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Supplementary Table 1. Data summary of previously published LHON patients with onset of visual loss ≤ 12 years of age.

Mutation	Patients	Sex			Age at onset (y)	BCVA [#]		
	n	f	m	m:f	Mean Median	Mean Median	≥ 0.5 (%)	< 0.05 (%)
11778	47	18	29	1.6 (3.9)*	8.3 8.0	0.15 0.10	4/59 (7)	18/59 (31)
3460	11	4	7	1.8	8.6 8.0	0.74 0.10	4/6 (67)	0/6 (0)
14484	11	6	5	0.8	9.2 9.0	0.82 0.80	10/14 (71)	0/14 (0)
All	69	28	41	1.5 (2.3)*	8.5 8.0	0.30 0.12	18/79 (23)	18/79 (23)

*Ratio after excluding two atypical families carrying the m.11778A>G mutation, one German and one Chinese, with a high proportion of affected female carriers.

Number of eyes with best-corrected visual acuity (BCVA) ≥ 0.5 or < 0.05 in Snellen decimal. One eye of a child carrying the m.11778G>A mutation was excluded from analysis due to unilateral anterior persistent faetal vasculature and pre-existing dense amblyopia.

Abbreviations: BCVA, best corrected visual acuity; f, female; m, male.

Supplementary Table 2. Meta-analysis of childhood-onset LHON patients.

Mutation	Patients	Sex			Age at onset (y)	BCVA [#]		
	n	f	m	m:f	Mean Median	Mean Median	≥ 0.5 (%)	< 0.05 (%)
11778	60	21	39	1.9	8.0 8.0	0.16 0.10	9/85 (11)	27/85 (32)
3460	18	6	12	2.0	8.17.7	0.63 0.73	12/20 (60)	2/20 (10)
14484	18	8	10	1.3	8.18.0	0.67 0.80	17/28 (61)	0/28 (0)
All	96	34	62	1.8	8.08.0	0.34 0.17	38/133 (29)	29/133 (22)

Number of eyes with best-corrected visual acuity (BCVA) ≥ 0.5 or < 0.05 in Snellen decimal.

Abbreviations: BCVA, best corrected visual acuity; f, female; m, male.

Supplementary appendix

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