Foresight®
Carrier Screen

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# Reproductive Carrier Screening Panel

The Foresight Carrier Screen focuses on serious, clinically-actionable, and prevalent conditions, as well as those recommended by ACOG and ACMG guidelines, to ensure you are providing meaningful information to your patients. This panel is designed to assess an individual or couple's risk for passing serious inherited conditions to their children.

| Disease | Gene | X-Linked Disorder |
|---|---|---|
| 3-Methylcrotonyl-CoA Carboxylase Deficiency, *MCC2*-Related | *MCCC2* | |
| 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency | *PTS* | |
| Abetalipoproteinemia | *MTTP* | |
| Adenosine Deaminase Deficiency | *ADA* | |
| Aicardi-Goutières Syndrome | *RNASEH2B* | |
| Alkaptonuria | *HGD* | |
| Alpha Thalassemia, *HBA1/HBA2*-Related | *HBA1/HBA2* | |
| Alpha-Mannosidosis | *MAN2B1* | |
| Alpha-Sarcoglycanopathy | *SGCA* | |
| Alport Syndrome, *COL4A3*-Related | *COL4A3* | |
| Alport Syndrome, *COL4A4*-Related | *COL4A4* | |
| Alstrom Syndrome | *ALMS1* | |
| Andermann Syndrome | *SLC12A6* | |
| Argininemia | *ARG1* | |
| Argininosuccinic Aciduria | *ASL* | |
| Arthrogryposis, Mental Retardation, and Seizures* | *SLC35A3* | |
| *ARX*-Related Disorders | *ARX* | ✓ |
| Asparagine Synthetase Deficiency | *ASNS* | |
| Aspartylglycosaminuria | *AGA* | |
| Ataxia with Vitamin E Deficiency | *TTPA* | |
| Ataxia-Telangiectasia | *ATM* | |
| *ATP7A*-Related Disorders | *ATP7A* | ✓ |
| Atransferrinemia | *TF* | |
| Autoimmune Polyglandular Syndrome Type 1 | *AIRE* | |
| Autosomal Recessive Osteopetrosis Type 1 | *TCIRG1* | |
| Autosomal Recessive Polycystic Kidney Disease, *PKHD1*-related | *PKHD1* | |
| Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay | *SACS* | |
| Bardet-Biedl Syndrome, *BBS1*-Related | *BBS1* | |
| Bardet-Biedl Syndrome, *BBS2*-Related | *BBS2* | |
| Bardet-Biedl Syndrome, *BBS10*-Related | *BBS10* | |
| Bardet-Biedl Syndrome, *BBS12*-Related | *BBS12* | |
| *BCS1L*-Related Disorders | *BCS1L* | |
| Beta Globin-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) | *HBB* | |
| Beta-Ketothiolase Deficiency | *ACAT1* | |
| Beta-Sarcoglycanopathy | *SGCB* | |
| Biotin-Thiamine–Responsive Basal Ganglia Disease | *SLC19A3* | |
| Biotinidase Deficiency | *BTD* | |
| Bloom Syndrome | *BLM* | |

| Disease | Gene | X-Linked Disorder |
|---|---|---|
| Calpainopathy | *CAPN3* | |
| Canavan Disease | *ASPA* | |
| Carbamoylphosphate Synthetase I Deficiency | *CPS1* | |
| Carnitine Palmitoyltransferase IA Deficiency | *CPT1A* | |
| Carnitine Palmitoyltransferase II Deficiency | *CPT2* | |
| Cartilage-Hair Hypoplasia | *RMRP* | |
| *CC2D2A*-Related Disorders | *CC2D2A* | |
| *CEP290*-Related Disorders | *CEP290* | |
| Cerebrotendinous Xanthomatosis | *CYP27A1* | |
| Choreoacanthocytosis | *VPS13A* | |
| Choroideremia | *CHM* | ✓ |
| Chronic Granulomatous Disease, Cytochrome B-Negative | *CYBA* | |
| Citrullinemia Type 1 | *ASS1* | |
| Classical-Like Ehlers-Danlos syndrome, *TNXB*-Related | *TNXB* | |
| *CLN3*-Related Disorders | *CLN3* | |
| *CLN5*-Related Neuronal Ceroid Lipofuscinosis | *CLN5* | |
| *CLN8*-Related Neuronal Ceroid Lipofuscinosis | *CLN8* | |
| *CNGB3*-Related Achromatopsia | *CNGB3* | |
| Cohen Syndrome | *VPS13B* | |
| Combined Pituitary Hormone Deficiency, *PROP1*-Related | *PROP1* | |
| Congenital Adrenal Hyperplasia, *CYP11A1*-Related | *CYP11A1* | |
| Congenital Adrenal Hyperplasia, CYP11B1-Related | *CYP11B1* | |
| Congenital Adrenal Hyperplasia, *CYP21A2*-Related* | *CYP21A2* | |
| Congenital Amegakaryocytic Thrombocytopenia | *MPL* | |
| Congenital Disorder of Glycosylation, PMM2-Related | *PMM2* | |
| Congenital Disorder of Glycosylation Type 1c | *ALG6* | |
| Congenital Disorder of Glycosylation, *MPI*-Related | *MPI* | |
| Congenital Hydrocephalus, *CCDC88C*-Related | *CCDC88C* | |
| Congenital Insensitivity to Pain with Anhidrosis | *NTRK1* | |
| Congenital Myasthenic Syndrome, *CHRNE*-Related | *CHRNE* | |
| Corticosterone Methyloxidase Deficiency* | *CYP11B2* | |
| Costeff Optic Atrophy Syndrome | *OPA3* | |
| Creatine Transporter Deficiency | *SLC6A8* | ✓ |
| Cystic Fibrosis | *CFTR* | |
| Cystinosis | *CTNS* | |
| D-Bifunctional Protein Deficiency | *HSD17B4* | |
| Delta-Sarcoglycanopathy | *SGCD* | |
| Dihydrolipoamide Dehydrogenase Deficiency | *DLD* | |

