genomic diagnostics
A Healius company

AUTHORIZED DISTRIBUTOR
natera™ | Horizon™ Advanced carrier screening

Expanded Carrier Screen - Conditions List

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
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**	•			O
3-Phosphoglycerate Dehydrogenase Deficiency	*PHGDH*	•			
5-Alpha-Reductase Deficiency	*SRD5A2*	•			
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	*PTS*	•			
Abetalipoproteinemia	*MTTP*	•			
Achondrogenesis, Type 1B	*SLC26A2*	•			O
Achromatopsia, CNGB3-Related	*CNGB3*	•			O
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*		•		O
Adrenoleukodystrophy, X-Linked	*ABCD1*		•		O
Agammaglobulinemia, X-Linked	*BTK*		•		
Aicardi-Goutières Syndrome	*SAMHD1*	•			
Aicardi-Goutières Syndrome, RNASEH2A-Related	*RNASEH2A*	•			
Aicardi-Goutières Syndrome, RNASEH2B-Related	*RNASEH2B*	•			O
Aicardi-Goutières Syndrome, RNASEH2C-Related	*RNASEH2C*	•			
Aicardi-Goutières Syndrome, TREX1-Related	*TREX1*	•			
Alkaptonuria	*HGD**	•			
Alpha-Mannosidosis	*MAN2B1*	•			
Alpha-Thalassemia	*HBA1/HBA2*	•		O	O
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*	•			
Androgen Insensitivity Syndrome	*AR*		•		
Arginine:Glycine Amidinotransferase Deficiency (Agat Deficiency)	*GATM*	•			
Argininemia	*ARG1*	•			
Argininosuccinate Lyase Deficiency	*ASL*	•			O
Aromatase Deficiency	*CYP19A1*	•			
Arts Syndrome	*PRPS1*		•		
Asparagine Synthetase Deficiency	*ASNS*	•			
Aspartylglycosaminuria	*AGA*	•			O
Ataxia with Vitamin E Deficiency	*TTPA*	•			
Ataxia-Telangiectasia	*ATM*	•			
Ataxia-Telangiectasia-Like Disorder 1	*MRE11*	•			
Atransferrinemia	*TF*	•			O
Autism Spectrum, Epilepsy and Arthrogryposis	*SLC35A3*	•			
Autoimmune Polyglandular Syndrome, Type 1	*AIRE*	•			O
Autosomal Recessive Congenital Ichthyosis (ARCI), SLC27A4-Related	*SLC27A4*	•			
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	•			
Bardet-Biedl Syndrome, ARL6-Related	*ARL6*	•			

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Bardet-Biedl Syndrome, BBS10-Related	*BBS10*	•			
Bardet-Biedl Syndrome, BBS12-Related	*BBS12*	•			
Bardet-Biedl Syndrome, BBS1-Related	*BBS1*	•			O
Bardet-Biedl Syndrome, BBS2-Related	*BBS2*	•			O
Bardet-Biedl Syndrome, BBS4-Related	*BBS4*	•			
Bardet-Biedl Syndrome, Bbs5-Related	*BBS5*	•			
Bardet-Biedl Syndrome, BBS7-Related	*BBS7*	•			
Bardet-Biedl Syndrome, BBS9-Related	*BBS9*	•			
Bardet-Biedl Syndrome, TTC8-Related	*TTC8*	•			
Bare Lymphocyte Syndrome, CIITA-Related	*CIITA*	•			
Barth Syndrome	*TAZ*		•		
Bartter Syndrome, BSND-Related	*BSND*	•			
Bartter Syndrome, KCNJ1-Related	*KCNJ1*	•			
Bartter Syndrome, SLC12A1-Related	*SLC12A1*	•			
Batten Disease, CLN3-Related	*CLN3*	•			
Beta-Hemoglobinopathies	*HBB*	•		O	O
Beta-Mannosidosis	*MANBA*	•			
Beta-Ureidopropionase Deficiency	*UPB1*	•			
Bilateral Frontoparietal Polymicrogyria	*GPR56*	•			
Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD)	*SLC19A3*	•			O
Bloom Syndrome	*BLM*	•		O	O
Brittle Cornea Syndrome 1	*ZNF469*	•			
Brittle Cornea Syndrome 2	*PRDM5*	•			
Canavan Disease	*ASPA*	•		O	O
Carbamoyl Phosphate Synthetase I Deficiency	*CPS1*	•			
Carnitine Deficiency	*SLC22A5*	•			
Carnitine Palmitoyltransferase IA Deficiency	*CPT1A*	•			
Carnitine Palmitoyltransferase II Deficiency	*CPT2*	•			O
Carnitine-Acylcarnitine Translocase Deficiency	*SLC25A20*	•			
Carpenter Syndrome	*RAB23*	•			
Cartilage-Hair Hypoplasia	*RMRP*	•			
Catecholaminergic Polymorphic Ventricular Tachycardia	*CASQ2*	•			
CD59-Mediated Hemolytic Anemia	*CD59*	•			
CEP152-Related Microcephaly	*CEP152*	•			
Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma (CEDNIK) Syndrome	*SNAP29*	•			
Cerebrotendinous Xanthomatosis	*CYP27A1*	•			O
Charcot-Marie-Tooth Disease with Deafness, X-Linked	*GJB1*		•		
Charcot-Marie-Tooth Disease, Recessive Intermediate C	*PLEKHG5*	•			
Charcot-Marie-Tooth Disease, Type 4D	*NDRG1*	•			
Chediak-Higashi Syndrome	*LYST*	•			
Choreoacanthocytosis	*VPS13A*	•			
Choroideremia	*CHM*		•		
Chronic Granulomatous Disease, CYBA-Related	*CYBA*	•			
Chronic Granulomatous Disease, NCF2-Related	*NCF2*	•			
Chronic Granulomatous Disease, X-Linked	*CYBB*		•		
Ciliopathies, RPGRIP1L-Related	*RPGRIP1L*	•			
Citrin Deficiency	*SLC25A13*	•			
Citrullinemia, Type 1	*ASS1*	•			
CLN10 Disease	*CTSD*	•			
Cohen Syndrome	*VPS13B*	•			
COL11A2-Related Conditions	*COL11A2*	•			
Combined Malonic and Methylmalonic Aciduria	*ACSF3*	•			
Combined Oxidative Phosphorylation Deficiency 1	*GFM1*	•			
Combined Oxidative Phosphorylation Deficiency 3	*TSFM*	•			
Combined Pituitary Hormone Deficiency 1	*POU1F1*	•			
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*	•			O
Congenital Adrenal Insufficiency, CYP11A1-Related	*CYP11A1*	•			O

