

Analysis of genetic risk factors for Leber hereditary optic neuropathy in the Polish population

Online Resource

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List of Tables

S1	Demographic and molecular data for each individual included in the study.	3
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List of Figures

S1	Representative agarose gel electrophoresis image of <i>Sac</i> II restriction digestion products obtained in the PCR-RFLP assay used for detection of the PRICKLE3 c.157C>T variant.	17
S2	A scatter plot illustrating the relationship between the approximate age of LHON symptom onset and mtCN in symptomatic carriers.	17

Table S1 Demographic and molecular data for each individual included in the study. Quantitative heteroplasmy data for the m.11778G>A variant were available for a subset of individuals analyzed by mtDNA next-generation sequencing and are presented in the ‘m.11778G>A mutation load’ column

ID	Group	Tissue	Age (years)	Sex	m.11778G>A mutation load	mtDNA copy number (mean <i>MT-ND1/B2M</i>)	mtDNA copy number (SD)	Presence of <i>PRICKLE3</i> variant (c.157C>A) (yes/no)
4106	Symptomatic carriers	blood	27	M	99.91	189.85	27.74	no
5529	Symptomatic carriers	blood	17	M	99.90	104.32	14.66	no
6501	Symptomatic carriers	blood	n/a	M	n/a	48.97	8.90	no
6586	Symptomatic carriers	blood	28	M	99.94	148.03	13.46	no
9594	Symptomatic carriers	blood	10	M	72.25	63.48	13.65	no
9922	Symptomatic carriers	blood	35	M	99.72	135.00	4.32	no
10911	Symptomatic carriers	blood	27	M	91.92	88.14	9.75	no
11266	Symptomatic carriers	blood	44	M	99.49	65.04	12.61	no
11922	Symptomatic carriers	blood	21	M	99.89	207.07	27.76	no
13933	Symptomatic carriers	blood	23	M	99.69	136.13	7.38	no
14164	Symptomatic carriers	blood	25	M	99.70	49.62	0.02	no
15046	Symptomatic carriers	blood	18	M	99.19	49.83	3.21	no
15415	Symptomatic carriers	blood	41	M	99.79	71.43	1.26	no
15591	Symptomatic carriers	blood	22	M	99.88	80.66	22.42	no
15745	Symptomatic carriers	blood	18	M	99.66	82.97	10.39	no
15923	Symptomatic carriers	blood	32	M	n/a	50.92	12.10	no

16159	Symptomatic carriers	blood	24	M	99.44	71.59	4.12	no
16170	Symptomatic carriers	blood	14	M	99.79	59.50	7.51	no
16799	Symptomatic carriers	blood	32	M	99.44	44.69	1.64	no
17367	Symptomatic carriers	blood	31	M	99.44	77.03	7.62	no
17545	Symptomatic carriers	blood	39	M	99.44	115.93	10.93	no
17791	Symptomatic carriers	blood	25	M	99.44	165.11	11.06	no
456L	Symptomatic carriers	blood	25	M	99.44	106.55	1.05	no
L1/2002	Symptomatic carriers	blood	24	M	n/a	39.10	0.67	no
L1/2009	Symptomatic carriers	blood	33	M	n/a	69.00	2.15	no
L11/2003	Symptomatic carriers	blood	18	M	n/a	77.03	1.22	no
L11/2012	Symptomatic carriers	blood	16	M	n/a	150.55	4.37	no
L12/2007	Symptomatic carriers	blood	37	M	84.05	120.18	16.08	no
L12/2008	Symptomatic carriers	blood	23	M	99.90	60.92	5.66	no
L12/2012	Symptomatic carriers	blood	n/a	M	99.82	37.07	2.55	no
L13/2006	Symptomatic carriers	blood	28	M	99.79	57.51	0.72	no
L15/2007	Symptomatic carriers	blood	34	M	n/a	46.37	1.45	no
L16/2008	Symptomatic carriers	blood	14	M	99.93	30.61	3.98	no
L17/2012	Symptomatic carriers	blood	21	M	n/a	24.00	0.30	no
L19/2018	Symptomatic carriers	blood	20	M	99.81	52.58	11.97	no
L2/2018	Symptomatic carriers	blood	28	M	99.83	74.60	3.23	no

