Rare Diseases, Hopeful Research

TEACHER NOTES

Objectives

- Students will learn about the lysosomal dysfunction behind the formation of Batten Disease.
- Students will learn about the probability of an individual having Batten Disease based on the genotype of the parents.
- Students will learn about STEM careers associated with rare genetic diseases such as Genetic Counselors and Medical Researchers.
- Students will learn that rare diseases, as a whole, affect almost 10% of the US population.
- Students will understand the mechanism behind an individual expressing the disease versus being a carrier of the allele for the disease.

Vocabulary

- Batten Disease
- rare genetic disease
- CoRDS
- lysosome
- lysosomal storage disorder
- enzyme
- protein
- probability
- Genetic Counselor
- DNA
- amino acid
- nucleus
- dominant
- recessive
- autosomal recessive
- carrier
- allele
- Punnett Square
- gene therapy

About the Lesson

- The lesson tells the story of Batten Disease, a lysosomal storage disorder which affects the nervous system and could lead to death.

TI-Nspire™ Navigator™

- Send out the *Rare Diseases, Hopeful Research.tns* file.
- Monitor student progress using Class Capture.
- Use Live Presenter to spotlight student answers.

Lesson Materials

- Compatible TI Technologies: TI- Nspire™ CX Handhelds, TI-Nspire™ Apps for iPad®, TI-Nspire™ Software

Tech Tips:

- This lesson includes screen captures taken from the TI-Nspire CX handheld. It is also appropriate for use with any of the TI-Nspire family of products, including TI-Nspire software and TI-Nspire App. Slight variations to these directions might be required if using other technologies besides the handheld.
- Watch for additional Tech Tips throughout the lesson for the specific technology you are using.
- Access free tutorials at http://education.ti.com/calculators/pd/US/Online-Learning/Tutorials.

Lesson Files:

Student Activity

- Rare Diseases Hopeful Research_Worksheet.pdf

TI-Nspire document

- Rare Diseases Hopeful Research.tns

Background

STEM CAREER—This activity introduces the concept of rare genetic diseases. Specifically, the activity explores the mechanism behind Batten Disease; a lysosomal storage disorder. There are several career opportunities in helping to cure and learn about rare genetic diseases. For example, Mr. Jason Flanagan is a certified Genetic Counselor who works with families. Genetic counselors help people decide whether to pursue genetic testing using medical and family histories and interpret results of the tests when they do.

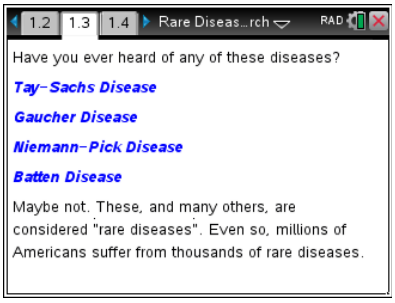
Dr. Jill Weimer is the Director of the Child's Health Research Center at Sanford Research. She leads a team of experts in search of treatments and a possible cure for Batten Disease. Dr. Weimer explains that conditions like Batten Disease are rare and might not receive the awareness they deserve. "It is critical that we do all we can to better understand rare diseases like Batten," Dr. Weimer explains.

Although Dr. Weimer and Mr. Flanagan have varied backgrounds and expertise, they are both working very hard to understand rare diseases like Batten Disease, to help families with therapies and options, and to help improve the quality of life for those afflicted.

OVERVIEW—Students will use a simulation of the mechanism behind Batten Disease to understand what's happening at the cellular level. The simulation gives a virtual model of the lysosome in which it accumulates by-products from the cell. Unfortunately, with Batten Disease, enzymes in the lysosome do not properly break down the by-products, and the result is that they accumulate. This accumulation of waste proteins eventually renders the cell non-functional.

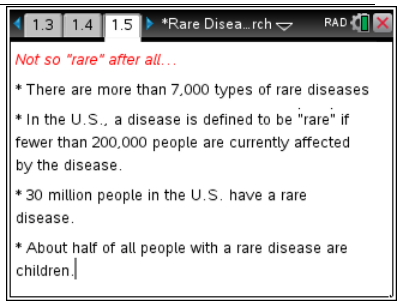
Move to pages 1.2–1.4.

1. Students are introduced to commonly known conditions such as heart disease, diabetes, and breast cancer in an effort to give some context to what makes some diseases "rare" while others are not.
2. Page 1.3 documents some actual rare genetic diseases that aren't commonly known. One of these is Batten Disease.



Move to page 1.5.

