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SNP Terminology development with VISTA

Designing a genomic variant ontology for common use within the open i2b2 community poses a unique challenge. Variants may be defined across several domains: dbSNP rs number, HGVS name, or gene/flanking sequence pairs. It is often difficult to know if variants from different domains are in fact equivalent. This project investigates ways to form a common basis for describing variants.

The project also examines necessary features required for display and comparison of genomic variants within external tools. An application has been developed to show interactive capability within the VISTA suite of visual analysis and functional annotation tools.

Paper and Presentations

Use of Genomic Variants in i2b2 - Lori C. Phillips MS1, Simon Minovitsky2, Igor Ratnere2, Inna Dubchak Ph.D.2,3, Isaac Kohane MD Ph.D.4, Shawn N. Murphy MD Ph.D.5 Partners Healthcare Systems, Charlestown, MA, 2DOE Joint Genome Institute, Walnut Creek, CA, 3Lawrence Berkeley National Laboratory, Berkeley, CA, 4Children's Hospital, Boston, MA, 5Massachusetts General Hospital, Boston, MA

View Presentation Slides from 2011 AMIA Summit Meeting on Translational Bioinformatics, San Francisco, CA, March 7-13, 2011

Software

Vista browser package for developers Package containing source code, data and developer's guide.

Vista browser package for users Package containing plugin jar, data files and User's guide for running the VISTA browser within version 1.6 of the i2b2 workbench.