

**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, 7th 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:**

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| **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:**

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| **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.

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| **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**