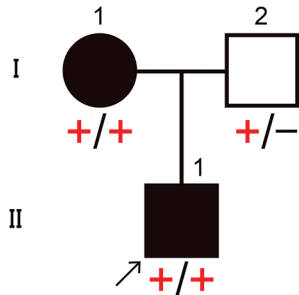


a **c.796C>T (p.(Arg266Ter))**



b

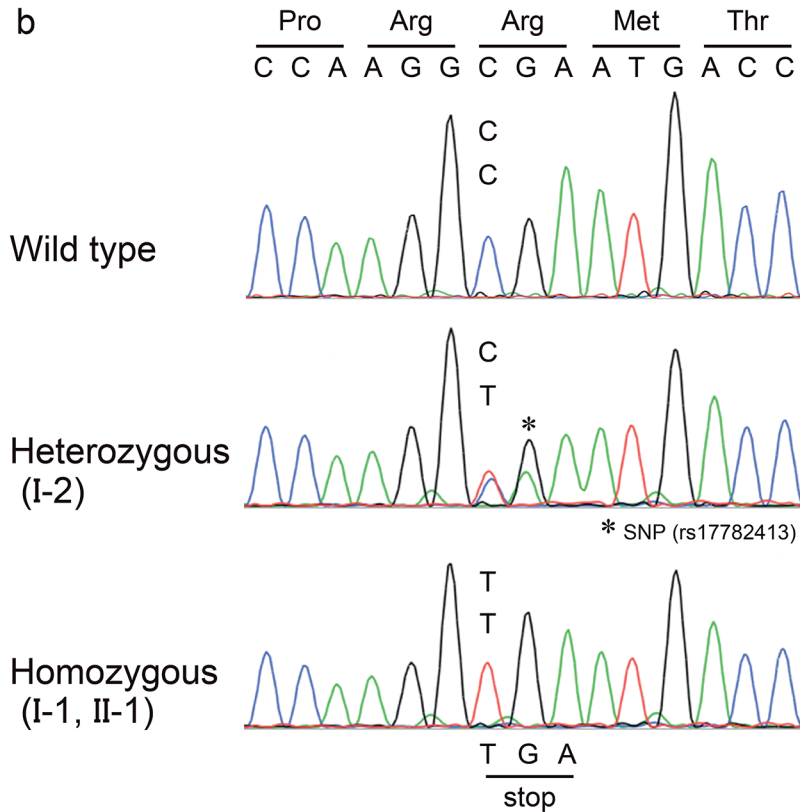


Fig. S2. Detection of *SERPINB7* mutation. (a) The pedigree of a 2-generation Japanese family with Nagashima-type palmoplantar keratosis (NPPK) shows pseudodominant inheritance. *Solid symbols*: affected individuals; *open symbols*: unaffected individuals. (b) The patient (II-1) and his mother (I-1) are homozygous for a nonsense mutation c.796C>T (p.(Arg266Ter)) in exon 8 of *SERPINB7*, whereas his father is heterozygous for the mutation. Note that the father (I-2) is also heterozygous for c.797G>A, which is a non-pathogenic single-nucleotide polymorphism (SNP) (rs17782413).