Enlis Genomics

Report Date: **August 19, 2015**
Software Annotation Version: **8**

Phenotype Report

Name: **NA12144 NW European**
Genome ID: **NA12144_S1**

Sequencing Provider: **Illumina**
Sequencing Type: **Exome**

Phenotype: **Retinitis Pigmentosa**

Description:
Retinitis pigmentosa (RP) is a group of inherited disorders in which abnormalities of the photoreceptors (rods and cones) or the retinal pigment epithelium (RPE) of the retina lead to progressive visual loss. Affected individuals first experience defective dark adaptation or "night blindness," followed by constriction of peripheral visual fields and, eventually, loss of central vision late in the course of the disease.

Modes of inheritance*: Autosomal Dominant
Autosomal Recessive
Mitochondrial
X-linked Inheritance
X-linked Recessive

Variation Summary:

Variant Classification	Num. Variants Found	Num. Positions Not Called (Missing data)	Num. Positions Matching Reference	Num. Variants Assessed
Pathogenic	0	7 (2.5%)	274 (97.5%)	281
Likely pathogenic	0	0 (0.0%)	61 (100%)	61
Risk factor	1	0 (0.0%)	0 (0.0%)	1

Total number of phenotype variations assessed: **343**

Variation Details:

- **Chr16: 53,720,436 C>T** — Risk factor ★☆☆☆☆†

Zygosity: Heterozygous dbSNP ID: rs61747071
Population Allele Frequency: 3.75%
Gene Impact: **RPGRIP1L** MISSENSE A-229-T
Supporting Publications: http://www.ncbi.nlm.nih.gov/pubmed/19430481

Limitations:

There are several limitations to the results presented here:

• 7 variation(s) known to be related to this phenotype were not called in the sequencing data. This data is missing and would likely require additional sequencing to determine.

• The quality of the sequencing data has not been verified by Enlis. It is possible that variations listed here do not actually exist in the listed genome, and it is possible that positions listed as matching reference are actually variant.

• Knowledge of the genomic factors that relate to this phenotype are incomplete. New variations and genes that relate to this phenotype may be discovered in the future. Variations and genes currently thought to be related to this phenotype may be found to be incorrect. Care should especially be taken in the interpretation of variations with confidence star levels of only 1 or 2.

• The Enlis Genome Research application, and all associated communication (Enlis Products) are for research and educational purposes only. Enlis Products together and separately do not constitute medical advice and should not be used as a source of professional medical advice or diagnosis.

Endnotes

* Mode of inheritance defintions:

Autosomal Dominant
Autosomal dominant inheritance refers to genetic conditions that occur when a mutation is present in one copy of a given gene (i.e., the person is heterozygous).

Autosomal Recessive
Autosomal recessive inheritance refers to genetic conditions that occur only when mutations are present in both copies of a given gene (i.e., the person is homozygous for a mutation, or carries two different mutations of the same gene, a state referred to as compound heterozygosity).

Mitochondrial
Describes a disorder in which a mutation in one of the mitochondrial genes is necessary to express the phenotype and that is always maternally inherited.

X-linked Inheritance
Describes a disorder with mutated alleles located on the X chromosome.

X-linked Recessive
X-linked recessive inheritance refers to genetic conditions associated with mutations in genes on the X chromosome. A male carrying such a mutation will be affected, because he carries only one X chromosome. A female carrying a mutation in one gene, with a normal gene on the other X chromosome, will be unaffected.

† Confidence Star Levels:


Review Status: Classified by single submitter with no evidence provided, or multiple conficting interpretations


Review Status: Classified by single submitter with evidence


Review Status: Classified by multiple submitters


Review Status: Reviewed by expert panel


Review Status: Reviewed by professional society

Human genome reference version: HomoSapiens_GRCh37

Appendix - Full Position List:

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr1: 26,764,719 >G	rs147394623	DHDDS	Pathogenic	Retinitis pigmentosa 59			Reference
Chr1: 68,895,518 >A	rs121917745	RPE65	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 68,896,843 >C	rs62637004	RPE65	Pathogenic	Retinitis pigmentosa 20			Reference
Chr1: 68,903,896 >G	rs62653011	RPE65	Pathogenic	Retinitis pigmentosa 20			Reference
Chr1: 68,903,911 >T	rs121917744	RPE65	Pathogenic	Retinitis pigmentosa 20			Reference
Chr1: 68,903,976 >G	rs61752909	RPE65	Pathogenic	Retinitis pigmentosa 20			Reference
Chr1: 68,910,315 >T	rs61752878	RPE65	Pathogenic	Retinitis pigmentosa 20			Reference
Chr1: 68,910,541 >A	rs61752871	RPE65	Pathogenic	Retinitis pigmentosa 20			Reference
Chr1: 68,912,520 >T	rs61751281	RPE65	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 94,528,222 DEL	rs387906386	ABCA4	Pathogenic	Retinitis pigmentosa 19			Reference
Chr1: 94,544,895 >A	rs61748550	ABCA4	Pathogenic	Retinitis pigmentosa 19			Reference
Chr1: 150,316,677 >A	rs121434243	PRPF3	Pathogenic	Retinitis pigmentosa 18			Reference
Chr1: 150,316,688 >T	rs121434242	PRPF3	Pathogenic	Retinitis pigmentosa 18			Reference
Chr1: 150,316,692 >T	rs121434241	PRPF3	Pathogenic	Retinitis pigmentosa 18			Reference
Chr1: 156,132,784 >C	rs267607033	SEMA4A	Pathogenic	Retinitis pigmentosa 35			Reference
Chr1: 156,132,800 >G	rs267607034	SEMA4A	Pathogenic	Retinitis pigmentosa 35			Reference
Chr1: 156,146,640 >A	rs41265017	SEMA4A	Pathogenic	Retinitis pigmentosa 35			Reference
Chr1: 197,390,534 >T	rs114342808	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr1: 197,396,689 >T	rs28939720	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,396,745 >T	rs62635654	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,396,856 >T	rs137853137	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,403,836 >A	rs62645748	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,403,976 >T	rs62635655	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,404,115 >C	rs62635656	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,404,300 >A	rs62636275	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,404,376 DEL		CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,404,534 >C	rs62636291	CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 197,446,905-197,446,914 DEL		CRB1	Pathogenic	Retinitis pigmentosa 12			Reference
Chr1: 211,844,558-211,844,565 >T	rs398122961	NEK2	Pathogenic	Retinitis pigmentosa 67			Reference
Chr1: 213,032,155 >G	rs267606820	FLVCR1	Pathogenic	Posterior column ataxia with retinitis pigmentosa			Reference
Chr1: 213,032,368 >C	rs267606821	FLVCR1	Pathogenic	Posterior column ataxia with retinitis pigmentosa			Reference
Chr1: 213,032,515 >A	rs267606819	FLVCR1	Pathogenic	Posterior column ataxia with retinitis pigmentosa			Reference
Chr1: 215,807,865 >C		USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,814,065 >A	rs146733615	USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 215,822,002 >T		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,823,990 >T	rs397517990	USH2A	Likely pathogenic	Retinitis pigmentosa 39			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr1: 215,824,034 >A		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,844,427 >C	rs80338904	USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,844,600 >A		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,847,787 >T		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,848,678 >T	rs199605265	USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 215,853,720 >C	rs397517978	USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 215,901,562-215,901,563 DEL		USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 215,956,121 >C		USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,963,510 >T	rs148660051	USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 215,972,456 >G		USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 215,990,485 >A	rs397518048	USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 216,040,512 DEL		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,040,521 >C	rs372347027	USH2A	Likely pathogenic	Retinitis pigmentosa 39			Reference
