

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

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

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

**STATEMENT OF MEDICAL NECESSITY (REQUIRED)**

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

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

## MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

### ETHNICITY

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

### SAMPLE

#### SAMPLE TYPE

- |   |  |                                    |
|---|--|------------------------------------|
| <input type="radio"/> Blood in EDTA-tube (purple-top) | <input type="radio"/> DNA (Specify): _____   | <input type="radio"/> Skin Biopsy* |
| <input type="radio"/> Buccal Swab <sup>†</sup>        | <input type="radio"/> Other (Specify): _____ |                                    |
| <input type="radio"/> Cord Blood                      | <input type="radio"/> Saliva                 |                                    |

#### DATE OF COLLECTION (MM/DD/YYYY)

\_\_\_\_ / \_\_\_\_ / \_\_\_\_

**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.

Blood should not be sent from patients who have had a bone marrow transplant.  
\* Only accepted for FMR1 CGG Repeat Expansion Analysis (test code 6573)

### INDICATION FOR TESTING (REQUIRED)

Symptomatic (Summarize below)

Symptomatic with Positive Family History

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 below and complete section to the right)

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

Full Gene Sequencing

Deletion/ Duplication Analysis

### FOR TARGETED TESTING SELECTION ONLY

Proband Last Name \_\_\_\_\_ Proband First Name \_\_\_\_\_

Relationship to Proband \_\_\_\_\_ Date (MM / DD / YYYY) \_\_\_\_\_

### PROBAND TESTING LOCATION (SELECT ONE)

Baylor Genetics

Another laboratory

Lab # \_\_\_\_\_ Family # \_\_\_\_\_

1. Attach a copy of the Proband test results
2. A positive control sample of the Proband is requested. Please provide, if available.

### MOLECULAR DIAGNOSTIC TESTS

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

TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/> 20100	Albinism Panel (13 genes)	BE, BUC, DNA
<input type="checkbox"/> 20400	Bardet-Biedl Syndrome Panel (18 genes)	BE, BUC, DNA
<input type="checkbox"/> 2105	Cholestasis Panel (7 genes)	BE, BUC, DNA
<input type="checkbox"/> 2100	CoQ10 Panel (PDSS1, PDSS2, COQ2, COQ9, and ADCK3)	BE, BUC, DNA
<input type="checkbox"/> 2120	Cobalamin Metabolism Panel + Severe MTHFR Deficiency (20 genes)	BE, BUC, DNA
<input type="checkbox"/> 2625	COL1A1/2-Related Disorders (COL1A1 & COL1A2)	BE, BUC, DNA
<input type="checkbox"/> 5095	Congenital Disorders of Glycosylation Panel (36 genes)	BE, BUC, DNA
<input type="checkbox"/> 2095	Fatty Acid Oxidation Deficiency Panel (20 genes)	BE, BUC, DNA
<input type="checkbox"/> 2125	Glycogen Storage Disease (GSD) Comprehensive Panel (23 genes)	BE, BUC, DNA
<input type="checkbox"/> 2126	Glycogen Storage Disease (GSD) Muscle Panel (13 genes)	BE, BUC, DNA
<input type="checkbox"/> 2127	Glycogen Storage Disease (GSD) Liver Panel (13 genes)	BE, BUC, DNA
<input type="checkbox"/> 2200	High Bone Mass Panel (14 genes)	BE, BUC, DNA
<input type="checkbox"/> 21700	Hyperinsulinism Panel (8 genes)	BE, BUC, DNA
<input type="checkbox"/> 21000	Hypoglycemia Panel (85 genes)	BE, BUC, DNA

\* 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.



## MOLECULAR DIAGNOSTIC TESTING REQUISITION

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### MOLECULAR DIAGNOSTIC TESTS

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

TEST CODE	TEST NAME	SAMPLE TYPE *	TEST CODE	TEST NAME	SAMPLE TYPE *
<input type="checkbox"/> 2090	Low Bone Mass Panel (23 genes)	BE, BUC, DNA	<input type="checkbox"/> 24001	Noonan Spectrum Disorders Panel (26 genes)	BE, BUC, DNA, SA
<input type="checkbox"/> 32870	Maple Syrup Urine Disease (MSUD) Panel (BCKHDA, BCKHDB, DBT, and DBD)	BE, BUC, DNA	<input type="checkbox"/> 22100	Peroxisomal Disorders Panel (22 genes)	BE, BUC, DNA
<input type="checkbox"/> 21900	Maturity-Onset Diabetes of the Young (MODY) Panel (25 genes)	BE, BUC, DNA	<input type="checkbox"/> 5274	Proximal Urea Cycle Disorders (PUCD Comprehensive (Seq. & Del/Dup) (CPS1, NAGS, OTC)	BE, BUC, DNA
<input type="checkbox"/> 2300	Myopathy/Rhabdomyolysis Panel (25 genes)	BE, BUC, DNA	<input type="checkbox"/> 2190	Retinitis Pigmentosa + RPGR orf15 by NGS (66 genes)	BE, BUC, DNA
<input type="checkbox"/> 20200	Nephronophthisis Panel (NPHP1, INVS/ NPHP2, NPHP3, and NPHP4)	BE, BUC, DNA	<input type="checkbox"/> 2110	Urea Cycle Disorders (UCD) and Hyperammonemia by NGS (8 genes)	BE, BUC, DNA

