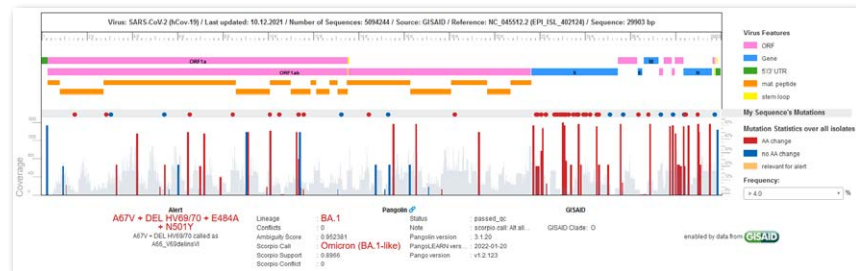
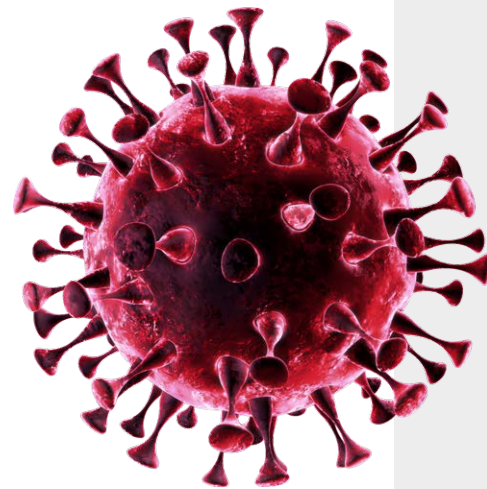


Contact

Are you also sequencing
SARS-CoV-2?

Check out
www.virSEAK.bio



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SEQUENCE Pilot

Software For Genetic Analysis

NEW!
VERSION 5.3

SEQNEXT
Next Generation Sequencing

varSEAK
Variant Interpretation

SEQPATIENT
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SEQARRAY
Genotyping-Microarray
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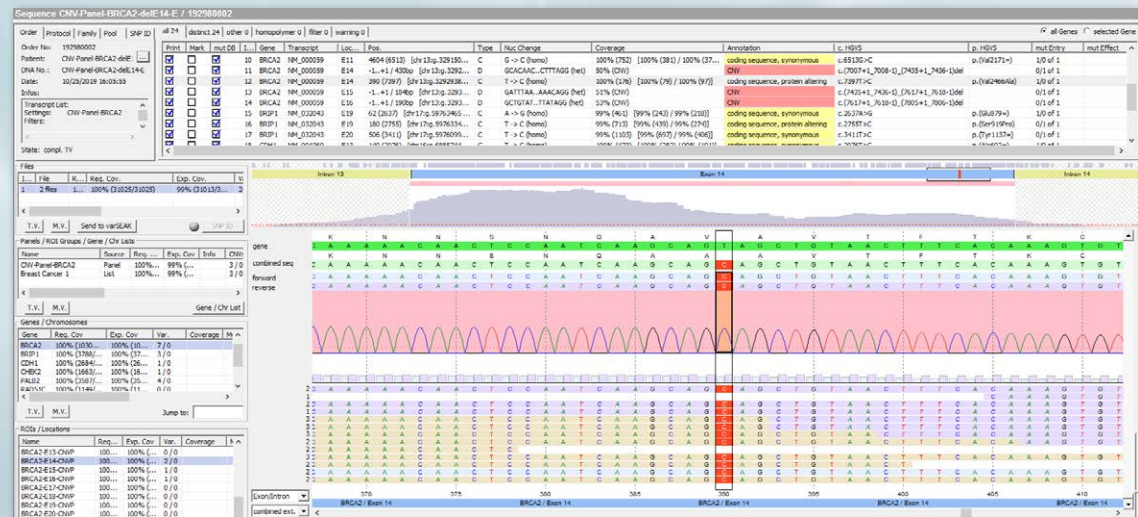
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A customer oriented company with customised solutions for your needs

JSI medical systems GmbH is a company with over 20 years of experience in software development for DNA sequencing analysis and interpretation. Our philosophy is to place customer satisfaction at the core of each of our business decisions. Thus competent support, availability, short response times, intensive on-site or online trainings and the implementation of customer needs are only some of our key strengths.

