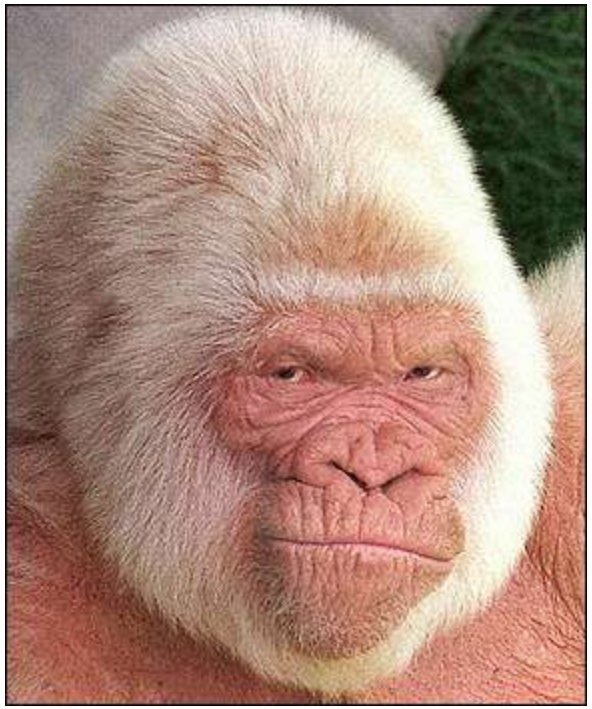
NOTES: 13.3 - MUTATIONS

MUTATIONS:

- **MUTATIONS** = changes in the DNA sequence that affect genetic information

MUTATIONS:

**any change in the DNA sequence can also change the protein it codes for



Mutations in Reproductive Cells:

- if a mutation occurs in a gene in a sperm or egg cell, the altered gene would become part of the genetic makeup of the offspring
- the result could be:

➔ a new trait (beneficial or harmful);

➔ a protein that does not work correctly;

➔ miscarriage

Mutations in Body Cells:

- if a mutation occurs in a nonreproductive cell (such as skin or muscle cell), it will not be passed to offspring
- the result could be:

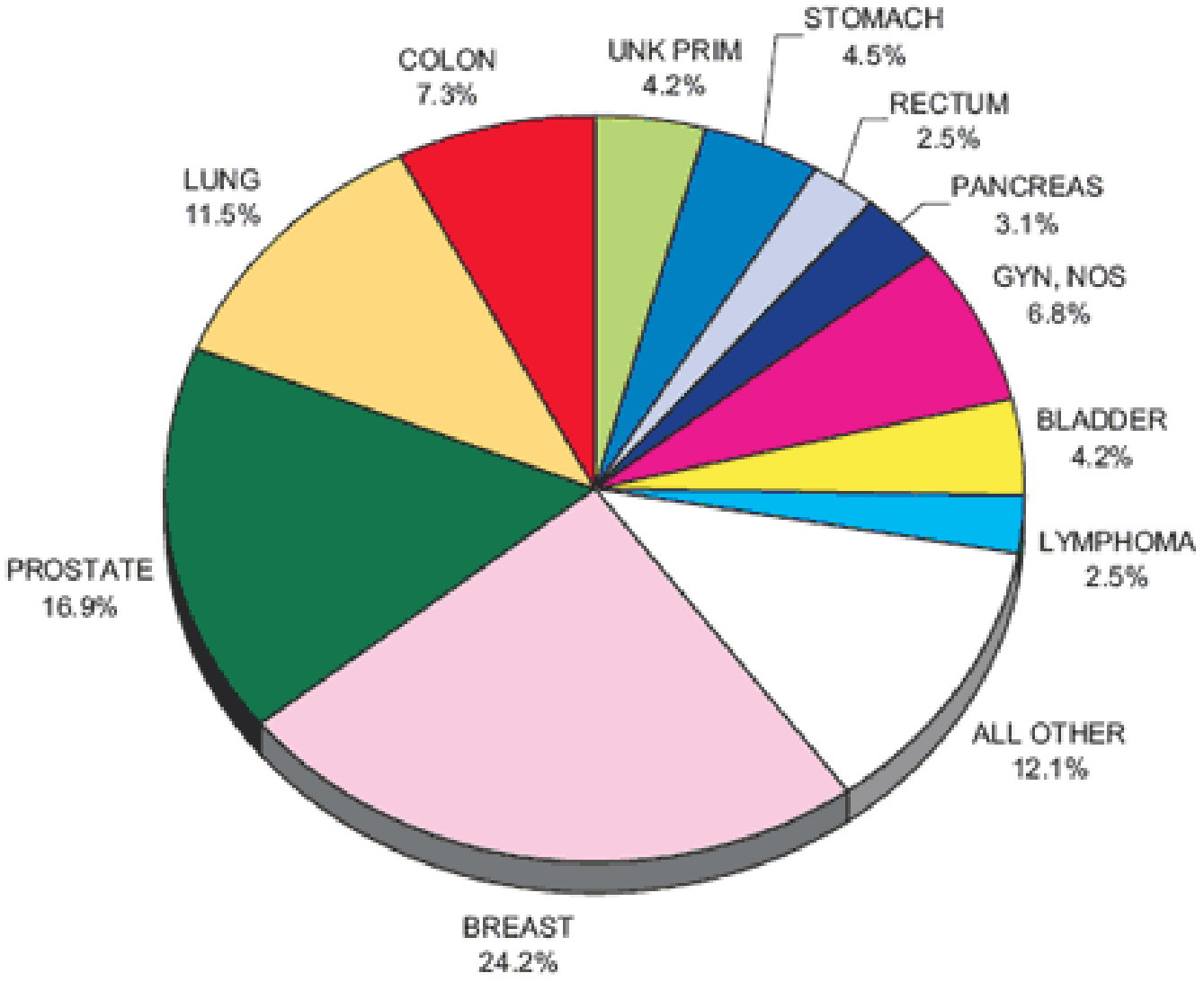
➔ impaired functioning of the cell;

➔ loss of control of cell division;

➔ cancer.

