

MAK-003-038502 Seat No. _____

B. Voc. Medical Laboratory & Molecular Diagnostic Technology (Sem. V) (CBCS) Examination October / November - 2016

MLMDT - 5.2 : Clinical Genetics

Faculty Code : 003 Subject Code : 038502

Time : $2\frac{1}{2}$ Hours] [Total Marks : 70

Instructions: (1) All questions are compulsory.

(2) Figures to right indicate marks.

1 Answer the following questions:

 $20 \times 1 = 20$

- (1) What is an organism with two identical alleles called?
- (2) How many types of gametes will be produced by an individual of AABbCcDd?
- (3) What is the allele which is unable to express its effect in the presence of another is called?
- (4) Blue Eye Color is recessive to brown eye color. A brown eyed man whose mother is blue eyed marries blue eyed woman. The children will be
 - (a) Both blue eye and brown eyed 1:1
 - (b) All brown eyed
 - (c) All blue eyed
 - (d) Blue eyed and brown eyed 3:1
- (5) What would be the probability of AABBCC individuals from a mating of two AaBbCc individuals?
- (6) If amount of DNA is 2X in G1 phase, what will be the amount of DNA in G2 phase?
- (7) Crossing over occurs at _____ stage of meiosis.
- (8) How many linkage groups are present in Drosophila, when it has 4 homologous pairs of Chromosomes?
- (9) Crossing over is more frequent in female; state whether it is true or false?
- (10) What is charge couple device?

- (11) What are results of teratogen exposure? (12) Define: genomic imprinting. (13) Define: Oligohydramnios. (14) Define: Philadelphia chromosome. (15) Define: Bcr-abl fusion protein (16) Name dye used in Q banding. (17) Define: WCP (18) Define: Pedigree analysis (19) Draw symbol of consangious marriage. (20) Give symbol of sex unidentified. Attempt any three of the following: $3 \times 2 = 6$ (a) (1) Define: Genotype and Phenotype. **(2)** Write Mendel's law of segregation. (3) Write down the role of MPF in prophase of mitosis. **(4)** Define synapsis and crossing over. **(5)** Explain uniparental disomy with example. (6) Differentiate between angelman and prader-willi syndrome. Attempt any three of the following: $3 \times 3 = 9$ (b) A crossing of two hybrid of a flower species the (1) result is shown below. Are these results consistent with expected proportion 9:3:3:1? Magenta Flower 120 Green Stigma
 - Magenta Flower
 Green Stigma

 Magenta Flower
 49
 Red Stigma

 Red Flower
 36
 Green stigma

 Red Flower
 12

Red Stigma

(According to chi square table the value at 3 degree of freedom is 7.82)

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- (2) State the rules for Inheritance of Autosomal Dominant Traits in Man.
- (3) List the factors affecting Linkage.
- (4) What are CDK inhibitors? Write the name of any two CDK inhibitors.
- (5) Explain mechanism of nondisjunction.
- (6) Enlist points of suspect for chromosomal defect.
- (c) Attempt any two of the following: $2\times 5=10$
 - (1) What is Monohybrid and Dihybrid Cross? State Mendel's Laws of Inheritance.
 - (2) What is protein kinase? What is the role of protein kinases in Cell Cycle?
 - (3) Define Gene frequency and genetic frequency. Explain the Hardy Weinberg law of equilibrium.
 - (4) Brief note on types of numerical abnormality.
 - (5) Elucidate reasons of getting chromosomal defect.
- 3 (a) Attempt any three of the following: $3\times2=6$
 - (1) Enlist non invasive methods of PND.
 - (2) What are effects of insertion?
 - (3) List function of amniotic fluid.
 - (4) Enlist types of probe labelling in FISH.
 - (5) Give importance of pedigree chart.
 - (6) Applications of phase contrast microscopy.
 - (b) Attempt any three of the following: $3\times3=9$
 - (1) Note on quadruple screen test.
 - (2) Note on Teratogen.
 - (3) How many check points are present in cell? Discuss the role of check point in cell cycle.
 - (4) Discuss variants of G banding.
 - (5) Give list of symbols used in pedigree analysis.
 - (6) Principle and advantages of dark field microscopy.

- (c) Attempt any two of the following: $2\times 5=10$
 - (1) Brief note on amniocentesis.
 - (2) Brief note on cytogenetic analysis of human embryo.
 - (3) Note on Q banding.
 - (4) Note on pedigree analysis.
 - (5) Note on types of inheritance pattern.