

Syllabus Fall 2023

# MCB 493: Genetic Disorders and Counseling

**3 credit hours**

**Location: 3117 Everitt Laboratory**

**Everitt Laboratory**

## Course Description

This course addresses key issues in human genetics at an advanced level and is designed for pre-health care professionals. The course covers basic principles of medical ethics, chromosomes, complex disorders like autism and Alzheimer's disease, gene therapy and emerging technologies like CRISPR. Relevance to counseling individuals with these disorders will be discussed throughout the course, including calculations of recurrence risk for each disorder.

## Course Goals and Objectives

The goals of this course are to:

- Prepare students to be proficient in medical genetics by understanding the principles identified in the MCAT topics in genetics and to prepare students for a career in genetic counseling.
- Prepare students to be able to evaluate emerging genetic technologies in the care of patients.

## Course Structure

This is a **3-credit hour** course that meets Tuesday and Thursday for the entire Fall semester from 11:00-12:20 PM. There will be opportunities for constructivism, in-class participation using case-based vignettes, problem-solving and writing.

## Instructor

Professor Stephanie Ceman is the instructor and has been teaching medical genetics to medical students and undergraduates since 2004. She received her PhD in Genetics from the University of Wisconsin-Madison. Her office is room 423 in the Medical Sciences building and she is happy to arrange appointments by email (sceman@illinois.edu). She will reserve 9-9:50 AM Tuesday morning as an office hour, beginning August 29th.

## Textbooks

The recommended text for this course is: *Thompson & Thompson Genetics in Medicine*. Nussbaum, McInnes, Willard. 8<sup>th</sup> edition. 2016. W.B. Saunders Company, which can be accessed in either link below

<https://www.clinicalkey.com#!/browse/book/3-s2.0-C2009059798X>

Medical Genetics by Jorde *et al* is also an excellent resource (linked below)

<https://www-clinicalkey-com.proxy2.library.illinois.edu#!/browse/book/3-s2.0-C20170009679> .

Readings from the textbook are optional but recommended for any topics with which you may have questions. You will not need to read entire chapters: please use the **index** or **search function**.

You can also order this textbook from the [Illini Union Bookstore \(IUB\)](#) (217-333-2050). Be certain you select the correct term from the "Search by Course" dropdown list. Used books are often available and can be provided at a cost savings to you. Note that the cost and postage for all books must be prepaid. You can charge your order using VISA, Discover, or MasterCard. If you are ordering books elsewhere (e.g., Amazon.com), please be sure to order the correct edition and year of publication.

## Prerequisites

MCB 316 or MCB 317, or equivalent, or consent of instructor

## Student Learning Outcomes

At the end of this course, students should be able to do the following:

1. Apply the four basic principles of Bioethics
2. Use online tools like OMIM or other NIH sponsored websites to facilitate diagnosis and prognosis
3. Describe the three liveborn trisomies and the karyotypes of the parents
4. Calculate the recurrence risk for multifactorial disorders using empirical data
5. Understand the genetic basis of common diseases like autism and Alzheimer's disease
6. Understand the role of a genetic counselor and how to pursue training in that field

## Course Outline

Tentative Dates and Lecture topics

<b>Date</b>	<b>Title/topic</b>	<b>Associated readings in Thompson and Thompson or other sources</b>
T, Aug. 22	1. Introduction to the course and principles of bioethics	Chapter 1 and 19
R, Aug. 24	2. Human variation and prevalence of human disease	Chapter 4
T, Aug 29	3. Review of Mendelian inheritance	Chapter 4, refresher ppt on Mendelian genetics

