Clinical Bioinformatics: Unlocking Genomics in Healthcare – Recommended Resources

Below is a list of web resources, please familiarise yourself with some of these resources to get a feel for their aim and scope. Have a look at some of the introductory pages e.g. FAQ's, tutorial pages to give you an idea of the terminology, and the appropriate context in which they are used as part of the clinical bioinformatician's workflow.

Nomenclature	
Accession number guide	www.ncbi.nlm.nih.gov/Sequin/acc.html
HGNC	www.genenames.org/
HGVS	www.hgvs.org/
LRG	www.lrg-sequence.org/
Genome Reference Consortium	www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/human/index.shtml
Mutalyzer	https://mutalyzer.nl/
Sequence Databases	
INSDC	www.insdc.org/
EBI	www.ebi.ac.uk/ena/
DDBJ	www.ddbj.nig.ac.jp/
GenBank	www.ncbi.nlm.nih.gov/genbank
RefSeq	https://www.ncbi.nlm.nih.gov/refseq/
RefSeqGene	http://www.ncbi.nlm.nih.gov/refseq/rsg/
UniProt	http://www.ebi.ac.uk/uniprot/
EBI training	http://www.ebi.ac.uk/training/online/
Genome Browsers	
Ensembl Genome Browser	www.ensembl.org/index.html
UCSC Genome Browser	www.genome.ucsc.edu/cgi-bin/hgGateway
Genome Projects	
Human Genome Project	http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml
Human Variome Project	http://www.humanvariomeproject.org/
1000 Genomes Project	http://www.1000genomes.org/
Mouse Genome Informatics	http://www.informatics.jax.org/

100000 Genomes Project	http://www.genomicsengland.co.uk/
UK 10000 genomes project	http://www.uk10k.org/
Personal genome project	http://www.personalgenomes.org/
Variation Databases	
dbSNP	www.ncbi.nlm.nih.gov/projects/SNP/
DGV	http://dgv.tcag.ca/dgv/app/home
Phenotype Databases	
OMIM	http://www.ncbi.nlm.nih.gov/omim
GEN2PHEN	www.gen2phen.org/
European Genome / Phenome archive	https://www.ebi.ac.uk/ega/home
Variant Databases	
HGVS LSDB listing	http://www.hgvs.org/dblist/glsdb.html
InSiGHT	www.insight-group.org/
Ecaruca	http://umcecaruca01.extern.umcn.nl:8080/ecaruca/
Decipher	http://decipher.sanger.ac.uk/
HGMD	http://www.hgmd.cf.ac.uk/ac/index.php
Unique	http://www.rarechromo.org/html/home.asp
dbVar	http://www.ncbi.nlm.nih.gov/dbvar/
ISCA	https://www.iscaconsortium.org/
LOVD	http://www.lovd.nl/2.0/www.ebi.ac.uk/Tools/msa/clustalo/
Exome Aggregation Consortium	http://exac.broadinstitute.org/
ClinVar	http://www.ncbi.nlm.nih.gov/clinvar/
Sequence Alignment	
Sequence Tools	http://www.lovd.nl/2.0/www.ebi.ac.uk/Tools/msa/clustalo/
Sequence Similarity	http://www.ebi.ac.uk/Tools/sss/
Sequence Alignment	http://www.ebi.ac.uk/Tools/msa/clustalo/
Variant Interpretation	
ALIGN-GVGD	www.agvgd.iarc.fr/agvgd_input.php
PolyPhen-2	http://www.genetics.bwh.harvard.edu/pph2/
SIFT	http://www.sift.jcvi.org/
Alamut	http://www.interactive-biosoftware.com/alamut.html
Missense tools	http://www.ngrl.org.uk/Manchester/page/missense-prediction-tools
Variant effect predictor	http://www.ensembl.org/info/docs/tools/vep/index.html

Literature Resources	
PubMed	http://www.ncbi.nlm.nih.gov/pubmed/
Google Scholar	http://www.scholar.google.co.uk
Ontologies	
Gene Ontology	http://www.geneontology.org/
HPO	http://human-phenotype-ontology.org/
Rare Disease Registries	
Orphanet	http://www.orpha.net/consor/cgi-bin/index.php
Rare diseases	https://www.rarediseases.org
Patient Fora	
Patients like me	http://www.patientslikeme.com/
Genetic disease charities	
Genetic Alliance	http://www.geneticalliance.org.uk/
Jeans for Genes	http://www.jeansforgenesday.org/aboutjeansforgenesday
UK Genetic Testing Network	http://ukgtn.nhs.uk/