Precision medicine: NGS variant analysis and interpretation for translational research

NGSII: Variant annotation

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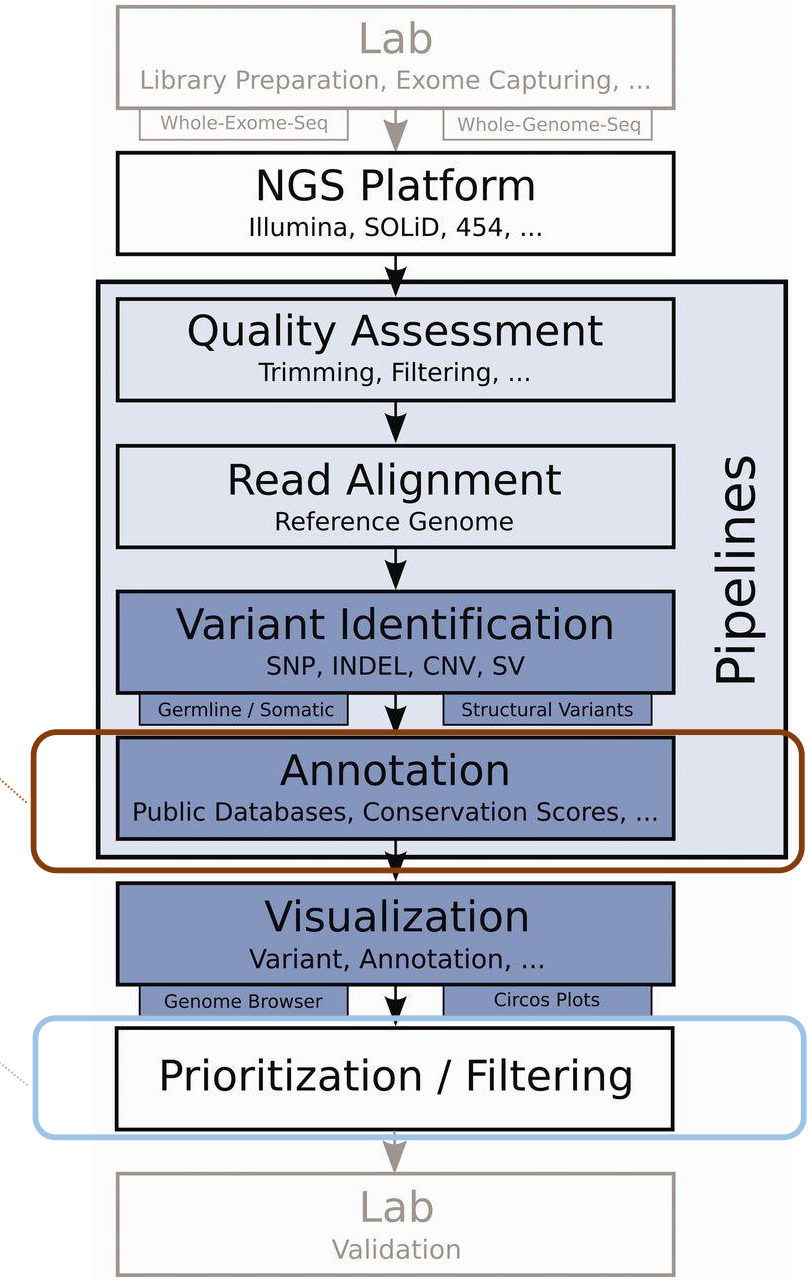
INSTITUTO GULBENKIAN DE CIÊNCIA

NGSII: Variant Annotation

Selecting the most relevant variants: How to filter



Stephan Pabinger et al. Brief Bioinform 2013;bib.bbs086

Variant annotation

- Technical information: quality parameters, filters, …
- Descriptive information: nomenclature, genotype, …
- Functional annotation: consequence, functional prediction, ...

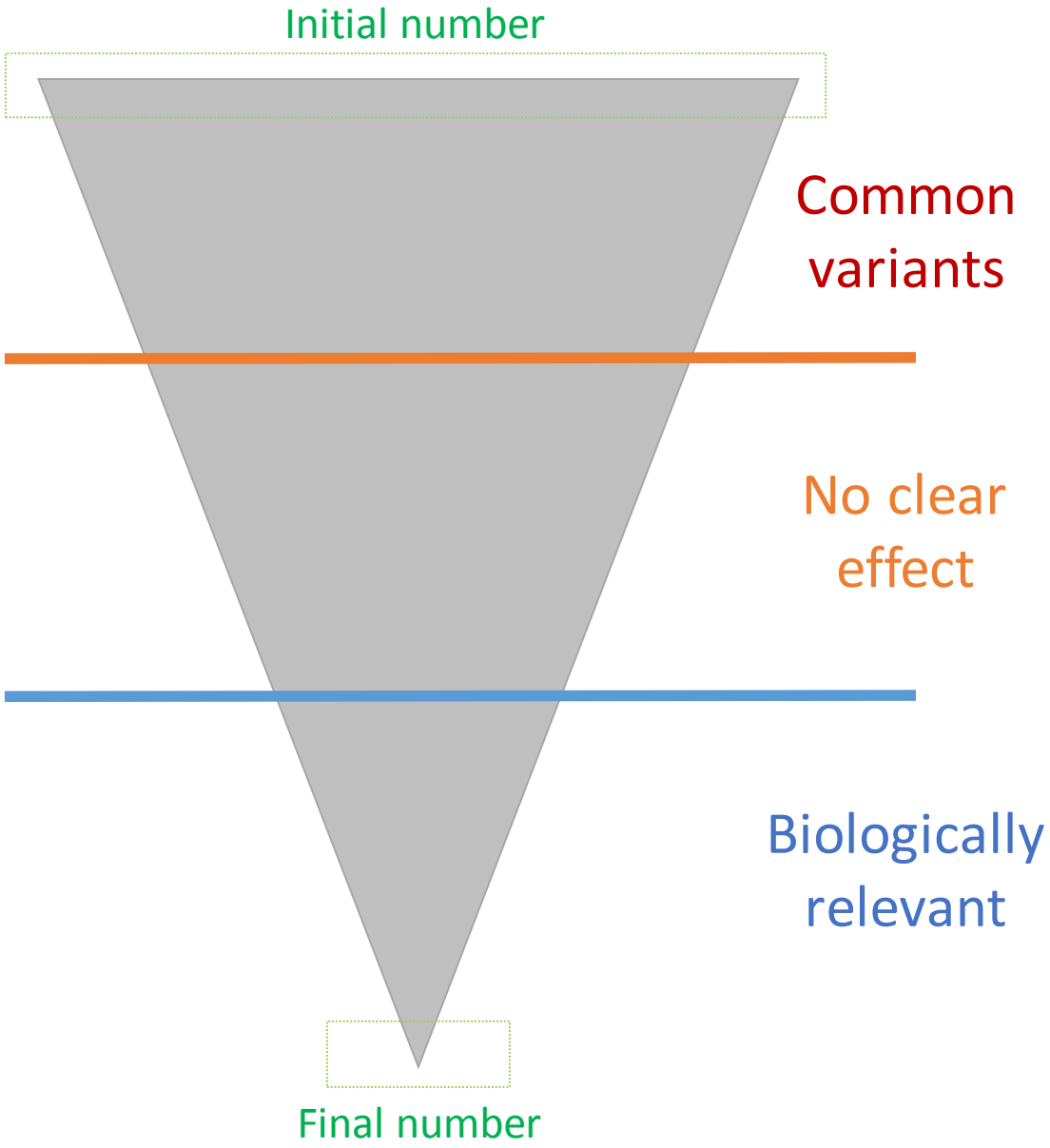
Variant callers provide technical parameters associated to each variant, that allow us to remove sequencing artifacts and select the most reliable variants.

Variant annotation

Not all the variants have a relevant impact in the phenotype of study. They are mainly polymorphisms (maybe predisposition) or variants without a clear effect.

To identify and select the most relevant variants we need to add descriptive and functional annotations.

There are different software programs that perform variant annotation.



Name	Input Format	Output Format	SNP	INDEL	CNV	GUI	CLI	Web	Notes
ABSOLUTE [91]	HAPSEG output, sample level variance, precomputed models of cancer types, sigma values	Plot showing the Purity/Ploidy, R data file	yes	no	yes	no	yes	no	Comes bundles with HAPSEG;
Align-GVGD [92]	FASTA, substitutions list	Web report	yes	no	no	no	no	yes	Estimates SNP risk;
ANNOVAR [93]	VCF4, Complete Genomics, GFF3-SOLiD, CSV in Annovar format;	Gene-based annotation; Region-based annotations; Filter-based annotation. For all categories	yes	yes	yes	no	yes	no	Integrated tool providing gene annotation, db ids and various scores;
AnnTools [94]	VCF, pileup, CSV	VCF	yes	yes	yes	no	yes	no	Provides a set of helper tools for custom annotation;
Auto-mute [95]	PDB ID, Chain, Mutation	Web report	yes	no	no	no	no	yes	The tool performs stability and disease potential predictions.
CandiSNPer [96]	dbSNP ID, population	Web report	yes	no	no	no	no	yes	
CHASM and SNVBox [97]	Passenger mutation rates, AA changes	CSV including CHASM score, p-value, and FDR	yes	no	no	no	yes	no	Predicts the functional significance of somatic missense mutations observed in the genomes of cancer cells and features prioritization of mutations;
CUPSAT [98]	PDB ID; PDB file format	Web report	yes	no	no	no	no	yes	Performs protein stability prediction;
dbNSFP [99]			yes	no	no	no	yes	no	Integrated SNP database; provides a simple JAVA CLI tool for searching;
VEP (Ensembl - Variant Effect Predictor) [100]	CSV, VCF, Pileup, HGVS, Variant Identifiers	Web report	yes	no	no	no	yes	yes	
ESEfinder [101]	FASTA	Web report, CSV	-	-	-	no	no	yes	Analyzes sequences for the presence of ESE motifs;
ESRSearch [102]	plain sequence; FASTA	Web report	-	-	-	no	no	yes	Finds ESR sequences;
FANS [103]	FASTA format; or variation information via web interface	Web report, CSV	yes	no	no	no	no	yes	Prioritized variations based on risk levels; divided into: Genome View, Gene View, Transcript View, Variation View;
FastSNP [104]	Gene Symbol, dbSNP ID	Web report	yes	no	no	no	no	yes	Outputs prioritized list of SNPs with risk assessment;
FESD [105]	Gene name	Web report	yes	no	no	no	no	yes	Output includes regions: promoter, CpG, islands, translation start, splice site, translation stop, poly(A) signal, transcript;

