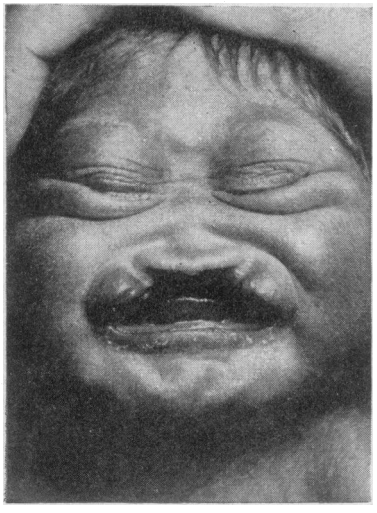

Fig 1 *Mid-line cleft of lip and palate with microcephaly*

The combination of these abnormalities in one infant appears to be rare.

The patient (Fig 1), a female infant, was born spontaneously after an uneventful thirty-four week pregnancy to an 18-year-old married woman, who has one other, normal, child, whose birth weight was 3 lb 15 oz. No family history of facial clefts, no consanguinity, no ingestion of teratogenic drugs.

The child's birth weight was 3 lb 8 oz, length 16½ in. (41·9 cm), head circumference 10¼ in. (26 cm). There is a central hare lip, absent premaxilla and globular process, and a complete central cleft palate. The nasal bones are hypoplastic. An air encephalogram (Fig 2) shows hydranencephaly. Three attempts to introduce air through the lumbar or cisternal route failed, and only direct ventriculography led to the diagnosis of hydranencephaly. This may be due to agenesis of the aqueduct, which is occasionally associated with hydranencephaly (Crome & Sylvester 1956).

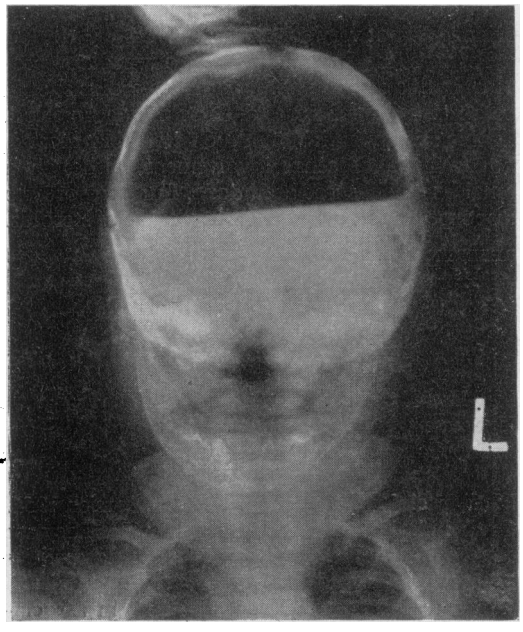

Fig 2 *Hydranencephaly and absent falx*

Since the age of 6 weeks the infant has had fits, which are now fairly well controlled by an anticonvulsant. Throughout, her body temperature has varied between 94°F and 104°F. This bizarre temperature has remained unexplained and is probably due to an abnormality of her heat-regulating centre. At the age of 15 weeks she has a marked microcephaly, her head circumference being 12⅛ in. (30·5 cm). There is gross mental retardation.

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Hereditary Ectodermal Dysplasia

John Lorber MD MRCP

P S, male, aged 3 years. Born 10.10.60. Birth weight 7 lb 1 oz

Principal physical features: Small, 24 lb (below 5th percentile). Very sparse hair and eyebrows. Low-set ears of unusual shape. Small head, circumference 18½ in. (47 cm). Only two, misshapen (cone-shaped), upper incisor teeth (Fig 1). No other deciduous teeth. Only four other deciduous and no permanent teeth demonstrable on radiograph of his jaws (has lost no teeth). Scaling erythema. Not photosensitive. Unable to sweat. Absence of nipples (Fig 2). Normal nails. Mental development adequate.

Family history: His father and all uncles, cousins, &c., are normal. His mother is unable to sweat. His first sib, aged 8, is normal and has normal dentition.

His second sib, who died at the age of 4 months in 1957 during a 'sweat test', was unable to sweat and was frequently treated for respiratory infections. Chance X-rays of jaws included on chest film show absence of teeth (retrospective observation). The diagnosis of ectodermal dysplasia was never considered in that infant.

