

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

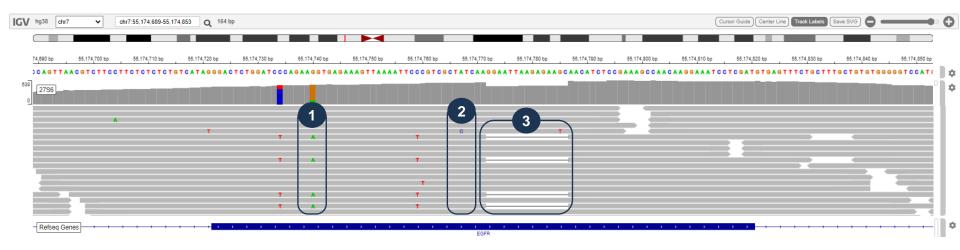
| | SampleID ≑ | Status ≑ | Total reads ≑ | Total Variants | | | | | | | | | | |
|--|-----------------------|-------------------------------------|--|---------------------------|--------------------------|-------------|------------|--|--|--|--|--|--|--|
| | 33H1 | PASSED | 1 485 196 | 170 | | | | | | | | | | |
| 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