BIOL 300 – Foundations of Biology
Summer 2017 – Telleen
Lecture Outline

Mendelian Genetics I: Mendel's First Law: The Law of Segregation

I. Gregor Mendel: The person

A. Mendel was an Austrian monk
B. He lived from 1822-1884
C. He was the first experimental scientist to discover the rules governing the inheritance of genetic traits from one generation to the next
D. He performed his famous experiments between 1843 and 1868, decades before chromosomes were discovered and a century before DNA was shown to be the genetic material
E. He was a contemporary of Charles Darwin and Alfred Russell Wallace (who independently came up with the idea of evolution by natural selection), but his work was unknown to them at the time and vice versa.

II. Mendel's Experiments

A. Mendel worked with pea plants to discover how genetic information is passed from parent to offspring
B. By carefully designed crosses (matings), he was able to deduce how traits are inherited generally (i.e. in most organisms) by following the inheritance patterns of several morphological characteristics through three generations in the garden pea (*Pisum sativum*)
C. The characteristics, or traits, he chose were each under the control of a single gene which had only two alternative forms (alleles)
D. Mendel's choice of traits was critical to his success. By choosing traits inherited in a very simple manner, he did not encounter obstacles in his research that arise when traits are under the control of multiple genes (polygenic traits) or under the control of many genes and the environment (multifactorial traits), which we'll talk about later.

III. Mendel's Laws reflect chromosome behavior in Meiosis

A. Mendel discovered two laws:
   1. **Law of Segregation (Mendel's 1st Law)**
   2. **Law of Independent Assortment (Mendel's 2nd Law)**
B. Although Mendel did not know it at the time, the laws are really describing the behavior of alleles (on chromosomes) as they move through the process of Meiosis
C. Understanding the behavior of chromosomes during Meiosis and how chromosome behavior dictates the behavior of alleles that lie along them is critical to understanding Mendel's Laws because *they are the same thing!*

IV. Mendel's Law of Segregation and the Monohybrid Cross

A. Mendel demonstrated segregation of alleles by performing a series of genetic matings between pea plants that he called monohybrid crosses
B. This type of cross begins with the mating of two homozygous plants, each of which expresses a different form of the same trait.
C. For example, YY (Yellow seeds) x yy (green seeds)

D. Although the term “homozygous” was not used in Mendel’s time, he was able to acquire seeds of “pure-breeding” plants. When such plants are selfed (fertilized by themselves, pea plants can self-fertilize), they always produce offspring with the same form of a particular trait as the parent. They do so because they only have one type of allele for that trait. Thus homozygous and pure-breeding plants are the same thing (for a particular trait).
E. When Mendel mated two homozygous parent plants (**P1**), he found that they always produced a generation of offspring (the **F1** generation) which all look like one of the two parents, but not both
F. For example, a cross between a pure-breeding (homozygous) smooth-seeded plant and a pure-breeding (homozygous) wrinkled-seeded plant always produced and **F1** generation with smooth seeds
G. Mendel called the form of the trait that was seen in the **F1**s dominant and he called the other form recessive. In the example above, smooth seeds are dominant over the wrinkled (recessive) form.
H. Mendel then self-fertilized the **F1** plants to produce an **F2** generation
I. To his surprise, the recessive (wrinkled) form of the trait reappeared among the **F2** plants!
J. In fact, he found that they always comprised about one-fourth of all the **F2** plants
K. Note that Mendel produced large numbers of offspring (i.e. large sample sizes) to calculate the **3:1 ratio** in the **F2**s. Large sample sizes allow the results to be obtained with statistical significance and are highly trustworthy.
L. Mendel repeated the monohybrid cross for each of the seven traits and got very similar results. He then generated a model to explain the results
M. The model is known as the **Law of Segregation**, which has three parts:
    1. **Alternative versions of factors (genes) account for the variations in inherited characters**. This is the concept of alleles. Alleles are different versions of genes that impart the same characteristic. Each pea plant has a gene that controls flower color, but some carry an allele for purple, while others carry an allele for white.
    2. **For each character, an organism inherits two genes, one from each parent**. This means that when somatic cells are produced from two gametes, one allele comes from the mother and one allele comes from the father. These alleles may be the same (homozygotes, e.g. WW or ww) or different (heterozygotes, e.g. Ww)
    3. **If the two alleles differ (e.g. Ww) then one, the dominant allele, is fully expressed while the other, recessive allele is not. However, the recessive allele segregates from the dominant allele during gamete production and is passed on unchanged to the heterozygote’s gametes.** The two alleles of the organism are separated into different gametes during gamete production (Meiosis), and the heterozygote therefore produces equal numbers of each type of gamete (e.g. W and w)

Mendelian Genetics II: The Dihybrid cross and the Law of Independent Assortment

I. The Dihybrid Cross

A. After completing his monohybrid cross, Mendel extended his experiments by performing a series of dihybrid crosses.

B. **Dihybrid crosses** are a mating between two pure-breeding (homozygous) parents, but this time the hereditary behavior of two separate traits are followed rather than just one.

