

Genetics Worksheet

Genetic Terminology

Locus – the location on the chromosome where a gene is found.

Gene - a unit of inheritance that typically is directly responsible for one trait or character.

Allele - an alternate form of a gene. Usually there are two alleles for every gene, sometimes as many as three or four.

Homozygous - when the two alleles are the same.

Heterozygous - when the two alleles are different, in such cases the dominant allele is typically expressed.

Dominant - a term applied to the trait (allele) that is expressed regardless of the second allele.

Recessive - a term applied to a trait that is only expressed when the second allele is the same (e.g. short plants are homozygous for the recessive allele).

Phenotype - the physical expression of the allelic composition for the trait under study.

Genotype - the allelic composition of an organism.

Punnett square - a probability diagram illustrating the possible offspring of a mating.

Introduction

An Austrian monk, Gregor Mendel, developed the fundamental principles that would become the modern science of genetics. Mendel demonstrated that heritable properties are parceled out in discrete units, independently inherited. These eventually were termed genes.

Mendel reasoned an organism for genetic experiments should have:

1. a number of different traits that can be studied
2. plant should be self-fertilizing and have a flower structure that limits accidental contact
3. offspring of self-fertilized plants should be fully fertile.

Mendel's experimental organism was a common garden pea (*Pisum sativum*), which has a flower that lends itself to self-pollination. Most flowers allow cross-pollination, which can be difficult to deal with in genetic studies if the male parent plant is not known. Since pea plants are self-pollinators, the genetics of the parent can be more easily understood. Peas are also self-compatible, allowing self-fertilized embryos to develop as readily as out-fertilized embryos. Mendel tested all 34 varieties of peas available to him through seed dealers. The garden peas were planted and studied for eight years. Each character studied had two distinct forms, such as tall or short plant height, or smooth or wrinkled seeds. Mendel's experiments used some 28,000 pea plants.

Table 14.1 The Results of Mendel's F₁ Crosses for Seven Characters in Pea Plants [true breeding: itself = itself]

Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant:Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6022:2001	3.01:1
Seed shape	Round 	×	Wrinkled 	5474:1850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

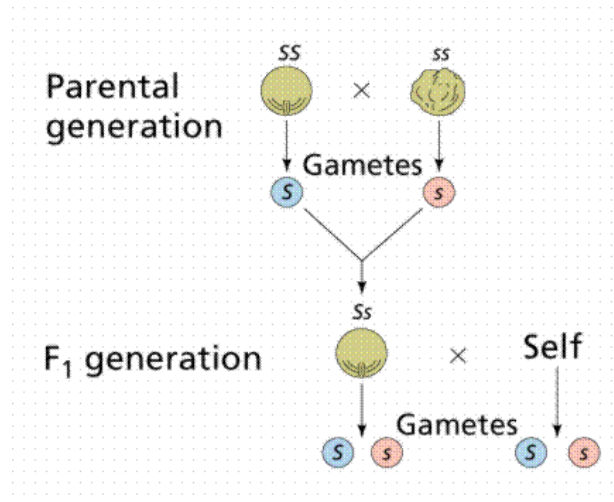
Traits as expressed in garden peas. Image copy-write 2005 © Pearson Education Inc., publishing as Benjamin Cummings. Used with permission.

Mendel's work showed:

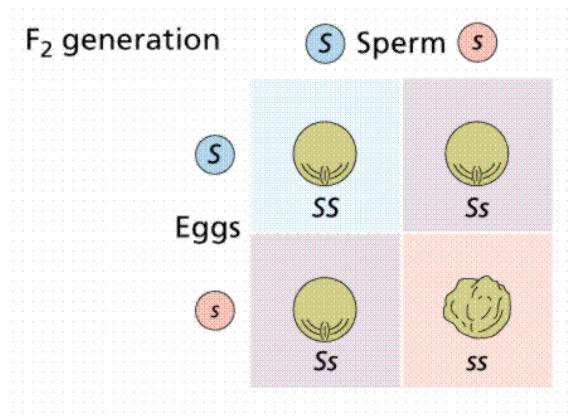
1. Each parent contributes one factor of each trait shown in offspring.
2. The two members of each pair of factors segregate from each other during gamete formation.
3. The blending theory of inheritance was discounted.
4. Males and females contribute equally to the traits in their offspring.
5. Acquired traits are not inherited.

