

Horizon Conditions List

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS			PANEL AVAILABILITY			
				ACOG*	ACMG	VICTOR CENTER	H 4	H 27	H 106	H 274
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	HSD3B2	•								•
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMGCL	•								•
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	MCCC1	•								•
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	MCCC2	•								•
3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH	•				o			•	•
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	PTS	•								•
Abetalipoproteinemia	MTTP	•				o			•	•
Achondrogenesis, Type 1B	SLC26A2	•								•
Achromatopsia, CNGB3-Related	CNGB3	•								•
Acrodermatitis Enteropathica	SLC39A4	•								•
Acute Infantile Liver Failure, TRMU-Related	TRMU	•							•	•
Acyl-CoA Oxidase I Deficiency	ACOX1	•								•
Adrenoleukodystrophy, X-Linked	ABCD1		•						•	•
Aicardi-Goutières Syndrome	SAMHD1	•								•
Alpha-Thalassemia Intellectual Disability Syndrome	ATRX		•							•
Alpha-Mannosidosis	MAN2B1	•								•
Alpha-Thalassemia	HBA1/HBA2	•		o			•	•	•	•
Alport Syndrome, COL4A3-Related	COL4A3	•				o			•	•
Alport Syndrome, COL4A4-Related	COL4A4	•								•
Alport Syndrome, X-Linked	COL4A5		•							•
Alstrom Syndrome	ALMS1	•								•
Andermann Syndrome	SLC12A6	•								•
Argininosuccinate Lyase Deficiency	ASL	•								•
Aromatase Deficiency	CYP19A1	•								•
Asparagine Synthetase Deficiency	ASNS	•							•	•
Aspartylglycosaminuria	AGA	•								•
Ataxia with Vitamin E Deficiency	TTPA	•								•
Ataxia-Telangiectasia	ATM	•								•
Autism Spectrum, Epilepsy and Arthrogyrosis	SLC35A3	•				o			•	•
Autoimmune Polyglandular Syndrome, Type 1	AIRE	•							•	•
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	•								•
Bardet-Biedl Syndrome, BBS10-Related	BBS10	•								•
Bardet-Biedl Syndrome, BBS12-Related	BBS12	•								•
Bardet-Biedl Syndrome, BBS1-Related	BBS1	•								•
Bardet-Biedl Syndrome, BBS2-Related	BBS2	•				o			•	•
Bare Lymphocyte Syndrome, CIITA-Related	CIITA	•								•
Bartter Syndrome, BSND-Related	BSND	•								•
Batten Disease, CLN3-Related	CLN3	•					•	•	•	•
Beta-Hemoglobinopathies (including sickle cell disease)	HBB	•		o			•	•	•	•
Bilateral Frontoparietal Polymicrogyria	GPR56	•								•
Biotinidase Deficiency	BTD	•								•
Bloom Syndrome	BLM	•		o	o	o	•	•	•	•
Canavan Disease	ASPA	•		o	o	o	•	•	•	•
Carbamoyl Phosphate Synthetase I Deficiency	CPS1	•								•
Carnitine Deficiency	SLC22A5	•								•
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	•								•
Carnitine Palmitoyltransferase II Deficiency	CPT2	•				o			•	•
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	•								•
Choreoacanthocytosis	VPS13A	•							•	•
Choroideremia	CHM		•							•
Chronic Granulomatous Disease, CYBA-Related	CYBA	•							•	•
Chronic Granulomatous Disease, X-Linked	CYBB		•							•
Ciliopathies, RPGRIP1L-Related	RPGRIP1L	•								•
Citrin Deficiency	SLC25A13	•								•
Citrullinemia, Type I	ASS1	•					•	•	•	•
Cohen Syndrome	VPS13B	•								•
Combined Malonic and Methylmalonic Aciduria	ACSF3	•								•
Combined Oxidative Phosphorylation Deficiency (Complex 4 Deficiency)	GFM1	•								•
Combined Oxidative Phosphorylation Deficiency 3	TSM	•								•
Combined Pituitary Hormone Deficiency-2	PROP1	•								•
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	CYP17A1	•								•
Congenital Amegakaryocytic Thrombocytopenia	MPL	•				o			•	•

