several investigators. The modal chromosomal number reported in all cases has been 48. This is the first case in which 49 chromosomes were found in a patient with double aneuploidy. The phenotypic characteristics of this patient most closely resemble those of trisomy 18. This observation is consistent with those described in other cases of double aneuploidy involving chromosome 18 and sex chromosomes. One explanation for this is that there are no specific phenotypic findings in the sex chromosome syndromes in the newborn period.

It is of interest to note the presence of unilateral microtia in our patient. This is not a characteristic finding in trisomy 18. Unilateral malformed ears have been reported in other cases of double aneuploidy with karyotypes of 48,XXY,+13 and 48,XXY,+18. This might be a reflection of the major chromosomal imbalance present since it is not syndrome specific.

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References


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Case reports

Proximal femoral focal deficiency associated with the Robin anomalad

SUMMARY A case of unilateral proximal femoral focal deficiency (PFFD) and the Robin anomalad is reported. Since bilateral PFFD and unusual facies have been reported before, we suggest an association between the Robin anomalad and PFFD.

The purpose of this report is to describe a patient with unilateral proximal femoral focal deficiency (PFFD) and coexisting Robin anomalad. Daentl and associates reported the occurrence of PFFD with unusual facies. In each of their cases the femoral defects were bilateral. The unusual facies described by Daentl, including micrognathia and cleft palate, would seem to be representative of the Robin anomalad.

Case report

A male term infant was born to a 22-year-old gravida 1 para 1 aborta 0 female after an uncomplicated gestation. The patient was delivered by caesarian section because of breech presentation. The Apgar scores at 1 and 5 minutes were 8 and 8,

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FIG 1 Profile of proband showing the pronounced mandibular hypoplasia that is seen with the Robin anomalad.
respectively. The infant weighed 2540 g and was not in acute distress. Family history was unremarkable. There was no juvenile or gestational diabetes in the mother. Physical examination showed slanting palpebral fissures, cleft soft palate, hypoplastic mandible (fig 1), floppy tongue, thin upper lip, slightly elongated philtrum, hypoplastic appearing pelvis with flexion contracture of the right hip, and talipes equinovarus. Neurological examination disclosed poor grasp, suckling, and Moro reflexes. The heart rate and rhythm were normal without a murmur initially; however, by 3 weeks of age a grade 2/6 systolic ejection murmur was heard over the pulmonic area. Further investigation showed a ventriculoseptal defect with pulmonary stenosis. There was no evidence of ophthalmological abnormalities.

Pelvic x-rays showed a hypoplastic right femur with the proximal end lateral and superior to the acetabular area (fig 2). The left femur was normal except for mild lateral subluxation of the hip. The chest x-ray was normal and the skull x-rays showed a small mandible.

Laboratory data was non-contributory. Chromosomal analysis with trypsin-Leishman banding revealed a 46,XY chromosome complement. The patient did well after the fitting of a cleft palate prosthesis and was discharged to be followed up in an orthopaedic clinic for management of the PFFD and talipes equinovarus.

Discussion

Proximal femoral focal deficiency is a rare congenital skeletal anomaly which may be associated with other osseous defects. The deficiency involves the proximal segment of the femur; however, a portion of the femur is always present, serving to differentiate this entity from complete femoral agenesis. Although the aetiology is unknown, Levinson et al list two possible mechanisms for the defect. One is localised cellular nutritional disturbance and the other is local vascular change. No chromosomal abnormalities have been reported with PFFD, but Kelly reported a familial case.

The Robin anomalad was originally described as micrognathia and glossoptosis, but cleft palate is often included because of its common association with the anomalad. The anomalad has also been associated with ocular disorders, bony abnormalities, talipes equinovarus, and congenital heart disease, all of which were present in this case. Daentl reported unusual facies occurring with PFFD. These were described as slanting palpebral fissures, elongated philtrum, thin upper lip, micrognathia, cleft palate, club foot, and other osseous abnormalities of the pelvis and spine. These findings may occur in the Robin anomalad.

The patient was different from all previous reported cases in that the femoral defect was unilateral. It would seem that the association of PFFD with the Pierre Robin anomalad is more than coincidence. When confronted with a patient who has PFFD, a careful physical examination should be performed to exclude the associated Robin anomalad.

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The penta-X syndrome

SUMMARY A child is presented with a 49,XXXXX chromosomal constitution bringing to 12 the total number of children described with this karyotype. Comparison of this child’s features with previously reported cases indicates a clinically recognisable specific pattern of malformations referred to as the penta-X syndrome. X chromosome replication studies using BrdU labelling in the patient’s cells clearly showed that the four presumably inactive X chromosomes were late replicating but not in a strictly synchronous fashion.

Since 1963, 11 children with a 49,XXXXX karyotype have been reported.\textsuperscript{1-11} The purpose of this publication is to describe an additional patient with a 49,XXXXX chromosomal constitution and to report the results of X chromosome replication studies using BrdU labelling in her cells. Comparison of the clinical features of this child with previously reported cases suggests that a 49,XXXXX chromosomal constitution is associated with a specific recognisable pattern of malformations, which has previously been referred to as the penta-X syndrome.

Case report

The patient, a Mexican American female, was born to a 19-year-old gravida 2, para 1 mother and a 20-year-old father. Fetal activity, which started at 4 months, was decreased throughout the remainder of the pregnancy. Delivery after 38 weeks’ gestation was uncomplicated. Birthweight was 1785 g and birth length was 42.5 cm. After being in stable condition initially, the infant suffered a dusky spell and at one week of age she developed circumoral cyanosis while feeding. Physical examination at 10 days of age showed a weight of 1800 g, length of 40 cm, and head circumference of 30 cm. The metopic suture was widely separated and the posterior fontanelle measured 3.5 x 2 cm. The hairline was low anteriorly. The inner canthal distance measured 2.2 cm (75th centile). There were external ear malformations consisting of appendages protruding from each tragus (fig 1), prominence of the antitragus bilaterally, and a preauricular pit on the left. There was incomplete development of the eyes manifest by persistence of the primary pupillary membrane and of the hyoid arteries bilaterally. Limb anomalies included decreased supination and pronation of the arms because of radioulnar synostosis, bilateral fifth finger clinodactyly, small hands which measured 5 cm in total length, and overlapping of toes. The digital dermatoglyphic patterns showed radial loops on both thumbs, one arch and three ulnar loops on the fingers of the right hand, and four arches on the left. She had a grade 3/6 systolic murmur at the left sternal border and bilateral rales at the lung bases.

FIG 1 The patient at 1 day of age. Note the low hair line anteriorly, the ocular hypertelorism, and the appendage protruding from the tragus of the right ear and the prominent antitragus.
Proximal femoral focal deficiency associated with the Robin anomaly.

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