Galactosemia (Galactokinase Deficiency): \textit{GALK1} Gene Deletion/Duplication

Test Code: JA  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1

Condition Description

Galactokinase (GALK) deficiency is one of the three known forms of galactosemia, along with galactose-1-phosphate uridylyltransferase (GALT) deficiency (classic galactosemia) and UDP-galactose-4'-epimerase (GALE) deficiency [1-2]. GALK deficiency is an autosomal recessive disorder characterized by an elevation of blood galactose concentration and diminished galactose-1-phosphate concentration, leading to production of alternative metabolic products such as galactitol [3]. Galactokinase-deficiency may present in the neonatal period with cataracts; no other clinical complications have been consistently associated with GALK-deficiency [2].

GALK deficiency should be considered in individuals with cataracts, elevated red cell galactose, galactosuria, or elevated urinary galactitol and normal GALT enzyme activity. GALK activity is used to rule-out variant galactosemia due to galactokinase deficiency which should not be confused with classical galactosemia secondary to GALT deficiency, or epimerase-deficiency galactosemia secondary to GALE deficiency. The vast majority of patients with biochemical diagnosis of GALK deficiency have mutations in the \textit{GALK1} (17q25) gene [4-6]. Gene sequence analysis is available to test for mutations in the \textit{GALK1} gene in patients with a biochemical diagnosis of GALK deficiency (IQ).


Click [here](http://www.ThinkGenetic.com) for the GeneReviews summary on this condition. Also, refer to the Comprehensive Galactosemia Panel for a disease overview.

References:
4. Sanguolo et al. Biochemical Characterization of Two \textit{GALK1} Mutations in Patients with Galaktokinase Deficiency Hum Mutat 2004 Apr;23(4):396-403
5. Park et al. Molecular and biochemical characterization of the \textit{GALK1} gene in Korean patients with galactokinase deficiency. Mol Genet Metab. 2007. 91:234-8

Genes

\textit{GALK1}

Indications

This test is indicated for:

- Individuals with elevated blood galactose but with normal GALT and GALE enzyme activities.
- Carrier testing for individuals with a family history of GALK deficiency.

Methodology

Detection is limited to duplications and deletions. Array CGH will not detect point mutations or intronic mutations.

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.

Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

Type: Whole Blood

Specimen Requirements:

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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.

### Related Tests

- **GALT and GALE Gene Sequencing** for transferase deficient and epimerase deficient galactosemia
- **Comprehensive Galactosemia Panel** includes: GALT enzyme activity, isozyme pattern, gal-1-P concentration
- **Urine Galactitol Concentration**
- Custom Diagnostic Mutation Analysis (KM) is available to family members if mutations are identified by sequencing.
- Prenatal testing is available for known familial mutations only. Please call the Laboratory Genetic Counselor before collecting a fetal sample.