

## Case reports

# Multiple abnormalities in a child with partial duplications of 10p and 13q from a 3:1 segregation of a maternal t(10;13) translocation

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### Abstract

**Partial duplications of 10p and 13q in association with partial deletions of other chromosome segments have been variously reported. We describe here a female child with multiple congenital abnormalities and combined partial duplications of 10p and 13q resulting from a 3:1 segregation of a maternal t(10;13)(p13;q22). In comparing the phenotypic features of the two chromosome imbalances, the expression of features typical of partial duplication 10p appeared more pronounced.**

The 'duplication 10p syndrome' generally involves most of the short arm of chromosome 10<sup>1</sup> while different segments of 13q have been variously implicated in the clinical expression of patients with partial or complete trisomy 13.<sup>2,3</sup> A female child with multiple congenital abnormalities and combined partial duplications of 13pter→q22 and 10p13→pter resulting from a 3:1 segregation of a maternal reciprocal translocation t(10;13)(p13;q22) is described.

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### Case report

The proband was the second female child of healthy, unrelated parents. At her birth, the mother was aged 34 and the father 49. There was no history of miscarriages.

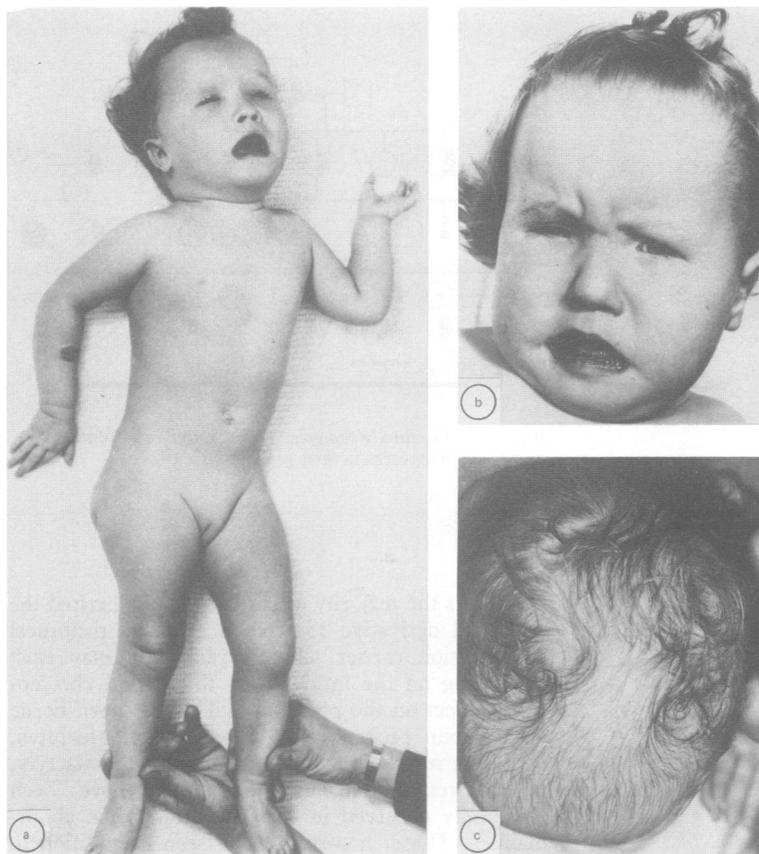
The proband was born at 30 weeks' gestation, weighing 1440 g (10th centile). Apgar scores were 1 at one minute and 8 at five minutes. Dysmorphic features and developmental delay were present from birth. These were reassessed at 21 months (gestational age 18 months) with the head circumference at 45 cm (10th centile), weight at 9.5 kg (3rd to 10th centile), and length at 79 cm (10th to 50th centile). She functioned at a 4 to 6 month level in all areas of development.

