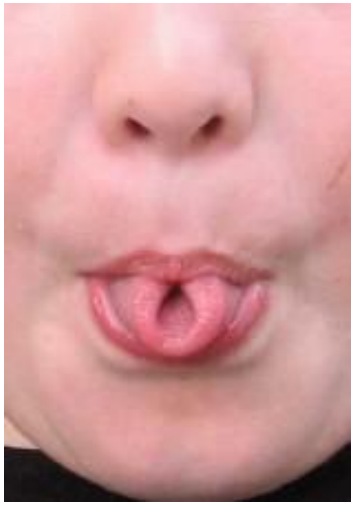
Genetics Notes

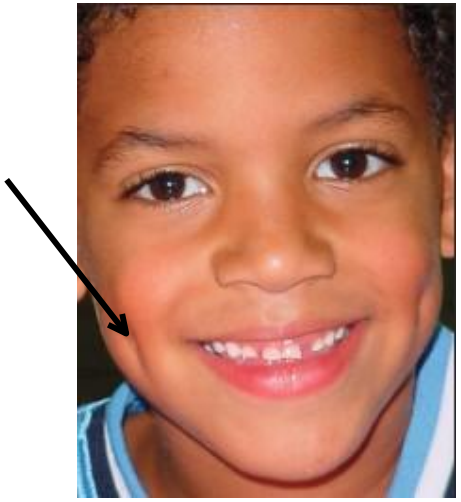
Who is Gregor Mendel?"Father of Genetics"

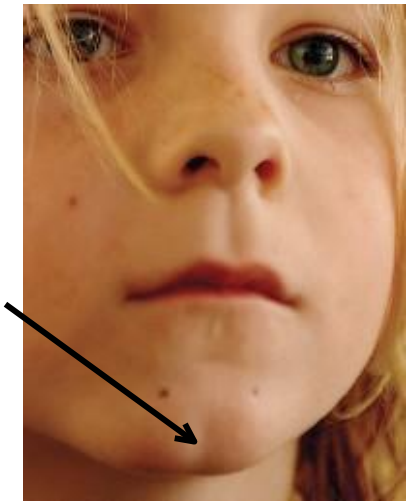
Principle of Independent Assortment – Inheritance of one trait has no effect on the inheritance of another trait

What is Genetics?

