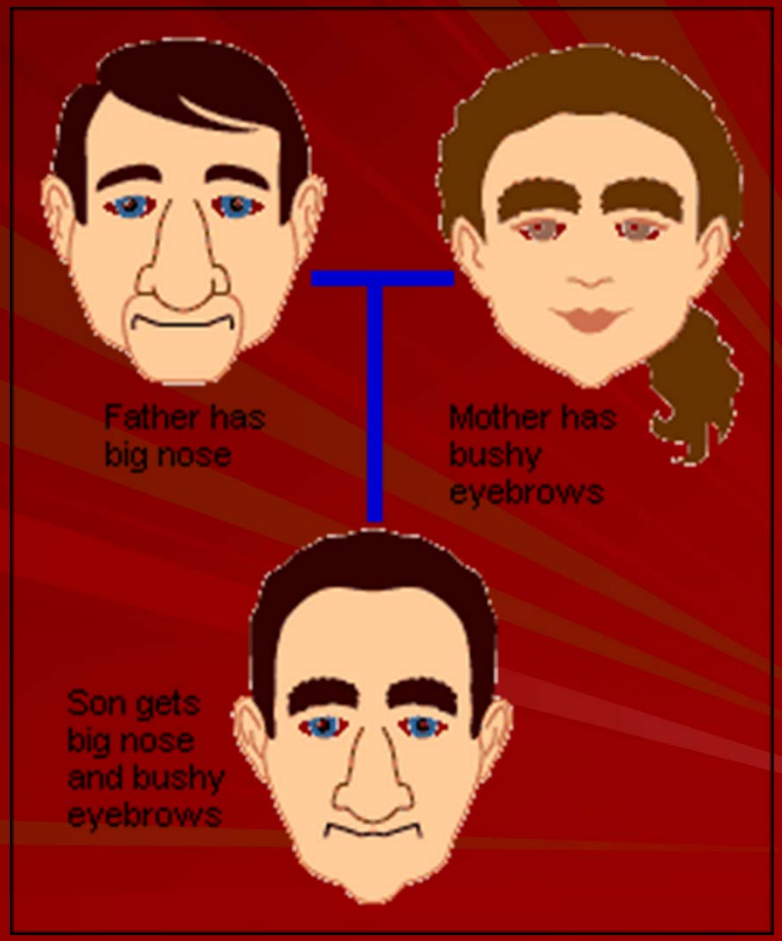
Mutations and Genetic Disorders

what is inheritance

Genetic Shuffling

- Shuffling of genes during sexual reproduction can introduce new gene combinations into a population.
- This is a source of genetic variation which can lead to evolutionary change.



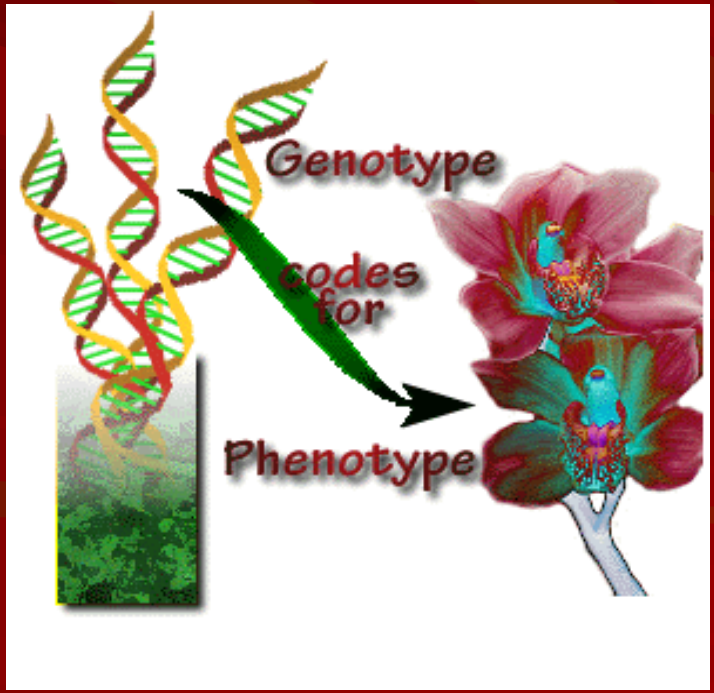
Gene vs. Alleles

- ***Gene:*** specific part of the chromosome that contains the coded information to make a specific polypeptide that influences a particular trait or characteristic in the organism.
- ***Allele:*** There are usually two or more ways for a trait to appear. Alleles are the alternate forms of the genes.

  Ex: in pea plants, genes carry information about the color of the seeds. Alleles are the actual color of the seeds – either green (G) or yellow (g).

Phenotype vs. Genotype

- **Phenotype:** physical or biochemical characteristics of an organism
- **Genotype:** genetic makeup of an organism



Genotype of a Trait

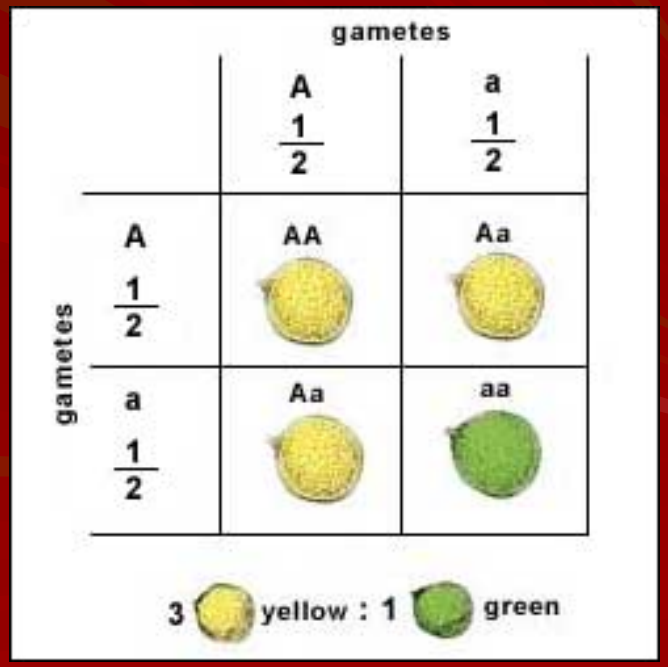
- In our example of pea seed color, seeds can either be green or yellow.
- If G is the allele for green color and g is the allele for yellow color then there can be three allelic combinations of genes for pea pod color.

Genotypes: ***Phenotypes:***

GG green

Gg green

gg yellow



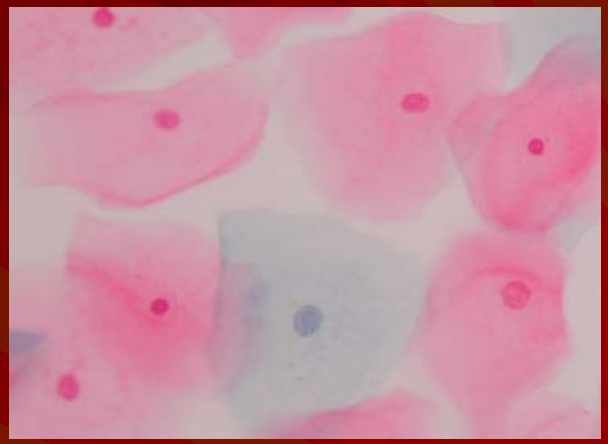
what is a trait

What is a Mutation?

- A **MUTATION** is a change in the bases in a section of DNA.
- Mutations are random and unrelated to how “useful” the change would be for the organism.
- Mutations can be beneficial, harmful or have no effect on the functioning or appearance of the organism in any way.

Where Do Mutations Occur?

- **Somatic Mutations:** occur in body cells and cannot be inherited.



- **Reproductive Mutations:** occur in sex cells and can be passed to offspring.

