Baylor Miraca Genetics Laboratories

geneaware

Date: May 14, 2015
To: Valued Clients
From: Baylor Miraca Genetics Laboratories
RE: Test Update Memorandum

The Baylor Miraca Genetics Laboratories (BMGL) is pleased to announce that effective May 26, 2015, the GeneAware carrier screen will now include over 150 genes analyzed by full gene sequencing with copy number analysis on selected genes. As of May 26, the following test codes for GeneAware version 1 will no longer be available.

60100	GeneAware Complete (Female)
60105	GeneAware Complete (Male)
60400	GeneAware Basic (Female)
60405	GeneAware Basic (Male)
60300	GeneAware ACMG/ACOG (Female)
60305	GeneAware ACMG/ACOG (Male)
60200	GeneAware Ashkenazi Jewish (Female)
60205	GeneAware Ashkenazi Jewish (Male)

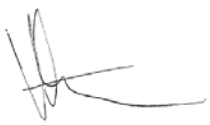
GeneAware v2 will fully replace the tests above and any new orders for version 1 will be changed to version 2. Please see attached information sheet for more details on our GeneAware version 2 panels.

60101	GeneAware Complete v2 (Female)
60106	GeneAware Complete v2 (Male)
60401	GeneAware Basic v2 (Female)
60406	GeneAware Basic v2 (Male)
60301	GeneAware ACMG/ACOG v2 (Female)
60306	GeneAware ACMG/ACOG v2 (Male)
60201	GeneAware Ashkenazi Jewish v2 (Female)
60206	GeneAware Ashkenazi Jewish v2 (Male)

Please direct any billing questions to 713-798-3566 or email us at medgenbilling@bcm.edu. Please visit our website for any additional notices or postings.

Thank you for your valued business. We look forward to providing you with the highest quality genetic testing services.

Sincerely,



Vito Oliveri
Director of Operations
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 screening, 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. Four panel options are offered for **GeneAware**, allowing flexibility based on your patient's needs. Females are screened for X-linked Duchenne and Becker muscular dystrophies and Fragile-X syndrome in all four panel options.

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.

ACMG and ACOG

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

** females only*

Basic

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

** females only*

Ashkenazi Jewish

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

** females only*

Complete Panel

GENEAWARE REPRODUCTIVE CARRIER SCREEN

3-Hydroxy-3-Methylglutaryl CoA lyase Deficiency (HMGCL)	
3-phosphoglycerate dehydrogenase deficiency (PHGDH)	○
Abetalipoproteinaemia (MTTP)	○
Adenosine Deaminase Deficiency (ADA)	
Adrenoleukodystrophy (ABCD1)	
Agammaglobulinemia, X-linked 1 (BTK)	
Alpha-1-Antitrypsin Deficiency (SERPINA1)	
Alpha-Mannosidosis (MAN2B1)	
Alpha-Thalassemia (HBA1 and HBA2)	● ■
Alport syndrome (COL4A3)	○
Angelman syndrome (UBE3A)	
Argininosuccinate Lyase Deficiency (ASL)	
Arthrogryposis, mental retardataion and seizures (SLC35A3)	○
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	
Aspartylglucosaminuria (AGA)	
Ataxia with Vitamin E Deficiency (TTPA)	
Ataxia-Telangiectasia (ATM)	
Atelosteogenesis Type 2 (SLC26A2)	
Autosomal Recessive Congenital Ichthyosis, TGM1 Related (TGM1)	
Autosomal Recessive Polycystic Kidney Disease (PKHD1)	○
Bardet-Biedl Syndrome: BBS1 Related (BBS1)	
Bardet-Biedl Syndrome: BBS10 Related (BBS10)	
Bardet-Biedl syndrome (BBS2)	○
Beta-Hemoglobinopathies (Beta-Thalassemia and Sickle Cell Disease, HBB)	● ■
BH4-Deficient Hyperphenylalaninemia A (PTS)	
Biotinidase Deficiency (BTD)	
Bloom Syndrome (BLM)	● ○
Canavan Disease (ASPA)	● ○
Carnitine deficiency, systemic primary (SLC22A5)	
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	
Carnitine Palmitoyltransferase II Deficiency (CPT2)	○
Cartilage-Hair Hypoplasia (RMRP)	
Cerebrotendinous Xanthomatosis (CYP27A1)	
Chronic granulomatous disease, X-linked (CYBB)	
Citrin Deficiency (SLC25A13)	
Citrullinemia Type 1 (ASS1)	
Congenital Amegakaryocytic Thrombocytopenia (MPL)	○
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2)	○
Congenital Disorder of Glycosylation: Type 1B: MPI Related (MPI)	
Congenital Myasthenic Syndrome, CHRNE-Related (CHRNE)	
Congenital Myasthenic Syndrome, DOK7-Related (DOK7)	
Congenital Myasthenic Syndrome, RAPSN-Related (RAPSN)	
Congenital Myasthenic Syndrome, CHAT-Related (CHAT)	