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Congenital Amegakaryocytic Thrombocytopenia	*MPL*	•			
Congenital Chronic Diarrhea	*DGAT1*	•			
Congenital Disorder Of Glycosylation Type 1, ALG1-Related	*ALG1*	•			
Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	*PMM2*	•			O
Congenital Disorder of Glycosylation, Type 1B	*MPI*	•			
Congenital Disorder of Glycosylation, Type 1C	*ALG6*	•			
Congenital Dyserythropoietic Anemia Type 2	*SEC23B*	•			
Congenital Finnish Nephrosis	*NPHS1*	•			O
Congenital Hydrocephalus 1	*CCDC88C*	•			O
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*	•			O
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, G6PC3-Related	*G6PC3*	•			
Congenital Neutropenia, HAX1-Related	*HAX1*	•			
Congenital Neutropenia, VPS45-Related	*VPS45*	•			
Congenital Secretory Chloride Diarrhea 1	*SLC26A3*	•			
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*		•		O
Cystic Fibrosis	*CFTR*	•		O	O
Cystinosis	*CTNS*	•			
Cytochrome C Oxidase Deficiency, PET100-Related	*PET100*	•			
Cytochrome P450 Oxioreductase Deficiency	*POR*	•			
D-Bifunctional Protein Deficiency	*HSD17B4*	•			
Deafness, Autosomal Recessive 77	*LOXHD1*	•			
Dent Disease, Type 1	*CLCN5*		•		
Dent Disease, Type 2 / Lowe Syndrome	*OCRL*		•		
Developmental And Epilieptic Encephalopathy 36	*ALG13*		•		
Dihydropteridine Reductase (DHPR) Deficiency	*QDPR*	•			
Dihydropyrimidine Dehydrogenase Deficiency	*DPYD*	•			
Donnai-Barrow Syndrome	*LRP2*	•			O
Dubin-Johnson Syndrome	*ABCC2*	•			
Duchenne/Becker Muscular Dystrophy	*DMD*		•		O
Dyskeratosis Congenita Spectrum Disorders	*TERT*	•			
Dyskeratosis Congenita, DKC1-Related	*DKC1*		•		
Dyskeratosis Congenita, RTEL1-Related	*RTEL1*	•			
Dystrophic Epidermolysis Bullosa, COL7A1-Related	*COL7A1*	•			O
Early Infantile Epileptic Encephalopathy, CAD-Related	*CAD*	•			
Ehlers-Danlos Syndrome Type VI	*PLOD1*	•			
Ehlers-Danlos Syndrome, Classic-Like, TNXB-Related	*TNXB*	•			O
Ehlers-Danlos Syndrome, Type VIIC	*ADAMTS2*	•			
Ellis-van Creveld Syndrome, EVC2-Related	*EVC2*	•			O
Ellis-van Creveld Syndrome, EVC-Related	*EVC*	•			
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	*EMD*		•		
Emery-Dreifuss Muscular Dystrophy 6, X Linked	*FHL1*		•		
Enhanced S-Cone Syndrome	*NR2E3*	•			
Epimerase Deficiency (Galactosemia Type III)	*GALE*	•			
Epiphyseal Dysplasia, Multiple, 7 / Desbuquois Dysplasia 1	*CANT1*	•			
ERCC6-Related Disorders	*ERCC6*	•			
ERCC8-Related Disorders	*ERCC8*	•			
Ethylmalonic Encephalopathy	*ETHE1*	•			
Fabry Disease	*GLA*		•		O

