

BIO 463 - Genetics of Human Disease

Winter 2016

Section 001: 2021 LSB on M W from 9:00 am - 10:15 am

Instructor/TA Info

Instructor Information

Name: Keoni Kauwe

Office Location: 4146 LSB

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Office Hours: Only By Appointment

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TA Information

Name: Ariel Hippen

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Course Information

Description

In this course we will examine the application of genetics to the understanding and treatment of human disease. We will also cover basic methods for design, analysis, interpretation and follow-up of linkage, candidate gene, genome-wide association and whole genome, whole exome studies. In addition, we will examine the various methods for understanding the functional consequences of genetic variants, the role of model organisms, the use of genetic information for treatment of disease and ethical considerations that arise from our advancing knowledge of the genetics of human disease.

Prerequisites

PWS 340 Genetics

Grading Scale

Grades	Percent
A	93%
A-	90%
B+	87%
B	83%
B-	80%

C+	77%
C	73%
C-	70%
D+	67%
D	63%
D-	60%
E	0%

Learning Outcomes

Genetic Variants

Understand and explain the different types of genetic variants, how they can be typed and how they can be used to understand the genetic architecture of human disease.

Linkage Disequilibrium

Understand linkage disequilibrium in the human genome and the role it plays in the search for genetic risk factors for human disease

Basic Statistics and Software

Understand and use basic statistics and software for analyzing the correlation between genetic markers and risk for disease.

Model Organisms and the Genetics of Human Disease

Understand and explain how model organisms contribute to our understanding of the genetics of human disease.

Acquiring and Presenting Information about Genetics of a Human

Demonstrate the ability to obtain, aggregate and present information about the current knowledge of the genetics of a human disease.

Grading Policy

Late Work:

I will not accept late work unless arrangements have been made PRIOR to the due date.

Grading:

If you have questions or concerns about a grade on an assignment or exam you have 2 school days after the grade has been posted on blackboard to discuss them with me. I WILL NOT discuss or change grades after that time.

Manuscript Grading:

A typical "A" paper presents a compelling argument, is well written and well organized. The introduction will provide an excellent, concise summary of the current literature. It will frame the question in a meaningful way, making the importance of the research clear. The methods section will provide a clear description of all of the samples, software and methods used to generate and

analyze the data, making it possible for the procedures to be replicated independently. The results section will provide clear statements of the outcomes of the experiments, including tables and figures that effectively communicate the information when necessary. Finally, the discussion/conclusions will make clear and justifiable conclusion from the data, linking it to the overall importance of the question as outlined in the discussion. In addition this section will clearly state the limitations of the experiment and its conclusions.

Participation Policy

This is a small, 400 level course. Class participation is important and will greatly affect your grade in the form of in class quizzes, assignments and a subjective participation grade. Students are expected to read the assigned materials BEFORE class and be prepared to participate in class discussions about those materials. Lectures, discussions and activities will be designed to extend the concepts presented in the reading assignments, not repeat them.

Attendance Policy

Attendance is important in this class. Makeup of missed quizzes and assignments will only be allowed under special circumstances. I am happy to make accommodations for planned absences such as Graduate or Professional school interviews and other important events.

Assignments

Assignment Description

Databases worksheet

Due: Monday, Jan 25 at 8:59 am

This is an in-class, group assignment. While you will work together, each person must fill out the worksheet found in the content tab and email it to kauwebio463@gmail.com by the next class period.

GWAS QC Lab Report

Due: Monday, Feb 08 at 8:59 am

Methods section describing QC criteria and results section summarizing the properties of the cleaned dataset.

GWAS Results and Follow-up Lab Report

Due: Wednesday, Feb 10 at 8:59 pm

Methods, Results and Discussion sections describing the GWAS analysis performed in lab.

Exam #1

Due: Wednesday, Feb 17 at 12:05 am

Exam #1 covers content through Feb 19th.

WGS causal variant identification Lab Report

Due: Monday, Feb 29 at 8:59 am

Methods and results sections
Sanger Sequence lab report

Due: Monday, Mar 14 at 8:59 am

Paragraph describing the identified variant and what is known about its possible function.

