Endomyocardial Anomalies of the Left Heart

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We discuss miscellaneous anomalies of the endocardium and myocardium of the left ventricle together with their diagnostic features and choices of medical or surgical management. Though each lesion individually is uncommon, the group of malformations is encountered sufficiently often to keep physicians aware of the challenge of diagnosis and proper treatment.

Malformations of the endomyocardium of the left heart are far less common than those affecting the valves or septa; yet they are encountered often enough to constitute a challenge to the physician in diagnosis and proper management. They are more often found in infants than in children or adults. Murmurs are absent or inconspicuous. The condition is apt to be recognized when an infant suddenly and expectedly goes into cardiac failure or when a roentgenogram of the chest obtained, perhaps for an unrelated reason, gives indications of it.

Endocardial fibroelastosis is the most common of these malformations. The thickened, restrictive endocardium lines either a contracted (Fig 1) or, more often, a dilated left ventricle. In the former, the signs and symptoms are those of pulmonary venous obstruction, pulmonary hypertension, and right-sided cardiac failure. In the latter, the manifestations are those of left ventricular failure. In either form, the thickening of the endocardium may extend onto the aortic or mitral valve (Fig 1).

Endocardial fibroelastosis has been found in postmortem examination of the fetus. In liveborn infants, the effects of the contracted type may be manifest in the first hours or days of life. In the dilated form, symptoms more often present only after a few weeks or months. Such an occurrence is illustrated in Figure 2, which compares the chest roentgenogram on the first day of life of a prematurely born baby with that taken 2½ months later on admission, almost moribund, in shock and cardiac failure. Death occurred a few minutes after admission, before emergency measures to treat the cardiac failure had time for effect.

The electrocardiogram in the dilated form shows left ventricular hypertrophy and "strain" (ST-T wave abnormalities) (Fig 3) with evidence sometimes of atrial enlargement. In the contracted form, the evidence of the hypertrophic left ventricular wall may be masked by the right ventricular hypertrophy.

Cardiac diagnostic studies, undertaken after control of cardiac failure, show, on catheterization, elevation of the end-diastolic pressure in the left ventricle and of the pressures in the more proximal chambers and vessels, secondary to left ventricular failure. Selective angiography shows a diminished range of contraction of the thick-walled left ventricle. On cineangiography, contrast medium can be seen to swirl in the left ventricle.

Diagnosis of the condition during life is based on the electrocardiogram and findings as described at cardiac catheterization with contrast studies, together with the exclusion by appropriate tests of other forms of endomyocardial disorders and of

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other causes of left ventricular failure, such as coarctation of the aorta and hypertension.

Treatment is directed toward the signs of cardiac failure; there is no specific treatment for the condition itself. In the contracted form, this intensive medical management does not usually succeed for long, but the prognosis for the dilated form is better.1 Many survive infancy, and as the child grows more rapidly than does the enlarged heart, the cardiothoracic ratio on chest roentgenograms becomes normal. Until that happens, and the electrocardiogram also improves, digitalization should be continued. Thereafter, the children may be allowed full activity but should continue under regular medical supervision since their ultimate prognosis is unknown. Perhaps they form the group who present as adults with a big heart, cardiac failure, and at autopsy, endocardial fibroelastosis.2 One of our patients, managed medically in infancy, was asymptomatic, off digitalis from the age of two years, when his chest roentgenogram and electrocardiogram became normal. However, cardiac decompensation returned at age eight, and he died in intractable failure and with cardiac arrhythmias, chiefly atrial flutter. At postmortem examination, in addition to the endocardial fibroelastosis, the left ventricle showed extensive scarring and fibrosis extending from the endocardial surface.

Nonobstructive cardiomyopathy or idiopathic hypertrophy of the heart is much less common than the previous condition but has some similar features.3 The hypertrophy can be so massive as to restrict the cavity of the left ventricle and obstruct pulmonary venous drainage, in which situation it resembles the contracted form of endocardial fibroelastosis. The electrocardiogram may show only the evidence of the right ventricular hypertrophy that is a sequel to the left-sided obstruction and, curiously, may show no evidence of left ventricular hypertrophy.

