

Molecular genetic analysis using targeted NGS analysis of 677 individuals with retinal dystrophy

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Table S1

Diagnosis	Number of individuals
1. Generalized retinal dystrophy (non-syndromic)	486
Retinitis pigmentosa	
Leber congenital amaurosis	
Cone-rod dystrophy	
Choroideremia	
Generalized choriocapillaris dystrophy (GCCD)	
Gyrate atrophy	
Unspecified generalized retinal and chorioretinal dystrophy	
2. Isolated cone disease	24
Cone dystrophy	
Achromatopsia	
Oligocone trichromacy	
3. Macular dystrophy	118
Best vitelliform macular dystrophy	
Adult –onset vitelliform macular dystrophy (including Pattern dystrophy)	
Stargardt disease (Stargardt macular dystrophy, fundus flavimaculatus)	
North Carolina macular dystrophy	
Macular dystrophy unspecified	
4. Vitreoretinopathies	5
Erosive vitreoretinopathies	
Enhanced S cone syndrome (ESCS) (incl Goldmann-Favre syndrome)	
Snowflake vitreoretinal degeneration	
Autosomal dominant vitreoretinopathies (ADVIRC)	
Wagner syndrome and erosive vitreoretinopathy	
Familial exudative vitreoretinopathies (FEVRs)incl Norrie disease	
X-linked juvenile retinoschisis	
Vitreoretinopathy unspecified	
5. Other non-syndromic retinal dystrophies	16
Congenital stationary night blindness	
Bietti crystalline retinopathy	
Oculocutaneous albinism	
Åland eye disease	
Sorsby pseudoinflammatory fundus dystrophy	
Fundus albipunctatus	
6. Syndromic retinal dystrophy	27
Usher syndrome .	
Bardet Biedl syndrome	
Senior Løken Syndrome/Nephronophthisis (SLSN)	
Refsum disease	

Joubert syndrome	
Alagille syndrome (ALGS)	
Alstrom syndrome (ALMS)	
Neuronal ceroid lipofuscinosis (NCLs or CLNs)	
Primary ciliary dyskinesia (PCD)	
Unspezifized syndrome	
7. Retinal vascular dystrophies	1

Table S2

Gene	OMIM gene	Inheritance	Phenotype	Number of mutations in HGMD (2018-2)	Function	NM-number	Gene ID
<i>ABCA4</i>	601691	AR	Stargardt disease; retinitis pigmentosa 19	1030	Visual cycle	NM_000350.2	NG_009073.1
<i>ADAM9</i>	602713	AR	Cone-rod dystrophy	8	Cell adhesion/structure	NM_003816.2	NG_016335.1
<i>ADGRA3</i>	612303	AR	Retinal dystrophy	4	Signal transduction (G-coupled 7 TM receptor)	NM_145290.3	NG_032963.1
<i>ADGRV1</i>	602851	AR	Usher syndrome 2C	184	Signal transduction (G-coupled 7 TM receptor)	NM_032119.3	NG_007083.2
<i>AIP1</i>	604392	AR	Leber congenital amaurosis 4	70	Nuclear transport, protein trafficking, chaperone activity	NM_014336.4	NG_008474.1
<i>ALMS1</i>	606844	AR	Alstrom syndrome	291	Cilia function	NM_015120.4	NG_011690.1
<i>ARL2BP</i>	615407	AR	Retinitis pigmentosa +/- situs inversus	4	Transcription (cofactor)	NM_012106.3	NG_033905.1
<i>ARL6</i>	608845	AR	Bardet-Biedl syndrome 3	21	Cilia function	NM_177976.2	NG_008119.2
<i>BBIP1</i>	613605	AR	Bardet-Biedl syndrome 18	1	Cilia function	NM_001195306.1	NG_041778.1
<i>BBS1</i>	209901	AR	Bardet-Biedl syndrome 1	94	Cilia function	NM_024649.4	NG_009093.1
<i>BBS10</i>	610148	AR	Bardet-Biedl syndrome 10	92	Cilia function	NM_024685.3	NG_016357.1
<i>BBS12</i>	610683	AR	Bardet-Biedl syndrome 12	50	Cilia function	NM_152618.2	NG_021203.1
<i>BBS2</i>	606151	AR	Bardet-Biedl syndrome 2	81	Cilia function	NM_031885.3	NG_009312.1
<i>BBS4</i>	600374	AR	Bardet-Biedl syndrome 4	47	Cilia function	NM_033028.4	NG_009416.2
<i>BBS5</i>	603650	AR	Bardet-Biedl syndrome 5	25	Cilia function	NM_152384.2	NG_011567.1

<i>BBS7</i>	607590	AR	Bardet-Biedl syndrome 7	38	Cilia function	NM_176824.2	NG_009111.1
<i>BBS9</i>	607968	AR	Bardet-Biedl syndrome 9	41	Cilia function	NM_198428.2	NG_009306.2
<i>BEST1 (AD)</i>	607854	AD	Macular dystrophy, vitelliform 2	292	Ion channel (Ca2+)	NM_004183.3	NG_009033.1
<i>BEST1 (AR)</i>	607854	AR	Bestrophinopathy		Ion channel (Ca2+)	NM_004183.3	NG_009033.1
<i>C1QTNF5</i>	608752	AD	Retinal degeneration late onset	6	Cell adhesion/structure	NM_015645.4	NG_012235.1
<i>C21ORF2</i>	603191	AR	Early onset retinal dystrophy	17	Cilia function	NM_004928.2	NG_032952.1
<i>PCARE</i>	613425	AR	Retinitis pigmentosa 54	40	Cilia function	NM_001029883.2	NG_021427.1
<i>C8ORF37</i>	614477	AR	Bardet-Biedl syndrome 21; Cone-rod dystrophy 16; Retinitis pigmentosa 64	12	Cilia function	NM_177965.3	NG_032804.1
<i>CA4</i>	114760	AD	Retinitis pigmentosa 17	8	Acid overload removal	NM_000717.3	NG_012050.2
<i>CABP4</i>	608965	AR	Cone-rod synaptic disorder, congenital non progressive	11	Synaptic function. Ca2+ influx regulator	NM_145200.3	NG_021211.1
<i>CACNA1F</i>	300110	XL	Åland eye disease; Night blindness congenital stationary	170	Ion channel (Ca2+)	NM_005183.3	NG_009095.2
<i>CACNA2D4</i>	608171	AR	Retinal cone dystrophy	4	Ion channel (Ca2+)	NM_172364.4	NG_012663.1
<i>CDH23</i>	605516	AR	Usher type 1D; deafness	288	Cell adhesion/structure	NM_022124.5	NG_008835.1
<i>CDH3</i>	114021	AR	Hypotrichosis congenital with juvenile macular dystrophy; Ectodermal	29	Cell adhesion/structure	NM_001793.4	NG_009096.1
<i>CDHR1</i>	609502	AR	Retinitis pigmentosa 65; Cone-rod dystrophy 15	34	Cell adhesion/structure	NM_033100.3	NG_028034.1
<i>CEP290</i>	610142	AR	Leber congenital amaurosis 10; Meckel syndrome 4; Senior Loken syndrome 6;	262	Cilia function	NM_025114.3	NG_008417.1

