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Making the diagnosis

→ Key points

- ◆ The diagnosis of autism and Asperger syndrome is often carried out by a multidisciplinary team, typically taking 2 or 3 hours, based on interview and observation. Classic autism is typically usually diagnosed by the age of 3, and can be diagnosed as young as 18 months. Asperger syndrome is often not diagnosed until at least 6, and often much later than this, including late diagnosis in adulthood.
- ◆ In the future, biological markers may enable diagnosis to be more objective. When any claims emerge for a biologically based diagnostic test, there will need to be research to establish if the test has good specificity (does the test just identify people with autism spectrum conditions, or does it also identify people with other conditions?) and good sensitivity (how many cases does the test identify correctly, how many does it miss, and how many does it identify incorrectly?).
- ◆ Diagnosis is of most value when it is not just a label but a passport into accessing all the necessary support services that each individual may need.

We have covered some of the history of autism and Asperger syndrome, and been introduced to the concept of autistic traits lying on a continuum. But for some readers of this book, the urgent question in their mind is where to obtain a diagnosis and what to expect from the doctor making the diagnosis. Appendix 2 contains a list of all the autism and Asperger associations that we know about throughout the world, who could at least direct you to a clinic local to you. Irrespective of whether you are the parent of a young child



with suspected autism or Asperger syndrome, or are an adult with suspected Asperger syndrome, the diagnostic assessment has a typical format that involves asking questions to collect information around three key questions:

1. Does the person have significant *social* difficulties?

The clinician conducting the diagnostic interview (who might be a child psychiatrist, clinical or educational psychologist, paediatrician or other health professional) might ask you questions such as (but not restricted to) the following:

- ◆ Has the person found it difficult to make and keep friends?
- ◆ Have they found it hard to understand and respond appropriately to other people's feelings?
- ◆ Can they join in larger, unstructured social groups (not just one-to-one social interaction)?
- ◆ Are they socially withdrawn?
- ◆ Do they tend to misinterpret other's intentions?
- ◆ Do they use *eye contact* inappropriately, either staring at others for too long, or looking at other people's eyes too briefly, or at the wrong times?
- ◆ Do they show a lack of normal *social awareness*?
- ◆ Are they to some extent oblivious of what others are thinking about them, or how they come across to others?
- ◆ Would they spontaneously comfort another person?
- ◆ Can they pick up on other people's feelings, not just using the extreme cues (such as someone crying), but using more subtle cues (e.g. to determine if someone is pretending to be happy)?
- ◆ In the second year of life, was there a delay in showing *joint attention* (such as following where others are looking, or using the pointing gesture to share interest or following another person pointing to share interest)?
- ◆ Has the person always been more comfortable in solitary pursuits?

These latter two examples are to determine if social abnormalities have been present right across the person's development, from infancy onwards.



2. The person's *communication* skills.

As mentioned in Chapter 1, social and communication skills cannot be truly separated, but for the diagnosis the clinician may try to cover these separately. The kinds of interviewing questions that they might ask include (but are not restricted to) the following:

- ◆ Does the person have a very *literal* understanding of language?
- ◆ Does the person have trouble understanding non-literal language (such as humour, sarcasm, irony and metaphor)?
- ◆ Is there a noticeable difference between the person's technical language skills (e.g. their vocabulary, or their syntactic ability) and their *pragmatic* language skills (i.e. being able to use language appropriately to the social context)?
- ◆ Does the person frequently say (as well as do) the wrong thing in a social situation (committing *faux pas*)?
- ◆ Do they tend to provide either *too little* information or *too much* information in their speech (or e-mails or other written communication)? Being able to judge what the listener needs to know is part of being able to take the other person's perspective into account.
- ◆ Do they have trouble with *turn-taking* in language, tending to hold forth in a monologue (and not realize that others are getting bored)?
- ◆ Was there any *delay* in their language skills? (Recall from Chapter 1 that a sign of delay is relevant to distinguishing classic autism from Asperger syndrome.) Delay is typically defined when the child is not using single words by 2 years old, or any phrase speech by 3 years old.
- ◆ Was the pattern of language acquisition atypical? For example, were first words highly unusual, such as being unusually specific or rare words?

