

Supplementary material

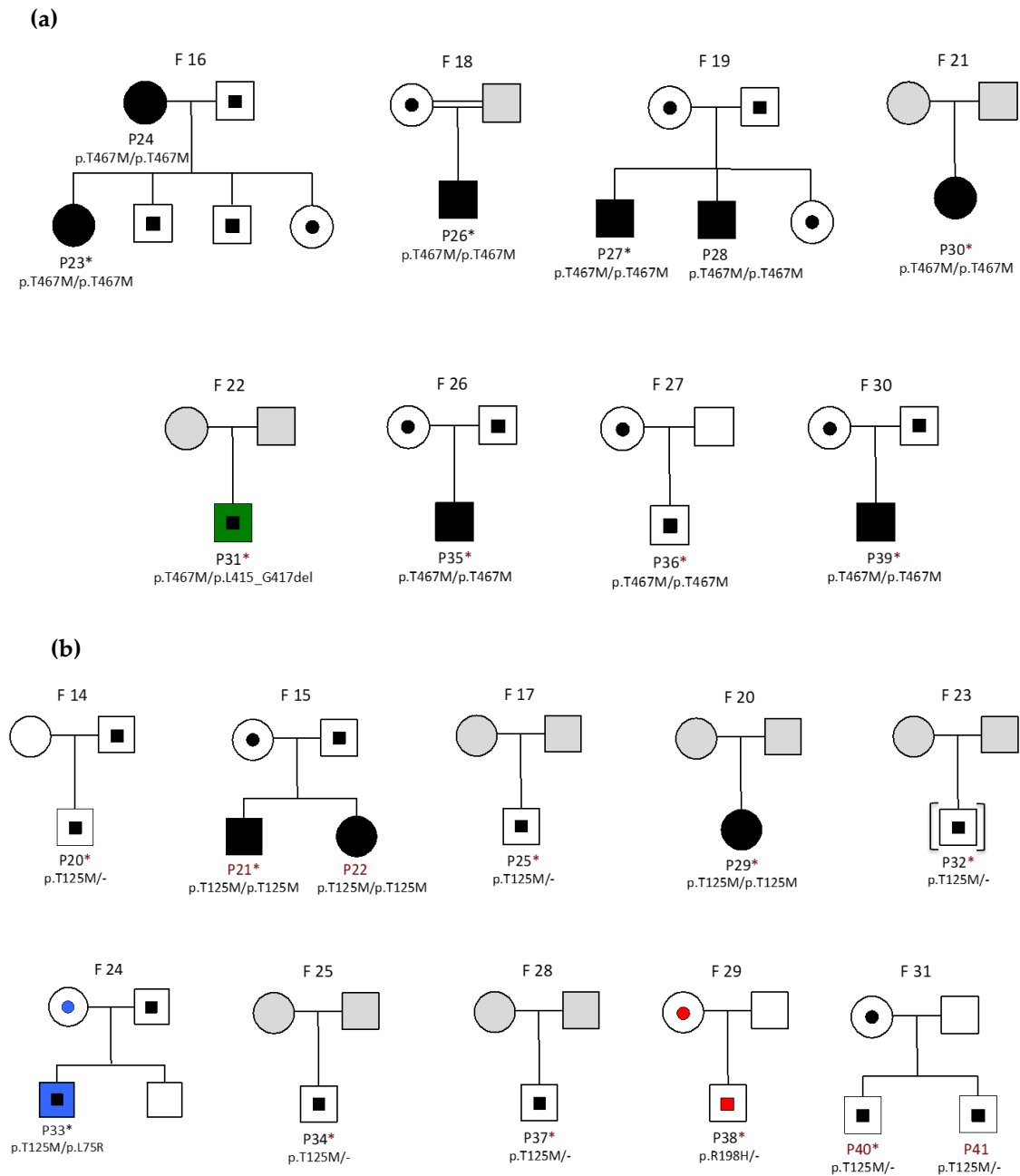


Figure S1. Pedigrees of families with RHUC type 1 (a) and type 2 (b) showing segregation of *SLC22A12* and *SLC2A9* variants, respectively. Asterisks indicate probands in each pedigree. Circles and squares correspond to female and male individuals, respectively. Filled circles and squares represent homozygous or compound heterozygous subjects; filled circles and squares within open frames represent heterozygous individuals; unfilled symbols denote subjects without mutation; grey symbols represent family members who were not available for genetic analysis. Double line indicates paternal consanguinity. F, family. Brackets indicates adoption. Black symbols indicate subjects harbouring the most frequent mutation in RHUC type 1 or RHUC type 2. Green symbols indicate individual

carrying mutation p.(L415_G417del). Blue symbols indicate individuals carrying mutation p.L75R. red symbols represent individuals harbouring novel mutation p.R198H.

Table S1: Primers used for PCR amplification of the variants genotyped for haplotype analysis. Human assembly GRCh38.p13

	Reference SNP	Position	Primers sequences (5' – 3')
SNP_1	rs6855911	chr4:9934286	rs6855911F: CTCCCAATCAAATCCCCTCT rs6855911R: TGAAAAGCCATCTGTGGTCA
SNP_2	rs7442295	chr4:9964756	rs7442295F: GGCTGGGGCTTAAAATCACT rs7442295R: GAGAGGCAAGAGGGCTGTC
SNP_3	rs10011206	chr4:9990331	rs10011206F: ATGAGGGAACGGAAAGTGTG rs10011206R: GCCCTTCAACGTAAGAACCA
SNP_4	rs7696536	chr4:9998612	rs7696536F: GCAGTGTGTTGTCTGCTGGT rs7696536R: GGCTGTTTCTCGTCTGGAGT
SNP_5	rs3756236	chr4:10011839	rs3756236F: GCCAATTGGAGCTAGTCACTG rs3756236R: GAAAAGGGAGGAGGCTCTTG
SNP_6	rs13133766	chr4:10018108	rs13133766F: CATCCATCACTCCACCTTCC rs13133766R: CACATTGTGCCTCTGGTTTG

Table S2. Allele frequencies distribution of the selected SNPs markers in cases and controls.

	Allele	Controls (%)	RH patients (%)
SNP_1 G/A	G	0.5	0.89
SNP_2 A/G	A	0.5	0.86
SNP_3 C/T	C	0.75	0.92
SNP_4 G/T	G	0.75	0.92
SNP_5 T/A	T	0.67	0.94
SNP_6 C/T	C	0.71	0.94