Table S3 MtDNA variants and their percentage detected in control subjects or both in control subjects and Mitochondrial Diabetic patients

	mtDNA			
Region	Nucleotide change	Amino acid change	Mitochondrial Diabetic patients n (%)	Control subjects n (%)
D-Loop	72T>C		2 (18.2)	2 (2.5)
D-Loop	73A>G		2 (18.2)	47 (58.8)
D-Loop	93A>G			2 (2.5)
D-Loop	114C>T			2 (2.5)
D-Loop	143G>A			3 (3.7)
D-Loop	146T>C		1 (9.1)	8 (10.0)
D-Loop	150C>T			11 (13.7)
D-Loop	151C>T			3 (3.7)
D-Loop	152T>C		3 (27.3) [a]	15 (18.8)
D-Loop	153A>G			6 (7.5)
D-Loop	183A>G			1 (1.2)
D-Loop	185G>A			3 (3.7)
D-Loop	189A>G			3 (3.7)
D-Loop	194C>T			2 (2.5)
D-Loop	195T>C		1 (9.1)	14 (17.5)
D-Loop	198C>T			2 (2.5)
D-Loop	199T>C			2 (2.5)
D-Loop	200A>G			1 (1.2)
D-Loop	204T>C			5 (6.2)
D-Loop	207G>A		1 (9.1)	3 (3.7)
D-Loop	210A>G			1 (1.2)
D-Loop	225G>A			4 (5.0)
D-Loop	228G>A			4 (5.0)
D-Loop	250T>C			2 (2.5)
D-Loop	263A>G		11 (100)	71 (88.7)
D-Loop	**281A>C**			**1 (1.2)**
D-Loop	294T>C			1 (1.2)
D-Loop	295C>T			7 (8.7)
D-Loop	310T>C		3 (27.3)	36 (45.0)
D-Loop	456C>T			2 (2.5)
D-Loop	462C>T			5 (6.2)
D-Loop	468C>T			1 (1.2)
D-Loop	482T>C			3 (3.7)
D-Loop	487A>G			1 (1.2)
D-Loop	489T>C			9 (11.2)
D-Loop	497C>T		1 (9.1)	2 (2.5)
D-Loop	499G>A			1 (1.2)
D-Loop	523 delAC		1 (9.1)	5 (6.2)
D-Loop	**574A>C**			**2 (2.5)**
MT-RNR1	709G>A		3 (27.3)	10 (12.5)
MTRNR1	748G>A			1 (1.2)
MT-RNR1	750A>G		11 (100)	69 (86.2)

MTRNR1	930G>A			3 (3.7)
MTRNR1	961T>C			2 (2.5)
MTRNR1	980T>C			2 (2.5)
MTRNR1	1018G>A			1 (1.2)
MT-RNR1	1189T>C		1 (9.1)	4 (5.0)
MTRNR1	1243T>C			2 (2.5)
MT-RNR1	1438A>G		10 (90.9)	74 (92.5)
MTRNR1	1598G>A			2 (2.5)
MTRNR2	1700T>C			3 (3.7)
MTRNR2	1719G>A			10 (12.5)
MT-RNR2	1811A>G		1 (9.1)	12 (15.0)
MTRNR2	1888G>A			5 (6.2)
MTRNR2	2259C>T			1 (1.2)
MTRNR2	2294A>G			2 (2.5)
MTRNR2	2352T>C			1 (1.2)
MTRNR2	2416T>C			1 (1.2)
MTRNR2	2639C>T			2 (2.5)
MT-RNR2	2706A>G		1 (9.1)	41 (51.2)
MTRNR2	2789C>T			1 (1.2)
MTRNR2	3010G>A			11 (13.7)
MTRNR2	3027T>C			2 (2.5)
MT-RNR2	3197T>C		1 (9.1)	6 (7.5)
MTRNR2	3200T>C			1 (1.2)
MTRNR2	3202T>C			1 (1.2)
MTRNR2	**3213A>G**			**1 (1.2)**
MT-TL1	3290T>C			1 (1.2)
MT-ND1	3316G>A	A-T		1 (1.2)
MT-ND1	3394T>C	Y-H		1 (1.2)
MT-ND1	3396T>C	syn		1 (1.2)
MT-ND1	3421G>A	V-I		1 (1.2)
MT-ND1	**3456T>C**	**syn**		**1 (1.2)**
MT-ND1	3480A>G	syn	1 (9.1)	3 (3.7)
MT-ND1	3505A>G	T-A		2 (2.5)
MT-ND1	3546C>A	syn		2 (2.5)
MT-ND1	3579A>G	syn		13 (16.2)
MT-ND1	3594C>T	syn		1 (1.2)
MT-ND1	3834G>A	syn		1 (1.2)
MT-ND1	3921C>A	syn		2 (2.5)
MT-ND1	**3984C>T**	**syn**		**1 (1.2)**
MT-ND1	3992C>T	T-M	1 (9.1)	1 (1.2)
MT-ND1	**4002T>C**	**syn**		**1 (1.2)**
MT-ND1	4011C>T	syn		1 (1.2)
MT-ND1	4029C>T	syn		1 (1.2)
MT-ND1	**4050C>T**	**syn**		**1 (1.2)**
MT-ND1	4093A>G	T-A		2 (2.5)
MT-ND1	4104A>G	syn		1 (1.2)
MT-ND1	4188A>G	syn		3 (3.7)
MT-ND1	4216T>C	Y-H		13 (16.2)

