

Discovery of mitochondrial DNA variants associated with
genome-wide blood cell gene expression: a
population-based mtDNA sequencing study

Supplementary tables

Table S1. Pairwise conditional analysis results. P_{bonf} indicates the mtSNP-specific Bonferroni-corrected p-value (i.e. $0.05 / \text{the number of pairwise conditional analyses made for each mtSNP}$) which was defined as the limit of significance. MtSNPs that survived all conditional analyses are shown in green. Cells in grey indicate mtSNPs that had pairwise $r^2 < 0.30$.

SPCS2P4

mtSNP	Original p-value	P_{Bonf}	Conditioned on															
			G9055A	A3480G	A10550G	C14167T	T16224C	T11299C	T1189C	T9698C	A9093G	T9903C	T14798C	A1811G	G11377A	A11251G	C15452A	A10398G
G9055A	1.81E-39	4.17E-03		1.80E-09	2.02E-09	1.90E-09	7.61E-12	4.07E-12	2.57E-14	4.91E-22	3.44E-23	2.02E-24	3.76E-26	2.82E-27	4.14E-29			
A3480G	4.84E-32	3.85E-03	4.88E-01		1.57E-09	6.07E-04	2.05E-05	2.17E-04	1.36E-06	2.83E-14	1.83E-15	7.38E-17	2.18E-18	1.36E-19	1.60E-21			
A10550G	9.49E-32	3.85E-03	5.16E-01	5.36E-08		6.39E-04	2.11E-04	2.25E-04	1.63E-06	2.94E-14	2.68E-15	1.16E-16	4.24E-18	1.61E-19	2.86E-21			
C14167T	9.49E-32	3.85E-03	5.07E-01	5.87E-08	1.97E-09		2.16E-04	2.28E-04	1.49E-06	3.13E-14	2.51E-15	1.13E-16	4.85E-18	1.69E-19	2.52E-21			
T16224C	1.77E-30	4.17E-03	2.19E-01	5.05E-02	4.43E-02	4.43E-02		1.76E-03	6.85E-05	2.00E-11	3.45E-14	1.06E-15		1.56E-17	1.50E-19			
T11299C	5.94E-29	3.85E-03	2.12E-01	1.16E-01	1.15E-01	1.15E-01	4.44E-03		2.95E-03	4.83E-11	8.65E-13	2.98E-14	2.14E-15	1.26E-16	6.86E-17			
T1189C	1.17E-27	3.85E-03	1.25E-01	7.43E-02	7.53E-02	7.55E-02	2.03E-02	5.40E-01		2.45E-09	4.17E-11	1.46E-12	6.69E-14	6.18E-16	6.83E-17			
T9698C	1.01E-19	4.55E-03	7.75E-02	7.86E-02	7.54E-02	7.75E-02	3.81E-01	5.20E-01	7.70E-01		3.56E-06	1.84E-07		3.69E-08	5.08E-10			
A9093G	4.72E-18	5.00E-03	6.73E-01	8.74E-01	8.72E-01	8.74E-01	7.86E-02	5.56E-01	6.69E-01	7.20E-04		8.02E-03			1.05E-07			
T9903C	7.24E-17	5.00E-03	9.11E-01	7.17E-01	7.15E-01	7.15E-01	6.69E-02	3.24E-01	3.95E-01	2.48E-04	3.74E-01				6.78E-06			
T14798C	9.74E-16	7.14E-03	8.77E-01	9.69E-01	9.86E-01	9.66E-01		5.46E-01	2.38E-01									4.58E-08
A1811G	4.86E-14	5.00E-02	8.40E-01	7.15E-01	7.33E-01	7.27E-01	7.64E-02	3.78E-01	1.93E-01	4.38E-02								
G11377A	2.19E-12	4.55E-03	2.12E-01	2.78E-01	2.72E-01	2.77E-01	6.45E-01	6.54E-01	5.07E-01	2.59E-02	1.18E-01	7.25E-01		2.38E-05				
A11251G	2.64E-11	5.00E-02															1.41E-03	
C15452A	5.57E-11	5.00E-02														4.28E-06		
A10398G	1.80E-09	5.00E-02											1.63E-01					

