

BCM-MEDICAL GENETICS LABORATORIES

PHONE: 800-411-GENE | FAX: 713-798-2787 | www.bcmgeneticlabs.org

SHIP TO: Medical Genetics Laboratories
Baylor College of Medicine
2450 Holcombe, Grand Blvd. -Receiving Dock
Houston, TX 77021-2024
Phone: 713-798-6555

MITOCHONDRIAL DNA (mtDNA) TEST REQUISITION

PATIENT INFORMATION	INDICATION FOR STUDY	
NAME: _____ LAST NAME FIRST NAME MI	DATE OF COLLECTION: ____ / ____ / ____ MM DD YY	
DATE OF BIRTH: ____ / ____ / ____ MM DD YY GENDER (Please select one): <input type="checkbox"/> FEMALE <input type="checkbox"/> MALE <input type="checkbox"/> UNKNOWN	HOSPITAL#: _____ ACCESSION#: _____	
-OR- PLACE PATIENT STICKER HERE	SAMPLE TYPE (Please select one): <input type="checkbox"/> BLOOD <input type="checkbox"/> SKELETAL MUSCLE <input type="checkbox"/> DNA (Specify Source): _____ <input type="checkbox"/> OTHER (Specify): _____	ETHNIC BACKGROUND (Select all that apply): <input type="checkbox"/> AFRICAN AMERICAN <input type="checkbox"/> ASIAN <input type="checkbox"/> ASHKENAZIC JEWISH <input type="checkbox"/> EUROPEAN CAUCASIAN <input type="checkbox"/> HISPANIC <input type="checkbox"/> NATIVE AMERICAN INDIAN <input type="checkbox"/> OTHER JEWISH <input type="checkbox"/> OTHER (Please specify): _____

REPORTING INFORMATION	ADDITIONAL PROFESSIONAL REPORT RECIPIENTS
PHYSICIAN: _____	NAME: _____
INSTITUTION: _____	PHONE: _____ *FAX: _____
PHONE: _____ *FAX: _____	NAME: _____
EMAIL (INTERNATIONAL CLIENT REQUIREMENT): _____	PHONE: _____ *FAX: _____

***BCM-MEDICAL GENETIC LABORATORIES HAS A FAX ONLY POLICY FOR REPORTING**

INDICATION FOR STUDY	
<input type="checkbox"/> SYMPTOMATIC (Summarize below.): <div style="border: 1px solid black; height: 40px; width: 100%;"></div>	<input type="checkbox"/> *FAMILIAL MUTATION/VARIANT ANALYSIS: Complete all fields below and attach the proband's report.
<input type="checkbox"/> ASYMPTOMATIC/POSITIVE FAMILY HISTORY: (ATTACH FAMILY HISTORY) RELATIONSHIP TO PROBAND: _____ <i>*If family mutation is known, complete the FAMILIAL MUTATION/ VARIANT ANALYSIS section.</i>	GENE NAME: _____
<input type="checkbox"/> ASYMPTOMATIC/POPULATION SCREENING	MUTATION/UNCLASSIFIED VARIANT: _____
<input type="checkbox"/> OTHER (Specify clinical findings below.): <div style="border: 1px solid black; height: 40px; width: 100%;"></div>	THIS INDIVIDUAL IS CURRENTLY: <input type="checkbox"/> SYMPTOMATIC <input type="checkbox"/> ASYMPTOMATIC
	NAME OF PROBAND: _____
	RELATIONSHIP TO PROBAND: _____
	BCM LAB#: _____
	<input type="checkbox"/> A COPY OF ORIGINAL RESULTS ATTACHED
	IF PROBAND TESTING WAS PERFORMED AT ANOTHER LAB, CALL TO DISCUSS PRIOR TO SENDING SAMPLE. A POSITIVE CONTROL MAY BE REQUIRED IN SOME CASES.

REQUIRED: NEW YORK STATE PHYSICIAN SIGNATURE OF CONSENT

I certify that the patient specified above and/or their legal guardian has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions. I have obtained informed consent from the patient or their legal guardian for this testing.

