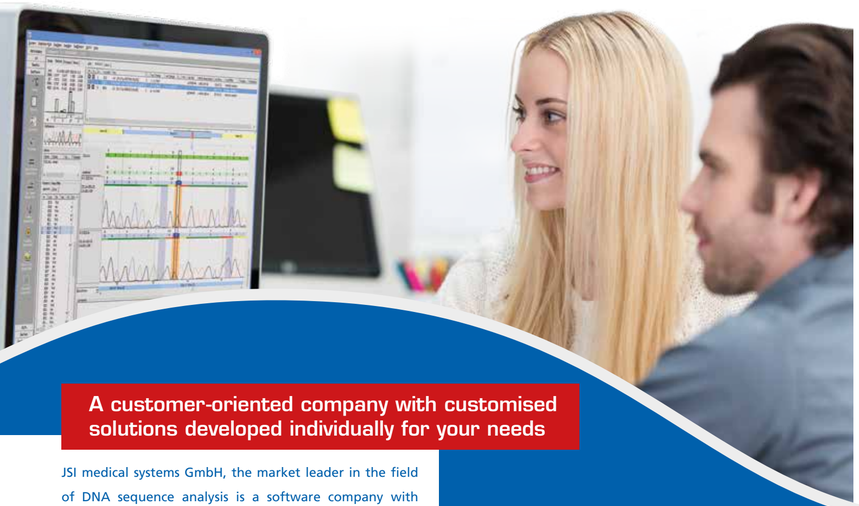
SEQUENCE PILOT

General product features

- Powerful all-in-one solution for robust high-throughput variant detection and interpretation (mapping, alignment, variant calling, QC-filtering, visualisation, filtering and data interpretation)
- Compatible with all common sequencing platforms
- User-friendly graphical interface with all necessary information and results in one screen
- Easy navigation in the sequence, moving to points of interest by a single click
- Patient-based work flow: automatic joining of result files to patient-related orders
- Collection of patient data independent of input format and **SEQUENCE PILOT** module in one order
- Technical and medical validation, secured by the user rights management
- Detailed and customisable analysis reports
- Available as single and multiple user application
- Runs on Linux and Windows server systems (32 and 64 bit)
- Efficient use of all available computing resources by parallel data analysis on multiple cores and computers
- Automated data import (ListenMaster) and export (TalkMaster) to LIMS systems



A customer-oriented company with customised solutions developed individually for your needs

JSI medical systems GmbH, the market leader in the field of DNA sequence analysis is a software company with decades of experience in developing software in the field of clinical diagnostics. The company philosophy is to place customer satisfaction at the core of each of its business decisions. Thus competent support, availability, short response times, intensive on-site or online trainings and the implementation of customer demands are only some of JSI's key strengths.

Discover the convenient and powerful SEQUENCE PILOT software modules!!!

Convince yourself of the benefit of the **SEQUENCE PILOT** modules by analysing your own data or demo data provided on our homepage.
Free trial licenses + software introduction (WebEx session) are available upon simple request.

www.jsi-medisys.com

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JSI medical systems

SEQUENCE PILOT

Software For Genetic Analysis

NEW!
SEQPILOT VERSION 5

SEQPATIENT
Sanger Sequencing

SEQNEXT
Next Generation Sequencing

SEQHLA
SBT Sanger Sequencing

SEQNEXT-HLA
SBT Next Generation Sequencing

MLPA®
Multiplex Ligation-dependent Probe Amplification

JSI medical systems

A fully automatable, cost and time efficient all-in-one solution

The utilisation of the **SEQUENCE PILOT** software as stand-alone tool or as a seamlessly integrated part of existing analysis pipelines, allows you to address current and future challenges related to the understanding, integration and use of biomedical and clinical diagnostic data. These challenges include the need to streamline the clinical testing process, manage the vast amounts of data generated in genetic testing, generate clinically useful interpretations from these data and channel this information efficiently and effectively to clinicians to impact patient care.

LIMS → Data Import → **ListenMaster** (JSI Interface) → **SEQUENCE PILOT** → **TalkMaster** (JSI Interface) → Data Export → LIMS

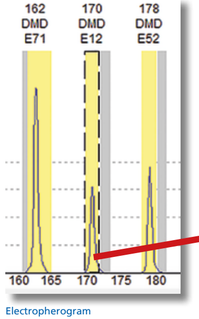
Peak area statistic

Electropherogram

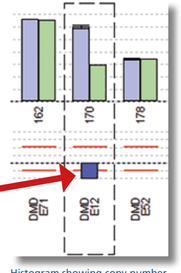
SEQPATIENT

The **SEQPATIENT** module facilitates the convenient and powerful analysis of Sanger sequencing data.

