



## MITOCHONDRIAL TESTS

Category	Test	Disease	Test Specification	Price in €
Mitochondrial Tests	MTTS2 (TRANSFER RNA, MITOCHONDRIAL, SERINE, 2)	ATAXIA, CATARACT AND DIABETES MELLITUS » RETINITIS PIGMENTOSA-DEAFNESS SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTV (TRANSFER RNA, MITOCHONDRIAL, VALINE)	ATAXIA, PROGRESSIVE SEIZURES, MENTAL DETERIORATION, AND HEARING LOSS	See Entire mitochondrial genome	
Mitochondrial Tests	C3254G	CARDIOMYOPATHY		250
Mitochondrial Tests	A3260G	CARDIOMYOPATHY		250
Mitochondrial Tests	C3303T	CARDIOMYOPATHY		250
Mitochondrial Tests	MTRNR1 (RIBOSOMAL RNA, MITOCHONDRIAL, 12S)	CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTG (TRANSFER RNA, MITOCHONDRIAL, GLYCINE)	CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTH (TRANSFER RNA, MITOCHONDRIAL, HISTIDINE)	CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTI (TRANSFER RNA, MITOCHONDRIAL, ISOLEUCINE)	CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL2 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 2)	CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	CARDIOMYOPATHY AND DEAFNESS	See Entire mitochondrial genome	
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	CARDIOMYOPATHY AND DEAFNESS	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	CARDIOMYOPATHY, INFANTILE HISTIOCYTOID » CARDIOMYOPATHY, INFANTILE XANTHOMATOUS » CARDIOMYOPATHY, FOCAL LIPID » CARDIOMYOPATHY, ONCOCYTIC	See Entire mitochondrial genome	
Mitochondrial Tests	MTRNR2 (RIBOSOMAL RNA, MITOCHONDRIAL, 16S)	CHLORAMPHENICOL RESISTANCE	See Entire mitochondrial genome	
Mitochondrial Tests	MTND1 (COMPLEX 1, SUBUNIT ND1, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND1NADH DEHYDROGENASE, SUBUNIT 1)	COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » NADH:Q(1) OXIDOREDUCTASE DEFICIENCY » NADH-COENZYME Q REDUCTASE DEFICIENCY » MITOCHONDRIAL NADH DEHYDROGENASE COMPONENT OF COMPLEX 1 DEFICIENCY OF	See Entire mitochondrial genome	
Mitochondrial Tests	MTND2 (COMPLEX 1, SUBUNIT ND2, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND2NADH DEHYDROGENASE, SUBUNIT 2)	COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » NADH:Q(1) OXIDOREDUCTASE DEFICIENCY » NADH-COENZYME Q REDUCTASE DEFICIENCY » MITOCHONDRIAL NADH DEHYDROGENASE COMPONENT OF COMPLEX 1 DEFICIENCY OF	See Entire mitochondrial genome	

