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 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

3.0 RELATED SOPS

Available on the PDMR website: https://pdmr.cancer.gov/sops

MCCRD_SOP0011: Whole Exome Sequencing Data Analysis Pipeline and Specifications

4.0 DESCRIPTION OF MSI DETECTION

4.1 The processed bam files are generated using whole exome sequence (WES) data following the WES data analysis pipeline in the SOP MCCRD_SOP0011.

4.2 MSI status is estimated using MSIsensor2 package[1,2].

5.0 CODE DESCRIPTION

5.1 MSIsensor2 is used to detect MSI score for tumor sample

- msisensor2 msi -M msisensor2/models_hg19 -t ${file}.bam -o ${file}

5.2 MSI status is determined based on the recommended msi score cutoff value 20%

- MSI-H: msi score $\geq$ 20%
- Otherwise, MSI-Stable

6.0 REVISION HISTORY:

Document No.	Version	Description of Revision	Effective Date
MCCRD-SOP0057 (EXT)	1.0	Original Release	7/3/2020