

PHONE 1.800.411.4363 FAX 1.800.434.9850

CONNECT





MITOCHONDRIAL TESTING REQUISITION

PATIENT INFORMATION (COMPLE	TE ONE FORM FOR EACH PERSON TESTE	D)		
				//
Patient Last Name	Patient First Nam	ne	MI	Date of Birth (MM / DD / YYYY)
Address	City	State Patient discharged from	Zip Genetic Sex:	Phone
Accession #	Hospital / Medical Record #	the hospital/facility: Yes No	Female Gender identity (if diffe	Male Unknown erent from above):
REPORTING RECIPIENTS				
Ordering Physician		Institution Name		
Email (Required for International Cli	ents)	Phone	Fax	
ADDITIONAL RECIPIENTS				
Name		Email	Fax	
Name		 Email	Fax	
PAYMENT (FILL OUT ONE OF THE	OPTIONS BELOW)			
Pay With Sample INSTITUTIONAL BILLING	Bill To Patient			
Institution Name	Institution Code In	stitution Contact Name In	stitution Phone	Institution Contact Email
O INSURANCE				
Do Not Perform Test Until	Patient is Aware of Out-Of-Pocket Costs (exclu	udes prenatal testing)		
REQUIRED ITEMS 1. Copy	of the Front/Back of Insurance Card(s) 2. ICD10	O Diagnosis Code(s) 3. Name of Ordering	g Physician 4. Insur	ed Signature of Authorization
	/ /	:		/ /
Name of Insured	Insured Date of Birth (MM / DD / YYY)	Y) Name of Insured		Insured Date of Birth (MM / DD / YYYY)
Patient's Relationship to Insured	Phone of Insured	Patient's Relationship to	Insured	Phone of Insured
Address of Insured		Address of Insured		
City	State Zip	City	<u> </u>	State Zip
Primary Insurance Co. Name	Primary Insurance Co. Phone	Secondary Insurance Co.	Name	Secondary Insurance Co. Phone
Primary Member Policy #	Primary Member Group #	Secondary Member Polic	y #	Secondary Member Group #
understand that I am responsible fo reasons including, but not limited to	ze Baylor Genetics to provide my insurance r any co-pay, co-insurance, and unmet deduct o, non-covered and non-authorized services. y in payment for this test. Please note that M	tible that the insurance policy dictates I understand that I am responsible for	, as well as any amou sending Baylor Gene	nts not paid by my insurance carrier for
				////
Patient's Printed Name	Patient's	s Signature		Date (MM / DD / YYYY)
STATEMENT OF MEDICAL NECES				
patient's medical management and	the risk assessment, diagnosis, or detection treatment decisions. The person listed as the n to the patient and they have consented to g	e Ordering Physician is authorized by l		
				//
Physician's Printed Name	Physicia	an's Signature		Date (MM / DD / YYYY)



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			/	
Patient Last Name	Patient First Name	MI	Date of Birth (MM / DD / YYYY) Ger	netic Sex
ETHNICITY				
African American	Hispanic American		Pacific Islander (Philippines, Micronesia,	Malaysia, Indonesia)
Ashkenazi Jewish	Mennonite		South Asian (India, Pakistan)	
East Asian (China, Japan, Korea)	Middle Eastern (Saudi Arabia, Qatar, Iraq,	Turkey)	Southeast Asian (Vietnam, Cambodia, T	hailand)
Finnish	Native American		Southern European Caucasian (Spain, I	taly, Greece)
French Canadian	Northern European Caucasian (Scandina)	vian, UK, Germany)	Other (Specify):	
SAMPLE		INDICATION F	FOR TESTING (REQUIRED)	
SAMPLE TYPE	DATE OF COLLECTION (MM/DD/YYYY)	Symptom	atic with Positive Family History	
Blood in EDTA (Purple-top)	/ /	Symptom	atic (Summarize below):	
Cord Blood				
O DNA, Extracted from:				
Liver				
Saliva		Asympton	natic	
Skin Fibroblast Culture	/	\circ	Population Screening Positive Family	History
Skeletal Muscle	//			
Tissue	//	Disease	Gene Varia	nt
TESTING OPTIONS Targeted Sequencing for Known Fa		MITOCHOND	RIAL TESTS RIAL PANELS	
(If selected, specify test code and gene a	nd complete section below)	TEST CODE	TEST NAME	SAMPLE TYPE *
Test Code	Gene	2085	Dual Genome Panel by Massively Parallel Sequencing (BCM-MitomeNGS SM)	BE, DNA, T, SFC
Proband Last Name	Proband First Name	20600	Dual Genome Leigh Disease Panel by Massively Parallel Sequencing (BCM-MitomeNGS SM)	BE, DNA, SFC, BUC, SA
Relationship to Proband	Date of Birth (MM/DD/YYYY)	2055	Comprehensive mtDNA by Massively Parallel Sequencing (BCM-MitomeNGS SM)	BE, DNA, T, SFC
Proband testing location (Select or	ne)			
Baylor Genetics		MASSIVELY	PARALLEL SEQUENCING (BCM-MITOMENGS SM	PANELS
		TEST CODE	TEST NAME	SAMPLE TYPE *
Lab #	Family #	20100	Albinism Panel (13 genes)	BE, DNA, SFC, BUC, SA
 Another Laboratory 1. Attach a copy of the Proba 	nd took requise	20400	Bardet-Biedl Syndrome Panel (18 genes)	BE, DNA, SFC, BUC, SA
	of the Proband is requested. Please provide, if available.	2105	Cholestasis Panel (7 genes)	BE, DNA, SFC, BUC, SA
Full Gene Sequencing		2120	Cobalamin Metabolism Panel + Severe MTHFR Deficiency (20 genes)	BE, DNA, SFC, BUC, SA
☐ Deletion/ Duplication Analysis				
		2625	COL1A1 and COL1A2 Panel	BE, DNA, SFC, BUC, SA



