

Genetic Variation Study Guide

Part A: Vocabulary. Define the following terms.

1. genotype	genetic makeup
2. allele	alternative forms of a gene
3. heterozygous	having two different alleles
4. homozygous	having two identical alleles
5. dominant	allele that masks the effect of a recessive allele
6. recessive	allele whose effect is masked by a dominant allele
7. gene pool	total collection of genes in a population
8. gene flow	movement of genes between populations
9. genetic drift	random change in allele frequencies
10. founder effect	genetic drift that occurs when a new population is established by a small number of individuals
11. bottleneck	sharp reduction in population size leading to genetic drift
12. mutation	change in the DNA sequence
13. point mutation	change in a single nucleotide
14. insertion	addition of nucleotides to a DNA sequence
15. deletion	removal of nucleotides from a DNA sequence
16. substitution	replacement of one nucleotide with another
17. frameshift	insertion or deletion of nucleotides that shifts the reading frame
18. silent mutation	mutation that does not change the amino acid sequence
19. missense mutation	mutation that changes one amino acid
20. nonsense mutation	mutation that creates a premature stop codon
21. neutral mutation	mutation that has no effect on the organism's fitness
22. beneficial mutation	mutation that increases the organism's fitness
23. harmful mutation	mutation that decreases the organism's fitness

Part B: Genetic Risk Factor: Huntington's Disease

Scenario

1. identify the location of the gene:
chromosome 4p16.3
2. identify the function of the gene:
huntingtin: protein that helps in neuronal development and regulation
huntingtin: protein that helps in neuronal development and regulation
huntingtin: protein that helps in neuronal development and regulation
3. identify the risk factor:
CAG repeat expansion

Mcdougal Genetic Variation Study Guide Answers

Joacim Rocklöv



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