<i>CERKL</i>	608381	AR	Retinitis pigmentosa 26	34	No information	NM_001030311.2	NG_021178.1
<i>CHM</i>	300390	XL	Choroideremia	273	Signal transduction	NM_000390.2	NG_009874.2
<i>CLRN1</i>	606397	AR	Usher syndrome type 3A; Retinitis pigmentosa 61	37	No information	NM_174878.2	NG_009168.1
<i>CNGA1</i>	123825	AR	Retinitis pigmentosa 49	27	Phototransduction	NM_000087.3	NG_009193.1
<i>CNGB1</i>	600724	AR	Retinitis pigmentosa 45	34	Phototransduction	NM_001297.4	NG_016351.1
<i>CNGB3</i>	605080	AR	Achromatopsia 3	114	Phototransduction	NM_019098.4	NG_016980.1
<i>CNNM4</i>	607805	AR	Jalili syndrome	23	No information	NM_020184.3	NG_016608.1
<i>CRB1</i>	604210	AR	Leber congenital amaurosis 8; Retinitis pigmentosa 12	299	Cell adhesion/structure	NM_201253.2	NG_008483.2
<i>CRX</i>	602225	AD	Cone-rod dystrophia; Leber congenital amaurosis 7	92	Transcription factor	NM_000554.4	NG_008605.1
<i>DHDDS</i>	608172	AR	Retinitis pigmentosa 59	8	No information	NM_024887.3	NG_029786.1
<i>ELOVL4</i>	605512	AD	Stargardt disease 3	11	Cell adhesion/structure	NM_022726.3	NG_009108.1
<i>EYS</i>	612424	AR	Retinitis pigmentosa 25	252	Cell adhesion/structure	NM_001142800.1	NG_023443.2
<i>FAM161A</i>	613596	AR	Retinitis pigmentosa 28	17	Cilia function	NM_001201543.1	NG_028125.1
<i>FLVCR1</i>	609144	AR	Ataxia, posterior column, with retinitis pigmentosa	14	Transport (heme)	NM_014053.3	NG_028131.1
<i>FSCN2</i>	607643	AD	Reported retinitis pigmentosa 30	1	No information	NM_001077182.2	NG_015964.1
<i>GNAT1</i>	139330	AD	Night blindness congenital stationary 3	7	Phototransduction	NM_144499.2	NG_009831.1

<i>GPR179</i>	614615	AR	Night blindness congenital stationary	14	Signal transduction (G-coupled 7 TM receptor)	NM_001004334.3	NG_032655.2
<i>GRK1</i>	180381	AR	Oguchi disease 2	16	Phototransduction	NM_002929.2	
<i>GRM6</i>	604096	AR	Night blindness congenital stationary 1B	35	Signal transduction (glutamate receptor)	NM_000843.3	NG_008105.1
<i>GUCA1A</i>	600364	AD	Cone dystrophy 3; Cone-rod dystrophy 14	20	Phototransduction	NM_000409.3	NG_009938.1
<i>GUCA1B</i>	602275	AD	Retinitis pigmentosa 48	4	Phototransduction	NM_002098.5	NG_016216.1
<i>GUCY2D (AD)</i>	600179	AD	Cone-rod dystrophy 6 (AD)	219	Phototransduction	NM_000180.3	NG_009092.1
<i>GUCY2D (AR)</i>	600179	AR	Leber congenital amaurosis 1 (AR);		Phototransduction	NM_000180.3	NG_009092.1
<i>IDH3B</i>	604526	AR	Retinitis pigmentosa 46	0	Citric acid cycle	NM_006899.3	NG_012149.1
<i>IMPDH1</i>	146690	AD	Retinitis pigmentosa 10; Leber congenital amaurosis 11	15	Cell growth	NM_000883.3	NG_009194.1
<i>IMPG2</i>	607056	AR	Retinitis pigmentosa 56; Macular dystrophy	34	Cell adhesion/structure	NM_016247.3	NG_028284.1
<i>INPP5E</i>	613037	AR	Joubert syndrome 1	46	Signal transduction	NM_019892.4	NG_016126.1
<i>IQCB1</i>	609237	AR	Senior Loken syndrome	39	Cilia function	NM_001023570.2	NG_015887.1
<i>KCNV2</i>	607604	AR	Retinal cone dystrophy	87	Ion channel (K+)	NM_133497.3	NG_012181.1
<i>KLHL7</i>	611119	AD	Retinitis pigmentosa 42	11	No information	NM_001031710.2	NG_016983.1
<i>LCA5</i>	611408	AR	Leber congenital amaurosis 5	47	Cilia function	NM_181714.3	NG_016011.1
<i>LRAT</i>	604863	AR	Leber congenital amaurosis 14	16	Visual cycle	NM_004744.4	NG_009110.1

<i>LRIT3</i>	615004	AR	Night blindness congenital stationary	5	Cell adhesion/structure	NM_198506.4	NG_033249.1
<i>LZTFL1</i>	606568	AR	Bardet-Biedl syndrome 17	3	Cilia function	NM_020347.3	NG_033917.1
<i>MAK</i>	154235	AR	Retinitis pigmentosa 62	16	Cilia function	NM_001242957.1	NG_030040.1
<i>MERTK</i>	604705	AR	Retinitis pigmentosa 38	58	Phagocytosis	NM_006343.2	NG_011607.1
<i>MKKS</i>	604896	AR	Bardet-Biedl syndrome 6; McKusick Kaufmann syndrome	55	Cilia function	NM_018848.3	NG_009109.1
<i>MKS1</i>	609883	AR	Bardet-Biedl syndrome13; Joubert syndrome; Meckel syndrome	46	Cilia function	NM_017777.3	NG_013032.1
<i>MVK</i>	251170	AR	Retinitis pigmentosa	169	Signal transduction	NM_000431.3	NG_007702.1
<i>MYO7A</i>	276903	AR	Usher syndrome 1B; isolated deafness	468	Cilia function	NM_000260.3	NG_009086.1
<i>NR2E3 (AD)</i>	604485	AD	Retinitis pigmentosa 37 (AD)	69	Transcription factor	NM_014249.3	NG_009113.2
<i>NR2E3 (AR)</i>	604485	AR	Enhanced S-cone syndrome (AR)		Transcription factor	NM_014249.3	NG_009113.2
<i>NRL</i>	162080	AD	Retinitis pigmentosa 27	23	Transcription factor	NM_006177.3	NG_011697.1
<i>NYX</i>	300278	XL	Night blindness congenital stationary	87	No information	NM_022567.2	NG_009112.1
<i>OFD1</i>	300170	XL	Joubert syndrome 10	152	Cilia function	NM_003611.2	NG_008872.1
<i>PCDH15</i>	605514	AR	Usher syndrome 1F; isolated deafness	101	Cell adhesion/structure	NM_033056.3	NG_009191.2
<i>PDE6A</i>	180071	AR	Retinitis pigmentosa 43	38	Phototransduction	NM_000440.2	NG_009102.1
<i>PDE6B (AD)</i>	180072	AD	NBSC (AD)	103	Phototransduction	NM_000283.3	NG_009839.1

<i>PDE6B (AR)</i>	180072	AR	Retinitis pigmentosa 40 (AR)		Phototransduction	NM_000283.3	NG_009839.1
<i>PDE6C</i>	600827	AR	Cone dystrophy 4	36	Phototransduction	NM_006204.3	NG_016752.1
<i>PDE6G</i>	180073	AR	Retinitis pigmentosa 57	2	Phototransduction	NM_002602.3	NG_009834.1
<i>PDZD7</i>	612971	?	Usher syndrome 2C	18	Cilia function	NM_001195263.1	NG_028030.1
<i>PITPNM3</i>	608921	AD	Cone-rod dystrophy 5	3	Signal transduction (phosphatidylinositol transfer)	NM_031220.3	NG_016020.1
<i>PRCD</i>	610598	AR	Retinitis pigmentosa 36	6	No information	NM_001077620.2	NG_016702.1
<i>PROM1</i>	604365	AR	Cone-rod dystrophy 12; Macular dystrophy 2; Retinitis pigmentosa 41 (1	58	Cellular structure	NM_006017.2	NG_011696.1
<i>PRPF3</i>	607301	AD	Retinitis pigmentosa 18	6	Splicing factor	NM_004698.2	NG_008245.1
<i>PRPF31</i>	606419	AD	Retinitis pigmentosa 11	142	Splicing factor	NM_015629.3	NG_009759.1
<i>PRPF6</i>	613979	AD	Retinitis pigmentosa 60	4	Splicing factor	NM_012469.3	NG_029719.1
<i>PRPF8</i>	607300	AD	Retinitis pigmentosa 13	37	Splicing factor	NM_006445.3	NG_009118.1
<i>PRPH2 (RDS)</i>	179605	AD + DIGEN (+ROM1)	Retinitis pigmentosa 7; Leber congenital amaurosis 18; Macualr	161	Cell adhesion/structure	NM_000322.4	NG_009176.1
<i>RAB28</i>	612994	AR	Cone-rod dystrophy 18	4	Intracellular trafficking	NM_004249.3	NG_033891.1
<i>RAX2</i>	610362	AD	Cone-rod dystrophy 11	4	Transcription	NM_032753.3	NG_011565.1
<i>RBP3</i>	180290	AR	?Retinitis pigmentosa 66	8	Visual cycle	NM_002900.2	NG_029718.1
<i>RBP4</i>	180250	AR	Retinal dystrophy plus colobom; microphthalmia plus colobom	7	Visual cycle	NM_006744.3	NG_009104.1

<i>RDH12</i>	608830	AR	Leber congenital amaurosis 13	92	Visual cycle	NM_152443.2	NG_008321.1
<i>RDH5</i>	601617	AR	Fundus albipunctatus	48	Visual cycle	NM_002905.3	NG_008606.1
<i>RGR</i>	600342	AR	Retinitis pigmentosa 44	8	Signal transduction (G-coupled 7 TM receptor)	NM_001012720.1	NG_009106.1
<i>RGS9</i>	604067	AR	Bradyopsia	2	Phototransduction	NM_003835.3	NG_013021.1
<i>RGS9BP</i>	607814	AR	Bradyopsia	6	Phototransduction	NM_207391.2	NG_016751.1
<i>RHO</i>	180380	AD	Retinitis pigmentosa 4	198	Phototransduction	NM_000539.3	NG_009115.1
<i>RIMS1</i>	606629	AD	Cone-rod dystrophy 7	4	Exocytosis	NM_014989.5	NG_016209.1
<i>RLBP1</i>	180090	AR	Rod-cone dystrophy; retinitis puctata albescens	31	Visual cycle	NM_000326.4	NG_008116.1
<i>ROM1</i>	180721	DIGEN (+PRPH)	Retinitis pigmentosa 7	11	Disc morphogenesis	NM_000327.3	NG_009845.1
<i>RP1 (AD)</i>	603937	AD	Retinitis pigmentosa 1	153	Cilia function	NM_006269.1	NG_009840.1
<i>RP1 (AR)</i>	603937	AR	Retinitis pigmentosa 1		Cilia function	NM_006269.1	NG_009840.1
<i>RP1L1</i>	608581	AR	Occult macular dystrophy	27	Cilia function	NM_178857.5	NG_028035.1
<i>RP2</i>	300757	XL	Retinitis pigmentosa 2	106	Signal transduction	NM_006915.2	NG_009107.1
<i>RP9</i>	607331	AD	?Retinitis pigmentosa 9	0	Splicing factor	NM_203288.1	NG_012968.1
<i>RPE65</i>	180069	AR	Leber congenital amaurosis 2; Retinitis pigmentosa 20	178	Visual cycle	NM_000329.2	NG_008472.1
<i>RPGR</i>	312610	XL	Cone-rod dystrophy 3; Retinitis pigmentosa 3	192	Cilia function	NM_000328.2	NG_009553.1

<i>RPGRIP1</i>	605446	AR	Leber congenital amaurosis 6; Cone-rod dystrophy 13	122	Cilia function	NM_020366.3	NG_008933.1
<i>SAG</i>	181031	AR	Oguchi disease; Retinitis pigmentosa 47	9	Phototransduction	NM_000541.4	NG_009116.1
<i>SDCCAG8</i>	613524	AR	Bardet-Biedl syndrome 16; Senior Loken syndrome	17	Cilia function	NM_006642.3	NG_027811.1
<i>SEMA4A</i>	607292	AR	Cone-rod dystrophy 10; Retinitis pigmentosa 35	7	Cell-cell signalling	NM_022367.3	NG_027683.1
<i>SLC24A1</i>	603617	AR	Night blindness congenital stationary	6	Phototransduction	NM_004727.2	NG_031968.2
<i>SNRNP200</i>	611664	AD	Retinitis pigmentosa 33	21	Splicing factor	NM_014014.4	NG_018973.1
<i>SPATA7</i>	609868	AR	Leber congenital amaurosis 3	36	Cilia function	NM_018418.4	NG_021183.1
<i>TEAD1</i>	189967	AD	Sveinsson chorioretinal atrophy	2	Transcription factor	NM_021961.5	NG_0021302.1
<i>TIMP3</i>	188826	AD	Sorsby fundus dystrophy	17	Matrix metalloproteinase	NM_000362.4	NG_009117.1
<i>TOPORS</i>	609507	AD	Retinitis pigmentosa 31	13	Cilia function	NM_005802.4	NG_017050.1
<i>TRIM32</i>	602290	AR	?Bardet-Biedl syndrome 11	15	E3 ubiquitin ligase	NM_012210.3	NG_011619.1
<i>TRPM1</i>	603576	AR	Night blindness congenital stationary	65	Ion channel (Ca ²⁺)	NM_002420.5	NG_016453.2
<i>TTC8</i>	608132	AR	Bardet-Biedl syndrome 8	13	Cilia function	NM_198309.3	NG_008126.1
<i>TULP1</i>	302280	AR	Leber congenital amaurosis 15; Retinitis pigmentosa 14	62	Cilia function	NM_003322.4	NG_009077.1
<i>USH1C</i>	605242	AR	Usher syndrome 1C; isolated deafness	38	Scaffolding	NM_005709.3	NG_011883.1
<i>USH1G</i>	607696	AR	Usher asynrome 1G	28	Scaffolding	NM_173477.4	NG_007882.2

<i>USH2A</i>	608400	AR	Usher syndrome 2A; Retinitis pigmentosa 39	974	Cilia function	NM_206933.2	NG_009497.1
<i>WHRN</i>	607928	AR	Usher type 1D; Deafness 31	21	Cilia function	NM_015404.3	NG_016700.1
<i>ZNF513</i>	613598	AR	?Retinitis pigmentosa 58	1	Transcription factor (possible)	NM_144631.5	NG_028219.1

Table S3

Target Capture Statistics	The average value for all samples
Target region (bp)	2,168,860
Reads mapped to genome	15,845,300
Bases mapped to target regions (Mb)	1156.22
Reads mapped to target region	12,841,500
Reads mapped to flanking region	1,557,260
Mean depth of target region (X)	533.1
Mean depth of flanking regions(X)	122.24
Coverage of target region (%)	98.74
Fraction of target covered $\geq 4X$ (%)	98.27
Fraction of target covered $\geq 10X$ (%)	97.83
Fraction of target covered $\geq 20X$ (%)	97.29

Table S5

No.	Type	Gene	Inheritance	Exons	Deletion/duplication	Second Variant
25	deletion	ABCA4	AR	ex 50	c.(6816+1_6817-1)(*1?)del#	c.1719G>A p.(Met573Ile)
39	deletion	AIPL1	AR	ex 1-2	c.(?-1)(276+1_277-1)del#	c.815G>C p.(Arg272Pro)
93	deletion	CNGB3	AR	ex 3	c.(211+1_212-1)(338+1_339-1)del# c.(211+1_212-1)(338+1_339-1)del#	
399	duplication	CRX	AD	entire gene	c.(?-1)(*1?)dup#	
121	deletion	EYS	AR	ex 13-22; 15-18;	c.(2023+1_2024-1)(3443+1_3444-1)del# c.(2023+1_2024-1)(3443+1_3444-1)del#	
129	deletion	EYS	AR	ex 17-18	c.(2641+1_2642-1)(2846+1_2847-1)del#	c.6714del p.(Ile2239Serfs*17)
403	deletion	EYS	AR	ex 30	c.(6078+1_6079-1)(6191+1_6192-1)del	
126	duplication	EYS	AR	ex 14	c.(2137+1_2138-1)(2259+1_2260-1)dup#	c.6714del p.(Ile2239Serfs*17)
122	duplication	EYS	AR	ex 30	c.(6078+1_6079-1)(6191+1_6192-1)dup# c.(6078+1_6079-1)(6191+1_6192-1)dup#	
139	deletion	GRM6	AR	ex 7-9	c.(1354+1_1355-1)(2436+1_2437-1)del#	c.1861C>T p.(Arg621*)
152	deletion	MERTK	AR	ex 1-7	c.(?-1)(1144+1_1145-1)del c.(?-1)(1144+1_1145-1)del	
155	deletion	MERTK	AR	ex 1-7	c.(?-1)(1144+1_1145-1)del	c.757+1G>A p.(?)#
424	duplication	MERTK	AR	ex 2	c.(61+1_62-1)(482+1_483-1)dup#	
189	deletion	PRPF31	AD	ex 2-14	c.(?-1)(*1?)del#	
183	deletion	PRPF31	AD	ex 2-14	c.(?-1)(*1?)del#	
194	deletion	PRPF31	AD	ex 2-3	c.(?-1)(238+1_239-1)del	
187	duplication	PRPF31	AD	ex 4-5	c.(238+1_239-1)(420+1_421-1)dup#	
466	deletion	SAG	AR	ex 8-10	c.(512+1_513-1)(806+1_807-1)del#	
271	deletion	USH2A	AR	ex 22-24	c.(4627+1_4628-1)(4987+1_4988-1)del	c.12161G>T p.(Ser4054Ile)
297	duplication	USH2A	AR	ex 12-13-14	c.(1971+1_1972-1)(2993+1_2994-1)dup#	c.10561T>C p.(Trp3521Arg)

Table S6

EYS

HGVS c. (Clinically Relevant)	HGVS p. (Clinically Relevant)	Chr:Pos	Ref/Alt	Identifier	Haplotype
NM_001142800:c.8779T>C	p.Cys2927Arg	6:64431148	A/G	rs373203896	REF
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NM_001142800:c.6835-124A>G		6:64694620	T/C	rs1482456	REF
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NM_001142800:c.-332-50864C>T	6:66256750	G/A	rs34586455	REF
NM_001142800:c.-332-51310T>C	6:66257196	A/G	rs9363391	ALT
NM_001142800:c.-332-52016T>A	6:66257902	A/T	rs1411839	ALT
NM_001142800:c.-332-52081C>T	6:66257967	G/A	rs1411840	REF
NM_001142800:c.-332-52214A>G	6:66258100	T/C	rs2351262	ALT
NM_001142800:c.-332-52293T>G	6:66258179	A/C	rs1411841	ALT
NM_001142800:c.-332-52347C>T	6:66258233	G/A	rs1929331	ALT
NM_001142800:c.-332-52717T>A	6:66258603	A/T	rs1929332	ALT
NM_001142800:c.-332-52736G>C	6:66258622	C/G	rs1929333	ALT
NM_001142800:c.-332-52765G>A	6:66258651	C/T	rs1929334	ALT
NM_001142800:c.-332-52892A>G	6:66258778	T/C	rs1331197	ALT
NM_001142800:c.-332-53065G>T	6:66258951	C/A	rs9363392	ALT
NM_001142800:c.-332-53332C>A	6:66259218	G/T	rs76838586	REF
NM_001142800:c.-332-53472A>G	6:66259358	T/C	rs6937452	ALT
NM_001142800:c.-332-53524A>C	6:66259410	T/G	rs6937478	ALT
NM_001142800:c.-332-53535C>T	6:66259421	G/A	rs6455046	ALT
NM_001142800:c.-332-53622T>C	6:66259508	A/G	rs6932966	ALT
NM_001142800:c.-332-53651C>T	6:66259537	G/A	rs6909711	ALT
NM_001142800:c.-332-53660T>C	6:66259546	A/G	rs6455047	REF
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NM_001142800:c.-332-56346C>T	6:66262232	G/A	rs4524589	ALT
NM_001142800:c.-332-56445A>C	6:66262331	T/G	rs9453326	ALT
NM_001142800:c.-332-56452C>T	6:66262338	G/A	rs9363393	ALT
NM_001142800:c.-332-56520A>G	6:66262406	T/C	rs4506027	ALT
NM_001142800:c.-332-56615A>T	6:66262501	T/A	rs9342485	ALT
NM_001142800:c.-332-56720T>G	6:66262606	A/C	rs9360135	ALT
NM_001142800:c.-332-56809A>G	6:66262695	T/C	rs9345650	ALT
NM_001142800:c.-332-56834C>T	6:66262720	G/A	rs3908650	ALT
NM_001142800:c.-332-56933A>G	6:66262819	T/C	rs3908651	ALT
NM_001142800:c.-332-56935C>T	6:66262821	G/A	rs9363394	ALT
NM_001142800:c.-332-57162G>A	6:66263048	C/T	rs3908652	ALT
NM_001142800:c.-332-57335T>C	6:66263221	A/G	rs7452712	ALT
NM_001142800:c.-332-57368T>G	6:66263254	A/C	rs9354265	ALT
NM_001142800:c.-332-57696A>T	6:66263582	T/A	rs3846802	ALT
NM_001142800:c.-332-57768A>G	6:66263654	T/C	rs3857534	ALT

Table S6

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NM_001142800:c.-332-58293G>A	6:66264179	C/T	rs2150619	ALT
NM_001142800:c.-332-58500T>A	6:66264386	A/T	rs2351265	ALT
NM_001142800:c.-332-58552A>C	6:66264438	T/G	rs6935790	ALT
NM_001142800:c.-332-58621T>C	6:66264507	A/G	rs1411847	ALT
NM_001142800:c.-332-58849A>G	6:66264735	T/C	rs1411848	ALT
NM_001142800:c.-332-58869C>T	6:66264755	G/A	rs1411849	ALT
NM_001142800:c.-332-59017delT	6:66264903	A/-	rs5876972	ALT
NM_001142800:c.-332-59045T>C	6:66264931	A/G	rs1929335	ALT
NM_001142800:c.-332-59217A>G	6:66265103	T/C	rs1929336	ALT
NM_001142800:c.-332-59938T>G	6:66265824	A/C	rs1999363	ALT
NM_001142800:c.-332-60418T>C	6:66266304	A/G	rs2351266	ALT
NM_001142800:c.-332-60657A>G	6:66266543	T/C	rs7743334	REF
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NM_001142800:c.-332-61647C>T	6:66267533	G/A	rs6926034	ALT
NM_001142800:c.-332-61961G>C	6:66267847	C/G	rs6932247	ALT
NM_001142800:c.-332-62068G>A	6:66267954	C/T	rs7767609	REF
NM_001142800:c.-332-62297G>A	6:66268183	C/T	rs9360136	ALT
NM_001142800:c.-332-62310T>C	6:66268196	A/G	rs1411852	ALT
NM_001142800:c.-332-62543C>T	6:66268429	G/A	rs1411853	REF
NM_001142800:c.-332-62874C>T	6:66268760	G/A	rs1473778	ALT
NM_001142800:c.-332-62900A>T	6:66268786	T/A	rs1473779	ALT
NM_001142800:c.-332-63513T>C	6:66269399	A/G	rs1331205	REF
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NM_001142800:c.-332-64078A>G	6:66269964	T/C	rs1473780	ALT
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NM_001142800:c.-332-65514G>T	6:66271400	C/A	rs2225457	ALT
NM_001142800:c.-332-65551G>A	6:66271437	C/T	rs13204681	REF
NM_001142800:c.-332-65557G>A	6:66271443	C/T	rs9363396	ALT
NM_001142800:c.-332-66014C>T	6:66271900	G/A	rs7349900	ALT
NM_001142800:c.-332-67582dupA	6:66273468	-/T	rs201551142	0/1
NM_001142800:c.-332-67590T>A	6:66273476	A/T	rs1331206	ALT
NM_001142800:c.-332-67724A>G	6:66273610	T/C	rs1331207	ALT
NM_001142800:c.-332-67918A>C	6:66273804	T/G		REF
NM_001142800:c.-332-67921C>T	6:66273807	G/A		REF
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NM_001142800:c.-332-70468G>T	6:66276354	C/A	rs12191952	ALT