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

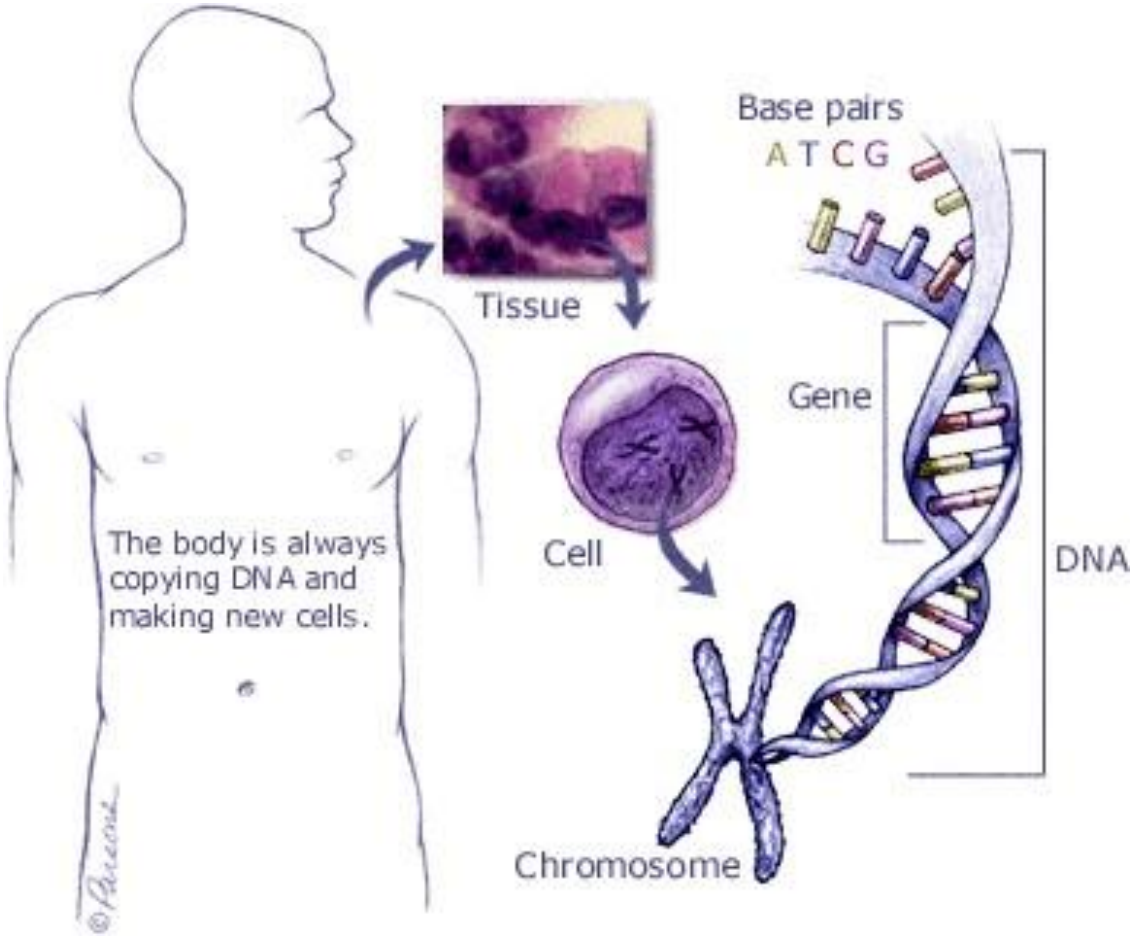


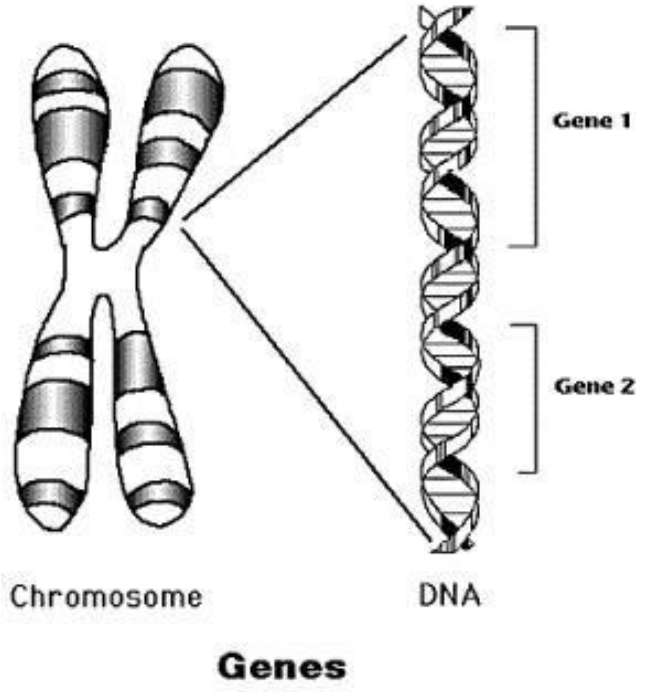




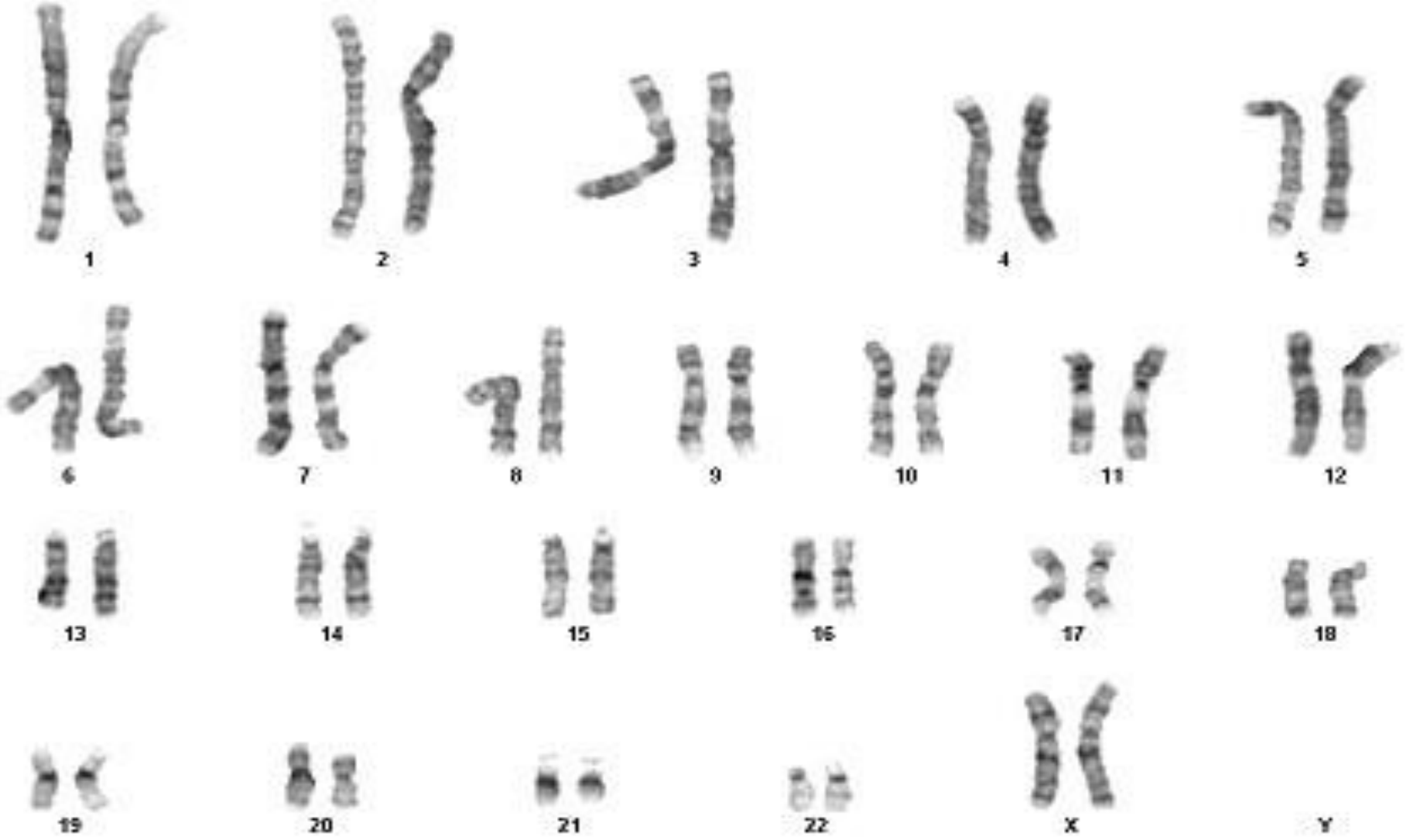
What are traits determined by?

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



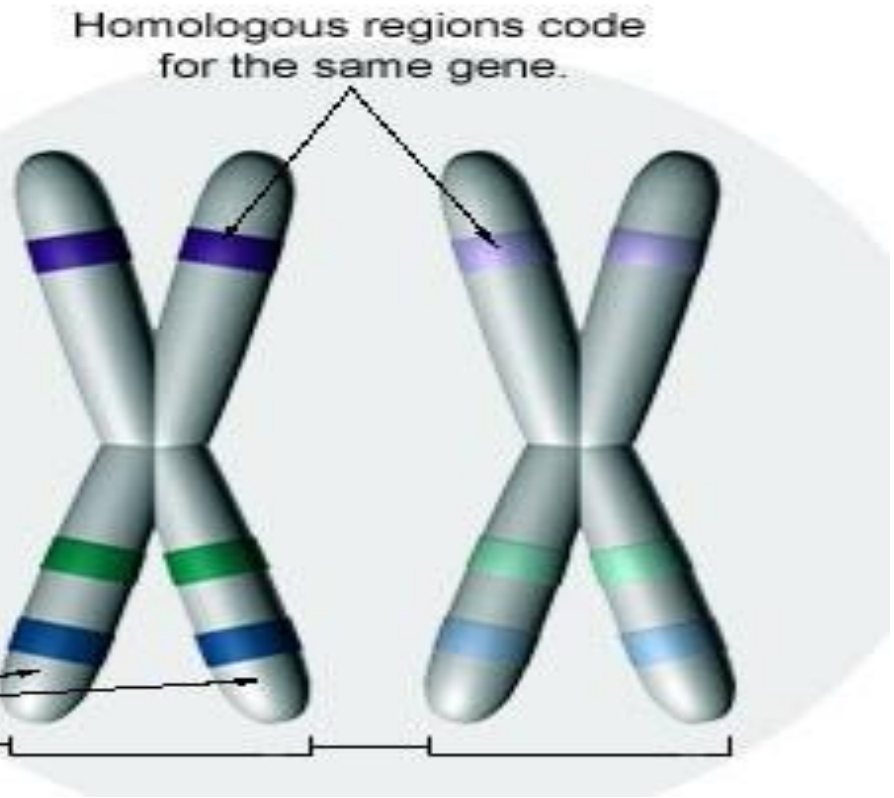


All of our chromosomes



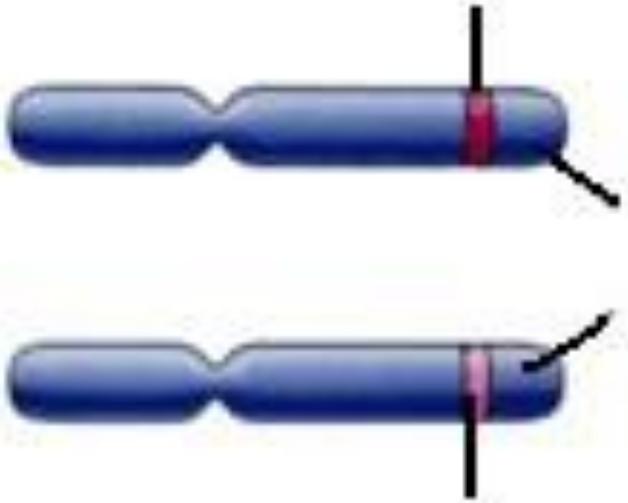
- Chromosomes come in homologous pairs, thus genes come in pairs.
  Homologous pairs – matching genes (not the same sequence of nucleotides) – one from female parent and one from male parent
- Example: Humans have 46 chromosomes or 23 pairs.
  One set from dad – 23 in sperm
  One set from mom – 23 in egg

Homologous chromosomes contain DNA that codes for the same genes. In this example, both chromosomes have all the same genes in the same locations (represented with colored strips), but different 'versions' of those genes (represented by the different shades of each color).



- One pair of Homologous Chromosomes: label on your notes

Gene for eye color (blue eyes)



Homologous pair of chromosomes

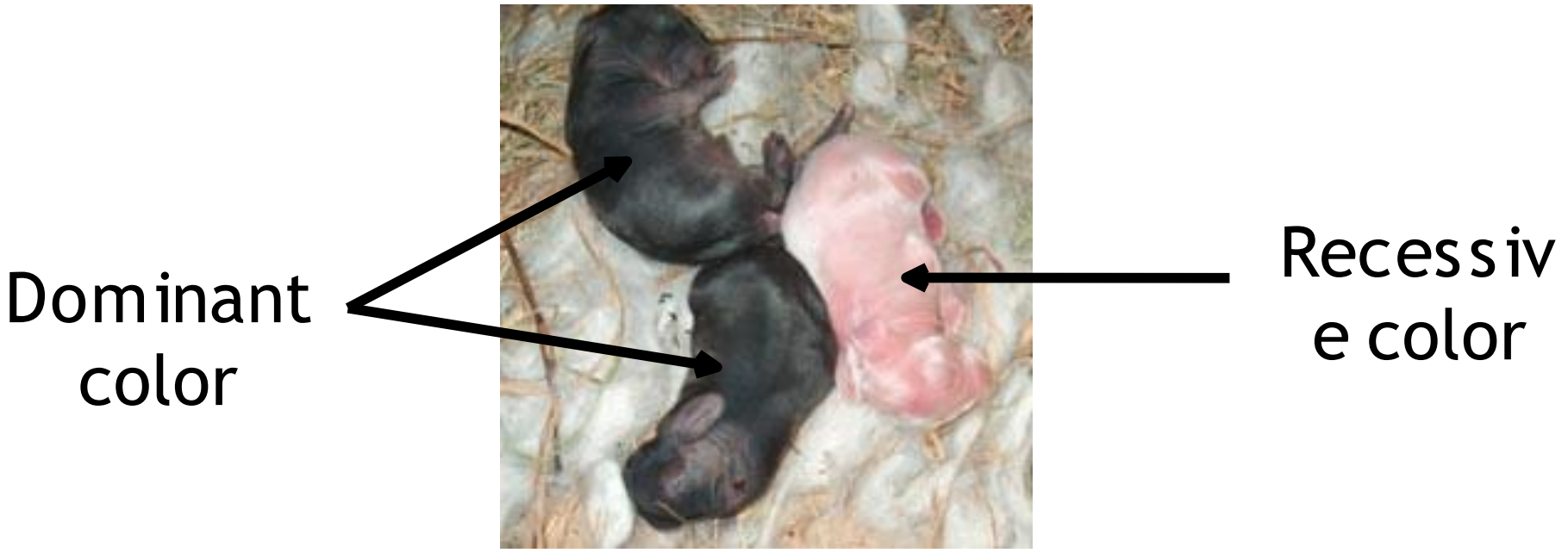
Gene for eye color (brown eyes)