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				/	/				
Patient Last Na	ame Patient First Nam	ne	MI	Date of Birth (MM /	DD / YYYY)	Genetic Sex			
MITOCHONDR	IAL TESTS								
MASSIVELY F	PARALLEL SEQUENCING (BCM-MITOMENG	S SM) PANELS ······							
TEST CODE	TECT NAME					CAMPLE TYPE *			
5095	TEST NAME	24 annual				SAMPLE TYPE *			
2100	Congenital Disorders of Glycosylation Panel (CoQ10 Deficiency Panel (PDSS1, PDSS2, COQ2		CAPC1\\			BE, DNA, SFC, BUC, SA BE, DNA, SFC, BUC, SA			
5260	Developmental Glaucoma Panel (8 genes)	., COQ7, and ADCN3(COQ07)	CADCIII			BE, DNA, SFC, BUC, SA			
5250	Familial Exudative Vitreoretinopathy Panel (F	7D/ I PP5 NDP and TSPA	(/12)			BE, DNA, SFC, BUC, SA			
2095	Fatty Acid Oxidation Panel (20 genes)	2D4, LINI 3, NDI , AND 131 AI	V12)			BE, DNA, SFC, BUC, SA			
2125	Glycogen Storage Disease (GSD) Panel (23 ge	unas)				BE, DNA, SFC, BUC, SA			
2126	Glycogen Storage Disease (GSD) Muscle Pane					BE, DNA, SFC, BUC, SA			
2127	Glycogen Storage Disease (GSD) Liver Panel					BE, DNA, SFC, BUC, SA			
2200	High Bone Mass Panel (14 genes)	(13 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	3)				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 (BC	CKHDA. BCKHDB. DBT and D	OLD)			BE, DNA, SFC, BUC, SA			
21900	Maturity-Onset Diabetes of the Young (MODY)					BE, DNA, SFC, BUC, SA			
2130	mtDNA Depletion/Integrity Panel (19 genes)		BE, DNA, SFC, BUC, SA						
2155	Mitochondrial Respiratory Chain Complex I D	BE, DNA, SFC, BUC, SA							
2160									
2165									
2170									
2175	Mitochondrial Respiratory Chain Complex V D					BE, DNA, SFC, BUC, SA 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)	<u> </u>				BE, DNA, SFC, BUC, SA			
20200	Nephronophthisis Panel (NPHP1, INVS, NPHP3	3, NPHP4)				BE, DNA, SFC, BUC, SA			
24001	Noonan Spectrum Disorders Panel (26 genes	s)				BE, DNA, SFC, BUC, SA			
2185	PDH & Mitochondrial RC Complex V Panel (9 g	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 Comprehension	ve (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 (6	6 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 N	DNA COPY NUMBER ANALYSIS								
TEST CODE	TEST NAME		SAMPLE TYPE *		SPECIFY GENE OF IN	TEREST			
3700	mtDNA Content (qPCR) Analysis - Skeletal Mu	ıscle	SM						
3720	mtDNA Content (qPCR) Analysis - Liver	· · · · · ·	L			/////////////////////////////////////			
2000	MitoMet®Plus aCGH Analysis		BE	<i>HHHHHH</i>		/////////////////////////////////////			
2001	Oligonucleotide Targeted Array Analysis (Sing	gle Target Gene)	BE	<u> </u>					

* Refer to Sample Specifications Table (Page 8)

Oligonucleotide Targeted Array Analysis (Up to 5 Target Genes)

2003

Test list continued on next page

09.07.23



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MITOCHONDRIAL TESTING REQUISITION

Patient Last N	ame Patient First Name		MI	Date of Birth (MM / DD / YYYY)	Genetic Sex
MITOCHONDE	RIAL TESTS				
MITOCHOND	DIAL DNA (+DNA) DECDIDATORY CHAIN FA	ZVME TECTS			
	RIAL DNA (mtDNA) RESPIRATORY CHAIN EN	ZYME IESIS ·····			
3200	TEST NAME	aia (ETC) Chalatal Musi	-1-		SAMPLE TYPE
3210	Mitochondrial Respiratory Chain Enzyme Analy Mitochondrial Respiratory Chain Enzyme Analy				SFC
	,	(,			
MITOCHOND	RIAL DNA (mtDNA) MUTATION SCREENS	•••••••••••	MITOCHOND	RIAL DNA (mtDNA) MUTATION SCREENS	•••••
TEST CODE	TEST NAME	SAMPLE TYPE *	TEST CODE	TEST NAME	SAMPLE TYPE
2010	Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGS)		3030	mtDNA Nonsyndromic Hearing Loss and Deafness Mutation Panel	BE, SA, SM, T
	Massivety Farattet sequencing (Behr Mitoritettes	,		Beamess Matation Failet	
SINGLE GEN	E ANALYSIS				
f a test is not	found on this form, please obtain the test code fro	m our website (www.BM	IGL.com) and write i	n the below space(s).	
Test Code	Gene	Test Code	Gene	Test Code	Gene
Test Name		Test Name		 Test Name	
icst ivallic		rest Name		restriante	
TEST CODE	TEST NAME		DISORDER		SAMPLE TYPE
3904	ACAD9 Comprehensive (Seq & Del/Dup Analysi		ACAD9 Deficiency		BE, DNA, BUC, S
2219	ATP5A1 Comprehensive (Seq & Del/Dup Analys	is)	ATP5A1-Related D	Disorders	BE, DNA, BUC, S
3614	TAZ Comprehensive (Seq & Del/Dup Analysis)		Barth Syndrome (TAZ-Related Disorders)	BE, DNA, BUC, S
3179 	C10orf2 (TWINKLE) Comprehensive (Seq & Del/	Dup Analysis)	C10orf2 (TWINKLE	E)-Related Disorders	BE, DNA, BUC, S
3854	CABC1(ADCK3) Comprehensive (Seq & Del/Dup	Analysis)	Coenzyme Q10 De	eficiency	BE, DNA, BUC, S
3419	COQ2 Comprehensive (Seq & Del/Dup Analysis)		Coenzyme Q10 De	ficiency	BE, DNA, BUC, S
3414	PDSS2 Comprehensive (Seq & Del/Dup Analysi	s)	Coenzyme Q10 De	eficiency	BE, DNA, BUC, S
2264	GFM1 Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidativ	ve Phosphorylation Deficiency	BE, DNA, BUC, S
3649	TSFM Comprehensive (Seq & Del/Dup Analysis)	Combined Oxidation	ve Phosphorylation Deficiency	BE, DNA, BUC, S
2289	MRPS22 Comprehensive (Seq & Del/Dup Analys	sis)	Combined Oxidation	ve Phosphorylation Deficiency	BE, DNA, BUC, S
2224	C12orf65 Comprehensive (Seq & Del/Dup Analy	rsis)	Combined Oxidation	ve Phosphorylation Deficiency	BE, DNA, BUC, S
2324	AARS2 Comprehensive (Seq & Del/Dup Analysi	s)	Combined Oxidativ	ve Phosphorylation Deficiency	BE, DNA, BUC, S
2664	FOXRED1 Comprehensive (Seq & Del/Dup Analy	rsis)	Complex I Deficier	псу	BE, DNA, BUC, S
3489	NDUFA1 Comprehensive (Seq & Del/Dup Analys	sis)	Complex I Deficier	псу	BE, DNA, BUC, S
2684	NDUFA11 Comprehensive (Seq & Del/Dup Analy	/sis)	Complex I Deficier	ncy	BE, DNA, BUC, S
3944	NDUFAF1 Comprehensive (Seq & Del/Dup Analy	ysis)	Complex I Deficier	ncy	BE, DNA, BUC, S
3539	NDUFAF2 Comprehensive (Seq & Del/Dup Anal	ysis)	Complex I Deficier	ncy	BE, DNA, BUC, S
2694	NDUFAF3 Comprehensive (Seq & Del/Dup Anal	/sis)	Complex I Deficier	ncy	BE, DNA, BUC, S
2704	NDUFS1 Comprehensive (Seq & Del/Dup Analys	sis)	Complex I Deficier		BE, DNA, BUC, S
3574	NDUFS3 Comprehensive (Seg & Del/Dup Analys		Complex I Deficier	•	BE, DNA, BUC, S

