University of Guelph College of Biological Sciences Department of Integrative Biology

BIOL\*3020 Population Genetics (F) 0.5 credits (3 lectures, 2 hour tutorial/lab per week, see WebAdvisor for meeting times and locations) Prerequisite: MBG\*2000 or MBG\*2040, Equate: MGB\*3000

# **Course Outline**

**Description:** This course provides an in-depth introduction to population genetics. Population genetics is the study of the genetic compositions of populations. The genetic composition of a population is characterized by things such as allele, haplotype and genotype frequencies as well as levels of linkage disequilibrium and the genealogical ancestry of alleles and haplotypes. This course will investigate how evolutionary processes shape genetic variation and how genetic variation can be used to infer evolutionary processes that are occurring in a population. Furthermore, population genetic theory is the foundation for understanding micro-evolutionary processes. This course will introduce a canonical set of models that form a foundation for micro-evolution.

#### Instructor:

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**Learning Objectives:** A student will begin to use population genetic principles to understand the causes of genetic variation and change in a population. Specifically, a student will gain expertise in the following areas:

(1) Conceptual skills:

(a) Mathematical concepts, such as frequency and probability

(b) Working usage of terminology, such as allele, locus, haplotype, linkage disequilibrium, selection coefficient and absolute, relative and marginal fitness (c) Identification of processes that cause genetic evolution: mutation, selection, migration and drift

(d) Analysis of basic models, such as one and two-locus selection, inbreeding and population structure, random genetic drift, recurrent mutation, the coalescent and ancestral selection graph, and the Mendelian basis of quantitative variation

(2) Inquiry skills:

(a) Elementary practice in data analysis in population genetics.

(b) Elementary practice in model building and hypothesis testing in population genetics.

(c) Connect course material and data analyses to variation and processes in nature through primary literature readings

(3) Basic skills:

- (a) Comprehension of scientific material.
- (b) Organized acquisition and filtering of scientific concepts, facts and methods through note taking and study.
- (c) Numeracy.
- (d) Scientific writing.

**Course Structure:** The course includes several elements: lectures, tutorials, problem sets, data analyses, suggested textbook readings, scientific literature assignments, exams and an essay. All elements of the course are integrated. Lectures introduce concepts, which are explored in greater depth in tutorial. Theoretical and practical applications of concepts are explored in problem sets and data analyses. Scientific literature readings illustrate how a concept informs us about variation in nature and link course material to modern scientific research. The essay provides a student an opportunity to explore a topic of their specific interest and illustrate an integrated understanding of how population genetics principles give insight into the topic.

Lecture: A list of lecture topics and suggested readings from the recommended textbook is provided below.

Tutorial: Tutorials are typically divided into two parts. During the first hour, students will have the opportunity to ask questions about and work through graded and practice problem sets. During the second hour, students will go to the CBS computer lab to get help with the data analysis that is due the following week. Data analyses involve using major international genetic databases, such as the human HapMap SNP database (http://hapmap.ncbi.nlm.nih.gov), the BOLD Barcode data base (http://www.boldsystems.org) or GenBank (www.ncbi.nlm.nih.gov/genbank).

Graded problem sets, data analyses and scientific literature assignments: These form an integrated set. They are posted on Wednesday by 5pm and are due the following Wednesday at the beginning of lecture.

Practice problem sets: These will also be posted on Wednesdays, but are not graded and form a basis of discussion in tutorial.

Exams: There are three exams. The exams are not comprehensive.

Essay: See the appended document that outlines the essay assignment. Note that you may hand in a preliminary outline of your essay. We will comment on the outline. Handing in an outline is not required, but may provide helpful feedback.

Lecture Topics: Below is a tentative list of lecture topics based on previous years.

Suggested readings are from *Population Genetics* by M. Hamilton, unless noted otherwise.

