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Familial amyloid polyneuropathy type I (Portuguese): distribution and characterization of renal amyloid deposits.

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Source

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Abstract

Renal amyloidosis has been considered rare and late in the evolution of the transthyretin (TTR) familial amyloid polyneuropathy (FAP) of the Portuguese type (type I). Renal biopsy has been performed systematically in 14 patients with FAP type I before liver transplantation. In all patients, TTR Met30 mutation was shown. Seven had proteinuria or abnormal microalbuminuria, whereas seven others had no urinary abnormalities. All had renal amyloid deposition predominantly in the medulla. Glomerular and vascular involvement was more prominent in patients with urinary abnormalities. Patients with the most extensive renal lesions represented a subgroup with a low score of polyneuropathy disability, a high prevalence of nephropathy in the proband generation, or a late onset for relatives with nephropathy. Immunohistochemistry studies showed that the amyloid substance corresponded to transthyretin. We have shown that renal TTR-derived amyloid deposition is common in patients with FAP type I, even in the absence of urinary abnormalities. The clinical presentation of nephropathy is not a late occurrence in the disease.

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