Mitochondrial Tests	MTND3 (COMPLEX 1, SUBUNIT ND3, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND3NADH DEHYDROGENASE, SUBUNIT 3)	COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » NADH:Q(1) OXIDOREDUCTASE DEFICIENCY » NADH-COENZYME Q REDUCTASE DEFICIENCY » MITOCHONDRIAL NADH DEHYDROGENASE COMPONENT OF COMPLEX 1 DEFICIENCY OF	See Entire mitochondrial genome	
Mitochondrial Tests	MTND4 (COMPLEX 1, SUBUNIT ND4, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND4NADH DEHYDROGENASE, SUBUNIT 4)	COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » NADH:Q(1) OXIDOREDUCTASE DEFICIENCY » NADH-COENZYME Q REDUCTASE DEFICIENCY » MITOCHONDRIAL NADH DEHYDROGENASE COMPONENT OF COMPLEX 1 DEFICIENCY OF	See Entire mitochondrial genome	
Mitochondrial Tests	MTND5 (COMPLEX 1, SUBUNIT ND5, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND5NADH DEHYDROGENASE, SUBUNIT 5)	COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » NADH:Q(1) OXIDOREDUCTASE DEFICIENCY » NADH-COENZYME Q REDUCTASE DEFICIENCY » MITOCHONDRIAL NADH DEHYDROGENASE COMPONENT OF COMPLEX 1 DEFICIENCY OF	See Entire mitochondrial genome	
Mitochondrial Tests	MTND6 (COMPLEX 1, SUBUNIT ND6, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND6NADH DEHYDROGENASE, SUBUNIT 6)	COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » NADH:Q(1) OXIDOREDUCTASE DEFICIENCY » NADH-COENZYME Q REDUCTASE DEFICIENCY » MITOCHONDRIAL NADH DEHYDROGENASE COMPONENT OF COMPLEX 1 DEFICIENCY OF	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	COMPLEX 3, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO1 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 1, CYTOCHROME c OXIDASE 1; COX1)	COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO2 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 2, CYTOCHROME c OXIDASE 2; COX2)	COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO3 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 3, CYTOCHROME c OXIDASE 3; COX3)	COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF » CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY	See Entire mitochondrial genome	
Mitochondrial Tests	4977 bp Deletion	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA)		300
Mitochondrial Tests	A3243G	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA)		250
Mitochondrial Tests	T3250C	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA)		250

Mitochondrial Tests	G3316A	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	T4274C	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	T4285C	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	G4298A	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	G4309A	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	T5628C	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	A5692G	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	G5703A	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	G8342A	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	A12308G	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	T12311C	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	G12315A	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)		250
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTY (TRANSFER RNA, MITOCHONDRIAL, TYROSINE)	CPEO (CHRONIC PROGRESSIVE EXTERNAL OPTHALMOPLÉGIA)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTS1 (TRANSFER RNA, MITOCHONDRIAL, SERINE, 1)	CYTOCHROME c OXIDASE DEFICIENCY	See Entire mitochondrial genome	
Mitochondrial Tests	A1555G	DEAFNESS		250
Mitochondrial Tests	7472insC	DEAFNESS		250
Mitochondrial Tests	T7511C	DEAFNESS		250
Mitochondrial Tests	A7445G	DEAFNESS		250
Mitochondrial Tests	961delT/insC	DEAFNESS		250
Mitochondrial Tests	T961G	DEAFNESS		250
Mitochondrial Tests	T1095C	DEAFNESS		250
Mitochondrial Tests	C1494T	DEAFNESS		250
Mitochondrial Tests	A827G	DEAFNESS		250
Mitochondrial Tests	T1005C	DEAFNESS		250
Mitochondrial Tests	T1291C	DEAFNESS		250
Mitochondrial Tests	T1243C	DEAFNESS		250
Mitochondrial Tests	A1116G	DEAFNESS		250

