Rare Diseases, Hopeful Research

Student Activity

Name _____

Class _____

Open the TI-Nspire document *Rare_Disease_Research.tns.*

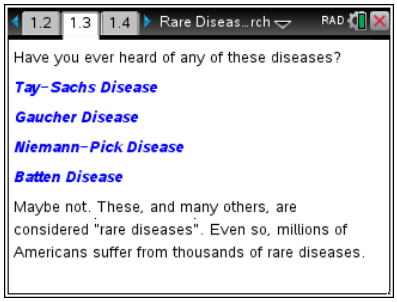
You've probably heard of breast cancer, diabetes, and heart disease. But have you ever heard of Tay-Sachs disease, Gaucher disease, Niemann-Pick disease, or Batten disease? Chances are that you haven't. That's because they are examples of "rare genetic diseases".

This activity contains information related to rare diseases with special focus on Batten disease. Along the journey, you'll also learn about a couple of very interesting STEM careers and how they relate to learning about and dealing with rare diseases.

Move to pages 1.2–1.4.

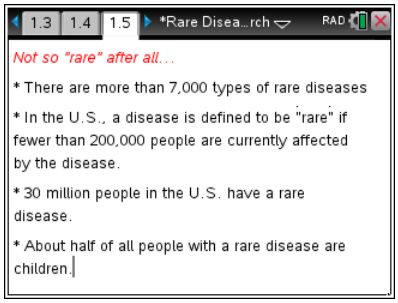
1. You are introduced to common conditions such as heart disease, diabetes, and breast cancer in an effort to give some context to what makes some diseases "rare" while others are not.

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 describes some numbers around the designation of "rare" when referring to diseases that affect fewer than 200,000 people in the US (out of a population of approximately 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!

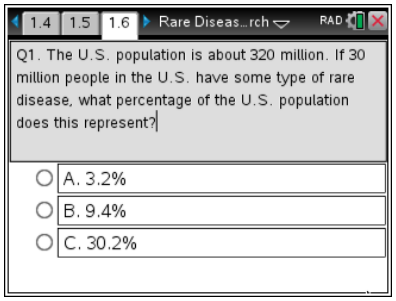


Move to page 1.6 – 1.9.

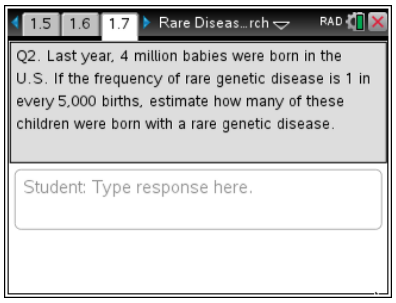
Answer the questions here and/or in the .tns file.

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?

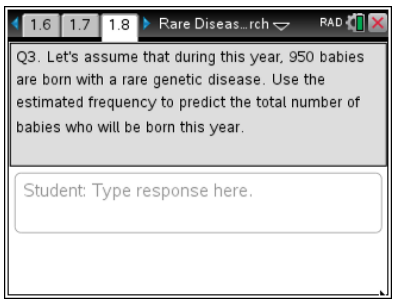
A. 3.2%
B. 9.4%
C. 30.2%



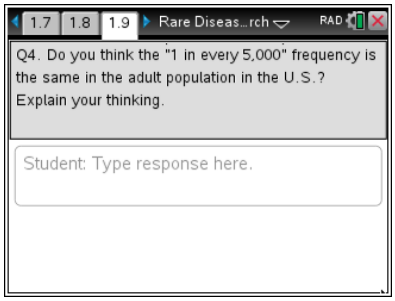
Q2. Last year, 4 million babies were born in the U.S. If the frequency of rare genetic disease is 1 in every 5,000 births, estimate how many of these children were born with a rare genetic disease.



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.



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.

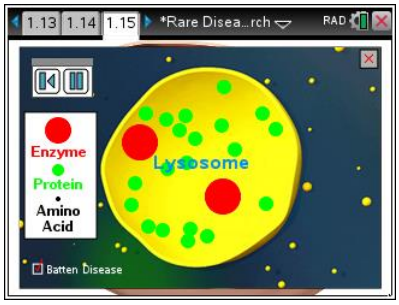


Move to pages 1.10 – 1.11.

