GUIDELINES

Recognition, referral, diagnosis, and management of adults with autism: summary of NICE guidance

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on behalf of the guideline development group

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This is one of a series of *BMJ* summaries of new guidelines based on the best available evidence; they highlight important recommendations for clinical practice, especially where uncertainty or controversy exists.

Further information about the guidance, a list of members of the guideline development group, and the supporting evidence statements are in the full version on bmj.com.

Autism is a lifelong condition characterised by difficulties in social interaction and communication and by rigid or repetitive behaviours; it affects about 1.1% of adults.[1] Although some people's autism is diagnosed in childhood, for every three known cases, there are two individuals without a diagnosis who might need assessment, support, and interventions for autism at some point in their lives.[2] Four out of five adults with autism find that obtaining a diagnosis in adulthood is difficult or not possible,[3] and many who have all the core symptoms do not receive a formal diagnosis.[4] Particular problems arise in identifying high functioning autism (Asperger's syndrome), which may not be recognised until adulthood[5] or may be misdiagnosed as depression, personality disorder, or a psychotic illness. Inadequate identification and assessment of adults with autism not only leads to inadequate care but can also result in inadequate recognition and treatment of coexisting mental and physical health problems. Whereas care for children and young people is relatively well coordinated[6] this is often not the case for adults. Falling between and being passed around services is a particular problem for adults with autism who have an IQ over 70 and do not have severe and enduring mental illness, as they may be excluded from both learning disabilities and mental health services.[3] Social and economic exclusion affects a large proportion of adults with autism. Unemployment or underemployment is a considerable problem for adults with autism, including the 44% of those who do not have a learning disability,[7] with almost 90% of this group unemployed.[8]

This article summarises the most recent recommendations from the National Institute for Health and Clinical Excellence (NICE) on autism in adults.[9]

Recommendations

NICE recommendations are based on systematic reviews of best available evidence and explicit consideration of cost effectiveness. When minimal evidence is available, recommendations are based on the Guideline Development Group's experience and opinion of what constitutes good practice. Evidence levels for the recommendations are in the full version of this article on bmj.com.

General principles

All staff working with adults with autism should have an understanding of:

- The nature, development, and course of autism
- The impact on personal, social, educational, and occupational functioning
- The impact of the social and physical environment.

Assessment for autism

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

	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 142). www.nice.org.uk/CG142

Key reference: Allison et al[11]

Autism Spectrum Quotient—10 items (AQ-10): a quick referral guide for adults with suspected autism who do not have a learning disability

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Previous articles in this series

- Management of venous thromboembolic diseases and the role of thrombophilia testing: summary of NICE guidance (*BMJ* 2012;344:e3979)
- Management of an acute painful sickle cell episode in hospital (*BMJ* 2012;344:e4063)
- Recognition, referral, diagnosis, and management of adults with autism (*BMJ* 2012;344:e4082)
- Management of acute upper gastrointestinal bleeding (*BMJ* 2012;344:e3412)
- Prescribing strong opioids for pain in adult palliative care (*BMJ* 2012;344:e2806)

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.

For adults with possible autism who do not have a moderate or severe learning disability, consider using the Autism Spectrum Quotient, 10 items (the AQ-10; figure).[11] (If a person has reading difficulties, read out the AQ-10.) If a person scores above 6 on the AQ-10, or clinical judgment suggests autism (taking into account any history provided by an informant), offer a comprehensive assessment for autism.

During a comprehensive assessment, inquire about and assess the following:

- Core autism signs and symptoms (difficulties in social interaction and communication and the presence of stereotypic behaviour, resistance to change or restricted interests) that were present in childhood and have continued into adulthood
- Early developmental history, where possible
- Behavioural problems
- Functioning at home, in education, or in employment
- Past and current physical and mental disorders (for example, schizophrenia, depression, or other mood disorders; and anxiety disorders—in particular, social anxiety disorder and obsessive compulsive disorder)
- Other neurodevelopmental conditions
- Neurological disorders (for example, epilepsy)
- Communication difficulties (for example, speech and language problems, and selective mutism)
- Hypersensory and/or hyposensory sensitivities and attention to detail.

Carry out direct observation of core autism signs and symptoms especially in social situations.