MT-ND1	**4221C>A**	**syn**		**1 (1.2)**
MT-ND1	4232T>C	I-T		1 (1.2)
MT-ND1	**4243A>G**	**syn**		**1 (1.2)**
MT-TQ	4336T>C	T-C		1 (1.2)
MT-ND2	**4512G>A**	**A-T**		**1 (1.2)**
MT-ND2	4529A>T	syn		2 (2.5)
MT-ND2	4561T>C	V-A		1 (1.2)
MT-ND2	4580G>A	syn		2 (2.5)
MT-ND2	4586T>C	syn		1 (1.2)
MT-ND2	4639T>C	I-T		1 (1.2)
MT-ND2	4640C>A	I-M		3 (3.7)
MT-ND2	4646T>C	syn		2 (2.5)
MT-ND2	4659G>A	A-T		1 (1.2)
MT-ND2	**4677C>T**	**L-F**		**1 (1.2)**
MT-ND2	4688T>C	syn		1 (1.2)
MT-ND2	4703T>C	syn		2 (2.5)
MT-ND2	4736T>C	syn		1 (1.2)
MT-ND2	**4749T>C**	**S-P**		**1 (1.2)**
MT-ND2	4769A>G	syn	11 (100)	73 (91.2)
MT-ND2	4793A>G	syn		1 (1.2)
MT-ND2	4907T>C	syn		1 (1.2)
MT-ND2	4917A>G	N-D		6 (7.5)
MT-ND2	**4953A>G**	**I-V**		**1 (1.2)**
MT-ND2	4960C>T	A-V		2 (2.5)
MT-ND2	5004T>C	syn	1 (9.1)	1 (1.3)
MT-ND2	5046G>A	V-I		2 (2.5)
MT-ND2	5054G>A	syn		2 (2.5)
MT-ND2	**5081T>C**	**syn**		**1 (1.2)**
MT-ND2	5130T>C	syn		1 (1.2)
MT-ND2	**5135C>T**	**syn**		**1 (1.2)**
MT-ND2	5147G>A	syn	1 (9.1)	3 (3.7)
MT-ND2	5198A>G	syn		2 (2.5)
MT-ND2	5261G>A	syn		1 (1.2)
MT-ND2	5319A>G	T-S		2 (2.5)
MT-ND2	5360C>T	syn		1 (1.2)
MT-ND2	5460G>A	A-T		3 (3.7)
MT-ND2	5471G>A	syn	1 (9.1)	3 (3.7)
MT-ND2	5495T>C	syn		1 (1.2)
MT-ND2	5498A>G	syn		1 (1.2)
MT-TW	**5530C>T**			**1 (1.2)**
MT-TA	5633C>T			3 (3.7)
MT-TA	5655T>C			1 (1.2)
MT-NC4	5656A>G			1 (1.2)
MT-CO1	5999T>C	syn		1 (1.2)
MT-CO1	6026G>A	syn		1 (1.2)
MT-CO1	6047A>G	syn		1 (1.2)
MT-CO1	6125A>G	syn		1 (1.2)
MT-CO1	**6126A>G**	**I-V**		**1 (1.2)**

