natera™ | Horizon™ Advanced carrier screening

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 Arthrogryposis	*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	*TSFM*	•								•
Combined Pituitary Hormone Deficiency-2	*PROP1*	•								•
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	*CYP17A1*	•								•
Congenital Amegakaryocytic Thrombocytopenia	*MPL*	•				o			•	•

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				ACOG*	ACMG	VICTOR CENTER	H 4	H 27	H 106	H 274
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 Hyperinsulinism, KCNJ11-Related	*KCNJ11*	•								•
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 Syndrome (3-Methylglutaconic Aciduria, Type 3)	*OPA3*	•							•	•
CRB1-Related Retinal Dystrophies	*CRB1*	•								•
Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1, X-Linked)	*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-Related	*RTEL1*	•				o			•	•
Dystrophic Epidermolysis Bullosa, COL7A1-Related	*COL7A1*	•								•
Ehlers-Danlos Syndrome, Type VIIC	*ADAMTS2*	•				o			•	•
Ellis-van Creveld Syndrome, EVC-Related	*EVC*	•								•
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	*EMD*		•							•
Enhanced S-Cone 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-Related	*ABCC8*	•		o		o			•	•
Familial Mediterranean Fever	*MEFV*	•							•	•
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	*AQP2*	•								•
Fanconi Anemia, Group A	*FANCA*	•		o					•	•
Fanconi Anemia, Group C	*FANCC*	•		o	o	o		•	•	•
Fanconi Anemia, Group G	*FANCG*	•		o						•
Fragile X Syndrome	*FMR1*		•	o		o	•	•	•	•
Fumarase Deficiency	*FH*	•								•
Galactokinase Deficiency (Galactosemia, Type II)	*GALK1*	•								•
Galactosemia	*GALT*	•		o		o		•	•	•
Gaucher Disease	*GBA*	•		o	o	o		•	•	•
Gitelman Syndrome	*SLC12A3*	•								•
Glutaric Acidemia, Type 2A	*ETFA*	•								•
Glutaric Acidemia, Type 2C	*ETFDH*	•								•
Glutaric Acidemia, Type 1	*GCDH*	•								•
Glycine Encephalopathy, GLDC-Related	*GLDC*	•								•
Glycine Encephalopathy, AMT-Related	*AMT*	•								•
Glycogen Storage Disease, Type 1A	*G6PC*	•		o		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	*BCS1L*	•								•
Guanidinoacetate Methyltransferase Deficiency	*GAMT*	•								•
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, HPS1-Related	*HPS1*	•								•
Hermansky-Pudlak Syndrome, HPS3-Related	*HPS3*	•							•	•
Holocarboxylase Synthetase Deficiency	*HLCS*	•								•
Homocystinuria, CBS-Related	*CBS*	•								•
Homocystinuria due to Deficiency of MTHFR	*MTHFR*	•							•	•
Homocystinuria, Type cblE	*MTRR*	•								•
Hydrolethalus Syndrome	*HYLS1*	•								•
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH Syndrome)	*SLC25A15*	•								•

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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 (Dihydrolipoamide 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*	•							•	•
Mucolipidosis II/IIIA	*GNPTAB*	•								•
Mucolipidosis III gamma	*GNPTG*	•								•
Mucolipidosis, Type IV	*MCOLN1*	•		o	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 (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 (MNGIE)	*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 Immunoosseous 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.

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