






## 3-MCC Deficiency Mutation Panel

	<b>Test Code</b>	D0410
	<b>Test Summary</b>	This test analyzes 1 variant of the MCCC2 gene
	<b>Turn-Around-Time (TAT)*</b>	10 - 12 days
	<b>Acceptable Sample Types</b>	Dried Blood Spots
	<b>Acceptable Billing Types</b>	Self (patient) Payment Institutional Billing Commercial Insurance

### Indications for Testing

- Individuals with symptoms of feeding difficulties, vomiting, diarrhea, lethargy, hypotonia, developmental delay, seizures, and coma
- Individuals with a family history of 3-Methylcrotonyl-CoA carboxylase deficiency

### Test Description

This test detects the 518insT variant of the *MCCC2* gene associated with 3-MCC deficiency type 2

### Condition Description

3-Methylcrotonyl-CoA carboxylase deficiency is a disease that prevents the breakdown of the amino acid leucine. Age of onset is typically in infancy or early childhood, and symptoms include feeding difficulties, vomiting, diarrhea, lethargy, hypotonia, developmental delay, seizures, and coma. A strict diet can manage the disease. The incidence of 3-Methylcrotonyl-CoA carboxylase deficiency is estimated to be ~1 in 36,000.

### 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