Acta Dermato-Venereologica

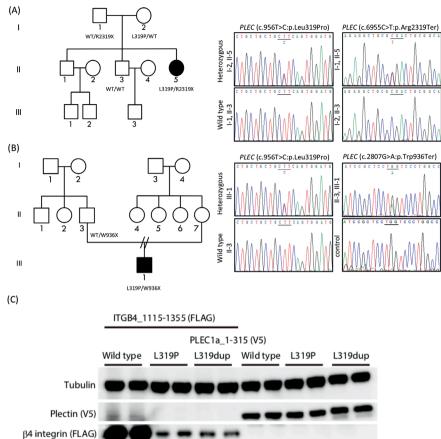


Fig. S1. Pedigree, mutational analysis, Western blotting of Cases 1 and 2. (A) Case 1 inherits c.956T>C (p.Leu319Pro) and c.6955C>T (p.Arg2319Ter) from her mother and father, respectively. (B) Case 2 inherits c.2807G>A (p.Trp936Ter) from his father. The DNA sample of patient's mother is not available; (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 $\beta4$ integrin were diminished when ITGB4 and PLEC mutants (p.Leu319Pro, p.Leu319dup) were cotransfected into HEK293 cells.