First Course Sheet

BSE654/BSE654A: Human Molecular Genetics [3-0-0-0 [9]] | 2017-18-II

Instructor: S.Ganesh (sganesh@) | Teaching Assistant: Akanksh Onkar (onkara@)

Class Schedule: 11:00 to 11:50 am (W/Th/F) | Venue: BSBE Seminar Room

Contact Hour: Friday, 5-6 pm (on advance request via email)

Course website: http://home.iitk.ac.in/~sganesh/hmg/

Objective: This course is expected introduce the rapid advancements in our understanding the role of human genome in health and disease. We would introduce key concepts of inheritance of human traits, pedigree analysis, and chromosome organization. Molecular biology tools used for understanding the genome, gene structure and gene mutations, gene mapping and gene cloning strategies will also be covered. Objectives and outcome of human genome project and the HapMap project will also be discussed at the end

Target students: It is an elective course for the UG and PG students of BSBE department. This course is not open to students from other departments since the students are expected to have sufficient introduction in cell and molecular genetics.

Attendance: 100% attendance unless leave approved by the DPGC (please inform the TA on approved leave duration). Students who are not regular to the class will be asked to deregister within the deadline announced by the DOAA.

Course materials: This course covers a number of research papers - close to 40 of them - throughout the semester and pdf of the papers are made available through the course website (http://home.iitk.ac.in/~sganesh/hmg/). While there is no prescribed text book for this course, students are advised to read the book, *Human Molecular Genetics* (Authors: *Tom Strachan and Andrew P. Read*), available at the central library, to learn the basics.

Course content: Simple Mendelian traits; Loss-of-function mutations; Gain-of-function mutations; Gene interactions; Dynamic mutations; Genetics of neoplasia; Genomic imprinting and human disease; X inactivation and DNA methylation; Gene mapping and positional cloning; Mutifactorial inheritance; Genetics of behavioral disorders; Pharmacogenetics and biochemical genetics; Animal models in human genetics: Methods used for diagnosis and detection of gene mutations; Gene Therapy.

Exams/Quiz: Two exams (mid and end); 40% weightage for each. Two pre-announced quizzes (open book) - one before and one after the mid-term exam; 10% weightage for each; No make-up for the quiz will be conducted. Two assignments and two presentations as group activities to encourage critical reading and discussion but they do not carry any weightage.

Grading policy: Student should secure minimum 40 % to get a pass grade.

S.Ganesh

Course instructor

Syllabus:

3 Lecture per week – Total 42 lectures

Course Content

Sl. No	Торіс	Lectures
1	Introduction to Human Molecular genetics, including	4
	concepts on genetic and physical mapping	
	Gene mapping in Mendelian disorders: Autosomal	2
	recessive disorder	
	Gene mapping in Mendelian disorders: Autosomal	2
	dominant disorder	
	Sex determination and Y-liked genes in humans	2
	Monogenic disorders: haploinsufficiency and long-range	2
	position effect	
	Dynamic mutations: neurological disorders associated	2
	with unstable repeats in the genes	
	Animal models for human disorders: Transgenic,	2
	knockout and knock-in mice models	
	Genomic imprinting and imprinting related disorders	2
	Genome rearrangements and genomic disorders.	2
	Beyond Mendel: Oligogenic and polygenic disorders	2
	International HapMap Project	2
	Whole genome association studies	2
	Genetics of Indian population	2
	Impact of population genetics in deciphering population-	2
	specific genetic risk factors	
	Ayurgenomics	2
	Genome variation and differential drug response	2
	Genetic risk factors for infection	2
	Epigenetics of lifestyle diseases	2
	Student presentation on selected topics	2
	Quiz	2