

Human Genome Biology (MMD-892)

Credit Hours 3 (3-0)

Course Description

Genomics, the study of the structure, function and evolution of the genome, is among the newest and most rapidly growing fields of both basic and applied science, and nearly all of the more traditional disciplines in biology are being revolutionized by genomic tools. The growing flood of data on the DNA, RNA and protein sequences of organisms provides unprecedented opportunities to address fundamental biological questions such as the causes of disease, the genetic basis of development, the extent and causes of adaptive evolution, and the nature of gene regulation. Genome biology is a highly interdisciplinary field, encompassing concepts and practices from such diverse fields as Cell and Molecular Biology, Evolutionary Genetics, and Computer Science.

Educational Objective

- Students in the Genome Biology program will receive a uniquely broad training in these concepts and practices, with a key focus on conceptual training in molecular biology, bioinformatics and evolutionary genetics, and practical training in both computational and wet-lab genomics research.
- A key focus of this is to train biologists in the breadth of knowledge and skills required to understand, generate, and use results from genomics.

Course Contents

1. Overview and Introduction to Genomics
 - Genomes
 - Transcriptomes
 - Proteomes
2. Structural Features of Genome Sequences
 - The Human Genome Anatomy, Composition, and Structure
3. Genome Mapping
 - Genetic Mapping
 - Physical Mapping

4. Genome Sequencing
 - The Methodology for DNA Sequencing
 - Assembly of a Contiguous DNA Sequence
 - The Human Genome Projects
 - Emerging Sequencing Methods: The Next Generation
5. Genome assembly
 - Hierarchical Sequencing
 - Shotgun Sequencing
 - Sequence Verification
6. Genome annotation, Gene prediction, Hidden Markov Models
 - EST Sequencing
 - Ab Initio Gene Discovery
 - Hidden Markov Models and Gene Finding
 - Regulatory Sequences
 - Non-Protein Coding Genes
 - Clustering of Genes by Sequence Similarity
 - Clusters of Orthologous Genes
 - Gene Ontology
7. Origins of heritable disease: SNPs, Repeats, Duplications and Rearrangements
 - The Nature of Single Nucleotide Polymorphisms
 - Classification of SNPs
 - Distribution of SNPs
8. Phylogenetics and Phylogenomics
 - Phylogenetic Classification of Genes
9. Genomic Variations and Population Genetics
 - Linkage Disequilibrium and Haplotype Maps
 - Population Genetics
 - Recombination Mapping
 - QTL Mapping
 - Linkage Disequilibrium Mapping
10. Population Genomics and Disease

- SNP Discovery
- SNP Genotyping
- High-throughput genotyping platforms
- Haplotype phasing methods

11. Transcriptomics: Measurement and analysis of Gene Expression

- Parallel Analysis of Gene Expression: Microarrays
- Applications of Microarray Technology
- Experimental Design
- Microarray Technologies
- Labeling and Hybridization of cDNAs
- Statistical Analysis of cDNA Microarray Data
- Microarray Data Mining
- ChIP Chips and Gene Regulation

12. Expression as a phenotype: eQTLs

- Functional genomics: high-throughput genetics
Functional Genomics
- Saturation Forward Genetics
- High-Throughput Reverse Genetics
- Fine-Structure Genetics

13. Epigenomics: beyond primary sequence

- Chromatin modifications and gene expression

14. Proteomics methods and applications

- Protein Separation and 2D-PAGE
- Mass Spectrometry
- Immunochemistry
- Protein Microarrays

15. Structural Proteomics

- Objectives of Structural Proteomics
- Protein Structure Determination
- Protein Structure Prediction and Threading

16. Interactome mapping: large-scale characterization of biological interactions

- Protein Interaction Maps

17. Comparative Genomics

18. Metabolomics

19. *In silico* Genomics

Recommended Books

1. Brown, T. A. (2002). Mapping genomes. In *Genomes. 2nd edition*. Wiley-Liss.
2. Dale, J. W., Schantz, M. V., & Klena, J. D. (2002). *From genes to genomes*. Wiley.
3. Reece, R. J. (2004). *Analysis of genes and genomes* (pp. 88-95). Hoboken, NJ: John Wiley & Sons.