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 that connects families that have a family member with a rare disease with the researchers who are studying the disease.

Move to pages 1.12 – 1.15.

5. Pages 1.12 to 1.15 introduce Batten disease. Batten disease is characterized by specific changes in the DNA that result in 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 entire organ systems.

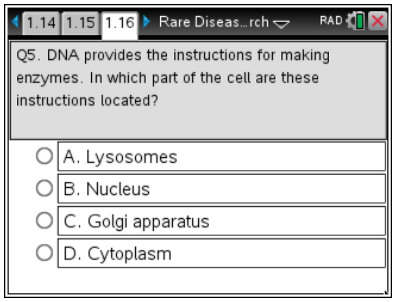


Move to page 1.16.

Answer the question here and/or in the .tns file.

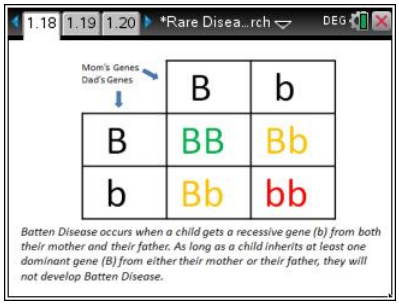
Q5. DNA provides the instructions for making enzymes. In which part of the cell are these instructions located?

A. Lysosomes
B. Nucleus
C. Golgi apparatus
D. Cytoplasm



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". From this, you will discover the probability of a child born with Batten disease.

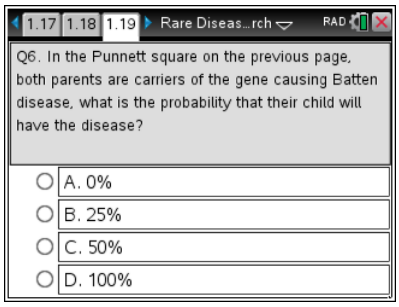


Move to pages 1.19 – 1.22.

Answer the questions here and/or in the .tns file.

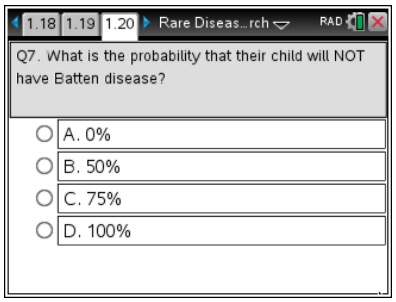
Q6. In the Punnett Square above, both parents are carriers of the gene causing Batten disease, what is the probability that their child will have the disease?

A. 0%
B. 25%
C. 50%
D. 75%



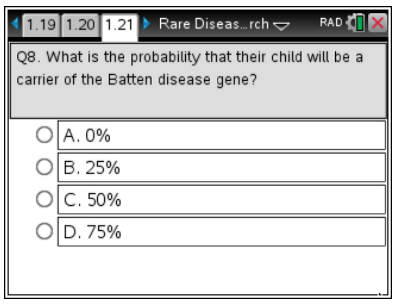
Q7. What is the probability that their child will NOT have Batten disease?

A. 0%
B. 25%
C. 50%
D. 75%

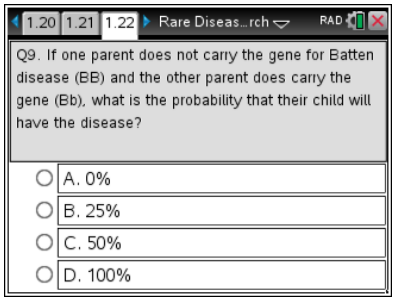


Q8. What is the probability that their child will be a carrier of the Batten disease gene?

A. 0%
B. 25%
C. 50%
D. 75%



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?



A. 0%
B. 25%
C. 50%
D. 75%

Move to pages 1.23 – 1.27.

7. These pages introduce 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 you to check out the great career options at places like Sanford Research.