3. The person's narrow, unusual and strong interests, as well as unusually repetitive behaviour.

The clinician might ask questions such as (but not confined to) the following:

- ◆ Has the person—right across their life—been capable of becoming *totally immersed* in one activity or interest to the exclusion of all else, concentrating for many hours each day on just one unusual topic?
- ◆ Did they tend to become an expert on that topic?

(continued)



- ◆ Would that topic have the quality of an *obsession*, in that efforts by others to shift the person on to new activities failed?
- ◆ Have they had a strong need to do the same thing over and over again, in a highly similar way? For example, positioning objects in the house and becoming very upset if other people moved them? Or taking the same route to school or work? Or performing some activity in a strict sequence that they refuse to change?
- ◆ Would they have become very upset if they had had to deviate from this routine?
- ◆ Do they invariably go through the *same* sequence of actions when beginning certain activities?
- ◆ Do they insist on wearing the *same* clothes, or eating the same food or going to the same places, over and over again?
- ◆ Do they resist change?

These latter characteristics, for me, go to the heart of autism and Asperger syndrome. Kanner identified this as the person's *need for sameness*. At other times he referred to it as their *resistance to change*. It is almost as if, just as a Type 1 diabetic starts to suffer if their blood sugar level rises too quickly, a person with autism or Asperger syndrome starts to suffer if they encounter unexpected change. We will see in Chapter 5 how this 'symptom' has been explained in terms of the person's strong drive to *systemize* events, to render them as near to predictable as they can. Anything that occurs in an unpredictable way is likely to throw the person into a panic and may trigger a withdrawal or an avoidance, or a desperate attempt to re-establish predictability by imposing a fixed pattern or sequence of behaviour.

The structured interview is conducted with a view to establishing if the person's behaviour in each of these three areas is significantly unusual, and if their difficulties in each of the three areas have interfered with everyday functioning. The interview is usually complemented by direct observation, in order to gather direct evidence for each of these abnormalities.

Standardized instruments

When diagnosing a child, it is increasingly usual to use a standardized method such as the ADI (*Autism Diagnostic Interview*) and/or the ADOS (the *Autism Diagnostic Observational Schedule*). These methods were developed by Michael Rutter in London and Cathy Lord in Michigan, and are sometimes referred to



as the ‘gold standard’ in diagnosis. Clinicians have to pay to be trained in these methods, and training takes about 1 week. During this training and after it, the trainee is assessed for how accurately they have learnt to make the diagnosis, i.e. how reliably they agree with other trained clinicians.

Such standardization of diagnostic methods was important to attempt, because previously all that was available was ‘clinical judgement’ or the doctor’s opinion. However, the latest research shows that these methods are not a gold standard in that they work best when combined with ‘clinical opinion’. That is, the original hope that they could replace the subjective opinion of the doctor has not turned out to be the case, because they miss some cases of Asperger syndrome. They are also less useful for the assessment of adults, though there are some standardized methods available for this purpose, such as the *Adult Asperger Assessment* (AAA).

One day, the hope is that accurate diagnosis will not depend on the vagaries of a clinical interview or of direct observation of behaviour, which invariably includes some subjective elements on the part of the doctor. Instead, it will be based on a biological marker or set of markers (e.g. a combination of specific gene variants, or a combination of specific protein levels), measured in the blood or in other bodily tissue or cells. This is how, for example, other conditions such as Down syndrome or phenylketonuria (PKU) are diagnosed. But for now, such a set of biological markers for autism or Asperger syndrome are not yet available, so we need to continue to rely on behavioural and interview-based methods.

