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Human Genetic Traits

Background

Modern genetics began with the work of Gregor Mendel and his study of pea plants. Mendel observed patterns in the number of each phenotype (physical appearance) of offspring from different parents. Mendel described genetic concepts that are used today to explain simple heredity (what traits are passed from one generation to the next) and variation (the differences between parents and their offspring). Through his experiments, Mendel determined that there can be multiple forms of the same gene. These alternate forms of genes, which are called alleles, code for slightly different expressions of a genetic trait. He recognized that some alleles exhibited dominant tendencies and others, recessive tendencies. When a dominant allele combined with a recessive allele, the dominant allele was expressed in the phenotype. Following the system that Mendel used in his experiments, a dominant allele is represented with a capital letter, and a recessive allele is represented with a lowercase letter.

For decades, teachers and professors have used a few classic human traits to demonstrate the concepts of simple autosomal inheritance. If this activity, you will observe the phenotypes of people in your class. You will apply your results, along with research findings, to predict whether each trait follows patterns of simple autosomal inheritance.

Mendelian Laws apply to the simple autosomal inheritance that Gregor Mendel studied, but these laws do not always apply to more-complex modes of genetic inheritance.

1. **The Law of Segregation:** Inherited traits are determined by genes, with two copies of each gene. Parental genes are randomly separated and segregated into each sex cell (gamete) with one copy of each gene per gamete. Offspring inherit one copy of each gene from each parent.
2. **The Law of Independent Assortment:** The inheritance of one trait is not dependent on the inheritance of another.

   It turns out that some genes, and therefore some traits, are linked. Therefore, they are not assorted independently. Genes in close proximity to one another on the same chromosome are, in fact, more likely to be inherited together.
3. **The Law of Dominance:** If the inherited genes have multiple alleles, the dominant allele will determine the phenotype of the organism.

   In fact, some alleles are codominant, meaning that multiple alleles contribute to the phenotype of the individual. Traits may have more than two alleles for the same gene. Furthermore, multiple genes may contribute to the phenotype of one trait.

The Traits in Question

Refer to the Genetic Traits Photosheets for images of phenotypes.

PTC Tasting

Phenylthiocarbamide, or PTC, is one type of molecule that some people perceive as bitter. Molecules similar to PTC occur naturally in some plants, and people who are sensitive to PTC are generally also sensitive to the bitterness in certain plants. Control Taste Paper serves as a control for the taste test. Any taste sensation that you have from this untreated paper is a result of the paper itself.

Tongue Rolling

Some people have the ability to roll their tongue into a tube shape.

Earlobe Attachment

Earlobes attach to the head at various angles. Depending on this angle, earlobes are referred to as either attached or unattached (or free).

Thumb Angle

The end of the thumb may be aligned with the rest of the thumb or bent back at an angle. If the end of the thumb is at an extreme angle, it is described as a "hitchhiker's thumb."

Cheek Dimples

When some people smile, small creases called dimples form in the cheek. People may have dimples on both sides of the face, on one side, or neither.

Hairline

The shape of the hairline across the forehead may vary. If the hairline in the front of the head comes to a point, it is called a "widow's peak." Other people have a straight hairline.

Pre-laboratory Questions

1. If the ability to taste PTC were controlled by only two alleles: one dominant (*T*) and one recessive (*t*), would there be any way to distinguish between the heterozygous (*Tt*) individuals and the homozygous dominant (*TT*) individuals without mating or performing DNA analysis? Explain your answer.

2. Explain how a child could display a dominant phenotype that was inherited from two recessive phenotype parents.

3. After reviewing the Procedure section, explain the purpose of using two different taste papers to test for the ability to taste PTC.

Materials

control taste paper (white)	paper towel
PTC taste paper (blue)	Genetic Traits Photosheet

Procedure

1. Use a pencil to label two different regions on the paper towel. Label one side "Control," and the other, "PTC." Your teacher will distribute the taste papers to your paper towel.
2. Place the untouched end of the control taste paper on your tongue. Move it around to be sure the sample becomes saturated with your saliva and contacts taste receptors in different regions of your tongue. Discard the control taste paper.
3. Place the untouched end of the PTC paper on your tongue. Move it around to be sure the sample becomes saturated with your saliva and contacts taste receptors in different regions of your tongue. Discard the PTC taste paper.
4. Record your observations of taste in your Data Table.
5. With your partner, use the Genetic Traits Photosheet to help determine your phenotype for the remaining traits. If your phenotype does not clearly match one of the two descriptions, select "other," and describe your phenotype in the comments section. Help your partner verify his or her phenotype for each trait.
6. Complete the Data Table with the results from the entire class.
7. Answer the Analysis Questions.

Analysis Questions

If a trait is simple and genetically inherited, then:

- the trait is not determined by environmental factors.
- two distinct phenotypes are observed.
- one allele is dominant and one allele is recessive. Two parents with recessive phenotypes will produce offspring with the recessive phenotype.
- identical twins express the same phenotype.

Do these traits follow Simple Mendelian Laws?

1. Bitter Tasting
   Claim: The ability to taste PTC is a simple, two-allele inherited trait, and the allele for tasting (*T*) is dominant over the nontasting allele (*t*).
   Analyze the following information to support or refute the claim.
   a. Approximately 70% of people tested are tasters of PTC.
   b. Sensitivity to PTC can vary by five orders of magnitude, meaning that some people can detect it at very low concentrations and others can only detect it at high concentrations. Most people fall into one of two categories: those who taste PTC at low doses and those who cannot taste it at all.
   c. Chimpanzees, like humans, show variable sensitivity to PTC bitterness.

