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 expert-authored, 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

July 16, 2025

- Updated 8.4 ACMG Secondary Findings Guidelines from v3.2 (2023) to v3.3 (2025) in text and reference list.
- Updated 8.5 ACMG Secondary Findings Guidelines from v3.2 (2023) to v3.3 (2025) in resource list.

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- First publication – view this version of the book in Print PDF or Digital PDF versions