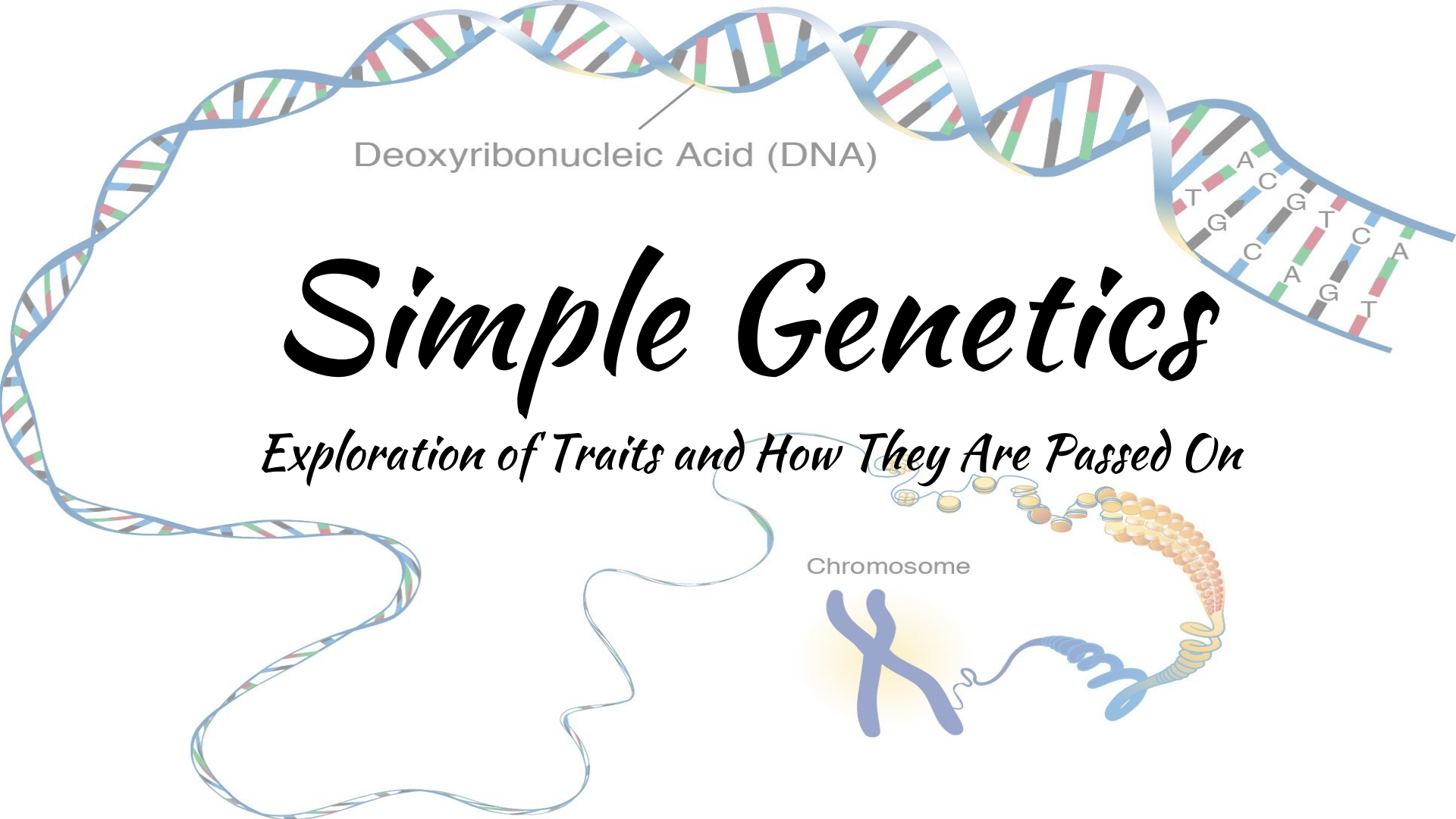


Simple Genetics

Exploration of Traits and How They Are Passed On

Putting together the Pieces...

Before completing a punnett square you must know the following

- **Allele** - a variant form of a gene
    - humans inherit one allele from each parent that determines a specific trait
    - alleles determine, if you have curly or straight hair, if you are tall or short
- **Dominant Allele** - shows their effect even if there is only one copy of the allele (dominant, strong, always there), represented by capital letters (A)
- **Recessive Allele** - will only show up if there is no dominant allele, (Recessive, weak, can be covered), represented by a lowercase letter (a)
- Dominant and Recessive alleles pair to determine a trait, (ex. AA, Aa, aa)

Genetic Traits

Attached ear lobe

Free ear lobe

Widow's peak

No widow's peak

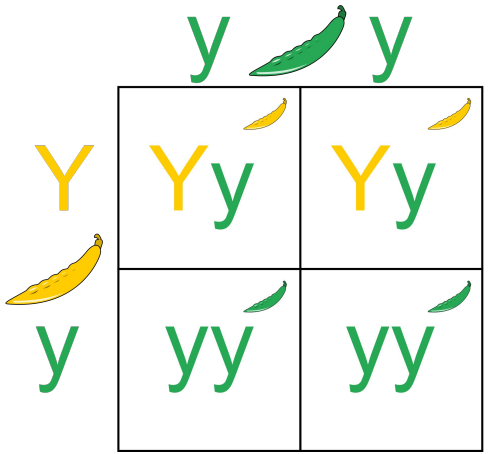
Cleft chin

No cleft chin

Punnett Squares

In order to determine the traits that a person might inherit, we use a simple diagram called a **Punnett Square**.

- They give us the probability of an offspring having or carry a specific trait.



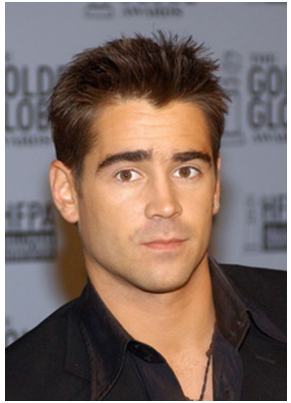
Homozygous vs. Heterozygous

So, alleles pair together (AA, aa or Aa)

- Homozygous - same alleles, identical, either both dominant or both recessive
  - Homo means same
  - A person with ¨AA¨ would be called homozygous dominant
  - A persons with ¨aa¨ would be called homozygous recessive
  - Ex. Let's say ¨A¨ represents the allele for a widow's peak,
  - ¨a¨ represents the allele for a straight hairline
- Heterozygous - alleles are different, one dominant (capital) and one recessive (lowercase), the alleles are heterozygous
  - ¨Hetero¨ means different
    - A person with ¨Aa¨ would be heterozygous
- Would the have a widow's peak or a straight hairline?

Phenotype and Genotype

All of the allele combinations tell us the phenotype and genotype of each person.



- **Phenotype** - the trait shown outwardly, Physical Appearance
  - Ex. Seeing a person's widow's peak.

- **Genotype** - the genetic makeup of the trait, Genetic Makeup
  - Ex. Aa (heterozygous), they can pass on either a dominant allele (A) or a recessive one (a)

Let's Practice

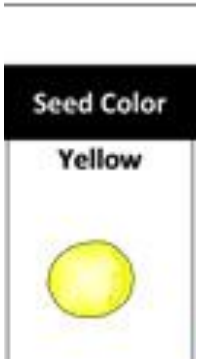
- Recessive or Dominant
  - A
  - b
  - B
- Heterozygous or Homozygous
  - AA
  - Bb
  - cc
- Phenotype or Genotype
  - She is homozygous recessive (rr).
  - She has blue eyes.

