UNIVERSITY OF THE PHILIPPINES
College of Medicine – Philippine General Hospital
University of the Philippines Manila

Genetics 201 – Principles of Human Genetics
1st Semester, AY 2024-2025

COURSE GUIDE

COURSE DESCRIPTION

This is the Principles of Human Genetics. It is one of the core courses for the Master of Science in Genetic Counseling and it is meant to provide a foundation on the basics of human genetics. This course runs for one semester. As with other courses, you are expected to read the prescribed text-books and attend the lectures which will supplement your learning.

COURSE LEARNING OUTCOMES

After completing this course, you should be able to achieve the following:

1. Comprehend the basics of human genetics
2. Analyze common genetic disorders and appreciate their etiologies, inheritance patterns and recurrence risk, appropriate diagnostic examinations
3. Appreciate the fundamentals of cytogenetics and molecular genetics
4. Appreciate the application of basic human genetics in the fields of dysmorphology, inborn errors of metabolism, cancer genetics and epigenetics

COURSE OUTLINE / Schedule

Date	Topic	Readings
Aug 26	Genes and how does it work? Introduction to chromosomal basis of heredity, genetic variations and patterns of inheritance	**Genetics in Medicine** Chapter 2: Introduction to the Human Genome Chapter 3: The Human Genome Gene Structure and Function Chapter 7: Patterns of Single-Gene Inheritance **New Clinical Genetics** Chapter 1: What can we learn from a family history? Chapter 3: How do genes work?
Sept 2	Clinical Cytogenetics Mendelian disorders with cytogenetic effects, cytogenetic analysis in cancer, autosomal and sex chromosome disorders, tools in cytogenetics	**Genetics in Medicine** Chapter 5: Principles of Clinical Cytogenetics and Genome Analysis Chapter 6: The Chromosomal and Genomic Basis of Disease: Disorders of Autosomes and Sex Chromosomes **New Clinical Genetics** Chapter 2: How can a patient's chromosome be studied?

Sept 9	How to analyze DNA: Analysis of DNA and RNA sequences, methods of nucleic acid and protein analysis (PCR, Southern blotting, Northern blotting, Western blotting, etc.)	**Genetics in Medicine** Chapter 10: Identifying the Genetic Basis for Human Disease Chapter 11: The Molecular Basis of Genetic Disease **New Clinical Genetics** Chapter 4: How can a patient's DNA be studied? Chapter 5: How can we check a patient's DNA for gene mutations?
Sept 16	What is the impact of having a mutation? Why does it matter?	**Genetics in Medicine** Chapter 4: Human Genetic Diversity: Mutation and Polymorphism Chapter 7: Patterns of Single-Gene Inheritance **New Clinical Genetics** Chapter 6: What do mutations do?
Sept 23	Inborn Errors of Metabolism	**Genetics in Medicine** Chapter 12: The Molecular, Biochemical and Cellular Basis of Genetic Disease **New Clinical Genetics** Chapter 9: Why are Some Conditions Common and Others Rare
Sept 30	**MIDTERM EXAM**	
Oct 7	Cancer Genetics Molecular basis of cancer (e.g., oncogenes and tumor suppressor genes), the two-hit hypothesis, examples of cancer syndromes	**Genetics in Medicine** Chapter 15: Cancer Genetics and Genomics **New Clinical Genetics** Chapter 7: Is cancer genetic?
Oct 14	Genetics of Skeletal Dysplasia	**Genetics in Medicine** Chapter 12: The Molecular, Biochemical and Cellular Basis of Genetic Disease
Oct 21	Molecular basis of Neurogenetic conditions	**Genetics in Medicine** Chapter 12: The Molecular, Biochemical and Cellular Basis of Genetic Disease
Nov 4	Genetics of the Hemoglobinopathies	**Genetics in Medicine** Chapter 11: The Molecular Basis of Genetic Disease
Nov 11	Genetic Aspects of Human Development Gene expression during in-utero development, examples of genetic conditions impacted by disorders of development (e.g., dysmorphology, etc.), Prenatal diagnostics (e.g., ultrasound)	**Genetics in Medicine** Chapter 14: Developmental Genetics and Birth Defects Chapter 17: Prenatal Diagnosis and Screening **New Clinical Genetics** Chapter 12: When is Screening Useful
Nov 18	Introduction to Epigenetics, Gene Therapy and Pharmacogenetics	**Genetics in Medicine** Chapter 18: Application of Genomics to Medicine and Personalized Health Care

		New Clinical Genetics Chapter 10: How do our genes affect our metabolism, drug responses and immune system? Chapter 11: How are genes regulated?
Nov 25	**FINAL EXAMS**	
Dec 2	**Video Presentations and Exam Feedback**	

MODE OF DELIVERY

All mode of instruction will be delivered through a learning management system. Supplemental lecture videos and readings will be found in VLE. Please take note that students are expected to have their own copies of the textbooks.

Please contact the Information Management System at ims@post.upm.edu.ph if you do not have an existing UP Manila email address or if you have any issues with logging in.

COURSE MATERIALS

The primary textbook that will be used are:

Genetics in Medicine, 8th ed. by Nussbaum, McInnes and Willard
New Clinical Genetics, 4th ed. by Read and Donnai

COURSE REQUIREMENTS

Course Requirement 1 – Quizzes (20%)

After each topic, there is a 7-10 point quiz that the students should answer. Take note that the quiz should only be answered AFTER the student has read the required readings and watched the supplemental video lectures. The student will be given 15 minutes to answer the quiz and s/he can only attempt this once. All quizzes will account for 20% of the student's final grade. Take note of the availability of the quizzes.

Course Requirement 2 – Video (20%)

Each student will produce a video not longer than 10 minutes which will be due on the last day of classes. The video should contain the following information: background of the disorder, etiology of the disorder and appropriate diagnostic test, inheritance pattern, relevant principles of genetics if applicable (ie., genetic anticipation, X-linked inactivation, penetrance, expressivity, etc.) and implications for genetic counseling. This will comprise 20% of the student's final grade. The following is the basis for grading:

Background of the disorder	15
Etiology of the disorder and appropriate diagnostic test	10
Inheritance Pattern	10
Relevant Principles of Genetics	30
Implications for Genetic Counseling	10
Organization, Communication Skills, Adherence to Time Limit, Creativity	20

References	5
TOTAL	100

A list of the suggested topics can be seen below:

1. Neurofibromatosis Type 1
2. Hunter Syndrome
3. Duchenne Muscular Dystrophy
4. MELAS
5. Fragile X Syndrome
6. Rett Syndrome
7. Osteogenesis Imperfecta
8. Turner Syndrome
9. Prader Willi Syndrome
10. Angelman Syndrome
11. William Syndrome
12. Maple Syrup Urine Disease
13. Marfan Syndrome
14. Alpha Thalassemia
15. Edward Syndrome

Course Requirement 3 – Written Exams (60%)
The students will have two 100-point examinations (midterms and final exams) covering the topics listed above. This will comprise 60% of their final grade.

ABOUT THE INSTRUCTOR

I am Dr Mary Ann Abacan and I am the current course coordinator of Genetics 201. The rest of the faculty members are: Dr Carmencita Padilla, Dr Eva Cutiongco-de la Paz, Dr Catherine Lynn Silao, Dr Melanie Alcausin and Dr Mary Anne Chiong. We are assisted by Dr Ma-am Joy Tumulak. I may be contacted through mrabacan@up.edu.ph.