

Models Systems of Genetic Disorders - GGS690/GGS490/STH299
Syllabus, Course Objectives and Policies
Fall Semester 2018

Credit hours: 3.0

Room: Heritage Hall room 227 (HHB 227)

Dates & Times: MWF 11:15 - 12:05

Instructor: Dr. Camerron M. Crowder

Office: HHB 521

Email: Camerron@uab.edu

Office hours: M 10:00-11:00 am & 12:15-1:15 pm

Course Objectives: This course will focus on the underlying genetic factors associated with disorders and disease states in humans and the importance of animal models in research. Through reading and reviewing scientific literature, we will explore the usage of animal models to understand gene function as it relates to genetic disorders. Topics covered, but not limited to, include neuromuscular disease, developmental abnormalities and cancer. All materials will be posted on Canvas and you are expected to come to class having read assigned materials and ready for discussion. Throughout the term we will be hearing from UAB faculty, as guest lecturers, to learn how different model systems are utilized in human disease research. To deepen our understanding of these models we will be touring research labs and facilities.

Course Goals:

- Understand why model systems are useful in investigating human genetic disorders
- Conceptualize the genetic relationship amongst eukaryotic organisms and concepts relating to genetic conservation
- Examine the benefits and considerations for working with various model organisms
- Determine how to ask a testable research question utilizing animal models
- Develop clear written and oral communication skills
- Improve skills of working on a team and reflecting on your own strengths and weaknesses

Student expectations:

- Arrive to class on time and ready to learn
- Read all assigned materials and complete all homework, prior to class
- Ask questions, share your thoughts on readings and be a willing participant in discussions and team work
- Respect yourself, your peers and all faculty

Recommended textbook: Molecular Biology of the Cell, 6th ed., Alberts *et al.* Garland Sciences

Grading:

Assignments	Points
Journal club assignments (10 pts. each x 6)	60
Individual paper (not group work) on Nobel laureate	10
Individual paper (not group work) on non-traditional model	10
Group presentation on Nobel laureate* (rubric)	15
Group presentation on non-traditional model* (rubric)	15
Quizzes (10 pts. each x 6)	60
Models in action - lab tour assignments (10 pts. x 3)	30
Models in action - lab tour presentation (20 pts. x 1)	15
Midterm exam	50
Final exam	100
Individual paper (not group work) on genetic disorder proposal (rubric)	15
Group presentation on genetic disorder proposal* (rubric)	15
Total course points	395

* Assignments graded as a group

Additional assignments for 690 level

Journal club discussion submission (4 pts. x 6)	24
Midterm exam take-home questions (30 pts)	30
Final exam take-home questions (30 pts)	30
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Journal club assignments: Throughout the semester you will be expected to read in depth about a variety of topics relating to genetic disorders and disease. Reading assignments will be posted via Canvas and you will need to complete all readings prior to the beginning of class. Journal club assignments (6 assignments total - 10 pts each) will be **due prior to class** and submitted on Canvas or printed out and brought with you to class. As a class and in small groups we will discuss the readings to gain a deeper understanding of the material, for participation it is required that you have completed readings and assignments.

Nobel laureate and non-traditional model paper and group presentation: You will work in small groups on a presentation and paper assignment on both the life, research, and animal models utilized by a particular Nobel laureate and the research, benefits and compensations of working with a particular non-traditional animal model. You will work on teams of 2-3 students per assignment on a topic chosen by your group from a pre-selected group of topics. As a group, you will design a 5-10 minute presentation that you will present to the class on a date listed on the syllabus and be graded (15 pts) using a rubric that will be available to you on Canvas. Separate from the presentation, you will write a paper (essay style) on your topic (2-3 pages in length) and turn it in individually (written on your own, not as a group) (10 pts).

Quizzes: Six 10-point quizzes will be administered at the beginning of the class throughout the semester. Quizzes are intended to assess your learning and to encourage you to keep up and study the materials in preparation for exams. Quizzes are all short answer and you will be required to write out terms, disorders or genetic concepts covered in lectures, speaker presentations and group work assignments.

