Gaucher's Disease with Mitral and Aortic Involvement: Echocardiographic Findings

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SUMMARY. Cardiac involvement in Gaucher's disease has been reported in only a few patients, mostly adults with pericardial changes. We describe findings in two siblings with Gaucher's disease, aged 15 and 9 years respectively, in whom mitral and aortic valve lesions were evaluated by auscultation and echocardiography. In both cases the mitral and aortic valves were thick and restricted in motion. Continuous Doppler echocardiography revealed significant mitral regurgitation and mitral stenosis. At the aortic valve level there was a systolic pressure gradient. Echocardiographic investigation of patients with suspected cardiac involvement with Gaucher's disease is recommended.

KEY WORDS: Gaucher's disease — Echocardiography — Cardiac involvement

Gaucher's disease is a rare, inherited metabolic disorder [9]. Although it most commonly affects the reticuloendothelial system cells of the spleen, liver, and bone marrow and, less frequently, the lymph nodes, kidneys, and lungs may also be involved [6]. Cardiac lesions are extremely rare, with only occasional reports [4]. All were adults, except a 15-year-old boy with aortic and mitral lesions reported by Casta et al. [2].

We here describe the cases of two siblings whose mitral and aortic valves were affected by Gaucher's disease, in particular the M-mode, two-dimensional, and Doppler echocardiographic findings.

Case Reports

Case 1

A 15-year-old boy with Gaucher's disease had been followed for 5 years. The diagnosis had been made by bone marrow and liver biopsies. There was no history of rheumatic fever and cardiac disease. The family history revealed second-degree consanguinity between the parents. The patient had been complaining about getting easily tired for the previous few months. Because of the development of a significant heart murmur the patient was referred to the department of Pediatric Cardiology for evaluation.

On examination, there was no cyanosis. His height was 148 cm, weight 34 kg. Arterial blood pressure was 110/60 mmHg, the heart rate was 88/min. There was no respiratory distress. At the cardiac apex a grade 4/VI blowing systolic murmur was heard; a diastolic murmur was time at mid and late diastole. Additionally, there was a grade 2/VI systolic ejection murmur at the aortic area and an early diastolic descrescendo murmur at the third left intercostal space, indicating aortic valve involvement. The liver edge was 6 cm below the right costal margin, while the spleen was palpable at 10 cm below the left costal margin.

The ECG showed a normal QRS axis in the frontal plane. There was left atrial enlargement, as well as left ventricular hypertrophy. The plain chest x-ray film revealed slight cardiomegaly. Hemoglobin (Hb) level was 11.80 g/dl, white blood cell count (WBC) 4600/mm³, blood sedimentation rate 10 mm/h, anti-streptolysin O (ASO) titer 330 units, and C-reactive protein (CRP) negative.

Echocardiography was carried out by a Toshiba Sonolayer-SSH 60 Echocardiograph using 3.7 MHz, 5 MHz, and 2.5 MHz transducers. The main cardiac structures were normal. Left ventricular ejection fraction was 60% and shortening fraction 39%. Left ventricular end-diastolic diameter was 46 mm, and left atrial diameter 51 mm. Aortic root diameter was 21 mm. The mitral and aortic valves were thick and restricted in motion (Figs. 1-3). Continuous Doppler echocardiography revealed significant mitral regurgitation and mitral stenosis.

Right and left heart catheterization and angiography were performed from the right femoral region by the percutaneous technique. Oxygen saturations revealed no significant change. Pressures were as follows: pulmonary wedge 21 mmHg (mean), pulmonary artery 62/66 mmHg (mean 43), right ventricle 80/5 mmHg, and right atrium 21 mmHg (mean). On retrograde left heart catheterization the pressures were 145/7 mmHg in the left ventricle and 112/60 mmHg (mean 90) in the aorta. Left ventricular and aortic biplane cineangiograms illustrated...
Fig. 1. M-mode and two-dimensional echocardiograms of case 1. Restricted aortic valve motion is seen in the M-mode (arrows). The aortic root shows dense echos. Left atrium (LA) is enlarged (51 mm). AO, aorta; M, mitral valve; LV, left ventricle; RV, right ventricle.

Fig. 2. Restricted mitral valve motion seen on M-mode recording (up and downward arrows). The posterior mitral valve moves anteriorly during diastole, indicating mitral stenosis (arrow). Parasternal long-axis two-dimensional study shows an echo-dense mitral valve (double arrows). IVS, interventricular septum (case 1).

rigid ascending aortic walls and valve cusps. There was a non-coronary cusp prolapse and minimal aortic regurgitation (Fig. 4). Left atrium was extremely enlarged and significant mitral regurgitation was seen. The parents did not accept surgical treatment. The patient was digitalized with digoxin and a diuretic was given. One month later we were informed that the patient had died in a local hospital. Postmortem studies were not permitted by the family.

Fig. 3. Echogenic aortic root is seen on both M-mode and two-dimensional recording (arrows) (case 1).

Fig. 4. Retrograde aortography. Rigid aortic cusps with non-coronary cusp prolapse and minimal aortic regurgitation are seen (case 1).

Case 2

A 9-year-old girl, the sister of case 1, had been followed for Gaucher's disease for 2 years. The diagnosis had been made by bone marrow. There was no history of rheumatic fever. The patient had been complaining of tiring easily and of nose bleeds for the last few months.

There was no cyanosis. Her height was 115 cm, weight 21 kg. The arterial blood pressure was 110/60 mmHg, at a heart rate of 85/min. There was a grade 2/VI pansystolic murmur at the cardiac apex. A faint and short systolic murmur was heard at the aortic area. The liver edge was 2 cm below the right costal margin, while the spleen enlarged to 6 cm below the costal margin. Hb level was 11.80 g/dl, WBC 8300/mm³, and ESR 12 mm/