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SOPHiA DDM™ for TSO500™

Enabling molecular laboratories to maximize the utility of Illumina's TSO500 panel

SOPHiA DDM™ for TSO500 is a fully integrated bioinformatic workflow (FASTQ to Report) for Illumina TruSight™ Oncology 500 panel. The SOPHiA DDM platform combines analytical performance* with streamlined interpretation of complex genomic variants in the context of comprehensive genomic profiling. Its intuitive interface and capabilities have been developed based on the feedback from our 1000+ customers. It offers several features to facilitate variant assessment and maximize the utility of genomic detection.

Analytical performance*

Detection of:

- SNVs & Indels in 523 genes
- Partner-agnostic detection of 55 fusion genes with full coverage of NTRK 1,2,3 fusions and splice variants
- CNVs in 495 genes (versus 59 genes with standard analysis solutions) [1]

Comprehensive immuno-oncology biomarkers assessment:

- Two TMB scores (mut/Mb) reported [2]
- MSI with qualitative results, benchmarked to PCR

Full access to analytical output:

- Ability to review the calculation behind MSI and TMB results access to source file and quality reports for analysis

The pipeline is compatible with Illumina NextSeq® sequencers.

Analysis time from FASTQ files: 8 hours [11]

	Performance metrics*[3]	Observed	Lower CI (95%)
DNA Variants			
SNV & Indels [4]	Sensitivity	95%	92.30%
	Precision	96%	93.40%
CNV/Amplifications [5]	Sensitivity	92.90%	66.13%
RNA Variants			
Fusions & Splice Variants [6]	Sensitivity	96.38%	91.8%
	True Positive	133/138	-
	False Negative	5/138	-
Biomarkers			
TMB [7]	Concordance (synonymous and non-synonymous mutations)	R2=0.994	-
	Concordance (non-synonymous mutations only)	R2=0.995	-
MSI	Sensitivity [8]	100%	86.80%
	Specificity [9]	100%	92.60%
	Precision [10]	100%	95.10%

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

The platform helps users to immediately focus on relevant and actionable genomic alterations. Several features facilitate their interpretation process:

- Hotspot screening
- Algorithm-supported variant pre-classification
- Fully customizable filters
- Comprehensive report

Confident decision-making

The SOPHiA DDM™ platform integrates the OncoPortal™. This feature provides the latest scientific evidence on the actionability and significance of each genomic alterations to support informed decision for research purpose.

Global support at every step

We offer local support available globally. Our Set Up Program provides assistance with assay setup for fast and worry-free transition to routine testing. All along the usage, our robust support including re-analysis and dedicated bioinformaticians help save time and resources, ensuring fast resolution of workflow disruptions.

Secure and unlimited data storage

The SOPHiA DDM™ platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with your local data security policies.

Access to SOPHiA community

Through the SOPHiA DDM™ platform genomics experts from >1000 healthcare institutions interpret their findings and flag the pathogenicity level of variants. This highly valuable information enriches the variant knowledge base and is safely shared among the members of the community, supporting their decision-making process for research purposes.

Match genomic profiles with clinical trials opportunities

SOPHiA DDM™ can support the identification of subjects who could benefit from investigational therapies. If the institution chooses to activate the feature, when a genomic profile matches a research opportunity, SOPHiA DDM™ notifies relevant healthcare professionals. As a result, identified subjects are offered to participate in corresponding clinical trials, contingent on eligibility assessment and consent.

1. Based on benchmarking studies comparing SOPHiA GENETICS with a standard analysis
2. Including all synonymous & non-synonymous and non-synonymous mutations
3. Performance metrics calculated in samples processed on Illumina NextSeq® 500/550
4. Based on analysis of 504 variants in the Acrometrix™ reference standard
5. Based on analysis of 14 amplifications variants in 9 FFPE samples including 5 different tumor types
6. Based on analysis of 65 confirmed fusions from 105 clinical and reference samples
7. Concordance with TSO500 LocalApp v2.2 comparing Mut/Mb in 64 samples, including 53 clinical FFPE tissue samples from more than 10 tumor types
8. Based on analysis of 26 MSI-H samples from at least 2 tumor types with MSI status determined by PCR or using Illumina BaseSpace apps
9. Based on analysis of 48 MSS samples from at least 8 tumor types with MSI status determined by PCR or using Illumina BaseSpace apps
10. Based on analysis of 26 MSI-H and 48 MSS samples from at least 8 tumor types with MSI status determined by PCR or using Illumina BaseSpace apps
11. Analysis time may vary depending on the number of genes, samples analyzed and server load

Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™).

***For Research Use Only. Not for Use in diagnostic procedures.**