Physician's Printed Name: _____ Signature: _____ Date (MM/DD/YY): _____

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LAST NAME FIRST NAME MI MM DD YY MALE
 UNKNOWN

COMPREHENSIVE mtDNA ANALYSIS BY MASSIVELY PARALLEL SEQUENCING (MitoNGS)

<input type="checkbox"/>	2055	- Sequence analysis of the entire mitochondrial genome with quantification of heteroplasmy levels for all nucleotide positions - Detection of deletions with breakpoints and heteroplasmy - Exceeds the combined capabilities of the mtDNA Common Mutation and Deletion Screening Panel (test 3000), mtDNA Whole Genome Sequencing (test 3055), and mtDNA point mutation quantification (test 3005)
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MASSIVELY PARALLEL SEQUENCING (BCM MitomeNGSSM) PANELS¹

<input type="checkbox"/>	2105	Cholestasis PANEL	4 GENES	ABCB4, ABCB11, ATP8B1, JAG1
<input type="checkbox"/>	2100	Coenzyme Q10 Deficiency PANEL	5 GENES	PDSS1, PDSS2, COQ2, COQ9, ADCK3 (COQ8/CABC1)
<input type="checkbox"/>	2120	Cobalamin Metabolism Disorders PANEL	8 GENES	TCN2, MMAA, MMAB, MMACHC, MMADHC, MTRR, LMBRD1, MUT
<input type="checkbox"/>	2125	Glycogen Metabolism Disorder PANEL	16 GENES	AGL, G6PC, GAA, GBE1, GYS1, GYS2, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PYGL, PYGM, SLC37A
<input type="checkbox"/>	2126	Glycogen Storage Disease (GSD) Muscle PANEL	9 GENES	AGL, GAA, GYS1, PFKM, PGAM2, PGM1, PHKA1, PHKB, PYGM
<input type="checkbox"/>	2127	Glycogen Storage Disease (GSD) Liver PANEL	10 GENES	AGL, G6PC, GAA, GBE1, GYS2, PHKA2, PHKB, PHKG2, PYGL, SLC37A4
<input type="checkbox"/>	2130	mtDNA Depletion/Integrity PANEL	14 GENES	C10orf2, DGUOK, MPV17, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, SUCLG2, TK2, TYMP
<input type="checkbox"/>	2140	Progressive External Ophthalmoplegia (PEO-NGS) PANEL	6 GENES	C10orf2 (TWINKLE), OPA1, POLG, POLG2, RRM2B, SLC25A4(ANT1)
<input type="checkbox"/>	2155	Mitochondrial Respiratory Chain Complex I Panel	25 GENES	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
<input type="checkbox"/>	2160	Mitochondrial Respiratory Chain Complex II Panel	6 GENES	SDHA, SDHB, SDHC, SDHD, SDHAF1, SDHAF2
<input type="checkbox"/>	2165	Mitochondrial Respiratory Chain Complex III Panel	4 GENES	BCS1L, TTC19, UQCRB, UQCRCQ
<input type="checkbox"/>	2170	Mitochondrial Respiratory Chain Complex IV Panel	12 GENES	COX10, COX15, COX4I1, COX4I2, COX6B1, COX7A1, FASTKD2, LRPPRC, SCO1, SCO2, SURF1, TACO1
<input type="checkbox"/>	2175	Mitochondrial Respiratory Chain Complex V Panel	3 GENES	ATPAF2 (ATP12), ATP5E, TMEM70
<input type="checkbox"/>	2180	Mitochondrial Respiratory Chain Complex I-V Panel	50 GENES	ATPAF2 (ATP12), ATP5E, BCS1L, C20orf7, COX10, COX15, COX4I1, COX4I2, COX6B1, COX7A1, FASTKD2, FOXRED1, LRPPRC, NDUFA1, NDUFA2, NDUFA7, NDUFA8, NDUFA10, NDUFA11, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFB6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV3, NUBPL, SCO1, SCO2, SDHA, SDHB, SDHC, SDHD, SDHAF1, SDHAF2, SURF1, TACO1, TMEM70, TTC19, UQCRB, UQCRCQ
<input type="checkbox"/>	2185	PDH & Mitochondrial RC Complex V Panel	9 GENES	PDHA1, PDHB, DLAT, DLD, PDHX, ATPAF2 (ATP12), ATP5E, TMEM70, PDP1
<input type="checkbox"/>	2300	Myopathy/Rhabdomyosis PANEL	25 GENES	ACADL, ACADM, ACADS, ACADVL, AGL, C10orf2, CPT1B, CPT2, GAA, GYS1, HADHA, HADHB, OPA1, OPA3, PFKM, PGAM2, PGM1, PHKA1, POLG, POLG2, PYGM, RRM2B, SUCLA2, TK2, TYMP

DNA COPY NUMBER ANALYSIS

<input type="checkbox"/>	3700	mtDNA Content (qPCR) Analysis- MUSCLE
<input type="checkbox"/>	3720	mtDNA Content (qPCR) Analysis- LIVER
<input type="checkbox"/>	3500	Mitochondrial/Metabolic (MitoMet [®]) Microarray Analysis Copy number analysis of selected nuclear genes + entire mtDNA REQUIRED INFORMATION BELOW. 1. Specific Disease/Gene: _____ 2. Indication: _____

