Horizon Conditions List

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CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY					
				ACOG*	ACMG	H4	H14	H27	H106	H274	H421
17-Beta Hydroxysteroid Dehydrogenase 3 Deficiency	HSD17B3	•									•
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	HSD3B2	•								•	•
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	•								•	•
3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADH	•									•
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	MCCC1	•								•	•
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	MCCC2	•								•	•
3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH	•							•	•	•
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	PTS	•								•	•
Abetalipoproteinemia	MTTP	•							•	•	•
Achondrogenesis, Type 1B	SLC26A2	•								•	•
Achromatopsia, CNGB3-Related	CNGB3	•								•	•
Acrodermatitis Enteropathica	SLC39A4	•								•	•
Action Myoclonus–Renal Failure (AMRF) Syndrome	SCARB2	•									•
Acute Infantile Liver Failure, TRMU-Related	TRMU	•							•	•	•
Acyl-CoA Oxidase I Deficiency	ACOX1	•								•	•
Adrenal Hypoplasia Congenita, X-Linked	NR0B1		•								•
Adrenoleukodystrophy, X-Linked	ABCD1		•						•	•	•
Agammaglobulinemia, X-Linked	BTK		•								•
Aicardi-Goutières Syndrome	SAMHD1	•								•	•
Aicardi-Goutières Syndrome, RNASEH2A-Related	RNASEH2A	•									•
Aicardi-Goutières Syndrome, RNASEH2B-Related	RNASEH2B	•									•
Aicardi-Goutières Syndrome, RNASEH2C-Related	RNASEH2C	•									•
Alpha-1 Antitrypsin Deficiency	SERPINA1	•									•
Alpha-Mannosidosis	MAN2B1	•								•	•
Alpha-Thalassemia	HBA1/HBA2	•		○			•	•	•	•	•
Alpha-Thalassemia Intellectual Disability Syndrome	ATRX		•							•	•
Alport Syndrome, COL4A3-Related	COL4A3	•							•	•	•
Alport Syndrome, COL4A4-Related	COL4A4	•								•	•
Alport Syndrome, X-Linked	COL4A5		•							•	•
Alstrom Syndrome	ALMS1	•								•	•
Amish Infantile Epilepsy Syndrome	ST3GAL5	•									•
Andermann Syndrome	SLC12A6	•								•	•
Argininemia	ARG1	•									•
Argininosuccinate Lyase Deficiency	ASL	•								•	•
Aromatase Deficiency	CYP19A1	•								•	•
Arts Syndrome	PRPS1		•								•
Asparagine Synthetase Deficiency	ASNS	•							•	•	•
Aspartylglycosaminuria	AGA	•								•	•
Ataxia with Vitamin E Deficiency	TTPA	•								•	•
Ataxia-Telangiectasia	ATM	•							•	•	•
Ataxia-Telangiectasia-Like Disorder 1	MRE11	•									•
Autism Spectrum, Epilepsy and Arthrogryposis	SLC35A3	•							•	•	•
Autoimmune Polyglandular Syndrome, Type 1	AIRE	•							•	•	•
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	•								•	•
Bardet-Biedl Syndrome, BBS1-Related	BBS1	•								•	•
Bardet-Biedl Syndrome, BBS2-Related	BBS2	•							•	•	•
Bardet-Biedl Syndrome, BBS4-Related	BBS4	•									•
Bardet-Biedl Syndrome, BBS7-Related	BBS7	•									•
Bardet-Biedl Syndrome, BBS9-Related	BBS9	•									•
Bardet-Biedl Syndrome, BBS10-Related	BBS10	•								•	•
Bardet-Biedl Syndrome, BBS12-Related	BBS12	•								•	•
Bardet-Biedl Syndrome, TTC8-Related	TTC8	•									•
Bare Lymphocyte Syndrome, CIITA-Related	CIITA	•								•	•
Barth Syndrome	TAZ		•								•
Bartter Syndrome, BSND-Related	BSND	•								•	•
Batten Disease, CLN3-Related	CLN3	•						•	•	•	•
Bernard-Soulier Syndrome, Type A1/A2	GP1BA	•									•
Bernard-Soulier Syndrome, Type C	GP9	•									•
Beta-Hemoglobinopathies	HBB	•		○			•	•	•	•	•
Beta-Ureidopropionase Deficiency	UPB1	•									•
