Autism spectrum disorders

The term autism spectrum disorder refers to a constellation of symptoms involving impairments in social interaction and communication, and restricted, repetitive behaviours and interests. This article highlights the disorder among professionals who may come into contact with people with autism spectrum disorder.

Public awareness of autism spectrum disorder has been increasing, leading to a need for professionals to have better knowledge and training in this area to aid early diagnosis. This article gives an overview of the historical background and terminology, epidemiology, aetiology, clinical features, assessment and interventions available for autism spectrum disorder.

Historical background and terminology

'Early infantile autism' (typical autism) or Kanner syndrome was first described by Leo Kanner in 1943 when he wrote about 11 children whom he said had 'fascinating peculiarities'. Shortly afterwards, in 1944, Hans Asperger described a group of high functioning patients, similar in many other ways to Kanner's patients, who were later diagnosed as having Asperger syndrome. The term autistic spectrum disorder was first suggested by Wing and Gould (1979) to describe children with impairments in social interaction, communication and imagination (which they named the 'triad' of impairments) as well as a repetitive, stereotyped pattern of activities. Although the difficulties experienced by these children did not exactly match Kanner's earlier description, they were all thought to fall within a broader 'autistic spectrum'.

Until the 1960s, autistic children were often misdiagnosed as having schizophrenia, but later research (Rutter et al, 1967; DeMeyer et al, 1972) confirmed that children with the behaviours described by Kanner could be reliably distinguished from children with learning disabilities or other psychiatric disorders.

There are currently two established classificatory systems for mental disorders: the *International Classification of Diseases* (ICD-10) published by the World Health Organization (1992) and the *Diagnostic and Statistical Manual of Mental Disorders* (DSM-IV), the classification system developed by the American Psychiatric Association (1994). The term pervasive developmental disorder is now incorporated in both ICD-10 (World Health Organization, 1992) and DSM-IV (American Psychiatric Association, 1994) as an umbrella term to describe a group of disorders characterized by qualitative abnormalities in reciprocal social interactions and in patterns of communication, and by a restricted, stereotyped, repetitive repertoire of interests and activities. The term autism spectrum disorder is currently favoured in the UK and is the one used in the National Autism Plan for Children (National Initiative for Autism Screening and Research, 2003).

Epidemiology

Several epidemiological studies have noted an increase in the prevalence of autism and other autism spectrum disorders over the decades. In 1966, Lotter published the first results of an epidemiological study of children with the behaviour pattern described by Kanner (1943), which gave an overall prevalence rate for autism of 4.5 per 10 000 children. Wing and Gould (1979) found a similar rate for typical (Kanner) autism of 5 for 10 000 in children with IQ less than 70 but this rose to 20 per 10 000 when children thought to fall within the broader 'autism spectrum' were also included. By 2001, the reported prevalence rate for (typical) autism was 16.8 per 10 000 and 45.8 per 10 000 for other pervasive developmental disorders (Chakrabarti and Fombonne, 2001). A more recent study of children aged 9–10 years reported a rate of 38.9 per 10 000 for childhood autism, 77.2 per 10 000 for other autism spectrum disorder and a rate of 116 per 10 000 for autism spectrum disorder overall (Baird et al, 2006). All studies have indicated a higher male preponderance with male to female ratio of 4:1 on average (Whiteley et al, 2010).

Aetiology

No one common cause for all cases of autism spectrum disorder has yet been identified. This is mainly because of the variation of phenotypic presentation and complexity in identifying the causative genes. A number of causative factors have been implicated and will now be briefly mentioned.

Research into genetic causes has recently intensified. The British twin study quoted concordance rates in monozygotic twins as 92% compared to 10% for dizygotic twins, for a broader spectrum of related cognitive or social abnormalities in turn suggesting a genetic cause for autism spectrum disorder symptoms (Bailey et al, 1995). The mode of inheritance appears to be via multiple interacting genes and the heritability of liability to autism is greater than 90%. In other words, 90% of the differences between autistic and non-autistic individuals is thought to be a result of genetic effects (Freitag, 2007).

Advanced paternal age and maternal age have been shown to be associated with an increased risk of having

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offspring with autism spectrum disorder, possibly because of de novo spontaneous mutations and/or alterations in genetic imprinting (Croen et al, 2007).

