REUSABLE CLASS SET | DO NOT WRITE ON

How mutations affect proteins

Science question: How can a tiny change in a gene affect an entire body? Read both pages, then complete your response handout on your own. You need only these sheets and a pencil.

A change in the instructions

A cell is building a protein, adding amino acids one after another. The instructions come from a **gene**, a section of DNA. DNA is packaged into **chromosomes**. A gene does not build a trait directly: it provides instructions for a protein, and the protein helps the body do a job.

Now imagine that a piece of the DNA changes. This change is a **mutation**. A substitution replaces DNA letters, an insertion adds letters, and a deletion removes letters. Will the protein stop working? That depends on what changes and what job the protein does.

Read a short stretch of DNA

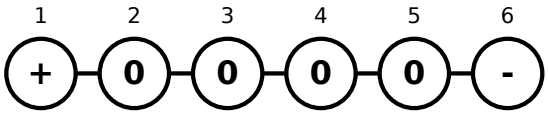
For this investigation, use the coding strand of a made-up gene. Each three-letter group specifies one amino acid. Cells use an RNA copy between DNA and protein; this shortcut table lets you go directly from DNA to amino acids. This is a short protein segment, not a complete gene.

DNA group	Amino acid	Charge in this model
AAA or AAG	Lysine	+
GAA or GAG	Glutamate	-
GGT	Glycine	0 (no charge)

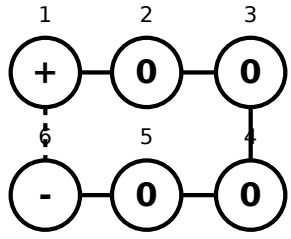
Original DNA: AAA | GGT | GGT | GGT | GGT | GAA
Amino acid charges: + | 0 | 0 | 0 | 0 | -

A chain that can bend

The circles below are amino acids. Solid lines connect neighbors in the chain. Opposite charges can attract, bringing distant parts closer together. The dashed line marks an attraction; it is not another link in the chain. Same charges repel. The 0 circles have no charge in this model.



Straight chain



One possible bend

Worked example: Replace the first AAA with AAG. The DNA changes, but both groups specify lysine (+). The amino acid sequence stays the same, so the original drawing still fits.

Investigate and explain

Three changes to try

Each case starts from the original DNA on page 1. Use the lookup table to find the new chain. Then draw one possible shape. Keep every amino acid in order, number the circles, and keep solid links between neighbors. Add a dashed line if opposite charges could attract.

Case	Changed DNA segment
A	AAA | GGT | GGT | GGT | GGT | AAA
B	AAA | GGT | GGT | AAA | GGT | GGT | GAA
C	AAA | GGT | GGT | GGT | GGT

Here, an insertion or deletion adds or removes a whole three-letter group. Adding or removing just one DNA letter can shift how the later groups are read and change many later amino acids.

Model limits: Real proteins fold in three dimensions and have many interactions besides charge. Your 2D drawing shows a possible mechanism. It does not prove the exact shape or whether the protein works.

When the protein helps hold a body together

Picture the lens inside an eye, held in position by tiny supporting fibers. Picture a large blood vessel stretching with each heartbeat. Both depend on **connective tissue**, tissue that connects and supports body parts. It needs strength and flexibility.

The **FBN1 gene** provides instructions for **fibrillin-1**. This protein helps form tiny fibers called microfibrils in connective tissue. Some FBN1 mutations change fibrillin or reduce the amount of working fibrillin available. The resulting fibers may not provide their usual support. Microfibrils also help regulate tissue growth, so their effects go beyond holding parts in place.

Evidence about Marfan syndrome	Body structure and function
FBN1 mutations can reduce usable fibrillin-1 and disrupt microfibrils.	Connective tissues can lose normal support and stability.
Fibers help hold the eye lens in position. In Marfan syndrome, the lens can shift.	A shifted lens can interfere with focusing light.
The aorta carries blood from the heart. In Marfan syndrome, its wall can weaken and stretch.	The vessel may enlarge and become more vulnerable to damage.

The short charge model on page 1 is not fibrillin and does not show an actual Marfan mutation.

The outcome depends on the change

Example 1: Changed DNA specifies the same amino acids; the protein and the organism keep working as before.
Example 2: A mutation reduces usable fibrillin and disrupts tissue support.
Example 3: In a hypothetical bacterium, a changed protein breaks down the food available in its environment more effectively. The bacterium survives and reproduces better.

Source for Marfan evidence: NIH MedlinePlus Genetics, Marfan syndrome, medlineplus.gov/genetics/condition/marfan-syndrome/.