Crigler-Najjar Syndrome (UGT1A1)	
Cystic Fibrosis (CFTR)	● ○ ■
Cystinosis (CTNS)	
D-Bifunctional Protein Deficiency (HSD17B4)	
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	○
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	
Duchenne/Becker Muscular Dystrophy (DMD)*	● ○ ■
Dyskeratosis Congenita (RTEL1)	○
Ehlers-Danlos syndrome VIIc (ADAMTS2)	○
Ethylmalonic Encephalopathy (ETHE1)	
Familial Dysautonomia (IKBKAP)	● ○
Familial Hyperinsulinism (ABCC8)	○
Fanconi Anemia (FANCC)	● ○
Fragile-X syndrome (FMR1)*	● ○ ■
Fukuyama Congenital Muscular Dystrophy (FKTN)	○
Fumarase Hydratase Deficiency (FH)	
Galactosemia (GALT)	○
Gaucher Disease (GBA)	● ○
Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)*	
Glutaric Acidemia I (GCDH)	
Glycine encephalopathy (AMT)	
Glycine encephalopathy (GLDC)	
Glycogen Storage Disease Type II (Pompe Disease) (GAA)	
Glycogen Storage Disease Type III (AGL)	
Glycogen Storage Disease: Type IA (G6PC)	○
Glycogen Storage Disease: Type IB (SLC37A4)	
GM1-gangliosidosis (GLB1)	
GRACILE syndrome (BCS1L)	
Hereditary Fructose Intolerance (ALDOB)	
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum (SLC12A6)	
Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3)	
Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3)	
Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2)	
Hermansky-Pudlak Syndrome: HPS3 Related (HPS3)	
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency (CBS)	
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome (SLC25A15)	
Hypophosphatasia (ALPL)	
Inclusion Body Myopathy: Type 2 (GNE)	
Infantile neuroaxonal dystrophy 1 (PLA2G6)	
Isovaleric Acidemia (IVD)	
Joubert Syndrome, NPHP1 Related (NPHP1)	
Joubert Syndrome, TMEM216 Related (TMEM216)	○