MT-CO1	6173C>T	syn		1 (1.2)
MT-CO1	6221T>C	syn	1 (9.1)	6 (7.5)
MT-CO1	6261G>A	A-T		1 (1.2)
MT-CO1	6266A>G	syn		1 (1.2)
MT-CO1	**6297T>C**	**syn**		**1 (1.2)**
MT-CO1	**6314C>A**	**syn**		**1 (1.2)**
MT-CO1	6359A>G	syn		1 (1.2)
MT-CO1	6365T>C	syn		1 (1.2)
MT-CO1	6371C>T	syn		5 (6.2)
MT-CO1	**6518C>T**	**syn**		**1 (1.2)**
MT-CO1	6524T>C	syn		2 (2.5)
MT-CO1	6546C>T	L-F		1 (1.2)
MT-CO1	6599A>G	syn		1 (1.2)
MT-CO1	6629A>G	syn		2 (2.5)
MT-CO1	6663A>G	syn		1 (1.2)
MT-CO1	6719T>C	syn		2 (2.5)
MT-CO1	6734G>A	syn		3 (3.7)
MT-CO1	6776T>C	syn	1 (9.1)	2 (2.5)
MT-CO1	**6899G>A**	**syn**		**1 (1.2)**
MT-CO1	**6975T>C**	**syn**		**1 (1.2)**
MT-CO1	7028C>T	syn	4 (36.4)	51 (63.7)
MT-CO1	7055A>G	syn		2 (2.5)
MT-CO1	7076A>G	syn		1 (1.2)
MT-CO1	7094T>C	syn		2 (2.5)
MT-CO1	7175T>C	syn		1 (1.2)
MT-CO1	7184A>G	syn		1 (1.2)
MT-CO1	7226G>A	syn		1 (1.2)
MT-CO1	7247C>T	syn		1 (1.2)
MT-CO1	7256C>T	syn		1 (1.2)
MT-CO1	7274C>T	syn		1 (1.2)
MT-CO1	7299A>G	M-V		1 (1.2)
MT-TS1	7476C>T			2 (2.5)
MT-TD	7521G>A			1 (1.2)
MT-TD	7547T>C			1 (1.2)
MT-TD	**7572T>C**			**1 (1.2)**
MT-CO2	7598G>A	A-T		1 (1.2)
MT-CO2	7621T>C	syn		1 (1.2)
MT-CO2	7645T>C	syn		2 (2.5)
MT-CO2	**7746A>G**	**N-S**		**1 (1.2)**
MT-CO2	7768A>G	syn		1 (1.2)
MT-CO2	7771A>G	syn		1 (1.2)
MT-CO2	7789G>A	syn		1 (1.2)
MT-CO2	7844A>G	T-A		1 (1.2)
MT-CO2	7858C>T	syn		1 (1.2)
MT-CO2	7961T>C	syn		1 (1.2)
MT-CO2	7963A>G	syn		1 (1.2)
MT-CO2	8137C>T	syn		1 (1.2)
MT-CO2	8152G>A	syn		3 (3.7)

