

Table SII. Distribution of R* (rs11547464, rs1805007, rs1805009, rs1805006) and r *(rs1805005, rs2228479) MC1R polymorphisms in association with CDKN2A status in familial melanoma patients

| MC1R status | CDKN2A status | | | p-value (overall) |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------|------------------------------------------|----------------------------|-------------------|
| | CDKN2A ⁺ (n = 18) n (%) | CDKN2A ⁻ (n = 24) n (%) | Total (n = 42) n (%) | |
| R: Homozygotes + heterozygotes (at least 1 of: <i>rs11547464ga</i> , <i>rs1805007ct</i> , <i>rs1805009gc</i> , <i>rs1805006ca</i>) | 3 (16.7) | 3 (12.5) | 6 (14.3) | 0.164** |
| r: Homozygotes + heterozygotes (at least 1 of <i>rs1805005gt</i> , <i>rs2228479ga</i>) | 10 (55.6) | 11 (45.8) | 21 (50.0) | |
| R & r – Homozygotes + heterozygotes (at least 1 of: <i>rs11547464ga</i> , <i>rs1805007ct</i> , <i>rs1805009gc</i> , <i>rs1805006ca</i> , <i>rs1805005gt</i> , <i>rs2228479ga</i>) | 2 (11.1) | 0 (0) | 2 (4.8) | |
| WT (%) | 3 (16.7) | 10 (41.7) | 13 (31.0) | |

**Fisher's exact test. p-value were obtained by testing for a difference in proportions between CDKN2A⁺ and CDKN2A⁻ with respect to a MC1R status using Fisher's exact test.
*Amino acid change of R and r: *rs11547464* (Arg142His), *rs1805007* (Arg151Cys), *rs1805009* (Asp294His), *rs1805006* (Asp84Glu), *rs1805005* (Val60Leu), *rs2228479* (Val92Met), *rs11547464* (Arg142His).