Isolated Congenital Mitral Stenosis

Report of Two Cases with Mitral Valvotomy in One


Of the 8 cases of isolated congenital mitral stenosis reported in the literature the authors add 2 cases personally observed. The postmortem findings in one of these patients is described; in the other, improvement was noted following surgical intervention. The literature is reviewed and the diagnostic criteria, including angiocardiographic findings, are described.

Isolated congenital mitral stenosis is an extremely rare condition. Only 8 cases have been reported in the literature, whereas 37 cases are recorded in association with other cardiac anomalies, such as patent ductus arteriosus, aortic stenosis, coarctation of the aorta, and aortic valve anomaly.1

This paper describes 2 female infants, 4 months and 3 months old, respectively, with isolated congenital mitral stenosis. The diagnosis was proved in one at autopsy; the second case was diagnosed clinically. The latter is alive and moderately well 16 months after mitral valvotomy.

The first case of isolated congenital mitral stenosis was recorded by Summons in 1906.2 In 1953, Bower and associates3 reported the first clinically proved case of isolated congenital mitral stenosis. The lesion was suspected after cardiac catheterization and confirmed by angiocardiography and operation.

The over-all prognosis is extremely poor; 4 cases died in the first year of life, 3 in the second, and 1 in the third year of life.

Since the advent of surgery for mitral stenosis, 3 patients with congenital mitral stenosis have been operated upon, 2 with isolated congenital mitral stenosis and 1 with a large patent ductus arteriosus. The first 2 patients both died postoperatively, one 30 hours4 and the other 6 weeks later.4 The third patient was alive 7 months after the operation.

Case Reports

Case 1

A girl, J. J., aged 3 months was the product of a full-term normal pregnancy and labor. At birth the weight was 6 lb. The mother noted grunting respiration soon after the child was born, and this symptom persisted until her first admission to hospital in severe congestive cardiac failure at the age of 3 months. Physical examination revealed a poorly nourished infant with marked dyspnea and substernal retraction. Slight cyanosis was present; there was no clubbing. The blood pressure was 110/65, pulse 160, and respiratory rate 50 per minute. The femoral pulses were easily palpable. The liver was enlarged 5 cm, below the right costal margin, nontender, and nonpulsatile. Jugular venous pressure was elevated to the angle of the jaw. The heart was enlarged and an apical systolic thrill and a right ventricular heave were noted. The anteroposterior diameter of the chest was increased. The first heart sound was loud and the second sound was accentuated. A rough grade 3 apical systolic murmur was present. It radiated to the base, but poorly to the axilla and back. No diastolic murmurs were noted. The breath sounds were normal. There was no peripheral edema or ascites.

Roentgenograms revealed cardiomegaly (cardiothoracic ratio 83 per cent) and marked increase in pulmonary vasculature. In the left anterior oblique film, the left main bronchus was displaced upwards by an enlarged left atrium. Enlargement of right ventricle and right atrium was also demonstrated (fig. 1). The electrocardiograph showed marked right ventricular hypertrophy with tall P waves in leads II and III (fig. 2). A diagnosis of congenital heart disease with pulmonary plethora was made. The features were not considered to be characteristic of a septal defect or patent ductus arteriosus. The patient responded well to digitalis therapy, with disappearance of the cyanosis, and was discharged undiagnosed.

Clinical Course. She remained fairly well on maintenance digitalis therapy until 1 month later, when she became dyspneic and cyanotic and was readmitted to hospital. The physical findings showed no change. Despite continuous oxygen and mercurial therapy she had several syncopal attacks and died 48 hours later.

Autopsy Findings. The heart was grossly enlarged (fig. 3). There was marked right ventricular hyper-
Fig. 1. Case 1. Teleroentgenogram shows gross cardiomegaly with increased pulmonary vasculature.

Fig. 2. The electrocardiogram of case 1 shows marked right atrial and right ventricular hypertrophy with probable left atrial hypertrophy in V1.

