



Division of Laboratory Services

630 Hart Lane
 Nashville, TN 37216
 615-262-6300

<https://www.tn.gov/newbornscreeninglab>

Newborn Screening Disorder:

Pompe Disease

Alternate Name(s)	<ul style="list-style-type: none"> • Acid maltase deficiency disease • Aglucosidase alfa • Alpha-1,4-glucosidase deficiency • Cardiomegalia glycogenica diffusa • Deficiency of alpha-glucosidase • GSD II • Deficiency of lysosomal alpha-glucosidase
Analyte(s) Tested	<ul style="list-style-type: none"> • Acid alpha-glucosidase
Methodology	Tandem Mass Spectrometry
TDH Requisition Form	<ul style="list-style-type: none"> • PH-1582 • Form Requests: Contact state lab by email or fax. Include facility name, address, phone number and contact person on your request. Fax: (615) 262-6455 Email: DCLAB.supply@tn.gov
Acceptable Specimen	Dried blood spots on filter paper collected from infants less than 6 months of age
Collection Information	<ul style="list-style-type: none"> • Optimal specimen: Collect at 24 hours + 1 minute of life • Acceptable specimen: Collect 24-48 hours of life • If transfused: Recollect 4 days post transfusion
Shipping Information	<ul style="list-style-type: none"> • Health Departments and Birthing Hospitals • Private Clinics and Midwives
Screening Results	<ul style="list-style-type: none"> • Tennessee Newborn Screening's Secure Remote Viewer (SRV) <ul style="list-style-type: none"> ○ Healthcare providers must be registered to view and print patient result reports. To Register: Complete and submit SRV Access Form (PH-3909). • NBS List of Screened Disorders and Mailer Explanations
Laboratory Location Performing Testing	Nashville, TN