

Autism and intellectual disability: diagnostic and treatment issues

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Estimates of the prevalence of autism and related disorders vary widely but it is clear that the rates are considerably higher than the 4–5 per 10 000 typically cited in early reports¹. A review² of 23 epidemiological studies conducted between 1966 and 1998 indicates that the combined rate for autism and autism spectrum disorders is around 18.7 per 10 000. Figures from the National Autistic Society in the UK³ are even higher, with prevalence rates for all autism-related disorders, including Asperger's syndrome, given as 91 per 10 000. Although the basis of this last statistic is controversial, the number of individuals who fall within the 'autistic spectrum' is clearly far greater than was once thought. Moreover, many of these individuals will have additional intellectual disabilities, and in the UK around 115 000 children and adults with autism have an IQ under 70⁴. Autism is also greatly over-represented amongst individuals with an IQ in the severe to profoundly retarded range, with 50–60% showing the 'triad' of autistic impairments, in language, social interaction and imagination⁵.

DIAGNOSIS

High figures for autism-related disorders have great implications for educational, social and health services. They also pose a challenge to diagnostic services, because of the difficulties that can arise in distinguishing autism-specific symptoms from those associated with very low cognitive functioning. Within the groups with most profound intellectual handicap (IQ below 20) it is frequently near-impossible to determine whether the absence of social and communication skills and the stereotyped and repetitive behaviours are attributable to gross cognitive impairment or to an additional autistic disorder. Indeed, the majority of (ambulant) individuals with this degree of disability will show the typical triad of autistic impairments⁵. It might also be argued that differential diagnosis within this group is somewhat academic, in that the overriding factor determining their care and outcome will be the severity of the cognitive handicap.

Differential diagnosis is much more important for those with a lesser degree of cognitive impairment. Thus, for

individuals with an IQ in the severe to mildly retarded range, the presence of autism will have substantial implications for prognosis and for the nature and intensity of intervention required. Individuals with intellectual disabilities *and* autism are likely to need support that is far more individualized, specialized and structured than do non-autistic individuals of the same IQ level⁶, and even then their prognosis will often be worse. To reach a differential diagnosis the following basic components are required⁷:

- Detailed assessment of cognitive level (verbal and non-verbal) and of receptive and expressive language abilities
- A developmental history covering progress from infancy onwards, preferably obtained with a standardized interview such as the Autism Diagnostic Interview⁸
- Observations of the child in both structured and non-structured settings (again a standardized instrument such as the Autism Diagnostic Observation Schedule Generic⁹ can be very informative)
- Routine screening for medical/genetic conditions
- Consideration of other relevant psychosocial factors.

On the basis of this evidence the clinician, or clinical team, must then consider whether the behaviours recorded are appropriate for the individual's chronological age, mental age and language age. If not, then special attention needs to be paid to problems in play, social skills, communication and behaviours of a stereotyped nature. In cases where another disorder also exists—for example Down's syndrome, fragile X, tuberous sclerosis, or severe physical or sensory impairments—clinicians must also consider whether the developmental and behavioural patterns are characteristic of that disorder. The association between autism and tuberous sclerosis is particularly strong, but autism can coincide with many other conditions and treatment may be seriously delayed if all the child's difficulties are incorrectly attributed to the prediagnosed disorder.

Adherence to the clinical recommendations described above, together with knowledge of both atypical and normal developmental patterns on the part of the diagnostic team, should then make it easy to determine whether the individual meets formal diagnostic criteria, as outlined in Box 1.

TREATMENTS FOR AUTISM

Whether or not autism is associated with additional intellectual impairments, there is no shortage of claims for therapies that are said to improve outcome or even lead to cure or 'recovery'¹⁰. Among such interventions are 'holding therapy', music therapy, scotopic sensitivity training, facilitated communication, auditory integration, intensive physical stimulation or 'conductive education', 'daily life therapy', gentle teaching, the 'options' method, and the Waldon approach. Reports on the extraordinary effects of other therapies, from swimming with dolphins, cranial osteopathy, or being swung around in nets, also make regular appearances in the media¹¹. Unfortunately, few of these methods have been subject to any form of experimental investigation. Amongst those that have, facilitated communication is now generally discredited¹²,

and auditory integration therapy has been found to have no positive effects¹³.

Many different drug and vitamin interventions have also been cited as having a dramatic impact on children with autism. However, again, adequate experimental research is limited and there is little if any information about long-term effects, especially when they are given to young children. Fenfluramine, for example, which in the USA, throughout much of the 1980s was widely used as a treatment of choice for autism, has now been largely withdrawn because of concerns about serious side-effects and long-term damage. Another recent treatment, extensively publicized on the Internet, involves the use of secretin infusions, which are claimed to have almost miraculous effects¹⁴. However, an increasing number of parents are reporting severe adverse effects, and initial results from controlled trials¹⁵ indicate

Box 1 ICD-10 criteria for autism

A Abnormal or impaired development is evident before the age of 3 years in at least one of the following areas:

- (1) receptive or expressive language as used in social communication
- (2) development of selective social attachments or of reciprocal social interaction
- (3) functional or symbolic play.

