



JOHNS HOPKINS
SCHOOL *of* MEDICINE

COURSE TITLE

Fundamentals of Clinical Genomics

SEMESTER

Spring 2024

PRE- AND CO-REQUISITES

None

RESPONSIBLE FOR COURSE

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COURSE WEBSITE

<https://jhu.instructure.com/courses/58618>

COURSE DESCRIPTION

This course provides the fundamental background and tools of genetics that are utilized by the genetic assistant when working in a laboratory or clinic. Topics include an overview of the genetic contribution to healthcare past and present, cell biology and genetics, genetic testing technology and application, and clinical genetic services. The course will wrap with a discussion of the pop culture and ethical considerations inherent to genetic and genomics. The course intends to provide an overview of various concepts of biology, medicine, genetic testing, and genetic counseling, so that students have background knowledge to learn their job-specific responsibilities.

COURSE OBJECTIVES

By the end of this course, you should be able to:

1. Identify the cellular structures and pathways that contribute to genetic variation and disease
2. Compare genetic and genomic technologies as each relates to the ability to identify genetic/genomic variation
3. Explain the indications for genetic testing through the lifespan
4. Describe the historical and current role of genetic testing and services in

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medicine

5. Utilize genetic and genomic resources
6. Discuss the ethical issues in genetic and genomics

REQUIRED READINGS AND OTHER COURSE MATERIALS

All required reading and other materials are included in the online course site.

ASSESSMENT SUMMARY

The following activities will be assessed in this course.

All assessments will be graded as Pass/ Fail, with a passing score being equal to or greater than 80%.

At the end of the course, all assessments will be averaged and you will need to have an average of 80% or greater in order to receive a passing grade for the course. You must submit work for each of the assignments to pass the course.

Assessment	Due Date	Points Possible
Quizzes (6)	2/11 2/18 2/25 3/3 3/10 3/17	100 (each)
Discussions (3)	3/28 & 3/31 4/4 & 4/7 4/10 & 4/12	100 (each)
Genetic Resources Assignment	3/24	100

LATE ASSESSMENT POLICY

All assignments must be turned in by the specified due date and time. Once the due date and time have passed, 5% of the total points of the assignment will be deducted per day (per 24-hour period).

Please contact the course instructor prior to the due date to request an extension on an assignment due to any extenuating circumstances. Your instructor will then work with you to determine a modified assignment due date.

Any assignment submitted later than 7 days past the assigned or modified assignment will receive a grade of Fail.

Any assignments with modified due dates cannot be submitted any later than 7 days after the end of the course.

ASSIGNMENT DEADLINES AND EXTENTIONS POLICY

All learners are required to inform the instructor in advance if they anticipate needing an extension on an assignment. Requests for extensions must be made before the assignment's due date, outlining the reasons for the extension request.

Extensions granted will generally not exceed one additional week beyond the original deadline. Please note that extensions are not guaranteed and will be considered on a case-by-case basis at the instructor's discretion.

It's imperative to note that all assignments, including those granted extensions, must be submitted no later than one week after the conclusion of the course. Failure to adhere to this deadline may result in a deduction of points or a grade penalty, as outlined in the course grading policy.

COURSE GRADING

The course will be graded on a Pass/Fail scale with a passing grade being equal to or greater than 80%. In order to receive a passing grade for the course, it is essential that you **submit work for each of the 10 modules** throughout the entire duration of the course. **Any incomplete weeks, where work is not submitted, will result in an automatic failure for the course.**

At the end of the course, all assessments will be averaged and you will need to have an average of 80% or greater in order to receive a passing grade for the course.

CERTIFICATE OF COMPLETION & LETTERS OF RECOMMENDATION

A certificate of completion can be achieved for the program by completing *both* the *Fundamentals of Clinical Genomics* and *Working as a Genetic Assistant* courses with a passing grade.

The courses do not have to be taken in the same semester, and you will receive the certificate of completion after successful completion of the second course. If you are in need of a letter of completion after completion of the first course you take, please contact JHOnline@jhmi.edu.

Please also note that we are unfortunately not able to write/ provide letters of recommendation.

COURSE REQUIREMENTS AND ASSESSMENTS RESOURCES

Most modules will include presentations and required resources, including readings and handouts. It is recommended that you listen to the presentations and read/review the resources, as this will best facilitate your successful completion of course assessments.

DISCUSSION POSTS

You are required to contribute to discussion board topics as posted.

