

MAMMOTH
SCIENCE

Unit 14- Biodiversity

- *Know that Gregor Mendel used pea plants to study heredity because*
- *Describe the principles of Mendelian Genetics*
- *Predict the results of a monohybrid genetic cross by using Punnett Squares and probability calculations*
- *Predict the parental genotypes for offspring*
- *Predict the results of a Dihybrid cross using Punnett Squares*
- *Determine the genotype of an organism with a dominant phenotype by applying a test cross*
- *Analyze a simple pedigree*
- *Identify the factors that influence patterns of heredity*
- *Describe how genetic disorders such as sickle cell anemia, cystic fibrosis, hemophilia, and Huntington's Disease may occur*



Menu

Who Was Gregor Mendel

Mendelian Genetics - Early Experiments

The Original Cross

Genes & Alleles

Dominant & Recessive

Genotype vs Phenotype

Homozygous vs Heterozygous





Menu

Monohybrid vs Dihybrid Crosses

Test Cross

Principles of Mendelian Genetics

Incomplete vs Codominance

Multiple Alleles

X-Linked Traits / Sex Linked Inheritance

Pedigrees





Menu

Genetic Disorders



Who Was Gregor Mendel?



Introduction

- An organism's heredity is the *set of characteristics it receives from its parents*.
- The *study of heredity* is known as **genetics**.
- **Genetics** can be divided into three major areas:
 - Molecular
 - Transmission
 - Population

Gregor Mendel

- Known as the *Father of Genetics*
- Born in 1822 - in what is now a part of the Czech Republic.
- 1843 entered a monastery in Austria.
- 1851 attended the University of Vienna and studied Mathematics & Science.
- Left Vienna and went back to the monastery where he taught and supervised the garden.



Mendelian Genetics - Early Experiments



- **Mendelian Genetics:**

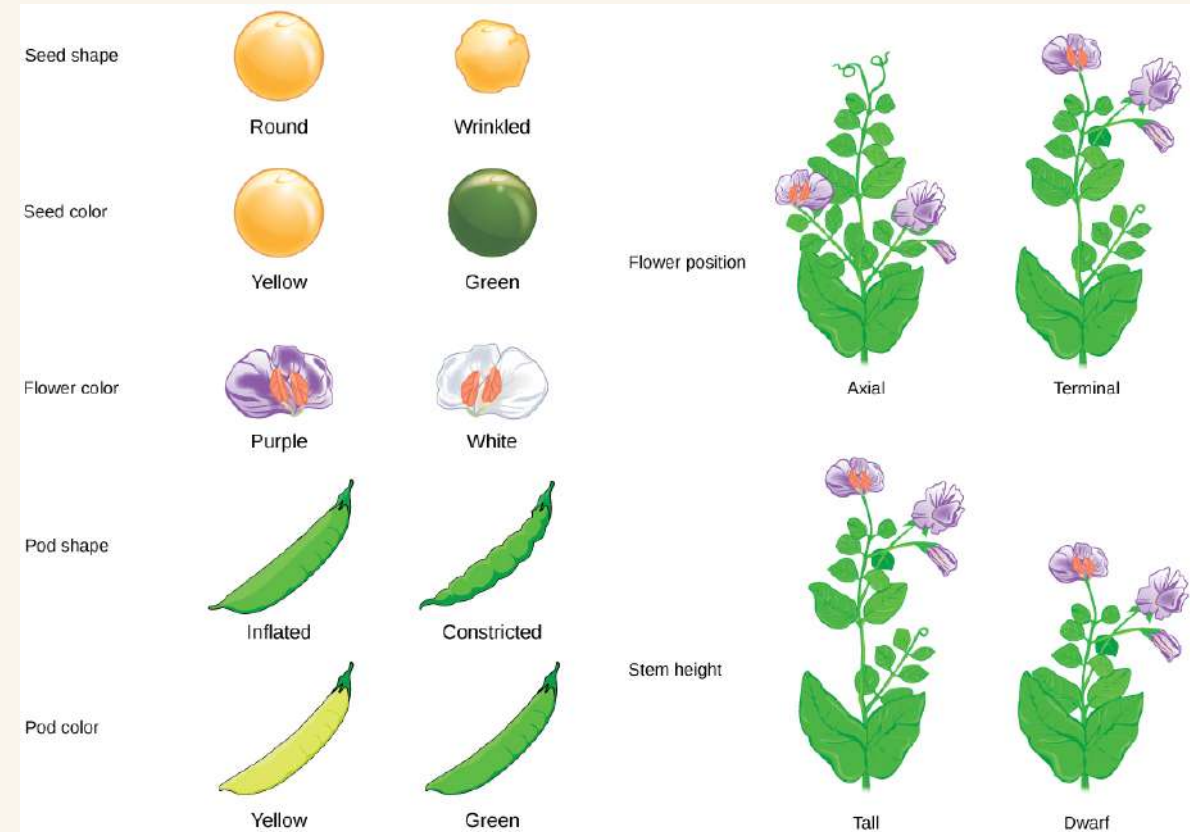
- The monks had developed ***true-breeding stock*** of pea plants.
- A true-breeding stock *always passes its characteristics* to the next generation.
 - Ex. A true-breeding tall plant passes on tall characteristics.
- Mendel used the pea plant because *pea plants normally reproduce by self-pollination*.
- It is very *easy to cross-pollinate* pea plants.
- **Traits:**
 - Mendel chose to study seven traits in pea plants
 - ★ A **trait** is a *characteristic that distinguishes one individual from another*.
 - ★ **Polygenic Trait:** *a characteristic of an organism that is determined by many genes* – Ex. Height & weight, eye color, ear lobes...



The Traits Mendel Studied



- *Flower position* = *Axial or Terminal*
- *Plant height* = *Tall or Short*
- *Pod color* = *Green or Yellow*
- *Pod appearance* = *Inflated or Constricted*
- *Seed color* = *Yellow or Green*
- *Seed appearance* = *Round or Wrinkled*
- *Flower color* = *Purple or White*

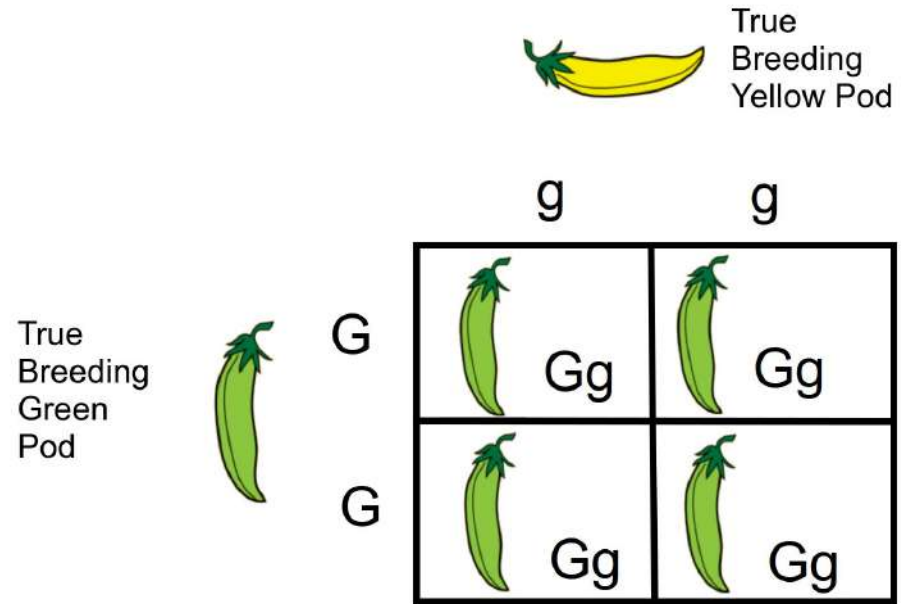


The Original Cross



– The Original Cross:

- Mendel crossed the pea plants.
- *True-breeding tall pea plants with a true-breeding short pea plant.*
- The *offspring* of the crosses are called **hybrids**.
- Mendel called the hybrids the ***F1 generation***. The letter *F* stands for *fillius* which means son.
- The true-breeding plants Mendel called the ***P generation*** stands for *parentis* meaning the parent.
- The traits *didn't blend instead one trait showed up* and one “vanished”.

