CS/B.Sc.(H)/(Genetics/Mol-Bio.)/SEM-6/MLG-603/2013

2013

MOLECULAR HUMAN GENETICS

Time Allotted : 3 Hours *Full Marks :* 70

The figures in the margin indicate full marks.

Candidates are required to give their answers in their own words as far as practicable.

GROUP – A

(Multiple Choice Type Questions)

1. Choose the correct alternatives for any *ten* of the following : 10x1 = 10

i) Which chromosome possesses lowest number/density of genes ?

a) 13

b) Y

c) 1

d) 22.

ii) Locus heterogeneity means

a) same disease by mutations at different loci

b) existence of many different disease-causing alleles at same locus

c) complex patterns of inheritance occurs from the same locus

d) variable expression of a gene in the same locus.

iii) Which year map based Cystic Fibrosis Gene was discovered on chromosome 7q ?

a) 1987

b) 1989

c) 1992

d) 1998.

iv) Which type of vectors are mostly used in gene therapy clinical trials ?

a) Retrovirus

b) Pox virus

c) Vaccinia virus

d) Adeno-associated virus.

v) First human gene therapy trial was done using

a) MoMLV-based delivery vector

b) Liposome-based delivery

c) Human Artificial chromosome based delivery

d) Non-viral based delivery.

vi) Repetitive sequence within coding region causes

a) Huntington disease

b) Fragile X syndrome

c) Baker Muscular dystrophy

d) Cystic fibrosis.

vii) Repetitive sequence outside coding region causes

a) Fragile X syndrome

b) Baker Muscular dystrophy

c) Huntington disease

d) Cystic fibrosis.

viii) Which year map based Huntington Disease Gene was discovered on chromosome 4p ?

a) 1990

b) 1993

c) 1997

d) 1998.

ix) Which year Human Genome Project was Proposed ?

a) 1982

b) 1985

c) 1988

d) 1990.

x) Diagnosis of Ambigous phenotype of a newborn baby is done by

a) Physical examination

b) Genetic test

c) Hormonal test

d) Ultrasound.

xi) The presence of both ovarian and testicular tissue in the same individual known as

a) female pseudohemaphroditism

b) true hermaphroditism

c) XY-FEMALE

d) mixed gonadal dysgenesis.

xii) The first artificial chromosome is synthesized in the year

a) 1981

b) 1983

c) 1986

d) 1984.

xiii) The DNA fingerprinting technique is developed in the year

a) 1978

b) 1982

c) 1984

d) 1990.

GROUP – B

(Short Answer Type Questions)

Answer any *three* of the following. 3x5 = 15

2. What is haploinsufficiency ? What is its effect ? 2 + 3

3. Why genetic map differ from physical map ?

4. How to decide whether a non-Mendelian character is genetic or not ?

5. Define haplotype and gene conversion. What is the difference between gene conversion and recombination ?

2 + 2 + 1

6. Chaisma count give an estimate of the total map length. Explain.

GROUP – C

(Long Answer Type Questions)

Answer any *three* of the following. 3x15 = 45

7. What are loss of function mutation and gain of function mutation ? What is dominant negative effect ? Discuss dominant negative effect in Fibrillar collagens.

$2_{1/2} + 2_{1/2} + 3 + 7$

8. Discuss Loss of function mutation in PAX3 gene ? What epigenetic mechanisms can bring about mutational effects in genes ? Discuss all of them in short. 7 + 2 + 6

9. What are positional cloning and candidate gene mapping ? How positional cloning and candidate gene mapping help to identify the human disease gene ? Explain with suitable example. What is complex genetic dieseas ?

2 + 2 + 4 + 4 + 3

10. How chromosomal aberrations provide a useful short-cut to locate a disease gene ? Why multipoint mapping is more efficient than two point mapping ? What are the different genetic markers used in human disease gene mapping ?

5 + 5 + 5

11. What are contiguous gene syndrome and segmental aneuploidy syndrome ? Give example. Allelic heterogeneity is a common feature of loss of function phenotype. Explain.

5 + 5 + 5

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