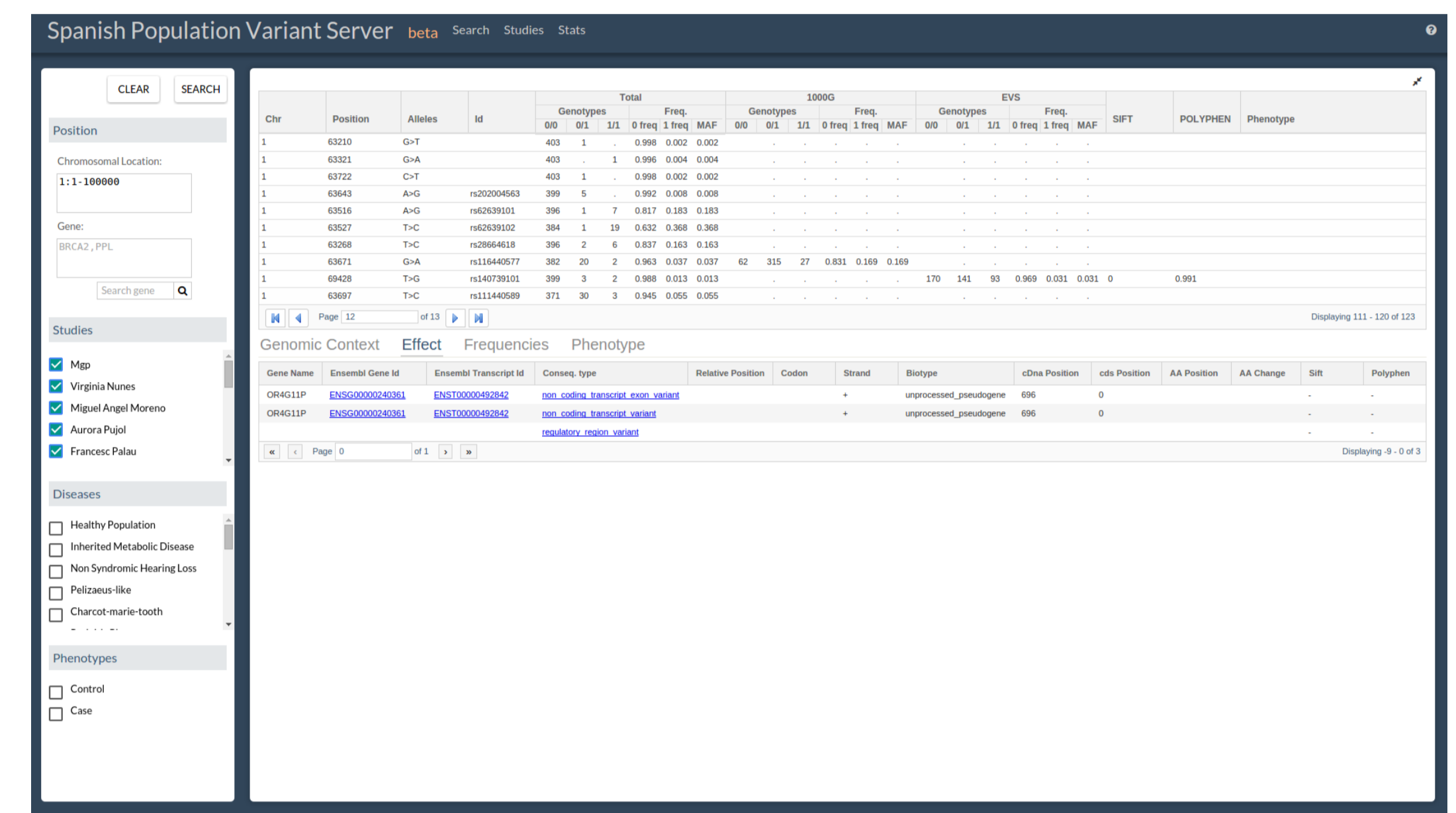
