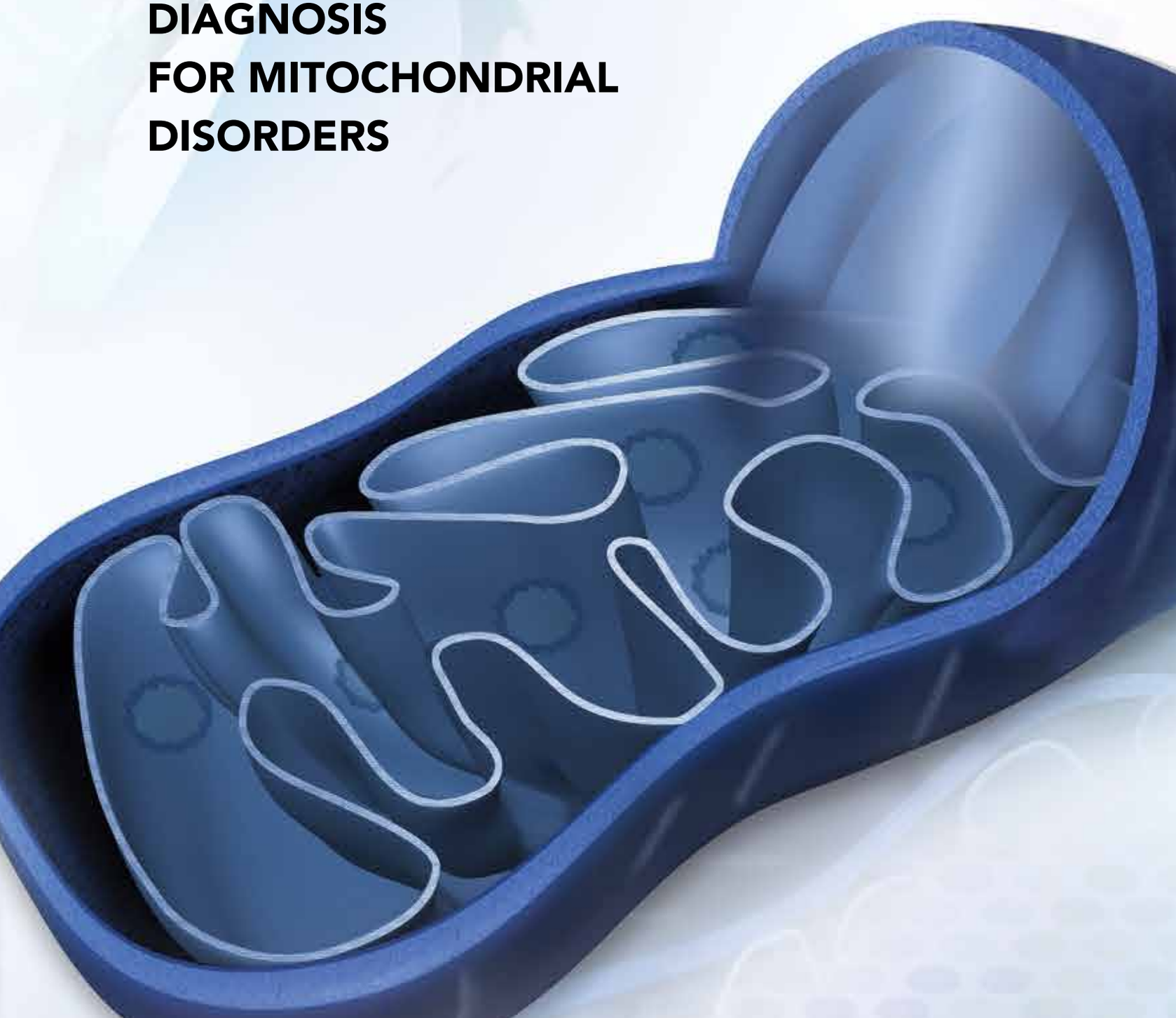
Baylor Miraca
Genetics Laboratories

COMPREHENSIVE DIAGNOSIS FOR MITOCHONDRIAL DISORDERS



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MITOCHONDRIAL DISORDERS NGS PANELS

