**GMS 6224 Medical Molecular Genetics (1 Cr Hr)**

**Summer 2019**

Delivery Format: Online Course
E-Learning Site (canvas) https://ufl.instructure.com/courses/

***Instructor:***

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Office: ARB RG-293 (ground floor)

Professor, Department of Molecular Genetics & Microbiology, College of Medicine

Preferred course communications: discussion board, email or by appointment

***Pre-Requisites*** Students must have basic knowledge of genetics and molecular biology.

***PURPOSE and OUTCOME***

***Course Overview***

This 5-week, 1-credit online graduate-level course will cover a variety of topics in human genetics, focusing on mutation/variant pathogenic mechanisms and applications in the context of human disease. This will provide a foundation for understanding mechanisms of human molecular pathogenesis at the gene and chromosome level, and understanding the application of genetic knowledge in modern medicine using real world examples.

***Relation to Program Outcomes:***
This course is a required, foundational course for the Precision Medicine Graduate Certificate Program, and covers information pertaining to the following overarching program competency:

* Apply current and emerging developments in genetics, genomics, pharmacogenomics, clinical informatics, value-based medicine, pathology, and ethics to integrate genomic medicine into practice.

***Course Objectives***

Develop a greater understanding of the role of the human genome in disease:

1. Describe the human genome, and modes of inheritance and factors affecting interpretation
2. Discuss the mechanisms by which DNA genetic variations contribute to human phenotypes and applications in the clinic.
3. Discuss molecular pathogentic mechanisms and implementation of techniques used to identify DNA-level high penetrance mutations.
4. Investigate pathogenesis at the RNA level
5. Discuss genetic association of multifactorial traits, and functional effects of alleles.
6. Discuss the following cancer genetics concepts: somatic mutations, oncogenes, tumor suppressors, and driver mutation theory.
7. Discuss epigenetics in hereditary disease and genetic susceptibility.

**Instructional Methods**

This is an online course that includes viewing of recorded lectures, quizzes, completion of assignments from required reading materials, and participation in an asynchronous discussion board, to assess understanding of, and provide their insight to, the course material. Each week, after students independently view recorded lectures, they should complete the online quiz for that week (30 minutes alloted from start to finish). After reading the assigned paper, they should complete the assigned questions with brief answers to demonstrate comprehension of the material. Students are also expected to initiate and contribute to the comments and questions on the Discussion Board, including from instructor prompts. Throughout all these learning activities, the instructor is available to clarify information via discussion board postings or email.

***DESCRIPTION OF COURSE CONTENT***

***Course Schedule (based on 5 week schedule) is shown in the table below.***

All assignments (in italics in table below) are due by midnight of the last day of the week.

|  |  |  |
| --- | --- | --- |
| ***Week*** | Topics | **Readings (assigned questions)** |
| ***Week 1******June 10-16*** | **lecture 1: clinical cytogenetics** **lecture 2: modes of inheritance**  | **Dib-Hajj et al., 2007, *Trends in Neuroscience*** |
| *Quiz 1*  |
| *Reading assignment*  |
| *Asynchronous Discussion Board Participation*  |
| ***Week 2******June 17-23*** | **lecture 3: factors affecting interpretation of human inheritance** | **Suri et al., 2009, *Ophthalmology*** |
| *Quiz 2*  |
| *Reading assignment*  |
| *Asynchronous Discussion Board Participation*  |
| ***Week 3******June 24-30*** | **lectures 4 and 5: molecular pathogenesis: mechanisms and interpretation** | **Keener 2018, *The Scientist*.****Sanoja et al. 2018, *Journal of Translational Genetics and Genomics*** |
| *Quiz 3*  |
| *Reading assignment*  |
| *Asynchronous Discussion Board Participation*  |
| ***Week 4******July 1-7*** | **lecture 6: pathogenesis at the RNA level****lecture 7: genetic association of multifactorial traits** | **Messiaen et al. 1999, *Genetics in Medicine*** |
| *Quiz 4*  |
| *Reading assignment*  |
| *Asynchronous Discussion Board Participation*  |
| ***Week 5******July 8-14*** | **lecture 8: cancer genetics** **lecture 9: epigenetics** | **Feinberg 2018, *New England Journal of Medicine***  |
| *Quiz 5*  |
| *Reading assignment*  |
| *Asynchronous Discussion Board Participation*  |

***Materials and Supply Fees*** N/A

***Course Materials and Technology***

***Recommended Textbooks***

There is no required textbook, but students may wish to refer to *Human Genetics and Genomics, 4th ed.* by Korf and Irons (2013, ISBN 0470654473, available online at UF libraries) or *Genetics and Genomics in Medicine* by Strachan and Goodship (2015, ISBN 0815344805) as independent resources. An older reference for the basic concepts and more-established technology, available in hard copy in the UF health science center library, is *Human Molecular Genetics, ed.4*  by Strachan and Read (2011, ISBN 815341490).

**For technical support for this class**, please contact the UF Help Desk at:

* [Learning-support@ufl.edu](file:///C%3A%5CUsers%5Chackg%5CDesktop%5CLearning-support%40ufl.edu)
* (352) 392-HELP - select option 2
* <https://lss.at.ufl.edu/help.shtml>

***ACADEMIC REQUIREMENTS AND GRADING***

***Grading****:*

A)online Quizzes will cover the material in the lectures, and will consist of 7 multiple choice questions, to be completed by end of day 7 of each week (see schedule above). The quiz will be open for 30 minutes once it has been started, so answers must be submitted by the end of 30 minutes. Quiz scores will be available within a few days.

B) Reading Assignment: students will provide written answers to questions about the assigned article, as provided in the assignment posted each week. These will be due by the end of day 7 of each week; scores will not be immediately provided but will be posted typically within the following week.

C) Discussion Board: students must follow directions in the prompt posted on the Discussion Board each week at the course website on Canvas. Students are also welcome to initiate or respond to questions or comments outside of the prompt, for any content related to the course (except quiz questions). Discussion board closes at midnight on day 7 of each week, and the score (based on participation) will be available within the following week.

Students who would like specific feedback on a graded assignment should contact the instructor.

|  |  |  |  |
| --- | --- | --- | --- |
| **Requirement** | **points** | **total points** | **Percentage of Final Grade** |
| Reading Assignments (N=5) | 70 each | 350 | 35% |
| Quizzes (N=5) | 70 each | 350 | 35% |
| Discussion Board Participation (all 5 weeks)  | 60 each | 300 | 30% |
|  |  | total 1000 | 100% |

***Grading Policy***

|  |  |  |
| --- | --- | --- |
| **Percent**  | **Grade**  | **Grade Points**  |
| 92.5 - 100.0  | A  | 4.00  |
| 89.5 – 92.49  | A-  | 3.67  |
| 86.5 – 89.49  | B+  | 3.33  |
| 82.5 – 86.49  | B  | 3.00  |
| 79.5 – 82.49  | B-  | 2.67  |
| 76.5 – 79.49  | C+  | 2.33  |
| 72.5 – 76.49  | C  | 2.00  |
| 69.5 – 72.49  | C-  | 1.67  |
| 66.5 – 69.49  | D+  | 1.33  |
| 62.5 – 66.49  | D  | 1.00  |
| 59.5 - 62.49  | D-  | 0.67  |
| < 59.50 | E  | 0.00  |

For greater detail on letter grades and university policies related to them, please see the Registrar’s Grade Policy regulations at the following websites:

<http://gradcatalog.ufl.edu/content.php?catoid=10&navoid=2020#grades>
<https://catalog.ufl.edu/ugrad/current/regulations/info/grades.aspx>

***POLICIES***

**Make-Up Policy for Quizzes, Reading Assignment submissions, and Discussion Board Participation:** Students will be allowed to make up quizzes, assignments, and discussion board activities for acceptable reasons as described in the Graduate Catalog. See: <http://gradcatalog.ufl.edu/content.php?catoid=10&navoid=2020#attendance>

Any requests for make-ups due to technical issues accessing the course website/content MUST be accompanied by the ticket number received from LSS when the problem was reported to them. The ticket number will document the time and date of the problem. You MUST e-mail the course director within 24 hours of the technical difficulty if you wish to request a make-up.

**Policy Related to Required Course Participation:**This is an online course and therefore, attendance means you are expected to complete the course learning activities so that you meet the established deadlines. Please note that all faculty are bound by the UF policy for excused absences. For information regarding the UF Attendance Policy see the Registrar website for additional details: <https://catalog.ufl.edu/ugrad/current/regulations/info/attendance.aspx>

Students who wish to drop from the course must do so by the drop/add deadline established by the Office of the University Registrar. Students must not assume they will be automatically dropped if they fail to participate in the course learning activities.

# *STUDENT EXPECTATIONS, ROLES, AND OPPORTUNITIES FOR INPUT*

# Expectations Regarding Course Behavior: Students are expected to complete course requirements, including ensuring that discussion board comments are posted on the correct discussion thread. Discussion board comments should be clearly written, relevant to the topic of discussion, use appropriate grammar and spelling, and be appropriately concise.

**Professionalism**: Students are expected to be courteous, turn in assignments on time, and to show professionalism in all active learning events and participation in this course.

