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.

Peroxisomal Diseases Section: 443-923-2788

Test Name	sample type	volume	preferred specimen	stability whole blood ambient / refrigerated	stability plasma ambient	stability plasma refrigerated 4°C	stability plasma frozen - 20°C	stability urine frozen -20°C	cause for rejection	ТАТ	Cost	CPT Code	Comments:
plasma very long chain fatty acids (includes phytanic and pristanic acids) (screen for peroxisomal disorders)	EDTA/Heparin plasma or whole blood; fasting/ preprandial	1 mL plasma (min: 0.25mL); 1-3mL [‡] whole blood	frozen plasma or serum	48hr*	1 week	1 month	3 months; >1 year in Nalgene / Nunc freezing vial			7-10 business days	\$165	82726	† If RBC plasmalogen content and/or plasma pipecolic acid and/or very long chain fatty acids
red blood cell plasmalogen content (screen for rhizomelic chondrodysplasia punctata)	EDTA whole blood	3mL [‡] (min: 1.5mL)	nonfrozen whole blood only**	48-96hr*	NA	NA	NA		frozen; hemolysis	10-14 business days	\$165	82542	
plasma total lipid fatty acid profile (C14 to C26 saturated, mono&polyunsaturated, trans, branched chain fatty acids, triene/tetraene ratio. essential fatty acids)	EDTA plasma or whole blood; fasting/ preprandial	1 mL plasma (min: 0.25mL); 1-3mL whole blood	frozen plasma or serum	48hr*	24hr	48hr	3 months; >1 year in Nalgene / Nunc freezing vial		gross hemolysis or gross lipemia	7-10 business days	\$220	82542	
to C26 saturated, mono@nolyunsaturated fatty acids and	EDTA whole blood	3mL wh.blood (min: 1.5mL)	nonfrozen whole blood only**	48hr*	NA	NA	NA		frozen; hemolysis	10-14 business days	\$220		*Whole blood may be acceptable up to 4 days for fatty acid profiling or longer for other
plasma pipecolic acid	EDTA/Heparin plasma or whole blood; fasting/ preprandial	1 mL plasma (min: 0.25mL); 1-3mL [‡] whole blood	frozen plasma	48hr*	24hr	48hr	3 months; >1 year in Nalgene / Nunc freezing vial			14 business days	\$250		assays if no hemolysis, although within 48hr is ideal.
urine pipecolic acid	random urine, clean	5mL (min: 1mL)	frozen urine	NA	NA	NA	NA	1 year		14 business days	\$250	82542	**Frozen washed RBC may be accepted with prior arrangement
C26:0-Lysophosphatidylcholine (lysoPC or LPC)	DBS; EDTA/Heparin plasma or whole blood	3cm DBS; 0.1mL plasma 1-3mL [‡] whole blood	frozen plasma	1 month	1 month	6 months	3 months; >1 year in Nalgene / Nunc freezing vial	NA		14 business days	\$100	82542	