Appendix A – Known RDH12 mutations (HGMD, July 2026)

Source: Human Gene Mutation Database (HGMD), July 2026; 211 entries. Variant class as assigned by HGMD: DM = disease-causing mutation; DM? = likely disease-causing (uncertain); R = retired entry. Companion to the RDH12 Landscape 2026 (3rd edition).

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.2T>C	p.M1?	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Astuti (2016) Eur J Hum Genet 24: 1071 [Leber congenital amaurosis] | Peter (2023) PNAS Nexus 2: pgad043 [Leber congenital amaurosis]	Chr14:67722644T/C
c.1A>G	p.M1?	DM	Leber congenital amaurosis	Suga (2022) Hum Mutat 43, 2251	Chr14:67722643A/G
c.38C>A	p.S13*	DM	Retinal degeneration	Fahim (2019) Br J Ophthalmol 103, 1789 | Yohe (2020) Mol Genet Genomic Med 8: e1081 [Rod cone dystrophy]	Chr14:67722680C/A
c.85G>A	p.G29R	DM?	Congenital heart disease	Dong (2025) Proc Natl Acad Sci U S A 122,	Chr14:67724489G/A
c.121G>T	p.V41L	DM?	Retinitis pigmentosa	Huang (2017) Sci Rep 7, 1948	Chr14:67724525G/T
c.125T>C	p.V42A	DM?	Leber congenital amaurosis	Sallum (2020) Am J Med Genet C Semin Med Genet 184, 728 | Sallum (2024) Am J Ophthalmol 265: 257 [Additional report]	Chr14:67724529T/C
c.133A>G	p.T45A	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Zolnikova (2017) Eur J Med Genet 60: 140 [Additional report] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Additional report]	Chr14:67724537A/G
c.137G>A	p.G46D	DM	Retinitis pigmentosa	Ozguc Caliskan (2024) Clin Genet 106, 258	Chr14:67724541G/A
c.139G>A	p.A47T	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Jin (2022) Brain Topogr : [Stargardt disease] | Patel (2016) Genet Med 18: 554 [Retinitis pigmentosa & rod cone dystrophy] | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa]	Chr14:67724543G/A
c.142A>G	p.N48D	DM?	Retinal disease	Lin (2024) Ophthalmol Retina 8, 699	Chr14:67724546A/G
c.146C>A	p.T49K	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Lin (2024) Ophthalmol Retina : [Retinal disease] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Han (2025) NPJ Genom Med 10: 6 [Leber congenital amaurosis]	Chr14:67724550C/A
c.146C>T	p.T49M	DM	Retinal dystrophy	Janecke (2004) Nat Genet 36, 850 | Wang (2018) Genes (Basel) 9: [Retinitis pigmentosa] | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Liu (2020) Clin Exp Ophthalmol 49: 46 [Retinitis pigmentosa]	Chr14:67724550C/T
c.148G>A	p.G50S	DM?	Retinitis pigmentosa	Gao (2019) Ophthalmology 126, 1549 | Li (2023) Invest Ophthalmol Vis Sci 64: [Retinal dystrophy] | Hitti-Malin (2024) Biomolecules 14: epub [Macular dystrophy] | Schlottmann (2023) NPJ Genom Med 8: 8 [Additional report]	Chr14:67724552G/A
c.152T>A	p.I51N	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639 | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinitis pigmentosa] | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinal dystrophy] | Lee (2010) FEBS Lett 584: 507 [Functional characterisation]	Chr14:67724556T/A
c.164C>A	p.T55K	DM	Leber congenital amaurosis	Li (2017) J Zhejiang Univ Sci B 18, 421 | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Xu (2020) Br J Ophthalmol 104: 932 [Leber congenital amaurosis / retinal dystrophy] | Zhu (2021) Ophthalmic Genet 42: 392 [Additional report]	Chr14:67724568C/A
c.164C>T	p.T55M	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Ehrenberg (2019) Mol Vis 25: 691 [Retinitis pigmentosa] | Griffith (2022) Genes (Basel) 13: epub [Retinitis pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa]	Chr14:67724568C/T
c.167C>A	p.A56D	DM?	Retinitis pigmentosa	Wang (2014) Hum Genet 133, 331 | Stone (2017) Ophthalmology 124: 1314 [Additional report]	Chr14:67724571C/A
c.176T>G	p.L59R	DM	Leber congenital amaurosis	Coppieters (2012) Genet Med 14, 576	Chr14:67724580T/G
c.179C>A	p.A60D	DM	Leber congenital amaurosis	Zhu (2021) Ophthalmic Genet 42, 392 | Matczyńska (2024) Biomedicines 12: 1355 [Additional report]	Chr14:67724583C/A
c.178G>C	p.A60P	DM	Cone dystrophy	Abu-Safieh (2013) Genome Res 23, 236 | Sallum (2020) Am J Med Genet C Semin Med Genet 184: 728 [Leber congenital amaurosis] | Sallum (2024) Am J Ophthalmol 265: 257 [Leber congenital amaurosis]	Chr14:67724582G/C

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.178G>A	p.A60T	DM	Cone-rod dystrophy	Zou (2018) Retina 39, 2040 | Jin (2022) Brain Topogr : [Stargardt disease] | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Kuo (2023) J Med Genet 60: 859 [Maculopathy, pseudocoloboma-like]	Chr14:67724582G/A
