BAYLOR GENETICS

BAYLOR GENETICS
2450 HOLCOMBE BLVD.
SUITE 2210
HOUSTON, TX 77021-2024

PHONE
1.800.411.4363
FAX
1.800.434.9850

CONNECT

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

Genetic 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)

Patient's Relationship to Insured | Phone of Insured

Address of Insured

City | State | Zip

Primary Insurance Co. Name | Primary Insurance Co. Phone

Primary Member Policy # | Primary Member Group #

Name of Insured | Insured Date of Birth (MM / DD / YYYY)

Patient's Relationship to Insured | Phone of Insured

Address of Insured

City | State | Zip

Secondary Insurance Co. Name | Secondary Insurance Co. Phone

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)

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ETHNICITY

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

SAMPLE

SAMPLE TYPE	DATE OF COLLECTION (MM/DD/YYYY)
◯ Blood in EDTA (Purple-top)	___ / ___ / ___
◯ Cord Blood	___ / ___ / ___
◯ DNA, Extracted from:	___ / ___ / ___
◯ Liver	___ / ___ / ___
◯ Saliva	___ / ___ / ___
◯ Skin Fibroblast Culture	___ / ___ / ___
◯ Skeletal Muscle	___ / ___ / ___
◯ Skin Biopsy†	___ / ___ / ___
◯ 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

1. Attach a copy of the Proband test results.
2. 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 *
☐ 2085	Dual Genome Panel by Massively Parallel Sequencing (BCM-MitomeNGS℠)	BE, DNA, T, SFC
☐ 20600	Dual Genome Leigh Disease Panel by Massively Parallel Sequencing (BCM-MitomeNGS℠)	BE, DNA, SFC, BUC, SA
☐ 2055	Comprehensive mtDNA by Massively Parallel Sequencing (BCM-MitomeNGS℠)	BE, DNA, T, SFC

MASSIVELY PARALLEL SEQUENCING (BCM-MITOMENGS℠) PANELS

TEST CODE	TEST NAME	SAMPLE TYPE *
☐ 20100	Albinism Panel (13 genes)	BE, DNA, SFC, BUC, SA
☐ 20400	Bardet-Biedl Syndrome Panel (18 genes)	BE, DNA, SFC, BUC, SA
☐ 2105	Cholestasis Panel (7 genes)	BE, DNA, SFC, BUC, SA
☐ 2120	Cobalamin Metabolism Panel + Severe *MTHFR* Deficiency (20 genes)	BE, DNA, SFC, BUC, SA
☐ 2625	*COL1A1* and *COL1A2* Panel	BE, DNA, SFC, BUC, SA

* This sample type incurs an additional fee and typically adds 14 days to the turnaround time, depending on sample quality.
† Baylor Genetics will store this sample for up to 14 days after the report is issued, allowing for follow-up testing if needed.

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MITOCHONDRIAL TESTS

MASSIVELY PARALLEL SEQUENCING (BCM-MITOMENGS℠) PANELS

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

DNA COPY NUMBER ANALYSIS

	TEST CODE	TEST NAME	SAMPLE TYPE *	SPECIFY GENE OF INTEREST
☐	3700	mtDNA Content (qPCR) Analysis - Skeletal Muscle**	SM	
☐	3720	mtDNA Content (qPCR) Analysis - Liver**	L	
☐	2000	MitoMet®Plus aCGH Analysis	BE	
☐	2001	Oligonucleotide Targeted Array Analysis (Single Target Gene)	BE	
☐	2003	Oligonucleotide Targeted Array Analysis (Up to 5 Target Genes)	BE	

** Skin biopsy sample type not available for this test

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MITOCHONDRIAL TESTS

MITOCHONDRIAL DNA (mtDNA) RESPIRATORY CHAIN ENZYME TESTS

	TEST CODE	TEST NAME	SAMPLE TYPE *
☐	3200	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skeletal Muscle**	SM
☐	3210	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts	SFC

MITOCHONDRIAL DNA (mtDNA) MUTATION SCREENS

	TEST CODE	TEST NAME	SAMPLE TYPE *
☐	2010	Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGS℠)	BE, DNA, SFC, T

MITOCHONDRIAL DNA (mtDNA) MUTATION SCREENS

	TEST CODE	TEST NAME	SAMPLE TYPE *
☐	3030	mtDNA Nonsyndromic Hearing Loss and Deafness Mutation Panel	BE, 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) and write in the below space(s).

