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 (ADGRG1)*	●								●	●
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 (ELP1)*	●		○	○		●	●	●	●	●
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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				ACOG*	ACMG	H4	H14	H27	H106	H274	H421
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 (HJV)*	●								●	●
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 (MMUT)*	●			○					●	●
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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				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 (USH3)*	●		○	○				●	●	●
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 (CRPPA)*	●									●
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 (ARSL)*		●								●
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.

1. American College of Obstetricians and Gynecologists, Committee Opinion # 690, March 2017.
2. American College of Obstetricians and Gynecologists, Committee Opinion # 691, March 2017.
3. Gregg et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG), July 2021.

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