RNF113A

mtSNP	Original p-value	P_{Bonf}	Conditioned on															
			G9055A	C14167T	A10550G	A3480G	T11299C	T1189C	T16224C	T9698C	A9093G	T9903C	T14798C	T4216C	A1811G	C15452C	A11251G	G11377A
G9055A	2.48E-21	4.17E-03		1.40E-04	1.51E-04	1.48E-04	2.68E-05	6.97E-06	1.61E-09	6.65E-11	3.62E-11	1.84E-12	4.56E-13		5.51E-14			2.82E-14
C14167T	1.87E-18	4.17E-03	9.81E-01		9.94E-05	3.72E-08	7.39E-02	1.63E-02	2.45E-06	8.93E-08	3.99E-08	1.79E-09	5.35E-10		8.10E-11			2.69E-11
A10550G	1.95E-18	4.17E-03	9.50E-01	7.49E-01		3.23E-08	7.28E-02	1.62E-02	2.36E-06	8.44E-08	3.95E-08	1.86E-09	9.07E-10		7.53E-11			2.80E-11
A3480G	2.70E-18	4.17E-03	9.94E-01	7.88E-01	1.00E-04		7.59E-02	1.67E-02	1.94E-05	8.47E-08	4.61E-08	2.28E-09	6.82E-10		8.08E-11			3.23E-11
T11299C	1.21E-17	4.17E-03	8.89E-01	8.14E-01	8.15E-01	8.09E-01		7.22E-01	1.63E-05	7.66E-07	1.75E-07	8.55E-09	9.68E-09		3.87E-10			7.27E-11
T1189C	4.42E-17	4.17E-03	9.20E-01	8.57E-01	8.53E-01	8.58E-01	3.97E-01		6.62E-05	3.40E-06	8.06E-07	3.50E-08	6.37E-09		1.35E-09			5.05E-10
T16224C	8.58E-14	4.55E-03	5.50E-01	4.33E-01	4.31E-01	4.92E-01	8.84E-01	7.16E-01		5.00E-03	8.24E-05	8.71E-06			9.30E-07			4.60E-07

Table S1 (continued). Pairwise conditional analysis results. P_{bonf} indicates the mtSNP-specific Bonferroni-corrected p-value (i.e. 0.05 / the number of pairwise conditional analyses made for each mtSNP) which was defined as the limit of significance. MtSNPs that survived all conditional analyses are shown in green. Cells in grey indicate mtSNPs that had pairwise $r^2 < 0.30$.

RNF113A

mtSNP	Original p-value	P_{Bonf}	Conditioned on															
			G9055A	C14167T	A10550G	A3480G	T11299C	T1189C	T16224C	T9698C	A9093G	T9903C	T14798C	T4216C	A1811G	C15452C	A11251G	G11377A
T9698C	2.57E-12	4.55E-03	4.47E-01	3.28E-01	3.23E-01	3.16E-01	6.69E-01	9.65E-01	3.95E-02		5.65E-04	7.29E-05			6.10E-05			5.12E-06
A9093G	7.40E-12	5.00E-03	6.93E-01	6.90E-01	6.91E-01	6.90E-01	4.54E-01	6.41E-01	2.30E-02	3.54E-03		1.32E-01						1.02E-04
T9903C	2.04E-10	5.00E-03	6.52E-01	6.40E-01	6.39E-01	6.37E-01	4.42E-01	6.05E-01	2.03E-02	5.70E-03	8.10E-01							2.90E-03
T14798C	4.76E-10	8.33E-03	7.24E-01	8.20E-01	7.86E-01	8.05E-01	5.95E-01	4.00E-01										
T4216C	1.44E-19	2.50E-02														7.38E-02	7.68E-02	
A1811G	3.43E-09	6.25E-03	5.53E-01	5.62E-01	5.74E-01	5.92E-01	4.14E-01	2.95E-01	3.13E-02	9.82E-02								
C15452C	5.22E-09	2.50E-02												2.00E-01			3.05E-02	
A11251G	7.84E-09	2.50E-02												2.22E-01		4.54E-03		
G11377A	1.18E-08	5.00E-03	8.46E-01	8.21E-01	8.23E-01	8.19E-01	9.84E-01	8.38E-01	1.05E-01	3.99E-02	3.56E-01	4.32E-01						

