

Biotinidase Deficiency

Test code

70132X [Biotinidase Activity]
 16526X [Mutation Analysis]
 16537X [Biotinidase Activity w/rfl Mutation Analysis]

Specimen Requirements

70132X: 2 mL frozen serum (red-top serum separator tube [SST]); 1 mL minimum
 Alternatively, submit 2 mL frozen plasma (EDTA, lavender-top tube)
 Separate serum or plasma within 1 hour of collection

16526X: 5 mL room temperature whole blood (EDTA, lavender-top tube); 3 mL minimum

16537X: Serum and whole blood; see individual assays

CPT Codes*

70132X: 82261

16526X: 83891; 83892 (x7); 83894 (x7); 83898 (x7); 83904 (x7); 83912

16537X: 82261. If reflex performed: 83891; 83892 (x7); 83894 (x7); 83898 (x7); 83904 (x7); 83912

Test Benefits

- Confirm biotinidase deficiency in infants identified by newborn screening (activity assay)
- Identify causative mutations (mutation assay)
- Identify biotinidase deficiency carriers (mutation assay)
- Prenatal diagnosis of biotinidase deficiency (mutation assay)
- No need to submit additional specimens to control for loss of enzyme activity during shipment

Clinical Summary

Biotinidase deficiency is an autosomal recessive disorder characterized by central nervous system and skin manifestations. Individuals with profound deficiency usually present around 3 months of age, but may present as early as the first week of life and as late as 10 years

of age. If untreated, the patient may experience seizures, developmental delay, skin lesions, and irreversible sensorineural deafness. Individuals with partial deficiency may have milder symptoms or may be asymptomatic. Patients are treated successfully with oral doses of free biotin, a water-soluble form of vitamin B.

Biotin is routinely recycled in the body when the enzyme biotinidase liberates biotin from endogenous and dietary proteins. Biotin acts as a coenzyme in gluconeogenesis, fatty acid synthesis, and branched chain amino acid catabolism. Biotinidase deficiency diminishes or prevents biotin recycling and coenzyme function. Thus, affected individuals may have ketolactic acidosis, organic aciduria, and mild hyperammonemia.

Biotinidase deficiency is detected via newborn screening (colorimetric, semiquantitative assessment of biotinidase activity) and follow-up quantitative measurement of biotinidase activity. *BTD* gene analysis can identify the causative mutation(s), of which there are about 100; identify carriers of a mutated gene (1:120 frequency in the general population); and diagnose the deficiency prenatally. Use of these laboratory tests enables early detection, which can lead to prevention of reversible and irreversible symptoms.

Quest Diagnostics, a Leader in Genetic Testing and Services

- **Extensive test menus**
 - Biochemical genetics, including confirmation of newborn screening test results
 - Cytogenetics, including a broad menu of FISH tests
 - Molecular genetics, including microarrays (ClariSure™ CGH)
- **Consultative services**
 - Board-certified Directors who can discuss complicated cases
 - Genetic Counselors who can help with test selection and interpretation

For assistance with test selection and interpretation, contact a Quest Diagnostics Genetic Counselor by calling 1-866-GENE-INFO (1-866-436-3463).

*The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payor being billed.

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