

Table S4. All of 123 variations validated by dideoxy sequencing.

Gene	Refseq ID	Nucleotide change	Amino acid change	Genotype	Category	SNP ID	Reported Mut.	PolyPhen-2	SIFT
ABCA4	NM_000350.2	c.4793C>A	p.Ala1598Asp	Hetero	Coding nonsynonymous	rs61750155	CM003386	Poss/0.638	Damaging/0.01
ABCA4	NM_000350.2	c.6320G>A	p.Arg2107His	Hetero	Coding nonsynonymous	rs62642564	CM990074	Prob/0.996	Damaging/0.00
ABCA4	NM_000350.2	c.3595C>T	p.Gln1199Ter	Hetero	STOP				
ABCA4	NM_000350.2	c.5882G>A	p.Gly1961Glu	Hetero	Coding nonsynonymous	rs1800553	CM970016	Prob/1.000	Damaging/0.00
ABCA4	NM_000350.2	c.4297G>A	p.Val1433Ile	Hetero	Coding nonsynonymous	rs56357060	CM990050	Benign/0.112	Tolerated/0.09
ABCA4	NM_000350.2	c.6730-3T>C		Hetero	Intron	rs1800717			
AIPL1	NM_014336.3	c.937G>T	p.Ala313Ser	Hetero	Coding nonsynonymous			Benign/0.065	Damaging/0.05
AIPL1	NM_014336.3	c.111C>T	p.Phe37Phe	Hetero	Coding synonymous	rs11650007			
AIPL1	NM_014336.3	c.111C>T	p.Phe37Phe	Hetero	Coding synonymous	rs11650007			
BBS1	NM_024649	c.316C>G	p.Leu106Val	Hetero	Coding nonsynonymous			Benign/0.000	Tolerated/1.00
BBS12	NM_001178007.1	c.1381A>C	p.Asn461His	Hetero	Coding nonsynonymous	rs10027479		Poss/0.706	Damaging/0.01
BBS12	NM_001178007.1	c.1847G>A	p.Ser616Asn	Hetero	Coding nonsynonymous	rs28507107		Benign/0.113	Tolerated/0.58
BBS2	NM_031885.3	c.1511C>T	p.Ala504Val	Homo	Coding nonsynonymous	rs16957538		Benign/0.000	Tolerated/0.29
BBS2	NM_031885.3	c.1657+3A>G		Hetero	Intron	rs6499838			
BBS5	NM_152384.2	c.551A>G	p.Asn184Ser	Hetero	Coding nonsynonymous		CM044580	Prob/0.986	Damaging/0.00
BEST1	NM_004183.3	c.727G>A	p.Ala243Thr	Hetero	Coding nonsynonymous		CM004434	Prob/0.946	Damaging/0.01
BEST1	NM_001139443.1	c.1748C>T	p.Ser583Leu	Hetero	Coding nonsynonymous			Benign/0.016	Tolerated/0.21
CA4	NM_000717.3	c.716G>A	p.Arg239Gln	Hetero	Coding nonsynonymous			Benign/0.000	Tolerated/0.59
CA4	NM_000717.3	c.700G>A	p.Val234Ile	Hetero	Coding nonsynonymous			Benign/0.097	Tolerated/0.62
CACNA1F	NM_005183.2	c.5789G>A	p.Arg1930His	Hetero	Coding nonsynonymous	rs33910054		Benign/0.000	Tolerated/0.43
CACNA1F	NM_005183.2	c.5789G>A	p.Arg1930His	Hetero	Coding nonsynonymous	rs33910054		Benign/0.000	Tolerated/0.43
CACNA1F	NM_005183.2	c.4161G>T	p.Glu1387Asp	Hemi	Coding nonsynonymous			Benign/0.266	Damaging/0.00
CACNA2D4	NM_172364.4	c.2517G>A	p.Ala839Ala	Hetero	Coding synonymous	rs33972365			
CACNA2D4	NM_172364.4	c.2517G>A	p.Ala839Ala	Hetero	Coding synonymous	rs33972365			