Unit 1: Framework and foundations

1. Overview of population genetics. (Chapter 1)

2. Introduction and review of genetic terminology and mathematical concepts that are central to population genetics. (Appendix)

3. Introduction to the HapMap database. (see handout on Courselink)

4. Review of the Hardy-Weinberg Law. (Chapter 2.1 - 2.3)

Unit 2: Selection

1. Haploid selection: Definitions of absolute, relative, and average fitness. (Chapter 6.1 [p. 185 - 189)

2. Diploid selection: Definition of marginal fitness. (Chapter 6.1 [p.189 – 193])

3. Single-locus selection dynamics: Introduction to the selection coefficient, dominance coefficient, heterozygote advantage and disadvantage, frequency-dependent selection and mutation-selection balance. (Chapters 6.2 - 6.3, 7.2 - 7.3)

4. Two-locus genetics: Definition of linkage disequilibrium (Chapter 2.1, 2.7)

5. Two-locus models of selection: Marginal fitness of haplotypes, average fitness and genetic hitch-hiking. (Chapter 7.1, 8.6)

Unit 3: Inbreeding and population structure

1. Definitions of identity-by-descent and inbreeding coefficient, and the effect of inbreeding on genotype frequencies. (Chapter 2.5 - 2.6)

2. Population structure:  $F_{IS}$ ,  $F_{ST}$ , and  $F_{IT}$ . (Chapter 4.1, 4.3 – 4.4)

3. Two-deme and island model of population structure. (Chapter 4.5)

Unit 4: Random genetic drift

- 1. Wright-Fisher model. (Chapter 3.1 3.2)
- 2. Effective population size. (Chapter 3.3 3.5)

Unit 5: Ancestral population genetics

1. The coalescent. (Chapter 3.6 - 3.7)

2. The coalescent with mutation and with population structure (Chapter 5.5, Chapter 4.6)

3. The ancestral recombination graph.

Unit 6: Molecular population genetics

1. Introduction to the BOLD database and the program MEGA. (see handout on Courselink)

2. Measuring sequence variation (Chapter 8.2 – 8.3)

- 3. Tests for selection. (Chapter 8.4)
- 4. Ancestral selection graph (Chapter 7.4)

5. Neutral and nearly neutral theory (Chapter 8.1)

Unit 7: Quantitative genetics

1. Decomposition of a phenotype into genetic and environmental components. (Chapter 9.1)

- 2. Evolutionary change in quantitative traits (Chapter 9.2)
- 3. Mendelian basis of quantitative traits (Chapter 10)

**Textbook:** *Population Genetics* by M.B. Hamilton (currently highly recommended)

**Essay:** This is the essay assignment from Fall 2013, requirements may change for the current offering.

Identify a field of scientific research that uses population genetic principles, methods and/or theory. Include a concise summary of the historical development of the field, citing the important literature. Indicate why the topic has developed in a particular sequence or if its development has been non-sequential. Indicate and describe the population genetic principles, methods and/or theory used and whether the field has introduced new principles, methods and/or theory to population genetics. Relate the principles, methods and/or theory to population genetics. Relate the principles, methods and/or theory to specific topics presented in BIOL\*3020. Comment on the historical and current state of the field, such as the robustness of its inferences and its promise to add insight into either an applied problem or to our basic understanding of nature.

Use 12 point Arial font, 2.5cm page margins (top, bottom, left and right), 1.5 line spacing and include page numbers. The maximum number of pages of text (excluding title page, references and tables and/or figures) is seven. A title page will include a title, your name, student number and course identification code. The reference section will begin on a new page, with a heading and the page limit is unrestricted. Tables and/or figures can be used, but only if they add positively to the essay and are to be placed after the reference section. Each figure or table is placed on its own page with a unique and chronologically numbered caption. Refer to the figure or table number in the text.

Given the limitation of 1.5 line spacing and seven pages of text, it is important to choose an appropriately focused topic. Accordingly, a well presented essay will require a student to be familiar with a narrowly defined field of research within a subfield of population genetics. For instance, a field of research that may be appropriate for an essay is "The use of principal component analysis to understand human population structure" would be workable within the restrictions of the essay. Likewise, an essay on "The theoretical basis of principal component analysis in population genetics" would also be defined narrowly enough. You may be interested in a field of research that has a large literature, such as altruism and cooperation. A workable essay topic under these conditions may be "The insight and generality of Trivers (1971) model of reciprocal altruism relative to Hamilton's (1964) rule." Here I have focused the essay's topic on two classic papers.

