Case reports

Duplication of distal 17q from a maternal translocation: an additional case with some unique features

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SUMMARY A female with multiple dysmorphic features was found to have an unbalanced karyotype with duplication of the distal long arm of chromosome 17 and deletion of the terminal region of the short arm of chromosome 12. This was derived from a reciprocal translocation in the mother, 46,XX,t(12;17)(p13.3;q23).

Clinical findings are presented and comparison with other reported cases of distal 17q duplication shows several unique features in our case.

Duplication of 17q is very rare with only a dozen reports published. Many of the dysmorphic features in the cases reported are similar and it is now becoming a recognisable syndrome. One de novo case has been interpreted as a tandem duplication of 17q25→qter based on the previously reported features of such duplications. Most cases, however, are inherited and are the result of segregation from a balanced translocation in one of the parents. The deletion of material from the other chromosome involved in the translocation appears to have a minimal clinical effect.

Case report

The proband was the first child of unrelated parents, the mother being 25 years of age. The pregnancy was normal apart from the mother taking lorazepam and diethylpropion hydrochloride throughout the pregnancy. Ultrasound scans carried out at 17, 21, and 25 weeks by dates indicated gestational ages of 12, 17, and 21 weeks, respectively. Dating at the last scan was assessed by femur length and abdominal circumference since biparietal diameter measurements were not possible owing to the fetal position.

The mother was induced at 38 weeks because of high blood pressure and a large increase in weight. She was given a trial of labour because of her short stature, but fetal distress ensued resulting in an emergency caesarian section.

After a difficult delivery by cephalic presentation, a markedly hydropic female baby was born, birth weight 3900 g. There was no respiratory effort and intubation was difficult owing to massive oedema (fig 1). Bilateral thoracentesis was performed and the infant was transferred to a ventilator and an exchange transfusion was performed. An in-
abdominal catheter was placed to drain ascitic fluid and repeated transfusions of albumin and blood were given for hypoalbuminaemia and anaemia. Dysmorphic features included low set ears, hypertelorism, widely spaced nipples, shield chest, and short neck. In addition, a heart murmur was noted which echocardiography showed to be the result of a patent ductus arteriosus. X rays showed increased bone density with calcified deposits in the sternum and gall bladder, the latter probably resulting from gall stones (fig 1). Abdominal ultrasound showed a bright echogenic pattern in the kidneys.

Chromosome analysis was requested shortly after birth owing to the grossly hydropic appearance and the dysmorphic features indicated.

The oedema lessened concurrent with a reduction in the infant's weight to 1.9 kg. There was a gradual improvement but survival was not possible without continued ventilatory support and she died aged two weeks. Permission for a necropsy was declined.

Haemoglobin electrophoresis was normal, no blood group incompatibility was detected, and both TORCH titres and viral cultures were negative.

**CYTOGENETIC FINDINGS**

Peripheral blood lymphocytes were cultured from the proband for chromosome analysis using GTG banding. The analysis showed an unbalanced karyotype with additional material on the short arm of chromosome 12. Only the mother was available for follow up studies and she was shown to have a balanced reciprocal translocation between chromosomes 12 and 17, 46,XX,t(12;17)(p13.3;q23). The proband's karyotype was therefore 46,XX,-12,+der (12)t(12;17)mat with a duplication of 17q23--qter and a terminal deletion of 12p13.3--pter (fig 2).

**Discussion**

Abnormalities of chromosome 17 appear to be fairly uncommon, with duplication of 17q being extremely rare. Most of the published reports of distal 17q duplication are the result of segregation from a parental balanced reciprocal translocation, but a few occur as de novo events. A distinct clinical picture is beginning to emerge which can include mental retardation, severe growth retardation, psychomo-
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Moebius' syndrome with unilateral cerebellar hypoplasia

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SUMMARY A case is reported of a child with Moebius' syndrome who also has unilateral cerebellar hypoplasia. We suggest that this combination of abnormalities could result from a vascular disruption occurring in the basilar artery early in its development.

Moebius' syndrome consists of congenital unilateral or bilateral facial weakness and loss of abduction of the eye, which are interpreted clinically as defects of the seventh and sixth cranial nerves. The involvement of other cranial nerves or abnormalities of the extremities, such as syndactyly, agenesis of digits, or defective branchial musculature, are common, but it is unusual to find gross structural brain abnormalities.

We report a patient with Moebius' syndrome who also has unilateral cerebellar hypoplasia.

Case report

A male infant was born at term by a normal vaginal delivery and his birth weight was 3250 g. An amniocentesis had been performed during the pregnancy owing to advanced maternal age, but the pregnancy was otherwise uneventful. In the neonatal period he was noticed to have a right convergent squint and right sided facial weakness, although he breast fed without difficulty.

He smiled at 10 weeks and from five months he was able to roll over from the prone to the supine position. He could pass objects from one hand to the other by six months, although his mother noticed that left hand preference had already become established and there were occasional jerky movements of his right arm.

When reviewed at seven months there had been no change in his condition. His head circumference (44.3 cm), weight (7.8 kg), and length (69 cm) were