

Supplementary Table 1.  
Summary of the genetic and clinical findings of the *NMNA1*-associated retinopathy

Reference #	Published year, Authors	Patient #	Status	Variant 1	Variant 1	Variant 2	Variant 2	Ethnicity	Age	Gender	Macular degeneration	Optic disk atrophy	Retinal degeneration	Visual acuity	Refractive errors	Electro-retinogram	Others
9	2012, Chiang et al.	1	HTZ	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Cherokee Indian, Asian, Hispanic, Caucasian	several months	M	Yes	NA	Yes	LP, lack of vision several hours after birth	NA	non-recordable	
9	2012, Chiang et al.	2	HTZ	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Brazil Caucasian (Poland, Portugal, Lebanon, Italy, Turkey, Germany)	6m	M	Yes	NA	Yes	NLP	NA	NA	nystagmus
9	2012, Chiang et al.	3	HTZ	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Brazil	4m	F	Yes	NA	Yes	NP-NLP	NA	NA	oculo-digital sign
9	2012, Chiang et al.	4	HTZ	c.457C>G c.817A>G	p.Leu153Val p.Asn273Asp	c.769G>A	p.Glu257Lys	Canada	6m	M	Yes	NA	Yes	20/200 can see colors	NA	cone dysfunction	cone-rod dysfunction, poor night vision
9	2012, Chiang et al.	5	HTZ	c.293T>G	p.Val98Gly	c.769G>A	p.Glu257Lys	Brazil	1	M	Yes	NA	Yes	no fixation able to see colors and shapes until 22 & 24 y.o., LP at 26 y.o.	NA	NA	Coats' disease OS at age 4 years
9	2012, Chiang et al.	6	HTZ	c.451G>T	p.Val151Phe	c.769G>A	p.Glu257Lys	Canada	6m	F	Yes	NA	Yes	LP	NA	profoundly abnormal	MD and relatively normal peripheral retina, later MD enlarged
9	2012, Chiang et al.	7	HTZ	c.104T>C	p.Met35Thr	c.769G>A	p.Glu257Lys	USA	10	F	Yes	NA	NA	no fixation	NA	profoundly abnormal	roving eye
9	2012, Chiang et al.	8	HTZ	c.255G>A	p.Trp85*	c.769G>A	p.Glu257Lys	Canada	1	M	NA	NA	NA	color vision lost thereafter, LP at age 22.	NA	NA	
9	2012, Chiang et al.	9	HTZ	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Australian, English	21	M	Yes	NA	Yes	LP	NA	severe dysfunction	nystagmus and roving eye
9	2012, Chiang et al.	10	HTZ	c.1A>G	p.Met1?	c.769G>A	p.Glu257Lys	Italy	30	M	NA	NA	NA	LP	NA	NA	
9	2012, Perrault et al.	11	HTZ	c.205A>G	p.Met69Val	c.769G>A	p.Glu257Lys	Spain/France	8w?	F	Yes	NA	NA	LP	NA	NA	
10	2012, Perrault et al.	P1	HTZ	c.362delA	p.Glu121Glufs'	c.769G>A	p.Glu257Lys	France	19	M	Yes	NA	NA	HM	high hyperopia	NA	
10	2012, Perrault et al.	P2	HTZ	c.439G>C	p.Ala147Pro	c.769G>A	p.Glu257Lys	France	15	F	NA	NA	NA	LP	NA	NA	photophobia
10	2012, Perrault et al.	P3	HTZ	c.439G>C	p.Ala147Pro	c.769G>A	p.Glu257Lys	France	3	M	Yes	NA	NA	LP	NA	NA	photophobia, neonatal and fast progressing macular atrophy
10	2012, Perrault et al.	P4	HTZ	c.439G>C	p.Ala147Pro	c.769G>A	p.Glu257Lys	France	9	M	Yes	Yes	Yes	LP	+7/+8	NA	night blindness
