

## MITOCHONDRIAL TESTING REQUISITION

### PATIENT INFORMATION (COMPLETE ONE FORM FOR EACH PERSON TESTED)

Patient Last Name \_\_\_\_\_ Patient First Name \_\_\_\_\_ MI \_\_\_\_\_ Date of Birth (MM / DD / YYYY) \_\_\_\_\_  
 Address \_\_\_\_\_ City \_\_\_\_\_ State \_\_\_\_\_ Zip \_\_\_\_\_ Phone \_\_\_\_\_  
 Accession # \_\_\_\_\_ Hospital / Medical Record # \_\_\_\_\_  
 Patient discharged from the hospital/facility:  Yes  No  
 Biological Sex:  Female  Male  Unknown  
 Gender identity (if different from above): \_\_\_\_\_

### REPORTING RECIPIENTS

Ordering Physician \_\_\_\_\_ Institution Name \_\_\_\_\_  
 Email (Required for International Clients) \_\_\_\_\_ Phone \_\_\_\_\_ Fax \_\_\_\_\_

### ADDITIONAL RECIPIENTS

Name \_\_\_\_\_ Email \_\_\_\_\_ Fax \_\_\_\_\_  
 Name \_\_\_\_\_ Email \_\_\_\_\_ Fax \_\_\_\_\_

### PAYMENT (FILL OUT ONE OF THE OPTIONS BELOW)

**SELF PAYMENT** .....  
 Pay With Sample  Bill To Patient  
 **INSTITUTIONAL BILLING** .....

Institution Name \_\_\_\_\_ Institution Code \_\_\_\_\_ Institution Contact Name \_\_\_\_\_ Institution Phone \_\_\_\_\_ Institution Contact Email \_\_\_\_\_

**INSURANCE** .....  
 Do Not Perform Test Until Patient is Aware of Out-Of-Pocket Costs (excludes prenatal testing)

REQUIRED ITEMS 1. Copy of the Front/Back of Insurance Card(s) 2. ICD10 Diagnosis Code(s) 3. Name of Ordering Physician 4. Insured Signature of Authorization

Name of Insured _____	Insured Date of Birth (MM / DD / YYYY) _____	Name of Insured _____	Insured Date of Birth (MM / DD / YYYY) _____
Patient's Relationship to Insured _____	Phone of Insured _____	Patient's Relationship to Insured _____	Phone of Insured _____
Address of Insured _____		Address of Insured _____	
City _____	State _____ Zip _____	City _____	State _____ Zip _____
Primary Insurance Co. Name _____	Primary Insurance Co. Phone _____	Secondary Insurance Co. Name _____	Secondary Insurance Co. Phone _____
Primary Member Policy # _____	Primary Member Group # _____	Secondary Member Policy # _____	Secondary Member Group # _____

By signing below, I hereby authorize Baylor Genetics to provide my insurance carrier any information necessary, including test results, for processing my insurance claim. I understand that I am responsible for any co-pay, co-insurance, and unmet deductible that the insurance policy dictates, as well as any amounts not paid by my insurance carrier for reasons including, but not limited to, non-covered and non-authorized services. I understand that I am responsible for sending Baylor Genetics any and all payments that I receive directly from my insurance company in payment for this test. Please note that Medicare does not cover routine screening tests.

Patient's Printed Name \_\_\_\_\_ Patient's Signature \_\_\_\_\_ Date (MM / DD / YYYY) \_\_\_\_\_

### STATEMENT OF MEDICAL NECESSITY (REQUIRED)

This test is medically necessary for the risk assessment, diagnosis, or detection of a disease, illness, impairment, symptom, syndrome, or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Physician is authorized by law to order the test(s) requested herein. I confirm that I have provided genetic testing information to the patient and they have consented to genetic testing.

Physician's Printed Name \_\_\_\_\_ Physician's Signature \_\_\_\_\_ Date (MM / DD / YYYY) \_\_\_\_\_

## MITOCHONDRIAL TESTING REQUISITION

Patient Last Name \_\_\_\_\_ Patient First Name \_\_\_\_\_ MI \_\_\_\_\_ Date of Birth (MM / DD / YYYY) \_\_\_\_\_ Biological Sex \_\_\_\_\_

### ETHNICITY

- |  |   |   |
|--|---|---|
| <input type="radio"/> African American                 | <input type="radio"/> Hispanic American                                       | <input type="radio"/> Pacific Islander (Philippines, Micronesia, Malaysia, Indonesia) |
| <input type="radio"/> Ashkenazi Jewish                 | <input type="radio"/> Mennonite   | <input type="radio"/> South Asian (India, Pakistan)                                   |
| <input type="radio"/> East Asian (China, Japan, Korea) | <input type="radio"/> Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)      | <input type="radio"/> Southeast Asian (Vietnam, Cambodia, Thailand)                   |
| <input type="radio"/> Finnish                          | <input type="radio"/> Native American   | <input type="radio"/> Southern European Caucasian (Spain, Italy, Greece)              |
| <input type="radio"/> French Canadian                  | <input type="radio"/> Northern European Caucasian (Scandinavian, UK, Germany) | <input type="radio"/> Other (Specify): _____  |

