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

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)

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	___ / ___ / ___
◯ 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.

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):

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

MITOCHONDRIAL TESTS

MITOCHONDRIAL PANELS

TEST CODE	TEST NAME	SAMPLE TYPE *
☐ 2085	Dual Genome Panel by Massively Parallel Sequencing (BCM-MitomeNGS^{SM})	BE , DNA, SFC, SM
☐ 20600	Dual Genome Leigh Disease Panel by Massively Parallel Sequencing (BCM-MitomeNGS^{SM})	BE, DNA
☐ 2055	Comprehensive mtDNA by Massively Parallel Sequencing (BCM-MitomeNGS^{SM})	BE, DNA, L, SM, T

MASSIVELY PARALLEL SEQUENCING (BCM-MITOMENGS^{SM}) PANELS

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

* Refer to Sample Specifications Table (Page 8)

Test list continued on next page

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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
☐	2100	CoQ10 Deficiency Panel (*PDSS1*, *PDSS2*, *COQ2*, *COQ9*, and *ADCK3*(*COQ8/CABC1*))	BE, DNA
☐	5260	Developmental Glaucoma Panel (8 genes)	BE, DNA
☐	5250	Familial Exudative Vitreoretinopathy Panel (*FZD4*, *LRP5*, *NDP*, and *TSPAN12*)	BE, DNA
☐	2095	Fatty Acid Oxidation Panel (20 genes)	BE, DNA
☐	2125	Glycogen Storage Disease (GSD) Panel (23 genes)	BE, DNA
☐	2126	Glycogen Storage Disease (GSD) Muscle Panel (13 genes)	BE, DNA
☐	2127	Glycogen Storage Disease (GSD) Liver Panel (13 genes)	BE, DNA
☐	2200	High Bone Mass Panel (14 genes)	BE, DNA
☐	21700	Hyperinsulinism Panel (8 genes)	BE, DNA
☐	21000	Hypoglycemia Panel (85 genes)	BE, DNA
☐	5090	Leber Congenital Amaurosis Panel (19 genes)	BE, DNA
☐	20601	Leigh Disease Panel (82 genes)	BE, DNA
☐	2090	Low Bone Mass Panel (23 genes)	BE, DNA
☐	32870	Maple Syrup Urine Disease (MSUD) Panel (*BCKHDA*, *BCKHDB*, *DBT* and *DLD*)	BE, DNA
☐	21900	Maturity-Onset Diabetes of the Young (MODY) Panel (25 genes)	BE, DNA
☐	2130	mtDNA Depletion/Integrity Panel (19 genes)	BE, DNA
☐	2155	Mitochondrial Respiratory Chain Complex I Deficiency Panel (21 genes)	BE, DNA
☐	2160	Mitochondrial Respiratory Chain Complex II Deficiency Panel (*SDHA*, *SDHB*, *SDHC*, *SDHD*, and *SDHAF1*)	BE, DNA
☐	2165	Mitochondrial Respiratory Chain Complex III Deficiency Panel (*BCS1L*, *TTC19*, *UQCRB*, and *UQCRQ*)	BE, DNA
☐	2170	Mitochondrial Respiratory Chain Complex IV Deficiency Panel (10 genes)	BE, DNA
☐	2175	Mitochondrial Respiratory Chain Complex V Deficiency Panel (*ATPAF2*, *ATP5E*, and *TMEM70*)	BE, DNA
☐	2086	Nuclear Panel (162 genes)	BE, SFC, DNA, SM
☐	2180	Mitochondrial Respiratory Chain Complex I-V Panel (50 genes)	BE, DNA
☐	2300	Myopathy/Rhabdomyolysis Panel (25 genes)	BE, DNA
☐	20200	Nephronophthisis Panel (*NPHP1*, *INVS*, *NPHP3*, *NPHP4*)	BE, DNA
☐	21400	Noonan Spectrum Disorders Panel (12 genes)	BE, DNA
☐	2185	PDH & Mitochondrial RC Complex V Panel (9 genes)	BE, DNA
☐	22100	Peroxisomal Disorders Panel (22 genes)	BE, DNA
☐	5255	Primary Open Angle Glaucoma Panel (MYOC, OPTN)	BE, DNA
☐	5274	Proximal Urea Cycle Disorders Comprehensive (Seq. & Del/Dup) (*CPS1*, *NAGS*, *OTC*)	BE, DNA
☐	2140	Progressive External Ophthalmoplegia Panel (10 genes)	BE, DNA
☐	2190	Retinitis Pigmentosa + RPGR orf15 by NGS (66 genes)	BE, DNA
☐	2110	Urea Cycle Disorders and Hyperammonemia (8 genes)	BE, DNA
☐	2195	Usher Syndrome Panel (9 genes)	BE, DNA