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Name	Input Format	Output Format	SNP	INDEL	CNV	GUI	CLI	Web	Notes
FOLD-X [106]			yes	no	no	no	yes	yes	It performs protein stability analysis.
F-SNP [107]	SNP ID; disease; gene; chromosomal region		yes	no	no				The software integrates information obtained from 16 bioinformatics tools and databases about the functional effects of SNPs.
GERP++ [108]		Web report	yes	no	no	no	yes	yes	It produces evolutionary conservation scores.
GSITIC [109]	Segmentation File, Markers File, FASTA, (Array List File, CNV File)	Lesions, Amplification Genes, Deletion Genes, Gistic Scores, Plots	no	no	yes	no	yes	no	Identifies regions of the genome that are significantly amplified or deleted across a set of samples;
HOPE [110]	FASTA, accession code for protein	Web report on structural differences between wild type and mutations	yes	no	no	no	no	yes	The web-based tool offers a simple web interface for entering protein sequence and amino acid mutation.
Human Splicing Finder (HSF) [111]	Ensembl / RefSeq ID, plain text sequences		yes	no	no	no	no	yes	
I-Mutant2.0 [112]	One letter residue code, sequence residue number	Web report	yes	no	no	no	yes	yes	The tool is based on support vector machines.
LS-SNP [113]	SwissProt ID, dbSNP ID, Kegg Pathway ID, HUGO Gene ID	Web report	yes	no	no	no	no	yes	The tool offers prediction of disease association and confidence of prediction and is based on support vector machines (SVM).
MAPP [114]	FASTA	CSV in MAPP format	yes	no	no	no	yes	no	
MuD [115]		Web report	yes	no	no	no	no	yes	
MutaGeneSys [116]		Web report / CSV	yes	no	no	no	yes	yes	The query Interface is not working.
MutationAssessor [117]	CSV in MutationAssessor format, Uniprot ID, Refseq ID	CSV in MutationAssessor format	yes	no	no	no	no	yes	
MutationTaster [118]	ORF, cDNA sequence, genomic sequence, alteration	Web report	yes	yes	no	no	yes	yes	
MutPred [119]	FASTA sequence, CSV file of mutations	Web report	yes	no	no	no	no	yes	Calculates the impact of mutation on different protein properties; is based on SIFT and offers precomputed dbSNP results;
MutSig [120]	List of mutations, regions to investigate	CSV	yes	yes	no	no	yes	no	Still in beta testing – available upon request
NGS-SNP [121]	VCF, pileup, CSV	VCF	yes	no	no	no	yes	no	
nsSNPAnalyzer [122]	FASTA, substitutions list	Web report	yes	no	no	no	no	yes	The tool outputs various SNP features and predicts the phenotypic class
Oncotator [123]	Oncotator format	CSV	yes	yes	no	no	no	yes	Annotations with data relevant to cancer researcher; collects Genomic Annotations, Protein Annotations, Cancer Annotations

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Name	Input Format	Output Format	SNP	INDEL	CNV	GUI	CLI	Web	Notes
PANTHER [124]	Protein sequence and Substitution	subPSEC score	yes	no	no	no	yes	yes	Uses subPSEC score;
Parepro [125]	Protein sequence and Substitution	-	yes	no	no	no	yes	no	It is based on support vector machines (SVM).
PESX [126]	plain sequence; FASTA	Web report	-	-	-	no	no	yes	Finds ESE sequences;
pfSNP [127]	SNP ID; chromosome region; Gene ID;	Web report	yes	no	no	no	no	yes	
PHAST [128]	FASTA, PHYLIP, MPM, MAF, SS	Conservation score	-	-	-	no	yes	no	Phylogenetic analysis toolbox, including phastCons and phyloP;
PhD-SNP [129]	One letter residue code, Swiss-Prot protein code, Sequence file	Effect preditction	yes	no	no	no	yes	no	
PMUT [130]	FASTA sequence/file SWISSProt code	Web report	yes	no	no	no	no	yes	Offers different prediction modes and is able to output detailed mutation analysis reports;
PolyDoms [131]	Gene/protein symbol(s), RefSeqID dbSNP ID	Web report	yes	no	no	no	no	yes	
PolyMAPr [132]	-	-	-	-	-	no	yes	no	No longer available;
PolyPhen-2 [133]	UniProt ID, FASTA, dbSNP ID	CSV in PolyPhen format	yes	no	no	no	yes	yes	
PupaSNP Finder [134]	dbSNP ID, Gene/Transcript ID; PED format	Web report	yes	no	no	no	no	yes	
QuickSNP [135]	genomic position; HUGO gene symbol	Web report	yes	no	no	no	no	yes	
RescueESE [136]	plain text; multi-FASTA	predicts sequences with ESE activity	-	-	-	no	no	yes	
SAPRED [137]	FASTA and mutation file		yes	no	no				The website is offline.
SCAN [138]		Web report	yes	no	yes	no	no	yes	
SCONE [139]	MAF	Conservation score	-	-	-	no	yes	no	
SeattleSeq Annotation [140]	Maq, GFFm CASAVA, VCF, GATK bed	VCF, own format	yes	yes	no	no	no	yes	
SeqAnt [141]	FASTA sequence file	Web report	yes	yes	no	no	no	yes	
SeqProfCod [142]	-	-	yes	no	no	-	-	-	Not available online;
SVA (Sequence Variant Analyser) [143]	VCF of variants, project file (for command line version)	--potential biological function --dbSNP/Kegg/GO/1000 Genomes/DGV annotation --identifies protein-truncating variants --filtering by function	yes	yes	no	yes	yes	no	

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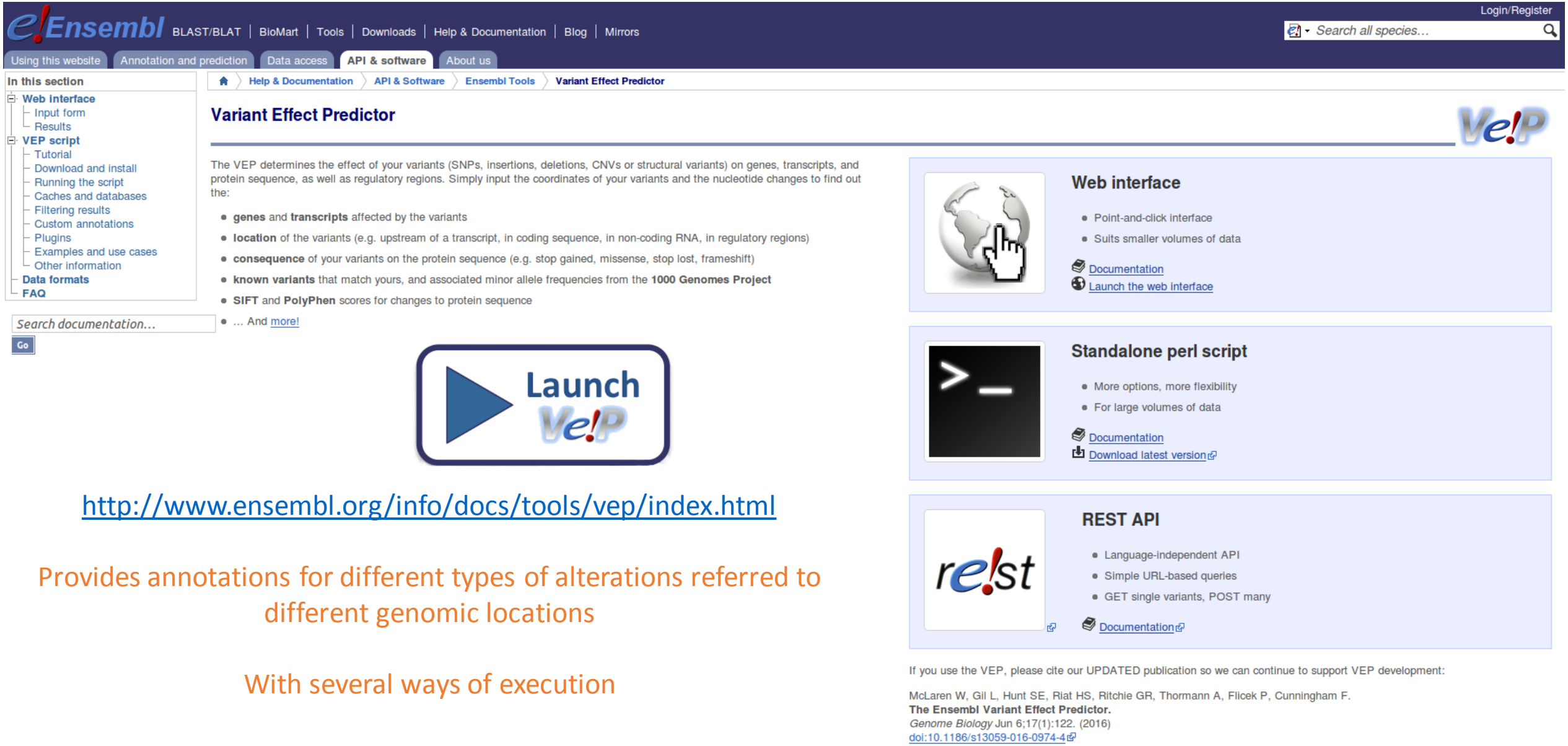
Name	Input Format	Output Format	SNP	INDEL	CNV	GUI	CLI	Web	Notes
SIFT [144]	Multiple proteins, dbSNP ID, NCBI GI number, protein sequence, protein sequence alignment, Pileup, VCF4, maq, soap, gff3, casava, cg	XXX in SIFT format	yes	no	no	no	yes	yes	
SIFT Indel [145]			no	yes	no	no	no	yes	
SiPhy [146]	FASTA, MAF, PHYLIP		-	-	-	no	yes	no	
SNAP [147]	AA in FASTA, substitutions format	Web report	yes	no	no	no	no	yes	This tool offers a user friendly web interface.
SNP Function Portal [148]	RefSNP Ids, OMIM Ids	Web report	yes	no	no	no	no	yes	
SNP@Domain [149]			yes	no	no	-	-	-	Not available anymore;
SNPdbe [150]	Gene/protein symbol, FASTA	Web report	yes	no	no	no	no	yes	The protein function is predicted using SNAP and SIFT and entries are augmented with experimental information from public databases.
SNPeffect 4.0 [151]	FASTA, PDB file, PDB ID, UniProt ID	Web report	yes	no	no	no	no	yes	This tool mainly uses protein structure information.
SNPHunter [152]	Gene symbol; dbSNP ID;	Web report	yes	no	no	yes	no	no	
SNPnexus [153]	CSV in SNPnexus input format	CSV in SNPnexus output format	yes	yes	yes	no	no	yes	Outputs CNV, INDELs, inversions;
SNPper [154]	dbSNP ID, TSC ID, position	Web report	yes	no	no	no	no	yes	
SNPs&GO [155]	One letter residue code; Swiss-Prot protein code; Sequence file; GO terms; CSV	Web report	yes	no	no	no	no	yes	Predicts neutral/deleterious; calculates reliability index and disease probability;
SNPs3D [156]	Gene symbol, SNP ID	Web report	yes	no	no	no	no	yes	
SNPseek [157]	-	-	-	-	-	-	-	-	Tool that performs neural network based protein stability prediction which is not available anymore;
SNPselector [158]	-	-	-	-	-	no	no	yes	No longer available;
SnpSIFT + snpEff [159]	VCF, SNPs, insertions, deletions, and MNPs	CSV	yes	yes	no	no	yes	no	A collection of tools to manipulate VCF files;
SPOT [160]	SNPs and p-values,	Web report	yes	no	no	no	no	yes	Outputs various DB ids and scores;
StSNP [161]	protein sequence; protein name; dbSNP ID; gene symbol	Web report	yes	no	no	no	no	yes	
TAMAL [162]	-	-	-	-	-	-	-	-	No longer available;
TopoSNP [163]	Protein ID, protein sequence	Web report	yes	no	no	no	no	yes	Predicts whether substitution is on surface of the protein structure; conservation score based on Pfam