Mitochondrial Tests	All 10 mutations in the MTRNR1 and MTTT1 genes associated with non-syndromic hearing loss : A1555G, 961delT/insC, T961G, T1095C, C1494T, A827G, T1005C, T1291C, T1243C and A1116G	DEAFNESS		600
Mitochondrial Tests	MTRNR1 (RIBOSOMAL RNA, MITOCHONDRIAL, 12S)	DEAFNESS	See Entire mitochondrial genome	
Mitochondrial Tests	MTRNR1 (RIBOSOMAL RNA, MITOCHONDRIAL, 12S)	DEAFNESS, AMINOGLYCOSIDE-INDUCED	See Entire mitochondrial genome	
Mitochondrial Tests	MTTO (TRANSFER RNA, MITOCHONDRIAL, GLUTAMINE)	DEAFNESS AND MIGRAINE	See Entire mitochondrial genome	
Mitochondrial Tests	A3243G	DEAFNESS AND DIABETES » MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	DEAFNESS AND DIABETES	See Entire mitochondrial genome	
Mitochondrial Tests	MTND1 (COMPLEX 1, SUBUNIT ND1, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND1NADH DEHYDROGENASE, SUBUNIT 1)	DYSTONIA, ADULT-ONSET	See Entire mitochondrial genome	
Mitochondrial Tests	MTND4 (COMPLEX 1, SUBUNIT ND4, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND4NADH DEHYDROGENASE, SUBUNIT 4)	DYSTONIA, FAMILIAL, WITH VISUAL FAILURE AND STRIATAL LUCENCIES » LEBER OPTIC ATROPHY AND DYSTONIA » MARSDEN SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTND6 (COMPLEX 1, SUBUNIT ND6, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND6NADH DEHYDROGENASE, SUBUNIT 6)	DYSTONIA, FAMILIAL, WITH VISUAL FAILURE AND STRIATAL LUCENCIES » LEBER OPTIC ATROPHY AND DYSTONIA » MARSDEN SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	ENCEPHALOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	ENCEPHALOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL2 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 2)	ENCEPHALOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTW (TRANSFER RNA, MITOCHONDRIAL, TRYPTOPHAN)	ENCEPHALOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTI (TRANSFER RNA, MITOCHONDRIAL, ISOLEUCINE)	ENCEPHALOPATHY, FAMILIAL PROGRESSIVE NECROTIZING	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	EXERCISE INTOLERANCE	See Entire mitochondrial genome	
Mitochondrial Tests	MTTG (TRANSFER RNA, MITOCHONDRIAL, GLYCINE)	EXERCISE INTOLERANCE	See Entire mitochondrial genome	
Mitochondrial Tests	MTTY (TRANSFER RNA, MITOCHONDRIAL, TYROSINE)	EXERCISE INTOLERANCE, CARDIOMYOPATHY, AND SEPTOOPTIC DYSPLASIA	See Entire mitochondrial genome	
Mitochondrial Tests	MTTY (TRANSFER RNA, MITOCHONDRIAL, TYROSINE)	FOCAL SEGMENTAL GLOMERULOSCLEROSIS AND DILATED CARDIOMYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTI (TRANSFER RNA, MITOCHONDRIAL, ISOLEUCINE)	HYPOMAGNESEMIA, HYPERTENSION, AND HYPERCHOLESTEROLEMIA	See Entire mitochondrial genome	
Mitochondrial Tests	4977 bp Deletion	KEARNS-SAYRE SYNDROME (KSS)		300
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	KEARNS-SAYRE SYNDROME (KSS)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTS1 (TRANSFER RNA, MITOCHONDRIAL, SERINE, 1)	KERATODERMA, PALMOPLANTAR, WITH DEAFNESS » DEAFNESS. NONSYNDROMIC SENSORINEURAL	See Entire mitochondrial genome	

Mitochondrial Tests	MTATP6 (ATP SYNTHASE 6, COMPLEX 5, ATP SYNTHASE, SUBUNIT ATPase 6, ATP6)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO1 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 1, CYTOCHROME c OXIDASE 1: COX1)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO3 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 3, CYTOCHROME c OXIDASE 3: COX3)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND1 (COMPLEX 1, SUBUNIT ND1, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND1NADH DEHYDROGENASE, SUBUNIT 1)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND2 (COMPLEX 1, SUBUNIT ND2, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND2NADH DEHYDROGENASE, SUBUNIT 2)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND4 (COMPLEX 1, SUBUNIT ND4, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND4NADH DEHYDROGENASE, SUBUNIT 4)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND6 (COMPLEX 1, SUBUNIT ND6, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND6NADH DEHYDROGENASE, SUBUNIT 6)	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)	See Entire mitochondrial genome	
Mitochondrial Tests	G3460A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	G11778A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	C3275A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	G3316A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	T3394C	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	T4216C	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	G7444A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	T9101C	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	G13708A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	T14484C	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	G14459A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	G15257A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		250
Mitochondrial Tests	3 common mutations accounting for ~90% of all LHON mutations: G11778A, T14484C, G3460A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		500
Mitochondrial Tests	12 common mutations accounting for > 95% of all LHON mutations: G11778A, T14484C, G3460A, C3275A, G3316A, T3394C, T4216C, G7444A, T9101C, G13708A, G14459A, G15257A	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)		650
Mitochondrial Tests	MTATP6 (ATP SYNTHASE 6, COMPLEX 5, ATP SYNTHASE, SUBUNIT ATPase 6, ATP6)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO3 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 3, CYTOCHROME c OXIDASE 3: COX3)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTND3 (COMPLEX 1, SUBUNIT ND3, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND3NADH DEHYDROGENASE, SUBUNIT 3)	LEIGH SYNDROME	See Entire mitochondrial genome	

