


































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

	Fam. 1		Family 2										Family 3					
																		
	79	82 ⁺	62	61	60*	59	36	34	33*	5	1	38*	37	12	9			
D12S85	19	12	19	18	12	12	12	18	12	12	12	12	12	12	12			
D12S2196	9	10	10	12	7	10	10	10	10	10	10	4	10	12	10			
rs2071219	A	G	A	G	G	G	A	G	G	G	A	A	A	A	G			
<u>c.625+1</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>C</u>	<u>C</u>			
rs706815	T	T	C	T	T	T	T	T	T	T	T	T	T	T	T			
rs706816	A	A	G	A	G	A	A	A	A	A	A	A	A	A	A			
D12S1677	20	20	21	20	21	20	21	20	20	20	20	19	20	21	20			
D12S1712	17	16	13	16	13	13	17	13	25	16	16	16	16	25	16			
D12S270	19	21	19	11	11	19	11	11	11	21	11	21	19	21	19			

	Family 4					Family 5					Family 6					Family 7		
																		
	58	56	54*	37	33	67	66	64	56	53*	33	30	1	67	61	38	68	65
D12S85	12	12	20	12	20	12	12	12	18	12	18*	12	18	12	12	12	20	12
D12S2196	10	11	11	10	11	10	10	7	7	10	7*	10	7	10	7	10	10	10
rs2071219	A	A	A	G	A	G	A	G	G	G	G	G	G	A	G	G	G	G
<u>c.625+1</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>G</u>	<u>C</u>	<u>G</u>	<u>C</u>	<u>C</u>
rs706815	T	C	C	T	C	T	C	T	T	T	T	T	T	T	T	T	T	T
rs706816	A	G	G	A	G	A	G	A	A	A	A	A	A	A	A	A	A	A
D12S1677	20	21	21	20	21	20	22	20	20	18	20	20	18	20	19	20	20	20
D12S1712	16	17	18	16	16	16	17	16	24	16	16	16	24	16	17	16	17	16
D12S270	11	11	21	21	11	21	11	11	11	21	9	11	9	21	9	9	11	11

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.