#### 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 *
<input type="checkbox"/> 5044	HSD17B10 Comprehensive (Seq & Del/Dup Analysis)	2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency	BE, DNA
<input type="checkbox"/> 5064	HMGCL Comprehensive (Seq & Del/Dup Analysis)	3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	BE, DNA
<input type="checkbox"/> 2874	MCCC1 and MCCC2 Comprehensive (Seq & Del/Dup Analysis)	3-Methylcrotonyl-CoA-Carboxylase Deficiency	BE, DNA
<input type="checkbox"/> 3639	MCCC1 Comprehensive (Seq & Del/Dup Analysis)	3-Methylcrotonyl-CoA-Carboxylase Deficiency	BE, DNA
<input type="checkbox"/> 3644	MCCC2 Comprehensive (Seq & Del/Dup Analysis)	3-Methylcrotonyl-CoA-Carboxylase Deficiency	BE, DNA
<input type="checkbox"/> 3914	AUH Comprehensive (Seq & Del/Dup Analysis)	3-Methylglutaconic Aciduria Type I	BE, DNA
<input type="checkbox"/> 6603	ABCA4 Comprehensive (Seq & Del/Dup Analysis)	ABCA4-Related Disorders	BE, DNA
<input type="checkbox"/> 3284	LPIN1 Comprehensive (Seq & Del/Dup Analysis)	Acute Recurrent Myoglobinuria (LPIN1-Related Disorders)	BE, DNA
<input type="checkbox"/> 2034	ACADSB Comprehensive (Seq & Del/Dup Analysis)	Acyl-CoA Dehydrogenase, Short/Branched Chain Deficiency	BE, DNA
<input type="checkbox"/> 2825	APRT Sequence Analysis	Adenine Phosphoribosyltransferase Deficiency	BE, DNA
<input type="checkbox"/> 5010	ADA Sequence Analysis	Adenosine Deaminase Deficiency	BE, DNA
<input type="checkbox"/> 3699	ADSL Comprehensive (Seq & Del/Dup Analysis)	Adenylosuccinase Deficiency	BE, DNA
<input type="checkbox"/> 5279	ABCD1 Comprehensive (Seq & Del/Dup Analysis)	Adrenoleukodystrophy	BE, DNA
<input type="checkbox"/> 3759	JAG1 Comprehensive (Seq & Del/Dup Analysis)	Alagille Syndrome	BE, DNA
<input type="checkbox"/> 2254	ALPL Comprehensive (Seq & Del/Dup Analysis)	ALPL-Related Disorders	BE, DNA
<input type="checkbox"/> 6490	AR Sequence Analysis	Androgen Insensitivity Syndrome	BE, DNA
<input type="checkbox"/> 6006	Angelman Syndrome (UBE3A) Methylation Analysis	Angelman Syndrome	BE, DNA
<input type="checkbox"/> 3429	ARG1 Comprehensive (Seq & Del/Dup Analysis)	Arginase Deficiency	BE, DNA
<input type="checkbox"/> 3459	GATM Comprehensive (Seq & Del/Dup Analysis)	Arginine: Glycine Amidinotransferase Deficiency	BE, DNA
<input type="checkbox"/> 6360	ASL Sequence Analysis	Argininosuccinate Lyase Deficiency	BE, DNA
<input type="checkbox"/> 6742	ARX Comprehensive (Seq & Del/Dup Analysis)	ARX-Related Disorders	BE, DNA

\* Refer to Sample Specifications Table (page 11)