L20/2007	Symptomatic carriers	blood	44	M	n/a	59.35	4.29	no
L21/2014	Symptomatic carriers	blood	33	M	n/a	142.93	1.24	no
L21/2017	Symptomatic carriers	blood	34	M	99.73	63.97	13.40	no
L23/2008	Symptomatic carriers	blood	31	M	99.60	117.63	0.67	no
L23/2017	Symptomatic carriers	blood	32	M	n/a	85.33	18.08	no
L24/2006	Symptomatic carriers	blood	n/a	M	99.74	77.45	0.26	no
L24/2008	Symptomatic carriers	blood	34	M	n/a	49.29	4.90	no
L24/2014	Symptomatic carriers	blood	53	M	n/a	159.44	9.31	no
L24/2018	Symptomatic carriers	blood	47	M	99.70	65.83	6.65	no
L25/2014	Symptomatic carriers	blood	26	M	n/a	125.81	14.34	no
L26/2002	Symptomatic carriers	blood	19	M	100.00	46.69	2.07	no
L28/2012	Symptomatic carriers	blood	22	M	n/a	122.52	7.95	no
L28/2016	Symptomatic carriers	blood	16	M	99.89	122.35	10.59	no
L29/2005	Symptomatic carriers	blood	n/a	M	99.76	82.30	3.19	no
L31/2006	Symptomatic carriers	blood	39	M	n/a	56.08	2.54	no
L31/2014	Symptomatic carriers	blood	26	M	n/a	90.66	18.28	no
L31/2017	Symptomatic carriers	blood	16	M	n/a	109.48	32.35	no
L32/2006	Symptomatic carriers	blood	30	M	99.74	131.21	29.88	no
L32/2016	Symptomatic carriers	blood	15	M	99.95	48.87	5.84	no
L39/2005	Symptomatic carriers	blood	31	M	n/a	68.55	11.44	no

L39/2008	Symptomatic carriers	blood	40	M	99.95	101.10	1.95	no
L41/2005	Symptomatic carriers	blood	38	M	99.70	141.57	7.53	no
L5/2014	Symptomatic carriers	blood	56	M	99.96	134.49	12.77	no
L5/2021	Symptomatic carriers	blood	16	M	n/a	67.41	5.37	no
L6/2013	Symptomatic carriers	blood	30	M	n/a	86.00	13.17	no
L7/2021	Symptomatic carriers	blood	33	M	n/a	56.67	10.04	no
L8/2019	Symptomatic carriers	blood	33	M	99.49	76.91	8.00	no
L8/2020	Symptomatic carriers	blood	43	M	n/a	77.55	20.07	no
L9/2019	Symptomatic carriers	blood	30	M	99.44	58.20	3.93	no
L17/2016	Symptomatic carriers	blood	35	F	n/a	141.93	6.44	no
L19/2005	Symptomatic carriers	blood	n/a	F	n/a	106.31	0.77	no
L20/2014	Symptomatic carriers	blood	46	F	n/a	80.43	4.78	no
L24/2017	Symptomatic carriers	blood	55	F	n/a	69.32	0.09	no
L10/2019	Asymptomatic carriers	blood	18	M	n/a	74.72	3.96	no
L12/2021	Asymptomatic carriers	blood	21	M	n/a	100.69	7.15	no
L13/2021	Asymptomatic carriers	blood	11	M	n/a	73.96	6.86	no
L30/2008	Asymptomatic carriers	blood	n/a	M	n/a	99.44	23.47	no
L42/2016	Asymptomatic carriers	blood	29	M	n/a	76.68	6.17	no
L7/2018	Asymptomatic carriers	blood	42	M	74.69	70.21	0.04	no
L1/2016	Asymptomatic carriers	blood	43	F	n/a	73.23	4.90	no

