Supplementary Table 2: Pathogenicity data for genetic variants that account for or are likely to account for disease in this cohort, displayed by variant subtype.

Insertion / Deletion Variants	Mutation Taster			SIFT INDEL				VEST INDEL		ExAC		Literature		
Nucleotide/ protein changes	Assignmental Score	PhyloP/ PhastCons	Causes NMD	Effect	Confidence score	Causes NMD	(%) Indel Location	Score	p Value	MAF	Homozygotes / Allele number	Occurrence	Functional studies	Comments
AIPL1 **(NM_014336.3)**														
c.356_359del p. His119Argfs*31	P 1.00	4.006; 4.835/ 1.000	Y	Dam	0.858	Y	31	P 0.848	0.001	No Data		-	-	**Novel**
CEP290 **(NM_025114.3)**														
c.3175dup p. Ile1059Asnfs*11	P 1.00		Y	Dam	0.858	Y	43	P 0.745	0.001	0.0003	0 / 113,552	[1-3]	-	-
c.3181_3182del p. Met1061Alafs*8	P 1.00	3.314; 3.237/ 1.000	Y	Dam	0.858	Y	43	P 0.787	0.001	No Data		[4]	-	-
c.4625_4626insCATG p. Ala1543Metfs*37	P 1.00		Y	Dam	0.858	Y	62	P 0.841	0.001	No Data		-	-	**Novel**
CRB1 **(NM_201253.2)**														
c.613_619del p. Ile205Aspfs*13	P 1.00		Y	Dam	0.858	Y	14	P 0.756	0.000	No Data		[5-7]	-	-
c.1793del p. Pro598Hisfs*23	P 1.00	0.999/ 0.098	Y	Dam	0.858	Y	42	P 0.861	0.000	No Data		-	-	**Novel**
GUCY2D **(NM_000180.3)**														
c.91dup p. Arg31Profs*288	P 1.00		Y	Dam	0.579	Y	3	B 0.214	0.372	No Data		[8]	-	-
c.2516del p. Thr839Argfs*27	P 1.00	5.69/ 1.000	Y	Dam	0.858	Y	76	P 0.731	0.001	No Data		[9]	-	-
c.2595del p. Lys866Argfs*14	P 1.00	1.564/ 1.000	Y	Dam	0.858	Y	78	P 0.913	0.000	No Data		[4, 10]	-	-

***LCA5* (NM_001122769.2)**													
c.1144_1147dup p. Asn383Thrfs*15	P 1.00		Pos	Dam	0.858	N	55	B 0.167	0.074	No Data	-	-	**Novel**
***NMNAT1* (NM_022787.3)**													
c.364del p. Arg122Glyfs*20	P 1.00	2.822/ 0.613	Pos	Dam	0.858	N	43	P 0.666	0.003	No Data	[11]	-	-
***RPE65* (NM_000329.2)**													
c.951_956del p. Tyr318_Glu319del	pm 1.00	1.000	N	Dam	0.894	N	59	P 1.000	0.000	No Data	-	-	**Novel**
***RPGRIP1* (NM_020366.3)**													
c.exon19del; chr14: g.(21798302_21798377) _(21798551_21799045) del (hg19,NC000014.8)	P 1.00		Y	No Data				No Data		No Data	-	-	**Novel;** Exon 17 deletion considered pathogenic for LCA [12]
***TULP1* (NM_003322.3)**													
c.524dup p. Pro176Thrfs*7	P 1.00		Y	Dam	0.858	Y	32	B 0.418	0.052 borde rline	No Data	-	-	**Novel**

Nonsense Variants	Mutation Taster					ExAC		Literature		
Nucleotide/ protein changes	Assignment and Score	PhyloP/ phastCons	Causes NMD	Position (AA) of stop codon		MAF	Homozygotes / Alelle number	Occurrence	Functional studies	Comments
				wt	mut					
AIPL1 **(NM_014336.3)**										
c.834G>A p. Trp278*	P 1.00	4.931/ 1.000	?	385	278	0.0004	0 119,090	[13-15]	[16]	-
CEP290 **(NM_025114.3)**										
c.1781T>A p. Leu594*	P 1.00	0.447/ 0.468	Y	2480	594	0.0001	0 27,218	[4]	-	-
c.2991+1655A>G[#] p. Cys998*	pm 1.00	0.983/ 0.109	Y	-	-	No Data		[17]	-	-
GUCY2D **(NM_000180.3)**										
c.2646C>G p. Tyr882*	P 1.00	0.754/ 1.000	Y	1104	882	No Data		-	-	**Novel**
NMNAT1 **(NM_022787.3)**										
c.507G>A p. Trp169*	P 1.00	6.004/ 1.000	?	280	169	0.0000	0 121,352	[11, 18, 19]	-	-

RDH12 **(NM_152443.2)**										
c.316C>T p. Arg106*	P 1.00	2.018/ 1.000	Y	317	106	0.0000	0 121,380	[20]	-	-
RPE65 **(NM_000329.2)**										
c.130C>T p. Arg44*	P 1.00	2.687/ 1.000	Y	534	44	0.0000	0 121,406	-	[21, 22]	-
RPGRIP1 **(NM_020366.3)**										
c.1219C>T p. Gln407*	P 1.00	1.128/ 0.598	Y	1287	407	0.0000	0 49,320	-	-	+1763-8C>G-NMD
c.1447C>T p. Gln483*	P 1.00	0.451/ 0.000	Y	1287	483	0.0000	0 119,622	[23]	-	(Referenced paper only found one heterozygous variant in RPGRIP1)
c.2935C>T p. Gln979*	P 1.00	1.473/ 0.016	Y	1287	979	No Data		-	-	**Novel**
SPATA7 **(NM_018418.4)**										
c.763C>T p. Gln255*	P 1.000	2.614/ 1.000	Y	600	255	No Data		[24]	[25]	-
TULP1 **(NM_003322.3)**										
c.1081C>T p. Arg361*	P 1.000	1.101/ 0.999	Y	543	361	No Data		[26, 27]	-	-

