Chromosomal Mutations

- Chromosome structure

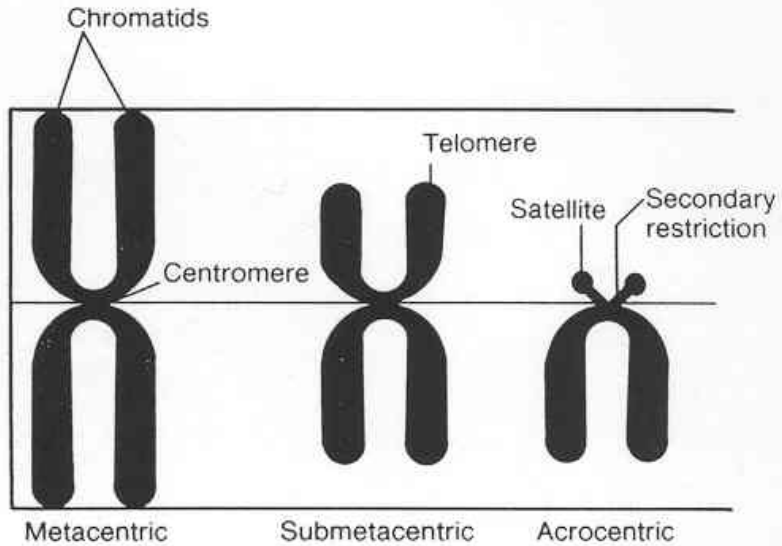
- Variation in Chromosome structure
- Duplication
- Deletions
- Inversions
- Translocations

- Change in Chromosome number and sets

Chromosome structure

- Human Karyotype
- Groups A-G
- P and q arms
- centromere location
- banding patterns

Figure 15.5 Chromosome Morphology Based on Location of the Centromere. The metacentric centromere divides the chromosome into arms of equal length. A submetacentric centromere is located off to one side, dividing the chromosome into arms of unequal lengths. The acrocentric centromere is located near one end of the chromosome. There are five pairs of acrocentric chromosomes in the human karyotype: pairs 13, 14, 15, 21, and 22. A satellite is a pale-staining chromosome segment located at the end of a secondary restriction. Each chromosome consists of a pair of chromatids. The telomere is the completed end region of each chromosome.



Karyotypes

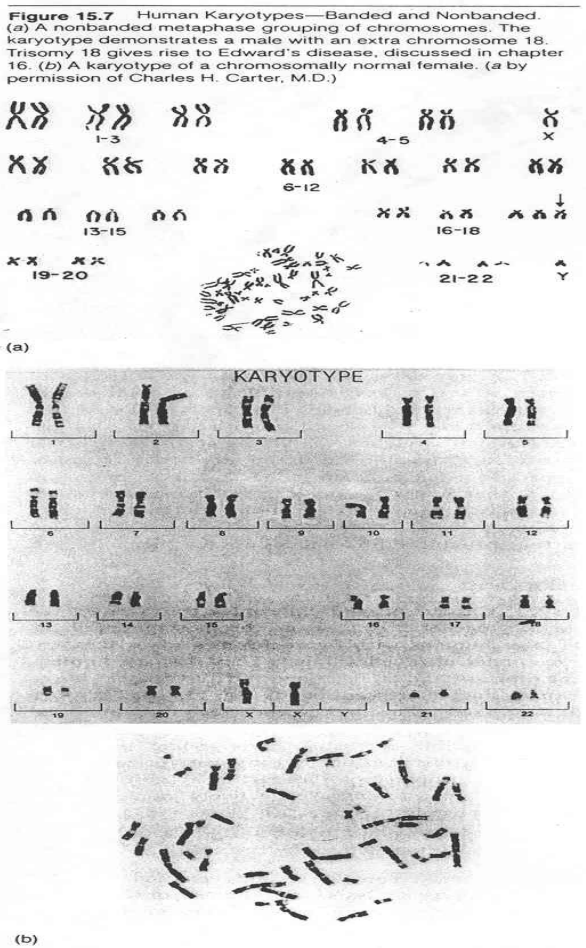
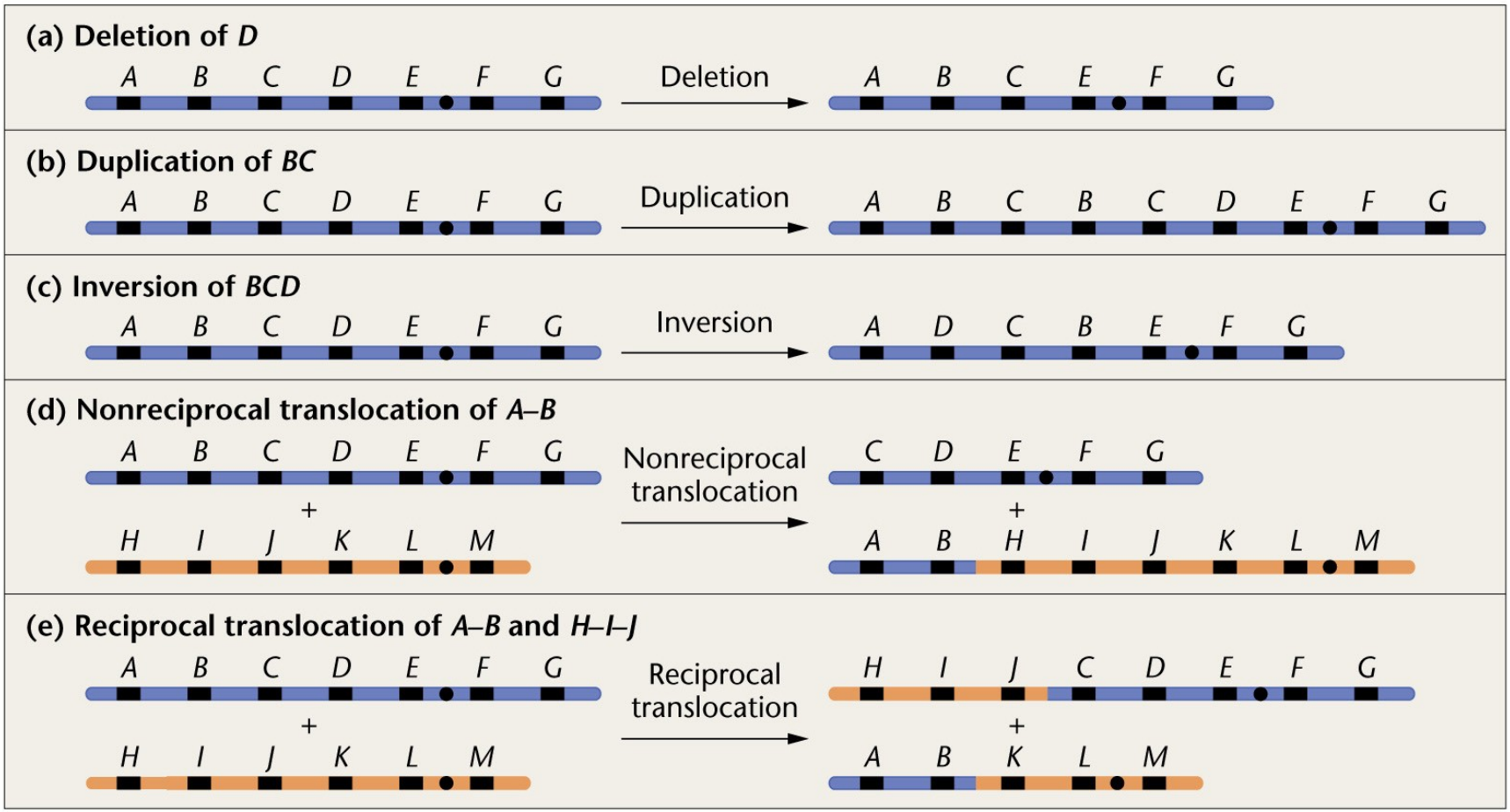


