

Using Genetic Testing for Diagnosis of Galactosemia

Indications:

- Positive newborn screening test for galactosemia
- Known familial mutations in *GALT*

Benefits:

Genetic testing for galactosemia

- Can confirm a clinical or biochemical diagnosis of galactosemia and identify the familial mutation(s)
- Can inform prognosis based on nature of familial mutations
- Can identify carriers among family members of patients and inform genetic counseling

Background:

- Galactosemia is an autosomal recessive disorder of galactose metabolism that often results in neonatal death or, if the patient survives the neonatal period, mental retardation.^{1,2}
- Prompt removal of lactose and galactose from the diet can resolve acute neonatal symptoms and prevent death and mental retardation.^{1,2}
- Most cases of galactosemia, which has a prevalence of about 1 in 30,000, are detected through newborn screening of galactose-1-phosphate uridylyltransferase (*GALT*) enzyme activity and/or galactose-1 phosphate concentration in red blood cells.^{1,2}
- Even with timely dietary treatment initiation, long-term consequences are common and can include verbal dyspraxia, delay of mental development, motor dysfunction, poor growth, cataracts, and, in females, ovarian failure.^{1,2}
- Prognosis may differ depending on the nature of the specific *GALT* mutations underlying the disease.¹⁻³
- Genetic testing can identify the *GALT* mutations in a patient and distinguish between the Duarte and the Los Angeles allele, which have very different prognosis but cannot be differentiated by commonly used techniques such as isoelectric focusing.¹

References: 1. Elsas LJ GeneReviews.
<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=galactosemia> Accessed 08/11/10.
2. Bosch AM (2006) *Inherit Metab Dis* 29:516-25. 3. Tyfield L, et al (1999) *Human Mutation* 13:417-30.

Ordering Information: Please see other side.

Ordering Information for Galactosemia Testing

Indications for Testing

- Positive newborn screening test for galactosemia
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Ordering Information

Gene(s)	Test Code
<i>GALT</i>	252816

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.