Identifying and managing autism in adults

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Autism may be underdiagnosed in women, as they may not show the expected markers of atypical social behaviour due to camouflaging of their autistic traits

The diagnosis of autism is frequently delayed, and it is important that GPs can quickly and effectively identify those who require specialist referral. Following on from our article on autism in children in the January issue of *Prescriber*, this article discusses how to recognise and screen for autism in adults, and the support and interventions that can be offered.

Autism is a lifelong, neurodevelopmental condition characterised by social communication difficulties, unusually restricted interests, repetitive behaviour,[1] challenges adjusting to unexpected change and sensory sensitivities. Autism affects 1–2% of the population, and the prevalence of autism continues to rise.[2,3] 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] In addition, autism is reported to be more common in males, with a ratio of approximately 3:1 males to females.[2,3] However, there is also evidence that autistic females may be underdiagnosed, misdiagnosed, or have later diagnosis due to differences in symptom presentation and 'camouflaging' of their symptoms.[2]

This article focuses on the identification and management of autism in adults. For more information on screening, diagnosis and support in children, refer to the article in the January 2020 issue of *Prescriber*.

What causes autism?

Both genetic and environmental factors appear to play a role in the aetiology of autism. It is highly heritable, as 64–91% of likelihood is associated with genetic factors.[6,7] Approximately 5% of autism cases can be attributed to a single rare genetic mutation, and these individuals may have a particular outcome or phenotype. For example, individuals with 15q11.2 duplication syndrome tend to show hypotonia, motor delays, intellectual disability, autism and epilepsy.[8] However, in most cases, many small mutations of both rare and common genetic

variants contribute to autism.

In addition, some parental factors (*eg* increased paternal or maternal age at birth) and pregnancy complications (*eg* maternal gestational diabetes and labour complications)[9] have been shown to increase the likelihood of autism in children. Prenatal sex steroid hormone exposure (*eg* to elevated testosterone or oestrogen levels) has also been implicated,[10,11] and such hormones may affect brain development of the foetus. Each of these factors only increases the likelihood of developing autism by a small amount. Most cases of autism likely result from a combination and interaction of both genetic and environmental factors. These change brain development (both structure and function), and features of the autistic brain include early brain overgrowth in a subgroup, and increased number of neurones and neuronal connections,[2] as well as different patterns of neural activity during social and non-social information processing.[2]

What are the signs of autism in adults?

In adults, signs of autism may appear as atypical social behaviour, resistance to change and rigid routines, differences in sensory processing (including both over- or under-reaction to sensory stimuli), and repetitive and stereotyped behaviours that may be more visible in emotionally-charged situations. Social difficulties can manifest in a variety of ways and may include social isolation or preference for being alone; conversational difficulties, including challenges with reciprocal interactions (*ie* talking 'at' others or seeking interaction with others only to fulfil their own needs), misunderstanding sarcasm or tone, difficulties with non-literal language, or appearing blunt or rude without intending to do so; and poor understanding of social relationships, particularly friendships. Autistic individuals also frequently have a cognitive profile that prefers logical, factual and precise information. Adults on the autism spectrum tend to focus on details and prefer predictable situations.

Social communication differences in autistic adults are associated with impairments in theory of mind, which is the ability to draw inferences about what others are thinking and feeling (including their desires, intentions, beliefs and emotions). Autistic individuals have been shown to be worse, on average, 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).[12]

Why is autism diagnosis sometimes delayed?

There are many reasons why individuals may not have been given a diagnosis of autism when they were children. One reason is that diagnostic criteria for autism have changed over time. Although autism was originally described in 1943 by Leo Kanner, it was not until 2013 (with the publication of DSM-5) that diagnostic criteria were formally expanded to support adult autism diagnosis.[13] In addition, awareness about autism used to be less widespread and many individuals who currently meet criteria may not have been assessed as children.