L10/2021	Asymptomatic carriers	blood	29	F	n/a	58.47	2.08	no
L11/2019	Asymptomatic carriers	blood	68	F	n/a	78.19	9.97	no
L11/2021	Asymptomatic carriers	blood	45	F	n/a	75.72	3.14	no
L12/2019	Asymptomatic carriers	blood	47	F	n/a	69.33	17.19	no
L14/2019	Asymptomatic carriers	blood	23	F	n/a	78.75	10.47	no
L17/2014	Asymptomatic carriers	blood	32	F	n/a	47.92	7.88	no
L18/2015	Asymptomatic carriers	blood	n/a	F	n/a	87.77	14.11	no
L18/2017	Asymptomatic carriers	blood	47	F	n/a	77.76	16.84	no
L19/2015	Asymptomatic carriers	blood	n/a	F	n/a	125.94	8.81	no
L19/2021	Asymptomatic carriers	blood	24	F	n/a	57.82	7.71	no
L2/2002	Asymptomatic carriers	blood	n/a	F	n/a	33.17	0.94	no
L20/2018	Asymptomatic carriers	blood	45	F	n/a	60.38	1.59	no
L28/2017	Asymptomatic carriers	blood	56	F	n/a	60.86	2.78	no
L29/2016	Asymptomatic carriers	blood	37	F	n/a	71.04	8.05	no
L29/2018	Asymptomatic carriers	blood	21	F	n/a	59.34	5.89	no
L3/2014	Asymptomatic carriers	blood	25	F	n/a	61.27	6.28	no
L30/2018	Asymptomatic carriers	blood	63	F	n/a	69.51	8.63	no
L31/2008	Asymptomatic carriers	blood	n/a	F	n/a	77.86	4.05	no
L32/2018	Asymptomatic carriers	blood	40	F	n/a	65.36	7.14	no
L37/2016	Asymptomatic carriers	blood	37	F	n/a	137.84	31.60	no

L41/2016	Asymptomatic carriers	blood	63	F	n/a	85.39	1.22	no
L6/2015	Asymptomatic carriers	blood	n/a	F	n/a	63.97	7.15	no
L8/2018	Asymptomatic carriers	blood	68	F	n/a	60.06	8.42	no
L8/2021	Asymptomatic carriers	blood	58	F	n/a	82.71	1.42	no
K101	Control group for mtCN	blood	21	M	n/a	63.98	1.42	n/a
K105	Control group for mtCN	blood	21	M	n/a	65.58	9.98	n/a
K106	Control group for mtCN	blood	21	M	n/a	93.94	14.21	n/a
K107	Control group for mtCN	blood	21	M	n/a	69.68	4.26	n/a
K119	Control group for mtCN	blood	23	M	n/a	70.47	7.51	n/a
K122	Control group for mtCN	blood	21	M	n/a	53.01	8.49	n/a
K124	Control group for mtCN	blood	22	M	n/a	52.29	5.79	n/a
K125	Control group for mtCN	blood	22	M	n/a	67.64	2.37	n/a
K126	Control group for mtCN	blood	23	M	n/a	96.78	0.00	n/a
K127	Control group for mtCN	blood	24	M	n/a	70.83	12.56	n/a
K133	Control group for mtCN	blood	23	M	n/a	66.02	0.75	n/a
K134	Control group for mtCN	blood	23	M	n/a	67.31	7.45	n/a
K136	Control group for mtCN	blood	23	M	n/a	77.03	7.17	n/a
K137	Control group for mtCN	blood	23	M	n/a	93.64	3.19	n/a
K140	Control group for mtCN	blood	23	M	n/a	69.29	5.02	n/a
K141	Control group for mtCN	blood	23	M	n/a	58.93	4.06	n/a

