

On-line Table 1: Demographic, clinical, and genetic features of the patients with OPA1-DOA and LHON and healthy subjects studied

	Sex	Age (yr)	Age at Onset (yr)	Disease Duration (yr)	Extravascular Symptoms	Idebenone Therapy	Visual Recovery	Mutation ^a	Haplogroup
Patients with OPA1-DOA									
1	M	60	6	54	None	No	No	c.2729 T>A (Val910Asp)	H
2	M	54	6	48	None	No	No	c.2729 T>A (Val910Asp)	H
3	M	54	30	24	None	Yes	No	c.703 C>T (Arg235X)	J
4	F	43	0	43	Deafness, PNP	No	No	c.893G>A (Ser298Asn)	H
5	M	42	5	37	None	Yes	No	c.984 + 3A>T	L
6	M	40	4	27	CPEO, deafness, PNP	Yes	No	c.1316G>T (Gly439Val)	HV
7	F	30	16	14	Deafness	Yes	No	c.869 G>A p.Arg290Gln	T
8	F	42	0	42	None	No	No	c.2823_26delAGTT	H
9	M	21	6	15	None	Yes	No	c.2823_26 delAGTT p.Lys941fsX966	H
10	M	25	6	19	None	No	No	c.2729 T>A (Val910Asp)	J
11	F	38	6	32	None	No	No	c.1770 G>A; splice defect	HV
12	F	39	6	34	None	Yes	No	c.2819-2 A>C; splice defect	H
13	F	33	3	30	None	Yes	No	c.2819-2 A>C; splice defect	H
14	M	46	0	46	None	No	No	c.984 + 3 A> T; splice defect	H
15	M	43	0	43	None	No	No	c.2196_2197dup AG > AC (×22)	NA
16	F	50	ND	ND	None	No	No	c.1669 C>T (R577X)	H
17	M	24	6	18	None	Yes	No	c.1669 C>T (R577X)	H
18	F	25	1	24	None	No	No	c.1778 T>C; p.Leu593Pro	H