Bilateral Frontoparietal Polymicrogyria	GPR56	•								•	•
Biotinidase Deficiency	BTD	•								•	•
Bloom Syndrome	BLM	•		○	○			•	•	•	•
Canavan Disease	ASPA	•		○	○		•	•	•	•	•
Carbamoyl Phosphate Synthetase I Deficiency	CPS1	•								•	•
Carnitine Deficiency	SLC22A5	•								•	•
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	•								•	•
Carnitine Palmitoyltransferase II Deficiency	CPT2	•							•	•	•
Carnitine-Acylcarnitine Translocase Deficiency	SLC25A20	•									•
Carpenter Syndrome	RAB23	•								•	•
Cartilage-Hair Hypoplasia	RMRP	•								•	•
Cerebrotendinous Xanthomatosis	CYP27A1	•							•	•	•
Charcot-Marie-Tooth Disease with Deafness, X-Linked	GJB1		•							•	•
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	•								•	•
Chediak-Higashi Syndrome	LYST	•									•
Choreoacanthocytosis	VPS13A	•							•	•	•
Choroideremia	CHM		•							•	•
Chronic Granulomatous Disease, CYBA-Related	CYBA	•							•	•	•
Chronic Granulomatous Disease, X-Linked	CYBB		•							•	•

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				ACOG*	ACMG	H4	H14	H27	H106	H274	H421
Ciliopathies, RPGRIP1L-Related	RPGRIP1L	•								•	•
Citrin Deficiency	SLC25A13	•								•	•
Citrullinemia, Type 1	ASS1	•						•	•	•	•
CLN10 Disease	CTSD	•									•
Cohen Syndrome	VPS13B	•								•	•
Combined Malonic and Methylmalonic Aciduria	ACSF3	•								•	•
Combined Oxidative Phosphorylation Deficiency 1	GFM1	•								•	•
Combined Oxidative Phosphorylation Deficiency 3	TSFM	•								•	•
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	•									•
Congenital Amegakaryocytic Thrombocytopenia	MPL	•							•	•	•
Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	PMM2	•							•	•	•
Congenital Disorder of Glycosylation, Type 1B	MPI	•								•	•
Congenital Disorder of Glycosylation, Type 1C	ALG6	•								•	•
Congenital Finnish Nephrosis	NPHS1	•								•	•
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	•								•	•
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, HAX1-Related	HAX1	•								•	•
Congenital Neutropenia, VPS45-Related	VPS45	•								•	•
Corneal Dystrophy and Perceptive Deafness	SLC4A11	•								•	•
Corticosterone Methyloxidase 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		•							•	•
Cystic Fibrosis	CFTR	•		○	○	•	•	•	•	•	•
Cystinosis	CTNS	•							•	•	•
Cytochrome C Oxidase Deficiency, PET100-Related	PET100	•									•
D-Bifunctional Protein Deficiency	HSD17B4	•								•	•
Deafness, Autosomal Recessive 77	LOXHD1	•							•	•	•
Dent Disease, Type 1	CLCN5		•								•
Dent Disease, Type 2 / Lowe Syndrome	OCRL		•								•
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	•									•
Duchenne/Becker Muscular Dystrophy	DMD		•			•	•	•	•	•	•
Dyskeratosis Congenita, DKC1-Related	DKC1		•								•
Dyskeratosis Congenita, RTEL1-Related	RTEL1	•							•	•	•
Dystrophic Epidermolysis Bullosa, COL7A1-Related	COL7A1	•								•	•
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2	•							•	•	•
Ellis-van Creveld Syndrome, EVC-Related	EVC	•								•	•
Ellis-van Creveld Syndrome, EVC2-Related	EVC2	•									•
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	EMD		•							•	•
Enhanced S-Cone Syndrome	NR2E3	•							•	•	•
Epiphyseal Dysplasia, Multiple, 7 / Desbuquois Dysplasia 1	CANT1	•									•
ERCC6-Related Disorders	ERCC6	•									•
ERCC8-Related Disorders	ERCC8	•									•
Ethylmalonic Encephalopathy	ETHE1	•								•	•
