



Fig. S1. Family pedigrees and mutation detection (a) and haplotype analysis (b) in patients with hyaline fibromatosis syndrome. Note consanguinity in all three families, with the proband being indicated by an arrowhead (a, left panel). Mutation analysis in Family 1 revealed a heterozygous splice site mutation in both parents of the deceased proband (arrows). In Family 2, there was an insertion of C resulting in an increase from 8Cs in the control to 9 Cs in the patient's DNA (arrow). Asterisk shows a homozygous polymorphism, p.Ala357Pro, common in Iranians. In Family 3, the patient has deletion of T in exon 13. (b) Haplotype analysis with SNPs covering the *CMG2* locus and flanking sequences in a 3 Mb region on chromosome 4. Note a 2 Mb homozygosity block in Patient 3 from Family 3 in comparison to a previously reported case (9) with the same c.1074delT mutation (green block). Note that the patient from Family 2 with a different mutation has a different haplotype. MAF: minor allele frequency; SNP: single nucleotide polymorphism.