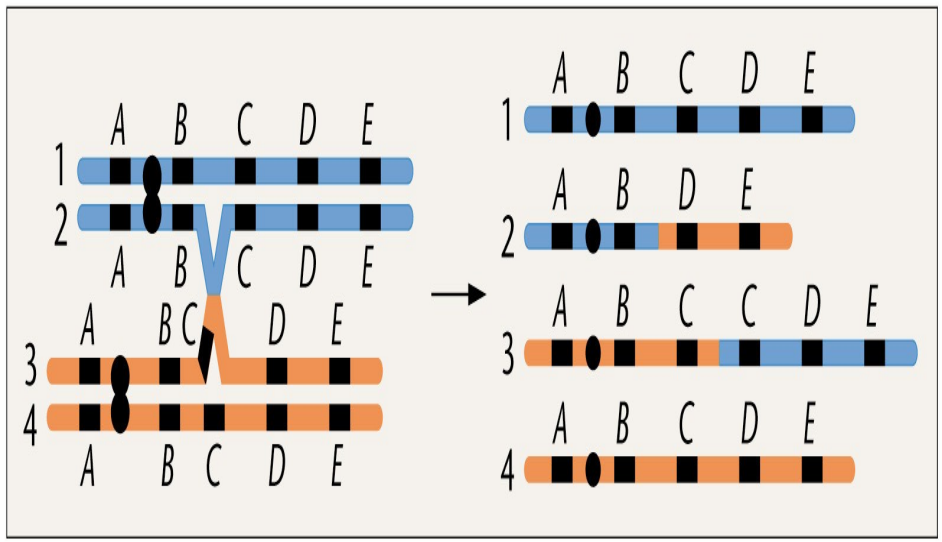
Figure 15.7 Human Karyotypes—Banded and Nonbanded. (*a*) A nonbanded metaphase grouping of chromosomes. The karyotype demonstrates a male with an extra chromosome 18. Trisomy 18 gives rise to Edward's disease, discussed in chapter 16. (*b*) A karyotype of a chromosomally normal female. (*a* by permission of Charles H. Carter, M.D.)

Group	Chromosome number	Description
A	1 to 3	The longest metacentrics. Distinguished from each other by centromere location.
B	4 and 5	The longest submetacentrics. Difficult to separate without banding.
C	6 to 12	Medium-sized metacentrics. Must use banding to identify.
D	13 to 15	Medium-sized acrocentrics with satellites.
E	16 to 18	Short metacentric 16 and submetacentrics 17 and 18.
F	19 and 20	Short metacentrics chromosomes.
G	21 and 22	Short acrocentrics with satellites.
XX	About the size and shape of the C-group chromosomes. In some karyotypes, the X chromosomes are placed to the right of the C group.	
Y	Similar in size and shape to the G-group chromosomes, but does not have satellites.	

