horizon™
natera carrier screen

Horizon Conditions List

CONDITION	GENE	SCREENING RECOMMENDATIONS			PANEL AVAILABILITY				
		ACOG	ACMG	VICTOR CENTER	H 4	H 27	H 106	H 137	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 (Diastrophic Dysplasia)	*SLC26A2*							•	•
Achromatopsia	*CNGB3*								•
Acrodermatitis Enteropathica	*SLC39A4*								•
Acute Infantile Liver Failure	*TRMU*						•		•
Acyl-CoA Oxidase I Deficiency	*ACOX1*								•
Adrenoleukodystrophy	*ABCD1*						•	•	•
Aicardi-Goutières Syndrome	*SAMHD1*								•
Alpha-Thalassemia and Mental Retardation	*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 (Early Onset Myocardial Infarction, Association with Leber Congenital Amaurosis, Diabetes Type 1)	*ALMS1*								•
Andermann Syndrome (Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum)	*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 Arthrogryposis	*SLC35A3*			o			•	•	•
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, Type II	*CIITA*								•
Bartter Syndrome	*BSND*								•
Batten Disease (Neuronal Ceroid Lipofuscinosis, CLN3-Related)	*CLN3*					•	•	•	•
Beta-Hemoglobinopathies	*HBB*	o				•	•	•	•
Bilateral Frontoparietal Polymicrogyria	*GPR56*								•
Biotinidase Deficiency	*BTD*							•	•
Bloom Syndrome	*BLM*		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, Cytochrome b-negative	*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	*TSFM*								•

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Combined Pituitary Hormone Deficiency-2	*PROP1*							•	•
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	*CYP17A1*								•
Congenital Amegakaryocytic Thrombocytopenia	*MPL*			o			•	•	•
Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	*PMM2*			o			•	•	•
Congenital Disorder of Glycosylation, Type 1C	*ALG6*								•
Congenital Disorder of Glycosylation, Type IB	*MPI*							•	•
Congenital Finnish Nephrosis	*NPHS1*							•	•
Congenital Insensivity to Pain with Anhidrosis (CIPA)	*NTRK1*						•		•
Congenital Myasthenic Syndrome, CHRNE-Related	*CHRNE*							•	•
Congenital Myasthenic Syndrome, RAPSN-Related	*RAPSN*						•	•	•
Congenital Neutropenia, HAX1-Related	*HAX1*								•
Congenital Neutropenia, VPS45-Related	*VPS45*								•
Corneal Dystrophy and Perceptive Deafness	*SLC4A11*								•
Corticosterone Methyloxidase Deficiency	*CYP11B2*						•		•
Costeff Disease Optic Atrophy (3-Methylglutaconic Aciduria, Type III)	*OPA3*						•		•
CRB1-Related Retinal Dystrophies	*CRB1*								•
Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1)	*SLC6A8*								•
Cystic Fibrosis	*CFTR*	o	o	o	•	•	•	•	•
Cystinosis	*CTNS*						•	•	•
D-Bifunctional Protein Deficiency	*HSD17B4*							•	•
Deafness, Autosomal Recessive 77	*LOXHD1*						•		•
Duchenne/Becker Muscular Dystrophy	*DMD*				•	•	•	•	•
Dyskeratosis Congenita	*RTEL1*			o			•	•	•
Dytrophic Epidermolysis Bullosa, Autosomal Recessive	*COL7A1*								•
Ehlers-Danlos Syndrome, Type VIIC	*ADAMTS2*			o			•	•	•
Ellis-van Creveld syndome (Weyers Acrofacial Dysostosis)	*EVC*								•
Emery-Dreifuss Muscular Dystrophy 1, X-linked	*EMD*								•
Enhanced S-Cone Syndrome (Goldmann-Favre Syndrome)	*NR2E3*						•		•
Ethylmalonic Encephalopathy	*ETHE1*							•	•
Fabry Disease	*GLA*								•
Factor IX Deficiency	*F9*								•
Factor XI Deficiency	*F11*						•		•
Familial Dysautonomia	*IKBKAP*	o	o	o		•	•	•	•
Familial Hypercholesterolemia, LDLRAP1-Related	*LDLRAP1*								•
Familial Hypercholesterolemia, LDLR-Related	*LDLR*						•		•
Familial Hyperinsulinism	*ABCC8*			o			•	•	•
Familial Mediterranean Fever	*MEFV*						•		•
Familial Neuropophyseal Diabetes Insipidus, Autosomal Recessive	*AQP2*								•
Fanconi Anemia, Group A	*FANCA*						•		•
Fanconi Anemia, Group C	*FANCC*		o	o		•	•	•	•
Fanconi Anemia, Group G	*FANCG*								•
Fragile X Syndrome	*FMR1*			o	•	•	•	•	•
Fumarase Deficiency	*FH*							•	•
Galactokinase Deficiency (Galactosemia, Type II)	*GALK1*								•
Galactosemia	*GALT*			o		•	•	•	•
Gaucher Disease	*GBA*		o	o		•	•	•	•
Gitelman Syndrome	*SLC12A3*								•
Glutaric Acidemia, Type IIA	*ETFA*								•
Glutaric Acidemia, Type IIC	*ETFDH*								•
Glutaryl-CoA Dehydrogenase Deficiency (Glutaric Acidemia, Type 1)	*GCDH*							•	•
Glycine Encephalopathy	*GLDC*							•	•
Glycine Encephalopathy, AMT-Related	*AMT*							•	•
Glycogen Storage Disease, Type 1A	*G6PC*			o		•	•	•	•
Glycogen Storage Disease, Type 2 (Pompe Disease)	*GAA*						•	•	•
Glycogen Storage Disease, Type 1B	*SLC37A4*							•	•
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 (Mitochondrial Respiratory Chain Complex III Deficiency)	*BCS1L*							•	•
Guanidinoacetate Methyltransferase Deficiency	*GAMT*								•
Hemochromatosis, Type 2A	*HFE2*								•
Hemochromatosis, Type 3, TFR2-Related	*TFR2*								•
Hereditary Fructose Intolerance	*ALDOB*							•	•
Hereditary Spastic Paraparesis, Type 49	*TECPR2*						•		•
Hermansky-Pudlak Syndrome, HPS1-Related	*HPS1*								•
Hermansky-Pudlak Syndrome, HPS3-Related	*HPS3*						•	•	•
Holocarboxylase Synthetase Deficiency	*HLCS*								•
Homocystinuria	*CBS*							•	•
Homocystinuria due to Deficiency of MTHFR	*MTHFR*						•		•
Homocystinuria, Type cblE	*MTRR*								•

