Variobox: Automatic Detection and Annotation of Human Genetic Variants

Paulo Gaspar[1], Pedro Lopes[1], Jorge Oliveira[2], Rosário Santos[2], Raymond Dalgleish[3], José Luís Oliveira[1]

Genetic variations not only dictate phenotypic differences between human beings, but are also the underlying cause of many gene-based disorders. Detecting, understanding, categorizing and associating human mutations with phenotypes is becoming the standard process to accomplish personalized medicine. Multiple software and hardware technologies already spanned from miscellaneous projects, leveraging an exponential growth of available genetics data, and it is now fairly easy and cheap to obtain sequence profiles for large cohorts, such as the 1000 Genomes Project, or for unique individuals, such as the ones performed by several genetic analysis companies and labs. Moreover, the overwhelming quantity of genetic patient data emerging from labs, along with available LSDBs, suggests the need for integration solutions that are able to use variations' knowledge for gene research and patient care scenarios.

Variobox is a desktop tool for the annotation, analysis and comparison of human genes. Variobox obtains variant annotation data from WAVe, protein metadata annotations from PDB and UniProt, and sequence metadata is obtained from Locus Reference Genomic (LRG) and RefSeq databases. To explore the retrieved data, Variobox provides an advanced sequence visualization that permits an agile navigation through the various genetic regions, and combines its features in an intuitive interface to analyse genes and mutations. At last, genes can be compared to sequences retrieved from LRG and RefSeq, finding and automatically annotating new potential variations. Variobox is a free cross-platform desktop application, available for download at *http://bioinformatics.ua.pt/variobox*.

1 — Department of Eletronics Telecommunications and Informatics & IEETA, University of Aveiro
2 — CGM, INSA
3 — University of Leicester

variobox