CASE REPORTS

Mitral Atresia and Occlusive Left Atrial Thrombus A Case with 11 Years of Survival

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Mitral atresia is a severe malformation, survival for more than one year being rare. Out of a total of about 160 published cases examined at necropsy, only 10 were found to have lived for more than a year; 9 of them had the great vessels transposed, the oldest patient being 22 years (Watson *et al.*, 1960; Fontana and Edwards, 1962; Eliot *et al.*, 1965; Gasul, Arcilla, and Lev, 1966; Meckel, 1922). Only one who lived 2 years (Eliot *et al.*, 1965) had the great vessels in the normal position. The relative frequency of occurrence of cases of mitral atresia with or without normally placed great vessels is 1:3 (Eliot *et al.*, 1965; Summerell *et al.*, 1968).

The following is a report of a patient with mitral atresia with normally placed great vessels who survived until the age of 11.

Case Report

An 11-year-old girl was referred (February 1964) for cardiac evaluation after an episode of "pneumonia". Cyanosis was first noted at 2 years of age and there was progressive dyspnoea on exertion.

Examination showed a girl 115 cm. in height, weighing 16 kg. There was moderate cyanosis and clubbing of the fingers. The pulse rate was 150/min., and the blood pressure 100/70 mm. Hg. The jugular veins were slightly distended; a prominence of the lower sternum and a forceful right ventricular parasternal systolic thrust were noted. On auscultation the second sound was single and loud. A grade 3/4 pansystolic murmur was heard loudest over the mid-precordium.

The chest x-ray films showed a moderate cardiomegaly with raised cardiac apex. The pulmonary artery was bulging, the left atrium was enlarged, and the vascularity of the lungs was increased.

The electrocardiogram showed first degree AV block, extreme axis deviation (-100°), right atrial enlargement, and a prominent R wave in V1, with Q waves in left

precordial leads of more than 2 mm., which was interpreted as indicating biventricular hypertrophy (Fig. 1).

Right heart catheterization revealed a pulmonary artery pressure of 90/40 mm. Hg, right ventricular pressure of 100/0 mm. Hg, and right atrial mean pressure of 8 mm. Hg. No satisfactory wedge pressure could be obtained. Peripheral saturation was 85 per cent and Hb 17.5 g./100 ml. A significant rise in the oxygen content in the right atrium and the rest of the right heart was detected. Angiography with right ventricular injection was unsatisfactory.

On second admission (August, 1966), she complained of transient episodes of dyspnoea, not related to effort, and recent left thoracic pain and haemoptysis. Examination showed increased cyanosis and jugular venous distension. The liver was moderately enlarged. There was atrial fibrillation, and an additional faint mid-diastolic murmur was heard occasionally over the praecordium. The chest x-ray film showed a rounded opacity in the left upper lobe, increased density of the hilar shadows, and Kerley B lines in the right costophrenic angle (Fig. 2).

A month later, on third admission, a new opacity in the right lower lobe was seen, the previous one having disappeared. During the period in hospital, the patient had several transient episodes of severe dyspnoea, cyanosis, hypotension, and lipothymia. No relation with changing posture was noticed, but this was not investigated. On the 15th day she died suddenly.

Necropsy. At necropsy the great vessels were normally placed, the right cavities and the pulmonary artery were dilated, and an apex was entirely formed by the left ventricle. The two venae cavae fed into the right atrium. The "foramen ovale" was closed. In the lower part of the atrial septum a small communication appeared in the location of the "ostium primum" defects (Fig. 3A). The atrium opened into the two ventricles by means of a wide five-leaflet "tricuspid" valve overriding a large ventricular septal defect of the "AV communis" type (Fig. 3A). The leaflets formed two funnels, each open-

FIG. 1.—Electrocardiogram showing extreme axis deviation, right atrial enlargement, prominent R wave in lead V1, and Q waves in left praecordium larger than 2 mm.

FIG. 2.—Chest x-ray films. (A)—Postero-anterior view. Note increased hilar markings, Kerley B lines in the right costophrenic angle, and a rounded opacity in the left upper lung. (B)—Right anterior oblique film showing enlargement of the left atrium.

ing into a ventricle and attached to the normal papillary muscles and the superior edge of the interventricular septum (Fig. 3D).

The pulmonary artery had a normal origin from the infundibulum of the right ventricle (Fig. 3C). Wall thickness was 0.8 cm.

