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**

# 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

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

## 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

- ◯ Blood in EDTA-tube (purple-top)
- ◯ Buccal Swab [1]
- ◯ Cord Blood
- ◯ Saliva
- ◯ DNA (Specify): _____
- ◯ Other (Specify): _____

**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 or recent blood transfusion

[1] 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℠) 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 |
| ☐ | 2100 | CoQ10 Panel (PDSS1, PDSS2, COQ2, COQ9, and ADCK3) | BE, DNA |
| ☐ | 2120 | Cobalamin Metabolism Panel + Severe MTHFR Deficiency (20 genes) | BE, DNA |
| ☐ | 2625 | COL1A1/2-Related Disorders (COL1A1 & COL1A2) | BE, DNA |
| ☐ | 5095 | Congenital Disorders of Glycosylation Panel (36 genes) | BE, DNA |
| ☐ | 2095 | Fatty Acid Oxidation Deficiency Panel (20 genes) | BE, DNA |
| ☐ | 2125 | Glycogen Storage Disease (GSD) Comprehensive 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 |

* Refer to Sample Specifications Table (page 11)

Panels continued on next page

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## MOLECULAR DIAGNOSTIC TESTS

### MASSIVELY PARALLEL SEQUENCING (BCM-MitomeNGS℠)

| TEST CODE | TEST NAME | SAMPLE TYPE * |
|---|---|---|
| ☐ 2090 | Low Bone Mass Panel (23 genes) | BE, DNA |
| ☐ 32870 | Maple Syrup Urine Disease (MSUD) Panel (BCKHDA, BCKHDB, DBT, and DBD) | BE, DNA |
| ☐ 21900 | Maturity-Onset Diabetes of the Young (MODY) Panel (25 genes) | BE, DNA |
| ☐ 2300 | Myopathy/Rhabdomyolysis Panel (25 genes) | BE, DNA |
| ☐ 20200 | Nephronophthisis Panel *(NPHP1, INVS/ NPHP2, NPHP3, and NPHP4)* | BE, DNA |
| ☐ 21400 | Noonan Spectrum Disorders Panel (12 genes) | BE, DNA |
| ☐ 22100 | Peroxisomal Disorders Panel (22 genes) | BE, DNA |
| ☐ 5274 | Proximal Urea Cycle Disordersz (PUCD Comprehensive (Seq. & Del/Dup) *(CPS1, NAGS, OTC)* | BE, DNA |
| ☐ 2190 | Retinitis Pigmentosa + RPGR orf15 by NGS (66 genes) | BE, DNA |
| ☐ 2110 | Urea Cycle Disorders (UCD) and Hyperammonemia by NGS (8 genes) | BE, 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 Name

Test Code | Gene | Test Name

Test Code | Gene | Test Name

| TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|
| ☐ 5044 | HSD17B10 Comprehensive (Seq & Del/Dup Analysis) | 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency | BE, DNA |
| ☐ 5064 | HMGCL Comprehensive (Seq & Del/Dup Analysis) | 3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency | BE, DNA |
| ☐ 29025 | HMGCS2 Sequence Analysis by NGS | 3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency | BE, DNA |
| ☐ 2874 | MCCC1 and MCCC2 Comprehensive (Seq & Del/Dup Analysis) | 3-Methylcrontonyl-CoA-Carboxylase Deficiency | BE, DNA |
| ☐ 3639 | MCCC1 Comprehensive (Seq & Del/Dup Analysis) | 3-Methylcrontonyl-CoA-Carboxylase Deficiency | BE, DNA |
| ☐ 3644 | MCCC2 Comprehensive (Seq & Del/Dup Analysis) | 3-Methylcrontonyl-CoA-Carboxylase Deficiency | BE, DNA |
| ☐ 3914 | AUH Comprehensive (Seq & Del/Dup Analysis) | 3-Methylglutaconic Aciduria Type I | BE, DNA |
| ☐ 6603 | ABCA4 Comprehensive (Seq & Del/Dup Analysis) | ABCA4-Related Disorders | BE, DNA |
| ☐ 6000 | Achondroplasia Mutation Panel (FGFR3) | Achondroplasia | BE, DNA |
| ☐ 3284 | LPIN1 Comprehensive (Seq & Del/Dup Analysis) | Acute Recurrent Myoglobinuria (LPIN1-Related Disorders) | BE, DNA |
| ☐ 2034 | ACADSB Comprehensive (Seq & Del/Dup Analysis) | Acyl-CoA Dehydrogenase, Short/Branched Chain Deficiency | BE, DNA |
| ☐ 2825 | APRT Sequence Analysis | Adenine Phosphoribosyltransferase Deficiency | BE, DNA |
| ☐ 5010 | ADA Sequence Analysis | Adenosine Deaminase Deficiency | BE, DNA |
| ☐ 3699 | ADSL Comprehensive (Seq & Del/Dup Analysis) | Adenylosuccinase Deficiency | BE, DNA |
| ☐ 5279 | ABCD1 Comprehensive (Seq & Del/Dup Analysis) | Adrenoleukodystrophy | BE, DNA |
| ☐ 29480 | SLC12A6 (KCC3A) Sequence Analysis by NGS | Agenesis of the Corpus Callosum with Peripheral Neuropathy | BE, DNA |
| ☐ 3759 | JAG1 Comprehensive (Seq & Del/Dup Analysis) | Alagille Syndrome | BE, DNA |
| ☐ 29390 | MAN2B1 Sequence Analysis by NGS | Alpha-Mannosidosis Types I and II | BE, DNA |
| ☐ 2254 | ALPL Comprehensive (Seq & Del/Dup Analysis) | ALPL-Related Disorders | BE, DNA |
| ☐ 22115 | AMACR Sequence Analysis by NGS | AMACR-Related Disorders | BE, DNA |
| ☐ 6490 | AR Sequence Analysis | Androgen Insensitivity Syndrome | BE, DNA |
| ☐ 6006 | Angelman Syndrome (UBE3A) Methylation Analysis | Angelman Syndrome | BE, DNA |
| ☐ 3429 | ARG1 Comprehensive (Seq & Del/Dup Analysis) | Arginase Deficiency | BE, DNA |
| ☐ 3459 | GATM Comprehensive (Seq & Del/Dup Analysis) | Arginine: Glycine Amidinotransferase Deficiency | BE, DNA |
| ☐ 6360 | ASL Sequence Analysis | Argininosuccinate Lyase Deficiency | BE, DNA |
| ☐ 20405 | ARL6 Sequence Analysis by NGS | ARL6-Related Disorders | BE, DNA |

