

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY			
				ACOG†	ACMG	H445	H569‡	H574‡	H613
17-Beta Hydroxysteroid Dehydrogenase 3 Deficiency	<i>HSD17B3</i>	•				•	•	•	•
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	<i>HSD3B2</i>	•				•	•	•	•
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	<i>HMGCL</i>	•				•	•	•	•
3-Hydroxyacyl-CoA Dehydrogenase Deficiency	<i>HADH</i>	•				•	•	•	•
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	<i>MCCC1*</i>	•				•	•	•	•
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	<i>MCCC2*</i>	•			o	•	•	•	•
3-Phosphoglycerate Dehydrogenase Deficiency	<i>PHGDH</i>	•				•	•	•	•
5-Alpha-Reductase Deficiency	<i>SRD5A2</i>	•				•	•	•	•
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	<i>PTS</i>	•				•	•	•	•
ABCA4-Related Conditions	<i>ABCA4</i>	•				•	•	•	•
Abetalipoproteinemia	<i>MTTP</i>	•				•	•	•	•
Achondrogenesis, Type 1B	<i>SLC26A2</i>	•			o	•	•	•	•
Achromatopsia, CNGB3-Related	<i>CNGB3</i>	•			o	•	•	•	•
Acrodermatitis Enteropathica	<i>SLC39A4</i>	•				•	•	•	•
Action Myoclonus-Renal Failure (AMRF) Syndrome	<i>SCARB2</i>	•				•			•
Acute Infantile Liver Failure, TRMU-Related	<i>TRMU</i>	•				•	•	•	•
Acyl-CoA Oxidase I Deficiency	<i>ACOX1</i>	•				•	•	•	•
Adrenal Hypoplasia Congenita, X-Linked	<i>NR0B1</i>		•		o	•	•	•	•
Adrenoleukodystrophy, X-Linked	<i>ABCD1</i>		•		o	•	•	•	•
Agammaglobulinemia, X-Linked	<i>BTK</i>		•			•	•	•	•
Aicardi-Goutières Syndrome	<i>SAMHD1</i>	•				•	•	•	•
Aicardi-Goutières Syndrome, RNASEH2A-Related	<i>RNASEH2A</i>	•				•	•	•	•
Aicardi-Goutières Syndrome, RNASEH2B-Related	<i>RNASEH2B</i>	•			o	•	•	•	•
Aicardi-Goutières Syndrome, RNASEH2C-Related	<i>RNASEH2C</i>	•				•	•	•	•
Aicardi-Goutières Syndrome, TREX1-Related	<i>TREX1</i>	•				•	•	•	•
Alkaptonuria	<i>HGD*</i>	•					•	•	•
Alpha-1 Antitrypsin Deficiency	<i>SERPINA1*</i>	•				•	•	•	•
Alpha-Mannosidosis	<i>MAN2B1</i>	•				•	•	•	•
Alpha-Thalassemia	<i>HBA1/HBA2</i>	•			o	•	•	•	•
Alpha-Thalassemia Intellectual Disability Syndrome	<i>ATRX</i>		•			•	•	•	•
Alport Syndrome, COL4A3-Related	<i>COL4A3</i>	•				•	•	•	•
Alport Syndrome, COL4A4-Related	<i>COL4A4</i>	•				•	•	•	•
Alport Syndrome, X-Linked	<i>COL4A5</i>		•			•	•	•	•
Alstrom Syndrome	<i>ALMS1</i>	•				•	•	•	•
Amish Infantile Epilepsy Syndrome	<i>ST3GAL5</i>	•				•	•	•	•
Andermann Syndrome	<i>SLC12A6</i>	•				•	•	•	•
Androgen Insensitivity Syndrome	<i>AR</i>		•				•	•	•
Arginine:Glycine Amidinotransferase Deficiency (Agat Deficiency)	<i>GATM</i>	•					•	•	•
Argininemia	<i>ARG1</i>	•				•	•	•	•
Argininosuccinate Lyase Deficiency	<i>ASL</i>	•			o	•	•	•	•
Aromatase Deficiency	<i>CYP19A1</i>	•				•	•	•	•
Arts Syndrome	<i>PRPS1</i>		•			•	•	•	•
Asparagine Synthetase Deficiency	<i>ASNS</i>	•				•	•	•	•
Aspartylglycosaminuria	<i>AGA</i>	•			o	•	•	•	•
Ataxia with Vitamin E Deficiency	<i>TTPA</i>	•				•	•	•	•
Ataxia-Telangiectasia	<i>ATM</i>	•				•	•	•	•
Ataxia-Telangiectasia-Like Disorder 1	<i>MRE11</i>	•				•	•	•	•
Atransferrinemia	<i>TF</i>				o	•	•	•	•
Autism Spectrum, Epilepsy and Arthrogyrosis	<i>SLC35A3</i>	•				•	•	•	•
Autoimmune Polyglandular Syndrome, Type 1	<i>AIRE</i>	•			o	•	•	•	•
Autosomal Recessive Congenital Ichthyosis (ARCI), SLC27A4-Related	<i>SLC27A4</i>	•				•	•	•	•
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	<i>SACS</i>	•				•	•	•	•
Bardet-Biedl Syndrome, ARL6-Related	<i>ARL6</i>	•					•	•	•
Bardet-Biedl Syndrome, BBS10-Related	<i>BBS10</i>	•				•	•	•	•
Bardet-Biedl Syndrome, BBS12-Related	<i>BBS12</i>	•				•	•	•	•
Bardet-Biedl Syndrome, BBS1-Related	<i>BBS1</i>	•			o	•	•	•	•
Bardet-Biedl Syndrome, BBS2-Related	<i>BBS2</i>	•			o	•	•	•	•
Bardet-Biedl Syndrome, BBS4-Related	<i>BBS4</i>	•				•	•	•	•
Bardet-Biedl Syndrome, Bbs5-Related	<i>BBS5</i>	•					•	•	•
Bardet-Biedl Syndrome, BBS7-Related	<i>BBS7</i>	•				•	•	•	•
Bardet-Biedl Syndrome, BBS9-Related	<i>BBS9</i>	•				•	•	•	•
Bardet-Biedl Syndrome, TTC8-Related	<i>TTC8</i>	•				•	•	•	•
Bare Lymphocyte Syndrome, CIITA-Related	<i>CIITA</i>	•				•	•	•	•
Barth Syndrome	<i>TAZ</i>		•			•	•	•	•
Bartter Syndrome, BSND-Related	<i>BSND</i>	•				•	•	•	•
Bartter Syndrome, KCNJ1-Related	<i>KCNJ1</i>	•					•	•	•