You should post a thoughtful and complete response and reply to at least two classmates' posts by the due date to receive full credit.

Suggested length is one to two paragraphs. High quality posts will contribute substantive content, illustrate a strong understanding of course material, reflect professionalism, and be free of grammatical errors. Please cite sources using APA guidelines and include links as appropriate. Guidelines for this style can be found at: [APA Style Guidelines](#)

QUIZZES

Regular quizzes will test your understanding of course content. Quizzes can be attempted three times each and you are encouraged to consult course materials as needed to complete each quiz.

These quizzes are not timed tests, but please understand that if you walk away from your quiz while taking it, depending on your computer and the length of time, you may be automatically logged out of Blackboard and may lose your quiz responses.

WRITTEN ASSIGNMENTS

Written assignments should be composed in complete sentences and include proper grammar, spelling, and punctuation. Files can be submitted in .docx, .pdf, and .png or .jpg formats.

All work submitted should include references for any resources consulted. Please cite work in formats consistent with the American Psychological Association (APA) Style. Guidelines for this style can be found at: [APA Style Guidelines](#)

LIVE Q & A SESSIONS (OPTIONAL)

Throughout the semester, there will be three live (synchronous) optional Q & A sessions with the course instructors for this course and the *Working as a Genetic Assistant* course. The instructors will be available to answer questions about the course and subjects related to the course material.

Additional information about the sessions, including dates, times, and web conference access information can be located in the **Web Conference – Q & A** area on the left menu of the online classroom.

COMMUNICATION POLICY

You may communicate with the instructor by email. The instructor will respond within 48 hours.

Assessment feedback will be provided within one week of each assessment due date.

E-CULTURE POLICY

All official communication, notices, and announcements will be distributed through e-mail (to the account you registered for the course with) via Canvas. You are accountable for checking your e-mail account regularly and for all course communication sent to it.

Students are responsible for reading “*Netiquette*” which is located under **Course Information** area on the Canvas site. *Netiquette* provides simple guidelines for civil on-line discourse & behavior, that participants are to follow and expect of one another.

ACADEMIC ETHICS POLICY

In this course, a fundamental principle is the emphasis on individual learning and achievement. Sharing work, in part or in whole, is not acceptable under any circumstances. However, we encourage active engagement with your peers, such as asking questions and studying together, to enhance your understanding of the material. Nevertheless, assignments are expected to be completed independently, and all work submitted must be your own. Any violations of this policy, including unauthorized collaboration, may result in penalties ranging from the loss of credit for submitted assignments to the extreme measure of removal from the Genetic Assistant Training Program. It is crucial that all students adhere to these guidelines to ensure the integrity and fairness of the learning experience.

This course places a strong emphasis on integrity and honesty, requiring learners to uphold these principles at all times when carrying out assignments, taking course examinations, fulfilling their patient obligations, and interacting with others. Both learners and instructors share the responsibility to report any instances of dishonesty within the course. In our commitment to maintaining an environment of complete trustworthiness, any act of dishonesty not only undermines a learner's suitability but is also regarded as unprofessional behavior. To reinforce these core values, the honor code underscores the significance of ethics in their development as scientists. The honor code delineates the School of Medicine's expectations for learners' conduct and necessitates each learner's formal declaration of personal honor. The code, as follows, articulates our shared commitment:

"As a learner at the Johns Hopkins University School of Medicine, I pledge to be honest in:

- Course work, including examinations and all assignments;
- The reporting and presentation of research data, with proper attribution and citation; and
- Professional interactions with all members of the scientific community."

It is essential for all learners to fully embrace and adhere to these standards to ensure the highest level of integrity.

DISABILITY STATEMENT

Johns Hopkins University adheres to the policies of the Americans with Disabilities Act

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to ensure that students, employees and visitors with disabilities have equal access to university programs, facilities, technology and websites. Blackboard Learn's course management system complies with the WCAG and Section 508 guidelines for accessible websites. The full disclosure statement from Blackboard may be found on Blackboard's Accessibility [webpage](#).

For complete information on Johns Hopkins University's Disability Services, please visit the Office of Institutional Equity's [website](#).

Blackboard has security and privacy policies that allow users to control who sees their information and files. Please refer to the information on their [Privacy Center](#) website and [Blackboard Cookie Statement](#) web page.

COURSE SCHEDULE

Presentations and Learning Resources and Assessments will be explained in detail within each learning module under **Learning Modules** area in Canvas.