10	2012, Perrault et al.	P5	HTZ	c.439G>C	p.Ala147Pro	c.769G>A	p.Glu257Lys	France	12	F	Yes	NA	NA	NLP/LE	+9.5/+7.75	NA	photophobia
10	2012, Perrault et al.	P6	HTZ	c.518A>G	p.Asp173Gly	c.769G>A	p.Glu257Lys	France	19	F	Yes	NA	NA	LP	+4	NA	photophobia
10	2012, Perrault et al.	P7.1	HTZ	c.542A>G	p.Tyr181Cys	c.769G>A	p.Glu257Lys	France	59	M	invisible due to cataract	invisible due to cataract	NA	NLP	hyperopia	NA	cataract
10	2012, Perrault et al.	P7.2	HTZ	c.542A>G	p.Tyr181Cys	c.769G>A	p.Glu257Lys	France	60	M	invisible due to cataract	invisible due to cataract	NA	NLP	hyperopia	NA	cataract
10	2012, Perrault et al.	P8	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	6	F	Yes	NA	NA	1/20	+5.5/+4.5	NA	
10	2012, Perrault et al.	P9	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	19	M	Yes	NA	NA	CF	+3/+0.75	NA	photophobia and night blindness
10	2012, Perrault et al.	P10	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	9	F	Yes	NA	NA	<1/10	+8.25/+9.75	NA	moderate photophobia
10	2012, Perrault et al.	P11.1	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	24	M	Yes	Yes	Yes	CF	+2/+3.5	NA	
10	2012, Perrault et al.	P11.2	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	20	M	Yes	Yes	Yes	CF	+2/+3.5	NA	
10	2012, Perrault et al.	P12	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	45	F	NA	NA	NA	1/20	NA	NA	
10	2012, Perrault et al.	P13	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	31	M	NA	NA	NA	NA	NA	NA	
10	2012, Perrault et al.	P14	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	France	49	M	Yes	Yes	Yes	<1/20	+8/+8	NA	night blindness
10	2012, Perrault et al.	P15	HTZ	c.643G>T	p.Glu215*	c.769G>A	p.Glu257Lys	France	6	M	Yes	NA	NA	NLP	+4/+5	NA	sun photophobia
10	2012, Perrault et al.	P16	HTZ	c.752A>C	p.His251Pro	c.769G>A	p.Glu257Lys	Spain	24	F	Yes	Yes	Yes	LP	+2.25/+6.75	NA	photophobia
10	2012, Perrault et al.	P17	HTZ	c.716T>C	p.Leu239Ser	c.769G>A	p.Glu257Lys	France	28	F	NA	NA	NA	CF	NA	NA	photophobia
10	2012, Perrault et al.	P18	HTZ	c.37G>A	p.Ala13Thr	c.468T>C	p.Leu153Pro	France	12	M	Yes	NA	NA	LP	+6/+7	NA	photophobia
10	2012, Perrault et al.	P19	HTZ	c.205A>G	p.Met69Val	c.650T>A	p.Ile217Asn	South Africa/France	3	M	None	Yes	Yes	NLP	NA	NA	
10	2012, Perrault et al.	P20	HTZ	c.362delA	p.Glu121Glufs'	c.532G>A	p.Val178Met	France	1	F	Yes	Yes	Yes	LP	+6.75/+7	NA	photophobia
10	2012, Perrault et al.	P21	HMZ	c.507G>A	p.Trp169*	c.507G>A	p.Trp169*	Algeria	16	F	Yes	NA	NA	CF	high hyperopia	NA	night blindness
10	2012, Perrault et al.	P22.1	HTZ	c.319G>T	p.Glu107*	c.709C>T	p.Arg237Cys	France	18	M	invisible due to retinal detachment and cataract	invisible due to retinal detachment and cataract	NA	NLP	NA	NA	retinal detachment and cataract
