TEACHING THE GENOME GENERATION

GENOME GENERATION

Sequencing for Rare Disease Diagnosis:
Scenario Introduction

The Jackson Laboratory
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Scenario

You are a parent of two daughters, both of whom have a rare, undiagnosed disorder. Your daughters have similar symptoms, including learning difficulties and ataxia, an inability to control body movements.

You want to find a medical diagnosis for your daughters so that you can connect with other patients and their families and learn more about how to best support your daughters. However, you've been to many doctors and none of them have been able to identify a known disorder that matches all of your daughters' symptoms.

Finally, a doctor connects your family to a group of researchers who study human disease. After hearing about your family's efforts to find a diagnosis and care for your daughters, the researchers suggest that they might be able to learn more by sequencing your daughters' DNA. After sequencing, they'll compare your daughters' DNA sequences to a reference sequence to look for any differences, or variants.

Reflection

1. How are these patients and their parents' experiences similar to or different from your own experience at the doctor?

2. What questions would you have for the researchers as the parent? As one of the daughters?

Guiding Questions

During these activities, you will be learning more about what the researchers did in their study and what they discovered. As you go, you'll be learning more about the questions below. Before you begin, write out your thoughts in response to these questions. Don't worry if you don't feel confident in your answers! You can revisit these after each activity so that you can see what you've learned.

Given what you know about genome sequencing and genetic variation:

1. What can we learn from comparing genetic information across individuals and species?

Given what you know about (a) how DNA codes for proteins and (b) the connection between protein structure and function:

2. How might a DNA variant affect protein sequence, structure, or function?