The National Autistic Society (NAS) was formed in 1962 on the initiative of a group of parents who were later joined by professionally interested people. Today, the Society has grown into the UK's foremost charity for people with autism and those who care for them, spearheading national and international initiatives and providing a strong voice for autism. The organisation works in many areas to help people with autism live their lives with as much independence as possible.

The NAS:

- runs education and adult centres
- supports local authorities in the development of their own specialist services
- publishes a range of books and leaflets
- has a library that parents and researchers can use by appointment
- runs an autism helpline for parents and carers and people with autistic spectrum disorders
- organises conferences and training programmes
- co-ordinates the work of volunteers on nationwide parent to parent and befriending schemes
- offers specialist diagnosis and assessment services
- encourages research into the causes of autism
- supports local groups and families around the country
- raises awareness and creates a better understanding of autism
- provides consultancy to professionals and organisations working in the field of autism
- offers an accreditation programme for autism-specific education and care services
- runs Prospects, a supported employment service for adults with autistic spectrum disorders

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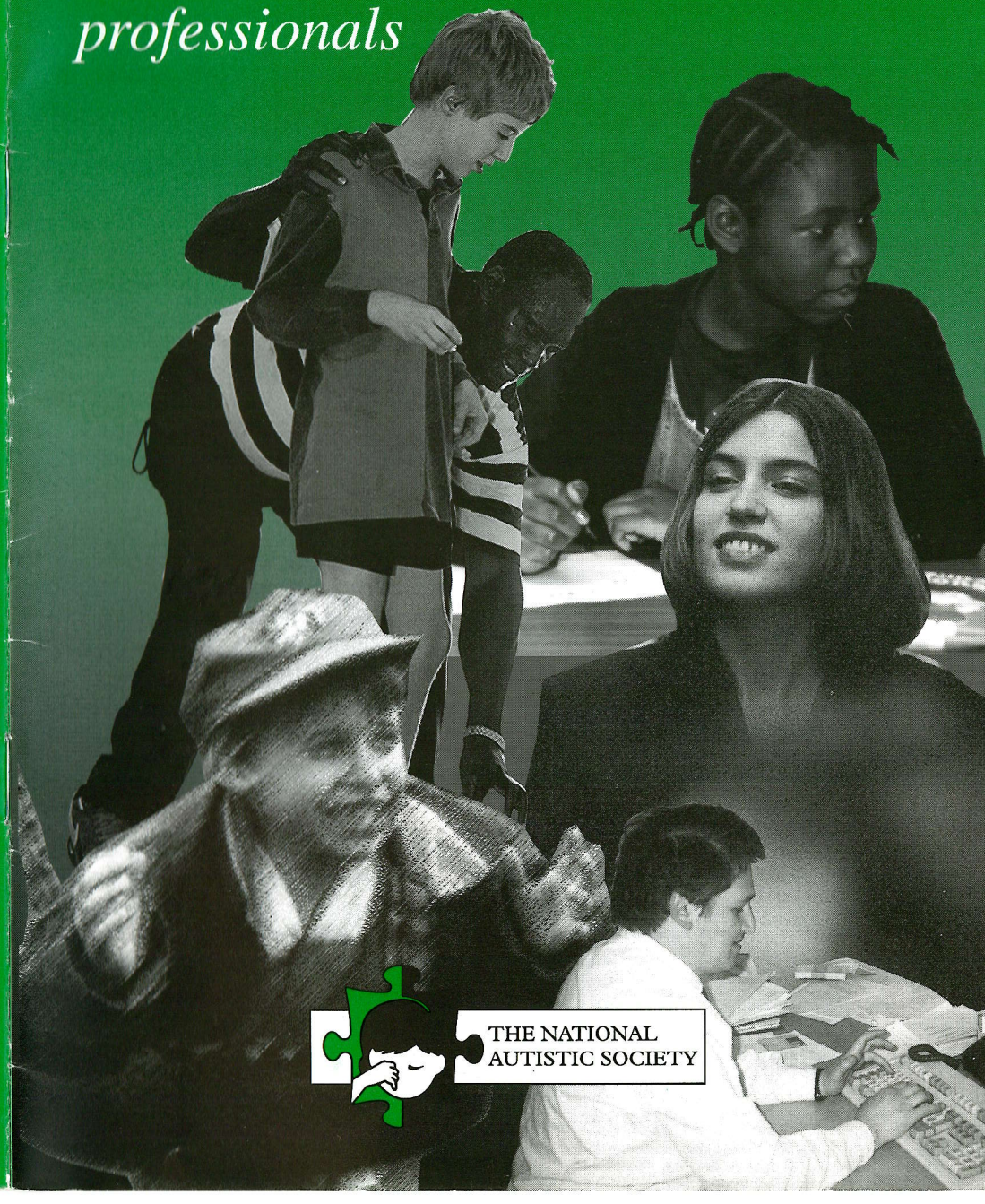
Code 99/1/76

Diagnosis: *a brief guide for health professionals*

OPENING THE DOOR



THE NATIONAL AUTISTIC SOCIETY

Introduction

Autistic spectrum disorder is a complex, developmental disability which can make diagnosis difficult. In this leaflet we present information to help health professionals in recognising the early signs of this disability.

As a parent my experience with diagnosis is that many medical professionals do not recognise an autistic spectrum disorder and do not want to label. As helpful as GPs are, they do not recognise the symptoms and it is to them whom a parent initially turns to.

Helen Birns

In an average list size of 2,000 patients, each GP may have up to 18 people on the autistic spectrum on his/her list.[1] Autistic spectrum disorders may be regarded as a low incidence disorder which, unlike for example hypertension, has no obvious active role for a GP. However, the role of the GP can be vitally important, particularly in the early stages of diagnosis and adjustment to that diagnosis.

Cathie Scothorne, GP

The first diagnosis

Leo Kanner (1943) was the first person to describe and name a pattern of behaviour he observed in a small group of young children, which he termed 'early infantile autism'. Asperger (1944), one year after Kanner's original paper, wrote about another behaviour pattern in older children and adolescents, which, though different in detail, clearly overlapped with Kanner's accounts. Asperger also used the term 'autistic' in relation to the behaviour he saw.

What is the autistic spectrum?

An autistic spectrum disorder is a complex life long developmental disability that affects the way a person communicates and relates to people around them. The autistic spectrum includes the syndromes described by Kanner and by Asperger but is wider than these two subgroups (Wing and Gould 1979).

Many people have a mixture of features from these two syndromes but do not fit neatly into either. The whole spectrum is defined by the presence of impairments affecting social interaction, communication and imagination, know as the triad of impairments. This is always accompanied by a narrow, repetitive range of activities.

[1] Estimated prevalence rate of people with autistic spectrum disorders in the UK is 91 people in every 10,000.

A range of other problems is also commonly found in association with the triad but the three basic impairments are the defining criteria.

Individuals who are considered to be on the autistic spectrum are all very different. The range of intellectual ability extends from severely learning disabled right up to normal or even above average levels of intellect. Similarly, linguistic skills range from those who are mute to those who display complex, grammatically correct speech.

What causes autistic spectrum disorders?

The exact cause or causes of autism have not yet been fully established although research is continuing on a number of fronts. It is evident from research that autism can be caused by a variety of conditions which affect brain development and which occur before, during or after birth. They include, for example, maternal rubella, tuberous sclerosis, lack of oxygen at birth and complications of childhood illnesses, such as whooping cough and measles. Twin and family studies suggest a genetic link in autism but the sites of relevant genes have yet to be identified.

Is there an effective treatment?

An autistic spectrum disorder is a life-long disability; children with the condition grow up into adults with the condition. However, with appropriate intervention early in life, specialised education and structured support, a child can be helped to maximise their skills and achieve their full potential as adults.

The importance of early diagnosis

It is crucial that an autistic spectrum disorder is recognised early in a person's life to enable the most effective intervention and management of the condition. Early diagnosis and intervention is also essential to ensure families and carers have access to appropriate services and professional support.

Certainly the signs are there to be recognised. In most cases the triad of impairments emerge in the first two to three years of life – indeed there are often indications of developmental problems within the first year. However, because autistic spectrum disorders are complex it is easy to miss important clues.

Although the characteristics of autism are generally evident in the first few years of life, the condition can go undetected for many years especially in those who are at the more able end of the spectrum where the signs are more subtle.

When I was diagnosed as having Asperger syndrome a year ago, it brought an overwhelming feeling of relief, together with a deep sense of pain of 25 lost years. Before I was diagnosed, any help I received failed due to a lack of knowledge and understanding of my difficulties. This is one of the reasons why it is vital that a diagnosis of autism or Asperger syndrome is made as early as possible.....A five year old has the potential to make so much more progress than someone who is 25.