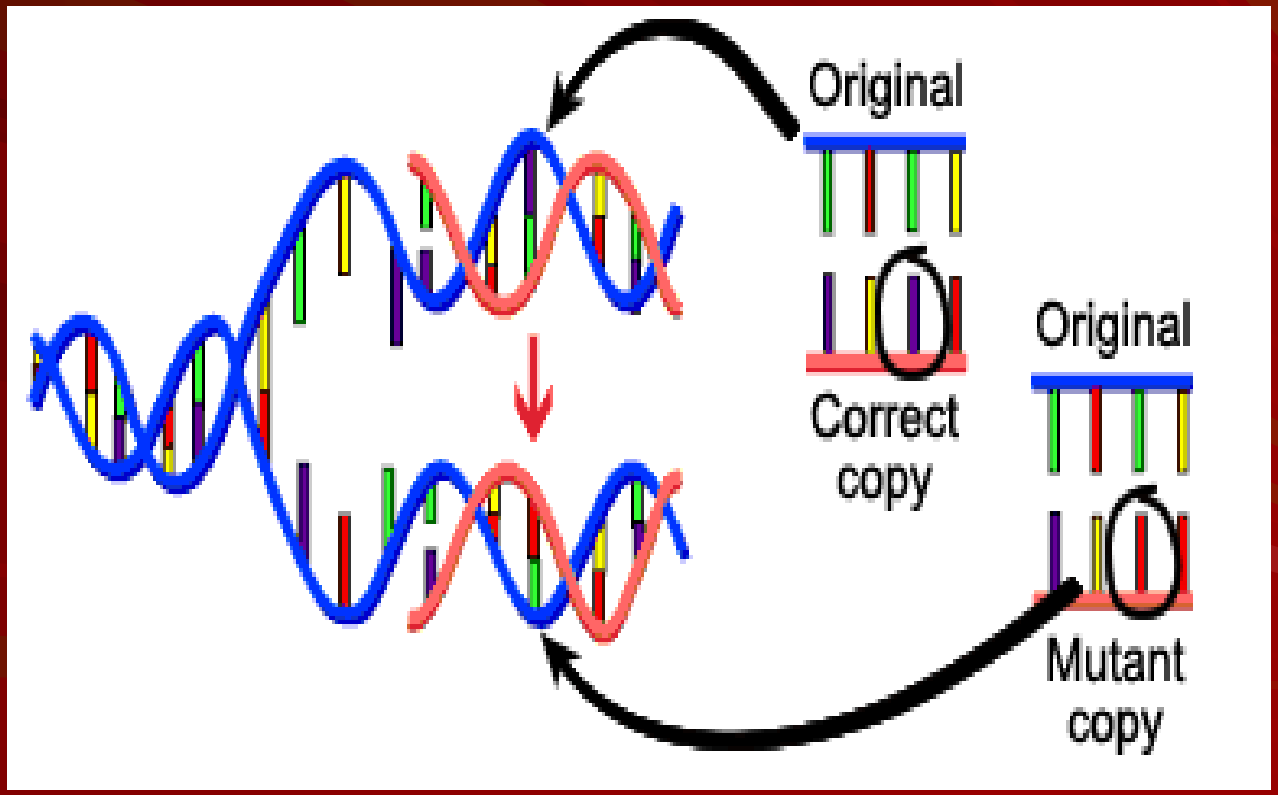
Causes of Mutations

- **Environmental Causes**

-- radiation
-- x-rays
-- cosmic rays
-- ultraviolet radiation
-- benzene
-- cigarette smoke
-- Agent Orange

Causes of Mutations

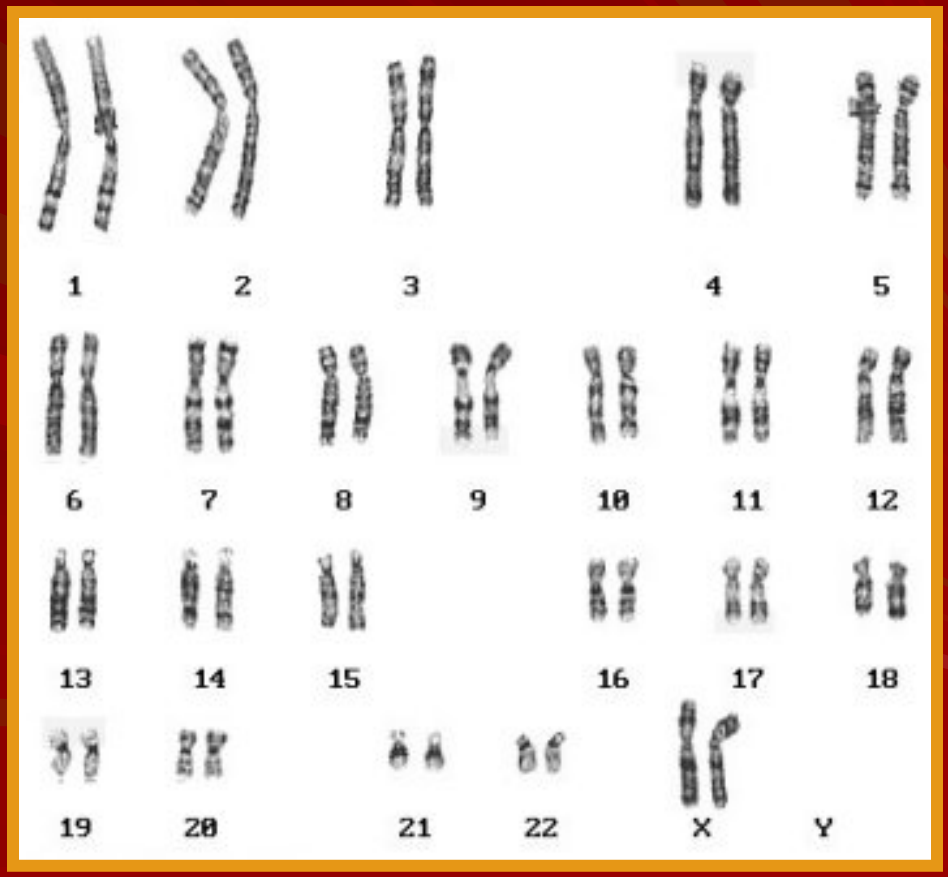
- DNA Fails to Copy Correctly resulting in **Gene Point Mutations**



Causes of Mutations

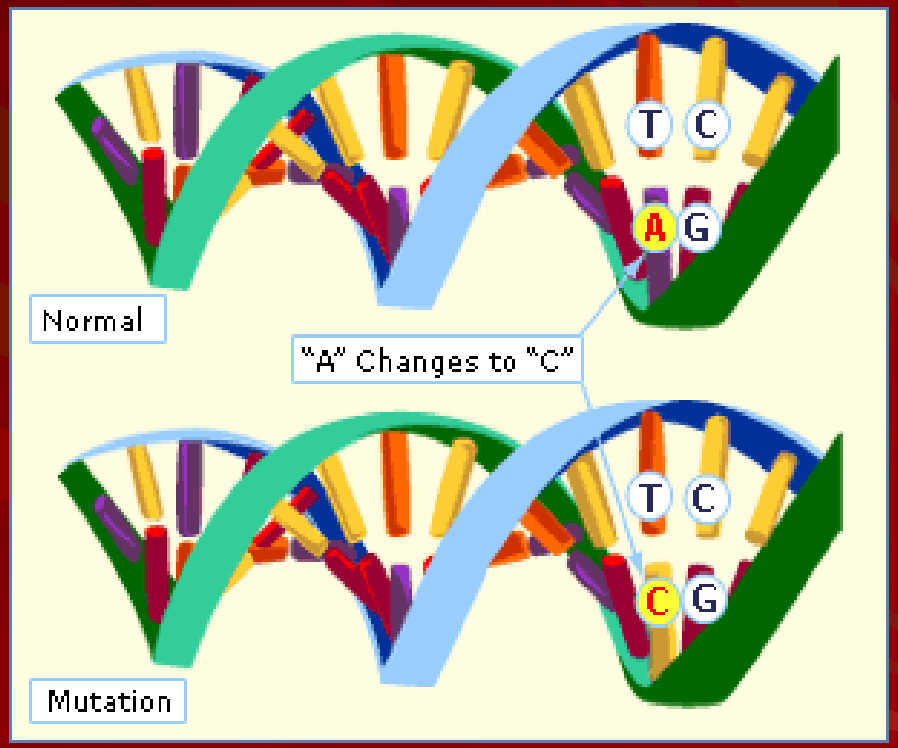
- Changes in chromosomal structure or number

Female Karyotype:
The photomicrograph of the 2n number of chromosomes displayed with respect to number, size, and form of the chromosomes.



