

# Please cite:

## Graphical Tools

### Cytoscape

Shannon P, Markiel A, Ozier O, et al. Cytoscape: a software environment for integrated models of biomolecular interaction networks. *Genome Res.* 2003;13(11):2498-2504. doi:10.1101/gr.1239303

### Gene Set Enrichment Analysis (GSEA)

Subramanian A, Tamayo P, Mootha VK, et al. Gene set enrichment analysis: a knowledge-based approach for interpreting genome-wide expression profiles. *Proc Natl Acad Sci U S A.* 2005;102(43):15545-15550. doi:10.1073/pnas.0506580102

### Integrative Genomics Viewer (IGV)

Robinson JT, Thorvaldsdóttir H, Winckler W, et al. Integrative genomics viewer. *Nat Biotechnol.* 2011;29(1):24-26. doi:10.1038/nbt.1754

### Jalview

Waterhouse AM, Procter JB, Martin DM, Clamp M, Barton GJ. Jalview Version 2--a multiple sequence alignment editor and analysis workbench. *Bioinformatics.* 2009;25(9):1189-1191. doi:10.1093/bioinformatics/btp033

## Online Databases and Related Tools

### Clustal Omega (EBI-EMBL)

Sievers F, Wilm A, Dineen D, et al. Fast, scalable generation of high-quality protein multiple sequence alignments using Clustal Omega. *Mol Syst Biol.* 2011;7:539. Published 2011 Oct 11. doi:10.1038/msb.2011.75

### Ensembl (EMBL-EBI)

Yates AD, Achuthan P, Akanni W, et al. Ensembl 2020. *Nucleic Acids Res.* 2020;48(D1):D682-D688. doi:10.1093/nar/gkz966

### Ensembl: Biomart

Kinsella RJ, Kähäri A, Haider S, et al. Ensembl BioMart: a hub for data retrieval across taxonomic space. *Database (Oxford).* 2011;2011:bar030. Published 2011 Jul 23. doi:10.1093/database/bar030

### Ensembl: Variant Effect Predictor (VEP)

McLaren W, Gil L, Hunt SE, et al. The Ensembl Variant Effect Predictor. *Genome Biol.* 2016;17(1):122. Published 2016 Jun 6. doi:10.1186/s13059-016-0974-4

### European Nucleotide Archive (ENA, EMBL-EBI)

Leinonen R, Akhtar R, Birney E, et al. The European Nucleotide Archive. *Nucleic Acids Res.* 2011;39(Database issue):D28-D31. doi:10.1093/nar/gkq967

### Filtering Expression Tables (FIESTA Viewer)

<https://bioinfoqp.cnb.csic.es/tools/FIESTA/index.php>

Gene Expression Omnibus (GEO, NCBI)

Edgar R, Domrachev M, Lash AE. Gene Expression Omnibus: NCBI gene expression and hybridization array data repository. *Nucleic Acids Res.* 2002;30(1):207-210. doi:10.1093/nar/30.1.207

The Gene Ontology Resource (NHGRI, NIH)

Ashburner M, Ball CA, Blake JA, et al. Gene ontology: tool for the unification of biology. The Gene Ontology Consortium. *Nat Genet.* 2000;25(1):25-29. doi:10.1038/75556

GMT files generator for GSEA (mygmt)

<https://bioinfogp.cnb.csic.es/tools/mygmt/>

MAFFT (EBI-EMBL)

Katoh K, Standley DM. MAFFT multiple sequence alignment software version 7: improvements in performance and usability. *Mol Biol Evol.* 2013;30(4):772-780. doi:10.1093/molbev/mst010

MUSCLE (EBI-EMBL)

Edgar RC. MUSCLE: multiple sequence alignment with high accuracy and high throughput. *Nucleic Acids Res.* 2004;32(5):1792-1797. Published 2004 Mar 19. doi:10.1093/nar/gkh340

Panther Classification System (NHGRI, NSF)