10	2012, Perrault et al.	P22.2	HTZ	c.319G>T	p.Glu107*	c.709C>T	p.Arg237Cys	France	13	F	invisible due to ocular phytis and cataract	invisible due to ocular phytis and cataract	NA	NLP	NA	NA	retinal detachment and cataract
10	2012, Perrault et al.	P23.1	HTZ	c.769G>A	p.Glu257Lys	?	?	Portugal	NA	NA	Yes	NA	Yes	NA	+6	NA	
10	2012, Perrault et al.	P24	HTZ	c.507G>A	p.Trp169*	?	?	France	22	NA	Yes	NA	Yes	CF	+3/+3.75	NA	photophobia
10	2012, Perrault et al.	P25	HTZ	c.769G>A	p.Glu257Lys	?	?	France	8	NA	Yes	NA	NA	LP	NA	NA	
10	2012, Perrault et al.	P26.1	HTZ	c.769G>A	p.Glu257Lys	?	?	Portugal/La	28	NA	Yes	Yes	Yes	LP	NA	NA	photophobia
10	2012, Perrault et al.	P26.2	HTZ	c.769G>A	p.Glu257Lys	?	?	Leunion (Fr)	24	NA	Yes	Yes	Yes	NLP	NA	NA	
10	2012, Perrault et al.	P27	HTZ	c.769G>A	p.Glu257Lys	?	?	France	3	NA	Yes	NA	Yes	LP	+5	NA	photophobia
10	2012, Perrault et al.	P28.1	HTZ	c.769G>A	p.Glu257Lys	?	?	France	6	NA	Yes	NA	NA	NLP	NA	NA	
10	2012, Perrault et al.	P29	HTZ	c.769G>A	p.Glu257Lys	?	?	La Leunion (Fr)	NA	NA	NA	NA	NA	NLP	NA	NA	photophobia
11	2012, Koenekoop et al.	MOGL208	HTZ	c.769G>A	p.Glu257Lys	c.817A>G	p.Asn273Asp	French Candadian	56	F	Yes	Yes	Yes	NLP/CF	hyperopia	NA	nystagmus
11	2012, Koenekoop et al.	MOGL512	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	French Candadian	13	M	Yes	Yes	Yes	CF	hyperopia	NA	nystagmus
11	2012, Koenekoop et al.	MOGL1269	HTZ	c.451G>A	p.Val151Phe	c.769G>A	p.Glu257Lys	European	NA	F	NA	NA	NA	NA	NA	NA	
11	2012, Koenekoop et al.	MOGL624	HMZ	c.769G>A	p.Glu257Lys	c.769G>A	p.Glu257Lys	Saudi Arabian	10	M	Yes	Yes	Yes	NLP	hyperopia	NA	nystagmus
11	2012, Koenekoop et al.	MOGL3174	HTZ	c.507G>A	p.Trp169*	?	?	Irish	33	F	Yes	Yes	Yes	20/2000	hyperopia	NA	nystagmus
11	2012, Koenekoop et al.	MOGL3294	HTZ	c.199G>T	p.Val67Phe	c.769G>A	p.Glu257Lys	Russian	41	NA	Yes	Yes	Yes	CF	hyperopia	NA	nystagmus
11	2012, Koenekoop et al.	MOGL3698	HTZ	c.37G>A	p.Ala13Thr	c.293T>A	p.Val98Gly	Haitian (African)	10	NA	Yes	Yes	Yes	CF	hyperopia	NA	nystagmus
11	2012, Koenekoop et al.	LCA9 IV:8								F	Yes	Yes	Yes				
11	2012, Koenekoop et al.	LCA9 IV:9								F	Yes	Yes	Yes				
11	2012, Koenekoop et al.	LCA9 IV:11								F	Yes	Yes	Yes				
11	2012, Koenekoop et al.	LCA9 IV:12	HMZ	c.838T>C	p.*280Gln	c.838T>C	p.*280Gln	Pakistani	2-30	F	Yes	NA	Yes	LP-NLP	NA	NA	nystagmus, night blindness, cataract (posterior subcapsular), one has keratoconus
11	2012, Koenekoop et al.	LCA9 IV:13								M	Yes	NA	Yes				
11	2012, Koenekoop et al.	LCA9 IV:16								F	Yes	NA	Yes				
11	2012, Koenekoop et al.	LCA9 IV:17								F	Yes	NA	Yes				
11	2012, Koenekoop et al.	LCA9 IV:18								F	Yes	NA	Yes				