c.185G>A	p.R62Q	DM	Maculopathy	De Zaeytijd (2021) Retina 41, 1346	Chr14:67724589G/A
c.185G>T	p.R62L	DM	Retinal degeneration	Fahim (2019) Br J Ophthalmol 103, 1789	Chr14:67724589G/T
c.184C>T	p.R62*	DM	Retinal dystrophy	Janecke (2004) Nat Genet 36, 850 | Beheshtian (2015) Arch Iran Med 18: 776 [Retinitis pigmentosa, autosomal recessive] | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa]	Chr14:67724588C/T
c.188G>A	p.G63E	DM?	Macular dystrophy	Weisschuh (2024) J Med Genet 61, 186	Chr14:67725099G/A
c.194G>A	p.R65Q	DM?	Retinal disease	Karali (2022) Sci Rep 12, | Zampaglione (2022) Genet Med 24: 332 [Retinal degeneration] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67725105G/A
c.193C>T	p.R65*	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67725104C/T
c.209G>A	p.C70Y	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67725120G/A
c.215A>G	p.D72G	DM	Retinitis pigmentosa	Scott (2020) Cold Spring Harb Mol Case Stud 6, a004754	Chr14:67725126A/G
c.226G>A	p.G76R	DM	Leber congenital amaurosis	Wang (2015) Invest Ophthalmol Vis Sci 56, 3642 | Weisschuh (2024) J Med Genet 61: 186 [Retinitis pigmentosa, autosomal recessive] | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa]	Chr14:67725137G/A
c.226G>C	p.G76R	DM	Leber congenital amaurosis	Aldahmesh (2009) Mol Vis 15, 2464 | Eisenberger (2013) PLoS One 8: e78496 [Retinitis pigmentosa, autosomal recessive] | Abu-Safieh (2013) Genome Res 23: 236 [Retinitis pigmentosa] | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa]	Chr14:67725137G/C
c.226G>T	p.G76W	DM	Retinitis pigmentosa	Jespersgaard (2019) Sci Rep 9, | Hayman (2024) J Med Genet 61: 224 [Retinal disease] | Han (2025) NPJ Genom Med 10: 6 [Leber congenital amaurosis]	Chr14:67725137G/T
c.229G>T	p.E77*	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Xiong (2015) Science 347: 1254806 [Additional report]	Chr14:67725140G/T
c.250C>T	p.R84*	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis]	Chr14:67725161C/T
c.275T>A	p.V92E	DM	Leber congenital amaurosis	Ardehaie (2025) Clin Genet 107, 300	Chr14:67725186T/A
c.278T>C	p.L93P	DM	Retinitis pigmentosa	Avila-Fernandez (2010) Mol Vis 16, 2550 | Panneman (2023) Front Cell Dev Biol 11: 1112270 [Retinitis pigmentosa] | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinitis pigmentosa] | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinal dystrophy]	Chr14:67725189T/C
c.284G>A	p.R95Q	DM	Macular dystrophy	Hitti-Malin (2024) Biomolecules 14, epub	Chr14:67725195G/A
c.295C>A	p.L99I	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639 | Schobers (2025) Eur J Hum Genet 33: 56 [Visual impairment] | Ge (2015) Sci Rep 5: 18287 [Retinitis pigmentosa, autosomal recessive] | Zhang (2016) Sci Rep 6: 32792 [Retinitis pigmentosa, autosomal recessive]	Chr14:67725206C/A
c.302A>G	p.D101G	DM?	Retinal disease	Aleman (2018) Invest Ophthalmol Vis Sci 59, 5225 | Fahim (2019) Br J Ophthalmol 103: 1789 [Retinal degeneration] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67725213A/G
c.315C>G	p.I105M	DM?	Retinitis pigmentosa	Collin (2011) Invest Ophthalmol Vis Sci 52, 2227	Chr14:67725226C/G
c.316C>T	p.R106*	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Lin (2024) Ophthalmol Retina : [Retinal disease] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis]	Chr14:67725227C/T
c.325G>C	p.A109P	DM	Retinal degeneration	Fahim (2019) Br J Ophthalmol 103, 1789 | Sheck (2021) Mol Genet Genomic Med 9: [Retinal disease] | Sallum (2020) Am J Med Genet C Semin Med Genet 184: 728 [Leber congenital amaurosis] | Sallum (2024) Am J Ophthalmol 265: 257 [Leber congenital amaurosis]	Chr14:67725236G/C

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.349A>T	p.K117*	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Xiong (2015) Science 347: 1254806 [Additional report]	Chr14:67726056A/T
c.362T>C	p.I121T	DM	Retinitis pigmentosa	Scott (2020) Cold Spring Harb Mol Case Stud 6, a004754	Chr14:67726069T/C
c.375T>A	p.N125K	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865	Chr14:67726082T/A
c.377C>A	p.A126E	DM?	Retinitis pigmentosa	Stone (2017) Ophthalmology 124, 1314 | Fahim (2019) Br J Ophthalmol 103: 1789 [Retinal degeneration] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67726084C/A
