



# Comprehensive Cancer Panel

Unlock the power of precision medicine and personalized cancer treatment

Despite the singular nomenclature, cancer is commonly considered as a diverse spectrum of diseases distinguished by the unrestrained proliferation and dissemination of atypical cells.

In precision medicine and personalized cancer treatment, understanding cancer genes and genome-wide copy number alterations in solid tumors gives valuable insights. These can help identify actionable variants, assess potential therapeutic strategies, and stratify patients for clinical trials. Consequently, such knowledge empowers healthcare professionals to make informed treatment decisions, ultimately leading to enhanced patient outcomes.

Detecting genetic variants in formalin-fixed paraffin-embedded (FFPE) tumor samples can be difficult due to technical artifacts associated with the formalin fixation process. This process can cause several types of DNA damage including fragmentation, cross-linking of DNA strands, and chemical modifications.

TruSight Oncology 500 DNA high-throughput (TSO500) (Illumina) is a next-generation sequencing (NGS) assay that enables in-house comprehensive genomic profiling of solid tumor samples. Its primary function involves converting DNA isolated from solid tumor samples into enriched libraries, targeting cancer-related genes with exceptional sensitivity, specificity and reproducibility.

The combination of a streamlined workflow together with a state-of-the-art bioinformatics pipeline makes GenomeScan's Comprehensive Cancer Panel solution an ideal choice for clinical laboratories and researchers seeking to advance precision medicine and personalized cancer treatment.

## Comprehensive Cancer Panel Workflow

Our workflow incorporates library preparation, quality control steps, and short-read sequencing by using the Illumina NovaSeq sequencing platform. This workflow has been validated using multiple types of input material, including fresh frozen tissue and FFPE, ensuring accuracy in our results.

### Comprehensive Cancer Panel Workflow

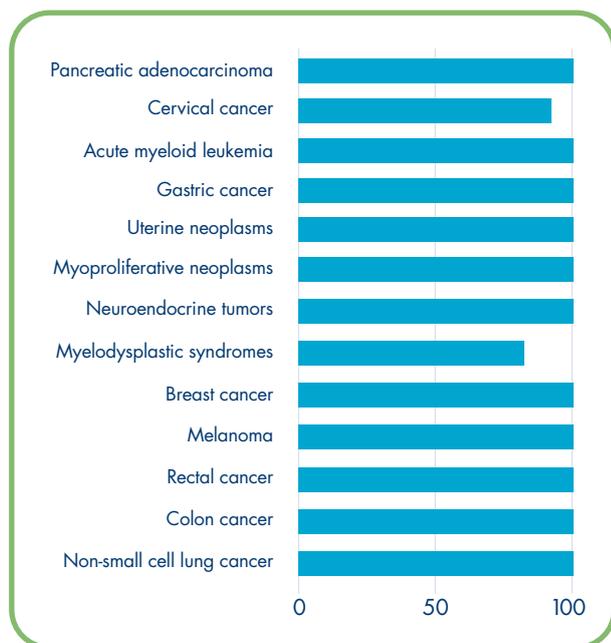


The Panel targets 523 genes that are frequently implicated in cancer, with high sensitivity and specificity, and provides uniform coverage of the target regions, which ensures reliable detection of low frequency variants in all target genes. Moreover, our sophisticated algorithms enable accurate analysis of tumor mutational burden (TMB) and microsatellite instability (MSI).

### Key benefits

- Analysis of hundreds of current and emerging cancer-related biomarkers, including immuno-oncology (IO) markers.
- Replace multiple tests with a single comprehensive panel.
- Fast turn-around-time.
- High sensitivity (>96%) and specificity (99.99%).
- Dedicated bioinformatics workflow.
- Flexibility in sample types, including FF and FFPE.

The data generated by our Comprehensive Cancer Panel can provide valuable insights for a wide range of clinical applications, such as cancer diagnostics, monitoring disease progression and treatment efficacy, identification of novel therapeutic targets and biomarkers, facilitation of precision medicine and personalized treatment planning.



The percentage of genetic markers generated by our Comprehensive Cancer Panel.

### Input material

#### Samples

- Fresh frozen (FF)
- Formalin-Fixed Paraffin-Embedded (FFPE)
- Fine-needle aspirates (FNAs)
- Core needle biopsies

#### Isolated genomic DNA

- Optimal DNA requirement:  $\geq 100\text{ng/sample}$
- DNA Purity: OD260/280 ~1.8-2.0 and OD260/230 ~2.0-2.2

#### Illumina NovaSeq Sequencing

- Paired-End 150 bp (PE150)
- Reads: 40 million paired-end reads/sample

#### Deliverables

- FastQ files
- Quality Score Q30  $\geq 80\%$
- Comprehensive Cancer Panel report

### Trustworthy results

#### Data quality guarantee

The purpose of data QC is to validate the precision and accuracy of your results. It helps to reduce the time you spend on sample verification.

#### ISO 15189 and ISO/IEC 17025 accreditation

GenomeScan strives for excellence and is committed to driving up standards in the field of NGS services. For our customers, this means consistency in our level of quality, service, and support.

#### Professional reporting

We deliver a comprehensive report that includes important information about the genetic alterations and biomarkers identified in the tumor sample.

