Assessment Task – Inheritance and the Beery Twins

Teacher Notes:

This assessment may take several class periods to complete. Plan on a minimum of 90 minutes to complete the exercise.

Teacher Tip: A color copy of the genome sequencing data will help students effectively analyze the data. We suggest that you may either show the data on a projector OR make a classroom set of color copies for the students to analyze.

A complete explanation of the data is shown to the right. Students will be provided with only the image so that they may focus on pattern recognition and not the specific mutations.

ENGAGE your students with a story about the Beery Family. A set of fraternal twins (one male and one female) had developmental delays, poor muscle control, excessive crying and frequent vomiting. Their older brother and parents appear to be unaffected and did not exhibit these symptoms.

Student Prior Knowledge:

- DNA structure
- DNA as the genetic material
- familiarity with Mendelian patterns of inheritance
- dominant versus recessive traits
- conceptual understanding of alleles
- Punnett squares as a model of inheritance

Student Learning Objectives:

Students will be able to ...

- Analyze and interpret information in a pedigree
- Predict inheritance patterns of traits from a pedigree analysis
- Create a Punnett square model predicting outcomes of a specific genetic cross
- Examine, compare and interpret DNA sequence data
- Create models of chromosomes depicting outcomes of genetic crosses

IV. Validation and Segregation of Two SPR Alleles

The Sanger sequencing traces (below) show the SPR genotype for each member of the Beery family.

The Arg150Gly mutation is an A \longrightarrow G mutation on chromosome 2 at nucleotide 72,969,094 leading to the replacement of Arginine with Glycine. The unaffected father is heterozygous (A/G) for the pathogenic Arg150Gly allele at the first locus and homozygous (A/A) for the wild-type allele at the second locus.

The Lys251X mutation is an A → T mutation on chromosome 2 at nucleotide 72,972,139

resulting in the conversion of a Lysine codon (AAG) to a STOP codon (UAG). The unaffected mother is homozygous (A/A) for the wild-type allele at the first locus but heterozygous (A/T) for the pathogenic Lys251X allele at the second locus.



Each affected twin is a compound heterozygote - (A/G and A/T) with a different pathogenic mutation at each allele.

Figure 2. Pedigree and validation of two deleterious SPR Alleles¹

Science and Engineering Practices (SEPs)

- Asking Questions and Defining Problems.
- Developing and using models.
- Analyzing and interpreting data.

Disciplinary Core Ideas (DCIs)

- HS-LS3-1. Ask questions to clarify relationships about the role of DNA and chromosomes in coding the instructions for characteristic traits passed from parents to offspring.
- LS1.A : Structure and Function
- LS3.A : Inheritance of Traits
- o LS3.B: Variation of Traits

Crosscutting Concepts (CCCs)

- o Patterns
- Cause & Effect: Mechanism and Explanation
- Structure & Function

What's Going On with the Beery Twins?

ENGAGE

The Beery twins are fraternal twins (one male and one female) that had developmental delays, poor muscle control, excessive crying and frequent vomiting. Their older brother and parents appear to be unaffected and did not exhibit any of these symptoms.

Formative Exploration Assessment Probe

EXPLORE

Examine the photo of Alexis and Noah Beery.

- A. Make at least three observations about the twin's condition.
- B. Ask at least three questions you might have about the twin's condition.
- C. Predict what you think may have caused the Beery Twin's condition.
- D. What further information you need to know to determine the cause of their condition?

EXPLAIN

Pedigree Analysis: Analyze the pedigree below, and consider the following:

1. Describe what is meant by each of the symbols (circle vs. square, color, letters, etc.)



- 2. What medical conditions were found in the twin's relatives, on both mom and dad's side of the family?
- 3. Predict if the Beery twins' condition is dominant or recessive? Create a Punnett square to support your claim.
- 4. We know now this appears to be a genetic disorder. Propose what steps you would take to further investigate this condition.



By now you have discovered that the Beery family consists of the parents, Retta and Joe, and three children: Zach, Noah and Alexis. The immediate family underwent whole genome sequencing that revealed the sequences of each member's DNA. The significant results from that genome sequencing are shown on the right. Read and analyze the experimental data and address the questions below. (Color copies of the data are highly recommended.)

- 5. Examine Figure 2. You may find it helpful to highlight or mark similarities directly in the data figure.
 - a. What do each of the columns of data represent in Figure 2? Why would the data be separated into 2 columns?
 - b. Compare Retta's peaks in column one to the rest of her family. Which family members share the most similarities to Retta?
 - c. Next, compare the peaks in column one to Joe's pattern. Which family members share the most similarities to Joe?
 - d. Now compare Retta's peaks in column two to the rest of her family. Which family members share the most similarities to Retta?
 - e. Finally, compare the peaks in column two to Joe's pattern. Which family members share the most similarities to Joe?

EVALUATE

6. Keeping in mind that Zach appears to be unaffected, what does this sequencing data reveal about the twins?

EXTEND

- 7. Create a model of Joe's chromosome 2 which demonstrates his genotype for the targeted trait.
- 8. Create a model of Retta's chromosome which demonstrates her genotype for the targeted trait.
- 9. Using the model created above predict the combination of chromosomes inherited by Zach, Noah and Alexis.



Figure 2: Results of a genome sequencing comparing each member of the Beery family at two points on the gene located on chromosome 2

Analyzing and Interpreting Data: Pedigree Analysis

| Basic | Progressing | Proficient | Exemplary |
|---|--|---|--|
| Question 1: Students can identify and interpret the symbols on the pedigree. | Question 2: Students can use the pedigree to identify medical conditions in the family. | Question 3: Students can create a Punnett square to provide evidence of inheritance pattern of the | Question 4: Students can propose further investigations to explain the condition. |
| | | condition. | |
| Students can accurately identify and explain at least five symbols shown in the key. | Students identify DRD, neurological disorders, fibromyalgia and depression in the pedigree. | Students show a Punnett square depicting a recessive inheritance pattern. | Students suggest additional medical testing including: DNA sequencing, blood testing, additional family history, etc. |

Analyzing and Interpreting Data: Sequencing Analysis

| Basic | Progressing | Proficient | Exemplary |
|---------------------------|-----------------------------|--------------------------|----------------------------|
| Question 5a: Students can | Questions 5b, c, d & e: | Question 6: Students can | Question 7, 8, & 9: |
| relate the caption to the | Students can compare and | use the sequence data to | Students can create and |
| graphic in the figure. | recognize similar DNA | determine how the twins | use a chromosome model |
| | sequence patterns. | inherited the condition. | to depict parental |
| | | | genotypes and predict the |
| | | | genotypes of the children. |
| Students recognize that | Students recognize that | Students recognize that | Students create accurate |
| the data is for two | the twins have the same | the twins inherited a | models of the genotypes of |
| different points in the | sequence pattern from Joe | mutated gene from each | parents and the children. |
| same gene on a | at one position in the gene | parent. | |
| chromosome. | and the same pattern from | | |
| | Retta at the second | | |
| | position in the gene. | | |