

General Information

MCB 270: Medical Genetics

Course Description

This is an online course that addresses key issues in medical genetics, defined as human genetics for pre-health care professionals. The course is informed by subject material on the MCAT, covering basic principles of medical ethics, modes of inheritance, the molecular basis of genetic disorders, chromosomes, treatment approaches, gene therapy and emerging technologies like whole genome sequencing.

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.
- Prepare students to be able to utilize emerging genetic technologies in the care of patients.

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. Interpret a pedigree
3. Identify disorders having Mendelian inheritance and calculate recurrence risks
4. Understand how ancestry increases the risks of certain common diseases
5. Use online tools (OMIM) to facilitate diagnosis and prognosis
6. Describe the advantages and challenges of sequencing technologies
7. Describe the three liveborn trisomies and the karyotypes of the parents.
8. Understand that multifactorial disorders are caused by the interaction of both genes and environment
9. Calculate recurrence risk for multifactorial disorders using empirical data
10. Describe how cancer is a genetic disorder

Course Structure

This is a **3-credit hour** course. The course is **8 weeks** long and consists of 8 content modules. Please be aware that this course is accelerated in nature; 16 weeks' worth of content will be covered in a 8-week time span. You should dedicate approximately **9–12 hours** per week to working on the course itself, but actual time commitments will vary depending on your input, needs, and personal study habits. You are required to log on to the course website a minimum of **4 days per week** but as discussions develop, you will probably need to do so more frequently.

We realize that you have a life beyond the scope of this course. However, if you are unable to complete an assignment because of professional obligations, you should notify the instructor or, better yet, prepare the assignment ahead of time and post it early. This will give your classmates a head start in reading and responding to your work. Most assignments are due by 10:55 PM of their respective due dates as listed

on the weekly overviews, giving you and your classmates time to read and comment on each other's work before the next module begins. **For various reasons, there can be problems uploading assignments. Please do not wait until 10:45 to upload!!! Late assignments will not be accepted.**

Note that instruction continues during Spring Break. You are still required to complete all assignments by the deadlines listed on the weekly overview page.

Readings and responses to discussion questions should be read and submitted during the module for which they are assigned in order to get the most benefit from the discussions. At the end of each content module, participants will have an opportunity to make sure that they have completed all the required activities and assignments.

Textbooks

The recommended text for this course is: *Thompson & Thompson Genetics in Medicine*. Nussbaum, McInnes, Willard. 8th edition. 2015. W.B. Saunders Company.

Readings from the textbook are optional but recommended for topics with which you may have questions beyond the presented material. You will not need to read entire chapters: please use the **index** to search for terms or topics that you need more information about.

This textbook is available electronically https://vufind.carli.illinois.edu/vf-uiu/Record/uiu_7761677

You may 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.

Note: In the course outline below, the chapters are indicated first, followed by page numbers. Case studies are indicated in ***bold italic***.

Course Outline

Week	Objectives	Lecture Titles	Associated Readings
Week 1: Introduction to Medical genetics and Genomic variation	<ul style="list-style-type: none"> Identify the basic ethical principles that guide the practice of medical genetics Explain how genetic variation can predict response to medications and effect proper dosing of medications. Identify the types of genetic variation observed in humans 	Introduction to Medical genetics, including medical ethics and personal genomics	Chapters 1, 18, 19; pg. <i>438-439</i>
		Genetic variation in individuals	Chapter 4
Week 2: Molecular basis of human DNA and types of mutation	<ul style="list-style-type: none"> Analyze a Southern blot Describe gene regulation, including epigenetics and microRNAs 	Molecular basis of chromosomes/Molecular Biology Tools	Chapters 3, 4