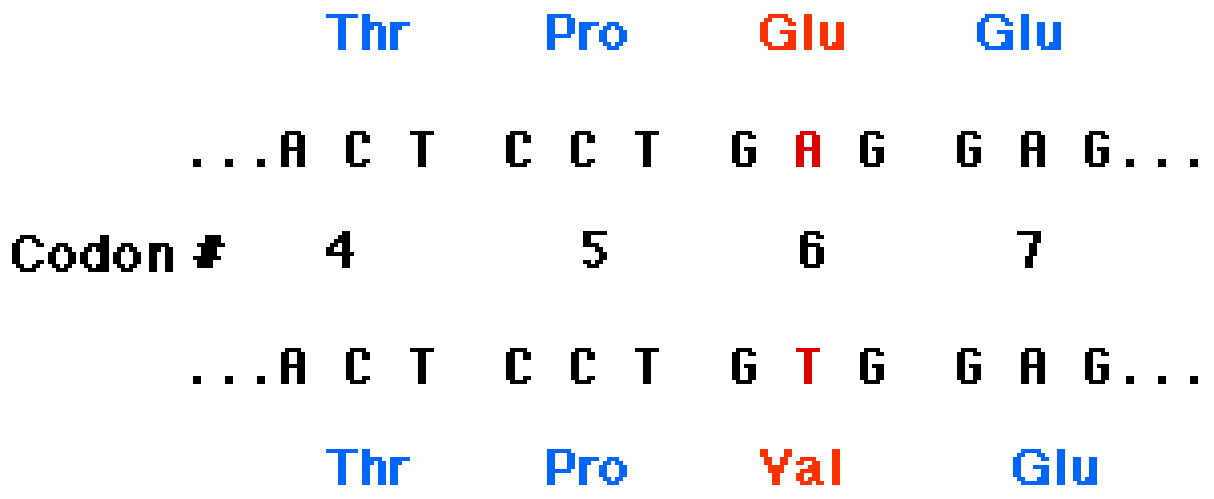
Frequency of Cancer

2005 Distribution of Primary Diagnosis



Types of GENE MUTATIONS:

- **Point Mutation**: *a change in a **single base pair in DNA***



3 types of POINT MUTATIONS:

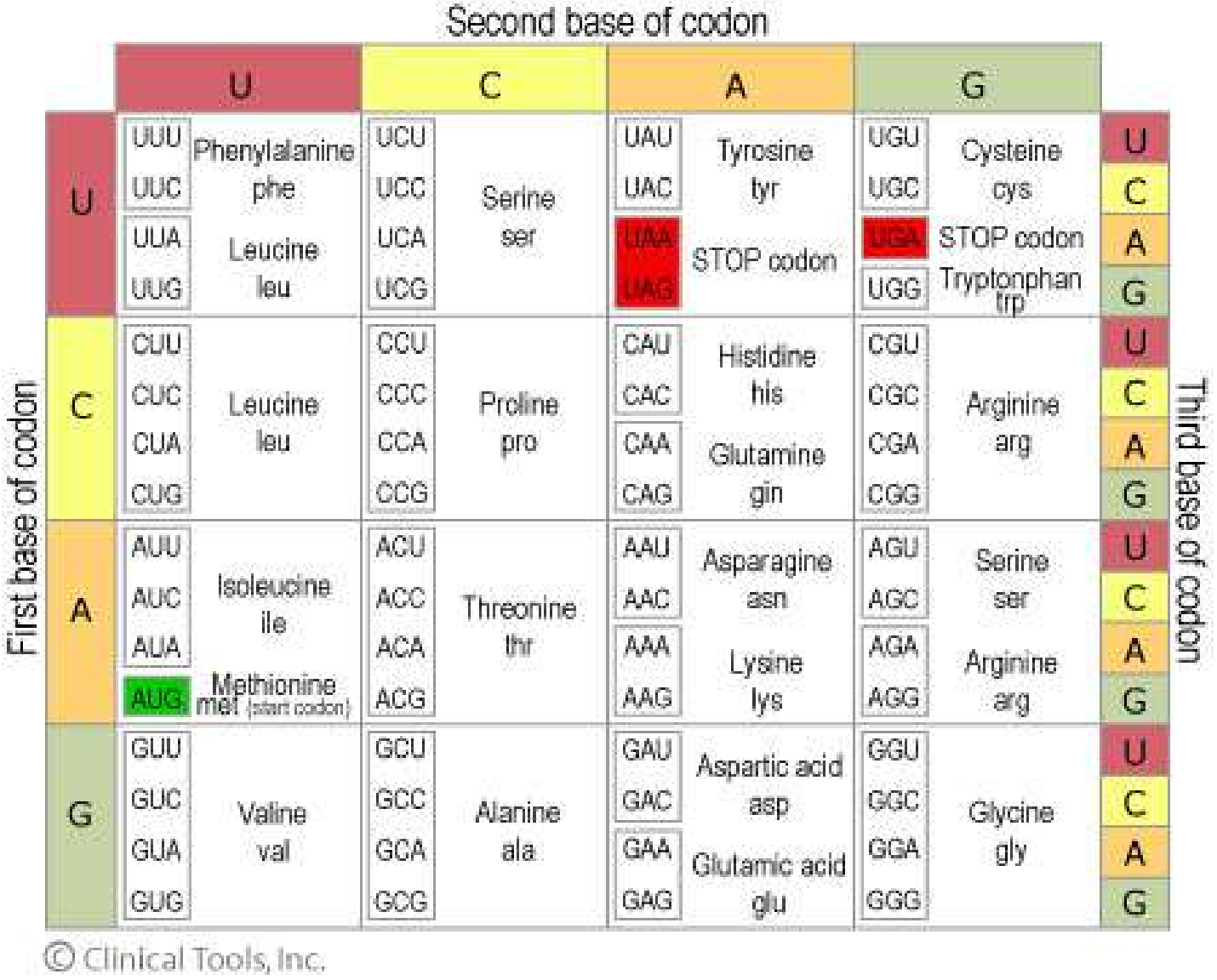
1) SUBSTITUTION

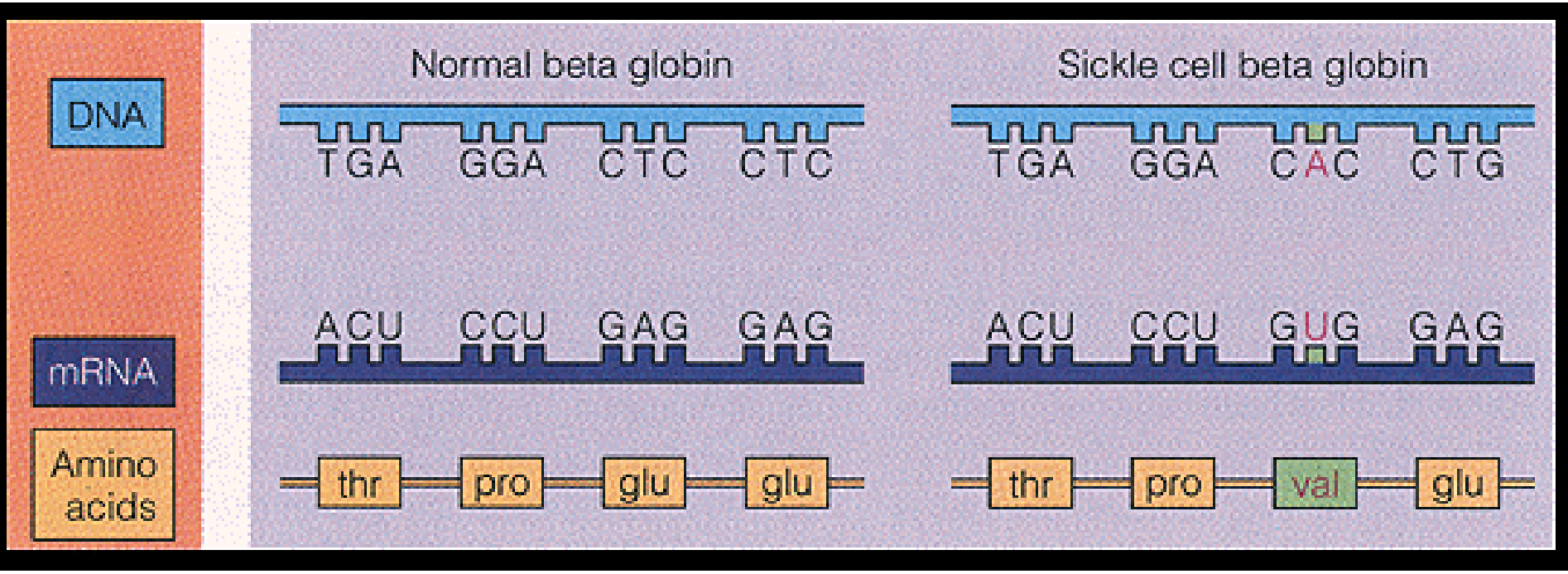
-One base pair is replaced by another base pair

-***Might*** result in the wrong amino acid *(why only "might"?)*

- Redundancy of the genetic code!

Redundancy of the code:

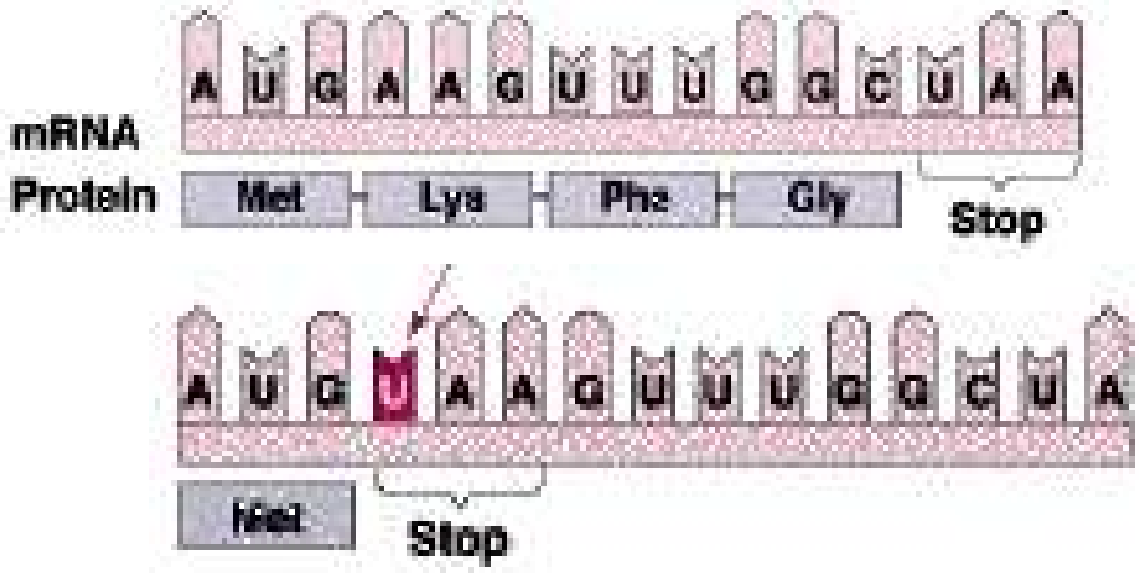



DNA
mRNA
Amino acids
Normal beta globin
TGA GGA CTC CTC
ACU CCU GAG GAG
thr pro glu glu
Sickle cell beta globin
TGA GGA CAC CTG
ACU CCU GUG GAG
thr pro val glu

3 types of POINT MUTATIONS:

2) BASE PAIR INSERTION

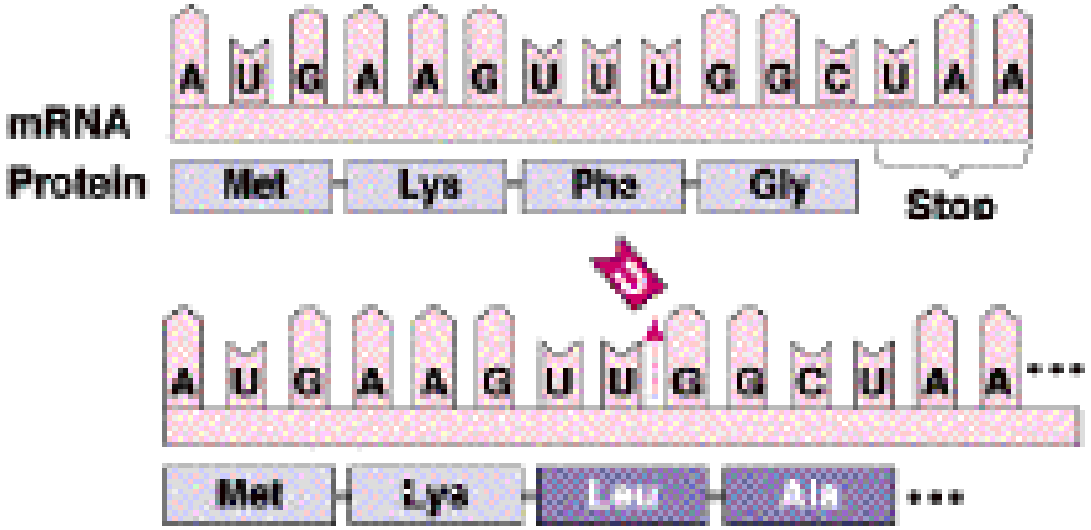
= insertion of 1 or more nucleotide pairs into a gene



3 types of POINT MUTATIONS:

3) BASE PAIR DELETION

= deletion of 1 or more nucleotide pairs from the gene



Frameshift mutations:

- Both base pair deletions and base pair insertions can result in a shift in the reading frame
- That can cause the wrong protein to be made!

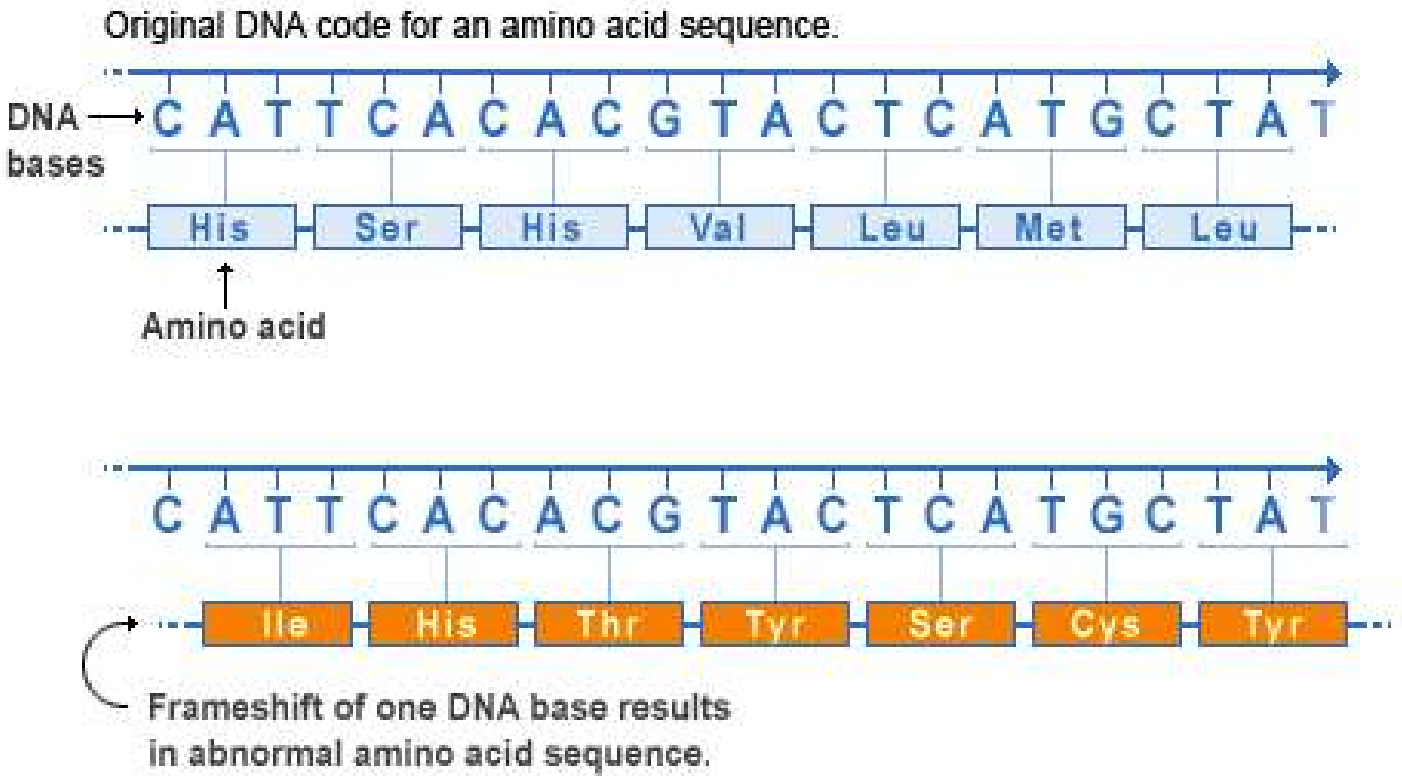
- ex: THE **C**AT ATE THE RAT

what happens

If we delete **"C"** ➔ **THE ATA TET HER AT-…**

Frameshift mutations:

