An unbalanced t(X;10) maternal translocation in a child with congenital abnormalities

We report a case of a two year old female child (fig 1), the second daughter of unrelated parents (mother 23, father 30). Birth weight was 2080 g, length 44 cm, and head circumference 30 cm. Other abnormalities noted at birth and during subsequent development were hypotonia, dolichocephaly, wide fontanelle, large dysmorphic ears, small jaw, short sternum, widely spaced nipples, flexion deformities, shortened carpals, equinovarus deformity of feet, stunted, deformed toes, psychomotor retardation, and hearing deficit.

Partial duplication of both 10p and 10q are now well defined syndromes, the features of which have been reviewed by Yunis.¹ Most cases arise as the result of balanced parental rearrangements. X-autosome translocations are rare events with a frequency of 1 to 3 per 10 000 live births, as estimated by Mattei *et al.*²

GTG banded metaphases from lymphocyte cultures showed an unbalanced X;10 translocation in the proband. Family chromosome studies showed the balanced form of the translocation t(X;10)(q11;q25) in the mother and sister of the proband. The father and both maternal grandparents had normal chromosome patterns. The karyotype of the proband was interpreted as 46,X,-X,+der(10),t(X;10)(10pter→10q25::Xq11→Xqter)mat (fig 2a, b, c).

X inactivation studies were carried out by the addition of 0.2 mg/ml BrdU for the last six hours of lymphocyte culture. In the mother, the normal X was late replicating in each of 30 cells examined. In the proband, the translocated X chromosome was inactivated in all of 50 cells examined, the spread of inactivation extending along the 10q segment to the centromere. The spread of inactivation results in the effective deletion of the short arm of the X and the duplication of the short arm of 10.

A balanced t(X;10) with a random pattern of inactivation

FIG 1 The proband at two years of age.

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FIG 2 (a) GTG banding pattern in the mother. (b) GTG banding pattern in the proband showing the unbalanced rearrangement and the late replicating X chromosome. (c) Diagrammatic representation of the translocation.

tion has been described by Sands³ in a mentally retarded female. This contrasts with the more usual pattern of inactivation as found in the mother of the proband in our case where the normal X was inactivated in all cells examined.

Notable common features in our case compared with published cases of 10p duplication are low birth weight, psychomotor retardation, dolichocephaly, large dysmorphic ears, and equinovarus deformity of the feet.

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