GenomeScan® OncoPair

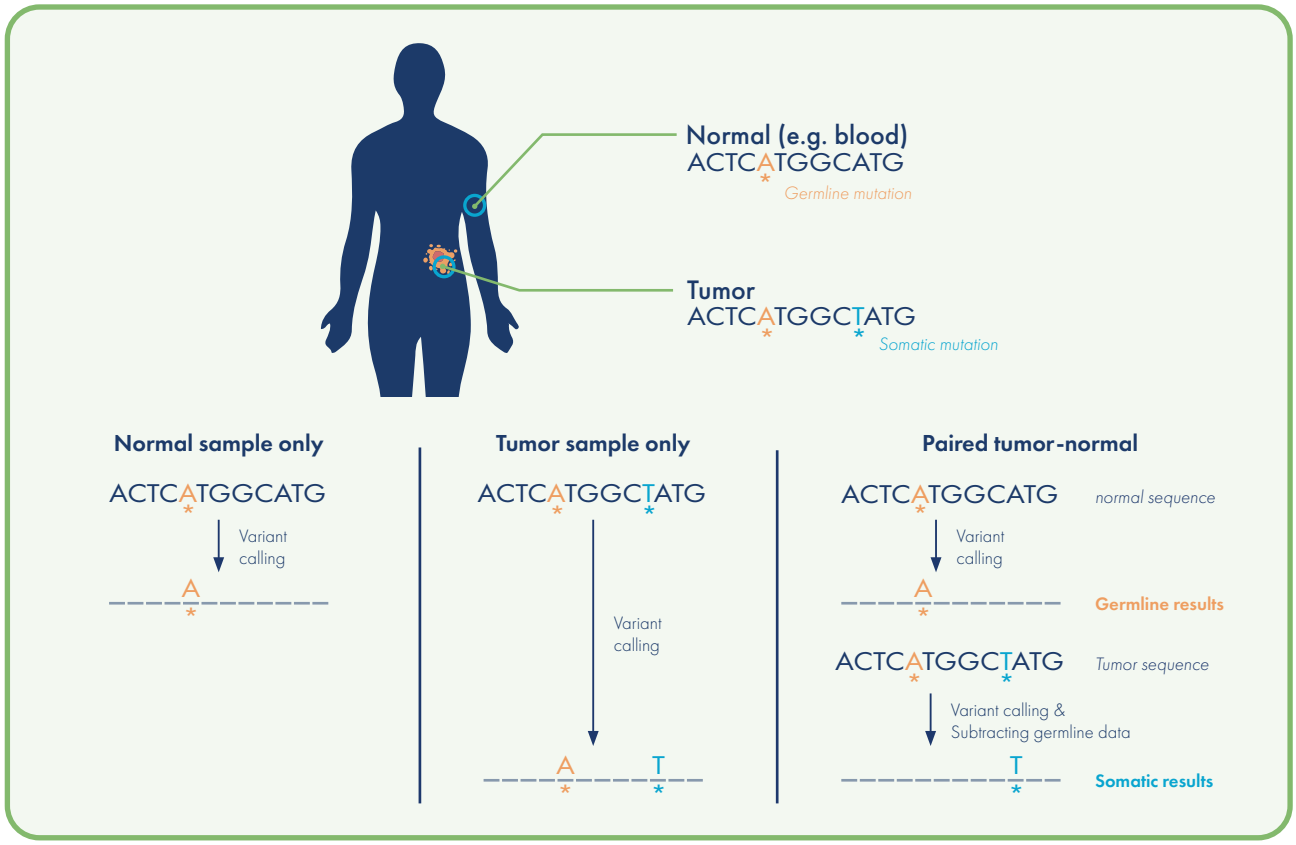
Matched tumor-normal sequencing for high-resolution cancer genomics

Cancer is a complex and heterogeneous disease, driven by a multitude of genetic alterations. Despite significant advances in diagnostics and therapeutics, a critical unmet need remains: the ability to accurately distinguish between inherited (germline) and acquired (somatic) mutations to guide personalized treatment strategies. Precision oncology demands tools that deliver reliable, high-resolution insights into the genomic landscape of each patient's tumor.

GenomeScan® OncoPair meets this need through a robust, ISO-accredited matched tumor-normal sequencing workflow.

By analyzing both tumor tissue and a matched normal sample, such as blood, saliva, or healthy tissue, OncoPair provides a comprehensive view of somatic mutations while filtering out germline background. This dual-sample strategy enhances the accuracy of variant interpretation, supports the identification of cancer-driving mutations, and facilitates informed clinical decision-making. The result is high-resolution genomic insights that drive both clinical and translational impact.

Note: OncoPair is for research use only and not intended for clinical diagnostics.