### SAMPLE

SAMPLE TYPE	DATE OF COLLECTION (MM/DD/YYYY)
<input type="radio"/> Blood in EDTA (Purple-top)	____ / ____ / ____
<input type="radio"/> Cord Blood	____ / ____ / ____
<input type="radio"/> DNA, Extracted from:	____ / ____ / ____
<input type="radio"/> Liver	____ / ____ / ____
<input type="radio"/> Saliva	____ / ____ / ____
<input type="radio"/> Skin Fibroblast Culture	____ / ____ / ____
<input type="radio"/> Skeletal Muscle	____ / ____ / ____
<input type="radio"/> Tissue	____ / ____ / ____

**NOTE:** Extracted DNA/RNA will only be accepted if the isolation of nucleic acids for clinical testing occurs in a CLIA-certified laboratory or a laboratory meeting equivalent requirements as determined by the CAP and/or the CMS.

### TESTING OPTIONS

- Targeted Sequencing for Known Familial Mutation  
(If selected, specify test code and gene and complete section below)
- |                               |                                  |
|-------------------------------|----------------------------------|
| Test Code _____               | Gene _____                       |
| Proband Last Name _____       | Proband First Name _____         |
| Relationship to Proband _____ | Date of Birth (MM/DD/YYYY) _____ |
- Proband testing location (Select one)
- Baylor Genetics
- |             |                |
|-------------|----------------|
| Lab # _____ | Family # _____ |
|-------------|----------------|
- Another Laboratory
- Attach a copy of the Proband test results.
  - A positive control sample of the Proband is requested. Please provide, if available.
- Full Gene Sequencing
- Deletion/ Duplication Analysis

### INDICATION FOR TESTING (REQUIRED)

- Symptomatic with Positive Family History
- Symptomatic (Summarize below):
- Asymptomatic
- Population Screening       Positive Family History

Disease \_\_\_\_\_ Gene \_\_\_\_\_ Variant \_\_\_\_\_

ICD10 Diagnosis Code(s):

### MITOCHONDRIAL TESTS

#### MITOCHONDRIAL PANELS

TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/> 2085	Dual Genome Panel by Massively Parallel Sequencing (BCM-MitomeNGS <sup>SM</sup> )	BE, DNA, SFC, SM
<input type="checkbox"/> 20600	Dual Genome Leigh Disease Panel by Massively Parallel Sequencing (BCM-MitomeNGS <sup>SM</sup> )	BE, DNA
<input type="checkbox"/> 2055	Comprehensive mtDNA by Massively Parallel Sequencing (BCM-MitomeNGS <sup>SM</sup> )	BE, DNA, L, SM, T

#### MASSIVELY PARALLEL SEQUENCING (BCM-MITOMENGS<sup>SM</sup>) PANELS

TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/> 20100	Albinism Panel (13 genes)	BE, BUC, DNA
<input type="checkbox"/> 20400	Bardet-Biedl Syndrome Panel (18 genes)	BE, BUC, DNA
<input type="checkbox"/> 2105	Cholestasis Panel (7 genes)	BE, BUC, DNA
<input type="checkbox"/> 2120	Cobalamin Metabolism Panel + Severe <i>MTHFR</i> Deficiency (20 genes)	BE, BUC, DNA
<input type="checkbox"/> 2625	<i>COL1A1</i> and <i>COL1A2</i> Panel	BE, BUC, DNA

\* Refer to Sample Specifications Table (Page 8)      Test list continued on next page

## MITOCHONDRIAL TESTING REQUISITION

\_\_\_\_\_  
 Patient Last Name                      Patient First Name                      MI                      Date of Birth (MM / DD / YYYY)                      Biological Sex

