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The Etiology of Infantile Autism: The Problem of Biological versus Psychological Causation

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The literature on autism contains many papers in which it is asserted rather than suggested that psychogenic factors play a major part in the etiology of the disease. Indeed, a substantial proportion of these papers carry no indication that biological factors may play even a minor part in the disease.

This chapter will be devoted to a detailed consideration of the problem of the etiology of infantile autism. The present writer disagrees with Eisenberg and Kanner's assertion that "Arguments that counterpose 'hereditary' versus 'environmental' as antithetical terms are fundamentally in error. Operationally defined, they are interpenetrating concepts" (1956, p. 563). That heredity and environment are "interpenetrating" cannot be denied. But the conclusion that their interpenetration precludes analysis does not follow. Complex problems require that we *increase,* not diminish our analytical efforts, if we are to have hope of solving the problems confronting us (Burt, 1958; Cattell, 1960).

There are several reasons for drawing close attention to the consideration of etiology in infantile autism.

1. The welfare of individual autistic children and their families hinges closely upon the problem of specific etiology, as van Krevelen has amply demonstrated (e.g., 1958, 1960). If the disease is psychogenic, the causative factors need to be identified. On the other hand, if autism is determined solely by organic factors, there is no need for the parents of these children to suffer the shame, guilt, inconvenience, financial expense, and marital discord which so often accompany the assumption of psychogenic etiology. (For examples of this, see May, 1958; Peck, Rabinovitch, and Cramer, 1949. Oppenheim, 1961; and Stuart, 1960 are also germane.)

2. So long as the practitioners who actually deal with autistic children

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feel satisfied that the disease is largely or entirely psychogenic, biologically trained research workers will feel disinclined to concentrate their efforts on the problem. It should be added at this point, in all frankness, that while the purpose of the review which follows was to investigate the specific etiology of early infantile autism, the issue is a broad one and a good deal of the material covered relates closely to the problem of causation of childhood behavioral disorders in general. The results of this work were surprising to the present writer and discordant with his previous beliefs. They may also be so to the reader. It is largely because of the large discrepancy between research findings and the remainder of the published literature that such detailed consideration is given the problem of etiology in this chapter.

Failing to find any adequate formulations of many of the inexplicit assumptions on both sides of the issue, the writer has attempted to articulate these, in his belief that a good part of the unique function of the psychologist is to try to articulate what seems ineffable.[1] No doubt much of this formulation will be challenged, and additional points will need to be added to those listed. We can do no better on this issue than to refer to Bacon's assertion that truth is more likely to emerge from error than from confusion.

The Arguments for Psychogenesis of Infantile Autism

The case for psychogenesis of autism would appear to rest on the following arguments and assumptions:

1. No consistent physical or neurological abnormalities have been found in autistic children which could account for their condition.
2. Many autistic children have been raised by parents apparently deficient in emotional responsiveness, which could have pathogenic effects on the child.
3. Certain children raised in hospitals or orphanages where maternal contact was sparse have been reported to show an undue frequency of emotional difficulties.
4. The behaviors of the child—his indifference or aggressiveness, his refusal to speak (or "elective mutism"), his apparent withdrawal from the outside world—are interpreted as signs of "punishment" or "retaliation" against the parents.
5. Certain incidents in the life of the autistic child appear to be pathogenic and permit the disorder to be traced to them.

[1] There are certain arguments, however, that defy our attempt to reformulate them in any testable way: "I believe that the child who shows autistic behavior has been traumatized in the early months of life since he symbolizes to the mother so definitely the hated sibling" (Ribble, in discussion of Despert, 1951, p. 350).

6. Psychotherapy or otherwise placing the child in a kind and understanding environment has beneficial effects.
7. The high incidence of first-born and only children suggests that parental attitudes may be causative.

Let us consider these points in turn:

1. *The absence of signs of organic impairment.* While the presence of physical symptoms is ordinarily regarded as conclusive proof of organicity, as in mongolism or phenylketonuria, the absence of such symptoms cannot be considered indicative of functional determination. This is so because "Neurological science thus far has been quite unable to furnish an adequate description of the neural processes involved in even the very simplest forms of mental activity" (Sperry, 1952, p. 292). Eisenberg and Kanner have said of autism, "neurologic investigations of the integrity of central function remain as yet in their clinical infancy and a negative result with current methods cannot be regarded as a conclusive demonstration of the lack of central nervous system pathology" (1956, p. 560).

There are numerous cases in the literature where even gross brain disease which eventually caused the death of the child had escaped intensive repeated neurological examination and was found only post-mortem (e.g., Ross, 1959). Heller's disease and phenylketonuria were both considered "functional" childhood psychoses until their organicity was determined (Benda, 1960; Kanner, 1949). Nevertheless writers such as Despert (1947) and Bettelheim (1959b) have written that the possibility of organic damage in the cases they cite was "ruled out" by physical and neurological examination. May (1958) and van Krevelen (1960) have cited cases in which the possibility of organic damage was ruled out without even examining the children.

2. *Parental personalities.* Eisenberg and Kanner are among the many writers who subscribe to the notion that parental behavior is a factor in producing autism in the child, although, unlike many others, they are careful to qualify their position:

> It is difficult to escape the conclusion that this emotional configuration in the home plays a dynamic role in the genesis of autism. But it seems to us equally clear that this factor, while important in the development of the syndrome, is not sufficient in itself to result in its appearance. There appears to be some way in which the children are different from the beginning of their extrauterine existence (1956, p. 563).

Kanner and Eisenberg do not specify *why* it is hard to escape the conclusion. They cite no supporting evidence. Plausible as the hypothesis may seem, there is no reason for accepting it in favor of competing hypotheses.

This is a clear case of subscription to the fallacies that *post hoc ergo propter hoc,* and that "correlation implies causation."