Mi H, Muruganujan A, Ebert D, Huang X, Thomas PD. PANTHER version 14: more genomes, a new PANTHER GO-slim and improvements in enrichment analysis tools. *Nucleic Acids Res.* 2019;47(D1):D419-D426. doi:10.1093/nar/gky1038

Sequence Read Archive (SRA, NCBI)

Leinonen R, Sugawara H, Shumway M; International Nucleotide Sequence Database Collaboration. The sequence read archive. *Nucleic Acids Res.* 2011;39(Database issue):D19-D21. doi:10.1093/nar/gkq1019

Single Nucleotide Polymorphisms Viewer (SNPer)

<https://bioinfogp.cnb.csic.es/tools/snper/>

STRING Database (SIB, CPR, EMBL)

Szklarczyk D, Gable AL, Lyon D, et al. STRING v11: protein-protein association networks with increased coverage, supporting functional discovery in genome-wide experimental datasets. *Nucleic Acids Res.* 2019;47(D1):D607-D613. doi:10.1093/nar/gky1131

Venn diagrams Generator (Venny)

<https://bioinfogp.cnb.csic.es/tools/venny/>

## Bioconductor Packages (R Language)

### DESeq2

Love MI, Huber W, Anders S. Moderated estimation of fold change and dispersion for RNA-seq data with DESeq2. *Genome Biol.* 2014;15(12):550. doi:10.1186/s13059-014-0550-8

### Rbowtie2

Wei Z, Zhang W, Fang H, Li Y, Wang X. esATAC: an easy-to-use systematic pipeline for ATAC-seq data analysis. *Bioinformatics.* 2018;34(15):2664-2665. doi:10.1093/bioinformatics/bty141

### Rsamtools

Morgan M, Pages H, Obenchain V, Hayden N. (2016). Rsamtools: Binary alignment (BAM), FASTA, variant call (BCF), and tabix file import. R package version, 1(0), 677-689

### Rsubread

Liao Y, Smyth GK, Shi W. The R package Rsubread is easier, faster, cheaper and better for alignment and quantification of RNA sequencing reads. *Nucleic Acids Res.* 2019;47(8):e47. doi:10.1093/nar/gkz114

## Command-Line Tools (UNIX)

### bwa

Li H, Durbin R. Fast and accurate short read alignment with Burrows-Wheeler transform. *Bioinformatics.* 2009;25(14):1754-1760. doi:10.1093/bioinformatics/btp324

### bcftools

Li H. A statistical framework for SNP calling, mutation discovery, association mapping and population genetical parameter estimation from sequencing data. *Bioinformatics.* 2011;27(21):2987-2993. doi:10.1093/bioinformatics/btr509

### bcftools csq

Danecek P, McCarthy SA. BCFtools/csq: haplotype-aware variant consequences. *Bioinformatics.* 2017;33(13):2037-2039. doi:10.1093/bioinformatics/btx100

### bedtools

Quinlan AR, Hall IM. BEDTools: a flexible suite of utilities for comparing genomic features. *Bioinformatics.* 2010;26(6):841-842. doi:10.1093/bioinformatics/btq033

### Genome Analysis Tool Kit (GATK)

McKenna A, Hanna M, Banks E, et al. The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. *Genome Res.* 2010;20(9):1297-1303. doi:10.1101/gr.107524.110

### samtools

Li H, Handsaker B, Wysoker A, et al. The Sequence Alignment/Map format and SAMtools. *Bioinformatics.* 2009;25(16):2078-2079. doi:10.1093/bioinformatics/btp352

### snpEFF

Cingolani P, Platts A, Wang le L, et al. A program for annotating and predicting the effects of single nucleotide polymorphisms, SnpEff: SNPs in the genome of *Drosophila melanogaster* strain w1118; iso-2; iso-3. *Fly (Austin).* 2012;6(2):80-92. doi:10.4161/fly.19695