FEATURED BIOMARKER TESTS

BENEFITS OF BIOMARKER TESTING

- Biomarker testing can provide further evidence to support a diagnosis of metabolic disease
- Requires minimal specimen and can often be performed on a previously collected specimen
- Cost-effective and time saving

C27 BILE ACIDS (PEROXISOMAL DISORDERS)  
MAYO TEST ID: BAIPD

- C27 bile acids are a diagnostic marker for peroxisomal biogenesis disorders and single enzyme defects of bile acid synthesis
- May be used to monitor treatment efficacy

CERAMIDE TRIHEXOSIDE/SULFATIDES (FABRY DISEASE, MLD, MSD, SAP-B DEFICIENCY)  
MAYO TEST ID: CTSA

- Urinary excretion of ceramide trihexosides can be suggestive of Fabry disease, while excretion of sulfatides with or without ceramide trihexosides can be suggestive of metachromatic leukodystrophy, multiple sulfatase deficiency, mucolipidosis II (I-cell disease), or saposin B deficiency
- Heightened sensitivity and specificity for Fabry, MLD, MSD, SAP-B, and some (likely more severe) cases of mucolipidosis II (I-Cell)

DERMATAN SULFATE AND HEPARAN SULFATE (MUCOPOLYSACCHARIDOSES TYPES I, II, III, AND IV)  
MAYO TEST ID: MPSBS

- Dermatan sulfate and heparan sulfate are markers for a subset of mucopolysaccharidoses (MPS)
- This test is used to aid in the diagnosis and monitoring of patients with MPS types I, II, III, and VI
- This test may also be used as a second tier newborn screen for MPS types I and II

FRATAXIN (FRIEDREICH ATAXIA)  
MAYO TEST ID: FFRBS

- A protein-based assay measuring concentration of frataxin that is suitable for use as a diagnostic test as well as for monitoring treatment in individuals with FA
- Frataxin protein analysis is a cost-effective and quick method for establishing a diagnosis of FA and will detect rare variants otherwise missed by common molecular-based trinucleotide repeat analysis
FEATURED BIOMARKER TESTS CONTINUED

GLUCOPSYCHOSINE (GAUCHER DISEASE)
MAYO TEST ID: GPSY
• Glucopsychosine is elevated in symptomatic patients with Gaucher disease and may be helpful in determining treatment response
• Quantification of glucopsychosine (glucosylsphingosine) in dried blood spots supports the biochemical diagnosis of Gaucher disease

KERATAN SULFATE QUANTITATIVE, URINE
MAYO TEST ID: KSQNU
• Keratan sulfate is a marker for MPS IV.
• This test is used to monitor keratin sulfate levels for patients on treatment

OXYSTEROLS (Niemann-Pick Disease)
MAYO TEST ID: OXNP
• Quantification of cholestane-3-beta, 5-alpha, 6-beta-triol, 7-ketocholesterol, and lyso-sphingomyelin in plasma for the suspicion of Niemann-Pick type A, Niemann-Pick type B, or Niemann-Pick type C disease
• May eliminate need for fibroblast analysis in Niemann-Pick type A/B/C
• May be useful as surrogate functional biochemical testing for VUS or yet non-published mutations
• May be a useful marker to monitor response to treatment

POLYOLS (Transaldolase Deficiency)
MAYO TEST ID: TALDO
• Polyols analysis in urine is the method of choice for the biochemical diagnosis of transaldolase deficiency (TALDO) and ribose-5-phosphate isomerase RPI deficiency
• This test should be ordered in all patients with liver disorder with normal LFTs

PSYCHOSINE (Krabbe Disease)
MAYO TEST ID: PSY
• Quantification of psychosine (galactosylsphingosine) in dried blood spots supports the biochemical diagnosis and follow-up of individuals affected with Krabbe disease
• Paired with enzyme analysis, it can provide clarity for newborn screen results