FUTU?ELAB+

LIVING EARTH

Genetic Detectives: Investigating Inherited Diseases

Genetic Mutations

Developed in partnership with: Discovery Education

In this Lesson Plan:

Print the Teacher Section \rightarrow rightarrow

01	For Teachers	
Lesso	n Overview	1
Make	Connections!	2
Pedag	ogical Framing	3
Slide	5	
	Day 1	4-5
	Day 2	6
	Day 3	7-8
	Day 4	9
	Day 5	10
	Day 6	11-12
National Standards 13		
Educa	tor Resources	
	Protein Synthesis	14-30

Print the Student Section \rightarrow

02	Student Resources	
DNA,	Genes, Proteins, and Mutations Review	
	Part 1 Jigsaw	1
	Part 2 Parts of a Cell Review	2
Muta	ion Investigation	
	Part 1 Intro to Mutations	3
	Part 2 Outcomes of Mutations	4
	Part 3 Types of Mutations	5
	Part 4 Patient's Inherited Disease	6
Trans	cription Investigation	
	Part 1 My Transcription Observations	7-8
	Part 2 My Transcription Questions	9
	Part 3 My Transcription Research	10
Trans	lation Investigation	
	Part 1 My Translation Observations	11-12
	Part 2 My Translation Questions	13
	Part 3 My Translation Research	14
Put It	All Together	15
Inheri	ted Disease Mutation Narrative	16-17



Cover Image Genetic research, pipetting into a test tube in front of a DNA autoradiogram This document is separated into two sections, For Teachers [T] and Student Resources [S], which can be printed independently.

Select the appropriate printer icon above to print either section in its entirety.

Follow the tips below in the Range field of your Print panel to print single pages or page ranges:

Single Pages (use a comma): T3, T6

Page Range (use a hyphen): T3-T6

LIVING EARTH / INVESTIGATING INHERITED DISEASES

Genetic Mutations

DRIVING QUESTION

How do you counsel a patient on their risk of genetic disease while considering their health literacy?

OVERVIEW

In this lesson, students start to develop a **Genetic Test Report** for the patient they selected in Lesson 2. They begin by learning about genetic tests, which are tools that allow patients to identify traits that have been passed from generation to generation. Students examine how patients can use the information in their genetic test to make important decisions about their health and family planning. As they develop their **Genetic Test Report**, students explore the profession of genetic counseling. Through this project, they simulate the role of genetic counselors to communicate test results in a manner that is empathetic, culturally sensitive, aware, and understanding of a patient's health literacy.

Students are also tasked with developing a narrative for their **Genetic Test Report** that provides the patient with background information on how genetic diseases arise. In order to do this, students learn about how genetic mutations lead to altered protein synthesis and review examples of genetic disease. Students' narratives must explain these complicated concepts in language that a patient with low health literacy would be able to understand.

ACTIVITY DURATION

Six days

ESSENTIAL QUESTIONS

What is the genetic code?

How is the information in DNA used to make proteins?

How do mutations give rise to altered proteins?

How do the altered proteins developed from mutations in DNA give rise to specific inherited diseases?

How do you counsel a patient with low health literacy on their risk of genetic disease?

OBJECTIVES

Students will be able to:

Discover how mutations give rise to altered proteins.

Investigate the genetic code.

Explore how information in DNA is used to create proteins through the process of protein synthesis.

Examine how genetic counselors communicate complicated information effectively to a patient.

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Day 1	Day 2	Day 3	Day 4	Day 5	Day 6
Review what they know about DNA and organelles.	Learn about mutations. Identify and research the mutation(s) associated with their patient's inherited disease.	Examine protein synthesis by investigating transcription.	Continue to examine protein synthesis by investigating translation.	Write a narrative that explains to their patient how the mutation associated with their inherited disease gives rise to faulty proteins. Conference with the teacher to demonstrate how to build proteins with physical models.	Identify ways to best communicate the mutation narrative to their patient.

MAKE CONNECTIONS!

How does this connect to the larger unit storyline?

This lesson examines different types of genetic disorders and highlights the genetic testing process. In this lesson, students identify the mutations that give rise to the inherited disease associated with their patient. They use this understanding to further develop their Genetic Test Report.

How does this connect to careers?

Genetic counselors help patients understand genetic diseases and explore treatment options. They also help people understand the risk of disease transmission to future generations.