Fabry Disease	GLA		•							•	•
Factor IX Deficiency	F9		•							•	•
Factor XI Deficiency	F11	•							•	•	•
Familial Dysautonomia	IKBKAP	•		○	○		•	•	•	•	•
Familial Hemophagocytic Lymphohistiocytosis, PRF1-Related	PRF1	•									•
Familial Hemophagocytic Lymphohistiocytosis, STX11-Related	STX11	•									•
Familial Hemophagocytic Lymphohistiocytosis, STXBP2-Related	STXBP2	•									•
Familial Hypercholesterolemia, LDLRAP1-Related	LDLRAP1	•								•	•
Familial Hypercholesterolemia, LDLR-Related	LDLR	•							•	•	•
Familial Hyperinsulinism, ABCC8-Related	ABCC8	•		○					•	•	•
Familial Mediterranean Fever	MEFV	•							•	•	•
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	AQP2	•								•	•
Fanconi Anemia, Group A	FANCA	•		○					•	•	•
Fanconi Anemia, Group B	FANCB		•								•
Fanconi Anemia, Group C	FANCC	•		○	○			•	•	•	•
Fanconi Anemia, Group D2	FANCD2	•									•
Fanconi Anemia, Group E	FANCE	•									•
Fanconi Anemia, Group F	FANCF	•									•
Fanconi Anemia, Group G	FANCG	•		○						•	•
Fanconi Anemia, Group I	FANCI	•									•
Fanconi Anemia, Group L	FANCL	•									•
Farber Lipogranulomatosis	ASAH1	•									•
Fragile X Syndrome	FMR1		•	○		•	•	•	•	•	•
Fumarase Deficiency	FH	•								•	•
GABA-Transaminase Deficiency	ABAT	•									•

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Galactokinase Deficiency (Galactosemia, Type II)	GALK1	•								•	•
Galactosemia	GALT	•					•	•	•	•	•
Galactosialidosis	CTSA	•									•
Gaucher Disease	GBA	•			○		•	•	•	•	•
Gitelman Syndrome	SLC12A3	•								•	•
Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD		•								•
Glutaric Acidemia, Type 1	GCDH	•								•	•
Glutaric Acidemia, Type 2A	ETFA	•								•	•
Glutaric Acidemia, Type 2B	ETFB	•									•
Glutaric Acidemia, Type 2C	ETFDH	•								•	•
Glycine Encephalopathy, AMT-Related	AMT	•								•	•
Glycine Encephalopathy, GLDC-Related	GLDC	•								•	•
Glycogen Storage Disease, Type 1A	G6PC	•		○				•	•	•	•
Glycogen Storage Disease, Type 1B	SLC37A4	•		○						•	•
Glycogen Storage Disease, Type 2 (Pompe Disease)	GAA	•							•	•	•
Glycogen Storage Disease, Type 3	AGL	•							•	•	•
Glycogen Storage Disease, Type 4	GBE1	•							•	•	•
Glycogen Storage Disease, Type 5 (McArdle Disease)	PYGM	•							•	•	•
Glycogen Storage Disease, Type 7	PFKM	•							•	•	•
GRACILE Syndrome	BCS1L	•								•	•
Guanidinoacetate Methyltransferase Deficiency	GAMT	•								•	•
Harlequin Ichthyosis	ABCA12	•									•
Hemochromatosis, Type 2A	HFE2	•								•	•
Hemochromatosis, Type 3, TFR2-Related	TFR2	•								•	•
Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Related	MPV17	•								•	•
Hereditary Fructose Intolerance	ALDOB	•								•	•
Hereditary Spastic Paraparesis, Type 49	TECPR2	•							•	•	•
Hermansky-Pudlak Syndrome, AP3B1-Related	AP3B1	•									•
Hermansky-Pudlak Syndrome, HPS1-Related	HPS1	•								•	•
Hermansky-Pudlak Syndrome, HPS3-Related	HPS3	•							•	•	•
Hermansky-Pudlak Syndrome, HPS4-Related	HPS4	•									•
Heterotaxy Syndrome, ZIC3-Related	ZIC3		•								•
Holocarboxylase Synthetase Deficiency	HLCS	•								•	•
Homocystinuria due to Deficiency of MTHFR	MTHFR	•							•	•	•
Homocystinuria, CBS-Related	CBS	•								•	•
Homocystinuria, Type cblE	MTRR	•								•	•
Hydrolethalus Syndrome	HYLS1	•								•	•
Hyper IgM Syndrome, X-Linked	CD40LG		•								•
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH Syndrome)	SLC25A15	•								•	•
