Ophthalmology

Ophthalmology

Single Gene

Code	Diseases	Gene
sCe3/0001	Achromatopsia type 2	CNGA3
sCe3/0002	Achromatopsia type 3	CNGB3
sCe3/0003	Achromatopsia type 4	GNAT2
sCe3/0004	Achromatopsia type 6	PDE6H
sCe3/0005	Aland Island eye disease	CACNA1F
sCe3/0006	Albinism, ocular type I, Nettleship-Falls type	GPR143
sCe3/0007	Albinism, oculocutaneous nonsyndromic	SLC24A5
sCe3/0008	Albinism, oculocutaneous type 1A	TYR
sCe3/0009	Albinism, oculocutaneous type 1B	TYR
sCe3/0010	Albinism, oculocutaneous type 2	OCA2
sCe3/0011	Albinism, oculocutaneous type 3	TYRP1
sCe3/0012	Albinism, oculocutaneous type 4	SLC45A2
sCe3/0013	Albinism, oculocutaneous type 5	C10ORF11
sCe3/0014	Alstrom syndrome	ALMS1
sCe3/0015	Aniridia	PAX6
sCe3/0016	Anterior segment mesenchymal dysgenesis	PITX3
sCe3/0017	Bardet-Biedl syndrome type 14	CEP290
sCe3/0018	Bestrophinopathy	BEST1
sCe3/0019	Bietti crystalline corneoretinal dystrophy	CYP4V2
sCe3/0020	Blepharophimosis, epicanthus inversus, and ptosis	FOXL2
sCe3/0021	Blepharophimosis-ptosis-intellectual disability syndrome	UBE3B
sCe3/0022	Bothnia retinal dystrophy	RLBP1
sCe3/0023	Bradyopsia	RGS9BP
sCe3/0024	Bradyopsia	RGS9
sCe3/0025	Branchiootorenal syndrome type 1	EYA1

sCe3/0026	Branchiootorenal syndrome type 2	SIX5
sCe3/0027	Brittle cornea syndrome	ZNF469
sCe3/0028	Cataract 11, multiple types	PITX3
sCe3/0029	Cataract type 17, multiple types	CRYBB1
sCe3/0030	Cataract type 23	CRYBA4
sCe3/0031	Cataract type 41	WFS1
sCe3/0032	Cataract type 43	UNC45B
sCe3/0033	Cataract, autosomal dominant	GCNT2
sCe3/0034	Cataract, autosomal recessive congenital nuclear type 2	CRYBB3
sCe3/0035	Cataract, autosomal recessive congenital type 1	CRYAA
sCe3/0036	Cataract, autosomal recessive congenital type 2	FYCO1
sCe3/0037	Cataract, autosomal recessive congenital type 4	TDRD7
sCe3/0038	Cataract, autosomal recessive type 38	AGK
sCe3/0039	Cataract, congenital	SORD
sCe3/0040	Cataract, congenital, associated with Marinesco-Sjogren Syndrome	SIL1
sCe3/0041	Cataract, cortical pulverulent, late-onset	LIM2
sCe3/0042	Cataract, lamellar	HSF4
sCe3/0043	Cataract, posterior polar type 2	CRYAB
sCe3/0044	Cataract, pulverulent or cerulean, with or without microcornea	MAF
sCe3/0045	Cataract, X-linked	NHS
sCe3/0046	Cataract-microcornea syndrome	GJA8
sCe3/0047	Cerebellar-retinal degeneration, infantile	ACO2
sCe3/0048	Choroidal dystrophy, central areolar type 2	PRPH2
sCe3/0049	Coat plus syndrome	CTC1
sCe3/0050	Coloboma of optic nerve	PAX6
sCe3/0051	Coloboma, ocular, autosomal dominant	PAX6
sCe3/0052	Colobomatous microphthalmia	TENM1
sCe3/0053	Cone-rod dystrophy	UNC119
sCe3/0054	Cone-rod dystrophy	AIPL1

