Supplementary Table 1 Patients with m.10197G>A mutation reported in the present study and previous literature.

No. of article	References	Country / District	No. of carrier	Gender / presenting age (year)	Onset age (year)	Family History	Mutation Load % (sample)	Phenotype Spectrum	Treatment	Outcome
1	Present study	China	P1	M / Early 20s	Young adult	Yes	58.1% (Leu) 77.1% (Mus)	Adult-onset LS / LDYT overlap syndrome	Idebenone, ubidecarenone	Stable at 1-year follow-up
2	Gilhooley, el al., 2024[1]	England	P2	NA / Child, NA	Child, NA	NA	Homoplasmic (NA)	LHON	NA	NA
3	Baldo, et al., 2023[2]	Portugal	P3	M / 7	Child, NA	NA	Homoplasmic (Mus)	LS and epilepsy	NA	NA
4	Nogueira, et al., 2023[3]	Portugal	P4	M / 4	Child, NA	NA	Homoplasmic (Mus)	LS	NA	NA
5	Durrleman, et al., 2023[4]	France	P5	NA / Child, NA	Child, NA	NA	NA	MELAS	NA	NA
6	Atdissone, et al., 2023[5]	Italy	P6	NA / Child, NA	Child, NA	NA	99.6% (Leu), 98% (Mus)	LS	Mitochondrial cofactor “cocktail”	NA
			P7	NA / Child, NA		NA		LS		NA
			P8	NA / Child, NA		NA		LS		NA
			P9	NA / Child, NA		NA	99-100% (Leu), 95-100% (Mus)	LS		NA
			P10	NA / Child, NA		NA		LS		NA
7	Stenton, et al., 2022[6]	China	P11	NA / NA	0	NA	Homoplasmic (Leu)	LS	NA	NA
			P12	NA / NA	5.75	NA	83% (Leu)	Late-onset LS	NA	NA
			P13	NA / NA	0.42	NA	Homoplasmic (Leu)	LS	NA	NA
			P14	NA / NA	4.93	NA	97% (Leu)	Late-onset LS	NA	NA
			P15	NA / NA	9.5	NA	98% (Leu)	Late-onset LS	NA	NA
			P16	NA / NA	2.25	NA	89% (Leu)	Late-onset LS	NA	NA
			P17	NA / NA	0.25	NA	99% (Leu)	LS	NA	NA
8	Wei, et al., 2022[7]	China		*Family*		Yes				
			P18	M / 19	14		68% (Leu), 93% (Uri)	Late-onset LS	NA	NA

			P19	F / 49	22		18.6% (Leu), 61% (Uri)	Adult-onset LS	NA	NA
9	Wu, et al., 2022[8]	China	P20	NA / Child, NA	Child, NA	None	99.6% (Leu)	LS	NA	NA
10	Zhao, et al., 2022[9]	China	P21	NA / Adult, NA	Adult, NA	NA	NA	LHON	NA	NA
11	Hechmi, et al., 2022[10]	Tunisia	P22	M / 5	1	Yes	87.7% (Leu)	LS	NA	NA
12	Cipriano, et al., 2021[11]	Italy	P23	M / 36	25	None	NA	Adult-onset LS	acetylcarnitine, ubidecarenone, tracheostomy with not-invasive ventilation	alive at 7 months after diagnosis
13	Wei, et al., 2021[12]	China	P24	M / 20	14	None	61.5% (Leu)	MELAS/LS overlap syndrome	NA	NA
14	Gramegna, et al., 2021[13]	Italy	P25	F / 36	12	NA	85.0% (Leu), 98.0% (Uri)	MELAS	NA	NA
15	Cui, et al., 2020[14]	China	P26	M / 19	19	None	Heteroplasmy (Leu)	LHON	NA	No obvious visual improvement at the 1-year
16	Chan, et al., 2020[15]	USA	P27	F / 20	20	NA	51% (NA)	LHON	Idebenone, vitamin C	Mild visual improvement
17	Tolomeo, et al., 2019[16]	Italy	P28	M / 7	6	None	Homoplasmic (Leu, Uri, Mus, Fib)	Dystonia and LS	NA	NA
			P29	M / 5	0.75	None	99.9% (Leu), 99.1% (Mus), 99.4% (Fib)	Dystonia and LS	NA	NA
			P30	M / 4	1.33	None	96.4% (Mus), 95.2% (Fib)	LS and arterial malformations	NA	NA

18	Solyman, et al., 2019[17]	USA	P31	F / 48	48	Yes	10-30% (NA)	LHON	Idebenone	Visual acuity improved at 4-month follow-up
19	Fantini, et al., 2019[18]	USA	*Family* P32	F / 48	48	Yes	NA	LHON	Idebenone, vitamin C, hormone replacement therapy	Complete reversal of vision loss by eight months post-therapy
			P33	F / NA	58		NA	LHON	NA	Vision loss
20	Pereira, et al., 2019[19]	Brazil	P34	M / 1.41	0.91	None	Homoplasmic (Mus)	LS	NA	Died 20 days later 0(3 years and 2 months of age)
21	Wei, et al., 2018[20]	China	P35	F / NA	2	None	92% (Leu)	Late-onset LS	Coenzyme Q10, vitamins C, E, B1, and B2.1	NA
			P36	M / NA	6	None	83% (Leu)	Late-onset LS	Coenzyme Q10, vitamins C, E, B1, and B2.	NA
			P37	M / NA	14	None	68% (Leu)	Late-onset LS	Coenzyme Q10, vitamins C, E, B1, and B2.	NA
			P38	M / NA	22	None	45% (Leu)	Adult-onset LS	Coenzyme Q10, vitamins C, E, B1, and B2.	NA
22	Huang, et al., 2017[21]	Taiwan	P39	M / 23	23	None	64.4% (Leu)	LHON	Idebenone, vitamin C	Visual recovery at 1-year follow-up
23	Zhang, et al., 2018[22]	China	P40	NA / NA	NA	NA	NA	MELAS	NA	NA
24	Lee, et al., 2016[23]	Korea	P41	F / 6	1.33	NA	NA	LS	NA	Stable
			P42	M / 16	2.25	NA	NA	Late-onset LS	NA	Deterioration
			P43	F / 13	3	NA	NA	Late-onset LS	NA	Stable

