

Date: October 18, 2016
To: All Zones, Physicians, Nursing Managers/Educators, and Gastrointestinal and Metabolic Clinics
From: Genetic Laboratory Services (GLS)
Re: **Galactosemia Testing in Alberta**

PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

Key Messages:

To ensure accurate and timely screening, confirmatory diagnosis, and dietary monitoring for patients with or suspected of having classical galactosemia, please see the ordering information outlined in this bulletin. Appropriate test selection will minimize unnecessary recollection on pediatric patients, and prevent family anxiety and delays in testing.

Action Required:

- Provide patient history – indicate whether test request is for screening, confirmation, or dietary monitoring. Also indicate if patient has received a recent blood transfusion.
- Spell out the name of the test (avoid using test acronyms).
- For galactosemia screening, write '*Galactosemia screen*' on the dried blood spot card (NMS use only box).
- For more information regarding each test, please contact the appropriate laboratory:
 - 780-407-7907 for galactosemia screening and monitoring in Edmonton and North Zones
 - 403-770-3600 for galactosemia screening and monitoring in Calgary, Central and South Zones
 - 403-955-7379 for confirmatory enzyme testing (galactose-1-phosphate uridyl transferase) in all Zones

Specimen Requirements:

| Clinical Indication | Test Name | Sample Type | Volume | Handling/Transport |
|---|--|--------------------------------|------------------------------|--|
| Screening – Initial investigation for galactosemia | Galactosemia screen | Dried blood spots | 2 full circles | Room temperature |
| Confirmatory enzyme test - For positive galactosemia screens | Galactose-1-phosphate uridyl transferase | Whole blood (Li or Na heparin) | 3 mL | On ice immediately, NO FREEZING |
| Dietary monitoring - Known galactosemia patients | Galactose-1-phosphate, quantitation | Whole blood (Li or Na heparin) | Pediatric 1.5mL Adult 2mL | FREEZE immediately, DO NOT SEPARATE |

Additional Information

- AHS Edmonton and Calgary, DynaLIFE Dx and CLS test directories
- Edmonton Client Response: 780-407-7484

Inquiries and feedback may be directed to:

- Biochemical Genetics Lab, Edmonton, Genetic Counselor, UAH at 780-407-7907
- Calgary Laboratory Services, Clinical Biochemist, ACH at 403-955-2277
- Biochemical Genetics Lab, Calgary, Biochemical Geneticist, ACH at 403-955-7379

This bulletin has been reviewed and approved by:

Dr. Martin Somerville, Medical / Scientific Director, GLS
 Dr. Carolyn O'Hara, Interim Medical Director, Laboratory Services