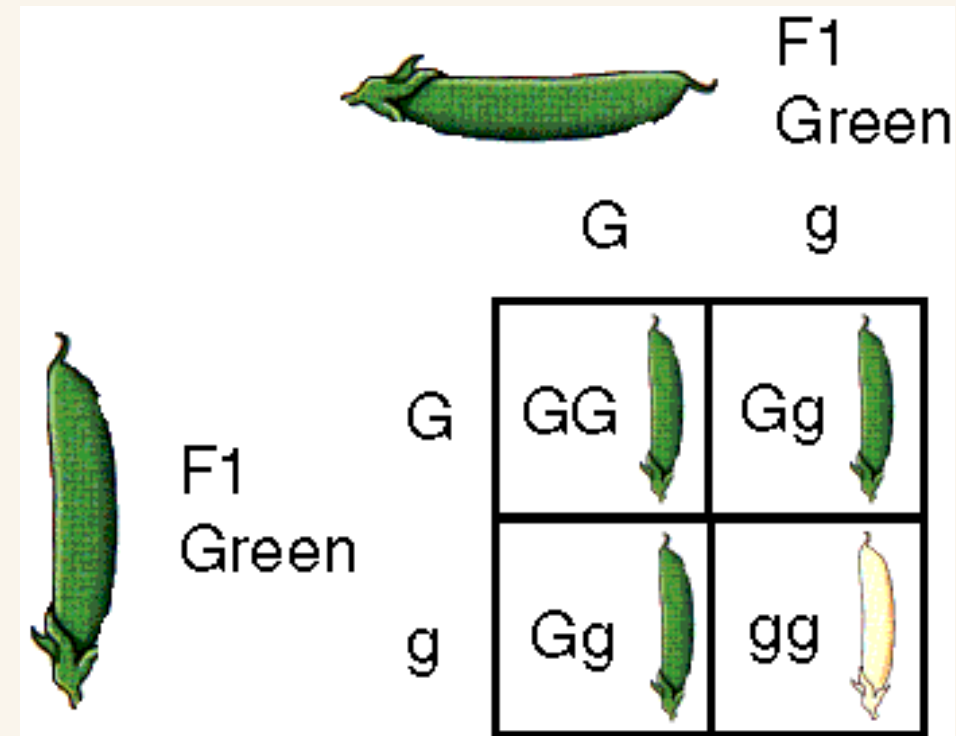


F1 vs F2 Generations

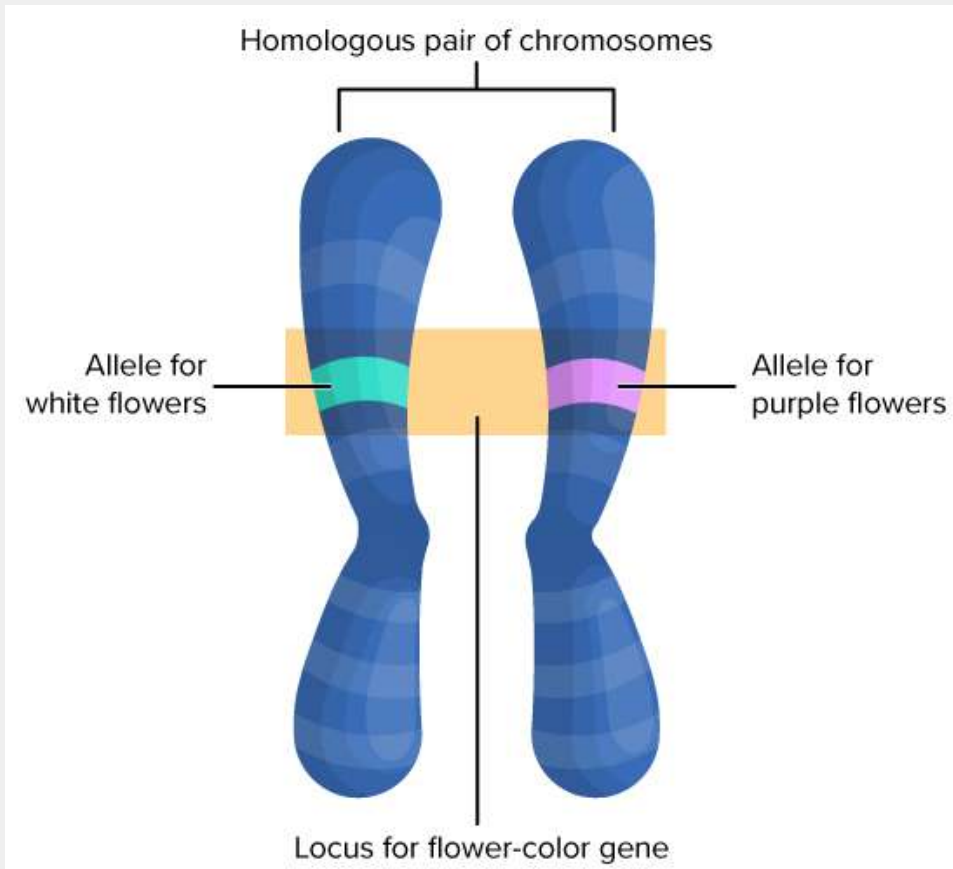


— F₁ vs. F₂

- Mendel *crossed* the plants of the **F₁** generation among themselves.
- He called the next generation **F₂**.
- *Some of the traits* that had “vanished” *reappeared* in the **F₂** and they reappeared in *approximately one fourth* of the plants. – Why?



