MARFAN'S SYNDROME

Samuel Ong & W.H. Ng

INTRODUCTION

IN 1896, Marfan first reported a skeletal anomaly in a 5 year old girl characterised by long slender extremities. Subsequent reports described similar skeletal anomalies associated with disorders of the connective tissues of the cardiovascular system and eyes (Mckusick, 1955). This disorder, Marfan's Syndrome, is transmitted as an autosomal dominant trait with variable penetrance. Complications arising in the cardiovascular system contribute to the premature deaths in these patients, while the skeletal and occular lesions cause considerable morbidity.

This report is of a 20 year old Malay girl with Marfan's Syndrome. The clinical features and echocardiographic findings are presented. Methods of treatment, and the causes of death in Marfan's Syndrome are discussed.

CASE REPORT

C.S., a 20 year old single Malay girl, was admitted on 10-1-79 for an upper respiratory tract infection. Further questioning established a reduced effort tolerance over the past 2-3 years. There were no past history of cardiac failure, rheumatic fever or visual disturbances. The nuclear family history is shown in Fig. 1.

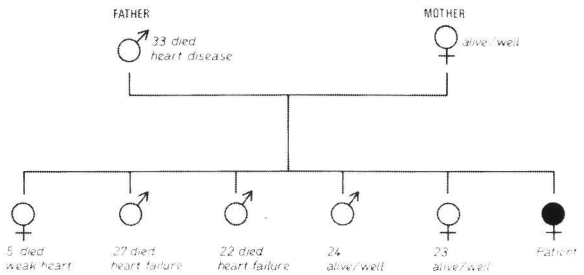

Fig. 1. Diagramatic representation of family history of the patient.

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Physical examination revealed a tall, thin, young lady measuring 170 cm. in height and 185 cm. in arms span. There was hyperextensibility of the joints and arachnodactyl. A high-arched palate was detected in te oral cavity and no abnormalities were detected in the eyes. In the cardiovascular system examination, her blood pressure was 160/60 mm. of Hg. with a regular pulse rate of 80/min. which was collapsing in character. There was gross cardiomegaly due to dilatation and hypertrophy of both ventricles. A loud pansystolic murmur of mitral incompetence was audible in the mitral area radiating to the axilla and an early diastolic murmur of aortic incompetence was heard along the left sternal edge. The rest of the physical examination was normal.

Chest x-ray (CXR) confirmed cardiomegaly predominantly of the left ventricle. The electrocardiogram (ECG) showed left ventricular hypertrophy but no strain pattern (Fig. 2). The haemoglobin was 10.2 gm. %, WBC 5,800 with a normal differential and the ESR was 2 mm. in the first hour. Serum electrolytes, blood urea and anti-streptolysin 0 titre were normal. Blood cultures, VDRL test and LE cells were all negative.

At echocardiography, with the echo beam directed at the aortic root, gross aortic root dilatation with normal aortic valves and left atrium were seen (Fig. 3). The end systolic aortic root

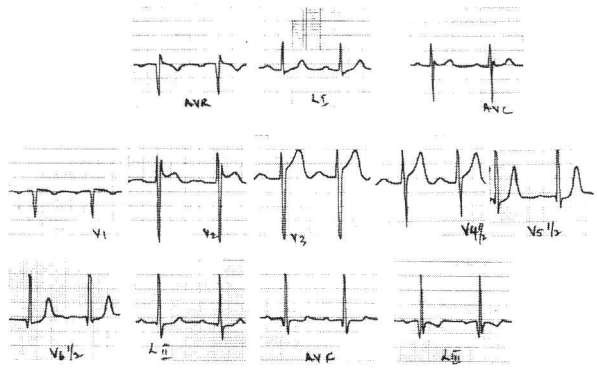

Fig. 2. ECG showing left ventricular hypertrophy.

diameter was 5.1 cm (normal 2 — 3.5 cm). There was no dissection of the aorta. In the mitral valve area (Fig. 4), fluttering of the anterior mitral leaflet indicating significant aortic incompetence, and holosystolic prolapse of the mitral valve were demonstrated.

