

Supplemental Data

Table S1. MtDNA variations in 49 patients with MELAS or MERRF.

Patient No (Phenotype)	Control region (1-576, 16024-16569)	Other region (577-16023)	Haplogroup
MT2-1 (MERRF)	73, 152, 235, 263, 309+C, 315+C, 522-523d, 16173, 16184, 16223, 16290, 16293, 16319, 16519	663, 750, 1438, 1736, 2706, 4248, 4769, 4824, 7028, 8344(H), 8794, 8860, 9650, 11719, 12705, 14766, 15326	A
MT7-1 (MELAS)	73, 150, 199, 263, 309+C, 315+C, 489, 16093, 16129, 16183C, 16189, 16223, 16239, 16297, 16298	750, 1438, 2706, 3243(H), 4048, 4071, 4164, 4769, 5351, 5460, 6455, 6680, 7028, 7684, 7853, 8701, 8860, 9540, 9824, 10345, 10398, 10400, 10873, 11365, 11719, 12405, 12705, 12811, 14766, 14783, 15043, 15301, 15326, 16093	M7b2
MT8-1 (MELAS)	73, 143, 152, 204, 263, 309+C, 315+C, 489, 16223, 16274, 16362, 16526	709, 750, 1438, 2706, 3426, 4769, 4833, 5108, 7028, 7621, 8701, 8854, 8860, 9540, 10398, 10873, 11719, 12705. 14569, 14766, 14783, 15043, 15301, 15326, 15746	G3a
MT11-1 (MELAS)	73, 150, 263, 309+CC, 315+C, 16172, 16223, 16257A, 16261, 16497	750, 961, 965+C ₃₋₄ (H), 1438, 2706, 4769, 5222, 5231, 5417, 7028, 8860, 11719, 12358, 12372, 12705, 14766, 15067, 15326	N9a2
MT13-1 (MERRF)	73, 152, 249d, 263, 315+C, 489, 16185, 16223, 16260, 16298	750, 1438, 2706, 3145, 4715, 4769, 6752, 7028, 7196A, 8188, 8584, 8701, 8860, 9090, 9380, 9540, 10398, 10400, 10873, 11719, 12705, 14668, 14766, 14783, 15067, 15301, 15326, 15355, 15487T, 15784	Z
MT21-1 (MELAS)	73, 146, 199, 263, 315+C, 489, 522-523d, 16223, 16294, 16295, 16519	750, 1438, 2706, 3882, 4071, 4769, 4850, 5442, 6053, 6455, 6620, 7028, 7961, 8701, 8860, 9540, 9824, 10398, 10400, 10873, 11665, 11719, 12091, 12705, 12804, 14755T, 14766, 14783, 15043, 15301, 15326	M7b'c
MT23-1 (MELAS)	73, 263, 315+C, 489, 573+C, 16223, 16278, 16362	750, 1438, 2706, 3010, 4343, 4769, 4883, 5178A, 7028, 8414, 8860, 9540, 10398, 10400, 10873, 11719, 12705, 13104, 14668, 14766, 14783, 15043, 15301, 15326, 15518	D4g1
MT25-1 (MERRF)	73, 150, 263, 309+CC, 315+C, 16129, 16189,	750, 1438, 2706, 2833, 4129, 4769, 5086, 5231, 5417, 7028, 8029, 8379, 8860, 9362, 11719,	N9

