

Supplemental table 2. The detailed results of *in silico* molecular genetic analysis of 245 detected variants

| Vt no | Position | dbSNP ID | Ref | Alt | Nucleotide change | Amino acid change | Effect | Pathogenicity | Primal screening Method | | | | Co-segregation | Total number of affected alleles in this study | allele Frequency | | Pathogenicity prediction programs | | | | Previous Reports | References | Supplemental references |
|-------|----------|-------------|-----|-----|-------------------|-------------------|--|---------------|-------------------------|-----|------|----|----------------|--|------------------|----------------------|-----------------------------------|-----------|-----------------|----------------------------------|------------------|------------|-------------------------|
| | | | | | | | | | NGS | GCA | SSCP | DS | | | ExAC browser | 1000 genomes browser | SIFT | Polyphen2 | Mutation taster | HSF | | | |
| 1 | 94586601 | | T | C | c.1A>G | p.Met1Val | Missense variant with significant splice site alteration | LP | ✓ | | ✓ | | 3 | ND | ND | NA | NA | DC | | Briggs CE, <i>et al.</i> | | 1 | |
| 2 | 94586586 | | G | A | c.16C>T | p.Gln6Ter | Stop | LP | ✓ | | | | 1 | ND | ND | NA | NA | DC | | This study | | | |
| 3 | 94586580 | | G | TG | c.21_22insA | p.Gln8ThrfsTer46 | Frame shift | LP | ✓ | | | | 1 | ND | ND | NA | NA | NA | | Fujinami K, <i>et al.</i> | 5 | | |
| 4 | 94586570 | | A | G | c.32T>C | p.Leu11Pro | Missense | LP | ✓ | | ✓ | | 1 | 8.237E-06 | ND | Del | PRD | DC | | Rozet JM, <i>et al.</i> | | 2 | |
| 5 | 94586557 | | C | T | c.45G>A | p.Trp15Ter | Stop | LP | ✓ | | | | 2 | ND | ND | NA | NA | DC | | Duncker T, <i>et al.</i> | | 3 | |
| 6 | 94586550 | rs121909205 | G | A | c.52C>T | p.Arg18Trp | Missense | LP | ✓ | | | | 1 | 2.471E-05 | 0.0004 | Del | PRD | DC | | Riveiro-Alvarez R, <i>et al.</i> | | 4 | |
| 7 | 94578623 | | C | G | c.67-1G>C | Splice acceptor | Splice site alteration | LP | | | ✓ | | 1 | ND | ND | NA | NA | NA | PRA | This study | | | |
| 8 | 94578524 | | C | T | c.160+5G>A | Splice region | Splice site alteration | LP | | | ✓ | | 2 | ND | ND | NA | NA | NA | PRA | This study | | | |
| 9 | 94578524 | | C | G | c.160+5G>C | Splice region | Splice site alteration | LP | | | ✓ | | 1 | ND | ND | NA | NA | NA | PRA | This study | | | |
| 10 | 94577135 | rs150774447 | C | T | c.161G>A | p.Cys54Tyr | Missense | LP | ✓ | ✓ | | | 2 | 0.0000165 | ND | Del | PRD | DC | | Lewis RA, <i>et al.</i> | | 5 | |
| 11 | 94577136 | | C | T | c.161-1G>A | Splice acceptor | Splice site alteration | LP | | | ✓ | ✓ | 1 | ND | ND | NA | NA | NA | PRA | This study | | | |
| 12 | 9457716 | | CT | T | c.180delG | p.Ala60AlafsTer18 | Frame shift | LP | ✓ | | | | 1 | ND | ND | NA | NA | NA | | Fujinami K, <i>et al.</i> | 17 | | |
| 13 | 94577112 | | C | G | c.183G>C | p.Met61Ile | Missense | LP | ✓ | | ✓ | | 2 | ND | ND | Tol | PRD | DC | | This study | | | |
| 14 | 94577102 | | C | A | c.194G>A | p.Gly65Glu | Missense | LP | ✓ | | | | 2 | 8.244E-06 | ND | Del | PRS | DC | | Genead MA, <i>et al.</i> | | 6 | |
| 15 | 94577059 | rs149381884 | A | G | c.237T>C | p.Asn79Asn | Synonymous variant with potential splice site alteration | LLP | ✓ | | | | 1 | 0.0009505 | 0.0028 | NA | NA | DC | NSA | This study | | | |
| 16 | 94577013 | | A | G | c.283T>C | p.Ser95Pro | Missense | LP | ✓ | | | | 2 | ND | ND | Del | PRD | DC | | This study | | | |
| 17 | 94577010 | rs61748529 | T | C | c.286A>G | p.Asn96Asp | Missense | LP | | | ✓ | | 1 | 2.506E-05 | ND | Del | Ben | DC | | Ducroq D, <i>et al.</i> | | 7 | |
| 18 | 94576993 | rs61751413 | C | T | c.302+1G>A | Splice donor | Splice site alteration | LP | ✓ | | | | 1 | 0.0000335 | ND | NA | NA | NA | PRA | Zemant J, <i>et al.</i> | | 8 | |
| 19 | 94574274 | | G | C | c.303-2A>G | Splice acceptor | Splice site alteration | LP | ✓ | | | | 1 | ND | ND | NA | NA | NA | PRA | This study | | | |
| 20 | 94574152 | | A | T | c.423T>A | p.Thr141Thr | Synonymous variant with potential splice site alteration | LLP | | | ✓ | | 1 | 8.237E-06 | ND | NA | NA | DC | NSA | This study | | | |
| 21 | 94568687 | | G | A | c.454C>T | p.Arg152Ter | Stop | LP | ✓ | ✓ | | | 3 | 8.275E-06 | ND | NA | NA | DC | | Rivera A, <i>et al.</i> | 20 | | |
| 22 | 94568686 | rs80309162 | C | T | c.455G>A | p.Arg152Gln | Missense | LLP | | ✓ | | | 1 | 0.002399 | 0.0016 | Tol | Ben | DC | NSA | Paloma E, <i>et al.</i> | | 9 | |
| 23 | 94568675 | rs112467008 | T | C | c.466A>G | p.Ile156Val | Missense | LLP | | ✓ | ✓ | | 2 | 0.001315 | 0.0024 | Tol | Ben | DC | NSA | Fujinami K, <i>et al.</i> | 17 | | |
