(n=1). No lead points were found in our 10 patients, although most similar series tend to implicate inverted appendiceal stump,² anastomotic suture line,³ unrecognised polyp,² hypertrophied lymph nodes,² Meckel's diverticulectomy area,^{3,5} traumatised intestinal wall.¹

Once operated on, all infants and children (ours and in other series¹⁻⁷) recovered well without an increase in their chances for future recurrent intussusceptions.

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Orocraniodigital (Juberg-Hayward) syndrome with growth hormone deficiency

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SUMMARY A boy is described with Juberg-Hayward syndrome. In addition to unilateral cleft lip and palate, bilateral absent thumbs, anomalous carpal bones, and deformity of the radial heads, there were other skeletal abnormalities. The patient showed clinical and biochemical evidence of growth hormone deficiency which appears to be an integral part of the syndrome.

The orocraniodigital syndrome was first described by Juberg and Hayward¹ in 1969, affecting 5 children from a sibship of six. The expression of the syndrome was variable in these individuals, the 2 brothers being more severely affected than the 3 sisters. The main features described were unilateral or bilateral cleft lip and palate, hypoplasia and distal displacement of the thumbs, bilateral forearm deformities with anterior displacement of the radial head, anomalous toes, and mild microcephaly. The inheritance appeared to be autosomal recessive with variable expression. The possible sex limitation suggested originally now seems unlikely since the case reported by Nevin *et al.*² in 1981, in which the patient was a female with full expression of the syndrome. That patient also had additional skeletal abnormalities, notably absence of the pituitary fossa and flattened vertebral bodies. No evidence of endocrine dysfunction was found to account for her short stature.

In this report we describe a boy with features of the orocraniodigital syndrome, who had growth hormone deficiency as well.

Case report

The patient, a 17-year-old boy, was the only child of healthy unrelated parents. At the time of conception the mother was aged 43 and the father 38 years, neither had oral, cranial, or digital abnormalities and each was of normal intelligence. Their heights were 155 cm (−2 SD) and 160 cm (−2 SD). There were no congenital abnormalities in more distant family members. The patient was born at term after a normal pregnancy, with a birthweight of 1.24 kg. Unilateral cleft lip and palate and bilateral absent thumbs were noted at birth. Surgical correction of the cleft lip was carried out at 9 months and of the palate at age 2 years. His progress was satisfactory, apart from short stature and hearing impairment. At age 17 years his height was 143.3 cm (−4.7 SD), weight 49.4 kg (−2 SD), and head circumference 54.4 cm (−1 SD). Upper to lower segment ratio was 1:1. The facies (Fig. 1) showed a prominent forehead with hypoplasia of the mid-face, micrognathia, and repaired unilateral cleft lip and palate. He had a short neck, broad chest, and micropenis. Testes measured 12 ml in volume bilaterally and pubic

hair was Tanner's stage 4. The thumbs were bilaterally absent, the left index finger having been pollicised. The left forearm was short and there was incomplete extension at the elbows with restricted supination of the forearms bilaterally. Intellectual function corresponded to the 50th centile on the matrices and the Mill Hill vocabulary scale.

On x-ray films the skull and pituitary fossa were normal. There was some flattening of the cervical vertebral bodies with narrowing of C6 and C7 disc spaces and spina bifida occulta of the sacral segments. The right 12th rib was absent. The long bones were of normal shape, except for shortening of the right radius. There was bilateral deformity

and dislocation of the radial head. The thumbs and first metacarpals were bilaterally absent, with abnormal carpal bones on the radial side of the wrist, associated on the right with subluxation of the carpus (Fig. 2). All metacarpal and phalangeal epiphyses were fused, indicating a skeletal age consistent with chronological age. Intravenous urography showed small rotated kidneys.

Laboratory studies. Chromosome analysis was normal 46XY. Hypothalamic-pituitary function was studied using a standard triple stimulation followed by arginine stimulation test.³ The results showed isolated growth hormone deficiency; basal level of GH < 1.0 mU/l, peak level after stimulation 3.0 mU/l.

