#flo #ref #ret #disorganized #incomplete

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1 | here we go

the test:

**

Biology 101

Assessment #1

4/28/21

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. PKU

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.

B child: pP, partner: pp

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

50% chance of having PKU disorder

1.2 | 2

1. **A promoter mutation that reduces expression of PAH protein to 50% of normal levels.**
    1. Given that "classic PKU" results from near complete loss of PAH function, a 50% loss would most likely be classified as "mild PKU"
2. **A missense mutation that changes an amino acid in the PAH enzyme's active site, preventing any phenylalanine from binding there.**
    1. This would completely inhibit PAH function, leading to "classic PKU"
3. **A frameshift mutation very early in the coding sequence of the PAH gene.**
    1. 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"

4. **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.**