Galactosemia (Epimerase): **GALE Gene Deletion/Duplication**

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

### Condition Description

UDP-Galactose-4-Epimerase (GALE) deficiency is one of the three known forms of galactosemia, along with galactose-1-phosphate uridylyltransferase (GALT) deficiency (classic galactosemia) and galactokinase (GALK) deficiency [1-2]. GALE deficiency has been defined as a continuous disorder with a spectrum of enzyme impairment and corresponding metabolic compromise impacting a variety of tissues in affected individuals [3]. The patients may present with symptoms reminiscent of classic galactosemia and demonstrate severely impaired GALE activity in both RBCs and lymphoblast resulting in accumulation of abnormally high levels of RBC galactose, gal-1P and UDP-gal. GALE deficiency should be considered in individuals with elevated RBC galactose, gal-1-P and UDP-gal but normal GALT enzyme activity. Mutations in the GALE gene, located on 1p36, are associated with a biochemical diagnosis of GALE deficiency [3]. Gene sequence analysis is available to test for mutations in the GALE gene in patients with a biochemical diagnosis of GALE deficiency (JU). Click here for the GeneReviews summary on this condition. Also, refer to the Comprehensive Galactosemia Panel for a disease overview.


### References:

### Genes

**GALE**

### Indications

This test is indicated for:
- Individuals with elevated gal-1-P and UDP-gal but with normal GALT and GALK enzyme activity and biochemical findings consistent with GALE deficiency  
- Carrier testing for individuals with a family history of GALE deficiency

### 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) 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 of EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.

Click here for the Epimerase Deficiency Clinical Information Form to send with the sample.

**Related Tests**

- Sequencing of GALT and GALK Genes for transferase deficient and galactokinase deficient galactosemia
- Comprehensive Galactosemia Panel includes: GALT enzyme activity, isozyme pattern, gal-1-P concentration, GALT common mutation panel
- Urine Galactitol Concentration
- Custom Diagnostic Mutation Analysis (KM) is available to family members if mutations are identified by sequencing.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.