| 24 | 94564548 | | T | A | c.571-1G>T | Splice acceptor | Splice site alteration | LP | | | ✓ | ✓ | 1 | ND | ND | NA | NA | NA | PRA | Duncker T, <i>et al.</i> | | 56 | |
| 25 | 94564549 | | T | A | c.571-2A>T | Splice acceptor | Splice site alteration | LP | ✓ | | | | 1 | 1.043E-05 | ND | NA | NA | NA | PRA | Verdina T, <i>et al.</i> | | 10 | |
| 26 | 94564500 | rs61748536 | G | C | c.618C>G | p.Ser206Arg | Missense | LLP | ✓ | | | | 1 | 0.001371 | 0.0042 | Tol | PRD | DC | NSA | Zemant J, <i>et al.</i> | | 58 | |
| 27 | 94564484 | rs61750200 | G | A | c.634C>T | p.Arg212Cys | Missense | LP | ✓ | ✓ | ✓ | ✓ | 8 | 0.0001166 | ND | Del | PRD | DC | | Fujinami K, <i>et al.</i> | 11 | | |
| 28 | 94564463 | | T | A | c.655A>T | p.Arg219Ter | Stop | LP | ✓ | ✓ | | | 1 | ND | ND | NA | NA | DC | | Fujinami K, <i>et al.</i> | 17 | | |

| | | | | | | | | | | | | | | | | | | | | | |
|----|----------|-----------------|----------------------|---|---------------------|--------------------|--|-----|---|---|---|-----------|-----------|-----------|-----|-----|-----------------------------|----------------------------|----|---------------------------|----|
| 29 | 94564462 | rs6174 8537 | C | G | c.656G>C | p.Arg219Thr | Missense | LP | | ✓ | 1 | 4.133E-05 | ND | Del | Ben | DC | Jaakson K, <i>et al.</i> | 11 | | | |
| 30 | 94564460 | | G | A | c.658C>T | p.Arg220Cys | Missense | LP | ✓ | ✓ | 2 | ND | ND | Tol | POD | Pol | Cideciyan AV, <i>et al.</i> | 12 | | | |
| 31 | 94564358 | | AAGG CACA CCAT | G | c.760_768+25delTTCC | p.Phe2541GlnfsTer8 | Frame shift | LP | ✓ | | 1 | ND | ND | NA | NA | NA | This study | | | | |
| 32 | 94563997 | rs7937 2932 | A | C | c.768+353T>C | Deep intron | Deep intronic variant with uncertain effect | LLP | ✓ | | 1 | ND | ND | NA | NA | NA | NSA | Zemant J, <i>et al.</i> | 23 | | |
| 33 | 94564350 | | C | A | c.768G>T | p.Val256Val | Synonymous variant with significant splice site alteration | LP | ✓ | ✓ | 6 | 0.0001072 | ND | NA | NA | DC | PRA | Fujinami K, <i>et al.</i> | 17 | | |
| 34 | 94548931 | | CA | C | c.834delT | p.Ser278SerfsTer22 | Frame shift | LP | ✓ | | 2 | 8.246E-06 | ND | NA | NA | DC | | This study | | | |
| 35 | 94546283 | | A | G | c.859-9T>C | Splice region | Splice site alteration | LP | ✓ | ✓ | 2 | ND | ND | NA | NA | NA | POA | Zemant J, <i>et al.</i> | 23 | | |
| 36 | 94546265 | | G | A | c.868C>T | p.Arg290Trp | Missense | LP | | ✓ | ✓ | 2 | 3.309E-05 | ND | Del | PRD | Pol | Yzer S, <i>et al.</i> | 13 | | |
| 37 | 94546261 | rs1905 40405 | G | A | c.872C>T | p.Pro291Leu | Missense | LP | | ✓ | 2 | 0.0005375 | 0.0004 | Del | PRD | Pol | Ernest PJ, <i>et al.</i> | 14 | | | |
| 38 | 94546247 | | AG | A | c.885delC | p.Asp295AspfsTer5 | Frame shift | LP | | ✓ | 1 | 4.127E-05 | ND | NA | NA | NA | | Zemant J, <i>et al.</i> | 58 | | |
| 39 | 94546118 | | A | C | c.1015T>G | p.Trp339Gly | Missense | LP | | ✓ | 1 | ND | ND | Tol | PRD | DC | | Yatsenko AN, <i>et al.</i> | 15 | | |
| 40 | 94546114 | | G | C | c.1019A<G | p.Tyr340Cys | Missense | LP | | ✓ | 1 | 1.648E-05 | ND | Tol | PRD | DC | | This study | | | |
| 41 | 94546111 | | G | T | c.1022A>G | p.Glu341Gly | Missense | LP | | ✓ | 1 | ND | ND | Del | Ben | DC | | Huang WC, <i>et al.</i> | 16 | | |
| 42 | 94546099 | | T | C | c.1034A>G | p.Tyr345Cys | Missense | LP | ✓ | | 1 | ND | ND | Tol | POD | DC | | This study | | | |
| 43 | 94546047 | | A | T | c.1086T>A | p.Tyr362Ter | Stop | LP | ✓ | ✓ | 2 | ND | ND | NA | NA | DC | | Rivera A, <i>et al.</i> | 20 | | |
| 44 | 94544916 | | T | G | c.1201A>C | p.Thr401Pro | Missense | LP | | ✓ | 1 | ND | ND | Tol | PRD | DC | | This study | | | |
| 45 | 94544235 | rs1380 44729 | G | A | c.1267C>T | p.His423Tyr | Missense | LLP | ✓ | ✓ | 2 | 0.0001483 | ND | Tol | Ben | Pol | NSA | This study | | | |
| 46 | 94544185 | rs6175 2391 | C | T | c.1317G>A | p.Trp439Ter | Stop | LP | ✓ | ✓ | 3 | 8.236E-06 | ND | NA | NA | DC | | Rivera A, <i>et al.</i> | 20 | | |
| 47 | 94543389 | rs1800 548 | C | T | c.1411G>A | p.Glu471Lys | Missense | LLP | | ✓ | 1 | 0.000939 | ND | Tol | Ben | DC | NSA | Scholl HP, <i>et al.</i> | 21 | | |
| 48 | 94543271 | | A | G | c.1529T>C | p.Leu510Pro | Missense | LP | | ✓ | 2 | ND | ND | Tol | PRD | DC | | This study | | | |
| 49 | 94543269 | | G | A | c.1531C>T | p.Arg511Cys | Missense | LP | ✓ | | 2 | 0.0002158 | ND | Tol | PRD | DC | | Zemant J, <i>et al.</i> | 58 | | |
| 50 | 94528873 | | C | T | c.1556G>A | p.Cys519Tyr | Missense | LP | | ✓ | 1 | ND | ND | Del | PRD | DC | | This study | | | |
| 51 | 94528854 | | A | C | c.1574T>C | p.Phe525Ser | Missense | LP | | ✓ | 2 | ND | ND | Del | PRD | DC | | Briggs CE, <i>et al.</i> | 1 | | |
