Supplementary table S1. Demographics of patients diagnosed with mtDNA disease at Turku University Hospital in 2010–2022. Patients ≥18 years of age in 2022 are shown. Patients from outside of Southwest Finland are shaded.

Patient	Year	mtDNA mutation	Tissue tested	Syndrome/disorder	Sex	Approximate age at diagnosis (y)	Family history	Specialty service diagnosing
1†	2010	m.11778G>A	B	LHON	M	60-70	Y	Opht
2	2011	m.3243A>G	Mu	MIDD	M	10-20	Y	Gen
3†	2011	m.3243A>G	Mu	MELAS	F	50-60	Y	Gen
4	2011	m.4296G>A	Mu	Parkinsonism, endocrinopathy, epilepsy	M	10-20	N	Neuro
5	2012	m.3260A>G	Mu	DM, epilepsy, MM	F	50-60	N	Neuro
6	2012	m.3260A>G	Mu	PEO	F	70-80	Y	Gen
7	2013	m.3243A>G	Mu	MM	F	60-70	Y	Neuro
8	2013	m.3243A>G	Mu	MELAS	M	40-50	N	Research
9†	2013	m.3243A>G	Mu	MELAS	F	60-70	N	Research
10	2013	m.7510T>C	Mu	SNHL	F	10-20	Y	Neuro
11	2013	m.7510T>C	Mu	SNHL	M	20-30	Y	Neuro
12	2013	m.7510T>C	Mu	SNHL	F	40-50	Y	Neuro
13	2016	m.3243A>G	Mu	SNHL	F	30-40	Y	Gen
14	2016	m.3243A>G	Mu	MM	F	50-60	Y	Opht
15	2016	m.8344A>G	Mu	SNHL	F	50-60	Y	Gen
16	2016	m.11778G>A	B	LHON	M	40-50	N	Opht

17	2016	m.11778G>A	B	LHON	M	50-60	N	Opht
18	2017	mtDNA deletion	Mu	PEO	M	60-70	N	Neuro
19	2017	mtDNA deletion	Mu	MM	F	60-70	N	Neuro
20	2018	m.3243A>G	B	MIDD	F	20-30	Y	Neuro
21†	2018	m.3271T>C	Mu	MELAS	F	60-70	N	Neuro
22	2018	m.4640C>A	B	LHON	M	30-40	N	Opht
23	2018	m.11778G>A	B	LHON	M	40-50	N	Opht
24	2019	m.3243A>G	B	MIDD	M	40-50	Y	Endo
25	2019	m.3243A>G	Mu	MM	F	50-60	Y	Opht
26	2019	m.3243A>G	B	MIDD	M	40-50	Y	Endo
27	2019	m.3243A>G	B	SNHL	F	70-80	Y	Gen
28†	2019	m.13513G>A	Mu	MELAS	M	60-70	N	Neuro
29	2020	m.3243A>G	B	MIDD	F	50-60	Y	Endo
30	2020	m.3243A>G	B	MM	F	50-60	Y	Gen
31	2020	m.3243A>G	Mu	PEO	F	60-70	N	Opht
32	2020	mtDNA deletion	Mu	PEO	M	30-40	N	Neuro
33	2021	m.3243A>G	Mu	MELAS	F	50-60	N	Card
34	2021	m.3243A>G	B	MIDD	M	20-30	Y	Endo
35	2021	m.3243A>G	B	MIDD	M	20-30	Y	Endo
36	2021	m.3243A>G	B	MM	M	20-30	Y	Gen

37	2021	m.3243A>G	B	SNHL	F	50-60	Y	Gen
38	2021	m.3243A>G	B	MELAS	F	50-60	N	Neuro
39	2021	m.8344A>G	Mu	MERRF	F	50-60	N	Neuro
40	2021	m.11778G>A	B	LHON	F	30-40	Y	Opht
41	2021	m.11778G>A	B	LHON	M	80-90	Y	Opht
42	2022	m.3243A>G	B	MIDD	M	20-30	Y	Gen

B = blood. Card = cardiology. DM = diabetes mellitus. Gen = clinical genetics. Endo = endocrinology. F = female. Family history = family history suggestive of mitochondrial disease. LHON = Leber's hereditary optic neuropathy. LS = Leigh syndrome. M = male. MELAS = mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes. MERRF = myoclonic epilepsy with ragged red fibres. MIDD = maternally inherited diabetes and deafness. MM = mitochondrial myopathy. Mu = muscle. N = no. Neuro = neurology. Opht = ophthalmology. PEO = progressive external ophthalmoplegia. Research = patient diagnosed in a research project. Y = yes. † = deceased. SNHL = sensorineural hearing loss.