



HCM-003-0038502

Seat No. _____

**B. Voc. (Medical Laboratory & Molecular
Diagnostic Technology) (Sem. V) (CBCS)
Examination**

October – 2017

MLMDT-5.2 - Clinical Genetics

Faculty Code : 003

Subject Code : 0038502

Time : $2\frac{1}{2}$ Hours]

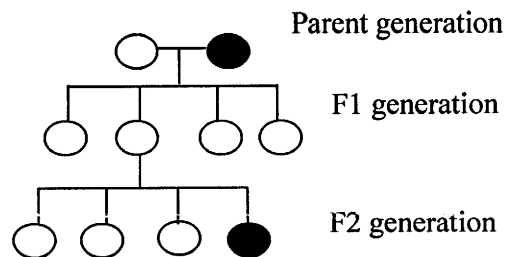
[Total Marks : 70

1 Answer the following questions : 20

- (1) When the genotype consists of a dominant and a recessive allele, the phenotype will be like _____ allele.
- (2) Assuming that both parent plants in the diagram below are homozygous, why would all of the f1 generation have yellow phenotypes?

○ Yellow pea

● Green pea



- (a) because the f1 genotypes are homozygous
 - (b) because yellow is dominant over green
 - (c) because both parents passed on yellow alleles
 - (d) None
- (3) How many types of gametes will be produced by an individual of AABbCcDd ?
 - (4) Which phase of cell cycle is associated with formation of synaptonemal complex?
 - (5) Which tumor suppressor protein is responsible for activation of p21 inhibitor protein?
 - (6) Growth factor binds with receptor at _____ point in mammals for activation of cyclin D.
 - (7) Crossing over is more frequent in female, state whether it is true or false?

- (8) Write Hardy - Weinberg law.
- (9) Define: isochromosome.
- (10) Give reason of arresting cell division at Metaphase for karyotyping.
- (11) Define gene and genome.
- (12) Define : teratogen
- (13) Define: spina bifida
- (14) Males are more often affected by sex-linked traits than females because
 - (a) males are hemizygous for the X chromosome.
 - (b) male hormones such as testosterone often alter the effects of mutations on the X chromosome.
 - (c) female hormones such as estrogen often compensate for the effects of mutations on the X.
 - (d) X chromosomes in males generally have more mutations than X chromosomes in females.
- (15) Which of the following statements describes genomic imprinting?
 - (a) It explains cases in which the gender of the parent from whom an allele is inherited affects the expression of that allele.
 - (b) It is greatest in females because of the larger maternal contribution of cytoplasm.
 - (c) It may explain the transmission of Duchenne muscular dystrophy.
 - (d) It involves an irreversible alteration in the DNA sequence of imprinted genes.
- (16) Which of the following is the most commonly used differential stain for chromosome analysis?
 - (a) Leishman
 - (b) Quinacrine
 - (c) Crystal violet
 - (d) Giemsa and Quinacrine
- (17) Women with Turner syndrome have a genotype characterized as karyotype 45, XX
True/False.
- (18) A cell that has $2n + 1$ chromosomes is
 - (a) trisomic
 - (b) monosomic
 - (c) euploid
 - (d) polyploid
- (19) Define : FISH
- (20) Which disease is common in family of Queen Victoria?

- 2 (a) Attempt any **three** of the following : **3×2=6**
- (1) Explain Test cross.
 - (2) Write Mendel's law of independent assortment.
 - (3) What is the importance of MPF in prophase of mitosis?
 - (4) What is the role of CDK in cell cycle?
 - (5) Define coupling gametes and repulsive gametes.
 - (6) Explain synapsis.
- (b) Attempt any three of the following : **3×3=9**
- (1) In a heterozygous, heterozygous dihybrid cross, the following data was obtained:
Dominant for both traits: 570,
Dominant for trait 1 and recessive for trait 2:185
Dominant for trait 2 and recessive for trait 1:190,
Recessive for both traits: 55
Perform a chi-square analysis to see if the data above agrees with the predicted outcome of this cross.
(According to chi square table the value at 3 degree of freedom is 7.82.)
 - (2) State the rules for Inheritance of Autosomal recessive traits in Man.
 - (3) How many check points are present in cell? Discuss the role of check point in cell cycle.
 - (4) Write down the events of anaphase promoting complex.
 - (5) Give the Statement of Hardy-Weinberg law of population genetics. Briefly write its significance.
 - (6) Discuss theories of crossing over. Give the significance of crossing over.
- (c) Attempt any **three** of the following : **2×5=10**
- (1) What is crossing over? Explain the mechanism of Crossing over.
 - (2) Define Linkage. Explain types of linkages with example.
 - (3) Discuss in detail Mendelian inheritance pattern and law of heredity.

- (4) Diagrammatically explain Meiosis I cell division including role of MPF in meiosis.
- (5) Write down the reasons and impact of error in cell division.

3 (a) Attempt any **three** of the following : **3×2=6**

- (1) Explain uniparental disomy with example.
- (2) Differentiate between types of inversion.
- (3) Differentiate transcervical and transabdominal CVS.
- (4) Enlist chromosomal abnormalities in CML.
- (5) Give advantages of Q banding.
- (6) Give symbol of dizygotic and monozygotic twins.

(b) Attempt any **three** of the following : **3×3=9**

- (1) Explain genomic imprinting in context of fragile X syndrome.
- (2) Note on dicentric chromosome.
- (3) Enlist diagnostic significance of PND.
- (4) Note on quadruple screen test.
- (5) Note on nomenclature of chromosome.
- (6) Advantages of charge coupled device cameras

(c) Attempt any **three** of the following : **2×5=10**

- (1) Write a brief note on types of numerical abnormality.
- (2) Brief note on chorionic villus sampling.
- (3) Brief note on FISH.
- (4) Discuss pedigree analysis and write its importance.
- (5) Explain Fluorescence microscopy.