19	M	23	6	17	None	Yes	No	c.1807G>C; p.Asp603His	U
Mean	—	39 ± 12	6 ± 8	31 ± 13	—	—	—	—	—
Patients with LHON									
1	M	27	16	11	None	Yes	No	3460_ND1	H
2	M	29	17	12	None	Yes	Yes	14484_ND6	X
3	M	31	14	17	None	No	No	11778_ND4	J
4	F	42	34	8	None	Yes	Yes	11778_ND4	W
5	M	29	28	30	None	Yes	No	11778_ND4	H
6	M	33	26	33	None	Yes	No	11778_ND4	H
7	F	42	34	8	None	No	No	11778_ND4	H
8	F	45	35	10	None	No	No	11778_ND4	H
9	M	29	14	15	None	Yes	Yes	11778_ND4	H
10	M	35	26	9	None	Yes	No	11778_ND4	H
11	M	26	14	12	None	Yes	Yes	3460_ND1	H
12	M	48	16	32	None	No	No	11778_ND4	K
13	M	53	16	37	None	No	No	11778_ND4	N
14	F	55	12	43	None	Yes	No	3460_ND1	K
15	F	43	30	13	None	Yes	No	11778_ND4	U
16	M	43	6	37	None	No	No	11778_ND4	U
17	M	25	24	1	None	Yes	No	11778_ND4	NA
Mean	—	37 ± 10	21 ± 9	19 ± 13	—	—	—	—	—
Healthy controls (19)	8:11 (M/F)	37 ± 10	—	—	—	—	—	—	—

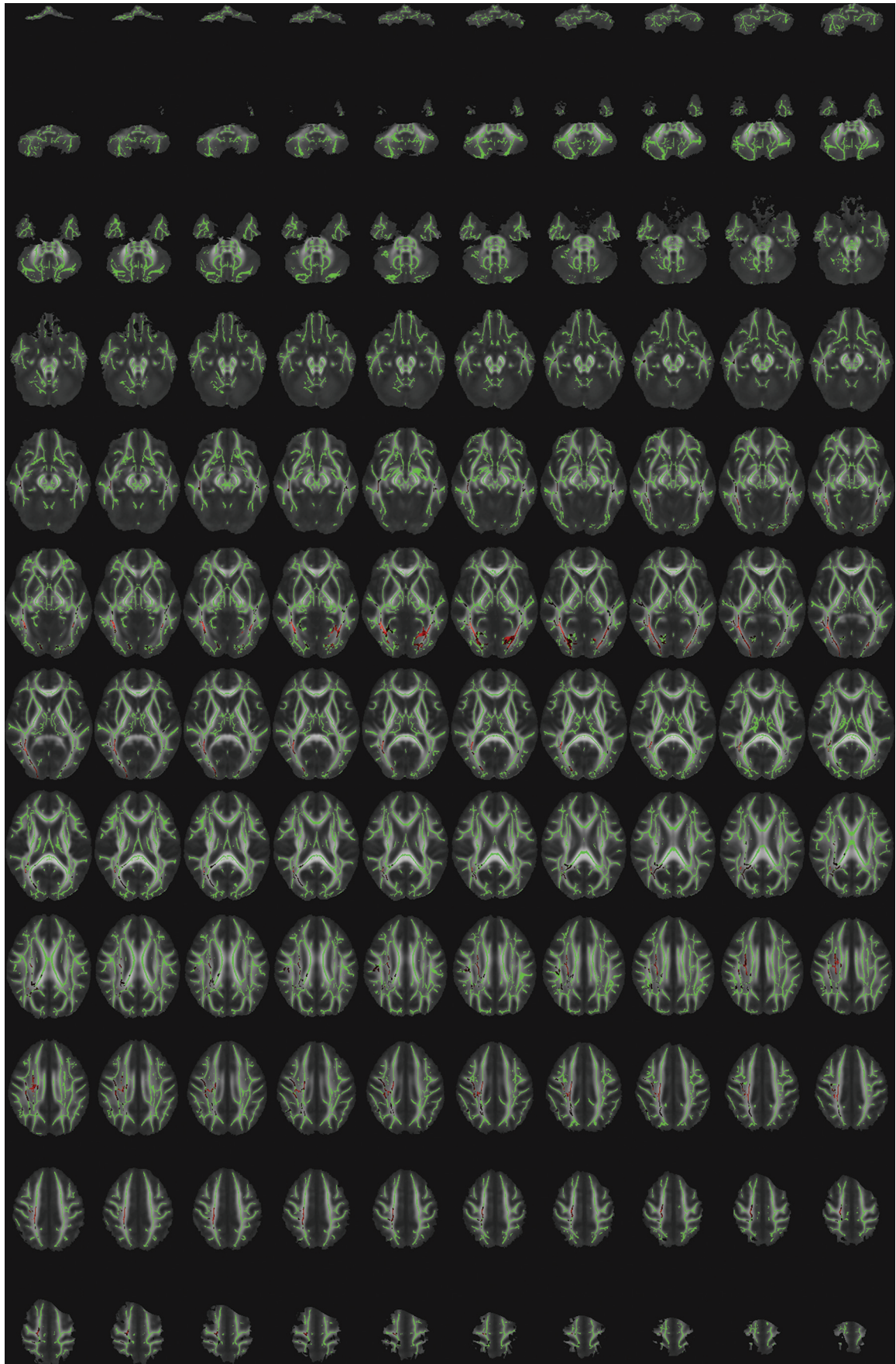
Note.—CPEO indicates chronic progressive external ophthalmoplegia; PNP, polynuropathy (confirmed by nerve-conduction study); ND, not definable (because mild optic atrophy was diagnosed by a routine ophthalmologic evaluation); NA, not available; —, not applicable.

^a Missense mutations are reported in italics, while mutations leading to haploinsufficiency are reported in Roman type.