Mitochondrial Tests	MTND5 (COMPLEX 1, SUBUNIT ND5, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND5NADH DEHYDROGENASE, SUBUNIT 5)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTND6 (COMPLEX 1, SUBUNIT ND6, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND6NADH DEHYDROGENASE, SUBUNIT 6)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTV (TRANSFER RNA, MITOCHONDRIAL, VALINE)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTW (TRANSFER RNA, MITOCHONDRIAL, TRYPTOPHAN)	LEIGH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	T8993C	LEIGH SYNDROME		250
Mitochondrial Tests	T8993G	LEIGH SYNDROME		250
Mitochondrial Tests	C1177A	LEIGH SYNDROME		250
Mitochondrial Tests	C1624T	LEIGH SYNDROME		250
Mitochondrial Tests	T9176C	LEIGH SYNDROME		250
Mitochondrial Tests	T9176G	LEIGH SYNDROME		250
Mitochondrial Tests	9537insC	LEIGH SYNDROME		250
Mitochondrial Tests	A13084T	LEIGH SYNDROME		250
Mitochondrial Tests	G13513A	LEIGH SYNDROME		250
Mitochondrial Tests	G13513A	LEIGH SYNDROME		250
Mitochondrial Tests	A3271G	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	T8356C	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	A13084T	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	G13513A	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	C3093G	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	A3252G	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	C3256T	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	A3260G	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	T3291C	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250

Mitochondrial Tests	T3308C	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	A13514G	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		250
Mitochondrial Tests	9 common mutations representing > 90% of all MELAS mutations: A3243G, T3271C, C3093G, A3252G, C3256T, A3260G, T3291C, T3308C, A13514G	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)		600
Mitochondrial Tests	MTND1 (COMPLEX 1, SUBUNIT ND1, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND1NADH DEHYDROGENASE, SUBUNIT 1)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND5 (COMPLEX 1, SUBUNIT ND5, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND5NADH DEHYDROGENASE, SUBUNIT 5)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND6 (COMPLEX 1, SUBUNIT ND6, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND6NADH DEHYDROGENASE, SUBUNIT 6)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTF (TRANSFER RNA, MITOCHONDRIAL, PHENYLALANINE)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTH (TRANSFER RNA, MITOCHONDRIAL, HISTIDINE)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTS1 (TRANSFER RNA, MITOCHONDRIAL, SERINE, 1)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTQ (TRANSFER RNA, MITOCHONDRIAL, GLUTAMINE)	MELAS SYNDROME (MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES)	See Entire mitochondrial genome	
Mitochondrial Tests	MTND5 (COMPLEX 1, SUBUNIT ND5, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND5NADH DEHYDROGENASE, SUBUNIT 5)	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTF (TRANSFER RNA, MITOCHONDRIAL, PHENYLALANINE)	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTH (TRANSFER RNA, MITOCHONDRIAL, HISTIDINE)	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)	See Entire mitochondrial genome	

Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)	See Entire mitochondrial genome	
Mitochondrial Tests	MTTS1 (TRANSFER RNA, MITOCHONDRIAL, SERINE, 1)	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)	See Entire mitochondrial genome	
Mitochondrial Tests	T8356C	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)		250
Mitochondrial Tests	A8344G	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)		250
Mitochondrial Tests	A8296G	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)		250
Mitochondrial Tests	G8363A	MERRF SYNDROME (MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS)		250
Mitochondrial Tests	MTRNR1, MTRNR2, MTND1, MTND2, MTND3, MTND4L, MTND4, MTND5, MTND6, MTCO1, MTCO2, MTCO3, MTATP8, MTATP6, MTCYB, MTTF, MTTV, MTTL1, MTTI, MTTQ, MTTM, MTTW, MTTA, MTTN, MTTT, MTTY, MTTS1, MTTD, MTK, MTTG, MTR, MTH, MTTS2, MTTL2, MTF, MTF, MTF	MITOCHONDRIAL DISEASE	Entire mitochondrial genome (16569 bp) including all the 37 genes (2 rRNA genes, 22 tRNA genes, and 13 protein-coding genes) and non-coding regions	750
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	MULTISYSTEM DISORDER	See Entire mitochondrial genome	
Mitochondrial Tests	MTTI (TRANSFER RNA, MITOCHONDRIAL, ISOLEUCINE)	MULTISYSTEM DISORDER	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	MYELODYSPLASTIC SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO1 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 1, CYTOCHROME c OXIDASE 1: COX1)	MYOGLOBINURIA, RECURRENT	See Entire mitochondrial genome	
Mitochondrial Tests	MTTK (TRANSFER RNA, MITOCHONDRIAL, LYSINE)	MYONEURAL GASTROINTESTINAL ENCEPHALOPATHY SYNDROME » MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME, MNGIE SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTC (TRANSFER RNA, MITOCHONDRIAL, CYSTEINE)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTD (TRANSFER RNA, MITOCHONDRIAL, ASPARTIC ACID)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL2 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 2)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTP (TRANSFER RNA, MITOCHONDRIAL, PROLINE)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTQ (TRANSFER RNA, MITOCHONDRIAL, GLUTAMINE)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTR (TRANSFER RNA, MITOCHONDRIAL, ARGININE)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTW (TRANSFER RNA, MITOCHONDRIAL, TRYPTOPHAN)	MYOPATHY	See Entire mitochondrial genome	
Mitochondrial Tests	T618C	MYOPATHY		250
Mitochondrial Tests	A3243T	MYOPATHY		250
Mitochondrial Tests	T3250C	MYOPATHY		250
Mitochondrial Tests	A3251G	MYOPATHY		250
Mitochondrial Tests	C3254G	MYOPATHY		250
Mitochondrial Tests	A3259G	MYOPATHY		250
Mitochondrial Tests	A3261G	MYOPATHY		250
Mitochondrial Tests	A3266G	MYOPATHY		250
Mitochondrial Tests	A3268G	MYOPATHY		250
Mitochondrial Tests	A3288G	MYOPATHY		250
Mitochondrial Tests	A3302G	MYOPATHY		250



