New methods to evaluate the impact of single point protein mutation on human health

Fact Sheet

Project Information

MUT2DIS

Grant agreement ID: 237225

Project closed

Start date
1 September 2009

End date
31 August 2012

Funded under
Specific programme "People" implementing the Seventh Framework Programme of the European Community for research, technological development and demonstration activities (2007 to 2013)

Total cost
€ 217 113,92

EU contribution
€ 217 113,92

Coordinated by
UNIVERSITAT DE LES ILLES BALEARS
Spain

Objective

The Single Nucleotide Polymorphisms (SNPs) are the main cause of the sequence variability in human. For this reason, the prediction of the impact of SNPs is one of the most challenging problems in computational biology and bioinformatics. The

increasing growth of the genome-scale studies is providing a large amounts of SNP data, allowing the application of rigorous statistical approaches for predicting whether a given single point mutation has an effect on human health. In spite of the scientific effort, the identification of disease-associated human SNPs remains a difficult task and a satisfactory solution of general applicability is yet unavailable. This proposal is focused on the study of those SNPs, called missense SNPs (mSNPs), that are responsible of single protein mutation, because the mSNPs can be related to the insurgence of pathologies decreasing the stability or the functional activity of the protein. The main aim of this project concerns the development of new machine learning based methods to predict the effect of a given single point protein mutation. A set of mSNPs corresponding to proteins with known with atomic resolution will be collected and studied evaluating the evolutionary information retrieved using the protein sequences and the structural information derived from the protein 3D structures. The description of the structural and evolutionary related features will allow the implementation of more accurate predictors able to discriminate between disease causing or neutral polymorphisms. In a second part of the project, the resulting predictors will be also used to elucidate the relationships between mSNPs and the resistance to a given drugs. This aspect will be vary important in order to evaluate which therapy could have better effect on a particular phenotype. All the results of this research as well as the implemented predictors will be made available online to scientific community.

Fields of science (EuroSciVoc)

natural sciences > biological sciences > biochemistry > biomolecules > **proteins**

natural sciences > biological sciences > genetics > **mutation**

medical and health sciences > basic medicine > **pathology**

natural sciences > biological sciences > genetics > **nucleotides**

natural sciences > computer and information sciences > artificial intelligence > **machine learning**

Keywords

Bioinformatics | Biological sciences | Computational biology | Computer science | Structural biology

Programme(s)

FP7-PEOPLE - Specific programme "People" implementing the Seventh Framework Programme of the European Community for research, technological development and demonstration activities (2007 to 2013)

Topic(s)

FP7-PEOPLE-IOF-2008 - Marie Curie Action: "International Outgoing Fellowships for Career Development"

Call for proposal

FP7-PEOPLE-IOF-2008
See other projects for this call

Funding Scheme

MC-IOF - International Outgoing Fellowships (IOF)

Coordinator

UNIVERSITAT DE LES ILLES BALEARS

EU contribution

€ 217 113,92

Total cost

No data

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Region

Este > Illes Balears > Mallorca

Activity type

Higher or Secondary Education Establishments

Links

Contact the organisation Website
Participation in EU R&I programmes
HORIZON collaboration network

Last update: 1 August 2019

Permalink: https://cordis.europa.eu/project/id/237225