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Hydrolethalus Syndrome	*HYLS1*								•
Hyperinsulinemic Hypoglycemia	*KCNJ11*								•
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH Syndrome)	*SLC25A15*							•	•
Hypohidrotic Ectodermal Dysplasia, X-linked	*EDA*								•
Hypophosphatasia, Autosomal Recessive	*ALPL*							•	•
Inclusion Body Myopathy 2	*GNE*						•	•	•
Infantile Cerebral and Cerebellar Atrophy	*MED17*						•		•
Isovaleric Acidemia	*IVD*					•	•	•	•
Joubert Syndrome 2	*TMEM216*			o			•	•	•
Ketothiolase Deficiency	*ACAT1*								•
Krabbe Disease	*GALC*							•	•
Lamellar Ichthyosis, Type 1	*TGM1*							•	•
Leber Congenital Amaurosis, Type LCA5	*LCA5*								•
Leber Congenital Amaurosis 2 (Retinitis Pigmentosa 20)	*RPE65*						•		•
Leber Congenital Amaurosis, Type CEP290	*CEP290*								•
Leber Congenital Amaurosis, Type RDH12	*RDH12*								•
Leigh Syndrome, French-Canadian Type	*LRPPRC*							•	•
Lethal Congenital Contracture Syndrome 1 (Lethal Arthrogryposis with Anterior Horn Cell Disease)	*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 (Maple Syrup Urine Disease, Type III)	*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			•	•	•
Meckel-Gruber Syndrome, Type 1	*MKS1*								•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*					•	•	•	•
Megalencephalic Leukoencephalopathy with Subcortical Cysts	*MLC1*						•	•	•
Metachromatic Leukodystrophy (Gaucher Disease, atypical)	*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*						•		•
Mitochondrial Complex 1 Deficiency, NDUFAF5-Related	*NDUFAF5*						•		•
Mitochondrial Complex 1 Deficiency, NDUFS6-Related	*NDUFS6*						•		•
Mitochondrial DNA Depletion Syndrome 6 (Hepatocerebral Type)	*MPV17*								•
Mitochondrial Myopathy and Sideroblastic Anemia (MLAS1)	*PUS1*						•		•
Mucolipidosis II/IIIA	*GNPTAB*							•	•
Mucolipidosis III gamma	*GNPTG*								•
Mucolipidosis, Type IV	*MCOLN1*		o	o		•	•	•	•
Mucopolysaccharidisis, 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	*GNS*								•
Mucopolysaccharidosis, Type IVB (GM1 Gangliosidosis)	*GLB1*							•	•
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	*ARSB*								•
Multiple Sulphatase Deficiency	*SUMF1*			o			•	•	•
Muscle-Eye-Brain Disease, POMGNT1-Related (Muscular dystrophy-dystroglycanopathy, Type C)	*POMGNT1*							•	•
Myoneurogastrointestinal Encephalopathy (MNGIE)	*TYMP*						•		•
Myotubular Myopathy, X-linked	*MTM1*								•
N-acetylglutamate Synthase Deficiency	*NAGS*								•
Nemaline Myopathy	*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*							•	•