SPCS2

mtSNP	Original p-value	P_{Bonf}	Conditioned on						
			G9055A	A3480G	A10550G	C14167T	T16224C	T11299C	T1189C
G9055A	9.59E-14	8.33E-03		3.68E-05	3.56E-05	3.72E-05	2.71E-05	1.42E-06	1.38E-06
A3480G	4.81E-10	8.33E-03	2.07E-01		6.50E-03	5.26E-03	4.25E-02	7.77E-03	1.44E-02
A10550G	5.04E-10	8.33E-03	2.06E-01	2.32E-03		5.46E-03	6.81E-02	8.21E-03	1.39E-02
C14167T	5.44E-10	8.33E-03	2.14E-01	2.22E-03	6.45E-03		6.87E-02	8.28E-03	1.37E-02
T16224C	9.72E-10	8.33E-03	8.66E-01	2.47E-01	2.57E-01	2.59E-01		5.16E-02	3.36E-02
T11299C	7.40E-09	8.33E-03	7.46E-02	1.20E-01	1.20E-01	1.20E-01	2.76E-01		2.75E-01
T1189C	8.27E-09	8.33E-03	1.26E-01	3.79E-01	3.79E-01	3.76E-01	2.59E-01	3.76E-01	

CRLS1

mtSNP	Original p-value	P_{Bonf}	Conditioned on		
			A8869G	T4639C	C5263T
A8869G	5.30E-13	2.50E-02		2.16E-08	3.15E-02
T4639C	8.45E-13	2.50E-02	4.60E-04		3.19E-02
C5263T	5.28E-11	2.50E-02	8.67E-01	8.71E-01	

Table S1 (continued). Pairwise conditional analysis results. P_{bonf} indicates the mtSNP-specific Bonferroni-corrected p-value (i.e. 0.05 / the number of pairwise conditional analyses made for each mtSNP) which was defined as the limit of significance. MtSNPs that survived all conditional analyses are shown in green. Cells in grey indicate mtSNPs that had pairwise $r^2 < 0.30$.

SLC25A14

mtSNP	Original p-value	P_{Bonf}	Conditioned on							
			A3505G	T1243C	A11947G	G8994A	G5046A	G15884C	T12414C	G5460A
A3505G	3.28E-10	7.14E-03		2.40E-07	7.41E-04	3.71E-04	6.42E-02	1.83E-04	1.02E-02	5.22E-03
T1243C	4.54E-10	7.14E-03	8.51E-08		1.49E-03	7.14E-04	3.83E-02	2.12E-03	1.00E-02	6.31E-03
A11947G	1.03E-09	7.14E-03	1.65E-07	1.88E-07		1.04E-03	4.51E-02	2.98E-03	1.20E-02	7.30E-03
G8994A	1.09E-09	7.14E-03	5.30E-08	2.30E-07	1.88E-03		4.77E-02	3.03E-03	1.18E-02	7.99E-03
G5046A	1.76E-09	7.14E-03	1.82E-07	2.76E-07	2.03E-03	1.45E-03		4.06E-03	1.29E-02	8.09E-03
G15884C	2.25E-09	7.14E-03	2.11E-07	3.38E-07	2.04E-03	1.36E-03	5.49E-02		1.36E-02	8.22E-03
T12414C	8.01E-09	7.14E-03	1.93E-01	1.91E-01	2.00E-01	2.05E-01	7.90E-02	2.06E-01		7.90E-03
G5460A	9.93E-09	7.14E-03	7.52E-01	7.54E-01	7.62E-01	7.71E-01	7.64E-01	7.69E-01	3.11E-01	

Table S2. Minor allele frequency and mitochondrial locus of 199 mtSNPs.

mtSNP	Mitochondrial locus	MAF
T72C	Control-Region	0.061
A93G	Control-Region	0.027
A153G	Control-Region	0.019
G185A	Control-Region	0.057
C186A	Control-Region	0.012
A188G	Control-Region	0.047
A189G	Control-Region	0.054
C194T	Control-Region	0.010
T199C	Control-Region	0.031
G203A	Control-Region	0.013
G207A	Control-Region	0.053
T217C	Control-Region	0.060
G225A	Control-Region	0.021
A227G	Control-Region	0.035
G228A	Control-Region	0.040
T250C	Control-Region	0.017
C295T	Control-Region	0.072
C456T	Control-Region	0.022
C462T	Control-Region	0.062
T477C	Control-Region	0.020
T485C	Control-Region	0.013
T489C	Control-Region	0.079
G709A	rRNA	0.124
G930A	rRNA	0.017
G951A	rRNA	0.022
T1189C	rRNA	0.035
T1243C	rRNA	0.045
C1341T	rRNA	0.032
G1719A	rRNA	0.044
C1721T	rRNA	0.010
A1811G	rRNA	0.077
G1888A	rRNA	0.050
G2098A	rRNA	0.029
C2259T	rRNA	0.028
A3434G	ND1	0.021
A3480G	ND1	0.037
A3505G	ND1	0.045
C3549T	ND1	0.020
A3720G	ND1	0.010
C3990T	ND1	0.012
C3992T	ND1	0.010
C4059T	ND1	0.036
T4216C	ND1	0.125
T4336C	tRNA	0.010
T4452C	tRNA	0.040
A4529T	ND2	0.017
G4580A	ND2	0.061
T4639C	ND2	0.021

