Billowing Mitral Valve Syndrome in Association with Absent Left Pericardium

A Case Report

W. A. POCOCK, J. B. LAKIER, J. D. BENJAMIN

SUMMARY

A patient with congenital complete absence of the left pericardium had a late systolic murmur and an intermittent non-ejection systolic click. A post-exercise ECG showed ST-segment depression and T-wave inversion, compatible with that described in the billowing mitral leaflet syndrome. Left ventricular cine-angiography confirmed prolapse of the mitral valve. It is suggested that an associated billowing mitral leaflet syndrome may be responsible for chest pain, variable auscultatory features and abnormal ECGs in some patients with absent left pericardium.


Congenital complete absence of the left pericardium is a rare disorder and Southworth and Stevenson encountered only 1 case in more than 14 000 necropsies at Johns Hopkins Hospital. Before 1970 not more than 200 cases had been reported. The majority of reported cases were diagnosed incidentally at necropsy or during surgery for an associated anomaly, but the condition is now being recognized clinically with increasing frequency. The anomaly may be isolated or associated with other congenital abnormalities, such as a patent ductus arteriosus, atrial septal defect, bronchogenic cysts and pulmonary sequestration.

This report describes a patient with congenital absence of the left pericardium and the billowing mitral leaflet syndrome, an association not hitherto recognized.

CASE REPORT

A 28-year-old man underwent an examination for insurance purposes in August 1975. Physical signs and electrocardiographic features led the physician to suspect idiopathic hypertrophic subaortic stenosis, and the patient was referred to the Cardiac Unit for evaluation.

For the past 2 - 3 months he had been aware of feelings of dizziness and faintness and of an occasional fleeting praecordial pain which was not related to effort. The patient stated that he had been born with a 'rotated heart'. There was no history of rheumatic fever and he had participated in active sport while at school. Examination showed a tall, well-built man. The pulse rate was 70/min, blood pressure 125/80 mmHg and the jugular venous pressure was normal. The apex beat was displaced to the 6th intercostal space in the mid-axillary line and was compatible with left ventricular hypertrophy, but had an unusual 'rolling' character. There was a third heart sound and a grade 2/6 late systolic murmur at the apex which became louder on inspiration. At the base a soft lower frequency murmur started just before midsystole and extended through the second heart sound. In the erect position, the late systolic murmur at the apex was still present and a non-ejection click was heard at this site. A diagnosis of a billowing (prolapsing) mitral valve with minimal mitral regurgitation was made, but the basal murmurs and the apparent marked cardiomegaly were not understood. The ECG showed a mean frontal plane QRS axis of +100° with a T-wave axis of +30°. A rSr' pattern was recorded in lead V1 and there was clockwise rotation with a dominant S wave in lead V5. The T waves were flat in leads V5 and V6. There was no evidence of left ventricular hypertrophy. The radiological features established the diagnosis of complete absence of the left pericardium (Fig. 1). The heart was displaced to the left and was

Fig. 1. Postero-anterior chest radiograph. The heart is displaced to the left and there are translucent bands between the left hemidiaphragm and base of the heart and between the pulmonary artery segment and the aortic knob. The latter two convexities, together with the left border of the heart, comprise the cardiac silhouette described as the 'Snoopy sign' by Morgan et al.
not enlarged. The cardiac silhouette demonstrated the 'Snoopy sign' with prominence of the pulmonary artery segment and elongation of the left heart border. Lung tissue was interposed between the aortic knuckle and the pulmonary artery segment and between the inferior surface of the heart and the diaphragm.

At a repeat clinical examination 10 days later the auscultatory signs were different, in that a soft, short, early diastolic murmur, indistinguishable from one arising at the aortic valve, was present at the base, during both natural respiration and held expiration. The late-onset basal systolic murmur extending into diastole had disappeared and the apical late systolic murmur started earlier. The non-ejection click was not heard. He was subjected to strenuous effort and the heart rate increased to 150/min. Immediately after effort (Fig. 2) the T waves became inverted in standard lead III, and there was a 2-mm ST-segment depression in leads V5 and V6 which appeared junctional rather than ischaemic. Five minutes after effort the T waves had become inverted in leads V5 and V6 and this was still present 5 minutes later. The voltages of the QRS complexes in leads V5, V7 were larger in the post-effort recording, and there was an unusual degree of variation in amplitude of the QRS complexes in the praecordial leads during natural respiration (Fig. 3, top), which was accentuated by held inspiration and expiration (Fig. 3, bottom).

**CONTROL**

**POST EXERCISE**

**Fig. 2.** ECG recorded immediately and at 2, 5 and 10 minutes after completion of exercise.

**Fig. 3.** Top: lead V4 recorded immediately after effort shows variation in amplitude of the QRS complex and in configuration of the P and T waves, consistent with an exaggerated respiratory electrical alternans. Sinus arrhythmia is present. Bottom: during held inspiration there is a decrease in the height of the R wave in lead V6, probably due to interposition of lung between the heart and chest wall.

Echocardiography did not demonstrate prolapse of the mitral valve. Abnormal septal motion and an enlarged right ventricular diameter suggested right ventricular volume overload.

Because of the reported incidence of unexplained, apparently innocent murmurs in patients with uncomplicated absent pericardium, we could not be certain whether the apical late systolic murmur indicated mild mitral regurgitation and billowing of the mitral leaflets, and
the patient was subjected to cardiac catheterization. The marked mobility of the heart was obvious on fluoroscopy. Right heart, pulmonary arterial wedge and left ventricular pressures were normal. Left ventricular cine-angiography in the right anterior oblique position demonstrated mild mitral regurgitation and slight prolapse of the middle scallop of the posterior leaflet of the mitral valve (Fig. 4). There was no aortic incompetence.

