2012

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 :

$$10 \times 1 = 10$$

i) Who coined the term "Genome" ?

a) Bateson b) Winkler

c) Johansson d) Mendel.

ii) Which Human chromosome is the largest acrocentric

chromosome ?

a) 13 b) 14

c) 15 d) Y.

iii) Which chromosome possesses lowest number/density of

genes ?

a) 13 b) Y

c) 1 d) 22.

iv) Allelic heterogeneity means

a) existence of many different disease-causing alleles

at same locus

b) same disease by mutations at different loci

c) variable expression of a gene in the same locus

d) complex patterns of inheritance occur from the

same locus.

v) In which year Map based Duchene Muscular Dystrophy

Gene was discovered on Chromsome Xp ?

a) 1982 b) 1986

c) 1987 d) 1988.

vi) In which year Human Genome Project was proposed ?

a) 1982 b) 1985

c) 1988 d) 1990.

vii) Diagnosis of ambigous phenotype of a newborn baby is

done by

a) Physical examination b) Genetic test

c) Hormonal test d) Ultrasound.

viii) The presence of both ovarian and testicular tissue in the

same individual is known as

a) Female pseudohermaphroditism

b) True hermaphroditism

c) XY-Female

d) Mixed gonadal dysgenesis.

ix) Interference of Gene Expression is due to

a) Missense mutation b) Epigenetic silencing

c) Nonsense mutation d) Unequal crossover.

x) In which year Celera Genomics published their draft of

the human genome in the journal *Science* ?

a) 1999 b) 2001

c) 2002 d) 2003.

xi) In which year Map based Roberts Syndrome Gene was

Discovered on chromosome 13q ?

a) 2000 b) 2005

c) 2006 d) 2008.

GROUP – B

(Short Answer Type Questions)

Answer any *three* of the following. $3 \times 5 = 15$

2. What are the methods of Gene Mapping ?

3. Define variable gene expression in complex genetic disorder.

4. What is Polygenic Disorder ? Explain with example.

5. What is Epigenetic silencing ? How is it related to Gene

Expression ?

6. How do Unstable Expanding Repeats in coding region affect

in Fragile X Syndrome ?

GROUP – C

(Long Answer Type Questions)

Answer any *three* of the following. $3 \times 15 = 45$

7. What is STS ? What are the type STSs available in Human

Genome ? What are the vectors used as a Library in Human

Genome Project ? Describe the Laboratory Protocol used in

Human Genome Project. $2 + 3 + 3 + 7$

8. What are Genetic Markers ? What are the types of Genetic

Markers used in Gene Mapping ? What is the molecular

basis of DNA fingerprinting ? Who is the inventor of the same

technology ? $2 + 3 + 8 + 2$

9. How do you sequence a whole Genome ? What is cDNA

sequencing ? What is current status of DNA sequencing

technology ? What is Bermuda agreement in HGP ?

10. What is Haplotype ? What is Genomic Variation ? How does

Haplotype help in searching genes in Complex Genetic

Disorders ?

11. What is Complex Genetic Disease ? Describe inheritance pattern in complex genetic disorders. What are the differences between monogenic and complex genetic disorders ? 3 + 6 + 6

============