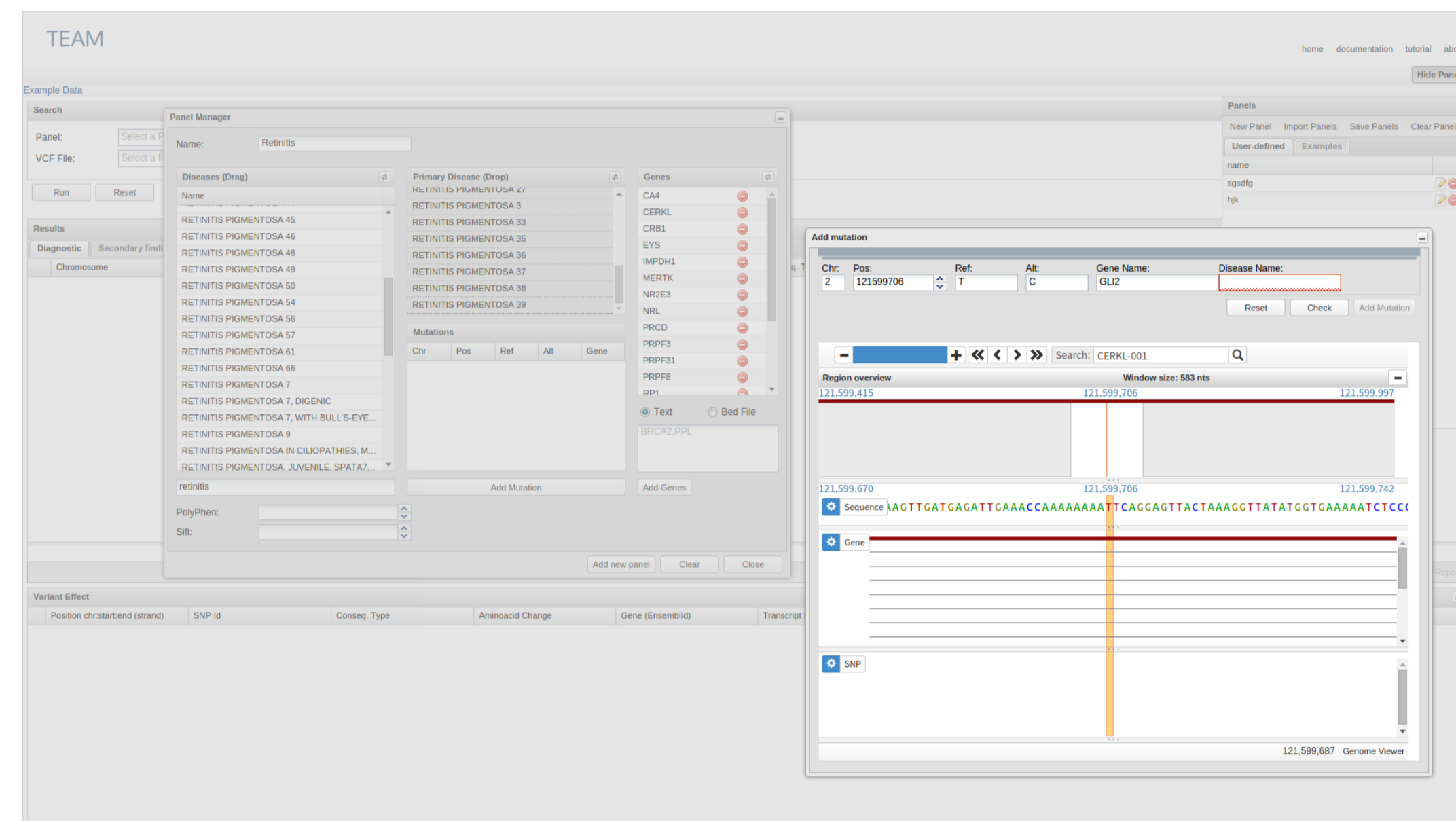