Principle of Segregation

A cross involving only one trait is referred to as a monohybrid cross. Mendel crossed true-breeding, smooth-seeded plants with a variety that had always produced wrinkled seeds. The parental generation is denoted as the P1 generation. The offspring of the P1 generation are the F1 generation (first filial). The self-fertilizing F1 generation produced the F2 generation (second filial).

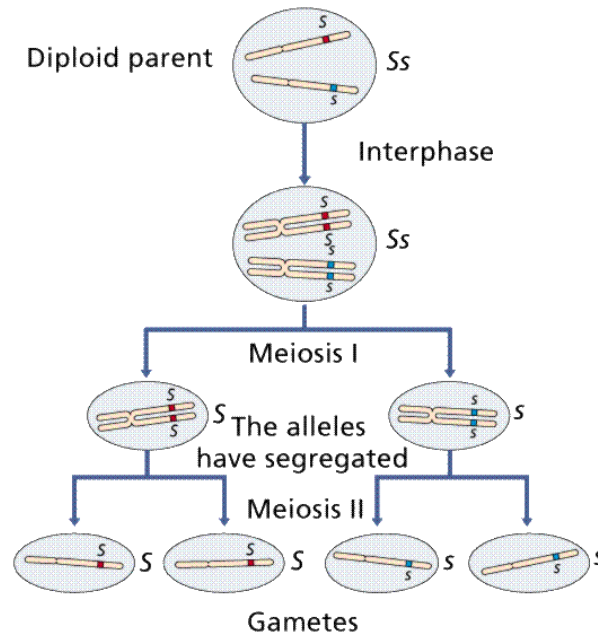


Inheritance of two alleles, S and s, in peas. Image from Purves et al., *Life: The Science of Biology*, 4th Edition, by Sinauer Associates (www.sinauer.com) and WH Freeman (www.whfreeman.com), used with permission.



Punnett square explaining the behavior of the S and s alleles. Image from Purves et al., *Life: The Science of Biology*, 4th Edition, by Sinauer Associates (www.sinauer.com) and WH Freeman (www.whfreeman.com), used with permission.

Meiosis, a process unknown in Mendel's day, explains how the traits are inherited.



The inheritance of the S and s alleles explained in light of meiosis. Image from Purves et al., *Life: The Science of Biology*, 4th Edition, by Sinauer Associates (www.sinauer.com) and WH Freeman (www.whfreeman.com), used with permission.

Mendel studied seven traits that appeared in two discrete forms, rather than continuous characters that are often difficult to distinguish. When "true-breeding" tall plants were crossed with "true-breeding" short plants, all of the offspring were tall plants. The parents in the cross were the P1 generation, and the offspring represented the F1 generation. The trait referred to as tall was considered dominant, while short was recessive. Dominant traits were defined by Mendel as those which appeared in the F1 generation in crosses between true-breeding strains. Recessives were those which "skipped" a generation, being expressed only when the dominant trait is absent. Mendel's plants exhibited complete dominance, in which the phenotypic expression of alleles was either dominant or recessive, not "in between".

When members of the F1 generation were crossed, Mendel recovered mostly tall offspring, with some short ones also occurring. Upon statistically analyzing the F2 generation, Mendel determined the ratio of tall to short plants was approximately 3:1. Short plants have skipped the F1 generation, and show up in the F2 and succeeding generations. Mendel concluded that the traits under study were governed by discrete (separable) factors. The factors were inherited in pairs, with each generation having a pair of trait factors. We now refer to these trait factors as alleles. Having traits inherited in pairs allows for the observed phenomena of traits "skipping" generations.

Summary of Mendel's Results:

1. The F1 offspring showed only one of the two parental traits, and always the same trait.
2. Results were always the same regardless of which parent donated the pollen (was male).
3. The trait not shown in the F1 reappeared in the F2 in about 25% of the offspring.
4. Traits remained unchanged when passed to offspring: they did not blend in any offspring but behaved as separate units.
5. Reciprocal crosses showed each parent made an equal contribution to the offspring.