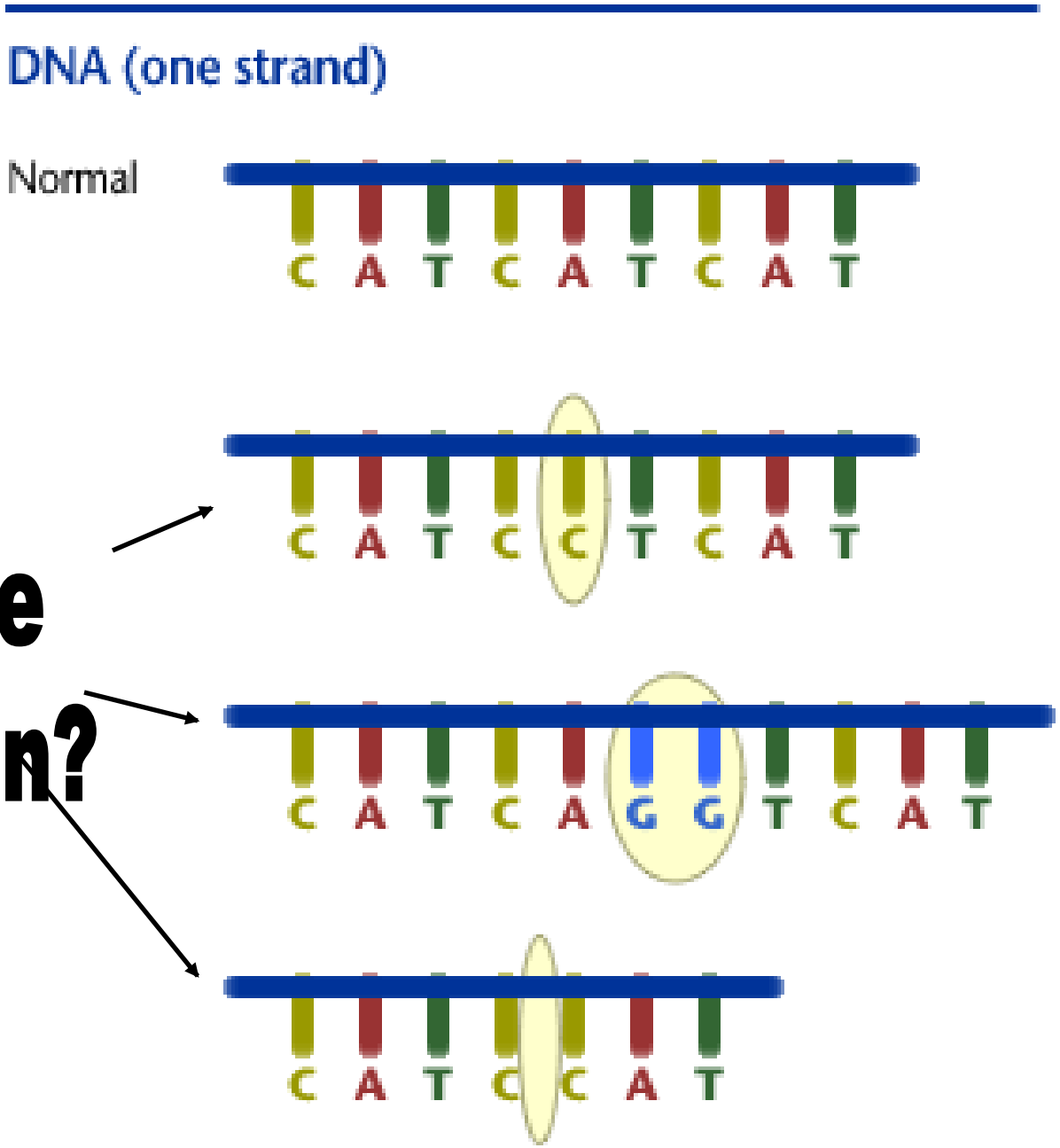
- nearly every amino acid in the protein after the mutation is changed!



SUMMARY: Types of Mutations

- Gene mutations
  - Base pair substitution
  - Base pair insertion
  - Base pair deletion

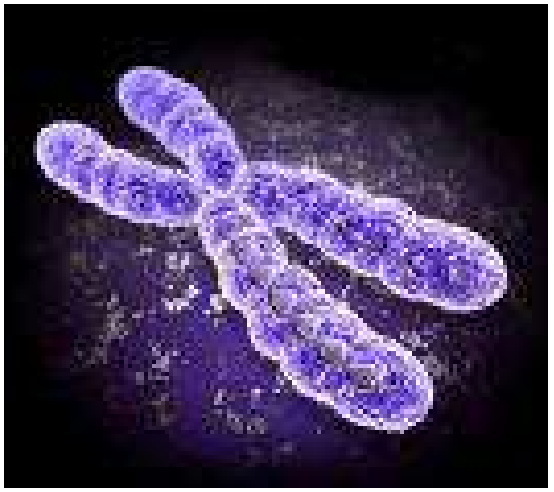
FRAMESHIFT MUTATIONS



What type of mutation?

Chromosomal Mutations

- **Chromosomal Mutations:**
  - **Deletion**
  - **Duplication**
  - **Inversion**
  - **Translocation**



Chromosomal Mutations:

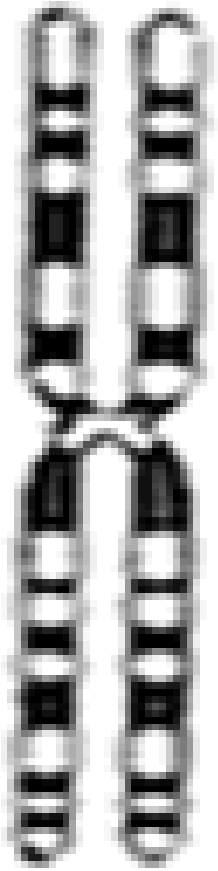
- **DELETION**: **lose all or part of a chromosome**

ABC-DEFAC-DEF →

- **DUPLICATION**: **segment of a chromosome is repeated**

ABC-DEFABBC-DEF →

Deletion Mutation

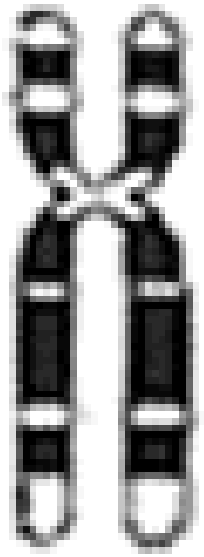


Chromosomal Mutations:

- **INVERSION: chromosome segment becomes oriented in reverse direction**

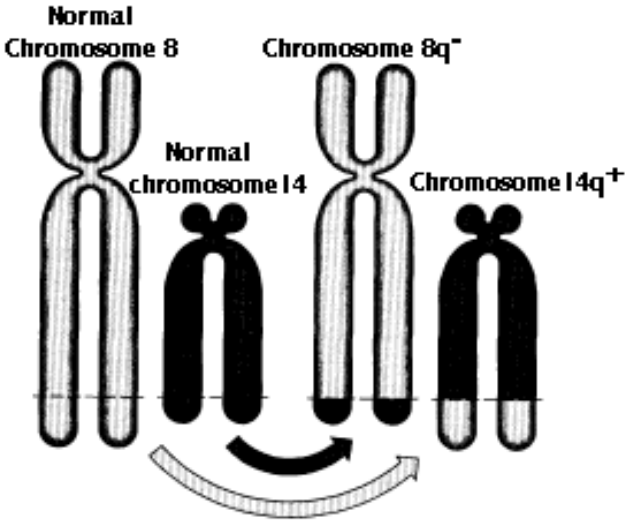
ABC-DEF ABE-DCF

Inversion Mutation



Chromosomal Mutations:

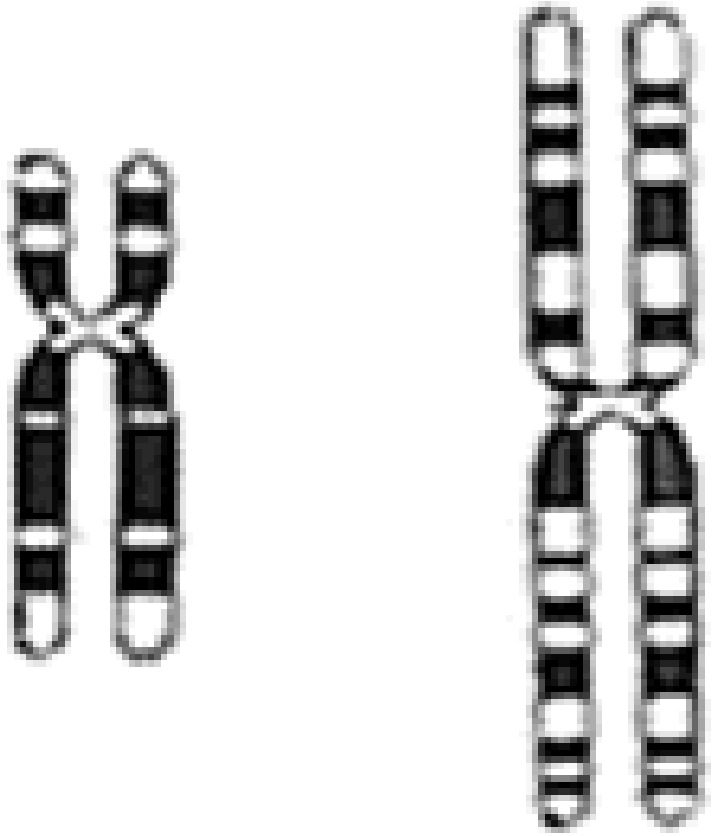
- **TRANSLOCATION: part of 1 chromosome breaks off & attaches to another non-homologous chromosome (segment is usually exchanged)**



