ECSEQ BIOINFORMATICS

DNA Variant Analysis

Study Genomic Variation With Ease.

KEY BENEFITS

Rapid Results
Our skilled team of professional bioinformaticians delivers results rapidly.

Expert Solutions
Rely on in-depth bioinformatics experience and up-to-date scientific methods.[1]

Excellent Support
Experts always on hand to address your bioinformatics concerns.

Flexible Analyses
Employ an analysis strategy that is adjusted to your data.

Trustworthy Results
Our DNA-Seq analyses have been tested and validated in many projects.

High Data Security
Securely transfer and access your data and the results.

Explore & Share your Results
Comprehensive interactive HTML reports included.

Best-in-class Methods
Get the most biologically relevant information out of your data.

[1]Discover our DNA-seq expertise at ecseq.com/analysis/DNA-Seq

The study of genetic variants has surged forward with the advent of current methods of next-generation sequencing (NGS). Steady improvements in sensitivity and decreasing costs enable widespread use of this innovative technology in applications like cancer diagnostics, biomarker discovery, precision medicine, and many more. The most prominent assays are whole genome, whole exome, and targeted amplicon sequencing.
DNA re-sequencing using NGS is a well-established method and our researchers have proven expertise in its analysis. Our proceedings are approved by renowned experts in the field and our analyses have been published in high-impact, peer-reviewed journals (find details under ecseq.com/analysis/DNA-Seq)

The Importance of Read Mapping

The alignment of DNA reads crucially affects how well SNPs and structural variations can be detected. Our mapping approach has been optimised for DNA sequences from NGS and provides high sensitivities with low false positive rates (see Table). Importantly, the error tolerance of the algorithm allows exploration of regions with clusters of sequence variants while simultaneously coping with sequencing errors. Conversely, our method automatically clips error-rich alignment ends and hence excludes remainders of sequencing adapters or low-quality read tails to avoid biasing subsequent variant calling.

Variant Calling and Filtering

Of major importance is the level of trust you can place in the variants obtained from your DNA-Seq analysis. Factors such as the selected assay and type of sequencing data affect variant calling. We therefore use a robust variant calling algorithm that resolves the challenges introduced by insertions and deletions as well as base quality inaccuracies. Depending on the type of sequencing data, it may be necessary to first detect and remove PCR clones to get accurate and reliable estimates of the variant frequency. In the case of targeted amplicon sequencing data, it is also essential to completely trim target primers from the alignments before variant calling. Otherwise, reference-matching primers of amplicons may dilute variant frequencies.