MT-CO2	8206G>A	syn		1 (1.2)
MT-CO2	8222T>C	syn		1 (1.2)
MT-CO2	8251G>A	syn		7 (8.7)
MT-ATP8	8389A>G	syn		1 (1.2)
MT-ATP8	8393C>T	P-S		1 (1.2)
MT-ATP8	8448T>C	M-T		1 (1.2)
MT-ATP8	8460A>G	N-S		1 (1.2)
MT-ATP8	8472C>T	P-L		2 (2.5)
MT-ATP8	**8555T>C**	**syn**		**1 (1.2)**
MT-ATP6	8572G>A	G-S		1 (1.2)
MT-ATP8	8572G>A	Ter-Ter		1 (1.2)
MT-ATP6	**8596A>G**	**I-V**		**1 (1.2)**
MT-ATP6	8614T>C	syn		1 (1.2)
MT-ATP6	**8633A>G**	**Y-L**		**1 (1.2)**
MT-ATP6	8684C>T	T-I		1 (1.2)
MT-ATP6	8697G>A	syn		6 (7.5)
MT-ATP6	8701A>G	T-A		2 (2.5)
MT-ATP6	8705T>C	M-T		1 (1.2)
MT-ATP6	**8806C>T**	**P-S**		**1 (1.2)**
MT-ATP6	8818C>T	syn		1 (1.2)
MT-ATP6	8836A>G	M-V		2 (2.5)
MT-ATP6	8860A>G	T-A	10 (90.9)	69 (86.2)
MT-ATP6	8865G>A	syn		1 (1.2)
MT-ATP6	8869A>G	M-V		1 (1.2)
MT-ATP6	8938A>G	I-V		1 (1.2)
MT-ATP6	8994G>A	syn		1 (1.2)
MT-ATP6	9033A>G	syn		1 (1.2)
MT-ATP6	9090T>C	syn		4 (5.0)
MT-ATP6	9111T>C	syn		1 (1.2)
MT-ATP6	9123G>A	syn		1 (1.2)
MT-ATP6	**9139G>A**	**A-T**		**1 (1.2)**
MT-CO3	**9221A>G**	**syn**		**1 (1.2)**
MT-CO3	**9266G>A**	**syn**		**1 (1.2)**
MT-CO3	**9318C>T**	**H-Y**		**1 (1.2)**
MT-CO3	9335C>T	syn		2 (2.5)
MT-CO3	9452G>A	syn		1 (1.2)
MT-CO3	9477G>A	V-I	1 (9.1)	6 (7.5)
MT-CO3	9540T>C	syn		2 (2.5)
MT-CO3	**9656T>C**	**syn**		**4 (5.0)**
MT-CO3	9698T>C	syn	1 (9.1)	4 (5.0)
MT-CO3	9716T>C	syn		1 (1.2)
MT-CO3	**9742C>T**	**S-L**		**1 (1.2)**
MT-CO3	9756T>G	S-A		1 (1.2)
MT-CO3	9758T>C	syn		1 (1.2)
MT-CO3	9899T>C	syn		1 (1.2)
MT-CO3	**9903T>C**	**F-L**		**1 (1.2)**
MT-CO3	9932G>A	syn		2 (2.5)
MT-CO3	9948G>A	V-I		2 (2.5)