Hyperphosphatemic Familial Tumoral Calcinosis, GALNT3-Related	GALNT3	•									•
Hypohidrotic Ectodermal Dysplasia, X-Linked	EDA		•							•	•
Hypophosphatasia, ALPL-Related	ALPL	•								•	•
Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome	FOXP3		•								•
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 Lissencephaly Sequence / Subcortical Band Heterotopia	DCX		•								•
Isovaleric Acidemia	IVD	•						•	•	•	•
Johanson-Blizzard Syndrome	UBR1	•									•
Joubert Syndrome 2 / Meckel Syndrome 2	TMEM216	•		○					•	•	•
Joubert Syndrome, AHI1-Related	AHI1	•		○							•
Joubert Syndrome, ARL13B-Related	ARL13B	•		○							•
Joubert Syndrome, B9D1-Related	B9D1	•		○							•
Joubert Syndrome, B9D2-Related	B9D2	•		○							•
Joubert Syndrome, C2CD3-Related / Orofaciodigital Syndrome 14	C2CD3	•		○							•
Joubert Syndrome, CC2D2A-Related / COACH Syndrome	CC2D2A	•		○							•
Joubert Syndrome, CEP104-Related	CEP104	•		○							•
Joubert Syndrome, CEP120-Related / Short-Rib Thoracic Dysplasia13 with or without Polydactyly	CEP120	•		○							•
Joubert Syndrome, CEP41-Related	CEP41	•		○							•
Joubert Syndrome, CPLANE1-Related / Orofaciodigital Syndrome 6	CPLANE1	•		○							•
Joubert Syndrome, CSPP1-Related	CSPP1	•		○							•
Joubert Syndrome, INPP5E-Related	INPP5E	•		○							•
Junctional Epidermolysis Bullosa, LAMA3-Related	LAMA3	•									•
Junctional Epidermolysis Bullosa, LAMB3-Related	LAMB3	•									•
Junctional Epidermolysis Bullosa, LAMC2-Related	LAMC2	•									•
Juvenile Retinoschisis, X-Linked	RS1		•							•	•
Ketothiolase Deficiency	ACAT1	•								•	•
Krabbe Disease	GALC	•								•	•
L1 Syndrome	L1CAM		•								•
Lamellar Ichthyosis, Type 1	TGM1	•								•	•
Leber Congenital Amaurosis 2	RPE65	•							•	•	•
Leber Congenital Amaurosis, IQCB1-Related / Senior-Loken Syndrome 5	IQCB1	•									•
Leber Congenital Amaurosis, Type CEP290	CEP290	•								•	•
Leber Congenital Amaurosis, Type LCA5	LCA5	•								•	•
Leber Congenital Amaurosis, Type RDH12	RDH12	•								•	•
Leigh Syndrome, French-Canadian Type	LRPPRC	•								•	•
Lesch-Nyhan Syndrome	HPRT1		•								•

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Lethal Congenital Contracture Syndrome 1	GLE1	•								•	•
Leukoencephalopathy with Vanishing White Matter	EIF2B5	•								•	•
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	•								•	•
Lipoamide Dehydrogenase Deficiency (Dihydrolipoamide Dehydrogenase Deficiency)	DLD	•							•	•	•
Lipoid Adrenal Hyperplasia	STAR	•								•	•
Lipoprotein Lipase Deficiency	LPL	•								•	•
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	•								•	•
Lysinuric Protein Intolerance	SLC7A7	•								•	•
Malonyl-CoA Decarboxylase Deficiency	MLYCD	•									•
Maple Syrup Urine Disease, Type 1A	BCKDHA	•		○						•	•
Maple Syrup Urine Disease, Type 1B	BCKDHB	•		○					•	•	•
Maple Syrup Urine Disease, Type 2	DBT	•		○							•
McKusick-Kaufman Syndrome	MKKS	•									•
Meckel Syndrome 7 / Nephronophthisis 3	NPHP3	•									•
Meckel-Gruber Syndrome, Type 1	MKS1	•								•	•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	•		○			•	•	•	•	•
MEDNIK Syndrome	AP1S1	•									•
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	•							•	•	•
Menkes Syndrome	ATP7A		•							•	•
Merosin-Deficient Muscular Dystrophy	LAMA2	•									•
Metabolic Encephalopathy and Arrhythmias, TANGO2-Related	TANGO2	•									•
Metachromatic Leukodystrophy, ARSA-Related	ARSA	•							•	•	•
