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 result interpretation.

Our biochemical genetics test menu includes:

Fatty acid oxidation disorders

Primary carnitine deficiency

- Carnitine, free and total, in urine and plasma
- *SLC22A5* sequencing and deletion/duplication

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* 5 mutation panel and sequencing

Tay-Sachs

- Hexosaminidase A and total hexosaminidase in plasma/serum and leukocytes
- Tay-Sachs (*HEXA*) 7 mutation panel and 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

Creatine disorders

- GAA and creatine panel in plasma/serum and urine
- *SLC6A8* sequencing and deletion/duplication
- *GATM* sequencing
- *GAMT* 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
- *PLOD1* sequencing and deletion/duplication

BIOCHEMICAL GENETICS TESTING

Test #	Test Name/Description
0040033	Acylcarnitine, Quantitative Profile, Plasma
0081170	Acylglycine, Quantitative, Urine
2014314	Autism and Intellectual Disability Comprehensive Panel
2014312	Autism and Intellectual Disability Metabolic Panel
2011415	Alpha-Iduronidase Enzyme Activity in Leukocytes
0080137	Amino Acids Quantitative, CSF
2009389	Amino Acids Quantitative by LC-MS/MS, Plasma
2009419	Amino Acids Quantitative by LC-MS/MS, Urine
0093362	Biotinidase, Serum (with paired normal control)
0081110	Carnitine Panel (Free and Total Carnitine, Acylcarnitine), Plasma or Serum
0080068	Carnitine, Free and Total, Plasma
0081308	Carnitine, Free and Total, Urine
0080065	Carnitine, Free, Plasma
0080067	Carnitine, Total, Plasma
2002328	Creatine Disorders Panel, Plasma or Serum
2002333	Creatine Disorders Panel, Urine
0081106	Cystine Quantitative, Urine
0081105	Cystinuria Panel (Arginine, Cystine, Lysine, and Ornithine), Urine
0080351	Ehlers-Danlos Syndrome Type VI Screen, Urine
2013518	Fatty Acids Profile, Essential Serum or Plasma
0081296	Galactose-1-Phosphate in Red Blood Cells
0080125	Galactose-1-Phosphate Uridyltransferase, in Red Blood Cells
0051175	Galactosemia Panel (Enzyme and DNA Testing for 7 Mutations and 2 Variants), Whole Blood
2014459	Gaucher Disease (GBA), Enzyme Activity in Leukocytes
2001510	Glutarylcarnitine Quantitative, Urine
2008121	Hexosaminidase A and Total Hexosaminidase, Plasma or Serum
2008125	Hexosaminidase A and Total Hexosaminidase, Leukocytes
2008129	Hexosaminidase A and Total Hexosaminidase in Plasma with Reflex to Hexosaminidase A and Total Hexosaminidase in Leukocytes
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 Electrophoresis and Quantitation, Urine
0081357	Mucopolysaccharides, Quantitative, Urine
2007599	Mucopolysaccharidosis Type I, Total HS and NRE (SensiPro) Quantitative, Serum or Plasma
2007488	Mucopolysaccharidosis Type I, Total HS and NRE (SensiPro) Quantitative, Urine

Test #	Test Name/Description
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
0080342	Pyridinoline and Deoxypyridinoline by HPLC, Urine
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
2011906	Adrenoleukodystrophy, X-linked (*ABCD1*) Sequencing and Deletion/Duplication*
2011144	Arginine: Glycine Amidinotransferase (*GATM*) Deficiency Sequencing
0051730	Biotinidase Deficiency (*BTD*) Sequencing*
2004203	Carnitine Deficiency, Primary (*SLC22A5*) Sequencing and Deletion/Duplication*
2006261	Citrin Deficiency (*SLC25A13*) Sequencing
2007069	Citrullinemia, Type I (*ASS1*) Sequencing
2008610	Creatine Transporter Deficiency (*SLC6A8*) Sequencing and Deletion/Duplication
2005559	Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Sequencing and Deletion/Duplication
2006697	Galactosemia (*GALT*) Sequencing*
2011140	Guanidinoacetate Methyltransferase (*GAMT*) Deficiency Sequencing
2006274	Inherited Insulin Resistance Syndrome (*INSR*) Sequencing
0051758	Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*) Sequencing*
2004896	Ornithine Transcarbamylase Deficiency (*OTC*) Sequencing and Deletion/Duplication*
2009298	Tay-Sachs Disease (*HEXA*) Sequencing and 7.6kb Deletion*
2004212	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) Sequencing and Deletion/Duplication*

*Additional genetic tests are available for this condition; please see the ARUP Laboratory Test Directory for more information.