Panels continued on next page



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### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 2205	AGA Sequence Analysis	Aspartylglycosaminuria	BE, DNA
<input type="checkbox"/> 6195	AIRE Sequence Analysis	Autoimmune Polyendocrinopathy 1	BE, DNA
<input type="checkbox"/> 3299	B4GALT7 Comprehensive (Seq & Del/Dup Analysis)	B4GALT7-Related Disorders	BE, DNA
<input type="checkbox"/> 3614	TAZ Comprehensive (Seq & Del/Dup Analysis)	Barth Syndrome (TAZ-Related Disorders)	BE, DNA
<input type="checkbox"/> 3499	BTD Comprehensive (Seq & Del/Dup Analysis)	Biotinidase Deficiency	BE, DNA
<input type="checkbox"/> 6012	Ashkenazic Mutation Panel (BLM)	Bloom Syndrome	BE, DNA
<input type="checkbox"/> 2429	LEMD3 Comprehensive (Seq & Del/Dup Analysis)	Buschke-Ollendorff Syndrome	BE, DNA
<input type="checkbox"/> 2589	TGFB1 Comprehensive (Seq & Del/Dup Analysis)	Camurati-Engelmann Disease	BE, DNA
<input type="checkbox"/> 6910	BRAF Sequence Analysis	Cardiofaciocutaneous Syndrome/ Costello Syndrome	BE, DNA
<input type="checkbox"/> 3439	SLC25A20 (CACT) Comprehensive (Seq & Del/Dup Analysis)	Carnitine Acylcarnitine Translocase Deficiency	BE, DNA
<input type="checkbox"/> 3364	SLC22A5 (OCTN2) Comprehensive (Seq & Del/Dup Analysis)	Carnitine Deficiency, Systemic	BE, DNA
<input type="checkbox"/> 3369	CPT1A Comprehensive (Seq & Del/Dup Analysis)	Carnitine Palmitoyltransferase IA Deficiency	BE, DNA
<input type="checkbox"/> 3374	CPT1B Comprehensive (Seq & Del/Dup Analysis)	Carnitine Palmitoyltransferase IB (CPT1B-Related Disorders)	BE, DNA
<input type="checkbox"/> 3164	CPT2 Comprehensive (Seq & Del/Dup Analysis)	Carnitine Palmitoyltransferase II Deficiency	BE, DNA
<input type="checkbox"/> 6125	RMRP Sequence Analysis	Cartilage Hair Hypoplasia (RMRP-Related Disorders)	BE, DNA
<input type="checkbox"/> 6733	CDKL5 Comprehensive (Seq & Del/Dup Analysis)	CDKL5-Related Disorders	BE, DNA
<input type="checkbox"/> 6376	CFTR Comprehensive Analysis (Seq, Del/Dup & 5T)	CFTR-Related Disorders (Cystic Fibrosis)	BE, DNA
<input type="checkbox"/> 6174	CHD7 Comprehensive (Seq & Del/Dup Analysis)	CHD7-Related Disorders (CHARGE Syndrome)	BE, DNA
<input type="checkbox"/> 6680	CHRNA7 Sequence Analysis	CHRNA7-Related Disorders	BE, DNA
<input type="checkbox"/> 3159	SLC25A13 (CTLN2) Comprehensive (Seq & Del/Dup Analysis)	Citrin Deficiency	BE, DNA
<input type="checkbox"/> 6180	ASS1 Sequence Analysis	Citrullinemia Type 1	BE, DNA
<input type="checkbox"/> 6150	RUNX2 Sequence Analysis	Cleidocranial Dysplasia	BE, DNA
<input type="checkbox"/> 3854	CABC1 (ADCK3) Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA
<input type="checkbox"/> 3419	COQ2 Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA
<input type="checkbox"/> 3414	PDSS2 Comprehensive (Seq & Del/Dup Analysis)	Coenzyme Q10 Deficiency	BE, DNA
<input type="checkbox"/> 7521	COL2A1 Comprehensive (Seq & Del/Dup Analysis)	COL2A1-Related Disorders	BE, DNA
<input type="checkbox"/> 6585	COL5A1 Sequence Analysis	COL5A1-Related Disorders	BE, DNA
<input type="checkbox"/> 6590	COL5A2 Sequence Analysis	COL5A2-Related Disorders	BE, DNA
<input type="checkbox"/> 3180	SDHA Sequence Analysis	Complex II Deficiency	BE, DNA
<input type="checkbox"/> 3185	SDHB Sequence Analysis	Complex II Deficiency	BE, DNA
<input type="checkbox"/> 3190	SDHC Sequence Analysis	Complex II Deficiency	BE, DNA
<input type="checkbox"/> 3195	SDHD Sequence Analysis	Complex II Deficiency	BE, DNA
<input type="checkbox"/> 2069	CYP17A1 Comprehensive (Seq & Del/Dup Analysis)	Congenital Adrenal Hyperplasia	BE, DNA
<input type="checkbox"/> 3259	CDG1A (PMM2) Comprehensive (Seq & Del/Dup Analysis)	Congenital Disorders of Glycosylation	BE, DNA
<input type="checkbox"/> 3454	CDG1B (MPI) Comprehensive (Seq & Del/Dup Analysis)	Congenital Disorders of Glycosylation	BE, DNA

\* Refer to Sample Specifications Table (page 11)

