When DNA Changes

Trace a mutation from DNA to a trait

FB.4.4 "Obtain and communicate information to describe how mutations may affect genetic expression and provide examples."

NAME _____ DATE _____

1 READ AND MARK

Read it twice

YOUR JOB Underline each DNA change in the passage, then circle its effect on gene activity or a protein. Number each underlined change and its matching circled effect so you can trace what happened.

Three Changes, Three Effects

1 A mutation is a change in DNA, but its effects depend on where the change occurs. Gene expression involves copying a gene into RNA and using that message to build a protein. Some mutations change a protein's instructions; others change when or how much of a protein is made. In airway cells, the CFTR gene provides instructions for a protein that helps move chloride ions across cell membranes. One common CFTR mutation removes three DNA letters. This deletion removes one amino acid, causing the protein to fold incorrectly. Much less CFTR reaches the cell surface, contributing to thick mucus in cystic fibrosis.

2 A different change can leave the amount of protein largely unchanged but alter what it does. In the HBB gene, a single DNA base substitution can replace one amino acid in hemoglobin, the protein that carries oxygen in red blood cells. A person with two copies of this sickle-cell variant makes hemoglobin that can clump when oxygen is low. Red blood cells may then become stiff and sickle shaped, blocking small blood vessels. The mutation changes the protein's structure, not simply whether the gene is switched on. This example shows why knowing a mutation's location and effect matters more than calling every mutation harmful.

3 Other mutations affect a gene's activity rather than the protein's amino acid sequence. The LCT gene provides instructions for lactase, an enzyme that digests milk sugar. In many people, LCT activity decreases after childhood. Certain DNA changes in a control region near LCT help keep the gene active into adulthood. Adults with these changes can keep producing lactase and often digest milk sugar more easily. The control region is not the recipe for lactase itself; it helps regulate when the recipe is used. Together, these cases show that a DNA change can affect protein structure, protein delivery, or the amount of protein produced.

2 ANSWER WITH EVIDENCE

Use the passage

1. Trace the CFTR mutation from the deleted DNA letters to its effect on mucus.

2. How does the HBB mutation change hemoglobin, and what can happen to red blood cells?

3. Where are the LCT-related DNA changes, and how can they affect an adult's ability to digest milk sugar?

4. Compare the HBB and LCT examples. Which changes a protein's structure, and which changes how long a gene stays active?

3 YOUR TURN

Your own words

Invent an organism with a gene for a visible trait. Describe a hypothetical mutation in that gene or its control region, trace its effect from DNA to the trait, and state whether it changes a protein's structure or the amount made.

When DNA Changes

PART 1 · READ AND MARK

Underline each DNA change in the passage, then circle its effect on gene activity or a protein. Number each underlined change and its matching circled effect so you can trace what happened. Accept any marking that follows this job and points at the right text.

PART 2 · ANSWERS

#	QUESTION	ANSWER
1	Trace the CFTR mutation from the deleted DNA letters to its effect on mucus.	**The deletion removes one amino acid, so CFTR folds incorrectly. Much less CFTR reaches the cell surface, contributing to thick mucus.**
2	How does the HBB mutation change hemoglobin, and what can happen to red blood cells?	**A base substitution replaces one amino acid in hemoglobin. The altered hemoglobin can clump when oxygen is low, making red blood cells stiff and sickle shaped.**
3	Where are the LCT-related DNA changes, and how can they affect an adult's ability to digest milk sugar?	**The changes are in a control region near LCT. They can keep LCT active into adulthood, so an adult continues making lactase and may digest milk sugar more easily.**
4	Compare the HBB and LCT examples. Which changes a protein's structure, and which changes how long a gene stays active?	**The HBB substitution changes hemoglobin's amino acid sequence and structure. The control-region changes near LCT help keep the gene active into adulthood.**

PART 3 · YOUR TURN (SAMPLE ANSWER)

In an invented moth, the GLO gene makes an enzyme that produces blue wing pigment. A deletion in a control region lowers GLO activity in wing cells, so they make less enzyme. With less pigment produced, the wings appear pale blue. This mutation changes the amount of enzyme made, not its structure.

Accept other biologically plausible chains from a DNA change to gene activity or protein function and then to a visible trait. For the CFTR question, students may describe less protein at the cell surface without naming protein folding.