Patterns of Inheritance
Gregor Mendel

- Austrian monk, gardener, scientist
- First acknowledged to study heredity – the passing on of characteristics from parents to offspring
- Traits – characteristics that are inherited
- Father of genetics – the branch of biology that studies heredity
The Peas

- Mendel chose garden peas
- They reproduce sexually, so they have both male and female sex cells, called gametes
- Pollen and egg unite in a process called fertilization
- Fertilization results in a fertilized cell, called a zygote, that develops into a seed
Pollination is the transfer of pollen grains from a male reproductive organ to a female productive organ. Both male and female organs are close together in the same pea flower. As a result, peas normally self-pollinate. This is what Mendel wanted in most cases. Mendel also removed the male organ and dusted pollen on flower of another plant. Called cross-pollination.
Mendel’s peas had been self-pollinating for a long time. This meant that the tall ones had been tall for a long time and the short ones had been short for generations. Called purebreds.
Mendel’s Monohybrid Crosses

- Mendel performed cross-pollination with a tall pea plant (6 foot purebred) and a short pea plant (2 foot purebred) – these are the parental generation (P₁)
- **Hybrid** – offspring of parents that have different forms of a trait
- All of the offspring grew as tall as the tall parent – first filial generation (F₁)
  - The “short trait” seemed to disappear
Monohybrid Crosses

- Mendel let his F1 plants self pollinate
  - Second filial generation ($F_2$)
- He counted over 1000 plants
  - About 75% were tall
  - About 25% were short
  - Ratio of 3:1
Mendel did these same types of crosses for 7 traits:
- Seed shape: round vs. wrinkled
- Seed color: yellow vs. green
- Flower color: purple vs. white
- Flower position: axial vs. terminal
- Pod color: green vs. yellow
- Pod shape: inflated vs. constricted
- Plant height: tall vs. short

### Table of Traits

<table>
<thead>
<tr>
<th>Character</th>
<th>Dominant trait</th>
<th>Recessive trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>Seed shape</td>
<td>Sphercial</td>
<td>Wrinkled</td>
</tr>
<tr>
<td>Seed color</td>
<td>Yellow</td>
<td>Green</td>
</tr>
<tr>
<td>Flower color</td>
<td>Purple</td>
<td>White</td>
</tr>
<tr>
<td>Flower position</td>
<td>Axial</td>
<td>Terminal</td>
</tr>
<tr>
<td>Pod shape</td>
<td>Inflated</td>
<td>Constricted</td>
</tr>
<tr>
<td>Pod color</td>
<td>Green</td>
<td>Yellow</td>
</tr>
<tr>
<td>Stem height</td>
<td>Tall</td>
<td>Dwarf</td>
</tr>
</tbody>
</table>

The Rule of Unit Factors

- Mendel concluded that each organism has two factors that control each of its traits.
- We now know these factors are **genes** and that they are located on chromosomes.
- Alternate forms of genes are called **alleles**.
- Each of Mendel’s traits had two forms of genes – two alleles.
  - One comes from mother, one from father.
The Rule of Dominance

- Some alleles are **dominant** over **recessive** alleles
  - Dominant alleles cover recessive alleles
- In genetics, capital letters are used to express dominant alleles and lower case letters are used to express recessive alleles
  - Ex: “T” for tall allele, “t” for short allele
- So what will a “TT” plant look like?
  - A Tt plant?
  - A tt plant?
- Earlobes, Forelock, Dimples, Straight thumb, Bent Pinky, Mid-digit hair
Simple Dominant Heredity

- Cleft chin, widow’s peak, unattached earlobes, hitchhiker’s thumb (back more than 30 degrees), almond-shaped eyes, thick lips, mid-digital hair
The Law of Segregation

- Mendel’s first law of heredity
- Every individual has two alleles of each gene and when gametes are produced, each gamete receives one of these alleles
- So how did this work in Mendel’s $F_2$ generation?
Phenotypes

- Two organisms can look alike on the outside, but have different allele combinations
- **Phenotype** is the way an organism looks and behaves
  - ex: yellow seeds can be TT or Tt
Genotypes

- The allele combination an organism contains is known as its genotype
  - You can’t always see this because of dominance
  - TT and Tt are different genotypes

- An organism is homozygous for a trait if the two alleles for the trait are the same
  - Ex: TT or tt

- An organism is heterozygous for a trait if the two alleles for the trait are different
  - Ex: Tt
Mendel’s Dihybrid Crosses

Mendel crossed peas that differed from each other in two traits.

