

# Supplementary Table 3.

## The clinical findings and variants in patients with *NMNAT1*-associated retinopathy

Refere ence #	Published year, Authors	Patient #	Age	Gender	Macular degeneration	Optic disk atrophy	Retinal degeneration	Visual acuity	Refractive errors	Electro- retinogram	Others	Variant 1	Variant 1	Variant 2	Variant 2	Ethnicity
9	2012, Chiang et al.	2	6m	M	Yes	NA	Yes	NLP	NA	NA	nystagmus	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Brazil Caucasian (Poland,
10	2012, Perrault et al.	P7.1	59	M		invisible due to cataract		NLP	hyperopia	NA	cataract	c.542A>G	p.Tyr181Cys	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P7.2	60	M		invisible due to cataract		NLP	hyperopia	NA	cataract	c.542A>G	p.Tyr181Cys	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P15	6	M	Yes	NA	NA	NLP	+4/+5	NA	sun photophobia	c.643G>T	p.Glu215*	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P19	3	M	No	Yes	Yes	NLP	NA	NA		c.205A>G	p.Met69Val	c.650T>A	p.Ile217Asn	South Africa/France
10	2012, Perrault et al.	P22.1	18	M		invisible due to retinal detachment and cataract		NLP	NA	NA	retinal detachment and cataract	c.319G>T	p.Glu107*	c.709C>T	p.Arg237Cys	France
10	2012, Perrault et al.	P22.2	13	F		invisible due to ocular phytis and cataract		NLP	NA	NA	retinal detachment and cataract	c.319G>T	p.Glu107*	c.709C>T	p.Arg237Cys	France
10	2012, Perrault et al.	P26.2	24	NA	Yes	Yes	NA	NLP	NA	NA		c.769G>A	p.Glu257Lys	?	?	Portugal/La Leunion (France)
10	2012, Perrault et al.	P28.1	6	NA	Yes	NA	NA	NLP	NA	NA		c.769G>A	p.Glu257Lys	?	?	France
10	2012, Perrault et al.	P29	NA	NA	NA	NA	NA	NLP	NA	NA	photophobia	c.769G>A	p.Glu257Lys	?	?	La Leunion (France)
11	2012, Koenekoop et al.	MOGL624	10	M	Yes	Yes	Yes	NLP	hyperopia	NA	nystagmus	c.769G>A	p.Glu257Lys	c.769G>A	p.Glu257Lys	Saudi Arabian
12	2012, Falk et al.	CHOP/MEEI-053-II-1	20	F	Yes	Yes	Yes	NLP	NA	non-recordable	33-week's gestational age, developmental delay	c.205A>G	p.Met69Val	c.769G>A	p.Glu257Lys	African American
12	2012, Falk et al.	UCL-1-II-1	4	F	Yes	Yes	Yes	NLP	+11	non-recordable		c.205A>G	p.Met69Val	c.769G>A	p.Glu257Lys	Caucasian descent
12	2012, Falk et al.	UCL-7-II-1	36m	M	Yes	Yes	Yes	NLP	NA	non-recordable		c.466G>C	p.Gly156Arg	c.769G>A	p.Glu257Lys	British (European descent)
23	2014, Siemiatkowska et al.	4	64	M	No	No	Yes	NLP	not performed	not performed	keratoconus	c.595G>T	p.Glu199*	c.769G>A	p.Glu257Lys	Danish
26	2015, Deng et al.	II:5	10	M		not-performed		NLP	NA	not-performed		c.721C>T	p.Pro241Ser	c.721C>T	p.Pro241Ser	Chinese
28	2016, Bravo-Gil et al.	RP157-II:3	6m	M	Yes	Yes	Yes	NLP	NA	NA	malformation syndrome	c.197G>A	p.Arg66Gln	c.769G>A	p.Glu257Lys	Spanish
18	2018, Hosono et al.	EYE42	16	M	Yes	No	Yes	NLP	NA	non-recordable	nystagmus from birth	c.1A>G		C.709C>T	p.Arg237Cys	Japanese
