Case reports

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De novo tandem duplication 9p (p12→p24) with normal GALT activity in red cells

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SUMMARY A 3 month old boy with a tandem duplication 9p (p12→p24) is reported. Both clinical and dermatoglyphic features were consistent with those of the trisomy 9p syndrome. However, the red cell galactose-1-P uridyl transferase (GALT) activity was normal despite the presence of the duplicated segment 9p13.

Case report

The proband, a 3 month old boy, was referred for evaluation of many anomalies and developmental delay. He was the third child of unrelated, healthy parents. There was no family history of congenital malformations. The mother was 31 and the father 35 years old at the time of his birth. Delivery at 39 weeks' gestation was uneventful. Birthweight was 2705 g, length 48.0 cm, head circumference 32.5 cm, and chest circumference 30.0 cm. He was a slightly hypotonic infant of short stature.

At 3 months of age his weight was 4520 g (−3.0 SD), height 55.6 cm (−2.6 SD), head circumference 38.5 cm (−2.0 SD), and chest circumference 37.0 cm (fig 1). The salient clinical findings included brachycephaly, microcephaly, large anterior fontanelle, hypertelorism, asymmetry and antimongoloid slant of the palpebral fissures, prominent nasal bridge, bulbous nose, cleft lip and palate, downturned corners of the mouth, cup-shaped ears, small hands and feet with hypoplastic nails, and left cryptorchidism.

Dermatoglyphic features included incomplete simian creases, brachymesophalangy with fusion of flexion creases and clinodactyly of the fifth fingers, absence of digital triradii *c*, A mainline termination in 5, axial triradii in *t'* position, excess of arches on

FIG 1 The proband at 3 months.

fingertips (4/10), and decreased total finger ridge count (TFRC=24). He began to smile and to vocalise at 2 months.

CYTOGENETIC STUDIES

Cultured peripheral lymphocytes were used for chromosome analyses. G banding was done with ethidium bromide pretreatment two hours before harvest, according to the method described by Ikeuchi.¹ One hundred cells were examined and, of these, 10 GAG banded cells, eight RBG banded cells, and three CBG banded cells were photographed and karyotyped. All cells examined showed an elongation of the short arm of chromosome 9 which was evaluated as dup 9p (p12→p24) on G banded chromosomes at the 850 band stage, and as dup 9p (p12 or 13→p24) on R banded chromosomes at the same band stage (fig 2). The karyotype was interpreted as 46,XY,dir dup(9p) (pter→p12::p24→qter). The chromosomes of both parents were normal.

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⁶ Meera Khan P, Smith M. Report of the committee on the genetic constitution of chromosomes 7, 8, and 9. *Cytogenet Cell Genet* 1984;37:71-102.

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A complex balanced rearrangement involving four chromosomes in an azoospermic man

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SUMMARY A complex chromosomal rearrangement involving chromosomes 1, 5, 10, and 12 is described. The patient was an infertile, phenotypically normal male. Cytogenetic analyses of his parents showed that the complex translocation arose de novo. Testicular histology showed spermatogenic arrest.

Complex chromosomal rearrangements involving exchanges between three or more chromosomes are not frequent¹ and their association with male infertility is rare.² In the present paper we describe an azoospermic man with a karyotype showing balanced translocations between four chromosomes.

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Case report

A 33 year old azoospermic man was referred to our laboratory. He was born after an uneventful pregnancy and was the only child of the family. At the time of his birth his mother was 23 and his father 32 years old. There was no history of miscarriages. Apart from his infertility he was phenotypically normal.

TESTICULAR HISTOLOGY

The histology of the testes appeared normal. The tubules showed Sertoli and spermatogenic cells but complete absence of spermatozoa was noted. Leydig cells appeared to be well represented. According to the classification of Chandley *et al*³ the biopsy was classified as grade 2.

FIGURE G banded partial karyotype showing derivative chromosomes 1, 5, 10, and 12. Arrows point to the breakpoints on the ideograms corresponding to the chromosomes.