

Understanding autism spectrum disorders

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Summary This paper considers a practical overview of the diagnosis and management of autism spectrum disorders, a group of neurodevelopmental disorders typified by childhood autism. It will focus primarily on the presentation of pre-school and school-aged children, as these children often first present to paediatricians in the pathway to care. Reference is made to recent national guidelines for assessment and management, and more controversial aetiological aspects are discussed.

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PRACTICE POINTS

- Observed rates of autism spectrum disorders (ASD) have increased significantly
- Whole-population screening for autism is not recommended
- Diagnosis relies on identification of a particular pattern of deficits
- 90% of cases are idiopathic. Clinical evidence of co-morbid medical conditions such as epilepsy should be sought but not tested for routinely
- Chromosome karyotyping and fragile-X DNA analysis are the only recommended routine special investigations
- Co-morbid behavioural and mental health problems are common
- Behavioural and educational interventions are effective, but no single approach has been shown to be more effective than others
- A multi-agency family care plan approach with clear ASD management strategies for all staff and parents to use is essential

RESEARCH AGENDA

- Further research on early screening and the impact of the diagnosis on carers is needed
- Evidence-based assessment of interventions and aetiology should continue and results should be disseminated

INTRODUCTION

Autism spectrum disorders (ASD) constitute a small but significant proportion of the disorders presenting at child health clinics. Language delay is the most common initial reason for referral,¹ and early diagnosis is key for the planning of interventions and educational provision. At a local level, paediatricians have a definite role to play in multi-agency assessment of children suspected of having ASD.

TERMINOLOGY

First used in 1943 by Kanner, the term 'autism' refers to a relatively narrowly defined group of children with marked deficits in three areas of function (see below). Over the past 20 years, the emphasis has shifted to a broader range of disorders that share the core deficits, but in which the degree of handicap may be less severe whilst service needs are similar. Although most research has been carried out using the narrower traditional diagnostic concept of core autism, the implications seem to apply more broadly.

The term 'pervasive developmental disorder' (PDD) came to be adopted in both the ICD-10 (World Health Organization 1992) and DSM IV (American Psychological Association 1994) as the umbrella term used to cover both core autism and a broader range of autistic-like disorders. Recently, there has been a movement from parents and professionals to replace the term 'PDD' with the term 'autism spectrum disorder' (ASD), reflecting the finding that there is more overlap than difference among subcategories. This paper refers to ASD as synonymous with the umbrella term 'PDD'.

The diagnosis of core autism/childhood autism requires onset before 36 months of age, and involves three key areas of deficit:

- difficulties in reciprocal social interaction;
- impaired communication;
- restricted, repetitive behaviour and interest.

No child with core autism shows all of the characteristics, and some show only one or two symptoms in each key area. Difficulties in these three key areas are described in some detail below.

The DSM IV category PDD-NOS (pervasive developmental disorder – not otherwise specified) is used to indicate a subgroup of ASD where some, but not all, of the criteria for core autism are present.

The term ‘Asperger syndrome’ has become increasingly popular amongst clinicians, parents and affected individuals themselves. It is often used to mark a subgroup at the more able end, in terms of social and communication handicaps, within the spectrum of ASD. Lack of delay in language development is a requirement, and although the ICD-10 recognizes this category, it remains a matter for debate and research to establish how distinct the subgroup is.²

Rett syndrome and childhood disintegrative disorder are two rarer categories which are currently included within ASD as they may mimic the picture of autism.

Rett syndrome is a rare X-linked dominant progressive disorder that affects girls almost exclusively. A developmental plateau at about 18 months is typically followed by gradual loss of speech, purposeful hand use and eventually mobility. Deceleration in head growth is an important clinical sign differentiating Rett syndrome from autism. A recent advance is the finding of mutations in the MECP2 gene,³ which may act as a marker of the disorder.

In childhood disintegrative disorder, children with normal development until the age of 2 years lose acquired skills such as language ability, bladder control and social development. The aetiology is mostly unknown.

EPIDEMIOLOGY

Controversies about the epidemiology of ASD have recently been highlighted by the proposed link between the combined measles, mumps and rubella vaccination, bowel disorders and autism. Current epidemiological evidence does not support this proposed link.⁴

Several large systematic studies have shown an increase in rates of both core autism and ASD. A recent study found rates as high as 16.8 per 10 000 for autism and approximately 60 per 10 000 for the broader category of ASD in a geographically defined population of pre-school children.⁵

The observed increase may be, at least partly, due to more complete ascertainment and a broader definition of ASD, but the matter requires further study.

Boys are three to four times more likely to be affected with autism than girls, and the male preponderance is even more marked at the higher functioning end of the spectrum.

AETIOLOGY

Genetic factors are important as highlighted by twin and family studies. It is likely that the phenotype results from a moderate number of genes, possibly involving 2q and 7q, acting together. The British twin study estimated broad heritability at 91–93%, with a concordance rate of 69% in monozygotic twins. Genetic liability extends to a broader phenotype of similar but milder deficits than those seen in autism.⁶

Psychological deficit theories have failed to identify one primary deficit which could account for all the features associated with the autistic phenotype. Disorder of mentalizing skills, a lack of the so-called ‘Theory of Mind’,⁷ has been implicated as a possible psychological mechanism underlying ASD.

