

# BIOL 3302

## MEDICAL GENETICS

### Fall 2021

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## Instructor: Dr. Laurel Fohn, MD, PhD

Email: [lfohn1@angelo.edu](mailto:lfohn1@angelo.edu)

Phone: 325-486-6644

Office: Cavness 107

**Office Hours:** Posted on office door and on first day of class. Additional office hours will be by appointment only. All office hours/ appointments will occur in a socially distanced manner and may occur via telephone, Blackboard Collaborate, outdoors, etc.

**Attendance:** There will be assigned seating and roll is taken at each class meeting for both in-person and remote students and factors into your grade as described below.

## Course Information

### Course Description

The goals of this course are to: [1] provide students with factual knowledge about the field of medical genetics; to introduce fundamental principles of the field. These represent progress points 1 & 2 on the IDEA course evaluation form to be filled out at the end of the semester.

### Prerequisite and Co-requisite Courses

Must have completed Biol 3301 with a grade of "A" or with special permission from the instructor.

### Prerequisite Skills

Proficiency in accessing and utilizing ASU Blackboard platform, internet websites, using ASU Library resources, and proficiency with Microsoft Word and/or PowerPoint are expectations of the course.

## Student Learning Outcomes

Upon completion of this course, students should:

1. understand major mechanisms of inheritance of genetic disorders
2. understand the applications of genetics in a broad array of medical disorders and diagnosis
3. be able to apply this understanding to unknown situations
4. begin understanding unusual mechanisms of genetic disease

## Course Delivery- Face to Face with Synchronous Remote Sessions

Classic lecture style utilizing Synchronous Remote Sessions supplemented with transparencies/figures/ powerpoints. Students will be assigned textbook readings as well as supplemental reading/ viewing material online. This course is designed for upper level science majors particularly those with health professions interests.

To maintain academic quality while accommodating quarantine/ isolation needs this semester, this course will use a split delivery model that combines face-to-face teaching with livestream instruction.

The goal is to provide face-to-face instruction to students who are eligible (no illness, no symptoms of illness, and no contact with COVID19), while also allowing students who may need to learn remotely to participate via virtual class sessions for lecture portions of class. These sessions may be recorded.

### How Does It Work?

When you are not in the physical class, you will attend live remote sessions at the same time as our scheduled course. You will also be expected to complete coursework via Blackboard.<sup>1</sup>

Please refer to this Health and Safety web page<sup>2</sup> for updated information about campus guidelines as they relate to the COVID-19 pandemic.

## Required Texts and Materials

**Textbook:** B. A. Pierce, "Genetics: A Conceptual Approach". W.H. Freeman and Co. 6<sup>th</sup> Edition. **ISBN-13:** 9781319050962 or **ISBN-10:** 1319050964

## **Technology/ Communication Requirements**

**Technology:** Students will need to utilize and check their **Angelo State University email** account for all email communications with the instructor and for course announcements. Additionally, students should maintain the ability to utilize **Blackboard** to access grades, reading assignments, synchronous lectures via Collaborate, exams, quizzes, assessments, and other course material.

To successfully complete this course, students need to have access to a **smart phone, reliable internet, and a computer (a PC or Mac computer/ laptop or iPad) with webcam** capable of utilizing **Respondus Lockdown Browser and Respondus Monitor**.

Access to some exams and quizzes may be through Blackboard and Respondus Lockdown Browser and will be video recorded via Respondus Monitor. Respondus requires a desktop computer or laptop (not a Chromebook) and a webcam. For best results, use an ethernet cable to connect to your Internet source instead of relying on Wifi. Refer to the Blackboard course for Respondus installation instructions.

This class will utilize Blackboard Collaborate (which requires a web cam).

**Communication:** Faculty will respond to email and/or telephone messages within 2 business days (Monday through Friday) during working hours.

All private email communication will be done exclusively through your ASU email address. Check frequently for announcements and policy changes. In your emails to faculty, include the course name and section number in your subject line.

**Virtual communication:** Office hours and/or advising may be done with the assistance of the telephone, Collaborate, Skype, etc.

## **Assessment/ Grading/Assignment and Activity Descriptions:**

Students will be assessed on the above objectives through exams and homework as detailed below. Course grades will be determined as indicated in the table below.

### **Evaluation and Grades-**

There will be three exams, one presentation, and various homework assignments each worth the indicated points. There will be additional points available in the homework/ quiz section to offset a single absence or poor grade, or to serve as bonus points.