A fully automatable, all-in-one solution for the analysis of sequencing data



Detailed analysis view of SEQNEXT

SEQUENCE Pilot – a fully automatable, cost and time efficient all-in-one solution

Discover the SEQUENCE Pilot software modules:

SEQNEXT | Next Generation Sequencing

Convenient and powerful analysis tool for next generation sequencing data including WES ▪ Supporting all common platforms and kit manufacturers ▪ Customizable settings for SNP, Deletion, Insertion, CNV, fusion gene detection ▪ Supporting SNP IDs, processing of smMIPs and UMIs, etc.

SEQPATIENT | Sanger Sequencing

Easy and fast analysis of Sanger sequencing data ▪ Configurable base caller with sequence-dependent thresholds ▪ Self-adapting algorithm based on peak area statistic for a sensitive detection of heterozygous positions (mosaics, somatic mutations, etc.) ▪ Sophisticated tools for frame shift and indel detection

SEQARRAY | Genotyping-Microarray (Illumina Infinium®)

Intuitive analysis tool for genotyping arrays (Infinium® Global Screening Array) ▪ Quick and user friendly import of GenomeStudio 2.0 data ▪ Convenient filters for genes and chromosomes ▪ Customizable variant table listing SNPs, insertions and deletions, plus CNV visualisation based on illumina cnvPartition algorithm (version 3.1.4)

MLPA® | Multiplex Ligation-dependent Probe Amplification

Powerful tool for detecting copy number changes ▪ Pre-configured SALSA MLPA® kit descriptions available for download and import ▪ Configurable analysis modes, control settings and RPA limits ▪ Data correction for probes with "tailing-off-effect"

varSEAK | Variant Interpretation

Fast and reliable tool to manage and interpret sequencing data ▪ Single click import of SEQNEXT, SEQPATIENT, SEQARRAY and VCF data ▪ Potent and highly flexible filter functions ▪ Automated calculation of Splice Site Prediction results ▪ ACMG-compliant classifications

virSEAK | SARS-CoV-2 NGS

User friendly analysis of SARS-CoV-2 next generation sequencing data ▪ Predefined settings including quality requirements (e.g. for RKI in Germany) available ▪ Pangolin integrated for lineage assignment ▪ Sequence exports in submission-ready fasta format

SEQNEXT-HLA | HLA Next Generation Sequencing

Convenient tool for the analysis of HLA typing based on next generation sequencing data ▪ All IMGT HLA database versions and updates available ▪ Display of results in different resolutions ▪ Calling of null alleles as well as new alleles

SEQHLA | HLA Sanger Sequencing

Comfortable tool for the analysis of HLA typing based on Sanger data ▪ All IMGT HLA database versions and updates available ▪ Display of results in different resolutions ▪ Splice site mutation warning

Comfortable filter function of varSEAK

Step	Operation	Description	„Remaining“ count after filter step	Count change	„Filtered“ count after filter step	Actions
	(Start empty)	Let „Remaining“ be the empty set and „Filtered“ be the set of all order variants				
1.	ExampleFilter	This is just an example description for the example filter.	0		15567	
1.1.	Include	Move those variants from „Filtered“ to „Remaining“ which fulfill these conditions: Variant Gene is in HPO abnormalities (Retinal degeneration (HP:0000546), Retinal dystrophy (HP:0000556), Retinopathy (HP:0000488)) (Sum)		+++ +1809 +++		
		Variant Gene is in HPO abnormalities Retinal degeneration (HP:0000546) Retinal dystrophy (HP:0000556) Retinopathy (HP:0000488)	1809		13758	
1.2.	Exclude	Move those variants from „Remaining“ to „Filtered“ which fulfill these conditions: Public DBs + ClinVar + Variant Clinical Significance isn't intersecting with (Likely pathogenic, Pathogenic, Pathogenic/Likely pathogenic)		+++ -1801 +++		
		Public DBs + ClinVar + Variant Clinical Significance isn't intersecting with Likely pathogenic Pathogenic Pathogenic/Likely pathogenic	8		15559	