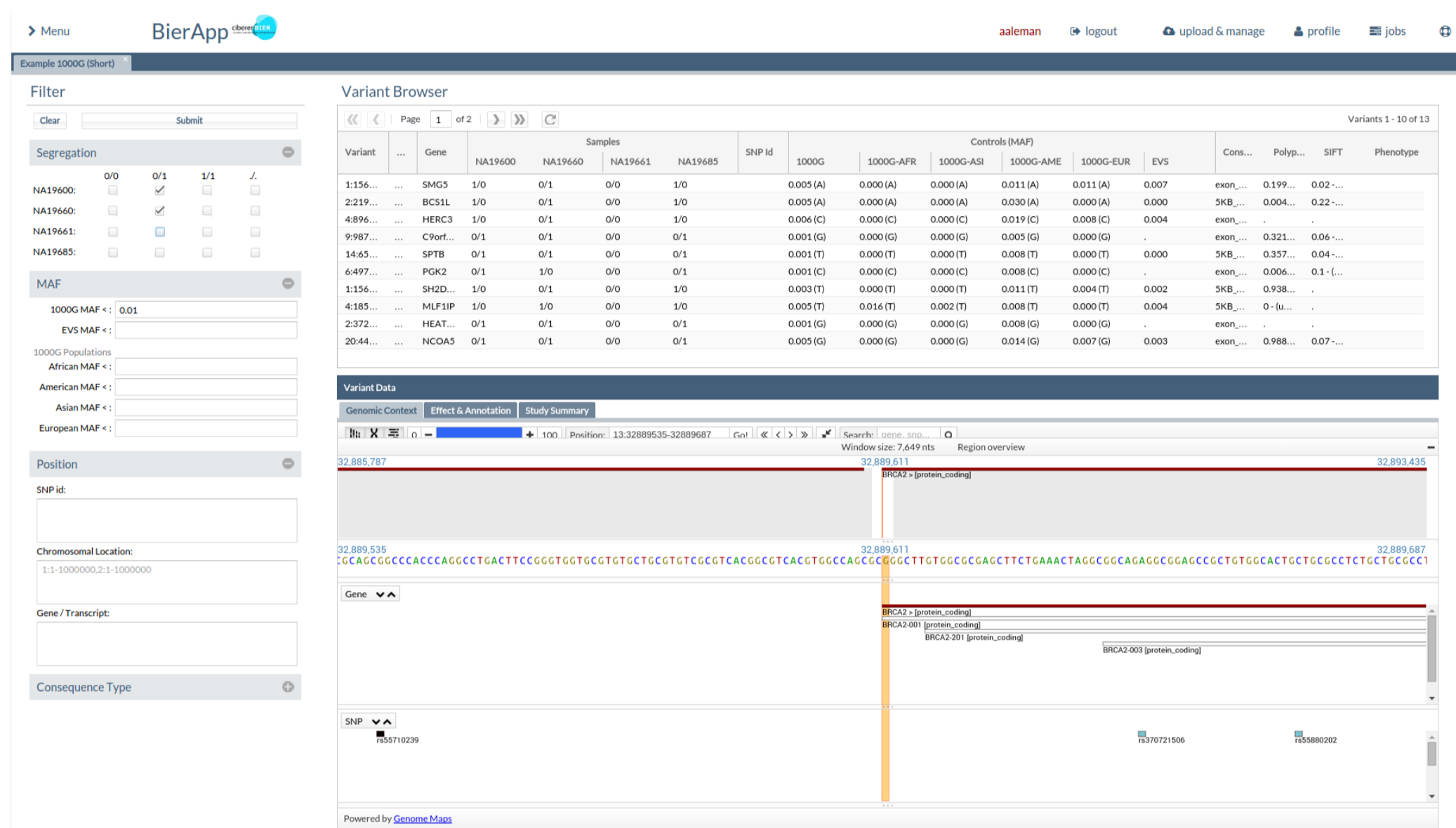




