SYNLAB

XX preconGEN
Understanding the risk of your next generation

LIST OF DISEASES ANALYZED BY preconGEN

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CYP11B1)

21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CYP21A2)*

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS)

ABCC8-Related Hyperinsulinism (ABCC8)

Adenosine Deaminase Deficiency (ADA)

Adrenoleukodystrophy: X-Linked (ABCD1) X-linked

Alpha Thalassemia (HBA1/HBA2)* ACOG ACMG

Alpha-Mannosidosis (MAN2B1)

Alpha-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2D) (SGCA)

Alport Syndrome, X-Linked (COL4A5) X-linked

Alstrom Syndrome (ALMS1)

AMT-Related Glycine Encephalopathy (AMT)

Andermann Syndrome (SLC12A6)

Argininemia (ARG1)

Argininosuccinic Aciduria (ASL)

ARSACS (SACS)

Aspartylglycosaminuria (AGA)

Ataxia with Vitamin E Deficiency (TTPA)

Ataxia-Telangiectasia (ATM)

ATP7A-Related Disorders (ATP7A) X-linked

Autosomal Recessive Osteopetrosis, Type 1 (TCIRG1)

Bardet-Biedl Syndrome, BBS1-Related (BBS1)

Bardet-Biedl Syndrome, BBS10-Related (BBS10)

Bardet-Biedl Syndrome, BBS12-Related (BBS12)

Bardet-Biedl Syndrome, BBS2-Related (BBS2)

Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) (SGCB)

Biotinidase Deficiency (BTD)

Bloom Syndrome (BLM) ACMG

Calpainopathy (CAPN3)

Canavan Disease (ASPA) ACOG ACMG

Carbamoylphosphate Synthetase I Deficiency (CPS1)

Carnitine Palmitoyltransferase IA Deficiency (CPT1A)

Carnitine Palmitoyltransferase II Deficiency (CPT2)

Cartilage-Hair Hypoplasia (RMRP)

Cerebrotendinous Xanthomatosis (CYP27A1)

Citrullinemia, Type 1 (ASS1)

CLN3-Related Neuronal Ceroid Lipofuscinosis (CLN3)

CLN5-Related Neuronal Ceroid Lipofuscinosis (CLN5)

CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (CLN6)

Cohen Syndrome (VPS13B)

COL4A3-Related Alport Syndrome (COL4A3)

COL4A4-Related Alport Syndrome (COL4A4)

Congenital Disorder of Glycosylation, Type Ia (PMM2)

Congenital Disorder of Glycosylation, Type Ib (MPI)

Congenital Disorder of Glycosylation, Type Ic (ALG6)

Congenital Finnish Nephrosis (NPHS1)

Costeff Optic Atrophy Syndrome (OPA3)

Cystic Fibrosis (CFTR) ACOG ACMG

Cystinosis (CTNS)

D-Bifunctional Protein Deficiency (HSD17B4)

Delta-Sarcoglycanopathy (SGCD)

Dysferlinopathy (DYSF)

Dystrophinopathies (including Duchenne / Becker Muscular Dystrophy) (DMD) X-linked

ERCC6-Related Disorders (ERCC6)

ERCC8-Related Disorders (ERCC8)

EVC-Related Ellis-Van Creveld Syndrome (EVC)

EVC2-Related Ellis-Van Creveld Syndrome (EVC2)

Fabry Disease (GLA) X-linked

Familial Dysautonomia (IKBKAP) ACOG ACMG

Familial Mediterranean Fever (MEFV)

Fanconi Anemia Complementation, Group A (FANCA)

Fanconi Anemia, Type C (FANCC) ACMG

FKRP-Related Disorders (FKRP)

FKTN-Related Disorders (including Walker-Warburg Syndrome) (FKTN)

Fragile X Syndrome (FMR1)* X-linked

Galactokinase Deficiency (GALK1)

Galactosemia (GALT)

Gamma-Sarcoglycanopathy (SGCG)

Gaucher Disease (GBA)* ACMG

GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) (GJB2)

GLB1-Related Disorders (GLB1)

GLDC-Related Glycine Encephalopathy (GLDC)

Glutaric Acidemia, Type 1 (GCDH)

Glycogen Storage Disease, Type Ia (G6PC)

Glycogen Storage Disease, Type Ib (SLC37A4)

Glycogen Storage Disease, Type III (AGL)

GNPTAB-Related Disorders (GNPTAB)

GRACILE Syndrome (BCS1L)

HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (HADHA)

Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (HBB) ACOG

Hereditary Fructose Intolerance (ALDOB)

Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related (LAMA3)

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related (LAMB3)

Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related (LAMC2)

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Hexosaminidase A Deficiency (including Tay-Sachs Disease) (HEXA) ACOG ACMG

HMG-CoA Lyase Deficiency (HMGCL)

Holocarboxylase Synthetase Deficiency (HLCS)

Homocystinuria caused by Cystathionine Beta-Synthase Deficiency (CBS)

Hydrolethalus Syndrome (HYLS1)

Hypophosphatasia, Autosomal Recessive (ALPL)

Inclusion Body Myopathy 2 (GNE)

