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 the context of precision medicine and personalized cancer treatment, the characterization of cancer genes and the analysis of genome-wide copy number alterations in solid tumors can provide valuable insights to clinicians. These insights, include the identification of actionable variants, assessment of potential therapeutic strategies, and patient stratification for clinical trials. Consequently, such knowledge empowers healthcare professionals to make informed treatment decisions, ultimately leading to enhanced patient outcomes.

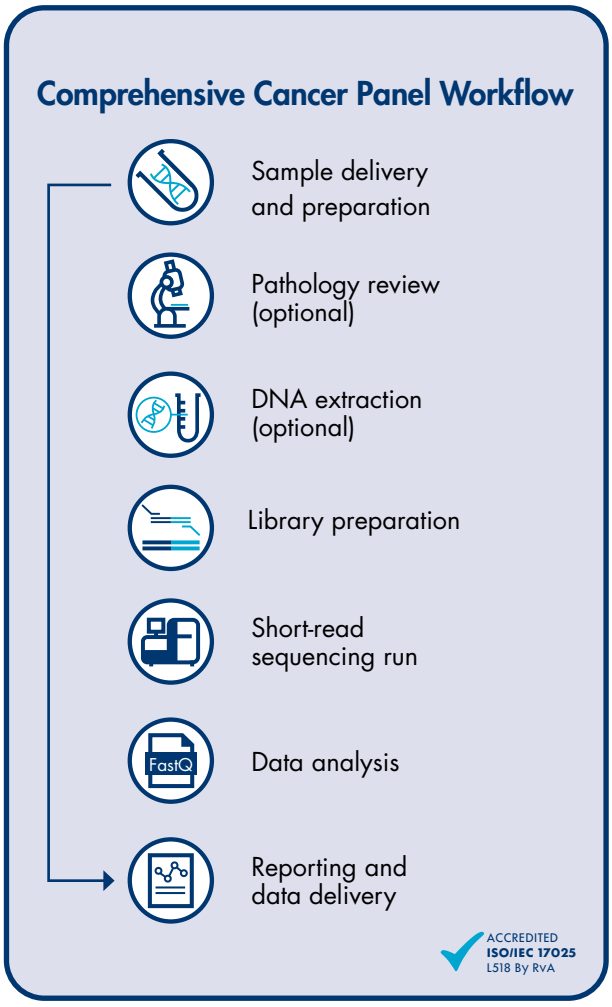
Detecting genetic variants in formalin-fixed paraffin-embedded (FFPE) tumor samples can be difficult due to technical artifacts associated with the formalin fixation process. This process can cause several types of DNA damage including fragmentation, cross-linking of DNA strands, and chemical modifications.

TruSight Oncology 500 DNA high-throughput (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.

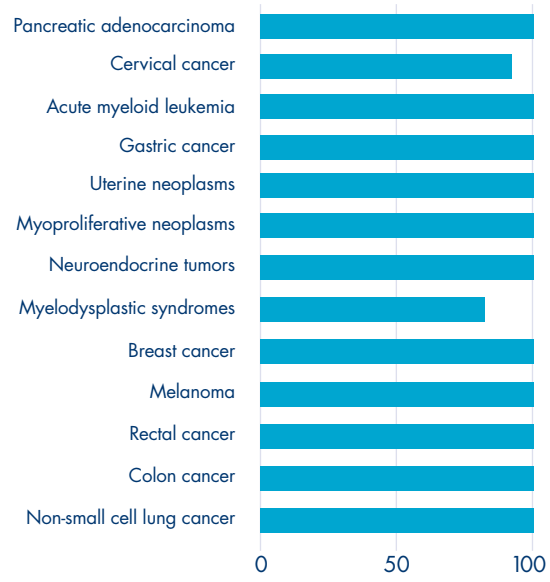


The Panel targets 523 genes that are frequently implicated in cancer, with high sensitivity and specificity, and provides uniform coverage of the target regions, which ensures reliable detection of low frequency variants in all target genes. Moreover, our sophisticated algorithms enable accurate analysis 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: ≥100ng/sample
- DNA Purity: OD260/280~1.8-2.0 and OD260/230 ~2.0-2.2

Illumina NovaSeq Sequencing

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

Deliverables

- FastQ file
- Quality Score Q30 ≥ 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.

