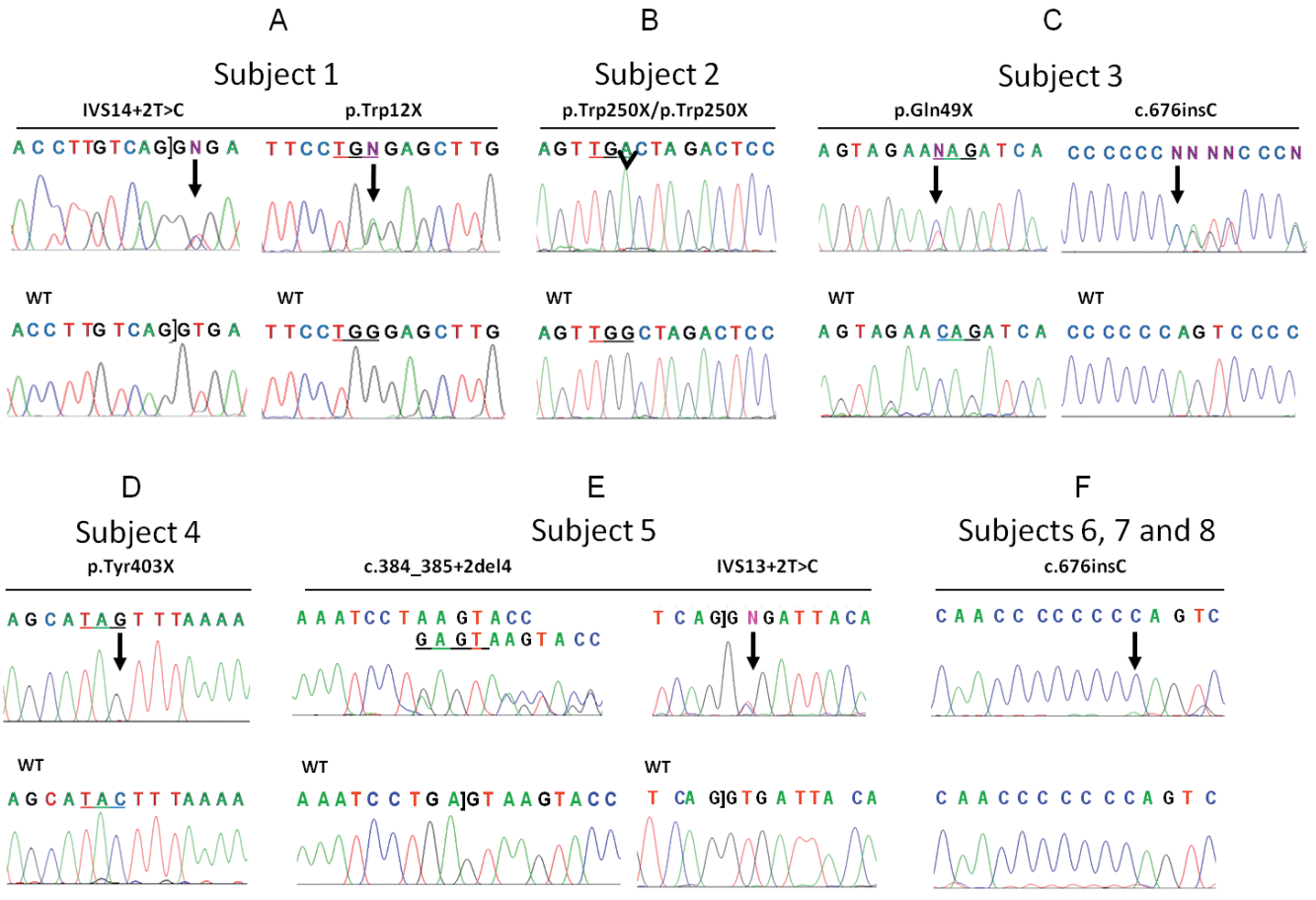

Fig. S1. Mutation analysis of the *FERMT1* gene. (A) In subject 1, there was a heterozygous donor splice site T>C transition within intron 14, denoted IVS14+2T>C and a heterozygous G>T transversion at nucleotide c.36, converting a tryptophan residue (TGG) to a stop codon (TGA), designated p.Trp12X. (B) In subject 2, there was a homozygous G>A transition at nucleotide c.750 converting a tryptophan residue (TGG) to a stop codon (TGA), designated p.Trp250X. (C) In subject 3, there was a heterozygous insertion of an extra cytosine base at position c.676, resulting in a frameshift mutation, c.676insC. In addition, there was a heterozygous C>T transition at nucleotide c.145, converting a glutamine residue (GAG) to a stop codon (TAG), designated p.Gln49X. (D) In subject 4, there was a homozygous C>G transversion at nucleotide c.1209 converting a tyrosine residue (TAC) to a stop codon (TAG), resulting in the nonsense mutation p.Tyr403X. (E) In subject 5, there was a heterozygous deletion of GA in exon 2 and a deletion of GT in the immediately adjacent intron. This mutation is denoted c384_385+2del4. In addition, there was a heterozygous donor splice site T>C transition within intron 13, denoted IVS13+2T>C. (F) In subjects 6, 7 and 8, there was a homozygous insertion of an extra cytosine at position c.676, resulting in the frameshift mutation, c.676insC. Illustrated sequence data is from subject 7.