Metabolic Brain Disease

Mitochondrial genome variation in male LHON patients with the m.11778G>A mutation

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Supplementary Table S1 mtDNA haplogroup details, total variant loads and variant loads using only non-synonymous variants with MutPred score above 0.5 threshold calculations for all participants in the study

LHON male subject ID	Age in years	mtDNA haplogroup	Total variant load	Number of variants	Variant load using variants with MutPred score >0.5	Number of variants with MutPred score >0.5
3595	42	H10b	1.712	4	0.638	1
4106	33	H5a1a	0.821	2	0.000	0
4806	35	X2b+226	2.349	6	0.569	1
5529	42	U4a2a2	1.466	5	0.000	0
6586	33	U4c1	1.850	6	0.000	0
9594	35	N1b1a2	3.005	7	1.210	2
9922	42	J2a1a1a	3.506	9	1.396	2
10911	33	U5b1e1	1.533	5	0.000	0
11266	35	K1c1h	2.257	6	0.609	1
11922	42	U5a1a1	2.625	7	0.561	1
13933	33	H7a1	0.821	2	0.000	0
14164	35	U5a1a1	2.625	7	0.561	1
15046	42	K1c1	2.257	6	0.609	1
15415	33	H1b1a	1.361	3	0.540	1
15591	35	J1c2e1	2.883	8	1.220	2
15745	42	W5a2	2.719	7	0.505	1
16159	33	W6a	3.915	9	1.701	3
16799	35	H1	0.821	2	0.000	0
17367	42	J1c2e1	2.883	8	1.220	2
17545	33	I2e	2.261	6	0.606	1
17791	35	H1a	0.821	2	0.000	0
194-L	42	J1c2c2a	3.506	10	1.220	2
456-L	33	H1e	1.326	3	0.505	1
L12-2007	37	K2a5	2.548	6	0.609	1
L12-2008	23	T1a1b	2.793	7	1.239	2
L12-2012	n/a	H5a1	1.357	3	0.536	1
L13-2006	28	T1a1b	2.793	7	1.239	2
L16-2008	14	V13	0.821	2	0.000	0
L19-2018	20	H4b1	1.485	3	0.664	1
L21-2015	20	J1c2e1	2.883	8	1.220	2
L21-2017	34	N1a1a1a1	2.197	6	0.606	1
L2-2018	28	U4c1	1.850	6	0.000	0
L23-2008	31	I2e	2.261	6	0.606	1
L24-2006	n/a	T1a1b	2.793	7	1.239	2
L24-2018	47	H1	0.821	2	0.000	0
L26-2002	19	H2a1c	0.821	2	0.000	0
L28-2016	16	U5b1b1a	1.235	4	0.000	0
L29-2005	n/a	H3g4	0.821	2	0.000	0
L32-2006	30	HV6a	1.004	3	0.000	0
L32-2016	15	K1c1h	2.257	6	0.609	1
L39-2008	40	U8b1a2b	2.557	6	0.688	1
L41-2005	38	U5a1d1	3.060	9	0.000	0
L4-2015	23	K1a1b1a	2.257	6	0.609	1

L5-2014	56	H3v	0.452	1	0.000	0
L7-2018	42	B4c1b2	2.541	6	1.142	2
L8-2019	33	H1c	0.821	2	0.000	0
L9-2010	35	H7c1	0.821	2	0.000	0
Control male subject ID	**Age in years**	**mtDNA haplogroup**	**Total variant load**	**Number of variants**	**Variant load using variants with MutPred score >0.5**	**Number of variants with MutPred score >0.5**
KJ2	84	H92	0.821	2	0.000	0
KJ6	71	H1c+152	0.821	2	0.000	0
KJ7	70	H1c+152	0.821	2	0.000	0
KJ8	64	H18	1.230	3	0.000	0
KJ12	60	U5a1d2a1	2.665	7	0.601	1
KJ13	52	H	0.821	2	0.000	0
KJ14	72	H3+152	0.821	2	0.000	0
KJ15	71	J1c2c1	3.253	9	1.220	2
KJ21	76	T2a1	2.588	7	1.239	2
KJ22	82	H2a1c	0.821	2	0.000	0
KJ23	69	U5a2a1	2.019	6	0.000	0
KJ26	63	H+16311	0.821	2	0.000	0
KJ31	77	X2b4	2.349	6	0.569	1
KJ38	64	W1	2.719	7	0.505	1
KJ40	81	J1c9	2.883	8	1.220	2
KJ45	83	H16d	0.821	2	0.000	0
KJ47	85	H5r	0.821	2	0.000	0
KJ49	77	J1c5	2.883	8	1.220	2
KJ52	60	H3	0.821	2	0.000	0
KJ55	77	H11a1	1.072	3	0.000	0
KJ56	61	V1a1	2.629	6	0.594	1
KJ61	78	V7a	0.821	2	0.000	0
KJ63	87	X2e2b	1.716	5	0.000	0
KJ66	83	J1c2	3.509	9	1.846	3
KJ67	62	H5e1a1	0.821	2	0.000	0
KJ68	68	V1a	1.885	4	0.594	1
KJ70	84	HV9	0.821	2	0.000	0
KJ75	69	U5a2a1	2.019	6	0.000	0
KJ76	72	I1a1a	2.714	7	1.188	2
KJ79	n/a	U4a2b	1.215	4	0.000	0
KJ104	60	H16	0.821	2	0.000	0
KJ106	56	N1b1a1	3.070	7	1.275	2
KJ112	65	J1c2	3.509	9	1.846	3
KJ116	83	H1c1	0.821	2	0.000	0
KJ117	76	H11b1	1.581	4	0.509	1
KJ118	69	V7a	0.821	2	0.000	0
KJ120	79	H7c3	0.821	2	0.000	0
KJ121	61	J1c5	2.883	8	1.220	2
KJ123	57	H28a	1.320	3	0.000	0
KJ129	65	V1a1	2.629	6	0.594	1
KJ131	66	J2a1a1a2	3.059	8	1.396	2
KJ153	n/a	U8a1a1b	2.630	6	1.331	2