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Bartter Syndrome, SLC12A1-Related	SLC12A1	•					•	•	•	•
Batten Disease, CLN3-Related	CLN3	•					•	•	•	•
Bernard-Soulier Syndrome, Type A1/A2	GP1BA*	•					•	•	•	•
Bernard-Soulier Syndrome, Type C	GP9*	•					•	•	•	•
Beta-Hemoglobinopathies	HBB	•		o	o		•	•	•	•
Beta-Mannosidosis	MANBA	•					•	•	•	•
Beta-Ureidopropionase Deficiency	UPB1	•					•			•
Bilateral Frontoparietal Polymicrogyria	GPR56	•					•	•	•	•
Biotinidase Deficiency	BTD*	•			o		•	•	•	•
Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD)	SLC19A3				o		•	•	•	•
Bloom Syndrome	BLM	•		o	o		•	•	•	•
Brittle Cornea Syndrome 1	ZNF469	•						•	•	•
Brittle Cornea Syndrome 2	PRDM5	•						•	•	•
Canavan Disease	ASPA	•		o	o		•	•	•	•
Carbamoyl Phosphate Synthetase I Deficiency	CPS1	•					•	•	•	•
Carnitine Deficiency	SLC22A5	•					•	•	•	•
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	•					•	•	•	•
Carnitine Palmitoyltransferase II Deficiency	CPT2	•			o		•	•	•	•
Carnitine-Acylcarnitine Translocase Deficiency	SLC25A20	•					•	•	•	•
Carpenter Syndrome	RAB23	•					•	•	•	•
Cartilage-Hair Hypoplasia	RMRP	•					•	•	•	•
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	•					•	•	•	•
CD59-Mediated Hemolytic Anemia	CD59	•						•	•	•
CEP152-Related Microcephaly	CEP152	•						•	•	•
Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma (CEDNIK) Syndrome	SNAP29	•						•	•	•
Cerebrotendinous Xanthomatosis	CYP27A1	•			o		•	•	•	•
Charcot-Marie-Tooth Disease with Deafness, X-Linked	GJB1		•				•	•	•	•
Charcot-Marie-Tooth Disease, Recessive Intermediate C	PLEKHG5	•						•	•	•
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	•					•	•	•	•
Chediak-Higashi Syndrome	LYST	•					•	•	•	•
Choreoacanthocytosis	VPS13A	•					•	•	•	•
Choroideremia	CHM		•				•	•	•	•
Chronic Granulomatous Disease, CYBA-Related	CYBA	•					•	•	•	•
Chronic Granulomatous Disease, NCF2-Related	NCF2	•						•	•	•
Chronic Granulomatous Disease, X-Linked	CYBB		•				•	•	•	•
Ciliopathies, RPGRIPL-Related	RPGRIPL	•					•	•	•	•
Citrin Deficiency	SLC25A13	•					•	•	•	•
Citrullinemia, Type 1	ASS1	•					•	•	•	•
CLN10 Disease	CTSD	•					•	•	•	•
Cohen Syndrome	VPS13B	•					•	•	•	•
COL11A2-Related Conditions	COL11A2	•						•	•	•
Combined Malonic and Methylmalonic Aciduria	ACSF3	•					•	•	•	•
Combined Oxidative Phosphorylation Deficiency 1	GFM1	•					•	•	•	•
Combined Oxidative Phosphorylation Deficiency 3	TSM	•					•	•	•	•
Combined Pituitary Hormone Deficiency 1	POU1F1	•						•	•	•
Combined Pituitary Hormone Deficiency-2	PROP1	•					•	•	•	•
Congenital Adrenal Hyperplasia, 11-Beta-Hydroxylase Deficiency	CYP11B1	•					•	•	•	•
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	CYP17A1	•					•	•	•	•
Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency	CYP21A2	•			o		•	•	•	•
Congenital Adrenal Insufficiency, CYP11A1-Related	CYP11A1				o		•	•	•	•
Congenital Amegakaryocytic Thrombocytopenia	MPL	•					•	•	•	•
Congenital Chronic Diarrhea	DGAT1	•						•	•	•
Congenital Disorder Of Glycosylation Type 1, ALG1-Related	ALG1	•						•	•	•
Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	PMM2	•			o		•	•	•	•
Congenital Disorder of Glycosylation, Type 1B	MPI	•					•	•	•	•
Congenital Disorder of Glycosylation, Type 1C	ALG6	•					•	•	•	•
Congenital Dyserythropoietic Anemia Type 2	SEC23B	•						•	•	•
Congenital Finnish Nephrosis	NPHS1	•			o		•	•	•	•
Congenital Hydrocephalus 1	CCDC88C				o		•	•	•	•
Congenital Hyperinsulinism, KCNJ11-Related	KCNJ11	•					•	•	•	•
Congenital Insensitivity to Pain with Anhidrosis (CIPA)	NTRK1	•					•	•	•	•
Congenital Myasthenic Syndrome, CHAT-Related	CHAT	•					•	•	•	•
Congenital Myasthenic Syndrome, CHRNE-Related	CHRNE	•			o		•	•	•	•
Congenital Myasthenic Syndrome, COLQ-Related	COLQ	•					•	•	•	•
Congenital Myasthenic Syndrome, DOK7-Related	DOK7	•					•	•	•	•
Congenital Myasthenic Syndrome, RAPSN-Related	RAPSN	•					•	•	•	•
Congenital Nephrotic Syndrome, PLCE1-Related	PLCE1	•					•	•	•	•
Congenital Neutropenia, G6PC3-Related	G6PC3	•					•	•	•	•
Congenital Neutropenia, HAX1-Related	HAX1	•					•	•	•	•
Congenital Neutropenia, VPS45-Related	VPS45	•					•	•	•	•

