

Disease discovery suites

Comprehensive software packages to explore the genetic background of diseases

A man in a red t-shirt and black pants is captured mid-air, jumping over a deep chasm between two rocky cliffs. The sky is bright blue with scattered white clouds. The man's arms are outstretched, and his legs are in a wide, jumping stance. The background is a vast, open sky.

CHRONIC DISEASES

RARE DISEASES

MALIGNANT DISEASES

INFECTIOUS DISEASES

Diseases discovery suites

Natural variations found in our genes influence our risk of developing a certain disease, and the degree to which it progresses. Understanding these genetic variations help researchers produce better diagnostics and drugs. The four comprehensive solutions offered by Astrid contribute to this discovery process.

Our discovery suites are based on the type of disease that is the focus of investigation: chronic, rare, malignant and infectious diseases. The purpose of the suites is the biological interpretation of structured NGS data – assisting the researcher in revealing the genetic background of diseases or providing an answer to related research questions. Disease discovery suites are built on modules that are selected and then customized according to the needs of the particular research project.

RARE DISEASES DISCOVERY SUITE

The suite aids research focusing on a certain inherited or acquired rare disease. It allows the researcher to discover more about the disease's unique genetic background and its possible consequences.

DE-NOVO MUTATION DISCOVERY MODULE

De-novo mutations usually play a huge role at the pathomechanism of rare diseases. The discovery of such mutations require a unique expertise.

SMALL N SAMPLE SIZE ASSOCIATION ANALYSIS MODULE

Small sample sizes require the application of specific statistical tests to discover relevant phenotype-variant associations during genetic characterization.

MALIGNANT DISEASES DISCOVERY SUITE

The suite allows the genetic characterization of malignancies and the establishment of genetics-tailored therapeutic modalities – contributing to a better understanding of cancer biology.

MUTATION ANALYSIS MODULE

The module combines expression analysis, variant detection, annotation and function prediction to help the researcher identify molecular alterations behind the phenotype transformation.