MT-CO3	9966G>A	V-I		2 (2.5)
MT-TG	10031T>C			1 (1.2)
MT-TG	10034T>C			2 (2.5)
MT-ND3	10115T>C	syn		1 (1.2)
MT-ND3	10142C>T	syn		1 (1.2)
MT-ND3	10143G>A	G-S		1 (1.2)
MT-ND3	10166T>C	syn		1 (1.2)
MT-ND3	10172G>A	syn		1 (1.2)
MT-ND3	10191T>C	S-P		1 (1.2)
MT-ND3	10217A>G	syn		3 (3.7)
MT-ND3	10237T>C	I-T		1 (1.2)
MT-ND3	10238T>C	syn		1 (1.2)
MT-ND3	10283A>G	syn		6 (7.5)
MT-ND3	**10322T>C**	**syn**		**1 (1.2)**
MT-ND3	**10334C>T**	**syn**		**1 (1.2)**
MT-ND3	**10356C>T**	**syn**		**1 (1.2)**
MT-ND3	10398A>G	T-A	1 (9.1)	13 (16.2)
MT-ND3	10400C>T	syn		1 (1.2)
MT-TR	10463T>C			7 (8.7)
MT-ND4L	10506A>G	T-A		2 (2.5)
MT-ND4L	10550A>G	syn	1 (9.1)	3 (3.7)
MT-ND4L	10586G>A	syn		1 (1.2)
MT-ND4L	**10739A>G**	**syn**		**1 (1.2)**
MT-ND4	10810T>C	syn		2 (2.5)
MT-ND4	10873T>C	syn		2 (2.5)
MT-ND4	10978A>G	syn		1 (1.2)
MT-ND4	11016G>A	S-N		1 (1.2)
MT-ND4	11025T>C	C-P		1 (1.2)
MT-ND4	11061C>T	S-F		1 (1.2)
MT-ND4	11242C>G	syn		1 (1.2)
MT-ND4	11251A>G	syn		13 (16.2)
MT-ND4	11299T>C	syn	1 (9.1)	4 (5.0)
MT-ND4	11332C>T	syn		1 (1.2)
MT-ND4	11362A>G	syn		2 (2.5)
MT-ND4	11437T>C	syn		1 (1.2)
MT-ND4	11440G>A	syn		1 (1.2)
MT-ND4	11467A>G	syn	2 (18.2)	18 (22.5)
MT-ND4	11653A>G	syn		1 (1.2)
MT-ND4	11674C>T	syn		2 (2.5)
MT-ND4	11719G>A	syn	2 (18.2)	45 (56.2)
MT-ND4	11812A>G	syn		5 (6.2)
MT-ND4	**11827T>C**	**syn**		**1 (1.2)**
MT-ND4	11914G>A	syn		3 (3.7)
MT-ND4	11944T>C	syn		1 (1.2)
MT-ND4	11947A>G	syn		2 (2.5)
MT-ND4	11971C>T	syn		1 (1.2)
MT-TS2	12234A>G			1 (1.2)
MT-TL2	**12246C>T**			**1 (1.2)**

MT-ND5	12346C>T	H-Y		2 (2.5)
MT-TL2	12308A>G		2 (18.2)	18 (22.5)
MT-ND5	12372G>A	syn	2 (18.2)	18 (22.5)
MT-ND5	**12378C>T**	**syn**		**1 (1.2)**
MT-ND5	12414T>C	syn		2 (2.5)
MT-ND5	12501G>A	syn		5 (6.2)
MT-ND5	12612A>G	syn		6 (7.5)
MT-ND5	12633C>T	syn		2 (2.5)
MT-ND5	12693A>G	syn		1 (1.2)
MT-ND5	12705C>T	syn		14 (17.5)
MT-ND5	12771G>A	syn		1 (1.2)
MT-ND5	12930A>G	syn		1 (1.2)
MT-ND5	12937A>G	M-V		1 (1.2)
MT-ND5	13368G>A	syn		6 (7.5)
MT-ND5	13617T>C	syn	1 (9.1)	8 (10.0)
MT-ND5	13708G>A	A-T		8 (10.0)
MT-ND5	13743T>C	syn		4 (5.0)
MT-ND5	13759G>A	A-T		2 (2.5)
MT-ND5	13780A>G	I-V		2 (2.5)
MT-ND5	13934C>T	T-M		3 (3.7)
MT-ND5	13966A>G	T-A		5 (6.2)
MT-ND5	14139A>G	syn		6 (7.5)
MT-ND6	14167C>T	syn	1 (9.1)	5 (6.2)
MT-ND6	14233A>G	syn		5 (6.2)
MT-ND6	14470T>C	syn		5 (6.2)
MT-ND6	14569G>A	syn	3 (27.3)	1 (1.2)
MT-CYB	14766C>T	T-I	2 (18.2)	45 (56.2)
MT-CYB	14769A>G	N-S		1 (1.2)
MT-CYB	14783T>C	syn		1 (1.2)
MT-CYB	14793A>G	H-R	1 (9.1)	5 (6.2)
MT-CYB	14798T>C	F-L	1 (9.1)	8 (10.0)
MT-CYB	14905G>A	syn		6 (7.5)
MT-CYB	15043G>A	syn		3 (3.7)
MT-CYB	15071T>C	Y-N		1 (1.2)
MT-CYB	15110G>A	A-T	1 (9.1)	1 (1.2)
MT-CYB	15218A>G	T-A		5 (6.2)
MT-CYB	15301G>A	syn		2 (2.5)
MT-CYB	15326A>G	T-A	11 (100)	71 (88.7)
MT-CYB	15452C>A	L-I		13 (16.2)
MT-CYB	15454T>C	syn		4 (5.0)
MT-CYB	15607A>G	syn		6 (7.5)
MT-TT	15904C>T			2 (2.5)
MT-TT	15910C>T			1 (1.2)
MT-TT	15924A>G			5 (6.2)
MT-TT	15928G>A			6 (7.5)
D-Loop	16069C>T			7 (8.7)
D-Loop	16074A>G			1 (1.2)
D-Loop	16086T>C			1 (1.2)

