

Simplifying cancer analysis

Bridging science and personalized medicine by uncovering hereditary and somatic mutations

Complete Solution

Helping laboratories turn sequencing data into meaningful insights, our NGS assays provide a complete solution comprising **library preparation**, **bioinformatics analysis** with **GENOVESA**, and generation of **genetic variant reports**.

NGS is becoming the new gold standard for human genetic disease testing

Cancer is one of the leading causes of death worldwide. It is estimated that 19.3 million people are diagnosed with cancer every year, and approximately 10 million die from it.¹ Therefore, an early and efficient diagnosis is indispensable for increasing the chances of effective treatment and survival.

Next Generation Sequencing (NGS) has revolutionized many areas of science and healthcare

including cancer detection. Applications of NGS on disease diagnosis, prognosis, and therapeutic decision have widely expanded, offering new opportunities for personalized medicine.²

Advantages of NGS technology

NGS-based kits provide **qualitative** and **quantitative data** in one assay and interrogate different types of **genetic variations simultaneously** (SNV, InDel, CNV, Fusion).

The BioVendor Group NGS Assays leverage NGS technology to examine multiple genomic regions simultaneously and provide a solution tailored to customer needs.



Key features

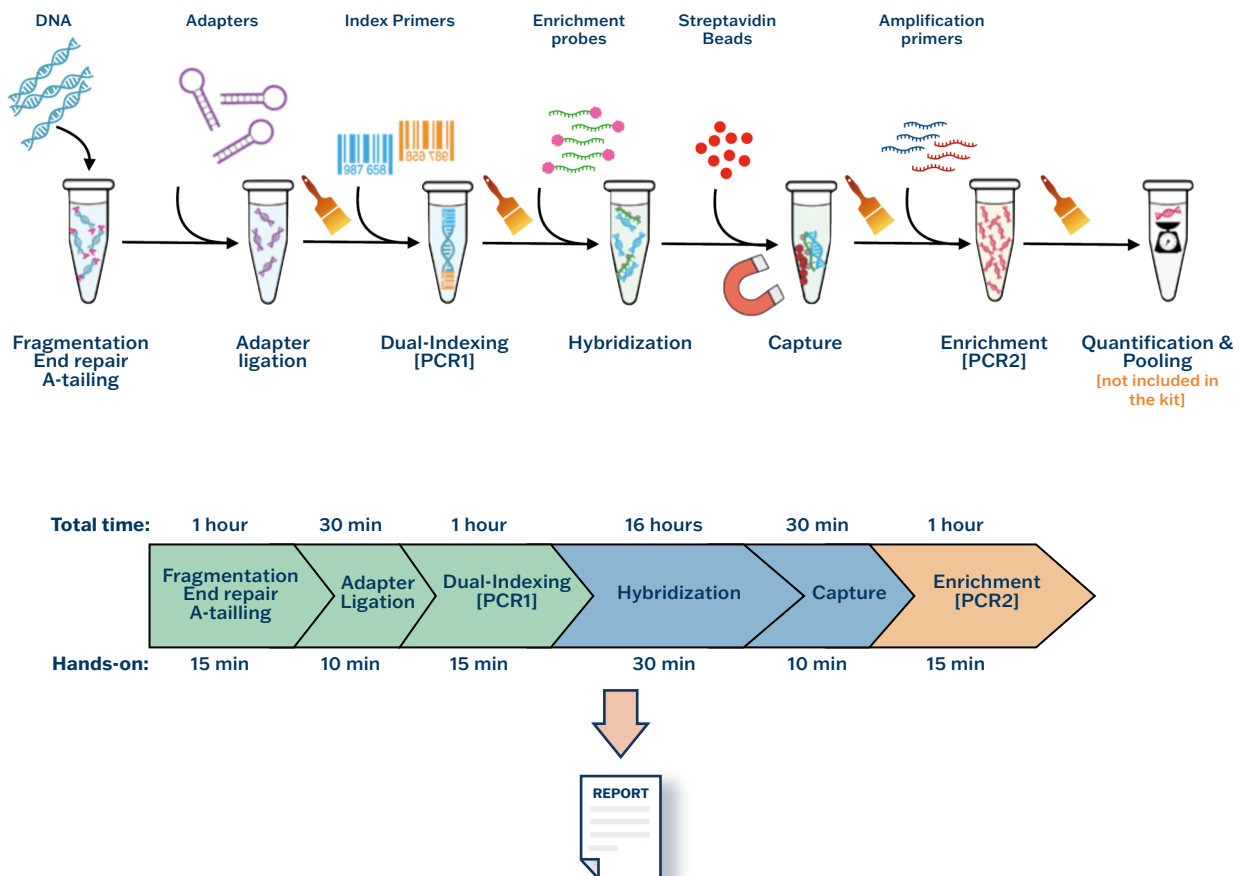
- Complete solution - from DNA to variant report generation
- Suitable for intact DNA and FFPE DNA*
- Robust and easy to follow protocol completed in 1.5 days
- Sensitive detection of low frequency variants
- Optimized workflow
- PanelCalc tool to support users with final library pooling and sample sheet generation
- User-friendly proprietary bioinformatics software (GENOVESA)
- Pre-set/custom filtering options
- Genetic variant report generation
- Compatible with Illumina sequencing platforms