Test list continued on next page

^{*} Refer to Sample Specifications Table (Page 8)



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MITOCHONDRIAL TESTING REQUISITION

Patient First Name	// 	Genetic Sex
TESTS	MI Date of Diff. (MIM / DD / 1111)	Genetic Sex
2515		
ALYSIS		
ST NAME	DISORDER	SAMPLE TYPE *
UFS4 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
UFS6 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
UFS8 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
UFV1 Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
BPL Comprehensive (Seq & Del/Dup Analysis)	Complex I Deficiency	BE, DNA, BUC, SA
HA Sequence Analysis	Complex II Deficiency	BE, DNA, BUC, SA
HB Sequence Analysis	Complex II Deficiency	BE, DNA, BUC, SA
HC Sequence Analysis	Complex II Deficiency	BE, DNA, BUC, SA
HD Sequence Analysis	Complex II Deficiency	BE, DNA, BUC, SA
HAF1 Comprehensive (Seq & Del/Dup Analysis)	Complex II Deficiency	BE, DNA, BUC, SA
S1L Comprehensive (Seq & Del/Dup Analysis)	Complex III Deficiency	BE, DNA, BUC, SA
C19 Comprehensive (Seq & Del/Dup Analysis)	Complex III Deficiency	BE, DNA, BUC, SA
X4I1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
X10 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
X15 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
01 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
02 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
RF1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
CO1 Comprehensive (Seq & Del/Dup Analysis)	Complex IV Deficiency	BE, DNA, BUC, SA
P5E Comprehensive (Seq & Del/Dup Analysis)	Complex V Deficiency	BE, DNA, BUC, SA
EM70 Comprehensive (Seq & Del/Dup Analysis)	Complex V Deficiency	BE, DNA, BUC, SA
1M8A Comprehensive (Seq & Del/Dup Analysis)	Deafness-Dystonia-Optic Neuropathy	BE, DNA, BUC, SA
UOK Comprehensive (Seq & Del/Dup Analysis)	DGUOK-Related Disorders	BE, DNA, BUC, SA
HE1 Comprehensive (Seq & Del/Dup Analysis)	Ethylmalonic Encephalopathy	BE, DNA, BUC, SA
RS2 Comprehensive (Seq & Del/Dup Analysis)	FARS2-Related Disorders	BE, DNA, BUC, SA
STKD2 Comprehensive (Seq & Del/Dup Analysis)	FASTKD2-Related Disorders	BE, DNA, BUC, SA
RS2 Comprehensive (Seq & Del/Dup Analysis)	HARS2-Related Disorders	BE, DNA, BUC, SA
RS Comprehensive (Seq & Del/Dup Analysis)	Intermediate Charcot-Marie-Tooth Neuropathy, KARS-Related	BE, DNA, BUC, SA
AT1 Comprehensive (Seq & Del/Dup Analysis)	Ketothiolase Deficiency	BE, DNA, BUC, SA
D Comprehensive (Seq & Del/Dup Analysis)	Maple Syrup Urine Disease Type 3	BE, DNA, BUC, SA
RS2 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 TESTING REQUISITION

		/ /	
Patient Last N	ame Patient First Name	MI Date of Birth (MM / DD / YYYY) Ger	netic Sex
MITOCHOND	RIAL 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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MITOCHONDRIAL TESTING REQUISITION