Alleles – different genes (possibilities) for the same trait –

ex: blue eyes or brown eyes

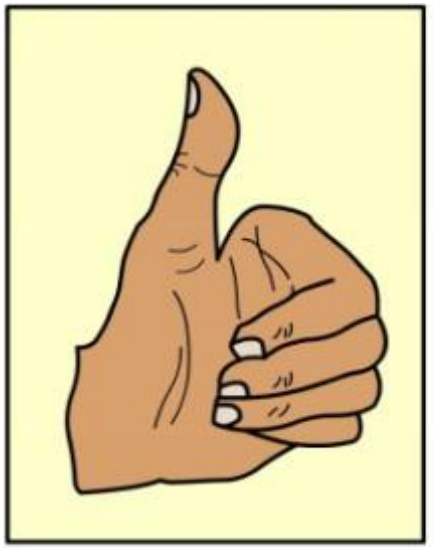
Dominant and Recessive Genes

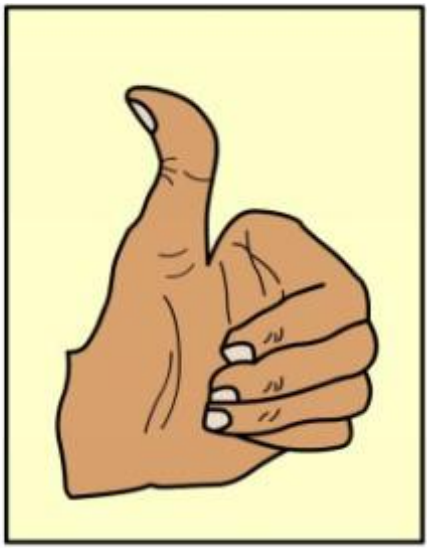
- Gene that prevents the other gene from "showing" – dominant
- Gene that does NOT "show" even though it is present – recessive
- Symbol – Dominant gene – upper case letter – T
  Recessive gene – lower case letter – t



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

- Both genes of a pair are the same – homozygous or purebred
  - TT – homozygous dominant
  - tt – homozygous recessive

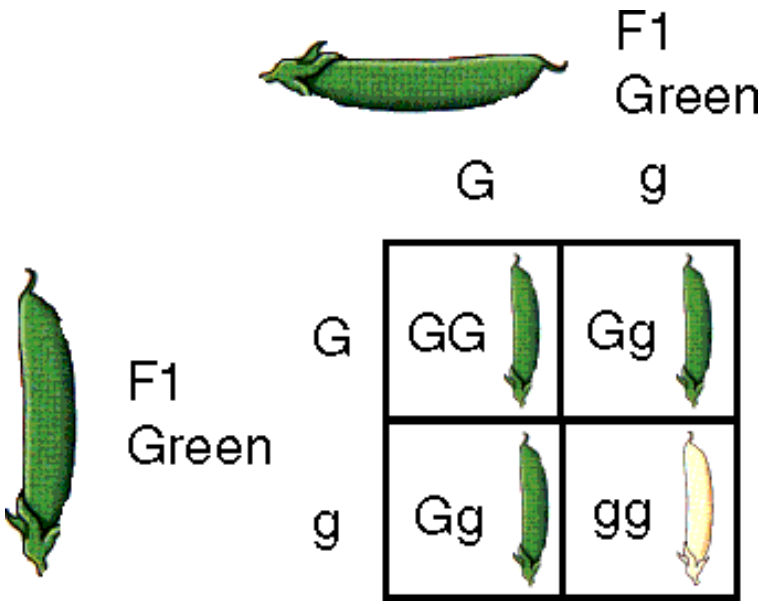
- One dominant and one recessive gene – heterozygous or hybrid
  - Tt – heterozygous

