

1. Indicate the true sentence. The effective size (N_e) of a population will be smaller than the census if:
 - a. There is a different number of males and females in the population
 - b. Bottlenecks have occurred in the population
 - c. The number of individuals fluctuates in different generations
 - d. All of the previous answers are correct

2. In a small population of 5 individuals there are two neutral alleles, B and b . In a particular generation, the frequency of B is 0.8. Indicate the false option:
 - a. The probability that the population is still polymorphic in the next generation is high
 - b. The probability that allele B is lost in the next generation is extremely low
 - c. The probability that allele B is fixed in the next generation is 0.1073
 - d. The probability that allele B frequency is exactly the same in the next generation is 0.00671

3. Which of the following is false?
 - a. Variants with alleles with frequencies higher than 1% are considered polymorphic
 - b. Variants located outside of coding regions cannot affect phenotype
 - c. SNPs, indels and inversions are mostly variants with two alleles
 - d. Copy number variants (CNVs) can have multiple alleles with different number of copies of a gene

4. Indicate which of the following statements about mutation is false:
 - a. As an evolutionary force, mutation alone is a weak force and allele frequency changes will take a long time
 - b. In a mutation-selection equilibrium the frequency in the population of a deleterious allele generated by mutation with a constant rate, will be higher if the deleterious allele is dominant
 - c. All variants found as polymorphisms in populations were generated by mutation as errors during DNA replication or repair in germ line cells
 - d. Mutations occur randomly

5. The fitness values for the genotypes AA , Aa , and aa in a population are 0.8, 1, and 0.4, respectively. Indicate the false sentence:
 - a. At equilibrium the average fitness of the population will be lower than 1
 - b. At equilibrium the frequency of heterozygotes will be 1
 - c. This population is evolving by balancing selection favoring the heterozygote
 - d. At equilibrium the population will have $p = 0.75$ and $q = 0.25$

6. A SNP with the alleles C and T in position 478 of human *MC1R* gene is responsible for the homozygote TT individuals having freckles and red hair. This gene encodes the melanocortin receptor and is involved in the synthesis of the melanin pigment in skin cells. In a random sample of 30 individuals of the United States with European origin we find 25 individuals with the CC genotype, 5 with the CT genotype, and none with the TT genotype. Indicate the false option:
 - a. The observed frequency of heterozygotes in this population is 0.167
 - b. The allele frequency of the red-haired allele T is 0.0833
 - c. The expected frequency of red-haired individuals is 0.00694
 - d. This population is not in Hardy-Weinberg equilibrium

7. Tay-Sachs disease is a fatal disorder in children causes a progressive degeneration of the central nervous system. It is caused by the absence of an enzyme called hexosaminidase A. This disease occurs in 1 in 27 newborns in persons of European Ashkenazi Jewish ancestry. Indicate the false answer:
- The frequency of carrier individuals in Ashkenazi Jews is 0.3108 -
 - We cannot test if the population is in Hardy-Weinberg equilibrium
 - A 32.3% of healthy individuals are carriers of the disease allele in the Ashkenazi Jews populations -
 - The allele frequency of the Tay-Sachs allele in Ashkenazi Jews is 0.037
8. In a population there is a gene with two alleles, A and a . Allele frequency of dominant allele A is 0.3. If, due to an environmental change, relative fitness values of genotypes AA , Aa , and aa become respectively 1, 1, and 0.36, which of the following statements is false?
- After the first round of selection, the frequency of allele A will be 0.437
 - When a new equilibrium is reached, allele frequencies will be 1 for allele A and 0 for allele a *
 - After the first round of selection, the frequency of aa individuals will be 0.1764
 - Allele a is recessive and detrimental
9. Indicate the true sentence. An allele with a frequency of 0.12 in a population of 25 individuals:
- Is more likely to be fixed than lost by genetic drift
 - Has a probability of fixation over time of 12%
 - Will have the same exact allele frequency in the next generation because no evolutionary mechanism is acting on this population
 - None of the other answers are true
10. Indicate if the following statements about migration among populations are true or false: (1) The allele frequency in a small population receiving migrants from a large population with constant allele frequency will eventually be the same than in the large population. (2) All populations involved in sending and receiving migrants will have the same intermediate allele frequency when they reach equilibrium.
- 1 false and 2 true
 - Both true
 - Both false
 - 1 true and 2 false
11. Indicate the false option among these statements about population structure:
- Without migration, allele frequencies in different populations will become different over time by the effects of genetic drift
 - If there is migration, the effects of genetic drift will be minimized and allele frequencies among different populations cannot diverge
 - The F_{ST} value will be higher with migration than without
 - An F_{ST} value of 1 indicates the maximum differentiation among populations