43	2020, Sallum, et al.	64.1	4m	NA	Yes/No	Yes	Yes	NLP	NA	NA		c.-57+21C>T		c.759delGins TA	p.Leu253Phef sTer5	Brazilian
43	2020, Sallum, et al.	65.1	3m	NA	Yes/No	Yes	Yes	NLP	NA	NA		c.507G>A	p.Trp169Ter	c.769G>A	p.Glu257Lys	Brazilian
43	2020, Sallum, et al.	66.1	since birth	NA	Yes/No	Yes	Yes	NLP	NA	NA		c.293T>G	p.Val98Gly	c.769G>A	p.Glu257Lys	Brazilian
27	2015, Hedergott et al.	II.3	4w	F	Yes	Yes	Yes	NLP/NLP	+4/+4	non-recordable	nystagmus, non-recordable VEP	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	CHOP/MEEI-047-IV-6	NA	M	NA	NA	NA	congenital blindness	NA	NA	LCA without developmetal delay	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	CHOP/MEEI-047-IV-7	NA	F	NA	NA	NA	congenital blindness	NA	NA	hearing loss and developmental delay (GJB2 mutations combined)	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	CHOP/MEEI-047-IV-8	NA	M	NA	NA	NA	congenital blindness	NA	NA	hearing loss and developmental delay (GJB2 mutations combined)	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	CHOP/MEEI-047-IV-3	4m	M	Yes(3y)	mild	Yes	LP > NLP	significant myopia	non-recordable	enophthalmos, neural development delay, non-verbal at age 4, autism, normal hearing	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
26	2015, Deng et al.	II:2	24	M	Yes	NA	NA	LP/NLP	NA	non-recordable		c.721C>T	p.Pro241Ser	c.721C>T	p.Pro241Ser	Chinese
11	2012, Koenekoop et al.	LCA9 IV:8	2-30	F	Yes	Yes	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:9	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:11	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:12	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA	nystagmus, night blindness, cataract (posterior subcapsular), one has keratoconus	c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:13	2-30	M	Yes	NA	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:16	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:17	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:18	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA		c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani
17	2017, Tompson et al.	-	NA	NA	NA	NA	NA	LP-NLP	NA	NA	photophobia	c.364del		c.769G>A	p.Glu257Lys	Australian
17	2017, Tompson et al.	-	NA	NA	NA	NA	NA	LP-NLP	NA	NA	photophobia	c.364del		c.769G>A	p.Glu257Lys	Australian
17	2017, Tompson et al.	-	NA	NA	NA	NA	NA	LP-NLP	NA	NA	photophobia	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Australian
10	2012, Perrault et al.	P5	12	F	Yes	NA	NA	NLP/LP	+9.5/+7.75	NA	night blindness	c.43911G>C	Splice	c.769G>A	p.Glu257Lys	France
27	2015, Hedergott et al.	II.1	5m	F	Yes	No	Yes	NLP/LP	+8/+10	non-recordable	nystagmus, non-recordable VEP, systemically normal, cataract	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