### MITOCHONDRIAL TESTS

#### MASSIVELY PARALLEL SEQUENCING (BCM-MITOMENGS<sup>SM</sup>) PANELS

TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/>	5095 Congenital Disorders of Glycosylation Panel (36 genes)	BE, BUC, DNA
<input type="checkbox"/>	2100 CoQ10 Deficiency Panel ( <i>PDSS1, PDSS2, COQ2, COQ9, and ADCK3(COQ8/CABC1)</i> )	BE, BUC, DNA
<input type="checkbox"/>	5260 Developmental Glaucoma Panel (8 genes)	BE, BUC, DNA
<input type="checkbox"/>	5250 Familial Exudative Vitreoretinopathy Panel ( <i>FZD4, LRP5, NDP, and TSPAN12</i> )	BE, BUC, DNA
<input type="checkbox"/>	2095 Fatty Acid Oxidation Panel (20 genes)	BE, BUC, DNA
<input type="checkbox"/>	2125 Glycogen Storage Disease (GSD) Panel (23 genes)	BE, BUC, DNA
<input type="checkbox"/>	2126 Glycogen Storage Disease (GSD) Muscle Panel (13 genes)	BE, BUC, DNA
<input type="checkbox"/>	2127 Glycogen Storage Disease (GSD) Liver Panel (13 genes)	BE, BUC, DNA
<input type="checkbox"/>	2200 High Bone Mass Panel (14 genes)	BE, BUC, DNA
<input type="checkbox"/>	21700 Hyperinsulinism Panel (8 genes)	BE, BUC, DNA
<input type="checkbox"/>	21000 Hypoglycemia Panel (85 genes)	BE, BUC, DNA
<input type="checkbox"/>	5090 Leber Congenital Amaurosis Panel (19 genes)	BE, BUC, DNA
<input type="checkbox"/>	20601 Leigh Disease Panel (82 genes)	BE, BUC, DNA
<input type="checkbox"/>	2090 Low Bone Mass Panel (23 genes)	BE, BUC, DNA
<input type="checkbox"/>	32870 Maple Syrup Urine Disease (MSUD) Panel ( <i>BCKHDA, BCKHDB, DBT and DLD</i> )	BE, BUC, DNA
<input type="checkbox"/>	21900 Maturity-Onset Diabetes of the Young (MODY) Panel (25 genes)	BE, BUC, DNA
<input type="checkbox"/>	2130 mtDNA Depletion/Integrity Panel (19 genes)	BE, BUC, DNA
<input type="checkbox"/>	2155 Mitochondrial Respiratory Chain Complex I Deficiency Panel (21 genes)	BE, BUC, DNA
<input type="checkbox"/>	2160 Mitochondrial Respiratory Chain Complex II Deficiency Panel ( <i>SDHA, SDHB, SDHC, SDHD, and SDHAF1</i> )	BE, BUC, DNA
<input type="checkbox"/>	2165 Mitochondrial Respiratory Chain Complex III Deficiency Panel ( <i>BCS1L, TTC19, UQCRB, and UQCRCQ</i> )	BE, BUC, DNA
<input type="checkbox"/>	2170 Mitochondrial Respiratory Chain Complex IV Deficiency Panel (10 genes)	BE, BUC, DNA
<input type="checkbox"/>	2175 Mitochondrial Respiratory Chain Complex V Deficiency Panel ( <i>ATPAF2, ATP5E, and TMEM70</i> )	BE, BUC, DNA
<input type="checkbox"/>	2086 Nuclear Panel (163 genes)	BE, BUC, DNA
<input type="checkbox"/>	2180 Mitochondrial Respiratory Chain Complex I-V Panel (50 genes)	BE, BUC, DNA
<input type="checkbox"/>	2300 Myopathy/Rhabdomyolysis Panel (25 genes)	BE, BUC, DNA
<input type="checkbox"/>	20200 Nephronophthisis Panel ( <i>NPHP1, INVS, NPHP3, NPHP4</i> )	BE, BUC, DNA
<input type="checkbox"/>	24001 Noonan Spectrum Disorders Panel (26 genes)	BE, BUC, DNA, SA
<input type="checkbox"/>	2185 PDH & Mitochondrial RC Complex V Panel (9 genes)	BE, BUC, DNA
<input type="checkbox"/>	22100 Peroxisomal Disorders Panel (22 genes)	BE, BUC, DNA
<input type="checkbox"/>	5255 Primary Open Angle Glaucoma Panel (MYOC, OPTN)	BE, BUC, DNA
<input type="checkbox"/>	5274 Proximal Urea Cycle Disorders Comprehensive (Seq. & Del/Dup) ( <i>CPS1, NAGS, OTC</i> )	BE, BUC, DNA
<input type="checkbox"/>	2140 Progressive External Ophthalmoplegia Panel (10 genes)	BE, BUC, DNA
<input type="checkbox"/>	2190 Retinitis Pigmentosa + RPGR orf15 by NGS (66 genes)	BE, BUC, DNA
<input type="checkbox"/>	2110 Urea Cycle Disorders and Hyperammonemia (8 genes)	BE, BUC, DNA
<input type="checkbox"/>	2195 Usher Syndrome Panel (9 genes)	BE, BUC, DNA