C. As in his previous experiments, he crosses the homozygous parents (P_1 generation) to produce an F_1 generation, and then crossed the F_1's to one another to produce an F_2 generation.

D. Consider this example:

P_1 – Smooth, Yellow x wrinkled gree

To produce:

F_1 – All Smooth, Yellow

Which are crossed to each other:

F_1 x F_1 – Smooth, Yellow x Smooth, Yellow

To produce:

F_2 – 9/16 Smooth, Yellow (both Dominant traits)
3/16 Smooth, green (Dominant trait 1, recessive trait 2)
3/16 wrinkled, Yellow (recessive trait 1, Dominant trait 2)
1/16 wrinkled, green (both recessive traits)

E. The experiment above has several notable features:

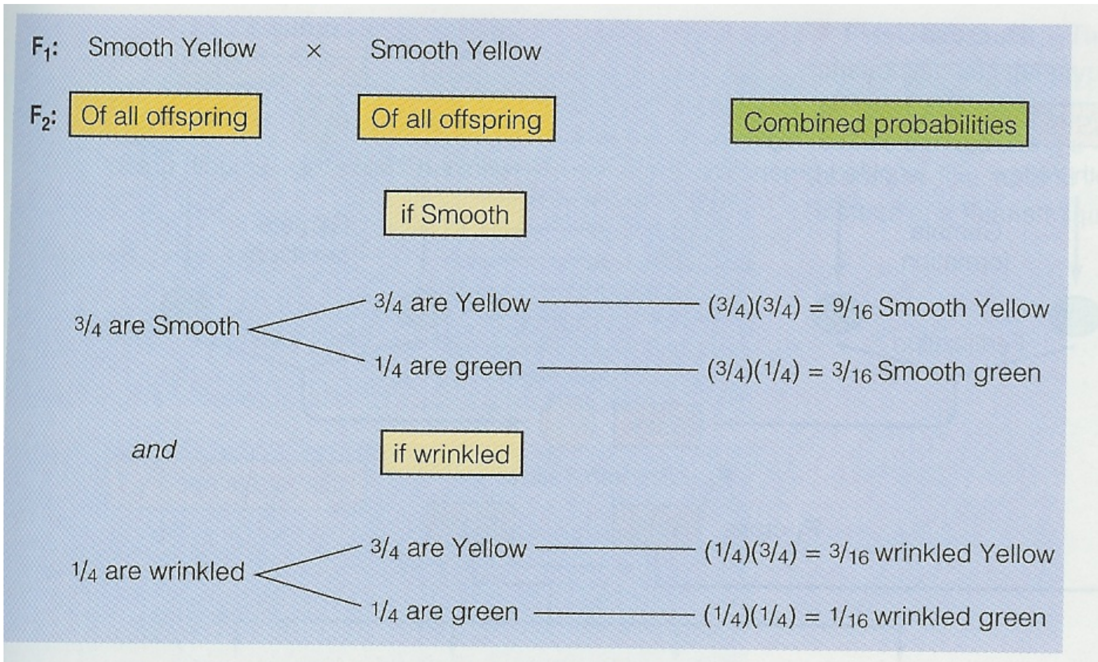
1. The F_1 generation all expressed both of the dominant traits, as expected for each trait individually.
2. The F_2 progeny always fell neatly into one of four phenotypic classes: Dominant, Dominant; Dominant, recessive; recessive, Dominant; recessive, recessive
3. If we look at each gene separately, we still get the expected 3:1 ratio e.g. Consider only seed color in the F_2 generation:

   Yellow (both Smooth and wrinkled together): 3/16+9/16 = 12/16 = ¾ or 75% expressed the Dominant trait, while only ¼ (3/16 + 1/16 = 4/16 = ¼) show the recessive trait.

F. Mendel repeated this experiment for all possible pairs of the seven traits he had originally selected. In each case (except one, which we'll discuss later) he got the same results: a 9:3:3:1 phenotypic ratio among the F_2 progeny.

