Supplementary Table 1. Whole-exome sequencing and identification of candidate variants in patient 2	
Total number of reads	75,285,819
Sample coverage	
2x coverage	97.0%
10x coverage	86.4%
20x coverage	74.5%
30x coverage	65.2%
Number of variants	
Total variants	56,901
Exonic variants	23,061
Functional variants[a]	10,983
MAF <0.5% in EVS	2,442
<5 pedigrees in GEM.app	204
In known CMT-related genes	4 – *NEFL*, *DST*, *SH3TC2*
Zygosity consistent with inheritance	2 – *NEFL*, *DST*
Known pathogenic mutations and consistent with phenotype	1 – *NEFL*

(a) Non-synonymous, splice-site and coding indel variants. CMT: Charcot-Marie-Tooth disease. EVS: Exome Variant Server (http://evs.gs.washington.edu/EVS/). GEM.app: GEnomes management application (https://genomics.med.miami.edu/). MAF: minor allele frequency.