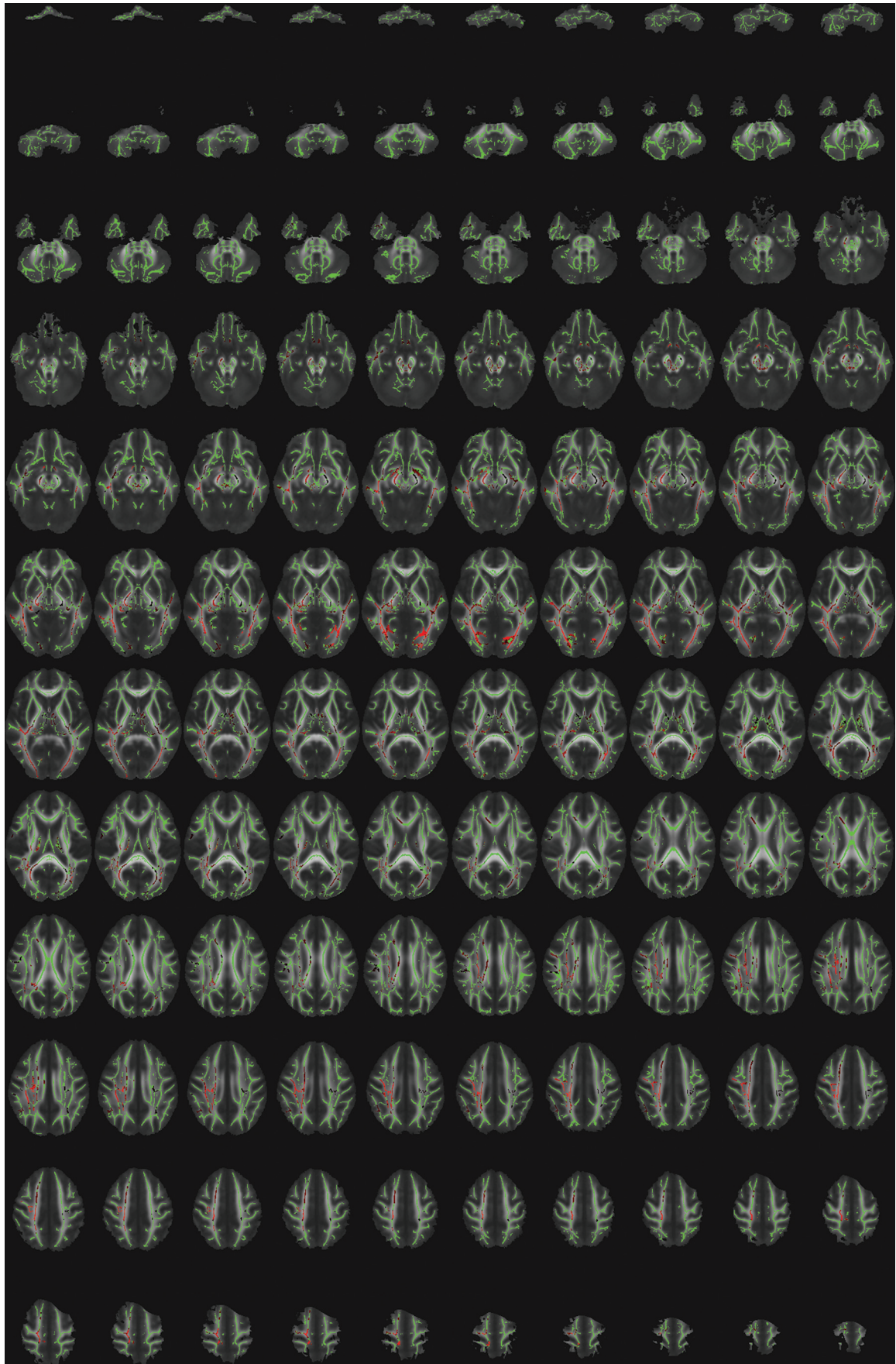
On-line Table 2: Ophthalmologic characteristics of the patients studied with OPAI-DOA and LHON

	Visual Acuity		Right Eye Retinal Nerve Fiber Layer Thickness (μm)				Left Eye Retinal Nerve Fiber Layer Thickness (μm)					
	Right Eye	Left Eye	Average	Temporal	Superior	Nasal	Inferior	Average	Temporal	Superior	Nasal	Inferior
Patients with OPAI-DOA												
1	0.2	0.6	56.2	34	65	51	74	61.93	35	85	47	80
2	0.1	0.05	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
3	0.63	0.2	58.13	34	80	54	65	53.67	31	76	51	57
4	0.05	0.05	43.02	25	56	39	52	46.14	39	58	43	44
5	0.01	0.06	53.71	47	42	49	77	57.16	44	71	36	78
6	0.05	0.1	51	59	44	33	68	NA	NA	NA	NA	NA
7	0.4	0.3	60.76	38	67	42	96	56.26	32	62	40	91
8	0.63	0.63	68.21	29	90	74	80	70.09	31	93	67	89
9	0.12	0.12	59.02	38	88	50	60	54.23	21	84	54	58
10	0.15	0.12	68.31	39	83	56	95	70.7	35	113	47	88