Test Code | Gene | Test Name

Test Code | Gene | Test Name

Test Code | Gene | Test Name

	TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
☐	3904	*ACAD9* Comprehensive (Seq & Del/Dup Analysis)	ACAD9 Deficiency	BE, DNA, BUC, SA
☐	2219	*ATP5A1* Comprehensive (Seq & Del/Dup Analysis)	ATP5A1-Related Disorders	BE, DNA, BUC, SA
☐	3614	*TAZ* Comprehensive (Seq & Del/Dup Analysis)	Barth Syndrome (TAZ-Related Disorders)	BE, DNA, BUC, SA
☐	3179	*C10orf2 (TWINKLE)* Comprehensive (Seq & Del/Dup Analysis)	C10orf2 (TWINKLE)-Related Disorders	BE, DNA, BUC, SA
☐	3854	*CABC1(ADCK3)* Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA, BUC, SA
☐	3419	*COQ2* Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA, BUC, SA
☐	3414	*PDSS2* Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA, BUC, SA
☐	2264	*GFM1* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA, BUC, SA
☐	3649	*TSFM* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA, BUC, SA
☐	2289	*MRPS22* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA, BUC, SA
☐	2224	*C12orf65* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA, BUC, SA
☐	2324	*AARS2* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE, DNA, BUC, SA
☐	2664	*FOXRED1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	3489	*NDUFA1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	2684	*NDUFA11* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	3944	*NDUFAF1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	3539	*NDUFAF2* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	2694	*NDUFAF3* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	2704	*NDUFS1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
☐	3574	*NDUFS3* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA

** Skin biopsy sample type not available for this test

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MITOCHONDRIAL TESTING REQUISITION

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MITOCHONDRIAL TESTS

SINGLE GENE ANALYSIS

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

* Refer to Sample Specifications Table (Page 8)

Test list continued on next page

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MITOCHONDRIAL TESTS

INDIVIDUAL MITOCHONDRIAL TESTS (LISTED BY DISORDER)

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

* Refer to Sample Specifications Table (Page 8)

Indications on next page

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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	Microencephaly
☐	703	sids	SIDS/Unexplained Death
☐	704	ca	Congenital Anomalies
☐	705	dys	Dysmorphic Features
☐	706	id	Immunodeficiency
☐	707	ma	Macrocytic Anemia
☐	708		Pancytopenia/Bone Marrow Failure
☐	709	np	Neutropenia
☐	710	mc	Macrocephaly
☐	711	cf	Course Features
☐	712	sa	Skeletal Anomalies
☐	713	art	Arthritis

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CONNECT

MITOCHONDRIAL TESTING REQUISITION

Patient Last Name | Patient First Name | MI | Date of Birth (MM / DD / YYYY) | Genetic Sex

INDICATION FOR TESTING - CONTINUED (REQUIRED)

FAMILY HISTORY

- ☐ 001 mut Mutation (Attach details)
- ☐ 002 mi Evidence of Maternal Inheritance

ELECTROPHYSIOLOGY

- ☐ 801 baers Abnormal BAERS
- ☐ 802 vers Abnormal VERS
- ☐ 803 eeg Abnormal EEG

HAIR/SKIN FINDINGS

- ☐ 714 rash Rashes with Hypopigmentation
- ☐ 715 htii Hyper Trichosis
- ☐ 716 alp Alopecia
- ☐ 717 ac Acrocyanosis
- ☐ 718 ak Angiokeratoma
- ☐ 719 ic Ichthyosis