Lab tour assignment and group presentation: In order to gain a deeper understanding as to what it is like to conduct research on various model systems we will be visiting the zebrafish and mice housing facilities. In addition to facility visits you will be touring and meeting graduate students, postdoc and faculty of two research labs. During these facility and lab visits you will be expected to ask questions and obtain information regarding various animal models. You will answer questions for a short worksheet assignment (15 pts per assignment) that you will turn in to me. For the lab tours, the class will be separated into two groups that will visit two different labs and make a presentation to present to the entire class on the individual lab your group toured.

Genetic disorder research presentation and individual paper: Working in small groups (2-3 students) you will be expected to pick a human genetic disorder from a selected group of disorders and develop a mini-research proposal. In a group, you will work together to identify a research question associated with your genetic disorder and write a persuasive argument as to which model system is ideal for investigating your research question. Clearly defining the benefits, limitations and range of testable hypotheses of your model system and introducing your disease and the importance of your research question will be expected. Together as a group you will present your disorder, model, question and testable hypotheses to the class and individually write up your work into a paper (3-4 pages).

Exams: A midterm (50 points) and final (100 pts) cumulative exam will be held in class at the middle and end of the semester. Exam will include multiple choice, short answer, long answer and drawing/labeling type questions. A review will be held in class prior to each exam where you will be expected to ask questions, draw out and label cellular and molecular events and discuss topics openly to prepare for the exams.

Letter Grade: In general, 90-100%=A; 80-89%=B; 70-79%=C; 60-69%=D; <60%=F

Missed classes/make-up assignments: If it is absolutely necessary or you are too ill to be present in class, you should notify me immediately about your absence, prior to the start of the class. If you have an acceptable excuse and have notified, me prior to your absence, we will work out the best option for you to make up an assignment or turn in work at a later time. All speakers and tours will not be available at a later time.

Audio recording, videotaping or photographing class lectures: In general, any form of audio and/or video capture of lectures is not permitted except with written consent of the instructor (with justification).

Electronic Devices in the Classroom: All cell phones should be silenced and removed from the desk top at the beginning of class. Utilization of laptops, tablets, or other electronic devices for classroom related activities is permitted. Use of these devices and other devices (cell phones, iPod's, etc.) for other activities such as searching the internet, checking email, or reviewing material unrelated to class is not permitted during class.

Class Attendance: Attendance at all lectures is expected and highly recommended. The majority of test and quiz questions will be derived from material discussed during lectures.

Academic Misconduct: Students are expected to maintain an acceptable quality of academic performance and to exhibit appropriate conduct. Students are expected to conduct themselves in a manner similar to accepted standards for practicing health care professionals. Academic misconduct may include, but is not necessarily limited to, acts such as plagiarism, cheating, misrepresentation, fabrication or giving or receiving unauthorized aid in tests, examinations, or other assigned work, and will be subject to disciplinary action. Any act of dishonesty in academic work constitutes academic misconduct. Academic misconduct will result in a grade of zero on the assignment/exam and may result in disciplinary action. A student who feels he or she has been unfairly disciplined should contact the program director or department chair to request a review of the disciplinary decision. A more detailed description of the *Grievance Procedures for Violations of Academic Standards* is available from the Office of the Associate Dean for Academic and Student Affairs.

Special Needs: If you have registered with the UAB DSS, please let me know ASAP if you have any concerns regarding the availability of accommodations that may be necessary, and to complete any necessary paperwork. You are expected to complete the exams on the scheduled exam dates. DSS contact email: dss@uab.edu

TITLE IX: The University of Alabama at Birmingham is committed to providing an environment that is free of bias, discrimination, and harassment. If you have been the victim of sexual discrimination, harassment, misconduct, or assault we encourage you to report the incident. UAB provides several avenues for reporting. For more information about Title IX, policy, reporting, protections, resources and supports, please visit [UAB's Title IX Policy](#) and [UAB's Equal Opportunity and Anti-Harassment Policy](#)

