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

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Abstract

Predictive 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 the SM2PH-Central (from Structural Mutation to Pathology Phenotypes in Human) knowledge base(1, 2) , http://decrypthon.igbmc.fr/sm2ph/cgi-bin/home, 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/proteins profiles, etc.), as well as automated services for specific applications, such as gene prioritization or variant prediction(3).

References

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Biography

Hoan Nguyen (PhD) is a Researcher at the Institut de Genetique et de Biologie Moleculaire 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**.**