| 52 | 94528818 | rs6175 2395 | C | T | c.1610G>A | p.Arg537His | Missense | LP | ✓ | ✓ | 2 | 0.001969 | 0.0008 | Tol | PRD | DC | | This study | | | |
| 53 | 94528806 | rs6175 1392 | A | G | c.1622T>C | p.Leu541Pro | Missense | LP | ✓ | ✓ | ✓ | ✓ | 18 | 0.0001235 | ND | Tol | PRD | DC | | Fujinami K, <i>et al.</i> | 11 |
| 54 | 94528780 | rs6174 8558 | C | T | c.1648G>A | p.Gly550Arg | Missense | LP | ✓ | | 3 | 8.237E-06 | ND | Del | PRD | DC | | Shroyer NF, <i>et al.</i> | 17 | | |
| 55 | 94528774 | rs1455 25174 | C | T | c.1654G>A | p.Val552Ile | Missense | LLP | ✓ | | 1 | 0.002636 | 0.0006 | Tol | Ben | DC | NSA | Rivera A, <i>et al.</i> | 20 | | |
| 56 | 94528713 | rs6174 8559 | C | T | c.1715G>A | p.Arg572Gln | Missense | LP | | ✓ | 1 | 4.942E-05 | 0.0002 | Del | PRD | DC | | Lewis RA, <i>et al.</i> | 5 | | |
| 57 | 94528702 | | C | G | c.1726G>C | p.Asp576His | Missense | LP | | ✓ | 1 | 8.236E-06 | ND | Del | PRD | DC | | Downs K, <i>et al.</i> | 18 | | |
| 58 | 94528671 | | T | C | c.1757A>G | p.Asp586Gly | Missense | LP | ✓ | | 2 | ND | ND | Del | POD | DC | | Fujinami K, <i>et al.</i> | 5 | | |
| 59 | 94528668 | | C | T | c.1760G>A | p.Arg587Lys | Missense variant with significant splice site alteration | LP | ✓ | | 1 | ND | ND | Tol | POD | DC | PRA | Fujinami K, <i>et al.</i> | 5 | | |
| 60 | 94528251 | | C | T | c.1819G>A | p.Gly607Arg | Missense | LP | ✓ | ✓ | 4 | 2.502E-05 | ND | Del | PRD | DC | | Scholl HP, <i>et al.</i> | 21 | | |

| | | | | | | | | | | | | | | | | | | | | | |
|----|----------|-------------|-----|----|------------------|--------------------|------------------------|-----|---|---|---|---|----|-----------|--------|-----|-----|-----|-----|---------------------------------|----|
| 61 | 94528248 | . | A | T | c.1822T>A | p.Phe608Ile | Missense | LP | | ✓ | | | 1 | 8.341E-06 | ND | Del | PRD | DC | | Lewis RA, <i>et al.</i> | 5 |
| 62 | 94528217 | | C | T | c.1853G>A | p.Gly618Glu | Missense | LP | | ✓ | | | 1 | ND | ND | Del | PRD | DC | | Ernest PJ, <i>et al.</i> | 14 |
| 63 | 94528164 | | G | A | c.1906C>T | p.Gln636Ter | Stop | LP | ✓ | ✓ | | ✓ | 3 | ND | ND | NA | NA | DC | | Zemant J, <i>et al.</i> | 58 |
| 64 | 94544918 | | G | A | c.1919C>T | p.Pro640Leu | Missense | LP | ✓ | | ✓ | | 2 | ND | ND | Del | PRD | DC | | Miraldi Utz V, <i>et al.</i> | 19 |
| 65 | 94528148 | | C | G | c.1922G>C | p.Cys641Ser | Missense | LP | | ✓ | | | 1 | ND | ND | Tol | POD | DC | | Steniri S, <i>et al.</i> | 20 |
| 66 | 94528143 | rs143548435 | C | T | c.1927G>A | p.Val643Met | Missense | LP | ✓ | | ✓ | | 3 | 0.001618 | 0.0074 | Del | PRD | DC | | Webster AR, <i>et al.</i> | 21 |
| 67 | 94528132 | rs61752401 | C | T | c.1937+1G>A | Splice donor | Splice site alteration | LP | | | ✓ | | 2 | 8.266E-06 | ND | NA | NA | NA | PRA | Eandi CM, <i>et al.</i> | 22 |
| 68 | 94526296 | | G | A | c.1957C>T | p.Arg653Cys | Missense | LP | ✓ | | ✓ | | 3 | 1.035E-05 | ND | Del | PRD | DC | | Fujinami K, <i>et al.</i> | 12 |
| 69 | 94526289 | | A | C | c.1964T>G | p.Phe655Cys | Missense | LP | | | ✓ | | 1 | 0.0006465 | ND | Del | PRD | DC | | Downs K, <i>et al.</i> | 18 |
| 70 | 94526244 | | GAC | C | c.2005_2006delAT | p.Met669AspfsTer96 | Frame shift | LP | | | ✓ | ✓ | 1 | ND | ND | NA | NA | NA | | Cideciyan AV, <i>et al.</i> | 23 |
| 71 | 94526230 | | C | T | c.2023G>A | p.Val675Ile | Missense | LP | ✓ | | | | 1 | 4.994E-05 | 0.0002 | Del | PRD | DC | | Duncker T, <i>et al.</i> | 56 |
| 72 | 94526212 | rs61749423 | G | A | c.2041C>T | p.Arg681Ter | Stop | LP | ✓ | | ✓ | ✓ | 5 | 2.483E-05 | ND | NA | NA | DC | | Bertelsen M, <i>et al.</i> | 24 |
| 73 | 94526091 | | A | C | c.2160+2T>G | Splice donor | Splice site alteration | LP | ✓ | | | | 1 | ND | ND | NA | NA | NA | | Mullins RF, <i>et al.</i> | 25 |
| 74 | 94522239 | rs61751395 | A | T | c.2300T>A | p.Val767Asp | Missense | LP | ✓ | | ✓ | | 3 | 8.38E-06 | ND | Del | PRD | DC | | Briggs CE, <i>et al.</i> | 1 |
| 75 | 94520872 | | C | T | c.2383-1G>A | Splice acceptor | Splice site alteration | LP | ✓ | | | | 1 | ND | ND | NA | NA | NA | PRA | This study | |
| 76 | 94520853 | | C | T | c.2401G>A | p.Ala801Thr | Missense | LP | ✓ | | | | 1 | ND | ND | Del | PRD | DC | | Zemant J, <i>et al.</i> | 58 |
| 77 | 94520801 | rs61750202 | C | T | c.2453G>A | p.Gly818Glu | Missense | LP | | | ✓ | ✓ | 2 | 0.0001978 | ND | Del | PRD | DC | | Webster AR, <i>et al.</i> | 21 |
| 78 | 94520793 | rs61749433 | A | T | c.2461T>A | p.Trp821Arg | Missense | LP | | | ✓ | | 1 | 8.241E-06 | ND | Del | Del | DC | | Lewis RA, <i>et al.</i> | 5 |