Date	Lecture Topic	Readings	Assignments due
Aug. 27 M	Introduction, syllabus	Muller (2010)	
Aug. 29 W	L1- Genomic sequencing	Katz (2016), Leonelli (2013)	NL topic & team
Aug. 31 F	<i>Journal club discussion 1</i>		JC assign. 1 due
Sept. 3 M	Labor Day - no class		
Sept. 5 W	L2- Gene conservation & homology		
Sept. 7 F	L3- Models of genetic disorders + NL team		Quiz 1 + NL team meet
Sept. 10 M	L4- Genetic & genomic research methods I	Review paper on NMD	NL paper due
Sept. 12 W	Nobel laureates presentations + GD topic		NL presentations
Sept. 14 F	L5- Genetic & genomic research methods II		
Sept. 17 M	L6- Disease Module I - Neuromuscular disease		Quiz 2
Sept. 19 W	Speaker - Dr. Michael Miller - <i>C. elegans</i>	<i>C. elegans</i> model	
Sept. 21 F	L7- Disease Module I - Neuromuscular disease	Journal club 2 paper	GD team meet
Sept. 24 M	L8- Disease Module I - Neuromuscular disease		Quiz 3
Sept. 26 W	<i>Journal club discussion 2</i>		JC assign. 2
Sept. 28 F	Speaker - Dr. Matt Alexander - <i>D. rerio</i>	Developmental review	
Oct. 1 M	Tour of zebrafish facility - Dr. Susan Farmer	Zebrafish facility tour	
Oct. 3 W	L9- Disease Module II - Development defects	Neural tube paper	Tour assign. 1 due
Oct. 5 F	L10- Disease Module II - Development defects		GD team meet
Oct. 8 M	Speaker - Dr. Heather Ray - <i>X. laevis</i>	<i>X. laevis</i> model	
Oct. 10 W	L11- Disease Module II - Developmental defects	Journal club 3 paper	Quiz 4
Oct. 12 F	L12- Disease Module II - Developmental defects		
Oct. 15 M	<i>Journal club discussion 3</i>		JC3 assign. 3
Oct. 17 W	Review for midterm + LT topic & team meet	<i>D. melanogaster</i> model	LT team meet
Oct. 19 F	Lab tours - Riddle lab and Miller lab	<i>D.m.</i> & <i>C.e.</i> lab tours	
Oct. 22 M	MIDTERM EXAM	Cancer review article	Midterm exam
Oct. 24 W	L13- Disease Module III - Cancer + LT teams		LT team meet
Oct. 26 F	L14- Disease Module III - Cancer		Quiz 5
Oct. 29 M	<i>Journal club discussion</i>		JC assign. 4
Oct. 31 W	Speaker - Dr. John Parant - <i>D. rerio</i> & <i>M.mus.</i>	<i>Danio rerio</i> model	
Nov. 2 F	Lab tour presentations + NTM topic + team		LT presentations
Nov. 5 M	L15- Disease Module III - Cancer + NTM team		NTM topic & team
Nov. 7 W	L16- Disease Module IV - Cancer		Quiz 6
Nov. 9 F	<i>Journal club discussion 5</i>		JC assign. 5
Nov. 12 M	Speaker - Dr. Karina Yoon - <i>M. musculus</i>	<i>M. musculus</i> model	
Nov. 14 W	Tour of the mice facility - Dr. Susan Farmer	<i>M. musculus</i> facility tour	
Nov. 16 F	Non-traditional models presentations	Journal club 6 paper	NTM presentations
Nov. 19-25	Thanksgiving Break - no class		
Nov. 26 M	L17- Stem cell research and applications		GD team meet
Nov. 28 W	<i>Journal club discussion 6</i>		JC assign. 6 due
Nov. 30 F	L18- Future of animal models of disease		Quiz 7
Dec. 3 M	Genetic disorder presentations		GD Presentations
Dec. 5 W	Genetic disorder presentations		GD paper due
Dec. 7 F	Review for final exam		
Dec. 10-14	Final Exam		Final exam

Team codes: **NL** = Nobel laureate, **GD** = Genetic disorder, **LT** = Lab tour, **NTM** = Non-traditional model