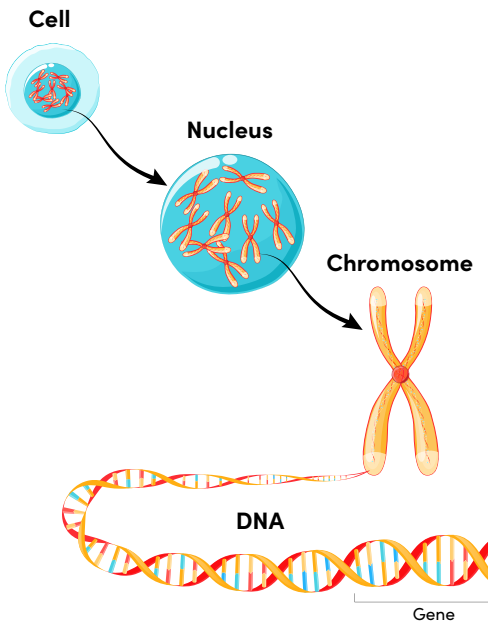
Genetic Mutations

DNA is the genetic code inside the nucleus of a cell. It contains instructions for making proteins. DNA is organized into structures called **chromosomes**.

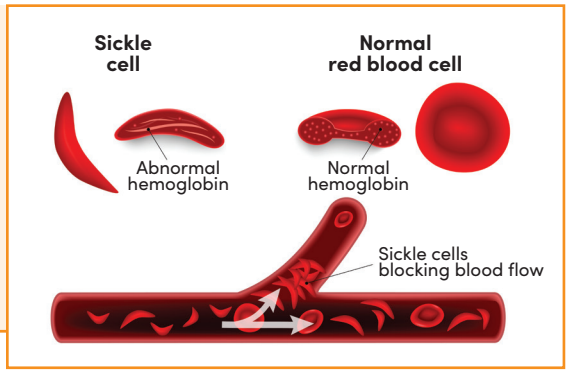
Genes are sections of DNA that code for specific proteins. The structure of a protein determines its function, and the function of proteins results in observable traits in organisms. So, an organism's genes determine many of its observable traits.

Sometimes, changes occur to genes. These are called **mutations**. Mutations can result in observable changes to traits. These changes can be beneficial, harmful, or neutral for organisms.



Let's check out an example! Read the passage below and answer the questions that follow.

1. Hemoglobin is a protein found in red blood cells. It is responsible for transporting oxygen throughout the body. Sometimes, a mutation occurs in the hemoglobin gene that changes the structure of the hemoglobin protein. This mutation results in sickle cell anemia, a disorder that changes the shape of red blood cells and can lead to health problems.



a. A DNA sequence is often modeled as a series of letters. The letters represent a specific genetic code. Below is the genetic code for hemoglobin. Circle the place in the mutated gene where you see a change, or mutation, from the normal gene.

b. Is this mutation beneficial, harmful, or neutral for humans? Why do you think so?

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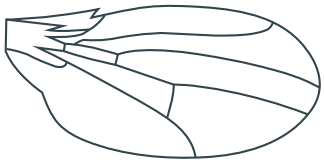
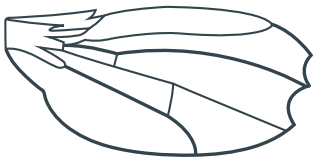
Keep going! Read the passages below and answer the questions that follow.

2. Most flies have rounded wings. Sometimes, a mutation occurs that deletes a small region of a specific chromosome. This mutation causes the fly to have notched wings. In most cases, flies with notched wings are still able to fly, and their ability to survive and reproduce is not affected by the mutation.



Normal Wing



Notched Wing

a. Circle the place on the mutated chromosome where a change, or mutation, occurred.

Normal chromosome **Mutated chromosome**

b. Is this mutation beneficial, harmful, or neutral for flies? Why do you think so?

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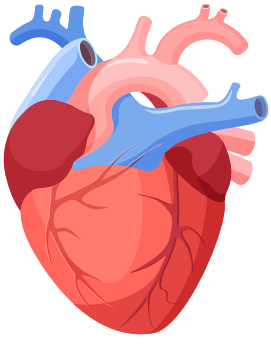
3. Apolipoprotein A1, or apoA-1 for short, is a protein found in humans. Sometimes, a mutation occurs in the apoA-1 gene that changes the protein's structure. People with this mutation are protected from cardiovascular disease, even if they have poor cholesterol. Scientists are studying this mutation to develop new therapies to treat cardiovascular disease.



a. Circle the place on the mutated protein where a change, or mutation, occurred.

Normal protein

Mutated protein

b. Is this mutation beneficial, harmful, or neutral for humans? Why do you think so?

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