# Mendel's Laws: Human Inheritance of Single Gene Traits

## A Brief Review of Mendel's Work with Garden Pea Plants

In garden pea plants, there are two character states for pea height, tall and short. Mendel began with pure lines of pea plants. In plants, **pure lines** are strains that consistently yield offspring with the same traits generation after generation. Mendel grew plants that were of pure line for tall and plants that were of pure line for short. He crossed a pure line tall parent plant with a pure line short parent plant (the **P generation**) to produce hybrids (the **$F_1$ generation** [first filial], in simple terms, the children of the parent plants). He then obtained a second hybrid generation (the **$F_2$ generation** [second filial], in simple terms, the grandchildren of the parent plants) by crossing the $F_1$ generation plants with each other. The following results were observed…

| | Crossing | Results |
|---|---|---|
| 1st cross | pure line tall X pure line short | 100% tall hybrids (children of the pure line parent plants that were as tall as the parent plant) |
| 2nd cross | hybrid tall $F_1$ X hybrid tall $F_1$ | ¾ tall to ¼ short plants (the grandchildren of the pure line parent plants) |

## Mendel made the following conclusions…

1. **Alleles** control an inherited characteristic & exist in individuals in pairs (You inherit one member of the pair from your father & one member of the pair from your mother). The two alleles of a pair are the same in **homozygous** individuals (e.g. the pure line short plants are homozygous for the character state of short. Their allele pair is short/short). The two alleles of the pair differ in **heterozygous** individuals (e.g. the hybrid $F_1$ generation inherited a tall allele from one parent and a short allele from the other parent. Thus their allele pair is tall/short). The **genotype** is the allele combination that produces a character state. The genotype of the hybrid $F_1$ generation is tall/short. The **phenotype** is the visible, physical trait. The phenotype of the hybrid $F_1$ generation is tall.

2. **LAW OF DOMINANCE**: Whenever the two alleles of a pair in a given individual differ, only one, the dominant one will be expressed. In the hybrid $F_1$ generation, the plants inherited one tall allele and one short allele. Yet, all of these plants appeared to be tall. Thus tall is the **dominant allele** (the allele that indicates the appearance of heterozygotes). One allele is said to be dominant over another if a heterozygous individual for that allele has the same appearance as an individual homozygous for it. The short allele is the **recessive allele** (an allele whose phenotype effects are masked in heterozygotes by the presence of a dominant allele).

3. **LAW OF SEGREGATION OF ALLELES**: When the gametes (egg & sperm) are formed by an individual, only one member of each allele pair is included in a gamete. Recall that gametes are **haploid**. When the hybrid $F_1$ generation plants produce gametes, each gamete will receive only one allele for plant height. So, an egg (or a sperm) will have an allele for tall or an allele for short, but not both. When the egg unites with the sperm during **fertilization**, the sperm will carry one allele for plant height, restoring the allele pair and the **diploid** condition.
YOU PASS ON ONLY ONE ALLELE PER TRAIT

4. **LAW OF INDEPENDENT ASSORTMENT**: All of the possible kinds of gametes that can be formed, will be formed in equal proportions. Alleles for different traits are inherited independently of each other if they are located on different homologous gene pairs.

If Mendel had a tall pea plant, how could he be certain that it was a pure line tall plant?

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# Human Inheritance of Single Gene Traits - Tasting

**Tasting:** Does broccoli taste bitter? Is eating hot peppers intensely painful? Scientific evidence suggests a genetic basis for food preferences – and it's all on the tip of the tongue. We will explore your ability to taste 3 chemicals.

1. **PTC** (phenylthiourea), is an organic compound that **either tastes very bitter or is virtually tasteless**, depending on your genetic makeup. **About 70% of people can taste PTC**. Food choice is related to a person's ability to taste PTC. One study found that non-smokers and those that don't often drink coffee or tea are more likely to be able to taste PTC. The ability to taste PTC (C) is dominant to the inability (c) to taste this chemical.

