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Comparative Analysis of Master Panel Data on MGI DNBSEQ-G50 and NovaSeq 6000

Abstract

This white paper presents a comparative analysis of the AmoyDx® Master Panel on two sequencing platforms: MGI DNBSEQ-G50 (MGI G50) and Illumina NovaSeq 6000. The study evaluated 61 DNA samples and 25 RNA samples, including reference materials and clinical FFPE specimens from various cancer types. Key metrics compared between the platforms included sequencing quality, variant detection performance (SNV/InDel, Fusion, and CNV), and biomarker assessment (HRD (GSS), MSI, TMB, EBV, TME, and GEP). The results demonstrate that the MGI G50 delivers accuracy and reliability comparable to the NovaSeq 6000, supporting its use as a viable alternative for comprehensive genomic profiling (CGP).

Introduction

Comprehensive genomic profiling (CGP) panels are advanced molecular diagnostic tools designed to analyse a broad range of genomic alterations across multiple cancer-related genes in a single test. These panels are increasingly used in clinical oncology to guide precision medicine, enabling clinicians to match patients with targeted therapies, immunotherapies, or relevant clinical trials based on their tumor's molecular profile.

The AmoyDx® Master Panel is a robust CGP solution that provides reliable detection of single nucleotide variants (SNVs), insertions/deletions (InDels), gene fusions, and copy number variations (CNVs) at the DNA level. It also supports accurate assessment of critical biomarkers such as homologous recombination deficiency (HRD, via GSS), microsatellite instability (MSI), and tumor mutational burden (TMB). At the RNA level, the panel enables detection of gene Fusions and evaluation of Epstein–Barr virus (EBV) status, tumor microenvironment (TME), and gene expression profile (GEP). Together, these features make the AmoyDx® Master Panel a comprehensive and reliable tool for molecular characterization in cancer diagnostics and treatment planning.

The AmoyDx® Master Panel has been optimized for use on Illumina sequencing platforms including NovaSeq 6000. With the growing demand for more flexible and efficient sequencing options, this study aims to validate the performance of AmoyDx® Master Panel on the MGI G50 platform. By comparing key performance metrics between the two platforms, this white paper explores the potential for expanding the use of AmoyDx® Master Panel beyond Illumina systems to the MGI G50 platform.