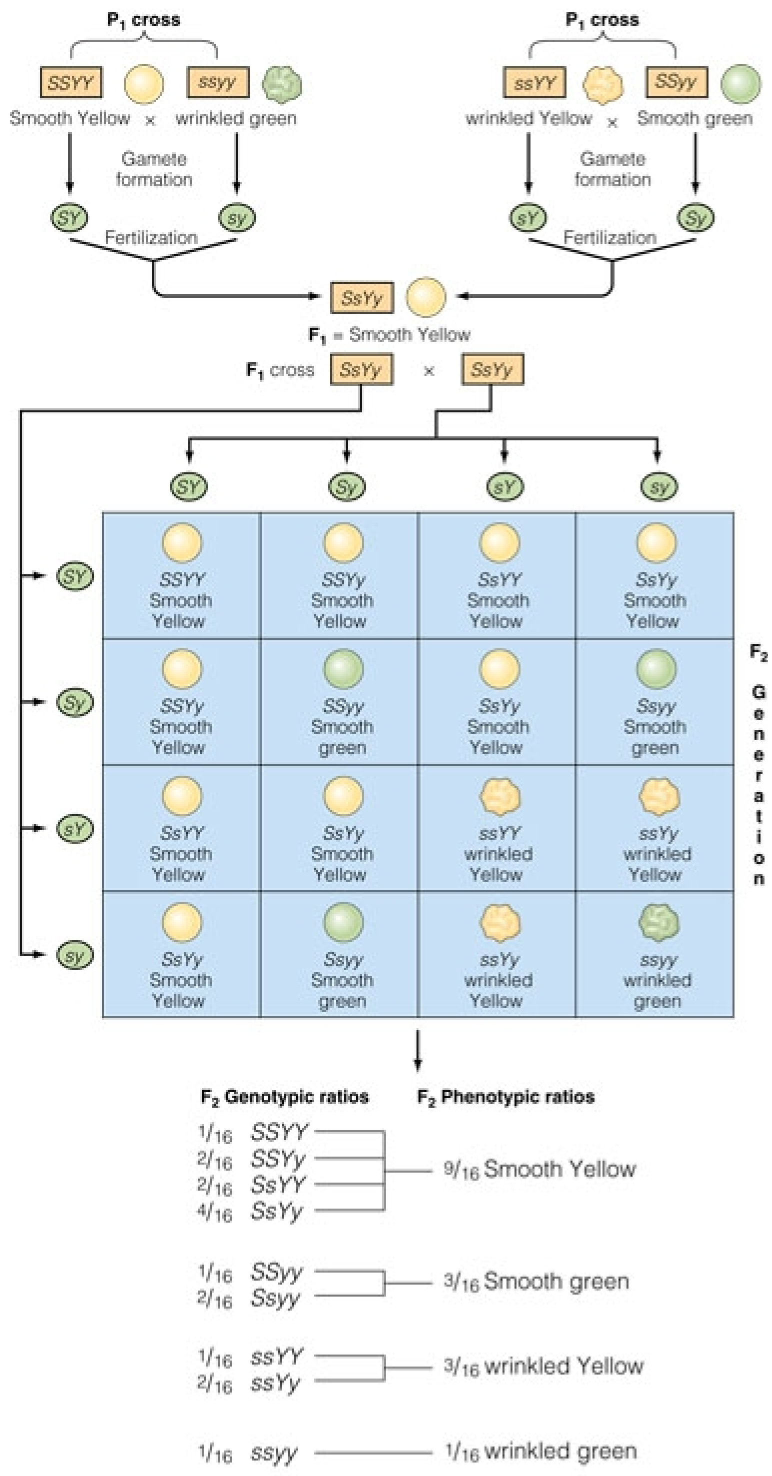
G. Mendel then used his Law of Segregation to build a model explaining the results of the dihybrid crosses:

1. The alleles for each trait should be visible in the F_2's in a 3:1 ratio, if the trait is considered alone.
2. If this is true and assuming the two traits being observed segregate independently during gamete production (i.e. the segregation of the alleles at the first locus does not affect or influence the segregation of the alleles at the second locus), the following prediction can be made:



H. This can also be described using letters to represent the alleles as we did for the monohybrid cross, which would give us F_1 plants with the genotype SsYy, which would produce the following gametes in equal numbers: SY, sy, Sy, sY. From this, it logically follows that we would end up with a 9:3:3:1 ratio of phenotypes in the F_2 (see the diagram above and the Punnett Square on the next page).