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Factor IX Deficiency	*F9*		•		O
Familial Dysautonomia	*IKBKAP*	•		O	O
Familial Hemophagocytic Lymphohistiocytosis, PRF1-Related	*PRF1*	•			O
Familial Hemophagocytic Lymphohistiocytosis, STX11-Related	*STX11*	•			
Familial Hemophagocytic Lymphohistiocytosis, STXBP2-Related	*STXBP2*	•			
Familial Hemophagocytic Lymphohistiocytosis, UNC13D-Related	*UNC13D*	•			
Familial Hypercholesterolemia, LDLRAP1-Related	*LDLRAP1*	•			
Familial Hypercholesterolemia, LDLR-Related	*LDLR*	•			
Familial Hyperinsulinism, ABCC8-Related	*ABCC8*	•		O	O
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	*AQP2*	•			
Fanconi Anemia Group J	*BRIP1*	•			
Fanconi Anemia, Group A	*FANCA*	•		O	
Fanconi Anemia, Group B	*FANCB*		•		
Fanconi Anemia, Group C	*FANCC*	•		O	O
Fanconi Anemia, Group D2	*FANCD2*	•			
Fanconi Anemia, Group E	*FANCE*	•			
Fanconi Anemia, Group F	*FANCF*	•			
Fanconi Anemia, Group G	*FANCG*	•		O	
Fanconi Anemia, Group I	*FANCI*	•			
Fanconi Anemia, Group L	*FANCL*	•			
Farber Lipogranulomatosis	*ASAH1*	•			
Foveal Hypoplasia	*SLC38A8*	•			
Fragile X Syndrome	*FMR1*		•	O	O
Fragile XE Syndrome	*AFF2*		•		O
Fraser Syndrome 3, GRIP1-Related	*GRIP1*	•			O
Fraser Syndrome, FRAS1-Related	*FRAS1*	•			
Fraser Syndrome, FREM2-Related	*FREM2*	•			
Friedreich Ataxia	*FXN*	•			O
Fructose-1,6-Bisphosphatase Deficiency	*FBP1*	•			
Fucosidosis	*FUCA1*	•			
Fumarase Deficiency	*FH*	•			
GABA-Transaminase Deficiency	*ABAT*	•			
Galactokinase Deficiency (Galactosemia, Type II)	*GALK1*	•			
Galactosemia	*GALT*	•			O
Galactosialidosis	*CTSA*	•			
Gaucher Disease	*GBA*	•			O
GCH1-Related Conditions	*GCH1*	•			
GDF5-Related Conditions	*GDF5*	•			
Geroderma Osteodysplastica	*GORAB*	•			
Gitelman Syndrome	*SLC12A3*	•			
Glanzmann Thrombasthenia	*ITGB3*	•			
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*	•			
Glutathione Synthetase Deficiency	*GSS*	•			
Glycine Encephalopathy, AMT-Related	*AMT*	•			
Glycine Encephalopathy, GLDC-Related	*GLDC*	•			
Glycogen Storage Disease Type IXb	*PHKB*	•			
Glycogen Storage Disease Type IXc	*PHKG2*	•			
Glycogen Storage Disease, Type 1A	*G6PC*	•		O	O
Glycogen Storage Disease, Type 1B	*SLC37A4*	•		O	O
Glycogen Storage Disease, Type 2 (Pompe Disease)	*GAA*	•			O
Glycogen Storage Disease, Type 3	*AGL*	•			
Glycogen Storage Disease, Type 4	*GBE1*	•			O
Glycogen Storage Disease, Type 5 (McArdle Disease)	*PYGM*	•			
Glycogen Storage Disease, Type 7	*PFKM*	•			
GRACILE Syndrome	*BCS1L*	•			
Guanidinoacetate Methyltransferase Deficiency	*GAMT*	•			
Harlequin Ichthyosis	*ABCA12*	•			

