Echocardiographic demonstration of isolated mitral valve involvement in a patient with mucopolysaccharidosis

Mukopolisakkaridozlu bir hastada izole mitral kapak tutulumunun ekokardiyografi ile gösterilmesi

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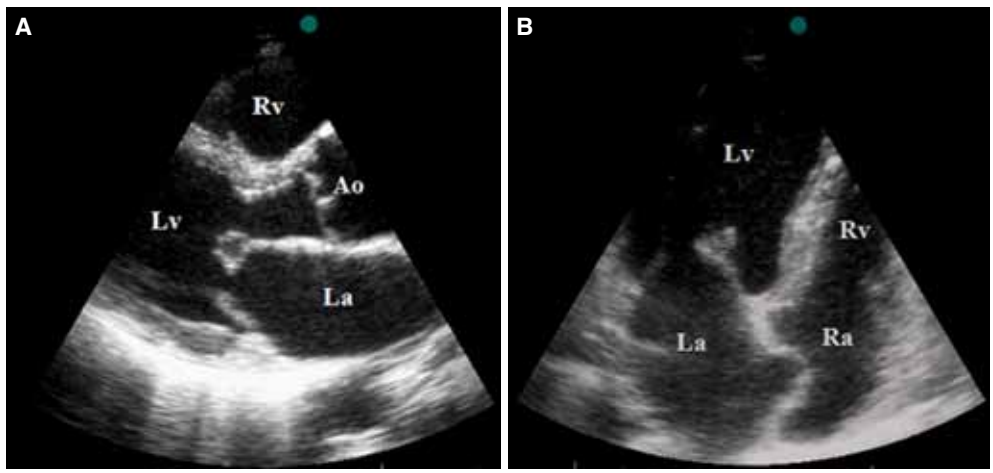
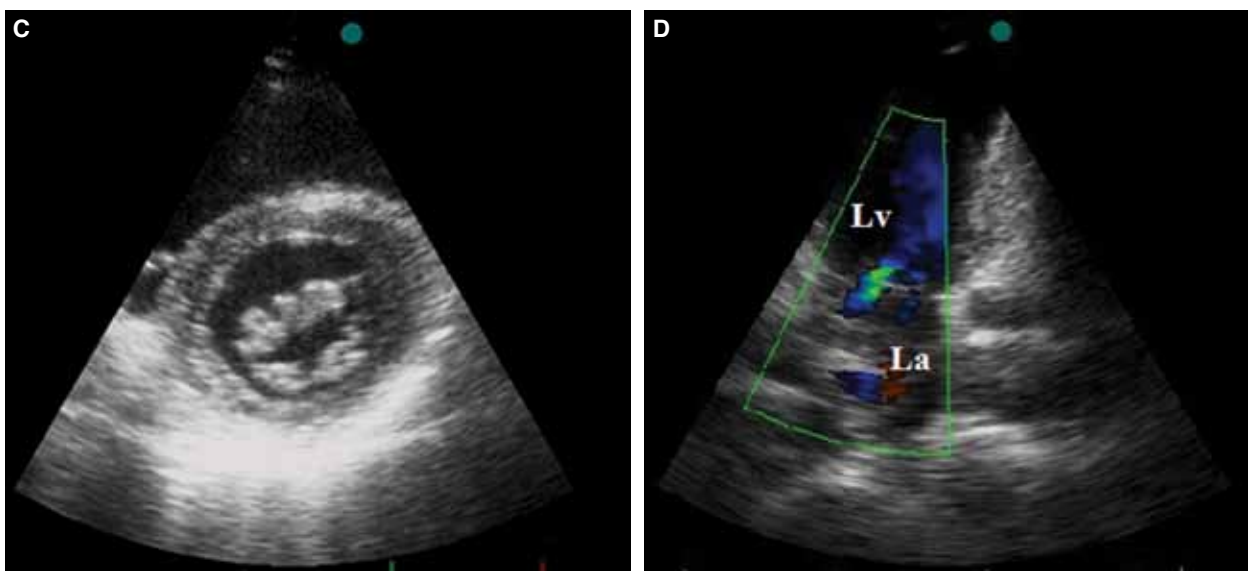


A 27-year-old female patient who had mucopolysaccharidosis type II and had been followed-up by medical treatment (enzyme replacement therapy) for three years was admitted to our cardiology clinic for further assessment of cardiac involvement. On physical examination, she had an apical 3/6 systolic ejection murmur of low intensity. Electrocardiography showed sinus rhythm with normal axis. The chest X-ray was unremarkable. Transthoracic echocardiography revealed mitral valve thickening with mild mitral regurgitation and an atrial septal aneurysm (Fig. A-D, supplementary video files 1-3*). Other echocardiographic findings were normal. Transesophageal echocardiography could not be performed due to the lack of patient's cooperation. We recommended regular routine echocardiographic follow-up to the patient.



Figures. (A) Parasternal long-axis, **(B)** apical 4-chamber, and **(C)** parasternal short-axis views showing mitral valve thickening. **(D)** Color Doppler echocardiography showing mild mitral regurgitation. **Supplementary video files associated with this case can be found in the online version.*