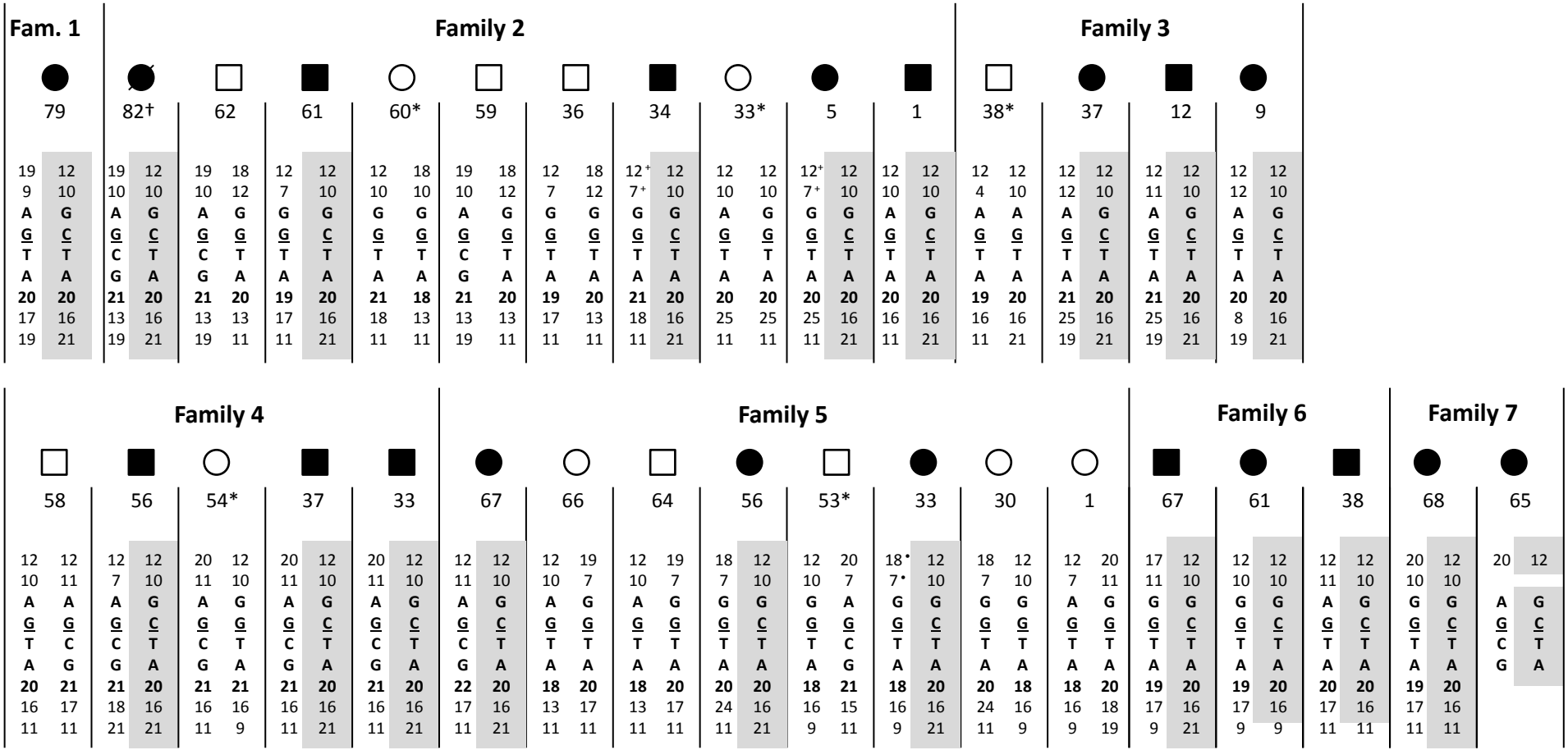
Figure S1. Haplotype analysis of patients and unaffected individuals in 7 families with the *ACVRL1* c.625+1G>C mutation [46].



Legends: blackened individuals are heterozygous, individual age at haplotype analysis is indicated; †: patient died in the year of haplotype analysis; *: non-consanguineous spouses; •: haplotype modification probably caused by recombination; intragenic polymorphic markers are highlighted with bold characters and the mutation site is additionally underscored; chromosomal location of the STR sequences: D12S85 $(AC)_n$: 12q13.11 (47,336,687-47,336,862); D12S2196 $(CTAT)_n$: 12q13.11 (48,700,248-48,700,547); D12S1677 $(GT)_n$: 12q13.13 (53,312,927-53,313,286); D12S1712 $(GT)_n$: 12q13.13 (52,445,897-52,446,222); D12S270 $(CA)_n$: 12q13.13 (52,709,971-52,710,294). Chromosomal location of the *ACVRL1* intragenic SNPs: rs2071219 (c.314-35A>G, MAF 0.437), rs706815 (c.1377+45T>C, MAF 0.343) and rs706816 (c.1377+65A>G, MAF 0.343); in the case of a 65-year-old female in Family 7 haplotype analysis was incomplete.