

BI 403/603: Medical Genetics

Spring 2017

Basic Information

Course: BI403/603	Section: 01	Medical Genetics
Credit Hours: 3	Prerequisites: BI283, CH341	
Semester: Spring 2017		
Time: 11:00 AM – 12:15 PM	Days: T R	
Location: MG 217		
Instructor Name: Rebecca Kurzhals		
Office Location: RH 218 (Lab RH215)		
Office Hours: Monday 11:00 AM – 1:00 PM, Tuesday 9:00 – 10:30 AM, and by appointment		
Office Phone Number: 573-651-2361		
Email Address: rkurzhals@semo.edu		
Website: http://learning.semo.edu		
Grade Book: http://learning.semo.edu		

Class Description

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This course will introduce students to topics, principles, and methods in genetics as it relates to inherited human genetic disorders. We will examine the biochemical, clinical, and molecular basis of several human genetic disorders and examine the importance of model organisms to better understand human disease. Critical reading of current scientific literature forms the basis of the discussions.

Student Learning Objectives:

1. Students will be able to interpret results from papers in the primary genetics literature.
2. Student will be able to summarize a human genetic disorder including how the mutation affects phenotype at the molecular level using a primary literature, online databases, and textbooks.
3. Students will be able to demonstrate knowledge of key terms used in genetics research.

Materials and Resources

Material	Location
Textbook	There is no textbook for this course.
Online resources	See course web site
Handouts	In class and at the course web page.

Expectations and Policies

Attendance

I do not require attendance, however there is no make-up for missed in-class assignments, you are responsible for all material presented in class. For university-sanctioned absences, I will schedule a make-up for any missed assignments with you.

The official University policy states: *Students are expected to attend all classes and to complete all assignments for courses in which they are enrolled. An absence does not relieve the student of the responsibility to complete all assignments. If an absence is associated with a university-sanctioned activity, the instructor will provide an opportunity for assignment make-up. However, it is the instructor's decision to provide, or not to provide, make-up work related to absences for any other reason.*

Academic Honesty

A link to the official university policy is below. If you are caught cheating or plagiarizing you will receive a zero on the exam or assignment. It will be brought to the attention of the Chair of the Biology Department and we will keep a record of the offense. Any subsequent offenses will be referred to the Judicial Coordinator.

<http://www.semo.edu/pdf/stuconduct-code-conduct.pdf>

<http://www.semo.edu/facultysenate/handbook/5d.html>

Civility

Every student at Southeast is obligated at all times to assume responsibility for his/her actions, to respect constituted authority, to be truthful, and to respect the rights of others, as to respect private and public property. In their academic activities, students are expected to maintain high standards of honesty and integrity and abide by the University's Policy on Academic Honesty. Alleged violations of the Code of Student Conduct are adjudicated in accordance with the established procedures of the judicial system.

Disabilities

Southeast Missouri State University and Disability Support Services remain committed to making every reasonable educational accommodation for students with disabilities. Many services and accommodations which aid a student's educational experience are available for students with various types of disabilities. It is the student's responsibility to contact Disability Support Services to become registered as a student with a disability in order to have accommodations implemented. Accommodations are implemented on a case by case basis. For more information visit the following site: www.semo.edu/lapdss or contact Disability Support Services at 573-651-2273

Technology

This course requires the use of Southeast's Learning Management System, Moodle. You can access this through <http://learning2.semo.edu>

Problems with the instructor of course:

Questions, comments, or requests regarding this course or program should be taken to your instructor. Unanswered questions or unresolved issues involving this class may be taken to Dr. Jim Champine, Biology Department Chairperson.

Undergraduate Student Grading

Grading Scale

100% - 90%	A
89% - 80%	B
79% - 70%	C
69% - 60%	D
59% and lower	F

Graduate Student Grading

Grading Scale

100% - 90%	A
89% - 80%	B
79% - 70%	C
69% and lower	F

Types of Assessment Undergraduate Students	Percentage of Grade
Exams	30%
Literature Review	30%
Scientific paper analyses, presentations, problem sets, in-class inquires	40%

Types of Assessment Graduate Students	Percentage of Grade
Exams	30%
Literature Review	30%
Scientific paper analyses, presentations, problem sets, in-class inquires, and Journal Club presentation	40%

Basic Breakdown of Assessments

Exams

There will be 3 exams. One following each topic and will be based on readings and material presented in class. Exams will be announced at least one week in advance.

