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BioEd℠

Complex Traits

Genotypes and Phenotypes

Nancy P. Moreno, Ph.D.

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Baylor College of Medicine

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OVERVIEW

Using short- and long-haired dachshunds as examples, students learn about how a change in the nucleotide sequence affects phenotype. They predict the outcomes of crosses between short- and long-haired dachshunds.

Genotypes and Phenotypes

Traditionally, biology books have begun the section on genetics with a discussion of the experimental findings of Gregor Mendel (1822–1884). Mendel was an Austrian friar, who studied variation in plants to answer questions about heredity. He created lines of pea plants that were pure-breeding for specific traits, such as flower color (purple or white) and seed surface texture (smooth or wrinkled). He tracked the inheritance of these traits through multiple generations of pea plants, and conducted many crosses of plants with different characteristics. Mendel concluded that hereditary information is transmitted from parents to offspring in the form of discreet "particles," which we now refer to as genes and alleles.

TIBETAN TERRIER

Phenotype is the physical appearance and characteristics of an individual.

Beginning with the discovery of DNA, biologists are learning that genetics is much more complex that the particulate model of inheritance proposed by Mendel. In fact, most characteristics (phenotypes) are shaped by the actions of multiple genes, rather than the "one gene—one trait" model that so often is taught to biology students. Traits that are influenced by more than one gene, and often also by behavioral and environmental factors, are referred to as complex traits. Many common human illnesses and conditions, such as cardiovascular disease, diabetes or asthma, are the result of many genes acting in concert with environmental conditions.

To identify genetic changes associated with a particular characteristic or disease, biologists survey large sections of DNA from many different individuals to find areas where changes sequence of bases occurs consistently in affected individuals. Because new technologies make it possible to determine the exact order of the nucleotide bases—adenine (A), thymine (T), guanine (G), and cytosine (C)—in a segment or multiple segments of DNA, sections of the genomes of many different individuals can be compared to find changes in the DNA sequence that might be related to particular traits. These traits may include genetic changes that promote or prevent diseases. This type of survey approach was used to find the mutation responsible for long hair in dogs that students explored in the previous activity.

Genotype is the genetic information of an individual.

DNA sequence changes can be simple substitutions at a single location. Other kinds of changes include the insertion of extra bases, the deletion of sections of DNA, and the repetition of one or more sequences. Changes in the sequence of bases are called mutations, and they occur as mistakes when DNA is copied. However, mutations are not always detrimental. In fact, many

mutations have no apparent effect on the organism, and in some cases, are beneficial.

As shown in the activity, "Mapping a Mutation," changes in the DNA sequence can (but do not always) affect the proteins that are produced. Overall, the effects of these changes might be neutral, detrimental (leading to death, disease or reduced ability to survive and reproduce) or advantageous (such as conferring disease resistance or a competitive advantage over other individuals).

MATERIALS

- Complex Traits slide set Slides 24–26), available at http://www.bioedonline.org/slides/classroom-slides1/genetics-and-inheritance/complex-traits/
- Computer and projector, or interactive whiteboard
- Copies of the student sheets (one set per student)

PROCEDURE

1. Remind students of the previous activity, "Mapping a Mutation." Ask, *What caused some dogs to grow long hair rather than short hair?* [substitution of a single base pair, which changed the protein responsible for ending hair growth; the new protein is defective and does not stop growth of the hair shaft.] Next ask, *Was this mutation (or substitution) harmful, neutral or beneficial?* Give students opportunities to discuss the ways in which long hair might be helpful or detrimental to different kinds of dogs, depending on where they live or their activities.

2. Now, ask students, *Do you think we could use genetic information to predict which puppies might end up with long or short hair?* Allow students time to discuss the idea. Clarify for students the differences between phenotype and genotype (Slide 24). Genotype is an individual's set of genes; phenotype is all of its observable characteristics. These two concepts are related: The genotype is expressed when the information encoded in the genes' DNA is used to RNA and protein molecules. The expression of the genotype contributes to the individual's observable traits, called the phenotype. Phenotype includes body characteristics, developmental patterns, biochemical properties and even behaviors. Environment (all of the external factors that influence an organism) also contributes to phenotype.

3. Prompt student thinking by asking the following questions. *What was the nucleotide at location 20 that resulted in long hair?* [T] *How many copies of the nucleotide substitution were necessary for a dog to have long hair?* [two] *Thus, what is genotype at location 20 of a dog with long hair?* [T T; tell students that when both chromosomes have the same information, the condition is called "homozygous."]

