**Appendix 6**. Variants selected for segregation studies.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| RefSeq\_ID | Gene | Variant | Protein change | Frequency | SIFT | PolyPhen | MutationTaster |
| NM\_144663.1 | *C11orf40* | c.599-600dupGT | p.Met201ValfsX211 | <0.01 | - | - | - |
| NM\_001837.3 | *CCR3* | c.478G>A | p.Val160Met | <0.01 | deleterious | probably damaging | disease causing |
| NM\_001852.3 | *COL9A2* | c.150C>T | p.Pro51Leu | <0.01 | n/a | n/a | n/a |
| NM\_153002.2 | *GPR156* | c.2426T>C | p.Leu809Ser | <0.01 | deleterious | probably damaging | disease causing |
| NM\_015112.2 | *MAST2* | c.3186G>A | p.Ser1062Ser | <0.01 | - | - | - |
| NM\_001137604.2 | *POLR1B* | c.1865C>G | p.Pro622Arg | <0.01 | deleterious | probably damaging | disease causing |
| NM\_006583.2 | *RRH* | c.900-4G>A | - | <0.01 | - | - |  |
| NM\_022112.2 | *ABCA7* | c.109G>A | p.Arg37His | New | n/a | n/a | n/a |
| NM\_001276713.1 | *ANKDD1B* | c.567-571delCAGTG | p.Ala190fsX201 | New | - | - | - |
| NM\_018186.2 | *C1orf112* | c.171A>G | p.Gln57Gln | New | - | - | - |
| **NM\_001102608.1** | ***COL6A6*** | **c.307G>A** | **p.Gly103Arg** | **New** | **deleterious** | **probably damaging** | **disease causing** |
| NM\_000308.2 | *CTSA* | c.459C>A | p.Asn153Lys | New | n/a | n/a | n/a |
| NM\_181453.3 | *GCC2* | c.3793A>G | p.Ile1265Val | New | deleterious | probably damaging | disease causing |
| NM\_002375.4 | *MAP4* | c.4777C>T | p.Leu1593Phe | New | n/a | n/a | n/a |
| NM\_001127178.1 | *PIGG* | c.661delC | p.Pro221GlnfsX222 | New | - | - | - |
| NM\_014330.3 | *PPP1R15A* | c.484A>T | p.Lys162X | New | n/a | n/a | n/a |
| NM\_175732.2 | *PTPMT1* | c.361A>C | p.Thr121Pro | New | n/a | n/a | n/a |
| NM\_001031709.2 | *RNLS* | c.871T>A | p.Ser291Thr | New | deleterious | probably damaging | disease causing |
| NM\_015540.3 | *RPAP1* | c.1877G>A | p.Arg626His | New | deleterious | probably damaging | disease causing |
| NM\_022112.2 | *TP53AIP1* | c.169G>A | p.Gly57Ser | New | n/a | n/a | n/a |
| NM\_003442.5 | *ZNF143* | c.791G>A | p.Arg264Gln | New | n/a | n/a | n/a |
| **Bold type** line shows the only identified genetic variant that co-segregated with RP in the RPT65 family. |