

Supplementary Table 2.
Variants and ethnicity of the patients with *NMNAT1*-associated retinopathy

Reference #	Published year, Authors	Patient #	Status	Genomic Variant	Protein Variant	Ethnicity
19	2020, Surl et al.	30	HTZ	exon 2 deletion		Korean
43	2020, Sallum, et al.	62.1	HTZ	exon 2-4 duplication		Brazilian
19	2020, Surl et al.	29	HTZ	exon 4-5 deletion		Korean
25	2015, Coppieters et al.	F3-II:1	HTZ	partial gene deletion		Japanese
25	2015, Coppieters et al.	F4-II:1	HTZ	partial gene deletion		Japanese
40	2020, Bedoni et al.	Family 1-P1	HMZ	7.1kb duplication		Italy
40	2020, Bedoni et al.	Family 2-P3	HMZ	7.1kb duplication		Italy
25	2015, Coppieters et al.	F1-IV:1	HMZ	c.-70A>T		Niger
25	2015, Coppieters et al.	F1-IV:1	HMZ	c.-70A>T		Niger
25	2015, Coppieters et al.	F2-IV:2	HMZ	c.-70A>T		Morocco
25	2015, Coppieters et al.	F2-IV:2	HMZ	c.-69C>T		Morocco
43	2020, Sallum, et al.	64.1	HTZ	c.-57+21C>T		Brazilian
10	2012, Perrault et al.	P1	HTZ	c.1A>G		Italy
18	2018, Hosono et al.	EYE42	HTZ	c.1A>G		Japanese
37	2018, Weisschuh, et al.	10	HTZ	c.12dup	p.Glu5Argfs*4	German
12	2012, Falk et al.	CHOP/MEEI-047-IV-1	HMZ	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	LVPEI-73-II-2	HMZ	c.25G>A	p.Val9Met	Indian
27	2015, Hedergott et al.	II.1	HMZ	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	CHOP/MEEI-047-IV-1	HMZ	c.25G>A	p.Val9Met	Pakistani
12	2012, Falk et al.	LVPEI-73-II-2	HMZ	c.25G>A	p.Val9Met	Indian
27	2015, Hedergott et al.	II.1	HMZ	c.25G>A	p.Val9Met	Pakistani
10	2012, Perrault et al.	P18	HTZ	c.37G>A	p.Ala13Thr	France
11	2012, Koenekoop et al.	MOGL3698	HTZ	c.37G>A	p.Ala13Thr	Haitian (African)
12	2012, Falk et al.	UCL-3-II-1	HTZ	c.37G>A	p.Ala13Thr	Caribbean
33	2017, Porto et al.	20-FBP_57	HTZ	c.37G>A	p.Ala13Thr	Brazilian
41	2020, Habibi et al.	F16-V.4	HMZ	c.37G>A	p.Ala13Thr	Tunisia
43	2020, Sallum, et al.	63.1	HMZ	c.37G>A	p.Ala13Thr	Brazilian
41	2020, Habibi et al.	F16-V.4	HMZ	c.37G>A	p.Ala13Thr	Tunisia
16	2016, Wang et al.	19	HTZ	c.43G>A	p.Gly15Ser	Han Chinese
23	2014, Siemiatkowska et al.	1	HTZ	c.53A>G	p.Asn18Ser	German/French
25	2015, Coppieters et al.	F9	HTZ	c.53A>G	p.Asn18Ser	NA
32	2017, Di Iorio et al.	27	HTZ	c.53A>G	p.Asn18Ser	Italy
35	2018, Nash et al.	Case 2	HTZ	c.53A>G	p.Asn18Ser	Caucasian
12	2012, Falk et al.	UCL-5-II-2	HTZ	c.59T>A	p.Ile20Asn	Polish
44	2020, Skorzyl-Werner et al.	9-29	HTZ	c.59T>A	p.Ile20Asn	Polish
44	2020, Skorzyl-Werner et al.	19-47	HTZ	c.65A>G	p.Asn22Ser	Polish
12	2012, Falk et al.	LVPEI-79-IV-1	HMZ	c.98A>G	p.Asp33Gly	Indian
12	2012, Falk et al.	LVPEI-79-IV-1	HMZ	c.98A>G	p.Asp33Gly	Indian
9	2012, Chiang et al.	7	HTZ	c.104T>C	p.Met35Thr	USA
24	2015, Wang et al.	94	HTZ	c.116-2A>G		Chinese
