Partial 10q trisomy with partial 12q monosomy

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Abstract

A case of partial trisomy 10q with partial monosomy 12q is reported. The chromosomal abnormalities resulted from a paternal balanced, reciprocal translocation involving chromosomes 10 and 12, which, to the best of our knowledge, has not been previously described.

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Case report

A 1 day old girl was referred to the neonatal intensive care unit of our hospital because of respiratory distress and multiple congenital anomalies (fig 1 A and B). She was born after 38 weeks' gestation to healthy unrelated parents with a birth weight of 1900 g (below third centile) a head circumference of 30.4 cm (below third centile), and a length of 43.5 cm (below third centile). Her mother had taken no medication before or during pregnancy. On examination on admission she showed cyanosis with tachypnoea and dysmorphic features, that is, a flat and round face, a low frontal hairline, a short neck, hypertelorism, a depressed nasal bridge, short palpebral fissures, a small nose, low set ears, a bow shaped mouth, a high arched palate, and micrognathia. She clenched her hands with the second and fourth fingers overlapping the third one, and had hypertrophy of the clitoris. On auscultation, a systolic ejection murmur along the left sternal border was detected. The Moro reflex and palmar grasp were present. Ophthalmoscopic examination showed no abnormality. A computed tomogram showed mild brain atrophy without distinct anomaly. Ultrasound scanning of the abdomen revealed no internal anomaly and no tumour.

A chest radiograph taken on admission showed decreased pulmonary blood flow. A two dimensional echocardiographical study on the fourth day of life showed pulmonary stenosis, a ventricular septal defect, right ventricular hypertrophy, and an overriding aorta. The diagnosis was tetralogy of Fallot. A blood examination showed no abnormality except for a low serum sodium concentration of 136 mmol/l and a high serum potassium concentration of 6.2 mmol/l. There was no abnormality on hormonal examination, which included urinary 17-hydroxycorticosteroids and 17-ketosteroids, serum cortisol, 17-hydroxyprogesterone, progesterone, aldosterone, and dehydroepiandrosterone.

Figure 1 The patient at 3 months of age. Note the unusual facial features (A) and the hypertrophied clitoris (B).

G-banded chromosome studies on the patient, her parents, and 18 months old sister were performed with peripheral leucocyte cultures. Analysis of the father's cells showed a balanced translocation between the long arms of chromosomes 10 and 12: 46,XY,t(10;12)(q24.1;q24.33). The patient's karyotype was 46,XX,-12+der(12)t(10;12)(q24.1;q24.33) pat (fig 2). The mother's karyotype was normal.

She has been evaluated every two months. She is presently 1 year old, and physical examination showed a weight of 5640 g (below third centile), a head circumference of 40.0 cm (below third centile), and a length of 63.6 cm (below third centile). She has severe psychomotor retardation: she cannot hold her head up, roll over, say any words, turn to a voice, or laugh.

Discussion

The patient has partial trisomy of the long arm of chromosome 10 (q24.1 qter). Distal 10q trisomy is well delineated and called the 'distal 10q trisomy syndrome' due to the clinical differences from proximal or middle 10q trisomy.¹ The main clinical findings in distal 10q trisomy syndrome are mental retardation, hypotonia, a high forehead, a flat face, hypertelorism, arched eyebrows, short palpebral

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Figure 2 (A) G-banding of chromosome 12 of the patient and (B) chromosomes 10 and 12 of her father.

fissures, a small nose, a bow shaped mouth, a short neck, microcephaly, micrognathia, and congenital heart disease.^{1,2} Our case showed many common features with other published cases. The hormonal examination was performed because of the hypertrophy of the clito-

ris. There was no hormonal abnormality suggestive of congenital adrenal hyperplasia. The genital finding was interpreted as a malformation caused by the chromosomal abnormality. A hypertrophied clitoris, however, is not a common feature of the distal 10q trisomy syndrome. Only one case has been reported, which had other urogenital anomalies including the absence of the right kidney and ureter, and a common urethrovaginal outlet.³ The different findings in the distal 10q trisomy syndrome are thought to be due to the simultaneous presence of partial monosomy of another autosome.² To the best of our knowledge, however, there has been no case reported of the partial monosomy 12q. It remains unclear from which chromosomal abnormality the hypertrophied clitoris actually resulted.

The patient had tetralogy of Fallot. Although congenital heart disease is seen in most cases, tetralogy of Fallot is quite rare. Only two cases were previously described,⁴ one of which had an extreme form of tetralogy of Fallot. Both had simultaneous partial trisomy 10q and partial monosomy 17p, so tetralogy of Fallot may result from partial trisomy 10q.

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