#### DNA COPY NUMBER ANALYSIS

TEST CODE	TEST NAME	SAMPLE TYPE *	SPECIFY GENE OF INTEREST
<input type="checkbox"/>	3700 mtDNA Content (qPCR) Analysis - Skeletal Muscle	SM	
<input type="checkbox"/>	3720 mtDNA Content (qPCR) Analysis - Liver	L	
<input type="checkbox"/>	2000 MitoMet <sup>®</sup> Plus aCGH Analysis	BE, DNA	
<input type="checkbox"/>	2001 Oligonucleotide Targeted Array Analysis (Single Target Gene)	BE, DNA	<input type="text"/>
<input type="checkbox"/>	2003 Oligonucleotide Targeted Array Analysis (Up to 5 Target Genes)	BE, DNA	<input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

\* Refer to Sample Specifications Table (Page 8)

Test list continued on next page

## MITOCHONDRIAL TESTING REQUISITION

\_\_\_\_\_  
 Patient Last Name                      Patient First Name                      MI                      Date of Birth (MM / DD / YYYY)                      Biological Sex

### MITOCHONDRIAL TESTS

#### MITOCHONDRIAL DNA (mtDNA) RESPIRATORY CHAIN ENZYME TESTS

TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/> 3200	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skeletal Muscle	SM
<input type="checkbox"/> 3210	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts	SFC

#### MITOCHONDRIAL DNA (mtDNA) MUTATION SCREENS

TEST CODE	TEST NAME	SAMPLE TYPE *	TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/> 2010	Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGS <sup>SM</sup> )	BE, DNA, SM, T	<input type="checkbox"/> 3030	mtDNA Nonsyndromic Hearing Loss and Deafness Mutation Panel	BA, DNA, SA, SM, T

#### SINGLE GENE ANALYSIS

If a test is not found on this form, please obtain the test code from our website ([www.BMGL.com](http://www.BMGL.com)) and write in the below space(s).

\_\_\_\_\_  
 Test Code                      Gene                      Test Code                      Gene                      Test Code                      Gene

\_\_\_\_\_  
 Test Name                      Test Name                      Test Name

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3904	ACAD9 Comprehensive (Seq & Del/Dup Analysis)	ACAD9 Deficiency	BE, DNA
<input type="checkbox"/> 2219	ATP5A1 Comprehensive (Seq & Del/Dup Analysis)	ATP5A1-Related Disorders	BE, DNA
<input type="checkbox"/> 3614	TAZ Comprehensive (Seq & Del/Dup Analysis)	Barth Syndrome (TAZ-Related Disorders)	BE, DNA
<input type="checkbox"/> 3179	C10orf2 (TWINKLE) Comprehensive (Seq & Del/Dup Analysis)	C10orf2 (TWINKLE)-Related Disorders	BE, DNA
<input type="checkbox"/> 3854	CABC1(ADCK3) Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA
<input type="checkbox"/> 3419	COQ2 Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA
<input type="checkbox"/> 3414	PDS2 Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA
<input type="checkbox"/> 2264	GFM1 Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA
<input type="checkbox"/> 3649	TSM Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA
<input type="checkbox"/> 2289	MRPS22 Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA
<input type="checkbox"/> 2224	C12orf65 Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA
<input type="checkbox"/> 2324	AARS2 Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA
<input type="checkbox"/> 2664	FOXRED1 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3489	NDUFA1 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 2684	NDUFA11 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3944	NDUFA1 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3539	NDUFA2 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 2694	NDUFA3 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 2704	NDUFS1 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3574	NDUFS3 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA

\* Refer to Sample Specifications Table (Page 8)

Test list continued on next page



## MITOCHONDRIAL TESTING REQUISITION

Patient Last Name \_\_\_\_\_

Patient First Name \_\_\_\_\_

MI \_\_\_\_\_

Date of Birth (MM / DD / YYYY) \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Biological Sex \_\_\_\_\_