Patient Las	st Name	Patient First N	ame		MI Da	/ ate of Birth (MM / D	_	Genetic Sex
INDICATIO	N FOR TE	ESTING (REQUIRED)						
Clinica	ıl manageı	ment of known diagnosis - Please spec	ify:					
Diagno	ostic Testir	ng – Please complete checklist below.						
CENTRAL	NERVOU	S SYSTEM	VISCERAL			···· SENSO	RY	
101	dd	Developmental Delay/ ID	301	gir	Gastrointestinal Reflux	<u> </u>	rp	Retinitis Pigmentosa
102	ht	Hypotonia	302	dge	Delayed Gastric Emptying	<u> </u>	opa	Optic Atrophy
103	au	Autistic Features	303	pan	Pancreatitis	<u> </u>	cat	Cataract
104	enc	Dementia/ Encephalopathy	304	dia	Diarrhea	<u> </u>	hl	Sensorineural Hearing Loss
105	ha	Headaches/ Migraines	305	cst	Constipation	<u> </u>	trv	Tortuous Retinal Vessels
106	stk	Stroke, Ischemic Episodes	306	cv	Cyclic Vomiting	<u> </u>	crs	Cherry Red Spot/Eye
107	atx	Ataxia	307	pob	Pseudoobstruction	507	со	Corneal Opacity
		Intractable/ Refractory/	308	hpf	Hepatic Failure	 508	el	Ectopia Lentis
108	SZ	Myoclonus/Myoclonic Seizures	309	eta	Elevated Transaminases	 	pp	Photophobia
109	pi	Perinatal Insult	310	rtd	Renal Tubular Disease			
110	ps	Pyramidal Signs	311	ар	Apnea/ Hypoventilation			
111	hp	Hemiparesis	312	rsf	Respiratory Deficiency/Failure	ENDOC	RINE	• • • • • • • • • • • • • • • • • • • •
112	spas	Spasticity	313	ren	Renal Dysfunction	☐ 601	db	Diabetes
113	dyst	Dystonia	314	lc	Liver Carcinoma	☐ 602	pd	Exocrine/Pancreatic Deficiency
114	cho	Chorea	315	jau	Jaundice	☐ 602	qf	Gonadal Failure
115	sib	Self-Injury	316	spm	Splenomegaly/Enlarged Sple	=	hth	Hypothyroidism
116	pan	Pancreatitis	317	hpm	Hepatomegaly/Enlarged Liver	=	hpt	** *
117	dia	Diarrhea	317	•		=		Hypoparathyroidism
☐ 118	cst	Constipation	□ 310	hd	Hepatic Dysfunction	<u></u> 606	adr	Hypo/Hyper-adrenal Function
119	cv	Cyclic Vomiting				<u></u> 607	ss	Short Stature
120	pob	Pseudoobstruction				□ 608	adc	Adrenal Calcification
120	pob	Pseudoobstruction				∐ 609	hf	Hydrops Fetalis
						∐ 610	pg	Pregnant
NEUROMU	JSCULAR		METABOL	ITES / M	METABOLIC	···· OTHER	CLINICAL	
201	pn	Peripheral Neuropathy	<u> </u>	nbs	Abnormal Newborn Screen		ftt	Failure to Thrive
202	exi	Exercise Intolerance	401	kto	Ketosis		mce	Microencephaly
203	pmw	Progressive Muscle Weakness	<u>402</u>	dca	Dicarboxylic Aciduria	703	sids	SIDS/Unexplained Death
204	smw	Static Muscle Weakness	403	la	Lactic Acidosis		ca	Congenital Anomalies
205	cr	Muscle Cramps after Exercise	404	csfl	High CSF Lactate		dys	Dysmorphic Features
206	fat	Easy Fatigability	405	oa	Organic Aciduria	706	id	Immunodeficiency
207	dcmyo	Dilated Cardiomyopathy	406	lpc	Low Plasma Carnitine	707	ma	Macrocytic Anemia
208	hcmyo	Hypertrophic Cardiomyopathy	 407	cpk	CPK Abnormalities	708		Pancytopenia/Bone Marrow Failure
209	hb	Heart Block	408	pyr	Elevated Pyruvate	709	np	Neutropenia
210	ar	Arrhythmia	409	ala	Elevated Alanine	710	mc	Macrocephaly
211	ор	Ophthalmoparesis, CPEO	410	3mg	3-Methylglutaconic Aciduria	711	cf	Course Features
211	emg	Abnormal EMG/NCV	410	acid	Acidosis	711	sa	Skeletal Anomalies
212	_	Ptosis	411	NH3	Hypoammonemia	☐ 712 ☐ 713		
=	pto		412		**	□ /13	art	Arthritis
214	eh	Cardiomegaly/Enlarged Heart	=	hypo	Hypoglycemia			
			414 (15	hyper	Hyperglycemia			
			415	uco	Unusual Color/Odor			



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60-80% confluence.

and stored at -80°C.

