Cleft palate and accessory metacarpal of index finger syndrome: possible familial occurrence

SUMMARY A case of cleft palate and accessory metacarpal of index finger syndrome is described and related to the presence of Pierre Robin syndrome in a stillborn sibling. The significance of this relationship is discussed.

The Pierre Robin syndrome is classically comprised of cleft palate, glossoptosis, and micrognathia (Smith, 1976). One variant of this condition is the cleft palate and accessory metacarpal of index finger syndrome which has so far been described in 3 cases, all of which have been sporadic in occurrence (Holthusen, 1972; Manzke, 1966; Farnsworth and Pacik, 1971). This report describes a further case together with a sib who had classical Pierre Robin syndrome. This may represent the first reported familial incidence of the syndrome.

Case report

R.P. was born at term by caesarean section to a 28-year-old gravida 2, para 1 mother. Caesarean was done because the mother had previously delivered a stillborn male infant with classical Pierre Robin syndrome and a 4 mm secundum atrial septal defect. Father was also aged 28 years. There was no history of parental consanguinity or of maternal drug or alcohol abuse during pregnancy. Birthweight was 3-21 kg. Respiratory difficulties developed immediately after birth and intubation was performed with difficulty because of micrognathia and cleft palate. After resuscitation he was managed prone on a specially constructed frame to ensure an optimum airway and to facilitate feeding.

Clinical examination revealed typical facial features of Pierre Robin syndrome (Fig. 1). In addition symmetrical angulation deformities of both index fingers, confirmed by x-ray examination as due to accessory metacarpals, were present (Fig. 2). X-rays of the feet were normal. Height and weight gain continued well below the third centile for age, and tachypnoea and recession persisted. Cardiac examination revealed a midsystolic murmur at the upper left sternal edge associated with wide, fixed splitting of the second heart sound. Electrocardiogram showed changes compatible with secundum atrial septal defect. He subsequently developed recurrent upper airway obstruction, respiratory tract infections, and a degree of cor
Case reports

Fig. 1  Facial features of Pierre-Robin syndrome present in this patient.

Fig. 2  Hand x-ray picture from patient showing bilateral symmetrical angulation deformities of the index fingers.
the Pierre Robin syndrome and thus the coexistence of the 'accessory metacarpal' may imply a substantial recurrence risk in sibs.

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References


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Announcement

The University of California, San Francisco, and the National Foundation—March of Dimes are sponsoring the 1978 Birth Defects Conference, 12–14 June 1978. For information write to Dr. Bryan D. Hall, M648, Department of Pediatrics, University of California, San Francisco, California 94143, USA.
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