

Editorial

QJM

Autism—what is it and where does it come from?

It is difficult to imagine how distressing it must be to have a son so sensitive to touch that he recoils whenever he is held. Perhaps it is easier to imagine the disruption if your daughter screamed with rage on leaving the house, unless you took her to the park she visited on her first excursion in a pram. Autism is a puzzling and frightening condition. As the recent furore concerning Andrew Wakefield's research illustrates, it commands the attention of both the public and the medical profession.¹ Given this high profile, it is surprising how few professional and lay people could explain what the diagnosis means, and that no-one has coherently explained its causes.

Like epilepsy or cerebral palsy, autism is not a single disease but a name (deriving from the Greek 'autos', meaning 'self') given to a behavioural phenotype that may have many aetiologies. Kanner first described autism in children in 1943, and coined the term to describe the 'extreme aloneness' that he saw as its characteristic.² Most authorities now agree on three criteria that are required for a diagnosis of autism.

Firstly, an inability to engage in reciprocal social interaction. Some have suggested that the autistic child fails to develop a 'theory of mind', and cannot imagine what other people may think or feel.³ In any event, they lack empathy. Humans, animals and objects are treated alike. Parents and siblings may be regarded as useful tools that can help the autistic child achieve certain ends, but rarely will they be sought for comfort. The child often becomes solitary, avoids eye contact, and displays little of the interested curiosity and exploratory play seen in normal infants and young children.

Secondly, there are communication problems, both verbal and non-verbal. Rather than having a problem with spoken language *per se*, it is almost as if the autistic child cannot grasp the point of communication. The affected child may not babble, is almost always late to speak, and will also have severe difficulties in understanding what is said to them. This defect in understanding spoken speech may be almost total, or may be subtle and merely

take the form of literal interpretation of language. One autistic child, when given a five-pound note by his grandmother and told to share it with his brother, promptly sought out his sibling, tore the money into two pieces and gave him half the note. If the autistic child does develop spoken language, and half will not, then there will almost always be abnormalities in the way it is used. Echolalia is common. Volume, pitch and tone of speech are often peculiar. Sometimes the affected child may remember and repeat endlessly whole conversations, but be incapable of explaining any of the content.

Thirdly, affected children or adults almost always show evidence of restricted imagination and a predilection for rigid routines. The autistic child may form strange attachments to certain everyday objects, such as pieces of plastic or stones, or become fascinated with flicking paper or running water from taps. An obsessive need for routine can have a disastrous impact on family life, with temper tantrums resulting from any small change in the usual pattern of life: a book out of place in a bookcase or an altered route home from school, for example.

The majority of autistic individuals are learning-disabled. Occasionally we are amazed by the autistic savant who appears to develop remarkable skills in one particular area. The children who can produce the most complex line drawings of buildings after one viewing, or who can play back a Beethoven piano concerto after hearing it for the first time, are extremely rare but fascinating examples of this phenomenon. More often, savants have less appealing skills, such as an ability to memorize bus or train timetables. Such a special talent is probably a product of the child's isolation. Deprived of company and friends because of their social incompetence, the lonely and bored child finds an area on which to focus, and returns obsessively to it. Debaene, when describing one autistic child's phenomenal ability to calculate days and dates on the calendar, remarked that he was 'like a Robinson Crusoe lost in an affective desert, his only

companions in solitude are called Friday or January'.⁴ Interestingly, the savant who is lucky enough to make some recovery from his autistic state often then loses his or her special ability.

Many professionals working in the area of developmental disorders say they are seeing increasing numbers of children with autism, and they may well be right. Although prevalence estimates since the 1960s have ranged from 3.3 to 31 per 10 000,^{5,6} the trend in recent studies is definitely upwards, and Gillberg and Coleman estimate a highly significant yearly increase of almost 4% between 1966 and 1997.⁷ Some of the recent increase in prevalence rates may be explained by changing and broadening diagnostic criteria, but probably not all of it. Accurate rates of prevalence are useful not only for public health purposes, but also for further scientific research. For example, the idea that autism has a genetic aetiology has been supported by family studies that have shown an increased risk (compared with the general population) of autism for siblings of probands. However, the extent of that increased risk, and therefore the amount of support it lends to an argument for a genetic aetiology, depends upon the prevalence rate in the general population.

We know little about what causes autism. There appear to be numerous different pathways to the behavioural disorder seen in these children. There must be a genetic component: Bailey showed a concordance rate of approximately 60% in monozygotic twins, and yet no concordance in dizygotic twins.⁸ However, it is unclear where the genetic abnormality lies. Reviewing the literature, every chromosome (except 14) appears to have been implicated at one time or another. Certainly there are single-gene disorders, such as tuberous sclerosis complex, that have much higher rates of autism than the general population. In tuberous sclerosis, the hamartomas in the brain have been implicated, and it has been suggested that tubers in the temporal lobe carry a high risk for the development of autism, but the strength of this association is probably not as strong as was first thought.⁹ At different times, abnormalities in the frontal lobe, parietal lobe, temporal lobe, occipital lobe, basal ganglia, brainstem and cerebellum have all been associated with autism, but no single anatomical location in the brain has been identified as leading to a final common pathway in the central nervous system that causes autistic symptoms. Numerous different biochemical abnormalities have also been reported in these patients. Low levels of serotonin and oxytocin, high levels of gangliosides, and abnormal growth hormone responses to insulin stimulation are just some of the anomalies that have been described.

It is not obvious when the insult to the developing central nervous system must occur if a child is to develop autistic features. The associations of autism with thalidomide embryopathy and fetal cocaine syndrome imply a first-trimester insult. The link with disorders of cortical development implicates the second trimester. There is even some evidence supporting the view that autism can result from the perinatal period, although others have suggested that it is previous abnormality in the fetus that predisposes it to obstetric complications. Postnatal insults can also lead to the autistic phenotype. There are several reports of autism developing after neonatal herpes encephalitis, and DeLong describes three children between the age of five and eleven years who demonstrated autistic features in association with herpes encephalitis that resolved after recovery from the viral illness.¹⁰

There has been little recent support for the idea that the postnatal environment could cause autism. Kanner attracted much controversy during his lifetime for proposing psychodynamic theories regarding the aetiology of the syndrome. In one infamous article in *Time* magazine he said that 'children with early infantile autism were the offspring of highly organised, professional parents, cold and rational, who just happened to defrost long enough to produce a child'. Such views find little support today, but Rutter's finding of 'quasi-autistic patterns' in children from Romanian orphanages who had suffered severe psychological privation provokes further questions about the role of postnatal environmental influences.¹¹

Autism is a phenomenological rather than an aetiological diagnosis. It describes a behavioural phenotype that may be arrived at by numerous different aetiological pathways, operating by mechanisms we do not yet understand. It is hardly surprising, given the severity of the condition and our relative lack of understanding of its causes, that any hypothesis that might explain this distressing illness gains such high exposure in the media.

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