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**Genetic Variations databases:**

* **Entrez SNP:** <http://www.ncbi.nlm.nih.gov/snp/>
* **HapMap:** <http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap27_B36/>
* **1000 Genomes:** <http://www.1000genomes.org/>
* **Database of genomic variants (DGV)**: <http://projects.tcag.ca/variation/>
* dbVar: <http://www.ncbi.nlm.nih.gov/projects/dbvar/>
* **NHGRI GWAS Catalog**: <http://www.genome.gov/gwastudies/>
* **EBI GWAS Catalog:** <https://www.ebi.ac.uk/gwas/>
* **Decipher**:<https://decipher.sanger.ac.uk/application/>
* dbRIP: <http://dbrip.brocku.ca/>
* HGVS mutation databases**:** <http://www.hgvs.org/dblist/dblist.html>
* **NCBI PheGenI**:<http://www.ncbi.nlm.nih.gov/gap/phegeni>
* **Clinical Genomic Database (NHGRI)**: <http://research.nhgri.nih.gov/CGD/>
* **PharmGKB:** <http://www.pharmgkb.org/>
* **HGMD:** <https://portal.biobase-international.com/cgi-bin/portal/login.cgi>
* **Cosmic:** <http://cancer.sanger.ac.uk/cancergenome/projects/cosmic/>
* **Tumor Portal:** <http://www.tumorportal.org/>

**Disease causing SNPs/Genes:**

* Disease/phenotype centric databases:
  + **OMIM:** <http://www.ncbi.nlm.nih.gov/omim/>
  + **HuGE Navigator**: <http://hugenavigator.net/>
  + **HGMD Public**: <http://www.hgmd.org/>
  + **CGD:** <http://research.nhgri.nih.gov/CGD/>
  + **NextBio**: [hsls.nextbio.com](http://www.hsls.nextbio.com)

**Genetic variations in the context of a genomic region:**

* NCBI Map Viewer: <http://www.ncbi.nlm.nih.gov/mapview/>
* UCSC Genome Browser: <http://genome.ucsc.edu/cgi-bin/hgGateway>
* UCSC Table Browser: <http://genome.ucsc.edu/cgi-bin/hgTables?command=start>

**Functional analysis of mutations:**

Variant Effect Predictor: <http://useast.ensembl.org/info/docs/tools/vep/index.html>

Biobase Genome Trax: <https://portal.biobase-international.com/cgi-bin/portal/login.cgi>

* Entrez Protein: <http://www.ncbi.nlm.nih.gov/sites/entrez?db=protein&cmd=search&term>=
* NCBI Amino Acid Explorer: <http://www.ncbi.nlm.nih.gov/Class/Structure/aa/aa_explorer.cgi>
* NCBI Mutation Analyzer:  <http://www.ncbi.nlm.nih.gov/Class/Structure/aa/aa_explorer.cgi?mode=translate>
* TMHMM Server: <http://www.cbs.dtu.dk/services/TMHMM-2.0/>
* **Amino acid properties:** <http://www.russell.embl.de/aas/>
* **F-SNP**: <http://compbio.cs.queensu.ca/F-SNP/>
* SIFT: <http://sift.jcvi.org/>
* PolyPhen: <http://genetics.bwh.harvard.edu/pph/>
* SNPs3D: <http://www.snps3d.org>[/](http://www.snps3d.org/)
* ESE Finder: <http://rulai.cshl.edu/cgi-bin/tools/ESE3/esefinder.cgi?process=home>
* **SPOT**: [http://spot.cgsmd.isi.edu](http://spot.cgsmd.isi.edu/)
* **Regulome DB:** <http://regulome.stanford.edu/>

**Health Sciences Library System:**

* InfoBooster: <http://hsls.pitt.edu/molbio/infobooster>
* Online Bioinformatics Resources Collection: <http://www.hsls.pitt.edu/guides/genetics/obrc/mutations_diseases>
* Search.HSLS.MolBio—query: “genetic variations” <http://www.hsls.pitt.edu/guides/genetics>
  + Databases/Tools
  + Articles on Databases/Tool

**Video Tutorials:**

* **Identify genetic variations associated with a human disease**

[**http://www.hsls.pitt.edu/molbio/videos/play?v=29**](http://www.hsls.pitt.edu/molbio/videos/play?v=29)

* **Identify genetic variations present is a human gene sequence (Part 1)**

[**http://www.hsls.pitt.edu/molbio/videos/play?v=27**](http://www.hsls.pitt.edu/molbio/videos/play?v=27)

* **Identify genetic variations present is a human gene sequence (Part 2)**

[**http://www.hsls.pitt.edu/molbio/videos/play?v=28**](http://www.hsls.pitt.edu/molbio/videos/play?v=28)

* **Retrieve all clinically linked Flagged SNPs associated with a list of genes (Part1)**

[**http://www.hsls.pitt.edu/molbio/videos/play?v=79**](http://www.hsls.pitt.edu/molbio/videos/play?v=79)

* **Retrieve all clinically linked Flagged SNPs associated with a list of genes (Part2)**

[**http://www.hsls.pitt.edu/molbio/videos/play?v=80**](http://www.hsls.pitt.edu/molbio/videos/play?v=80)

**Suggested Readings:**

* PLOS Computational Biology: Translational Bioinformatics

<http://www.ploscollections.org/article/browseIssue.action?issue=info:doi/10.1371/issue.pcol.v03.i11>

* Currect Topics in Genome Analysis:

<http://www.genome.gov/12514288>