Biotinidase Deficiency Enzyme Analysis (Complete/Partial)

Test Code	B0001
Test Summary	This test measures biotinidase to aid in the diagnosis of biotinidase deficiency.
Turn-Around-Time (TAT)*	3 days
Acceptable Sample Types	Dried Blood Spots
Acceptable Billing Types	Self (patient) Payment Institutional Billing

Indications for Testing

This test may be used to aid in the diagnosis of biotinidase deficiency. This test may be appropriate for individuals with a clinical history concerning for biotinidase deficiency or to confirm a diagnosis after a positive newborn screening result.

Test Description

Decreases biotinidase enzyme activity is associated with biotinidase deficiency.

Condition Description

Biotinidase deficiency is an inherited disorder in which the body is unable to 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

Colorimetry is used to measure certain analytes in solution, if the analyte is known to produce a measurable color change in reaction to certain reagents. The Colorimeter instrument applies a light of known wavelength to the solutions and measures the absorbance through the solution. The analyte's concentration is determinable by the use of known concentrated standards to calibrate the test, and then measuring the analyte's color change in reference to the standards.

Detailed Sample Requirements

Dried Blood Spots