11	0.25	0.2	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
12	0.32	0.4	82.71	31	101	105	93	72.66	26	102	81	82
13	0.125	0.1	65.46	36	94	68	64	63.89	34	88	60	73
14	0.15	0.15	64.74	41	83	58	77	61.98	32	89	57	70
15	0.6	0.6	66.4	35	101	50	80	72.49	32	112	68	79
16	0.6	0.6	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
17	0.2	0.25	66.4	35	101	50	80	72.49	32	112	68	79
18	0.05	0.05	51.71	31	60	50	66	42.55	31	54	39	47
19	0.05	0.01	60.73	33	66	71	85	43.18	36	53	41	43
Mean	0.24 ± 0.22	0.23 ± 0.21	61.03 ± 9.19	36.56 ± 7.86	76.31 ± 19.65	56.25 ± 17	75.75 ± 12.77	59.96 ± 10.64	32.73 ± 5.23	83.47 ± 20.88	53.27 ± 13.28	70.53 ± 16.58
Patients with LHON												
1	0.05	0.05	47.3	45	61	35	48	49	37	65	44	50
2	0.4	0.16	41.06	29	45	42	48	54.48	57	71	44	47
3	0.0025	0.0025	53.3	43	50	56	64	48.48	30	70	39	54
4	0.3	1	97.43	43	123	91	133	97.56	37	123	88	142
5	0.01	0.01	41.44	33	53	27	53	53.36	38	64	46	65
