genzyme

Carrier Testing

Patient Name: [redacted]
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

DOB: [redacted]
Sex: M
SSN: [redacted]

Date Collected: 07/23/2009
Date Received: 07/25/2009
Lab ID:
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Not Provided

Indication: Not Provided

Disease	Result	Interpretation
Niemann-Pick Type A	Negative	This negative result may need further interpretation depending on the clinical indication.
Tay-Sachs - DNA	Negative	This negative result may need further interpretation depending on the clinical indication.
Tay-Sachs - Enzyme	Hex. Activity: 846 nmol/mg protein Hex. Percent A: 59	Non carrier Plasma/Serum WBC Non carrier range: Hex A >= 55% >= 55% Carrier range : Hex A 20 - 48% 20 - 49%
Bloom Syndrome	Negative	This negative result may need further interpretation depending on the clinical indication.
Canavan Disease	Negative	This negative result may need further interpretation depending on the clinical indication.
Cystic Fibrosis	Negative	This negative result may need further interpretation depending on the clinical indication.

SCAN
MM

AUG 07 2009

Category Lab results
Subject

(REPORT CONTINUED ...)

Date: 08/03/2009

Ruth A. Heim, Ph.D., FACMG
Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

[redacted]

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Carrier Testing

Patient Name: [redacted] Referring Physician: [redacted] Specimen #: [redacted] Patient ID: [redacted]

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Disease	*Result*	*Interpretation*
Familial Dysautonomia	Negative	This negative result may need further interpretation depending on the clinical indication.
Fanconi Anemia - C	Negative	This negative result may need further interpretation depending on the clinical indication.
Glycogen Storage Disease Type 1a	Negative	This negative result may need further interpretation depending on the clinical indication.
Gaucher Disease	Negative	This negative result may need further interpretation depending on the clinical indication.
Mucolipidosis Type IV	Negative	This negative result may need further interpretation depending on the clinical indication.
Maple Syrup Urine Disease	Negative	This negative result may need further interpretation depending on the clinical indication.

COMMENTS:

DNA:
The negative results from this analysis cannot eliminate the possibility that this individual carries a mutation not detected by this test. Unless otherwise noted, interpretations are based on a negative family history and the absence of symptoms.

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.

Enzyme: [White Blood Cells]
This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

METHOD:

DNA is isolated from the sample and amplified for disease specific regions using the polymerase chain reaction (PCR). Mutations are identified by hybridization to allele specific oligonucleotides or by solution-phase multiplex allele-specific primer extension with subsequent mutation-specific hybridization and detection.

False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation or somatic heterogeneity of the tissue sample. This test was developed and its performance characteristics determined by Genzyme. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.

(REPORT CONTINUED ...)

Date: 08/03/2009

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