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		ACOG	ACMG	VICTOR CENTER	H 4	H 27	H 106	H 137	H 274
Neuronal Ceroid-Lipofuscinosis, CLN6-Related	*CLN6*							•	•
Neuronal Ceroid-Lipofuscinosis, CLN8-Related	*CLN8*							•	•
Niemann-Pick Disease, Types C1/D	*NPC1*							•	•
Niemann-Pick Disease, Type C2	*NPC2*								•
Niemann-Pick Disease, Types A/B	*SMPD1*		o	o		•	•	•	•
Nijmegen Breakage Syndrome	*NBN*							•	•
Non-syndromic Hearing Loss	*GJB2*						•	•	•
Occipital Horn Syndrome (Motor neuropathy, distal)	*ATP7A*								•
Odonto-onycho-dermal Dysplasia (Schopf-Schulz-Passarge Syndrome)	*WNT10A*								•
Omenn Syndrome	*RAG2*						•		•
Ornithine Aminotransferase Deficiency	*OAT*						•		•
Ornithine Transcarbamylase Deficiency	*OTC*							•	•
Osteopetrosis, Infantile Malignant (Osteopetrosis, autosomal recessive)	*TCIRG1*						•		•
Pendred Syndrome	*SLC26A4*							•	•
Phenylketonuria	*PAH*						•	•	•
Pituitary Hormone Deficiency, Combined 3	*LHX3*								•
Polycystic Kidney Disease, Autosomal Recessive	*PKHD1*			o		•	•	•	•
Polyglandular Autoimmune Syndrome	*AIRE*						•		•
Pontocerebellar Hypoplasia, RARS2-Related	*RARS2*						•		•
Pontocerebellar Hypoplasia, Type 1A	*VRK1*						•		•
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 Cerebello-Cerebral Atrophy	*SEPSECS*						•		•
Progressive Familial Intrahepatic Cholestasis, Type 2	*ABCB11*								•
Propionic Acidemia, alpha subunit	*PCCA*							•	•
Propionic Acidemia, beta subunit	*PCCB*							•	•
Pycnodysostosis	*CTSK*							•	•
Pyruvate Dehydrogenase Deficiency, Autosomal Recessive	*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 (Akyl-DHAP Synthase Deficiency)	*AGPS*								•
Rhizomelic Chondrodysplasia Punctata, Type 1	*PEX7*					•	•	•	•
Riboflavin Responsive Complex 1 Deficiency (Acyl-CoEnzyme Dehydrogenase 9 Deficiency)	*ACAD9*								•
Roberts Syndrome	*ESCO2*								•
Salla Disease	*SLC17A5*							•	•
Sandhoff Disease	*HEXB*							•	•
Schimke Immunoosseous Dysplasia	*SMARCAL1*								•
Segawa Syndrome, Autosomal Recessive	*TH*							•	•
Severe Combined Immunodeficiency (Adenosine Deaminase Deficiency)	*ADA*							•	•
Severe Combined Immunodeficiency, Type Athabaskan	*DCLRE1C*							•	•
Sjogren-Larsson Syndrome	*ALDH3A2*							•	•
Smith-Lemli-Opitz Syndrome	*DHCR7*			o		•	•	•	•
Spinal Muscular Atrophy	*SMN1*		o	o	•	•	•	•	•
Spondylothoracic Dysostosis	*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			•	•	•
Usher Syndrome, Type 2A	*USH2A*						•	•	•
Usher Syndrome, Type 3	*CLRN1*			o			•	•	•
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	*ACADVL*							•	•
Walker-Warburg Syndrome	*FKTN*			o			•	•	•
Wilson Disease	*ATP7B*			o			•	•	•
Wolman Disease	*LIPA*						•		•
X-Linked Juvenile Retinoschisis	*RS1*								•
X-linked Severe Combined Immunodeficiency	*IL2RG*							•	•
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*						•		•

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