# **Optional - Essay Outline**

Maximum of two pages, single spaced, 12 point Arial font, 2.5 cm margins (top, bottom, left right). Include tentative title.

# Grading rubric for essay (weighting in parenthesis)\*

Thoroughness and correctness of historical development (10 %) Insight with respect to the sequence of historical development (15%) Thoroughness and correctness of explanation of population genetic principles used and/or introduced (20%) Thoroughness and correctness relating principles to topics presented in BIOI \*3020

Thoroughness and correctness relating principles to topics presented in BIOL\*3020 (15%)

Correctness and insight with respect to historical and current state of the field (15%) Logical structure of document, paragraphs, and sentences (15%)

Format, punctuation, grammar and spelling (10%)

\*Each criterion will be graded on a 0-100 scale and then weighted for a final grade.

**Methods of Assessment:** As discussed under "Course Structure," there are several types of assessment used in the course: problem sets, data analyses, primary literature assignments, exams and an essay. There will be approximately six problem sets, five data analyses and three literature assignments. Problem sets, data analyses and literature assignments are assigned evenly throughout the course and typically cover course material from the previous week or two. Two mid-terms will occur, one covering Units 1.1 - 2.3 in mid-October and the other Units 2.4 - 5.3 in mid-November. The final exam is not comprehensive and covers Units 6.1 - 7.3. The optional essay outline is due in early November and the final essay is due during the last week of class.

A student's final grade is currently planned to be weighted such that performance on problem sets accounts for 15%, data analyses accounts for 10%, literature assignments accounts for 15%, low, medium and high exams accounts for 10%, 15% and 15%, respectively, and the final essay accounts for 20%. This weighting may be subject to change based on revisions to the course.

Grading standards follows the standards outlined in the Undergraduate Calendar, which is copied below:

**80 - 100 (A) Excellent.** An outstanding performance in which the student demonstrates a superior grasp of the subject matter, and an ability to go beyond the given material in a critical and constructive manner. The student demonstrates a high degree of creative and/or logical thinking, a superior ability to organize, to analyze, and to integrate ideas, and a thorough familiarity with the appropriate literature and techniques.

**70 - 79 (B) Good.** A more than adequate performance in which the student demonstrates a thorough grasp of the subject matter, and an ability to organize and examine the material in a critical and constructive manner. The student demonstrates a good understanding of the relevant issues and a familiarity with the appropriate literature and techniques.

**60 - 69 (C) Acceptable.** An adequate performance in which the student demonstrates a generally adequate grasp of the subject matter and a moderate ability to examine the material in a critical and constructive manner. The student displays an adequate understanding of the relevant issues, and a general familiarity with the appropriate literature and techniques.

**50 - 59 (D) Minimally Acceptable.** A barely adequate performance in which the student demonstrates a familiarity with the subject matter, but whose attempts to examine the material in a critical and constructive manner are only partially successful. The student displays some understanding of the relevant issues, and some familiarity with the appropriate literature and techniques.

0 - 49 (F) Fail. An inadequate performance.

#### Polices related to missed assignments and exams

Missed assignment(s): Late problem sets, data analyses, literature assignments and a late essay are not accepted and will be given a zero. Answers to problem sets are posted soon after they are handed in and answers to data analyses and literature assignments may be discussed in tutorial the following day.

An exception to the late assignment rules is if a student misses a due date as a result of an event that falls under an acceptable Academic Consideration or Accommodation (see General University Policies below). If this should occur, please contact the course instructor.

Missed exam: If an exam is to be missed the student is required to make a reasonable attempt to notify the instructor in advance. Furthermore, the reason for the missed exam must fall under an acceptable Academic Consideration or Accommodation (see General University Policies below). If a mid-term exam is missed, a make-up exam will be given and the student is expected to accommodate a scheduled time for the make-up exam that allows for it to co-occur with other students making up the exam. A missed final exam is made-up during the deferred period.