9	2012, Chiang et al.	3	4m	F	Yes	NA	Yes	LP-NLP	NA	NA	oculo-digital sign	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Brazil
11	2012, Koenekoop et al.	MOGL208	56	F	Yes	Yes	Yes	NLP/CF	hyperopia	NA	nystagmus	c.769G>A	p.Glu257Lys	c.817A>G	p.Asn273Asp	French Candadian
9	2012, Chiang et al.	7	10	F	Yes			LP	NA	profoundly abnormal	MD and relatively normal peripheral retina, later MD enlarged	c.104T>C	p.Met35Thr	c.769G>A	p.Glu257Lys	USA
9	2012, Chiang et al.	11	8w?	F	Yes	NA	NA	LP	NA	severe dysfunction	nystagmus and roving eye	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Australian, English
10	2012, Perrault et al.	P1	19	M	Yes	Yes	NA	LP	+9	NA		c.1A>G	p.Met1?	c.769G>A	p.Glu257Lys	Italy
10	2012, Perrault et al.	P3	3	M	Yes	NA	NA	LP	NA	NA	photophobia	c.362delA	p.Glu121Glufs*20	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P4	9	M	Yes	Yes	Yes	LP	+7/+8	NA	photophobia, neonatal and fast progressing macular atrophy	c.439G>C	p.Ala147Pro	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P6	19	F	Yes	NA	NA	LP	+4	NA	photophobia	c.518A>G	p.Asp173Gly	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P16	24	F	Yes	Yes	Yes	LP	+2.25/+6.75	NA	photophobia	c.752A>C	p.His251Pro	c.769G>A	p.Glu257Lys	Spain
10	2012, Perrault et al.	P18	12	M	Yes	NA	NA	LP	+6/+7	NA	photophobia	c.37G>A	p.Ala13Thr	c.468T>C	p.Leu153Pro	France
10	2012, Perrault et al.	P20	1	F	Yes	Yes	Yes	LP	+6.75/+7	NA	photophobia	c.362delA	p.Glu121Glufs*20	c.532G>A	p.Val178Met	France
10	2012, Perrault et al.	P25	8	NA	Yes	NA	NA	LP		NA		c.769G>A	p.Glu257Lys	?	?	France
10	2012, Perrault et al.	P26.1	28	NA	Yes	Yes	NA	LP	NA	NA	photophobia	c.769G>A	p.Glu257Lys	?	?	Portugal/La Leunion (France)
10	2012, Perrault et al.	P27	3	NA	Yes	NA	Yes	LP	+5	NA	photophobia	c.769G>A	p.Glu257Lys	?	?	France
12	2012, Falk et al.	LVPEI-79-IV-1	1	NA	Yes	moderate	Yes	LP	NA	NA	motor developmental delay and it was improved	c.98A>G	p.Asp33Gly	c.98A>G	p.Asp33Gly	Indian
12	2012, Falk et al.	UCL-2-II-2	13m	F	Yes	NA	Yes	LP	+3	non-recordable		c.161C>T	p.Ala54Val	c.293T>G	p.Val98Gly	Caribbean & Sri Lankan
12	2012, Falk et al.	UCL-4-II-1	13m	M	Yes	NA	Yes	LP	+6.5/+6.5	non-recordable		c.723delA	p.Pro241Profs*45	c.769G>A	p.Glu257Lys	Caribbean & Irish

12	2012, Falk et al.	UCL-5-II-2	18m	F	Yes	NA	Yes	LP	+6/+6		enophthalmos, older sibling was developmentally delay	c.59T>A	p.Ile20Asn	c.769G>A	p.Glu257Lys	Polish
23	2014, Siemiatkowska et al.	1	23	M	Yes	No	Yes	LP	+7/+7.5	non-recordable	cataract (posterior subcapsular)	c.53A>G	p.Asn18Ser	c.472G>C	p.Asp158His	German/French
23	2014, Siemiatkowska et al.	2	25	F	Yes	Yes	Yes	LP	not performed	non-recordable	none	c.253T>C	p.Trp85Arg	c.769G>A	p.Glu257Lys	German
23	2014, Siemiatkowska et al.	3.2	2	F	Yes	NA	Yes	LP	+6.5	not performed	none	c.617A>G	p.His206Ala	c.769G>A	p.Glu257Lys	Dutch
