Clinical Questionnaire for **Tay-Sachs Disease Screening**

This form should be filled out when Tay-Sachs disease biochemical or DNA testing is ordered (test numbers 510412, 511246, 510404, 333561, or 332859). The form should be completed by the ordering physician's office and should accompany the sample. Please call 800-345-4363 with any questions. Tay-Sachs disease is a lysosomal storage disease that causes progressive neurological deterioration. People of Ashkenazi Jewish and French-Canadian ancestry are at increased risk to be carriers of this disorder.

There are several methods available for carrier screening, including enzyme testing in serum or leukocytes and direct DNA screening for common mutations. Enzyme testing is not mutation-dependent and is suitable for testing in all ethnic groups. Please note that the serum enzyme test is not accurate in pregnant women and women who take oral contraceptives. LabCorp's DNA test will identify greater than 94% of carriers who are Ashkenazi Jewish, 80% of carriers who are French-Canadian, and approximately 25% of carriers who are non-Jewish Caucasian. A detection rate for LabCorp's DNA test is not available for other ethnic backgrounds.[1-6]

Patient's name: _____ **Date of birth:** _____

Gender: ○ Male ○ Female Name of person completing form: _____

Physician's signature: _____ Physician's telephone: _____

Patient Ethnicity

○ Ashkenazi Jewish (Eastern European) ○ Sephardic Jewish (Spanish, Portuguese, or North African)

○ French-Canadian ○ Non-Jewish Caucasian

○ Other _____

Patient History

Is patient/spouse pregnant? ○ Yes ○ No What is the gestational age? _____

Is the patient taking oral contraceptives? ○ Yes ○ No

Any other medications in the past two weeks? (Please list) _____

Has the patient's spouse been identified as a Tay-Sachs carrier? ○ Yes ○ No

Indications for Testing

Routine Screening: ○ Yes ○ No

Family history of Tay-Sachs

Has anyone in this patient's family been diagnosed with Tay-Sachs disease? ○ Yes ○ No

If yes, what is the relationship to the patient (brother, sister, niece, first cousin, etc)? _____

Has anyone in this patient's family been identified as a carrier of Tay-Sachs disease? ○ Yes ○ No

If yes, what is the relationship to the patient (brother, sister, niece, first cousin, etc)? _____

Suspected diagnosis. Symptoms: _____

Sandhoff disease screening: _____

Other: _____

References

1. Kaback M. Hexosaminidase A deficiency. GeneReviews. Available at www.genetests.org. Accessed: May 19, 2006.
2. American College of Obstetricians and Gynecologists. *Prenatal and Preconceptional Carrier Screening for Genetics Diseases in Individuals of Eastern European Jewish Descent*. Washington, DC: ACOG; August, 2004. Technical Bulletin 298.
3. Triggs-Raine B, Richard M, Wasel N, Prence EM, Natowicz MR. Mutational analyses of Tay-Sachs Disease: Studies on Tay-Sachs Carriers of French Canadian Background Living in New England. *Am J Hum Genet*. 1995;56:870-879..
4. Akerman BR, Zielenski, Triggs-Raine BL, et al. A mutation common in non-Jewish Tay-Sachs Disease: Frequency and RNA Studies. *Human Mutat*. 1992; 1: 303-309.
5. Gross SJ, Pletcher BA, Monaghan KG. Carrier screening in individuals of Ashkenazi Jewish descent. *Genet Med*. 2008; 10(1):54-56.
6. Monaghan KG, Feldman GL, Palomaki GE, et al. Technical standards and guidelines for reproductive screening in the Ashkenazi Jewish population. *Genet Med*. 2008; 10(1):57-72.

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