MAYO CLINIC LABORATORIES

1-800-533-1710

CSP53

TP53 Gene Somatic Mutation Pre-Analysis Cell Sorting, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00130908	TESTINGRNV, P53CAREPORT		1999-09-09	M	20
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00130908	SA00130908	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Jul 2020 08:15			

TP53 Pre-Analysis Cell Sorting, V

TP53 Pre-Analysis Cell Sort

1 MCR

Performed

ADDITIONAL INFORMATION

Flow cytometric cell selection was performed with antibodies to the following antigens: CD19, CD20, CD45, surface kappa and lambda.

Specimen enrichment for certain cell types is necessary, in order to enhance the sensitivity of genetic/molecular abnormalities detection in the cell population of interest, and to avoid unwanted contamination from other cell types. Flow cytometric cell sorting is the most direct and robust method of obtaining a pure population for subsequent genetic/molecular analysis, through assessment of a characteristic combination of cell surface antigens.

Received: 14 Jul 2020 13:04 **Reported:** 14 Jul 2020 13:20

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

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SA00130908	TESTINGRNV, P53CAREPORT		1999-09-09	M	20
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C7028846 DLMP Rochester		13 Jul 2020 08:15			

TP53 gene somatic mutation analysis

Specimen Type: MCR

Peripheral blood

Signing Pathologist MCR

BENJAMIN BAILEY

Final Diagnosis: [1] MCR

Peripheral blood, TP 53 Analysis for Tumor-associated Genetic Alterations, Sequencing:

Positive. A genetic alteration in the TP 53 gene was detected.

The genetic alteration identified is: c.610del; p.Glu20Serfs*fs.

The predicted effect of this genetic alteration is: likely pathogenic. Somatic TP 53 genetic alterations often result in loss of protein function, although in some cases, the changes could result in an aberrant gain of function (http:/www-p53iarc.fr/FunctionCriteria.asp and http:/p53free.fr/Database/p53_recommentations.html).

The identified TP 53 genetic alteration in this specimen is most likely a somatic alteration in the neoplastic cell population; however, this test does not differentiate between a germline versus somatic alteration. A small possibility exists that this abnormality may be a germline nucleotide change (i.e. present in all cells at birth and typically inherited from a parent rather than somatically acquired). Congenital TP 53 alterations are known to cause an inherited cancer syndrome known as Li-Fraumeni syndrome. Correlation with the patient's clinical presentation and family history is suggested. If there is concern for a familial condition, further genetic testing and/or counseling could be considered.

Peripheral blood B-lymphocytes were enriched by flow cytometry to >95% purity prior to DNA isolation.

ADDITIONAL INFORMATION

Method Summary: DNA was extracted from the sample and PCR was performed to amplify exon regions 4 to 9 of the TP53 gene. The presence or absence of acquired (somatic) TP53 mutations involving exons 4 to 9 and associated splice junctions is assessed by Sanger sequencing. The analytic sensitivity of the assay is approximately 20%. However, mutations outside of the analyzed region, or mutations present at low level or in small subclonal populations cannot be excluded by this method. The reference gene transcript used for variant annotation is: GRCh37(hg19)NM_000546.4.

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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.

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