**https://annovar.openbioinformatics.org/en/latest/**

**ANNOVAR (ANNOtate VARiation)** is a software tool used to annotate genetic variants, mainly in Whole Genome Sequencing (WGS).

Given a list of variants chromosome with , start position, end position, reference nucleotide and observed nucleotides, ANNOVAR can perform:

**Gene-based annotation:** identify whether SNPs or CNVs cause protein-coding changes and the amino acids that are affected. Users can flexibly use RefSeq genes, UCSC genes, ENSEMBL genes, GENCODE genes, AceView genes, or many other gene definition systems.

**Region-based annotation:** identify variants in specific genomic regions, for example, conserved regions among 44 species, predicted transcription factor binding sites, segmental duplication regions, GWAS hits, database of genomic variants, DNAse I hypersensitivity sites, ENCODE H3K4Me1/H3K4Me3/H3K27Ac/CTCF sites, ChIP-Seq peaks, RNA-Seq peaks, or many other annotations on genomic intervals.

**Filter-based annotation:** identify variants that are documented in specific databases, for example, whether a variant is reported in dbSNP, what is the allele frequency in the 1000 Genome Project, NHLBI-ESP 6500 exomes or Exome Aggregation Consortium (ExAC) or Genome Aggregation Database (gnomAD), calculate the SIFT/PolyPhen/LRT/MutationTaster/MutationAssessor/FATHMM/MetaSVM/MetaLR scores, find intergenic variants with GERP++ score<2 or CADD>10, or many other annotations on specific mutations.

**Other functionalities:** Retrieve the nucleotide sequence in any user-specific genomic positions in batch, identify a candidate gene list for Mendelian diseases from exome data, and other utilities.

BWA-MEM (Burrows-Wheeler Aligner - Maximal Exact Matches)