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PostGWAS Knowledge Miner- An integrative resource for cardiovascular Post-GWAS research

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Abstract

Genome-wide association studies (GWASs) have identified several variants robustly associated with various human diseases. Using large-scale meta-analyses of GWAS data, we recently increased to 46 the number of susceptibility loci to coronary artery disease (CAD) and myocardial infarction (MI). In addition, whole exome/genome sequencing have accelerated the discovery of gene mutations that are implicated in cardiovascular diseases (CVD) and related quantitative traits. These results provide a starting point for elucidating the molecular mechanisms of CVD.

Several initiatives have been launched to make GWAS results and genotype-phenotype data broadly accessible to the research community. However, there is an urgent need to develop an integrative resource that allows integration of GWAS results with different omics data. We conceived PostGWAS Knowledge Miner (PostGWAS.KM), the first integrative resource for cardiovascular Post-GWAS research. We integrated GWAS results, mutations identified by next-generation sequencing, biological knowledge, gene expression, eQTL data, SNP-miRNA associations, as well as information on relationship between genetic variants and drug targets.

PostGWAS.KM is a software package with three main components: 1) a knowledge base for cardiovascular research called CVDsKB, 2) a module for predicting the functional effect of SNPs, and 3) a module providing functionalities for SNP annotation and prioritization. SNP, mutations, and genes in CVDsKB are linked to public databases, including disease-centered databases, gene-centered databases, SNP databases, and online catalogs for GWAS results.

Although PostGWAS.KM is dedicated to cardiovascular research, other disciplines can benefits from its flexible architecture; only the content of CVDsKB database need to be updated.

Biography

After she obtained an Engineer Degree in computer sciences and a Master of Research in Medical Informatics and Communication Technologies, Seraya Maouche performed her Ph.D. at the University Pierre Marie Curie (Paris) and at the French National Institute of Medical research (INSERM). She was funded by the French Foundation for Medical Research. She performed her first postdoctoral fellowship at the University of Lübeck. She is co-leading a bioinformatics research group working on methods for large-scale genomics data. Seraya Maouche is co-authors of several articles published in highly ranked journals and she is finalizing a volume on bioinformatics for cardiovascular research.