Web tools for the analysis of genomic data and the discovery of new disease genes

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Personalized Medicine

BiERapp

BiERapp is an interactive web application for facilitating 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 MedicalGenome Project (MPG).

http://bierapp.babelomics.org

Databases

CIBERER Spanish Variant Server

The CSVS is the interface to database of genetic variability of the Spanish population. The database provides aggregated information on the frequency at which variants are present in genes or genomic regions in a sample of the Spanish population, along with complementary information that includes other population variability data (1000 genomes, ESP, ExAC) and several indexes of pathogenicity, conservation, and known phenotypes. CSVS is useful for filtering polymorphisms and local variations in the process of prioritizing candidate disease genes. CSVS currently stores information on 790 unrelated Spanish individuals, collected from different CIBERER groups and associated diagnostic laboratories, which includes almost 400 healthy controls and individuals with different diseases that can be selected or deselected for the calculations of genotypic frequencies of (pseudo-)controls.

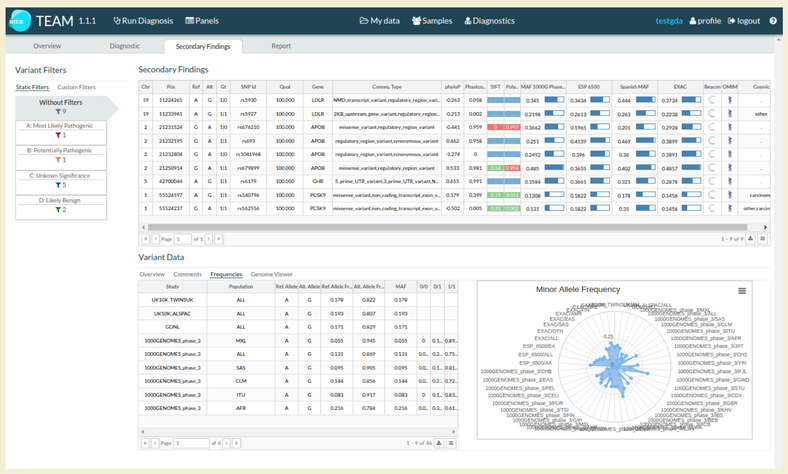
https://csvs.babelomics.org

Precision Medicine

hiPathia

hiPathia is a web tool for the interpretation of the consequences of the combined changes of gene expression levels and/or genomic mutations in the context of signaling pathways. hiPathia uses a model that infers signaling pathway activity from gene expression and/or genomic variation data. Such activities carry information on the different cell functionalities triggered by them. Signaling activities from hipathia not only account for the underlying molecular mechanisms of diseases or the mode of action of drugs but they can also be used as mechanistic biomarkers for the prediction of complex phenotypes.

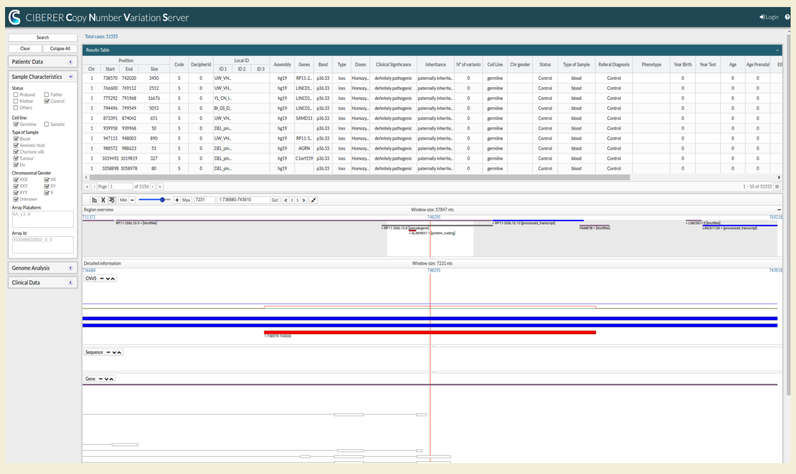
http://hipathia.babelomics.org/



TEAM

TEAM (Targeted Enrichment Analysis and Management) is an interactive web application for the management of targeted enrichment and massive sequencing data for diagnostic applications.
TEAM allows users to define their own panels of genes or genomic regions. The definition is very intuitive and 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). If found, a report is generated, otherwise provides tools for the prioritization for variants of uncertain significance. TEAM has different options to manage unexpected findings.

