

Genetics and Heredity Notes

I. Introduction

- A. It was known for 1000s of years that traits were inherited but scientists were unsure about the laws that governed this inheritance.
- B. Gregor Mendel (1822-1884) was an Augustinian monk in Austria who experimented with garden peas and developed the foundation of modern genetics. He noticed that peas had several traits and always showed only one of a pair rather than a blend which was previously believed. He crossed plants with different traits to see what the offspring would look like.
- C. In a typical breeding experiment, Mendel would cross-pollinate (**hybridize**) two contrasting, true-breeding pea varieties (true-breeding means that the offspring all have the same trait).
 1. The true-breeding parents are the **P generation**, and their hybrid offspring are the **F₁ generation**. (filius - L, son)
 2. Mendel would then allow the F₁ hybrids to self-pollinate to produce an **F₂ generation**.
- D. When Mendel crossed a purple-flowered plant and a white-flowered plant the F₁ offspring were all purple.
 1. When Mendel allowed the F₁ plants to self-fertilize, the F₂ generation included both purple-flowered and white-flowered plants. The white trait, absent in the F₁, reappeared in the F₂.
 2. Mendel reasoned that the heritable factor for white flowers was present in the F₁ plants, but somehow did not affect flower color.
 3. Purple flower color is a **dominant** trait, and white flower color is a **recessive** trait.
 4. The offspring were always present in a 3-to-1 ratio in F₂. That is, three showed the dominant trait for every one showing the recessive trait.
- E. Mendel found that no matter what combinations he tried, one trait always dominated and masked the other.
 1. It didn't matter if the trait came from the male or female parent.
 2. The traits were controlled by factors which were later known as genes.
- F. Mendel worked out a hypothesis consisting of four statements to explain the results he observed.
 1. Alternative versions of genes account for variations in inherited traits.
 - a. These alternate versions are called **alleles** (short for allelomorph. allos - G, other; morph - G, form). For example, the gene that controls the color of the flowers in Mendel's peas has two alleles - purple and white.
 - b. Each gene resides at a specific locus on a specific chromosome.
 2. For each trait, an organism inherits two alleles, one from each parent.
 - a. In **homozygous** individuals, these two alleles are the same.
 - b. In **heterozygous** individuals, the two alleles are different.
 - c. An organism's traits are called its **phenotype**.
 - d. Its genetic makeup is called its **genotype**.
 3. If the two alleles for a trait are different, then one, the **dominant** allele, masks the presence of the other, **recessive** allele, and determines the appearance of the organism.
 - a. It is important to remember that the term "dominant" does not mean that the allele is the most common in the population. The recessive allele might be

more common.

4. Mendel's **law of segregation** states that the two alleles for a trait separate during gamete production and end up in different gametes. The **law of independent assortment** says that the alleles segregate independent of one another.
- If an organism has two identical alleles for a particular trait, then that allele is present as a single copy in all gametes.
 - If different alleles are present, then 50% of the gametes will receive one allele and 50% will receive the other.

II. Mendel's first experiments considered only one trait.

- A. An individual that is heterozygous for one trait is called a **monohybrid** and a cross between two heterozygotes is a **monohybrid cross**.

e.g.,
P round seed wrinkled
RR x rr
gametes R, R r, r
F₁ Rr (all round)

use two F₁ individuals as new Parents

P Rr x Rr
gametes R, r R, r
F₂ RR, Rr, Rr, rr (round, round, round, wrinkled; 3:1)

- B. A Punnett square can be used to show genotype, phenotype, and probability.

e.g., heterozygous purple (Pp) x white (pp)

	p	p
P	Pp	Pp
p	pp	pp

F₁ 1 purple: 1 white

e.g., two heterozygous tall plants (Tt)

	T	t
T	TT	Tt
t	Tt	tt

F₁ 3 tall: 1 short

C. Test cross

- Imagine that you have an organism showing a dominant phenotype. Is the individual homozygous or heterozygous? To be able to say for certain, a test cross is performed.
- The unknown individual is crossed with a homozygous recessive individual.
- The genotype of the unknown parent can be deduced from the appearance of the

offspring.