**Communication Guidelines:** In all course communications, including emails and treaded discussions, students are expected to follow Netiquette Guidelines. These guidelines promote an environment that encourages everyone to ask questions and learn from each other. Discussion board posts that are not respectful of other opinions discourage a positive learning environment. The following link provides guidelines:

<http://teach.ufl.edu/wp-content/uploads/2012/08/NetiquetteGuideforOnlineCourses.pdf>

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# Academic Integrity

Students are expected to act in accordance with the University of Florida policy on academic integrity.  As a student at the University of Florida, you have committed yourself to uphold the Honor Code, which includes the following pledge:

 *“We, the members of the University of Florida community, pledge to hold ourselves and our peers to the highest standards of honesty and integrity.”*

You are expected to exhibit behavior consistent with this commitment to the UF academic community, and on all work submitted for credit at the University of Florida, the following pledge is either required or implied:

*“On my honor, I have neither given nor received unauthorized aid in doing this assignment.”*

It is your individual responsibility to know and comply with all university policies and procedures regarding academic integrity and the Student Honor Code.  Violations of the Honor Code at the University of Florida will not be tolerated.  Violations will be reported to the Dean of Students Office for consideration of disciplinary action.  For additional information regarding Academic Integrity, please see Student Conduct and Honor Code or the Graduate Student Website for additional details:

<https://sccr.dso.ufl.edu/policies/student-honor-code-student-conduct-code/>

<https://www.dso.ufl.edu/sccr/process/student-conduct-honor-code/>

<http://gradschool.ufl.edu/students/introduction.html>

Cheating, lying, misrepresentation, or plagiarism in any form is unacceptable and inexcusable behavior and will lead to sanctions.

# Online Faculty Course Evaluation Process

Students are expected to provide feedback on the quality of instruction in this course by completing an online evaluation at <https://evaluations.ufl.edu>. Students will be notified by email of specific times when this course evaluation is open. Summaries of these assessments are available to students at <https://evaluations.ufl.edu/results/>.

***SUPPORT SERVICES***

# Accommodations for Students with Disabilities

If you require classroom accommodation because of a disability, you must register with the Dean of Students Office <http://www.dso.ufl.edu> within the first week of class. The Dean of Students Office will provide you a letter documenting accommodations for you, which you must then give to the Instructor of the course to receive the accommodations. Please make sure you provide this letter to the Instructor by the end of the second week of the course. UF is committed to providing reasonable accommodations to assist students in their coursework.

# Counseling and Student Health

Students sometimes experience stress from academic expectations and/or personal and interpersonal issues that may interfere with their academic performance. If you find yourself facing issues that have the potential to or are already negatively affecting your coursework, you are encouraged to talk with the Instructor and/or seek help through University resources available to you:

* The Counseling and Wellness Center 352-392-1575 offers a variety of support services such as psychological assessment and intervention and assistance for math and test anxiety. Visit their web site for more information: <http://www.counseling.ufl.edu>. Online and in-person assistance is available.
* You Matter We Care website: <http://www.umatter.ufl.edu/>. If you are feeling overwhelmed or stressed, you can reach out for help through the You Matter We Care website, which is staffed by Dean of Students and Counseling Center personnel.
* The Student Health Care Center at Shands is a satellite clinic of the main Student Health Care Center (the latter of which is located at 280 Fletcher Drive on campus, 392-1161). Both sites offer a variety of clinical services. The Shands clinic is located on the second floor of the Dental Tower in the Health Science Center (D2-49, 294-5700). For more information, go to the web site: [shcc.ufl.edu/](https://shcc.ufl.edu/)
* Crisis intervention is always available 24/7 from Alachua County Crisis Center: (352) 264-6789
<http://www.alachuacounty.us/DEPTS/CSS/CRISISCENTER/Pages/CrisisCenter.aspx>

Do not wait to reach a crisis before getting assistance. Many students have been helped through stressful situations impacting academic performance. You are not alone, so don't hesitate to ask for help.

***Software Use***

All faculty, staff, and students of the University are required and expected to obey the laws and legal agreements governing software use. Failure to do so can lead to monetary damages and/or criminal penalties for the individual violator. Because such violations are also against University policies and rules, disciplinary action will be taken as appropriate. We, the members of the University of Florida community, pledge to uphold ourselves and our peers to the highest standards of honesty and integrity.

***Student Privacy***

Federal laws protect your privacy in regard to grades earned in courses and on individual assignments. For more information, please see: <http://registrar.ufl.edu/catalog0910/policies/regulationferpa.html>

***Campus Academic Resources:***

**E-learning technical suppor***t*, 352-392-4357 (select option 2) or e-mail to Learning-support@ufl.edu. <https://lss.at.ufl.edu/help.shtml>.

**Career Resource Center**, Reitz Union, 392-1601. Career assistance and counseling. <https://www.crc.ufl.edu/>.

**Library Support**, <http://cms.uflib.ufl.edu/ask>. Various ways to receive assistance with respect to using the libraries or finding resources.

**Teaching Center**, Broward Hall, 392-2010 or 392-6420. General study skills and tutoring. <https://teachingcenter.ufl.edu/>.

**Writing Studio, 302 Tigert Hall***,* 846-1138. Help brainstorming, formatting, and writing papers. <https://writing.ufl.edu/writing-studio/>.

**Student Complaints Campus***:* <https://www.dso.ufl.edu/documents/UF_Complaints_policy.pdf>.

**On-Line Students Complaints***:* <http://www.distance.ufl.edu/student-complaint-process>.