He crossed plants that were homozygous for round yellow seeds (RRYY) with plants that were homozygous for green wrinkled seeds (rryy).

F₁ generation

All plants produced round, yellow seeds.

What was dominant?
The Second Generation (F₂)

- Let F₁ plants self-pollinate (were heterozygous for two traits)
- He got all four combinations in a certain ratio
  - Round, yellow – 9
  - Round, green – 3
  - Wrinkled, yellow – 3
  - Wrinkled, green – 1
The Law of Independent Assortment

- Mendel’s second law of heredity
- Genes for different traits are inherited independently of each other
  - Inheritance of one trait has no influence on another trait
- Instead of a ratio of 9:3:3:1, what would the dihybrid cross have looked like?
Punnett Squares

1905, Reginald Punnet, English biologist created a shorthand way of finding EXPECTED proportions of possible genotypes in the offspring of a cross.

Monohybrid crosses
See overhead

Dihybrid crosses
See overhead
Making a Pedigree

- A pedigree is a graphic representation of the genetic inheritance of ONE trait
  - Generally used to study historical inheritance – make inferences

- Symbols
  - Circle - female
  - Square - male
  - Shaded in - shows the trait
  - Half shaded in - is a carrier, heterozygous individual that does not show the trait
  - Each horizontal row is a generation - represented by roman numerals
  - Parents are connected horizontally
  - Children are connected to parents with a vertical line
Pedigree Example

- Pedigree a)
- Pedigree b)

Diagram Icons:
- Unaffected male
- Affected male
- Unaffected female
- Affected female
- Person whose sex is not known
- Vertical line = offspring (in this case, son)
- Marriage (mating)
- Consanguineous marriage
- A family of four brothers and sisters. The last two are non-identical twins
- Identical twins
Section Review

1. What structural features of pea plants made them suitable for Mendel’s genetic studies?

2. What are genotypes of a homozygous and a heterozygous tall pea plant?

3. One parent is homozygous tall and the other is heterozygous. How many offspring will be heterozygous?

4. How many different gametes can an RRYy parent form? What are they?

5. What is the law of segregation?

6. What is the law of independent assortment?

7. What is the rule of dominance?

8. In garden peas, the allele for yellow peas is dominant to the allele for green peas. Suppose you have a plant that produces yellow peas, but you don’t know whether it is homozygous dominant or heterozygous. What experiment could you do to find out?
1. What do these symbols represent in a pedigree: square, circle, unshaded circle, shaded square, horizontal line, vertical line?

2. Describe one trait that you inherited by simple dominance. Will you pass it on to your offspring?

3. Suppose that a child with unattached earlobes has a mother with attached earlobes. Can a man with attached earlobes be the child’s father?
Incomplete Dominance

The phenotype of heterozygous individuals is intermediate between those of the two homozygotes.

Snap Dragon
- Red Flowered (RR) x White Flowered (R’R’)
- All F₁ will be pink (RR’)
- What will the F₂ generation look like?
Codominance

- The phenotypes of both homozygotes is expressed in heterozygous individuals

- Chickens
  - Black-feathered (BB) x White-feathered (WW)
  - All F₁ will have both black feathers and white feathers (BW)
  - What will F₂ look like?
Sickle-Cell Disease

رز An example of codominance in humans
رز Common in African Americans and Americans with ancestry near the Mediterranean Sea
رز Homozygous – hemoglobin differs from normal by 1 amino acid
  رز Changes the shape of the red blood cells (rbc)
  رز Slow blood flow, block small vessels, and result in tissue damage and pain
Sickle-Cell Disease

- Heterozygous – produce both normal and sickle hemoglobin (codominance)
  - Enough that they don’t have major health problems
  - Show sickle-cell related disorders when oxygen isn’t readily available
Multiple Phenotypes from Multiple Alleles