* Refer to Sample Specifications Table (page 11)

Panels continued on next page

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# MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

## MOLECULAR DIAGNOSTIC TESTS

### SINGLE GENE ANALYSIS

| | TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|---|
| ☐ | 6742 | ARX Comprehensive (Seq & Del/Dup Analysis) | ARX-Related Disorders | BE, DNA |
| ☐ | 2205 | AGA Sequence Analysis | Aspartylglycosaminuria | BE, DNA |
| ☐ | 29530 | TTPA Sequence Analysis by NGS | Ataxia with Isolated Vitamin E Deficiency | BE, DNA |
| ☐ | 29155 | ATP6V0A2 Sequence Analysis by NGS | ATP6V0A2-Related Disorders | BE, DNA |
| ☐ | 6195 | AIRE Sequence Analysis | Autoimmune Polyendocrinopathy 1 | BE, DNA |
| ☐ | 3299 | B4GALT7 Comprehensive (Seq & Del/Dup Analysis) | B4GALT7-Related Disorders | BE, DNA |
| ☐ | 3614 | TAZ Comprehensive (Seq & Del/Dup Analysis) | Barth Syndrome (TAZ-Related Disorders) | BE, DNA |
| ☐ | 29445 | PTS Sequence Analysis by NGS | BH4-Deficient Hyperphenylalaninemia A | BE, DNA |
| ☐ | 29110 | AKR1D1 Sequence Analysis by NGS | Bile Acid Synthesis Defect, Congenital, 2 | BE, DNA |
| ☐ | 3499 | BTD Comprehensive (Seq & Del/Dup Analysis) | Biotinidase Deficiency | BE, DNA |
| ☐ | 6012 | Ashkenazic Mutation Panel (BLM) | Bloom Syndrome | BE, DNA |
| ☐ | 2429 | LEMD3 Comprehensive (Seq & Del/Dup Analysis) | Buschke-Ollendorff Syndrome | BE, DNA |
| ☐ | 2589 | TGFB1 Comprehensive (Seq & Del/Dup Analysis) | Camurati-Engelmann Disease | BE, DNA |
| ☐ | 29285 | ASPA Sequence Analysis by NGS | Canavan Disease | BE, DNA |
| ☐ | 3349 | CPS1 Comprehensive (Seq & Del/Dup Analysis) | Carbamoyl Phosphate Synthetase I Deficiency | BE, DNA |
| ☐ | 6910 | BRAF Sequence Analysis | Cardiofaciocutaneous Syndrome/ Costello Syndrome | BE, DNA |
| ☐ | 3439 | SLC25A20 (CACT) Comprehensive (Seq & Del/Dup Analysis) | Carnitine Acylcarnitine Translocase Deficiency | BE, DNA |
| ☐ | 3364 | SLC22A5 (OCTN2) Comprehensive (Seq & Del/Dup Analysis) | Carnitine Deficiency, Systemic | BE, DNA |
| ☐ | 3369 | CPT1A Comprehensive (Seq & Del/Dup Analysis) | Carnitine Palmitoyltransferase IA Deficiency | BE, DNA |
| ☐ | 3374 | CPT1B Comprehensive (Seq & Del/Dup Analysis) | Carnitine Palmitoyltransferase IB (CPT1B-Related Disorders) | BE, DNA |
| ☐ | 3164 | CPT2 Comprehensive (Seq & Del/Dup Analysis) | Carnitine Palmitoyltransferase II Deficiency | BE, DNA |
| ☐ | 6125 | RMRP Sequence Analysis | Cartilage Hair Hypoplasia (RMRP-Related Disorders) | BE, DNA |
| ☐ | 6733 | CDKL5 Comprehensive (Seq & Del/Dup Analysis) | CDKL5-Related Disorders | BE, DNA |
| ☐ | 29330 | CYP27A1 Sequence Analysis by NGS | Cerebrotendinous Xanthomatosis | BE, DNA |
| ☐ | 6376 | CFTR Comprehensive Analysis (Seq, Del/Dup & 5T) | CFTR-Related Disorders (Cystic Fibrosis) | BE, DNA |
| ☐ | 6174 | CHD7 Comprehensive (Seq & Del/Dup Analysis) | CHD7-Related Disorders (CHARGE Syndrome) | BE, DNA |
| ☐ | 6680 | CHRNA7 Sequence Analysis | CHRNA7-Related Disorders | BE, DNA |
| ☐ | 3159 | SLC25A13 (CTLN2) Comprehensive (Seq & Del/Dup Analysis) | Citrin Deficiency | BE, DNA |
| ☐ | 6180 | ASS1 Sequence Analysis | Citrullinemia Type 1 | BE, DNA |
| ☐ | 29315 | CLN6 Sequence Analysis by NGS | CLN6-Related Disorders | BE, DNA |
| ☐ | 29320 | CLN8 Sequence Analysis by NGS | CLN8-Related Disorders | BE, DNA |
| ☐ | 6150 | RUNX2 Sequence Analysis | Cleidocranial Dysplasia | BE, DNA |
| ☐ | 3854 | CABC1 (ADCK3) Comprehensive (Seq & Del/Dup Analysis) | Coenzyme Q10 Deficiency | BE, DNA |
| ☐ | 3419 | COQ2 Comprehensive (Seq & Del/Dup Analysis) | Coenzyme Q10 Deficiency | BE, DNA |
| ☐ | 3414 | PDSS2 Comprehensive (Seq & Del/Dup Analysis) | Coenzyme Q10 Deficiency | BE, DNA |
| ☐ | 2639 | COL1A2 Comprehensive (Seq & Del/Dup Analysis) | COL1A2-Related Disorders | BE, DNA |
| ☐ | 7521 | COL2A1 Comprehensive (Seq & Del/Dup Analysis) | COL2A1-Related Disorders | BE, DNA |
| ☐ | 6585 | COL5A1 Sequence Analysis | COL5A1-Related Disorders | BE, DNA |
| ☐ | 6590 | COL5A2 Sequence Analysis | COL5A2-Related Disorders | BE, DNA |
| ☐ | 29440 | PROP1 Sequence Analysis by NGS | Combined Pituitary Hormone Deficiency 2 | BE, DNA |
| ☐ | 3180 | SDHA Sequence Analysis | Complex II Deficiency | BE, DNA |

