Glutaric Acidemia Type I Mutation Panel

Test Code	D0406
Test Summary	This test analyzes 2 variants of the GCDH gene
Turn-Around-Time (TAT)*	10 - 12 days
Acceptable Sample Types	Dried Blood Spots (Preferred sample type)
Acceptable Billing Types	Self (patient) Payment Institutional Billing Commercial Insurance

Indications for Testing

- Individuals with symptoms of intellectual disability, large heads, spasms, hypotonia, jerky movement, bleeding in the brain and eyes, organic acidemia, organic aciduria, and damage to the basal ganglia
- Individuals with a family history of glutaric aciduria type I

Test Description

This test detects the A421V and R402W variants of the *GCDH* gene associated with glutaric acidemia type I

Condition Description

Glutaric acidemia type I is a disease that causes problems breaking down the amino acids lysine, hydroxylysine, and tryptophan. The age of onset and symptoms are variable and can include intellectual disability, large heads, spasms, hypotonia, jerky movement, bleeding in the brain and eyes, organic acidemia, organic aciduria, and damage to the basal ganglia. The disease can be managed with a strict diet. The incidence of glutaric aciduria type I is estimated to be ~ 1 in 100,000.

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 (Preferred sample type)