

Gene	[Chromosome:Position]	[REF/ALT]	rs#	Exonic function	Cyto band	Reference sequence	Sequence variant (hg19)	Predicted protein change	Region	gnomAD	CADD phred score	VEST3 score	Meta LR score	Family ID	Proband call	Father call	Mother call	Origin	Phenotype
AGRN	1:985283	G/C		nonsynonymous SNV	1p36.33	NM_198576	c.G4745C	p.G1582A	exon27	0	23.7	0.76	0.98	#42	G/C	G/C	G/C	Maternal	Unilateral microphthalmia, isolated
RYR2	1:125812	G/C		nonsynonymous SNV	1p36.33	NM_02156462	c.T214C	p.Y81H	exon4	NA	27.1	0.951	0.207	#2	A/G	A/A	A/A	Maternal	Unilateral microphthalmia, isolated
TAS1R3	1:126894	T/C		nonsynonymous SNV	1p36.33	NM_152228	c.T1709C	p.L570P	exon6	NA	22.9	0.745	0.79	#50	T/C	T/C	T/T	Paternal	Bilateral microphthalmia, isolated
EPB41	1:2936587	C/G		nonsynonymous SNV	1p35.3	NM_203343	c.C1480G	p.P494A	exon10	NA	27.4	0.735	0.818	#34	C/G	C/G	C/C	Paternal	Unilateral microphthalmia, isolated
CEB2	1:150939675	A/G		nonsynonymous SNV	1q21.3	NM_020275	c.T616C	p.F26L	exon8	NA	28.5	0.862	0.771	#30	A/G	A/A	A/A	Maternal	Unilateral microphthalmia, isolated
RC3H1	1:173962102	A/G		nonsynonymous SNV	1q25.1	NM_001300850	c.T22C	p.W8R	exon1	NA	26.4	0.991	0.896	#42	A/G	A/A	A/A	Paternal	Unilateral microphthalmia, isolated
IARS	1:220279347	C/T		nonsynonymous SNV	1q41	NM_018060	c.C1181T	p.A394V	exon9	NA	34	0.789	0.758	#68	C/T	C/C	C/T	Maternal	Unilateral microphthalmia, isolated
MAP3K21	1:233464192	C/T		nonsynonymous SNV	1q42.2	NM_032435	c.C418T	p.R140C	exon1	NA	33	0.755	0.779	#28	C/T	C/C	C/T	Maternal	Unilateral microphthalmia with accompanying malformations
TBC1	1:235564818	G/C		nonsynonymous SNV	1q42.3	NM_001079515	c.G101C	p.G34A	exon3	NA	26.4	0.744	0.757	#33	G/C	G/G	G/C	Maternal	Bilateral microphthalmia with accompanying malformations
RYR2	1:237608723	A/G		nonsynonymous SNV	1q43	NM_0010035	c.A1193G	p.H398R	exon14	NA	21.6	0.905	0.74	#20	A/G	A/G	NA	Unknown	Unilateral microphthalmia with accompanying malformations
JPH1	1:238048119	C/T		nonsynonymous SNV	1q43	NM_021186	c.G1304A	p.C447Y	exon10	NA	27.8	0.833	0.863	#54	C/T	NA	NA	C/T	Unknown
FMNL2	1:25343446	G/T	rs75741476	nonsynonymous SNV	2q32.3	NM_025205	c.G750T	p.E250D	exon8	0	24.6	0.798	0.717	#2	G/T	G/G	G/T	Maternal	Unilateral microphthalmia, isolated
OSGEPL1	1:290617696	C/A		nonsynonymous SNV	2q32.2	NM_001354347	c.G1060T	p.G336C	exon6	0	34	0.985	0.903	#21	C/A	C/C	C/A	Maternal	Bilateral microphthalmia with accompanying malformations
AQX1	2:201533443	C/T		nonsynonymous SNV	2q33.1	NM_001159	c.C3715T	p.P1239S	exon33	NA	33	0.947	0.722	#27	C/T	C/T	C/C	Paternal	Unilateral microphthalmia, isolated
