Partial trisomy 12q

SUMMARY A partial trisomy 12q243→qter resulting from a maternal balanced translocation, 46,XX,t(9;12)(p243;q243), was detected in a male newborn with multiple congenital abnormalities. The maternal grandmother was also a carrier of the 9;12 translocation. Our patient exhibited a number of clinical features similar to two others reported, who were also trisomic for the distal part of 12q.

Aberrations of chromosome 12 are very rare. There have been only two reports of partial trisomy 12q, both the result of a familial translocation.1,2 We describe a third unbalanced case.

Case report

The proband was the second child of a 30-year-old mother and a 32-year-old father. Both the non-consanguineous parents and a 2½-year-old sister were phenotypically normal. The first pregnancies of the mother and the maternal grandmother ended in spontaneous abortion during the first trimester (fig 1). After a 41-week pregnancy, complicated by vaginal bleeding during the third month, delivery was by caesarian section. Birthweight was 3060 g, length 47 cm, and head circumference 34.5 cm. Histological examination of the placenta showed no pathological findings.

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![Pedigree of the family.](image)

![Proband at the age of 2 days.](image)
Clinical examination revealed brachycephaly with flattened occiput and peculiar facial features (fig 2a), including a small broad-bridged nose with a down-turned tip, hypertelorism, wide mouth with downturned corners and thin lips, slight micrognathia, high arched palate, and low set malformed ears. Further multiple clinical abnormalities were present (fig 2b): elastic skin with loose folds in the posterior and anterior regions of the neck and the left axilla; short, thick neck with low hairline; broad chest and widely spaced nipples; short proximal arms; flexion and ulnar deviation of both hands, palmar simian creases, and relatively long fingers; subluxation of both hips and deep coccygeal dimple; absence of the right patella; bilateral cryptorchidism and small penis.

Computerised tomography of the head showed mild hydrocephalus externus and internus, but the electroencephalogram was normal. A systolic murmur along the left sternal border, biventricular hypertrophy on the electrocardiogram, and abnormal configuration of the heart on radiographic examination suggested a congenital heart defect. Barium study of the gastrointestinal tract showed cardiochlasia and malrotation of the duodenum. Lumbar dystopia of the right kidney and malrotation of the left kidney were disclosed by intravenous pyelography. Skeletal x-rays showed only a dysplastic thorax and shortened ribs.

The clinical course was complicated by feeding difficulties and frequent vomiting, owing to a weak sucking reflex and cardiochlasia, respectively. Although feeding by gastric tube was performed, the baby did not gain weight. Recurrent bacterial urinary tract infections were noted. Muscular hypotonia developed during the first weeks of life. Physical and psychomotor development were retarded.

At the age of 2½ months the child died from fulminant viral hepatitis type B, which was proved histologically by percutaneous liver biopsy. A necropsy was refused.

**Cytogenetic Studies**

Chromosome analysis after G banding in the proband revealed extra genetic material on the short arm of chromosome 9 (46,XY,9p+).

The karyotype of the mother showed a balanced translocation between chromosomes 9 and 12: 46,XX,t(9;12)(p243;q243). The distal part of the long arm of chromosome 12 (q243→qter) had been translocated onto the terminal band of the short arm of chromosome 9 (fig 3a).

The proband inherited the 9p+, resulting in partial trisomy of the long arm of chromosome 12: 46,XY,—9,+der(9), t(9;12)(p243;q243)mat (fig 3c).

The maternal grandmother proved also to be a carrier of the balanced 9;12 translocation (fig 3b). The father, maternal grandfather, and sister of the proband were all found to have normal karyotypes (fig 1).

**Discussion**

Until now, structural rearrangements involving chromosome 12 have rarely been observed. A possible explanation for this fact could be that larger
partial monosomies and trisomies are not compatible with intrauterine and postnatal survival and aberrations with more distal breakpoints were difficult to detect without the new banding techniques. Partial trisomy 12q has been described in only two cases, both resulting from different familial translocations.\(^1\)\(^2\) In both children the breakpoints of chromosome 12 were shown to be at band q24 resulting in a partial trisomy 12q24→qter. The banding pattern in our patient allowed a clear determination of the breakpoint at band 12q243 (fig 3c). It appears that the breaks occurred more distally to band 12q243 in the case reported by Hobolth et al\(^2\) and more proximally in the case of Hemming and Brown.\(^1\)

Clinical features common to the three patients are an unusual craniofacial dysmorphism including brachycephaly, flattened occiput, and a characteristic facial appearance, comprising a small and broad-bridged nose with a downturned tip, hypertelorism, wide mouth with downturned corners and thin lips, low set malformed ears, short thick neck with loose posterior skin folds, short proximal arms, palmar simian creases, and cryptorchidism. The patient of Hemming and Brown\(^1\) and our case presented with a pelvic kidney and malrotation of the intestine. In addition, congenital heart defect was assumed in the child observed by Hobolth et al\(^2\) and in our case. These two patients showed signs of mental retardation, while the case of Hemming and Brown\(^1\) died a few days after birth. The similarities, especially in the patient of Hemming and Brown\(^1\) and in our case, may suggest a new clinically distinguishable cytogenetic syndrome, but further observations are necessary for the exact delineation of this chromosomal aberration.

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References


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A case of trisomy of chromosome 15

Summary We describe a case of trisomy of chromosome 15 in an infant who presented at birth with numerous abnormalities. As far as we are aware this chromosomal abnormality has not been described before. On the basis of this one case there appear to be no features which are specific to this chromosomal abnormality.

We describe a female infant with trisomy of chromosome 15. As far as we can ascertain, no other patients with this chromosomal abnormality have been reported, although details of a number of patients with partial trisomy have been published.\(^1\)\(^2\)\(^3\)

Case report

The infant was born in May 1979, the father being 32 years old and the mother 31. There had been three previous pregnancies. The first and third were complicated by hypertension but resulted in normal term deliveries of live female infants (birthweights 3.5 kg and 3.8 kg). The second pregnancy resulted in a miscarriage at 26 weeks' gestation. No details of the fetus are available.

During the first trimester the mother had taken an antihistamine (for hay fever) and an antiemetic. She had taken an iron and folic acid preparation throughout the pregnancy. The clinical impression of growth retardation was confirmed by ultrasound scans. The pregnancy went to term and delivery was normal. The baby had an Apgar score of 6 at 1 minute and 7 at 5 minutes. The birthweight was 2.0 kg, the length 47.5 cm, and the head circumference 31 cm.

The infant was hypotonic and there were numerous anatomical abnormalities (fig 1). The face showed small palpebral fissures, wide epicanthic folds, a broad flat nasal bridge, a small mouth, and micrognathia. There was a large anterior fontanelle and the posterior fontanelle was open. The ears were small, low set, and convoluted. The neck was webbed and the hairline low. The chest shape was abnormal with a short sternum and widely spaced nipples. There was webbing and overriding of the fingers and toes, though the palmar skin creases were normal. The external genitalia were normal, but the anus was displaced anteriorly. Abnormalities of the joints included bilateral talipes equinovarus, bilateral dislocation of the hips, and dislocation of the left wrist. The femoral pulses were weak, the