Supplementary material contains:

Supplementary table 1. OMIM codes for mitochondrial diseases (MitD) and phenotype descriptions.

OMIM Entry	OMIM code	Phenotype description
1	# 500000	CARDIOMYOPATHY, INFANTILE HISTIOCYTOID
2	# 500001	LEBER OPTIC ATROPHY AND DYSTONIA
3	# 500002	MITOCHONDRIAL MYOPATHY WITH DIABETES
4	# 500003	STRIATONIGRAL DEGENERATION, INFANTILE, MITOCHONDRIAL
5	# 500004	RETINITIS PIGMENTOSA-DEAFNESS SYNDROME
6	# 500005	HYPOMAGNESEMIA, HYPERTENSION, AND HYPERCHOLESTEROLEMIA, MITOCHONDRIAL
7	# 500006	CARDIOMYOPATHY, INFANTILE HYPERTROPHIC
8	# 500007	CYCLIC VOMITING SYNDROME (CYCLIC VOMITING SYNDROME WITH NEUROMUSCULAR DISEASE, INCLUDED)
9	# 500008	DEAFNESS, NONSYNDROMIC SENSORINEURAL, MITOCHONDRIAL
10	# 500009	MITOCHONDRIAL MYOPATHY, INFANTILE, TRANSIENT
11	# 500010	ATAXIA AND POLYNEUROPATHY, ADULT-ONSET
12	# 500011	MYOPATHY, LACTIC ACIDOSIS, AND SIDEROBLASTIC ANEMIA 3
13	# 500013	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, MITOCHONDRIAL FORM, 1
14	# 500014	MITOCHONDRIAL COMPLEX I DEFICIENCY, MITOCHONDRIAL TYPE 1
15	# 500015	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, MITOCHONDRIAL TYPE 1; MC5DM1
16	# 502000	AGING
17	# 502500	ALZHEIMER DISEASE, SUSCEPTIBILITY TO, MITOCHONDRIAL
18	# 515000	CHLORAMPHENICOL TOXICITY (CHLORAMPHENICOL RESISTANCE, INCLUDED)
19	# 520000	DIABETES AND DEAFNESS, MATERNALLY INHERITED
20	# 520100	DIARRHEA, CHRONIC, WITH VILLOUS ATROPHY
21	# 530000	KEARNS-SAYRE SYNDROME
22	# 535000	LEBER OPTIC ATROPHY
23	# 540000	MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES
24	# 545000	MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS
25	# 550500	MYOGLOBINURIA, RECURRENT
26	# 551000	MITOCHONDRIAL MYOPATHY, LETHAL, INFANTILE
27	# 551200	NEPHROPATHY, CHRONIC TUBULOINTERSTITIAL
28	# 551500	NEUROPATHY, ATAXIA, AND RETINITIS PIGMENTOSA
29	# 553000	ONCOCYTOMA
30	# 556500	PARKINSON DISEASE, MITOCHONDRIAL
31	# 557000	PEARSON MARROW-PANCREAS SYNDROME
32	# 560000	RENAL TUBULOPATHY, DIABETES MELLITUS, AND CEREBELLAR ATAXIA
33	# 598500	WOLFRAM SYNDROME, MITOCHONDRIAL FORM

Supplementary table 2. OMIM codes for mtDNA genes and associated phenotypes.

OMIM Entry	OMIM code	Gene description
34	* 516000	COMPLEX I, SUBUNIT ND1; *MTND1*
35	* 516001	COMPLEX I, SUBUNIT ND2; *MTND2*
36	* 516002	COMPLEX I, SUBUNIT ND3; *MTND3*
37	* 516003	COMPLEX I, SUBUNIT ND4; *MTND4*
38	* 516004	COMPLEX I, SUBUNIT ND4L; *MTND4L*
39	* 516005	COMPLEX I, SUBUNIT ND5; *MTND5*
40	* 516006	COMPLEX I, SUBUNIT ND6; *MTND6*
41	* 516020	CYTOCHROME b OF COMPLEX III; *MTCYB*
42	* 516030	COMPLEX IV, CYTOCHROME c OXIDASE SUBUNIT I; *MTCO1*
43	* 516040	COMPLEX IV, CYTOCHROME c OXIDASE SUBUNIT II; *MTCO2*
44	* 516050	CYTOCHROME c OXIDASE III; *MTCO3*
45	* 516060	ATP SYNTHASE 6; *MTATP6*
46	* 516070	ATP SYNTHASE 8; *MTATP8*
47	* 561000	RIBOSOMAL RNA, MITOCHONDRIAL, 12S; *MTRNR1*
48	* 561010	RIBOSOMAL RNA, MITOCHONDRIAL, 16S; *MTRNR2*
49	* 590000	TRANSFER RNA, MITOCHONDRIAL, ALANINE; *MTTA*
50	* 590005	TRANSFER RNA, MITOCHONDRIAL, ARGININE; *MTTR*
51	* 590010	TRANSFER RNA, MITOCHONDRIAL, ASPARAGINE; *MTTN*
52	* 590015	TRANSFER RNA, MITOCHONDRIAL, ASPARTIC ACID; *MTTD*
53	* 590020	TRANSFER RNA, MITOCHONDRIAL, CYSTEINE; *MTTC*
54	* 590025	TRANSFER RNA, MITOCHONDRIAL, GLUTAMIC ACID; *MTTE*
55	* 590030	TRANSFER RNA, MITOCHONDRIAL, GLUTAMINE; *MTTQ*
56	* 590035	TRANSFER RNA, MITOCHONDRIAL, GLYCINE; *MTTG*
57	* 590040	TRANSFER RNA, MITOCHONDRIAL, HISTIDINE; *MTTH*
58	* 590045	TRANSFER RNA, MITOCHONDRIAL, ISOLEUCINE; *MTTI*
59	* 590050	TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1; *MTTL1*
60	* 590055	TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 2; *MTTL2*
61	* 590060	TRANSFER RNA, MITOCHONDRIAL, LYSINE; *MTTK*
62	* 590065	TRANSFER RNA, MITOCHONDRIAL, METHIONINE; *MTTM*
63	* 590070	TRANSFER RNA, MITOCHONDRIAL, PHENYLALANINE; *MTTF*
64	* 590075	TRANSFER RNA, MITOCHONDRIAL, PROLINE; *MTTP*
65	* 590080	TRANSFER RNA, MITOCHONDRIAL, SERINE, 1; *MTTS1*
66	* 590085	TRANSFER RNA, MITOCHONDRIAL, SERINE, 2; *MTTS2*
67	* 590090	TRANSFER RNA, MITOCHONDRIAL, THREONINE; *MTTT*
68	* 590095	TRANSFER RNA, MITOCHONDRIAL, TRYPTOPHAN; *MTTW*
69	* 590100	TRANSFER RNA, MITOCHONDRIAL, TYROSINE; *MTTY*
70	* 590105	TRANSFER RNA, MITOCHONDRIAL, VALINE; *MTTV*

