Large left ventricular thrombus in a patient with Friedreich’s ataxia

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A 25 year old woman was admitted to the hospital because of fatigue, palpitation, and dyspnoea on minor exertion. Nineteen years before, Friedreich’s ataxia had been diagnosed. On physical examination, the patient presented characteristic findings of Friedreich’s ataxia, including marked ataxia, dysarthria, generalised weakness, severe scoliosis, and cavus deformity of the foot. No heart murmur was audible. The electrocardiogram showed atrial flutter with 2:1 atrioventricular conduction; the QRS rate was 170/min. Echocardiography revealed biventricular hypertrophy with left ventricular wall thickness of 13 mm, severe global hypokinesia of a left ventricle of normal size, and a large apical thrombus (3.5 × 2.5 cm) (figure).

About 20 hours after the echocardiographic examination, the patient suddenly lost consciousness; she was intubated because of respiratory insufficiency. Although pulses of the carotid arteries were strongly palpable, pulses at all four extremities were not. Thrombus embolisation was therefore suspected, and confirmed by echocardiography. The neurological disorder and the apparent cerebrovascular involvement ruled out vascular surgical intervention.

At necropsy residual thrombotic material was present in the apex of the left ventricle. Occlusive thrombotic material was found in the left internal carotid artery, the left subclavian and right brachial artery, and the aortic bifurcation.

Although Friedreich’s ataxia is primarily an inherited progressive neurological disease, heart failure is the most common cause of death in these patients. Concentric or, less frequently, asymmetrical left ventricular wall thickening may readily be detected by echocardiography: rarely, a picture of dilated cardiomyopathy is found. Echocardiography also allows assessment of systolic and diastolic ventricular function. Increasing physical immobility can mask myocardial insufficiency for a long time. Atrial arrhythmias raise suspicions of left ventricular dysfunction and indicate a poor prognosis. Occasional peripheral emboli resulting from mural thrombi or, more usually, from atrial thrombi in cases of atrial fibrillation may occur in Friedreich’s ataxia. The thrombus was exceptionally large in this case and there was massive lethal embolisation. Anticoagulation should be considered in patients with Friedreich’s ataxia and impaired ventricular wall motion or atrial fibrillation.

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