Fig. S1. Sequencing and haplotype analysis. (a) The DNA sequence of the affected Hungarian patient III/3 shows a heterozygous single nucleotide substitution c.2806C>T, which converts codon 936 CGA for arginine to TGA for stop codon; this mutation is designated p.Arg936X. (b) Wild-type sequence of genomic DNA spanning codons 933–939 of the CYLD gene. (c) Genotyping of the surrounding, common polymorphisms demonstrated that the same mutation carried by the 2 geographically distant pedigrees is surrounded by different haplotypes and was the result of 2 independent mutational events.