

Laboratory Service Report

1-800-533-1710

REPORTED 12/11/2012 15:54

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT, FRAPS	SA00051867	46	M	SA00051867
Ordering Phys				DOB
CLIENT, CLIENT				05/25/1966
Client Order #	Account Information			Report Notes
SA00051867				
Collected	C7028846-DLMP ROC	HESTER		
12/10/2012		3050 SUPERIOR DRIVE		
Printed	ROCHESTER,MN 5590)1		
12/18/2012 14:41				

Test Flag Results Unit Value Site*

TRAPS (TNFRSF1A) Exons 2-5

Result:

Y03

Positive: Heterozygous for T79M Mutation in the TNFRSF1A Gene.

This individual is heterozygous for a C>T nucleotide substitution in exon 3 of the TNFRSF1A gene, resulting in the replacement of a Threonine codon (ACG) with a Methionine codon (ATG) at amino acid position 79. This mutation is denoted c.236C>T at the cDNA level or p.Thr70Met (T79M) at the protein level. Please note, this mutation is also referred to as T50M using alternative nomenclature based on the mature, processed protein. Interpretation:

The T79M missense mutation in the TNFRSF1A gene has been reported previously, as T50M, in association with TRAPS (McDermott et al., 1999; Aksentijevich et al., 2001), and its presence is consistent with the diagnosis in this patient.

Recommendation:

Y03

Y03

This result permits mutation-specific molecular testing for at-risk family members. Genetic counseling is recommended. Genomic DNA from this specimen was PCR-amplified for analysis of exons 2-5, their flanking splice sites, as well as introns 2 and 4 of the TNFRSF1A gene. Bidirectional sequence was obtained and DNA sequence was analyzed and compared to the published gene sequence. The methods used by GeneDx are expected to be greater than 99% sensitive in detecting mutations identifiable by sequencing.

This test was developed and its performance characteristics determined by GeneDx. 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. Pursuant to the requirements of CLIA '88, this laboratory has established and verified the test's accuracy and precision.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status	
SAMPLEREPORT, FRAPS	12/10/2012	Final	
Page 1 of 2		>> Continued on Next Page >>	

^{*} Report times for Mayo performed tests are CST/CDT



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				Reference	e Perform
Test	Flag	Results	Unit	Value	Site

* Performing Site:

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Y039	GeneDx, Inc.	Lab Director:
1039	207 Perry Parkway Gaithersburg MD 20877	Lab Director.

Patient Name SAMPLEREPORT.FRAPS	Collection Date and Time	Report Status Final	
Page 2 of 2		** End of Report **	

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