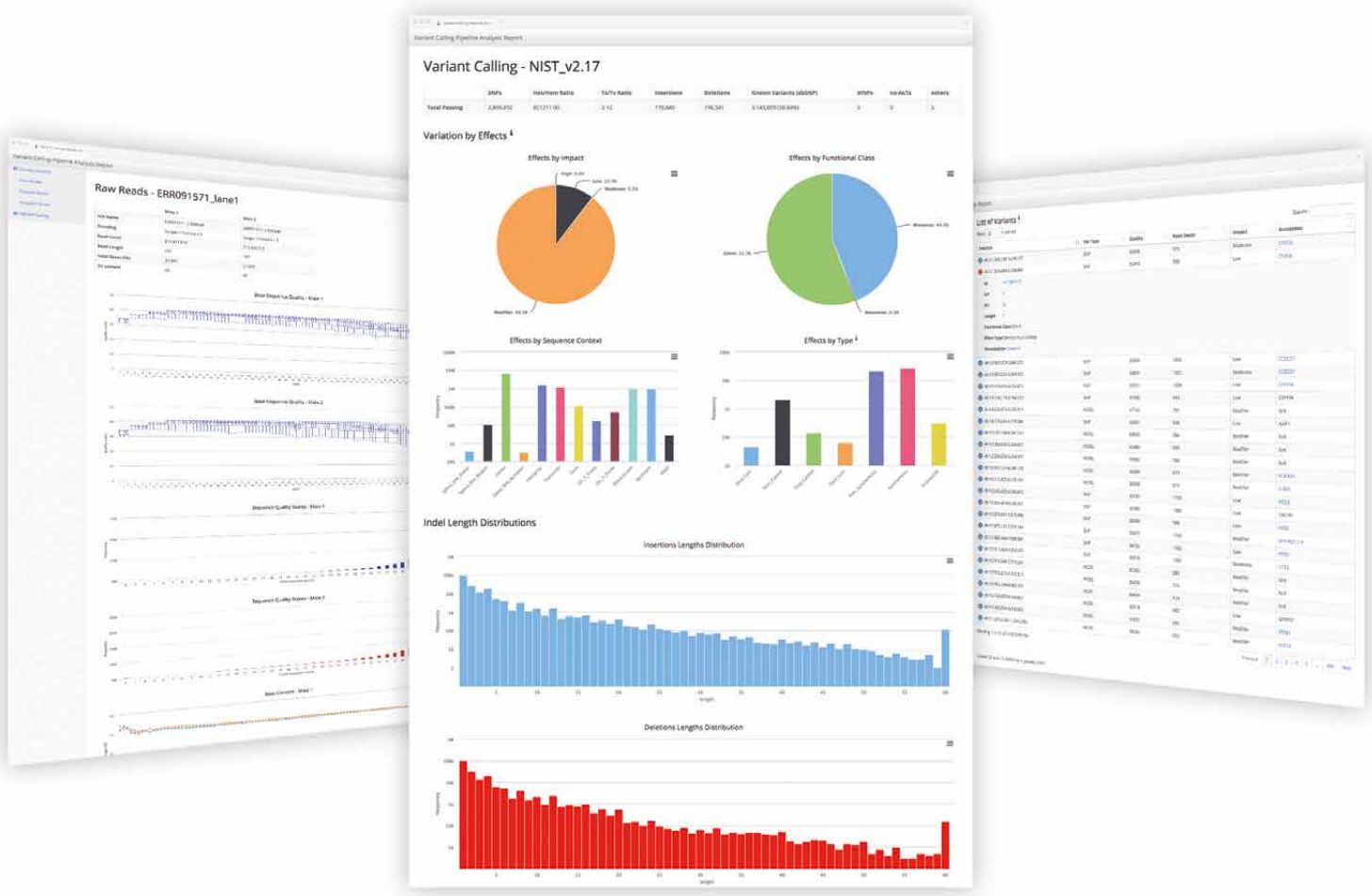
DNA-Seq

	Single-End	Paired-End
Sensitivity	96.39%	99.29%
False Positives	0.03%	0.42%

Annotation and Data Integration

Stringent and biologically relevant filtering of the discovered sequence variants is essential for DNA variant analyses, as well as the integration of additional data sources. After all, you want to be sure you are investing your time and resources into trustworthy and relevant variants. Besides the integration of common variation databases (e.g. dbSNP, ClinVar, etc.) to identify whether a variant is known, the annotation of variants with respect to their genomic context is important since this allows the differentiation of true but silent variants from ones that may impact the function of the associated protein. In this way, this variant annotation process enables classification of the potential severity of novel variants using functional prediction algorithms. In addition, the sequence variation data can be integrated, for example, with sequence data from RNA sequencing to allow selection of candidates affecting expressed transcripts, thereby reducing the list of variants to a set of the most interesting ones.

Interactive HTML Reports



Basic Analyses

- Sequencing Data Quality Control
- Adapter Clipping
- Bad-End Quality Trimming
- High-Sensitivity Read Mapping
- Enrichment Analysis
- Removal of PCR Clones
- Robust Variant Calling
- Post-Filtering Options
- Annotation of Variants and Prediction of their Effects

Additional Analyses

- GO-Term Analyses
- Somatic Mutation Analyses in Cancer
- Trio/Cohort/Pedigree Analyses
- Data Integration with RNA-Seq
- Custom Analyses

End-to-end NGS Solutions

For DNA-Seq applications, we also offer end-to-end analysis, including

- Sample Preparation
- Next-Generation Sequencing
- Bioinformatics Data Analysis

Contact one of our scientists and discuss your project!

Use the callback service on our website **www.ecseq.com** or write to support@ecseq.com

About Us

ecSeq GmbH is a bioinformatics solution provider focusing on next-generation sequencing technologies. Since 2012, ecSeq GmbH has been providing data analysis services and bioinformatics training for various NGS applications.

ecSeq GmbH
Brandvorwerkstrasse 43 · 04275 Leipzig · Germany
linkedin.com/company/ecseq · twitter.com/ecSeq
support@ecseq.com · www.ecseq.com