

BViennaLab[®]

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

Group GENOVESA		PRO V	LIGHT	✓ FASTGE	ViennaLab Diagnostics GmbH ≜▼									
Projects														
5	~				ADD PROJECT	0		Search O ₄						
ID \$	Project Name 🌲	Workflow 🔶	SI	tatus	Created	Finished	Description 🚔	Actions						
289	Run 5	Vienna Lab He	ereditary Cancer	ASSED	22. 10. 2021	09. 12. 2021								
281	Run 4	Vienna Lab He	ereditary Cancer	ASSED	18. 10. 2021	18. 10. 2021								
279	Run 3	Vienna Lab So	omatic Mutations	ASSED	15. 10. 2021	15. 10. 2021								
278	Run 2	Vienna Lab He	ereditary Cancer	ASSED	15. 10. 2021	19. 10. 2021								
277	Run 1	Vienna Lab So	omatic Mutations	ASSED	12. 10. 2021	12. 10. 2021								
Showing 11 To	o 15 Of 53 Entries							« 1 2 3 4 5 6 7 »						

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ViennaLab filter – preset filters optimized for simplified and time-saving identification of clinically relevant variants

Examples for each workflow:

- ViennaLab Hereditary Cancer: from **170** variants down to **2** relevant variants
- ViennaLab Somatic Mutations: from 107 variants down to 8 relevant variants
- ViennaLab CES: from **12854** variants down to **307** relevant variants

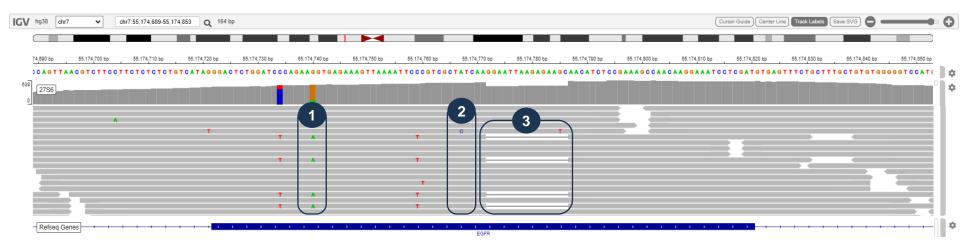
	SampleID ≑	Status ≑	Total reads ≑	Total Variants										
	33H1	PASSED	1 485 196	170										
Vernat.ab-Filter All Variants Custom Filter Diagnoses Preselected Variants Signed Out Variants														
Transcript system: Filter by: Filter by:				Q X 167										
Sample ID Int. CI. A Int. CI. B C	inVar 🗢 🛛 Freq / Proj	Freq / Ger m Gene / Transcript 💠	Chr:Position \Leftrightarrow dbSNP \Leftrightarrow	HGVSc / HGVSp 💠	Type 💠 Consequence 🗢	Var Quality	V/ Actions							
> 33H1 5 0	5 2/8	2 / 3652 BRCA1 NM_007294.4	chr17:43099773 rs80358047	<u>c.547+2T>A</u>	SNV splice donor variant	good								
> 33H1 5 0	5 2/8	2 / 3652 ATM NM_000051.4	chr11:108315856 rs375783941	c.6040G>T p.Glu2014Ter	SNV stop gained	good								



2

3

IGV visualization of SNV and InDel variants



- Single Nucleotide Variant (SNV) shown as colored letter
- SNV variant observed only once/few times, likely sequencing artifact
- InDel variant (short deletion: sequence missing shown as line)
- → NGS allows qualitative and quantitative information of each sequenced DNA molecule

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Key features of the new ViennaLab NGS assays

- ✓ Suitable for FFPE and intact DNA
- ✓ Compatible with Illumina sequencing platforms
- ✓ Full package: library preparation, bioinformatic analysis, genetic variant report generation
 - ✓ Web-based data analysis pipeline: GENOVESA
 - ✓ Automatic QC and data processing
 - ✓ **Preset filters** optimized to identify clinically relevant variants
 - Custom filters allow comprehensive investigation of data
 - ✓ Visual display of variants for both SNVs, InDels one-click integrated IGV
 - ✓ CNV and SVs analysis available upon request
 - ✓ Meaningful report