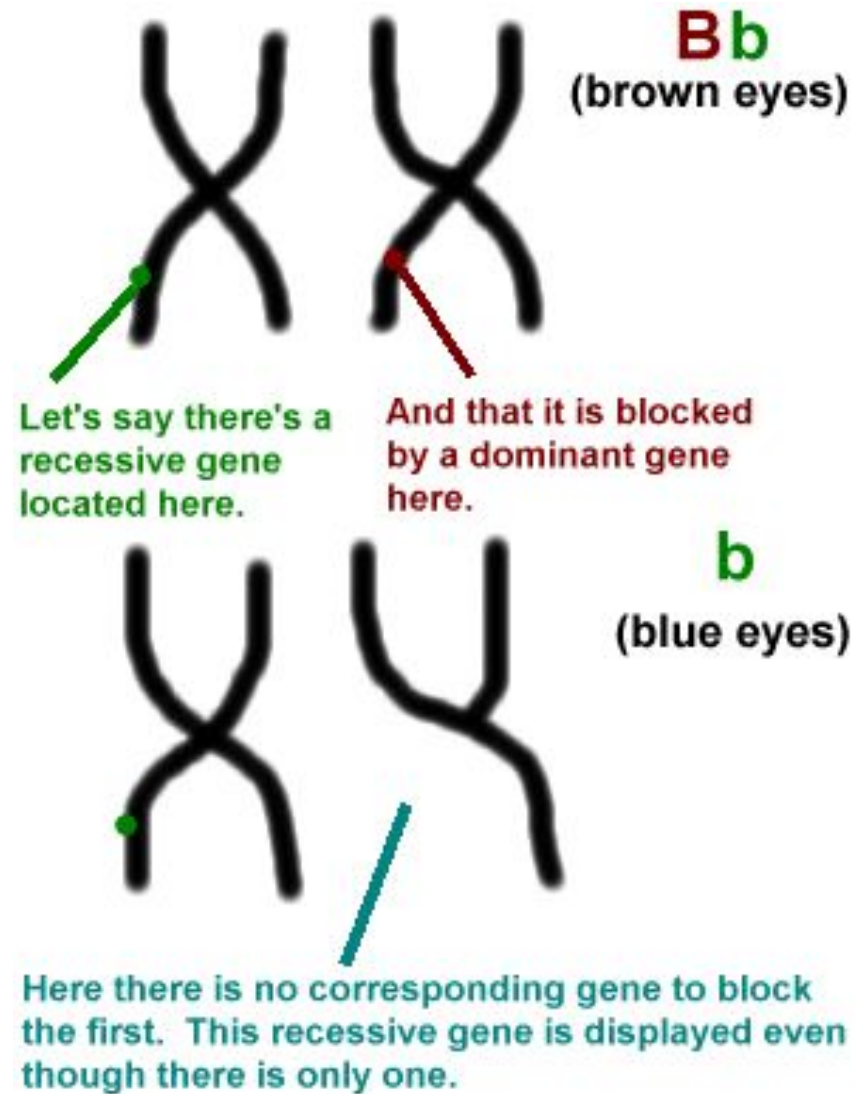


Genetic Disorders

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

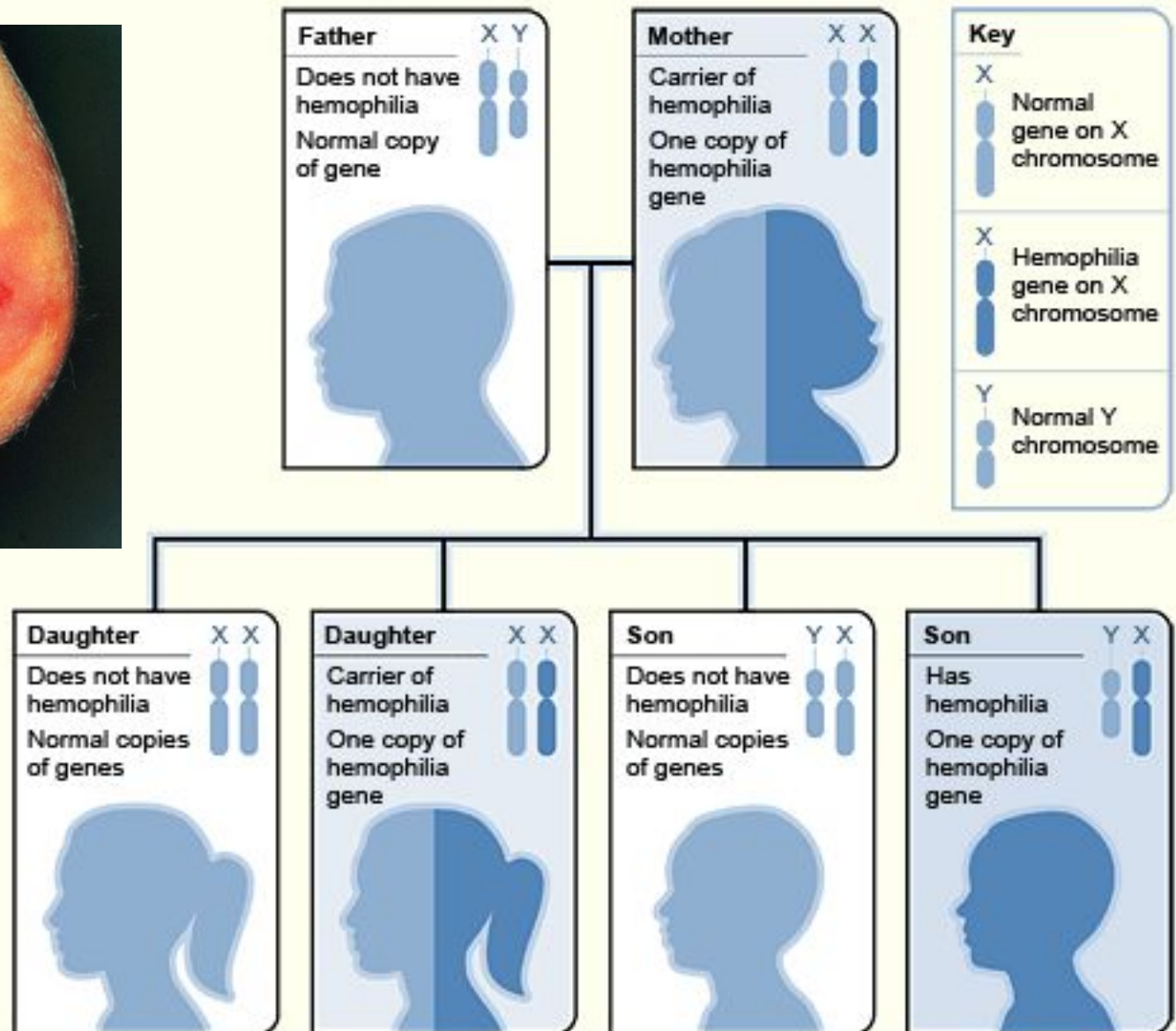


You should see **58**
(upper left), **18**
(upper right), **E**
(lower left) and **17**
(lower right).

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 carrier for colorblindness marries a male with normal vision.

Give the expected phenotypes of their children.