Chr1: 216,051,224 >C	rs397518039	USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,173,831 >T		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,419,934 >C	rs201527662	USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,420,437 DEL	rs80338903	USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,420,460 >A	rs80338902	USH2A	Pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,420,527 >A	rs111033334	USH2A	Pathogenic	Retinitis pigmentosa 39			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr1: 216,465,634 INS TC		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,592,017 >A		USH2A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr1: 216,595,438 INS GATC		USH2A	Pathogenic	Retinitis pigmentosa 39			Reference
Chr2: 27,601,023 >G	rs267607182	ZNF513	Pathogenic	Retinitis pigmentosa 58			Reference
Chr2: 27,668,796 >C		KRTCAP3, IFT172	Pathogenic	Retinitis pigmentosa 71			Reference
Chr2: 27,669,181 >T		KRTCAP3, IFT172	Pathogenic	Retinitis pigmentosa 71			Reference
Chr2: 27,680,627 >T		IFT172	Pathogenic	Retinitis pigmentosa 71			Reference
Chr2: 27,693,963 >T	rs370540673	IFT172	Pathogenic	Retinitis pigmentosa 71			Reference
Chr2: 27,703,928 >G		IFT172	Pathogenic	Retinitis pigmentosa 71			Reference
Chr2: 29,294,140 INS G		C2ORF71	Likely pathogenic	Retinitis pigmentosa			Reference
Chr2: 29,295,002 DEL		C2ORF71	Likely pathogenic	Retinitis pigmentosa			Reference
Chr2: 29,296,527 >A	rs267606690	C2ORF71	Pathogenic	Retinitis pigmentosa 54			Reference
Chr2: 29,296,572 >A	rs267606691	C2ORF71	Pathogenic	Retinitis pigmentosa 54			Reference
Chr2: 62,063,210 >A	rs267606793	FAM161A	Pathogenic	Retinitis pigmentosa 28			Reference
Chr2: 62,066,572 >A	rs202193201	FAM161A	Pathogenic	Retinitis pigmentosa 28			Reference
Chr2: 62,066,783-62,066,784 DEL	rs397704718	FAM161A	Pathogenic	Retinitis pigmentosa 28			Reference
Chr2: 62,066,830 >A	rs200691042	FAM161A	Pathogenic	Retinitis pigmentosa 28			Reference
Chr2: 62,067,454 >A	rs267606794	FAM161A	Pathogenic	Retinitis pigmentosa 28			Reference
Chr2: 96,953,697 >A	rs397514574	SNRNP200	Pathogenic	Retinitis pigmentosa 33			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr2: 96,953,706 >A	rs267607077	SNRNP200	Pathogenic	Retinitis pigmentosa 33			Reference
Chr2: 96,956,153 >C	rs397514575	SNRNP200	Pathogenic	Retinitis pigmentosa 33			Reference
Chr2: 96,958,823 >A		SNRNP200	Pathogenic	Retinitis pigmentosa			Reference
Chr2: 96,958,828 >T		SNRNP200	Pathogenic	Retinitis pigmentosa			Reference
Chr2: 96,959,219 >T		SNRNP200	Likely pathogenic	Retinitis pigmentosa			Reference
Chr2: 112,686,860 DEL		MERTK	Likely pathogenic	Retinitis pigmentosa			Reference
Chr2: 112,687,005 >T		MERTK	Likely pathogenic	Retinitis pigmentosa 38			Reference
Chr2: 112,751,981 >A		MERTK	Likely pathogenic	Retinitis pigmentosa			Reference
Chr2: 112,758,776 >G		MERTK	Pathogenic	Retinitis pigmentosa 38			Reference
Chr2: 112,766,043 >T	rs119489105	MERTK	Pathogenic	Retinitis pigmentosa 38			Reference
Chr2: 112,777,100 >T	rs371956016	MERTK	Pathogenic	Retinitis pigmentosa 38			Reference
Chr2: 112,779,132 >T	rs387907314	MERTK	Pathogenic	Retinitis pigmentosa 38			Reference
Chr2: 182,423,333 DEL	rs398122964	CERKL	Pathogenic	Retinitis pigmentosa 26			Reference
Chr2: 182,423,344 >A	rs121909398	CERKL	Pathogenic	Retinitis pigmentosa 26			Reference
Chr2: 182,438,495 >A	rs398122963	CERKL	Pathogenic	Retinitis pigmentosa 26			Reference
Chr2: 182,468,625 DEL	rs398122962	CERKL	Pathogenic	Retinitis pigmentosa 26			Reference
Chr2: 234,243,725 DEL		SAG	Pathogenic	Retinitis pigmentosa 47			Reference
Chr3: 97,503,810 >T		ARL6	Pathogenic	Retinitis pigmentosa 55			Reference
Chr3: 100,949,961 >A	rs199867882	IMPG2	Pathogenic	Retinitis pigmentosa			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr3: 100,961,664 >A	rs267606875	IMPG2	Pathogenic	Retinitis pigmentosa 56			Reference
Chr3: 100,962,459 >A	rs267606876	IMPG2	Pathogenic	Retinitis pigmentosa 56			Reference
Chr3: 100,994,538 >C	rs267606874	IMPG2	Pathogenic	Retinitis pigmentosa 56			Reference
Chr3: 129,247,620 >G	rs104893786	RHO	Pathogenic	Retinitis pigmentosa			Reference
