

## 1. General Information

**Genetics** – the branch of biology that seeks to explain biological variation  
Nothing on this planet is exactly alike. There are many different variations of all species  
Variation fuels natural selection, evolution. Some variations are due to the environment.  
**Human Genome Project** – a project to map the entire human genetic code.  
**Drosophila Neanogastro** – scientific name for the fruit fly (there's no need to memorize this)  
**Autosomes** – the first 22 pairs of chromosomes in a human  
**Sex Chromosome** – the 23rd pair of chromosomes  
In humans, if the 23 pair contains a Y, then the human is male  
If it does NOT contain a Y, then the human is female

## 2. Genes and Alleles

**Genes** – the part of nuclear material that contain instructions that guide the development of organisms  
Genes are passed down from generation to generation, providing the evolutionary link  
Defined as a segment of DNA whose sequence of nucleotides codes for a specific trait  
A single gene determines some traits, while multiple genes determine others  
**Alleles** – same gene, different variation. Example: Black and brown hair. Same gene, different allele  
Defined as an alternate form of a gene that can have different base sequences  
**Chromosomes** – packages of genes. Remember that eukaryotic DNA has introns (See Ch. 9)

## 3. Techniques for Studying Chromosomes and Sex Chromosomes

Chromosomes are most easily observable when the cell is dividing.  
Chromosomes can be identified by their shape, size, banding pattern, staining, etc.  
All chromosomes come in matching pairs except the last pair – the sex chromosome  
In humans, chromosomes are made up of X and Y chromatids  
Males – XY      Females – XX  
The egg contains an X chromatid, so the chromatid of the sperm determines the sex  
About 50% of the sperm produced contains X chromosomes, 50% contain Y chromosomes  
**Karyotypes** – a display of human chromosomes, arranged as homologous pairs  
Prepared by cutting individual chromosomes from a photograph and matching them pair by pair  
In other creatures, the make up of sex chromosomes are different  
Insects – X and O system. Females are XX, males X.  
Birds, some fish – ZA system. Males are ZZ, females, ZW  
Most plants and some animals have no sex chromosomes

## 4. Probability and Genetics

**Probability** – the branch of mathematics that predicts the chances that a certain event will occur  
Remember that the outcome of one event doesn't influence other events  
1 coin is tossed, 50% chance it will be heads. If it's tossed again, the chance is still 50%  
For more information on probability, go read a math book =)  
Probability is used to predict the genotypes of the offspring of various crosses  
**Chi Square Test** – a test for determining deviation from predicted results  
Probability deals with chance, not absolute numbers  
Not every family will have an equal number of boys and girls, even though the chance is 50/50  
Theoretically, the higher the number of trials, the closer the actual results are to predicted results  
If you toss a coin 10 times, you could easily get 7 heads and 3 tails, meaning 70/30  
If you toss it 100 times, you probably would get maybe 55 and 45  
If you toss is 1000 times, you might get 505 and 495  
As the number of tosses increases, the actual results get closer to the predicted results

## 5. Mendel and Simple Genetics

Mendel experimented with pea plants and determined the patterns of inheritance for single gene traits  
He used peas because they were easy to grow, grew fast, didn't need a lot of space, and cheap  
**Traits** – certain characteristics of organisms  
Mendel stated that peas were either round or wrinkled, yellow or green, tall or short, etc.  
He worked with one trait at a time, breeding round peas with wrinkled peas, or tall with short, etc.  
**Dominant trait** – when both traits are present, the dominant one will show. Represented by capital letter  
**Recessive trait** – will only show when no dominant trait is present. Represented by lower case letter  
**Genotypes** – the genetic makeup, or makeup of alleles (AA, Aa, AB, etc)  
**Phenotype** – the physical appearance, observable factors (green, round, tall, etc.)

**Homozygous** – the genotype contains alleles that are all the same (AA, BB, aa, bb, etc)

**Heterozygous** – the genotype contains alleles that are NOT the same (Aa, Bb, etc.)

**Homozygous dominant** – two dominant alleles (AA, BB, etc.)

**Homozygous recessive** – two recessive alleles (aa, bb, etc.)

**Heterozygous** – One dominant and one recessive (Aa, Bb, etc.)

Mendel's Question: If I breed a tall plant and a short plant, would I get a medium sized plant?