Table S2. Minor allele frequency and mitochondrial locus of 199 mtSNPs.

mtSNP	Mitochondrial locus	MAF
T4646C	ND2	0.025
A4745G	ND2	0.017
A4917G	ND2	0.053
T4928C	ND2	0.014
G5046A	ND2	0.045
G5147A	ND2	0.016
C5263T	ND2	0.019
A5390G	ND2	0.015
T5426C	ND2	0.015
G5460A	ND2	0.047
T5495C	ND2	0.020
A5656G	Non-Coding	0.138
T5999C	CO1	0.024
A6047G	CO1	0.024
T6152C	CO1	0.010
T6221C	CO1	0.021
G6261A	CO1	0.014
C6371T	CO1	0.021
G6734A	CO1	0.013
T6776C	CO1	0.026
G7013A	CO1	0.016
T7309C	CO1	0.040
G7337A	CO1	0.016
A7385G	CO1	0.080
G7444A	CO1	0.022
C7476T	tRNA	0.010
C7864T	CO2	0.037
G8251A	CO2	0.066
G8269A	CO2	0.012
T8448C	ATP8	0.023
A8537G	ATP8	0.012
G8616T	ATP6	0.012
G8697A	ATP6	0.051
T8705C	ATP6	0.024
T8715C	ATP6	0.012
C8818T	ATP6	0.016
A8842G	ATP6	0.013
T8843C	ATP6	0.015
A8869G	ATP6	0.021
C8898T	ATP6	0.013
A8923G	ATP6	0.018
G8994A	ATP6	0.045
G9039A	ATP6	0.013
G9053A	ATP6	0.010
G9055A	ATP6	0.043
A9066G	ATP6	0.040
A9093G	ATP6	0.022
C9391T	CO3	0.013

Table S2. Minor allele frequency and mitochondrial locus of 199 mtSNPs.

mtSNP	Mitochondrial locus	MAF
G9612A	CO3	0.014
A9667G	CO3	0.017
T9698C	CO3	0.048
T9903C	CO3	0.019
G9947A	CO3	0.014
T10034C	tRNA	0.017
A10086G	ND3	0.014
T10238C	ND3	0.023
A10398G	ND3	0.140
T10463C	tRNA	0.055
A10550G	ND4L	0.037
A10754G	ND4L	0.013
A10876G	ND4	0.010
T10915C	ND4	0.015
T10927C	ND4	0.080
C11191T	ND4	0.012
A11251G	ND4	0.123
T11299C	ND4	0.038
C11332T	ND4	0.024
G11377A	ND4	0.027
C11674T	ND4	0.047
A11812G	ND4	0.040
T11899C	ND4	0.022
G11914A	ND4	0.052
A11947G	ND4	0.045
G12127A	ND4	0.013
G12192A	tRNA	0.014
T12414C	ND5	0.046
G12501A	ND5	0.024
A12612G	ND5	0.072
G12613A	ND5	0.014
G12618A	ND5	0.136
G12630A	ND5	0.016
C12633A	ND5	0.010
C12669T	ND5	0.015
C12705T	ND5	0.096
A13152T	ND5	0.012
T13326C	ND5	0.014
G13368A	ND5	0.053
A13637G	ND5	0.010
C13680T	ND5	0.014
G13708A	ND5	0.079
T13734C	ND5	0.010
A13780G	ND5	0.023
A13827G	ND5	0.031
G13928C	ND5	0.031
A13966G	ND5	0.022
C14167T	ND6	0.037

Table S2. Minor allele frequency and mitochondrial locus of 199 mtSNPs.