- Peak area statistic based on previously analysed data (internal data base) for a sensitive detection of heterozygous positions (mosaics, somatic mutations etc.)
- Configurable base caller with sequence dependent thresholds; self-adaptive algorithm based on peak area statistic
- Definition of reference electropherograms from previously analysed orders
- Sophisticated tools for frame shift and indel detection



Electropherogram

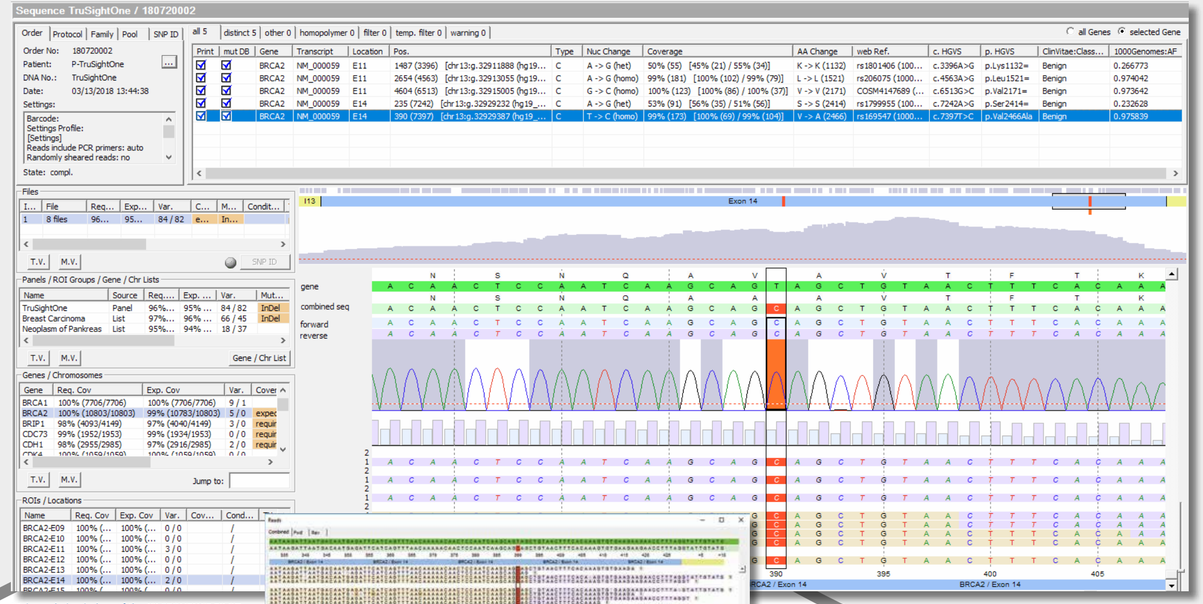


Histogram showing copy number changes

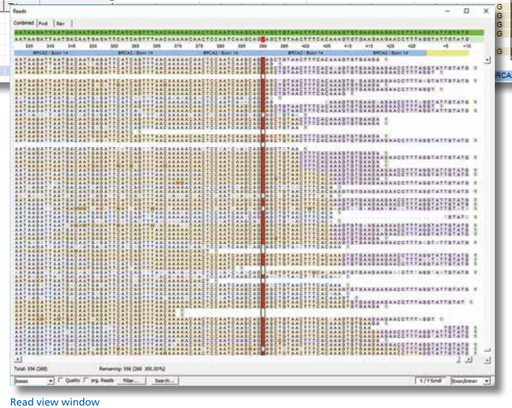
MLPA®

The **MLPA®** module is a convenient and powerful tool for detecting copy number changes.

- Pre-configured SALSA **MLPA®** kit descriptions for download and import
- Different analysis modes for each **MLPA®** kit
- Configurable control settings and RPA limits
- Data correction for probes with „tailing-off-effect"



Main analysis window of the **JSI SEQUENCE PILOT** Software module **SEQNEXT**.
The main results and features are clearly represented. You can retrieve additional information (up to read level) by just one click!



Read view window

SEQNEXT

The **SEQNEXT** module facilitates the convenient and powerful analysis of next-generation sequencing data.

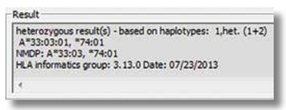
- Fast and easy definition of regions of interest (ROIs) via bed, manifest or CSV files
- Manual generation / import of specific gene lists adressing your interest
- Customizable filters for variants in pseudogenes and homologic regions
- Efficient standards for mapping, alignment and quality
- Personalized settings for analysis and detection of variants (≥ 0.1 %)
- High sensitivity and specificity for detecton of SNPs, deletions and insertions as well as detection of CNV and fustion genes
- Analysis of FFPE samples and processing of Tags (e.g. smMIPs, UMIs etc.)
- Patient identification via SNP IDs
- For small panel to **Whole Exome Sequencing** analysis
- **High speed** analysis: Complete WES in 2.5 hours*

* dependent on your server hardware requirements.
28 Cores à 3.1 GHz, 64GB RAM

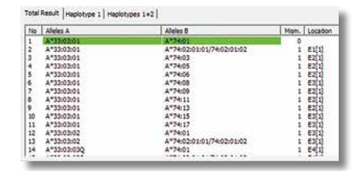
SEQHLA & SEQNEXT-HLA

The **SEQHLA** and **SEQNEXT-HLA** module are convenient and powerful tools for the interpretation of your **HLA** typing data.

- Availability of all IMGT **HLA** database versions and updates for download and import
- Constant positions can be ignored
- Display of results in different resolutions (2-digits, 4-digits and max. resolution)
- Intron sequences can be displayed (not used for result calculation)
- Extended DRB check against pseudogenes
- Splice site mutation warning



HLA typing results



Likely allele combinations