

Input
VCF file

**Annotate LoF mutations
with variant- and transcript-specific features**

Mismapping

Segmental duplication;
pseudogene; paralog

Functional

NMD prediction; Loss of functional, structural domains, disordered regions, post translational modification sites; gene expression in GTex...

Annotation Issue

Non-canonical splice site;
LoF position...

Conservation

GERP score; dN/dS; 1000G, ESP6500 allele frequency; heterozygosity of genes...

Network

Shortest path to disease genes; network centralities...

Pathogenicity of premature Stop mutations

Prediction model

trained on benign, dominant and recessive disease-causing premature-stop mutations

Output