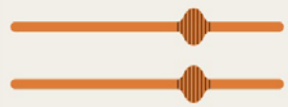
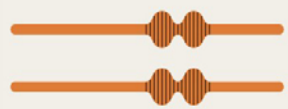
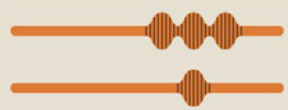
Variation in Chromosome structure



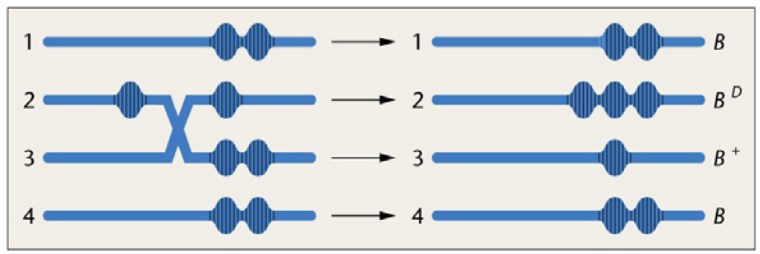
Duplication



(a) Genotypes and Phenotypes

Genotype	Facet Number	Phenotype	= 16A segments
B^+ / B^+	779		
B / B^+	358		
B / B	68		
B^D / B^+	45		

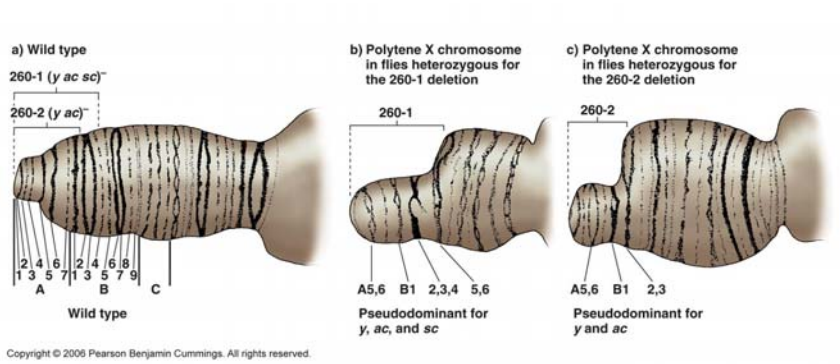
(b) Origin of B^D allelle as a result of unequal crossing over