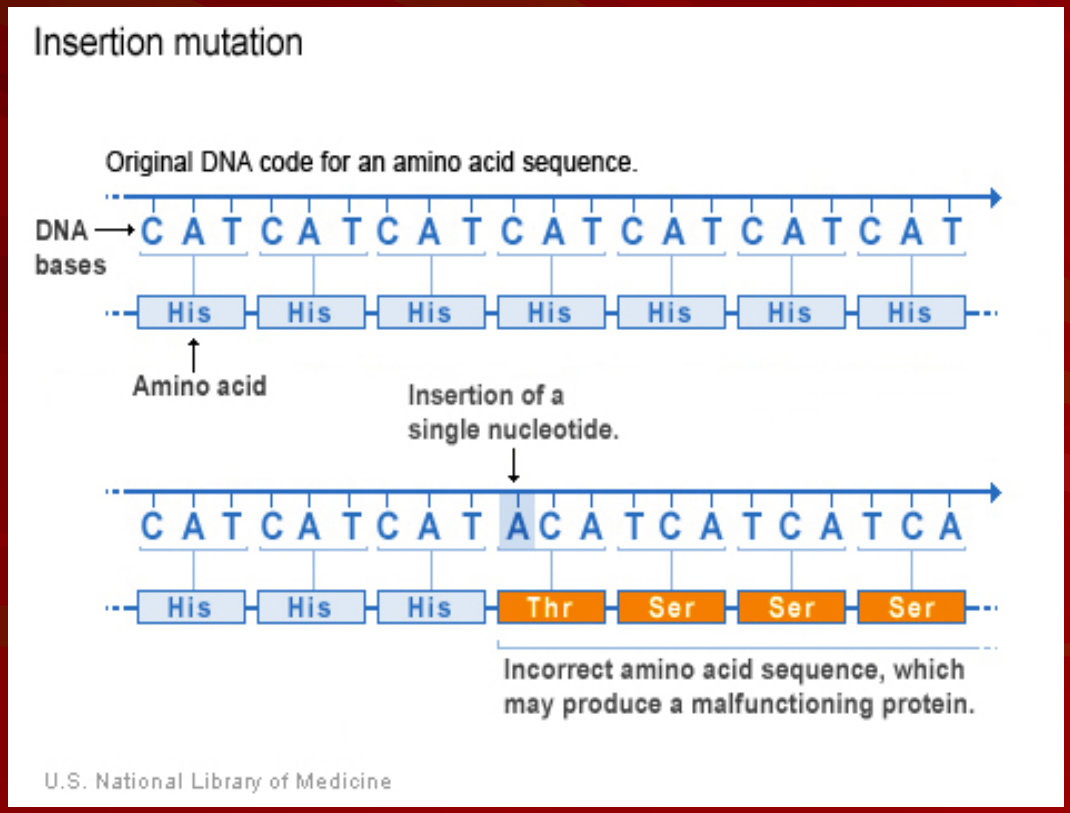
Types of Gene Point Mutations

- **Substitution:**
  a change in the structure of DNA in which one base is exchanged for another.



Types of Gene Point Mutations

- **Insertion:** extra base or bases are inserted into a new place in the sequence of the DNA.



Types of Gene Point Mutations

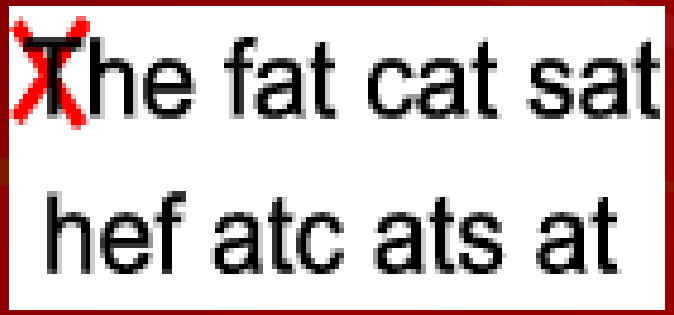
- **Inversion:** mutations where there is a reversal in a DNA triplet base pair as seen below.

Original DNA: C A T T A G G A T

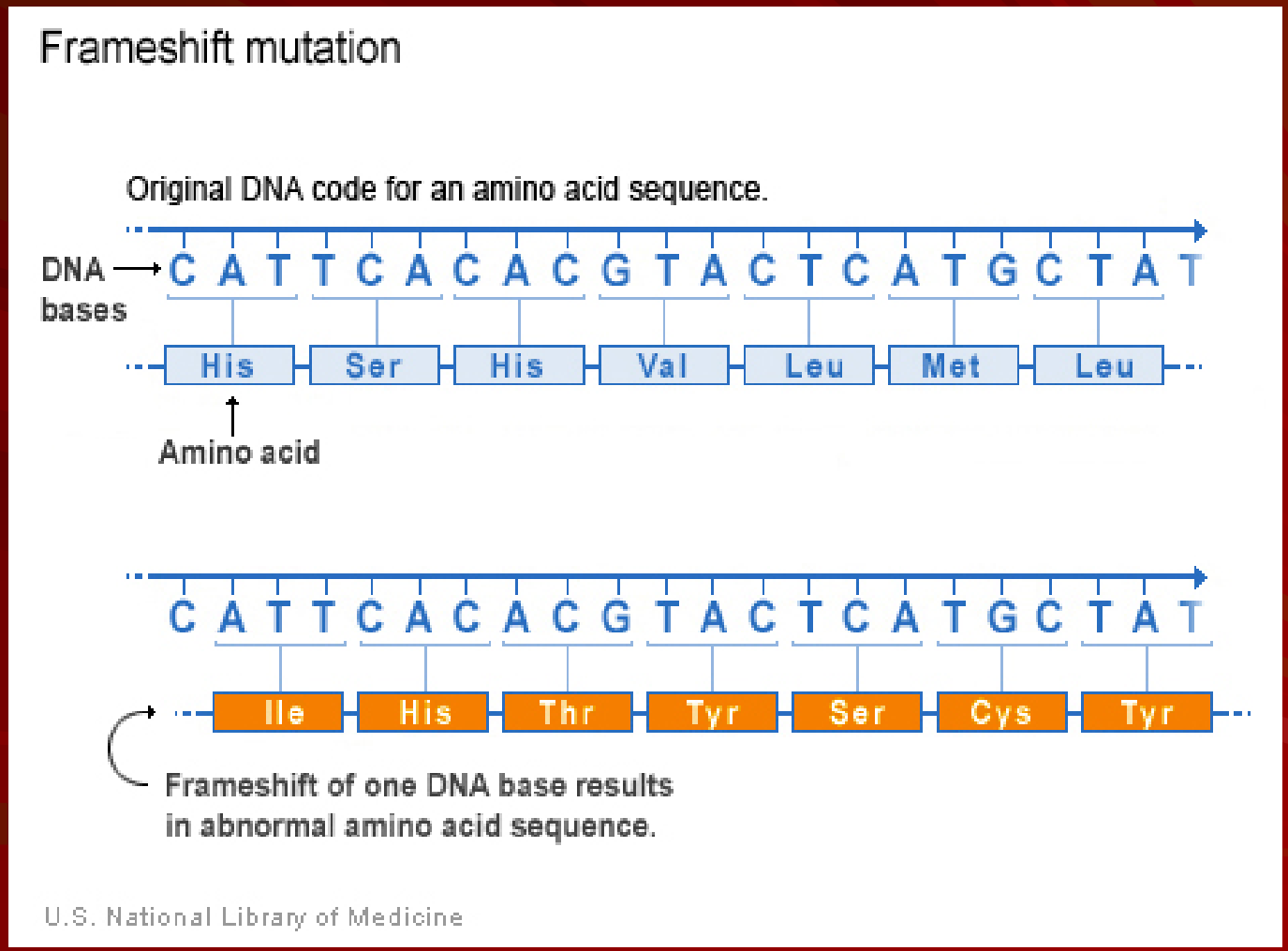
Inversion: C A T G A T G A T

Types of Gene Point Mutations

- **Frameshift:** DNA is divided into triplets that are three bases long. Insertions and deletions can alter a gene so its message is no longer correctly paired.
- In frameshifts, errors that occur at the DNA level cause the triplets to be paired, read, and translated incorrectly, generating proteins that are often useless.



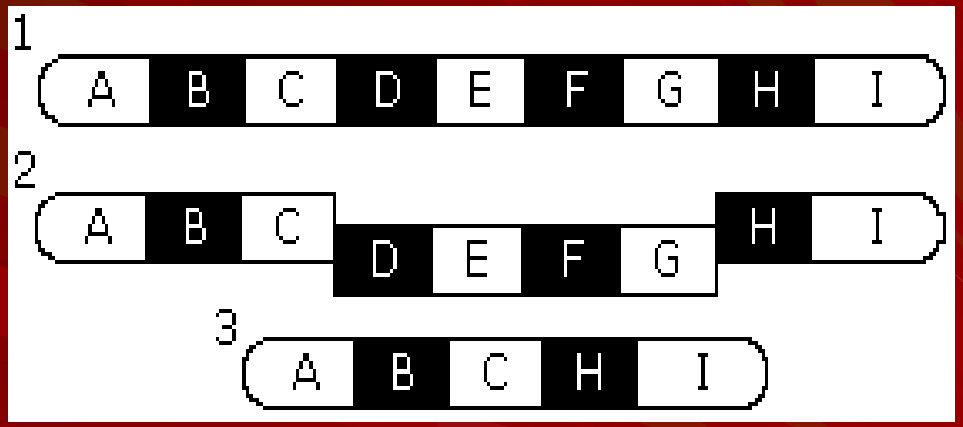
Types of Gene Point Mutations



Chromosomal Mutations: Deletion

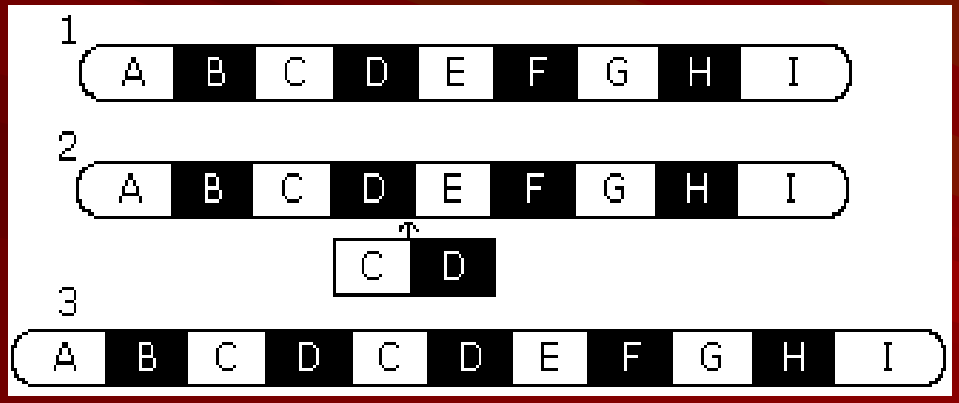
- Image 1: normal chromosome with genes ABCDERGHI
- Image 2: Genes DEFG separate from chromosome and are lost
- Image 3: New chromosome only with genes ABCHI
- New chromosome lacks certain genes which may prove fatal depending on how important these genes are

