論 文 要 旨

Clinical and genetic features of Charcot–Marie–Tooth disease 2F and hereditary motor neuropathy 2B in Japan

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Mutations in small heat shock protein beta-1 (HspB1) have been linked to Charcot–Marie–Tooth (CMT) disease type 2F and distal hereditary motor neuropathy type 2B. Only four cases with *HSPB1* mutations have been reported to date in Japan. In this study between April 2007 and October 2014, we conducted gene panel sequencing in a case series of 1,030 patients with inherited peripheral neuropathies (IPNs) using DNA microarray, targeted resequencing, and whole-exome sequencing. We identified *HSPB1* variants in 1.3 % (13 of 1,030) of the patients with IPNs, who exhibited a male predominance. Based on neurological and electrophysiological findings, seven patients were diagnosed with CMT disease type 2F, whereas the remaining six patients were diagnosed with distal hereditary motor neuropathy type 2B. P39L, R127W, S135C, R140G, K141Q, T151I, and P182A mutations identified in 12 patients were described previously, whereas a novel K123* variant with unknown significance was found in one patient. Diabetes and impaired glucose tolerance were detected in 6 of the 13 patients. Our findings suggest that *HSPB1* mutations result in two phenotypes of inherited neuropathies and extend the phenotypic spectrum of *HSPB1*-related disorders.