

Need to know - autistic spectrum conditions

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Autism researcher Professor Simon Baron-Cohen answers GP Dr Mandy Fry's questions on screening, treatment and the prognosis in autism.

Take-home points

- There are validated screening tools for autism that can be used in primary care
- CHAT can be used to screen for 'classic autism' in toddlers
- Q-CHAT is a more sensitive tool that can pick up milder cases such as Asperger's
- CAST can be used in older children to screen for Asperger's
- A therapy called applied behavioural analysis focuses on shaping target behaviours and has a solid evidence base
- Secondary depression is common in adults with Asperger's and needs to be recognised
- The increase in numbers of patients diagnosed with these conditions is largely because of the broadening of autism into a spectrum
- Not everyone on the autism spectrum needs a diagnosis – this should be reserved for those who need it to access help

1. When should GPs suspect an autistic spectrum condition? How can these children be distinguished from 'variations of normal'? Are there screening instruments available?

If a parent reports that their child is not talking by two years old, is not pointing at objects to share interest, is not looking up at other people's faces, is trying to impose strict order on their world by lining things up or throwing a tantrum if things are moved, and is rocking back and forth in repetitive movements, albeit in a self-soothing way, the GP can suspect autism.

The Checklist for Autism in Toddlers (CHAT) is a questionnaire that a GP or health visitor can use to screen for such cases of 'classic autism', though this

does not pick up milder autism spectrum conditions, such as Asperger syndrome.

Researchers have instead developed a more sensitive screening instrument for this, known as the Q-CHAT (or Quantitative Checklist for Autism in Toddlers). As the name implies, this screening instrument recognises that autism blends into 'normality' and the difference between

a child who needs a diagnosis of autism and one who does not may simply be whether the atypical behaviours are interfering with the child's life or not. If the parent or teacher is unconcerned, then the child may not need a diagnosis.

These screening tools – and CAST (see below) – can be downloaded from the Autism Research Centre website

2. What about an older child?

With an older child, Asperger's can be screened for using the Childhood Autism Spectrum Test (CAST), and here the risks of missing a child's diagnosis may be leaving a child unmonitored, and prey to bullying.

Children with Asperger's are noticeable because they prefer the company of adults to other children, they tend to 'lecture' in a monologue style rather than have a chat, their interests are narrow and unusual, and they may avoid the social areas of school such as the playground, preferring to spend break-time in the library.

The benefits of a diagnosis are that the child may be 'rescued' from dropping out of school, since many children with Asperger's feel like giving up if they are constantly marginalised, have no friends, are teased for being different, and if their learning style is at odds with the expectations of the mainstream classroom.

Such children are often talented in terms of their remarkable attention to detail, their ability to go into depth, their good memory, and their speed at detecting patterns in information – for instance, in maths. With the right support, these strengths can be harnessed to help their self-esteem and keep them on a path to higher education and eventual employment.

3. What are the principles of treatment of autism?

For children with classic autism, applied behavioural analysis (ABA), also known as behavioural therapy, is used widely, either in the classroom and/or at home. This focuses on shaping target behaviours such as sitting at a desk, speech, remaining on task or making eye contact. ABA grew out of the Lovaas approach to treatment.

The evidence for its efficacy is good. Critics regard it as shaping behaviour rather than necessarily changing the child's comprehension, and tying the child into a series of rules and rewards.

An alternative, newer approach is to work with the child's natural interests – their obsessions – and encourage social interaction around these interests.

For example, Lego therapy builds on the child's natural desire to build models, but puts children with autism into groups so they collaborate around an activity they enjoy.

Software such as Mindreading DVD or the Transporters DVD aim to teach emotion recognition by presenting the information in an autism-friendly way. This is done in the context of mechanical systems or in a rule-based, repetitive system, since people with autism tend to get anxious if information is ambiguous or cannot be checked and rechecked.

To find out whether a treatment has been evaluated, www.researchautism.net is a trusted source of information.

4. Sometimes it is siblings that find it hardest to cope with a child with autism. What are good sources of help available to the families of children with autism?

The best source of support for the family – not just siblings, but parents, grandparents, partners and involved professionals and carers – is the National Autistic Society. It has a free telephone help line and information about local support groups, both for people with a diagnosis, and for those in their circle who might also benefit.