- For many traits, though you only have two alleles, many can exist in a population
  - The trait is said to have multiple alleles
- Pigeons
  - Three alleles govern feather color
  - $B^A$ is dominant - ash red feathers
  - $B$ allele is dominant to $b$, but recessive to $B^A$ - blue feathers
  - $b$ is recessive to both - chocolate-colored feathers
Blood Type

- There are three alleles for the gene, “I”
  - $I^A$, $I^B$, and i
  - $I^A, I^A$ (AA) or $I^A, i$ (AO) – blood type A
  - $I^B, I^B$ (BB) or $I^B, i$ (BO) – blood type B
  - $I^A, I^B$ (AB) – codominance, blood type AB
  - $ii$ (OO) – blood type O
- There are different molecules that are produced on the surface of the rbc – represented by A and B
- Your immune system fights against blood cells with different molecules
- So who can donate blood to whom?
- $I^A, i \times I^B, i$ – What will be produced?
Rh Factor

- Separate gene from ABO blood type
- Simple heredity - “Rh+” is dominant over “Rh-”
- If an antiserum agglutinates your red cells, you are “Rh+” If it doesn't, you are “Rh-”
**Blood Type Frequencies**

- **O+** 38%
- **A+** 34%
- **B+** 9%
- **O-** 7%
- **A-** 6%
- **AB+** 3%
- **B-** 2%
- **AB-** 1%

<table>
<thead>
<tr>
<th>Recipient's blood</th>
<th>ABO antigens</th>
<th>ABO antibodies</th>
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<th>Donor type O cells</th>
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Compatible

Not compatible
Sex-Linked Traits

- Traits controlled by genes located on sex chromosomes are called sex-linked traits.
- The alleles are written as superscripts of the X or Y chromosome.
  - Alleles on Y chromosomes don’t have a corresponding allele on an X chromosome.
  - In males, if they have a recessive allele on their X chromosomes, there isn’t another allele to cover it up.

Virtual Fruit Fly Lab
Fruit Flies

Thomas Hunt Morgan (1910) experimented with fruit fly eye color

Red Eyed Female \((X^R X^R)\)  
\* White-Eyed Male \((X^r Y)\)

Assume two females and two males, what will the kids look like?

Females - all red \((X^R X^r)\)

Males - all red \((X^R Y)\)
Sex-Linked Traits in Humans

- Red-Green Color Blindness
- Recessive allele on the X chromosome
- How will a boy get it? A girl?
Sex-Linked Traits in Humans

- Hemophilia
- Recessive disease that prevents the blood’s ability to clot
- In males – 1 in 10,000
- In females – 1 in 100,000,000
- Why the difference?
Polygenic Inheritance

- The inheritance pattern of a trait is controlled by two or more genes
  - Skin color, height, corn cob length
  - Genes may be on the same chromosome or different chromosomes
  - Each gene may have two or more alleles

- Each allele represented by an uppercase letter contributes a small, but equal, portion to the trait being expressed
  - The result is that phenotypes show a continuous range of variability
### Polygenic Inheritance

<table>
<thead>
<tr>
<th>Gene 1</th>
<th>Gene 2</th>
<th>Gene 3</th>
<th>Total number of dark-skin genes</th>
<th># of light &quot;d&quot; alleles</th>
<th># of dark &quot;D&quot; alleles</th>
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<td>$D'^3D'^3$</td>
<td>6</td>
<td>0</td>
<td>6</td>
</tr>
</tbody>
</table>

- **Very light**
- **Medium**
- **Very dark**
Eye Color

Polygenic & Multiple Allelic

This means there are multiple alleles than can show up on multiple genes.

Many irises have multiple colors
MiniLab

Human eye color, like skin color, is determined by polygenic inheritance. You can detect several shades of eye color, especially if you look closely at the iris with a magnifying glass. Often, the pigment is deposited so that light reflects from the eye, causing the iris to appear blue, green, gray, or hazel (brown-green). In actuality, the pigment may be yellowish or brown, but not blue.

Procedure:

1. Use a magnifying glass to observe the patterns and colors of pigments in the eyes of 5 classmates.
2. Use crayons to make drawings of the 5 irises.
3. Describe your observations.

