

Course ID:	Course Title:	Winter 2020
BIO 327	Medical Genetics	Prerequisite: BIO 211
		Credits: 3

Class Information		Instructor Information		Important Dates	
Days:	Tuesday and Thursday	Instructor:	Dr. Chris Wang	First day of classes:	Tue, Jan 7
Time:	16:00 – 17:15	Email:	chris.wang@ambrose.edu	Last day to add/drop, or change to audit:	Sun, Jan 19
Room:	A2210	Phone:	(403) 410-2000 ext. 6910	Last day to request revised exam:	Mon, Mar 9
Lab/ Tutorial:	N/A	Office:	L2113	Last day to withdraw from course:	Fri, Mar 20
	N/A	Office Hours:	by appointment (open door policy)	Last day to apply for coursework extension:	Mon, Mar 30
Final Exam:				Last day of classes:	Thurs, Apr 9

Course Description

This course studies and applies the principles of genetics in human and medical context. The topics include: Mendelian and multifactorial inheritance, linkage and gene mapping, pedigree analysis, molecular basis of diseases and screening, cytogenetics and developmental genetics, genomic imprinting, population and cancer genetics, gene therapy, and ethical issues.

Expected Learning Outcomes

Upon successful completion of this course, students will be able to

1. provide an understanding of the genetic basis of human disease and current approaches to diagnose, treat, and prevent the genetic disorders
2. relate the study of human genetics to major advances in molecular, biochemical, and cellular genetics
3. provide an awareness of the relationship of the individual genome to the genetic makeup of the human population and its ramifications to human diversity
4. relate human genetics to current issues in genetic screening, genetic counseling and genetic engineering

Textbooks (Recommended)

- Medical Genetics. L. Jorde, J. Carey and M. Bamshad. 5th Edition. 2016. Elsevier.
- Thompson & Thompson Genetics in Medicine. Robert L. Nussbaum, Roderick R. McInnes, and Huntington F Willard. 8th Edition. 2015. Elsevier. (a copy of 7th edition is reserved in the library)
- Lewin's Genes XII. Jocelyn E. Krebs, Elliott S. Goldstein, and Stephen T. Kilpatrick. 12th Edition. 2017. Jones & Bartlett Learning. (a copy of chapter 27 and 28 is reserved in the library)

Course Schedule

Date	Topic	Reading
Jan. 07	Introduction to the Course	
Jan. 09	Topic 1 - Introduction to Human Genome	Ch. 1 and 2
Jan. 14	Topic 2 - Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Jan. 16	Topic 2 - Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Jan. 21	Topic 2 - Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Jan. 23	Topic 2 - Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Jan. 28	Topic 2 - Principal of Clinical Cytogenetics and Genome Analysis	Ch. 6
Jan. 30	<i>Program Day (No Class)</i>	
Feb. 04	Topic 3 - The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Feb. 06	Topic 3 - The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Feb. 11	In-Class Mid-term Exam # 1	
Feb. 13	Topic 3 - The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Feb. 18	<i>Winter Reading Week (No Class)</i>	
Feb. 20	<i>Winter Reading Week (No Class)</i>	
Feb. 25	Topic 3 - The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Feb. 27	Topic 4 - The Molecular, Biochemical, and Cellular Basis of Genetic Disease	Ch. 7
Mar. 03	Topic 4 - The Molecular, Biochemical, and Cellular Basis of Genetic Disease	Ch. 7
Mar. 05	Topic 5 - Genetic Variation, Mutation, and Polymorphism	Ch. 3
Mar. 10	Topic 5 - Genetic Variation, Mutation, and Polymorphism	Ch. 3
Mar. 12	Preparation for Ambrose Research Conference (ARC) presentation on March 25 th	
Mar. 17	Topic 6 - Cancer Genetics	Ch. 11
Mar. 19	In-Class Mid-term Exam # 2	
Mar. 24	Topic 6 - Cancer Genetics	Ch. 11
Mar. 26	Topic 7 - Epigenetics	Lewin's Genes XII Ch. 27 - 28
Mar. 31	Topic 7 - Epigenetics	Lewin's Genes XII Ch. 27 - 28
Apr. 02	Topic 8 - Multifactorial Inheritance and Common Disease	Ch. 12
Apr. 07	Topic 8 - Multifactorial Inheritance and Common Disease	Ch. 12
Apr. 09	Topic 9 – Genetics and Precision Medicine	Ch. 14

Requirements:

- no late assignment will be accepted unless pre-arrangement was made or evidence of legitimate excuses was presented

Evaluation:

Evaluation Methods	Due Date	Weighting
Midterm Exam 1	Feb. 11 th	20%
Midterm Exam 2	Mar. 19 th	20%
Disease-Gene Identification Assignment (Chapter 8)		5%
ARC Presentation	Mar. 25 th	10%
Research Proposal	Apr. 2 nd	10%
Final Exam (Cumulative)		35%

Attendance:

- students are required to attend all the scheduled lectures and evaluation sessions
- a medical note (original and dated) is required to justify for the absence
- ARC attendance is required

Grade Summary:

Percent (%) to Letter Grade Conversion	Grade	Grade Point	Description
92.00% - 100%	A+	4.0	Excellent
85.00% - 91.99%	A	4.0	
80.00% - 84.99%	A-	3.7	
77.00% - 79.99%	B+	3.3	Good
73.00% - 76.99%	B	3.0	
70.00% - 72.99%	B-	2.7	
67.00% - 69.99%	C+	2.3	Satisfactory
63.00% - 66.99%	C	2.0	
60.00% - 62.99%	C-	1.7	
55.00% - 59.99%	D+	1.3	Minimal Pass
50.00% - 54.99%	D	1.0	
00.00% - 49.99%	F	0	Fail

Because of the nature of the Alpha 4.00 system, there can be no uniform University-wide conversion scale. The relationship between raw scores (e.g. percentages) and the resultant letter grade will depend on the nature of the course and the instructor's assessment of the level of each class, compared to similar classes taught previously.

Please note that final grades will be available on student registration system. Printed grade sheets are not mailed out.

Other:

Potential genetic disorders to be presented in ARC:

- selection of topic is on *first-come, first-serve* basis (topic selection due on **Jan. 23rd** (1 of the 10% project mark)
- poster presentation in ARC on **March 25th**
- each topic can only be chosen once
- only topics that are not discussed in lectures can be selected

_____ Abacavir-Induced Stevens-Johnson Syndrome

_____ Achondroplasia

_____ Age-Related Macular Degeneration

_____ Alzheimer Disease

_____ Autism/16p11.2 Deletion Syndrome

_____ Beckwith-Wiedemann Syndrome

_____ Hereditary Breast and Ovarian Cancer

_____ Charcot-Marie-Tooth Disease Type 1A

_____ CHARGE Syndrome

_____ Chronic Myelogenous Leukemia

_____ Crohn Disease

_____ Cystic Fibrosis

_____ Deafness (Nonsyndromic)

_____ Duchenne Muscular Dystrophy

_____ Familial Adenomatous Polyposis

_____ Familial Hypercholesterolemia

_____ Fragile X Syndrome

_____ Type I (Non-neuronopathic) Gaucher Disease

_____ Glucose-6-Phosphate Dehydrogenase Deficiency

_____ Hereditary Hemochromatosis

_____ Hemophilia

_____ Hirschsprung Disease

_____ Holoprosencephaly (Nonsyndromic Form)

_____ Hypertrophic Cardiomyopathy

_____ Insulin-Dependent (Type 1) Diabetes

_____ Intrauterine Growth Restriction

_____ Long QT Syndrome

_____ Lynch Syndrome

_____ Marfan Syndrome

_____ Medium-Chain Acyl-CoA Dehydrogenase Deficiency

_____ Miller-Dieker Syndrom

_____ Myoclonic Epilepsy with Ragged Red Fiber

_____ Neurofibromatosis

_____ Non-Insulin-Dependent (Type 2) Diabetes

_____ Ornithine Transcarbamylase Deficiency

_____ Polycystic Kidney Disease

_____ Sex Development Disorder (46, XX Male)

_____ Sickle Cell Disease

_____ Thalassemia

_____ Thiopurine S-Methyltransferase Deficiency

_____ Thrombophilia

_____ Tuberous Sclerosis

_____ Others

<https://rarediseases.info.nih.gov/diseases/diseases-by-category/5>

Contents for Poster Presentation:

- history and physical finding
- disease etiology and incidence
- pathogenesis
- phenotypes
- inheritance risk
- diagnosis
- other related diseases
- management and treatments
- prevention

Research Proposal:

- based on the topic chosen for ARC presentation
- no online (hyperlink) or Wikipedia references allowed
- citing *at least 5 primary literatures no older than 2015*
- limiting the writing to 5 double-spaced pages, *excluding* reference list and figures, but including:
 - questions to be answered
 - hypothesis/hypotheses
 - objectives
 - significance of the research
 - layout of 3 proposed experiments and their expected outcomes
 - a conclusion section

Ambrose University Academic Policies:

Communication

All students have received an Ambrose e-mail account upon registration. It is the student's responsibility to check this account regularly as the Ambrose email system will be the professor's instrument for notifying students of important matters (cancelled class sessions, extensions, requested appointments, etc.) between class sessions. If students do not wish to use their Ambrose accounts, they will need to forward all messages from the Ambrose account to another personal account.

