






## Biotinidase Deficiency Mutation Panel

	<b>Test Code</b>	D0402
	<b>Test Summary</b>	This test analyzes 12 variants in the BTB 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

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

### Test Description

This test analyzes the G98:d7i3, Q456H, R157H, R538C, D252G, and D444H; D444H; A171T, D444H; F403V, D444H; and R157H variants in the *BTB* gene

### Condition Description

Biotinidase deficiency is an inherited disorder in which the body cannot recycle the vitamin biotin. Without treatment, signs and symptoms appear within the first few months of life. Profound biotinidase deficiency can cause seizures, weak muscle tone, breathing problems, hearing and vision loss, problems with movement and balance, skin rashes, hair loss, a fungal infection called candidiasis, and delayed development. Biotinidase deficiency occurs in approximately 1 in 60,000 newborns. (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