

**Date:** April 24, 2014

**To:** Edmonton Zone, North Zone and Central Zone  
Physicians, Nurse Managers, Laboratory Directors and Managers

**From:** AHS Laboratory Services – University of Alberta Hospital (UAH)

**Re:** Lysosomal Storage Disorder Enzyme Testing Temporarily Unavailable at the UAH Laboratory

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## PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

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### Key Messages:

- Due to instrument failure, lysosomal storage disorder enzyme activity testing for Tay-Sachs, Sandhoff, Fabry, Gaucher, GM1 Gangliosidosis, and Pompe performed at the UAH Newborn Screening & Biochemical Genetics Lab in Edmonton will be **temporarily** unavailable.
- This change is effective immediately.
- In the interim, samples collected for serum hexosaminidase will be referred out to the Alberta Children's Hospital Biochemical Genetics Laboratory in Calgary. Enzyme testing for Fabry (alpha galactosidase), Gaucher (beta glucosidase), GM1 Gangliosidosis (beta galactosidase), and Pompe disease (alpha glucosidase) will be referred out to a reference lab in Ontario.
- Interpretive reports from the referral laboratories including reference intervals will be mailed to the ordering and "copy to" physicians. Turnaround times are expected to be within 1 month.

### Why this is important:

- Lysosomal enzyme activity assays help establish the diagnosis of lysosomal storage disorders.
- Sample collection and handling requirements will change to accommodate the needs of the referral laboratories performing these tests.
- Appropriate specimen collection and handling is crucial in ensuring optimal specimen volume, quality specimens, and timely reporting of accurate laboratory test results.

### Action Required:

If Clinical Suspicion for	Indicate on Requisition/Card	Provide
Tay-Sachs or Sandhoff Disease	Hexosaminidase	Detailed patient history
Pompe Disease	Pompe Disease	Patient with instructions to have bloodspot collection done between Monday and Wednesday
Fabry Disease Gaucher Disease GM1 Gangliosidosis	Alpha galactosidase Beta glucosidase Beta galactosidase	Detailed patient history Patient with instructions to have blood collection done between Monday and Friday before noon

**Additional information:**

- Alberta Health Services Edmonton Zone Test Directory

**For questions and feedback contact:**

- Edmonton Client Response Centre at :780-407-7484
- Dr. Kareena Schnabl, Clinical Biochemist, UAH Laboratory at: 780-407-3186.

**This bulletin has been reviewed and approved by:**

Dr. Carolyn O'Hara, Medical/Scientific Director and Zone Clinical Department Head, AHS Laboratory Services, Edmonton Zone