* FFPE DNA is best suited for Somatic Mutations NGS Assay. It can be used for Hereditary Cancer NGS Assay.

Workflows of the NGS Assays

Using a hybridization capture-based approach covering the **whole coding sequence** of genes of interest, these assays deliver uniform coverage

of targeted regions while enabling the detection of co-occurring mutations and complex genomic rearrangements across multiple genes.



References:

¹ Ferlay J et al. Cancer statistics for the year 2020: An overview. Int J Cancer 2021

² Yadav D et al. Next-Generation sequencing transforming clinical practice and precision medicine. Clin Chim Acta. 2023

GENOVESA

Genovesa - a web-based platform for automated bioinformatic analysis of NGS data

To accelerate variant detection a proprietary, easy to use web-based software called GENOVESA is used. It allows powerful data analysis of FASTQ files with only a few clicks. GENOVESA supplies the user with an automated pipeline for the detection of SNVs, InDels and in case of the Hereditary Cancer NGS Assay also CNVs. Subsequently, it annotates the variants with up-to-date information from renowned databases. GENOVESA is an **essential part** of the NGS Assays.

GENOVESA allows visual inspection of the alteration in a user-friendly way directly in the same interface. It provides information about **MSI status** indispensable for investigation of **Lynch Syndrome** and immunotherapy decisions*. It uses qualitative and quantitative information from each sequenced DNA molecule **(A)**.

It offers carefully selected pre-set filters (Default filter) to ease the variant discovery. The Default filter parameters were specifically designed for optimized

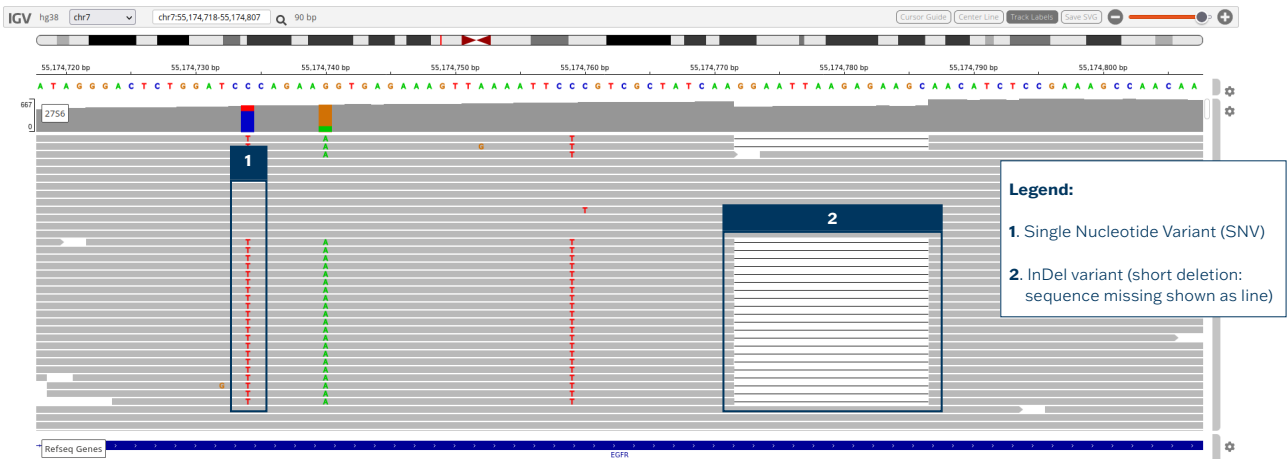
* MSI detection possible for Hereditary Cancer NGS Assay.

variant identification for each of our assays. In the example below, our Default Filter reduces the variant list from 171 total variants down to 2 relevant variants **(B)**.

For more experienced users, custom filters are available, with the possibility to modify parameters accordingly. Furthermore, GENOVESA offers the possibility to use pseudopanels - custom subsets of genes important for specific phenotypes or diseases - in order to narrow down the analysis according to your needs.

GENOVESA allows for automated processing and quality control metrics of sequencing data. Subsequently, a meaningful report is generated which shows the upfront selected variants. GENOVESA has been successfully used in-house to accurately detect, annotate, filter, and select clinically relevant SNVs and InDels using the correct up-to-date HGVS/p nomenclature in recent EQA evaluations.

A



B

ID	SampleID	Status	Total reads	Total Variants
5945	33H7	SIGNED OUT	1 429 132	171

Sample	CL_A	CL_B	AOMG	ClinVar	Predict	Freq / Proj	Freq / Germ	Gene	dbSNP	HGVS / HGVS p	VAF	Total DP	Ref. : Alt DP	Type	Consequence	Var Qual
33H7	5	0	5	5	NA	2	2	BRCA1 NM_007294.4	rs80359047 Links	c.547>2T>A	51.67	389	188 : 201	SNV	splice donor varian t	good
33H7	5	0	5	5	NA	2	2	ATM NM_000051.4	rs375783941 Links	c.6040G>T p.Glu2014Ter	52.48	463	220 : 243	SNV	stop gained	good
Total 2																



NGS Assays at a glance

Early detection of cancer constitutes a key point for successful treatment. Preventive screening to detect and analyse a potential harmful variant in families at risk is crucial for early diagnosis. To help with this arduous task, the **Hereditary Cancer NGS Assay** interrogates the key genes associated with inherited cancer variants.

Tumor molecular profiling helps to establish the most accurate therapy. It allows physicians to make the best decision in regard to targeted therapy depending

on the individual mutations detected in each patient. The **Somatic Mutations NGS Assay** is a panel carefully designed for that purpose.

Whether your focus is hereditary cancer screening or somatic mutation profiling, our assays combine an optimized workflow with streamlined downstream data analysis to provide an all-in-one solution. A panel content carefully selected to address a broad range of testing needs.

 Hereditary Cancer NGS Assay [REF 9-221] (16rxn)	RUO
Covered Genes	APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PRSS1, PTEN, RAD50, RAD51C, RAD51D, SLX4, SMAD4, STK11, TP53, VHL
Variants	SNV, InDel, CNV, MSI
Target Region	Whole CDS (Target size: 97Kb)
Preferred Sequencer	Illumina MiSeq, MiSeq i100 series (up to 16 samples), MiniSeq, iSeq 100
Bioinformatic Analysis (Genovesa) included	✓
 Somatic Mutations NGS Assay [REF 9-231] (16rxn)	RUO
Covered Genes	ALK*, APC, BRAF, EGFR, ERBB2, KRAS, MET, NRAS, PIK3CA, RET*, ROS1*, SMAD4, TP53
Variants	SNV, InDel, Fusions*#
Target Region	Whole CDS and hotspot introns for fusions* (Target size: 62Kb)
Preferred Sequencer	Illumina MiSeq, MiSeq i100 series (up to 16 samples), MiniSeq, iSeq 100
Bioinformatic Analysis (Genovesa) included	✓

MANUFACTURER:

DISTRIBUTOR:



ViennaLab Diagnostics GmbH

Gaudenzdorfer Guertel 43-45
A-1120 Vienna, Austria
+43 1 812 0156
info@viennalab.com
www.viennalab.com

June 2026