

Chapter 9 – Patterns of Inheritance

Modern genetics began with Gregor Mendel's quantitative experiments with pea plants

History of Heredity

Blending theory of heredity - the heredity "stuff" of the parents blend together to produce the characteristics observed in the offspring

Particulate theory of heredity - each heredity factor from the parent (what we now call genes) will remain unchanged in the hybrid

Mendel's Critical Test

Mendel tested his idea by checking the offspring of the hybrid generation
Mendel could easily control matings among the plants because peas **self-fertilize**
When Mendel wanted to he could **cross-fertilize** plants very easily

Mendel worked with plants until he was sure he had **pure breeding** varieties - plants that when self-fertilized always produce offspring with traits like themselves
Mendel asked what would happen when he crossed his different varieties

Mendel carried out what are called **monohybrid crosses** - which are matings between individuals that differ in only one trait, or the tracking of the inheritance of a single trait
The offspring of a cross between 2 different varieties are called **hybrids**
And the cross-fertilization is referred to as **hybridization** or simply a **cross**

The parental plants are called the **parental (P) generation**
Their hybrid offspring represent the next generation called the **first filial (F₁) generation**
Mendel allowed the F₁ generation to self-fertilize to produce the next generation called the **second filial (F₂) generation**

A Monohybrid Cross

From his experimental data, Mendel deduced that an organism has two genes (alleles) for each inherited characteristic
One characteristic comes from each parent

From these kinds of results Mendel developed 4 hypotheses:

1. There are alternative forms of **genes** (discrete portions or sequences of the DNA molecule in chromosomes), the units that determine heritable traits
We now call alternative forms of genes **alleles**
2. For each inherited trait, an organism has 2 genes (alleles), one from each parent
These genes may be both the same allele, or they may be different alleles
3. A sperm or an egg carries only one allele for each inherited trait, because allele pairs separate (segregate) from each other during the formation of the gametes
4. When the 2 genes of a pair are different alleles, one is fully expressed and the other has no noticeable effect on the organism's appearance
These are called the **dominant allele** and **recessive allele** respectively

A pure breeding organism which has a pair of identical alleles for a trait is said to be **homozygous** for that trait

An organism with 2 different alleles for a trait is said to be **heterozygous** for that trait

Phenotype and Genotype

Because an organism's appearance does not always reveal its genetic composition, geneticists distinguish between an organisms expressed traits and actual genetic makeup

Genotype - the actual genetic makeup

If a gene has identical alleles at a locus the condition is called **homozygous**

If the 2 alleles happen to be the dominant form of the gene the condition is **homozygous dominant**

If the 2 alleles are the recessive form the condition is **homozygous recessive**

If a locus has different alleles at a locus the condition is called **heterozygous**

Phenotype - the observable expression of the genotype; the physical make up

The mechanism underlying the pattern of inheritance Mendel observed is stated by the **principle of segregation** - *pairs of genes segregate (separate) during gamete formation: the fusion of gametes at fertilization pairs genes once again*

We can see the connection between Mendel's principles and homologous chromosomes:
Alleles (alternative forms) of a gene reside at the same locus on homologous chromosomes

A Test Cross to Distinguish Genotypes

A **test cross** can allow you to determine the genotype of the plant

In a test cross an individual plant of unknown genotype is crossed with a plant whose genotype is homozygous recessive for the trait

The Inheritance of Two Independent Traits - The Dihybrid Cross

A **dihybrid cross**, is a mating between parents that differ with respect to 2 traits

Because the outcome of the dihybrid cross could be predicted by assuming that 2 gene pairs assort independently during the formation of gametes, Mendel proposed the second rule of heredity called the **principle of independent assortment**

Namely, different traits segregate independently of each other during the formation of the gametes

Note:

There is a connection between the behavior of chromosomes and the behavior of Mendel's heritable factors

The **chromosomal theory of inheritance** - it states that genes are located on chromosomes and that the behavior of chromosomes during meiosis and fertilization accounts for inheritance patterns

Mendel's Principles and Human Traits

There are a number of human traits that are thought to be determined by simple dominant-recessive inheritance

dominant traits

widow's peak
finger hair
freckles
double jointed thumb
free earlobe

recessive traits

straight hairline
no finger hair
no freckles
tight thumb ligaments
attached earlobe

The word "dominant" in genetics does not imply that a phenotype is either normal or more common than a recessive phenotype

