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the test:

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Biology 101

Assessment #1

4/28/21

Honor Code

I, ___Full Name___, affirm that I will only utilize the internet during this assessment for the purpose of accessing my class notes and documents linked on the class Canvas site. I will close all other internet browser windows before beginning the assessment. I will not use any other resources, including using search engines to look up terms. I will not discuss the assessment with anyone but Jehnna, including after it's completed. If I am confused about wording or terminology used on the assessment, I will reference the allowed materials and/or ask Jehnna to clarify by sending a private zoom message to her. I agree that I will learn best by authentically engaging with the assessment rather than searching for answers on the internet or from my friends. I understand that I will be offered a reassessment opportunity if I need it. I affirm that I, ___Full Name___, have read this honor code and will abide by it.

Instructions

This in-class written assessment has been designed to take approximately 45 minutes to complete. This is a rough estimation of an average completion time; individual experiences may vary. As such, anyone who needs it can spend more than 45 minutes on the assessment, but must turn in their final product before the end of class today. When you are finished with your assessment, please save it as a PDF and turn it in on Canvas.

Evaluation

I believe that assessments can be a learning opportunity. In reviewing for this assessment, you have helped to sharpen and solidify your conceptual understanding, and during the act of answering the following questions, you'll be recalling and applying that understanding in new ways. This application, doing something with what you've learned, is key! Just like you learn from projects, you can learn from assessments as well. I want everyone to understand what we're learning, so I will offer a reassessment opportunity to anyone who needs it (these may be oral one-on-one format or written format). This assessment provides opportunities to demonstrate content understanding from the following two template objectives:

Standard

Objective 1:

Objective 2:

Cell division, Genetics, and Evolution (n=2)

Cell division:

Students can explain and connect concepts within mitosis and meiosis, including cell cycle stage identification, mechanisms of chromosomal replication and segregation, and outcomes of genetic homogeneity vs. diversity.

Genetics:

Students can explain how alleles contribute to organismal phenotypes through impacts on protein function, and can reason through Medelian and non-Mendelian genetic inheritance patterns.

1. Phenylketonuria (PKU) is a genetic disorder with varying levels of severity in affected individuals; it's caused by mutations in the PAH enzyme, which normally breaks down the amino acid phenylalanine. If phenylalanine breakdown is not properly regulated, toxic levels of this amino acid can build up and damage cells, especially brain cells, leading to intellectual disability and other symptoms. Luckily, many PKU symptoms can be controlled/prevented via early dietary changes if the disorder is noticed in time.

PKU is a recessive disorder, meaning that it only occurs when an individual has two mutant PAH alleles. A single mutant PAH allele would not cause a PKU disease phenotype. The PAH gene is located on chromosome 12. Use the following genetic notation within your answers: capital P = normal PAH allele; lowercase p = PKU PAH allele.

1. A woman with confirmed PKU disorder and her unaffected male partner are considering having a child. The man has undergone genetic testing and has two normal PAH alleles. What is the likelihood that their child would have PKU disorder? Explain your reasoning using words in addition to the above genetic notation.
2. If the child from part (a) grew up and met a partner who had confirmed PKU disorder, what would be the likelihood that their child would have PKU disorder? Explain your reasoning using words in addition to the above genetic notation.

Individual cases of PKU can be caused by different PAH loss-of-function mutations - there is more than one possible genotype for this disease. "Classic PKU" is the severe form of the disease, which causes irreversible brain damage in infancy if left untreated. In contrast, "mild PKU" doesn't require as much dietary intervention and would not result in severe symptoms if left untreated. This spectrum of severity exists because the various PAH gene mutations have different impacts on PAH enzyme function. Classic PKU is associated with a near-complete loss of PAH function. For each of the following types of PAH gene mutation, predict whether classic or mild PKU is more likely and briefly explain your reasoning:

3. A promoter mutation that reduces expression of PAH protein to 50% of normal levels.
4. A missense mutation that changes an amino acid in the PAH enzyme's active site, preventing any phenylalanine from binding there.
5. A frameshift mutation very early in the coding sequence of the PAH gene.
6. A missense mutation that changes an amino acid in an allosteric site (an enzyme site that is not directly involved in breaking down phenylalanine), leading to a 40% reduction in the rate of enzyme activity.
7. Hemophilia is an inherited sex-linked disorder characterized by the inability to properly form blood clots. The gene associated with hemophilia is located on the X chromosome. Hemophilia is now treatable, but before the mid-1900s, only a few hemophiliacs survived to reproductive age because even minor injuries could prove fatal through loss of blood.
8. Hemophilia affects males much more frequently than females. Propose an explanation for the increased frequency of hemophilia in males.
9. A healthy woman whose father had hemophilia wants to start a family of her own. She is concerned that her potential biological children could have hemophilia like her father did. Based on this information, around 50% of the woman's eggs should contain the hemophilia-associated allele. In contrast, 100% of the woman's somatic cells should contain the hemophilia-associated allele. Explain how the process of meiosis created this discrepancy between egg cells and somatic cells.
10. If the woman's male partner is healthy, what are the chances that their future son would have the same hemophilia as his grandfather? Briefly explain your reasoning.

11. Familial cancer syndromes are inherited genetic disorders that vastly increase the risk of developing particular cancer types as compared to the overall population (cancer is an uncontrolled overgrowth of cells) . One of these familial cancer syndromes, Li-Fraumeni Syndrome, is caused by inheriting a particular type of mutation in the p53 gene. P53 is an important cell cycle regulator; it will halt cell cycle progression until any DNA damage is cleanly repaired or initiate cell death if the damage is too extensive to be fully repaired.
12. What type of impact do Li-Fraumeni mutations likely have on p53 function? Explain your reasoning by connecting your knowledge of mutational impacts and the cell cycle.

Another familial cancer syndrome, Multiple Endocrine Neoplasia Syndrome Type 2 (MEN2), is caused by inheriting a particular type of mutation in the RET gene. The RET protein is a receptor that receives growth factor signals at the surface of some cells. These signals are passed on by RET so that cell division can be pushed forward.

1. What type of impact do MEN2 mutations likely have on RET function? Explain your reasoning by connecting your knowledge of mutational impacts and the cell cycle.

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