Familial Occurrence of Pierre Robin Anomalad

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The Pierre Robin anomalad is a congenital defect consisting of micrognathia, glossoptosis, and cleft palate. While these three stigmata represent the classical triad of anomalies first described by Pierre Robin in 1923,[1] less severe forms of the anomalad are seen with a variable expression of the microsigns of the disease. Dennison[2] suggests that the determination of exactly what constitutes a mild degree of Pierre Robin anomalad is a subjective matter. While cleft palate is commonly present, it may sometimes be replaced by a high-arched palate. He reports that radiography shows that there is a short mandibular body joining the ramus at a more acute angle than normal. The more severe cases can lead to respiratory obstruction and death. Anatomically, the defect is quite clear; the body of the mandible is posteriorly displaced causing the tongue to block normal respiratory and alimentary pathways.

No mode of inheritance has yet been clearly established. While the majority of cases of this rare disorder (1:50,000 births)[3] appear to be sporadic, there have been several reports of familial occurrence; cases of affected brothers have been presented[4, 5] and an X-linked subvariety exhibiting persistence of the left superior vena cava and atrial septal defect has been described.[6] A family having maternal half-siblings with the Pierre Robin anomalad is reported here. The mother bore a single affected male and four normal females by her first husband and two affected females by her second husband. Strong evidence exists for dominant transmission through the mother, who herself exhibits mild characteristics of the disorder. We therefore propose that this family represents an autosomal dominant form of the Pierre Robin anomalad with variable expressivity.

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Case Report

The proband (Fig 1, III,1) was born on July 22, 1976, of a 26-year-old mother and a 25-year-old father. She was the 8 pound 10½ ounce full-term product of a pregnancy complicated by maternal vaginal discharge which was treated with a cream and resolved in the second trimester, and leaking of amniotic fluid with spontaneous resolution from the fourth through the sixth months. The patient was born by a normal vaginal delivery, cried immediately, and did not need resuscitation. Midline cleft palate and micrognathia were noted at birth, with the lips intact. Other than two left preauricular skin tags and possible bilateral high frequency hearing loss, no other abnormalities were present. She breathed rapidly and became cyanotic when allowed to lie flat on her back with resultant backward displacement of the tongue. Postnatal jaundice was treated with phototherapy. She was discharged with special feeding instructions, but readmitted a month later due to poor weight gain. At two months she was noted to have a grade I

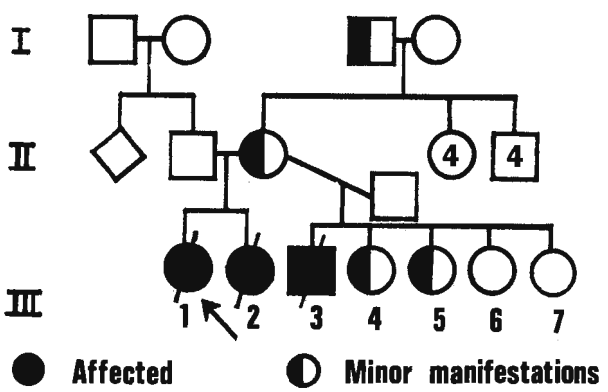

Fig 1—Pedigree of proband's family showing affected half-siblings.

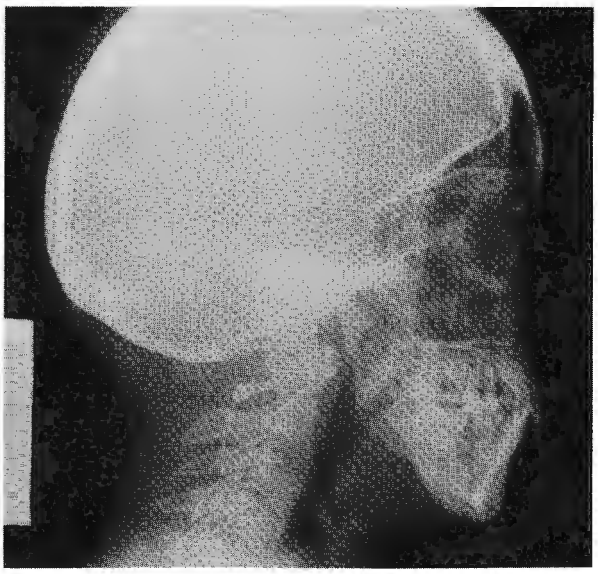

Fig 2—Lateral x-ray of proband's mother showing steep mandibular angle.

to II/VI systolic ejection murmur, but chest x-ray, electrocardiogram, and a cardiology consultation suggested that there was no organic heart disease present. The patient was hospitalized many times during the first months of life due to multiple episodes of pneumonia and the inability to maintain adequate nutrition. At age 7 months a tracheostomy was done which resulted in improved breathing and weight gain. At age 11 months the patient was admitted for bronchoscopy and removal of the tracheostomy tube; she was unable to breathe adequately without it, and the tube was reinserted. She died suddenly at the age of one year, but no postmortem examination was permitted.

The proband's sister (Fig 1, III,2), also affected with Pierre Robin anomalad, died two months after birth as a result of poor weight gain and a terminal bout of aspiration pneumonia. Their half-brother (Fig 1, III,3), who died 28 days after birth, had severe congenital heart disease, including persistent left superior vena cava, tricuspid atresia, atrial septal defect, right-sided aortic arch, and descending aorta in addition to the Pierre Robin anomaly. Of the four normal half-sisters of the proband (Fig 1, III,4,5,6,7), two have slightly high, narrow palates and small chins. The mother has a very high-arched palate and micrognathia. A lateral x-ray of the mother's jaw demonstrated that one mandibular ramus was smaller and narrower than the other, presenting as asymmetrical appearance, and the mandibular angle was steeper than normal (Fig 2).

Discussion

We have presented a family in which three children (1 male and 2 females) who had two different fathers were affected with the Pierre Robin anomalad. The mother exhibits some of the characteristic stigmata of this syndrome which strongly suggests that an autosomal dominant form of the Pierre Robin anomalad exists with variable expressivity.

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