K147	Control group for mtCN	blood	23	M	n/a	83.49	3.52	n/a
K154	Control group for mtCN	blood	22	M	n/a	100.74	20.21	n/a
K156	Control group for mtCN	blood	22	M	n/a	95.89	18.53	n/a
K196	Control group for mtCN	blood	24	M	n/a	87.28	1.02	n/a
K202	Control group for mtCN	blood	23	M	n/a	123.01	6.57	n/a
K216	Control group for mtCN	blood	21	M	n/a	63.28	6.79	n/a
K217	Control group for mtCN	blood	22	M	n/a	91.46	24.03	n/a
K218	Control group for mtCN	blood	22	M	n/a	107.22	12.09	n/a
K219	Control group for mtCN	blood	22	M	n/a	103.90	24.69	n/a
K220	Control group for mtCN	blood	23	M	n/a	73.33	0.87	n/a
K221	Control group for mtCN	blood	22	M	n/a	95.26	11.26	n/a
K222	Control group for mtCN	blood	25	M	n/a	97.60	12.15	n/a
K223	Control group for mtCN	blood	22	M	n/a	83.06	5.46	n/a
K224	Control group for mtCN	blood	22	M	n/a	92.79	9.88	n/a
K229	Control group for mtCN	blood	22	M	n/a	91.73	13.52	n/a
K232	Control group for mtCN	blood	21	M	n/a	109.30	4.28	n/a
K238	Control group for mtCN	blood	21	M	n/a	103.26	10.27	n/a
K243	Control group for mtCN	blood	25	M	n/a	82.21	4.32	n/a
K246	Control group for mtCN	blood	21	M	n/a	101.08	18.81	n/a
K247	Control group for mtCN	blood	21	M	n/a	70.03	1.77	n/a

K248	Control group for mtCN	blood	21	M	n/a	66.02	0.09	n/a
K249	Control group for mtCN	blood	21	M	n/a	116.56	8.35	n/a
K250	Control group for mtCN	blood	21	M	n/a	84.15	9.31	n/a
K251	Control group for mtCN	blood	21	M	n/a	106.09	4.65	n/a
K252	Control group for mtCN	blood	22	M	n/a	91.55	2.78	n/a
K256	Control group for mtCN	blood	23	M	n/a	115.51	25.73	n/a
K258	Control group for mtCN	blood	22	M	n/a	92.42	7.30	n/a
K259	Control group for mtCN	blood	21	M	n/a	89.18	1.71	n/a
K260	Control group for mtCN	blood	21	M	n/a	79.65	7.88	n/a
K262	Control group for mtCN	blood	21	M	n/a	63.39	3.85	n/a
K263	Control group for mtCN	blood	21	M	n/a	73.83	3.60	n/a
K270	Control group for mtCN	blood	22	M	n/a	70.09	1.37	n/a
K271	Control group for mtCN	blood	21	M	n/a	110.51	6.70	n/a
K272	Control group for mtCN	blood	22	M	n/a	76.26	6.12	n/a
K274	Control group for mtCN	blood	21	M	n/a	69.19	3.86	n/a
K277	Control group for mtCN	blood	21	M	n/a	108.44	11.65	n/a
K283	Control group for mtCN	blood	22	M	n/a	63.86	7.51	n/a
K291	Control group for mtCN	blood	20	M	n/a	77.27	9.58	n/a
K292	Control group for mtCN	blood	21	M	n/a	71.89	3.11	n/a
K297	Control group for mtCN	blood	20	M	n/a	102.62	15.06	n/a

K298	Control group for mtCN	blood	20	M	n/a	75.41	10.04	n/a
K3	Control group for mtCN	blood	22	M	n/a	70.97	0.45	n/a
K301	Control group for mtCN	blood	20	M	n/a	100.36	11.86	n/a
K309	Control group for mtCN	blood	21	M	n/a	89.23	9.12	n/a
K318	Control group for mtCN	blood	20	M	n/a	69.24	3.64	n/a
K336	Control group for mtCN	blood	20	M	n/a	73.54	0.07	n/a
K5	Control group for mtCN	blood	22	M	n/a	71.82	0.17	n/a
K70	Control group for mtCN	blood	23	M	n/a	79.69	10.77	n/a
K87	Control group for mtCN	blood	22	M	n/a	53.73	1.26	n/a
K88	Control group for mtCN	blood	21	M	n/a	64.86	8.54	n/a
K91	Control group for mtCN	blood	23	M	n/a	81.21	9.95	n/a
K98	Control group for mtCN	blood	21	M	n/a	64.26	0.18	n/a
KJ101	Control group for <i>PRICKLE3</i>	blood	77	F	n/a	n/a	n/a	no
KJ102	Control group for <i>PRICKLE3</i>	blood	74	F	n/a	n/a	n/a	no
KJ103	Control group for <i>PRICKLE3</i>	blood	72	F	n/a	n/a	n/a	no
KJ104	Control group for <i>PRICKLE3</i>	blood	60	M	n/a	n/a	n/a	no
KJ105	Control group for <i>PRICKLE3</i>	blood	69	F	n/a	n/a	n/a	no
KJ106	Control group for <i>PRICKLE3</i>	blood	56	M	n/a	n/a	n/a	no
KJ107	Control group for <i>PRICKLE3</i>	blood	85	F	n/a	n/a	n/a	no
KJ108	Control group for <i>PRICKLE3</i>	blood	80	F	n/a	n/a	n/a	no

