

Patient Name TESTINGRNV,AJPWOREPORT	Patient ID SA00052905	Age 27	Gender M	Order # SA00052905
Ordering Phys CLIENT,CLIENT				DOB 02/11/1985
Client Order # SA00052905	Account Information			Report Notes
Collected 01/20/2013	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 01/22/2013 11:48				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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Ashkenazi Jewish Panel Without CF

REPORTED 01/21/2013 16:20

Specimen		Blood			MCR
Specimen ID		1039363			MCR
Order Date		21 Jan 2013 16:08			MCR
Result					MCR

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.
 Mucopolipidosis IV: A mutation was NOT identified.
 Niemann-Pick types A and B: A mutation was NOT identified.
 Tay-Sachs mutation analysis: A mutation was NOT identified.

Hexosaminidase enzyme analysis:

Hexosaminidase A and Total:
 20.8 nmol/min/mg Normal Range: 16.4-36.2 nmol/min/mg

% Hexosaminidase A:
 68% Normal Range: 63-75%

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

Interpretation

MCR

Having excluded the presence of the listed mutations, the risk that this individual is a carrier of another mutation not tested by the method described is approximated below for each disease. 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. The residual risk that this individual is a carrier of Tay-Sachs is based on a negative molecular test result in addition to hexosaminidase enzyme activity within the normal range.

	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)
Mucopolipidosis IV:	1/2521 (95%, 1/127)
Niemann-Pick A:	1/2968 (97%, 1/90)

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* Report times for Mayo performed tests are CST/CDT

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Tay-Sachs:	1/3001	(99%, 1/31)			

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 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

Carrier screen for diseases included in the Ashkenazi Jewish panel.

MCR

Method

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); mucopolipidosis IV (IVS3(2)A>G and delta6.4kb); and Niemann-Pick types A and B (L302P, R496L, fsP330, and deltaR608).

MCR

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Hexosaminidase enzyme activity is measured on blood leukocytes for Tay-Sachs screening.

Reviewed By	Jessica Rose Chavey	MCR
Released Date	21 Jan 2013 16:17	MCR

* Performing Site:

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