Fig. 4. Cine-angiogram from the left ventricle (LV), recorded in the right anterior oblique position, demonstrates slight opacification of the left atrium (LA) and mild prolapse (arrow) of the middle scallop of the posterior mitral leaflet.

In view of the abnormal mitral valve the patient has been advised to observe precautions against infective endocarditis, but has been reassured as to the benign prognosis of the absent pericardium.

DISCUSSION

The clinical, radiological, electrocardiographic and echocardiographic features in our patient are characteristic of those reported in congenital complete absence of the left pericardium. Some of the unusual features of the ECG, such as the marked variation in direction and amplitude of the QRS complex with respiration (Fig. 3), can also be accounted for by the increased mobility of the heart, and were due, probably, to varying interposition of lung between the heart and anterior chest wall. The increased amplitude of the R wave, suggestive of left ventricular hypertrophy, in the post-effort tracing (Fig. 2) as compared to the control one, may possibly be due to a difference in the relationship between the heart and chest wall when the patient resumed the supine position. Tubbs and Yacoub recorded electrical alternans in one of their patients and attributed it to increased cardiac mobility. The abnormal T waves observed in our patient have as yet not been reported as a feature of absent left pericardium, but are quite compatible with the billowing mitral leaflet syndrome. It is noteworthy that the ECGs of 3 of the 6 young men with congenital absence of the left pericardium reported by Morgan et al. show inverted or flat T waves in the inferior leads and in 1 instance in leads V5 and V6 as well; these patterns are characteristic of the billowing mitral leaflet syndrome. The authors did not comment on these abnormalities, nor on whether billowing of the mitral valve was observed on left ventricular cine-angiography, and it is possible that minor degrees of prolapse were overlooked. Three of their patients had apical systolic murmurs, but in none was mitral regurgitation observed.

The auscultatory features of absent left pericardium have not been elucidated. Apical systolic and diastolic murmurs have been documented. Unexplained early diastolic murmurs, similar to the murmur heard in our patient, have also been reported, but basal systolic murmurs, ascribed to a pulmonary origin, are a more common finding. Ellis et al. attributed the murmurs to "... turbulence, set up by varying mechanical deformations at the base of an unusually mobile heart", and most authors have concurred. Pulmonary regurgitation is unlikely in our patient, as the murmur was not of low frequency. Although aortography did not demonstrate aortic regurgitation, it cannot be assumed that the early diastolic murmur was extracardiac in origin, since an intermittent minor degree of aortic regurgitation, produced by distortion of the aortic root, cannot be excluded. However, the late systolic murmur, which was also variable, was intracardiac and due to minimal mitral incompetence in association with billowing of the posterior leaflet. It is possible that some of the apical systolic murmurs documented in patients with absent pericardium are on this basis, and certainly if a non-ejection systolic click is present, a prolapsing mitral valve should be strongly suspected. The association may be coincidental, since this abnormality of the mitral valve is relatively common. However, a developmental anomaly on an embryological basis is possible, and both patent ductus arteriosus and atrial septal defect, which have been reported with absent pericardium, are found together with the billowing mitral leaflet syndrome. Chest pain is a symptom in some patients with absent pericardium and has been attributed to increased stress on the anchoring structures of the cardiac base and to tension on cardiopleural adhesions. In such patients it would be necessary to exclude the billowing mitral leaflet syndrome, in which chest pain is a recognized feature, particularly if a late systolic murmur is audible. The presence of a non-ejection click would make the diagnosis virtually certain. The importance lies in the fact that uncomplicated absent pericardium is compatible with a normal life expectancy whereas the prognosis of the billowing mitral leaflet syndrome is less certain, and patients are at risk of infective endocarditis, arrhythmias and, rarely, sudden death.

REFERENCES

History of Medicine

Professor F. P. Scott (1915-1976) — his Contribution to Dermatology in South Africa

G. H. FINDLAY

SUMMARY
Tribute is paid to F. P. Scott, and the nature and circumstances of his contribution to dermatology are described. Conditions prevailing in the Transvaal and the Orange Free State during his lifetime are related to his professional, academic and research achievements.

BACKGROUND AND EDUCATION
As a Freestater born and bred, he was the right man in the right place. He started life as a farmer’s son with a farm-school education in the Winburg area. The Free State reclaimed him for general practice in Edenville and Vrede, later for specialist practice during 10 years in Bloemfontein, and for his last 6 years as full-time professor with his own department of dermatology at the University of the Orange Free State. Besides being the obvious local choice for the University, his unique character and remarkable ability would have graced a civilized community anywhere.

By temperament and background he possessed a stoical and resourceful nature with abundant insight and perceptiveness, but a straightforward rural upbringing dominated him entirely until he was nearly 20 years of age. Then as a medical student the impact of Johannesburg and later Groningen released him from the remaining traces of country naivety. Cities, with their variety of people, attitudes and accomplishments, opened an entirely new world. Instead of deteriorating under the wicked influence of the towns, Scott made up for lost time by becoming absorbed in art, books, science, people and ideas. While completing his medical training in wartime Holland, he was pitchforked into the complexities caused by the German occupation, persecution of Jews, underground movements, care of the sick, and struggles against privation. He married, he delivered his first child, and was evacuated almost empty-handed with his wife on a ship's deck to England after the liberation, carrying the baby with them in a basket. In Britain he caught a glimpse of a people who could keep their good humour and courtesy in trying circumstances, a trait which he himself possessed to a rare degree. These are a few of the thoughts which Scott sometimes mentioned about his earlier life. One