KJ109	Control group for <i>PRICKLE3</i>	blood	83	F	n/a	n/a	n/a	no
KJ11	Control group for <i>PRICKLE3</i>	blood	66	F	n/a	n/a	n/a	no
KJ110	Control group for <i>PRICKLE3</i>	blood	61	F	n/a	n/a	n/a	no
KJ111	Control group for <i>PRICKLE3</i>	blood	79	F	n/a	n/a	n/a	no
KJ112	Control group for <i>PRICKLE3</i>	blood	65	M	n/a	n/a	n/a	no
KJ113	Control group for <i>PRICKLE3</i>	blood	70	F	n/a	n/a	n/a	no
KJ114	Control group for <i>PRICKLE3</i>	blood	72	F	n/a	n/a	n/a	no
KJ115	Control group for <i>PRICKLE3</i>	blood	77	F	n/a	n/a	n/a	no
KJ117	Control group for <i>PRICKLE3</i>	blood	76	M	n/a	n/a	n/a	no
KJ119	Control group for <i>PRICKLE3</i>	blood	73	F	n/a	n/a	n/a	no
KJ12	Control group for <i>PRICKLE3</i>	blood	60	M	n/a	n/a	n/a	no
KJ120	Control group for <i>PRICKLE3</i>	blood	79	M	n/a	n/a	n/a	no
KJ121	Control group for <i>PRICKLE3</i>	blood	61	M	n/a	n/a	n/a	no
KJ122	Control group for <i>PRICKLE3</i>	blood	70	F	n/a	n/a	n/a	no
KJ123	Control group for <i>PRICKLE3</i>	blood	57	M	n/a	n/a	n/a	no
KJ124	Control group for <i>PRICKLE3</i>	blood	67	F	n/a	n/a	n/a	no
KJ125	Control group for <i>PRICKLE3</i>	blood	65	F	n/a	n/a	n/a	no
KJ126	Control group for <i>PRICKLE3</i>	blood	65	F	n/a	n/a	n/a	no
KJ127	Control group for <i>PRICKLE3</i>	blood	84	F	n/a	n/a	n/a	no
KJ128	Control group for <i>PRICKLE3</i>	blood	69	F	n/a	n/a	n/a	no

