We report the case of a 32-year-old man who had suffered from orthostatic syncope and body weight loss since he was 27 years old. As years passed by, he also showed muscle weakness and abnormal sensations in both legs, hyporeflexia in 4 limbs, and autonomic failure (impotence, urinary and fecal incontinence, and edema in lower limbs) suggesting the presence of peripheral somatic and autonomic polyneuropathy. His mother, mother’s father, and mother’s paternal aunt also had similar symptoms. Both the sensory nerve action potential and the sensory nerve conduction velocity of his right sural nerve were low (1.26 μV and 47.2 m/s, respectively), and the motor nerve conduction velocity of his right tibial nerve was 41.1 m/s (normal >45 m/s). A DNA test on the man disclosed a missense mutation in the transthyretin gene (Ser50Arg), which is relatively rare in familial transthyretin-related systemic amyloidosis. Transthyretin-immunoreactive amyloid deposition was demonstrated in the biopsied gastroduodenal mucosa (Figure 1). Echocardiography showed a markedly thickened ventricular wall (thickness of interventricular septum 22.3 mm [normal <12 mm]) with normal wall motion (fractional shortening 37.6% [normal 28–42%]), indicating that he also had cardiac...
amyloidosis (Figure 2A). Contrast magnetic resonance imaging revealed focal late gadolinium enhancement in the thickened ventricular wall (Figure 2B). The patient had been treated with orthotopic live-donor liver transplantation when he was 31 years old to alleviate and prevent exacerbation of his neuronal and cardiac symptoms. His condition, including the neurological disability, gradually improved, and he started to work again 10 months after liver transplantation.

In order to visualize amyloid deposition in the myocardium, the patient underwent a cardiac positron-emission tomography study with $[^{11}C]-BF-227$ that sensitively and specifically binds to aggregated amyloid fibrils. The positron-emission tomography images revealed significantly robust retention of $[^{11}C]-BF-227$ in the patient’s heart compared with that of the normal control (Figure 3). Biopsy specimens from the patient’s duodenum also showed higher signals of BF-227 compared with that of the normal control (Figure 1, C and D). The present result provides evidence that our amyloid-specific positron-emission tomography tracer, $[^{11}C]-BF-227$, can successfully detect amyloid deposition in the heart. Several molecules, such as $^{99m}$Tc-aprotinin and $^{99m}$Tc-labeled phosphate derivatives, have been investigated to visualize cardiac amyloidosis. None of the previous tracers, however, could specifically bind to aggregated amyloid, which forms a $\beta$-pleated sheet structure. In any of the amyloidogenic disorders, such as transthyretin-related systemic amyloidosis and Alzheimer’s disease, it is surmised that the monomer of the amyloid protein itself is not very toxic, whereas misfolded oligomers could cause damage to human organs. It is therefore truly important to detect the accumulation of real amyloid fibrils for the early and accurate diagnosis of amyloidosis. To our knowledge, this is the first report showing the usefulness of a $\beta$-pleated sheet structure-specific positron-emission tomography in investigating visceral organ amyloidosis.

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**Disclosures**

None.

**References**


Cardiac Positron-Emission Tomography Images With an Amyloid-Specific Tracer in Familial Transthyretin-Related Systemic Amyloidosis

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