Variant Effect Predictor (VEP)



http://www.ensembl.org/info/docs/tools/vep/index.html

Provides annotations for different types of alterations referred to different genomic locations

With several ways of execution

Standalone execution

perl **variant_effect_predictor.pl** --format vcf --sift b --polyphen b --ccds --uniprot --hgvs --symbol --numbers --domains --regulatory --canonical --protein --biotype --uniprot --tsl --gmaf --variant_class --xref_refseq --maf_1kg --maf_esp --maf_exac --dir /home/epineiro/analysis/pancancer/vep/ensembl-tools-release-85/scripts/variant_effect_predictor /.vep -i /home/epineiro/analysis/pancancer/genotypes/0a6be23a-d5a0-4e95-ada2-a61b2b5d9485.vcf --config /home/epineiro/analysis/pancancer/vep/ensembl-tools-release-85/scripts/variant_effect_predictor/registry.local --output_file / /home/epineiro/analysis/pancancer/genotypes/0a6be23a-d5a0-4e95-ada2-a61b2b5d9485.vcf_output_VEP.txt --force_overwrite --vcf --no_progress --plugin Condel,/home/epineiro/analysis/pancancer/vep/ensembl-tools-release-85/scripts/variant_effect_predictor/.vep/Plugins/config/Condel/config,b --fork 8 --offline

Configuration options

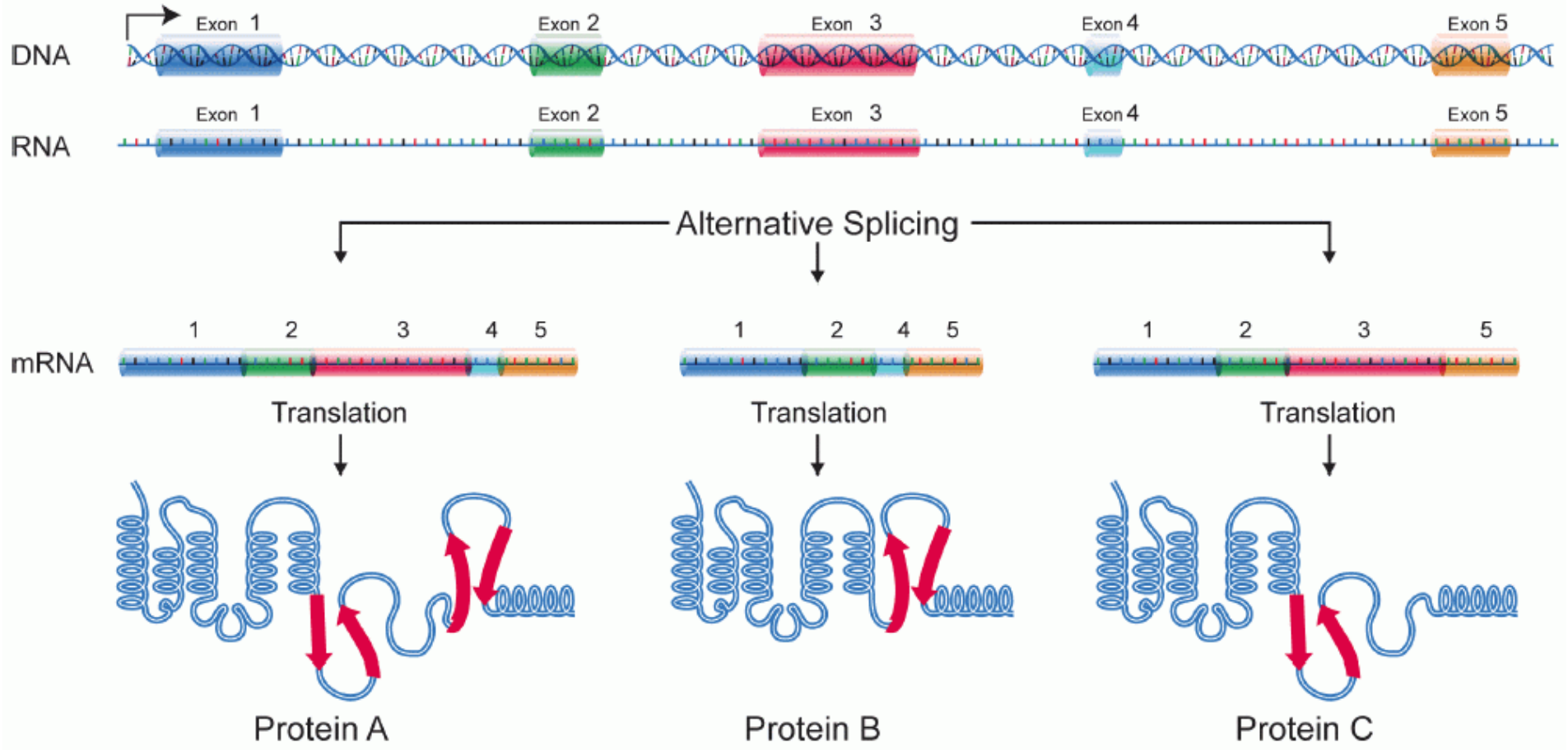
Input, output and format

Annotations

Plugins

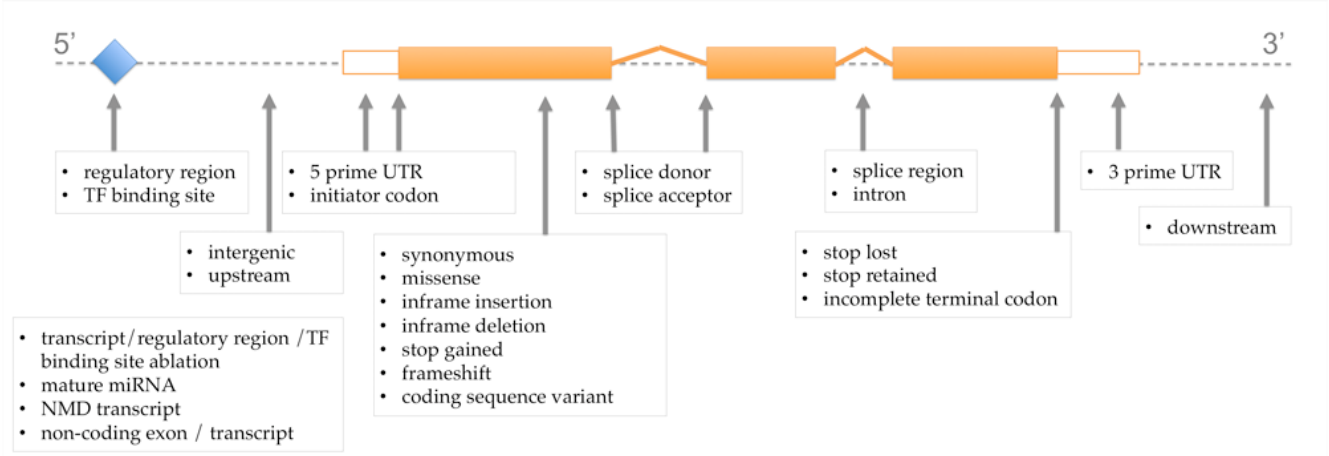
http://www.ensembl.org/info/docs/tools/vep/script/vep_options.html#basic