Genes & Alleles



Genes and Alleles

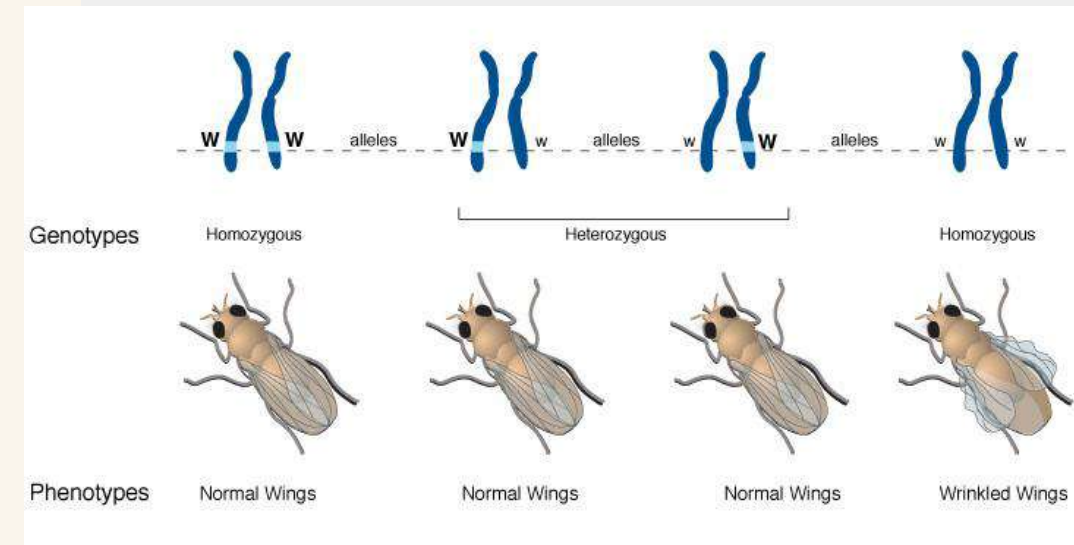
- What Mendel called *characteristics* we now call a **gene**.
- True-breeding tall pea plants contain a gene for tallness.
- Mendel concluded that for each of the seven traits he investigated, *a plant must contain two genes-one from each parent*.
- These *different forms of a gene* are called **alleles**.
- How is genetic Information passed?
 - *Parents pass on one copy of a gene to each new generation then each has one, from each parent.*
 - The *Dominant allele* will mask over a **recessive allele** or the **dominant** gene is expressed.
 - Use *capital letters for dominant alleles* and *lowercase for recessive alleles*.

Genotype vs Phenotype



- **Phenotype and Genotype**

- An organism's *phenotype* is the form of a trait it displays
- An organism's *genotype* is its genetic makeup.
- **Phenotype:** *the appearance of an organism resulting from the interaction of the genotype and the environment.*
- **Genotype:** *the genetic makeup of an organism or group of organisms regarding a single trait, set of traits, or an entire complex of traits.*

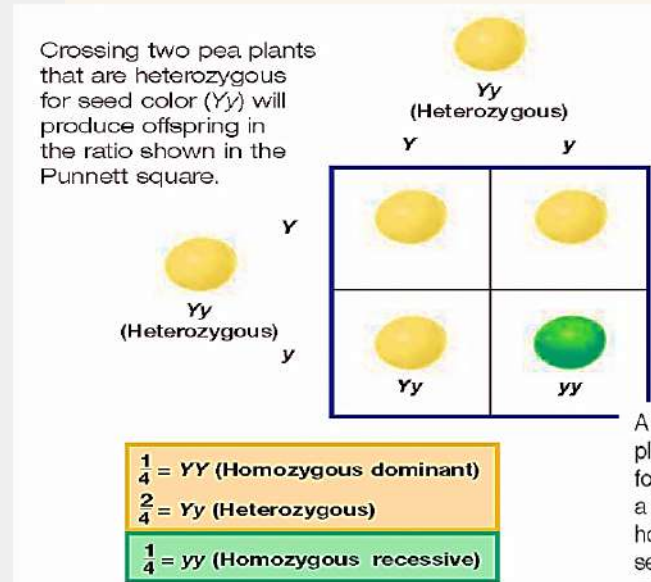


Homozygous vs Heterozygous

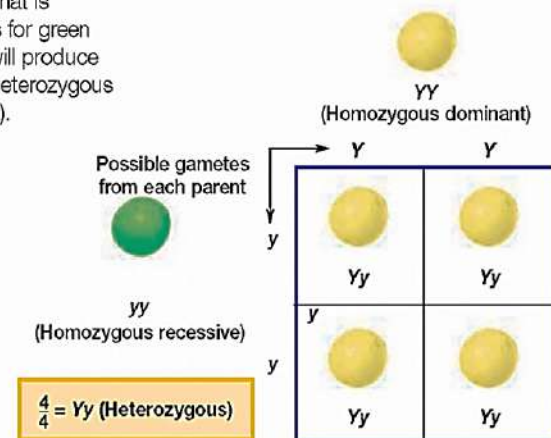


- **Homozygous vs. Heterozygous**

- If an organism is true-breeding, *having identical pair of alleles* (both upper and both lower case) it is **homozygous**.
- if an organism has a *mixed pair of alleles* (one upper and one lower) it is **heterozygous**.



A cross between a pea plant that is homozygous for yellow seeds (YY) and a pea plant that is homozygous for green seeds (yy) will produce only yellow heterozygous offspring (Yy).

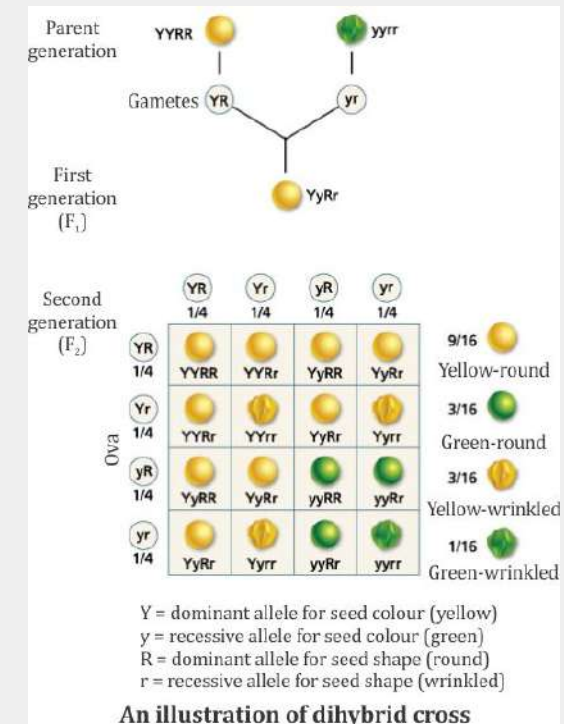
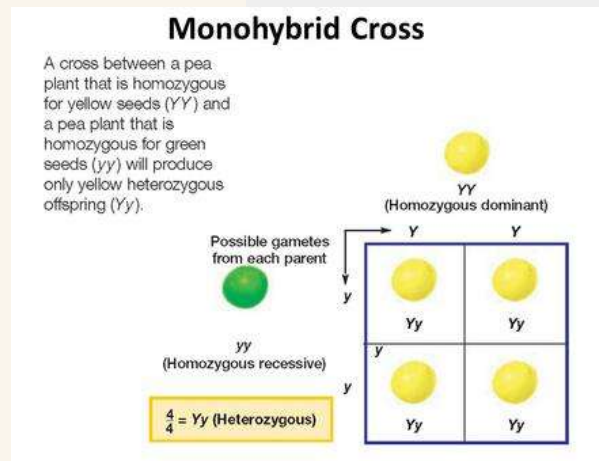


