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A mutant Thr95Ile ATTR cardiac amyloidosis with polyneuropathy

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A 72-year-old man was referred for progressive dyspnea and peripheral edema since last year. He had a history of advanced gastric cancer and subsequent radical subtotal gastrectomy with adjuvant chemotherapy (Tegafur/Gemercil/Oteracil) two years ago. There was no family history of cardiovascular disease. A chest X-ray showed cardiomegaly, and an electrocardiogram showed sinus bradycardia with low QRS voltage in limb leads and left axis deviation. An echocardiogram showed thickened biventricular walls with granular sparkling appearance and biatrial enlargement. Global left ventricular (LV) systolic function was reduced to ejection fraction (EF) 43%, and in diastole, LV showed restrictive filling pattern. Cardiac magnetic resonance (CMR) imaging showed diffuse transmural delayed gadolinium enhancement of LV. A cardiac biopsy revealed deposition of amorphous material in interstitium. However, immunofixation of serum and urine proteins demonstrated no abnormal finding, while Technetium-99m Pyrophosphate (PYP) scan was suggestive of TTR-related amyloidosis (ATTR) with intensely diffuse PYP uptake in myocardium of LV. DNA sequencing of the exon 3 in TTR gene revealed Thr95Ile (c.284C>T) mutation, which is reported as the second case in the world. His LVEF was progressively decreased to 32% in spite of optimal medical treatment for heart failure. He underwent implantable cardioverter-defibrillator implantation for primary prevention. He also suffered from paresthesia in both legs, which was diagnosed as sensori-motor polyneuropathy. Therefore, he is scheduled to be treated with tafamidis, proven to be an effective therapy for ATTR cardiomyopathy and peripheral neuropathy.