Mutations

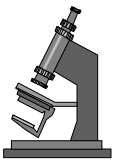


Fig. 18.1

Cancer Cells

Fig. 18.2a

Mutation in *Drosophila*

Fig. 18.2b

Types of Mutation

Mutation	Example result
NO MUTATION	Normal B protein is produced by the *B* gene.
POINT MUTATION	
Base substitution — Substitution of one or a few bases	B protein is inactive because changed amino acid disrupts function.
Insertion — Addition of one or a few bases	B protein is inactive because inserted material disrupts proper shape.
Deletion — Loss of one or a few bases	B protein is inactive because portion of protein is missing.
CHANGES IN GENE POSITION	
Transposition	*B* gene or B protein may be regulated differently because of change in gene position.
Chromosomal rearrangement	*B* gene may be inactivated or regulated differently in its new location on chromosome.

Fig. 18.3

Pyrimidine Dimer

Fig. 18.4

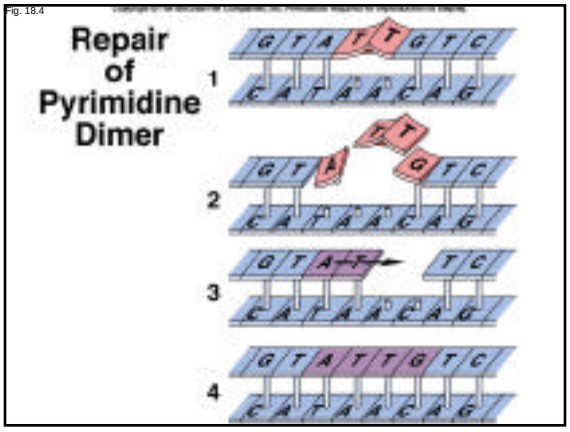


Fig. 18.5

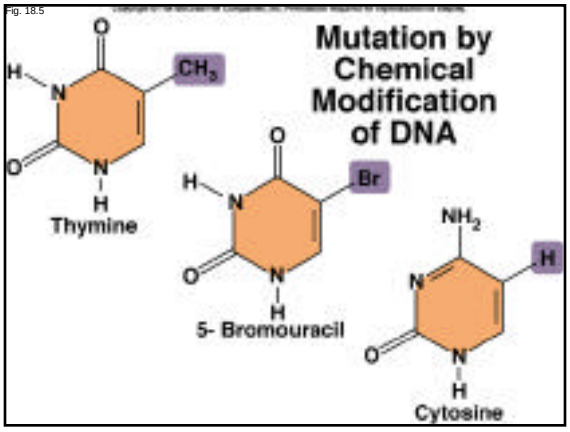


Fig. 18.6

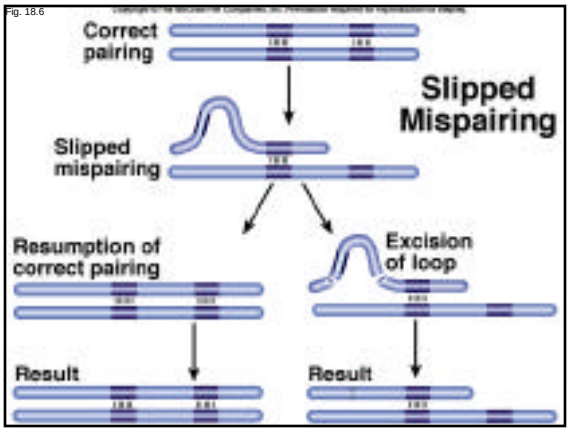
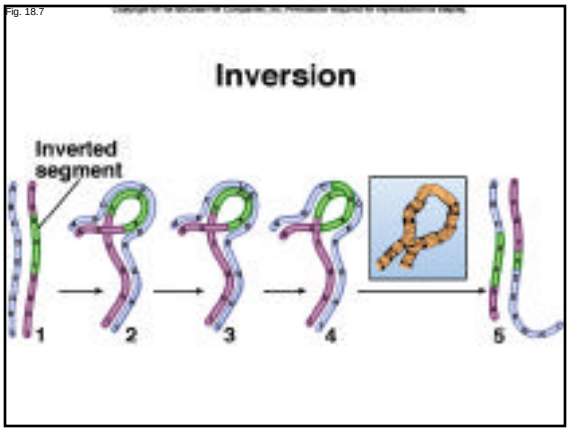