mtSNP	Mitochondrial locus	MAF
A14233G	ND6	0.041
T14470C	ND6	0.021
C14482T	ND6	0.014
A14564G	ND6	0.014
C14620T	ND6	0.024
T14798C	CYB	0.096
T14857C	CYB	0.012
C14872T	CYB	0.028
G14905A	CYB	0.048
G15043A	CYB	0.036
A15218G	CYB	0.022
G15257A	CYB	0.012
C15452A	CYB	0.123
A15607G	CYB	0.050
T15693C	CYB	0.024
A15758G	CYB	0.014
G15884C	CYB	0.045
C15904T	tRNA	0.062
A15924G	tRNA	0.024
G15928A	tRNA	0.050
A16051G	Control-Region	0.012
C16069T	Control-Region	0.072
T16093C	Control-Region	0.075
C16114A	Control-Region	0.034
T16126C	Control-Region	0.119
T16144C	Control-Region	0.079
G16145A	Control-Region	0.013
G16153A	Control-Region	0.023
A16162G	Control-Region	0.061
T16172C	Control-Region	0.025
C16186T	Control-Region	0.010
C16223T	Control-Region	0.099
T16224C	Control-Region	0.045
G16255A	Control-Region	0.018
C16256T	Control-Region	0.066
C16261T	Control-Region	0.043
T16271C	Control-Region	0.016
C16278T	Control-Region	0.040
C16291T	Control-Region	0.022
C16292T	Control-Region	0.050
C16294T	Control-Region	0.086
C16296T	Control-Region	0.027
T16298C	Control-Region	0.074
C16301T	Control-Region	0.016
T16304C	Control-Region	0.035
T16311C	Control-Region	0.118
A16316G	Control-Region	0.016
G16319A	Control-Region	0.012

Table S2. Minor allele frequency and mitochondrial locus of 199 mtSNPs.

mtSNP	Mitochondrial locus	MAF
C16354T	Control-Region	0.022
C16355T	Control-Region	0.010
T16356C	Control-Region	0.038
T16362C	Control-Region	0.029
G16391A	Control-Region	0.018
A16399G	Control-Region	0.026
G16526A	Control-Region	0.038

Table S3. Minor allele frequency and mitochondrial locus of 156 mtSNPs used in sex-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (females)	MAF (males)
T72C	Control-Region	0.061	0.064	0.057
A93G	Control-Region	0.027	0.027	0.027
A153G	Control-Region	0.019	0.020	0.017
G185A	Control-Region	0.057	0.051	0.064
A188G	Control-Region	0.047	0.042	0.054
A189G	Control-Region	0.054	0.053	0.057
T199C	Control-Region	0.031	0.029	0.034
G203A	Control-Region	0.013	0.013	0.012
G207A	Control-Region	0.053	0.055	0.052
T217C	Control-Region	0.060	0.062	0.057
G225A	Control-Region	0.021	0.022	0.020
A227G	Control-Region	0.035	0.044	0.022
G228A	Control-Region	0.040	0.042	0.037
T250C	Control-Region	0.017	0.015	0.020
C295T	Control-Region	0.072	0.068	0.079
C456T	Control-Region	0.022	0.026	0.017
C462T	Control-Region	0.062	0.058	0.066
T477C	Control-Region	0.020	0.018	0.022
T485C	Control-Region	0.013	0.013	0.012
T489C	Control-Region	0.079	0.073	0.086
G709A	rRNA	0.124	0.128	0.118
G930A	rRNA	0.017	0.018	0.015
G951A	rRNA	0.022	0.020	0.025
T1189C	rRNA	0.035	0.029	0.042
T1243C	rRNA	0.045	0.046	0.044
C1341T	rRNA	0.032	0.031	0.034
G1719A	rRNA	0.044	0.042	0.047
A1811G	rRNA	0.077	0.060	0.101
G1888A	rRNA	0.050	0.049	0.052
G2098A	rRNA	0.029	0.036	0.020
C2259T	rRNA	0.028	0.036	0.017
A3434G	ND1	0.021	0.027	0.012
A3480G	ND1	0.037	0.033	0.042
A3505G	ND1	0.045	0.046	0.044
C3549T	ND1	0.020	0.020	0.020
C4059T	ND1	0.036	0.036	0.034
T4216C	ND1	0.125	0.119	0.133
T4452C	tRNA	0.040	0.047	0.029
A4529T	ND2	0.017	0.015	0.020
G4580A	ND2	0.061	0.062	0.059
T4639C	ND2	0.021	0.024	0.017
T4646C	ND2	0.025	0.020	0.032
A4745G	ND2	0.017	0.020	0.012
A4917G	ND2	0.053	0.053	0.054
G5046A	ND2	0.045	0.046	0.044
G5147A	ND2	0.016	0.016	0.015
C5263T	ND2	0.019	0.020	0.017
G5460A	ND2	0.047	0.047	0.047

