**Table S1.** Genetic variations detected in the full-length *ERMAP* gene of the family

| Location | Nucleotide Change* | dbSNP reference no. | Protein residue change† | Franklin classification‡ | Zygosity | |
|---|---|---|---|---|---|---|
| | | | | | Patient | Sister 1/Sister 2 |
| Intron 1 | c.-121-795T>C | rs6600425 | NA | Benign | Homozygous | Homozygous |
| Intron 2 | c.-6+1397G>A | rs6658001 | NA | Benign | NO | Heterozygous |
| | c.-6+1564T>G | Novel | NA | VUS | NO | Heterozygous |
| | c.-6+1835C>A | rs1471747 | NA | Benign | Homozygous | Homozygous |
| Intron 3 | c.86-85G>T | rs12727498 | NA | Benign | NO | Heterozygous |
| Exon 4§ | c.424C>G | Novel | Gln142Glu | VUS | Homozygous | Heterozygous |
| Intron 4 | c.433+993T>C | rs34721801 | NA | Benign | Homozygous | Heterozygous |
| | c.433+1239A>G | rs11210725 | NA | Benign | Homozygous | Homozygous |
| | c.433+1281T>C | rs11580112 | NA | Benign | NO | Heterozygous |
| | c.433+1548A>G | rs10789427 | NA | Benign | Homozygous | Homozygous |
| | c.433+1763A>G | rs56161641 | NA | Benign | Homozygous | Homozygous |
| | c.434-1588A>G | rs11210726 | NA | Benign | Homozygous | Homozygous |
| | c.434-1409G>A | rs58724795 | NA | Benign | Homozygous | Heterozygous |
| | c.434-1061G>A | rs7548186 | NA | Benign | Homozygous | Homozygous |
| | c.434-874T>C | rs7540604 | NA | Benign | Homozygous | Homozygous |
| | c.434-46T>C | rs4660672 | NA | Benign | Homozygous | Homozygous |
| Intron 5 | c.550+96C>G | rs4660673 | NA | Benign | Homozygous | Homozygous |
| | c.550+197_550+198delCC | rs59816087 | NA | Benign | Homozygous | Homozygous |
| Intron 7 | c.616+719C>T | rs7522285 | NA | Benign | NO | Heterozygous |
| | c.617-398C>A | rs11210728 | NA | Benign | NO | Heterozygous |
| Intron 11 | c.712+250G>A | rs1466549 | NA | Benign | NO | Heterozygous |
| | c.712+542T>C | rs1237396071 | NA | VUS | Homozygous | Heterozygous |
| | c.713-606A>G | rs11210730 | NA | Benign | Homozygous | Homozygous |
| Exon 12 (3' UTR) | c.*645G>A | rs1484324 | NA | Benign | NO | Heterozygous |
| | c.*939G>C | rs10789428 | NA | Benign | NO | Heterozygous |

* relative to NCBI Reference Sequence NM_001017922.1
† relative to NCBI Reference Sequence NP_001017922.1
‡ VUS-variant of uncertain significance[37]
§ The patient and his 2 sisters were homozygous for the reference nucleotide "G" at position c.169 in exon 4 indicative of the SC:1 phenotype.
NA- Not available
NO- Not observed

**Table S2.** Genetic variations detected in the *RHCE* gene of the family

| Location | Nucleotide Change* | dbSNP reference no. | Protein residue change† | Franklin classification | Zygosity | |
|---|---|---|---|---|---|---|
| | | | | | Patient | Sister 1/Sister 2 |
| Promoter | c.-378G>A | rs2281179 | NA | Benign | Homozygous | Homozygous |
| | c.-369C>T | rs2072933 | NA | Benign | Homozygous | Homozygous |
| | c.-296G>A | rs2072932 | NA | Benign | Homozygous | Homozygous |
| | c.-122C>A | rs2072931 | NA | Benign | Homozygous | Homozygous |
| Intron 3 | c.486+157C>A | rs28631635 | NA | Benign | Homozygous | Homozygous |
| Intron 4 | c.635-435C>T | rs607116 | NA | Benign | Homozygous | Homozygous |
| Exon 5 | c.676G>C | rs609320 | Ala226Pro (“E”) | Benign | Homozygous | Homozygous |
| Intron 5 | c.801+101G>A | rs636889 | NA | Benign | Homozygous | Homozygous |
| | c.802-281G>T | rs760938 | NA | Benign | Homozygous | Homozygous |
| Intron 8 | c.1154-222_1154-221insAAATA | rs71014358 | NA | Benign | Homozygous | Homozygous |