Dysmorphic features included plagiocephaly, an abnormal hair pattern with multiple whorls, and an upswept hairline (fig 1). She had a box shaped, prominent forehead and short palpebral fissures (palpebral fissure length 1.8 cm; mean at 18 months 2.3 cm). Dextrocardia, a gut malrotation, and a thoracic scoliosis were present. There was bilateral clinodactyly and a left simian crease. Several strawberry naevi in various stages of involution were present.

Coarse nystagmus was present although visual acuity was sufficient to enable her to fix and follow a 0.5 cm object. Optic fundi were normal. Audiological assessment showed normal hearing. Muscle tone and tendon reflexes were generally and symmetrically decreased.

### CYTOGENETIC STUDIES

GTG banding studies were done on peripheral blood cultures from the proband, her mother, and 6 year old sister. The father was unavailable for analysis. The mother and sister had a balanced reciprocal translocation involving the short arm of chromosome 10 and the long arm of chromosome 13. Their karyotypes were 46,XX,t(10;13)(p13;q22). The proband



*Figure 1 Clinical features of the proband. (a, b) Frontal view and facial dysmorphism of proband at 1 year 9 months. (c) Abnormal hair pattern with multiple whorls in the neonatal period.*

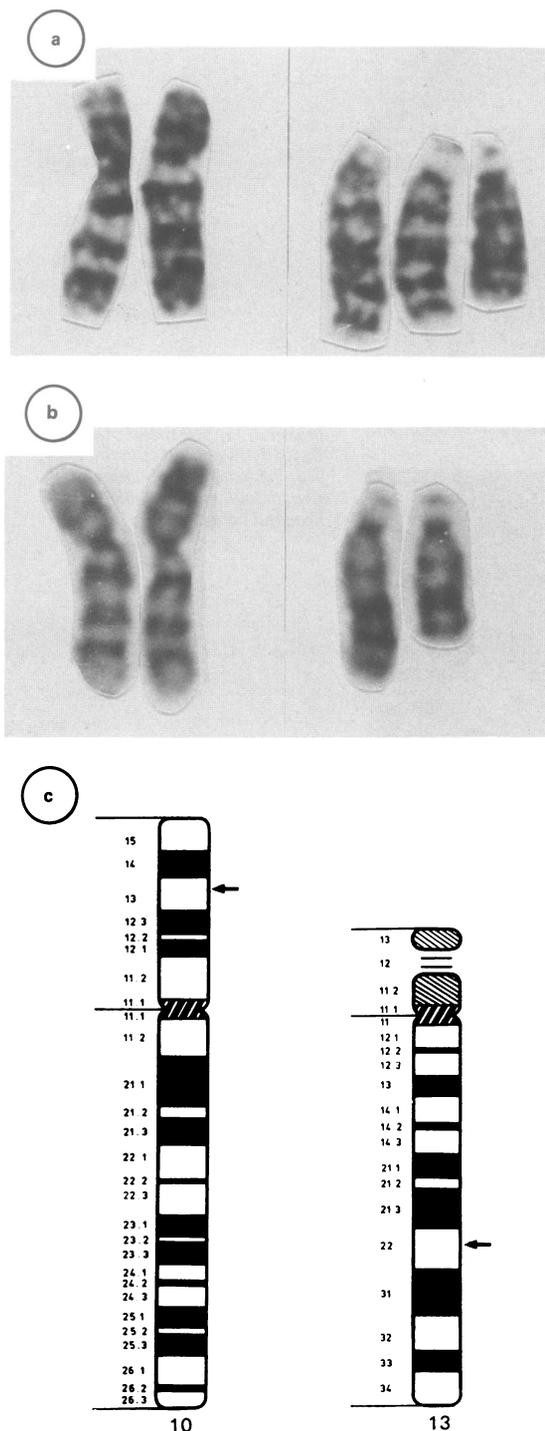
had 47 chromosomes with an extra derivative chromosome 13, two-thirds the size of a normal 13 and derived from a 3:1 maternal meiotic segregation. The proband's karyotype is interpreted as 47,XX,+der(13),t(10;13)(p13;q22), involving duplication for segments 13pter→q22 and 10p13→pter (fig 2). Extensive family studies in three generations showed seven of 12 maternal relatives tested to be balanced translocation carriers (fig 3). There were five pregnancy losses to known carriers, none of whom were available for cytogenetic analysis.