Monohybrid vs Dihybrid

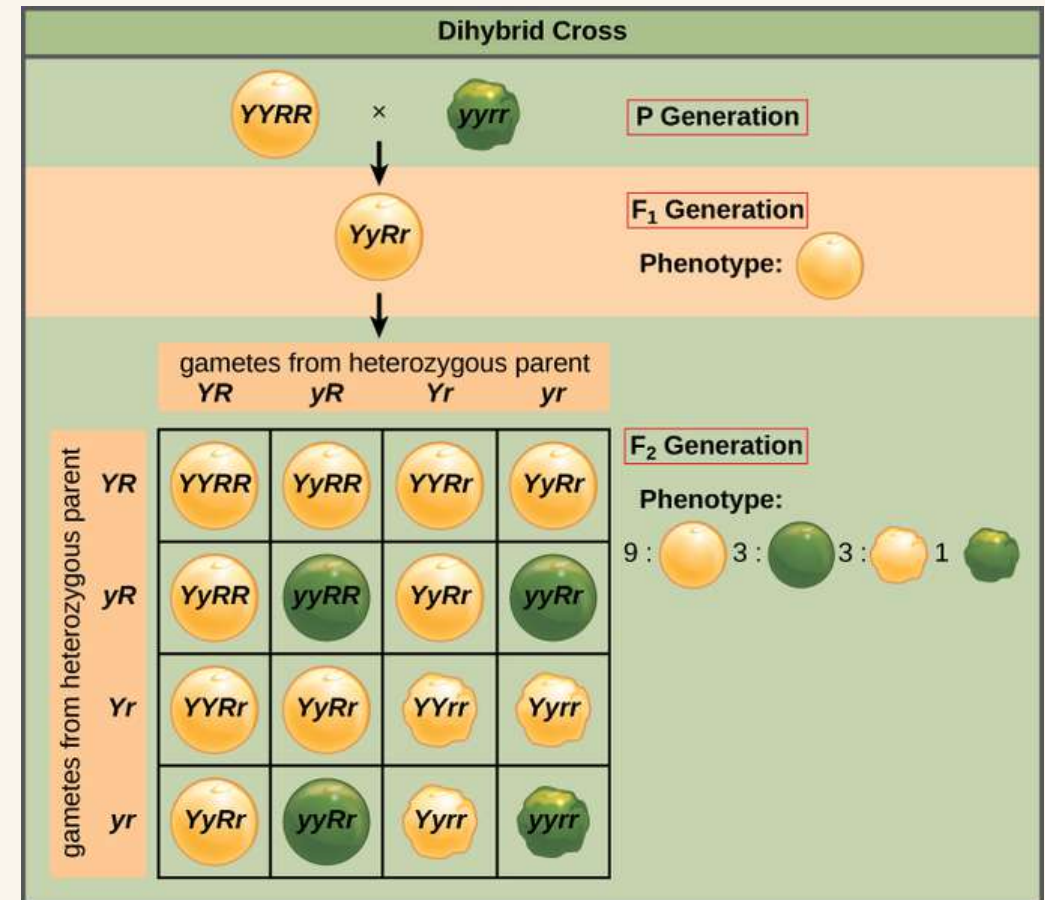
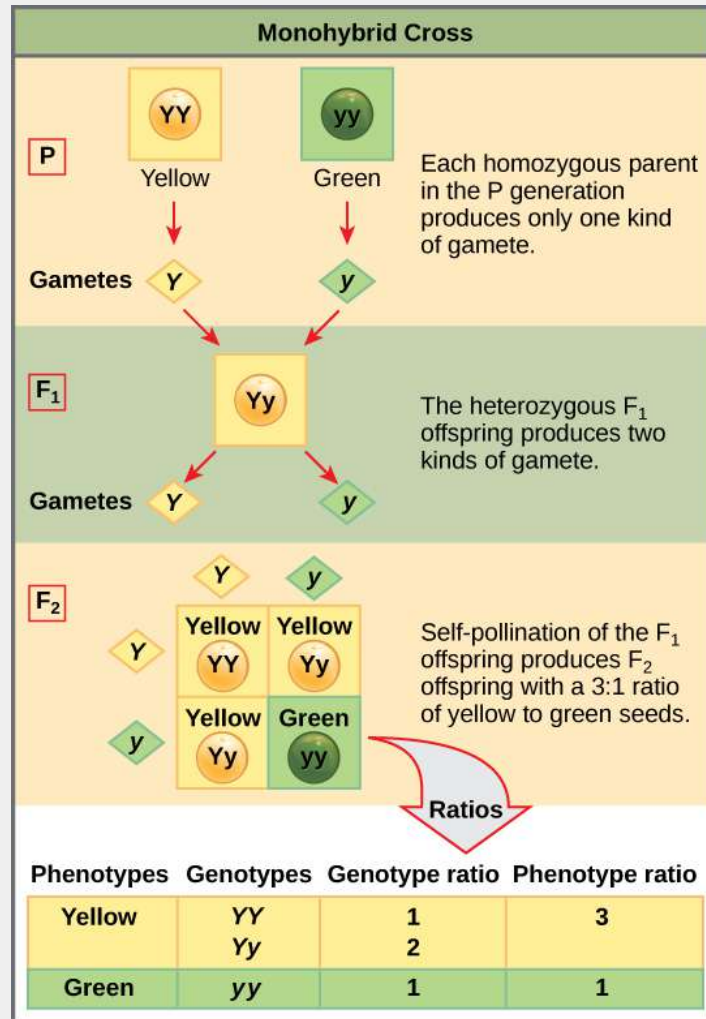


Monohybrid vs. Dihybrid Crosses:

- A cross between individuals that involves one pair of contrasting traits in called a **monohybrid cross**.
- A **Dihybrid cross** is a cross between individuals that involves two pairs of contrasting traits.
- Biologists use a **Punnett Square** to aid them in predicting probabilities, and Mendel's principles to predict the likely outcome of genetic crosses.
- Probabilities inside Crosses:
 - **Probability:** the likelihood that a possible future event will occur in any given instance of the event; the mathematical ratio of the number of times one outcome of any event is likely to occur to the number of possible outcomes of the event
 - **Homo/Homo - Mono:** all the same
 - **Homo/Hetero - Mono:** $\frac{1}{2}$ homo, $\frac{1}{2}$ hetero
 - **Hetero / Hetero:** 1 homo dominant, 2 hetero, 1 homo recessive
 - **General Dihybrid Crosses:** 9:3:3:1



Probabilities

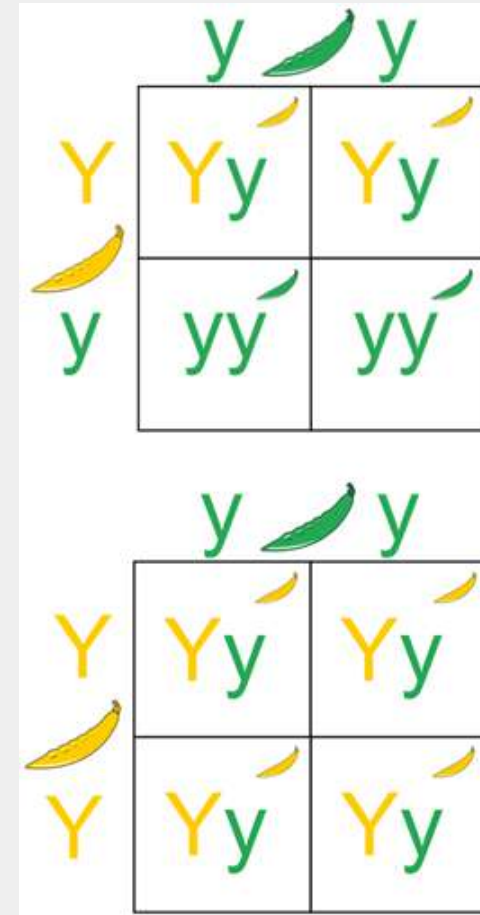


Test-Cross



- **Test Cross:**

- To find out if an organism's genotype is homozygous or heterozygous for a trait you perform a *test cross*.
- A **test cross** is *when an individual of unknown genotype is crossed with a homozygous recessive individual*. A testcross can determine the genotype of any individual whose phenotype is dominant.