5. Autism used to be a specific diagnosis. Now we hear much more about autistic spectrum conditions. Why the change and what does it mean?

Part of the change has been the discovery of different subgroups, the two key ones being classic autism and Asperger's. The former also involves language delay and can involve learning difficulties to varying degrees, whereas the latter does not.

Other subgroups include atypical autism, and PDD-NOS (pervasive developmental disorder, not otherwise specified). These vary in terms of how many of the diagnostic features have to be present, but all share the common thread of social and communication difficulties, alongside narrow interests and a strong love of routine or 'resistance to change'.

The idea of a spectrum is not just a spectrum of severity within those attending clinics, but a spectrum that runs right through the population. But a diagnosis is restricted to those who need it because the features are causing suffering to some degree.

The upshot of the change to thinking about a spectrum is that there is no longer such a clear point at which one can say 'this person needs a diagnosis' and 'this person does not', because of intermediate cases.

The other main consequence of broadening autism into a spectrum is that the number of cases being diagnosed has increased from the old days of four in 10,000, to the new figure of one in 100.

6. Are there any useful techniques that we as GPs could employ in trying to engage adults with Asperger syndrome to participate in healthcare?

Arguably the biggest risk is GPs not realising how adults with Asperger syndrome may become suicidal from the secondary depression, which is common.

This depression is associated with social isolation, the high levels of unemployment, the lack of close friends, the lack of a partner and the abuse that adults with Asperger's may experience on the bus or in the supermarket or in other everyday situations.

GPs need to give reassurance to adults with Asperger's that just because their disability is invisible, they recognise the patient is suffering underneath and will help them find the right support.

7. What are the current theories about the causes of autistic spectrum conditions?

The main theory is the genetic theory, based on the evidence from family and twin studies. And in the past few years, there has been an intensive effort by scientists to identify genes for autism.

Currently about 130 genes have been associated with autism in one way or another, and the puzzle for research is to figure out which of these are necessary to cause autism, what the functions of these genes are, and whether there are different causes (different gene subgroups) that can appear similar in their developmental outcome.

The relevant genes typically affect brain development and brain function, making autism a neurodevelopmental condition.

The parts of the brain that are developing atypically include those areas involved in understanding other people, particularly understanding other minds, leaving the child unable to empathise or put themselves into someone else's shoes with the same ease that a typically developing child can.

There is also evidence of early brain overgrowth, resulting in some children with autism having larger than average head size and brain size.

8. Autism is more common in males than females. Why is this?

Autism may be more common in males for three reasons: genetic, hormonal or diagnostic factors.

It may turn out the genes for autism are sex-linked or sex-limited in some way. This is not yet known. What is known is that autism runs in families and has a strong genetic component.

Twin studies suggest heritability is as high as 90%. It has been shown that foetal testosterone (FT) levels measured in the womb (in amniotic fluid, in

studies of women who had amniocentesis during pregnancy) are related to the child's social and language development postnatally.

Because male foetuses produce at least twice as much testosterone as females (since males produce it from their testes, whereas females only produce it from their adrenal glands), and because we know from animal studies that FT is known to affect brain development and later behaviour, this factor may turn out to play an important role in the cause of autism. FT may be both the result of genetic factors, and itself serve as an environmental factor (in the womb).

Finally diagnostic factors may play a role. It may be that girls are better at masking their autistic traits by imitation of a peer, just enough to not get noticed or raise concern among parents or teachers, so that girls are referred less often to clinics for diagnosis. Even when girls are seen in clinics, their learned social behaviour is judged to be good and their 'symptoms' may therefore be judged not to be severe enough to warrant a diagnosis.

Some evidence supporting this theory is that those females who do receive a diagnosis tend to have more severe traits, suggesting they may only come to a professional's attention when their behaviour is strikingly atypical.

9. What is the prognosis in autism?

Prognosis is best predicted by the child's initial level of language and IQ, and can be excellent, with the right support. In the AS subgroup, it is not unusual to meet patients who as adults have achieved high levels of educational and occupational success. Sheltered employment schemes and sheltered housing can also increase quality of life, even for the less able individuals on the autistic spectrum.

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Further reading

Simon Baron-Cohen. Autism and Asperger Syndrome: The Facts. Oxford University Press 2008