



Table S1. Results of the macular and optic nerve head optical coherence tomography imaging.

Retinal layer	Layer thickness (μm) (mean \pm SD)			
	Affected LHON (n=26 eyes)	p *	Unaffected LHON (n=18 eyes)	Control (n=48 eyes)
Macula				
Retina	306.3 \pm 24.8	<0.001	349.0 \pm 8.5	340.8 \pm 13.3
RNFL	19.2 \pm 2.8	<0.001	24.8 \pm 1.1	24.2 \pm 2.1
GCL-IPL	47.1 \pm 14.6	<0.001	94.9 \pm 3.4	93.5 \pm 7.8
INL	45.1 \pm 4.7	<0.001	39.2 \pm 2.3	39.6 \pm 3.5
OPL	33.0 \pm 8.2	0.955	34.9 \pm 4.7	32.3 \pm 4.0
OPL-ONL	110.9 \pm 10.0	<0.001	107.4 \pm 8.1	102.0 \pm 8.6
Outer retina	83.9 \pm 3.4	0.002	82.9 \pm 2.6	81.5 \pm 2.7
Peripapillary RNFL				
Inferotemporal	83.2 \pm 47.3		152.4 \pm 18.6	147**
Temporal	33.4 \pm 17.2		72.7 \pm 2.5	78**
Superotemporal	73.5 \pm 46.0		122.3 \pm 20.4	138**
Superonasal	77.1 \pm 35.2		111.4 \pm 30.8	102**
Nasal	54.0 \pm 21.1		83.3 \pm 13.7	72**
Inferonasal	79.7 \pm 35.0		126.4 \pm 26.4	108**
Average	66.8 \pm 30.0		111.4 \pm 12.4	108**

Abbreviations: GCL-IPL, ganglion cell layer - inner plexiform layer complex; INL, inner nuclear layer; LHON, Leber hereditary optic neuropathy; OPL, outer plexiform layer; OPL-ONL, outer plexiform layer - outer nuclear layer complex; RNFL, retinal nerve fiber layer.

* Mann-Whitney *U* Test

** Heidelberg OCT normative mean

Table S2. L-cone critical flicker fusion variables.

Observer	L-cone				
	The lowest radiance (log ₁₀ quanta s ⁻¹ deg ⁻²)	Ferry-Porter slope Per decade R ²		Frequency (Hz) at 8.5 log ₁₀ quanta s ⁻¹ deg ⁻² radiance	Plateau frequency (Hz) at 10.3-10.6 log ₁₀ quanta s ⁻¹ deg ⁻² radiance
Normal mean±SE	6.65±0.06	9.16±0.35	0.998	27.90±0.91	40.18±0.88
LHON					
A1	10.00	0.295	0.017	ND	11.60
A2	8.99	2.873	0.933	ND	13.00
A3	9.71	4.594	0.931	ND	6.78
A4	10.63	ND	ND	ND	8.92
A6	10.37	ND	ND	ND	16.00
A7	8.05	2.914	0.762	ND	19.67
A9	8.45	10.847	0.896	ND	21.84
A10	10.94	ND	ND	ND	0.67
A11					
LHON mean±SE	9.64±0.43	4.30±1.59	0.71±0.18		12.31±2.84
Mann-Whitney U test, p	<0.001	<0.001			<0.001
Carrier					
U1	6.89	8.62	0.99	26.08	41.89
U2	7.46	5.37	0.97	19.32	29.11
U3	7.21	7.85	0.99	17.02	33.17
U4	5.42	9.24	0.92	24.13	37.22
U5	7.21	8.20	0.94	21.34	34.80
U6	7.16	6.55	0.93	18.83	30.72
U7	7.43	9.30	0.99	19.07	28.89
U8	6.84	8.00	0.99	27.21	39.56
U9	7.18	7.17	0.95	19.48	32.89
All carriers Mean±SE	6.98±0.19	7.81±0.43	0.96±0.01	21.39±1.19	34.25±1.43
Mann-Whitney U test, p	0.004	0.035*		0.001*	0.004*

Abbreviation: LHON, Leber hereditary optic neuropathy; ND, not determined.

Table S3. S-cone critical flicker fusion variables.

Observer	S-cone				
	The lowest radiance (log ₁₀ quanta s ⁻¹ deg ⁻²)	Ferry-Porter slope Per decade	R ²	Frequency (Hz) at 8.5 log ₁₀ quanta s ⁻¹ deg ⁻² radiance	Plateau frequency (Hz) at 9.2-9.4 log ₁₀ quanta s ⁻¹ deg ⁻² radiance
Normal	6.77±0.09	7.28±0.29	0.998	18.99±0.89	23.88±0.98
Affected LHON					
A7	8.13	2.20	0.8219	ND	10.44
A9	8.14	0.1074	0.055	ND	7.63
All affected	8.14±0.00	4.30±1.59			9.04±0.53
Unaffected					
U1	7.44	10.12	0.99	20.50	26.50
U2	7.11	7.27	0.91	18.80	22.25
U3	9.31	2.57	0.70	1.06	3.39
U4	7.05	9.66	0.95	16.07	22.17
U5	7.18	6.14	0.98	17.15	22.33
U6	6.89	6.30	0.94	17.93	21.11
U7	7.44	6.97	0.98	14.62	18.33
U8	7.42	5.64	0.98	10.33	16.11
U9 TC	6.91	6.39	0.91	15.86	23.28
All unaffected	7.42±0.23	6.78±0.74	0.95±0.01	14.70±1.95	19.50±2.11
Mann-Whitney U test, p	0.001*	0.411		0.055	0.049

Abbreviation: LHON, Leber hereditary optic neuropathy; ND, not determined.