II. The Punnett Square for the Dihybrid Cross:

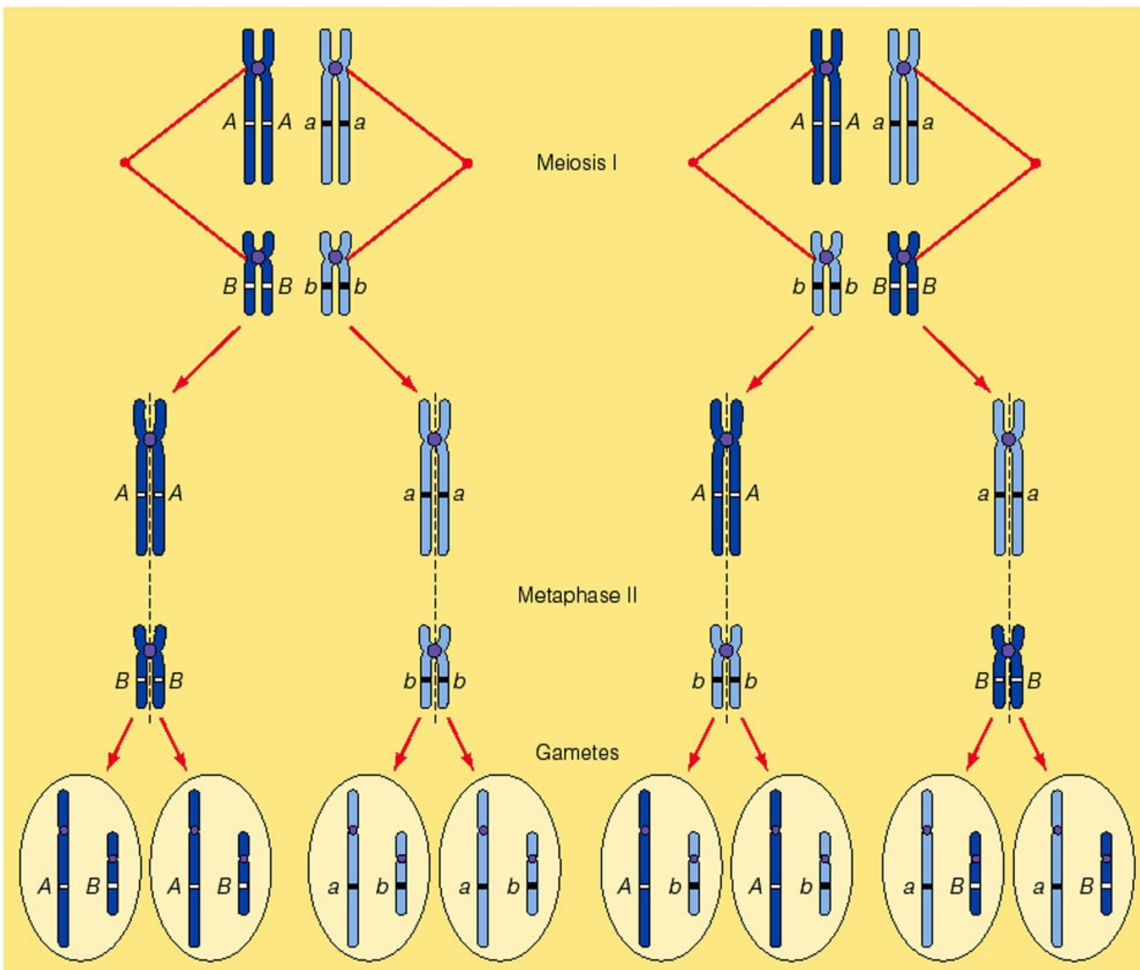


III. The Law of Independent Assortment

A. From these observations, Mendel derived his Law of Independent Assortment:

1. During fertilization, the alleles of one gene pair segregate independently of the alleles belonging to other gene pairs
2. Therefore, an individual who is heterozygous for two different genes (e.g, AaBb) will produce gametes containing all possible combinations of the four alleles in equal numbers (¼ AB, ¼ ab, ¼ Ab, ¼ aB).

B. Bringing it back to Meiosis. Let’s imagine that genes A and B are on different chromosomes and follow the alleles of a heterozygote (AaBb) through Meiosis. Notice how the way the homologous pairs of chromosomes line up during Metaphase I influences the possible gametes formed (left side vs. right side). Each of the two possibilities shown below occurs in 50% of the cells undergoing Meiosis.



IV. Putting it all together:

A. The Law of Segregation simply states what we already learned studying Meiosis:
    1. Homologs segregate into different cells during the reductional division ($2n \rightarrow n$) of Meiosis I, and the sister chromatids segregate during Meiosis II. As a result, each gamete receives only one allele for each gene.
    2. This law only concerns the behavior of the alleles at a single gene locus.

B. The Law of Independent Assortment states that the homolog pairs line up on the metaphase plate during Meiosis I independently of one another. Thus, a heterozygote for two loci will produce gametes in equal proportions containing each of the four possible combinations of alleles.
    1. This law addresses the behavior of the alleles of two gene loci considered together
    2. This behavior can only be observed in a double heterozygote (because the alleles can be followed through the cross), but it also occurs when an individual is not heterozygous at both loci (we just can't follow it).