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Heme Oxygenase 1 Deficiency	*HMOX1*	•			
Hemochromatosis, Type 2A	*HFE2*	•			
Hemochromatosis, Type 3, TFR2-Related	*TFR2*	•			
Hemophilia A	*F8*		•		O
Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Related	*MPV17*	•			
Hereditary Fructose Intolerance	*ALDOB*	•			O
Hereditary Hemochromatosis Type 2B	*HAMP*	•			
Hereditary Spastic Paraparesis, Type 49	*TECPR2*	•			
Hereditary Spastic Paraplegia, CYP7B1-Related	*CYP7B1*	•			
Hermansky-Pudlak Syndrome, AP3B1-Related	*AP3B1*	•			
Hermansky-Pudlak Syndrome, BLOC1S3-Related	*BLOC1S3*	•			
Hermansky-Pudlak Syndrome, BLOC1S6-Related	*BLOC1S6*	•			
Hermansky-Pudlak Syndrome, HPS1-Related	*HPS1*	•			O
Hermansky-Pudlak Syndrome, HPS3-Related	*HPS3*	•			O
Hermansky-Pudlak Syndrome, HPS4-Related	*HPS4*	•			
Hermansky-Pudlak Syndrome, HPS5-Related	*HPS5*	•			
Hermansky-Pudlak Syndrome, HPS6-Related	*HPS6*	•			
Heterotaxy Syndrome, ZIC3-Related	*ZIC3*		•		
Holocarboxylase Synthetase Deficiency	*HLCS*	•			
Homocystinuria And Megaloblastic Anemia Type Cblg	*MTR*	•			
Homocystinuria due to Deficiency of MTHFR	*MTHFR*	•			
Homocystinuria, CBS-Related	*CBS*	•			O
Homocystinuria, Type cblE	*MTRR*	•			
HSD10 Disease	*HSD17B10*		•		
Hydrolethalus Syndrome	*HYLS1*	•			
Hyper IgM Syndrome, X-Linked	*CD40LG*		•		
Hyper-IgM Immunodeficiency	*CD40*	•			
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH Syndrome)	*SLC25A15*	•			
Hyperphosphatemic Familial Tumoral Calcinosis, GALNT3-Related	*GALNT3*	•			
Hypohidrotic Ectodermal Dysplasia, X-Linked	*EDA*		•		
Hypomyelinating Leukodystrophy 12	*VPS11*	•			
Hypophosphatasia, ALPL-Related	*ALPL*	•			O
Imerslund-Gräsbeck Syndrome 2	*AMN*	•			
Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome	*FOXP3*		•		
Immunodeficiency-Centromeric Instability-Facial Anomalies (ICF) Syndrome, DNMT3B-Related	*DNMT3B*	•			
Immunodeficiency-Centromeric Instability-Facial Anomalies (ICF) Syndrome, ZBTB24-Related	*ZBTB24*	•			
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 Ectopia Lentis	*ADAMTSL4*	•			
Isolated Lissencephaly Sequence / Subcortical Band Heterotopia	*DCX*		•		
Isolated Sulfite Oxidase Deficiency	*SUOX*	•			
Isolated Thyroid-Stimulating Hormone Deficiency	*TSHB*	•			
Isovaleric Acidemia	*IVD*	•			
Johanson-Blizzard Syndrome	*UBR1*	•			
Joubert Syndrome 2 / Meckel Syndrome 2	*TMEM216*	•		O	O
Joubert Syndrome And Related Disorders (JSRD), TMEM67-Related	*TMEM67*	•			
Joubert Syndrome, AHI1-Related	*AHI1*	•		O	O
Joubert Syndrome, ARL13B-Related	*ARL13B*	•		O	
Joubert Syndrome, B9D1-Related	*B9D1*	•		O	
Joubert Syndrome, B9D2-Related	*B9D2*	•		O	
Joubert Syndrome, C2CD3-Related / Orofaciodigital Syndrome 14	*C2CD3*	•		O	
Joubert Syndrome, CC2D2A-Related / COACH Syndrome	*CC2D2A*	•		O	O
Joubert Syndrome, CEP104-Related	*CEP104*	•		O	
Joubert Syndrome, CEP120-Related / Short-Rib Thoracic Dysplasia 13 with or without Polydactyly	*CEP120*	•		O	
Joubert Syndrome, CEP41-Related	*CEP41*	•		O	
Joubert Syndrome, CPLANE1-Related / Orofaciodigital Syndrome 6	*CPLANE1*	•		O	

