



## SeqOne platform in Oncology

- A comprehensive software solution for analyzing somatic variants based on DNA, RNA or ctDNA
- To provide support for diagnosis and therapeutic decision, SeqOne platform detects several types of somatic variants including:
  - Single Nucleotide Variant (SNV), Multi Nucleotide Variant (MNV), and short indels
  - Splicing event and gene fusion for RNA sequencing

## Data-specific solutions



**RNA** solution for splicing events, SeqOne platform provides functional analysis of the impact of splicing in the new protein



**ctDNA** Bioinformatics pipeline have been tuned to be able to detect very low variant allele frequency



### DNA

- MSI and TMB Computation of biomarkers to determine patients eligible to targeted drugs including Tumor Mutational Burden (TMB) and Microsatellite instability (MSI)
- CNV Call of Copy Number variation (CNV) when cohorts of individuals are available
- ALU insertion Detection of ALU insertion, medium-size deletions and duplications (<300 bp) including FLT3-ITD variation

## Advantages of SeqOne platform for Oncology

- Compatibility SeqOne bioinformatic pipelines are compatible with most gene panels and have been designed to be agnostic to specificities of the wet lab
- Annotation Fusion, splicing events & SNV are enriched with annotations arising from several knowledge databases including CIVIC, OMIM, CLINVAR, GNOMAD and COSMIC
- Pooled analysis in which SeqOne platform computes variant frequency for a sample of individuals to detect sequencing artifacts
- Drug and trial recommendation provided for each variant based on the Molecular Match knowledge database
- Pathogenicity prediction provided with ACMG classes using ComPerMed framework, which is a technical guideline to harmonize clinical interpretation of variants
- UMI adapters SeqOne platform implements dedicated platform to account for UMI adapters:
  - UMI-specific pipeline increase variant detection sensitivity
  - UMI-specific pipeline is critical to improve SNV calling precision for samples with very low tumoral content and ctDNA

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## From FastQ to final report

A true end-to-end solution for efficiency and traceability from raw data (FastQ or Dragen VCF) to easily understandable and actionable clinical report.

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## Supports both somatic and constitutional analysis

SeqOne supports both somatic and constitutional analysis on a unified platform

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## Bioinformatic excellence

SeqOne's platform offers advanced bioinformatics that detect complex variants and support all the important usages including UMIs for liquid biopsy analysis, noise and artefact reduction for a lower false positive rate

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## Easy-to-use interpretation environment

An intuitive visual environment providing tools to boost interpretation efficiency. SeqOne's platform offers specific applications to address the requirements of each genomic test

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## ML driven variant classification

Biological and clinical pre-classification based on European International Guidelines (ACMG, AMP, ComPerMed) and on machine learning systems

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## Quality control

QC base-per-base coverage ensuring true negative assertion

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## World-class support

Qualified support team of genomic specialists Method validation for accreditation bodies

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## Certification

A platform built for clinical diagnostic routine: CE-IVD, HDS and ISO27001 certified

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## Data security

SeqOne stores data on an ISO27001 compliant platform and offers additional user-specific data encryption to ensure data stays safe

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## Up-to-date knowledge

Regularly updated molecular and clinical knowledge bases (biological, clinical and therapeutic) with update notification to enable retrospective analysis of variants, treatments and clinical trials.