Table S3. Minor allele frequency and mitochondrial locus of 156 mtSNPs used in sex-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (females)	MAF (males)
T5495C	ND2	0.020	0.013	0.029
A5656G	Non-Coding	0.138	0.150	0.123
T5999C	CO1	0.024	0.020	0.029
A6047G	CO1	0.024	0.020	0.029
T6221C	CO1	0.021	0.022	0.020
C6371T	CO1	0.021	0.022	0.020
G6734A	CO1	0.013	0.013	0.012
T6776C	CO1	0.026	0.029	0.022
G7013A	CO1	0.016	0.016	0.015
T7309C	CO1	0.040	0.047	0.029
G7337A	CO1	0.016	0.016	0.015
A7385G	CO1	0.080	0.084	0.074
G7444A	CO1	0.022	0.024	0.020
C7864T	CO2	0.037	0.035	0.039
G8251A	CO2	0.066	0.064	0.069
T8448C	ATP8	0.023	0.022	0.025
G8697A	ATP6	0.051	0.051	0.052
T8705C	ATP6	0.024	0.027	0.020
C8818T	ATP6	0.016	0.015	0.017
T8843C	ATP6	0.015	0.015	0.015
A8869G	ATP6	0.021	0.024	0.017
A8923G	ATP6	0.018	0.013	0.025
G8994A	ATP6	0.045	0.046	0.044
G9039A	ATP6	0.013	0.011	0.015
G9055A	ATP6	0.043	0.040	0.047
A9066G	ATP6	0.040	0.047	0.029
A9093G	ATP6	0.022	0.020	0.025
A9667G	CO3	0.017	0.020	0.012
T9698C	CO3	0.048	0.038	0.061
T9903C	CO3	0.019	0.020	0.017
T10034C	tRNA	0.017	0.015	0.020
T10238C	ND3	0.023	0.020	0.027
A10398G	ND3	0.140	0.128	0.157
T10463C	tRNA	0.055	0.055	0.057
A10550G	ND4L	0.037	0.033	0.042
T10915C	ND4	0.015	0.016	0.012
T10927C	ND4	0.080	0.084	0.074
A11251G	ND4	0.123	0.117	0.130
T11299C	ND4	0.038	0.035	0.042
C11332T	ND4	0.024	0.020	0.029
G11377A	ND4	0.027	0.024	0.032
C11674T	ND4	0.047	0.047	0.047
A11812G	ND4	0.040	0.035	0.047
T11899C	ND4	0.022	0.024	0.020
G11914A	ND4	0.052	0.062	0.039
A11947G	ND4	0.045	0.046	0.044
G12192A	tRNA	0.014	0.013	0.015
T12414C	ND5	0.046	0.047	0.044

Table S3. Minor allele frequency and mitochondrial locus of 156 mtSNPs used in sex-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (females)	MAF (males)
G12501A	ND5	0.024	0.020	0.029
A12612G	ND5	0.072	0.068	0.079
G12618A	ND5	0.136	0.148	0.120
G12630A	ND5	0.016	0.018	0.012
C12705T	ND5	0.096	0.095	0.098
G13368A	ND5	0.053	0.053	0.054
G13708A	ND5	0.079	0.071	0.088
A13780G	ND5	0.023	0.020	0.027
A13827G	ND5	0.031	0.033	0.029
G13928C	ND5	0.031	0.033	0.029
A13966G	ND5	0.022	0.022	0.022
C14167T	ND6	0.037	0.033	0.042
A14233G	ND6	0.041	0.036	0.047
T14470C	ND6	0.021	0.022	0.020
C14482T	ND6	0.014	0.015	0.012
A14564G	ND6	0.014	0.015	0.012
C14620T	ND6	0.024	0.020	0.029
T14798C	CYB	0.096	0.088	0.108
C14872T	CYB	0.028	0.036	0.017
G14905A	CYB	0.048	0.049	0.047
G15043A	CYB	0.036	0.029	0.044
A15218G	CYB	0.022	0.022	0.022
C15452A	CYB	0.123	0.117	0.130
A15607G	CYB	0.050	0.049	0.052
T15693C	CYB	0.024	0.020	0.029
G15884C	CYB	0.045	0.046	0.044
C15904T	tRNA	0.062	0.064	0.059
A15924G	tRNA	0.024	0.018	0.032
G15928A	tRNA	0.050	0.049	0.052
C16069T	Control-Region	0.072	0.068	0.079
T16093C	Control-Region	0.075	0.084	0.064
C16114A	Control-Region	0.034	0.033	0.034
T16126C	Control-Region	0.119	0.113	0.128
T16144C	Control-Region	0.079	0.084	0.071
G16145A	Control-Region	0.013	0.011	0.015
G16153A	Control-Region	0.023	0.026	0.020
A16162G	Control-Region	0.061	0.064	0.057
T16172C	Control-Region	0.025	0.027	0.022
C16223T	Control-Region	0.099	0.099	0.101
T16224C	Control-Region	0.045	0.038	0.054
G16255A	Control-Region	0.018	0.020	0.015
C16256T	Control-Region	0.066	0.071	0.059
C16261T	Control-Region	0.043	0.042	0.044
T16271C	Control-Region	0.016	0.018	0.012
C16278T	Control-Region	0.040	0.035	0.047
C16291T	Control-Region	0.022	0.022	0.022
C16292T	Control-Region	0.050	0.051	0.049
C16294T	Control-Region	0.086	0.086	0.086