# Bioinformatics solutions for diagnosis by panels and discovery of new variants of disease.

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## BIERApp

BIERApp is an interactive web application for assisting in gene prioritization in whole exome sequencing (WES) experiments. BIERApp is mainly oriented to disease gene finding in Mendelian disorders, although it can be applied to other contexts, such as case-control comparisons. BIERApp has been used by the BIER team (Bioinformatics for Rare Diseases) in different versions during the last year for the analysis of more than 1000 exomes of patients of more than 70 different inherited pathologies, produced by the Spanish Network for Research in Rare Diseases (CIBERER) and the Medical Genome Project (MPG).  
<http://bierapp.babelomics.org>

## TEAM

TEAM (Targeted Enrichment Analysis and Management) is an open web-based tool for the design and management of panels of genes for targeted enrichment and massive sequencing for diagnostic applications.

TEAM allows users to define their own panels of genes. The definition is based on disease terms obtained from different databases (HGMD-public, HUMSAVAR, ClinVar and COSMIC). Each disease term has the corresponding disease genes associated. Thus, when a VCF file is uploaded in the systems, TEAM searches for known diagnostic mutation(s).

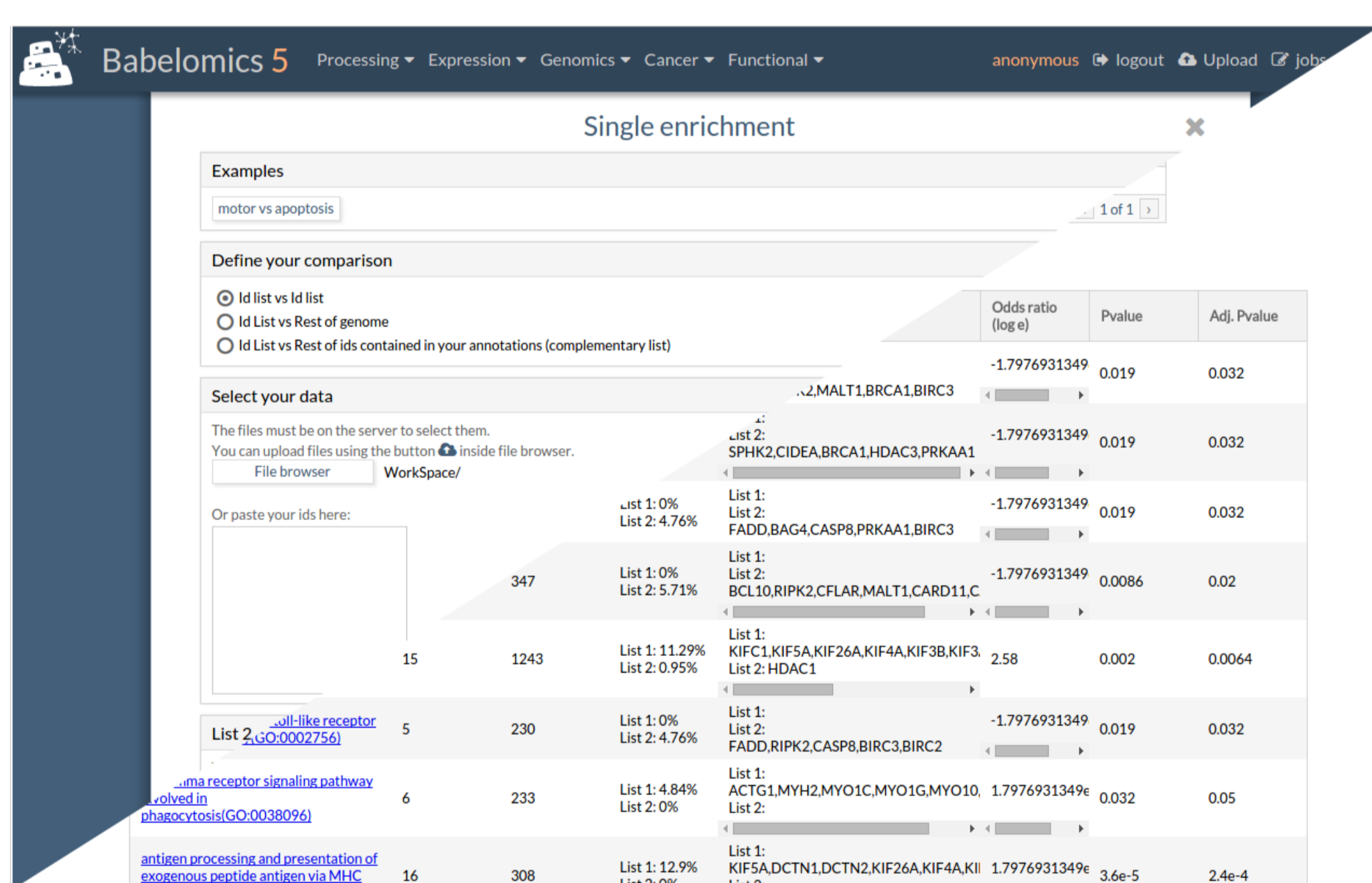
<http://team.babelomics.org>

## CIBERER Spanish Variant Server

This tool was created to provide information about the variability of the Spanish population to the scientific/medical community. It is useful for filtering polymorphisms and local variations in the process of prioritizing candidate disease genes. CSVS currently stores information on 400 unrelated Spanish individuals. We accept submissions from WES or WGS. See the protocol for sending samples.