	16223, 16257A, 16261,	11914, 12354, 12358, 12372, 12612, 12705, 14766, 15326	
MT27-1 (MELAS)	73, 152, 235, 263, 309+C, 315+C, 16129, 16213, 16223, 16290, 16319	663, 750, 1438, 1736, 2706, 4248, 4769, 4824, 7028, 8563, 8794, 8860, 10691, 11536, 11719, 12705, 14053, 14766, 15326	A5c
MT29-1 (MELAS/ PEO)	73, 194, 263, 315+C, 489, 522-523d, 16172, 16362, 16519	750, 1382C, 1438, 2706, 3010, 4769, 4883, 5178A, 7028, 7630(H), 8020, 8414, 8701, 8860, 8964, 9296, 9540, 9824A, 10398, 10400, 10873, 11719, 12358, 12705, 14668, 14766, 14783, 15043, 15301, 15326	D4b2b
MT30-1 (MELAS)	73, 199, 202, 207, 263, 309+CC, 315+C, 499, 16136, 16182C, 16183C, 16189, 16217, 16284, 16357, 16519	750, 1438, 2706, 2831, 3243(H), 4117, 4769, 4820, 6023, 6413, 7028, 8206, 8271-8279d, 8440, 8860, 11719, 13590, 14766, 15236, 15326, 15535	B4b1a1
MT32-1 (MELAS)	73, 146, 194, 263, 309+C, 315+C, 489, 522-523d, 16223, 16362, 16519	750, 1382C, 1438, 2706, 3010, 4769, 4883, 5178A, 7028, 8020, 8414, 8701, 8860, 8964, 9296, 9540, 9824A, 10398, 10400, 10873, 11719, 12705, 14605, 14608, 14668, 14766, 14783, 15043, 15301, 15326	D4b2b1
MT33-1 (MELAS)	73, 146, 199, 263, 309+C, 315+C, 373, 489, 522-523d, 16223, 16295, 16519	750, 1438, 2706, 3243(H), 3882, 4071, 4452, 4769, 4850, 5442, 6455, 7028, 7960, 8701, 8860, 9540, 9824A, 10398, 10400, 10873, 11389, 11665, 11719, 11914, 12091, 12705, 14766, 14783, 15043, 15301, 15326, 15787, 15924	M7b'c
MT34-1 (MELAS)	73, 150, 263, 309+C, 315+C, 524+AC, 16172, 16223, 16257A, 16261	750, 961, 965+C ₁₋₃ (H), 1438, 2706, 3243(H), 3537, 4769, 5231, 5417, 7028, 8860, 11719, 12358, 12372, 12705, 14766, 15067, 15326	N9a2
MT35-1 (MERRF)	73, 150, 263, 315+C, 489, 522-523d, 16092, 16164(H), 16172, 16182C, 16183C, 16189, 16223, 16266, 16362	750, 752, 1107, 2706, 4769, 4883, 5178A, 5301, 7028, 8701, 8860, 9180, 9540, 10397, 10398, 10400, 10873, 11719, 11944, 12026, 12705, 14766, 14783, 15043, 15301, 15326	D5a2
MT36-1 (MELAS)	73, 195, 249d, 263, 315+C, 522-523d, 16092A, 16291, 16304	750, 1005, 1438, 1824, 2706, 3531, 3970, 4769, 4811, 6392, 6962, 7028, 7828, 8860, 10310, 10535, 10586, 11719, 12338, 13708, 13928C, 14766, 15218, 15326, 15849	F
MT40-1 (MELAS)	73, 199, 202, 207, 263, 309+CC, 315+C, 499,	750, 827, 1438, 2706, 2831, 4117, 4769, 4820, 6023, 6413, 7028, 8188, 8206, 8271-8279d, 8860,	B4b1a1

