



Fig. S1. Pedigree, mutational analysis, Western blotting of Cases 1 and 2. (A) Case 1 inherits c.956T>C (p.Leu319Pro) and c.6955C>T (p.Arg2319Ter) from her mother and father, respectively. (B) Case 2 inherits c.2807G>A (p.Trp936Ter) from his father. The DNA sample of patient's mother is not available; (C) HEK293 cells are transfected with wild type *ITGB4* complementary DNA (cDNA) and different *PLEC* cDNA (wild type, mutant *PLEC* cDNA containing c.956T>C/p.Leu319Pro, or c.954_956dup/p.Leu319dup). Immunoblot of cell lysates showed that plectin and $\beta 4$ integrin were diminished when *ITGB4* and *PLEC* mutants (p.Leu319Pro, p.Leu319dup) were cotransfected into HEK293 cells.