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)

Clinician
Clinician Name: Not provided
Medical Facility: Dr Lal Pathlabs
Pathologist: Dr Atul Thatai

Patient Demographic
Name: Perin R Patel
Sex: Male
Date of Birth/Age: 18 years
Disease: Colon Adenocarcinoma

Specimen
Site: Colon
Sample Type: FFPE block 19H-16379 A2
Date of Collection: 28-02-2020
Date of Booking: 28-02-2020

iMSI Rapid™ Assay

Result — Microsatellite - High (MSI-H)

BIOMARKER FINDINGS

ACVR2A	Mutation detected
BTBD7	Mutation detected
DIDO1	Mutation detected
MRE11	Mutation detected
RYR3	Mutation detected
SEC13A	No mutation detected
SULF2	No mutation detected

INTERPRETATION

Mutations are detected in 5 of the 7 markers

*MSS	<2 of the 7 markers demonstrate instability
#MSI-H	≥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*, *DIDO1*, *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

February 29, 2020

Dr Gulshan Yadav, MD, Consultant Pathology — Date