Autism Spectrum Disorders: Tier 1 Biochemical Panel

Test Code: BB021  
Turnaround time: 2 weeks  
CPT Codes: 82139 x1, 82542 x1, 82570 x1, 83789 x1, 83864 x1, 83918 x1, 84375 x1, 84377 x1

### Condition Description

#### Genetics of Autism Spectrum Disorders

Autism spectrum disorders (ASDs) are a group of neurodevelopmental disorders which include autism, pervasive developmental delay—not otherwise specified (PDD-NOS), and Asperger syndrome. ASDs are characterized by impairments in social relationships, variable degrees of language and communication deficits, and repetitive behaviors and/or a narrow range of interests. The age of onset is prior to age 3 with a variable clinical presentation, ranging in severity both amongst individuals as well as amongst the various subtypes of ASDs. Additional clinical features may also be observed in individuals with an ASD, such as intellectual disability (up to ~50%) and seizures (~25%).

Known genetic causes of autism include cytogenetically visible chromosome abnormalities (3-5%), copy number variants—which include submicroscopic deletions and duplications (~6-7%), and single gene disorders (~5%).

Emory Genetics Laboratory’s integrated testing strategy allows for a comprehensive cytogenetic, metabolic, and molecular analysis of ASD in your patient. For a summary of autism testing at EGL, please click here.

References:


### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of autism or an autism spectrum disorder.
- Carrier testing in adults with a family history of autism or an autism spectrum disorder.

### Methodology

#### Urine Organic Acids:
Qualitative and quantitative determination performed by gas chromatography/mass spectrometry.

#### Plasma amino acids:
Quantitative ion exchange chromatography, reported as micromoles/L.

#### Smith-Lemli-Opitz Screen:
Isotope dilution method by LC-MSMS

#### Lysosomal Storage Disease Screen

Oligosaccharides: MALDI-TOF/TOF  
The traditional one-dimensional thin-layer chromatography method for urine oligosaccharides analysis has limited specificity and sensitivity and provides no structural information that is often needed for diagnosis. This test provides a sensitive screening method for structural analysis of urinary oligosaccharides, glycan and glycoamino acids by Matrix-Assisted Laser Desorption/Ionization-Time of flight/Time of flight (MALDI-TOF/TOF).

GAGS: Dimethylene Blue Binding Quantitation and Thin Layer Chromatography

### Detection

#### Urine Organic Acids:
Test results can be influenced by the age and eating status of the patient. A second test (amino acids analysis/acylcarnitine profile) is typically required to confirm a diagnosis.

#### Plasma Amino Acids:
This test is very sensitive for specific amino acid disorders, but detection can be sensitive to the age and eating status of the patient.

#### Smith-Lemli-Opitz Screen:
Mild elevations of 7-dehydrocholesterol can occur in patients with hypercholesterolemia and those receiving treatment with haloperidol. Only patients with Smith-Lemli-Opitz syndrome have elevated 7-dehydrocholesterol/cholesterol ratios.

#### Lysosomal Storage Disease Screen:
This test provides a comprehensive tool for initial screening of lysosomal storage disorders. The combination of urine oligosaccharide/free glycan and urine quantitative and fractionation of GAG's increases the sensitivity and specificity of urinary screening for lysosomal storage disorders. Due to the extreme sensitivity of the MALDI-TOF/TOF method, the urine oligo/free glycan profiles can detect lysosomal dysfunctions due to the accumulation of charged storage material such as glycosaminoglycans. While the quantification and fractionation of glycosaminoglycans provides additional information that differentiates between the different types of mucopolysaccharidosis. Abnormal results should be confirmed by enzyme and molecular analysis.

### Reference Range

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Urine Organic Acids: Click here for reference range.

Plasma Amino Acids: NA

Smith-Lemli-Opitz Screen: Reporting will be normal or abnormal based on the value of 7-dehydrocholesterol and the ratio of 7-dehydrocholesterol/cholesterol.

Normal ranges:

- 7-dehydrocholesterol (plasma) <2.67 ug/ml
- Cholesterol (plasma) <2098 ug/ml
- Ratio of 7-dehydrocholesterol/cholesterol 0.003

Lysosomal Storage Disease Screen: Interpretation of the urinary oligosaccharide and free glycan profiles is by pattern recognition.

Specimen Requirements

Submit both of the following specimens

**Type: Urine**

Specimen Requirements:

In a clean container without preservatives: 15-30 ml. Freeze.

Fasting or first void sample is preferable.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

**Type: Plasma**

Specimen Requirements:

In sodium heparin (green top) tube: 2-5 ml

Sample should be collected while fasting or 2-4 hours postprandial.

Centrifuge to separate plasma and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

**Related Tests**

- Autism Panel: Complete Tier 1
- Autism Panel: Tier 1 Cytogenetic and Molecular
- Autism Panel: Tier 2
- The components of Autism Panel: Tier 1 Biochemical are also available individually: urine organic acids, plasma amino acids, Smith-Lemli-Opitz Screen, and lysosomal storage disease screen.