MAYO CLINIC LABORATORIES

1-800-533-1710

TMSI

Microsatellite Instability, Tumor

Patient ID	Patient Name		Birth Date	Gender	Age
SA00140250	TESTINGTMSI, NORMAL		1950-11-11	F	70
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00140250	SA00140250	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		11 Nov 2020 00:00			

Tumor, Microsatellite Instability

Result Summary

MCR

MSS

Result

MCR

Provided diagnosis: colorectal adenocarcinoma

MSI Result: MSS (instability observed in 0 of 7 informative markers)

Interpretation

1 MCR

An MSS phenotype suggests the presence of normal DNA mismatch repair function within the tumor.

THERAPEUTIC IMPLICATIONS

Current data suggest that advanced stage solid tumors with intact mismatch repair (MSS) are less likely to be responsive to treatment with immunotherapies such as anti-PD-1 therapies (Science. 2017 Jul 28;357(6349):409–413(PMID 28596308); J Clin Oncol. 2018 Jan 20:JCO2017769901 (PMID 29355075)). In stage II colon cancers with intact mismatch repair (MSS) and high risk features, 5-FU based therapy may be effective (J Clin Oncol. 2010 Jul 10;28(20):3219–26 (PMID 20498393)).

HEREDITARY IMPLICATIONS

These results decrease the likelihood but do not eliminate the possibility that this individual has HNPCC/Lynch syndrome. These results do not rule out the possibility that this individual's tumor is due to an inherited defect in another gene not involved in DNA mismatch repair. A significant fraction of clinically defined HNPCC cases (30% or more) do not have defective DNA mismatch repair as the underlying genetic basis of their disease. Additionally, we cannot rule out the possibility that this tumor could represent a sporadic occurrence. If there is a strong personal or family history of HNPCC/Lynch syndrome related cancers for this patient or if this individual has multiple tumors, consider microsatellite instability (MSI) and immunohistochemical staining (IHC) on a different tumor to further evaluate the possible role of defective DNA mismatch repair for this individual or family. A genetic consult may be of benefit.

PROGNOSTIC IMPLICATIONS

The presence of intact mismatch repair (MSS) is considered to be an unfavorable prognostic factor for patients with colorectal cancer (J Clin Oncol. 2005 Jan 20;23(3):609–18 (PMID 15659508)).

ADDITIONAL INFORMATION

Consideration of these results, in light of other clinical information, may aid in clinical management decisions for this patient.

Of note, the literature suggests that MSI analysis on neoadjuvant chemoradiated tumor specimens may influence MSI status and lead to an erroneous interpretation of results (Int J Radiat Oncol Biol Phys. 2007 68(5):1584).

These data should be interpreted in the context of the histopathologic findings. A surgical pathology consult may be ordered separately. If immunohistochemistry (IHC) for the mismatch repair proteins was also ordered on this specimen, the results will be reported separately under test code IHC (IHC / MMR Protein, IHC Only, Tumor). For questions regarding the interpretation of IHC and MSI results, please contact the Genomics Laboratory at 1–800–533–1710.

ADDITIONAL INFORMATION

Microscopic examination was performed by a pathologist to identify areas of normal and tumor for enrichment by macrodissection. A PCR-based assay is used to test for tumor microsatellite instability (TMSI) with the use of 7 mononucleotide repeat markers (ACVR2A, BTBD7, DIDO1, MRE11, RYR3, SEC31A, and SULF2). The tumor tissue is classified as MSS (instability detected in 0 or 1 out of 7 markers), or MSI-H (instability in 2 or more of 7 markers tested). Due to the sensitivity of the method being used, microsatellite instability cannot be reliably detected in colorectal samples containing less than 20% tumor DNA or samples from other tumors containing less than 40% tumor DNA. Samples are routinely macrodissected to enrich for tumor cells, with colorectal samples less than 20% and other tumor types less than 40% rejected from further testing. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. If results

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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obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Specimen MCR

Tissue, Tumor

Tissue ID MCR

1234

Released By MCR

Kevin C. Halling, M.D., Ph.D.

Received: 11 Nov 2020 13:10 **Reported:** 11 Nov 2020 13:58

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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