Panel Name	Test code	Gene Name
Mitome200-Nuclear Panel	2086 (164 nuclear genes)	AARS2, ACACA, ACACB, ACAD9, ACADL, ACADM, ACADVL, ACAT1, ADCK3, AGL, ARG1, ATP5A1, ATP5E, ATPAF2, AUH, BCS1L, BTD, C10orf2, C12orf65, COQ2, COQ9, COX15, COX4I1, COX4I2, COX6B1, COX7A1, CPS1, CPT1A, CPT1B, CPT2, DARS2, DGUOK, DLAT, DLD, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FOXRED1, G6PC, GAA, GBE1, GFM1, GYS1, GYS2, HADHA, HADHB, HARS2, HLCS, ISCU, IVD, KARS, LARS2, LMBRD1, LPIN1, LRPPRC, MARS2, MCCC1, MCCC2, MGME1, MMAA, MMAB, MMACHC, MMADHC, MPI, MPV17, MRPL40, MRPL44, MRPS16, MRPS18A, MRPS2, MRPS22, MRRF, MTFMT, MTRR, MUT, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA13, NDUFA2, NDUFA7, NDUFA8, NDUFAF2, NDUFAF5, NDUFB6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV3, NUBPL, OPA1, OPA3, OTC, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PMM2, POLG, POLG2, PUS1, PYGL, PYGM, RARS2, RRM2B, SARS2, SCO1, SCO2, SDHAF1, SDHAF2, SDHB, SDHC, SLC22A5, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A3, SLC25A4, SLC37A4, SUCLA2, SUCLG1, SUCLG2, SURF1, TACO1, TAZ, TCN2, TFAM, TFB1M, TIMM8A, TK2, TMEM70, TOMM20, TRMU, TSFM, TUFM, TYMP, UQCRB, UQCRQ, YARS2
Leigh Syndrome-Nuclear Panel	20601 (91 nuclear genes) 20600 (plus mitochondrial genome)	AARS2, ACAD9, ADCK3, APTX, ATP5E , ATPAF2 (ATP12), BCS1L, C10ORF2, COQ2, COQ9 , COX10, COX15, COX4I1, COX4I2, COX6B1, COX7A1, DARS2, DGUOK, DLAT, DLD, ETFDH, ETHE1, FASTKD2, FH, FOXRED1, GFER, GFM1, LARS2, LMBRD1, LRPPRC, MPV17, MRPS16, MTFMT, NDUFA1, NDUFA10, NDUFA11, NDUFA13, NDUFA2, NDUFA7, NDUFA8, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFB6, NDUFS1, NDUFS2 , NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV3, NUBPL , PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, POLG, RARS2, RRM2B, SCO1 , SCO2 , SDHA, SDHAF1, SDHAF2, SDHB, SDHC , SDHD, SUCLA2, SUCLG1, SUCLG2, SURF1, TACO1, TK2, TMEM70, TOMM20, TRMU, TSFM, TTC19, TUFM, TUSC3, TYMP, UQCRB, UQCRQ, YARS2
CoQ10 Deficiency Panel	2100 (5 genes)	ADCK3 (COQ8/CABC1), COQ2, COQ9, PDSS1, PDSS2

Panel Name	Test code	Gene Name
mtDNA Depletion/ Integrity Panel	2130 (20 genes)	APTX, C10orf2, DGUOK, DNA2, FBXL4, GFER, MGME1, MPV17, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A4, SPG7, SUCLA2, SUCLG1, SUCLG2, TK2, TYMP
Progressive External Ophthalmoplegia (PEO)	2140 (6 genes)	C10orf2, OPA1, POLG, POLG2, RRM2B, SLC25A4
Mitochondrial Respiratory Chain Complex I – V Panel	2180 (50 genes)	All genes as listed below for Mitochondrial Respiratory Chain Complex I - V tests
Mitochondrial Respiratory Chain Complex I Panel	2155 (25 genes)	FOXRED1, NDUFA1, NDUFA2, NDUFA7, NDUFA8, NDUFA10, NDUFA11, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5 (C20ORF7), NDUFB6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV3, NUBPL
Mitochondrial Respiratory Chain Complex II Panel	2160 (6 genes)	SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD
Mitochondrial Respiratory Chain Complex III Panel	2165 (4 genes)	BCS1L, TTC19, UQCRB, UQCRQ
Mitochondrial Respiratory Chain Complex IV Panel	2170 (12 genes)	COX10, COX15, COX4I1, COX4I2, COX6B1, COX7A1, FASTKD2, LRPPRC, SCO1, SCO2, SURF1, TACO1
Mitochondrial Respiratory Chain Complex V Panel	2175 (3 genes)	ATP5E, ATPAF2 (ATP12), TMEM70
PDH & Mitochondrial Respiratory Chain Complex V Panel	2185 (9 genes)	ATP5E, ATPAF2 (ATP12), DLAT, DLD, PDHA1, PDHB, PDHX, PDP1, TMEM70