R, Aug 31	4. Prenatal diagnosis and the newborn screen.	Chapter 17; Video interview with genetic counselor Judy Miller on how to give a diagnosis
T, Sept 5	5. Chromosome nomenclature	Chapter 5
R, Sept 7 and T, Sept 12	6. Numerical chromosome disorders	Chapter 6
R, Sept 14	Review	
T, Sept 19	Exam 1	
R, Sep 21, T Sept 26	7. Calculating disease incidence and population genetics. These calculations are relevant for risk calculations in families of specific ancestries	Chapters 9 and 16 Watch "Sunkissed" documentary
R, Sept. 28	8. Hemoglobinopathies	Chapter 11 <i>New Engl J Med</i> 2017;376:1561-67.
T, Oct 3	9. Principles of cloning and stem cell production	Chapter 4
R, Oct 5	10. Introduction to cystic fibrosis and gene linkage	Chapter 11
T, Oct 10	11. Gene Therapy and treatments for cystic fibrosis	Chapter 13
R, Oct 12	Review	
T, Oct. 17	Exam 2	
R, Oct 19	12. Imprinting and its associated diseases: Prader-Willi and Angelman syndrome.	Chapter 6
T, Oct 24, R, Oct 26	13. Complex inheritance and gene by environment interactions. These calculations are relevant for recurrence risk calculations in families with an affected individual	Chapter 8
T, Oct.31	14. Genetics underlying personality and autism.	Supplemental article: <i>Ceman and DeThorne</i>
R, Nov 2	Review	

T, Nov 7	Exam 3	
R, Nov 9	15. Genetics of neurodevelopmental disorders with a focus on trinucleotide repeat disorders	Chapter 8
T, Nov 14	16. Genetics of intelligence	Supplemental article: <i>Sniekers, 2017 Nat. Genetics</i>
R, Nov 16	17. Genetics and etiology of Alzheimer's Disease.	Chapter 12
	Thanksgiving break	
T, Nov 28	18. Hereditary cancers	Chapter 15
R, Nov 30	19. Genetics of spontaneous cancers	Chapter 15
T, Dec 5	Exploring a career in genetic counseling; review of relevant websites	

**Final exam: TBA** please note that the final is **not** cumulative

The final will be Friday December 8<sup>th</sup> 8-11 AM

## Course Activities

You are expected to complete your work independently, in accordance with [University policy](#). Failure to do so will result in strict disciplinary action, including loss of all credit for the assignment, notification of a dean, and possible dismissal from the University. You may work with others on homework, but the final product must be your own.

### Assignments, Weights, and Deliverables

You can access your scores on the Moodle site

Points for weekly Reflections, authored exam questions, in-class participation points (please note that the latter cannot be made-up) are described below.

#### Reflections

At the end of each week, you will take time to reflect on what you have learned in the week, which is worth 15% of the total grade. The focus of this reflection is a review of your initial thoughts and how your knowledge about the week's content has changed. The grading rubric is on the course website.

#### Authored exam questions.

Writing a question that tests knowledge of concepts and does not merely reflect memorization is an important learning tool. 10% of your grade will be earned by authoring an exam question/test so they are worth 2.5% apiece. Please note, you may see your question on the exam!

## Essay

My challenge to you is teach me something I do not know!

Please prepare an essay that explores any topic you learned about this semester. The broad goal is to pitch an idea or a next direction that **builds** on what you learned in the course.

I will give a specific example that was not covered in this course, thus, is not a possible topic: the genetics of longevity. The question is “why do some human beings live to be 100 years or older?” One hypothesis could be that centenarians have specific alleles at the apolipoprotein C3 locus that contribute to their longevity. Now provide the evidence that supports or refutes this hypothesis. If the hypothesis is refuted, state a new hypothesis. In the last sentence or 2 of the assignment, pitch or propose a way to increase human longevity through a possible treatment approach supported by your research: examples might be a specific diet, or small molecule activators or inhibitors, or microbiome changes, etc..

In your essay, identify a topic or disease that you found interesting (indicate lecture number or date) and explain why you found it interesting. Next and importantly, describe what you would explore **next**, based on your hypothesis and what you think the next break-through will be. This is really the heart of the essay and you will likely have pages of research exploring this. This should be a novel interpretation or a new model. You may turn in everything you have found; however, I will only grade **the first page**. Some students have referred to this as the "abstract" of your research. It should **not exceed 300** words, which is about one double-spaced page.

You should **include references**, which are **not** included in the word limit. This activity is worth 10% of your grade (100 pts) and is due on Monday Dec 11 at 11:59 PM.