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Congenital Secretory Chloride Diarrhea 1	SLC26A3	•					•	•	•	•
Corneal Dystrophy and Perceptive Deafness	SLC4A11	•					•	•	•	•
Corticosterone Methylxidase Deficiency	CYP11B2	•					•	•	•	•
Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3)	OPA3	•					•	•	•	•
Cowchock Syndrome	AIFM1		•				•			•
CRB1-Related Retinal Dystrophies	CRB1	•					•	•	•	•
Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1, X-Linked)	SLC6A8		•		o		•	•	•	•
Cystic Fibrosis	CFTR	•		o	o		•	•	•	•
Cystinosis	CTNS	•					•	•	•	•
Cytochrome C Oxidase Deficiency, PET100-Related	PET100	•					•	•	•	•
Cytochrome P450 Oxidoreductase Deficiency	POR	•						•	•	•
D-Bifunctional Protein Deficiency	HSD17B4	•					•	•	•	•
Deafness, Autosomal Recessive 77	LOXHD1	•					•	•	•	•
Dent Disease, Type 1	CLCN5		•				•			•
Dent Disease, Type 2 / Lowe Syndrome	OCRL		•				•	•	•	•
Developmental And Epileptic Encephalopathy 36	ALG13		•					•	•	•
Dihydropteridine Reductase (DHPR) Deficiency	QDPR	•						•	•	•
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	•					•			•
Donnai-Barrow Syndrome	LRP2				o		•	•	•	•
Dubin-Johnson Syndrome	ABCC2	•						•	•	•
Duchenne/Becker Muscular Dystrophy	DMD		•		o		•	•	•	•
Dyskeratosis Congenita Spectrum Disorders	TERT	•						•	•	•
Dyskeratosis Congenita, DKC1-Related	DKC1		•				•	•	•	•
Dyskeratosis Congenita, RTEL1-Related	RTEL1	•					•	•	•	•
Dystrophic Epidermolysis Bullosa, COL7A1-Related	COL7A1	•			o		•	•	•	•
Early Infantile Epileptic Encephalopathy, CAD-Related	CAD	•						•	•	•
Ehlers-Danlos Syndrome Type VI	PLOD1	•						•	•	•
Ehlers-Danlos Syndrome, Classic-Like, TNXB-Related	TNXB				o		•		•	•
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2	•					•	•	•	•
Ellis-van Creveld Syndrome, EVC2-Related	EVC2	•			o		•	•	•	•
Ellis-van Creveld Syndrome, EVC-Related	EVC	•					•	•	•	•
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	EMD		•				•	•	•	•
Emery-Dreifuss Muscular Dystrophy 6, X Linked	FHL1		•					•	•	•
Enhanced S-Cone Syndrome	NR2E3	•					•	•	•	•
Epimerase Deficiency (Galactosemia Type III)	GALE	•						•	•	•
Epiphyseal Dysplasia, Multiple, 7 / Desbuquois Dysplasia 1	CANT1	•					•	•	•	•
ERCC6-Related Disorders	ERCC6	•					•	•	•	•
ERCC8-Related Disorders	ERCC8	•					•	•	•	•
Ethylmalonic Encephalopathy	ETHE1	•					•	•	•	•
Fabry Disease	GLA		•		o		•	•	•	•
Factor IX Deficiency	F9		•		o		•	•	•	•
Factor V Deficiency	F5*	•						•	•	•
Factor XI Deficiency	F11*	•					•	•	•	•
Familial Dysautonomia	IKBKAP	•		o	o		•	•	•	•
Familial Hemophagocytic Lymphohistiocytosis, PRF1-Related	PRF1	•			o		•	•	•	•
Familial Hemophagocytic Lymphohistiocytosis, STX11-Related	STX11	•					•	•	•	•
Familial Hemophagocytic Lymphohistiocytosis, STXBP2-Related	STXBP2	•					•	•	•	•
Familial Hemophagocytic Lymphohistiocytosis, UNC13D-Related	UNC13D	•						•	•	•
Familial Hypercholesterolemia, LDLRAP1-Related	LDLRAP1	•					•	•	•	•
Familial Hypercholesterolemia, LDLR-Related	LDLR	•					•	•	•	•
Familial Hyperinsulinism, ABCC8-Related	ABCC8	•		o	o		•	•	•	•
Familial Mediterranean Fever	MEFV*	•					•	•	•	•
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	AQP2	•					•	•	•	•
Fanconi Anemia Group J	BRIP1	•						•	•	•
Fanconi Anemia, Group A	FANCA	•		o			•	•	•	•
Fanconi Anemia, Group B	FANCB		•				•	•	•	•
Fanconi Anemia, Group C	FANCC	•		o	o		•	•	•	•
Fanconi Anemia, Group D2	FANCD2	•					•	•	•	•
Fanconi Anemia, Group E	FANCE	•					•	•	•	•
Fanconi Anemia, Group F	FANCF	•					•	•	•	•
Fanconi Anemia, Group G	FANCG	•		o			•	•	•	•
Fanconi Anemia, Group I	FANCI	•					•	•	•	•
Fanconi Anemia, Group L	FANCL	•					•	•	•	•
Farber Lipogranulomatosis	ASAH1	•					•			•
Foveal Hypoplasia	SLC38A8	•						•	•	•
Fragile X Syndrome	FMR1		•	o	o		•	•	•	•
Fragile XE Syndrome	AFF2		•		o		•		•	•
Fraser Syndrome 3, GRIP1-Related	GRIP1				o		•	•	•	•
Fraser Syndrome, FRAS1-Related	FRAS1	•						•	•	•
Fraser Syndrome, FREM2-Related	FREM2	•						•	•	•