Hyperdynamic left septum and posterior wall motion indicating left ventricular volume overload with gross dilatation of the left ventricular cavity and dilated right ventricle were seen with the echo beam directed at the ventricles (Fig. 5). The left ventricular internal diameter in the diastole was 7.8 cms. (normal 3.0 — 5.5 cm).

DISCUSSION

The association of cardiovascular lesions with Marfan's Syndrome was first noted by Salle in 1912. Goyette and Palmer (1953) in a study estimated that the cardiovascular complications occur in 30-60% of such patients. It has been shown that these cardiovascular lesions, predominantly aortic incompetence and its complications, contribute to the significantly reduced life span. Murdoch *et al.* (1972), in a study of 257 patients with the Marfan's Syndrome found the average age of death was 32 years in 74 cases. The cause of death from cardiac complications occured in 52 of these 74 cases (70.3%). Thev include rupture of aortic aneurysm, dissecting aneurysm, aortic incompetence with cardiac failure, myocardial infarction, infective endocarditis, cardiac arrythmias and acute heart blocks.

Due to its non-invasive qualities, echocardiography would be an ideal method of studying the cardiovascular complications of Marfan's Syndrome. Contrary to popular belief, aortic lesions occur less frequently than involvement of the mitral valve. Spangler *et al.* (1975), in an echocardiographic study of patients with the Marfan's Syndrome, found mitral lesions in 50%, aortic lesions in 18%, and mixed aortic and mitral lesions in 15% of cases. The remaining patients had no cardiac abnormalities. The cardiovascular complications include aortic or pulmonary ring dilatation with consequent incompetence, aortic aneurysm, dissection of the aorta, mitral incompetence, varying degrees of mitral valvular or annular prolapse and infective endocarditis.

Echocardiography is used in measuring the dimensions of the aortic root, left atrium and left ventricle at initial assessment. Sequential measurements aid in assessing the progress of the disease process, thus deciding the time for cardiac catherization or surgical intervention. Detection of aortic dissection and aortic aneurysm and its progression

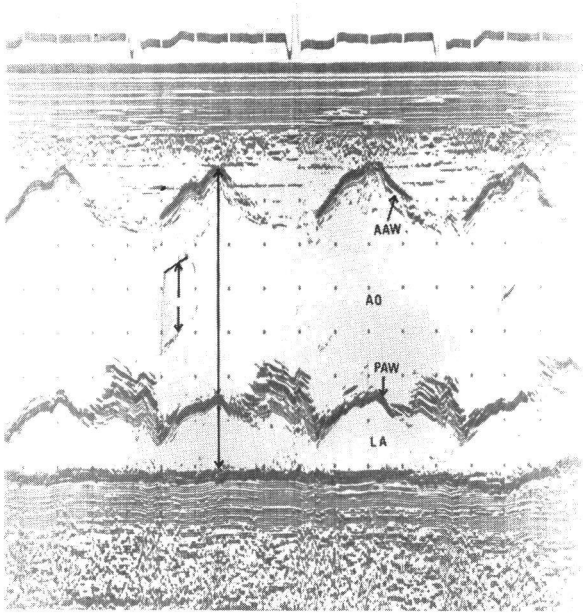

Fig. 3. Echocardiogram of the aortic root showing gross aortic root dilatation; normal aortic valves and left atrial size. (AAW — anterior aortic wall, PAW — posterior aortic wall, LA — left atrium, AO — pos- AV — aortic valve)

