

Name :

CS/B.Sc.(H)/GENETICS/MOL. BIO/SEM-6/MLG-603/2012 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

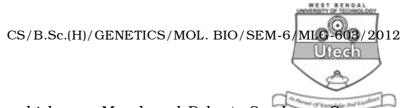
 c) 1
 d) 22.
 - iv) Allelic heterogeneity means
 - a) existence of many different disease-causing alleles at same locus
 - b) same disease by mutations at different loci
 - c) variable expression of a gene in the same locus
 - d) complex patterns of inheritance occur from the same locus.

[Turn over

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v)	In w	which year Map based Duchene Muscular Dystrophy				
	Gene was discovered on Chromsome Xp ?					
	a)	1982	b)	1986		
	c)	1987	d)	1988.		
vi)	In w	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)	viii) The presence of both ovarian and testicular tissue in same individual is known as			d testicular tissue in the		
	a)	Female pseudohermaphroditism				
	b)	`rue hermaphroditism				
	c)	XY-Female				
	d)	Mixed gonadal dysgenesis.				
ix)	x) 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.

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- xi) In which year Map based Roberts Syndrome Gene was Discovered on chromosome 13q ?
 - a) 2000 b) 2005
 - c) 2006 d) 2008.

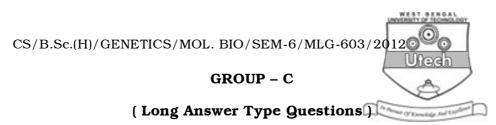
GROUP – B

(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 ?

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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

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