ARUP LABORATORIES

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)

[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] 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

[ ] Skin rash [ ] Alopecia [ ] Speech problems [ ] Hearing loss
[ ] Candidiasis [ ] Seizures [ ] Developmental delay [ ] Other _____
[ ] Hypotonia [ ] Ataxia [ ] 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