# Year 12 Science: Mutations, metabolic pathways and the environment

Name: _____ Date: _____

Try these without your notes first, then check the answer key.

## Practice

1. The mRNA codon UAC changes to UAA. Name the type of mutation.

2. The mRNA codon GGU changes to GGC. What happens to the amino acid?

3. Which shifts the reading frame: deleting 1 base or deleting 3 bases?

4. In the pathway A → B → C, enzyme 2 (B to C) does not work. Which substance builds up, and which is missing?

5. Name two mutagens.

6. What environmental factor makes a Siamese cat's ears dark?

## Exam-style question: melanin and mutations

**Gene 2 codes for tyrosinase, the enzyme that turns tyrosine into melanin. A short, made-up section of its template strand, 3' to 5', reads TAC AAG CTT GTA ACC. Mutation 1 reads TAC AAG CTC GTA ACC, and mutation 2 reads TAC AAG CTT GTA ACT.**

7. Give the amino acid sequence for the normal gene and for each mutation.

8. Name the type of each mutation.

9. Explain which mutation is more likely to cause albinism.

10. Discuss how a mutation in gene 1 would show differently from one in gene 2, and how the environment can change melanin production without changing the genes.

## Fix the mistake

11. Correct this statement: "A substitution always changes the protein."

12. Correct this statement: "A deletion removes one amino acid, so it is mild."

13. Correct this statement: "The Siamese cat's cold ears changed its genes for dark fur."

14. Correct this statement: "If enzyme 2 is blocked, the substrate of enzyme 1 builds up."

## Answer key (for parents and tutors)

1. The mRNA codon UAC changes to UAA. Name the type of mutation.
   **Nonsense: UAC codes for Tyr, but UAA is a stop codon, so the polypeptide ends early.**
2. The mRNA codon GGU changes to GGC. What happens to the amino acid?
   **Nothing: GGU and GGC both code for glycine, a silent mutation.**
3. Which shifts the reading frame: deleting 1 base or deleting 3 bases?
   **Deleting 1 base; deleting 3 removes one codon without a frameshift.**
4. In the pathway A → B → C, enzyme 2 (B to C) does not work. Which substance builds up, and which is missing?
   **B builds up and C is missing.**
5. Name two mutagens.
   **Any two of: UV light, X-rays, gamma rays, chemicals in tobacco smoke, some viruses.**
6. What environmental factor makes a Siamese cat's ears dark?
   **The lower temperature of the ears lets the temperature-sensitive tyrosinase work there and make melanin.**
7. Give the amino acid sequence for the normal gene and for each mutation.
   **Normal mRNA AUG UUC GAA CAU UGG: Met-Phe-Glu-His-Trp. Mutation 1 mRNA AUG UUC GAG CAU UGG: Met-Phe-Glu-His-Trp. Mutation 2 mRNA AUG UUC GAA CAU UGA: Met-Phe-Glu-His, then stop (Achieved for a correct sequence).**
8. Name the type of each mutation.
   **Both are substitutions; mutation 1 is silent and mutation 2 is nonsense (Achieved).**
9. Explain which mutation is more likely to cause albinism.
   **Mutation 2, because the stop codon ends translation early, so the tyrosinase is too short to fold into its working shape and no melanin is made; mutation 1 still gives Glu (GAA and GAG both code for it) so the enzyme is unchanged (Merit). A person needs two faulty copies of the gene to have albinism.**
10. Discuss how a mutation in gene 1 would show differently from one in gene 2, and how the environment can change melanin production without changing the genes.
    **Excellence: a block at gene 1 stops phenylalanine becoming tyrosine, so phenylalanine builds up (PKU), but melanin can still be made from tyrosine in food; a block at gene 2 leaves tyrosine unused, so there is no melanin and the phenotype is albinism. The environment changes the phenotype without changing the genotype: a temperature-sensitive tyrosinase makes melanin only in a Siamese cat's cool ears, paws and tail, and a low-phenylalanine diet prevents the brain damage of PKU.**
11. Correct this statement: "A substitution always changes the protein."
    **Because the code is degenerate, a substitution can give a codon for the same amino acid: a silent mutation.**
12. Correct this statement: "A deletion removes one amino acid, so it is mild."
    **Deleting one base shifts the reading frame, so every codon after it changes; only deleting three bases removes a single amino acid.**
13. Correct this statement: "The Siamese cat's cold ears changed its genes for dark fur."
    **The genotype is the same everywhere; the cool temperature only lets the existing enzyme work there.**
14. Correct this statement: "If enzyme 2 is blocked, the substrate of enzyme 1 builds up."
    **The substance just before the block builds up: the substrate of enzyme 2, which is the product of enzyme 1.**