ARUP LABORATORIES

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

PATIENT HISTORY FOR TAY-SACHS DISEASE (HEX A DEFICIENCY) TESTING

Patient 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_____

Does the patient have SYMPTOMS? [ ] No [ ] Yes [ ] Unknown If yes, check all that apply:
[ ] Developmental delay [ ] Hypotonia [ ] Seizures
[ ] Loss of motor skills [ ] Spasticity [ ] Cherry-red spot on macula
[ ] Blindness [ ] Ataxia [ ] Liver disease
[ ] Neurodegeneration (describe: _____) [ ] Other _____

Is patient PREGNANT? (Females) [ ] No [ ] Yes (Gestational Age _____) [ ] Unknown

Is patient on any MEDICATIONS? [ ] No [ ] Yes [ ] Unknown If yes, check all that apply:

[ ] Oral Contraceptives (list type) _____ [ ] Hormone Therapy (describe) _____

LABORATORY FINDINGS
HexA enzyme (serum) [ ] Normal [ ] Abnormal (result: _____) [ ] Not performed [ ] Unknown
HexA enzyme (leukocytes) [ ] Normal [ ] Abnormal (result: _____) [ ] Not performed [ ] Unknown

Is there a FAMILY HISTORY of TAY-SACHS DISEASE? [ ] No [ ] Yes [ ] Unknown
If yes, attach a PEDIGREE or specify the RELATIONSHIP of the family member(s) to the patient and detail the symptoms in each relative.
_____

Has DNA testing for the *HEXA* gene been performed for family member(s)? [ ] No [ ] Yes [ ] Unknown
If yes, please attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Has the patient undergone previous DNA testing? [ ] No [ ] Yes [ ] Unknown
If yes, please describe the test, methodology used, and the results _____

DESCRIPTION OF TAY-SACHS DISEASE TESTING

2008129 Hexosaminidase A and Total Hexosaminidase in Plasma with Reflex to Hexosaminidase A and Total Hexosaminidase in Leukocytes: 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.

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: Targeted sequencing for gene mutation(s) previously identified in a family member; a copy of a relative's lab result is REQUIRED.

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

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