23	2014, Siemiatkowska et al.	5	16	F	Yes	NA	No	LP	-0.25	non-recordable	none	c.731T>C	p.Val244Ala	c.769G>A	p.Glu257Lys	German
25	2015, Coppieters et al.	F4-II:1	3m	M	Yes	No	No	LP	+7.32	non-recordable	night blindness	c.709C>T	p.Arg237Cys	partial gene deletion		Japanese
18	2018, Hosono et al.	S132	14	F	Yes	Yes	Yes	LP	NA	non-recordable	nystagmus from birth	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Japanese
37	2018, Weisschuh, et al.	10	48	F	NA	NA	NA	LP	NA	NA	cataract, nystagmus	c.12dup	p.E5Rfs*4	c.769G>A	p.Glu257Lys	German
40	2020, Bedoni et al.	Family 1-P1	1m	M	Yes	Yes	Yes	LP	NA	non-recordable	SHILCA	7.1kb duplication		7.1kb duplication		Italy
40	2020, Bedoni et al.	Family 1-P2	1m	F	Yes	NA	Yes	LP	+8	non-recordable	SHILCA	7.1kb duplication		7.1kb duplication		Italy
43	2020, Sallum, et al.	61.1	since birth	NA	Yes/No	Yes	Yes	LP	NA	NA		c.716T>C	p.Leu239Ser	c.769G>A	p.Glu257Lys	Brazilian
43	2020, Sallum, et al.	63.1	since birth	NA	Yes/No	Yes	Yes	LP	NA	NA	Coats disease in some of them	c.37G>A	p.Ala13Thr	c.293T>G	p.Val98Gly	Brazilian
43	2020, Sallum, et al.	67.1	since birth	NA	Yes/No	Yes	Yes	LP	NA	NA		c.507G>A	p.Trp169Ter	c.769G>A	p.Glu257Lys	Brazilian
44	2020, Skorczyk-Werner et al.	9-29	10	F	NA	Yes	Yes	LP	hyperopia with astigmatism	NA	photophobia, nystagmus, oculo-digital sign	c.59T>A	p.Ile20Asn	c.769G>A	p.Glu257Lys	Polish
12	2012, Falk et al.	CHOP/MEEI-007-II-2	3	F	Yes	Yes	Yes	LP at age 4	NA	NA	significant fixing and following at age 3	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Asian American
9	2012, Chiang et al.	1	several months	M	Yes	NA	Yes	LP, lack of vision several hours after birth	NA	non-recordable		c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Cherokee Indian, Asian, Hispanic, Caucasian
24	2015, Wang et al.	SRF120	22	M	Yes	NA	Yes	LP/LP	myopia	NA	keratoconus, nystagmus	c.709C>T	p.Arg237Cys	c.713A>G	p.Tyr238Cys	Cinese
24	2015, Wang et al.	SRF600	1	F	Yes	NA	Yes (dark retina)	LP/LP	NA	NA	oculo-digital sign	c.713A>G	p.Tyr238Cys	c.721C>T	p.Pro241Ser	Cinese
19	2020, Surl et al.	25	6.3	M	Yes	NA	Yes	LP/LP	+4.5/+5.5	non-recordable	systemically normal	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	27	1.4	M	Yes	NA	No	LP/LP	+4.5/+4.5	non-recordable	systemically normal	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	28	0.5	F	Yes	NA	Yes	LP/LP	+4/+5	non-recordable	systemically normal	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
23	2014, Siemiatkowska et al.	6	72	M	Yes	Yes	Yes	LP/HM	not performed	non-recordable	panretinal degeneration	c.619C>T	p.R207W	c.769G>A	p.Glu257Lys	Dutch
16	2016, Wang et al.	20a	10m	M	Yes	Yes	Yes	LP/light following	+5	NA	night blindness, oculo-digital sign	c.721C>T	p.Pro241Ser	c.721C>T	p.Pro241Ser	Han Chinese
9	2012, Chiang et al.	9	21	M	Yes	Yes	Yes	color vision lost thereafter, LP at age 22.	NA	NA		c.255G>A	p.Trp85*	c.769G>A	p.Glu257Lys	Canada