	16136, 16182C, 16183C, 16189, 16217, 16284, 16357, 16519	11719, 13590, 14766, 15236, 15326, 15535	
MT41-1 (MELAS)	73, 146, 152, 200, 235, 263, 309+CC, 315+C, 522-523d, 16223, 16290, 16319, 16362	663, 750, 1438, 1736, 2706, 3316, 4248, 4769, 4824, 7028, 8794, 8860, 10410, 10882, 11719, 12705, 13437, 14766, 15326	A
MT42-1 (MELAS)	73, 263, 309+C, 315+C, 489, 16192, 16223	750, 1438, 2706, 3010, 4769, 4883, 5178A, 7028, 8414, 8701, 8860, 9540, 10398, 10400, 10873, 11719, 12092A, 12705, 14668, 14766, 14783, 15043, 15301, 15326	D4
MT44-1 (MELAS/ PEO)	73, 263, 309+C, 315+C, 522- 523d, 16178, 16182C, 16183C, 16189, 16217, 16240, 16261,	750, 1438, 2706, 4769, 6863, 7028, 8271-8279d, 8860, 9717, 10398, 11719, 14410, 14766, 15326	B4a
MT46-1 (MELAS)	73, 151, 153, 263, 315+C, 522-523d, 16182C, 16183C, 16189, 16217, 16261, 16288, 16311, 16519	750, 1438, 2706, 4769, 5465, 7028, 8271-8279d, 8860, 9123, 9932A, 9977, 10238, 10915, 11719, 13143, 14766, 15326	B4a1b
MT49-1 (MERRF)	73, 150, 263, 309+C, 315+C, 16172, 16223, 16257A, 16261, 16355, 16497	750, 961, 965+C ₀₋₂ (H), 1438, 2706, 4769, 5231, 5417, 7028, 8860, 11719, 12358, 12372, 12705, 13224, 14766, 15067, 15326	N9a2ab
MT50-1 (MERRF)	73, 150, 263, 315+C, 489, 522-523d, 16092, 16164(H), 16172, 16182C, 16183C, 16189, 16223, 16266, 16362, 16519	750, 752, 1107, 2706, 3528, 4769, 4883, 5178A, 5301, 7028, 8701, 8802, 8860, 9180, 9540, 10397, 10398, 10400, 10873, 11719, 11944, 12026, 12705, 14766, 14783, 15043, 15301, 15326	D5a2
MT53-1 (MELAS)	73, 152, 189, 263, 309+C, 315+C, 489, 16184, 16223, 16298, 16319	750, 1438, 2706, 2835, 4670, 4715, 4769, 6179, 6671, 7028, 7196A, 8584, 8684, 8701, 8860, 9540, 10398, 10400, 10873, 11719, 12705, 13050, 13438, 14470, 14766, 14783, 15043, 15301, 15326, 15487T	M8a2b
MT55-1 (MERRF)	53, 143, 152, 263, 315+C, 489, 573+C ₃₋₅ (H), 16189, 16223, 16265C, 16274, 16362	709, 750, 1438, 2706, 4769, 4833, 5108, 6086, 6221, 7028, 7621, 8701, 8860, 9540, 10398, 10400, 10873, 11719, 12705, 14569, 14766, 14783, 15043, 15301, 15326, 15746	G3a
MT59-1 (MELAS)	73, 152, 263, 309+C, 315+C, 489, 16126, 16145, 16188, 16189, 16223, 16381	750, 1438, 2706, 3644, 4769, 5773, 6023, 6253, 6620, 7028, 8701, 8860, 9540, 10256, 10398, 10400, 10411, 10790, 10873, 11719, 12705,	M13a