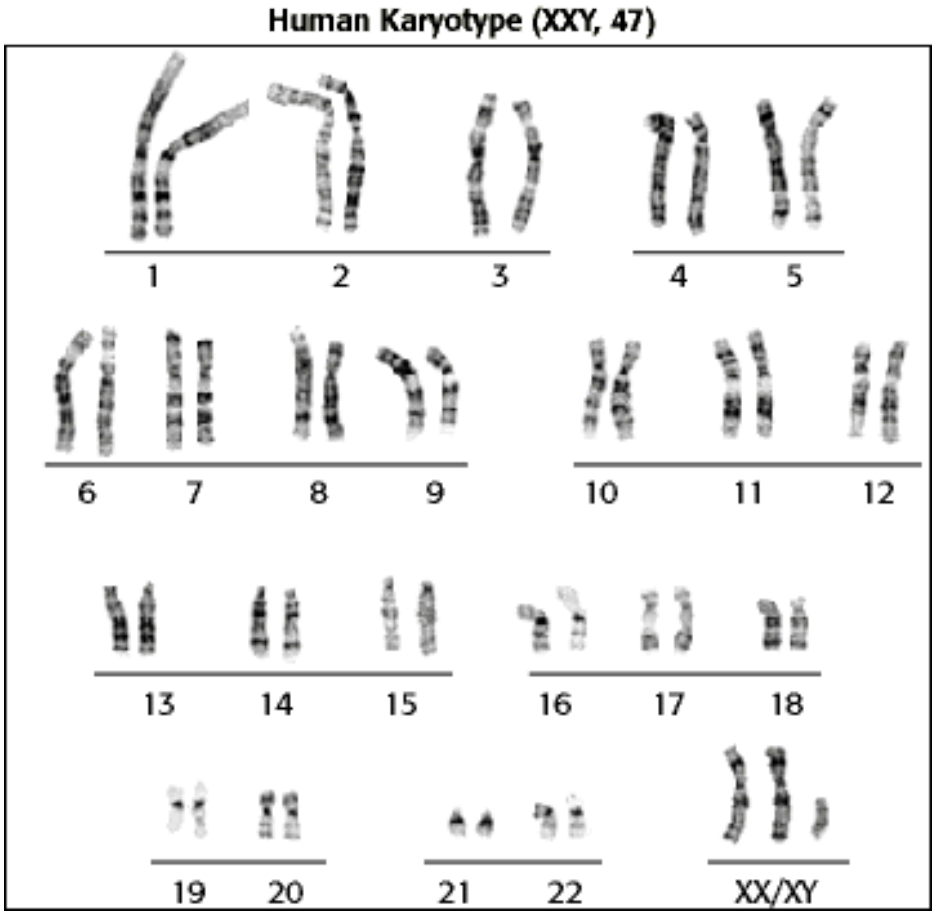
ABC-DEF ⟶ ABC-JKL

GH-IJKL ⟶ GH-IDEF

Translocation



Chromosomal mutations could also involve having the incorrect # of chromosomes…



-ex: 47 human chromosomes instead of 46

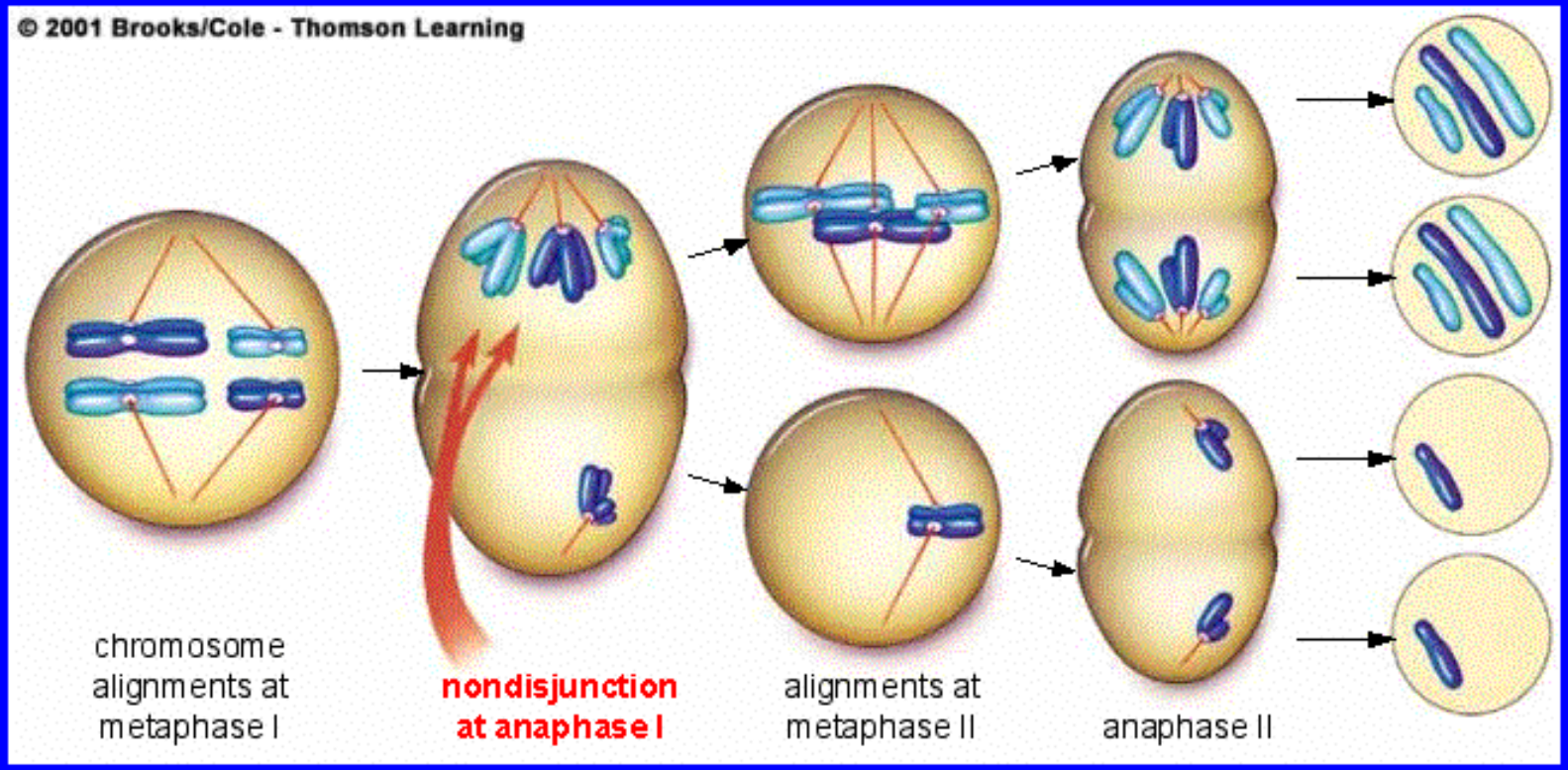
-we will discuss this in chapter 12

Chromosomal Mutations (change in number)

- **NONDISJUNCTION**: the failure of the chromosomes to separate properly during cell division