10	2012, Perrault et al.	P2	15	F	NA	NA	NA	HM	high hyperopia	NA		c.205A>G	p.Met69Val	c.769G>A	p.Glu257Lys	Spain/France
22	2014, Jin et al.	Family-035-II:2	6	F	Yes	Yes	Yes	HM	NA	non-recordable	normal mental/physical health	c.196C>T	p.Arg66Trp	c.272A>G	p.Glu91Gly	Chinese
22	2014, Jin et al.	Family-035-II:3	8	M	Yes	Yes	Yes	HM	NA	non-recordable	normal mental/physical health	c.196C>T	p.Arg66Trp	c.272A>G	p.Glu91Gly	Chinese
16	2016, Wang et al.	22	4	M	Yes	Yes	Yes	HM	NA	NA	night blindness	c.500A>G	p.Asn167Ser	c.668C>T	p.Ser223Phe	Han Chinese
43	2020, Sallum, et al.	62.1	3m	NA	Yes/No	Yes	Yes	HM	NA	NA		exon 2-4 duplication		c.769G>A	p.Glu257Lys	Brazilian
44	2020, Skorczyk-Werner et al.	19-47	15	M	Yes	Yes	NA	HM	hyperopia with astigmatism	NA	mild nystagmus, oculo-digital sign	c.65A>G	p.Asn22Ser	c.769G>A	p.Glu257Lys	Polish
12	2012, Falk et al.	UCL-6-II-1	41	M	Yes	NA	Yes	HM/HM	NA	NA	IOL OU	c.552A>G	p.Ile184Met	c.769G>A	p.Glu257Lys	British (European descent)
9	2012, Chiang et al.	5	1	M	Yes	NA	Yes	no fixation	NA	NA	Coats' disease OS at age 4 years	c.293T>G	p.Val98Gly	c.769G>A	p.Glu257Lys	Brazil
9	2012, Chiang et al.	8	1	M	NA	NA	NA	no fixation	NA	profoundly abnormal	roving eye	c.104T>C	p.Met35Thr	c.769G>A	p.Glu257Lys	USA
16	2016, Wang et al.	21	1	M	Yes	Yes	Yes	no following and fixation	NA	NA	oculo-digital sign	c.C619T	p.Arg207Trp	c.721C>T	p.Pro241Ser	Han Chinese
19	2020, Surl et al.	22	1.4	F	Yes	NA	Yes	no maintained fixation	+5/+5.5	non-recordable	multiple "bear-foot like" colobomatous MD, systemically normal	c.275G>A	p.Trp92*	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	23	0.5	M	Yes	NA	No	no maintained fixation	+4.75/+7.25	non-recordable	systemically normal	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	24	4.2	M	Yes	NA	No	no maintained fixation	+4/+4.5	non-recordable	systemically normal	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	26	1.2	F	Yes	NA	No	no maintained fixation	+3.5/+3.5	non-recordable	systemically normal	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	29	0.8	F	Yes	NA	No	no maintained fixation	+6.5/+6.5	non-recordable	systemically normal	exon 4-5 deletion		c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	30	1.3	F	Yes	NA	No	no maintained fixation	+5/+5	non-recordable	systemically normal	exon 2 deletion		c.709C>T	p.Arg237Cys	Korean
12	2012, Falk et al.	CHOP/MEEI-047-IV-1	11	F	Yes	Yes	Yes	not fix or follow objects	NA	non-recordable	enophthalmos, corneal opacity OS, sensorineural hearing loss, developmental delay, non-verbal autism (GJB2 mutations combined)	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani
31	2017, Han et al.	P4	13m	M	Yes	NA	Yes	severe visual impairment	+5.5/+5.5	non-recordable		c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