Intelligence tests (IQ), and related educational and cognitive measures

It is important that the clinic has some measure of the person’s overall IQ, since for the diagnosis of Asperger syndrome the person must have an IQ in the average (or above average) range, i.e. have no signs of general learning difficulties. We discussed in Chapter 2 how different IQ bands in some sense enable a clinician to refine the subgroup into which the person’s diagnosis falls. IQ is also important because it remains a very strong predictor of prognosis, and because the specific profile on an IQ test (e.g. strengths in visual spatial tests or difficulties in verbal tests) can be used to plan individual educational programmes for the child.

For a child, it is also important to have a measure of language ability (both comprehension and expression): recall that the diagnosis of Asperger



syndrome is only made if the person shows no signs of language delay. Noting the size of the language delay will also be of practical importance in planning interventions such as speech therapy for a child with classic autism. Finally, in some clinics, the doctor will undertake some other psychological tests (of everyday planning abilities, or of memory, for example) not because these are intrinsic to the diagnosis (they are not) but because they might help to understand the person's unique pattern of strengths and difficulties.

What to expect when you go for a diagnostic assessment

Most clinics use a multidisciplinary team. What this means is that you might expect a mix of professionals in the interview or in the room observing you or your child. As mentioned earlier, these might include a child psychiatrist, clinical psychologist, speech therapist, educational psychologist, paediatrician or other related disciplines (neurologists, for example). Sometimes in order not to be too overwhelming, the team may observe behind a one-way mirror, always with your prior knowledge and consent.

Typically a diagnostic assessment takes at least 2–3 hours, and in some clinics it takes a whole day, with breaks. The team should be able to give you the outcome (a diagnosis if appropriate) on the same day, and you should try to prepare some questions to ask the team, in the event that they confirm the diagnosis is on the autistic spectrum. That way, you make the most of being with the specialists, and can benefit the most from their advice.

What should happen immediately following a diagnosis

People react differently to hearing the words 'I think your child has autism' or 'I think you have Asperger syndrome'. Some are relieved that finally there is a name for the condition that has always made them feel they, or their child, are different; and relieved that finally they have a signpost for where to go for the most relevant help. Some react with a sense of shock that they, or their child, have a condition that is understood to be genetic, affecting brain development (see Chapter 6). The shock can sometimes turn to sadness if hopes turn to disappointments about the future. People vary in how quickly they adjust to the diagnosis.

The clinician giving you the diagnosis should tell you about the National Autistic Society (NAS) in the UK, the Autism Society of America (ASA) in



the US, or the equivalent in your state or country (see Appendix 2 for a list of such associations). These mostly began as parent-led charities and in many countries have become powerful lobbying groups, running services such as special schools, adult day centres, sheltered housing or sheltered employment services, social groups, playgroups, etc.

For a diagnosis of Asperger syndrome (or high-functioning autism), it is possible to see the diagnosis in a positive light. It is a statement that the person has followed an atypical path of brain development. In the language of people with Asperger syndrome, they are not 'neurotypical'. As we will see in Chapter 5, the psychology of autism and Asperger syndrome involves areas of strength as well as areas of difficulties. The difficulties (in socializing and communicating through small-talk) can be disabling unless environments are chosen to minimize this. But the strengths (in attention to detail, the ability to concentrate for hours and hours on a single topic, the thoroughness with which narrow topics are explored and the systematic approach to certain activities) can be great assets if they can be harnessed usefully (in education, in work, or in hobbies, for example).

Where to find help and support

The list of agencies at the end of this book may provide a useful starting point for support groups near you.

How early can a diagnosis be made?

Autism can now be reliably diagnosed by 18 months of age. Many clinics are unaware that this is possible, but studies have been conducted showing that diagnoses made at that age using the established instruments such as the ADI and ADOS are reliable, and predict later diagnosis.

