

Genetics – Mutations

Teacher's Guide

1.0 Summary

The *Mutations* activity is the seventh core Genetics activity and should be run after *XLinkage*. It should take students approximately 45 minutes to complete this activity.

2.0 Learning Goals

Driving Question: What happens when you change the DNA? How are mutations inherited?

Mutations and *Mutations II* are two activities that enable students to use BioLogica's DNA model to make changes to the base pair sequences of dragon DNA and investigate the impact of these changes on phenotypes and the inheritance patterns that emerge when dragons with mutated alleles are bred. Because both *Mutations* and *Mutations II* use pedigrees, students should complete *Monohybrid* first.

Mutations introduces students to mutations through the appearance of a novel trait in a pedigree. Students then explore the role of DNA in mutations. They can modify the base pair sequences of particular dragon alleles and examine the impact of these newly created alleles on the appearance of a dragon.

Mutations II builds on **Mutations** and **Monohybrid** by enabling students to investigate how mutations are inherited. It also gives the students more practice in using Punnett squares to determine the probability of inheriting a mutated trait. It should be used after *X-Linkage*.

Learning Goals

- Students will understand that genes are base-pair sequences in DNA which determine traits.
- Students will recognize that the four bases form complementary base pairs (A-T and G-C) that link the two strands of DNA.
- Students will identify the four bases of DNA as adenine, thymine, guanine, and cytosine.
- Students will understand that mutations are changes in the base-pair sequences.
- Students will know that genetic mutations create new alleles.
- Students will understand that not all genetic mutations result in a change of phenotype.
- Students will comprehend that some mutations are dominant and some are recessive.
- Students will be able to differentiate between somatic and inheritable mutations.

Additional Teacher Background

DNA is the nucleic acid that stores and transmits genetic information from one generation to another. The structure of DNA is a double helix in which two strands are wound around each other. Each strand is made up of a chain of nucleotides. The two strands are held together by hydrogen bonds between adenine and thymine and between guanine and cytosine.

Mutations are changes in the DNA sequence that affect how that code is translated. Mutations can range from imperceptible to lethal. Gene mutations result from changes in a single gene. Chromosomal mutations involve changes in whole chromosomes. Somatic mutations occur in body cells such as the skin and cannot be inherited. Germline mutations occur in the germline cells or gametes of organisms and can be inherited.

The *DNA Model* deals with gene mutation. Most gene mutations are point mutations involving the substitution of one nucleotide for another. Substitutions generally change one of the amino acids in a protein. However, when a point mutation also involves the deletion or insertion of a nucleotide the situation is more severe. In this case, frameshift mutation occurs and every amino acid that follows the point of insertion or deletion is affected. The protein can be changed so drastically that it cannot perform normal functions.

Students will be able to make insertions, deletions, and substitutions to the DNA of the Dragon model. They will then evidence the change in the nucleotide sequences as well as the resultant physical changes to the dragon Elvira.

3.0 Standards Alignment

Alignment to National Math and Science Standards (NCTM or NSES)

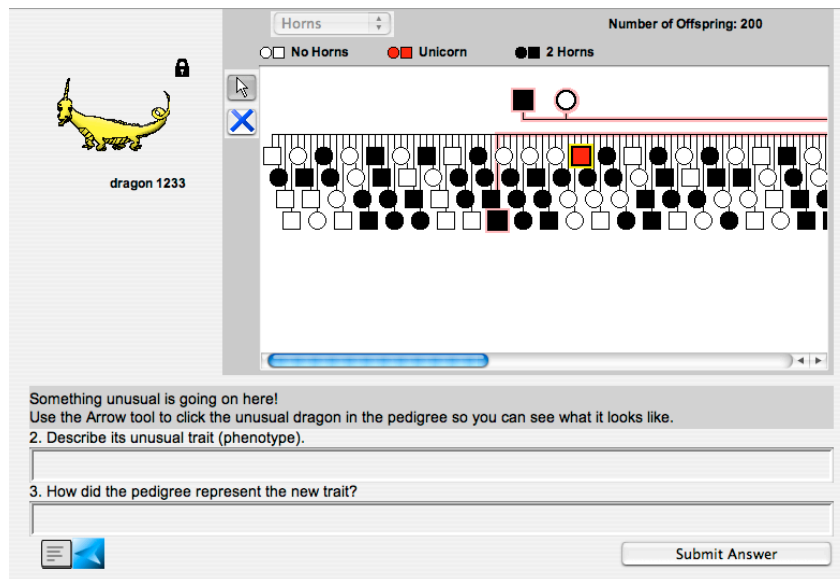
Objective		Standards	
Students will learn that DNA is a complex molecule.		Students should know the information passed down from parents to offspring is coded in DNA molecules. Students should know that each DNA molecule in a cell forms a single chromosome.	
Students will learn that changes in the DNA of sex cells result in changes in the genotype of the individual.		Students should know that only mutations in germ cells could create the variation that changes cells and organisms.	
Students will learn about the modes of inheritance of mutations.		Students should know that in all organisms, the instructions for specifying the characteristics of the organisms are carried in DNA.	
Students will further their study of probability.		Students should be able to use simulations to construct empirical probability distributions.	

