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SM2PH-Central: An integrative knowledge base to investigate the genotype to phenotype relationships involved in human genetic diseases

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Abstract

The goal of SM2PH Central is to provide biologists and clinicians with easy access to various types of data linking genotype to phenotype via many standard web services, as well as numerous tools to characterize the structural and functional impact of missense mutations through the new KD4v predictio and MSV3d database associated .We have created the computational infrastructure associated with interactive analysis resources supporting in-depth studies and interpretation of the molecular consequences of mutations involved in only human monogenic disease. This included i) the integration of data and information concerning sequence, structure, mutation and OMIM. ii) the development of an automated cascade of programs including the creation and annotation of MSAs (Multiple Sequence Alignments), the creation of 3D models, the formalization and extraction of dedicated features and parameters. Recently, we have incorporated new tools (Gepetto-Gene Prioritization Extended Toolkit) and additional genomics and functional genomics data, in order to take into account molecular and cellular context in the analysis of mutation-to-phenotype relationships involved in all human protein (20219 proteins), including: (i) Introduction of all human SNPs defined in the dbSNP database and identification of the sequence variations affecting the protein sequence, (ii) Linking of genes involved in human diseases with functional genomics data: interactome, transcriptome and KEGG pathways, (iii) Enhanced quality of the 3D models and variant annotations, (iv) Identification of related proteins at the fold level using SCOP classification.

Biography

Hoan Nguyen (PhD) is a Researcher at the Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC), Illkrich, France where he works on projects related to automated variant annotation and prediction, database design and management and large-scale computation. His is responsible for SM2PH-Central framework.