Deletion of genes DEFG



Chromosomal Mutations: Duplication

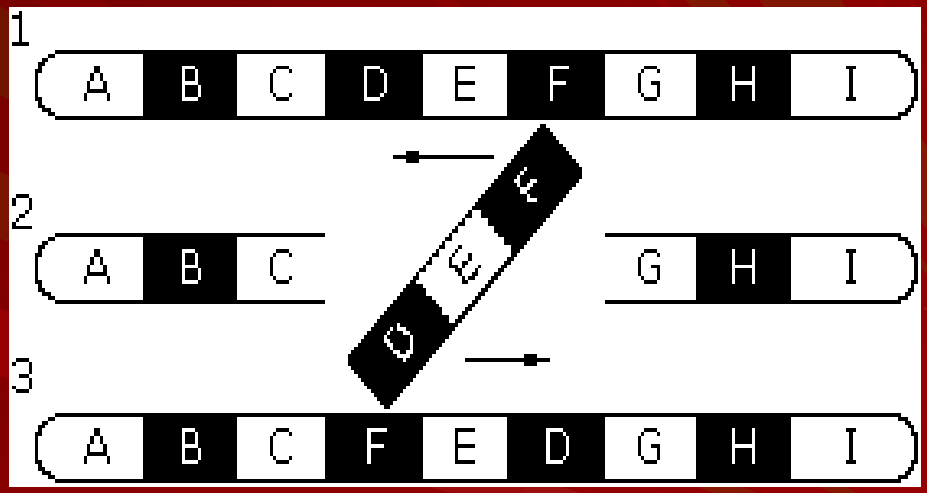
***Duplication* of Genes C & D**



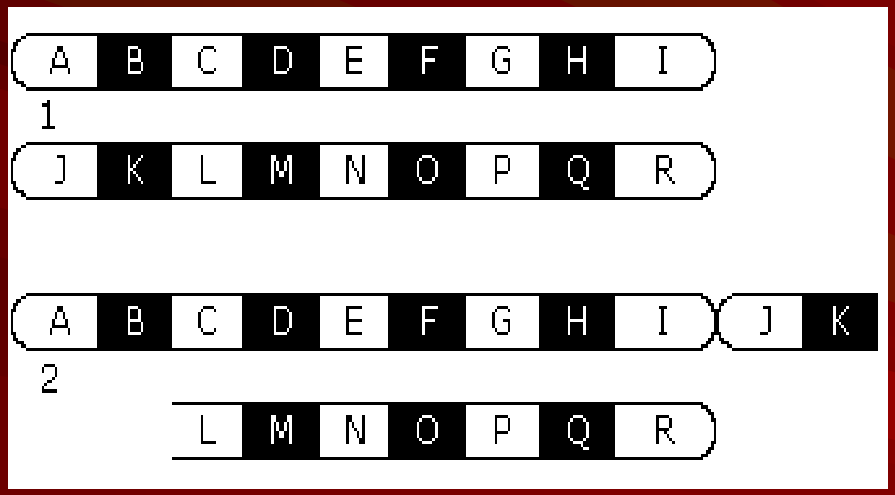
- Image 1: Normal chromosome before mutation
- Image 2: Genes from the homologous chromosome are copied and inserted into the genetic sequence
- Image 3: New chromosome possesses all its initial genes plus a duplicated one, which is usually harmless

Chromosomal Mutation: Inversion of Genes

- Image 1: Normal chromosome before mutation
- **Image 2:** The connection between genes break and the sequence of these genes are reversed
- Image 3: The new sequence may not be viable to produce an organism, depending on which genes are reversed. Advantageous characteristics from this mutation are also possible



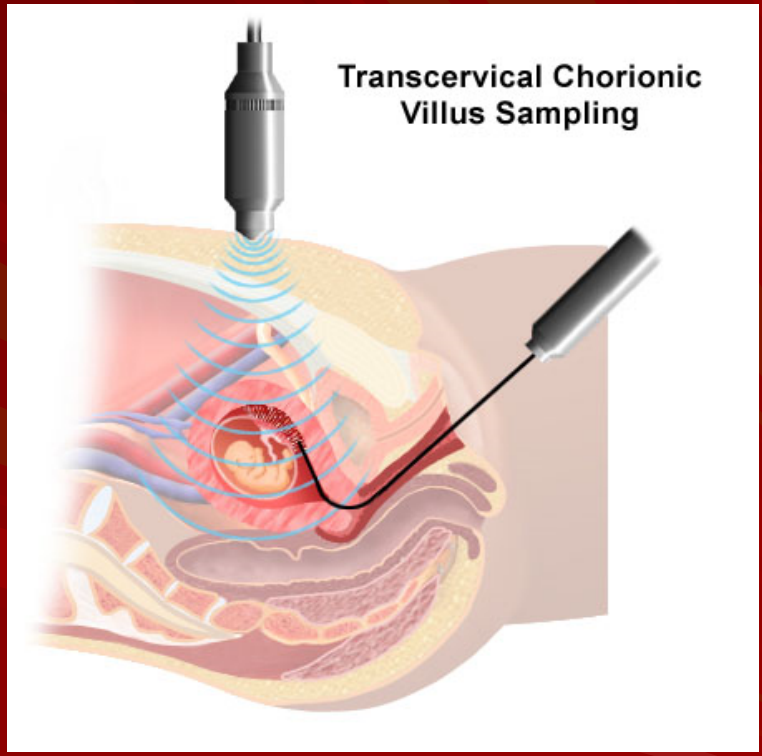
Chromosomal Mutations: Translocation of Genes



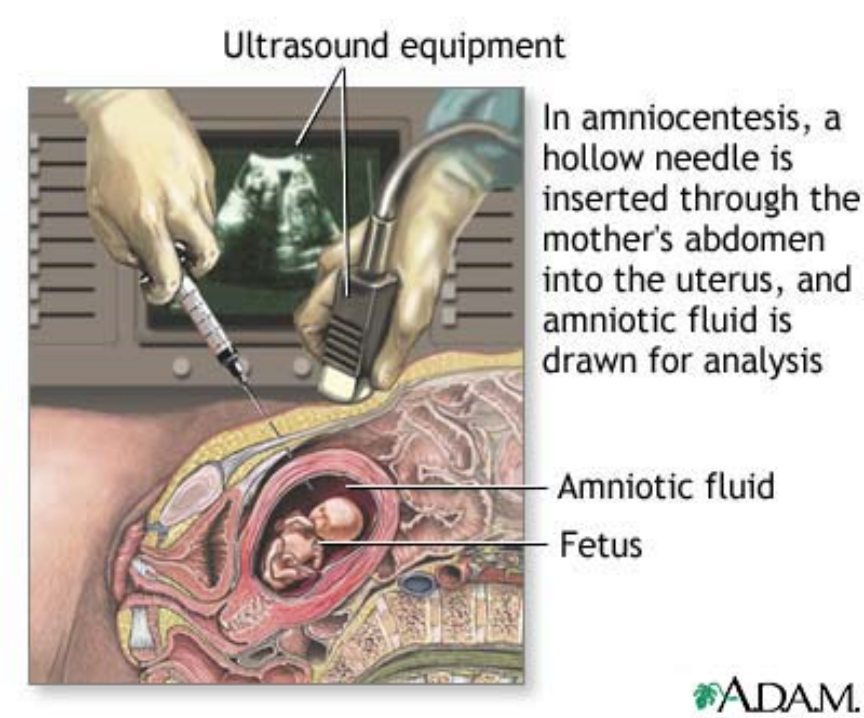
- This is where information from one of two homologous chromosomes breaks and binds to the other.
- **Image 1:** Usually this sort of mutation is lethalAn un-altered pair of homologous chromosomes
- **Image 2:** Translocation of genes has resulted in some genes from one of the chromosomes attaching to the opposing chromosome

