Familial radioulnar synostosis

**SUMMARY** A family with proximal radioulnar synostosis segregating in three generations is described. Familial radioulnar synostosis is a rare anomaly; however, the sporadic form is a frequent feature in cases of sex chromosome abnormalities and other syndromes. This disorder has been reported in several ethnic groups, but this is apparently the first example from the black population.

Congenital synostosis of the proximal radius and ulna was first described by Sandifort in 1793, with over 265 cases reported since. Though most cases are sporadic, a small number are familial. The first report of familial radioulnar synostosis was by Abbott in 1892, and approximately 18 families have been described to date (Abbott, 1892; Davenport et al., 1924; Fahlstrom, 1932; Hansen and Andersen, 1970; Berant and Berant, 1973). Almost all of these families are of western European origin, several of Jewish descent.

Familial radioulnar synostosis tends to be bilateral, and a majority of the affected are males. The inheritance pattern is autosomal dominant, but incomplete penetrance is common.

This report describes bilateral proximal radioulnar synostosis in three generations of a black American family. The clinical syndrome, its embryology, and treatment are discussed.

**Case report**

The propositus, a 3-year-old black boy, was the product of an uncomplicated 36-week pregnancy. Birthweight was 2350 g (25-50%), length 46 cm (25-50%), head circumference 32 cm (25-50%), and chest circumference 30 cm (25%). One- and five-minute Apgar scores were 8 and 9, respectively. Bilateral limitation of forearm supination was noted soon after birth. The neonatal period was otherwise uncomplicated, and subsequent growth and development were normal.

When examined at 3 years, the patient was a bright, well-developed boy. Height was 98 cm (75-90%) and weight was 13.2 kg (25%). There was distinct limitation of forearm supination from the vertical, bilaterally (left 20°, right 15°). Deep tendon reflexes were depressed. The remainder of the physical examination was normal.

Radiographs revealed bilateral synostosis of the proximal ends of the radius and ulna (Fig. 1). CBC was normal. Buccal smear was 100% chromatin negative.

**PEDIGREE**

The family pedigree is illustrated in Fig. 2. Affected individuals occur in three successive generations. The patient's mother (III.2) has almost total restriction of forearm motion subsequent to bilateral osteotomies in childhood. A maternal uncle (III.4) and great uncle (II.5) of the patient are also affected with bilateral radioulnar synostosis, but have not been studied. The maternal grandmother (II.2) of the propositus is unaffected, showing incomplete penetrance.

**Comment**

The kindred illustrates several of the typical features of the familial congenital radioulnar synostosis syndrome, including bilateral involvement, autosomal dominant inheritance, and incomplete penetrance. Though the maternal grandmother of the propositus must carry the abnormal gene, she is unaffected. Males are more often affected than females. Familial radioulnar synostosis tends to be bilateral, but some families tend to exhibit unilateral involvement. Radioulnar synostosis has been associated with
congenital hyperthyroidism, familial craniosynostosis (Berant and Berant, 1973), and other skeletal malformations (Davenport et al., 1924; Hansen and Andersen, 1970). It is often seen in cases of sex chromosome abnormalities (Cleveland et al., 1969; Jancu, 1971), and sporadic cases of congenital radioulnar synostosis should be investigated by karyotyping. Familial radioulnar synostosis, however, usually occurs as an isolated abnormality, and the karyotype may be expected to be normal.

Congenital radioulnar synostosis results from an arrest of development at the fifth to sixth week. The radius, ulna, and humerus derive from a single precartilagenous mesenchymatous plate. Radioulnar synostosis results from a failure of differentiation of this plate into distinct cartilagenous rods. Another type of radioulnar synostosis develops in late fetal life from periosteal abrasion and new bone formation at points of contact between the radius and ulna (Wilkie, 1914). Rarely, acquired radioulnar synostosis may arise as a result of fracture of the radius or ulna.

Surgical treatment of radioulnar synostosis has led to disappointing results. Resection of the synostosis, with separation of the radius and ulna by fat, muscle, or inert material, usually leads to severe limitation of forearm mobility. Insertion of a stainless steel swivel
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 sibling 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 consanginity or of maternal drug or alcohol abuse during pregnancy. Birthweight was 3.2 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 were 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 mid-systolic 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

References


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