GENE 514: Current Concepts in Clinical Molecular Genetics and Molecular Diagnostics

The objective of this course is to provide a review of molecular diagnosis of common hereditary or neoplastic disorders for which DNA-based diagnosis is now in routine use. Topics include FGFR3 disorders, fetal blood typing, thrombophilias, hemochromatosis, fragile X syndrome, polyglutamine disorders, hereditary breast cancers, Charcot Marie Tooth and spinal muscular atrophy, PraderWilli and Angelman syndromes, mitochondrial diseases, Duchenne and Becker muscular dystrophy, cystic fibrosis, and Smith-Lemli-Opitz Syndrome. Sessions also include genetic risk prediction, using linkage and Bayesian analysis as well as DNA forensics and paternity testing. The course is designed as part of the required curriculum for Clinical Genetics residents and Fellows preparing for the Clinical Molecular Genetics Boards given by the American Board of Medical Genetics.

Learning Objectives

- Appreciate the types of techniques used in molecular genetic diagnostic laboratories, including the limitations of each assay
- Acquire skills in calculating residual risks after molecular testing

Credits: 1
Class Type: Graduate Course
Prerequisites:
GENE 500 and permission of the instructor.
Program: Biology, Genetics, and Medicine