

of these, Meckel's syndrome, do cardiac anomalies and postaxial polydactyly also occur. The absence of an encephalocele and renal abnormalities in this case makes the diagnosis of Meckel's syndrome unlikely, particularly since cystic renal dysplasia may be a prerequisite for this diagnosis.² The hydroletharus syndrome³ also features in the differential diagnosis, but points against this are the holoprosencephaly with its associated facial dysmorphism, plus the absence of micrognathia and pulmonary abnormalities.

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Unknown syndrome: abnormal facies, congenital heart defects, hypothyroidism, and severe retardation

SUMMARY We present a female infant with (1) abnormal facies: microcephaly, blepharophimosis, small, low set, posteriorly rotated ears, bulbous nose, carp shaped mouth, micrognathia; (2) congenital heart abnormalities: large atrial and ventricular septal defects; (3) congenital hypothyroidism; (4) severe global retardation.

History

Prenatal. No cigarettes or alcohol. Debendox from seven to eight weeks. Amniocentesis at 16 weeks for fetal sexing. Normal intrauterine movements.

Birth. Forceps delivery after induction at 41 weeks.

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Required resuscitation. Apgar scores 2 at one minute, 2 at five minutes, 8 at 10 minutes.

Family. First child of healthy unrelated parents. Father aged 29 years, mother aged 24 years. Maternal brother had Duchenne muscular dystrophy.

Medical history

Dysmorphism noted at birth. Initially hypotonic, then increasing hypertonia. Hypothyroidism treated from two weeks. Pneumonia at eight months and one year. Single febrile convulsion at eight months. Cardiac failure treated from 14 months. Cause of death at 25 months bronchopneumonia.

Clinical examination

At birth. Weight 2.97 kg (10th centile), length 52 cm (50th centile), head circumference 34 cm (10th centile). Slanting forehead, prominent occiput, three fontanelles. Posteriorly rotated, low set ears, both 3 cm long (3rd centile). Right preauricular pit. Blepharophimosis, palpebral fissure width 18 mm. Bulbous nasal tip. Narrow palate with broad alveolar margins. Carp shaped mouth. Micrognathia. Loud pansystolic murmur. Single right palmar crease.

At one year (figs 1 and 2). Weight 6 kg (<<3rd centile), length 69 cm (3rd centile), head circumference 40.8 cm (<<3rd centile). Severe global retardation. Visual

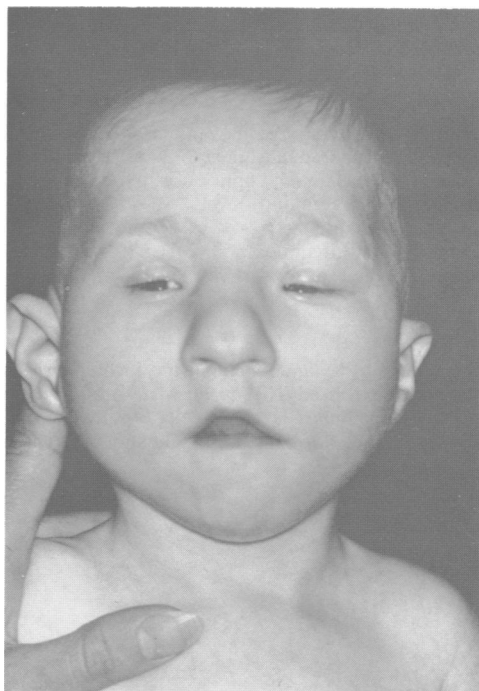


FIG 1 AP view of the patient at one year. Note the short and narrow palpebral fissures and carp shaped mouth.

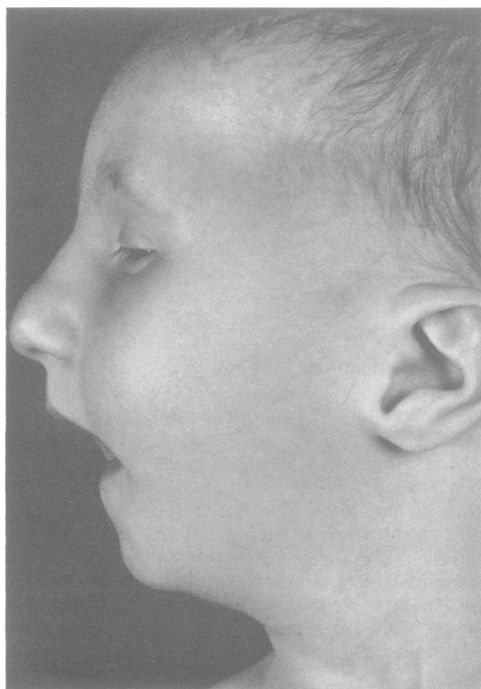


FIG 2 Lateral facial view at one year. Note the low set ear with prominent antihelix, bulbous nasal tip, and marked micrognathia.

inattention with roving eye movements and intermittent nystagmus. Spastic quadriplegia. No structural eye abnormality other than mild optic nerve hypoplasia. Motor development at three month level.

At two years. Weight 6.8 kg (<<3rd centile), length 79 cm (3rd centile), head circumference 42.5 cm (<<3rd centile). Motor development at four month level.

Investigations

Routine haematology and biochemistry, including creatine kinase, normal. Raised TSH (>60 MIU/l) and very low thyroxine (<30 nmol/l) at two weeks of age. Normal G banded karyotype. Normal parental G banded karyotypes.

Skeletal survey showed 11 pairs of ribs. Normal cerebral ultrasound scan in neonatal period. Cross sectional echocardiogram showed moderate ventricular septal defect and large secundum type atrial septal defect. Necropsy was not performed.

Discussion

No satisfactory diagnosis has been reached in this child. The London dysmorphology database¹ lists nine syndromes in which hypothyroidism occurs. In one of these, microcephaly, nystagmus, cardiac anomalies, mental retardation, and spasticity also occur but with a very different facies and much better prognosis.² Congenital abnormalities have been noted in seven out of 34 infants identified through a hypothyroid screening programme.³ None of these infants showed features similar to those in this patient, nor has review of syndromes with blepharophimosis⁴ been fruitful. Only the Marden-Walker syndrome⁵ is similar, but hypothyroidism is not characteristic of this syndrome and our patient had no congenital contractures, cleft palate, or kyphoscoliosis.

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