**Single gene Sanger Sequencing and Targeted Deletion/Duplication analysis are also available.*

SAMPLE & SHIPPING INFORMATION

TECHNICAL INFORMATION	
Methodology:	Targeted Capture followed by Massively Parallel Sequencing
Test Type:	Next Generation Sequencing
SAMPLE & SHIPPING INFORMATION	
Specimen Type:	Blood
Requirements:	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc (Adults/ Children).
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze. Sample must arrive within 48 hrs.
Specimen Type:	Purified DNA
Requirements:	Send 5-10 ug of purified DNA (minimal concentration of 50ng/uL; A260/A280 of ~1.7).
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Turn Around Time: 6-10 weeks	
SHIPPING ADDRESS	
2450 Holcombe, Grand Blvd. Receiving Dock, Houston, TX 77021-2024	

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METABOLIC DISORDERS

Panel Name	Test code	Number of Genes	Gene name*
Cholestasis/PFIC Panel	2105	7	ABCB4, ABCB11, AKR1D1, ATP8B, JAG1, SLC25A13, SERPINA1
Cobalamin Metabolism Disorders Panel	2120	20	ABCD4, ACSF3, CBS, CD320, GIF, HCFC1, IVD, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, SUCLA2, SUCLG1, SUCLG2, TCN2
Congenital Disorders of Glycosylation (CDG) Panel	5095	36	ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM3, GNE, MGAT2, MOGS, MPDU1, MPI, NGLY1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SRD5A3, TMEM165, TUSC3
CoQ10 Deficiency Panel	2100	5	ADCK3 (COQ8/CABC1), COQ2, COQ9, PDSS1, PDSS2
Fatty Acid Oxidation Deficiency Panel	2095	22	ACAD9, ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT1B, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, SLC22A5, SLC25A20, TAZ
Glycogen Storage Disease (GSD) Comprehensive Panel	2125	23	AGL, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PCK1, PCK2, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PYGL, PYGM, SLC2A2, SLC37A4
Glycogen Storage Disease (GSD) Liver Panel	2127	13	AGL, FBP1, G6PC, GAA, GBE1, GYS2, PCK1, PHKA2, PHKB, PHKG2, PYGL, SLC2A2, SLC37A4
Glycogen Storage Disease (GSD) Muscle Panel	2126	13	AGL, GAA, GYG1, GYS1, LAMP2, LDHA, PCK2, PFKM, PGAM2, PGM1, PHKA1, PHKB, PYGM
Hemochromatosis Panel	5405	5	HAMP, HFE, HFE2, SLC40A1, TFR2
Hyperinsulinism Panel	21700	8	ABCC8, GCK, GLUD1, HADH, HNF4A, INSR, KCNJ11, SLC16A1
Ketotic Hypoglycemia & Gluconeogenesis Panel	3784	4	ALDOB, FBP1, GYS2, PC
3-Methylcrotonyl-CoA Carboxylase Panel (3MCC) Panel	3782	2	MCCC1, MCCC2
Maple Syrup Urine Disease (MSUD) Panel	32870	4	BCKDHA, BCKDHB, DBT, DLD
Maturity Onset Diabetes of the Young (MODY) Panel	21900	25	ABCC8, AKT2, BLK, CISD2, CP, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, INS, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PTF1A, RFX6, SLC2A2, WFS1
Methylmalonic Acidemia Panel	3780	3	MUT, MMAA, MMAB
Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) Panel	2349	3	ETFA, ETFB, ETFDH

Panel Name	Test code	Number of Genes	Gene name*
Myopathy/ Rhabdomyolysis Panel	2300	27	ACAD9, ACADL, ACADM, ACADVL, AGL, C10orf2, CPT1B, CPT2, GAA, GYS1, HADHA, HADHB, LPIN1, OPA1, OPA3, PFKM, PGAM2, PGM1, PHKA1, POLG, POLG2, PYGM, RRM2B, SLC22A5 (OCTN2), SUCLA2, TK2, TYMP
Peroxisomal Disorders Panel	22100	22	ACOX1, AGPS, AMACR, DNM1L, GNPAT, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH, SCP2
Propionic Acidemia	2347	2	PCCA, PCCB
Proximal Urea Cycle Disorders Panel	5270	3	CPS1, NAGS, OTC
Trifunctional Protein Deficiency Panel	2345	2	HADHA, HADHB
UCD and Hyperammonemia Panel	2110	8	ARG1, ASL, ASS1, CPS1, NAGS, OTC, SLC25A13, SLC25A15