Although the organic basis of ASD is well recognized, a coherent picture of the neuropathological deficits that explains autistic symptomatology has yet to be achieved. Physiological abnormalities affecting the gastrointestinal tract and the immune system have been investigated as possible causal factors, but the only consistent finding is non-specific raised levels of blood serotonin.⁸

CLINICAL CHARACTERISTICS

‘A child in rural Guatemala peers out of the corner of his eye at flags made of leaves and wooden sticks in the same way that a child in suburban Chicago squints at flags made from McDonald’s swizzle sticks and sweetener packets. Yet identical twins with autistic spectrum disorder may vary in IQ scores by as much as 50 points and lead very different lives, with one twin driving his own car to work and the other requiring assistance with basic hygiene.’⁹

The above abstract demonstrates the extraordinary consistency across individuals with autistic disorders in some behaviours, whilst associated skills and deficits may vary considerably.

I. Social deficits

Pre-school children with autism typically lack interest in other children, have a limited range of socially directed facial expressions, have unusual eye contact and are less likely to comfort others or share enjoyment. If social interest subsequently develops, problems persist

in social responsiveness, reciprocity and capacity for empathy.

2. Communication

Many children with autism have severe language delays. Up to 50% have no useful speech, and about 25% have continuing severe receptive and expressive language impairment.¹⁰

In those with language, the most distinctive feature is its unusual social quality. Children with autism have difficulty carrying on conversations that do not relate to their pre-occupations, and their speech is often more of a monologue than a socially directed communication (talking *at* rather than *with* other people). Interestingly, however, this manifestation of shared interest may provide a route to social success.

Language may include idiosyncratic elements such as pronominal reversal (referring to themselves as 'you' instead of 'I'), echolalia ('parroting' of words), stereotypic speech (reliance on stock phrases) and invented words. Abnormalities of pitch and rhythm occur more frequently in those with ASD than in general mental retardation/learning disability.

Deficits in non-verbal communication also occur. A young child with autism who wants a drink may not use eye contact, gesture or vocalization to communicate the request, but may have a tantrum or put the adult's hand on the bottle to pour it. Children with ASD may also display limited ability to imitate or engage in social play (such as peek-a-boo or hide and seek), and have significant difficulties with pretend and spontaneous imaginative play, such as with dolls or action figures. Shared play with other children is also often impaired.

3. Stereotyped interests and behaviours

The third key area includes unusual pre-occupations (e.g. with street signs, drain pipes, birth dates), circumscribed interests (e.g. in train timetables, car prices), attachments to unusual objects (such as toilet brushes), and adherence to non-functional rituals or routines.

Stereotyped movements of the hands and fingers, frequently within peripheral vision, hand flapping and twirling are also common. Sometimes mannerisms are more complex and may involve the whole body.

Some have strong reactions to sensations such as touch, smell or sound. They may have unusual sensory interests or peer at linear patterns.

Some children have a 'need for sameness' and are upset by minor changes in their own routines or environments (e.g. placement of a vase on the table). Repetitive behaviours and play (e.g. lining up video cassettes or cars) are often seen.

ASSOCIATED FEATURES

Cognitive impairment

Eighty percent of people with narrowly defined autism have a full-scale IQ below 70.¹¹ Most individuals with ASD are likely to have an uneven cognitive profile with areas of significant cognitive deficit and areas of relative strength.

Seizures

Further evidence of the organic basis of autism is the high prevalence of seizures in the disorder. At least one-third to one-quarter of individuals with autism will develop epilepsy, with one peak in age of onset of seizures in early childhood and another peak in adolescence.

Behavioural and psychiatric co-morbidity

Many children with ASD have additional difficulties such as severe and frequent temper tantrums, self-injurious behaviour, sleeping problems, toileting problems (sometimes due to sensitivity to the sound of a flushing toilet) and extreme food fads.¹² Co-morbid diagnoses include obsessive-compulsive disorder, attention-deficit hyperactivity disorder, depression, social anxiety and social phobia.

Associated medical conditions

Ten to fifteen percent of cases of autism are associated with identifiable medical disorders.¹⁰ Around 20–60% of individuals with tuberose sclerosis will have autism. The fragile-X anomaly occurs in approximately 4% of individuals with autism.

Deafness is slightly more prevalent in children with autism, and therefore some authors recommend definitive hearing tests in all these children, with the exception of those who speak clearly in sentences.¹³

DIFFERENTIAL DIAGNOSIS

Global developmental delay

As mentioned above, there is a strong association between autism and learning disability (ICD-10: 'mental retardation'). Diagnosis of ASD can be particularly problematic at either end of the intellectual range, despite the observation that profoundly learning disabled individuals without autism show different patterns of behaviour from those with autism.