When the hypertrophied wall permits more nearly normal filling and emptying of the left ventricle, the condition can resemble the dilated form
of endocardial fibroelastosis, except that in the electrocardiogram, ST segments and T waves are usually not abnormal. The angiocardiogram demonstrates a thick-walled left ventricle which contracts and relaxes and may show irregularities of contour of the cavity if eccentrically hypertrophied muscle bulges into the cavity. Radioisotope scanning over the ventricle has been reported to disclose a pattern resembling a tumor or abnormal area of myocardium.4

Occasionally idiopathic hypertrophy merges with the condition of hypertrophic subaortic stenosis, for the thickened myocardium of the free wall and the ventricular septum may approximate one another during systole so as to obstruct left ventricular outflow. This situation can lead to further hypertrophy.

Neither the cause nor the cure for idiopathic hypertrophy is known. If it has reached a stage of hypertrophic subaortic stenosis, cardiac surgery to incise the hypertrophied septum may afford relief of the obstructive component of the condition.

Glycogen-storage disease of the myocardium5 is a rare condition in which glycogen is stored in the heart but also throughout the body in a generalized fashion, in skeletal muscle, tongue, liver and kidneys. The disorder has been shown to be due to a defect in the enzyme alpha-1,4-glucosidase.6 The cardiac form of generalized glycogenosis, known as Pompe’s disease, produces symptoms of cardiac failure early in infancy and usually, death before the first birthday. The heart is markedly enlarged. There may be no cardiac murmurs, but this is a condition in which hypertrophic subaortic stenosis can occur so that a murmur of aortic stenosis may be heard.5

The electrocardiogram is of great diagnostic value (Fig 4). In addition to marked increase in voltage of the QRS complex, greater than in almost any other type of left ventricular hypertrophy, there is shortening of the PR interval. The tracing may be confused with that of Wolff-Parkinson-White syndrome. T waves may be normal in direction or inverted in leads V5 and 6, aVL, and 2 and 3.

The combination of these findings in the setting of a hypotonic, poorly nourished infant with large tongue strongly suggests the diagnosis of cardiac glycogenosis. The diagnosis can be confirmed by a biopsy of skeletal muscle, stained for glycogen. The swollen, distorted skeletal muscle filled with glycogen is similar to that which will be found in the heart at postmortem examination.

Cardiac catheterization and angiocardiography can measure the degree of cardiac impairment and evaluate the existence of accompanying hypertrophic subaortic stenosis.

No treatment is known. Response to digitalis and anticongestive measures is only temporary. Now that the specific enzyme deficiency has been identified, it may in the future be possible to use replacement therapy of some form, but at present the prognosis is hopeless.

Cardiac tumors of the myocardium are usually rhabdomyoma(ta), or rhabdomyobroma, or rhabdomyobrosarcoma. The condition is recognized because of several situations that can occur: (1) the tumor obstructs the flow of blood through a cardiac chamber or valve, or coronary artery;7 (2) it interferes with proper function of the myocardium so that the heart fails; (3) the strategic location of the tumor interferes with the conduction system, producing heart block or causing an arrhythmia; (4) it deforms the contour of the heart on chest roentgenogram; or (5) fragments of tumor embolize to the periphery or other organs. The next cases, from our experience with cardiac tumors in the pediatric age group, illustrate these points.

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2. The heart.

I. In the newborn period, heart failure or sudden death in the newborn.

II. In another patient, followed from infancy till her death in chronic cardiac failure at age eight years, we suspected the diagnosis of rhabdomyomatosis because of electrocardiographic findings: changing atrioventricular block and the appearance of episodes of atrial flutter (Fig 6). When a rash appeared over the bridge of her nose around the age of two years, we made the diagnosis of adenoma sebaceum and searched the eyegrounds for the plaque-like lesions of tuberous sclerosis, the condition with which rhabdomyomata and adenoma sebaceum are associated. When the typical eyeground changes were found, the diagnosis of cardiac rhabdomyomatosis became more confident.

For the next six years the cardiac failure and arrhythmias were controlled with digitalis, without causing progression of the partial heart block. The heart enlarged progressively over this period (Fig 7). She died during a respiratory illness. Postmortem examination confirmed the diagnosis of rhabdomyomatosis and tuberous sclerosis.

Another patient, a young baby with cardiac failure, had an electrocardiogram that suggested the diagnosis of cardiac tumor interfering with the anterior descending coronary artery. The tracing resembled an anterior myocardial infarction. That tumor was a rhabdomyosarcoma.

A third form of tumor of heart muscle led to a more fortunate outcome in a young boy. The sudden, unexpected onset of cardiac failure at age two years was due to ventricular tachycardia. A bulge at the apex of the heart suggested a ventricular tumor, whose presence was confirmed by selective left ventricular angiocardiography. At operation, an encapsulated rhabdomyofibroma was successfully removed.