At the start of the semester, the first two learning modules (Modules 1 & 2) will be open for your review. Then, beginning with Module 3, each learning module will open on the Sunday night (by 5 p.m. ET) prior to the module start date.

Unless otherwise noted, **all assessments are due at 11:59 p.m. ET on the date noted.**

Permission for late submission of assignments must be requested from instructor before the due date under special circumstances.

Module	Topic(s)	Presentation & Learning Resources	Assessments
Module 1: Basic Cell Biology and Genetic Concepts – Part 1	<ul style="list-style-type: none"> • Basic cell biology • Basic genetics • Chromosomes • Meiosis • Mitosis • DNA structure • DNA replication • Transcription • Translation • Gene Expression 	<p>Presentations Module 1 Presentations</p> <p>Learning Resources Basic Biology. (n.d.). Introduction to genetics. Hamilton, New Zealand: Author. Retrieved June 3, 2019 from https://basicbiology.net/biology-101/introduction-to-genetics</p> <p>Centers for Disease Control and Prevention. (n.d.). Public health genomics: Genetics basics. Atlanta, GA: Author. Retrieved June 3, 2019 from https://www.cdc.gov/genomics/about/basics.htm</p>	<p>Quiz Due: Sunday, Feb 11</p> <p>Course Introduction Survey Due: Sunday, Feb 11</p>
Module 2: Basic Cell Biology and	<ul style="list-style-type: none"> • Genome structure and function • Types of genetic 	<p>Presentations Module 2 Presentations</p>	<p>Quiz Due: Sunday, Feb 18</p>

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Module	Topic(s)	Presentation & Learning Resources	Assessments
Genetic Concepts – Part 2	variation <ul style="list-style-type: none"> • Nomenclature • Repair Pathways • Mitochondrial DNA 	Learning Resources Basic Biology. (n.d.). Introduction to genetics . Hamilton, New Zealand: Author. Retrieved June 3, 2019 from https://basicbiology.net/biology-101/introduction-to-genetics Centers for Disease Control and Prevention. (n.d.). Public health genomics: Genetics basics . Atlanta, GA: Author. Retrieved June 3, 2019 from https://www.cdc.gov/genomics/about/basics.htm	
Module 3: Genetic Services	<ul style="list-style-type: none"> • Reasons for referral • What happens at a genetic clinic visit? • What is genetic counseling? • Who works in a genetics clinic? • What are urgent referrals in genetics? 	Presentations Module 3 Presentations Learning Resources National Society of Genetic Counselors (NSGC). (n.d.). About genetic counselors . Chicago, IL: Author. Retrieved June 18, 2019 from https://www.nsgc.org/page/aboutgeneticcounselors ACMG Board of Directors. (2015). Scope of practice: A statement of the American College of Medical Genetics and Genomics (ACMG) . <i>Genetics in Medicine</i> , 17(9), e3.	Quiz Due: Sunday, Feb 25
Module 4: Genetic Testing through the	<ul style="list-style-type: none"> • Prenatal diagnostic testing and screening • Newborn 	Presentations Module 4 Presentations Learning Resources	Quiz Due: Sunday, March 3

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Module	Topic(s)	Presentation & Learning Resources	Assessments
Lifespan	screening <ul style="list-style-type: none"> • Newborn & Pediatrics • Adult Genetic • Predictive/ presymptomatic testing • Post-mortem testing 	<p><i>Newborn Screening</i> Health Resources & Services Administration (HRSA). (2019). Advisory Committee on Heritable Disorders in Newborns and Children: Recommended uniform screening panel. Rockville, MD: Author.</p> <p><i>Universal Carrier Screening</i> American College of Obstetricians and Gynecologists (ACOG). (2017. February 27). ACOG recommends offering additional carrier screening to all women, regardless of ethnicity or family history [News release]. Washington, DC: Author.</p> <p><i>Noninvasive Prenatal Diagnosis</i> National Institutes of Health U.S. National Library of Medicine. (n.d.). Genetic home reference: What is noninvasive prenatal testing (NIPT) and what disorders can it screen for? Bethesda, MD: Author. Retrieved June 18, 2019 from https://ghr.nlm.nih.gov/primer/testing/nipt</p> <p><i>Genetic Testing for Cancer</i> American Cancer Society. (2018). Family cancer syndrome. Atlanta, GA: Author.</p> <p><i>Post-Mortem Testing</i> National Society of Genetic Counselors (NSGC). (2018). Postmortem genetic testing FAQs. Chicago, IL: Author.</p>	

