Gene	Location	Encoded Protein and Function	Mendelian disease	Mode of inheritance
Genes associated with rare monogenic retinal diseases				
CDHR1[a,c]	10q23.1	Cadherin-related family member 1 (maintains rod and cone outer segment structure)	Cone-rod dystrophy; macular dystrophy	Recessive
CERKL[a]	2q31.3	Ceramide kinase-like protein (role in viability of cells with membranes rich in sphingolipids)	Rod-cone dystrophy; cone-rod dystrophy; widespread retinal degeneration	Recessive
GNAT2[a]	1p13.3	Cone-specific transducin (part of the phototransduction cascade in cone photoreceptors)	Achromatopsia (non-functioning cone photoreceptors)	Recessive
GRK1[a]	13q34	G-protein receptor kinase 1, rhodopsin kinase (phosphorylates activated rhodopsin to permit shut-off of the light response)	Oguchi disease, retinitis pigmentosa	Recessive
IMPG1[a]	6q14.1	Interphotoreceptor matrix proteoglycan 1 (role in retinal cell adhesion/ interphotoreceptor matrix)	Retinitis pigmentosa; vitelliform macular dystrophy	Dominant and recessive
IMPG2[a]	3q12.3	Interphotoreceptor matrix proteoglycan 2 (role in retinal cell adhesion/ interphotoreceptor matrix)	Retinitis pigmentosa; vitelliform macular dystrophy	Dominant and recessive
JAG1[a]	20p12.2	Jagged protein 1 (ligand for Notch proteins; function in cell-cell signalling)	Alagille syndrome	Dominant
PRPH2[a]	6p21.1	Peripherin-2 (important for rod and cone outer segment disc structure)	Macular pattern dystrophy; retinitis pigmentosa	Dominant and recessive

RAX2[a,c]	19p13.3	Retina and anterior neural fold homeobox 2 transcription factor	Retinitis pigmentosa; cone-rod dystrophy	Dominant and recessive
RBP3[a]	10q11.22	Retinol binding protein 3 (role in retinoid recycling)	Retinal dystrophy associated with high myopia	Recessive
RDH5[a,b,d]	14q24.1	Retinol dehydrogenase 5 (role in retinoid recycling)	Fundus albipunctatus, retinitis pigmentosa	Recessive
RHO[a]	3q22.1	Rhodopsin (forms visual pigment in rods)	Retinitis pigmentosa	Dominant and recessive
RLBP1[a]	15q26.1	Retinaldehyde-binding protein 1 (role in retinoid recycling)	Bothnia dystrophy, retinitis pigmentosa	Recessive
RP1L1[a]	15q26.1	Retinitis pigmentosa 1-like protein 1 (role in photoreceptor connecting cilia)	Occult macular dystrophy; retinitis pigmentosa	Dominant and recessive
SAG[a]	2q37.1	S-antigen, arrestin (binds activated rhodopsin to permit shut-off of the light response)	Oguchi disease, retinitis pigmentosa	Recessive and dominant
TULP1[a]	6p21.31	Tubby-like protein 1 (role in rhodopsin transport within photoreceptors)	Retinitis pigmentosa; Leber congenital amaurosis	Recessive
CFH[d]	1q31.3	Complement factor H (regulates alternative complement pathway)	Early onset macular drusen	Dominant
MERTK[d]	2q13	c-mer protooncogene receptor tyrosine kinase (loss of function leads to defective phagocytosis of photoreceptor outer segments by RPE)	Retinitis pigmentosa; early onset retinal dystrophy	Recessive

ABCA4[e]	1p22.1	ATP-binding cassette transporter A4 (functions as a flippase for N-retinylidene-phosphatidylenthanolamine; aids clearing of all-trans retinaldehyde)	Stargardt disease macular dystrophy; cone-rod dystrophy	Recessive
NR2E3[e]	15q23	Nuclear receptor subfamily 2 group E3 (transcription factor, role in rod photoreceptor development; loss of function leads to excess of S-cones)	Enhanced S-cone syndrome, Goldmann-Faver syndrome, retinitis pigmentosa	Recessive and dominant
MYO7A[e]	11q13.5	Myosin VIIA (motor protein functions in transport of opsin in photoreceptors and melanosomes in RPE)	Type 1 Usher syndrome (retinitis pigmentosa and deafness)	Recessive
Genes associated with oculocutaneous albinism (and foveal hypoplasia)				
OCA2[a,e]	15q12-15q13.1	Oculocutaneous albinism 2 melanosomal transmembrane protein (protein regulates pH of melanosomes)	Oculocutaneous albinism	Recessive
TYR[a]	11q14.3	Tyrosinase (role in melanin synthesis)	Oculocutaneous albinism	Recessive
Genes associated with rare monogenic ocular maldevelopment (including microphthalmia)				
VSX2[a,b]	14q24.3	Visual system homeobox 2 (role in eye development)	Microphthalmia	Recessive
PITX2[e]	4q25	Paired-like homeodomain transcription factor 2 (role in eye development)	Anterior segment dysgenesis; microphthalmia	Dominant
ATOH7[a]	10q21.3	Atonal basic helix-loop-helix transcription factor 7 (role in eye development)	Persistent primary hypoplastic vitreous; ocular maldevelopment, including microphthalmia	Recessive

CHD7[a]	8q12.2	Chromodomain helicase DNA-binding protein 7 (role in control of neurocrest gene expression and in regulation of cel motility)	CHARGE syndrome	Dominant
RARB[a]	3p24.2	Retinoic acid receptor beta (role in eye and other organ development)	Microphthalmia, syndromic	Dominant and recessive
RAX[a]	18q21.32	Retina and anterior neural fold homeobox (role in eye development)	Microphthalmia	Recessive