**Grading** will be as follows: Identify a topic or disease that you find interesting and identify the question and hypothesis that you will test (25 pts), indicate lecture number or date (5 pts) explain why you found it interesting (10 pts), what aspects you would explore next (20 pts) and speculate on the next break-through or treatment approach (20 pts). You will be awarded 10 pts for accurate spelling and grammar. As references, you must have at least one primary paper and one review (10 pts) but they are outside of the word limit. This activity is worth 100 pts or 10% of your grade and is due on Monday Dec.13 at 11:59 PM. Exceeding 350 words will result in loss of 20 pts.

## Exams

Four exams will be given

**Final grade:** There are 15 weeks of instruction. The contribution of each activity to your total grade is as follows

Reflections: 15% (1.0%/week)

Authored exam questions: 10%

Participation points identified by iClicker polling 5.0% (0.25% x 20 lectures)

Essay: 10%

Exams:

60% (15% for each exam).

Scale:

Letter Grade	Point Ranges	Grade Point Value
A+	1000–920	4.000
A	919–880	4.000
A-	879–840	3.667

B+	839–800	3.333
B	799–760	3.000
B-	759–720	2.667
C+	719–680	2.333
C	679–640	2.000
C-	639–600	1.667
D+	599–560	1.333
D	559–520	1.000
D-	519–480	0.667
F	479–0	0.000

**A note about sources of information:** It is highly recommended that you only consult the following sources of information in studying for this class. Use of another source (such as Internet sites found via Google) may provide information that is unreliable.

- Suggested books and readings
- Supplemental information posted on course website
- Internet links provided in class or on course website
- Any website that is sponsored by the National Institutes of Health (NIH)

### **Course Policies and Resources** (ex. MCB Core Curriculum Policies)

#### **For non-academic campus assistance and support:**

- See Office of Diversity, Equity and Access (ODEA) information at the end of this document.

#### **Student Advocacy Resources:**

- For student-centered advocacy programs and services visit:  
[mcb.illinois.edu/undergrad/advising/resources](http://mcb.illinois.edu/undergrad/advising/resources).

#### **Contacting MCB Course Personnel:**

- MCB course personnel are more than happy to assist students.
- Emails to instructors, TAs, or course coordinators will only be answered if they come from an @illinois.edu account. We will only use this account in order to protect your educational information and profile. As a student, please remember that when you email a staff member, it is important to include all pertinent information so that we can assist you in the most efficient and effective manner possible. This information includes:
  - The course rubric in the subject line
  - Your full first and last name
  - Your NetID (the first part of your illinois.edu email account)
  - Your UIN (9 digit number that can be found on your ICard)
  - The course that you are concerned about (the course personnel often work with multiple courses)
  - Your section letter/number
  - The previous email "thread" or previous communicated information pertinent to the situation
- Your cooperation will help us respond much more quickly to your concerns.

**Religious Observances and Practices:**

- Students are required to submit the Request for Accommodation for Religious Observances Form (which can be found at [www.odos.illinois.edu/.../Religious\\_Observance\\_Accommodation\\_Request\\_Form.docx](http://www.odos.illinois.edu/.../Religious_Observance_Accommodation_Request_Form.docx)) to their instructors and the Office of the Dean of Students requesting accommodation by the end of the second week of the course. Requests that are not submitted within this time frame may not be granted. Information about accommodations can be found in the Student Code: <http://studentcode.illinois.edu/>.

**DRES Accommodations:**

- If a student has DRES (Disability Resources and Educational Services) accommodations, documentation must be submitted to course personnel by the end of the second week of class.
- If a student believes that they need DRES accommodations, they should contact DRES at [disability@illinois.edu](mailto:disability@illinois.edu).