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				ACOG†	ACMG	H445	H568†	H574†	H613
Friedreich Ataxia	<i>FXN</i>				o	•		•	•
Fructose-1,6-Bisphosphatase Deficiency	<i>FBP1</i>	•					•	•	•
Fucosidosis	<i>FUCA1</i>	•					•	•	•
Fumarase Deficiency	<i>FH</i>	•				•	•	•	•
GABA-Transaminase Deficiency	<i>ABAT</i>	•				•			•
Galactokinase Deficiency (Galactosemia, Type II)	<i>GALK1</i>	•				•	•	•	•
Galactosemia	<i>GALT</i>	•			o	•	•	•	•
Galactosialidosis	<i>CTSA</i>	•				•	•	•	•
Gaucher Disease	<i>GBA</i>	•			o	•	•	•	•
GCH1-Related Conditions	<i>GCH1</i>	•					•	•	•
GDF5-Related Conditions	<i>GDF5</i>	•					•	•	•
Geroderma Osteodysplastica	<i>GORAB</i>	•					•	•	•
Gitelman Syndrome	<i>SLC12A3</i>	•				•	•	•	•
Glanzmann Thrombasthenia	<i>ITGB3</i>	•					•	•	•
Glucose-6-Phosphate Dehydrogenase Deficiency	<i>G6PD*</i>		•			•	•	•	•
Glutaric Acidemia, Type 1	<i>GCDH</i>	•				•	•	•	•
Glutaric Acidemia, Type 2A	<i>ETFA</i>	•				•	•	•	•
Glutaric Acidemia, Type 2B	<i>ETFB</i>	•				•	•	•	•
Glutaric Acidemia, Type 2C	<i>ETFDH</i>	•				•	•	•	•
Glutathione Synthetase Deficiency	<i>GSS</i>	•					•	•	•
Glycine Encephalopathy, AMT-Related	<i>AMT</i>	•				•	•	•	•
Glycine Encephalopathy, GLDC-Related	<i>GLDC</i>	•				•	•	•	•
Glycogen Storage Disease Type IXb	<i>PHKB</i>	•					•	•	•
Glycogen Storage Disease Type IXc	<i>PHKG2</i>	•					•	•	•
Glycogen Storage Disease, Type 1A	<i>G6PC</i>	•			o	o	•	•	•
Glycogen Storage Disease, Type 1B	<i>SLC37A4</i>	•			o	o	•	•	•
Glycogen Storage Disease, Type 2 (Pompe Disease)	<i>GAA</i>	•				o	•	•	•
Glycogen Storage Disease, Type 3	<i>AGL</i>	•					•	•	•
Glycogen Storage Disease, Type 4	<i>GBE1</i>	•			o		•	•	•
Glycogen Storage Disease, Type 5 (McArdle Disease)	<i>PYGM</i>	•					•	•	•
Glycogen Storage Disease, Type 7	<i>PFKM</i>	•					•	•	•
GRACILE Syndrome	<i>BCS1L</i>	•					•	•	•
Guanidinoacetate Methyltransferase Deficiency	<i>GAMT</i>	•					•	•	•
Harlequin Ichthyosis	<i>ABCA12</i>	•					•	•	•
Heme Oxygenase 1 Deficiency	<i>HMOX1</i>	•					•	•	•
Hemochromatosis, Type 2A	<i>HFE2</i>	•					•	•	•
Hemochromatosis, Type 3, TFR2-Related	<i>TFR2</i>	•					•	•	•
Hemophilia A	<i>F8</i>		•		o		•	•	•
Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Related	<i>MPV17</i>	•					•	•	•
Hereditary Fructose Intolerance	<i>ALDOB</i>	•			o		•	•	•
Hereditary Hemochromatosis Type 1	<i>HFE*</i>	•					•	•	•
Hereditary Hemochromatosis Type 2B	<i>HAMP</i>	•					•	•	•
Hereditary Spastic Paraparesis, Type 49	<i>TECPR2</i>	•					•	•	•
Hereditary Spastic Paraplegia, CYP7B1-Related	<i>CYP7B1</i>	•					•	•	•
Hermansky-Pudlak Syndrome, AP3B1-Related	<i>AP3B1</i>	•					•		•
Hermansky-Pudlak Syndrome, BLOC1S3-Related	<i>BLOC1S3</i>	•					•	•	•
Hermansky-Pudlak Syndrome, BLOC1S6-Related	<i>BLOC1S6</i>	•					•	•	•
Hermansky-Pudlak Syndrome, HPS1-Related	<i>HPS1</i>	•				o	•	•	•
Hermansky-Pudlak Syndrome, HPS3-Related	<i>HPS3</i>	•				o	•	•	•
Hermansky-Pudlak Syndrome, HPS4-Related	<i>HPS4</i>	•					•	•	•
Hermansky-Pudlak Syndrome, HPS5-Related	<i>HPS5</i>	•					•	•	•
Hermansky-Pudlak Syndrome, HPS6-Related	<i>HPS6</i>	•					•	•	•
Heterotaxy Syndrome, ZIC3-Related	<i>ZIC3</i>		•				•		•
Holocarboxylase Synthetase Deficiency	<i>HLCS</i>	•					•	•	•
Homocystinuria And Megaloblastic Anemia Type Cblg	<i>MTR</i>	•					•	•	•
Homocystinuria due to Deficiency of MTHFR	<i>MTHFR</i>	•					•	•	•
Homocystinuria, CBS-Related	<i>CBS</i>	•			o		•	•	•
Homocystinuria, Type cblE	<i>MTRR</i>	•					•	•	•
HSD10 Disease	<i>HSD17B10</i>		•				•	•	•
Hydrolethalus Syndrome	<i>HYLS1</i>	•					•	•	•
Hyper IgM Syndrome, X-Linked	<i>CD40LG</i>		•				•	•	•
Hyper-Igm Immunodeficiency	<i>CD40</i>	•					•	•	•
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH Syndrome)	<i>SLC25A15</i>	•					•	•	•
Hyperphosphatemic Familial Tumoral Calcinosis, GALNT3-Related	<i>GALNT3</i>	•					•	•	•
Hypohidrotic Ectodermal Dysplasia, X-Linked	<i>EDA</i>		•				•	•	•
Hypomyelinating Leukodystrophy 12	<i>VPS11</i>	•					•	•	•
Hypophosphatasia, ALPL-Related	<i>ALPL</i>	•			o		•	•	•
Imerslund-Gräsbeck Syndrome 2	<i>AMN</i>	•					•	•	•
Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome	<i>FOXP3</i>		•				•		•
Immunodeficiency-Centromeric Instability-Facial Anomalies (ICF) Syndrome, DNMT3B-Related	<i>DNMT3B</i>	•					•	•	•