IMAGING/OTHER STUDIES

- ☐ 804 bg Increased Signal Basal Ganglia
- ☐ 805 dmy Delayed Myelination
- ☐ 806 cea Cerebellar Atrophy
- ☐ 807 pstk Posterior Stroke
- ☐ 808 leuk Leukodystrophy
- ☐ 809 mrsl MRS/Lactate Peak
- ☐ 810 mri Abnormal MRI

MUSCLE BIOPSY

- ☐ 901 his Abnormal Histology
- ☐ 902 em Abnormal Ultrastructure
- ☐ 903 enz Abnormal Respiratory Enzymes
- ☐ 904 prol Large Mitochondria/Proliferation
- ☐ 905 cox COX Deficiency
- ☐ 906 rrf Ragged Red Fibers

SAMPLE SPECIFICATIONS TABLE

ABBREVIATION	SAMPLE NAME	RECOMMENDED AMOUNT (2 YRS - ADULT)	RECOMMENDED AMOUNT (NEWBORN - 2YRS)	SHIPPING INSTRUCTIONS	SPECIAL NOTES
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.	
BUC	Buccal Swab	See Special Notes	See Special Notes	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze. Sample must arrive within 72 hours.	Collected with ORAcollect.Dx (OCD-100) self-collection kit (provided by Baylor Genetics with instructions). It is highly recommended the sample be collected by a healthcare professional.
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. **Surgical pathology report required.** 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.

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INFORMED CONSENT FOR MITOCHONDRIAL TESTING

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			/ /	

GENERAL GENETIC TESTING CONSENT

This consent form cannot be used for whole exome sequencing (WES), whole genome sequencing (WGS), biochemical testing, or Huntington disease testing. Consent forms for other tests are located at Baylor Genetics' website (https://www.baylorgenetics.com/consent/).

For the purposes of this consent, "I", "my", "you", and "your" can refer to you, your child, your unborn child, or other individual you are the legal representative of.

TEST INFORMATION

Your healthcare provider (doctor, genetic counselor, or other person with medical training) wants to order one or more tests to find a cause for your health issues. This testing can see if there is a cause for your health issues or if there is an increased chance for a health issue to happen to you or your family. Some of these tests look for changes, called variants, in a person's DNA. DNA is our genetic material. You might have testing for variants in one or more genes, specific parts of DNA that are needed for our health. Variants can also be found in other places in the genome (all of the DNA that a person has). Some tests might look for changes in proteins or analytes that cause health issues. The testing ordered will depend on your health issues as well as what is already known about you and your family's genetics. These tests may also explain health issues that your family may have. Even if this test finds the cause of your health issues, this may not help treat or manage those issues.

Before you sign this consent form, you should speak with your healthcare provider. They can help you understand this testing and what it means for your health.

TEST RESULTS

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

- **Positive:** A variant in the DNA was found that is related to your health issues or a health issue that you are at an increased risk of having in the future. These changes that cause disease are also known as pathogenic variants.
- **Negative:** No variants in the DNA were found that are related to your health issues or that would increase your risk of a health issue in the future.
- **Variant of Uncertain Clinical Significance (VUS):** A variant in the DNA was found that we do not know its effect, if any, on health. More testing may be needed for you or your family if a VUS is found that may be associated with your health issues.
- **Secondary and Incidental Findings:** Testing can sometimes find a variant in the DNA not related to the reason for testing. If this result is expected to affect your health, it is called a secondary or incidental finding.

CONSIDERATIONS AND LIMITATIONS

- You should speak with your provider before signing this consent form.
- Testing may show you have, or are at increased chance of having, a health issue. In addition, it may show that you have an increased chance of having a child with a health issue.
- Even if the variant(s) causing your health issues are found, this may not predict how these issues might progress or improve with treatment. Affected family members with the same variant might not be affected like you are.
- Depending on the results of testing, additional testing may be needed to understand any variant found. This testing might be needed for you and/or other family members.
- Variants and how they cause disease may change over time as we learn more about genetics.
- Testing, while very accurate, might not find an answer for your health issues. Reasons for this may include limited information about the genes tested or limitations of what the test can find.
- An additional sample may be needed if the first sample is not sufficient to complete testing.