Annotations provided for each affected transcript



Annotations about consequence: Transcriptional

Sequence Ontology



*	SO term	SO description	SO accession	Display term	IMPACT
	transcript_ablation	A feature ablation whereby the deleted region includes a transcript feature	SO:0001893	Transcript ablation	HIGH
	splice_acceptor_variant	A splice variant that changes the 2 base region at the 3' end of an intron	SO:0001574	Splice acceptor variant	HIGH
	splice_donor_variant	A splice variant that changes the 2 base region at the 5' end of an intron	SO:0001575	Splice donor variant	HIGH
	stop_gained	A sequence variant whereby at least one base of a codon is changed, resulting in a premature stop codon, leading to a shortened transcript	SO:0001587	Stop gained	HIGH
	frameshift_variant	A sequence variant which causes a disruption of the translational reading frame, because the number of nucleotides inserted or deleted is not a multiple of three	SO:0001589	Frameshift variant	HIGH
	stop_lost	A sequence variant where at least one base of the terminator codon (stop) is changed, resulting in an elongated transcript	SO:0001578	Stop lost	HIGH
	start_lost	A codon variant that changes at least one base of the canonical start codo	SO:0002012	Start lost	HIGH
	transcript_amplification	A feature amplification of a region containing a transcript	SO:0001889	Transcript amplification	HIGH
	inframe_insertion	An inframe non synonymous variant that inserts bases into in the coding sequenc	SO:0001821	Inframe insertion	MODERATE
	inframe_deletion	An inframe non synonymous variant that deletes bases from the coding sequenc	SO:0001822	Inframe deletion	MODERATE
	missense_variant	A sequence variant, that changes one or more bases, resulting in a different amino acid sequence but where the length is preserved	SO:0001583	Missense variant	MODERATE
	protein_altering_variant	A sequence_variant which is predicted to change the protein encoded in the coding sequence	SO:0001818	Protein altering variant	MODERATE

HIGH

MODERATE

High

Moderate

Low

Modifier

Annotations about consequence: Functional impact prediction

SIFT PREDICTION (--sift [p|s|b]) predicts whether an amino acid substitution affects protein function based on sequence homology and the physical properties of amino acids. The VEP can output the **p**rediction term, **s**core or **b**oth

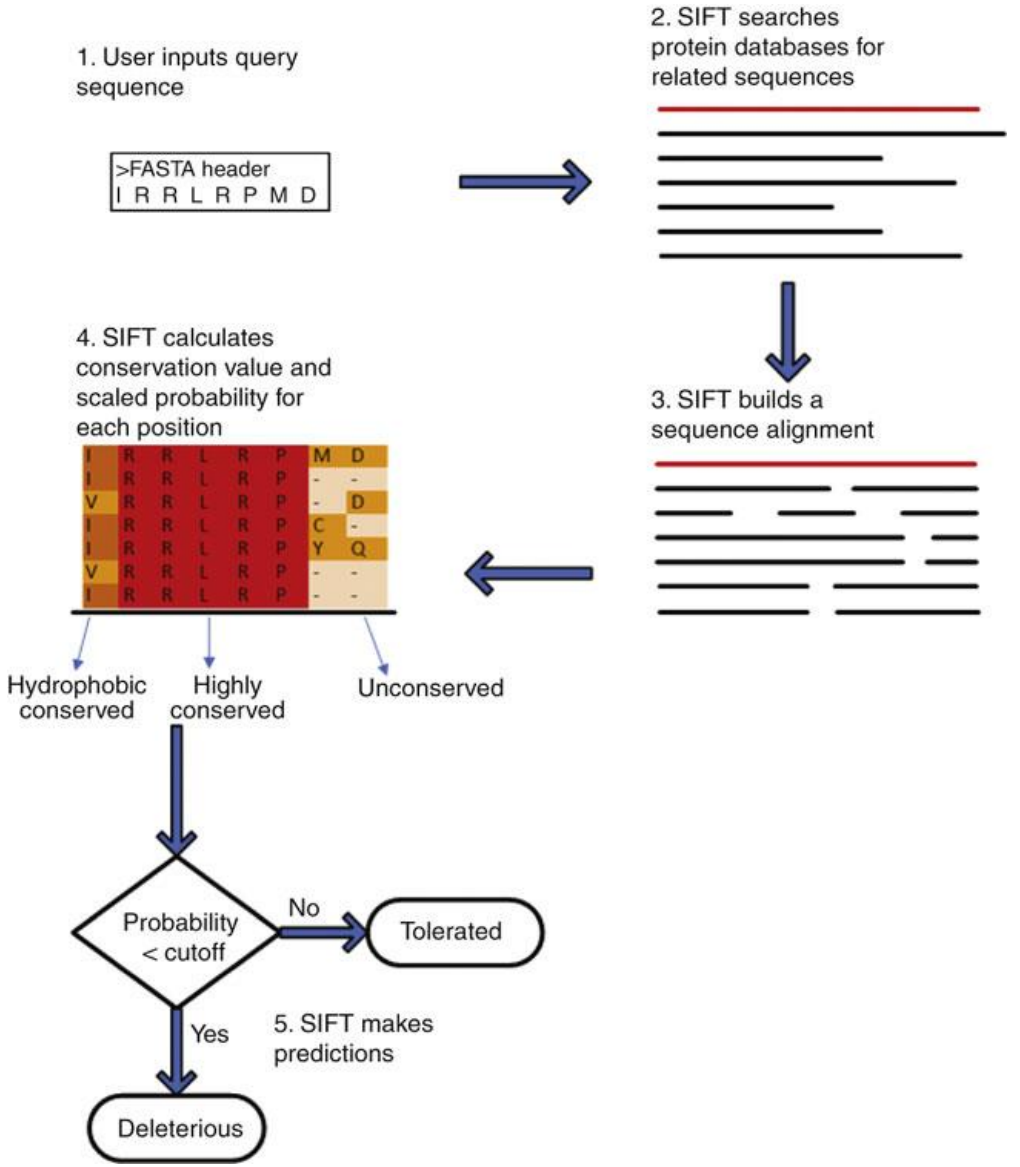
Deleterious – Tolerated

< 0.05 Deleterious

PolyPhen PREDICTION (--polyphen [p|s|b]) predicts the possible impact of an amino acid substitution on the structure and function of a human protein using straightforward physical and comparative considerations. The VEP can output the **p**rediction term, **s**core or **b**oth.

Probably damaging - Possibly damaging - Benign – Unknown

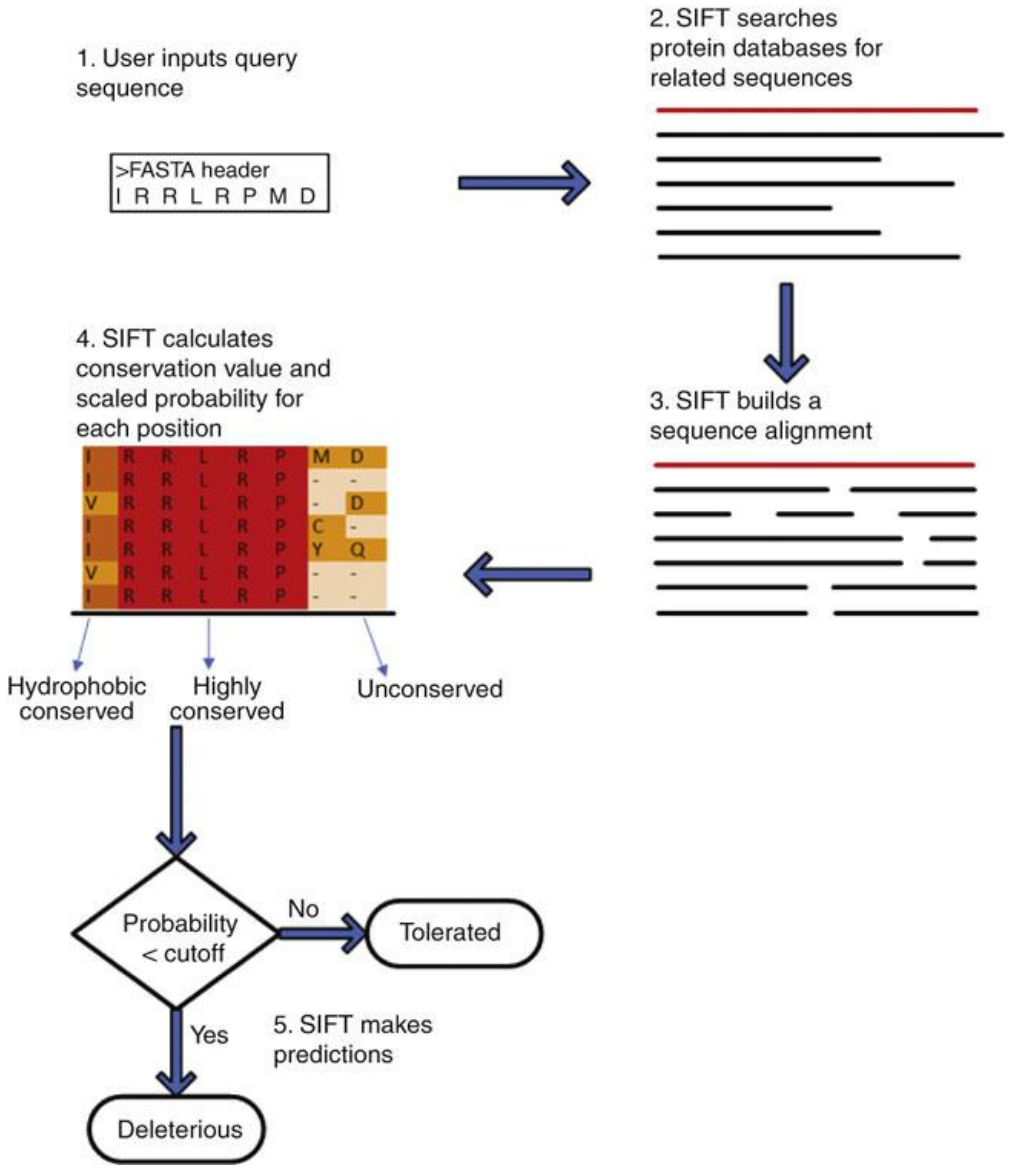
Benign < ~0.435 < Damaging



Annotations about consequence: Functional impact prediction

The built-in functionality in VEP of these predictors allows only the prediction of coding non-synonymous SNV

CONDEL (--plugin) Condel **CONsensus DELeteriousness score** runs an adapted version of CONDEL computing a score based on pre-calculated SIFT and PolyPhen-2 scores from the Ensembl API.