31	2017, Han et al.	P5	7m	F	Yes	NA	Yes	severe visual impairment	+4/+5	non-recordable		c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean
31	2017, Han et al.	P6	12m	M	Yes	NA	Yes (marbled fundus)	severe visual impairment	+4/+3.5	non-recordable		c.709C>T	p.Arg237Cys	? (not detected)		Korean
30	2017, Jinda et al.	LCATH3	13m	M	Yes	Yes	Yes	wondering eye	+4.5/-0.3	non-recordable		c.196C>T	p.Arg66Trp	c.634G>A	p.Val212Met	Thai
9	2012, Chiang et al.	6	6m	F	Yes	NA	Yes	able to see colors and shapes until 22 & 24 y.o., LP at 26 y.o.	NA	NA		c.451G>T	p.Val151Phe	c.769G>A	p.Glu257Lys	Canada
12	2012, Falk et al.	LVPEI-73-II-2	5m	NA	Yes	minimal	Yes	fixing on and following light	NA	NA		c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Indian
12	2012, Falk et al.	LVPEI-100-II-2	3m	NA	Yes	minimal	Yes	following and fixing light	+9	NA	7-year follow-up	c.709C>T	p.Arg237Cys	c.565delG	p.Ala189Leufs*25	Indian
10	2012, Perrault et al.	P9	19	M	Yes	NA	NA	CF	+3/+0.75	NA	photophobia and night blindness	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P11.1	24	M	Yes	Yes	Yes	CF	+2/+3.5	NA		c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P11.2	20	M	Yes	Yes	Yes	CF	+2/+3.5	NA		c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P17	28	F	NA	NA	NA	CF	NA	NA	photophobia	c.716T>C	p.Leu239Ser	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P21	16	F	Yes	NA	NA	CF	high hyperopia	NA	night blindness	c.507G>A	p.Trp169*	c.507G>A	p.Trp169*	Algeria
10	2012, Perrault et al.	P24	22	NA	Yes	NA	Yes	CF	+3/+3.75	NA	photophobia	c.507G>A	p.Trp169*	?		France
11	2012, Koenekoop et al.	MOGL512	13	M	Yes	Yes	Yes	CF	hyperopia	NA	nystagmus	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	French Candadian
11	2012, Koenekoop et al.	MOGL3294	41	NA	Yes	Yes	Yes	CF	hyperopia	NA	nystagmus	c.199G>T	p.Val67Phe	c.769G>A	p.Glu257Lys	Russian
11	2012, Koenekoop et al.	MOGL3698	10	NA	Yes	Yes	Yes	CF	hyperopia	NA	nystagmus	c.37G>A	p.Ala13Thr	c.293T>A	p.Val98Gly	Haitian (African)
16	2016, Wang et al.	18a	5	F	Yes	Yes	Yes	CF	+2.5	NA	night blindness	c.695G>A	p.Arg232Lys	c.695G>A	p.Arg232Lys	Han Chinese
34	2018, Khan, et al.	II-3	10	M	Yes	No	No	2/200	+4.25/+5.25	NA	unlike II-1, no apparent difference in day/night vision	c.500A>G	p.Asn167Ser	c.500A>G	p.Asn167Ser	Arab
11	2012, Koenekoop et al.	MOGL3174	33	F	Yes	Yes	Yes	20/2000	hyperopia	NA	nystagmus	c.507G>A	p.Trp169*			Irish
34	2018, Khan, et al.	II-1	13	F	Yes	No	No	4/200	+6/+6	cone-rod dysfunction	CRD rather than LCA, but fundus findings were LCA9, CCDC66 mutations were also found but probably not pathogenic	c.500A>G	p.Asn167Ser	c.500A>G	p.Asn167Ser	Arab
25	2015, Coppieters et al.	F3-II:1	3m	M	Yes	No	No	0.03	+1.31	non-recordable	night blindness	c.709C>T	p.Arg237Cys	partial gene deletion		Japanese
12	2012, Falk et al.	UCL-3-II-1	10	F	Yes	NA	Yes	1.5 logMAR	+3/+5	non-recordable		c.37G>A	p.Ala13Thr	c.293T>G	p.Val98Gly	Caribbean