Metachromatic Leukodystrophy, PSAP-Related	PSAP	•								•	•
Methylmalonic Aciduria and Homocystinuria, Type cblC	MMACHC	•						•	•	•	•
Methylmalonic Aciduria and Homocystinuria, Type cblD	MMADHC	•								•	•
Methylmalonic Aciduria, MMAA-Related	MMAA	•								•	•
Methylmalonic Aciduria, MMAB-Related	MMAB	•								•	•
Methylmalonic Aciduria, Type mut(0)	MUT	•								•	•
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 17	NDUFAF6	•									•
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	PUS1	•							•	•	•
Mitochondrial Trifunctional Protein Deficiency, HADHB-Related	HADHB	•									•
Molybdenum Cofactor Deficiency, Type A	MOCS1	•									•
Mucolipidosis II/IIIA	GNPTAB	•								•	•
Mucolipidosis III gamma	GNPTG	•								•	•
Mucolipidosis, Type IV	MCOLN1	•		○	○			•	•	•	•
Mucopolysaccharidosis, Type I (Hurler Syndrome)	IDUA	•						•	•	•	•
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	•								•	•
Myoneurogastrointestinal Encephalopathy (MNGIE)	TYMP	•							•	•	•
Myotubular Myopathy, X-Linked	MTM1		•							•	•
N-acetylglutamate Synthase Deficiency	NAGS	•								•	•
Nemaline Myopathy, NEB-Related	NEB	•							•	•	•
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	•								•	•
Niemann-Pick Disease, Type C1/D	NPC1	•		○						•	•
Niemann-Pick Disease, Type C2	NPC2	•		○						•	•
Niemann-Pick Disease, Types A/B	SMPD1	•		○	○			•	•	•	•
Nijmegen Breakage Syndrome	NBN	•								•	•
Nonsyndromic Hearing Loss, GJB2-Related	GJB2	•							•	•	•
Nonsyndromic Hearing Loss, MYO15A-Related	MYO15A	•									•
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	WNT10A	•								•	•

Horizon Conditions List

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CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY					
				ACOG*	ACMG	H4	H14	H27	H106	H274	H421
Omenn Syndrome, RAG2-Related	RAG2	•							•	•	•
Ornithine Aminotransferase Deficiency	OAT	•							•	•	•
Ornithine Transcarbamylase Deficiency	OTC		•							•	•
Osteopetrosis, Infantile Malignant, TCIRG1-Related	TCIRG1	•							•	•	•
Pendred Syndrome	SLC26A4	•								•	•
Perlman Syndrome	DIS3L2	•									•
Phenylketonuria	PAH	•							•	•	•
Pituitary Hormone Deficiency, Combined 3	LHX3	•								•	•
POLG-Related Disorders	POLG	•									•
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	•					•	•	•	•	•
Pontocerebellar Hypoplasia, EXOSC3-Related	EXOSC3	•									•
Pontocerebellar Hypoplasia, RARS2-Related	RARS2	•							•	•	•
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, 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	•								•	•
Primary Hyperoxaluria, Type 3	HOGA1	•							•	•	•
Progressive Familial Intrahepatic Cholestasis, Type 1 (PFIC1)	ATP8B1	•									•
Progressive Familial Intrahepatic Cholestasis, Type 2 (PFIC2)	ABCB11	•								•	•
Progressive Familial Intrahepatic Cholestasis, Type 4 (PFIC4)	TJP2	•									•
Prolidase Deficiency	PEPD	•									•
Propionic Acidemia, PCCA-Related	PCCA	•								•	•
Propionic Acidemia, PCCB-Related	PCCB	•								•	•
Pseudocholinesterase Deficiency	BCHE	•									•
Pseudoxanthoma Elasticum	ABCC6	•									•
Pycnodysostosis	CTSK	•								•	•
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 25	EYS	•							•	•	•
Retinitis Pigmentosa 26	CERKL	•							•	•	•
Retinitis Pigmentosa 28	FAM161A	•							•	•	•
Retinitis Pigmentosa 59	DHDDS	•							•	•	•
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	•						•	•	•	•