Panels continued on next page



## MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

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MI \_\_\_\_\_

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

Genetic Sex \_\_\_\_\_

### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 5119	CDG1M (DOLK) Comprehensive (Seq & Del/Dup Analysis)	Congenital Disorders of Glycosylation	BE, DNA
<input type="checkbox"/> 6545	HRAS Sequence Analysis	Costello Syndrome	BE, DNA
<input type="checkbox"/> 3150	SLC6A8 (CT1) Sequence Analysis	Creatine Transporter (CRTR) Deficiency-Related Disorders	BE, DNA
<input type="checkbox"/> 6949	RPS19 Comprehensive (Seq & Del/Dup Analysis)	Diamond Blackfan Anemia-RPS19 Related Disorders	BE, DNA
<input type="checkbox"/> 5310	TBX1 Sequence Analysis	DiGeorge Syndrome	BE, DNA
<input type="checkbox"/> 3464	DLD Comprehensive (Seq & Del/Dup Analysis)	Dihydroliipoamide Dehydrogenase Deficiency	BE, DNA
<input type="checkbox"/> 6350	DMD Deletion/Duplication Analysis	DMD-Related Disorders	BE, DNA
<input type="checkbox"/> 2634	SLC39A13 Comprehensive (Seq & Del/Dup Analysis)	Ehlers-Danlos Syndrome, Spondylodysplastic Type 3	BE, DNA
<input type="checkbox"/> 6930	Type 4, STXBP1 Sequence Analysis	Epileptic Encephalopathy, Early Infantile	BE, DNA
<input type="checkbox"/> 7110	Type 7, KCNQ2 Sequence Analysis	Epileptic Encephalopathy, Early Infantile	BE, DNA
<input type="checkbox"/> 3749	ETHE1 Comprehensive (Seq & Del/Dup Analysis)	Ethylmalonic Encephalopathy	BE, DNA
<input type="checkbox"/> 6011	GLA Comprehensive (Seq & Del/Dup Analysis)	Fabry Disease	BE, DNA
<input type="checkbox"/> 2579	FAM20C Comprehensive (Seq & Del/Dup Analysis)	FAM20C-Related Disorders	BE, DNA
<input type="checkbox"/> 6740	LDLR Comprehensive (Seq & Del/Dup Analysis)	Familial Hypercholesterolemia	BE, DNA
<input type="checkbox"/> 6520	RUNX1 Sequence Analysis	Familial Platelet Disorder w/ Associated Myeloid Malignancy	BE, DNA
<input type="checkbox"/> 6573	FMR1 CGG Repeat Expansion	FMR1-Related Disorders (Fragile X Syndrome)	BE, BUC, DNA, SA
<input type="checkbox"/> 6570	FMR1 Sequence Analysis	FMR1-Related Disorders (Fragile X Syndrome)	BE, DNA
<input type="checkbox"/> 6345	PORCN Sequence Analysis	Focal Dermal Hypoplasia	BE, DNA
<input type="checkbox"/> 6690	FOXF1 Sequence Analysis	FOXF1-Related Disorders	BE, DNA
<input type="checkbox"/> 6031	Friedreich Ataxia Repeat Expansion Analysis	Friedreich Ataxia Syndrome	BE, DNA
<input type="checkbox"/> 6365	FXN Sequence Analysis	Friedreich Ataxia Syndrome	BE, DNA
<input type="checkbox"/> 3939	FBP1 Comprehensive (Seq & Del/Dup Analysis)	Fructose 1,6 Bisphosphatase Deficiency	BE, DNA
<input type="checkbox"/> 3740	FH Sequence Analysis	Fumarate Hydratase Deficiency (FH-Related Disorders)	BE, DNA
<input type="checkbox"/> 3279	GALE Comprehensive (Seq & Del/Dup Analysis)	Galactosemia	BE, DNA
<input type="checkbox"/> 3249	GALT Comprehensive (Seq & Del/Dup Analysis)	Galactosemia	BE, DNA
<input type="checkbox"/> 3799	GALK1 Comprehensive (Seq & Del/Dup Analysis)	Galactokinase Deficiency	BE, DNA
<input type="checkbox"/> 6955	SLC2A1 (GLUT1) Sequence Analysis	Glucose Transporter Type 1 Deficiency Syndrome	BE, DNA
<input type="checkbox"/> 3689	Type 1, GCDH Comprehensive (Seq & Del/Dup Analysis)	Glutaric Acidemia	BE, DNA
<input type="checkbox"/> 2044	Type 3, C7orf10 Comprehensive (Seq & Del/Dup Analysis)	Glutaric Acidemia	BE, DNA
<input type="checkbox"/> 5034	AMT Comprehensive (Seq & Del/Dup Analysis)	Glycine Encephalopathy	BE, DNA
<input type="checkbox"/> 3534	Type 0 Liver Isoform, GYS2 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3839	Type 0 Muscle Isoform, GYS1 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3134	Type 1a (GSD1A), G6PC Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA

\* Refer to Sample Specifications Table (page 11)

