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Supplementary Table S1

Supplementary Table S1 presents a summary of known mtDNA mutations related to the main forms of diabetes (T1D, T2D, MIDD and GDM). It combines data from case studies (one patient/family) and wide population analyses. All mentioned mutations were identified retrospectively, from patients with diagnosed diabetes (internationally recognized and clinically approved criteria, but still could vary depending on the local clinical practice).

Table S1. Diabetes mellitus-related mtDNA mutations.

Mutation	Gene	DM type	Patient population	Reference
m.T58C		T2D	Italian T2D patients	Cormio et al. 2009
m.C151T	OriH			
m.568 poly C length variation (up to 12 bp)	D-loop	MIDD	Turkish family case study	Janssen et al. 2006
m.8281 length variation, 2 9-bp repeats	$tRNA^{Lys}$			
m.C1310T	12S rRNA	T2D	Japanese T2D patients	Tawata et al. 1998
m.G1438A				
m.A12026G	ND4			
m.A1382C	MOTS-c/12S rRNA	T2D	Asian T2D patients	Zempo et al. 2021
m.A3156G*	16S rRNA	T1D, T2D	Japanese T1D, T2D and GDM patients	Ohkubo et al. 2001
m.G3357A*	ND1			
m.C3375A*				
m.T3394C*				
m.T3200C	16S rRNA	T2D	Chinese T2D patients	Yang et al. 2002
m.A3243G	$tRNA^{Leu}$	MIDD	Caucasian family case study	Van den Ouweland et al. 1992
m.C3256T	$tRNA^{Leu}$	MIDD	Japanese family case study	Hirai et al. 1998
m.T3264C	$tRNA^{Leu}$	MIDD	Japanese family case study	Suzuki et al. 1997
m.T3271C*	$tRNA^{Leu}$	MIDD	Taiwanese family case study	Chen et al. 2004
m.A3302G	$tRNA^{Leu}$	T2D	Han Chinese family case study	Ding et al. 2016
m.G3316A	ND1	T2D	Japanese patients	Odawara et al. 1996
m.T3394C	ND1	T2D	Chinese patients	Tang et al. 2006
m.A14693G	$tRNA^{Glu}$	T2D	Indonesian patients	Pranoto et al. 2005
m.C3254A	$tRNA^{Leu}$	GDM	Healthy subjects with no diabetes history and symptoms, Singapore	Chen et al. 2000
m.T3398C	ND1			
m.A3399T				
m.T3394C	ND1	T2D		Liao et al. 2008

m.G4491A	ND2		Han Chinese patients	
m.C5178A				
m.A10398G	ND3			
m.T16189C	mtDNA control region			
m.T16519C				
m.T3548C	ND1	MIDD	Brazilian patients	Crispim et al. 2008
m.A8348G	$tRNA^{Lys}$			
m.C8393T	ATP8			
m.C8478T				
m.T8551C	ATP6, ATP8			
m.A12026G	ND4			
m.G4284A	$tRNA^{Ile}$	MIDD	Italian patient sith multisystem progressing disorders	Corona et al. 2002
m.A4738C**	ND2	T1D	ALR/Lt, a mouse straing with strong resistance to T1D	Mathews et al. 2005
m.5172 11 and 12 as polymorphic locus**	OriL	T2D	AKR/J (C57BL/6J-mtAKR/J; B6-mtAKR) on a C57BL/6J (B6) mouse strain	Hirose et al. 2018
m.C5178A	ND2	T1D, T2D	Japanese population	Wang et al. 2001, Uchigata et al. 2002
m.G7778T natural polymorphism**	ATP8	T1D, T2D	C57BL/6J-mtFVB/N (B6-mtFVB) mouse strain	Weiss et al. 2012, Yu et al. 2009
m.A8296G	$tRNA^{Lys}$	MIDD	Japanese patients	Kameoka et al. 1998
m.A8344G	$tRNA^{Lys}$	T2D	Japanese family case	Suzuki et al. 1994
m.A8344G*	$tRNA^{Lys}$	GDM	Asian Indian population with no diabetes history	Khan et al. 2015
m.T8356C	$tRNA^{Lys}$	T2D	Coimbatore T2D patients	Vijaya Padma et al. 2010
m.T8414G	ATP8	T2D	Chinese Uyghur T2D patients	Jing et al. 2017
m.G2706A	16S RNA	T2D		
m.C8561G	ATP6, ATP8	T2D	Case study of one family	Kytövuori et al. 2016
m.A8860G*	ATP6	MIDD	Chinese family case study with MIDD and MELAS	Li et al. 2015a
m.A15326G*	CYB			
m.G9267C*	COX3	MIDD	Tunesian family case with MIDD	Tabebi et al. 2015
m.G5913A*	COX1			
m.A9827G**	COX3	T1D	ALR/Lt strain, NOD/Lt and NOD/LtDvs mouse sub-strains	Mathews et al. 2005
m.T10003C	$tRNA^{Gly}$	MIDD	Han Chinese family with MIDD	Li et al. 2015b, Liu et al. 2015

m.C12258A	$tRNA^{Ser}$	MIDD	British family case with MIDD	Lynn et al. 1998, Choo-Kang et al. 2002
m.G13997A**	ND6	T2D	Aged mito-mice $ND6^{M}$ mouse strain	Hashizume et al. 2012
m.T14577C	ND6	T2D	Japanese family	Tawata et al. 2000
m.T14577C*	$tRNA^{Glu}$	MIDD	Han Chinese patients	Wang et al. 2016
m.A14693G*	$tRNA^{Glu}$	MIDD	Taiwan MELAS patient with DM mother and healthy siblings	Tzen et al. 2003
m.T14709C	$tRNA^{Glu}$	T2D	Patient with muscle weakness due to COX deficiency	Hao et al. 1995
m.A15746G	CYB	T2D	Taiwan T2D patients	Loo et al. 2014
m.T16093C*	Non-coding region	MIDD	Chinese patients with MIDD	Jiang et al. 2019
m.T16189C	mtDNA control region for replication and transcription	T2D	Asian patients	Park et al. 2008
			UK patients	Poulton et al. 2002
			Chinese patients	Weng et al. 2005
			Finnish patients	Soini et al. 2012

*- mutation identified in addition to m.3243G;
**- mouse model

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