CACNA2D4	NM_172364.4	c.2065A>G	p.Ile689Val	Hetero	Coding nonsynonymous			Benign/0.001	Tolerated/0.1
CDH23	NM_022124.4	c.4310G>A	p.Arg1437Gln	Hetero	Coding nonsynonymous	rs56181447		Poss/0.711	Tolerate/0.37
CDH23	NM_022124.4	c.7C>T	p.Arg3Cys	Hetero	Coding nonsynonymous	rs7902757		Benign/0.106	Tolerate/0.12
CDH23	NM_022124.4	c.8411A>G	p.Lys2804Arg	Hetero	Coding nonsynonymous			Poss/0.730	Tolerate/0.28
CDH23	NM_022124.4	c.9373T>C	p.Phe3125Leu	Hetero	Coding nonsynonymous	rs45583140		Benign/0.023	Tolerate/0.07

CDH23	NM_022124.4	c.9373T>C	p.Phe3125Leu	Hetero	Coding nonsynonymous	rs45583140		Benign/0.023	Tolerate/0.07
CDH23	NM_022124.4	c.3625A>G	p.Thr1209Ala	Hetero	Coding nonsynonymous	rs41281314	CM021540	Poss/0.742	Tolerate/0.46
CDH23	NM_022124.4	c.5023G>A	p.Val1675Ile	Hetero	Coding nonsynonymous	rs17712523		Benign/0.491	Tolerate/0.39
CDH23	NM_022124.4	c.5023G>A	p.Val1675Ile	Hetero	Coding nonsynonymous	rs17712523		Benign/0.491	Tolerate/0.39
CDH23	NM_022124.4	c.5023G>A	p.Val1675Ile	Hetero	Coding nonsynonymous	rs17712523		Benign/0.491	Tolerate/0.39
CDH23	NM_022124.4	c.5023G>A	p.Val1675Ile	Hetero	Coding nonsynonymous	rs17712523		Benign/0.491	Tolerate/0.39
CDH23	NM_022124.4	c.6847G>A	p.Val2283Ile	Hetero	Coding nonsynonymous	rs41281334		Benign/0.001	Tolerate/0.45
CDH3	NM_001793.4	c.1285G>A	p.Val429Ile	Hetero	Coding nonsynonymous	rs34394404		Benign/0.163	Tolerated/0.43
CEP290	NM_025114.3	c.1991A>G	p.Asp664Gly	Hetero	Coding nonsynonymous			Poss/0.754	Tolerated/0.14
CEP290	NM_025114.3	c.2827A>G	p.Ile943Val	Homo	Coding nonsynonymous			Benign/0.000	Tolerated/0.27
CEP290	NM_025114.3	c.2512A>G	p.Lys838Glu	Homo	Coding nonsynonymous	rs11104738		Benign/0.000	Tolerated/1.00
CNGB1	NM_001297.4	c.2882C>T	p.Ala961Val	Hetero	Coding nonsynonymous			Benign/0.211	Tolerated/0.22
CNGB1	NM_001297.4	c.2747G>A	p.Arg916His	Hetero	Coding nonsynonymous			Prob/0.994	Damaging/0.00
CNGB1	NM_001297.4	c.2233C>A	p.Leu745Ile	Hetero	Coding nonsynonymous			Benign/0.165	Tolerated/0.33
CNGB1	NM_001297.4	c.1473G>A	p.Pro491Pro	Hetero	Coding synonymous				
CNGB1	NM_001297.4	c.1604T>C	p.Val535Ala	Hetero	Coding nonsynonymous			Benign/0.467	Tolerated/0.15
CNGB1	NM_001297.4	c.1604T>C	p.Val535Ala	Hetero	Coding nonsynonymous			Benign/0.467	Tolerated/0.15
CNGB3	NM_019098.4	c.384C>T	p.Ala128Ala	Hetero	Coding synonymous				
CNGB3	NM_019098.4	c.80A>G	p.Asn27Ser	Hetero	Coding nonsynonymous	rs35807406		Benign/0.025	Tolerated/0.41