Presentation

Due: Wednesday, Mar 30 at 8:59 pm

This is in INDIVIDUAL assignment, not a group assignment.

You will choose a trait with a complex genetic basis and provide two manuscripts (one a review of the genetics of the disease, another an example of current research in the genetics of the disease) upon which your presentation will be based.

You will give a 12 minute presentation to the class followed by a 3 minute question period. Your presentation should include an overview of the disease, disease genetics and current progress in understanding the genetics of disease as well as treatment strategies. You will be graded using the following rubric.

Bio463 Presentation Rubric.docx [Download](#)

Reading Quiz #3

Due: Monday, Apr 04 at 11:59 pm

Attendance quiz on 3/23 and 3/30.

Reading Quiz #2

Due: Monday, Apr 04 at 11:59 pm

Reading Quiz #5

Due: Monday, Apr 04 at 11:59 pm

Attendance on 4/4.

Reading Quiz #4

Due: Monday, Apr 04 at 11:59 pm

Reading Quiz #1

Due: Monday, Apr 04 at 11:59 pm

Evaluation Extra Credit

Due: Thursday, Apr 14 at 11:59 pm

Participation

Due: Thursday, Apr 14 at 11:59 pm

Assessment will be based on attendance, participation in class meetings, contributions to the group project and labs and participation in student presentations (attentiveness, asking questions, etc.)

Group Manuscript

Due: Thursday, Apr 14 at 11:59 pm

Full manuscript in PLoS ONE format describing the WGS findings.

See formatting guidelines here: <http://www.plosone.org/static/guidelines>

Instructions for writing a paper (slides from class) Writing and Reviewing Papers.pdf Download

Extra Credit 1

Due: Thursday, Apr 14 at 11:59 pm

Reporting on one of the seminars or presentations about human genetics in campus

Final Exam

Due: Monday, Apr 18 at 7:00 am

Schedule

Date	Column 1	Column 2
M Jan 04 Monday	First Day of Winter Semester (01/04/2016 - 04/12/2016)	Syllabus
	Syllabus, Group	

	Assignments, Overview	
W Jan 06 Wednesday	Types of genetic variation	Ch 13
M Jan 11 Monday	Heritability, Segregation Analysis, Linkage Analysis	Ch 3, Ch 14
W Jan 13 Wednesday	Linkage Disequilibrium	Manuscript Download
M Jan 18 Monday	Martin Luther King Jr Day	Visit and explore: NCBI, HapMap. Ensembl, 1000Genomes, UCSC Genome Browser websites
W Jan 20 Wednesday	Lab: Databases that facilitate the study of human disease genetics	Ch 15, Manuscript Download
M Jan 25 Monday	Statistics of Association analysis	Manuscripts Download; Download
W Jan 27 Wednesday	Genome-wide association studies, CD/CV	Ch 16;
M Feb 01 Monday	Lab: GWAS Quality Control	PLINK Tutorial
W Feb 03 Wednesday	Presentations	Manuscripts Download, Download, Download
M Feb 08 Monday	GWAS findings, limitations	
W Feb 10 Wednesday	Lab: GWAS results and follow-up	
	Guest Lecture: Ariel Hippen	
M Feb 15 Monday	Presidents Day	

T Feb 16 Tuesday	Monday Instruction	
	Review for Exam 1	
W Feb 17 Wednesday	Exam #1	
M Feb 22 Monday	Introduction to whole genome based approaches, alignment, variant calling, databases	Ch 12, Manuscript Download, Download
	Guest Lecture: Mark Ebbert	
W Feb 24 Wednesday	Lab: WGS causal variant identification	Manuscripts Download, Download
	Guest Lecture: Mark Ebbert	
M Feb 29 Monday	Presentations	
W Mar 02 Wednesday	Test Discussion / Project Data	
M Mar 07 Monday	Presentations	
W Mar 09 Wednesday	Lab: Rare variants in Sanger sequence data from TREM2	Download Geneious or sequencher demos and explore the software. Presentation will be done using sequencher.
M Mar 14 Monday	Presentations	
T Mar 15 Tuesday	Withdraw Deadline (Full Semester)	