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Methodology

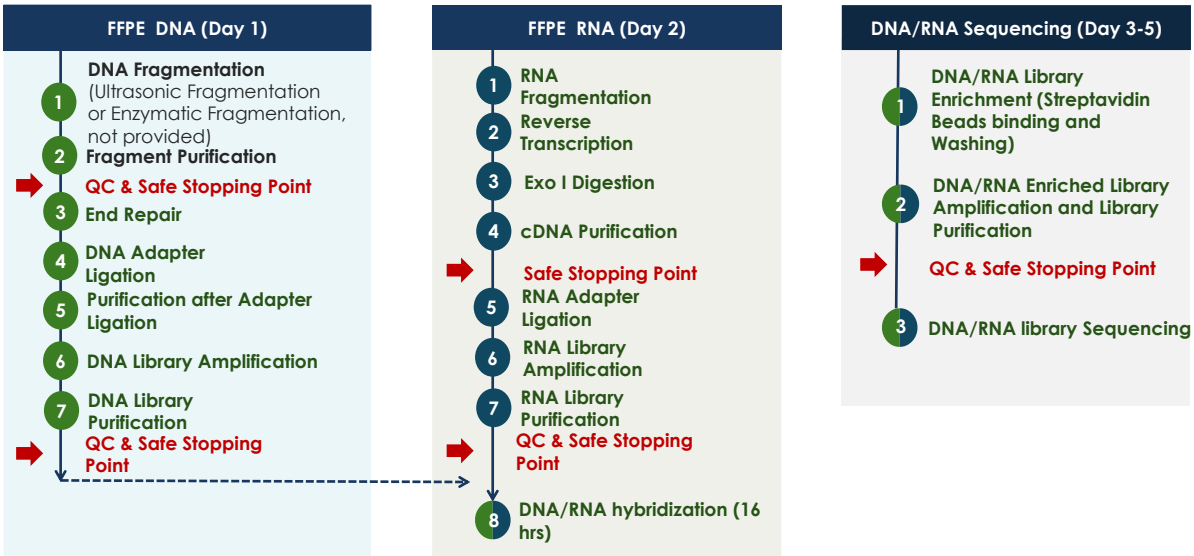
1. Sample Collection

61 DNA samples and 25 RNA samples were used for performance testing of the MGI G50 & NovaSeq 6000 in this report as stated below:

a) DNA samples: A total of 61 DNA samples, including cell line-derived reference materials, and FFPE samples from multiple types of cancers were tested.
b) RNA samples: A total of 25 RNA samples, including cell line-derived reference materials, and FFPE samples from multiple types of cancers were tested.

2. Extraction & Library Preparation

DNA extraction was conducted using AmoyDx® Magnetic FFPE DNA Extraction Kit. Library preparation was performed according to the AmoyDx® Master Panel protocol. Sequencing was carried out for each sample on both the MGI G50 and NovaSeq 6000 platforms for comparative analysis.



3. Data Processing and Analysis

Data analysis was conducted using the ADXMaster-DNA-Int module for DNA samples, and the ADXMaster-RNA-Int module for RNA samples. Key metrics including sequencing depth, quality control parameters, mutation detection efficiency, variant frequency, and biomarker analysis results were evaluated to assess performance.

Results

1. Sequencing and Data QC Performance

Sequencing and Data QC Performance Comparison for DNA Samples

Metric	MGI G50	NovaSeq 6000
cleanQ30	≥ 75%	≥ 75%
Coverage	≥ 95%	≥ 95%
HotUNIQUni-20%	≥ 90%	≥ 90%
NonHotUNIQUni-20%	≥ 80%	≥ 80%
HotUNIQDepth	≥ 1000×	≥ 1000×
NonHotUNIQDepth	≥ 500×	≥ 500×

Sequencing and Data QC Performance Comparison for RNA Samples

Metric	MGI G50	NovaSeq 6000
cleanQ30	≥ 75%	≥ 75%
Mapping	≥ 80%	≥ 80%
End2SenseRate	≥ 90%	≥ 90%
effectiveReads	≥ 4 Million	≥ 4 Million

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2. Variant Detection and Biomarker Analysis at the DNA-level

Comparison of Illumina NovaSeq 6000 and MGI G50 for Variant Detection at the DNA-level

Hotspot SNV/InDel (N=61)		Illumina NovaSeq 6000			Positive percent agreement (PPA)	Negative percent agreement (NPA)	Overall percent agreement (OPA)
		Positive	Negative	Total			
MGI G50	Positive	93	1	94	95.88%	100.00%	100.00%
	Negative	4	66636	66640			
	Total	97	66637	66734			

Fusion (N=61)		Illumina NovaSeq 6000			Positive percent agreement (PPA)	Negative percent agreement (NPA)	Overall percent agreement (OPA)
		Positive	Negative	Total			
MGI G50	Positive	8	0	8	100.00%	100.00%	100.00%
	Negative	0	53	53			
	Total	8	53	61			

CNV (N=59*)		Illumina NovaSeq 6000			Positive percent agreement (PPA)	Negative percent agreement (NPA)	Overall percent agreement (OPA)
		Positive	Negative	Total			
MGI G50	Positive	11	0	11	100.00%	100.00%	100.00%
	Negative	0	1737	1737			
	Total	11	1737	1748			

