

The surgical treatment of restrictive cardiomyopathy in pseudoxanthoma elasticum

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SUMMARY A patient with pseudoxanthoma elasticum presented in pulmonary oedema with restrictive left ventricular cardiomyopathy caused by calcified endocardial bands that were confirmed on echocardiography and at catheterisation. The bands were resected as far as possible and the involved mitral valve was replaced by a heterograft. A year later calcification of the heterograft forced its replacement by a St Jude prosthesis. Relief of symptoms has been good in the medium term.

Case report

A 27 year old white man, known to have pseudoxanthoma elasticum, presented with a rapidly progressive nine month history of exertional dyspnoea, orthopnoea, paroxysmal nocturnal dyspnoea, and the striking symptom of exertional haemoptysis. Over the nine month period his exercise tolerance had decreased from that of an active amateur footballer to being unable to walk more than one hundred yards on the flat.

Physical examination showed the classic skin lesions of pseudoxanthoma elasticum—yellow pseudoxanthomatous plaques, mainly in the neck region. There was evidence of congestive cardiac failure, with a sinus tachycardia, a loud left ventricular third heart sound, bilateral basal crepitations, jugular venous pressure raised 3-4 cm with a deep “y” descent, an enlarged pulsatile liver, and oedema of the legs. The electrocardiogram showed no abnormality and in particular the QRS complexes and T waves were normal. The chest x ray showed considerable calcification within the cardiac outline—in particular in the region of the left ventricle (figure). The classic appearances of pulmonary oedema were present. Cross sectional and M mode echocardiography showed several abnormal features (table 1). The left ventricular endocardium was markedly thickened, particularly in the posterior wall region. There was “flattening” of the left ven-

tricular posterior wall diastolic motion, although this could not be assessed quantitatively. There appeared to be diastolic restriction of left ventricular filling. The mitral valve E-F slope was at the lower limit of normal and mitral valve excursion was reduced. Left ventricular systolic function appeared to be normal, as shown by a normal ejection fraction and left ventricular fractional shortening. Left ventricular systolic and end diastolic internal dimension were within normal limits. The left atrium was markedly enlarged, in keeping with restriction of diastolic filling of the left ventricle in the absence of mitral stenosis. There was minor prolapse of the anterior mitral leaflet. The pericardium appeared to be of normal thickness. The right ventricle appeared to be normal.

The clinical and echocardiographic findings suggested a diagnosis of a restrictive left ventricular cardiomyopathy secondary to pseudoxanthoma elasticum. To obtain a definitive diagnosis, cardiac catheterisation was undertaken. Table 2 gives the right and left heart pressures obtained before angiography. The left ventricular end diastolic pressure was markedly raised at 25-30 mm Hg. The pulmonary artery and right ventricular pressures were also raised, although the right ventricular end diastolic pressure was normal. The pulmonary artery wedge pressure was raised with considerable “x” and “y” descents. Right atrial pressures were normal. There were no valve gradients. Left ventricular angiography showed a heavily calcified left ventricular endocardium with overall reduction in left ventricular contractility and a prolonged diastolic phase. There was mild mitral regurgitation. The

