

TESTING RESUMED

NOTIFICATION DATE: April 17, 2014 **EFFECTIVE DATE:** Immediately

GALACTOSEMIA REFLEX, BLOOD Test ID: GCT

EXPLANATION: Test ID GALT, a component of test ID GCT, will be immediately orderable using an improved method. Please note the changes to acceptable specimen types, reference ranges, and methodology listed below.

TESTING ALGORITHM

Testing begins with galactose-1-phosphate uridyltransferase (GALT) enzyme analysis. If GALT is > or =24.5 nmol/h/mg of hemoglobin, testing is complete. No molecular test will be performed. If GALT is <24.5 nmol/h/mg of hemoglobin, GAL6 / Galactosemia Gene Analysis (6-Mutation Panel) will be performed at an additional charge. See Galactosemia Testing Algorithm in Special Instructions.

PREVIOUS METHODOLOGY

GALT: Ultraviolet, Kinetic

GAL6: A real-time polymerase chain reaction (PCR)-based assay is utilized to examine DNA for 6 alterations.

NEW METHODOLOGY

GALT: Enzyme Reaction Followed by Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

GAL6: A real-time polymerase chain reaction (PCR)-based assay is utilized to examine DNA for 6 alterations.

PREVIOUS SPECIMEN REQUIREMENTS

Container/Tube: Lavender top (EDTA)

Specimen Volume: 5 mL

Additional Information: Patient's age is required.

1. New York Clients-Informed consent is required.

Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

2. If ordering electronically, submit a Biochemical Genetics Request Form (Supply T439) with the specimen.

NEW SPECIMEN REQUIREMENTS

Container/Tube:

Preferred: Lavender top (EDTA) **Acceptable**: Yellow top (ACD) **Specimen Volume**: 5 mL

Additional Information: Patient's age is required.

Forms:

 $1. \ New \ York \ Clients-Informed \ consent \ is \ required.$

Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special

Instructions.

2. If ordering electronically, submit a Biochemical Genetics Request Form (Supply T439) with the specimen.

CURRENT REFERENCE VALUES

> or =18.5 U/g of hemoglobin

CURRENT RESULTABLES		
Result ID	Result Name	
8333	Gal-1-P Uridyltransferase, RBC	
2296	Comment	

NEW REFERENCE VALUES > or =24.5 nmol/h/mg of hemoglobin

NEW RESULTABLES		
Result ID	Result Name	
8333	Gal-1-P Uridyltransferase, RBC	
2296	Comment	
58115	Reviewed By	