Week	Objectives	Lecture Titles	Associated Readings
	<ul style="list-style-type: none"> Predict how mutations occur and lead to disease 		
Week 3: Mendelian inheritance and Population Genetics	<ul style="list-style-type: none"> Construct a three-generation pedigree and from this, determine the mode of inheritance of single gene disorders Derive allele frequencies from genotype frequencies Derive genotype frequencies from allele frequencies using the Hardy-Weinberg law Define the terms 'Founder Effect' and 'Genetic drift' Use probability to assess risk for a genetic disease 	Mendelian Inheritance/ Pedigree Analysis	Chapters 7, 12
		Genetic variation in Populations/Risk	Chapter 9
Week 4: The role of genes in development and features of genes on the X chromosome	<ul style="list-style-type: none"> Explain how patterns of gene expression vary during development and recognize the anomalies that result from disordered embryonic development, focusing on HOX genes and SHH Understand how stem cells are derived and the role of transcription factors in 'stemness' Discuss and diagnose the X chromosome-encoded disorders of muscular dystrophy, fragile X and Lesch nyhan disease 	Single Gene Disorders: Development	Chapter 14; pg. 436-7
		X-linked Disorders	418-19; 424-425
Week 5: Chromosome nomenclature and disorders	<ul style="list-style-type: none"> Compare mitosis and meiosis and the consequence of nondisjunction Define the structure and function of chromosomes and utilize cytogenetic nomenclature Identify the features of the live-born trisomies and the sex chromosome aneuploidies 	Chromosome nomenclature and Numerical Chromosome Disorders	Chapters 2, 5, 6; 484-5
		Prenatal Screening and Diagnosis	Chapter 17

Week	Objectives	Lecture Titles	Associated Readings
	<ul style="list-style-type: none"> Predict the impact of balanced translocation carriers on gamete formation Explain the roles of 1st and 2nd trimester ultrasound and analyte testing and the common indications, risks and benefits of chorionic villi sampling and amniocentesis 		
Week 6: Cystic Fibrosis as a disease model for understanding linkage and gene therapy, along with a discussion of two common disorders of imprinting	<ul style="list-style-type: none"> Describe the pathophysiology of cystic fibrosis and how it demonstrates allelic heterogeneity 	Cystic fibrosis as a model for linkage and gene therapy	Chapters 2, 5, 6; 414-5
	<ul style="list-style-type: none"> Describe the pathophysiology of cystic fibrosis and how it demonstrates allelic heterogeneity Explain gene therapy, including its successes and failures Describe the mechanisms of imprinting, recognizing the clinical features of the imprinting disorders Prader-Willi and Angelman syndrome 	Imprinting	
Week 7: Multifactorial disorders	<ul style="list-style-type: none"> Compare and contrast the genetic contribution to single gene disorders vs. multifactorial disorders 	Multifactorial Inheritance	Chapter 8; 398-9; 442-3
	<ul style="list-style-type: none"> Describe the inheritance patterns of multifactorial disorders, focusing on cleft lip/palate and Type 1 diabetes Describe how twin studies are used to estimate the relative influence of genes and environment Describe mapping genes of complex traits, using association studies and odds ratios 	Genetics of Personality	400-401

Week	Objectives	Lecture Titles	Associated Readings
	<ul style="list-style-type: none"> Assess the role of genes and environment in autism and schizophrenia 		
Week 8: The role of genes in cancer	<ul style="list-style-type: none"> Evaluate the impact of cancer in the US Describe the genetic basis of cancer Compare and contrast the three major classes of genes involved in cancer: proto-oncogenes, tumor suppressor genes and cellular maintenance genes Discuss the use of microarray analysis in cancer diagnosis and prognosis and RNA-seq 	Cancer – part 1	Chapter 15; <i>410-11</i>
		Cancer-part 2	Chapter 15; <i>404-5; 420-1; 468-9;</i>

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 by clicking the **Grades** link from the left column of the course home page.

All interim and final deliverables have due dates. Failure to meet deadlines results in a reduction of the assignment points. For the due dates of each assignment, please see the weekly overviews.

Students will receive specific deadlines online for when the material described above is due. **No credit will be given for late submissions.**

If you add the class late, i.e, after it begins, you are still responsible for meeting the deadlines

Weekly Overviews

Each module will begin with the module overview, explain what the module is about, what learning goals you are expected to achieve, how long the module will take, and in what activities you will participate. Each module is designed with the same structure and activities unless otherwise specified. The module activities are explained in greater detail below. You can find the due dates of specific assignments on each week's Overview page.