Analysis

1. Observe How many different pigments were you able to detect in each eye?
2. Critique From your data, do you suspect that eye color might not be inherited by simple Mendelian rules? Explain.
3. Analyze Suppose that two people have brown eyes. They have two children with brown eyes, one with blue eyes, and one with green eyes. What pattern might this suggest?
Genetics only determine potential

External Environmental Influences

- Temperature, nutrition, light, chemicals and infectious agents can all influence gene expression
- In Siamese cats and arctic foxes, temperature has an effect on coat color
- Leaves can have different sizes, thicknesses, and shapes depending on the amount of sunlight they receive
Environmental Influences

- Internal environmental influences
- Horn size in males and female mountain sheep is different due to differing internal environments
  - Also applies to baldness in humans and feather color in peacocks
- Age can also affect gene expression, though this isn’t completely understood
- Life choices
  - Live a healthy life, reduce your risk of heart disease, diabetes, etc.
Epigenetics

- The study of the changes in phenotype of gene expression caused by something other than the DNA
  - May be passed on
- Deals with certain genes being turned on or off in an individual
  - Cell memory – cells “remember” which genes were turned on or off through changes to the chromatin proteins
- May be one reason we can have so few genes
Section Review

1. A cross between an purebred animal with red hairs and a purebred animal with white hairs produces an animal that has both red hairs and white hairs. What type of inheritance pattern is involved?

2. If a white-eyed male fruit fly were crossed with a heterozygous red-eyed female fruit fly, what ratio of genotypes would be expected in the offspring?

3. A red-flowered plant is crossed with a white-flowered plant. All of the offspring are pink. What inheritance pattern is expressed?

4. The color of wheat grains shows variability between red and white with multiple phenotypes. What is the inheritance pattern?

5. Armadillos always have four offspring that have identical genetic makeup. Suppose that, within a litter, each young armadillo is found to have a different phenotype for a particular trait. How could you explain this?
Genetic Variation

In humans, how many possible combinations are there in a single sperm or egg?

$2^{23} = 8,388,608$ combinations

How many possible combinations with fertilization

$8,388,608 \times 8,388,608 = 7.04 \times 10^{13}$ (over 70 trillion)
Crossing Over in Meiosis

Crossing Over (recombination)
The homologous pair moves close together. The chromatids may exchange genes.

First cell division
Homologous pair of replicated chromosomes separate
Sister chromatids separate
Four haploid daughter cells

Genes that have crossed over

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Genetic Variation

- Crossing over during meiosis provides variability
- How many different kinds of gametes can a pea plant produce?
- Each cell has 7 pairs of chromosomes
  - Each can line up at the equator in two different ways and separate by segregation
  - \( 2^n = 2^7 = 128 \) possible combinations without crossing over
- When you include the egg, \( 128 \times 128 = 16,384 \) different combinations of offspring
With crossing over, additional variation is added providing an almost endless amount of variation possible.

This reassortment of chromosomes and the genetic information they carry, either by crossing over or by independent segregation of homologous chromosomes is called genetic recombination.

Variation is the raw material that forms the basis for evolution to act on.

Genetic Recombination
Genetic Diversity

FF to Minute 5!!!!!!!
Gene Linkage

- Genes that are close together on a chromosome are often inherited together
- Crossing over rarely works for just one gene
- These genes are said to be linked
- So chromosomes, not genes follow Mendel’s independent assortment
Chromosome Mapping

- Crossing over occurs
- Geneticists use the frequency of crossing over to map the relative position of genes on a chromosome
- Genes that are further apart are more likely to have crossing over occur
Fruit Fly Chromosome
Suppose there are 4 genes on a chromosome – A, B, C, D.

Frequencies of recombination as follows:
- Between A & B: 50% (50 map units)
- Between A & D: 10% (10 map units)
- Between B & C: 5% (5 map units)
- Between C & D: 35% (35 map units)

These give a relative distance between genes:
- A - 10 units - D - 35 units - C - 5 units - B

(whole thing is 50 units)
Section Review

1. What two processes contribute to the amount of genetic variation in humans?
2. How does crossing over occur?
3. What are “linked genes”?
4. How can you use crossing over to map a chromosome?