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY				
				ACOG†	ACMG	H445	H568†	H574†	H613	
Immunodeficiency-Centromeric Instability-Facial Anomalies (ICF) Syndrome, ZBTB24-Related	ZBTB24	•					•	•	•	•
Inclusion Body Myopathy 2	GNE	•					•	•	•	•
Infantile Cerebral and Cerebellar Atrophy	MED17	•					•	•	•	•
Infantile Nephronophthisis	INVS	•					•	•	•	•
Infantile Neuroaxonal Dystrophy	PLA2G6	•					•	•	•	•
Infantile Spinal Muscular Atrophy, X-Linked	UBA1		•				•	•	•	•
Isolated Ectopia Lentis	ADAMTSL4	•						•	•	•
Isolated Lissencephaly Sequence / Subcortical Band Heterotopia	DCX		•				•			•
Isolated Sulfite Oxidase Deficiency	SUOX	•						•	•	•
Isolated Thyroid-Stimulating Hormone Deficiency	TSHB	•						•	•	•
Isovaleric Acidemia	IVD	•					•	•	•	•
Johanson-Blizzard Syndrome	UBR1	•					•	•	•	•
Joubert Syndrome 2 / Meckel Syndrome 2	TMEM216	•		o	o		•	•	•	•
Joubert Syndrome And Related Disorders (JSRD), TMEM67-Related	TMEM67	•						•	•	•
Joubert Syndrome, AHI1-Related	AHI1	•		o	o		•	•	•	•
Joubert Syndrome, ARL13B-Related	ARL13B	•		o			•			•
Joubert Syndrome, B9D1-Related	B9D1	•		o			•			•
Joubert Syndrome, B9D2-Related	B9D2	•		o			•			•
Joubert Syndrome, C2CD3-Related / Orofaciodigital Syndrome 14	C2CD3	•		o			•			•
Joubert Syndrome, CC2D2A-Related / COACH Syndrome	CC2D2A	•		o	o		•	•	•	•
Joubert Syndrome, CEP104-Related	CEP104	•		o			•			•
Joubert Syndrome, CEP120-Related / Short-Rib Thoracic Dysplasia 13 with or without Polydactyly	CEP120	•		o			•			•
Joubert Syndrome, CEP41-Related	CEP41	•		o			•			•
Joubert Syndrome, CPLANE1-Related / Orofaciodigital Syndrome 6	CPLANE1	•		o			•			•
Joubert Syndrome, CSPP1-Related	CSPP1	•		o			•			•
Joubert Syndrome, INPP5E-Related	INPP5E	•		o			•			•
Junctional Epidermolysis Bullosa, COL17A1-Related	COL17A1	•						•	•	•
Junctional Epidermolysis Bullosa, ITGA6-Related	ITGA6	•						•	•	•
Junctional Epidermolysis Bullosa, ITGB4-Related	ITGB4	•						•	•	•
Junctional Epidermolysis Bullosa, LAMA3-Related	LAMA3	•					•	•	•	•
Junctional Epidermolysis Bullosa, LAMB3-Related	LAMB3	•					•	•	•	•
Junctional Epidermolysis Bullosa, LAMC2-Related	LAMC2	•					•	•	•	•
Juvenile Retinoschisis, X-Linked	RS1		•		o		•	•	•	•
Ketothiolase Deficiency	ACAT1	•			o		•	•	•	•
Krabbe Disease	GALC	•					•	•	•	•
L1 Syndrome	L1CAM		•		o		•	•	•	•
Lamellar Ichthyosis, Type 1	TGM1	•					•	•	•	•
Laron Syndrome	GHR	•						•	•	•
Leber Congenital Amaurosis 2	RPE65	•					•	•	•	•
Leber Congenital Amaurosis Type AIPL1	AIPL1	•						•	•	•
Leber Congenital Amaurosis Type LRAT	LRAT	•						•	•	•
Leber Congenital Amaurosis Type TULP1	TULP1	•						•	•	•
Leber Congenital Amaurosis, IQCB1-Related / Senior-Loken Syndrome 5	IQCB1	•					•			•
Leber Congenital Amaurosis, Type CEP290	CEP290	•			o		•	•	•	•
Leber Congenital Amaurosis, Type LCA5	LCA5	•					•	•	•	•
Leber Congenital Amaurosis, Type RDH12	RDH12	•					•	•	•	•
Leber Congenital Amaurosis Type GUCY2D	GUCY2D	•						•	•	•
Leigh Syndrome, French-Canadian Type	LRPPRC	•					•	•	•	•
Lesch-Nyhan Syndrome	HPRT1		•				•	•	•	•
Lethal Congenital Contracture Syndrome 1	GLE1	•					•	•	•	•
Leukoencephalopathy with Vanishing White Matter	EIF2B5	•					•	•	•	•
Leukoencephalopathy With Vanishing White Matter, EIF2B1-Related	EIF2B1	•						•	•	•
Leukoencephalopathy With Vanishing White Matter, EIF2B2-Related	EIF2B2	•						•	•	•
Leukoencephalopathy With Vanishing White Matter, EIF2B3-Related	EIF2B3	•						•	•	•
Leukoencephalopathy With Vanishing White Matter, EIF2B4-Related	EIF2B4	•						•	•	•
LIG4 Syndrome	LIG4	•						•	•	•
Limb-Girdle Muscular Dystrophy Type 8	TRIM32	•						•	•	•
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3	•					•	•	•	•
Limb-Girdle Muscular Dystrophy, Type 2B	DYSF	•					•	•	•	•
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	•					•	•	•	•
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	•					•	•	•	•
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	•					•	•	•	•
Limb-Girdle Muscular Dystrophy, Type 2F	SGCD	•					•	•	•	•
Limb-Girdle Muscular Dystrophy, Type 2I	FKRP	•				o	•	•	•	•
Lipoamide Dehydrogenase Deficiency (Dihydropolipoamide Dehydrogenase Deficiency)	DLD	•			o		•	•	•	•
Lipoid Adrenal Hyperplasia	STAR	•					•	•	•	•
Lipoprotein Lipase Deficiency	LPL	•					•	•	•	•
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	•					•	•	•	•
Lung Disease, Immunodeficiency, And Chromosome Breakage Syndrome (LICS)	NSMCE3	•						•	•	•
Lysinuric Protein Intolerance	SLC7A7	•					•	•	•	•

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY			
				ACOG†	ACMG	H445	H568‡	H574‡	H613
Malonyl-CoA Decarboxylase Deficiency	MLYCD	•				•	•	•	•
Maple Syrup Urine Disease, Type 1A	BCKDHA	•		o		•	•	•	•
Maple Syrup Urine Disease, Type 1B	BCKDHB	•		o	o	•	•	•	•
Maple Syrup Urine Disease, Type 2	DBT	•		o		•	•	•	•
McKusick-Kaufman Syndrome	MKKS	•				•	•	•	•
Meckel Syndrome 7 / Nephronophthisis 3	NPHP3	•				•	•	•	•
Meckel-Gruber Syndrome, Type 1	MKS1	•				•	•	•	•
MECP2-Related Conditions	MECP2		•				•	•	•
MECR-Related Neurologic Disorder	MECR	•					•	•	•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	•		o	o	•	•	•	•
MEDNIK Syndrome	AP1S1	•				•	•	•	•
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	•			o	•	•	•	•
Menkes Syndrome	ATP7A		•			•	•	•	•
Merosin-Deficient Muscular Dystrophy	LAMA2	•				•	•	•	•
Metabolic Encephalopathy and Arrhythmias, TANGO2-Related	TANGO2	•				•	•	•	•
Metachromatic Leukodystrophy, ARSA-Related	ARSA	•			o	•	•	•	•
Metachromatic Leukodystrophy, PSAP-Related	PSAP	•				•	•	•	•
Methylmalonic Acidemia And Homocystinuria Type Cblf	LMBRD1	•					•	•	•
Methylmalonic Acidemia And Homocystinuria Type Cblx	HCFC1		•				•	•	•
Methylmalonic Aciduria and Homocystinuria, Type cblC	MMACHC	•			o	•	•	•	•
Methylmalonic Aciduria and Homocystinuria, Type cblD	MMADHC	•				•	•	•	•
Methylmalonic Aciduria, MCEE-Related	MCEE	•					•	•	•
Methylmalonic Aciduria, MMAA-Related	MMAA	•				•	•	•	•
Methylmalonic Aciduria, MMAB-Related	MMAB	•				•	•	•	•
Methylmalonic Aciduria, Type mut(0)	MUT	•			o	•	•	•	•
Mevalonic Kinase Deficiency	MVK				o	•	•	•	•
Microcephalic Osteodysplastic Primordial Dwarfism Type II	PCNT	•					•	•	•
Microphthalmia/Anophthalmia, VSX2-Related	VSX2	•				•	•	•	•
Mitochondrial Complex 1 Deficiency, ACAD9-Related	ACAD9	•				•	•	•	•
Mitochondrial Complex 1 Deficiency, NDUFAF5-Related	NDUFAF5	•				•	•	•	•
Mitochondrial Complex 1 Deficiency, NDUFS6-Related	NDUFS6	•				•	•	•	•
Mitochondrial complex I Deficiency, Nuclear Type 1	NDUFS4	•				•	•	•	•
Mitochondrial Complex I Deficiency, Nuclear Type 10	NDUFAF2	•					•	•	•
Mitochondrial Complex I Deficiency, Nuclear Type 17	NDUFAF6	•				•			•
Mitochondrial Complex I Deficiency, Nuclear Type 19	FOXRED1	•					•	•	•
Mitochondrial Complex I Deficiency, Nuclear Type 3	NDUFS7	•					•	•	•
Mitochondrial Complex I Deficiency, Nuclear Type 4	NDUFV1	•					•	•	•
Mitochondrial Complex IV Deficiency, Nuclear Type 2, SCO2-Related	SCO2				o	•	•	•	•
Mitochondrial Complex IV Deficiency, Nuclear Type 6	COX15	•					•	•	•
Mitochondrial Dna Depletion Syndrome 2	TK2	•					•	•	•
Mitochondrial Dna Depletion Syndrome 3	DGUOK	•					•	•	•
Mitochondrial Myopathy and Sideroblastic Anemia (MLSA1)	PUS1	•				•	•	•	•
Mitochondrial Trifunctional Protein Deficiency, HADHB-Related	HADHB	•				•	•	•	•
Molybdenum Cofactor Deficiency Type B	MOCS2	•					•	•	•
Molybdenum Cofactor Deficiency, Type A	MOCS1	•				•	•	•	•
Mucopolipidosis II/IIIA	GNPTAB	•			o	•	•	•	•
Mucopolipidosis III gamma	GNPTG	•				•	•	•	•
Mucopolipidosis, Type IV	MCOLN1	•		o	o	•	•	•	•
Mucopolysaccharidosis, Type I (Hurler Syndrome)	IDUA	•			o	•	•	•	•
Mucopolysaccharidosis, Type II (Hunter Syndrome)	IDS		•			•	•	•	•
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	SGSH	•				•	•	•	•
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	NAGLU	•				•	•	•	•
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	HGSNAT	•				•	•	•	•
Mucopolysaccharidosis, Type IIID (Sanfilippo D)	GNS	•				•	•	•	•
Mucopolysaccharidosis, Type IVA (Morquio Syndrome)	GALNS	•				•	•	•	•
Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	GLB1	•				•	•	•	•
Mucopolysaccharidosis, Type IX	HYAL1	•				•	•	•	•
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	ARSB	•				•	•	•	•
Mucopolysaccharidosis, Type VII	GUSB	•				•	•	•	•
Mulibrey Nanism	TRIM37	•				•	•	•	•
Multiple Pterygium Syndrome, CHRNG-Related / Escobar Syndrome	CHRNG	•				•	•	•	•
Multiple Sulfatase Deficiency	SUMF1	•				•	•	•	•
Muscle-Eye-Brain Disease, POMGNT1-Related	POMGNT1	•				•	•	•	•
Muscular Dystrophy-Dystroglycanopathy	RXYLT1	•					•	•	•
MUSK-Related Congenital Myasthenic Syndrome	MUSK	•					•	•	•
Myoneurogastrointestinal Encephalopathy (MNGIE)	TYMP	•				•	•	•	•
Myotonia Congenita	CLCN1				o	•	•	•	•
Myotubular Myopathy, X-Linked	MTM1		•			•	•	•	•
N-acetylglutamate Synthase Deficiency	NAGS	•				•	•	•	•
Nemaline Myopathy, NEB-Related	NEB	•			o	•	•	•	•