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)
12	2012, Falk et al.	CHOP/MEEI-047-IV-3	HMZ	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani	4m	M	Yes(3y)	mild	Yes	LP > NLP	significant myopia	non-recordable	enophthalmos, neural development delay, non-verbal at age 4, autism, normal hearing
12	2012, Falk et al.	CHOP/MEEI-047-IV-6							NA	M	NA	NA	NA	congenital blindness	NA	NA	LCA without developmetal delay
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)
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)

12	2012, Falk et al.	CHOP/MEEI-007-II-2	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Asian American	3	F	Yes	Yes	Yes	LP at age 4	NA	NA	significant fixing and following at age 3
12	2012, Falk et al.	CHOP/MEEI-053-II-1	HTZ	c.205A>G	p.Met69Val	c.769G>A	p.Glu257Lys	African American	20	F	Yes	Yes	Yes	NLP	NA	non-recordable	33-week's gestational age, developmental delay
12	2012, Falk et al.	LVPEI-73-II-2	HMZ	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Indian	5m	NA	Yes	minimal	Yes	fixing on and following light	NA	NA	
12	2012, Falk et al.	LVPEI-79-IV-1	HMZ	c.98A>G	p.Asp33Gly	c.98A>G	p.Asp33Gly	Indian	1	NA	Yes	moderate	Yes	LP	NA	NA	motor developmental delay and it was improved
12	2012, Falk et al.	LVPEI-100-II-2	HTZ	c.709C>T	p.Arg237Cys	c.565delG	p.Ala189Leufs*25	Indian	3m	NA	Yes	minimal	Yes	following and fixing light	+9	NA	7-year follow-up
12	2012, Falk et al.	LVPEI-128-II-3	HMZ	c.215T>A	p.Leu72His	c.215T>A	p.Leu72His	Indian	28	M	Yes	minimal	moderate	20/280, 20/150	NA	NA	
12	2012, Falk et al.	UCL-1-II-1	HTZ	c.205A>G	p.Met69Val	c.769G>A	p.Glu257Lys	Caucasian descent	4	F	Yes	Yes	Yes	NLP	+11	non-recordable	
12	2012, Falk et al.	UCL-2-II-2	HTZ	c.161C>T	p.Ala54Val	c.293T>G	p.Val98Gly	Caribbean & Sri Lankan	13m	F	Yes	NA	Yes	LP	+3	non-recordable	
12	2012, Falk et al.	UCL-3-II-1	HTZ	c.37G>A	p.Ala13Thr	c.293T>G	p.Val98Gly	Caribbean	10	F	Yes	NA	Yes	1.5 logMAR	+3/+5	non-recordable	
12	2012, Falk et al.	UCL-4-II-1	HTZ	c.723delA	p.Pro241Profs*	c.769G>A	p.Glu257Lys	Caribbean & Irish	13m	M	Yes	NA	Yes	LP	+6.5/+6.5	non-recordable	
12	2012, Falk et al.	UCL-5-II-2	HTZ	c.59T>A	p.Ile20Asn	c.769G>A	p.Glu257Lys	Polish	18m	F	Yes	NA	Yes	LP	+6/+6	non-recordable	enophthalmos, older sibling was developmentally delay
12	2012, Falk et al.	UCL-6-II-1	HTZ	c.552A>G	p.Ile184Met	c.769G>A	p.Glu257Lys	British (European descent)	41	M	Yes	NA	Yes	HM/HM	NA	NA	IOL OU
12	2012, Falk et al.	UCL-7-II-1	HTZ	c.466G>C	p.Gly156Arg	c.769G>A	p.Glu257Lys	British (European descent)	36m	M	Yes	Yes	Yes	NLP	NA	non-recordable	
22	2014, Jin et al.	Family-035-II:2	HTZ	c.196C>T	p.Arg66Trp	c.272A>G	p.Glu91Gly	Chinese	6	F	Yes	Yes	Yes	HM	NA	non-recordable	normal mental/physical health
