Dysmorphology reports

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Unknown syndrome: congenital heart disease, ptosis, hypodontia, and craniosynostosis

SUMMARY We report a child with total anomalous pulmonary venous drainage, ptosis, hypoplastic teeth, sagittal craniosynostosis, and developmental delay, together with several unusual features.

Case report

The proband, a two year old male, was the first child born to unrelated parents. The father was 36 and mother 33 years at the time of birth. A maternal cousin was born with tricuspid atresia, but otherwise the family history was negative.

There was no history of infections or medication during pregnancy. He was born by caesarean section 18 days after term with a birth weight of 2820 g and a head circumference of 34 cm. He developed respiratory distress and had a successful repair of total anomalous pulmonary venous drainage at six days of age. He has subsequently shown some delay in psychomotor development. He sat up at 10 months and has begun to walk at 18 months.

He was of slender build with height on the 50th centile and weight below the 3rd centile. His head circumference was below the 3rd centile. There was ridging of the cranial sutures and an absent anterior fontanelle. Bilateral ptosis was present with more marked narrowing of the right palpebral fissure (figure). The ears were low set and prominent. There was a long philtrum and prominent lower lip and mandible. The teeth were small, conical, and widely spaced. The structure and growth of the hair and nails were normal. There was generalised joint laxity and long fingers and toes. No joint contractures were present.

Full chromosome studies were normal. Routine haematology and biochemistry were also normal. The skull X ray showed premature fusion of the sagittal suture.

Discussion

The combination of blepharophimosis and congenital heart disease has been reported in the Marden Walker syndrome and in a report of two sisters by Ohdo et al. In the Marden Walker syndrome joint contractures are a prominent feature and were certainly not present in this case. In the report by Ohdo et al both sisters had a combination of congenital heart disease, blepharophimosis, ptosis, hypoplastic teeth, and severe mental retardation, but craniosynostosis and the other facial features we have noted were not present.

The association between craniosynostosis and total anomalous pulmonary venous drainage is also unusual. The syndrome described by Gorlin et al combines many of the features seen in this case, for example, craniosynostosis, oligodontia, congenital heart disease, and mental retardation, but in our opinion is probably a different disorder. The two girls in the paper of Gorlin et al were of normal intelligence and showed additional features such as labial hypoplasia, hypertrichosis, and microphthalmia. We have used the London Dysmorphology Database and not been able to place the clinical features of this case with a recognised syndrome. It would appear that this child has a new cardiofacial syndrome.

FIGURE Frontal and lateral photographs illustrating the facial appearance. Note the presence of ptosis, low set ears, and prognathism.
Unknown syndrome in sibs: microcephaly, seizures, mental retardation, congenital heart disease, and skeletal abnormalities

SUMMARY We present two male sibs with a series of malformations including microcephaly, mental retardation, congenital heart disease, skeletal abnormalities, microgenesis, and mild hypothyroidism. Both have had seizures. While the pattern of abnormalities is similar to that previously reported in this journal as an unknown syndrome, the facies is clearly distinct, the hypothyroidism is mild, microgenesis is present, and there are additional minor skeletal abnormalities.

History


Family. No other sibs, no stillbirths or abortions. Parents healthy and unrelated. Father aged 30 years, mother 29 years at birth of sib 1, and 32 and 31, respectively, at birth of sib 2. Four maternal cousins with osteogenesis imperfecta.

Clinical examination

Sib 1
At birth. Weight 2700 g (10th centile), length 49.5 cm (25th centile), head circumference 31 cm (less than 3rd centile). Transient nystagmus, hypotonia, atrial septal defect, micropenis, and left undescended testicle. Hypothermia and seizures in neonatal period.

At 30 months. Weight 9 kg (less than 3rd centile), height 81.2 cm (less than 3rd centile), head circumference 42.7 cm (less than 3rd centile) (fig 1). Hypotonia, marked delay in psychomotor development, poorly controlled seizures.

FIG 1 AP and lateral view of sib 1 at 35 months of age. Note depressed nasal bridge.
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