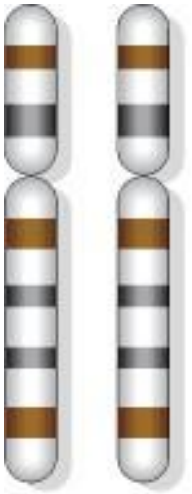
BB – Black
Bb – Black w/ white gene

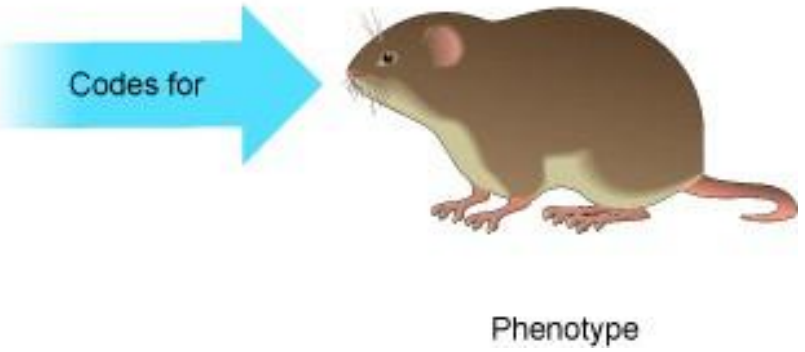
bb – White

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







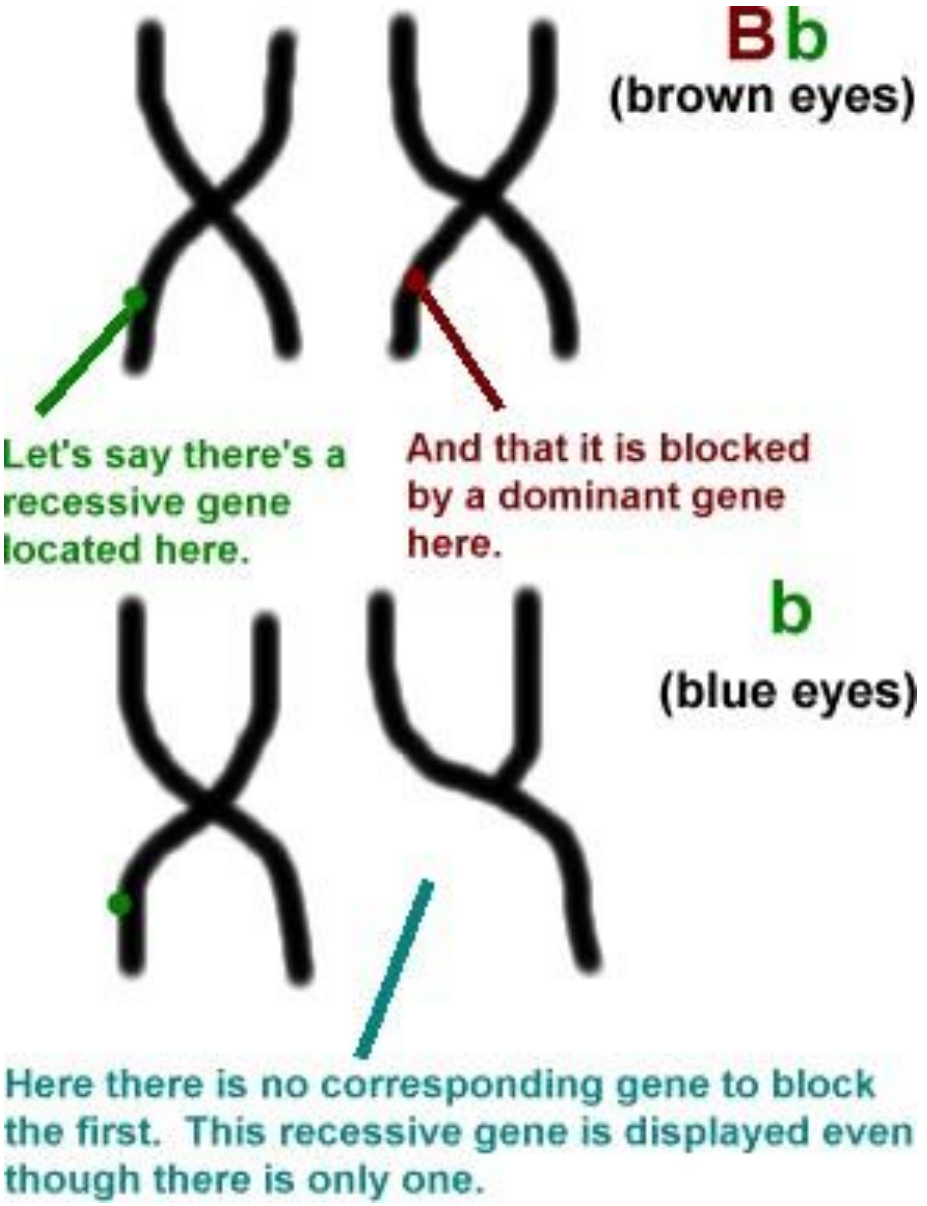
Diploid and Haploid

- Diploid- 2 copies of the chromosomes – 46

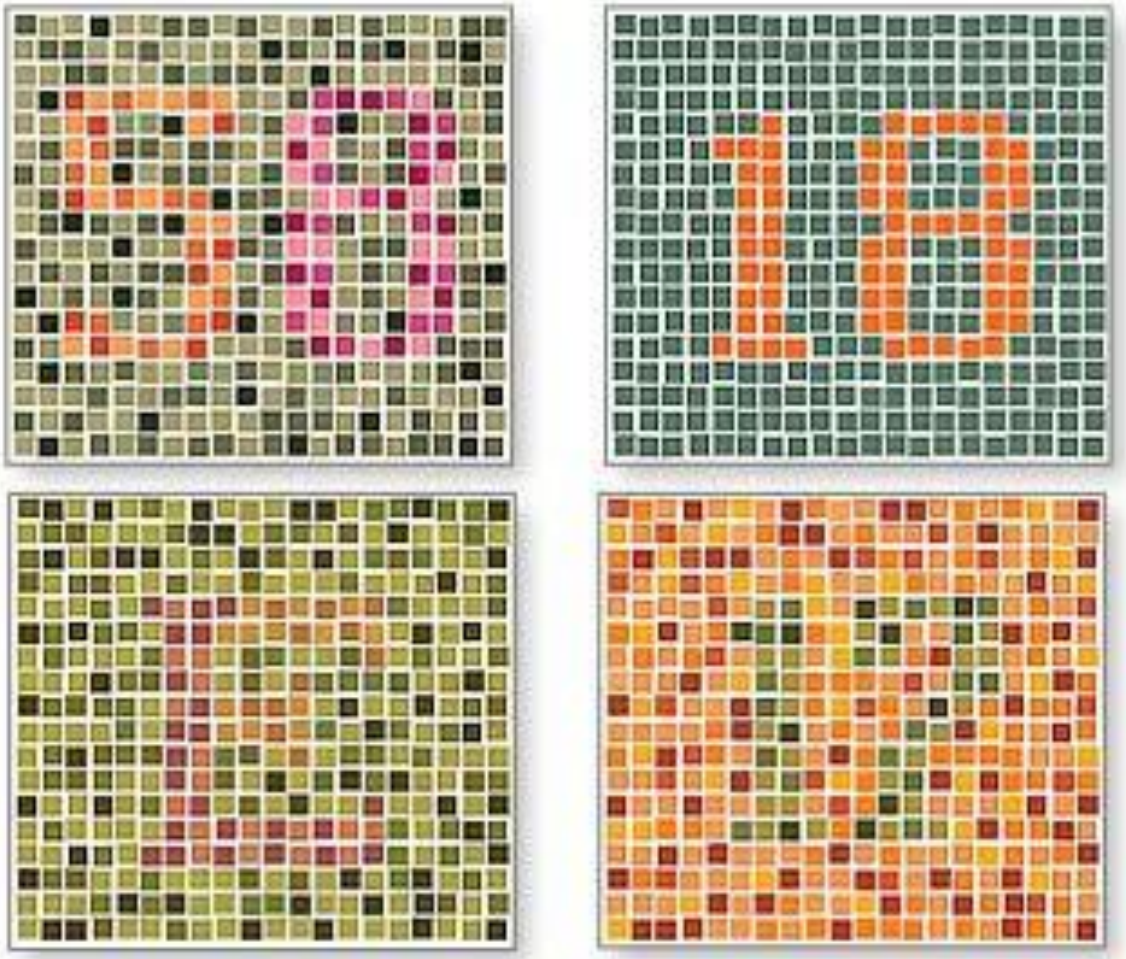
    All body cells are diploid except sperm and eggs

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

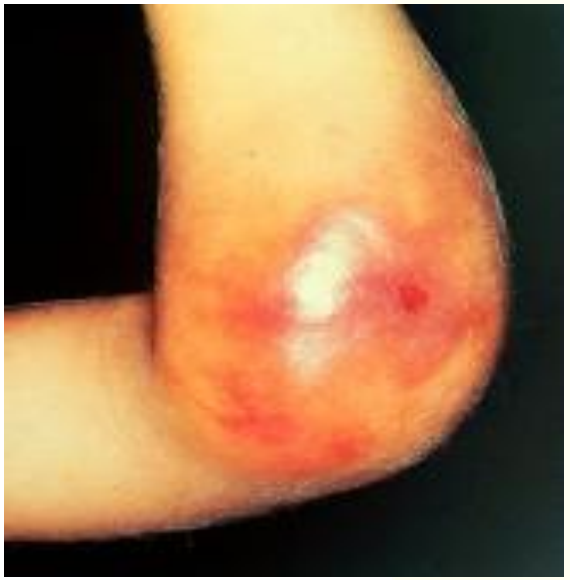


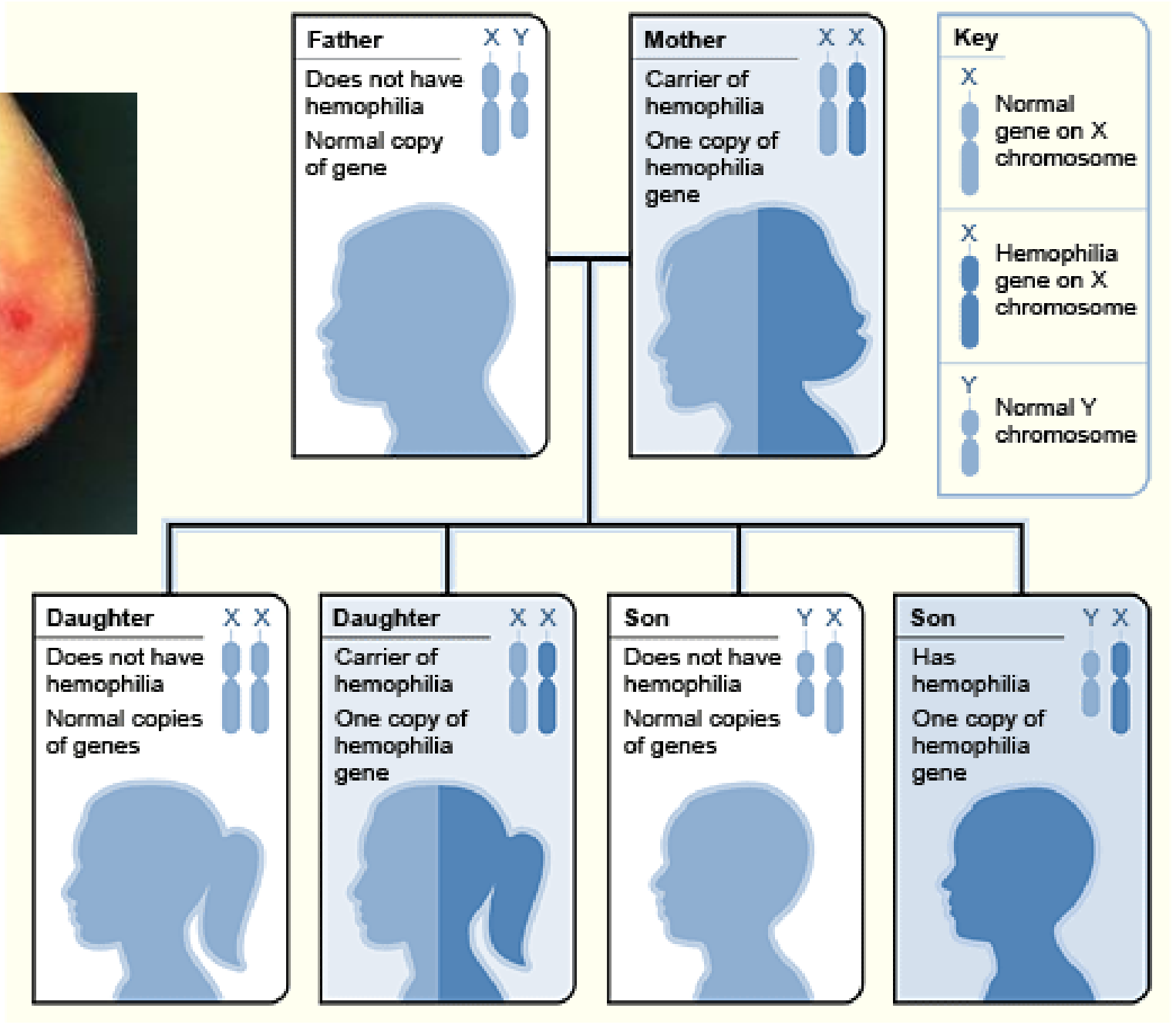
You should see 58 (upper left), 18 (upper right), E (lower left) and 17 (lower right).