<http://bioinfo.cipf.es/apps-beta/spvs/1.0.0/>

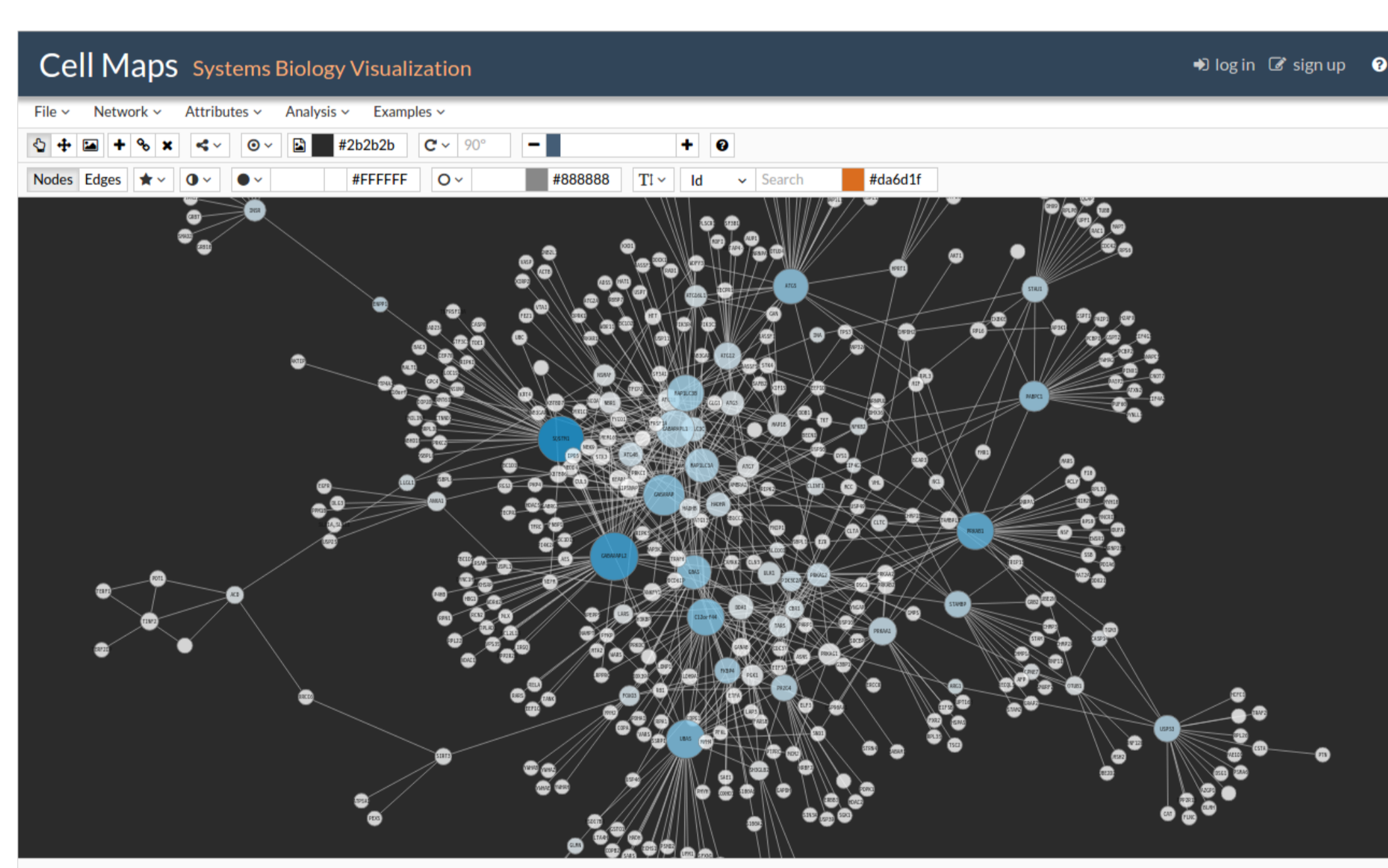
Protocol:  
<http://bioinfo.cipf.es/apps-beta/spvs/protocol.pdf>



## Babelomics 5

Babelomics is an integrative platform for the analysis of Transcriptomics, Proteomics and Genomics data with advanced functional profiling. This new version of Babelomics integrates primary (normalization, calls, etc.) and secondary (signatures, predictors, associations, TDTs, clustering, etc.) analysis tools within an environment that allows relating genomic data and/or interpreting them by means of different functional enrichment or gene set methods. Such interpretation is made using functional definitions, protein-protein interactions...

