Dysmorphology reports

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Unknown syndrome: abnormal facies, hypothyroidism, postaxial polydactyly, and severe retardation: a third patient

SUMMARY Young and Simpson¹ in 1987 and Fryns and Moerman² in 1988 each reported a case of a new unknown syndrome with hypothyroidism, severe global retardation, and abnormal facies, including microcephaly, blepharophimosis, bulbous nose, thin upper lip, low set ears, and micrognathia. A male infant with a similar pattern of malformations and postaxial polydactyly is reported here.

History

Prenatal. No cigarettes or alcohol. Unspecified vitamins and analgesics for chest pain in the first trimester. Normal intrauterine movements.

Birth. Vaginal delivery at 38 weeks. Apgar scores of 7 at one minute and 9 at 10 minutes. Weight 2520 g (3rd centile), length 44 cm (<3rd centile), head circumference 32.5 cm (3rd centile).

Family. Third child of healthy, unrelated parents. Both previous children are healthy males and aged nine and six years. Father aged 35 and mother aged 33 years.

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FIGURE AP view of the patient at seven months. Note the short palpebral fissures and long and prominent philtrum.

Medical history

Hypothyroidism diagnosed and treated from the first month. Ear infection at five months. Constipation from the first month. Referred for genetic counselling at six months.

Clinical examination

At seven months (figure). Weight 7150 g (<3rd centile), length 62.5 cm (<3rd centile), head circumference 40.5 cm (<3rd centile). Severe global retardation. High, sloping forehead, thick metopic suture, small palpebral fissures (17 mm), flat nasal bridge, bulbous nasal tip, long and prominent philtrum, microstomia, microretrognathia, low set, malformed ears, short neck, diastasis of recti, unilateral cryptorchidism, and sacral dimple. Bilateral upper limb postaxial polydactyly, postaxial polydactyly on left foot, and narrow, hyperconvex toe nails.

At one year. Weight 8300 g (3rd centile), length 68 cm (<3rd centile), head circumference 42.5 cm (<3rd centile). Severe global retardation.

Investigations

Low neonatal thyroxine (4.4 µg/100 ml, normal range 6.0 to 15.0). Serum TSH at five months was 2.6 µl/ml (normal, up to 8.0 µl/ml). Radiographs of the knees and wrists at three months showed absence of the distal femoral, proximal tibial, and distal ulnar and radial epiphyseal centres. A CT scan of the head at one year was normal. Normal G banded male karyotype in cultured lymphocytes and fibroblasts.

Discussion

This patient has similar features to the two cases reported previously in this journal by Young and Simpson¹ and Fryns and Moerman.² The most important clinical signs of this syndrome seem to be the congenital hypothyroidism and facial appearance with blepharophimosis, bulbous nose, thin upper lip, and micrognathia. The two major discordant findings are the cardiac anomaly present only in the case of Young and Simpson¹ and the postaxial polydactyly occurring only in the present case.

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Unknown syndrome: microcephaly, hypoplastic nose, exophthalmos, gum hyperplasia, cleft palate, low set ears, and osteosclerosis

SUMMARY We report a neonate, born at term, with microcephaly, a hypoplastic nose, exophthalmos, gum hyperplasia, cleft palate, and low set ears, who died at 86 minutes of age. Radiographs showed diffuse sclerosis of the bones.

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FIG 1 AP view of face.

FIG 2 Lateral view of face.

History

Prenatal. No infections or drugs in pregnancy.

Birth. Cephalic delivery at term. Female infant. Weight 2730 g (10th centile), length 44.5 cm (<3rd centile), and occipitofrontal circumference 30 cm (<3rd centile). Apgar scores 8 at one and five minutes. Noted to be hypotonic.

Family. Healthy, unrelated, Caucasian parents. One healthy daughter. One stillbirth at term after placental abruption: normal baby. One first trimester miscarriage. No family history of congenital abnormalities.

Clinical examination (figs 1, 2, and 3)

Microcephaly. Sagittal suture 5 cm wide with large anterior and posterior fontanelles. Ridged coronal sutures. Marked exophthalmos. Maxillary hypoplasia with a sunken nasal bridge and micrognathia. Bowed upper lip. Cleft in soft palate and uvula. Hyperplastic premaxillary and mandibular gums. Low set ears.

Subsequent course

Placed in a cot in the special care baby unit. Respiratory distress developed within 30 minutes of birth, presumably secondary to pulmonary hypoplasia. Because of the multiple severe abnormalities a policy of conservative treatment was adopted. Gasping at 80 minutes at age, she died at 86 minutes of age.