* Refer to Sample Specifications Table (page 11)

Panels continued on next page

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# MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

## MOLECULAR DIAGNOSTIC TESTS

### SINGLE GENE ANALYSIS

| | TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|---|
| ☐ | 3185 | SDHB Sequence Analysis | Complex II Deficiency | BE, DNA |
| ☐ | 3190 | SDHC Sequence Analysis | Complex II Deficiency | BE, DNA |
| ☐ | 3195 | SDHD Sequence Analysis | Complex II Deficiency | BE, DNA |
| ☐ | 2069 | CYP17A1 Comprehensive (Seq & Del/Dup Analysis) | Congenital Adrenal Hyperplasia | BE, DNA |
| ☐ | 3259 | CDG1A (PMM2) Comprehensive (Seq & Del/Dup Analysis) | Congenital Disorders of Glycosylation | BE, DNA |
| ☐ | 3454 | CDG1B (MPI) Comprehensive (Seq & Del/Dup Analysis) | Congenital Disorders of Glycosylation | BE, DNA |
| ☐ | 5119 | CDG1M (DOLK) Comprehensive (Seq & Del/Dup Analysis) | Congenital Disorders of Glycosylation | BE, DNA |
| ☐ | 29510 | TGM1 Sequence Analysis by NGS | Congenital Ichthyosis, Autosomal Recessive 1 | BE, DNA |
| ☐ | 6805 | Coronary Heart Disease Risk Factor (9p21 rs10757278) | Coronary Heart Disease (CHD) | BE, DNA |
| ☐ | 6545 | HRAS Sequence Analysis | Costello Syndrome | BE, DNA |
| ☐ | 3150 | SLC6A8 (CT1) Sequence Analysis | Creatine Transporter (CRTR) Deficiency-Related Disorders | BE, DNA |
| ☐ | 29325 | CTNS Sequence Analysis by NGS | CTNS-Related Disorders | BE, DNA |
| ☐ | 6949 | RPS19 Comprehensive (Seq & Del/Dup Analysis) | Diamond Blackfan Anemia-RPS19 Related Disorders | BE, DNA |
| ☐ | 5310 | TBX1 Sequence Analysis | DiGeorge Syndrome | BE, DNA |
| ☐ | 3464 | DLD Comprehensive (Seq & Del/Dup Analysis) | Dihydrolipoamide Dehydrogense Deficiency | BE, DNA |
| ☐ | 29340 | DPYD Sequence Analysis by NGS | DPYD-Related Disorders | BE, DNA |
| ☐ | 20145 | DMD Comprehensive Sequence and CNV Analysis by NGS | DMD-Related Disorders | BE, DNA |
| ☐ | 6350 | DMD Deletion/Duplication Analysis | DMD-Related Disorders | BE, DNA |
| ☐ | 2634 | "Spondylocheirodysplastic Form, SLC39A13 (ZnT) Comprehensive (Seq & Del/Dup Analysis)" | Ehlers-Danlos Syndrome | BE, DNA |
| ☐ | 2754 | COL3A1 Comprehensive (Seq & Del/Dup Analysis) | Ehlers-Danlos Syndrome | BE, DNA |
| ☐ | 22120 | DNM1L Sequence Analysis by NGS | Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission | BE, DNA |
| ☐ | 6930 | Type 4, STXBP1 Sequence Analysis | Epileptic Encephalopathy, Early Infantile | BE, DNA |
| ☐ | 7110 | Type 7, KCNQ2 Sequence Analysis | Epileptic Encephalopathy, Early Infantile | BE, DNA |
| ☐ | 3749 | ETHE1 Comprehensive (Seq & Del/Dup Analysis) | Ethylmalonic Encephalopathy | BE, DNA |
| ☐ | 6011 | GLA Comprehensive (Seq & Del/Dup Analysis) | Fabry Disease | BE, DNA |
| ☐ | 6028 | Factor V Leiden (F5) Mutation Panel | Factor V Leiden | BE, DNA |
| ☐ | 2579 | FAM20C Comprehensive (Seq & Del/Dup Analysis) | FAM20C-Related Disorders | BE, DNA |
| ☐ | 29370 | IKBKAP Sequence Analysis by NGS | Familial Dysautonomia | BE, DNA |
| ☐ | 6740 | LDLR Comprehensive (Seq & Del/Dup Analysis) | Familial Hypercholesterolemia | BE, DNA |
| ☐ | 6520 | RUNX1 Sequence Analysis | Familial Platelet Disorder w/ Associated Myeloid Malignancy | BE, DNA |
| ☐ | 29345 | FANCC Sequence Analysis by NGS | Fanconi Anemia | BE, DNA |
| ☐ | 2339 | FBN1 Comprehensive (Seq & Del/Dup Analysis) | FBN1-Related Disorders | BE, DNA |
| ☐ | 6573 | FMR1 CGG Repeat Expansion | FMR1-Related Disorders (Fragile X Syndrome) | BE, BUC, DNA, SA |
| ☐ | 6570 | FMR1 Sequence Analysis | FMR1-Related Disorders (Fragile X Syndrome) | BE, DNA |
| ☐ | 6345 | PORCN Sequence Analysis | Focal Dermal Hypoplasia | BE, DNA |
| ☐ | 6690 | FOXF1 Sequence Analysis | FOXF1-Related Disorders | BE, DNA |
| ☐ | 6031 | Friedreich Ataxia Repeat Expansion Analysis | Friedreich Ataxia Syndrome | BE, DNA |
| ☐ | 6365 | FXN Sequence Analysis | Friedreich Ataxia Syndrome | BE, DNA |
| ☐ | 3939 | FBP1 Comprehensive (Seq & Del/Dup Analysis) | Fructose 1,6 Bisphosphatase Deficiency | BE, DNA |
| ☐ | 3740 | FH Sequence Analysis | Fumarate Hydratase Deficiency (FH-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 * |
|---|---|---|---|
| ☐ 29355 | G6PD Sequence Analysis by NGS | G6PD-Related Disorders | BE, DNA |
| ☐ 3279 | GALE Comprehensive (Seq & Del/Dup Analysis) | Galactosemia | BE, DNA |
| ☐ 3249 | GALT Comprehensive (Seq & Del/Dup Analysis) | Galactosemia | BE, DNA |
| ☐ 3799 | GALK1 Comprehensive (Seq & Del/Dup Analysis) | Galactokinase Deficiency | BE, DNA |
| ☐ 6955 | SLC2A1 (GLUT1) Sequence Analysis | Glucose Transporter Type 1 Deficiency Syndrome | BE, DNA |
| ☐ 3689 | Type 1, GCDH Comprehensive (Seq & Del/Dup Analysis) | Glutaric Acidemia | BE, DNA |
| ☐ 2044 | Type 3, C7orf10 Comprehensive (Seq & Del/Dup Analysis) | Glutaric Acidemia | BE, DNA |
| ☐ 5034 | AMT Comprehensive (Seq & Del/Dup Analysis) | Glycine Encephalopathy | BE, DNA |
| ☐ 3534 | Type 0 Liver Isoform, GYS2 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3839 | Type 0 Muscle Isoform, GYS1 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3134 | Type 1a (GSD1A), G6PC Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3834 | Type 1 (b,c,d), SLC37A4 (GSD1B) Comprehensive (Seq & Del/ Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3404 | Type II, GAA Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3674 | Type III, AGL Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3829 | Type IV (GSDIV), GBE1 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3804 | Type V (GSDV), PYGM Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3794 | Type VI (GSDVI), PYGL Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3824 | Type VII (GSDVII), PFKM Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3979 | Type IX (GSDIX), PHKB Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3984 | Type IX (GSDIX), PHKG2 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3989 | Type IX (GSDIX), PHKA1 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3994 | Type IX (GSDIX), PHKA2 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 3809 | Type X (GSDX), PGAM2 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 2529 | Type XIII (GSDXIII), ENO3 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 2524 | Type XIV (GSDXIV), PGM1 Comprehensive (Seq & Del/Dup Analysis) | Glycogen Storage Disease | BE, DNA |
| ☐ 5129 | GNE Comprehensive (Seq & Del/Dup Analysis) | GNE-Related Disorders | BE, DNA |
| ☐ 3149 | GAMT Comprehensive (Seq & Del/Dup Analysis) | Guanidinoacetate Methyltransferase Deficiency | BE, DNA |
| ☐ 6019 | Connexin 26 - GJB2 Sequence Analysis | Hearing Loss | BE, DNA |
| ☐ 6355 | Connexin 30 - GJB6 (232kb and 309kb) Deletion/Duplication Analysis | Hearing Loss | BE, DNA |
| ☐ 3030 | Mitochondrial Nonsyndromic Hearing Loss and Deafness Mutation Panel (MT-RNR1, MT-TS1, MT-TS2, MTRNR1) | Hearing Loss | BE, DNA |
| ☐ 6395 | MYO7A Sequence Analysis | Hearing Loss | BE, DNA |
| ☐ 6655 | CDH23 Sequence Analysis | Hearing Loss | BE, DNA |

