

MCCRD-SOP0057: Detection of Microsatellite Instability (MSI) Status from Whole Exome Sequencing Data

Laboratory: Molecular Characterization and Clinical Assay Development Laboratory

Revision Date: 7/3/2020

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Effective Date: 7/3/2020

**Please check for revision status of the SOP at**

<https://pdmr.cancer.gov/sops/>

**PDMR** **NCI Patient-Derived Models Repository**  
An NCI Precision Oncology Initiative<sup>SM</sup> Resource

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## VERSION INFORMATION

### 1. Change History

| Revision | Description   |
|----------|---|
|          | Internal SOP used by MOCHA Laboratory   |
| 7/3/2020 | Standardize SOP for posting to PDMM internal site for use by designated NCI intramural laboratories |

### 2. Related SOPs

|   |
|---|
| MCCRD_SOP0011: Whole Exome Sequencing Data Analysis Pipeline and Specifications |
|---|

### 3. References

- [1] Beifang Niu\*, Kai Ye\*, Qunyuan Zhang, Charles Lu, Mingchao Xie, Michael D. McLellan, Michael C. Wendl and Li Ding#.MSIsensor: microsatellite instability detection using paired tu-mor-normal sequence data. Bioinformatics 30, 1015–1016 (2014).
- [2] <https://github.com/niu-lab/msisensor2>

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## 1.0 PURPOSE/SCOPE

This Standing Operating Procedure (SOP) describes procedures for detection of microsatellite instability (MSI) status using whole exome sequencing (WES) data for reporting in the NCI Patient-Derived Models database as performed by the Molecular Characterization Laboratory (MoCha) at the Frederick National Laboratory for Cancer Research. **This SOP is for research-use purposes only; do not use for clinical sample analysis.**

## 2.0 DESCRIPTION OF MSI DETECTION

- b. The processed bam files are generated using whole exome sequence (WES) data following the WES data analysis pipeline in the SOP MCCRD\_SOP0011.
- c. MSI status is estimated using MSIsensor2 package<sup>[1,2]</sup>.

## 3.0 CODE DESCRIPTION

- d. MSIsensor2 is used to detect MSI score for tumor sample
  - `msisensor2 msi -M msisensor2/models_hg19 -t ${file}.bam -o ${file}`
- e. MSI status is determined based on the recommended msi score cutoff value 20%
  - MSI-H: msi score  $\geq$  20%
- f. Otherwise, MSI-Stable