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Portuguese-type amyloidosis (transthyretin amyloidosis, ATTR V30M)

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Abstract

Portuguese-type amyloidosis (transthyretin amyloidosis, ATTR V30M) is the most common form

of systemic hereditary amyloidosis, inherited in autosomal dominant mode. The disease, also called familial amyloid polyneuropathy type I (FAP-I), is caused by a mutant transthyretin (TTR) protein, which is synthesized by the liver. A single amino acid substitution of methionine for valine at position 30 of the TTR molecule (TTR V30M) was found in Portuguese patients. The clinical disease usually manifests as a peripheral sensory, motor and autonomic neuropathy starting in the 3rd or 4th decade of life. Renal manifestations of ATTR V30M, like other amyloidoses, are different levels of proteinuria and renal insufficiency. In ATTR V30M a large amyloid deposition in the medullary zone of the kidney and tubules is characteristic. A more extensive glomerular and vascular involvement is present only in patients with renal manifestations. A prospective survey in the north of Portugal showed that a stage of microalbuminuria (MA) could precede nephropathy and neurological disease. Nephropathy in FAP-I is present in one-third of affected patients and tends to aggregate in families. The progression towards end-stage renal disease (ESRD) affects 10% of the patients, and the survival after initiation of dialysis is a mean of 21 months. Patients who progress to ESRD have a late onset of neuropathy and lower prevalence of clinical disease in their families. Liver transplantation is a widely accepted treatment for FAP-I, and combined liver-kidney transplantation is also an option for selected patients with FAP-I and ESRD.

Full Text - PDF

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