| 79 | 94520732 | | T | G | c.2522A>C | p.Gln841Pro | Missense | LLP | ✓ | | | | 1 | ND | ND | Tol | Ben | Pol | NSA | This study | |
| 80 | 94520708 | rs61749435 | A | G | c.2546T>C | p.Val849Ala | Missense | LLP | | | ✓ | | 1 | 0.001335 | 0.0046 | Del | Ben | Pol | NSA | Webster AR, <i>et al.</i> | 21 |
| 81 | 94520693 | | C | CC | c.2560_2561insG | p.Ala854GlyfsTer5 | Frame shift | LP | | | ✓ | | 1 | ND | ND | NA | NA | NA | | This study | |
| 82 | 94520690 | rs61752406 | C | T | c.2564G>A | p.Trp855Ter | Stop | LP | ✓ | | ✓ | | 4 | 3.296E-05 | ND | NA | NA | DC | | Rosenberg T, <i>et al.</i> | 26 |
| 83 | | | NA | NA | exon7del | Large deletion | Large exonic deletion | LP | | | ✓ | | 1 | ND | ND | NA | NA | NA | | Cideciyan AV, <i>et al.</i> | 23 |
| 84 | 94517254 | rs121909203 | C | G | c.2588G>C | p.Gly863Ala | Missense | LP | ✓ | ✓ | ✓ | ✓ | 40 | 0.005072 | 0.0012 | Del | PRD | DC | | Fujinami K, <i>et al.</i> | 17 |
| 85 | 94517234 | | G | A | c.2608C>T | p.Pro870Ser | Missense | LP | ✓ | | | | 1 | ND | ND | Tol | POD | DC | | Zemant J, <i>et al.</i> | 8 |
| 86 | 94517233 | | G | A | c.2609C>T | p.Pro870Leu | Missense | LP | ✓ | | | | 1 | 8.303E-06 | ND | Del | PRD | DC | | This study | |
| 87 | 94512589 | rs61749444 | A | G | c.2804T>C | p.Val935Ala | Missense | LP | | | ✓ | | 1 | 8.262E-06 | ND | Del | PRD | DC | | Briggs CE, <i>et al.</i> | 1 |
| 88 | 94512580 | rs149071415 | A | G | c.2813T>C | p.Phe938Ser | Missense | LP | | | ✓ | | 1 | 4.128E-05 | ND | Del | PRD | DC | | Duncker T, <i>et al.</i> | 41 |
| 89 | 94512574 | rs144995371 | G | C | c.2819C>G | p.Pro940Arg | Missense | LLP | | | ✓ | | 1 | 0.0002394 | ND | Tol | Ben | Pol | NSA | Souied EH, <i>et al.</i> | 27 |
| 90 | 94512532 | | T | G | c.2861A>C | p.Tyr954Ser | Missense | LP | ✓ | | | | 1 | ND | ND | Tol | PRD | DC | | Aguirre-Lamban J, <i>et al.</i> | 28 |
| 91 | 94512526 | | G | T | c.2868C>A | p.Asn956Lys | Missense | LP | | | ✓ | | 1 | ND | ND | Del | Ben | DC | | This study | |
| 92 | 94512499 | | T | C | c.2894A>G | p.Asn965Ser | Missense | LP | | ✓ | ✓ | | 4 | 0.0001731 | ND | Del | PRD | DC | | Rosenberg T, <i>et al.</i> | 26 |

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|----|----------|-----------------|--------------------|-----------|----------------------|---------------------|-----|---|---|----|-----------|--------|-----|-----|----|----------------------------------|-----------------------------|----|
| 93 | 94512481 | G | T | c.2912C>A | p.Thr971Asn | Missense | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | Webster AR, <i>et al.</i> | 21 | |
| 94 | 94512478 | rs6174 9451 | G | A | c.2915C>A | p.Thr972Asm | LP | | ✓ | 2 | 1.649E-05 | ND | Del | PRD | DC | Lewis RA, <i>et al.</i> | 5 | |
| 95 | 94510277 | rs1478 26775 | G | A | c.2942C>T | p.Pro981Leu | LP | ✓ | | 1 | 8.237E-06 | ND | Del | PRD | DC | Fujinami K, <i>et al.</i> | 17 | |
| 96 | 94510271 | | G | A | c.2948C>T | p.Thr983Ile | LP | | ✓ | 1 | ND | ND | Del | PRD | DC | This study | | |
| 97 | 94514477 | rs6174 9440 | G | A | c.2690C>T | p.Thr897Ile | LP | ✓ | | 1 | 0.001138 | 0.0016 | Tol | PRD | DC | Simonelli F, <i>et al.</i> | 29 | |
| 98 | 94510253 | rs1392 96587 | A | G | c.2966T>C | p.Val989Ala | LP | ✓ | ✓ | 5 | 0.0002965 | 0.0016 | Del | Ben | DC | Webster AR, <i>et al.</i> | 21 | |
| 99 | 94510248 | rs1474 84266 | C | G | c.2971G>C | p.Gly991Arg | LP | ✓ | ✓ | 3 | 0.0007166 | 0.0016 | Del | PRD | DC | Yatsenko AN, <i>et al.</i> | 15 | |
| ## | 94510242 | rs2818 65401 | UIG1 AACT TT | T | c.2977_2984delGACAT | p.Asp993AsnfsTer27 | LP | ✓ | | 2 | ND | ND | NA | NA | NA | Webster AR, <i>et al.</i> | 21 | |
| ## | 94510212 | | TC | TGC | c.3007_3008insC | p.Gln1003ArgfsTer20 | LP | ✓ | | 1 | ND | ND | NA | NA | NA | This study | | |
| ## | 94510178 | | A | C | c.3041T>G | p.Leu1014Arg | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | Webster AR, <i>et al.</i> | 21 | |
| ## | 94509032 | | C | T | c.3051-1G>A | Splice acceptor | LP | | ✓ | 1 | ND | ND | NA | NA | NA | PRA | Cideciyan AV, <i>et al.</i> | 23 |
| ## | 94509026 | | G | A | c.3056C>T | p.Thr1019Met | LP | ✓ | ✓ | 2 | 4.133E-05 | ND | Del | PRD | DC | Rozet JM <i>et al.</i> | 2 | |
| ## | 94509018 | rs6174 9459 | C | T | c.3064G>A | p.Glu1022Lys | LP | | ✓ | 1 | 8.259E-06 | ND | Del | PRD | DC | Stenirri S, <i>et al.</i> | 20 | |
| ## | 94508969 | rs6175 1374 | G | A | c.3113C>T | p.Ala1038Val | LLP | ✓ | ✓ | 27 | 0.001426 | 0.0008 | Tol | Ben | DC | NSA | Allikmets R, <i>et al.</i> | 30 |
| ## | 94508455 | | C | A | c.3191-1G>T | Splice acceptor | LP | ✓ | | 1 | ND | ND | NA | NA | NA | PRA | Fujinami K, <i>et al.</i> | 17 |
