Supplementary Information

Subjects
Informed consent was obtained from the patient according to the protocol approved by the Institutional Review Board of Gangnam Severance Hospital, Korea.

Targeted sequencing
Genomic DNA from blood of patient was extracted with Qiagen DNeasy blood & tissue kits (Qiagen, Valencia, CA, USA). For mutation analysis, the coding exons and flanking introns of 69 myopathy-causative genes were enriched by hybridization capture. The captured library was sequenced by an Illumina Hiseq2000 platform with the 2X100 bp paired-end read module. Total sequencing yield was 528.54 Mbp. The coverage of target region (≥10X) was 98.7%.

Variant analysis
The raw sequencing data were aligned to the hg19 reference genome with Burrows-Wheeler Aligner (BWA) (ver. 0.7.5a) algorithm. Output SAM files were converted to BAM files and sorted with SAMtools (ver. 0.1.18). Duplicate removal was performed with Picard tools (ver. 1.95) markduplicates. Realignment around known indel sites and base quality score recalibration (BQSR) were performed with the genome analysis toolkit (GATK) (ver. 2.6-5) to create final BAM files.

Variants were called using the GATK v2.6 Unified Genotyper algorithm for loci with sequencing depth greater than or equal to 20X. Variants were annotated with ANNOVAR (ver. 2013-06-21) using RefGene, dbSNP 138, and the 1000 Genomes Project SNP (2014 Sep release) of Asian population and all-population databases. Among called variants, only nonsynonymous single nucleotide variant (SNV), frameshift indel and splicing site variants were chosen. Polymorphisms in Korean populations (n=352) were filtered out. Variants with low functional score in SIFT, PolyPhen-2 (v2.2.2) and GERP were considered benign mutations and discarded. Pathogenic and likely pathogenic variants were validated by Sanger sequencing.