Dysmorphology short reports


Discussion

This male neonate presents a number of striking similarities with the unknown syndrome presented by Young and Simpson. Among these similarities the most important seem to be the facial appearance with bulbous nose and microtretognathia and the hypothyroidism. The only major discordant finding is the absence of a cardiac anomaly in the present patient. Complete necropsy did not reveal other important internal malformations.

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Reference


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A further patient with the lethal type of Larsen syndrome

SUMMARY We present a female infant with multiple joint dislocations, flat facies, cleft soft palate, redundant neck skin, pulmonary hypoplasia, and skeletal abnormalities.

History

Prenatal. No medication, alcohol, or cigarettes. Normal liquor volume. Poor fetal movements.


Family. Both parents healthy; 40 year old mother and unrelated 41 year old father. Two previous pregnancies had ended in spontaneous abortion at eight weeks and fetal death at 22 weeks. Necropsy in the latter case did not reveal any abnormality. The parents had a total of seven healthy children by previous partners.

Clinical examination

External (fig 1). Weight 2550 g (3rd to 10th centile), length 45 cm (3rd centile), OFC 32 cm (3rd centile). Flat occiput, large posterior fontanelle and very narrow, diamond shaped anterior fontanelle. Low set ears, flat nasal bridge, and cleft soft palate. Short, broad neck owing to an excess of subcutaneous tissue. Cervical kyphosis. Dislocation of both hips and knees and talipes equinovarus deformity of the feet. Rhizomelic shortening of upper limbs. Bilateral single palmar creases.

Necropsy. Severely hypoplastic lungs, weight 13 g and 11 g (normal 25 g), small spherical brain weighing 309 g with formation of microgyri in the frontal lobes. Fusion of frontal and parietal bones.

Radiology (fig 2). Hypoplasia of the vertebral bodies from T2 to T8. Severely hypoplasia of the fibulae, especially proximally.

Discussion

This patient has similar features to the two cases reported previously by Chen et al. The Larsen syndrome of multiple congenital dislocations associated with characteristic facial abnormality appears to arise as a result of a generalised mesenchymal disorder and collagen fibre abnormalities have been reported. This is a heterogeneous

FIG 1 Postmortem view of the infant showing joint dislocations, unusual facies, and redundant neck skin.
disorder and Chen postulated that the autosomal dominant form runs a more benign course than the recessive form. All three reported lethal cases have been isolated and in the case above a new dominant mutation seems likely in view of the relatively increased parental age.

We would like thank Dr J D Bell at Furness General Hospital for referring this patient.

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References

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