| ## | 94508433 | | G | GAC | c.3210_3211insGT | p.Ser1071CysfsTer14 | LP | ✓ | ✓ | 5 | 3.296E-05 | ND | NA | NA | NA | Allikmets R, <i>et al.</i> | 2 | |
| ## | 94508421 | | G | T | c.3224C>A | p.Ala1075Asp | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | This study | | |
| ## | 94508386 | rs6175 1398 | C | T | c.3259G>A | p.Glu1087Lys | LP | ✓ | ✓ | 4 | 3.296E-05 | ND | Del | PRD | DC | Fujinami K, <i>et al.</i> | 12 | |
| ## | 94508385 | | T | C | c.3260A>G | p.Glu1087Gly | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | This study | | |
| ## | 94508358 | | G | A | c.3287C>T | p.Ser1096Leu | LP | ✓ | | 1 | 8.24E-06 | ND | Del | PRD | DC | This study | | |
| ## | 94508353 | | G | A | c.3292C>T | p.Arg1098Cys | LP | ✓ | ✓ | 3 | 3.296E-05 | ND | Del | PRD | DC | Fujinami K, <i>et al.</i> | 17 | |
| ## | 94508340 | rs1436 89372 | T | A | c.3305A>T | p.Asp1102Val | LP | ✓ | | 1 | 8.241E-06 | ND | Del | POD | DC | Ernest PJ, <i>et al.</i> | 14 | |
| ## | 94508333 | | A | G | c.3311T>C | p.Leu1104Pro | LP | | ✓ | 1 | ND | ND | Del | PRD | DC | This study | | |
| ## | 94508323 | rs6175 0120 | G | A | c.3322C>T | p.Arg1108Cys | LP | ✓ | ✓ | 14 | 0.0001154 | 0.0006 | Del | PRD | DC | Fujinami K, <i>et al.</i> | 12 | |
| ## | 94508322 | | C | T | c.3323G>A | p.Arg1108His | LP | | ✓ | 2 | 1.649E-05 | ND | Del | PRD | DC | Webster AR, <i>et al.</i> | 21 | |
| ## | 94506958 | | C | A | c.3329G>T | p.Gly1110Val | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | This study | | |
| ## | 94506923 | rs6175 1399 | C | T | c.3364G>A | p.Glu1122Lys | LP | ✓ | ✓ | 5 | 2.471E-05 | ND | Del | PRD | DC | Cideciyan AV, <i>et al.</i> | 31 | |
| ## | 94506902 | | G | A | c.3385C>T | p.Arg1129Cys | LP | ✓ | ✓ | 3 | 1.648E-05 | ND | Del | PRD | DC | Cideciyan AV, <i>et al.</i> | 12 | |
| ## | 94506901 | rs1801 269 | C | A | c.3386G>T | p.Arg1129Leu | LP | | ✓ | 8 | 0.0002471 | 0.0006 | Del | PRD | DC | Riveiro-Alvarez R, <i>et al.</i> | 4 | |
| ## | 94506898 | | A | G | c.3389T>C | p.Ile1130Thr | LP | | ✓ | 1 | ND | ND | Del | PRD | DC | Jiang F, <i>et al.</i> | 32 | |
| ## | 94506895 | | GG | C | c.3392_3393delCCinsG | p.Ala1131GlyfsTer17 | LP | ✓ | | 1 | ND | ND | NA | NA | NA | Fujinami K, <i>et al.</i> | 17 | |
| ## | 94506806 | | G | A | c.3481C>T | p.Arg1161Cys | LP | ✓ | | 1 | 4.119E-05 | 0.0002 | Del | PRD | DC | This study | | |

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|----|----------|----------------------|-----|------------|---------------------|----------------------|---|-----|---|---|---|-----------|--------|-----|-----|-----|------------------------------------|--------------------------------|----|
| ## | 94490550 | rs6264 2574 | C | T | c.4594G>A | p.Asp1532Asn | Missense | LP | ✓ | ✓ | 3 | 9.061E-05 | ND | Del | Ben | DC | Lewis RA, <i>et al.</i> | 5 | |
| ## | 94488976 | | C | T | c.4653G>A | p.Trp1551Ter | Stop | LP | | ✓ | 1 | ND | ND | NA | NA | DC | Duno M, <i>et al.</i> | 37 | |
| ## | 94488946 | | GT | | c.4663_4664delCA | p.Gln1555GlufsTer41 | Frame shift | LP | ✓ | | 1 | ND | ND | NA | NA | NA | This study | | |
| ## | 94488897 | | A | G | c.4667+45T>C | Deep intron | Deep intronic variant with uncertain effect | LLP | ✓ | | 1 | 0.0002085 | ND | NA | NA | NA | NSA | This study | |
| ## | 94487490 | rs1762 111 | A | G | c.4685T>C | p.Ile1562Thr | Missense | LP | ✓ | ✓ | 1 | 0.001305 | ND | Tol | Ben | DC | Fujinami K, <i>et al.</i> | 17 | |
| ## | 94487466 | | G | A | c.4709C>T | p.Pro1570Leu | Missense | LLP | ✓ | | 1 | ND | ND | Tol | PRD | Pol | NSA | This study | |
| ## | 94487455 | | C | A | c.4720G>T | p.Glu1574Ter | Stop | LP | | ✓ | 1 | ND | ND | NA | NA | DC | Maia-Lopes S, <i>et al.</i> | 38 | |
| ## | 94487300 | rs1115 05866 | C | T | c.4774-30G>A | Deep intron | Deep intronic variant with uncertain effect | LLP | ✓ | | 1 | 0.001663 | ND | NA | NA | NA | NSA | This study | |
| ## | 94487436 | | A | G | c.4739T>C | p.Leu1580Ser | Missense | LLP | ✓ | ✓ | 2 | 2.472E-05 | ND | Tol | Ben | Pol | NSA | Passerini I, <i>et al.</i> | 39 |
| ## | 94487404 | rs1131 06943 | C | T | c.4771G>A | p.Gly1591Arg | Missense | LP | | ✓ | 1 | 0.002617 | 0.0004 | Tol | PRD | DC | Müller PL, <i>et al.</i> | 40 | |
| ## | 94487251 | rs6175 0155 | G | T | c.4793C>A | p.Ala1598Asp | Missense | LP | ✓ | | 2 | 2.639E-05 | ND | Tol | PRD | DC | Boulanger-Scemama E, <i>et al.</i> | 55 | |
| ## | 94486896 | rs6175 1404 | G | A | c.4918C>T | p.Arg1640Trp | Missense | LP | ✓ | ✓ | 7 | 8.238E-06 | ND | Tol | PRD | DC | Rozet JM, <i>et al.</i> | 2 | |
| ## | 94486895 | rs6175 1403 | C | T | c.4919G>A | p.Arg1640Gln | Missense | LP | | ✓ | 5 | 5.766E-05 | ND | Del | PRD | DC | Briggs CE, <i>et al.</i> | 1 | |