In some cases, autism spectrum disorder can be associated with an identifiable syndrome, for example, fragile X syndrome is the most common genetic cause of autism in males (Rogers et al, 2001). Other conditions associated with autism spectrum disorder include neurocutaneous disorders like tuberous sclerosis, phenylketonuria, fetal alcohol syndrome, Angelman syndrome and Rett syndrome (Johnson and Myers, 2007).

Clinical features

Autism spectrum disorder encompasses impairments in three core domains which include social skills deficits, communication difficulties and restrictive, repetitive and stereotyped behaviours, interests and activities. However, the degree of deficits may vary extensively.

Many parents first become concerned about their child between 15 and 18 months of age but formal diagnosis may be delayed for a number of months or even years afterwards (Howlin and Moorf, 1997). Autism spectrum disorder can also go unnoticed until school age where deficits may then be noticed in the child's interaction with peers and other differences between the child and his/her peer group may become more apparent.

Social skill deficits

Children with autism spectrum disorder have deficits in connecting with others and sharing complementary feeling states. In the first year of life this may present as inability to point to request an object, which is followed by looking alternatively between the object of interest and the parent, to share the social experience. It may also present as inability to turn consistently to respond to one's own name. As the child grows older there may be difficulties noted in playing with other children which can result in inappropriate attempts at joint play manifesting as aggressive or disruptive behaviour. Children with autism spectrum disorder are often happy playing on their own, make poor eye contact, ignore parents' attempts for attention and may show extreme reaction to invasion of personal space. They have deficits in recognizing the emotional states of others. The neurotypical child responds to novel situations by mimicking the facial expressions of the mother, whereas children with autism spectrum disorder have limited ability to do this and are easily overwhelmed by social and other novel stimulation. They may be unable to relate to others and are unable to follow current trends, for example children's dressing sense, style of speech or interests.

Communication difficulties

This can range from muteness to different degrees of deficits in speech. There may be a lack of desire to communicate and speech may not be fluent. The difference in speech can be noticeable from the first year of life, and may include reduced babbling, use of unusual gestures and diminished responsiveness. In the older age groups, speech is often found to be stereotypic. Echolalia may be present and there may be a monotone quality. Facial expressions, movements and gestures may not always match what the child is saying. Children with autism spectrum disorder may reverse pronouns, for example, the child may refer to himself using his proper name or using 'you' or 'he'. Some children use single 'giant words' like 'whatsit? Idontknow', but may be unable to combine words in novel or original phrases or sentences that convey true meaning (Johnson and Myers, 2007).

Restricted, repetitive and stereotyped patterns of behaviour, interests and activities

These include motor mannerisms such as finger flicking, habitual toe walking and stereotypes like hand flapping, finger movements or rocking. There may also be self-injurious behaviour, e.g. biting and hitting. Children with autism spectrum disorder may have an increased degree of interest in certain topics which they tend to know in extreme detail, e.g. train timetables, types of aeroplanes. Some of these children prefer spending time arranging their toys in a particular order rather than engaging in imaginative play. They can also find it difficult to cope with changes in their routine and even a slight change in mealtime, bedtime, dressing or taking a bath can upset them easily.

Other clinical features

Regression

Regression in acquired skills is well documented in autism spectrum disorder (Baird et al, 2008). Regression in language skills can be seen in up to 30% of children with narrowly defined (typical) autism. There can also be regression noted in gestural communication or social skills as well.

Imagination and creativity

Children with autism spectrum disorder may have deficits in their ability to play imaginatively and may lack creativity.

Response to sensory stimuli

There may also be evidence of unusual responses to sensory stimuli, for example the child may not be able to tolerate wearing certain types of materials, may complain about labels on clothes or may be overtly sensitive to certain smells, the texture of food or loud noises.

Assessment

Early identification is thought to be beneficial as effective interventions need to be focussed and start early (Dawson and Osterling, 1997). A few tools have been developed to screen the population for autism spectrum disorder. Screening questionnaires include the Modified Checklist for Autism in Toddlers (M-CHAT) (Robins et al, 2001),

the Early Screening of Autistic Traits Questionnaire (Dietz et al, 2006) and the First Year Inventory (Watson et al, 2007). Initial data on the M-CHAT and its predecessor, the CHAT (Baron-Cohen et al, 1992), on children aged 18–30 months suggest that it is best used in a clinical setting and that it has low sensitivity (many false negatives) but good specificity (few false positives) (Landa, 2008).