### MITOCHONDRIAL TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3564	NDUFS4 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3569	NDUFS6 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3849	NDUFS8 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3594	NDUFV1 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 2714	NUBPL Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA
<input type="checkbox"/> 3180	SDHA Sequence Analysis	Complex II Deficiency	BE, DNA, SA
<input type="checkbox"/> 3185	SDHB Sequence Analysis	Complex II Deficiency	BE, DNA, SA
<input type="checkbox"/> 3190	SDHC Sequence Analysis	Complex II Deficiency	BE, DNA, SA
<input type="checkbox"/> 3195	SDHD Sequence Analysis	Complex II Deficiency	BE, DNA, SA
<input type="checkbox"/> 3679	SDHAF1 Comprehensive (Seq & Del/Dup Analysis)	Complex II Deficiency	BE, DNA
<input type="checkbox"/> 3114	BCS1L Comprehensive (Seq & Del/Dup Analysis)	Complex III Deficiency	BE, DNA
<input type="checkbox"/> 2719	TTC19 Comprehensive (Seq & Del/Dup Analysis)	Complex III Deficiency	BE, DNA
<input type="checkbox"/> 2734	COX4I1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 3104	COX10 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 3549	COX15 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 3099	SCO1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 3094	SCO2 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 3089	SURF1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 2749	TACO1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA
<input type="checkbox"/> 3294	ATP5E Comprehensive (Seq & Del/Dup Analysis)	Complex V Deficiency	BE, DNA
<input type="checkbox"/> 3739	TMEM70 Comprehensive (Seq & Del/Dup Analysis)	Complex V Deficiency	BE, DNA
<input type="checkbox"/> 3344	TIMM8A Comprehensive (Seq & Del/Dup Analysis)	Deafness-Dystonia-Optic Neuropathy	BE, DNA
<input type="checkbox"/> 3079	DGUOK Comprehensive (Seq & Del/Dup Analysis)	DGUOK-Related Disorders	BE, DNA
<input type="checkbox"/> 3749	ETHE1 Comprehensive (Seq & Del/Dup Analysis)	Ethylmalonic Encephalopathy	BE, DNA
<input type="checkbox"/> 2249	FARS2 Comprehensive (Seq & Del/Dup Analysis)	FARS2-Related Disorders	BE, DNA
<input type="checkbox"/> 3559	FASTKD2 Comprehensive (Seq & Del/Dup Analysis)	FASTKD2-Related Disorders	BE, DNA
<input type="checkbox"/> 2314	HARS2 Comprehensive (Seq & Del/Dup Analysis)	HARS2-Related Disorders	BE, DNA
<input type="checkbox"/> 2329	KARS Comprehensive (Seq & Del/Dup Analysis)	Intermediate Charcot-Marie-Tooth Neuropathy, KARS-Related	BE, DNA
<input type="checkbox"/> 2269	ACAT1 Comprehensive (Seq & Del/Dup Analysis)	Ketothiolase Deficiency	BE, DNA
<input type="checkbox"/> 3464	DLD Comprehensive (Seq & Del/Dup Analysis)	Maple Syrup Urine Disease Type 3	BE, DNA
<input type="checkbox"/> 2229	MARS2 Comprehensive (Seq & Del/Dup Analysis)	MARS2 Related Disorders	BE, DNA

\* Refer to Sample Specifications Table (Page 8)

Test list continued on next page



## MITOCHONDRIAL TESTING REQUISITION

Patient Last Name

Patient First Name

MI

Date of Birth (MM / DD / YYYY)

Biological Sex

### MITOCHONDRIAL TESTS

#### INDIVIDUAL MITOCHONDRIAL TESTS (LISTED BY DISORDER)

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3964	SUCLG2 Comprehensive (Seq & Del/Dup Analysis)	mtDNA Depletion Syndrome, SUCLG2-Related	BE, DNA
<input type="checkbox"/> 3074	TK2 Comprehensive (Seq & Del/Dup Analysis)	mtDNA Depletion Syndrome, Myopathic Form (TK2-Related Disorders)	BE, DNA
<input type="checkbox"/> 3064	TYMP Comprehensive (Seq & Del/Dup Analysis)	MNGIE/MNGIE like Syndrome	BE, DNA
<input type="checkbox"/> 3324	MPV17 Comprehensive (Seq & Del/Dup Analysis)	MPV17-Related Disorders	BE, DNA
<input type="checkbox"/> 2294	MRPL44 Comprehensive (Seq & Del/Dup Analysis)	MRPL44-Related Disorders	BE, DNA
<input type="checkbox"/> 2235	MTFMT Sequence Analysis	MTFMT-Related Disorders	BE, DNA, SA
<input type="checkbox"/> 3659	ISCU Comprehensive (Seq & Del/Dup Analysis)	Myopathy with Deficiency of ISCU	BE, DNA
<input type="checkbox"/> 3654	PUS1 Comprehensive (Seq & Del/Dup Analysis)	Myopathy, Mitochondrial, and Sideroblastic Anemia	BE, DNA
<input type="checkbox"/> 3959	YARS2 Comprehensive (Seq & Del/Dup Analysis)	Myopathy, Mitochondrial, and Sideroblastic Anemia	BE, DNA
<input type="checkbox"/> 2309	NARS2 Comprehensive (Seq & Del/Dup Analysis)	NARS2-Related Disorders	BE, DNA
<input type="checkbox"/> 3529	OPA3 Comprehensive (Seq & Del/Dup Analysis)	Optic Atrophy Type 3	BE, DNA
<input type="checkbox"/> 3169	PDHA1 Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3899	PDHB Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3894	PDP1 Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3924	PDHX Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3919	DLAT Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3069	POLG Comprehensive (Seq & Del/Dup Analysis)	POLG-Related Disorders	BE, DNA
<input type="checkbox"/> 3384	POLG2 Comprehensive (Seq & Del/Dup Analysis)	POLG2-Related Disorders	BE, DNA
<input type="checkbox"/> 3754	PC Comprehensive (Seq & Del/Dup Analysis)	Pyruvate Carboxylase Deficiency	BE, DNA
<input type="checkbox"/> 3424	RRM2B Comprehensive (Seq & Del/Dup Analysis)	RRM2B-Related Disorders	BE, DNA
<input type="checkbox"/> 3174	SLC25A4 (ANT1) Comprehensive (Seq & Del/Dup Analysis)	SLC25A4-Related Disorders	BE, DNA
<input type="checkbox"/> 5335	SPG7 Sequence Analysis	Spastic Paraplegia 7, Autosomal Recessive	BE, DNA, SA
<input type="checkbox"/> 3379	SUCLA2 Comprehensive (Seq & Del/Dup Analysis)	SUCLA2-Related Disorders	BE, DNA
<input type="checkbox"/> 3394	SUCLG1 Comprehensive (Seq & Del/Dup Analysis)	SUCLG1-Related Disorders	BE, DNA

