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SM2PH-Central: Intelligent integrative system for comprehensive analysis of human diseases

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A nalysis of genetic variation is a major goal of biomedical research and disease studies, especially for the development of diagnostics and effective therapeutic solutions. Next generation sequencing technologies (NGS) are now used routinely to identify variations in individuals, families or populations. To achieve this, we developed a comprehensive knowledge discovery infrastructure SM2PH-CENTRAL for the analysis of genetic variations involved in human diseases, including an original "big data" management system (BIRD), a knowledge base of annotated variants (MSV3d), a prediction service (KD4v) to estimate their phenotypic effects, and an original open-source framework (GEPETTO) is for gene prioritization on a desktop computer that ensures confidentiality of personal data. The complete software and data environment is available for local or distributed computing. Case studies identified novel mutations in rare diseases (ciliopathies, retinopathies). This development 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. Our work helps elucidate the chain of events leading from a molecular defect to the associated pathology and thus represents an important step towards the practical application of translational medicine.

Biography

Hoan Nguyen is a big-data specialist at the Institute of Genetics and Molecular and Cellular Biology (IGBMC), France. He received a PhD degree in Applied Computer Science at the Strasbourg University, France in 2006. He subsequently joined the IGBMC where he works on projects related to automated annotation and prediction of genetic variants in human diseases, big-data design and large-scale computation. He is interested in the application of machine learning for the analysis of biomedical big-data and the extraction of biologically meaningful knowledge. He is responsible for SM2PH-Central framework. He is an author of 19 technical and scientific papers.

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