

## BIOLOGY 262 - HUMAN GENETICS

Spring 2006

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Course Description: Human genetics is an applied genetics course which reviews Mendelian patterns of inheritance, chromosomal syndromes, molecular implications in oncogenetics and immunogenetics, strategies of gene screening, counseling and therapy, and relevant issues of population genetics. The lab will emphasize human genetic methods via in class and field trip experiences. Students will be expected to research a selected topic for a paper and class presentation. This course is an elective course for the biology major.

Texts: HUMAN HEREDITY - Principles and Issues, Seventh Edition (2006)  
by Michael R. Cummings  
MUTANTS by Armand Marie Leroi (2003)

Lecture: Tuesday and Thursday 3b in PPHAC 301

Laboratory: Thursday 12:45 to 3:30 Collier Hall of Science 300

### Tentative Lecture and Laboratory Schedule

Tue Jan 17	Analysis of human genomes Human Genome Project Icelandic Population Study (deCode) Other studies Ethical implications - past and present Meaning (significance) of normal - who is a mutant? Huntington Disease (web site and video)	Chapter 1 (C)       Chapter 1 (M)
	(Draw a student presentation number.)	
Thr Jan 19	Stem Cell Controversy Genetic terminology Chromosome structure Karyotypes Introduction to pedigree analysis-Single gene patterns Questions on cell division - mitosis and meiosis Cystic fibrosis (video, counseling)	Chapter 2 & 3 (C)
<b>LAB1</b>	<b>Bone marrow cultures</b> <b>Meiotic preparations</b> <b>Video: Skeletal syndromes</b> <b>Visit OMIM and Gene Tests Web Sites</b>	Chapter 5 & 6 (M)

Tue Jan 24	Sex linked Patterns X-linked dominant recessive Y linked Maternal inheritance - mitochondrial genes Orphan syndromes - NORDs	Chapter 4 (C)
Thr Jan 26	Complex patterns of inheritance Importance of twins Studies Conjoined twins	Chapter 5 (C) Chapter 2 (M)
<b>LAB 2</b>	<b>Exam 1 Chapters 1-4 (C) and 1, 5,6 (M)</b> <b>Down's syndrome</b> <b>Prenatal testing</b> <b>Amniocentesis</b> <b>Chorionic villi methods</b> <b>Preimplantation genetic diagnosis</b> <b>Set up skin cultures</b>	
Tue Jan 31	Chromosomal syndromes Autosomal syndromes Sex chromosome syndromes Various types of banding methods G, C, T, NOR Chromosome painting methods Additional chromosomal variation	Chapter 6 (C)
Thr Feb 2	Early human development Sexual determination and development Sexual variation - hermaphrodites and pseudohermaphrodites Gender pursuit	Chapter 7 (C) Chapter 3 & 4 (M) Chapter 7 (M)
<b>LAB 3</b>	<b>Field trip: Local Hospital</b> <b>Amniocentesis</b> <b>Chorionic villi method</b>	
Tue Feb 7	Molecular structure of the genome Genomics Sequencing technology Hybridization technology	Chapter 8 (C)
Thr Feb 9	Gene expression Proteomics	Chapter 9 (C)
<b>LAB 4</b>	<b>Exam 2 Chapters 5-8 (C) 3-4 (M)</b> <b>Harvest skin cultures</b>	

	<b>X-inactivation-sex chromatin test</b> <b>Video: Genetic counseling for cystic fibrosis</b>	
<b>Monday</b>	<b>Set up student blood samples</b>	
Tue Feb 14	Genetic implications in metabolic pathways Chapter 10 (C) Genetic screening newborns - PKU High risk populations Jewish population - Tay sachs African American population - sickle cell anemia Caucasion population - cystic fibrosis	
Thr Feb 16	Molecular gene mutations DNA repair Genomic imprinting Syndromes involving skin and hair	Chapter 11 (C)   Chapter 8 (M)
<b>LAB 5</b>	<b>Harvest student blood sample</b> <b>Stain and study slides</b> <b>Video: PKU screening technologies</b>	
Tue Feb 21	Oncogenetics Oncogenes <i>myc</i> and <i>ras</i> Tumor suppressor genes retinoblastoma gene BRCA 1 and 2 Cancer models Colon cancer Melanoma Chromosomal changes associated with specific cancers Philadelphia chromosome Burketts lymphoma	Chapter 12 (C)
Thr Feb 23	Recombinant DNA technologies Transgenic organisms Molecular clones PCR Southern blotting	Chapter 13 (C)
<b>LAB 6</b>	<b>Exam 3 - Chapters 9-12 (C); Chapter 8 (M)</b> <b>Electrophoresis lab</b> Agarose electrophoretic separation Southern blotting <b>Continue to study and photograph chromosome slide</b>	
Tue Feb 28	Identifying disease related genes microarrays high throughput screening <i>in situ</i> hybridization Cancer screening - new technologies	Chapter 14 (C) (324-335)

Thr Mar 2 <b>LAB 7</b>	Human genome project <b>Field trip to Molecular Toxicology Lab</b> <b>High Throughput screening</b> <b>Culture Media</b>	Chapter 15 (C)
Tue Mar 14	Reproductive technologies	Chapter 16 (C)
Thr Mar 16	Immunogenetics Immunoglobulin genes T-cell receptor genes	Chapter 17 (C)
<b>LAB 8</b>	<b>Exam 4 Chapters 13-16 (C)</b> <b>Dermatoglyphics</b> <b>Video: Genetic counseling for cardiovascular disease</b>	
Tue Mar 21	Behavior genetics Schizophrenia Depression	Chapter 18 (C)
Thr Mar 23	Population genetics/Evolution	Chapter 19 (C)
<b>LAB 9</b>	<b>Population genetics lab</b> <b>Video: Williams syndrome</b> <b>Video: Tourette's syndrome</b>	
Tue Mar 28	Genetics of Racial groups Eugenics (past and present) Aging	Chapter 9 (M)
Thr Mar 30	Genetically modified organisms applications to human genetics	Chapter 14: (C) (314-324)
<b>Lab 10</b>	<b>Guest speaker on cancer screening</b> <b>Video: Genetic counseling for breast cancer</b>	
Tue Apr 4	<b>Exam 5 Chapters 17-19 and Chapter 14 pgs; Chapter 9(M) (take home exam)</b>	
Thr Apr 6	<b>Field trip to Philadelphia</b> <b>morning - Mutter Museum</b> <b>afternoon - Fox Chase</b>	
<b>Lab 11</b>		Tue Apr 11
Tue Apr 11	Student presentations	(1 & 2)
Thr Apr 13	Student presentations	(3 & 4)
<b>Lab 12</b>	Student presentations <b>Dermatoglyphics</b>	(5 & 6)

Tue Apr 18	Student presentations (7 & 8)	
Thr Apr 20	Trip to New York area	
<b>Lab 13</b>	<b>Quest diagnostics -cytogenetics lab Mt. Sinai Hospital - Jewish genetics</b>	
Tue Apr 25	Gene therapy Gene screening Genetic counseling	Chapter 10 (M)
Thr Apr 27	Forensic genetics	
<b>Lab 14</b>	<b>DNA Fingerprinting</b>	

**FINAL EXAM:** Essay exam based on guideline focus questions and Chapters 2 and 10 (M)

**Components of your grade:**

Paper and presentation - 200 pts

Final exam - 200 pts

Unit exams - 375 pts (75 pts each)

Lab grade - 280 pts

Attendance/participation grade - 100 pts

**Final grade = earned points/possible points = %**

90 - 100% = A

80- 89% = B

70- 79% = C

60 - 69% = D

below = F

