Euformatics

# Sequence variations in their context

## Annotate, classify, report

omnomicsNGS

The **omnomicsNGS** software provides a platform for genomic variant analysis in oncology, constitutive genetics and for rare genetic diseases. It allows diagnostic laboratories to annotate, share, and report on NGS data.

The package comes with annotations and knowledge structures from the most central public sources and provides mechanisms for the users to add and grow their domain-specific knowledge. Use the package on the cloud, as local stand-alone or integrate it into larger IT infrastructures for making patient-centred information securely available to authorised users.

## omnomicsNGS is unique

### Integrate expert knowledge and grow your own

- Integrate the latest annotations from multiple sources including variant, gene, disease, biological pathway, population frequency, structure conservation, protein and drug interaction databases.
- Integrate variant annotations from the user organisation or specialised consortia in addition to CIViC, ClinVar and others.
- Curate variants manually or through batch processes in accordance with ACMG or ASCO best practice guidelines.

### Variant annotation and interpretation at scale

- Store data from and filter any number of subjects and variants.
- Select virtual panels for reporting narrow regions of interest.
- Define biomarkers for immediate reporting on multiple variants.
- Search using multiple filters, which can be saved and modified.
- Zero in on results at the cohort, subject, gene or variant level.
- Produce report content for third party systems and clinicians.

### Automated re-evaluation of variants

- Store sequenced variants, including unclassified variants.
- Store all observed variants to enable calculation of local varianat allele frequencies.
- Report automatically and retroactively when the classification of variants changes as a consequence of knowledge source updates.

Euformatics Oy
Tekniikantie 12, FI-02150 Espoo, Finland

+358 400 359 846
contact@euformatics.com

Euformatics

# Benefits

## Quality

- Integrate with existing secondary analysis tools
- Develop virtual gene panel, filter strategies and biomarker scan routines
- Apply standard ontologies
- Upload NGS data from gene panels, WES, WGS, somatic and germline tests
- Create and freeze routines for, data browsing, and reporting procedures

## Speed

- Scale from single patient testing to support national testing programs: cohorts, carriers, paired samples, trios or families
- Automate variant reporting
- Batch reporting when integrated to supporting IT infrastructure

## Future-Readiness

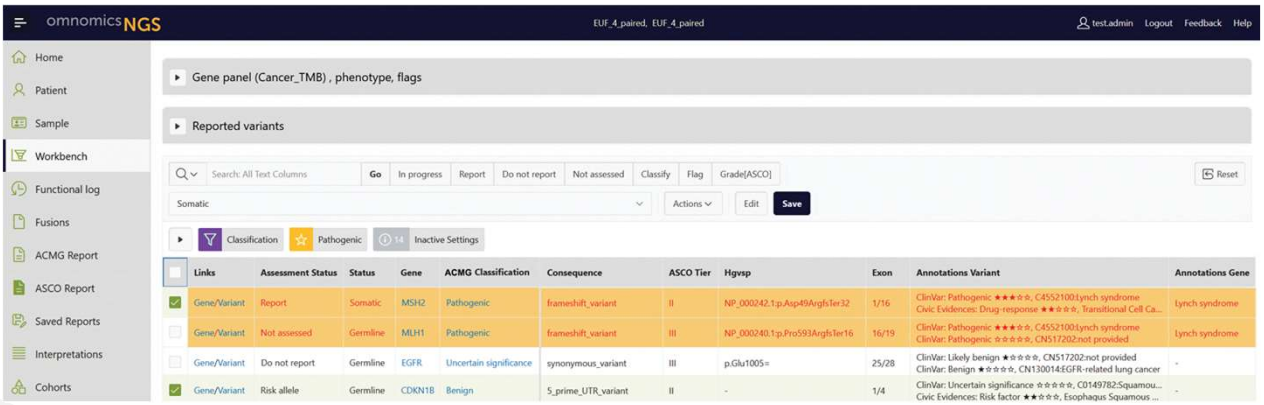
- Add new tests, change strategy, widen the scope of the analyses
- Grow analytic capacity on the basis of accumulating variant data
- Stay up-to-date with best practice reporting guidelines(ACMG, AMP etc.)
- Customise as user the lay-out of the interface and analysis procedures

## Confidence

- Operate securely on the cloud, or inside the user organisation safety boundaries
- HIPAA-compliant cloud platform storage.
- Role-based access

## Validation and QC

Integrate coverage analysis and combine with the automated assay validation and regular quality control with the **omnomicsQ** tool




Euformatics Oy
Tekniikantie 12, FI-02150 Espoo, Finland

+358 400 359 846
contact@euformatics.com