Biology

Chapter 10
Study Guide

Trait

A trait is a variation of a particular character (e.g. color, height). Traits are passed from parents to offspring through genes.

Genes

Genes are located on chromosomes and consist of DNA. They are passed from parents to their offspring through reproduction.

Alleles

Alleles are alternative forms of a gene (e.g. a flower color may be white, another flower may be purple). This means that the gene for color is expressed in two different forms, either white or purple.

Homozygous

The two alleles representing the trait are identical (e.g. PP for purple color, pp for white color).

Heterozygous

The two alleles representing the trait are different (e.g. Pp for purple color). Although the pea plant appears purple in color (its phenotype is purple), its genotype (genetic make-up) is a mixture of a dominant "P" and a recessive 'p" trait.

Recessive Characteristics:

These are the traits that are masked by the presence of the more dominant traits. The recessive allele is inherited unchanged and can reappear.

Gregor Mendel (1822-1884)

Gregor Mendel was an Austrian Monk who discovered the basic rules or principals of heredity. Mendel chose the garden pea plant for his experiments and observations. He chose the pea plant for various reasons:

a. easily grown
b. easily manipulated
c. self pollination (the pea plant has both the male and female reproductive organs "stamen", and "carpel" within one plant).

Mendel experimented with 7 pea plant traits (each trait had 2 forms or two alleles):

1. flower color (purple or white)
2. flower position (axial "near the middle of flower" or terminal "near the end of stem")
3. pod color (yellow or green)
4. pod shape (inflated or constricted)
5. seed color (yellow or green)
6. stem length (long or short)
7. seed shape (round or wrinkled)

In some of his first experiments **(FIRST CROSS EXPERIMENTS)**, Mendel crossed a pure breeding purple flowered pea plant (PP) with a pure breeding white flowered pea plant (pp):

PP X pp (Parents-true breeding and Homozygous)

Pp Pp Pp Pp (F1 generation-offspring are Heterozygous)

Results: All the 2nd generation pea plants (F1) were purple in color. No blending of colors.

Again, Mendel crossed a pure breeding round seeded variety (RR) with a pure breeding wrinkled seeded pea plant (rr):

RR X rr (Parents)

Rr Rr Rr Rr (Offspring-F1)

Results: All 2nd generation pea plants (F1) were round seeded plants.

Mendel discovered that certain traits were masked (recessive characters, as the white flower color, and the wrinkled seed) while others appeared in the offspring without any blending of parent characteristics. This meant that F1 pea plants were either purple or white-no intermediate colors.

Mendel then allowed the 2nd generation plants (F1) to self pollinate **(SECOND CROSS EXPERIMENTS)**, and they produced:

PP X pp (Parents)

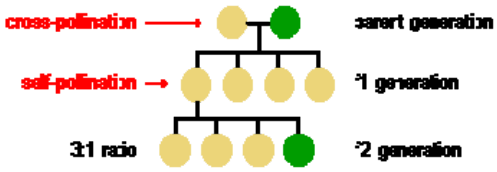
Pp Pp Pp Pp (F1)

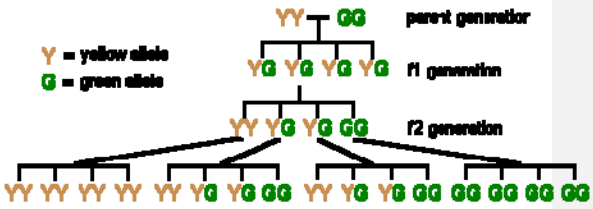
X

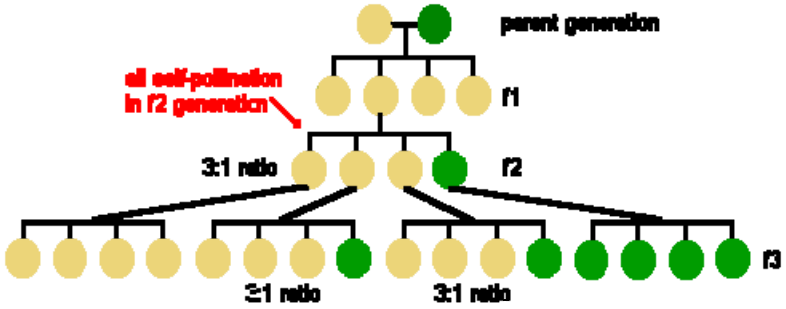
PP Pp Pp pp (F2)

Results: The white color trait reappeared in 25% (homozygous-pp) of the 3rd generation (F2). 50% of the flowers were purple (homozygous-PP) and 25% were purple (heterozygous-Pp).