6	0.01	0.01	53.53	37	51	65	60	48.37	37	57	39	60
7	0.0025	0.03	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
8	0.01	0.01	47.59	35	50	56	50	45.16	38	49	49	45
9	0.2	0.125	51.8	31	69	49	58	56.38	33	85	52	56
10	0.005	0.05	67.65	56	82	55	78	63.08	44	82	61	65
11	0.05	0.01	52.14	40	61	44	64	28.41	23	53	23	14
12	0.04	0.02	61.47	31	86	50	78	58.47	34	72	52	76
13	0.02	0.5	50.77	33	61	41	68	65.2	33	69	65	95
14	0.1	0.05	49.28	40	59	45	52	47.79	28	66	43	54
15	0.01	0.01	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
16	1	0.5	73.23	32	106	67	88	71.53	25	106	73	83
17	0.05	0.08	79.27	38	107	99	73	84.95	41	106	96	97
Mean	0.13 ± 0.25	0.15 ± 0.27	57.81 ± 15.52	37.73 ± 7.10	70.93 ± 24.35	54.80 ± 19.47	67.67 ± 21.79	58.15 ± 16.90	35.67 ± 8.24	75.87 ± 21.10	54.27 ± 19.42	66.87 ± 29.45

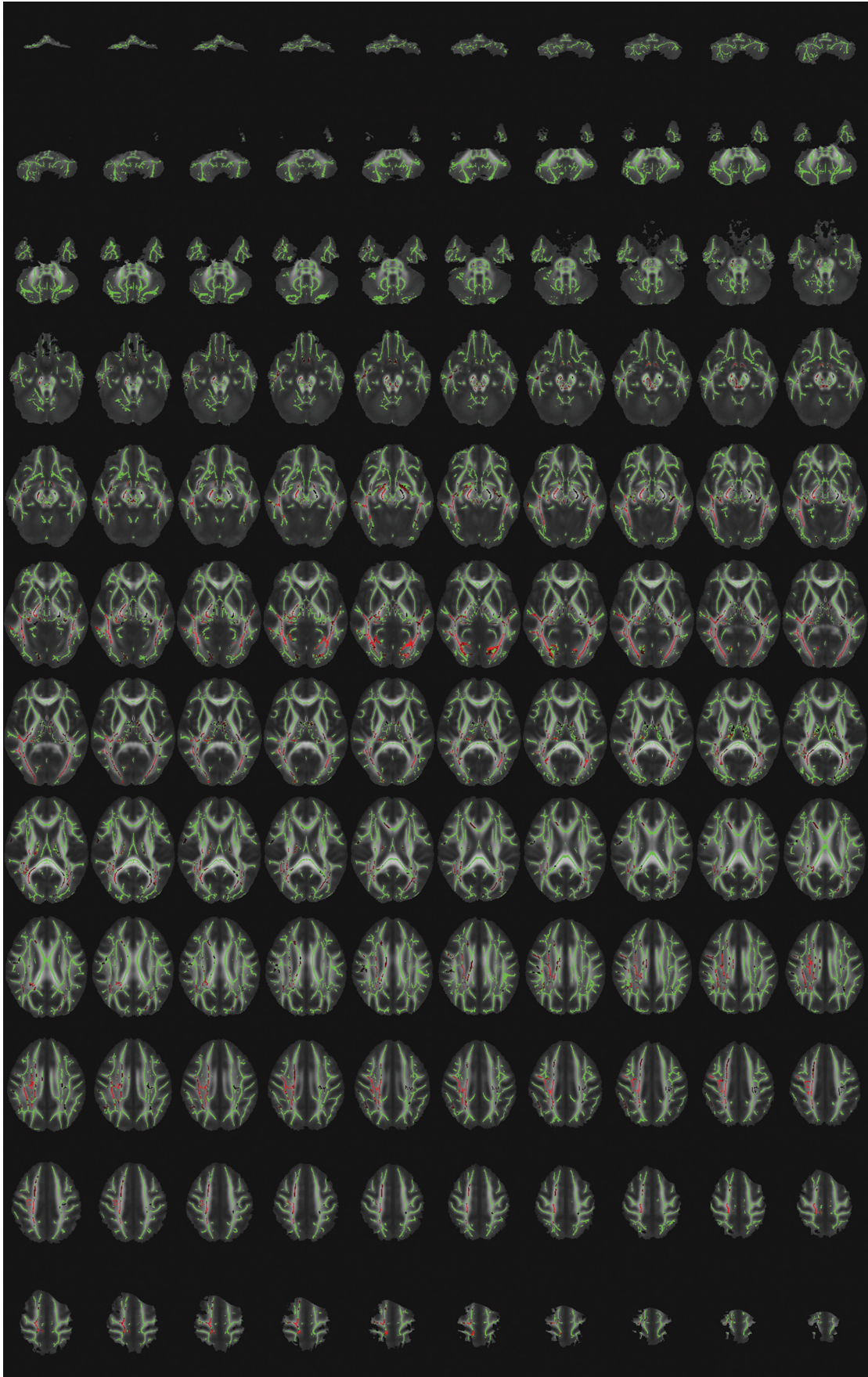
Note:—NA indicates not available.