c.377C>T	p.A126V	DM	Retinitis pigmentosa	Benayoun (2009) Am J Med Genet A 149A, 650 | Ge (2015) Sci Rep 5: 18287 [Retinitis pigmentosa, autosomal recessive] | Sharon (2020) Hum Mutat 41: 140 [Retinitis pigmentosa] | Zou (2018) Retina 39: 2040 [Retinal dystrophy, early-onset, severe]	Chr14:67726084C/T
c.379G>T	p.G127*	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639 | Patel (2016) Genet Med 18: 554 [Retinitis pigmentosa & rod cone dystrophy] | Weisschuh (2020) Hum Mutat 41: 1514 [Retinitis pigmentosa] | Sahin (2024) Cureus 16: e53742 [Retinitis pigmentosa]	Chr14:67726086G/T
c.383T>C	p.V128A	DM?	Retinitis pigmentosa	Gao (2019) Ophthalmology 126, 1549 | Li (2023) Invest Ophthalmol Vis Sci 64: [Leber congenital amaurosis] | Jin (2022) Brain Topogr : [Additional report]	Chr14:67726090T/C
c.383T>G	p.V128G	DM?	Retinal disease	Ellingford (2016) J Med Genet 53, 761 | Fahim (2019) Br J Ophthalmol 103: 1789 [Retinal degeneration] | Lin (2024) Ophthalmol Retina : [Additional report]	Chr14:67726090T/G
c.387G>A	p.M129I	DM	Retinal disease	Maddirevula (2020) Genome Biol 21, 145	Chr14:67726094G/A
c.393T>A	p.C131*	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67726100T/A
c.401C>T	p.S134F	DM	Retinitis pigmentosa	Jin (2022) Brain Topogr epub, epub	Chr14:67726108C/T
c.400T>C	p.S134P	DM?	Retinal disease	Aleman (2018) Invest Ophthalmol Vis Sci 59, 5225 | Fahim (2019) Br J Ophthalmol 103: 1789 [Retinal degeneration] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67726107T/C
c.403A>G	p.K135E	DM	Cone/cone-rod dystrophy	Boulanger-Scemama (2015) Orphanet J Rare Dis 10, 85	Chr14:67726110A/G
c.407C>G	p.T136R	DM	Leber congenital amaurosis	Jin (2022) Brain Topogr epub, epub	Chr14:67726114C/G
c.407C>T	p.T136I	DM	Retinal degeneration	Zampaglione (2020) Genet Med 22, 1079	Chr14:67726114C/T
c.433G>A	p.G145R	DM	Retinitis pigmentosa	Zou (2018) Retina 39, 2040 | Jin (2022) Brain Topogr : [Additional report]	Chr14:67726140G/A
c.434G>A	p.G145E	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865	Chr14:67726141G/A
c.437T>A	p.V146D	DM	Retinitis pigmentosa	Fu (2013) Invest Ophthalmol Vis Sci 54, 4158 | Liu (2020) Clin Exp Ophthalmol 49: 46 [Stargardt disease] | Jin (2022) Brain Topogr : [Retinal dystrophy, early onset] | Li (2023) Invest Ophthalmol Vis Sci 64: [Retinal dystrophy]	Chr14:67726144T/A
c.440A>C	p.N147T	DM	Macular dystrophy	Scott (2020) Cold Spring Harb Mol Case Stud 6, a004754 | Zampaglione (2020) Genet Med 22: 1079 [Retinal degeneration]	Chr14:67726147A/C
c.442C>A	p.H148N	DM?	Rod-cone dystrophy	Yohe (2020) Mol Genet Genomic Med 8, e1081	Chr14:67726149C/A
c.444C>A	p.H148Q	DM	Retinitis pigmentosa	Sahin (2024) Cureus 16, e53742	Chr14:67726151C/A
c.446T>C	p.L149P	DM	Retinitis pigmentosa	Chacon-Camacho (2013) Gene 528, 178 | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67726153T/C
c.451C>A	p.H151N	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639	Chr14:67726983C/A
c.451C>G	p.H151D	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639 | Weisschuh (2020) Hum Mutat 41: 1514 [Retinitis pigmentosa] | Mackay (2011) Mol Vis 17: 2706 [Retinal dystrophy] | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67726983C/G
c.454T>A	p.F152I	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67726986T/A
c.454T>C	p.F152L	DM	Macular dystrophy/cone-rod dystrophy	Suga (2022) Hum Mutat 43, 2251	Chr14:67726986T/C

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.464C>A	p.T155N	DM	Macular dystrophy	Hitti-Malin (2024) Biomolecules 14, 367	Chr14:67726996C/A
c.464C>T	p.T155I	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Zou (2018) Retina 39: 2040 [Retinitis pigmentosa, autosomal recessive] | Weisschuh (2020) Hum Mutat 41: 1514 [Retinitis pigmentosa] | Panneman (2023) Front Cell Dev Biol 11: 1112270 [Retinitis pigmentosa]	Chr14:67726996C/T
c.468C>G	p.Y156*	DM	Retinal dystrophy	Zou (2018) Retina 39, 2040	Chr14:67727000C/G
c.482G>A	p.R161Q	DM?	Stargardt disease	Zolnikova (2017) Eur J Med Genet 60, 140 | De Zaeytijd (2021) Retina 41: 1346 [Retinal dystrophy, intermediate]	Chr14:67727014G/A
c.481C>T	p.R161W	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Weisschuh (2024) J Med Genet 61: 186 [Retinitis pigmentosa, autosomal recessive] | Koyanagi (2019) J Med Genet 56: 662 [Retinitis pigmentosa] | Sahin (2024) Cureus 16: e53742 [Retinitis pigmentosa]	Chr14:67727013C/T