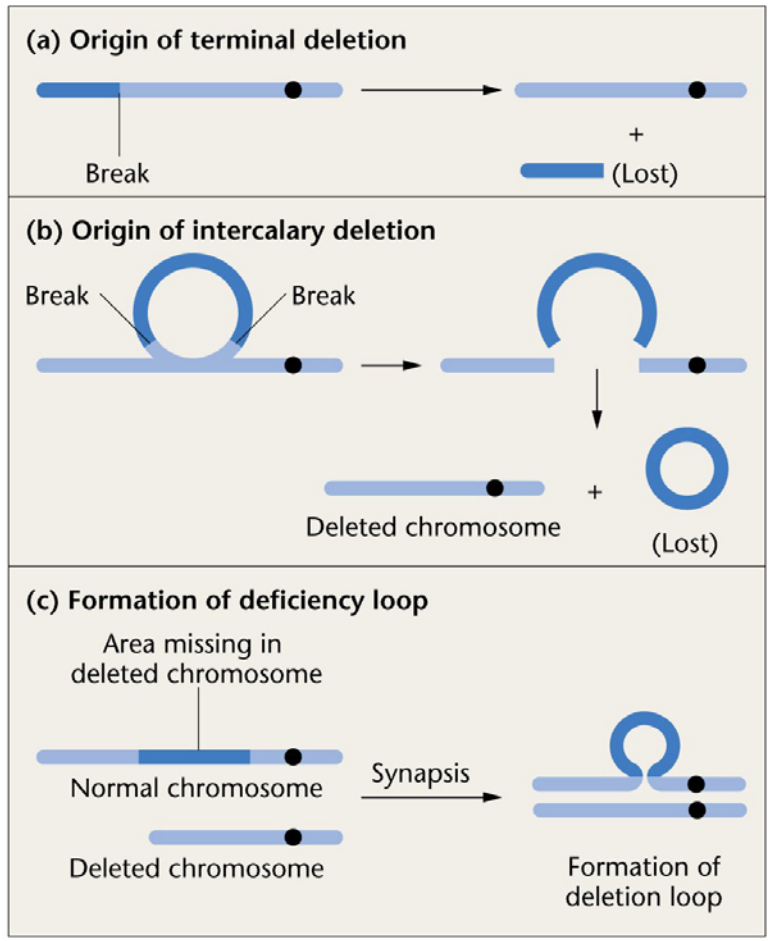


Deletions

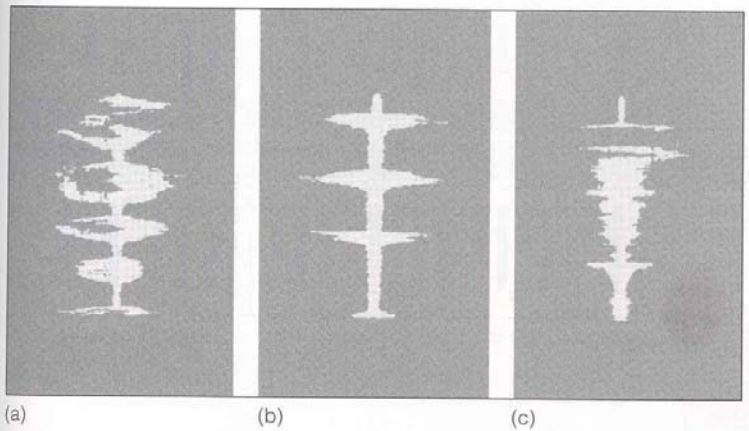
- terminal
- intercalary

- Pseudodominance



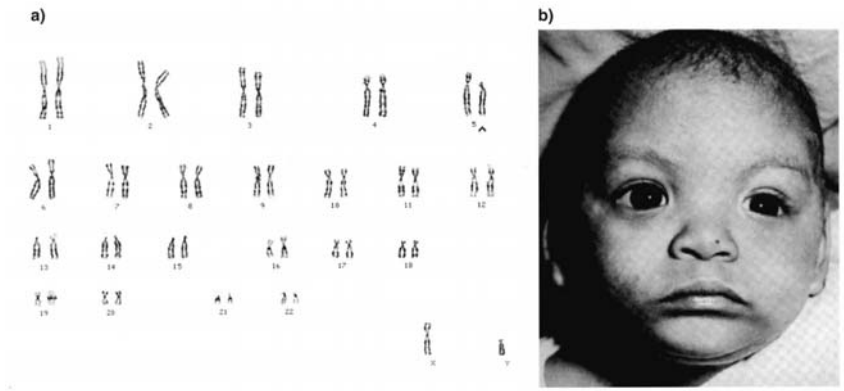


Cri cu chat

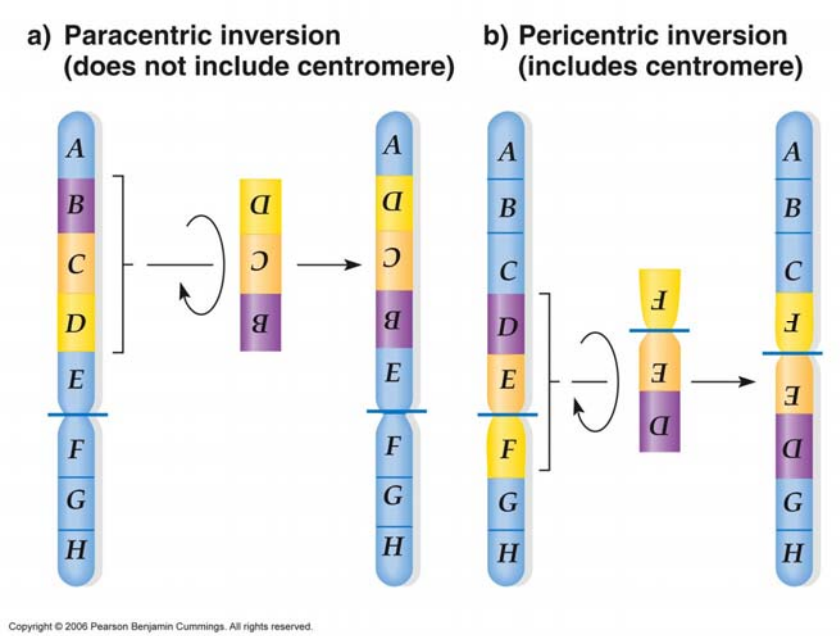


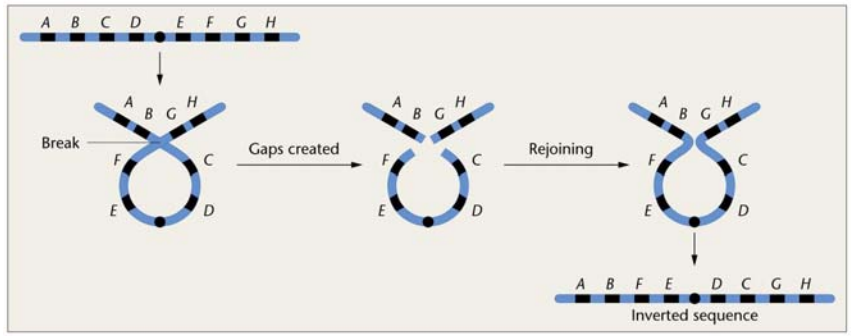
(a) (b) (c)

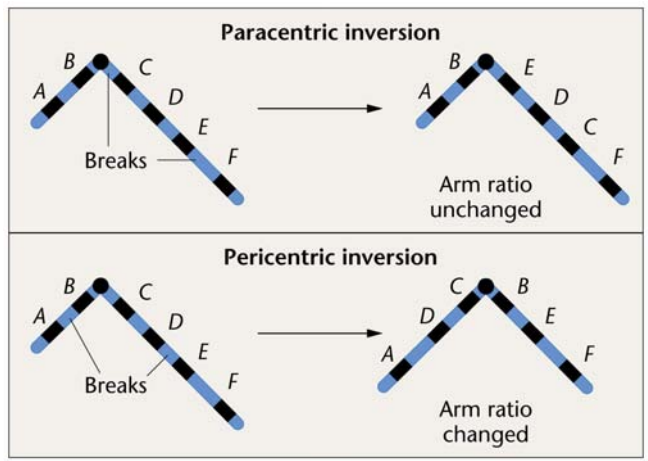
FIGURE 6.24 Sound recordings of (a) a normal infant, (b) an infant who has cri du chat syndrome, and (c) a cat. The cry of the affected infant is much closer in sound pattern to that of the cat than to that of a normal infant, giving rise to the name of the syndrome, cry of the cat.