Panels continued on next page



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### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3834	Type 1 (b,c,d), SLC37A4 (GSD1B) Comprehensive (Seq & Del/ Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3404	Type II, GAA Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3829	Type IV (GSDIV), GBE1 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3804	Type V (GSDV), PYGM Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3794	Type VI (GSDVI), PYGL Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3824	Type VII (GSDVII), PFKM Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3984	Type IX (GSDIX), PHKG2 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 3809	Type X (GSDX), PGAM2 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 2529	Type XIII (GSDXIII), ENO3 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 2524	Type XIV (GSDXIV), PGM1 Comprehensive (Seq & Del/Dup Analysis)	Glycogen Storage Disease	BE, DNA
<input type="checkbox"/> 5129	GNE Comprehensive (Seq & Del/Dup Analysis)	GNE-Related Disorders	BE, DNA
<input type="checkbox"/> 3149	GAMT Comprehensive (Seq & Del/Dup Analysis)	Guanidinoacetate Methyltransferase Deficiency	BE, DNA
<input type="checkbox"/> 6019	Connexin 26 - GJB2 Sequence Analysis	Hearing Loss	BE, DNA
<input type="checkbox"/> 6355	Connexin 30 - GJB6 (232kb and 309kb) Deletion/Duplication Analysis	Hearing Loss	BE, DNA
<input type="checkbox"/> 3030	Mitochondrial Nonsyndromic Hearing Loss and Deafness Mutation Panel (MT-RNR1, MT-TS1, MT-TS2, MTRNR1)	Hearing Loss	BE, DNA
<input type="checkbox"/> 6395	MYO7A Sequence Analysis	Hearing Loss	BE, DNA
<input type="checkbox"/> 6655	CDH23 Sequence Analysis	Hearing Loss	BE, DNA
<input type="checkbox"/> 6670	POU3F4 Sequence Analysis	Hearing Loss	BE, DNA
<input type="checkbox"/> 3344	TIMM8A Comprehensive (Seq & Del/Dup Analysis)	Hearing Loss	BE, DNA
<input type="checkbox"/> 5405	Hemochromatosis Panel by Sanger Sequencing (HAMP, HFE, HFE2, SLC40A1, TFR2)	Hemochromatosis	BE, DNA
<input type="checkbox"/> 3129	ALDOB Comprehensive (Seq & Del/Dup Analysis)	Hereditary Fructose Intolerance	BE, DNA
<input type="checkbox"/> 3784	ALDOB, FBP1, GYS2, & PC Sequence Analysis	Hereditary Fructose Intolerance	BE, DNA
<input type="checkbox"/> 2145	SEPT9 Targeted Mutation Analysis	Hereditary Neuralgic Amyotrophy (HNA)	BE, DNA
<input type="checkbox"/> 6925	HEXA Sequence Analysis	Hexosaminidase A Deficiency/ Tay-Sachs Disease	BE, DNA
<input type="checkbox"/> 5390	HNRNPA1 Sequence Analysis	HNRNPA1-Related Disorders	BE, DNA
<input type="checkbox"/> 3544	HLCS Comprehensive (Seq & Del/Dup Analysis)	Holocarboxylase Synthetase Deficiency	BE, DNA
<input type="checkbox"/> 3974	CBS Comprehensive (Seq & Del/Dup Analysis)	Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	BE, DNA
<input type="checkbox"/> 2075	HPD Sequence Analysis	HPD-Related Disorders	BE, DNA
<input type="checkbox"/> 6034	Huntington Disease Repeat Expansion Analysis	Huntington Disease (Disease Specific Consent Required)	BE, DNA
<input type="checkbox"/> 5285	GLUD1 Sequence Analysis	Hyperinsulinism-Hyperammonemia Syndrome	BE, DNA
<input type="checkbox"/> 2070	GNMT Sequence Analysis	Hypermethioninemia	BE, DNA
<input type="checkbox"/> 2135	AHCY Sequence Analysis	Hypermethioninemia with S-Adenosylhomocysteine Hydrolase Deficiency	BE, DNA

\* Refer to Sample Specifications Table (page 11)

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Patient Last Name

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MI

Date of Birth (MM / DD / YYYY)

Genetic Sex

### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3239	SLC25A15 (HHH) Comprehensive (Seq & Del/Dup Analysis)	Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome	BE, DNA
<input type="checkbox"/> 5139	ALDH4A1 Comprehensive (Seq & Del/Dup Analysis)	Hyperprolinemia Type II	BE, DNA
<input type="checkbox"/> 2654	SLC34A1 (NPT2) Comprehensive (Seq & Del/Dup Analysis)	Hypophosphatemic Nephrolithiasis/Osteoporosis, 1	BE, DNA
<input type="checkbox"/> 5045	IYD Sequence Analysis	Hypothyroidism, Congenital	BE, DNA
<input type="checkbox"/> 5395	HNRNPA2B1 Sequence Analysis	Inclusion Body Myopathy with Early-Onset Paget Disease with or without Frontotemporal Dementia 2	BE, DNA
<input type="checkbox"/> 6036	Incontinentia Pigmenti Common Deletion Analysis	Incontinentia Pigmenti (IKBKG-Related Disorders)	BE, DNA
<input type="checkbox"/> 7100	IKBKG Sequence Analysis	Incontinentia Pigmenti (IKBKG-Related Disorders)	BE, DNA
<input type="checkbox"/> 3314	ABCB11 Comprehensive (Seq & Del/Dup Analysis)	Intrahepatic Cholestasis	BE, DNA
<input type="checkbox"/> 3319	ABCB4 Comprehensive (Seq & Del/Dup Analysis)	Intrahepatic Cholestasis	BE, DNA
<input type="checkbox"/> 3309	ATP8B1 Comprehensive (Seq & Del/Dup Analysis)	Intrahepatic Cholestasis	BE, DNA
<input type="checkbox"/> 2029	ACAD8 Comprehensive (Seq & Del/Dup Analysis)	Isobutyryl-CoA Dehydrogenase Deficiency	BE, DNA
<input type="checkbox"/> 3684	IVD Comprehensive (Seq & Del/Dup Analysis)	Isovaleric Acidemia	BE, DNA
<input type="checkbox"/> 6037	Kennedy Disease Repeat Expansion Analysis	Kennedy Disease	BE, DNA
<input type="checkbox"/> 5370	KIF11 Sequence Analysis	KIF11-Related Disorders	BE, DNA
<input type="checkbox"/> 6415	GALC Sequence Analysis	Krabbe Disease	BE, DNA
<input type="checkbox"/> 3389	ACADL Comprehensive (Seq & Del/Dup Analysis)	LCAD Deficiency	BE, DNA
<input type="checkbox"/> 3124	HADHA Comprehensive (Seq & Del/Dup Analysis)	LCHAD Deficiency (HADHA-Related Disorders)	BE, DNA
<input type="checkbox"/> 6065	PTPN11 Sequence Analysis	LEOPARD Syndrome	BE, DNA
<input type="checkbox"/> 6475	RAF1 Sequence Analysis	LEOPARD Syndrome	BE, DNA
<input type="checkbox"/> 6240	HPRT Sequence Analysis	Lesch-Nyhan Syndrome	BE, DNA
<input type="checkbox"/> 3719	DARS2 Comprehensive (Seq & Del/Dup Analysis)	Leukoencephalopathy	BE, DNA
<input type="checkbox"/> 3819	TRMU Comprehensive (Seq & Del/Dup Analysis)	Liver Failure, Acute Infantile	BE, DNA
<input type="checkbox"/> 6039	OCRL Sequence Analysis	Lowe Syndrome	BE, DNA
<input type="checkbox"/> 2039	ACSF3 Comprehensive (Seq & Del/Dup Analysis)	Malonic & Methylmalonic Aciduria, Combined	BE, DNA
<input type="checkbox"/> 2774	Type 1A, BCKDHA Comprehensive (Seq & Del/Dup Analysis)	Maple Syrup Urine Disease	BE, DNA
<input type="checkbox"/> 2884	Type 1B, BCKDHB Comprehensive (Seq & Del/Dup Analysis)	Maple Syrup Urine Disease	BE, DNA
<input type="checkbox"/> 3869	Type 2, DBT Comprehensive (Seq & Del/Dup Analysis)	Maple Syrup Urine Disease	BE, DNA
<input type="checkbox"/> 3119	ACADM Comprehensive (Seq & Del/Dup Analysis)	MCAD Deficiency	BE, DNA
<input type="checkbox"/> 6380	ARSA Sequence Analysis	Metachromatic Leukodystrophy (Arylsulfatase A Deficiency)	BE, DNA
<input type="checkbox"/> 2569	cbIE Type, MTRR Comprehensive (Seq & Del/Dup Analysis)	Methylcobalamin Deficiency	BE, DNA
<input type="checkbox"/> 2054	cbIG Type, MTR Comprehensive (Seq & Del/Dup Analysis)	Methylcobalamin Deficiency	BE, DNA
<input type="checkbox"/> 3602	Methylmalonic Acidemia Comprehensive Panel (MUT, MMAA, MMAB)	Methylmalonic Acidemia	BE, DNA
<input type="checkbox"/> 3399	MCEE Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Acidemia	BE, DNA
<input type="checkbox"/> 3579	MMAA Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Acidemia	BE, DNA