Patient advocates help patients with genetic diseases coordinate care among doctors and identify all available treatment options.

Health service managers oversee the services provided by health care organizations. They help clinical teams overcome operational challenges and keep practitioners abreast of current laws and regulations.

Medical interpreters translate complex health information to patients that are non-native English speakers. Medical interpreters are often mediators between doctors and patients and help the patient gain a full understanding of the health information provided.

How does this connect to our world?

Students use their knowledge, understanding, and personal connection to their patient to consider how to effectively communicate the results from the genetic test.

It is important to stress that medical technology and personnel vary widely around the world, and not every community has access to high-quality care. Students can investigate equity and social justice issues related to access to healthcare, including access to genetic testing worldwide.



Pedagogical Framing

Instructional materials are designed to meet national education and industry standards to focus on in-demand skills needed across the full product development life cycle from molecule to medicine—which will also expose students and educators to the breadth of education and career pathways across biotechnology.

Through this collection, educators are equipped with strategies to engage students from diverse racial, ethnic, and cultural groups, providing them with quality, equitable, and liberating educational experiences that validate and affirm student identity.

Units are designed to be problembased and focus on workforce skill development to empower students with the knowledge and tools to be the change in reducing health disparities in communities.



SOCIAL-EMOTIONAL LEARNING

In this lesson, students research how medical professionals can deliver and communicate genetic testing results accurately in an empathetic, culturally sensitive, aware, and understanding manner that is considerate of patients' health literacy. They will practice social awareness and management skills by working collaboratively, resisting inappropriate social pressure, negotiating solutions to conflict, and seeking help when needed.

CULTURALLY AND LINGUISTICALLY RESPONSIVE INSTRUCTION

This lesson applies culturally and linguistically responsive instruction to student research into how medical professions can deliver and communicate genetic testing results. Students begin to get a clear picture of how genetic mutations arise, and will deepen their understanding of how heritable diseases disproportionately impact communities of color. The lesson offers opportunities for understanding of patients' health literacy challenges, while creating culturally and linguistically responsive, empathetic, and socially aware presentation options, treatment choices, and healthcare information reports.

ADVANCING INCLUSIVE RESEARCH

At present, the vast majority of genetic data available for clinical trials comes from northern Europe. The lack of diversity in these data means that scientists cannot get a clear picture of how a new therapy works across a global population. In this lesson, students explore how genetic mutations arise and deepen their understanding of how heritable diseases disproportionately impact communities of color. By the end of the unit, students will be able to identify the reasons why the current genetic data are so heavily skewed toward those with northern European ancestry, and articulate the importance of collecting data from historically marginalized communities in order to develop effective therapies.

COMPUTATIONAL THINKING PRACTICES

DNA is the algorithm that tells the body how to make proteins. When there is an error in executing that algorithm, genetic mutations result. In this lesson, students will practice the computational thinking strategy of developing algorithms to identify the steps of the transcription process and the computational thinking strategy of decomposition to drill down into genetic sequences and identify mutations.

CONNECTION TO THE PRODUCT LIFE CYCLE

In this lesson, students learn how mutations arise during DNA transcription. They then connect their knowledge of how genetic mutations develop to a selection of heritable diseases such as Sickle Cell and Cystic Fibrosis. As they collect data on these diseases, students gain hands-on experience with the Discover phase of the product life cycle. Students will understand how little is currently known about a wide range of genetic mutations and develop strategies on how to advocate for more research funding for rare heritable diseases.

COMPUTATIONAL THINKING IN ACTION

As students participate in the telephone exercise, they gain hands-on experience with the computational thinking strategy of developing algorithms. Algorithms are sequences of steps that computer programmers use to tell computers how to complete tasks. The game of telephone demonstrates to students how errors can happen in the transcription process (which is how cells enact the algorithm provided in DNA).



COMPUTATIONAL THINKING IN ACTION

Metaphors and similes are tools that help students gain experience with the computational thinking strategy of abstraction, which is the process of distilling information down into its most central components.