sCe3/0055	Cone-rod dystrophy type 11	RAX2
sCe3/0056	Cone-rod dystrophy type 12	PROM1
sCe3/0057	Cone-rod dystrophy type 13	RPGRIP1
sCe3/0058	Cone-rod dystrophy type 14	GUCA1A
sCe3/0059	Cone-rod dystrophy type 15	CDHR1
sCe3/0060	Cone-rod dystrophy type 17	CD3G
sCe3/0061	Cone-rod dystrophy type 2	CRX
sCe3/0062	Cone-rod dystrophy type 20	POC1B
sCe3/0063	Cone-rod dystrophy type 3	ABCA4
sCe3/0064	Cone-rod dystrophy type 4	PDE6C
sCe3/0065	Cone-rod dystrophy type 5	PITPNM3
sCe3/0066	Cone-rod dystrophy type 7	RIMS1
sCe3/0067	Cone-rod dystrophy type 9	ADAM9
sCe3/0068	Cone-rod dystrophy, C21orf2 related	C21orf2
sCe3/0069	Cone-rod dystrophy, X-linked type 3	CACNA1F
sCe3/0070	Corneal dystrophy, epithelial basement membrane	TGFBI
sCe3/0071	Corneal dystrophy, posterior polymorphous, type 1	VSX1
sCe3/0072	Corneal endothelial dystrophy type 2	SLC4A11
sCe3/0073	Corneal intraepithelial dyskeratosis and ectodermal dysplasia	NLRP1
sCe3/0074	Corneal opacification and other ocular anomalies	PXDN
sCe3/0075	Doyne honeycob retinal dystrophy	EFEMP1
sCe3/0076	Duane Retraction syndrome	CHN1
sCe3/0077	Duane Retraction syndrome	SALL4
sCe3/0078	Dyschromatosis symmetrica hereditaria	ADAR
sCe3/0079	Early onset glaucoma, phenotype modifier of, COL15A1 related	COL15A1
sCe3/0080	Ectopia lentis et papillae	ADAMTSL4
sCe3/0081	Ectopia lentis, familial	FBN1
sCe3/0082	Ectopia lentis, isolated, autosomal recessive	ADAMTSL4
sCe3/0083	Exfoliation syndrome, susceptibility to	LOXL1

sCe3/0084	Exudative vitreoretinopathy	FZD4
sCe3/0085	Exudative vitreoretinopathy type 2	NDP
sCe3/0086	Exudative vitreoretinopathy type 5	TSPAN12
sCe3/0087	Fibrosis of extraocular muscles, congenital type 1	KIF21A
sCe3/0088	Fibrosis of extraocular muscles, congenital type 3a	TUBB3
sCe3/0089	Fleck retina, familial benign	PLA2G5
sCe3/0090	Foveal hypoplasia type 1	PAX6
sCe3/0091	Fundus albipunctatus	PRPH2
sCe3/0092	Fundus albipunctatus	RDH5
sCe3/0093	Gaze palsy, horizontal, with progressive scoliosis	ROBO3
sCe3/0094	Gillespie syndrome	ITPR1
sCe3/0095	Glaucoma, open angle type 1A	MYOC
sCe3/0096	Glaucoma, open angle type 1E	OPTN
sCe3/0097	Glaucoma, open angle type 1F	NTF4
sCe3/0098	Glaucoma, open angle type 1F	ASB10
sCe3/0099	Glaucoma, open angle type 1G	WDR36
sCe3/0100	Glaucoma, primary type 3A	CYP1B1
sCe3/0101	Glaucoma, primary type 3D	LTBP2
sCe3/0102	Gyrate atrophy of choroid and retina with or without ornithinemia	OAT
sCe3/0103	Hereditary Retinoblastoma	RB1
sCe3/0104	Hermansky Pudlak syndrome type 4	HPS4
sCe3/0105	Hermansky-Pudlak syndrome type 1	HPS1
sCe3/0106	Hermansky-Pudlak syndrome type 2	AP3B1
sCe3/0107	Hermansky-Pudlak syndrome type 3	HPS3
sCe3/0108	Hermansky-Pudlak syndrome type 5	HPS5
sCe3/0109	Hermansky-Pudlak syndrome type 6	HPS6
sCe3/0110	Hermansky-Pudlak syndrome type 7	DTNBP1
sCe3/0111	Hermansky-Pudlak syndrome type 8	BLOC1S3
sCe3/0112	Hyperferritinemia-cataract syndrome	FTL