N = normal vision

n = colorblindness

X^N : Xⁿ



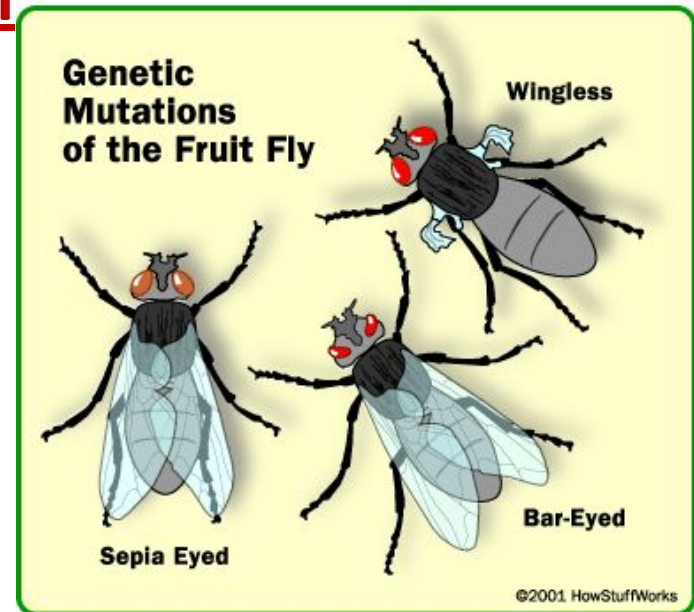
Phenotype:

1 normal

1 colorb

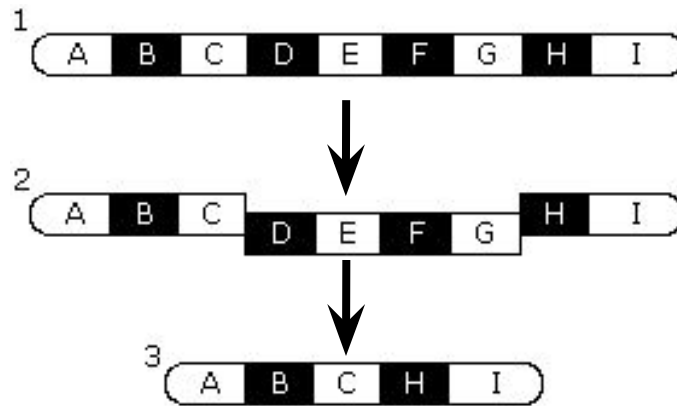
Mutations

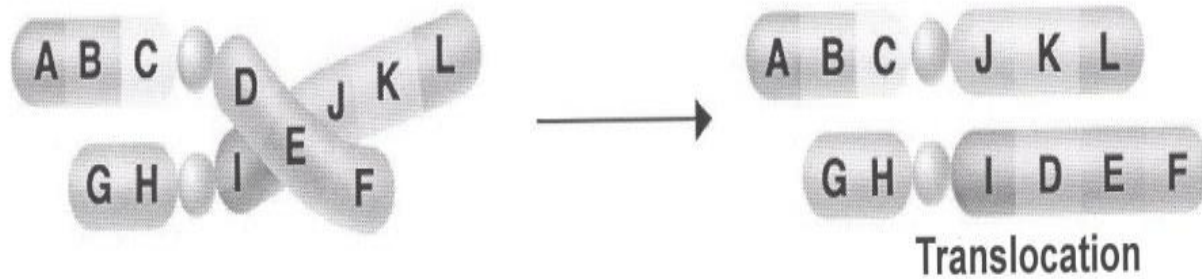
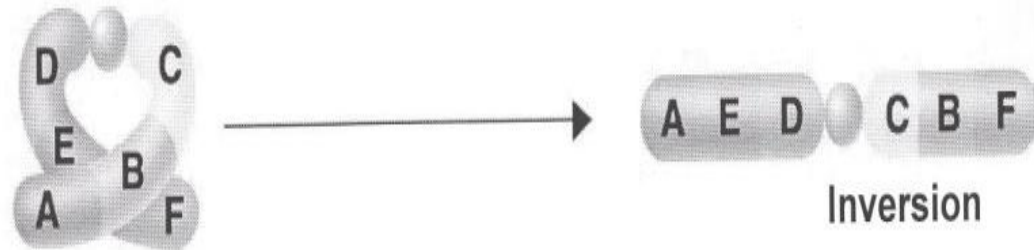
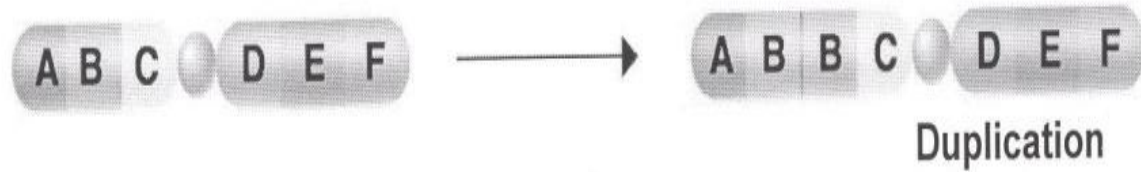
- Mutation – sudden genetic change (change in base pair sequence of DNA)
- Can be :
 - Harmful mutations – organism less able to survive: genetic disorders, cancer, death
 - Beneficial mutations – allows organism to better survive: provides genetic variation
 - Neutral mutations – neither harmful nor helpful to organism
- Mutations can occur in 2 ways: chromosomal mutation or gene/point mutation