| ## | 94486888 | rs6175 3017 | G | C | c.4926C>G | p.Ser1642Arg | Missense | LP | ✓ | ✓ | 2 | 3.295E-05 | ND | Del | Ben | DC | Fujinami K, <i>et al.</i> | 17 | |
| ## | 94486794 | | A | G | c.5018+2T>C | Splice donor | Splice site alteration | LP | ✓ | ✓ | 4 | ND | ND | NA | NA | NA | PRA | Fujinami K, <i>et al.</i> | 17 |
| ## | 94486791 | | A | T | c.5018+5G>A | Splice region | Splice site alteration | LP | ✓ | | 1 | ND | ND | NA | NA | NA | PRA | This study | |
| ## | 94486788 | rs1888 00817 | T | C | c.5018+8A>G | Splice region | Potential splice site alteration | LLP | ✓ | | 1 | 0.0009637 | ND | NA | NA | NA | NSA | Zemant J, <i>et al.</i> | 23 |
| ## | 94485279 | A1CC AACG CTAC | C | T | c.5041_5055delGTGGT | p.Val1681_Cys1685del | In-frame deletion | LP | ✓ | ✓ | 2 | ND | ND | NA | NA | NA | | Fujinami K, <i>et al.</i> | 17 |
| ## | 94485278 | rs6175 3019 | C | T | c.5056G>A | p.Val1686Met | Missense | LP | | ✓ | 1 | 0.0005179 | 0.0004 | Del | PRD | DC | Duncker T, <i>et al.</i> | 57 | |
| ## | 94485257 | rs6175 0563 | C | T | c.5077G>A | p.Val1693Ile | Missense | LP | ✓ | ✓ | 2 | 0.0002382 | 0.0008 | Del | PRD | Pol | Tomas R, <i>et al.</i> | 42 | |
| ## | 94485220 | | C | T | c.5114G>A | p.Arg1705Gln | Missense | LP | | ✓ | 1 | 7.758E-05 | ND | Del | PRD | DC | Aleman TS, <i>et al.</i> | 43 | |
| ## | 94485220 | | C | A | c.5114G>T | p.Arg1705Leu | Missense | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | Rivera A, <i>et al.</i> | 20 | |
| ## | 94485197 | | G | T | c.5137C>A | p.Gln1713Lys | Missense | LP | | ✓ | 1 | 4.362E-05 | ND | Del | POD | DC | This study | | |
| ## | 94485172 | | ACC | C | c.5161_5162delAC | p.Thr1721TfsTer65 | Frame shift | LP | ✓ | | 1 | ND | ND | NA | NA | NA | | Briggs CE, <i>et al.</i> | 1 |
| ## | 94485160 | | C | T | c.5172G>A | p.Trp1724Ter | Stop | LP | | ✓ | 1 | ND | ND | NA | NA | DC | Jiang F, <i>et al.</i> | 32 | |
| ## | 94485157 | | G | T | c.5177C>A | p.Thr1726Asn | Missense | LP | ✓ | ✓ | 2 | ND | ND | Tol | PRD | DC | Zemant J, <i>et al.</i> | 58 | |
| ## | 94484001 | | C | T | c.5196+1137G>A | Deep intron | Deep intronic variant with uncertain effect | LLP | ✓ | | 2 | ND | ND | NA | NA | NA | NSA | Braun TA, <i>et al.</i> | 22 |
| ## | 94485137 | rs6175 1377 | C | T | c.5196+1G>A | Splice donor | Splice site alteration | LP | ✓ | ✓ | 4 | 6.017E-05 | ND | NA | NA | NA | PRA | Kitiratschky VB, <i>et al.</i> | 33 |
| ## | 94481390 | CGA CGAC GA | CGA | CGAC GA | c.5216_5218dupGCT | p.Ala1739dup | Duplication | LP | ✓ | | 1 | ND | ND | NA | NA | NA | | This study | |
| ## | 94481339 | | A | G | c.5288T>C | p.Leu1763Pro | Missense | LP | | ✓ | 1 | ND | ND | Del | PRD | DC | Rivera A, <i>et al.</i> | 20 | |
| ## | 94481296 | | C | T | c.5311G>A | p.Gly1771Arg | Missense | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | Weisschuh N, <i>et al.</i> | 44 | |
| ## | 94481295 | | C | T | c.5312+1G>A | Splice donor | Splice site alteration | LP | | ✓ | 1 | ND | ND | NA | NA | NA | | Zemant J, <i>et al.</i> | 58 |

| | | | | | | | | | | | | | | | | | | | | | | |
|----|----------|------------|---|---|--------------------|--------------------|-----------------|---|-----|---|---|---|-----------|-----------|-----------|-----|-----|----------------------------------|---------------------------------|---------------------------|-------------------------|----|
| ## | 94480241 | . | G | A | c.5318C>T | p.Ala1773Val | Missense | LP | ✓ | | | 1 | 0.0001153 | ND | Del | PRD | DC | Chacón-Camacho OF, <i>et al.</i> | 45 | | | |
| ## | 94480222 | | T | A | c.5337C>G | p.Tyr1779Ter | Stop | LP | ✓ | | | 1 | ND | 0.0002 | NA | NA | DC | Papaoannou M, <i>et al.</i> | 54 | | | |
| ## | 94480164 | | T | C | c.5395A>G | p.Asn1799Asp | Missense | LP | | ✓ | | 2 | ND | ND | Del | PRD | DC | Paloma E, <i>et al.</i> | 9 | | | |
| ## | 94480095 | . | T | A | c.5460+4A>G | | Splice region | Splice site alteration | LP | | ✓ | 1 | 8.237E-06 | ND | NA | NA | NA | PRA | This study | | | |
| ## | 94476951 | rs1800728 | A | G | c.5461-10T>C | | Splice region | Splice site alteration | LP | ✓ | ✓ | ✓ | 31 | 0.0002231 | ND | NA | NA | NA | POA | Fujinami K, <i>et al.</i> | 17 | |
| ## | 94476886 | | A | G | c.5516T>C | p.Phe1839Ser | Missense | LP | | ✓ | | 1 | ND | ND | Del | PRD | DC | Fujinami K, <i>et al.</i> | 12 | | | |
| ## | 94476882 | | G | T | c.5520C>A | p.Cys1840Ter | Stop | LP | | ✓ | | 1 | ND | ND | NA | NA | DC | This study | | | | |
| ## | 94476822 | . | G | A | c.5578C>T | p.Arg1860Trp | Missense | LP | ✓ | | | 1 | ND | ND | Del | Ben | DC | Fujinami K, <i>et al.</i> | 17 | | | |