Various tests for color blindness

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 <u>carrier for colorblindness</u> marries a male with <u>normal vision</u>. Give the expected phenotypes of their children.

N = normal vision

n = colorblindness

Phenotype <u>females</u>

1 normal

1 colorblind male

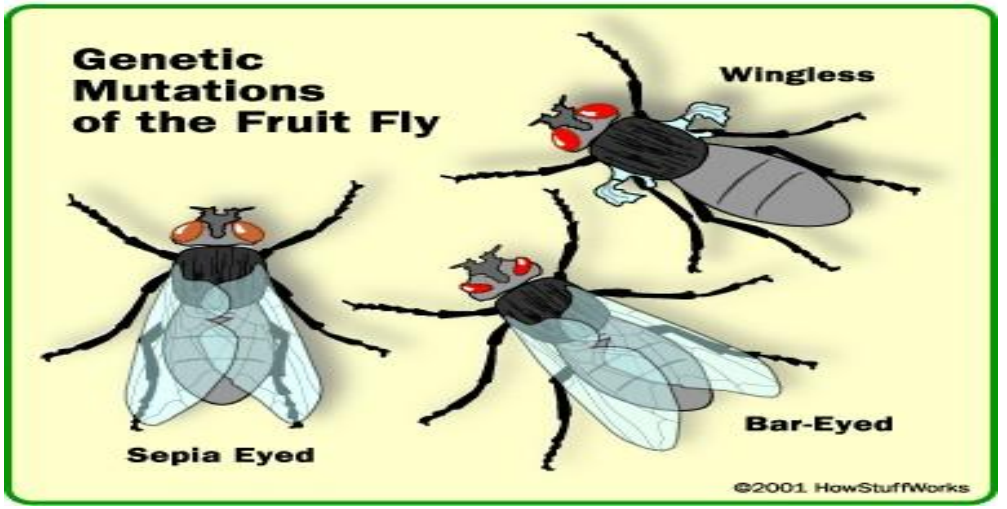
Mutations

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

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

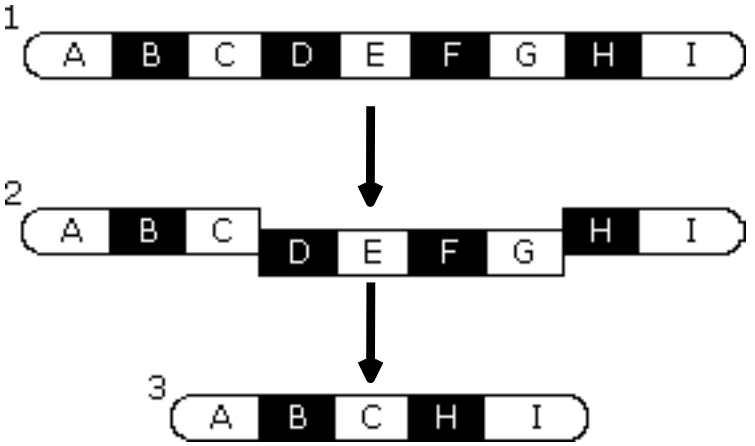
Beneficial mutations – allows organism to better

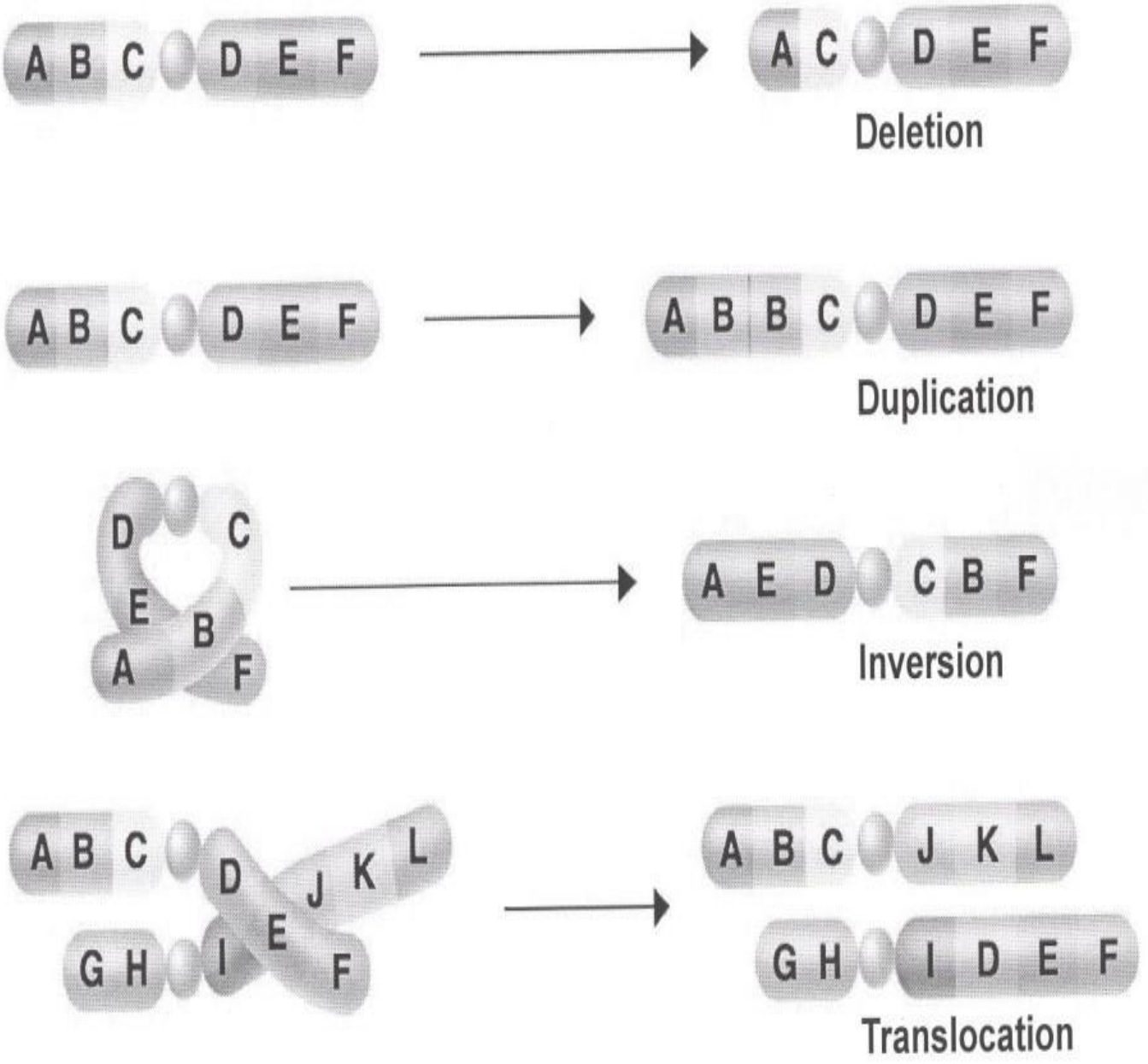
Mutations can occur in 2 ways: chromosomal mutation or gene/point mutation