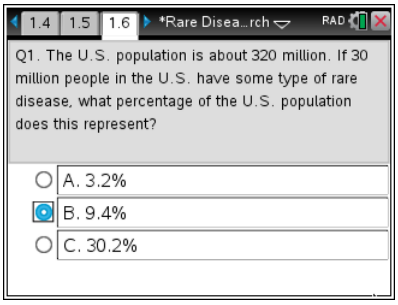
3. Page 1.5 offers students some numbers around the designation of "rare" when referring to diseases that affect fewer than 200,000 people in the US (out of an approximate population of 320 million). It also shows that there are more than 7,000 known rare diseases which, taken as a whole, affect almost 10% of the US population!



Move to page 1.6.

Q1. The U.S. population is about 320 million. If 30 million people in the U.S. have some type of rare disease, what percentage of the U.S. population does this represent?

Answer: B. 9.4%

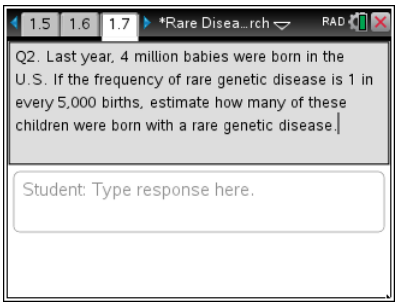


Move to page 1.7.

Q2. Last year, 4 million babies were born in the U.S. If the frequency of rare disease is 1 in every 5,000 births, estimate how many of these children were born with a rare genetic disease.

Suggested Answer: Since the rate of rare disease births is 1 in every 5,000 births and there were 4 million babies born in the U.S., students would simply divide 4 million by 5,000 to find the number of babies born with a rare disease.

4,000,000 ÷ 5,000 = 800 babies

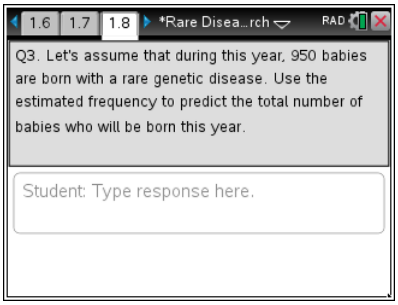


Move to page 1.8.

Q3. Let's assume that during this year, 950 babies are born with a rare genetic disease. Use the estimated frequency to predict the total number of babies who will be born this year.

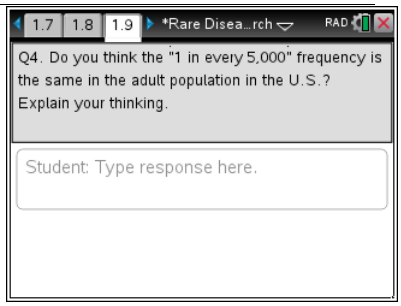
Suggested Answer: Students should use the average rate from Q2, 1 in 5,000 babies are born with a rare disease and the number of babies born this year, 950, to determine how many total babies will be born this year.

950 babies born with a rare disease x 5,000 babies = 4,750,000 total babies should be born



Move to page 1.9.

Q4. Do you think the "1 in every 5,000" frequency is the same in the adult population in the U.S.? Explain your thinking.



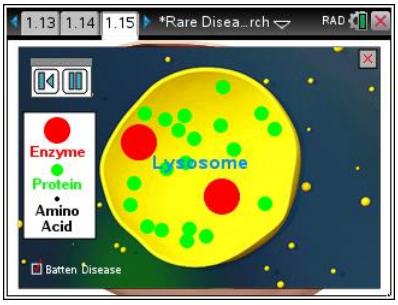
Suggested Answer: Answers will vary, but in general students should realize that many of these diseases are terminal in children so they never make it to adulthood sadly. Therefore, for every adult with a rare disease, there will be some number greater than 5,000 adults without a rare disease.

Move to pages 1.10 – 1.11.

4. Pages 1.10 and 1.11 introduce Sanford Research's CoRDS program, which stands for "Coordination of Rare Diseases at Sanford". It is a database, which connects families with a member affected by a rare disease with the researchers who are studying that disease.

Move to pages 1.12 – 1.15.

5. Pages 1.12 to 1.15 introduce students to Batten Disease. Batten Disease is characterized by a poorly or non-functional lysosome, an organelle responsible for removing and breaking down proteins the cell doesn't need. Because the enzymes in the lysosome are not able to break down these proteins, they accumulate and eventually reduce the functionality of the cell.