23	2014, Siemiatkowska et al.	3.1	7	M	Yes	No	Yes (subtle)	2/60	+3	non-recordable	none	c.617A>G	p.H206A	c.769G>A	p.Glu257Lys	Dutch
16	2016, Wang et al.	19	5	F	Yes	Yes	Yes	0.05	+2	NA	night blindness, exdative change	c.43G>A	p.Gly15Ser	c.634G>A	p.Val212Met	Han Chinese
10	2012, Perrault et al.	P8	6	F	Yes	NA	NA	1/20	+5.5/+4.5	NA		c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P12	45	F	NA	NA	NA	1/20	NA	NA		c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P14	49	M	Yes	Yes	Yes	<1/20	+8/+8	NA	night blindness	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
35	2018, Nash et al.	Case 2	7	F	Yes	No	No	6/90 > 6/120	+1/+2	cone-rod dysfunction	fundi are similar to Case 1	c.53A>G	p.Asn18Ser	c.769G>A	p.Glu257Lys	Caucasian

39	2020, Ruberto et al.	1	1y1m	F	Yes	Yes	Yes	0.7/10	NA	non-recordable	peripheral coloboma, pendular nystagmus	c.619C>T	p.Arg207Tyr?	c.769G>A	p.Glu257Lys	Italy
10	2012, Perrault et al.	P10	9	F	Yes	NA	NA	<1/10	+8.25/+9.75	NA	moderate photophobia	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
9	2012, Chiang et al.	4	6m	M	Yes	NA	Yes	20/200 can see colors	NA	cone dysfunction	cone-rod dysfunction, poor night vision	c.457C>G	p.Leu153Val	c.769G>A	p.Glu257Lys	Canada
12	2012, Falk et al.	LVPEI-128-II-3	28	M	Yes	minimal	moderate	20/280, 20/150	NA	NA		c.817A>G	p.Asn273Asp	c.215T>A	p.Leu72His	Indian
35	2018, Nash et al.	Case 1	26	F	Yes	No	No	6/45, 1/120	-1.7/-2	cone dysfunction	large macular degeneration beyond the vascular arcade	c.271G>A	p.Glu91Lys	c.271G>A	p.Glu91Lys	Indian
39	2020, Ruberto et al.	16	1	NA	Yes	Yes	No	1.5/10	NA	NA	CGH array deletion in Crl p36.22 involving NMNAT1 gene, Jerky nystagmus	c.769G>A	p.Glu257Lys	c.769G>A	p.Glu257Lys	Italy
24	2015, Wang et al.	SRF94	19	M	Yes	NA	Yes	0.03/0.2	hyperopia	NA	nystagmus	c.116-2A>G		c.634G>A	p.Val212Met	Cinese
41	2020, Habibi et al.	F16-V.1	23	F	No	No	Yes	5/10, 5/10	NA	NA	RP, OCT normal, relatively preserved visual acuity until the third decade	c.37G>A	p.Ala13Thr	c.37G>A	p.Ala13Thr	Tunisia
41	2020, Habibi et al.	F16-V.4	21	M	No	Yes	Yes	7/10, 6/10	NA	NA	RP, OCT normal, relatively preserved visual acuity until the third decade	c.37G>A	p.Ala13Thr	c.37G>A	p.Ala13Thr	Tunisia
9	2012, Chiang et al.	10	30	M	NA	NA	NA	NA	NA	NA		c.255G>A	p.Trp85*	c.769G>A	p.Glu257Lys	Canada
10	2012, Perrault et al.	P13	31	M	NA	NA	NA	NA	NA	NA		c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P23.1	NA	NA	Yes	NA	Yes	NA	+6	NA		c.769G>A	p.Glu257Lys	?	?	Portugal
11	2012, Koenekoop et al.	MOGL1269	NA	F	NA	NA	NA	NA	NA	NA		c.451G>A	p.Val151Phe	c.769G>A	p.Glu257Lys	European
25	2015, Coppieters et al.	F1-IV:1	NA	F	Yes	NA	NA	NA	NA	NA		c.-70A>T	p.?	c.-70A>T	p.?	Niger
25	2015, Coppieters et al.	F1-IV:2	NA	F	Yes	NA	NA	NA	NA	NA		c.-70A>T	p.?	c.-70A>T	p.?	Niger
