geneaware

Baylor Miraca Genetics Laboratories

GENEAWARE REPRODUCTIVE CARRIER SCREEN

GeneAware is a reproductive carrier screen that detects disease-causing variants in over 150 genes by full gene sequencing, supplemented with copy number analysis for genes with frequent deletions. These pathogenic variants are associated with serious disorders such as Duchenne muscular dystrophy, alpha-thalassemia, and MECP2 duplication syndrome, which are not routinely included in other carrier testing panels.

We know that the risk for carrying certain genetic conditions varies from patient to patient based on several factors. Because of this diversity, we offer four different **GeneAware** panels to better meet the needs of your patients. Females are screened for X-linked Duchenne and Becker muscular dystrophies and Fragile-X syndrome in all four panel options.

ACMG and ACOG – disorders recommended for screening by the ACMG and ACOG
Ashkenazi Jewish – over 35 disorders specific for individuals of Ashkenazi Jewish descent
Basic – several of the most commonly requested disorders
Complete – over 150 disorders included for the most comprehensive screening

ACMG and ACOG

Alpha-Thalassemia (HBA1 and HBA2)		C	S
Beta-Hemoglobinopathies (Beta-Thalassemia and Sickle Cell Disease, HBB)		C	S
Bloom Syndrome (BLM)			S
Canavan Disease (ASPA)			S
Cystic Fibrosis (CFTR)		C	S
Duchenne/Becker Muscular Dystrophy (DMD)*		C	S
Familial Dysautonomia (IKBKAP)			S
Fanconi Anemia (FANCC)			S
Fragile-X Syndrome (FMR1)*	CGG		
Gaucher Disease (GBA)			S
Mucolipidosis IV (MCOLN1)		C	S
Niemann-Pick Disease, Type A (SMPD1)			S
Spinal Muscular Atrophy (SMN1)		C	
Tay-Sachs Disease (HEXA)		C	S

Basic

Alpha-Thalassemia (HBA1 and HBA2)		C	S
Beta-Hemoglobinopathies (Beta-Thalassemia and Sickle Cell Disease, HBB)		C	S
Cystic Fibrosis (CFTR)		C	S
Duchenne/Becker Muscular Dystrophy (DMD)*		C	S
Fragile-X Syndrome (FMR1)*	CGG		
Spinal Muscular Atrophy (SMN1)		C	

Ashkenazi Jewish

3-Phosphoglycerate Dehydrogenase Deficiency (PHGDH)			S
Abetalipoproteinaemia (MTTP)			S
Alport Syndrome (COL4A3)			S
Arthrogryposis, Mental Retardataion and Seizures (SLC35A3)			S
Autosomal Recessive Polycystic Kidney Disease (PKHD1)			S
Bardet-Biedl Syndrome: BBS2 Related (BBS2)			S
Bloom Syndrome (BLM)			S
Canavan Disease (ASPA)			S
Carnitine Palmitoyltransferase II Deficiency (CPT2)			S
Congenital Amegakaryocytic Thrombocytopenia (MPL)			S
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2)			S
Cystic Fibrosis (CFTR)		C	S
Dihydrolipoamide Dehydrogenase Deficiency (DLD)			S
Duchenne/Becker Muscular Dystrophy (DMD)*		C	S
Dyskeratosis Congenita (RTEL1)			S
Ehlers-Danlos Syndrome VIIc (ADAMTS2)			S
Familial Dysautonomia (IKBKAP)			S
Familial Hyperinsulinism (ABCC8)			S
Fanconi Anemia (FANCC)			S
Fragile-X Syndrome (FMR1)*	CGG		
Fukuyama Congenital Muscular Dystrophy/Walker-Warburg Congenital Muscular Dystrophy (FKTN)			S
Galactosemia (GALT)		C	S
Gaucher Disease (GBA)			S
Glycogen Storage Disease: Type IA (G6PC)			S
Joubert Syndrome, TMEM216 Related (TMEM216)			S
Maple Syrup Urine Disease: Type 1B (BCKDHB)			S
Mucolipidosis IV (MCOLN1)		C	S
Multiple Sulphatase Deficiency (SUMF1)			S
Nemaline Myopathy: NEB Related (NEB)		C	
Niemann-Pick Disease, Type A (SMPD1)			S
Retinitis Pigmentosa, Autosomal Recessive (DHDDS)			S
Smith-Lemli-Opitz Syndrome (DHCR7)			S
Spinal Muscular Atrophy (SMN1)		C	
Tay-Sachs Disease (HEXA)		C	S
Tyrosinemia: Type I (FAH)			S
Usher Syndrome: Type 1F (PCDH15)			S
Usher Syndrome: Type 3A (CLRN1)			S
Wilson Disease (ATP7B)			S
Zellweger Spectrum Disorders: PEX2 Related (PEX2)			S

KEY

C	Copy Number Analysis	S	Full Sequencing Analysis
CGG	CGG Repeat Analysis	*	Females Only