Assessment of challenging behaviour

When assessing challenging behaviour, do a functional analysis (see "Interventions for challenging behaviour" below), including identifying and evaluating any factors that may trigger or maintain the behaviour, such as:

- Physical disorders
- The social environment (including relationships with family members, partners, carers, and friends)
- The physical environment, including sensory factors
- Coexisting mental disorders (including depression, anxiety disorders, and psychosis)
- Communication problems
- Changes to routines or personal circumstances.

Interventions for autism

For adults with autism without a learning disability or with a mild learning disability, who are having difficulty obtaining or maintaining employment, consider an individual supported employment programme.

An individual supported employment programme should typically include:

- Help with writing CVs and job applications and preparing for interviews
- Training for the identified work role and work related behaviours
- Carefully matching the person with autism with the job
- Advice to employers about making reasonable adjustments to the workplace
- Continuing support for the person after they start work
- Support for the employer before and after the person starts work, including autism awareness training.

For the management of core symptoms of autism in adults, do not use anticonvulsants, chelation, drugs specifically designed to improve cognitive functioning (for example, cholinesterase inhibitors), oxytocin, secretin, drugs for testosterone regulation, hyperbaric oxygen therapy, antipsychotic medication, or antidepressant medication.

Interventions for challenging behaviour

When deciding on the nature and content of a psychosocial intervention for challenging behaviour, use a functional analysis. The functional analysis should facilitate the targeting of interventions by:

- Providing information, from a range of environments, on factors that seem to trigger the challenging behaviour; and on the consequences of the behaviour (that is, the reinforcement received as a result of their behaviour)
- Identifying trends in the occurrence of challenging behaviour, factors that may be evoking that behaviour, and the needs that the person is trying to meet by behaving in that manner.

Psychosocial interventions for challenging behaviour should include:

- Clearly identified target behaviour(s)
- A focus on outcomes that are linked to quality of life
- Assessment and modification of environmental factors that may contribute to initiating or maintaining the behaviour
- A clearly defined intervention strategy
- A clear schedule of reinforcement, and capacity to offer reinforcement promptly and contingently on demonstration of the desired behaviour
- A specified timescale to meet intervention goals (to promote modification of intervention strategies that do not lead to change within a specified time)
- A systematic measure of the target behaviour(s) taken before and after the intervention to ascertain whether the agreed outcomes are being met.

Consider antipsychotic medication in conjunction with a psychosocial intervention for challenging behaviour when there has been no or limited response to psychosocial or other interventions (such as environmental adaptations). Antipsychotic medication should be prescribed by a specialist and quality of life outcomes monitored carefully. Review the effects of the medication after three to four weeks, and discontinue it if there is no indication of a clinically important response at six weeks.

Support for families and carers

Offer families, partners, and carers of adults with autism an assessment of their own needs, including personal, social, and emotional support; support in their caring role, including respite care and emergency plans; advice on and support in obtaining practical support; planning of future care for the person with autism. [*Based on the experience and opinion of the GDG*]

Offer information, advice, training, and support to families, partners, and carers if they need help with the personal, social, or emotional care of the person with autism; or if they are involved in supporting the delivery of an intervention (in collaboration with professionals).

Overcoming barriers

Primary and secondary care professionals currently have a limited knowledge of autism and its various presentations,[12] and this lack will need specific attention if the recommendations in this guideline are to be of real benefit. Two areas are of particular concern: initial identification in primary care, and autism symptoms being masked by comorbid conditions in secondary care.[13] Training in the identification and assessment of autism should be more prominent in the undergraduate and postgraduate education of health and social care professionals.

People with autism, particularly those who are more disabled by it, often fall through the gaps between medical and social care, especially if they do not present with a mental health disorder or learning disability. This presents challenges in developing integrated health and social care services that engage people with autism. Access to treatment for adults with autism is also limited and extends beyond mental healthcare to physical healthcare. The establishment of local autism strategy groups (which should be responsible for developing and managing local care pathways) and of associated multiprofessional teams should help to resolve this problem of access to treatment, but care is needed to ensure that adults with autism have access to the full range of healthcare services. However, it is the families of adults with autism who provide much of the care and support. The guideline highlights that they too should receive specific support as without their involvement many interventions will be of limited benefit.