Inversions

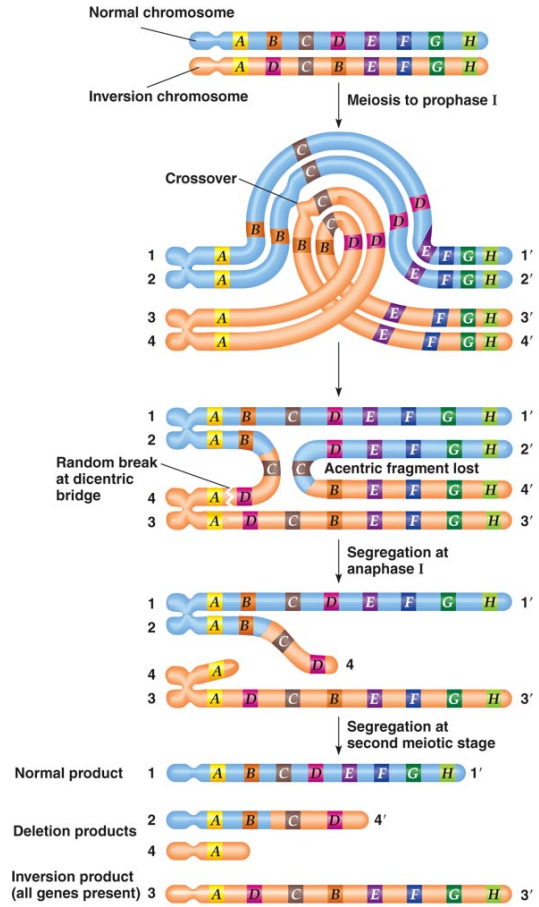




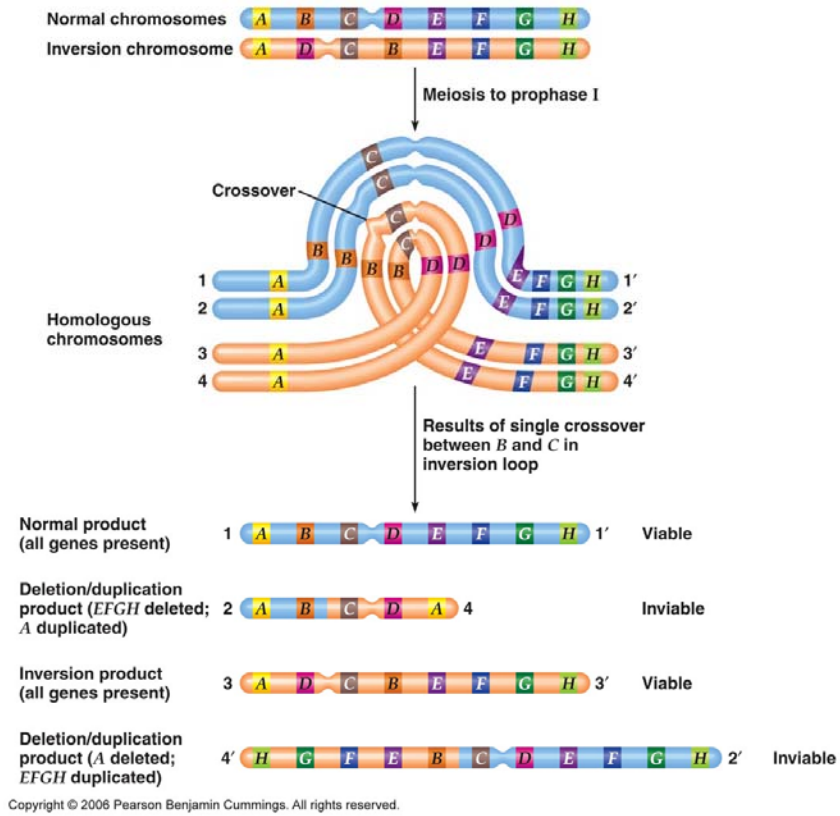


Inversions

Paracentric inversion

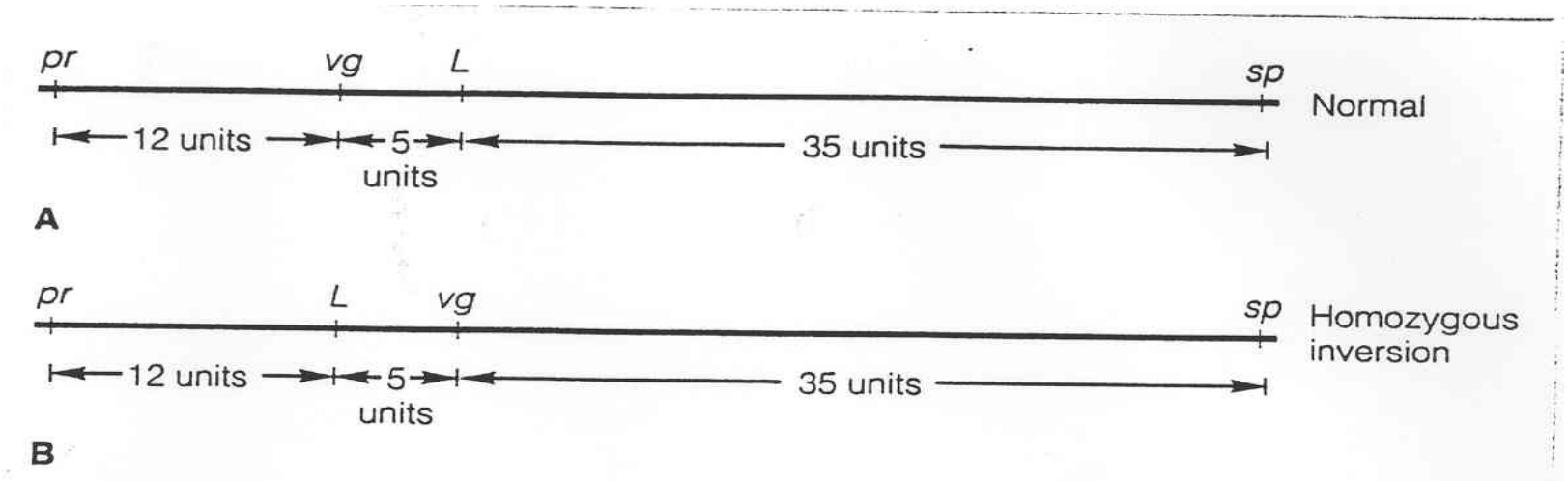


Pericentric inversion

