

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform Biotinidase DNA testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR BIOTINIDASE DEFICIENCY TESTING

Patient's Name _____ **Date of Birth** ____/____/____ **Gender** F M

Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Did the patient have an abnormal NEWBORN SCREEN? No Yes Unknown

Does the patient have REDUCED SERUM BIOTINIDASE ACTIVITY? No Yes Unknown

If yes, the reduced enzymatic level is consistent with:

- partial deficiency complete deficiency Unknown

Does the patient have SYMPTOMS? No Yes Unknown If yes, what was age of onset? _____

If yes, check all that apply

- | | | | |
|--------------------------------------|-----------------------------------|--|---------------------------------------|
| <input type="checkbox"/> Skin rash | <input type="checkbox"/> Alopecia | <input type="checkbox"/> Speech problems | <input type="checkbox"/> Hearing loss |
| <input type="checkbox"/> Candidiasis | <input type="checkbox"/> Seizures | <input type="checkbox"/> Developmental delay | <input type="checkbox"/> Other _____ |
| <input type="checkbox"/> Hypotonia | <input type="checkbox"/> Ataxia | <input type="checkbox"/> Vision problems | |

Does the patient have a FAMILY HISTORY OF BIOTINIDASE DEFICIENCY? No Yes Unknown

If yes, please attach a PEDIGREE or specify the **RELATIONSHIP** of family member(s) to the patient. _____

Is the relative a healthy carrier affected with biotinidase deficiency?

What are the *BTD* mutations in the family member? _____

Circle the Biotinidase test you intend to order

0093362 Biotinidase, Serum (with paired normal control): Biotinidase enzyme testing; ordered as an initial test.

0051730 Biotinidase Deficiency (BTD) Sequencing: Sequencing of the *BTD* coding region and intron/exon boundaries. Clinical sensitivity is 99% for both diagnostic and carrier detection.

0051700 Biotinidase Deficiency (BTD) 5 Mutations: Tests for five common *BTD* gene mutations. Clinical sensitivity is 60%.

2001961 Familial Mutation, Targeted Sequencing: A copy of the relative's DNA lab result is REQUIRED for this test.

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141

Master Label