Date	Assessment	Percent of Total Grade
9/21	Exam 1 (100 points)	20%
11/2	Exam 2 (100 points)	20%
November Dates	Presentation (100 points)	20%
Thurs. Dec. 9 10:30-12:30	Exam 3- Comprehensive Final (125 points)	25%
Various	Homework and Quizzes (75 points- up to 100 points available)	15% (with possibility of 5% bonus)
	*Note: Portions of all exam grades above may be derived from homework assignments.	

**NOTE: November 22, 2021** is currently listed as the **last day to Drop/Withdraw from class without a grade** (ie. these students will receive a W on their transcript). **Any student remaining on the official role after this date will receive a grade.**

## Grading System

Course grades will be dependent upon completing course requirements and meeting the student learning outcomes.

The following grading scale is in use for this course: 500 points total

- A = 90.0-105 percent (450+ points)
- B = 80.0-89.9 percent (400-449 points)
- C = 70.0-79.9 percent (350-399 points)
- D = 60.0-69.9 percent (300-349 points)
- F = 0-59.9 percent (299 or fewer points)

## Assignment and Activity Descriptions (Exams, Attendance, Presentations, etc.):

### EXAMS

On exam days, exams will be given during the normally allocated lecture time. All exams will be turned in promptly at the end of the period. Extra time will not be allocated for finishing the exam unless academic accommodations have been previously applied for/ made. Exams will be proctored either in person or online using Respondus Lockdown Browser and Monitor. No learning materials (notes, books, handouts, etc) or electronic devices (other than the exam device), including calculators, are permitted on desktops/ surrounding exam areas during exams and no

assistance from any other being is permitted.

Exams will be composed of practical questions arising from lecture material, readings, or presentations. Exams will contain multiple choice, short answer, matching, essay and application/ clinical case study questions. While approximately 50% of questions will be based on factual recall, the remaining 50% of each exam will be composed of questions that will require a “synthesis” of information or an “application” of knowledge to solve an unknown situation.

**No make-up exams will be administered.** Therefore, attendance is imperative. In the event of an excused absence/ special extenuating circumstance, arrangements should be made prior to the absence. In the event of an emergency, notification should be made as soon as possible with documentation and a mutually agreeable alternative will be determined.

#### NOTE for exams:

- Additional reading assignments/ study guides may be given during lecture or posted to blackboard. Students are responsible for this information as well as that presented in class/ textbook. You may AUDIO tape (but NOT VIDEO OR PHOTOGRAPH lectures/slides).

- Assignments, take-home review questions, homework, etc. should be used to assist your study for exams. Some of these questions/assignments will be designed to test your understanding of genetic principles and their application in medical genetics and may include clinical/case-study questions.

→ You may use any resource available to you to answer these homework/ assignment questions- textbook, reliable internet sources, publications, the library, or colleagues. However, these are take-home assignments-not take-Dr. Fohn assignments- I will not answer these questions for you.

#### Attendance:

Lectures-Students may attend lectures at their own discretion; however, in accord with University policy, attendance will be taken at each class meeting. Attendance records are maintained for administrative reasons and will not be a direct negative factor in determining your final grade. Note, however, consistent lecture attendance generally proves beneficial to successful examination performance. It is highly unlikely that persons with sporadic attendance will be able to maintain a successful level of performance; they might find it more profitable therefore to devote their time and energy to other endeavors. The exceptions related to attendance and a negative factor on your grade is for presentations, **your attendance at your and your classmates presentations will factor into your presentation grade.**

Exams- Students are required to take all exams and to complete all assignments on time. If you miss an exam for any reason you will receive a grade of zero (0) unless prior arrangements have been made or unless an emergency situation is documented.

Exceptions will be made only in rare and unusual circumstances.

## **Student Presentations**

Each student will be assigned a presentation topic which will cover a particular medical genetic disorder. Students will be assigned a topic and presentation date. Each student will be responsible for researching/organizing/presenting a 25-30 minute PowerPoint to the class. The student will give a case study, background information (with scientific literature references) about the disease, address relevant genetic topics, and discuss how the disease is diagnosed and potentially treated. They will also prepare a summary and review questions for their topic. Presentation assignments (and more extensive instructions) will be made following the first lecture exam. A portion of the student's presentation grade will come from attending and participating (asking questions, being involved in discussions) in their classmates presentations.

## **General Policies Related to This Course**

All students are required to follow the policies and procedures presented in these documents:

- [Angelo State University Student Handbook](#)<sup>3</sup>
- [Angelo State University Catalog](#)<sup>4</sup>

## **Academic Integrity**

Students are expected to maintain complete honesty and integrity in all work. Any student found guilty of any form of dishonesty in academic work is subject of disciplinary action and possible expulsion from ASU.