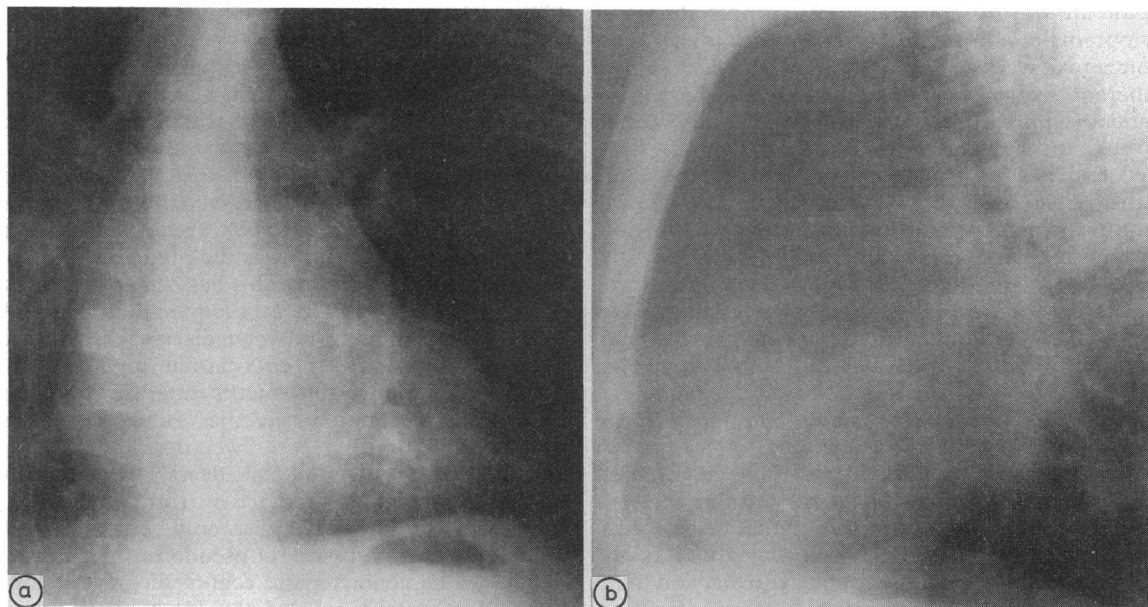


Fig Chest x rays from posterior-anterior (a) and lateral views (b) showing calcification in the region of the left ventricle.

right ventricle contracted normally. Coronary arteriography revealed a solitary stenosis of 40% beyond the first septal branch of the left anterior descending coronary artery. These findings confirmed our diagnosis of a restrictive cardiomyopathy solely affecting the left ventricle. The only cause appeared to be the extensive, but discrete, bands of endocardial calcification extending into the myocardium which were theoretically amenable to resection. In view of his age and rapidly deteriorating condition it was decided to attempt surgical resection.

At operation the lining of the left ventricle was thickened, fibrous, and mainly calcified, although there seemed to be three distinct ridges of calcified tissue along the posterior wall; the last of these was particularly restricting dilatation of the left ventri-

cle. The calcification was friable and went deeply into the myocardium. The mitral valve leaflets were normal, but the chordae were inserted into calcified papillary muscles. The calcification in the posterior wall and to some extent in the two lateral walls was excised as far as possible.

The mitral valve was replaced with a size 31 Carpentier-Edwards heterograft in view of the papillary muscle involvement. The coronary artery lesion was not considered to be haemodynamically important.

After operation his general condition improved

Table 1 Echocardiographic features before operation

Feature		Normal limits
Mitral valve E-F slope	53	(50-120 mm/s)
Mitral valve excursion	1.7	(2.0-2.5 cm)
Aortic root diameter	2.9	(2.0-3.7 cm)
Aortic valve cusp separation	2.0	(1.6-2.6 cm)
LA internal dimension	5.9	(1.9-3.8 cm)
Septal thickness (diastole)	1.2	(0.7-1.1 cm)
Posterior LV wall thickness (diastole)	1.8	(0.7-1.1 cm)
LV internal dimension (systolic)	2.6	(2.5-4.1 cm)
LV internal dimension (end diastolic)	4.2	(3.5-5.6 cm)
Cubed formula LV stroke volume	57	(55-95 ml)
Ejection fraction	77	(65-75%)
LV fractional shortening	0.38	(0.28-0.41)

LA, left atrium; LV, left ventricle.

Table 2 Catheter findings before and after operation

Vessel or chamber	Pressure (mm Hg)	Mean or end diastolic pressure
<i>Before operation</i>		
Right atrium	a = 2.5 v = 0 x = -5 y = -1	Mean = -1.5
Right ventricle	90/0	EDP 2-5
Pulmonary artery	90/30	Mean = 50
PAWP	a = 30 v = 35 x = 15 y = 10	Mean = 20
Left ventricle	110/0	EDP 23-30
Aorta	110/60	Mean = 85
<i>After operation</i>		
Right atrium	a = -1 v = 1 x = -3 y = -3	Mean = -1
Right ventricle	40/-6	
Pulmonary artery	32/12	Mean = 20
PAWP	a = 10 v = 11 x = 9 y = 8	Mean = 10
Left ventricle	120/-5	EDP 10
Aorta	120/70	

PAWP, pulmonary artery wedge pressure.

markedly with disappearance of both his abnormal symptoms and physical signs. He was discharged home two weeks after operation. In view of the inherent danger of prescribing anticoagulants to patients with pseudoxanthoma elasticum he was not given anticoagulants. Three months later he was well and had no symptoms or abnormal physical signs. Echocardiography confirmed his clinical improvement. Posterior left ventricular wall motion was normal and there did not seem to be any restriction of diastolic left ventricular filling. Cardiac catheterisation was repeated. Table 2 shows the considerable improvement in left ventricular end diastolic pressure, which had fallen to 10 mm Hg. Pulmonary artery pressure was correspondingly reduced. Left ventricular angiography showed an overall improvement in contractility.