KJ129	Control group for <i>PRICKLE3</i>	blood	65	M	n/a	n/a	n/a	no
KJ13	Control group for <i>PRICKLE3</i>	blood	52	M	n/a	n/a	n/a	no
KJ130	Control group for <i>PRICKLE3</i>	blood	70	F	n/a	n/a	n/a	no
KJ131	Control group for <i>PRICKLE3</i>	blood	66	M	n/a	n/a	n/a	no
KJ14	Control group for <i>PRICKLE3</i>	blood	72	M	n/a	n/a	n/a	no
KJ15	Control group for <i>PRICKLE3</i>	blood	71	M	n/a	n/a	n/a	no
KJ153	Control group for <i>PRICKLE3</i>	blood	n/a	M	n/a	n/a	n/a	no
KJ16	Control group for <i>PRICKLE3</i>	blood	80	F	n/a	n/a	n/a	no
KJ17	Control group for <i>PRICKLE3</i>	blood	n/a	F	n/a	n/a	n/a	no
KJ18	Control group for <i>PRICKLE3</i>	blood	56	F	n/a	n/a	n/a	no
KJ19	Control group for <i>PRICKLE3</i>	blood	78	F	n/a	n/a	n/a	no
KJ2	Control group for <i>PRICKLE3</i>	blood	84	M	n/a	n/a	n/a	no
KJ21	Control group for <i>PRICKLE3</i>	blood	76	M	n/a	n/a	n/a	no
KJ22	Control group for <i>PRICKLE3</i>	blood	82	M	n/a	n/a	n/a	no
KJ23	Control group for <i>PRICKLE3</i>	blood	69	M	n/a	n/a	n/a	no
KJ26	Control group for <i>PRICKLE3</i>	blood	63	M	n/a	n/a	n/a	no
KJ27	Control group for <i>PRICKLE3</i>	blood	84	F	n/a	n/a	n/a	no
KJ28	Control group for <i>PRICKLE3</i>	blood	71	F	n/a	n/a	n/a	no
KJ29	Control group for <i>PRICKLE3</i>	blood	80	F	n/a	n/a	n/a	no
KJ30	Control group for <i>PRICKLE3</i>	blood	53	F	n/a	n/a	n/a	no

KJ32	Control group for <i>PRICKLE3</i>	blood	82	F	n/a	n/a	n/a	no
KJ33	Control group for <i>PRICKLE3</i>	blood	65	F	n/a	n/a	n/a	no
KJ34	Control group for <i>PRICKLE3</i>	blood	68	F	n/a	n/a	n/a	no
KJ35	Control group for <i>PRICKLE3</i>	blood	83	F	n/a	n/a	n/a	no
KJ36	Control group for <i>PRICKLE3</i>	blood	74	F	n/a	n/a	n/a	no
KJ37	Control group for <i>PRICKLE3</i>	blood	60	F	n/a	n/a	n/a	no
KJ38	Control group for <i>PRICKLE3</i>	blood	64	M	n/a	n/a	n/a	no
KJ39	Control group for <i>PRICKLE3</i>	blood	61	F	n/a	n/a	n/a	no
KJ40	Control group for <i>PRICKLE3</i>	blood	81	M	n/a	n/a	n/a	no
KJ41	Control group for <i>PRICKLE3</i>	blood	67	F	n/a	n/a	n/a	no
KJ42	Control group for <i>PRICKLE3</i>	blood	76	F	n/a	n/a	n/a	no
KJ43	Control group for <i>PRICKLE3</i>	blood	85	F	n/a	n/a	n/a	no
KJ44	Control group for <i>PRICKLE3</i>	blood	80	F	n/a	n/a	n/a	no
KJ45	Control group for <i>PRICKLE3</i>	blood	83	M	n/a	n/a	n/a	no
KJ46	Control group for <i>PRICKLE3</i>	blood	84	F	n/a	n/a	n/a	no
KJ47	Control group for <i>PRICKLE3</i>	blood	85	M	n/a	n/a	n/a	no
KJ48	Control group for <i>PRICKLE3</i>	blood	82	F	n/a	n/a	n/a	no
KJ49	Control group for <i>PRICKLE3</i>	blood	77	M	n/a	n/a	n/a	no
KJ50	Control group for <i>PRICKLE3</i>	blood	88	F	n/a	n/a	n/a	no
KJ51	Control group for <i>PRICKLE3</i>	blood	82	F	n/a	n/a	n/a	no