**Principle of Segregation** – The offspring receives one allele from each parent

**Monohybrid crosses** – crosses between parents that differ in only one characteristic

**Dihybrid crosses** – crosses between parents that differ in two characteristics

**The P Generation** – the parental cross. The cross between the two parents.

**The F1 Generation** – the offspring of the Parental cross

**The F2 Generation** – the offspring of breeding two members of the F1 generation

Dominant alleles are dominant over recessive alleles (duh)

If green peas (G) are dominant over yellow peas (g), then

GG – green (two dominants, no discussion needed)

Gg – still green (dominant trait exists, recessive doesn't show)

gg – yellow (no dominant trait present, recessive trait shows)

Punnett Square – a "tool" used to visually determine genotypes of different types of crosses

Consider breeding a parent that's homozygous dominant in height (TT) with homozygous recessive (tt)

Use the FOIL method from math or Punnett Square. TT and tt become Tt, Tt, Tt, Tt

	T	T
t	Tt	Tt
t	Tt	Tt

As you can see from the square, we get 4 offspring with genotype Tt. All the plants are tall

If pure tall with pure short cross in the P generation, result will be 4 tall in the F1 generation

The F2 generation will be:

	T	t
T	TT	Tt
t	Tt	tt

As you can see, you'll get 1 TT, two Tt, and 1 tt. This is a 1:2:1 genotype ratio

However, since TT and Tt both produce tall plants, the phenotype ratio is 3:1 (3 tall, 1 short)

Now, consider the dihybrid cross, a cross between plants that differ in two characteristics

Breed a homozygous dominant in both height and color (TTGG) with homozygous recessive (ttgg)

FOIL each of the alleles with itself. TTGG becomes TG, TG, TG, TG, ttgg becomes tg, tg, tg, tg

	TG	TG	TG	TG
tg	TtGg	TtGg	TtGg	TtGg
tg	TtGg	TtGg	TtGg	TtGg
tg	TtGg	TtGg	TtGg	TtGg
tg	TtGg	TtGg	TtGg	TtGg

When breeding a pure tall green plant with a pure short yellow plant, we get all tall green plants in F1

Now, the F2 generation: We're breeding TtGg with TtGg

FOIL first. In this case, the top and side of the square will be the same

TtGg becomes TG, Tg, tG, and tg

	TG	Tg	tG	tg
TG	TTGG	TTGg	TtGG	TtGg
Tg	TTGg	TTgg	TtGg	Ttgg
tG	TtGG	TgGg	ttGG	ttGg
tg	TtGg	Ttgg	ttGg	ttgg

As you can see, you'll get 1 TTGG, 2 TTGg, 2 TtGG, 4 TtGg, 1 TTgg, 2 Ttgg, 1 ttGG, 2 ttGg, 1 ttgg

However, since anything with a T and G will be tall or green or both, the phenotypes are:

Tall Green: 9

Tall Yellow: 3

Short Green: 3

Green Short: 1

The ratio for the F2 generation in the dihybrid cross of two pure parents will always be 9:3:3:1

**Principle of Independent Assortment** – alleles of one trait have no effect on alleles of another trait

### 6. Co dominance and Multiple Alleles

In Mendel's experiments, there was always a dominant and a recessive

Some traits, such as the color of snapdragons. The traits are equally dominant.

Consider a red snapdragon, RR, with a white one, WW.

F1 will be RW, RW, RW, RW, which is pink

When both traits are dominant, then the cross is a mix of the two

F2 will be RR, RW, RW, and WW – 1 red, 2 pink, 1 white. 1:2:1 ratio.

Multiple alleles – Before, there are only two choices, tall or short, green or yellow, etc.

Some traits have more than 2 choices, but only 2 alleles can be chosen, cuz only 2 parents

**Blood Type** – there's I<sup>A</sup>, I<sup>B</sup>, and i.

I<sup>A</sup>I<sup>A</sup> - forms type A

I<sup>B</sup>I<sup>B</sup> - Forms type B

I<sup>A</sup>I<sup>B</sup> - Forms type AB

ii - Forms type o

There are 3 different alleles, but only two are used at a time.

