

Genetics Notes

Mendel Laid the Groundwork for Genetics

- **Traits** are distinguishing characteristics that are inherited, such as eye color, leaf shape, and tail length.
- **Genetics** is the study of biological inheritance patterns and variation in organisms.
- The father of modern genetics is **Gregor Mendel**, who bred thousands of plants, carefully counting and recording his results.
- From his data, he was able to predict the results of meiosis long before chromosomes were discovered. He was also able to describe how traits were passed between generations.

Mendel's Data Revealed Patterns of Inheritance

- Mendel worked with pea plants in the garden at the monastery. He made three key choices in his work:
 1. He had control over breeding (no random mating).
 2. He used purebred plants.
 3. He observed “either-or” traits that appeared in only two alternate forms.

Experimental Design

- Mendel chose pea plants because they reproduce quickly, and he could easily control how they mate.
- Plants contain both male and female reproductive organs. Because of this, plants can self pollinate or reproduce by themselves. If a line of plants has self-pollinated for long enough, that line becomes genetically uniform, or **purebred**.
- Mendel controlled the breeding of his pea plants by removing the male parts so the plants could not self-pollinate.
- Mendel chose to work with 7 traits in the plants: pea shape, pea color, pod shape, pod color, plant height, flower color, and flower position. All traits were “either-or”.

Results

- In genetics, the mating of two organisms is called a **cross**.
- Mendel crossed a purebred white-flowered pea plant with a purebred purple-flowered pea plant. These plants are the **P (parental) generation**.
- The resulting offspring are the **first filial (F1)** generation. The F1 generation was all purple flowers. The trait for white seemed to disappear.
- When Mendel allowed the F1 to cross, the resulting F2 produced BOTH purple and white flowers.
- He concluded that the trait for white color had not disappeared but had simply been hidden.
- Mendel continued to cross plants and found patterns in inheritance.

Conclusions

- Mendel drew 3 important conclusions.
- His first is the **Law of Segregation** which states:
 - Organisms inherit two copies of each gene, one from each parent.
 - Organisms donate only one copy of each gene in their gametes, so the two copies of each gene segregate (separate) during gamete formation.

The Same Gene Can Have Many Versions

- A **gene** is a piece of DNA that provides a set of instructions to a cell to make a certain protein.
- Each gene has a specific position on a pair of homologous chromosomes and has an alternative form known as an **allele**.
- Your cells have two alleles for each gene, one on each of the homologous chromosomes.
- Each parent donates one allele.
- **Homozygous alleles** are the SAME.
 - EX: Homozygous alleles for Tall pea plants would be TT, for short would be tt
- **Heterozygous alleles** are DIFFERENT.
 - Ex: Heterozygous alleles for Tall pea plants would be Tt.

Genes Influence the Development of Traits

- A **genome** is all of an organism's genetic material. Everyone has a unique genome that determines their traits.
- A **genotype** typically refers to the genetic makeup of a specific set of genes.
 - Ex: the genotype for a tall plant can be TT or Tt.
 - Ex: the genotype for a short plant is tt.
- A **phenotype** is the physical characteristics or traits of a specific organism.
 - Ex: the phenotype for TT or Tt is Tall.
 - Ex: the phenotype for tt is short.

Dominant and Recessive Alleles

- If individuals are heterozygous for a trait, which trait is expressed?
- A **dominant allele** is the allele that is expressed when two different alleles are present.
 - Ex: The tall allele (T) is dominant over the short allele (t).
 - Ex: In the genotype TT and Tt the tall gene is expressed.
- A **recessive allele** is the allele that is only expressed when two copies are present.
 - Ex: The short trait is ONLY expressed when the genotype is tt.
- Dominant alleles are ALWAYS expressed with **capital letters**, while recessive alleles are ALWAYS expressed with **lower case letters**.

Alleles and Phenotypes

- Two genotypes can produce the dominant trait.
 - Homozygous dominant will display the dominant trait.
 - Heterozygous will also display the dominant trait.
- The ONLY way to get the recessive trait is to be Homozygous Recessive.

Punnett Squares Illustrate Genetic Crosses

- R.C. Punnett developed the Punnett Square, which is a grid system for predicting all possible genotypes resulting from a cross. The outside of the grid represent the possible gamete genotypes of each parent. The inside of the box shows all the possible outcomes of that genetic cross.

A Monohybrid Cross Involves One Trait

- **Monohybrid crosses** examine the inheritance of only ONE specific trait.

Homozygous – Homozygous

- Ex: Cross a pea plant that is homozygous dominant for purple flowers with a pea plant that is homozygous recessive for white flowers. The resulting F1 can ONLY be heterozygous and purple.