KJ53	Control group for <i>PRICKLE3</i>	blood	64	F	n/a	n/a	n/a	no
KJ54	Control group for <i>PRICKLE3</i>	blood	78	F	n/a	n/a	n/a	no
KJ55	Control group for <i>PRICKLE3</i>	blood	77	M	n/a	n/a	n/a	no
KJ56	Control group for <i>PRICKLE3</i>	blood	61	M	n/a	n/a	n/a	no
KJ58	Control group for <i>PRICKLE3</i>	blood	62	F	n/a	n/a	n/a	no
KJ6	Control group for <i>PRICKLE3</i>	blood	71	M	n/a	n/a	n/a	no
KJ60	Control group for <i>PRICKLE3</i>	blood	66	F	n/a	n/a	n/a	no
KJ61	Control group for <i>PRICKLE3</i>	blood	78	M	n/a	n/a	n/a	no
KJ63	Control group for <i>PRICKLE3</i>	blood	87	M	n/a	n/a	n/a	no
KJ64	Control group for <i>PRICKLE3</i>	blood	72	F	n/a	n/a	n/a	no
KJ65	Control group for <i>PRICKLE3</i>	blood	69	F	n/a	n/a	n/a	no
KJ66	Control group for <i>PRICKLE3</i>	blood	83	M	n/a	n/a	n/a	no
KJ67	Control group for <i>PRICKLE3</i>	blood	62	M	n/a	n/a	n/a	no
KJ68	Control group for <i>PRICKLE3</i>	blood	68	M	n/a	n/a	n/a	no
KJ69	Control group for <i>PRICKLE3</i>	blood	65	F	n/a	n/a	n/a	no
KJ7	Control group for <i>PRICKLE3</i>	blood	70	M	n/a	n/a	n/a	no
KJ70	Control group for <i>PRICKLE3</i>	blood	84	M	n/a	n/a	n/a	no
KJ71	Control group for <i>PRICKLE3</i>	blood	63	F	n/a	n/a	n/a	no
KJ72	Control group for <i>PRICKLE3</i>	blood	68	F	n/a	n/a	n/a	no
KJ75	Control group for <i>PRICKLE3</i>	blood	69	M	n/a	n/a	n/a	no

KJ76	Control group for <i>PRICKLE3</i>	blood	72	M	n/a	n/a	n/a	no
KJ77	Control group for <i>PRICKLE3</i>	blood	n/a	F	n/a	n/a	n/a	no
KJ79	Control group for <i>PRICKLE3</i>	blood	n/a	M	n/a	n/a	n/a	no
KJ8	Control group for <i>PRICKLE3</i>	blood	64	M	n/a	n/a	n/a	no
KJ89	Control group for <i>PRICKLE3</i>	blood	n/a	F	n/a	n/a	n/a	no
KJ9	Control group for <i>PRICKLE3</i>	blood	71	F	n/a	n/a	n/a	no

