Overview

Useful For
Preferred test for diagnosing biotinidase deficiency
Follow-up testing for certain organic acidurias

Genetics Test Information
Preferred test to rule-out biotinidase deficiency.
Second-tier molecular testing is available, see BTDZ / Biotinidase Deficiency, BTD Full Gene Analysis.

Highlights
Enzymatic testing for the diagnosis of biotinidase deficiency, usually in follow-up to an abnormal newborn screen.
Biotinidase deficiency is treatable with oral biotin supplements.
Individuals who are diagnosed presymptomatically (eg, by newborn screening) and who are treated with biotin supplementation do not develop the associated clinical features of biotinidase deficiency.

Special Instructions
- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Informed Consent for Genetic Testing (Spanish)

Method Name
Colorimetric

NY State Available
Yes

Specimen

Specimen Type
Serum

Specimen Required
Collection Container/Tube:

Preferred: Red top
Acceptable: Serum gel
Submission Container/Tube: Plastic vial

Specimen Volume: 1 mL
Collection Instructions: Spin down immediately and remove serum.
Test Definition: BIOTS
Biotinidase, S

Forms
1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:
   - **Informed Consent for Genetic Testing** (T576)
   - **Informed Consent for Genetic Testing-Spanish** (T826)
2. **Biochemical Genetics Patient Information** (T602) in Special Instructions.
3. If not ordering electronically, complete, print, and send an **Inborn Errors of Metabolism Test Request** (T798) with the specimen.

Specimen Minimum Volume
0.5 mL

Reject Due To

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<tr>
<td>Gross lipemia</td>
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<td>Gross icterus</td>
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Specimen Stability Information

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Clinical and Interpretive

Clinical Information

Biotinidase deficiency is an autosomal recessive disorder caused by mutations in the biotinidase gene (*BTD*). Age of onset and clinical phenotype vary among individuals depending on the amount of residual biotinidase activity. Profound biotinidase deficiency occurs in approximately 1 in 137,000 live births and partial biotinidase deficiency occurs in approximately 1 in 110,000 live births, resulting in a combined incidence of about 1 in 61,000. The carrier frequency for biotinidase deficiency within the general population is about 1 in 120.

Untreated profound biotinidase deficiency typically manifests within the first decade of life as seizures, ataxia, developmental delay, hypotonia, sensorineural hearing loss, vision problems, skin rash, and alopecia. Partial biotinidase deficiency is associated with a milder clinical presentation, which may include cutaneous symptoms without neurologic involvement. Certain organic acidurias, such as holocarboxylase synthase deficiency, isolated carboxylase synthase deficiency, and 3-methylcrotonylglycinuria, present similarly to biotinidase deficiency. Serum biotinidase levels can help rule out these disorders.

Treatment with biotin is successful in preventing the clinical features associated with biotinidase deficiency. In symptomatic patients, treatment will reverse many of the clinical features except developmental delay, vision, and...
hearing complications. As a result, biotinidase deficiency is included in most newborn screening programs. This enables early identification and treatment of presymptomatic patients.

Molecular tests form the basis of confirmatory or carrier testing. When biotinidase enzyme activity is deficient, sequencing of the entire BTD gene (BTDZ / Biotinidase Deficiency, BTD Full Gene Analysis) allows for detection of disease-causing mutations in affected patients. Identification of familial mutations allows for testing of at-risk family members (FMTT / Familial Mutation, Targeted Testing).

While genotype-phenotype correlations are not well established, it appears that certain mutations are associated with profound biotinidase deficiency, while others are associated with partial deficiency.

**Reference Values**
3.5-13.8 U/L

**Interpretation**
The reference range is 3.5 U/L to 13.8 U/L.

Partial deficiencies and carriers may occur at the low end of the reference range.

Values below 3.5 U/L are occasionally seen in specimens from unaffected patients.

**Cautions**
A diet high in biotin may result in normal clinical presentation even when the biotinidase level is low.

**Clinical Reference**


**Performance**

**Method Description**

**PDF Report**
No

**Day(s) and Time(s) Test Performed**
Monday, Thursday; set up at 8 a.m.

**Analytic Time**
4 days
Maximum Laboratory Time
8 days

Specimen Retention Time
30 days

Performing Laboratory Location
Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

82261

LOINC® Information

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