William Rice

People at the more able end of the spectrum are often aware that they are different from other people and that they have difficulty in forming or maintaining relationships. If undiagnosed their behaviour can appear odd, which can lead to bullying or teasing at school. Depression, therefore can be a feature as the person gets older.

The triad of impairments

The items making up the triad can be shown in a wide range of different ways. The manifestations vary with the severity of the disability. Changes occur with age, especially in those with higher levels of ability; different aspects of the behaviour pattern are more obvious at some ages than at others.

Education and the social environment can have marked effects on overt behaviour; in a very structured setting, with one to one attention, the autistic behaviour may not be shown in any obvious way. It should also be remembered that all children have their own personality, which affects their reaction to their disabilities.

1 Impairment of social interaction

The most severe form is aloofness and indifference to other people although most enjoy certain forms of active physical contact and show attachment on a simple level to parents or carers.

In less severe forms, the individual passively accepts social contact, even showing some pleasure in this, though he or she does not make spontaneous approaches. Some children or adults with the triad approach other people spontaneously, but do so in an odd, inappropriate, repetitive way and pay little or no attention to the responses of the people they approach.

Among the most able adolescents and adults, the social impairment may have evolved into an inappropriately stilted and formal manner of interaction with family and friends as well as strangers.

It has been suggested that the problem underlying social impairment is lack of the in-built ability to recognise that other people have thoughts and feelings – the absence or impairment of a so-called 'theory of mind'.

2 Impairment of social communication

A lack of appreciation of the social uses and the pleasure of communication is always present in one form or another. This is true even of those who have a lot of speech, which they use to talk 'at' others and not with them. A lack of understanding that language is a tool for conveying information to others is another typical example of the communication impairment. Some are able to ask for their own needs but have difficulty in talking about feelings or thoughts and in understanding the emotions, ideas and beliefs of other people.

Many are unable to convey or comprehend information by using gesture, miming, facial expression, bodily posture, vocal intonation etc. Some more able people do use gestures but these tend to be odd and inappropriate. Those with good vocabularies have a pedantic, concrete understanding and use of words, an idiosyncratic, sometimes pompous choice of words and phrases, and limited content of speech. Some verbal autistic people are fascinated with words and word games but do not use their vocabularies as tools of social interaction and reciprocal communication.

3 Impairment of imagination

In children, inability to play imaginatively with objects or toys or with other children or adults is an outward manifestation of this impairment. A tendency to select for attention minor or trivial aspects of things in the environment instead of an imaginative understanding of the meaning of the whole scene is often found (eg attending to one earring instead of the whole person, a wheel instead of the whole toy train).

Some of these children display a limited range of imaginative activities, which may be copied, for example, from TV programmes, but they pursue these repetitively and cannot be influenced by suggestions from other children. Such play may seem very complex, but careful observation shows its rigidity and stereotyped nature. Some watch soap operas or read particular types of books, such as science fiction, but the interest is limited and repetitive.

Some confuse fiction and reality and tell rambling stories they seem to believe are true. Some do not know the difference between dreams and

reality. Many lack understanding of the purpose of any pursuits that involve comprehension of words and their complex associations, eg social conversation, literature, especially fiction, subtle verbal humour (though simple jokes may be enjoyed). There is a consequent lack of motivation to indulge in these activities, even if the necessary skills are available.

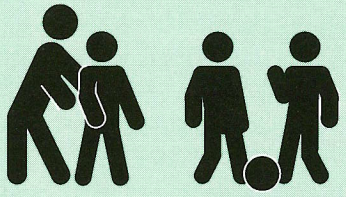
In adults, the proper development of imagination is shown in the ability to use past and present experiences (both one's own and other people's, the latter from personal social communication and books, plays and films) in order to predict consequences of actions and make plans for the short and long term future. This aspect of the mature imagination is conspicuously lacking in people with autistic spectrum disorders, whatever their level of ability. The consequence of the impairment of imagination is a very narrow range of repetitive activities or special interest.