How can one say that both the child's and the parent's behavior are not related consequences of the same genetic factors? Granted, it may be *possible* that psychological factors contribute, but it has not yet been demonstrated that they do, to even a minor extent; nor has it been demonstrated that any familial environment, no matter how favorable, would have prevented the emergence of the disease.

Perhaps it is in relation to the "parental personalities" that reference should be made to the recent work on "affectional systems" in monkeys (e.g., Harlow and Harlow, 1962a; 1962b). These points appear to be most relevant: The behavior of infant monkeys is relatively independent of the personality (or response repertoire) of the mother. Infant monkeys raised by cruel, rejecting, unsympathetic and indifferent mothers reacted not by "autistic" withdrawal but by persistent and vigorous attempts to obtain the mother's attention. Even infant monkeys brutally beaten by mothers attempting to discourage contact showed no signs of maladjustment. Infant monkeys raised only with inanimate cloth-covered "mother surrogate" effigies showed no signs of maladjustment until adulthood, when their sexual functioning was found to be severely impaired.

The only way in which any of Harlow's infant monkeys could be treated to cause them to simulate "autistic" withdrawal was through total social deprivation for extended periods. But even twenty minutes a day of contact with other infant monkeys seemed sufficient to produce normal development.

With regard to the problem of *absence* of maternal contact, as differentiated from the nature of those with whom the subject is in contact, there is ample evidence at the human infant level.

3. *Maternal deprivation and hospitalism.* The works of Goldfarb, Spitz, Ribble, Bowlby, and others on the syndrome of "hospitalism" or "maternal deprivation" are frequently cited as analogous evidence that early infantile autism is psychogenic. The analogy with autism is a poor one, since the symptoms which characterize these infants do not resemble autism, as noted by Eisenberg and Kanner (1956), Keeler (1957), and others, and do not begin to approach autism in severity. Additionally, the special circumstances relating to the sampling and physical environment of these maternally-deprived infants make them a poor basis on which to construct a psychogenic theory.

In 1951 Bowlby wrote that there was "no room for doubt . . . the prolonged deprivation of the young child of maternal care may have grave and far-reaching effects on his character" (p. 46).

Despite Bowlby's assurance on the matter, psychologists have been very

skeptical and critical of the hypothesis. The hospitalism studies have been subjected to criticisms of the gravest nature. Pinneau (1955), who earlier pointed out severe deficiencies in the work of Ribble in this area, has also called attention to what appear to be disqualifying errors in the work of Spitz and Fischer. Pinneau notes, to cite one example, that of the 59-point drop in the Development Quotient Spitz reported for his "maternally deprived" group of infants, 55 points were lost *prior* to the time at which most of the children were separated from their mothers! Stevenson (1957) has observed that the screened-off cribs Spitz has described may have resulted in the infants being deprived of adequate physical sensory stimulation, rather than of maternal affection.

With regard to Fischer's study of hospitalism, Pinneau noted that Fischer selected for study, at a home for unwed mothers, those infants with IQs under 90. Then, after observing their behavior in much the same way as is done in *arriving* at IQ scores for infants, she concluded that their poor performance was due to "hospitalism"!

Pasamanick and Knobloch have recently noted that the maternal deprivation hypothesis may turn out to be simply another instance of misdirection by the "*post hoc ergo propter hoc*" fallacy:

> Further, since a large number of children exhibit no significant difficulties after hospitalization, we must consider the possibility that it may be largely those children having some brain injury, with a consequent lowering of thresholds to stress, who are affected by hospitalization during infancy (1961, p. 87).

They also note that a number of the hospitalized children included in Goldfarb's study were grossly defective or brain-damaged and suggest that this was why the children remained hospitalized rather than being placed in foster homes. This could readily account for the so-called "symptoms of hospitalism."

Faced with a rising tide of evidence disconfirming the maternal deprivation hypothesis from both his own subsequent work and the reports of other investigators, *Bowlby reversed his stand.* In 1956 he concluded that some of the workers who drew attention to the dangers of deprivation, including himself, had tended to "overstate their case." "In particular, statements implying that children who experience institutionalization . . . early in life *commonly* develop psychopathic or affectionless characters are incorrect" (Bowlby, Ainsworth, Boston, and Rosenbluth, 1956, p. 242). He noted that "only a small minority develop those very severe disabilities of personality which first drew attention to the pathogenic nature of the experiences" (p. 240).

Inasmuch as a small minority of *any* group of infants will show personality disorders, especially if they had been selected because of adverse circumstances surrounding pregnancy or birth, it does not seem that the proponents of the psychogenic view may look to the maternal deprivation hypothesis for support.

Actually, our attention to the maternal deprivation studies results not from their actual relevance to the problem of etiology of autism, but only from their having frequently been cited as analogous evidence favoring psychogenesis of autism. In actuality, the case history materials supplied by Kanner and others show very little reason for believing the children were neglected. Kanner observes that very few of the children were at least overtly rejected (1949). The mothers "were anxious to do a good job," and they performed like "overconscientious gasoline station attendants." The mother of Donald T., Kanner's first case of autism, for instance, tried to help her son by spending "all her time developing ways of keeping him at play with her" (1943, p. 217). The verbal behavior of the children is also suggestive of a good deal of contact with adults.

Bowlby's original statement that there was "no room for doubt" about the adverse effects of maternal deprivation is very similar to a statement made with equal assurance more than half a century before by the noted Langdon Down (Down, 1887, p. 89)—except that Down was sure that the *opposite* conclusion was correct—maternal emotionality causes idiocy!

4. *Child's behavior as suggestive of etiology.* Many writers consider the child's hostility or indifference to his parents as evidence that the parents are guilty of causing the disease. Noting that many autistic children were exposed to parental "coldness, obsessiveness, and a mechanical type of attention," Kanner observed that the children's "withdrawal seems to be an act of turning away from such a situation" (1949, p. 425). Elsewhere Kanner uses the term "retaliation." Others also have written in this vein.

