Galactosemia: GALT Gene Deletion/Duplication

**Test Code:** DGALT  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Galactosemia is an autosomal recessive disorder of galactose metabolism that often presents in the newborn period with poor suck, vomiting, diarrhea, bleeding diathesis, lethargy, jaundice, and sepsis. If left untreated, an individual may progress to irreversible liver disease and mental retardation. The prevalence of galactosemia is approximately 1 in 30,000 newborns. Most often galactosemia is caused by a deficiency of the galactose-1-phosphate uridylytransferase (GALT) enzyme, which catalyzes the conversion of the galactose-1-phosphate to glucose-1-phosphate. This is a critical step in the metabolism of galactose to glucose.

The GALT gene is encoded by 11 exons on chromosome 9p13. Galactosemia caused by mutations in the GALT gene is inherited in an autosomal recessive manner.

Family members of individuals with galactosemia are at risk to be carriers of the disorder. Biochemical and molecular screening is available to family members or partners of individuals with galactosemia using a combination of mutation analysis and enzyme activity.

Refer to the comprehensive galactosemia panel (GS) for a disease overview. Refer to the galactosemia carrier panel (GR).


### References:

- GeneReviews Clinical Summary

### Genes

**GALT**

### Indications

This test is indicated for individuals with biochemical diagnosis of a galactosemia or carrier status and a negative or unknown result from mutation analysis for common GALT gene mutations. This test is **NOT** recommended for galactosemia screening or diagnosis in persons with a positive newborn screen. Please refer to the Comprehensive Galactosemia Panel.

### Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient’s phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### Detection

Detection is limited to duplications and deletions. Array CGH will not detect point mutations or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

#### Type: Whole Blood

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:  
Infants (2 years): 3-5 ml  
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

#### Type: Saliva

Specimen Requirements:
Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Special Instructions**

Submit copies of diagnostic biochemical test results with the sample. Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.

**Related Tests**

- [Comprehensive GALT Analysis](#) is the standard galactosemia test offered by EGL and includes GALT enzyme activity, isozyme pattern (phenotyping), and gal-1-p level.
- [Carrier Testing for Galactosemia](#) is accomplished by looking at GALT and isozyme.
- Prenatal testing may be available to couples who are confirmed carriers of galactosemia by molecular and biochemical analysis. Please contact the laboratory genetic counselor to determine the availability of prenatal testing.