Fig. 3. Case 1. Cardiomegaly, marked thickening of the right ventricular musculature, and gross dilatation of the main pulmonary artery.

trophy, the wall measuring 1 cm. in diameter. The right atrium was dilated but not hypertrophied. The left atrium was also greatly enlarged (fig. 4). The mitral valve was severely stenosed (fig. 5) and the tricuspid valve incompetent. The atrial and ventricular septa were intact, and there was no patent ductus arteriosus or coarctation of the aorta. The pulmonary artery was dilated, and the lungs were slightly edematous. There was no macroscopic pulmonary arterial disease. The enlarged liver exhibited the nutmeg pattern of severe congestive failure. Microscopic sections of the heart demonstrated generalized fibroelastosis of the left atrial endocardium. There was no evidence of rheumatic inflammation in either myocardium or endocardium. Stains for collagen were negative. The lung was not sectioned.

Case 2
A female infant, B. P., was first seen at 3 weeks of age on September 7, 1954. The pregnancy and labor were normal and the birth weight was 6 lb. 14 oz. The complaints were difficulty with feeding, vomiting, failure to thrive, and excessive sweating. Physical examination revealed a dyspneic infant with profuse sweating and pronounced substernal retraction. The face and extremities were cyanosed. The blood pressure was 80/50, pulse 180, and respiratory rate 80 per minute. Femoral pulses were palpable. Jugular venous pressure was elevated to the angle of the jaw. The liver was enlarged 4 cm. below the right costal margin. Peripheral edema and ascites were not evident. The heart was enlarged clinically. There were no thrills; a vigorous systolic thrust and diastolic shock were palpable in the second and third left intercostal spaces. The first heart sound was loud. The pulmonic second sound was booming. At the apex a blowing systolic murmur, grade I to II, was heard. No diastolic murmurs...
were audible. After the heart rate had been slowed by digitalis the first heart sound was noted to be split as was the pulmonic second. A fourth heart sound became audible. The cyanosis disappeared and was replaced by pallor.

Roentgenograms showed cardiomegaly (cardiothoracic ratio 63 per cent) with right ventricular and right atrial enlargement, double cardiac density, and pulmonary plethora. Isolated left atrial enlargement was noted in the right anterior oblique film. The electrocardiogram showed marked right ventricular hypertrophy with tall P waves in lead II and V1 (fig. 6). Phonocardiography showed a systolic murmur, a presystolic gallop, and a fourth heart sound.

Cardiac catheterization was performed at 7 weeks of age under general anesthesia. The pressure findings are recorded in table 1. Unfortunately, all the blood samples were hemolyzed, except that from the axillary artery. Despite numerous attempts, the catheter did not enter the descending aorta. Even under rectal pentothal anesthesia the axillary artery sample showed a nearly normal oxygen content, which excluded a large right-to-left shunt. The marked increase in right ventricular pressure over systemic arterial pressure indicated an intact ventricular septum. The elevated pulmonary capillary pressure was compatible with an obstruction at the mitral valve. A tentative diagnosis of congenital isolated mitral stenosis was made.

Angiocardiography was carried out at 3 months of age. Under general anesthesia 12 ml of 70 per cent Diodrast was injected into the superior vena cava through a polythene catheter (figs. 7 and 8). The first film taken 3 seconds after the injection outlined the right atrium, right ventricle, and pulmonary artery. The subsequent films showed the dye returning from the lungs into a large left atrium and functioning left ventricle (fig. 7). The last film, (2 minutes after the injection), showed the left ventricle practically free from dye with the left atrium still full of contrast medium (fig. 8). The above features indicated a tight mitral stenosis.