Dominance simply means that an allele is expressed in a heterozygote, as well as in an individual homozygous for that allele

By contrast, the corresponding recessive allele is expressed only in a homozygote

Non-Mendelian Genetics

Incomplete dominance

For some traits, the F1 hybrids have an appearance somewhat in between the phenotypes of the 2 parental varieties

This effect is called **incomplete dominance** - a condition whereby neither allele in a heterozygote is dominant; instead, both alleles contribute to the phenotype

Multiple Allele Traits

So far we have discussed the inheritance patterns involving only 2 alleles per gene locus
But many genes have multiple alleles

Although an individual can only have 2 alleles for a trait at a particular locus, in the entire population more than 2 alleles may be present

The ABO blood groups in humans are one example of multiple alleles

4 blood types result from various combinations of 3 alleles: I^A (for the ability to make substance A), I^B (for B), and i (for neither A nor B)

Because there are 3 alleles there are 6 possible genotypes

Both the I^A , I^B alleles are dominant to the i allele

The I^A , I^B alleles are said to be **codominant**, meaning that both alleles are expressed in heterozygous individuals who have type AB blood

Note:

The matching of compatible blood types is critical for blood transfusions

If a donor's blood cells have a carbohydrate (A or B) that is foreign to the recipient, then the recipient produces blood proteins called antibodies that bind specifically to the foreign carbohydrates and cause the donor blood cells to clump together

This clumping can kill the recipient

Pleiotropy

In many cases one gene can influence several traits

The impact of a single gene on 2 or more traits is called **pleiotropy**

An example of pleiotropy in humans is sickle-cell anemia,

Traits Controlled by many Genes

Many traits are not qualitatively distinct like this, but vary quantitatively over a range of values

These kind of traits are called **quantitative traits**

Quantitative traits are somewhat complicated to study because they are controlled by several interacting genes, rather than by a pair of alleles at a single locus

This phenomenon is referred to as **polygenic inheritance**

A case in point of polygenic inheritance is skin pigmentation in humans

Linkage

The inheritance patterns are different if 2 traits happen to be on the same chromosome

Linkage is defined as the presence of 2 or more traits on the same chromosome

Crossing-Over

Crossing Over - a process during which corresponding parts of homologous chromosomes are exchanged

Caused due to breaks that occur in chromosomes during Meiosis I

Some of the key experiments that first demonstrated the effects of crossing over were performed by T. H. Morgan in the early 1900s using the fruit fly *Drosophila melanogaster*

Morgan performed the following experiment: a cross between a normal fruit fly (gray body and long wings) and an abnormal fly (black body and vestigial wings)

Morgan knew the genotypes of these flies from previous experiments (GGLL x ggll)

e.g. G = gray body (dominant)
g = black body (recessive)
L = long wings (dominant)
l = vestigial wings (recessive)

In mating these flies e.g. gray fly with long wings (GgLl) with a black fly with vestigial wings (ggll), Morgan performed a test cross

If the genes had not been linked then independent assortment would have produced offspring in a phenotypic ratio of 1:1:1:1

But, because these genes were linked, he obtained results in which most of the offspring had parental genotypes, but 17% of the flies were recombinants

Crossing-over and Mapping Genes

Crossing over data can be very useful in mapping the relative position of gene loci on chromosomes

The usefulness of crossing over data in this fashion was first discovered by Sturtevant

Assuming that the chance of crossing over is approximately equal at all points on a chromosome, he hypothesized that the further apart 2 genes are on a chromosome, the higher the probability that a cross-over would occur between them

Sex-Linked Inheritance

Chromosome pairs, with 2 identical members in both males and females are called the **autosomes**

Chromosomes that may have dissimilar members in the sexes are called **sex chromosomes**

In humans females have 2 identical sex chromosomes called **X chromosomes**

Males have only one X chromosome, and a smaller unpaired chromosome called the **Y chromosome**

The inheritance pattern is different when one considers traits on the sex chromosomes because many genes on the X chromosome are missing on the Y chromosome

For certain traits the males are effectively haploid

Recessive traits can be expressed in males with only one recessive allele

The gene for hemophilia is carried on the X chromosome (X^h)

Also, its a recessive trait: females must be X^hX^h to express the condition

Males need only have one X^h to exhibit the condition, because there is no corresponding allele on the Y chromosome to mask the expression of the "h" allele on the X chromosome