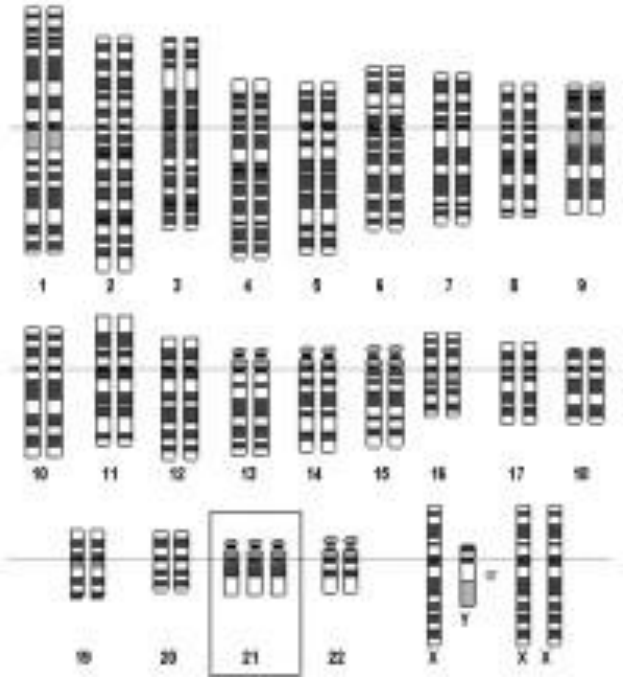
Chromosomal mutation:

- 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

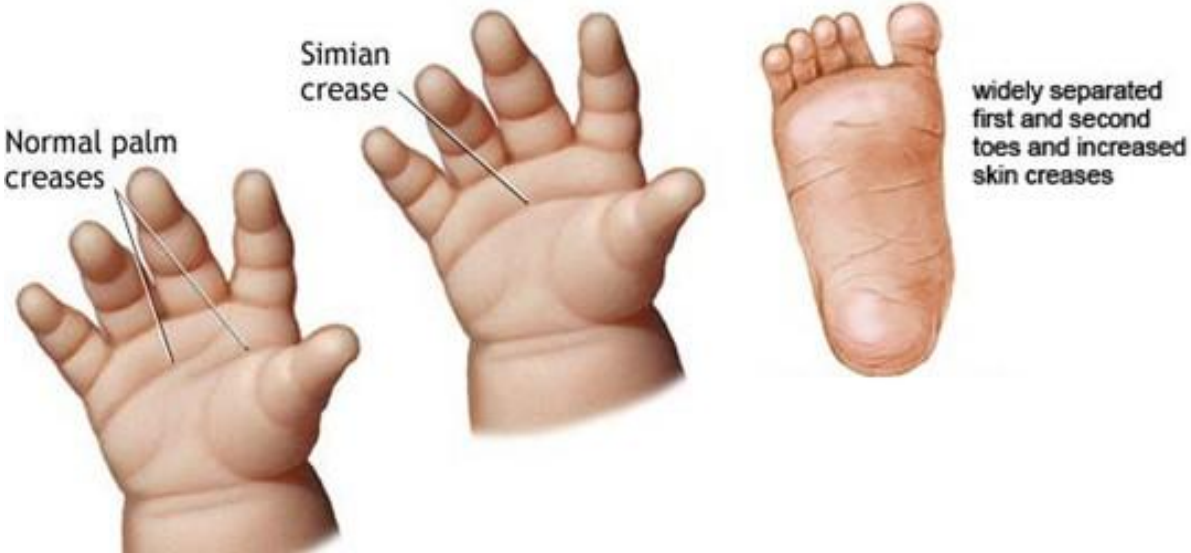



A B C D E F
A C D E F
Deletion
A B C D E F
A B B C D E F
Duplication
D C E B A F
A E D C B F
Inversion
A B C D J K L G H I E F
A B C J K L
G H I D E F
Translocation

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



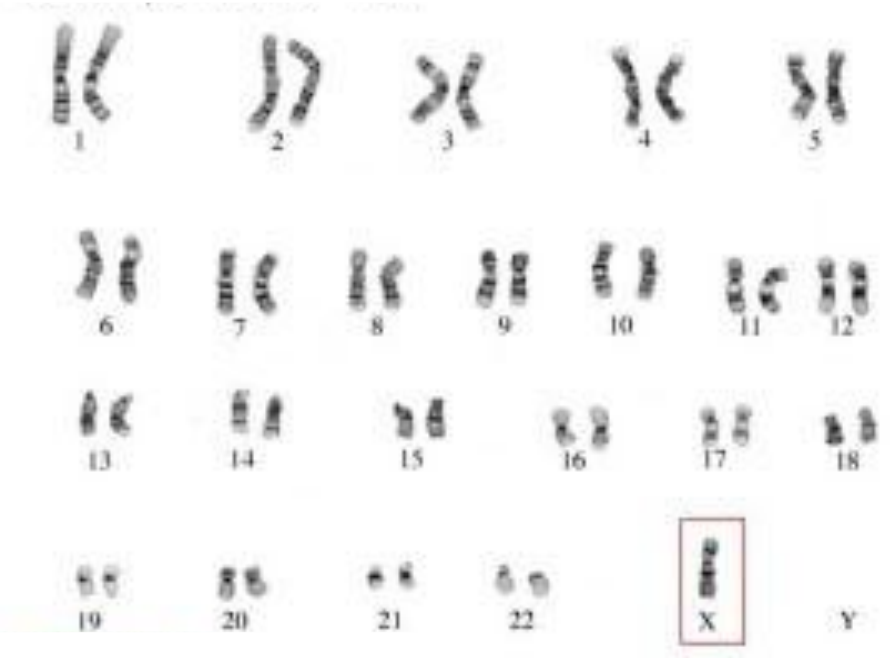
flattened nose and face, upward slanting eyes,



widely separated first and second toes and increased skin creases

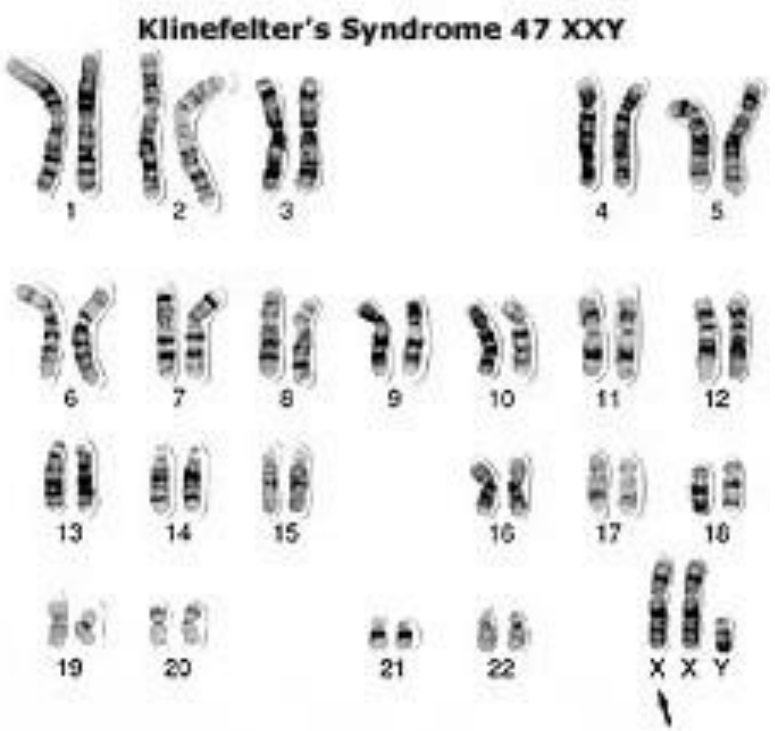
Turner's syndrome – only 45 chromosomes, missing a sex chromosome (X)