The left atrium was distended and received the four pulmonary veins. It opened into the right atrium by way of the narrow septal defect. No mitral orifice or anomalous venous drainage was found. The cavity was nearly obliterated by a massive, partially organized thrombus attached to the posterior and lateral walls

(Fig. 3B). Its free portion was lying over the small defect leaving a narrow passage between its surface and the free walls. There was no doubt that the thrombus could modify the degree of obstruction to flow depending on the position of the heart.

The left ventricle and aorta appeared to be normal in size (Fig. 3D). The left ventricular wall thickness was 0.7 cm. There was no coarctation or persistent ductus.

The left main pulmonary artery was occupied by a large thrombus. Some smaller branches were occluded by old thromboemboli. Three recent infarctions were found in both lungs, the largest being 6 × 5 cm.

FIG. 3.—(A)—Right atrium (RA) and right ventricle (RV) opened showing tricuspid orifice and valvular apparatus. Note the atrial septal defect (D) in the “ostium primum” location, and the funnel-shaped left-sided leaflets entering the left ventricle (LV). (B)—View of opened left atrium (LA) showing the absence of the mitral valve, the atrial defect (D), and the section of the massive thrombus (T) partially removed. (C)—The opened right ventricle (RV) showing aorta (A) and pulmonary artery (P) normally placed. (D)—Internal view of the left ventricle (LV) showing origin of the aorta and attachment of the left-sided leaflets of the AV valve in the papillary muscles and superior edge of the interventricular septum (VS).

Discussion

Cases of mitral atresia with normally interrelated great vessels (Group I of the classification of Eliot *et al.*, 1965, and Summerell *et al.*, 1968) show a graded spectrum of severity according to the size of the aorta. Few patients with aortic atresia (Type Ia) live more than a month (Kanjuh, Eliot, and Edwards, 1965). One patient with aortic hypoplasia (Type Ib) lived 2 years, the only one in this group to survive a year. Patients with normal aorta (Type Ic) have a mean survival of 6 months (Watson *et al.*, 1960; Gasul *et al.*, 1966). Our patient is the oldest in the group and can be classified as a variety of Type Ic, presenting with the distinct anatomical features of an "AV communis" type of ventricular defect, with an overriding "tricuspid" valve and a left ventricle of good size. These characteristics probably bear some relation to the long survival. The additional presence of an "ostium primum" type of septal defect suggests that, as well as the ontogenic factors that determined the mitral atresia, there was an anomalous formation of the endocardial cushions.

These findings explain the peculiar electrocardiogram which shows evidence of significant left ventricular forces and an extreme axis deviation, as frequently seen in the cushion defects (Keith, Rowe, and Vlad, 1967).

The massive thrombus was a complication of the long-standing difficulty of the left atrium in emptying through the narrow septal defect. Contributory factors were heart failure, polycythaemia, and atrial fibrillation. We regard the thrombus as occlusive, implying that in addition to the progressive obliteration of the atrium there was the possibility of intermittent obstruction preventing its emptying, as suggested by necropsy findings and the episodes of paroxysmal dyspnoea and the variable diastolic murmur (Hahne and Climie, 1962).

The clinical manifestations of left atrial obstruction and thrombosis, apart from cyanosis, resemble those seen in mitral stenosis. The more significant findings for the diagnosis are (a) left atrial enlargement, rarely mentioned in the published reports, and related to the size of the atrial septal defect (Kjellberg *et al.*, 1959); (b) signs of pulmonary venous congestion; and (c) recurrent pulmonary infarction, perhaps the first reported in cyanotic congenital heart disease in the paediatric group.

Due to the rarity of peripheral venous thrombosis in this group of patients (Keith *et al.*, 1967), we suggest, with reasonable certainty, that the pulmonary infarction was due to emboli that parted from the left atrium and crossed into the right heart: they could be described as "paradoxical emboli". These pulmonary emboli would be the equivalent of the usual and expected systemic emboli of massive left atrial thrombus in acquired heart disease (Hahne and Climie, 1962).

In view of the anatomy, it is theoretically conceivable that this malformation can be corrected by closure of the defects and by implanting a prosthetic mitral valve before severe pulmonary arterial hypertension has developed.

Summary

An 11-year-old patient with mitral atresia, with normally interrelated great vessels, showing the distinct anatomical features of an "AV communis" type of ventricular septal defect and an overriding "tricuspid" valve, survived longer than any other reported patient without transposition of the great vessels.

The unique complications were an occlusive left atrial thrombus and recurrent pulmonary infarctions.

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