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY				
				ACOG†	ACMG	H445	H568‡	H574‡	H613	
Nephrogenic Diabetes Insipidus, AVPR2-Related	AVPR2		•				•	•	•	•
Nephronophthisis 1	NPHP1	•					•	•	•	•
Neuronal Ceroid Lipofuscinosis, CLN5-Related	CLN5	•					•	•	•	•
Neuronal Ceroid Lipofuscinosis, CLN6-Related	CLN6	•					•	•	•	•
Neuronal Ceroid Lipofuscinosis, CLN8-Related	CLN8	•					•	•	•	•
Neuronal Ceroid Lipofuscinosis, MFSD8-Related	MFSD8	•					•	•	•	•
Neuronal Ceroid Lipofuscinosis, PPT1-Related	PPT1	•					•	•	•	•
Neuronal Ceroid Lipofuscinosis, TPP1-Related	TPP1	•					•	•	•	•
Ngly1-Congenital Disorder Of Glycosylation	NGLY1	•						•	•	•
Niemann-Pick Disease, Type C1/D	NPC1	•		o			•	•	•	•
Niemann-Pick Disease, Type C2	NPC2	•		o			•	•	•	•
Niemann-Pick Disease, Types A/B	SMPD1	•		o	o		•	•	•	•
Nijmegen Breakage Syndrome	NBN	•					•	•	•	•
Non-Syndromic Hearing Loss, GJB2-Related	GJB2	•			o		•	•	•	•
Nonsyndromic Hearing Loss, MYO15A-Related	MYO15A	•					•	•	•	•
Nonsyndromic Hearing Loss, OTOA-Related	OTOA	•						•	•	•
Nonsyndromic Hearing Loss, OTOF-Related	OTOF	•						•	•	•
Nonsyndromic Hearing Loss, PJVK-Related	PJVK	•						•	•	•
Nonsyndromic Hearing Loss, SYNE4-Related	SYNE4	•						•	•	•
Nonsyndromic Hearing Loss, TMC1-Related	TMC1	•						•	•	•
Nonsyndromic Hearing Loss, TMPRSS3-Related	TMPRSS3	•						•	•	•
Nonsyndromic Intellectual Disability	CC2D1A	•						•	•	•
Normophosphatemic Tumoral Calcinosis	SAMD9	•						•	•	•
Oculocutaneous Albinism Type III	TYRP1	•						•	•	•
Oculocutaneous Albinism Type IV	SLC45A2	•						•	•	•
Oculocutaneous Albinism, OCA2-Related	OCA2				o		•	•	•	•
Oculocutaneous Albinism, Type 1A and 1B	TYR				o		•	•	•	•
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	WNT10A	•					•	•	•	•
Omenn Syndrome, RAG2-Related	RAG2	•					•	•	•	•
Opitz G/BBB Syndrome, X-Linked	MID1		•		o		•	•	•	•
Ornithine Aminotransferase Deficiency	OAT	•					•	•	•	•
Ornithine Transcarbamylase Deficiency	OTC		•		o		•	•	•	•
Osteogenesis Imperfecta Type VII	CRTAP	•						•	•	•
Osteogenesis Imperfecta Type VIII	P3H1	•						•	•	•
Osteogenesis Imperfecta Type XI	FKBP10	•						•	•	•
Osteogenesis Imperfecta Type XIII	BMP1	•						•	•	•
Osteopetrosis, Infantile Malignant, TCIRG1-Related	TCIRG1	•					•	•	•	•
Osteopetrosis, OSTM1-Related	OSTM1	•						•	•	•
Pantothenate Kinase-Associated Neurodegeneration	PANK2	•						•	•	•
Papillon Lefèvre Syndrome	CTSC	•						•	•	•
Parkinson Disease 15	FBXO7	•						•	•	•
Pendred Syndrome	SLC26A4	•			o		•	•	•	•
Perlman Syndrome	DIS3L2	•					•			•
Pgm3-Congenital Disorder Of Glycosylation	PGM3	•						•	•	•
Phenylketonuria	PAH	•			o		•	•	•	•
Pign-Congenital Disorder Of Glycosylation	PIGN	•						•	•	•
Pituitary Hormone Deficiency, Combined 3	LHX3	•					•	•	•	•
PLP1 Disorders	PLP1		•		o		•	•	•	•
POLG-Related Disorders	POLG	•			o		•	•	•	•
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	•			o		•	•	•	•
Pontocerebellar Hypoplasia, EXOSC3-Related	EXOSC3	•					•	•	•	•
Pontocerebellar Hypoplasia, RARS2-Related	RARS2	•			o		•	•	•	•
Pontocerebellar Hypoplasia, TSEN2-Related	TSEN2	•					•			•
Pontocerebellar Hypoplasia, TSEN54-Related	TSEN54	•					•	•	•	•
Pontocerebellar Hypoplasia, Type 1A	VRK1	•					•	•	•	•
Pontocerebellar Hypoplasia, Type 2D	SEPSECS	•					•	•	•	•
Pontocerebellar Hypoplasia, VPS53-Related	VPS53	•					•	•	•	•
Primary Ciliary Dyskinesia, CCDC103-Related	CCDC103	•						•	•	•
Primary Ciliary Dyskinesia, CCDC39-Related	CCDC39	•						•	•	•
Primary Ciliary Dyskinesia, DNAH11-Related	DNAH11	•						•	•	•
Primary Ciliary Dyskinesia, DNAH5-Related	DNAH5	•					•	•	•	•
Primary Ciliary Dyskinesia, DNAI1-Related	DNAI1	•					•	•	•	•
Primary Ciliary Dyskinesia, DNAI2-Related	DNAI2	•					•	•	•	•
Primary Congenital Glaucoma / Peters Anomaly	CYP1B1	•					•	•	•	•
Primary Hyperoxaluria, Type 1	AGXT	•					•	•	•	•
Primary Hyperoxaluria, Type 2	GRHPR	•				o	•	•	•	•
Primary Hyperoxaluria, Type 3	HOGA1	•					•	•	•	•
Primary Microcephaly 1, Autosomal Recessive	MCPH1				o		•	•	•	•
Progressive Early-Onset Encephalopathy With Brain Atrophy And Thin Corpus Callosum	TBCD	•						•	•	•
Progressive Familial Intrahepatic Cholestasis, ABCB4-Related	ABCB4	•						•	•	•