KEY ● ACMG and ACOG ○ Ashkenazi Jewish ■ Basic

Krabbe Disease (GALC)	
Leigh Syndrome: French-Canadian Type (LRPPRC)	
Leukoencephalopathy with Vanishing White Matter, EIF2B5 Related (EIF2B5)	
Limb-Girdle Muscular Dystrophy, Type 2A (CAPN3)	
Limb-Girdle Muscular Dystrophy, Type 2C (SGCG)	
Limb-Girdle Muscular Dystrophy, Type 2D (SGCA)	
Limb-Girdle Muscular Dystrophy, Type 2E (SGCB)	
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA)	
Lowe syndrome (OCRL)	
Lysinuric Protein Intolerance (SLC7A7)	
Maple Syrup Urine Disease: Type 1A (BCKDHA)	
Maple Syrup Urine Disease: Type 1B (BCKDHB)	○
Maple syrup urine disease, type II (DBT)	
MECP2 duplication syndrome (MECP2)*	
Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	
Megalencephalic Leukoencephalopathy with Subcortical Cysts, MLC1 Related (MLC1)	
Metachromatic Leukodystrophy (ARSA)	
Methylmalonic Aciduria and Homocystinuria: Type cblC (MMACHC)	
Mucolipidosis II (GNPTAB)	
Mucolipidosis IV (MCOLN1)	● ○
Mucopolysaccharidosis Type I (IDUA)	
Mucopolysaccharidosis, Type IIIA (Sanfilippo Syndrome A) (SGSH)	
Multiple sulphatase deficiency (SUMF1)	○
Muscle-Eye-Brain Disease (POMGNT1)	
Nemaline Myopathy: NEB Related (NEB)	○
Nephrotic Syndrome: Type 1 (NPHS1)	
Nephrotic Syndrome: Type2 (NPHS2)	
Neuronal Ceroid Lipofuscinosis, CLN3-Related (CLN3)	
Neuronal Ceroid Lipofuscinosis, CLN5-Related (CLN5)	
Neuronal Ceroid Lipofuscinosis, CLN6-Related (CLN6)	
Neuronal Ceroid Lipofuscinosis, CLN8-Related (CLN8)	
Neuronal Ceroid Lipofuscinosis, PPT1-Related (PPT1)	
Neuronal Ceroid Lipofuscinosis, TPP1-Related (TPP1)	
Niemann-Pick Disease, Type A (SMPD1)	● ○
Niemann-Pick Disease, Type C (NPC1)	
Nijmegen Breakage Syndrome (NBN)	
Nonsyndromic Hearing Loss and Deafness: GJB2 related DFNB1 (GJB2)	
Nonsyndromic Hearing Loss and Deafness: GJB6 related DFNB1 (GJB6)	
Ornithine transcarbamylase deficiency (OTC)	
Pendred Syndrome (SLC26A4)	
Phenylalanine Hydroxylase Deficiency (PAH)	
POLG-Related Disorders (POLG)	

Primary Hyperoxaluria: Type 1 (AGXT)	
Primary Hyperoxaluria: Type 2 (GRHPR)	
Primary Congenital Glaucoma (CYP1B1)	
PROP1-Related Combined Pituitary Hormone Deficiency (PROP1)	
Propionic Acidemia, PCCA Related (PCCA)	
Propionic Acidemia, PCCB Related (PCCB)	
Pycnodysostosis (CTSK)	
Pyruvate Carboxylase Deficiency (PC)	
Retinitis pigmentosa, autosomal recessive (DHDDS)	○
Rhizomelic Chondrodysplasia Punctata: Type I (PEX7)	
Salla Disease (SLC17A5)	
Sandhoff Disease (HEXB)	
Severe Combined Immunodeficiency, Athabascan type (DCLRE1C)	
Severe combined immunodeficiency, X-linked (IL2RG)	
Sjogren-Larsson Syndrome (ALDH3A2)	
Smith-Lemli-Opitz Syndrome (DHCR7)	○
Spinal Muscular Atrophy (SMN1)	● ○ ■
Tay-Sachs Disease (HEXA)	● ○
Tyrosine Hydroxylase Deficiency (TH)	
Tyrosinemia: Type I (FAH)	○
Usher Syndrome: Type 1B (MYO7A)	
Usher Syndrome: Type 1C (USH1C)	
Usher Syndrome: Type 1D (CDH23)	
Usher Syndrome: Type 1F (PCDH15)	○
Usher Syndrome: Type 2A (USH2A)	
Usher Syndrome: Type 3A (CLRN1)	○
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	
Wilson Disease (ATP7B)	○
Wiskott-Aldrich syndrome (WAS)	
Zellweger Spectrum Disorders: PEX1 Related (PEX1)	
Zellweger syndrome (PEX2)	○

** females only*