Fully optimized workflow of SOPHiA DDM Solid Tumor Solution on MGI Sequencing and Automation platforms offers a sample to result solution for highest convenience, quality and new insights from solid tumor samples

Highlight

- Automated library preparation on the MGISP-100 enables researchers to generate reliable and reproducible results with limited hands-on time.
- Together with the DNBSEQ-G50* which offers great coverage uniformity and output, researchers are gaining optimal sequencing capacity utilization.
- The expertly designed panel of 42 clinically relevant genes covering a wide range of solid tumors offers comprehensive variant detection through a capture-based assay and the tailor-made sequencer-specific analysis pipeline. Variant analysis, interpretation and reporting is performed in a single workflow powered by the SOPHiA DDM Platform.

Sample to result workflow

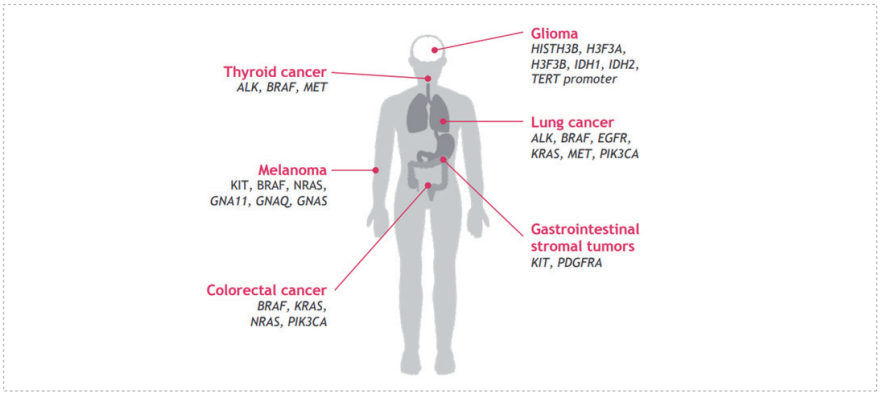
Researchers can successfully process FFPE or fresh-frozen samples from a broad range of tissue, requiring a minimum input of 10ng FFPE DNA (50ng is recommended).

Panel content

The panel content has been designed by experts in the field and consists of 42 genes selected in consultation with leading institutions to enable the detection of:

SNVs / Indels (all 42 genes)	Gene amplifications (24 genes)	MSI status (6 loci associated with colorectal cancer)

If more flexibility is needed, the gene panel can be fully customized and processed with the established workflow.



The following genes are included in the panel:

AKT1 (3)
ALK (21-25) ❖
BRAF (11,15) ❖
CDK4 (2) ❖
CDKN2A (1*,2,3) ❖
CTNNB1 (3)
DDR2 (18)
DICER1 (24,25)
EGFR (18-21) ❖
ERBB2 (8,17,20) ❖
ERBB4 (10,12)
FBXW7 (7-11) ❖
FGFR1 (12,14) ❖
FGFR2 (7,12,14) ❖
FGFR3 (7,9,14,16) ❖
FOXL2 (1*)
GNA11 (4,5)
GNAQ (4,5)
GNAS (8)
H3F3A (2*)
H3F3B (2*)
HISTH13B (1)
HRAS (2-4) ❖
KIT (8-11,13,17,18) ❖
KRAS (2-4) ❖
MAP2K1 (2,3)
MET (2,14-20) ❖
MYOD1 (1) ❖
NRAS (2-4) ❖
PDGFRA (12,14,18) ❖
PIK3CA (2*,3,6*,8,10,21) ❖
PTPN11 (3)
RAC1 (3)
RAF1 (7,10,12,13*,14*,15*) ❖
RET (11,13,15,16) ❖
ROS1 (38*,41*) ❖
SF3B1 (15-17) ❖
SMAD4 (8-12)
TERT (promoter*,1*,8*,9*,13*) ❖
TP53 (2-11) ❖
❖ Gene amplifications
* Hotspots only

Automated Library Preparation using the MGISP-100

Optimized convenience and reduced hands-on time is offered through the MGISP-100 liquid handler. The instrument contains an integrated PCR machine, magnetic rack and heating module which enables researchers to process 8- or 16-samples at a time. The intuitive deck setup is easy to navigate and a validated script as well as a dedicated user guide is available from MGI. This capture-based assay can be processed with limited hands-on time in 1.5 days using the MGISP-100.

Sequencing

MGI recommends sequencing the libraries generated with the SOPHiA DDM Solid Tumor Solution panel on the DNBSEQ-G50*. It is a compact bench-top sequencing platform that can adapt to the needs of the lab and is a perfect companion for the SOPHiA DDM Solid Tumor Solution. Researchers can sequence up to 48 samples per run on the FCS flow cell using PE100, generating 100M reads in less than 20h run time.

