

Childhood Autism - II: Differential Diagnosis

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A major problem in distinguishing autistic disability from the whole range of child language disorders is that its basic components of communication defect and social impairment are to be found in different forms and varying degrees across the whole range of language disorders in children. Any child struggling ineffectively to communicate will be prevented from relating normally to their parents, teachers or other children. It is from the distinctive nature of the communication disorder that autistic children can be recognised and, as argued in the first paper, the distinctive nature of this disorder can best be understood from its developmental origins. There are certain significant stages in language acquisition which are either absent or distorted in autistic children, whereas they may be delayed through more or less normal or compensated for in particular ways in other, e.g deaf or retarded or primarily brain damaged children. The very young autistic child often presents as a particularly 'good' baby presumably because his highly selective attention and piecemeal efforts to construct an awareness of his world preoccupies him, making him very self contained. Often as the baby grows into a toddler and hence become more exploratory, the autistic child becomes prone to extremes. He combines placid self-isolating behaviour which alarms his parents because he seems so cut off, with temper tantrums, episodes of confusion amounting almost to panic and strange protracted fears - presumably as he struggles with the unfamiliar or the unexpected that inevitably confronts him. Later in childhood, he often settles into a preoccupied, rigidly obsessional child with set ways and stereotyped responses, presumably as he settles into his own version of the world in which he moves. Later as puberty approaches he may become very settled, more confident and assertive and indeed become a very manipulative youngster. This distinctive natural history will of course vary from one autistic child to the other because it is important always to emphasise that autistic children, like any other group of handicapped children, are individuals to the same extent as normal children.

Another obstacle to confident differential diagnosis is that autistic disability may be combined, not only (and understandably) with a psychiatric overlay, but with other disabilities, such as fluctuating hearing loss, epileptic attacks and attention defect. This is inevitable in a condition which can be caused by a variety of pathological processes. It has been argued in the first paper that the core defects of autism can be the result of organically distorted development but a virtually identical clinical picture can be produced by some biochemical disorders, such as untreated phenylketonuria, by some forms of epileptic illness such as infantile spasms and other causes. Each of these can inflict on the disabled child, fits both overt and subclinical, attention defects, and a whole range of learning problems which can complicate or mimic the clinical picture of autism. One always has to remember that the autistic child can suffer from fluctuating hearing loss, minor epileptic attacks and other disorders much in the same way as other mentally handicapped children. These possibilities should always be considered and properly investigated however taking this may be to the investigating department concerned. Finally each of these disorders can produce symptomatology which closely resembles autism, particularly in the young child, so it is important to review briefly those features which distinguish disorders such as hearing loss, brain damage with attention defect, and global retardation from the autistic child.

Hearing loss

The tragic disability of deafness is usually apparent even if the young child, not least through the

harsh, dissonant quality of their vocal output. However, it is always worth remembering that the intelligent deaf child is often remarkably capable of giving the impression of hearing. The main culprit, among hearing disorders in producing a picture similar to autism is recurrent hearing impairment particularly in the dull child. This is often the product of frequent catarrhal otitis producing a 'glue ear' and often associated with a snuffly, irritable and inattentive child. It is a condition furthermore which may be almost asymptomatic without pyrexia, earache or discharge and therefore not clinically evident. Such a child can suffer from a widely fluctuating level of acoustic awareness, often combined with a general malaise which makes them disinterested, passive and almost inaccessible. It will also interfere with their language acquisition. However, it does not seem seriously to interfere with the child's awareness of pitch change and inflection patterns, nor in their capacity to produce these. A flat 30-40 db loss, characteristic of these children often as a resting state, usually enables them to respond and produce appropriate inflection patterns and normal vocal signals. Their social response, too differs from the autistic child; their visual scanning of faces will be normal and not have the highly selective impersonal quality of the autistic child, nor will they have the idiosyncratic interest in particular acoustic stimuli, which one often finds as a puzzling feature of autism. The warmth of their response to their parents compared with strangers and the emergence of simple representational play will also distinguish them from the autistically disabled child.

Attention defect

The child with diffuse cortical hypoxaemic damage, often the consequence of a toxæmic pregnancy may well be a motor intact, highly distractible child with massive attention defect and poor or no language. Presumably his difficulty in acquiring language is due to his inability to focus and sustain his attention sufficiently to make categories effectively out of the array of information his senses provide. This is fundamentally different from the autistic child's problem. The brain damaged distractible child is able, and above all is interested in attending to and labelling events which occur repeatedly, are usually conventionally i.e. predictably of interest and have some value to him, such as items of food or drink. His speech is slowly acquired and often confined to utilitarian situations, and does not appear suddenly like the autistic child's but nor does it usually involve drawing attention to diverse objects of interest as does verbal labelling in the normal child. In addition the child with attention defect will not recognise or retain the detail of any item or situation as does the autistic child. Similarly his hyperactive behaviour is different; he is much more prone to shift his attention rapidly from one activity to the next, whereas the autistic child will often concentrate with total absorption on one favoured activity such as turning taps or light switches on or off after an initial brief period of busy exploratory searching. Finally the brain damaged child often impresses with his efforts to communicate as may do the child with hearing loss so that the little he does say is laboriously produced with great deal of intent, often including gesture contrasted with the flat, facile vocal output of the autistic child when he feels so disposed to produce it.

