MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,AJPWO	SA00007356	32	F	SA00007356
Ordering Phys				**DOB** 04/04/1980
Client Order # SA00007356	**Account Information**			**Report Notes**
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/15/2012 14:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Ashkenazi Jewish Panel Without CF			REPORTED 08/14/2012 16:52		
Specimen		Blood			MCR
Specimen ID		976453			MCR
Order Date		01 Aug 2012 13:02			MCR
Result					MCR

Tay-Sachs mutation analysis: One copy of the G269S mutation was identified.

Hexosaminidase enzyme analysis:

Hexosaminidase A and Total:
20.5 U/g prot Normal Range: 16.4-36.2 U/g prot

% Hexosaminidase A:
49% Normal Range: 63-75%

Hexosaminidase enzyme activity is consistent with POSITIVE carrier status for Tay-Sachs.

Gaucher:	A mutation was NOT identified.
Canavan:	A mutation was NOT identified.
Familial dysautonomia:	A mutation was NOT identified.
Bloom syndrome:	A mutation was NOT identified.
Fanconi anemia(FANCC):	A mutation was NOT identified.
Mucolipidosis IV:	A mutation was NOT identified.
Niemann-Pick types A and B:	A mutation was NOT identified.

Interpretation MCR

Biochemical and molecular analyses for Tay-Sachs are in agreement. This result indicates that this individual is a carrier of Tay-Sachs. This interpretation assumes that this individual is healthy and is not clinically affected with Tay-Sachs.

Since a mutation has been identified, genetic testing of at risk family members is possible. Genetic testing should be offered to this individual's reproductive partner to further clarify their risk of having a child with this disease.

Having excluded the presence of the remaining listed mutations, the risk that this individual is a carrier of another mutation not tested by the method described is approximated below for each of the remaining diseases. These risk calculations assume that there is no family

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* Report times for Mayo performed tests are CST/CDT

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history of the disease and are based on the mutation detection rates and carrier frequencies listed.

	Risk (Detection rate, Carrier freq)
Gaucher:	1/341 (95%, 1/18)
Canavan:	1/2001 (98%, 1/41)
Familial dysautonomia:	1/3001 (99%, 1/31)
Bloom syndrome:	1/10,601 (99%, 1/107)
Fanconi anemia(FANCC):	1/8801 (99%, 1/89)
Mucolipidosis IV:	1/2521 (95%, 1/127)
Niemann-Pick A/B:	1/2968 (97%, 1/90)

The risk calculation provided is specific for the Ashkenazi Jewish population. There is considerable variability both in the carrier frequency and detection rate among different ethnic groups. Because there is little information about the carrier frequency and mutation detection rate for individuals of non-Ashkenazi Jewish ancestry, we are unable to provide a specific revised risk assessment for individuals of other ethnicities.

A genetic consultation may be of benefit.

CAUTIONS:
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Laboratory developed test.

Reason For Referral MCR

Carrier screen for diseases included in the Ashkenazi Jewish panel.

Method MCR

A PCR-based assay was used to test for mutations common in

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the Ashkenazi Jewish population. These include: Canavan (E285A, Y231X(C>A, C>T), 433(-2)A>G, and A305E); familial dysautonomia (IVS20(+6)T>C and R696P); Tay-Sachs (1278insTATC, G269S, IVS12(+1)G>C, IVS9(+1)G>A, delta7.6kb, R247W, R249W); Gaucher (N370S, IVS2(+1)G>A, 84G>GG, R496H, L444P, delta55bp, V394L, and D409H); Bloom syndrome (2281del6/ins7); FANCC related Fanconi anemia (IVS4(+4)A>T and 322delG); mucolipidosis IV (IVS3(-2)A>G and delta6.4kb); and Niemann-Pick types A and B (L302P, R496L, fsP330, and deltaR608).					
Hexosaminidase enzyme activity is measured on blood leukocytes for Tay-Sachs screening.					
Reviewed By		D Brian Dawson PhD			MCR
Released Date		14 Aug 2012 16:46			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

TEST SITE