Phone: 1-800-411-GENE (4363) • E-mail: geneticstest@bcm.edu

3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency (HMGCL)			S
3-Phosphoglycerate Dehydrogenase Deficiency (PHGDH)			S
Abetalipoproteinaemia (MTTP)			S
Adenosine Deaminase Deficiency (ADA)			S
Adrenoleukodystrophy (ABCD1)*			S
Agammaglobulinemia, X-linked 1 (BTK)*			S
Alpha-1-Antitrypsin Deficiency (SERPINA1)			S
Alpha-Mannosidosis (MAN2B1)			S
Alpha-Thalassemia (HBA1 and HBA2)		C	S
Alport Syndrome (COL4A3)			S
Angelman Syndrome (UBE3A)			S
Argininosuccinate Lyase Deficiency (ASL)			S
Arthrogryposis, Mental Retardataion and Seizures (SLC35A3)			S
Aspartylglucosaminuria (AGA)			S
Ataxia with Vitamin E Deficiency (TTPA)			S
Ataxia-Telangiectasia (ATM)			S
Atelosteogenesis Type 2 (SLC26A2)			S
Autosomal Recessive Congenital Ichthyosis, TGM1 Related (TGM1)			S
Autosomal Recessive Polycystic Kidney Disease (PKHD1)			S
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)			S
Bardet-Biedl Syndrome: BBS1 Related (BBS1)			S
Bardet-Biedl Syndrome: BBS2 Related (BBS2)			S
Bardet-Biedl Syndrome: BBS10 Related (BBS10)			S
Beta-Hemoglobinopathies (Beta-Thalassemia and Sickle Cell Disease, HBB)		C	S
BH4-Deficient Hyperphenylalaninemia A (PTS)			S
Biotinidase Deficiency (BTD)			S
Bloom Syndrome (BLM)			S
Canavan Disease (ASPA)			S
Carnitine Deficiency, Systemic Primary (SLC22A5)			S
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)			S
Carnitine Palmitoyltransferase II Deficiency (CPT2)			S
Cartilage-Hair Hypoplasia (RMRP)			S
Cerebrotendinous Xanthomatosis (CYP27A1)			S
Chronic Granulomatous Disease, X-linked (CYBB)*			S
Citrin Deficiency (SLC25A13)			S
Citrullinemia Type 1 (ASS1)			S
Congenital Amegakaryocytic Thrombocytopenia (MPL)			S
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2)			S
Congenital Disorder of Glycosylation: Type 1B: MPI Related (MPI)			S
Congenital Myasthenic Syndrome, CHRNE Related (CHRNE)			S
Congenital Myasthenic Syndrome, DOK7 Related (DOK7)			S
Congenital Myasthenic Syndrome, RAPSN Related (RAPSN)			S
Congenital Myasthenic Syndrome, CHAT Related (CHAT)			S
Crigler-Najjar Syndrome (UGT1A1)			S
Cystic Fibrosis (CFTR)		C	S
Cystinosis (CTNS)		C	S
D-Bifunctional Protein Deficiency (HSD17B4)			S
Dihydrolipoamide Dehydrogenase Deficiency (DLD)			S
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)			S
Duchenne/Becker Muscular Dystrophy (DMD)*		C	S
Dyskeratosis Congenita (RTEL1)			S
Ehlers-Danlos Syndrome VIIc (ADAMTS2)			S
Ethylmalonic Encephalopathy (ETHE1)			S
Familial Dysautonomia (IKBKAP)			S
Familial Hyperinsulinism (ABCC8)			S
Fanconi Anemia (FANCC)			S
Fragile-X Syndrome (FMR1)*	CGG		
Fukuyama Congenital Muscular Dystrophy/Walker-Warburg Congenital Muscular Dystrophy (FKTN)			S
Fumarase Hydratase Deficiency (FH)			S
Galactosemia (GALT)		C	S
Gaucher Disease (GBA)			S
Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)*			S
Glutaric Acidemia I (GCDH)			S
Glycine Encephalopathy (AMT)			S
Glycine Encephalopathy (GLDC)			S
Glycogen Storage Disease: Type IA (G6PC)			S
Glycogen Storage Disease: Type IB (SLC37A4)			S
Glycogen Storage Disease: Type II (Pompe Disease) (GAA)			S
Glycogen Storage Disease: Type III (AGL)			S
GM1-Gangliosidosis (GLB1)			S
GRACILE Syndrome (BCS1L)			S
Hereditary Fructose Intolerance (ALDOB)			S
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum (SLC12A6)			S
Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3)			S
Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3)			S
Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2)			S
Hermansky-Pudlak Syndrome: HPS3 Related (HPS3)			S
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency (CBS)			S
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome (SLC25A15)			S

Hypophosphatasia (ALPL)			S
Inclusion Body Myopathy: Type 2 (GNE)			S
Infantile Neuroaxonal Dystrophy 1 (PLA2G6)			S
Isovaleric Acidemia (IVD)			S
Joubert Syndrome, TMEM216 Related (TMEM216)			S
Juvenile Nephronopthisis (NPHP1)		C	
Krabbe Disease (GALC)		C	S
Leigh Syndrome: French-Canadian Type (LRPPRC)			S
Leukoencephalopathy with Vanishing White Matter, EIF2B5 Related (EIF2B5)			S
Limb-Girdle Muscular Dystrophy, Type 2A (CAPN3)			S
Limb-Girdle Muscular Dystrophy, Type 2C (SGCG)			S
Limb-Girdle Muscular Dystrophy, Type 2D (SGCA)			S
Limb-Girdle Muscular Dystrophy, Type 2E (SGCB)			S
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA)			S
Lowe Syndrome (OCRL)*			S
Lysinuric Protein Intolerance (SLC7A7)			S
Maple Syrup Urine Disease: Type 1A (BCKDHA)			S
Maple Syrup Urine Disease: Type 1B (BCKDHB)			S
Maple Syrup Urine Disease: Type II (DBT)			S
MECP2 Duplication Syndrome (MECP2)*		C	
Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)			S
Megalencephalic Leukoencephalopathy with Subcortical Cysts, MLC1 Related (MLC1)			S
Metachromatic Leukodystrophy (ARSA)			S
Methylmalonic Aciduria and Homocystinuria: Type cblC (MMACHC)			S
Mucolipidosis II (GNPTAB)			S
Mucolipidosis IV (MCOLN1)		C	S
Mucopolysaccharidosis, Type I (IDUA)			S
Mucopolysaccharidosis, Type IIIA (Sanfilippo Syndrome A) (SGSH)			S
Multiple Sulphatase Deficiency (SUMF1)			S
Muscle-Eye-Brain Disease (POMGNT1)			S
Nemaline Myopathy: NEB Related (NEB)		C	
Nephrotic Syndrome: Type 1 (NPHS1)			S
Nephrotic Syndrome: Type 2 (NPHS2)			S
Neuronal Ceroid Lipofuscinosis, CLN3 Related (CLN3)		C	
Neuronal Ceroid Lipofuscinosis, CLN5 Related (CLN5)			S
Neuronal Ceroid Lipofuscinosis, CLN6 Related (CLN6)			S
Neuronal Ceroid Lipofuscinosis, CLN8 Related (CLN8)			S
Neuronal Ceroid Lipofuscinosis, PPT1 Related (PPT1)			S
Neuronal Ceroid Lipofuscinosis, TPP1 Related (TPP1)			S
Niemann-Pick Disease, Type A (SMPD1)			S
Niemann-Pick Disease, Type C (NPC1)			S
Nijmegen Breakage Syndrome (NBN)			S
Nonsyndromic Hearing Loss and Deafness: GJB2 Related DFNB1 (GJB2)			S
Nonsyndromic Hearing Loss and Deafness: GJB6 Related DFNB1 (GJB6)		C	
Ornithine Transcarbamylase Deficiency (OTC)*			S
Pendred Syndrome (SLC26A4)			S
Phenylalanine Hydroxylase Deficiency (PAH)			S
POLG-Related Disorders (POLG)			S
Primary Hyperoxaluria: Type 1 (AGXT)			S
Primary Hyperoxaluria: Type 2 (GRHPR)			S
Primary Congenital Glaucoma (CYP1B1)			S
PROP1-Related Combined Pituitary Hormone Deficiency (PROP1)			S
Propionic Acidemia, PCCA Related (PCCA)			S
Propionic Acidemia, PCCB Related (PCCB)			S
Pycnodysostosis (CTSK)			S
Pyruvate Carboxylase Deficiency (PC)			S
Retinitis Pigmentosa, Autosomal Recessive (DHDDS)			S
Rhizomelic Chondrodysplasia Punctata: Type I (PEX7)			S
Salla Disease (SLC17A5)			S
Sandhoff Disease (HEXB)			S
Severe Combined Immunodeficiency, Athabascan Type (DCLRE1C)			S
Severe Combined Immunodeficiency, X-linked (IL2RG)*			S
Sjogren-Larsson Syndrome (ALDH3A2)			S
Smith-Lemli-Opitz Syndrome (DHCR7)			S
Spinal Muscular Atrophy (SMN1)		C	
Tay-Sachs Disease (HEXA)		C	S
Tyrosine Hydroxylase Deficiency (TH)			S
Tyrosinemia: Type I (FAH)			S
Usher Syndrome: Type 1B (MYO7A)			S
Usher Syndrome: Type 1C (USH1C)			S
Usher Syndrome: Type 1D (CDH23)			S
Usher Syndrome: Type 1F (PCDH15)			S
Usher Syndrome: Type 2A (USH2A)			S
Usher Syndrome: Type 3A (CLRN1)			S
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)			S
Wilson Disease (ATP7B)			S
Wiskott-Aldrich Syndrome (WAS)			S
Zellweger Spectrum Disorders: PEX1 Related (PEX1)			S
Zellweger Spectrum Disorders: PEX2 Related (PEX2)			S

KEY

C	Copy Number Analysis	S	Full Sequencing Analysis
CGG	CGG Repeat Analysis	*	Females Only

ORDERING AND REPORTING INFORMATION

Ordering, reporting, and kit fulfillment are fast and easy with our online provider portal. Results are available within two weeks after samples are received in our laboratory. Complimentary consultations with our certified genetic counselors are available for your patients. Visit www.GeneAware.clinical.bcm.edu to learn more.

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