* Refer to Sample Specifications Table (page 11)

Panels continued on next page

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# MOLECULAR DIAGNOSTIC TESTING REQUISITION

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

## MOLECULAR DIAGNOSTIC TESTS

### SINGLE GENE ANALYSIS

| TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|
| ☐ 6670 | POU3F4 Sequence Analysis | Hearing Loss | BE, DNA |
| ☐ 3344 | TIMM8A Comprehensive (Seq & Del/Dup Analysis) | Hearing Loss | BE, DNA |
| ☐ 5405 | Hemochromatosis Panel by Sanger Sequencing (HAMP, HFE, HFE2, SLC40A1, TFR2) | Hemochromatosis | BE, DNA |
| ☐ 6035 | HFE Mutation Panel | Hemochromatosis | BE, DNA |
| ☐ 3129 | ALDOB Comprehensive (Seq & Del/Dup Analysis) | Hereditary Fructose Intolerance | BE, DNA |
| ☐ 3784 | ALDOB, FBP1, GYS2, & PC Sequence Analysis | Hereditary Fructose Intolerance | BE, DNA |
| ☐ 2145 | SEPT9 Targeted Mutation Analysis | Hereditary Neuralgic Amyotrophy (HNA) | BE, DNA |
| ☐ 20110 | BLOC1S3 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 20115 | DTNBP1 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 20120 | HPS1 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 20125 | HPS3 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 20130 | HPS4 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 20135 | HPS5 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 20140 | HPS6 Sequence Analysis by NGS | Hermansky-Pudlak Syndrome | BE, DNA |
| ☐ 6925 | HEXA Sequence Analysis | Hexosaminidase A Deficiency/ Tay-Sachs Disease | BE, DNA |
| ☐ 5390 | HNRNPA1 Sequence Analysis | HNRNPA1-Related Disorders | BE, DNA |
| ☐ 3544 | HLCS Comprehensive (Seq & Del/Dup Analysis) | Holocarboxylase Synthetase Deficiency | BE, DNA |
| ☐ 3974 | CBS Comprehensive (Seq & Del/Dup Analysis) | Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency | BE, DNA |
| ☐ 20640 | MTHFR Sequence Analysis by NGS | Homocystinuria Caused by MTHFR Deficiency | BE, DNA |
| ☐ 2075 | HPD Sequence Analysis | HPD-Related Disorders | BE, DNA |
| ☐ 22130 | HSD17B4 Sequence Analysis by NGS | HSD17B4-Related Disorders | BE, DNA |
| ☐ 6034 | Huntington Disease Repeat Expansion Analysis | Huntington Disease (Disease Specific Consent Required) | BE, DNA |
| ☐ 5285 | GLUD1 Sequence Analysis | Hyperinsulinism-Hyperammonemia Syndrome | BE, DNA |
| ☐ 2070 | GNMT Sequence Analysis | Hypermethioninemia | BE, DNA |
| ☐ 2135 | AHCY Sequence Analysis | Hypermethioninemia with S-Adenosylhomocysteine Hydrolase Deficiency | BE, DNA |
| ☐ 3239 | SLC25A15 (HHH) Comprehensive (Seq & Del/Dup Analysis) | Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome | BE, DNA |
| ☐ 5139 | ALDH4A1 Comprehensive (Seq & Del/Dup Analysis) | Hyperprolinemia Type II | BE, DNA |
| ☐ 2654 | SLC34A1 (NPT2) Comprehensive (Seq & Del/Dup Analysis) | Hypophosphatemic Nephrolithiasis/Osteoporosis, 1 | BE, DNA |
| ☐ 5045 | IYD Sequence Analysis | Hypothyroidism, Congenital | BE, DNA |
| ☐ 5395 | HNRNPA2B1 Sequence Analysis | Inclusion Body Myopathy with Early-Onset Paget Disease with or without Frontotemporal Dementia 2 | BE, DNA |
| ☐ 6036 | Incontentia Pigmenti Common Deletion Analysis | Incontinentia Pigmenti (IKBKG-Related Disorders) | BE, DNA |
| ☐ 7100 | IKBKG Sequence Analysis | Incontinentia Pigmenti (IKBKG-Related Disorders) | BE, DNA |
| ☐ 21720 | INSR Sequence Analysis by NGS | INSR-Related Disorders | BE, DNA |
| ☐ 3314 | ABCB11 Comprehensive (Seq & Del/Dup Analysis) | Intrahepatic Cholestasis | BE, DNA |
| ☐ 3319 | ABCB4 Comprehensive (Seq & Del/Dup Analysis) | Intrahepatic Cholestasis | BE, DNA |
| ☐ 3309 | ATP8B1 Comprehensive (Seq & Del/Dup Analysis) | Intrahepatic Cholestasis | BE, DNA |
| ☐ 29120 | GIF Sequence Analysis by NGS | Intrinsic Factor Deficiency | BE, DNA |
| ☐ 2029 | ACAD8 Comprehensive (Seq & Del/Dup Analysis) | Isobutyryl-CoA Dehydrogenase 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) | Biological Sex