In view of the presence of congestive cardiac failure at 3 weeks of age and ultimate poor prognosis, mitral valvotomy was performed on November 16, 1954. The patient was 3 months of age at this stage and weighed 7 lb. At thoracotomy the left lung appeared normal, but there was gross cardiac enlargement. The pulmonary artery felt exceptionally tense, but the aorta was small and of low tension. The left atrium, anterior to the pulmonary veins, was markedly enlarged. Through a left atrial incision the surgeon felt the mitral orifice to be rounded with neither irregularity, commissure, nor palpable regurgitation. The tip of the finger was pushed through the narrowed orifice with immediate, audible, splitting to admit the finger almost to the first knuckle. No regurgitation had ensued and it was estimated that the orifice, originally of a diameter of less than 3/4 cm., was opened to about 2 cm. The pulmonary artery pressure was then markedly reduced, the aortic pressure felt increased, and no systolic thrill was felt at the back of the atrium. The child's condition appeared excellent at the end of the procedure.

Clinical Course. The profuse sweating disappeared soon after the operation and she steadily gained weight, reaching 16 lb 2 oz. at 16 months of age. Three bouts of congestive cardiac failure occurred from January to August 1955, each associated with an acute bacterial infection. Digitalis, Diamox, and

Figs. 4 and 5. Left. Case 1. The very large left atrium is demonstrated. Right. Case 1. Demonstrates the stenosed mitral valve and marked thickening of the left atrial endocardium.
mercurial therapy was finally stopped in August 1955, and the patient has since been well. A recent bacterial infection did not result in congestive failure. At the time of writing she has progressed normally, and can walk and say a few words. The physical findings have altered. There is no evidence of congestive cardiac failure. The systolic thrust and diastolic shock in the pulmonary area are much less prominent and the pulmonary second sound, although loud, is no longer booming. The apical systolic murmur has increased in intensity to grade III. There are no diastolic murmurs and the fourth heart sound is no longer audible. The X-ray findings of the heart are unchanged. The electrocardiogram still shows marked right ventricular hypertrophy, but the P waves have decreased in height and the left ventricular R wave in V6 is more prominent. The operation has undoubtedly prolonged her life.

**Discussion**

The main features of all the cases of isolated congenital mitral stenosis reported in the literature together with our 2 cases are detailed in table 2. Symptoms frequently commence at birth but may be delayed until 2 years of age. Failure to thrive and dyspnea were noted soon after birth in our 2 patients and are common to most of the described cases. Congestive cardiac failure with cardiomegaly, apical systolic murmur, and accentuated pulmonic second sound are the most common physical signs. Persistent cyanosis is not a feature. Temporary cyanosis was present on admission in both our cases but disappeared after digitalization. X-ray examination reveals cardiomegaly, pulmonary plethora, left and usually right atrial enlargement together with right ventricular hypertrophy. The electrocardiogram shows marked right ventricular hypertrophy together with either left or right or combined atrial hypertrophy. Angiocardiography is diagnostic. The enlarged left atrium retains the dye for an abnormally long time because of the obstruction offered by the stenosed mitral valve. In our case 2 the contrast medium was still demonstrable in the left atrium 2 minutes after its injection into the axillary vein, yet the left ventricle was completely devoid of dye. Intracardiac or extracardiac shunts are not present. Cardiac catheter findings comprise elevated pulmonary artery and pulmonary capillary pressures.

Clinical diagnosis is suggested by the above symptoms and signs. Other forms of a cyanotic congenital heart disease may present similar findings. Patent ductus arteriosus, ventricular septal defect, including the Eisenmenger complex, and atrial septal defect are easily differentiated by cardiac catheterization. The presence of isolated left atrial enlargement excludes primary pulmonary hypertension. In
Figs. 7 and 8. Left. Case 2. Angiocardiogram 20 seconds after injection shows the dye returning from the lungs into a large left atrium and definite left ventricle. Right. Case 2. Film taken 2 minutes after injection shows the left ventricle practically free from the dye with the left atrium still full of contrast medium.

