If the individual scores **more than 6 out of 10**, consider referring them for a specialist diagnostic assessment.
USE: This is the child version of the test recommended in the NICE clinical guideline CG142. www.nice.org.uk/CG142

Key reference: Allison C, Auyeung B, and Baron-Cohen S, (2012) *Journal of the American Academy of Child and Adolescent Psychiatry* 51(2):202-12.

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AQ-10 (Adolescent Version)

Autism Spectrum Quotient (AQ)

Box 3

A quick referral guide for parents to complete about a teenager aged 12-15 years old with suspected autism who does not have a learning disability.

Please tick one option per question only:		**Definitely Agree**	**Slightly Agree**	**Slightly Disagree**	**Definitely Disagree**
1	S/he notices patterns in things all the time				
2	S/he usually concentrates more on the whole picture, rather than the small details				
3	In a social group, s/he can easily keep track of several different people's conversations				
4	If there is an interruption, s/he can switch back to what s/he was doing very quickly				
5	S/he frequently finds that s/he doesn't know how to keep a conversation going				
6	S/he is good at social chit-chat				
7	When s/he was younger, s/he used to enjoy playing games involving pretending with other children				
8	S/he finds it difficult to imagine what it would be like to be someone else				
9	S/he finds social situations easy				
10	S/he finds it hard to make new friends				

SCORING: Only 1 point can be scored for each question. *Score 1 point for Definitely or Slightly Agree on each of items 1, 5, 8 and 10. Score 1 point for Definitely or Slightly Disagree on each of items 2, 3, 4, 6, 7 and 9.* If the individual scores **more than 6 out of 10**, consider referring them for a specialist diagnostic assessment.
USE: This is the adolescent version of the test recommended in the NICE clinical guideline CG142. www.nice.org.uk/CG142

Key reference: Allison C, Auyeung B, and Baron-Cohen S, (2012) *Journal of the American Academy of Child and Adolescent Psychiatry* 51(2):202-12.

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sustaining employment or education, difficulties in initiating or sustaining social relationships, previous or current contact with mental health or learning disability services, a history of a neurodevelopmental condition (including learning disabilities and ADHD) or a psychiatric condition.

The potential health benefits of screening for patients include faster referral to specialist diagnostic services, which may help to alleviate uncertainty, as well as secondary symptoms such as depression and anxiety.[39,40] Patients with undiagnosed autism often find medical examinations and procedures make them anxious, which may be challenging for the clinician,[41] and frustrating for the patients.

CONFIRMING DIAGNOSIS

Diagnostic assessment should be multidisciplinary. NICE recommends that detailed questions about the caregiver's concerns and, if appropriate, the patient's concerns, be taken into consideration. A developmental history should be taken, using an autism-specific tool to gather information on developmental and behavioural features consistent with the diagnostic criteria. There should be: an assessment (through interaction with, and observation of, the child or young person) of social and communication skills and behaviours; a medical history; a physical examination; consideration of any differential diagnosis; systematic assessment for conditions that may coexist with autism; development of a profile of the child's or young person's strengths, skills, impairments and needs that can be used to create a needs-based management plan. The family and educational context also need to be taken into account.

'Clinicians should not rely on any autism-specific diagnostic tool alone to diagnose autism'

NICE recommends that information from all sources, together with clinical judgment, should be used to diagnose autism based on ICD-10 or DSM-5 criteria. NICE also states that clinicians should not rely on any autism-specific diagnostic tool alone to diagnose autism. A revised edition (ICD-11) is expected in 2019 and will closely align with the latest edition of the American Diagnostic and Statistical Manual (DSM-5).

Assessment of adults can be challenging since the diagnosis relies on an informant to provide information about the person's early development. This can be particularly problematic when parents have died and there is nobody else to provide this early developmental history.

MANAGEMENT APPROACHES

Early intensive behavioural intervention (EIBI) is the most studied treatment model for autistic children. This is a variation of the applied behaviour analysis (ABA) approach and focuses on the premise that early intervention will be successful if: it occurs early (toddler/pre-school age), is intensive

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AQ-10

Autism Spectrum Quotient (AQ)

Box 4

A quick referral guide for adults with suspected autism who do not have a learning disability.

	Please tick one option per question only:	**Definitely Agree**	**Slightly Agree**	**Slightly Disagree**	**Definitely Disagree**
1	I often notice small sounds when others do not				
2	I usually concentrate more on the whole picture, rather than the small details				
3	I find it easy to do more than one thing at once				
4	If there is an interruption, I can switch back to what I was doing very quickly				
5	I find it easy to 'read between the lines' when someone is talking to me				
6	I know how to tell if someone listening to me is getting bored				
7	When I'm reading a story I find it difficult to work out the characters' intentions				
8	I like to collect information about categories of things (e.g. types of car, types of bird, types of train, types of plant etc)				
9	I find it easy to work out what someone is thinking or feeling just by looking at their face				
10	I find it difficult to work out people's intentions				

SCORING: Only 1 point can be scored for each question. *Score 1 point for Definitely or Slightly Agree on each of items 1, 7, 8, and 10. Score 1 point for Definitely or Slightly Disagree on each of items 2, 3, 4, 5, 6, and 9.* If the individual scores **more than 6 out of 10**, consider referring them for a specialist diagnostic assessment.