It would seem more reasonable to regard the child's actions as *symptoms,* not as indications of etiology. In the case of the adult who insists that he is being persecuted by the Communists, the F.B.I. or by little green men from Mars, one does not take his statements at face value but only as an indication that he is ill. Unless there is a reason for believing otherwise, it seems best also to regard the autistic child's symptoms solely as symptoms, even if "the patient acts as if his mother is the source of his psychotic fears, and he attributes the potentiality of the same responses onto other humans" (Weiland & Rudnik, 1961, p. 552).

Does "elective mutism" really represent the child's *refusal* to speak? One cannot conclude this unless one is willing also to conclude that adult aphasics who are virtually mute until an emergency arises (Brian, 1960, p. 180) also had "elected" their disability.

Is the fact that many autistic children ignore humans and are interested solely in objects indicative only of some sort of "willful" rejection of man-

kind, or can this result from organic pathology? Nielsen (1951, p. 185 ff.) describes a fully conscious patient who, after occipital lobe damage, was apparently blind with respect to "animate" objects, including the surgeon's fingers, a doll, and her own artificial teeth, but had no trouble in perceiving non-living objects. Another patient could perceive *only* animate objects after occipital and temporal lobe damage. Autos and mountains were not recognized, but flowers and an animal grazing near the autos were readily identified.

Nor does it seem to be widely appreciated that brain pathology can influence affection as well as cognition. Those who believe that an autistic child's behavior represents withdrawal from his parents should also believe, it would seem, that the sweet disposition and loving nature that is almost universal among Mongoloid children stems from the pleasure the parents of these children must experience on viewing their offsprings' malady.

5. *Pathogenic incidents.* Many discussions of etiology of autism refer to certain specific events which are said to be traumatic to the child and thus causative of his disorder. The birth of a sibling, a stay in the hospital, the absence of one or both parents are examples of the incidents cited.

That such incidents can produce disorder has not been demonstrated. Many infants and children suffer exceedingly traumatic lives with no evidence of autism or other disorder, while some autistic children have backgrounds free of at least obvious pathogenic incidents.

Renaud and Estess (1961) have published a study of the "pathogenic incidents of childhood" as derived from intensive interviews with 100 above-average young men (military officers). Renaud and Estess discovered, to their considerable surprise, that there was just as much material of a supposedly "pathogenic" sort in the childhoods of these men as would have been expected in a clinically abnormal group. The implications of this finding are clear.

Stevenson (1957) explored the literature relevant to the proposition that personality is "plastic" in infancy and came to the conclusion that "if the experiences of childhood importantly influence the later personality, we should expect to find some correlation between such experiences and the later occurrence of mental disorders. In fact, no such correlations have ever been shown" (p. 153).

6. *Psychotherapy.* Indirect support for the psychogenic hypothesis is often inferred by writers who point to the improvement wrought through psychotherapy or by other means of modifying the child's social environment. As Kuten and Kuten (1958) point out, such evidence for psychogenesis typically comes from writers who cite only very small numbers of cases. Control groups are not used.

Kanner has discussed the results of psychotherapy for the first 42 of his cases with infantile autism, and has observed that: "29 did not get anywhere," including some who had "what is regarded as good psychotherapy." On the other hand, the 13 who recovered sufficiently to go to school "are children who have not had anything that is regarded as good psychotherapy or as psychotherapy at all" (1954b, p. 471).

It might be argued that Kanner's group is too small to provide conclusive evidence that psychotherapy is ineffective in infantile autism, but taken in aggregate, the evidence relating to the efficacy of psychotherapy in childhood (or adulthood, for that matter) is generally quite negative and thus lends no support for the psychogenic hypothesis (Astin, 1961; Eysenck, 1961; Hood-Williams, 1960; Levitt, 1957, 1960).

It should be noted that the force of this argument does not rest on the adequacy of the studies of the utility of psychotherapy, since it is not asserted here that the efficacy of psychotherapy has been disproven, but only that it is unproven.

In the absence of evidence that psychotherapy produces improvement in excess of base rate, writers who cite improvement in individual case studies as evidence that autism was psychogenically induced are taking an untenable position. This untenability is underlined by the further point that even if psychotherapy *could* be proven to be effective, it could not be taken as indicative of psychogenic causation any more than the efficacy of oxygen in heart failure proves that the heart failure was caused by a lack of oxygen.

7. *Birth order.* The high frequency of the first-born with autism is sometimes taken as evidence of psychogenicity. There are two obvious objections to this line of reasoning. First, there are many children with autism who have both younger and older normal siblings. Second, a high frequency of first-borns is also consistent with organic causation, since there are many physical disorders associated with primogeniture, especially in male infants. Pyloric stenosis, a digestive system disorder which becomes evident shortly after birth, has long been known to occur primarily in first-born male infants.

Among specific conditions having "mental" involvement, mongolism and anencephaly are reported to be disproportionately represented among the first-born (MacMahon and Sowa, 1961; Stott, 1960) as is epilepsy (Colver and Kerridge, 1962).

Studies which have attempted to relate birth order to parental behaviors and attitudes have far more often produced negative than positive results (Lasko, 1954). In view of these considerations, one can hardly argue that birth order data support psychogenesis in autism.

I have discussed the various factors that I have been able to identify or isolate as having been offered as evidence that early infantile autism is psychogenic in origin. In no case can it be argued that the psychogenic aspect of the factor is more potent than the biological aspect, nor, in fact that the psychogenic factor can be shown to have *any* potency at all in the causation of infantile autism. We are not saying that psychogenesis is an imaginary in-

fluence; we are merely saying that there appears to be no evidence that it is anything but imaginary.

It may appear that the case for psychogenic hypothesis has been understated. Yet the present writer is only one of many who have made a search for evidence and come to the conclusion that evidence is lacking. Stevenson (1957) has observed that:

> The literature of psychiatry abounds in articles asserting causal connections between the early experiences of life (especially training practices) and the later personality. The far fewer articles reporting objective studies of such relationships fail to support the assertions made (p. 152).