Type A blood produces a certain protein, and an antiprotein to type B

Type B blood produces another protein, and an antiprotein to type A

Type AB produces both proteins, and no antiproteins

Type O produces no proteins, and both antiproteins

**Antibodies** – proteins found the blood plasma that fight off foreign material

If a type A person gives blood to a Type B person, the type B person will die, and vice versa

Since O has no proteins, O is the universal donor – O people can donate to anyone

Since AB has no antiproteins, AB is universal receiver – AB people can receive from anyone

### 7. Multifactorial Inheritance and Linked Genes

Some traits are determined by more than one gene. These traits deviate from Mendelian ratios

The most common example is eye color. There are many different shades, tones.

**Modifier genes** – controls the shades of pigments in the eye

Controlled by multiple genes, and very graded in differences (in eye color, this means shades)

Height, weight, intelligence, hair color, and skin color are other examples of multiple gene traits

Many of these traits are also affected by the environment

**Linked Genes** – genes located on the same chromosome

Not all genes on a single chromosome are inherited together

When genes cross over, some genes are cut and replaced by other genes

The closer two link genes are positioned on the chromosome, the better the chance of staying together during crossing over

Using information on how often genes are separated, scientists can create gene maps

### 8. X Linked Traits (Sex Linked Traits)

These traits are located on the X chromosome. Discovered by **Morgan, Columbia University**

**Fruit Flies example** – bred white eye and red flies, got a 3:1 red to white ratio

However, all the white eyed ones were male.

The single recessive trait was only present on the X chromosome, not the Y

The recessive X, paired with a "neutral" Y, forms a male with white eyes

**Humans** – partial color blindness

X<sup>C+</sup> - normal      X<sup>C</sup> - color blind

Breed color blind mother with normal father, all sons will be colorblind, all daughters normal

Color blind mother genotype - X<sup>C</sup>X<sup>C</sup>

Normal father genotype - X<sup>C+</sup>Y

	X <sup>C</sup>	X <sup>C</sup>
X <sup>C+</sup>	X <sup>C+</sup> X <sup>C</sup>	X <sup>C+</sup> X <sup>C</sup>
Y	X <sup>C</sup> Y	X <sup>C</sup> Y

As you see, only the sons show the recessive color blind trait

## 9. Nondisjunction, X Inactivation, and Mutations Due to the Environment

**Nondisjunction** – failure of chromosomes in a homologous pair to separate during mitosis

Cells either end up with three chromosomes (XXX, XXY) or one (XO)

Discovered by Calvin Bridges, using vermilion (red-orange) eyed fruit flies

Since eye color is a sex-linked trait, only males had vermilion eyes (See above)

Vermilion eyes is recessive to dark red eyes (dominant)

However, 1/2000 was a female, due to nondisjunction. Had genotype  $X^V X^V X^V$

Nondisjunction can be considered a mutation

Nondisjunction in humans – humans with either 47 or 45 chromosomes

Doesn't have to be the sex chromosome, can occur to any pair of chromosomes

**Turner Syndrome** – XO in the 23 pair – short, underdeveloped, retarded, sterile, females only

**Klinefelter syndrome** - XXY in the 23 pair – tall, underdeveloped, retarded, males only

**Down Syndrome** – nondisjunction in the 21<sup>st</sup> pair. Retarded, short, characteristic facial features

Caused by Trisomy 21, or 3 chromosomes in the 21<sup>st</sup> pair.

Women over 35 make up of only 5% of mothers, but 25% of all Down Syndrome mothers

Most common serious birth defect. Affects 1/700 babies in the US

Nondisjunction in the autosomes (1<sup>st</sup> 22 pairs of chromosomes) often lead to miscarriages

X Inactivation – 1 X chromosome is inactivated in females after embryo develops

Since females have 2 X chromosomes, one is deactivated in each cell, randomly

Males have only 1 X chromosome, so X inactivation doesn't occur

**Barr Bodies** – condensed, deactivated, X chromosomes. They serve no important purpose

Discovered by **Mary Lyon**

The Environment and Genes

Radiation, and other factors, can mutate and affect genes

Smoking and drinking during pregnancy affects genes

**Nature VS Nurture** – nature are the genes, nurture is after birth, environment