Rhizomelic Chondrodysplasia Punctata, Type 2	GNPAT	•									•
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	•								•	•
Roberts Syndrome	ESCO2	•								•	•
Salla Disease	SLC17A5	•								•	•
Sandhoff Disease	HEXB	•								•	•
Schimke Immunoosseous Dysplasia	SMARCAL1	•								•	•
Segawa Syndrome, TH-Related	TH	•								•	•
Senior-Loken Syndrome 4 / Nephronophthisis 4	NPHP4	•									•
Severe Combined Immunodeficiency, ADA-Related	ADA	•								•	•
Severe Combined Immunodeficiency, RAG1-Related	RAG1	•									•
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	•								•	•
Severe Combined Immunodeficiency, X-Linked	IL2RG		•							•	•
Shwachman-Diamond Syndrome, SBDS-Related	SBDS	•									•
Sialidosis	NEU1	•									•
Sjögren-Larsson Syndrome	ALDH3A2	•								•	•
Smith-Lemli-Opitz Syndrome	DHCR7	•		○			•	•	•	•	•
Spastic Paraplegia, Type 15	ZFYVE26	•									•
Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly (SPATCCM)	SLC1A4	•									•
Spinal Muscular Atrophy	SMN1	•		○	○	•	•	•	•	•	•
Spinocerebellar Ataxia, Autosomal Recessive 12	WWOX	•									•
Spondylothoracic Dysostosis, MESP2-Related	MESP2	•								•	•
Steel Syndrome	COL27A1	•									•
Steroid-Resistant Nephrotic Syndrome	NPHS2	•								•	•
Stuve-Wiedemann Syndrome	LIFR	•								•	•
Tay-Sachs Disease	HEXA	•		○	○		•	•	•	•	•
Trichohepatoenteric Syndrome, TTC37-Related	TTC37	•									•
Trichothiodystrophy 1 / Xeroderma Pigmentosum, Group D	ERCC2	•									•
Triple A Syndrome	AAAS	•									•
Tyrosinemia, Type 1	FAH	•						•	•	•	•
Tyrosinemia, Type 2	TAT	•									•
Usher Syndrome, Type 1B	MYO7A	•								•	•
Usher Syndrome, Type 1C	USH1C	•								•	•
Usher Syndrome, Type 1D	CDH23	•								•	•
Usher Syndrome, Type 1F	PCDH15	•		○					•	•	•

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CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS		PANEL AVAILABILITY					
				ACOG*	ACMG	H4	H14	H27	H106	H274	H421
Usher Syndrome, Type 1J / Deafness, Autosomal Recessive, 48	CIB2	•									•
Usher Syndrome, Type 2A	USH2A	•							•	•	•
Usher Syndrome, Type 2C	ADGRV1	•									•
Usher Syndrome, Type 3	CLRN1	•		◦					•	•	•
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	•								•	•
Vitamin D Dependent Rickets, Type 1A	CYP27B1	•									•
Walker-Warburg Syndrome, FKTN-Related	FKTN	•							•	•	•
Walker-Warburg Syndrome, ISPD-Related	ISPD	•									•
Walker-Warburg Syndrome, LARGE1-Related	LARGE1	•									•
Walker-Warburg Syndrome, POMT1-Related	POMT1	•									•
Walker-Warburg Syndrome, POMT2-Related	POMT2	•									•
Werner Syndrome	WRN	•									•
Wilson Disease	ATP7B	•							•	•	•
Wiskott-Aldrich Syndrome	WAS		•								•
Wolcott-Rallison Syndrome	EIF2AK3	•									•
Wolman Disease	LIPA	•							•	•	•
Xeroderma Pigmentosum, Group A	XPA	•									•
Xeroderma Pigmentosum, Group C	XPC	•									•
X-Linked Chondrodysplasia Punctata 1	ARSE		•								•
X-Linked Lissencephaly with Abnormal Genitalia	ARX		•								•
Zellweger Spectrum Disorders, PEX1-Related	PEX1	•						•	•	•	•
Zellweger Spectrum Disorders, PEX2-Related	PEX2	•							•	•	•
Zellweger Spectrum Disorders, PEX6-Related	PEX6	•							•	•	•
Zellweger Spectrum Disorders, PEX10-Related	PEX10	•								•	•
Zellweger Spectrum Disorders, PEX12-Related	PEX12	•									•
Zellweger Spectrum Disorders, PEX26-Related	PEX26	•									•

* 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.

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