2. Tongue Rolling

   Claim: Tongue rolling is a simple, two-allele inherited trait, and the allele for rolling (*R*) is dominant over the nonrolling allele (*r*).

   Analyze the following information to support or refute the claim.

   a. Values compiled from a number of studies reveal that the tongue rolling phenotype occurs in 65–82% of individuals.
   b. Some individuals can slightly lift the edges of their tongue but cannot roll it completely.
   c. Another study revealed that tongue rolling can be learned. Many children cannot roll their tongues when first asked, but develop the ability over time.
   d. A study conducted by Philip Matlock in 1940 examined the tongue rolling ability of 33 pairs of identical twins. Eighteen pairs of twins were capable of rolling their tongues, and eight pairs of twins were not. In seven pairs, one identical twin could perform tongue rolling, but the other could not.

3. Earlobe Attachment

   Claim: The attachment of the earlobe is a simple, two-allele inherited trait. The allele for unattached earlobes (*F*) is dominant and the allele for attached earlobes is recessive (*f*).

   Analyze the following information to support or refute the claim.

   a. Although most people have distinctly attached or unattached lobes, a subset of the population possesses intermediate or partially attached earlobes.
   b. A 1973 study conducted by Mohanraju and Mukherjee in India found that the offspring of two people with unattached earlobes produced children with unattached earlobes 93% of the time. A pairing of one parent with attached earlobes and one with unattached earlobes produced children with unattached earlobes 50% of the time.

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Data Table

PTC Tasting			
	Bitter Taster	Nontaster	Comments
Personal			
Class Totals			
% Class			

Tongue Rolling				
	Roller	Nonroller	Other	Comments
Personal				
Class Totals				
% Class				

Earlobe Attachment				
	Unattached	Attached	Other	Comments
Personal				
Class Totals				
% Class				

Thumb Angle				
	Straight	Hitchhiker's	Other	Comments
Personal				
Class Totals				
% Class				

Cheeks				
	Dimples	Nondimpled	Other	Comments
Personal				
Class Totals				
% Class				

Hairline				
	Straight	Widow's Peak	Other	Comments
Personal				
Class Totals				
% Class				

Appendix A: Hardy–Weinberg Background

Genetic studies are often carried out with controlled mating of genetically uniform stocks of plants and animals, but selective breeding among humans is rare. Therefore, much of what we know about human heredity is based on frequency analysis.

You will use the Hardy–Weinberg equation, a mathematical formula, for estimating the PTC tasting alleles of the classroom population. For this activity, we will assume that there are only two alleles for the tasting trait (there are in fact five additional rare forms). The equation $(p + q)^2 = 1$ indicates that the proportion of p alleles plus the proportion of q alleles is 1. However, this gives only the allele frequencies in a population. To determine genotypic frequencies, the equation is expanded by squaring both sides to get $p^2 + 2pq + q^2 = 1$. In this equation, p^2 represents the proportion of individuals with a homozygous dominant genotype, $2pq$ represents the proportion of individuals with a heterozygous genotype, and q^2 represents the proportion of individuals with a homozygous recessive genotype.

The Hardy–Weinberg equation is dependent on several specific conditions. If these conditions are met, the population's allelic and genotypic frequencies will remain statistically constant over time, a condition referred to as Hardy–Weinberg equilibrium.

Hardy–Weinberg Equation

$p^2 + 2pq + q^2 = 1$

p^2 represents the frequency of *TT*.

$2pq$ represents the frequency of *Tt*.

q^2 represents the frequency of *tt*.

Hardy–Weinberg Conditions

1. The population size is infinite or very large.
2. Mating within the population is random. This means that there is no mating preference for any specific phenotype over another.
3. There is no mutation occurring in the population.
4. There is no exchange of genetic information with other populations—no immigration or emigration of individuals.
5. There is no selection for one phenotype over another. All phenotypes have an equal chance of survival and of having their genes passed on to future generations.

These conditions are not completely met in your classroom, but we can still use the equation to predict approximate genotypic frequencies.

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Appendix B: Hardy–Weinberg Analysis Worksheet

What is the recessive allele in this population? This will be represented by q in the following equations.

____

What is the dominant allele in this population? This will be represented by p in the following equations.

____

Fill in the following information based on the population being studied.

Step 1: Find the distribution of phenotypes within a population.

	Number of Individuals
Dominant Phenotype (PTC tasters)	
Recessive Phenotype (PTC nontasters)	
Total	

Step 2: Find the frequency of each allele within the population. Let p represent the frequency of *T*, and q represent the frequency of *t*. Therefore, q^2 represents individuals with the genotype *tt*. Solve for q.

a. Divide the number of individuals expressing the recessive phenotype by the total number of individuals. This value is q^2.

   ________ ÷ ________ = ________

b. Take the square root of q^2 to find q.

   √________ = ________

c. Subtract q from 1 to find p.

   1 – ________ = ________

Step 3: Based on the values determined for p and q, find the proportion of the class with each genotype. Added together, these values should equal 1.

a. homozygous dominant = p^2 = ________^{2} = ________

b. heterozygous = 2pq = 2 × ________ × ________ = ________

c. homozygous recessive = q^2 = ________^{2} = ________

Step 4: Multiply each of the proportion values to determine the number of individuals in the population (class) with that genotype.

a. homozygous dominant = p^2 × total # individuals = ______ × ______ = ______

b. heterozygous = 2pq × total # individuals = ______ × ______ = ______

c. homozygous recessive = q^2 × total # individuals = ______ × ______ = ______