¹ Single Gene Tests Available - See Molecular Diagnostic Requisition

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mtDNA RESPIRATORY CHAIN ENZYME TESTS

<input type="checkbox"/>	3200	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skeletal Muscle
<input type="checkbox"/>	3210	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts

MITOCHONDRIAL DNA (mtDNA) MUTATION SCREENS

<input type="checkbox"/>	3000	mtDNA COMMON MUTATIONS AND DELETIONS SCREEN: Screens for deletions and 13 common point mutations in MELAS, MERRF, NARP, Leigh Syndrome, LHON, Cardiomyopathy, Deafness and/or Diabetes, Pearson Syndrome, and Kearns-Sayre Syndrome.
<input type="checkbox"/>	3025*	LHON POINT MUTATIONS SCREEN: Includes m.11778G>A, m.3460G>A, m.14484T>C, and m.14459G>A which are observed in > 95% of LHON cases.

Reflex Policy for test 3000 and 3025: qPCR to measure the level of mutation heteroplasmy will be performed as a reflex test for point mutations detected by Allele-Specific Oligonucleotide hybridization. Results of the reflexive test will be reported separately under test code 3006 at no charge.

*Consider mtDNA SCREENING PANEL - POINT MUTATIONS AND DELETIONS (test code 3000) instead of test 3025.

mtDNA SANGER SEQUENCE ANALYSIS TESTS

For Familial Mutation/Variant Analysis, complete indication information on page 1.

<input type="checkbox"/>	3055*	mtDNA Whole Genome Sanger Sequence Analysis: subsumes tests 3025-3050
<input type="checkbox"/>	3050*	mtDNA Complex I Subunits Sequence Analysis
<input type="checkbox"/>	3040*	mtDNA Complex III Deficiency Sequence Analysis
<input type="checkbox"/>	3035*	mtDNA Complex IV Deficiency Sequence Analysis
<input type="checkbox"/>	3045*	mtDNA ATPase Subunits Sequence Analysis
<input type="checkbox"/>	3030	mtDNA Nonsyndromic Hearing Loss

*Consider BCM MitoNGSSM (test code 2055). BCM MitoNGSSM provides a more thorough evaluation.

NUCLEAR GENE SANGER SEQUENCE ANALYSIS PANELS*

<input type="checkbox"/>	3300	ad-PEO PANEL	3 GENES	POLG, SLC25A4 (ANT1), C10orf2 (TWINKLE)
<input type="checkbox"/>	3105	Complex IV (COX) Deficiency PANEL (nuclear genes)	4 GENES	COX10, SCO1, SCO2, SURF1
<input type="checkbox"/>	3335	mtDNA Depletion, Hepatocerebral Form PANEL	3 GENES	POLG, DGUOK, MPV17
<input type="checkbox"/>	3080	mtDNA Depletion & Multiple Deletions PANEL	4 GENES	POLG, DGUOK, SUCLA2, TK2
<input type="checkbox"/>	3620	Complex I Deficiency PANEL (nuclear genes)	13 GENES	NDUFA1, NDUFA7, NDUFAF1, NDUFAF2, NDUFAF4 (C6orf66), NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1

*Consider BCM MitomeNGSSM panels.

INDIVIDUAL NUCLEAR GENE SANGER SEQUENCE ANALYSIS TESTS

For Familial Mutation/Variant Analysis, complete indication information on page 1.

Barth Syndrome (TAZ-Related Disorders)		TAZ	Coenzyme Q10 Deficiency			
<input type="checkbox"/>	3610	TAZ Sequence Analysis	<input type="checkbox"/>	3850	ADCK3(CABC1) Sequence Analysis	ADCK3
C10orf2 (TWINKLE)-Related Disorders		C10orf2	<input type="checkbox"/>	3415	COQ2 Sequence Analysis	COQ2
<input type="checkbox"/>	3179	C10orf2 (TWINKLE) Comprehensive (Seq & Del/Dup Analysis)	<input type="checkbox"/>	3775	COQ9 Sequence Analysis	COQ9
<input type="checkbox"/>	3175	C10orf2 (TWINKLE) Sequence Analysis	<input type="checkbox"/>	3405	PDSS1 Sequence Analysis	PDSS1
<input type="checkbox"/>	3178	C10orf2 (TWINKLE) Deletion/Duplication Analysis	<input type="checkbox"/>	3410	PDSS2 Sequence Analysis	PDSS2
			<input type="checkbox"/>	4800	Coenzyme Q10 Analyte Analysis - Skeletal Muscle	

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INDIVIDUAL NUCLEAR GENE SANGER SEQUENCE ANALYSIS TESTS

For Familial Mutation/Variant Analysis, complete indication information on page 1.