22	2014, Jin et al.	Family-035-II:3	HTZ	c.196C>T	p.Arg66Trp	c.272A>G	p.Glu91Gly	Chinese	8	M	Yes	Yes	Yes	HM	NA	non-recordable	normal mental/physical health
23	2014, Siemiatkowska et al.	1	HTZ	c.53A>G	p.Asn18Ser	c.472G>C	p.Asp158His	German/French	23	M	Yes	None	Yes	LP	+7/+7.5	non-recordable	cataract (PSC)
23	2014, Siemiatkowska et al.	2	HTZ	c.253T>C	p.Trp85Arg	c.769G>A	p.Glu257Lys	German	25	F	Yes	Yes	Yes	LP	not performed	non-recordable	none
23	2014, Siemiatkowska et al.	3.1	HTZ	c.617A>G	p.His206Ala	c.769G>A	p.Glu257Lys	Dutch	7	M	Yes	None	Yes (subtle)	2/60	+3	non-recordable	none
23	2014, Siemiatkowska et al.	3.2	HTZ	c.617A>G	p.His206Ala	c.769G>A	p.Glu257Lys	Dutch	2	F	Yes	NA	Yes	LP	+6.5	not performed	none
23	2014, Siemiatkowska et al.	4	HTZ	c.595G>T	p.Glu199*	c.769G>A	p.Glu257Lys	Danish	64	M	None	None	Yes	NLP	not performed	not performed	keratoconus
23	2014, Siemiatkowska et al.	5	HTZ	c.731T>C	p.Val244Ala	c.769G>A	p.Glu257Lys	German	16	F	Yes	NA	None	LP	-0.25	non-recordable	none
23	2014, Siemiatkowska et al.	6	HTZ	c.619C>T	p.Arg207Trp	c.769G>A	p.Glu257Lys	Dutch	72	M	Yes	Yes	Yes	LP/HM	not performed	non-recordable	panretinal degeneration
24	2015, Wang et al.	SRF120	HTZ	c.709C>T	p.Arg237Cys	c.713A>G	p.Tyr238Cys	Cinese	22	M	Yes	NA	Yes	LP/LP	myopia	NA	keratoconus, nystagmus
24	2015, Wang et al.	SRF94	HTZ	c.116-2A>G	c.634G>A	c.634G>A	p.Val212Met	Cinese	19	M	Yes	NA	Yes	0.03/0.2	hyperopia	NA	nystagmus
24	2015, Wang et al.	SRF600	HTZ	c.713A>G	p.Tyr238Cys	c.721C>T	p.Pro241Ser	Cinese	1	F	Yes	NA	Yes (dark retina)	LP/LP	NA	NA	oculo-digital sign
25	2015, Coppieters et al.	F1-IV:1							NA	F	Yes	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F1-IV:2	HMZ	c.070A>T		c.070A>T		Niger	NA	F	Yes	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F1-IV:4							NA	F	Yes	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F2-IV:2							NA	M	Yes	NA	Yes	NA	NA	NA	
25	2015, Coppieters et al.	F2-IV:3	HMZ	c.069C>T		c.070A>T		Morocco	NA	F	Yes	NA	Yes	NA	NA	NA	
25	2015, Coppieters et al.	F3-II:1	HTZ	c.709C>T	p.Arg237Cys	partial gene deletion		Japanese	3m	M	Yes	None	None	0.03	+1.31	non-recordable	night blindness
25	2015, Coppieters et al.	F4-II:1	HTZ	c.709C>T	p.Arg237Cys	partial gene deletion		Japanese	3m	M	Yes	None	None	LP	+7.32	non-recordable	night blindness
25	2015, Coppieters et al.	F5	HTZ	c.364del	p.Arg122fs	c.769G>A	p.Glu257Lys	NA	NA	NA	NA	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F6	HTZ	c.542A>G	p.Tyr181Cys	c.769G>A	p.Glu257Lys	NA	NA	NA	NA	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F7	HTZ	c.679C>T	p.Arg227Trp	c.769G>A	p.Glu257Lys	NA	NA	NA	NA	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F8	HTZ	c.562C>T	p.Arg188Trp	? (not detected)		NA	NA	NA	NA	NA	NA	NA	NA	NA	
