



## The Lantern Project Specimen Requirements Lighting the way to rare disease diagnosis



Test	Testing	Acceptable Sample Type	DBS Card	EDTA	Heparin	Saliva
ASM (Niemann Pick A/B)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
ABG (Gaucher)	Enzyme	Dried Blood Spot / Heparin*	Υ	N	Υ	N
GAA (Pompe)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
GLA (Fabry)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
IDUA (MPS-I)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
ID2S (MPS-II)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
NAGLU (MPS-IIIB)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
GALNS (MPS-IVA)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
b-GAL (MPS-IVB)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
ARSB (MPS- VI)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
GUSB (MPS-VII)	Enzyme	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
LysoGB-1	Biomarker	Dried Blood Spot / EDTA / Heparin*	Υ	Υ	Y*	N
LysoGB-3	Biomarker	Dried Blood Spot / EDTA	Υ	Υ	N	N
Single Gene/Multi-Gene	Sequencing	Dried Blood Spot / EDTA / Saliva+	Υ	Υ	N	Υ

<sup>\*</sup>Heparin is not preferred and will not allow for bundle testing if ordered.

<sup>+</sup>Saliva will not allow for additional enzyme or biomarker testing if ordered.

SAMPLE TYPES	CODE	REQUIREMENTS
DRIED BLOOD SPOTS PREFERRED SAMPLE TYPE	DBS	Collection Container(s): Dried blood spot card Collection and Processing Instructions: Follow kit instructions to fill all spots. Briefly, allow blood to saturate card until indicated areas are filled and blood has soaked through card. Air dry card at ambient temperature for at least 3 hours. Preferred Sample Condition: Follow kit instructions. Store at ambient temperature. Shipping Instructions: Follow kit instructions. Double bag and ship overnight at ambient temperature.
WHOLE BLOOD EDTA (purple top) Heparin (green top) HEPARIN CANNOT BE USED FOR ANY DNA TESTING OR LYSO-GB3	WB	Collection Container(s): EDTA (purple top), Heparin (green top) Collection and Processing Instructions: Infants (< 2-years): 2 to 3 mL; Children (>2-years): 3 to 5 mL; Older children and adults: 5 to 10 mL. Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze. Shipping Instructions: Ship overnight at ambient temperature ensuring receipt within 5-days of collection. Comments: Clotted or hemolyzed samples are not accepted.
SALIVA CANNOT BE USED FOR ENZYME ASSAY OR BIOMARKER TESTING	sv	Collection Container(s): Oragene™ Saliva Collection Kit Collection and Processing Instructions: Collect saliva on an Oragene™ Saliva Collection Kit according to the manufacturer's instructions. Please contact PKIG to request the saliva collection kit for patients that cannot provide a blood sample.  Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.  Shipping Instructions: Ship overnight at ambient temperature.

For samples that are collected not utilizing the PerkinElmer Genomics test packs, ship sample, test requisition form, and informed consent form by preferred shipping method to PerkinElmer Genomics at:

PerkinElmer Genetics 250 Industry Dr. Suite 400 Pittsburgh, PA 15275 For general questions on the collection and return of sample results, please call: PerkinElmer Genetics at +1 (866) 354-2910 (Monday-Friday, 8:00AM - 5:00PM EST) or by emailing Genomics@perkinelmer.com