12	2012, Falk et al.	UCL-2-II-2	HTZ	c.161C>T	p.Ala54Val	Caribbean & Sri Lankan
12	2012, Falk et al.	CHOP/MEEI-007-II-2	HTZ	c.196C>T	p.Arg66Trp	Asian American
22	2014, Jin et al.	Family035-II-2	HTZ	c.196C>T	p.Arg66Trp	Chinese
29	2016, Xu et al.	LH01-II:1	HTZ	c.196C>T	p.Arg66Trp	Chinese
30	2017, Jinda et al.	LCATH3	HTZ	c.196C>T	p.Arg66Trp	Thai
31	2017, Han et al.	P4	HTZ	c.196C>T	p.Arg66Trp	Korean
31	2017, Han et al.	P5	HTZ	c.196C>T	p.Arg66Trp	Korean
18	2018, Hosono et al.	EYE159	HTZ	c.196C>T	p.Arg66Trp	Japanese
18	2018, Hosono et al.	S132	HTZ	c.196C>T	p.Arg66Trp	Japanese
38	2020, Xu et al.	-	HTZ	c.196C>T	p.Arg66Trp	Chinese
19	2020, Surl et al.	23	HTZ	c.196C>T	p.Arg66Trp	Korean
19	2020, Surl et al.	24	HTZ	c.196C>T	p.Arg66Trp	Korean
19	2020, Surl et al.	25	HTZ	c.196C>T	p.Arg66Trp	Korean
19	2020, Surl et al.	26	HTZ	c.196C>T	p.Arg66Trp	Korean
19	2020, Surl et al.	27	HTZ	c.196C>T	p.Arg66Trp	Korean
19	2020, Surl et al.	28	HTZ	c.196C>T	p.Arg66Trp	Korean
45	2021, Han et al.	-	HTZ	c.196C>T	p.Arg66Trp	Korean
28	2016, Bravo-Gil et al.	RP157-II:3	HTZ	c.197G>A	p.Arg66Gln	Spanish
11	2012, Koenekoop et al.	MOGL3294	HTZ	c.199G>T	p.Val67Phe	Russian
10	2012, Perrault et al.	P2	HTZ	c.205A>G	p.Met69Val	Spain/France
10	2012, Perrault et al.	P19	HTZ	c.205A>G	p.Met69Val	South Africa/France

12	2012, Falk et al.	CHOP/MEEI-053-II-1	HTZ	c.205A>G	p.Met69Val	African American
12	2012, Falk et al.	UCL-1-II-1	HTZ	c.205A>G	p.Met69Val	European descent
12	2012, Falk et al.	LVPEI-128-II-3	HMZ	c.215T>A	p.Leu72His	Indian
12	2012, Falk et al.	LVPEI-128-II-3	HMZ	c.215T>A	p.Leu72His	Indian
23	2014, Siemiatkowska et al.	2	HTZ	c.253T>C	p.Trp85Arg	German
9	2012, Chiang et al.	9	HTZ	c.255G>A	p.Trp85*	Canada
35	2018, Nash et al.	Case 1	HMZ	c.271G>A	p.Glu91Lys	Indian
35	2018, Nash et al.	Case 1	HMZ	c.271G>A	p.Glu91Lys	Indian
22	2014, Jin et al.	Family035-II-2	HTZ	c.272A>G	p.Glu91Gly	Chinese
19	2020, Surl et al.	22	HTZ	c.275G>A	p.Trp92*	Korean
11	2012, Koenekoop et al.	MOGL3698	HTZ	c.293T>A	p.Val98Gly	Haitian (African)
9	2012, Chiang et al.	5	HTZ	c.293T>G	p.Val98Gly	Brazil
43	2020, Sallum, et al.	66.1	HTZ	c.293T>G	p.Val98Gly	Brazilian
12	2012, Falk et al.	UCL-2-II-2	HTZ	c.293T>G	p.Val98Gly	Caribbean & Sri Lankan
12	2012, Falk et al.	UCL-3-II-1	HTZ	c.293T>G	p.Val98Gly	Caribbean
33	2017, Porto et al.	20-FBP_57	HTZ	c.293T>G	p.Val98Gly	Brazilian
43	2020, Sallum, et al.	63.1	HTZ	c.293T>G	p.Val98Gly	Brazilian
10	2012, Perrault et al.	P22.1	HTZ	c.319G>T	p.Glu107*	France
10	2012, Perrault et al.	P3	HTZ	c.362delA	p.Glu121Glufs*20	France
10	2012, Perrault et al.	P20	HTZ	c.362delA	p.Glu121Glufs*20	France
25	2015, Coppieters et al.	F5	HTZ	c.364del	p.Arg122fs	NA