c.485T>C	p.L162P	DM	Leber congenital amaurosis	Aleman (2024) Invest Ophthalmol Vis Sci 65, 30	Chr14:67727017T/C
c.503C>T	p.A168V	DM?	Retinal disease	Karali (2022) Sci Rep 12,	Chr14:67727035C/T
c.506G>A	p.R169Q	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Yohe (2020) Mol Genet Genomic Med 8: e1081 [Rod-cone dystrophy] | Stone (2017) Ophthalmology 124: 1314 [Retinitis Pigmentosa] | Jin (2022) Brain Topogr : [Retinal dystrophy, early onset]	Chr14:67727038G/A
c.505C>G	p.R169G	DM	Leber congenital amaurosis	Wang (2015) Invest Ophthalmol Vis Sci 56, 3642 | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Zou (2018) Retina 39: 2040 [Retinal dystrophy, early-onset, severe] | Li (2023) Invest Ophthalmol Vis Sci 64: [Retinal dystrophy]	Chr14:67727037C/G
c.505C>T	p.R169W	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Fu (2013) Invest Ophthalmol Vis Sci 54: 4158 [Retinitis pigmentosa] | Wang (2018) Genes (Basel) 9: [Retinitis pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa]	Chr14:67727037C/T
c.508G>A	p.V170M	DM	Leber congenital amaurosis / retinal dystrophy	Xu (2020) Br J Ophthalmol 104, 932	Chr14:67727040G/A
c.524C>T	p.S175L	DM	Leber congenital amaurosis	Coppieters (2010) Hum Mutat 31, E1097 | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Xiao (2023) J Med Case Rep 17: 226 [Retinitis pigmentosa] | Neveling (2013) Hum Mutat 34: 1721 [Retinal dystrophy]	Chr14:67727056C/T
c.523T>C	p.S175P	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639	Chr14:67727055T/C
c.529G>C	p.A177P	DM?	Retinal disease	Villanueva-Mendoza (2021) Genes (Basel) 12,	Chr14:67727061G/C
c.535C>G	p.H179D	DM	Leber congenital amaurosis	Li (2017) J Zhejiang Univ Sci B 18, 421 | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Zou (2018) Retina 39: 2040 [Retinal dystrophy, early-onset, severe] | Li (2023) Invest Ophthalmol Vis Sci 64: [Retinal dystrophy]	Chr14:67727067C/G
c.542G>A	p.G181D	DM	Retinitis pigmentosa	Jin (2022) Brain Topogr epub, epub	Chr14:67727074G/A
c.559G>A	p.D187N	DM?	Maculopathy	De Zaeytijd (2021) Retina 41, 1346 | Panneman (2023) Front Cell Dev Biol 11: 1112270 [Retinal dystrophy]	Chr14:67727091G/A
c.560A>G	p.D187G	DM?	Retinitis pigmentosa	Griffith (2022) Genes (Basel) 13,	Chr14:67727092A/G
c.565C>T	p.Q189*	DM	Retinal dystrophy	Janecke (2004) Nat Genet 36, 850 | Karali (2022) Sci Rep 12: [Retinal disease] | Xiong (2015) Science 347: 1254806 [Additional report]	Chr14:67727097C/T
c.571G>A	p.E191K	DM?	Congenital heart disease	Dong (2025) Proc Natl Acad Sci U S A 122,	Chr14:67727103G/A
c.578G>A	p.R193H	DM?	Retinitis pigmentosa	Maltese (2022) Front Genet 13, 914345	Chr14:67727110G/A
c.582C>G	p.Y194*	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Wang (2013) J Med Genet 50: 674 [Leber congenital amaurosis] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67727114C/G
c.595G>C	p.A199P	DM?	Retinitis pigmentosa	Gao (2019) Ophthalmology 126, 1549 | Jin (2022) Brain Topogr : [Additional report] | Li (2023) Invest Ophthalmol Vis Sci 64: [Additional report]	Chr14:67727127G/C

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.599A>G	p.Y200C	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Xu (2014) Hum Genet 133: 1255 [Retinitis pigmentosa] | Sahin (2024) Cureus 16: e53742 [Retinitis pigmentosa] | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67727131A/G
c.598T>C	p.Y200H	DM	Retinitis pigmentosa	Xu (2014) Hum Genet 133, 1255 | Sallum (2020) Am J Med Genet C Semin Med Genet 184: 728 [Leber congenital amaurosis]	Chr14:67727130T/C
c.599A>C	p.Y200S	DM?	Retinitis pigmentosa	Weisschuh (2020) Hum Mutat 41, 1514	Chr14:67727131A/C
c.601T>C	p.C201R	DM	Leber congenital amaurosis	Sun (2007) Vision Res 47, 2055 | Midgley (2024) Mol Genet Genomic Med 12: e70046 [Retinitis pigmentosa] | Mackay (2011) Mol Vis 17: 2706 [Retinal dystrophy] | Sheck (2021) Mol Genet Genomic Med 9: [Retinal disease]	Chr14:67727133T/C
c.609C>A	p.S203R	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Holtan (2020) Acta Ophthalmol 98: 286 [Retinal disease] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis] | Karali (2022) Sci Rep 12: [Additional report]	Chr14:67727141C/A
c.610A>C	p.K204Q	DM	Retinitis pigmentosa	Jespersgaard (2019) Sci Rep 9,	Chr14:67727142A/C
c.617C>A	p.A206D	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67727149C/A