Impact prediction – Other predictors

- **dbNSFP**

functional predictions and annotations for human nonsynonymous single-nucleotide variants and splice-site variants

- **LoFtool**

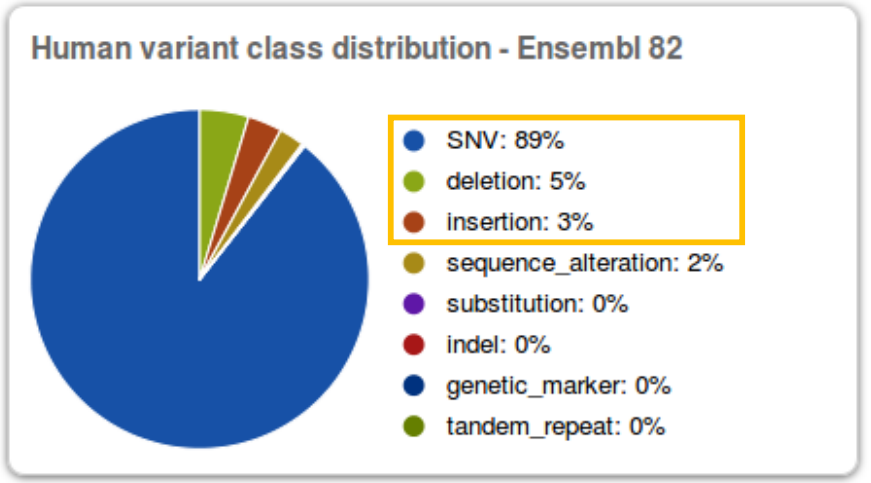
gene intolerance ranking system, based on the ratio of Loss-of-function (LoF) to synonymous mutations for each gene from ExAC data

- **PROVEAN**

Functional prediction for nonsynonymous mutations or indels

Variant level annotations

VARIANT CLASS (--variant_class): Sequence Ontology variant class

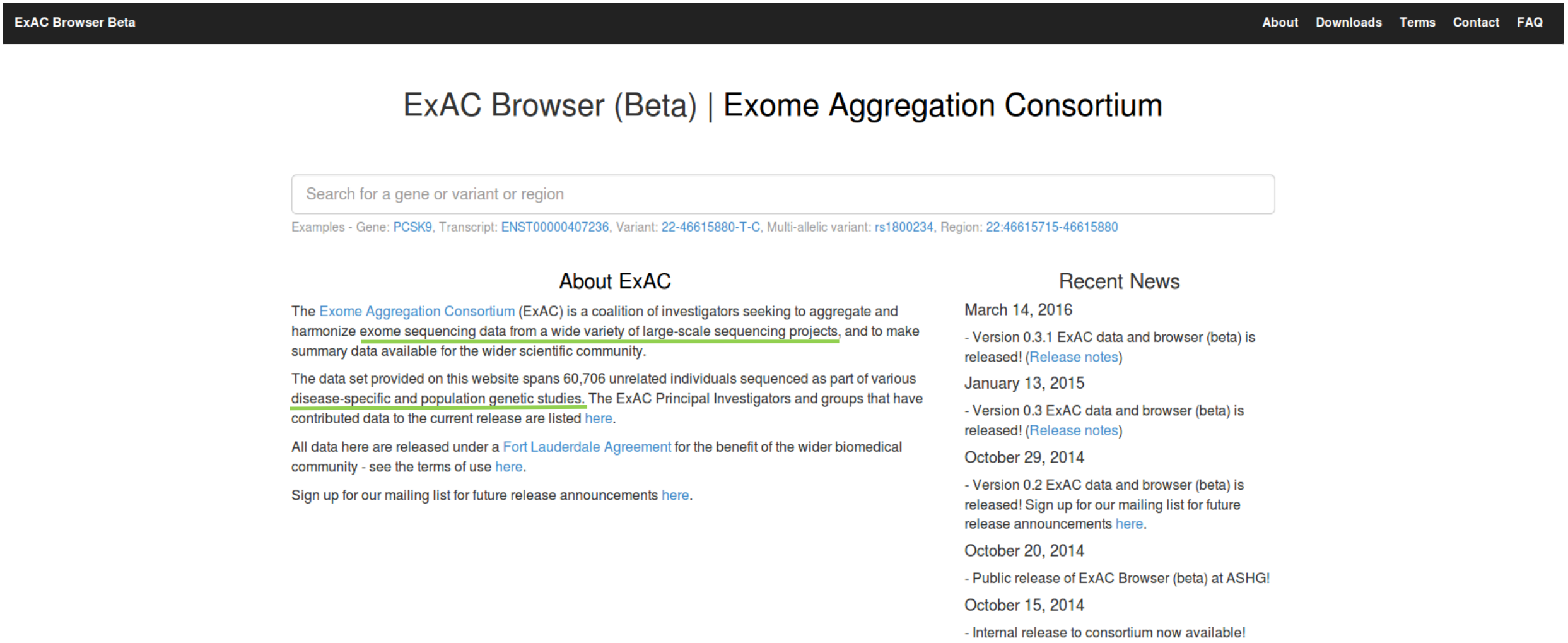


HGVS NOMENCLATURE (--hgvs): HGVS nomenclature additional to Ensembl identifier. Both coding and protein sequence. e.g. ENST00000263967.3:c.3140A>G

GMAF (--gmaf): *Global minor allele frequency* (MAF) from 1000 Genomes Phase 1. e.g. G:0.4036

ExAC (--maf_exac)

http://exac.broadinstitute.org/



ExAC (--maf_exac)

Variant: 22:46615880 T / C

Note: This variant is multiallelic! The other alt alleles are:

- 22-46615880-T-A

Filter Status	PASS
dbSNP	rs1800234
Allele Frequency	0.009613
Allele Count	1163 / 120986
UCSC	22-46615880-T-C
ClinVar	Click to search for variant in Clinvar

Genotype Quality Metrics

Site Quality Metrics

Annotations

This variant falls on 7 transcripts in 1 genes:

missense

- PPARA Transcripts

non coding transcript exon

- PPARA - ENST00000493286

intron

- PPARA - ENST00000434345

Note: This list may not include additional transcripts in the same gene that the variant does not overlap.

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
Latino	649	11522	28	0.05633
East Asian	361	8618	8	0.04189
Other	10	904	0	0.01106
European (Finnish)	22	6606	1	0.00333
South Asian	42	16440	2	0.002555
European (Non-Finnish)	73	66602	0	0.001096
African	6	10294	0	0.0005829
Total	**1163**	**120986**	**39**	**0.009613**

Gene and genetic region

GENE SYMBOL (--symbol) e.g. MYC

Standard nomenclature for the human genes

EXON/INTRON NUMBER (--numbers) The format is Number/Total.

OVERLAPPING REGULATORY REGIONS (--regulatory) Script also detect if the variant falls in a **transcription factor binding site**. Output lines have a Feature type of **RegulatoryFeature** or **MotifFeature**.

BIOTYPE (--biotype) Adds the biotype of the transcript or regulatory feature. e.g. protein_coding, processed_pseudogene...

Transcript annotation

CCDS (--ccds) Adds the CCDS transcript identifier. e.g. CCDS1639.1

CCDS: Consensual CoDing Sequence

TRANSCRIPT SUPPORT LEVEL (--tsl)

The Transcript Support Level (TSL) indicates if the transcript model is well or poorly supported.

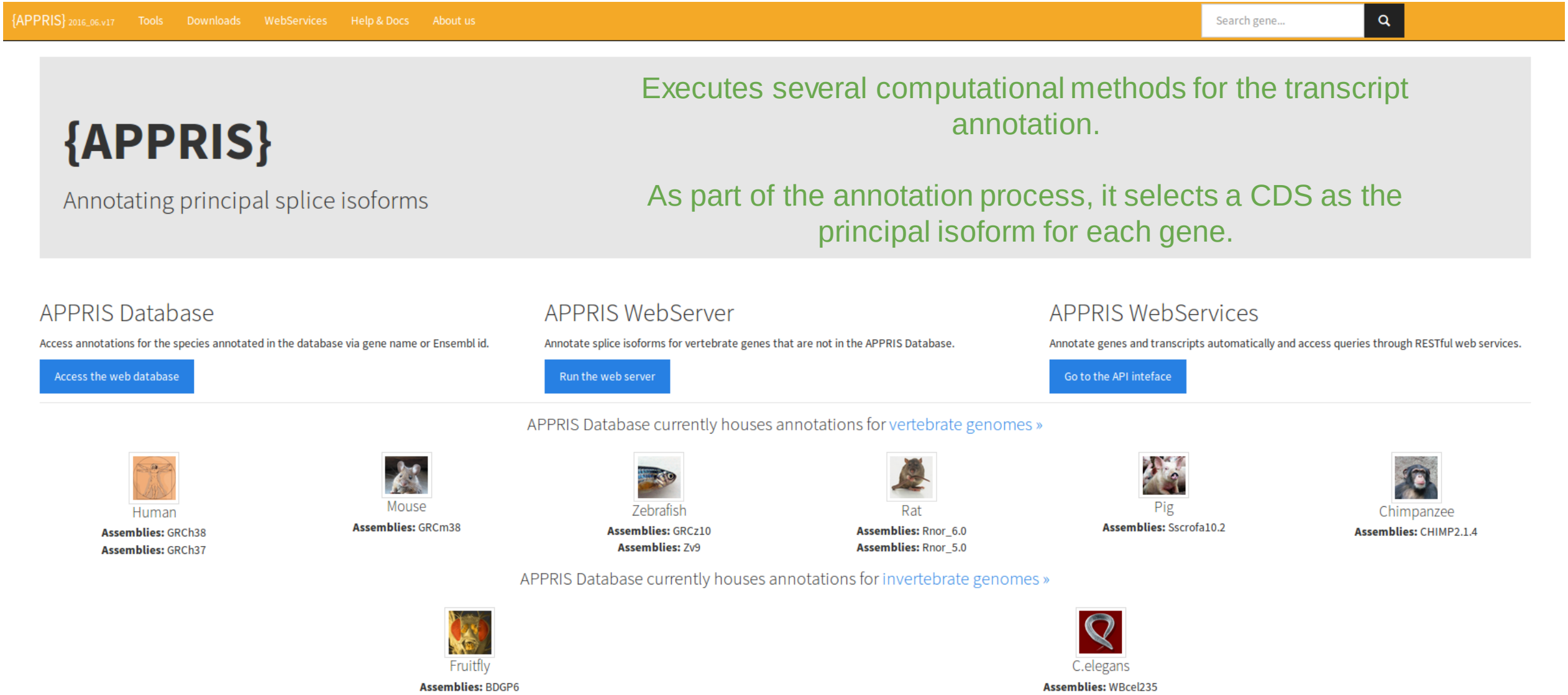
tsl1 > tsl2 > tsl3 > tsl4 > tsl5 > tslNA (the transcript was not analyzed)