Some Health Visitors and GPs/paediatricians use screening instruments such as the Checklist for Autism in Toddlers (CHAT), which look for the absence of behaviours that one would expect to be present in a typically developing toddler (such as joint attention) as well as the presence of behaviours that are not usually present in a typically developing toddler (such as rocking back and forth for hours). The CHAT has been modified into the Modified-CHAT (M-CHAT) and the Quantitative-CHAT (Q-CHAT). [See www.autismresearchcentre.com for the latter.] These are not diagnostic, but help to indicate if a child might warrant a full diagnostic assessment.



Will my child grow out of it? What will happen to him or her when they grow up?

Autism and Asperger syndrome are life-long in that they reflect the make up of the brain. Whilst the brain changes and adapts, ultimately the core of autism and Asperger syndrome is part of who that person is. In the case of Asperger syndrome we can think of the condition as being a form of personality type. It is possible to adapt your personality to the outside world when needed, but one's personality ultimately is who you are, and aspects of the core (such as excellent attention to detail or sensory hyper-sensitivity) do not fundamentally change across one's life. For some people, social skills improve to varying degrees, with age and experience.

Can the diagnosis be removed later?

A person who receives a diagnosis will not necessarily need that diagnosis all their lives. A diagnosis is made at a particular snapshot in time, at a point in that person's life when things had got so difficult that they needed the diagnosis in order to access support and help.

In the case of Asperger syndrome it may, for example, have been useful as a diagnosis as a teenager, when they weren't coping with mainstream school and all of the social pressures that this implies. By adulthood, that same person might have found a niche in which they not only feel they fit, but in which they are thriving, and feel they no longer need the diagnosis. I have come across people who seek the diagnosis and I have come across people who seek to be undiagnosed. The latter are just as valid as the former, but will need just as thorough a reassessment of the individual. This is to check it is the case that they are coping sufficiently such that the autistic traits that they have no longer interfere with their daily life. If this is the case, then they no longer meet the criteria for a diagnosis. The clinician needs to discuss with them the pros and cons of removing the diagnosis.

In the case of classic autism, we need to be realistic that the person may need the diagnosis all of their lives. We discussed in Chapter 2 how in some sense this is the more 'severe' subgroup on the autistic spectrum. The rare exceptions to this are individuals with high-functioning autism, who in terms of daily living skills may achieve the same level of independence as someone with Asperger syndrome. But the medium- and low-functioning individuals on the autistic spectrum will need their diagnosis all their lives, to ensure they obtain help with sheltered living, sheltered employment and protection as a vulnerable person.



Horror stories

There are examples of bad practice out there, and there is still a lot of ignorance or misunderstanding about autism and Asperger syndrome. I wish this book could contain all positive stories, but to do this would be pure spin. We need to look at what is actually happening on the ground, not how things should be in an ideal world. Here are two examples that make me very worried:

- ◆ Social workers who refuse to believe that a child has autism or Asperger syndrome, and instead think the child's difficult behaviour (their social difficulties, their tantrums at change, their lack of social conformity, their learning difficulties) are signs of *inadequate parenting and neglect*, rather than signs of a neurological condition.
- ◆ Parents who are accused of *Munchausen's by proxy*, a clinician's term for suggesting the parent wants his or her child to have problems, in order to satisfy some disturbed need for the attention of doctors.

I have heard of parents whose child is put on the 'at risk' register because the social workers believe such things. Such attitudes are horrific and set the clock back by 50 years in perceiving autism and Asperger syndrome as reactions to poor parenting, instead of recognizing that autism and Asperger syndrome are medical conditions in need of sensitive support. Recall in Chapter 2 how we discussed Bettelheim's inaccurate theory (in the 1960s) about parents causing their child's autism, and Rutter's refutation of this idea (in the 1970s).

Naturally, social workers and others need to be open to the possibility of abuse or neglect, but this should not be at the cost of dismissing an alternative diagnosis. The horror cases I have come across tend to involve a dogmatic bigotry on the part of social services who may say that they do not believe that Asperger syndrome exists, or believe that there is some fashion involving over-diagnosis of Asperger syndrome, and a refusal to acknowledge such cases as genuine. The despair of such parents on the receiving end of such dismissive attitudes is heart breaking.

