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Causes and pathology of early childhood psychoses

I. KOLVIN

In the majority of the studies reported here, the subjects are referred to by the authors as 'autistic'. The criteria for case selection vary. Some authors use a narrow definition of autism based on Kanner's publications, whereas others cover a wider group, but they are all concerned with children in whom the psychosis began before five and, in the great majority, before three years of age (Kolvin, 1971; Kolvin, Ounsted, Humphrey & McNay, 1971).

Social and psychological factors

In the past, it was commonly assumed that the fundamental determinants of psychiatric abnormalities in childhood were parental personality, attitudes and emotional disturbance, so it was to be expected that research workers seeking explanations for autism would initially investigate such factors.

Studies of children referred to particular clinical services have usually reported that the parents of classically autistic children come predominantly from the upper social strata and are of above average intelligence (Creak & Ini, 1960; Kolvin, 1971; Kolvin, Ounsted, Richardson & Garside, 1971; Rutter & Lockyer, 1967). Only a few groups of workers (e.g. Ritvo *et al.*, 1971; Schopler, Andrews & Strupp, 1979) have to date published studies of autistic children in which the findings on the parents were out of step with those of the other major research groups. The first epidemiological study of autism (Lotter, 1967) and the largest in scale found a tendency to higher

social class, especially among the parents of the most typically autistic children.

As discussed in chapter 34 some recent evidence (Wing, 1980b) has failed to confirm these findings, and more detailed investigation is needed to examine the relationship of social class to various subgroups among children with autism and other early childhood psychoses. However, as most studies have reported a social-class bias in the former, it is not surprising that this has been used to support the hypothesis of an environmental origin for childhood autism. The earlier descriptions of social formality and lack of overtly expressed warmth in the mothers (Kanner, 1943; 1949) and in fathers (Eisenberg, 1957) led to the concept of 'refrigerator parents', who were said to have obsessive and cold personalities, rather than to these features being seen as relatively common characteristics of the public manners of the middle and upper classes.

Unfortunately, these early theoretical explanations were considered to have been proved without checking the validity of the basic premise. A plethora of similar theories has been advanced, mainly stemming from clinical practice, most of which are unacceptable on the grounds of being based on heterogeneous, biased, or unduly small series; similarly, reliable and objective techniques have not been used to accumulate empirical data upon which a theory could be based (Kolvin, Garside & Kidd, 1971). Not only were some of the theories that were developed incapable of generating testable hypotheses, some theorists did not even attend to the two axiomatic methodological steps of demonstrating a correlation between child and parent variables and of carrying out experimental or observational studies in an attempt to validate a cause–effect relationship. In addition, most theorists did not consider the possibility that the syndrome might have a multifactorial basis.

Modern Anglo-American research has not confirmed the parental personality stereotype, irrespective of whether it has been based on self-rating schedules such as the Maudsley Personality Inventory (Kolvin, Garside & Kidd, 1971), assessments of parental warmth based on clinical interview (Creak & Ini, 1960), parent attitude scales (Pitfield & Oppenheim, 1964), objective tests of thought disorder (Schopler & Loftin, 1969a, b), or combinations of clinical assessment and objectively rated interviews (DeMyer *et al.*, 1972b). The last-mentioned did not identify any unusual rearing or attitudinal patterns in parents of autistic children with regard to warmth in nurturing, acceptance of their infants, or degree of general stimulation. They found that child-care practices of parents of autistic children were similar to those of the parents of matched normal children. Finally, there is the study of Cox, Rutter, Newman, and Bartak (1975), who used objective clinical interviews plus parental self-rating inventories. These researchers reported that mothers showed somewhat less warmth to their autistic than to their non-autistic children, but interpreted it as difficulty in interacting with an unresponsive child, especially since the difference was small and half the mothers were rated as very warm to their autistic children.

