

Medical Genetics Spring 2004

Course Director:
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Lectures and exam in Bakst Auditorium

Required text:

Genetics in Medicine, Nussbaum, McInnes, and Willard, 6th edition revised reprint, 2004.

Date	Time	Class	Instructor
May 19	12:30- 1:30	Gene and Genome Organization Gene structure, chromatin organization, gene expression and imprinting, genotypes and phenotypes, inheritance of chromosomes, and basic Mendelian principles	Dr. Shoumita Dasgupta
May 20	11:30- 12:30	Chromosomes and Chromosomal Abnormalities Mitotic and meiotic nondisjunction, abnormalities of chromosome number and structure, uniparental disomy, mosaicism, FISH and karyotyping	Dr. Beth Sullivan
May 24	10:30- 11:30	Molecular Genetic Abnormalities Mutation and disease, monogenic syndromes, trinucleotide repeat expansion diseases, and mitochondrial disorders	Dr. Shoumita Dasgupta
May 26	1:30- 3:30	Transmission of Genetic Abnormalities Pedigree Workshop	Dr. Shoumita Dasgupta
May 27	11:30- 12:30	Gene Regulation and Development Transcription factors, role of environment and teratogens in development, and congenital malformations	Dr. Kenn Albrecht
May 28	10:30- 11:30	Identification of Genetic Basis of Disease Linkage and gene mapping, association studies, twin studies, pleiotropy, multifactorial disease, modifier genes, and penetrance	Dr. Shoumita Dasgupta
June 2	1:30- 3:30	Patient panel: Trisomies 18 and 13	Dr. Shoumita Dasgupta

June 3	11:30- 12:30	Cancer Genetics and Animal Models of Disease Tumor suppressor genes and oncogenes, loss of heterozygosity, gene activation by duplication or translocation, DNA repair and genome instability, and knock out mouse models	Dr. Michael Christman
June 4	10:30- 11:30	Testing for disease susceptibility Molecular methods and limitations of linked marker and disease locus tests	Dr. Shoumita Dasgupta
June 7	10:30- 11:30	Genetic Frontiers The Human Genome Project, microarrays, pharmacogenomics, RNAi and gene therapy	Dr. Shoumita Dasgupta
June 9	8:30- 9:30	Genetic Screening and Prenatal Diagnosis Types and methods of screening: population screening, newborn screening, screening for heterozygotes, and screening in pregnancy	Dr. Jeff Milunsky
June 9	1:30- 3:30	Small Group: Case studies	Dr. Shoumita Dasgupta
June 10	11:30- 12:30	Genetic Counseling: Principles and Practice Knowing when to seek the help of a genetic counselor, principles of communication: accuracy of tests, non-directive counseling, confidentiality	Ms. Nathalie McIntosh, M.Sc., CGC, CCGC
June 11	9:30- 10:30	Ethics and the New Genetics Eugenics, information privacy, health insurance, racial discrimination	Dr. Shoumita Dasgupta
June 16	9:00- 10:30	Final Exam	