These can take simple or complex forms. Children of higher levels of ability tend to show more complex routines. The following are only some examples of stereotyped activities. The possible variations on this theme are endless. Simple stereotyped activities include:

- flicking fingers, objects, pieces of string
- watching things that spin
- tapping and scratching on surfaces
- inspecting, walking along and tracing lines and angles
- feeling special textures
- rocking, especially standing up and jumping from back foot to front foot
- tapping, scratching or otherwise manipulating parts of the body
- repetitive head banging or self injury
- teeth grinding
- repetitive grunting, screaming or other noises.

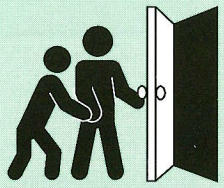
These pin people illustrate some ways in which autism is displayed

Displays indifference

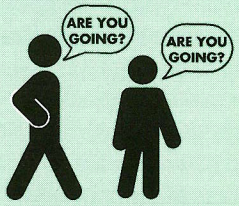


Joins in only if adult insists and assists

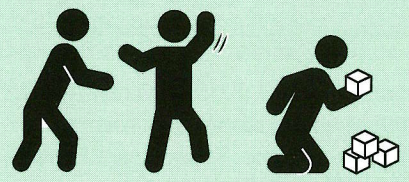
Handles or spins objects



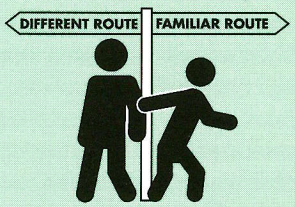
Indicates needs by using an adult's hand



Echolalic – copies words like parrot

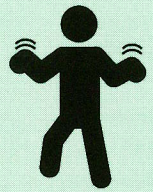


Does not play with other children



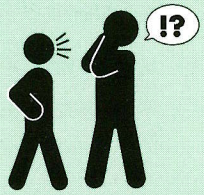
Variety is not the spice of life

Inappropriate eye contact

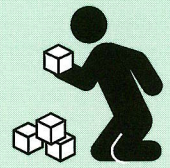


Bizarre behaviour

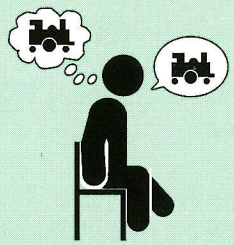
Inappropriate laughing or giggling



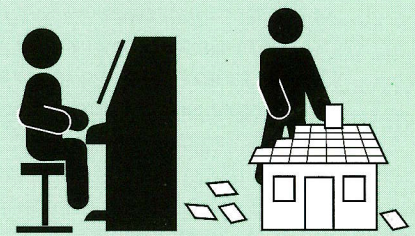
One-sided interaction



Lack of creative, pretend play



Talks incessantly about only one topic



But some can do some things very well, very quickly but not tasks involving social understanding

Complex stereotyped activities involving objects include:

- intense attachment to particular objects for no apparent purpose
- a fascination with regular repeated patterns of objects, sounds
- arranging objects in lines or patterns
- the collection, for no apparent purpose, of large numbers of particular objects, such as plastic bottles, pebbles, or the tops from tubes of Smarties.

Complex stereotyped activities involving routines include:

- insistence on a lengthy bedtime ritual
- repetition of a sequence of odd bodily movements.

Complex verbal or abstract repetitive activities include:

- fascination with certain topics, such as electricity, astronomy, birds, train timetables, even specific persons, asking the same series of questions and demanding standard answers.

Other features that may be present

In addition to the essential diagnostic criteria listed, there are some other features that are common but not essential for diagnosis.

1 Problems affecting formal language (in addition to the essential communication impairments mentioned above) including difficulties in comprehension and use of speech as in developmental language disorders

2 Odd responses to sensory stimuli, such as hypersensitivity to sound, fascination with visual stimuli, dislike of gentle touch but enjoyment of firm pressure etc.

3 Poor motor co-ordination including clumsiness, odd gait and posture.

4 Over or under activity

5 Abnormalities of mood, such as excitement, misery

6 Abnormalities of eating, drinking, sleeping

7 Physical disabilities, such as epilepsy, sensory impairments, Down's syndrome or any other.

8 Additional developmental disorders affecting language, reading, writing, number work etc

9 Psychiatric conditions, such as depressions, anxiety, catatonia, 'psychotic states'.

10 Disturbance of behaviour such as aggression, self injury, running away, screaming etc.

11 Special skills. About 10% of children with autistic spectrum disorders have some special skill at a much higher level than the rest of their abilities – for example, music, art, numerical calculations or jigsaw puzzles.