Principles of Mendelian Genetics



Principles of Mendelian Genetics:

- **Principle of Dominance and Recessiveness**

- If an organism *inherits different alleles for the same trait one allele may be dominant over the other.*

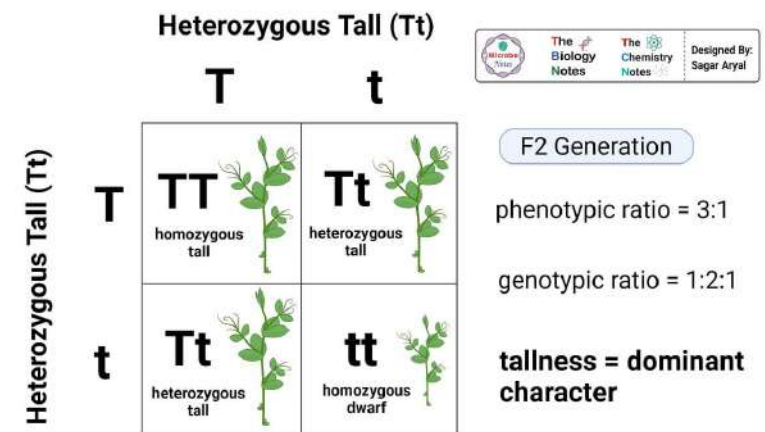
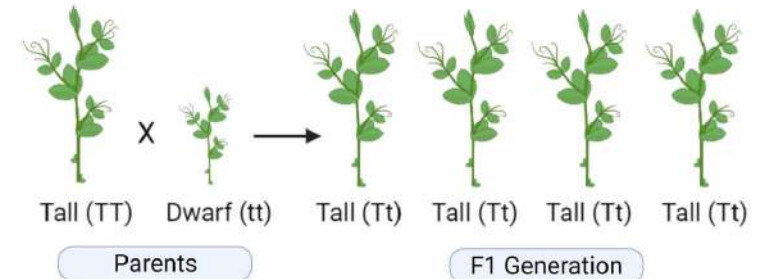
- **Principle of Segregation**

- For *each gene*, an *organism receives one allele from each parent*. The alleles *separate from each other when reproductive cells are formed.*