CRB1	NM_201253.2	c.99G>T	p.Arg33Ser	Hetero	Coding nonsynonymous			Benign/0.090	Tolerated/0.56
CYP4V2	NM_207352.3	c.1114G>A	p.Val372Ile	Hetero	Coding nonsynonymous	rs61755911		Benign/0.004	Tolerated/0.24
FSCN2	NM_001077182.2	c.49G>A	p.Val17Ile	Hetero	Coding nonsynonymous			Benign/0.001	Tolerated/1.00
GPR98	NM_032119.3	c.18273A>G	p.Ala6091Ala	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.10872A>G	p.Gln3624Gln	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.10872A>G	p.Gln3624Gln	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.10577T>C	p.Met3526Thr	Hetero	Coding nonsynonymous			Benign/0.001	Tolerated/0.59
GPR98	NM_032119.3	c.10577T>C	p.Met3526Thr	Homo	Coding nonsynonymous			Benign/0.001	Tolerated/0.59
GPR98	NM_032119.3	c.10577T>C	p.Met3526Thr	Hetero	Coding nonsynonymous			Benign/0.001	Tolerated/0.59
GPR98	NM_032119.3	c.4506C>T	p.Pro1502Pro	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.4506C>T	p.Pro1502Pro	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.4506C>T	p.Pro1502Pro	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.5780C>T	p.Thr1927Met	Hetero	Coding nonsynonymous			Benign/0.024	Tolerated/0.08

GPR98	NM_032119.3	c.5780C>T	p.Thr1927Met	Hetero	Coding nonsynonymous			Benign/0.024	Tolerated/0.08
GPR98	NM_032119.3	c.16308C>A	p.Thr5436Thr	Hetero	Coding synonymous				
GPR98	NM_032119.3	c.5525-7C>T		Hetero	Intron				
GRK1	NM_002929.2	c.693C>T	p.Gly231Gly	Hetero	Coding synonymous	rs62621170			
GRM6	NM_000843.3	c.2420C>T	p.Ala807Val	Hetero	Coding nonsynonymous	rs17078874		Poss/0.505	Tolerated/0.19
GUCA1B	NM_002098.5	c.253G>A	p.Val85Met	Hetero	Coding nonsynonymous		CM045052	Benign/0.023	Damaging/0.02
GUCY2D	NM_000180.3	c.1720C>T	p.Arg574Cys	Hetero	Coding nonsynonymous			Prob/0.933	Damaging/0.02
HMCN1	NM_031935.2	c.14158G>A	p.Ala4720Thr	Hetero	Coding nonsynonymous	rs6693069		Benign/0.002	Tolerated/0.69
HMCN1	NM_031935.2	c.6917G>A	p.Arg2306Gln	Hetero	Coding nonsynonymous			Benign/0.021	Tolerated/0.54
HMCN1	NM_031935.2	c.4586A>G	p.Asn1529Ser	Hetero	Coding nonsynonymous	rs41317471		Poss/0.586	Tolerated/0.16
HMCN1	NM_031935.2	c.5482A>G	p.Ile1828Val	Hetero	Coding nonsynonymous			Prob/0.896	Tolerated/0.4
HMCN1	NM_031935.2	c.9397A>G	p.Ile3133Val	Hetero	Coding nonsynonymous			Benign/0.118	Tolerated/0.29
HMCN1	NM_031935.2	c.13313-4G>A		Hetero	Intron	rs41317497			
HTRA1	NM_002775.4	c.753C>T	p.Ile251Ile	Hetero	Coding synonymous				
