ARUP LABORATORIES

MARCH 2022

BIOCHEMICAL GENETICS

Committed to providing high-quality biochemical genetic testing

ARUP's Biochemical Genetics Laboratory offers a comprehensive metabolic test menu. Consultation with a medical director or genetic counselor is available for providers to discuss test orders or results interpretation.

Our biochemical genetics test menu includes tests for:

Fatty acid oxidation disorders

Primary carnitine deficiency

- Carnitine, free and total, in urine and plasma

MCAD deficiency

- Acylcarnitine profile, plasma
- Acylglycine profile, urine
- Organic acids, urine
- *ACADM* 2 mutation panel and sequencing

VLCAD deficiency

- Acylcarnitine profile, plasma
- Organic acids, urine
- *ACADVL* sequencing and deletion/duplication

Biotinidase deficiency

- Biotinidase enzyme, serum
- Organic acids, urine
- Acylcarnitine profile, plasma
- *BTD* sequencing

Galactosemia

- Galactosemia panel (enzyme plus DNA testing for 9 mutations)
- Galactose-1-phosphate uridyltransferase, red blood cells
- Galactose-1-phosphate in red blood cells
- *GALT* 9 mutation panel and sequencing

Ornithine transcarbamylase deficiency

- Amino acids, plasma
- Orotic acid, urine
- *OTC* sequencing and deletion/duplication

Ehlers-Danlos syndrome type VI (kyphoscoliotic form)

- EDS type VI screen, urine

ARUP is a nonprofit enterprise of the University of Utah and its Department of Pathology.

BIOCHEMICAL GENETICS TESTING

Test #	Test Name/Description
0040033	Acylcarnitine Quantitative Profile, Plasma
0081170	Acylglycines, Quantitative, Urine
2011415	Alpha-Iduronidase Enzyme Activity in Leukocytes
0080137	Amino Acids Quantitative by LC-MS/MS, CSF
2009389	Amino Acids Quantitative by LC-MS/MS, Plasma
2009419	Amino Acids Quantitative by LC-MS/MS, Urine
2014314	Autism and Intellectual Disability Comprehensive Panel
2014312	Autism and Intellectual Disability Metabolic Panel
0092610	Bile Acids, Fractionated and Total by LC-MS/MS
0093362	Biotinidase, Serum (with Paired Normal Control)
0080068	Carnitine, Free and Total (Includes Carnitine, Esterified)
0081308	Carnitine, Free and Total, Urine
0081110	Carnitine Panel (Free and Total Carnitine, Acylcarnitine), Plasma or Serum
2002328	Creatine Disorders Panel, Serum or Plasma
2002333	Creatine Disorders Panel, Urine
0081106	Cystine Quantitative, Urine
0081105	Cystinuria Panel
0080351	Ehlers-Danlos Syndrome Type VI Screen, Urine
3003086	Fatty Acids Profile, Essential in Red Blood Cells
2013518	Fatty Acids Profile, Essential Serum or Plasma
0081296	Galactose-1-Phosphate in Red Blood Cells
3001790	Galactose-1-Phosphate Uridyltransferase (GALT Enzyme), RBC
0051175	Galactosemia (GALT) Enzyme Activity and 9 Mutations
2014459	Gaucher Disease (GBA), Enzyme Activity in Leukocytes
2001510	Glutarylcarnitine Quantitative, Urine
2012259	Keratan Sulfate, Quantitative by LC-MS/MS, Urine
2012266	Lysosomal Acid Lipase Activity, Dried Blood Spot
2005255	Methylmalonic Acid, Serum or Plasma (Metabolic Disorders)
0081352	Mucopolysaccharides Screen - Electrophoresis and Quantitation, Urine
0081357	Mucopolysaccharides, Quantitative, Urine
3003566	Mucopolysaccharidoses Type 1/2, Total Heparan Sulfate and NRE (Sensi-Pro®) Quantitative, Serum or Plasma
3003552	Mucopolysaccharidoses Type 1/2, Total Heparan Sulfate and NRE (Sensi-Pro®) Quantitative, Urine
3003487	Mucopolysaccharidoses Type 4A/6 Total Chondroitin Sulfate and Dermatan Sulfate with NRE (Sensi-Pro®) Quantitative, Serum
3003539	Mucopolysaccharidoses Type 4A/6 Total Chondroitin Sulfate and Dermatan Sulfate with NRE (Sensi-Pro®) Quantitative, Urine
0098389	Organic Acids, Urine
3000704	Orotic Acid, Urine
0080336	Phenylalanine and Tyrosine, Plasma (monitoring only)
0080315	Phenylalanine Monitoring, Plasma (monitoring only)
2007406	Pipecolic Acid, Serum or Plasma
2008131	Pipecolic Acid, Urine
2014463	Pompe Disease (GAA), Enzyme Activity in Leukocytes
2013352	Pyridoxine-Dependent Epilepsy Panel, Serum or Plasma
2013355	Pyridoxine-Dependent Epilepsy Panel, Urine
2007401	Succinylacetone, Quantitative, Urine
0080355	Tyrosine, Plasma (monitoring only)
2004250	Very Long-Chain and Branched-Chain Fatty Acids Profile

MOLECULAR TESTING
FOR BIOCHEMICAL DISORDERS

Test #	Test Name/Description
0051730	Biotinidase Deficiency (*BTD*) Sequencing
3001851	Fatty Acid Oxidation Disorders Panel, Sequencing
2006697	Galactosemia (*GALT*) Sequencing
0051176	Galactosemia (*GALT*) 9 mutations
2004212	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) Sequencing and Deletion/Duplication*