In the UK, the National Autism Plan for Children (National Initiative for Autism Screening and Research, 2003) does not recommend primary screening for autism spectrum disorder by the use of tests applied to the whole population. Instead it recommends regular reviews of a child's development in the pre-school years, e.g. by a health visitor who is trained to be aware of the 'alerting signals' for autism spectrum disorder. Some absolute indicators for referral to a specialist for a general developmental assessment include:

- No babbling, pointing or other gesture by 12 months
- No single words by 18 months
- No two-word spontaneous phrases by 24 months
- Any loss of any language or social skills at any age (Filipek et al, 1999).

The initial presentation of a child with (suspected) autism spectrum disorder could be to any professional in primary care, education or social services. Therefore it is important that front-line professionals are aware of and trained in identifying the possible features of autism spectrum disorder. This primary assessment could be useful in identifying the nature and severity of the problem. If autism spectrum disorder is suspected, the child should be referred for a specialist assessment, the aim of which is to gather and record information to enable a diagnosis and provide multidisciplinary management. In many areas of the UK, multi-agency referral and assessment pathways have been developed or are in the process of being developed for children suspected of having autism spectrum disorder (e.g. Salmon et al, 2006). As autism spectrum disorder is a 'lifelong' label, it is important that a thorough multi-professional assessment is conducted before arriving at the diagnosis. Such an assessment is necessarily comprehensive and may involve a number of the following professionals: educational and/or clinical psychologist, specialist teacher, speech and language therapist, community paediatrician and/or child and adolescent psychiatrist, and occupational therapist and/or physiotherapist.

The initial step in the assessment is to coordinate information from various settings in order to avoid repetition for the child and the family. This could include collating reports submitted previously for the purposes of assessing a child for a statement of special educational need. Such reports might include advice from parents, school or early educational setting, speech and language therapy, medical, psychological and social services. Information from the paediatrician which includes physical and neurodevelopmental examination helps to rule out other possible medical causes for the child's presentation.

The specialist leading the assessment will then usually meet with the child and interview the parents to obtain an autism spectrum disorder-specific developmental history which includes family history and a description of the current functioning of the child.

There are various autism spectrum disorder-specific diagnostic instruments which are used in clinical practice to aid diagnosis. These include the Autism Diagnostic Interview-Revised (ADI-R) (Lord et al, 1994), the Diagnostic Interview for Social and Communication Disorders (DISCO) (Leekam et al, 2002) and the Developmental, Dimensional and Diagnostic interview (3Di) (Skuse et al, 2004). Observation of the child in various settings such as the clinic, the school and at home is an essential next step. The Autism Diagnostic Observational Schedule-Generic (ADOS-G) (Lord et al, 2000) is a reliable diagnostic instrument which is often used to supplement clinical history and informal observations. Through the use of structured activities and materials, the ADOS-G provides standard contexts in which social interactions, communication and other behaviours relevant to autism spectrum disorder can be observed. Based on the child's individual needs, further assessments by a speech and language therapist, occupational therapist and/or physiotherapist may also be helpful.

The medical assessment also includes identifying comorbidities such as epilepsy, attention deficit hyperactivity disorder, developmental coordination disorder, visual impairment, hearing impairment, mental health and behavioural problems. When there is a high degree of suspicion, aetiological factors like tuberous sclerosis, fragile X syndrome and differential diagnoses like Rett's syndrome should be investigated.

The National Autism Plan for Children (National Initiative for Autism Screening and Research, 2003) sets a standard for feedback to be provided to the family within 17 weeks of the start of the multiagency specialist assessment. In practice, this may be difficult to achieve, especially in areas where the different professionals involved in the autism spectrum disorder assessment work in isolation from each other.

Interventions

There are many different interventions and treatments available for autism spectrum disorder, and the evidence base is stronger for some than for others (Francis, 2005). A broad overview of interventions available is given.

Interventions involving family support and information

Following the diagnosis of autism spectrum disorder, families should be informed about the availability of local support groups and training opportunities. These include group sessions to improve parenting skills and individual support from a trained professional, for example, a parent support worker who specializes in working with families in which there is a child with autism spec-

trum disorder. Parent-mediated interventions are commonly used to help families interact with the autistic child, to promote development and offer practical advice and support to parents.