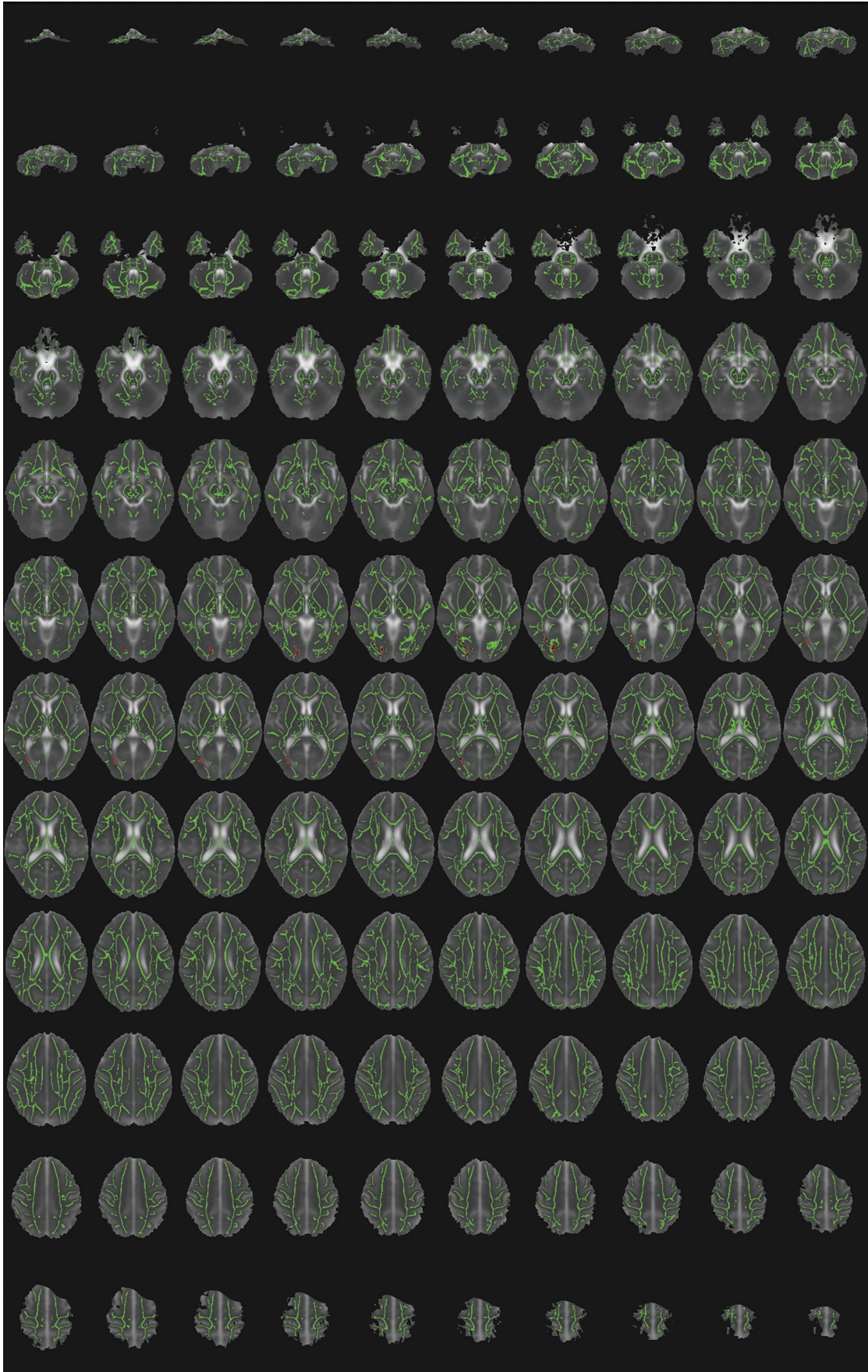
ON-LINE FIG 1. LHON FA < control FA: all axial sections.