KCNV2	NM_133497.2	c.-2C>T		Hetero	5' untranslated region	rs75316505			
LRAT	NM_004744.3	c.74T>A	p.Phe25Tyr	Hetero	Coding nonsynonymous	rs75368761		Prob/0.98	Tolerated/0.11
LRP5	NM_002335.2	c.1999G>A	p.Val667Met	Hetero	Coding nonsynonymous	rs4988321	CM014727	Prob/0.957	Tolerated/0.07
LRP5	NM_002335.2	c.1999G>A	p.Val667Met	Hetero	Coding nonsynonymous	rs4988321	CM014727	Prob/0.957	Tolerated/0.07
LRP5	NM_002335.2	c.1999G>A	p.Val667Met	Hetero	Coding nonsynonymous	rs4988321	CM014727	Prob/0.957	Tolerated/0.07
MERTK	NM_006343.2	c.60A>T	p.Arg20Ser	Hetero	Coding nonsynonymous	rs35898499		Benign/0.018	Tolerated/0.15
MERTK	NM_006343.2	c.878G>A	p.Arg293His	Hetero	Coding nonsynonymous	rs34072093		Benign/0.002	Tolerated/0.62
MERTK	NM_006343.2	c.878G>A	p.Arg293His	Hetero	Coding nonsynonymous	rs34072093		Benign/0.002	Tolerated/0.62
MERTK	NM_006343.2	c.2608G>A	p.Val870Ile	Hetero	Coding nonsynonymous	rs2230517		Benign/0.008	Tolerated/0.74
MKKS	NM_018848.2	c.1015A>G	p.Ile339Val	Hetero	Coding nonsynonymous		CM021299	Benign/0.004	Tolerated/0.13
MKKS	NM_018848.2	c.1015A>G	p.Ile339Val	Hetero	Coding nonsynonymous		CM021299	Benign/0.004	Tolerated/0.13
PCDH15	NM_001142763.1	c.1153G>A	p.Gly385Ser	Hetero	Coding nonsynonymous	rs10825269		Benign/0.002	Tolerated/1.00
PCDH15	NM_001142763.1	c.1153G>A	p.Gly385Ser	Homo	Coding nonsynonymous	rs10825269		Benign/0.002	Tolerated/1.00
PCDH15	NM_001142763.1	c.1278T>C	p.Thr426Thr	Hetero	Coding synonymous	rs7921598			
PCDH15	NM_001142763.1	c.1278T>C	p.Thr426Thr	Hetero	Coding synonymous	rs7921598			
PCDH15	NM_001142763.1	c.1278T>C	p.Thr426Thr	Homo	Coding synonymous	rs7921598			
PDE6A	NM_000440.2	c.1546C>G	p.Leu516Val	Hetero	Coding nonsynonymous	rs6173362		Benign/0.016	Tolerated/0.52
PDE6A	NM_000440.2	c.879G>A	p.Pro293Pro	Hetero	Coding synonymous				

PDE6B	NM_001145291.1	c.143G>A	p.Arg48Gln	Hetero	Coding nonsynonymous		Benign/0.004	Tolerated/0.51
PDE6B	NM_001145291.1	c.143G>A	p.Arg48Gln	Hetero	Coding nonsynonymous		Benign/0.004	Tolerated/0.51
PROM1	NM_006017.2	c.1557C>A	p.Tyr519Ter	Hetero	STOP			
PROM1	NM_006017.2	c.1768-5C>T		Hetero	Intron	rs55708318		
PRPH2	NM_003926.5	c.933C>T	p.Ser311Ser	Hetero	Coding synonymous			
RGR	NM_002921.3	c.734C>T	p.Ser245Phe	Hetero	Coding nonsynonymous	rs1730895	Benign/0.001	Tolerated/0.33
RP1	NM_006269.1	c.6055G>A	p.Gly2019Ser	Hetero	Coding nonsynonymous		Benign/0.001	Tolerated/0.45
RP1	NM_006269.1	c.2255C>T	p.Thr752Met	Hetero	Coding nonsynonymous		Benign/0.025	Tolerated/0.49
