



The Search for a Mutated Gene

Time 0:00 Introduction

You will watch a short video that presents the approaches scientists used to identify a mutation that causes retinitis pigmentosa (RP) in a patient. At various points, the video will pause and you will be asked to think about the research. You will not be able to continue to watch the video, until you have answered and saved your response to the prompt.

As you answer the prompts, keep in mind that some questions do not have a "right answer." You will have the opportunity to revisit your responses at the end of the video.

Time 0:10 Embedded Prompt

In a few words, describe what you know about gene therapy.

Time 1:35 Embedded Question

Sam's parents don't have retinitis pigmentosa (RP). How can RP be an inherited condition?

Extension Questions:

- How do our brains convert information from light entering our eyes into an image?
- What is disease? How do organisms get diseases?
- What is a genetic mutation and what are its effects?
- How is searching for a treatment for a genetic disease different from that for a disease caused by something else, such as bacteria or viruses?

Time 2:56 Embedded Question

You collect blood samples, which contain DNA, from a patient with RP and their relatives. Some of the relatives have RP and some do not. Outline a strategy for using these samples to identify the disease-causing mutation in the patient.

Time 3:22 Embedded Question

How can mutations in different genes be associated with a single disease?

Extension Questions:

- Why would a researcher use a pedigree to study a disease?
- Many characteristics of an organism are controlled by more than one gene. Likewise, many of the biochemical processes and pathways involve proteins produced by different genes. Explain how this relates to the existence of many different mutations that cause RP.
- Can you think of any other diseases or conditions that are caused by mutations in different genes?

Time 4:02 Embedded Question

A scan of Sam's genome for the 100 most common mutations known to cause RP didn't find anything. How can you explain this result?

Time 5:16 Embedded Question

Sam has a mutation in a gene that affects the function of a transfer RNA (tRNA). Human cells have 20 different types of tRNAs, and each adds a different amino acid to growing peptide chains, which form proteins.

Based on what you know so far, would you expect this mutation to affect **all** proteins produced in Sam's cells? Explain your reasoning.

Time 6:30 Embedded Question

What evidence could you collect to confirm that the mutation identified in Sam's DNA causes symptoms of RP?

Extension Question:

- Some mutations are lethal and others are not. Explain the difference between these two types of mutations in terms of cellular function.

Time 7:56 Embedded Question

Doctors may be able to inject a functioning copy of the gene mutated in Sam's DNA in the cells of his eyes. If the procedure were successful, would you expect Sam to regain his vision? Why or why not?

Extension Question:

- In your own words, explain why researchers use model organisms such as zebrafish. Can you name any other model organisms?

Time 8:30 Embedded Question

Other than identifying a target for gene therapy, how does identifying a disease-causing mutation help a patient and their family?

Extension Questions:

- Genetic medicine is an emerging technology that holds great promise for many diseases. Are there any ethical concerns with genetic medicine as described in this case study? Explain your perspective.
- Are there any other diseases you know of that currently are or could potentially be treated or cured through genetic medicine?
- Search newspapers, magazines, or the scientific literature for diseases being treated through genetic medicine. Have there been successes? Failures?