

Hematologic Neoplasms, TP53 Somatic Mutation,
DNA Sequencing Exons 4-9, Varies

Patient ID SA00179273	Patient Name TESTING, P53CA_ABNORMAL	Birth Date 2005-05-05	Sex F	Age 20
Order Number SA00179273	Client Order Number SA00179273	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 13 Feb 2026 12:03		

TP53 gene somatic mutation analysis

Specimen Type:

MCR

Bone marrow

Signing Pathologist

MCR

LAUREN SCHULTZ

Final Diagnosis:

1 MCR

Bone marrow, TP53 Analysis for Tumor-associated Genetic Alterations, Sequencing:

Positive. A genetic alteration in the TP53 gene was detected.

The genetic alteration identified is:g.7577559G>A; c.722C>T; p.Ser241Phe.

The predicted effect of this genetic alteration is: likely pathogenic. Somatic TP53 genetic alterations often result in loss of protein function, although in some cases, the changes could result in an aberrant gain of function (<https://p53.iarc.fr/TP53GeneVariations.aspx>).

The identified TP53 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 TP53 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.

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.

Indication for Test

MCR

SOB

Received: 16 Feb 2026 12:16

Reported: 16 Feb 2026 12:42

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	Nikola Baumann Ph.D.	24D0404292