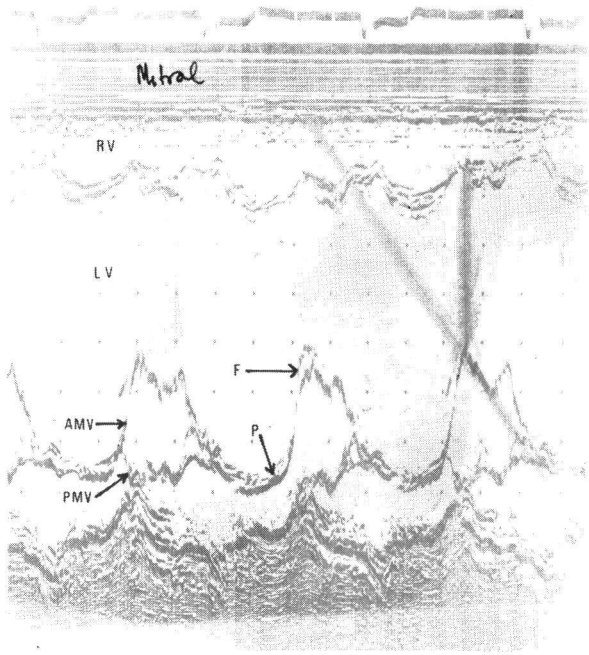

Fig. 4. Echocardiogram of the mitral valve showing fluttering of anterior mitral leaflet of aortic incompetence, and prolapse of the mitral valve. (RV — right ventricle, LV — left ventricle, AMV — anterior mitral valve, PMV — posterior mitral valve, F — fluttering of anterior mitral leaflet, P — prolapse of mitral valve in systole).

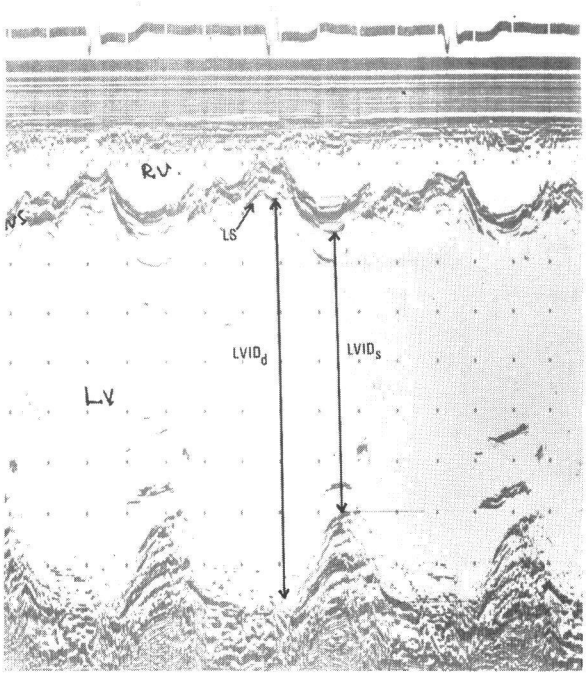

Fig. 5. Left ventricular echocardiogram showing left ventricular volume overload and bi-ventricular dilatation. (LVIDd — left ventricular internal diameter in diastole, LVIDs — left ventricular internal diameter is systole, LS — left septum).

may also be made echocardiographically (Nanda, Gramiak and Shah, 1973; Kronzon and Mehta, 1974). It may also detect mitral valvular abnormalities in the absence of clinical signs or radiological changes.

Medical treatment is directed primarily to the prevention of the progression of the cardiovascular complications, especially in situations of dissecting aortic aneurysms and aneurysmal rupture. Reserpine and hexamethonium have been shown to decrease myocardial contractility, which is the more important factor than the lowering of blood pressure in preventing aortic dissection (Beaven and Murphy. 1956). The beta-blockers, a new anti-hypertensive and anti-anginal agent, also decrease myocardial contractility. However, few studies of its use in Marfan's Syndrome have been reported. Halpern *et. al.* (1971), in a preliminary trial with propranolol claimed encouraging results. They however cautioned that it remained to be established that long term beta-blockade could prolong life in such cases. Patients with combined aortic and mitral insufficiency, as with this patient, have a rapidly deteriorating clinical course with extremely high mortality. Surgical correction of aneurysms and prosthetic valve replacement of defective valves can be offered in the management of these patients. Nelson and Vaughn (1969) reported good results with double valve replacement in the Marfan's Syndrome. Similarly Singh and Bentall (1972), obtained encouraging results with complete replacement of the ascending aorta with aortic valve replacement in the treatment of aortic aneurysm.

As seen from Fig. 1, the family history of heart disease with premature deaths, presumably from Marfan's Syndrome, is strong. Cardiovascular assessment of the surviving members would detect complications arising from the Marfan's Syndrome and also serve to chart their progress.

SUMMARY

A case of Marfan's Syndrome is described. Echocardiographic features and the role of echocardiography in the management of such patients are discussed. The methods of treatment are also highlighted.

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