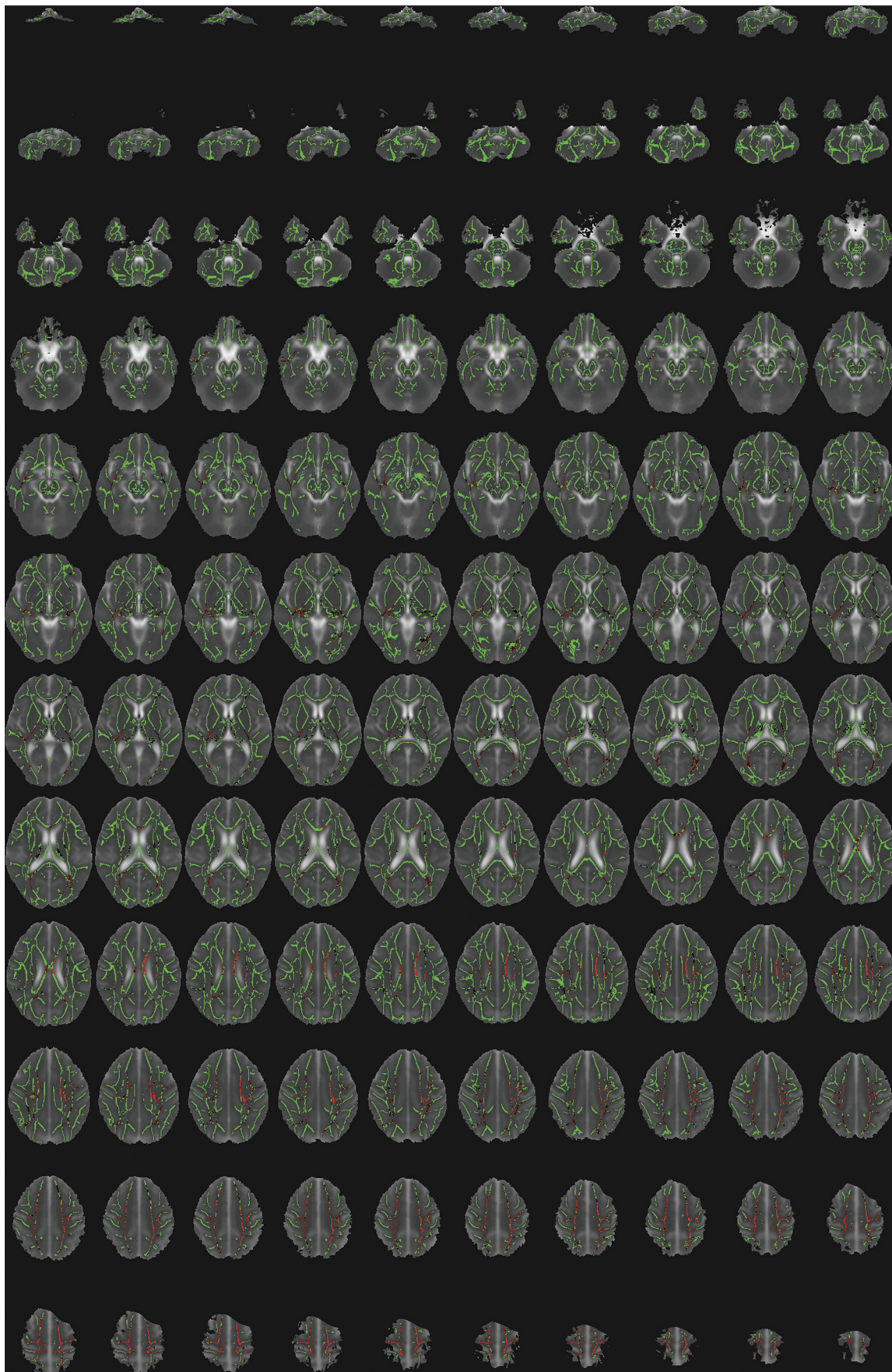
ON-LINE FIG 2. OPA1-DOA FA < control FA: all axial sections.