Some have a remarkable memory for dates and things that particularly interest them. For example, people with Asperger syndrome often develop an almost obsessive interest in a hobby or collection. Usually their interest involves arranging or memorising facts about a specialist subject, such as train timetables, Derby winners or the dimensions of cathedrals.

Problems of diagnosis

The difficulties in diagnosing autistic spectrum conditions experienced by clinicians arise mainly for the following reasons.

The non-specific and variable nature of the autistic spectrum does not make it easy for a parent to identify a problem. It is easier to describe a pain in the leg than a feeling that something is not quite right.

Cathie Scothorne GP

1 Variation in manifestations of features

As described above, the manifestations of the diagnostic criteria vary widely. Examples are listed briefly in the ICD 10 research criteria manual and the DSM IV manual, but they cannot cover the whole range. For example, most diagnosticians would classify insistence on doing nothing but lining up toy cars in a precise sequence as an elaborate repetitive routine, but some may not recognise insistence on verbally retelling facts about cars, regardless of the social situation, as another manifestation of the same phenomenon.

Most recognise social impairment in a child who ignores people but is fascinated by trains, but some fail to see that a child who wants to have friends but approaches other children in bizarre, inappropriate, repetitive ways is also socially impaired. Eye contact is a particular pitfall. Visual avoidance is easy to detect; it is harder to recognise inappropriate use of eye contact as equally diagnostic.

2 Differentiation from other conditions

Language impairments resembling those in the specific disorders of language development, or poor co-ordination as in dyspraxia, may be diagnosed but the presence of the autistic triad of impairments may be overlooked. The many other conditions that can occur together with an autistic disorder can take attention away from the triad, which can be completely missed. This can also happen with adults contacting the psychiatric service who were never diagnosed as autistic during childhood. It is particularly likely to happen with those who are most able.

In differential diagnosis, the task is to decide whether the triad of impairments is present, whether or not there is some other condition as well.

3 Confusion concerning sub-groups

The ICD 10 system divides 'pervasive developmental disorders' into sub-groups, including 'childhood autism' and 'Asperger syndrome'. DSM IV has similar sub-groups, though the names are slightly different.

Some clinicians are reluctant to diagnose an autistic disorder if the clinical features shown by an individual do not fit any of the sub-groups. The confusion is exacerbated by the fact that ICD 10 and DSM IV define Asperger syndrome differently from Asperger himself. Some clinicians use the international systems, while others use criteria based on Asperger's own description. Faced with these diagnostic difficulties, some take refuge in the category of 'pervasive developmental disorder not otherwise specified'. This vague diagnosis may lead to problems when the parents try to obtain appropriate education for their child.

There are more children with a mixture of features of different sub-groups than there are 'pure' cases (Wing and Gould, 1979). It is much more important for clinicians to diagnose the presence of an autistic spectrum disorder than to worry about the sub-group. Research workers may decide to study only 'pure' cases, but clinicians should be concerned with the needs of the individual they are seeing.

As professionals we have a responsibility to be well informed so that, after listening carefully to the anxieties presented to us, we may act appropriately.

However, we must all be aware of our limitations and such reassurance must be based on an adequate history and examination and assessment as in all our medical practice. GPs must have access to a secondary referral centre with proven expertise in the assessment of child development. Many parents feel obliged to seek a tertiary referral because of lack of local expertise.

Cathie Scothorne, GP

4 Relying on observation of the child

There is a common source of diagnostic confusion, particularly with more able children. In structured situations with experienced staff and close supervision, such as during psychological testing, or in a well organised classroom, an individual with an autistic disorder may show no obvious signs of autistic behaviour, and the subtle signs can easily be missed. The best setting in which to observe autistic behaviour is one that is unstructured and in which the individual is unsupervised and undirected. For children, free play in the school playground with no staff supervision is particularly likely to bring out the autistic pattern.

5 Blaming the parents

The social impairment in autism is, in most cases, present from birth or early in life. This inevitably leads to major difficulties in the mother-child relationship. The problems are due to the autism in the child but mothers tend to blame themselves before the diagnosis is made. Some clinicians always look for psychopathology in the mother whenever they see a child with behaviour problems who looks physically normal. When they see how poorly the child relates to their mother, they take this as corroboration of their diagnosis. This view confirms and exacerbates the mother's feeling of guilt and does nothing to help the child or the family. The distress is made even worse if the clinician decides that the mother is causing or inventing the child's problems and diagnoses 'Münchhausen's syndrome by proxy'.