\* Refer to Sample Specifications Table (Page 8)

Indications on next page



## MITOCHONDRIAL TESTING REQUISITION

Patient Last Name \_\_\_\_\_

Patient First Name \_\_\_\_\_

MI \_\_\_\_\_

Date of Birth (MM / DD / YYYY) \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Biological Sex \_\_\_\_\_

### INDICATION FOR TESTING (REQUIRED)

Clinical management of known diagnosis - Please specify: \_\_\_\_\_

Diagnostic Testing - Please complete checklist below.

#### CENTRAL NERVOUS SYSTEM .....

- 101 dd Developmental Delay/ ID
- 102 ht Hypotonia
- 103 au Autistic Features
- 104 enc Dementia/ Encephalopathy
- 105 ha Headaches/ Migraines
- 106 stk Stroke, Ischemic Episodes
- 107 atx Ataxia
- 108 sz Intractable/ Refractory/ Myoclonus/Myoclonic Seizures
- 109 pi Perinatal Insult
- 110 ps Pyramidal Signs
- 111 hp Hemiparesis
- 112 spas Spasticity
- 113 dyst Dystonia
- 114 cho Chorea
- 115 sib Self-Injury
- 116 pan Pancreatitis
- 117 dia Diarrhea
- 118 cst Constipation
- 119 cv Cyclic Vomiting
- 120 pob Pseudoobstruction

#### VISCERAL .....

- 301 gir Gastrointestinal Reflux
- 302 dge Delayed Gastric Emptying
- 303 pan Pancreatitis
- 304 dia Diarrhea
- 305 cst Constipation
- 306 cv Cyclic Vomiting
- 307 pob Pseudoobstruction
- 308 hpf Hepatic Failure
- 309 eta Elevated Transaminases
- 310 rtd Renal Tubular Disease
- 311 ap Apnea/ Hypoventilation
- 312 rsf Respiratory Deficiency/Failure
- 313 ren Renal Dysfunction
- 314 lc Liver Carcinoma
- 315 jau Jaundice
- 316 spm Splenomegaly/Enlarged Spleen
- 317 hpm Hepatomegaly/Enlarged Liver
- 318 hd Hepatic Dysfunction

#### SENSORY .....

- 501 rp Retinitis Pigmentosa
- 502 opa Optic Atrophy
- 503 cat Cataract
- 504 hl Sensorineural Hearing Loss
- 505 trv Tortuous Retinal Vessels
- 506 crs Cherry Red Spot/Eye
- 507 co Corneal Opacity
- 508 el Ectopia Lentis
- 509 pp Photophobia

#### ENDOCRINE .....

- 601 db Diabetes
- 602 pd Exocrine/Pancreatic Deficiency
- 603 gf Gonadal Failure
- 604 hth Hypothyroidism
- 605 hpt Hypoparathyroidism
- 606 adr Hypo/Hyper-adrenal Function
- 607 ss Short Stature
- 608 adc Adrenal Calcification
- 609 hf Hydrops Fetalis
- 610 pg Pregnant

#### NEUROMUSCULAR .....

- 201 pn Peripheral Neuropathy
- 202 exi Exercise Intolerance
- 203 pmw Progressive Muscle Weakness
- 204 smw Static Muscle Weakness
- 205 cr Muscle Cramps after Exercise
- 206 fat Easy Fatigability
- 207 dcmyo Dilated Cardiomyopathy
- 208 hcmyo Hypertrophic Cardiomyopathy
- 209 hb Heart Block
- 210 ar Arrhythmia
- 211 op Ophthalmoparesis, CPEO
- 212 emg Abnormal EMG/NCV
- 213 pto Ptosis
- 214 eh Cardiomegaly/Enlarged Heart