Mendel repeated the same experiments on the **pod color of pea plants (yellow or green):**







Results: Mendel found that the 1st offspring (F1) always had yellow seeds (dominant trait), however the following generation (F2), had yellow to green in the ratio of 3:1. The 3:1 ratio continued in later generations as well (F3).

Mendel came to the following conclusions:

1. Inheritance of each trait is determined by "units" or "factors" that are passed on to descendants unchanged (units or factors are what we know today as GENES).

2. An individual inherits a unit from each parent for each trait.

3. A trait may not show up in an individual but can still be passed on to the next generation.

Summary of Mendel's Observations:

1. Starting plants (Parents) were all homozygous (had same allele) for flower color, pod color, and seed shape. The parents were all pure breeding plants.

2. F1 generations were all heterozygous (each offspring had inherited 2 different alleles- one from each parent). This means that when gametes are formed, the factors separate and are distributed as units to each gamete (rule of segregation).

3. If an organism had 2 unlike forms of a gene (2 different alleles) for a certain characteristic, one allele is expressed as dominant versus the second allele which is masked and expressed as recessive. The recessive trait may affect an individual's phenotype, but can still be passed on to the next generations and have an effect.

Mendel's Principals

1. Principal of Segregation

For any trait, the pair of alleles of each parent separate and only 1 allele passes on from each parent to offspring. Which allele is inherited is random. This separation occurs during meiosis.

2. Principal of Independent Assortment

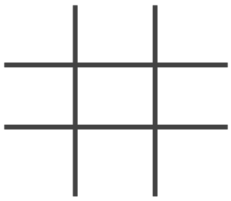
Different pairs of alleles are passed to offspring independently of each other. This means if you inherit a particular eye color that does not mean that this trait increases the likelihood or chances of you becoming tall or having 6 fingers! We know this today because we understand that genes are located on different chromosomes for different traits.

Probability of Inheritance and Punnett Squares

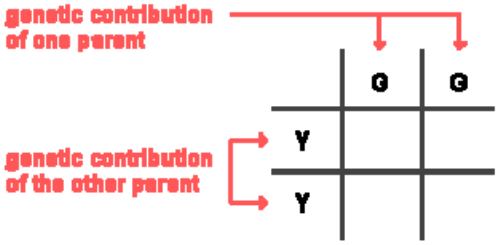
We can predict the likelihood of inheriting particular traits. This help with breeding animals and plants, as well as help humans to explain inheritance of certain traits and diseases within family lines.

Reginald Punnett was an English 20th century geneticist who developed a technique to calculate the probability of inheriting a specific trait. His method was used to discover the potential combinations that can occur in children given the genotypes of the parents.

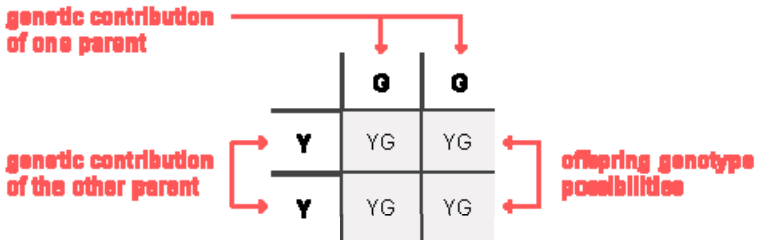
1. First you draw a grid of perpendicular lines



2. You put the genotype of 1 parent across top, and the genotype of the other parent down the left side.



3. Copy the row and column head letters across or down into the empty squares. In this case both parents are homozygous. Y (yellow color) is the dominant allele which masks the recessive G (green color). All offspring are yellow.



4. In this case, both parents are heterozygous. The masked green color (GG) reappears in 1 out of 4 of the offspring (25%).

	Y	G
Y	YY	YG
G	YG	GG

If both parents are heterozygous (YG genotypes), there will be 25% YY, 50% YG, and 25% GG

Punnett's Squares and Human disorders

Case 1

	A	a
A	AA	Aa
a	Aa	aa

If both parents are carriers of the recessive allele for a disorder, all of their children will face the following odds of inheriting it:
25% chance of having the recessive disorder
50% chance of being a healthy carrier
25% chance of being healthy and not have the recessive allele at all

Case 2

	A	a
a	Aa	aa
a	Aa	aa

If one parent is a carrier and the other has a recessive disorder, their children will have the following odds of inheriting it:
50% chance of being a healthy carrier
50% chance having the recessive disorder

Case 3

Some disorders are caused by dominant alleles for genes. Inheriting just 1 copy of this dominant allele will cause the disorder (e.g. Huntington disease, polydactyl).