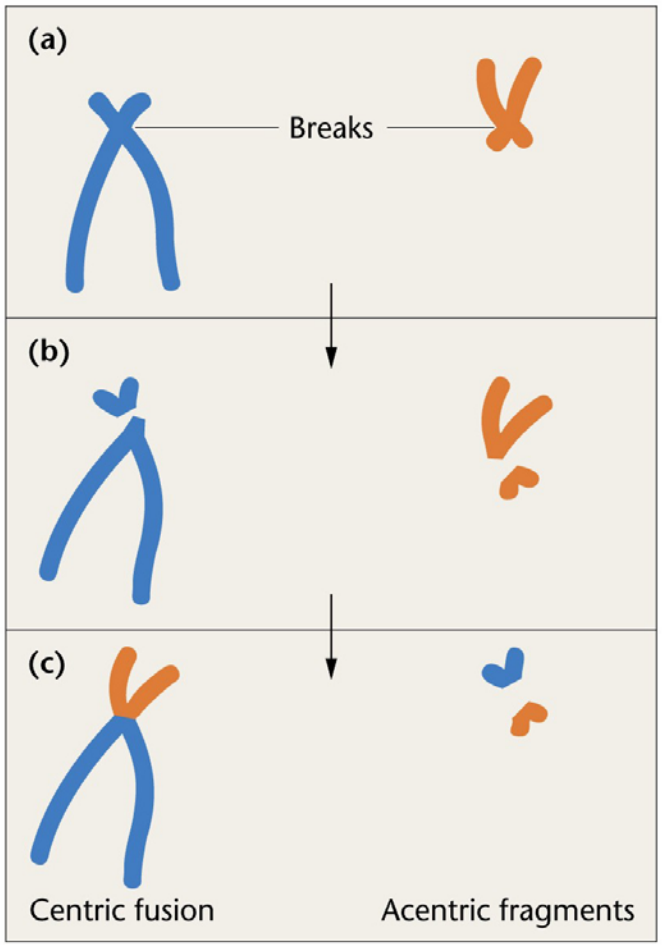


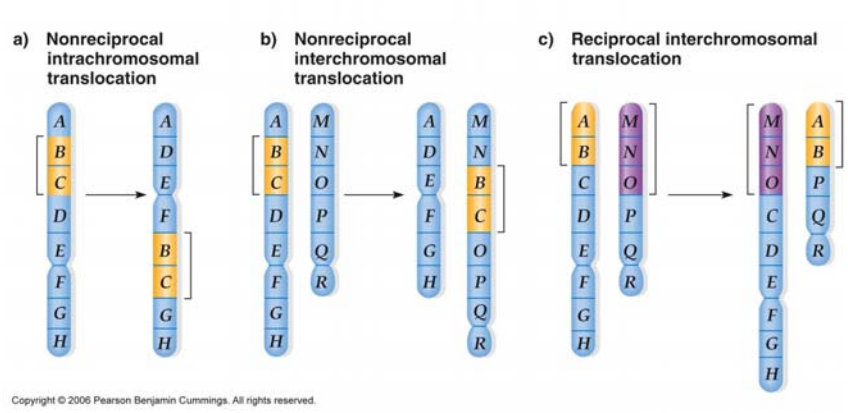
Inversions

- Suppression of recombinant recovery
- Maintenance of a set of specific alleles at adjacent loci
- Can lead to evolutionary advantage