25	2015, Coppieters et al.	F1-IV:4	NA	F	Yes	NA	NA	NA	NA	NA		c.-70A>T	p.?	c.-70A>T	p.?	Niger
25	2015, Coppieters et al.	F2-IV:2	NA	M	Yes	NA	Yes	NA	NA	NA		c.-69C>T	p.?	c.-70A>T	p.?	Morocco
25	2015, Coppieters et al.	F2-IV:3	NA	F	Yes	NA	Yes	NA	NA	NA		c.-69C>T	p.?	c.-70A>T	p.?	Morocco
25	2015, Coppieters et al.	F5	NA	NA	NA	NA	NA	NA	NA	NA		c.364del	p.Arg122fs	c.769G>A	p.Glu257Lys	NA
25	2015, Coppieters et al.	F6	NA	NA	NA	NA	NA	NA	NA	NA		c.542A>G	p.Tyr181Cys	c.769G>A	p.Glu257Lys	NA
25	2015, Coppieters et al.	F7	NA	NA	NA	NA	NA	NA	NA	NA		c.679C>T	p.Arg227Trp	c.769G>A	p.Glu257Lys	NA
25	2015, Coppieters et al.	F8	NA	NA	NA	NA	NA	NA	NA	NA		c.562C>T	p.Arg188Trp	?	?	NA
25	2015, Coppieters et al.	F9	NA	NA	NA	NA	NA	NA	NA	NA		c.53A>G	p.Asn18Ser	?	?	NA
29	2016, Xu et al.	LH01-II:1	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)	c.196C>T	p.Arg66Trp	c.G634A	p.V212M	Chinese
29	2016, Xu et al.	LH01-II:2	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)	c.196C>T	p.Arg66Trp	c.G634A	p.V212M	Chinese
29	2016, Xu et al.	QT1118-II:1	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)	c.619C>T	p.Arg207Trp	c.721C>T	p.Pro241Ser	Chinese
29	2016, Xu et al.	QT1118-II:3	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)	c.619C>T	p.Arg207Trp	c.721C>T	p.Pro241Ser	Chinese
29	2016, Xu et al.	LH05-II:1	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)	c.737A>T		c.737A>T		Chinese
32	2017, Di Iorio et al.	27	5	NA	NA	NA	Yes	NA	NA	NA	onset: 9m	c.53A>G	p.Asn18Ser	c.542A>G	p.Tyr181Cys	Italy
33	2017, Porto et al.	20-FBP_57	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)	c.37G>A	p.Ala13Thr	c.293T>G	p.Val98Gly	Brazilian
18	2018, Hosono et al.	EYE159	NA	NA	NA	NA	NA	NA	NA	NA		c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Japanese
36	2018, Han et al.	-	60	M	Yes	Yes	Yes	NA	NA	NA		NA		NA	NA	NA
38	2020, Xu et al.	-	NA	NA	NA	NA	NA	NA	NA	NA		c.196C>T	p.Arg66Trp	c.721C>T	p.Pro241Ser	Chinese
40	2020, Bedoni et al.	Family 2-P3	1	M	NA	NA	NA	NA	NA	NA		7.1kb duplication		7.1kb duplication		Italy
42	2020, Hull et al.	-	NA	NA	NA	NA	NA	NA	NA	NA		c.553T>C	p.Cys185Arg	c.769G>A	p.Glu257Lys	New Zealand
45	2021, Han et al.	-	6m	F	Yes	Yes	Yes	NA	NA	NA	Coats' disease	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean

NA: not available, y: years, m: months, w: weeks, NLP: no light perception, LP: light perception, CF: counting fingers, CRD: cone-rod dystrophy, LCA: Leber congenital amaurosis, RP: retinitis pigmentosa, OCT: Optical coherence tomographty, y.o.: years old, MD: macular degeneration, IOL: intraocular lens, VEP: visual evoked potential, SHILCA: spondylo-epiphyseal dysplasia, sensorineural hearing loss, intellectual disability and Leber congenital amaurosis

The variants are transcribed exactly as presented in the original article.