BAYLOR GENETICS

BAYLOR GENETICS
2450 HOLCOMBE BLVD.
GRAND BLVD. RECEIVING DOCK
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 Code | Gene | Test Code | Gene

Test Name | Test Name | 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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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

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

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. To assist you in understanding the reason for this testing, we have provided information about the testing process and potential results below.

The purpose of genetic testing is to determine if a genetic disease may be present or if there is an increased risk for a genetic disease to occur in a patient or their family. DNA is the genetic material that we receive from our parents. Genes are made of DNA and are the instructions for maintaining the health of our body. Each person has a unique set of DNA and most of the differences in our DNA do not impact our health. Genetic testing analyzes DNA to find any abnormal changes (mutations also called variants) that might cause disease, make it more likely to develop disease, and/or increase the chance of having a child affected by disease.

The testing ordered by your healthcare provider can determine if you or your child have a variant associated with a genetic disease. "Your child" can also mean your unborn child, for the purposes of this consent.

Depending on why genetic testing is needed, you might be tested for:

- A known variant that has already been found in your family
- A single gene or variant that causes a specific, suspected disease.
- Multiple genes at the same time. These genes might cause similar diseases or might cause diseases that are unrelated to each other.
- Multiple types of testing that each test for different variants.

RESULTS

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

- Positive: Positive or "abnormal" results mean there is a change in the DNA found that is related to your/your child's medical issues or that you/your child are at an increased risk of developing a disease in the future. It is possible to test positive for more than one variant. Positive results might include pathogenic variants (variants known to be associated with disease) and likely pathogenic variants (variants that are likely to be associated with disease).
- Negative: Negative or "normal" results mean no relevant variants related to your/your child's medical issues were detected or that you/your child are not expected to be at an increased risk for developing a disease in the future. This might indicate that there are no variants associated with disease in the gene(s) tested. Genetic testing, while highly accurate, might not detect a variant present in the gene(s) tested. This can be due to limitations of the information available about the gene(s) being tested, or limitations of the testing technology.
- Variant of Uncertain Significance: Testing can detect variant(s) in DNA which we do not yet fully understand. These are also referred to as variants of uncertain significance (VUS). Additional testing may be recommended for you or your family if a VUS is identified in a gene that may be associated with your/your child's medical condition.
- Secondary / Incidental Findings: Testing can sometimes detect a variant in a person's DNA unrelated to the reason for testing. If this variant is expected to have medical or reproductive significance, it is called a secondary or incidental finding.

CONSIDERATIONS AND LIMITATIONS

- This consent form cannot be used for whole exome sequencing (WES), whole genome sequencing (WGS), or Huntington's disease testing. These tests have specific consents that are located at https://www.baylorgenetics.com/consent/.
- Results may indicate you have a genetic disease, are at increased risk to develop a genetic disease, and/or be at an increased risk to have a child with a genetic disease. It is important to understand that genetic tests, even if negative, cannot rule out every variant. It is not possible to exclude risks for all genetic diseases for you and your family members.
- Depending on the type of genetic testing performed and the results, additional genetic testing or other testing may be needed to fully understand the likelihood of your developing the disease or the severity of the disease. This additional testing might be needed for you/your child or other members of your family.
- It is recommended that you discuss genetic testing with your healthcare provider or genetic counselor before signing this consent and again after results are made available.
- It may not always be possible to complete testing. as sometimes the sample does not have enough DNA to perform testing or other reasons. In these cases, another sample may need to be sent to the laboratory to perform testing.

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION

- If several family members are tested, the correct interpretation of the results depends on the information provided about the relationships amongst 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 difference is identified, it may be necessary to share this information with the healthcare provider who ordered the testing.

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PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION (CONT.)

- Genetic testing is highly accurate, however in rare cases, inaccurate results may occur. Reasons for this 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 the healthcare provider who ordered the test to cancel the test. If you wish to cancel testing, the laboratory must be notified of the cancellation request before 5 PM CST the business day after the sample has begun testing. If the laboratory is not notified of your cancellation request until after this time, you will be charged for the full cost of the test.
- Only Baylor Genetics and Baylor Genetics contracted partners will have access to the sample(s) provided to conduct the requested testing. Results will only be released to the following person(s): (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. I understand that I have the right to access any test results directly from Baylor Genetics by providing a written request. I also understand that laboratory raw data, while not routinely released as part of the testing process, can be requested by providing a written request or HIPAA Authorization Form.
- 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.
- 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 heath care providers unless specific prior arrangements have been made.
- Samples from residents of New York State will not be included in research studies without your written consent and will not be retained for more than 60 days after receipt of the sample. No tests other than those authorized shall be performed on the biological sample.
- By signing this consent form, I understand and agree that variants identified may also be submitted to public databases, such as ClinVar. Such submission serves to contribute knowledge to the medical community. I understand that limited clinical information is also required for the submission of information to ClinVar's database and further that the contents of this limited clinical information may, although unlikely, include information that may identify me personally.
- It is possible that even if the test identifies the underlying genetic cause for the disease in your family, this information may not help in predicting the progression of disease or change management or treatment of disease.

FINANCIAL AGREEMENT AND GUARANTEE

By signing this consent form, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider. For insurance billing, I hereby authorize Baylor Genetics to bill my health insurance plan on my behalf, and further authorize Baylor Genetics to release any information to my insurance carrier which is reasonably required for billing. I additionally 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. I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by Baylor Genetics as part of a verification of benefits investigation. I agree to be financially responsible for all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for unpaid services performed by Baylor Genetics on my behalf, I agree to endorse the insurance check as appropriate and forward such check to Baylor Genetics within thirty (30) days of receipt thereof, as payment towards Baylor Genetics' claim for services rendered. If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by Baylor Genetics.

I understand that a completed Advance Beneficiary Notice (ABN) is required for Medicare patients if the service is deemed not medically necessary.

RECONTACT FOR RESEARCH CONSENT

Baylor Genetics participates in research relating to health, disease prevention, drug development, and other scientific purposes. Baylor Genetics may contact patients or their provider(s) directly as part of this research. I agree to allow Baylor Genetics to contact me or my provider(s) about possible research involving the sample(s) and/or information associated with this testing. I understand that patients generally receive no compensation for this participation in research. For more information on research at Baylor Genetics, please visit baylorgenetics.com.

If I wish to opt out of being recontacted for research purposes by Baylor Genetics, I understand that I may check the box below:

☐ Please do not contact me regarding any research that uses information obtained from this testing.

For any research I may be contacted about, I prefer contact through the following methods (please check all that apply – if no choices are selected, contact will be made via secure email if possible):

☐ Email ☐ Phone ☐ Mail

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			____ / ____ / ____	

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

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

Relationship of Personal Representative to the Patient	Ordering Provider's Signature	Date (MM / DD / YYYY)
		____ / ____ / ____

*If you are signing as a person with legal authority to act on behalf of the patient, you may be required to provide evidence of your authority.