Thus, there is no evidence that autism is secondary to abnormal parental personalities, nor unusual child-rearing practices. As pointed out by Rutter (1972b), there is also no evidence of a link with extremely depriving circumstances. Ferster (1961) suggested that autism was due to faulty conditioning, but none of the relevant findings have supported this view. Wing (1966) notes that it would be difficult to imagine how any of these extreme environmental experiences could give rise to the complex but specific patterns of impairments in such diverse areas as cognitive, perceptual, motor, and autonomic functioning. Bartak, Rutter, and Cox (1975) studied autistic children of normal intelligence in whom no overt neurological disorder could be identified. Even in this group they found evidence of cognitive disorder and concluded that 'it is most unlikely that the condition is primarily psychogenic' (Cox, Rutter, Newman & Bartak, 1975). The sum total of these studies and reviews provides strong refutation of a psychogenic hypothesis. In addition, there is suggestive evidence (Bell, 1968, 1971; Cox, Rutter, Newman & Bartak, 1975) that some of the parental social reactions may be secondary to the autism and that social isolation, when present in mothers of autistic children, appears to follow the onset of the disorder (Kolvin, Garside & Kidd, 1971).

Biological factors

Gross organic causes

Current research suggests that early childhood psychosis may be the final common behavioural expression of a wide variety of organic-cum-developmental influences. The evidence for this is impressively extensive. First, presumptive evidence of cerebral injury has been obtained from studies of perinatal complications, with particularly high rates being

reported in clinical studies (Gittelman & Birch, 1967; Gubbay, Lobascher & Kingerlee, 1970; Lobascher, Kingerlee & Gubbay, 1970; Kolvin, Ounsted & Roth, 1971; Knobloch & Pasamanick, 1966; Folstein & Rutter, 1977) and also, but to a less marked degree, in population studies (Lotter, 1967; Treffert, 1970). Other evidence of neurological involvement comes from DeMyer and colleagues (1972b), who reported reduced alertness in infancy and more overt signs of brain damage.

Second, in about half of the cases in two major hospital series, evidence of cerebral dysfunction was found (Rutter & Lockyer, 1967; Kolvin, Ounsted & Roth, 1971). In the course of time, autistic children and adolescents develop epileptic fits far more frequently than might be expected by chance (Rutter, 1970; Kolvin, Ounsted & Roth, 1971; DeMyer *et al.*, 1973). Early childhood psychosis can also be associated with previous episodes of infantile spasms (Kolvin, Ounsted & Roth, 1971; Taft & Cohen, 1971). The association is discussed in chapter 31.

Third, certain identifiable organic conditions appear to have a close relationship with the behavioural abnormalities characteristic of early childhood psychosis. Chess (1971) reported that, in a series of 243 children of mothers who had had rubella in pregnancy, 10 children had classic autism (a rate of 412 per 10,000) and 8 had a 'partial autistic syndrome' (329 per 10,000). These rates are more than one hundred times greater than would be expected in the general population (see chapter 34). Not all of these rubella children had visual impairments. Early childhood psychosis can also be found in children with congenital blindness, or partial sight, combined with brain damage due, for example, to retrolental fibroplasia (Keeler, 1958; Freedman, 1971).

Encephalitis or encephalopathy from causes other than maternal rubella have been reported as occurring during the first two or three years of life in some children with early childhood psychosis (Greenbaum & Lurie, 1948; Wing & Gould, 1979). The pattern of behaviour has also been described in untreated phenylketonuria (Jervis, 1963), tuberose sclerosis (Critchley & Earl, 1932; Earl, 1934; Creak, 1963; Lotter, 1974), and cerebral lipoidosis (Creak, 1963). In some reported cases, the diagnosis of the neurological condition was not made until after death (Creak, 1963; Lotter, 1974). Darby (1976) summarized the results of all the cases of childhood autism and psychosis where post-mortem examination of the brain had been carried out; several cerebral pathologies were found.

