BIO 463 - Genetics of Human Disease

Winter 2018

Section 001: C261 ESC on T Th 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: Tue 2:30pm-3:00pm
Or By Appointment
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Name: Marie Emmett
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TA Information

Name: Josue Gonzalez Murcia
Office Location: 4049 LSB
Office Hours: Thu 12:00pm-1:30pm
Or By Appointment
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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. Final 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: Tuesday, Jan 30 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: Thursday, 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: Thursday, Feb 15 at 8:59 pm

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

Exam #1

Due: Tuesday, Feb 27 at 12:05 am

Exam #1 covers content through Feb 19th.

WGS causal variant identification Lab Report

Due: Tuesday, Mar 13 at 8:59 am

Methods and results sections

Sanger Sequence lab report

Due: Tuesday, Mar 20 at 8:59 am

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

Presentation

Due: Thursday, Apr 12 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 (~2min), overview disease genetics (~3min) and current progress in understanding the genetics of disease (~6min) as well as current treatment strategies (~1min). You will be graded using the following rubric.
Bio463 Presentation Rubric.docx Download
follow the link to pick a date and disease to present:

https://docs.google.com/spreadsheets/d/1pLRcHoMzJFPzH-BjXfNiL7__HS9sgBW203OEg5XG7h8/edit?usp=sharing

Final Exam

Due: Monday, Apr 16 at 7:00 am

Reading Quiz #3

Due: Tuesday, Apr 17 at 11:59 pm

Attendance quiz on 3/23 and 3/30.

Reading Quiz #1

Due: Tuesday, Apr 17 at 11:59 pm

Reading Quiz #5

Due: Tuesday, Apr 17 at 11:59 pm

Attendance on 4/4.

Reading Quiz #4

Due: Tuesday, Apr 17 at 11:59 pm

Reading Quiz #2

Due: Tuesday, Apr 17 at 11:59 pm

Evaluation Extra Credit

Due: Wednesday, Apr 18 at 11:59 pm

Participation

Due: Wednesday, Apr 18 at 11:59 pm

Assessent 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: Wednesday, Apr 18 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

Schedule

Date	Topic	Assignments	Readings/Pre-class Preparation
T Jan 09 Tuesday	Syllabus, Group Assignments, Overview		Syllabus
Th Jan 11 Thursday	Types of genetic variation		Ch 13 Genetic of complex diseases Download
M Jan 15 Monday	**Martin Luther King Jr Day**		
T Jan 16 Tuesday	Lab: Databases that facilitate the study of human disease genetics		Visit and explore: NCBI, HapMap. Ensembl, 100 Browser websites, genecard
Th Jan 18 Thursday	Heritability, Segregation Analysis, Linkage Analysis		Ch 3, Ch 14
T Jan 23 Tuesday	Linkage Disequilibrium		Manuscript Download
Th Jan 25 Thursday	Statistics for Association analysis		Ch 15, Manuscript Download
T Jan 30 Tuesday	Genome-wide association studies, genetic architecture of complex traits	**Databases worksheet**	Ch 16; Manuscripts Download; Download
Th Feb	Lab: GWAS		Plink Tutorial - Get familiar with PLINK. Read the

01 Thursday	Quality Control		(Item 1 on the left side of the screen), and comm the left side of the screen)
T Feb 06 Tuesday	GWAS findings, limitations, Post-GWAS use of association study data and samples		10_years_of_GWAS_discovery_review.pdf Down
Th Feb 08 Thursday	Presentations	**GWAS QC Lab Report**	Genetics_T1D_Review.pdf Download T1D_Current_Research.pdf Download Obesity_current_research.pdf Download Obesity_review.pdf Download Parkinsons_review.pdf Download Parkinsons_association.pdf Download hyperten hypertension_current_research.pdf Download
T Feb 13 Tuesday	Lab: GWAS results and follow-up		gwaspart2_lab3.pptx Download
Th Feb 15 Thursday	Presentations	**GWAS Results and Follow-up Lab Report**	asthma_current_research.pdf Download asthma_review.pdf Download celiac's_current_research.pdf Download celiac's_review.pdf Download Intelligence Current Research.pdf Download Intelligence Review.pdf Download angelman_current_research.pdf Download angelman_review.pdf Download Arthritis_current research.PDF Download arthritis_review.pdf Download
T Feb 20 Tuesday	**Monday Instruction**		

Th Feb 22 Thursday	Review Session		
T Feb 27 Tuesday	Exam #1		
Th Mar 01 Thursday	Test Discussion / Project Data		Gall Bladder Disease IRB Application Materials https://www.dropbox.com/sh/yirhz6f4pp00pfq/AA
T Mar 06 Tuesday	Introduction to whole genome based approaches, alignment, variant calling, databases Guest Lecture: Dr. Perry Ridge		Ch 12, Manuscript Download, Download
Th Mar 08 Thursday	Lab: WGS causal variant identification Guest Lecture: Dr. Perry Ridge		Manuscripts Download, Download
T Mar 13 Tuesday	Lab: Rare variants in Sanger sequence data from TREM2	**WGS causal variant identification Lab Report**	Download and activate Geneious Trial. Download Sanger Sequence data. BIO 463 Sanger Data.zip Download Get familiar with Geneious software. Sanger Sequence Analysis Lab Assignment.doc

Th Mar 15 Thursday	Presentations		
T Mar 20 Tuesday	Presentations	**Sanger Sequence lab report**	
Th Mar 22 Thursday	Lab: Group Project planning and work		
T Mar 27 Tuesday	Molecular techniques in human genetics: model organisms, gene editing, etc. Guest Lecture: Dr. Julianne Grose		Ch 20
Th Mar 29 Thursday	Genetics and human disease treatment Guest Lecture: Josue Gonzalez		Ch 19
T Apr 03 Tuesday	Presentations		
Th Apr 05 Thursday	Presentations		

T Apr 10 Tuesday	Presentations, Group Project Work Session		
Th Apr 12 Thursday	Writing/Reviewing scientific manuscripts Guest Lecture: TBA	**Presentation**	Ch 21
T Apr 17 Tuesday	Genes and Behavior, ethics, summary		
Th Apr 19 Thursday	**Winter Exam Preparation (04/19/2018 - 04/19/2018)**		
F Apr 20 Friday	**First Day of Winter Final Exams (04/20/2018 - 04/25/2018)**		

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

In accordance with Title IX of the Education Amendments of 1972, Brigham Young University prohibits unlawful sex discrimination against any participant in its education programs or activities. The university 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 all university employees in a teaching, managerial, or supervisory role to report all incidents of Sexual Misconduct that come to their attention in any way, including but not limited to face-to-face conversations, a written class assignment or paper, class discussion, email, text, or social media post. Incidents of Sexual Misconduct should be reported to the Title IX Coordinator at t9coordinator@byu.edu or (801) 422-8692. Reports may also be submitted through EthicsPoint at https://titleix.byu.edu/report or 1-888-238-1062 (24-hours a day).

BYU offers confidential resources for those affected by Sexual Misconduct, including the university's Victim Advocate, as well as a number of non-confidential resources and services that may be helpful. Additional information about Title IX, the university's Sexual Misconduct Policy, reporting requirements, and resources can be found at http://titleix.byu.edu or by contacting the university's Title IX Coordinator.

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.