20/02/2025

Printing Pages :1

Paper Code: PGDCG-101 C (SVSU:2024-25/R)

Enrollment No.

Post Graduate Diploma in Clinical Genetics (PGDCG) Ist Semester

Subject: Basic Human Genetics

Time: 3:00 Hours **Max. Marks: 70**

Q.I: Write long notes on any three. **(10x3=30)**

a) Describe the principles and application of non-Mendelian inheritance?
b) Discuss Genetic Susceptibility & risk factors.
c) Describe the role of pedigree analysis in human genetics.
d) Describe the complication in pedigree analysis in details.
e) Describe the Replication and their application in flow of genetic information.

Q.II: Write short notes on any five. **(5x5=25)**

a) What are the applications of gene mapping?
b) Describe the role of mRNA in details.
c) What is the role of migration and mutation in changing allele frequencies?
d) Describe the LOD equation for genetics?
e) Describe the Genetic code.
f) What is the purpose of a family pedigree?
g) Differentiate between dominance, co-dominance and incomplete dominance.

Q.III: Write Very short notes on any five. **(1x5=05)**

a. What is the purpose of a family pedigree?
b. Write the name to purine and pyrimidine.
c. How to calculate map distance between two genes?
d. How are the alleles of a gene different from each other?
e. What are the two basic rules of probability in genetics?
f. Define Linkage.
g. Write the features of double helix structure of DNA.

Q.IV Attempt all multiple Choice Questions. Select the correct answer only. **(1x10=10)**

a. The tendency of two or more than two genes to stay together during inheritance is called.....
i. Genetics ii. Gene interaction iii. Crossing over iv. Linkage

b. Failure in which of the following phenomenon would result in linkage?
i. Law of dominance ii. Law of segregation iii. Law of segregation iv. Law of separation

c. Height of humans is a....
i. Monogenic Trait ii. Polygenic Trait iii. Both of these iv. None of these

d. If a person had Hh in the H antigen locus and I^A i in the blood group locus what would be the blood group?
i. A ii. B iii. AB iv. O

e. Colour blindness is an _____ linked recessive trait
i. Z chromosome ii. X chromosome iii. Y Chromosome iv. None of these

f. An individual's collection of genes is called
i. Genetype iii. Phenotype iii. Trait iv. None of these

g. Name the phenomenon which can break the occurrence of linkage?
i. Crossing over ii. Linkage iii. Reconstruction iv. Breakage.

h. Name the organism whose first genetic map was made?
i. Rat ii. Arabidopsis iii. Fly iv. Drosophilla

i. How many loci the genes responsible for skin colour are present?
i. 2 ii. 3 iii. 4 iv. 5

j. ABO blood group is not an example of:
i. Co dominance ii. Multiple allele iii. Epistasis iv. Mendalian relations

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29/02/2025

Printing Pages :1
Paper Code: PGDCG-102 A (SVSU:2024-25/R)
Enrollment No.

PG Diploma in Clinical Genetics Ist Semester
Subject: Human Cytogenetics

Time: 3 Hours **M.M. 70**

Note: All questions are compulsory. Make diagrams wherever required.

Q1. Long answer type questions. Attempt any three. **(10x3=30)**

a. Write a detail note on the cell cycle with illustration.
b. Explain the different type of chromosome banding with proper illustration.
c. Elaborate the process of cryopreservation of human cell lines.
d. Elaborate the disorders associated with different autosomal anomalies.
e. Elaborate the different types of human cell cultures based on their culture technique.

Q2. Short answer type questions. Attempt any five. **(5x5=25)**

a. What is ploidy? Describe with appropriate examples.
b. Explain the clinical significance of ring chromosome.
c. Explain the salient features of cell-line maintenance.
d. Explain the significance of interphase.
e. Explain the function of Barr body.
f. Elaborate the clinical significance of chromosomal structural duplication.
g. Write a note on *in-situ* hybridization and its significance in diagnostics.