Combined Oxidative Phosphorylation Deficiency				Complex III Deficiency			
<input type="checkbox"/>	3760	MRPS16 Sequence Analysis	MRPS16	<input type="checkbox"/>	3110	BCS1L Sequence Analysis	BCS1L
<input type="checkbox"/>	3645	TSFM Sequence Analysis	TSFM	<input type="checkbox"/>	2720	UQCRB Sequence Analysis	UQCRB
<input type="checkbox"/>	3810	TUFM Sequence Analysis	TUFM	<input type="checkbox"/>	2725	UQCRCQ Sequence Analysis	UQCRCQ
Complex I Deficiency				<input type="checkbox"/>	2715	TTC19 Sequence Analysis	TTC19
<input type="checkbox"/>	2655	C20orf7 Sequence Analysis	C20orf7	Complex IV Deficiency			
<input type="checkbox"/>	2660	FOXRED1 Sequence Analysis	FOXRED1	<input type="checkbox"/>	2730	COX4I1 Sequence Analysis	COX4I1
<input type="checkbox"/>	3485	NDUFA1 Sequence Analysis	NDUFA1	<input type="checkbox"/>	2735	COX4I2 Sequence Analysis	COX4I2
<input type="checkbox"/>	2665	NDUFA2 Sequence Analysis	NDUFA2	<input type="checkbox"/>	3625	COX6B1 Sequence Analysis	COX6B1
<input type="checkbox"/>	3260	NDUFA7 Sequence Analysis	NDUFA7	<input type="checkbox"/>	2740	COX7A1 Sequence Analysis	COX7A1
<input type="checkbox"/>	2670	NDUFA8 Sequence Analysis	NDUFA8	<input type="checkbox"/>	3100	COX10 Sequence Analysis	COX10
<input type="checkbox"/>	2675	NDUFA10 Sequence Analysis	NDUFA10	<input type="checkbox"/>	3545	COX15 Sequence Analysis	COX15
<input type="checkbox"/>	2680	NDUFA11 Sequence Analysis	NDUFA11	<input type="checkbox"/>	3555	FASTKD2 Sequence Analysis	FASTKD2
<input type="checkbox"/>	2685	NDUFA13 Sequence Analysis	NDUFA13	<input type="checkbox"/>	3240	LRPPRC Sequence Analysis	LRPPRC
<input type="checkbox"/>	3940	NDUFAF1 Sequence Analysis	NDUFAF1	<input type="checkbox"/>	3095	SCO1 Sequence Analysis	SCO1
<input type="checkbox"/>	3535	NDUFAF2 Sequence Analysis	NDUFAF2	<input type="checkbox"/>	3090	SCO2 Sequence Analysis	SCO2
<input type="checkbox"/>	2690	NDUFAF3 Sequence Analysis	NDUFAF3	<input type="checkbox"/>	3085	SURF1 Sequence Analysis	SURF1
<input type="checkbox"/>	3480	NDUFAF4 Sequence Analysis	NDUFAF4	<input type="checkbox"/>	2745	TACO1 Sequence Analysis	TACO1
<input type="checkbox"/>	2500	NDUFB6 Sequence Analysis	NDUFB6	Complex V Deficiency			
<input type="checkbox"/>	2700	NDUFS1 Sequence Analysis	NDUFS1	<input type="checkbox"/>	3270	ATPAF2 Sequence Analysis	ATPAF2
<input type="checkbox"/>	3930	NDUFS2 Sequence Analysis	NDUFS2	<input type="checkbox"/>	3290	ATP5E Sequence Analysis	ATP5E
<input type="checkbox"/>	3570	NDUFS3 Sequence Analysis	NDUFS3	<input type="checkbox"/>	3735	TMEM70 Sequence Analysis	TMEM70
<input type="checkbox"/>	3560	NDUFS4 Sequence Analysis	NDUFS4	Deafness-Dystonia-Optic Neuropathy			
<input type="checkbox"/>	3250	NDUFS5 Sequence Analysis	NDUFS5	<input type="checkbox"/>	3340	TIMM8A Sequence Analysis	TIMM8A
<input type="checkbox"/>	3565	NDUFS6 Sequence Analysis	NDUFS6	DGUOK-Related Disorders			
<input type="checkbox"/>	3605	NDUFS7 Sequence Analysis	NDUFS7	<input type="checkbox"/>	3079	DGUOK Comprehensive (Seq & Del/Dup Analysis)	DGUOK
<input type="checkbox"/>	3845	NDUFS8 Sequence Analysis	NDUFS8	<input type="checkbox"/>	3075	DGUOK Sequence Analysis	
<input type="checkbox"/>	3590	NDUFV1 Sequence Analysis	NDUFV1	<input type="checkbox"/>	3078	DGUOK Deletion/Duplication Analysis	
<input type="checkbox"/>	2705	NDUFV3 Sequence Analysis	NDUFV3	Ethylmalonic Encephalopathy			
<input type="checkbox"/>	2710	NUBPL Sequence Analysis	NUBPL	<input type="checkbox"/>	3745	ETHE1 Sequence Analysis	ETHE1
Complex II Deficiency				MPV17-Related Disorders			
<input type="checkbox"/>	3180	SDHA Sequence Analysis	SDHA	<input type="checkbox"/>	3324	MPV17 Comprehensive (Seq & Del/Dup Analysis)	MPV17
<input type="checkbox"/>	93185	SDHB Sequence Analysis	SDHB	<input type="checkbox"/>	3320	MPV17 Sequence Analysis	
<input type="checkbox"/>	93190	SDHC Sequence Analysis	SDHC	<input type="checkbox"/>	3323	MPV17 Deletion/Duplication Analysis	
<input type="checkbox"/>	93195	SDHD Sequence Analysis	SDHD	mtDNA Depletion Syndrome, SUCLG2 - Related			
<input type="checkbox"/>	3675	SDHAF1 Sequence Analysis	SDHAF1	<input type="checkbox"/>	3960	SUCLG2 Sequence Analysis	SUCLG2
<input type="checkbox"/>	3320	SDHAF2 Sequence Analysis	SDHAF2				