\* Refer to Sample Specifications Table (page 11)

Panels continued on next page



## MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3584	MMAB Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Acidemia	BE, DNA
<input type="checkbox"/> 3444	MMACHC Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Acidemia	BE, DNA
<input type="checkbox"/> 3889	MMADHC Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Acidemia	BE, DNA
<input type="checkbox"/> 3589	MUT Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Acidemia	BE, DNA
<input type="checkbox"/> 2564	cbfI Type, LMBRD1 Comprehensive (Seq & Del/Dup Analysis)	Methylmalonic Aciduria and Homocystinuria	BE, DNA
<input type="checkbox"/> 3064	TYMP Comprehensive (Seq & Del/Dup Analysis)	MNGIE Syndrome	BE, DNA
<input type="checkbox"/> 3599	MOCS1 Comprehensive (Seq & Del/Dup Analysis)	Molybdenum Cofactor Deficiency	BE, DNA
<input type="checkbox"/> 3619	MOCS2 Comprehensive (Seq & Del/Dup Analysis)	Molybdenum Cofactor Deficiency	BE, DNA
<input type="checkbox"/> 6385	Type I (MPS I), IDUA Sequence Analysis	Mucopolysaccharidosis	BE
<input type="checkbox"/> 6814	Type II (MPS II), IDS Comprehensive (Seq & Del/Dup w/Inv Analysis)	Mucopolysaccharidosis	BE, DNA
<input type="checkbox"/> 3604	Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) Comprehensive Panel (Seq & Del/Dup Analysis) (ETFA, ETFB, ETFDH)	Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)	BE, DNA
<input type="checkbox"/> 3859	ETFA Comprehensive (Seq & Del/Dup Analysis)	Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)	BE, DNA
<input type="checkbox"/> 3864	ETFB Comprehensive (Seq & Del/Dup Analysis)	Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)	BE, DNA
<input type="checkbox"/> 3844	ETFDH Comprehensive (Seq & Del/Dup Analysis)	Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)	BE, DNA
<input type="checkbox"/> 6041	Myotonic Dystrophy Type 1 Repeat Expansion Analysis	Myotonic Dystrophy Type 1	BE, DNA
<input type="checkbox"/> 3354	NAGS Comprehensive (Seq & Del/Dup Analysis)	N-Acetylglutamate Synthase (NAGS) Deficiency	BE, DNA
<input type="checkbox"/> 7523	LMX1B Comprehensive (Seq & Del/Dup Analysis)	Nail-Patella Syndrome	BE, DNA
<input type="checkbox"/> 6900	SHOC2 Sequence Analysis	Noonan-like Syndrome	BE, DNA
<input type="checkbox"/> 6845	LEP Sequence Analysis	Obesity, Monogenic Nonsyndromic	BE, DNA
<input type="checkbox"/> 6850	LEPR Sequence Analysis	Obesity, Monogenic Nonsyndromic	BE, DNA
<input type="checkbox"/> 6855	PCSK1 Sequence Analysis	Obesity, Monogenic Nonsyndromic	BE, DNA
<input type="checkbox"/> 6860	POMC Sequence Analysis	Obesity, Monogenic Nonsyndromic	BE, DNA
<input type="checkbox"/> 6083	X-Linked, GPR143 Comprehensive (Seq & Del/Dup Analysis)	Oculocutaneous Albinism	BE, DNA
<input type="checkbox"/> 3529	Type 3, OPA3 Comprehensive (Seq & Del/Dup Analysis)	Optic Atrophy Type 3	BE, DNA
<input type="checkbox"/> 3144	OTC Comprehensive (Seq & Del/Dup Analysis)	Ornithine Transcarbamylase (OTC) Deficiency	BE, DNA
<input type="checkbox"/> 2574	AMER1 Comprehensive (Seq & Del/Dup Analysis)	Osteopathia Striata with Cranial Sclerosis	BE, DNA
<input type="checkbox"/> 2624	TCIRG1 Comprehensive (Seq & Del/Dup Analysis)	Osteopetrosis	BE, DNA
<input type="checkbox"/> 2604	CA2 Comprehensive (Seq & Del/Dup Analysis)	Osteopetrosis with Renal Tubular Acidosis	BE, DNA
<input type="checkbox"/> 6885	PCDH19 Sequence Analysis	PCDH19-Related X Linked Female-Limited Epilepsy w/MR	BE, DNA
<input type="checkbox"/> 3169	PDHA1 Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3899	PDHB Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3924	PDHX Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA
<input type="checkbox"/> 3894	PDP1 Comprehensive (Seq & Del/Dup Analysis)	PDH Complex Deficiency	BE, DNA