25	2015, Coppieters et al.	F9	HTZ	c.53A>G	p.Asn18Ser	? (not detected)		NA	NA	NA	NA	NA	NA	NA	NA	NA	
26	2015, Deng et al.	II:2							24	M	Yes	NA	NA	LP/NLP	NA	non-recordable	
26	2015, Deng et al.	II:5	HMZ	c.721C>T	p.Pro241Ser	c.721C>T	p.Pro241Ser	Chinese	10	M		not-performed		NLP	NA	not-performed	
27	2015, Hedergott et al.	II.1	HMZ	c.25G>A	p.Val9Met	c.25G>A	p.Val9Met	Pakistani	5m	F	Yes	None	Yes	NLP/LP	+8/+10	non-recordable	nystagmus, non-recordable VEP, systemically normal, cataract
27	2015, Hedergott et al.	II.3							4w	F	Yes	Yes	Yes	NLP/NLP	+4/+4	non-recordable	nystagmus, non-recordable VEP
28	2016, Bravo-Gil et al.	RP157-II:3	HTZ	c.197G>A	p.Arg66Gln	c.769G>A	p.Glu257Lys	Spanish	6m	M	Yes	Yes	Yes	NA	NA	NA	malformation syndrome
29	2016, Xu et al.	LH01-II:1	HTZ	c.196C>T	p.Arg66Trp	c.634G>A	p.Val212Met	Chinese	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)
29	2016, Xu et al.	LH01-II:2							NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)
29	2016, Xu et al.	QT1118-II:1	HTZ	c.619C>T	p.Arg207Trp	c.721C>T	p.Pro241Ser	Chinese	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)
29	2016, Xu et al.	QT1118-II:3							NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)
29	2016, Xu et al.	LH05-II:1	HMZ	c.737A>T		c.737A>T		Chinese	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)
16	2016, Wang et al.	18a	HMZ	c.695G>A	p.Arg232Lys	c.695G>A	p.Arg232Lys	Han Chinese	5	F	Yes	Yes	Yes	CF	+2.5	NA	night blindness
16	2016, Wang et al.	19	HTZ	c.43G>A	p.Gly15Ser	c.634G>A	p.Val212Met	Han Chinese	5	F	Yes	Yes	Yes	0.05	+2	NA	night blindness, exdative change
16	2016, Wang et al.	20a	HMZ	c.721C>T	p.Pro241Ser	c.721C>T	p.Pro241Ser	Han Chinese	10m	M	Yes	Yes	Yes	LP/light following	+5	NA	night blindness, oculo-digital sign
16	2016, Wang et al.	21	HTZ	c.C619T	p.Arg207Trp	c.721C>T	p.Pro241Ser	Han Chinese	1	M	Yes	Yes	Yes	no following and fixation	NA	NA	oculo-digital sign
16	2016, Wang et al.	22	HTZ	c.500A>G	p.Asn167Ser	c.668C>T	p.Ser223Phe	Han Chinese	4	M	Yes	Yes	Yes	HM	NA	NA	night blindness
30	2017, Jinda et al.	LCATH3	HTZ	c.196C>T	p.Arg66Trp	c.634G>A	p.Val212Met	Thai	13m	M	Yes	Yes	Yes	wondering eye	+4.5/-0.3	non-recordable	
31	2017, Han et al.	P4	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	13m	M	Yes	NA	Yes	severe visual impairment	+5.5/+5.5	non-recordable	
31	2017, Han et al.	P5	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	7m	F	Yes	NA	Yes	severe visual impairment	+4/+5	non-recordable	
