Web Tools
- Web Tools
  - BLAST/BLAT
  - Variant Effect Predictor
  - Linkage Disequilibrium Calculato
  - File Chameleon
  - Assembly Converter
  - ID History Converter
  - VCF to PED Converter
  - Data Slicer

Configure this page
Custom tracks
Export data
Share this page
Bookmark this page

Variant Effect Predictor

New job | Clear form | Close

Species: Human (Homo sapiens)

Assembly: GRCh38.p12 *(If you are looking for VEP for Human GRCh37, please go to GRCh37 website.)*

Name for this job (optional):

Input data:

Either paste data:

Examples: Ensembl default, VCF, Variant identifiers, HGVS notations, SPDI

Or upload file: Выберите файл Файл не выбран

Click here to download the previously uploaded file.

Or provide file URL:

Transcript database to use:

- (•) Ensembl/GENCODE transcripts
- ( ) Ensembl/GENCODE basic transcripts
- ( ) RefSeq transcripts
- ( ) Ensembl/GENCODE and RefSeq transcripts

Additional configurations:

Identifiers — *Additional identifiers for genes, transcripts and variants*

Identifiers

- [x] Gene symbol:
- [x] Transcript version:
- [ ] CCDS:
- [ ] Protein:
- [ ] UniProt:
- [ ] HGVS:

Variants and frequency data — *Co-located variants and frequency data*

Variants and frequency data

- Find co-located known variants: Yes
- Frequency data for co-located variants:
  - [x] 1000 Genomes global minor allele frequency
  - [x] 1000 Genomes continental allele frequencies
  - [x] ESP allele frequencies
  - [x] gnomAD (exomes) allele frequencies
- [x] PubMed IDs for citations of co-located variants:
- [ ] Include flagged variants:

Additional annotations — *Addtional transcript, protein and regulatory annotations*

Transcript annotation

- [x] Transcript biotype:
- [ ] Exon and intron numbers:
- [x] Transcript support level:
- [x] APPRIS:
- [ ] Identify canonical transcripts:
- Upstream/Downstream distance (bp): 5000
- [ ] miRNA structure:

Protein annotation

- [ ] Protein domains:

Regulatory data

- Get regulatory region consequences: Yes

Phenotype data

- [ ] Phenotypes:

Predictions — *Variant predictions, e.g. SIFT, PolyPhen*

Pathogenicity predictions

- SIFT: Prediction and score
- PolyPhen: Prediction and score
- dbNSFP:
  - (•) Disabled
  - ( ) Enabled

CADD:

Condel:
- Disabled
- Enabled

LoFtool:

Splicing predictions

dbscSNV:

MaxEntScan:

Conservation

BLOSUM62:

Ancestral allele:

Filtering options *Pre-filter results by frequency or consequence type*

Filters

Filter by frequency:
- No filtering
- Exclude common variants
- Advanced filtering

Return results for variants in coding regions only:

Restrict results: Show all results

NB: Restricting results may exclude biologically important data!

Advanced options *Settings to optimise VEP*

Advanced options

Buffer size: 5000

NB: When the **Regulatory data** option is selected, due to the large amount of regulatory data available, the **maximum** buffer size is automatically set to **500**. However you can still select a value lower than 500.

Run ›

Recent jobs

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Analysis	Jobs	Submitted at
Variant Effect Predictor	VEP analysis of forVEP.vcf in Homo_sapiens Done [View results]	23/07/2019, 10:21 (BST)
Variant Effect Predictor	VEP analysis of forVEP.vcf in Homo_sapiens Done [View results]	17/07/2019, 10:44 (BST)