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Joubert Syndrome, CSPP1-Related	*CSPP1*	•		O	
Joubert Syndrome, INPP5E-Related	*INPP5E*	•		O	
Junctional Epidermolysis Bullosa, COL17A1-Related	*COL17A1*	•			
Junctional Epidermolysis Bullosa, ITGA6-Related	*ITGA6*	•			
Junctional Epidermolysis Bullosa, ITGB4-Related	*ITGB4*	•			
Junctional Epidermolysis Bullosa, LAMA3-Related	*LAMA3*	•			
Junctional Epidermolysis Bullosa, LAMB3-Related	*LAMB3*	•			
Junctional Epidermolysis Bullosa, LAMC2-Related	*LAMC2*	•			
Juvenile Retinoschisis, X-Linked	*RS1*		•		O
Ketothiolase Deficiency	*ACAT1*	•			O
Krabbe Disease	*GALC*	•			
L1 Syndrome	*L1CAM*		•		O
Lamellar Ichthyosis, Type 1	*TGM1*	•			
Laron Syndrome	*GHR*	•			
Leber Congenital Amaurosis 2	*RPE65*	•			
Leber Congenital Amaurosis Type AIPL1	*AIPL1*	•			
Leber Congenital Amaurosis Type LRAT	*LRAT*	•			
Leber Congenital Amaurosis Type TULP1	*TULP1*	•			
Leber Congenital Amaurosis, IQCB1-Related / Senior-Loken Syndrome 5	*IQCB1*	•			
Leber Congenital Amaurosis, Type CEP290	*CEP290*	•			O
Leber Congenital Amaurosis, Type LCA5	*LCA5*	•			
Leber Congenital Amaurosis, Type RDH12	*RDH12*	•			
Leber Conghenital Amaurosis Type GUCY2D	*GUCY2D*	•			
Leigh Syndrome, French-Canadian Type	*LRPPRC*	•			
Lesch-Nyhan Syndrome	*HPRT1*		•		
Lethal Congenital Contracture Syndrome 1	*GLE1*	•			
Leukoencephalopathy with Vanishing White Matter	*EIF2B5*	•			
Leukoencephalopathy With Vanishing White Matter, EIF2B1-Related	*EIF2B1*	•			
Leukoencephalopathy With Vanishing White Matter, EIF2B2-Related	*EIF2B2*	•			
Leukoencephalopathy With Vanishing White Matter, EIF2B3-Related	*EIF2B3*	•			
Leukoencephalopathy With Vanishing White Matter, EIF2B4-Related	*EIF2B4*	•			
LIG4 Syndrome	*LIG4*	•			
Limb-Girdle Muscular Dystrophy Type 8	*TRIM32*	•			
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*	•			O
Lipoamide Dehydrogenase Deficiency (Dihydrolipoamide Dehydrogenase Deficiency)	*DLD*	•			O
Lipoid Adrenal Hyperplasia	*STAR*	•			
Lipoprotein Lipase Deficiency	*LPL*	•			
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*	•			
Lung Disease, Immunodeficiency, And Chromosome Breakage Syndrome (LICS)	*NSMCE3*	•			
Lysinuric Protein Intolerance	*SLC7A7*	•			
Malonyl-CoA Decarboxylase Deficiency	*MLYCD*	•			
Maple Syrup Urine Disease, Type 1A	*BCKDHA*	•		O	
Maple Syrup Urine Disease, Type 1B	*BCKDHB*	•		O	O
Maple Syrup Urine Disease, Type 2	*DBT*	•		O	
McKusick-Kaufman Syndrome	*MKKS*	•			
Meckel Syndrome 7 / Nephronophthisis 3	*NPHP3*	•			
Meckel-Gruber Syndrome, Type 1	*MKS1*	•			
MECP2-Related Conditions	*MECP2*		•		
MECR-Related Neurologic Disorder	*MECR*	•			
Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*	•		O	O
MEDNIK Syndrome	*AP1S1*	•			
Megalencephalic Leukoencephalopathy with Subcortical Cysts	*MLC1*	•			O
Menkes Syndrome	*ATP7A*		•		
Merosin-Deficient Muscular Dystrophy	*LAMA2*	•			