D-Loop	16093T>C		4 (5.0)
D-Loop	16126T>C		15 (18.7)
D-Loop	16129G>A	1 (9.1)	3 (3.8)
D-Loop	16134C>T		1 (1.2)
D-Loop	16140T>C		2 (2.5)
D-Loop	16142C>T		1 (1.2)
D-Loop	16145G>A		2 (2.5)
D-Loop	16148C>T		1 (1.2)
D-Loop	16150C>T		1 (1.2)
D-Loop	16153G>A		1 (1.2)
D-Loop	16154T>C		1 (1.2)
D-Loop	16162A>G		1 (1.2)
D-Loop	16163A>G		1 (1.2)
D-Loop	16172T>C		1 (1.2)
D-Loop	16176C>G		2 (2.5)
D-Loop	16180A>G		1 (1.2)
D-Loop	16182A>C		1 (1.2)
D-Loop	16183A>C		4 (5.0)
D-Loop	16184C>T		1 (1.2)
D-Loop	16185C>T		1 (1.2)
D-Loop	16186C>T		1 (1.2)
D-Loop	16188C>T	3 (27.3)	1 (1.2)
D-Loop	16189T>C	1 (9.1)	12 (15.0)
D-Loop	16192C>T	1 (9.1)	7 (8.8)
D-Loop	16193C>T		4 (5.0)
D-Loop	16224T>C	1 (9.1)	5 (6.2)
D-Loop	16233A>G	1 (9.1)	13 (16.2)
D-Loop	16256C>T	1 (9.1)	9 (11.2)
D-Loop	16260C>T		2 (2.5)
D-Loop	16270C>T	1 (9.1)	9 (11.2)
D-Loop	16278C>T	1 (9.1) [a]	6 (7.5)
D-Loop	**16285A>C**		**1 (1.2)**
D-Loop	16286C>A		1 (1.2)
D-Loop	16288T>C		3 (3.7)
D-Loop	16289A>G		5 (6.2)
D-Loop	16290C>A		1 (1.2)
D-Loop	16292C>T		3 (3.7)
D-Loop	16293A>C		9 (11.2)
D-Loop	16294C>T		7 (8.7)
D-Loop	16296C>T		4 (5.0)
D-Loop	16298T>C	2 (18.2)	3 (3.7)
D-Loop	16304T>C		7 (8.7)
D-Loop	16311T>C	3 (27.3)	13 (16.2)
D-Loop	16325T>C		3 (3.7)
D-Loop	16328C>A		4 (5.0)
D-Loop	16352T>C		4 (5.0)
D-Loop	16355C>T		2 (2.5)
D-Loop	16362T>C	1 (9.1)	4 (5.0)

D-Loop	16384G>A		6 (7.5)
D-Loop	16390G>A		6 (7.5)
D-Loop	16391G>A		2 (2.5)
D-Loop	**16402A>C**		**2 (2.5)**
D-Loop	16519T>C	7 (63.6)	29 (36.2)
D-Loop	16527C>T		1 (1.2)

In bold are reported novel variants; [a]Variant present at heteroplasmic level.

 For previously described variants see www.mitomap.org.

The variants detected in Mitochondrial diabetic affected patients were always present also in their mothers.