Short reports

Journal of Medical Genetics, 1984, 21, 396-400

Pericentric inversion inv(3)(p11q21)

A familial inv(3)(p11q21) is reported, ascertained through a mildly dysmorphic non-recombinant infant.

The proband, a female, was born in April 1983 after an uncomplicated 38 week pregnancy and normal delivery. Her birth weight, height, and head circumference were 2650 g, 47 cm, and 33 cm respectively. Her face appeared slightly peculiar because of a prominent forehead and receding chin, a left preauricular tag, a long philtrum, and pendulous cheeks (fig 1). Premature closure of the anterior fontanelle, long fingers and toes, and shortening of the anovulvar distance were also noted. Radiological studies did not show other abnormalities. Routine laboratory and neonatal screening tests were normal.

The proband's mother and father were 25 and 27 years old respectively and both phenotypically normal. The mother had had two previous pregnancies resulting in phenotypically normal offspring and one spontaneous abortion (amenorrhoea for 3 months).

GTG, CBG, and QFQ banded preparations were obtained from 72 hour lymphocyte cultures according to standard techniques. The proband's karyotype was 46,XX,inv(3)(p11q21) (fig 2). Chromosome studies were extended to the proband's family and the same abnormality was found in her father, one of her two brothers, a paternal aunt, and her daughter, all of whom

FIG 2 Left to right: GTG banded chromosomes 3 from the proband's father, GTG banded inv(3) of the proband, and CBG and QFQ banded chromosomes 3 from the proband.

were phenotypically normal. Normal karyotypes were found in the other relatives studied. Reproductive histories were negative for malformed offspring.

Band 3q21 has been involved in other cases of familial inv(3),¹ and band 3p11 must be a not uncommon site of breakage and reunion if we consider the relatively high frequency of inversion of the proximal Q brilliant band in the long arm of chromosome 3 reported in some populations.²

In this familial inv(3), mild phenotypical abnormalities were present in the proband but not in the carrier relatives. A similar situation has been reported by Rivera *et al.*³ In both cases the karyotype-phenotype relationship, if any, remains obscure.

The absence of either proven or suspected liveborn recombinants in this kindred may be related to the relative shortness (approximately 18% of the total chromosome length) of the segment involved in the inversion.

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References

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- 3 Rivera H, Hernandez A, Rolon A, Ibarra B, Alcaraz A, Cantu JM. Familial pericentric inversion (3)(p14q24). *Ann Genet (Paris)* 1979;22:43-6.

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FIG 1 Lateral view of the proband's face at the age of 3 months. Note the left preauricular tag.