In addition to his own well-documented review, Stevenson cites three previous review articles. More recently, Hebb (1958b) and O'Connor and Franks (1961) have come to similar conclusions after reviewing the relevant research.

The Case for Biological Causation

Unlike the hypothesis that autism is psychogenically determined, there are a number of points of information which support the hypothesis that autism may result from a rare recessive trait, or be otherwise determined by biological factors. Kanner, in his various publications (especially with Eisenberg, 1956), has cited the first five points listed below as evidence against the psychogenic view. The remaining points have been identified by the present writer or others who have concerned themselves with this problem.

1. Some clearly autistic children are born of parents who do not fit the autistic parent personality pattern.
2. Parents who do fit the description of the supposedly pathogenic parent almost invariably have normal, non-autistic children.
3. With very few exceptions, the siblings of autistic children are normal.
4. Autistic children are behaviorally unusual "from the moment of birth."
5. There is a consistent ratio of three or four boys to one girl.
6. Virtually all cases of twins reported in the literature have been identical, with both twins afflicted.
7. Autism can occur or be closely simulated in children with known organic brain damage.
8. The symptomatology is highly unique and specific.
9. There is an absence of gradations of infantile autism which would create "blends" from normal to severely afflicted.

1. *Parents who do not fit the pattern.* "Some 10 per cent" of parents of autistic children are warm and friendly and do not fit the personality stereotype reported so commonly for these parents (Eisenberg and Kanner, 1956). This point is also made in Eisenberg's article on the fathers of autistic children (1957). Keeler (1957) reports autism in a child adopted by warm and loving foster parents within ten days after birth. No mention of the natural parents was made. The parents of van Krevelen's case are specifically described as warm and affectionate, as are those mentioned by Chapman (1957), Schachter (1958), and others. The father described by van Krevelen does fit the "intelligent-driving personality" stereotype, however. While the physician and lawyer described by Polan and Spencer fit Kanner's pattern of intelligence and seriousness, the mothers did not. One was a "warm and gentle person" and the other was an "intelligent and efficient individual of normal emotional spontaneity." There are numerous other examples in the literature.

2. *"Autistic-type parents" with only normal children.* Many parents who do fit the personality stereotype exactly and therefore might be expected to have autistic children if the psychogenic theory were correct, instead have perfectly normal children, as noted by Kanner and others (see especially Kanner, 1957), and as very probably can be verified from the reader's own experience.

The experiment conducted by Dennis (1941) on the development of infants under conditions of "minimum social stimulation" seems relevant to this point. Dennis raised fraternal twins from the end of their first month to the end of the fourteenth month by attending strictly to their material needs only. He gave no encouragement or approval of any sort. Despite this systematic attempt to serve the child in what must undoubtedly be a more detached and indifferent manner than Kanner's parents could have achieved inadvertently, Dennis concluded from his experiment that if the child's well-being was assured, his behavioral development would take its normal course.

Dennis' experimental treatment of children is certainly not to be recommended as routine. No one would willfully run the risk of injuring the child's sense of security, or any other aspect of his personality, even though the available research suggests that adverse effects, if there are any, may be too small to be detected by presently available methods. One is cautious even with a gun one has good reason to believe is not loaded. But Dennis' work certainly does suggest that the case for psychogenically induced adverse effect has been greatly overstated.

3. *Siblings are normal.* Kanner and a number of others have noted that many normal children are raised as siblings to an autistic child. It is a rarity to have more than one autistic child in a family, except in the case of twins. Of 131 siblings to Kanner's first 100 autistic children, only three could be

regarded as probably autistic. Seven others showed evidence of emotional disturbance (Kanner and Lesser, 1958). This is a small proportion, especially when it is considered that these siblings were probably under closer scrutiny than normal and under severe stress as well (Creak & Ini, 1960). Rattner & Chapman (1959) have reported an autistic child in the middle of a series of eleven normal children. Polan & Spencer (1959) reported two normal siblings for each of the three cases of autism they described. Phillips (1957) reports all siblings of his cases to be normal. Both siblings of van Krevelen's (1952) case were normal, as were also both siblings of the girl described by Poppella (1955) and by Sutton & Read (1958). Similar instances are plentiful.

4. *Autistic children unusual from the "moment of birth."* Kanner has often referred to autism as "inborn" and "innate." Even the psychogenically oriented writers do not appear to question this point. It is difficult to understand why pathology severe enough to be recognized so early is not considered severe enough to cause later behavior disorder without the compounding of it with psychogenic factors which have yet to be demonstated to be other than fictional.

5. *The high sex ratio.* That males are less viable than females from conception to old age is common knowledge. The attrition of males before birth is even greater than after. Males are susceptible not only to a great variety of sex-linked hereditary diseases, but also to later acquired infections and other adverse somatic conditions. Thus the occurrence of autism in three or four times as many boys as girls is consistent with the known greater vulnerability of males to organic damage. It would be hard to find a convincing psychogenically oriented explanation for this ratio, especially since boys tend to be more often welcome in our culture than girls, and especially so as first children.

6. *Twins with autism.* One of the strongest lines of evidence against psychogenic etiology of autism has come to light only in recent years. Keeler (1957, 1958) sems to have been the first to call attention to the incidence of autism in identical twins. He did not tabulate the cases of such twins known to him, but in a later personal communication to the writer (1960) Keeler referred to a set of identical twins in addition to the blind twins he had mentioned in 1958. Keeler pointed out that there were no known cases where only one of monozygotic twins was afflicted, nor where both of dizygotic twins exhibited autism. His 1957 paper refers to a set of fraternal twins where only one child was afflicted.

Kallman, Barrera, and Metzger (1940) also reported blind identical twins. These twin boys, congenitally blind with microphthalmia, showed motility patterns "strikingly similar" to those displayed by the blind children Keeler (1958) described as manifesting the syndrome of infantile autism. The verbal behavior of the twins described by Kallman et al. would also warrant their

being considered as possibly autistic. They could, for example, repeat anything said to them, but showed little comprehension of the meaning. This is very typical of autism.