Isovaleric Acidemia (IVD)

Joubert Syndrome 2 (TMEM216)

KCNJ11-Related Familial Hyperinsulinism (KCNJ11)

Krabbe Disease (GALC)

LAMA2-Related Muscular Dystrophy (LAMA2)

Leigh Syndrome, French-Canadian Type (LRPPRC)

Lipoamide Dehydrogenase Deficiency (DLD)

Lipoid Congenital Adrenal Hyperplasia (STAR)

Lysosomal Acid Lipase Deficiency (LIPA)

Maple Syrup Urine Disease, Type Ia (BCKDHA)

Maple Syrup Urine Disease, Type IB (BCKDHB)

Maple Syrup Urine Disease, Type II (DBT)

Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)

Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)

Metachromatic Leukodystrophy (ARSA)

Methylmalonic Acidemia, cblA Type (MMAA)

Methylmalonic Acidemia, cblB Type (MMAB)

Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)

MKS1-Related Disorders (MKS1)

Mucolipidosis III Gamma (GNPTG)

Mucolipidosis IV (MCOLN1) ACMG

Mucopolysaccharidosis, Type I (including Hurler Syndrome) (IDUA)

Mucopolysaccharidosis, Type II (IDS) X-linked

Mucopolysaccharidosis, Type IIIA (SGSH)

Mucopolysaccharidosis, Type IIIB (NAGLU)

Mucopolysaccharidosis, Type IIIC (HGSNAT)

Muscle-Eye-Brain Disease (POMGNT1)

MUT-Related Methylmalonic Acidemia (MUT)

MYO7A-Related Disorders (MYO7A)

NEB-Related Nemaline Myopathy (NEB)

Niemann-Pick Disease, Type C (NPC1)

Niemann-Pick Disease, Type C2 (NPC2)

Niemann-Pick Disease, SMPD1-Associated (SMPD1) ACMG

Nijmegen Breakage Syndrome (NBN)

Northen Epilepsy (CLN8)

Ornithine Transcarbamylase Deficiency (OTC) X-linked

PCCA-Related Propionic Acidemia (PCCA)

PCCB-Related Propionic Acidemia (PCCB)

PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (PCDH15)

Pendred Syndrome (SLC26A4)

Peroxisome Biogenesis Disorder, Type 3 (PEX12)

Peroxisome Biogenesis Disorder, Type 4 (PEX6)

Peroxisome Biogenesis Disorder, Type 5 (PEX2)

Peroxisome Biogenesis Disorder, Type 6 (PEX10)

PEX1-Related Zellweger Syndrome Spectrum (PEX1)

Phenylalanine Hydroxylase Deficiency (PAH)

PKHD1-Related Autosomal Recessive Polycystic Kidney Disease (PKHD1)

Polyglandular Autoimmune Syndrome, Type 1 (AIRE)

Pompe Disease (GAA)

PPT1-Related Neuronal Ceroid Lipofuscinosis (PPT1)

Primary Carnitine Deficiency (SLC22A5)

Primary Hyperoxaluria, Type 1 (AGXT)

Primary Hyperoxaluria, Type 2 (GRHPR)

Primary Hyperoxaluria, Type 3 (HOGA1)

PROP1-Related Combined Pituitary Hormone Deficiency (PROP1)

Pycnodysostosis (CTSK)

Pyruvate Carboxylase Deficiency (PC)

Rhizomelic Chondrodysplasia Punctata, Type 1 (PEX7)

RTEL1-Related Disorders (RTEL1)

Salla Disease (SLC17A5)

Sandhoff Disease (HEXB)

Segawa Syndrome (TH)

Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)

Sjogren-Larsson Syndrome (ALDH3A2)

Smith-Lemli-Opitz Syndrome (DHCR7)

Spastic Paraplegia, Type 15 (ZFYVE26)

Spinal Muscular Atrophy (SMN1)* ACMG

Spondylothoracic Dysostosis (MESP2)

Steroid-Resistant Nephrotic Syndrome (NPHS2)

Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2)

TGM1-Related Autosomal Recessive Congenital Ichthyosis (TGM1)

TPP1-Related Neuronal Ceroid Lipofuscinosis (TPP1)

Tyrosinemia, Type I (FAH)

Tyrosinemia, Type II (TAT)

USH1C-Related Disorders (USH1C)

USH2A-Related Disorders (USH2A)

Usher Syndrome, Type 3 (CLRN1)

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)

Wilson Disease (ATP7B)

X-Linked Congenital Adrenal Hypoplasia (NR0B1) X-linked

X-Linked Juvenile Retinoschisis (RS1) X-linked

X-Linked Myotubular Myopathy (MTM1) X-linked

X-Linked Severe Combined Immunodeficiency (IL2RG) X-linked

Xeroderma Pigmentosum, Group A (XPA)

Xeroderma Pigmentosum, Group C (XPC)

ACOG Indicates testing recommended by ACOG (American College of Obstetricians and Gynecologists)
ACMG Indicates testing recommended by ACMG (American College of Medical Genetics and Genomics)
X-linked Indicates X-linked disorders
* Use of differentiating specialized technology (technically challenging genes)
Diseases included on the donor panel

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