Communication delay and language disorders

Language delay 'outside the limit of two standard deviations' suggests the diagnosis of a specific developmental

disorder of language. Language disorders may have some symptoms that overlap with ASD, but unusual pre-occupations and rituals are much less common than in autism.

Deafness

Although deaf children may have difficulties in recognizing the emotional states of other people, social impairment is qualitatively different from that seen in autism.

When present, ASD is frequently diagnosed later in deaf children than in other children, because of the confounding effect of 'diagnostic overshadowing'.¹⁴

Severe psychosocial deprivation

Children who have experienced severe institutional deprivation can show a quasi-autistic presentation, although there is usually more social reciprocity.¹⁵

Selective mutism

These children, who are more commonly girls, typically have reciprocal relationships with parents whilst their behaviour is restricted at school. Specific language abnormalities associated with autism (e.g. pronominal reversal) are uncommon.

Acquired aphasia with epilepsy (Landau-Kleffner syndrome)

In this rare syndrome of unknown aetiology, expressive language skills are lost with a relative preservation of general intellectual function and social relationships. Onset is accompanied by epileptic seizures, and although it occasionally mimics autism, differentiation is usually straightforward.

GUIDELINES FOR ASSESSMENT AND SCREENING

The National Initiative for Autism Screening and Assessment (NIASA) has been supported by the Royal College of Paediatrics and Child Health and the Royal College of Psychiatrists to produce the 'National Autism Plan for Children'.¹⁶ The plan provides national guidelines for best practice in screening, investigation, early diagnosis and intervention.

Rather than whole-population screening for autism, NIASA suggests regular opportunities to discuss a child's development with parents as part of 'surveillance' to detect and respond rapidly to any developmental concerns. All involved professionals should be trained in 'alerting' signals of possible ASD, both at pre-school and school age, to detect those who require formal assessment.

Here, a three-stage assessment framework is recommended, with stages 1 and 2 at local level.

Stage 1 is a general developmental assessment for any child with a possible developmental problem, usually under leadership of a community paediatrician. Those in whom an ASD is suspected rapidly proceed to stage 2.

Stage 2 of the assessment process is a multi-agency assessment, usually led by a paediatrician in the case of pre-school children.

Multi-agency assessments should be available in all local areas and include psychometric, educational, communication, language, motor competency, behaviour and mental health assessments of the child.

A specific ASD developmental and family history should be taken. It may be useful to use a semi-structured interview such as the autism diagnostic interview (ADI-R).¹⁷

Focused observations should be taken across more than one setting. This could include tools such as the autism diagnostic observation schedule.¹⁸

A full physical examination should be performed including appropriate medical tests. Choice of tests depends on each child's clinical presentation, but chromosome karyotyping and fragile-X DNA analysis are the only current routine recommendations. Clinical evidence of co-morbid medical conditions such as epilepsy should be sought but not tested for routinely, unless there are symptoms. Other assessments may be required to investigate unusual sensory responses, motor planning and co-ordination difficulties, and self-care problems.

A written report of findings and recommended interventions should be produced and discussed with the parents, and a named key worker appointed.

The local area team may need to refer to a stage 3, tertiary ASD assessment team for several reasons, including a second opinion, diagnostic doubt, complexity and specific advice about treatments.

MANAGEMENT

Open and sensitive feedback about the diagnosis is the first step in establishing a therapeutic alliance with parents. The family should receive information about local parent groups, education and training, as well as information about support services and social benefits.

The National Autistic Society is a well-recognized resource and advocacy group with an excellent directory of books, literature and references (www.nas.org.uk), some written by children and adults with autism (e.g. see Luke Jackson¹⁹). Disability living allowance (DLA) constitutes a cash payment to offset some of the extra costs of disability, and may serve as a marker of increased need.²⁰

Research has supported the effectiveness of a range of intervention approaches,² with the emphasis on behavioural and educational approaches. Involving parents as

'co-therapists' to foster social-communicative interactions may well prevent the development of secondary behavioural problems. Although programmes such as ABA and TEACCH (Treatments and Education of Children, Adolescents and Adults with Autism) have many features in common, there have been few direct comparisons of different teaching strategies.²¹

The 'National Autism Plan for Children' recommends that pre-school children should have access to 15 h/week of appropriate ASD-specific programmes. For pre-school and school-aged children, the Family Care and Individual Education Plans (IEP) ideally include clear ASD management strategies for staff and parents, and access to ASD-specific individual or small group therapeutic educational opportunities as dictated by needs. Close collaboration between school and home is needed to ensure consistency of approach across settings.

Some studies have shown that non-autistic peers can be involved to assist autistic children in gaining social skills in group settings, and such interventions are increasingly used in schools.

There is no definitive evidence to date that pharmacological intervention alters long-term prognosis, although psychopharmacological interventions for specific indications may make the child more amenable to educational interventions.

The child's and the family's needs will change over time and all professional contact should aim to facilitate continuity of care, especially at times of transition.

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