

Autism in children: improving screening, diagnosis and support

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Although symptoms of autism often first appear before the age of three years, many children experience significant delays in identification, diagnosis and access to specialist services. This article discusses the signs and symptoms of autism in children, screening and referral, and the management options available.

Autism is a spectrum of lifelong, neurodevelopmental conditions characterised by social communication difficulties, markedly restricted interests and repetitive behaviour,¹ alongside difficulties adjusting to unexpected changes and sensory hypersensitivity. Current estimates suggest that autism affects 1–2% of the population, though the prevalence of autism continues to rise.² Autism affects a wide spectrum of individuals in terms of both symptoms and IQ; traditionally it has been thought that approximately 45% of autistic individuals may be non-verbal or have intellectual disability (ID), while the other 55% are in the average or higher range of IQ.² However, some recent studies suggest these rates of co-occurring conditions are changing, with ID or language delay only being seen in 20–30% of autistic individuals.^{3–5} Autism also tends to be more common in males, with a ratio of 3:1 males to females.² However, there is also evidence that autistic females may be underdiagnosed or have a later diagnosis due to differences in symptom presentation and camouflaging of their symptoms.²

Symptom presentation and severity varies significantly, and autism is also characterised by particular non-clinical and cognitive features. Differences in sensory experience are very common, with many people experiencing hyper- and hypo-sensitivities across any of the sensory modalities. In terms of cognitive profile, autistic individuals tend to prefer factual, logical and precise information; they tend to focus on details and prefer predictability, which can also manifest as difficulty adjusting to change. These differences in sensory experience and cognitive profile affect the way in which autistic individuals interact with their environment, and can have a significant impact on their quality of life. Situations that are highly social, ambiguous, unpredictable or provide great sensory stimulation may be overwhelming; however, appropriate adjustments and modifications of the environment that may seem small can significantly reduce distress and secondary symptoms, such as anxiety and depression.

Over 70% of autistic individuals have co-morbid mental and/or physical health conditions,² including anxiety, schizophrenia, depression, suicide/self-harming behaviour, attention deficit hyperactivity disorder (ADHD), epilepsy and sleep disturbances, as well as gastrointestinal, autoimmune, allergic and thyroid conditions.⁶ In addition, autistic individuals are at higher risk of obesity and type 2 diabetes, which may have further implications for other health risks.⁶ We do not know if this higher health burden is explained by genetic factors, lifestyle choices, lack of social support and/or vulnerability to adverse life experiences (eg bullying at school).

What causes autism?


There is no single known cause for autism. We know that it is a highly heritable condition, with 64–91% of risk being associated with genetic factors.^{7,8} About 5% of autism cases are syndromic, meaning that a single gene mutation causes a particular set of symptoms and phenotype, eg individuals with 15q11.2 duplication syndrome tend to show hypotonia, motor delays, intellectual disability, autism and epilepsy.⁹ However, in most cases autism is polygenic, meaning that many genes contribute to its aetiology. Genetic mutations include contributions from both rare and common genetic variants.

Many different types of evidence suggest that autism is highly heritable, including evidence from twins and family units. Studies show that having an autistic child increases the risk that the next child born will be autistic.¹⁰ In addition, approximately 20% of parents of autistic children have the 'broader autism phenotype',¹¹ a term used to describe individuals who show a constellation of subtle language, cognitive, social and personality characteristics that parallel the defining autism characteristics, including a higher than average number of autistic traits without necessarily meeting criteria for a formal diagnosis.

Some environmental factors may play a role in the development of autism. Risks related to pregnancy (eg pregnancy complications, maternal gestational diabetes and labour complications), as well as certain parental factors (eg increased paternal or maternal age at birth)¹² have been shown to increase the likelihood of autism in children. However, each of these individual risks is small, and it is likely that most autism cases result from a combination of both genetic and environmental factors.

What are the signs and symptoms of autism in children?

As autism is a lifelong condition that develops prenatally, symptoms often first appear before three years of age. However, many individuals experience significant delays in identification, diagnosis and access to specialist support services. Common early signs in childhood include behavioural and motor differences, including delays or deficits in visual attention, sharing attention (following cues from parents to look at or point to objects to share interest), imaginative play and imitation, as well as difficulties with both verbal and non-verbal communication. Children may also acquire unusual



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AQ-10 (Child Version)
Autism Spectrum Quotient (AQ)

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 6 or above, 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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Figure 1. Shorter, 10-item version of the Autism Spectrum Quotient (AQ-10) for use in children

repetitive behaviours and exhibit extreme temperament, eg being very shy, anxious or prone to anger when self-chosen routines are disrupted. Finally, some autistic children experience delays in acquisition of motor skills and display atypical visuomotor exploration, eg showing a preference for tiny details. However, early signs vary immensely across children and age groups.