This test is recommended in 'Autism: recognition, referral, diagnosis and management of adults on the autism spectrum' (NICE clinical guideline CG142). www.nice.org.uk/CG142

Key reference: Allison C, Auyeung B, and Baron-Cohen S, (2012) *Journal of the American Academy of Child and Adolescent Psychiatry* 51(2):202-12.

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(> 20 hours/week), and long-term (> 2 years). EIBI includes the use of discrete trial training, is delivered one to one by an adult therapist, has individualised goal setting guided by typical developmental sequences and involves parents as co-therapists. The most recent Cochrane review[42] found there to be weak evidence that EIBI may be an effective behavioural treatment for some autistic children. The studies were small and not of the optimum randomised design.

Alternatives to this approach include the Early Start Denver Model (ESDM) and Joint Attention Symbolic Play Engagement and Regulation (JASPER). A recent systematic review of 48 randomised controlled trials (RCTs)[43] found positive significant outcomes in many trials (even at long-term follow-up beyond two years).[44,45] However, these studies show small effect sizes, wide confidence intervals and there remain major methodological challenges for future trials.

A parent-mediated approach is another alternative in which parents are trained by professionals to deliver support to their children. However, a Cochrane review[46] of 17 RCTs found no significant evidence of gain in children's language and communication, initiation in parent-child interaction, adaptive behaviour, or parents' stress. However, there were positive changes in parent-child interaction pattern on shared attention and parent synchrony as well as evident reduction of children's autism characteristics.

There is now a body of evidence that indicates targeted intervention,[47,48] designed to address a relatively narrow but pivotal goal or set of skills, in contrast to the comprehensiveness of EIBI, can be helpful to autistic children.[49] For example, interventions that focus on enhancing social skills and peer relationships report potential positive outcomes in improving overall social competence and quality of friendships and reducing loneliness, with a recent meta-analysis indicating overall improvement in social functioning.[50] Other kinds of interventions include supported employment schemes, supported living schemes, autism self-advocacy or support from an autism advocate, mentoring, and peer support groups. In the UK, the National Autistic Society website (see Useful information box, p16) is a useful resource for identifying local agencies and services.

CONCLUSIONS

The increasing prevalence of autism over time, (previously rare and now very common, due to better awareness and changes in diagnostic criteria), is putting pressure on specialist diagnostic services to offer assessment and post-diagnostic support, which in many areas remains insufficient. GPs are often the gatekeepers to the referral for a diagnostic assessment, but there is a need for more and better training especially about the more subtle manifestations of autism.

Providing GPs with screening tools may enable them to make informed decisions about whether the patient presenting to them is showing signs of autism which might warrant further investigation. Identification is the first step towards autistic people accessing the support they need to lead fulfilling and rewarding lives.

Competing interests: None

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key points

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Autism is a set of neurodevelopmental conditions characterised by difficulties in social communication, alongside unusually narrow interests and strongly repetitive behaviour. Autism occurs in about 1% of people, which may be as many as 605,000 individuals in the UK. Individuals with autism can function well in certain environments, where there are fewer demands to multitask and factual information and pattern recognition are required, but they may not function well in highly social environments, or situations characterised by rapid and unpredictable change.

Autism has high heritability. Having an older sibling with autism increases the likelihood of autism in the next sibling by 20-32.2% and around 20% of parents of children with autism have a high number of autistic traits. NICE recommends that, when considering referral to the autism team, account should be taken of factors associated with increased prevalence of autism. GPs need to consider the family history and to take a detailed obstetric history when considering the possibility of autism. Screening tools may enable GPs to make informed decisions about whether the patient presenting to them is showing signs of autism which might warrant further investigation. However, the guideline also recommends that patients should be referred if autism is suspected based on clinical judgement.

GPs may be the first point of contact for parents who may be concerned that their child has autism, as well as for adults who feel that they themselves may be autistic. Autism symptoms generally manifest in early childhood. Signs and symptoms of autism in young children include: deficits or delays in the emergence of joint attention and pretend play, reduced reciprocal affective behaviour, decreased response to hearing one's own name, reduced imitation, delayed verbal and nonverbal communication, motor delay, and unusually repetitive behaviours. In older children, social or motor coordination skills may be poorly developed, while particular areas of knowledge, reading or vocabulary are advanced for chronological or mental age.

Diagnostic assessment should be multidisciplinary. NICE recommends that detailed questions about the caregiver's concerns and, if appropriate, the patient's concerns, be taken into consideration. Early intensive behavioural intervention (EIBI) is the most studied treatment model for autistic children, it is intensive (> 20 hours/week), and long-term (> 2 years). There is now a body of evidence that indicates targeted intervention, designed to address a relatively narrow but pivotal goal or set of skills, in contrast to the comprehensiveness of EIBI, can be helpful to autistic children.

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Useful information

NICE. CG 128. Autism spectrum disorder in under 19s: recognition, referral and diagnosis
www.nice.org.uk/guidance/cg128

NICE. CG142. Autism spectrum disorder in adults: diagnosis and management
www.nice.org.uk/guidance/cg142

The National Autistic Society
www.autism.org.uk

The Autism Research Trust
www.autismresearchtrust.org

Autistica
www.autistica.org

Online versions of screening tools
The full adult AQ
www.wired.com/2001/12/aqtest

The full Q-CHAT
https://transformingautism.org/q-chat/