Short report

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Trisomy 9p due to unusual maternal translocation (3;9)

Cytogenetic studies in a child referred for dysmorphic features, bilateral club foot, and mental retardation showed a chromosome complement of 47,XY,+der(9),t(3;9)(q29;q21·1) mat, the mother being the carrier of the balanced translocation: 46,XX,t(3;9)(q29;q21·1) (figure).

Autosomes of all groups can be involved in reciprocal translocations with chromosome 9. Most frequently, acrocentric chromosomes have been found to be involved, especially chromosomes 15 and 22. To our knowledge, tertiary trisomy resulting in partial trisomy 9p in a child of a balanced 3;9 translocation carrier mother has not been described.

The proband was born in 1979 after an uncomplicated term pregnancy and normal delivery. The birth weight was 3000 g and the length 50 cm. The parents were healthy, unrelated, and had negative family history. At his birth the father and the mother were 33 and 29 years old respectively. The mother had had a previous spontaneous abortion at 12 weeks but chromosome analysis was not performed.

The proband had all the typical features of trisomy 9p syndrome¹: microcephaly (head circumference 32 cm, 10th centile), hypertelorism, enophthalmos, anti-mongo-

loid slant, epicanthic folds, strabismus, globular prominent nose, simple low set ears, large downturned mouth, short neck, clinodactyly of the 5th finger, club feet, and general hypotonia. Results of routine laboratory studies, including full blood count, urine analysis, and EEG, were normal.

Chromosomes from PHA stimulated lymphocytes were studied in the proband and his parents with trypsin Giemsa (GTG) banding and cell synchronisation techniques. Heterochromatic polymorphism was evaluated with CBG banding. Cytogenetic investigations of the family of the mother showed no abnormalities. The grandmother was not available for examination because she was dead, so it was not possible to establish if the translocation in the mother was de novo or inherited.

The problem of translocations with 3:1 disjunction resulting in 47 or 45 chromosomes has been reviewed by many authors.² Factors influencing this type of disjunction are the nature of the chromosome involved and the origin of the translocation. Apparently 3:1 disjunction is more frequent when the translocation affects a chromosome 9 and when it is of maternal origin.³

In reviewing reports of trisomy 9p, the present case seems to be the first tertiary trisomy 9p derived from a translocation 3;9 mat. The biological reasons for the rarity of this translocation remain to be established.

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FIGURE Partial karyotype (RBA banded after cell synchronisation technique) of the mother (a) and the proband (b). Arrows indicate breakpoints.

References

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