	P	P
P	Pp	Pp
p	Pp	Pp

Heterozygous – Heterozygous

- Ex: Cross 2 heterozygous purple pea plants.

	P	p
P	PP	Pp
p	Pp	pp

- The resulting F1 generation produces: 1 PP – Homozygous Dominant Purple; 2 Pp – Heterozygous Purple; 1 pp – Homozygous Recessive white

Heterozygous – Homozygous

- Ex: Cross a heterozygous purple flower with a homozygous recessive white flower.

	P	p
p	Pp	pp
p	Pp	pp

- The resulting F1 generation produces: 2 Pp – Heterozygous Purple and 2 pp – Homozygous Recessive white
- If we did not know the genotype of the purple flowers, we could use a **testcross**. A testcross is a cross between an organism with an unknown phenotype and an organism with the recessive phenotype. The offspring will show whether the unknown is homozygous or heterozygous.

A Dihybrid Cross Involves Two Traits

- A **dihybrid cross** examines the inheritance of TWO different traits.
- Mendel wondered if both traits would always appear together or if they would be expressed independently of each other.
- Ex: Mendel crossed a homozygous plant with yellow round peas with a homozygous plant with green wrinkled peas.
 - Remember: Yellow is dominant (Y) and Round is dominant (R).
 - The cross is written: YYRR x yyrr

1234 1234
 - To figure out the parent's genes do the following: 1&3, 1&4, 2&3, 2&4

	YR	YR	YR	YR
yr	YyRr	YyRr	YyRr	YyRr
yr	YyRr	YyRr	YyRr	YyRr
yr	YyRr	YyRr	YyRr	YyRr
yr	YyRr	YyRr	YyRr	YyRr

- The resulting offspring will ALL be YyRr or Yellow and Round.
- NOW YOU TRY: Cross the F1 generation: YyRr x YyRr**

- The Resulting Phenotypes are: _____

Heredity Patterns Can Be Calculated with Probability

- **Probability** is the likelihood that a particular event will happen. It predicts the average number of occurrences, not the exact number of occurrences.
- Probability = $\frac{\text{Number of ways a specific event can occur}}{\text{Number of total possible outcome}}$
- Ex: If you flip a coin, the number of total possible outcomes is 2: heads up or tails up. The probability of heads is $\frac{1}{2}$ and the probability of tails is $\frac{1}{2}$.
- Ex: You flip 2 coins. The results of one have no effect on the next outcome.
- To calculate the probability of independent events. Multiply the two probabilities.
 - $\frac{1}{2} * \frac{1}{2} = \frac{1}{4}$
- Now look at a Punnett square: Cross two Heterozygous tall plants (Tt x Tt)

	T	t
T	TT	Tt
t	Tt	tt

- The resulting cross yields: Genotype: $\frac{1}{4}$ TT, $\frac{1}{2}$ Tt, $\frac{1}{4}$ tt
Phenotype: $\frac{3}{4}$ Tall, $\frac{1}{4}$ short

Sexual Reproduction Creates Unique Gene Combinations

- The major advantage of sexual reproduction is that it gives rise to a great deal of genetic variation within a species. This variation results largely from:
 1. The independent assortment of chromosomes during meiosis.
 2. The random fertilization of gamete.
- Independent assortment and fertilization play key roles in creating and maintaining genetic diversity in all sexually reproducing organisms.
- The possible combinations vary from species to species.

Crossing Over During Meiosis Increases Genetic Diversity

- **Crossing over** is the exchange of chromosome segments between homologous chromosomes during prophase I of meiosis I. Part of one chromatid from each chromosome breaks off and reattaches to the other chromosome.
- Crossing over happens any time a germ cell divides.
- Crossing over is also known as **genetic recombination**.

Two Copies of Each Autosomal Gene Affect Phenotype

- Some genetic traits depend on dominant and recessive alleles.
- Gene expression is often related to whether a gene is located on an autosome or on a sex chromosome.
- Remember **autosomes** are all but the sex chromosomes and sex chromosomes determine gender.

Disorders Caused by Recessive Alleles

- Some human genetic disorders are caused by recessive alleles on autosomes.
- This means both alleles must be recessive for the trait to be displayed in the phenotype.
- These disorders often appear in offspring of parents who are BOTH heterozygous.
- A **Carrier** does not show disease symptoms, but can pass on the disease-causing allele to offspring.

Disorders Caused by Dominant Alleles

- Dominant genetic disorders are far less common than recessive disorders.
- Huntington's disease is an example.