Girls affected – short, slow growth, heart problems



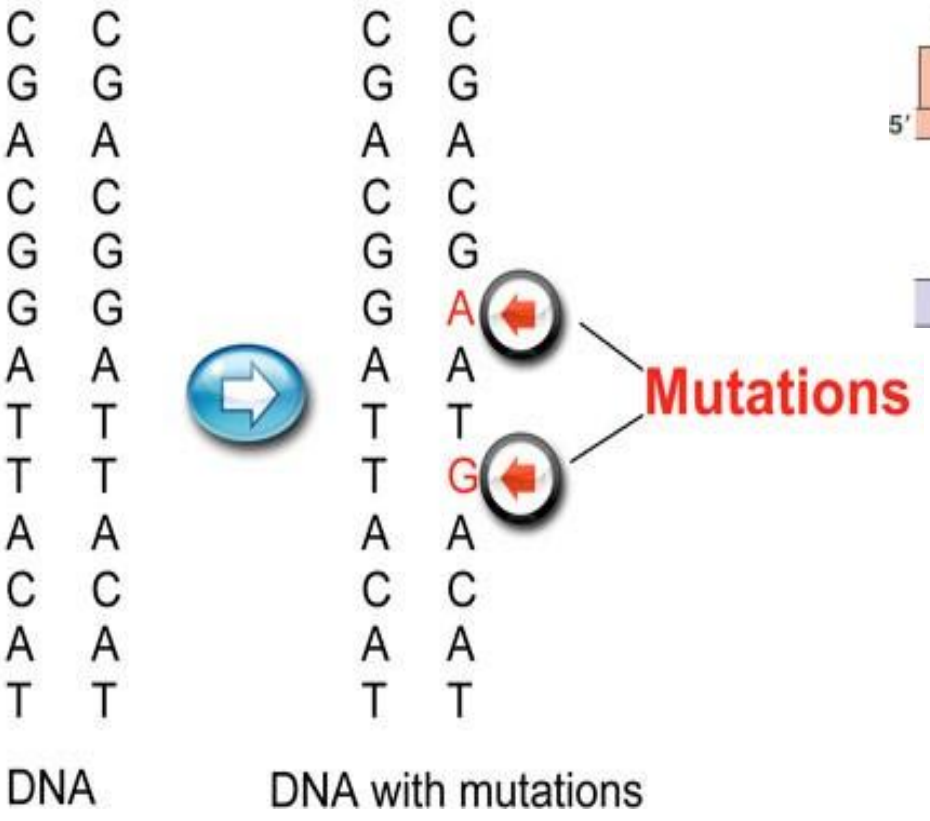
Klinefelter's syndrome – 47 chromosomes, extra X chromosomes (XXY)

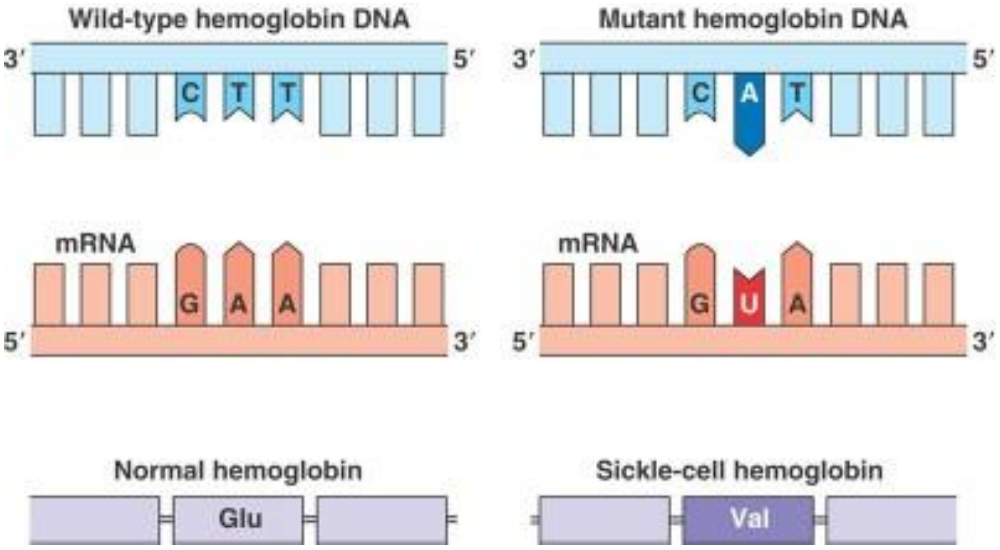
Boys affected – low testosterone levels, underdeveloped muscles, sparse facial hair