It is of interest that Down's syndrome is comparatively rarely associated with early childhood psychosis (Wing, 1969, 1971; Wakabayashi, 1979; Wing & Gould, 1979).

It has been suggested that, in at least some cases, autism has a genetic basis. The evidence for and against this is discussed in chapter 39.

Biochemical, metabolic, or allergic theories of aetiology have been put forward but remain not proven. Possible lines of investigation in this field are discussed in chapter 37.

Localization of the brain pathology

So far we have little idea of the nature of the specific brain dysfunction. The diversity of EEG and seizure patterns, which range from focal epilepsy to the widespread disorganization of hypsarrhythmia (Rutter & Lockyer, 1967; Kolvin, Ounsted & Roth, 1971; Taft & Cohen, 1971), appears to argue against a single underlying homogeneous pathological mechanism. On the basis of knowledge available at the time he was writing, Rutter (1974) found it impossible to suggest a site for the lesion and favoured the hypothesis of a non-specific syndrome of biological impairment. Wing and Wing (1971), on the other hand, attempting to account for the complex clinical picture, argued that multiple neurological deficits do not necessarily imply multiple lesions. 'A number of different brain functions could be affected by, for example, a single genetic or biochemical abnormality, or anatomical proximity could make different centres vulnerable to the same lesion.' On this hypothesis, any condition that produces abnormality or delayed maturation of the relevant brain areas could, theoretically, lead to the impairments found in childhood autism or other early childhood psychoses.

There are a variety of views as to which brain areas have to be involved to produce the characteristic behaviour. Crawley (1971) considered that the problem might be in the association areas. The pattern of deficiencies in language and symbolic processes, with better retention of visuo-spatial skills, has led some workers to suggest that the lesion or dysfunction underlying childhood autism affects the dominant hemisphere of the brain (Hermelin, 1966; Barry & James, 1978). One group of workers (Hauser, DeLong & Rosman, 1975) reported enlargement of the temporal horn of the lateral ventricle with atrophy of the adjacent area of the left medial temporal lobe. But there is doubt about the methodology and the homogeneity of the sample. It has been suggested that

the subjects were highly selected with heavy loadings of neurological disorder (Lancet, 1976).

Ross and Mesulam (1979) and Sackeim and Gur (1978) produced evidence that the non-dominant hemisphere is concerned in the comprehension and expression of emotion in non-verbal ways. Impairment of non-verbal as well as verbal communication is characteristic of autism and other early childhood psychoses. If the involvement of the non-dominant hemisphere in non-verbal communication is confirmed, this would suggest that the problems in such conditions are probably not confined to the dominant hemisphere alone. The lesion or dysfunction must be capable of impairing all aspects of language and communication, as well as explaining the other features of the clinical picture.

Damasio and Maurer (1978) considered the possibility of dysfunction of a single and yet widely based system consisting of the bilateral neural structures of the mesolimbic cortex. They postulated that autism might be the result of macroscopic or microscopic changes in the above-mentioned target areas or structure influencing them. On the basis of this hypothesis, the broadly homogeneous functional aberration of autism might be the result of very different types of structural disorganization. Unfortunately, this theory is so far-ranging that it would be difficult to test. Other workers have suggested subcortical dysfunction, which directly or indirectly affects the reticular activating system involving either under- or over-arousal (Hutt, Hutt, Lee & Ounsted, 1964; 1965) or an imbalance between these systems (Des Lauriers & Carlson, 1969). The empirical evidence advanced in support of these conflicting hypotheses is rather questionable (Hermelin & O'Connor, 1968).

Cognitive deficits

The nature of the cognitive deficits is discussed in chapter 35. They are so marked, so characteristic and, beyond a certain point, so unresponsive to environmental modification, that it is tempting to suggest a common pathological origin. However, none of the theories so far put forward is able to explain all the abnormalities of language, perceptual, motor and autonomic function and there is, as yet, no direct evidence of a necessary and sufficient pathology.