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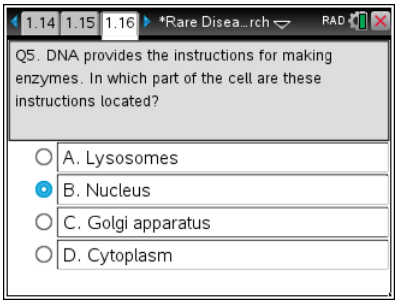
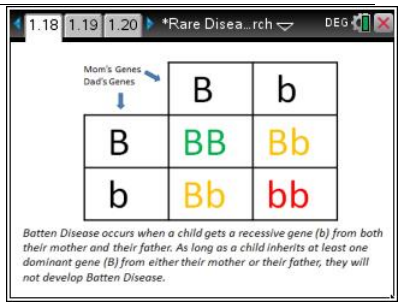
Q5. DNA provides the instructions for making enzymes. In which part of the cell are these instructions located?



Answer: B. Nucleus

Move to pages 1.17 – 1.18.

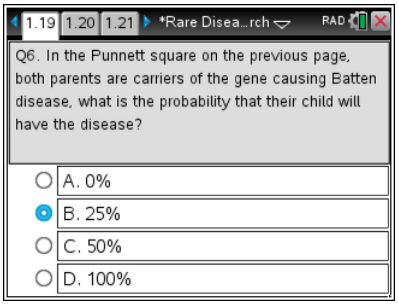
6. Pages 1.17 to 1.18 describe the inheritance of Batten Disease from parents who are both "carriers". Students will understand the probability of a child born with Batten Disease.



Move to pages 1.19 – 1.22.

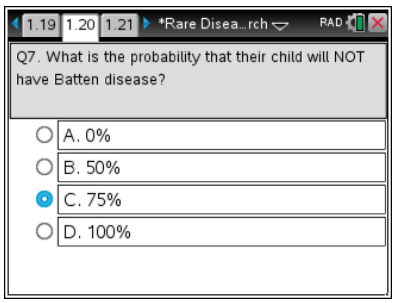
Q6. In the Punnett Square above, both parents are carriers of the gene causing Batten Disease, what is the probability that their child will have the disease?

Answer: B. 25%



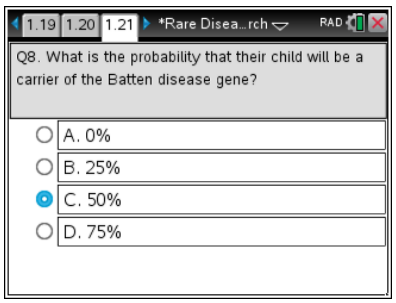
Q7. What is the probability that their child will NOT have Batten Disease?

Answer: C. 75%

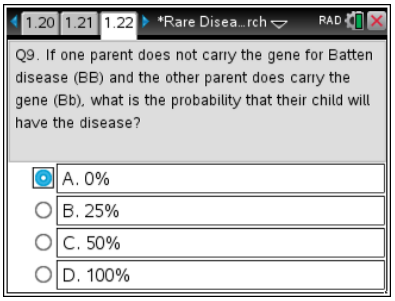


Q8. What is the probability that their child will be a carrier of the Batten Disease gene?

Answer: C. 50%



Q9. If one parent does not carry the gene for Batten Disease (BB) and the other parent does carry the gene (Bb), what is the probability that their child will have the disease?



Answer: A. 0%

Move to pages 1.23 – 1.27.

7. These pages introduce students to Mr. Jason Flanagan, a certified Genetic Counselor who works with families to interpret information based on their genetic makeup. Mr. Flanagan also discusses his career choice and the areas of study required to become a certified Genetic Counselor.

Move to pages 1.28 – 1.30.

8. Dr. Jill Weimer is a medical researcher and is currently the Director of the Children's Health Research Center at Sanford Research. Dr. Weimer leads a team of researchers and experts to better understand Batten Disease in an effort to create therapies and ideally a cure.

Move to page 1.31.

9. Page 1.31 concludes the activity by directing students to check out the great career options at places like Sanford Research.

TI-Nspire Navigator Opportunities

Make a student the Live Presenter to demonstrate his or her asteroid simulation graphs.

Assessment

- Students will answer questions throughout the lesson to ensure they understand the concepts of rare diseases and STEM careers involved.

Going Further

- To add to this lesson, you can share personal stories, ask students to share some of the things they already know about rare diseases, and talk about what's been reported n the news.

For more information about rare diseases and the work at Sanford Research check out these links:

- **CoRDS—www.sanfordresearch.org/cords/**
- **Sanford Research—www.sanfordresearch.org**

For more information about personal genetics, check out the Personal Genetics Education Project:

- **www.pged.org**