\* Refer to Sample Specifications Table (page 11)

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## MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 6550	GJC2 Sequence Analysis	Pelizaeus-Merzbacher-Like Disease	BE, DNA
<input type="checkbox"/> 5365	PGM3 Sequence Analysis	PGM3-Related Disorders	BE, DNA
<input type="checkbox"/> 3139	PAH Comprehensive (Seq & Del/Dup Analysis)	Phenylalanine Hydroxylase Deficiency (PKU)	BE, DNA
<input type="checkbox"/> 6149	PLP1 Comprehensive (Seq & Del/Dup Analysis)	PLP1-Related Disorders	BE, DNA
<input type="checkbox"/> 3729	RARS2 Comprehensive (Seq & Del/Dup Analysis)	Pontocerebellar Hypoplasia Type 6	BE, DNA
<input type="checkbox"/> 6050	Prader-Willi Syndrome Methylation Analysis	Prader-Willi Syndrome	BE, DNA
<input type="checkbox"/> 7105	MAGEL2 Sequence Analysis	Prader-Willi-like Syndrome; Intellectual Disability; Autism	BE, DNA
<input type="checkbox"/> 3622	Propionic Acidemia Comprehensive Panel (Seq & Del/Dup Analysis) (PCCA & PCCB)	Propionic Acidemia	BE, DNA
<input type="checkbox"/> 6048	Prothrombin Mutation Panel (F2)	Prothrombin	BE, DNA
<input type="checkbox"/> 6790	PTEN Comprehensive (Seq & Del/Dup Analysis)	PTEN-Related Disorders	BE, DNA
<input type="checkbox"/> 5025	PNP Sequence Analysis	Purine Nucleoside Phosphorylase Deficiency	BE, DNA
<input type="checkbox"/> 2444	CTSK Comprehensive (Seq & Del/Dup Analysis)	Pycnodysostosis	BE, DNA
<input type="checkbox"/> 6950	ALDH7A1 Sequence Analysis	Pyridoxine-Dependent Seizures	BE, DNA
<input type="checkbox"/> 3919	DLAT Comprehensive (Seq & Del/Dup Analysis)	Pyruvate Dehydrogenase E2 Deficiency	BE, DNA
<input type="checkbox"/> 3754	PC Comprehensive (Seq & Del/Dup Analysis)	Pyruvate Carboxylase Deficiency	BE, DNA
<input type="checkbox"/> 5300	RAG2 Sequence Analysis	RAG2-Related Disorders	BE, DNA
<input type="checkbox"/> 6736	MECP2 Comprehensive (Seq & Del/Dup Analysis)	Rett Syndrome (MECP2-Related Disorders)	BE, DNA
<input type="checkbox"/> 6635	FOXP1 Sequence Analysis	Rett Syndrome, Congenital Variant	BE, DNA
<input type="checkbox"/> 6565	VDR Sequence Analysis	Rickets-Alopecia Syndrome	BE, DNA
<input type="checkbox"/> 6758	CREBBP Comprehensive (Seq & Del/Dup Analysis)	Rubinstein-Taybi Syndrome	BE, DNA
<input type="checkbox"/> 3929	ACADS Comprehensive (Seq & Del/Dup Analysis)	SCAD Deficiency	BE, DNA
<input type="checkbox"/> 6285	COL10A1 Sequence Analysis	Schmid Metaphyseal Chondrodysplasia (SMCD)	BE, DNA
<input type="checkbox"/> 6745	DHCR7 Sequence Analysis	Smith-Lemli-Opitz Syndrome	BE, DNA
<input type="checkbox"/> 6760	RAI1 Sequence Analysis	Smith-Magenis Syndrome	BE, DNA
<input type="checkbox"/> 6059	SMN1/SMN2 Copy Number Analysis	Spinal Muscular Atrophy (SMA) Diagnostic Test	BE, DNA
<input type="checkbox"/> 2899	PRKCG Comprehensive (Seq & Del/Dup Analysis)	Spinocerebellar Ataxia 14 (SCA14)	BE, DNA
<input type="checkbox"/> 6060	SRY Molecular Analysis	SRY-Related Phenotypes	BE, DNA
<input type="checkbox"/> 5024	ALDH5A1 Comprehensive (Seq & Del/Dup Analysis)	Succinic Semialdehyde Dehydrogenase Deficiency	BE, DNA
<input type="checkbox"/> 2510	TMLHE Sequence Analysis	TMLHE Deficiency	BE, DNA
<input type="checkbox"/> 2513	TMLHE Exon 2 Deletion Analysis	TMLHE Deficiency	BE, DNA
<input type="checkbox"/> 3969	TCN2 Comprehensive (Seq & Del/Dup Analysis)	Transcobalamin II Deficiency	BE, DNA
<input type="checkbox"/> 3624	Trifunctional Protein Deficiency Comprehensive Panel (Seq & Del/Dup Analysis) (HADHA and HADHB)	Trifunctional Protein Deficiency	BE, DNA