Another reason for later diagnosis may be because of the significant heterogeneity in symptom presentation and severity among autistic individuals. The autism spectrum encompasses all levels of intellectual capacity.[2] Thus, depending on intellectual capacity, autistic individuals may have entirely different lifestyles, life experiences and healthcare needs. In addition, autistic individuals tend to show differences in sensory experience compared to others, but these differences are not uniform. Some autistic individuals may experience heightened sensitivity in particular senses, decreased sensitivity, or even both.[2] Furthermore, these differences span all sensory modalities, meaning that one autistic individual might have heightened sensitivity to sound, while another might have decreased sensitivity to visual stimuli, and another might have heightened sensitivity to touch, sound and visual stimuli.

Consider assessment for possible autism when a person has:
One or more of the following:
- Persistent difficulties in social interaction
- Persistent difficulties in social communication
- Stereotypic (rigid and repetitive) behaviours, resistance to change or restricted interests

And one or more of the following:
- Problems in obtaining or 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 attention deficit hyperactivity disorder) or mental disorder

Table 1. NICE criteria for autism assessment in adults[15]

Individuals without clear signs of developmental delay tend to be diagnosed later than those with more severe or obvious difficulties.[13] This can be, in part, because of their ability to 'camouflage' or mask their autism symptoms, which may be done intentionally or subconsciously. For example, individuals may learn appropriate social behaviours, such as maintaining eye contact during conversation or pre-planning jokes or anecdotes to keep a conversation flowing. While non-autistic adults also attempt to manage their social behaviours and interactions, it appears that autistic individuals may have a reduced capacity to do so (potentially due to reduced abilities in theory of mind[12]), and pressures to conform to social norms and conceal autism symptoms can cause exhaustion and distress.[14]

What about autism in women and girls?

For many years, autism was diagnosed far more often in males than females, with some estimates of the gender ratio in diagnosis as high as 10:1, in favour of males. Research now suggests that the gender ratio is likely closer to 3:1, and that underdiagnosis of women has been a significant issue.[2,3,14] In part, this may be due to qualitative differences in autistic trait presentation between males and females. For example, evidence of a strong, restricted interest in a particular area is a core feature of autism. Stereotypically, autistic males focus on objects, such as trains or cars, and may acquire a great deal of

knowledge on that subject. However, autistic females may have restricted interests that are more people-focused, including things like make-up or a particular rock band. This may mean that screening tools are less sensitive at detecting autistic women and girls. It may also mean that a clinician considering screening a female patient for autism may have relied too heavily on these stereotypes due to a limited knowledge of autism, thereby failing to refer them for a diagnostic assessment.

Furthermore, autistic females/women may remained undetected if they do not show the expected markers of atypical social behaviour or social isolation due to camouflaging of their autistic traits. Thus, a woman may not have been referred for a diagnosis if she was able to maintain friendships or romantic relationships, or to follow social norms for behaviour. As autism was traditionally conceptualised as more prevalent in males, GPs may have only chosen to refer women with more obvious autistic traits or difficulties.

How should we identify autism in adults?

GPs have a key role in the identification of autistic individuals who may warrant a specialist diagnostic assessment. This is particularly so in adults, as GPs may be the first person to recognise autism symptoms in a patient, or may be the first person that a patient speaks to if they suspect that they are autistic. Patients often experience significant delays in referral and eventual diagnosis, as well as delays in receiving support and interventions; therefore, it is important that GPs can quickly and effectively identify patients who may require a specialist referral for a diagnostic assessment.

NICE has released guidance specifically to help practitioners identify the signs of autism in adults (see Table 1). The guideline recommends referring an individual for assessment if they show evidence of autism symptoms (including difficulties with social interaction or communication, repetitive behaviours, restricted interests or resistance to change), along with evidence of other challenges (including difficulties in maintaining employment, education or social relationships; contact with mental health or learning disability services; or history of a neurodevelopmental condition, such as ADHD or intellectual disability).[15]

For individuals with an IQ within the average range or mild learning disabilities, multiple screening tools are available. The Autism Spectrum Quotient (AQ)[16] was designed to assess autistic traits of individuals; there are 50-item and 10-item versions available and they have been translated into various languages. Both these versions have shown excellent specificity, sensitivity and positive predictive value (all >0.80).[17, 18] There are both 50-item and 10-item versions available and they have been translated into various languages. The English version of the AQ-10 for use in adults is shown in Figure 1.