c.616G>C	p.A206P	DM?	Retinitis pigmentosa	Gao (2019) Ophthalmology 126, 1549 | Li (2023) Invest Ophthalmol Vis Sci 64: [Additional report]	Chr14:67727148G/C
c.617C>T	p.A206V	DM	Retinitis pigmentosa	Martin-Merida (2019) Ophthalmology 126, 1181	Chr14:67727149C/T
c.619A>G	p.N207D	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Sheck (2021) Mol Genet Genomic Med 9: [Retinal disease] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67727151A/G
c.623T>A	p.V208E	DM	Leber congenital amaurosis	Wang (2015) Invest Ophthalmol Vis Sci 56, 3642 | Jin (2022) Brain Topogr : [Additional report]	Chr14:67727155T/A
c.632C>T	p.T211I	DM?	Retinitis pigmentosa	Weisschuh (2024) J Med Genet 61, 186	Chr14:67727164C/T
c.635G>A	p.R212H	DM?	Retinitis pigmentosa	Talib (2021) Br J Ophthalmol ,	Chr14:67727167G/A
c.641T>C	p.L214P	DM?	Retinal disease	Aleman (2018) Invest Ophthalmol Vis Sci 59, 5225 | Weisschuh (2024) J Med Genet 61: 186 [Retinitis pigmentosa, autosomal recessive] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67727173T/C
c.664G>A	p.G222R	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis]	Chr14:67729196G/A
c.667G>T	p.V223F	DM	Cone-rod dystrophy	Liu (2020) Clin Exp Ophthalmol 49, 46 | Colombo (2021) Invest Ophthalmol Vis Sci 62: 13 [Retinitis pigmentosa, autosomal recessive] | Karali (2022) Sci Rep 12: [Retinal disease] | Hitti-Malin (2024) Biomolecules 14: epub [Macular dystrophy]	Chr14:67729199G/T
c.671C>T	p.T224I	DM	Retinal degeneration	Fahim (2019) Br J Ophthalmol 103, 1789 | Liu (2020) Clin Exp Ophthalmol 49: 46 [Stargardt disease]	Chr14:67729203C/T
c.677A>G	p.Y226C	DM	Retinal dystrophy	Janecke (2004) Nat Genet 36, 850 | Weisschuh (2020) Hum Mutat 41: 1514 [Retinitis pigmentosa] | Sheck (2021) Mol Genet Genomic Med 9: [Retinal disease] | Hitti-Malin (2024) Biomolecules 14: epub [Macular dystrophy]	Chr14:67729209A/G
c.685C>T	p.H229Y	DM	Leber congenital amaurosis / retinal dystrophy	Xu (2020) Br J Ophthalmol 104, 932	Chr14:67729217C/T
c.688C>G	p.P230A	DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639	Chr14:67729220C/G
c.689C>G	p.P230R	DM	Retinitis pigmentosa	Martin-Merida (2019) Ophthalmology 126, 1181 | Rodríguez-Muñoz (2020) J Mol Diagn 22: 532 [Leber congenital amaurosis]	Chr14:67729221C/G
c.692G>A	p.G231D	DM	Leber congenital amaurosis	Wang (2013) J Med Genet 50, 674 | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Additional report]	Chr14:67729224G/A

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.698T>A	p.V233D	DM	Leber congenital amaurosis	Coppieters (2010) Hum Mutat 31, E1097 | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Sallum (2020) Am J Med Genet C Semin Med Genet 184: 728 [Additional report] | Panneman (2023) Front Cell Dev Biol 11: 1112270 [Additional report]	Chr14:67729230T/A
c.697G>A	p.V233I	DM	Cone-rod dystrophy	Scott (2020) Cold Spring Harb Mol Case Stud 6, a004754 | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67729229G/A
c.697G>C	p.V233L	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Thompson (2017) Mol Genet Genomic Med 5: 652 [Leber congenital amaurosis] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67729229G/C
c.697G>T	p.V233F	DM?	Retinitis pigmentosa	Jespersgaard (2019) Sci Rep 9,	Chr14:67729229G/T
c.701G>A	p.R234H	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinitis pigmentosa] | De Zaeytijd (2021) Retina 41: 1346 [Retinal dystrophy, intermediate] | Zampaglione (2020) Genet Med 22: 1079 [Retinal degeneration]	Chr14:67729233G/A
c.710T>C	p.L237P	DM	Leber congenital amaurosis	Zou (2018) Retina 39, 2040	Chr14:67729242T/C
c.716G>A	p.R239Q	DM	Retinal dystrophy	Beryozkin (2014) Invest Ophthalmol Vis Sci 55, 1149 | Griffith (2022) Genes (Basel) 13: epub [Retinitis pigmentosa] | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinal dystrophy] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis]	Chr14:67729248G/A
c.715C>G	p.R239G	DM	Leber congenital amaurosis	Ma (2021) BMC Med Genomics 14, 74	Chr14:67729247C/G
c.716G>T	p.R239L	DM	Leber congenital amaurosis	Zanolli (2020) Ophthalmic Genet 41, 36 | Sharon (2020) Hum Mutat 41: 140 [Retinitis pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Schlottmann (2023) NPJ Genom Med 8: 8 [Retinitis pigmentosa]	Chr14:67729248G/T
c.715C>T	p.R239W	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Lin (2024) Ophthalmol Retina : [Retinal disease] | Mackay (2011) Mol Vis 17: 2706 [Additional report]	Chr14:67729247C/T
