Familial Occurrence of Single Ventricle

Single ventricle, which accounts for approximately 2.5% of major congenital heart anomalies at necropsy (Fontana and Edwards, 1962; Neel, Shaw, and Schull, 1965; Bello, 1968), has been the subject of several recent reviews (Cruz and Miller, 1968; Lev et al., 1969), but to our knowledge the condition has never been reported to be familial. The purpose of this paper is to call attention to the occurrence of this anomaly in two members of a sibship, with the hope that with the accumulation of more data an accurate estimate of familial recurrence risk may be assigned and a mode of inheritance determined.

Case Report

The proband was the product of a normal, term pregnancy, birthweight 3260 g. At 8 weeks she was moderately cyanotic, weight was 4593 g, heart rate 140, blood pressure 90 mmHg by flush. There was a grade 1/6 systolic ejection murmur which was heard best along the left sternal border. There were palpable brachial and femoral pulses. The dermatoglyphs were normal.

Chromosome study was normal (46, XX), and other laboratory studies, including serum electrolytes, haemogram, urinalysis, and liver function studies showed no abnormalities.

Chest x-rays revealed an enlarged globular heart and diminished pulmonary vascularity. The electrocardiogram showed an abnormal conduction pathway suggestive of the Wolff-Parkinson-White syndrome.

At cardiac catheterization ventricular pressures obtained through the tricuspid valve and beneath the aortic valve were identical. Catheter position in the frontal projection indicated that the ascending aorta and aortic valve were posteriorly placed. These observations establish laevo or corrected transposition of the aorta. A venous catheter passed from the right atrium into the ventricle and injection of radio-opaque material revealed a large ventricular chamber and an aortic outflow chamber (Fig.).

Paint opacification of the pulmonary arteries was noted with the ventricular injection, and even fainter pulmonary artery opacification was seen with an aortic root injection. From these observations, plus the decreased pulmonary vasculature of the chest film, it was concluded that pulmonary valve stenosis was present. Cine studies of the left atrium revealed that it also emptied into the common ventricle. The final diagnosis was single ventricle with laevo (corrected) transposition and pulmonary stenosis.

The infant died at 7½ months, but necropsy was not permitted.

The parents have had one other child, a girl, who died at 4 days of age. Necropsy revealed the presence of a heart with a single ventricle and two atria. The pulmonary artery had vestigial valves but a normal diameter. The foramen ovale and the ductus arteriosus were patent. The right atrium was very dilated. There is no history of parental consanguinity, but three maternal first cousins of the proband were reported to have died of congenital heart disease.

Comment

Single ventricle as defined by Rahimtoola, Ongley and Swan (1966) is a ventricular chamber which receives blood from both atrioventricular valves. It is due to failure of development at the bulboventricular loop stage. At this time in the embryology of the heart, the bulbus cordis has moved to lie ventrally and to the right of the common ventricle. As the heart tube folds, the

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FIG.—Outlined cine print of ventricular injection in a lateral position. The catheter entered the ventricle from the right atrium. The ventricle was outlined and was made up of two parts: (B) the body of the ventricle and (O) an outflow chamber from which the aorta (A) arose. The vertebral bodies are outlined as landmarks.
bulboventricular spur is formed, projecting between the common ventricle and the bulbus cordis. Normally, this large spur is absorbed into the ventral wall of the ventricle and is reduced to the bulboatrial spur, while at the same time the bulbus cordis shifts and grows to the left to form most of the right ventricle. Concurrent with both the above events, the muscular portion of the interventricular septum is formed. It has been postulated that the haemodynamic consequences of a developmental arrest at the time of ventricular septation may in turn give rise to the various other findings which occur with the single ventricle such as rudimentary chamber, transposition, and corrected transposition of the great vessels (Harley, 1958).

Campbell, Reynolds, and Trounce (1953) reported that partial pulmonary obstruction improved prognosis. The proband, in contrast to her sib with a normal-sized pulmonary valve, lived 7 months, possibly because pulmonary stenosis limited the pulmonary blood flow.

Environmental factors such as drugs or viruses are known to cause congenital heart malformations, but in general these teratogens cause multiple system anomalies, and affect only one member of a sibship. In the present family the occurrence of two sibs with such strikingly similar, isolated, and rare heart malformations is strong evidence for genetic determination. The most likely modes of genetic transmission appear to be an autosomal recessive trait or a discontinuous polygenic trait.

Summary

Two sisters with single ventricle are presented as evidence for genetic determination of this malformation. Embryogenesis of single ventricle is reviewed.

References


Cruz, M. V. de la, and Miller, B. L. (1968). Double-inlet left ventricle. Circulation, 37, 249.


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Endocarditis in the Neonatal Period

Vegetations on the heart valves of newborn infants are rare at necropsy. Hudson (1965) in his textbook mentions the presence of non-bacterial vegetations on the heart valves in the neonatal period only in the context of Lambl's excrescences. In diseased valves, these contribute to the general roughening of the contact margin, and may build up as a granular and friable mass of platelets and fibrin. The underlying valve shows no cellular reaction and the whole lesion resembles the terminal or cachectic vegetations seen in the elderly and the degenerative verrucal endocarditis of Allen and Sirota (1944). While these lesions are usually papillary, some may be more warty and may vary from the size of a pinhead to several mm in diameter. Hudson does not recognize infective endocarditis in this age group, though three fairly well-documented cases can be found in the literature, as well as one rather incompletely recorded case (see Table). We here report a further case.

Case Report

The infant was the third child of a Rhesus positive mother aged 29 years, who developed pre-eclamptic toxoaemia at 37 weeks, and was delivered by lower segment caesarian section at 39 weeks. The mother had chronic bronchitis with poor respiratory function, but suffered no acute exacerbations during the pregnancy. Two previous pregnancies had terminated in forceps deliveries; the first infant (male, 2300 g) died aged 1 week from intracranial haemorrhage, the second (male, 4500 g) died during delivery. The present infant (male, 3133 g), who cried early, but then went limp and was resuscitated, appeared normal with a clear chest and normal heart sounds. By 24 hours, the infant had developed respiratory distress and began to have apnoeic attacks. Chest x-ray was consistent with hyaline membrane disease and the infant was maintained on intermittent positive pressure respiration with oxygen until death at 40 hours.

At necropsy the only significant gross findings were confined to the thorax. The lungs were fully expanded and a dull cherry red colour. There was no evidence of pulmonary embolism or infarction, and no broncho-
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