Chapter for today: Chap. XI

Major points for the day:

1. Dominant & recessive traits
2. 3:1 segregation in the second generation of a monohybrid cross
3. 9:3:3:1 segregation in the second generation of a dihybrid cross

Introduction

Today we will begin to look at the ways that this process affects distribution of traits by looking at the genetic outcome of meiosis as first studied by Gregor Mendel. The science which deals with this issue is called **genetics**.

- The term "genetics" was introduced in 1905 by William Bateson who was one of the rediscoverers of Mendel's work (he was the first to show that Mendel's concepts could be applied to an animal—chickens).

Mendel's experiments with peas (*Pisum sativum*) provided the basis for genetics

Mendel was a monk in a monastery in Brünn (now Brno) in what is now the Czech Republic but was then, in the mid–19th century part of Austria.

He became interested in the nature of inheritance, and performed experiments with the system which was easiest for him to use, the garden pea.

These experiments were eventually presented to scientific societies and published in 1866.

- Though well-educated at the University of Vienna, Mendel was not a member of the scientific elite of his day. It is likely that the elite were not able to fully appreciate the work
- Though his work was known by many, it was not really appreciated for several decades, and were rediscovered by three scientists who unwittingly reproduced Mendel's work, confirming his conclusions. Bateson
- William Bateson, who was a professor at Cambridge University, popularized the term "genetics" for the science that Mendel had created.though they were aware of his work through its publication

Mendel did experiments in what was termed "hybridization" at the time

- He studied what happened when "true breeding" plants were crossed to each other
- True breeding plants are plants that always produce offspring that look the same, e.g. plants with yellow peas producing more plants with yellow peas

People thought at the time that hereditary information from each parent were mixed with each other in their offspring

Mendel's leap was to imagine that heredity consisted of units ("characters")

- Hereditary units could be associated with particular observable traits (e.g, those yellow peas)
- Each parent contributed one unit to the offspring, so there were two of each—one maternal and one paternal

<u>How did Mendel come to this hypothesis?</u>

It is hard to know since the abbot that succeeded Mendel was a lifelong rival who opposed Mendel's scientific experiments

The new abbot destroyed all of Mendel's notebooks on his death!

One conjecture is that Mendel noted that there was one obvious trait that had an “either / or” nature: gender

- Offspring of animals, including humans, are either male or female
- They don’t show a “mixture” of sexual characteristics
- In large populations of animals the ratio of male to female is 1:1
- This suggests a simple model of sex determination in which one of the sexes carries a single unit which determines maleness (or femaleness)
- A child receiving that unit was that gender, one not receiving it was the other

Mendel wanted to demonstrate the existence of such a unit. His work didn’t directly involve gender since pea plants do not have a gender (their flowers have both male and female structures)

Instead, he studied other observable traits:

Trait	Types	
Flower color	Purple	White
Stem length (plant height)	Tall	Dwarf
Flower position	Along stem	At tip
Pod color	Green	Yellow
Pod shape	Inflated	Wrinkled
Pea color	Yellow	Green
Pea shape	Round	Wrinkled

In each case, Mendel was able to create a true-breeding strain in which all individuals had a the same feature, showing that the appearance of the individual was hereditary; Tall breed Tall and Dwarf breed Dwarf.

- The alternative would have been, like humans, for there to be a variety of heights ranging from very tall to very short, but we will come back to look at that idea later

What Mendel was interested in was how these traits interacted with each other when they were introduced into new plants by what he called "hybridization", but what we would now call a genetic cross.

Would the progeny of a Tall and a Dwarf plant be divided equally among Tall and Dwarf progeny much as a cross of a male and a female animal leads to 50% male and 50% female? Or would there be a more complicated answer? It is a credit to Mendel that he was able to understand the more complicated answer that he received from these crosses.

With these simple observable traits he was able to demonstrate that genetic units existed, and that they were inherited in pairs, one from each parent.

Some genetic nomenclature

To discuss Mendel’s results it helps to use the genetic nomenclature (also largely invented by Bateson)

- **gene**: a unit of hereditary information; each is at a unique location on a chromosome, also called a locus
- **allele**: genes can come in various forms which carry distinct information—each distinct form is called an allele.
  - A gene concerned with pea color might specify yellow versus green
  - A particular form of the gene specifying green would be called an allele

  - There can be multiple forms which have give the same observable trait; each is a unique allele
- **phenotype** vs. **genotype**: the observable effect of an allele is its phenotype; genotype is just the nature of the genes carried by an individual (for example, identifying what alleles he carries)
- **homozygous** vs. **heterozygous**: since each individual carries two of these alleles, they can either be identical (homozygous) or different (heterozygous)
  - "homo" means same while "hetero" means different
- **dominant** vs. **recessive**: if an individual carries two alleles with different phenotypes (e.g., yellow peas versus green peas) he can not express both of them—they are mutually exclusive
  - most of the time one is expressed to the exlusion of the other; the one whose phenotype is expressed is dominant (e.g., when a yellow and green allele are present the peas appear yellow—yellow is dominant)
  - the allele whose phenotype is not expressed is recessive (green is recessive)

In addition to these genetic terms there are some common genetic symbols:

- Dominant and recessive genes are represented by uppercase and lowercase letters, respectively
  - The dominant allele might be referred to as "***A***" while the recessive is symbolized by an "***a***"
  - A dominant homozygote would be ***AA***, a recessive homozygote is aa; the heterozygote is ***Aa***
  - A second gene could be symbolized as ***B*** and ***b***
- To track inheritance the generations of a genetic cross are termed:

  P parental generation
  F_1 first–generation offspring
  F_2 second–generation offspring

Mendel's test of his theory

Mendel wanted to demonstrate experimentally that his theory of inheritance was correct. We are going to look at the way he set up that experiment and what his results were.

