Galactosemia Mutation Panel

Test Code	D0405
Test Summary	This test analyzes 5 common mutations associated with Galactosemia.
Turn-Around-Time (TAT)*	10 - 12 days
Acceptable Sample Types	Dried Blood Spots
Acceptable Billing Types	Self (patient) Payment Institutional Billing Commercial Insurance

Indications for Testing

This test may be appropriate for individuals with a clinical history consistent with galactosemia.

Test Description

This panel analyzes the N314D (Duarte), Q188R, S135L, K285N, and L195P (Classical) variants in the *GALT* gene associated with galactosemia.

Condition Description

Galactosemia is a condition that affects how the body processes a simple sugar called galactose. Galactose is present in many foods and is primarily a part of a larger sugar called lactose found in all dairy products and many baby formulas. The symptoms of galactosemia result from the body's inability to use galactose to produce energy. (NIH, genetics home reference)

Test Methods and Limitations

Gene analysis for the various targeted mutations is performed by polymerase chain reaction and melting curve analysis to detect the mutant and wild-type forms of the gene. Sequence-specific oligonucleotide probes are labeled with fluorescent dyes, which hybridize to their complementary sequence target in PCR products. The fluorescence resonance energy transfer (FRET) from one fluorophore to another adjacent fluorophore is measurable and is directly proportional to the amount of target DNA generated during PCR. Allele-specific melting curves are generated by slow thermal denaturing of the probe: template hybrid. Melting curves are generated by monitoring fluorescence throughout denaturation, and melting peaks are generated by plotting the inverse derivative of fluorescence versus temperature (?dF/dT).

Detailed Sample Requirements

Dried Blood Spots