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, DNA	
☐	2001	Oligonucleotide Targeted Array Analysis (Single Target Gene)	BE	
☐	2003	Oligonucleotide Targeted Array Analysis (Up to 5 Target Genes)	BE	

* Refer to Sample Specifications Table (Page 8)

Test list continued on next page

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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, SM, 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
☐	2889	*ACACA* Comprehensive (Seq & Del/Dup Analysis)	Acetyl-CoA Carboxylase Deficiency (ACACA-Related Disorders)	BE
☐	20520	*AIFM1* Sequence Analysis	AIFM1 Related Disorders	BE, DNA
☐	29005	*APTX* Sequence Analysis by NGS	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	BE, DNA
☐	2219	*ATP5A1* Comprehensive (Seq & Del/Dup Analysis)	ATP5A1-Related Disorders	BE
☐	3614	*TAZ* Comprehensive (Seq & Del/Dup Analysis)	Barth Syndrome (TAZ-Related Disorders)	BE
☐	3179	*C10orf2 (TWINKLE)* Comprehensive (Seq & Del/Dup Analysis)	C10orf2 (TWINKLE)-Related Disorders	BE
☐	3854	*CABC1(ADCK3)* Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE
☐	3419	*COQ2* Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE
☐	3414	*PDSS2* Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE
☐	4800	Coenzyme Q10 Analyte Analysis - Skeletal Muscle	Coenzyme Q10 Deficiency	SM
☐	2264	*GFM1* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE
☐	3649	*TSFM* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE
☐	2289	*MRPS22* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE
☐	2224	*C12orf65* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE
☐	2324	*AARS2* Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidative Phosphorylation Deficiency	BE
☐	20555	*MRPL3* Sequence Analysis	Combined Oxidative Phosphorylation Deficiency 9	BE, DNA
☐	20565	*MTO1* Sequence Analysis	Combined Oxidative Phosphorylation Deficiency 10	BE, DNA
☐	20540	*EARS2* Sequence Analysis	Combined Oxidative Phosphorylation Deficiency 12	BE, DNA
☐	2664	*FOXRED1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3489	*NDUFA1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	2684	*NDUFA11* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3944	*NDUFAF1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3539	*NDUFAF2* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	2694	*NDUFAF3* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	2704	*NDUFS1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3574	*NDUFS3* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE

* Refer to Sample Specifications Table (Page 8)