Diagnostic Tests: Chorionic Villi Testing

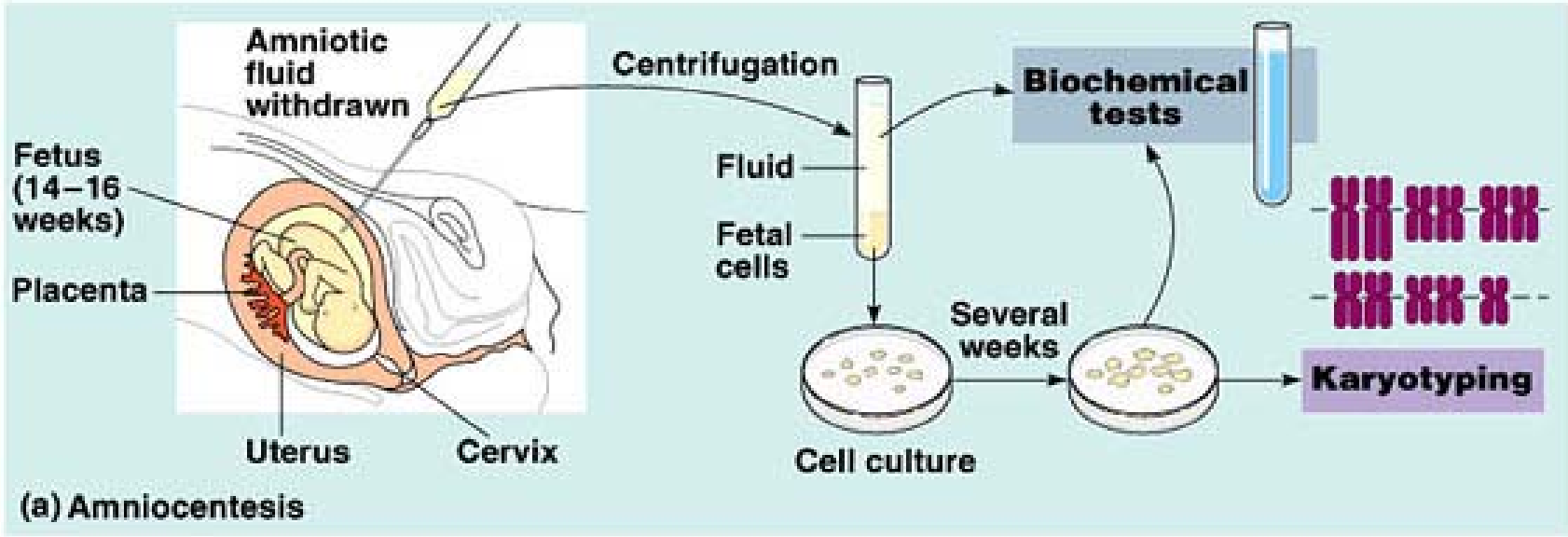
- Used to diagnose certain birth defects in the first trimester
- A small bit of placental tissue is gently suctioned into a syringe. Since placental tissue is formed from embryonic tissue, it is genetically identical to the fetus and can be analyzed for potentially dangerous birth defects.
- The incidence of miscarriage is a bit higher when compared to amniocentesis.

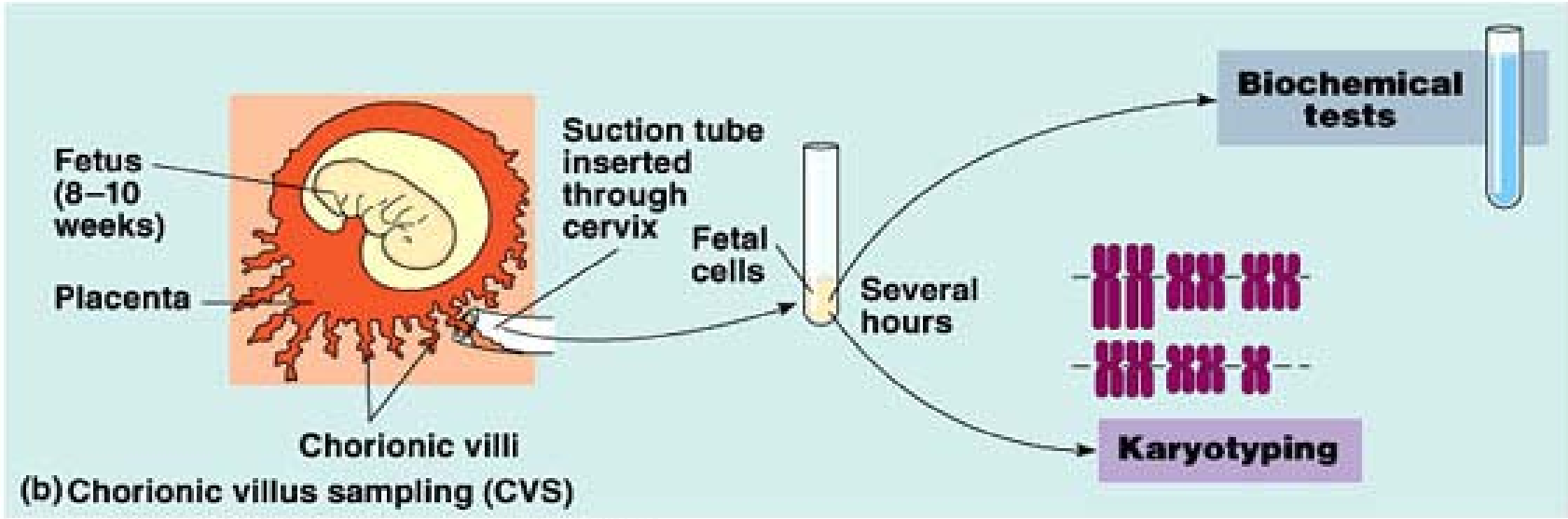


Diagnostic Tests: Amniocentesis



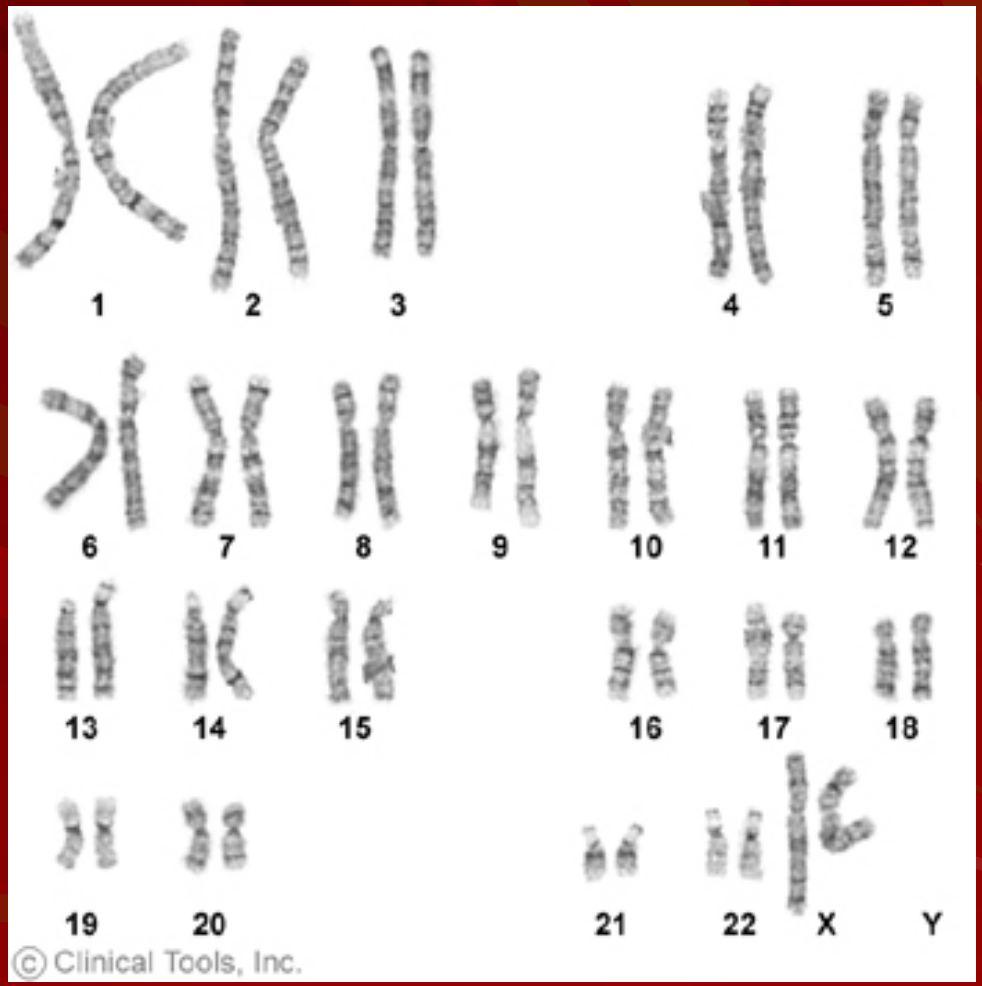
- By examining the amniotic fluid, chromosome abnormalities, neural tube defects, and genetic disorders can be detected.
- Generally performed during the 14th -20th week of pregnancy.
- Risks of miscarriage are from 1:200 to 1:400.





