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Congenital Heart Disease in the Pierre Robin Syndrome

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SUMMARY. **Congenital heart disease occurs in about 20 percent of patients with Pierre Robin syndrome. Ventricular septal defect, patent ductus arteriosus, and atrial septal defect are the most common congenital cardiac lesions in this syndrome. The associated upper airway obstruction can produce cor pulmonale, cardiomegaly, pulmonary edema, and cyanosis.**

KEY WORDS: **Pierre Robin syndrome — Congenital heart disease**

The Pierre Robin syndrome consists of hypoplasia of the mandible (micrognathia), together with a posterior and downward displacement of the tongue (glossoptosis) that acts as a ball valve preventing air from entering the glottis [4]. This syndrome was first described by Fiarbian in 1846 [5]. In 1923, Pierre Robin described a series of children having micrognathia, glossoptosis, and respiratory distress [14]. In 1934, he indicated that cleft palate, which occurs in half of these patients, should also be considered part of the syndrome that now bears his name [15].

Congenital heart disease occurs in approximately 20 percent of children with Pierre Robin syndrome [12].

The following case is an example of the Pierre Robin syndrome with congenital heart disease:

Case Report

This 12-month-old white female was born weighing 2,860 gm, following a gestation estimated to be of 43 weeks duration. The amniotic fluid was heavily stained with meconium, and her Apgar scores were 2 at 1 minute and 6 at 5 minutes. The neonatal period was complicated by persistent feeding difficulties and recurrent aspirations, necessitating the construction of a feeding gastrostomy at 1 month of age.

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The patient is in mild respiratory distress and developmentally delayed. Her weight (5.5 kg) is well below the third percentile, and her height (69 cm) is at the third percentile. Her pulse is 120 beats per minute, her respiratory rate is 44 per minute, and her blood pressure is 98/50 mm Hg. The mandible is hypoplastic (Fig. 1), and there is a cleft of the soft palate. Rhonchi are audible over the bases of both lungs. The precordium is hyperactive at the left lower sternal border. The first heart sound is single and of normal intensity. The second heart sound is also of normal intensity and persistently split. There is a Grade III/VI midsystolic crescendo-decrescendo murmur heard best at the second left intercostal space. Diastole is silent. The liver and spleen are not palpable, and a feeding gastrostomy is in place. The radial and femoral pulses are simultaneous and of normal volume. There is no clubbing, cyanosis, or edema.

The electrocardiogram has an indeterminate mean frontal QRS axis. It indicates right atrial enlargement and right ventricular hypertrophy. The chest roentgenogram reveals a slightly enlarged heart with an elevated apex. The pulmonary vasculature is normal. Right- and left-sided cardiac catheterization and angiography demonstrated an atrial septal defect of the ostium secundum variety. The pulmonary to systemic flow ratio was 3.4 to 1, and the pulmonary artery pressure was 25/7 (mean = 17) mm Hg.

Discussion

Infants with the Pierre Robin syndrome generally develop choking, swallowing difficulties, and intermittent cyanosis within a few days of birth [20]. Their hypoplastic mandibles are variously described as "bird-face", "shrew-face", or "Andy Gump."

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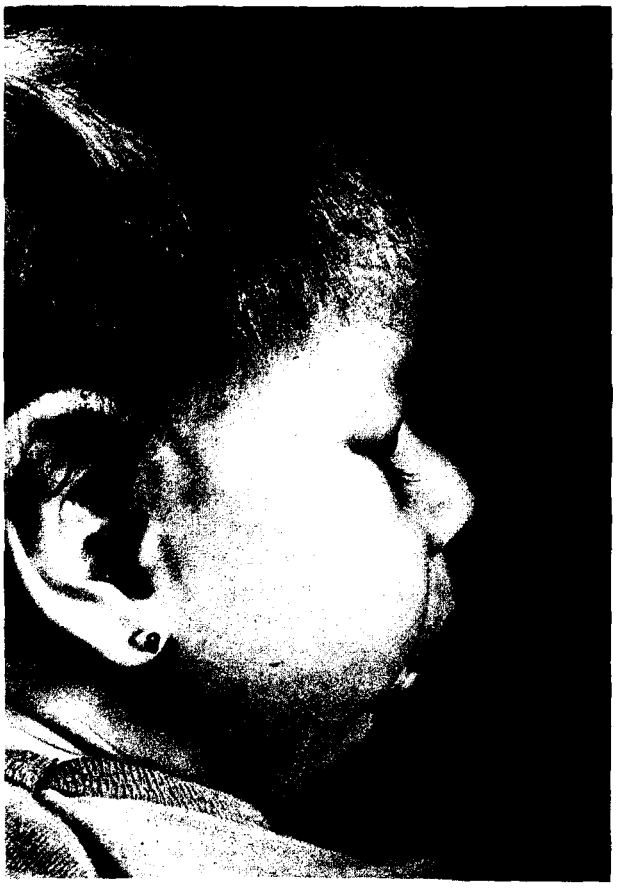

Fig. 1. Patient at 12 months of age demonstrating hypoplastic mandible.

Although congenital heart disease is frequently suspected in dyspneic growth-retarded patients with Pierre Robin syndrome, a search of the world's literature revealed only 37 cases in which congenital heart disease was proved [1, 3, 4, 6–8, 11–13, 16–20]. These cases, together with the present case, are summarized in the Table. The number of diagnoses (56) exceeds the number of cases (38) because multiple defects were present in several patients. Three of the 11 cases listed in the Table as ventricular septal defects were noted to have a biventricular or overriding aorta [3, 6, 12], suggesting that they might have developed into tetralogies of Fallot.

There are many additional reports in the literature of patients with Pierre Robin syndrome in whom congenital heart disease was clinically suspected. However, since the upper airway obstruction of Pierre Robin syndrome can produce cor pulmonale, cardiomegaly, pulmonary edema, and cyanosis in the absence of congenital heart disease [9, 10], diagnoses not substantiated by cardiac catheterization, surgery, or necropsy have been excluded for this review.

Table. Cardiac defects in 38 patients with Pierre Robin syndrome

Lesion	No. of Patients	Percentage
Ventricular septal defect	13	23.2
Patient ductus arteriosus	11	19.6
Atrial septal defect	9	16.1
Coarctation of aorta	6	10.7
Tetralogy of Fallot	6	10.7
Endocardial cushion defect	2	3.6
Hypoplastic left heart	2	3.6
Persistent left superior vena cava	2	3.6
Cor triloculare	1	1.8
Dextrocardia	1	1.8
Mitral stenosis	1	1.8
Pulmonary atresia with ventricular septal defect	1	1.8
Transposition	1	1.8
Total	56	100

Other major congenital anomalies are found in about half of the patients with Pierre Robin syndrome. In addition to congenital heart disease, deformities of the ears, eyes, and skeletal system are common [20].

There are numerous reports of the occurrence of Pierre Robin syndrome in siblings and in successive generations [2]. Most of these are compatible with a dominant mode of inheritance with incomplete penetrance.

Tube feeding, gastrostomy, tracheostomy, creating an artificial tongue tie, and maintaining the infant in a prone position with the back of the head suspended by means of an elastoplast cap are all accepted modes of therapy [3]. Whichever method is used, the importance of scrupulous nursing care cannot be overemphasized.

Although deaths in infancy from aspiration and respiratory tract infections are not rare, the long-term prognosis for patients not having additional uncorrectable anomalies is excellent. The mandible grows to a normal size. Hypoplastic mandibles in adults are not related to the Pierre Robin syndrome [4].

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