Mendel's Conclusions:

1. Evidence indicated factors could be hidden or unexpressed, these are the recessive traits.
2. The term phenotype refers to the outward appearance of a trait, while the term genotype is used for the genetic makeup of an organism.
3. Male and female contributed equally to the offsprings' genetic makeup: therefore the number of traits was probably two (the simplest solution).
4. Upper case letters are traditionally used to denote dominant traits, lower case letters for recessives.

Mendel reasoned that factors must segregate from each other during gamete formation (remember, meiosis was not yet known!) to retain the number of traits at 2. The Principle of Segregation proposes the separation of paired factors during gamete formation, with each gamete receiving one or the other factor. Diploid organisms carry two alleles for every trait. These traits (on homologous chromosomes) separate during the formation of gametes.

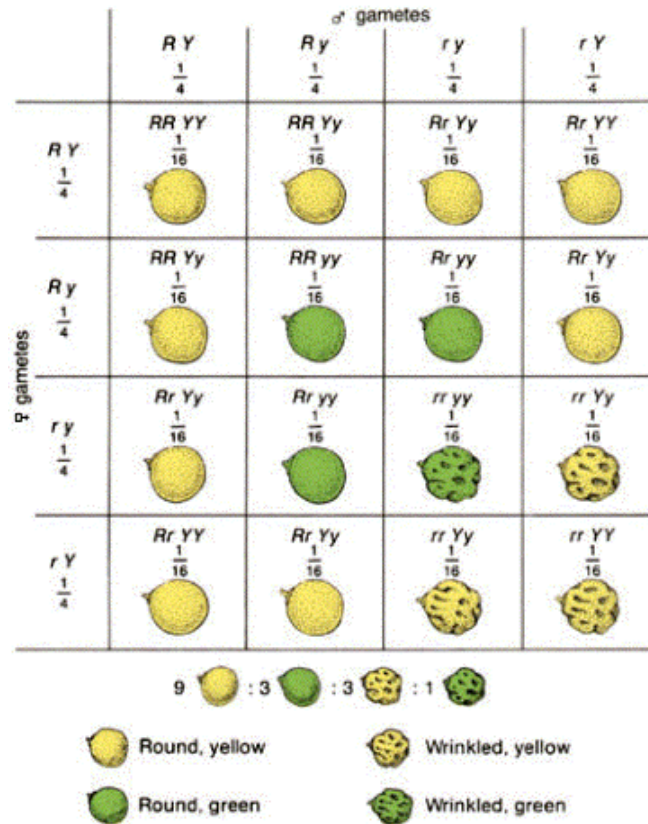
Dihybrid Crosses

When Mendel considered two traits per cross (dihybrid, as opposed to single-trait-crosses, monohybrid), The resulting (F2) generation did not have 3:1 dominant:recessive phenotype ratios. The two traits, if considered to inherit independently, fit into the principle of segregation. Instead of 4 possible genotypes from a monohybrid cross, dihybrid crosses have as many as 16 possible genotypes.

Mendel realized the need to conduct his experiments on more complex situations. He performed experiments tracking two seed traits: shape and color. A cross concerning two traits is known as a dihybrid cross.

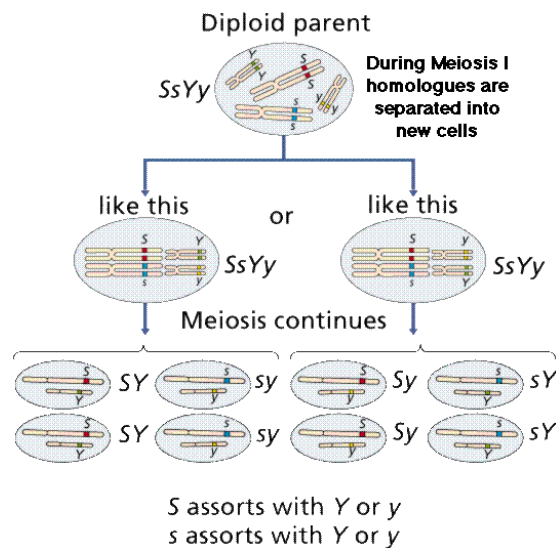
Crosses With Two Traits:

- The characteristic of Seed Roundness (R,r) can be either Smooth and dominant (R) or wrinkled and recessive (r).
- Yellow seed color (Y) is dominant over green (y).