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NM_001142800:c.-332-71287G>A	6:66277173	C/T	rs4235971	ALT
NM_001142800:c.-332-71530C>T	6:66277416	G/A	rs9342486	REF
NM_001142800:c.-332-71742C>G	6:66277628	G/C	rs34412399	REF
NM_001142800:c.-333+71756C>A	6:66277915	G/T	rs7753548	ALT
NM_001142800:c.-333+71469delA	6:66278202	T/-	rs61492530	./1
NM_001142800:c.-333+71096C>T	6:66278575	G/A	rs13203580	REF
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NM_001142800:c.-333+70136G>C	6:66279535	C/G	rs9345654	ALT
NM_001142800:c.-333+69967C>G	6:66279704	G/C	rs9363398	REF
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NM_001142800:c.-333+69606T>C	6:66280065	A/G	rs66812553	ALT
NM_001142800:c.-333+69487T>C	6:66280184	A/G	rs9360138	ALT
NM_001142800:c.-333+68564G>A	6:66281107	C/T	rs34268718	REF
NM_001142800:c.-333+68363G>C	6:66281308	C/G	rs6455048	REF
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NM_001142800:c.-333+67447G>A	6:66282224	C/T	rs6650991	ALT
NM_001142800:c.-333+67298G>A	6:66282373	C/T	rs1331203	ALT
NM_001142800:c.-333+67285C>G	6:66282386	G/C	rs1331204	ALT
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NM_001142800:c.-333+65485C>T	6:66284186	G/A	rs9360139	ALT
NM_001142800:c.-333+65425C>G	6:66284246	G/C	rs9363400	ALT
NM_001142800:c.-333+65421T>A	6:66284250	A/T	rs1590965	ALT
NM_001142800:c.-333+65029A>G	6:66284642	T/C	rs76704490	REF
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NM_001142800:c.-333+63615G>C	6:66286056	C/G	rs354346	ALT
NM_001142800:c.-333+63216A>G	6:66286455	T/C	rs354347	ALT
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NM_001142800:c.-333+62306G>T	6:66287365	C/A	rs354349	ALT
NM_001142800:c.-333+62129A>G	6:66287542	T/C	rs354350	ALT
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NM_001142800:c.-333+55810delT	6:66293861	A/-	rs61362090	REF
NM_001142800:c.-333+54846A>G	6:66294825	T/C	rs354358	ALT
NM_001142800:c.-333+53899C>T	6:66295772	G/A	rs374738	ALT
NM_001142800:c.-333+53641C>T	6:66296030	G/A	rs35936441	REF
NM_001142800:c.-333+53476G>C	6:66296195	C/G	rs354359	ALT
NM_001142800:c.-333+52809T>C	6:66296862	A/G	rs188298223	REF
NM_001142800:c.-333+51495A>G	6:66298176	T/C	rs354361	ALT
NM_001142800:c.-333+51358G>A	6:66298313	C/T	rs6902867	REF
NM_001142800:c.-333+51214delA	6:66298457	T/-	rs35933552	ALT
NM_001142800:c.-333+49538T>C	6:66300133	A/G	rs13194431	REF
NM_001142800:c.-333+49184A>G	6:66300487	T/C	rs34018715	REF
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NM_001142800:c.-333+48921dupA	6:66300750	-/T		0/1
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NM_001142800:c.-333+48659G>A	6:66301012	C/T	rs62407719	REF
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NM_001142800:c.-333+48461C>G	6:66301210	G/C	rs115835301	ALT
NM_001142800:c.-333+48447A>T	6:66301224	T/A	rs116265847	ALT
NM_001142800:c.-333+48441G>C	6:66301230	C/G	rs9363403	ALT
NM_001142800:c.-333+48228G>A	6:66301443	C/T	rs12199868	ALT
NM_001142800:c.-333+47764T>G	6:66301907	A/C	rs354362	ALT
NM_001142800:c.-333+47719T>C	6:66301952	A/G	rs9351513	REF
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NM_001142800:c.-333+45508A>G	6:66304163	T/C	rs354364	REF
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NM_001142800:c.-333+43773T>C	6:66305898	A/G	rs9354267	ALT
NM_001142800:c.-333+43435dupA	6:66306236	-/T	rs11443686	ALT
NM_001142800:c.-333+42528T>C	6:66307143	A/G	rs2183675	ALT
NM_001142800:c.-333+41716A>G	6:66307955	T/C	rs189550	REF
NM_001142800:c.-333+41544T>G	6:66308127	A/C		0/1

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NM_001142800:c.-333+39364G>T	6:66310307	C/A	rs12206843	ALT
NM_001142800:c.-333+39185G>A	6:66310486	C/T	rs16896896	REF
NM_001142800:c.-333+36525C>T	6:66313146	G/A	rs9453339	REF
NM_001142800:c.-333+35789G>A	6:66313882	C/T	rs168514	ALT
NM_001142800:c.-333+35748G>A	6:66313923	C/T	rs1929323	ALT
NM_001142800:c.-333+35645T>G	6:66314026	A/C	rs1387793	ALT
NM_001142800:c.-333+35521A>G	6:66314150	T/C	rs16896908	REF
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NM_001142800:c.-333+34080A>C	6:66315591	T/G	rs354400	REF
NM_001142800:c.-333+33938C>T	6:66315733	G/A		REF
NM_001142800:c.-333+33750A>G	6:66315921	T/C	rs6930884	ALT
NM_001142800:c.-333+33523G>T	6:66316148	C/A	rs1907018	ALT
NM_001142800:c.-333+33444G>A	6:66316227	C/T	rs354398	ALT
NM_001142800:c.-333+33308T>G	6:66316363	A/C	rs1907017	ALT
NM_001142800:c.-333+33211T>A	6:66316460	A/T	rs1907016	ALT
NM_001142800:c.-333+33129G>T	6:66316542	C/A	rs1907015	ALT
NM_001142800:c.-333+32893dupA	6:66316778	-/T	rs140167236	1/.
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NM_001142800:c.-333+32334A>G	6:66317337	T/C	rs354394	ALT
NM_001142800:c.-333+32304G>A	6:66317367	C/T	rs7748738	ALT
NM_001142800:c.-333+31531T>A	6:66318140	A/T	rs354392	ALT
NM_001142800:c.-333+31086G>A	6:66318585	C/T	rs10447346	ALT
NM_001142800:c.-333+30946T>G	6:66318725	A/C	rs10447316	ALT