Chromosomal mutation:

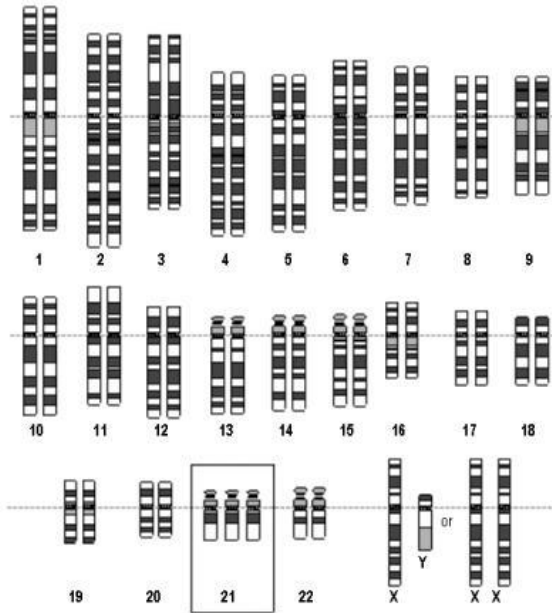
- less common than a gene mutation
- more drastic – affects entire chromosome, so affects many genes rather than just one
- caused by failure of the homologous chromosomes to separate normally during meiosis
- chromosome pairs no longer look the same – too few or too many genes, different shape





- Examples:

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

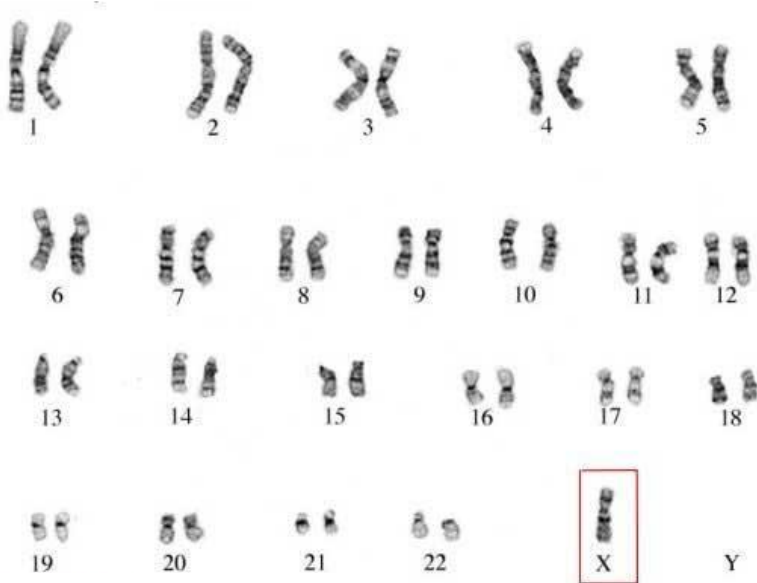


flattened
nose and face,
upward slanting
eyes,



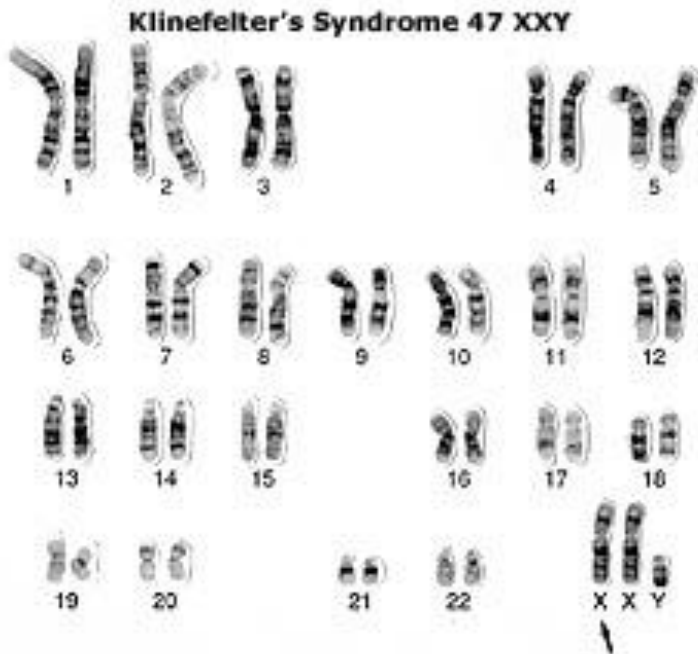
Turner's syndrome – only 45 chromosomes, missing a sex chromosome (X)

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



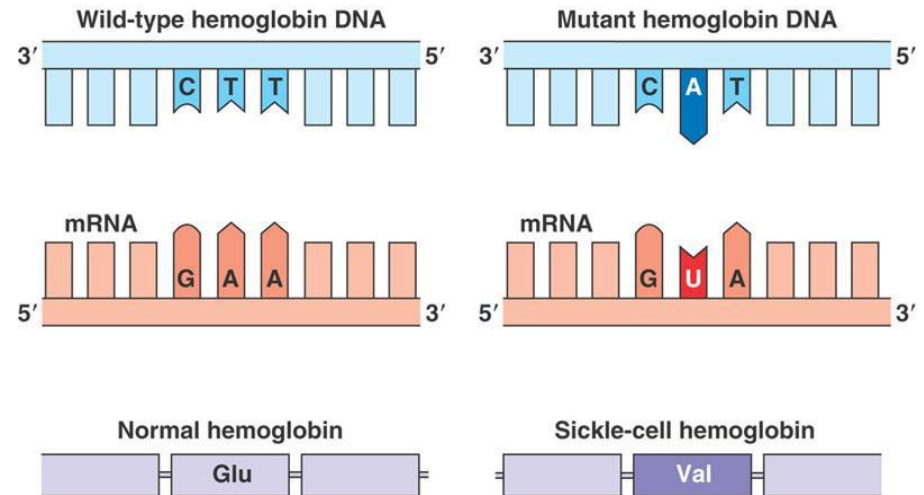
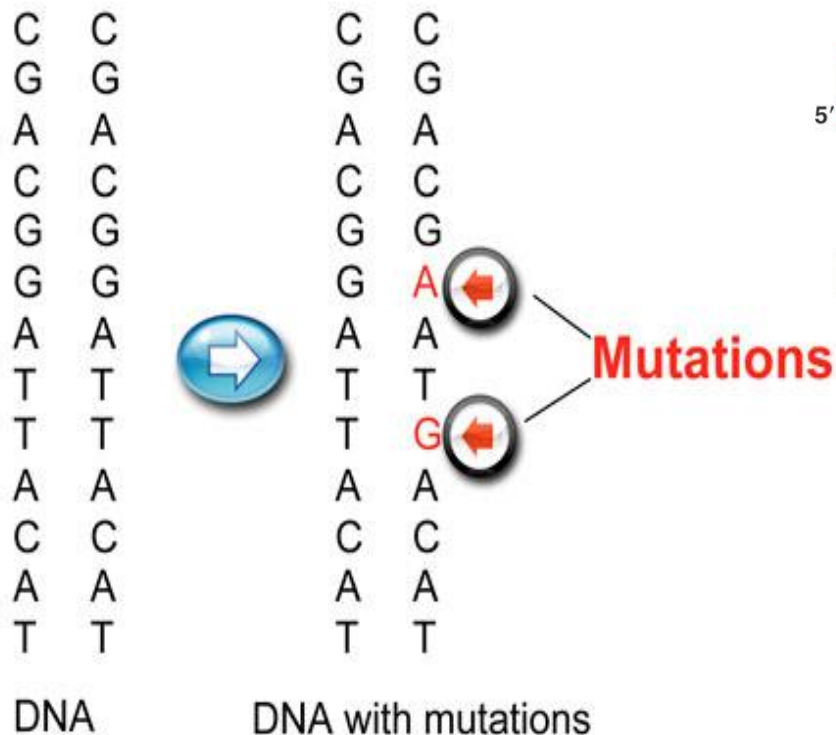
- Having an extra set of chromosomes is fatal in animals, but in plants it makes them larger and hardier.