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Module	Topic(s)	Presentation & Learning Resources	Assessments
Module 5: Genetic Testing Technologies – Part 1	<ul style="list-style-type: none"> • Cytogenetic testing technologies • Molecular testing technologies • Alternative molecular techniques 	<p>Presentations Module 5 Presentations</p> <p>Learning Resources Howard Hughes Medical Institute (HHMI) BioInteractive. (n.d.). Sanger sequencing. Chevy Chase, MD: Author. Retrieved June 18, 2019 from https://www.biointeractive.org/classroom-resources/sanger-sequencing</p>	<p>Quiz Due: Sunday, March 10</p>
Module 6: Genetic Testing Technologies – Part 2	<ul style="list-style-type: none"> • Targeted variant testing • Single gene testing • Panel testing • Exome sequencing • Genome sequencing • Sample origin and types • Diagnostic utility • Clinical and lab sensitivity and specificity • Research and clinical testing 	<p>Presentations Module 6 Presentations</p> <p>Learning Resources Biesecker, L. G., & Green, R. C. (2014). Diagnostics clinical genome and exome sequencing. <i>New England Journal of Medicine</i>, 371(12), 1170.</p>	<p>Quiz Due: Sunday, March 17</p>
Module 7: Genetic	<ul style="list-style-type: none"> • OMIM • GeneReviews 	<p>Presentations Module 7 Presentations</p>	<p>Genetic Resources Due: Sunday, March</p>

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Module	Topic(s)	Presentation & Learning Resources	Assessments
Resources	<ul style="list-style-type: none"> • GTR • UNIQUE • NSGC • ACMG • Genetic Alliance • GARD • Support Groups 	<p>Learning Resources Online Mendelian Inheritance in Man® (OMIM)</p> <p>Online Mendelian Inheritance in Man® (OMIM). (2018). How to search tutorial [Video]. Baltimore, MD: Author.</p> <p>Online Mendelian Inheritance in Man® (OMIM). (2018). MIMmatch tutorial [Video]. Baltimore, MD: Author.</p>	24
Module 8: Ethical Issues in Genetics and Genomics	<ul style="list-style-type: none"> • Genetic discrimination • Recontakt & duty to warn • Identifiability • Gene editing 	<p>Presentations Module 8 Presentations</p> <p>Learning Resources Rodriguez, L. L., Brooks, L. D., Greenberg, J. H., & Green, E. D. (2013). The complexities of genomic identifiability. <i>Science</i>, 339(6117), 275-276.</p> <p>Ross, L. F., Saal, H. M., Anderson, R. R., American Academy of Pediatrics, & American College of medical Genetics and Genomics. (2013). Technical report: Ethical and policy issues in genetic testing and screening of children. <i>Genetics in Medicine</i>, 15(3), 234-245.</p>	<p>Discussion Initial Post Due: Thursday, March 28</p> <p>Response Posts Due: Sunday, March 31</p>
Module 9: Current Trends of Genetics in the Media	<ul style="list-style-type: none"> • Direct to consumer testing • Public health issues 	<p>Presentations Module 9 Presentations</p>	<p>Discussion Initial Post Due: Thursday, April 4</p>

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Module	Topic(s)	Presentation & Learning Resources	Assessments
			Response Posts Due: Sunday, April 7
Module 10: Past, Present, and Future of Genetics and Genetic Medicine	<ul style="list-style-type: none"> • History of medicine • History of genetics • Rise of clinical DNA sequencing • Pre-symptomatic detection and prevention • Where are we going 	<p>Presentations Module 10 Presentations</p> <p>Learning Resources American Society of Human Genetics (ASHG). (n.d.). Genetics in healthcare practice. Rockville, MD: Author. Retrieved June 18, 2019 from http://www.ashg.org/policy/healthcare.shtml</p> <p>Howard Hughes Medical Institute (HHMI) BioInteractive. (n.d.). Central dogma and genetic medicine. Chevy Chase, MD: Author. Retrieved June 18, 2019 from https://www.biointeractive.org/classroom-resources/central-dogma-and-genetic-medicine</p>	<p>Discussion Initial Post Due: Wednesday, April 10</p> <p>Response Posts Due: Friday, April 12</p>