**Supp.Table S1: CASQ1 intragenic SNPs analysis**

| SNP | Family 1 | | | Family 2 | | | Family 3 | | | Family 4 | | | Sporadic | HAPL |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | III:5 | II:4 | HAPL | II:1 | I:1 | HAPL | II:3 | III:2 | HAPL | II:2 | I:1 | HAPL | | |
| rs617698 | G/G | A/G | **G** | A/G | G/G | **G** | G/G | A/G | **G** | A/G | G/G | **G** | G/G | **G** |
| rs61803844 | G/G | G/G | **G** | A/G | G/G | **G** | G/G | G/G | **G** | A/G | G/G | **G** | A/G | **?** |
| rs617599 | A/A | A/G | **A** | A/G | A/A | **A** | A/G | A/G | **?** | A/G | A/A | **A** | A/A | **A** |
| rs3747621 | T/T | T/C | **T** | T/T | T/T | **T** | T/C | T/C | **?** | T/T | T/T | **T** | T/C | **?** |
| rs3747622 | G/G | G/G | **G** | G/C | G/G | **G** | G/G | G/G | **G** | G/C | G/G | **G** | G/C | **?** |
| rs3747623 | T/T | T/C | **T** | T/C | T/T | **T** | T/C | T/C | **?** | T/C | T/T | **T** | C/C | **C** |
| **c.731A>G** | +/- | +/- | + | +/- | +/- | + | +/- | -/- | + | +/- | +/- | + | +/- | + |
| rs822450 | A/G | A/G | **?** | A/G | A/A | **A** | A/G | A/G | **?** | A/G | A/A | **A** | G/G | **G** |

+/- = individuals heterozygous for the c.731A>G variant; -/- = individuals homozygous for the wild type allele; HAPL = Haplotype phased with the c.731A>G variant. Question marks indicate heterozygous SNPs in both individuals in a family. The differences between haplotypes are highlighted in red and blue.