Chapman (1957), in reporting a case of identical autistic twin girls, also raised the issue of twinness, and called attention to the cases of identical twin boys reported earlier by Sherwin (1953) and Bakwin (1954).

Polan and Spencer (1959) reported still another set of identical twin boys. They referred to this set as the fifth known to them.

The present writer, who had independently become impressed with the frequency with which twins with autism were being reported, has attempted to tabulate all cases of probable autism in the literature involving multiple births. The list follows:

1. Kallman, et al. (1940) identical twin boys.
2. Sherwin (1953) reported identical twin boys.
3. Bakwin (1954), another case of identical twin boys.
4. Eisenberg & Kanner (1956) described a set of twin boys, one of whom died in infancy before it could be discovered whether he was autistic like his brother. No reference to zygosity.
5. Keeler's (1957) fraternals, only one being autistic.
6. Chapman's (1957) identical twin girls.
7. Lehman, Haber & Lesser (1957) reported identical twin boys.
8. Keeler's (1958) blind twins. Presumably, from Keeler's report, these are boys.
9. Polan & Spencer's (1959) identical boys.
10. Bruch (1959) referred to twin Negro boys with autism. In personal communication to the writer (1961), she identified these as identical twins.
11. & 12. Chapman (1960) reported knowledge of two additional sets of identical twins.
13. Keeler (1960, personal communication) one other set of identical autistic twins. Sex not stated.
14. Ward & Hoddinott (1962) described a set of concordant fraternal twin girls who were "typical examples of early infantile autism."

This compilation raises the number of known multiple births involving at least one case of autism to fourteen. Known cases involving twins reported to be monozygotic is thus raised from five (Polan and Spencer) to a new total of eleven (excluding sets numbered 4, 5, and 14). Each of these sets is concordant (i.e., both twins similar with regard to affliction-nonaffliction). Kallman's set, reported prior to Kanner's delineation of autism, is the only one not so classified by the original author.

The identical autistic twin sons described by May (1958) in his book, *A*

Physician Looks at Psychiatry, have been included in the above list.

Kanner has made no specific mention of twins in his group, with the exception of set 4, above, referred to incidentally in illustrating a point concerning the twin's parents. No mention was made of this or other multiple births in either Kanner's 1954a listing nor Kanner & Lesser's 1958 detailed listing of the birth conditions and order of the first 100 cases, nor in the listing of the mental status of his autistic patients' siblings. (Correspondence has determined that the twins referred to by Eisenberg and Kanner were contained in separate sacs. Although dichorionic twins are usually fraternal, zygosity cannot be stated with certainty in this case.)

In general, only one of three twin births is monozygotic, so one would expect to find 22 cases of dizygotic twins for each eleven monozygotic cases reported. Even allowing for errors in establishing zygosity, this appears to represent a strong overloading of monozygotic twins with autism, as opposed to dizygotic. Additionally, since only about one birth in 285 is of monozygotic twins (Allen & Kallman, 1955), there would also appear to be an overloading of such twins in the absolute total number of cases of reported autism. Including the fewer than 150 cases Kanner has reported (1958b), there are probably no more than about 200 bona-fide cases of autism referred to in the literature, not nearly enough to support finding eleven twin cases, let alone eleven monozygotic twin cases, on a chance-only basis.

Luxenburger (cited by Rosanoff, Handy, & Plesset, 1937) has demonstrated that one may expect to find a disproportionately high incidence of monozygotic and concordant twins in twin studies based on search of the literature. The degree of overloading reported by Luxenburger (which was considerably greater than that reported by Rosanoff et al.) would not begin to account for the high incidence of twins with autism reported above. Autism seems especially likely to be reported, seen in single births, not only because its manifestations are so striking, but because the parents frequently take the child from clinic to clinic in the hope of finding someone who understands the disease (May, 1958; Rothenberg, 1960). Additionally, the early age of onset of autism, before irrevocable separation of twins is likely to take place, as well as an increased understanding of the importance of twin data to etiological research, make it appear unlikely that Luxenburger-type bias is an overriding or even important variable in the present instance.

The finding of at least eleven sets of monozygotic twins, all concordant, seems highly significant in terms of the biological etiology of the disease. Kety (1959) has remarked that Chapman's (1957) reference to three sets of identical autistic twins was suggestive. Ward & Hoddinott (1962), in describing their set of concordant fraternal twins, observe that for infantile autism, "to appear in twins, particularly fraternal twins, is an event beyond statistical probability" (p. 191). That one of the two cases of reported fraternal twins is discordant (set Number 5) serves only to enhance this finding. (The twin

who died at five months in set 4, probably fraternal, was reported only as "more responsive" than his definitely autistic brother.)

The high susceptibility of twins to neurological defects does not adequately account for the high incidence of *identical* twins with autism. While the ratio of *defective* monozygotic to dizygotic twins is lower than the one-to-one ratio found for nondefective twins (Rosanoff, et al., 1937; Allen & Kallman, 1955), the preponderance of monozygotic twins with autism is none the less striking.

Kallman's data (1953, 1956), widely accepted by the scientific community as providing the clearest evidence of the strong hereditary component in mental disorder (e.g., Knobloch & Pasamanick, 1961; Rosenthal, 1961; Meehl, 1962), show 9 per cent of non-twin siblings to be schizophrenic when one sibling is affected, 15 per cent of dizygotic twins to be concordant if one twin is schizophrenic and 86 per cent concordance if one of monozygotic twins is schizophrenic. When schizophrenia was diagnosed at an early age, between five and eleven years, only 71 per cent of identical twins were concordant. Judged by these standards, the genetic element in autism would appear to be unusually strong.

