Partial trisomy 14q24→qter

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SUMMARY A newborn male with partial trisomy for the distal part of the long arm of chromosome 14 (14q24→qter) is described. The anomaly arose as an adjacent 1 meiotic segregation product from a balanced translocation t(11;14)(q25;q24) in the mother (figure). To our knowledge only one previous case involving the same segment has been reported. The karyotype was confirmed as 46,XY,der(11),t(11;14)(q25;q24) mat.

Case report

The mother has had six previous pregnancies, five of which resulted in spontaneous abortions, four between 12 and 20 weeks and one at 8 weeks' gestation. She has one normal child. No chromosome studies had been performed on herself or her

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FIGURE Ag/G and G banded partial karyotype of patient's mother showing balanced translocation 46,XX,t(11;14)(q25;q24). The proband has inherited the derivative 11q+.

husband or any of the aborted material. She is aged 26.

The proband's birth weight was 2480 g, length was 47 cm, and head circumference was 35 cm. He was born at term by dates, confirmed by Dubowitz assessment. His overall tone was poor with marked truncal hypotonia and an absent Moro reflex. There was no sucking reflex and absent asymmetrical tonic neck reflexes. Tendon reflexes were reduced. Pulses were palpable and of normal volume. Heart sounds were dual with a normally split second sound and no bruits audible. The respiratory system was clear. Abdominal examination revealed no hepatosplenomegaly or renal masses. There were bilateral inguinal hernias and bilateral cryptorchidism. The penis and scrotum were normal.

There was a prominent occiput with narrow bifrontal diameter, wide sutures, and a large anterior and posterior fontanelle. The ears, though not low set, were slightly angled and mildly dysplastic. He had micrognathia, carp-shaped mouth, and narrow palatal arch but no evidence of a cleft palate. The nose was broad. The hands were mainly clenched with overlapping of the index finger over the third and the fifth finger over the fourth. There was no nail hypoplasia. The hallux was normal but there was a prominent calcaneus bilaterally. There was no obvious thoracic abnormality. X-rays of skull, chest, and limbs were normal.

By day 7 he was beginning to feed from the breast. Hypotonia persisted though he subsequently developed a partial sluggish Moro reflex. Bilateral herniorrhaphy with orchidopexy was performed on day 11, at which operation both the hernial sacs contained the testes which were situated in the lower third of the inguinal canal. Both testes were of reasonable size and colour.

At one month his tone was basically unchanged though he had a better suck reflex. On this occasion there was a soft grade 1/4 ejection type systolic murmur maximal over the aortic area. The heart sounds, ECG, and chest x-ray were normal.

Discussion

Complete and partial trisomy 14 has been reviewed

by Johnson *et al*¹ and Wyandt *et al*.² Trisomy for the most distal part of 14q has been described only four times³⁻⁵ (Luzzatti, 1977, personal communication²). Of these cases that of Fryns *et al*³ is similar to our own both clinically and chromosomally, the only differences being the breakpoint at 14q23 and the parental translocation of paternal origin. The patient of Luzzatti (1977, personal communication²), on the other hand, has the same breakpoint as our case and although no great detail is given clinically, what is available compares favourably.

Until more reports appear it remains difficult to make specific karyotype-phenotype correlations.

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X chromosome replication patterns in a case of X;9 balanced translocation

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SUMMARY A case of X;9 balanced translocation in a female with amenorrhoea is reported. The X breakpoint was at Xq21, inside the 'critical region'. The normal X was consistently late replicating in blood lymphocytes and skin and ovary fibroblasts.

In somatic cells of mammalian females one X chromosome is randomly inactivated at the onset of embryonic development.¹ The inactivated X is late replicating and can be cytogenetically distinguished from the active one by means of ³H-thymidine or BUdR incorporation in late S period.^{2 3}

The analysis of balanced X;autosome translocations has revealed that in most cases the normal X is constantly late replicating.⁴ Hellkhul *et al*⁵ have recently reported in an X;3 balanced translocation that this general rule is restricted to blood lymphocytes; in skin fibroblasts the normal and the trans-

located X were alternatively inactivated. They have pointed out that in the majority of the reported cases only blood lymphocytes have been examined.

Our study concerns the X chromosome replication patterns in blood lymphocytes and skin and ovary fibroblasts in a case of X;9 balanced translocation.

Case report

The proband, aged 22, was referred because of secondary amenorrhoea. She was the third child of a family of five. Her mother was 28 and her father 33 when she was born and the delivery was uneventful. Both parents and the proband's brothers have normal karyotypes. At the age of 3 the proband underwent surgery for persistent ductus arteriosus. Menarche was at the age of 12, followed by three normal periods and then subsequently amenorrhoea. At the age of 18, she was treated with sequential oestrogestins and pseudo-menstruations ensued.

When seen by us the patient was 162 cm tall and was in apparent good health. Secondary sexual