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Metabolic Encephalopathy and Arrhythmias, TANGO2-Related	*TANGO2*	•			
Metachromatic Leukodystrophy, ARSA-Related	*ARSA*	•			O
Metachromatic Leukodystrophy, PSAP-Related	*PSAP*	•			
Methylmalonic Acidemia And Homocystinuria Type Cblf	*LMBRD1*	•			
Methylmalonic Acidemia And Homocystinuria Type Cblx	*HCFC1*		•		
Methylmalonic Aciduria and Homocystinuria, Type cblC	*MMACHC*	•			O
Methylmalonic Aciduria and Homocystinuria, Type cblD	*MMADHC*	•			
Methylmalonic Aciduria, MCEE-Related	*MCEE*	•			
Methylmalonic Aciduria, MMAA-Related	*MMAA*	•			
Methylmalonic Aciduria, MMAB-Related	*MMAB*	•			
Methylmalonic Aciduria, Type mut(0)	*MUT*	•			O
Mevalonic Kinase Deficiency	*MVK*	•			O
Microcephalic Osteodysplastic Primordial Dwarfism Type II	*PCNT*	•			
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 10	*NDUFAF2*	•			
Mitochondrial Complex I Deficiency, Nuclear Type 17	*NDUFAF6*	•			
Mitochondrial Complex I Deficiency, Nuclear Type 19	*FOXRED1*	•			
Mitochondrial Complex I Deficiency, Nuclear Type 3	*NDUFS7*	•			
Mitochondrial Complex I Deficiency, Nuclear Type 4	*NDUFV1*	•			
Mitochondrial Complex IV Deficiency, Nuclear Type 2, SCO2-Related	*SCO2*	•			O
Mitochondrial Complex IV Deficiency, Nuclear Type 6	*COX15*	•			
Mitochondrial Dna Depletion Syndrome 2	*TK2*	•			
Mitochondrial Dna Depletion Syndrome 3	*DGUOK*	•			
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	*PUS1*	•			
Mitochondrial Trifunctional Protein Deficiency, HADHB-Related	*HADHB*	•			
Molybdenum Cofactor Deficiency Type B	*MOCS2*	•			
Molybdenum Cofactor Deficiency, Type A	*MOCS1*	•			
Mucolipidosis II/IIIA	*GNPTAB*	•			O
Mucolipidosis III gamma	*GNPTG*	•			
Mucolipidosis, Type IV	*MCOLN1*	•		O	O
Mucopolysaccharidosis, Type I (Hurler Syndrome)	*IDUA*	•			O
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*	•			
Muscular Dystrophy-Dystroglycanopathy	*RXYLT1*	•			
MUSK-Related Congenital Myasthenic Syndrome	*MUSK*	•			
Myoneurogastrointestinal Encephalopathy (MNGIE)	*TYMP*	•			
Myotonia Congenita	*CLCN1*	•			O
Myotubular Myopathy, X-Linked	*MTM1*		•		
N-acetylglutamate Synthase Deficiency	*NAGS*	•			
Nemaline Myopathy, NEB-Related	*NEB*	•			O
Nephrogenic Diabetes Insipidus, AVPR2-Related	*AVPR2*		•		
Nephronophthisis 1	*NPHP1*	•			
Neuronal Ceroid Lipofuscinosis, CLN5-Related	*CLN5*	•			
Neuronal Ceroid Lipofuscinosis, CLN6-Related	*CLN6*	•			
Neuronal Ceroid Lipofuscinosis, CLN8-Related	*CLN8*	•			

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Neuronal Ceroid Lipofuscinosis, MFSD8-Related	*MFSD8*	•			
Neuronal Ceroid Lipofuscinosis, PPT1-Related	*PPT1*	•			
Neuronal Ceroid Lipofuscinosis, TPP1-Related	*TPP1*	•			
Ngly1-Congenital Disorder Of Glycosylation	*NGLY1*	•			
Niemann-Pick Disease, Type C1/D	*NPC1*	•		O	
Niemann-Pick Disease, Type C2	*NPC2*	•		O	
Niemann-Pick Disease, Types A/B	*SMPD1*	•		O	O
Nijmegen Breakage Syndrome	*NBN*	•			
Non-Syndromic Hearing Loss, GJB2-Related	*GJB2*	•			O
Nonsyndromic Hearing Loss, MYO15A-Related	*MYO15A*	•			
Nonsyndromic Hearing Loss, OTOA-Related	*OTOA*	•			
Nonsyndromic Hearing Loss, OTOF-Related	*OTOF*	•			
Nonsyndromic Hearing Loss, PJVK-Related	*PJVK*	•			
Nonsyndromic Hearing Loss, SYNE4-Related	*SYNE4*	•			
Nonsyndromic Hearing Loss, TMC1-Related	*TMC1*	•			
Nonsyndromic Hearing Loss, TMPRSS3-Related	*TMPRSS3*	•			
Nonsyndromic Intellectual Disability	*CC2D1A*	•			
Normophosphatemic Tumoral Calcinosis	*SAMD9*	•			
Oculocutaneous Albinism Type III	*TYRP1*	•			
Oculocutaneous Albinism Type IV	*SLC45A2*	•			
Oculocutaneous Albinism, OCA2-Related	*OCA2*	•			O
Oculocutaneous Albinism, Type 1A and 1B	*TYR*	•			O
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	*WNT10A*	•			
Omenn Syndrome, RAG2-Related	*RAG2*	•			
Opitz G/BBB Syndrome, X-Linked	*MID1*		•		O
Ornithine Transcarbamylase Deficiency	*OTC*		•		O
Osteogenesis Imperfecta Type VII	*CRTAP*	•			
Osteogenesis Imperfecta Type VIII	*P3H1*	•			
Osteogenesis Imperfecta Type XI	*FKBP10*	•			
Osteogenesis Imperfecta Type XIII	*BMP1*	•			
Osteopetrosis, Infantile Malignant, TCIRG1-Related	*TCIRG1*	•			
Osteopetrosis, OSTM1-Related	*OSTM1*	•			
Pantothenate Kinase-Associated Neurodegeneration	*PANK2*	•			
Papillon Lefèvre Syndrome	*CTSC*	•			
Parkinson Disease 15	*FBXO7*	•			
Pendred Syndrome	*SLC26A4*	•			O
Perlman Syndrome	*DIS3L2*	•			
Pgm3-Congenital Disorder Of Glycosylation	*PGM3*	•			
Phenylketonuria	*PAH*	•			O
Pign-Congenital Disorder Of Glycosylation	*PIGN*	•			
Pituitary Hormone Deficiency, Combined 3	*LHX3*	•			
PLP1 Disorders	*PLP1*		•		O
POLG-Related Disorders	*POLG*	•			O
Polycystic Kidney Disease, Autosomal Recessive	*PKHD1*	•			O
Pontocerebellar Hypoplasia, EXOSC3-Related	*EXOSC3*	•			
Pontocerebellar Hypoplasia, RARS2-Related	*RARS2*	•			O
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, CCDC103-Related	*CCDC103*	•			
Primary Ciliary Dyskinesia, CCDC39-Related	*CCDC39*	•			
Primary Ciliary Dyskinesia, DNAH11-Related	*DNAH11*	•			
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*	•			O
Primary Hyperoxaluria, Type 2	*GRHPR*	•			
Primary Hyperoxaluria, Type 3	*HOGA1*	•			

