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NGS Genomics Import Tool

The characterization of a patient, whether for clinical care or clinical research, is destined to become heavily dependent upon knowing an individual's full genome. The development of next generation sequencing (NGS) has allowed the whole genome to be readily expressed. This tool enables an i2b2 data loading process that transforms and organizes the products of NGS pipelines into genomic variant observations expressed in the Sequence Ontology. These observations can then co-exist with many other clinical observations about a patient and allow the formation of complex translational queries.

This project provides scripts to create the requisite Sequence Ontology metadata as well as software to transform NGS products into ready to load i2b2 observations.

Presentations

View Presentation Slides from 2013 i2b2 AUG Meeting, Aug 18, 2013, Boston, MA

Demonstration of sequence ontology Demonstration of a query for patients with an HLA-DQB1 protein log level < 0, with missense and nonsense variants on gene HLA-DQB1.

Software

Genomics import package for users Package containing plugin jar, Sequence Ontology metadata files, data files and User's guide for running the Genomics import tool within version 1.7 of the i2b2 workbench. This software converts output from VCF, ANNOVAR or GVF to I2B2 observation_fact format. Perl scripts are used to convert files to GVF; the import tool software converts GVF to I2B2 facts described by Sequence Ontology concepts and modifiers. Software source files can be downloaded from GitHub: https://github.com/i2b2plugins/wb-genomic-import-tool

The Perl scripts are additionally provided here for convenience: vcf2gvf.pl vcfANNO2gvf.pl

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Feb 28, 2018 • updated by Lori Phillips • view change

vcfANNO2gvf.pl
Dec 06, 2016 • attached by Lori Phillips

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Genomic NGS Tool
Mar 16, 2016 • updated by Janice Donahoe

GVF to I2B2
May 07, 2014 • updated by Lori Phillips • view change

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Future work
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Genome Variant Format

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Variant Call Format

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