It is of interest that in several cases of identical twins stricken with autism, the degree of affliction, while invariably severe, is not quite identical (Sherwin; Lehman et al.; Polan & Spencer). This suggests that while genetic factors may predispose toward autism post-conceptional factors could be operative (see Burt, 1958; and Price, cited by Rosenthal, 1961, for references on prenatal identical twin differences).

7. *Austim caused or simulated by known organic disease.* A line of evidence which suggests the organicity of early infantile autism stems from the many cases in which the symptomatology may be traceable to a postencephalitic condition or other form of central nervous system injury. Some of these cases are those described by Anthony (1958), Rattner and Chapman (1959), Frankl (1943; see Sarason & Gladwin, 1958, p. 216); Ross (1959), Sutton and Read (1958), van Krevelen (1960), and possibly the cases of Anastasi and Levce (1960), Kallman et al. (1940), Poppella (1955), Schacter (1958), and Vaillant (1962). The appearance of the syndrome in children given oxygen shortly after birth is also germane to this point.

These cases do not prove autism is *invariably* organic, but they at least indicate that the symptoms *can* be caused by organic agents. It is not believed that there is parallel evidence supporting the view that the symptoms of autism *can* be produced by sociopsychological factors.

8. *Uniqueness and specificity.* The high degree of interindividual similarity in the symptomatology of autism would seem to identify it as biological. As May (personal communication, 1959) has observed:

> I do not know any one of these children who did not have the rocking and banging of head . . . the particular interest in some machinery . . . the

> typical speech patterns . . . This identity of symptomatology pleads strongly for a well-localized lesion . . . .

Much experimental research in genetics has shown that what appears to be a complicated and varied syndrome can be traced back to a single initial fault in development (Roberts, 1959). It should be noted, however, that while a single abnormal gene may produce a remarkable variety of symptoms *within* an individual, the syndrome from individual to individual tends to be highly uniform, especially when it is a recessive gene that is at fault. In phenylketonuria, for example, there is a characteristic diversity of problems: motor involvement, seizures, eye and skin pigmentation effects, and skin sensitivity in addition to mental retardation. Yet each victim exhibits the *same* syndrome, because there is but a single recessive underlying defect.

That gene effects within the nervous system can be highly specific does not seem to be widely appreciated. C. E. Keeler (1940) has described an interesting study in which the exact locus of the inherited nerve lesion causing "waltzing" in guinea pigs was located. It was then possible to duplicate the inherited waltzing tendency by making carefully placed surgical lesions in normal animals. Keeler observed that, "So steady is the hand of genetics and so accurate in repeating these operations that you may have a thousand identically operated specimens to study quite as readily as one" (p. 97).

Moorhead, Mellman, and Wenar (1961) have recently made a related finding in humans by tracing a family defect involving speech and mental retardation to a heritable chromosome translocation.

It may be of interest to note the many similarities in the symptoms of autism with those described for "brain-injured" children by Lewis, Strauss, and Lehtinen (1951); and by Strauss and Lehtinen in other publications. The similarities between autism and the symptomatology of brain injury should not permit the confusion of autism with the general brain-injured syndrome. The more general term "brain-injured" encompasses many cases whose lesions apparently include involvement of the brain areas in which profound but localized dysfunction, usually in genetically sensitive infants, may produce true early infantile autism.

Kanner has observed that some autisticlike symptoms are found in brain-injured (and innately retarded) children (1958b). However, the "brain-injured" in this sense will rarely show the *composite* of symptoms which defines autism. In Kanner's words, "The symptom combination in most instances warrants an unequivocal diagnostic formulation" (1951, p. 23).

9. The absence of "blends." If autism were a reaction to environmental factors we would expect it to exhibit not only the diversity of manifestations from case to case as a consequence of situational differences (above), but, in addition, the usual gradation in intensity, depending on the adverseness of the environment. Environmental adversity would ordinarily be assumed to

follow a continuous, probably Gaussian distribution, rather than a dichotomous one. While there is variation in severity and in prognosis, the degree of variation does not account for the large void between autism and normal behavior. There have been few serious attempts to deny the existence of this void.

Psychogenesis as an Inadequate and Pernicious Hypothesis

Perhaps it should be made explicit at this point that the writer does not presume to have shown that autism is biologically determined and that the psycho-social environment plays no part in its etiology. What the writer *does* assert is that a careful review of the evidence has revealed no support for the psychogenic point of view. The evidence is instead highly consistent with expectation based on organic pathology.

Our finding with regard to autism coincides with the more general view formulated by the participants of a recent conference on the causes of mental disorder (Milbank Memorial Fund, 1961):

> There seems to be no clearly demonstrated instance of either a cultural or social factor being known to be a predisposing factor in mental illness. . . . The absence of clear-cut evidence does not show that the hypothesis is incorrect but only that it has not been demonstrated even once (p. 379).

Neither Creak and Ini (1960), who intensively studied 100 sets of parents of psychotic children, nor Peck, Rabinovitch, and Cramer (1949), who studied 50 sets of parents, were able to find evidence of a psychogenic nature. What they did find was a good deal of suffering brought on by the child's behavior and a good deal of intense (and we might add unnecessary) feelings of guilt.

It is probably too early to suggest that psychogenesis as a *hypothesis* no longer be considered. ("Hypothesis" is used advisedly, because there appears to be too little evidence to support use of the term "theory.") No avenue for learning all that we can about the etiology of mental disorder should be unexplored. The detailed explication in this chapter of the arguments concerning the etiology of autism was in part intended to facilitate, and perhaps even to provoke, some long-overdue, rational, and articulate consideration of the problem, even at the expense of jointly provoking a measure of articulate and inarticulate wrath.

It is not questioned that distinction should be maintained between a disproven and an unproven hypothesis, but neither should there be a failure to distinguish between an unproven and an uninvestigated hypothesis. The psychogenic hypothesis is by no means uninvestigated.