- **Principle of Independent Assortment**

- Some *genes segregate independently.*

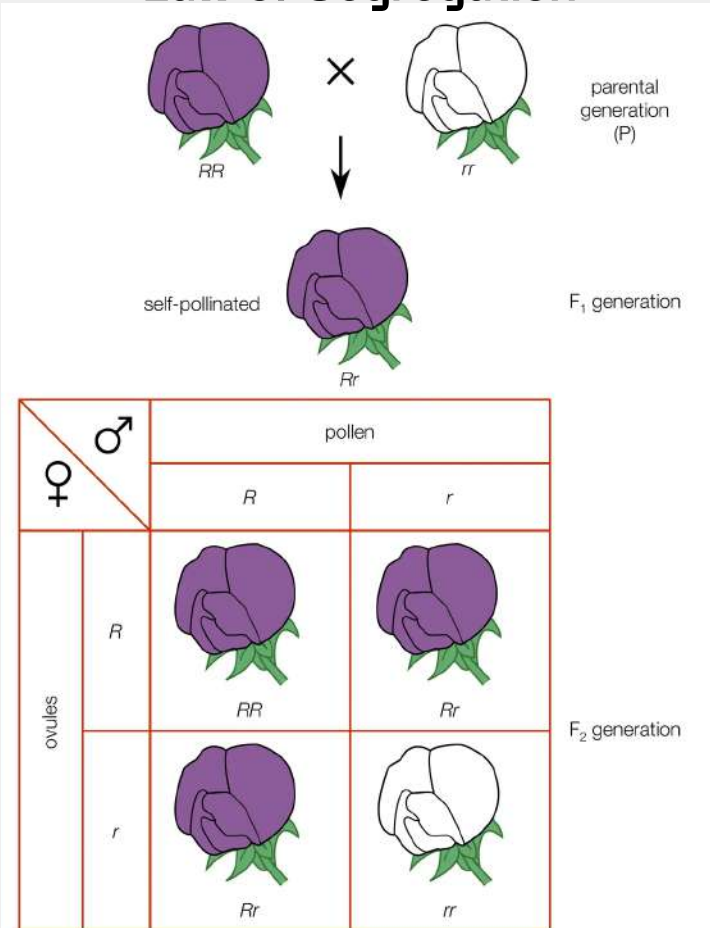
Mendel's Law of Dominance



Laws of Segregation and Independent Assortment

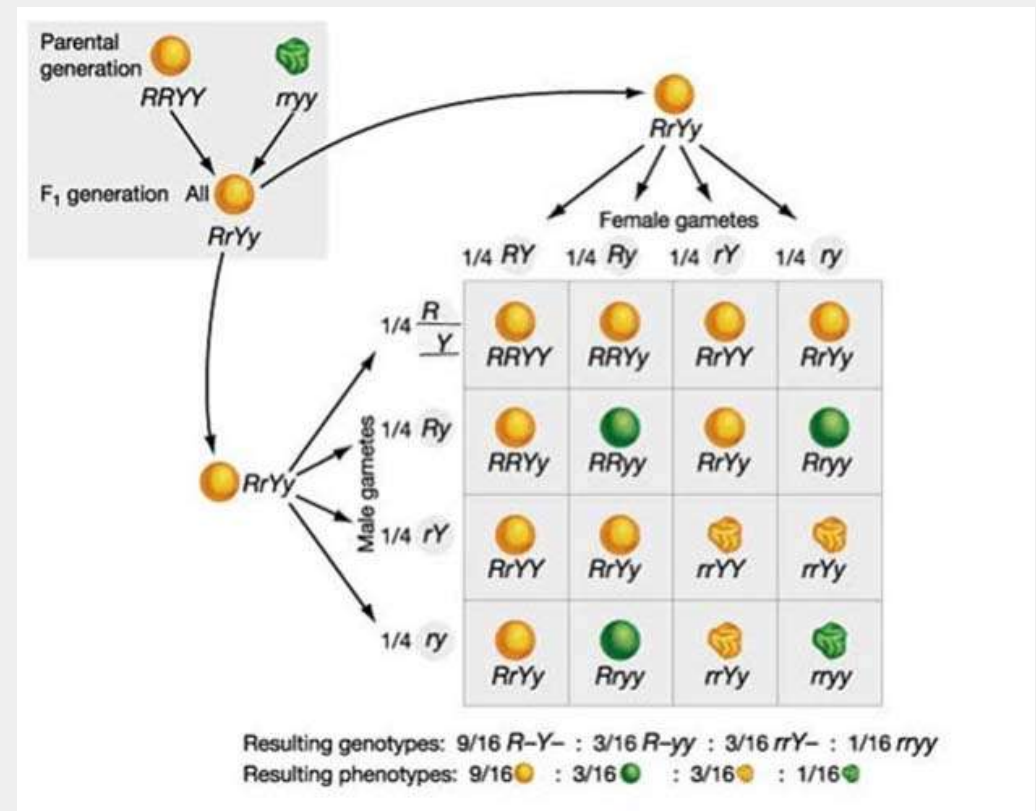


Law of Segregation



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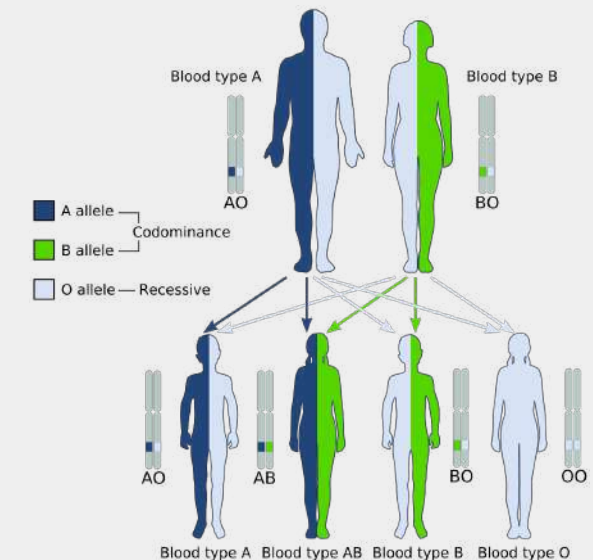
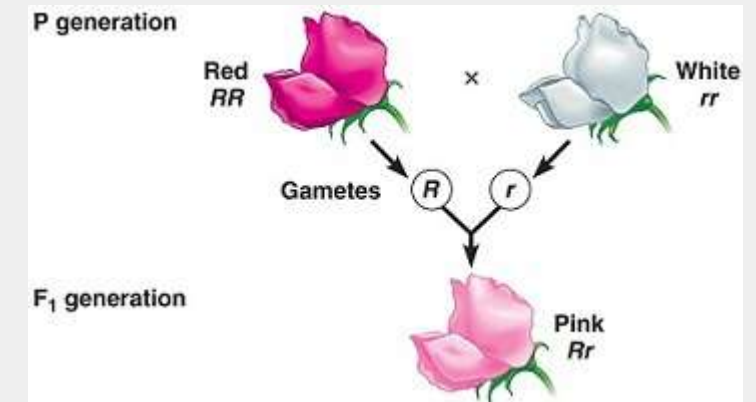
Law of Independent Assortment



Non-Mendelian: Incomplete vs Codominance

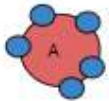
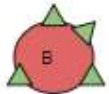
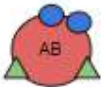


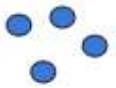

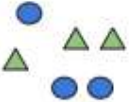




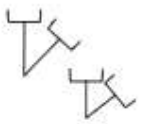


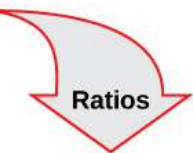
- **Incomplete dominance:**
 - Many genes have alleles that are *neither dominant nor recessive*. Genes with these kinds of alleles give rise to a *variety* of different *phenotypes*.
 - In **incomplete dominance** (non-Mendelian crossing), a condition in which a trait in an individual is intermediate between the phenotype of the individual's two parents because the dominant allele is unable to express itself fully
 - Ex: Snapdragons
 - RR = red flower color
 - Rr = pink flower color
 - rr = white flower color
- **Codominance:**
 - **Codominance** - a condition in which both alleles for a gene are fully expressed
 - In Codominance, neither allele is dominant or recessive, nor do the alleles blend in the *phenotype*.
 - Ex: *Human Blood Type*



Codominance - Human Blood Types



	Blood Type A	Blood Type B	Blood Type AB	Blood Type O	Rh+	Rh-
Red Blood Cells						
Antigens				N/A		N/A
Antibodies			N/A		N/A	

Codominant Cross	
I^A $I^A I^A$ A $I^A I^B$ AB I^B $I^B I^B$ B	I^B $I^A I^B$ AB $I^B I^B$ B
 Ratios	
Genotypes	Genotype ratio
AA	1
AB	2
BB	1







Codominance & Multiple Alleles



- **Multiple Alleles:**

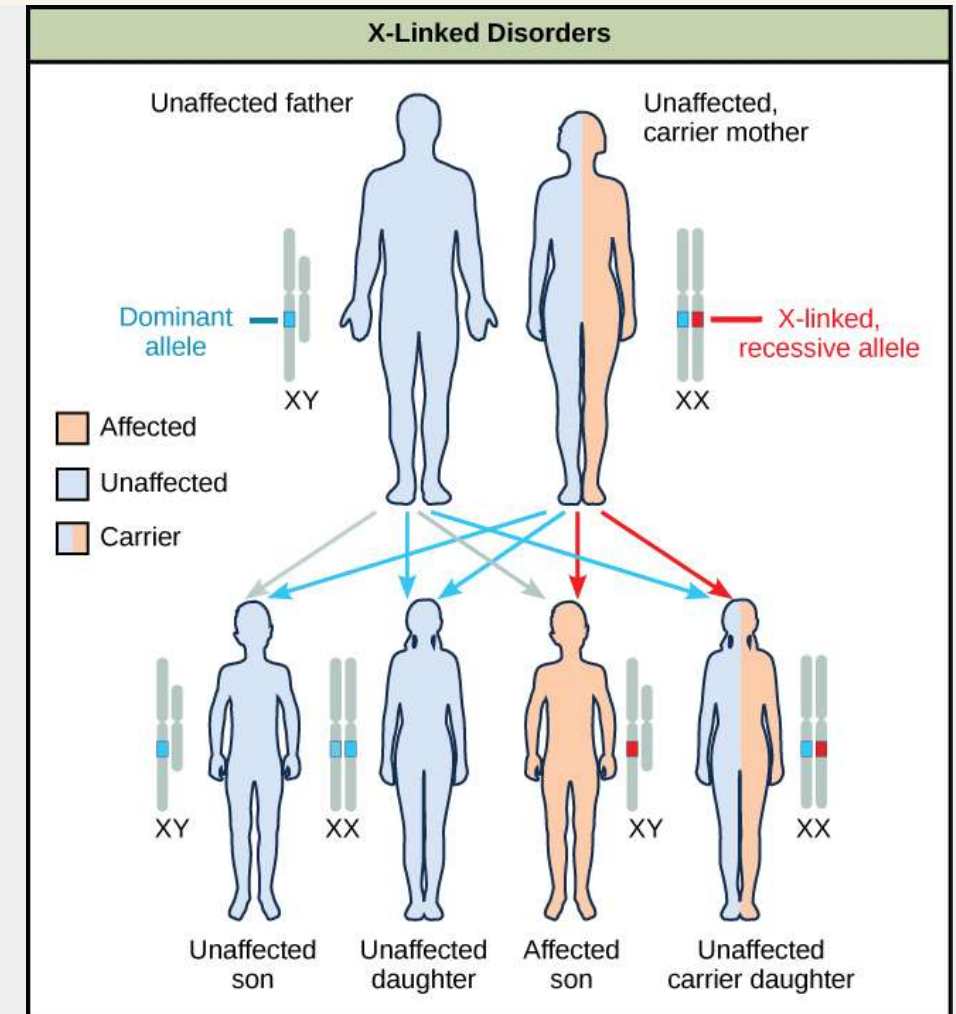
- For some genes, there can be more than *two alternative alleles* at the *single gene locus*.
- A *diploid* individual can inherit just two of the *possible alleles*.
- The *ABO Blood groups* are an example of a *multiple allele* trait.
- There are *three alleles*: I^A , I^B , i
- The I^A and (A blood type) and I^B (B blood type) alleles are *codominant*.

Allele			
C	C^{ch}	C^h	c
Genotype			
CC	$C^{ch}C^{ch}$	C^hC^h	cc
Phenotype			
WILD TYPE: Brown fur	CHINCHILLA: Black-tipped white fur	HIMALAYAN: White fur with black paws, nose, ears, tail	ALBINO: White fur
			

X-Linked / Sex Linked Traits



- **X - Linked / Sex-Linked Traits**
 - **Sex linked Traits** - *a trait that is determined by a gene found on one of the sex chromosomes, such as the X chromosomes or the Y chromosomes in humans*
 - Ex: *Red-Green Color Blindness*



Pedigree



- **Pedigree:**

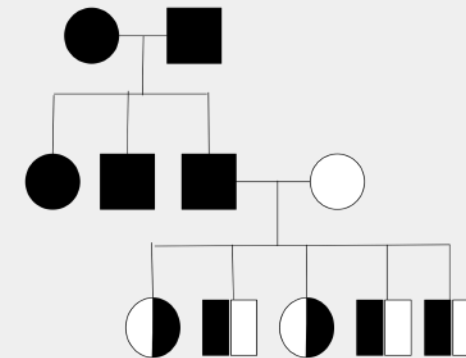
- **Pedigree:** *a diagram that shows the occurrence of a genetic trait in several generations of a family*
 - A pedigree is a family record that shows how a trait is inherited over several generations.
 - Biologists discover how traits are inherited by studying phenotypes among members of the same species from generation to the next.
 - When analyzing pedigrees, biologists find that certain phenotypes are usually repeated in predictable patterns from one generation to the next.
 - These patterns are called patterns of inheritance.
 - Individuals who have one copy of a recessive autosomal allele are called carriers.

