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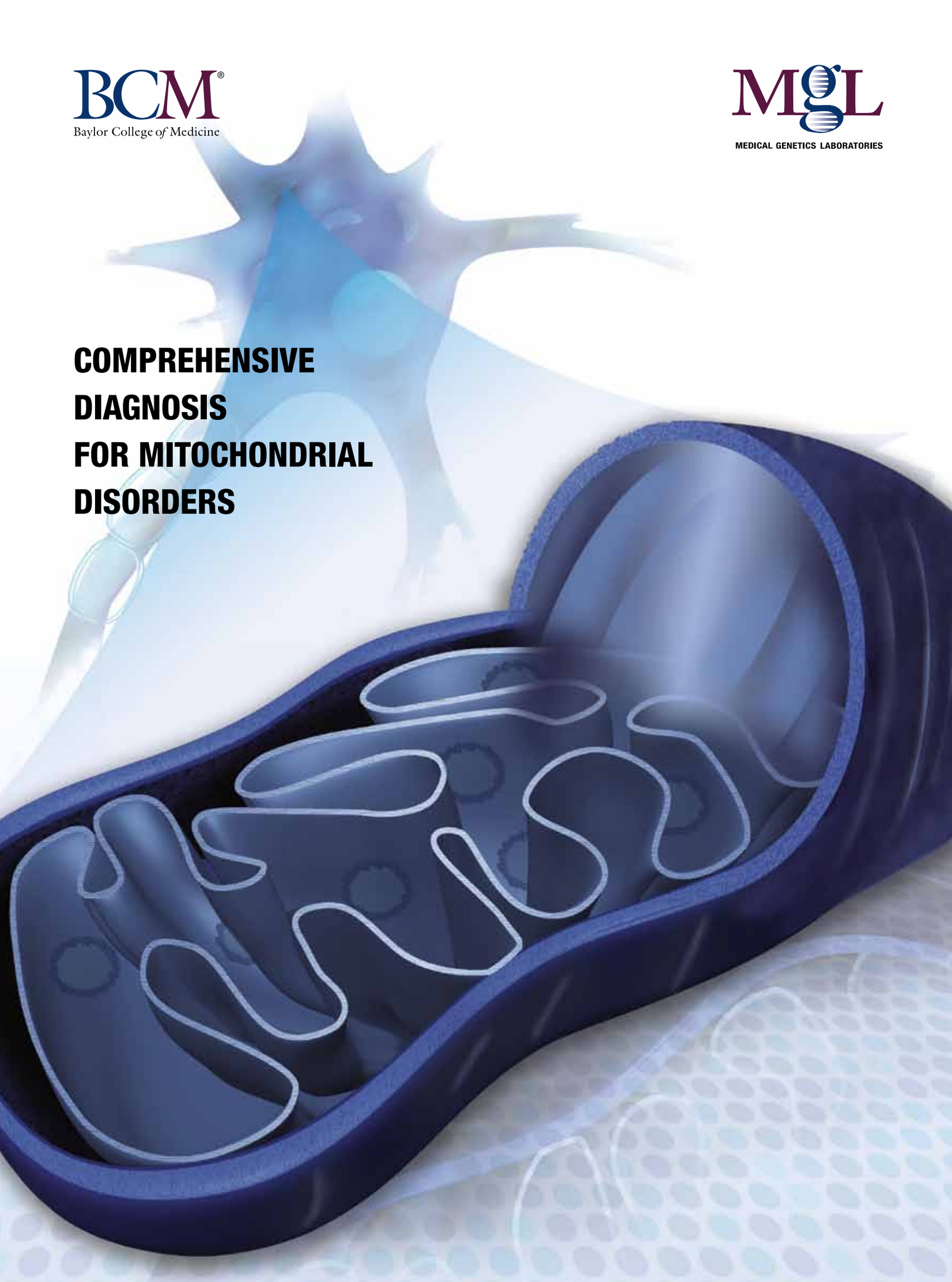
MEDICAL GENETICS LABORATORIES

COMPREHENSIVE DIAGNOSIS FOR MITOCHONDRIAL DISORDERS



MITOCHONDRIAL DISORDERS NGS PANELS

Panel Name	Test code	Gene name*
Mitome200-Nuclear	2086 (164 nuclear genes) 2085 (plus mitochondrial genome)	A ARS2, ACACA, ACACB, ACAD9, ACADL, ACADM, ACADVL, ACAT, 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 Disease 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	PDSS1, PDSS2, COQ2, COQ9, ADCK3, (COQ8/CABC1)

continued

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MITOCHONDRIAL DISORDERS NGS PANELS *continued*