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| Disease | Gene | X-Linked Disorder |
|---|---|---|
| Dihydropyrimidine Dehydrogenase Deficiency | *DPYD* | |
| *DNAH5*-Related Primary Ciliary Dyskinesia | *DNAH5* | |
| *DNAI1*-Related Primary Ciliary Dyskinesia | *DNAI1* | |
| *DNAI2*-Related Primary Ciliary Dyskinesia | *DNAI2* | |
| Donnai–Barrow Syndrome | *LRP2* | |
| DYNC2H1-Related Disorders | *DYNC2H1* | |
| Dysferlinopathy | *DYSF* | |
| Dystrophic Epidermolysis Bullosa | *COL7A1* | |
| Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) | *DMD* | ✓ |
| Ehlers-Danlos Syndrome Type VIIC | *ADAMTS2* | |
| ERCC2-Related Disorders | *ERCC2* | |
| *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* | ✓ |
| Factor XI Deficiency | *F11* | |
| Familial Dysautonomia | *ELP1* | |
| Familial Hemophagocytic Lymphohistiocytosis, *PRF1*-Related | *PRF1* | |
| Familial Hyperinsulinism, *ABCC8*-Related | *ABCC8* | |
| Familial Hyperinsulinism, *KCNJ11*-Related | *KCNJ11* | |
| Familial Mediterranean Fever | *MEFV* | |
| Fanconi Anemia Complementation Group A | *FANCA* | |
| Fanconi Anemia, *FANCC*-Related | *FANCC* | |
| *FKRP*-Related Disorders | *FKRP* | |
| *FKTN*-Related Disorders | *FKTN* | |
| Fragile X Syndrome* | *FMR1* | ✓ |
| Fragile XE Syndrome | *AFF2* | ✓ |
| Fraser Syndrome, GRIP1-Related | *GRIP1* | |
| Free Sialic Acid Storage Disorders | *SLC17A5* | |
| Galactokinase Deficiency | *GALK1* | |
| Galactosemia | *GALT* | |
| Gamma-Sarcoglycanopathy | *SGCG* | |
| Gaucher Disease* | *GBA* | |
| *GBE1*-Related Disorders | *GBE1* | |
| *GJB2*-Related *DFNB1* Nonsyndromic Hearing Loss and Deafness | *GJB2* | |
| *GLB1*-Related Disorders | *GLB1* | |
| Glucose-6-Phosphate Dehydrogenase Deficiency* | *G6PD* | ✓ |
| Glutaric Acidemia, *GCDH*-Related | *GCDH* | |
| Glycine Encephalopathy, *AMT*-Related | *AMT* | |
| Glycine Encephalopathy, *GLDC*-Related | *GLDC* | |
| Glycogen Storage Disease Type Ia | *G6PC1* | |
| Glycogen Storage Disease Type Ib | *SLC37A4* | |
| Glycogen Storage Disease Type III | *AGL* | |
| Glycogen Storage Disease Type V | *PYGM* | |
| Glycogen Storage Disease Type VII | *PFKM* | |
| *GNE* Myopathy | *GNE* | |
| *GNPTAB*-Related Disorders | *GNPTAB* | |
| *HADHA*-Related Disorders | *HADHA* | |
| Hemophilia B | *F9* | ✓ |

