**Supplemental Figure 1.** The pedigrees of 5 Chinese families with IEL caused by *ADAMTS4* mutations. The wild-type *ADAMTS4* allele is indicated by a plus sign, the affected individuals are represented as filled symbols, and black arrows indicate the probands.

**Supplemental Figure 2.** Schematic view of the *ADAMTS4* structure, isoform 1(NM_019032.5). Exons are shown as rectangles with exon number and size indicated above them. The novel *ADAMTS4* mutations are also depicted.

**Supplemental Figure 3.** Novel homozygous mutation identified in family ID 043 via Sanger sequencing. The homozygous mutation is shown in the forward reads, and the red arrows indicate the mutation’s location.

**Supplemental Figure 4.** Novel heterozygous mutations identified in 4 families, including patient ID 058 (A), ID 222 (B), ID 246 (C) and ID 366 (D) via Sanger sequencing. All mutations are shown in the forward reads, and the red arrows indicate the mutations’ locations.