

HG 542: Molecular Basis of Human Genetic Disease (3credit hours)

Winter Term 2009

Location: 5915 Buhl

Time: 10-11 AM

Course Director: Glover

Instructors: Antonellis, Camper, Mustapha-Chaib, Burmeister, Li, Scott, Gruber, Moran, Glover, Sekiguchi

HG 542 will emphasize the principles and methods of genetics and molecular genetics as they relate to human disease. The course covers the topics of Mendelian inheritance, monogenic traits, complex genetic disorders, non-Mendelian inheritance, cytogenetics and copy number variants, and cancer genetics. In each section, principles of genetics are presented by way of illustration with human genetic diseases. Papers from the current and classic literature will supplement lecture material. HG 541 is a prerequisite for this course.

Note: An expanded syllabus with reading assignments is available on ctools

Date/ Faculty/Lecture Title

1/7	W	Antonellis	Mapping human disease genes I: inheritance patterns and linkage analysis
1/9	F	Antonellis	Mapping human disease genes II: identifying disease-associated variants
1/12	M	Antonellis	Functional characterization of human mutations
1/14	W	Antonellis	Molecular pathology I: introduction and loss of function mutation
1/16	F	Antonellis	Molecular pathology II: gain of function and dominant negative mutation
1/19	M	<i>No Class - MLK Day</i>	
1/21	W	Antonellis	Monogenic diseases: windows into more complex issues
1/23	F	Recitation	
1/26	M	Camper	X-linked inheritance: Color blindness
1/28	W	Camper	
1/30	F	Mustapha-Chaib	Deafness

2/2	M	Mustapha-Chaib	
2/4	W	Recitation	
2/6	F	EXAM I	
2/9	M	Burmeister	Introduction to complex genetics
2/11	W	Burmeister	Heterogeneity in rare disorders: founder effects, homozygosity, etc.
2/13	F	Burmeister	Gene x environment interactions: Behavior (Neurogenetics)
2/ 16	M	Burmeister	Pharmacogenetics
2/18	W	Burmeister	Personalized medicine and recreational genomics
2/20	F	Recitation	
2/23-2/27		No Classes - Spring break	
3/2	M	Scott	Primary analysis and meta-analysis of genome-wide association (GWAS) studies for complex diseases
3/4	W	Li	Following-up GWAS findings: validation, bioinformatics, rare variants, etc.
3/6	F	Gruber	Genetic Epidemiology of Cancer
3/9	M	Li	Homozygosity in the human genome
3/11	W	Moran	Sex determination
3/13	F	Moran	Prion disease
3/16	M	Recitation	
3/18	W	EXAM II	
3/20	F	Glover	Fragile X syndrome and unstable repeat disorders

3/23	M	Glover	Aneuploidy
3/25	W	Glover	Structural rearrangements: implications of genomic architecture
3/27	F	Glover	Structural variation in the human genome
3/24	M	Glover	Copy number variation in genetic disease and cancer
4/1	W	Recitation	
4/3	F	Sekiguchi	Cancer genetics I
4/6	M	Sekiguchi	Cancer genetics II
4/8	W	Sekiguchi	Inherited genome instability disorders
4/10	F	Sekiguchi	Chromosomal abnormalities in cancer
4/13	M	Sekiguchi	Mechanisms of chromosomal anomalies in cancer
4/15	W	Recitation	
4/17	F	EXAM III	