

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

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

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	
Friederich Ataxia*	FXN	
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	

Disease	Gene	X-Linked Disorder
Hemophilia A	F8	✓
Hemophilia B	F9	✓
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	
Hydroletharus 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	
Methylmalonic 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	
Mucopolipidosis III Gamma	GNPTG	
Mucopolipidosis 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	

Disease	Gene	X-Linked Disorder
NAGA-Related Disorders	NAGA	
NDUFAF5-Related Mitochondrial Complex 1 Deficiency*	NDUFAF5	
NDUFS4-Related Mitochondrial Complex 1 Deficiency	NDUFS4	
NDUFS6-Related Mitochondrial Complex 1 Deficiency	NDUFS6	
NEB-Related Nemaline Myopathy	NEB	
Nephrotic Syndrome, <i>NPHS1</i> -Related	NPHS1	
Nephrotic Syndrome, <i>NPHS2</i> -Related	NPHS2	
Neuronal Ceroid Lipofuscinosis, <i>CLN6</i> -Related	CLN6	
Niemann-Pick Disease Type C1	NPC1	
Niemann-Pick Disease Type C2	NPC2	
Niemann-Pick Disease, <i>SMPD1</i> -Related	SMPD1	
Nijmegen Breakage Syndrome	NBN	
Normophosphatemic Familial Tumoral Calcinosis	SAMD9	
NR2E3-Related Disorders	NR2E3	
Oculocutaneous Albinism, <i>OCA2</i> -Related	OCA2	
Oculocutaneous Albinism, <i>TYR</i> -Related	TYR	
Opitz G/BBB Syndrome, <i>MID1</i> -Related	MID1	✓
Ornithine Aminotransferase Deficiency	OAT	
Ornithine Transcarbamylase Deficiency	OTC	✓
PCCA-Related Propionic Acidemia	PCCA	
PCCB-Related Propionic Acidemia	PCCB	
<i>PCDH15</i> -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	
<i>PHYH</i> -Related Refsum Disease	PHYH	
<i>PLP1</i> -Related Disorders	PLP1	✓
<i>POLG</i> -Related Disorders	POLG	
<i>POMGNT1</i> -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	
<i>PPT1</i> -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, <i>MCPH1</i> -Related	MCPH1	
Primary Trimethylaminuria	FMO3	

Disease	Gene	X-Linked Disorder
Pycnodysostosis	CTSK	
Pyruvate Carboxylase Deficiency	PC	
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	
<i>RPE65</i> -Related Disorders	RPE65	
<i>RTEL1</i> -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, <i>ANO10</i> -Related	ANO10	
Spondylothoracic Dysostosis	MESP2	
Surfactant Deficiency, <i>ABCA3</i> -Related	ABCA3	
<i>TGM1</i> -Related Autosomal Recessive Congenital Ichthyosis	TGM1	
<i>TPP1</i> -Related Neuronal Ceroid Lipofuscinosis	TPP1	
Transient Infantile Liver Failure	TRMU	
Tyrosine Hydroxylase Deficiency	TH	
Tyrosinemia Type I	FAH	
Tyrosinemia Type II	TAT	
<i>USH1C</i> -Related Disorders	USH1C	
<i>USH2A</i> -Related Disorders	USH2A	
Usher Syndrome Type 3	CLRN1	
Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	
Vitamin D-Dependent Rickets, <i>CYP27B1</i> -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	NROB1	✓
X-Linked Adrenoleukodystrophy	ABCD1	✓
X-Linked Alport Syndrome	COL4A5	✓
X-Linked Juvenile Retinoschisis	RS1	✓
X-Linked Myotubular Myopathy	MTM1	✓
X-Linked Retinal Dystrophy, <i>RPGR</i> -Related	RPGR	✓
X-Linked Severe Combined Immunodeficiency	IL2RG	✓

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