PRINCIPAL ISOFORM (--canonical) Adds a flag indicating if the transcript is the canonical transcript for the gene.

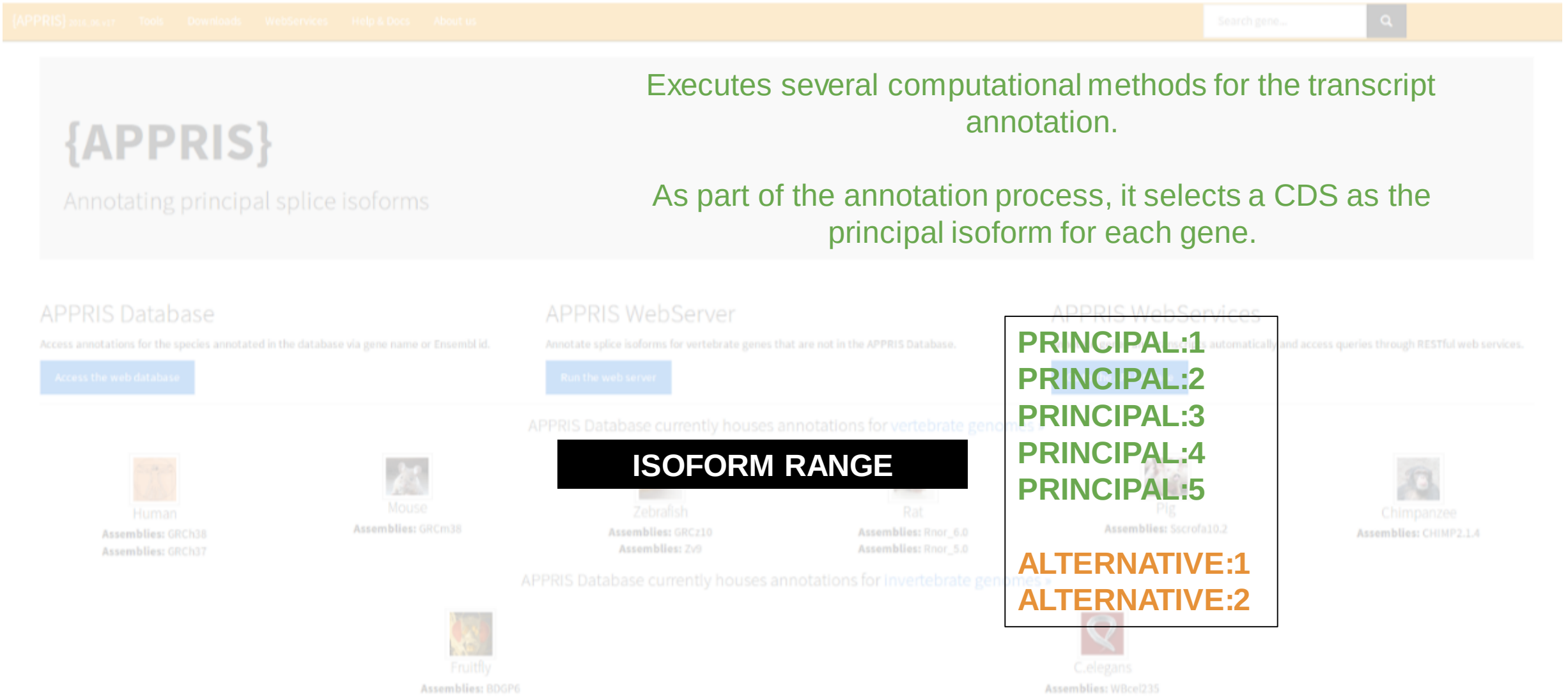
1. Longest CCDS with no stop codons
2. Longest Ensembl/Havana merged translation with no stop codons
3. Longest translation with no stop codons
4. Longest non-protein-coding transcript (if no translation)

Principal Isoform - APPRIS

http://appris.bioinfo.cnio.es/#/



Principal Isoform - APPRIS



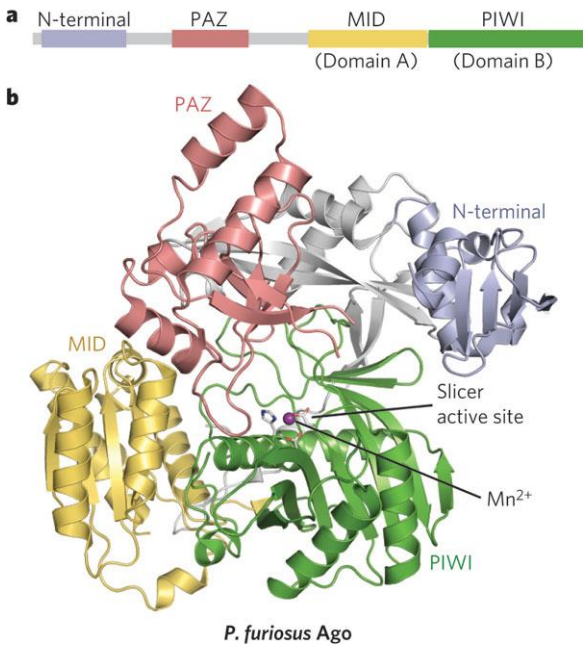
Protein annotation

PROTEIN(--protein) Ensembl protein identifier e.g. ENSP00000470877

UNIPROT IDENTIFIERS (--uniprot) Protein identifiers in UniProt databases (SWISSPROT, TREMBL y UniParc) e.g. SH3Y1_HUMAN (SWISSPROT), C9J4Z8_HUMAN (TREMBL), UPI0000208A67 (UniParc)

DOMAINS (--domains) Protein overlapping domains

Pfam, Prosite, InterPro
e.g. Pfam_domain:PF00071



P. furiosus Ago

VEP standalone script output

Links

- Top of page
- VEP run statistics
- General statistics
- Variant classes
- Consequences (most severe)
- Consequences (all)
- Coding consequences
- SIFT summary
- PolyPhen summary
- Variants by chromosome
- Position in protein

VEP run statistics

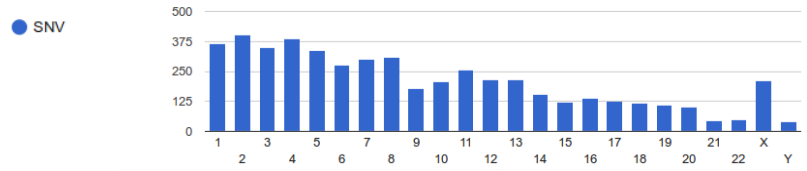
VEP version (API)	85 (85)
Cache/Database	/home/epineiro/analysis/pancancer/vep/ensembl-tools-release-85/scripts/variant_effect_predictor/.vep/homo_sapiens/85_GRCh37
Species	homo_sapiens
Command line options	`--format vcf --sift b --polyphen b --ccds --uniprot --hgvs --symbol --numbers --domains --regulatory --canonical --protein --biotype --uniprot --tsl --gmaf --variant_class --xref_refseq --maf_1`
Start time	2016-09-03 09:01:35
End time	2016-09-03 09:43:38
Run time	2523 seconds
Input file (format)	/home/epineiro/analysis/pancancer/genotypes/0a6be23a-d5a0-4e95-ada2-a61b2b5d9485.vcf (VCF)
Output file	/home/epineiro/analysis/pancancer/genotypes/0a6be23a-d5a0-4e95-ada2-a61b2b5d9485.vcf_output_VEP.txt [text]

General statistics

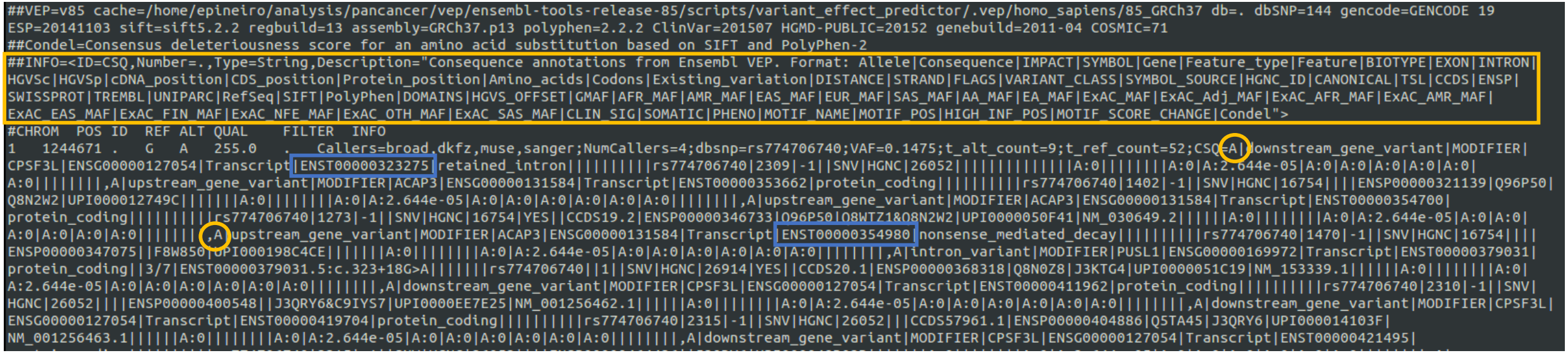
Lines of input read	5025
Variants processed	5013
Variants remaining after filtering	5013
Lines of output written	5013
Novel / existing variants	4273 (85.2%) / 740 (14.8%)
Overlapped genes	2761
Overlapped transcripts	10274
Overlapped regulatory features	490