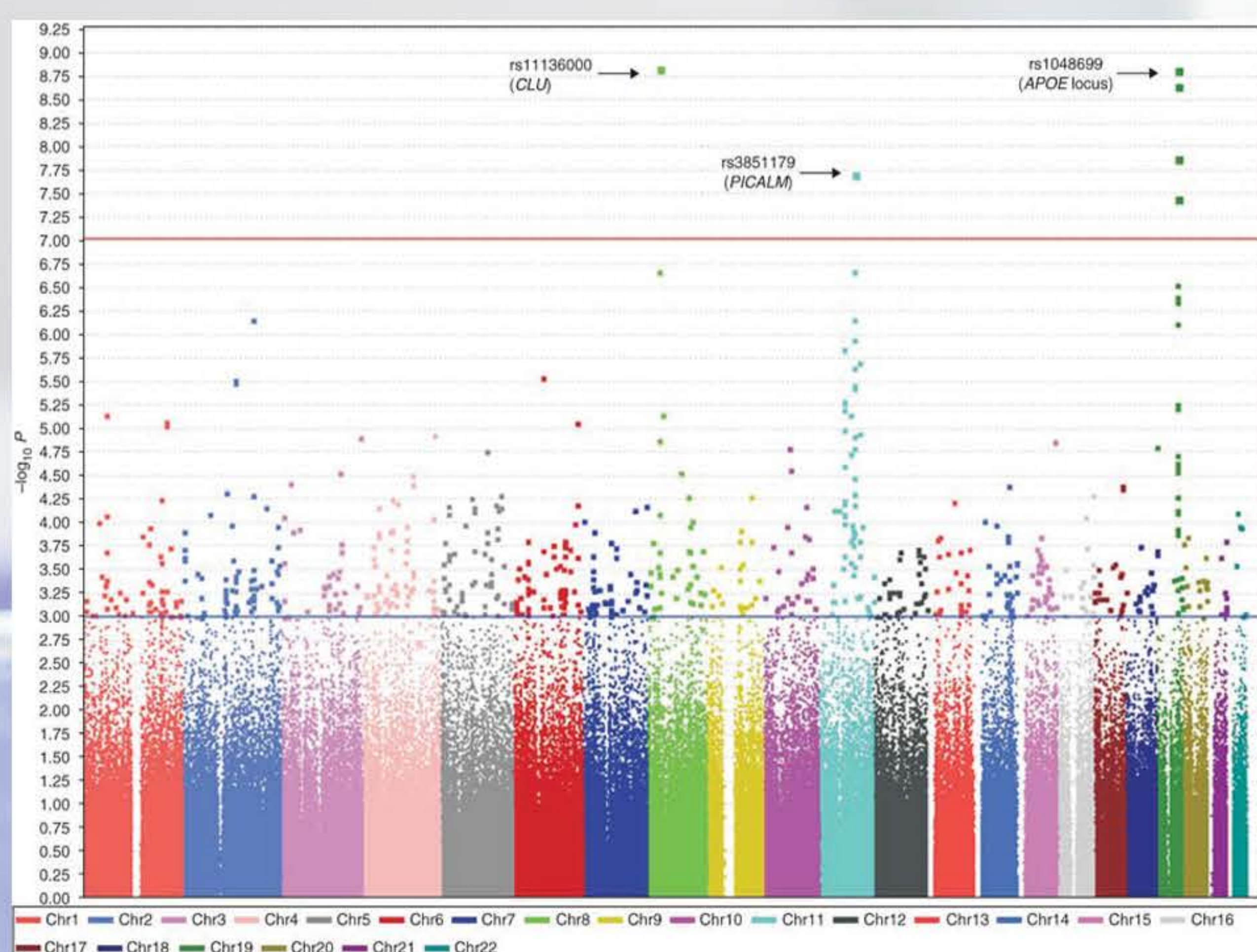
CANCER EVOLUTION ANALYSIS MODULE

Parallel discovery of the individual germline or somatic cell, the primary cancer and the metastasis genotype – revealing genetic alterations behind the cancer evolution sequence.

