Horizon Conditions List

natera™ | Horizon™ Advanced carrier screening

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 Dysplasia 13 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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