Fig. 18.7



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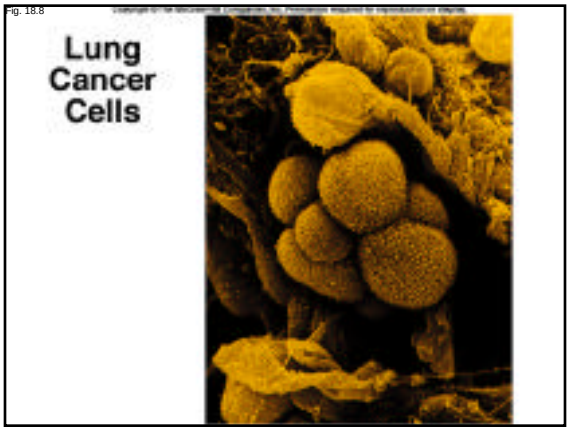


Fig. 18.9

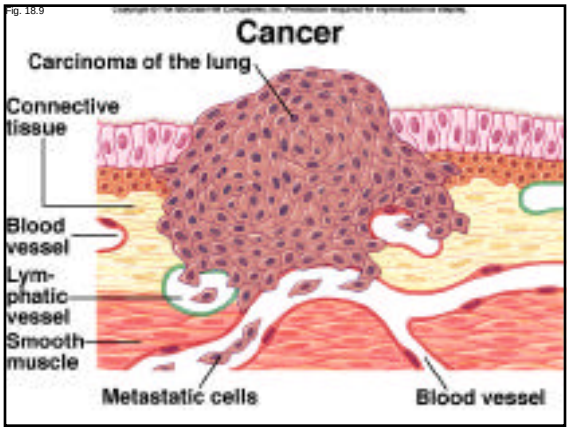


Table 18.2 Incidence of Cancer in the United States in 2000

Type of Cancer	New Cases	Deaths	% of Cancer Deaths
Lung	164,100	156,900	28
Colon and rectum	93,700	56,300	10
Leukemia/lymphoma	95,200	49,200	9
Breast	184,300	41,200	8
Prostate	180,400	31,900	7
Pancreas	28,300	28,200	5
Ovary	23,100	14,000	3
Stomach	21,500	13,800	2
Liver	15,300	13,800	2
Nervous system/eye	18,700	13,200	2
Bladder	35,300	12,200	2
Oral cavity	30,200	7,800	2
Kidney	31,200	11,900	2
Cervix/uterus	46,900	11,300	2
Malignant melanoma	47,700	7,700	1
Sarcoma (connective tissue)	10,600	6,800	1
All other cancers	199,400	77,800	14

In the United States in 2000 there were 1,220,100 reported cases of new cancers and 552,200 cancer deaths, indicating that roughly half the people who develop cancer die from it.

Source: Data from the American Cancer Society, Inc., 2000.