# **General University Policies**

# When You Cannot Meet a Course Requirement

When you find yourself unable to meet an in-course requirement because of illness or compassionate reasons, please advise the course instructor (or designated person, such as a teaching assistant) in writing, with your name, id#, and e-mail contact. See the undergraduate calendar for information on regulations and procedures for Academic Consideration:

http://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-ac.shtml

# Accessibility

The University of Guelph is committed to creating a barrier-free environment. Providing services for students is a shared responsibility among students, faculty and administrators. This relationship is based on respect of individual rights, the dignity of the individual and the University community's shared commitment to an open and supportive learning environment. Students requiring service or accommodation, whether due to an identified, ongoing disability or a short-term disability should contact the Centre for Students with Disabilities as soon as possible.

For more information, contact CSD at 519-824-4120 ext. 56208 or email csd@uoguelph.ca or see the website: http://www.csd.uoguelph.ca/csd/

# Academic Misconduct

The University of Guelph is committed to upholding the highest standards of academic integrity and it is the responsibility of all members of the University community – faculty, staff, and students – to be aware of what constitutes academic misconduct and to do as much as possible to prevent academic offences from occurring. University of Guelph students have the responsibility of abiding by the University's policy on academic misconduct regardless of their location of study; faculty, staff and students have the responsibility of supporting an environment that discourages misconduct. Students need to remain aware that instructors have access to and the right to use electronic and other means of detection.

Please note: Whether or not a student intended to commit academic misconduct is not relevant for a finding of guilt. Hurried or careless submission of assignments does not excuse students from responsibility for verifying the academic integrity of their work before submitting it. Students who are in any doubt as to whether an action on their part could be construed as an academic offence should consult with a faculty member or faculty advisor.

The Academic Misconduct Policy is detailed in the Undergraduate Calendar: http://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08amisconduct.shtml

#### E-mail Communication

As per university regulations, all students are required to check their <uoguelph.ca> email account regularly. E-mail is the official route of communication between the University and its students.

# Drop Date

The last date to drop one-semester courses, without academic penalty, is the 40<sup>th</sup> class day. To confirm the actual date, please see the schedule of dates in the Undergraduate Calendar. For regulations and procedures for Dropping Courses, see the Undergraduate Calendar:

http://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-drop.shtml

# Copies of out-of-class assignments

Keep paper and/or other reliable back-up copies of all out-of-class assignments: you may be asked to resubmit work at any time.

# Recording of Materials

Presentations which are made in relation to course work—including lectures—cannot be recorded or copied without the permission of the presenter, whether the instructor, a classmate or guest lecturer. Material recorded with permission is restricted to use for that course unless further permission is granted.

# Resources

The Academic Calendars are the source of information about the University of Guelph's

procedures, policies and regulations which apply to undergraduate, graduate and diploma programs:

http://www.uoguelph.ca/registrar/calendars/index.cfm?index

# Campus Resources

The Academic Calendar is the source of information about the University of Guelph's procedures, policies and regulations which apply to undergraduate, graduate and diploma programs:

http://www.uoguelph.ca/registrar/calendars/index.cfm?index

If you are concerned about any aspect of your academic program:

 make an appointment with a program counsellor in your degree program. <u>http://www.bsc.uoguelph.ca/index.shtml</u> or <u>https://www.uoguelph.ca/uaic/programcounsellors</u>

If you are struggling to succeed academically:

 There are numerous academic resources offered by the Learning Commons including, Supported Learning Groups for a variety of courses, workshops related to time management, taking multiple choice exams, and general study skills. You can also set up individualized appointments with a learning specialist. http://www.learningcommons.uoguelph.ca/

If you are struggling with personal or health issues:

- Counselling services offers individualized appointments to help students work through personal struggles that may be impacting their academic performance. <u>https://www.uoguelph.ca/counselling/</u>
- Student Health Services is located on campus and is available to provide medical attention.
  <a href="https://www.uoguelph.ca/studenthealthservices/clinic">https://www.uoguelph.ca/studenthealthservices/clinic</a>
- For support related to stress and anxiety, besides Health Services and Counselling Services, Kathy Somers runs training workshops and oneon-one sessions related to stress management and high performance situations. <u>http://www.uoguelph.ca/~ksomers/</u>

If you have a documented disability or think you may have a disability:

The Centre for Students with Disabilities (CSD) can provide services and support for students with a documented learning or physical disability. They can also provide information about how to be tested for a learning disability. For more information, including how to register with the centre please see: <u>https://www.uoguelph.ca/csd/</u>