W Mar 16 Wednesday	Lab: Group Projects	
M Mar 21 Monday	Presentations	
W Mar 23 Wednesday	Presentations	Ch 20
M Mar 28 Monday	Genetics and human disease treatment	1-s2.0-S1474442211700394- main.pdf Download Ch 18, Ch 21
W Mar 30 Wednesday	Presentations	
M Apr 04 Monday	Writing/Reviewing scientific manuscripts	Ch 19
W Apr 06 Wednesday	Model organisms and human disease genetics	
	Guest Lecture: Dr. Arminda Suli	
M Apr 11 Monday	Genes and Behavior, ethics, summary	
T Apr 12 Tuesday	Last Day of Winter Semester (01/04/2016 - 04/12/2016)	
W Apr 13 Wednesday	First Day of Winter Exam Preparation (04/13/2016 - 04/14/2016)	
Th Apr 14 Thursday	Last Day of Winter Exam Preparation (04/13/2016 - 04/14/2016)	

M Apr 18 Monday	Final Exam: 2021 LSB 11:00am - 2:00pm
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W Apr 20 Wednesday

University Policies

Honor Code

In keeping with the principles of the BYU Honor Code, students are expected to be honest in all of their academic work. Academic honesty means, most fundamentally, that any work you present as your own must in fact be your own work and not that of another. Violations of this principle may result in a failing grade in the course and additional disciplinary action by the university. Students are also expected to adhere to the Dress and Grooming Standards. Adherence demonstrates respect for yourself and others and ensures an effective learning and working environment. It is the university's expectation, and every instructor's expectation in class, that each student will abide by all Honor Code standards. Please call the Honor Code Office at 422-2847 if you have questions about those standards.

Sexual Misconduct

As required by Title IX of the Education Amendments of 1972, the university prohibits sex discrimination against any participant in its education programs or activities. Title IX also prohibits sexual harassment-including sexual violence-committed by or against students, university employees, and visitors to campus. As outlined in university policy, sexual harassment, dating violence, domestic violence, sexual assault, and stalking are considered forms of "Sexual Misconduct" prohibited by the university.

University policy requires any university employee in a teaching, managerial, or supervisory role to report incidents of sexual misconduct that come to their attention through various forms including face-to-face conversation, a written class assignment or paper, class discussion, email, text, or social media post. If you encounter Sexual Misconduct, please contact the Title IX Coordinator at t9coordinator@byu.edu or 801-422-2130 or Ethics Point at <https://titleix.byu.edu/report> or 1-888-238-1062 (24-hours). Additional information about Title IX and resources available to you can be found at <http://titleix.byu.edu>.

Student Disability

Brigham Young University is committed to providing a working and learning atmosphere that reasonably accommodates qualified persons with disabilities. If

you have any disability which may impair your ability to complete this course successfully, please contact the University Accessibility Center (UAC), 2170 WSC or 422-2767. Reasonable academic accommodations are reviewed for all students who have qualified, documented disabilities. The UAC can also assess students for learning, attention, and emotional concerns. Services are coordinated with the student and instructor by the UAC. If you need assistance or if you feel you have been unlawfully discriminated against on the basis of disability, you may seek resolution through established grievance policy and procedures by contacting the Equal Employment Office at 422-5895, D-285 ASB.

Academic Honesty

The first injunction of the Honor Code is the call to "be honest." Students come to the university not only to improve their minds, gain knowledge, and develop skills that will assist them in their life's work, but also to build character.

"President David O. McKay taught that character is the highest aim of education" (The Aims of a BYU Education, p.6). It is the purpose of the BYU Academic Honesty Policy to assist in fulfilling that aim. BYU students should seek to be totally honest in their dealings with others. They should complete their own work and be evaluated based upon that work. They should avoid academic dishonesty and misconduct in all its forms, including but not limited to plagiarism, fabrication or falsification, cheating, and other academic misconduct.