Fig. 18.10

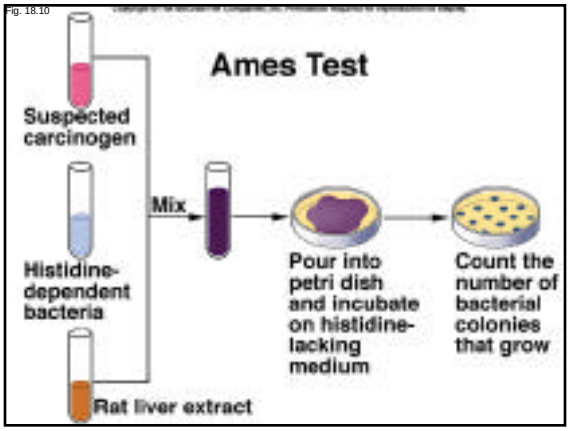


Fig. 18.11a

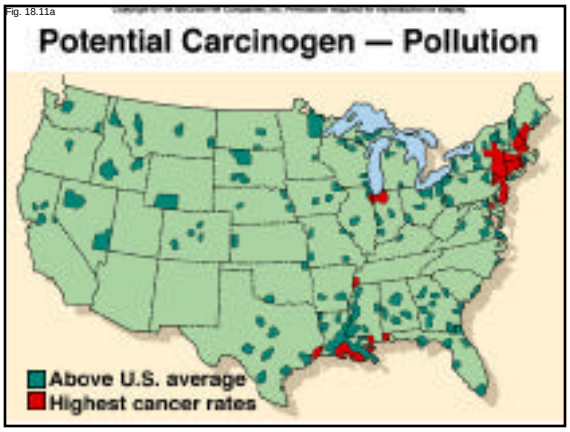


Fig. 18.11b

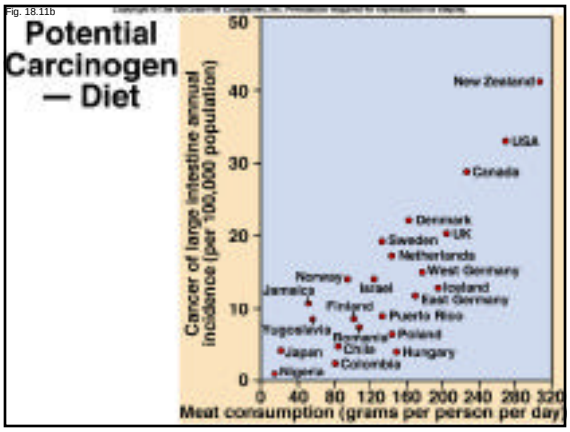
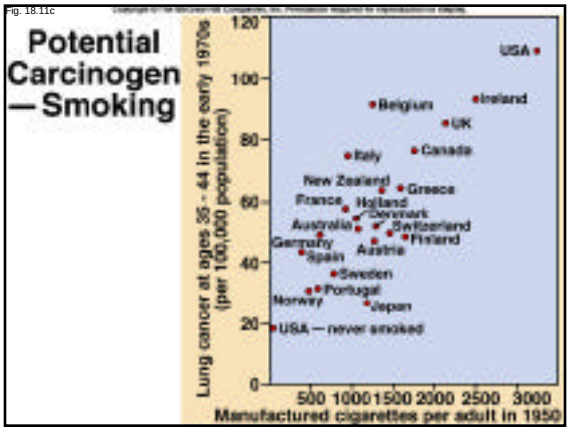


Fig. 18.11c



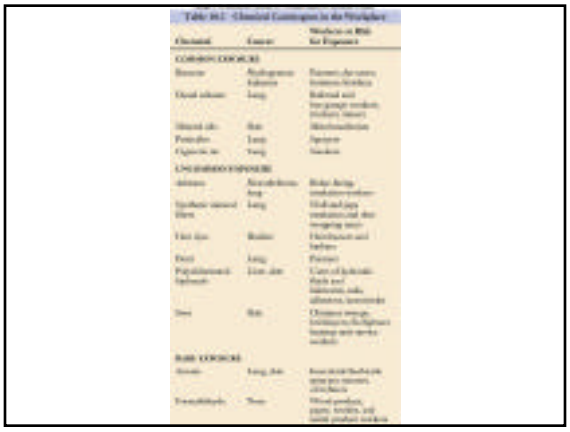


Fig. 18.12

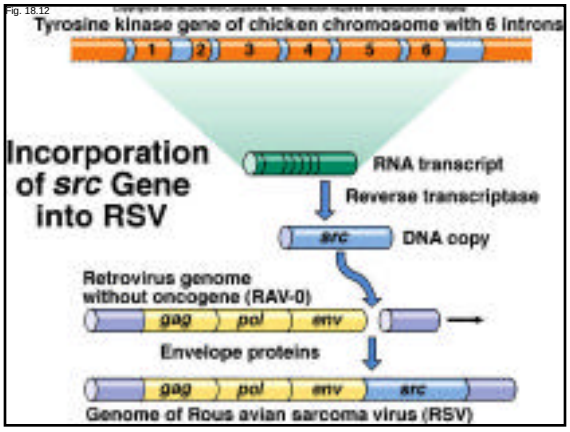


Fig. 18.13

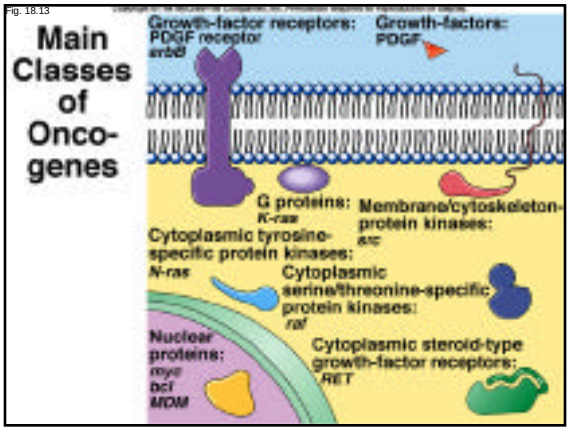


Table 18.4 Some Genes Implicated in Human Cancers

Gene	Product	Cancer
ONCOGENES		
Genes Encoding Growth Factors or Their Receptors		
erb-B	Receptor for epidermal growth factor	Glioblastoma (a brain cancer); breast cancer
erb-B2	A growth factor receptor (gene also called *neu*)	Breast cancer; ovarian cancer; salivary gland cancer
PDGF	Platelet-derived growth factor	Glioma (a brain cancer)
RET	A growth factor receptor	Thyroid cancer
Genes Encoding Cytoplasmic Relays in Intracellular Signaling Pathways		
K-ras	Protein kinase	Lung cancer; colon cancer; ovarian cancer; pancreatic cancer
N-ras	Protein kinase	Leukemias
Genes Encoding Transcription Factors That Activate Transcription of Growth-Promoting Genes		
c-myc	Transcription factor	Lung cancer; breast cancer; stomach cancer; leukemias
L-myc	Transcription factor	Lung cancer
N-myc	Transcription factor	Neuroblastoma (a nerve cell cancer)
Genes Encoding Other Kinds of Proteins		
bcl-2	Protein that blocks cell suicide	Follicular B cell lymphoma
bcl-1	Cyclin D1, which stimulates the cell cycle clock (gene also called *PRAD1*)	Breast cancer; head and neck cancers
MDM2	Protein antagonist of p53 tumor-supressor protein	Wide variety of sarcomas (connective tissue cancers)
TUMOR-SUPRESSOR GENES		
Genes Encoding Cytoplasmic Proteins		
APC	Step in a signaling pathway	Colon cancer; stomach cancer
DPC4	A relay in signaling pathway that inhibits cell division	Pancreatic cancer
NF-1	Inhibitor of ras, a protein that stimulates cell division	Neurofibroma; myeloid leukemia
NF-2	Inhibitor of ras	Meningioma (brain cancer); schwannoma (cancer of cells supporting peripheral nerves)
Genes Encoding Nuclear Proteins		
MTS1	p16 protein, which slows the cell cycle clock	A wide range of cancers
p53	p53 protein, which halts cell division at the G_1 checkpoint	A wide range of cancers
Rb	Rb protein, which acts as a master brake of the cell cycle	Retinoblastoma; breast cancer; bone cancer; bladder cancer
Genes Encoding Proteins of Unknown Cellular Locations		
BRCA1	?	Breast cancer; ovarian cancer
BRCA2	?	Breast cancer
VHL	?	Renal cell cancer

Fig. 18.14

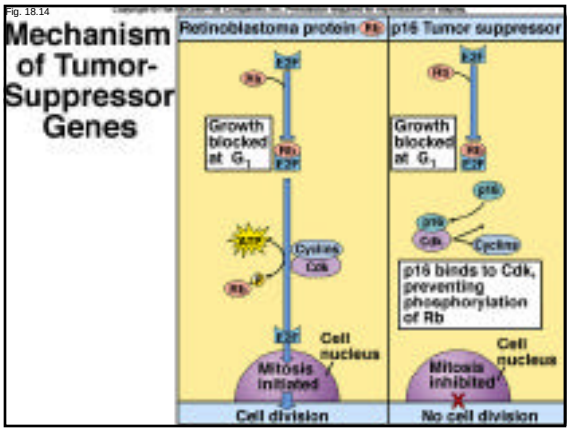


Fig. 18.15

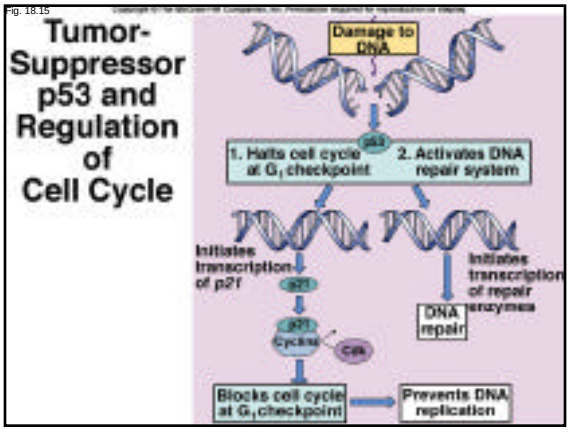


Fig. 18.16

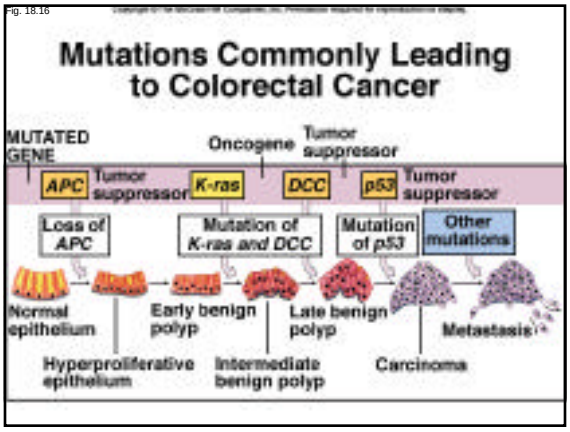


Fig. 18.17

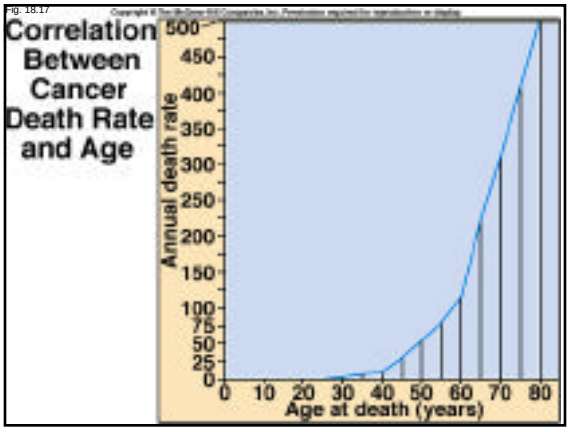
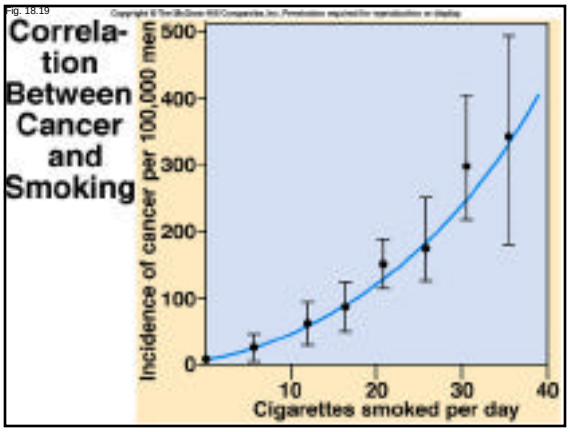


Fig. 18.18

Fig. 18.19



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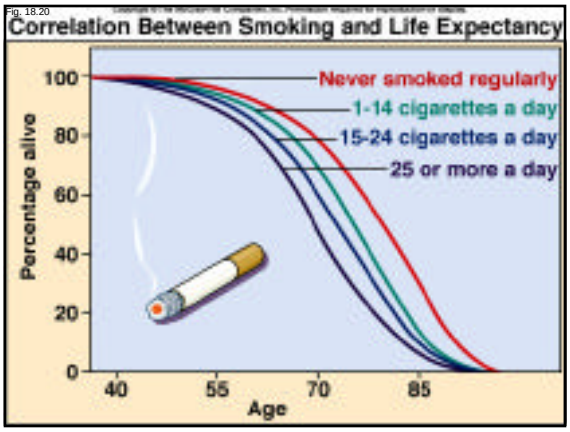

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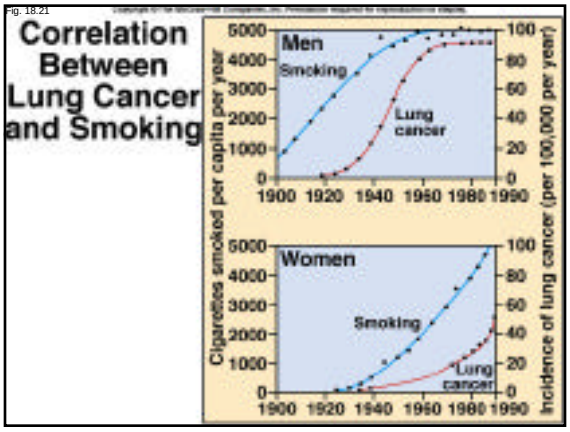

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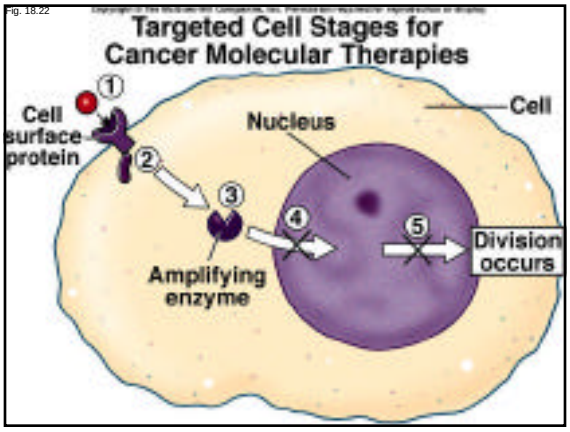


Fig. 18.23

Discoverer of Gene Transfer by Transposition – Barbara McClintock

Table 18.5 Classes of Genetic Recombination

Class	Occurrence
GENE TRANSFERS	
Conjugation	Occurs predominantly but not exclusively in bacteria and is targeted to specific locations in the genome
Transposition	Common in both bacteria and eukaryotes; genes move to new genomic locations, apparently at random
RECIPROCAL RECOMBINATIONS	
Crossing over	Requires the pairing of homologous chromosomes and may occur anywhere along their length
Unequal crossing over	The result of crossing over between mismatched segments; leads to gene duplication and deletion
Gene conversion	Occurs when homologous chromosomes pair and one is "corrected" to resemble the other
Independent assortment	Haploid cells produced by meiosis contain only one randomly selected member of each pair of homologous chromosomes

Fig. 18.24

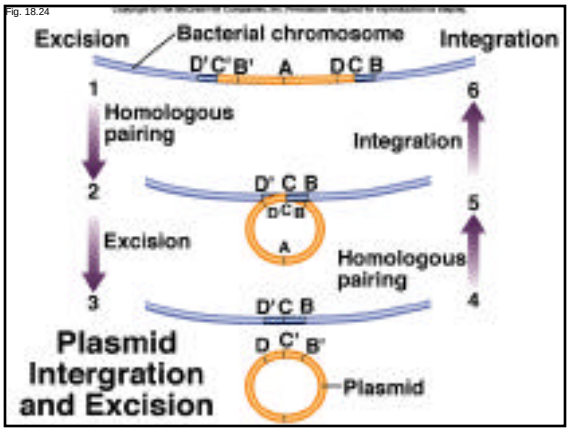


Fig. 18.25

Pilus Contact

Fig. 18.26

Gene Transfer Between Bacteria

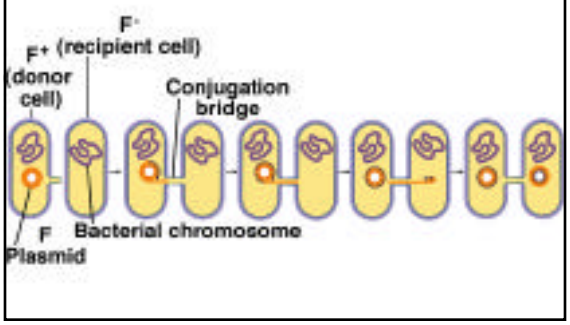


Fig. 18.27

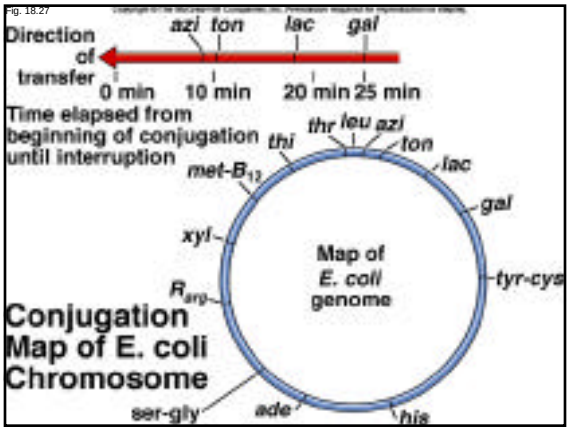


Fig. 18.28

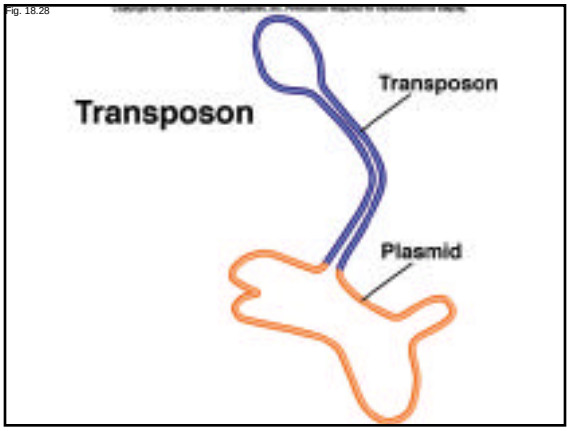


Fig. 18.29

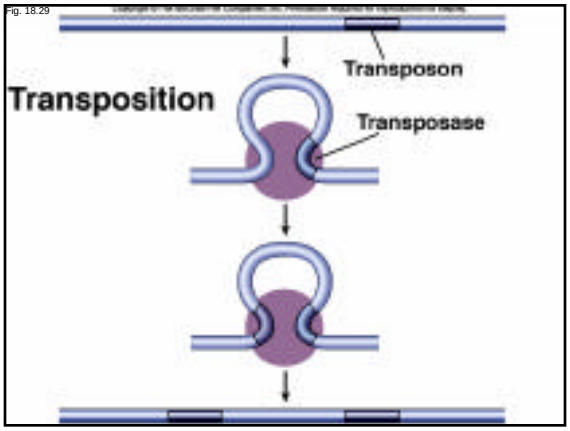


Fig. 18.30

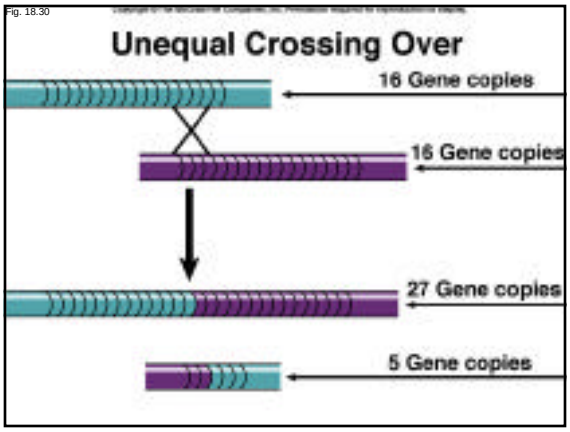
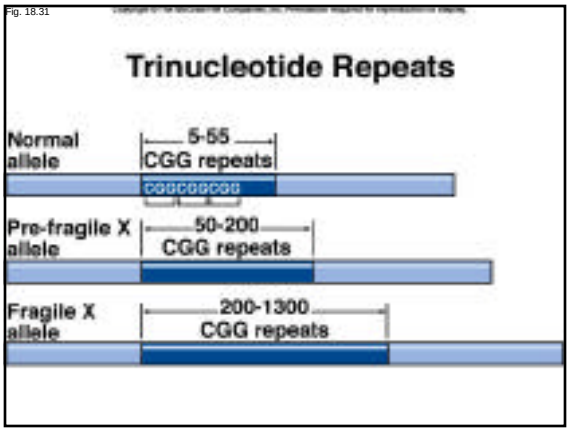


Fig. 18.31



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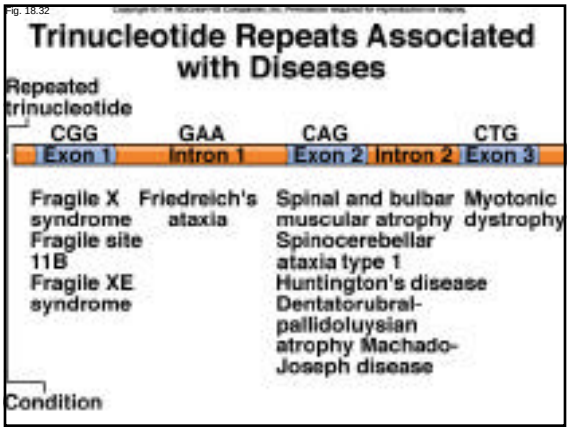
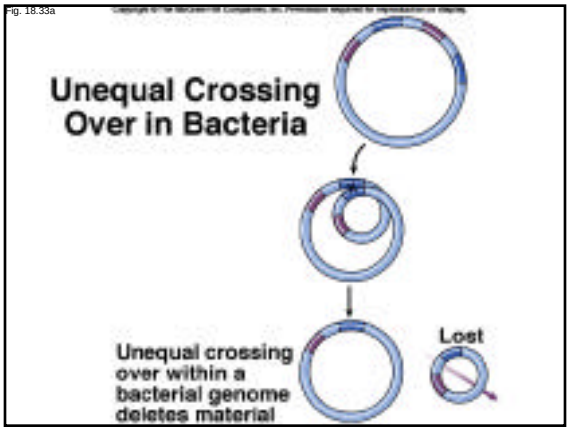


Fig. 18.33a



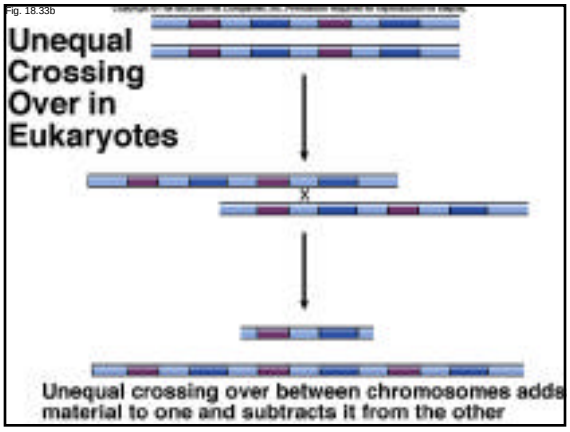


Fig. 18.33b
Unequal Crossing Over in Eukaryotes
Unequal crossing over between chromosomes adds material to one and subtracts it from the other

The End.