## **APPENDIX**

# APPENDIX A: GENETIC AND GENOMIC ONLINE RESOURCES

#### **Compilation of Genetic and Genomic Online Resources**

- **OMIM**: OMIM: Online Mendelian Inheritance in Man (https://omim.org/) A comprehensive database of human genes and genetic phenotypes, focusing on the relationship between genotype and phenotype.
- The Human Protein Atlas: The Human Protein Atlas (https://www.proteinatlas.org/)— Provides detailed information on the tissue and cell distribution of proteins in the human body.
- **GeneCards**: GeneCards (https://www.genecards.org/)— An integrated database offering comprehensive information on all annotated and predicted human genes.
- **ClinGen**: ClinGen (https://www.clinicalgenome.org/) Defines the clinical relevance of genes and variants for use in precision medicine and research.
- ClinVar: ClinVar (https://www.ncbi.nlm.nih.gov/clinvar/) Archives and aggregates information about relationships among genomic variation and human health.
- GeneReviews: GeneReviews (https://www.ncbi.nlm.nih.gov/books/NBK1116/)

   Provides expertauthored, peer-reviewed disease descriptions focusing on diagnosis, management, and genetic counseling.
- **NIH Gene**: NIH Gene (https://www.ncbi.nlm.nih.gov/genbank/)— A searchable database of genes, focusing on genomes that have been completely sequenced.
- **Human Disease Genes**: Human Disease Genes (https://humandiseasegenes.nl/) Offers professional information about genes and their clinical consequences.
- ExpressAnalyst: ExpressAnalyst (https://expressanalyst.ca/) A web-based platform for gene expression data analysis.
- Reactome: Reactome (https://reactome.org/) A free, curated database of biological pathways.
- Comparative Toxicogenomics Database: Comparative Toxicogenomics Database
   (https://ctdbase.org/about/) Provides information on the effects of environmental chemicals on human health.
- National Comprehensive Cancer Network: NCCN (https://www.nccn.org/) Provides guidelines and resources for cancer care.
- National Library of Medicine (Gene): NLM Gene (https://www.ncbi.nlm.nih.gov/) Offers access to a wide range of biomedical and genomic information.

- The Expression Atlas: The Expression Atlas (https://www.ebi.ac.uk/gxa/home) Provides information on gene expression patterns under different biological conditions.
- **Ensembl**: Ensembl (https://useast.ensembl.org/index.html) A genome browser for vertebrate genomes.
- Gene Omnibus: Gene Omnibus (https://www.ncbi.nlm.nih.gov/genbank/)

   A repository for gene expression data.
- NCBI Genome Data Viewer: NCBI Genome Data Viewer (https://www.ncbi.nlm.nih.gov/genbank/)— A genome browser for visualizing genomic data.
- UCSC Genome Browser: UCSC Genome Browser (https://genome.ucsc.edu/cgi-bin/hgGateway)

   A tool for exploring the human genome.
- **SFARI Gene**: SFARI Gene (https://gene.sfari.org/) A database for autism research.
- PhenX Toolkit: PhenX Toolkit (https://www.phenxtoolkit.org/) A repository of standardized measurement protocols.
- Gabriella Miller Kids First Data Resource Center: Kids First Data Resource Center
   (https://d3b.center/kidsfirst/) Provides biospecimens and genetic data related to pediatric cancer and
   structural birth defects.
- **Health and Retirement Study**: Health and Retirement Study (https://hrs.isr.umich.edu/about) A longitudinal study of health and economic conditions in older adults.
- MetaboAnalyst: MetaboAnalyst (https://www.metaboanalyst.ca/) A tool for metabolomics data analysis.
- Plink: Plink (https://zzz.bwh.harvard.edu/plink/) A toolset for whole-genome association analysis.
- **Plink 1.9 Update**: Plink 1.9 Update (https://www.cog-genomics.org/plink/) An updated version of the Plink toolset.
- IGV Integrative Genomics Viewer: IGV (https://software.broadinstitute.org/software/igv/) A tool for interactive exploration of large genomic datasets.
- **GeneMania**: GeneMania (https://genemania.org/) A tool for predicting gene function and gene-gene interactions.
- Cytoscape: Cytoscape (https://cytoscape.org/) A software platform for visualizing complex networks.
- OpenEpi.com: OpenEpi.com (https://www.openepi.com/Menu/OE\_Menu.htm) Provides statistical
  calculators for epidemiology.
- Coriell Institute: Coriell Institute A biobank for human cells and DNA.
- NHANES: NHANES A program of studies designed to assess the health and nutritional status of adults and children in the United States.
- Pharmacogenomics:
  - ° CPIC (https://cpicpgx.org/guidelines/) Provides guidelines for pharmacogenomics.
  - ° PharmGKB (https://www.pharmgkb.org/) A resource for pharmacogenomics knowledge.
  - ° PharmVar (https://www.pharmvar.org/) A database of pharmacogenetic variants.

#### • Cancer/Control Data Sets:

- GTEx (https://gtexportal.org/home/) Provides data on gene expression and regulation in multiple human tissues.
- ° cBioPortal (https://www.cbioportal.org/) A resource for exploring cancer genomics data.

#### • miRNA:

- ° miRDB A database for miRNA target prediction.
- ° miRBase A database of published miRNA sequences and annotations.
- miRNet A tool for miRNA network analysis.
- ° TargetScan A tool for predicting biological targets of miRNAs.

## **UPDATE & CHANGE LOG**

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• First publication