In older children, symptoms of autism may manifest as social isolation (and even preference for being alone); challenges with reciprocal conversation (talking 'at' others rather than engaging in conversation in a 'to-and-fro' manner); poor understanding of social relationships, particularly friendship; atypical gestures, facial expressions and bodily co-ordination; atypical eye contact; differences in sensory processing resulting in over- or under-reaction to sensory stimuli; highly repetitive behaviours or rituals that impair daily function; and narrow areas of extreme interest or obsession. Finally, autistic children and adolescents may show inconsistent profiles in motor and cognitive skill acquisition, eg development in specific areas of knowledge or academic skills but deficits in motor control or social engagement.

Differences in social communication skills have been associated with decreased skills in theory of mind, which is the ability to draw inferences about what others are thinking (including their beliefs, desires, intentions and emotions). Across age groups, autistic individuals have been shown to be worse at

tasks involving theory of mind, including measures of both cognitive empathy (ie the ability to identify another person's emotional state) and affective empathy (ie the ability to respond to another person's emotional state appropriately).¹³

What tools exist for screening children for autism?


The Autism Spectrum Quotient (AQ)¹⁴ was originally developed to measure autistic traits in adults. In a clinically referred sample, the AQ showed excellent sensitivity, specificity and positive predictive value (all >0.80).¹⁵ Both child and adolescent versions of the AQ^{16,17} have been developed, as well as a version for young children in early development called the Quantitative Checklist for Autism in Toddlers (Q-CHAT).¹⁸ These tests have been translated into various languages and shorter, 10-item versions (AQ-10 and Q-CHAT-10) have also been created. The English versions of AQ-10 for children and adolescents are shown in Figures 1 and 2 and Q-CHAT-10 in Figure 3.

These tools should not be used to diagnose autism, but rather to serve as a screening mechanism for referral. This is mainly because of a lack of research into their use in primary care settings, as most studies have been conducted on a clinical population of patients who have already been referred. Furthermore, while a positive score on the AQ or Q-CHAT may support referral, a failure to meet the screening cut-off point does not rule out autism. Thus, these tools can be useful in helping GPs to assess the likelihood of autism in a child, but clinical judgement should take priority when considering referrals for a developmental assessment.

Even with these limitations in mind, the AQ and Q-CHAT can be an effective way to identify potential autism cases and mitigate the significant delays experienced by so many children and families in receiving diagnoses and support services. Autistic individuals may receive a diagnosis at any age from toddlerhood to adulthood, and differences in age of diagnosis typically reflect how soon parents seek help, as well as how sensitive healthcare professionals are to a possible diagnosis. In many cases, the GP may be the first person to whom parents express concerns that their child may be showing atypical behaviour or even that they suspect autism. It is therefore important that GPs are knowledgeable about autism and are able to appropriately refer children to specialist services efficiently and effectively.

Clinical guidelines for autism and confirming a diagnosis

It is important that the diagnostic assessment is multidisciplinary and robust. The NICE guideline on autism spectrum disorders in under 19s (CG128)¹⁹ recommends that concerns of both the parents and patient (if appropriate) are taken into consideration, along with clinical judgement, to diagnose autism based on ICD-11 or DSM-5 criteria. Though specific diagnostic recommendations for autism have changed significantly in recent years, both ICD-11²⁰ and DSM-5¹ highlight social communication difficulties, as well as restricted interests and repetitive behaviours as the core features of autism. In



AQ-10 (Adolescent Version)

Autism Spectrum Quotient (AQ)

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 **6 or above**, 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

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Figure 2. Shorter, 10-item version of the Autism Spectrum Quotient (AQ-10) for use in adolescents

addition, ICD-11 and DSM-5 mention atypical sensory processing or sensitivities and possible co-morbidity with intellectual disability, both of which should be taken into account when making assessments.

To make or confirm a diagnosis, NICE states that clinicians should take a complete developmental history using an autism-specific tool to assess developmental and behavioural features associated with autism. In addition, clinicians should perform a wide-ranging clinical assessment, including evaluation of social communication skills and behaviours (by interaction with or observation of the child or adolescent), medical history, physical examination, consideration of differential diagnoses, and consideration of possible co-morbidities. Furthermore, as certain pregnancy and parental factors have been associated with risk of autism, NICE recommends taking full obstetric and family medical histories when making or confirming diagnostic assessments.

Management and support

Once a diagnosis is established and confirmed, a needs-based management plan should be implemented that takes into account the child or adolescent's impairments as well as their strengths and skills. It should also take their family and educational situation into account so that the plan can effectively improve the patient's quality of life.