I have also heard of other horror stories:

- ◆ Parents of children with autism not receiving appropriate support and feeling so desperate with a child who does not sleep, cannot adjust to change, and is occasionally even aggressive, that the parent attempts suicide or actually commits suicide. Such tragedies are preventable with a little bit of humanity and care on the part of local services.



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- ◆ Adults with Asperger syndrome who, having got the diagnosis, find that their local services do nothing. Such adults sometimes decide that since no-one cares, and they have slipped into a depression, they will end their own life. Again, such suicides are preventable if agencies are properly 'joined up'.
- ◆ The local services sometimes pass the buck by saying that Asperger syndrome is the responsibility of the local mental health team. They in turn say that Asperger syndrome is not a mental health condition but a learning disability. The learning disabilities team then say that Asperger syndrome is not a learning disability because the person has an IQ above 70 and is an educational responsibility. The educational authority may then pass the buck by saying it is a social services responsibility, who pass the responsibility back to the mental health team. You can see how this can go round and round in circles, whilst the person with Asperger syndrome feels more and more isolated.

Parents still need to be advocates for their child with autism

From the above, it will become apparent that our world is still not as autism- or as Asperger-friendly as it could be. Until it is, parents and others continue to have a role in educating their local school, social services, GP or education authority about the nature of autism and Asperger syndrome, and in fighting on behalf of their child for appropriate support. They should not have to take on this role, because they may already have a lot of stress to deal with. Realistically, it is important to recognize that such parental involvement may be essential in helping a person with autism or Asperger syndrome get the right support. Joining a parent support group can make it feel less like having to fight alone.

Students with Asperger syndrome

There are other equally avoidable sad outcomes, such as students with Asperger syndrome dropping out of university because the university system is not being flexible in making allowances for their Asperger syndrome. I know for example of students with Asperger syndrome whose learning style is more suited to private study (from textbooks, or from journal articles available via electronic libraries on the web), but who are required to attend lectures and seminars as part of their degree.



Lectures and seminars were not designed for people with Asperger syndrome, because these educational formats typically

- ◆ involve large social groups
- ◆ are noisy
- ◆ expect the student to edit what the lecturer is saying into short-hand notes
- ◆ expect the student to switch topics after 55 minutes
- ◆ expect the student to do two things (listening and writing) at once
- ◆ expect the student to sit in any available place
- ◆ expect the student to concentrate even with whispering from other students.

In contrast, many students with Asperger syndrome may

- ◆ prefer to work in silence
- ◆ prefer to go slowly and methodically
- ◆ prefer not to have to edit (for fear of losing important detail)
- ◆ prefer to error-check, to be sure that a fact is a fact
- ◆ prefer conditions to remain unchanged (same seat, same lighting, etc.)
- ◆ prefer lack of distractions
- ◆ prefer to see all of the logical steps or evidence for each statement, rather than accepting assertions in the absence of explanations
- ◆ prefer, once they start a topic, to stay on that topic for many hours, ignoring lunch or drinks or even the need to go to the bathroom
- ◆ become irritated by the intrusion of other people into their space
- ◆ become anxious if other people talk to them unexpectedly
- ◆ become irritated by human errors in lecture handouts
- ◆ become irritated by a chatty style of lecturing
- ◆ become irritated by the whispering students in the row behind them in the lecture theatre who want to talk about who is going out with who.



Universities are places of learning, and there should not be a dogmatic attitude that assumes that all students learn in the same way. Some students will learn best through lectures, but others will learn best far away from the hustle and bustle of lecture theatres or even seminar groups.

