ABS-936 Genomics and Transcriptomics 3 (3-0)

Educational Objectives

1. The course aims to produce graduates who can understand the concepts of genomics and advanced techniques to sequence and identify disease causing transcripts.

    a. To understand theory and underlying mechanisms related to structure and function of genomes and transcriptomes.
    b. To use different software as tools of research in genomics and transcriptomics.
    c. To explore online databases for faster and quicker way to analyze results from genomic and transcriptomic data.

Course Outcomes

2. After the completion of this course, students will be able to understand the concepts of human genome, genomics, variations in the DNA, how these variations cause human genetic diseases, new techniques and analysis related to genome data; new insights how to overcome genetic disease burden and how to improve medical practices to help early right diagnosis of human ailments. They will also able to have knowledge about genome variations and their role in personalized medicine. At the end students will understand the legislation issues regarding human genomics and data privacy ethics.

3. Course Contents

a. **Structure of the genome**
    (1) Introduction: Gene to genome to genomics
    (2) Prokaryotic and eukaryotic genome structure
    (3) Gene Finding and Sequence Annotation
    (4) Genome evolution
b. **Genome variations**

(1) Types of variations in the genome (DNA) e.g. single nucleotide variants, small insertions and deletions, copy number variation, rearrangements and tandem repeats

(2) What is the extent of variations in populations (e.g. HapMap)

c. **Introduction to Transcriptomics**

(1) Prokaryotic and eukaryotic Gene Expression

(2) ESTs and cDNA liberaries

(3) Gene-chips/Microarrays and data analysis

(4) Assembly algorithms and Functional annotation

d. **Regulatory and Epigenetic Landscapes of Eukaryotic Genomes**

(1) Functional elements in linear sequence

(2) Envisioning the Genome and Pervasive transcription

(3) Conserved and ultra-conserved elements

(4) Epigenetic regulation and eukaryotic genome evolution

e. **Genome analysis through Bioinformatics**

(1) Data-mining, Prediction of open reading frames

(2) Characteristic sequence features, and regulatory sequences

(3) Familiarity with whole genome browsers

(4) Bioinformatics of genome annotation, current status of genome sequencing projects

f. **Genomic Browsers and databases**

(1) Orthology prediction (comparative genomics)

(2) Search for transcription factor binding sites (TFBS)

(3) Computational prediction of miRNA target genes

(4) De novo prediction of regulatory motifs in genome

g. **Next generation sequencing technologies**

(1) Illumina (Solexa) sequencing

(2) Roche 454 sequencing

(3) Ion torrent: Proton / PGM sequencing

(4) SOLiD sequencing

h. **Legislation regarding human genomics data and data privacy issues**

(1) Privacy, Confidentiality, and Identifiability in Genomic Research

(2) Single nucleotide polymorphisms (SNP), Genome medicine and Pharmacogenomics

(3) HHS, Standards for Privacy of Individually Identifiable Health Information

(4) EU Data Protection Directive

Recommended Books

1. **Genes IX (9th edition),** by Benjamin Lewin, B. Jones and Bartlett publishers, 2011

2. **Genomes (2nd edition),** by Terence A Brown. Oxford: Wiley-Liss; 2002. ISBN-10: 0-471-25046-5

3. **Discovering Genomics, Proteomics and Bioinformatics 2nd edition -** by A. Malcolm Campbell and Laurie J. Heyer. Published by Cold Spring Harbor Laboratory Press

4. **Bioinformatics and Functional Genomics (3rd Edition),** by Jonathan Pevsner. Wiley-Blackwell, 2015