c.728T>A	p.L243Q	DM?	Retinitis pigmentosa	Yohe (2020) Mol Genet Genomic Med 8, e1081	Chr14:67729260T/A
c.731T>A	p.L244H	DM?	Leber congenital amaurosis/retinitis pigmentosa	Avela (2019) Acta Ophthalmol 97, 805	Chr14:67729263T/A
c.746G>T	p.R249L	DM	Leber congenital amaurosis	Srikrupa (2018) Clin Genet 93, 329	Chr14:67729278G/T
c.749T>C	p.L250P	DM	Retinitis pigmentosa	Gupta (2022) Mol Vis 28, 203	Chr14:67729281T/C
c.764T>G	p.V255G	DM	Macular dystrophy	Hitti-Malin (2024) Biomolecules 14, epub	Chr14:67729296T/G
c.770C>T	p.T257M	DM	Retinitis pigmentosa	Jin (2022) Brain Topogr epub, epub	Chr14:67729302C/T
c.781G>A	p.G261R	DM?	Retinitis pigmentosa	Panneman (2023) Front Cell Dev Biol 11, 1112270 | De Zaeytijd (2021) Retina 41: 1346 [Maculopathy] | Gopinath (2023) Graefes Arch Clin Exp Ophthalmol 261: 2003 [Additional report]	Chr14:67729313G/A
c.785C>G	p.A262G	DM?	Leber congenital amaurosis	Hitti-Malin (2022) Hum Mutat 43, 2234 | Hitti-Malin (2024) Biomolecules 14: epub [Macular dystrophy]	Chr14:67729317C/G
c.788A>C	p.Q263P	DM	Retinitis pigmentosa	Panneman (2023) Front Cell Dev Biol 11, 1112270	Chr14:67729320A/C
c.795C>A	p.S265R	DM	Leber congenital amaurosis	Rodríguez-Muñoz (2020) J Mol Diagn 22, 532 | Del Pozo-Valero (2022) Invest Ophthalmol Vis Sci 63: [Cone-rod dystrophy]	Chr14:67729327C/A
c.800A>C	p.H267P	DM?	Leber congenital amaurosis	Aleman (2024) Invest Ophthalmol Vis Sci 65, 30	Chr14:67729332A/C
c.803G>T	p.C268F	DM	Retinitis pigmentosa	Coppieters (2014) Genet Med 16, 671	Chr14:67729335G/T
c.806C>G	p.A269G	DM	Cone-rod dystrophy	Huang (2016) Exp Eye Res 146, 252 | Kao (2024) NPJ Genom Med 9: 4 [Retinitis pigmentosa] | Kuo (2023) J Med Genet 60: 859 [Maculopathy, pseudocoloboma-like] | Chen (2021) NPJ Genom Med 6: [Macular dystrophy]	Chr14:67729338C/G

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.821T>C	p.L274P	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Beryozkin (2014) Invest Ophthalmol Vis Sci 55: 1149 [Retinitis pigmentosa, early-onset] | Sharon (2020) Hum Mutat 41: 140 [Retinitis pigmentosa] | Hayman (2024) J Med Genet 61: 224 [Retinal disease]	Chr14:67729353T/C
c.823G>T	p.E275*	DM	Leber congenital amaurosis	Wang (2013) J Med Genet 50, 674 | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Additional report]	Chr14:67729355G/T
c.844T>G	p.F282V	DM	Macular dystrophy	Consugar (2015) Genet Med 17, 253 | Zampaglione (2022) Genet Med 24: 332 [Retinal degeneration] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis]	Chr14:67729376T/G
c.854G>A	p.C285Y	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865	Chr14:67733751G/A
c.867G>A	p.W289*	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67733764G/A
c.869T>G	p.V290G	DM	Retinitis pigmentosa	Eisenberger (2013) PLoS One 8, e78496 | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Lee (2015) Am J Ophthalmol 160: 354.e9 [Additional report]	Chr14:67733766T/G
c.872C>T	p.S291F	DM	Retinal dystrophy	Neveling (2013) Hum Mutat 34, 1721 | Panneman (2023) Front Cell Dev Biol 11: 1112270 [Retinitis pigmentosa] | De Zaeytijd (2021) Retina 41: 1346 [Maculopathy]	Chr14:67733769C/T
c.883C>T	p.R295*	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Stone (2017) Ophthalmology 124: 1314 [Retinitis Pigmentosa] | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67733780C/T
c.910T>C	p.W304R	DM?	Retinal disease	Ellingford (2016) J Med Genet 53, 761 | Gao (2019) Ophthalmology 126: 1549 [Retinitis pigmentosa] | Li (2023) Invest Ophthalmol Vis Sci 64: [Retinal dystrophy] | Fahim (2019) Br J Ophthalmol 103: 1789 [Retinal degeneration]	Chr14:67733807T/C
c.912G>A	p.W304*	DM	Leber congenital amaurosis	Sodi (2010) J AAPOS 14, 349	Chr14:67733809G/A
c.917T>G	p.V306G	DM?	Leber congenital amaurosis	Eisenberger (2013) PLoS One 8, e78496	Chr14:67733814T/G
c.938T>G	p.I313S	DM	Macular dystrophy	Hitti-Malin (2024) Biomolecules 14, epub	Chr14:67733835T/G
c.940C>T	p.R314W	DM?	Retinitis pigmentosa	Huang (2017) Sci Rep 7, 1948	Chr14:67733837C/T
		DM	Leber congenital amaurosis	Sundaramurthy (2016) J Hum Genet 61, 951	
c.68+1G>A		DM	Leber congenital amaurosis	Wang (2016) Graefes Arch Clin Exp Ophthalmol 254, 2227 | Jin (2022) Brain Topogr : [Retinitis pigmentosa] | Xu (2020) Br J Ophthalmol 104: 932 [Leber congenital amaurosis / retinal dystrophy] | Zhu (2021) Ophthalmic Genet 42: 392 [Additional report]	Chr14:67722711G/A