Test list continued on next page

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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
☐	3569	*NDUFS6* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3849	*NDUFS8* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3594	*NDUFV1* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	2714	*NUBPL* Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE
☐	3180	*SDHA* Sequence Analysis	Complex II Deficiency	BE, SA
☐	3185	*SDHB* Sequence Analysis	Complex II Deficiency	BE, SA
☐	3190	*SDHC* Sequence Analysis	Complex II Deficiency	BE, SA
☐	3195	*SDHD* Sequence Analysis	Complex II Deficiency	BE, SA
☐	3679	*SDHAF1* Comprehensive (Seq & Del/Dup Analysis)	Complex II Deficiency	BE
☐	3114	*BCS1L* Comprehensive (Seq & Del/Dup Analysis)	Complex III Deficiency	BE
☐	2719	*TTC19* Comprehensive (Seq & Del/Dup Analysis)	Complex III Deficiency	BE
☐	2734	*COX4I1* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3104	*COX10* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3549	*COX15* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3244	*LRPPRC* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3099	*SCO1* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3094	*SCO2* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3089	*SURF1* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	2749	*TACO1* Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE
☐	3294	*ATP5E* Comprehensive (Seq & Del/Dup Analysis)	Complex V Deficiency	BE
☐	3739	*TMEM70* Comprehensive (Seq & Del/Dup Analysis)	Complex V Deficiency	BE
☐	3344	*TIMM8A* Comprehensive (Seq & Del/Dup Analysis)	Deafness-Dystonia-Optic Neuropathy	BE
☐	3079	*DGUOK* Comprehensive (Seq & Del/Dup Analysis)	DGUOK-Related Disorders	BE
☐	3749	*ETHE1* Comprehensive (Seq & Del/Dup Analysis)	Ethylmalonic Encephalopathy	BE
☐	2249	*FARS2* Comprehensive (Seq & Del/Dup Analysis)	FARS2-Related Disorders	BE
☐	3559	*FASTKD2* Comprehensive (Seq & Del/Dup Analysis)	FASTKD2-Related Disorders	BE
☐	2314	*HARS2* Comprehensive (Seq & Del/Dup Analysis)	HARS2-Related Disorders	BE
☐	2329	*KARS* Comprehensive (Seq & Del/Dup Analysis)	Intermediate Charcot-Marie-Tooth Neuropathy, KARS-Related	BE
☐	2269	*ACAT1* Comprehensive (Seq & Del/Dup Analysis)	Ketothiolase Deficiency	BE
☐	20585	*SIRT3* Sequence Analysis	Li-Fraumeni Syndrome with Brain Tumor	BE, DNA
☐	3464	*DLD* Comprehensive (Seq & Del/Dup Analysis)	Maple Syrup Urine Disease Type 3	BE
☐	2229	*MARS2* Comprehensive (Seq & Del/Dup Analysis)	MARS2 Related Disorders	BE
☐	20550	*MFN2* Sequence Analysis	MFN2 Related Disorders	BE, DNA
☐	20570	*NDUFV2* Sequence Analysis	Mitochondrial Complex I Deficiency	BE, DNA
☐	20525	*ATP5O* Sequence Analysis	Mitochondrial Complex V Deficiency - ATP5O Related	BE, DNA
☐	20615	*UQCRC2* Sequence Analysis	Mitochondrial Complex III Deficiency Nuclear Type 5	BE, DNA
☐	20620	*UQCR10* Sequence Analysis	Mitochondrial Complex III Deficiency - UQCR10 Related	BE, DNA
☐	20560	*MTHFD1L* Sequence Analysis	Mitochondrial Disorders - MTHFD1L Related	BE, DNA

* Refer to Sample Specifications Table (Page 8)

Test list continued on next page

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MITOCHONDRIAL TESTING REQUISITION

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MITOCHONDRIAL TESTS

INDIVIDUAL MITOCHONDRIAL TESTS (LISTED BY DISORDER)