#### METABOLITES / METABOLIC .....

- 400 nbs Abnormal Newborn Screen
- 401 kto Ketosis
- 402 dca Dicarboxylic Aciduria
- 403 la Lactic Acidosis
- 404 csfl High CSF Lactate
- 405 oa Organic Aciduria
- 406 lpc Low Plasma Carnitine
- 407 cpk CPK Abnormalities
- 408 pyr Elevated Pyruvate
- 409 ala Elevated Alanine
- 410 3mg 3-Methylglutaconic Aciduria
- 411 acid Acidosis
- 412 NH3 Hypoammonemia
- 413 hypo Hypoglycemia
- 414 hyper Hyperglycemia
- 415 uco Unusual Color/Odor

#### OTHER CLINICAL .....

- 701 ftt Failure to Thrive
- 702 mce Microcephaly
- 703 sids SIDS/Unexplained Death
- 704 ca Congenital Anomalies
- 705 dys Dysmorphic Features
- 706 id Immunodeficiency
- 707 ma Macrocytic Anemia
- 708 pcbm Pancytopenia/Bone Marrow Failure
- 709 np Neutropenia
- 710 mc Macrocephaly
- 711 cf Course Features
- 712 sa Skeletal Anomalies
- 713 art Arthritis



## MITOCHONDRIAL TESTING REQUISITION

Patient Last Name \_\_\_\_\_ Patient First Name \_\_\_\_\_ MI \_\_\_\_\_ Date of Birth (MM / DD / YYYY) \_\_\_\_\_ Biological Sex \_\_\_\_\_

### INDICATION FOR TESTING - CONTINUED (REQUIRED)

**FAMILY HISTORY** ..... **ELECTROPHYSIOLOGY** .....

- |                              |     |                                  |                              |       |                |
|------------------------------|-----|----------------------------------|------------------------------|-------|----------------|
| <input type="checkbox"/> 001 | mut | Mutation (Attach details)        | <input type="checkbox"/> 801 | baers | Abnormal BAERS |
| <input type="checkbox"/> 002 | mi  | Evidence of Maternal Inheritance | <input type="checkbox"/> 802 | vers  | Abnormal VERS  |
|                              |     |                                  | <input type="checkbox"/> 803 | eeg   | Abnormal EEG   |

**HAIR/SKIN FINDINGS** ..... **IMAGING/OTHER STUDIES** ..... **MUSCLE BIOPSY** .....

- |                              |      |                              |                              |      |                                |                              |      |                                  |
|------------------------------|------|------------------------------|------------------------------|------|--------------------------------|------------------------------|------|----------------------------------|
| <input type="checkbox"/> 714 | rash | Rashes with Hypopigmentation | <input type="checkbox"/> 804 | bg   | Increased Signal Basal Ganglia | <input type="checkbox"/> 901 | his  | Abnormal Histology               |
| <input type="checkbox"/> 715 | htii | Hyper Trichosis              | <input type="checkbox"/> 805 | dmy  | Delayed Myelination            | <input type="checkbox"/> 902 | em   | Abnormal Ultrastructure          |
| <input type="checkbox"/> 716 | alp  | Alopecia                     | <input type="checkbox"/> 806 | cea  | Cerebellar Atrophy             | <input type="checkbox"/> 903 | enz  | Abnormal Respiratory Enzymes     |
| <input type="checkbox"/> 717 | ac   | Acrocyanosis                 | <input type="checkbox"/> 807 | pstk | Posterior Stroke               | <input type="checkbox"/> 904 | prol | Large Mitochondria/Proliferation |
| <input type="checkbox"/> 718 | ak   | Angiokeratoma                | <input type="checkbox"/> 808 | leuk | Leukodystrophy                 | <input type="checkbox"/> 905 | cox  | COX Deficiency                   |
| <input type="checkbox"/> 719 | ic   | Ichthyosis                   | <input type="checkbox"/> 809 | mrs1 | MRS/Lactate Peak               | <input type="checkbox"/> 906 | rrf  | Ragged Red Fibers                |
|                              |      |                              | <input type="checkbox"/> 810 | mri  | Abnormal MRI                   |                              |      |                                  |

### SAMPLE SPECIFICATIONS TABLE

ABBREVIATION	SAMPLE NAME	RECOMMENDED AMOUNT		SHIPPING INSTRUCTIONS	SPECIAL NOTES
		(2 YRS - ADULT)	(NEWBORN - 2YRS)		
BE	Blood in EDTA (purple-top)	3 - 5 cc	3 - 5 cc	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	
CB	Cord Blood	N/A	1 - 2 cc	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	Ensure properly labeled. Also send 3 cc of maternal blood in properly labeled EDTA tube for MCC studies at no charge as needed.
DNA	DNA, Extracted	10 - 15 µ	10 - 15 µ	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	Minimal concentration of 50ng/µ; A260/A280 of ~1.7
L	Liver	50 mg	50 mg	Ship frozen sample in insulated container, with 3 - 5 lbs dry ice, by overnight courier.	Liver should be flash frozen in liquid nitrogen at collection with no media added and stored at -80°C.
SA	Saliva	See Special Notes	See Special Notes	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	Collected with Oragene DNA Self-Collection Kit.
SFC	Skin Fibroblast Culture	(3) T25 flasks	(3) T25 flasks	Ship at ambient temperature in an insulated container by overnight courier.	Send three (3) T25 flasks at approximately 60-80% confluence.
SM	Skeletal Muscle	150 mg	150 mg	Ship frozen sample in insulated container, with 3 - 5 lbs dry ice, by overnight courier.	Skeletal Muscle should be flash frozen in liquid nitrogen at collection with no media added, and stored at -80°C. <b>Surgical pathology report required.</b> If a pathology report is not available at this time, please send a clinical summary and the results of any pertinent ancillary testing.
T	Tissue	50 mg	50 mg	Ship frozen sample in insulated container, with 3 - 5 lbs dry ice, by overnight courier.	Tissue should be flash frozen in liquid nitrogen at collection with no media added, and stored at -80°C.



