Birla Institute of Technology & Science, Pilani
Hyderabad Campus

FIRST SEMESTER 2019-2020

(Course Handout Part II)

Dated: 01.08.2019

In addition to Part-I (General Handout for all courses appended to the time table) this portion gives further specific details regarding the course.

Course No. **: BIO G612**
Course title **: Human Genetics**
Instructor-in-charge **: Piyush Khandelia**
Instructors **: N/A**

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 populations, health and disease. Different human genetic approaches will be discussed in detail in the context of disorders due to inherited or acquired mutations.

3. Text Books:

1. Human Molecular Genetics: Strachen, T. and Read, A., 4th Edition; Garland Science Publishers, UK, 2011. (This would be the main book from where the lectures will be delivered)
2. Thompson and Thompson's Genetics in Medicine: Nussbaum, R.L., McInnes, R.R. and Willard H.F, 7 th Edition; Saunders-Elsevier Publishers, Indian Edition, 2011. (Gives basic outline of the course, but not as exhaustive as T1)

4. Reference books:

1. Vogel and Motulsky's Human Genetics: Problems and Approaches: Speicher M., Antonarakis, S.E. and Motulsky, A.G., 4th Edition; Springer Publishers, 2011

5. Resources:

Study material related to some topics will be provided as .pdf versions.

6. Course plan:

Lectures	Learning objectives	Topics to be covered	Reference
1-3	Knowing the history of Human Genetics and its current status	Introduction, Garrod's in-born errors of metabolism, discovery of sickle cell mutation, human cytogenetics, mapping the human genome, human genome project, human genetics in the post-genome project era.	Class Notes T1: Chapter 2 T1: Chapter 9 T1: Chapter 12
4-7	Understanding the mode of transmission of genes and interactions between genes	Mendelian Inheritance in man, pedigree analysis, epistasis, X-linked inheritance, probability and gene ratios, multiple alleles and polygenic inheritance	T1: Chapter 3 T2: Chapter 2 Class Notes
8-12	Identification of a disease-causing locus for single gene disorders	History of genetic maps: two- and three-point mapping for determining gene order, tetrad analysis in yeast and *Neurospora*; mapping disease-causing genes using human pedigrees, LOD scores, Mapping function, Multi-point mapping	T1: Chapter 14 Class Notes
13-17	Identification of genes conferring susceptibility to complex disorders	Family studies of complex diseases, Segregation analysis, Linkage analysis of complex characters, Association studies and linkage disequilibrium, Association studies in practice, Limitations of association studies.	T1: Chapter 15
18-21	Understanding the variability in phenotypes because of mutations and allelic interactions	Types of variation between human genomes, pathogenic DNA variants, Molecular pathology, Genotype-phenotype correlations	T1: Chapter 13
22-27	Studying how gene frequencies vary in populations and understanding the factors influencing the gene frequencies	Hardy-Weinberg equilibrium and its extensions, Calculation of mutation rates, inbreeding coefficients in pedigrees, mutation-selection effects, human migration, small population sizes and genetic drift	Class Notes
28-31	Understanding the genetic susceptibility of cancer and cancer-causing mechanisms	The evolution of cancer, Oncogenes and tumor suppressor genes, Cell cycle dysregulation in cancer, instability of the genome, genome-wide views of cancer	T1: Chapter 17 Class Notes
32-36	How genetic tests are conducted in a clinical setting	What to test and why, Scanning a gene for mutations, testing for a specified sequence change, some special tests, gene tracking, DNA profiling	T1: Chapter 18
37-39	Understanding how genetic variants can provide prognostic value and criteria for personalized medicine	Pharmacogenetics and Pharmacogenomics, Personalized Medicine: prescribing the best drug and testing for susceptibility to complex diseases.	T1: Chapter 19 T1: Chapter 17 T2: Chapter 16

40-42	Current status and future approaches to treat genetic disorders	Treatment using drugs, recombinant proteins and vaccines, Cell-therapy, Gene therapy and gene transfection systems, RNA and oligonucleotide therapeutics, Gene therapy in practice.	T1: Chapter 21

Laboratory plan:

S. No.	List of experiments
1	Genotyping of single nucleotide polymorphisms by amplification refractory mutation system (ARMS) PCR
2	Profiling of variable number tandem repeats (VNTR) in genetically distinct individuals by PCR
3	To perform site-directed mutagenesis by inverse PCR
4	Analysis of DNA methylation by bisulphite conversion followed by PCR

7. Evaluation scheme:
Open book component would be 50% and would be components of laboratory evaluation, assignments and comprehensive exams.

Component	Duration	Marks	%	Date and Time	Venue	Remarks
Mid-semester exam	90 min	40	20%	04.10.19 (3.30-5.00PM)		Closed book
Laboratory Evaluation	-	60	30%	TBA		Open Book
Assignments		40	20%	TBA		Open Book
Comprehensive exam	180 min	60	30%	12.12.19 (AN)		Closed book

8. Chamber consultation hour: To be announced

9. Notices: All notices will be displayed on the Biological Sciences Group notice board.

10. Grading policy: Students missing one or more component of evaluation completely will be given an NC.

11. Make-up policy: As per the clause 4.07 in the Academic regulations booklet. Make-up will be granted only if candidate is sick and hospitalized. No make-up will be granted in quizzes under any circumstances.

12. Academic Honesty and Integrity Policy: Academic honesty and integrity are to be maintained by all the students throughout the semester and no type of academic dishonesty is acceptable.

INSTRUCTOR-IN-CHARGE
BIO G612