- In doing this we will use the nomenclature created and popularized by the people who rediscovered Mendel's work
- If you were to look at Mendel's original papers you would be hard pressed to understand it; the trick to gaining scientific acceptance is often in the way that the work is presented as much as in the data themselves!

The central idea that Mendel started from was that a single genetic unit could govern an observable trait. An observable trait should then appear in offspring of the original plant in predictable ways.

For example, let's look at a cross Mendel did between a wild type plant with the normal Purple flowers and another plant with White flowers

- A cross involving alternative alleles of one gene is termed a **monohybrid cross**.
- Crosses of the Purple flower plant to themselves always gave Purple flowered plants and the White flowered plants always produced White.
- But in the cross all of the plants were Purple.
- Any experiment should be seen as testing alternative hypotheses. In this case, Mendel is contrasting his theory of discrete genetic determinants against a "blending" hypothesis. That alternative hypothesis might have predicted that all

of the plants in the cross would have been pale Purple. A third hypothesis might have predicted that half of the F1 plants would have been Purple, and half White.

- The fact that all of the plants were Purple suggests this phenotype is in some sense "stronger" than White. In genetic terms we call Purple "dominant".

Remember that the plants are diploid, so they carry one chromosome from each parent. If we call the Purple plants ***AA*** (the "A" gene specifying flower color) and the White plants as ***aa*** (double recessive). The progeny plants, having gotten one copy of the gene from each parent are by definition ***Aa*** (one copy of each gene).

- The nomenclature is designed to show that there are two types of the gene present, the dominant ***A*** (Purple) and the recessive ***a*** (White).

Mendel knew that some Purple flowered plants, when crossed to each other, could produce many white progeny. The recessive ***a*** gene could "go into hiding" and reveal itself in later plants. This is akin to red-haired children who can appear in alternating generations in humans (as in my own family).

Mendel's theory predicted that crossing two Purple flowered plants from the F1 generation should produce a very specific ratio of Purple to White flowered plants. We can demonstrate this prediction with something called a Punnett Square, another device invented far after Mendel's time:

	A	a
A	AA	Aa
a	Aa	aa

- The ***Aa*** plants should produce equal numbers of ***A*** and ***a*** bearing gametes, which when fused produce a 3:1 ratio of Purple to White plants.

Mendel’s first law

Experiments of this type form the basis for Mendel’s first law, the Principle of Independent Segregation.

This “law” describes how genetic characters (genes) behave during meiosis.

The Principle of Independent Segregation states that during meiosis pairs of alleles separate from each other.

In the example, a plant carrying both the ***A*** and the ***a*** allele generates two types of gametes in equal numbers, one type carrying ***A*** and the other ***a***.

This principle held true for alleles at all of the seven loci tested by Mendel.

The dihybrid cross

Given that alleles of a gene segregate from each other during meiosis the next obvious question is how alleles of multiple genes behave in meiosis. We can again propose some alternative hypotheses to describe how the genes might behave.

A cross involving alternative alleles at two genes is termed a dihybrid cross. This is the cross that Mendel used to test how multiple genes assort.

Mendel believed that the genetic characters would assort independently, that is that gametes had a random chance to receive any of the alleles present. A plant that was heterozygous for flower color (Purple vs. White) and for height (Tall vs. Dwarf) gametes carrying all possible combinations of color and height genes: Purple Tall, Purple Dwarf, White Tall and White Dwarf.

An alternative hypothesis might have said that characters from one parent would tend to be inherited together, as would those of the other parent.

- Perhaps Purple and Tall originated from one true breeding parent prior to hybridization, and White and Dwarf from another.
- Under this hypothesis one would expect only to see Purple and Tall alleles assorting together, and only White and Dwarf.

Of course, the blending hypothesis would predict a range of types between Purple and White and between Tall and Dwarf, though that hypothesis seems unlikely in light of the first experiment.

Again a more complex Punnet Square can demonstrate what Mendel observed.

- Let ***A*** and ***a*** stand for Purple and White again, and ***B*** and ***b*** stand for Tall and Dwarf.
- As before, just as Purple by White (***AA*** x ***aa***) produced Purple (***Aa***) progeny, Tall by Dwarf (***BB*** x ***bb***) produced only Tall plants (***Bb***). So it appeared that Tall like Purple was a dominant allele.
- Therefore, the F1 generation all the progeny of the cross **AA BB** x ***aa bb*** were ***Aa Bb*** and appear Tall with Purple flowers.
- A cross among these F1 progeny would test how alleles of multiple genes assort during meiosis. The Punnett Square predicts the result if the alleles of the two genes assort entirely randomly:

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

- The Punnett Square predicts a 9:3:3:1 ratio of Purple Tall, Purple Dwarf, White Tall and White Dwarf plants and this is the ratio that Mendel observed in multiple dihybrid crosses.

Mendel's second law

The results of this experiment showed that the genes for flower color and plant height assort entirely independently of each other.

Mendel's second law states that during a meiosis involving alternative alleles of multiple genes the distinct gene pairs (e.g. ***Aa*** and ***Bb***) assort entirely independently of each other.

If the genes segregated together to any degree (e.g., ***A*** tending to assort with ***B*** and ***a*** with ***b*** then the ratio of progeny plants seen in the dihybrid cross would diverge from the 9:3:3:1 ratio

Question for next time: What assumptions about inheritance do Mendel's experiments make?