Fig S1. Genomic DNA sequencing.
(a) Sequencing of family member DNA reveals a C>A transversion at nucleotide 321 that converts cysteine (TGC) to a stop codon (TGA); the mutation is designated p.Cys107X and homozygous, heterozygous and wild-type sequences are illustrated;
(b) Sequencing of family member DNA reveals a T>A transversion at nucleotide 682 that converts phenylalanine (TTT) to isoleucine (ATT); the mutation is designated p.Phe228Ile and homozygous, heterozygous and wild-type sequences are illustrated.