* relative to NCBI Reference Sequence NM_020485.4
† relative to NCBI Reference Sequence NP_065231.3
NA- Not available

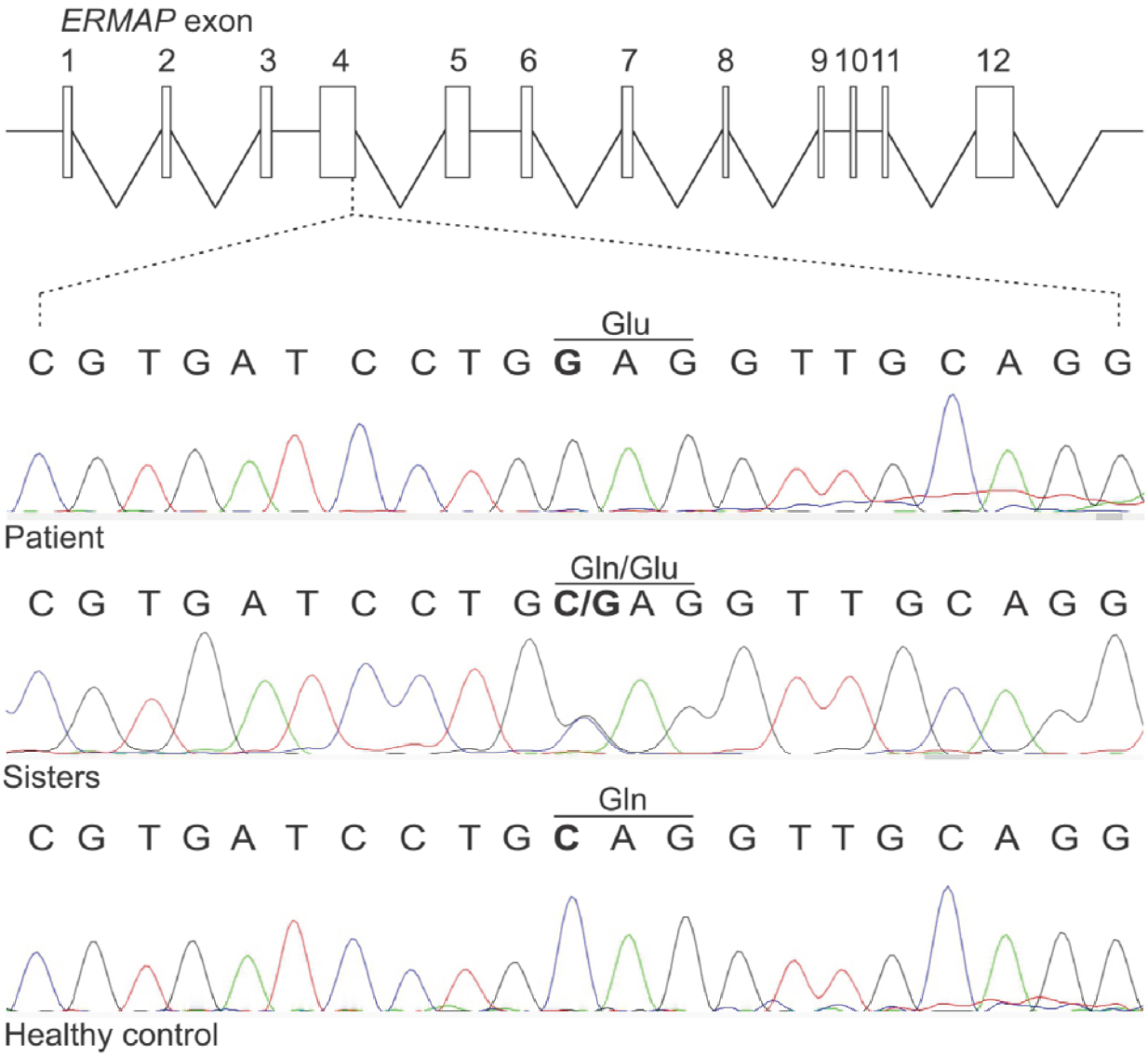
ERMAP exon
1 2 3 4 5 6 7 8 9 10 11 12
Glu
C G T G A T C C T G G A G G T T G C A G G
Patient
Gln/Glu
C G T G A T C C T G C/G A G G T T G C A G G
Sisters
Gln
C G T G A T C C T G C A G G T T G C A G G
Healthy control

**Fig. S1**