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY			
				ACOG†	ACMG	H445	H568‡	H574‡	H613
Progressive Familial Intrahepatic Cholestasis, Type 1 (PFIC1)	ATP8B1	•				•	•	•	•
Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	•				•	•	•	•
Progressive Familial Intrahepatic Cholestasis, Type 4 (PFIC4)	TJP2	•				•			•
Progressive Pseudorheumatoid Dysplasia	CCN6	•					•	•	•
Prolidase Deficiency	PEPD	•				•	•	•	•
Propionic Acidemia, PCCA-Related	PCCA	•				•	•	•	•
Propionic Acidemia, PCCB-Related	PCCB	•				•	•	•	•
Prothrombin-Related Thrombophilia	F2*	•					•	•	•
Pseudochoolinesterase Deficiency	BCHE	•				•			•
Pseudoxanthoma Elasticum	ABCC6	•				•			•
Pterin-4 Alpha-Carbinolamine Dehydratase (PCD) Deficiency	PCBD1	•					•	•	•
Pycnodysostosis	CTSK	•				•	•	•	•
Pyridoxal 5'-Phosphate-Dependent Epilepsy	PNPO	•					•	•	•
Pyridoxine-Dependent Epilepsy	ALDH7A1	•				•	•	•	•
Pyruvate Carboxylase Deficiency	PC	•				•	•	•	•
Pyruvate Dehydrogenase Deficiency, PDHB-Related	PDHB	•				•	•	•	•
Pyruvate Dehydrogenase Deficiency, X-Linked	PDHA1		•			•	•	•	•
Refsum Disease, PHYH-Related	PHYH	•				•	•	•	•
Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	ATP6V1B1	•				•	•	•	•
Renal Tubular Acidosis, Proximal, with Ocular Abnormalities and Mental Retardation	SLC4A4	•				•			•
Retinitis Pigmentosa 2	RP2		•				•	•	•
Retinitis Pigmentosa 25	EYS	•				•	•	•	•
Retinitis Pigmentosa 26	CERKL	•				•	•	•	•
Retinitis Pigmentosa 28	FAM161A	•				•	•	•	•
Retinitis Pigmentosa 36	PRCD	•					•	•	•
Retinitis Pigmentosa 59	DHDDS	•			o	•	•	•	•
Retinitis Pigmentosa 62	MAK	•					•	•	•
Retinitis Pigmentosa, X-Linked, RPGR-Related	RPGR		•		o	•	•	•	•
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	•				•	•	•	•
Rhizomelic Chondrodysplasia Punctata, Type 2	GNPAT	•				•	•	•	•
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	•				•	•	•	•
RLBP1-Related Retinopathy	RLBP1	•					•	•	•
Roberts Syndrome	ESCO2	•				•	•	•	•
RYR1-Related Conditions	RYR1	•					•	•	•
Salla Disease	SLC17A5	•				•	•	•	•
Sandhoff Disease	HEXB	•				•	•	•	•
Schimke Immunososseous Dysplasia	SMARCAL1	•				•	•	•	•
Schindler Disease	NAGA				o	•	•	•	•
Segawa Syndrome, TH-Related	TH	•				•	•	•	•
Senior-Loken Syndrome 4 / Nephronophthisis 4	NPHP4	•				•			•
Sepiapterin Reductase Deficiency	SPR	•					•	•	•
Severe Combined Immunodeficiency (SCID), CD3D-Related	CD3D	•					•	•	•
Severe Combined Immunodeficiency (SCID), CD3E-Related	CD3E	•					•	•	•
Severe Combined Immunodeficiency (SCID), FOXP1-Related	FOXP1	•					•	•	•
Severe Combined Immunodeficiency (SCID), IKBKB-Related	IKBKB	•					•	•	•
Severe Combined Immunodeficiency (SCID), IL7R-Related	IL7R	•					•	•	•
Severe Combined Immunodeficiency (SCID), JAK3-Related	JAK3	•					•	•	•
Severe Combined Immunodeficiency (SCID), PTPRC-Related	PTPRC	•					•	•	•
Severe Combined Immunodeficiency, ADA-Related	ADA	•				•	•	•	•
Severe Combined Immunodeficiency, RAG1-Related	RAG1	•				•	•	•	•
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	•				•	•	•	•
Severe Combined Immunodeficiency, X-Linked	IL2RG		•			•	•	•	•
Short-Rib Thoracic Dysplasia 3 with or without Polydactyly	DYNC2H1				o	•	•	•	•
Shwachman-Diamond Syndrome, SBDS-Related	SBDS	•				•			•
Sialidosis	NEU1	•				•	•	•	•
Sjögren-Larsson Syndrome	ALDH3A2	•				•	•	•	•
Smith-Lemli-Opitz Syndrome	DHCR7	•			o	•	•	•	•
Spastic Paraplegia, Type 15	ZFYVE26	•				•	•	•	•
Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly (SPATCCM)	SLC11A4	•				•	•	•	•
SPG11-Related Conditions	SPG11	•				•	•	•	•
Spinal Muscular Atrophy	SMN1	•			o	•	•	•	•
Spinal Muscular Atrophy With Respiratory Distress Type 1	IGHMBP2	•					•	•	•
Spinocerebellar Ataxia, Autosomal Recessive 10	ANO10				o	•	•	•	•
Spinocerebellar Ataxia, Autosomal Recessive 12	WVVOX	•				•			•
Spondylocostal Dysostosis 1	DLL3	•					•	•	•
Spondylothoracic Dysostosis, MESP2-Related	MESP2	•				•	•	•	•
Steel Syndrome	COL27A1	•				•	•	•	•
Steroid-Resistant Nephrotic Syndrome	NPHS2	•				•	•	•	•
Stuve-Wiedemann Syndrome	LIFR	•				•	•	•	•
SURF1-Related Conditions	SURF1	•					•	•	•