### Table 2.—The Salient Symptoms, Signs, and Investigations in Ten Cases of Isolated Congenital Mitral Stenosis

<table>
<thead>
<tr>
<th>No., age and reference no.</th>
<th>Onset of symptoms (months)</th>
<th>Symptoms</th>
<th>Signs</th>
<th>X-ray</th>
<th>ECG</th>
<th>Angiocardiogram</th>
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<tr>
<td></td>
<td></td>
<td>Failure to thrive</td>
<td>Cough</td>
<td>Dyspnea</td>
<td>Cyanotic attacks</td>
<td>Congestive failure</td>
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<td>1. Birth</td>
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<td>2. Birth</td>
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<td>3. Birth</td>
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* Our 2 cases.
our case 2, the diagnostic possibility of congenital mitral stenosis was suggested by the catheter findings of severe pulmonary hypertension and raised pulmonary capillary pressure in the absence of intracardiac or extracardiac shunts. Angiocardiography was considered mandatory and revealed the presence of severe mitral stenosis.

The pathologic changes in the mitral valve are uniform. It is usually thickened, hard, nodular, and white with a pearly semitranslucent appearance. It may be semicartilaginous in consistency. The chordae tendineae are thickened, shortened, and fused. The valve aperture is extremely small even during life: the diameters in the 3 operated cases were 0.5

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**Table 3.—Catheterization and Operative Data**

<table>
<thead>
<tr>
<th>No.</th>
<th>Right-to-left shunt</th>
<th>Left-to-right shunt</th>
<th>Pulmonary artery pressure (mm. Hg)</th>
<th>Elevated pulmonary capillary pressure (mm. Hg)</th>
<th>Age at operation (months)</th>
<th>Size of valve</th>
<th>Result</th>
<th>Age at death (month)</th>
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<td>6</td>
<td>Abs.†</td>
<td>Abs.</td>
<td>67</td>
<td></td>
<td>9</td>
<td>0.5 cm.</td>
<td>Died 36 hours postop.</td>
<td>19</td>
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<tr>
<td>7</td>
<td>Abs.</td>
<td>Abs.</td>
<td>50/18</td>
<td>25/13</td>
<td>4</td>
<td>1.0 cm.</td>
<td>Died 6 weeks postop.</td>
<td>24</td>
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<tr>
<td>9.*</td>
<td>Abs.</td>
<td>Abs.</td>
<td>100/48</td>
<td>20 mm. (syst.)</td>
<td>3</td>
<td>0.5 cm.</td>
<td>Alive 16 months postop.</td>
<td>4</td>
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</table>

† Absent.

* Our 2 cases.

**Figs. 9 and 10.** Left. Case 2. Section of the left lingula under low power showing cellular infiltrate and thickened media of the pulmonary arterioles. Right. Case 2. High power showing thickened media and prominent internal and external elastic laminae of the pulmonary arterioles. There is no evidence of endarteritis.
cm. in 2 and 1 cm. in 1. The endocardium of the left atrium and right ventricle frequently shows thickening due to fibroelastosis. The capillaries in the lung are usually distended and the alveoli may contain heart-failure cells characteristically seen in cases of severe acquired mitral stenosis. Medial hypertrophy of the pulmonary arterioles has been described in 7 cases of congenital mitral stenosis. Three cases exhibited persistence of the fetal state of the arterioles. No single instance of intimal thickening of the pulmonary vessels has been recorded. The biopsy of the left lingula removed at operation in our case 2 showed similar findings (figs. 9 and 10). However, we are unable to account for the striking cellular infiltrate of the peribronchiolar connective tissue, respiratory bronchioles, and alveolar ducts.

In view of the poor prognosis—8 out of 9 children dying before the age of 2 years—the question of surgery warrants consideration. In rheumatic heart disease one is dealing with acquired pathology imposed upon a normal valve, whereas in congenital mitral stenosis the valve structure is congenitally distorted. In some cases of acquired mitral stenosis valvotomy results in restoration of normal function by the separation of the fused segments of the valve cusps, the valves themselves being otherwise relatively normal; on the other hand, congenital mitral stenosis is the result of a developmental malformation of the entire valve and commissurotomy will do no more than enlarge the stenosed aperture without restoring valve function. Hence the stenosis must be partly replaced by incompetence, and both structure and function remain abnormal. The operation is indicated only in the presence of a functioning left ventricle. Since her operation, our patient has had 3 bouts of congestive cardiac failure associated with bacterial infection. Between these bouts she was in mild failure and required maintenance digitalis and mercurial or Diamox therapy. For the past 6 months all treatment has been discontinued and she has remained well. We feel that valvotomy has definitely prolonged her life.