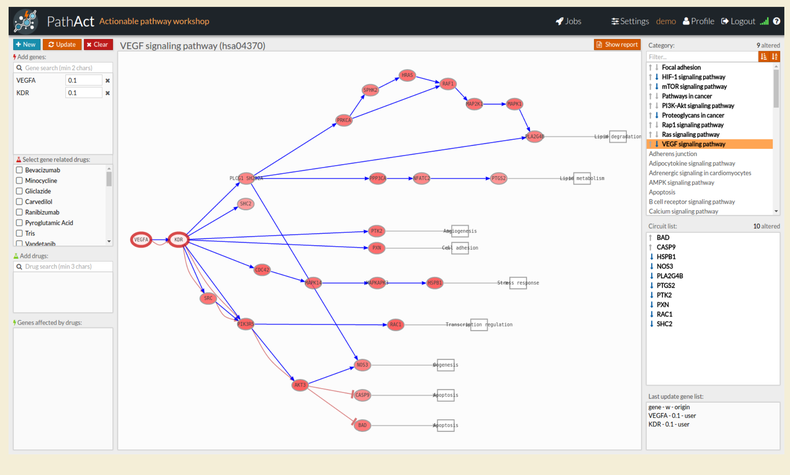
http://team.babelomics.org



Copy Number Variation Server

The Copy Number Variation Server (CNVS) gathers information of copy number variants (CNVs) found in Spanish patients of numerous diseases as well as the CNVs from the 1000 genomes populations. The CNVS can be queries in many different ways and provides a comprehensive perspective of the variability at the level of CNVs, including data on ethnicity, geographic location, genes affected, phenotype (if available) and other. Queries include patient-related data (ethnicity, geographic location, age, etc.), sample-related data (status, type of sample, array platform, etc.) genomic data (gene, chromosomal region, type of variant, doses, etc.) and clinical data (referral diagnosis, phenotype, syndrome, inheritance, etc.) Results provise a list of the variants found in the query than can be represented in their genomic context, along with genes, SNPs, and other genomic features.

https://cnvs.babelomics.org

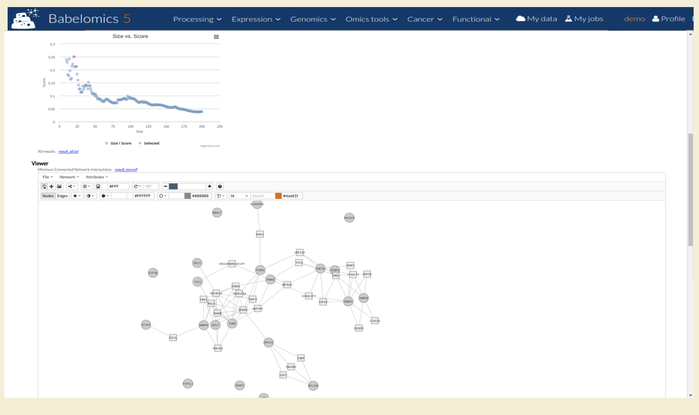


PathAct

PathAct (from Actionable Pathway) is a translational application of the hiPathia models. PathAct enables the study of the consequences that knock-outs(KOs) or over-expressions of genes can have over signaling pathways and, consequently, in the cell functionality. PathAct is a web tool that implements the hiPathia models within an advanced graphical interface that provide a unique interactive working environment in which actionable genes, that could become potential drug targets, can be easily assayed alone or in combinations. Also the effect of drugs with known targets over the different signaling pathways can be studied. Since signals trigger functions across the pathways, the direct and long-distance functional consequences of interventions over genes can be straightforwardly revealed through this actionable pathway scenario. PathAct enables the design of rational personalized interventions in patients.

http://pathact.babelomics.org/

Genomic Analysis



Babelomics

Babelomics is an integrative platform for the analysis of Transcriptomics, Proteomics and Genomics data with advanced downstream functional analysis. Babelomics integrates primary (normalization, calls, etc.) and secondary (signatures, predictors, associations, clustering, etc.) analysis tools within an environment that allows relating genomic data and/or interpreting them by means of different functional enrichment, gene set or network analysis methods. Such interpretation is made using functional definitions, protein-protein interactions, etc. Different versions of Babelomics are running for more than a decade. Currently, the number of citations amounts to more than 2000, has a record of approximately 50,000 uses per year and has about 1000 registered users.

http://babelomics.org