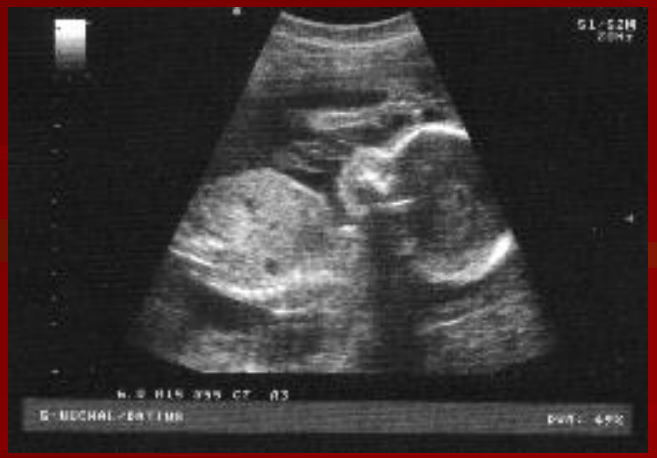
Diagnostic Tests: Karyotyping

- Enlarged picture of homologous chromosomes in a cell.
- Chromosomes are examined for abnormalities.
- Sex of fetus can also be determined.



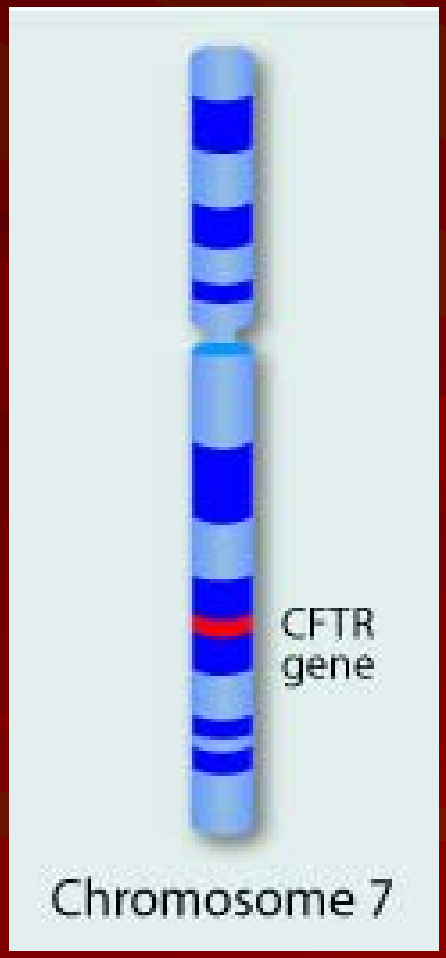
Diagnostic Tests : Obstetric Ultrasound

- The examination can be performed transabdominally, usually with a full bladder to achieve better visualization, or transvaginally with a special vaginal transducer.
- Having a full bladder for the transabdominal portion of the exam is helpful because sound travels through fluid which helps to better visualize the uterus which lies posteriorly to the bladder.
- The procedure is painless, noninvasive, and safe.



Fetus at 22 weeks

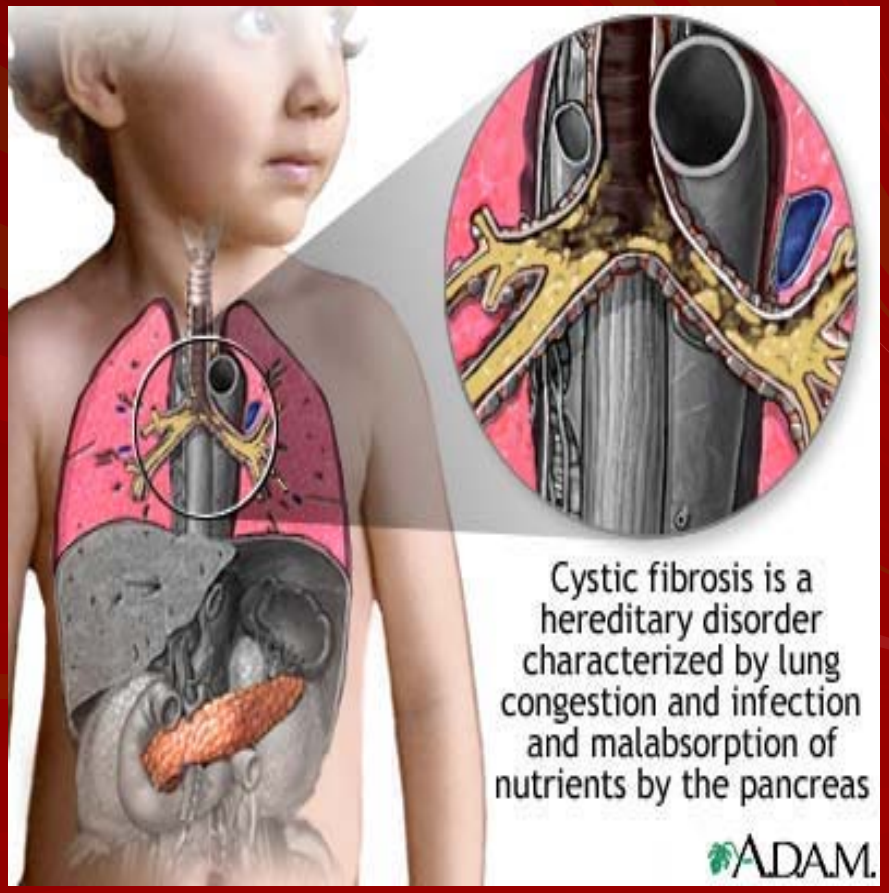
Genetic Disorders: Cystic Fibrosis



- CF occurs in about 1 out of every 2,500 births among Caucasian children, with a far lower incidence rate among Asian and Black children.
- CF is caused by a mutation to a gene on Chromosome 7.

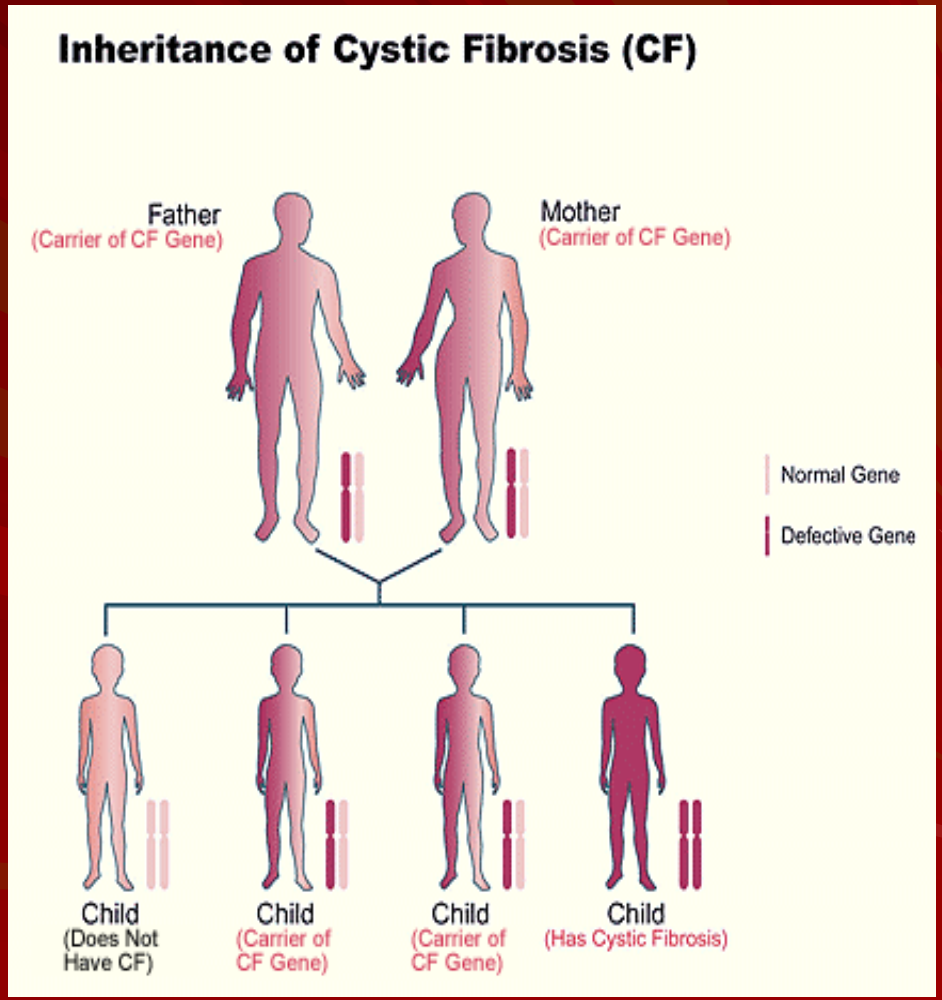
Genetic Disorders: Cystic Fibrosis

- CF is a inherited, life threatening disease that causes mucus build-up in the lungs and gastrointestinal tract.
- Thick mucus causes breathing problems and promotes bacterial infection, causing inflammation of the airways, leading to lung damage.
- Mucus build-up in the digestive tract and pancreas prevents enzymes to get to the small intestine, so proper digestion can occur.