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NM_001142800:c.-333+30191A>C	6:66319480	T/G	rs354391	ALT
NM_001142800:c.-333+29854A>T	6:66319817	T/A	rs7745746	ALT
NM_001142800:c.-333+29768G>A	6:66319903	C/T	rs1027188	ALT
NM_001142800:c.-333+29674A>C	6:66319997	T/G	rs35472556	REF
NM_001142800:c.-333+29501G>A	6:66320170	C/T	rs2351267	ALT
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NM_001142800:c.-333+29152G>T	6:66320519	C/A	rs1490142	ALT
NM_001142800:c.-333+28901A>G	6:66320770	T/C	rs354387	ALT
NM_001142800:c.-333+28799C>T	6:66320872	G/A	rs191001672	REF
NM_001142800:c.-333+28599dupT	6:66321072	-/A	rs200048825	ALT
NM_001142800:c.-333+28545A>T	6:66321126	T/A	rs12198998	ALT
NM_001142800:c.-333+28521G>A	6:66321150	C/T	rs354386	ALT
NM_001142800:c.-333+27625T>C	6:66322046	A/G	rs12199458	ALT
NM_001142800:c.-333+27555T>C	6:66322116	A/G	rs183631341	REF
NM_001142800:c.-333+27548A>G	6:66322123	T/C	rs12200913	ALT
NM_001142800:c.-333+27090G>A	6:66322581	C/T	rs7747445	ALT
NM_001142800:c.-333+27006dupT	6:66322665	-/A	rs11385339	ALT
NM_001142800:c.-333+26470G>A	6:66323201	C/T	rs13201651	REF
NM_001142800:c.-333+26464G>A	6:66323207	C/T	rs10806512	ALT
NM_001142800:c.-333+26261C>T	6:66323410	G/A	rs354382	ALT
NM_001142800:c.-333+25792C>T	6:66323879	G/A	rs2130498	ALT
NM_001142800:c.-333+25678A>C	6:66323993	T/G	rs1980987	ALT
NM_001142800:c.-333+25409T>G	6:66324262	A/C	rs7739112	ALT
NM_001142800:c.-333+25311C>G	6:66324360	G/C	rs168512	ALT
NM_001142800:c.-333+25297dupA	6:66324374	-/T	rs35160142	REF
NM_001142800:c.-333+25176C>T	6:66324495	G/A	rs354381	ALT
NM_001142800:c.-333+24894_-333+24895insT	6:66324777	-/A	rs5876977	ALT
NM_001142800:c.-333+24879G>A	6:66324792	C/T	rs7758595	ALT
NM_001142800:c.-333+24652A>T	6:66325019	T/A	rs971620	REF
NM_001142800:c.-333+24033A>G	6:66325638	T/C	rs2046837	REF
NM_001142800:c.-333+24028G>A	6:66325643	C/T	rs2046836	REF
NM_001142800:c.-333+23938C>T	6:66325733	G/A	rs2046835	REF
NM_001142800:c.-333+23789A>T	6:66325882	T/A	rs12208402	ALT
NM_001142800:c.-333+23372delT	6:66326299	A/-	rs66800400	ALT
NM_001142800:c.-333+23067C>G	6:66326604	G/C	rs421733	REF
NM_001142800:c.-333+23063C>G	6:66326608	G/C	rs12213513	ALT
NM_001142800:c.-333+22994dupT	6:66326677	-/A	rs34322899	REF
NM_001142800:c.-333+22986delT	6:66326685	A/-		REF
NM_001142800:c.-333+22726A>G	6:66326945	T/C	rs13207080	REF
NM_001142800:c.-333+22654C>G	6:66327017	G/C	rs973300	REF
NM_001142800:c.-333+22641G>C	6:66327030	C/G	rs973299	ALT

Table S6

NM_001142800:c.-333+22620C>T	6:66327051	G/A	rs2351268	ALT
NM_001142800:c.-333+22501G>A	6:66327170	C/T	rs2351269	ALT
NM_001142800:c.-333+22039A>G	6:66327632	T/C	rs12210732	ALT
NM_001142800:c.-333+22025G>T	6:66327646	C/A	rs10944817	ALT
NM_001142800:c.-333+18242G>C	6:66331429	C/G	rs168511	REF
NM_001142800:c.-333+18158A>G	6:66331513	T/C	rs373700804	REF
NM_001142800:c.-333+18062T>C	6:66331609	A/G	rs187126	ALT
NM_001142800:c.-333+17802A>C	6:66331869	T/G	rs4298329	ALT
NM_001142800:c.-333+17323G>A	6:66332348	C/T	rs354371	ALT
NM_001142800:c.-333+17022delA	6:66332649	T/-	rs35201653	./1
NM_001142800:c.-333+16967T>C	6:66332704	A/G	rs354370	ALT
NM_001142800:c.-333+16771A>G	6:66332900	T/C	rs6926767	ALT
NM_001142800:c.-333+16566G>A	6:66333105	C/T	rs6900513	ALT
NM_001142800:c.-333+16152T>A	6:66333519	A/T	rs354369	ALT
NM_001142800:c.-333+16049G>C	6:66333622	C/G	rs728880	ALT
NM_001142800:c.-333+15999C>T	6:66333672	G/A	rs902287	ALT
NM_001142800:c.-333+15945G>C	6:66333726	C/G	rs728881	ALT
NM_001142800:c.-333+15397C>G	6:66334274	G/C	rs902286	ALT
NM_001142800:c.-333+15344C>T	6:66334327	G/A	rs4446546	ALT
NM_001142800:c.-333+14811A>T	6:66334860	T/A	rs12215492	ALT
NM_001142800:c.-333+14207C>T	6:66335464	G/A	rs1490139	ALT
NM_001142800:c.-333+14047T>A	6:66335624	A/T	rs354367	ALT
NM_001142800:c.-333+13705C>T	6:66335966	G/A	rs766199	ALT
NM_001142800:c.-333+13273C>T	6:66336398	G/A	rs6917909	ALT
NM_001142800:c.-333+13204G>T	6:66336467	C/A	rs6919315	ALT
NM_001142800:c.-333+13154A>T	6:66336517	T/A	rs968893	ALT
NM_001142800:c.-333+12908A>G	6:66336763	T/C	rs6903740	ALT
NM_001142800:c.-333+12872G>T	6:66336799	C/A	rs6919950	REF
NM_001142800:c.-333+12351T>C	6:66337320	A/G	rs9453347	ALT
NM_001142800:c.-333+12327G>A	6:66337344	C/T	rs139174916	REF
NM_001142800:c.-333+12079C>T	6:66337592	G/A	rs12197389	ALT
NM_001142800:c.-333+12029C>T	6:66337642	G/A	rs111411564	REF
NM_001142800:c.-333+11998G>A	6:66337673	C/T	rs12211326	ALT
NM_001142800:c.-333+11893G>A	6:66337778	C/T	rs12211385	ALT
NM_001142800:c.-333+11204C>T	6:66338467	G/A	rs6930269	ALT
NM_001142800:c.-333+11092T>C	6:66338579	A/G	rs62407730	REF
NM_001142800:c.-333+10904A>G	6:66338767	T/C	rs10455583	ALT
NM_001142800:c.-333+10823C>T	6:66338848	G/A	rs10455188	ALT
NM_001142800:c.-333+10537A>G	6:66339134	T/C	rs76385824	REF
NM_001142800:c.-333+10511C>G	6:66339160	G/C	rs10455189	ALT
NM_001142800:c.-333+10166T>C	6:66339505	A/G	rs12195089	ALT
NM_001142800:c.-333+9468G>A	6:66340203	C/T	rs1873292	ALT
NM_001142800:c.-333+9239G>A	6:66340432	C/T	rs2130493	ALT
NM_001142800:c.-333+9192A>G	6:66340479	T/C	rs10944827	ALT
NM_001142800:c.-333+9092G>A	6:66340579	C/T	rs10944828	ALT
NM_001142800:c.-333+8453A>G	6:66341218	T/C	rs1387800	ALT
NM_001142800:c.-333+8302dupT	6:66341369	-/A	rs5876981	0/1
NM_001142800:c.-333+7859C>A	6:66341812	G/T	rs6916915	ALT
NM_001142800:c.-333+7725G>A	6:66341946	C/T	rs16896967	REF

