

MSK-IMPACT® Flex powered with SOPHiA DDM™ enables sample-specific comprehensive genomic profiling through selective analysis of DNA and RNA biomarkers

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SOPHiA GENETICS, Rolle, Switzerland. Conflicts of interest: All authors are employees of SOPHiA GENETICS. Poster #ST041.

1 Highlights



Modular and flexible design

MSK-IMPACT® Flex powered with SOPHiA DDM™ enables selective inclusion of DNA, RNA, and shallow WGS modules, allowing laboratories to tailor comprehensive genomic profiling to each sample.



High analytical performance across analytes

A unified workflow and analytical pipeline deliver accurate detection of diverse biomarker types, including SNV/Indels, CNVs, fusions, gene expression, MSI, TMB, HRD, and tumor purity.

3 Aims and Methods

- ✓ Demonstrate a complete and modular workflow for flexible genomic profiling.
- ✓ Assess analytical performance of the solution across key biomarker types.
- ✓ Evaluate the benefit of multimodal analysis for complex biomarker detection and interpretation.

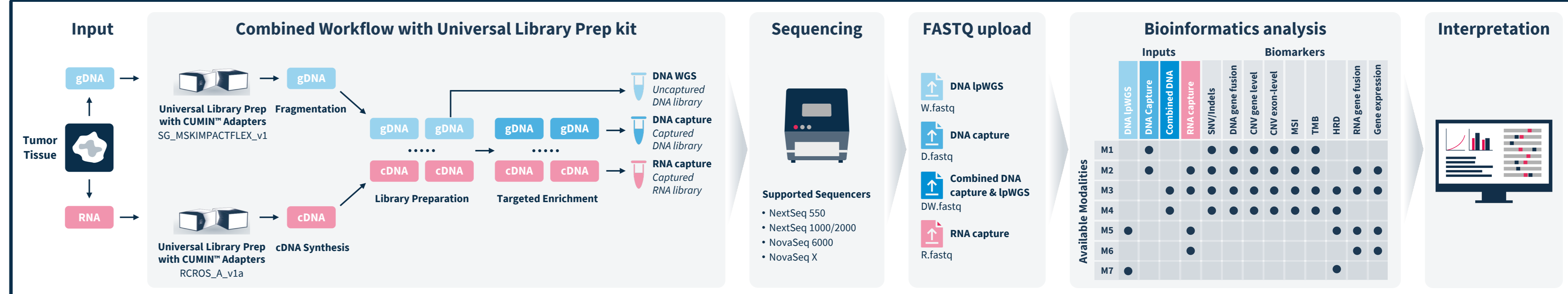
To evaluate performance and modularity, targeted DNA sequencing, targeted RNA sequencing, and shallow WGS were performed within a single, harmonized workflow using unified library preparation chemistry. Each analyte was processed independently and in combination to assess analytical performance across configurations. Clinical FFPE tumor samples with known biomarker status were analyzed, and results were compared with established orthogonal reference assays. Sequencing data were processed using SOPHiA DDM™, which automatically adapts to the available data types to perform SNV/Indel, CNVs, fusions, exon-skipping, gene expression, MSI, TMB, HRD, and tumor purity analysis.