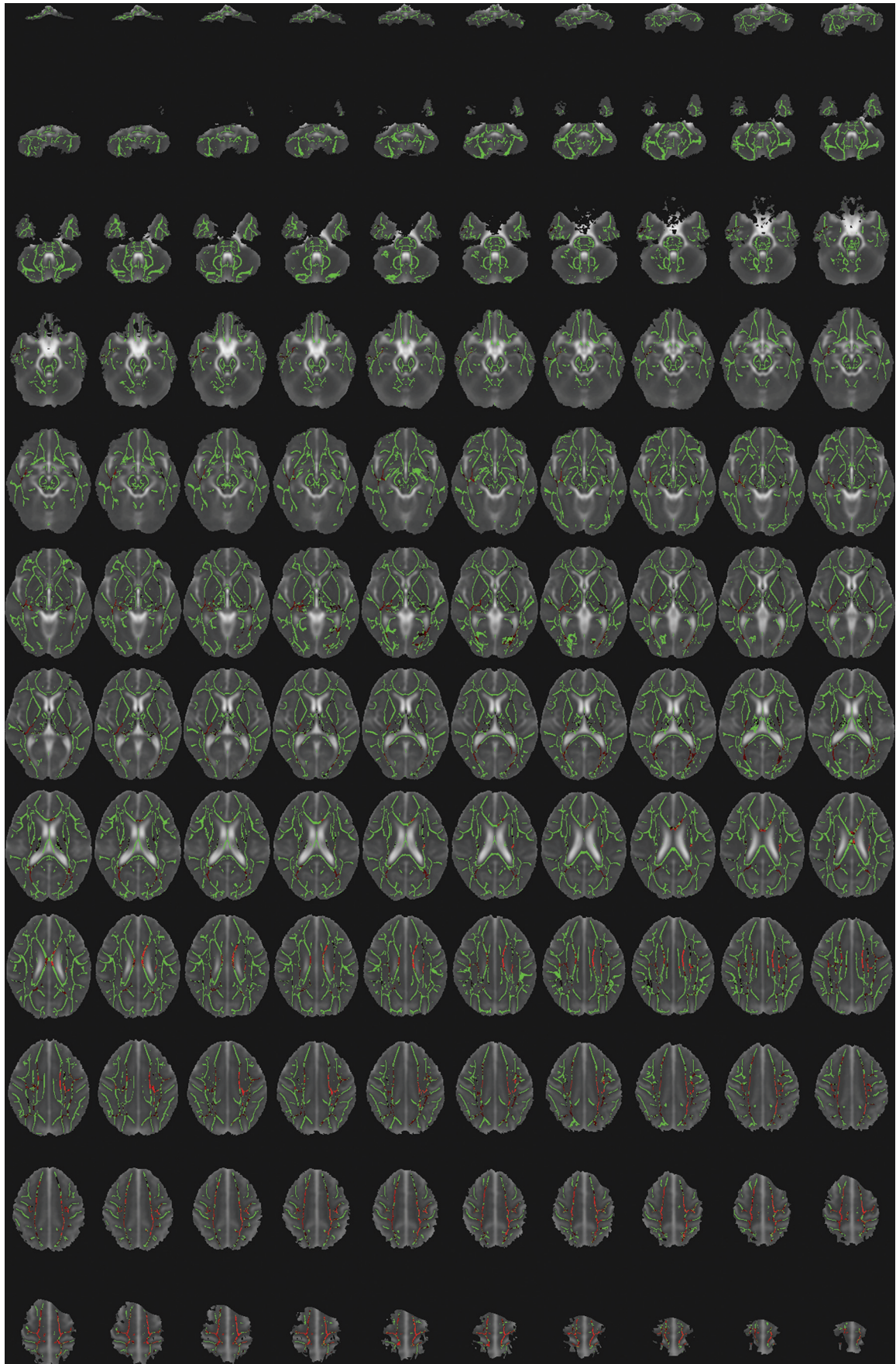
ON-LINE FIG 3. Pure OPA1-DOA FA < control FA: all axial sections.



ON-LINE FIG 4. LHON MD > control MD: all axial sections.



ON-LINE FIG 5. *OPA1*-DOA MD > control MD: all axial sections.



ON-LINE FIG 6. Pure OPA1-DOA MD > control MD: all axial sections.