Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,AJPWO A	SA00062942	46	M	SA00062942
Ordering Phys CLIENT,CLIENT				**DOB** 10/06/1966
Client Order # C7028846	**Account Information**			**Report Notes**
Collected 10/04/2013 00:00	C7002670-MML Unknown Client Account 3050 Superior Drive NW Rochester, MN 55901			
Printed 10/08/2013 11:43				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Ashkenazi Jewish Panel Without CF					
RECEIVED: 10/04/2013 13:54 **REPORTED:** 10/04/2013 16:18					
Specimen		Blood			MCR
Specimen ID		1062523			MCR
Order Date		04 Oct 2013 14:53			MCR
Result					MCR
Tay-Sachs mutation analysis: One copy of the G269S mutation was identified.					
Hexosaminidase enzyme analysis:					
Hexosaminidase A and Total:					
9.4 nmol/min/mg nmol/min/mg		Normal Range (</=15 years): >/=20			
		Normal Range (>/=16 years): 16.4-36.2 nmol/min/mg			
% Hexosaminidase A:					
49%		Normal Range (</=15 years): 20-80%			
		Normal Range (>/=16 years): 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					

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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
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:	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.

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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 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		04 Oct 2013 16:15			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: