

Focused or Comprehensive ?

ViennaLab NGS Assays - Bridging science and personalized medicine. ViennaLab NGS Assays uncover hereditary and somatic mutations

Complete Solution

ViennaLab NGS assays enable focused analysis of germline and somatic variants in selected cancer-associated genes as well as comprehensive Clinical Exome analysis. ViennaLab offers a complete solution comprising **library preparation, bioinformatic analysis** with our software **GENOVESA** and generation of a **genetic variant report**.

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 ViennaLab NGS Assays **take advantage of the NGS technology** to examine many genomic regions at the same time, and offer a solution for customer's needs.

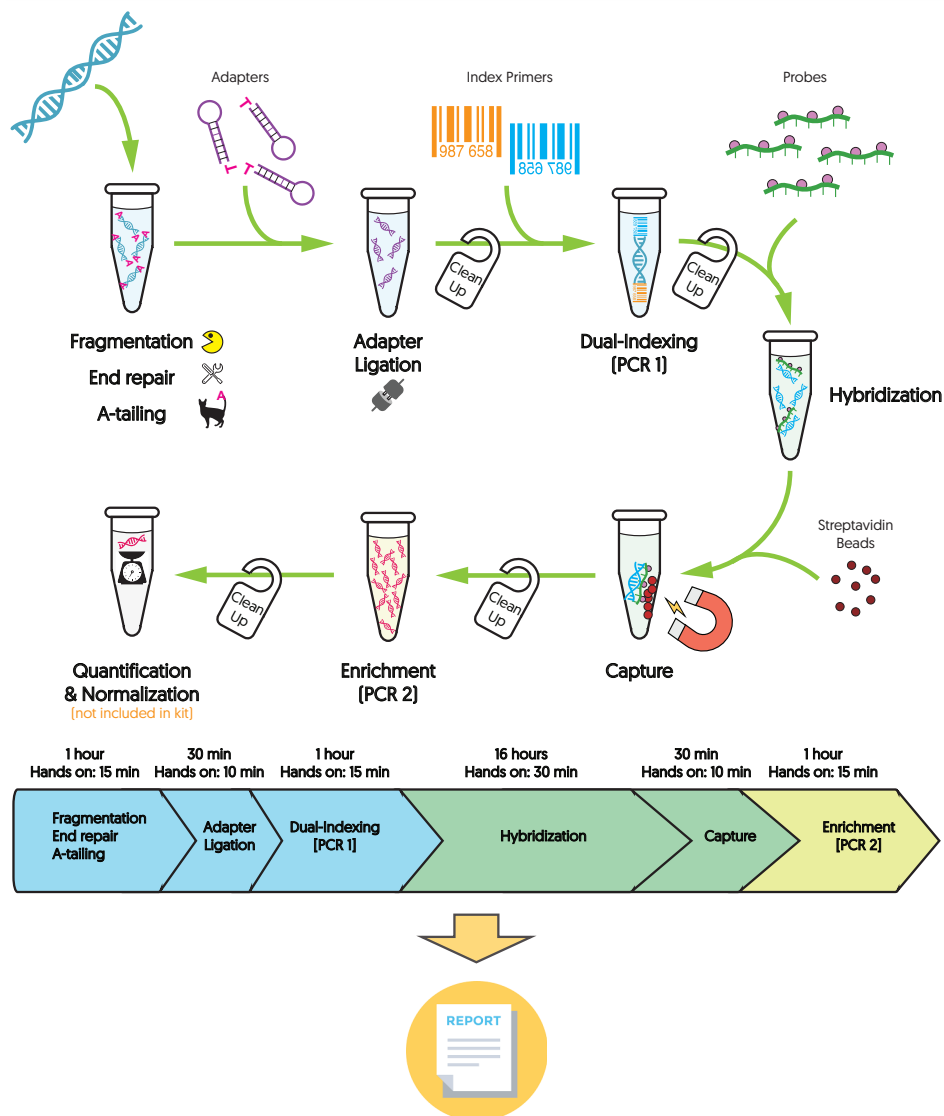
Key features

- Complete solution - from DNA to variant report generation
- Suitable for intact DNA and FFPE DNA
- Robust and easy to follow protocol done 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 bioinformatic software (GENOVESA)
- Pre-set/custom filtering options
- Genetic variant report generation
- Compatible with Illumina sequencing platforms

Workflow of the ViennaLab NGS Assay

The ViennaLab NGS protocol relies on a hybridization capture-based approach covering the **whole coding sequence** of genes of interest. It provides uniform coverage of the investigated regions and is flexible

to detect co-occurring mutations or more complex genomic rearrangements. Furthermore, hybridization allows the simultaneous interrogation of many genes with a single assay.