Any parent of a child with a problem will have anxieties: about the nature of the problem and its short and long term implications; about day to day management: and about reactions of siblings, relations and friends. This may well be compounded by feeding and sleeping problems so prevalent in children on the autistic spectrum. These anxieties need to be acknowledged, and, where possible, practical advice given. Although a clear-cut diagnosis may not be possible, this does not mean that professionals are unable to support and advise the family in behavioural management while assessment is in progress.

Cathie Scothorne, GP

The process of diagnosis

The pitfalls listed above can be avoided only if the clinician always bears in mind the possibility of an autistic spectrum disorder when faced with a worried and puzzled parent describing odd behaviour in their child.

Autistic spectrum conditions are developmental disorders caused by physical abnormalities in parts of the brain. In most cases, complex genetic factors are important in the causation. The pattern of abnormal development unfolds over time. The correct diagnosis can be made only be taking a detailed developmental history from infancy and obtaining equally detailed information concerning behaviour in different settings. The right questions must be asked in a systematic fashion. A diagnosis cannot be made solely by observing or testing in the artificial setting of a clinic.

The results of psychological assessment by an experienced psychologist are invaluable for deciding the child's educational needs. The profile of sub-tests may back up the diagnosis, but the absence of the typical discrepant pattern does not rule out an autistic disorder. Even the simpler 'theory of mind' tests are often passed by more able, older children and adults, despite their lack of empathy with others in real life.

Parents often feel bewildered by the involvement of many professionals and are often unsure who to approach for information. Many return to their GP at this stage. The GP should be in the position of being fully informed as to the progress of the assessment by liaising with the key worker. Parents will then remain confident that their case is in good hands.

Cathie Scothorne, GP

When diagnosing an autistic disorder, there is no substitute for a good history taken with the aid of a schedule that lists, systematically, the questions to be asked. One example is the Diagnostic Interview for Social and Communication Disorders (DISCO) being developed at The Centre for Social and Communication Disorders. When using such a schedule, the parents must be allowed time to talk about things that concern them, although the interviewer must also ensure that the right questions are asked.

Time taken to listen to the parents' story with attention and interest gives a real understanding of the child and helps to establish the foundation for a good relationship with the family.

Many families feel that they are left dangling in mid-air, all management plans being delayed until a firm diagnosis is made. Achieving a diagnosis then becomes an overwhelming priority for the families since it appears as if it is the golden ticket to all the treatment/management programmes the child may require. Imagine how it must feel to believe that you are being denied a diagnosis, and, by implication, the route to help for your child. It is easy to understand why many parents feel relieved when a diagnosis is made. It may not be until later that the full implications being to take their toll.

Cathie Scothorne, GP

It is essential to listen to the person with autism and Asperger syndrome, whether or not they have been diagnosed, for they are the real experts, living with the problems of being autistic, every moment of every day of their lives. Until I was diagnosed, professionals failed to listen to me, and dismissed my difficulties, interacting with others and my very limited diet, as phases, that I would grow out of.

William Rice

Help and training

If a local expert cannot be found, professionals can contact the NAS or the Centre for Social and Communication Disorders. They will be able to advise about the nearest source of expertise and help.

The Centre which is a service provided by the NAS offers training for professionals in the complexities of making a diagnosis and offers specific training on the use of the Diagnostic Interview for Social and Communication Disorders (DISCO).

The Centre for Social and Communication Disorders
Tel 0181 466 0098
(From June 1999: 020 8466 0098)

The NAS provides a wide range of services. This includes specific training for GPs through its training services department as part of the GP tutorial systems.

NAS Training 0115 911 3363

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http://www.oneworld.org/autism_uk/

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Email: autismhelpline@nas.org.uk

Fundraising: 0171 903 3522
(From June 1999: 020 7903 3522)

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(From June 1999: 020 7903 3599)
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Press: 0171 903 3593
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I saw one of my GPs in March 1996 and received the usual 'You're shy, quiet and lack confidence, it's a phase, you'll grow out of it'. This phase had now lasted more than 20 years..... The words 'I told you so', can never compensate for those 25 lost years.

William Rice

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