Sequence Ontology

The Sequence Ontology for i2b2

A subset of the Sequence Ontology (http://www.sequenceontology.org) describing variants was repurposed for this project. The ontology organizes a set of concepts describing the structural change of the variant, such as a SNP/SNV (Single nucleotide polymorphism/Single nucleotide variant), or the insertion or the deletion of bases. Attributes of the variant are expressed as "modifiers" for that variant. In their simplest form shown below they may represent information relating to the location of the variant: start and end location of the variant within a chromosome; the gene in which the variation is thought to occur; or the type of variant (exonic, intronic, intergenic). The tree shown here appears on the http;//www.i2b2.org/webclient demo site.