Slides 1-12

Slides 1-8

Play telephone as a way to introduce mutations. (10 minutes)

- 1 Have students form a circle or a line.
- 2 Ask students if they have ever played telephone before. Have students share how the game is played. If no one knows, share the rules with students:
 - **a.** "The rules are simple, I'm going to tell the first person something, and they have to repeat EXACTLY what I said to the person next to them, then that person repeats EXACTLY what they hear to the next person, and so on until the final person is reached. When the final person is reached, they share EXACTLY what they hear out loud for the class to hear."
- 3 Before you begin, ask students what they predict will happen.
- 4 Play two or three rounds of telephone with students.
- 5 When done, have students respond to the following prompts through a *Think-Pair-Share* discussion protocol:
 - **a.** "How do you think the telephone activity is connected to DNA and inherited diseases?
 - b. "What do you think will happen when changes occur in your DNA?"

Slides 9-12

Students review background information on cell parts that are connected to DNA, traits, and mutations. (35 minutes)

- 1 Have students do the first part of the *3-2-1 Bridge* thinking routine using the topic Mutations.
 - **a.** Students write down 3 thoughts or ideas, 2 questions, and come up with 1 metaphor or simile about mutations.
 - **b.** When done, have a few students share their examples.
 - **c.** Students will complete Part 2 of the thinking routine at the beginning of class on Day 2 when students have completed their *DNA*, *Genes*, *Proteins*, & *Mutations* capture sheet.

Continues next page >

Day 1 Continued



Slides 9-12

2	Inform students they are going to investigate how changes in DNA, aka mutations, give rise to the inherited disease on which they are focusing. The next step involves a review of what they already know about DNA, where it is located, and how information in DNA is used.
3	Have students split into four equal-size groups. Have one group watch the <i>DNA</i> & <i>Genes video</i> , one group watch the <i>What are Proteins video</i> , another watch the <i>Inheritance of Traits video</i> , and the fourth group watch the <i>Outcomes of Mutation</i> .
4	Students in the four groups should use an <i>AEIOU</i> chart to summarize and clarify the information from their video, so they can teach other students in the next step.

Teacher Note > *Distribute* <u>*AEIOU*</u> graphic organizers, or have students create their own on a blank piece of paper. Explain the meaning of each vowel for the purposes of this strategy.

- A = Adjective: List a word or two that describes something you saw or learned.
- E = Emotion: Describe how a particular part of the video made you feel.
- I = Interesting: Write something you found interesting about the content or topic.
- O = Oh!: Describe something that caused you to say "Oh!"
- U = Um?: Write a question you have, or what you want to learn more about.
- 5 When groups are finished with their *AEIOU* charts, form new groups with one person from each of the other four groups. Then, have students answer Part 1 in the *DNA*, *Genes*, *Proteins*, & *Mutations Review* capture sheet. In order to answer the questions in Part 1, students will need to utilize the knowledge of all four people in the group.
- 6 Have students complete Part 2. Any unfinished tasks can be completed as homework.

Slides 13–16



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Students identify and learn about the mutation associated with the inherited disease their patient. (25 minutes)

- 1 Review the capture sheet students completed on Day 1. Clarify any questions students have. 2 Tell students they are going to identify the mutations associated with their patient's inherited disease so they can include it in their report about their patient. 3 To do this work, give students the *Mutation Investigation* capture sheet. Slides 15-16 Students collaborate to compare their findings on their mutation. (20 minutes) 1 Pair up students up based on inherited disease. 2 With their partner, have students compare their research to identify any gaps. 3 Have partners identify, highlight, or underline any parts of their research they do not understand. Then prompt students to turn the highlighted text into questions. 4 Have each pair share aloud one or two questions. Questions students ask could include: What is a three base sequence? а.
 - b. What is an abnormal gene?
- 5 Add these questions to the student N2K list.



Slides 17–23

Slides 17-19

Students complete the 3-2-1 Bridge thinking routine about mutations. (10 minutes)

1		Have students complete the second of the <i>3-2-1 Bridge</i> thinking protocols using the following prompts:
	a.	Using what you learned about mutations, write new responses:
		 What are three new thoughts or ideas you have about mutations? What are two new questions you have about mutations? What is a new metaphor or simile you can make about mutations? Now, create your bridge between your first response and today's response. Explain how your new responses connect to your initial response.
2		Have a few students share how today's 3-2-1 reflections differed from their initial 3-2-1 responses on the topic of mutations.
3		Tell students they will need to identify the mutation responsible for the inherited disease associated with their patient.
4		Remind students that, in their Genetic Test Report , they will need to explain to their patient which mutation is responsible for the inherited disease, and why the mutation in DNA gives rise to the disease. To explain this, they will need to learn how the information in DNA is used to make protein through the process called Protein Synthesis.