Q 3. Very short answer type questions. Attempt any five. **(1x5=5)**

a. What is band resolution?
b. What is the numerical significance of chromosomes in meiosis?
c. What is a chromatin?
d. Define polyploidy.
e. What are isochromosomes?
f. How chromosomal translocation can be diagnosed?
g. What is C-banding?

Q 4. Multiple choice questions.Attempt all. **(1x10=10)**

a. Mark the process which does not involve in histone modification
i. Acetylation ii. Methylation iii. Phosphorylation iv. Dehydration
b. Structure of DNA and protein found in the nucleus of eukaryotic cells.
i. Nucleic acid ii. Nucleosome iii. Chromatin iv. Tetraplex
c. Which of the following disease is caused by the mutation of desmosomes?
i. Parkinson's disease ii. Down's syndrome
ii. iii. Cardiomyopathy iv. Bubble Boy syndrome
d. In FISH technology, which fluorescence confirms a BCR/ABL translocation?
i. Red ii. Green iii. Yellow iv. Orange
e. Nucleosome is made up of --
i. DNA, histone core protein ii. DNA, histone core protein, linker H1
iii. RNA, histone core protein iv. RNA, histone core protein, linker H1
f. Cells that have stopped dividing and are arrested in a state preceding that of DNA synthesis, are said to be in the --
i. S-phase ii. G1-phase iii. G0-phase iv. G2-phase
g. The X-chromosome once inactivated remains inactive throughout the life in all the cells of the body:
i. True ii. False
h. BRCA1 and BRCA2 can cause which cancer?
i. Mammary adenocarcinomas ii. Colorectal adenomas
iii. Skin tumors iv. Lung adenocarcinomas
i. Pseudogenes are --
i. Non-functional ii. Non-existent
iii. Existent in plants only iv. Cannot be sequenced
j. Which of the following plays a substantial role in linking together sister chromatids immediately after replication?
i. Cohesins ii. Condensins iii. Histones iv. Topoisomerases

24/02/2025

Printing Pages :1

Paper Code: PGDCG-103 **C (SVSU:2024-25/R)**

Enrollment No.													

Post Graduate Diploma in Clinical Genetics (PGDCG) Ist Semester
Subject: Biochemistry and Instrumentation
Paper Code: PGDCG-103

Time: 3:00 Hours **Max. Marks: 70**

Q.I: Write long notes on any three. **(10x3=30)**

a. Describe the principle, application and types of Centrifuge.
b. Discuss the symptoms, diagnosis and treatment of Phenylketonuria.
c. Describe the symptoms and diagnosis of hypo and hyperthyroidism.
d. Describe the classification and functions of Vitamin.
e. Describe the principle, application of Mass spectrometry.

Q.II: Write short notes on any five. **(5x5=25)**

a. Describe the essential and nonessential amino acid.
b. Write the biochemical function and deficiency symptoms of Vitamin K.
c. Write the Biochemical function and deficiency symptoms of Calcium.
d. Describe the symptoms, diagnosis and treatment of Duchenne Muscular Dystrophy (DMD).
e. Describe the nucleic acid with their structure.
f. What are radioisotopes? Write the use of radioisotopes in medicine
g. Describe the starch and glycogen.

Q.III: Write Very short notes on any five. **(1x5=05)**

a. Write the deficiency symptoms of Vitamin E.
b. Write the deficiency symptoms of Sodium.
c. What is the use of Iodine-131.
d. Write the name of Basic amino acid.
e. Write the principle of ELISA.
f. Write the hormones related to fertility.
g. Which test is used to detect urea in urine?