Tumor-Normal pair sequencing for mutation classification (adapted from Mandelker, D, & Ceyhan-Birsoy, O. (2020))

Input material requirements

Sample types

- Tumor samples: FFPE or Fresh Frozen tumor tissue
- Matched normal samples: Blood, saliva or healthy tissue

DNA input requirements

High Molecular Weight (HMW) genomic DNA

- PCR-free workflow: ≥ 200 ng per sample
- With PCR amplification: ≥ 10 ng per sample
- Minimum volume: 25 µl per sample
- Quality: free from impurities and RNA

FFPE genomic DNA

- Minimum input: ≥ 10 ng per sample
- Recommended input: ≥100 ng per sample
- Quality: DV200 ≥ 50% (HMW DNA not expected due to fragmentation)
- Free from impurities and RNA
- Minimum volume: 25 µl per sample

Sequencing Specifications

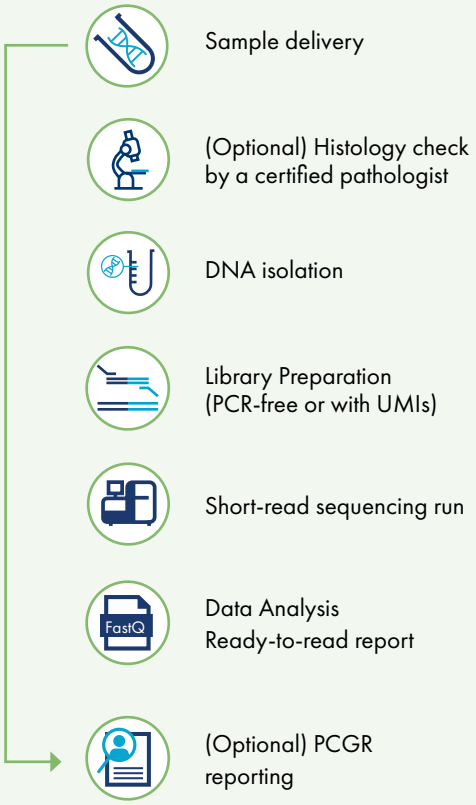
- Platform: Illumina NovaSeq (PE150)
- Total reads:
  - Tumor sample:
    - 90x coverage
    - ~1 billion paired end reads per sample
  - Matched normal sample:
    - 30x coverage
    - ~300 million paired end reads per sample
- PCR-free libraries preferred
- Unique Molecular Identifiers (UMIs) for PCR-based samples
- Unique dual-indexed sequencing adapters

Deliverables

- Data format: FastQ files, BAM files
- Quality metrics: ≥ 80% of bases with Q30 or higher*
- Variant types
  - Somatic SNVs, indels, CNVs, MNPs, SVs
- Standard reporting
- (Optional) Comprehensive analysis report (PCGR)

**Due to the degraded nature of FFPE samples, quality expectations are adjusted. We aim for at least 70% of bases to have a Q-score of 30 or higher.*

GenomeScan® OncoPair Workflow



Key features

Matched Tumor-Normal Design

Accurate somatic variant calling by filtering inherited variants; identifies tumor-specific mutations and drivers.

High-Depth Sequencing

90x tumor / 30x normal coverage enables detection of low-frequency variants, SVs, and clonal architecture.

Flexible Sample Types

Compatible with fresh frozen, FFPE, blood, or adjacent tissue; supports broad clinical and research use.

Comprehensive Variant Spectrum

Covers SNVs, indels, CNVs, SVs, offering a complete genomic profile in one package.

Neoantigen Insights

Enables identification of tumor-specific neoantigens for immunotherapy development and patient stratification.

Pathology Review

Optional histopathology ensures reliable tumor content estimation for confident variant interpretation.

Accredited Workflow

ISO/IEC 17025-accredited process ensures high-quality data for clinical trials and biomarker discovery.

Optional Reporting (PCGR)

Includes actionable insights, visualizations, and clinical annotations for therapy selection and stratification.

Precision Oncology Applications

These insights support personalized treatment strategies, such as:

- Designing targeted therapies based on unique somatic mutations.
- Developing immunotherapies by identifying neoantigens.
- Monitoring minimal residual disease and tumor recurrence.

Trustworthy results

Data Quality Guarantee

Depth of coverage, base quality and data quality are essential metrics to evaluate the quality of your NGS data.

Transparent Reporting

We deliver comprehensive, consistent, and transparent project reports that describe all experimental procedures and include relevant QC metrics.

ISO 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.

Committed to your project

GenomeScan® OncoPair is backed by GenomeScan's ISO/IEC 17025 accreditation, ensuring that all laboratory processes meet the highest standards of quality, technical competence, and reproducibility. Each project begins with a pilot phase to assess sample quality and optimize workflow parameters, reducing the risk of suboptimal data and enabling informed decision-making before scaling up.

Key quality guarantees

- High mapping rates across exonic, intronic, and intergenic regions.
- Reproducibility across a wide range of input amounts.
- Expert QC at every stage, from sample intake to data delivery.

About GenomeScan

GenomeScan is a trusted ISO-accredited partner for pharmaceutical companies seeking high-quality, customizable Next Generation Sequencing (NGS) solutions. With a strong foundation in clinical-grade genomics, we support drug discovery, biomarker development, and translational research through robust sequencing workflows and expert bioinformatics.

Our services are designed to accelerate therapeutic innovation by delivering precise, reproducible, and regulatory-compliant genomic data. Whether supporting early-stage research or clinical development, GenomeScan empowers pharma partners with the tools and insights needed to advance personalized medicine and targeted therapies.

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GUID026, Version 1, Authorized by QA