	A	a
a	Aa	aa
a	Aa	aa

If only one parent has a single copy of a dominant allele for a dominant disorder, their children will have a 50% chance of inheriting the disorder and 50% chance of being entirely normal.

From this case we can determine that AA individuals are not healthy (have the disorder because they have both alleles of dominant trait), and Aa individuals are also not healthy (have 1 copy of the dominant allele), while the individuals with aa are healthy and are not carriers of the disease since they do not have a copy of the dominant allele).

Exceptions to Mendel's Pattern

1. Polygenic Traits

Traits are determined by the combined effect of more than 1 pair of genes (polygenic or continuous traits).

e.g. human stature, human skin color, hair color, and eye color

Whether an individual achieves his or her genetically programmed height is affected significantly by HGH (human growth hormone) produced by the pituitary gland.

A deficiency in these hormones during childhood or puberty results in stunted growth. So although we can calculate or estimate the height of offspring using parents' height, other factors may intervene to alter this.

2. Intermediate Expression

Blending occurs in the phenotype when there is incomplete dominance resulting in intermediate expression of a trait.

e.g. In some flowers (as four o'clock), the red or white flowers are homozygous, while the pink are heterozygous. The pink flowers are a result of the red allele being unable to code for production of enough pigment to make the petals dark red.

e.g. Tay-Sachs disease; this disease is caused by incomplete dominance (child killer disease at young age). Heterozygous individuals are genetically programmed to produce only 40-60% of the normal amount of an enzyme that prevents the disease.

3. Co-dominance

Individual exhibits the characteristics of 2 dominant traits.

e.g. Human Blood Type AB

Individuals have characteristics of type A and type B blood.

4. Incomplete Penetrance

Some genes are incompletely penetrant (this means their effect does not normally occur unless certain environmental factors are present).

e.g. You may inherit the genes that are responsible for type 2 diabetes, but you never get the disease unless you become overweight, or suffer from stress.

5. Sex-Related Genetic Effects

a. sex limited genes

These are inherited by both men and women, but are normally expresses in the phenotype of only one pf them.

e.g. Male beard. Women have facial hair, but it is sparse.

b. sex controlled genes

These are expressed in both sexes, but differently
e.g. gout-a disease that causes more sever pain in joints for men that if it occurs in women.

c. genome effects

These genes have different effects on individuals depending on which parent (the gender) from whom they were inherited.

e.g. Angelman Syndrome- mental retardation.

Environmental Influences on Inheritance and Genes

An individuals' phenotype depends on environment as well as genes. The sunlight affects the color of your skin. Nutrition plays an important role as well. Some diets do not promote healthy or normal growth. The health of pregnant women, and breast feeding mothers is also important for a child's normal growth.

BLOOD TYPES

There are 4 blood types A, B, AB, and O

Type A→ have A carbohydrate molecules on their red blood cells.

Type B→ have B carbohydrate molecules on their red blood cells.

Type AB→ have A and B carbohydrate molecules on their red blood cells

Type O→ have neither A nor B carbohydrate molecules on their red blood cells.

The A and B carbohydrate molecules are called "antigens" because they stimulate your body to produce an immune response, including antibodies.

Antibodies

These are proteins which travel in your blood to help destroy viruses or bacteria.

Antibodies do not attack their own body cells. So this means that people with type A blood do not make antibodies against type A antigen which is present on their red blood cells, but they make antibodies against type B antigen.

If you are given a blood transfusion that doesn't match your blood type, antibodies in your blood type can react with antigens present on the red blood cell and this can be fatal!

e.g. Type A blood person is given a type B blood transfusion, will cause the person's anti-B antibodies to react with the type B antigens on the donated red blood cells and cause a harmful reaction.

The Universal Donor = Type O
The Universal Receiver = Type AB

Comment [a1]:

Blood Type	Receives From	Gives Blood To
A	A and O	A and AB
B	B and O	B and AB
AB	A, B, AB, and O	AB
O	O only	A, B, AB, and O

Genetics of Blood Types

You receive one blood type gene from your mother and one from your father. The two genes combined determine your blood type. The blood type gene has 3 alleles:

I^A results in A antigen
I^B results in B antigen
ii doesn't result in either antigen

I^A I^A (from 1 parent) and I^Ai (from another parent)→ result in Type A blood

I^B I^B (from 1 parent) and I^Bi (from another parent)→ result in Type B blood

ii and ii→ result in Type O blood