Variant classes

Variants by chromosome



VEP standalone script output



Allele
Consequence
IMPACT
SYMBOL (--symbol)
Gene
Feature_type (--regulatory)
Feature
BIOTYPE (--biotype)
EXON (--numbers)
INTRON (--numbers)

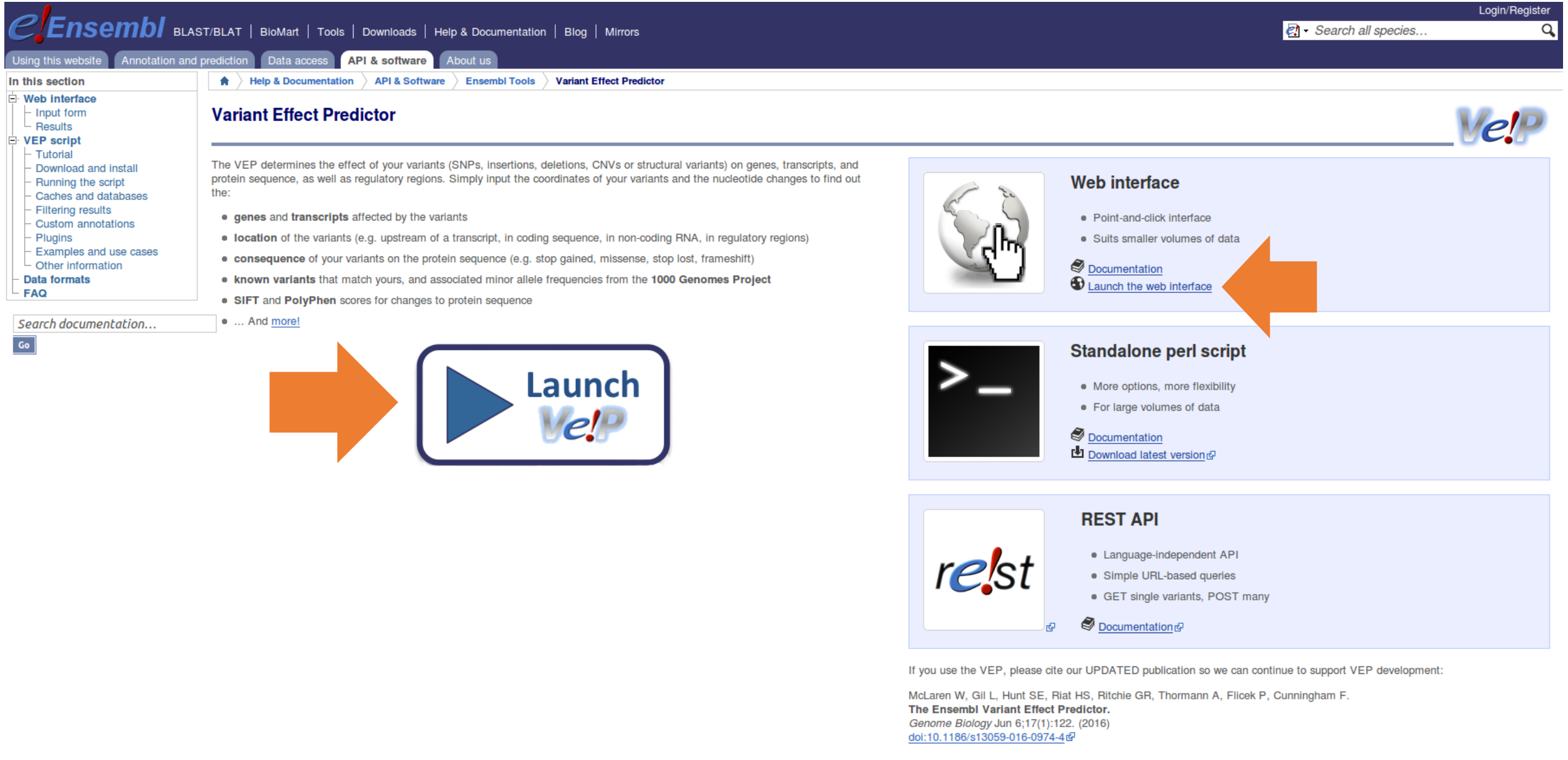
HGVSc (--hgvs)
HGVSp (--hgvs)
cDNA_position
CDS_position
Protein_position
Amino_acids
Codons
Existing_variation
DISTANCE
STRAND

FLAGS
VARIANT_CLASS (--variant_class)
SYMBOL_SOURCE
HGNC_ID
CANONICAL (--canonical)
TSL (--tsl)
CCDS (--ccds)
ENSP (--protein)
SWISSPROT (--uniprot)
TREMBL (--uniprot)

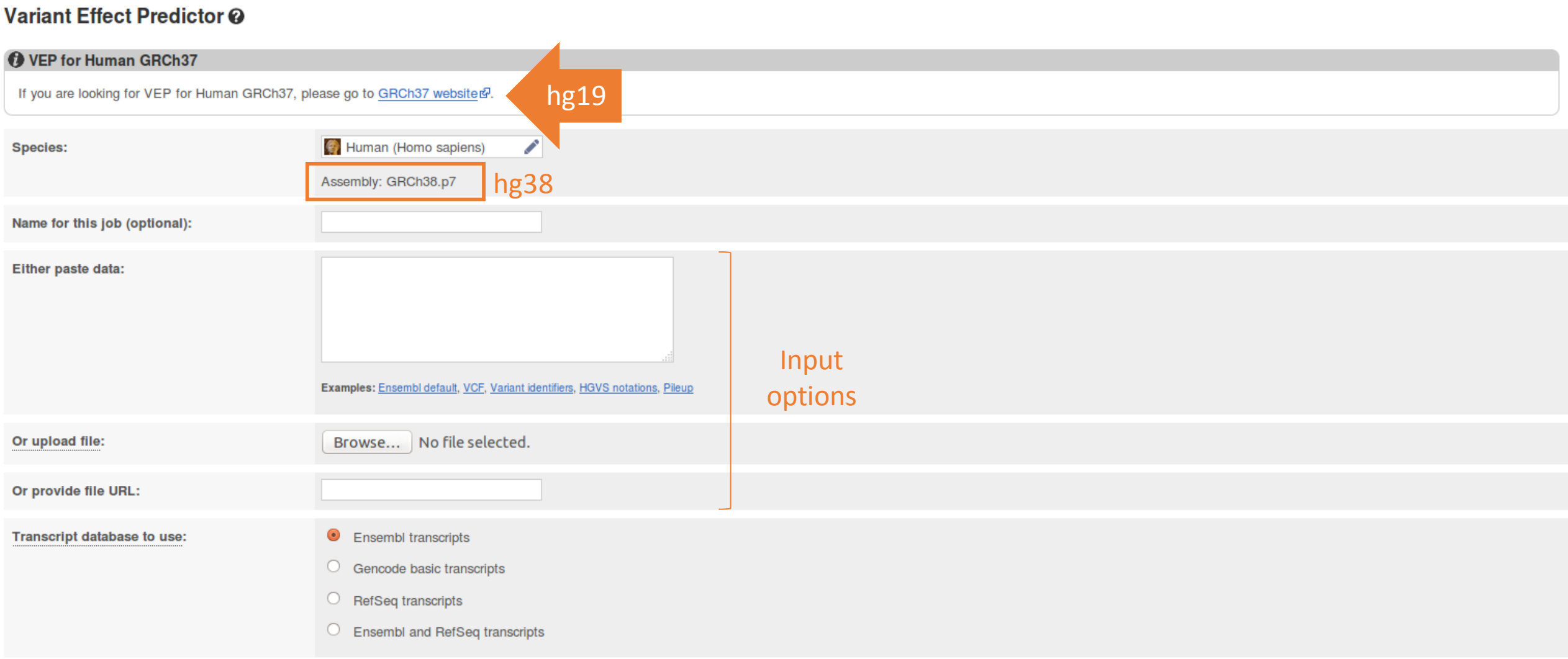
UNIPARC (--uniprot)
RefSeq (--xref_refseq)
SIFT (--sift)
PolyPhen (--polyphen)
DOMAINS (--domains)
HGVS_OFFSET
GMAF (--gmaf)
*_MAF (--maf_1kg)
AA_MAF (--maf_esp)
EA_MAF (--maf_esp)

ExAC_MAF (--maf_exac)
ExAC_*_MAF (--maf_exac)
CLIN_SIG
SOMATIC
PHENO
MOTIF_NAME
MOTIF_POS
HIGH_INF_POS
MOTIF_SCORE_CHANGE
Condel (--condel)

web execution



web execution



web execution

Identifiers and frequency data *Additional identifiers for genes, transcripts and variants; frequency data*

Identifiers

Gene symbol:	☑
CCDS:	☐
Protein:	☐
Uniprot:	☐
HGVS:	☐
CSN(p):	☐
Unshifted HGVS(p):	☐

Frequency data

Find co-located known variants:	Yes
Frequency data for co-located variants:	☑ 1000 Genomes global minor allele frequency ☐ 1000 Genomes continental allele frequencies ☐ ESP allele frequencies ☐ ExAC allele frequencies
PubMed IDs for citations of co-located variants:	☑
Include flagged variants:	☐

