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CASE REPORT

Gaucher disease causing sudden cardiac death

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KEYWORDS

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Abstract A 17-year-old male patient with Gaucher disease was presented to our institution complaining of rapid irregular palpitations. Echocardiography showed the presence of critical aortic stenosis due to Gaucher disease.

1. Introduction

Gaucher disease is a lysosome storage disease that leads to accumulation of glycolipids in the cells and can be treated by enzyme replacement therapy.[1] Patients usually present with hepatosplenomegaly, anemia, thrombocytopenia, bleeding tendency, bone pain, osteopenia, pathologic fractures, growth retardation, neurological manifestations and rarely cardiac affection.[2]

2. Case report

A 17-year-old male patient, unemployed and single, has been diagnosed as having Gaucher disease in 2003 when he was presented with growth retardation. The patient had hepatosplenomegaly, anemia and thrombocytopenia. A bone marrow biopsy revealed Gaucher cells and a reduced glucocerebrosidase activity was detected in peripheral leukocytes. Hence, enzyme replacement therapy (recombinant glucocerebrosidases) was given once every two weeks. The patient reported that after therapy he became of average height and weight within two years. In 2011, the patient started manifesting poor performance in school that led to school dropout in 2013. In addition, he complained of a head thrusting movement since 2011 that was diagnosed as oculomotor nerve apraxia.

The patient was presented to our facility in 9/2013 complaining of exertional, irregular recurrent palpitations that terminate spontaneously within 30 min by rest. He denied any other cardiac symptoms including dyspnea, orthopnea, paroxysmal nocturnal dyspnea, chest pain nor syncope. Upon presentation, his blood pressure was 100/70 mmHg, heart rate 90 bpm and peripheral pulses well felt. Cardiac examination revealed an ejection systolic murmur heard over aortic area and a pansystolic murmur heard over the mitral area. Abdominal examination revealed hepatosplenomegaly.

The electrocardiogram showed normal sinus rhythm with left ventricular hypertrophy and strain pattern. The laboratory workup was unremarkable except for microcytic hypochromic anemia with hemoglobin: 7.7 g/dl, platelets: 59,000/Ul and the chest X-ray was unremarkable.

Echocardiography revealed left ventricular hypertrophy with a good systolic function (Ejection fraction: 70%) (Fig. 1). The aortic valve was heavily calcific leading to critical stenosis with a peak systolic velocity of 6 m/s, aortic valve area 0.5 cm^2, mean systolic gradient 88 mmHg and mild aortic

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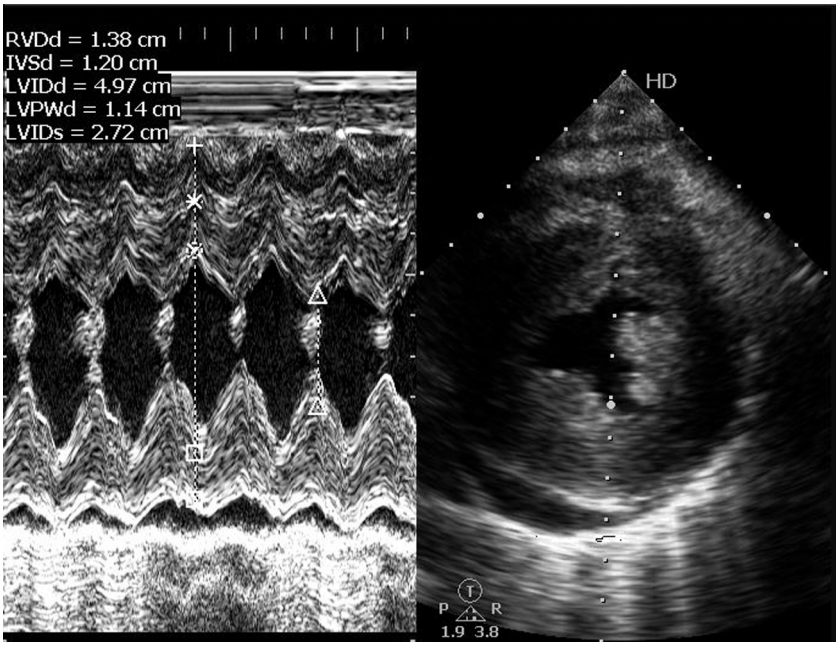

Figure 1 M-mode across the left ventricular cavity showing a good systolic function and left ventricular hypertrophy.