Mitochondrial Tests	4370insC	MYOPATHY		250
Mitochondrial Tests	T4409C	MYOPATHY		250
Mitochondrial Tests	G4450A	MYOPATHY		250
Mitochondrial Tests	G5521A	MYOPATHY		250
Mitochondrial Tests	A12320G	MYOPATHY		250
Mitochondrial Tests	G15762A	MYOPATHY		250
Mitochondrial Tests	C15990T	MYOPATHY		250
Mitochondrial Tests	MTTA (TRANSFER RNA, MITOCHONDRIAL, ALANINE)	MYOPATHY, MYOTONIC DYSTROPHY-LIKE	See Entire mitochondrial genome	
Mitochondrial Tests	MTTE (TRANSFER RNA, MITOCHONDRIAL, LUTAMIC ACID)	MYOPATHY, WITH DIABETES MELLITUS » MITOCHONDRIAL MYOPATHY, LIPID TYPE	See Entire mitochondrial genome	
Mitochondrial Tests	MTATP6 (ATP SYNTHASE 6, COMPLEX 5, ATP SYNTHASE, SUBUNIT ATPase 6, ATP6)	NARP SYNDROME (NEUROPATHY WITH ATAXIA AND RETINITIS PIGMENTOSA)	See Entire mitochondrial genome	
Mitochondrial Tests	T8993C	NARP SYNDROME (NEUROPATHY WITH ATAXIA AND RETINITIS PIGMENTOSA)		250
Mitochondrial Tests	T8993G	NARP SYNDROME (NEUROPATHY WITH ATAXIA AND RETINITIS PIGMENTOSA)		250
Mitochondrial Tests	MTTV (TRANSFER RNA, MITOCHONDRIAL, VALINE)	NEONATAL DEATH	See Entire mitochondrial genome	
Mitochondrial Tests	MTTW (TRANSFER RNA, MITOCHONDRIAL, TRYPTOPHAN)	NEUROGASTROINTESTINAL SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	NEUROPSYCHIATRIC DISORDER AND EARLY-ONSET CATARACT	See Entire mitochondrial genome	
Mitochondrial Tests	MTATP8 (ATP SYNTHASE 8, COMPLEX 5, ATP SYNTHASE, SUBUNIT ATPase 8, ATP8)	NO DISEASE	See Entire mitochondrial genome	
Mitochondrial Tests	MTTM (TRANSFER RNA, MITOCHONDRIAL, METHIONINE)	NO DISEASE	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	OBESITY	See Entire mitochondrial genome	
Mitochondrial Tests	MTTN (TRANSFER RNA, MITOCHONDRIAL, ASPARAGINE)	OPHTHALMOPLEGIA	See Entire mitochondrial genome	
Mitochondrial Tests	MTND1 (COMPLEX 1, SUBUNIT ND1, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND1NADH DEHYDROGENASE, SUBUNIT 1)	PARKINSON DISEASE	See Entire mitochondrial genome	
Mitochondrial Tests	MTTP (TRANSFER RNA, MITOCHONDRIAL, PROLINE)	PARKINSON DISEASE	See Entire mitochondrial genome	
Mitochondrial Tests	MTTT (TRANSFER RNA, MITOCHONDRIAL, THREONINE)	PARKINSON DISEASE	See Entire mitochondrial genome	
Mitochondrial Tests	MTCYB (CYTOCHROME b OF COMPLEX 3, COMPLEX 3, CYTOCHROME b SUBUNITUBIQUINONE-CYTOCHROME c OXIDOREDUCTASE, CYTOCHROME b SUBUNIT)	PARKINSONISM / MELAS OVERLAP SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTATP6 (ATP SYNTHASE 6, COMPLEX 5, ATP SYNTHASE, SUBUNIT ATPase 6, ATP6)	SEIZURES AND LACTIC ACIDOSIS	See Entire mitochondrial genome	
Mitochondrial Tests	MTCO1 (COMPLEX 4, CYTOCHROME c OXIDASE SUBUNIT 1, CYTOCHROME c OXIDASE 1: COX1)	SIDEROBLASTIC ANEMIA, ACQUIRED IDIOPATHIC	See Entire mitochondrial genome	
Mitochondrial Tests	MTATP6 (ATP SYNTHASE 6, COMPLEX 5, ATP SYNTHASE, SUBUNIT ATPase 6, ATP6)	STRIATONIGRAL DEGENERATION, INFANTILE » BILATERAL STRIATAL NECROSIS, INFANTILE, MITOCHONDRIAL	See Entire mitochondrial genome	
Mitochondrial Tests	MTND1 (COMPLEX 1, SUBUNIT ND1, NADH-UBIQUINONE OXIDOREDUCTASE, SUBUNIT ND1NADH DEHYDROGENASE, SUBUNIT 1)	SUDDEN INFANT DEATH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTG (TRANSFER RNA, MITOCHONDRIAL, GLYCINE)	SUDDEN INFANT DEATH SYNDROME	See Entire mitochondrial genome	
Mitochondrial Tests	MTTL1 (TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1)	SUDDEN INFANT DEATH SYNDROME	See Entire mitochondrial genome	

Mitochondrial Tests	9 Mutations: 4977 bp Deletion, G3460A, G11778A, T8993C, T8993G, A3271G, A3243G, T8356C, A8344G	Screening for 9 Mitochondrial Mutations: CPEO, KEARNS-SAYRE SYNDROME, LHON, LEIGH SYNDROME, MELAS SYNDROME, MERRE SYNDROME, NARP SYNDROME	650
Mitochondrial Tests	Deletion Testing MTDNA	VARIOUS DISEASES	320