

Table E1. Visual outcome in 44 LHON subjects with the m.11778G>A mutation

	Recovery definition: Improvement from baseline of at least 15 letters		Worsening definition: Losing from baseline of at least 15 letters		Stabilization definition: gaining < 15 letters or losing < 15 letters from baseline	
88 Eyes 44 Patients	Eyes n (%)	Patients n (%) Recovery in at least one eye	Eyes n (%)	Patients n (%) Worsening in at least one eye	Eyes n (%)	Patients n (%) Stabilization in at least one eye
YES	13 (15%)	8 (18%)	7 (8%)	6 (14%)	68 (77%)	38 (86%)
NO	75 (85%)	36 (82%)	81 (92%)	38 (86%)	20 (23%)	6 (14%)

Source: Lam et al. 2014

Table E2. Population with vision loss \leq 1 year and with a follow-up of at least 24 months (from 24 months to 36 months)

N=9	Recovery definition: Improvement from baseline of at least 15 letters	Worsening definition: Losing from baseline of at least 15 letters	Stabilization definition: gaining < 15 letters or losing < 15 letters from baseline
	Patients n (%) Recovery in at least one eye	Patients n (%) Worsening in at least one eye	Patients n (%) Stabilization in at least one eye
All population (N=9)			
Number	2 / 9 (22%)	4 / 9 (44%)	7 / 9 (78%)
Patient Numbers	14, 53	34, 39, 40, 58	2, 5, 34, 39, 53, 58, 59
Subjects \geq 15 years old (N=8)			
Number	1/8 (12.5%)	4/8 (50%)	7/8 (87.5%)
Patient Numbers	53	34, 39, 40, 58	2, 5, 34, 39, 53, 58, 59

Source: Lam et al. 2014

Table E3. Visual outcomes in LHON patients with age of onset below 18 by age category

	Age of onset (years)				
	< 10	10-12	12-14	15-17	\geq 18
N (patients)	3	5	7	12	48
Number of eyes	6	10	14	24	
Average Visual Acuity	20/200-20/400	20/400-20/1000	20/100-20/200	20/400-20/1000	20/400-20/1000

Source: van Senus 1963 after molecular confirmation using Howell 2003
Original visual acuities were changed into Snellen acuities

Table E4. Visual outcomes in LHON patients with age of onset below 18 years by age category

		Age of onset (years)				
		<10	<12	12-14	15-17	All <18
N (patients)		3	9	11	22	42
Patients with good outcome	N	3	7	3	1	11
	%	100	78	27	5	26

Sources: Newman et al. 1991, Hotta et al. 1995

Good outcome was defined as vision better than 20/200 in at least one eye.

Table E5. Visual outcomes in LHON patients with age of onset below 12 years

Mutation	Number of Patients	Mean age of onset (years)*	Mean BCVA	Number of Eyes with		
				Visual Recovery	BCVA \geq 0.5	BCVA < 0.05
3 Primary	27	6.8	0.20	20/54	20/54	11/54
m.11778G>A	13	6.9	0.39	6/26	5/26	9/26

Source: Majander et al. 2017

* All reported patients had LHON onset \leq 12 years old.