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removed. He has been well in the four years of postoperative follow-up.

Atrial myxoma is the most common intracardiac tumor, and though it has been reported chiefly in adults suspected of having mitral stenosis, it does also occur in children. The myxomatous tumor arises from a stalk attached to the atrial septum in the region of the fossa ovalis and protrudes either into the left or right atrium so that it obstructs either pulmonary or systemic venous drainage.

The diagnosis of left atrial myxoma was suspected in one child at our hospital because of a changing diastolic murmur at the apex of the heart and episodes of peripheral embolic phenomena which suggested tumor emboli. The diagnosis of a left atrial tumor was confirmed by the filling defect in the left atrium on angiocardiography. Although that patient lived and died before the advent of extracorporeal circulation, others now can be cured when the diagnosis is made and the tumor excised at open heart surgery.

Diverticulum of the left ventricle is exceedingly rare. We have not seen an example of the elongated diverticulum which protrudes as a pulsatile mass in the upper abdomen. We have, however, seen one girl, admitted because of painless hematuria, who had an abnormal electrocardiogram with an unusual degree of left axis deviation and a cardiac silhouette with slight irregularities of the left ventricular border. On intravenous (Fig 9) and selective left ventricular angiocardiography, these areas were shown to be due to multiple diverticula of the left ventricle. Perhaps clots within these diverticula were the source of renal emboli that resulted in painless hematuria.

Anomalous origin of the left coronary artery from the pulmonary artery is less rare than was once thought when it was recognized only in the form...
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Figure 9. Supravalvular aortogram in child with anomalous origin of the left coronary artery from the pulmonary artery. In the film on the left, injection into the root of the aorta resulted in opacification of the right coronary artery only. In the film on the right, delicate, opacified intercoronary collateral vessels are filled and then in retrograde fashion, the left coronary artery (arrow).

Described by Bland, White, and Garland. That form depicted a fretful infant with anginal episodes and in cardiac failure with a greatly dilated left ventricle and an electrocardiogram characteristic of myocardial infarction and aneurysm or both. The diagnosis, suspected clinically, could be confirmed by selective injection of contrast medium into the base of the aorta. There would visualize a single right coronary artery with filling of smaller vessels and then, through collateral channels, a left coronary artery that communicated with the pulmonary artery (Fig 9). In a baby with extreme coronary insufficiency, little could be accomplished with medical or surgical management, and the diagnosis was regularly confirmed at postmortem examination.

Not all babies born with this anomaly were so severely affected, however, and the other two forms of the anomaly came to be recognized in those surviving infancy: one, the child with an apical systolic murmur of mitral insufficiency and an electrocardiogram of left ventricular myocardial damage and the other, the individual with a soft, continuous murmur over the precordium of a coronary arteriovenous fistula.

Survival was possible because of the development of adequate intercoronary collateral circulation over the left ventricle. The murmur of mitral insufficiency resulted from the anoxic damage to the papillary muscles, with scarring and stiffening or shortening so that the mitral leaflets could not approximate normally. The continuous murmur of a coronary arteriovenous fistula resulted from establishment of large or rich anastomotic channels between the normally arising right coronary artery and the anomalous left coronary so that an arteriovenous fistula developed between aortic blood, leaving via the right coronary artery and communicating via collateral pathways and the left coronary artery with venous blood in the pulmonary artery.

Diagnosis in either situation depends upon thinking of the possibility of anomalous origin of the left coronary artery as an explanation for the findings and then demonstrating, by cardiac catheterization and selective injection of contrast medium into the pulmonary artery and into the aorta, the abnormal connections and function described above.

Prognosis for these two forms is not yet known. The two oldest patients in our experience with these two forms will be described briefly. One child, with mitral insufficiency, referred to us at age ten years after she had been asymptomatic from the second year of life, died suddenly while play-
ing. Another of our patients who functions with a small coronary arteriovenous fistula was asymptomatic at age 15 years when she under went cardiac diagnostic studies because of the continuous murmur.

Ideal treatment of patients with anomalous origin of the left coronary artery would seem to be severance of the abnormal communication with the pulmonary artery and anastomosis directly or through a prosthesis to the ascending aorta before sequelae of inadequate coronary perfusion of the left ventricle and papillary muscles become severe or irreparable (if that could be judged in other than the extremely ill, moribund young baby where damage is probably already too extensive for help). Such an operation could improve the natural history of those able to survive infancy only if it can be demonstrated that the anastomosis of such a small, thin-walled vessel as the anomalous left coronary artery would remain patent and would grow with the child. This has yet to be demonstrated, but an early report by Nora and others18 appears hopeful.

