



Fig. S1. Patients' pedigrees, wild type and mutated CASP14 protein and sequencing results of family A. (a) Family A. The father of patients 1 and 2 married a first cousin once removed. (b) Family B. Both unaffected siblings present the wild-type sequence. Alleles are shown in parentheses. (c) Diagram of the wild-type and the hypothetical mutated CASP14 protein structure. The wild-type protein consists of a short pro-domain (*yellow*), the large catalytic subunit p17 (*blue*) and the small subunit p11 (*green*). The hypothetical protein structure illustrates the amino acid sequence alteration (aa 154) and the truncation of the subunit p11 (aa 180; red). (d) The healthy parents are heterozygous, while the patients are homozygous for the c.462_463delCA deletion (CASP14, NM_012114).