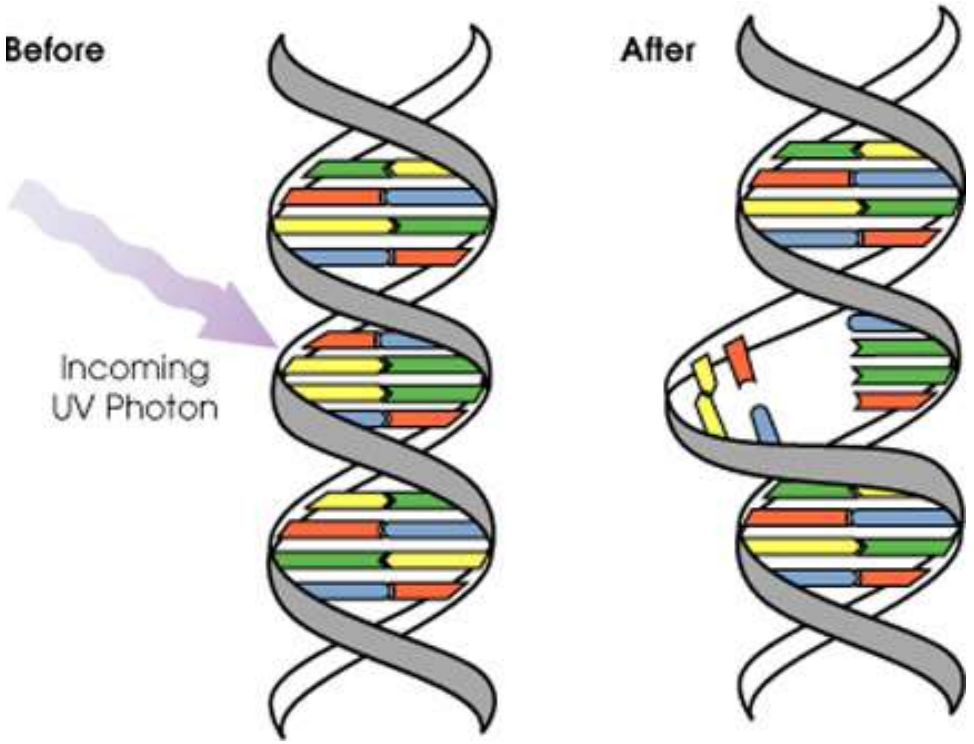
(specifically, MEIOSIS, the type of cell division that produces the gametes)

Nondisjunction



Causes of Mutations:

- **MUTAGEN**: any agent that can cause a change in DNA



MUTAGENS:

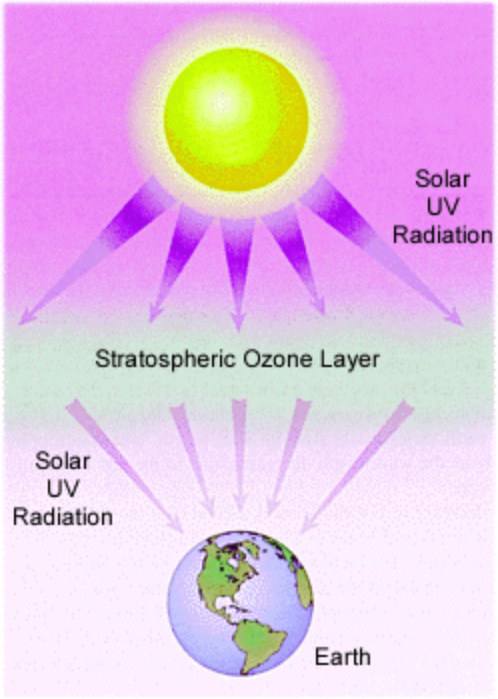
1) HIGH ENERGY RADIATION:

> X-rays

> cosmic rays

> ultraviolet light

> nuclear radiation



Asymmetry

Border irregularity

Color

Diameter: ¼ inch or 6mm

MUTAGENS:

2) CHEMICALS:

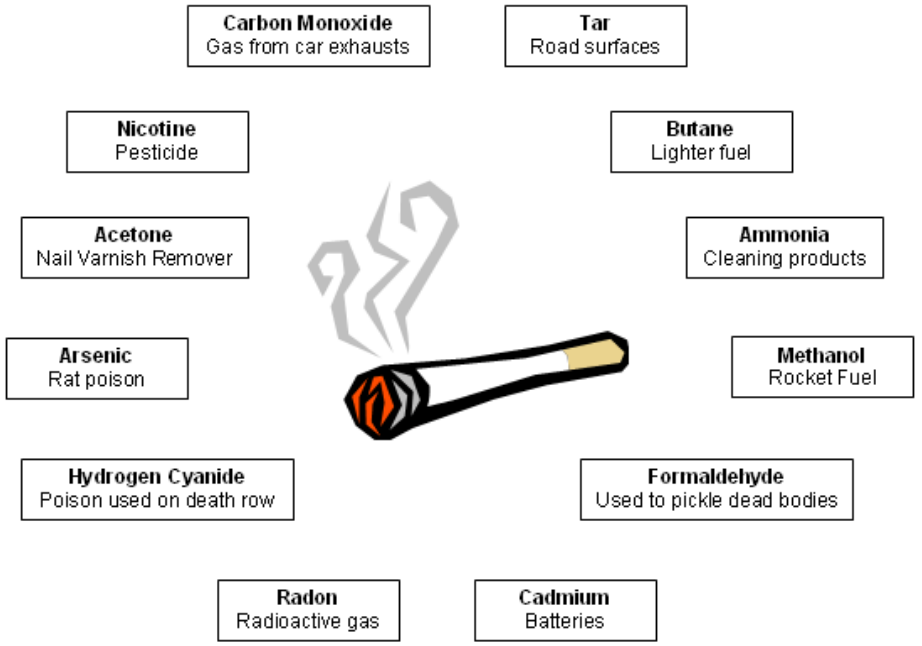
> dioxins

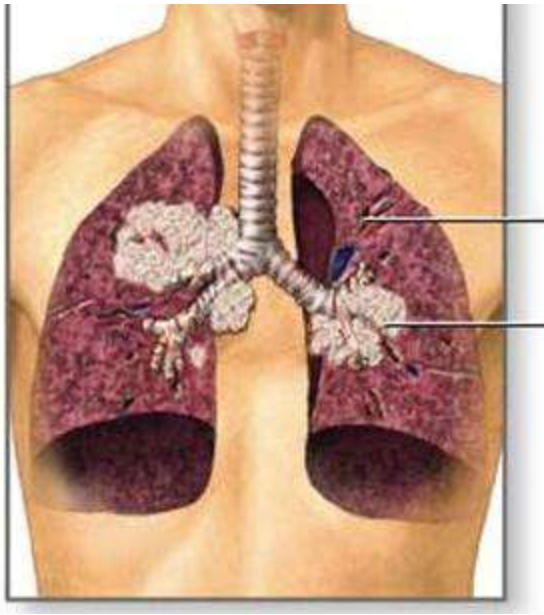
> asbestos

> benzene

> formaldehyde

> cigarette smoke





Lungs of a smoker("brown")

Cancer