17	2017, Tompson et al.	-	HTZ	c.364del	p.Arg122fs	Australian
17	2017, Tompson et al.	-	HTZ	c.364del	p.Arg122fs	Australian
10	2012, Perrault et al.	P5	HTZ	c.439+1G>C	Splice	France
10	2012, Perrault et al.	P4	HTZ	c.439G>C	p.Ala147Pro	France
11	2012, Koenekoop et al.	MOGL1269	HTZ	c.451G>A	p.Val151Phe	European
9	2012, Chiang et al.	6	HTZ	c.451G>T	p.Val151Phe	Canada
9	2012, Chiang et al.	4	HTZ	c.457C>G	p.Leu153Val	Canada
12	2012, Falk et al.	UCL-7-II-1	HTZ	c.466G>C	p.Gly156Arg	British (European descent)
10	2012, Perrault et al.	P18	HTZ	c.468T>C	p.Leu153Pro	France
23	2014, Siemiatkowska et al.	1	HTZ	c.472G>C	p.Asp158His	German/French
16	2016, Wang et al.	22	HTZ	c.500A>G	p.Asn167Ser	Han Chinese
34	2018, Khan, et al.	II-1	HMZ	c.500A>G	p.Asn167Ser	Arab
34	2018, Khan, et al.	II-1	HMZ	c.500A>G	p.Asn167Ser	Arab
9	2012, Chiang et al.	1	HTZ	c.507G>A	p.Trp169*	Cherokee Indian
9	2012, Chiang et al.	2	HTZ	c.507G>A	p.Trp169*	Brazil
9	2012, Chiang et al.	3	HTZ	c.507G>A	p.Trp169*	Brazil
9	2012, Chiang et al.	11	HTZ	c.507G>A	p.Trp169*	Australian
10	2012, Perrault et al.	P21	HMZ	c.507G>A	p.Trp169*	Algeria
10	2012, Perrault et al.	P24	HTZ	c.507G>A	p.Trp169*	France
11	2012, Koenekoop et al.	MOGL3174	HTZ	c.507G>A	p.Trp169*	Irish
17	2017, Tompson et al.	-	HTZ	c.507G>A	p.Trp169*	Australian
43	2020, Sallum, et al.	65.1	HTZ	c.507G>A	p.Trp169Ter	Brazilian
43	2020, Sallum, et al.	67.1	HTZ	c.507G>A	p.Trp169Ter	Brazilian
10	2012, Perrault et al.	P21	HMZ	c.507G>A	p.Trp169*	Algeria
10	2012, Perrault et al.	P6	HTZ	c.518A>G	p.Asp173Gly	France
10	2012, Perrault et al.	P20	HTZ	c.532G>A	p.Val178Met	France
10	2012, Perrault et al.	P7.1	HTZ	c.542A>G	p.Tyr181Cys	France
25	2015, Coppieters et al.	F6	HTZ	c.542A>G	p.Tyr181Cys	NA
32	2017, Di Iorio et al.	27	HTZ	c.542A>G	p.Tyr181Cys	Italy
12	2012, Falk et al.	UCL-6-II-1	HTZ	c.552A>G	p.Ile184Met	British (European descent)
42	2020, Hull et al.		HTZ	c.553T>C	p.Cys185Arg	New Zealand
25	2015, Coppieters et al.	F8	HTZ	c.562C>T	p.Arg188Trp	NA
12	2012, Falk et al.	LVPEI-100-II-2	HTZ	c.565delG	p.Ala189Leufs*25	Indian
23	2014, Siemiatkowska et al.	4	HTZ	c.595G>T	p.Glu199*	Danish
23	2014, Siemiatkowska et al.	3.1	HTZ	c.617A>G	p.His206Ala	Dutch
10	2012, Perrault et al.	P8	HTZ	c.619C>T	p.Arg207Trp	France
10	2012, Perrault et al.	P9	HTZ	c.619C>T	p.Arg207Trp	France
10	2012, Perrault et al.	P10	HTZ	c.619C>T	p.Arg207Trp	France
10	2012, Perrault et al.	P11.1	HTZ	c.619C>T	p.Arg207Trp	France
10	2012, Perrault et al.	P12	HTZ	c.619C>T	p.Arg207Trp	France
10	2012, Perrault et al.	P13	HTZ	c.619C>T	p.Arg207Trp	France
10	2012, Perrault et al.	P14	HTZ	c.619C>T	p.Arg207Trp	France
11	2012, Koenekoop et al.	MOGL512	HTZ	c.619C>T	p.Arg207Trp	French Canadian
