#ref #ret

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1 | **begin!**

Honor Code

I, Huxley Marvit, 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, Huxley Marvit, have read this honor code and will abide by it.

1.1 | **1**

1. A mom: pp, dad: PP pP -> P In any combination, their would not be two mutant PAH alleles. Thus, the child would have a 0% chance of having the disorder.
2. B child: pP, partner: pp

   pp, (p1p1) -> p pp, (p1p2) -> p Pp, (Pp1) -> P Pp, (Pp2) -> P

   50% chance of having PKU disorder

   (a) **A promoter mutation that reduces expression of PAH protein to 50% of normal levels.**
      i. Given that "classic PKU" results from near complete loss of PAH function, a 50% loss would most likely be classified as "mild PKU"

   (b) **A missense mutation that changes an amino acid in the PAH enzyme's active site, preventing any phenylalanine from binding there.**
      i. This would completely inhibit PAH function, leading to "classic PKU"

   (c) **A frameshift mutation very early in the coding sequence of the PAH gene.**
      i. A frameshift mutation early on would cause almost the entire sequence to be translated incorrectly. Most likely, this would lead to near complete loss of function, and thus, "classic PKU"

   (d) *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.*
      i. 40% reduction is not near-complete, and would most likely be classified as "mild PKU"

1.2 | **2**

1. A Most woman have two X chromosomes, whereas most men have a X chromosome and a Y chromosome. Since hemophilia is located on the X chromosome, in men, it doesn't have a chance to be dominated.

2. B For the woman to be healthy and have any hemophilia-associated alleles, they must have a singular recessive mutation.

   Somatic cells, carrying 23 pairs of chromosomes, have all the genetic information which is all copied by mitosis. Thus, they will contain the hemophilia-associated allele.

   Meiosis produces haploid cells with only 23 singular 2-chromatid chromosomes. Thus, only half would have the mutant x chromosome.

3. C woman: xX, man: XY

   For their child to be male, the man has to pass down his Y, leaving options:

   xY, XY.

   Thus, their is a 50% chance of hemophilia.

1.3 | **3**

1. A Given that p53 is a negative regulator, it most likely has a loss of function mutation, making it less effective at doing its job of pausing cell cycle progression when needed and initiating cell death. When p53 is less effective, an uncontrolled overgrowth of cells is more likely to occur.

2. B Given that RET is a positive regulator, MEN2 mutations would most likely cause a gain of function. The RET protein would be more prone to signaling for cell progression, pushing cell division forward before everything is necessarily ready. This would increase the likelihood of cancer.