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Intelligent integrative knowledge bases: Bridging genomics, systems biology and personalized medicine

Hoan Nguyen

Institut de Genetique et de Biologie Moleculaire et Cellulaire(IGBMC), France

Abstract

Precision medicine relies on efficient genome annotation, 'omic' data integration and system level analyses to develop new approaches for personalized health care in which patients are treated based on their individual characteristics. We present a knowledgebase (http://decrypthon.igbmc.fr/sm2ph/cgi-bin/home), called SM2PH-Central (from Structural Mutation to Pathology Phenotypes in Human) which is part of an overall strategy aimed at the development of a transversal system to better understand and describe the networks of causality linking a particular phenotype, and one or various genes or networks. It incorporates tools and data related to all human genes, including their evolution, tissue expressions, genomic features and associated phenotypes, in a single infrastructure. It also provides access to systematic annotation tools, including sequence database searches, multiple alignment and 3D model exploitation, physico-chemical, functional, structural and evolutionary characterizations of variants. All information is accessible via standardized reports (gene profiles, error reports, etc.), as well as automated services for specific applications, such as gene prioritization (Gepetto framework). The structuration of the data and information in the SM2PH-Central facilitates the application of intelligent modules to search for hidden patterns and extract pertinent knowledge. For instance, we can cite MSV3d, devoted to the characterization of all missense variants and the KD4v system, which uses the 3D structures and information to characterize and predict the phenotypic effect of a mutation. A crucial feature of this infrastructure is the ability to create specialized knowledgebase, called SM2PH-Instances, which can be managed independently and allow the integration, management and distribution of domain-specific data (e.g. individual genomes/exomes). An Instance facilitates the construction of test sets to develop, compare and optimize methods to identify the genes and processes involved in the disease. The potential of this infrastructure has been demonstrated in a number of recent studies devoted to complete congenital stationary night blindness.

nguyen@igbmc.fr