23	2014, Siemiatkowska et al.	6	HTZ	c.619C>T	p.Arg207Trp	Dutch
16	2016, Wang et al.	21	HTZ	c.619C>T	p.Arg207Trp	Han Chinese
29	2016, Xu et al.	QT1118-II:1	HTZ	c.619C>T	p.Arg207Trp	Chinese
39	2020, Ruberto et al.	1	HTZ	c.619C>T	p.Arg207Tyr?	Italy
24	2015, Wang et al.	94	HTZ	c.634G>A	p.Val212Met	Chinese
29	2016, Xu et al.	LH01-II:1	HTZ	c.634G>A	p.Val212Met	Chinese
16	2016, Wang et al.	19	HTZ	c.634G>A	p.Val212Met	Han Chinese
30	2017, Jinda et al.	LCATH3	HTZ	c.634G>A	p.Val212Met	Thai

10	2012, Perrault et al.	P15	HTZ	c.643G>T	p.Glu215*	France
10	2012, Perrault et al.	P19	HTZ	c.650T>A	p.Ile217Asn	South Africa/France
16	2016, Wang et al.	22	HTZ	c.668C>T	p.Ser223Phe	Han Chinese
25	2015, Coppieters et al.	F7	HTZ	c.679C>T	p.Arg227Trp	NA
16	2016, Wang et al.	18a	HMZ	c.695G>A	p.Arg232Lys	NA
16	2016, Wang et al.	18a	HMZ	c.695G>A	p.Arg232Lys	Han Chinese
12	2012, Falk et al.	CHOP/MEEI-007-II-2	HTZ	c.709C>T	p.Arg237Cys	Asian American
12	2012, Falk et al.	LVPEI-100-II-2	HTZ	c.709C>T	p.Arg237Cys	Indian
24	2015, Wang et al.	120	HTZ	c.709C>T	p.Arg237Cys	Cinese
25	2015, Coppieters et al.	F3-II:1	HTZ	c.709C>T	p.Arg237Cys	Japanese
25	2015, Coppieters et al.	F4-II:1	HTZ	c.709C>T	p.Arg237Cys	Japanese
31	2017, Han et al.	P6	HTZ	c.709C>T	p.Arg237Cys	Korean
10	2012, Perrault et al.	P22.1	HTZ	c.709C>T	p.Arg237Cys	France
31	2017, Han et al.	P4	HTZ	c.709C>T	p.Arg237Cys	Korean
31	2017, Han et al.	P5	HTZ	c.709C>T	p.Arg237Cys	Korean
18	2018, Hosono et al.	EYE42	HTZ	c.709C>T	p.Arg237Cys	Japanese
18	2018, Hosono et al.	EYE159	HTZ	c.709C>T	p.Arg237Cys	Japanese
18	2018, Hosono et al.	S132	HTZ	c.709C>T	p.Arg237Cys	Japanese
19	2020, Surl et al.	22	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	23	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	24	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	25	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	26	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	27	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	28	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	29	HTZ	c.709C>T	p.Arg237Cys	Korean
19	2020, Surl et al.	30	HTZ	c.709C>T	p.Arg237Cys	Korean
45	2021, Han et al.	-	HTZ	c.709C>T	p.Arg237Cys	Korean
24	2015, Wang et al.	600	HTZ	c.713A>G	p.Tyr238Cys	Cinese
24	2015, Wang et al.	120	HTZ	c.713A>G	p.Tyr238Cys	Cinese
10	2012, Perrault et al.	P17	HTZ	c.716T>C	p.Leu239Ser	France
43	2020, Sallum, et al.	61.1	HTZ	c.716T>C	p.Leu239Ser	Brazilian
26	2015, Deng et al.	II:2	HMZ	c.721C>T	p.Pro241Ser	Chinese
24	2015, Wang et al.	600	HTZ	c.721C>T	p.Pro241Ser	Cinese
26	2015, Deng et al.	II:2	HMZ	c.721C>T	p.Pro241Ser	Chinese
16	2016, Wang et al.	20a	HMZ	c.721C>T	p.Pro241Ser	Han Chinese
16	2016, Wang et al.	20a	HMZ	c.721C>T	p.Pro241Ser	Han Chinese
