Web tools to analyze and prioritize genes or mutations for diseases

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Aim

- Recent advances in genome sequencing technologies have enabled a breakthrough in the investigation of the genetic basis of disease. However, the resolving power of these technologies has shifted the bottleneck of the discovery process from the production to the **statistical data analysis** phase.
- Our team (**BIER**: Bioinformatics Platform for Rare Diseases; http://www.ciberer.es/bier) has developed several we tools aimed at different kind of statistical analysis: **discovery of new variants, disease diagnosis and visualization of genomics results**.

Methods

- BiER has designed pipelines for Genomics and Transcriptomics sequencing data analysis.
- These web applications makes an intensive use of new web technologies and standards like HTML5. R (free software environment for statistical computing) was used for analysis modules.
- Several training activities were carried out to facilitate the understanding and management of data.

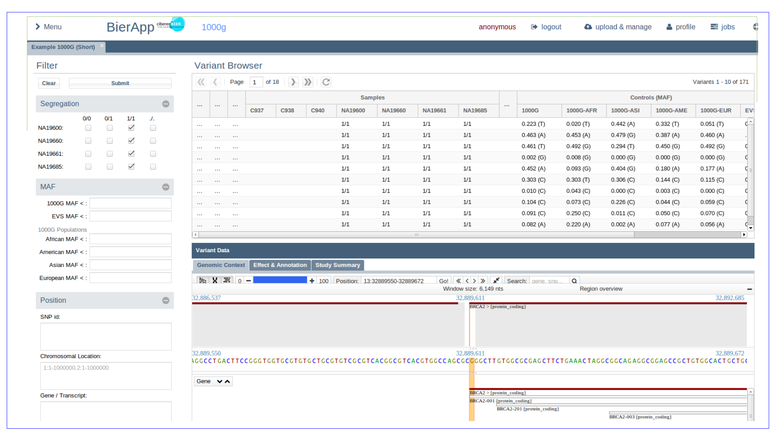
Results

- Scientific collaborations took place among **19 CIBERER research CIBERER.**

- **Recent publications** include the discovery of two new mutations in the BCKDK gene, responsible of a neurobehavioral deficit in pediatric patients (1), new mutations in different genes causing inherited retinal dystrophies (2) and metabolic diseases (3).

- These **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 secuencing studies.
- **ExomeServer**. Created with the intention to provide the scientific and medical community, information about the variability in the Spanish population. It is useful for filtering polymorphisms and local variants.
- **TEAM** (5). A web tool for the design and management of panels of genes for targeted enrichment and massive sequencing for clinical applications.
- **BABELOMICS** (6). Gene expression, genome variation and functional profiling analysis suite (http://babelomics.bioinfo.cipf.es/).



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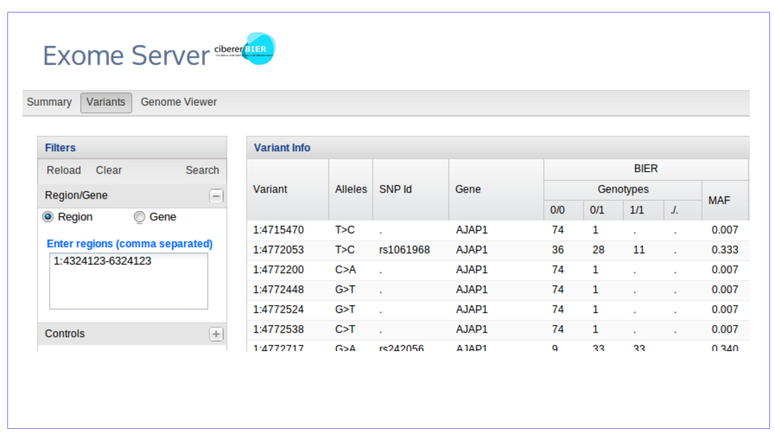

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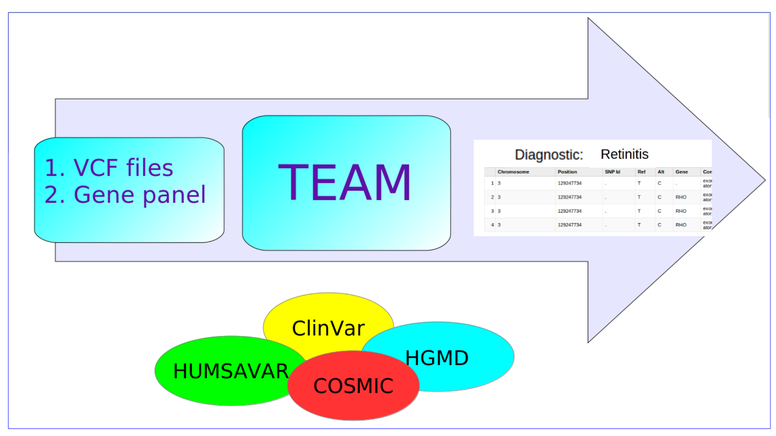

http://team.babelomics.org/

Conclusions

- Interaction between research groups and BIER platform has been an important factor in web design and adjustment tools 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.
- The use of web tools improved the skills of researchers in the statistical analysis of genomic data.

References

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PRINCIPE FELIPE CENTRO DE INVESTIGACION Computational Genomics

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