Registration

During the **Registration Revision Period** students may enter a course without permission, change the designation of any class from credit to audit and /or voluntary withdraw from a course without financial or academic penalty or record. Courses should be added or dropped on the student portal by the deadline date; please consult the List of Important Dates. After that date, the original status remains and the student is responsible for related fees.

Students intending to withdraw from a course after the Registration Revision Period must apply to the Office of the Registrar by submitting a "Request to Withdraw from a Course" form or by sending an email to the Registrar's Office by the **Withdrawal Deadline**; please consult the List of Important Dates on the my.ambrose.edu website. Students will not receive a tuition refund for courses from which they withdraw after the Registration Revision period. A grade of "W" will appear on their transcript.

Students wishing to withdraw from a course, but who fail to do so by the applicable date, will receive the grade earned in accordance with the course syllabus. A student obliged to withdraw from a course after the Withdrawal Deadline because of health or other reasons may apply to the Registrar for special consideration.

Exam Scheduling

Students, who find a conflict in their exam schedule must submit a Revised Examination Request form to the Registrar's Office by the deadline date; please consult the List of Important Dates. Requests will be considered for the following reasons only: 1) the scheduled final examination slot conflicts with another exam; 2) the student has three final exams within three consecutive exam time blocks; 3) the scheduled final exam slot conflicts with an exam at another institution; 4) extenuating circumstances. Travel is not considered a valid excuse for re-scheduling or missing a final exam.

Electronic Etiquette

Students are expected to treat their instructor, guest speakers, and fellow students with respect. It is disruptive to the learning goals of a course or seminar and disrespectful to fellow students and the instructor to use electronics for purposes unrelated to the course during a class session. Turn off all cell phones and other electronic devices during class. Laptops should be used for class-related purposes only. Do not use iPods, MP3 players, or headphones. Do not text, read, or send personal emails, go on Facebook or other social networks, search the internet, or play computer games during class. Some professors will not allow the use of any electronic devices in class. The professor has the right to disallow the student to use a

laptop in future lectures and/or to ask a student to withdraw from the session if s/he does not comply with this policy. Repeat offenders will be directed to the Dean. If you are expecting communication due to an emergency, please speak with the professor before the class begins.

Academic Policies

It is the responsibility of all students to become familiar with and adhere to academic policies as stated in the Academic Calendar. Personal information (information about an individual that may be used to identify that individual) may be required as part of taking this class. Any information collected will only be used and disclosed for the purpose for which the collection was intended. For further information contact the Privacy Compliance Officer at privacy@ambrose.edu.

Extensions

Although extensions to coursework in the semester are at the discretion of the instructor, students may not turn in coursework for evaluation after the last day of the scheduled final examination period unless they have received permission for a course Extension from the Registrar's Office. Requests for course extensions or alternative examination time must be submitted to the Registrar's Office by the deadline date; please consult the List of Important Dates. Course extensions are only granted for serious issues that arise "due to circumstances beyond the student's control."

Appeal of Grade

An appeal for change of grade on any course work must be made to the course instructor within one week of receiving notification of the grade. An appeal for change of final grade must be submitted to the Registrar's Office in writing and providing the basis for appeal within 30 days of receiving notification of the final grade, providing the basis for appeal. A review fee of \$50.00 must accompany the appeal. If the appeal is sustained, the fee will be refunded.

Academic Integrity

We are committed to fostering personal integrity and will not overlook breaches of integrity such as plagiarism and cheating. Academic dishonesty is taken seriously at Ambrose University as it undermines our academic standards and affects the integrity of each member of our learning community. Any attempt to obtain credit for academic work through fraudulent, deceptive, or dishonest means is academic dishonesty. Plagiarism involves presenting someone else's ideas, words, or work as one's own. Plagiarism is fraud and theft, but plagiarism can also occur by accident when a student fails or forgets to acknowledge to another person's ideas or words. Plagiarism and cheating can result in a failing grade for an assignment, for the course, or immediate dismissal from the university. Students are expected to be familiar with the policies in the current Academic Calendar that deal with plagiarism, cheating, and the penalties and procedures for dealing with these matters. All cases of academic dishonesty are reported to the Academic Dean and become part of the student's permanent record.

Note: Students are strongly advised to retain this syllabus for their records.