

An Enhanced Sequence Ontology for i2b2

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Many tools exist to annotate NGS output vcf data files. With these annotations come opportunities to represent even more modifiers on the Navigation Tree. For example we can expand the tree to add exon-centric features such as dbSNP numbers, non/synonymous variant classification, PolyPhen scores, and functional attributes such as stop_loss or stop_gain. In essence, you can grow or shrink the ontology (and the observation fact data collected) as your system requirements dictate.

