BIER platform: analyzing and understanding genomic and biomedical data

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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).

Methods

- This bioinformatic and technological support includes advice on the experimental design, analysis strategy and interpretation of data.
- BiER has optimized 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.

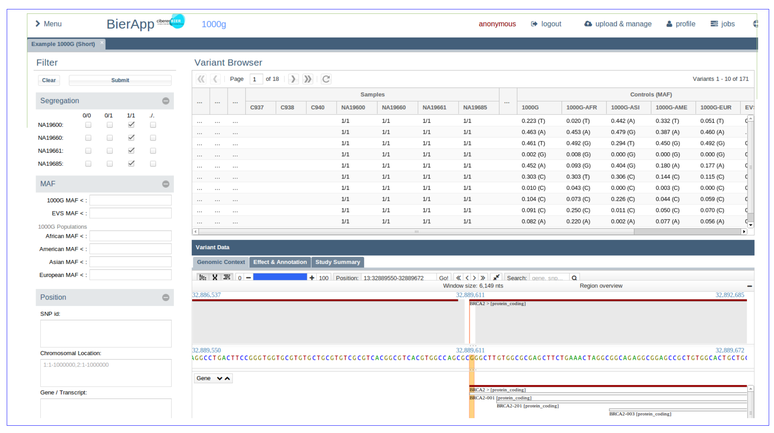
Results

Scientific collaborations took place among several research groups (this involved a total of 24 publications in 2014) :

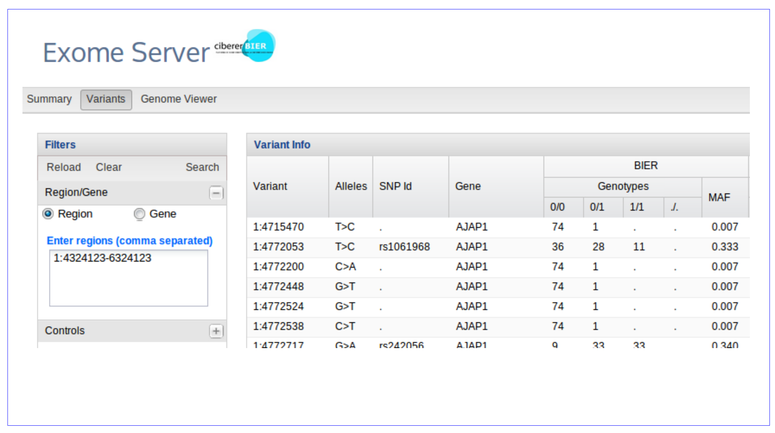
- Medical Genome Project (http://www.medicalgenomeproject.com/)
- Collaborations between CIBER groups: CIBERESP, Carlos Martín.
- CIBERER activity:
  - 212 exomes were analyzed in 131 different families.
  - After optimizing the analysis pipeline, we reanalyzed 72 of the previous exomes to refine the selection of candidate variants.
  - In addition, we have studied a total of 76 samples of panels of different diseases.
  - 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).

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

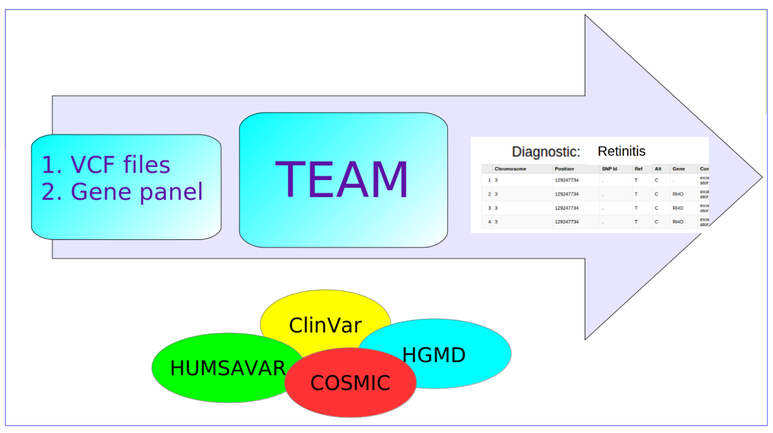
- BiERapp (4). A web-based interactive framework to assist in the prioritization of disease candidate genes in whole-exome secuencing studies.
- ExomeServer. Created with the intention to provide the scientific and medical community, information about the variability in the Spanish population. It is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



Exome Server



http://ciberer.es/bier/team

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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