Hardier

Gene or Point Mutation

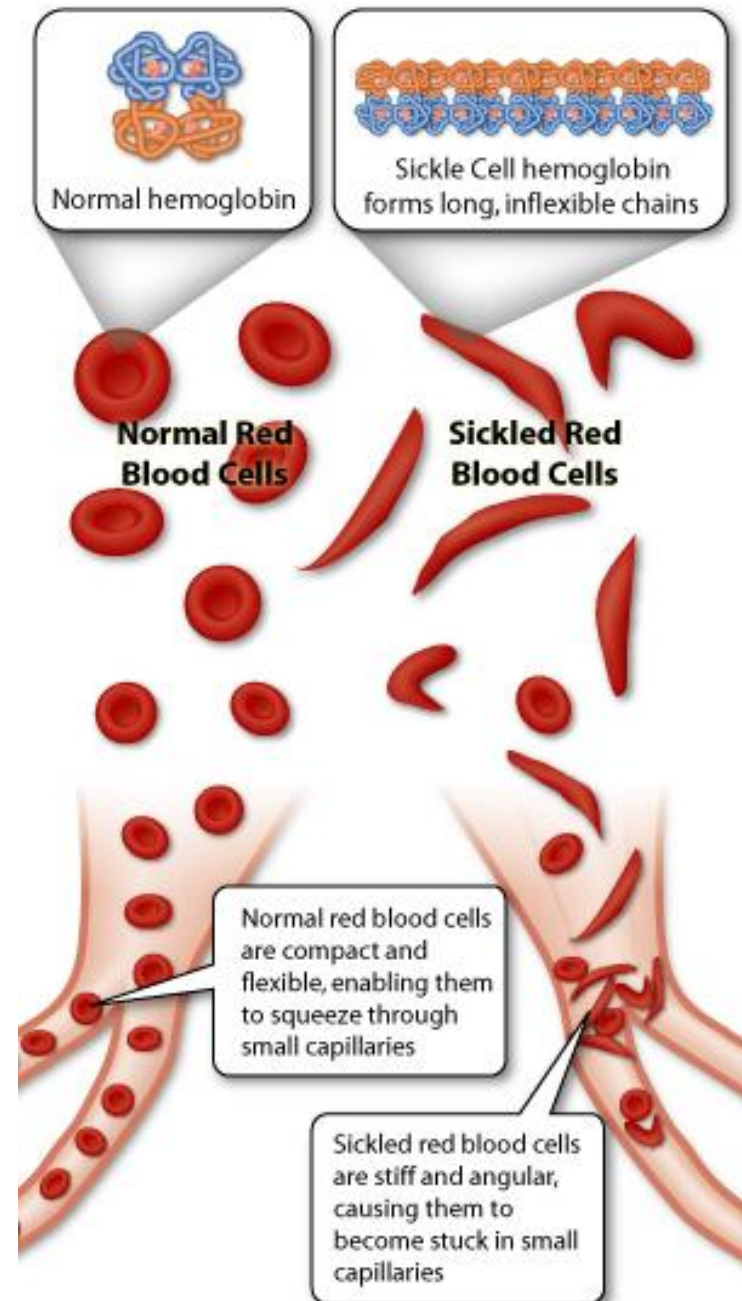
- most common and least drastic
- only one gene is altered