PRKAG3	2:219689275	C/A		nonsynonymous SNV	2q35	NM_017431	c.G1193T	p.R398L	exon11	NA	35	0.909	0.83	#14	C/A	C/C	C/A	Maternal	Unilateral microphthalmia, isolated
GMPPA	2:220368850	A/T		nonsynonymous SNV	2q35	NM_013335	c.A535T	p.I179F	exon7	NA	28.4	0.95	0.939	#14	A/T	A/A	A/A	Maternal	Unilateral microphthalmia, isolated
SNM1A	3:4458913	C/T		nonsynonymous SNV	3p26.1	NM_001164674	c.G664A	p.G222S	exon5	NA	35	0.971	0.975	#53	C/T	C/C	C/T	Maternal	Unilateral microphthalmia with accompanying malformations
SCN11A	3:38908840	A/G		nonsynonymous SNV	3q22.2	NM_014139	c.T3923C	p.I1308T	exon23	NA	28.6	0.82	0.961	#10	A/G	A/A	A/A	Maternal	Unilateral microphthalmia, isolated
CACNA2D3	3:55002482	G/A		nonsynonymous SNV	3p14.3	NM_018398	c.G2455A	p.G819S	exon28	NA	25.1	0.945	0.714	#55	G/A	G/G	G/A	Maternal	Unilateral microphthalmia, isolated
P2RY24	3:150931230	G/C		nonsynonymous SNV	3q25.1	NM_001081455	c.C875G	p.P292R	exon3	NA	26.4	0.978	0.975	#6	G/C	G/G	G/C	Maternal	Unilateral microphthalmia, isolated
MCC1	3:182751828	T/G		nonsynonymous SNV	3q27.1	NM_001293273	c.A1281C	p.R427S	exon12	NA	27.3	0.95	0.876	#6	T/G	T/G	T/T	Paternal	Unilateral microphthalmia, isolated
UPH1	3:185241924	C/G		nonsynonymous SNV	3q27.2	NM_139248	c.G653C	p.G218A	exon5	NA	26.3	0.908	0.904	#7	C/G	C/G	C/C	Paternal	Unilateral microphthalmia, isolated
PDE6B	4:628611	A/G		nonsynonymous SNV	4p16.3	NM_000283	c.A614G	p.D205P	exon2	NA	24.5	0.927	0.718	#53	A/G	A/G	A/A	Paternal	Bilateral microphthalmia with accompanying malformations
FRAS1	4:79238624	G/A		nonsynonymous SNV	4q21.21	NM_001166133	c.G1922A	p.C641Y	exon17	NA	25.2	0.993	0.724	#34	G/A	G/G	G/A	Maternal	Unilateral microphthalmia, isolated
PRKG2	4:82096091	C/A		nonsynonymous SNV	4q21.21	NM_001282485	c.G484T	p.A162S	exon3	NA	32	0.713	0.761	#33	C/A	C/C	C/A	Maternal	Bilateral microphthalmia with accompanying malformations
USP38	4:144135410	C/A		nonsynonymous SNV	4q31.21	NM_001290325	c.C2281A	p.L761I	exon9	NA	27.5	0.75	0.856	#60	C/A	C/A	C/C	Paternal	Unilateral microphthalmia with accompanying malformations
TRIM2	4:154191643	C/A		nonsynonymous SNV	4q31.3	NM_001130067	c.C106A	p.L36I	exon2	NA	31	0.706	0.73	#62	C/A	C/C	C/A	Maternal	Unilateral anophthalmia with accompanying malformations
PALLD	4:169630200	T/G		nonsynonymous SNV	4q32.3	NM_001166109	c.T377G	p.I126S	exon8	NA	25.7	0.983	0.814	#1	T/G	NA	T/T	Unknown	Unilateral microphthalmia, isolated
CBR4	4:169923345	C/A		nonsynonymous SNV	4q32.3	NM_032783	c.G412T	p.G138C	exon4	NA	33	0.899	0.927	#53	C/A	C/C	C/C	De novo	Bilateral microphthalmia with accompanying malformations