PATIENT CONFIDENTIALITY AND SAMPLE RETENTION

- If several family members are tested, knowing the correct biological relationships among them is important. In rare cases, testing can show that family members are not related as expected. If this is found, we may contact the provider who ordered your testing.
- If this testing is requested to be cancelled after the order and sample are sent to the laboratory, please see our Test Cancellation Policy at www.baylorgenetics.com/cancel-test/.
- Only Baylor Genetics and its contracted partners will have access to your sample for the ordered testing. Results from testing will only be released to: (i) a licensed healthcare provider, (ii) those authorized in writing, (iii) the patient or their personal representative, and (iv) those allowed access to test results by law. You have the right to access your test results from Baylor Genetics by providing a written request. You also have the right to request raw data obtained from your sample by providing a written request or HIPAA Authorization Form.
- In rare cases, people with genetic diseases may have problems with health insurance and employment. The U.S. Federal Government has several laws that prohibit discrimination based on test results by health insurance companies and employers. These laws also prohibit unauthorized disclosure of this information. For more information, please visit www.genome.gov/10002077.
- Samples will be kept in the laboratory based on our retention policy. Once testing completes, de-identified sample may be used for test development, quality assurance, and training purposes. Samples are not returned to patients or providers unless requested prior to testing.
- Baylor Genetics performs scientific research and may contact you to see if you would like to be involved.
- Variants found may be submitted to databases. The medical community uses these databases to collect information about how variants might cause disease to improve testing and treatment for patients. An example is ClinVar, a free, public archive of reports on human genetics. Limited clinical information may need to be shared with these databases. In rare cases, this information may be enough to allow you or your family members to be identified.
- For more information on privacy practices at Baylor Genetics, please visit www.baylorgenetics.com/privacy-practices/.

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FOR SAMPLES FROM NEW YORK STATE RESIDENTS

Samples from New York State residents shall not be included in research without written consent. Samples will not be retained for more than sixty (60) days after receipt by Baylor Genetics, unless authorized by marking below. No tests other than those authorized shall be performed on the samples.

☐ I authorize Baylor Genetics to retain sample(s) longer based on our retention policy for test development, quality assurance, and training purposes.

FINANCIAL AGREEMENT

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. I designate Baylor Genetics as my designated representative for purposes of appealing any denial of benefits by my insurance carrier. I irrevocably assign associated payment to Baylor Genetics, and direct that payment be made directly to Baylor Genetics. Please note, some payers may not cover certain screening tests.

If my health insurer does not cover the test or I do not have health insurance, I have received a good faith estimate of the cost for the genetic testing ordered by my provider and agree to pay for the cost of the genetic testing billed to me by Baylor Genetics based on that good faith estimate. More information is available in Baylor Genetics' No Surprises Act and Good Faith Estimate Notice located at https://www.baylorgenetics.com/no-surprises-act/.

A Medicare Advance Beneficiary Notice (ABN) is required for services Medicare identifies as not medically necessary.

PATIENT AUTHORIZATION

By signing this statement of consent, I acknowledge that I have read, understand, and hereby grant my informed consent for genetic testing. I have received appropriate explanations from my healthcare provider about the planned genetic test(s) and possible results. I have been informed by my healthcare provider about the availability and importance of genetic counseling and have been provided with written information identifying a genetic counselor or medical geneticist who can provide such counseling services. All my questions have been answered, and I have had the necessary time to make an informed decision about the genetic test(s).

I hereby give permission to Baylor Genetics to conduct genetic testing as recommended by my physician*.

Patient Name	Patient's Signature	Date Signed (MM / DD / YYYY)
		/ /

Patient's Parent / Personal Representative* Name	Patient's Parent / Personal Representative Signature	Date Signed (MM / DD / YYYY)
		/ /

*If you are signing on behalf of the patient as the parent(s) and/or person with legal authority to act on behalf of the patient or parent, you may be required to provide evidence of your authority.