Chr3: 129,247,626 >T	rs104893769	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,643 >G	rs104893797	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,644 >A	rs104893768	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,709 >C	rs104893770	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,727 >C	rs104893792	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,734 >G	rs28933395	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,749 >G	rs28933394	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,756 >A		RHO	Likely pathogenic	Retinitis pigmentosa			Reference
Chr3: 129,247,836 >A	rs104893771	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,842 >A	rs104893772	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,892 >A	rs104893773	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,892 >T	rs104893773	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,905 >A	rs104893787	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,247,917 >A	rs104893788	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,249,760 >T	rs104893775	RHO	Pathogenic	Retinitis pigmentosa			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr3: 129,249,761 >T	rs104893774	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,249,805 >A	rs104893791	RHO	Pathogenic	Retinitis pigmentosa 4, autosomal recessive			Reference
Chr3: 129,249,848 >A	rs104893793	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,249,868 >T	rs104893794	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,249,877 >A		RHO	Pathogenic	Retinitis pigmentosa			Reference
Chr3: 129,251,096 >G	rs104893776	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,104 >A		RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,107 >A	rs104893780	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,125 >A		RHO	Pathogenic	Retinitis pigmentosa			Reference
Chr3: 129,251,131 >A	rs104893779	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,131 >T	rs104893779	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,132 >G	rs104893777	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,183 >G	rs104893782	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,195 >C	rs28933993	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,424 >T	rs104893783	RHO	Pathogenic	Retinitis pigmentosa 4, autosomal recessive			Reference
Chr3: 129,251,468-129,251,470 DEL		RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,479 >T	rs104893781	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,251,565 >G	rs29001653	RHO	Pathogenic	Retinitis pigmentosa 4			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr3: 129,252,491-129,252,494 DEL		RHO	Likely pathogenic	Retinitis pigmentosa			Reference
Chr3: 129,252,544 >T	rs104893778	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,252,547 >A	rs104893795	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,252,547 >C	rs104893795	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,252,553 >T	rs29001637	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,252,554 >T	rs29001566	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,252,554 >A	rs29001566	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 129,252,554 >G	rs29001566	RHO	Pathogenic	Retinitis pigmentosa 4			Reference
Chr3: 150,690,404 >A		CLRN1-AS1,RP11-166N6.2, CLRN1	Pathogenic	Retinitis pigmentosa 61			Reference
Chr3: 170,185,037 >C		CLDN11, SLC7A14	Pathogenic	Retinitis pigmentosa 68			Reference
Chr3: 170,198,680 >A	rs79668755	CLDN11, SLC7A14	Pathogenic	Retinitis pigmentosa 68			Reference
Chr3: 170,201,230 >T	rs2276717	CLDN11, SLC7A14	Pathogenic	Retinitis pigmentosa 68			Reference
Chr3: 170,219,044 >A		CLDN11, SLC7A14	Pathogenic	Retinitis pigmentosa 68			Reference
Chr4: 619,584 INS ACGGCGC...		PDE6B	Pathogenic	Retinitis pigmentosa 40			Reference
Chr4: 647,908 >T	rs121918579	PDE6B, RP11-1191J2.2	Pathogenic	Retinitis pigmentosa 40			Reference
Chr4: 649,728 >C		PDE6B	Likely pathogenic	Retinitis pigmentosa			Reference
Chr4: 652,807 >C		PDE6B	Pathogenic	Retinitis pigmentosa			Reference
Chr4: 654,274 DEL		PDE6B	Pathogenic	Retinitis pigmentosa 40			Reference
Chr4: 654,328 DEL	rs398123298	PDE6B	Pathogenic	Retinitis pigmentosa 40			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr4: 654,364 >A		PDE6B	Likely pathogenic	Retinitis pigmentosa			Reference
Chr4: 654,379 >T	rs121918580	PDE6B	Pathogenic	Retinitis pigmentosa 40			Reference
Chr4: 654,392 >A		PDE6B	Pathogenic	Retinitis pigmentosa			Reference