Generation

I

II

III

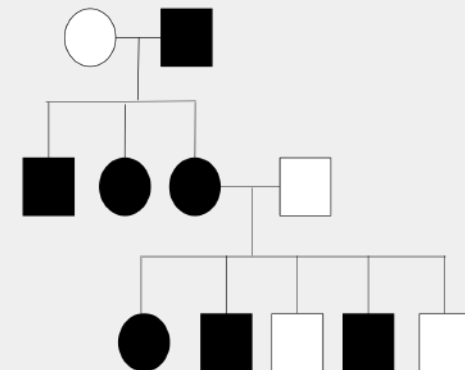


Generation

I

II

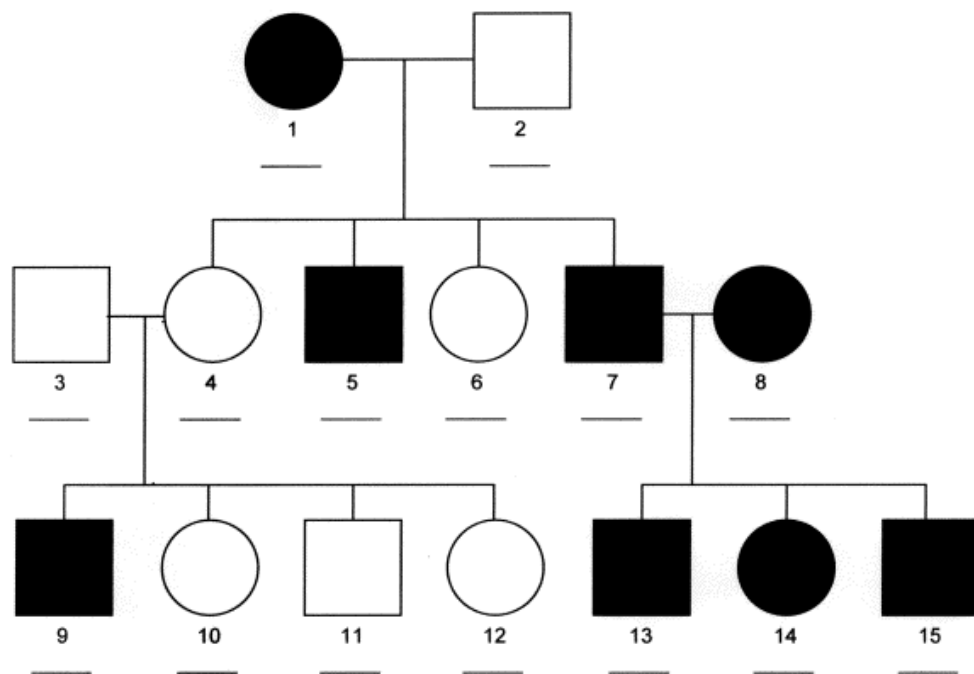
III



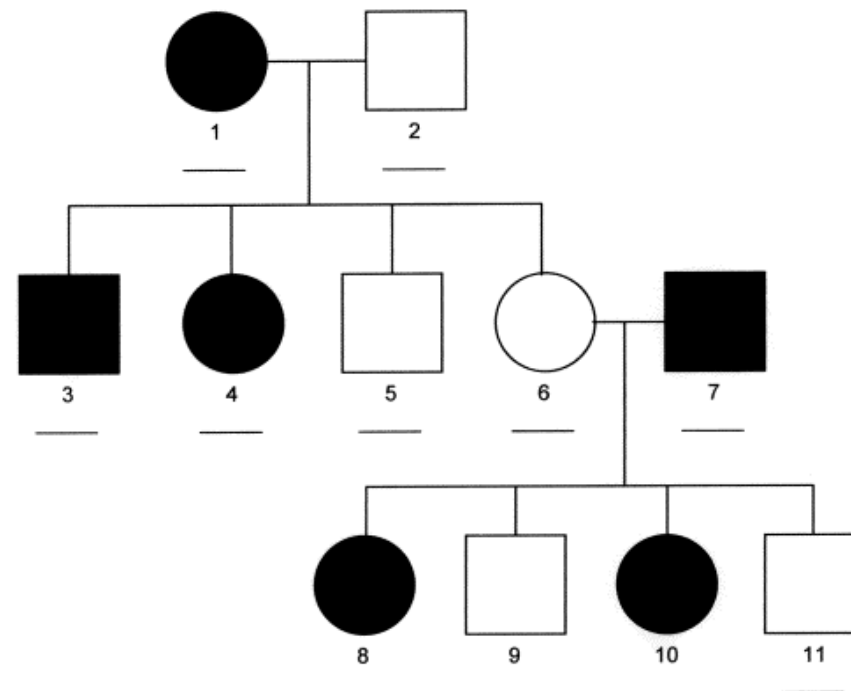
Pedigree - X-Linked



X-linked
Recessive



X-linked
Dominant



Genetic Disorders - Sickle Cell



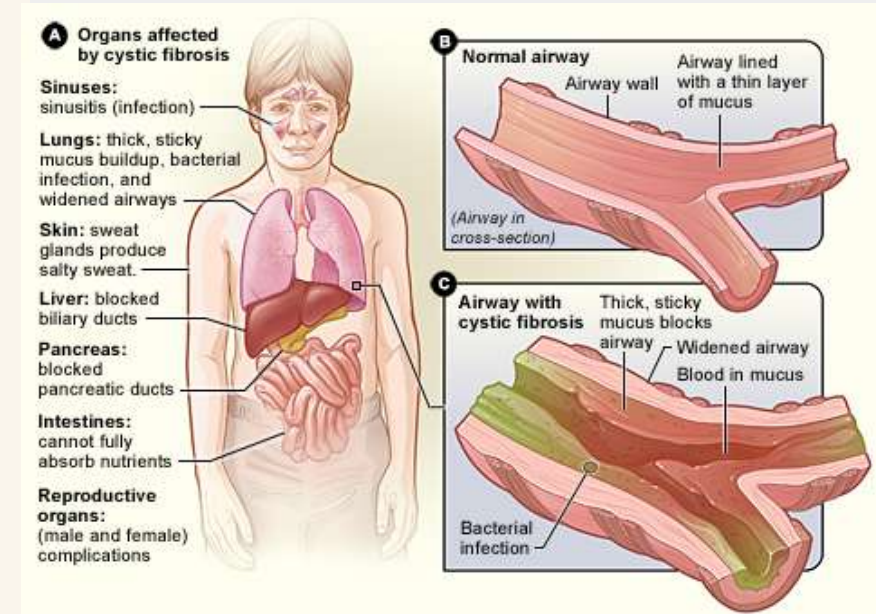
- **Sickle cell anemia:** is a disease passed down through families in which red blood cells form an abnormal sickle or crescent shape. Red blood cells carry oxygen to the body and are normally shaped like a disc.
 - **Causes, incidence, and risk factors**
 - Sickle cell anemia is caused by an abnormal type of hemoglobin called hemoglobin S. Hemoglobin is a protein inside red blood cells that carries oxygen.
 - ★ Hemoglobin S changes the shape of red blood cells. The red blood cells become shaped like crescents or sickles.
 - ★ The fragile, sickle-shaped cells deliver less oxygen to the body's tissues.
 - ★ They can also get stuck more easily in small blood vessels, as well as break into pieces that can interrupt healthy blood flow. These problems decrease the amount of oxygen flowing to body tissues even more.
 - Sickle cell anemia is inherited from both parents. If you inherit the sickle cell gene from only one parent, you will have sickle cell trait. People with sickle cell trait do not have the symptoms of sickle cell anemia.
 - Sickle cell disease is much more common in people of African and Mediterranean descent. It is also seen in people from South and Central America, the Caribbean, and the Middle East.



Genetic Disorders - Cystic Fibrosis



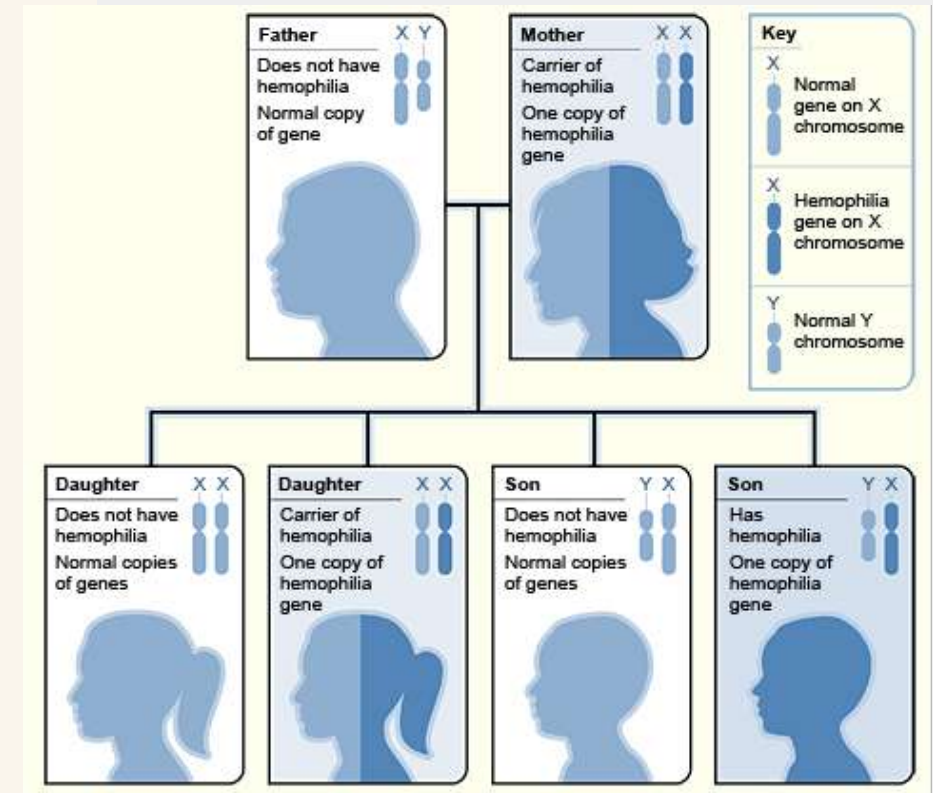
- **Cystic Fibrosis:** Cystic fibrosis is a disease passed down through families that causes thick, sticky mucus to build up in the lungs, digestive tract, and other areas of the body. It is one of the most common chronic lung diseases in children and young adults. It is a life-threatening disorder.
- **Causes, incidence, and risk factors**
 - Cystic fibrosis (CF) is caused by a defective gene which causes the body to produce abnormally thick and sticky fluid, called mucus. This mucus builds up in the breathing passages of the lungs and in the pancreas, the organ that helps to break down and absorb food. This collection of sticky mucus results in life-threatening lung infections and serious digestion problems. The disease may also affect the sweat glands and a man's reproductive system.
 - Millions of Americans carry the defective CF gene, but do not have any symptoms. That's because a person with CF must inherit two defective CF genes -- one from each parent. An estimated 1 in 29 Caucasian Americans have the CF gene. The disease is the most common, deadly, inherited disorder affecting Caucasians in the United States. It's more common among those of Northern or Central European descent.
 - Most children with CF are diagnosed by age 2. A small number, however, are not diagnosed until age 18 or older. These patients usually have a milder form of the disease.



Genetic Disorders - Hemophilia



- **Hemophilia:** refers to a group of bleeding disorders in which it takes a long time for the blood to clot.
 - **Causes, incidence, and risk factors**
 - When you bleed, the body launches a series of reactions that help the blood clot. This is called the coagulation cascade. The process involves special proteins called coagulation factors. When one or more of these clotting factors are missing, there is usually a higher chance of bleeding.
 - Hemophilia is caused by a lack of enough factor VIII or IX. In most cases, hemophilia is passed down through families (inherited). It most often affects males.



Genetic Disorders - Huntington's



- **Huntington's disease:** is a disorder passed down through families in which nerve cells in certain parts of the brain waste away, or degenerate.
 - **Causes, incidence, and risk factors**
 - Huntington's disease is caused by a genetic defect on chromosome 4. The defect causes a part of DNA, called a CAG repeat, to occur many more times than it is supposed to. Normally, this section of DNA is repeated 10 to 28 times. But in persons with Huntington's disease, it is repeated 36 to 120 times.
 - As the gene is passed down through families, the numbers of repeats tend to get larger. The larger the number of repeats, the greater your chance of developing symptoms at an earlier age. Therefore, as the disease is passed along in families, symptoms develop at younger and younger ages.
 - There are two forms of Huntington's disease.
 - * The most common is adult-onset Huntington's disease. Persons with this form usually develop symptoms in their mid-30s and 40s.
 - * An early-onset form of Huntington's disease accounts for a small number of cases and begins in childhood or adolescence.
 - If one of your parents has Huntington's disease, you have a 50% chance of getting the gene for the disease. If you get the gene from your parents, you will develop the disease at some point in your life, and can pass it onto your children. If you do not get the gene from your parents, you cannot pass the gene onto your children.





Thank you!

Do you have any questions?

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