He subsequently remained well for one year but later presented with a two month history of dyspnoea and orthopnoea. On examination he was once again found to be in pulmonary oedema with bilateral basal crepitations, a sinus tachycardia, and loud third heart sound. There was a loud mid-diastolic murmur at the apex. Cross sectional echocardiography showed that the mitral heterograft was thickened and rigid. The left ventricular filling rate was reduced. Overall left ventricular contractility appeared to be normal. Cardiac catheterisation was again repeated. The left ventricular end diastolic pressure was within normal limits at 5 mm Hg. There was a gradient of 19 mm Hg across the mitral prosthesis. Left ventricular angiography confirmed the continued improvement in left ventricular contractility. The mitral heterograft appeared rigid and immobile. These findings suggested that the return of his symptoms was related to stenosis of the mitral heterograft and not to a recurrence of the restrictive cardiomyopathy.

Further operation was indicated and the mitral heterograft was replaced with a St Jude prosthesis and treatment with anticoagulants was started and continued. His recovery was uneventful and associated with complete loss of all his symptoms. At follow up over the next year he was free of symptoms and leading a normal life.

Discussion

The rare condition of pseudoxanthoma elasticum, first described by Rigal in 1881, is inherited as an autosomal recessive.¹ It is a multisystem disorder affecting the skin, mucous membranes, eyes, blood vessels, and heart. The pathogenesis is poorly understood, but there is an important defect in the metabolism and subsequent synthesis of elastic

fibres. The resulting elastic tissue, which is abnormal, calcifies and degenerates.²

Cardiovascular complications are well documented and include angina pectoris caused by premature coronary artery disease,³ with sudden death from acute myocardial infarction,⁴ hypertension,⁵ premature peripheral vascular disease,⁶ mitral valve prolapse,⁷ and congestive cardiac failure. The case we describe had mild coronary artery disease, but congestive cardiac failure was the predominant cardiovascular complication. This has been reported in up to 70% of cases. The aetiology is multifactorial with hypertension, mitral valve disease, myocardial ischaemia,⁸ and direct endocardial involvement⁹ implicated, although the exact manner in which the latter leads to congestive cardiac failure is poorly understood. In the case we describe diffuse endocardial fibrosis and calcification resulted in the development of restrictive cardiomyopathy. Restrictive cardiomyopathy has only been described on one previous occasion in pseudoxanthoma elasticum.¹⁰ The characteristic echocardiographic features of a restrictive cardiomyopathy have been described in other infiltrative cardiac disorders including primary cardiac amyloidosis.¹¹ These features include a normally sized ventricular cavity with normal ventricular systolic function, "flattening" of ventricular posterior wall motion,¹² diastolic restriction of ventricular filling, and reduction in the E-F slope of the mitral valve. There is usually considerable endocardial thickening, and the left atrium is enlarged in the absence of mitral valve disease. In our case all of these features affected the left ventricle.

The standard 12 lead electrocardiogram is usually non-specific, although T wave changes and conducting disturbances have been recorded. The chest x ray is similarly not usually a help in establishing the diagnosis, but calcification was a striking feature in our patient. There is little information on the echocardiographic features of cardiac involvement in pseudoxanthoma elasticum, and we believe our report to be the first to describe echocardiographic features of a restrictive cardiomyopathy in this condition.

Cardiac surgery has rarely been undertaken in pseudoxanthoma elasticum, although triple coronary artery bypass grafting has been performed in an 18 year old girl with a seven year history of angina pectoris.¹³ We believe that our case is the first in which relief of a restrictive cardiomyopathy by limited resection of the calcified bands has been attempted. The duration of such palliation cannot be certain but in the medium term relief of symptoms has been good. A Carpentier-Edwards mitral heterograft was initially inserted in this case, in the

hope that lifelong anticoagulant treatment could be avoided, because of the danger of gastrointestinal bleeding in this condition. Valvar heart disease such as mitral calcification, mitral and tricuspid valve stenosis, and aortic insufficiency, although rare, have all been reported.^{14 15} It therefore seems likely that the rapid onset of heterograft calcification in our patient was directly related to the disease process. Thus despite the obvious risks of anticoagulation, a mechanical prosthesis is preferable to a heterograft.

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