Slides 20-23

Through an inquiry process, students begin to learn about the first step of protein synthesis, transcription. (35 minutes)

- 1 Using the *Transcription Investigation*, have students record their observations of the picture of transcription that is projected for the whole class to see. Record a list of student observations on the board by using *Train or Pass It On*—in this activity, students call on one another to add observations to the board. If a student is uncertain of what to add, they may "pass it on" to another student.
- 2 Have student pairs use the class observations to develop questions about the image of transcription.

Continues next page >

Day 3 Continued

Slides 20–23

3		Have students share aloud one or two questions from their list about transcription. These can be added to the N2K list.
4		Through the process of developing the class list of transcription questions, guide students to identify the two key questions for their research in Part 2:
	a.	What are the steps of transcription?
	b.	How does the information in DNA get to the ribosomes?
5		Use these two questions to transition into the research in Part 2 of the document.





Slides 24-27

Slides 24-27

Through an inquiry process, students begin to learn about the second step of protein synthesis, translation. (45 minutes)

1		Using the <i>Translation Investigation</i> , have students record their observations of the picture of translation that is projected for the whole class to see. Record a list of student observations on the board by using <i>Train or Pass It On</i> —in this activity, students call on one another to add observations of the text to the board. If a student is uncertain of what to add, they may "pass it on" to another student.
2		Have student pairs use the class observations to develop questions about the image of translation.
3		Have students turn one closed-ended question into an open-ended question and vice versa similar to Day 2.
4		Have students share aloud one or two questions from their list to make a class list of questions about translation.
5		Through the process of developing the class list of translation questions, guide students to identify the two key questions for their research in Day 2:
	a.	What are the steps of translation?
	b.	What is the code that is used to read the information in DNA to make proteins?
6 Use these two questions to transition into the research in Part 2 of the document.		

INDUSTRY AND CAREER CONNECTION

As students craft their narrative of how genetic mutations result in inherited disease, they are gaining experience with the career of genetic counseling.

COMPUTATIONAL THINKING IN ACTION

As students hone in on specific genetic mutations, they are practicing the computational thinking strategy of decomposition. This strategy allows students to break down the transcription process and genetic code into component parts and focus on one specific sequence that results in an inherited disease.

COMPUTATIONAL THINKING IN ACTION

As students use the manipulatives to build the mRNA, they are using the computational thinking strategy of building models. This strategy allows students to test and iterate in order to solve problems in a design.

Slides 29

Slide 29

Students create a description of the mutations associated with their inherited disease for their patient's **Genetic Test Report**. (5 minutes)

- Have students create the narrative of how the mutation gives rise to the faulty proteins associated with the inherited disease.
 To do their initial writing, students will use the *Inherited Disease Mutation Narrative* capture sheet.
 Tell students they will use what they have learned about protein synthesis and the mutation associated with their patient's inherited disease to write an informative narrative that describes:

 the mutation in the DNA that gives rise to the inherited disease
 how the mutation in the DNA, through the process of protein synthesis, gives rise to the proteins associated with the disease
- 4
- Have students work on their narrative for the remainder of class time.

Small Group

While students are finalizing their investigation documents, have some groups of students pull out with the teacher to practice building proteins with physical models. (40 minutes)

1

2

- **Prepare Ahead of Time:** Cut out the Protein Synthesis cards in the *Educator Resources section* so students can manipulate the different parts of protein synthesis.
- Call groups of students up one at a time to a section of DNA laid out on a table. Have students use the manipulatives to build the mRNA as they describe the steps of transcription using the vocabulary words. Adjust and clarify student understanding during these small group conferences. Then have students do the same thing for translation, using the mRNA to build a protein.
 - **a.** Consider having multiple cut-outs that students can use for peer-to-peer tutoring. Allow students who have successfully demonstrated their understanding of the model to tutor other students with the extra cut-outs.

INDUSTRY AND CAREER CONNECTION

As students work together to make the narrative easier to understand, they practice skills that are used by patient advocates and medical interpreters in their daily work.



INDUSTRY AND CAREER CONNECTION

As students complete the debrief section of the lesson, they utilize skills that are essential in the career of health service management. These professionals must be able to evaluate the operations of a medical facility, identify challenges, propose solutions, and assess outcomes.

