

**Answer the following questions. (60%)**

1. Please define the following biological terms: (20%, 4% each)
  - (1) Next generation sequencing techniques
  - (2) Linkage disequilibrium
  - (3) Bottleneck effects
  - (4) Comparative genomics
  - (5) Epistasis
2. What is meant by the term **pseudogene** and how are **pseudogenes** formed? (5%)
3. Please describe in details on the **transposition** of a **retrotransposon**? (5%)
4. What is meant by the term **suppressor mutation** and please list the types of suppressor mutations. (5%)
5. What is an **allele-specific** oligonucleotide and briefly describe its use? (5%)
6. In a cross between a heterozygous tall pea plant and a dwarf plant, predict the ratios of the offspring's genotypes and phenotypes. (6%)
7. What are "Genetic Model Organisms"? List three examples (species) and describe the characteristics that made them especially suitable for genetic research. (8%)
8. Why do mutations in non-coding regions of a gene still have major effects on phenotypic expression? (6%)
9. **Choose the correct answer (only one answer) for each of the questions. (40%, 2% each)**
  - (1) If an X-linked disease results from a rare recessive allele
    - A. there will be more females exhibiting the trait than males
    - B. there will be more males exhibiting the trait than females
    - C. the frequency of affected males and females will be equal
    - D. among females there will be more affected than carriers
    - E. all sons of affected males will be affected
  - (2) Mitochondrial DNA
    - A. carries all of the cell genes necessary for respiration
    - B. is a linear molecule
    - C. utilizes slightly different genetic code than nuclear genome
    - D. carries genes for all components necessary for its function
    - E. evolves slower than nuclear DNA

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- (3) Loss of function of p53
- A. activates apoptosis
  - B. eliminates the DNA damage checkpoint
  - C. increases contact inhibition
  - D. blocks activation of the anaphase promoting complex
  - E. suppresses assembly of the spindle
- (4) Ultraviolet light primarily damages DNA by
- A. pyrimidine dimers
  - B. forming purine dimers
  - C. alkylating DNA
  - D. depurinating DNA
  - E. none of the above
- (5) A test for carcinogens and mutagens that looks for an increased reversion frequency in a His<sup>-</sup> strain of bacteria is called the
- A. carcinogen test
  - B. mutagen test
  - C. Ames test
  - D. *Salmonella* reversion test.
  - E. auxotrophic reversion test.
- (6) A homeobox is a
- A. DNA binding site
  - B. sequence that codes for DNA binding motif
  - C. transcriptional activator of other genes
  - D. protein involved in the control of meiosis
  - E. protein involved in the control of replication
- (7) The nucleotide sequence of the attenuator region
- A. contains features of a termination site
  - B. triggers degradation of RNA transcripts containing homologous sequences
  - C. interacts with the Polycomb group of transcriptional silencer protein
  - D. regulates *lac* operon transcription
  - E. is the binding site of the cAMP-CRP complex

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- (8) Which of the following is true regarding RNA processing?
- A. spliceosomes are present in organelles and nuclei
  - B. it involves removal of exons
  - C. it may be autocatalytic
  - D. it occurs in prokaryotes
  - E. none of the above
- (9) The genetic code is degenerate because
- A. mRNA is rapidly degraded
  - B. the code is not universal among organisms
  - C. some amino acids have more than one codon
  - D. frameshift mutations are tolerated
  - E. stop codons may have corresponding tRNA molecules
- (10) Which of the following mutations is more likely to produce a severe rather than mild phenotypic consequence(s)?
- A. synonymous mutations
  - B. nonsense mutations
  - C. missense mutations
  - D. none of the above
  - E. all of the above
- (11) If mature eukaryotic mRNA is hybridized with its corresponding DNA coding strand (heteroduplex analysis) and visualized by electron microscopy, looping strands of nucleic acid are seen. What do these structures represent?
- A. lariat structures
  - B. inverted repeats
  - C. introns
  - D. exons
  - E. overlapping genes
- (12) Which of the following is unique to prokaryotes?
- A. coupled transcription-translation
  - B. removal of introns
  - C. 3' polyadenylation
  - D. mRNA capping
  - E. promoter sequences

- (13) The presence of a bacteriophage can be visualized on a lawn of *E. coli* cells by the formation of lytic foci called
- A. plaques
  - B. lysates
  - C. prophages
  - D. cistrons
  - E. lysogens
- (14) Some XY individuals are phenotypically females. What chromosomal abnormality could account for this?
- A. mosaicism
  - B. dosage compensation
  - C. a deletion of the portion of the Y chromosome containing the testis-determining factor
  - D. mitotic segregation
  - E. fragile X syndrome
- (15) In *Drosophila* the recessive alleles for brown and scarlet eyes (of two independent genes) interact so that  $bw/bw; st/st$  is white. If a pure-breeding brown is crossed to a pure--breeding scarlet, what proportion of the  $F_2$  will be white?
- A. 1/16
  - B. 1/4
  - C. 7/16
  - D. 3/4
  - E. 13/16
- (16) If an individual has 10 gene pairs, how many different gametes can be formed if five of the gene pairs are homozygous and the remaining 5 gene pairs are heterozygous?
- A. 42
  - B. 109
  - C. 32
  - D. 1024
  - E. cannot be determined

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(17) How many telomeres are there in a nondividing human liver cell?

- A. 1
- B. 2
- C. 23
- D. 46
- E. 92

(18) A wild type chromosome can be represented as ABC \* DEFGH, and from this a chromosomal aberration arises that can be represented ABC \* DEGFH. This is known as (\* = centromere)

- A. deletion
- B. translocation
- C. duplication
- D. pericentric inversion
- E. paracentric inversion

(19) An enhancer is best described as a:

- A. specialized DNA sequence that acts to promote expression of specific genes
- B. a transcription factor that acts to promote expression of specific genes
- C. a binding site for RNA polymerase
- D. a protein that binds to RNA polymerase, thereby modulating the rate at which RNA polymerase transcribes a given gene
- E. TATA box-containing DNA element

(20) In eukaryotes, translation usually begins:

- A. at the first set of three nucleotides adjacent to the methyl guanine cap
- B. at the first codon downstream of the methyl guanine cap
- C. at the first AUG downstream of the methyl guanine cap
- D. at the first AAG downstream of the methyl guanine cap
- E. at the first anticodon