Missense Variants	Mutation Taster			Align GVGD		PolyPhen2		SIFT			ExAC		Literature		Comments
Nucleotide / protein changes	**Assignment and Score**	**PhyloP/ phastCons**	**Grantham Score**	**Class**	**GV/GD**	**Hum Div**	**Hum Var**	**Pred**	**Score**	**Med Seq Cons**	**MAF**	**Homozygotes / Allelle number**	**Occurrence**	**Functional studies**	
CRB1 **(NM_201253.2)**															
c.2843G>A p. Cys948Tyr	P 1.00	5.31/ 1.00	194	C15	138.77 112.21	PD 1.00	PD 0.92	Del	0.04	2.69	0.0002	0 119,820	[4, 6, 28-36]	-	-
GUCY2D **(NM_000180.3)**															
c.307G>A p. Glu103Lys	pm 0.65	2.78/ 0.74	56	C0	240.48 0.00	PD 1.00	PD 0.99	Tol	0.29	3.22	No Data		[9, 37-39]		-
c.2302C>T p. Arg768Trp	P 1.00	3.75/ 1.00	101	C0	241.31 94.79	PD 1.00	PD 1.00	Del	0.00	3.22	0.0001	0 121,264	[8, 40-44]	[45, 46]	-
c.2345T>A p. Leu782His	pm 1.00	0.87/ 1.00	99	C0	258.55 0.00	B 0.00	B 0.00	Tol	0.29	3.22	0.1303	1537 121,268	-	-	**Benign**
c.2383C>T p. Arg795Trp	P 1.00	2.35/ 0.98	101	C0	241.31 94.79	PD 1.00	PD 1.00	Del	0.00	3.22	0.0001	0 121,100	[47, 48]	-	[43, 49, 50] indicate that other variants at this codon position are considered pathogenic for LCA

NMNAT1 **(NM_022787.3)**														
c.500A>G p. Asn167Ser	P 1.00	3.45/ 1.00	46	C0	217.79 0.00	PD 0.82	B 0.30	Tol	0.08/ 9	3.12	No Data		-	-	**Novel**
c.769G>A p. Glu257Lys	P 1.00	3.87/ 1.00	56	C0	118.33 21.98	B 0.09	B 0.03	Tol	0.25/ 7	3.11	0.0006	0 120,500	[11, 18, 19, 51, 52, 53]	[19, 54]	Incomplete penetrance/ Hypo-morphic allele
RDH12 **(NM_152443.2)**														
c.697G>C p. Val233Leu	P 1.00	6.21/ 1.00	32	C0	134.16 4.86	PD 0.96	PD 0.74	Tol	0.22	2.87	0.0000	0 120,420	[20, 55]	-	-
RPE65 **(NM_000329.2)**														
c.1040G>C p. Arg347Pro	P 1.00	4.11/ 1.00	103	C15	56.64 66.83	PD 1.00	PD 0.99	Del (Low conf)	0.02	3.30	No Data But Arg347His = 0.000008295		-	-	**Novel**
RPGRIP1 **(NM_020366.3)**														
c.1639G>T p. Ala547Ser	pm 0.97	0.39/ 0.95	99	C0	244.82 0.00	PD 1.00	PD 1.00	Tol	0.59	2.80	0.2105	2911 117,052	[56-61]	-	**Benign**

Splicing Variants	Mutation Taster			Spliceman		Other nomenclature	ExAC		Literature		Comments
Nucleotide/ protein changes	Assignment and Score	PhyloP	PhastCons	L1 Distance	Ranking (%)		MAF	Homozygotes / Allele number	Occurrence	Functional Studies	
AIPL1 **(NM_014336.3)**											
c.277-2A>G	Consensus Dinucleotides					IVS2-2A>G	0.00002	0 120,944	[25, 62]	[63]	-
CEP290 **(NM_025114.3)**											
c.297+3A>G	P 1.000	2.768	1.000	33753	61	-	No Data		-	-	**Novel**
c.5587-1G>C	Consensus Dinucleotides					IVS40 as G-C-1	No Data		[25, 35, 64]	-	-
RPE65 **(NM_000329.2)**											
c.726-1G>A	Consensus Dinucleotides					-	No Data		-	-	**Novel**
TULP1 **(NM_003322.3)**											
c.999+5G>C	P 0.999	1.463	0.782	37074	78	IVS10 ds G-C +5	No Data		[65, 66]	-	-

Abbreviations: **Mutation Taster**; P = pathogenic, pm = polymorphism, NMD = nonsense-mediated decay: **SIFT INDEL**; Dam = damaging: **VEST INDEL**; P = pathogenic, B = benign: **ExAC**; MAF = minor allele frequency; **PolyPhen2**; PD = probably damaging, B = benign: **SIFT**; Del = deleterious, Tol = tolerated, Med Seq Cons = median sequence conservation.

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