Variant annotation from Supplementary Materials (Pabinger, et al)

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;
FOLD-X [106]			yes	no	no	no	yes	yes	It performs protein stability analysis.

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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
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).

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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/1000Genomes/DGV annotation --identifies protein-truncating variants -- filtering by function	yes	yes	no	yes	yes	no	
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.

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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 protein alignments;
VARIANT [164]	VCF	Web report, text	yes	no	no	no	yes	yes	

CSV = comma separated value

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