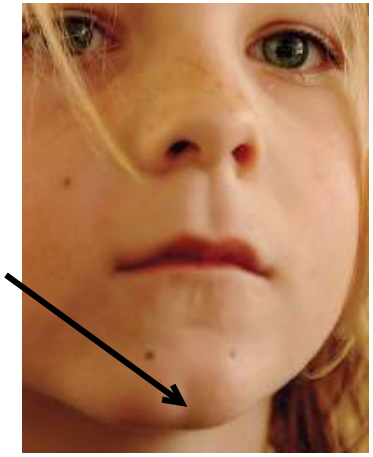
Genetics Notes

Who is Gregor Mendel? "**Father of Genetics**"

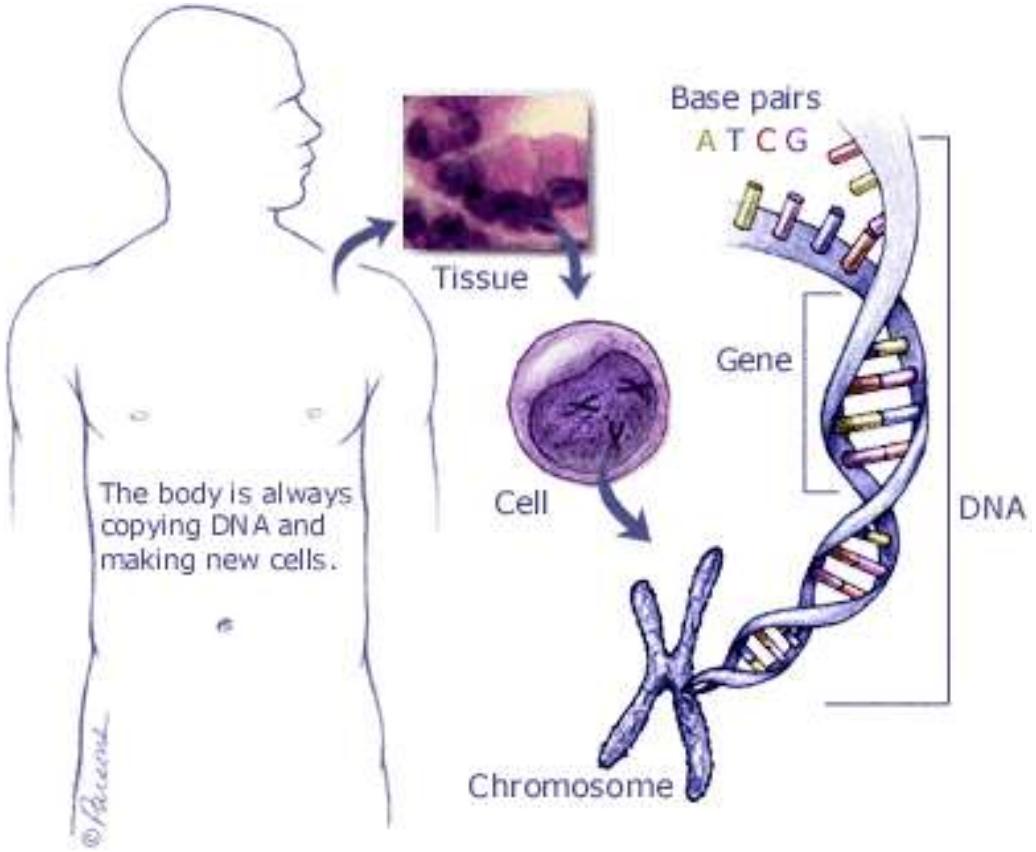
Principle of **Independent Assortment** – Inheritance of one trait has **no effect** on the inheritance of another trait

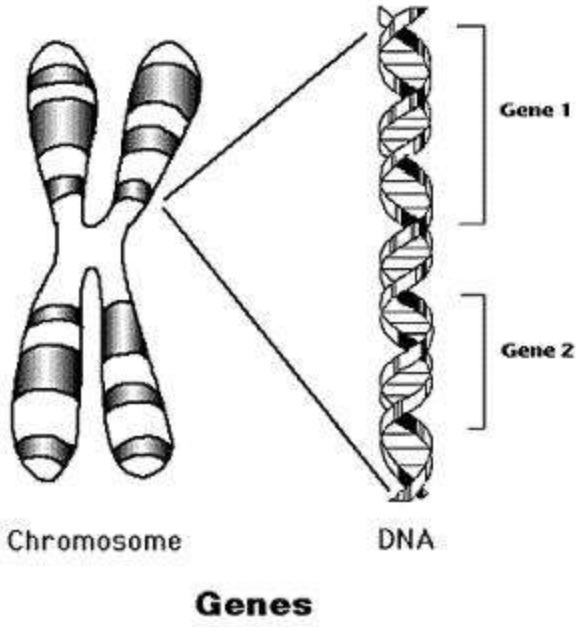
Traits

- Genetics – study of how **traits** are passed from **parent** to **offspring**

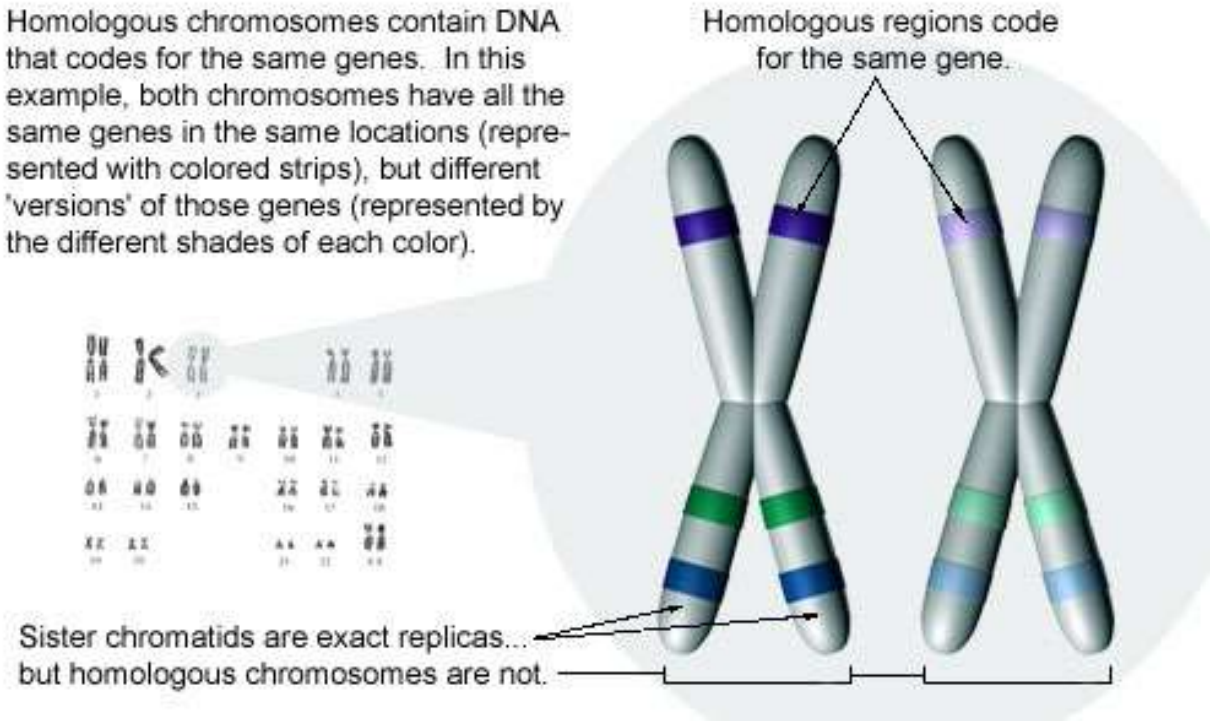


- Traits are determined by the **genes** on the **chromosomes**. A gene is a segment of **DNA** that determines a **trait**.



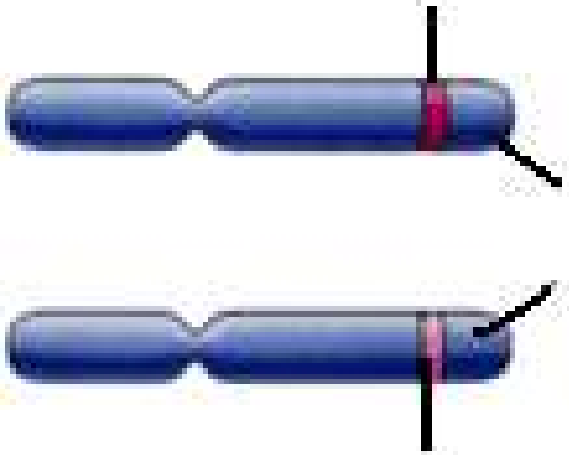


- Chromosomes come in **homologous** pairs, thus **genes** come in pairs.
  Homologous pairs – **matching** genes – one from female parent and one from male parent
- Example: Humans have 46 chromosomes or **23** pairs.
  One set from dad – 23 in **sperm**
  One set from mom – 23 in **egg**



- One pair of Homologous Chromosomes:

Gene for **eye color**
(**blue** eyes)



Homologous pair
of chromosomes

Gene for **eye color**
(**brown** eyes)

Alleles – different **genes** (possibilities) for the same **trait** –
ex: blue eyes or brown eyes

Dominant and Recessive Genes

- Gene that **prevents** the other gene from "showing" – **dominant**
- Gene that **does NOT** "show" even though it is **present** – **recessive**
- Symbol – Dominant gene – **upper** case letter – **T**
  Recessive gene – **lower** case letter – **t**

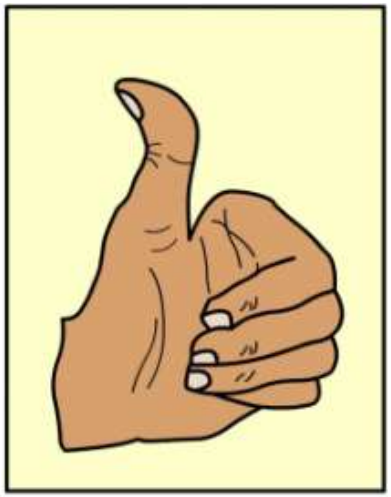
Dominant color

Recessive color

Example: Straight thumb is **dominant** to hitchhiker thumb

T = straight thumb **t** = hitchhikers thumb

(Always use the same letter for the same alleles—
No S = straight, h = hitchhiker's)



Straight thumb = TT

Straight thumb = Tt

Hitchhikers thumb = tt

* Must have **2** recessive **alleles** for a recessive trait to "**show**"

- Both genes of a pair are the same – **homozygous** or **purebred**

  TT – homozygous **dominant**

  tt – homozygous **recessive**

- One dominant and one recessive gene – **heterozygous** or **hybrid**

  Tt – heterozygous

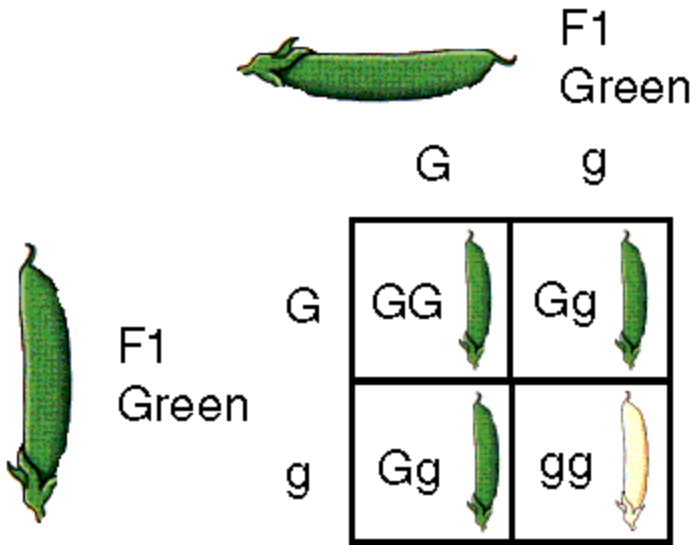
BB – Black

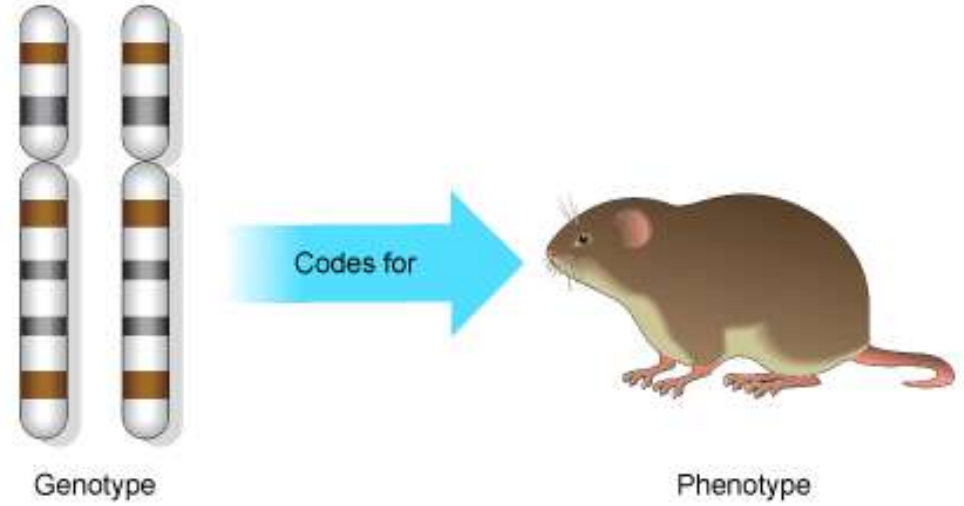
Bb – Black w/ white gene

bb – White

Genotype and Phenotype

- Combination of genes an organism has (**actual gene makeup**) – **genotype**
  Ex: TT, Tt, tt
- **Physical appearance** resulting from gene make-up – **phenotype**
  Ex: hitchhiker's thumb or straight thumb

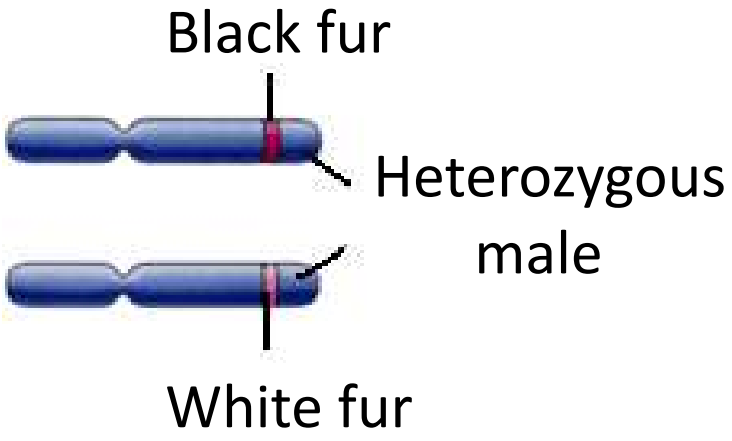


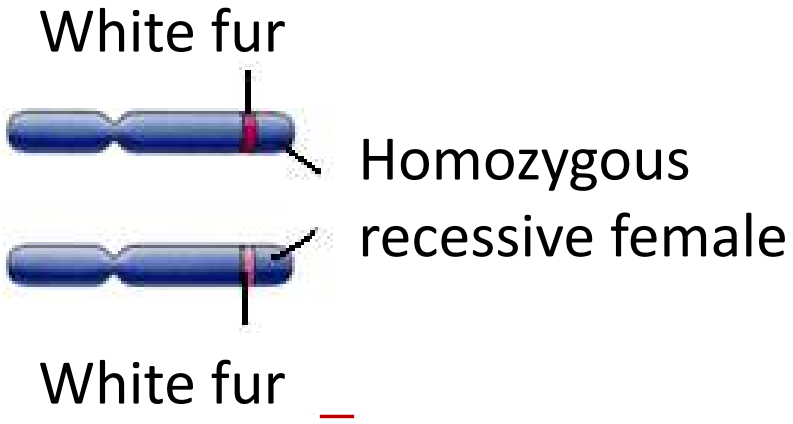


Punnett Square and Probability

- Used to **predict** the **possible** gene makeup of **offspring** – **Punnett Square**
- Example: Black fur (B) is dominant to white fur (b) in mice

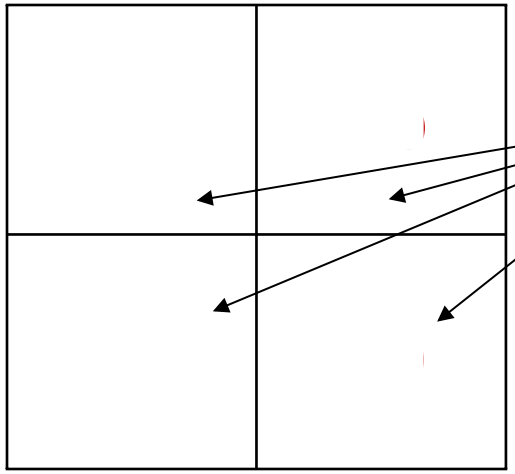
1. Cross a heterozygous male with a homozygous recessive female.





Male = ____ **X** Female = ____

Female gametes – N
(One gene in egg)



Possible offspring – 2N

Male gametes - N
(One gene in sperm)

Genotypic ratio = 2 : 2
50% Bb : 50% bb

Phenotypic ratio = 2 : 2
50% black : 50% white

Write the ratios in the following orders:

Genotypic ratio

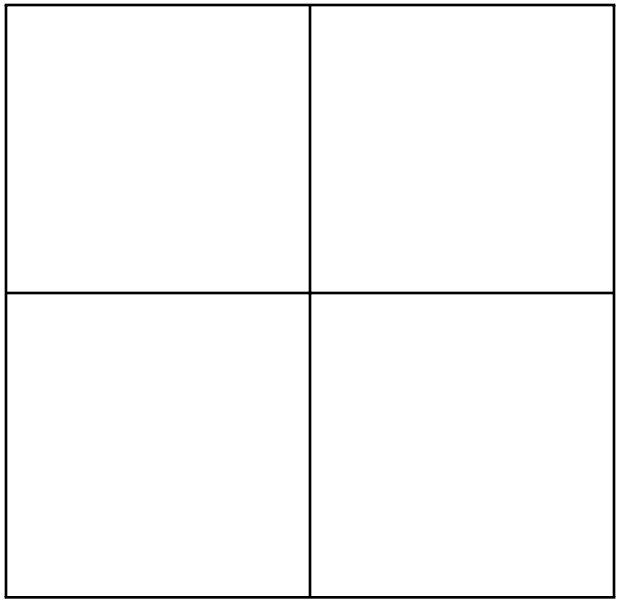
homozygous : heterozygous : homozygous
dominant recessive

Phenotypic ratio

dominant : recessive

Cross 2 hybrid mice and give the genotypic ratio and phenotypic ratio.

X



typic ratio = ____ : : ____

25% BB : 50% Bb : 25% bb

typic ratio = :

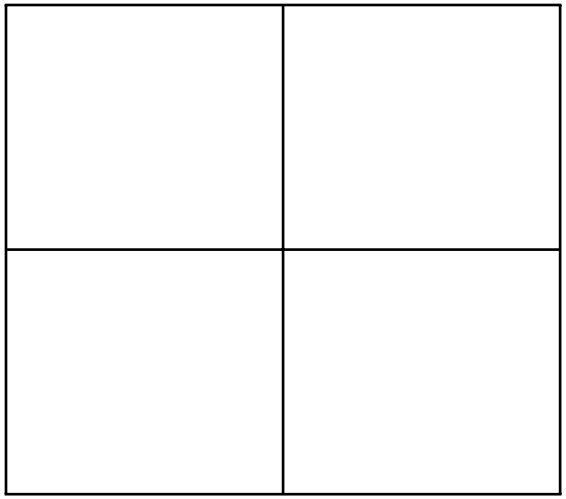
75% black : 25% white

Example: A man and woman, both with brown eyes (B) marry and have a blue eyed (b) child. What are the genotypes of the man, woman and child?

X

Man = ___

Woman =



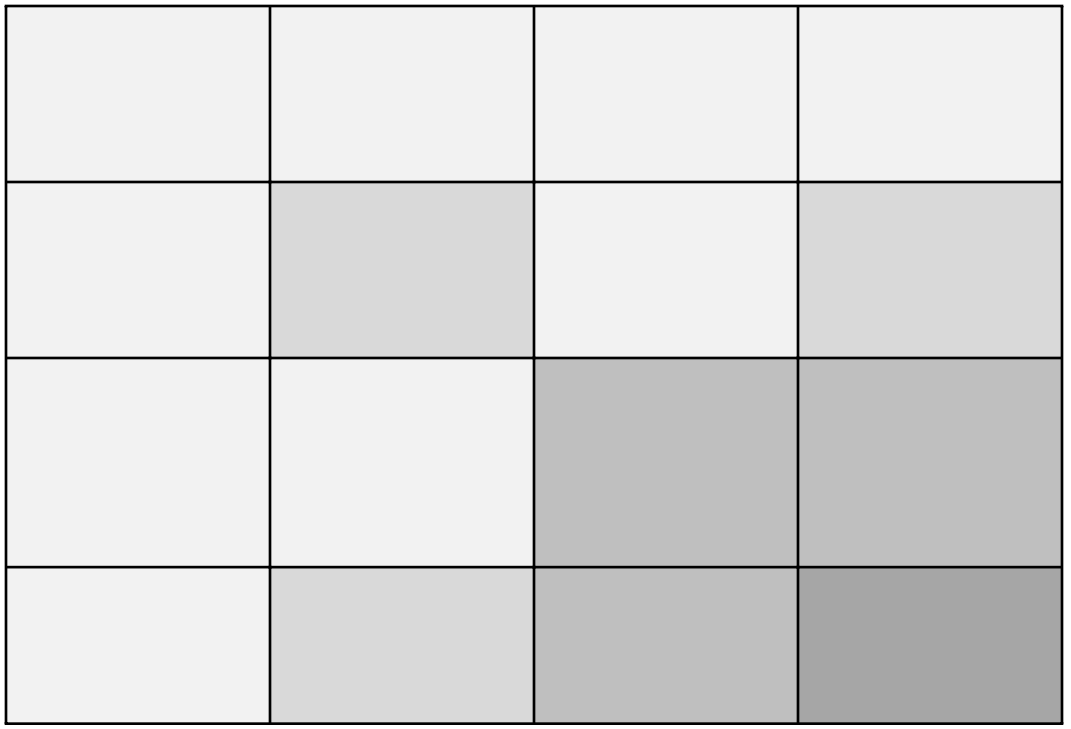
Crossing involving 2 traits – Dihybrid crosses

- Example: In rabbits black coat (B) is dominant over brown (b) and straight hair (H) is dominant to curly (h). Cross 2 hybrid rabbits and give the phenotypic ratio for the first generation of offspring.

Possible gametes:

X

Gametes



Phenotypes - 9:3:3:1

9 _____ _____

3 _____ _____

3 _____ _____

1 _____ _____

- Example: In rabbits black coat (B) is dominant over brown (b) and straight hair (H) is dominant to curly (h). Cross a rabbit that is homozygous dominant for both traits with a rabbit that is homozygous dominant for black coat and heterozygous for straight hair. Then give the phenotypic ratio for the first generation of offspring.

X

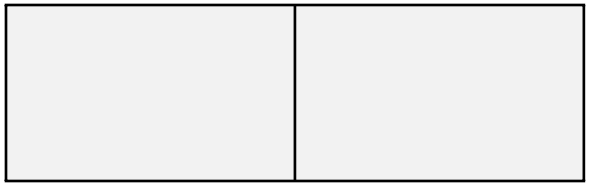
Possible gametes:

Gametes

Phenotypes:

100% _____ _____

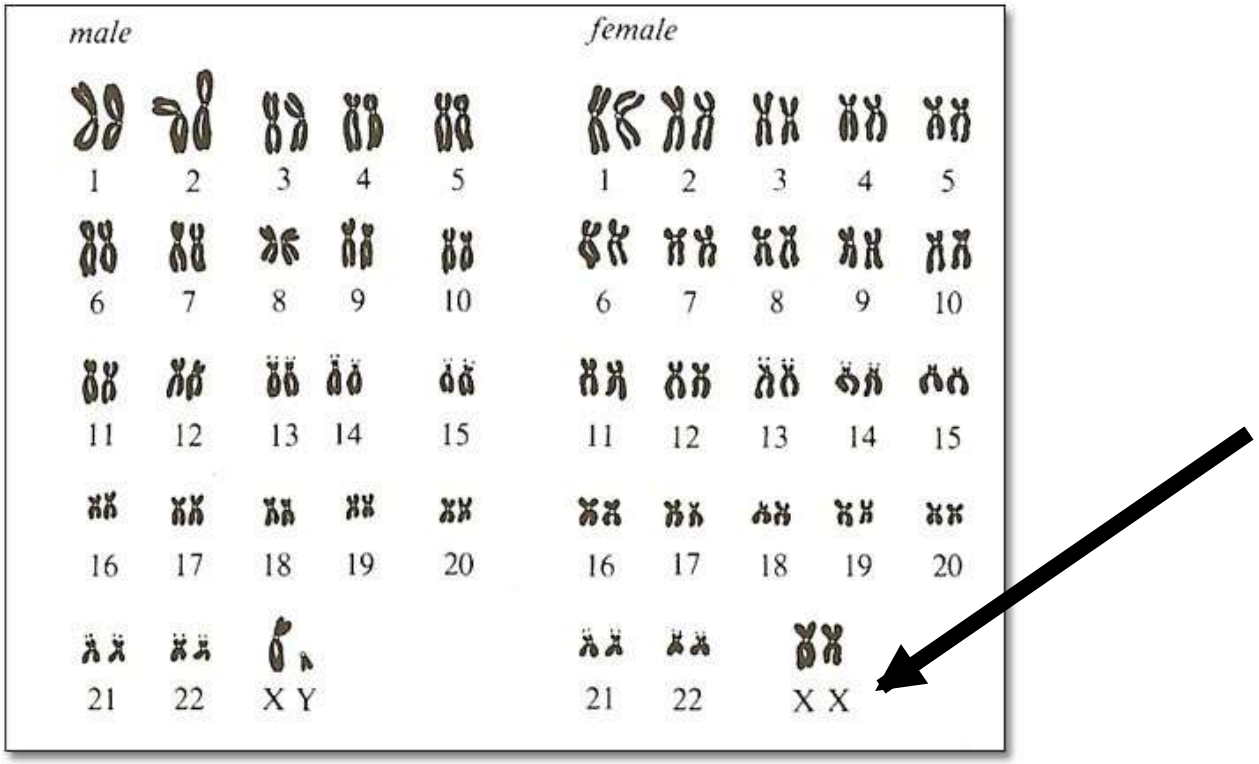
Gametes



(Hint: Only design Punnett squares to suit the number of possible gametes.)

Sex Determination

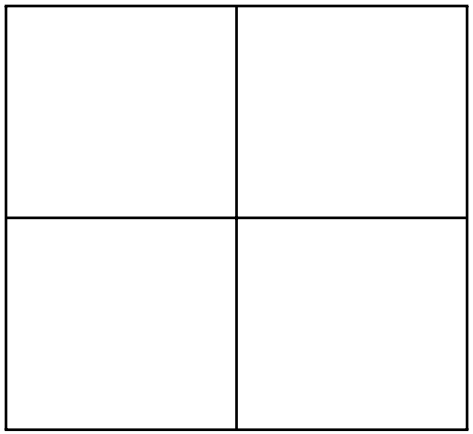
- People – **46** chromosomes or **23** pairs
- 22 pairs are **homologous** (look alike) – called **autosomes** – determine body traits
  1 pair is the **sex** chromosomes – determines sex (male or female)
- Females – sex chromosomes are **homologous** (look alike) – label **XX**
  Males – sex chromosomes are different – label **XY**



- What is the probability of a couple having a boy? Or a girl?

Chance of having female baby?

male baby?



Who determines the sex of the child? _____

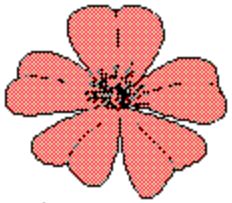
Incomplete dominance and Codominance

- When one allele is **NOT** completely **dominant** over another (they **blend**) – **incomplete dominance**

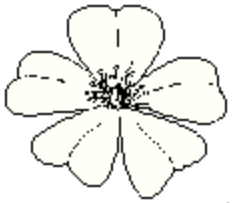
  Example: In carnations the color red (R) is incompletely dominant over white (W). The hybrid color is pink. Give the genotypic and phenotypic ratio from a cross between 2 pink flowers.

X

homozygous dominant



heterozygous



homozygous recessive

Genotypic = _____ : _____ : _____

Phenotypic = _____ : _____ : _____

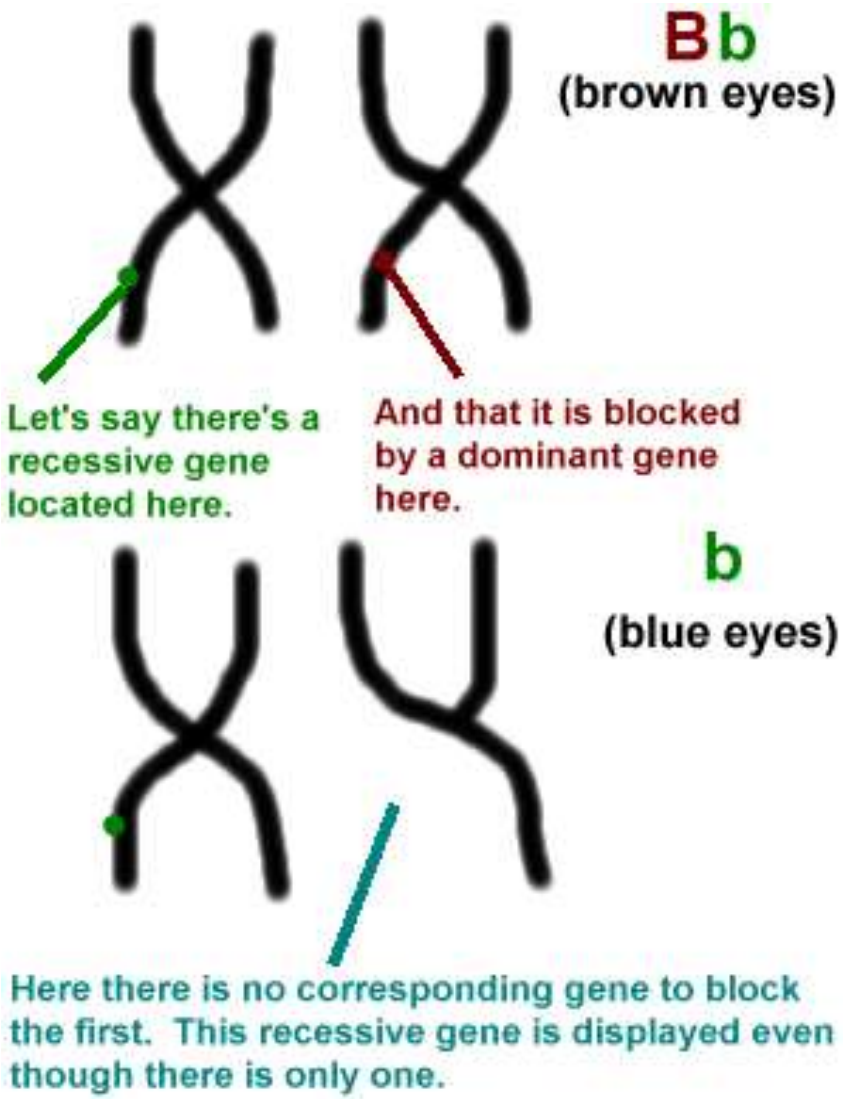
- When **both** alleles are **expressed** – **Codominance**

  Example: In certain chickens black feathers are codominant with white feathers.

  Heterozygous chickens have black and white speckled feathers.

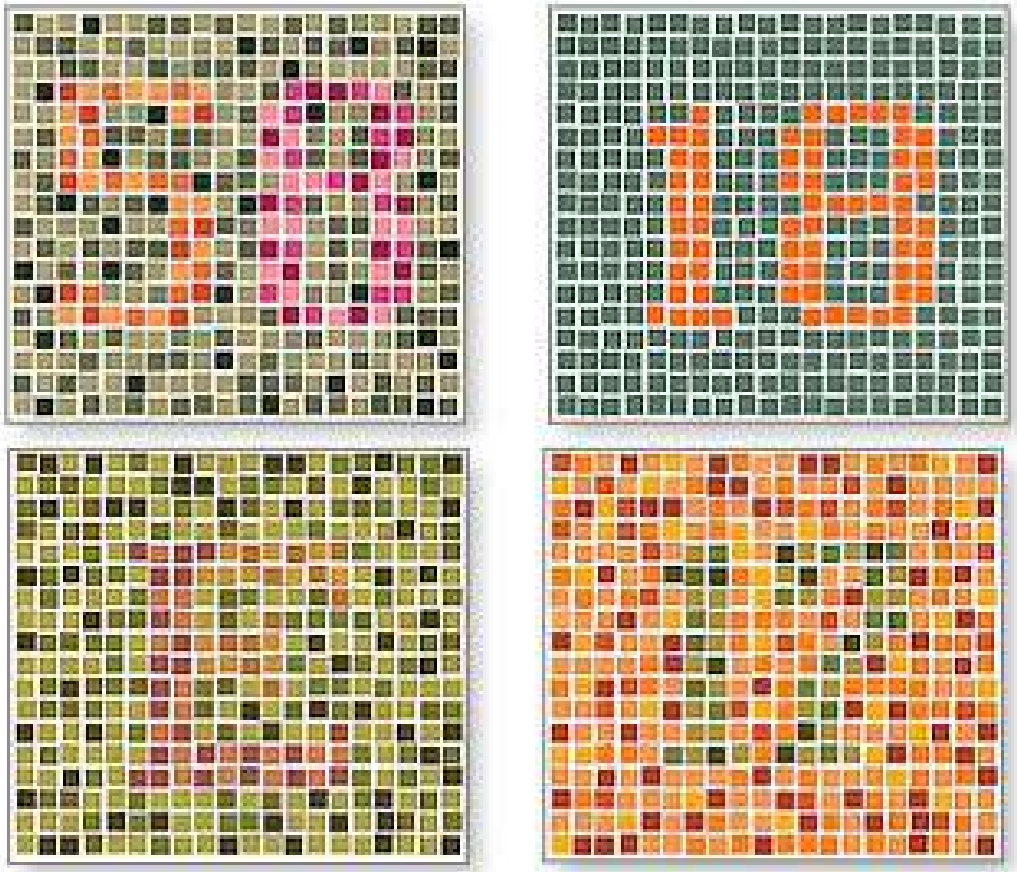
Sex – linked Traits

- **Genes** for these **traits** are located **only** on the **X** chromosome (NOT on the Y chromosome)
- X linked alleles **always** show up in **males** whether **dominant** or **recessive** because males have only **one** X chromosome



- Examples of **recessive** sex-linked disorders:

  1. colorblindness – inability to distinguish between certain colors

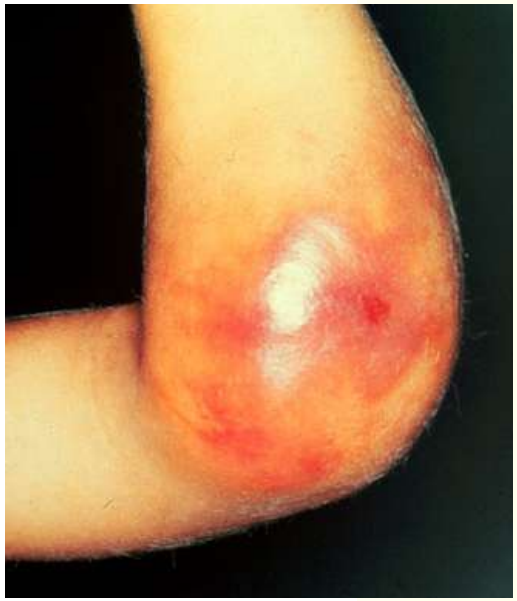


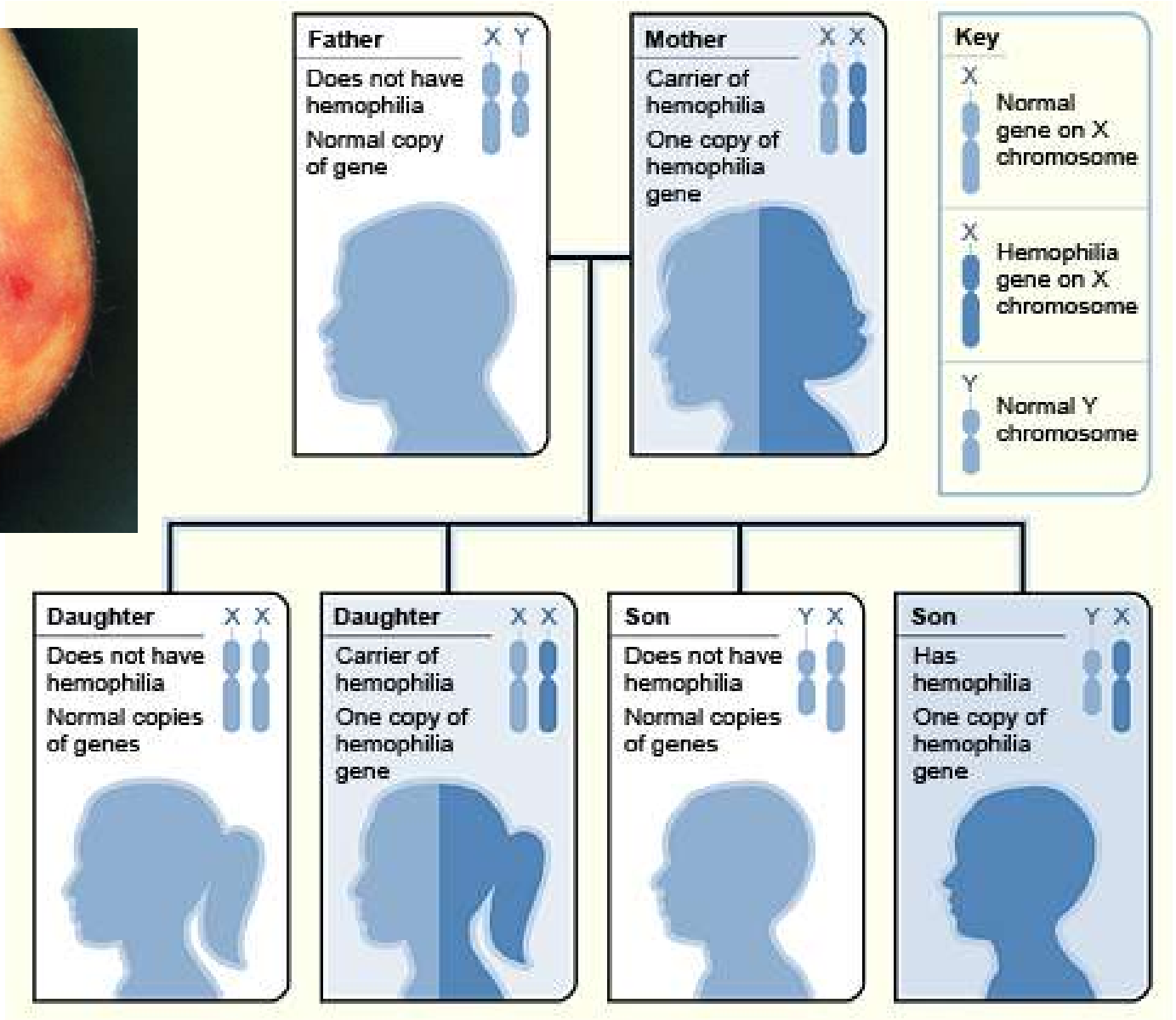
Various tests for color blindness

You should see **58** (upper left), **18** (upper right), **E** (lower left) and **17** (lower right).

Color blindness is the inability to distinguish the differences between certain colors. The most common type is red-green color blindness, where red and green are seen as the same color.

2. **hemophilia** – blood won't clot





- Example: A female that has normal vision but is a carrier for colorblindness marries a male with normal vision. Give the expected phenotypes of their children.

  N = normal vision

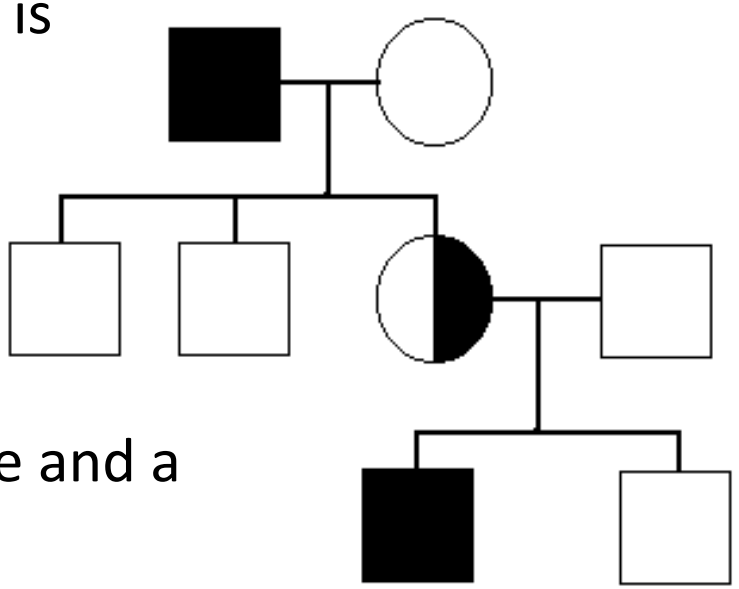
  n = colorblindness

X

Phenotype:

Pedigrees

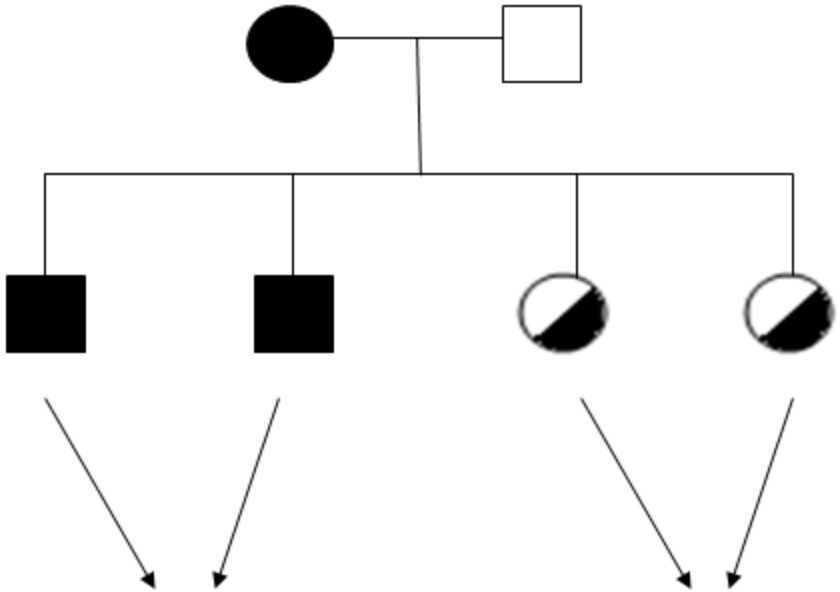
- **Graphic** representation of how a **trait** is passed from parents to **offspring**
- Tips for making a pedigree
  1. **Circles** are for females
  2. **Squares** are for males
  3. **Horizontal lines** connecting a male and a female represent a **marriage**
  4. **Vertical line** and **brackets** connect parent to offspring
  5. A **shaded** circle or square indicates a person **has** the trait
  6. A circle or square **NOT shaded** represents an individual who does NOT have the trait
  7. **Partial** shade indicates a **carrier** – someone who is **heterozygous** for the trait



- Example: Make a pedigree chart for the following couple. Dana is color blind; her husband Jeff is not.

  They have two boys and two girls.

  HINT: Colorblindness is a recessive sex-linked trait.



Has trait

Can pass trait to offspring

Multiple Alleles

- **3 or more alleles** of the **same** gene that code for a **single** trait
- In humans, **blood type** is determined by 3 alleles – **A**, **B**, and **O**

BUT each human can only **inherit 2** alleles

1. Dominant – A and B (codominance)

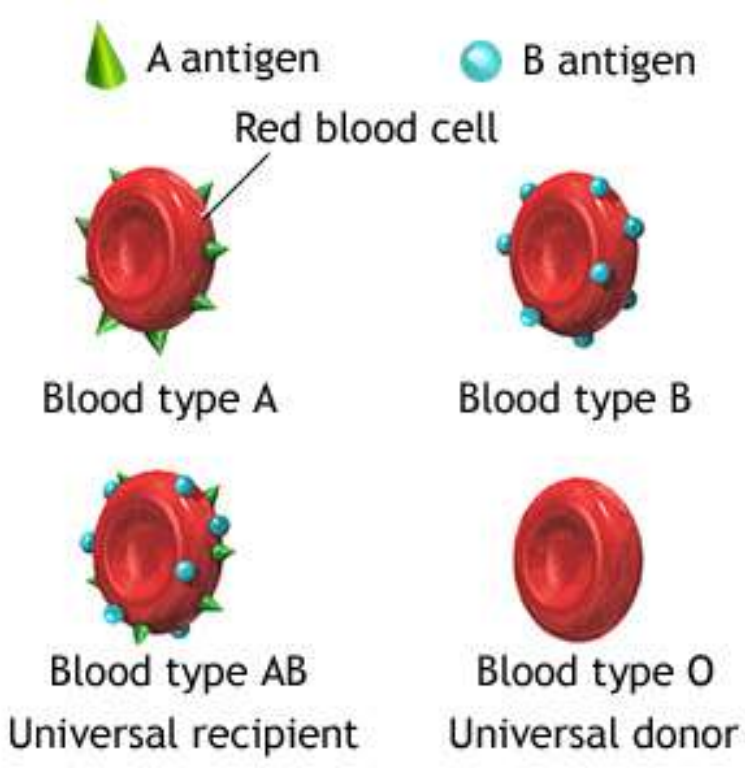
   Recessive – O

2. Blood type – A = AA or AO

   B = BB or BO

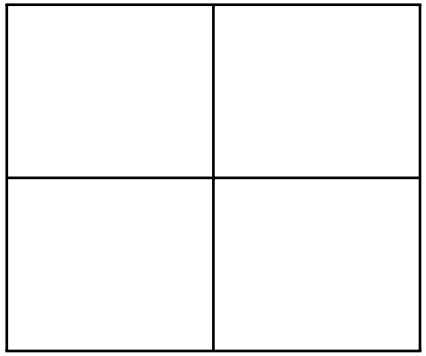
   AB = AB

   O = OO



Example: What would be the possible blood types of children born to a female with type AB blood and a male with type O blood?

X



Children would be type __ or __ only

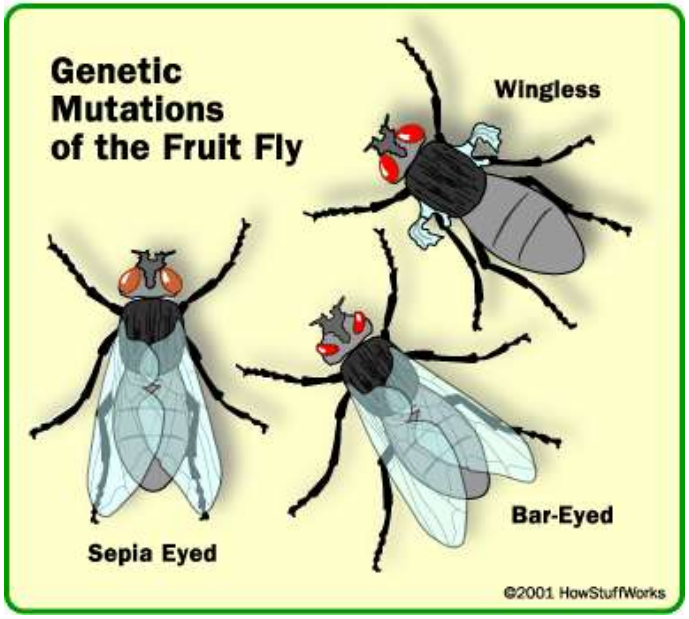
Mutations

- Mutation – sudden **genetic change** (change in **base** pair sequence of **DNA**)
- Can be :

  Harmful mutations – organism **less able** to survive: genetic disorders, cancer, death

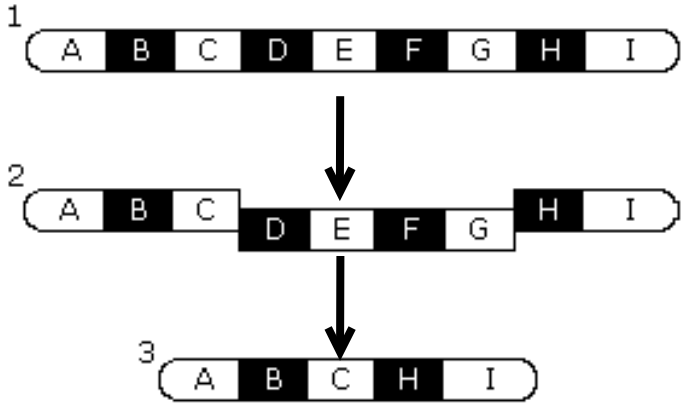
  Beneficial mutations – allows organism to **better survive**: provides **genetic variation**

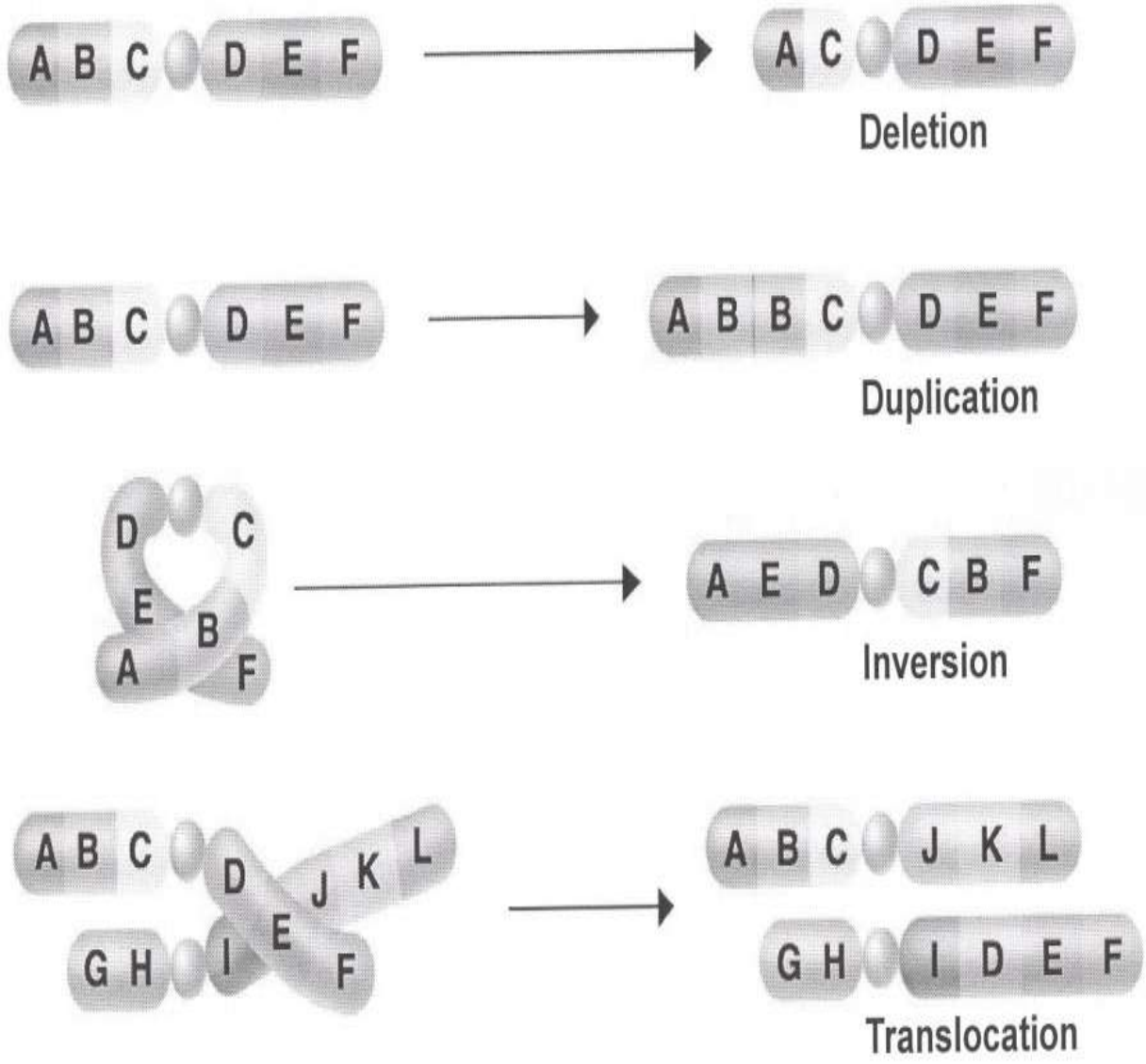
  Neutral mutations – **neither** harmful nor helpful to organism
- Mutations can occur in 2 ways: **chromosomal** mutation or **gene/point** mutation



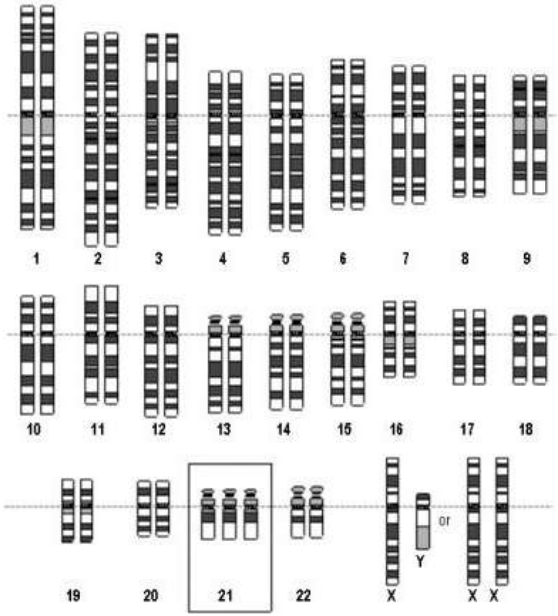
Chromosomal mutation:

- **less common** than a gene mutation
- more **drastic** – affects entire **chromosome**, so affects **many genes** rather than just one
- caused by failure of the **homologous** chromosomes to **separate** normally during **meiosis**
- **chromosome pairs** no longer look the same – too few or too many genes, different shape

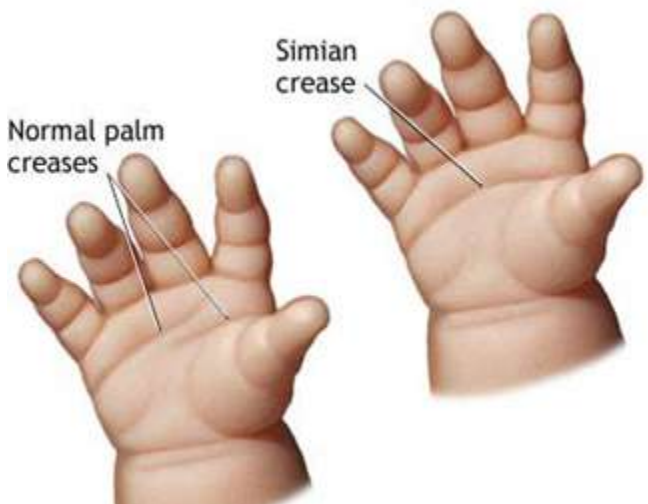



A B C D E F
A C D E F
Deletion
A B C D E F
A B B C D E F
Duplication
D C E B A F
A E D C B F
Inversion
A B C D J K L E G H I F
A B C J K L
G H I D E F
Translocation

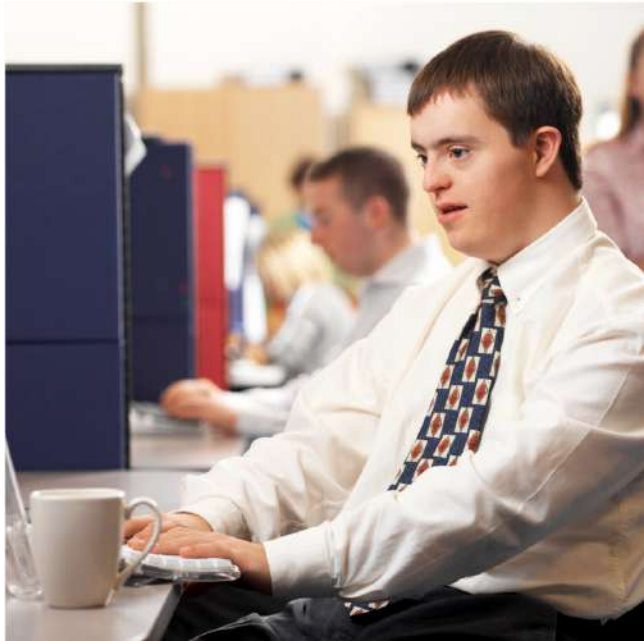
- Examples:
  Down's syndrome – (Trisomy 21) **47** chromosomes, extra chromosome at pair **#21**



flattened nose and face, upward slanting eyes,

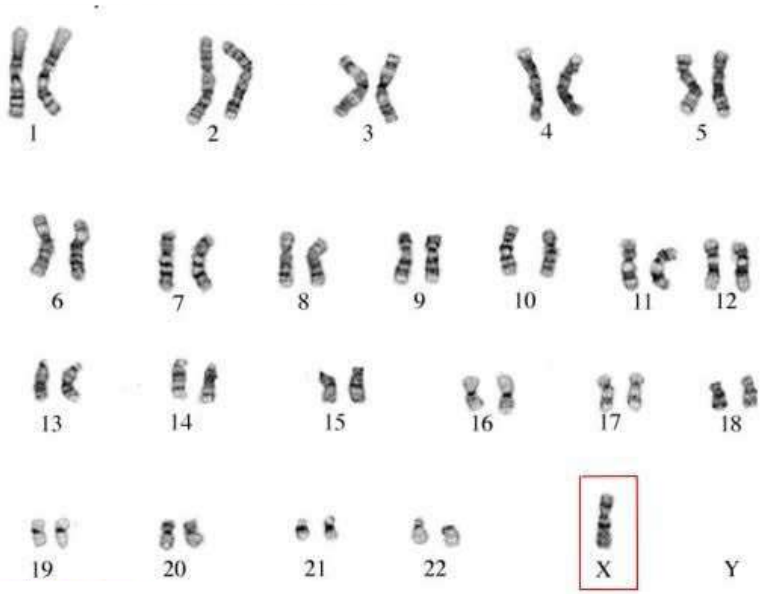


widely separated first and second toes and increased skin creases



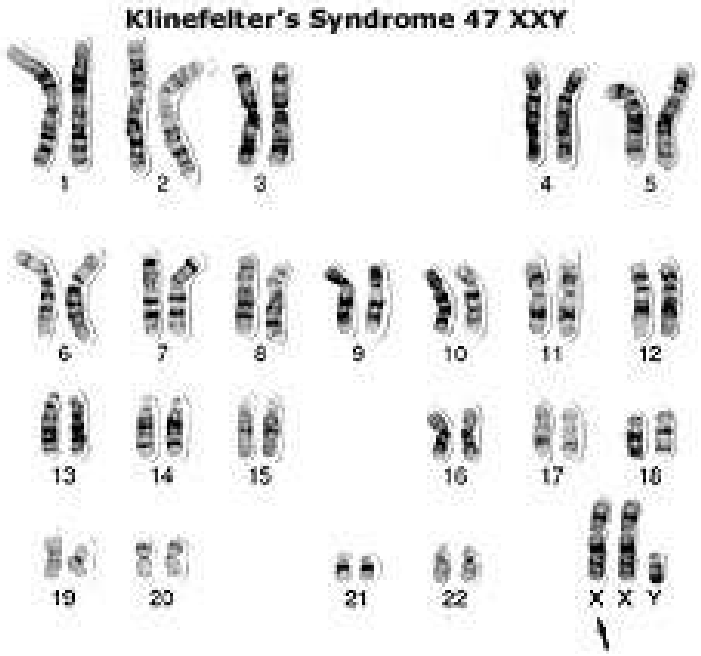
Turner's syndrome – only **45** chromosomes, missing a **sex** chromosome (X)

Girls affected – short, slow growth, heart problems

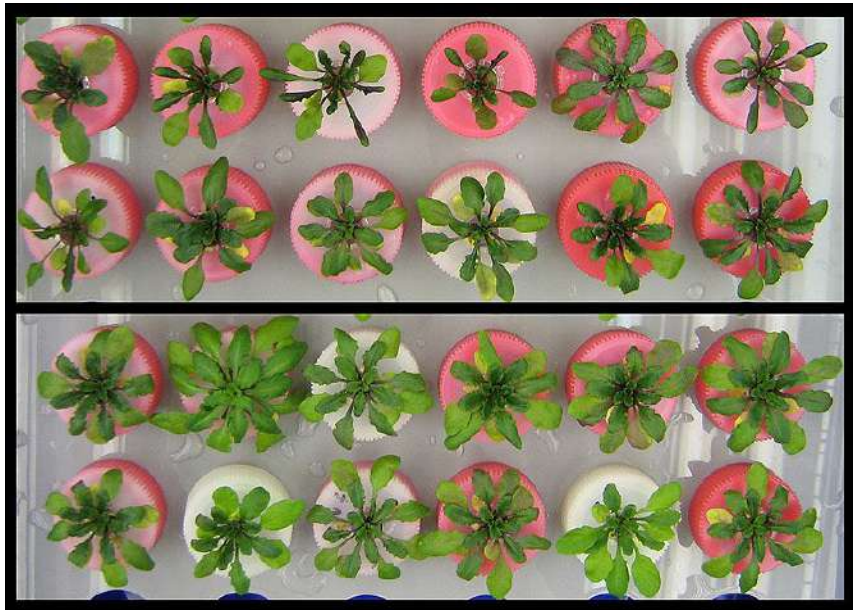


Klinefelter's syndrome – **47** chromosomes, **extra X** chromosomes (XXY)

Boys affected – low testosterone levels, underdeveloped muscles, sparse facial hair



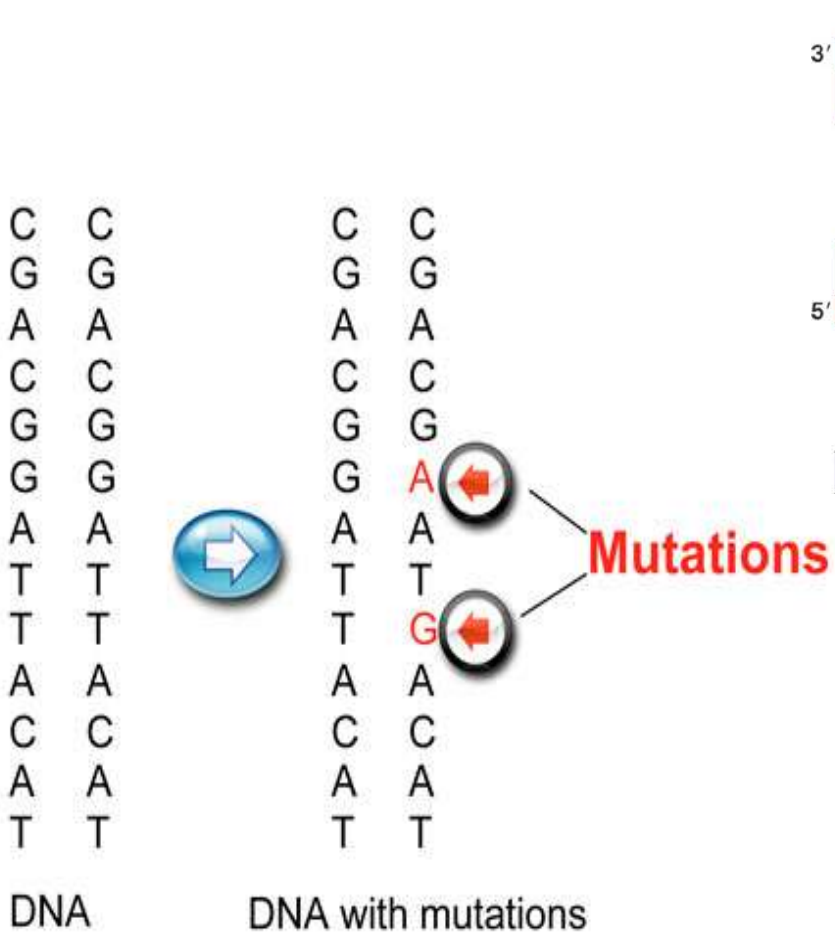
- Having an **extra set** of chromosomes is **fatal** in **animals**, but in **plants** it makes them **larger** and **hardier**.

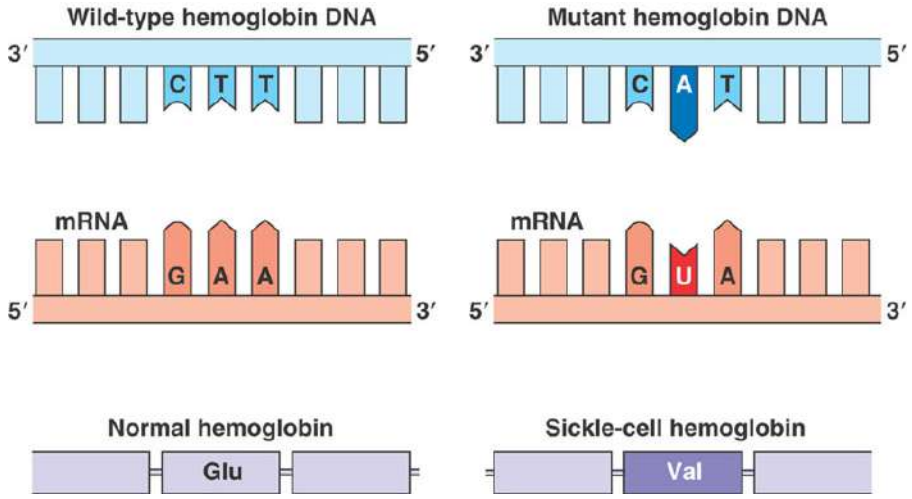


Hardier

Gene or Point Mutation

- **most common** and **least drastic**
- only **one gene** is altered

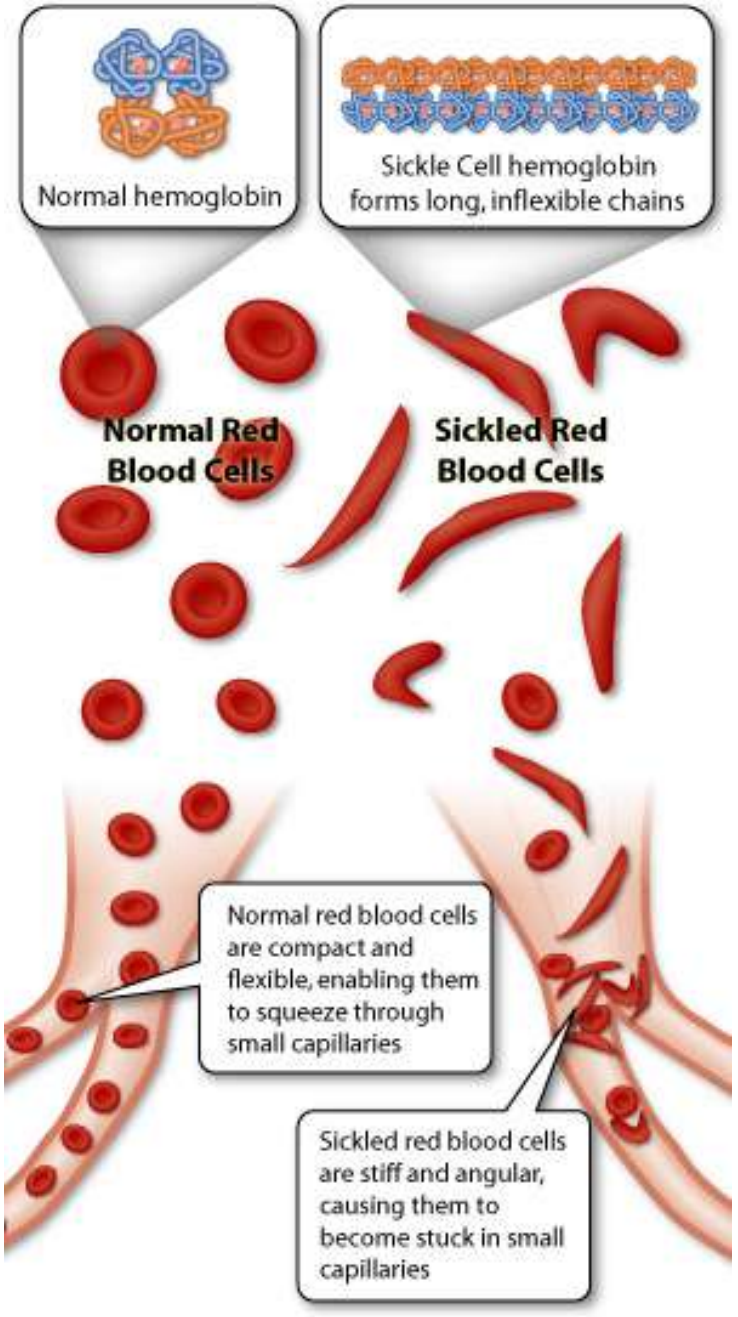




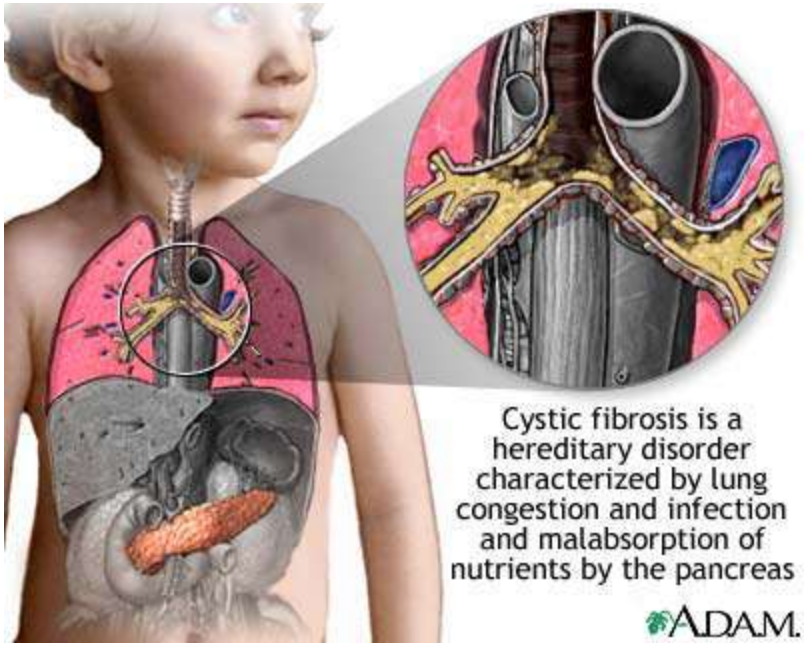
- Examples:

Recessive gene mutations:

Sickle cell anemia – **red blood cells** are sickle shaped instead of round and cannot carry enough **oxygen** to the body tissues – heterozygous condition protects people from **malaria**

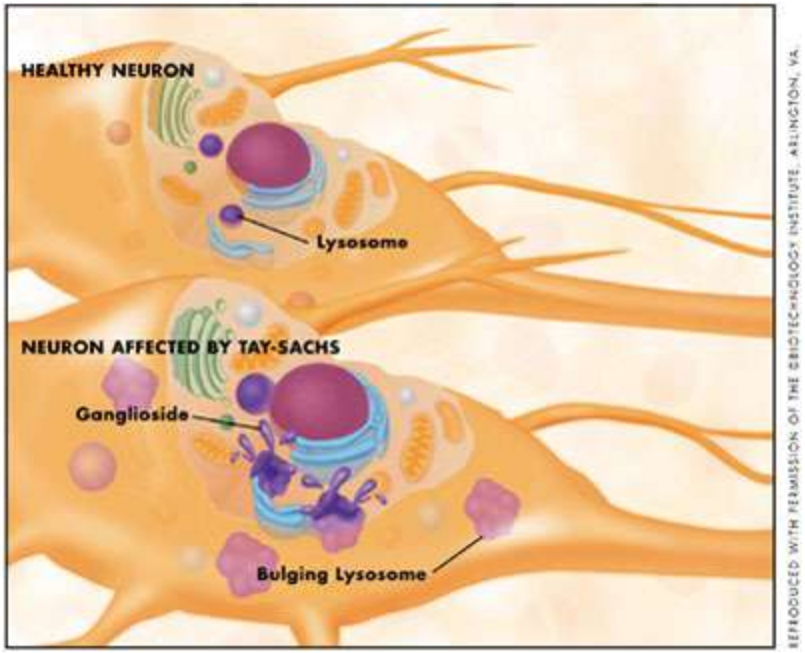


Cystic fibrosis – **mucous** builds up in the **lungs**



Tay-Sachs Disease – deterioration of the **nervous system** – early death

Mutated genes produce enzymes that are less effective than normal at breaking down fatty cell products known as gangliosides. As a result, gangliosides build up in the lysosomes and overload cells. Their buildup ultimately causes damage to nerve cells.



Phenylketonuria (PKU) – an **amino acid** common in **milk** cannot be broken down and as it builds up it causes **mental retardation** – newborns are tested for this

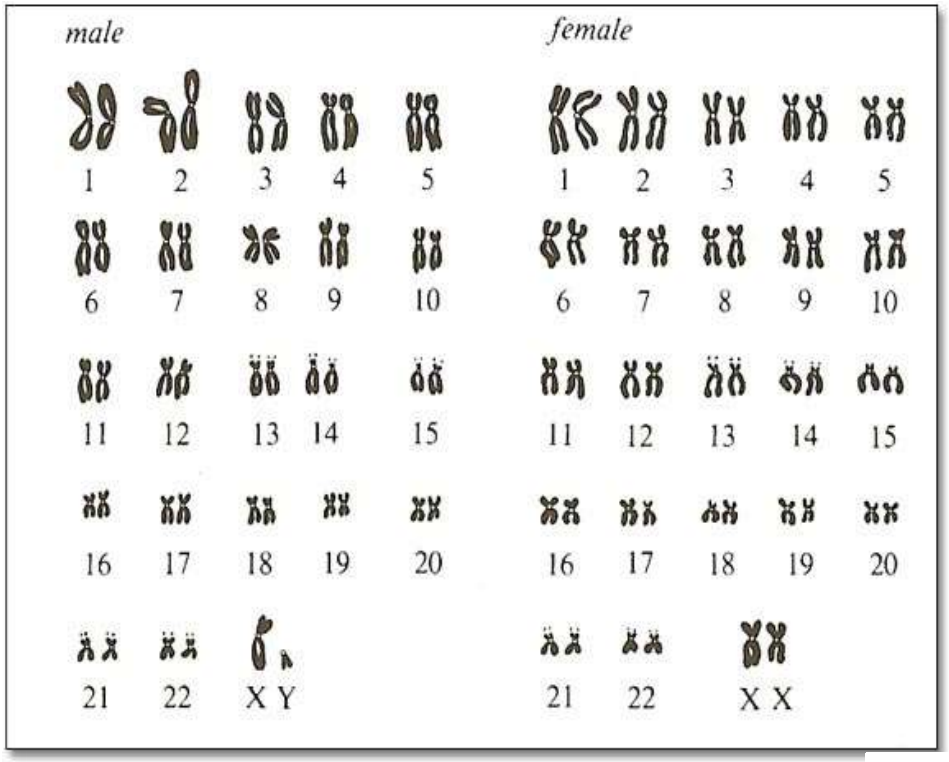
Dominant gene mutations:

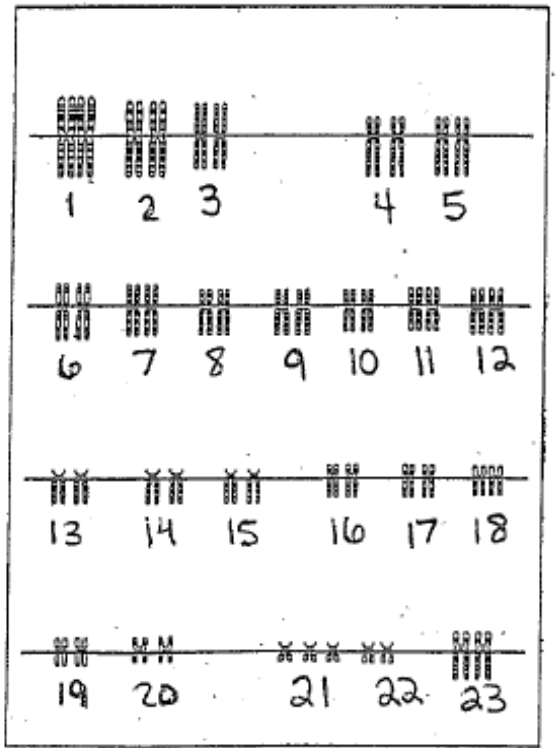
Huntington's disease – gradual **deterioration** of **brain tissue**, shows up in **middle age** and is **fatal**

Dwarfism – variety of **skeletal** abnormalities

Detecting Genetic Disorders

- picture of an individual's chromosomes – **karyotype**
- amniotic fluid surrounding the embryo is removed for analysis – **amniocentesis**





Female with **Down's** syndrome