



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.