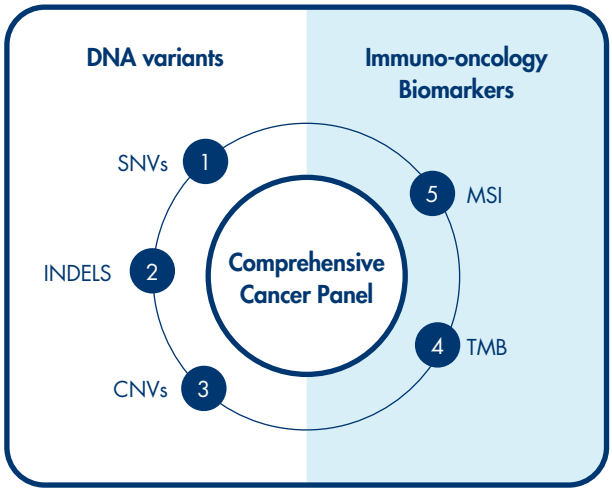
Data analysis

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 that empower informed decision-making in the field of 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.

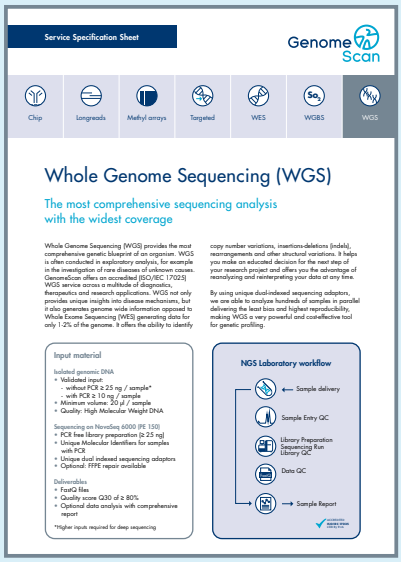


Comprehensive Cancer Panel insights.

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


Service Specification Sheet

Genome Scan

Chip | Longreads | Methyl arrays | Targeted | WES | WGBS | 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 conducted in exploratory analysis, for example in the investigation of rare diseases of unknown causes. GenomeScan offers an accredited (ISO/IEC 17025) WGS service across a multitude of diagnostics, therapeutics and research applications. WGS not only provides unique insights into disease mechanisms, but it also generates genome wide information opposed to Whole Exome Sequencing (WES) generating data for only 1-2% of the genome. It offers the ability to identify copy number variations, insertions-deletions (indels), rearrangements and other structural variations. It helps you make an educated decision for the next step of your research project and offers you the advantage of reanalyzing and reinterpreting your data at any time.

By using unique dual-indexed sequencing adaptors, we are able to analyze hundreds of samples in parallel delivering the least bias and highest reproducibility, making WGS a very powerful and cost-effective tool for genetic profiling.

Input material

Isolated genomic DNA
- Validated input:
  - without PCR ≥ 25 ng / sample*
  - with PCR ≥ 10 ng / sample
- Minimum volume: 20 µl / sample
- Quality: High Molecular Weight DNA

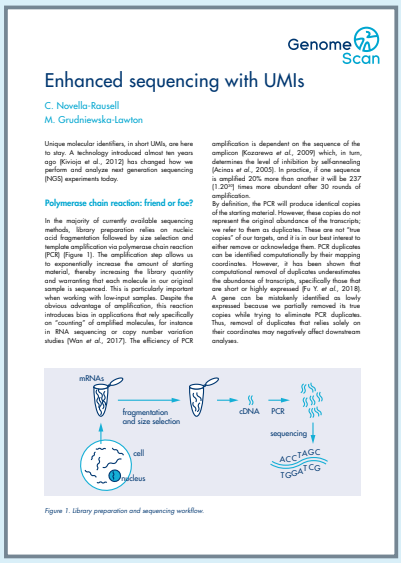
Sequencing on NovaSeq 6000 (PE 150)
- PCR free library preparation (≥ 25 ng)
- Unique Molecular Identifiers for samples with PCR
- Unique dual indexed sequencing adaptors
- Optional: FFPE repair available

Deliverables
- FastQ files
- Quality score Q30 of ≥ 80%
- Optional data analysis with comprehensive report

*Higher inputs required for deep sequencing

NGS Laboratory workflow

Sample delivery
Sample Entry QC
Library Preparation Sequencing Run Library QC
Data QC
Sample Report


Genome Scan

Enhanced sequencing with UMIs

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Unique molecular identifiers, in short UMIs, are here to stay. A technology introduced almost ten years ago (Kivioja 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 warranting 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 (Wan et al., 2017). The efficiency of PCR amplification is dependent on the sequence of the amplicon (Kozarewa et al., 2009) which, in turn, determines the level of inhibition by self-annealing (Acinas et al., 2005). In practice, if one sequence is amplified 20% more than another it will be 237 (1.20^{30}) 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 acknowledge 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 lowly 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 themost accurate services, look no further. Contact us today to learn how we can help you.

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