## MOLECULAR DIAGNOSTIC TESTS

### SINGLE GENE ANALYSIS

| | TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|---|
| ☐ | 3684 | IVD Comprehensive (Seq & Del/Dup Analysis) | Isovaleric Acidemia | BE, DNA |
| ☐ | 6037 | Kennedy Disease Repeat Expansion Analysis | Kennedy Disease | BE, DNA |
| ☐ | 5370 | KIF11 Sequence Analysis | KIF11-Related Disorders | BE, DNA |
| ☐ | 6415 | GALC Sequence Analysis | Krabbe Disease | BE, DNA |
| ☐ | 29375 | LAMA3 Sequence Analysis by NGS | LAMA3-Related Disorders | BE, DNA |
| ☐ | 29380 | LAMB3 Sequence Analysis by NGS | LAMB3-Related Disorders | BE, DNA |
| ☐ | 29385 | LAMC2 Sequence Analysis by NGS | LAMC2-Related Disorders | BE, DNA |
| ☐ | 3389 | ACADL Comprehensive (Seq & Del/Dup Analysis) | LCAD Deficiency | BE, DNA |
| ☐ | 3124 | HADHA Comprehensive (Seq & Del/Dup Analysis) | LCHAD Deficiency (HADHA-Related Disorders) | BE, DNA |
| ☐ | 6065 | PTPN11 Sequence Analysis | LEOPARD Syndrome | BE, DNA |
| ☐ | 6475 | RAF1 Sequence Analysis | LEOPARD Syndrome | BE, DNA |
| ☐ | 6240 | HPRT Sequence Analysis | Lesch-Nyhan Syndrome | BE, DNA |
| ☐ | 3719 | DARS2 Comprehensive (Seq & Del/Dup Analysis) | Leukoencephalopathy | BE, DNA |
| ☐ | 22220 | SCP2 Sequence Analysis by NGS | Leukoencephalopathy with dystonia and motor neuropathy | BE, DNA |
| ☐ | 3819 | TRMU Comprehensive (Seq & Del/Dup Analysis) | Liver Failure, Acute Infantile | BE, DNA |
| ☐ | 6039 | OCRL Sequence Analysis | Lowe Syndrome | BE, DNA |
| ☐ | 29500 | SLC7A7 (LAT1) Sequence Analysis by NGS | Lysinuric Protein Intolerance | BE, DNA |
| ☐ | 2039 | ACSF3 Comprehensive (Seq & Del/Dup Analysis) | Malonic & Methylmalonic Aciduria, Combined | BE, DNA |
| ☐ | 2774 | Type 1A, BCKDHA Comprehensive (Seq & Del/Dup Analysis) | Maple Syrup Urine Disease | BE, DNA |
| ☐ | 2884 | Type 1B, BCKDHB Comprehensive (Seq & Del/Dup Analysis) | Maple Syrup Urine Disease | BE, DNA |
| ☐ | 3869 | Type 2, DBT Comprehensive (Seq & Del/Dup Analysis) | Maple Syrup Urine Disease | BE, DNA |
| ☐ | 3119 | ACADM Comprehensive (Seq & Del/Dup Analysis) | MCAD Deficiency | BE, DNA |
| ☐ | 29400 | MEFV Sequence Analysis by NGS | MEFV-Related Disorders | BE, DNA |
| ☐ | 29405 | MLC1 Sequence Analysis by NGS | Megalencephalic Leukoencephalopathy with Subcortical Cysts | BE, DNA |
| ☐ | 2549 | ATP7A Comprehensive (Seq & Del/Dup Analysis) | Menkes Disease | BE, DNA |
| ☐ | 6380 | ARSA Sequence Analysis | Metachromatic Leukodystrophy (Arylsulfatase A Deficiency) | BE, DNA |
| ☐ | 2569 | cblE Type, MTRR Comprehensive (Seq & Del/Dup Analysis) | Methylcobalamin Deficiency | BE, DNA |
| ☐ | 2054 | cblG Type, MTR Comprehensive (Seq & Del/Dup Analysis) | Methylcobalamin Deficiency | BE, DNA |
| ☐ | 3602 | Methylmalonic Acidemia Comprehensive Panel (MUT, MMAA, MMAB) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 3399 | MCEE Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 3579 | MMAA Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 3584 | MMAB Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 3444 | MMACHC Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 3889 | MMADHC Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 3589 | MUT Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Acidemia | BE, DNA |
| ☐ | 29115 | HCFC1 Sequence Analysis by NGS | Methylmalonic Acidemia and Homocystinuria, cblX Type | BE, DNA |
| ☐ | 2564 | cblF Type, LMBRD1 Comprehensive (Seq & Del/Dup Analysis) | Methylmalonic Aciduria and Homocystinuria | BE, DNA |
| ☐ | 29125 | CD320 Sequence Analysis by NGS | Methylmalonic Aciduria due to Transcobalamin Receptor Defect | BE, DNA |
| ☐ | 20455 | MKKS Sequence Analysis by NGS | MKKS-Related Disorders | BE, DNA |
| ☐ | 20460 | MKS1 Sequence Analysis by NGS | MKS1-Related Disorders | 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) | Biological Sex