Chr4: 655,977 >T	rs121918581	PDE6B	Pathogenic	Retinitis pigmentosa			Reference
Chr4: 658,734 >A		PDE6B	Pathogenic	Retinitis pigmentosa 40			Reference
Chr4: 661,711 >A	rs121918583	PDE6B	Pathogenic	Retinitis pigmentosa 40			Reference
Chr4: 15,995,651 >A	rs137853005	PROM1	Pathogenic	Retinitis pigmentosa 41			Reference
Chr4: 16,002,140 >T	rs137853907	PROM1	Pathogenic	Retinitis pigmentosa 41			Reference
Chr4: 47,938,878 >T		CNGA1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr4: 47,939,522 >C		CNGA1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr4: 47,939,552 >A	rs62625014	CNGA1	Pathogenic	Retinitis pigmentosa 49			Reference
Chr4: 47,939,672 >T	rs375412499	CNGA1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr4: 47,945,220 >A	rs121909600	CNGA1	Pathogenic	Retinitis pigmentosa 49		Missing Data	
Chr4: 47,951,911 >A	rs121909599	CNGA1	Pathogenic	Retinitis pigmentosa 49			Reference
Chr4: 47,953,415 DEL		CNGA1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr4: 155,665,641 >T		LRAT	Likely pathogenic	Retinitis pigmentosa			Reference
Chr5: 149,245,756-149,245,759 DEL		PDE6A	Pathogenic	Retinitis pigmentosa 43			Reference
Chr5: 149,263,074 >T	rs121909835	PDE6A	Pathogenic	Retinitis pigmentosa 43			Reference
Chr5: 149,264,342 >T		PDE6A	Pathogenic	Retinitis pigmentosa 43			Reference
Chr5: 149,265,917 >C	rs121918576	PDE6A	Pathogenic	Retinitis pigmentosa 43			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr5: 149,274,791 >T	rs121918578	PDE6A	Pathogenic	Retinitis pigmentosa 43			Reference
Chr5: 149,286,908 >T	rs121918577	PDE6A	Pathogenic	Retinitis pigmentosa 43			Reference
Chr6: 10,804,063 >T		RP11-637O19.3, SYCP2L, MAK	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 10,804,119 >T	rs387906648	RP11-637O19.3, SYCP2L, MAK	Pathogenic	Retinitis pigmentosa 62			Reference
Chr6: 10,804,120 >A		RP11-637O19.3, SYCP2L, MAK	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 10,809,146 >G	rs387906646	RP11-637O19.3, SYCP2L, MAK	Pathogenic	Retinitis pigmentosa 62			Reference
Chr6: 10,813,895 INS C		RP11-637O19.3, SYCP2L, MAK	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 10,830,845 >T	rs387906647	RP11-637O19.3, SYCP2L, MAK	Pathogenic	Retinitis pigmentosa 62			Reference
Chr6: 35,467,782 >G	rs121909074	TULP1	Pathogenic	Retinitis pigmentosa 14			Reference
Chr6: 35,467,809 >A	rs121909077	TULP1	Pathogenic	Retinitis pigmentosa 14			Reference
Chr6: 35,467,877 >T	rs121909075	TULP1	Pathogenic	Retinitis pigmentosa 14			Reference
Chr6: 35,471,400 >G	rs121909073	TULP1	Pathogenic	Retinitis pigmentosa 14			Reference
Chr6: 35,471,593 >G	rs121909076	TULP1	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 35,479,425 >T		TULP1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 42,153,424 >T	rs121909124	GUCA1B	Pathogenic	Retinitis pigmentosa 48			Reference
Chr6: 42,672,100 >A	rs281865373	PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr6: 42,672,195 >G	rs61755817	PRPH2	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 42,672,199 >T	rs61755816	PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 42,672,273-42,672,275 DEL	rs61755807	PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 42,672,284 >A	rs61755806	PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 42,689,519 >G	rs121918563	PRPH2	Pathogenic	Retinitis pigmentosa 7, digenic			Reference
Chr6: 42,689,555 >A	rs61755794	PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 42,689,574 >T		PRPH2	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 42,689,610-42,689,612 DEL		PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 42,689,663 >T		PRPH2	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 42,689,715-42,689,717 DEL		PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 42,689,937 >A	rs61755771	PRPH2	Pathogenic	Retinitis pigmentosa 7			Reference