**Class Absences:**

- Regular class attendance is expected of all students at the University. ([http://odos.illinois.edu/studentAssistance/absence/revise\\_code.asp](http://odos.illinois.edu/studentAssistance/absence/revise_code.asp))
- The Office of the Dean of Students will only provide informative letters to instructors for protracted illness of 3 or more days, certain emergencies and to be present during the serious illness of immediate family members (parents, legal guardian, spouse/partner, siblings, children, or grandparents). These letters do not excuse you from class but merely provide information for the instructor to consider with regard to excusing the absence and permitting make-up work. Students must request absence letters from the Office of the Dean of Students after the student has returned to class but not more than 10 business days after the last date of absence.
- Absences that may be excused without a letter include circumstances beyond the student's control such as medical treatment, surgery related to prolonged illness or injury, pregnancy, legal matters, citizenship or naturalization processes, or acts of nature which cause destruction to a primary residence or disrupt air travel. All will require documentation.
- Absences that may also be excused without a letter include job, graduate or professional school interviews, though a best effort should be made to schedule these events to minimize class attendance disruption.
- Absences planned for the items listed in previous bullet point must be communicated to your instructor or course coordinator at least two weeks in advance of the absence. Failure to do so may result in the loss of opportunity to reschedule the missed class period and the portion of the grade associated with this class period.
- Absences that will not be excused include family events such as reunions or weddings, or presence during serious illness of extended family members (aunt, uncle, niece, nephew, or cousin).
- Unplanned absences may result in the loss of opportunity to reschedule the missed class period and, therefore, the portion of the grade associated with this class period.
- Absences will be handled according to individual course policy.

**Exam Conflicts:**

- If you have a regularly scheduled University course that conflicts with the exam, you should complete the online Conflict Exam Request Form on the course website. This request must be made by 5:00 pm not less than 3 business days prior to the exam. Requests made after 5:00 pm and less than 3 business days prior to the exam will not be granted. See course policies for a specific deadline for your course.
- Work schedules should be adjusted, if at all possible, in order to eliminate a conflict with scheduled exams. Please plan accordingly at the beginning of the semester. If eliminating a conflict is not possible, the student should complete the online Conflict Exam Request Form on the course website. This request must be made by 5:00 pm not less than 3 business days prior to the exam. Requests made after 5:00 pm and less than 3 business days prior to the exam will not be granted.
- Students that are formally participating in officially recognized groups, such as athletic teams and performing groups, with a conflict should request a conflict exam by 5:00 pm not less than 3 days prior to the exam via the online Conflict Request Form. Formal participation does not include general meetings of RSOs or any other recognized groups. Documentation of the event will be required prior to scheduling the conflict exam. Requests made after 5:00 pm and less than 3 business days prior to the exam will not be granted.
- Students with DRES accommodations should also submit the online Conflict Request Form by 5:00 pm no later than 3 business days prior to the exam. Requests made after 5:00 pm and less than 3 business days prior to the exam will not be granted.

#### **Final Exam Absence:**

- If you must miss a final exam due to unforeseen circumstances, you are required to contact your instructor or course coordinator within 24 hours of the absence. You must also contact the Dean of your college. Finally, you must submit an online Absence Form if one is available on your course website. You will receive an ABS (absent) in the course if you miss the final exam. This ABS will result in an F in the course unless action is taken. The Dean can approve the change of the ABS to an Incomplete, which then allows a limited window of time for you to complete the final exam and earn a grade in the course.
- There will be instances when the student must make an individual choice about their ability to perform on an exam and will need to accept any and all consequences for that choice.
- If the absence is a result of a protracted illness, you should follow the procedure for obtaining a letter from the Office of the Dean of Students. The request may be made once the student recovers but not more than 10 business days after the date of absence.
- Information about final exams can be found in the Student Code: <http://studentcode.illinois.edu/>.

#### **Final Exam Conflict:**

- Conflict final exams may only be granted for any one of the following situations:
  - Students with three final exams scheduled within a 24 hour period as defined in Section 82.A.4). Final Examinations of the *Code of Policies and Regulations Applying to All Students* which can be found at: [www.illinois.edu/admin/manual/code/](http://www.illinois.edu/admin/manual/code/)
  - Students who have two final exams scheduled at the same time. Final conflict exam requests should be made to the course with larger enrollment. Course personnel can assist with information to determine which course this would be.
  - Students who have a verified personal problem, and who have received written permission to take a conflict final exam from a dean in their college.

