Trisomy 10p syndrome owing to maternal pericentric inversion

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Abstract

A female infant with karyotype 46,XX,rec(10),dup p inv(10)(p11.2q25.2)mat is presented. She had both duplication of 10p and deletion of distal 10q, but only had the constellation of specific features characteristic of duplication of 10p.

It is commonly thought that duplication of 10p results in a clinically recognisable chromosomal syndrome. Almost all such cases are inherited from translocation carriers. Only three cases of trisomy 10p syndrome owing to parental pericentric inversion have been reported.¹⁻³ It has been noted that patients with deletion of distal 10q have no pathognomonic features.

Figure 1 Clinical features of the proband at 2 months.

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Figure 2 Partial karyotypes of chromosome 10 of mother and proband. (a) Partial karyotype of the mother, 46,XX,inv(10)(p11.2q25.6). (b) Recombinant chromosome of the proband. This recombinant chromosome represents both duplication of 10p (p11.2→pter) and deletion of distal 10q (q25.2→qter).

Case report

The proband, a female, was born on 2.2.88 at 41 weeks' gestation to healthy and unrelated parents; the father was 28 and the mother 29 years old. The mother's first conception resulted in spontaneous

abortion, and the second in delivery of a healthy male infant.

The birth weight of the proband was 2322 g. She was referred to our outpatient clinic aged 11 days because of peculiar facies and thrombocytopenia. At that time length was 47.5 cm (-2.0 SD), weight 2315 g (-2.2 SD), and head circumference 32 cm (-0.7 SD). Physical findings included a high, protruding forehead, a small face in relation to the cranium, large horizontal palpebral fissures, sparse eyebrows, broad and protruding nasal bridge, round and sagging cheeks, thin upper lips, turtle beak mouth, micrognathia, low set, malformed auricles, narrow thorax, hyperflexed upper limbs, clinodactyly of the fifth fingers, clenched hands, narrow pelvis, abducted hip joint, and club foot (fig 1). Dermatoglyphic analysis showed bilateral palmar transverse creases, 10 whorls, and high axial palmar triradii. There was no cardiac or renal malformation. Although the platelet count on the sixth day after birth was $2.1 \times 10^4/\text{mm}^3$, it rose to $24 \times 10^4/\text{mm}^3$ on the 40th day. Cultured blood lymphocytes showed a karyotype of 46,XX,rec(10), dup p, inv(10)(p11.2q25.2) owing to a pericentric inversion of a maternal chromosome 10 (fig 2). Both her maternal grandmother and maternal aunt had this pericentric inversion. Her father and brother had normal karyotypes.

At 12 months DQ was about 25.

Discussion

The present case had both duplication of 10p (p11.2→pter) and deletion of distal 10q (q25.2→qter), but had specific features characteristic only of duplication of 10p. The table shows the extent of recombinant chromosomes resulting from pericentric inversion of a parental chromosome 10 and the clinical features of previously reported patients with this karyotype. None of the cases reported by de la Chapelle *et al.*,⁷ Husselein *et al.*,⁸ and Simpson *et al.*⁹ with this pericentric inversion has had a liveborn infant with this recombinant chromosomal aberration. Patients reported by Rodriguez *et al.*⁵ and Dutrillaux *et al.*⁶ had clinical features characteristic of trisomy 10p syndrome (table). In a case with almost the same breakpoints in chromosome 10 as those of our case, none of the characteristic clinical features noted in trisomy 10p syndrome was found, and Yunis and de Caballero⁴ doubted the existence of a trisomy 10p syndrome. In view of the fact that a case studied by Nomoto *et al.*² with duplication of 10p13→pter had phenotypic features characteristic of the trisomy 10p syndrome, it is not likely that the extent of duplication in the case of Yunis and de Caballero was too small to cause characteristic features of the trisomy 10p syndrome. Although the reason is still unclear, four cases¹⁻³ including our own had a constellation of features typically seen in the trisomy 10p syndrome,

Clinical features in cases with recombinant chromosome owing to pericentric inversion of chromosome 10.

	Yunis and de Caballero ⁴	Lansky-Shafer <i>et al.</i> ¹	Nomoto <i>et al.</i> ²	Sekhon <i>et al.</i> ³	Rodriguez <i>et al.</i> ⁵	Dutrillaux <i>et al.</i> ⁶	Present case
Extent of parental inversion	p11→q25	p11→q25	p13→q26	Not described	p15→q24	p15→q24	p11.2→q25.5
<i>Findings in trisomy 10p</i>							
Dolichocephaly	—	+	+	*	+	*	—
High, protruding forehead	+	+	+	*	+	—	+
Large, horizontal palpebral fissures	—	+	*	*	—	—	+
Low set, large, malformed ears	—	+	+	+	—	*	+
Broad, protruding nasal bridge	+	+	+	*	—	—	+
Round, sagging cheeks	—	*	+	*	*	*	+
Turtle beak mouth	—	+	+	*	*	*	+
Hyperflexed upper limbs	*	+	*	*	*	*	+
Abducted, flexed lower limbs	*	+	+	*	*	*	+
Club foot	*	+	—	+	*	*	+
Hypotonia	*	+	+	*	*	+	+
Excess of whorls	—	*	*	*	*	*	+
Cardiac malformation	*	*	—	+	—	—	—
Renal malformation	+	—	—	+	*	*	—
Ocular abnormalities	*	*	*	*	+	+	+
Hypoplastic genitalia	*	+	+	+	*	*	—
<i>Findings in trisomy 10q</i>							
Microcephaly	+	*	*	+	—	—	—
Round, wide, flat face	*	—	—	*	*	+	—
Narrow palpebral fissures	+	—	—	*	+	+	—
Microphthalmos	*	*	*	*	+	+	—
Hypoplastic nasal bridge	*	—	—	+	+	+	—
Ligamentary hyperlaxity	*	*	*	*	*	*	—
Clinodactyly	*	—	*	*	+	+	+
Phenotype	NC	Trisomy 10p	Trisomy 10p	Trisomy 10p	Trisomy 10q	Trisomy 10q	Trisomy 10p

*=not described or unclear.

NC=no characteristic features of trisomy 10p.

so it can be assumed that this syndrome might be found in many cases with this karyotype.

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