| ## | 94476495 | | A | G | c.5585-10T>C | | Splice region | Splice site alteration | LP | | ✓ | 1 | ND | ND | NA | NA | NA | POA | Rosenberg T, <i>et al.</i> | 26 | | |
| ## | 94476804 | | C | T | c.5584+14G>A | | Deep intron | Deep intronic variant with uncertain effect | LLP | ✓ | | 1 | ND | ND | NA | NA | NA | NSA | This study | | | |
| ## | 94476812 | . | A | G | c.5584+6T>C | | Splice region | Splice site alteration | LP | ✓ | | 1 | 8.271E-06 | ND | NA | NA | NA | POA | Bax NM, <i>et al.</i> | 46 | | |
| ## | 94476467 | rs1801466 | T | A | c.5603A>T | p.Asn1868Ile | Missense | LP | | ✓ | | 5 | 0.04456 | 0.0208 | Tol | PRD | Pol | NSA | Aguirre-Lamban J, <i>et al.</i> | 18 | | |
| ## | 94476377 | rs1800552 | C | T | c.5693G>A | p.Arg1898His | Missense | LLP | | ✓ | ✓ | 2 | 0.001784 | 0.0004 | Tol | Ben | Pol | NSA | Lewis RA, <i>et al.</i> | 5 | | |
| ## | 94476351 | rs61751407 | C | T | c.5714+5G>A | | Splice region | Splice site alteration | LP | | ✓ | ✓ | ✓ | 13 | 0.0003478 | ND | NA | NA | NA | PRA | Rivera A, <i>et al.</i> | 20 |
| ## | 94474428 | . | C | A | c.5715-1G>T | | Splice acceptor | Splice site alteration | LP | ✓ | | 1 | ND | ND | NA | NA | NA | PRA | This study | | | |
| ## | 94474369 | | T | C | c.5773A>G | p.Arg1925Gly | Missense | LP | | ✓ | | 1 | 8.236E-06 | ND | Del | PRD | DC | Huang WC, <i>et al.</i> | 16 | | | |
| ## | 94474365 | | T | G | c.5777A>C | p.Gln1926Pro | Missense | LP | ✓ | | | 1 | ND | ND | Del | PRD | DC | This study | | | | |
| ## | 94485250 | | G | T | c.5084C>A | p.Ala1695Asp | Missense | LP | | ✓ | | 1 | ND | ND | Del | PRD | DC | Lazow MA, <i>et al.</i> | 48 | | | |
| ## | 94474323 | rs61753033 | A | G | c.5819T>C | p.Leu1940Pro | Missense | LP | | ✓ | | 1 | 8.237E-06 | ND | Del | PRD | DC | Paloma E, <i>et al.</i> | 9 | | | |
| ## | 94474302 | | C | T | c.5835+5G>A | | Splice region | Splice site alteration | LP | | ✓ | 1 | ND | ND | NA | NA | NA | PRA | This study | | | |
| ## | 94473790 | . | C | T | c.5898+1G>A | | Splice donor | Splice site alteration | LP | ✓ | | 1 | ND | ND | NA | NA | NA | PRA | Alapati A, <i>et al.</i> | 49 | | |
| ## | 94473807 | rs1800553 | C | T | c.5882G>A | p.Gly1961Glu | Missense | LP | ✓ | ✓ | ✓ | ✓ | 91 | 0.005054 | ND | Del | PRD | DC | Burke TR, <i>et al.</i> | 34 | | |
| ## | 94473287 | rs28938473 | G | A | c.5908C>T | p.Leu1970Phe | Missense | LP | | ✓ | | 2 | 0.002925 | 0.0024 | Tol | PRD | DC | Rozet JM, <i>et al.</i> | 2 | | | |
| ## | 94473266 | rs61750639 | C | T | c.5929G>A | p.Gly1977Ser | Missense | LP | | ✓ | | 2 | 8.304E-06 | ND | Del | PRD | DC | Lewis RA, <i>et al.</i> | 5 | | | |
| ## | 94473259 | | G | A | c.5936C>T | p.Thr1979Ile | Missense | LP | | ✓ | ✓ | 3 | ND | ND | Del | PRD | DC | This study | | | | |
| ## | 94473234 | CCTG T | T | | c.5961_5964delGGAC | p.Gly1987GlyfsTer4 | Frame shift | LP | | ✓ | ✓ | 2 | ND | ND | NA | NA | NA | Passerini I, <i>et al.</i> | 39 | | | |
| ## | 94473189 | . | C | A | c.6005+1G>T | | Splice donor | Splice site alteration | LP | ✓ | | 2 | ND | ND | NA | NA | NA | Lewis RA, <i>et al.</i> | 5 | | | |
| ## | 94471067 | | A | G | c.6077T>C | p.Leu2026Pro | Missense | LP | | ✓ | ✓ | 2 | ND | ND | Tol | PRD | DC | This study | | | | |
| ## | 94471065 | rs61751408 | G | A | c.6079C>T | p.Leu2027Phe | Missense | LP | ✓ | ✓ | ✓ | ✓ | 13 | 0.0002142 | 0.0002 | Del | PRD | DC | Fujinami K, <i>et al.</i> | 6 | | |
| ## | 94471055 | rs61750641 | C | T | c.6089G>A | p.Arg2030Gln | Missense | LP | ✓ | ✓ | ✓ | 9 | 0.0003871 | 0.0006 | Del | PRD | DC | Cideciyan AV, <i>et al.</i> | 50 | | | |
| ## | 94471056 | rs61751383 | G | A | c.6088C>T | p.Arg2030Ter | Stop | LP | ✓ | | | 4 | 2.471E-05 | ND | NA | NA | DC | Fishman GA, <i>et al.</i> | 51 | | | |
| ## | 94471032 | rs61750643 | G | A | c.6112C>T | p.Arg2038Trp | Missense | LP | ✓ | | | 2 | 2.471E-05 | ND | Del | PRD | DC | Lewis RA, <i>et al.</i> | 5 | | | |

| | | | | | | | | | | | | | | | | | | | |
|----|----------|---------------------|----|---|---------------------|---------------------|--|-----|---|---|----|-----------|--------|-----|-----|-----|----------------------------|---------------------------|----|
| ## | 94471026 | rs6175 3038 | G | A | c.6118C>T | p.Arg2040Ter | Stop | LP | ✓ | | 1 | 1.648E-05 | ND | NA | NA | DC | Rosenberg T, <i>et al.</i> | 26 | |
| ## | 94467548 | rs4129 2677 | C | G | c.6148G>C | p.Val2050Leu | Missense | LP | ✓ | ✓ | 4 | 0.002752 | 0.0028 | Del | PRD | DC | Fujinami K, <i>et al.</i> | 10 | |
| ## | 94467512 | TGAC A | A | A | c.6181_6184delACTG | p.Thr2061SerfsTer53 | Frame shift | LP | | ✓ | 2 | ND | ND | NA | NA | NA | This study | | |
| ## | 94467487 | | G | C | c.6209C>G | p.Thr2070Arg | Missense | LP | ✓ | | 2 | ND | ND | Tol | PRD | DC | Fujinami K, <i>et al.</i> | 17 | |
| ## | 94467467 | | G | A | c.6229C>T | p.Arg2077Trp | Missense | LP | ✓ | | 1 | ND | ND | Del | PRD | DC | Rivera A, <i>et al.</i> | 20 | |
| ## | 94467475 | | C | A | c.6221G>T | p.Gly2074Val | Missense | LP | | ✓ | 1 | 0.0000412 | ND | Del | PRD | DC | Alapati A, <i>et al.</i> | 49 | |
| ## | 94467445 | | G | A | c.6250G>A | p.Ala2084Thr | Missense | LP | | ✓ | 1 | ND | ND | Del | PRD | DC | This study | | |
| ## | 94467407 | rs1711 0761 | C | T | c.6282+7G>A | Splice region | Potential splice site alteration | LLP | | ✓ | 1 | 0.08091 | 0.134 | NA | NA | NA | NSA | Tsipi M, <i>et al.</i> | 52 |
| ## | 94466658 | rs6175 0646 | C | T | c.6286G>A | p.Glu2096Lys | Missense | LP | | ✓ | 2 | 8.278E-06 | ND | Del | PRD | DC | Lewis RA, <i>et al.</i> | 5 | |
| ## | 94466624 | | G | A | c.6319C>T | p.Arg2107Cys | Missense | LP | | ✓ | 1 | 0.0000165 | ND | Tol | PRD | DC | Briggs CE, <i>et al.</i> | 1 | |
| ## | 94466624 | rs6264 2564 | C | T | c.6320G>A | p.Arg2107His | Missense | LP | ✓ | ✓ | 12 | 0.001872 | 0.0046 | Del | PRD | DC | Fujinami K, <i>et al.</i> | 12 | |
| ## | 94466561 | | T | C | c.6383A>G | p.His2128Arg | Missense | LP | ✓ | | 1 | 8.238E-06 | ND | Del | PRD | DC | Fishman GA, <i>et al.</i> | 51 | |
| ## | 94466556 | | G | C | c.6386+2C>G | Splice donor | Splice site alteration | LP | ✓ | | 3 | ND | | NA | NA | NA | PRA | Stenirri S, <i>et al.</i> | 20 |
| ## | 94466426 | rs1401 42529 | G | A | c.6445C>T | p.Arg2149Ter | Stop | LP | | ✓ | 1 | 1.647E-05 | ND | NA | NA | DC | Lewis RA, <i>et al.</i> | 5 | |
| ## | 94463499 | | G | A | c.6647C>T | p.Ala2216Val | Missense | LP | ✓ | | 1 | ND | ND | Del | POD | DC | Rivera A, <i>et al.</i> | 20 | |
| ## | 94466422 | rs6175 1384 | C | T | c.6449G>A | p.Cys2150Tyr | Missense | LP | ✓ | | 1 | 2.471E-05 | ND | Del | PRD | DC | Yatsenko AN, <i>et al.</i> | 15 | |
| ## | 94463624 | rs1439 09323 | G | A | c.6522G>A | p.Pro2174Pro | Synonymous variant with potential splice site alteration | LLP | ✓ | | 1 | ND | 0.0004 | NA | NA | DC | NSA | This study | |
| ## | 94463617 | rs1800 555 | C | T | c.6529G>A | p.Asp2177Asn | Missense | LLP | ✓ | | 1 | 0.01018 | 0.004 | Tol | Ben | Pol | NSA | Rivera A, <i>et al.</i> | 20 |
| ## | 94463453 | | GC | C | c.6693delC | p.Ile2231MetfsTer16 | Frame shift | LP | | ✓ | 1 | ND | ND | NA | NA | DC | This study | | |
| ## | 94463432 | | G | A | c.6712C>T | p.Gln2238Ter | Stop | LP | ✓ | | 1 | ND | ND | NA | NA | DC | This study | | |
| ## | 94463425 | rs6174 8521 | G | C | c.6721C>G | p.Leu2241Val | Missense | LP | ✓ | | 2 | 0.0001652 | 0.0002 | Del | PRD | DC | Rivera A, <i>et al.</i> | 20 | |
| ## | 94463412 | CAAC CGG CAAC | C | C | c.6729+5_19delGTTGG | Splice region | Splice site alteration | LP | ✓ | | 5 | ND | ND | NA | NA | NA | NA | Littink KW, <i>et al.</i> | 53 |
| ## | 94461754 | rs1800 717 | A | G | c.6730-3T>C | Splice region | Potential splice site alteration | LLP | | ✓ | 2 | 0.07276 | ND | NA | NA | NA | NSA | Stenirri S, <i>et al.</i> | 20 |
| ## | 94461717 | rs6666 652 | C | A | c.6764G>T | p.Ser2255Ile | Missense | LLP | | ✓ | 3 | 0.08023 | ND | Del | Ben | Pol | NSA | Souied EH, <i>et al.</i> | 27 |
| ## | 94458800 | | T | G | c.6817-2A>C | Splice acceptor | Splice site alteration | LP | ✓ | | 2 | ND | ND | NA | NA | NA | PRA | Fujinami K, <i>et al.</i> | 5 |

Vt no. = variant number; Ref = reference; Alt = alternate; Position = genome position; LP = likely pathogenic; LLP = less likely pathogenic; NGS = PCR enrichment based targeted next generation sequencing; GCA = gene chip array; SSCP = single-strand conformation polymorphism; DS = direct sequencing; ND = not detected; ExAC Browser = The Exome Aggregation Consortium Browser; SIFT = Sorting Intolerant from Tolerant; HSF = Human Splicing finder program version 3.0; Del = deleterious; Tol = tolerant; PRD = probably damaging; POD = possibly damaging; Ben = benign; DC = disease causing; Pol = polymorphism; PRA = probably affecting splicing; POA = possibly affecting splicing; NSA = not significantly affecting splicing.

Reference genome and transcript: UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly and ENST00000370225 (50 coding exons).

Novel variants are shown in *Italic*.

Supplemental references are listed below.

1. Briggs CE, Rucinski D, Rosenfeld PJ, Hirose T, Berson EL, Dryja TP. Mutations in ABCR (ABCA4) in patients with Stargardt macular degeneration or cone-rod degeneration. *Invest Ophthalmol Vis Sci* 2001;42:(10):2229-36