Table S6

NM_001142800:c.-333+7615A>C	6:66342056	T/G	rs13195796	REF
NM_001142800:c.-333+6998A>G	6:66342673	T/C	rs7745805	REF
NM_001142800:c.-333+5471G>C	6:66344200	C/G	rs10944829	ALT
NM_001142800:c.-333+4816G>A	6:66344855	C/T	rs1873291	REF
NM_001142800:c.-333+4680A>T	6:66344991	T/A	rs7760787	ALT
NM_001142800:c.-333+4573G>A	6:66345098	C/T	rs79739949	REF
NM_001142800:c.-333+4522G>A	6:66345149	C/T	rs1907010	ALT
NM_001142800:c.-333+4382C>T	6:66345289	G/A	rs4621617	ALT
NM_001142800:c.-333+3984C>T	6:66345687	G/A	rs7775327	ALT
NM_001142800:c.-333+3938G>T	6:66345733	C/A	rs6899544	ALT
NM_001142800:c.-333+3732G>A	6:66345939	C/T	rs6904116	ALT
NM_001142800:c.-333+3582C>A	6:66346089	G/T	rs1586656	ALT
NM_001142800:c.-333+3555A>G	6:66346116	T/C	rs138554316	REF
NM_001142800:c.-333+3540C>G	6:66346131	G/C	rs1586657	ALT
NM_001142800:c.-333+3483T>A	6:66346188	A/T	rs10944830	ALT
NM_001142800:c.-333+3324T>G	6:66346347	A/C	rs1586658	ALT
NM_001142800:c.-333+3081A>G	6:66346590	T/C	rs1490126	ALT
NM_001142800:c.-333+2789A>T	6:66346882	T/A	rs4458657	ALT
NM_001142800:c.-333+2735A>C	6:66346936	T/G	rs2046838	ALT
NM_001142800:c.-333+2726A>G	6:66346945	T/C	rs2046839	ALT
NM_001142800:c.-333+2170T>G	6:66347501	A/C	rs6933242	ALT
NM_001142800:c.-333+1849G>A	6:66347822	C/T	rs6911669	ALT
NM_001142800:c.-333+1760C>T	6:66347911	G/A	rs12207870	ALT
NM_001142800:c.-333+1506C>T	6:66348165	G/A	rs6915285	ALT
NM_001142800:c.-333+1292G>A	6:66348379	C/T	rs12195141	ALT
NM_001142800:c.-333+1049G>A	6:66348622	C/T	rs6917401	ALT
NM_001142800:c.-447-123A>C	6:66349908	T/G	rs17779372	ALT
NM_001142800:c.-447-529delA	6:66350314	T/-	rs11312384	ALT
NM_001142800:c.-447-1257C>A	6:66351042	G/T	rs7739824	ALT
NM_001142800:c.-447-1551C>T	6:66351336	G/A	rs72648382	ALT
NM_001142800:c.-447-2595C>G	6:66352380	G/C	rs6907955	ALT
NM_001142800:c.-447-2617A>T	6:66352402	T/A	rs3919983	ALT
NM_001142800:c.-447-2642delA	6:66352427	T/-	rs11299994	ALT
NM_001142800:c.-447-2753C>A	6:66352538	G/T	rs6908305	ALT
NM_001142800:c.-447-2816C>A	6:66352601	G/T	rs6908347	ALT
NM_001142800:c.-447-3877T>C	6:66353662	A/G	rs9363410	ALT
NM_001142800:c.-447-4228G>T	6:66354013	C/A	rs2351499	ALT
NM_001142800:c.-447-4603T>C	6:66354388	A/G	rs1826149	ALT
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NM_001142800:c.-447-6123G>A	6:66355908	C/T	rs9363411	ALT
NM_001142800:c.-447-6804T>C	6:66356589	A/G	rs12207318	ALT
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NM_001142800:c.-447-7944T>G	6:66357729	A/C	rs6919287	ALT
NM_001142800:c.-447-8424C>T	6:66358209	G/A	rs112653175	REF
NM_001142800:c.-447-8426C>T	6:66358211	G/A		REF
NM_001142800:c.-447-8467T>C	6:66358252	A/G	rs56361312	ALT
NM_001142800:c.-447-8508A>C	6:66358293	T/G	rs1826151	ALT

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NM_001142800:c.-447-8816dupT	6:66358601	-/A	rs75589705	ALT
NM_001142800:c.-447-9018A>G	6:66358803	T/C	rs7765481	ALT
NM_001142800:c.-447-9038C>T	6:66358823	G/A	rs12190480	ALT
NM_001142800:c.-447-9071C>T	6:66358856	G/A	rs12191907	ALT
NM_001142800:c.-447-9246dupT	6:66359031	-/A	rs71677714	O/1
NM_001142800:c.-447-9275C>T	6:66359060	G/A	rs374536933	REF
NM_001142800:c.-447-9290A>T	6:66359075	T/A	rs372295707	REF
NM_001142800:c.-447-10184T>A	6:66359969	A/T	rs1387795	ALT
NM_001142800:c.-447-10443A>G	6:66360228	T/C	rs1387796	ALT
NM_001142800:c.-447-12600A>G	6:66362385	T/C	rs9345665	ALT
NM_001142800:c.-447-12982A>G	6:66362767	T/C	rs958705	ALT
NM_001142800:c.-447-13032G>A	6:66362817	C/T	rs958706	ALT
NM_001142800:c.-447-13214A>T	6:66362999	T/A	rs11758603	REF
NM_001142800:c.-447-15087dupT	6:66364872	-/A	rs36088825	REF
NM_001142800:c.-447-15820C>G	6:66365605	G/C	rs1844528	ALT
NM_001142800:c.-447-16373C>A	6:66366158	G/T	rs12195278	ALT
NM_001142800:c.-447-16546C>T	6:66366331	G/A	rs1602235	ALT
NM_001142800:c.-447-16662T>G	6:66366447	A/C	rs1602234	ALT
NM_001142800:c.-447-17970A>C	6:66367755	T/G	rs2200941	ALT
NM_001142800:c.-447-19519A>G	6:66369304	T/C	rs9345666	ALT
NM_001142800:c.-447-20781G>A	6:66370566	C/T	rs1387807	ALT
NM_001142800:c.-447-20994A>G	6:66370779	T/C	rs2171902	ALT
NM_001142800:c.-447-24071T>G	6:66373856	A/C	rs9345667	ALT
NM_001142800:c.-447-24072G>A	6:66373857	C/T	rs9363416	ALT
NM_001142800:c.-447-25019G>A	6:66374804	C/T	rs923537	ALT
NM_001142800:c.-447-25759A>C	6:66375544	T/G	rs7761627	ALT
NM_001142800:c.-447-25886T>A	6:66375671	A/T	rs7757773	ALT
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NM_001142800:c.-447-26179delA	6:66375964	T/-		REF
NM_001142800:c.-447-26209C>T	6:66375994	G/A	rs7741090	ALT
NM_001142800:c.-447-26281C>T	6:66376066	G/A	rs9354272	ALT
NM_001142800:c.-447-27066delT	6:66376851	A/-		REF
NM_001142800:c.-447-28089A>T	6:66377874	T/A	rs923534	ALT
NM_001142800:c.-447-28154A>G	6:66377939	T/C	rs923535	ALT
NM_001142800:c.-447-29854G>T	6:66379639	C/A	rs10455190	ALT
NM_001142800:c.-447-30126A>G	6:66379911	T/C	rs9283820	ALT
NM_001142800:c.-447-31642T>A	6:66381427	A/T	rs993015	ALT
NM_001142800:c.-447-32008A>G	6:66381793	T/C	rs1490120	ALT
NM_001142800:c.-447-32381T>C	6:66382166	A/G	rs7739376	ALT
NM_001142800:c.-447-33215C>T	6:66383000	G/A	rs12197906	ALT
NM_001142800:c.-447-33552T>C	6:66383337	A/G	rs6907252	ALT
NM_001142800:c.-448+33470G>A	6:66383558	C/T	rs6928284	ALT
NM_001142800:c.-448+32697T>C	6:66384331	A/G	rs10455585	ALT
NM_001142800:c.-448+30776T>G	6:66386252	A/C	rs7761171	ALT
NM_001142800:c.-448+30007C>T	6:66387021	G/A	rs200465206	REF
NM_001142800:c.-448+29090C>A	6:66387938	G/T	rs1907009	ALT
NM_001142800:c.-448+28329A>C	6:66388699	T/G	rs1490121	ALT

