



Bioinformatics Challenges in Personalized Medicine

Bioinformatics is expected to play a key role in the advancement of new diagnostic, therapeutic and predictive solutions in personalised medicine. This session is aimed at promoting an open debate in the room about what is needed to facilitate a bioinformatics pipeline in this context of Personalised medicine. We shall discuss which is the state of the art, some of the success stories and scientific and technical challenges in the area. We expect that the audience will share their own experiences and involvement in related projects.

Introduction: Alfonso Valencia, CNIO

Chairs:

- Jose Pereira Leal, Instituto Gulbenkian de Ciência
- Fernando Martin-Sánchez, Instituto de Salud Carlos III

Date: Wednesday 27th October.

Place: Hotel Son Don Pablo. Sala Segovia.

Program:

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| 17-17:15 | Introduction. Setting the scene. Introduction: Alfonso Valencia Moderators: José Leal Pereira (Gulbenkian)/ Fernando Martin-Sanchez (ISCIII) Case studies: 1) Core signaling pathways in Human pancreatic cancer (CNIO) 2) Clinical assesment incorporating a Personal Genome (Stanford U.) |
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| 17:15-18:00 | <p>Open debate: Bioinformatics challenges in a personalized medicine pipeline</p> <p>Chairs: José Leal Pereira/Fernando Martin-Sanchez</p> <p>STRUCTURAL & FUNCTIONAL ANALYSIS OF THE PATIENT GENOME</p> <p>1.- Analysis of NGS data. Finding mutations, assigning confidence values and validating them. 2.- Analysis of mutations in coding and non-coding regions and assigning them disease-risk. 3.- Treatment of structural variations and copy number variations. 4.- Functional analysis (gene expression, proteomics, miRNA, epigenetics,...).</p> <p>SYSTEMS BIOLOGY</p> <p>5.- Network/Pathway based interpretation of patient genomic data in the context of disease knowledge. 6.- Network/Pathway based interpretation of patient genomic data in the context of drug knowledge (previous compilation of drugs target and mutation disease relations). 7.- Analysis of Networks/Pathways altered among different diseases</p> <p>CLINICAL RESEARCH</p> <p>8.- Analysis of genome specific toxicity/drug responses and medical history of the patient 9.- Analysis of the results in animal models (if possible) 10.- Handling of clinical data through disease ontologies, information extraction from text and related phenomic issues</p> <p>CLINICAL PRACTICE</p> <p>11.- Linking the patient clinical record with individual genomic data. 12.- Assisting the physician in medical decision making for diagnosis and treatment(i.e. complementary tests, prognosis, suggestions of potential and dosages). 13.- Consultation with the patient (who may have its own genome data) and follow-up of the results and education of health professionals in Bioinformatics</p> |
| 18:00:18:30 | <p>Contributions from the audience on specific on-going work and projects on related areas.</p> |