4.0 Activity Sections

In this activity, students explore the appearance of a novel trait in the pedigree and the types of genetic changes that might be responsible. Students have a chance to view, identify, and modify DNA to produce the new allele and new trait. Students identify the letters ATCG as part of the DNA code. They then match letters as the base pairs A-T and C-G. Students identify the names of the molecules adenine, thymine, cytosine, and guanine. Students create substitution, deletion and insertion mutations to create new alleles. Students investigate how these new traits are inherited through the generations in order to determine dominance relationships among new and existing alleles.

4.1 A New Trait

Students investigate a new trait that appears in a pedigree.



The screenshot shows a software interface for a genetics simulation. On the left, there is a yellow dragon icon labeled "dragon 1233". The main area displays a pedigree chart with three generations. The legend at the top indicates three horn types: "No Horns" (white square/circle), "Unicorn" (red square/circle), and "2 Horns" (black square/circle). The "Number of Offspring" is set to 200. Below the pedigree, there are two text input fields for questions: "2. Describe its unusual trait (phenotype)." and "3. How did the pedigree represent the new trait?". A "Submit Answer" button is located at the bottom right.

Using their prior knowledge and pedigree tools, students determine that the unicorn trait can be inherited and that the new HU allele is dominant when paired with existing alleles.

Horns Number of Offspring: 280

No Horns Unicorn 2 Horns

10. What phenotypes are present among the offspring? Click all that apply.

No horns Unicorn Two horns

Submit Answer

Unicorn

Unicorn

Chromosome: 1

Chromosome: 1

HU Horns

h Horns

Now that you've determined that the Unicorn trait can be inherited, use the pull-down menus to check your answers.

17. Which alleles produce unicorn dragons?

HH Hh hh HUh HUHU HHU

Submit Answer

Students explore then predict how likely unicorns are to appear now that the allele is present in the population.

18. Using the information in the Punnett square, what is the probability that an offspring will be a unicorn?

0 out of 4 boxes or none
 1 box out of 4 or 1/4
 2 boxes out of 4 or 2/4
 3 boxes out of 4 or 3/4
 4 boxes out of 4 or 4/4

Buttons: Help, Show Me, Submit Answer

We then stress the rarity of mutations

4.2 DNA Model

Students then dive into the DNA model to become familiar with its parts and how they relate to the chromosomes and alleles encountered in Introduction and Meiosis.

Elvis

Chromosome: 1 H Horns

Chromosome: 1 h Horns

Let's start with Elvis. Those are his chromosomes on the right. As you can see, his Horns genotype is "Hh." As you know, our chromosome model represents the different alleles as a big H and a little h. Surely, there's more to it than that. Click one of his Horns genes (the red line in the chromosome) to learn more about the differences.

Click on red line.

View of Elvis' chromosomes, click on a gene (red line) to view DNA.

A series of screens familiarize students with BioLogica's DNA model. Questions help students focus on the base pairs that form DNA.

Elvis

Chromosome: 1 H Horns

Chromosome: 1 h Horns

Organism Allele Pair: H and h

H

h

295 T C A G T T A T A A C T G A G G C G C G C G C C A A T C T G 325

295 T C A G T T A T A A C T G A G G C G C G C G C C A A T C T G 325

Let's take a closer look; focus on just one line of boxes. You can see that it is made up of alphabet blocks.

24. What letters do you see in those blocks? Click all that apply.

P S C N A Y G X D T Y R M W O K

Submit Answer

View of DNA of Horns alleles. Red bar above base pairs shows where base pairs differ for the different alleles.

Students learn how to make changes in the base pairs and are challenged to change the dominant allele to match the recessive one.