### MOLECULAR DIAGNOSTIC TESTS

#### SINGLE GENE ANALYSIS

| | TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|---|
| ☐ | 3064 | TYMP Comprehensive (Seq & Del/Dup Analysis) | MNGIE Syndrome | BE, DNA |
| ☐ | 3599 | MOCS1 Comprehensive (Seq & Del/Dup Analysis) | Molybdenum Cofactor Deficiency | BE, DNA |
| ☐ | 3619 | MOCS2 Comprehensive (Seq & Del/Dup Analysis) | Molybdenum Cofactor Deficiency | BE, DNA |
| ☐ | 29410 | MPL Sequence Analysis by NGS | MPL-Related Disorders | BE, DNA |
| ☐ | 6045 | MTHFR 677 C>T Variant Analysis | MTHFR Deficiency | BE, DNA |
| ☐ | 29395 | MCOLN1 Sequence Analysis by NGS | Mucolipidosis IV | BE, DNA |
| ☐ | 6385 | Type I (MPS I), IDUA Sequence Analysis | Mucopolysaccharidosis | BE |
| ☐ | 6814 | Type II (MPS II), IDS Comprehensive (Seq & Del/Dup w/Inv Analysis) | Mucopolysaccharidosis | BE, DNA |
| ☐ | 3604 | Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) Comprehensive Panel (Seq & Del/Dup Analysis) (ETFA, ETFB, ETFDH) | Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) | BE, DNA |
| ☐ | 3859 | ETFA Comprehensive (Seq & Del/Dup Analysis) | Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) | BE, DNA |
| ☐ | 3864 | ETFB Comprehensive (Seq & Del/Dup Analysis) | Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) | BE, DNA |
| ☐ | 3844 | ETFDH Comprehensive (Seq & Del/Dup Analysis) | Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) | BE, DNA |
| ☐ | 6041 | Myotonic Dystrophy Type 1 Repeat Expansion Analysis | Myotonic Dystrophy Type 1 | BE, DNA |
| ☐ | 3354 | NAGS Comprehensive (Seq & Del/Dup Analysis) | N-Acetyglutamate Synthase (NAGS) Deficiency | BE, DNA |
| ☐ | 7523 | LMX1B Comprehensive (Seq & Del/Dup Analysis) | Nail-Patella Syndrome | BE, DNA |
| ☐ | 29415 | Type 1, NPHS1 Sequence Analysis by NGS | Nephrotic Syndrome | BE, DNA |
| ☐ | 29420 | Type 2, NPHS2 Sequence Analysis by NGS | Nephrotic Syndrome | BE, DNA |
| ☐ | 29435 | Type 1, PPT1 Sequence Analysis by NGS | Neuronal Ceroid Lipofuscinosis | BE, DNA |
| ☐ | 29305 | Type 3, CLN3 Sequence Analysis by NGS | Neuronal Ceroid Lipofuscinosis | BE, DNA |
| ☐ | 29310 | Type 5, CLN5 Sequence Analysis by NGS | Neuronal Ceroid Lipofuscinosis | BE, DNA |
| ☐ | 6555 | NPC1 Sequence Analysis | Niemann-Pick Disease Type C | BE, DNA |
| ☐ | 6560 | NPC2 Sequence Analysis | Niemann-Pick Disease Type C | BE, DNA |
| ☐ | 6900 | SHOC2 Sequence Analysis | Noonan-like Syndrome | BE, DNA |
| ☐ | 20205 | NPHP1 Sequence Analysis by NGS | NPHP1-Related Disorders | BE, DNA |
| ☐ | 20215 | NPHP3 Sequence Analysis by NGS | NPHP3-Related Disorders | BE, DNA |
| ☐ | 20220 | NPHP4 Sequence Analysis by NGS | NPHP4-Related Disorders | BE, DNA |
| ☐ | 6845 | LEP Sequence Analysis | Obesity, Monogenic Nonsyndromic | BE, DNA |
| ☐ | 6850 | LEPR Sequence Analysis | Obesity, Monogenic Nonsyndromic | BE, DNA |
| ☐ | 6855 | PCSK1 Sequence Analysis | Obesity, Monogenic Nonsyndromic | BE, DNA |
| ☐ | 6860 | POMC Sequence Analysis | Obesity, Monogenic Nonsyndromic | BE, DNA |
| ☐ | 6083 | X-Linked, GPR143 Comprehensive (Seq & Del/Dup Analysis) | Oculocutaneous Albinism | BE, DNA |
| ☐ | 3469 | Type 1, OPA1 Comprehensive (Seq & Del/Dup Analysis) | Optic Atrophy Type 1 | BE, DNA |
| ☐ | 3529 | Type 3, OPA3 Comprehensive (Seq & Del/Dup Analysis) | Optic Atrophy Type 3 | BE, DNA |
| ☐ | 3144 | OTC Comprehensive (Seq & Del/Dup Analysis) | Ornithine Transcarbamylase (OTC) Deficiency | BE, DNA |
| ☐ | 2574 | AMER1 Comprehensive (Seq & Del/Dup Analysis) | Osteopathia Striata with Cranial Sclerosis | BE, DNA |
| ☐ | 2614 | CLCN7 Comprehensive (Seq & Del/Dup Analysis) | Osteopetrosis | BE, DNA |
| ☐ | 2624 | TCIRG1 Comprehensive (Seq & Del/Dup Analysis) | Osteopetrosis | BE, DNA |
| ☐ | 2604 | CA2 Comprehensive (Seq & Del/Dup Analysis) | Osteopetrosis with Renal Tubular Acidosis | BE, DNA |
| ☐ | 6885 | PCDH19 Sequence Analysis | PCDH19-Related X Linked Female-Limited Epilepsy w/MR | BE, DNA |
| ☐ | 3169 | PDHA1 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) | Biological Sex