sCe3/0113	Iridogoniodysgenesis type 1	FOXC1
sCe3/0114	Jalili syndrome	CNNM4
sCe3/0115	Keratoconus type 1	VSX1
sCe3/0116	Knobloch syndrome type 1	COL18A1
sCe3/0117	Leber congenital amaurosis type 1	GUCY2D
sCe3/0118	Leber congenital amaurosis type 10	CEP290
sCe3/0119	Leber congenital amaurosis type 11	IMPDH1
sCe3/0120	Leber congenital amaurosis type 12	RD3
sCe3/0121	Leber congenital amaurosis type 16	KCNJ13
sCe3/0122	Leber congenital amaurosis type 17	GDF6
sCe3/0123	Leber congenital amaurosis type 3	SPATA7
sCe3/0124	Leber congenital amaurosis type 5	LCA5
sCe3/0125	Leber congenital amaurosis type 6	RPGRIP1
sCe3/0126	Leber congenital amaurosis type 7	CRX
sCe3/0127	Leber congenital amaurosis type 8	CRB1
sCe3/0128	Leber congenital amaurosis type 9	NMNAT1
sCe3/0129	Leber congenital amaurosis with myopathy	DTHD1
sCe3/0130	Leber optic atrophy	MT-ND4
sCe3/0131	Leber optic atrophy	MT-ND2
sCe3/0132	Leber optic atrophy	MT-ATP6
sCe3/0133	Leber optic atrophy	MT-CYB
sCe3/0134	Leber optic atrophy	MT-ND1
sCe3/0135	Leber optic atrophy	MT-ND6
sCe3/0136	Leber optic atrophy	MT-ND5
sCe3/0137	Leber optic atrophy	MT-CO1
sCe3/0138	Leber optic atrophy	MT-CO3
sCe3/0139	Leber optic atrophy	MT-ND4L
sCe3/0140	Macular degeneration, age-related type 11	CST3
sCe3/0141	Macular degeneration, age-related type 3	FBLN5

sCe3/0142	Macular degeneration, age-related type 6	RAX2
sCe3/0143	Macular degeneration, age-related type 8, association with	ARMS2
sCe3/0144	Macular degeneration, early-onset	FBN2
sCe3/0145	Macular dystrophy retinal type 2	PROM1
sCe3/0146	Macular dystrophy with central cone involvement	MFSD8
sCe3/0147	Macular dystrophy, BEST2-related	BEST2
sCe3/0148	Macular dystrophy, BEST3-related	BEST3
sCe3/0149	Macular dystrophy, BEST4-related	BEST4
sCe3/0150	Macular dystrophy, vitelliform	PRPH2
sCe3/0151	Macular dystrophy, vitelliform	BEST1
sCe3/0152	Mainzer Saldino syndrome	IFT140
sCe3/0153	Marshall syndrome	COL11A1
sCe3/0154	Megalocornea, X-linked	CHRDL1
sCe3/0155	MELAS syndrome, MT-TL1 related	MT-TL1
sCe3/0156	Microphthalmia syndromic type 2	BCOR
sCe3/0157	Microphthalmia syndromic type 3	SOX2
sCe3/0158	Microphthalmia syndromic type 5	OTX2
sCe3/0159	Microphthalmia syndromic type 6	SIX6
sCe3/0160	Microphthalmia syndromic type 6	BMP4
sCe3/0161	Microphthalmia syndromic type 7	HCCS
sCe3/0162	Microphthalmia syndromic type 8	ALDH1A3
sCe3/0163	Microphthalmia syndromic type 9	STRA6
sCe3/0164	Microphthalmia, isolated type 2	VSX2
sCe3/0165	Microphthalmia, isolated type 3	RAX
sCe3/0166	Microphthalmia, isolated type 4	GDF6
sCe3/0167	Microphthalmia, isolated type 5	MFRP
sCe3/0168	Microphthalmia, isolated type 6	PRSS56
sCe3/0169	Microphthalmia, isolated type 9	GDF3
sCe3/0170	Microphthalmia, isolated with coloboma type 3	VSX2

