Two patients with GMPPB mutation: The overlapping phenotypes of limb-girdle myasthenic syndrome and limb-girdle muscular dystrophy dystroglycanopathy.

Mutations in the guanosine diphosphate-mannose pyrophosphorylase-B gene (GMPPB) have been identified in congenital muscular dystrophies, limb-girdle muscular dystrophy (LGMD2T), and congenital myasthenic syndromes (CMSs); overall, 41 patients have been described. Two patients presented with a myasthenic syndrome (patient 1; 74 years old) and rhabdomyolysis (patient 2; 23 years old). Examinations included repetitive nerve stimulation, muscle biopsy and whole-body MRI (WBMRI); next generation sequencing facilitated diagnosis. We identified the following GMPPB mutations: c.79G>C/c.859C>T in the 23-year-old man with LGMD2T-phenotype and c.79G>C homozygosity in the 74-year-old woman with CMS phenotype. WBMRI showed fatty degeneration of paraspinal, thigh adductor, and calf muscles in patient 1 and edematous changes of the soleus muscle in patient 2. This case of c.79G>C homozygosity causing a mild, late-onset CMS phenotype, confirms the mild nature of this common mutation. The descriptions of these 2 new GMPPB cases add to the knowledge regarding this recently discovered, heterogeneous disease. Muscle Nerve 56: 334-340, 2017.