BiER collaborative projects

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Aim

BiER (Bioinformatics Platform for Rare Diseases; http://www.ciberer.es/bier) is a transversal working group whose function, in collaboration with the INB, is to provide bioinformatic and technological support to experimental and clinical groups for the integration, analysis and interpretation of biomedical data (structural and functional genomics, modeling and molecular dynamics, metabolism, relationship networks genes-phenotypes/disease), with an intense collaborative relationship within the context of intramural sequencing projects.

Methods

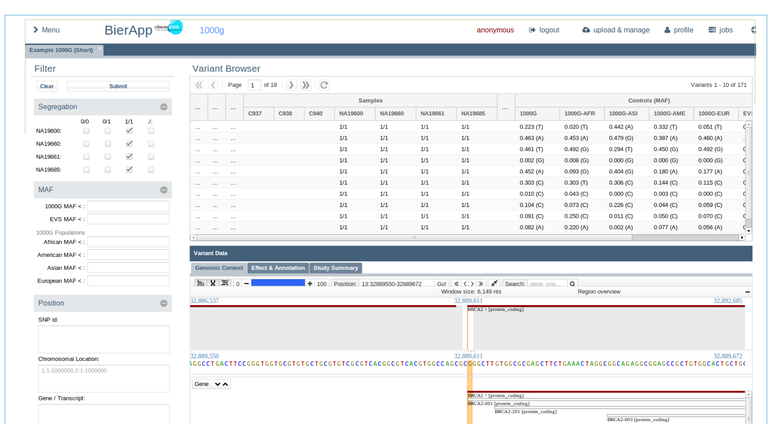
- This bioinformatic and technological support includes advice on the experimental design, analysis strategy and interpretation of data.
- BiER has designed and improved pipelines for Genomics and Transcriptomics sequencing data analysis and developed web tools and methods to analyze and prioritize genes or mutations for diseases.
- Several training activities were carried out to facilitate the understanding and management of data.

Results

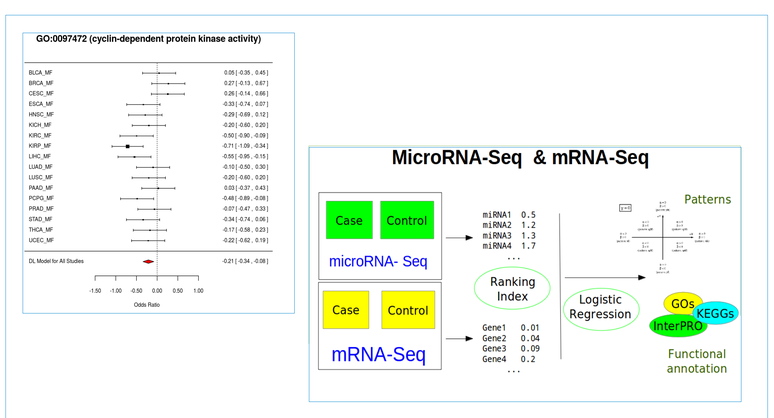
During the last years the BiER platform has collaborated with more than **25 CIBERER groups**:

- Technological and bioinformatic support in **70 projects**.
- Collaboration **intra-groups** receiving more than **50 reserarchers** for stays in our unit.
- Results of these analysis and bioinformatic developments have contributed to the discovery of **13 new disease genes in which 27 new mutations** were identified and the identification of 36 new causal mutations in known disease genes, and also generated **56 collaborative scientific publications** in the last three years (http://bioinfo.cipf.es/publications) (1)(2)(3)

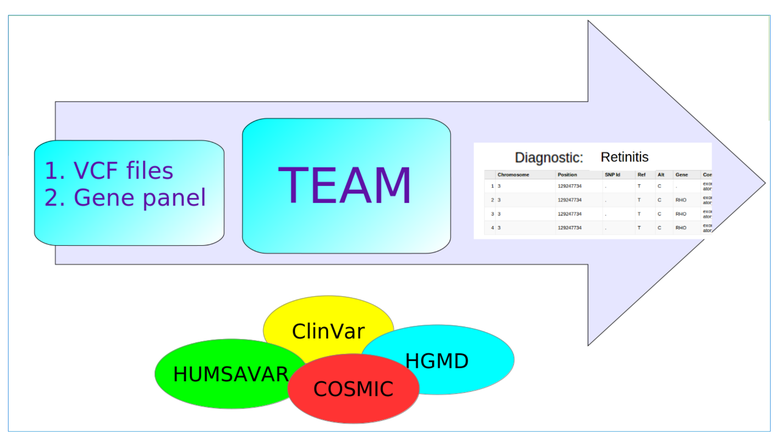
- **Training activity** "NGS course: from reads to candidate genes which has been held during the last 4 years with an average of 25 attendants from different groups CIBERER per edition.
- Several **web tools** were generated to analyze and improve the management of results: **BiERapp**(4), a web-based interactive framework to assist in the prioritization of disease candidate genes in whole-exome sequencing studies. **TEAM** (5), a web tool for the design and management of panels of genes for targeted enrichment and massive sequencing for clinical applications. **CSVS** (CIBERER Spanish Variant Server) and **Babelomics** (6),
- **Development of new methods** of transcriptome analysis in the context of signaling pathways, functional meta-analysis and functional enrichment analysis for microRNAs.



http://bierapp.babelomics.org/



Development of new analysis methods



http://team.babelomics.org/

Conclusions

- Interaction between research groups and BIER platform has been an important factor in web design, adjustment tools and methods for analyzing sequencing data and its interpretation.
- The results obtained from the analyzes have provided a better understanding of the genomic data of these diseases, as well as the detection of biomarkers that can be used in the prevention, diagnosis and clinical therapy design.

References

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