Whether ligation of the anomalous artery at its source offers an improvement over the natural history of those who can develop their own coronary collateral circulation is not clear. The child has demonstrated he has coronary circulation adequate for survival and would not appear to need this operation if there is reasonable expectation for a better one. In the young infant, judgment is more difficult, but we believe that only if the infant fails to improve on medical management with digitalization, should he be considered a candidate for ligating the anomalous vessel. For the individual with the continuous murmur, it is unlikely that the arteriovenous runoff contributes much to inadequate perfusion of the left ventricle, since patients with large coronary arteriovenous fistulae as the only malformation do not have evidence of myocardial ischemia.

The syndrome of apical systolic click, late systolic murmur and abnormal T waves14-18 is the last of the endomyocardial conditions to be discussed. It is the most recently recognized of this group and may prove to be the most common. Since we have observed it in children without a history to suggest rheumatic fever or myocarditis, we presume it to be congenital rather than acquired.

It is recognized by characteristic auscultatory and electrocardiographic features. At the apex of the heart are heard a systolic click, usually in midsystole, and a late systolic murmur. The electrocardiogram may vary from time to time in the same individual, but on all examinations in our patients, there is some abnormality of T waves, with inversion or flattening in leads V5-6, aVF, 2 and 3. U waves may be prominent. These findings are noted in asymptomatic individuals, almost always females, with normal cardiac series of chest roentgenograms.

Development of the diagnostic techniques of intracardiac phonocardiography and left ventricular angiocardiology led to the explanation of these findings. The late systolic murmur is due to a mild degree of mitral regurgitation occurring late in systole as a mitral leaflet, usually the posterior, balloons into the left atrium. The systolic click is thought to be due to the sudden tautening of slack chordae tendineae in midsystole.

The reasons why the chordae become slack, then taut, but too late to permit proper coaptation of the mitral leaflets in the latter part of systole has been found in six young women whom we have studied by biplane selective left ventricular angiocardiology. An abnormal contraction develops in the floor of the left ventricle midway through systole and disappears in diastole.

Our interest in this condition began in 1952 when we saw our first three girls and recognized that they had identical findings not adequately explained. Our attention was directed to the left ventricle when two of them were studied first by intravenous angiocardiology. During systole, the left ventricle in the frontal view seemed to partition into two chambers but became normal in contour with each diastole. Later, selective left ventricular angiocardiology in these two individuals and in four others with the syndrome showed in lateral and oblique views a bulging inward at midsystole of the posteroinferior aspect of the floor of the left ventricle. In the frontal view, this bulge appeared to separate the ventricle into two chambers. The posterior papillary muscle attaches in this area of the left ventricle, and a bulging inward of the floor of the ventricle could displace the papillary muscle, allowing it to slacken and permitting the attached mitral leaflet to prolapse late in systole into the left atrium so that mitral regurgitation occurs.

The electrocardiographic abnormalities are consistent with coronary ischemic changes in the posteroinferior aspect of the left ventricle. Perhaps the abnormal contraction interferes with regional coronary arterial perfusion there. The course and distribution of major coronary arteries have been shown to be normal.

The next question, why does the left ventricle
contract in this manner, is not answered. Fortunately for the patients who have the syndrome, the condition appears benign so that neither operative nor postmortem findings are available to clarify this point. The electrocardiogram offers no clue, for QRS duration and form are normal and do not permit an answer to speculation about a disturbed time sequence of left ventricular depolarization.

The six patients have continued asymptomatic in periods of follow-up that range from four to 15 years. Thus far, into young adulthood, their prognosis is good, and from reports appearing in the literature in increasing numbers, the prognosis is good into later adulthood. The only medical management that is indicated is penicillin prophylaxis at times of predictable risk of endocarditis.

Differential diagnosis of these endomyocardial anomalies of the left heart must be made, one from the other, and also from the acquired conditions such as myocarditis and myocardiopathies, as well as from those left ventricular abnormalities that develop in other syndromes, such as Friedreich's ataxia, muscular dystrophy, and the Hurler syndrome. In addition, the form of endocardial fibroelastosis and of idiopathic hypertrophy that severely constricts the chamber of the left ventricle requires differentiation from other forms of the hypoplastic left heart syndrome.

REFERENCES


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