MOLECULAR EPIDEMIOLOGY

20405 - BIOL439 - A01 January 8 – April 8, 2024

COURSE OUTLINE

LECTURER: JOHN S. TAYLOR

Office: Petch 012 Tel: 250-472-5206 email: taylorjs@uvic.ca

Lectures: TWF: 9:30 AM – 10:20 PM – C118, David Strong Bldg.

COURSE DESCRIPTION. This course provides an introduction to the basic principles and applications of molecular epidemiology. We focus on the identification of genes that play a role in disease in humans (e.g., using linkage and association studies, exome and genome sequencing) and the implications of such discoveries for diagnosis, screening, and treatment. Cystic fibrosis, cancer, HIV progression, and the human HapMap are among the subjects covered. A key component of the course is the completion and presentation of semester-long group projects.

EVALUATION

- 1. ASSIGNMENTS: (50 pts)
 - i) OR assignment (5)
 - ii) Reading assignment: Pre-implantation genetic diagnosis (5)
 - iii) CanRisk breast cancer risk assignment (5)
 - iv) HapMap assignment: Selecting tagging SNPs (5)
 - v) Group presentation (10)
 - vi) Research report (20)
- 2. MID-TERM EXAM: (20 pts)
- 3. FINAL EXAM: (30 pts)

Grading scheme: A+ (90%-100%), A (85-89%), A- (80-84%), B+ (77-79), B (73-76%), B- (70-72%), C+ (65-69%), C (60-64%), D (50-59%), F (<50%), N (max. = 49%): Failure to complete one or more of the following: Mid-term exam, Final exam, Research Report.

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Lecture schedule

| 1 | Jan. 9 | Exposure, Spot Maps and Odds Ratio | Start Assignment 1 |
|----|--------|---|--------------------|
| 2 | 10 | Cystic Fibrosis, Consanguineous marriage, LOD Score | |
| 3 | 12 | CF, RFLPs, Linkage | Assignment 1 due |
| 4 | 16 | F508del | |
| 5 | 17 | Pre-implantation Genetics Diagnosis | Start Assignment 2 |
| 6 | 19 | Gene Therapy/ Personalized Medicine and CF | |
| 7 | 23 | Newborn Screening and PPV | |
| | 24 | Groups meet in class | Assignment 2 due |
| 8 | 26 | Cancer Linkage Studies | |
| 9 | 30 | Odds Ratio, Relative Risk, and BRCA1& BRCA2 | Start Assignment 3 |
| 10 | 31 | Interactions among loci | |
| 11 | Feb. 2 | Hazard Ratio/ Tumor Transcription | |
| 12 | 6 | Oncotyping by Dr. Jennifer Nuk | Assignment 3 due |
| 13 | 7 | Tumor Transcription/Over-diagnosis | |
| | 9 | Midterm | |
| 14 | 13 | HIV-AIDS | |
| 15 | 14 | The Hap Map, GWAS and Macular Degeneration | Start Assignment 4 |
| 16 | 16 | Groups meet in class | |
| | 20 | Reading Break | |
| | 21 | Reading Break | |
| | 23 | Reading Break | |
| 17 | 27 | Transcriptome sequencing. | Assignment 4 due |
| 18 | 28 | Exome sequencing | Last day to drop |
| 19 | Mar. 1 | Whole genomes | |
| 20 | 5 | Genetic Resilience | |
| 21 | 6 | Genome Prediction. | |
| | 8 | Group Presentations Begin | |

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Learning Objectives

- 1. Students will learn that risk, in the context of epidemiology, can be estimated using the odds ratio (OR) and hazard ratio (HR).
- 2. Students will learn how the logarithm of odds score (LOD score) is used to identify loci that influence disease risk.
- 3. Students will learn about Genome Wide Association Studies (GWAS) and Genome Prediction studies and be able to explain how they differ from Linkage Studies.
- 4. Students will be able to explain in detail methodologies/tools used to screen human populations for disease-associated loci.
- 5. Students will be able to discuss the benefits and costs associated with genetic screening and reflect upon specific studies when doing so.