		12954, 13135, 14766, 14783, 15043, 15301, 15326, 15924	
MT60-1 (MERRF)	73, 263, 294, 298, 309+C, 315+C, 489, 16223, 16362, 16519	750, 1438, 2294, 2706, 3010, 3398, 4769, 4883, 5178A, 5231, 7028, 7119, 8414, 8701, 8860, 9540, 10398, 10400, 10873, 11719, 12705, 13104, 14668, 14766, 14783, 15043, 15301, 15326	D4
MT61-1 (MELAS)	73, 150, 263, 309+C, 315+C, 524+AC, 16172, 16223, 16257A, 16261,	750, 961, 965+C ₁₋₃ (H), 1438, 2706, 3243(H), 3537, 4769, 5231, 5417, 7028, 8860, 11719, 12358, 12372, 12705, 14766, 15067, 15326	N9a2
MT62-1 (MELAS)	73, 263, 309+C, 489, 573+C ₃₋₅ (H), 16066, 16223, 16311	709, 750, 1438, 2706, 3172+C, 4035, 4140, 4769, 7028, 7250, 8701, 8793, 8856, 8860, 9540, 10398, 10400, 10648, 10873, 11719, 12549, 12705, 13152, 14502, 14766, 14783, 15040, 15043, 15071, 15218, 15301, 15326	M10b
MT63-1 (MERRF)	73, 263, 309+C, 315+C, 489, 16223, 16356, 16362	592, 709, 750, 1438, 2706, 4769, 4833, 4853, 5108, 5601, 7028, 8363(H), 8701, 8860, 8877, 9540, 10398, 10400, 10873, 11151, 11719, 12705, 13563, 14569, 14766, 14783, 15043, 15301, 15326	M
MT67-1 (MELAS)	73, 263, 315+C, 489, 16223, 16245, 16362	750, 1438, 2706, 3010, 4769, 4833, 5178A, 7028, 8383, 8414, 8419, 8701, 8860, 9431, 9540, 10398, 10400, 10873, 11719, 12705, 14668, 14766, 14783, 15043, 15301, 15326	D4c2
MT68-1 (MERRF)	73, 152, 207, 263, 309+C, 315+C, 489, 16129, 16223, 16362, 16519	750, 1438, 2706, 3010, 4769, 4883, 5178A, 7028, 7822, 8414, 8473, 8701, 8860, 9540, 10398, 10400, 10410, 10873, 11719, 12705, 14668, 14766, 14783, 14979, 15043, 15301, 15326	D4a1
MT69-1 (MERRF)	73, 150, 263, 315+C, 489, 16051, 16223, 16325, 16362, 16519	709, 750, 1438, 2706, 4769, 4793, 4833, 5108, 7028, 7867, 8200, 8701, 8860, 9132, 9540, 10398, 10400, 10873, 11719, 12705, 14569, 14766, 14783, 15043, 15301, 15323, 15326, 15497, 15860	G1a1
MT70-1 (MELAS)	73, 263, 309+C, 315+C, 489, 16223, 16224, 16234, 16316, 16362	750, 1041, 1438, 2706, 3394, 4491, 4769, 7028, 8701, 8860, 9242, 9540, 10398, 10400, 10873, 11719, 11854, 11963, 12705, 13731, 14308, 14766, 14783, 15043, 15301, 15326	M9a
MT71-1 (MELAS)	73, 195, 204, 263, 309+C, 315+C, 522-523d, 16140,	709, 750, 960+C, 1438, 1598, 1766, 2706, 4769, 7028, 8271-8279d, 8584, 8784, 8829, 8860, 9950,	B5b1

	16183C, 16189, 16243, 16278, 16324, 16519	10398, 11146, 11719, 12361, 13707, 14470, 14766, 15223, 15326, 15508, 15662, 15891, 15927	
MT72-1 (MELAS)	73, 194, 263, 309+C, 315+C, 489, 522-523d, 16111, 16186, 16223, 16362, 16519	709, 750, 1382C, 1438, 2706, 3010, 3243(H), 4769, 4883, 5178A, 5320G, 7028, 8020, 8414, 8701, 8860, 8964, 9296, 9540, 9824A, 10398, 10400, 10873, 11719, 12705, 14668, 14766, 14783, 15043, 15301, 15326, 15397	D4b2b
MT73-1 (MERRF)	73, 263, 315+C, 489, 16051, 16150, 16223, 16278, 16362	709, 750, 1438, 2706, 4769, 4833, 5108, 5601, 7028, 7600, 8701, 8860, 9377, 9540, 9575, 10398, 10400, 10873, 11017, 11719, 12705, 13563, 14200, 14569, 14766, 14783, 15043, 15301, 15326	G2a
MT75-1 (MELAS)	73, 263, 316+C, 489, 16129, 16223, 16362, 16519	750, 1438, 3010, 4538, 4769, 4883, 5178A, 6899, 7028, 7270, 8414, 8701, 8764, 8860, 9540, 10295, 10398, 10400, 10873, 11255, 11719, 12705, 14668, 14766, 14783, 15043, 15301, 15326	D4
MT77-1 (MELAS)	73, 263, 309+C, 315+C, 489, 16184, 16223, 16298, 16319,	750, 1438, 2706, 2835, 3290, 3394, 4715, 4769, 6179, 7028, 7196A, 8584, 8684, 8701, 8860, 9540, 10398, 10400, 10873, 11719, 12705, 12952, 14470, 14766, 14783, 15043, 15244, 15257, 15301, 15326, 15487T, 15565	M8a2
MT78-1 (MELAS)	73, 263, 309+C, 315+C, 489, 16223, 16293, 16362	750, 1438, 1664, 2706, 3010, 4769, 4883, 5178A, 7028, 8414, 8701, 8860, 9540, 10398, 10400, 10873, 11696, 11719, 12705, 14668, 14766, 14783, 15043, 15301, 15326	D4j
MT79-1 (MELAS)	73, 93, 210, 263, 309+C, 315+C, 522-523d, 16129, 16140, 16187, 16189, 16266, 16519	750, 1438, 2706, 3537, 4769, 7028, 8271-8279d, 8584, 8860, 9950, 9962, 10040, 10398, 11149, 11151, 11719, 12234, 14149, 14766, 15235, 15326, 15466	B5
MT80-1 (MERRF)	73, 103, 131, 263, 309+CC, 315+C, 522-523d, 16111, 16140, 16183C, 16189, 16234, 16243, 16463, 16519	709, 750, 1438, 1598, 2706, 4769, 4895, 5964, 7028, 8271-8279d, 8473, 8584, 8829, 8860, 9950, 10398, 11719, 12361, 14766, 15223, 15326, 15508, 15662, 15850, 15851, 15927	B5b2
MT81-1 (MELAS)	73, 146, 185, 189, 195, 196, 263, 309+C, 315+C, 513, 16172, 16182C, 16183C, 16189, 16217, 16223,	750, 827, 1438, 2706, 4769, 4926, 5277, 5460, 6026, 6614, 7028, 8271-8279d, 8860, 9921, 10750, 11719, 14133, 14766, 15326, 15535	B4b'd