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

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

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MITOCHONDRIAL DISORDERS

Panel Name	Test code
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

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METABOLIC DISORDERS

Panel Name	Test code	Number of Genes	Gene name*
Cholestasis/PFIC Panel	2105	7	ABCB11, ATP8B, ABCB4, JAG1, SLC25A13, SERPINA1, AKR1D1
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, and TUSC3
CoQ10 Deficiency Panel	2100	5	PDSS1, PDSS2, COQ2, COQ9, ADCK3 (COQ8/CABC1)
Fatty Acid Oxidation Deficiency Panel	2095	22	ACAD9, ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT1B, CPT2, ETFA, ETFB, ETFDH, HADHA, HADHB, LPIN1, SLC22A5, SLC25A20, TAZ, GLUD1, HMGCL, HMGCS2, HSD17B10, HADH
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

continued

METABOLIC DISORDERS *continued*

Panel Name	Test code	Number of Genes	Gene name*
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
Hyperinsulinism	21700	8	ABCC8, GCK, GLUD1, HADH, HNF4A, INSR, KCNJ11, SLC16A1
Maple Syrup Urine Disease (MSUD) Panel	32870	4	BCKDHA, BCKDHB, DBT, DLD
Maturity Onset Diabetes of the Young (MODY)	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
Myopathy/Rhabdomyolysis Panel	2300	27	ACADL, ACADM, ACADVL, ACAD9, 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	AP3B1, BLOC1S3, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS,5 HPS6, OCA2, SLC45A2, TYR & TYRP1
Proximal Urea Cycle Disorders Panel	5270	3	CPS1, NAGS, OTC
UCD and Hyperammonemia Panel	2110	8	CPS1, NAGS, OTC, ASS1, ASL, ARG1, SLC25A13, SLC25A15

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

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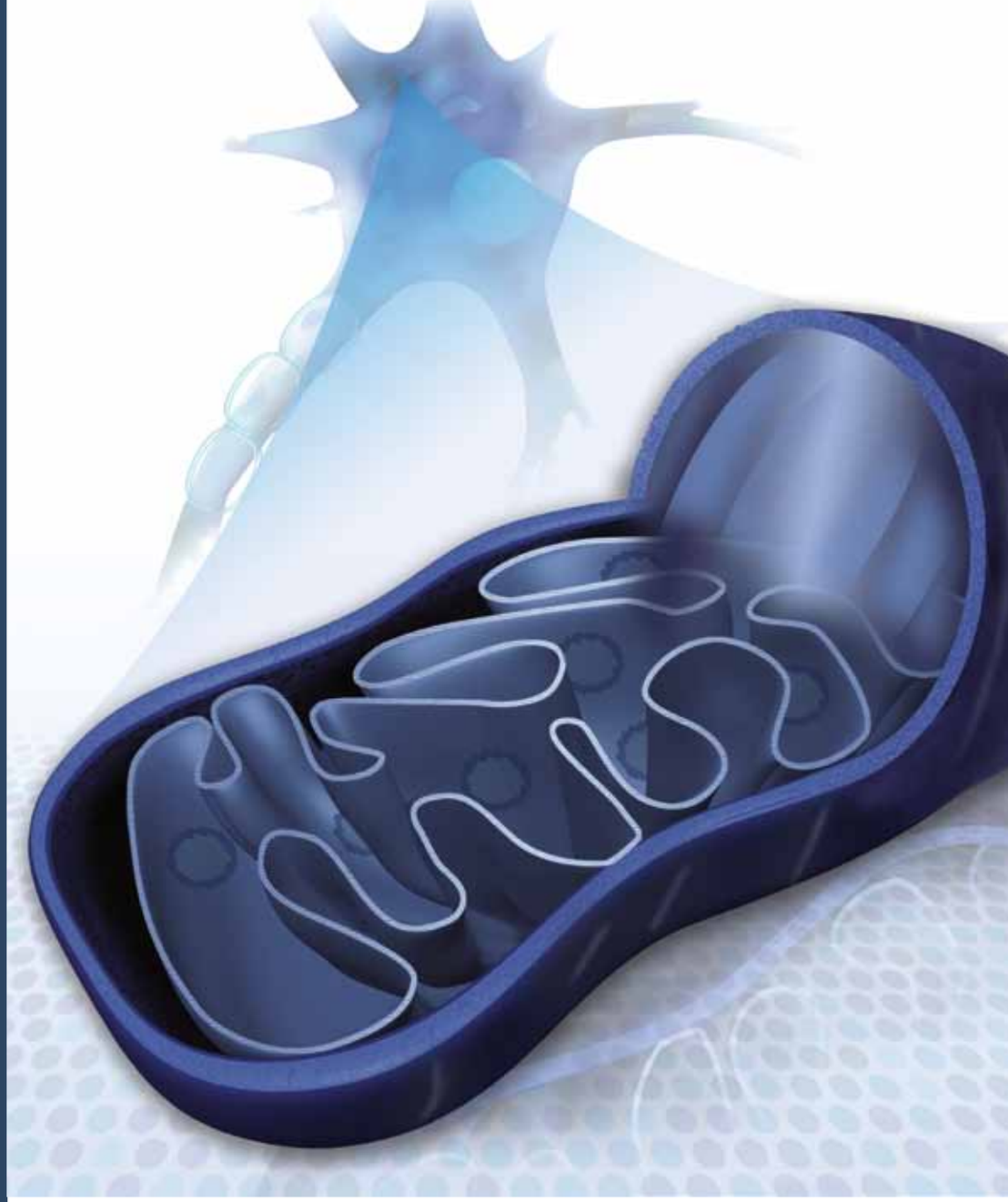
Mitochondrial disorders are a group of genetically and clinically heterogeneous multi-systemic disorders. The Mitochondrial laboratory offers the most comprehensive molecular DNA analysis of the common point mutations and large deletions in the mitochondrial genome, the whole mitochondrial genome sequence 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 recently developed 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.bcm.geneticlabs.org 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:

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