Summary

Two cases of isolated congenital mitral stenosis are described, both of which presented with congestive cardiac failure before the age of 3 months. The physical examination was inconclusive. The diagnostic possibility of congenital mitral stenosis was suggested by the catheter findings of severe pulmonary hypertension and raised pulmonary capillary pressure in the absence of intracardiac or extracardiac shunts. Angiocardiography was diagnostic. The left atrium in 1 case remaining full of contrast medium after the left ventricle had emptied.

One patient was subjected to surgery at the age of 3 months. She is alive 16 months after mitral valvotomy.

Acknowledgment

The authors would like to thank Dr. I. Webster and Dr. W. J. Pepler of the South African Institute of Medical Research for the pathologic findings in the 2 cases. They are also indebted to Drs. B. Van Lingen, J. Kaye, and other members of the Cardiac Clinic of the Johannesburg Hospital for their assistance in the cardiac catheterization and angiocardiography of our second case. Mr. A. M. Shewitz was responsible for the excellent photographs.

Summario in Interlingua

Es describite duo casus de isolate congenite stenosis mitral. Ambes esseva presentate con congestive disfallimento cardiaco ante le etate de 3 menses. Le examine physic esseva indecise. Le possibilitate de congenite stenosis mitral esseva suggerite per le constatation catheterica de sever hypertension pulmonary e de elevate pressiones pulmono-capillari in le absentia de derivationes intra- o extracardiac. Le diagnose esseva establite definitivamente per medio de angiogiardiographia. In un caso le atri sinistre remaneva plen de substantia de contrasto post que le ventriculo sinistre esseva vacuate.

Un del patientes esseva subjicite a un opera- tion chirurgie al etate de 3 menses. Illa vive 16 menses post valvotomia mitral.
REFERENCES


Chronic discoid lupus erythematosus has been regarded as primarily a skin disease with rare systemic manifestations. In order to determine the truth of this statement the authors studied a series of 41 patients with chronic discoid lupus erythematosus. The patients were divided into 2 groups: the localized discoid form, with skin lesions above the chin, and the generalized discoid form with cutaneous involvement on the face and elsewhere. Sixteen of the 26 patients (62 per cent) of the localized discoid group had evidence at some time in the course of their illness of arthritis, fever, Raynaud's phenomenon, pleurisy, or other systemic changes by history and physical examination alone. Fourteen of the 15 cases of generalized discoid disease had such changes. If, in addition, laboratory abnormalities such as leukopenia, elevated sedimentation rate, hyperglobulinemia, or abnormal flocculation tests were considered, then 24 of the 26 with localized discoid disease and all 15 of the generalized discoid group showed such changes. Therefore, there was evidence of systemic involvement in 96 per cent of this group of patients with chronic discoid lupus. Three different modes of onset of discoid lupus were found. Thirty-three patients (72 per cent) had cutaneous changes initially, followed, in 45 per cent of this group, by rheumatoid-like arthritis. Seven patients had rheumatoid arthritis prior to the appearance of discoid lesions. The classification of lupus erythematosus is an arbitrary one. There are many transitions between the types. In this report it is shown that discoid lupus, from its inception, is a systemic disorder that is a variant of the more malignant acute disseminated form. The "benign"-appearing cutaneous lesion may be a herald of advanced systemic manifestations which may be present at the same time or at a later date, when the skin changes have healed. Therefore, all patients with discoid lupus erythematosus should have a thorough general medical survey. The form of therapy instituted depends entirely upon the extent of the disease.

Wendkos
Isolated Congenital Mitral Stenosis: Report of Two Cases with Mitral Valvotomy in One
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Circulation. 1957;15:358-365
doi: 10.1161/01.CIR.15.3.358

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