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 TAY-SACHS DISASE (HEX A 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: _____

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): _____

Is patient pregnant? (females) ☐ No ☐ Yes (gestational age: _____) ☐ Unknown

Is patient on any medications? ☐ No ☐ Yes (check all that apply and describe) ☐ Unknown

☐ Oral contraceptives: _____
☐ Hormone therapy: _____

Laboratory Findings:

HexA enzyme (serum) ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown
HexA enzyme (leukocytes) ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown
If yes, describe the test(s), methodology, 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:

Has DNA testing for the *HEXA* gene 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).

Check the test you intend to order.

☐ **2008129 Hexosaminidase A and Total Hexosaminidase in Plasma with Reflex to Hexosaminidase A and Total Hexosaminidase in Leukocytes:** Most comprehensive initial test to determine carrier or affected status.
☐ **2008121 Hexosaminidase A and Total Hexosaminidase, Plasma or Serum:** Initial test to determine carrier or affected status in male and non-pregnant females.
☐ **2008125 Hexosaminidase A and Total Hexosaminidase in Leukocytes:** Order for carrier testing on pregnant women, women taking oral contraceptives, or anyone with an inconclusive serum enzyme result.
☐ **0051428 Tay-Sachs Disease (*HEXA*) 7 Mutations:** Molecular confirmation of common pathogenic and pseudodeficiency mutations
☐ **2009298 Tay-Sachs Disease (*HEXA*) Sequencing and 7.6kb Deletion:** Order to confirm the specific *HEXA* mutation(s) in a suspected carrier or symptomatic individual.
☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation 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