4. Follow by asking, *Can you determine the genotype of a dog with short hair?* [In this case, the answer is no, because a short-haired dog could be homozygous for the original form of the gene that codes for short hair; or it could carry one copy of the substitution. Tell students that when an individual carries two different versions (alleles) of a gene, the condition is described as "heterozygous."]

DACHSHUND PUPPIES

5. Project Slide 25, shortly followed by Slide 26 (for use with question 6 on the student sheet). Divide students into groups and have them work through the examples on the "Dachshunds: Predict the Puppies" student sheets. Or, work through most of the questions with the class as a group. Assign question 7 (student sheet, p. 20) as homework or as an exercise for students to conduct in small groups.

Dachshunds: Predict the Puppies

MALE PARENT: FIDO FEMALE PARENT: FLUFFY

Take a careful look at the two parent dachshund dogs in the photo, then answer the questions below on the back of this sheet.

1. Which dog has the mutation that leads to long hair? Does the dog you identified have one or two copies of the mutation? Explain your answer.

2. Look at the other dog. Can you tell whether this dog also has the mutation for long hair? Explain your answer.

3. Which term refers to the appearance of each of the dogs; for example, the presence of long or short hair (phenotype or genotype)? Which term refers to the DNA variations that are present in each of the dogs (phenotype or genotype)?

4. Just like they use letters to refer to nucleotides (A, C, T, G), biologists also assign letters to specific characters and traits that they observe. For example, the letter "L" can stand for hair length. However, an upper-case "L" is used when only one copy of an allele enables the trait to be seen. A lower-case "l" is used when two copies of an allele are necessary for the trait to be present.

   Based on this information, how would you write the possible genotype or genotypes of Fido? How would you write the possible genotype or genotypes of Fluffy?

5. If you know the genotype of each parent, is it possible to predict the genotypes *and* phenotypes (appearance) of each of the offspring? Explain your answer.

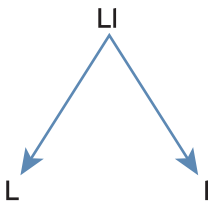
(*Continued on page 20.*)

Dachshunds: Predict the Puppies

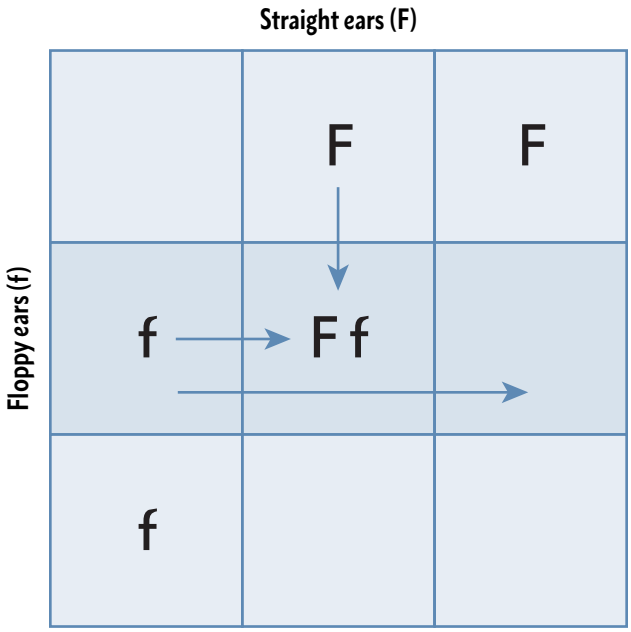
(*Continued from page 19.*)

6. During reproduction, every offspring receives a single copy of each gene from each parent. Logically, since every offspring has two parents—in the end, each puppy once again will have two copies of each gene.

   Thus, if a parent has the genotype, Ll, half of the offspring will receive an "L" version of the gene, and the other half will receive an "l" version of the gene.



   This process can be represented using a simple table that allows you to plot the outcomes of any possible cross between two parents with known genotypes. The genotype of one parent is written across the top. The genotype of the other parent is written along the left side. The example below has the genetic contribution of one parent already written across the top, and the contribution of another parent along the side. It is using a hypothetical example of floppy ears (f) vs. straight ears (F).



   Next, fill in the boxes by copying the row and column-head letters into each square. An example with the first square is completed for you. Create a similar table and fill in the remaining squares. Each of the squares represents the genotype of a single possible offspring.

   Assume that capital letter "F" refers to a fictitious gene for ear type, and signifies straight ears, while lower case "f" leads to floppy ears. When two copies of "f" are present are present, a dog will have floppy ears. What are the phenotypes of the offspring?

7. Now, based on what you know about the phenotypes and genotypes of Fido and Fluffy, make one or more tables to estimate what their puppies might look like. If you do not know the genotype of one of the parents, make a separate table for each of the possible genotypes.