Literature Review

Each student will write a Literature Review about an inherited genetic disease disorder of the student's choice with instructor approval. Early in the semester each student will choose a topic and meet a series of benchmarks throughout the semester. The literature review will be based information found in primary literature, review articles, online databases, policy review, etc. Students will present their findings to the students in an oral presentation and a final paper. All points relating to the literature review are worth 30% of your final grade. More information to follow.

Scientific paper analysis and presentations

Throughout the semester we will be reading primary genetics literature and review articles. You will learn to read and analyze the data presented in papers. You are responsible for reading and working through the papers in order to prepare for class discussion. See separate hand-out for more details.

In addition to the final paper analysis, each student will be responsible for presenting one figure from the paper to the class. This will give students a chance to present part of an article in a way that is common and expected in graduate school and in medical school.

Problem sets and in-class inquiries

I may occasionally provide a set of questions to accompany an assigned reading. The questions are meant to help you focus on the important concepts and/or facts in the reading. These are meant to increase retention and learning and to aid in exam preparation.

Late Assignments and make-ups

There are no make-ups for missed assignments except for University sanctioned events and documented illnesses. Late assignments will receive a **20% deduction of points** for each day that they are late. The schedule of homework due dates and exam will be provided in advance and are available on Moodle. If you will be out of town or are unable to hand in an assignment on time, it is up to you to make arrangements to hand your assignment early. If you are ill it is your responsibility to contact me as soon as you can to make arrangements regarding turning in homework late

Graduate Student Requirements

In addition to the assessments described above, graduate students will do a 'Journal Club' style presentation of a primary literature article. Students will present the necessary background information, describe the techniques used, and data presented in the paper. The article will be related to the topic we are currently discussing in class (i.e. Down syndrome, etc.)

Example of Topics Covered and Assigned Readings

Clinical Cytogenetics: The chromosomal basis of human disease

Down Syndrome

- Asim et al. 2015: "Down syndrome: an insight of the disease"
- Tejedor et al. 1995: "Minibrain: A new protein kinase family involved in postembryonic neurogenesis in *Drosophila*"
- Yabut et al. 2010: "Dyrk1A overexpression inhibits proliferation and induces premature neuronal differentiation of neural progenitor cells"
- Hammerle et al. 2011: "Transient expression of Mnb/Dyrk1A couples cell cycle exit and differentiation of neuronal precursors by inducing p27^{KIP1} expression and suppressing NOTCH signaling"

Cystic Fibrosis

- Tsui and Dorfman 2013: "The cystic fibrosis gene: a molecular genetic perspective"
- De Lisle and Borowitz 2013: "The cystic fibrosis intestine"
- Drumm et al. 2005: "Genetic modifiers of lung disease in cystic fibrosis"
- Jin et al. 2006: "The cystic fibrosis transmembrane conductance regulator (Cftr) modulates the timing of puberty in mice"

Fragile X

- Bassell and Warren 2008: "Fragile X syndrome: loss of local mRNA regulation alters synaptic development and function"
- Jin et al. 2003: "RNA-mediated neurodegeneration caused by the fragile X premutation rCGG repeats in *Drosophila*"
- Myrick et al. 2011: "Independent role for presynaptic FMRP revealed by an FMR1 missense mutation associated with intellectual disability and seizures"
- Zhou et al. 2016: "CGG-repeat dynamics and FMR1 gene silencing in fragile X syndrome stem cells and stem cell-derived neurons"

Schedule

This schedule and syllabus are subject to change.