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
☐ 20575	*POLRMT* Sequence Analysis	Mitochondrial Disorders - POLRMT Related	BE, DNA
☐ 20590	*SIRT5* Sequence Analysis	Mitochondrial Disorders - SIRT5 Related	BE, DNA
☐ 20595	*TOP1MT* Sequence Analysis	Mitochondrial Disorders - TOPMT Related	BE, DNA
☐ 20610	*TRIT1* Sequence Analysis	Mitochondrial Disorders - TRIT1 Related	BE, DNA
☐ 3964	*SUCLG2* Comprehensive (Seq & Del/Dup Analysis)	mtDNA Depletion Syndrome, SUCLG2-Related	BE
☐ 3074	*TK2* Comprehensive (Seq & Del/Dup Analysis)	mtDNA Depletion Syndrome, Myopathic Form (TK2-Related Disorders)	BE
☐ 29015	*FBXL4* Sequence Analysis by NGS	mtDNA Depletion Syndrome I3, Encephalomyopathic type	BE, DNA
☐ 3064	*TYMP* Comprehensive (Seq & Del/Dup Analysis)	MNGIE/MNGIE like Syndrome	BE
☐ 3324	*MPV17* Comprehensive (Seq & Del/Dup Analysis)	MPV17-Related Disorders	BE
☐ 2294	*MRPL44* Comprehensive (Seq & Del/Dup Analysis)	MRPL44-Related Disorders	BE
☐ 2235	*MTFMT* Sequence Analysis	MTFMT-Related Disorders	BE, SA
☐ 3659	*ISCU* Comprehensive (Seq & Del/Dup Analysis)	Myopathy with Deficiency of ISCU	BE
☐ 3654	*PUS1* Comprehensive (Seq & Del/Dup Analysis)	Myopathy, Mitochondrial, and Sideroblastic Anemia	BE
☐ 3959	*YARS2* Comprehensive (Seq & Del/Dup Analysis)	Myopathy, Mitochondrial, and Sideroblastic Anemia	BE
☐ 29010	*GFER* Sequence Analysis by NGS	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	BE, DNA
☐ 2309	*NARS2* Comprehensive (Seq & Del/Dup Analysis)	NARS2-Related Disorders	BE
☐ 33465	*OPA1* Sequence Analysis by NGS	Optic Atrophy Type 1	BE, SA
☐ 3529	*OPA3* Comprehensive (Seq & Del/Dup Analysis)	Optic Atrophy Type 3	BE
☐ 3169	*PDHA1* Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE
☐ 3899	*PDHB* Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE
☐ 3894	*PDP1* Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE
☐ 3924	*PDHX* Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE
☐ 3919	*DLAT* Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE
☐ 3069	*POLG* Comprehensive (Seq & Del/Dup Analysis)	POLG-Related Disorders	BE
☐ 3384	*POLG2* Comprehensive (Seq & Del/Dup Analysis)	POLG2-Related Disorders	BE
☐ 3754	*PC* Comprehensive (Seq & Del/Dup Analysis)	Pyruvate Carboxylase Deficiency	BE
☐ 20530	*COX6A1* Sequence Analysis	Recessive Intermediate D Charcot-Marie-Tooth Disease	BE, DNA
☐ 3424	*RRM2B* Comprehensive (Seq & Del/Dup Analysis)	RRM2B-Related Disorders	BE
☐ 20580	*SIRT1* Sequence Analysis	SIRT1 Related Disorders	BE, DNA
☐ 3174	*SLC25A4 (ANT1)* Comprehensive (Seq & Del/Dup Analysis)	SLC25A4-Related Disorders	BE
☐ 5335	*SPG7* Sequence Analysis	Spastic Paraplegia 7, Autosomal Recessive	BE, SA
☐ 3379	*SUCLA2* Comprehensive (Seq & Del/Dup Analysis)	SUCLA2-Related Disorders	BE
☐ 3394	*SUCLG1* Comprehensive (Seq & Del/Dup Analysis)	SUCLG1-Related Disorders	BE
☐ 20545	*GFM2* Sequence Analysis	Wolcott-Rallison Syndrome	BE, DNA
☐ 20535	*DNAJC19* Sequence Analysis	3-Methyglutaconic Aciduria Type V	BE, DNA

* 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	pcbm	Pancytopenia/Bone Marrow Failure
☐	709	np	Neutropenia
☐	710	mc	Macrocephaly
☐	711	cf	Course Features
☐	712	sa	Skeletal Anomalies
☐	713	art	Arthritis

BAYLOR GENETICS

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CONNECT

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

☐	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.	
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. **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)	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.
- 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.

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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

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION (CONT.)

- Samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by your selection. No tests other than those authorized shall be performed on the biological sample.
- Information including results, indications for testing and clinical status obtained from this testing may be shared with healthcare providers, scientists and healthcare databases or used in scientific publications or presentations, but the personal identifying information of all persons studied will not be revealed in such data sharing or publications/presentations.

RESEARCH & RECONTACT CONSENT

For more information on research at Baylor Genetics, please visit baylorgenetics.com. Please read the below statements carefully and check the appropriate box.

Note: If left blank, consent is interpreted as "NO."

☐ I agree to use of my de-identified specimen for research to improve genetic testing for all patients and contribute to scientific research.

☐ I am a New York State Resident, and I give Baylor Genetics permission to store my specimen in accordance to the laboratory retention policy for internal quality assurance and possible research studies.

☐ In addition to agreeing above, I agree to be contacted by Baylor Genetics regarding research opportunities.

PATIENT AUTHORIZATION

By signing this statement of consent, I acknowledge that I have read and understand the informed consent for genetic testing. I have received appropriate explanations from my physician regarding the purpose, scope, type and significance of the planned genetic testing and achievable results. All my questions have been answered and I have had the necessary time to make an informed decision about the genetic test.

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

Patient Signature | Date (DD/MM/YYYY)

Printed Name