**Single gene Sanger Sequencing and Targeted Deletion/Duplication analysis are also available.*

SAMPLE & SHIPPING INFORMATION

TECHNICAL INFORMATION	
Methodology:	Targeted Capture followed by Massively Parallel Sequencing
Test Type:	Next Generation Sequencing
SAMPLE & SHIPPING INFORMATION	
Specimen Type:	Blood
Requirements:	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc (Adults/ Children).
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze. Sample must arrive within 48 hrs.
Specimen Type:	Purified DNA
Requirements:	Send 5-10 ug of purified DNA (minimal concentration of 50ng/uL; A260/A280 of ~1.7).
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Turn Around Time: 6-10 weeks	
SHIPPING ADDRESS	
2450 Holcombe, Grand Blvd. Receiving Dock, Houston, TX 77021-2024	

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MITOCHONDRIAL DISORDERS

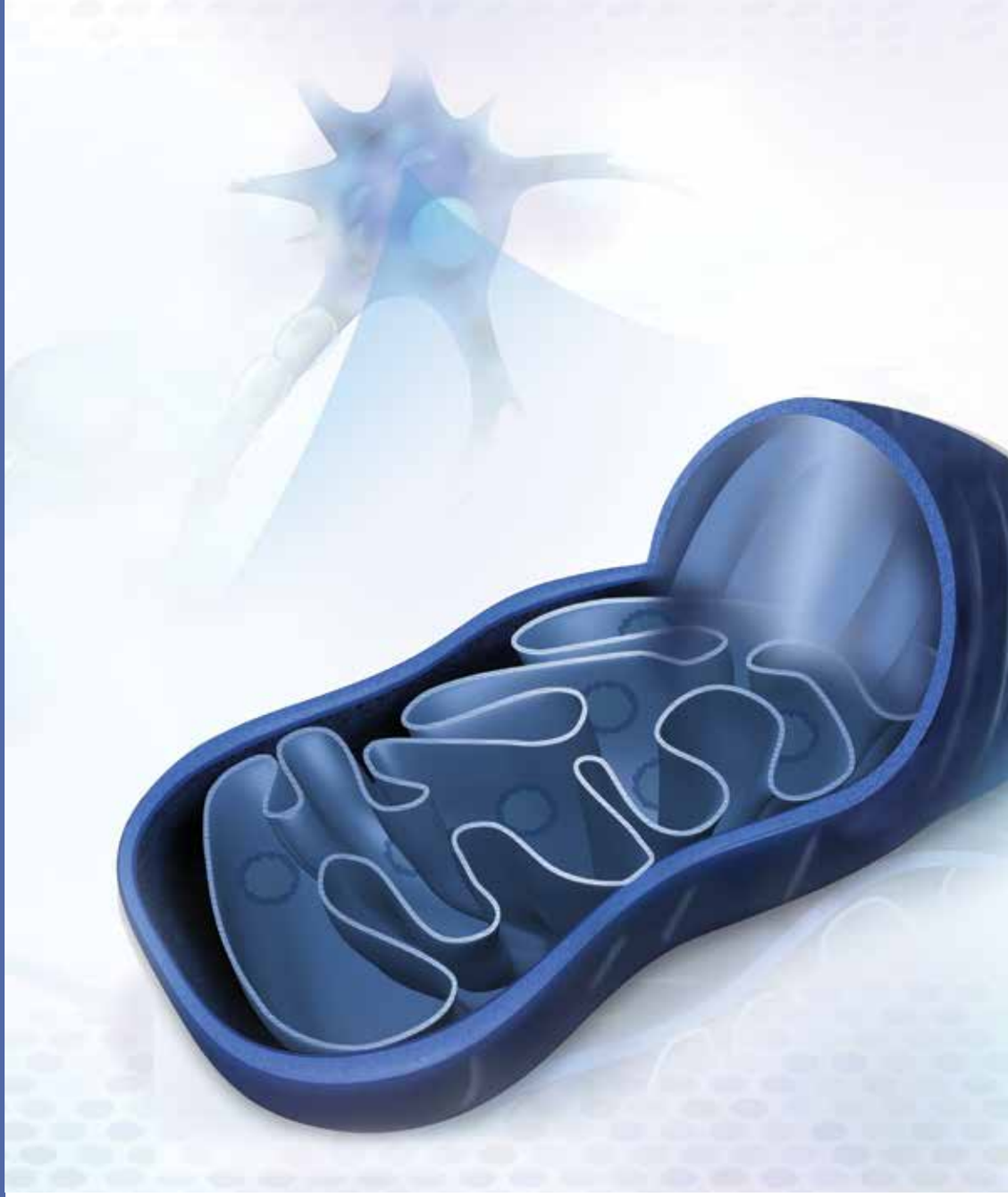
Panel Name	Gene name*
Mitochondrial respiratory chain enzyme analysis (ETC)	Skeletal Muscle 3200 Skin Fibroblasts 3210
Mitochondrial DNA copy number analysis	Skeletal Muscle 3700 Liver 3720
Mitochondrial/Metabolic (MitoMet®Plus) Microarray Analysis: Copy number analysis of the entire mtDNA and 1635 nuclear genes.	2000
Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing Screens for mtDNA deletions and 36 point mutations in MELAS, MERRF, NARP, Leigh Syndrome, LHON, Cardiomyopathy, Deafness and/or Diabetes, Pearson Syndrome, and Kearns-Sayre Syndrome.	2010
Comprehensive mtDNA analysis by next generation sequencing (MitoNGS) MitoNGS is able to detect and quantify heteroplasmic single nucleotide changes down to as low as 1.5% and determine the exact breakpoints of deletions in the mitochondrial genome.	2055