B A total of at least six symptoms from (1), (2), and (3) must be present, with at least two from (1) and at least one from each of (2) and (3):

- (1) *Qualitative abnormalities in reciprocal social interaction are manifest in at least two of the following areas.*
 - (a) failure adequately to use eye-to-eye gaze, facial expression, body posture, and gesture to regulate social interaction
 - (b) failure to develop (*in a manner appropriate to mental age*, and despite ample opportunities) peer relationships that involve mutual interests, activities, and emotions
 - (c) lack of socio-emotional reciprocity as shown by an impaired or deviant response to other people's emotions; or lack of modulation of behaviour according to social context; or a weak integration of social, emotional, and communicative behaviours
 - (d) lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. lack of showing, bringing, or pointing out to other people objects of interest to the individual).
- (2) *Qualitative abnormalities in communication are manifest in at least one of the following areas:*
 - (a) delay in, or total lack of, development of spoken language that is *not* accompanied by an attempt to compensate through the use of gesture or mime as an alternative mode of communication (often preceded by a lack of communicative babbling)
 - (b) relative failure to initiate or sustain conversational interchange (*at whatever level of language skills is present*) in which there is reciprocal responsiveness to the communications of the other person
 - (c) stereotyped and repetitive use of language or idiosyncratic use of words or phrases
 - (d) lack of varied spontaneous make-believe or (when young) social initiative play.
- (3) *Restricted, repetitive, and stereotyped patterns of behaviour, interests, and activities are manifest in at least one of the following areas:*
 - (a) an encompassing preoccupation with one or more stereotyped restricted patterns of interest that are abnormal in their intensity and circumscribed in nature though not in content or focus
 - (b) apparently compulsive adherence to specific, non-functional routines or rituals
 - (c) stereotyped and repetitive motor mannerisms that involve either hand or finger flapping or twisting, or complex whole body movements
 - (d) preoccupations with part-objects or non-functional elements of play materials (such as their odour, the feel of their surface, or the noise or vibration that they generate).

few differences in short-term outcome between children receiving secretin and those given placebo injections.

Preschool interventions

Of the many different interventions claimed to have a positive impact on outcome for children with autism and/or intellectual disabilities, those with an emphasis on the *early* development of cognitive, play, social and communication skills are better supported by empirical evidence. The Head Start Program, for example, which is widely available across the USA for children at risk of learning and behavioural difficulties, has consistently been found to improve (at least in the short term) cognitive, social and physical development, and to reduce rates of problem behaviours¹⁶. Evaluation of home-based interventions specifically designed for children with autism¹⁷ indicates that if parents can be helped to develop appropriately structured and consistent management strategies during the child's early years, this can enhance social, cognitive and linguistic development and lessen subsequent behavioural troubles. Early specialist educational programmes likewise have positive effects¹⁸. The most successful programmes involve intensive support (of 15 or more hours per week), tend to last at least six months, and require a high adult:child ratio. However, because of questionable methodology and research design, their effectiveness, especially in the longer term, remains doubtful. Progress seems to be enhanced if children begin intervention before 5 years, but there is no information on exactly how early they should begin. Similarly, although some research indicates that IQ and language levels are important predictive variables, with the most handicapped children making least progress¹⁹, other groups have found no relationship between pretreatment level and outcome²⁰.

Many other questions remain to be answered concerning the specific effects of early intervention—for example, the relative importance of the different components of treatment and the treatment variables responsible for change; the roles of parents and other professionals; and the comparative merits of one-to-one versus group teaching, or of home-based versus school-based programmes. There is an urgent need, too, for development of standardized protocols to assess functioning before and after treatment, so that different interventions can be reliably compared.

Intensive home-based behavioural programmes

Probably the most controversial approach to early intervention has been the intensive home-based programme of Lovaas and his colleagues. Therapy, which is provided one-to-one for 40 or more hours a week and lasts for at least two years, is claimed to bring about 'recovery' from autism²¹. The children involved are reported to have shown

large gains in IQ (30 points or more), rates of integration into mainstream school are high and around 40% are said to have achieved normal functioning, with the gains persisting into adolescence²². Although this work is methodologically stronger than other early intervention projects, there have been criticisms concerning the experimental design and particular controversy over the use of terms such as recovery and normal functioning²³. The intensity of involvement also limits opportunities for replication and the few studies that have been conducted have generally used a modified design, with smaller numbers of children^{19,20}. Generally, progress seems to have been accelerated by these interventions, but children are not reported as achieving normal functioning. It is also evident that not all children respond equally favourably to intervention. In particular, progress is limited among those with an IQ in the severe-profound range, or those who do not acquire verbal imitation within the first 4–6 months²⁴. Sheinkopf and Siegel²⁵ conclude that, 'while these studies suggest that home-based behavioral therapy is a *good* option for children with autism, they do not indicate whether such a treatment approach is *better* than other treatments of similar intensity or structure'.