sCe3/0171	Microphthalmia, isolated with coloboma type 6, digenic	GDF6
sCe3/0172	Microphthalmia, isolated with coloboma type 6, digenic	GDF3
sCe3/0173	Microphthalmia, isolated with coloboma type 9	TENM3
sCe3/0174	Microphthalmia, syndromic type 1	NAA10
sCe3/0175	Microphthalmia, syndromic type 11	VAX1
sCe3/0176	Microphthalmia, VAX2 related	VAX2
sCe3/0177	Microspherophakia and/or megalocornea	LTBP2
sCe3/0178	Nance-Horan syndrome	NHS
sCe3/0179	Nanophthalmia type 2	MFRP
sCe3/0180	Night blindness type 1, congenital stationary, autosomal dominant	RHO
sCe3/0181	Night blindness, congenital stationar type 1B	GRM6
sCe3/0182	Night blindness, congenital stationar type 1C	TRPM1
sCe3/0183	Night blindness, congenital stationary type 1A	NYX
sCe3/0184	Night blindness, congenital stationary type 2A	CACNA1F
sCe3/0185	Night blindness, congenital stationary type 2B	CABP4
sCe3/0186	Night blindness, congenital stationary type 3	GNAT1
sCe3/0187	Night blindness, congenital stationary, autosomal dominant type 2	PDE6B
sCe3/0188	Night blindness, congenital stationary, type 1E	GPR179
sCe3/0189	Nonarteritic anterior ischemic optic neuropathy	GP1BA
sCe3/0190	Nystagmus type 1	FRMD7
sCe3/0191	Nystagmus type 6	GPR143
sCe3/0192	Occult macular dystrophy	RP1L1
sCe3/0193	Oguchi disease	GRK1
sCe3/0194	Oguchi disease	SAG
sCe3/0195	Ophthalmoplegia, isolated, MT-TN related	MT-TN
sCe3/0196	Optic atrophy type 1	OPA1
sCe3/0197	Optic atrophy type 3	OPA3
sCe3/0198	Optic atrophy type 7	TMEM126A
sCe3/0199	Optic atrophy type 9	ACO2

sCe3/0200	Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia, and neuropathy	OPA1
sCe3/0201	Patterned dystrophy of retinal pigment epithelium	PRPH2
sCe3/0202	Peters Anomaly	PITX2
sCe3/0203	Peters anomaly	PAX6
sCe3/0204	Peters Anomaly	CYP1B1
sCe3/0205	Peters-Plus syndrome	B3GLCT
sCe3/0206	Pigmented paravenous chorioretinal atrophy	CRB1
sCe3/0207	Plasminogen deficiency type 1	PLG
sCe3/0208	Progressive external ophthalmoplegia with mitochondrial deletions type 1	POLG
sCe3/0209	Progressive external ophthalmoplegia with mitochondrial deletions type 3	TWNK
sCe3/0210	Progressive external ophthalmoplegia with mitochondrial deletions type 4	POLG2
sCe3/0211	Progressive external ophthalmoplegia with mitochondrial deletions type 6	DNA2
sCe3/0212	Progressive external ophthalmoplegia with mitochondrial deletions, autosomal recessive	POLG
sCe3/0213	Retinal cone dystrophy type 3B	KCNV2
sCe3/0214	Retinal cone dystrophy type 4	CACNA2D4
sCe3/0215	Retinal degeneration, late-onset, autosomal dominant	C1QTNF5
sCe3/0216	Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities	ITM2B
sCe3/0217	Retinal nonattachment nonsyndromic congenital	ATOH7
sCe3/0218	Retinitis pigmentosa juvenile	LRAT
sCe3/0219	Retinitis pigmentosa SEMA4C related	SEMA4C
sCe3/0220	Retinitis pigmentosa type 1, autosomal dominant	RP1
sCe3/0221	Retinitis pigmentosa type 10, autosomal dominant	IMPDH1
sCe3/0222	Retinitis pigmentosa type 11, autosomal dominant	PRPF31
sCe3/0223	Retinitis pigmentosa type 12, autosomal recessive	CRB1
sCe3/0224	Retinitis pigmentosa type 13, autosomal dominant	PRPF8
sCe3/0225	Retinitis pigmentosa type 14, autosomal recessive	TULP1
sCe3/0226	Retinitis pigmentosa type 17, autosomal dominant	CA4

