Partial trisomy 16p due to maternal balanced translocation

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SUMMARY A newborn oriental male with multiple malformations was found to have partial trisomy of 16p. The mother was found to be a translocation carrier: 46,XX,t(14;16)(q32;p12).

While trisomy 16 is the most frequent trisomy detected in first trimester spontaneous abortions, it has not been found in surveys of perinatal and neonatal deaths or in surveys of newborn infants.^{1,2} Although trisomy 16 appears uniformly lethal early in pregnancy, fetuses with partial trisomy 16p or 16q occasionally survive to birth. Reports³⁻⁵ of liveborn infants with partial or complete trisomy of 16p or 16q material indicate that such infants have multiple congenital abnormalities and limited postnatal survival.

We report a newborn oriental male with multiple malformations who was found to be trisomic for 16p12→16pter. The mother was a carrier of a 14;16 apparently balanced translocation.

Case report

The proband, a newborn oriental male, was the 2350 g product of an unremarkable pregnancy. He was born to a 31 year old gravida 2, para 2 mother and a 28 year old father. The marriage was consanguineous, the maternal and paternal grandmothers being sisters. Premature rupture of the membranes occurred at 36 weeks' gestation, 38 hours before delivery. Physical examination showed an infant with a head circumference of 32 cm; a length of 47.5 cm; a floppy protuberant right ear with minimal helix formation (fig 1); depressed nasal bridge; high arched palate; micrognathia; hypoplastic mandible; flexed fingers; toe 3 on left foot dorsiflexed under toe 2; large right hernia (fig 2); and bilateral palpable testes. The infant had acrocyanosis and was feeble. A grade 2/6 systolic murmur was not present at birth but developed over the first few weeks of life. This was found to be due to an atrial septal defect. Neurological examination revealed a hypotonic infant who had a weak cry and

a poorly coordinated suck and swallow requiring nasogastric feeding. When seen again at 9 months of age the infant had developed the ability to nipple feed. However, he had no head control and was showing severe developmental delay. He had a seizure activity and burst suppression pattern in sleep which were consistent with hypsarrhythmia. A CT scan showed a probable cyst of the septum pellucidum and carum vergae.

FIG 1 Asymmetry of the ears. The right ear is floppy with minimal helix formation; the left ear is normal.

FIG 2 Lower torso showing large inguinal hernia and dorsiflexion of toe 3 under toe 2 on the left foot.

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FIG 3 Partial karyotypes showing the partial trisomy 16p in the proband and the balanced translocation in the mother.

CYTOGENETIC STUDIES

Metaphase cell preparations were obtained from 72 hour cultures of phytohaemagglutinin stimulated lymphocytes. Prophase cells were obtained using the technique of Yunis.⁶ Cells were analysed primarily by GTG banding. Initial analysis of the proband's karyotype revealed 46,XY,14q+ (fig 3a). The father and brother had normal karyotypes. The mother was carrying a translocation 46,XX,t(14;16)(q32;p12) (fig 3b). Thus, the proband's karyotype would be interpreted as 46,XY,-14,+der(14),t(14;16)(q32;p12→pter). Thus, he is trisomic for 16p12→pter.

Discussion

In 1978 Roberts and Duckett³ reported a case of trisomy 16p and reviewed published reports on partial and full trisomy 16. Yunis *et al*⁵ reported a liveborn infant with partial trisomy of the short arm and part of the long arm of chromosome 16. Leschot *et al*⁷ reported a familial 16;21 translocation which resulted in five cases of partial trisomy 16p. Additional cases of partial trisomy 16

due to familial 16;21 translocations have been recently reported.⁸ While most of these familial translocations involved chromosome 21, other chromosomes (18 and 12) have also been reported.^{9,10} In our patient, the trisomy 16p resulted from maternal translocation between chromosomes 14 and 16. All of the patients with partial trisomy 16p had multiple congenital anomalies and, with one exception, died within the first year of life.¹⁰ The most common anomalies were hypoplastic mandible, low set ears, asymmetry of ears, cardiac defect, clinodactyly, dorsiflexion of toes, and generalised hypotonia.

In summary, while full trisomy 16 is apparently incompatible with life, partial trisomy 16 can result in a liveborn infant who would be expected to have multiple congenital anomalies, severe developmental retardation, and limited postnatal survival.

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