References:

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

² Gonzales-Garay, M. L. The road from next-generation sequencing to personalized medicine. 2014. Per Med. 11:523-544

GENOVESA

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

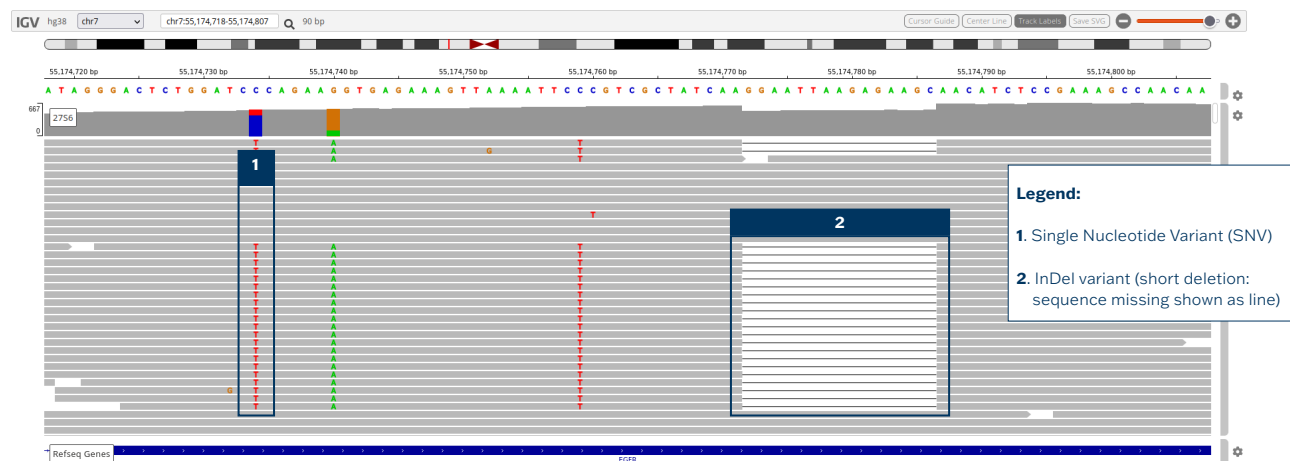
To accelerate variant detection a proprietary, easy to use web-based software called GENOVESA is used. It allows powerful data analysis of FASTQ files with only a few clicks. GENOVESA supplies the user with an automated pipeline for the detection of SNVs and InDels as well as variant annotation with up-to-date information from renowned databases. GENOVESA is an **essential part** of the ViennaLab NGS Assays.

GENOVESA can accurately detect and annotate relevant variants and allows visual inspection of the alteration in a user-friendly way directly in the same interface. It uses qualitative and quantitative information from each sequenced DNA molecule **(A)**. It offers carefully selected ViennaLab pre-set filters to ease the variant discovery. In this example, the ViennaLab 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 pseudopanel - 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

ViennaLab Filter	All Variants	Custom Filter	Diagnoses	Preselected Variants	Signed Out Variants
<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Transcript system: Ensembl RefSeq

Sample ID	Int. Cl. A	Int. Cl. B	ACMG	ClinVar	Freq / Proj	Freq / Ge rm	Gene / Transcript	Chr:Position	dbSNP	HGVS / HGVS p	Type	Consequence	Var Quality
33H7	5	0	5	5	2 / 8	2 / 294	BRCA1 NM_007294.4	chr17:43099773	rs80358047	c.547+2T>A	SNV	splice donor variant	good
33H7	5	0	5	5	2 / 8	2 / 294	ATM NM_000051.4	chr11:108315856	rs375783941	c.6040G>T p.Glu2014Ter	SNV	stop gained	good

Total 2

Pre-set ViennaLab Filter optimizes the identification of clinically relevant variants

ViennaLab 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, ViennaLab offers the **Hereditary Cancer NGS Assay** to interrogate 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.

ViennaLab offers the most comprehensive panel - the **Clinical Exome Sequencing (CES) NGS Assay** - to even identify the genetic basis of complex and challenging phenotypes. It contains all clinically relevant genes and hot spot regions which can be screened simultaneously in a cost-efficient way.

Depending on **your needs**, we offer a **focused or comprehensive** approach. With our optimized and efficient protocol and downstream data analysis, we provide an all-in-one solution. The content of each panel was carefully selected to address the needs of many users.

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 [#]	
Target Region	Whole CDS (Target size: 97Kb)	
Preferred Sequencer	Illumina MiSeq (up to 16 samples)	
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 (up to 16 samples)	
Bioinformatic Analysis (Genovesa) included	✓	
CES NGS Assay [REF 9-241] (4x4 rxn)		RUO
Covered Genes	7500+ genes that are known contributors of disease pathogenesis	
Variants	SNV, InDel	
Target Region	Whole CDS, hotspots, mitochondrial genome (Target size: 19,7 Mb)	
Preferred Sequencer	Illumina NextSeq, NovaSeq [§] (at least 16 samples)	
Bioinformatic Analysis (Genovesa) included	✓	

[#] Analysis available upon request; [§] Most economical option

MANUFACTURER:

DISTRIBUTOR:



ViennaLab Diagnostics GmbH

Gaudenzdorfer Guertel 43-45

A-1120 Vienna, Austria

+43 1 812 0156

info@viennalab.com

www.viennalab.com

January 2023