Poster H-41

Large-scale detection of mutations potentially responsible of genetic disorders

Authors:
Kais Ghedira (*Institute Pasteur of Tunis*)
Walid Bedhiafi (*Institut Pasteur de Tunis*)
Hela Ouaili (*Ecole de Supérieure des Statistique et de l'Analyse de l'Information*)
Koussay Dellagi (*Institute Pasteur of Tunis*)
Alia Benkahla (*Institute Pasteur of Tunis*)

Short Abstract: The scientific challenge of the present analysis is to extract meaningful and realistic hypotheses about links between mutations and human genetic disorders by taking advantage of all knowledge and data available in public databases.

Long Abstract:
Amino-acid positions that are conserved across vertebrates have been the target of selection during vertebrate evolution. Our hypothesis here is that substitutions at these positions are responsible for genetic disorders or other distinctive phenotypes. The scientific challenge of the present analysis is to extract meaningful and realistic hypotheses about links between mutations and human genetic disorders (for instance cancers, and cardiac diseases) by taking advantage of all knowledge and data available in public databases. The abundance and availability of three independent data quality suggested this analysis: (1) the synteny data and gene catalogs for five nearly completely sequenced vertebrate genomes (human, mouse, rat, chimpanzee, and dog) are available in the ENSEMBL database, and a number of other catalogs are available in UniGene; (2) the detailed descriptions of the tissue of origin of more then 7,000,000 human Expressed Sequence Tags (ESTs) are available in dbEST; (3) several datasets of human Single Nucleotide Polymorphisms (SNPs) are available for diverse disorders. We present here the strategy that was followed in order to develop a freely available database in which will be displayed: (i) positions conserved across all vertebrates; (ii) mutation in ESTs from disease tissues at theses positions (generated in i); (iii) SNPs from disease tissues at these positions (generated in i). We will also present the statistical tests that were used to evaluate the relevance of the resulting substitutions to biomedical research.