Late-Onset Familial Amyloid Polyneuropathy Unrelated to Known Endemics

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A 77-year-old man born in Yamagata Prefecture, Japan was referred to our hospital because of paresthesia in the distal portion of his extremities. His medical history consisted of several episodes of syncope and constipation since he was 50 years old. Neurological and electrophysiological examinations revealed a sensori-motor neuropathy. The sural nerve biopsy showed amyloid deposition as revealed by direct fast scarlet (DFS) stain (Picture 1A). No demyelination or axonal changes were apparent. Electron-microscopic observation revealed aggregated fibrils (10-15nm in width), (Picture 1B), aligned parallel to each other (Picture 1C). The gene analysis demonstrated transthyretin gene mutation of Val30Met (transthyretin Val30Met-associated familial amyloid polyneuropathy, FAP ATTR Val30Met). Family history revealed that one of his brothers had been diagnosed as having FAP ATTR Val30Met based on gene analysis. Patients with FAP ATTR Val30Met unrelated to the two Japanese foci in Kumamoto and Nagano generally have milder symptoms (1).

Reference