Month	Date	Day	Topics
Jan	17	T	Chromosomal basis of human disease
Jan	19	R	Chromosomal basis of human disease
Jan	24	T	Chromosomal basis of human disease
Jan	26	R	Down syndrome
Jan	31	T	Down syndrome
Feb	2	R	Down syndrome
Feb	7	T	Down syndrome
Feb	9	R	Down syndrome
Feb	14	T	Down syndrome
Feb	16	R	Down syndrome
Feb	21	T	Library and Citation Manager
Feb	23	R	Guest Speaker
Feb	28	T	Cystic Fibrosis
March	2	R	Cystic Fibrosis
March	7	T	Cystic Fibrosis
March	9	R	Cystic Fibrosis
March	14	T	Spring Break
March	16	R	Spring Break
March	21	T	Cystic Fibrosis
March	23	R	Cystic Fibrosis
March	28	T	Cystic Fibrosis
March	30	R	Cystic Fibrosis
April	4	T	Fragile X
April	6	R	Fragile X
April	11	T	Fragile X
April	13	R	Fragile X
April	18	T	Fragile X
April	20	R	Fragile X
April	25	T	Fragile X
April	27	R	Presentations
May	2	T	Presentations
May	4	R	Presentations
May	9	T	Final Exam 12:00 PM
May	11	R	

Dates to keep in mind:

21 April 2017 – Last day to drop a class without a failing grade.

16 May 2017 – Final grades posted to the web.



To: Dr. James Champine, Chair, Department of Biology

From: Barbara Glackin, Dean of Kent Library

CC: Dr. Chris McGowan, Dean of the College of Science, Technology & Agriculture
Katlyn Griffin, Information Literacy Librarian, Kent Library
Vicki Howell, Administrative Assistant, Department of Biology

RE: BI 403/603 Medical Genetics course review

Katlyn Griffin, library liaison to the Department of Biology has reviewed the course approval document and sample syllabus for the proposed Medical Genetics course and has determined that Kent Library does have sufficient resources, including databases and journals, and services to support this course.

Accompanying this document is her review of specific library resources and services that support this course.

Thank you.

Kent Library Resources Review
BI 403 / 603: Medical Genetics

Major Databases

Biology (ProQuest)

ProQuest's Biology database provides access to primary literature on a wide range of biology topics, including biological chemistry, molecular biology, and genetics.

CINAHL Plus with Full Text

Cumulative Index of Nursing and Allied Health Literature (CINAHL) is the authoritative resource for nursing and allied health professionals, students, educators, and researchers. Topics include nursing, biomedicine, alternative/complementary medicine, consumer health, and 17 allied health disciplines.

MEDLINE (EBSCOhost)

MEDLINE is a database provided by the National Library of Medicine and indexes citations from over 5,000 biomedical journals.

PubMed & PubMed Central

PubMed comprises more than 26 million citations for biomedical literature from MEDLINE, life science journals, and online books. Citations may include links to full-text content from Kent Library holdings, PubMed Central, and publisher web sites.

Selected Journals

Clinical Genetics (1999-present, most recent year not available)

Cold Spring Harbor Perspectives in Medicine (2011-2015, most recent two years not available)

Development (1953-present, most recent 6 months not available)

Genetics in Medicine (1998-2013; 2015-present, most recent year not available)

Genome Research (1991-present, most recent 6 months not available)

Human Mutation (1997-2007)

Journal of Biomedical Science (1998-present)

Journal of Medical Genetics (1964-present)

Molecular Autism (2010-present)

Neuron (2002-present)

New England Journal of Medicine (1990-present, most recent 3 months not available)

Proceedings of the National Academy of Sciences of the United States of America (1915-present)

Library Instruction

BI 403 / 603 will include a “Library Literacy” unit and a session with a librarian. As part of this session, students will learn how to:

- Navigate PubMed and other library databases to find primary literature on their research topics.
- Use Kent Library’s E-Journal Portal to find primary literature referenced in other articles.
- Request articles through Interlibrary Loan.
- Use a citation management tool to keep track of their research.

Interlibrary Loan & Document Delivery

Interlibrary Loan and document delivery obtain items for Kent Library users that are not owned or accessible locally. These services give access to millions of items from all over the world. Users can request books as well as digital copies of journal articles. These services are free of charge to Southeast Missouri State University patrons.

Research Guides

Citation Management Software