Chr6: 64,430,522 >T	rs137853190	EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 64,430,718 >G	rs183589498	EYS	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,431,122 >T		EYS	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,431,518-64,431,519 >AT		EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 64,431,547 INS TGCA		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,472,413 >T		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,472,506 >T		EYS	Pathogenic	Retinitis pigmentosa			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr6: 64,488,004 >T		EYS	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,498,027 DEL		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,574,212 >C	rs398123575	EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 64,694,283 DEL		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,791,763 >T		EYS	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 64,940,493 >T		EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 65,146,137 >A	rs137853189	EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 65,300,557-65,300,558 DEL		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 65,300,746 >A		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 65,300,803 INS T		EYS	Pathogenic	Retinitis pigmentosa			Reference
Chr6: 65,301,357 INS CCTCTTGA		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 65,301,373 DEL		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 65,301,404-65,301,410 DEL		EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 65,523,270 >A	rs373441420	EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 65,707,540 >A		EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 65,767,589 >T	rs372354156	EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr6: 66,044,889 >A		EYS	Likely pathogenic	Retinitis pigmentosa			Reference
Chr6: 66,063,465 >A		EYS	Likely pathogenic	Retinitis pigmentosa			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr6: 66,204,814 >A		EYS	Pathogenic	Retinitis pigmentosa 25			Reference
Chr7: 23,180,394 >A	rs137853112	KLHL7	Pathogenic	Retinitis pigmentosa 42			Reference
Chr7: 23,180,402 >A	rs137853114	KLHL7	Pathogenic	Retinitis pigmentosa 42			Reference
Chr7: 23,180,403 >T	rs137853113	KLHL7	Pathogenic	Retinitis pigmentosa 42			Reference
Chr7: 33,135,003 >C	rs104894039	RP9	Pathogenic	Retinitis pigmentosa 9			Reference
Chr7: 128,038,485 >T	rs121912551	IMPDH1	Pathogenic	Retinitis pigmentosa 10			Reference
Chr7: 128,038,611 >T	rs121912550	IMPDH1	Pathogenic	Retinitis pigmentosa 10			Reference
Chr7: 128,038,616 >G	rs121912552	IMPDH1	Pathogenic	Retinitis pigmentosa 10			Reference
Chr8: 10,469,636 >A		RP1L1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr8: 10,480,477 >A	rs377269054	RP1L1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr8: 55,534,710 DEL		RP1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr8: 55,537,560 >T	rs77775126	RP1	Pathogenic	Retinitis pigmentosa 1			Reference
Chr8: 55,537,628 >T	rs201493928	RP1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr8: 55,538,471 >T	rs104894082	RP1	Pathogenic	Retinitis pigmentosa 1			Reference
Chr8: 55,538,477 >T	rs104894083	RP1	Pathogenic	Retinitis pigmentosa 1			Reference
Chr8: 55,539,142-55,539,143 >AC		RP1	Pathogenic	Retinitis pigmentosa 1			Reference
Chr8: 55,540,992 DEL		RP1	Pathogenic	Retinitis pigmentosa 1			Reference
Chr8: 55,541,318 >A		RP1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr8: 55,541,461 >G	rs398124220	RP1	Pathogenic	Retinitis pigmentosa 1			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr8: 55,542,239 >T	rs118031911	RP1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr8: 63,985,549 >C	rs121917849	TTPA	Pathogenic	Ataxia and retinitis pigmentosa with isolated vitamin e deficiency			Reference
Chr8: 96,259,924 >C	rs387907137	C8ORF37-AS1, C8ORF37	Pathogenic	Retinitis pigmentosa 64			Reference
Chr9: 32,541,966-32,541,969 DEL		TOPORS	Pathogenic	Retinitis pigmentosa			Reference
Chr9: 116,037,910-116,037,927 DEL	rs541873609	PRPF4	Pathogenic	Retinitis pigmentosa 70			Reference
Chr9: 116,050,463 >T		PRPF4	Pathogenic	Retinitis pigmentosa 70			Reference
Chr10: 48,385,854 >T	rs146150511	RBP3	Pathogenic	Retinitis pigmentosa 66			Reference
Chr10: 86,007,463 >C	rs104894187	RGR	Pathogenic	Retinitis pigmentosa 44			Reference
Chr11: 61,723,360 >G	rs267606678	BEST1	Pathogenic	Retinitis pigmentosa 50			Reference
Chr11: 61,724,448 >C	rs267606680	BEST1	Pathogenic	Retinitis pigmentosa 50			Reference
Chr11: 61,724,902 >G	rs267606677	BEST1	Pathogenic	Retinitis pigmentosa, concentric			Reference
Chr11: 61,724,904 >A	rs267606676	BEST1	Pathogenic	Retinitis pigmentosa 50			Reference