31	2017, Han et al.	P6	HTZ	c.709C>T	p.Arg237Cys	? (not detected)		Korean	12m	M	Yes	NA	Yes (marbled fundus)	severe visual impairment	+4/+3.5	non-recordable	
17	2017, Tompson et al.	-	HTZ	c.364del		c.769G>A	p.Glu257Lys	Australian	NA	NA	NA	NA	NA	LP-NLP	NA	NA	photophobia
17	2017, Tompson et al.	-	HTZ	c.364del		c.769G>A	p.Glu257Lys	Australian	NA	NA	NA	NA	NA	LP-NLP	NA	NA	photophobia
17	2017, Tompson et al.	-	HTZ	c.507G>A	p.Trp169*	c.769G>A	p.Glu257Lys	Australian	NA	NA	NA	NA	NA	LP-NLP	NA	NA	photophobia
32	2017, Di Iorio et al.	27	HTZ	c.53A>G	p.Asn18Ser	c.542A>G	p.Tyr181Cys	Italy	5	NA	NA	NA	Yes	NA	NA	NA	onset: 9m
33	2017, Porto et al.	20-FBP_57	HTZ	c.37G>A	p.Ala13Thr	c.293T>G	p.Val98Gly	Brazilian	NA	NA	NA	NA	NA	NA	NA	NA	(no clinical data was presented)
34	2018, Khan, et al.	II-1	HMZ	c.500A>G	p.Asn167Ser	c.500A>G	p.Asn167Ser	Arab	13	F	Yes	None	None	4/200	+6/+6	cone-rod dysfunction	CRD rather than LCA, but fundus findings were LCA9, CCDC66 mutations
34	2018, Khan, et al.	II-3							10	M	Yes	None	None	2/200	+4.25/+5.25	NA	were also found but probably not pathogenic
35	2018, Nash et al.	Case 1	HMZ	c.271G>A	p.Glu91Lys	c.271G>A	p.Glu91Lys	Indian	26	F	Yes	None	None	6/45, 1/120	-1.7/-2	cone dysfunction	unlike II-1, no apparent difference in day/night vision
35	2018, Nash et al.	Case 2	HTZ	c.53A>G	p.Asn18Ser	c.769G>A	p.Glu257Lys	Caucasian	7	F	Yes	None	None	6/90 > 6/120	+1/+2	cone-rod dysfunction	large macular degeneration beyond the vascular arcade
18	2018, Hosono et al.	EYE42	HTZ	c.1A>G		c.709C>T	p.Arg237Cys	Japanese	16	M	Yes	None	Yes	NLP	NA	non-recordable	nystagmus from birth
18	2018, Hosono et al.	EYE159	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Japanese	NA	NA	NA	NA	NA	NA	NA	non-recordable	
18	2018, Hosono et al.	S132	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Japanese	14	F	Yes	Yes	Yes	LP	NA	non-recordable	nystagmus from birth
36	2018, Han et al.	-		NA		NA		NA	60	M	Yes	Yes	Yes	NA	NA	NA	
37	2018, Weissschuh, et al.	10	HTZ	c.12dup	p.Glu5Argfs*4	c.769G>A	p.Glu257Lys	German	48	F	NA	NA	NA	LP	NA	NA	cataract, nystagmus
38	2020, Xu et al.	-	HTZ	c.196C>T	p.Arg66Trp	c.721C>T	p.Pro241Ser	Chinese	NA	NA	NA	NA	NA	NA	NA	NA	
19	2020, Surl et al.	22	HTZ	c.275G>A	p.Trp92*	c.709C>T	p.Arg237Cys	Korean	1.4	F	Yes	NA	Yes	no maintained fixation	+5/+5.5	non-recordable	multiple "bear-foot like" colobomatous MD, systemically normal
19	2020, Surl et al.	23	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	0.5	M	Yes	NA	None	no maintained fixation	+4.75/+7.25	non-recordable	systemically normal
19	2020, Surl et al.	24	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	4.2	M	Yes	NA	None	no maintained fixation	+4/+4.5	non-recordable	systemically normal