\* Refer to Sample Specifications Table (page 11)

Panels continued on next page

## MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

TEST CODE	TEST NAME	DISORDER	SAMPLE TYPE *
<input type="checkbox"/> 3634	HADHB Comprehensive (Seq & Del/Dup Analysis) (HADHB)	Trifunctional Protein Deficiency	BE, DNA
<input type="checkbox"/> 5005	TSHR Sequence Analysis	TSHR-Related Disorders	BE, DNA
<input type="checkbox"/> 3449	Type I, FAH Comprehensive (Seq & Del/Dup Analysis)	Tyrosinemia	BE, DNA
<input type="checkbox"/> 2084	Type II, TAT Comprehensive (Seq & Del/Dup Analysis)	Tyrosinemia	BE, DNA
<input type="checkbox"/> 6650	USH2A Sequence Analysis	Usher Syndrome 2A	BE, DNA
<input type="checkbox"/> 6660	CLRN1 Sequence Analysis	Usher Syndrome 3A	BE, DNA
<input type="checkbox"/> 3359	ACADVL Comprehensive (Seq & Del/Dup Analysis)	VLCAD Deficiency	BE, DNA
<input type="checkbox"/> 2554	ATP7B Comprehensive (Seq & Del/Dup Analysis)	Wilson Disease	BE, DNA
<input type="checkbox"/> 6430	LIPA Sequence Analysis	Wolman Disease	BE, DNA

### CUSTOM FAMILY SEQUENCING INFORMATION

Test Code 1580 is to be used when requesting sequencing of a known familial variant(s) for which the Baylor Genetics does not provide a separate test code. These should only be used when Baylor Genetics has already identified the sequence change in the proband/original patient. If proband testing was performed at another lab, call to discuss prior to sending a sample. A positive control may be required in some cases. If testing of proband is needed, see separate requisition "Proband Custom Sequencing Analysis," which can be found at [www.baylorgenetics.com](http://www.baylorgenetics.com).

Name of First Patient Studied \_\_\_\_\_ Relationship to Patient Studied \_\_\_\_\_ Baylor Genetics Lab # \_\_\_\_\_ Family # \_\_\_\_\_

This Family Member is Currently:

- ASYMPTOMATIC** If SYMPTOMATIC, please provide details. Please attach additional pages, if needed.
- SYMPTOMATIC**

*Include a pedigree showing familial relationships.*

Copy of Original Results Attached (REQUIRED)

### CUSTOM FAMILY SEQUENCING TEST

TEST CODE	TEST NAME	GENE NAME (REQUIRED)	MUTATION/UNCLASSIFIED VARIANT (REQUIRED)
<input type="checkbox"/> 1580	Custom Family Member Sequence Analysis - Gene 1	<input type="text"/>	<input type="text"/>

### SAMPLE SPECIFICATIONS TABLE

ABBREVIATION	SAMPLE NAME	RECOMMENDED AMOUNT		SHIPPING INSTRUCTIONS	SPECIAL NOTES
		(2 YRS - ADULT)	(NEWBORN - 2 YRS)		
BE	Blood in EDTA tube (purple-top)	3 - 5 cc	2 - 3 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		Collected with ORAcollect•Dx (OCD-100) self-collection kit (provided by Baylor Genetics with instructions). It is highly recommended that the sample be collected by a healthcare professional. Buccal swab is an accepted sample type for FMR1 CGG Repeat Expansion Analysis (test code 6573) only.
CB	Cord Blood	N/A	1 - 2 cc		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 ug	10 -15 ug		Minimal concentration of 50ng/uL; A260/A280 of ~1.7
SA	Saliva	See Special Notes	See Special Notes		Collected with Oragene DNA Self-Collection Kit.

**INFORMED CONSENT FOR MOLECULAR DIAGNOSTIC 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.

## INFORMED CONSENT FOR MOLECULAR DIAGNOSTIC TESTING

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

### 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](http://www.genome.gov/10002077).
- Samples will be retained in the laboratory in accordance with the laboratory retention policy.
- After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring health care providers unless specific prior arrangements have been made.
- 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](http://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



## INFORMED CONSENT FOR MOLECULAR DIAGNOSTIC TESTING

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

**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.