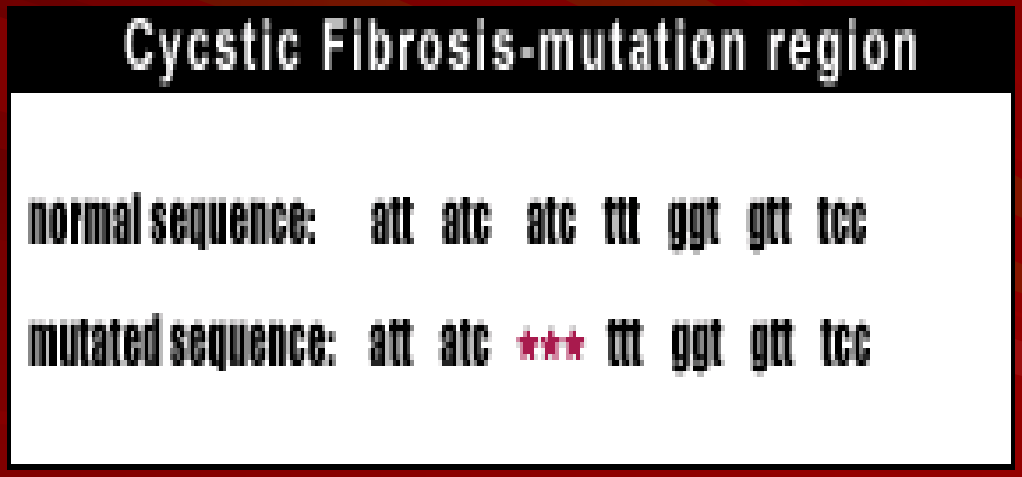
Genetic Disorders: Cystic Fibrosis

- People with cystic fibrosis have two mutant genes (one from each parent) that do not make the proper protein to permit the movement of chloride ions across the cell membrane.



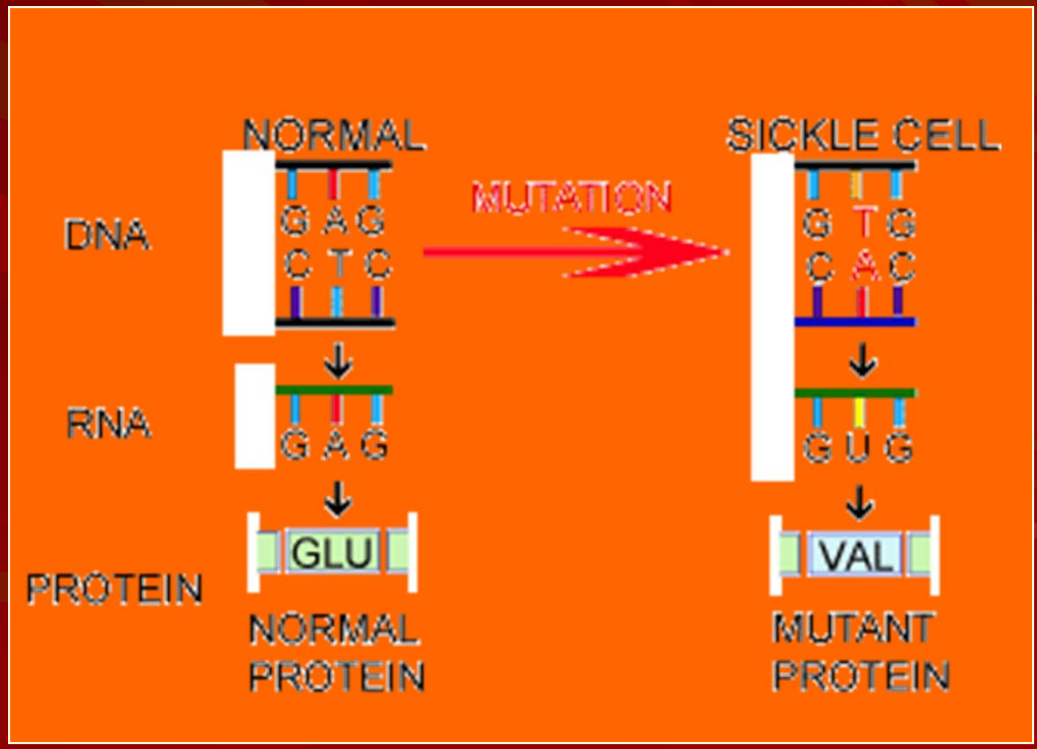
Genetic Disorders: Cystic Fibrosis

- There are now more than 500 different mutations of the CFTR gene that are known to cause CF.



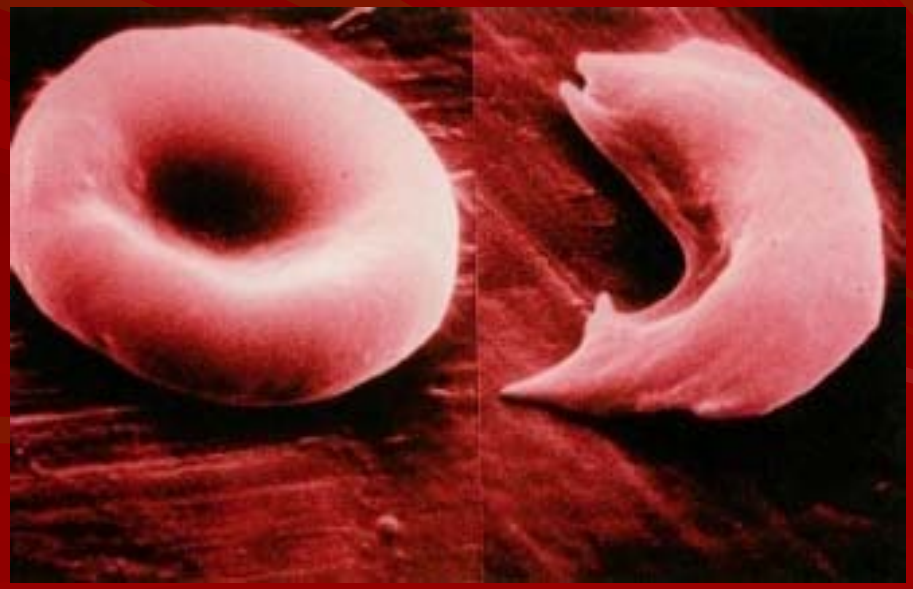
Genetic Disorders: Sickle Cell Anemia

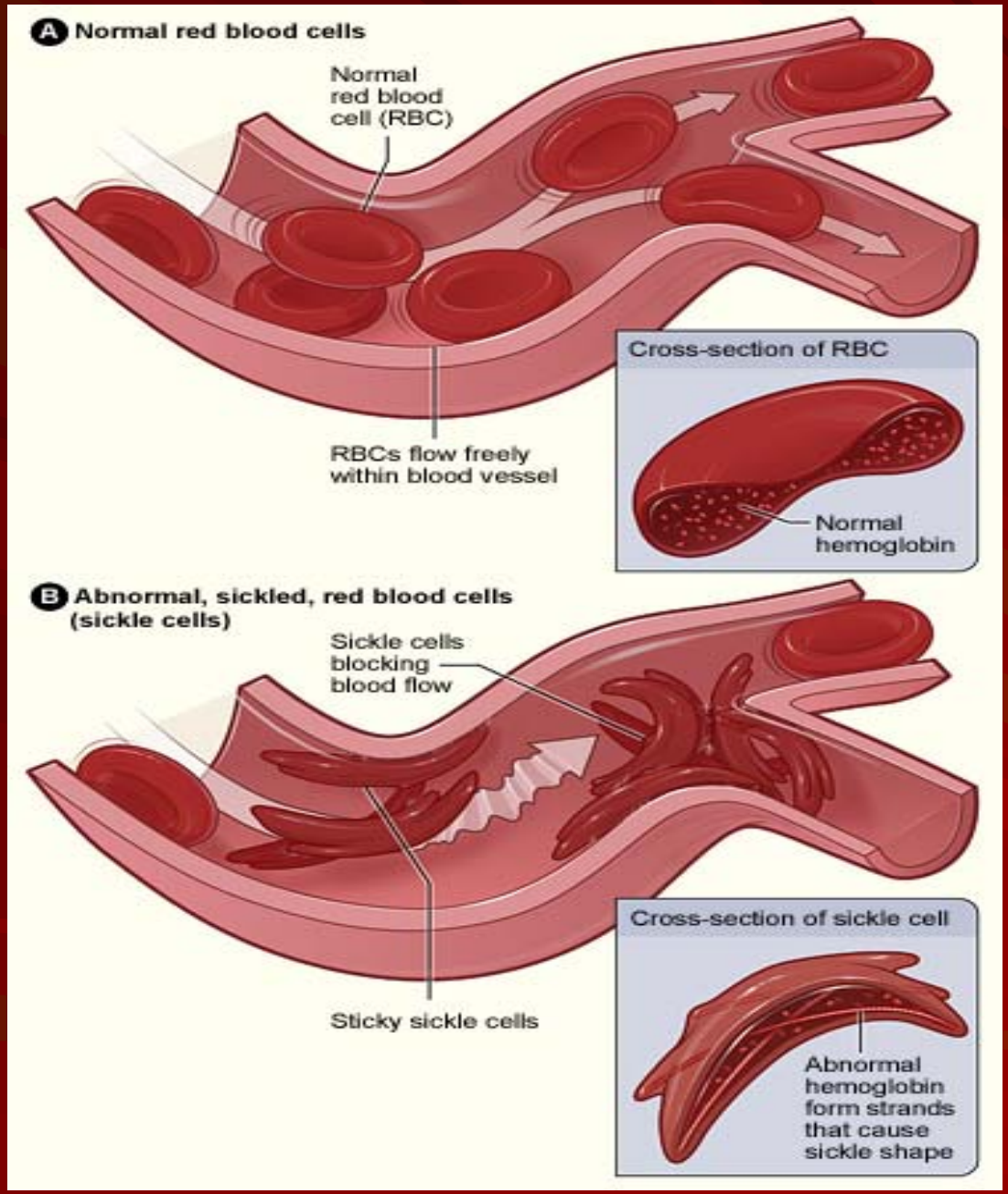
- This disease is caused by a mutated version of the gene that helps to make hemoglobin, the red respiratory pigment that carries oxygen on the red blood cells. People with two copies of this gene have the disease; those with one copy of the gene do not have the disease but can pass the gene onto their children.