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LAST NAME FIRST NAME MI MM DD YY MALE UNKNOWN

INDIVIDUAL NUCLEAR GENE SEQUENCE ANALYSIS TESTS (CONT.) For Familial Mutation/Variant Analysis, complete indication information on page 1.

mtDNA Depletion Syndrome, Myopathic Form (TK2-Related Disorders)		<i>TK2</i>
<input type="checkbox"/>	3074	<i>TK2</i> Comprehensive (Seq & Del/Dup Analysis)
<input type="checkbox"/>	3070	<i>TK2</i> Sequence Analysis
<input type="checkbox"/>	3073	<i>TK2</i> Deletion/Duplication Analysis
Mitochondrial Phosphate Carrier Deficiency		<i>SLC25A3</i>
<input type="checkbox"/>	3490	<i>SLC25A3</i> Sequence Analysis
Mitochondrial Myopathy and Sideroblastic Anemia		<i>PUS1</i>
<input type="checkbox"/>	3650	<i>PUS1</i> Sequence Analysis
MNGIE/MNGIE like Syndrome		<i>TYMP</i>
<input type="checkbox"/>	3060	<i>TYMP</i> Sequence Analysis
Myopathy with Deficiency of ISCU		<i>ISCU</i>
<input type="checkbox"/>	3655	<i>ISCU</i> Sequence Analysis
Optic Atrophy Type 1		<i>OPA1</i>
<input type="checkbox"/>	3465	<i>OPA1</i> Sequence Analysis
Optic Atrophy Type 3		<i>OPA3</i>
<input type="checkbox"/>	3525	<i>OPA3</i> Sequence Analysis
PDH Complex Deficiency		
<input type="checkbox"/>	3169	<i>PDHA1</i> Comprehensive (Seq & Del/Dup Analysis) <i>PDHA1</i>
<input type="checkbox"/>	3165	<i>PDHA1</i> Sequence Analysis <i>PDHA1</i>
<input type="checkbox"/>	3168	<i>PDHA1</i> Deletion/Duplication Analysis <i>PDHA1</i>
<input type="checkbox"/>	3895	<i>PDHB</i> Sequence Analysis <i>PDHB</i>
<input type="checkbox"/>	3890	<i>PDP1</i> Sequence Analysis <i>PDP1</i>
<input type="checkbox"/>	3920	<i>PDHX</i> Sequence Analysis <i>PDHX</i>
<input type="checkbox"/>	3460	<i>DLD</i> Sequence Analysis <i>DLD</i>
<input type="checkbox"/>	3915	<i>DLAT</i> Sequence Analysis <i>DLAT</i>