2 Background

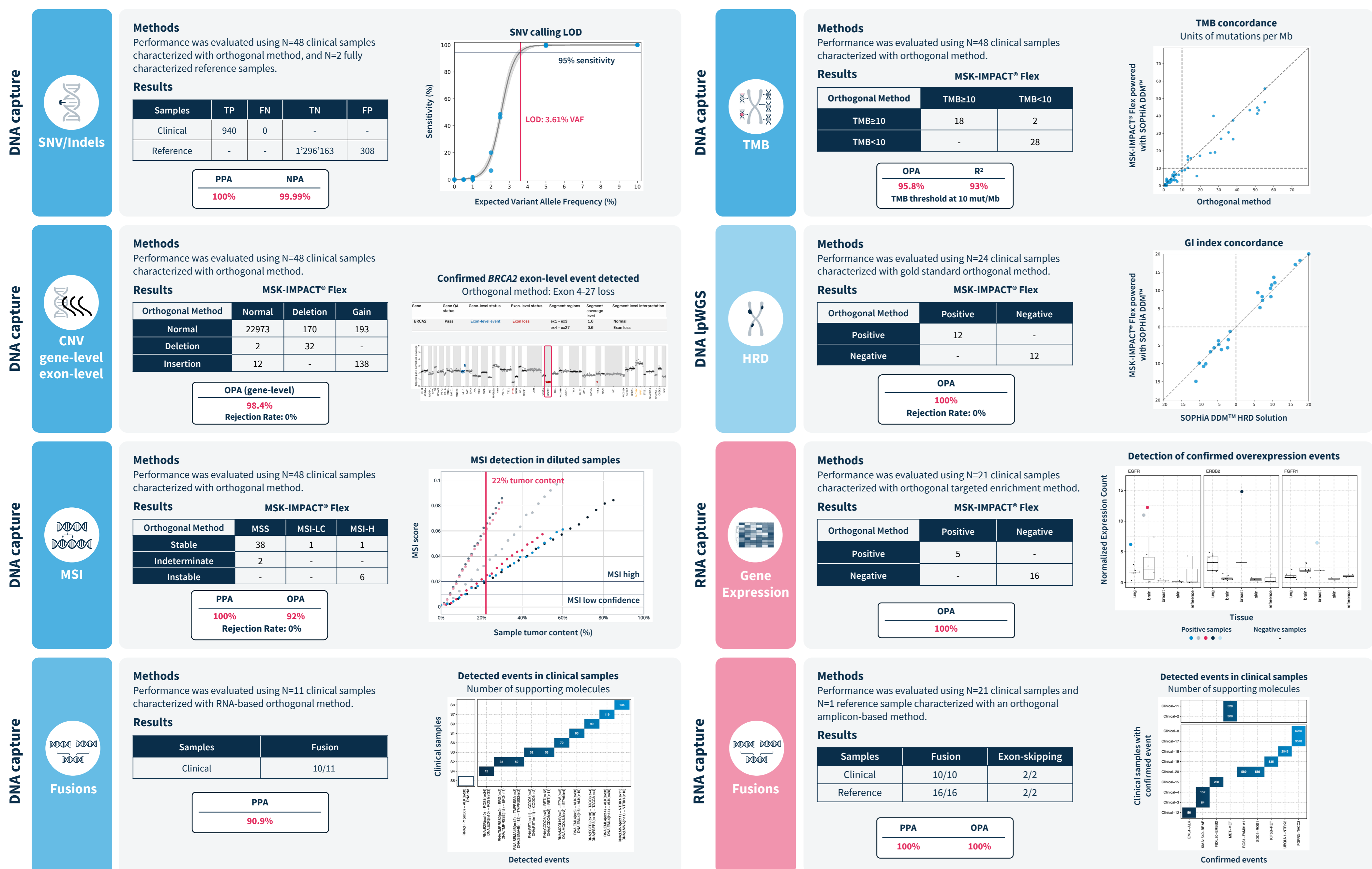
Comprehensive genomic profiling (CGP) has transformed precision oncology by enabling simultaneous detection of multiple clinically relevant biomarkers. However, conventional workflows often rely on multiple, purpose-specific assays, resulting in inefficiency, higher costs, and limited adaptability to case-specific needs. **MSK-IMPACT® Flex powered with SOPHiA DDM™** was developed to overcome these

limitations by combining targeted DNA, targeted RNA, and shallow whole-genome sequencing (WGS) within a single, harmonized workflow. This modular design allows laboratories to flexibly generate and integrate genomic data based on clinical context, supporting a broad range of biomarker types while maintaining analytical performance equivalent to dedicated assays.

4 MSK-IMPACT® Flex powered with SOPHiA DDM™ Workflow



5 Analytical performance across DNA and RNA biomarkers



6 Combined workflow enhances complex biomarker analysis