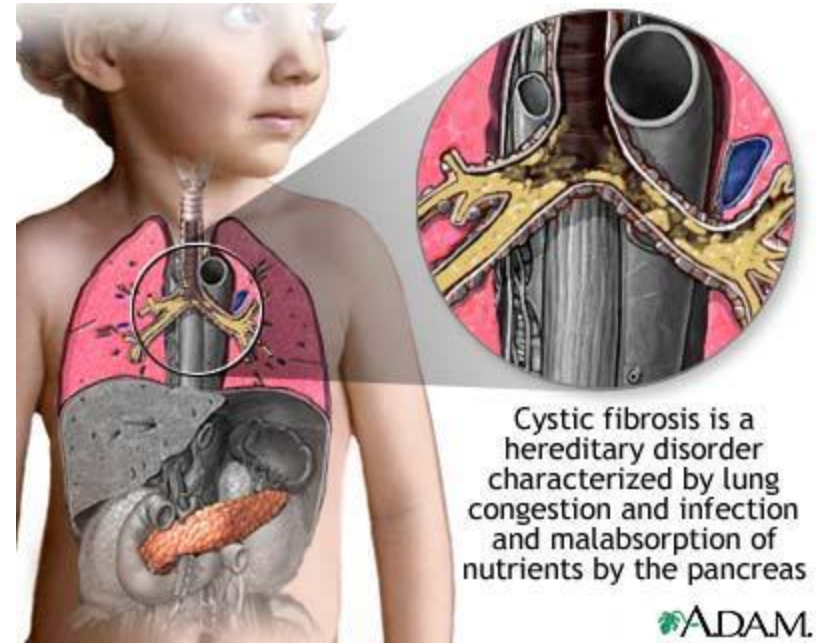
- Examples:

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

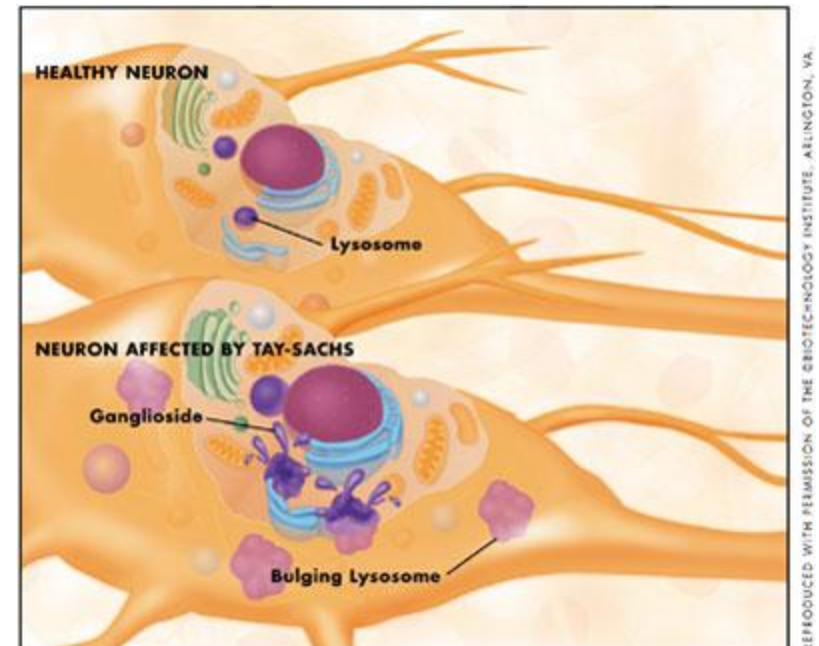


Cystic fibrosis – **mucous** builds up in the **lungs**



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:

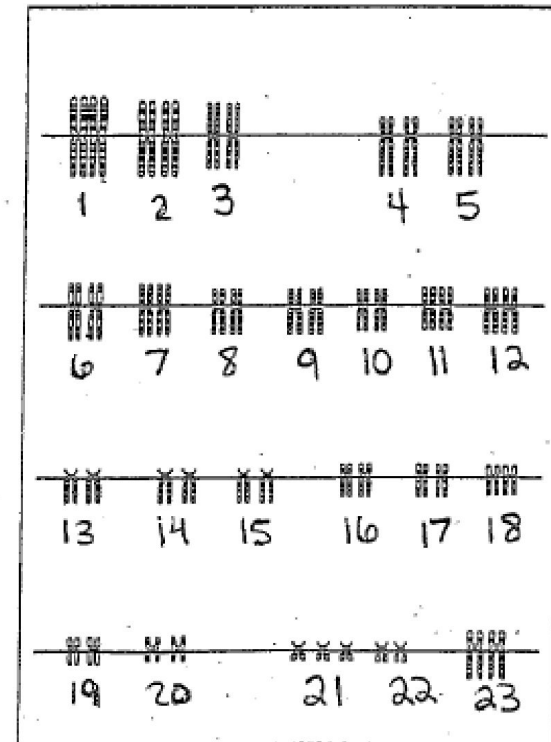
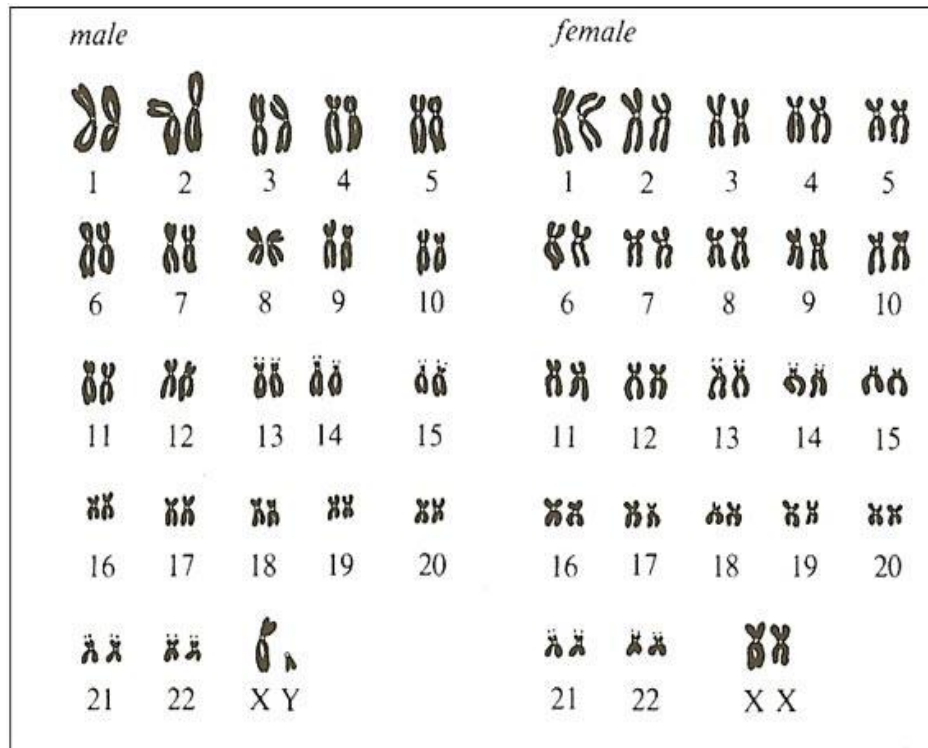
Huntington's disease – gradual **deterioration** of **brain tissue**, shows up in **middle age** and is **fatal**

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