Slides 30-37

Slides 30-34

Students further develop their Genetic Test Report. (35 minutes)

1	Have students take out their previously written narrative.
2	Pair students with a partner who has a different inherited disease.
3	Have students brainstorm how they can add to or modify the information in the narrative in a way that will help their patient understand the information more clearly.
4	Have students share aloud their ideas with a partner who has a different patient. As students share, add their ideas to an anchor chart for students to refer to later when they work on their Genetic Test Report .
	ideas include:
	• graphics
	 graphs infographics
	a combination of these ideas
5	Have students select one or two ideas to create and include when they write the narrative into the "More Information and Support" section of their Genetic Test Report .
6	Have students work on developing the "More Information and Support" section of their Genetic Test Report .

Slides 35-37

Debrief lesson 3 with students. (10 minutes)

- 1 Facilitate a class discussion, based on the prompts below. Have students first discuss in their groups, and then have groups share aloud one item from their group discussion.
- 2 Use the following prompts to facilitate a class debrief:
 - **a.** Take a look at our N2K questions. Which questions can we answer now based on the work we've done so far?

Continues next page >

Day 6 Continued

Slides 35–37

- **b.** What new questions do you have that we need to add to our N2K questions?
- c. Other questions to consider or discuss:
 - What are the challenges of communicating complicated scientific information to your patient?
 - How do you think your Genetic Test Report will impact your patient?
 - Forget about the technical information for a minute that you are putting in your report. How do you think the formatting, organization, and colors of the **Genetic Test Report** can help your patient understand the information you put in?
- 3 At the end of the class debrief, give students the following exit ticket prompt to do a quick *turn and talk*:
 - a. What goes into making an effective Genetic Test Report for your patient?
 - **b.** As a genetic counselor, how would you know the **Genetic Test Report** you provided to your patient is effective?



National Standards

Next Generation Science Standards

Science and Engineering Practices (SEP)

Practice 1

Asking Questions and Defining Problems

Ask questions that arise from examining models or a theory to clarify relationships.

Disciplinary Core Ideas (DCI)

LS1.A Structure and Function

All cells contain genetic information in the form of DNA molecules. Genes are regions in the DNA that contain the instructions that code for the formation of proteins.

Crosscutting Concepts (CC)

Cause and Effect

Empirical evidence is required to differentiate between cause and correlation and make claims about specific causes and effects.



































Protein Synthesis



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DNA, Genes, Proteins, and Mutations Review, Part 1 *Jigsaw*

Directions

In your group, share the information you learn while you watch the videos and make connections by answering the questions below.

Outcomes of Mutation







Proteins





DNA and Genes

1. What are the parts of DNA and how are they structured?

2. How are genes connected to proteins?

3. How are proteins connected to traits?

4. How are mutations connected to DNA and traits?

DNA, Genes, Proteins, and Mutations Review, Part 2 Parts of a Cell Review

Directions

Use the **Cell Anatomy Viewer** to identify and review the parts of the cell associated with DNA and proteins.



1. Where is the DNA located in the cell?

2. Based on the information in Part 1, you learned that information in DNA is used to make proteins. What organelles in the cell are connected to this process?

3. Does the DNA ever leave the nucleus?

4. What questions do you have about how the information in DNA is used to make proteins?

Mutation Investigation, Part 1

Intro to Mutations

Directions

Open up the page *Mutation and Genetic Variation*. Watch the video and read the article. Then answer the questions that follow.



1. How do mutations happen?

4. What does the article say about mutations being considered bad?

- 2. What is the relationship between mutations and alleles?
- 5. Think about your inherited disease. Does your mutation result from an environmental factor or from a germ-line (inherited) factor?
- 3. How does the environment play a role in the development of mutations?

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Mutation Investigation, Part 2 *Outcomes of Mutations*

Directions

Open the Outcomes of Mutation interactive. Fill out the table below. Select any three of the traits except for Cystic Fibrosis.



Selected Trait	Description of Trait	What is different in the DNA structure?	What changes in the protein?

- 1. How do mutations happen and get shared with offspring?
- 2. How do mutations lead to the development of a disease?

Mutation Investigation, Part 3

Types of Mutations

Directions

Open the Different Types of Mutations interactive.

First identify the types of mutation, and then describe how the DNA is changed when this mutation occurs.

