Ambry Genetics®

Mitochondrial Mutation List

Exome testing includes optional mtDNA analysis for this defined list of established disease-causing variants.

HGVS	HUGO Gene Name	Disease
m.583G>A	MT-TF	MELAS / MM & EXIT
m.1494C>T	MT-RNR1	DEAF
m.1555A>G	MT-RNR1	DEAF
m.1606G>A	MT-TV	AMDF
m.1644G>A	MT-TV	HCM+MELAS
m.3243A>G	MT-TL1	MELAS / LS/DMDF / MIDD / SNHL / FSGS / CPEO
m.3251A>G	MT-TL1	MELAS
m.3252A>G	MT-TL1	MELAS
m.3256C>T	MT-TL1	MELAS
m.3260A>G	MT-TL1	MMC
m.3271T>C	MT-TL1	MELAS
m.3291T>C	MT-TL1	MELAS / Myopathy / Deafness+Cognitive Impairment
m.3302A>G	MT-TL1	MM
m.3303C>T	MT-TL1	MMC
m.3460G>A	MT-ND1	LHON
m.3635G>A	MT-ND1	LHON
m.3697G>A	MT-ND1	MELAS / LS / LDYT
m.3700G>A	MT-ND1	LHON
m.3733G>A	MT-ND1	LHON
m.3890G>A	MT-ND1	Progressive Encephalomyopathy / LS / Optic Atrophy
m.4171C>A	MT-ND1	LHON
m.4269A>G	MT-TI	FICP
m.4274T>C	MT-TI	CPEO / Motor Neuron Disease
m.4298G>A	MT-TI	CPEO / MS
m.4300A>G	MT-TI	MICM
m.4308G>A	MT-TI	CPEO
m.4332G>A	MT-TQ	Encephalopathy / MELAS
m.5537_5538insT	MT-TW	Leigh Syndrome
m.5650G>A	MT-TA	Myopathy
m.5703G>A	MT-TN	CPEO / MM
m.7222A>G	MT-CO1	Peripheral neuropathy, muscle weakness, ptosis, abnormal muscle pathology
m.7445A>G	MT-TS1	SNHL
m.7471dupC	MT-TS1	PEM / AMDF / Motor Neuron Disease-like
m.7497G>A	MT-TS1	MM / EXIT
m.7511T>C	MT-TS1	SNHL
m.8344A>G	MT-TK	MERRF
m.8356T>C	MT-TK	MERRF
m.8363G>A	MT-TK	MICM+DEAF / MERRF / Autism/ LS / Ataxia+Lipomas
m.8969G>A	MT-ATP6	Mitochondrial Myopathy, Lactic Acidosis

m.8993T>C	MT-ATP6	NARP / Leigh Disease / MILS / other
m.8993T>G	MT-ATP6	NARP / Leigh Disease / MILS / other
m.9176T>C	MT-ATP6	FBSN / Leigh Disease
m.9176T>G	MT-ATP6	Leigh Disease / Spastic Paraplegia
m.9185T>C	MT-ATP6	Leigh Disease / Ataxia / NARP-like Disease
m.10010T>C	MT-TG	PEM
m.10158T>C	MT-ND3	Leigh Disease
m.10191 T>C	MT-ND3	Leigh Disease / Leigh-like Disease / ESOC
m.10197G>A	MT-ND3	Leigh Disease / Dystonia / Stroke / LDYT
m.10663T>C	MT-ND4L	LHON
m.11777C>A	MT-ND4	Leigh Disease
m.11778G>A	MT-ND4	LHON, Progressive Dystonia
m.12147G>A	MT-TH	MERRF-MELAS / Enchephalopathy
m.12258C>A	MT-TS2	DMDF / RP+SNHL
m.12315G>A	MT-TL2	CPEO/KSS
m.12320A>G	MT-TL2	Myopathy
m.12706T>C	MT-ND5	Leigh Disease
m.13513G>A	MT-ND5	Leigh Disease / MELAS / LHON-MELAS Overlap Syndrome
m.13514A>G	MT-ND5	Leigh Disease / MELAS
m.14459G>A	MT-ND6	LDYT / Leigh Disease
m.14482C>G	MT-ND6	LHON
m.14484T>C	MT-ND6	LHON
m.14487T>C	MT-ND6	Dystonia / Leigh Disease /Ataxia
m.14495A>G	MT-ND6	LHON
m.14568C<T	MT-ND6	LHON
m.14674T>C	MT-TE	Reversible COX Deficiency Myopathy
m.14709T>C	MT-TE	MM+DMDF / Encephalomyopathy
m.14849T>C	MT-CYB	EXIT / Septo-Optic Dysplasia
m.15579A>G	MT-CYB	Multisystem Disorder, EXIT

One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

MKT-SPEC-FLYR-50084-EN v1 07.29.24