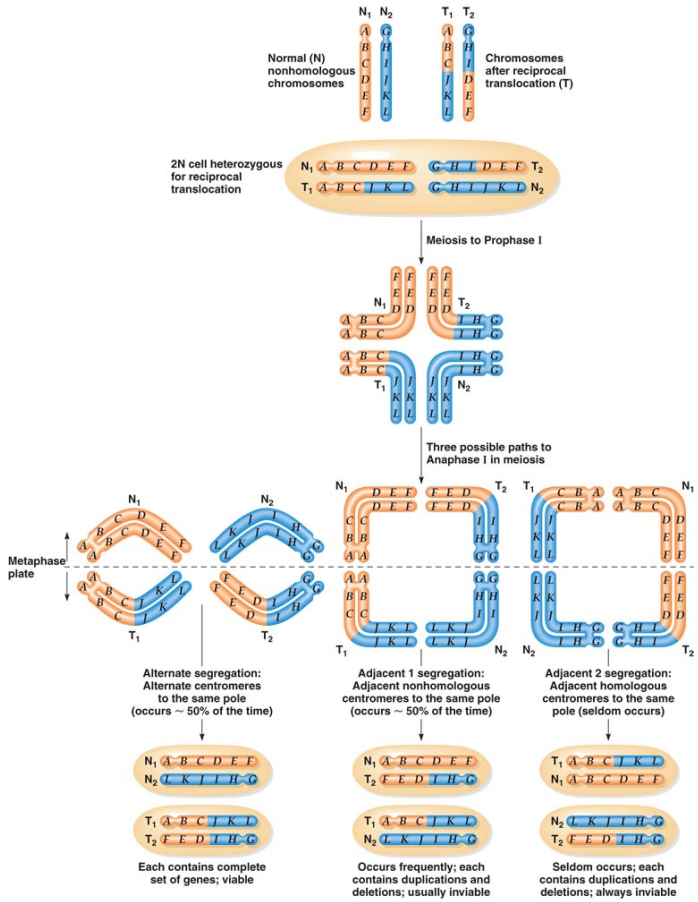


Translocations

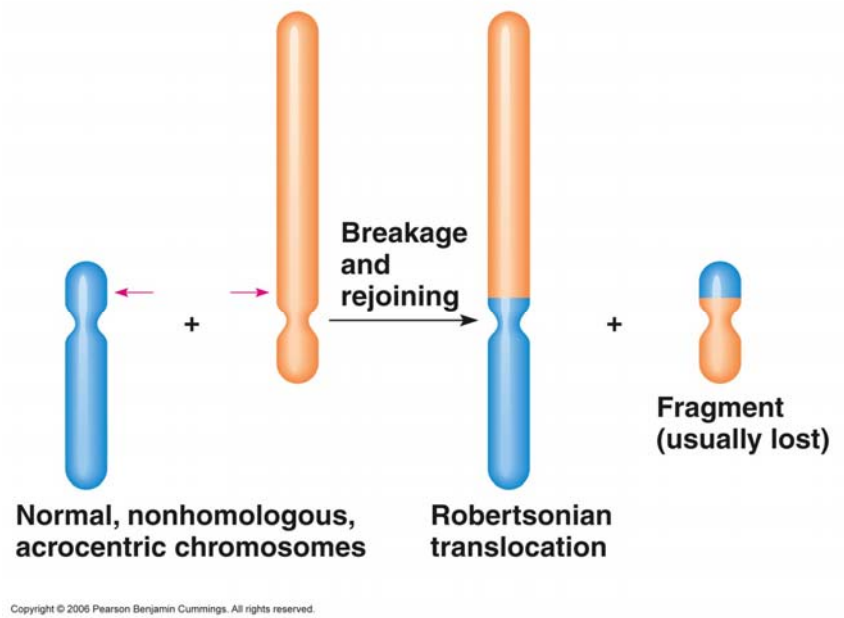


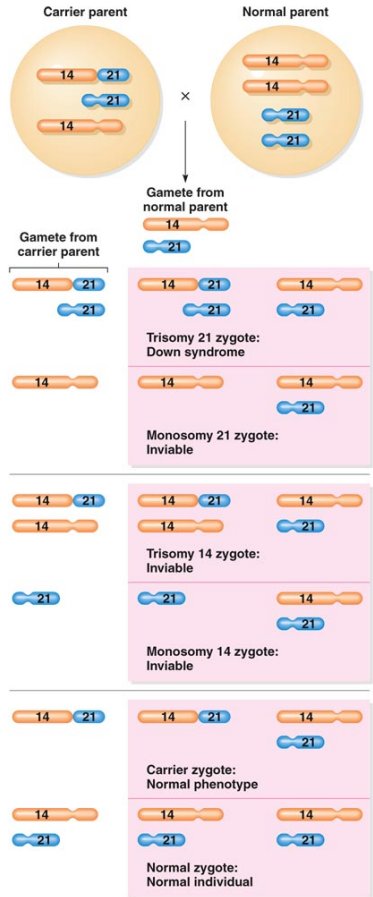


Meiosis



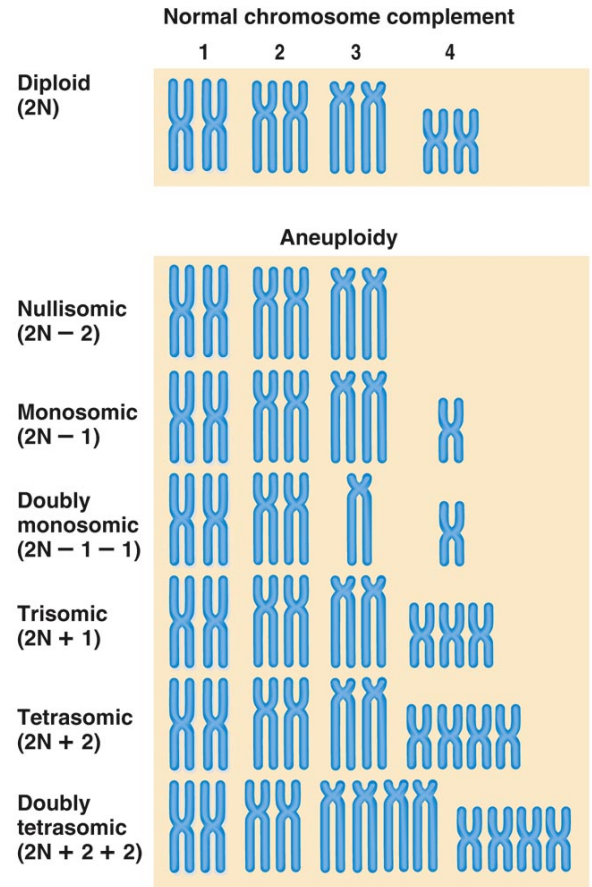
Familial Down syndrome



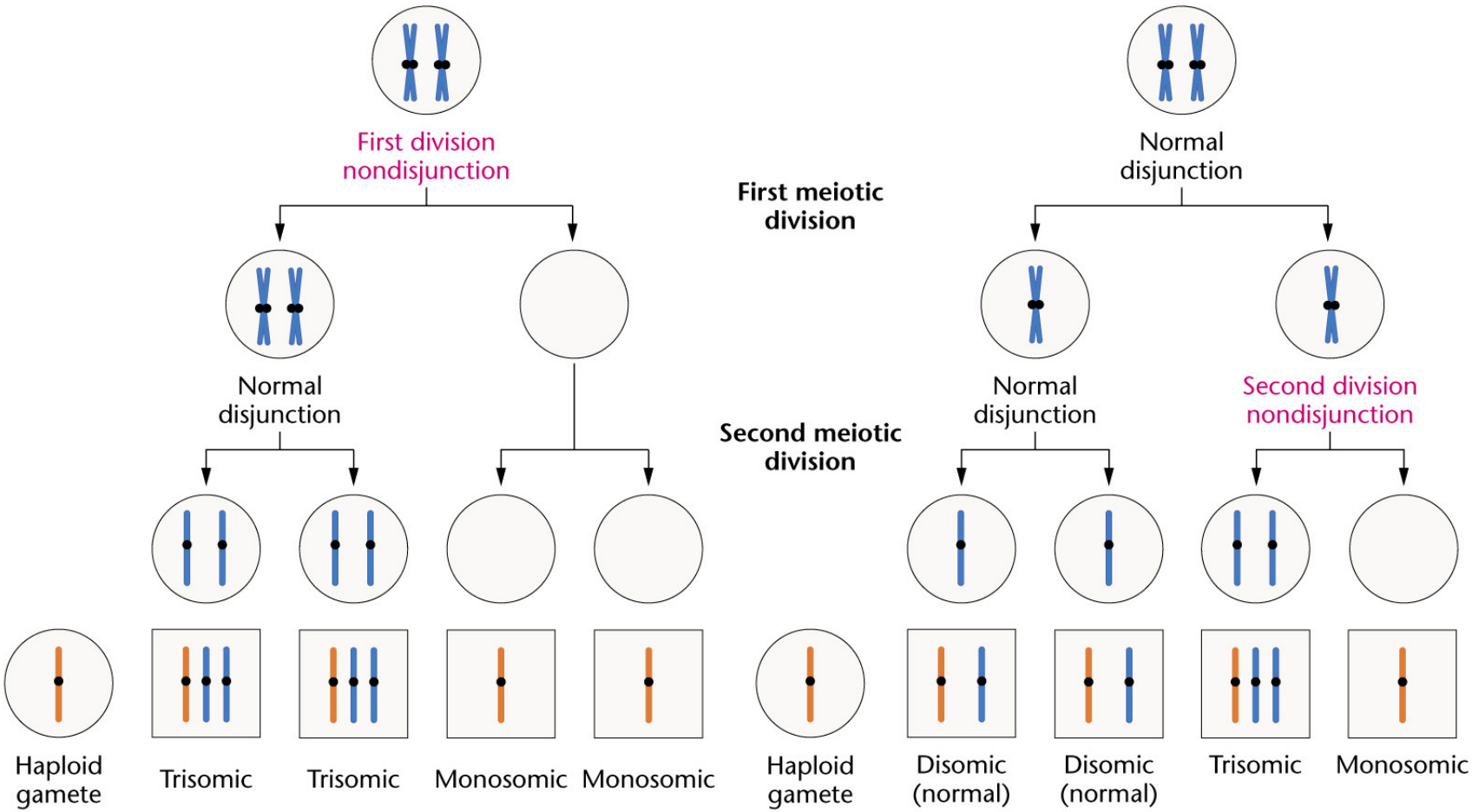


Change in Chromosome number and sets

- Aneuploidy
- Non-disjunction
- Monosomy
- Trisomy
- Sex-chromosomes
- Polyploidy



Non-Disjunction



Monosomy & Trisomy

Chromosome constitution	*Number among spontaneously aborted fetuses*	*Number among live births*