2. **Thiourea** is another organic molecule that **either tastes very bitter or is virtually tasteless**. A study at Yale University found that people could be classified into three groups, "**supertasters**" who could not abide the taste test paper at all, **medium tasters** who did not like it but tolerated it, and **nontasters** who could not taste anything. **Supertasters will not consume any bitter foods**, like dark-green leafy vegetables, coffee or chocolate, and **represent perhaps 10 to 15%** of the population. Nontasters have higher rates of thyroid disease - and of course thiourea compounds are antithyroid agents. The ability to taste thiourea is dominant to the inability to taste it.
   a. Super tasters are homozygous dominant (TT).
   b. Medium tasters are heterozygous (Tt).
   c. Nontasters are recessive (tt)

3. **Sodium benzoate** is a type of salt that may occur naturally in some foods but is more likely to be added as a preservative to foods. It is used in small amounts only (usually in acidic foods like sodas and fruit juices) because too much makes the food taste very bitter. Sodium benzoate tastes differently to different people. Some may perceive a **salty taste** while others may claim the sodium benzoate tastes **sour, bitter or sweet**. Others may not taste the sodium benzoate at all. Approximately **75% of people can taste it**. The ability to taste Sodium benzoate (S) is dominant to the inability (s) to taste this chemical.

| GENETIC TRAITS | | YOUR RESULTS | | CLASS TABULATION | |
|---|---|---|---|---|---|
| **Phenotype** | **Genotype** | **Phenotype** | **Genotype** | **# Dominants** | **# Recessives** |
| PTC taster<br>Nontaster | CC or Cc<br>cc | | | | |
| Thiourea Supertaster<br>Medium taster<br>Nontaster | TT<br>Tt<br>tt | | | | |
| Sodium Benzoate taster<br>Non taster | SS or Ss<br>ss | | | | |

How are allele frequencies calculated? Using the Hardy-Weinberg Principle

$$p^2 + 2pq + q^2 = 1.0$$

A. **p** = frequency of dominant allele. **q** = the frequency of the recessive allele.

B. **Let's consider the example of the ability to taste PTC.**

1. **We know that 70% of people can taste it**.
    a. The ability to taste it is controlled by the dominant allele, C. People who can taste PTC are either CC or Cc.
2. 30% of people are unable to taste PTC and must be cc.
3. Let's apply this knowledge to the Hardy-Weinberg formula.

| Formula | $p^2$ | 2pq | $q^2$ | = 100% |
|---|---|---|---|---|
| Genotypes | % Homozygous dominant | % Heterozygous dominant | % Homozygous recessive | = 100% |
| | % CC | % Cc | % cc | = 100% |

a. The value we know is $q^2$ = .3 (or 30%)
    1) $q = \sqrt{q^2}$
    2) $q = \sqrt{.3} = .55$ (or 55%)

b. Now we can figure p because p + q = 1.0 (or 100%)
    1) p = 1.0 – q = 1.0 - .55 = .45 (or 45%)

c. We will put this information into our formula.
    1) $p^2 + (2)(.45)(.55) + .3 = 1.0$
    2) $p^2 + 0.495 + .3 = 1.0$
    3) $p^2 = .205$

d. **Using Hardy-Weinberg, we now know what percent of the population is homozygous dominant, heterozygous dominant, and homozygous recessive**.

| Formula | $p^2$ | 2pq | $q^2$ | = 100% |
|---|---|---|---|---|
| Genotypes | % Homozygous dominant | % Heterozygous dominant | % Homozygous recessive | = 100% |
| | 20.5% | 49.5% | 30% | = 100% |

C. **Let's consider another example: the ability to roll your tongue.** Tongue rolling (R) is dominant to the inability to roll the tongue (r). The two alleles can result in only 3 possible genotypes: RR, Rr, and rr.