Whatever may be the merit in being patient with psychogenesis as a hypothesis, there is much less in being patient with it as an assumed force-in-fact. The all too common practice of blatantly assuming that psychogenic etiology *can* exist or *does* exist in any individual case or in any given class of disorders is not only unwarranted but actively pernicious.

It is perhaps permissible for writers such as Weiland & Rudnik (1961) to "postulate" that "the expectation of murderous attack or of symbiotic engulfment by a psychotogenic mother results in a failure to progress beyond autism and in panicky attempts to escape from symbiosis into autism" (p. 552), especially when they add (in a footnote) "We do not believe this has been demonstrated conclusively. . . ." It is something else again when the implications of this view are translated into action, and psychogenic causation is assumed to be a reality rather than merely a hypothetical possibility.

Ross (1959) presents an interesting and instructive case history of an autistic girl whose mother had been held to be responsible for her child's plight because the mother's affection was considered "intellectualized and objectified." Only when the child died did the fact of extensive brain damage become evident. Intensive neurological examination had failed to reveal the difficulty before the child's death, which interrupted intensive psychotherapy. It was fortunate for the mother that the brain damage was of a sort that present-day techniques could disclose, else she would to this day be held responsible for her daughter's death.

Consider the case of "Jonny" (Rothenberg, 1960). After stating that Jonny, at 1½ pounds, was one of the smallest premature babies ever born in the United States to survive, and after noting that 3½ months spent in an incubator under high oxygen tension and heat lamps had turned the infant's hair orange and his skin chocolate brown, the author attributed his later severe behavior disturbance to lack of mothering while being incubated. "Cure" was said to be greatly facilitated by suddenly confronting Jonny with a model of the incubator—a personification of the mother who supplies only material needs and no nourishment of the ego. The possibility that organic brain damage might have resulted from such adverse physical conditions was apparently not seriously considered, yet it had been known for some time that a high concentration of oxygen is able to cause destruction of nerve tissue in infants.

Bettelheim (1959a) interpreted the psychosis of "Joey, the Mechanical Boy" as a reaction to his mother's hostility when the evidence was also quite consistent with hereditary determination, since the mother appeared to be severely mentally disturbed herself. (See also the reply by May to this article.) In this case, as in Rothenberg's, the appeal of the psychogenic concept appeared to preclude consideration of concealed organic defect. Somehow the adherents of the psychogenic hypothesis tend to overlook the possibility that

the complex and little understood cerebrum could be structurally or chemically impaired.

In another case Bettelheim (1959b) found emotional isolation to have caused the psychosis of a girl who had been conceived, born, and raised by her Jewish parents in World War II in a small hole beneath a farm building in Poland. The hole was too cramped to permit an adult to stretch out. German soldiers were nearby (sometimes firing shots into the building) so that the mother had to smother the infant's cries. Not considered by Bettelheim was research showing the adverse effects on the offspring's emotionality of prenatal stress in the mother (e.g., Thompson, 1957), nor of sensory deprivation in the child.

It should not be thought, however, that workers in this field are universal in accepting environmental determination. Some writers have been frank in rejecting the psychogenic hypothesis. Keeler (1957), for example, has said, "I certainly do not adhere to the opinion put forth by some that infantile autism stems from a very specific type of pathological parent-child (especially mother-child) relationship." Anthony (1958) notes, "I do not think that traumata which sometimes seem to precipitate a psychosis in childhood are anything greater than normal developmental hazards (sibling birth, etc.). It is the predisposition that makes them vulnerable." Chapman (1960) also believes the role of the psycho-social environment has been overestimated: "the degree of interpersonal pathology between parents and child rarely seems sufficient to explain the catastrophic interpersonal disorder of the child." Goldstein (1959), in his very illuminating discussion of autism, has pointed out clearly the gratuitousness of assuming psychogenesis as an etiologic factor in the disease.

In discussing the obvious prejudice against the hereditary viewpoint, Nolan Lewis (1954) points out, "It would seem that most of the prejudice against genetic inheritance stems from a feeling in the realm of wish fulfillment, based on the idea that acceptance of genetic factors would create an attitude of therapeutic hopelessness." Williams (1956) cites this point among others in his attempt to penetrate the prejudice against heredity. He notes that hopelessness is by no means justified by the evidence, and cites the ready correction of the effects of diabetes, phenylketonuria, and hypothydroidism as examples.

It should not be necessary to ask for recognition of the role played by genetic factors among persons trained in scientific thinking, but Williams has seen the need to do so:

> We therefore make a plea for an unprejudiced facing of the facts of heredity. We urge that such facts be accepted with as great readiness as any others. This plea seems necessary in view of the attitude which we have

> repeatedly noted, namely, that of willingness to arrive at "environmentalistic" conclusions on the basis of slender evidence while rejecting points of view which would emphasize the role of heredity, even though the weight of the evidence, viewed without prejudice, appears overwhelming (p. 16).

When dark-haired and dark-eyed parents produce a dark-complexioned child, we all are quick to agree, "Mendel was right!" But when introverted parents produce a child who similarly shows little interest in socialization, the refrain inexplicably changes to "Aha, Freud was right!"

In arguing for more critical use of the diagnosis of autism, Kanner says:

> The misuse of the diagnosis of autism has played havoc with the comfort and finances of many parents of retarded children, who were made to feel that their attitudes and practices were primarily responsible for their offspring's problems, were made to submit themselves and the child to lengthy, expensive, and futile therapy, and were pauperized and miserable to the time the true state of affairs was brought to light (1958a, p. 111).

Kanner's appeal for the protection of the parents of children misdiagnosed as autistic is certainly to be commended, but what of the parents of *accurately* diagnosed cases? In view of the present status of research on the efficacy of psychotherapy, and of the fact that the evidence for psychogenic etiology of autism is not, to use Kanner's term, "unequivocal" (1958a), it would seem that the parents of properly diagnosed autistic children might also be deferred from being made "pauperized and miserable," for the time being.