MCF30	5:126732827	G/A		nonsynonymous SNV	5q23.2	NM_00125645	c.G830A	p.C277Y	exon8	NA	27.7	0.998	0.701	#40	G/A	G/A	G/G	Paternal	Unilateral microphthalmia, isolated
GRM6	5:178413258	G/C		nonsynonymous SNV	5q35.3	NM_000843	c.C1979G	p.S666C	exon9	NA	25.7	0.901	0.872	#11	G/C	G/G	G/C	Paternal	Unilateral microphthalmia, isolated
NHLRC1	6:18121930	A/G		nonsynonymous SNV	6p22.3	NM_198586	c.T908C	p.L303P	exon1	NA	24.6	0.894	0.919	#41	A/G	A/G	A/A	Paternal	Unilateral microphthalmia, isolated
CAPN11	6:44147870	G/C		nonsynonymous SNV	6p22.1	NM_007058	c.G610C	p.R577P	exon14	NA	31	0.989	0.892	#2	A/G	A/G	A/A	Paternal	Unilateral microphthalmia, isolated
MAIAN1	6:119501094	A/G		nonsynonymous SNV	6q22.31	NM_005907	c.T1852C	p.F618L	exon13	NA	32	0.968	0.754	#53	A/G	A/A	A/A	Maternal	Unilateral microphthalmia with accompanying malformations
ANG1	6:131802421	T/G		nonsynonymous SNV	6q22.2	NM_000045	p.V123G		exon4	NA	28.6	0.864	0.962	#29	T/G	T/T	T/G	Maternal	Unilateral microphthalmia, isolated
LRP11	6:150174143	A/C		nonsynonymous SNV	6q25.1	NM_032832	c.T767G	p.M256R	exon2	NA	23.8	0.846	0.901	#21	A/C	A/A	A/C	Maternal	Bilateral microphthalmia with accompanying malformations
THSD7A	7:11457141	C/T	rs76166757	nonsynonymous SNV	7p21.3	NM_015204	c.G3473A	p.C1158Y	exon16	NA	32	0.766	0.755	#67	C/T	T/T	C/T	Paternal	Bilateral microphthalmia, isolated
GCK	7:44187297	T/G		nonsynonymous SNV	7p13	NM_000162	c.A815S	p.E272A	exon7	NA	24.8	0.881	0.907	#67	T/G	T/G	T/T	Paternal	Bilateral microphthalmia, isolated
ABCA13	7:48626814	A/G		nonsynonymous SNV	7p12.3	NM_152701	c.A14570G	p.Y4857C	exon57	NA	29.1	0.958	0.871	#39	A/G	A/G	A/A	Paternal	Unilateral microphthalmia, isolated
CFTR	7:117304758	T/G		nonsynonymous SNV	7q31.2	NM_000492	c.T3989G	p.V1327G	exon25	NA	23.8	0.809	0.771	#17	T/G	T/G	T/T	Paternal	Bilateral microphthalmia, isolated
GRM8	7:126173652	A/T		nonsynonymous SNV	7q31.33	NM_001171087	c.T1159A	p.I990N	exon8	NA	27.4	0.844	0.828	#19	A/T	A/T	A/A	Paternal	Unilateral microphthalmia, isolated
PRKAG2	7:151262820	A/G		nonsynonymous SNV	7q36.1	NM_024429	c.T662C	p.V221A	exon8	NA	32	0.862	0.976	#17	A/G	A/A	A/A	Maternal	Bilateral microphthalmia, isolated
FGF20	8:16859328	C/G		nonsynonymous SNV	8p22	NM_019851	c.C214G	p.R72G	exon1	NA	34	0.949	0.727	#6	G/C	G/G	G/C	Maternal	Unilateral microphthalmia, isolated
ST18	8:53079484	A/G		nonsynonymous SNV	8q11.23	NM_001352848	c.C1132T	p.H378R	exon7	NA	28.2	0.944	0.809	#2	G/A	G/G	G/A	Maternal	Unilateral microphthalmia, isolated
CHD7	8:61734662	G/A		nonsynonymous SNV	8q12.2	NM_017780	c.A2915G	p.Q972R	exon11	NA	25.6	0.973	0.996	#60	A/G	A/A	A/A	De novo	Unilateral microphthalmia with accompanying malformations
