

I. Choose the best explanation (A-J) for the following terms (1-10) (2 points each)

1. alternative splicing
2. attenuation
3. carrier
4. epigenetic
5. epistasis
6. haplotype
7. kinetochore
8. linkage disequilibrium
9. nondisjunction
10. RNA editing

- A. a gene interaction in which the effects of an allele at one gene hide the effects of alleles at another gene.
- B. a specialized chromosomal structure composed of DNA and proteins that is the site at which chromosomes attach to the spindle fibers.
- C. a state of gene functionality that is not encoded within the DNA sequence but that is still heritable from one generation to the next.
- D. a type of gene regulation in which transcription of a gene terminates in the regulatory region before a complete mRNA transcript is made.
- E. production of different mature mRNAs from the same primary transcript by joining different combinations of exons.
- F. specific alteration of the genetic sequence carried within an RNA molecule after transcription is completed.
- G. specific combination of linked alleles in a cluster of related genes.
- H. the failure of two sister chromatids to separate during mitotic anaphase generates reciprocal trisomic and monosomic daughter cells.
- I. unaffected parents who bear a dominant normal allele that masks the effects of an abnormal recessive one.
- J. when alleles at separate loci (such as marker alleles and disease alleles) are associated with each other at a significantly higher frequency than would be expected by chance.

II. Multiple choice (choose one best answer, 4 points each)

1. Which of the following is correct regarding the blending theory of inheritance?
 - A. It believed that hereditary traits blended from one generation to the next
 - B. It was possible for the blending to change the trait from one generation to the next
 - C. It was supported by early research by Joseph Kölreuter
 - D. It was the prevailing theory of inheritance prior to Mendel
 - E. All of the answers are correct

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2. In four-o'clock plants, red flower color is dominant to white flower color. However, heterozygous plants have a pink color. If a pink-flowered plant is crossed with a white-flowered plant, what will be the phenotypic ratios of their offspring?
- ¼ red, ½ pink, ¼ white
 - All pink
 - All white
 - ½ pink, ½ white
 - ½ red, ½ pink
3. Assume that genes C and D are located on the same chromosome. On one chromosome, alleles C and D are found, while the homologue contains alleles c and d. Which of the following would be an example of a recombination event?
- Alleles C and D together on one chromosome
 - Alleles c and d together on one chromosome
 - Alleles C and d together on one chromosome
 - Alleles c and D together on one chromosome
 - Both alleles C and d together on one chromosome and alleles c and D together on one chromosome
4. GGGCCATTCGAACGTCCGAAAATGCCCTGAATGAAAATTTGGCCC. The primer used for replication *in vitro* is CCCGGTAAGCTT. Where is the 5' end for the template and primer, respectively?
- left, left
 - right, left
 - left, right
 - right, right
 - not enough information
5. Based on the following mature mRNAs, what exons are constitutive?
- I. 1-2-3-4-7-8-10 II. 2-4-5-6-7-9 III. 1-4-6-7-8 IV. 1-2-4-7-10
- 1 and 2
 - 1 and 4
 - 2 and 7
 - 4 and 7
 - 1 and 8
6. Which of the following would indicate that a disease has a genetic, rather than environmental, cause?
- The disease has a specific age of onset
 - A high level of concordance among monozygotic twins
 - The disease does not occur in individuals that are exposed to a similar environment
 - The disease is similar to a known genetic disease in an animal such as a mouse
 - All of the answers are correct
7. In the following sequence of DNA, the italicized base has been mutated. What type of mutation is this?
- 5' - GATCTCCGAATT - 3' original strand
5' - GATCTCCCAATT - 3' mutated strand
- Transition
 - Transversion
 - Inversion
 - Deletion
 - None of above

8. After growing a culture of *E. coli* bacteria in the presence of the T1 phage, you discover that sub-cultures taken from the original exposure show a tremendous fluctuation in the number of colonies that are resistant to the T1 phage. This fluctuation supports which of the following theories?
- Physiological adaptation theory
 - Spontaneous mutation theory
 - Chromosome theory
 - Two-hit theories
 - None of above
9. After screening a colony of bacteria for a given gene, you discover 100 mutant colonies out of 3 million total colonies. What is the mutation frequency for this gene in the population?
- 1.0×10^5
 - 1.0×10^{-5}
 - 3.0×10^5
 - 3.3×10^{-5}
 - None of the answers are correct
10. Which of the following are examples of suppressor mutations?
- An intragenic mutation that restores protein structure
 - An intergenic mutation that increases the activity of a protein performing the same function as the mutated protein
 - An intergenic mutation that activates a transcription factor that dramatically upregulates expression of the mutant protein
 - Both A and C
 - All of the answers are correct
11. Using Mendel's flower color (purple is dominant, white is recessive), if a two heterozygous plants are crossed, what is the probability that the first two offspring will have purple flowers?
- 1/2
 - 1/4
 - 6/4
 - 9/16
 - 1/16
12. An individual with type A blood and an individual with type B blood mate and have offspring. What blood type is not possible in their offspring?
- Type O blood
 - Type A blood
 - Type B blood
 - Type AB blood
 - All blood types are possible
13. If a geneticist describes a trait as being 70% penetrant, what would they mean?
- The expression of the trait varies by individual
 - It is lethal in 30% of the individuals who have the trait
 - Only 70% of the individuals who carry the trait express the trait
 - The trait is present in 70% of the population
 - None of above
14. A heritability value of 0.997 indicates which of the following?
- The majority of the phenotypic variation has a genetic basis
 - The majority of the phenotypic variation has an environmental basis
 - The trait is polygenic

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- D. There is a significant difference between the two strains
- E. None of the answers are correct

15. Which of the following is NOT a characteristic by which a population is in equilibrium according to Hardy-Weinberg equation?
- A. The population is large.
 - B. There is no migration into or out of the population.
 - C. There is no selection against a given genotype.
 - D. There is no mutation in the gene being studied.
 - E. There is nonrandom mating

III. Short answer questions (20%)

1. What are the differences between iPS cells and ES cells? Are they both pluripotent cells? (5 points)
2. Explain genome-wide association studies. Do you know any trait(s) which were mapped/suggested by genome-wide association studies? (5 points)
3. In a given population of *Drosophila*, curly wings (c) is recessive to the wild-type condition of straight wings (c+). You isolate a population of 35 curly winged flies, 70 flies that are heterozygous for straight wings and 45 that are homozygous for straight wings. What is the total number of alleles in the gene pool? Using the information from the above question, what is the frequency of alleles, c and c+, in this population? (5 points)
4. List one classical model organisms and an key experiment in Genetics. (5 points)

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