### Discussion

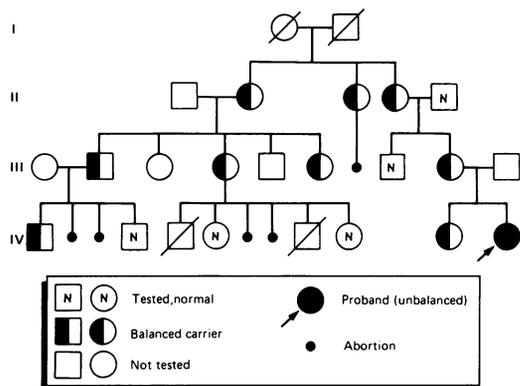
The female infant described, with a modal number of 47 chromosomes, is a tertiary trisomy derived from a 3:1 segregation of a balanced t(10;13) translocation present in the mother. The pachytene configuration from a maternal translocation is asymmetrical, involving two chromosomes markedly different in size, and including an acrocentric (chromosome 13), all of which would appear to predispose it to a 3:1 disjunction.<sup>4 5</sup>

Stene and Stengel-Rutkowski<sup>6</sup> characterised the range of breakpoints for chromosomes involved in reciprocal translocations, carrying risks for liveborn abnormal offspring with a tertiary trisomy. The breakpoints tend to be on the proximal long arm of a chromosome for which the short arm and proximal long arm segments are compatible with survival in the trisomic state. In our proband, for chromosome 13, the region involved includes the segments 13pter→q22. Compatibility with survival to term of such a large imbalance with more than half of chromosome 13 is not unusual, as trisomy 13 for the entire chromosome is seen in Patau's syndrome. The proband has a duplication of chromosome 13pter→q22 and exhibits few of the features associated with complete trisomy 13. Duplication for the distal segment 13q22→qter has been shown to be sufficient to produce such features.<sup>3 7</sup>

Phenotype-karyotype correlations in patients with partial duplication of various segments of chromosome 13 have been reviewed.<sup>2</sup> Features common to proximal duplication 13 up to band 13q22 include strabismus, depressed nasal bridge, and cleft lip and



**Figure 2** (a) Partial karyotype of proband with additional derivative chromosome 13. (b) Chromosomes 10 and 13 involved in the reciprocal translocation in the balanced carriers including the mother. The derivative chromosomes are on the right. (c) Diagrammatic illustration of the breakpoints.



**Figure 3** The family pedigree. The balanced reciprocal translocation was seen in three generations.

palate. As the majority of patients have inherited the abnormal derivative 13 from a balanced reciprocal translocation carrier, clinical variability may exist depending on the influence of the second chromosome defect on the phenotype. This has been borne out in our proband whose non-specific features, including microphthalmia, nystagmus, clinodactyly, mental retardation, and hypotonia, have been commonly observed in various chromosome abnormalities. These, however, when seen in addition to the box shaped, prominent forehead, abnormal hair whorl pattern and backswept hair, mid-thoracic scoliosis, and dextrocardia, do represent features of the readily recognisable duplication 10p syndrome, generally associated with most of the short arm of chromosome 10 from 10p11.<sup>1-8</sup> The breakpoint in the proband was more distal, at p13, involving a smaller segment from 10p13→pter, and the extent of dysmorphism and the severity of the growth and motor delay was less marked than in complete duplication 10p. Craniofacial changes have included plagiocephaly rather than dolichocephaly, short and not large palpebral fissures, with absence of mouth and palate deformities. Longer survival in partial distal duplication 10p has been noted.<sup>1</sup> Presumably, the severe organ malformations and skeletal anomalies often observed in serious cases of complete duplication 10p could involve the proximal short arm segments.

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