BHLHE22	8:65493297	T/C		nonsynonymous SNV	8q12.3	NM_154144	c.T50C	p.L17P	exon1	NA	23	0.769	0.886	#68	T/C	T/T	T/C	Maternal	Unilateral microphthalmia, isolated
PKHD1L1	8:110493680	G/A		nonsynonymous SNV	8q23.1	NM_177531	c.G9346A	p.G3116S	exon56	NA	34	0.869	0.949	#21	G/A	G/G	G/C	Maternal	Bilateral microphthalmia with accompanying malformations
WASHC5	8:126068971	C/A		nonsynonymous SNV	8q24.13	NM_001330609	c.G1520T	p.R507L	exon15	NA	35	0.94	0.809	#62	C/A	C/A	C/C	Paternal	Unilateral anophthalmia with accompanying malformations
CCM4	8:134237720	G/T		nonsynonymous SNV	8q24.22	NM_080838	c.G437T	p.G146V	exon3	NA	31	0.983	0.856	#8	G/T	G/T	G/G	Paternal	Unilateral microphthalmia, isolated
KIF2	8:145698370	C/T		nonsynonymous SNV	8q24.3	NM_001369769	c.C2054T	p.S685L	exon17	NA	33	0.883	0.863	#55	C/T	C/C	C/T	Maternal	Unilateral microphthalmia, isolated
KIAA2026	9:5968445	C/T		nonsynonymous SNV	9p24.1	NM_001017969	c.G1786A	p.G599R	exon3	NA	20.7	0.763	0.829	#26	C/T	C/T	C/C	Paternal	Unilateral microphthalmia with accompanying malformations
ABCA1	9:107560781	A/C		nonsynonymous SNV	9q31.1	NM_005502	c.T5042G	p.V1681G	exon37	NA	23.5	0.915	0.779	#16	A/C	A/C	A/A	Paternal	Unilateral microphthalmia, isolated
MSRB2	10:23409702	G/A		nonsynonymous SNV	10p12.2	NM_012228	c.G460A	p.G154S	exon5	NA	34	0.972	0.948	#34	G/A	G/G	G/A	Maternal	Unilateral microphthalmia, isolated
HCTD2	10:93272038	C/G		nonsynonymous SNV	10q23.32	NM_001348365	c.C1907G	p.T636S	exon21	NA	28.9	0.801	0.788	#64	C/G	C/G	C/C	Paternal	Unilateral microphthalmia, isolated
MFSO23A	10:104228908	T/G		nonsynonymous SNV	10q24.32	NM_024789	c.T122G	p.F41C	exon3	NA	28.1	0.964	0.748	#47	T/G	T/G	T/T	Paternal	Unilateral microphthalmia, isolated
VTG14	10:114229574	G/T		nonsynonymous SNV	10q25.2	NM_001218205	c.G557T	p.R186E	exon7	NA	25.4	0.932	0.702	#15	G/T	G/G	G/G	De novo	Bilateral microphthalmia, isolated
CLU2D1	10:124553376	A/C		nonsynonymous SNV	10q26.13	NM_022334	c.C1143G	p.L488W	exon8	NA	28	0.836	0.753	#23	A/C	A/A	A/C	Maternal	Bilateral microphthalmia, isolated
PAX6	11:31824340	C/T		nonsynonymous SNV	11p13	NM_001389111	c.G56A	p.G19E	exon2	NA	26.4	0.787	0.991	#59	C/T	C/C	C/C	Maternal	Bilateral microphthalmia, isolated
MYBPC3	11:47353698	C/G		nonsynonymous SNV	11p11.2	NM_000256	c.G379C	p.D1247H	exon32	NA	26.6	0.979	0.732	#28	C/G	C/C	C/G	Maternal	Unilateral microphthalmia with accompanying malformations
TYR	11:88924544	A/T		nonsynonymous SNV	11q14.3	NM_000972	c.A994T	p.M332L	exon2	NA	24.8	0.877	0.947	#24					