



**Bioinformatic Platform for MSB workflows**



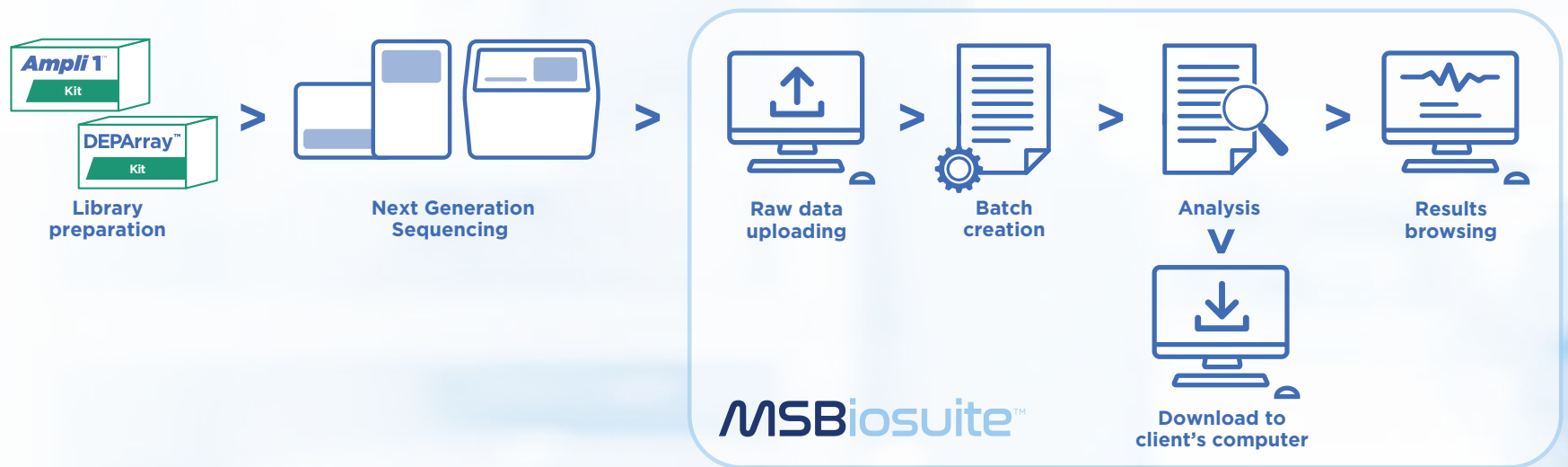
**MENARINI**  
silicon biosystems

## Completing our sample to results solution

**MSBiosuite** is a cloud based platform which automates processing of sequencing data generated from **Ampli1™** and **DEPArray™** NGS kits.

MSBiosuite manages your entire workflow from uploading of raw data, data processing, to the downloading of results directly onto your computer. The tailored proprietary analysis pipeline guarantees highly confident variant calling and generation of copy number aberration profiles.

### MSBiosuite workflow



## Two level of reporting available



**Analytical report with basic annotation of variant calling**



**Clinical interpretation report with advance annotation**  
(available for OncoSeek pipelines only)

## Bridge the gap between your raw NGS data and consolidated results

### Key Features



#### EASY DATA PROCESSING

- Automated analysis of sequencing data generated from *Ampli*™ and *DEPArray*™ NGS kits
- No bioinformatic skills required



#### USER FRIENDLY

- Custom graphical views of browsing results
- Remote support always available
- Easily share results of specific analysis runs with colleagues and collaborators



#### SECURE

- Data are encrypted both “in transit” and “at rest”
- Strong authentication mechanisms



#### DATA RESIDENCY

- Global data center deployment
- Processing and storage of genomics data are guaranteed to be in the users’ region of choice



#### AFFORDABLE

- Pre-paid analysis packages and monitoring of remaining analysis
- No hardware costs or license fees

# MSBiosuite pipelines for single cell application



## Ampli<sup>1</sup>™ LowPass pipeline



Sample of interest can be easily selected for a visual comparison of multiple copy number profiles (Fig 1). An interactive searchable and zoomable display allows browsing specific genomic regions. A hierarchical clustering of the samples included in the batch is provided to help in investigating the relationships between samples and to study tumor heterogeneity (Fig 2). Data and publication quality figures can be downloaded to your computer.