The College of Science and Engineering adheres to the university's [Statement of Academic Integrity](#).<sup>5</sup>

## **Accommodations for Students with Disabilities**

ASU is committed to the principle that no qualified individual with a disability shall, on the basis of disability, be excluded from participation in or be denied the benefits of the services, programs or activities of the university, or be subjected to discrimination by the university, as provided by the Americans with Disabilities Act of 1990 (ADA), the Americans with Disabilities Act Amendments of 2008 (ADAAA) and subsequent

legislation. **Please note: I must have all documentation at least one week prior to requested accommodations being implemented. It is the responsibility of the student to remind me and work with me prior to each exam/ assignment requiring accommodations to make this process as effective as possible.**

Student Disability Services is located in the Office of Student Affairs, and is the designated campus department charged with the responsibility of reviewing and authorizing requests for reasonable accommodations based on a disability. It is the student's responsibility to initiate such a request by contacting an employee of the Office of Student Affairs, in the Houston Harte University Center, Room 112, or contacting the department via email at [ADA@angelo.edu](mailto:ADA@angelo.edu). For more information about the application process and requirements, visit the [Student Disability Services website](#).<sup>6</sup> The employee charged with the responsibility of reviewing and authorizing accommodation requests is:

Dr. Dallas Swafford  
Director of Student Disability Services  
Office of Student Affairs  
325-942-2047  
[dallas.swafford@angelo.edu](mailto:dallas.swafford@angelo.edu)  
Houston Harte University Center, Room 112

## **Incomplete Grade Policy**

It is policy that incomplete grades be reserved for student illness or personal misfortune. Please contact faculty if you have serious illness or a personal misfortune that would keep you from completing course work. Documentation may be required. See ASU Operating Policy 10.11 [Grading Procedures](#)<sup>7</sup> for more information.

## **Plagiarism**

Plagiarism is a serious topic covered in ASU's [Academic Integrity policy](#)<sup>8</sup> in the Student Handbook. Plagiarism is the action or practice of taking someone else's work, idea, etc., and passing it off as one's own. Plagiarism is literary theft.

In your discussions and/or your papers, it is unacceptable to copy word-for-word without quotation marks and the source of the quotation. It is expected that you will summarize or paraphrase ideas giving appropriate credit to the source both in the body of your paper and the reference list.

Papers are subject to be evaluated for originality. Resources to help you understand this policy better are available at the [ASU Writing Center](#).<sup>9</sup>

## **Student Absence for Observance of Religious Holy Days**

A student who intends to observe a religious holy day should make that intention known in writing to the instructor prior to the absence. See ASU Operating Policy 10.19 [Student Absence for Observance of Religious Holy Day](#)<sup>10</sup> for more information.

## **Title IX at Angelo State University**

Angelo State University is committed to providing and strengthening an educational, working, and living environment where students, faculty, staff, and visitors are free from sex discrimination of any kind. In accordance with Title VII, Title IX, the Violence Against Women Act (VAWA), the Campus Sexual Violence Elimination Act (SaVE), and other federal and state laws, the University prohibits discrimination based on sex, which includes pregnancy, and other types of Sexual Misconduct. Sexual Misconduct is a broad term encompassing all forms of gender-based harassment or discrimination and unwelcome behavior of a sexual nature. The term includes sexual harassment, nonconsensual sexual contact, nonconsensual sexual intercourse, sexual assault, sexual exploitation, stalking, public indecency, interpersonal violence (domestic violence or dating violence), sexual violence, and any other misconduct based on sex.

You are encouraged to report any incidents involving sexual misconduct to the Office of Title IX Compliance and the Director of Title IX Compliance/Title IX Coordinator, Michelle Miller, J.D. You may submit reports in the following manner:

Online: [Incident Reporting Form](#)<sup>11</sup>

Face to Face: Mayer Administration Building, Room 210

Phone: 325-942-2022

Email: [michelle.miller@angelo.edu](mailto:michelle.miller@angelo.edu)

Note, as a faculty member at Angelo State, I am a mandatory reporter and must report incidents involving sexual misconduct to the Title IX Coordinator. Should you wish to speak to someone in confidence about an issue, you may contact the University Counseling Center (325-942-2371), the 24-Hour Crisis Helpline (325-486-6345), or the University Health Clinic (325-942-2171).