Longer-term evaluations, covering many different aspects of functioning, are required if the true cost-effectiveness of the time, effort and energy expended by families in these programmes is to be adequately assessed. Future research also needs to concentrate on developing strategies to compare different intervention models, and to examine more closely the differential effects of treatment techniques, intensity, structure and setting on different subgroups of children with the autistic spectrum. The important question is not so much 'does this or that technique work?' but rather 'which approaches offer most to which type of children?'

The importance of appropriate education

Whatever the controversy over other forms of treatment, the importance of specialized educational programmes for children with autism and other forms of severe intellectual impairment is extensively documented²⁶. The TEACCH programme²⁷, developed in North Carolina but now widely used throughout the USA and in many parts of Europe, is an educational approach founded on the need for structure. It emphasizes the need for appropriate environmental organization and the use of clear visual cues to circumvent communication difficulties. The programme takes account of developmental levels and the importance of individually based teaching as well as incorporating behavioural and cognitive approaches. However, despite many anecdotal reports of its effectiveness²⁸ there is little in the way of independent evaluation.

Many other approaches to education, focusing specifically on strategies to overcome the fundamental impairments in autism, have been reported although, again, evaluative research is limited. Quill²⁹, for example, provides detailed information on programmes designed to improve social and communication functioning. The Picture Exchange Communication System (PECS)³⁰ uses symbols, pictures or objects to enhance communication skills in the classroom. Other researchers^{31,32} report on the effectiveness of using normal peers to enhance social interactions and play, or programmes such as Bright Start that concentrate on the development of cognitive and metacognitive abilities³³. Jordan and Powell³⁴ describe various innovative techniques (including the use of computers) that can be applied to enhance learning, and others³⁵ provide practical suggestions for teaching children with autism within mainstream settings. Koegel and Koegel³⁶ illustrate how traditional behavioural techniques can be successfully adapted for use in more natural school settings.

The principal components of effective treatments

Most professionals involved in the education and treatment of children with autism make no extravagant claims for success. They recognize the enormous differences between children and their very uneven and individualistic profiles of behavioural, cognitive, social and communicative functioning; nor do they insist that one approach is successful for all. Instead, whether the basic approach is developmental, educational, behaviourist, or a combination of all three, there is recognition of the need for individually designed programmes taking into account not only the deficits that are fundamental to the disorder but also the individual child's pattern of skills and difficulties. Most interventions will involve a range of different techniques, and the choice will be partly determined by family factors.

In general, the most successful programmes, whether predominantly home or school based, have the following components in common:

- A combination of behaviourally oriented strategies with developmental and educational approaches relevant to the individual child's profile of skills and deficits
- Recognition of the need for structured teaching programmes, with a particular emphasis on visually based cues; these provide the child with a predictable and readily understandable environment, which helps to limit confusion and distress
- A focus on the development of social-communication and play activities, especially with peers, and if possible the implementation of specialist training programmes

(for example to improve 'mind-reading' skills³⁷ or social understanding³⁸)

- Acknowledgment that many so-called undesirable or challenging behaviours are a reflection of the child's limited behavioural repertoire or poor communication skills; a focus on skill enhancement is therefore often the most effective means of reducing difficult or disruptive behaviours²⁹
- Understanding of the importance of obsessions and rituals, not just as an underlying cause of many behaviour problems, but also as having a vital role in reducing anxiety and as powerful sources of motivation and reinforcement¹⁰
- Treatment approaches that are family centred rather than exclusively child oriented⁴
- Management strategies that can be implemented consistently without excessive sacrifice of time, money or other aspects of family life.

Finally, and perhaps most importantly, there is a crucial need for early diagnosis and the provision of suitable advice and support for parents, which can do much to lessen subsequent difficulties. Unfortunately, delays in diagnosis are still common. A recent large-scale survey⁴¹ showed that, although most parents became worried in the first months or year of the child's life, there were many delays and frustrations before they obtained a diagnosis. Many children were not diagnosed until the age of 5 or later, well after the optimum period for intervention.

CONCLUSIONS

In 1973, Kanner⁴² noted that, in the 30 years since he had first described the condition,

'There has been a hodge-podge of theories, hypotheses and explanations . . . yet no-one has succeeded in finding a therapeutic setting, drug, method or technique that has yielded the same or lasting results for all children. It is expected that the next 20 or 30-year follow-up will be able to present a report of . . . more hopeful prognosis.'

Unfortunately, confusion and controversy regarding both treatment and causes still prevail. Moreover, although therapeutic and educational provision, and to some extent prognosis, have improved over time⁴³, many children with autism and their families still do not receive the support and guidance they need. Despite the lack of a cure for autism, appropriate treatment and education can prevent secondary behavioural problems and ensure that children develop their existing skills to the full. Intervention in early childhood can greatly improve quality of life in adulthood, and is likely to prove far more cost effective than crisis management in

later life. A focus on *prevention* of behavioural complications, and on the development of appropriate skills, will be more productive than searches for a cure.

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