	16295, 16519		
MT83-1 (MERRF)	73, 263, 315+C, 489, 522- 523d, 16223, 16362	750, 1438, 2706, 3010, 4769, 4883, 5178A, 6305, 6366, 7028, 8414, 8701, 8860, 9540, 10398, 10400, 10685, 10873, 11215, 11719, 12705, 14668, 14766, 14783, 15043, 15301, 15326, 15874	D4
MT84-1 (MELAS)	73, 152, 235, 263, 309+C, 315+C, 522-523d, 16093, 16223, 16290, 16319, 16362	663, 750, 1438, 1736, 2706, 4248, 4769, 4824, 7028, 8860, 11719, 12705, 14766, 15258, 15326	A4
MT87-1 (MELAS)	73, 146, 183, 263, 315+C, 489, 16189, 16223, 16362	750, 1009, 1438, 2706, 3010, 3336, 3644, 4655, 4769, 4883, 5048, 5178A, 7028, 8414, 8701, 8860, 9494, 9540, 10398, 10400, 10873, 11719, 12396, 12705, 13819, 14668, 14766, 14783, 15043, 15301, 15326	D4
MT90-1 (MELAS)	73, 200, 263, 309+CC, 315+C, 499, 16136, 16172, 16183C, 16189, 16217, 16218, 16428, 16519	750, 827, 1438, 1719, 2706, 4769, 4820, 7028, 7521, 8065, 8271-8279d, 8860, 9101G, 10586, 11009, 11239, 11719, 13590, 14587, 14766, 15326, 15535	B4b1
MT91-1 (MELAS)	73, 200, 204, 207, 263, 309+CC, 315+C, 489, 522- 523d, 16182C, 16183C, 16189, 16223, 16360, 16362	750, 752, 1107, 1438, 2706, 3702, 4769, 4883, 5178A, 5301, 7028, 8701, 8860, 9180, 9540, 10397, 10398, 10400, 10873, 11719, 11944, 12026, 12705, 12708, 13759, 14766, 14783, 14798, 15043, 15301, 15725	D
MT98-1 (MELAS)	73, 152, 263, 309+C, 315+C, 489, 16129, 16172, 16223, 16362, 16519	750, 1438, 2706, 3010, 3206, 4769, 4883, 5178A, 7028, 8414, 8473, 8701, 8860, 9540, 10398, 10400, 10410, 10873, 11383, 11719, 12705, 14668, 14766, 14783, 14979, 15043, 15301, 15326	D4a1

Nucleotide numbers were followed the rCRS ((NC_012920). +: insertion, d: deletion, (H): heteroplasmic variation.