

Supplemental Table S1a. Nonsense variants (stopgain) in probands with anophthalmia/microphthalmia identified by exome sequencing

Gene	[Chromosome: Position]	[REF/ALT]	rs#	Exonic function	Cyto band	Reference sequence	Sequence variant (hg19)	Predicted protein change	Region	gnomAD	pLI score	Family ID	Proband call	Father call	Mother call	Origin	Sanger sequencing results summary	Phenotype
VPS13D a	1:12557600	C/T		stopgain	1p36.22	NM_018156	c.C12634T	p.Q4212X	exon67	NA	1.0000	#46	C/T	C/C	C/C	De novo	Proband result confirmed; Parents were not tested due to insufficient amount of DNA	Unilateral microphthalmia with accompanying malformations
<i>XKR4</i>	8:56015062	C/G		stopgain	8q12.1	NM_052898	c.C14G	p.S5X	exon1	NA	0.9776	#54	C/G	NA	C/C	Unknown	Trios were NOT tested due to no remaining DNA for proband	Unilateral microphthalmia, isolated
CHD7 b	8:61735063	C/T		stopgain	8q12.2	NM_017780	c.C2959T	p.R987X	exon12	NA	1.0000	#55	C/T	C/C	C/C	De novo	Trio results confirmed	Unilateral microphthalmia, isolated
<i>CHD7</i>	8:61750761	C/T	rs587783442	stopgain	8q12.2	NM_017780	c.C4480T	p.R1494X	exon19	NA	1.0000	#20	C/T	C/C	NA	Unknown	Proband result confirmed; Parents were not tested due to insufficient amount of DNA	Unilateral microphthalmia with accompanying malformations
<i>LRP5</i>	11:68191161	C/T		stopgain	11q13.2	NM_001291902	c.C1489T	p.R497X	exon14	NA	0.9984	#17	C/T	C/T	C/C	Paternal	Proband failed PCR; Parent results confirmed	Bilateral microphthalmia, isolated
<i>NUMA1</i>	11:71726895	G/A		stopgain	11q13.4	NM_006185	c.C1654T	p.Q552X	exon15	NA	1.0000	#45	G/A	G/A	G/G	Paternal	Proband result confirmed; Parents were not tested due to insufficient amount of DNA	Bilateral microphthalmia, isolated
<i>TUBGCP3</i>	13:113181728	G/T		stopgain	13q34	NM_001286277	c.C1376A	p.S459X	exon12	NA	0.9525	#16	G/T	G/T	G/G	Paternal	Proband result confirmed; Parents were not tested due to insufficient amount of DNA	Unilateral microphthalmia, isolated
<i>FAM192A</i>	16:57206739	G/A		stopgain	16q13	NM_001354078	c.C175T	p.Q59X	exon3	NA	0.9441	#64	G/A	G/G	G/A	Maternal	Proband result confirmed; parents were not tested due to insufficient amount of DNA	Unilateral microphthalmia, isolated
<i>KRT31</i>	17:39553461	C/A		stopgain	17q21.2	NM_002277	c.G331T	p.E111X	exon1	NA	0.9285	#61	C/A	C/C	C/A	Maternal	Trios were NOT tested due to insufficient amount of DNA for all samples	Unilateral microphthalmia, isolated

Abbreviations: ALT, alternate; A/M, anophthalmia/microphthalmia; gnomAD, Genome Aggregation Database; hg19: human genome 19; NA: not available; NM: mRNA; pLI, probability of being loss-of-function intolerant; REF, reference.

a de novo not confirmed by Sanger sequencing (proband result was confirmed, but parent samples were not available)

b de novo confirmed by Sanger sequencing