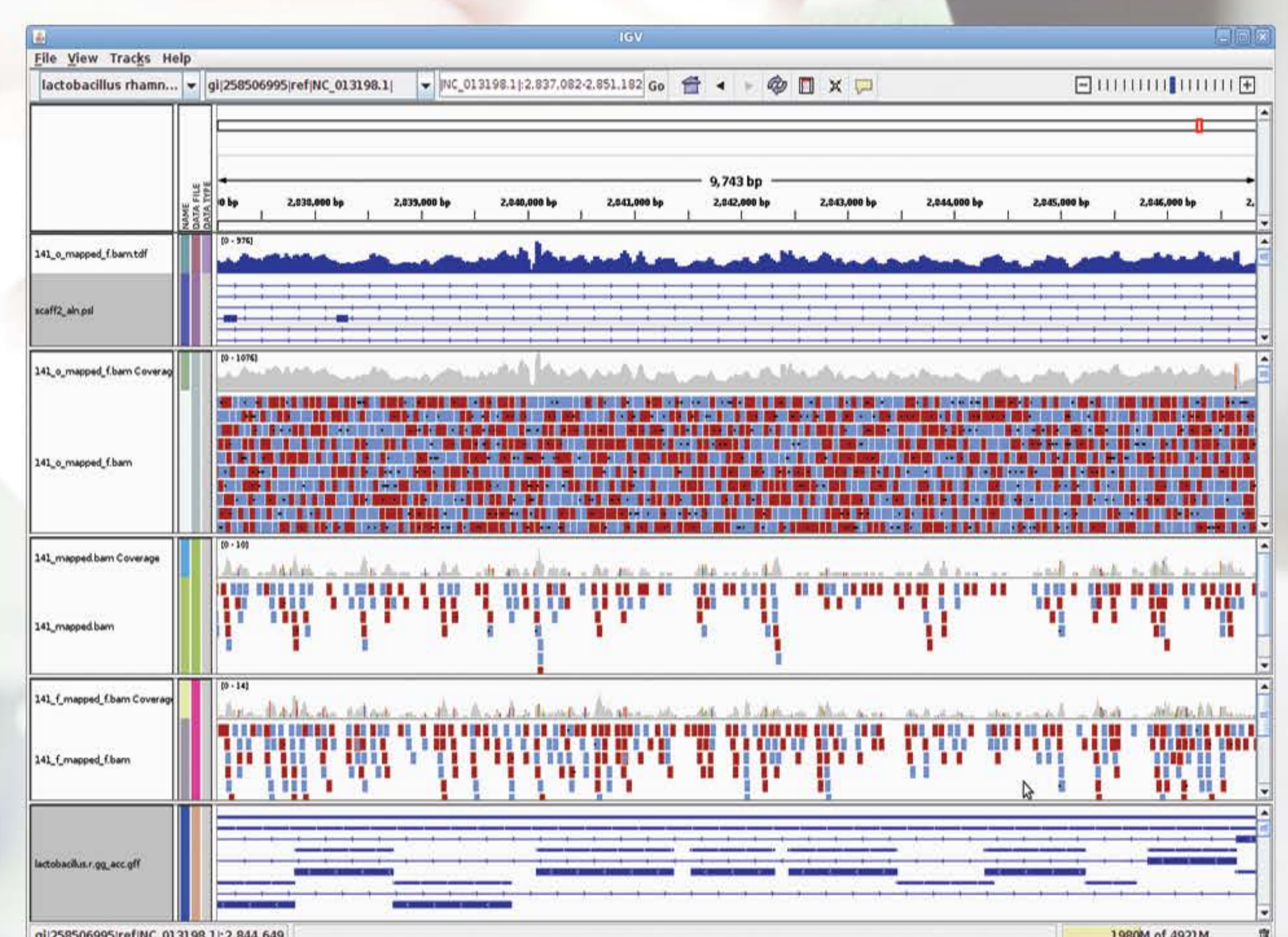
Study design

Biobanking

High-throughput experiment



Visualization of genome-wide association mapping



Visualization of multi-genome comparison

CHRONIC DISEASES DISCOVERY SUITE

Revealing the genetic background of chronic non-communicable diseases is a considerable challenge. The functional interpretation of NGS reads is complicated by the large number of variations and their combined effects contributing to a multifactorial disease.

VARIANT DETECTION AND ANNOTATION MODULE

The result of raw data analysis is an assembled and aligned nucleotide sequence. Further analysis starts with annotation: gene location and function need to be identified. Revealed variants are first filtered then annotated, resulting in dbSNP rs IDs, overlapping-gene accession numbers, SNP function (e.g. missense coding), etc.

PHENOTYPE PREDICTION MODULE

Mapped sequences and variants need to be functionally annotated and assigned. The module performs pathway analysis, function prediction and protein-protein interaction prediction – providing genotype-phenotype associations.

GENOME-WIDE ASSOCIATION ANALYSIS MODULE

Genetic characterization of multifactorial diseases is a complex matter. The output of genome-wide association mapping is a whole genome/exome scanning analysis for statistically strong associations between a set of SNVs/structural variants and a particular trait or disease.

DATA INTEGRATION MODULE

Data originating from different platforms or procedures (microarray, DNA/mRNA/ChIP/miRNA sequencing, proteomic methods, etc.) need to be integrated into a single holistic data warehouse for simultaneous interpretation. The omics module presents data modelling in a system wide context.

INFECTIOUS DISEASES DISCOVERY SUITE

The suite aids at metagenomic studies, at mapping and identifying genes responsible for variable susceptibility to major infectious diseases and at genetic epidemiology studies.

METAGENOMICS MODULE

The module helps at microbial genome sequencing, at studying not easily cultureable organisms or studying organisms in their natural environment.

NON-HOST GENETICS MATERIAL ASSESSMENT MODULE

Provides microbiological research-demanded solutions.

INDIVIDUAL SUSCEPTIBILITY ASSESSMENT MODULE

The module provides assistance at studies examining infectious disease susceptibility and resistance genes.

Raw data
analysis

Data
integration

Data mining,
biostatistics

DISEASE
DISCOVERY
SUITES

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