SOPHiA GENETICS™ | Element Biosciences

Expand your insights. Not your costs.

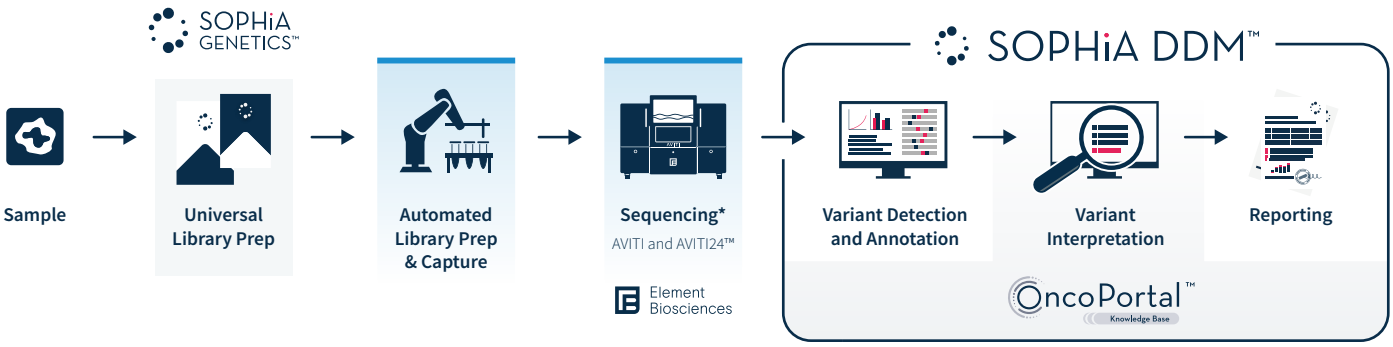
Comprehensive genomic profiling with SOPHiA GENETICS + Element Biosciences

Streamline your path from sample to decision with MSK-IMPACT® Flex powered with SOPHiA DDM™ and Element Biosciences AVITI/AVITI24™.

A fully integrated, end-to end offering

Bring comprehensive genomic profiling (CGP) in-house with a modular solution that combines content curated by MSK experts, cost-efficient sequencing, and deep AI-driven insights in a single workflow.

MSK-IMPACT® Flex powered with SOPHiA DDM™ + Element AVITI/AVITI24™ workflow



*Flow cells not provided for routine usage. Sequencers are available on a case-by-case basis.

Modular design

Tailor biomarker coverage at the sample level with flexible combinations of DNA capture (533 genes), RNA capture (139 genes), and low-pass WGS.

Cost efficient

Benefit from low cost per sample with Element AVITI/AVITI24™, making sequencing economical for labs at any scale.

Excellent performance

Achieve high analytical concordance to ground truth across variant classes with the AI-powered SOPHiA DDM™ Platform.

> *"From an economic standpoint, TomaLab has seen a huge advantage in cost savings on reagents. In fact, compared to the previous technology used in the lab, we've found that we've managed to cut costs by 30-40%.*

Dr. Francesca Dulcetti | Director, Laboratory of Medical Genetics | TomaLab, Italy

Based on experience with migration of SOPHiA DDM™ Hereditary Cancer Solution, Whole Exome Solution, and Clinical Exome Solution to Element AVITI™ Platform for research.

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For Research Use Only (RUO); RUO results must not be used for clinical diagnosis or patient management without further validation by the laboratory.

Flexibility at every step

MSK-IMPACT® Flex powered with SOPHiA DDM™ supports different sequencing modalities – DNA capture, RNA capture, and low-pass WGS – which can be analyzed separately or in any combination at the sample level, within a single run.

INPUT	BIOMARKERS
DNA capture	SNVs/Indels, MSI, TMB, CNVs (gene- and exon-level), gene fusions
Low-pass WGS	HRD genomic instability (GI)
Combined DNA capture and low-pass WGS	Tumor content & ploidy, allele-specific copy number (absolute copy number & LOH)
RNA capture	Gene fusions, exon skipping, gene expression

AVITI/AVITI24™ add flexibility in how you sequence, letting you seamlessly adapt to changing analysis needs and sample volume requirements.

Dual flow cells and **individually addressable lanes** allow for flexible, parallel sequencing and let different modalities run side by side.



High analytical concordance

MSK-IMPACT® powered with SOPHiA DDM™ performance with AVITI24™ was verified using real-world samples with established ground truth.

INPUT	BIOMARKERS	OPA	PPA	REFERENCE METHOD AND VERIFICATION COHORT
DNA capture				
	SNV	-	99.7%	**MSK-IMPACT®** (centralized test used at MSK) N=48 FFPE DNA (pan-cancer) – MSK cohort
	Indel	-	100%	
	MSI	97.9%	100%	
	TMB	100%	100%	
	CNV (gene-level)		93.5%	
	CNV (exon-level)	-	3/4[a]	**Myriad MyChoice®** N=24 FFPE DNA (ovarian cancer) – PAOLA-1 cohort
Low-pass WGS				
	HRD GI	100%	100%	
RNA capture				
	Gene fusion and exon-skipping	100%	100%	**Amplicon-based RNA assay** N=12 FFPE RNA (pan-cancer) – internal SOPHiA GENETICS cohort

[a]3 out of 4 positive events detected

≥97.7% concordance across biomarkers between Element AVITI24™ and Illumina NovaSeq™ X.

Ready to explore a cost-efficient, end-to-end workflow?
Contact your SOPHiA GENETICS representative to schedule an assessment for your institution.

CNV, copy number variant; FFPE, formalin-fixed paraffin-embedded; GI, genomic instability; HRD, homologous recombination deficiency; Indel, insertion and deletion; LOH, loss of heterozygosity; MSI, microsatellite instability; MSK, Memorial Sloan Kettering Cancer Center; OPA, overall percent agreement; PPA, positive percent agreement; SNV, single nucleotide variant; TMB, tumor mutational burden; WGS, whole genome sequencing.

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