Methylmalonic Acidemia Mutation Panel

Test Code	D0411
Test Summary	This test analyzes 2 variants of the MUT 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

- Infants with symptoms of vomiting, dehydration, hypotonia, developmental delay, lethargy, hepatomegaly, failure to thrive, feeding problems, intellectual disability, chronic kidney disease, pancreatitis, coma, and early death
- Individuals with a family history of methylmalonic aciduria

Test Description

This test detects the N219Y and G717V variants of the *MUT* gene associated with methylmalonic acidemia

Condition Description

Methylmalonic aciduria is a disease that prevents the body from processing certain proteins and fats. The age of onset is typically in early infancy, and variable symptoms include vomiting, dehydration, hypotonia, developmental delay, lethargy, hepatomegaly, failure to thrive, feeding problems, intellectual disability, chronic kidney disease, pancreatitis, coma, and early death. The incidence of methylmalonic aciduria is estimated to be ~ 1 in 50,000.

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)