Comment

This child presents most of the characteristic features of hereditary ectodermal dysplasia. There is failure of development of his hair, teeth,

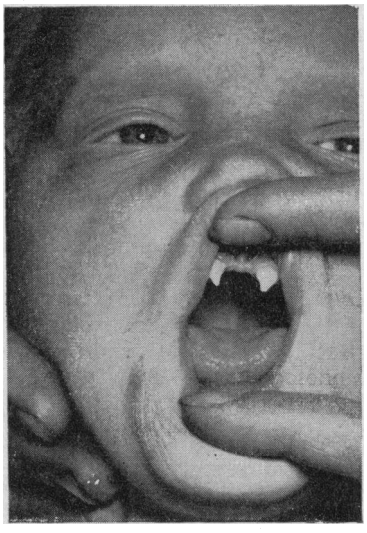

Fig 1 *Showing two mis-shapen cone-like upper incisor teeth and sparse eyebrows*

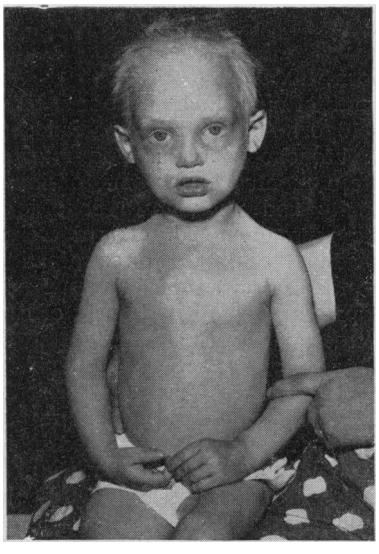

Fig 2 *General appearance with sparse hair, scaly erythema on face and absence of nipples*

sweat and sebaceous glands and he has no nipples. He cannot bear hot weather and one summer he nearly died of heat exhaustion. His brother died due to overheating. This is the most important feature of the disease, as tragedies can be avoided if this diagnosis is borne in mind. Adequate orthodontic treatment is also desirable from an early age. The hereditary pattern may vary; the condition is often sex linked and males are more commonly affected.

Nonprogressive Neurogenic Muscular Atrophy with 'Voluntary Fasciculation'

Victor Dubowitz MD MRCP

M K, male, born 31.3.55

History: The pregnancy, labour and neonatal period were normal. He sat with support and started to crawl at 10 months. He tried to stand at one year, but was very floppy. By the age of 3 years he was able to walk with a walking machine, to ride a tricycle and to stand for short periods with support. After calipers were fitted at the age of 4¾ he was able to stand and to walk. His walking has gradually improved since then. He is now also able to stand for short periods without calipers. His intellectual development has been normal.

On examination (August 1961): Generalized weakness of the trunk and limb muscles; no facial involvement. The tongue showed marked atrophy and fasciculation, both at rest and on protrusion. He stood with marked pes planus and eversion of the feet (Fig 1). No fixed scoliosis or contractures. Ankles showed an increased range of mobility. Tendon jerks all absent; plantar response flexor.

No fasciculation was noted in his skeletal muscles, but voluntary flexion of the right middle finger produced a spontaneous large amplitude 'tremor' of that finger and an associated palpable 'fasciculation' of the belly of the flexor digitorum muscle. The movement continued until he extended the finger again. No similar response could be elicited elsewhere.

Course and progress: Over a two-year period there has been no deterioration. His locomotion has shown definite improvement. The atrophy and fasciculation of the tongue and the unusual movement of the finger remain unchanged.

Investigations

Serum enzymes: Glutamic oxaloacetic transaminase 53, aldolase 13, creatine phosphokinase 6·3 units.

Muscle biopsy (triceps): Typical picture of long-standing neurogenic atrophy with numerous groups of atrophic fibres, 5–10μ in diameter, and some residual normal-sized and giant fibres 60–150μ in diameter (Fig 2).

Electromyography: Denervation pattern. Frequency of contractions in flexor digitorum muscle (after flexing finger): 10/sec.

Comment

The clinical picture and course in this child are consistent with a benign congenital hypotonia, but the fasciculation and atrophy of the tongue suggest a denervation atrophy. This is supported by the histological and electromyographic findings.