A 38-year-old woman suffered from fatigue, stress-induced dyspnea, and a chronic cough. She also was suffering from loss of body weight (4 kg within 3 months) and constipation.

The patient is the third daughter of a Vietnamese family with inherited heart disease. The members of this family (third generation known to have the heart disease) suffer from fatigue, muscle weakness, atrophy, and cardiac arrhythmias. Symptoms become manifest in the early forties and lead to death after 3 to 10 years (Figure 1).

Echocardiography revealed a moderate cardiac hypertrophy with slight accentuation in the basal septum, as well as good left ventricular systolic function. Normal valves and atria could be documented.

In the $^{99}$Tc-methylene-diphosphonate scan (Tc-MDP-scan), a significantly increased incorporation in the heart was observed, indicating the deposition of amyloid (Figure 2B).

In cardiac catheterization, a normal left ventricular function could be shown angiographically. Normal left ventricular and right ventricular systolic pressures were observed. Only end-diastolic pressures were significantly elevated, with a typical dip-plateau phenomenon (Figure 3). This dip-plateau phenomenon indicates the restrictive component of this form of cardiomyopathy.

In biopsies taken from the stomach and rectum, the deposit of excessive amounts of amyloid could be visualized. Extensive deposits of amyloid were also observed in biopsies taken from the left ventricle (Figure 4). The definitive diagnosis made after noninvasive testing using the $^{99}$Tc-MDP scan can be based on the invasively taken myocardial biopsies. In the neurological test, a loss of electrical evoked sympathetic skin reaction was observed, which indicated an autonomic nerve defect.

Figure 1. Pedigree of the Vietnamese family. The index patients are indicated by the arrows. The third generation of the family is known to suffer from an inherited heart disease (see text).
Figure 2. Shown is the $^{99}$Tc-MDP scan of a normal control (A) and of a patient with amyloidosis (index patient 2, B). $^{99}$Tc-MDP, which binds in the calcium-binding site of the amyloid, can clearly identify cardiac amyloidosis noninvasively with high sensitivity and specificity and thus is of major prognostic and therapeutic value, especially in families with suspected amyloidosis due to missense mutations of the transthyretin gene.

Figure 3. Left ventricular pressure recordings with typical dip-plateau phenomenon in the 38-year-old patient with the suspected cardiac amyloidosis (index patient 2 in the pedigree). Elevated end-diastolic pressures with a typical dip-plateau phenomenon are parameters for a restrictive form of cardiomyopathy. These are indicative of but not specific for cardiac amyloidosis.

Figure 4. Histology of left ventricular biopsies of the 38-year-old patient. Cardiomyocytes are surrounded by fine fibrillar, kongo red-positive amyloid (A). In polarized light, the amyloid appears apple-green (B). The definitive diagnosis after noninvasive testing using the $^{99}$Tc-MDP scan can be based on the invasively taken myocardial biopsies.
Restrictive Cardiomyopathy in Familial Amyloidosis TTR-Arg-50
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