sCe3/0227	Retinitis pigmentosa type 18, autosomal dominant	PRPF3
sCe3/0228	Retinitis pigmentosa type 19, autosomal recessive	ABCA4
sCe3/0229	Retinitis pigmentosa type 2 X-linked	RP2
sCe3/0230	Retinitis pigmentosa type 20, autosomal recessive	RPE65
sCe3/0231	Retinitis pigmentosa type 23 X-linked	OFD1
sCe3/0232	Retinitis pigmentosa type 25	EYS
sCe3/0233	Retinitis pigmentosa type 26, autosomal recessive	CERKL
sCe3/0234	Retinitis pigmentosa type 27, autosomal dominant	NRL
sCe3/0235	Retinitis pigmentosa type 28, autosomal recessive	FAM161A
sCe3/0236	Retinitis pigmentosa type 3 X-linked	RPGR
sCe3/0237	Retinitis pigmentosa type 30, autosomal dominant	FSCN2
sCe3/0238	Retinitis pigmentosa type 31, autosomal dominant	TOPORS
sCe3/0239	Retinitis pigmentosa type 33, autosomal dominant	SNRNP200
sCe3/0240	Retinitis pigmentosa type 35, autosomal dominant/recessive	SEMA4A
sCe3/0241	Retinitis pigmentosa type 36, autosomal recessive	PRCD
sCe3/0242	Retinitis pigmentosa type 37, autosomal dominant/recessive	NR2E3
sCe3/0243	Retinitis pigmentosa type 38, autosomal recessive	MERTK
sCe3/0244	Retinitis pigmentosa type 39	USH2A
sCe3/0245	Retinitis pigmentosa type 4, autosomal dominant/recessive	RHO
sCe3/0246	Retinitis pigmentosa type 40, autosomal recessive	PDE6B
sCe3/0247	Retinitis pigmentosa type 41, autosomal recessive	PROM1
sCe3/0248	Retinitis pigmentosa type 42, autosomal dominant	KLHL7
sCe3/0249	Retinitis pigmentosa type 43, autosomal recessive	PDE6A
sCe3/0250	Retinitis pigmentosa type 44, autosomal dominant/recessive	RGR
sCe3/0251	Retinitis pigmentosa type 45, autosomal recessive	CNGB1
sCe3/0252	Retinitis pigmentosa type 46, autosomal recessive	IDH3B
sCe3/0253	Retinitis pigmentosa type 47, autosomal recessive	SAG
sCe3/0254	Retinitis pigmentosa type 48, autosomal dominant	GUCA1B
sCe3/0255	Retinitis pigmentosa type 49, autosomal recessive	CNGA1

sCe3/0256	Retinitis pigmentosa type 50, autosomal dominant	BEST1
sCe3/0257	Retinitis pigmentosa type 51, autosomal recessive	TTC8
sCe3/0258	Retinitis pigmentosa type 53, autosomal recessive	RDH12
sCe3/0259	Retinitis pigmentosa type 54, autosomal recessive	C2ORF71
sCe3/0260	Retinitis pigmentosa type 55, autosomal recessive	ARL6
sCe3/0261	Retinitis pigmentosa type 56, autosomal recessive	IMPG2
sCe3/0262	Retinitis pigmentosa type 57, autosomal recessive	PDE6G
sCe3/0263	Retinitis pigmentosa type 58, autosomal recessive	ZNF513
sCe3/0264	Retinitis pigmentosa type 59, autosomal recessive	DHDDS
sCe3/0265	Retinitis pigmentosa type 60	PRPF6
sCe3/0266	Retinitis pigmentosa type 61, autosomal recessive	CLRN1
sCe3/0267	Retinitis pigmentosa type 62, autosomal recessive	MAK
sCe3/0268	Retinitis pigmentosa type 64, autosomal recessive	C8ORF37
sCe3/0269	Retinitis pigmentosa type 66, autosomal recessive	RBP3
sCe3/0270	Retinitis pigmentosa type 7	ROM1
sCe3/0271	Retinitis pigmentosa type 7, autosomal dominant	PRPH2
sCe3/0272	Retinitis pigmentosa type 74, autosomal recessive	BBS2
sCe3/0273	Retinitis pigmentosa type 9, autosomal dominant	RP9
sCe3/0274	Retinitis pigmentosa, juvenile, autosomal recessive	SPATA7
sCe3/0275	Retinitis punctata albescens	RHO
sCe3/0276	Retinoschisis	RS1
sCe3/0277	Revesz syndrome	TINF2
sCe3/0278	Ring dermoid of cornea	PITX2
sCe3/0279	Senior-Loken syndrome type 5	IQCB1
sCe3/0280	Sorsby fundus dystrophy	TIMP3
sCe3/0281	Stargardt Disease type 1	ABCA4
sCe3/0282	Stargardt Disease type 1	CNGB3
sCe3/0283	Stargardt Disease type 3	ELOVL4
sCe3/0284	Stargardt Disease type 4	PROM1

