Foresight®

Carrier Screen

Universal Plus 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.

Disease	Gene	ACOG Guidelines[1]	ACMG Guidelines[2]	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-1 Antitrypsin Deficiency^	*SERPINA1*			
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*		✓	

Disease	Gene	ACOG Guidelines	ACMG Guidelines	X-Linked Disorder
Biotinidase Deficiency	*BTD*		✓	
Bloom Syndrome	*BLM*		✓	
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*			

Disease	Gene	ACOG Guidelines	ACMG Guidelines	X-Linked Disorder
Delta-Sarcoglycanopathy	*SGCD*			
Dihydrolipoamide Dehydrogenase Deficiency	*DLD*		✓	
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 V Leiden Thrombophilia**^	*F5*			
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*			

Disease	Gene	ACOG Guidelines	ACMG Guidelines	X-Linked Disorder
GNPTAB-Related Disorders	*GNPTAB*		✓	
HADHA-Related Disorders	*HADHA*			
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*	✓	✓	
HFE-Associated Hereditary Hemochromatosis*^	*HFE*			
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*			
Methylmalonic Acidemia, *MMUT*-Related	*MMUT*		✓	
Methylmalonic Acidemia and Homocystinuria, cblC Type	*MMACHC*		✓	
Mevalonate Kinase Deficiency	*MVK*		✓	
Mild *MTHFR* Deficiency*^	*MTHFR*			
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*			

Disease	Gene	ACOG Guidelines	ACMG Guidelines	X-Linked Disorder
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*			
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*		✓	

Disease	Gene	ACOG Guidelines	ACMG Guidelines	X-Linked Disorder
Prothrombin Thrombophilia‡^	*F2*			
Pseudocholinesterase Deficiency	*BCHE*			
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*			
RPE65-Related Disorders	*RPE65*			
RTEL1-Related Disorders	*RTEL1*			
Sandhoff Disease	*HEXB*			
Short-Chain Acyl-CoA Dehydrogenase Deficiency	*ACADS*			
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
**Targeted analysis, must be ordered with prothrombin
‡ Targeted analysis, must be ordered with factor V Leiden
# Does not include mild *A222V* variant
^ Opt-in condition

Myriad genetics®

Carrier screening panels designed with *every* patient in mind

Myriad Genetics pioneered the first expanded carrier screening to maximize detection of at-risk couples. Our goal is to produce not simply more information, but *meaningful* clinical information. We offer four standard panels:

Fundamental

Cystic fibrosis, spinal muscular atrophy, and fragile X

Fundamental Plus

Focused set of 14 conditions, including ACOG-recommended conditions associated with ethnicity

Universal

176 conditions associated with serious and prevalent inherited conditions

Universal Plus

Comprehensive panel of 267 conditions including ACMG Tier 3 category recommended conditions*

Panels may be customized based on clinical needs

Meet the Pantier family

Real-world impact:

Ashley Pantier shares the positive health impact the Foresight® Carrier Screen has had on her family.

Scan to watch Ashley's video

**F8* and *FXN* genes recommended by ACMG are not included

1. Carrier screening in the age of genomic medicine. Committee Opinion No. 690. American College of Obstetricians and Gynecologists. Obstet Gynecol 2017;129:e35–40. **2.** Gregg AR, Aarabi M, Klugman S, 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) [published correction appears in Genet Med. 2021 Aug 27;:]. Genet Med. 2021;23(10):1793-1806. doi:10.1038/s41436-021-01203-z.

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