Gene or Point Mutation

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

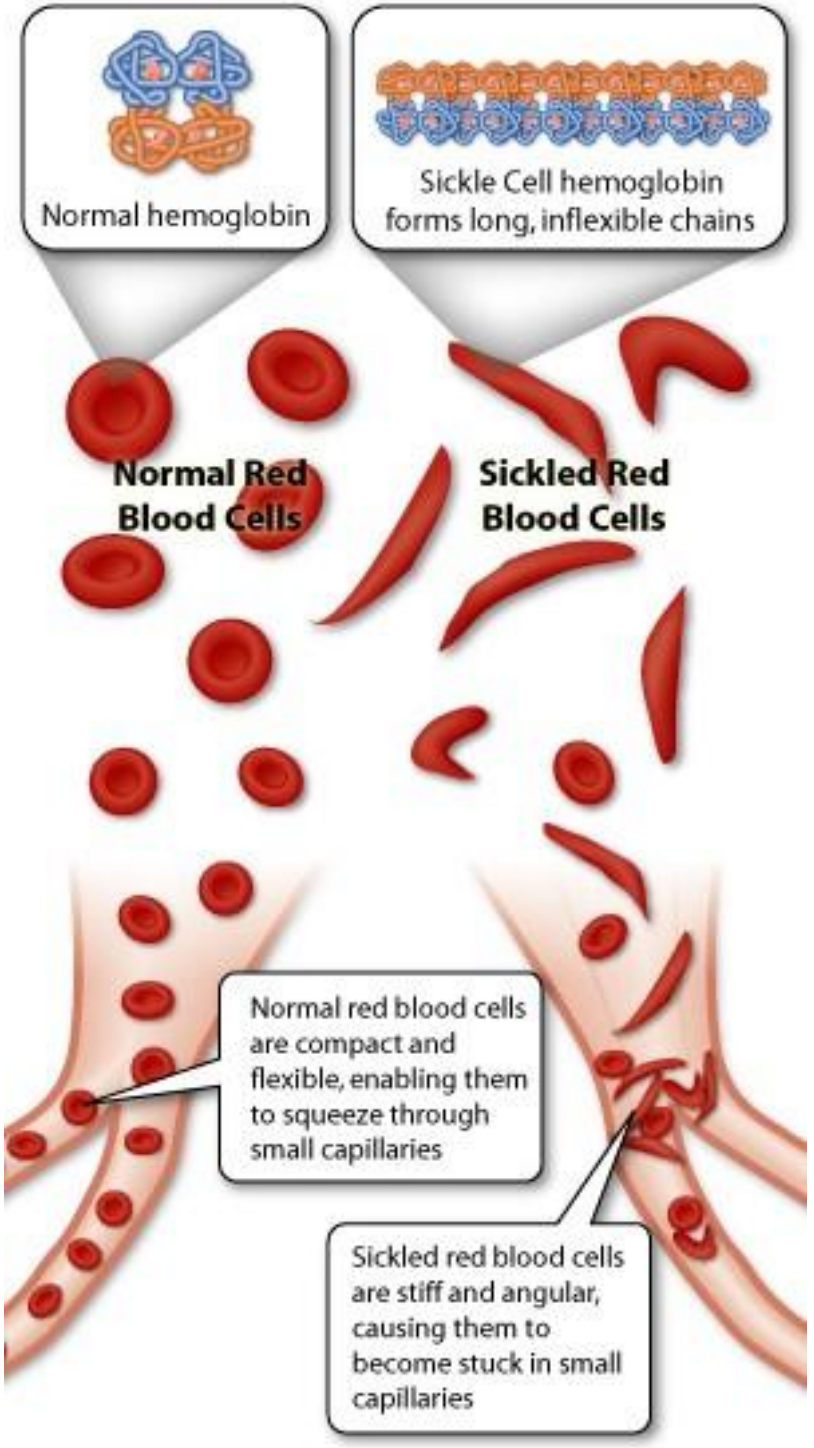




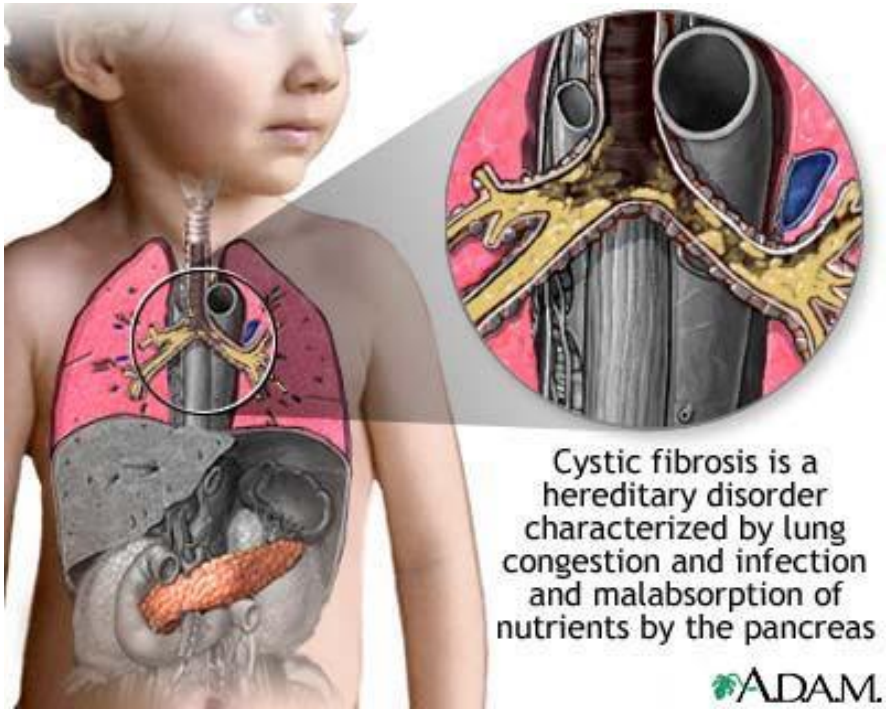
- Examples:

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

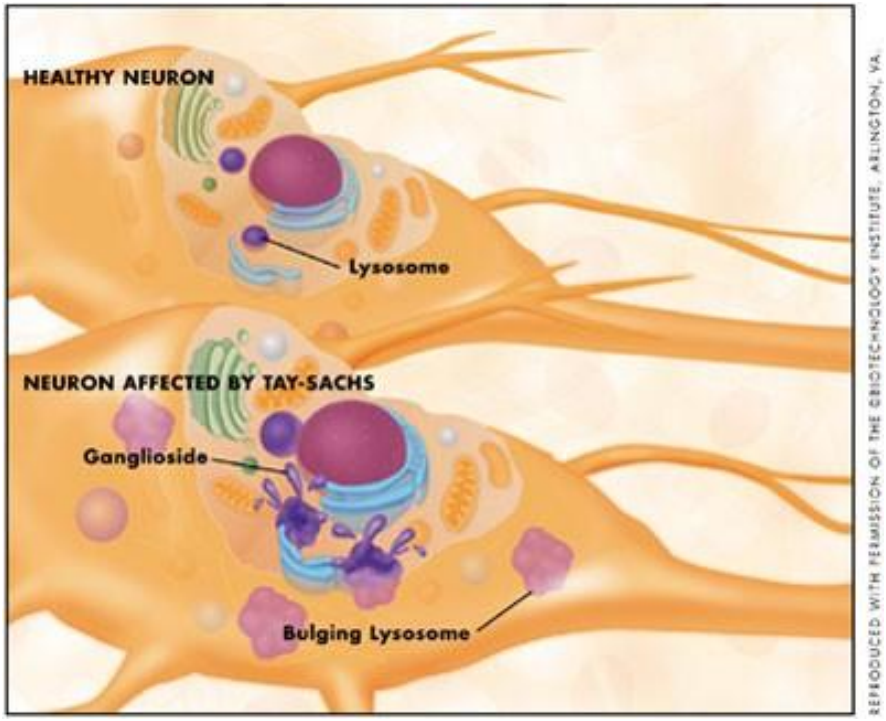


Cystic fibrosis – mucous builds up in the lungs



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

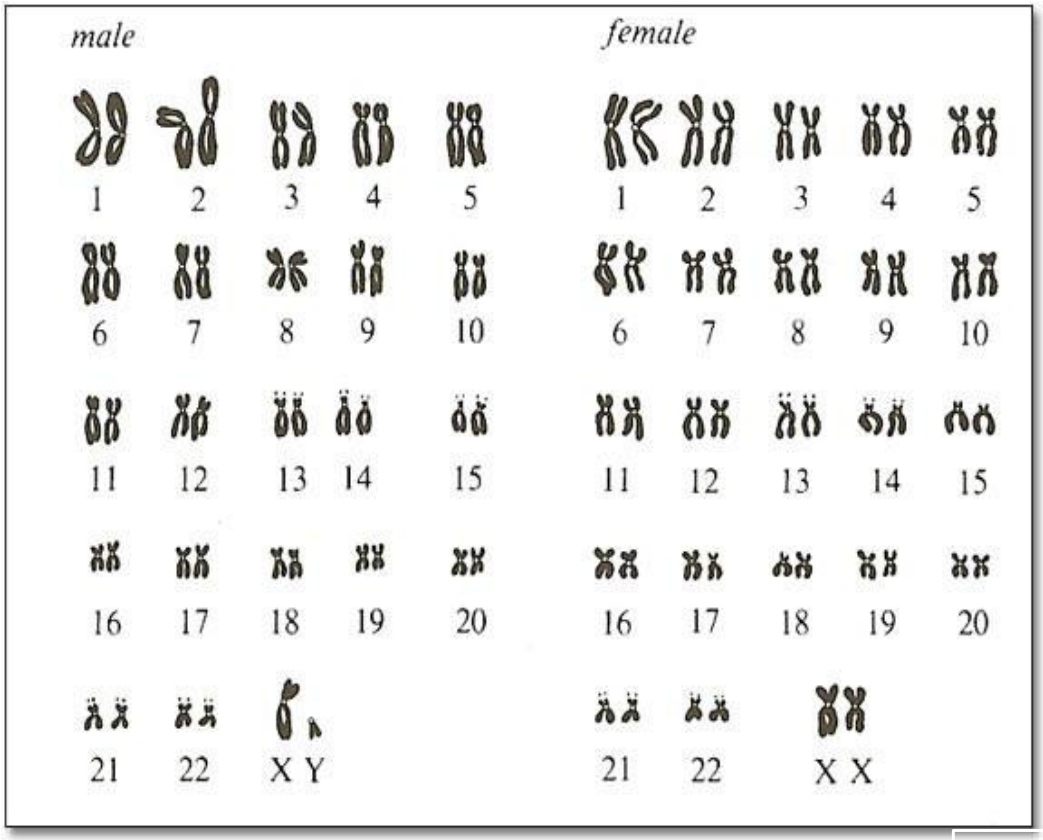
Dominant gene mutations:

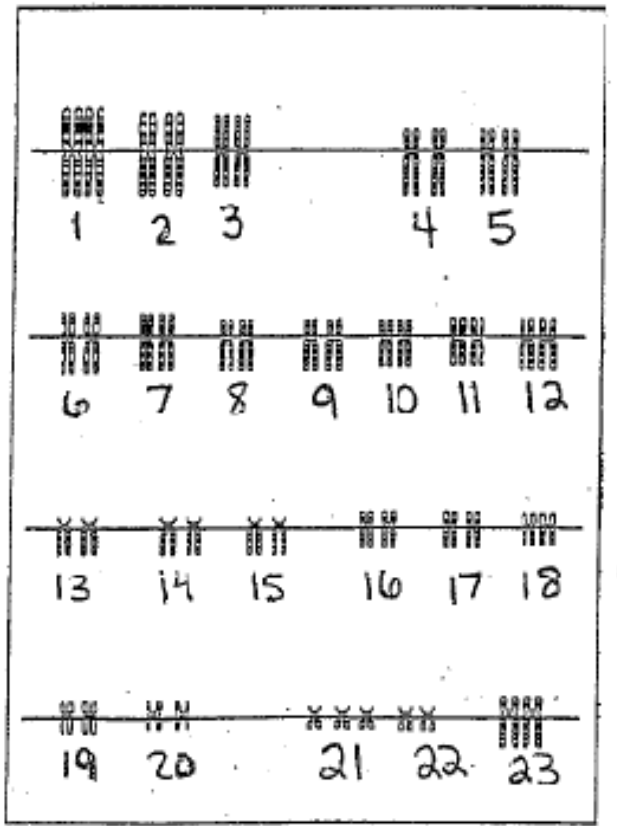
Huntington's disease – gradual deterioration of brain tissue, shows up in middle age and is fatal

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

THANK YOU