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Mitral valve prolapse and mitral insufficiency in two siblings with Gaucher's disease

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Abstract

Gaucher's disease is an autosomal recessive storage disorder. We report two siblings with Gaucher's disease, both of which had mitral valve prolapse and mitral insufficiency. One of the siblings died of bacterial endocarditis and pneumonia, while the other continues under followup.

MeSH: Gaucher's disease, Mitral valve prolapse, Endocarditis

Introduction

Gaucher's disease is the commonest sphingolipidosis, and was first described by Gaucher in 1882. This condition is autosomal recessive, and leads to deposition of glucocerebrosides in various organs, especially in the reticuloendothelial system.^{1,2}

Three major clinical phenotypes have been identified:³

Type 1: Adult nonneuropathic

Type 2: Infantile neuropathic

Type 3: Juvenile subacute neuropathic.

We report two siblings with Gaucher's disease, both of which had mitral valve prolapse and mitral insufficiency. One of the siblings died of bacterial endocarditis and pneumonia, while the other continues under followup.

Case 1:

A 17 year old girl was admitted to our institution with palpitations, fatigue and fever. She was known to have hydrocephalus, bilateral corneal opacities, deafness, left ventricular hypertrophy and clinodactyly related to Gaucher's disease.¹ Physical examination showed a temperature of 39C, pulse of 120/min, BP 90/60, prominent apical impulse in the 5th left intercostal space in the midclavicular line without a thrill. Auscultation showed a pansystolic murmur (2-3/6) at the left sternal edge radiating to the axilla and a softer (1-2/6) systolic murmur in the aortic area which radiated up the right side of the neck.

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Blood count showed Hb: 10g/dl, HCT 31%, WBC $16 \times 10^9/l$; ESR 96mm in one hour. CXR showed marked cardiomegaly with pneumonic infiltration in the left paracardiac area.

Echocardiography showed hypertrophy and calcification of both right and left ventricular myocardium which also involved all valve leaflets. Neither the mitral nor the aortic valves were stenotic. The mitral valve also had a semimobile calcified nodule on its anterior leaflet which measured 5 by 5 mm, which was demonstrated on both transthoracic and transoesophageal echocardiography (fig 1). Blood cultures failed to grow any microorganisms.

Figure 1 Transoesophageal echocardiographic image of nodule on mitral valve (case 1)



(LA: left atrium, LV: left ventricle)

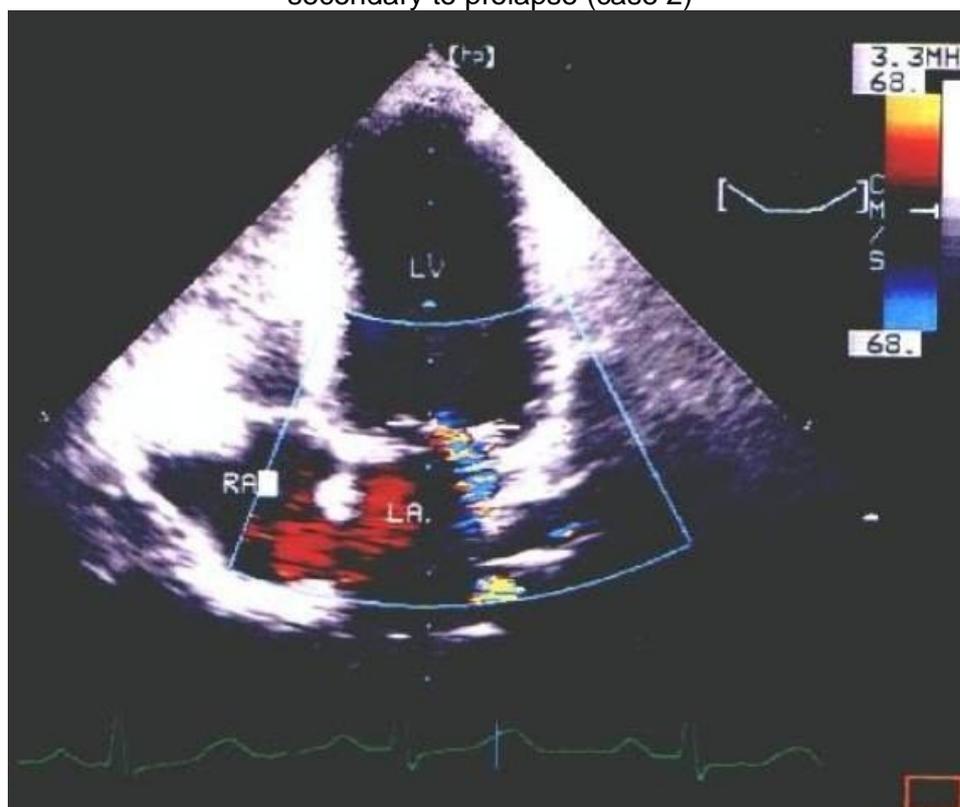
The initial diagnosis was that of pneumonia and endocarditis and she was treated with intravenous vancomycin and gentamicin. However, she expired after three days of treatment. The family did not allow a postmortem study.

Case

A 12 year old, a known case of Gaucher's disease, was admitted to our institution with palpitations and syncope. Manifestations of Gaucher's disease included deafness, corneal opacities and hydrocephalus. Examination showed a pulse of 96/min with a BP of 120/70. Auscultation showed an apical midsystolic click and a late systolic murmur. ECG and CXR were normal. Echocardiography showed mitral valve prolapse of the anterior leaflet with mild mitral insufficiency (fig 2).

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Figure 2 Transthoracic echocardiographic image mitral valve with mitral insufficiency secondary to prolapse (case 2)



(LA: left atrium, LV: left ventricle, RV: right ventricle)

Discussion

Although it had been initially believed that Gaucher's disease does not involve the heart, this has now been shown to be incorrect.⁴⁻⁷ Cardiac manifestations include constrictive pericarditis, rheumatic aortic valve disease and annulo-aortic ectasia with Debakey II dissecting aneurysm. Valvular calcifications in this condition have been linked with corneal opacities.⁵ Simultaneous mitral and aortic valvar involvement with Gaucher's disease has been reported in two siblings aged 9 and 15 years.⁶ Gaucher's disease with mitral valve calcification has also been reported,⁷ and was followed by a report of 3 cases, two of whom had severe simultaneous mitral and aortic involvement, and one of whom died of heart failure.³

Myocardial infiltration with Gaucher cells has been demonstrated in both myocardium and in valve tissue,⁸ and it was speculated that the valvar damage was by a cell mediated mechanism which involved both bone matrix proteins and integrins.

We speculate that cardiac involvement in Gaucher's disease may be common, and that such involvement is now more easily demonstrated with accessibility to echocardiography, both transthoracic and transoesophageal. Our report, to the best of our knowledge, is the first that describes transoesophageal echocardiography in an individual with Gaucher's disease and in addition, this is also the first report mitral valve prolapse with mitral insufficiency in patients with Gaucher's disease.

If there are any doubts as to the possibility of cardiac involvement in patients with Gaucher's disease, we suggest transthoracic echocardiography, followed by transoesophageal echocardiography if deemed necessary.

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