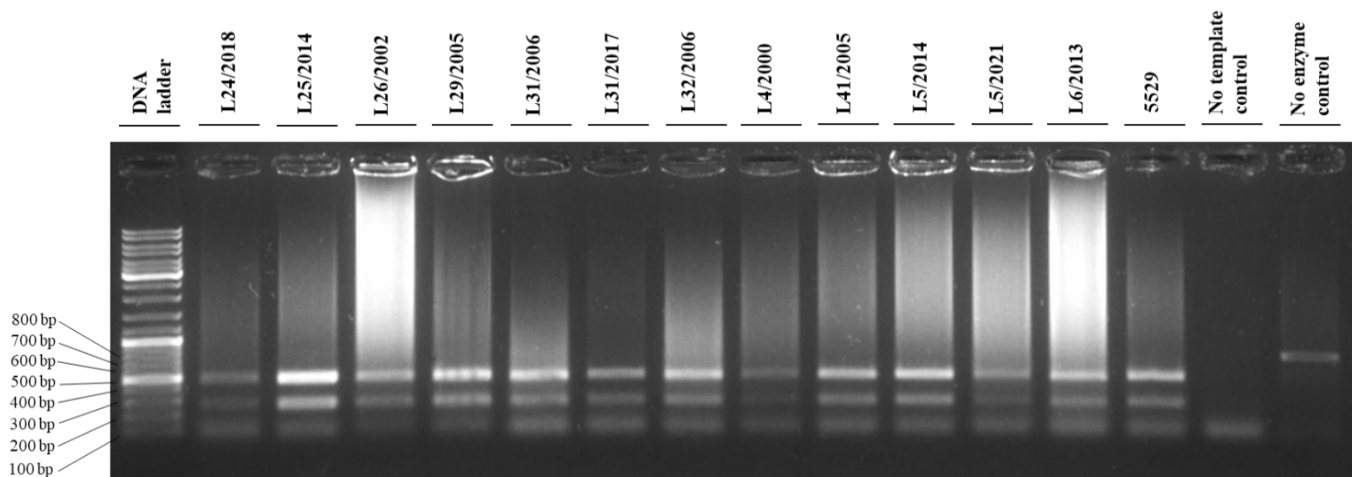


Fig. S1 Representative agarose gel electrophoresis image of *SacII* restriction digestion products obtained in the PCR-RFLP assay used for detection of the *PRICKLE3* c.157C>T variant. DNA isolated from peripheral blood of 13 symptomatic male carriers of the m.11778G>A variant was analyzed. The c.157C>T substitution abolishes the *SacII* restriction site; therefore, in the presence of the variant, an undigested 724 bp fragment would be observed. In wild-type samples, the PCR product is cleaved into two fragments of 475 bp and 249 bp. All analyzed samples showed the wild-type digestion pattern. A low-molecular-weight band visible in all lanes, including the NTC, most likely represents primer dimers. NTC, no template control – negative control without DNA template; No enzyme control – undigested PCR product control

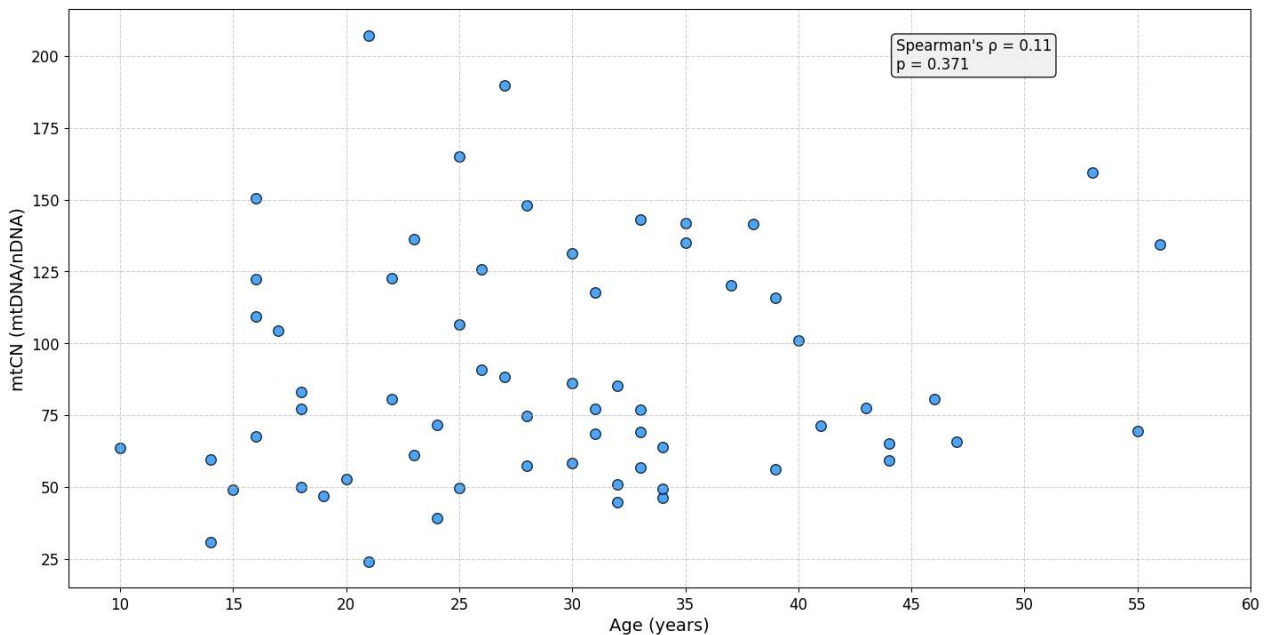


Fig. S2 A scatter plot illustrating the relationship between the approximate age of LHON symptom onset and mtCN in symptomatic carriers. Spearman's rank correlation test revealed a correlation coefficient ρ of 0.114 with a p -value of 0.371, indicating no statistically significant association