Point Distributions

Assignments	We ek 1	We ek 2	We ek 3	We ek 4	We ek 5	We ek 6	We ek 7	We ek 8	Total points per assignment	Relative weight
Lecture Questions	10	10	10	10	10	10	10	10	80	8%
Quizzes	25	25	25	25	25	25	25	25	200	20%
Wiki – submission score	10	20	80				80		190	19%
Wiki – grader's score				25				25	50	5%
Exam question authoring				25			25		50	5%
Reflection	15	15	15	15	15	15	15	15	120	12%
Exams					150			160	310	31%
Weekly Totals	60	70	130	100	200	50	155	225	1000	100%

Quizzes

Each week there will be an online quiz worth 2.5% for a total of 20% of the final grade

Wiki

Each student will be required to work on a Wiki about a genetic disease, which will be graded twice during the course for a total of 16% of the final grade. You will be asked to comment on each other's Wikis twice during the course, which counts for 4% of the grade.

Exam Questions

Students will be asked to write exam questions, which are worth 4% of the grade.

Reflections

At the end of each week, you will take time to reflect on what you have learned in the week, which is worth 6.4% 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.

Exams

Two exams will be given: one midterm, one at the end, each worth 14% of the total grade. For on-campus students, these exams will be taken at arranged times and locations your instructor will tell you about as the time draws near. For off-campus students, you are responsible for identifying a suitable proctor and proctoring location. Contact your instructor for additional information.

Exam Proctoring Options

Option 1: On-Campus: If you wish to take the exams at the Urbana-Champaign campus, they will be held in [BLDG. & RM. LOCATION] on [DATE]. TO BE ANNOUNCED.

Option 2: ProctorU: ProctorU is an online proctoring service that allows students to take exams online while ensuring the integrity of the exam for the institution. The service authenticates your identity and monitors both your computer screen and webcam to ensure academic integrity:

- Fee based exam (\$14.75 /1-hour exam, \$21.50 /90-min. and 2-hour exam, \$30.25 /3-hour exam) billed when you schedule the exam. Refunds are given only if the exam is canceled within 48 hours before the scheduled exam time.
- All appointments should be made at least 3 days in advance, since reservations made within 72 hours of your exam are subject to a \$5 late reservation fee.
- Web cam, microphone, and computer with internet connection are required.
- [ProctorU System Requirements and System Test](#)
- [How to Schedule Your Exam with ProctorU](#)

Option 3: Proctored Location Off Campus (other than ProctorU): If you choose a proctored location off campus (other than ProctorU), it must allow for web-based testing. You must also complete the [Third-Party Proctor Request Form](#) and email it to your instructor no later than two weeks before the exam date to indicate your third-party proctored exam choice and your off-campus proctor information. You may consult the NCTA links below for possible testing center locations. Note that not all testing centers listed have web-based testing, so please be sure to choose a testing center that will allow you to take your tests online (for Illinois Compass/WebCT, see the [java and browser requirements](#)). Once you have chosen your proctor, it is your responsibility to schedule the exam with the proctor. Your instructor will notify the proctor with instructions on how to access your exam. Please verify with the proctor that they will be open and able to proctor the exam for you before contacting your instructor about this proctored exam choice. NOTE: the final decision to approve a third-party proctor is up to the instructor.

- [NCTA Consortium of College Testing Centers](#)
- [NCTA College Testing Centers in Illinois](#)

Checklist

Each week will present a checklist at the end to help you verify with confidence that you have completed all of the required activities for that week.

A note about sources of information: It is highly recommend 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 required 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)

Getting Help

If you need help:

- Only contact your instructor directly if you have a personal question.
- For all other questions about course content, activities, deadlines, technical problems, etc., please check the [General Q & A](#) forum to see if someone else has already asked your same question and received a response.
- If your question isn't there yet, post your question to the [General Q & A](#) forum. Feel free to help your peers out if you know the answer!
- If you have technical problems, please fill out [this form](#).

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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.

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) 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.

Class Absences:

- Regular class attendance is expected of all students at the University. (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 Absences:

- There are no excused absences for exams because one exam will be dropped

Exam Conflicts:

- If you have a regularly scheduled University course that conflicts with the exam, you need to schedule an exam on ProctorU. Please see the course website for details under “Exams

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/
 - 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 your 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

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

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
 - Discrimination & Harassment Prevention
 - Title IX
 - Accessibility & Accommodations
 - Inclusive Illinois

Copyright Indication/Symbol