POLG-Related Disorders		<i>POLG</i>
<input type="checkbox"/>	3069	<i>POLG</i> Comprehensive (Seq & Del/Dup Analysis)
<input type="checkbox"/>	3065	<i>POLG</i> Sequence Analysis
<input type="checkbox"/>	3068	<i>POLG</i> Deletion/Duplication Analysis
POLG2 -Related Disorders		<i>POLG2</i>
<input type="checkbox"/>	3380	<i>POLG2</i> Sequence Analysis
Pyruvate Carboxylase Deficiency		<i>PC</i>
<input type="checkbox"/>	3750	<i>PC</i> Sequence Analysis
RRM2B-Related Disorders		<i>RRM2B</i>
<input type="checkbox"/>	3420	<i>RRM2B</i> Sequence Analysis
SLC25A4-Related Disorders		<i>SLC25A4</i>
<input type="checkbox"/>	3170	<i>SLC25A4(ANT1)</i> Sequence Analysis
SUCLA2-Related Disorders		<i>SUCLA2</i>
<input type="checkbox"/>	3379	<i>SUCLA2</i> Comprehensive (Seq & Del/Dup Analysis)
<input type="checkbox"/>	3375	<i>SUCLA2</i> Sequence Analysis
<input type="checkbox"/>	3378	<i>SUCLA2</i> Deletion/Duplication Analysis
SUCLG1 -Related Disorders		<i>SUCLG1</i>
<input type="checkbox"/>	3390	<i>SUCLG1</i> Sequence Analysis
TOMM20 - Related Disorders		<i>TOMM20</i>
<input type="checkbox"/>	3475	<i>TOMM20</i> Sequence Analysis

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PATIENT NAME:

LAST NAME

FIRST NAME

MI

MITOCHONDRIAL DNA (mtDNA) DIAGNOSTICS CHECK LIST

Please indicate whether each feature is: PRESENT (Y); ABSENT (N); NOT KNOWN/TESTED (?); or IF THERE IS A FAMILY HISTORY (FHx).

If more detailed clinical information is required, please provide the name, e-mail address, and phone number of the contact person below.

CENTRAL NERVOUS SYSTEM

101	dd	Developmental Delay/MR	Y	N	?	FHx
102	ht	Hypotonia/Floppy Baby	Y	N	?	FHx
103	au	Autistic Features	Y	N	?	FHx
104	enc	Dementia/Encephalopathy	Y	N	?	FHx
105	ha	Headaches/Migraines	Y	N	?	FHx
106	stk	Stroke, Ischemic Episodes	Y	N	?	FHx
107	atx	Ataxia	Y	N	?	FHx
108	cm	Episodic Coma	Y	N	?	FHx
109	sz	Seizures	Y	N	?	FHx
110	mc	Myoclonus or Myoclonic Seizures	Y	N	?	FHx
111	pi	Perinatal Insult	Y	N	?	FHx
112	ps	Pyramidal Signs	Y	N	?	FHx
113	hp	Hemiparesis	Y	N	?	FHx
114	isz	Intractable/Refractory Seizures	Y	N	?	FHx
115	spas	Spasticity	Y	N	?	FHx
116	dyst	Dystonia	Y	N	?	FHx
117	cho	Chorea	Y	N	?	FHx

NEUROMUSCULAR

201	pn	Peripheral Neuropathy	Y	N	?	FHx
202	exi	Exercise Intolerance	Y	N	?	FHx
203	mw	Muscle Weakness/DMD-like	Y	N	?	FHx
204	cr	Muscle Cramps after Exercise	Y	N	?	FHx
205	fat	Easy Fatigability	Y	N	?	FHx
206	cmyo	Cardiomyopathy	Y	N	?	FHx
207	hb	Heart Block	Y	N	?	FHx
208	ar	Arrhythmia	Y	N	?	FHx
209	op	Ophthalmoparesis, CPEO	Y	N	?	FHx
210	emg	Abnormal EMG/NCV	Y	N	?	FHx
211	pto	Ptosis	Y	N	?	FHx

VISCERAL

301	gir	Gastrointestinal Reflux	Y	N	?	FHx
302	dge	Delayed Gastric Emptying	Y	N	?	FHx
303	pan	Pancreatitis	Y	N	?	FHx
304	dia	Diarrhea	Y	N	?	FHx
305	cst	Constipation	Y	N	?	FHx
306	cv	Cyclic Vomiting	Y	N	?	FHx
307	pob	Pseudoobstruction	Y	N	?	FHx
308	hpf	Hepatic Failure	Y	N	?	FHx
309	eta	Elevated Transaminases	Y	N	?	FHx
310	rtd	Renal Tubular Disease	Y	N	?	FHx
311	ap	Apnea/Hypoventilation	Y	N	?	FHx
312	rsf	Respiratory Deficiency/Failure	Y	N	?	FHx

