



## *The Search for a Mutated Gene*

### INTRODUCTION

You will watch a short video that explores how scientists identified a mutation that causes **retinitis pigmentosa (RP)**, a disease that leads to blindness. You'll also learn how this information can inform treatments for RP and other genetic diseases.

### PROCEDURE

Play the interactive video [The Search for a Mutated Gene](#). At various points, the video will pause and ask you to think about the content. You will not be able to continue watching the video until you have answered and saved your response to the prompt. You can record your answers in this worksheet or as directed by your instructor.

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.

1. In a few words, describe what you know about gene therapy.
2. Sam's parents don't have RP. How can it be an inherited condition?
3. 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.
4. How can mutations in different genes be associated with a single disease?
5. 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?
6. 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.

7. What evidence could you collect to confirm that the mutation identified in Sam's DNA causes symptoms of RP?
  
  
  
  
  
  
  
  
  
  
8. Doctors may be able to inject a functioning copy of the gene mutated in Sam's DNA into the cells of his eyes. If the procedure were successful, would you expect Sam to regain his vision? Why or why not?
  
  
  
  
  
  
  
  
  
  
9. Other than identifying a target for gene therapy, how does identifying a disease-causing mutation help a patient and their family?