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Primary Microcephaly 1, Autosomal Recessive	*MCPH1*	•			O
Progressive Early-Onset Encepahlopathy With Brain Atrophy And Thin Corpus Callosum	*TBCD*	•			
Progressive Familial Intrahepatic Cholestasis, ABCB4-Related	*ABCB4*	•			
Progressive Familial Intrahepatic Cholestasis, Type 1 (PFIC1)	*ATP8B1*	•			
Progressive Familial Intrahepatic Cholestasis, Type 2	*ABCB11*	•			
Progressive Familial Intrahepatic Cholestasis, Type 4 (PFIC4)	*TJP2*	•			
Progressive Pseudorheumatoid Dysplasia	*CCN6*	•			
Prolidase Deficiency	*PEPD*	•			
Propionic Acidemia, PCCA-Related	*PCCA*	•			
Propionic Acidemia, PCCB-Related	*PCCB*	•			
Pseudocholinesterase Deficiency	*BCHE*	•			
Pterin-4 Alpha-Carbinolamine Dehydratase (PCD) Deficiency	*PCBD1*	•			
Pycnodysostosis	*CTSK*	•			
Pyridoxal 5'-Phosphate-Dependent Epilepsy	*PNPO*	•			
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 2	*RP2*		•		
Retinitis Pigmentosa 25	*EYS*	•			
Retinitis Pigmentosa 26	*CERKL*	•			
Retinitis Pigmentosa 28	*FAM161A*	•			
Retinitis Pigmentosa 36	*PRCD*	•			
Retinitis Pigmentosa 59	*DHDDS*	•			O
Retinitis Pigmentosa 62	*MAK*	•			
Retinitis Pigmentosa, X-Linked, RPGR-Related	*RPGR*		•		O
Rhizomelic Chondrodysplasia Punctata, Type 1	*PEX7*	•			
Rhizomelic Chondrodysplasia Punctata, Type 2	*GNPAT*	•			
Rhizomelic Chondrodysplasia Punctata, Type 3	*AGPS*	•			
RLBP1-Related Retinopathy	*RLBP1*	•			
Roberts Syndrome	*ESCO2*	•			
RYR1-Related Conditions	*RYR1*	•			
Salla Disease	*SLC17A5*	•			
Sandhoff Disease	*HEXB*	•			
Schimke Immunoosseous Dysplasia	*SMARCAL1*	•			
Schindler Disease	*NAGA*	•			O
Segawa Syndrome, TH-Related	*TH*	•			
Senior-Loken Syndrome 4 / Nephronophthisis 4	*NPHP4*	•			
Sepiapterin Reductase Deficiency	*SPR*	•			
Severe Combined Immunodeficiency (SCID), CD3D-Related	*CD3D*	•			
Severe Combined Immunodeficiency (SCID), CD3E-Related	*CD3E*	•			
Severe Combined Immunodeficiency (SCID), FOXN1-Related	*FOXN1*	•			
Severe Combined Immunodeficiency (SCID), IKBKB-Related	*IKBKB*	•			
Severe Combined Immunodeficiency (SCID), IL7R-Related	*IL7R*	•			
Severe Combined Immunodeficiency (SCID), JAK3-Related	*JAK3*	•			
Severe Combined Immunodeficiency (SCID), PTPRC-Related	*PTPRC*	•			
Severe Combined Immunodeficiency, ADA-Related	*ADA*	•			
Severe Combined Immunodeficiency, RAG1-Related	*RAG1*	•			
Severe Combined Immunodeficiency, Type Athabaskan	*DCLRE1C*	•			
Severe Combined Immunodeficiency, X-Linked	*IL2RG*		•		
Short-Rib Thoracic Dysplasia 3 with or without Polydactyly	*DYNC2H1*	•			O
Shwachman-Diamond Syndrome, SBDS-Related	*SBDS*	•			
Sialidosis	*NEU1*	•			
Sjögren-Larsson Syndrome	*ALDH3A2*	•			
Smith-Lemli-Opitz Syndrome	*DHCR7*	•		O	O
Spastic Paraplegia, Type 15	*ZFYVE26*	•			

