BIRLA INSTITUTE OF TECHNOLOGY AND SCIENCE, PILANI
INSTRUCTION DIVISION
FIRST SEMESTER 2016-2017
(Course Handout)

Dated: 02.08.2016

In addition to part I (general handout for all courses appended to the timetable) this portion gives further specific details regarding the course.

Course No : **BIO G612**
Course Title : **Human Genetics**
Instructor-In charge : **Dr. Sudeshna Mukherjee** :

1. Course Description:
The course will provide a survey on the current status of human genetics with an equal emphasis on molecular-genetic, genomic and population genetic approaches. Specific problems in human genetics will be addressed with examples from molecular genetics of common traits and genetic disorders.

2. Scope & Objective:
The course is aimed at making the student well-versed with the methods of identification and analysis of human genes in health and disease. Different human genetic approaches will be discussed in the context of disorders due to inherited or acquired mutations.

Text Books:
1. Strachen,T. and Read A. (2011). Human Molecular Genetics, 4th Edition. Garland Science Publishers, Abingdon UK.
   This would be the main book from where the lectures will be delivered.

2. Nussbaum, R.L, MicInnes, R.R and Willard H.F. (2011). Thompson and Thompson's Genetics in Medicine, 7th Edition, Saunders-Elsevier Publishers, Indian Edition, Noida UP.
   A relatively cheaper book which gives some basic outlines but not as exhaustive as T1.

Resources:
Study material related to some topics will be provided as and when required.

Reference books:
1. Speicher M, Antonarakis, S.E. and Motulsky, A.G. (2010). Vogel and Motulsky's Human Genetics: Problems and Approaches, 4th Edition. Springer Publishers.
2. Sudbery P., Sudbery I. (2009) Human Molecular Genetics, 3rd Edition. Pearson (Benjamin Cummings) Publishers.

BIO G612 Human Genetics
Tentative course handout

Lectures	Learning objective	Topic	Ref.
1	**Outline of the course, discussion with students and history of Human Genetics**	**Introduction**	**Lecture Notes**
2-6	**Sequences constituting the Human genome, Comparative genomics, Human chromosomes and their abnormalities.**	**Organization of the Human Genome and Chromosomes**	**Lecture Notes, T1: Chapters 9-10. T2: Chapter 2**
7-10	**DNA replication, Repair and Recombination and Defects in DNA transactions.**	**Maintenance of the Human Genome**	**T1: Chapter 13 Lecture Notes**
11-16	**General concepts of gene regulation, epigenetics, epigenome mapping, X-inactivation, genomic imprinting and its role in development.**	**Human Gene expression & Regulation**	**T1: Chapter 11 Lecture Notes T2: Chapter 3, parts of chapter 7**
17-22	**Genes in pedigrees & population genetics, Human Genome project, Identifying human disease genes and susceptibility factors**	**Molecular-Genetic Methods of Analysis**	**T1: Chapters 3, 14,16 ,Chapters 1-2 Lecture Notes T2: Chapters 10-12**
22-25	**Oncogenes & tumor suppressor genes, cell cycle dysregulation, genomic instability, genome-wide views of cancer and multi-stage evolution of tumor.**	**Cancer Genetics**	**T1: Chapter 17 Lecture Notes T2: Chapter 16**
28-33	**Types of variation between human genomes, pathogenic DNA variants, understanding the effects of variants and genotype-phenotype relationships.**	**Human Genetic variability and its consequences**	**T1: Chapter 13 Lecture Notes T2: Chapter 9**
32-34	**Mutation detection, gene tracking and DNA profiling**	**Genetic testing of individuals**	**T1: Chapter 18 T2: Chapter 17**
35-40	**Treatment using drugs, recombinant proteins and vaccines, Cell-therapy, Gene therapy and gene transfection systems.**	**Genetic approaches to treating disease**	**T1: Chapter 21 T2: Chapter 13**

Evaluation scheme:

Component	Duration	Weightage %	Date & Time	Remarks
Mid Sem	90 mins	20	<TEST_1>	CB
Quizzes, Assignments/(in lecture hours or announced)	Distributed throughout the semester either in lecture hrs or announced	20		CB/OB
Seminars and poster presentation	Distributed throughout the semester either in lecture hrs or announced	20		
Comprehensive	3 hrs	40	<TEST_C>	Partly OB

Chamber consultation hour: To be announced

Notices: All notices will be displayed on the Dept. of Biological Sciences notice board.

Make-up policy: Make-up will be granted only if candidate is hospitalized and in genuine cases as decided by the IC. No make-up will be granted in quizzes under any circumstances.

Instructor-in- charge
BIO G612