Global retardation

Some clinical conditions produce global delay in a motor intact child which may be so severe that the child has little social response and therefore resembles an autistic child. Such children, however, tend to have normal inflection of the limited vocal output they possess: their vocal signalling occurs in socially appropriate conditions, i.e. they make a requesting inflection if they want something. Similarly their phonetic repertoire tends to be delayed often restricted to simple back vowel/front consonant combinations even as childhood progresses, so that they do not reveal the articulatory skill of the autistic child, however little he may communicate. They are much more likely to have delayed motor milestones, protracted incontinence and very slowly acquired self help skills than is the autistic child. But one of the major contrasts in the retarded child with autistic features and the truly autistically disabled child is the absence in the former of certain specific autistic skills. Retarded children are not usually skilled mimics; nor are they particularly concerned about spatial arrangements or routes followed, nor do they have the strange visuo-spatial skills with eg. inset puzzles or jigsaws that one finds in the autistic child. A final point is that often their facial expression and indeed its mobility are much more normal, so that they give the impression more of a slow, placid, apathetic child than one living in a world of his own, which is such a sad feature of autism.

Maladjustment

The intelligent autistic child is often misdiagnosed because they rarely have any episode of cerebral insult, pass their motor milestones normally, do not suffer from fits and have syntactically correct, well-articulated (though very oddly used) language. They are often regarded as psychiatrically disordered children, i.e. as maladjusted. In such children the social impairment inherent in their autistic disability is the most overwhelming source of complaint. They behave very oddly, though often quite intelligently, particularly in rote or mathematical skills; they may have prodigious memories, all of which deter people from regarding them as a disabled child. However, they should be recognised through the oddly inflected monotonous way they speak, their total inability to cope with abstract ideas in spite of their academic skills in other directions, their frequently obsessional tidiness, their concern to maintain routine, spatial arrangements and familiar routes, but above all from their complete inability to appreciate what is normal socially conforming behaviour. They will make the most outrageous personally offensive comments on eg. people's appearance which, though an accurate observation, may be socially quite unacceptable. The fact that they are quite unaware of this is often ignored or simply disbelieved. This is particularly painful for the child's parents because they will often be regarded as indifferent, over-anxious or inadequate and therefore accountable for the child's rudeness when they have themselves struggled to understand why their child should behave so oddly. It is inevitable that any parent attempting to rear an autistically disabled child will run into all sorts of interpersonal conflict, so the child often presents at a psychiatric clinic where they may be regarded as the casualty of parental mismanagement. In such cases it is extremely important that the psychiatrist takes a careful history and recognises the odd and distinctive language and cognitive peculiarities of this sadly disabled patient.

As yet the differential diagnosis of autism depends on defining distinctive behaviour characteristics. It will of course be helped by more objective investigation which excludes other and similarity disabling conditions, e.g. impedance tests or cochleograms for hearing discharges; amino-acid screening to

exclude metabolic disorders and so on. It should always be remembered that positive findings from these investigations do not necessarily exclude autism since an autistic child may have other, associated, pathology. Eventually tests will be developed based on our better understanding of the biological origins of autistic development. At the moment a variety of such tests exist which act as indicators because they are positive in a proportion, sometimes a high proportion of autistically disabled children, but as yet we do not know enough to define any specific biological indicator which is pathognomonic for the condition. A specific cognitive profile can be revealed in careful psychological testing [1] some autistic children (as will be further demonstrated by Dr. Shivashankar in a paper at this conference) have abnormal acoustic brain stem responses with intact hearing [2] chromosome abnormalities occur in some autistic children, particularly the Frail X [3], [4] and again a further contribution in this direction will be made by Dr Manjunath. Examples of abnormal levels of neurotransmitters such as serotonin, and of trace elements and nutrients have been suggested [8], [10], but the evidence here is conflicting so that it is clear we have a long way to go before our understanding of this distressing condition reaches down to the level of reliable biological mechanisms which we can identify and test. Progress in this direction, will not only better equip us to differentiate autistic disability from other similar conditions but by improving our understanding will hopefully put into clearer perspective the parents' role and indeed the effectiveness of our own efforts to help the disabled child.

1. Lockyer L & Rutter M, 5-15 year follow-up study of infantile psychosis: IV Patterns of cognitive ability

British Journal of Social Clinical Psychology Page: 9: 152-163, 1970

2. Tanguay P E & Edwards R E, Electrophysiological studies of autism: The whisper of the bang
Journal of Autism & Developmental Disorders Page: 12: 172-185, 1982

3. Meryash D L, Szymanski L S & Gerald P S, Infantile autism associated with the fragile-X syndrome

Journal of Autism & Developmental Disorders Page: 12: 295-301, 1982

4. Gilberg C & Wahlstorm J, Chromosome abnormalities in infantile autism and other childhood psychosis: A population study of 66 cases

Developmental Medicine & Child Neurology Page: 27: 293-304, 1975

5. Hoshino Y, Yamamoto T, Kaniko M, Tachibana R, Watanabe M, Ono Y & Kumashiro H, Blood serotonin and free tryptophan concentration in autistic children

Neuropsychobiology Page: 11: 22-27, 1984

6. Geller E, Ritvo E, Freeman B & Yuwiler A, Preliminary observations on the effect of fenfluramine on blood serotonin and symptoms in three autistic boys

New England Journal of Medicine Page: 307: 165-169, 1982

7. August G, Raz N, Papanicolaou A, Baird T, Hirsh S & Hsu L, Fenfluramine treatment of infantile autism Neuroclinical, electrophysiological and behavioural effects

Journal of Nervous & Mental Diseases Page: 172: 604-612, 1984

8. Cohen D, Johnson W & Caparulo B, Pica and elevated blood levels in autistic and atypical children

American Journal of Diseases in Childhood Page: 130: 47-48, 1976

9. Shearer T R & Larson K, Minerals in the hair and nutrient intake of autistic children

Journal of Autism & Developmental Disorders Page: 12: 25-34, 1982

10. Wecker L, Miller S, Cochran S, Dugger D & Johnson W, Trace element concentrations in hair from autistic children

