



Fig. S1 A blood reverse transcription polymerase chain reaction (RT-PCR) assay reveals GLS (NM_014905.5):c.736-406A>G induced aberrant splicing in ID 342.

A. RT-PCR analysis shows the mRNA isoforms encompassing GLS exon 3 to exon 5 (3-4-5) from blood lymphocytes of the carrier mother (M), father (F) and healthy brother (B). D indicates 100 bp DNA molecular weight markers; the enhanced band has a size of 500 bp.

B. Electropherograms of aberrant splicing isoform 4-4'-5 of the carrier mother. The shaded blue box indicates the 128 bp cryptic exon 4' generated by the mutation c.736 - 406A>G (ID 342 DNA), and the blue squiggle represents the ellipsis of partial sequences of cryptic exon4'. The sequences of cryptic splice sites in DNA c.736 -411 of individual ID 342 and c.DNA of the carrier mother are indicated.