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A PATIENT'S JOURNEY

After repair of tracheo-oesophageal atresia

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Despite surgery for short gap tracheo-oesophageal atresia and fistula as a baby, a doctor describes how she suffered associated cough and illness throughout childhood and as a student before receiving the diagnosis and care she needed to manage her symptoms

When my first feed in hospital resulted in coughing, choking, and going blue, it was evident that I couldn't swallow. I was quickly transferred to a larger hospital and short gap tracheo-oesophageal atresia and fistula was diagnosed. At the time (1975), although repair of this condition had first been performed in 1948, survival was far from guaranteed. However, despite needing a thoracotomy and trans-sternal incision, the repair was successful, and I left hospital three months later, supposedly cured.

The "cure"

Although I was the first child of my young parents, they quickly realised I wasn't normal. I slept very badly, choked, and constantly coughed and was irritable and unwell most of the time. Thus began my mother's frequent visits to general practitioners and (initially) paediatric surgeons. Because I was a normal weight, doctors assumed my mother was simply overprotective owing to my initial defect. In addition, my lungs were always clear, so no infection was evident. The one concession to the frequent visits was the diagnosis of reflux, but at this time, Gaviscon was the only option.

In between visits to the doctor, my mother had to cope with having a child with a near constant "TOF cough": the TOF (tracheo-oesophageal fistula) cough is a result of tracheobronchomalacia and tracheal scarring, and sounds like a seal barking. My mother was often accosted by strangers chiding her for taking a child with obvious whooping cough out in the streets, and people pointed and stared.

Despite the ungodly noises coming out of the child in front of them, the doctors continually told us there was nothing wrong with me, and no need for any investigations or treatment. Ignoring my frequent absences from school (I missed at least two weeks each term) due to "chest infections" and the sequelae of prolonged coughing (black eyes owing to petechial haemorrhage, torn intercostals, and frequent haemoptysis), the doctors did not acknowledge that I had any problem.

A closed mind

By my late teens, I had internalised the unspoken message of the consultations with doctors. I believed that I was a hypochondriac or simply not as stoical as others and that this explained my frequent absences from school and medical school. Even when I questioned a paediatric surgeon at medical school about whether there was any long term impact of TOF, I was again told there were none.

My mind became closed to any suggestion of disease. Once I started working as a doctor, colleagues suggested there might be a problem, but I could not acknowledge it in the light of my past experiences. When I was a house officer on call, my senior house officer threatened to admit me as my oxygen saturation levels were 92%, but I shrugged it off as a joke. A doctor housemate complained I kept her awake all night coughing, but, as I slept fine, I thought she was over-reacting. It was not until my consultant and the occupational health doctor insisted on a respiratory opinion when I was 30 (after I had had four "chest infections" in four months) that I was forced to accept there might be a problem.

Looking for answers

My first respiratory history revealed how deeply in denial I was about the problem. It had not occurred to me that others do not cough every day, cough up sputum daily, or suffer "infections" as frequently as I did. A range of investigations revealed severe tracheobronchomalacia and a fluid filled pouch at the base of the trachea. At this point I was referred to the Liverpool cardiothoracic unit. Further investigation there showed poor oesophageal motility, that my trachea, oesophagus, and aorta were stuck together, and my lungs were completely normal. However, I was getting sicker and sicker as each investigation exacerbated my chest problems.

Cognitive dissonance

The discovery that I did have a real health problem had a major psychological impact on me. I was extremely angry. I was angry first at my senior colleagues for making me face up to the truth, and then at all the doctors who had treated me in the past. I was angry at the doctors not because they had not known what to do, but because they were too arrogant to admit their ignorance and seek further information, instead blaming my family and me for the problem. No one could expect a GP to know what the long term outlook of this new patient group would be, but in refusing to accept there might be one, they failed me. The cognitive dissonance that this anger caused was hard to deal with. On one hand I was a hospital doctor treating patients, but on the other, I resented the medical profession and the attitudes that had brought me to this point. I also grieved for the child

WHAT THIS HAS TAUGHT ME

As a doctor

- Patients prefer to hear "I don't know" to unfounded explanations or reassurances
- It is not a professional failure to seek advice from other doctors who may know more
- It cannot be underestimated how rude people can be about differences, visible (such as rashes and marks) or otherwise. Strangers, friends, and family may be culprits, even if well meaning. This has been invaluable experience for my work in dermatology
- The medical profession is not good at accepting and dealing with sick colleagues—there is an unwritten rule that doctors are not allowed to be ill
- People with lifelong chronic disease become institutionalised; this may be through becoming a medic or through medical attention-seeking behaviour
- Patients with rare diagnoses also get common problems