There are various options for behavioural interventions,

particularly for young children. The most studied intervention is Early Intensive Behavioural Intervention (EIBI), which suggests that intervention will be effective if it is early, intensive and long term. This intervention follows the same principles as Applied Behaviour Analysis (ABA) and requires one-on-one attention from a trained therapist for 20 or more hours per week for two or more years and individualised goal setting with parents as co-therapists. Access to EIBI and ABA through the NHS and other publicly sector services may be available, depending on your area. If the interventions are available, waiting times for interventions are typically very long.

There are many other options for interventions. Some interventions focus on naturalistic play to boost social communication and emotional skills, including the Early Start Denver Model (ESDM) and Joint Attention Symbolic Play Engagement and Regulation (JASPER), while others involve peer or parent training. Studies show that these interventions may only have low to moderate effects or may only affect functioning in one area,^{2,21} yet even moderate improvements in skills and behaviours can improve the child or adolescent's quality of life. Finally, some autistic individuals find that participation in a peer support group or mentoring scheme is useful, as it connects them with a social network of individuals who may have shared experience. In the UK, the National Autistic Society helps individuals identify local support services and agencies.

Prescribing

Autism is now understood as a type of 'neurodiversity', in which the brain is simply structured or wired differently. As such, there are no recommended pharmacological treatments for autism and none that improve social-communication difficulties. There is much debate both in the autistic community and among scientists as to whether pharmacological treatments of any kind are wanted or needed. In some studies, antipsychotics (eg risperidone, aripiprazole) and SSRIs (eg citalopram, escitalopram, fluoxetine) have been used to manage challenging and repetitive behaviours in autism; however, caution should be used in prescribing these medications as a significant risk of adverse reactions has also been reported.² For this reason, psychological interventions, such as cognitive behavioural therapy (CBT, particularly for symptoms of anxiety, depression and OCD, which are common in autism), as well as environmental adjustments (eg finding a more autism-friendly school, work, home and/or leisure environment), should always be considered when creating a management plan and will often precede discussions about medication.

Co-morbidity or associated difficulties

Autism frequently co-occurs with other conditions that may benefit from pharmacological treatments, including epilepsy, depression, anxiety, OCD and ADHD.⁶ Stimulants such as methylphenidate have been prescribed to manage symptoms of ADHD in autism, yet relatively little evidence of improvement exists while a significant risk of adverse effects has been reported.² Furthermore, autism frequently co-occurs with both mental and physical health conditions (see Figure 4), leading to

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 3 or above, the health professional may consider referring your child for a multi-disciplinary assessment.

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Figure 3. Shorter, 10-item version of the Quantitative Checklist for Autism in Toddlers (Q-CHAT-10)

complex health needs for autistic children, particularly for those with intellectual or learning disabilities. It is essential that GPs take care when making decisions about which drugs to prescribe for particular individuals.

Summary

Autism includes a wide spectrum of individuals with a range of symptoms and needs. Autistic individuals may also experience a variety of secondary physical or mental health conditions that should be managed carefully by their GP. As the prevalence of autism continues to rise (due to improved recognition and changes in diagnostic criteria), there is a greater need to provide timely diagnosis and additional support services. As such, GPs are also put under additional pressure to appropriately

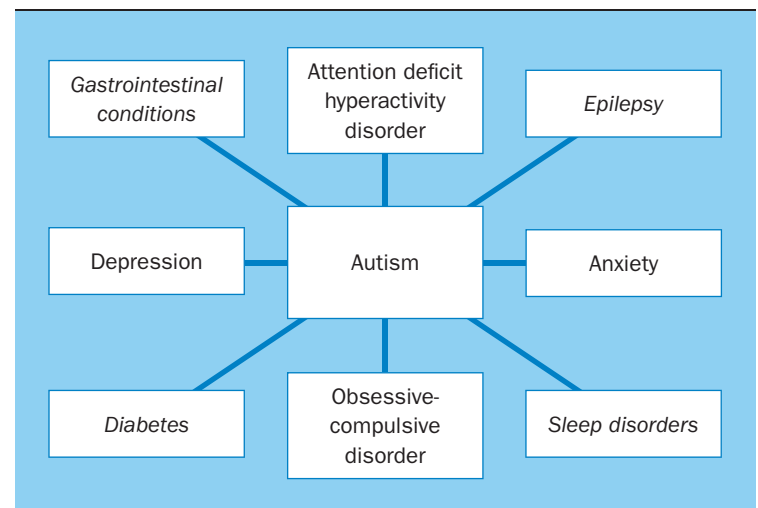


Figure 4. Common physical and mental co-morbidities of autism

refer children who warrant an autism assessment, in an efficient and effective manner.

Brief screening measures are available that could help front-line healthcare professionals make decisions about whether or not to refer a patient, though clinical reports and thorough history of symptoms should take precedence in these decisions. Improving diagnosis and effective management of symptoms and secondary physical and mental health conditions will help autistic children and adolescents to lead healthy and fulfilling lives.

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Declaration of interests

None to declare.

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