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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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)

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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)

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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)

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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)

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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 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 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 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

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MOLECULAR DIAGNOSTIC TESTS

SINGLE GENE ANALYSIS

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☐	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.