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HEREDITY, ENVIRONMENT AND MENTAL SUBNORMALITY

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In the good old days when A. F. Tredgold (1908) wrote his textbook it was acceptable to divide subnormality—then called amentia—into primary and secondary types. Primary cases were caused by heredity and secondary by environment (see Table 1). Equivalent terms favoured abroad were endogenous and exogenous and these are still employed to distinguish supposed origins of psychotic states. The separation of the two influences is of great antiquity and they were called nature and nurture. Literally, of course, nature means what we are born with and nurture means how we are nourished. In this sense the contrast is between what is congenital and what is acquired. This is a distinction commonly made in medicine and it is much easier to apply than more philosophical concepts which imply a knowledge of the hereditary processes. The traditional view was that the influence of nature was paramount, e.g., "The Tempest," Act IV, Sc. 1, Prospero's assessment of Caliban, "A devil, a born devil, on whose nature nurture can never stick; on whom my pains, humanely taken, all, all lost, quite lost, and as, with age, his body uglier grows, so his mind cankers."

TABLE 1

Estimates of Primary and Secondary Cases

	Hereditary (Primary)	Environmental (Secondary)
Tredgold (1929)	80%	20%
Larsen (1931)	76%	24%
Penrose (1933)	29% —(62%)—	9%

In the last row, 62% are not considered classifiable under either heading

One of the earliest attempts to examine the problem scientifically was made by Francis Galton who pointed out, in 1875, that identical and fraternal twin pairs might be particularly useful in this respect. The principle he relied on was that identical twins would have the same hereditary qualities as one another and so differences which developed must be attributed to environment.

An enormous amount of investigation has been carried out on the intelligence, and on physical characters associated with intellectual defects, in twin pairs. The traditional methods of analysis of the data (e.g., J. C. Smith, 1930), indicate, in most examples, that heredity has a very strong effect in determining these variations (e.g., 98%) (see Table 2). The key information concerns the degree of "concordance" of monozygotic twins. Features that remain constant in both members of

the pair, in spite of exposure to environmental influences of all kinds, are assumed to have been determined by inborn constitution. As identical twins are in fact nearly always very similar to one another the inference is that almost all physical and mental traits are essentially inborn. It used also to be supposed that any differences found between such twin pairs must be environmental.

TABLE 2
Mental defect in twins*

Monozygotic		Dizygotic	
++ (a)	+− (b)	++ (c)	+− (d)
14	2	4	46

* from Smith (1930)

Estimate of hereditary influence, equivalent of Holzinger's (1929) coefficient,
 $H^2 = (ad - bc) / (ad + \frac{1}{2}bd) = 0.92$

++ indicates both affected, (a) and (c)

+− indicates one twin affected, one normal, (b) and (d)

Measurements of likeness can be obtained by correlation techniques; they are especially useful in the study of metrical characters such as stature, weight and intelligence. If there were no environmental effects, in theory, every correlation for identical twins would be +1.0; for intelligence tests they are round +0.9 and this can be interpreted as meaning that 90 per cent of causes of variations in intellect are hereditary. These arguments are very convincing and, at first, seem to be watertight. However, there are some curious discrepancies. Correlation measurements of abilities of children who are unrelated but reared together in the same family group have proved to be surprisingly high, +0.27 for intelligence and about +0.5 for arithmetic, as reported by Burt (1966). These results suggest that from one quarter to a half of variations in mental capacity are environmentally determined.

There is one obvious difficulty in the interpretation of correlation coefficients and this is that they only tell us what happens in the mass. They do not help to explain the origin of defect in a given case and so cannot be used to divide patients individually into primary and secondary groups. Besides, there are additional, practical difficulties which arise out of the study of twins and predictions from them. Some of these are exemplified by a Swedish study on cattle in order to determine how far milk yield and muscle development could be improved by breeding. Investigations on twin beasts showed that about 90 per cent of the variation was hereditary because identical twin pairs were so much alike. Direct breeding, however, showed that only about 40 per cent of variations in these characters were hereditary. It would appear that there is something basically unsound in the twin method—or, if not perhaps unsound, something hidden and treacherous.

Let us look more closely into the nature of twins. First, there are our old friends the conjoined twins. These, it is almost always supposed, must be identical and derived from one imperfectly divided embryo. H. H. Newman (1917) however, believed that they were normal identical pairs inadvertently united by growing too

close together in the earliest developmental stages. Anyway, these conjoined pairs are usually less similar than those normally separated. The original Siamese twins were very different from one another in temperament and used to come to blows occasionally. Sometimes one conjoined twin fails altogether to develop or may be grossly malformed (e.g. acardiac or anencephalic). Such defects may be caused by differences in interuterine environment but they can also arise in consequence of genetical mutations. The modern genetical methods of chromosome identification have shown that it is even possible for a pair of identical twins to have different karyotypes, i.e. different numbers of chromosomes.

An example turned up in France of a man and a woman, born in 1944, whose blood groups were in perfect agreement and who were monochorionic. Their intelligences were both above normal—I.Q's 125 and 104 respectively. Cell cultures revealed that the man was a normal male but that the woman was an example of Turner's syndrome with 45 chromosomes. She had lost a Y at a very early cell division or at the time of twinning. This account might have been regarded with suspicion by some had not a somewhat similar pair been discovered soon after by J. H. Edwards, T. Dent and J. Kahn (1966) in Birmingham. Though Shakespeare may have been wrong about heredity and environment he was unexpectedly right about the possibility of male and female identical twins. In "Twelfth Night," the whole play depends upon this phenomenon.

More relevant, in the context of subnormality, was the startling twin pair, found in Switzerland, apparently identical but with one twin a normal boy and the other a quite typical mongol. Again doubts have been eased by the advent of another similar case in female twins reported by Dekaban (1966) from America. What are we to say about these mongols? Are they primary or secondary cases?

One could go on indefinitely pointing out peculiarities of twins. Indeed the main characteristic of twins is that they are peculiar. They are exceptional and so should not be used, without great caution, for prediction about the general population. Twins are abnormally light at birth, many of them have poor health in early life and they are liable to malformation. Moreover, they are subject to influences in embryonic and foetal life which do not affect single births. Besides the risk of being conjoined there may be interchange of tissues. This is characteristic of fraternal twins, who can partly share each other's blood groups. Most influences, however, act on both twins equally and tend to increase their similarity. We can summarize the situation in the general rule that the presence of a trait in both members of a pair of identical twins is consistent with this trait's being determined by heredity; but it does not prove it.

Getting away from twins and coming back to the practical problem of deciding whether, in a given case, subnormal intelligence is to be attributed to nature or to nurture, the first necessity is accurate diagnosis of concomitant disease. It may be, of course, that a disease observed is only a chance accompaniment—if so, we must search for other abnormalities. At the present time, in a general mental deficiency hospital, we can, for example, find diagnosable single gene hereditary conditions, such as phenylketonuria, true microcephaly, hereditary diplegia and hereditary epilepsy, in about 5 per cent of our patients, dominant defects, like epiloia, in 1 per cent and sex-linked defects, like hydrocephalus, in 1 per cent; that is to say, not more than 7 per cent (or about one case in 15) are typically hereditary in the Mendelian sense.

Chromosome abnormalities, represented by mongols, make up 10 per cent. Klinefelters, triple-X females and double-Y males, as well as all kinds of rare aberrations, probably add another 5 per cent.

If all the cases of genetical origin are scored as hereditary cases, then 22 per cent of patients are definitely in the primary category of Tredgold. On the other extreme, cases with a definite history of specific illnesses which can have injured the brain, encephalitis, birth injury, etc., probably do not make up more than about 5 per cent altogether (see Table 3). This means that, for certain, we know the cause in little more than one quarter of all cases of subnormality.

TABLE 3
Causes of Mental Subnormality

Clinical group (mainly severe)		Residual group (mainly mild)		Total
Single gene effects	7%	Multiple gene effects	15%	37%
Recessive	5%			
Dominant	1%			
Sex-linked	1%			
Chromosomal errors	15%			
Mongolism	10%			
Other autosomal	2%			
Sex	3%			
Specific illnesses, infections, foetal injuries, etc.	5%	Miscellaneous environmental influences	15%	20%
Unknown	43%		—	43%
Total	70%		30%	100%

So far, however, I have left out what E. O. Lewis (1933) called the subcultural group which may account for 30 per cent. of all our patients. These are the residual, aclinical, relatively high grade cases who are not physically ill but who may, as well as being scholastically inadequate, suffer from emotional disturbances, epilepsy and even psychoses. After discounting the effect of such illnesses, in none of which is the hereditary background satisfactorily understood, we still have to deal with the low mental ability which may be, in theory, inherited or acquired. It is here that we may need the population surveys and the familial correlations to decide on the relative significance of heredity and environment. Originally Lewis believed that the whole group could be considered to be of hereditary origin. It was the lower part of the intelligence distribution determined by normal hereditary variation in the human species. On this basis all subcultural defectives would be primary. Applying the results of genetical studies, twin studies and the like, most investigators in the past have preferred an approximate figure of, say, one part environment to two parts heredity. More recently, encouraged by the uncertainty of intelligence tests as prognostic criteria and by observed effects of good teaching, many psychologists have almost denied that heredity has any perceptible effect on the distribution of mental ability.