- Students who have DRES academic accommodations.
- Students that find themselves in any of the above situations should complete the online Conflict Final Exam Request Form which can be found on the course website. This request must be made by 5:00 pm on the last day of class in order for the request to be granted. Any requests made after this time may not be granted. If a conflict final exam is granted, it may be scheduled at any time during the final examination period and is at the discretion of the instructor or course coordinator.

### **Academic Integrity:**

- Science cannot exist without honesty. The faculty and staff in MCB require students, as scientists-in-the-making, to hold the highest standards of scientific and academic conduct. Any form of cheating on any graded work in courses is unacceptable, and will be dealt with as outlined below, and in accordance with the University-wide standards in the Code of Policies and Regulations Applying to All Students.
- We require that all graded work be entirely your own, and that anything you write using the words of other writers be correctly attributed. Some specific points follow.
- On exams, the answers that you turn in for grading must be your own, formulated during the exam from your own understanding of the material and without any supporting information, be it written, verbal or electronic. Copying the work of another student, or allowing another to copy your work, or copying work from any other source, is unacceptable. Since we cannot always monitor you as you complete your work, we must rely upon appearance of your work from which to judge. If the work you submit resembles that of another student or another source too closely, we may conclude that it was not your original work. Always make a conscious effort to complete your work on your own and to protect it from the view of others, in order to ensure that it will be seen as your own. Failure to adhere to these standards for any portion of an exam may result in a grade of zero for the entire exam or quiz for all persons involved.
- Texting, or the use of a cell phone or any other device for any purpose, during a quiz or exam is prohibited. Doing so may earn you a zero or a more extreme penalty on the quiz or exam at the discretion of the instructor.
- Use of any social or electronic media to share information, request information or make confidential information public is prohibited. Any use of this type may earn you a zero on the exam or a more extreme penalty at the discretion of the instructor.
- On written or electronic assignments, the answers that you turn in for grading must be written in your own words, formulated from your own understanding of the material. While you may be working with other students in the course, you must formulate and submit your own answers. Copying or paraphrasing the work of another student, or allowing another to copy or paraphrase your work, is unacceptable. Since we cannot monitor you as you complete your work, we have only the appearance of your work from which to judge. If the work you submit resembles that of another student too closely, we may conclude that it was not your original work. Always make a conscious effort to complete your work on your own and to protect it from the view of others, in order to ensure that it will be seen as your own. You must also make a conscious effort to protect your passwords and accounts. Failure to adhere to these standards may result in a grade of zero for the entire assignment for all persons involved.
- On written or electronic assignments, if you use a statement taken directly from any book or other publication, including the course textbook, you must provide a citation. That is, you must put the text in quotes and put the author of the publication in parentheses after the quotation. Failure to do so will result in zero credit for that answer. Further, using only the words of another author as

your entire answer or as the majority of your answer to any question is never sufficient to earn credit. If the majority of your work has been taken directly from a publication, you are likely to receive no credit for the work, since you would not be demonstrating knowledge beyond the ability to copy. Even if you quote another, your answer must be substantially your own words, drawn from your own understanding of the material.

#### **Social Media Use:**

- Use of any social or electronic media to share course information, request course information or make confidential course information public is prohibited. Any use of this type may earn you a zero or a more extreme penalty at the discretion of the instructor on an assignment or exam.
- Any social media sites created in relation to MCB courses must grant access to course personnel upon request. Failure to provide access will result in a failing grade in the course for the group/site's administrator(s).

#### **Course Material:**

- Students are welcome and encouraged to make audio recordings of course lectures.
- The material recorded is intellectual and copyrighted property of the University of Illinois Board of Trustees and may be made for personal use only.
- Video recordings of any kind are strictly prohibited.
- Posting of audio recordings or transcriptions on social or electronic media platforms is strictly prohibited.
- Posting or redistributing of course material in any format is strictly prohibited.

#### **University Information on Student Safety**

##### **Active Threats:**

- **General Emergency Response Recommendations** ([Emergency Response Guide](#)):
- Security Threat. The Department of Homeland Security and the University of Illinois at Urbana-Champaign Office of Campus Emergency Planning recommend the following three responses to any emergency on campus: **RUN > HIDE > FIGHT**
  
- **Only follow these actions if safe to do so.** When in doubt, follow your instincts - you are your best advocate!
  