METABOLITES/METOBOLIC

401	kto	Ketosis	Y	N	?	FHx
402	dca	Dicarboxylic Aciduria	Y	N	?	FHx
403	la	Lactic Acidosis	Y	N	?	FHx
404	csfl	High CSF Lactate	Y	N	?	FHx
405	oatgg	Organic Aciduria, Tiglylglycine	Y	N	?	FHx
406	lpc	Low Plasma Carnitine	Y	N	?	FHx
407	cpk	CPK Abnormalities	Y	N	?	FHx
408	pyr	Elevated Pyruvate	Y	N	?	FHx
409	csfp	Hi CSF Protein	Y	N	?	FHx
410	ala	Elevated Alanine	Y	N	?	FHx
412	3mg	3-Methylglutaconic Aciduria	Y	N	?	FHx

SENSORY

501	rp	Retinitis Pigmentosa	Y	N	?	FHx
502	opa	Optic Atrophy	Y	N	?	FHx
503	cat	Cataract	Y	N	?	FHx
504	hl	Sensorineural Hearing Loss	Y	N	?	FHx
505	trv	Tortuous Retinal Vessels	Y	N	?	FHx

ENDOCRINE

601	db	Diabetes	Y	N	?	FHx
602	pd	Exocrine/Pancreatic Deficiency	Y	N	?	FHx
603	gf	Gonadal Failure	Y	N	?	FHx
604	hth	Hypothyroidism	Y	N	?	FHx
605	hpt	Hypoparathyroidism	Y	N	?	FHx
606	adr	Hypo/Hyper-adrenal Function	Y	N	?	FHx
607	ss	Short Stature	Y	N	?	FHx

OTHER CLINICAL

701	ftt	Failure to Thrive	Y	N	?	FHx
702	mce	Microcephaly	Y	N	?	FHx
703	sids	SIDS/Unexplained Death	Y	N	?	FHx
704	ca	Congenital Anomalies	Y	N	?	FHx
705	dys	Dysmorphic Features	Y	N	?	FHx
706	id	Immunodeficiency	Y	N	?	FHx
707	ma	Macrocytic Anemia	Y	N	?	FHx
708	pcbm	Pancytopenia/Bone Marrow Failure	Y	N	?	FHx
710	np	Neutropenia	Y	N	?	FHx

HAIR/SKIN FINDINGS

711	rash	Rashes w/Hypopigmentation	Y	N	?	FHx
712	htii	Hypertrichosis	Y	N	?	FHx
713	alp	Alopecia	Y	N	?	FHx
714	ac	Acrocyanosis	Y	N	?	FHx
001	mut	Mutation	Y	N	?	FHx
003	mi	Evidence of Maternal Inheritance	Y	N	?	FHx

ELECTROPHYSIOLOGY

801	baers	Abnormal BAERS	Y	N	?	FHx
802	vers	Abnormal VERS	Y	N	?	FHx
803	eeg	Abnormal EEG	Y	N	?	FHx

IMAGING/OTHER STUDIES

804	bg	Increased Signal Basal Ganglia	Y	N	?	FHx
805	dmy	Delay Myelination	Y	N	?	FHx
806	cea	Cerebellar Atrophy	Y	N	?	FHx
807	pstk	Posterior Stroke	Y	N	?	FHx
808	leuk	Leukodystrophy	Y	N	?	FHx
809	mrsf	MRS/Lactate Peak	Y	N	?	FHx
810	mri	Abnormal MRI	Y	N	?	FHx

MUSCLE BIOPSY

901	his	Abnormal Histology	Y	N	?	FHx
902	em	Abnormal Ultrastructure (EM)	Y	N	?	FHx
903	enz	Abnormal Respiratory Enzymes	Y	N	?	FHx
904	prol	Large Mitochondrial/Proliferation	Y	N	?	FHx
905	cox	COX Deficiency	Y	N	?	FHx
906	rrf	Ragged Red Fibers	Y	N	?	FHx

BCM-MEDICAL GENETICS LABORATORIES

PHONE: 800-411-GENE | FAX: 713-798-2787 | www.bcmgeneticlabs.org

SHIP TO: Medical Genetics Laboratories
Baylor College of Medicine
2450 Holcombe, Grand Blvd. -Receiving Dock
Houston, TX 77021-2024
Phone: 713-798-6555

