The fact that it was observed in a 69-year-old woman supports the thesis that cholesterol lowering in the elderly can reduce the risk of coronary morbidity and mortality in this population group.

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References


Specific heart muscle disease associated with glycogen storage disease type III: clinical similarity to the dilated phase of hypertrophic cardiomyopathy

Glycogen storage disease type III (GSDIII) is an autosomal recessive disorder characterized by a deficiency of the glycogen debranching enzyme. We present a patient with a specific heart muscle disease associated with GSDIII, with clinical findings comparable to those in the dilated phase of hypertrophic cardiomyopathy.

A 38-year-old Japanese woman was hospitalized with fatigue and dyspnoea on exertion. Her parents were first cousins, but her family history was otherwise unremarkable. The patient’s physical development during childhood was normal, although liver dysfunction and cardiomegaly had been noted at 17 and 23 years, respectively.

On admission, her blood pressure was 118/74 mmHg, and her pulse rate was 96 beats min⁻¹. Hepatosplenomegaly and mild muscle weakness were revealed. There were persistent elevations in concentrations of serum creatine kinase (455 U. l⁻¹, MB: 5-3%). An electrocardiograph was abnormal, demonstrating left ventricular hypertrophy, pathological Q waves, and inverted T waves in leads I, II, aVL, and V₃₋₆. A two-dimensional echocardiographic study revealed that parts of the anterolateral and posterolateral portions of the left ventricular wall were akinetic, with visible thinning of the wall. The left ventricle was enlarged (left ventricular end-diastolic dimension: 56 mm) with decreased contractility (ejection fraction: 24%). Left ventricular hypertrophy (left ventricular mass index: 225 g m⁻², left ventricular septum thickness: 18 mm) was present (Fig. 1, panel (A)). During exercise scintigraphy, akinetic portions of the ventricle showed no uptake of thallium-201 (Fig. 1, panel (B)). The coronary arteries appeared normal on cardiac catheterization. An endomyocardial
biopsy specimen demonstrated central vacuolar degeneration of myocytes, with PAS-positive depositions within the myofibres (Fig. 1, panel (C)). Debranching enzyme activity was determined in endomyocardial biopsy specimens, but it was absent (0.04 nmol. min \(^{-1}\). mg \(^{-1}\) protein vs 4.4 units in control subjects) in a spectrophotometric assay\(^1\). Cardiac symptoms were not relieved, and the patient died suddenly 5 months later.

Moderate left ventricular hypertrophy with scattered and patchy fibrosis was present at autopsy. Fibrosis was prominent in the anterior and posterior walls. This was consistent with akinetic motion and visible wall thinning, demonstrated by echocardiography and the absence of thallium-201 uptake during exercise scintigraphy. Microscopic findings revealed regions of focal fibrosis distributed throughout the myocardium (Fig. 1, panel (D)).

This patient was diagnosed as having a specific heart muscle disease associated with GSD III on the basis of enzymatic analysis. In addition to hypertrophy, the left ventricle was dilated. Some portions of it were akinetic and replaced by fibrous tissue. These conditions caused the patient to suffer from symptoms of heart failure and probably resulted in the sudden death. A review of the literature has suggested that electrocardiographic abnormalities have been identified in approximately 70% of the patients with GSD III, and echocardiographic abnormalities in approximately 50%\(^2,3\). Almost all patients demonstrate hypertrophic cardiomyopathy, although manifestations of symptoms are very rare\(^4,5\). In this patient, serum concentrations of myocardial enzymes were elevated, which suggested continuous disruption of the myocardium. Fibrotic changes in the heart may be an additional factor leading to the deterioration of cardiac function and a poor prognosis. However, study of additional cases is required to establish the details of cardiac involvement in this disease.

This case is a good example of how the clinical picture of a patient with a specific heart muscle disease may be consistent with that of the dilated phase of hypertrophic cardiomyopathy. Hence, one should consider that a specific heart muscle disease may be associated with GSD III, when diagnosing a patient with signs symptomatic of the dilated phase of hypertrophic cardiomyopathy.

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References


Congenital heart disease

Congenital heart disease now features in adult cardiology as an entity, albeit a small one. In no European language is there another word for congenital, which is not surprising as the word comes from the Latin derivation, which is the origin of many European languages. Thus it is inevitable we use the initials CHD in English. Unfortunately these letters are also used for coronary heart disease so when one

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