The authorities need to keep in mind that many people with Asperger syndrome dream of a planet where they are the only human being, where there are no interruptions, where events happen with regularity and predictability. Many pine for the lifestyles that were adopted by monks in monasteries, where a calm tranquility allowed for routines in domestic life combined with solitary work. If universities want students with Asperger syndrome to come to study, they need to accept that all that matters is that the person is learning.

How they learn should not be the purview of the university governing body. *What* they are examined in is a reasonable area for universities to legislate on, and the test of the student will still be in exam performance, alongside all other students. The student with Asperger syndrome may require a quiet room away from the hundreds of other students in which to take his or her exam papers. Thankfully, many universities now have a Disability Resource Centre with specialists in Asperger syndrome, who can help assess what such students need in order to ensure that they enjoy their years in college and fulfil their potential.

Prenatal screening and diagnosis: potential benefits and dangers

Let us return to the main focus of this chapter: diagnosis. What will have become apparent is that diagnosis is still based on behavioural criteria. This is true for most of psychiatry, where we still do not have diagnostic biological markers for most conditions. (The exceptions to this are some of the learning disabilities, such as Down syndrome, and some of the dementias.)

There are some questions about how biological markers, if they were available to detect autism or Asperger syndrome, would be used or abused. In the final section of this chapter, we explore some of the ethical issues this raises. If it were possible to diagnose purely on the basis of genes or proteins, for example, this could open the door to *prenatal* diagnosis, or at least prenatal *screening*. Just as it is now possible to screen for Down syndrome using the ‘triple test’ (a blood test from the pregnant mother) or to detect Down syndrome using *amniocentesis* during pregnancy (during which some of the amniotic fluid in



which the foetus is bathed is sampled using a long needle, because this fluid contains lots of cells from the foetus itself), so it might one day be possible to screen or detect autism or Asperger syndrome from a maternal blood test or foetal amniotic fluid test.

Currently the results of such tests for Down syndrome are used to enable parents to make a decision about whether to continue with the pregnancy or opt for a termination. The fear from at least some of the higher-functioning individuals with autism or with Asperger syndrome is that these methods, if and when they become available, could lead to *prevention* of autism or Asperger syndrome, or to some form of *eugenics* (genetic and social engineering). Understandably, those who feel autism or Asperger syndrome is a central part of their identity, or who feel it is responsible for their strengths (not just their difficulties), feel this is a major threat to their very existence, and that society would lose potentially valuable genes from the gene pool.

We will see in Chapters 5 and 6 how the genes for autism and Asperger syndrome not only lead to difficulties but (in some of the other family members, or even in those with a diagnosis) may lead to talents in areas such as exceptional attention to detail, ability to focus deeply to develop expertise, extraordinary memory and remarkable ability to detect patterns (useful in fields such as maths, music, engineering, craftsmanship and the ‘hard’ sciences). In this way, the fears expressed by some people with Asperger syndrome are not just about their civil liberties—their right to life—but also about the relationship between the genes underlying autism and Asperger syndrome, and the genes that have enabled human beings to produce great art, science and technology.

On the other side of the argument are parents’ right to choose whether to continue with a pregnancy or not, especially if the child is likely to have major disabilities such as severe learning difficulties. A second argument in support of prenatal screening is the potential for such methods to enable early intervention to be provided, in theory from birth. That is, prenatal screening does not have to lead to termination; it can lead to early intervention.

The latter argument is, for me, one of the most important reasons for pushing ahead with research into prenatal diagnosis, since at present many children (and adults) have to wait far too long to obtain their diagnosis. If the means were available, it could then be determined if early intervention (beginning in infancy) leads to a greater reduction in later difficulties, compared with intervention that begins later. We return to the whole topic of intervention in Chapter 7.

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The other major benefit of research into prenatal factors (e.g., as genes, or hormones produced by the foetus) is to help us understand the fundamental causes of autism and Asperger syndrome. Greater understanding of causal factors is important in any area of science and medicine. The above discussion is necessary to flag up the ethical issues surrounding this area, so that there is proper debate and caution in how science and clinical practice proceed.