| Disease | Gene | X-Linked Disorder |
|---|---|---|
| Hereditary Fructose Intolerance | *ALDOB* | |
| Hermansky-Pudlak Syndrome, HPS1-Related | *HPS1* | |
| Hermansky-Pudlak Syndrome Type 3 | *HPS3* | |
| Hexosaminidase A Deficiency (including Tay-Sachs disease) | *HEXA* | |
| *HMG-CoA* Lyase Deficiency | *HMGCL* | |
| Holocarboxylase Synthetase Deficiency | *HLCS* | |
| Homocystinuria Caused By *MTHFR* Deficiency# | *MTHFR* | |
| Homocystinuria, *CBS*-Related | *CBS* | |
| Hydrolethalus Syndrome | *HYLS1* | |
| Hypophosphatasia | *ALPL* | |
| Isovaleric Acidemia | *IVD* | |
| Joubert Syndrome 2 | *TMEM216* | |
| Joubert Syndrome, AHI1-Related | *AHI1* | |
| Junctional Epidermolysis Bullosa, *LAMA3*-Related | *LAMA3* | |
| Junctional Epidermolysis Bullosa, *LAMB3*-Related | *LAMB3* | |
| Junctional Epidermolysis Bullosa, *LAMC2*-Related | *LAMC2* | |
| Krabbe Disease | *GALC* | |
| L1 Syndrome | *L1CAM* | ✓ |
| Leigh Syndrome, French-Canadian Type | *LRPPRC* | |
| Lipoid Congenital Adrenal Hyperplasia | *STAR* | |
| *LOXHD1*-Related *DFNB77* Hearing Loss and Deafness | *LOXHD1* | |
| 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* | |
| Methlymalonic Acidemia, *MMUT*-Related | *MMUT* | |
| Methylmalonic Acidemia and Homocystinuria, cblC Type | *MMACHC* | |
| Mevalonate Kinase Deficiency | *MVK* | |
| Mitochondrial Complex IV Deficiency, SCO2-Related | *SCO2* | |
| Mitochondrial Neurogastrointestinal Encephalopathy Disease | *TYMP* | |
| *MKS1*-Related Disorders | *MKS1* | |
| Mucolipidosis III Gamma | *GNPTG* | |
| Mucolipidosis IV | *MCOLN1* | |
| Mucopolysaccharidosis Type I | *IDUA* | |
| Mucopolysaccharidosis Type II | *IDS* | ✓ |
| Mucopolysaccharidosis Type IIIA | *SGSH* | |
| Mucopolysaccharidosis Type IIIB | *NAGLU* | |
| Mucopolysaccharidosis Type IIIC | *HGSNAT* | |
| Multiple Sulfatase Deficiency | *SUMF1* | |
| Muscular Dystrophy, *LAMA2*-Related | *LAMA2* | |
| *MYO7A*-Related Disorders | *MYO7A* | |
| Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 | *PUS1* | |
| Myotonia Congenita | *CLCN1* | |
| *NAGA*-Related Disorders | *NAGA* | |
| *NDUFAF5*-Related Mitochondrial Complex 1 Deficiency* | *NDUFAF5* | |