The AQ should only be used as a referral tool and not as a diagnostic measure for autism. This is primarily because of a lack of research in a primary care setting; more research needs to be done in this area when patients present to their GP with concerns about autism, or if the GP suspects the patient may be autistic. In addition, a positive score on the AQ may bolster

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AQ-10

Autism Spectrum Quotient (AQ)

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

the evidence for referral; however, patients who fail to meet the cut-off point may still be autistic. Clinical judgement must take priority when considering referrals for an autism assessment. Thus, it is critical that GPs are aware of the signs of autism and can directly apply this knowledge to refer their patients appropriately.

The AQ is not appropriate for individuals with moderate or severe learning disabilities. In these cases, an assessment should be made to identify possible signs of autism. Information on current or past symptoms may need to be supplemented or provided by caregivers, family members or partners.

Are there any other risks GPs should be aware of?

Autistic individuals may have greater risk of premature mortality compared with non-autistic individuals. Recent research from Sweden suggests that autistic individuals without intellectual disability die on average 12 years younger than others, and those with moderate or severe intellectual disability die on average 30 years younger than others, for a variety of reasons related to both physical and mental health. In particular, mortality was associated with diseases of the nervous system (including epilepsy) and suicide. More than 70% of autistic people experience other co-morbid physical and mental health conditions, which may also affect quality of life.[2] Autistic individuals are at higher risk of anxiety, schizophrenia, depression, suicide, self-harming behaviour, attention deficit hyperactivity disorder

(ADHD), epilepsy, obesity and sleep disturbances, as well as gastrointestinal, autoimmune, allergic, thyroid and diabetic conditions.[19]

High rates of physical and mental health conditions, and in particular suicidality, among autistic adults may be explained by a number of different factors: genetic risks, lifestyle choices, lack of social support and/or vulnerability to adverse life experiences (*eg* bullying at school).[20] It should be a priority to provide both safeguarding and to assess the individual for what kinds of support they may need. Unfortunately, there is still little research on the physical health risks of autistic adults over the age of 35 years. More work needs to be done to better understand how physical and mental health co-morbidities may be affecting the quality and length of life of autistic adults.

Management

We know that autism is a type of 'neurodiversity', or a difference in the way that an individual's brain is structured. Other conditions, such as dyslexia and ADHD also sit on this spectrum of neurodiversity. There are a variety of non-pharmacological interventions available for specific symptoms of autism that patients may be interested in. These include supported employment or living schemes, autism self-advocacy or support groups, or mentoring. The National Autistic Society provides information on local agencies and services that can provide support. In addition, cognitive behavioural therapy (CBT) may be recommended to help manage anxiety, depression and OCD, which commonly co-occur with autism.[2,19]

Currently, there are no effective pharmacological interventions for improving social communication in autistic individuals. Furthermore, many patients, practitioners and researchers have strong reservations as to whether any pharmacological intervention would be helpful in supporting autistic individuals with social communication difficulties. Antipsychotics (*eg* risperidone and aripiprazole) and SSRIs (*eg* citalopram, escitalopram, fluoxetine) have been used previously to manage repetitive and/or challenging behaviours in autistic patients; however, a significant risk of adverse reactions has also been reported.[2] NICE recommends combining psychosocial interventions with pharmacological ones.

Summary

Autism encompasses a wide spectrum of individuals, with a range of symptoms and intellectual abilities. Autistic adults may have a variety of healthcare needs related directly to their autism symptoms, or other physical or mental health co-morbidities. There are a number of reasons why autistic adults may not have been diagnosed as children, including changes to diagnostic criteria and camouflaging of symptoms in women and girls, in particular. Thus, GPs must have the means to identify whether adult patients show autistic traits that are causing impairment in their lives and, when there is sufficient evidence, must refer as soon as possible, to minimise delays in diagnosis and support.

There are brief tools available that can help with decision making about whether or not to refer an individual, including the 50-item and 10-item versions of the AQ. Further guidance should be sought via the NICE guideline that has been specifically designed to aid screening and support for autistic adults. There are also significant risks to mental and physical health associated with autism in adults; however, improving identification and diagnosis, and providing earlier interventions to manage autistic traits and secondary health conditions may greatly improve quality of life.

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Declaration of interests

None to declare.

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