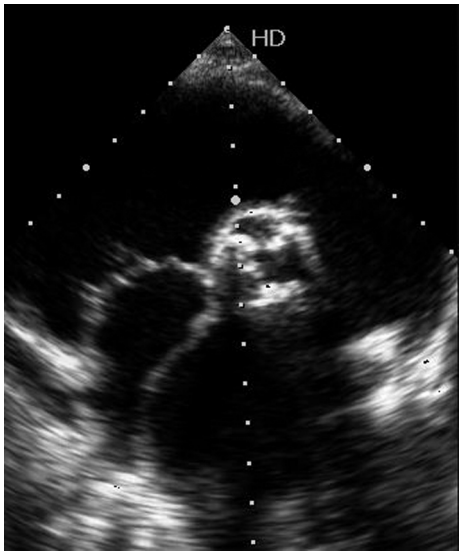

Figure 2 Parasternal short view showing a heavily calcific aortic valve.

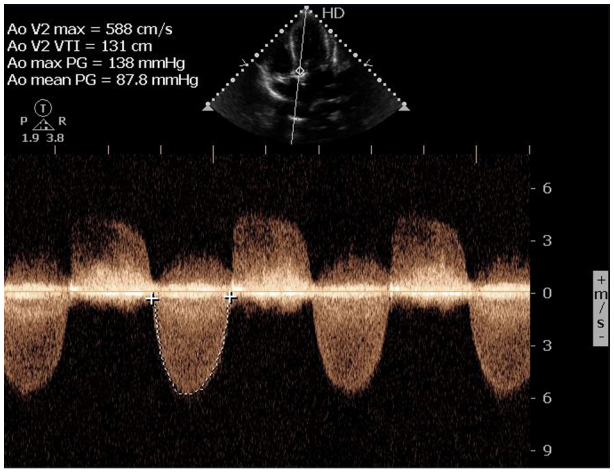

Figure 3 Continuous Doppler wave across the aortic valve showing critical aortic stenosis and mild aortic regurgitation.

regurgitation (Figs. 2 and 3). The aortic root was small with echodense walls. The aortic annulus, bisinus, Sinotubular and ascending aorta measured were 16 mm, 20 mm, 17 mm and 19 mm respectively (Figs. 4 and 5). The aortic calcifications were extending to the aortomitral intervalvular fibrosa and to the anterior mitral valve leaflet, and focal calcifications were visualized at the attachment of the posterior leaflet to the lateral mitral annulus (Figs. 5 and 6). Both leaflets showed mild restriction of mobility and the subvalvular involvement was mild. All of the previous findings led to moderate regurgitation and mild stenosis (Mitral valve area: 1.8 cm^2 measured by 2D planimetry) (Figs. 7 and 8). The pericardium appeared to be echodense and thickened particularly at the posterior basal segment but there was no evidence of calcifications (Fig. 6). In addition to the previous findings there was mild pericardial effusion localized mainly at the posterior segment, left atrial dilation, mild tricuspid regurgitation, and mild pulmonary hypertension (predicted pulmonary artery systolic pressure: 40 mmHg).

The patient was referred to the cardiothoracic surgery department for the possibility of double valve replacement, surgery was deferred as the patient was not complaining of any symptoms of heart failure and the surgery was considered to be of high risk due to thrombocytopenia plus the technical difficulty in aortic valve replacement due to the small aortic annulus. A follow-up was scheduled every three months.