Inheritance of two traits simultaneously, a dihybrid cross. The above graphic is from the Genetics pages at McGill University (<http://www.mcgill.ca/nrs/dihyb2.gif>).

Again, meiosis helps us understand the behavior of alleles.



The inheritance of two traits on different chromosomes can be explained by meiosis. Image from Purves et al., *Life: The Science of Biology*, 4th Edition, by Sinauer Associates (www.sinauer.com) and WH Freeman (www.whfreeman.com), used with permission.

Mendel started with true-breeding plants that had smooth, yellow seeds and crossed them with true-breeding plants having green, wrinkled seeds. All seeds in the F₁ had smooth yellow seeds. The F₂ plants self-fertilized, and produced four phenotypes:

315 smooth yellow

108 smooth green

101 wrinkled yellow

32 wrinkled green

Mendel analyzed each trait for separate inheritance as if the other trait were not present. The 3:1 ratio was seen separately and was in accordance with the Principle of Segregation. The segregation of S and s alleles must have happened independently of the segregation of Y and y alleles. The chance of any gamete having a Y is 1/2; the chance of any one gamete having a S is 1/2. The chance of a gamete having both Y and S is the product of their individual chances (or $1/2 \times 1/2 = 1/4$). The chance of two gametes forming any given genotype is $1/4 \times 1/4$ (remember, the product of their individual chances). Thus, the Punnett Square has 16 boxes. Since there are more possible combinations to produce a smooth yellow phenotype (SSYY, SsYy, SsYY, and SSYy), that phenotype is more common in the F₂.

From the results of the second experiment, Mendel formulated the Principle of Independent Assortment -- that when gametes are formed, alleles assort independently. If traits assort independent of each other during gamete formation, the results of the dihybrid cross can make sense. Since Mendel's time, scientists have discovered chromosomes and DNA. We now interpret the Principle of Independent Assortment as alleles of genes on different chromosomes are inherited independently during the formation of gametes. This was not known to Mendel.

Punnett squares deal only with probability of a genotype showing up in the next generation. Usually if enough offspring are produced, Mendelian ratios will also be produced.

Step 1 – Determination of parental genotypes (from phenotypic descriptions).

Step 2 - Determination of all possible gametes produced by parents.

Step 3 - Construction of the square with parental gametes shown on adjacent sides.

Step 4 - Recombination of alleles into each small square.

Step 5 - Determination of Genotype and Phenotype ratios in the next generation.

While answering genetics problems, there are certain forms and protocols that will make unintelligible problems easier to do. The term "true-breeding strain" is a code for homozygous. Dominant alleles are those that show up in the next generation in crosses between two different "true-breeding strains". The key to any genetics problem is the recessive phenotype (more properly the phenotype that represents the recessive genotype). It is that organism whose genotype can be determined by examination of the phenotype. Usually homozygous dominant and heterozygous individuals have identical phenotypes (although their genotypes are different). This becomes even more important in dihybrid crosses.

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PART A: Mendelian Genetics Problems

The following problems are designed to give you practice with genetic inheritance and the associated terminology. For most problems, you should draw out your response including writing the genotype of the parents and a Punnett square to show inheritance. For dominant and recessive traits the dominant trait is described using a capital letter and the recessive trait is described using the lower case version of the same letter (Rr).

When describing inheritance remember the parent has two alleles of the same trait (diploid) but the gametes will only have one of those two traits (haploid). On a Punnett square you are drawing the possible gametes a parent could produce.

Labeling: Develop the habit of labeling everything: who's the mom, who's healthy and who is sick, who is a boy or girl, what does the capital letter signify, etc. etc.