12. If a population is NOT in Hardy-Weinberg equilibrium, it means that:
- This population does not fulfill some of the assumptions of an ideal population: diploid organism, sexual reproduction, same allele frequencies in both sexes, or random mating
 - One of the evolutionary mechanisms that cause changes in allele frequencies over time is acting on this population
 - The observed genotype frequencies cannot be calculated from allele frequencies as p^2 , $2pq$ and q^2
 - The three previous answers are true

1. A SNP with the alleles C and T in position 478 of human MC1R gene is responsible for the homozygote TT individuals having freckles and red hair. This gene encodes the melanocortin receptor and is involved in the synthesis of the melanin pigment in skin cells. In a random sample of 30 individuals of the United States with European origin we find 25 individuals with the CC genotype, 5 with the CT genotype, and none with the TT genotype. Indicate the false option:

- The expected frequency of red-haired individuals is 0.00694
- This population is not in Hardy-Weinberg equilibrium
- The observed frequency of heterozygotes in this population is 0.167
- The allele frequency of the red-haired allele T is 0.0833

2. In a population there is a gene with two alleles, A and a. Allele frequency of A is 0.65. If, due to an environmental change, relative fitness values of genotypes AA, Aa, and aa become respectively 1, 1, and 0.27, which of the following statements is false?

- When a new equilibrium is reached, allele frequencies will be 1 for allele A and 0 for allele a.
- After the first round of selection, the frequency of allele A will be 0.714
- Allele a is recessive and detrimental
- After the first round of selection, the frequency of aa individuals will be 0.0331.

3. Which of the following is false?

- Variants with alleles with frequencies higher than 1% are considered polymorphic
- SNPs, indels and inversions are mostly variants with two alleles.
- Variants located outside of coding regions cannot affect phenotype
- Copy number variants (CNVs) can have multiple alleles with different number of copies of a gene.

4. Indicate the true sentence. The effective size (N_e) of a population will be smaller than the census if:

- There is a different number of males and females in the population
- Bottlenecks have occurred in the population
- The number of individuals fluctuates in different generations
- All of the previous answers are correct

5. Indicate the true sentence. An allele with a frequency of 0.15 in a population of 20 individuals:

- Is more likely to be fixed than lost by genetic drift
- Has a probability of fixation of 15% (true (?))
- Will have the same allele frequency in the next generation
- None of the other answers are true

6. The fitness values for the genotypes AA, Aa, and aa in a population are 0.8, 1, and 0.4, respectively.

Indicate the false sentence:

$$\Delta p = 0$$



$$\hat{p} = \frac{s_2}{s_1 + s_2}$$

- This population is evolving by balancing selection favoring the heterozygote
- At equilibrium the population will have $p = 0.75$ and $q = 0.25$
- At equilibrium the frequency of heterozygotes will be 1
- At equilibrium the average fitness of the population will be 0.85

7. Indicate which of the following statements about mutation is false:

- Mutations occur randomly
- In a mutation-selection equilibrium the frequency in the population of a deleterious allele generated by mutation with a constant rate, will be higher if the deleterious allele is dominant
- All variants found as polymorphisms in populations were generated by mutation as errors during DNA replication or repair in germ line cells.
- As an evolutionary force, mutation alone is a weak force and allele frequency changes will take a long time

8. In a small population of 8 individuals there are two neutral alleles, B and b. In a particular generation, the frequency of B is 0.8125. Indicate the false option:

- The probability that allele B is fixed in the next generation is 0.036
- The probability that allele B frequency is exactly the same in the next generation is 0.00044
- The probability that allele B is lost in the next generation is extremely low
- The probability that the population is still polymorphic in the next generation is high

Increase of fixation index with genetic drift

$$F_{t+1} = \frac{1}{2N} + \left(1 - \frac{1}{2N}\right) F_t$$



Fixation index in generation t in a finite population

$$F_t = 1 - \left(1 - \frac{1}{2N}\right)^t$$

9. There are two populations from the same species located very far away from each other on different continents. One of them is polymorphic for a gene with two alleles, A and a. In the other population, allele A is fixed. Which evolutionary mechanisms can provide an explanation for this situation?

- Genetic drift
- Natural selection acting on the population with the fixed allele
- Neither genetic drift or natural selection could result in this scenario
- Both genetic drift and local adaptation could explain this situation

10. Cystic fibrosis is an autosomal recessive disease that causes severe damage to the lungs, digestive system and other organs in the body, and is caused by alleles with an inactivated CFTR gene. This disease occurs in 1 in 3000 newborns of European origin. Indicate the false answer:

- We cannot test if the population is in Hardy-Weinberg equilibrium
- The allele frequency of the cystic fibrosis allele in European populations is 0.00033
- The probability that two healthy individuals have an affected child is 0.00032
- The frequency of carrier individuals in European populations is 0.0358