[A Japanese family with POMT2-related limb girdle muscular dystrophy]

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Abstract

Mutations in the gene encoding the protein O-mannosyl-transferase 2 (POMT2) are known to cause autosomal recessive limb girdle muscular dystrophy type 14 (LGMDR14). No Japanese patient with LGMDR14 has been reported previously. Here, we report three patients with LGMDR14 in one family. The first and second patients harbored a novel homozygous mutation of c.1568A>G, while the third harbored a compound heterozygous mutation of c.1568A>G and c.869C>T. The novel c.1568A>G mutation is classified as likely pathogenic by the guideline of the American College of Medical Genetics and Genomics. Similar to previous cases, all three patients presented difficulty walking and cognitive impairment, and the hamstring muscles were severely affected. Although eye abnormality has only been reported in one previous case, two our patients showed eye abnormalities. As POMT2 enzymatic activity has been demonstrated in the mammalian retina, an eye abnormality may represent a phenotype associated with POMT2 mutation.

Keywords: LGMDR14; POMT2 gene; eye abnormality; familial onset; α-dystroglycanopathy.