Sample Punnett Square – monohybrid cross

Mother - Aa X Father - AA

	(Eggs) A	or	a
(Sperm) A	AA		Aa
or			
A	AA		Aa

Dominant/recessive inheritance

1. The seeds of the pea plant Mendel used are either yellow or green. The allele for yellow is dominant and the allele for green is recessive. What would the possible genotypes of the offspring be if a plant heterozygous for seed color is crossed with a plant having a homozygous dominant allele combination? (Y = yellow y=green)

2. Mendel also studied pea plant height, which demonstrates dominant and recessive inheritance of traits (T = tall t=dwarf). He had both tall and short pea plants. He conducted multiple crosses of plants of unknown genotype (i.e. one tall and one short, two tall, two short) =F1 generation, and further crossed the offspring of initial crosses by crossing F1 offspring with siblings =F2 generation. By the second offspring generation of every cross except the one descending from the cross between two short plants had dwarf plants and tall plants. The F2 generation from the two short cross contained all short plants. He knew his dwarf plants were homozygous. How did he know this (or how do you know this)?

He then wanted to determine the genotypes of the tall plants in this second generation. He crossed each of the tall plants with a dwarf plant to find his answer. Why would this work?

3. Mendel also compared multiple traits on pea plants. Assuming that seed shape and petal color traits are found on different chromosomes and are dominant and recessive traits, show the possible offspring for a plant that is heterozygous for seed shape and petal color crossed with a plant that is heterozygous for seed shape and homozygous recessive for petal color. List both the offspring genotype and phenotype. (P=purple p=white, R=round r=wrinkled)
To help you get started, answer the following 3 questions:

Heterozygous for seed shape (use the letters for the alleles) = _____

Homozygous recessive for petal color = _____

A plant heterozygous for shape & heterozygous for color has these 4 letters: _____

A plant heterozygous for shape & homozygous recessive for color has these 4 letters: _____

Now make a Punnett square similar to the dihybrid crosses from the above two genotypes.

Genetic Disorders

4. Autosomal recessive: Cystic fibrosis is caused by a recessive trait (c) on chromosome 7. A cell membrane protein is not effective at ion transport in the lungs leading to the development of thick mucus. To exhibit the disease, an individual must be homozygous recessive for the trait. If two parents who do not have cystic fibrosis have a child who does and a child who does not, what can you say about each person's genotype in the family?

5. Autosomal dominant: Huntington's disease is caused by a dominant allele (H) and leads to nervous system problems that develop after middle age. A heterozygous woman develops Huntington's in her late forties after she has had children. Her husband does not have Huntington's. Write two **workable possibilities** (there are several) for the genotypes for the woman's parents.

Woman's mother

Woman's father

Possibility 1:

Possibility 2:

What are the phenotypes of both parents in both possibilities above?

What is the possibility of this woman's children developing Huntington's? (Make a Punnett square to show your answer).

Incomplete dominance: This is a situation in which a heterozygous genotype produces a phenotype that is intermediate between two extremes.

6. Among Caucasians, straight or curly hair, are the two extremes and wavy is the intermediate phenotype. a) If two people with wavy hair have children, show what all the possible genotypes of the children could be using a Punnett square. b) Then state the phenotype ratio of the children. Use C for curly, s for straight and Cs for wavy.

Codominance

Human blood type is a codominant trait where more than 2 allele types are present in the population (ABO). If a person receives alleles for A or B they will be expressed (if both are received they will have the blood type AB). Type O is a recessive blood type when neither A or B is received. If a person receives an O allele along with an A or B allele they will have the blood type A or B. Use I^A for A blood. I^B for B and i for O blood.

7. If a heterozygous man with type B blood mates with a woman with type O blood, what are their genotypes and what are the possible genotypes and phenotypes of their offspring. (Can you fully determine all of the genotypes?)

8. How can blood type help determine who the parents are for a given child? Give an example where blood type would determine the father of a child.

X linked inheritance (aka sex linked)

9. Hemophilia is an X linked (on the X chromosome) recessive disorder that occurs when a blood clotting protein is mutated. James has hemophilia but his parents do not. What is James' genotype and what are the genotypes of his parents? Draw a Punnett square that shows the likelihood of James siblings having the disorder. X^h is hemophilia; X^H is healthy. A healthy man would be $X^H Y$.

10. Red/green colorblindness is an X linked recessive trait. If a man and woman have a daughter who is colorblind, what is the genotype and phenotype of each of the parents? Assume mom is not colorblind. What is the probability that her brother will be colorblind?