BIER platform: analyzing and understanding genomic and biomedical data

Francisco García-García[1], Alejandro Alemán[1,2], Joaquín Dopazo[1,2,3]

1 Systems Genomics Lab, Computational Genomics, Centro de Investigación Príncipe Felipe (CIPF), Valencia, Spain.
2 BIER, CIBER de Enfermedades Raras (CIBERER). 3 Functional Genomics Node (INB)

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).

Methods

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

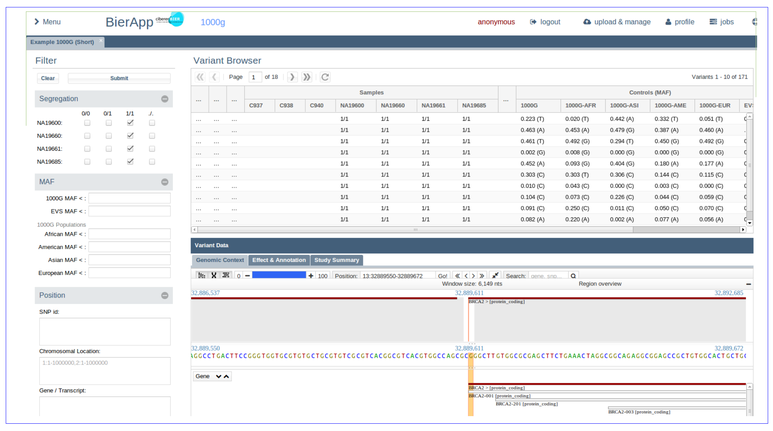
Results

Scientific collaborations took place among **19 groups CIBERER**:

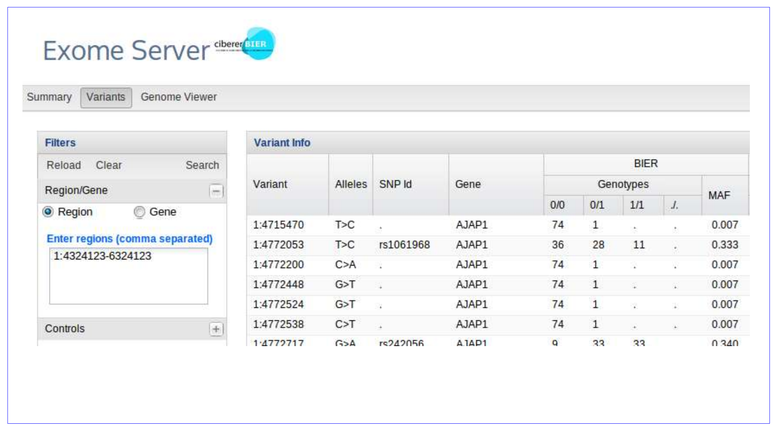
- **173 exomes were analyzed** in 94 different families.
- After including new methods in the pipeline, **we reanalyzed 72** of the previous exomes to refine the selection of candidate variants.
- **Recent publications** include the discovery of two new mutations in the BCKDK gene, responsible of a neurobehavioral deficit in pediatric patients (1), new mutations in different genes causing inherited retinal dystrophies (2) and metabolic diseases (3).

Several **web tools** were generated to analyze and improve the management of results:

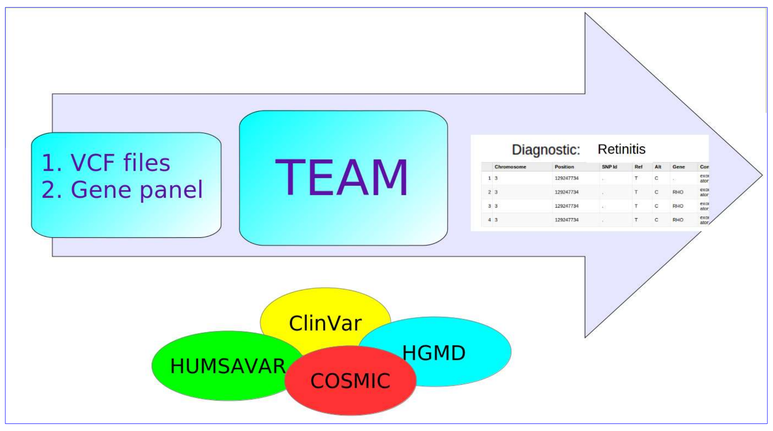
- **BiERapp** (4). A web-based interactive framework to assist in the prioritization of disease candidate genes in whole-exome secuencing studies.
- **ExomeServer**. Created with the intention to provide the scientific and medical community, information about the variability in the Spanish population. 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.



http://bierapp.babelomics.org/



Exome Server



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.

References

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