- III. Mendel later wondered if traits always travelled together or if they were inherited separately.
- A. To answer the question he considered two traits at once - a **dihybrid cross**.
- In one cross he studied the inheritance of seed color and seed shape.
 - The allele for yellow seeds (Y) is dominant to the allele for green seeds (y).
 - The allele for round seeds (R) is dominant to the allele for wrinkled seeds (r).
 - Mendel crossed true-breeding plants that had yellow, round seeds (YYRR) with true-breeding plants that has green, wrinkled seeds (yyrr).
 - If the two traits are transmitted from parents to offspring as a package, the F₁ spring would produce yellow, round seeds. The F₂ offspring would produce two phenotypes (yellow + round; green + wrinkled) in a 3:1 ratio, just like a monohybrid cross.
 - If the alleles separated independent of one another, we should observe four different combinations. The four different kind of sperm and four different kinds of eggs should give 16 different combinations in the offspring.
 - Mendel found a 9:3:3:1 ratio in the F₂ generation.
- B. This showed that traits are inherited independently.

e.g., YyRr x YyRr

	YR	Yr	yR	yr
YR	YYRR	YYRr	YyRR	YyRr
Yr	YYRr	YYrr	YyRr	Yyrr
yR	YyRR	YyRr	yyRR	yyRr
yr	YyRr	Yyrr	yyRr	yyrr

F₁ 9 yellow, round: 3 yellow, wrinkled: 3 green, round; 1 green, wrinkled

- IV. Mendelian inheritance is governed by the laws of probability.
- A. The probability scale ranges from 0 to 1, where 0 means there is no chance the event will occur and 1 means the event will occur every time.
- Probability can be calculated using the equation:
$$P = \frac{\text{\#correct outcomes}}{\text{\# total outcomes}}$$
 - The probability of tossing heads with a coin is $\frac{1}{2}$.
 - The probability of rolling a 3 with a six-sided die is $\frac{1}{6}$, and the probability of rolling any other number is $1 - \frac{1}{6} = \frac{5}{6}$.
- B. If the outcome of one event has no effect on the outcome of future events, they are said to be **independent events**. e.g., the chance of getting heads in a coin toss is $\frac{1}{2}$; the chance of getting heads a second time is $\frac{1}{2}$
- C. The **rule of multiplication** can be used to determine the chance that two or more independent events will occur together in some specific combination.
- Calculate the probability of each independent event.
 - Multiply the individual probabilities to obtain the overall probability of these events occurring together.

3. The probability that two coins tossed at the same time will land heads up is $1/2 \times 1/2 = 1/4$.
4. Similarly, the probability that a heterozygous pea plant (Pp) will self-fertilize to produce a white-flowered offspring (pp) is the chance that a sperm with a white allele will fertilize an ovum with a white allele.
5. This probability is $1/2 \times 1/2 = 1/4$.
6. The rule of multiplication also applies to dihybrid crosses.
 - a. For a heterozygous parent ($YyRr$) the probability of producing a YR gamete is $1/2 \times 1/2 = 1/4$.
 - b. The probability that an F_2 plant from heterozygous parents will have a $YYRR$ genotype is $1/16$ ($1/4$ chance for a YR ovum and $1/4$ chance for a YR sperm).

D. The **rule of addition** also applies to genetic problems.

1. The chance of either one of two possible outcomes occurring is the sum of the two individual probabilities.
 - a. For example, there are two ways that F_1 gametes can combine to form a heterozygote.
 - b. The dominant allele could come from the sperm and the recessive from the ovum (probability = $1/4$).
 - c. Or the dominant allele could come from the ovum and the recessive from the sperm (probability = $1/4$).
 - d. The probability of obtaining a heterozygote is $1/4 + 1/4 = 1/2$.

V. Not all traits follow the simple rules of Mendelian inheritance (*i.e.*, **complete dominance**). Some alleles show different degrees of dominance and recessiveness in relation to each other.

A. In **codominance**, both alleles are both expressed and affect the phenotype.

1. The heterozygote phenotype appears to be a blend of the two homozygous phenotypes.
2. An example is roan cattle. A cross between a red bull and a white cow yields roan calves. They calves appear reddish in color but on closer inspection, they have both red and white hairs. In other words, BOTH alleles are expressed..
3. A 1:2:1 is characteristic of codominance.