- Huntington's damages the nervous system and usually appears during adulthood.
- Because the disease is dominant, the child of a parent who has the disease has a 50/50 chance of getting Huntington's.
- Because Huntington's appears later in life, most people have children before they are aware that they have the disease.

Males and Females Can Differ in Sex-Linked Traits

Sex-Linked Genes

- **Sex-Linked Genes** are those located on the sex chromosomes (X and Y Chromosomes).
- XX female; XY male - Females only pass on X chromosomes. Males can pass on X or Y chromosomes.

Expression of Sex-Linked Genes

- Sex linked traits are expressed differently because the X and Y chromosomes are NOT the same.
- Females can pass on traits to males that are carried on the X chromosome.
- Because the Y does not carry these traits they are passed on from mother to son.
- That means any recessive allele on the X chromosome is expressed in a male.
- In females, one of the two X chromosomes is randomly "turned off" by a process called X Chromosome Inactivation.

Phenotype can Depend on Interactions of Alleles

Incomplete Dominance

- In **Incomplete Dominance**, a heterozygous phenotype is somewhere between the two homozygous phenotypes. Neither allele is completely dominant or completely recessive.
- Ex: Four-O'clock Plants
 - When homozygous Red (RR) is crossed with homozygous white (WW) you get heterozygous pink (RW).

Codominance

- **Codominance** occurs when BOTH traits are expressed in the heterozygous form.
- Ex: A codominant trait in Red and White flowers would form a heterozygous Spotted flower that was BOTH red and white.
- In humans, blood types are also codominant.
 - The possible blood types are A, B, AB, and O
 - A and B are dominant while O is the recessive.
 - The possible genotypes for blood are:
 - $I^A I^A$ or $I^A i$ make Blood type A
 - $I^B I^B$ or $I^B i$ make Blood type B
 - Only ii makes blood type O (universal donor)
 - $I^A I^B$ makes Blood type AB (universal recipient)
- PRACTICE
 - If a mother has type AB blood and a father has type O blood, what are the possible blood types of the offspring?
 - If a father is heterozygous blood type A and a mother is homozygous blood type B, what are the possible blood types of the offspring?

Many Genes May Interact to Produce One Trait

Polygenic Traits

- Traits produced by two or more genes are called **Polygenic Traits**. Ex: Human skin color and eye color are polygenic.

Epistasis

- In this case, one gene can overpower all other genes in terms of the traits. Ex: one brown eye, one blue eye and albinism in mammals.

The Environment Interacts with Genotype

- Some traits are determined by both genes and environment. Ex: sea turtle eggs become male or female turtles based on the temperature at which they incubate.
- Nutrition can effect body growth and development in humans causing genotypes not to be expressed.

Gene Linkage was explained through Fruit Flies.

- **Gene linkage** was first described by William Bateson and R.C.Punnett.
- American scientist Thomas Hunt Morgan, who worked with fruit flies, found connections. He noticed that some traits were inherited together. Morgan called these linked traits. Morgan concluded that linked genes were carried on the same chromosome.

Linkage Maps Estimate Distances Between Genes

- Linkage maps show the relative locations, or loci, of genes on a chromosome.
- On a linkage map, one map unit is equal to one cross-over for each 100 offspring, or one percentage point.

Females can Carry Sex-Linked Genetic Disorders

- Remember, a carrier can carry a gene for a trait, but does NOT express the trait.
- Only females can be carriers of sex-linked disorders.
 - Ex: Colorblindness, hemophilia, male-pattern baldness.
 - The royal family in England has many members with hemophilia because there was much inbreeding with cousins.

A Pedigree is a Chart for Tracing Genes in a Family

- A **Pedigree** chart can help trace the phenotypes and genotypes in a family to determine whether people carry recessive alleles. When enough family phenotypes are known, genotypes can be figured out.

Tracing Autosomal or Sex-Linked Genes

- Squares = males
- Circles = Females
- ----- = mating (wed)
- Shaded = has trait
- Empty = no trait
- Half shaded = Carrier

Several Methods help Map Human Chromosomes.

- Pedigrees are useful for studying genetics in a family.
- A **Karyotype** is a picture of all of the chromosomes in a cell. These can be used to study genetic disorders caused by nondisjunction. A Karyotype is most commonly used to diagnose Trisomy 21 (downs syndrome).
- Chromosome mapping can be done directly by searching for a particular gene.

Genetic Mutations

- A mutation is a change in a base sequence of DNA. A change in DNA can lead to a change in the protein that the gene codes.
- A change in the protein structure can lead to certain disorders, such as sickle cell anemia.