## MOLECULAR DIAGNOSTIC TESTS

### SINGLE GENE ANALYSIS

| | TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|---|
| ☐ | 3899 | PDHB Comprehensive (Seq & Del/Dup Analysis) | PDH Complex Deficiency | BE, DNA |
| ☐ | 3924 | PDHX Comprehensive (Seq & Del/Dup Analysis) | PDH Complex Deficiency | BE, DNA |
| ☐ | 3894 | PDP1 Comprehensive (Seq & Del/Dup Analysis) | PDH Complex Deficiency | BE, DNA |
| ☐ | 6550 | GJC2 Sequence Analysis | Pelizaeus-Merzbacher-Like Disease | BE, DNA |
| ☐ | 22105 | ACOX1 Sequence Analysis by NGS | Peroxisomal Acyl-CoA Oxidase Deficiency | BE, DNA |
| ☐ | 5365 | PGM3 Sequence Analysis | PGM3-Related Disorders | BE, DNA |
| ☐ | 3139 | PAH Comprehensive (Seq & Del/Dup Analysis) | Phenylalanine Hydroxylase Deficiency (PKU) | BE, DNA |
| ☐ | 29045 | Cytostolic, PCK1 Sequence Analysis by NGS | Phosphoenolpyruvate Carboxykinase Deficiency | BE, DNA |
| ☐ | 29050 | Mitochondrial, PCK2 Sequence Analysis by NGS | Phosphoenolpyruvate Carboxykinase Deficiency | BE, DNA |
| ☐ | 6149 | PLP1 Comprehensive (Seq & Del/Dup Analysis) | PLP1-Related Disorders | BE, DNA |
| ☐ | 29425 | PKHD1 Sequence Analysis by NGS | Polycystic Kidney and Hepatic Disease | BE, DNA |
| ☐ | 3729 | RARS2 Comprehensive (Seq & Del/Dup Analysis) | Pontocerebellar Hypoplasia Type 6 | BE, DNA |
| ☐ | 6050 | Prader-Willi Syndrome Methylation Analysis | Prader-Willi Syndrome | BE, DNA |
| ☐ | 7105 | MAGEL2 Sequence Analysis | Prader-Willi-like Syndrome; Intellectual Disability; Autism | BE, DNA |
| ☐ | 3622 | Propionic Acidemia Comprehensive Panel (Seq & Del/Dup Analysis) (PCCA & PCCB) | Propionic Acidemia | BE, DNA |
| ☐ | 3769 | PCCA Comprehensive (Seq & Del/Dup Analysis) | Propionic Acidemia | BE, DNA |
| ☐ | 3774 | PCCB Comprehensive (Seq & Del/Dup Analysis) | Propionic Acidemia | BE, DNA |
| ☐ | 29545 | Type 1, AGXT Sequence Analysis by NGS | Primary Hyperoxaluria | BE, DNA |
| ☐ | 29365 | Type 2, GRHPR Sequence Analysis by NGS | Primary Hyperoxaluria | BE, DNA |
| ☐ | 6048 | Prothrombin Mutation Panel (F2) | Prothrombin | BE, DNA |
| ☐ | 6790 | PTEN Comprehensive (Seq & Del/Dup Analysis) | PTEN-Related Disorders | BE, DNA |
| ☐ | 5025 | PNP Sequence Analysis | Purine Nucleoside Phosphorylase Deficiency | BE, DNA |
| ☐ | 2444 | CTSK Comprehensive (Seq & Del/Dup Analysis) | Pycnodysostosis | BE, DNA |
| ☐ | 6950 | ALDH7A1 Sequence Analysis | Pyridoxine-Dependent Seizures | BE, DNA |
| ☐ | 3919 | DLAT Comprehensive (Seq & Del/Dup Analysis) | Pyruvate Dehydrogenase E2 Deficiency | BE, DNA |
| ☐ | 3754 | PC Comprehensive (Seq & Del/Dup Analysis) | Pyruvate Carboxylase Deficiency | BE, DNA |
| ☐ | 5300 | RAG2 Sequence Analysis | RAG2-Related Disorders | BE, DNA |
| ☐ | 22215 | PHYH Sequence Analysis by NGS | Refsum Disease | BE, DNA |
| ☐ | 6736 | MECP2 Comprehensive (Seq & Del/Dup Analysis) | Rett Syndrome (MECP2-Related Disorders) | BE, DNA |
| ☐ | 6635 | FOXG1 Sequence Analysis | Rett Syndrome, Congenital Variant | BE, DNA |
| ☐ | 22125 | Type 2, GNPAT Sequence Analysis by NGS | Rhizomelic Chondrodysplasia Punctata | BE, DNA |
| ☐ | 22110 | Type 3, AGPS Sequence Analysis by NGS | Rhizomelic Chondrodysplasia Punctata | BE, DNA |
| ☐ | 6565 | VDR Sequence Analysis | Rickets-Alopecia Syndrome | BE, DNA |
| ☐ | 6758 | CREBBP Comprehensive (Seq & Del/Dup Analysis) | Rubinstein-Taybi Syndrome | BE, DNA |
| ☐ | 3929 | ACADS Comprehensive (Seq & Del/Dup Analysis) | SCAD Deficiency | BE, DNA |
| ☐ | 6285 | COL10A1 Sequence Analysis | Schmid Metaphyseal Chondrodysplasia (SMCD) | BE, DNA |
| ☐ | 29515 | TH Sequence Analysis by NGS | Segawa Syndrome Recessive | BE, DNA |
| ☐ | 29105 | SERPINA1 Sequence Analysis by NGS | SERPINA1-Related Disorders | BE, DNA |
| ☐ | 6053 | Sickle Cell Disease Mutation Analysis | Sickle Cell Disease | BE, DNA |
| ☐ | 29550 | ALDH3A2 Sequence Analysis by NGS | Sjogren-Larsson Syndrome | 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) | Biological Sex

