

Pathology 4600B: Genetic and Epigenetic Basis of Human Disease

Short title: Genetic Basis of Human Disease

This course encompasses the origins of genetic and epigenetics changes found in patients, manifestations of these in disease, and treatment approaches. Most disease scenarios will focus on inheritance of developmental and degenerative diseases. Mouse models will highlight causative changes as a complement to the analysis of human clinical examples.

Prerequisite(s): Biology 2581A/B; Pathology 3500

Extra Information: 2 lecture hours

Course Weight: 0.50

Learning Outcomes:

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

- describe molecular origins of genetic and epigenetic changes associated with diseases, such as cancer, and developmental anomalies in humans.
- describe and compare modern molecular genetic diagnostic methods.
- explain how DNA sequence aberrations resulted in selected examples of human disease conditions.
- explain how epigenetic alterations resulted in selected examples of human disease conditions.
- distinguish different clinical trials and therapeutic repurposing in the management of select examples of diseases caused by DNA sequence aberrations and epigenetic alterations.

Draft course outline

Course Coordinator: Dr. Fred Dick.

Others will include clinical geneticists and faculty members with specialized knowledge related to the lecture themes.

Assessments

Assessment	Format (written, oral, mixed)	Weight (%; must total 100)
Online assignments (6)	Written, submitted through OWL	30
Online quiz	Written, submitted through OWL	10
Mid-term test	Written, in person	25
Final exam	Written, in person	35

Draft Course Schedule

Week #	Topic	Instructor
Week 1	Introduction - Interphase nuclear structure and mitosis	Dr. Dick
Week 2	Introduction – DNA replication, DNA damage, DNA repair mechanisms, and error types	
Week 3	Introduction - Epigenetic silencing and retrotransposons	
Week 4	Diagnostic tools – DNA sequence based	
Week 5	Diagnostic tools - Cytogenetics	
Week 6	Diseases – Intellectual disabilities, Cohesinopathies, Condensinopathies, and Chromothripsis	
Week 7	Diseases – Diabetes and circulatory diseases, epigenetics and long non-coding RNAs	
Week 8	Diseases – Rare cancers and epigenetic misregulation	
Week 9	Applications - Clinical trials basics and strategies	
Week 10	Applications - Synthetic lethality and therapeutics	
Week 11	Applications - Viral mimicry and therapeutics	
Week 12	Applications - Model organisms and drug repurposing	