TCAG New Technologies Seminar

Alamut: A Decision Support System for Mutation Interpretation

Date: Tuesday, Sept. 21
Time: 10:30-11:30 AM
Location: Room 14-203, MaRS TMDT
101 College St.

Speaker: Dr. André Blavier - MD, MSc
Interactive Biosoftware

Alamut is a software application dedicated to variant interpretation in medical genetics with four complementary functions, data integration from public data sources (i.e. sequences),conservation information, polymorphisms, published mutations, and protein annotations. Visualization of integrated data is represented in a user-friendly graphical environment, designed to apprehend information easily. Variant interpretation assisted by *in silico* predictions of pathogenicity and splicing effects. Data collection, storage and visualization of variants along with computed and manually-entered annotations. Basically, Alamut is a sophisticated viewer over thousands of annotated human genes, enabling users to enter variations and study their effects on transcription, splicing and translation. Interpretation is assisted by automated access to web-based variant scoring systems (PolyPhen, SIFT, Align GVGD) and by integrated splicing predictions methods (SpliceSiteFinder, MaxEntScan, NNSPLICE, GeneSplicer). Alamut displays a number of gene-related annotations, including nucleotide-level conservation, SNPs, published mutations collected by HGMD (Human Gene Mutation Database), protein domains, and multiple alignments of orthologues. As the software also generates mutation reports with full HGVS nomenclature, and manages user's own mutation sets, it puts together a very practical working environment for both diagnostic and research labs.

Hosted by The Centre for Applied Genomics and the Ontario Genomics Institute

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