Chr11: 61,725,666 >T	rs372989281	BEST1	Pathogenic	Retinitis pigmentosa			Reference
Chr11: 62,381,084 INS G	rs71458427	ROM1	Pathogenic	Retinitis pigmentosa			Reference
Chr11: 76,874,011 >T		MYO7A	Likely pathogenic	Retinitis pigmentosa			Reference
Chr14: 24,551,771 >G	rs397514516	NRL	Pathogenic	Retinitis pigmentosa 27			Reference
Chr14: 24,551,907 >A		NRL	Pathogenic	Retinitis pigmentosa 27			Reference
Chr14: 24,551,910 >T	rs104894459	NRL	Pathogenic	Retinitis pigmentosa 27			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr14: 24,552,035 DEL		NRL	Likely pathogenic	Retinitis pigmentosa			Reference
Chr14: 68,192,801 >T	rs202126574	RDH12	Pathogenic	Retinitis pigmentosa			Reference
Chr14: 68,196,025 DEL		RDH12	Pathogenic	Retinitis pigmentosa			Reference
Chr14: 89,300,035 >G		TTC8	Pathogenic	Retinitis pigmentosa 51			Reference
Chr15: 72,103,847-72,103,848 >AGTGTGC...		NR2E3	Pathogenic	Retinitis pigmentosa 37			Reference
Chr15: 72,103,870 >A	rs121912631	NR2E3	Pathogenic	Retinitis pigmentosa 37			Reference
Chr15: 72,104,309 >T		NR2E3	Likely pathogenic	Retinitis pigmentosa			Reference
Chr16: 53,720,436 >T	rs61747071	RPGRIP1L	Risk factor	Retinitis pigmentosa in ciliopathies, modifier of	Zygosity 1		
Chr16: 57,282,482 >G	rs398123053	ARL2BP, RP11-407G23.3	Pathogenic	Retinitis pigmentosa without situs inversus			Reference
Chr16: 57,931,817 >A	rs121918532	CNGB1	Pathogenic	Retinitis pigmentosa 45			Reference
Chr16: 57,938,715-57,938,716 >TG		CNGB1	Pathogenic	Retinitis pigmentosa 45			Reference
Chr16: 57,938,748 INS T		CNGB1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr16: 57,984,367 >A	rs372504780	CNGB1	Pathogenic	Retinitis pigmentosa 45			Reference
Chr16: 57,998,386 >G		CNGB1	Likely pathogenic	Retinitis pigmentosa			Reference
Chr17: 1,554,175 >T	rs121434238	PRPF8	Pathogenic	Retinitis pigmentosa 13			Reference
Chr17: 1,554,178 >G	rs121434236	PRPF8	Pathogenic	Retinitis pigmentosa 13			Reference
Chr17: 1,554,178 >C	rs121434236	PRPF8	Pathogenic	Retinitis pigmentosa 13			Reference
Chr17: 1,554,192 >C	rs121434240	PRPF8	Pathogenic	Retinitis pigmentosa 13			Reference
Chr17: 1,554,203 >T	rs121434239	PRPF8	Pathogenic	Retinitis pigmentosa 13			Reference
Chr17: 1,556,852 >A	rs387906971	PRPF8	Pathogenic	Retinitis pigmentosa 13			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr17: 6,328,871-6, 328,882 DEL	rs281865195	AIPL1	Pathogenic	Juvenile retinitis pigmentosa, AIPL1-related		Missing Data	
Chr17: 58,227,435 >T	rs104894559	CA4	Pathogenic	Retinitis pigmentosa 17			Reference
Chr17: 58,234,014 >A	rs121434552	CA4	Pathogenic	Retinitis pigmentosa 17			Reference
Chr17: 58,235,718 >A	rs121434551	CA4	Pathogenic	Retinitis pigmentosa 17			Reference
Chr17: 74,536,225 >C		CYGB, PRCD	Pathogenic	Retinitis pigmentosa			Reference
Chr17: 74,536,228 >A	rs121918369	CYGB, PRCD	Pathogenic	Retinitis pigmentosa 36			Reference
Chr17: 74,536,287 >T	rs387907268	CYGB, PRCD	Pathogenic	Retinitis pigmentosa 36			Reference
Chr17: 79,495,629 DEL	rs376633374	FSCN2	Pathogenic	Retinitis pigmentosa 30			Reference
Chr19: 48,339,592 >C		CRX, TPRX2P	Pathogenic	Retinitis pigmentosa			Reference
Chr19: 48,343,221 >C		CRX, TPRX2P	Likely pathogenic	Retinitis pigmentosa			Reference
Chr19: 54,626,942 >G		PRPF31, AC012314.8	Pathogenic	Retinitis pigmentosa 11			Reference
Chr19: 54,627,162 >T		PRPF31, AC012314.8	Likely pathogenic	Retinitis pigmentosa			Reference
Chr19: 54,627,181 >A	rs119475043	PRPF31, AC012314.8	Pathogenic	Retinitis pigmentosa 11			Reference
Chr19: 54,627,215 >G		PRPF31, AC012314.8	Likely pathogenic	Retinitis pigmentosa			Reference
Chr19: 54,627,246 >C	rs119475042	PRPF31, AC012314.8	Pathogenic	Retinitis pigmentosa 11			Reference
Chr19: 54,627,944 >T		PRPF31, AC012314.8	Likely pathogenic	Retinitis pigmentosa			Reference
Chr19: 54,631,576 >A		PRPF31	Pathogenic	Retinitis pigmentosa 11			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr19: 54,631,719-54,631,729 DEL		PRPF31	Pathogenic	Retinitis pigmentosa 11			Reference
Chr19: 54,632,558 >T		PRPF31	Pathogenic	Retinitis pigmentosa 11			Reference
Chr19: 54,633,399 >G		PRPF31	Pathogenic	Retinitis pigmentosa 11		Missing Data	
Chr20: 2,641,558 >G	rs137853020	IDH3B	Pathogenic	Retinitis pigmentosa 46			Reference
