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# **Software and Datasets**

We develop and maintain a variety of software, genomic annotations and web services.

#### **Annotation Datasets**

- Roulette is a mutation rate model identifying the mutagenic effect of Polymerase III transcription at the basepair resolution.
- $s_{het}$  are gene-based estimates of selection strength.

#### **Tools for Assessment of Variants**

- PolyPhen-2 is a tool for predicting the effect of an amino acid substitution on protein structure and function, based on comparative genomics and experimentally determined protein structures. It is available as a web service, and can also be downloaded as a standalone application.
- SNPtrack is a simple interface for mutation mapping and identifying causal mutations from wholegenome sequencing studies. It is available as a web service.

#### **Tools for Mass Spectrometry and Proteomics**

• MS-BLAST is a tool for searching protein sequences identified with tandem mass spectrometry against databases of protein sequences. It is available as a web service and as a standalone software.

#### **Tools for Statistical Genetics**

- Joint Likelihood Mapping (JLIM) is a tool to test for shared genetic effect between two genetic association data, for example, a disease GWAS study and gene expression QTL (eQTL) study.
- Joint Likelihood Mapping 2 (JLIM\_2.0) is a version of JLIM which supports meta-analysis across more than one cohort of matching ancestry.
- Joint Likelihood Mapping (JLIM) 2.5 is a new version of JLIM based on summary statistics.

- NPS is a tool for polygenic risk scoring based on partitioning-based non-parametric shrinkage algorithm.
- RVTT is a novel statistical test of trend that assesses the relationship of the frequency of qualifying rare variants in a pathway with dichotomous disease phenotypes leveraging the Cochran-Armitage test statistic.

### **Tools for Cancer Genomics**

- MutPanning is designed to detect rare cancer driver genes from aggregated whole-exome sequencing data.
- CBaSE enables cancer type and gene-specific estimation of the strength of negative and positive selection. It is available as a browser-based tool as well as for download as a standalone package.

## **Tools for Population Genetics**

• simDoSe is a fast and flexible Wright-Fisher simulator for arbitrary diploid selection evolving through realistic human demography.

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