**GEORGE MASON UNIVERSITY**

**College of Science**

**BIOL666** (3 credits)

**Human Genetics Concepts for Health Care**

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**Prince William: SciTech (PW): Colgan Hall 203;**

**MONDAYS 4:30 pm**

**Course Pre-requisites:** Students in BIOS/BIOL program will be able to count either this course, either BIOL572 Human Genetics, but not both of them.

**Course Title:** Human Genetic Concepts for Health Care

**Catalogue Description:** Principles of genetically-determined diseases with emphasis on clinical aspects of these diseases, genetic counseling, and laboratory methods used in human genetics. Extended Studies students preparing to enter Med- or Dental Schools are welcome.

***Pre-requisite:*** B.S. degree completed or enrollment in accelerated Masters program. At least one Cell or Molecular Biology undergraduate course. ***Not available to students who have taken BIOL 572.***

**Course Objectives:**

The course will integrate knowledge of genetic principles and framework of genetically-determined human diseases with a special emphasis on the pathophysiological aspects of monogenic and multifactorial diseases, and on genetic counseling. Students will learn to interpret the results obtained using research methods commonly used by human geneticists.

***Note:*** If you are a student with a disability and you need academic accommodations, please see instructor, and contact the Disability Resource Center (DRC) at 708-993-2474. All academic accommodations must be arranged through the DRC.

**Recommended Texts:**

Nussbaum, R.L., McInnes, R.R.& Willard, H.F. (2004). *Thompson & Thompson: Genetics in Medicine*, 6th ed. Philadelphia: Saunders.

Young, I.D. (2005). *Medical Genetics*. New York: Oxford University Press.

**Or any other, more recent editions of these two books**

**OTHER REQUIRED READING: (before 2nd exam)**

1. Seleman M, Hoyos-Bachiloglu R, Geha RS, Chou J. Uses of Next-Generation Sequencing Technologies for the Diagnosis of Primary Immunodeficiencies. Front Immunol. 2017 Jul 24;8:847. doi: 10.3389/fimmu.2017.00847. eCollection 2017. Review. PubMed PMID: 28791010; PubMed Central PMCID: PMC5522848.
2. Ramos E, Weissman SM. The dawn of consumer-directed testing. Am J Med Genet C Semin Med Genet. 2018 Mar;178(1):89-97. doi: 10.1002/ajmg.c.31603. Epub 2018 Mar 7. PubMed PMID: 29512889
3. Sawyer SL, Hartley T, Dyment DA, Beaulieu CL, Schwartzentruber J, Smith A,.. FORGE Canada Consortium; Care4Rare Canada Consortium, Majewski J, Boycott KM. Utility of whole-exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clin Genet.2016 Mar;89(3):275-84. doi: 10.1111/cge.12654. Epub 2015 Sep 22. Review. PubMed PMID: 26283276; PubMed Central PMCID: PMC5053223.

**Course Grading :**

**Evaluation will be based upon:**

Closed Book Exams 100% (25% Mid-term, 25% Mid-term, 50% Final exam)

The website for the GMU honor code: http://www.gmu.edu/facstaff/handbook/aD.html

**Course Schedule**

| **Week** | Activity |
| --- | --- |
| **WEEK 1, January 28th** | **Lecture 1. Acute and chronic diseases: blurry borders. EBV infection as an example. Genetic classification of human diseases. Family studies, twin studies, adoption studies. Polygenic inheritance. Heritability. Genes and Environment. “Liability-Threshold model”. Susceptibility genes. Common disease-common variant (CDCV) hypothesis. Linkage and association.**  |
| **February 4th**  | **Lecture 2. Examples of multifactorial diseases: detailed analysis. 1. Alzheimer disease 2. Coronary Artery disease 3. Diabetes Type I (childhood onset) 4. Diabetes type II (adult onset) 5.Hirschprung disease 6. Neural tube defects 7. Schizophrenia**  |
| **February 11th** | **Lecture 3. Mutations and their consequences to expression of the gene or function of its product. Functional types of mutations, loss and gain of function. Spontaneous and induced mutations. Mutagens. Instability of the human genome. Paternal gain of mutations. Common types of mutations in DNA. Rate of mutations in humans. Mutations vs. Aneuploidy. Truncation selection in human populations. Forced gain of mutations. HPRT Assay. Comet assay. Environmental carcinogens.DNA repair and DNA checkpoints. Ataxia-Telangioectasia.Direct-to-Consumer (DTC) genetic testing.** |
| **February 18th** | **Lecture 3 continued.** |
| **February 25th** | **Lecture 4.** **Genetics of human development. Human reproduction as an inefficient process. Critical periods for developmental defects. Classification of birth defects. Teratogenesis. Hox and Pax genes. FGF pathway. Sonic hedgehog pathway. Neural crest cell migration network. Sex determination pathway.**  |
| **March 4th**  | **EXAM1**  |
| **March 18th** | **Lecture 5. Chromosome instability syndromes. Telomeric instability.** |
| **March 25th** | **Lecture 5 continued. Chromosome instability syndromes. Telomeric instability.** **Lecture 6. Mendelian diseases. Important definitions of classic genetics. Expressivity. Penetrance. Consanguinity. X-chromosome inactivation. Anticipation. Mosaicism. Mitochondrial inheritance.**  |
| **April 1st** | **Lecture 7. NextGen sequencing to find out gene defects. Exome sequencing for rare diseases. Recent advances. READ THE FRONTIERS MS before exam****Lecture 8. Human Polymorphisms. Types of Variations in Human Genome. Types of SNPs. Life cycles of SNPs and mutations. Disease risk and treatment response associated types of variation. ApoE4.TP53 Arg/Pro.** |
| **April 8th**  | **Exam 2. Lecture 8 CONTINUED. Human Polymorphisms. Isoniazide, methotrexate, tamoximen response and its genetic modifiers. INDEL polymorphisms. ACE Alu-based indel. CCR5 indel. Copy Number Variations. CCL3L1. CNVs and autism. Allelic associations. Ankylosing spondylitis and B27 allele. Genome-wide association studies. HapMap project. Biomedical differences between chimps and human.** |
| **April 15th** | **Lecture 9. Obesity as an example of complex disease. (backed up as VIDEO RECORDED LECTURE).** |
| **April 22st** | **Lecture 11. Tumor genetics (backed up as VIDEO RECORDED LECTURE)** |
| **April 29th**  | **Lecture 10. REACTIVE OXIGEN SPECIES AS CONTRIBUTORS TO HUMAN DISEASES/**  |
| **May 6th** | **Lecture 12. Epigenetics contributions to human disorders** |
| **May 13th** | **FINAL EXAM MAY 13th** |