ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

PATIENT HISTORY FOR TAY-SACHS DISEASE (HEXA DEFICIENCY) TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor's Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Ashkenazi Jewish ☐ Asian ☐ French Canadian

☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

Does the patient have symptoms? ..... ☐ No ☐ Yes (check all that apply)

☐ Ataxia ☐ Developmental delay ☐ Loss of motor skills

☐ Blindness ☐ Hypotonia ☐ Seizures

☐ Cherry-red spot on macula ☐ Liver disease ☐ Spasticity

☐ Neurodegeneration (describe): _____

☐ Other symptom(s): _____

Laboratory Findings:

HEX A enzyme (serum) ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

HEX A enzyme (leukocytes) ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

Has the patient undergone previous DNA testing for Tay-Sachs Disease? ..... ☐ No ☐ Yes ☐ Unknown

If yes, describe the test(s) and results: _____

_____

Is there any relevant family history of Tay-Sachs disease? ..... ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

_____

_____

Has DNA testing for the *HEXA* been performed for the family member(s)? ..... ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Is the patient's reproductive partner a carrier of Tay-Sachs disease? ..... ☐ No ☐ Yes ☐ Unknown

If yes, please list the variant _____

Check the test you intend to order.

☐ **3004486 Tay-Sachs Disease (*HEXA*) Sequencing and Deletion/Duplication:** Order to confirm the *I FYB* pathogenic or pseudodeficiency variant(s) in suspected carriers or symptomatic individuals.

☐ **0051428 Tay-Sachs Disease (*HEXA*) 7 Variations:** Molecular confirmation of common pathogenic and pseudodeficiency variants

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for pathogenic sequence variant(s) previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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