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. Tissue should be flash frozen in liquid

nitrogen at collection with no media added,

CONNECT







Culture

Skeletal Muscle

Tissue

 SM

Т

MITOCHOND	RIAL TESTING RE	QUISITION	N						
							/	/	
Patient Last Name	F	atient First Nam	ne		MI	Date of B	rth (MM / DD	, / YYYY)	Genetic Sex
INDICATION FOR T	ESTING - CONTINUED (I	REQUIRED)							
FAMILY HISTORY			ELECT	ROPHYSIOL	OGY				
001 mut	Mutation (Attach details	-1	□ 80	1 baers	Abnormal BAERS				
002 mi	Evidence of Maternal In				Abnormal VERS				
002	Evidence of Maternatin	meritance	80		Abnormal EEG				
HAIR/SKIN FINDIN	NGS		IMAGII	NG/OTHER S	TUDIES		MUSCLE	BIOPSY	
714 rash	Rashes with Hypopigm	entation	□ 80	4 bg	Increased Signal Basal	Ganglia	901	his	Abnormal Histology
715 htii	Hyper Trichosis		80	5 dmy	Delayed Myelination	-	902	em	Abnormal Ultrastructure
	Alopecia		 80	6 cea	Cerebellar Atrophy		903	enz	Abnormal Respiratory Enzymes
717 ac	Acrocyanosis		 80	7 pstk	Posterior Stroke		904	prol	Large Mitochondria/Proliferation
	Angiokeratoma		 80	8 leuk	Leukodystrophy		905	cox	COX Deficiency
719 ic	Ichthyosis		 80	9 mrsl	MRS/Lactate Peak		906	rrf	Ragged Red Fibers
_	·		 81	0 mri	Abnormal MRI		_		
SAMPLE SPECIFICA	ATIONS TABLE								
		RECOM	IMENDE	AMOUNT					
ABBREVIATION	SAMPLE NAME	(2 YRS - ADUL	_T) (NEWBORN - 2YF		INSTRUCTIONS			SPECIAL NOTES
BE	Blood in EDTA (purple-top)	3 - 5 cc		3 - 5 cc	Ship at room tempe container by overnig or freeze.				
BUC	Buccal Swab	See Specia Notes	ıl	See Special Notes	Ship at room tempe container by overniq or freeze. Sample m hours.	ght courier. Do	not heat	collection with instr	with ORAcollect.Dx (OCD-100) self- n kit (provided by Baylor Genetics ructions). It is highly recommended ale be collected by a healthcare anal.
DNA	DNA, Extracted	10 - 15 μ		10 - 15 μ	Ship at room tempe container by overnig or freeze.			Minimal of ~1.7	concentration of 50ng/µ; A260/A280
L	Liver	50 mg		50 mg	Ship frozen sample with 3 -5 lbs dry ice				uld be flash frozen in liquid nitrogen ion with no media added and stored
SA	Saliva	See Specia Notes	ıl	See Special Notes	Ship at room tempe container by overnig or freeze.			Collected	with Oragene DNA Self-Collection Kit.
SFC	Skin Fibroblast	(3) T25 flask	KS	(3) T25 flasks	Ship at ambient tem	perature in a	n insulated	Send thre	ee (3) T25 flasks at approximately

150 mg

50 mg

150 mg

50 mg

container by overnight courier.

Ship frozen sample in insulated container,

with 3 -5 lbs dry ice, by overnight courier.

Ship frozen sample in insulated container,

with 3 -5 lbs dry ice, by overnight courier.



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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.

