BIO 426/526 Human Genetic Diseases, Spring 2018

The aim of this class is to help students gain a sufficient understanding of human genetics so that they can appreciate the genetic basis of human diseases, and the importance of genetic considerations in the prevention and treatment of diseases. This course will use a literature-based approach to cover the fundamentals, from the chromosomal basis for hereditary to the molecular and biochemical basis of genetic diseases as well as the current molecular diagnostic and treatment methods available for genetic diseases. A combination of classic and current papers will be used. In Bio 526, graduate students will gain better insight into the current literature, by analyzing and presenting recent research papers in great detail.

Text: None. Based on preliminary literature.

Credit hours: 3 units

Prerequisite: Genetics (319) or permission from instructor

Time: Fridays 10.30 PM - 1:10 PM
Location: Cooke Rm 248

Instructor: Dr Shermali Gunawardena
Office hours: Monday 10-11am, Cooke Rm 329.

Grading: Final grades will be determined from in class quizzes (20%), class presentation (25%), class participation/discussion (28%) and a written report (28%). No extra credit will be given. Missed classes cannot be made up. Coming late to class will be considered as an absence from class. Undergraduate students will each present and discuss a topic as detailed in the syllabus. Graduate students will be responsible to present and discuss a primary research paper on a topic as detailed in the syllabus. Presentations will be graded on delivery, content and the stimulation of class discussion, and is 24% of the grade. Each student will also be graded on class participation/discussion during each class, which is 28% of the grade. All students will write a written report on the topic they present, which is due the week AFTER the presentation (28% of the grade).

Grading scale: ≥90% = A, 80–89% = B, 70–79% = C, 60–69% = D, <60% = F
A curve may also be applied and plusses and minuses assigned at the instructors' discretion.

Quizzes=	200 (40x5)
Class presentation=	240
Class participation=	280 (20x14)
Written Report=	280

Makeup policy: Make-up presentations will only be given for medical reasons and will be evaluated on a case by case. You must submit a signed doctor's note to be considered for a make-up. Attendance in every class is a MUST unless for medical reasons with a doctor's note. No make-ups will be given for in class quizzes. A missed class will result in a zero grade for discussion/participation for that day.
Other policies: Since this is a discussion class, **late attendance** is highly discouraged. Further it is a distraction to the presenter. Any late attendance will result in the deduction of participation points.

SYLLABUS SUMMARY

Cytogenetics, chromosomes, inheritance patterns.
Tools of cytogenetics and molecular genetics and patterns of inheritance
Prenatal diagnosis, genetic counseling/introduce pedigees and risk assessment and ethics

Mendelian inheritance
Autosomes: Autosomal recessive: Tay sacs disease, cystic fibrosis
Autosomal dominance: neurofibromatosis, familial hypercholesterolemia
Incomplete dominance: Achondroplasia/dwarfism
Sex chromosomes: x-linked recessive: inherited hemophilia, x-linked dominate disorders: Rett syndrome, X inactivation, dosage compensation: Duchenne Muscular Dystrophy

Chromosome non disjunction
Review meiosis, mitosis, mechanisms of nondisjunction.
Disorders of autosomes, Trisomy 21/18/13
Sex chromosome abnormalities: X-linked mental retardation, Klinefelter syndrome, turner syndrome. Trisomy X, XYY syndrome

Genomic disorders: Deletion/duplication syndromes
Mechanisms of genomic duplication/deletion. Cri du chat syndrome, Smith-Magnenis syndrome
Microdeletion syndromes: DiGeorge syndrome, Velocardiofacial syndrome or conotruncal anomaly face syndrome.
Duplication syndromes: Charcot-Marie Tooth disease

Genomic imprinting
Mechanisms of genomic imprinting
Prader-Willie and angelman syndrome
Uniparental disomy of imprinted region Beckwith-Weidemann syndrome

Multi factorial traits with genetic and environmental factors are known.
Introduction to genetic and environmental factors, mechanisms of multi factorial traits.
Retinitis pigmentosa, venous thrombosis, Hirschsprung disease, type 1 diabetes
Neuronal tube defects cleft palette

Mitochondrial disorders
Mechanisms of mitochondrial disorders, Leber hereditary optic neuropathy

Mosaics and chimeras
Mechanisms of somatic mosaics. Discussion of human chimera case studies.

Detecting genetics abnormalities
Benefits and problems

Treatment of genetic diseases

Pharmacogenetics, gene therapy, stem cell therapy

ASSESSMENT

0=not covered, 1=moderately covered, 2=extensively covered
The course learning objectives will be met when 90% of the enrolled students achieve a final course grade of C or higher.

Number	Program Learning Outcome	Depth	Specific outcome objectives for Bio 426/526	Assessment instrument
1	Provide breadth of knowledge of basic principles and concepts	1	Apply basic genetic concepts of both mendelian and non-mendalian genetics to human genetic diseases	Oral presentation and Written report
2	Provide depth within specialized areas	2	Understand how mutations in genes function to cause human genetic diseases.	Presentation, report and Quizzes (discussion of primary literature -526)
			Understand how molecular pathways are affected by mutations in genes during disease.	Presentation, report and Quizzes (discussion of primary literature -526)
			Understand the current diagnostic, preventive care or treatment strategies available for genetic disease.	Presentation, report (discussion of primary literature discussion-526)
			Understand the ethical and social issues of human genetic diseases and the counseling and social services that are currently available.	Presentation, report and discussions. (discussion of primary literature -526)
			Understand current research activities on gene and stem cell therapies for human genetic diseases.	Presentation, report and discussions. (discussion of primary literature -526)
3	Provide an understanding of experimental design and methodology	1	Know critical experiments that were used to expose the mechanisms involved in human genetic disease.	Presentation, report and Quizzes (discussion of primary literature -526)
4	Develop approaches for integration of information	1	Understand the contribution of genetics, cell and molecular biology and pharmacology to human genetic disease.	Presentation, report and Quizzes (discussion of primary literature -526)

			Understand the molecular and genetic information of human genetic diseases	Presentation, report and Quizzes (discussion of primary literature -526)
5	Encourage critical thinking and hypothesis building	1	Initiate discussions on novel approaches for genetic diseases	Discussions (discussion of primary literature -526)
6	Provide skills in scientific communication	2	Develop skills to communicate both orally and in writing the current findings for a particular human genetic disease.	Presentation and report (discussion of primary literature -526)
7	Provide contemporary information	2	Understand key recent findings in genetic and molecular basis for human genetic disease	Presentation, report and Quizzes (discussion of primary literature -526)
8	Encourage appreciation of scientific values	1	Understand the research contributions in elucidating the mechanisms of human genetic disease.	Presentations, report and discussions.
			Appreciate the ethical and social issues involved in diagnosis and treatment of these diseases.	Presentation, report and discussions.