As a patient

- Be your own expert patient—my treatment as a patient improved immeasurably once I added MBChB after my name, and I understand my health better than most GPs
- Having a rare and poorly understood condition is difficult to live with and to explain to others
- Clear boundaries are essential for staying well and being able to work—pacing oneself and not agreeing to extra work is essential (I'm still working on that one)
- My biggest struggle has been to accept that I have only limited control of my health, and am not to blame if it deteriorates, but at the same time I must take appropriate measures to protect it
- There are benefits to having been left to fend for myself healthwise for many years. Because I was "cured," there was an expectation I should achieve academically and get a job. I'm not sure I would have achieved as much if there had been an excuse not to

RESOURCES FOR PATIENTS AND CLINICIANS

- TOFS (www.tofs.org.uk)—A UK based charity providing emotional support to families of children born with tracheo-oesophageal fistula (TOF), oesophageal atresia, and associated conditions. The site also links to a range of European TOF groups in other languages
- ea/TEF (Esophageal Atresia/Tracheoesophageal) Child and Family Support Connection (www.eatef.org/)—A US non-profit organisation supporting families and educating them and medical professionals about the condition
- OARA (Oesophageal Atresia Research Auxillary) (http://rchfoundation.com.au/event/OARA)—An Australian support group based at the Royal Children's Hospital, Melbourne that supports families, organises research into the condition, and funds specialist nurses
- Living With Reflux (www.livingwithreflux.org)—A UK based support group for families with children with severe gastro-oesophageal reflux disease
- PAGER (Pediatric/Adolescent Gastroesophageal Reflux Association) (www.reflux.org)—A US non-profit organisation offering advice and support for families with a child with gastro-oesophageal reflux disease

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Previous articles in this series

- Lyme neuroborreliosis (*BMJ* 2012;344:e3250)
- Gitelman syndrome (*BMJ* 2012;344:e3590)
- Ulcerative colitis (*BMJ* 2012;344:e2947)
- At either end of the tube (*BMJ* 2012;344:e2971)
- Amyloidosis (*BMJ* 2011;343:d6326)

This is one of a series of occasional articles by patients about their experiences that offer lessons to doctors. The *BMJ* welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance.

I had been—the breathless toddler learning to speak quietly and quickly in between coughs, the schoolgirl bullied for my coughing, and the teenager cancelling social events owing to ill health.

Finding answers

Although the doctor at the Liverpool cardiothoracic unit had begun to improve things for me, the real turning point came when I moved to Yorkshire. I was referred to Professor Morice's cough clinic. The first sign of what was to come was the reflux severity index I completed on arrival. I quickly realised I scored highly on all counts. When this was followed by a consultation in which my condition was both understood and explained, and a treatment plan devised, I felt hope for the first time in many months. I learnt that my "chest infections" were episodes of aspiration, hence their unresponsiveness to steroids, and my other symptoms were manifestations of gastro-oesophageal reflux. I was started on a range of medications, to see what would control the symptoms, and I have found a regimen that keeps things settled. I am still learning about my condition—at a recent conference I learnt that the reason for my frequent choking episodes is that in people with a tracheo-oesophageal fistula the pharynx may also be dysfunctional, resulting in an unsafe swallow. Such information doesn't alter anything but helps me understand my problems better. As I've improved, I realise I am luckier than many with my symptoms, most of which come from an irritant tracheitis, which has prevented deeper damage, but others in similar situations develop chronic obstructive pulmonary disease or bronchiectasis.

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A DOCTOR'S PERSPECTIVE

Chronic cough is a common but neglected symptom. In the 19th century its origin was suggested to be a form of reflux, and it is only recently that irritation by a gaseous, non-acid, mist has been shown to cause exquisite hypersensitivity of the upper airways. When aspirated, this reflux can give rise to wheezing, leading to the mistaken diagnosis of asthma, and consequent lung damage leads to many other respiratory conditions, such as bronchiectasis, exacerbations of chronic obstructive pulmonary disease, and even pulmonary fibrosis. No objective tests reliably detect the cough hypersensitivity syndrome, but the clinical history of associated features is characteristic. A validated questionnaire (available at www.issc.info) allows the identification of airway reflux as the cause of otherwise mysterious respiratory and upper airway symptoms.