Table S3. Minor allele frequency and mitochondrial locus of 156 mtSNPs used in sex-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (females)	MAF (males)
C16296T	Control-Region	0.027	0.024	0.032
T16298C	Control-Region	0.074	0.071	0.079
C16301T	Control-Region	0.016	0.016	0.015
T16304C	Control-Region	0.035	0.038	0.029
T16311C	Control-Region	0.118	0.122	0.113
A16316G	Control-Region	0.016	0.011	0.022
C16354T	Control-Region	0.022	0.020	0.025
T16356C	Control-Region	0.038	0.033	0.044
T16362C	Control-Region	0.029	0.022	0.039
G16391A	Control-Region	0.018	0.016	0.020
A16399G	Control-Region	0.026	0.027	0.025
G16526A	Control-Region	0.038	0.040	0.034

Table S4. Minor allele frequency and mitochondrial locus of 127 mtSNPs used in prediabetes-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (prediabetes)	MAF (controls)
T72C	Control-Region	0.056	0.040	0.063
G185A	Control-Region	0.056	0.044	0.062
A188G	Control-Region	0.047	0.036	0.051
A189G	Control-Region	0.058	0.056	0.058
T199C	Control-Region	0.032	0.028	0.034
G207A	Control-Region	0.055	0.044	0.060
T217C	Control-Region	0.062	0.064	0.062
A227G	Control-Region	0.036	0.028	0.039
G228A	Control-Region	0.040	0.032	0.043
T250C	Control-Region	0.018	0.020	0.017
C295T	Control-Region	0.071	0.072	0.070
C456T	Control-Region	0.024	0.024	0.024
C462T	Control-Region	0.060	0.048	0.065
T489C	Control-Region	0.074	0.076	0.074
G709A	rRNA	0.124	0.120	0.125
G951A	rRNA	0.023	0.028	0.021
T1189C	rRNA	0.031	0.032	0.031
T1243C	rRNA	0.047	0.036	0.051
C1341T	rRNA	0.035	0.044	0.031
G1719A	rRNA	0.047	0.040	0.050
A1811G	rRNA	0.077	0.076	0.077
G1888A	rRNA	0.049	0.048	0.050
G2098A	rRNA	0.029	0.020	0.033
C2259T	rRNA	0.025	0.032	0.022
A3480G	ND1	0.034	0.032	0.034
A3505G	ND1	0.047	0.036	0.051
C4059T	ND1	0.038	0.052	0.033
T4216C	ND1	0.124	0.129	0.122
T4452C	tRNA	0.041	0.032	0.045
A4529T	ND2	0.018	0.020	0.017
G4580A	ND2	0.056	0.036	0.065
T4646C	ND2	0.026	0.024	0.027
A4917G	ND2	0.052	0.052	0.051
G5046A	ND2	0.047	0.036	0.051
T5426C	ND2	0.016	0.020	0.014
G5460A	ND2	0.048	0.036	0.053
A5656G	Non-Coding	0.142	0.157	0.135
T5999C	CO1	0.026	0.024	0.027
A6047G	CO1	0.026	0.024	0.027
G6261A	CO1	0.014	0.024	0.010
T6776C	CO1	0.028	0.028	0.027
T7309C	CO1	0.041	0.032	0.045
A7385G	CO1	0.080	0.096	0.074
C7864T	CO2	0.037	0.024	0.043
G8251A	CO2	0.067	0.056	0.072
T8448C	ATP8	0.026	0.032	0.024
G8697A	ATP6	0.050	0.048	0.051
T8705C	ATP6	0.025	0.024	0.026

