

Test Description

MSI testing is used for Hereditary Cancer screening (Hereditary Non-Polyposis Colorectal Cancer -HNPCC or Lynch syndrome); As a biomarker (Prognostic and predictive biomarker for the response of Immunotherapy)

Patient Demographic

Name: Aruna Mittal Sex: Female Date of Birth/Age: 48 years Disease: Endometrial Cancer

| PATIENT | REPORT DATE | BOOKING ID |
|--------------|-------------|--------------|
| Aruna Mittal | 28 Mar 2019 | 011903270470 |

Clinician

Clinician Name: Dr Amit Verma Medical Facility: Max Hospital Pathologist: Not Provided

Specimen

Site: Endometrium Sample Type: FFPE block 1173/19 Date of Collection: 27-03-2019 Date of Booking: 28-03-2019

iMSI Rapid[™] Assay

Result

Microsatellite - High (MSI-H)

BIOMARKER FINDINGS

| ACVR2A | Mutation detected |
|--------|----------------------|
| BTBD7 | Mutation detected |
| DID01 | Mutation detected |
| MRE11 | No mutation detected |
| RYR3 | No mutation detected |
| SEC13A | Mutation detected |
| SULF2 | mutation detected |

INTERPRETATION

| Mutations are detected in 5 of the 7 markers | | |
|--|---|--|
| *MSS | <2 of the 7 markers demonstrate instability | |
| #MSI-H | \geq 2 of the 7 markers demonstrate instability | |
| *Microsatellite stable | | |
| # Microsatellite Instability-High | | |
| For valid batch test results specific controls are being run with every batch. | | |

METHODOLOGY

Multiplex detection of seven mononucleotide repeats using molecular beacon probe-based polymerase chain reaction followed by high resolution melt-curve analysis. The assay uses seven novel biomarkers *ACVR2A*, *BTBD7*, *DID01*, *MRE11*, *RYR3*, *SEC31A* and *SULF2* as this set of biomarkers is stable over different cancer types and ethnicities and show high performance than other known assays like *Bethesda Panel*. This test is carried out on Idylla platform using the MSI/1.0 Cartridge based kit which is CE IVD approved.

 REFERENCES
 Zhao et al. (2014) eLife 3: e02725, 1-26.

 De Craene B. et al. (2018) ASCO Abstract #e15639.
 Zhao et al. (2018) ASCO Abstract #e15654

March 28 2019

Dr Gulshan Yadav, MD, Consultant Pathology

Date