Supplementary table 3. PRISMA checklist of items to include when reporting a systematic review or meta-analysis.

Section/topic	#	Checklist item	Reported on page #
TITLE			
Title	1	Identify the report as a systematic review, meta-analysis, or both.	1
ABSTRACT			
Structured summary	2	Provide a structured summary including (as applicable): background; objectives; data sources; study eligibility criteria, participants, and interventions; study appraisal and synthesis methods; results; limitations; conclusions and implications of key findings; systematic review registration number.	2–5
INTRODUCTION			
Rationale	3	Describe the rationale for the review in the context of what is already known.	9
Objectives	4	Provide an explicit statement of questions being addressed with reference to participants, interventions, comparisons, outcomes, and study design (PICOS).	-
METHODS			
Protocol and registration	5	Indicate if a review protocol exists, if and where it can be accessed (e.g., Web address), and, if available, provide registration information including registration number.	-
Eligibility criteria	6	Specify study characteristics (e.g., PICOS, length of follow-up) and report characteristics (e.g., years considered, language, publication status) used as criteria for eligibility, giving rationale.	9
Information sources	7	Describe all information sources (e.g., databases with dates of coverage, contact with study authors to identify additional studies) in the search and date last searched.	9
Search	8	Present the full electronic search strategy for at least one database, including any limits used, such that it could be repeated.	9
Study selection	9	State the process for selecting studies (i.e., screening, eligibility, included in systematic review, and, if applicable, included in the meta-analysis).	9–10
Data collection process	10	Describe method of data extraction from reports (e.g., piloted forms, independently, in duplicate) and any processes for obtaining and confirming data from investigators.	9
Data items	11	List and define all variables for which data were sought (e.g., PICOS, funding sources) and any assumptions and simplifications made.	-
Risk of bias in individual studies	12	Describe methods used for assessing risk of bias of individual studies (including specification of whether this was done at the study or outcome level) and how this information is to be used in any data synthesis.	-
Summary measures	13	State the principal summary measures (e.g., risk ratio, difference in means).	-
Synthesis of results	14	Describe the methods of handling data and combining results of studies, if done, including measures of consistency (e.g., I^2) for each meta-analysis.	-

Section/topic	#	Checklist item	Reported on page #
Risk of bias across studies	15	Specify any assessment of risk of bias that may affect the cumulative evidence (e.g., publication bias, selective reporting within studies).	-
Additional analyses	16	Describe methods of additional analyses (e.g., sensitivity or subgroup analyses, meta-regression), if done, indicating which were prespecified.	-
RESULTS			
Study selection	17	Give numbers of studies screened, assessed for eligibility, and included in the review, with reasons for exclusions at each stage, ideally with a flow diagram.	10
Study characteristics	18	For each study, present characteristics for which data were extracted (e.g., study size, PICOS, follow-up period) and provide the citations.	Tables 1–3
Risk of bias within studies	19	Present data on risk of bias of each study and, if available, any outcome level assessment (see item 12).	-
Results of individual studies	20	For all outcomes considered (benefits or harms), present, for each study: (a) simple summary data for each intervention group (b) effect estimates and confidence intervals, ideally with a forest plot.	-
Synthesis of results	21	Present results of each meta-analysis done, including confidence intervals and measures of consistency.	-
Risk of bias across studies	22	Present results of any assessment of risk of bias across studies (see Item 15).	-
Additional analysis	23	Give results of additional analyses, if done (e.g., sensitivity or subgroup analyses, meta-regression [see Item 16]).	-
DISCUSSION			
Summary of evidence	24	Summarize the main findings including the strength of evidence for each main outcome; consider their relevance to key groups (e.g., healthcare providers, users, and policy makers).	
Limitations	25	Discuss limitations at study and outcome level (e.g., risk of bias), and at review-level (e.g., incomplete retrieval of identified research, reporting bias).	18
Conclusions	26	Provide a general interpretation of the results in the context of other evidence, and implications for future research.	18
FUNDING			
Funding	27	Describe sources of funding for the systematic review and other support (e.g., supply of data) and the role of funders in the systematic review.	3

From: Moher D, Liberati A, Tetzlaff J, Altman DG, The PRISMA Group (2009). Preferred Reporting Items for Systematic Reviews and Meta-Analyses: The PRISMA Statement. PLoS Med 6(7): e1000097. doi:10.1371/journal.pmed1000097

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