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Hypohidrotic Ectodermal Dysplasia, X-Linked	EDA		•							•
Hypophosphatasia, ALPL-Related	ALPL	•								•
Inclusion Body Myopathy 2	GNE	•							•	•
Infantile Cerebral and Cerebellar Atrophy	MED17	•							•	•
Isovaleric Acidemia	IVD	•						•	•	•
Joubert Syndrome 2 / Meckel Syndrome 2	TMEM216	•		o		o			•	•
Juvenile Retinoschisis, X-Linked	RS1		•							•
Ketothiolase Deficiency	ACAT1	•								•
Krabbe Disease	GALC	•								•
Lamellar Ichthyosis, Type 1	TGM1	•								•
Leber Congenital Amaurosis	LCA5	•								•
Leber Congenital Amaurosis 2	RPE65	•							•	•
Leber Congenital Amaurosis, Type CEP290	CEP290	•								•
Leber Congenital Amaurosis, Type RDH12	RDH12	•								•
Leigh Syndrome, French-Canadian Type	LRPPRC	•								•
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 2I	FKRP	•								•
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	•								•
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	•								•
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	•								•
Lipoamide Dehydrogenase Deficiency (Dihydropyridine Dehydrogenase Deficiency)	DLD	•				o			•	•
Lipoid Adrenal Hyperplasia	STAR	•								•
Lipoprotein Lipase Deficiency	LPL	•								•
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	•								•
Lysinuric Protein Intolerance	SLC7A7	•								•
Maple Syrup Urine Disease, Type 1A	BCKDHA	•								•
Maple Syrup Urine Disease, Type 1B	BCKDHB	•		o		o			•	•
Meckel-Gruber Syndrome, Type 1	MKS1	•								•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	•		o				•	•	•
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	•							•	•
Menkes Syndrome	ATP7A		•							•
Metachromatic Leukodystrophy, PSAP-Related	PSAP	•								•
Metachromatic Leukodystrophy, ARSA-Related	ARSA	•							•	•
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 Myopathy and Sideroblastic Anemia (MLASA1)	PUS1	•							•	•
Mucopolysaccharidosis II/IIIA	GNPTAB	•								•
Mucopolysaccharidosis III gamma	GNPTG	•								•
Mucopolysaccharidosis, Type IV	MCOLN1	•		o	o	o		•	•	•
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	SGSH	•								•
Mucopolysaccharidosis Type IX	HYAL1	•								•
Mucopolysaccharidosis, Type I (Hurler Syndrome)	IDUA	•						•	•	•
Mucopolysaccharidosis, Type II (Hunter Syndrome)	IDS		•							•
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	NAGLU	•								•
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	HGSNAT	•								•
Mucopolysaccharidosis, Type IIID (Sanfilippo D)	GNS	•								•
Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	GLB1	•								•
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	ARSB	•								•
Multiple Sulphatase Deficiency	SUMF1	•				o			•	•
Muscle-Eye-Brain Disease, POMGNT1-Related	POMGNT1	•								•
Myoneurogastrointestinal Encephalopathy (MINGIE)	TYMP	•							•	•
Myotubular Myopathy, X-Linked	MTM1		•							•
N-acetylglutamate Synthase Deficiency	NAGS	•								•
Nemaline Myopathy, NEB-Related	NEB	•				o			•	•
Neuronal Ceroid Lipofuscinosis, CLN5-Related	CLN5	•								•
Neuronal Ceroid Lipofuscinosis, MFSD8-Related	MFSD8	•								•
Neuronal Ceroid Lipofuscinosis, PPT1-Related	PPT1	•								•
Neuronal Ceroid Lipofuscinosis, TPP1-Related	TPP1	•								•
Neuronal Ceroid-Lipofuscinosis, CLN6-Related	CLN6	•								•
Neuronal Ceroid-Lipofuscinosis, CLN8-Related	CLN8	•								•
Niemann-Pick Disease, Types C1/D	NPC1	•		o						•
Niemann-Pick Disease, Type C2	NPC2	•		o						•