c.188-1G>A		DM?	Retinitis pigmentosa	Gao (2019) Ophthalmology 126, 1549 | Li (2023) Invest Ophthalmol Vis Sci 64: [Retinal dystrophy] | Jin (2022) Brain Topogr : [Additional report]	Chr14:67725098G/A
c.188G>T		DM	Leber congenital amaurosis	Eisenberger (2013) PLoS One 8, e78496	Chr14:67725099G/T
c.344-8C>T		DM?	Leber congenital amaurosis	Srilekha (2015) PLoS One 10, e0131679	Chr14:67726043C/T
c.343+1G>A		DM	Retinitis pigmentosa	Zou (2018) Retina 39, 2040 | Zhu (2021) Ophthalmic Genet 42: 392 [Leber congenital amaurosis] | Wang (2021) Front Genet 12: 715100 [Functional characterisation] | Jin (2022) Brain Topogr : [Additional report]	Chr14:67725255G/A
c.343+2T>G		DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67725256T/G
c.448+1G>A		DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Lin (2024) Ophthalmol Retina : [Retinal disease] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Ba-Abbad (2020) Ophthalmic Genet 41: 612 [Additional case report]	Chr14:67726156G/A
c.448+1G>C		DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67726156G/C
c.659-2A>T		DM	Retinal disease	Karali (2022) Sci Rep 12, 20815	Chr14:67729189A/T
c.658+1G>A		DM	Leber congenital amaurosis	Perrault (2004) Am J Hum Genet 75, 639 | Beryozkin (2014) Invest Ophthalmol Vis Sci 55: 1149 [Retinitis pigmentosa, early-onset] | Sharon (2020) Hum Mutat 41: 140 [Retinitis pigmentosa] | Ezer (2022) Am J Med Genet A 188: 2555 [Retinitis pigmentosa]	Chr14:67727191G/A

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.849-2A>G		DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67733744A/G
c.848+1G>A		DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67729381G/A
c.848+1G>C		DM?	Retinitis pigmentosa	Griffith (2022) Genes (Basel) 13,	Chr14:67729381G/C
c.848+2T>C		DM	Retinal disease	Ellingford (2016) J Med Genet 53, 761	Chr14:67729382T/C
c.848+82C>G		DM?	Retinal dystrophy	AlBakri (2015) J AAPOS 19, 568	Chr14:67729462C/G
c.-123C>T		DM	Macular dystrophy	Dueñas Rey (2024) Genome Med 16, 7	Chr14:67722520C/T
c.43delC	p.(Leu15Cysfs*4)	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67722683TC/T
c.52delG	p.(Val18*)	DM?	Developmental and epileptic encephalopathy	Takata (2019) Nat Commun 10, | Suga (2022) Hum Mutat 43: 2251 [Leber congenital amaurosis]	Chr14:67722692TG/T
c.57_59delTCC	p.(Pro20del)	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67722697GCTC/G
c.63_66delCATC	p.(Ile22Glyfs*19)	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Gupta (2022) Mol Vis 28: 203 [Retinitis pigmentosa] | Demas (2025) BMC Med Genomics 18: 139 [Retinal disease] | Aleman (2018) Invest Ophthalmol Vis Sci 59: 5225 [Additional report]	Chr14:67722698CTCCA/C
c.101delA	p.(Asn34Metfs*8)	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67724502CA/C
c.105delG	p.(Gln36Serfs*6)	DM	Cone-rod dystrophy	Habibi (2021) Front Cell Dev Biol 9,	Chr14:67724508TG/T
c.189delA	p.(Ala64Profs*11)	DM	Retinitis pigmentosa	Gupta (2022) Mol Vis 28, 203	Chr14:67725099GA/G
c.381delA	p.(Val128*)	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706	Chr14:67726087GA/G
c.448+4_448+7delAGTA		DM	Leber congenital amaurosis	Han (2025) NPJ Genom Med 10, 6	Chr14:67726155GGTAA/G
c.495_499delTGCCC	p.(Ala166Cysfs*5)	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791	Chr14:67727025TCTGCC/T
c.546delG	p.(Ile183Phefs*95)	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67727077AG/A
c.554delT	p.(Phe185Serfs*93)	DM	Leber congenital amaurosis	Chau (2022) NPJ Genom Med 7,	Chr14:67727084CT/C
c.601delT	p.(Cys201Alafs*77)	DM	Leber congenital amaurosis	Wang (2015) Invest Ophthalmol Vis Sci 56, 3642	Chr14:67727131AT/A
c.698_711del14	p.(Val233Glyfs*35)	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67729228CGTCCGCTCTGAGCT/C
c.723delC	p.(Ser242Profs*36)	DM	Leber congenital amaurosis / retinal dystrophy	Xu (2020) Br J Ophthalmol 104, 932	Chr14:67729253TC/T
c.724_726delTCC	p.(Ser242del)	DM	Leber congenital amaurosis	Wang (2015) Invest Ophthalmol Vis Sci 56, 3642	Chr14:67729251ACTC/A
c.735_743delCCTGCTCTG	p.(Cys245_Leu247del)	DM	Retinal dystrophy	Ba-Abbad (2020) Ophthalmic Genet 41, 612 | Lin (2024) Ophthalmol Retina : [Retinal disease] | Dueñas Rey (2024) Genome Med 16: 7 [Macular dystrophy]	Chr14:67729257CCCTGCTCTG/C
c.743_751delGGCGGCTCT	p.(Trp248_Leu250del)	DM	Retinitis pigmentosa	Zou (2018) Retina 39, 2040	Chr14:67729269TGCTCTGGCG/T
