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 al2 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-nosed 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 Brown1 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 al2 and in our case. These two patients showed signs of mental retardation, while the case of Hemming and Brown1 died a few days after birth. The similarities, especially in the patient of Hemming and Brown1 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.1–3

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 epicantal 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
heart was enlarged, and a loud systolic murmur was audible over the precordium. There was no visceromegaly.

The infant became increasingly cyanotic and tachypnoeic and died on the fourth day from intractable heart failure.

INVESTIGATIONS

Chest radiography confirmed cardiomegaly. On skeletal survey only eleven pairs of ribs were present and there was an associated rib deformity in the left axillary margin. The right clavicle was small and hypoplastic. Both acetabular angles were abnormally steep with dislocation of both hips. There was incomplete bony fusion of almost all the spinal neural arches in the cervicodorsal region.

At necropsy the lungs were found to be small and hypoplastic. The heart had a very small left atrium, and the right atrium and both ventricles were moderately dilated. The foramen ovale was valvar, and the ductus arteriosus was widely patent. There was an oval defect of the middle third of the ventricular septum and there was coarctation of the aorta between the left subclavian orifice and ductus.

Chromosome studies were performed on peripheral blood lymphocytes using standard techniques. Giemsa trypsin banding was carried out using a modification of the method of Seabright. One hundred metaphases were counted and five banded metaphases were karyotyped. The chromosome count in all cells examined was 47,XX,(15+)(fig 2).

Both parents had normal chromosome counts in their peripheral blood lymphocytes.

This infant had trisomy of chromosome 15. She showed some of the features previously described in children with partial trisomy 15 including hypotonia, microcephaly with frontal prominence, broad depressed nose, low set ears, and low hairline. Clinodactyly of the fifth digit and syndactyly of the second and third digits were also present. However, she also had cardiovascular and skeletal abnormalities, including a ventricular septal defect, coarctation of the aorta, bilateral talipes, and congenital dislocation of the hips. None of these features has been described in association with partial trisomy 15. On the basis of our findings in this child trisomy 15 does not appear to have particular features which make an immediate clinical diagnosis possible at birth.

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Congenital dislocation of the knees in a child with Down-mosaic Turner syndrome

SUMMARY A further case of Down-mosaic Turner syndrome is discussed. Both the cytogenetic and the dermatoglyphic data support the clinical diagnosis. The association with dislocated knees and the diagnosis of this polysyndrome at birth have not been reported before.

Approximately one in 200 liveborn children have a chromosomal abnormality. It appears that aneuploidy and structural chromosomal anomalies are among the most common causes of abnormal fetal development. Klinefelter syndrome with Down syndrome is the most frequent double aneuploidy recognised.

In this paper a case of Down-mosaic Turner syndrome associated with congenitally dislocated knees is presented. No previous report of a similar combination could be traced.

Case report

After an uneventful pregnancy, the proband, a Caucasian female, was born weighing 3 kg and measuring 50 cm in length. The father was 28 years old and the mother 26 years at the time of the child’s birth. The proband had mongoloid features (fig 1a), bilaterally dislocated knees (fig 1b), webbed neck, a transverse palmar crease, low hairline, oedema of both feet, hypoplastic nipples, and cubitus valgus. X-rays confirmed bilateral dislocation of her knees. She is now 5 years old and has both mental and physical retardation. She is the only child and her mother has not had any miscarriages.

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CYTOGENETIC INVESTIGATIONS

A buccal smear showed a single Barr body in 14% of the cells examined. Chromosomes from lymphocyte cultures taken from the proband and both parents were examined in standard and trypsin-Giemsa banded preparations. The proband had 46 chromosomes in 37 of the 50 cells analysed. In each of these cells there was an extra chromosome 21.

FIG 1a Proband at the age of 5 years.

FIG 1b Congenitally dislocated knees (only one shown).