- **RUN** — Action taken to leave an area for personal safety.
  - Take the time to learn the different ways to leave your building **before** there is an emergency.
  - Evacuations are mandatory for fire alarms and when directed by authorities! No exceptions!
  - Evacuate immediately. Pull manual fire alarm to prompt a response for others to evacuate.
  - Take critical personal items only (keys, purse, and outerwear) and close doors behind you.
  - Assist those who need help, but carefully consider whether you may put yourself at risk.
  - Look for **Exit** signs indicating potential egress/escape routes.
  - If you are not able to evacuate, go to an Area of Rescue Assistance, as indicated on the front page of this plan.
  - Evacuate to Evacuation Assembly Area, as indicated on front page of this plan.
  - Remain at Evacuation Assembly Area until additional instructions are given.

- Alert authorities to those who may need assistance.
- Do not re-enter building until informed by emergency response personnel that it is safe to return.
- Active Threat: IF it is safe to do so, run out of the building. Get as far away as possible. Do NOT go to the Evacuation Assembly Area.
- **HIDE** — Action taken to seek immediate shelter indoors when emergency conditions do not warrant or allow evacuation.
  - Severe Weather:
    - If you are outside, proceed to the nearest protective building.
    - If sheltering-in-place due to severe weather, proceed to the identified Storm Refuge Area or to the lowest, most interior area of the building away from windows or hazardous equipment or materials.
  - Active Threat:
    - Lock or barricade your area.
    - Get to a place where the threat cannot see you.
    - Place cell phones on silent.
    - Do not make any noise.
    - Do not come out until you receive an Illini-Alert advising you it is safe.
- **FIGHT** — Action taken as a last resort to increase your odds of survival.
  - Active Threat: If you cannot run away safely or hide, be prepared to fight with anything available to increase your odds for survival.

#### **We Care at Illinois:**

- For sexual misconduct support, response and prevention visit: [wecare.illinois.edu](http://wecare.illinois.edu)

Title IX makes it clear that violence and harassment based on sex and gender are Civil Rights offenses subject to the same kinds of accountability and the same kinds of support applied to offenses against other protected categories such as race, national origin, etc. If you or someone you know has been harassed or assaulted, you can find the appropriate resources here:

[http://oiir.illinois.edu/sites/prod/files/SexualMisconduct\\_ResourceGuide.pdf](http://oiir.illinois.edu/sites/prod/files/SexualMisconduct_ResourceGuide.pdf)

University Police Department, Emergency, 9, -911; Non-emergency, 217.333.8911

Counseling Center, 110 Student Services Bldg., 610 E. John St., 217.333.3704

McKinley Health Center, General Information, 217.333-2701

McKinley Mental Health Center, 1109 S. Lincoln, 217.333.2705

Dean of Students, 300 Turner Students Services Bldg, 610 E. John St., 217.333.0050

Local Sexual Assault Center, RACES, 217.384.4444

Women's Resources Center, 703 South Wright Street, 2nd Floor, 217-333-3137

#### **The Office of Diversity, Equity and Access (ODEA):**

- For non-academic support visit: [diversity.illinois.edu](http://diversity.illinois.edu)
  - Discrimination & Harassment Prevention
  - Title IX
  - Accessibility & Accommodations
  - Inclusive Illinois

#### **Mental Health Statement**

- Significant stress, mood changes, excessive worry, substance/alcohol misuse or interferences in eating or sleep can have an impact on academic performance, social development, and emotional wellbeing. The University of Illinois offers a variety of confidential services including individual and group counseling, crisis intervention, psychiatric services, and specialized screenings which are covered through the Student Health Fee. If you or someone you know experiences any of the above mental health concerns, it is strongly encouraged to contact or visit any of the University's resources provided below. Getting help is a smart and courageous thing to do for yourself and for those who care about you.
- Counseling Center (217) 333-3704
- McKinley Health Center (217) 333-2700
- National Suicide Prevention Lifeline (800) 273-8255
- Rosecrance Crisis Line (217) 359-4141 (available 24/7, 365 days a year)

This statement is approved by the University of Illinois [Counseling Center](#).