RPGR	NM_001034853.1	c.1274G>A	p.Arg425Lys	Hetero	Coding nonsynonymous	rs1801687	Benign/0.083	Tolerated/0.31
RPGR	NM_001034853.1	c.223A>G	p.Ile75Val	Hetero	Coding nonsynonymous	CM011007	Benign/0.015	Tolerated/1.00
RPGRIP1	NM_020366.3	c.3358A>G	p.Ile1120Val	Hetero	Coding nonsynonymous	CM076486	Poss/0.833	Tolerated/0.09
TLR4	NM_138554.3	c.842G>A	p.Cys281Tyr	Hetero	Coding nonsynonymous	CM035921	Poss/0.900	Tolerated/0.25
TLR4	NM_138554.3	c.1196C>T	p.Thr399Ile	Hetero	Coding nonsynonymous	rs4986791 CM001362	Benign/0.013	Tolerated/0.19
TRIM32	NM_012210.3	c.1222C>T	p.Arg408Cys	Hetero	Coding nonsynonymous	rs3747835	Poss/0.795	Damaging/0.04
TTC8	NM_198310.2	c.505-5C>T		Hetero	Intron			
USH2A	NM_206933.2	c.6875G>A	p.Arg2292His	Hetero	Coding nonsynonymous		Benign/0.001	Tolerated/0.58
USH2A	NM_206933.2	c.15091C>T	p.Arg5031Trp	Hetero	Coding nonsynonymous		Benign/0.058	Tolerated/0.19
USH2A	NM_206933.2	c.1931A>T	p.Asp644Val	Hetero	Coding nonsynonymous	rs1805048	Benign/0.077	Tolerated/0.44
USH2A	NM_206933.2	c.6713A>C	p.Glu2238Ala	Hetero	Coding nonsynonymous	UMD-USH2A	Prob/0.995	Damaging/0.02
USH2A	NM_206933.2	c.6713A>C	p.Glu2238Ala	Hetero	Coding nonsynonymous	UMD-USH2A	Prob/0.995	Damaging/0.02
USH2A	NM_206933.2	c.6713A>C	p.Glu2238Ala	Hetero	Coding nonsynonymous	UMD-USH2A	Prob/0.995	Damaging/0.02
USH2A	NM_206933.2	c.2137G>C	p.Gly713Arg	Hetero	Coding nonsynonymous	rs696723 CM001809	Prob/1.000	Damaging/0.01
USH2A	NM_206933.2	c.2137G>C	p.Gly713Arg	Hetero	Coding nonsynonymous	rs696723 CM001809	Prob/1.000	Damaging/0.01
USH2A	NM_206933.2	c.4714C>T	p.Leu1572Phe	Hetero	Coding nonsynonymous		Poss/0.673	Damaging/0.01
USH2A	NM_206933.2	c.7584C>T	p.Thr2528Thr	Hetero	Coding synonymous			
USH2A	NM_206933.2	c.9343A>G	p.Thr3115Ala	Hetero	Coding nonsynonymous		Benign/0.002	Tolerated/1.00
USH2A	NM_206933.2	c.13297G>T	p.Val4433Leu	Hetero	Coding nonsynonymous	UMD-USH2A	Benign/0.000	Tolerated/0.36
USH2A	NM_206933.2	c.13297G>T	p.Val4433Leu	Hetero	Coding nonsynonymous	UMD-USH2A	Benign/0.000	Tolerated/0.36
USH2A	NM_206933.2	c.14792-2A>G		Hetero	Intron			
USH3A	NM_017926.2	c.6A>C	p.Pro2Pro	Hetero	Coding synonymous			

Note: Under the reported mutations, either the HGMD accession number or UMD-USH2A mutation database was cited. PolyPhen-2 or SIFT scores in bold represent the probable/possible damaging effect (PolyPhen-2) or damaging effect (SIFT). Hetero, heterozygous; homo, homozygous; hemi, hemizygous.