IMAGES IN CARDIOLOGY

Intracellular cardiac amyloidosis in familial amyloid polyneuropathy of the Portuguese type

We report on a 38 year old woman of Portuguese origin with peripheral ascending polyneuropathy for several years. Her family history was positive for hereditary systemic amyloidosis. A familial amyloidotic polyneuropathy with a Portuguese type I variant (Val→Met30) of the transthyretin molecule (prealbumin) was diagnosed. In the following months, symptoms worsened, the gait became stepping, and urinary incontinence increased. In order to prevent disease progression it was decided to perform liver transplantation. Electrocardiographic monitoring showed continuous bradycardic phases without variability of the heart frequency, suggesting cardiac involvement of the amyloidosis. However, echocardiography did not reveal any pathological alterations. Histological examination of endomyocardial biopsy specimens of the right ventricle demonstrated variably configured cardiomyocytes (panel 1). Amyloid was detected by the Congo red reaction which showed a typical apple green birefringence under polarised light (panel 2). Conspicuously, amyloid was localised intracellularly. Immunohistochemical labelling with antibodies directed against human prealbumin (anti-AF, DAKO, Glostrup, Denmark) revealed strong AF staining in the cardiomyocytes (panel 3). Furthermore, intracellular amyloid fibrils with an average diameter of 10.5 nm could be observed by transmission electron microscopy (panel 4).

Amyloidosis is defined as a group of diseases characterised by typical interstitial deposition of congophilic fibrillar proteins. The heart may be infiltrated in different forms including hereditary amyloidosis such as the familial amyloidotic polyneuropathy of the Portuguese type, but never with intracellular amyloid deposits. However, it is still uncertain whether the intracellular amyloid deposits influence the course and prognosis of cardiac amyloidosis.

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Panel 1. Light microscopy of tissue from cardiac biopsy (haematoxylin and eosin stain; bar = 100 µm). Panel 2. Congo red stain with polarised light showing characteristic apple green birefringence (bar = 50 µm). Arrows: intracellular amyloid-deposits. Panel 3. Immunohistochemical labelling with anti-AF amyloid (red) demonstrating strong intracellular staining (counterstaining with haematoxylin; Bar = 20 µm). Panel 4. Electron microscopy with intracellular amyloid fibrils (bar = 1 µm). Arrows: intracellular amyloid fibrils; N, nuclei; M, mitochondria; F, myofibrils.