Case reports

This research was supported in part by the UCLA Mental Retardation/Child Psychiatry Program, and NIH grants MCH-927, HD-04612, HD-05615, and HD-06576.

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Variation in chromosome 19

SUMMARY Variations in centromeric staining of chromosome 19 appear to be an uncommon polymorphism inherited in a Mendelian manner and easily seen in G-banded cells. It should not be misinterpreted as a structural cytogenetic abnormality.

Although Craig-Holmes *et al.* (1973) were the first to draw attention to additional centromeric banding in the F group of chromosomes, it was Crossen (1975) who specifically implicated chromosome 19. However, there has been very little documentation of this variant. McKenzie and Lubs (1975) and Nakagome *et al.* (1977) paid scant attention to it and other reviews of chromosome polymorphism (Buckton *et al.*, 1976; Muller and Klinger, 1976) fail to mention it entirely.

We report variations in centromeric banding of chromosome 19 detected in the fetus during amniocentesis and also present in the mother.

Results and discussion

Amniocentesis was performed routinely because the mother would have been 35 years of age by the time the baby was born. Twenty-two cells representing a primary culture with 7 colonies and a secondary culture with an additional 7 colonies were banded by the trypsin-Giemsa technique. In all cells, one

Fig. One pair of chromosomes 19 from fetus (above) and mother (below) stained by the trypsin-Giemsa method. The variant is placed to the left in each pair. It is larger and submetacentric, with an increased amount of subcentromeric heterochromatin, and readily seen by G-banding.

chromosome 19 was submetacentric and longer than its homologue, because of enlargement of the band at q12 (Fig., top row).

In order to rule out a *de novo* alteration with its implications for the fetus, blood from the mother and father was examined, and the mother was found to have the same chromosome 19 variation (Fig., bottom row). Our conclusions were that this variation was inherited by the baby from the mother in a natural manner and, as the mother was healthy, there was no reason to infer that it would be the cause of any problems in the baby. The mother later delivered a normal baby boy.

Although easily seen in G-banded cells by the large submetacentric band, variation in chromosome 19 has been infrequently described. Nakagome *et al.* (1977), using the LBA technique on 4 patients, noted 'frequently observed' variation not obvious by quinacrine fluorescence. McKenzie and Lubs (1975) studied 77 patients and noted a '19C+' variant (their Fig. 8) with a very low frequency (1 to 2%). Muller and Klinger (1976), with 136 patients, and Buckton *et al.* (1976), with 708 patients, did not observe any alteration in chromosome 19.

It was Crossen (1975) who analysed these variations and enumerated 4 types depending upon the location of the heterochromatic area.

- Variant 1; confined to the centromere.
- Variant 2; extending into the short arms.
- Variant 3; extending into the long arms.
- Variant 4; extending into both short and long arms.

The variant chromosome in our family was type 3.

It seems, therefore, that this is not a common marker. Though this appears to be contradicted by Nakagome *et al.* (1977), they were dealing with a very small number of subjects and using a non-routine technique. The centromeric variation is easily seen in G-banded cells. It appears to segregate

in a Mendelian manner and therefore may be useful in family studies. It is presumably without phenotypic significance and thus should not be considered pathogenic.

The authors are grateful to Dr Paul Reid for referring the patient, to Dr Louise Knight for technical assistance, and to Mrs Iris Haas for secretarial help. This study was supported by Ontario Ministry of Health Grant PR 279.

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