Elvis

Chromosome: 1 H Horns

Chromosome: 1 h Horns

Organism Allele Pair: H and h

Apply Revert

H

h

295 T C A G T T A T A A C T G A G G C G C G C G C C A A T C T G 325

295 T C A G T T A T A A C T G A G G C G C G C G C C A A T C T G 325

Reset

Try it! Click the DNA strand to the right of the base pair you want to change. Hit the backspace key to delete the base pair, then type the abbreviation for the nucleotide you want to replace it with.

When you've made the change you want, click the Apply button.

If you get confused, click the Revert button to start over again.

Change the DNA to make Elvis hornless.

Students are asked about the relationships among DNA, chromosomes, genes and alleles and receive scores on their answers.

33. What is the relationship between chromosomes and DNA molecules?

- Chromosomes typically contain more than one DNA molecule.
- DNA molecules typically contain more than one chromosome.
- The number of chromosomes and DNA molecules in a typical cell nucleus is the same.

34. What is the physical relationship between genes and alleles?

- An allele is a particular part of a gene.
- A gene is a particular part of an allele.
- An allele is a particular version of a gene.
- A gene is a particular version of an allele.

35. What is the physical relationship between genes and chromosomes?

- Genes are part of chromosomes.
- Chromosomes are part of genes.
- Genes are on chromosomes.
- Chromosomes are on genes.



Submit Answer

36. What is the numerical relationship between genes and chromosomes in a typical cell nucleus?

- It contains more chromosomes than genes.
- It contains more genes than chromosomes.
- It contains the same number of genes and chromosomes.
- It doesn't contain chromosomes or genes.

37. What is the physical relationship between genes and DNA?

- Genes and DNA molecules are identical segments of information.
- Genes are segments of information attached to DNA molecules.
- Genes are segments of information within DNA molecules.

38. What is the numerical relationship between genes and DNA?

- Genes typically contain more than one DNA molecule.
- DNA molecules typically contain more than one gene.
- The number of genes and DNA molecules in a typical cell nucleus is about the same.



Submit Answer

That completes our exploration of the DNA model. You have explored the base pairs that form the critical information-bearing part of the DNA molecule and learned that:

- Each chromosome is a separate DNA molecule.
- Chromosomes contain regions of coding and non-coding DNA.
- Genes are regions of coding DNA.
- Alleles are particular sequences of base pairs for a gene.
- Different alleles have different sequences.

But what is a mutation? Continue to Part 3 and see!

Summary for Part 2

4.3 Change the DNA, Get a Mutation

In the following section students explore how the HU allele came to be (a substitution) and investigate other mutations (insertions and deletions).

You did it! You created the HU allele that gives Elvis a single horn. Of course, in the real world, we can't yet make those kinds of changes. However, since BioLogica is a model, we can do things that we can't do in real life. Let's investigate another mutation.

Students substitute a base pair to create the HU allele.

41. Wow! This is a dragon of a different color! What does this tell you about the 'aw' allele?

- The 'aw' allele is dominant to the 'a' allele.
- The 'aw' allele is recessive to the 'a' allele.
- The 'aw' allele is incompletely dominant to the 'A' allele.

42. Explain your choice.

Submit Answer

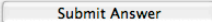
Inserting a base pair into the sequence creates a frameshift and a new allele *aw* that is recessive to both existing Color1 alleles.

Let's review!

43. In your own words, what is a mutation?
 A mutation is any change in the base pair sequences of DNA that produces a new allele.

44. The distinction between a gene and an allele is one of the most important concepts in genetics. Use the models of DNA and Chromosomes to help someone who doesn't know genetics understand the difference between a gene and an allele.
 Genes are particular locations within a DNA molecule that code for particular characteristics. Alleles are specific sequences of base pairs that combined produce specific traits. Alleles are particular versions of genes.

45. Explain what happens to the sequence of the DNA when an insertion or deletion occurs?
 The base-pair sequences are shifted so that the three base pair sequences that code for a particular amino acid might not make sense.



Students answer review questions and receive brief summary.

In this part of the activity you have learned that

- A mutation is a change in the DNA of an organism.
- Substitutions, deletions, and insertions change the sequence of base pairs in DNA and produce mutations.
- These mutations produce new alleles if they are in the coding portion of the DNA.

Keep in mind that you've been using a model of what happens in nature. It's easy for us to make these changes. It is a lot harder in real life but it does happen.

In Mutations 2, you will explore other mutations in dragons and how they are inherited.

5.0 Student Reports

Your students' work with *Mutations* is logged and viewable on the MAC Project Web Portal at <http://mac.concord.org>. For each student, you can view a report containing questions and answers.

The next activity that students should use is *Mutations2*, an optional activity in which students explore x-linked and lethal mutations through pedigree analysis.