Genetic Disorders: Sickle Cell Anemia

- Sickle Cell Anemia is genetically inherited disease most commonly associated with people of African American descent. People with sickle cell anemia are very tired, lack energy due to an insufficient amount of oxygen in their cells to carry on aerobic respiration to release needed energy for metabolic processes, and are often in severe pain from the abnormally shaped red blood cells as they move through the capillaries.





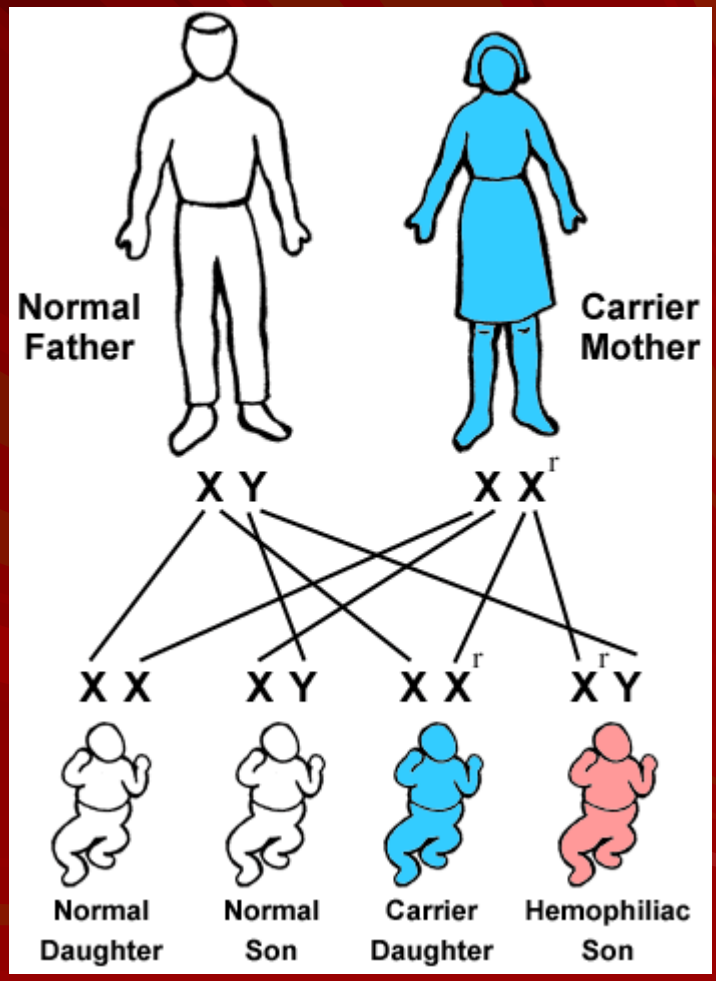
Blood Flow with Normal and Sickled Cells

Genetic Disorders: Albinism

- Inherited condition that is recessive (must have a gene from each parent)
- All people with albinism have vision problems. Most have blue eyes, but some individuals have red or violet eyes.
- Pigmentation is weak or non-existent, leaving individuals with very pale skin, white hair and eyelashes.
- Very sensitive to light and with little melanin, must wear sun block to prevent severe burning in the sun.

Genetic Disorders: Hemophilia

- Rare inherited blood disorder due to the inability to code for a clotting factor required to form normal blood cells.
- Hemophilia more common among boys since it is a sex-linked recessive disorder inherited from the mother.
- Afflicted individual can bleed from gums, in joints, urine and stool. Children often have multiple bruising.



Genetic Disorders: Tay-Sachs

- More common among individuals of eastern European Jewish origin.
- A mutated gene results in the insufficient activity of an enzyme that breaks down fatty materials in the brain.
- The accumulation of fat causes progressive damage to nerve cells.
- During the first year of life, symptoms begin with loss of peripheral vision and abnormal startl response.
- Baby slowly loses motor skills &experiences swallowing and breathing difficulties.
- The child will become blind, mentally retarded, paralyzed and unresponsive to the external environment. Most afflicted children die by the age of 5.

Genetic Disorders: Phenylketonuria (PKU)

- An abnormal gene results in the inability to metabolize the amino acid phenylalanine.
- Phenylalanine accumulates in the body tissues and brain, causing irreversible brain damage and other neurological problems.
- Treatment involves a lifetime maintenance of a diet low in phenylalanine consumption.

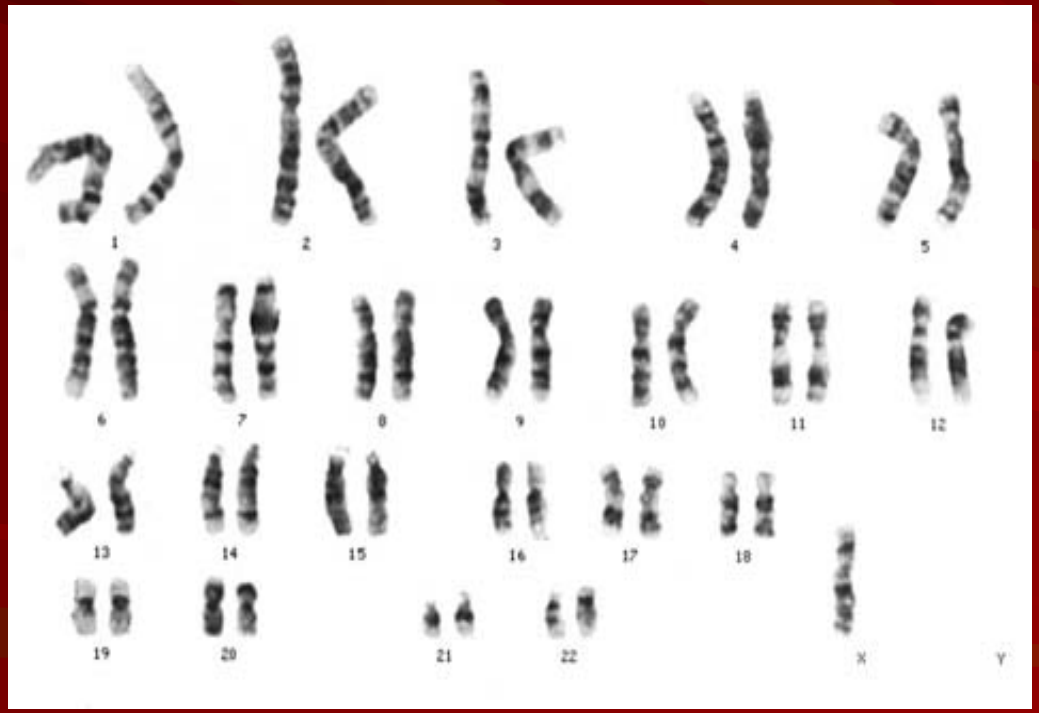
Just some foods that PKU sufferers have to limit
Image credit: pdphoto.org

Genetic Disorders: Down Syndrome

- Caused by an extra 21st chromosome in the nucleus of each cell.
- Occurs more often with older mothers or moms who have a Down's syndrome baby.
- Common characteristics include:
  - ✓ Mental retardation
  - ✓ Heart disease, hearing problem
  - ✓ Typified physical appearance
  - ✓ Thyroid issues and digestive problems

Genetic Disorders: Turner Syndrome

- Individuals all or part of one X (female) chromosome.
- Characteristics associated with this disorder include:



Karyotype of Individual with Turner Syndrome