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NM_001142800:c.-448+28318dupT	6:66388710	-/A	rs34345454	0/1
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NM_001142800:c.-448+26177G>A	6:66390851	C/T	rs2130491	ALT
NM_001142800:c.-448+25747C>T	6:66391281	G/A	rs2087538	ALT
NM_001142800:c.-448+24395C>G	6:66392633	G/C	rs1158424	ALT
NM_001142800:c.-448+23745A>T	6:66393283	T/A	rs117516822	REF
NM_001142800:c.-448+22870A>C	6:66394158	T/G	rs10944833	ALT
NM_001142800:c.-448+22430G>T	6:66394598	C/A	rs1490124	ALT
NM_001142800:c.-448+22266C>T	6:66394762	G/A	rs2130492	ALT
NM_001142800:c.-448+21737T>A	6:66395291	A/T	rs6904807	ALT
NM_001142800:c.-448+21499C>T	6:66395529	G/A	rs6924750	ALT
NM_001142800:c.-448+21324C>T	6:66395704	G/A	rs6924950	ALT
NM_001142800:c.-448+20795A>T	6:66396233	T/A	rs12208064	ALT
NM_001142800:c.-448+20646G>A	6:66396382	C/T	rs9354275	ALT
NM_001142800:c.-448+20345G>A	6:66396683	C/T	rs2351500	ALT
NM_001142800:c.-448+20048C>A	6:66396980	G/T	rs2351501	ALT
NM_001142800:c.-448+19541T>C	6:66397487	A/G	rs6455052	ALT
NM_001142800:c.-448+19517T>G	6:66397511	A/C	rs7754201	ALT
NM_001142800:c.-448+19337C>T	6:66397691	G/A		REF
NM_001142800:c.-448+19336G>C	6:66397692	C/G		REF
NM_001142800:c.-448+17964A>T	6:66399064	T/A		ALT
NM_001142800:c.-448+17183T>C	6:66399845	A/G	rs6902244	ALT
NM_001142800:c.-448+17131G>C	6:66399897	C/G	rs6923019	ALT
NM_001142800:c.-448+16406A>G	6:66400622	T/C	rs72648383	ALT
NM_001142800:c.-448+16232T>C	6:66400796	A/G	rs112562414	ALT
NM_001142800:c.-448+15354A>C	6:66401674	T/G	rs9445566	REF
NM_001142800:c.-448+14500T>C	6:66402528	A/G	rs35543758	ALT
NM_001142800:c.-448+13730delA	6:66403298	T/-	rs58238248	0/1
NM_001142800:c.-448+13191T>C	6:66403837	A/G	rs12211518	ALT
NM_001142800:c.-448+12992G>A	6:66404036	C/T	rs12204750	ALT
NM_001142800:c.-448+12846T>C	6:66404182	A/G	rs12529251	ALT
NM_001142800:c.-448+12733C>A	6:66404295	G/T	rs375062337	REF
NM_001142800:c.-448+12392dupT	6:66404636	-/A	rs34494006	0/1
NM_001142800:c.-448+12041C>T	6:66404987	G/A	rs12192822	ALT
NM_001142800:c.-448+11499delA	6:66405529	T/-	rs71815460	ALT
NM_001142800:c.-448+10943T>C	6:66406085	A/G	rs72648385	ALT
NM_001142800:c.-448+10270C>T	6:66406758	G/A	rs6916913	ALT
NM_001142800:c.-448+9020A>G	6:66408008	T/C	rs2812772	ALT
NM_001142800:c.-448+8980T>C	6:66408048	A/G	rs2812773	ALT
NM_001142800:c.-448+8824A>G	6:66408204	T/C	rs2636123	ALT
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NM_001142800:c.-448+8058G>A	6:66408970	C/T	rs1873293	ALT
NM_001142800:c.-448+7273G>A	6:66409755	C/T	rs2812775	ALT
NM_001142800:c.-448+7244T>C	6:66409784	A/G	rs2812776	ALT
NM_001142800:c.-448+7239A>G	6:66409789	T/C	rs2814115	ALT

Table S6

NM_001142800:c.-448+6942C>G	6:66410086	G/C	rs2812777	ALT
NM_001142800:c.-448+6328G>T	6:66410700	C/A	rs2812778	ALT
NM_001142800:c.-448+5697A>G	6:66411331	T/C	rs1490130	ALT
NM_001142800:c.-448+4330C>T	6:66412698	G/A	rs1387801	ALT
NM_001142800:c.-448+3949A>G	6:66413079	T/C	rs2814114	ALT
NM_001142800:c.-448+1760T>G	6:66415268	A/C	rs1021350	ALT
NM_001142800:c.-448+1338C>A	6:66415690	G/T	rs997075	ALT

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HGVS c. (Clinically Relevant)	HGVS p. (Clinically Relevant)	Chr:Pos	Ref/Alt	Identifier	Haplotype
NM_006269:c.-13+191A>G		8:55528953	A/G	rs702761	ALT
NM_006269:c.-13+311C>T		8:55529073	C/T	rs9643828	ALT
NM_006269:c.-13+725T>A		8:55529487	T/A	rs6473949	ALT
NM_006269:c.-13+850A>G		8:55529612	A/G	rs145290	ALT
NM_006269:c.-12-1998G>A		8:55531517	G/A	rs436527	ALT
NM_006269:c.788-202G>A		8:55537028	G/A		REF
NM_006269:c.788-193C>A		8:55537037	C/A		REF
NM_006269:c.788-92T>C		8:55537138	T/C	rs429668	ALT
NM_006269:c.2615G>A	p.Arg872His	8:55539057	G/A	rs444772	ALT
NM_006269:c.2690_2695delCTTTG A	p.Ser897Ter	8:55539132	CTTTGA/-		ALT
NM_006269:c.2953A>T	p.Asn985Tyr	8:55539395	A/T	rs2293869	REF
NM_006269:c.5008G>A	p.Ala1670Thr	8:55541450	G/A	rs446227	ALT
NM_006269:c.5071T>C	p.Ser1691Pro	8:55541513	T/C	rs414352	ALT
NM_006269:c.5175A>G	p.Gln1725=	8:55541617	A/G	rs441800	ALT
NM_006269:c.6098G>A	p.Cys2033Tyr	8:55542540	G/A	rs61739567	REF
NM_006269:c.*247_*248insTT		8:55543161	-/TT	rs10654889	REF