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly (SPATCCM)	*SLC1A4*	•			
SPG11-Related Conditions	*SPG11*	•			
Spinal Muscular Atrophy	*SMN1*	•		O	O
Spinal Muscular Atrophy With Respiratory Distress Type 1	*IGHMBP2*	•			
Spinocerebellar Ataxia, Autosomal Recessive 10	*ANO10*	•			O
Spinocerebellar Ataxia, Autosomal Recessive 12	*WWOX*	•			
Spondylocostal Dysostosis 1	*DLL3*	•			
Spondylothoracic Dysostosis, MESP2-Related	*MESP2*	•			
Steel Syndrome	*COL27A1*	•			
Steroid-Resistant Nephrotic Syndrome	*NPHS2*	•			
Stuve-Wiedemann Syndrome	*LIFR*	•			
SURF1-Related Conditions	*SURF1*	•			
Surfactant Dysfunction, ABCA3-Related	*ABCA3*	•			O
Tay-Sachs Disease	*HEXA*	•		O	O
TBCE-Related Conditions	*TBCE*	•			
Thiamine-Responsive Megaloblastic Anemia Syndrome	*SLC19A2*	•			
Thyroid Dyshormonogenesis 1	*SLC5A5*	•			
Thyroid Dyshormonogenesis 2A	*TPO*	•			
Thyroid Dyshormonogenesis 3	*TG*	•			
Thyroid Dyshormonogenesis 6	*DUOX2*	•			
Transcobalamin II Deficiency	*TCN2*	•			
Trichohepatoenteric Syndrome, SKIV2L-Related	*SKIC2*	•			
Trichohepatoenteric Syndrome, TTC37-Related	*TTC37*	•			
Trichothiodystrophy 1 / Xeroderma Pigmentosum, Group D	*ERCC2*	•			O
Trimethylaminuria	*FMO3*	•			O
Triple A Syndrome	*AAAS*	•			
TSHR-Related Conditions	*TSHR*	•			
Tyrosinemia Type III	*HPD*	•			
Tyrosinemia, Type 1	*FAH*	•			O
Tyrosinemia, Type 2	*TAT*	•			
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 1J / Deafness, Autosomal Recessive, 48	*CIB2*	•			
Usher Syndrome, Type 2A	*USH2A*	•			O
Usher Syndrome, Type 2C	*ADGRV1*	•			
Usher Syndrome, Type 3	*CLRN1*	•		O	O
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	*ACADVL*	•			O
Viatmin D-Dependent Rickets Type 2A	*VDR*	•			
Vici Syndrome	*EPG5*	•			
Vitamin D Dependent Rickets, Type 1A	*CYP27B1*	•			O
Vldlr-Associated Cerebellar Hypoplasia	*VLDLR*	•			
Walker-Warburg Syndrome, FKTN-Related	*FKTN*	•			O
Walker-Warburg Syndrome, ISPD-Related	*ISPD*	•			
Walker-Warburg Syndrome, LARGE1-Related	*LARGE1*	•			
Walker-Warburg Syndrome, POMT1-Related	*POMT1*	•			
Walker-Warburg Syndrome, POMT2-Related	*POMT2*	•			
Warsaw Breakage Syndrome	*DDX11*	•			
Werner Syndrome	*WRN*	•			
Wilson Disease	*ATP7B*	•			O
Wiskott-Aldrich Syndrome	*WAS*		•		
Wolcott-Rallison Syndrome	*EIF2AK3*	•			
Wolman Disease	*LIPA*	•			
Woodhouse-Sakati Syndrome	*DCAF17*	•			
Xeroderma Pigmentosum Variant Type	*POLH*	•			
Xeroderma Pigmentosum, Group A	*XPA*	•			
Xeroderma Pigmentosum, Group C	*XPC*	•			O
X-Linked Chondrodysplasia Punctata 1	*ARSL*		•		
X-Linked Lissencephaly with Abnormal Genitalia	*ARX*		•		O

Condition	Gene	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS	
				ACOG†	ACMG
Zellweger Spectrum Disorder, PEX13-Related	*PEX13*	•			
Zellweger Spectrum Disorder, PEX16-Related	*PEX16*	•			
Zellweger Spectrum Disorder, PEX5-Related	*PEX5*	•			
Zellweger Spectrum Disorders, PEX10-Related	*PEX10*	•			
Zellweger Spectrum Disorders, PEX12-Related	*PEX12*	•			
Zellweger Spectrum Disorders, PEX1-Related	*PEX1*	•			
Zellweger Spectrum Disorders, PEX26-Related	*PEX26*	•			
Zellweger Spectrum Disorders, PEX2-Related	*PEX2*	•			
Zellweger Spectrum Disorders, PEX6-Related	*PEX6*	•			

*Low penetrance genes with variable presentation.

†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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