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Niemann-Pick Disease, Types A/B	SMPD1	•		o	o	o		•	•	•
Nijmegen Breakage Syndrome	NBN	•								•
Non-Syndromic Hearing Loss, GJB2-Related	GJB2	•							•	•
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	WNT10A	•							•	•
Omenn Syndrome, RAG2-Related	RAG2	•							•	•
Ornithine Aminotransferase Deficiency	OAT	•							•	•
Ornithine Transcarbamylase Deficiency	OTC		•							•
Osteopetrosis, Infantile Malignant, TCIRG1-Related	TCIRG1	•							•	•
Pendred Syndrome	SLC26A4	•								•
Phenylketonuria	PAH	•		o					•	•
Pituitary Hormone Deficiency, Combined 3	LHX3	•								•
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	•				o		•	•	•
Pontocerebellar Hypoplasia, RARS2-Related	RARS2	•							•	•
Pontocerebellar Hypoplasia, Type 1A	VRK1	•							•	•
Pontocerebellar Hypoplasia, Type 2D	SEPSECS	•							•	•
Primary Ciliary Dyskinesia, DNAH5-Related	DNAH5	•							•	•
Primary Ciliary Dyskinesia, DNAI1-Related	DNAI1	•							•	•
Primary Ciliary Dyskinesia, DNAI2-Related	DNAI2	•							•	•
Primary Hyperoxaluria, Type 1	AGXT	•								•
Primary Hyperoxaluria, Type 2	GRHPR	•								•
Primary Hyperoxaluria, Type 3	HOGA1	•							•	•
Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	•								•
Propionic Acidemia, PCCA-Related	PCCA	•								•
Propionic Acidemia, PCCB-Related	PCCB	•								•
Pycnodysostosis	CTSK	•								•
Pyruvate Dehydrogenase Deficiency, PDHB-Related	PDHB	•								•
Pyruvate Dehydrogenase Deficiency, X-Linked	PDHA1		•							•
Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	ATP6V1B1	•							•	•
Retinitis Pigmentosa 25	EYS	•							•	•
Retinitis Pigmentosa 26	CERKL	•							•	•
Retinitis Pigmentosa 28	FAM161A	•							•	•
Retinitis Pigmentosa 59	DHDDS	•				o			•	•
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	•								•
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	•						•	•	•
Roberts Syndrome	ESCO2	•								•
Salla Disease	SLC17A5	•								•
Sandhoff Disease	HEXB	•								•
Schimke Immunoskeletal Dysplasia	SMARCAL1	•								•
Segawa Syndrome, TH-Related	TH	•								•
Severe Combined Immunodeficiency, ADA-Related	ADA	•								•
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	•								•
Severe Combined Immunodeficiency, X-Linked	IL2RG		•							•
Sjogren-Larsson Syndrome	ALDH3A2	•								•
Smith-Lemli-Opitz Syndrome	DHCR7	•		o		o		•	•	•
Spinal Muscular Atrophy	SMN1	•		o	o	o	•	•	•	•
Spondylothoracic Dysostosis, MESP2-Related	MESP2	•								•
Steroid-Resistant Nephrotic Syndrome	NPHS2	•								•
Stuve-Wiedemann Syndrome	LIFR	•								•
Tay-Sachs Disease	HEXA	•		o	o	o		•	•	•
Tyrosinemia, Type I	FAH	•				o		•	•	•
Usher Syndrome, Type 1B	MYO7A	•								•
Usher Syndrome, Type 1C	USH1C	•								•
Usher Syndrome, Type 1D	CDH23	•								•
Usher Syndrome, Type 1F	PCDH15	•		o		o			•	•
Usher Syndrome, Type 2A	USH2A	•							•	•
Usher Syndrome, Type 3	CLRN1	•		o		o			•	•
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	•								•
Walker-Warburg Syndrome, FKTN-Related	FKTN	•				o			•	•
Wilson Disease	ATP7B	•				o			•	•
Wolman Disease	LIPA	•							•	•
Zellweger Spectrum Disorders, PEX10-Related	PEX10	•								•
Zellweger Spectrum Disorders, PEX1-Related	PEX1	•						•	•	•
Zellweger Spectrum Disorders, PEX2-Related	PEX2	•				o			•	•
Zellweger Spectrum Disorders, PEX6-Related	PEX6	•							•	•

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