

his mother. Though his children show neither of these conditions at present, they will be kept under observation to see if either or both disorders develop in the future.

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A giant short arm of no. 21 chromosome in mother of 21/21 translocation mongol

Summary. An extreme variation of the short arm of no. 21 chromosome in the mother of a 21/21 translocation mongol is described. The possible relation between the very long short arm of chromosome no. 21 in the mother and a centric fusion type of translocation mongolism in the offspring is discussed.

We recently encountered an interesting family where the mother had an abnormal no. 21 chromosome and the child had a centric fusion type of translocation Down's syndrome.

Case report

The patient was a 10-year-old boy with the typical features of Down's syndrome. The mother was 20 and the father was 25 years old at the time of his birth. The mother's phenotype was entirely normal.

Chromosome studies

The patient's chromosome analysis showed that he had 46 chromosomes with 21/21 centric fusion type translocation (or isochromosome of no. 21 chromosome). This cytogenetic finding was confirmed by trypsin banding technique (Fig. 1).

The father's karyotype was normal. The pheno-

typically normal mother's modal chromosome number was also 46, but one of the G group chromosomes was different from the other members of this group, with grossly enlarged short arms. Trypsin banded preparations showed the chromosome concerned to be a no. 21 (Fig. 2). The short arms of this abnormal chromosome were positively stained in C-banded preparations (Fig. 2). They were dull when stained with quinacrine, apart from tiny fluorescent satellites on their tips.

The abnormal no. 21 chromosome was interpreted as an extreme example of the well-recognized variation in size of the heterochromatic short arms of acrocentric chromosomes (21ph+).

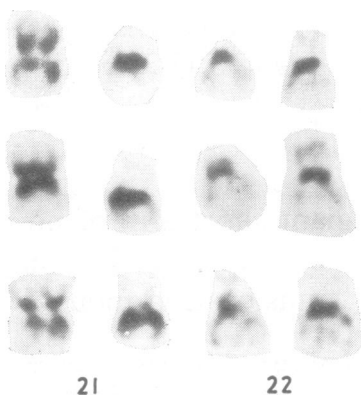


FIG. 1. Partial karyotype of the patient from trypsin banded preparations.

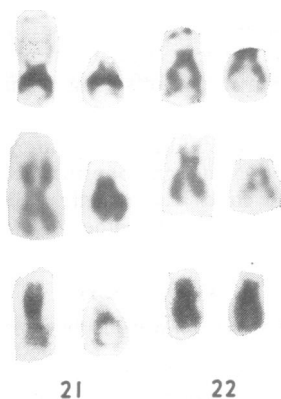


FIG. 2. Partial karyotype of the mother. From the top, first row with trypsin banding, second row with normal Giemsa stain, and the last row with C banding.

Discussion

It is well known that the short arms of the acrocentric chromosomes show great variations in their length. According to Lubs and Ruddle (1970) about 6% of the normal population has an obvious variant of this sort. However, the frequency of very long short arms of the G group chromosome is extremely low (0.04%).

Satellited short arms of the acrocentric chromosomes and the satellite association phenomenon have long been considered as a factor in the causation of nondisjunction and Robertsonian translocations (Ohno *et al*, 1961, Zellweger, Abbo, and Cuany, 1966; Vamos-Hurwitz *et al*, 1967). However recent studies done by banding techniques showed no difference in the satellite association frequency of no. 21 chromosome between the parents of trisomy 21 children and their controls (Cooke and Curtis, 1974; Taysi, 1975). Therefore, the relation between the most extreme variation in the short arms of no. 21 chromosome in the mother and the translocation type of trisomy 21 in the child, as in our case, could be the result of chance alone.

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