Treatment programmes that seem to be the most widely effective have the following features in common (Howlin et al, 1987; Schopler et al, 1990):

- Use predictability, routine and consistency as important elements in the teaching of new skills and reducing problem behaviour
- Make use of visual strategies to emphasize meaning
- Offer small group social opportunities for children and their families
- Teach an understanding of the role played by the fundamental deficits in autism in both causing and maintaining behavioural problems
- Use a functional approach to problem behaviours
- Build on basic behavioural strategies such as prompting and shaping techniques, to develop more complex skills and the systematic breakdown of complex tasks into their component but meaningful tasks to enhance learning
- Use naturally occurring reinforcers (i.e. resulting from the successful completion of the task itself, or using the child's special or obsessive interest) (adapted from National Initiative for Autism Screening and Research, 2003).

Interventions to improve communication

Many children with autism spectrum disorder are referred to a speech and language therapist who can provide assessment of and interventions for pragmatic and social communication difficulties.

Here again, strategies such as offering additional support to verbal social initiations by using tactile prompting or visual reinforcement are adopted.

Behavioural and psychological interventions

Based on the individual child's need, this could vary from an intensive behavioural programme to improve overall functioning and altering outcome to addressing specific behavioural difficulties such as sleep disturbance or improving positive behaviour.

Other non-pharmacological interventions

There are other forms of therapies available which include auditory integration training to target the discomfort experienced by children with autism spectrum disorder, e.g. listening to certain sound frequencies, music therapy, occupational therapy (Scottish Intercollegiate Guidelines Network, 2007).

Research into biomedical and nutritional interventions is ongoing. There is currently insufficient evidence to suggest that diets free in gluten or casein (Millward et al, 2004) or diets rich in vitamin B and magnesium are of benefit to children with autism spectrum disorder (Nye and Brice, 2004).

Research has supported the effectiveness of a range of non-pharmacological intervention approaches, but there is no strong evidence that one approach is more effective than the others (Dawson and Osterling, 1997).

Pharmacological interventions

Pharmacological interventions may be necessary to treat comorbid psychiatric or neurodevelopmental disorders or as a short-term intervention for specific severe symptoms, e.g. aggression. They should always be used in combination with other modalities of treatment and as a part of an integrated multidisciplinary care plan. Drugs frequently prescribed for children with autism spectrum disorder include: risperidone which is useful for the short-term treatment of significant aggression, tantrums or self injury (McCracken et al, 2002); methylphenidate as a treatment for attention difficulties and hyperactivity (Research units on Paediatric Psychopharmacology Autism Network, 2005), fluoxetine for repetitive behaviour and melatonin for sleep problems.

Education

Schools and other specialist educational placements play a major role in the management of autism spectrum disorder children. Many children with autism spectrum disorder attend mainstream school with extra individual help. Many schools in the UK undertake educational assessments and provide any extra educational support that is required. Children with special educational needs are supported via an individual educational plan which helps staff to teach and review the progress of these children. Many children with autism spectrum disorder have co-existing motor difficulties affecting handwriting skills and playing sports, and may also have dyslexia which will need addressing.

Prognosis

There is no cure for autism spectrum disorder (Levy et al, 2009). However, there are reports that 3–25% of children recover from autism spectrum disorder after intensive therapy or spontaneously (Helt et al, 2008). The outcome in school, work or social functioning is varied: 75% of autistic individuals have a poor outcome while 25% have a better prognosis. Acquisition of language before the age of 6 years and IQ levels above 50 predict a better outcome (Tidmarsh and Volkmar, 2003).

Conclusions

There have been huge developments in the field of autism spectrum disorder in recent years. Public awareness is increasing which in turn is putting huge demands on the health service. It is becoming increasingly important for all health professionals to be aware of early alerting symptoms of autism spectrum disorder. With a wide range of screening questionnaires being available which are easy to administer, health professionals can play an important part in the early identification of autism spectrum disorder and aid better outcomes. BJHM

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KEY POINTS

- Raised public awareness about autism spectrum disorders has led to a need for better professional knowledge and training.
- Epidemiological studies have noted an increase in prevalence of autism spectrum disorders over the decades.
- Early identification is beneficial as effective interventions need to be focused and started early.