RETZ

RET Proto-Oncogene, Full Gene Analysis, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00001030	SAMPLEREPDMGLM, VLD20150713A0291		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00001030	SA00001030	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 19:22			

Result Summary

MCR

Negative

Result

MCR

No reportable variants were identified.

Interpretation

1 MCR

This result decreases the likelihood but does not rule out a diagnosis of multiple endocrine neoplasia type 2A (MEN2A) or familial medullary thyroid carcinoma (FMTC). We predict that there are individuals with a diagnosis of MEN2A/FMTC who have a pathogenic mutation that is not detectable by the method described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations). Additionally, this assay does not rule out the presence of mutations in other genes associated with hereditary thyroid cancer or paragangliomas.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

ADDITIONAL INFORMATION

Next-generation sequencing was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the RET gene (GenBank accession number NM_0020975; build GRCh37 (hg19)). Sanger sequencing is used to confirm reported alterations detected by next-generation sequencing when appropriate. An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

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 false-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 Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:29 **Reported:** 24 Jul 2015 20:53

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

Patient ID	Patient Name		Birth Date	Gender	Age
SA00001030	SAMPLEREPDMGLM, VLD20150713A0291		1981-01-01	M	34
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00001030	SA00001030	CLIENT, CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		12 Jul 2015 19:22			

Laboratory Notes

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292