

NOTES: 13.3 - MUTATIONS



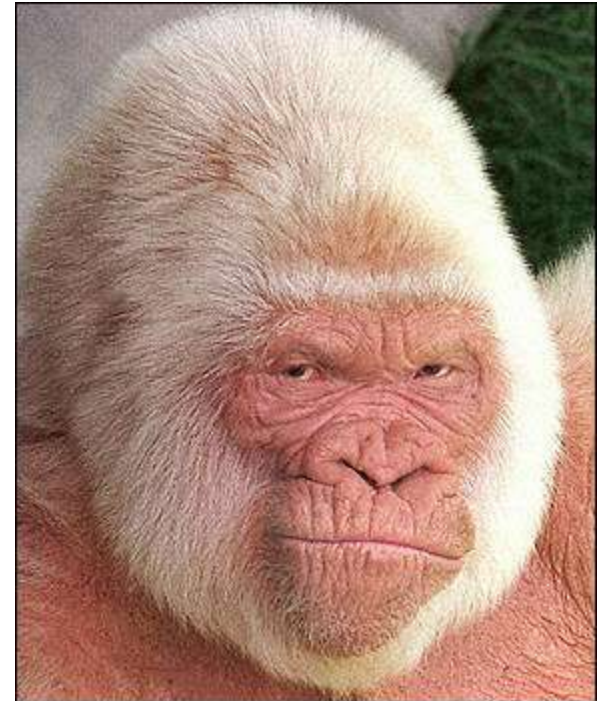
MUTATIONS:

- 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:

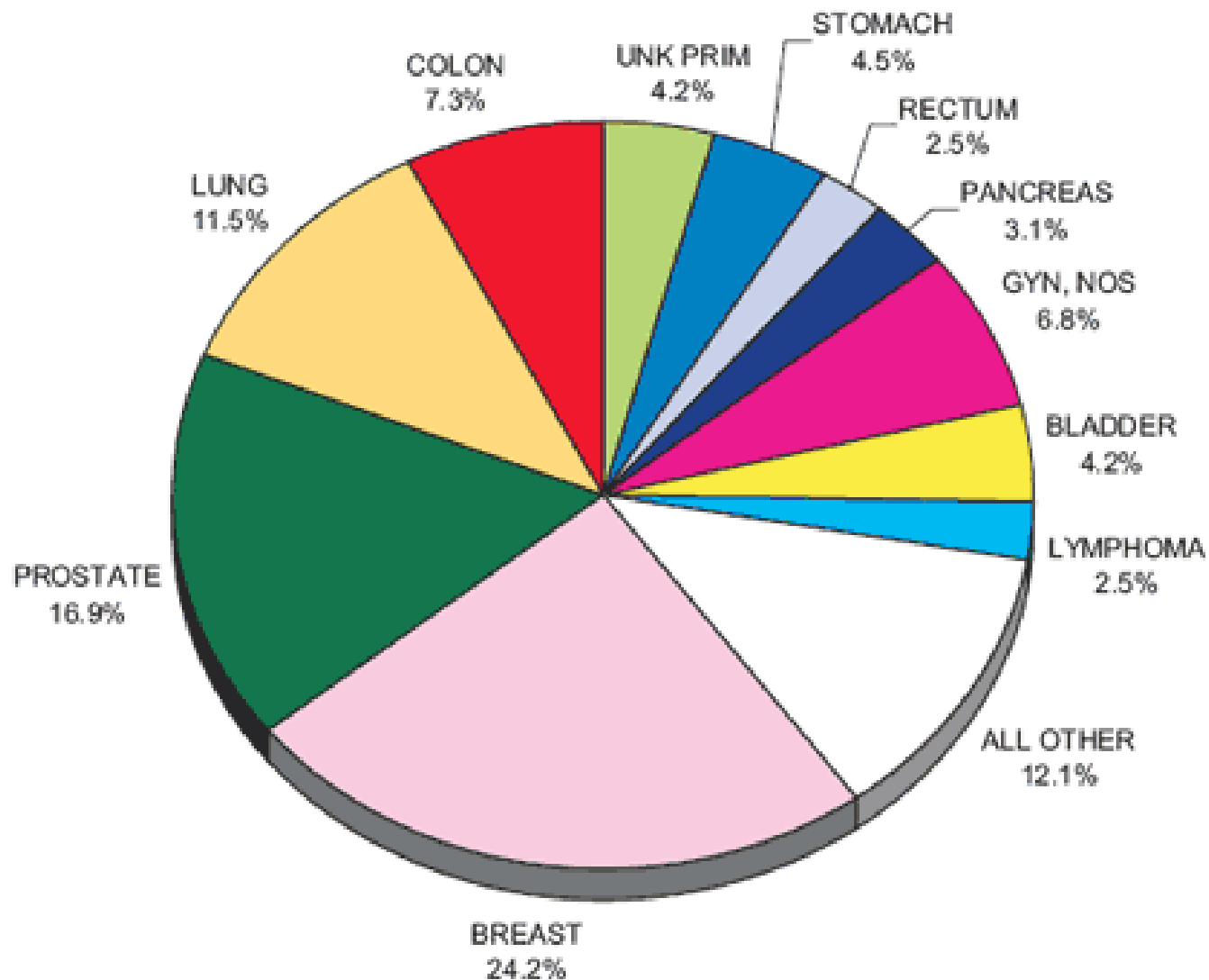
- if a mutation occurs in a gene in a sperm or egg cell, the altered gene would become part of the genetic makeup of the offspring
- the result could be:
 - ➔ a new trait (beneficial or harmful);
 - ➔ a protein that does not work correctly;
 - ➔ miscarriage

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*

	Thr	Pro	Glu	Glu
	... A C T	C C T	G A G	G A G ...
Codon #	4	5	6	7
	... A C T	C C T	G T G	G A G ...
	Thr	Pro	Val	Glu

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!

CTGGAG

CTGGGG

Redundancy of the code:

		Second base of codon											
		U		C		A		G					
First base of codon	U	UUU	Phenylalanine phe	UCU	Serine ser	UAU	Tyrosine tyr	UGU	Cysteine cys	U			
		UUC		UCC		UAC		UGC			C		
		UUA	Leucine leu	UCA		STOP codon	UGA	STOP codon	A				
		UUG		UCG			UAG					UGG	Tryptophan trp
	C	CUU	Leucine leu	CCU	Proline pro	CAU	Histidine his	CGU		Arginine arg		U	
		CUC		CCC		CAC		CGC			C		
		CUA		CCA		CAA	Glutamine gin	CGA	A				
		CUG		CCG		CAG		CGG					G
	A	AUU	Isoleucine ile	ACU	Threonine thr	AAU	Asparagine asn	AGU		Serine ser		U	
		AUC		ACC		AAC		AGC			C		
		AUA		ACA		AAA	Lysine lys	AGA	Arginine arg	A			
		AUG	Methionine met (start codon)	ACG		AAG		AGG					G
	G	GUU	Valine val	GCU	Alanine ala	GAU	Aspartic acid asp	GGU	Glycine gly			U	
		GUC		GCC		GAC		GGC			C		
		GUA		GCA		GAA	Glutamic acid glu	GGA		A			
		GUG		GCG		GAG		GGG					G

DNA

mRNA

Amino
acids

Normal beta globin



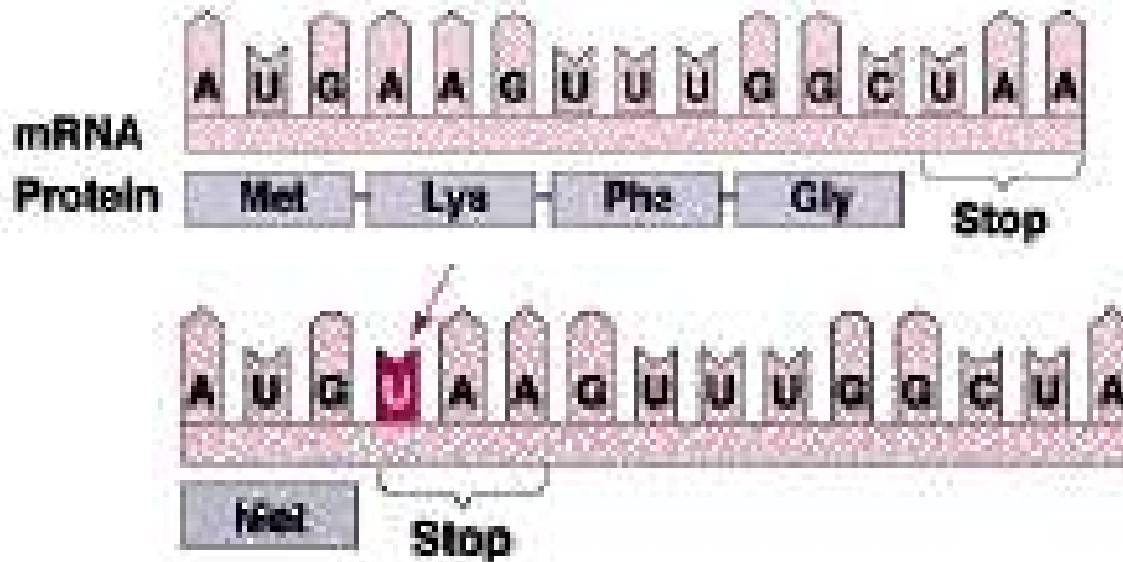
Sickle cell beta globin



3 types of POINT MUTATIONS:

2) BASE PAIR INSERTION

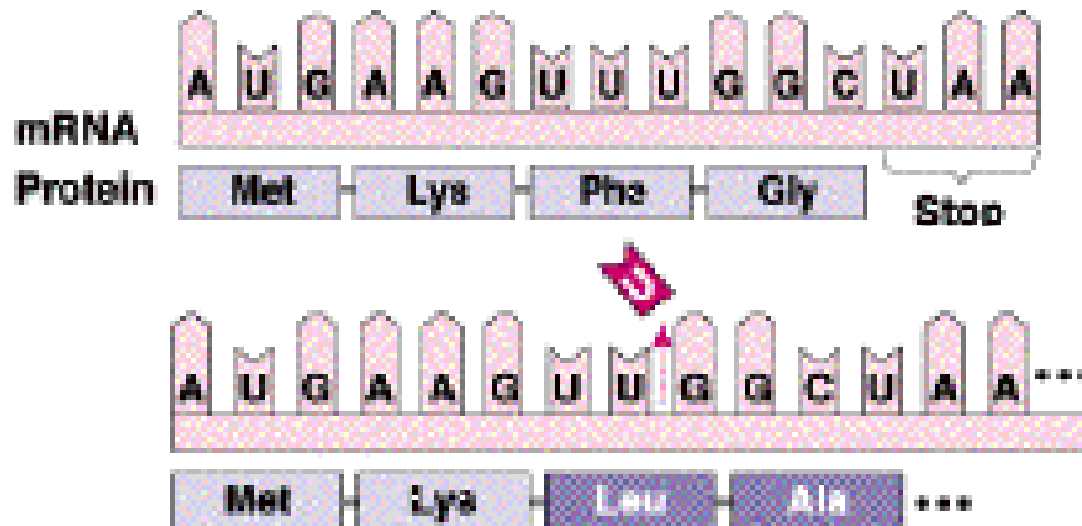
= insertion of 1 or more
nucleotide pairs into a gene



3 types of POINT MUTATIONS:

3) BASE PAIR DELETION

= deletion of 1 or more nucleotide pairs from the gene

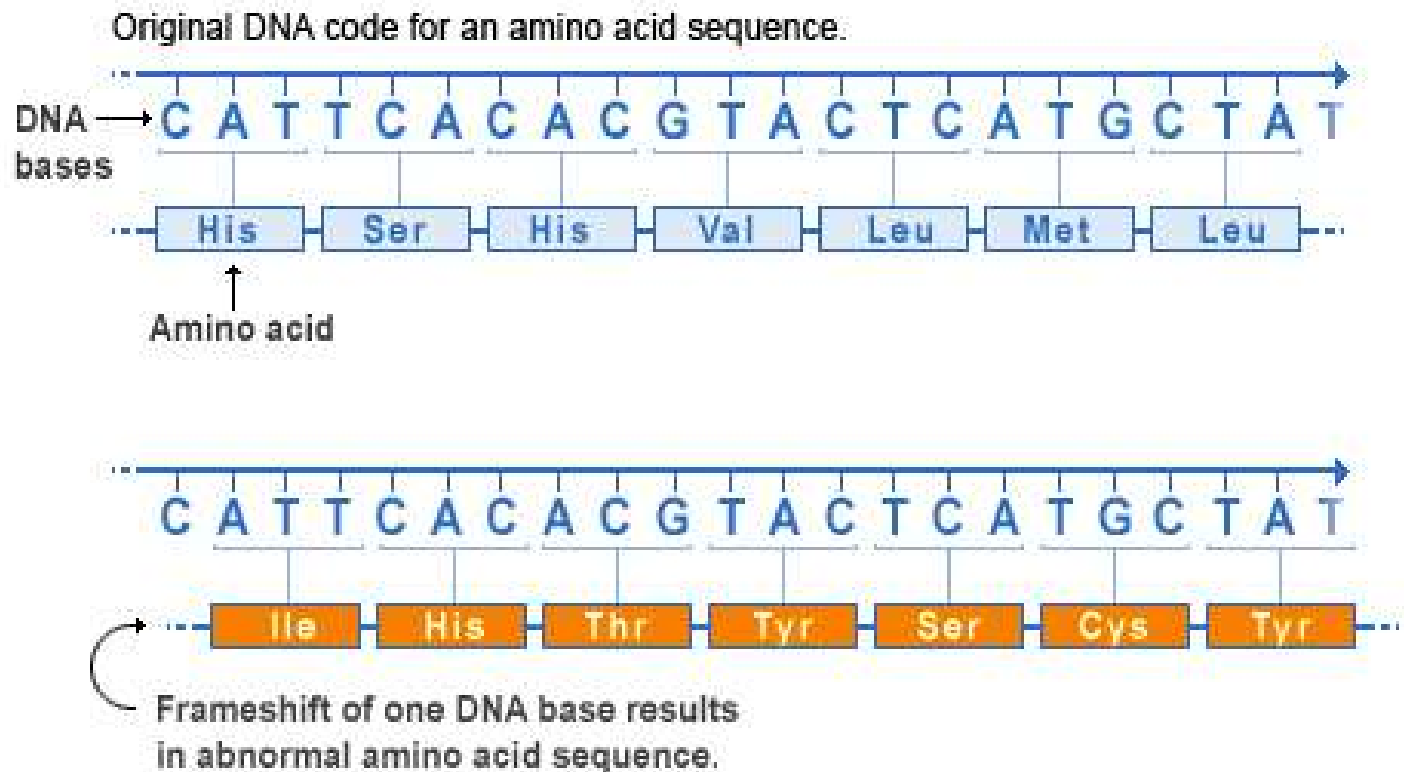


Frameshift mutations:

- 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 CAT ATE THE RAT
what happens
If we delete "C" → THE ATA TET HER AT-...

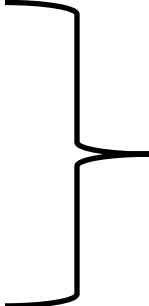
Frameshift mutations:

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



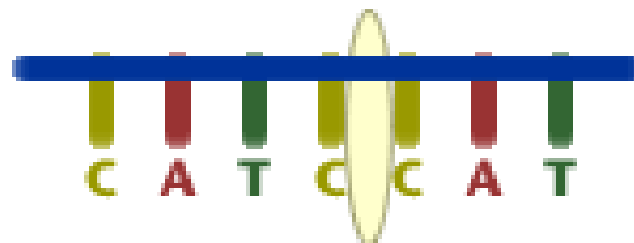
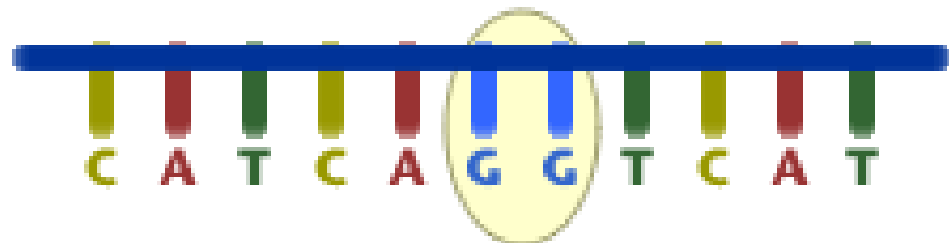
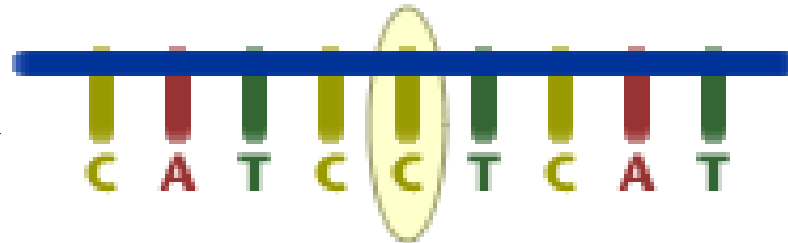
SUMMARY:

Types of Mutations

- Gene mutations
 - Base pair substitution
 - Base pair insertion
 - Base pair deletion
- FRAMESHIFT
MUTATIONS**
- 

DNA (one strand)

Normal

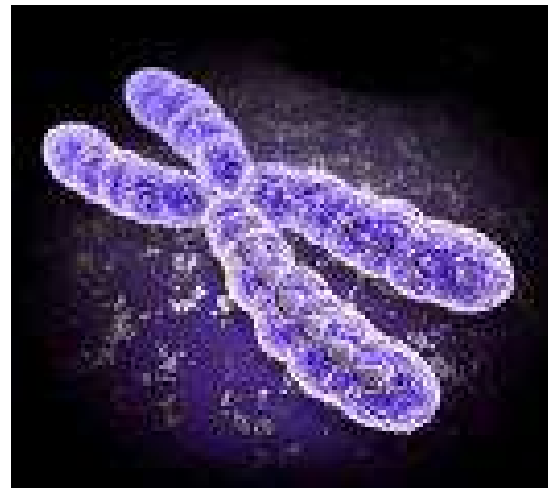


**What type
of mutation?**

Chromosomal Mutations

- **Chromosomal Mutations:**

- Deletion
- Duplication
- Inversion
- Translocation



Chromosomal Mutations:

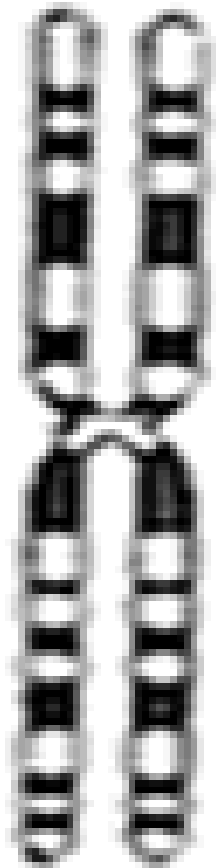
- DELETION: lose all or part of a chromosome

ABC-DEFAC-DEF →

- DUPLICATION: segment of a chromosome is repeated

ABC-DEFABBC-DEF →

Deletion Mutation

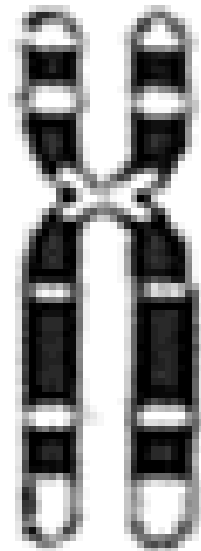


Chromosomal Mutations:

- INVERSION: chromosome segment becomes oriented in reverse direction

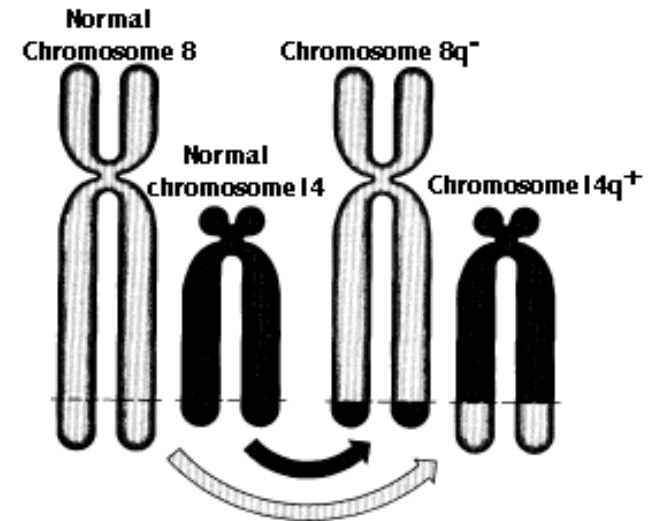
ABC-DEF ~~ABE-DCF~~

Inversion Mutation



Chromosomal Mutations:

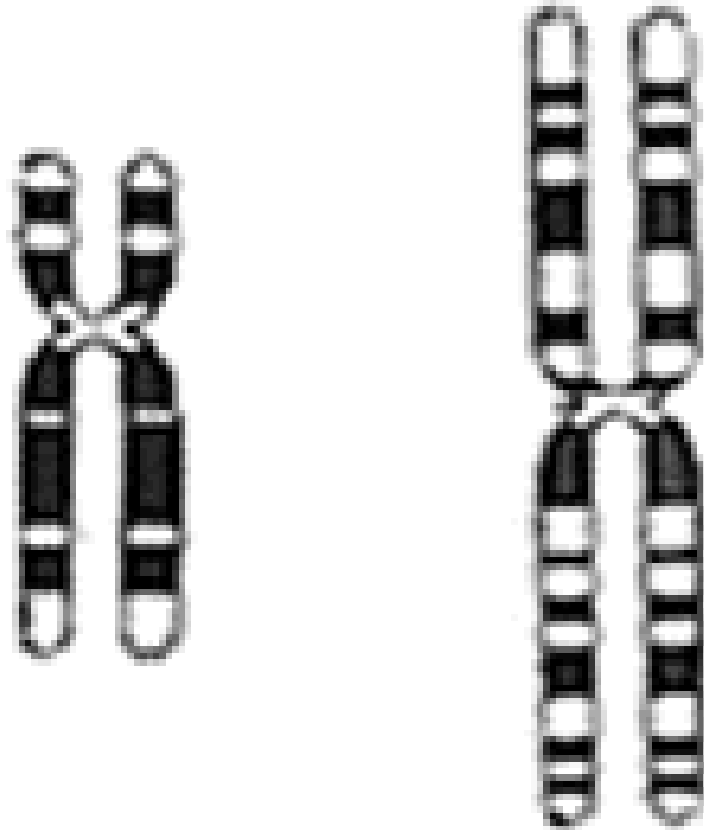
- TRANSLOCATION:
part of 1
chromosome breaks
off & attaches to
another non-
homologous
chromosome
(segment is usually
exchanged)



ABC-DEF \longrightarrow ABC-JKL

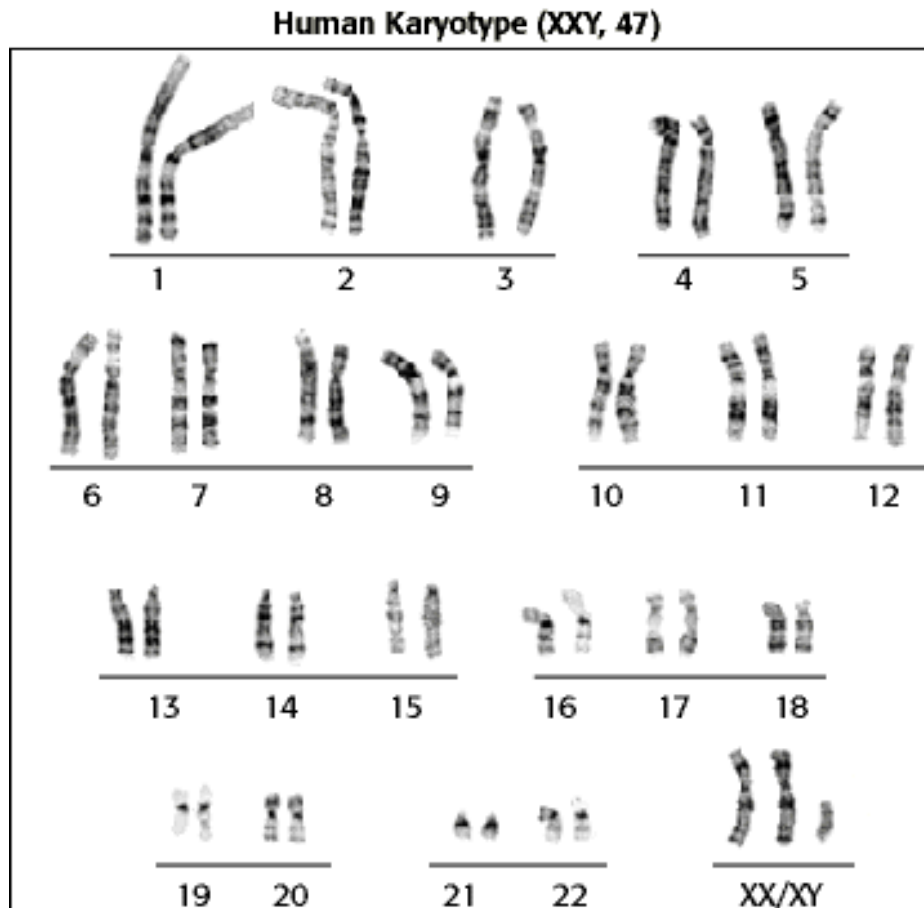
GH-IJKL \longrightarrow GH-IDEF

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



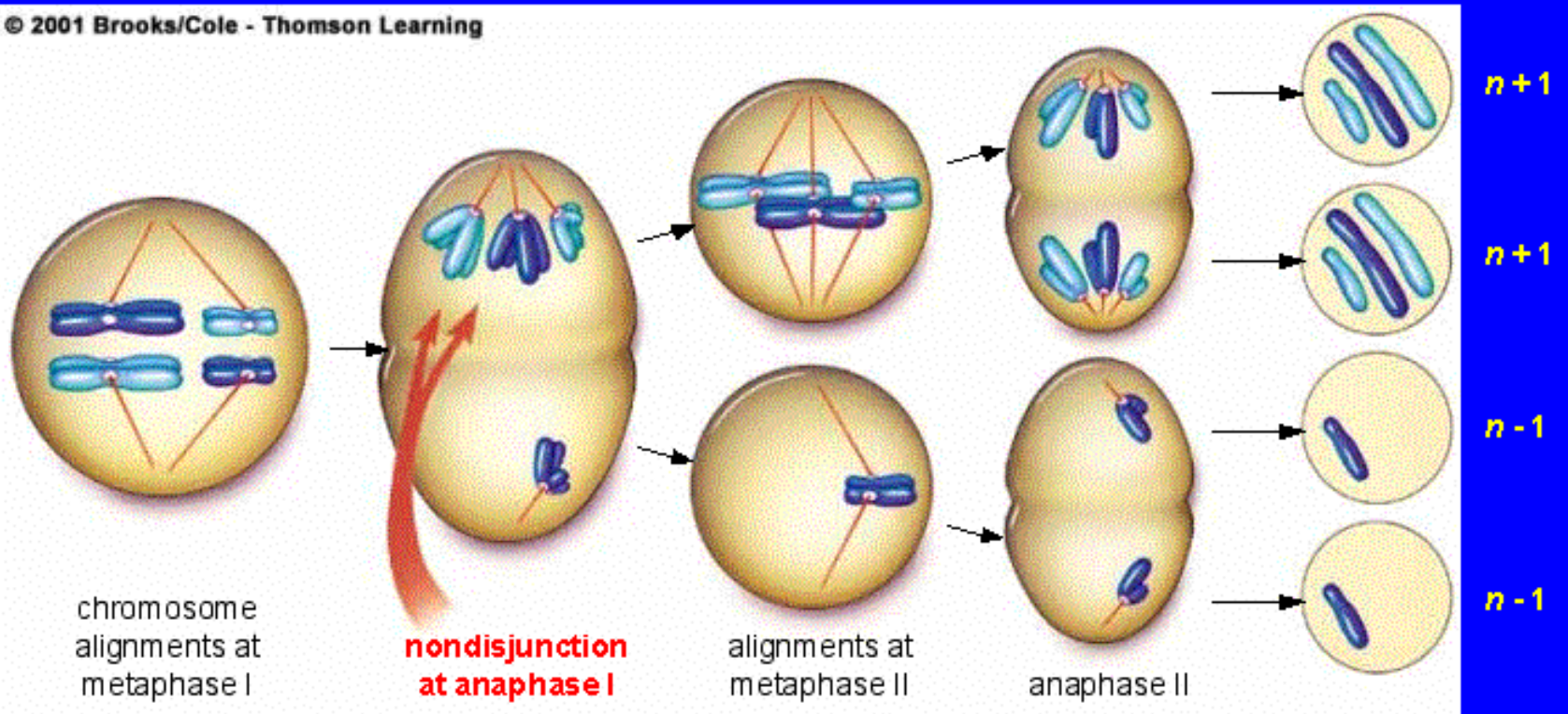
Chromosomal Mutations (change in number)

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

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

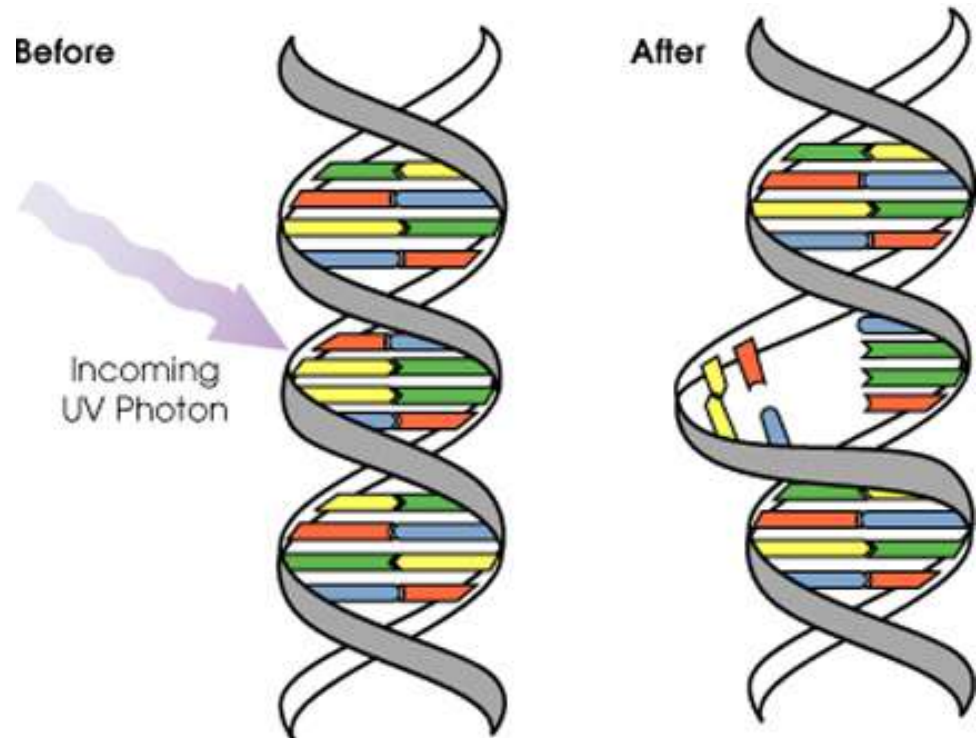
Nondisjunction

© 2001 Brooks/Cole - Thomson Learning



Causes of Mutations:

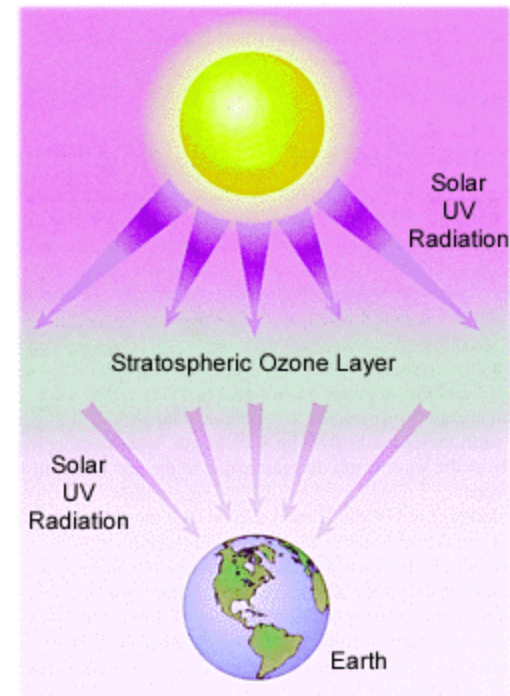
- **MUTAGEN**: any agent that can cause a change in DNA



MUTAGENS:

1) HIGH ENERGY RADIATION:

- > X-rays
- > cosmic rays
- > ultraviolet light
- > nuclear radiation





Asymmetry



Border
irregularity



Color

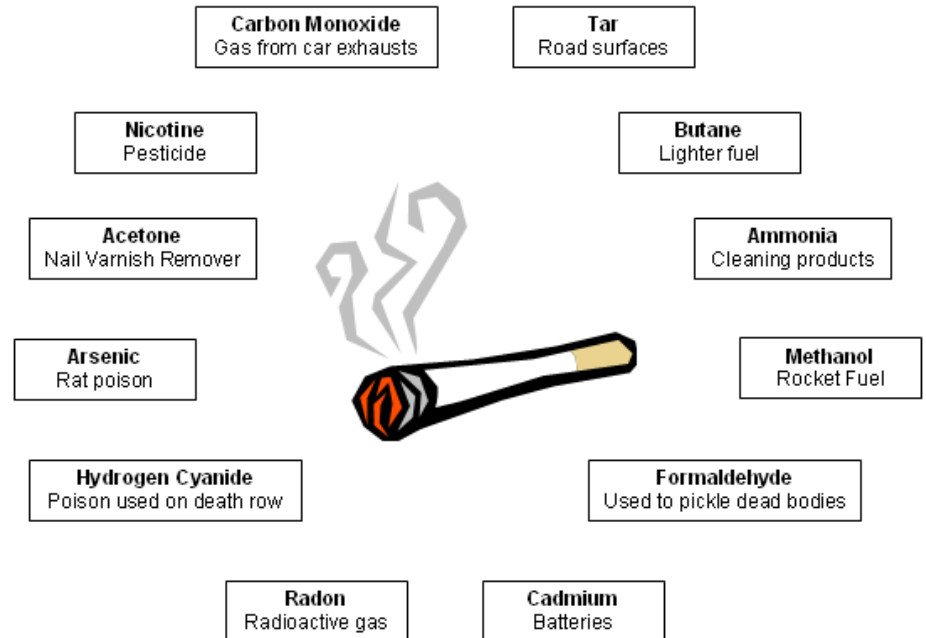


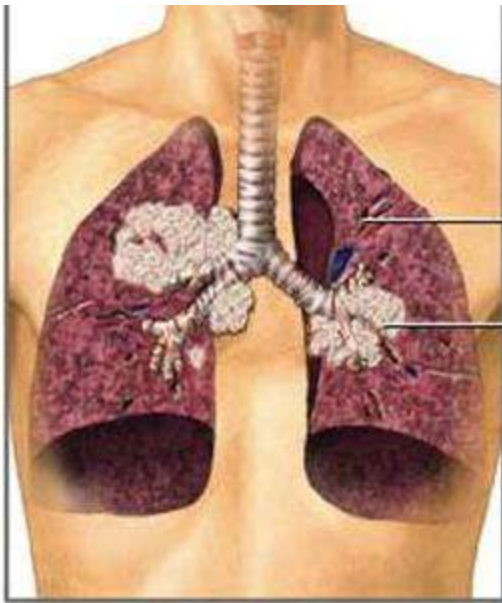
Diameter:
 $\frac{1}{4}$ inch or
6mm

MUTAGENS:

2) CHEMICALS:

- > dioxins
- > asbestos
- > benzene
- > formaldehyde
- > cigarette smoke





Lungs of a
smoker("brown")

Cancer