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| Disease | Gene | X-Linked Disorder |
|---|---|---|
| *NDUFS4*-Related Mitochondrial Complex 1 Deficiency | *NDUFS4* | |
| *NDUFS6*-Related Mitochondrial Complex 1 Deficiency | *NDUFS6* | |
| *NEB*-Related Nemaline Myopathy | *NEB* | |
| Nephrotic Syndrome, *NPHS1*-Related | *NPHS1* | |
| Nephrotic Syndrome, *NPHS2*-Related | *NPHS2* | |
| Neuronal Ceroid Lipofuscinosis, *CLN6*-Related | *CLN6* | |
| Niemann-Pick Disease Type C1 | *NPC1* | |
| Niemann-Pick Disease Type C2 | *NPC2* | |
| Niemann-Pick Disease, *SMPD1*-Related | *SMPD1* | |
| Nijmegen Breakage Syndrome | *NBN* | |
| Normophosphatemic Familial Tumoral Calcinosis | *SAMD9* | |
| *NR2E3*-Related Disorders | *NR2E3* | |
| Oculocutaneous Albinism, *OCA2*-Related | *OCA2* | |
| Oculocutaneous Albinism, *TYR*-Related | *TYR* | |
| Opitz G/BBB Syndrome, *MID1*-Related | *MID1* | ✓ |
| Ornithine Aminotransferase Deficiency | *OAT* | |
| Ornithine Transcarbamylase Deficiency | *OTC* | ✓ |
| *PCCA*-Related Propionic Acidemia | *PCCA* | |
| *PCCB*-Related Propionic Acidemia | *PCCB* | |
| *PCDH15*-Related Disorders | *PCDH15* | |
| Pendred Syndrome | *SLC26A4* | |
| Peroxisome Biogenesis Disorder Type 1 | *PEX1* | |
| Peroxisome Biogenesis Disorder Type 3 | *PEX12* | |
| Peroxisome Biogenesis Disorder Type 4 | *PEX6* | |
| Peroxisome Biogenesis Disorder Type 5 | *PEX2* | |
| Peroxisome Biogenesis Disorder Type 6 | *PEX10* | |
| Phenylalanine Hydroxylase Deficiency | *PAH* | |
| Phosphoglycerate Dehydrogenase Deficiency | *PHGDH* | |
| *PHYH*-Related Refsum Disease | *PHYH* | |
| *PLP1*-Related Disorders | *PLP1* | ✓ |
| *POLG*-Related Disorders | *POLG* | |
| *POMGNT*-Related Disorders | *POMGNT1* | |
| Pompe Disease | *GAA* | |
| Pontocerebellar Hypoplasia Type 1A | *VRK1* | |
| Pontocerebellar Hypoplasia Type 2D | *SEPSECS* | |
| Pontocerebellar Hypoplasia Type 2E | *VPS53* | |
| Pontocerebellar Hypoplasia Type 6* | *RARS2* | |
| Postnatal Progressive Microcephaly with Seizures and Brain Atrophy* | *MED17* | |
| *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* | |
| Primary Microcephaly, *MCPH1*-Related | *MCPH1* | |
| Primary Trimethylaminuria | *FMO3* | |
| Pycnodysostosis | *CTSK* | |
| Pyruvate Carboxylase Deficiency | *PC* | |

| Disease | Gene | X-Linked Disorder |
|---|---|---|
| *RAG2*-Related Disorders | *RAG2* | |
| *RAPSN*-Related Disorders | *RAPSN* | |
| Renal Tubular Acidosis with Deafness | *ATP6V1B1* | |
| Retinitis Pigmentosa Type 25 | *EYS* | |
| Retinitis Pigmentosa Type 26 | *CERKL* | |
| Retinitis Pigmentosa Type 28 | *FAM161A* | |
| Retinitis Pigmentosa Type 59* | *DHDDS* | |
| Rhizomelic Chondrodysplasia Punctata, Type 1 | *PEX7* | |
| *RPE65*-Related Disorders | *RPE65* | |
| *RTEL1*-Related Disorders | *RTEL1* | |
| Sandhoff Disease | *HEXB* | |
| Sjogren-Larsson Syndrome | *ALDH3A2* | |
| *SLC26A2*-Related Disorders | *SLC26A2* | |
| Smith-Lemli-Opitz Syndrome | *DHCR7* | |
| Spastic Paraplegia 49 | *TECPR2* | |
| Spastic Paraplegia Type 15 | *ZFYVE26* | |
| Spinal Muscular Atrophy* | *SMN1* | |
| Spinocerebellar Ataxia, *ANO10*-Related | *ANO10* | |
| Spondylothoracic Dysostosis | *MESP2* | |
| Surfactant Deficiency, *ABCA3*-Related | *ABCA3* | |
| *TGM1*-Related Autosomal Recessive Congenital Ichthyosis | *TGM1* | |
| *TPP1*-Related Neuronal Ceroid Lipofuscinosis | *TPP1* | |
| Transient Infantile Liver Failure | *TRMU* | |
| Tyrosine Hydroxylase Deficiency | *TH* | |
| 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* | |
| Vitamin D–Dependent Rickets, *CYP27B1*-Related | *CYP27B1* | |
| VSX2-Related Microphthalmia, Anophthalmia and Coloboma | *VSX2* | |
| Wilson Disease | *ATP7B* | |
| Xeroderma Pigmentosum Group A | *XPA* | |
| Xeroderma Pigmentosum Group C | *XPC* | |
| X-Linked Adrenal Hypoplasia Congenita | *NR0B1* | ✓ |
| X-Linked Adrenoleukodystrophy | *ABCD1* | ✓ |
| X-Linked Alport Syndrome | *COL4A5* | ✓ |
| X-Linked Juvenile Retinoschisis | *RS1* | ✓ |
| X-Linked Myotubular Myopathy | *MTM1* | ✓ |
| X-Linked Retinal Dystrophy, *RPGR*-Related | *RPGR* | ✓ |
| X-Linked Severe Combined Immunodeficiency | *IL2RG* | ✓ |

*Targeted analysis and/or custom assay
# Does not include mild *A222V* variant

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