

Test Definition: GALTP

Galactose-1-Phosphate Uridyltransferase Biochemical Phenotyping, Erythrocytes

Reporting Title: Gal-1-Phos Urdyltrns Phenotype, RBC

Performing Location: Rochester

Ordering Guidance:

The preferred test to evaluate for possible diagnosis of galactosemia, routine carrier screening, and follow-up of abnormal newborn screening results is GCT / Galactosemia Reflex, Blood.

For monitoring of dietary compliance, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

Necessary Information:

Patient's age is required.

A quantitative galactose-1-phosphate uridyltransferase level (GALT / Galactose-1-Phosphate Uridyltransferase, Blood) is required for accurate interpretation.

<u>Biochemical Genetics Patient Information</u> (T602) is recommended, but not required, to be filled out and sent with the specimen to aid in the interpretation of test results.

Specimen Requirements:

Multiple whole blood tests for galactosemia can be performed on 1 specimen. Prioritize order of testing when submitting specimens. For a list of tests that can be ordered together see <u>Galactosemia-Related Test List</u>.

Container/Tube: Lavender top (EDTA)

Specimen Volume: 3 mL

Forms:

- 1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
- -Informed Consent for Genetic Testing (T576)
- -Informed Consent for Genetic Testing-Spanish (T826)
- 2. <u>Biochemical Genetics Patient Information</u> (T602) is recommended.
- 3. If not ordering electronically, complete, print, and send a <u>Biochemical Genetics Test Request</u> (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Refrigerated (preferred)	28 days	
	Ambient	14 days	

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
80341	Gal-1-Phos Urdyltrns Phenotype,RBC	Alphanumeric		33780-8
34524	Reviewed By	Alphanumeric		18771-6



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 $\ensuremath{\mathsf{LOINC}}\xspace^{\ensuremath{\texttt{@}}}$ and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82664

82775

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
GALT	Gal-1-P Uridyltransferase, RBC	1	82775	Yes	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
GALT	8333	Gal-1-P Uridyltransferase, RBC	Numeric	nmol/h/mg Hb	24082-0
GALT	2296	Interpretation (GALT)	Alphanumeric		59462-2
GALT	58115	Reviewed By	Alphanumeric		18771-6

Reference Values:

An interpretative report will be provided.