16	2016, Wang et al.	21	HTZ	c.721C>T	p.Pro241Ser	Han Chinese
29	2016, Xu et al.	QT1118-II:1	HTZ	c.721C>T	p.Pro241Ser	Chinese
38	2020, Xu et al.	-	HTZ	c.721C>T	p.Pro241Ser	Chinese
12	2012, Falk et al.	UCL-4-II-1	HTZ	c.723delA	p.Pro241Profs*45	Caribbean & Irish
23	2014, Siemiatkowska et al.	5	HTZ	c.731T>C	p.Val244Ala	German
29	2016, Xu et al.	LH05-II:1	HMZ	c.737A>T		Chinese
29	2016, Xu et al.	LH05-II:1	HMZ	c.737A>T		Chinese
10	2012, Perrault et al.	P16	HTZ	c.752A>C	p.His251Pro	Spain
43	2020, Sallum, et al.	64.1	HTZ	c.759delGinsTA	p.Leu253PhefsTer5	Brazilian
10	2012, Perrault et al.	P23.1	HTZ	c.769G>A	p.Glu257Lys	Portugal
10	2012, Perrault et al.	P25	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P26.1	HTZ	c.769G>A	p.Glu257Lys	Portugal/La Leunion (France)
10	2012, Perrault et al.	P27	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P28.1	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P29	HTZ	c.769G>A	p.Glu257Lys	La Leunion (France)
11	2012, Koenekoop et al.	MOGL208	HTZ	c.769G>A	p.Glu257Lys	French Canadian
11	2012, Koenekoop et al.	MOGL624	HMZ	c.769G>A	p.Glu257Lys	Saudi Arabian
39	2020, Ruberto et al.	16	HMZ	c.769G>A	p.Glu257Lys	Italy
9	2012, Chiang et al.	1	HTZ	c.769G>A	p.Glu257Lys	Cherokee Indian
9	2012, Chiang et al.	2	HTZ	c.769G>A	p.Glu257Lys	Brazil
9	2012, Chiang et al.	3	HTZ	c.769G>A	p.Glu257Lys	Brazil
9	2012, Chiang et al.	4	HTZ	c.769G>A	p.Glu257Lys	Canada
9	2012, Chiang et al.	5	HTZ	c.769G>A	p.Glu257Lys	Brazil
9	2012, Chiang et al.	6	HTZ	c.769G>A	p.Glu257Lys	Canada
9	2012, Chiang et al.	7	HTZ	c.769G>A	p.Glu257Lys	USA
9	2012, Chiang et al.	9	HTZ	c.769G>A	p.Glu257Lys	Canada
9	2012, Chiang et al.	11	HTZ	c.769G>A	p.Glu257Lys	Australian
10	2012, Perrault et al.	P1	HTZ	c.769G>A	p.Glu257Lys	Italy
10	2012, Perrault et al.	P2	HTZ	c.769G>A	p.Glu257Lys	Spain/France
10	2012, Perrault et al.	P3	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P4	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P5	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P6	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P7.1	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P8	HTZ	c.769G>A	p.Glu257Lys	France

10	2012, Perrault et al.	P9	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P10	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P11.1	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P12	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P13	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P14	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P15	HTZ	c.769G>A	p.Glu257Lys	France
10	2012, Perrault et al.	P16	HTZ	c.769G>A	p.Glu257Lys	Spain