Chr20: 3,893,179 >C	rs28939088	PANK2	Pathogenic	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration			Reference
Chr20: 3,897,602 >T	rs137852968	PANK2	Pathogenic	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration			Reference
Chr20: 3,899,342 >A	rs137852959	PANK2	Pathogenic	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration			Reference
Chr20: 3,899,364 >T	rs137852967	PANK2	Pathogenic	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration			Reference
Chr20: 21,106,808 >T		KIZ	Pathogenic	Retinitis pigmentosa 69			Reference
Chr20: 21,112,767-21,112,770 DEL		KIZ	Pathogenic	Retinitis pigmentosa 69			Reference
Chr20: 21,117,104 >T	rs202210819	KIZ	Pathogenic	Retinitis pigmentosa 69			Reference
Chr20: 25,282,896 >C		ABHD12	Pathogenic	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract			Reference
Chr20: 25,282,958 >A	rs267606624	ABHD12	Pathogenic	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract			Reference
Chr20: 25,288,617 INS GCTCTTA		ABHD12	Pathogenic	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract			Reference
Chr20: 25,297,700 >G		ABHD12	Pathogenic	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
Chr20: 25,300,900 >T		ABHD12	Pathogenic	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract			Reference
Chr20: 25,304,045-25,304,046 >AAA	rs387906217	ABHD12	Pathogenic	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract			Reference
Chr20: 62,626,380 >C		ZNF512B, PRPF6	Likely pathogenic	Retinitis pigmentosa			Reference
Chr20: 62,658,491 >T	rs387907100	ZNF512B, PRPF6	Pathogenic	Retinitis pigmentosa 60			Reference
ChrX: 13,768,358 >G		OFD1	Pathogenic	Retinitis Pigmentosa 23		Missing Data	
ChrX: 38,145,602 >A	rs137852549	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15		Missing Data	
ChrX: 38,145,825-38,145,826 DEL		TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa		Missing Data	
ChrX: 38,145,846-38,145,847 DEL	rs398122960	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15		Missing Data	
ChrX: 38,146,271 >A		TM4SF2, RPGR	Likely pathogenic	Retinitis pigmentosa			Reference
ChrX: 38,158,366 INS CTAC		TM4SF2, RPGR	Likely pathogenic	Retinitis pigmentosa			Reference
ChrX: 38,163,900 >G		TM4SF2, RPGR	Likely pathogenic	Retinitis pigmentosa			Reference
ChrX: 38,163,927-38,163,928 DEL		TM4SF2, RPGR	Likely pathogenic	Retinitis pigmentosa			Reference
ChrX: 38,163,999 >T	rs62642057	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,164,016 >T	rs398123336	TM4SF2, RPGR	Likely pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,169,943 >A	rs62638651	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,169,990-38,170,004 >G		TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,176,671 >G	rs137852550	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa, x-linked, and sinorespiratory infections, with deafness			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
ChrX: 38,176,683 >A		TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,178,081 >T	rs62638646	TM4SF2, RPGR	Likely pathogenic	Retinitis pigmentosa			Reference
ChrX: 38,178,162 >C	rs62638644	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,180,294 >T	rs62638637	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,182,174 >A	rs62638634	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 38,182,200 >C	rs62638632	TM4SF2, RPGR	Pathogenic	Retinitis pigmentosa 15			Reference
ChrX: 46,696,550-46,696,552 DEL		RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrX: 46,696,611 >T	rs104894925	RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrX: 46,713,160 DEL		RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrX: 46,713,161 >A	rs28933687	RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrX: 46,713,161 >T	rs28933687	RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrX: 46,713,166 >T	rs104894927	RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrX: 46,713,261 >G	rs104894926	RP2	Pathogenic	Retinitis pigmentosa 2			Reference
ChrM: 8,618 INS T	rs199476139	MT-ATP6	Pathogenic	Neuropathy ataxia retinitis pigmentosa syndrome			Reference
ChrM: 8,993 >G		MT-ATP6	Pathogenic	Neuropathy ataxia retinitis pigmentosa syndrome			Reference
ChrM: 12,258 >A		MT-TS2	Pathogenic	Retinitis pigmentosa-deafness syndrome			Reference