sCe3/0285	Stickler syndrome type 1	COL2A1
sCe3/0286	Stickler syndrome type 1, nonsyndromic ocular	COL2A1
sCe3/0287	Stickler syndrome type 2	COL11A1
sCe3/0288	Stickler syndrome type 3	COL11A2
sCe3/0289	Stickler syndrome type 5	COL9A2
sCe3/0290	Stickler syndrome, autosomal recessive	COL9A1
sCe3/0291	Sveinsson choreoretinal atrophy	TEAD1
sCe3/0292	Usher syndrome type 1D	CDH23
sCe3/0293	Usher syndrome type 1D/F	PCDH15
sCe3/0294	Usher syndrome type 1G	USH1G
sCe3/0295	Usher syndrome type 1J	CIB2
sCe3/0296	Usher syndrome type 2C	PDZD7
sCe3/0297	Usher syndrome type 2C	ADGRV1
sCe3/0298	Usher syndrome type 3A	CLRN1
sCe3/0299	Vitreoretinochoroidopathy	BEST1
sCe3/0300	Waardenburg syndrome/albinism	MITF
sCe3/0301	Waardenburg syndrome/albinism	TYR
sCe3/0302	Wagner syndrome	VCAN
sCe3/0303	Warburg micro syndrome 3	RAB18
sCe3/0304	Warburg micro syndrome type 1	RAB3GAP1
sCe3/0305	Weill-Marchesani syndrome – AR	ADAMTS10
sCe3/0306	Weill-Marchesani syndrome, dominant type 2	FBN1
sCe3/0307	Wolfram syndrome type 1	WFS1
sCe3/0308	Wolfram syndrome type 2	CISD2
sCe3/0309	Wolfram-like syndrome, autosomal dominant	WFS1

Panel

Code	product name	TAT-day
	Clinical Exome Whole exome Solo	
pCe3/0001	Achromatopsia panel	25
pCe3/0002	Albinism panel	25
pCe3/0003	Cataract panel	25
pCe3/0004	Cone-rod and cone dystrophy panel	25
pCe3/0005	Flecked retina panel	25
pCe3/0006	Glaucoma panel	25
pCe3/0007	Hermansky-Pudlak syndrome panel	25
pCe3/0008	Leber congenital amaurosis panel	25
pCe3/0009	Leber optic atrophy panel	25
pCe3/0010	Microphthalmia/anophthalmia/coloboma spectrum panel	25
pCe3/0011	Oculomotor apraxia panel	25
pCe3/0012	Ophthalmoplegia (progressive external) panel	25
pCe3/0013	Optic atrophy panel	25
pCe3/0014	Retinitis pigmentosa panel, autosomal dominant	25
pCe3/0015	Retinitis pigmentosa panel, autosomal recessive	25
pCe3/0016	Stargardt disease panel	25
pCe3/0017	Stickler syndrome panel	25
pCe3/0018	Usher syndrome panel	25
pCe3/0019	Vitreoretinopathy and Wagner syndrome panel	25