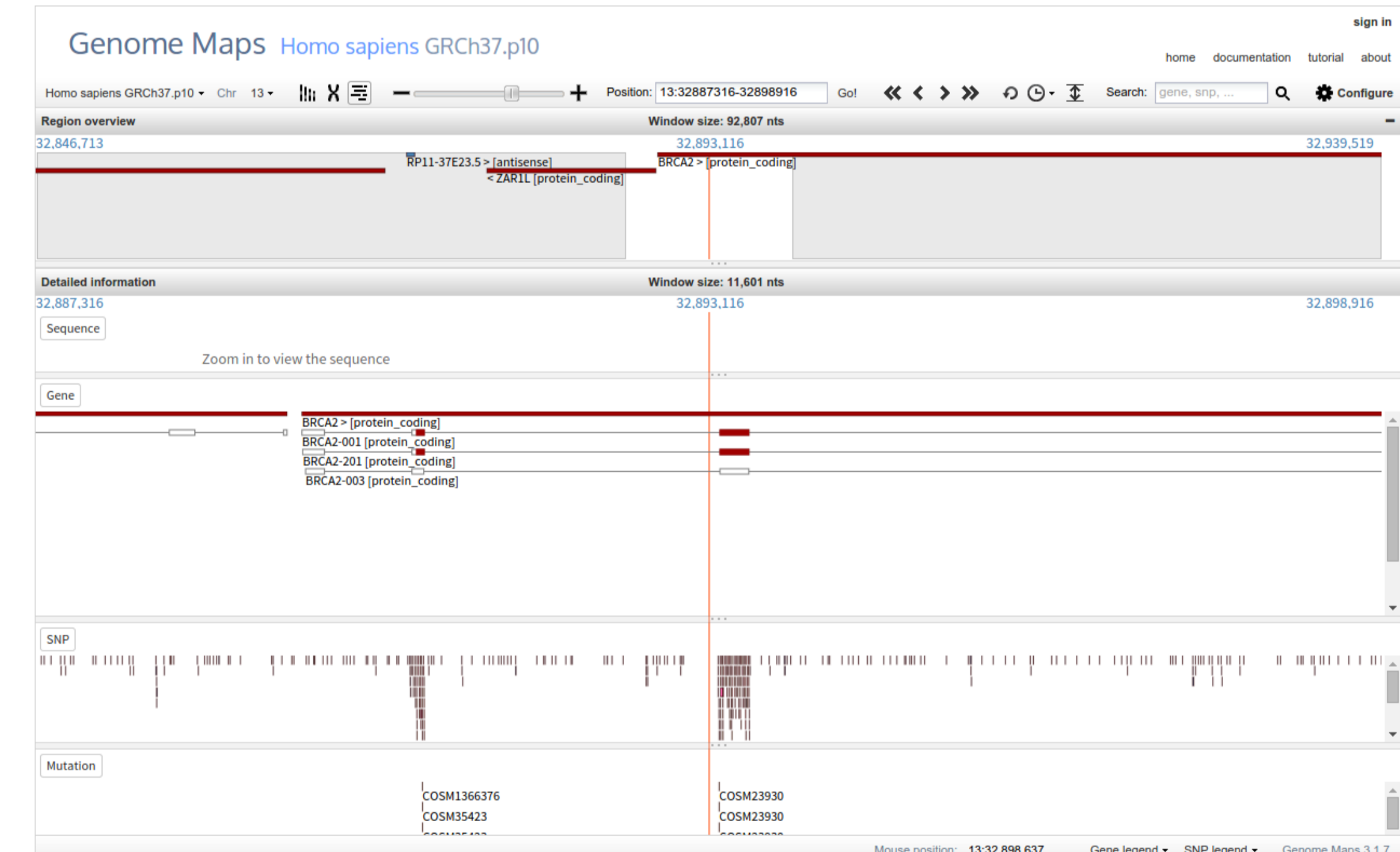
<http://babelomics.org>



## Cell Maps

CellMaps is an open source HTML5 web-based application that allows researchers to easily model, visualize, integrate data and analyse biological networks inside a web browser. It has been designed in a modular fashion to be easily integrated into different web sites, and implemented using modern web technologies to be interactive even when large networks with hundreds of nodes are loaded.

<http://cellmaps.babelomics.org>



## Genome Maps

Genome Maps is an open source and high-performance HTML5 web-based genome browser. Genome Maps allows to browse several genomes and annotations by fetching data from CellBase high-performance REST web services. Custom data (BAMs, VCFs, ...) can also be loaded from remote servers and local disk by using OpenCGA server.

<http://genomemaps.org>