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BOOK OF ABSTRACTS

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BIER platform: analyzing and understanding genomic and biomedical data

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INTRODUCTION

BiER (Bioinformatics Platform for Rare Diseases; http://www.ciberer.es/bier) is a transversal working group whose function is to provide experimental and clinical groups CIBERER, bioinformatic and technological support needed for the integration, analysis and interpretation of biomedical data (structural and functional genomics, modeling and molecular dynamics, metabolism, relationship networks genes-phenotypes/disease).

METHODS

BiER has designed pipelines for Genomics and Transcriptomics sequecing data analysis and developed web tools to analyze and prioritize genes or mutations for diseases. This bioinformatic and technological support includes advice on the experimental design, analysis strategy and interpretation of data. 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, new mutations in different genes causing inherited retinal dystrophies and metabolic diseases.

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

1. BiERapp. A web-based interactive framework to assist in the prioritization of disease candidate genes in whole-exome secuencing studies.

2. 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.

3. TEAM. A web tool for the design and management of panels of genes for targeted enrichment and massive sequencing for clinical applications.

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.

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Integrated Gene Set Analysis for microRNA Studies

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INTRODUCTION From a systems biology perspective, gene set analysis (GSA) allow us to understand the molecular basis of a genome-scale experiments. Gene set methods are much more sensitive than single enrichment methods in detecting gene sets (defined as sets of genes with a common annotation) with a joint implication in a genomic experiment. But currently there are not GSA methods tailored for the miRNA context. In this work we present a novel approach to the functional interpretation of miRNA studies which keeps the advantages of the GSA.

METHODS We downloaded 20 datasets from The Cancer Genome Atlas (http://cancergenome.nih.gov/), containing tumoral and normal samples. Differential expression analysis was carried out for mRNA and miRNA levels (Bioconductor library edgeR). Information from miRNA was transferred to gene level by adding its effects and generating a new index which ranks genes according to their differential inhibition by miRNA activity across biological conditions. Given such ranking statistics of the genes for each functional class, we apply the logistic regression models for GSA. P-values were corrected for multiple testing using the method Benjamini and Yekutieli.

RESULTS This new approach has allowed to obtain a genomic functional profiling for different cancers when using miRNA data. In our study we used Gene Ontology terms (http://www.geneontology.org/) to define gene sets, obtaining detailed functional results for each ontology (biological process, cellular component and molecular function).

CONCLUSIONS This method may be successfully applied in genomic functional profiling, transferring miRNA data to gene level so that GSA can be properly aplyed. Functional results take advantage of the knowledge already available in biological databases and can help to understand large-scale experiments from a systems biology perspective.