Dysmorphology short reports

of these, Meckel’s syndrome, do cardiac anomalies and postaxial polydactyly also occur. The absence of an
type 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. The hydrolethalus syndrome 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 abnor-
malities.

References
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delineation of a ‘new’ lethal malformation syndrome based on

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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 hypo-
thyroidism; (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.

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 broncho-
pneumonia.

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. Post-
teriorly 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.
marked micrognathia. 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 database lists nine syndromes in which hypothyroidism occurs. In one of these, microcephaly, nystagmus, cardiac abnormalities, mental retardation, and spasticity also occur but with a very different facies and much better prognosis. Congenital abnormalities have been noted in seven out of 34 infants identified through a hypothyroid screening programme.

None of these infants showed features similar to those in this patient, nor has review of syndromes with blepharophimosis been fruitful. Only the Marden-Walker syndrome is similar, but hypothyroidism is not characteristic of this syndrome and our patient had no congenital contractures, cleft palate, or kyphoscoliosis.

References


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