BAYLOR GENETICS
READY WITH ANSWERS

SHIPPING Baylor Genetics
2450 Holcombe Blvd., Grand Blvd. Receiving Dock
Houston, TX 77021-2024
Phone: 713.798.6555
Fax: 713.798.2787

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/YY)

Address | Accession # | Hospital/ Medical Record #

Biological Sex: ◯ M ◯ F ◯ Unknown

City | State | Zip | Phone | Gender identity (if different from above):

REPORTING RECIPIENTS

Ordering Physician | Institution Name

Email (Required for International Clients) | Phone | Fax

◆ **ADDITIONAL RECIPIENTS**

Name | Name

Email | Fax | Email | Fax

PAYMENT (FILL OUT ONE OF THE OPTIONS BELOW)

☐ **SELF PAYMENT**

◯ Bill Patient For Laboratory Testing

☐ **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/YY) | Address of Insured

Patient's Relationship to Insured | Phone of Insured | City | State | Zip

Primary Insurance Co. Name | Primary Insurance Co. Phone | Primary Member Policy # | Primary Member Group #

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 designated insurance carrier any information necessary, including test results, for processing my insurance claim. I also authorize benefits to be payable exclusively to Baylor Genetics. I understand that my insurance carrier may not approve or reimburse my medical genetic services in full or any portion thereof, due to a variety of reasons, including, but not limited to: the contract status of my insurance provider with Baylor Genetics, usual and customary rate limits, benefit exclusions, coverage limits, lack of authorization, or medical necessity. I understand that I am responsible for any co-pay, co-insurance, and unmet deductible that the insurance policy dictates. I understand that I am responsible for 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 Name | Patient's Signature | Date

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

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ETHNICITY

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

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) _____

SAMPLE

Date of Collection (MM/DD/YY)

◆ **SAMPLE TYPE**
- ◯ Blood in EDTA-tube (purple-top)
- ◯ Cord Blood
- ◯ Saliva
- ◯ DNA (Specify) _____
- ◯ Other (Specify) _____

Note: Blood should not be sent from patients who have had a bone marrow transplant or recent blood transfusion

◆ 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

Date of Birth (MM/DD/YY) | Relationship of Proband to Patient

Proband testing location (Select one)

☐ Baylor Genetics
Lab # | Family #

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

MOLECULAR DIAGNOSTIC TESTS

◆ **MASSIVELY PARALLEL SEQUENCING (BCM-MitomeNGS^{SM}) PANELS**

TEST CODE	TEST NAME	SAMPLE TYPE *
☐ 20100	Albinism Panel (13 genes)	BE, DNA
☐ 20400	Bardet-Biedl Syndrome Panel (18 genes)	BE, DNA
☐ 2105	Cholestasis Panel (7 genes)	BE, DNA
☐ 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

TEST CODE	TEST NAME	SAMPLE TYPE *
☐ 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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◆ MASSIVELY PARALLEL SEQUENCING (BCM-MitomeNGS^{SM})

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

TEST CODE	TEST NAME	SAMPLE TYPE *
☐ 21400	Noonan Spectrum Disorders Panel (12 genes)	BE, DNA
☐ 22100	Peroxisomal Disorders Panel (22 genes)	BE, DNA
☐ 5274	Proximal Urea Cycle Disorders (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 Code | Gene | Test Code | Gene

Test Name | Test Name | 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)

Test list continued on next page

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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)

Test list continued on next page

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◆ 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, DNA
☐ 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)

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☐ 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)

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◆ **SINGLE GENE ANALYSIS**

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☐ 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)

Test list continued on next page

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Patient Last Name _____ Patient First Name _____ MI _____ Date of Birth (MM/DD/YY) ___/___/___ Biological Sex _____

◆ **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)

Test list continued on next page

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◆ 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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◆ 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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◆ 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* Deletion 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 - 2 yrs)	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.	
CB	Cord Blood	N/A	1 - 2 cc	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	Ensure properly labeled. Also send 3 cc of maternal blood in properly labeled EDTA tube for MCC studies at no charge as needed.
DNA	DNA, Extracted	10 -15 ug	10 -15 ug	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	Minimal concentration of 50ng/uL; A260/A280 of ~1.7
SA	Saliva	See Special Notes	See Special Notes	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.	Collected with Oragene DNA Self-Collection Kit.