Performance data

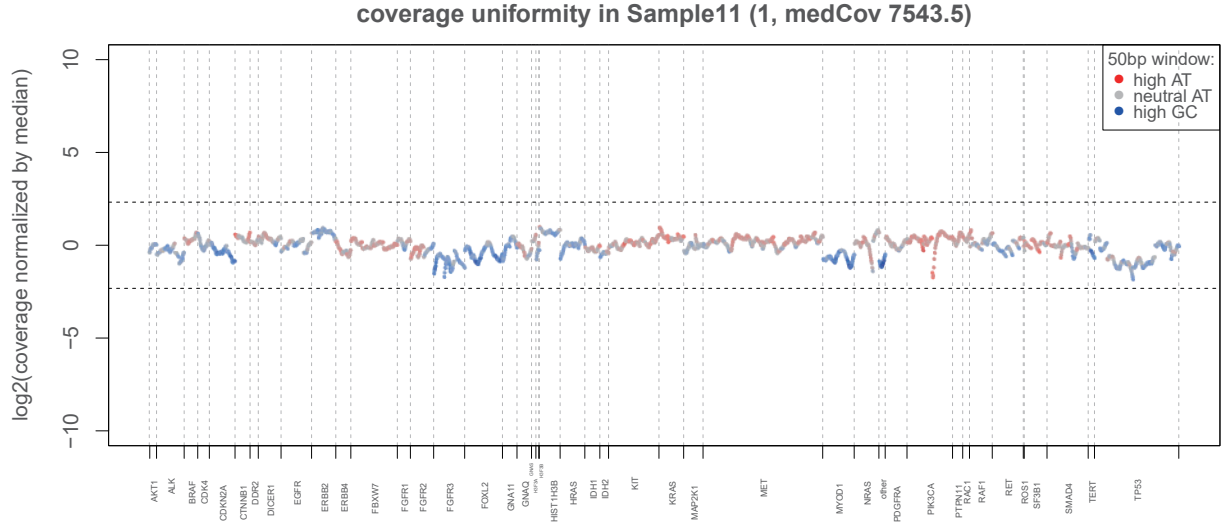
The SOPHiA DDM Platform analyzes complex NGS data by detecting, annotating and pre-classifying multiple types of genomic alterations in one unique experiment.

Analysis time from FASTQ: from 4 hours

	Observed (%)	Lower 95% CI
Sensitivity	98.59	86.19
Accuracy	99.99	99.97
Precision	100	91.89
Repeatability	100	100
Reproducibility	99.99	99.95
Coverage uniformity	99.7	100*

Performance values have been calculated based on SNVs and Indels only in 36 samples processed on MGI DNBSEQ-G50 using FCS PE100 sequencing set.
*5% quantile

High on-target rate and coverage uniformity throughout the target region. Maximize multiplexing and get confident variant detection even in "challenging" regions like TERT promoter C228T and C250T. Below is a representative sample from a run with 32 samples.



A true sample to result workflow, combining MGI's automation for library preparation, the DNBSEQ-G50* sequencer and a capture-based target enrichment kit with the intuitive SOPHiA DDM Platform.

Visualization and Reporting

The SOPHiA DDM Platform's user-friendly features facilitate variant visualization, simplify interpretation, and expedite reporting. Interpretation is further supported by access to the global SOPHiA GENETICS Community, one of the largest networks of healthcare institutions, where experts can indicate variant pathogenicity levels with the flagging feature.

Ordering information

Analysis time from FASTQ: from 4 hours

Model	Product Name	Intended Market	Part number
MGISP-100RS	DNA Sequencing Library Preparation System	CE RUO	900-000206-00
MGISP-100	DNA Sequencing Library Preparation System	CE IVD	900-000207-00
DNBSEQ-G50RS*	Genetic Sequencer DNBSEQ-G50RS (Config 2) Software version 4.0	RUO	900-000354-00
Sequencing Set	DNBSEQ-G50RS High-throughput Rapid Sequencing Set (FCS PE100)	RUO	1000019861

SOPHiA DDM Solid Tumor Solution is a Research Use Only product and is not intended for purposes other than research. Please contact SOPHiA GENETICS for ordering information.

MGI Tech Co.,Ltd.
Building 11, Beishan Industrial Zone, Yantian District,
Shenzhen, CHINA, 518083

400-096-6988
mgi-tech.com
MGI-service@mgi-tech.com

*Unless otherwise informed, StandardMPS and CoolMPS sequencing reagents, and sequencers for use with such reagents are not available in Germany, USA, Spain, UK, Hong Kong, Sweden, Belgium, Italy, Finland, Czech Republic, Switzerland and Portugal.