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 from 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 $2 \times 150$ bp paired-end read module. The sample was pooled into a part of a single lane on a single flow cell and sequenced together. A 6 bp index sequence (Illumina) and 6 bp inhouse barcode sequence were used to differentiate between samples. Total sequencing yield was 526.428 Mbp. The coverage of target region ($\geq 10X$) was 98.6%.

Variant analysis
From HiSeq2000 raw data, sequencing data of all samples were sorted by index and barcode sequences. Sorted fastq files were aligned to the hg19 reference genome with Burrows-Wheeler Aligner (ver. 0.7.5a) algorithm. Output SAM files were converted to BAM files and sorted with SAM tools (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 were performed with the genome analysis toolkit (GATK) (ver. 2.6–5) to create final BAM files. Variants were called using the GATK v3.3.0 Unified Genotyper algorithm. Called variants were filtered by the following criteria: 1) loci depth $\geq 10$, variant frequency $\geq 0.5$, 2) loci depth $\geq 20$, variant frequency $\geq 0.25$. Functional annotation of genetic variants was performed by ANNOVAR (ver. 2014-11-12). Additionally, we rescreened total variants for splicing sites and variable promoter regions, which are likely to be missed by conventional annotation programs. We also filtered polymorphisms found in the Korean population ($n=298$) and public databases [dbSNP 135 and 1,000 Genome project SNP (2014 September release)] from East Asian, South Asian, and all-population databases. All variants were classified into five categories (pathogenic, likely pathogenic, uncertain significance, likely benign, and benign variants) according to standards and guidelines by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology.