**Appendix. OPN1LW, OPN1MW, OPN1SW and Rho SNPs Identified by LR-PCR-Seq**

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| **GENE; Location;****Genomic Reference** | **rs Number** | **Functional Consequence/****Clinical Significance** | **Incidence****(Subject number)** | **Amino Acid Change** | **Minor allele frequency****(Sequencing Project)** |
| **OPN1LW; Chromosome X;** **Reference NG\_009105.2** |  |  |  | **Protein Reference NP\_064445.2** |  |
| X:154152987g.[13737A>C](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NG_009105.2&search=NG_009105.2:g.13737A%3eC&v=1:100&content=5) | rs713 | Missense | 1,2,3,4,5,6,7 | p.[Met153Leu](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Met153Leu&v=1:100&content=5)  | A=0.0013 (ExAC) |
| X:154153051g.13801C>T | rs149897670 | Missense | 0 | p.[Ala174Val](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Ala174Val&v=1:100&content=5) | T=0.1011 (ExAC)T=0.1348 (1000 Genomes)T=0.1235 (GO-ESP) |
| X:154153062g.[13812A>G](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NG_009105.2&search=NG_009105.2:g.13812A%3eG&v=1:100&content=5) | rs145009674 | Missense/Benign | 0 | p.[Ile178Val](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Ile178Val&v=1:100&content=5) | G=0.0221 (ExAC)G=0.0366 (1000 Genomes)G=0.0214 (GO-ESP) |
| X:154153068g.[13818G>T](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NG_009105.2&search=NG_009105.2:g.13818G%3eT&v=1:100&content=5) | rs949431 | Missense/Benign | 5,7,8 had A1,3,4,6 had A/S; 2 had S | p.[Ala180Ser](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Ala180Ser&v=1:100&content=5) | G=0.2053 (ExAC)G=0.1674 (1000 Genomes) |
| X:154154684g.[15434T>C](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NG_009105.2&search=NG_009105.2:g.15434T%3eC&v=1:100&content=5) | rs148583295 | Missense | 2I/T1,4 | p.[Ile230Thr](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Ile230Thr&v=1:100&content=5)  | C=0.0032 (ExAC)C=0.0008 (1000 Genomes)C=0.0021 (GO-ESP)C=0.0024 (TOPMED) |
| X:154154692-94g.15442G>Ag.15443C>Tg.15444T>C | Triplet of variants causing amino acid change | Missense/Benign | 1,4 | [p.Ala233Ser](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Ala233Thr&v=1:100&content=5) | A=0.000072 (GnomAD) |
| X:154156369g.17119A>G | None | Unknown | 8 | p.Ile274Val | N/A |
| X:154156379g.17129A>T | rs2315126  | Missense | 8,9 | [p.Tyr277Phe](http://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Tyr277Phe&v=1:100&content=5)  | N/A |
| X:154156384g.17134G>A | rs2315127 | Missense | 5,8,9 | p.Val279Phe | A=0.00001 (ExAC)T=0.0002 (GO-ESP)T=0.00004 (TOPMED) |
| X:154156402g.17152A>G | rs200528091 | Missense | 8,9 | p.[Thr285Ala](http://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Thr285Ala&v=1:100&content=5) | G=0.0006 (ExAC)G=0.0005 (1000 Genomes)G=0.0006 (GO-ESP)G=0.0005 (TOPMED) |
| X:154156441g.17191G>A | rs1065440 | Missense | 8,9 | [p.Ala298Pro](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Ala298Pro&v=1:100&content=5) | C=0.0003 (ExAC)C=0.0003 (TOPMED) |
| X:154156475g.17225A>T | rs145631912  | Missense | 7,8,9 | [p.Tyr309Phe](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_064445.2&search=NP_064445.2:p.Tyr309Phe&v=1:100&content=5) | T=0.0004 (ExAC)T=0.0008 (1000 Genomes)T=0.0005 (TOPMED) |
| **GENE; Location;****Genomic Reference** | **rs Number** | **Functional Consequence/****Clinical Significance** | **Incidence****(Subject number)** | **Amino Acid Change** | **Minor allele frequency****(Sequencing Project)** |
| **OPN1MW; Chromosome X;** **Reference NG\_01166.1** |  |  |  | **Protein Reference NP\_000504.1** |  |
| X:154190165g.12570C>T | rs372044027 | Missense | 1 | [p.Ala174Val](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_000504.1&search=NP_000504.1:p.Ala174Val&v=1:100&content=5) | T=0.0488 (ExAC) |
| X:154190176g.12581A>G | rs375538821 | Missense | 1,3 | [p.Ile178Val](http://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_000504.1&search=NP_000504.1:p.Ile178Val&v=1:100&content=5) | G= 0.02853 (GnomAD) |
| X:154190182g.12587G>T | rs949431  | Missense | 9 | [p.Ala180Ser](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_000504.1&search=NP_000504.1:p.Ala180Ser&v=1:100&content=5) | T=0.074(GnomAD) |
| X:154191800g.14205C>T | None | Unknown | 0 | p.Thr230Ile | N/A |
| X:154191808-10g.14213A>Gg.14214G>Cg.14215C>T | None | Unknown | 0 | p.Ser233Ala | N/A |
| X:15419817g.14222G>A | None | Unknown | 0 | p.Val236Met | N/A |
| X:154193483g.15890G>A | None | Unknown | 8 | p.Val274Ile | N/A |
| X:154193493g.15900A>G | None | Unknown | 8 | p.Phe277Tyr | N/A |
| X:154193498g.15905T>G | rs782251342 | Missense | 8 | p.Phe279Val | G=0.0010 (ExAC) |
| X:154193516g.15923G>A | rs782051833 | Missense | 8 | [p.Ala285Thr](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_000504.1&search=NP_000504.1:p.Ala285Thr&v=1:100&content=5)  | A=0.00001 (ExAC) |
| X:154193555g.15962C>G | rs201292561 | Missense | 8 | [p.Pro298Ala](http://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_000504.1&search=NP_000504.1:p.Pro298Ala&v=1:100&content=5) | G=0.0002 (ExAC)G=0.0021 (1000 Genomes)G=0.0017 (TOPMED) |
| X:154193589g.15996T>A | rs782593302 | Missense | 8 | [p.Phe309Tyr](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NP_000504.1&search=NP_000504.1:p.Phe309Tyr&v=1:100&content=5)  | A=0.0002 (ExAC)A=0.0053 (TOPMED) |
| **GENE;Location;****Genomic Reference** | **rs Number** | **Functional Consequence/****Clinical Significance** | **Incidence****(Subject number)** | **Amino Acid Change** | **Minor allele frequency****(Sequencing Project)** |
| **OPN1SW; Chromosome 7;** **Reference NG\_009094** |  |  |  | **Protein Reference NP\_001699.1** |  |
| 7:128775236g. 5555G>A | rs17476081 | Intron variant | 5,6,8 | None | T=0.1791 (1000 Genomes)T=0.2762 (TOPMED) |
| 7:128775141g. 5650A>C | rs1799922 | Synonymous codon | 3,5,6,8 | p.Gly122 | G=0.3026 (ExAC)G=0.1871 (1000 Genomes)G=0.3169 (GO-ESP)G=0.2910 (TOPMED) |
| 7:128774430g. 6361A>G | rs17397588 | Intron variant | 5,6,8 | None | C=0.1789 (1000 Genomes)C=0.2764 (TOPMED) |
| 7:128774281g. 6510C>T | rs73238109 | Intron variant | 3,4 | None | A=0.1090 (1000 Genomes)A=0.0701 (TOPMED) |
| 7:128774141g. 6650C>T | rs73238106 | Intron variant | 3,4 | None | A=0.0847 (1000 Genomes)A=0.0466 (TOPMED) |
| 7:128774034g. 6757C>T | rs45486505 | Intron variant | 5,6,8 | None | A=0.1787 (1000 Genomes)A=0.2761 (TOPMED) |
| **GENE;Location;****Genomic Reference** | **rs Number** | **Functional Consequence/****Clinical Significance** | **Incidence****(Subject number)** | **Amino Acid Change** | **Minor allele frequency****(Sequencing Project)** |
| **RHODOPSIN; Chromosome 3;** **Reference NG\_009115.1** |  |  |  | **Protein Reference NP\_000530.1** |  |
| 3:129528683g. 5045G>A | rs2269736 | utr variant 5 prime/ Benign | 3,9 | None | A=0.0641 (ExAC)A=0.1697 (1000 Genomes)A=0.0651 (GO-ESP)A=0.0990 (TOPMED) |
| 3:129528708g. 5070A>G | rs7984 | utr variant 5 prime/ Benign | 1,2,3,9 | None | G=0.2869 (ExAC)A=0.4716 (1000 Genomes)G=0.3793 (GO-ESP)G=0.4116 (TOPMED) |
| 3:129528885g. 5247G>C | rs149079952 | Missense/likely Benign | 0 | p.Gly51Ala | C=0.0011 (ExAC)C=0.0046 (1000 Genomes)C=0.0025 (GO-ESP)C=0.0025 (TOPMED) |
| 3:129529093g.5455C>T | rs79765751 | Synonymous codon/ likely Benign | 0 | p.Gly120 | T=0.0032 (ExAC)T=0.0036 (GO-ESP)T=0.0041 (TOPMED) |
| 3:129531031g.7393G>A | rs139731264 | Missense | 0 | p.Ala173Thr | A=0.0002 (ExAC)A=0.0004 (GO-ESP)A=0.0003 (TOPMED) |
| 3:129532420g.8782C>T | rs56340615 | Intron variant/Benign | 0 | None | T=0.0824 (ExAC)T=0.0857 (1000 Genomes)T=0.0820 (GO-ESP)T=0.0797 (TOPMED) |
| 3:129533585g.9947G>A | rs2071092 | Intron variant | 1,3 | None | A=0.0538 (ExAC)A=0.1150 (1000 Genomes)A=0.0484 (GO-ESP)A=0.0635 (TOPMED) |
| 3:129533640g.10002C>T | rs142771862 | Synonymous codon/ likely Benign | 0 | p.Cys323 | T=0.0006 (ExAC)T=0.0013 (GO-ESP)T=0.0007 (TOPMED) |
| 3:12953375 g.10113C>T | rs113310993 | utr variant 3 prime/ likely Benign | 0 | None | T=0.0038 (ExAC)T=0.0086 (1000 Genomes)T=0.0124 (GO-ESP)T=0.0113 (TOPMED) |
| 3:129533761g.10123C>A | rs2071093 | utr variant 3 prime/ Benign | 2 | None | A=0.0828 (ExAC)A=0.0841 (1000 Genomes)A=0.0807 (GO-ESP)A=0.0794 (TOPMED) |

Minor allele frequency (MAF):  MAF is the frequency of the minor allele. MAF is often reported in the context of allele frequencies established by the 1000 Genomes and other large sequencing projects. When there are more than two alleles, MAF refers to the **second** most frequent allele.

N/A: No frequency data noted in NCBI dbSNP data base