**2 samples were excluded as they are mixtures of multiple tumor cell lines and are not suitable for CNV analysis.*

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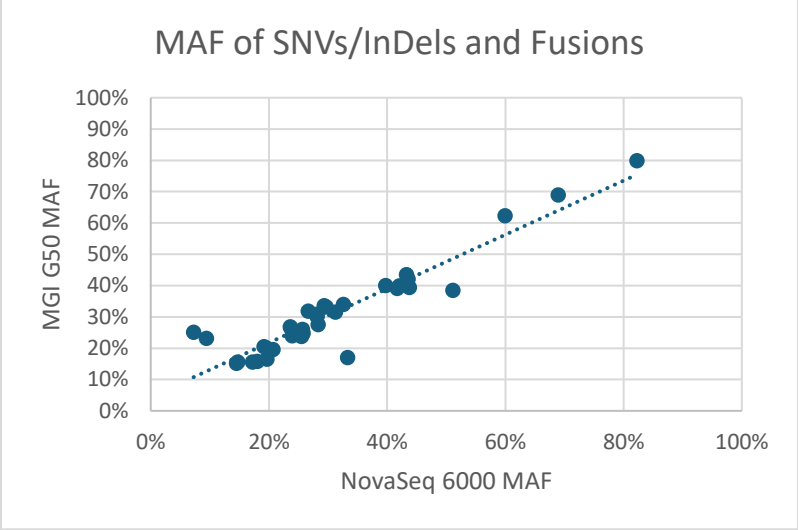
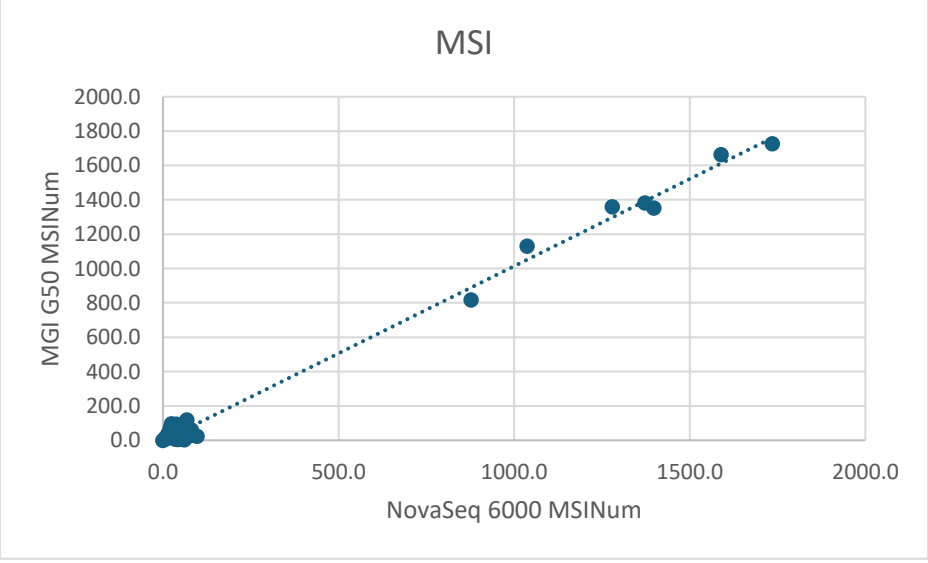
Allele Frequency Comparison of Illumina NovaSeq 6000 and MGI G50 for SNVs/InDels and Fusions at the DNA-level



Comparison of Illumina NovaSeq 6000 and MGI G50 for Biomarker Analysis at the DNA-level

For the determination of MSI, TMB, and GSS status, both sequencing platforms showed complete consistency across all tested samples, with PPA, NPA, and OPA all achieving 100%. The following figures illustrate the consistency analysis of each biomarker's specific values, all demonstrating high consistency.