(p) = functionality from VEP plugin

web execution

Extra options — e.g. SIFT, PolyPhen and regulatory data

Miscellaneous

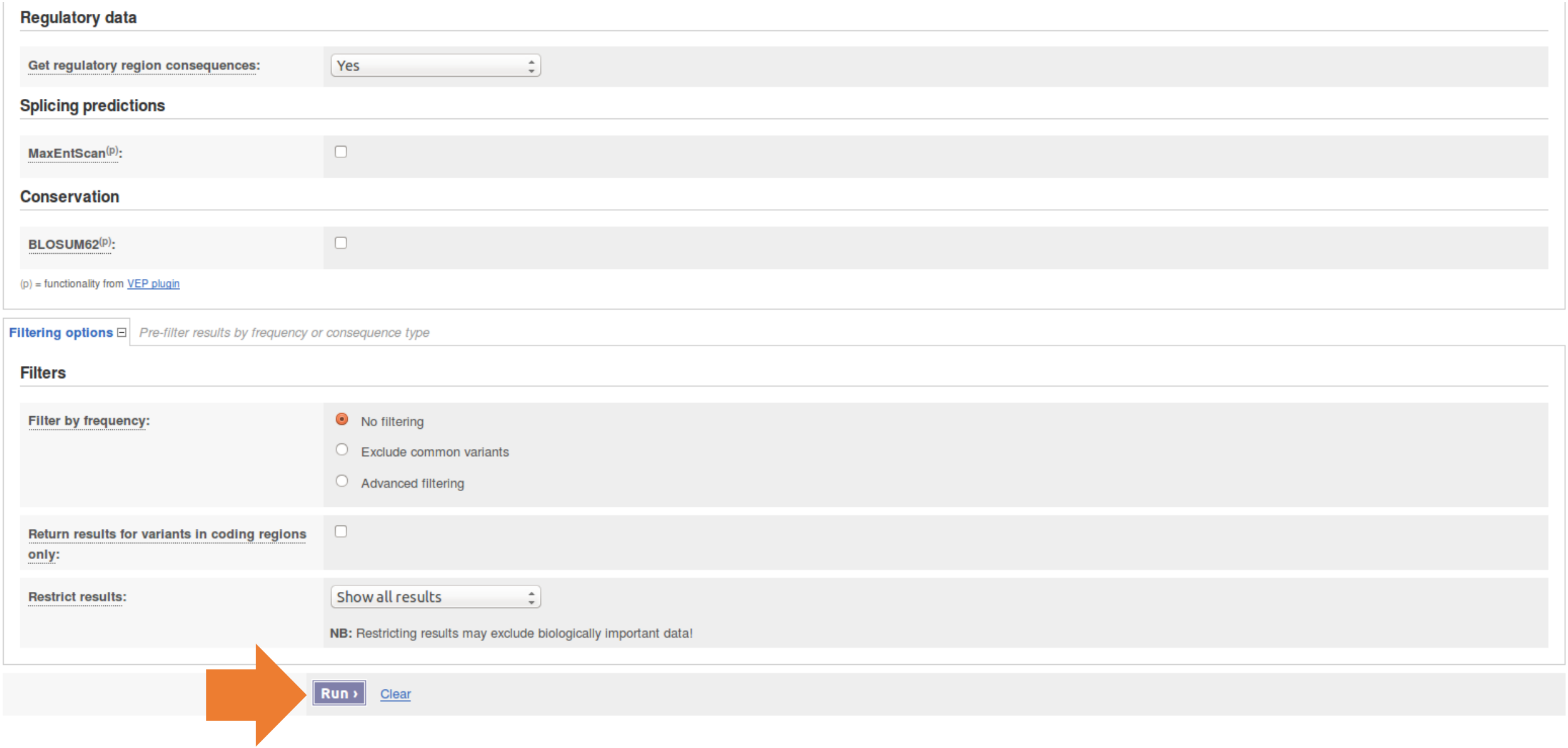
protein coding, pseudogene, processed pseudogene, miRNA, rRNA, scRNA, snoRNA and snRNA

Transcript biotype:	☑
Protein domains:	☐
Exon and intron numbers:	☐
Transcript support level:	☑
APPRIS:	☑
Identify canonical transcripts:	☐
miRNA structure(p):	☐
Upstream/Downstream distance(p):	◉ Disabled ○ Enabled

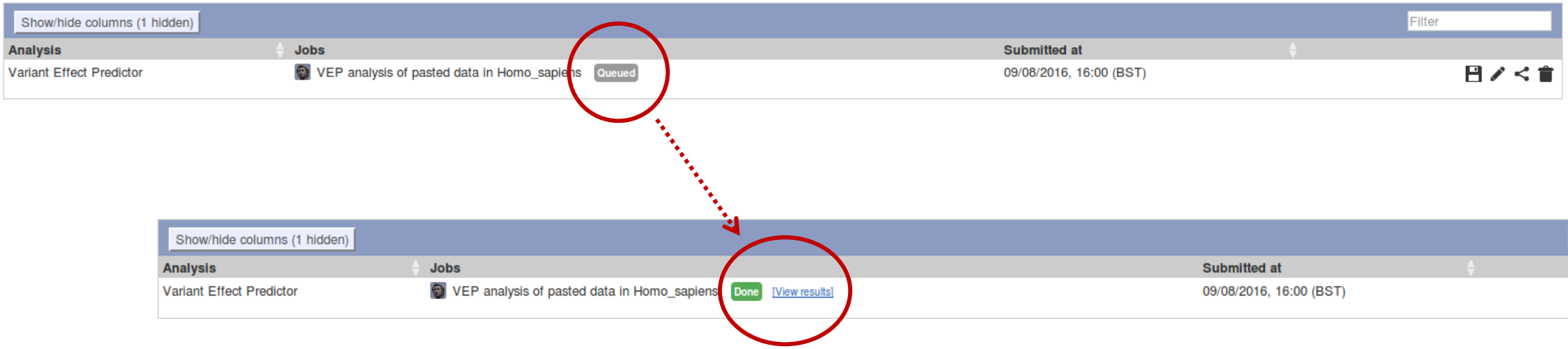
Pathogenicity predictions

SIFT:	Prediction and score
PolyPhen:	Prediction and score
Condel(p):	◉ Disabled ○ Enabled
LoFtool(p):	☐

web execution



web execution



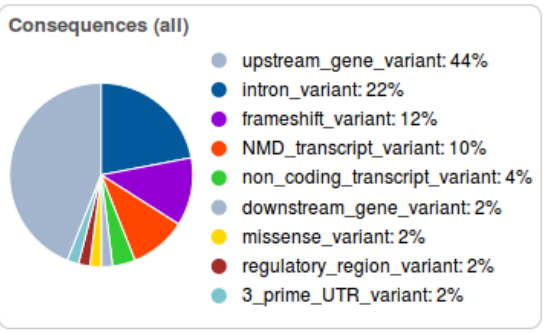
web execution

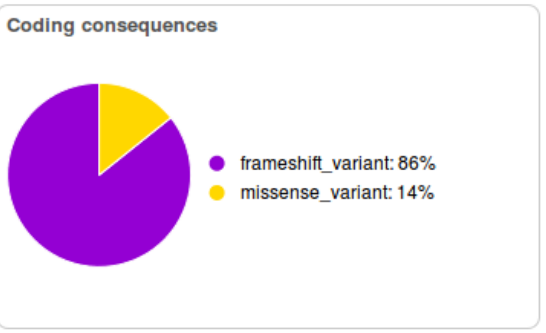
Variant Effect Predictor results

Job details

Summary statistics

Category	Count
Variants processed	3
Variants remaining after filtering	3
Novel / existing variants	-
Overlapped genes	4
Overlapped transcripts	42
Overlapped regulatory features	1





Summary

Results preview

Filtering

Navigation

Page: 1 of 1 | Show: 1 All variants

Filters

Uploaded variant | is | defined | Add

Download

All: VCF VEP TXT

BioMart: Variants Genes

Download

Results table

Show/hide columns

Uploaded variant	Location	Allele	Consequence	Impact	Symbol	Gene	Feature type	Feature	Biotype	Exon	Intron	HGVSc	HGVSp	cDNA position	CDS position	Pr po
1_818046_T/C	1:818046-818046	C	missense_variant	MODERATE	AL645608.2	ENSG00000269308	Transcript	ENST00000594233	protein_coding	1/3	-	-	-	4	4	2
2_265023_C/A	2:265023-265023	A	intron_variant	MODIFIER	ACP1	ENSG00000143727	Transcript	ENST00000272065	protein_coding	-	1/5	-	-	-	-	-
2_265023_C/A	2:265023-265023	A	intron_variant	MODIFIER	ACP1	ENSG00000143727	Transcript	ENST00000272067	protein_coding	-	1/5	-	-	-	-	-
2_265023_C/A	2:265023-265023	A	upstream_gene_variant	MODIFIER	SH3YL1	ENSG00000035115	Transcript	ENST00000356150	protein_coding	-	-	-	-	-	-	-
2_265023_C/A	2:265023-265023	A	upstream_gene_variant	MODIFIER	SH3YL1	ENSG00000035115	Transcript	ENST00000402632	protein_coding	-	-	-	-	-	-	-
2_265023_C/A	2:265023-265023	A	upstream_gene_variant	MODIFIER	SH3YL1	ENSG00000035115	Transcript	ENST00000403657	protein_coding	-	-	-	-	-	-	-
2_265023_C/A	2:265023-265023	A	upstream_gene_variant	MODIFIER	SH3YL1	ENSG00000035115	Transcript	ENST00000403658	protein_coding	-	-	-	-	-	-	-
2_265023_C/A	2:265023-265023	A	upstream_gene_variant	MODIFIER	SH3YL1	ENSG00000035115	Transcript	ENST00000403712	protein_coding	-	-	-	-	-	-	-

THE END

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