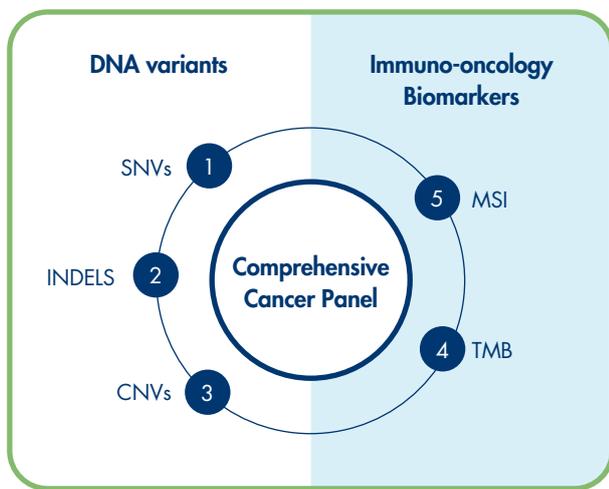
### Data analysis

Our data analysis workflows are designed using cutting-edge bioinformatics tools, ensuring the extraction of meaningful insights from tumor genomic data. With a focus on accuracy and precision, our state-of-the-art methodologies enable comprehensive analysis that uncovers crucial genetic information relevant to your project. From alignment and variant calling to annotation, our robust bioinformatics pipelines deliver reliable results empowering informed decision-making in cancer genomics.

## Biological Insights

The biological insights that can be inferred from your data include:

- Identification of genomic alterations, including single nucleotide variants (SNVs), small insertions/deletions (indels), copy number variations (CNVs) and structural variants.
- Tumor heterogeneity by identifying subclonal populations and their genetic alterations within a tumor sample.
- Biomarkers associated with specific cancer types or treatment responses.
- Estimation of Tumor Mutational Burden (TMB).
- Microsatellite Instability (MSI) assessment.



Comprehensive Cancer Panel insights.

## About GenomeScan

As an ISO-accredited leading Dutch Next Generation Sequencing service provider, GenomeScan develops customizable NGS solutions for pharmaceutical and biotech companies, healthcare providers and academic institutions. By providing state-of-the-art tools to analyze genetic disorders fast, affordably, and effectively, GenomeScan fosters innovation through partnership with medical centers and research laboratories.

## Explore related resources

**DNA Service Specification Sheet**

GenomeScan

Long Read, Methylation Arrays, Targeted, WES, WGS

### Whole Genome Sequencing (WGS)

The most comprehensive sequencing analysis with the widest coverage

Whole Genome Sequencing (WGS) provides the most comprehensive genetic blueprint of an organism. WGS is often combined in a multi-omic analysis, for example in the investigation of rare diseases of unknown cause. Genomics offers an unbiased (SCV/SCN/TCV) WGS service across a multitude of diagnostic, therapeutic and research applications. WGS not only provides unique insights into disease mechanisms, but it also generates genomic data (chromosomes) to Whole Exome Sequencing (WES) generating data for only 1-2% of the genome. It offers the ability to identify copy number variations, insertion-deletions (indels), rearrangements and other structural variants. It helps you make informed decisions for the next step of your research project and offers you the advantage of re-analyzing and re-sequencing your data at any time.

To bring diverse individual sequencing objectives, we are able to analyze hundreds of samples in parallel delivering the best price and highest reproducibility, making WGS a very powerful and cost-effective tool for genetic profiling.

**Input material**

**Isolated genomic DNA**

- Required input:
  - without PCR:  $\geq 25 \text{ ng}$  / sample
  - with PCR:  $\geq 1 \text{ ng}$  / sample
- Minimum volume: 20  $\mu\text{l}$  / sample
- Quality: High Molecular Weight DNA
- Custom: FFPE, repeat available

**Sequencing on Illumina NextSeq PE 150**

- PCR free library preparation ( $\geq 25 \text{ ng}$ )
- Unique Molecular Identifiers for samples
- Unique dual indexed sequencing adapters

**Deliverables**

- FastQ files
- Quality score Q30 of  $\geq 80\%$
- Customized data analysis with comprehensive report

\*Higher input is required for 100-ug for deep sequencing.

**NGS Laboratory workflow**

- ← Sample delivery
- Sample Entry QC
- Library Preparation (sequencing adapters, Unique Molecular Identifiers)
- Data QC
- ← Sample Report

GenomeScan

### Enhanced sequencing with UMIs

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Unique molecular identifiers, or short UMIs, are here to stay. A technology introduced almost ten years ago [Kozma et al., 2012] has changed how we perform and analyze next generation sequencing (NGS) experiments today.

**Polymerase chain reaction: friend or foe?**

In the majority of currently available sequencing methods, library preparation relies on nucleic acid fragmentation followed by size selection and template amplification via polymerase chain reaction (PCR) (Figure 1). The amplification step allows us to exponentially increase the amount of starting material, thereby increasing the library quantity and ensuring that each molecule in our original sample is sequenced. This is particularly important when working with low-input samples, despite the obvious advantage of amplification, this reaction introduces bias in applications that rely specifically on "counting" of amplified molecules, for instance in RNA sequencing or copy number variation studies [Ditts et al., 2017]. The efficiency of PCR amplification is dependent on the sequence of the amplicon [Kuznetsov et al., 2007] which, in turn, determines the level of inhibition by self-sequencing [Aronis et al., 2005]. In practice, if one sequence is amplified 20x, more than another it will be 237 [(1.20)<sup>20</sup>] times more abundant after 30 rounds of amplification.

By definition, the PCR will produce identical copies of the starting material. However, these copies do not represent the original abundance of the transcripts; we refer to them as duplicates. These are not "true copies" of our targets, and it is in our best interest to either remove or outbalance them. PCR duplicates can be identified computationally by their mapping coordinates. However, it has been shown that computational removal of duplicates underestimates the abundance of transcripts, specifically those that are short or highly expressed [Fu, Y. et al., 2018]. A gene can be mistakenly identified as being expressed because we partially removed its true copies while trying to eliminate PCR duplicates. Thus, removal of duplicates that relies solely on their coordinates may negatively affect downstream analyses.

Figure 1. Library preparation and sequencing workflow.

## Contact us

If you are looking for the most accurate services, look no further. Contact us today to learn how we can help you.