**GALT Gene Sequencing Assay (Galactosemia): Test Ordering Information**

**Test Information:**
GALT Gene Sequencing Assay (Galactosemia) (CMGDL test code 3002)

For additional information please refer to the CMGDL [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu).

**Indications for Testing**
- Individuals with a biochemical testing confirmed diagnosis of galactosemia or abnormal biochemical testing with neither or only one disease-causing mutation detected by targeted mutation analysis. In these individuals gene sequencing may help in identifying mutations not included in the targeted mutation analysis panel.

**Contraindications**
- Testing should not be ordered for individuals with previously identified familial GALT mutations. To test for a specific mutation, it is recommended to order Family Testing, Gene Sequencing Targeted Mutation Analysis (CMGDL test code 3000) and provide a copy of the laboratory report stating the familial mutation.
- Prenatal testing is not performed by the CMGDL.

**Specimen Requirements**
2.0 ml of whole blood (minimum)
Collection Tube: Lavender Top Tube /w EDTA
Handling: Room Temp- specimen processed within 72 hours

**Turn-Around Times**
21 to 28 business days
(see [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu) for eventual TAT changes or updates)

**Methodology and Assay Characteristics**
This test is performed by using the Sanger’s method, which is also referred to as dideoxy sequencing or chain termination sequencing. Briefly, genomic DNA segments of 100-500 nucleotides (in general containing the gene exons of interest plus an additional 20 base pairs from the exon/intron junction into the intron) are enriched by polymerase chain reaction (PCR). The products of this amplification reaction are then sequenced bi-directionally.

**Analytical Sensitivity and Specificity**
99 percent of analytical sensitivity and specificity for the sequenced gene regions (see Test Limitations)

**Test Limitations**
(see [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu) for additional information)
- Gene sequencing by Sanger’s method cannot detect large deletions as well as it cannot detect other genomic rearrangements;
- Gene sequencing tests may reveal or suggest non-paternity in certain cases;
- This method is affected by allele-dropout, although this phenomenon is minimized by the selection of primer binding sites not containing known variants. If the allele dropout occurs, only one allele of two will be amplified: in this case eventual variants on the dropped allele will not be detected and eventual variants located on the amplified allele may be falsely detected as homozygous;
- This test is designed to detect all the gene exons and around 20 nucleotides into the intronic regions: mutations that occur outside of these regions would not be detected;
- Clinical molecular genetics is a fast-moving field. The gene variant interpretation may change as new medical and scientific information becomes available;
- Results of this test should be interpreted in the context of the clinical presentation and in consultation with a clinical geneticist.

This test was developed and its performance determined by the CMGDL. The CMGDL has clinically validated it and determined its accuracy and precision. It has not been cleared or approved by the U.S. Food and Drug Administration. This test is used for clinical purposes. Pursuant to the requirements of CLIA ’88, this laboratory has verified the test accuracy and precision. Genetic testing using the methods applied by the CMGDL is expected to be highly accurate. However, the chance of a false positive or a false negative result, due to laboratory errors incurred during any phase of the testing, cannot be completely excluded.

**Clinical Presentation, Diagnosis and Medical Genetics of Galactosemia** (Refer to GeneReviews Galactosemia [http://www.ncbi.nlm.nih.gov/books/NBK1518/](http://www.ncbi.nlm.nih.gov/books/NBK1518/) and OMIM 230400)
References

References
1. OMIM 230400
2. GeneReviews Galactosemia

Related Tests
- Family Testing, Gene Sequencing Targeted Mutation Analysis (test code 3000)
(visit our website at www.medgen.med.miami.edu)

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