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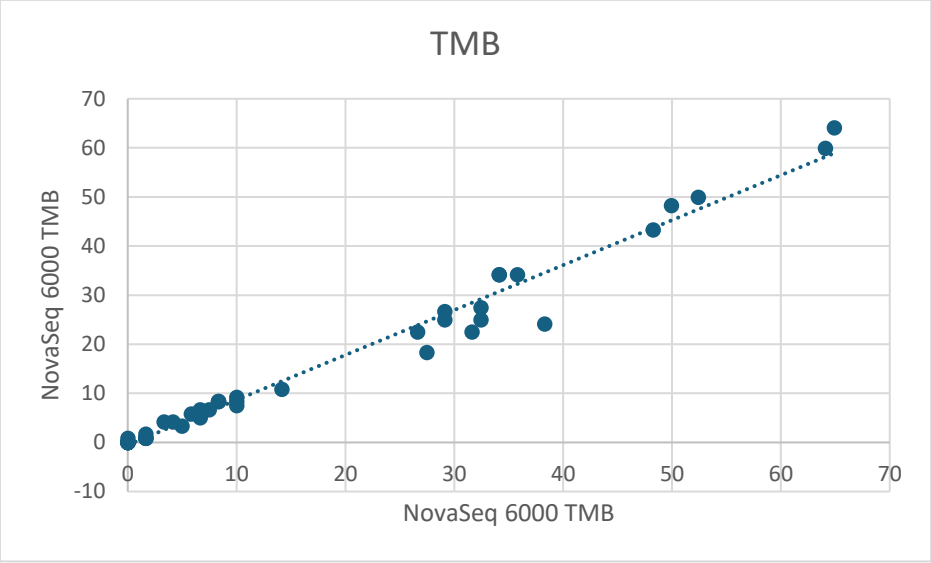
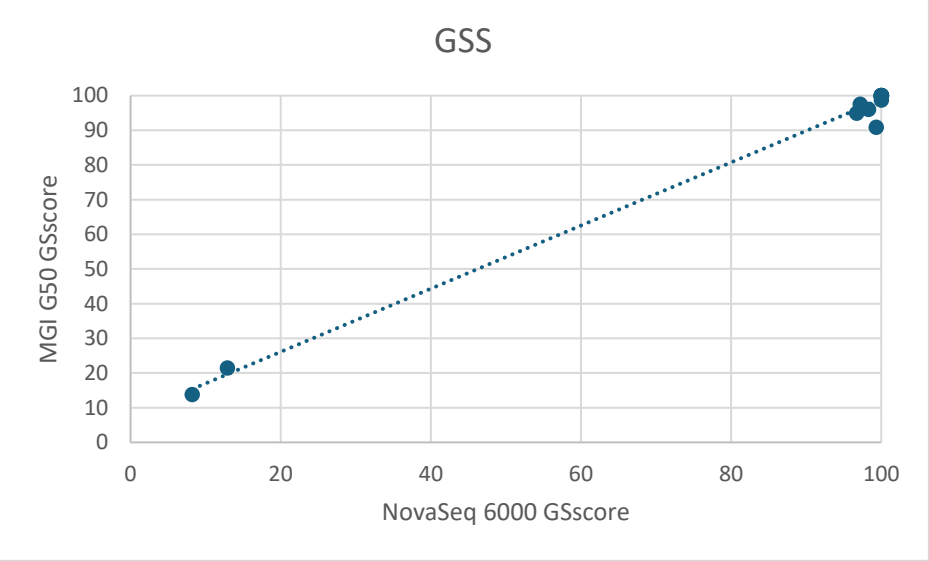




3. Variant Detection and Biomarker Analysis at the RNA-level

Comparison of Illumina NovaSeq 6000 and MGI G50 for Fusion Detection at the RNA-level