1. The probability of receiving R from both parents (RR genotype) is p x p, or $\mathbf{p^2}$.
2. The probability of receiving r from both parents (rr genotype) is q x q, or $\mathbf{q^2}$.
3. Receiving the Rr combination is described by **2pq** since it is possible for the R or r allele to come from either parent, thereby doubling the chance.
4. **Consider this example: In a class of 28 students, 23 were rollers (RR or Rr), while 5 were not (rr).**
    a. 5 out of 28 can't roll their tongues. 5 ÷ 28 = 0.18 (18% are non-rollers).
    b. $\mathbf{q^2 = 0.18}$. q is the square root of 0.18, or 0.42.
        1) The frequency of the recessive allele in this class is 42%.
    c. There are only two possible alleles for this trait (R and r).
        1) Therefore, p + q = 100% (that's the same as p + q = 1.0)
        2) p = 1 – q
        3) p = 1 - .42
        4) p = .58
    d. The frequency of the heterozygous condition would be **2pq or 2(.42)(.58) = .49**

5. Going back to our original formula…

a. $p^2 + 2pq + q^2 = 1.0$

b. $p^2 + .49 + .18 = 1$

c. **$p^2 = .33$**

## Calculate the Allele Frequencies for our Class

Remember the following: **$p^2 + 2pq + q^2 = 100\%$**

**p** is the frequency of the dominant allele

**q** is the frequency of the recessive allele

- $p^2$ = frequency of homozygous dominant individuals
- $2pq$ = frequency of heterozygous individuals
- $q^2$ = frequency of homozygous recessive individuals

| | PTC | Thiourea | Sodium Benzoate |
|---|---|---|---|
| Frequency of the recessive genotype<br>% of nontasters (homozygous recessive)<br>($q^2$) = # NONTASTERS / ALL STUDENTS | | | |
| Frequency of the recessive allele<br>% of recessive allele in the gene pool<br>(q) = $\sqrt{q^2}$ | | | |
| Frequency of the dominant allele<br>% of dominant allele in the gene pool<br>(p) = 100% - q | | | |
| Frequency of the homozygous dominant genotype<br>% of homozygous dominant tasters<br>($p^2$) = (p)(p) | | | |
| Frequency of the heterozygous genotype<br>% of heterozygous tasters<br>(2pq) = (2)(p)(q) | | | |

## Human Inheritance of Single Gene Traits – A Classroom Inventory

Determine your own phenotype for each of the following traits. For some of the more easily observed traits you may be able to recall the phenotypes of your parents. If it is possible, determine your own genotype by comparing phenotypes of your parents and your own phenotype. If you have the dominant phenotype, give both possibilities for your genotype where it cannot be determined definitely. Record your phenotypes in the table below. Group data of the entire class will be tabulated. Record in your table the class results.

| **GENETIC TRAITS** | | **YOUR RESULTS** | | **CLASS TABULATION** | |
|---|---|---|---|---|---|
| **Phenotype** | **Genotype** | **Phenotype** | **Genotype** | **# Dominants** | **# Recessives** |
| Double-jointed thumb<br>Tight joints | JJ or Jj<br>jj | | | | |
| Widow's Peak<br>Straight hair line | WW or<br>Ww<br>ww | | | | |
| Curly hair<br>Wavy hair<br>Straight hair | CC<br>CC'<br>C'C' | | | | |
| Nonred hair<br>Red hair | NN or Nn<br>nn | | | | |
| Dark hair<br>Blond hair | MM or<br>Mm<br>mm | | | | |
| Freckles<br>No freckles | FF or Ff<br>ff | | | | |
| Cheek dimple<br>No cheek dimple | DD or Dd<br>dd | | | | |
| Free earlobes<br>Attached earlobes | EE or Ee<br>ee | | | | |
| Tongue rolling present<br>Tongue rolling absent | RR or Rr<br>rr | | | | |
| Digital hair present<br>Digital hair absent | MM or<br>Mm<br>mm | | | | |
| Straight thumb<br>Hitchhikers's thumb | HH or Hh<br>hh | | | | |
| Left thumb over right<br>Right thumb over left | LL or Ll<br>ll | | | | |
| Bent little finger<br>Straight little finger | BB or Bb<br>bb | | | | |
| Cleft chin present<br>Absence | CC or Cc<br>cc | | | | |
| Unibrow<br>Separate eyebrows | UU or Uu<br>uu | | | | |

For how many traits are you recessive? ______________________

On the basis of the tabulated results for the entire class, are there any recessive traits which are more common among the class members than the dominant condition? _________ If so, highlight them.

What difference would you expect in the tabulation if 1000 students were included in the table?

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