**Single gene Sanger Sequencing and Targeted Deletion/Duplication analysis are also available.*

SAMPLE & SHIPPING INFORMATION

TECHNICAL INFORMATION	
Specimen Type:	Blood (accepted for test codes 2010, 2055 and 2000)
Requirements:	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc (Adults/Children) and 3 cc (Infant<2yrs).
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Specimen Type:	Purified DNA (accepted for test codes 2010, 2055 and 2000)
Requirements:	Send at least 5ug of purified DNA (minimal concentration of 50ng/uL; A260/A280 of ~1.7).
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Specimen Type:	Skeletal Muscle (accepted for test codes 3200, 3700, 2010, 2055 and 2000)
Requirements:	50mg for Muscle
Shipping Conditions:	Skeletal Muscle should be flash frozen in liquid nitrogen at collection with no media added, stored at -80C, and shipped by overnight courier on 3-5 lbs of dry ice.
Specimen Type:	Liver (accepted for test codes 3720, 2010 and 2055)
Requirements:	50mg for Liver
Shipping Conditions:	Fresh Tissue should be flash frozen in liquid nitrogen at collection with no media added, stored at -80C, and shipped by overnight courier on 3-5 lbs of dry ice.
Specimen Type:	Cultured Skin Fibroblasts(accepted for test codes 3210, 2010, 2055 and 2000)
Requirements:	Send 1-3 T25 flasks at 60-80% confluence.
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
SHIPPING ADDRESS	
2450 Holcombe, Grand Blvd. Receiving Dock, Houston, TX 77021-2024	

Mitochondrial disorders are a group of genetically and clinically heterogeneous multi-systemic disorders. The Mitochondrial laboratory offers the most comprehensive molecular DNA analysis of the common point mutations and large deletions in the mitochondrial genome, the whole mitochondrial genome sequence analysis, and DNA sequence analysis of nuclear genes that are involved in mitochondrial DNA biosynthesis, respiratory chain enzyme complexes assembly, and metabolic disorders, including urea cycle, fatty acid oxidation, creatine pathway, etc. In addition, the laboratory offers the MitoMet oligonucleotide array CGH for the detection of copy number changes in both nuclear and mitochondrial genomes.



Shipping Information:

- Fill out the appropriate laboratory requisition form - an incomplete form will delay testing. Go to www.BMGL.com to see specific specimen requirements and shipping conditions.
- Label all specimen tubes with full name and date of birth of the patient. Provide billing information. If we are billing patient insurance, provide a copy of the front and back of the insurance card.
- For additional information, contact the laboratories at 713-798-6555 or 1-800- 411-GENE (4363).

Sample Shipping Address:

Baylor Miraca Genetics Laboratories
2450 Holcombe, Grand Blvd. Receiving Dock, Houston, TX 77021

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