Type of Mutation			How does the DNA change when this mutation occurs?
Point Mutation	1	Missense	
	2	Nonsense	
	3	Silent	
Insertion Mutatio	n		
Deletion Mutation			

Mutation Investigation, Part 4

Patient's Inherited Disease

Directions

Use the online resources to find information about the mutation that causes the inherited disease associated with your patient. Make note of where you found the information in the Sources column.

What You Discovered prompts

Which gene is associated with the mutation?

Which type of mutation leads to your inherited disease?

What changes in the DNA structure occur due to the mutation?

How does the protein change as a result of the mutation?

What is the effect of the changed protein on the person's body?

Inherited Disease	What You Discovered	Sources

Transcription Investigation, Part 1

My Transcription Observations

Directions

Write down all the observations you make from the this diagram depicting protein synthesis.

Protein Synthesis



Continues next page >

Transcription Investigation, Part 1

My Transcription Observations

Continued

1.	6.
	7
Z	<i>.</i>
3.	8.
4.	9.
5.	10.

Transcription Investigation, Part 2

My Transcription Questions

Directions

- a. Write down as many questions about the picture as you can. How many questions can you come up with?
- b. Identify which questions are open ended and which are closed ended.
- c. Turn one closed-ended question into an open-ended question and vice versa.
- d. Select three questions to share aloud to create a Class Transcription Questions list.

1.	6.
2.	7.
3.	8.
4.	9.
5.	10.

Transcription Investigation, Part 3

My Transcription Research

Directions

Describe the steps of transcription in detail using the resources to help.

Resources									
Article	Steps of Transcription								
Audio	Transcription described								
Video	Narrated Transcription Description (w/ subtitles)								
Video	Transcription Explained (basic)								
Video	Transcription Explained (advanced)								
Video	Triplet Code								

Step	Name	Description
1		
2		
3		

Translation Investigation, Part 1 *My Translation Observations*

Directions

Write down all the observations you make from the this diagram depicting peptide synthesis

Peptide Synthesis



Continues next page >

Translation Investigation, Part 1

My Translation Observations

Continued

1.	6.
2	7
3.	8.
4.	9.
5.	10.

Translation Investigation, Part 2

My Translation Questions

Directions

- a. Write down as many questions about the picture as you can. How many questions can you come up with?
- b. Identify which questions are open ended and which are closed ended.
- c. Turn one closed-ended question into an open-ended question and vice versa.
- d. Select three questions to share aloud to create a Class Transcription Questions list.

1.	6.
2.	7.
3.	8.
4.	9.
5.	10.

Translation Investigation, Part 3

My Translation Research

Directions

Describe the steps of transcription in detail using the resources to help.

Resour	ces
Article	Steps of Translation
Article	About Translation
Table	Codon Table
Audio	Translation described
Video	Translation Explained (basic)
Video	Translation Explained (advanced)

Step	Name	Description
1		
2		
3		

Put It All Together

Transcription and Translation Investigation

Directions

Now put what you learned about Transcription and Translation together

Re	S	ונ	ır	C	e	5																							
							 	••	 																				

Video Transcribe and Translate a Gene

Video DNA to RNA to Protein

Question	Answer
What is the information in DNA that is used to make proteins?	
Why do humans look different from one another?	

Inherited Disease Mutation Narrative

Directions

- 1. Write two paragraphs that help your patient understand:
 - a. the type of mutation in the DNA that gives rise to the inherited disease.
 - b. how the mutation in the DNA, through the process of protein synthesis, gives rise to the proteins associated with the disease.
- 2. Be sure to include the vocabulary in the word bank below. Also, use the sequence words to help.
- 3. You will include this summary in your **Genetic Test Report**. So be sure to communicate this information clearly and in a way your patient will understand.

Transcription Vocabulary	Translation Vocabulary	Sequence Words
Transcription	Transcription	First,
RNA Polymerase	Translation	Next,
Nucleotides (Thymine, Adenine, Cytosine, Guanine, Uracil)	Nucleotides (Thymine, Adenine, Cytosine, Guanine, Uracil)	Then,
DNA	Ribosome	Second,
Promoter	RNA Polymerase	Later,
Coding/encode	DNA	After that,
Termination sequence	Messenger RNA (mRNA)	Finally,
Messenger RNA (mRNA)	Amino Acid	Lastly,
Ribosome	Codon	

Continues next page >

Your Inherited Disease Mutation Narrative

Continued

Narrative