Discussion

The severity and extent of the abnormalities in the orocraniodigital syndrome are variable. The minimum criteria suggested by Juberg⁴ for the diagnosis in an isolated case are unilateral or bilateral cleft lip and palate, anomalous thumbs and toes, and mild microcephaly. In addition to these abnormalities, low birthweight and short stature have been constant findings, while features of a widespread skeletal disorder are more variable, and there may be an associated renal abnormality.^{1,2} IQ and chromosome analysis appear to be normal.

No endocrine studies were reported in the cases of Juberg and Hayward,¹ and although the patient of Nevin *et al.*² had an absent pituitary fossa on skull x-rays films, no endocrine abnormality could be found to explain the short stature. Our patient has documented isolated growth hormone deficiency. However, because of his age and skeletal maturation he is likely to achieve limited benefit from growth hormone replacement therapy. Early diagnosis and treatment of growth hormone deficiency in such cases is essential in order to achieve maximum growth potential.

It has been estimated that growth hormone deficiency occurs 40 times more often in short children with cleft lip and palate than in those without.⁵ An embryonal developmental defect is the likely cause of this association, since the lip, palate, and adeno-hypophysis arise in close proximity. The association of radial defects and cleft lip and palate also appears to be non-random. Kato⁶ in 1924 found orofacial malformations in 2.5% of cases with radial defects. Any relationship between cleft lip and palate with growth hormone deficiency, or cleft lip and palate with radial defects and the full Juberg-Hayward syndrome requires further elucidation.

In addition to the minimum criteria for the

Fig. 1 The patient aged 17 years.

Fig. 2 X-ray film of hands and wrists.

diagnosis of the orocraniodigital syndrome, our patient has a more widespread skeletal dysplasia, with absence of thumbs, and isolated growth hormone deficiency. As he is the only child of non-consanguineous parents, no further information is gained regarding the mode of inheritance.

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Neonatal rickets in one of identical twins

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SUMMARY We report a case of rickets in one of identical low birthweight twin infants. Plasma 25-hydroxycholecalciferol concentrations were normal in both infants; there was good clinical response to 1- α hydroxycholecalciferol. We suggest that there was a delay in maturation of renal 25-hydroxycholecalciferol-1 α hydroxylase enzyme.

The problem of rickets in the low birthweight infant has been known for some time.¹ The diagnosis is made on clinical, biochemical, and radiological criteria; serial measurement of plasma alkaline phosphatase activity is useful to screen for disease.² The aetiology of neonatal rickets is multi-factorial. We report a case of rickets in one of identical twins.

Case report

Female twins (I vertex, II breech) were born at 30 weeks' gestation after a normal pregnancy to a 23-year-old healthy well-nourished white primigravida. The membranes of twin I ruptured spontaneously 36 hours before, and those of twin II at delivery. The placenta weighed 680 g and was mono chorionic and diamniotic. The blood group of both infants was ORh⁺ (mother ARh⁺) and identical antigens for the common groups, including Lutheran, Lewis, and Duffy were found. The genotype was M⁺ N⁺ S⁻ $\bar{s}$ ⁺ P⁺ Lu^a-Le^a (a-b⁺) Fy^(a+b+) for both children.

Plasma alkaline phosphatase, calcium, and inorganic phosphorus were measured serially from age 2 weeks.² Rickets was diagnosed in twin II at age 6 weeks (36 weeks' post-conceptual age). There was no evidence of rickets in twin I either at this age

Childhood reference ranges²: Plasma alkaline phosphatase upper limit 850 U/l, plasma calcium 2.20 - 2.67 mmol/l, plasma inorganic phosphorus 1.02 - 1.96 mmol/l.

Conversion: SI to traditional units - plasma calcium: 1 mmol/l $\approx$ 4.0 mg/100 ml. Plasma inorganic phosphorus: 1 mmol/l $\approx$ 3.0 mg/100 ml.

Figure Serial plasma alkaline phosphatase, calcium, and inorganic phosphorus in both twins.