10	2012, Perrault et al.	P17	HTZ	c.769G>A	p.Glu257Lys	France
11	2012, Koenekoop et al.	MOGL512	HTZ	c.769G>A	p.Glu257Lys	French Canadian
11	2012, Koenekoop et al.	MOGL1269	HTZ	c.769G>A	p.Glu257Lys	European
11	2012, Koenekoop et al.	MOGL624	HMZ	c.769G>A	p.Glu257Lys	Saudi Arabian
11	2012, Koenekoop et al.	MOGL3294	HTZ	c.769G>A	p.Glu257Lys	Russian
12	2012, Falk et al.	CHOP/MEEI-053-II-1	HTZ	c.769G>A	p.Glu257Lys	African American
12	2012, Falk et al.	UCL-1-II-1	HTZ	c.769G>A	p.Glu257Lys	European descent
12	2012, Falk et al.	UCL-4-II-1	HTZ	c.769G>A	p.Glu257Lys	Caribbean & Irish
12	2012, Falk et al.	UCL-5-II-2	HTZ	c.769G>A	p.Glu257Lys	Polish
12	2012, Falk et al.	UCL-6-II-1	HTZ	c.769G>A	p.Glu257Lys	British (European descent)
12	2012, Falk et al.	UCL-7-II-1	HTZ	c.769G>A	p.Glu257Lys	British (European descent)
23	2014, Siemiatkowska et al.	2	HTZ	c.769G>A	p.Glu257Lys	German
23	2014, Siemiatkowska et al.	3.1	HTZ	c.769G>A	p.Glu257Lys	Dutch
23	2014, Siemiatkowska et al.	4	HTZ	c.769G>A	p.Glu257Lys	Danish
23	2014, Siemiatkowska et al.	5	HTZ	c.769G>A	p.Glu257Lys	German
23	2014, Siemiatkowska et al.	6	HTZ	c.769G>A	p.Glu257Lys	Dutch
25	2015, Coppieters et al.	F5	HTZ	c.769G>A	p.Glu257Lys	NA
25	2015, Coppieters et al.	F6	HTZ	c.769G>A	p.Glu257Lys	NA
25	2015, Coppieters et al.	F7	HTZ	c.769G>A	p.Glu257Lys	NA
28	2016, Bravo-Gil et al.	RP157-II:3	HTZ	c.769G>A	p.Glu257Lys	Spanish
17	2017, Tompson et al.	-	HTZ	c.769G>A	p.Glu257Lys	Australian
17	2017, Tompson et al.	-	HTZ	c.769G>A	p.Glu257Lys	Australian
17	2017, Tompson et al.	-	HTZ	c.769G>A	p.Glu257Lys	Australian
35	2018, Nash et al.	Case 2	HTZ	c.769G>A	p.Glu257Lys	Caucasian
37	2018, Weisschuh, et al.	10	HTZ	c.769G>A	p.Glu257Lys	German
39	2020, Ruberto et al.	1	HTZ	c.769G>A	p.Glu257Lys	Italy
39	2020, Ruberto et al.	16	HMZ	c.769G>A	p.Glu257Lys	Italy
42	2020, Hull et al.	-	HTZ	c.769G>A	p.Glu257Lys	New Zealand
43	2020, Sallum, et al.	61.1	HTZ	c.769G>A	p.Glu257Lys	Brazilian
43	2020, Sallum, et al.	62.1	HTZ	c.769G>A	p.Glu257Lys	Brazilian
43	2020, Sallum, et al.	65.1	HTZ	c.769G>A	p.Glu257Lys	Brazilian
43	2020, Sallum, et al.	66.1	HTZ	c.769G>A	p.Glu257Lys	Brazilian
43	2020, Sallum, et al.	67.1	HTZ	c.769G>A	p.Glu257Lys	Brazilian
44	2020, Skorczyk-Werner et al.	9-29	HTZ	c.769G>A	p.Glu257Lys	Polish
44	2020, Skorczyk-Werner et al.	19-47	HTZ	c.769G>A	p.Glu257Lys	Polish
9	2012, Chiang et al.	4	HTZ	c.817A>G	p.Asn273Asp	Canada
11	2012, Koenekoop et al.	MOGL208	HTZ	c.817A>G	p.Asn273Asp	French Canadian
11	2012, Koenekoop et al.	LCA9 IV:8	HMZ	c.838T>C	p.*280Gln	Pakistani
11	2012, Koenekoop et al.	LCA9 IV:8	HMZ	c.838T>C	p.*280Gln	Pakistani

HMZ: homozygous, HTZ: heterozygous, NA: not available

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