

Supplementary Material 3. Analysis of the relationship of phenotypes and genotypes

First, we extracted the protein sequences of *SYNE1* variants in East Asia via National Center for Biotechnology Information (NCBI) database (<https://www.ncbi.nlm.nih.gov/>, accessed on September 17, 2022) and ExPASy translate tool (<https://web.expasy.org/translate/>, accessed on September 17, 2022). The splice sites were excluded due to their unpredictable and unavailable changes on protein sequence level. Next, we performed Basic Local Alignment Search Tool (BLAST) on The Universal Protein Resource (UniProt, <https://www.uniprot.org/>, accessed on September 17, 2022) and analyzed all the available 33 protein sequences from the stop-gain and frameshift mutations. Meanwhile, we retrieved and diagrammed the structure of all the affected Nesprin-1 isoforms (Supplementary Figure 2 in the online-only Data Supplement). Finally, we identified the locations of the protein sequences of *SYNE1* variants in Nesprin-1 to understand the relationship between phenotypes and genotypes (Supplementary Table 6 in the online-only Data Supplement).