I hesitate to go deeply into this problem on this occasion. There is much to be said on both sides. As an approximation, there is no harm in attributing about half the variation to nature and about half to nurture. This, in one way, is formally

equivalent to diagnosing half the subcultural patients as primary and half as secondary. Such a decision would bring in almost another 30 per cent of cases with known cause. Our group attributable to heredity would rise to 37 per cent and those attributable to environment to 20 per cent. Still this leaves about half our patients non-classifiable yet in such terms.

Personally, I have always deprecated the attempt to divide mental subnormality into primary and secondary classes. As L. Hogben (1933) once said, the only thing which is simple about simple primary amentia is the mind of the doctor who diagnoses it. In the great majority of cases we have to deal with a complex interaction of factors, inborn and acquired, which lead to the end result of a patient recognized to be retarded. Each factor has to be separately investigated and appreciated in its own right before the combined aetiological effect can be appreciated.

Consider such a disease as kernicterus caused by *Rhesus* immunization. Undoubtedly the origin is hereditary but the effect is produced by unfavourable foetal environment. When it produces subnormality, this cannot be classed either as primary or secondary. Again, what is to be done with a fresh mutation of a gene or a chromosomal error, caused by irradiation, by poisoning or perhaps by virus infection just before or after fertilization? The cause is environmental but the effect is genetical. So the case belongs to both classes. In the study of mentally ill patients, it is commonplace to attribute psychotic breakdown to the interaction of internal and external factors: clearly this way of thinking is also reasonable in mental deficiency.

In the past, one of the main urges, to classify defectives into the two traditional groups, was the idea that secondary cases could most likely be treated favourably, possibly even cured, whereas primary cases could not be treated but might be prevented. This is an obvious fallacy; for some intractable hereditary conditions, like phenylketonuria and Wilson's disease, can be controlled, if not absolutely cured, by diet or drugs. Some diseases of partly genetical origin, like kernicterus, can be preventively controlled by treating the mother or the infant. In the old days sterilization would have been the only proposal for dealing with such situations.

Many of the diseases caused by chromosomal aberrations, particularly those concerned with non-disjunction, could be much reduced in numbers by avoiding pregnancies at late maternal ages. Others, possibly caused by virus infections affecting germ cells of the parents, may also eventually be guarded against in the future by immunization if not by avoiding infection.

In the subcultural group, there is reason to believe that improved standards of living and education, in the general community, are reducing the numbers of such patients who should require hospital treatment. In any case, increasing opportunities for employment, as compared with 30 years ago, have reduced the pressure for admission of these subculturals at the present time. There has never been more than a small fraction of the population section, whose I.Q.'s range between say 55 and 85, who needed to be admitted to hospital, say 1/10, and this proportion is sensitive to the attitude of society as well as to economic conditions.

Although, in recent years, many of the extensions in our knowledge of the causes and pathology of mental defect have come from increased understanding of heredity, the result has, paradoxically, been to emphasise the value of manipulating the environment in the interests of therapy (rather than the attack on bad genes, popular earlier). The belief that a family history of tuberculosis, alcoholism or moral depravity was a determining factor in producing imbecility has long since been (I hope) discarded. In its place we can now, in many instances, after accurate diagnostic procedures have been carried out, give exact information to parents about here-

ditary causes. They need feel no more disgraced by this knowledge than they would have formerly felt on the attribution of their child's illness to infectious disease. Even if this may not yet help with the treatment of the child, it is good mental therapy for the parents. The stigma of mental defect has almost disappeared and this, itself, is a great therapeutic advance.

The battle, between those who believe that nature is all powerful and those who hold the same view but about nurture, continues. The present trend of thought in mental deficiency work, however, is to expand our knowledge of both processes to the utmost and so aim to make use of nurture to defeat the errors of nature. Successes, so far, have only been in small fields but the way has been opened, I believe, for continuing steady advances.

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