

## The Kennedy Krieger Institute

Genetics Laboratories  
707 North Broadway  
Baltimore, MD 21205

Hours of Operation: Monday - Friday 8:00am -5:00pm

### Sample Requirements

For patient samples already collected but not meeting the criteria listed below, contact the laboratory for further guidance.

## Biochemical Genetics Section: 443-923-2782

Test Name	Sample Type	Volume	Preferred Specimen	Stability ambient	Stability refrigerated 4°C	Stability frozen -20°C	TAT	Cost	CPT Code	Comments:
Amino Acid Analysis	Plasma, CSF, Urine	0.5 mL	Frozen	Sub-Optimal	Sub-Optimal	14 days, optimal	3-5 business days	\$185	82139	
Organic Acid Analysis	Urine, random, no preservative	5 mL	Frozen	Sub-Optimal	14 days	12 months	5-10 business days	\$200	83919	This test is performed on Tuesdays and Thursdays. Contact the laboratory for urgent testing
	CSF	1 mL								
<i>Organic Acid Quantification by Stable Isotope Dilution GC/MS or LC/MS</i>										
N-Acetylaspartic acid	Urine	5 mL	Frozen	Sub-Optimal	1 month	10 months	5-10 business days	\$150	83921	
3-Methylglutaconic acid								\$150	83921	
Mevalonic acid								\$150	83921	
Methylmalonic acid	Urine	1 mL	Frozen	Sub-Optimal	1 month	10 months	5-10 business days	\$150	83921	
Orotic acid	Urine	1 mL	Frozen	Sub-Optimal	7 days	1 month	5-10 business days	\$150	83921	
3-Methylglutaconic acid	Plasma	2 mL	Frozen	Sub-Optimal	1 month	10 months	5-10 business days	\$150	83921	
Methylmalonic acid		1 mL					3-5 business days	\$150	83921	
Guanidinoacetate and Creatine	Plasma	1 ml	Frozen	Sub-Optimal	7 days	12 months	5-10 business days	\$150	82542	This test is performed on Mondays
	Urine	1 mL	Frozen	Sub-Optimal	7 days	12 months		\$150	82542	
<i>Sterols</i>										
7-Dehydrocholesterol Plus Other Cholesterol Precursors	Plasma	0.5 mL	Frozen	24 hours as whole blood	7 days	12 months	3-5 business days	\$150	82542	Sterol testing is performed on Wednesdays and Fridays. Contact the laboratory for urgent testing.
Cholestanol, Sitosterol, Campesterol	Plasma	0.5 mL	Frozen	24 hours as whole blood	1 month	12 months		\$150	82542	
7-Dehydrocholesterol, prenatal	Amniotic Fluid	5 mL (*contact lab if < 2 mL available)	Frozen	1 month	1 month	12 months	3-5 business days	\$200	82542	
	CVS	5-10 mg direct tissue	Frozen	24 hours	1 month	12 months		\$200	82542	
Carnitine	Plasma	0.5 mL	Frozen	Sub-Optimal	21 days	60 days	5-10 business days	\$125	82379	Testing is performed on Wednesdays.
Acylcarnitine	Plasma	0.5 mL	Frozen	Sub-Optimal	64 days	92 days		\$185	82139	Contact the laboratory for urgent testing.