Familial postaxial acrofacial dysostosis syndrome

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Most known cases of postaxial acrofacial dysostosis syndrome (POADS) are sporadic. The familial case of Robin sequence and oligodactyly occurring in a mother and her two sons, reported by Robinow and Chen¹ as a previously unrecognised malformation syndrome, was later regarded as an example of POADS² and autosomal dominant inheritance of this condition was suggested. However, other familial cases have been reported in three pairs of sibs born to healthy, unrelated parents.³⁻⁵ In addition, one patient was born to consanguineous parents.⁶ These observations argue for autosomal recessive inheritance of this syndrome (MIM 263750). We report here a family with two affected sibs which support this.

Case 1 (figure A,B) is a male infant, the last child of a sibship of four. The parents were non-consanguineous and clinically normal.

The baby was delivered by caesarean section at term after an uneventful pregnancy. Birth weight was 2700 g, length 48 cm, and head circumference 35 cm. Clinical evaluation at 1 month showed downward slanting palpebral fissures, bilateral coloboma of the eyelids, cleft of the hard palate, glossoptosis, micrognathia, low set, dysmorphic ears, ulnar deviation of the hands at the wrist, and absence of the fifth fingers and toes. X ray examination of the upper and lower limbs showed short forearms with bilateral radioulnar synostosis and absence of the fifth digital ray of the hands and feet. The karyotype was normal.

An older sister of the patient (case 2, figure C,D) died at 13 days of life. She had a constellation of anomalies similar to the ones detected in the patient, including cleft hard palate, micrognathia, low set and malformed ears, absence of the fifth digital rays of the upper and lower limbs, and bilateral ocular glaucoma. The neonatal period was complicated by recurrent cyanosis and apnoea which required continuous medical care. Photographic evaluation of this patient suggested a diagnosis of POADS.

These two sibs, together with four previously documented familial cases of POADS, strongly support autosomal recessive inheritance of this condition. Thus, a one in four recurrence risk should be given to the patient's parents. At present, the existence of genetic heterogeneity in POADS, in our opinion, is supported only by the familial observation of Robinow and Chen.¹ However, critical re-evaluation of that syndrome does not obviously suggest that it should be lumped with POADS.

Facial appearance and feet of case 1 (A,B) and case 2 (C,D).

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