"TOF" (derived from tracheo-oesophageal fistula) is a generic term for a variety of congenital abnormalities of the trachea and oesophagus. Embryologically the lungs are a mere "out-pouching" of the gut. Most patients present in the neonatal period with choking attacks associated with feeding. Pioneering surgery developed in the 1960s and '70s corrected the anatomical defects and the patient was apparently "cured." However, the oesophagus is an extremely complex organ, which not only conveys the food into the stomach but is also responsible for the retrograde transport of excessive gas. The delicate physiology of this biphasic movement remains disordered in the overwhelming majority of patients with TOF. This leads to recurrent episodes of non-acid reflux and aspiration. In the very active patients' association TOFS, this is known as TOF cough. Because many operations have been performed over the years in our cardiothoracic centre, I have seen many such patients and, indeed, have been labelled as the Prof of TOF cough.

Unlike for patients with simple chronic cough, for patients with TOF cough, imaging such as barium studies may be rewarding, delineating the large dilated sections of oesophagus or colonic transposition, which develop after surgery. Mechanical approaches, such as dilation or myotomy can then be helpful. As in chronic cough, drug treatment is targeted at enhancing motility and relieving any oesophagogastric obstruction. Promotility agents such as the dopamine agonists, metoclopramide, and domperidone may help. The GABA (γ-aminobutyric acid) agonist baclofen can, by enhancing lower oesophageal tone, prevent frank gastroesophageal reflux. The macrolide antibiotics, erythromycin and azithromycin, act as agonists of the hormone motilin, enhancing the poor peristalsis; hence, their activity in other respiratory conditions such as exacerbations of chronic obstructive pulmonary disease and cystic fibrosis. Finally, trazodone acts via 5-HT$_2$ receptors to enhance oesophageal transit.

For intractable coughing, low dose morphine can in some patients bring significant relief, and the dexbrompheniramine component of some combination antihistamine preparations is an antagonist of the TRPV1 cough receptor. Cough suppression, however, must be used cautiously since a cough is primarily a protective reflex against aspiration and the clinician must balance symptom relief against possible increase in end organ damage.

Alyn H Morice, professor of respiratory medicine

ANSWERS TO ENDGAMES, p 48

For long answers go to the Education channel on bmj.com

CASE REPORT Chest pain and ST elevation

1 The most important immediate cause of ST elevation is acute myocardial infarction. Other causes include pericarditis, early repolarisation, left bundle branch block, left ventricular hypertrophy, coronary spasm, hyperkalaemia, and Brugada syndrome.

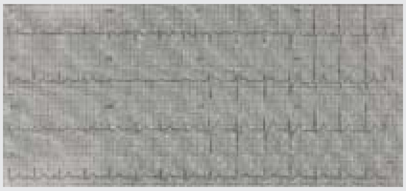

Fig 1 | **ECG on presentation**

Fig 2 | **ECG with maximal J point elevation**

2 The ECG appearances on presentation are non-specific (fig 1), but the subsequent ECG pattern of coved ST elevation and T wave inversion in the right precordial leads is typical of a type 1 Brugada pattern ECG (fig 2). However, given the history of chest pain (which is not a feature of Brugada syndrome), myocardial ischaemia should be excluded first.

3 Patients with Brugada syndrome should be provided with a list of drugs to avoid and be advised to take antipyretic agents in the event of a fever. Family screening should also be recommended.

STATISTICAL QUESTION
Cluster randomised controlled trials

Statements *a* and *b* are true, while *c* and *d* are false.

PICTURE QUIZ
A reticulated eruption on the lower abdomen in a 17 year old girl

1 A localised area of reddish brown reticulate discoloration affecting the lower abdomen.

2 The most likely diagnosis is erythema ab igne.

3 ab igne may be a sign of underlying organic disease because local application of heat is an old but effective remedy for pain relief. There is also a small but real risk of erythema ab igne undergoing malignant transformation.

4 The mainstay of treatment is to eliminate exposure to the source of infrared radiation.