TEST ID: GALE
UDP-GALACTOSE 4’ EPIMERASE (GALE), BLOOD

USEFUL FOR
Diagnosis of UDP-galactose 4’ epimerase deficiency.

GENETICS TEST INFORMATION
Enzyme testing only.

CLINICAL INFORMATION
Galactosemia is an autosomal recessive disorder that results from a deficiency of 1 of the 3 enzymes catalyzing the conversion of galactose to glucose: galactose-1-phosphate uridyltransferase (GALT), galactokinase (GALK), and uridine diphosphate galactose-4-epimerase (GALE). Epimerase deficiency galactosemia can be categorized into 3 types: generalized, peripheral, and intermediate. Generalized epimerase deficiency galactosemia results in profoundly decreased enzyme activity in all tissues, whereas peripheral epimerase deficiency galactosemia results in decreased enzyme activity in red and white blood cells, but normal enzyme activity in all other tissues. This is compared to intermediate epimerase deficiency galactosemia which results in decreased enzyme activity in red and white blood cells and less than 50% of normal enzyme levels in other tissues.

Clinically, infants with generalized epimerase deficiency galactosemia develop symptoms such as liver and renal dysfunction and mild cataracts when on a normal milk diet, while infants with peripheral or intermediate epimerase deficiency galactosemia do not develop any symptoms. Generalized epimerase deficiency galactosemia is treated by a galactose- and lactose-restricted diet, which can improve or prevent the symptoms of renal and liver dysfunction and mild cataracts. Despite adequate treatment from an early age, individuals with generalized epimerase deficiency galactosemia remain at increased risk for developmental delay and intellectual disability. Unlike patients with classic galactosemia resulting from a GALT deficiency, females with generalized epimerase deficiency galactosemia experience normal puberty and are not at increased risk for premature ovarian failure. Based upon reports by newborn screening programs, the frequency of epimerase deficiency galactosemia in the United States ranges from approximately 1 in 6,700 in African American infants to 1 in 70,000 infants of European ancestry.

Galactose-1-phosphate (Gal-1-P) accumulates in the erythrocytes of patients with galactosemia due to either GALT or GALE deficiency. The quantitative measurement of Gal-1-P (GAL1P / Galactose-1-Phosphate (Gal-1-P), Erythrocytes) is useful for monitoring compliance with dietary therapy. Gal-1-P is thought to be the causative factor for development of liver disease in these patients and, because of this, patients should maintain low levels and be monitored on a regular basis.

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
Newborn screening, which identifies potentially affected individuals by measuring total galactose (galactose and Gal-1-P) and/or determining the activity of the GALT enzyme, varies from state to state. The diagnosis of galactosemia is established by follow-up quantitative measurement of GALT enzyme activity. If enzyme levels are normal, but an infant has an elevated Gal-1-P, then epimerase deficiency galactosemia is to be considered. Molecular testing via sequencing of the GALE gene may be performed.

See Galactosemia Testing Algorithm in Special Instructions for additional information.

**INTERPRETATION**

An interpretive report will be provided.

See Galactosemia Testing Algorithm in Special Instructions for additional information. For galactokinase deficiency, see GALK / Galactokinase, Blood.

For galactose-1-phosphate uridylyltransferase deficiency, see GALT / Galactose-1-Phosphate Uridylyltransferase, Blood.

**CLINICAL REFERENCE**