19	2020, Surl et al.	25	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	6.3	M	Yes	NA	Yes	LP/LP	+4.5/+5.5	non-recordable	systemically normal
19	2020, Surl et al.	26	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	1.2	F	Yes	NA	None	no maintained fixation	+3.5/+3.5	non-recordable	systemically normal
19	2020, Surl et al.	27	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	1.4	M	Yes	NA	None	LP/LP	+4.5/+4.5	non-recordable	systemically normal
19	2020, Surl et al.	28	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	0.5	F	Yes	NA	Yes	LP/LP	+4/+5	non-recordable	systemically normal
19	2020, Surl et al.	29	HTZ	Exon 4-5 deletion		c.709C>T	p.Arg237Cys	Korean	0.8	F	Yes	NA	None	no maintained fixation	+6.5/+6.5	non-recordable	systemically normal
19	2020, Surl et al.	30	HTZ	Exon 2 deletion		c.709C>T	p.Arg237Cys	Korean	1.3	F	Yes	NA	None	no maintained fixation	+5/+5	non-recordable	systemically normal
39	2020, Ruberto et al.	1	HTZ	c.619C>T	p.Arg207Tyr?	c.769G>A	p.Glu257Lys	Italy	1y1m	F	Yes	Yes	Yes	0.7/10	NA	non-recordable	peripheral coloboma, pendular nystagmus
39	2020, Ruberto et al.	16	HMZ	c.769G>A	p.Glu257Lys	c.769G>A	p										

43	2020, Sallum, et al.	61.1	HTZ	c.716T>C	p.Leu239Ser	c.769G>A	p.Glu257Lys	Brazilian	since birth	NA		Yes	Yes	LP	NA	NA	
43	2020, Sallum, et al.	62.1	HTZ	Exon 204 duplication		c.769G>A	p.Glu257Lys	Brazilian	3m	NA		Yes	Yes	HM	NA	NA	
43	2020, Sallum, et al.	63.1	HTZ	c.37G>A	p.Ala13Thr	c.293T>G	p.Val98Gly	Brazilian	since birth	NA		Yes	Yes	LP	NA	NA	
43	2020, Sallum, et al.	64.1	HTZ	c.-57+21C>T		c.759delGinsTA	p.Leu253PhefsTer5	Brazilian	4m	NA	some of them	Yes	Yes	NLP	NA	NA	Coats disease in some of them
43	2020, Sallum, et al.	65.1	HTZ	c.507G>A	p.Trp169Ter	c.769G>A	p.Glu257Lys	Brazilian	3m	NA		Yes	Yes	NLP	NA	NA	
43	2020, Sallum, et al.	66.1	HTZ	c.293T>G	p.Val98Gly	c.769G>A	p.Glu257Lys	Brazilian	since birth	NA		Yes	Yes	NLP	NA	NA	
43	2020, Sallum, et al.	67.1	HTZ	c.507G>A	p.Trp169Ter	c.769G>A	p.Glu257Lys	Brazilian	since birth	NA		Yes	Yes	LP	NA	NA	
44	2020, Skorczyk-Werner et al.	9-29	HTZ	c.59T>A	p.Ile20Asn	c.769G>A	p.Glu257Lys	Polish	10	F	NA	Yes	Yes	LP	hyperopia with astigmatism	NA	photophobia, nystagmus, oculo-digital sign
44	2020, Skorczyk-Werner et al.	19-47	HTZ	c.65A>G	p.Asn22Ser	c.769G>A	p.Glu257Lys	Polish	15	M	Yes	Yes	NA	HM	hyperopia with astigmatism	NA	mild nystagmus, oculo-digital sign
45	2021, Han et al.	-	HTZ	c.196C>T	p.Arg66Trp	c.709C>T	p.Arg237Cys	Korean	6m	F	Yes	Yes	Yes	NA	NA	NA	Coats' disease

HMZ: homozygous, HTZ: heterozygous, 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 tomography, 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.