c.759delC	p.(Phe254Leufs*24)	DM	Retinitis pigmentosa	Sarkar (2020) Front Genet 11, 335 | Hayman (2024) J Med Genet 61: 224 [Retinal disease] | Lin (2024) Ophthalmol Retina : [Retinal disease] | Han (2025) NPJ Genom Med 10: 6 [Leber congenital amaurosis]	Chr14:67729286TC/T
c.763delG	p.(Val255Serfs*23)	DM	Retinitis pigmentosa	Muthiah (2022) Br J Ophthalmol 106, 1274 | Sheck (2021) Mol Genet Genomic Med 9: [Retinal disease] | Lin (2024) Ophthalmol Retina : [Additional report]	Chr14:67729294TG/T
c.767delA	p.(Lys256Argfs*22)	DM	Retinitis pigmentosa	Jin (2022) Brain Topogr epub, epub	Chr14:67729297CA/C
c.778delG	p.(Glu260Argfs*18)	DM	Retinitis pigmentosa	Fingert (2008) Arch Ophthalmol 126, 1301 | Lin (2024) Ophthalmol Retina : [Retinal disease]	Chr14:67729307CG/C
c.784delG	p.(Ala262Argfs*16)	DM	Leber congenital amaurosis	Stone (2007) Am J Ophthalmol 144, 791 | Yohe (2020) Mol Genet Genomic Med 8: e1081 [Retinitis pigmentosa]	Chr14:67729311AG/A
c.793_796delAGCC	p.(Ser265Cysfs*12)	DM?	Retinal dystrophy	Basdemirci (2024) Mol Syndromol 15, 202	Chr14:67729322ACCAG/A

DNA	Protein	Variant class	Disease	References	VCF (hg38)
c.806_810delCCCTG	p.(Ala269Glyfs*2)	DM	Retinal dystrophy	Janecke (2004) Nat Genet 36, 850 | Ge (2015) Sci Rep 5: 18287 [Retinitis pigmentosa] | Rodríguez-Muñoz (2020) J Mol Diagn 22: 532 [Retinitis pigmentosa] | Panneman (2023) Front Cell Dev Biol 11: 1112270 [Retinitis pigmentosa]	Chr14:67729336CGCCCT/C
c.829_830delCT	p.(Leu277Glufs*7)	DM?	Leber congenital amaurosis	Hanany (2020) Proc Natl Acad Sci U S A 117, 2710	Chr14:67729360CCT/C
c.99_102dupAAAT	p.(Val35Lysfs*28)	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865	Chr14:67724502C/CAAAT
c.207dupC	p.(Cys70Leufs*13)	R	Retinitis pigmentosa	Perez-Carro (2016) Sci Rep 6, 19531	Chr14:67725116G/GC
c.210dupC	p.(Arg71Glnfs*12)	DM	Retinitis pigmentosa	Martin-Merida (2019) Ophthalmology 126, 1181 | Capalbo (2019) PLoS Genet 15: e1008409 [Specific phenotype not reported] | Hanany (2020) Proc Natl Acad Sci U S A 117: 2710 [Leber congenital amaurosis] | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67725120G/GC
c.630dupT	p.(Thr211Tyrfs*3)	DM	Retinitis pigmentosa	Panneman (2023) Front Cell Dev Biol 11, 1112270	Chr14:67727157C/CT
c.658+2dupT		DM	Retinitis pigmentosa	Panneman (2023) Front Cell Dev Biol 11, 1112270	Chr14:67727191G/GT
c.697_698dupGT	p.(Arg234Serfs*45)	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706	Chr14:67729228C/CGT
c.697_698insGT	p.(Val233Glyfs*46)	DM?	Retinal disease	Lin (2024) Ophthalmol Retina 8, 699	Chr14:67729229G/GGT
c.712dupG	p.(Val238Glyfs*35)	DM	Leber congenital amaurosis	Aleman (2024) Invest Ophthalmol Vis Sci 65, 30	Chr14:67729242T/TG
c.715dupC	p.(Arg239Profs*34)	DM	Retinal dystrophy	Mackay (2011) Mol Vis 17, 2706 | Lin (2024) Ophthalmol Retina : [Retinal disease] | Moya (2024) Int J Mol Sci 25: epub [Leber congenital amaurosis] | Aleman (2018) Invest Ophthalmol Vis Sci 59: 5225 [Additional report]	Chr14:67729245T/TC
c.759dupC	p.(Phe254Leufs*19)	DM	Retinal disease	Abolhassani (2024) NPJ Genom Med 9, 12	Chr14:67729286T/TC
c.784dupG	p.(Ala262Glyfs*11)	DM	Leber congenital amaurosis	Borck (2015) Genome Res 25, 155 | Karali (2022) Sci Rep 12: [Retinal disease] | Del Pozo-Valero (2022) Invest Ophthalmol Vis Sci 63: [Cone-rod dystrophy]	Chr14:67729311A/AG
c.429_432delCCTGinsGGT	p.(His143Glnfs*20)	DM	Retinal dystrophy	Thompson (2005) Hum Mol Genet 14, 3865 | Aleman (2024) Invest Ophthalmol Vis Sci 65: 30 [Leber congenital amaurosis]	Chr14:67726135ACCTG/AGGT
c.680_684delCAGTGinsT	p.(Ala227Valfs*50)	DM	Retinal degeneration	Fahim (2019) Br J Ophthalmol 103, 1789 | Colombo (2021) Invest Ophthalmol Vis Sci 62: 13 [Retinitis pigmentosa, autosomal dominant]	Chr14:67729211GCAGTG/GT
c.698_699delTCinsAA	p.(Val233Glu)	DM?	Leber congenital amaurosis	Sallum (2020) Am J Med Genet C Semin Med Genet 184, 728	Chr14:67729229GTC/GAA
		DM	Leber congenital amaurosis	Turro (2020) Nature 583, 96	
c.648_658+20del31	p.(Arg217Hisfs*16)	DM	Retinal disease	Ellingford (2016) J Med Genet 53, 761 | Zhu (2022) Genes (Basel) 13: [Additional report]	Chr14:67727178AAGAGGCTCCAAGGTAAGTCTGGAGAAAGAGG/A
c.658+591_*603delinsCT		DM	Retinitis pigmentosa	Neveling (2012) Hum Mutat 33, 963	
		DM?	Retinal disease	Lin (2024) Ophthalmol Retina 8, 699	
		DM	Retinitis pigmentosa	Collin (2011) Invest Ophthalmol Vis Sci 52, 2227	