BLOOD GROUPING AND HUMAN TRISOMY

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LEJEUNE, Gautier and Turpin (1959) have shown that the human abnormality rather unfortunately termed mongoloid imbecility is usually due to trisomy, that is to say the presence of three, instead of two, of one of the 22 kinds of autosome. Penrose, Ellis and Delhanty (1960) have shown that it is sometimes due to partial trisomy resulting from translocation in an earlier generation. At least two other types of abnormality due to trisomy of other chromosomes have been described. These abnormalities no doubt occur in India, though they may be rarer than in Western Europe and North America, because mongolism, at least, is mainly found in the children of mothers over 40 years old, and such births are relatively rare in India.

Consider a mother belonging to group AB who bears a trisomic mongoloid infant. It probably receives two chromosomes from its mother. Let us consider what is expected if these chromosomes include the ABO locus. In the present state of our ignorance we cannot deny that they may sometimes be two A's or two B's. However, on almost any hypothesis they should sometimes be an A and a B. Thus if the ABO locus is on the chromosome of which a mongoloid possesses three, some mongoloid children of AB mothers and O fathers should belong to group AB.

Two other types of marriage could give evidence of the same kind. These are :—

$$\begin{array}{c} A_1B \times A_2 \\ | \\ A_1B \end{array} \quad , \text{ and } \quad \begin{array}{c} A_2B \times A_1 \\ | \\ A_2B \end{array} \quad ,$$

provided that in the last case, the A_1 parent can be shown, either from examination of other children, or of the parents of the A_1 parent, to be A_1O and not A_1A_2.

In other cases the evidence from a single trisomic could not be decisive, but from a group of trisomics it could be so. For example, from AB × B we expect a little over ¼ AB offspring. If this was significantly exceeded, we should have evidence for the location of the ABO locus. In fact I only know of one mother of a mongoloid who belonged to group AB. Her case is recorded by Lang-Brown, Lawler, and Penrose (1953). She was A_1B, married an A_1 man, and had two mongoloid children of groups A_1B and B. It is thus certain that she produced one gamete not carrying both A and B, and she may have produced two. The argument is not essentially different in mongoloidism and other abnormalities due to partial trisomy following translocation, except that the marker locus must not be at the opposite end of the chromosome from the section whose trisomy is responsible for the abnormality.

The frequency of group AB in Britain is very low, about 3%. In India it probably averages about 8%, and many samples contain over 10%. No such high frequencies are found in Europe or North America, though they are equalled in Japan. Thus there is good reason for a search in India on the lines indicated. With suitable antisera the Rh and MNS loci could be studied in the same way ; but such sera are rare in India, and the genotype frequencies at these loci give India no advantage over Europe.

I therefore appeal to Indian physicians and geneticists to investigate the blood groups of mongoloid and other trisomic human beings and of their parents, brothers and sisters. A fuller account of the theory will be published elsewhere. It is of course likely that the conclusions will be negative. But a proof that the ABO locus was not on the chromosome responsible for mongoloidism would be a contribution to human biology.

1. Lang-Brown, H., Lawler, S. D. and Penrose, L. S., *Ann. Eugen.*, 1953, **17**, 307-36.
2. Lejeune, J., Gautier, M. and Turpin, R., *C.R. Acad. Sci.*, 1959, **248**, 1721.
3. Penrose, L. S., Ellis, J. R. and Delhanty, J. D. A., *Lancet II*, 1960, 409.