MITOCHONDRIAL DNA (mtDNA) TEST REQUISITION

BILLING INFORMATION

IMPORTANT NOTICE: ONE OF THE THREE FOLLOWING BILLING OPTIONS MUST BE INDICATED BELOW.
PLEASE FORWARD ALL BILLING QUESTIONS TO: MEDGENBILLING@BCM.EDU

PATIENT INFORMATION

PATIENT NAME (LAST, FIRST, MI): _____ PATIENT DATE OF BIRTH (MM/DD/YY): _____
ADDRESS: _____ CITY, STATE, ZIP: _____
PHONE: _____ EMAIL: _____

PAYMENT OPTION 1 - INSTITUTION

INSTITUTION NAME: _____ INSTITUTION CODE: _____
CONTACT NAME: _____ EMAIL (REQUIRED): _____
BILLING ADDRESS: _____ CITY, STATE, ZIP: _____
PHONE: _____ FAX: _____

PAYMENT OPTION 2 - SELF-PAY (PAYMENT MUST ACCOMPANY SAMPLE)

CREDIT CARD (PLEASE SELECT ONE): AMEX DISCOVER MC VISA
VALID CARD #: _____ EXPIRATION DATE (MM/YY): _____ CVC CODE: _____
BILLING ADDRESS: _____ CITY, STATE, ZIP: _____
CARDHOLDER PRINTED NAME: _____ CARDHOLDER SIGNATURE: _____
 CHECK/MONEY ORDER: CHECK/MONEY ORDER #: _____ AMOUNT ENCLOSED: _____

PAYMENT OPTION 3 - INSURANCE

PROVIDE A LEGIBLE PHOTOCOPY OF THE FRONT & BACK OF THE INSURANCE CARD OR HMO/MEDICAID HMO AUTHORIZATION/REFERRAL.

Please refer to the Financial Policy at www.bcmgeneticlabs.org for complete insurance filing information and managed care contract list. Insurance is filed to our contracted carriers as a client service courtesy. Patients are responsible for non-covered services, deductibles, co-insurance, contract exclusions, non-authorized services, and remaining balances after insurance reimbursement. HMO policies must have required approved authorizations. BCM-Medical Genetic Laboratories cannot bill out-of-state welfare programs. We accept authorized Texas Medicaid HMO covered charges for genetic testing. Please contact our office prior to submitting a Texas Medicaid sample. Contact medgenbilling@bcm.edu with questions.

ICD9 Diagnosis Code(s) - must be provided or insurance cannot be filed: _____

- PPO, POS, Commercial Insurance - Provide complete member information with legible front & back photocopy of insurance card.
 HMO - Provide approved authorization #: _____ and attach legible front & back photocopy of insurance card.
 Texas Medicaid HMO - Provide approved authorization #: _____ and contact Billing at 713-798-6555.

INSURED MEMBER'S INFORMATION

MEMBER NAME (Last, First, MI): _____ MEMBER DATE OF BIRTH (MM/DD/YY): _____ GENDER: FEMALE MALE
MEMBER POLICY #: _____ MEMBER SS #: _____ MEMBER GROUP #: _____
INSURANCE CO. NAME: _____ PHONE: _____
INSURANCE CO. ADDRESS: _____ CITY, STATE, ZIP: _____

I AUTHORIZE BCM-MEDICAL GENETICS LABORATORIES TO FURNISH ANY MEDICAL INFORMATION REQUESTED ON MYSELF, OR MY COVERED DEPENDENTS. IN CONSIDERATION OF SERVICES RENDERED, I TRANSFER AND ASSIGN ANY BENEFITS OF INSURANCE TO BCM-MEDICAL GENETICS LABORATORIES. I UNDERSTAND I AM RESPONSIBLE FOR ANY CO-PAY, DEDUCTIBLES, OR NON-AUTHORIZED SERVICES AND REMAINING BALANCES AFTER INSURANCE REIMBURSEMENT. I UNDERSTAND I AM FULLY RESPONSIBLE FOR PAYMENT OF MY ACCOUNT IF THE BCM-MEDICAL GENETICS LABORATORIES IS NOT A PARTICIPANT WITH MY HEALTH PLAN, AND MY HEALTH PLAN DOES NOT FULLY REIMBURSE MY MEDICAL SERVICES DUE TO LACK OF AUTHORIZATION OR MEDICAL NECESSITY.

PRINTED NAME: _____ SIGNATURE: _____ DATE (MM/YY): _____