## MOLECULAR DIAGNOSTIC TESTS

### SINGLE GENE ANALYSIS

| | TEST CODE | TEST NAME | DISORDER | SAMPLE TYPE * |
|---|---|---|---|---|
| ☐ | 21730 | SLC16A1 (HHF7) Sequence Analysis by NGS | SLC16A1-Related Disorders | BE, DNA |
| ☐ | 29485 | SLC17A5 (NSD) Sequence Analysis by NGS | SLC17A5-Related Disorders | BE, DNA |
| ☐ | 29490 | SLC26A2 (DTDST) Sequence Analysis by NGS | SLC26A2-Related Disorders | BE, DNA |
| ☐ | 29495 | SLC26A4 (PENDRIN) Sequence Analysis by NGS | SLC26A4-Related Disorders | BE, DNA |
| ☐ | 6745 | DHCR7 Sequence Analysis | Smith-Lemli-Opitz Syndrome | BE, DNA |
| ☐ | 6760 | RAI1 Sequence Analysis | Smith-Magenis Syndrome | BE, DNA |
| ☐ | 29505 | SMPD1 Sequence Analysis by NGS | SMPD1-Related Disorders | BE, DNA |
| ☐ | 29455 | SACS Sequence Analysis by NGS | Spastic Ataxia Charlevoix-Saguenay Type | BE, DNA |
| ☐ | 6059 | SMN1/SMN2 Copy Number Analysis | Spinal Muscular Atrophy (SMA) Diagnostic Test | BE, DNA |
| ☐ | 2899 | PRKCG Comprehensive (Seq & Del/Dup Analysis) | Spinocerebellar Ataxia 14 (SCA14) | BE, DNA |
| ☐ | 29210 | SRD5A3 Sequence Analysis by NGS | SRD5A3-Related Disorders | BE, DNA |
| ☐ | 6060 | SRY Molecular Analysis | SRY-Related Phenotypes | BE, DNA |
| ☐ | 5024 | ALDH5A1 Comprehensive (Seq & Del/Dup Analysis) | Succinic Semialdehyde Dehydrogenase Deficiency | BE, DNA |
| ☐ | 6062 | Thrombophilia Mutation Panel (F5, MTHFR, F2) | Thrombophilia | BE, DNA |
| ☐ | 20465 | TMEM67 Sequence Analysis by NGS | TMEM67-Related Disorders | BE, DNA |
| ☐ | 29520 | TMEM216 Sequence Analysis by NGS | TMEM216-Related Disorders | BE, DNA |
| ☐ | 2510 | TMLHE Sequence Analysis | TMLHE Deficiency | BE, DNA |
| ☐ | 2513 | TMLHE Exon 2 Deletion Analysis | TMLHE Deficiency | BE, DNA |
| ☐ | 29525 | TPP1 Sequence Analysis by NGS | TPP1-Related Disorders | BE, DNA |
| ☐ | 3969 | TCN2 Comprehensive (Seq & Del/Dup Analysis) | Transcobalamin II Deficiency | BE, DNA |
| ☐ | 3624 | Trifunctional Protein Deficiency Comprehensive Panel (Seq & Del/Dup Analysis) (HADHA and HADHB) | Trifunctional Protein Deficiency | BE, DNA |
| ☐ | 3634 | HADHB Comprehensive (Seq & Del/Dup Analysis) (HADHB) | Trifunctional Protein Deficiency | BE, DNA |
| ☐ | 5005 | TSHR Sequence Analysis | TSHR-Related Disorders | BE, DNA |
| ☐ | 3449 | Type I, FAH Comprehensive (Seq & Del/Dup Analysis) | Tyrosinemia | BE, DNA |
| ☐ | 2084 | Type II, TAT Comprehensive (Seq & Del/Dup Analysis) | Tyrosinemia | BE, DNA |
| ☐ | 29535 | UGT1A1 Sequence Analysis by NGS | UGT1A1-Related Disorders | BE, DNA |
| ☐ | 6650 | USH2A Sequence Analysis | Usher Syndrome 2A | BE, DNA |
| ☐ | 6660 | CLRN1 Sequence Analysis | Usher Syndrome 3A | BE, DNA |
| ☐ | 3359 | ACADVL Comprehensive (Seq & Del/Dup Analysis) | VLCAD Deficiency | BE, DNA |
| ☐ | 2554 | ATP7B Comprehensive (Seq & Del/Dup Analysis) | Wilson Disease | BE, DNA |
| ☐ | 6430 | LIPA Sequence Analysis | Wolman Disease | BE, DNA |

## SAMPLE SPECIFICATIONS TABLE

| ABBREVIATION | SAMPLE NAME | RECOMMENDED AMOUNT (2 YRS - ADULT) | RECOMMENDED AMOUNT (NEWBORN - 2YRS) | SHIPPING INSTRUCTIONS | SPECIAL NOTES |
|---|---|---|---|---|---|
| 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. |