25	Leng, et al., 2015[24]	China	P44	M / 14	14	None	61.1% (Leu), 75.8% (Uri), 85.1% (Mus)	MELAS/LS overlap syndrome	Vitamin B and several coenzymes	Stable
26	Chen, et al., 2015[25]	China		*Family*		Yes				
			P45	F / 16	15		95.0% (Leu), 97.0% (Mus)	Late-onset LS	Coenzyme Q10	Improved at 3-month follow-up
			HC1	F / NA (mother)			95% (Leu)	Asymptomatic carrier	/	/
			HC2	M / NA (brother)			39% (Leu)	Asymptomatic carrier	/	/
27	Calvo, et al., 2010[26]	Australia	P46	NA / NA	3	None	90% (Leu)	Mitochondrial encephalopathy	NA	NA
28	Wang, et al., 2009[27]	China		*Family*		Yes				
			P47	M / NA	5		Homoplasmic (Leu)	LDYT	NA	NA
			P48	M / NA	8		Homoplasmic (Leu)	LDYT	NA	NA
			P49	F / NA	8		Homoplasmic (Leu)	LDYT	NA	NA
			P50	F / NA	5		Homoplasmic (Leu)	LDYT	NA	NA
			P51	F / 20	8		Homoplasmic (Leu)	LDYT	NA	NA
			P52	M / 18	5		Homoplasmic (Leu)	LDYT	NA	NA
			P53-62	5M / NA and 5F / NA	14-30		Homoplasmic (Leu)	LHON	NA	NA
			HC3-HC4	1M / NA and 1F / NA	/		Homoplasmic (Leu)	Asymptomatic carrier	/	/
29	Naess, et al., 2009[28]	Sweden	P63	M / NA	7	NA	50% (Mus), 31% (Fib)	Late-onset LS	NA	NA
30	Valente, et al., 2009[29]	Italy	P64	NA / NA	0.58	NA	NA	LS	NA	NA
31	Chae, et al., 2007[30]	Korean	P65	F / 9	7	Yes	98% (Mus)	Late-onset LS	NA	NA
			P66	M / NA	4	Yes	86% (Mus)	Late-onset LS	NA	NA
			P67	M / NA	0.25	None	80% (Mus)	LS	NA	NA
32	Sarzi, et al., 2007[31]	France		*Family 1*		Yes				
			P68	F / 41 (mother)	14		67% (Leu)	Dystonia	NA	Stable

			P69	M / NA	0.08		Homoplasmic (Mus)	Dystonia and LS	NA	Died at 5 mon
			P70	M / NA	0.16		Homoplasmic (Leu, liv)	Dystonia and LS	NA	Died at 2 mon
			P71	F / NA	0.16		Homoplasmic (Mus)	Dystonia	NA	Died at 8 mon
				Family 2		Yes				
			HC5	F / 34 (mother)	/		50% (Leu)	Asymptomatic carrier	/	/
			P72	M / NA	0.33		/	Dystonia and LS, epilepsy	NA	Died at 11-mon
			P73	M / 5	0.41		Homoplasmic (Mus, Fib)	Dystonia and LS, epilepsy	NA	Developmental delay and deterioration
				Family 3		Yes				
			P74	F / 37 (mother)	6		74% (Leu), 96% (Mus)	Dystonia and late-onset LS	NA	Stable
			HC6	F / 7	/		73% (Leu)	Asymptomatic carrier	/	/
			P75	F / 5	0.41		Homoplasmic (Leu)	Seizure and development delay	NA	Seizure and developmental delay
			P76	M / NA	2		/	Encephalopathy and seizure	NA	Died at 2-year
			P77	M / NA	0.41		Homoplasmic (Leu, Mus, Cybrids)	Dystonia and LS, seizure	NA	Died at 1-year
33	Tchikviladzé, et al. 2007[32]	France	P78	M / 30	6	NA	<50% (Leu), 100% (Mus), 70% (buccal mucosa cells)	Late-onset LS	NA	Deterioration
34	Kirby, et al., 2004[33]	Australia	P79	F / NA	2	Yes	Homoplasmic (Mus)	LS-like	NA	Alive at 13 years

F, Female. LDTY, Leber hereditary optic neuropathy and dystonia. HC, healthy carrier. Leu, leukocytes. LHON, Leber hereditary optic neuropathy. Liv, liver. LS, Leigh syndrome. M, male. MELAS, mitochondrial encephalopathy

with lactate acidosis and stroke-like episodes. Mon, months. Mus, muscle. NA, not available. Uri, urine sediment. P, patient.

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