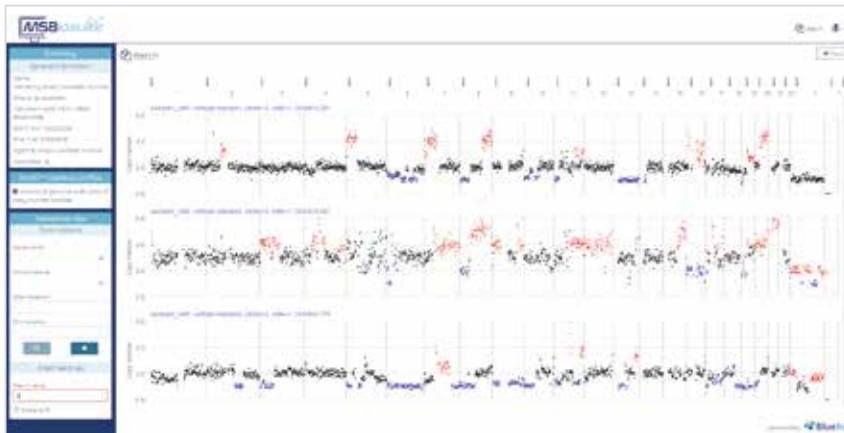


Fig 1 Each track corresponds to the copy number profile of the selected single cell. The track header includes sample name, cell type, ploidy, cell number and DLRS information. In the plot, absolute copy numbers (y-axis) are displayed along the genome chromosomes (x-axis).



Fig 2 Hierarchical clustering is displayed as a dendrogram on the left side of a heatmap showing the copy number profiles arranged in rows, where copy-gains and losses are highlighted using a red and blue gradient respectively.

# MSBiosuite pipelines for single cell application



## Ampli<sup>1</sup>™ OncoSeek pipeline



Putative somatic variants are listed and grouped by sample.

The allelic frequencies of each variant and basic annotation are also listed (Fig 3).

A more comprehensive list of all variants, both somatic and germline, listing also putative LOH events is available as download in Excel format.

Variant ID	Position	Allelic Frequency	Annotations
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)
chr17:78000000	78,000,000	0.05	... (somatic variant)

Figure 3. Somatic variants list

The copy number amplifications section shows, both in text form and as a barchart (Fig. 4), the copy number amplifications detected in 19 target genes, grouped by group/patient.

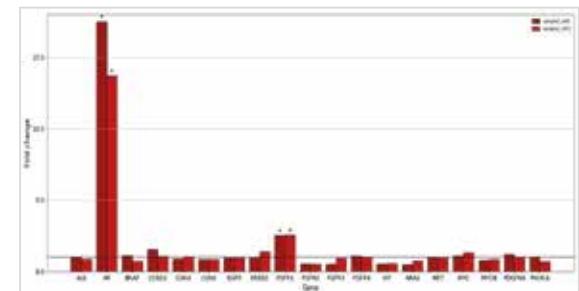


Figure 4. Barchart of copy number amplifications. Copy number gains whose fold change value is >2 and >5 Z-score calculated on reference samples are highlighted with an asterisk.

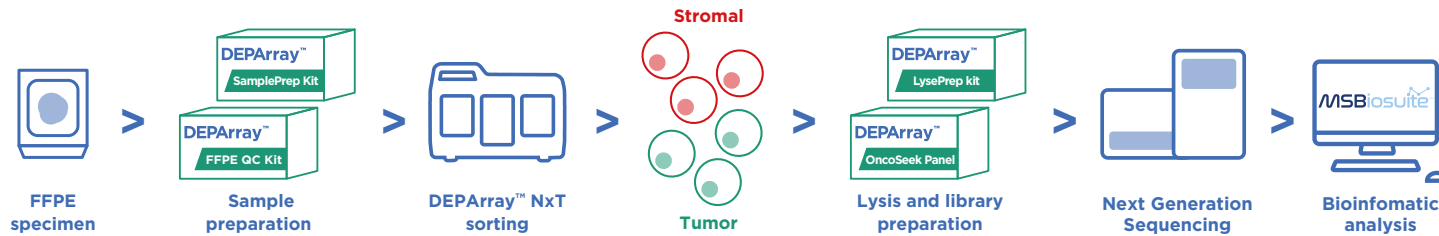
Optional advanced annotation is available on demand for OncoSeek pipeline. (Powered by N-Of-One)



# MSBiosuite Pipeline for FFPE Application



## DEPArray™ OncoSeek pipeline



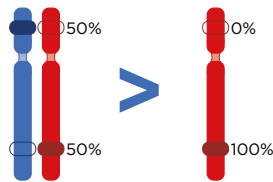
Visualize putative somatic variants with basic annotation and a table/barchart view of focal copy-number amplifications (see Fig.3 and 4 as an example). Result files can be downloaded to your computer and provide more comprehensive insight, including information on germline and LOH variants, target coverage and additional annotation.

A variant report table can be derived providing unambiguous identification of genetics event in each cell population (Fig. 5).