In a court of law it is impermissible to convict a person solely on evidence consistent with the hypothesis that he is guilty—the evidence must also be inconsistent with the hypothesis that he is innocent. This simple point of justice has been neglected, consistently, by those who deal with families having children afflicted with autism, and the damage and torment this practice has wrought upon parents whose lives and hopes have already been shattered by their child's illness is not easy to imagine nor pleasant to contemplate. To add a heavy burden of shame and guilt to the distress of people whose hopes, social life, finances, well-being, and feelings of worth have been all but destroyed seems heartless and inconsiderate in the extreme. Yet it is done, as May (1958), Oppenheim (1961), Stuart (1960), and van Krevelen (1960) amply illustrate.

In view of these pernicious implications and the absence of scientific evidence, the wide acceptance of the psychogenic view is difficult to understand. A partial explanation for the prevalence of this view may be found in Kanner's unguarded admission that he is perplexed by the fact that the great

majority of the parents of autistic children have been able to rear non-autistic children, while other parents who fit the parental typology perfectly, raised children who responded aggressively rather than by withdrawal: "the existence of these exceptions is puzzling. . . . It is not easy to account for this difference of reaction" (1949, p. 426). The same "puzzling" inconsistency is clearly present in other childhood mental disorders, such as mongolism, Tay-Sachs disease, and phenylketonuria, and is readily explained in terms of recessive inheritance.

Despert provides another example of a child psychiatrist who does not apply what she must certainly know of genetics to her thoughts on etiology:

> It is sometimes argued that these mothers had other children who were normal or relatively normal, but it must be remembered that a mother, biogenetically identical for all her children, may nevertheless psychogenetically differ widely from one child to the other (1951, p. 345).

"Biogenetically identical"—100 years after Mendel!

Perhaps we are painting too dark a picture. There are signs of a growing recognition that the failure to find support for psychogenesis may possibly lie in the inadequacy of the concept rather than in a lack of resourcefulness among its investigators. Despert, whose 1951 view was quoted immediately above and who in 1947 wrote of cases in which neurological disorders had been "ruled out" by examination, has written in 1958 that the possibility of finding constitutional factors in infantile autism was particularly strong (Despert & Sherwin, 1958). Ekstein, Bryant, and Friedman (1958) show a willingness to question "our prejudiced, one-sided consideration of etiological factors" (p. 653); and Bettelheim, for years a leader of the psychogenic school, was recently willing to "reserve judgment" about what causes autism, although he is still "pretty sure" that psychogenic factors "contribute" (1959b, p. 463).

Szurek, who started in 1946 "to test the hypothesis that the etiology of psychotic disorders of childhood are entirely psychogenic" (Boatman & Szurek, 1960; p. 389) takes a much weaker stand today in stating that certain "facts" "seem to lend weight to the possibility that psychogenic factors are at least important" (p. 430). (The reader may wish to see these "facts.")

To the present writer these indications of a retreat from the psychogenic hypothesis, like Bowlby's previously cited disavowal of the maternal deprivation hypothesis, represent a timely and welcome willingness to let conviction be subordinated to evidence. The history of science proves this to be the first step toward progress.

COMMENTARY by David L. Holmes

Rimland's chapter, "The Etiology of Infantile Autism: The Problem of Biological Versus Psychological Causation," in his book, *Infantile Autism* (1964), is probably the most significant statement on the psychogenic versus biogenic causation of autism. This treatise set the tone for more critical analysis of the etiology of autism, resulting in reduced credibility of the psychogenic theory of causation, and increased interest in viewing autism from a biogenic vantage point. Although criticized at the time of its publication as the subjective ravings of a distraught father, Rimland held to his posture that the evidence is lacking for a psychogenic causation of autism, and that there is far more support for the hypothesis that autism is a biologically determined disorder.

Rimland's hypothesis of a biogenic causation of autism has held the test of time, and, in fact, is the foundation upon which research into the biophysical nature of autism has followed from the late 1960s through the present. Further, Rimland's chapter was also successful in stimulating work with parents, viewing them not as causative agents in autism, but rather, as potential members of a helping team to reduce the effects of the syndrome on the child.

It is this reviewer's opinion that Rimland's work, "The Etiology of Infantile Autism: The Problem of Biological Versus Psychological Causation," is one of the most significant treatises in the development of current research trends into the etiology of autism, as well as being an effective training source for parents who act as cotherapists in the treatment of their children with autism.

6

Epidemiology of Autistic Conditions in Young Children

VICTOR LOTTER

PART 1. PREVALENCE

Introduction

In spite of an increasingly voluminous literature about "psychotic" disturbances in children, etiology remains obscure (Kanner, 1957), the lack of agreed behavioural criteria reflects the nosological uncertainties and prevents the useful comparison of findings (Despert and Sherwin, 1958; Tramer, 1962; Rimland, 1964) and prevalence is unknown (Bellak, 1958).

Adequate behavioural descriptions are rare. The most detailed are still those made for "early infantile autism" by Kanner over 20 years ago, and in a number of papers since then (e.g., Kanner, 1943, 1946, 1951, 1954; Eisenberg and Kanner, 1956; Kanner and Lesser, 1958). In addition, detailed descriptions of certain aspects of behaviour were made by Norman (1954, 1955) for a group of 25 "schizophrenic" children. In 1936, Earl described in some detail the behaviour of a small group with what he called "the catatonic psychosis of idiocy," while three children with Heller's syndrome (classified as a "schizophrenic" reaction) were reported in detail by Yakovlev et al. (1948). Since the present survey was completed, two detailed behavioural studies of "psychotic" children (Rutter, 1966) and "schizophrenic" children (Wolff and Chess, 1964) have been reported.

The differences between the behaviour manifested in "psychotic" children who become ill during early childhood and those whose illness is of later onset have usually been attributed to the immaturity of the former (Potter, 1933; Kanner, 1957). However, from Kanner's study of "infantile autism" much other evidence may be derived to suggest that the disturbances in the

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