Table S4. Minor allele frequency and mitochondrial locus of 127 mtSNPs used in prediabetes-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (prediabetes)	MAF (controls)
C8818T	ATP6	0.018	0.020	0.017
T8843C	ATP6	0.014	0.020	0.012
C8898T	ATP6	0.014	0.020	0.012
G8994A	ATP6	0.047	0.036	0.051
G9055A	ATP6	0.041	0.036	0.043
A9066G	ATP6	0.041	0.032	0.045
A9093G	ATP6	0.020	0.020	0.021
C9391T	CO3	0.014	0.024	0.010
A9667G	CO3	0.018	0.024	0.015
T9698C	CO3	0.043	0.040	0.045
T9903C	CO3	0.017	0.020	0.015
T10034C	tRNA	0.018	0.020	0.017
T10238C	ND3	0.025	0.024	0.026
A10398G	ND3	0.136	0.141	0.134
T10463C	tRNA	0.055	0.052	0.057
A10550G	ND4L	0.034	0.032	0.034
T10915C	ND4	0.016	0.020	0.014
T10927C	ND4	0.080	0.096	0.074
A11251G	ND4	0.120	0.120	0.120
T11299C	ND4	0.035	0.036	0.034
C11332T	ND4	0.026	0.024	0.027
G11377A	ND4	0.025	0.028	0.024
C11674T	ND4	0.048	0.036	0.053
A11812G	ND4	0.041	0.036	0.043
G11914A	ND4	0.054	0.040	0.060
A11947G	ND4	0.047	0.036	0.051
T12414C	ND5	0.048	0.036	0.053
G12501A	ND5	0.026	0.028	0.026
A12612G	ND5	0.071	0.072	0.070
G12618A	ND5	0.139	0.157	0.132
G12630A	ND5	0.017	0.024	0.014
C12705T	ND5	0.098	0.084	0.104
G13368A	ND5	0.052	0.052	0.051
G13708A	ND5	0.077	0.076	0.077
A13780G	ND5	0.025	0.024	0.026
A13827G	ND5	0.032	0.048	0.026
G13928C	ND5	0.032	0.048	0.026
C14167T	ND6	0.034	0.032	0.034
A14233G	ND6	0.042	0.040	0.043
C14620T	ND6	0.026	0.024	0.027
T14798C	CYB	0.092	0.080	0.098
C14872T	CYB	0.025	0.032	0.022
G14905A	CYB	0.048	0.048	0.048
G15043A	CYB	0.035	0.036	0.034
A15218G	CYB	0.023	0.028	0.021
C15452A	CYB	0.120	0.120	0.120
A15607G	CYB	0.049	0.048	0.050
T15693C	CYB	0.026	0.024	0.027

Table S4. Minor allele frequency and mitochondrial locus of 127 mtSNPs used in prediabetes-specific analysis.

mtSNP	Mitochondrial locus	MAF (all)	MAF (prediabetes)	MAF (controls)
G15884C	CYB	0.047	0.036	0.051
C15904T	tRNA	0.058	0.040	0.065
A15924G	tRNA	0.025	0.024	0.026
G15928A	tRNA	0.049	0.048	0.050
C16069T	Control-Region	0.071	0.072	0.070
T16093C	Control-Region	0.078	0.084	0.075
C16114A	Control-Region	0.035	0.056	0.026
T16126C	Control-Region	0.116	0.120	0.115
T16144C	Control-Region	0.079	0.092	0.074
A16162G	Control-Region	0.059	0.072	0.053
C16223T	Control-Region	0.102	0.088	0.108
T16224C	Control-Region	0.041	0.044	0.039
G16255A	Control-Region	0.019	0.020	0.019
C16256T	Control-Region	0.068	0.088	0.060
C16261T	Control-Region	0.038	0.048	0.034
T16271C	Control-Region	0.017	0.024	0.014
C16278T	Control-Region	0.042	0.048	0.039
C16291T	Control-Region	0.023	0.028	0.021
C16292T	Control-Region	0.053	0.048	0.055
C16294T	Control-Region	0.085	0.104	0.077
C16296T	Control-Region	0.029	0.020	0.033
T16298C	Control-Region	0.068	0.056	0.074
T16304C	Control-Region	0.036	0.028	0.039
T16311C	Control-Region	0.112	0.124	0.106
A16316G	Control-Region	0.018	0.028	0.014
C16354T	Control-Region	0.023	0.028	0.021
T16356C	Control-Region	0.037	0.024	0.043
T16362C	Control-Region	0.030	0.024	0.033
G16391A	Control-Region	0.019	0.020	0.019
A16399G	Control-Region	0.028	0.032	0.026
G16526A	Control-Region	0.038	0.056	0.031