## INFORMED CONSENT FOR MITOCHONDRIAL TESTING

\_\_\_\_\_  
Patient Last Name                      Patient First Name                      MI                      Date of Birth (MM / DD / YYYY)                      Biological Sex

### INFORMED CONSENT FOR GENETIC TESTING

#### TEST INFORMATION

This consent form will provide you with information regarding genetic testing, which you should discuss with your healthcare provider or a genetic counselor. In order to ensure that you have understood the purpose and significance of genetic testing, we have provided information about the testing process and potential results below.

The purpose of genetic testing is to identify the cause of a suspected disease in you or your family. The testing analyzes your genetic material (DNA) for an abnormal change (variant) that could explain the disease you or members of your family are experiencing. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic, and results may require additional testing.

The purpose of this test is to see if you or your child may have a genetic variant or chromosome rearrangement. This may cause a genetic disorder or may determine the chance that you or your child will develop or pass on a genetic disorder in the future. "Your child" can also mean your unborn child, for the purposes of this consent.

In a genetic test, depending on the case, you can be tested for:

- A single gene/variant responsible for a specific, suspected genetic disease.
- Multiple genes in parallel.

The sample/specimen that is needed to perform the genetic test is stated in the test order form and is typically blood or purified DNA, but may also be tissue, saliva or buccal swab.

#### RESULTS

There are several categories of test results that may be reported including:

- **Positive:** Positive or "abnormal" results mean there is a change in the genetic material found that is related to your/your child's medical issues or that you/your child are at an increased risk of developing the disorder in the future. It is possible to test positive for more than one genetic variant.
- **Negative:** Negative or "normal" results mean no relevant genetic change related to your/your child's medical issues was detected. This does not mean there is no genetic change, but it may mean that the type of testing performed could not detect it.
- **Results of Unclear Significance:** Testing can detect change(s) in DNA which we do not yet fully understand. These alterations are also referred to as variants of uncertain significance (VUS). Additional studies may be recommended if a VUS is identified in a gene that may be associated with your/your child's medical concerns.
- **Secondary / Incidental Findings:** Testing can sometimes detect a change in a person's DNA unrelated to the reason for testing. If this change has medical or reproductive significance, it is called a secondary or incidental finding.

#### CONSIDERATIONS AND LIMITATIONS

- Results may indicate affected status, increased risk to someday be affected with, and/or reproductive risk for a genetic disorder. It is important to understand that genetic tests, even if negative, are not exhaustive. It is not possible to exclude risks for all possible genetic diseases for yourself and your family members.
- A positive test result is an indication that the individual(s) being tested may be predisposed to or have the specific disease or condition which prompted testing. You might consider additional independent testing, consult a personal physician, or pursue genetic counseling.
- It is possible that the knowledge of the test results may result in psychological stress for you and your family. It is always recommended to discuss the results with your healthcare provider or genetic counselor.
- If several family members are tested, the correct interpretation of the results depends on the provided relationships between family members. In rare cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. If a discrepancy is identified, it may be necessary to report this to the physician who ordered the testing.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to, mislabeled samples, inaccurate reporting of clinical/medical information, or rare technical errors.
- If you sign this consent form, but you no longer wish to have your sample(s) tested, you can contact your physician to cancel the test. If testing is complete, but you have not received your results yet, you can inform your physician that you no longer wish to receive the results. If you withdraw consent for testing after 5pm CST the next business day following sample receipt by the laboratory, you will be charged for the full cost of the test.

#### PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION

- Results will only be released to a licensed healthcare provider, to those allowed access to test results by law, and to those authorized in writing.
- In rare cases, persons with genetic diagnoses have experienced problems with insurance coverage and employment. The U.S. Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, you can visit [www.genome.gov/10002077](http://www.genome.gov/10002077).
- Samples will be retained in the laboratory in accordance with the laboratory retention policy.
- After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring health care providers unless specific prior arrangements have been made.