B. In **incomplete dominance**, the phenotype is also a blend of both alleles.

1. The offspring of a cross between heterozygotes show three phenotypes: each parental phenotype and a blended phenotype.
2. An example is seen in flower color of snapdragons.
 - a. A cross between a white-flowered plant and a red-flowered plant will produce all pink F_1 offspring.
 - b. Self-pollination of the F_1 offspring produces 25% white, 25% red, and 50% pink F_2 offspring.
3. A 1:2:1 is characteristic of incomplete dominance.

C. Some traits are controlled by genes for which more than two alleles exist. This is called **multiple alleles**.

1. Remember that, even if more than two alleles exist in the population, each individual only possesses two - one inherited from each parent.

D. In **epistasis**, one gene affects the phenotypic expression of a separate gene.

1. For example, in mice and many other mammals, coat color depends on two genes.
2. One, the epistatic gene, determines whether pigment will be deposited in hair or not.

- a. Presence (*C*) is dominant to absence (*c*) of pigment.
 3. The second gene determines whether the pigment to be deposited is black (*B*) or brown (*b*).
 4. An individual that is *cc* has a white (albino) coat regardless of the genotype of the second gene.
- E. Some traits appear to be distributed across a spectrum, like skin color or height. These are examples of **polygenic inheritance**.
1. Skin color in humans is controlled by at least three independent genes.
 2. An AAB^BCC individual is very dark; an aabbcc individual is very light.

VI. Many human genetic disorders are controlled by Mendelian inheritance.

- A. A **recessively inherited** disorder shows up only in homozygous individuals who inherit a recessive allele from each parent.
1. Genes code for proteins. An allele that causes a genetic disorder codes for a non-functional protein or no protein at all.
 - a. Homozygous dominant (AA) and heterozygous (Aa) individuals are normal in phenotype because the one copy of the normal allele produces a sufficient quantity of the protein to prevent the disorder.
 - b. A homozygous recessive (aa) individual is unable to produce enough of the protein and would show the disease phenotype.
 2. While heterozygotes may lack obvious phenotypic effects, they are **carriers** who may transmit a recessive allele to their offspring.
 3. If an allele is rare, it is usually unlikely that two carriers in the general population will meet and mate. Matings between close relatives, however, increase the risk because the allele exists in the family. This explains why “inbred” matings result in an increased risk of offspring having a recessive genetic disorder.
 4. If the disorder is lethal before reproductive age, no homozygous recessive individuals will reproduce.
- B. Although most harmful alleles are recessive, some human disorders are caused by alleles that are **dominant**.
1. A lethal dominant allele is more rare because even heterozygotes are affected (*i.e.*, die).
 2. If the disorder is lethal before reproductive age, the allele will not be passed on. The allele can be perpetuated in the population if it is late-acting.
- C. The risk that a particular genetic disorder will occur can sometimes be assessed before a child is conceived or early in pregnancy. This is part of **genetic counselling**.
1. **Carrier recognition** involves identifying parents who might be carriers of a particular allele.
 - a. Because most children with recessive disorders are born to parents with a normal phenotype, it is important to identify parents who might be carriers before they reproduce.
 - b. DNA fingerprinting can be used to identify carriers.
 - c. Embryos can also be screened to determine if they are affected by a disorder. Affected embryos can be destroyed while unaffected ones can be implanted into the mother.
 - d. There are obvious ethical questions that arise with this type of screening.
- D. A pedigree chart can also be used to evaluate the risk. This is a diagram that shows the occurrence and appearance or phenotypes of a particular gene or organism and its ancestors

from one generation to the next.

1. In a pedigree, squares represent males and circles represent females. Horizontal lines connecting a male and female represent mating. Vertical lines extending downward from a couple represent their children. Subsequent generations are therefore written underneath the parental generations and the oldest individuals are found at the top of the pedigree.
2. If the purpose of a pedigree is to analyze the pattern of inheritance of a particular trait, it is customary to shade in the symbol of all individuals that possess this trait.