For more information about resources related to sexual misconduct, Title IX, or Angelo State's policy please visit the [Title IX website](#).<sup>12</sup>

## **Information About COVID-19**

Please refer to ASU's [COVID-19 \(Coronavirus\) Updates](#)<sup>13</sup> web page for current information about campus guidelines and safety standards as they relate to the COVID-19 pandemic.



You **are required to complete the daily wellness screen** each day prior to attending class. Faculty members may also ask you to display your daily screening badge. If you are not feeling well, please complete the screen and do not return to class until you are cleared for return through the system.

Current research on the COVID-19 virus suggests there is a significant reduction in the potential for transmission of the virus from person to person by wearing a mask/facial covering that covers the nose and mouth areas. Therefore, as of the start of the semester, the CDC and I highly recommend and encourage the wearing of an appropriate mask/facial covering indoors before, during, and after class. You are also asked to maintain safe distancing practices, hygiene, and hand washing to the best of your ability.

**No food or drink may be consumed in the classroom.**

## Modifications to the Syllabus

This syllabus, including grade evaluation and course schedule, is subject to modification on potentially short notice based on developing circumstances. In particular, the COVID-19 pandemic may require significant changes in course delivery and content on potentially short notice.

## Course Schedule

Week/Date	Topic	Special Applications	Due Date
1. Aug 24,26	Syllabus/ Intro to Med. Genetics		
2. Aug 31, Sept 2	Review/ Mitosis & Meiosis		
3. Sept 7, 9	Review Chromosomes/ transcription/ translation/ Protein structure		
4. Sept 14, 16	Types of Mutations & causes (chromosomal, molecular, germline, somatic, LOF/GOF, dominant/ recessive/ dominant negative, etc)	OMIM PubMed NCBI Blast	
5. Sept 21, 23	<b>Exam 1</b> / Genetic Analysis- Macroscopic Medical evaluation/ genetic counseling		<b>Sept 21</b>

Week/Date	Topic	Special Applications	Due Date
6. Sept 28, 30	Genetic Analysis- Modes of Transmission (typical and special situations including anticipation, triplet repeats, imprinting, mosaicism, mitochondrial inheritance)	Pedigrees	
7. Oct 5, 7	Genetic Analysis- Chromosomal	Karyotype FISH CGH	
8. Oct 12, 14	Genetic Analysis- Molecular	RFLP; Southern Blot; PCR	
9. Oct 19, 21	Genetic Analysis- Molecular	MSI; SNP analysis; WGS	
10. Oct 26, 28	Applications: Prenatal dx/ IVF/ post-natal/ telomeres/ viral integration/ cancer/ treatments (vaccines/ gene therapy/ cancer diagnostics/ personalized medicine)		
11. Nov 2, 4	<b>Exam 2</b> Presentations 1-2	Likely topics: CF; Hemophilia; Sickle Cell/ Thalassemia; Collagen Vascular Disease	<b>Nov 2</b>
12. Nov 9, 11	Presentations 3-6	Fragile X, Prader Willi, Mosaic Down's Syndrome	
13. Nov 16, 18	Presentations 7-10	HNPCC; MSI-high/low	
14. Nov 23/ <b>25</b> <b>Thanksgiving</b>	Presentation 11-12 <b>25 is Holiday</b>	Mitochondrial Diseases	
15. Nov 30 Dec. 2	Presentations 13-14; Review	Cancer Diagnosis using FISH; Personalized Medicine	
16. Dec.9- Thurs: 10:30am to 12:30 pm	<b>FINAL EXAM</b>		<b>Dec. 9</b>

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- 1 <https://blackboard.angelo.edu/>
  - 2 <https://www.angelo.edu/covid-19/returning-to-campus/health-and-safety.php>
  - 3 <https://www.angelo.edu/current-students/student-handbook/>
  - 4 <https://www.angelo.edu/academics/catalog/>
  - 5 <https://www.angelo.edu/live/files/27603-student-handbook-2020-21#page=96>
  - 6 <https://www.angelo.edu/current-students/disability-services/>
  - 7 <https://www.angelo.edu/content/files/14197-op-1011-grading-procedures>
  - 8 <https://www.angelo.edu/live/files/27603-student-handbook-2020-21#page=96>
  - 9 [https://www.angelo.edu/current-students/writing-center/academic\\_honesty.php](https://www.angelo.edu/current-students/writing-center/academic_honesty.php)
  - 10 <https://www.angelo.edu/content/files/14206-op-1019-student-absence-for-observance-of>
  - 11 <https://www.angelo.edu/incident-form>
  - 12 <https://www.angelo.edu/title-ix>
  - 13 <https://www.angelo.edu/covid-19/>