

<b>Course ID:</b>	<b>Course Title:</b>	<b>Fall 2017</b>
<b>BIO 327</b>	<b>Medical Genetics</b>	<b>Prerequisite:</b>
		<b>Credits: 3</b>

Class Information		Instructor Information		Important Dates	
<b>Days:</b>	Wednesday and Friday	<b>Instructor:</b>	Dr. Chris Wang	<b>First day of classes:</b>	Wed., Sept. 6
<b>Time:</b>	14:30 – 15:45	<b>Email:</b>	chris.wang@ambrose.edu	<b>Last day to add/drop, or change to audit:</b>	Sun, Sept 17
<b>Room:</b>	L 2084	<b>Phone:</b>	(403) 410-2000 ext. 6910	<b>Last day to request revised exam:</b>	Mon, Oct. 23
<b>Lab/ Tutorial:</b>		<b>Office:</b>	L 2113	<b>Last day to withdraw from course:</b>	Mon, Nov 13
		<b>Office Hours:</b>	Wednesday and Friday: 11 am – 12 pm or by appointment (open door policy)	<b>Last day to apply for coursework extension:</b>	Mon, Nov 20
<b>Final Exam:</b>	Dec. 19			<b>Last day of classes:</b>	Mon, Dec 11

### 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

Medical Genetics, L. Jorde, J. Carey and M. Bamshad, 5<sup>th</sup> Edition - 2016, Elsevier.

## Course Schedule

Date	Topic	Reading
Sep. 06	Introduction to the Course	
Sep. 08	Introduction to Human Genome	Ch. 1 and 2
Sep. 13	Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Sep. 15	Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Sep. 20	Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Sep. 22	Principles of Clinical Cytogenetics and Genome Analysis	Ch. 6
Sep. 27	<i>Spiritual Emphasis Day (No Class)</i>	
Sep. 29	Principal of Clinical Cytogenetics and Genome Analysis	Ch. 6
Oct. 04	The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Oct. 06	<b>Mid-term Exam # 1 – In Class</b>	
Oct. 11	The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Oct. 13	The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Oct. 18	The Chromosomal and Genomic Basis of Disease	Ch. 5 and 13
Oct. 20	Human Genetic Diversity: Mutation and Polymorphism	Ch. 3
Oct. 25	Human Genetic Diversity: Mutation and Polymorphism	Ch. 3
Oct. 27	Identifying the Disease-Causing Gene	Ch. 3 and 14
Nov. 01	Identifying the Disease-Causing Gene	Ch. 3 and 14
Nov. 03	Lecture Overflow	
Nov. 08	<i>Fall Module Week (No Class)</i>	
Nov. 10	<i>Fall Module Week (No Class)</i>	
Nov. 15	<b>Mid-term Exam # 2 – In Class</b>	
Nov. 17	The Molecular, Biochemical, and Cellular Basis of Genetic Disease	Ch. 7
Nov. 22	The Molecular, Biochemical, and Cellular Basis of Genetic Disease	Ch. 7
Nov. 24	Cancer Genetics	Ch. 11
Nov. 29	Cancer Genetics	Ch. 11
Dec. 01	In-Class Presentation	
Dec. 06	In-Class Presentation	
Dec. 08	TBD	

**Requirements:****Evaluation:**

<b>Evaluation Methods</b>	<b>Due Date</b>	<b>Weighting</b>
Midterm Exam 1	Oct. 06	25%
Midterm Exam 2	Nov. 15	25%
In-Class Presentation (Individual)	Dec. 01 and 06	10%
Report (Individual)	Dec. 08	10%
Final Exam	Dec. 19	30%

**Grade Summary:**

<b>Percent (%) to Letter Grade Conversion</b>	<b>Grade</b>	<b>Grade Point</b>	<b>Description</b>
90.00% - 100%	A+	4.0	Excellent
85.00% - 89.99%	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.

**Case Selection for In-Class Presentation and Paper:**

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 (Nonsyndromatic Form)  
 Huntington Disease  
 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  
 Prader-Willi Syndrome  
 Retinoblastoma  
 Rett Syndrome  
 Sex Development Disorder (46, XX Male)  
 Sickle Cell Disease  
 Tay-Sachs Disease  
 Thalassemia  
 Thiopurine S-Methyltransferase Deficiency  
 Thrombophilia  
 Tuberous Sclerosis  
 Turner Syndrome  
 Xeroderma Pigmentosum  
 Others  
<https://rarediseases.info.nih.gov/diseases/diseases-by-category/5>

- case selection for in-class presentation and paper is on first-come, first-serve basis
- each case can only be chosen once (*i.e.* no overlapping case between peers, presentations, or papers)

**Contents for In-Class Presentation and Paper:**

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

**In-Class Presentation:**

- 12 minutes presentations followed by 3 minutes questions

**Paper:**

- the paper must be written in your own words and NO QUOTES are allowed (plagiarism is an academic misconduct)
- the paper should be limited to 5-6 double-space pages, excluding figures and a reference list, with 11 to 12 fonts,
- follow the formats of Genetics for both in-text citations and the reference list (<http://www.genetics.org/content/prep-manuscript#references>)
- at least 3, and only, peer-reviewed articles are cited (*i.e.* don't use Wikipedia or web information)
- only submit the print-out copy
- a 10% penalty per day would be applied for late submission

**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

## 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](mailto: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 college. 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.