ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR BIOTINIDASE DEFICIENCY TESTING

Patient Name _____ **Date of Birth** _____ **Sex** ☐ F ☐ M

Physician _____ **Physician Phone** _____

Practice Specialty _____ **Physician Fax** _____

Genetic Counselor _____ **Counselor Phone** _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ 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 (check all that apply) ☐ Unknown

☐ Age of onset: _____ ☐ Hypotonia

☐ Alopecia ☐ Seizures

☐ Ataxia ☐ Skin rash

☐ Candidiasis ☐ Speech problems

☐ Developmental delay ☐ Vision problems

☐ Hearing loss ☐ Other symptom(s): _____

Is there any relevant family history of biotinidase deficiency? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relationship of family member(s) to the patient: _____

The relative is: ☐ a healthy carrier ☐ affected with biotinidase deficiency

List the *BTD* mutations in the family member(s): _____

_____

Check the 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