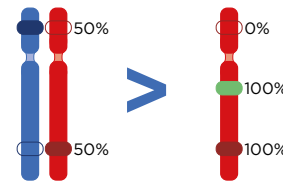
gene	chromosome	position	reference	alternative	stromal		tumor		unsorted	event
					lib1	lib2	lib3	lib4	lib5	
PDGFRA	chr4	55.149.258	G	A	55,22	43,54	0,00	0,00	15,29	LOH
KIT	chr4	55.563.683	T	G	40,42	71,27	100,00	100,00	91,34	LOH
CDK6	chr7	92.313.733	A	G	82,94	56,58	0,00	0,00	17,78	LOH
CDK6	chr7	92.333.408	A	G	50,93	32,86	0,00	5,26	19,05	LOH
TP53	chr17	7.577.094	G	A	0,00	0,00	100,00	100,00	56,01	SOM + LOH

Fig. 5. Pure tumor and stromal populations from primary adenocarcinoma were sorted and analysed with the DEPArray FFPE workflow. Relevant variants in the sorted pure populations of tumor (orange), stromal (blue) cells and unsorted fractions (green) are shown in the table. Loss Of Heterozygosity (LOH), somatic variant (SOM) and dual events (SOM + LOH) are easily detected in the sorted cell populations. Adapted from a case report, Isidori F et al., BMC Cancer, 2018.

LOH



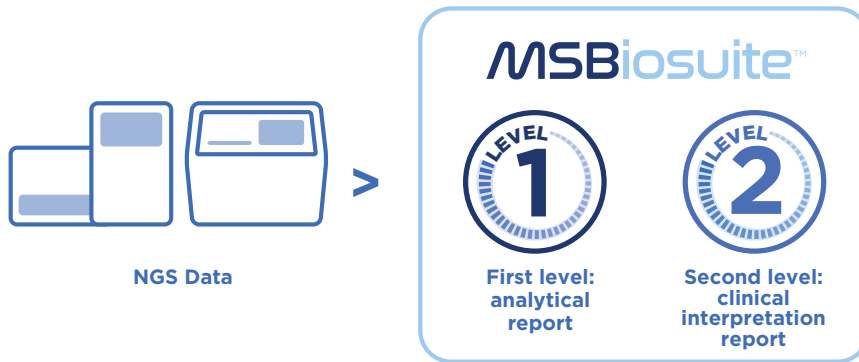
SOM + LOH



Optional advanced annotation is available on demand for OncoSeek pipeline. (Powered by N-Of-One)



# Clinical interpretation report with advanced annotation for Oncoseek pipelines



The service takes advantage of RapidInsights™ by clinical interpretation company N-of-One. The service includes also TrialMatch™, N-of-One proprietary process for incorporating deep scientific and clinical analysis of each patient's molecular data, to identify and prioritize the best clinical trial options for that patient.\*

## Information provided:

- Clinically relevant alterations divided in variants of strong, potential and uncertain clinical significance
- Predictive, prognostic and diagnostic level of evidence
- For each predictive variant listed the therapies approved in the specific indication and in other indications, and if the variant may indicate resistance to therapy
- Detailed biomarkers informations
- Matching clinical trial prioritized by clinical specificity and by region.



\*The content contained herein (Content) is a publication of Menarini Silicon Biosystems (MSB). MSB does not provide medical services, nor is any MSB employee engaged in the practice of medicine for or on behalf of MSB. Some tests, drugs and biomarkers identified in this report may not be approved for a particular use or validated for that use. The Content is compiled from sources believed to be reliable. Extensive efforts have been made to make the Content as accurate and as up-to-date as possible. Despite our best efforts, the Content may contain typographical errors and omissions. The Content is for professional medical and scientific use only. N-of-One, Inc. has provided to Menarini Silicon Biosystems the research, analysis and interpretation in this report. Additional information about N-of-One is available on its website at [www.n-of-one.com](http://www.n-of-one.com)

## Ordering information

MSBiosuite products	Code
<b>MSBiosuite <i>AmpI</i><sup>™</sup> LowPass Analysis (Ion Torrent)</b>	KI0129 (16 samples), KI0130 (48 samples), KI0131 (320 samples)
<b>MSBiosuite <i>AmpI</i><sup>™</sup> LowPass Analysis (Illumina)</b>	KI0132 (16 samples), KI0133 (48 samples), KI0134 (320 samples)
<b>MSBiosuite <i>AmpI</i><sup>™</sup> Oncoseek Analysis</b>	KI0138 (9 samples), KI0139 (24 samples), KI0140 (120 samples)
<b>MSBiosuite DEPAArray<sup>™</sup> OncoSeek Analysis</b>	KI0135 (9 samples), KI0136 (24 samples), KI0137 (120 samples)
<b>Clinical Interpretation Report (available only for OncoSeek panels)</b>	KI0141 (1 sample)

Valid only for EU Customers.

*AmpI*<sup>™</sup> Kits, DEPAArray<sup>™</sup> Kits and MSBiosuite<sup>™</sup> are for research use only, not to use in diagnostic procedures.

MSBiosuite<sup>™</sup> is powered by BlueBee

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