FIGURE (a) Facial appearance of proband. (b) Bronchography showing absence of right lung and mainstem bronchus. The tip of the orogastric catheter at the blind end of the oesophagus is marked by an arrow.

left lung. There were only two pulmonary veins draining the left lung. A small spleniculus was found at the hilus of the spleen. The cerebellum was markedly hypoplastic.

Discussion

Since the first description of the Goldenhar anomalad in 1952,² variable combinations of facial, oral, auricular, vertebral, ocular, and cardiac malformations have been described. Other occasional abnormalities include branchial cleft remnants, laryngeal anomalies, hypoplasia or aplasia of the lung, occipital encephalocele, and renal and limb anomalies.¹ The combination of oesophageal atresia and pulmonary agenesis has been reported in 14 cases, recently summarised.³ Such an association with the Goldenhar anomalad has not been previously described. Bowen and Parry,⁴ however, described a patient with bronchopulmonary-foregut communication and hypoplasia of the right lung but with a patent oesophagus. Thus, our report is the second

instance where tracheobronchial-oesophageal communication was found in association with the Goldenhar anomalad. Awareness of this combination may facilitate future diagnosis and treatment of the anomaly.

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Severe pseudoachondroplasia with parental consanguinity

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SUMMARY A boy who showed features of the severe form of pseudoachondroplasia, whose parents were first cousins, is reported. Published reports supporting the existence of an autosomal recessive form of this disorder are reviewed.

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Pseudoachondroplasia comprises a relatively common form of short limbed dwarfism with involvement of the spine, metaphyses, and epiphyses, but not the skull. Clinically different grades of severity can be recognised and provisionally four forms, severe and mild, dominant and recessive, have been identified.¹ In this report a child who showed features of severe pseudoachondroplasia is described. The parents were first cousins.

Case report

The patient, a 6 year old boy, was the second child of healthy Indian Muslim parents aged 28 and 27 years at the time of his conception. An older brother and younger sister were healthy. The paternal grandmother and maternal grandfather were sibs.

Birth weight was 2.7 kg at term. No problems were noted until the age of 23 months when he presented with short stature (height 70 cm) and 'swollen joints'. Investigations excluded rickets and a mucopolysaccharidosis. From the age of 2 years the short stature became increasingly obvious and he developed a waddling gait. Locomotion beyond a short distance was limited by joint pain. Apart from

these problems his general health was good and he was of normal intelligence.

On examination at 6 years (fig 1) growth parameters were head circumference 47 cm, height 78.5 cm, lower segment 30 cm, and span 66.5 cm. He had a weak high pitched voice, pigeon shaped chest, excessive skin folds, increased lumbosacral lordosis, and generalised shortening of all limb segments with mild genu varum and bowing. Large osseous swellings could be palpated at all limb joints which showed a restricted range of movement in association with increased joint laxity. His fingers were short and stubby and of an unusually soft consistency.

Radiographs of the spine (fig 2) revealed irregu-

FIG 1 The patient aged 6 years. His hips, elbows, and knees are fully extended.

FIG 2 Lateral view of the spine at 6 years. There is a mild thoracic kyphosis with exaggerated lumbar lordosis.

FIG 3 The pelvis at 6 years. Note the very small upper femoral epiphyses with irregular acetabular walls.

larity of the vertebral bodies with anterior 'tonguing'. In the pelvis (fig 3) the acetabulae were dysplastic with small femoral epiphyses. The tubular bones of the arms and legs (fig 4) showed shortening with irregular modelling of the diaphyses, pronounced metaphyseal flaring, and small irregular epiphyses. Views of the skull were normal.

Examination of the parents and sibs failed to reveal any abnormality.

Discussion

The clinical and radiological findings in this child are consistent with a very severe form of pseudoachondroplasia and the parental consanguinity points to autosomal recessive inheritance. Variability in the severity of pseudoachondroplasia is well documented both between^{1 2} and within^{3 4} families, so that while some have attempted to subdivide on clinical and genetic grounds,^{1 2} others have concluded that clinical severity and radiological changes

may represent a continuum rather than clear cut discrete entities.^{3 4}

The difficulty in establishing genetic heterogeneity has been compounded by recent reports that one of the two sibs originally reported as having the severe recessive variety¹ has had an affected child, thus raising the possibility of gonadal mosaicism in one of the sibs' parents.^{4 5} However, a review of published reports reveals at least two other sibships of normal parents, consanguineous in one instance, each containing more than one child with a severe form of pseudoachondroplasia,^{6 7} so that it seems likely that a severe recessive variety exists.

It is interesting that the heights of these published cases all fall below -1 SD from the mean for patients with pseudoachondroplasia,⁸ as does the subject of this report. It is not suggested that referral to these growth curves for pseudoachondroplasia provides an accurate means of distinguishing between dominant and recessive forms, although likelihood odds for one or the other might be

FIG 4 (a) The upper limb at 6 years. There is shortening of the tubular bones with mild radial bowing, metaphyseal flare, and tiny irregular epiphyses showing delayed maturation. (b) The lower limb at 6 years. The lower femoral metaphyseal flare is striking.

Case reports

derived if information about sufficient cases was forthcoming. Continued progress at the biochemical level⁹ may also provide a means of separating the different genetic entities. In the meantime it is suggested that the present case lends additional support for the existence of a severe autosomal recessive form of pseudoachondroplasia.

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