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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INFORME	D CONSENT FOR	MITOCHONDRIAL TESTIN	IG		
				/ /	
Patient Last Na	ame	Patient First Name	MI	Date of Birth (MM / DD / YYYY)	Genetic Sex
PATIENT CON	IFIDENTIALITY AND SE	PECIMEN RETENTION (CONT.) ······			
		te, however in rare cases, inaccura of clinical/medical information, or r		Reasons for this include, but are not li	mited to, mislabeled
cancel the	test. If you wish to ca	ncel testing, the laboratory must b	e notified of the cance	an contact the healthcare provider wl llation request before 5 PM CST the b Itil after this time, you will be charged	usiness day after the
will only b represent Genetics b	e released to the follo ative, and (iv) those al by providing a written	wing person(s): (i) a licensed healt lowed access to test results by law	hcare provider, (ii) thos v. I understand that I ha oratory raw data, whil	mple(s) provided to conduct the reque se authorized in writing, (iii) the patier ave the right to access any test results e not routinely released as part of the	nt or their personal s directly from Baylor
enacted s	everal laws that prohi		test results by health	e coverage and employment. The U.S insurance companies and employers www.genome.gov/10002077.	
• Samples v	will be retained in the	laboratory in accordance with the l	aboratory retention po	olicy.	
assurance		es. DNA specimens are not returne		elopment and improvement, internal eferring heath care providers unless	
		York State will not be included in r ple. No tests other than those auth		ut your written consent and will not b ned on the biological sample.	e retained for more than
submission of information	n serves to contribute	knowledge to the medical commu	nity. I understand that	submitted to public databases, such limited clinical information is also re nformation may, although unlikely, ind	quired for the submission
		t identifies the underlying genetic o ge management or treatment of dis		n your family, this information may no	t help in predicting the
FINANCIAL A	GREEMENT AND GUAR	ANTEE			
billing, I here to my insura of appealing directly to Ba part of a veri health insura to endorse th Baylor General	eby authorize Baylor G nce carrier which is re any denial of benefits aylor Genetics. I under fication of benefits invance plan. If my insura ne insurance check as tics' claim for services	enetics to bill my health insurance easonably required for billing. I add by my insurance carrier. I irrevocate stand that my out-of-pocket costs vestigation. I agree to be financially ince provider sends a payment direction appropriate and forward such che	plan on my behalf, and ditionally designate Ba ably assign associated may be different than to responsible for all am actly to me for unpaid s ck to Baylor Genetics w	ic testing ordered by my healthcare p d further authorize Baylor Genetics to ylor Genetics as my designated repre payment to Baylor Genetics, and dire the estimated amount indicated to me nounts as indicated on the explanation services performed by Baylor Genetic within thirty (30) days of receipt there y for the full cost of the genetic testin	release any information sentative for purposes ct that payment be made by Baylor Genetics as n of benefits issued by my s on my behalf, I agree of, as payment towards
I understand	that a completed Adv	ance Beneficiary Notice (ABN) is re	quired for Medicare pa	atients if the service is deemed not m	edically necessary.
RECONTACT	FOR RESEARCH CONS	ENT			
contact patie research inv	ents or their provider(solving the sample(s) a	s) directly as part of this research.	I agree to allow Baylor this testing. I understa	pment, and other scientific purposes. Genetics to contact me or my provide nd that patients generally receive no baylorgenetics.com.	er(s) about possible
If I wish to op	ot out of being reconta	cted for research purposes by Bay	lor Genetics, I underst	and that I may check the box below:	
□ Please do	not contact me regard	ling any research that uses informa	ation obtained from thi	s testing.	
	arch I may be contacto via secure email if po		he following methods	(please check all that apply – if no cho	pices are selected, contact
□ Email □ F	Phone □ Mail				



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INFORMED CONSENT FOR MITOCHONDRIAL TESTING

			/	1	
Patient Last Name	Patient First Name	MI	Date of Birth (MM / D	_ ,	Genetic Sex
PATIENT AUTHORIZATION					
By signing this statement of consent, appropriate explanations from my he provider about the availability and im or medical geneticist who can provide informed decision about the genetic t	althcare provider about the portance of genetic counsel e such counseling services. est(s).	planned genetic test(s) and ling and have been provide All my questions have bee	d possible results. I hav d with written informal n answered and I have	ve been informed b tion identifying a g	by my healthcare enetic counselor
Patient's Printed Name		atient's Signature		Date (N	// MM / DD / YYYY)
Patient's Parent / Personal Representative*	Name Pa	atient's Parent / Personal Repre	esentative Signature	Date (M	/ MM / DD / YYYY)
Relationship of Personal Representative to	the Patient Or	rdering Provider's Signature			// 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.