Let's Practice Answers

- Recessive or Dominant
  - A - Dominant
  - b - Recessive
  - B - Dominant
- Heterozygous or Homozygous
  - AA - Homozygous
  - Bb - Heterozygous
  - cc - Homozygous
- Phenotype or Genotype
  - She is homozygous recessive (rr). - Genotype
  - She has blue eyes. - Phenotype

Building a Punnett Square

- Consider the following alleles for pea plant color:
  - AA (Yellow-homozygous dominant)
  - Aa (Yellow-heterozygous)
  - aa (Green-homozygous recessive)



A

a

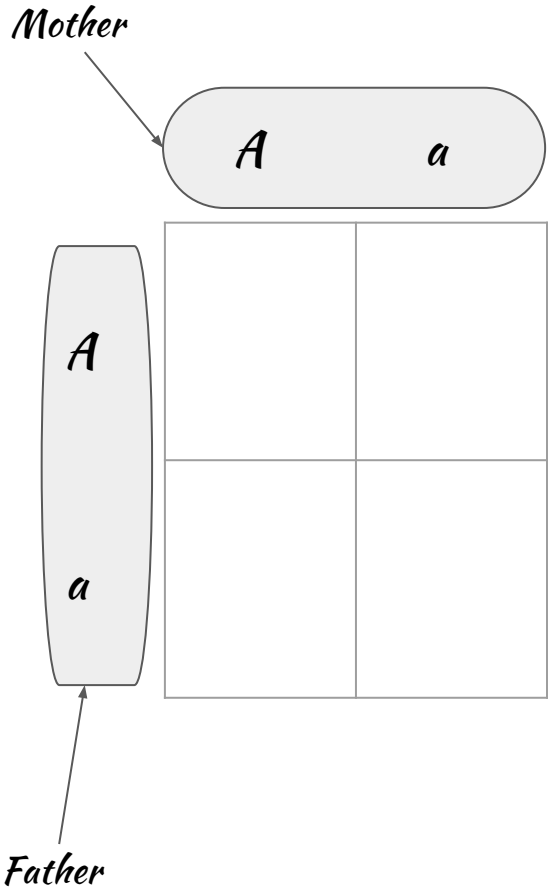
- Let's say that you have two pea plants, both heterozygous yellow (Aa), and you wanted to know what color plants would be produced. How would you find out?
  - Set up and solve a Punnett Square

Building a Punnett Square

- What we know: Both parents are Aa

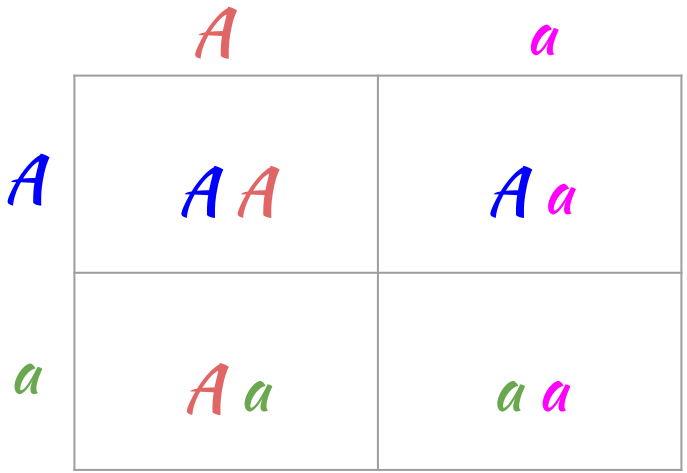
- So, we will cross the mother and father as Aa x Aa

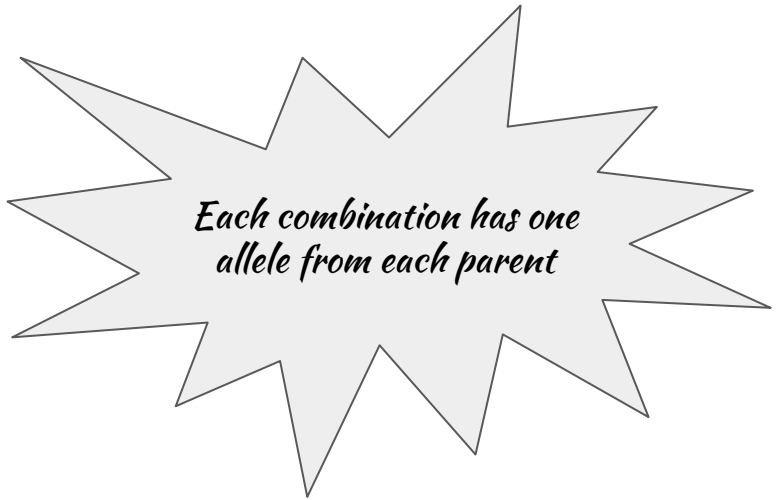
Either way it is setup - the results will be the same



Building a Punnett Square, continued

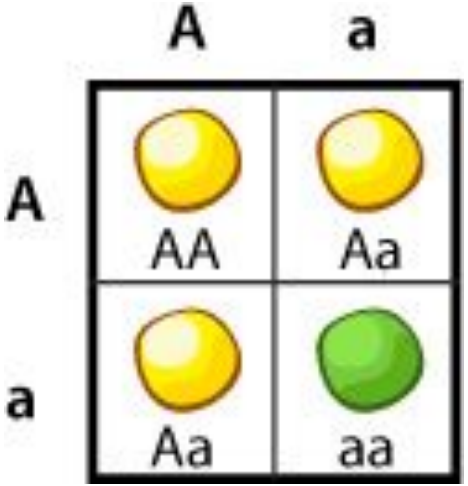
- Carrying out the cross
  - Father's letters are crossed to the right
  - Mother's alleles cross down , filling in the squares to complete the allele





Interpreting the Final Square

- Once the square is finished, we have to determine ratios for the offspring:
  - Genotype
  - Phenotype



Genotype: 1 : 2 : 1

- 1 AA, 2 Aa and 1 aa

Phenotype: 3 : 1

- 3 Yellow and 1 Green

***Note: Each ratio is equally probable for each offspring, each offspring has the same chance of inheriting the traits!

Practice: Complete the following

- Monohybrid Cross of
  - Heterozygous Tall Male and a Recessive Short Female
  - Tall - T
  - Short - t

Genotype:

Phenotype:

Practice: Complete the following

W – long whiskers

w – short whiskers

- What is the probability of producing offspring that have short whiskers from a cross of two long whiskered seals, one homozygous dominant and one heterozygous?

Genotype:

Phenotype:

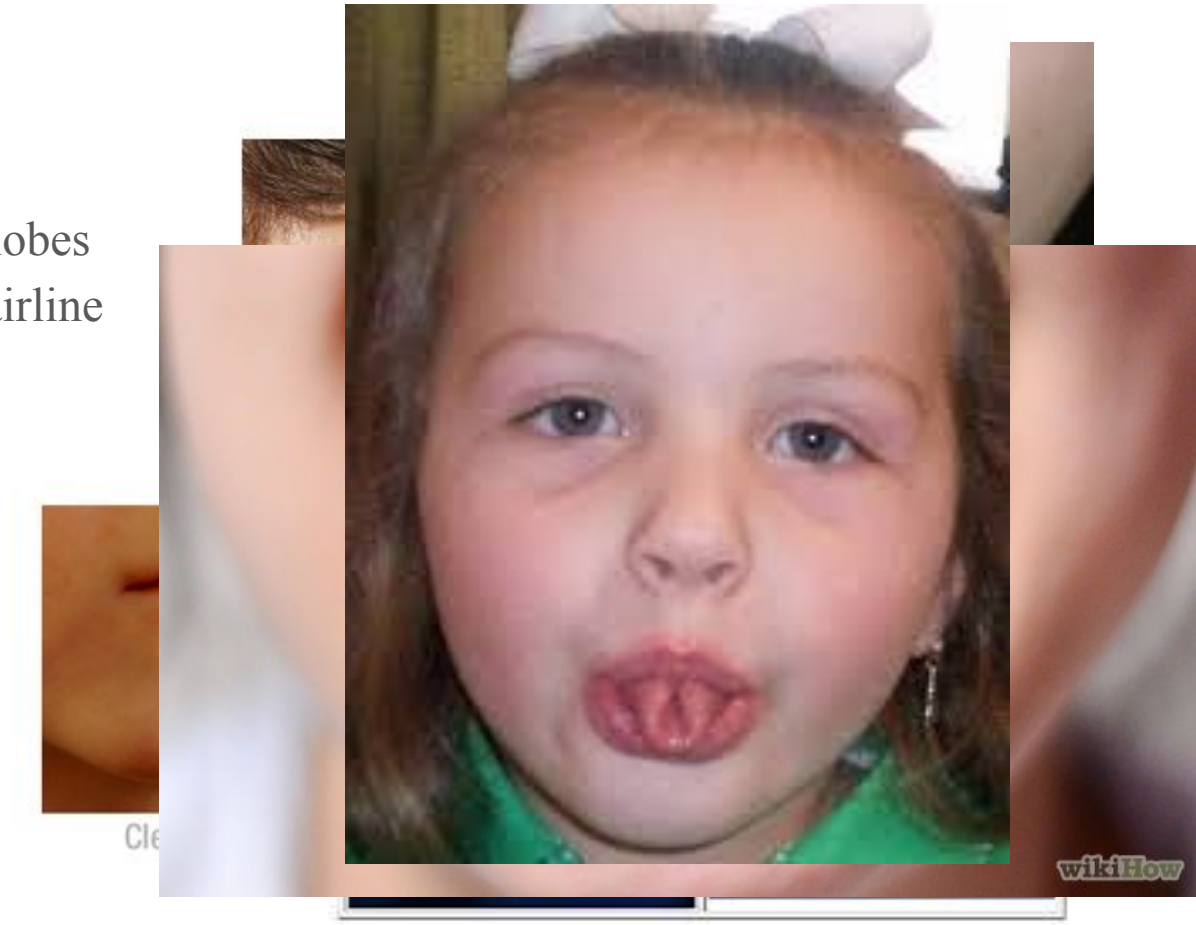
Traits

- Some traits are controlled by more than one pair of genes, and so present a wide range of phenotypes
  - (ex. skin, hair, and eye color)

- All Traits depend on both genetic and environmental factors
  - Heredity determines your traits, but the environment may play a role in how they act

Traits

- Attached or Unattached Earlobes
- Widow's Peak or Straight hairline
- Cleft Chin
- Vulcan Fingers
- Hitchhiker's Thumb
- Cloverleaf Tongue
- Rolling Tongue Over
- Curling Tongue



Genetic Diseases

How Diseases are Carried Through Generations

Dominant and Recessive

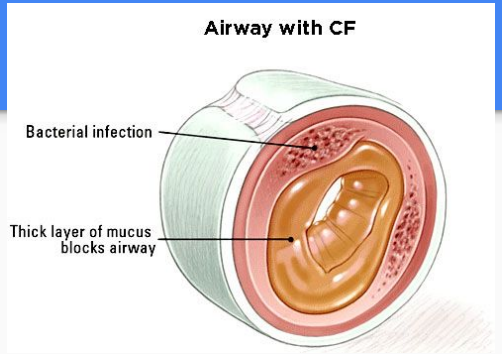
- If a disease is recessive, **both** parents have to pass on a mutated allele to the offspring
  - Those who are heterozygous (Aa) are carriers, meaning they have the mutated allele and can pass it on, but are healthy themselves
- If a disease is dominant, **only one** parent has to pass on the mutated allele for offspring to have it
  - Can a person with a dominant disease be a carrier?

NO, Why? If it is dominant it will show up in that individual

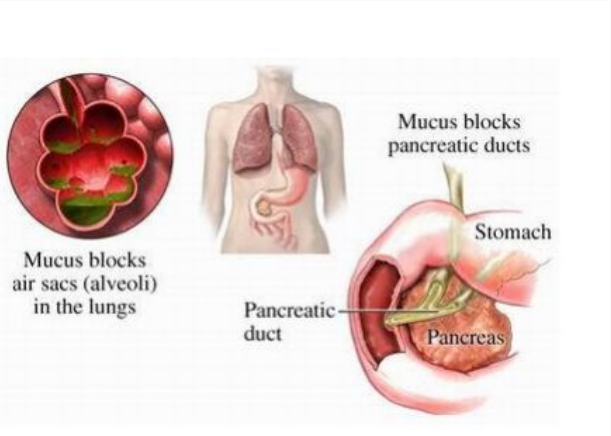
Specific Genetic Diseases

- Cystic Fibrosis
- Sickle Cell Anemia
- PKU
- Huntington's Disease

Cystic Fibrosis (cf) - Recessive

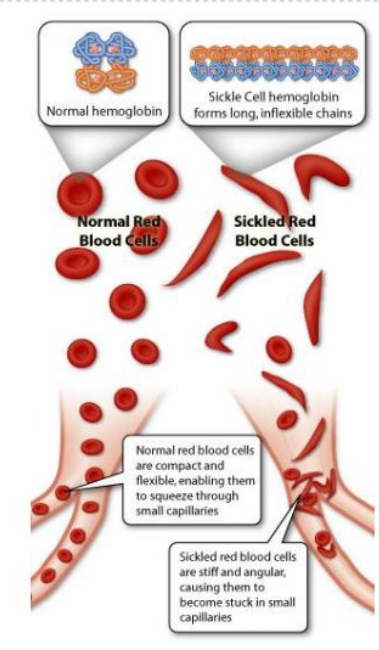


- Causes mucus to build up in the lungs and digestive system
- Those affected have difficulty breathing and poor growth
- Common in those with European Ancestry
- Life Expectancy - up to 35 years with Gene Therapy



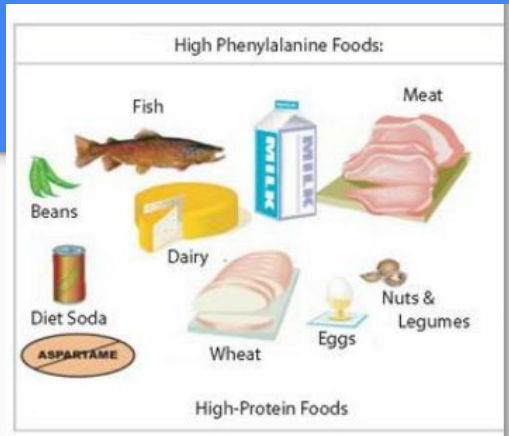
Sickle Cell Anemia (sca) - Recessive

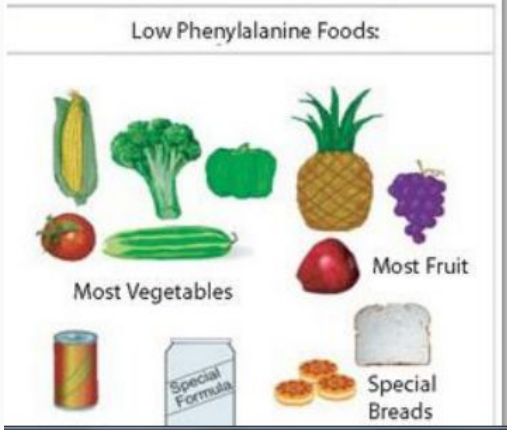
- Affects a person's red blood cells
  - Mutation causes the cells to be long and bent (sickle shaped)
- The odd shape causes the cells to get stuck in blood vessels
  - cuts off blood flow and circulation
- Common in those with African Ancestry
  - Carrieres associated with malaria resistance



Phenylketonuria (pku) - Recessive

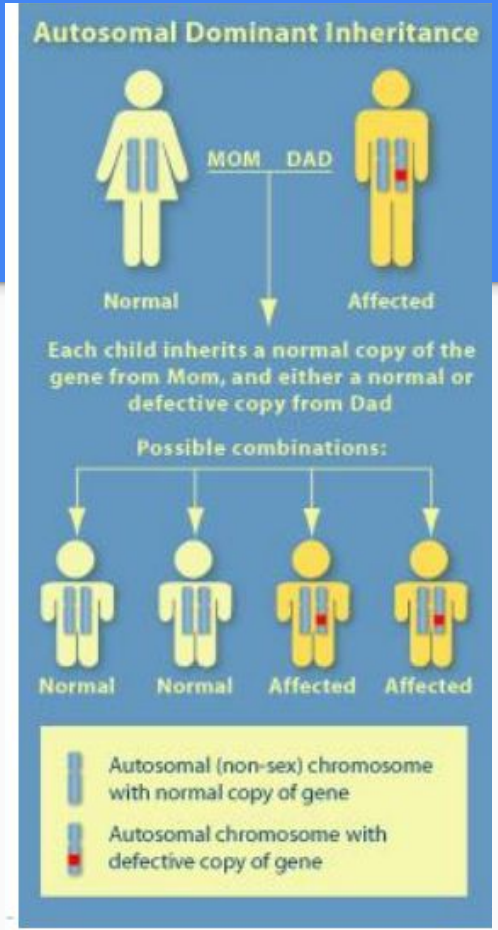
- Usually called PKU
- condition where the body is **missing the enzym**e that breaks down phenylalanine, an amino acid
- Phenylalanine builds up in the body, causes mental deterioration
  - Memory Loss
  - Identity Loss
- Affected people have to keep a **low protein** diet and have the enzyme injected regularly
  - Has to be done their entire life!





Huntington’s (H) - Dominant

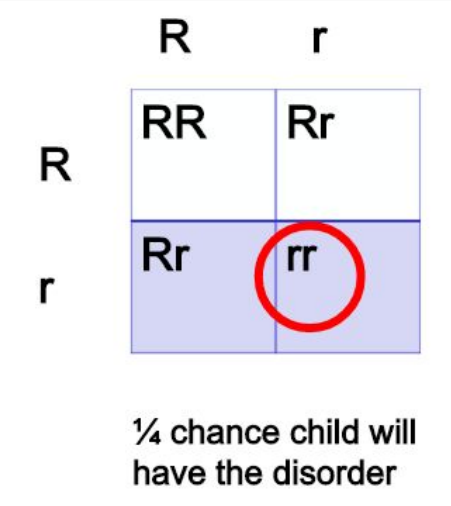
- Affects the muscles of the body
- Causes muscle function decline
  - painful, similar to muscle cramping that will not go away
- Doesn’t show signs until the person reaches mid 30’s, can take up to 20 years to cause death
  - the person has usually already had children and they have already passed the allele on
- Affects anyone!



Punnett Square: Genetic Disease

- A woman is a carrier for Cystic Fibrosis, marries a man who is also a carrier. What is the probability they will have a child with Cystic Fibrosis?

Cystic Fibrosis

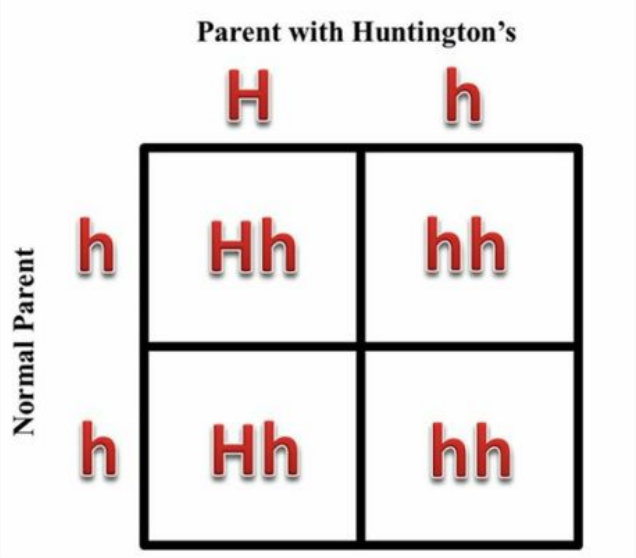


Punnett Square: Genetic Disease

- A woman is concerned that she may develop Huntington's Disease because her father has it. What is the probability that she has Huntington's? (*the mother does not have Huntington's Disease)

Huntington’s Disease

Dominant Disease

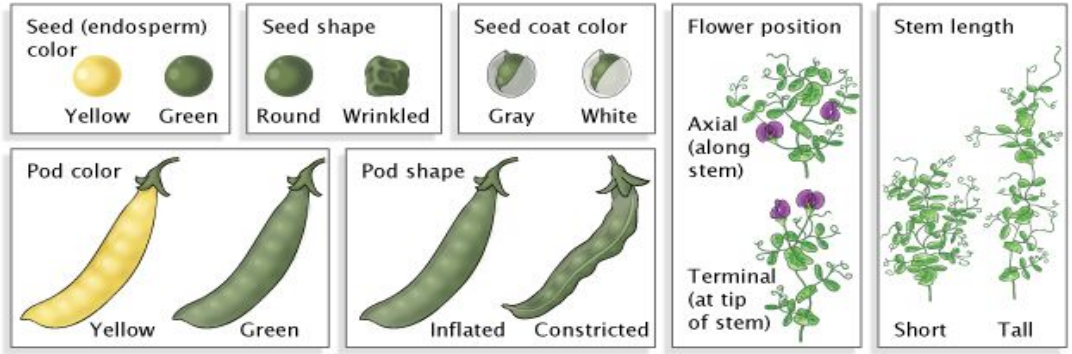


Complex Genetics

Codominance, Incomplete Dominance, Sex-Linked Traits and Blood Typing

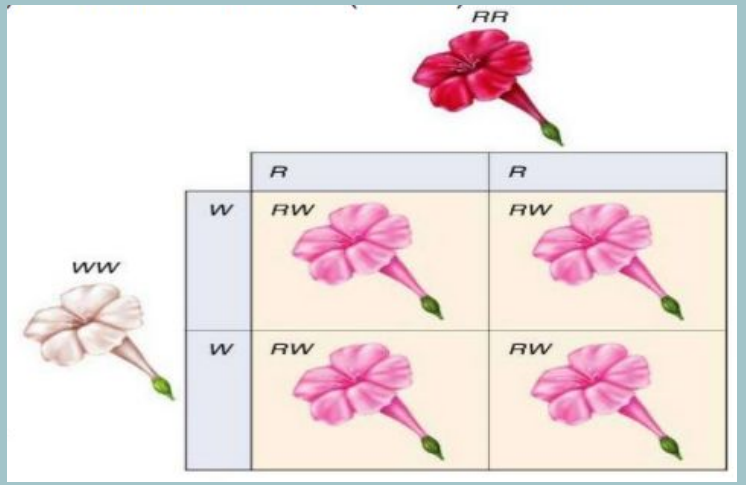
Gregor Mendel

- Father of Genetics
- Discovered that genes come in pairs and are inherited as distinct units
- tracked genes as dominant and recessive
- recognized their mathematical patterns
- Examples Pea Plants
  - Color
  - Shape
  - Seed coat



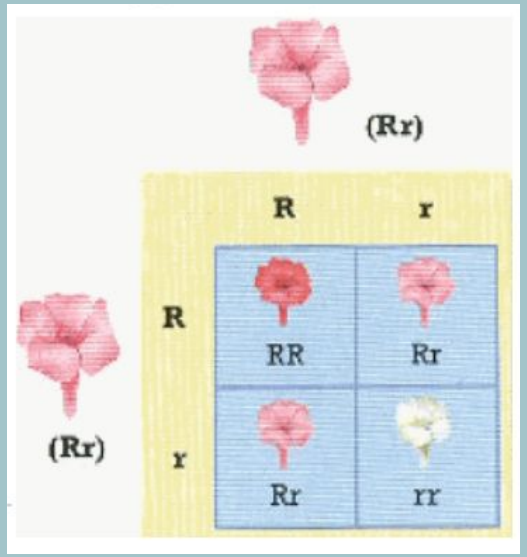
Incomplete Dominance

- Sometimes, one allele is not completely dominant over another allele
  - In this case, the offspring are **blends** of both parents!
  - The heterozygote has a unique phenotype because one gene does not dominate the other
  - Example: Red Flower (RR) X White Flower (WW), produces a Pink Flower (RW)



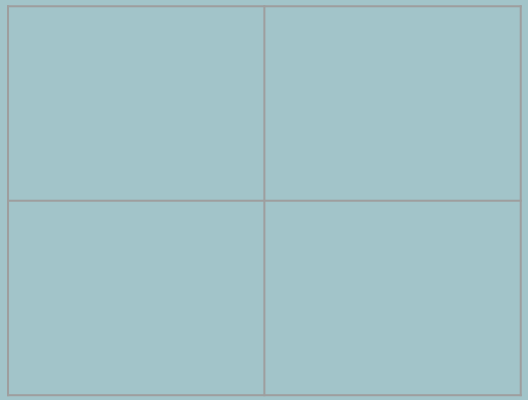
Incomplete Dominance, Continued

- During incomplete dominance, organisms who are heterozygous for a trait have the opportunity to pass on either gene to the offspring
- Therefore, a cross between two heterozygous organisms have the following results:
  - 1 RR : 2Rr: 1 rr
  - 1 Red: 2 Pink : 1 White



Example of Incomplete Dominance

- A red carnation is crossed with a white carnation and the resulting offspring are all pink (indicating incomplete dominance).
  - Cross the following: a red carnation and a white carnation



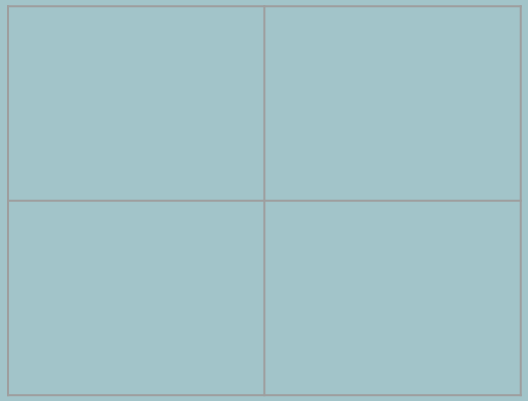
Example of Incomplete Dominance

- A red carnation is crossed with a white carnation and the resulting offspring are all pink (indicating incomplete dominance).
  - Cross the following: a pink carnation and a white carnation

	R	r
r	Rr	rr
r	Rr	rr

Cross:

- Cross the following: a pink carnation and a white carnation, give the phenotypes and genotypes
  - Rr X rr



Cross:

- Cross the following: a pink carnation and a white carnation, give the genotype and phenotype.
  - RR X rr

	R	r
r	Rr	rr
r	Rr	rr

Genotype:
2 Rr : 2 rr
1:1

Phenotype:
2 Pink : 2 White
1:1

Codominance

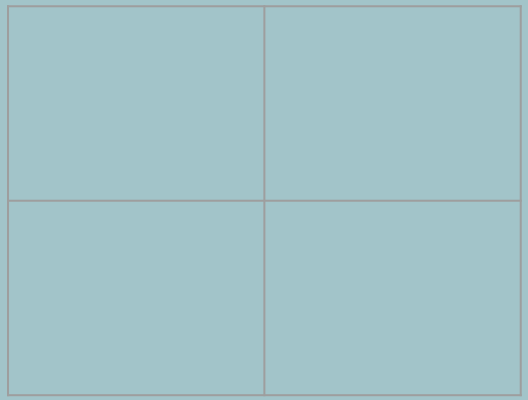
- Sometimes both alleles are equally dominant
  - Therefore, both alleles will be expressed in the phenotype
  - In this case, the offspring display the traits of both parents!
  - Example: White Cow (WW) X Brown Cow (BB) produces a White and Brown Cow (BW)


+

=

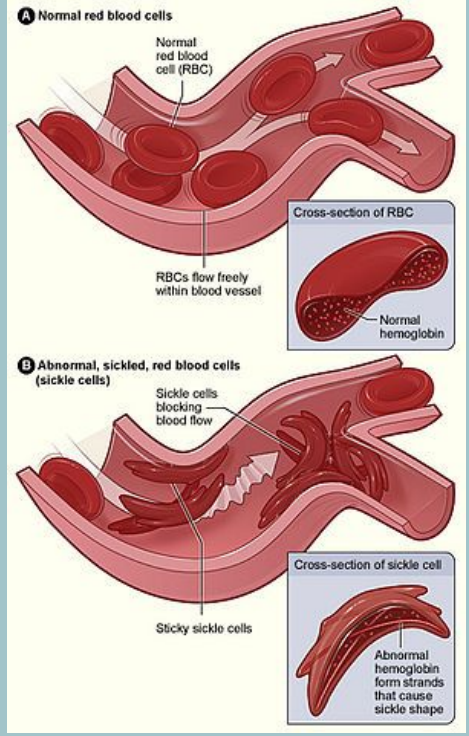

Cross:

- White Cow (WW) X Brown Cow (BB) produces a White and Brown Cow (BW)



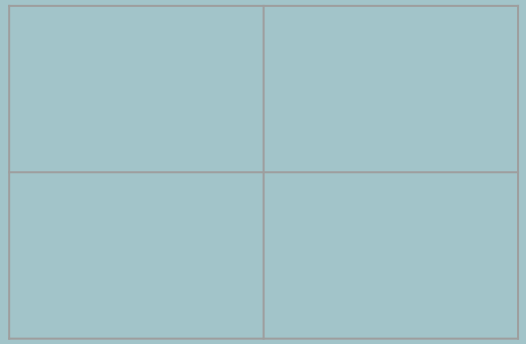
Case Study: Sickle Cell Anemia

- Although we discussed Sickle Cell Anemia has a recessive inheritance pattern, the disease does display a certain codominance
  - Those homozygous affected (aa) have misshapen cells
  - Those homozygous unaffected (AA) have normal cells
  - Those heterozygous (Aa) are unaffected carries, but can have some misshapen cells mixed in
    - This is the unique feature that makes those heterozygous individuals are resistant to malaria!



Example: Codominance

- A bird with white and blue feathers mates with another white and blue feathered bird (BW X BW).
  - What are the phenotypic ratios of the offspring?



Example: Codominance - *(BW X BW)*

- What are the phenotypic ratios of the offspring?

	B	W
B	BB	BW
W	BW	WW

Phenotype:

1 Blue

2 Blue & White

1 White

1 : 2 : 1

Blood Typing

- Human blood types include a codominance pattern
  - Humans can have type A, B, AB or O Blood
  - The alleles involved are two codominant alleles (I^A and I^B), and one recessive allele (i)

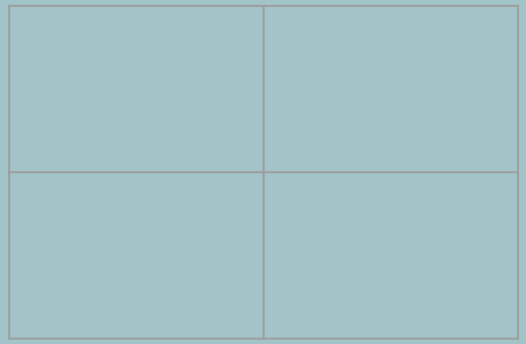
Genotype	Blood Type
I^AI^A or I^Ai	A
I^BI^B or I^Bi	B
I^AI^B	AB
ii	O

Blood Typing Information

Blood Type	Genotype		Can Receive Blood From:
A	$i^A i$ $i^A i^A$	AA AO	A or O
B	$i^B i$ $i^B i^B$	BB BO	B or O
AB	$i^A i^B$	AB	A, B, AB, O
O	$i\,i$	OO	O

Example: Blood Typing

- The father is type A homozygous and the mother is type O.
  - What are the possible blood types of their offspring?



Example: Blood Typing

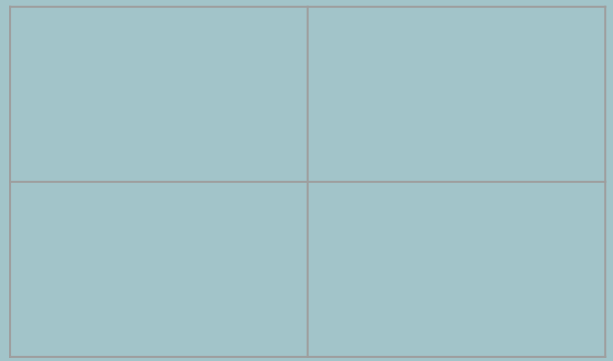
- The father is type A homozygous and the mother is type O.
  - What are the possible blood types of their offspring?

	i	i
I^A	$I^A i$	$I^A i$
I^A	$I^A i$	$I^A i$

All will have Type A Blood!

Possible Parent?

- Ralph has type B blood and his wife Rachel has type A blood, They are very shocked to hear that their baby has type O blood, and they think that a switch might have been made at the hospital. Can this baby be theirs?, Explain.



Possible Parent?

- Ralph has type B blood and his wife Rachel has type A blood, They are very shocked to hear that their baby has type O blood, and they think that a switch might have been made at the hospital.
  - Can this baby be theirs?
  - Explain.

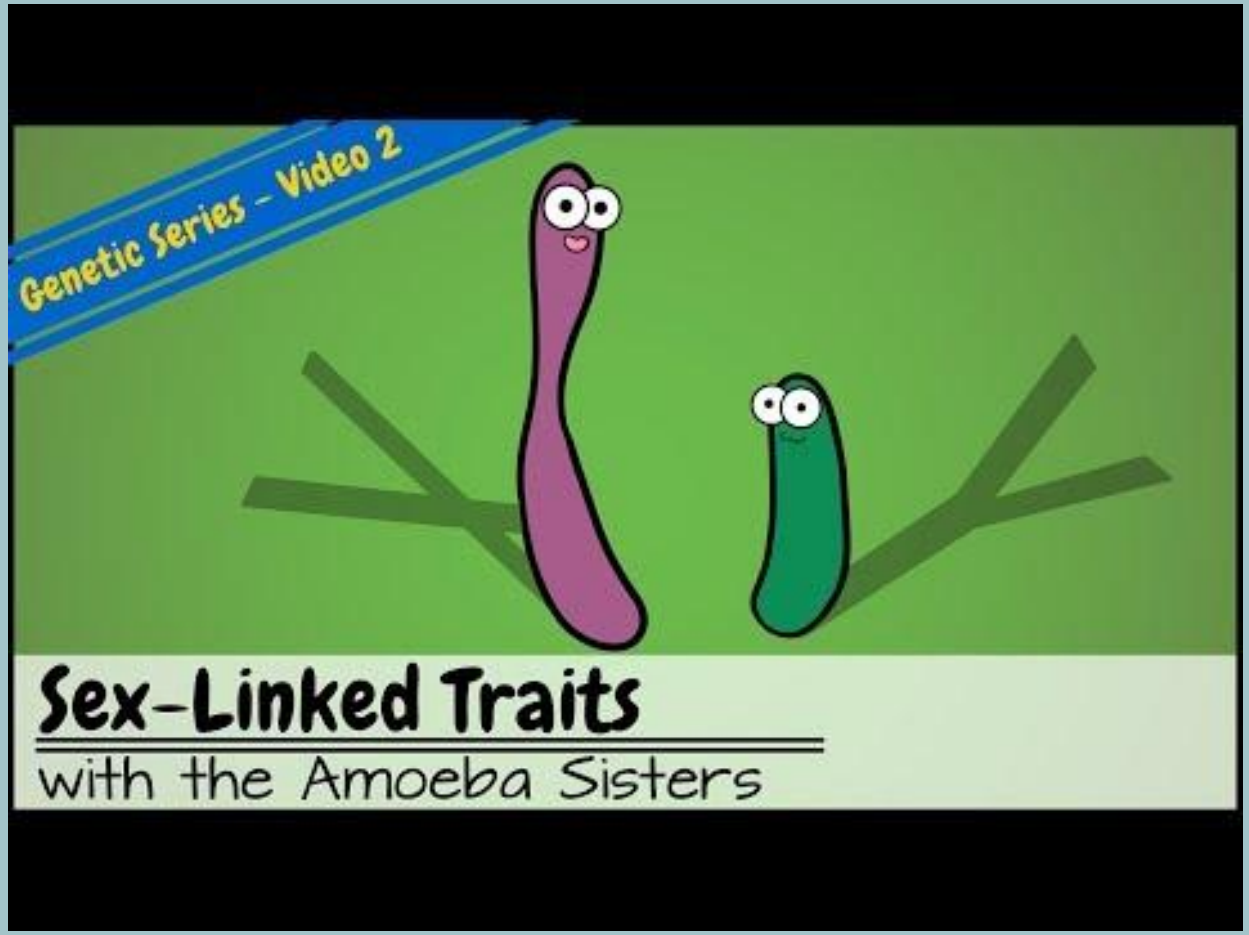
Possible Parent?

- Ralph has type B blood and his wife Rachel has type A blood, They are very shocked to hear that their baby has type O blood, and they think that a switch might have been made at the hospital.
  - Can this baby be theirs?
  - Explain.

	I^B	i
I^A	I^AI^B	I^Ai
i	I^Bi	ii

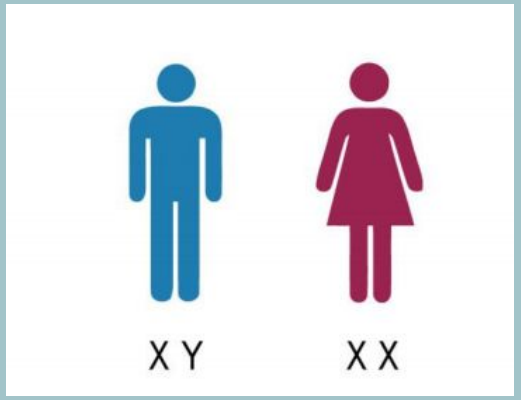
Yes the baby can be theirs!

They both carry i, and pass on the recessive allele for O Blood Type.


Genetic Series - Video 2
Sex-Linked Traits
with the Amoeba Sisters

Sex-Linked Traits

- Genes located on the sex chromosomes *(are called sex-linked traits)*
- usually found on the X chromosomes
- Because of this, males are much more likely to express a sex linked trait than females, males only have one X chromosome!



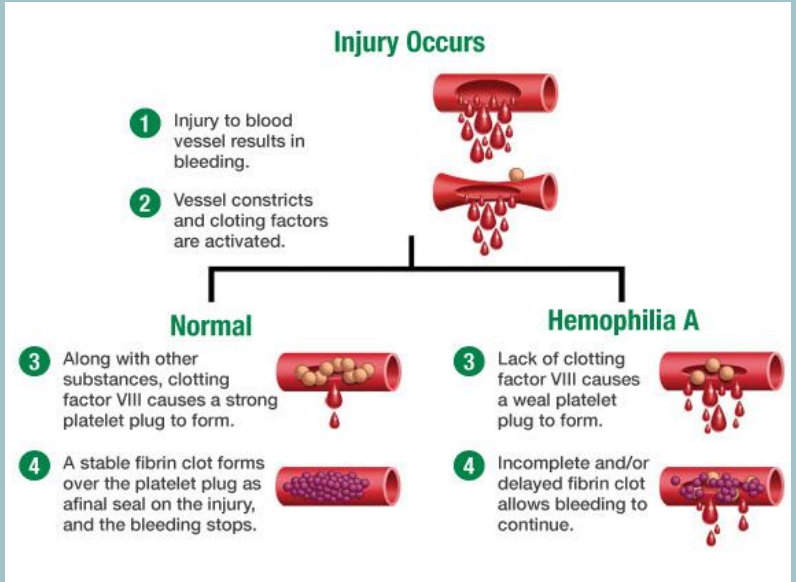
Sex-Linked Traits

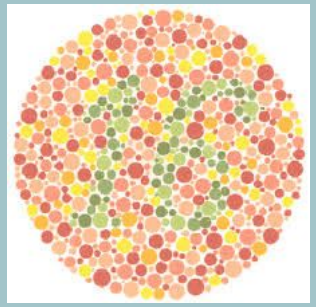
- Let's take a look at the possible alleles in a recessive sex-linked disease:
  - Female: X^AX^A, X^AX^a, X^aX^a
    - Notice that the female must inherit 2 recessive copies to display the trait. It is also possible for females to be carriers of sex-linked traits.
  - Male: X^AY, X^aY
    - Notice the males does not have a second X to help mask the trait, this means that if he inherit the recessive copy, he will display the trait

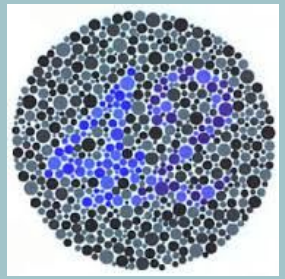
****Because of this, males are much more likely to inherit a sex-linked trait

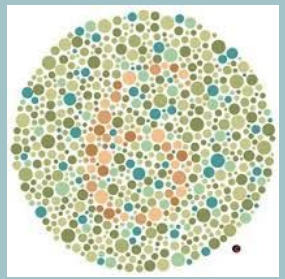
Common Sex-Linked Traits

- Hemophilia - A bleeding disorder that slows the blood clotting process
- Color Blindness - inability to see color, or perceive color differences
- Because it is X-Linked, these conditions are more common in men



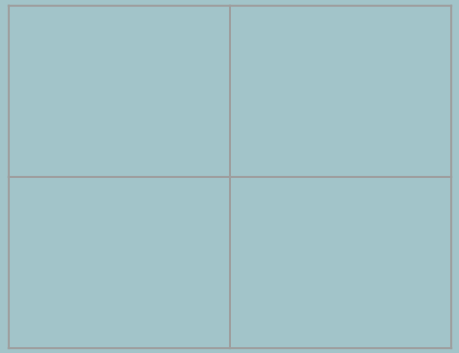






Sex-Linked Problems

- Cross a woman carrier for hemophilia to a hemophiliac man
  - What could the percentage of the females could inherit the trait?
  - Males?



FYI - Positive or Negative

- Blood types are identified with a letter and a positive or negative sign.
- The sign is linked directly to the protein, Rhesus (aka Rh) or Rh Factor
  - If you have the protein Rh you are Positive (most people do)
  - If you don't have the protein you are Negative

DOES IT MATTER - females who are Rh negative and their fetus is Rh positive, during deliver sometimes the blood is transferred and the mothers body will start to release antibodies to eliminate the foreign material.

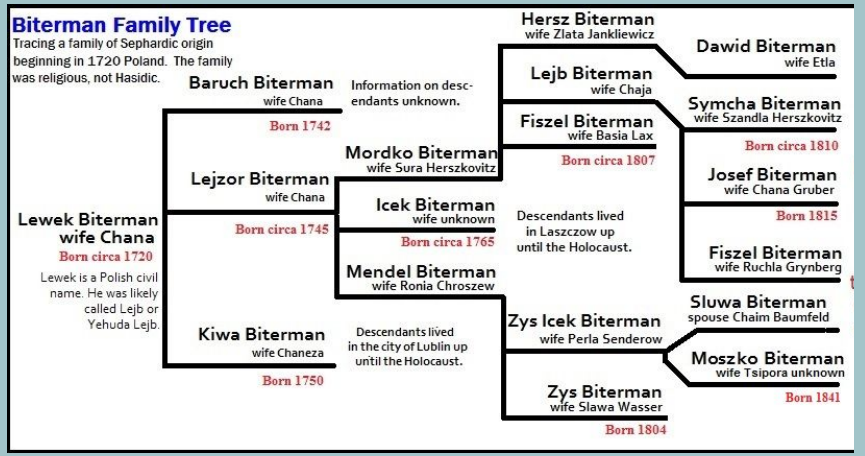
Pedigrees

Inheritance Patterns Through Generations

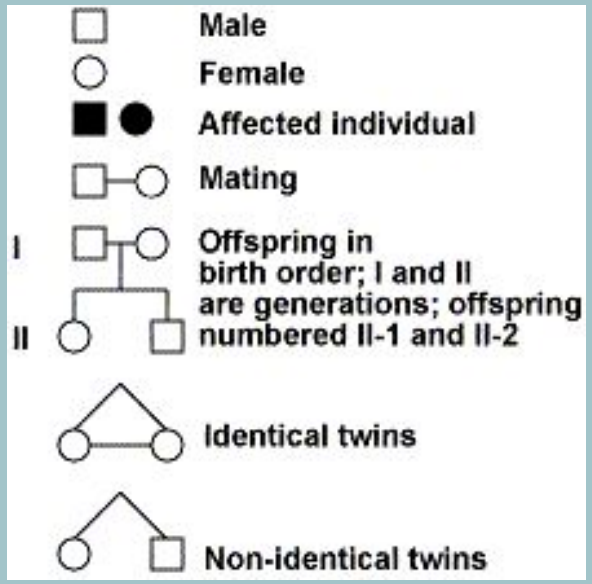
What is a Pedigree?

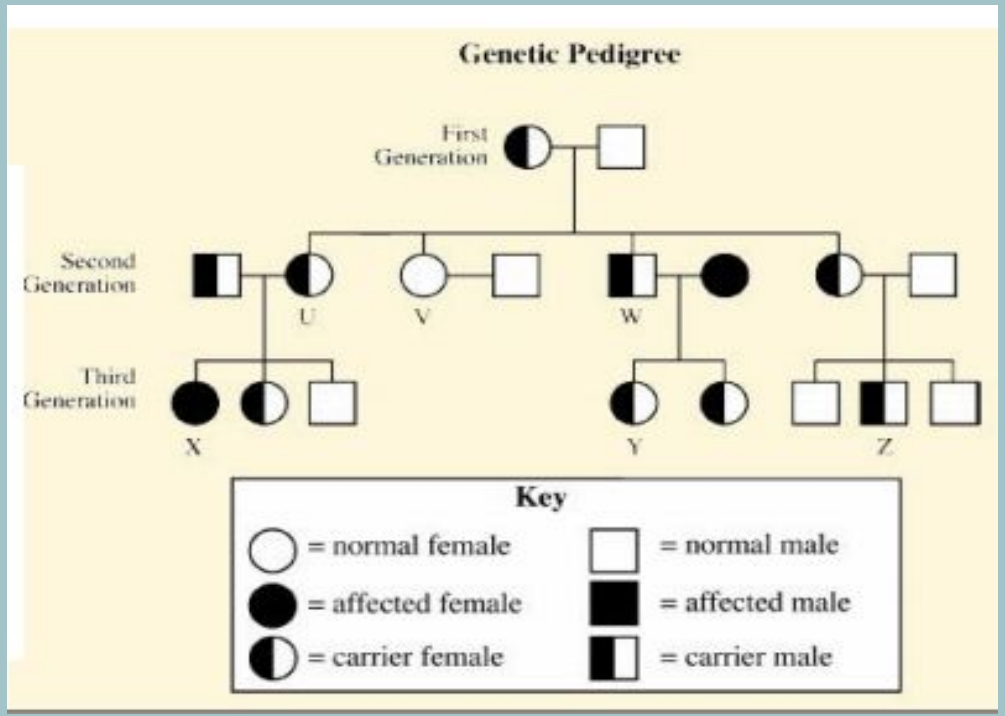
- A pedigree is a recorded ancestry of a family

- Pedigree allow us to see the genotypes and inheritance patterns through the generations of a family



Pedigree Symbols and Sample

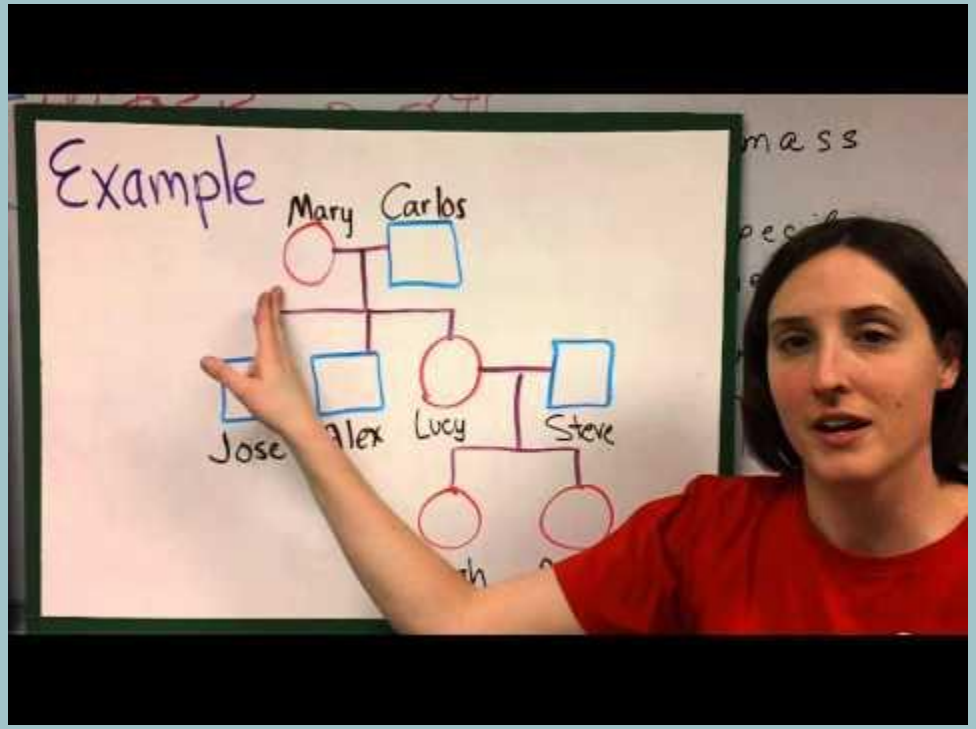




What Does a Pedigree Tell Us?

- Pedigrees can give us insight into how traits are passed through families. You can tell a lot through a pedigree.
  - Example
    - The phenotypes and genotypes of individuals
    - The inheritance patterns.
      - If more males are affected, it is likely sex linked
      - If there are no carriers then one of the parents must it, the disorder is dominant
      - If carrier parents produce some affected children, the disease must be recessive

Pedigree Charts



Pedigree: Example Problems

- What type of inheritance pattern is shown below?
- What would be the genotype of individual 1-1?