Q.IV: Attempt any four multiple Choice Questions. Select the correct answer only. **(1x10=10)**

a. Which test is used to detect albumin in urine?:-
i. Benedict's test ii. Heat and acetic acid test iii. DAMO Test iv. All of these.

b. The structure of Hemoglobin is
i. Primary ii. Secondary iii. Tertiary iv. Quaternary

c. The deficiency of Vit D is
i. Beri beri ii. Rikets iii. Xerophthalmia iv. None of these

d. Spectrophotometer is based on
i. Lambert law ii. Beer's law iii. Both of these iv. None of these

e. LH is secreted by
i. Pituitary ii. Testes iii. Ovary iv. Adrenal cortex.

f. Which of the following is act as the most essential nutrient for a woman during the initial stages of pregnancy to prevent birth defects?
i. Thiamine ii. Folic acid iii. Vitamin C iv. Vitamin E

g. Riboflavin is the-
i. Vitamin B_3 ii. Vitamin B_1 iii. Vitamin B_2 iv. Vitamin

h. Ultraviolet spectrum of light is
i. 200-360 nm ii. 380-720 nm iii. 720-1000 nm iv. 100-200 nm

i. The sugar present in RNA is
i Ribose ii. Deoxyribose iii. Both of these iv. None of these

j. The m-RNA is used in the synthesis of.
i. Lipid ii. Carbohydrate iii. Protein iv. None of these

27/02/2025

Printing Pages :1
Paper Code: PGDCG-104 A (SVSU:2024-25/R)

Enrollment No.														

PG Diploma in Clinical Genetics
I Year/ Ist Semester
Subject: Genomics

Time: 3 Hours **M.M. 70**

Note: All questions are compulsory. Make diagrams wherever required.

Q1. Long answer type questions. Attempt any three. **(10x3=30)**

a. Explain the different components of eukaryotic genome.
b. Explain the type of heritance observed in mitochondrial genome.
c. Explain with proper example the significance of pharmacogenomics towards better clinical outcome
d. Explain the clinical significance of human mutation database with appropriate examples
e. Differentiate between intron and exon in terms of both structure and function.

Q2. Short answer type questions. Attempt any five. **(5x5=25)**

a. Differentiate between gain-of-function and loss-of-function mutation with appropriate example.
b. Differentiate between non-sense mutation and missense mutation.
c. Which one is lethal ? a) insertion, b) deletion or c) frame-shift mutation. Provide justification.
d. Explain the characteristic feature of mitochondrial genome.
e. Write a note on the database/s of human mutation.
f. What is meant by mutation rate?
g. Write a short note on genetic code with appropriate illustration.

Q 3. Very short answer type questions. Attempt any five. **(1x5=5)**

a. Necessity of DNA compaction.
b. Significance of missense mutation.
c. Loss-of-function mutation.
d. Functional significance of euchromatin.
e. Pharmacogenetics
f. Microsatellite marker usage
g. Phocomelia.

Q 4. Multiple choice questions.Attempt all. **(1x10=10)**

a. Which of the following is the start codon?.
i. CUC ii. ACC/CCA iii. GUA iv. AUG/GUG
b. Multiple copies of RNA could be formed at the same time.
i. True ii. False
c. Which of the following statements is true with respect to the DNA double helix?
i. Composed of two or more polynucleotide chains ii. The base pairs have opposite polarity
iii. Covalent bond exists in base pairing iv. T is transcribed as U
d. What proportion of mitochondrial proteins are coded by nuclear DNA?
i. 40% ii. 0% iii. 10% iv. 95%
e. Mitochondrial DNA is one of the best marker tools for population biologists and evolutionary biologist because
i. Absence of genetic recombination in mt-DNA ii. Mitochondrial genes are specific to mt-DNA
iii. It can be easily isolated iv. It undergoes spontaneous mutation
f. Which of the following is the clinical application of SNP and human mutation database?
i. Biomarker discovery ii. Molecular diagnostics iii. Prognosis iv. All of the above
g. Why compaction of DNA is necessary?
i. Inhibit transcription process ii. Protect DNA damage
iii. Fit large amounts of DNA into the cellular structure iv. All of the above
h. Which type of DNA structure possess the capability of condensation and decondensation?
i. Heterochromatin ii. Telomeric iii. Euchromatin iv. Centromeric
i. Metabolomics is employed for which of the following?
i. New born screening ii. Precision medicine iii. Drug development iv. All of the above
j. The triplet code allows many amino acids to be specified by more than one codon. Such a code is said to be degenerate?
i. True ii. False