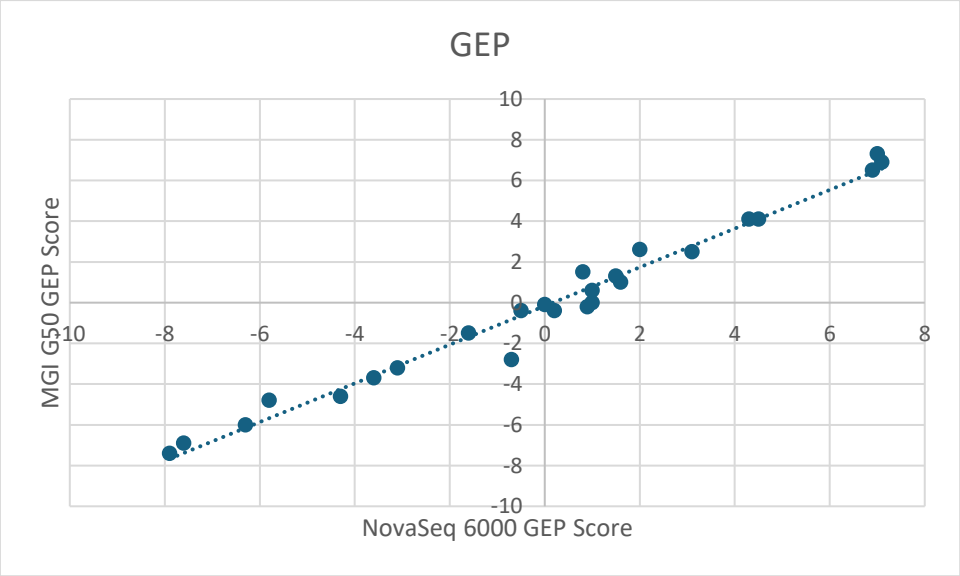
The 25 samples analysed for Fusion detection showed high consistency between two sequencing platforms, with an OPA at 99.93%:

Fusion (N=25)		Illumina NovaSeq 6000			Positive percent agreement (PPA)	Negative percent agreement (NPA)	Overall percent agreement (OPA)
		Positive	Negative	Total			
MGI G50	Positive	22	2	24	95.65%	99.95%	99.93%
	Negative	1	4325	4326			
	Total	23	4327	4350			

Note: All three inconsistent Fusions are near the Fusion detection cut-off.

Comparison of Illumina NovaSeq 6000 and MGI G50 for Biomarker Analysis at the RNA-level

The analysis of EBV and GEP status both achieved 100% consistency between the two sequencing platforms, while TME status were consistent for 24 out of 25 samples, showing a consistency of 96%. The following figure illustrates the consistency analysis of GEP scores, demonstrating high consistency between the two sequencing platforms.



Discussion

The comparative analysis highlights several key findings:

Sequencing Quality and Data QC Performance:

Both sequencing platforms demonstrated high sequencing quality and data QC performance for the analysis of both DNA and RNA libraries.

The CleanQ30 values exceeded 75% for both platforms. This indicates that both the MGI G50 and NovaSeq 6000 can generate accurate sequencing data suitable for downstream biomarker assessment and clinical analysis.

The quality control (QC) data on both MGI G50 and NovaSeq 6000 platforms met all QC criteria for the analysis of both DNA and RNA libraries, showing high concordance between the two sequencing platforms. This demonstrates the capability and reliability of the MGI G50 to match the performance of the NovaSeq 6000, enabling relevant variant detection and biomarker analysis on both DNA and RNA levels.

The consistency in sequencing quality and data QC values between the platforms suggests comparable base-calling accuracy and data QC performance, reinforcing the reliability of the MGI G50 for the variant detection and biomarker analysis for both DNA and RNA libraries.

Variant Detection and Biomarker Analysis Performance:

Variant detection and biomarker analysis performance on both DNA and RNA levels were comprehensively compared between MGI G50 and NovaSeq 6000. These two platforms demonstrated over 95% consistency across all analysed mutation types and biomarkers. This includes the detection of SNVs/InDels, Fusions, and CNVs, as well as the analysis of MSI, TMB, and GSS at the DNA level, and the detection of Fusions along with the analysis of EBV, GEP, and TME at the RNA level. These data confirm the high accuracy and consistency of the MGI G50 platform in variant detection and biomarker analysis performance at both DNA and RNA levels, positioning it as a reliable alternative to the NovaSeq 6000.

Conclusion

The study confirms that the AmoyDx® Master Panel performs exceptionally well on both MGI G50 and NovaSeq 6000 platforms. The MGI G50 demonstrated superior sequencing quality and data QC performance while maintaining comparable accuracy in variant detection and biomarker analysis performance at both DNA and RNA levels. These findings validate MGI G50 as a robust and reliable platform for comprehensive genomic profiling (CGP).