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY			
				ACOG†	ACMG	H445	H568‡	H574‡	H613
Surfactant Dysfunction, ABCA3-Related	ABCA3				o	•	•	•	•
Tay-Sachs Disease	HEXA	•			o	•	•	•	•
TBCE-Related Conditions	TBCE	•					•	•	•
Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2	•					•	•	•
Thyroid Dyshormonogenesis 1	SLC5A5	•					•	•	•
Thyroid Dyshormonogenesis 2A	TPO	•					•	•	•
Thyroid Dyshormonogenesis 3	TG	•					•	•	•
Thyroid Dyshormonogenesis 6	DUOX2	•					•	•	•
Transcobalamin II Deficiency	TCN2	•					•	•	•
Trichohepatoenteric Syndrome, SKIV2L-Related	SKIC2	•					•	•	•
Trichohepatoenteric Syndrome, TTC37-Related	TTC37	•				•	•	•	•
Trichothiodystrophy 1 / Xeroderma Pigmentosum, Group D	ERCC2	•			o	•	•	•	•
Trimethylaminuria	FMO3				o	•	•	•	•
Triple A Syndrome	AAAS	•				•	•	•	•
TSHR-Related Conditions	TSHR	•					•	•	•
Tyrosinemia Type III	HPD	•					•	•	•
Tyrosinemia, Type 1	FAH	•			o	•	•	•	•
Tyrosinemia, Type 2	TAT	•				•	•	•	•
Usher Syndrome, Type 1B	MYO7A	•				•	•	•	•
Usher Syndrome, Type 1C	USH1C	•				•	•	•	•
Usher Syndrome, Type 1D	CDH23	•				•	•	•	•
Usher Syndrome, Type 1F	PCDH15	•			o	•	•	•	•
Usher Syndrome, Type 1J / Deafness, Autosomal Recessive, 48	CIB2	•				•			•
Usher Syndrome, Type 2A	USH2A	•			o	•	•	•	•
Usher Syndrome, Type 2C	ADGRV1	•				•	•	•	•
Usher Syndrome, Type 3	CLRN1	•			o	•	•	•	•
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	•			o	•	•	•	•
Vitamin D-Dependent Rickets Type 2A	VDR	•					•	•	•
Vici Syndrome	EPG5	•					•	•	•
Vitamin D Dependent Rickets, Type 1A	CYP27B1	•			o	•	•	•	•
Vldlr-Associated Cerebellar Hypoplasia	VLDLR	•					•	•	•
Walker-Warburg Syndrome, FKTN-Related	FKTN	•			o	•	•	•	•
Walker-Warburg Syndrome, ISPD-Related	ISPD	•				•			•
Walker-Warburg Syndrome, LARGE1-Related	LARGE1	•				•	•	•	•
Walker-Warburg Syndrome, POMT1-Related	POMT1	•				•	•	•	•
Walker-Warburg Syndrome, POMT2-Related	POMT2	•				•	•	•	•
Warsaw Breakage Syndrome	DDX11	•						•	•
Werner Syndrome	WRN	•				•	•	•	•
Wilson Disease	ATP7B	•			o	•	•	•	•
Wiskott-Aldrich Syndrome	WAS		•			•	•	•	•
Wolcott-Rallison Syndrome	EIF2AK3	•				•	•	•	•
Wolman Disease	LIPA	•				•	•	•	•
Woodhouse-Sakati Syndrome	DCAF17	•					•	•	•
Xeroderma Pigmentosum Variant Type	POLH	•					•	•	•
Xeroderma Pigmentosum, Group A	XPA	•				•	•	•	•
Xeroderma Pigmentosum, Group C	XPC	•			o	•	•	•	•
X-Linked Chondrodysplasia Punctata 1	ARSL		•			•	•	•	•
X-Linked Lissencephaly with Abnormal Genitalia	ARX		•		o	•	•	•	•
Zellweger Spectrum Disorder, PEX13-Related	PEX13	•					•	•	•
Zellweger Spectrum Disorder, PEX16-Related	PEX16	•					•	•	•
Zellweger Spectrum Disorder, PEX5-Related	PEX5	•					•	•	•
Zellweger Spectrum Disorders, PEX10-Related	PEX10	•				•	•	•	•
Zellweger Spectrum Disorders, PEX12-Related	PEX12	•				•	•	•	•
Zellweger Spectrum Disorders, PEX1-Related	PEX1	•				•	•	•	•
Zellweger Spectrum Disorders, PEX26-Related	PEX26	•				•	•	•	•
Zellweger Spectrum Disorders, PEX2-Related	PEX2	•				•	•	•	•
Zellweger Spectrum Disorders, PEX6-Related	PEX6	•				•	•	•	•

*Low penetrance genes with variable presentation. Horizon 556 is available to clinicians who would like to exclude screening for these 13 genes.

†Note that ACOG screening recommendations listed here include diseases in ACOG Committee Opinion 690 example expanded carrier screening panel, as well as the diseases listed in ACOG Committee Opinion 691

‡Panel is named based on the total number of genes, counting HBA1 and HBA2 separately.

1. American College of Obstetricians and Gynecologists, Committee Opinion # 690, March 2017.

2. American College of Obstetricians and Gynecologists, Committee Opinion # 691, March 2017.

3. Gregg et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG), July 2021.