Normal	7500	84,450
Trisomy		
13	128	17
18	223	13
21	350	113
Other autosomes	3176	0
Sex chromosomes		
47,XYY	4	46
47,XXY	4	44
45,X	1350	8
47,XXX	21	44
Translocations		
Balanced (euploid)	14	164
Balanced (aneuploid)	225	52
Polyploid		
Triploid	1275	0
Tetraploid	450	0
Others (mosaics, etc.)	280	49
Total	15,000	85,000

Trisomy and Triploid

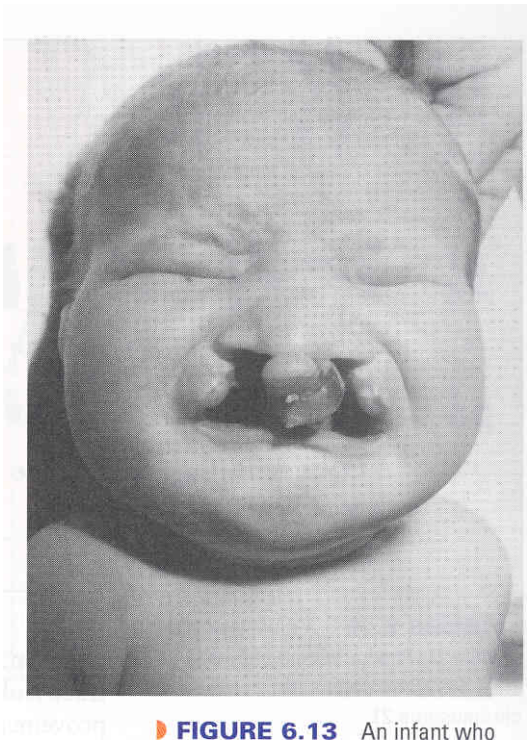


FIGURE 6.13 An infant who

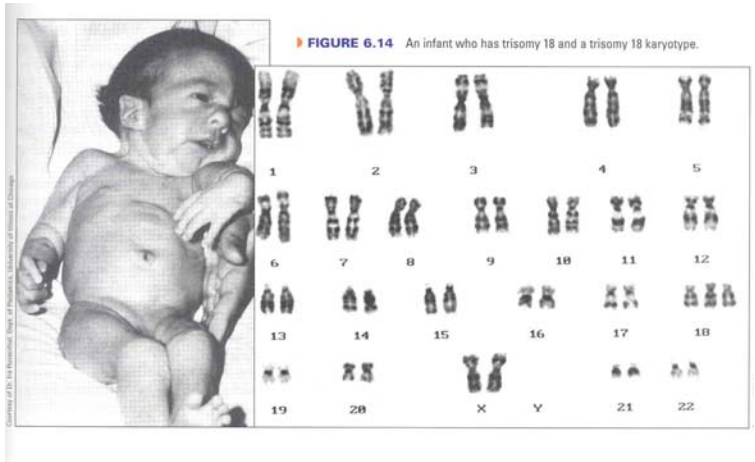
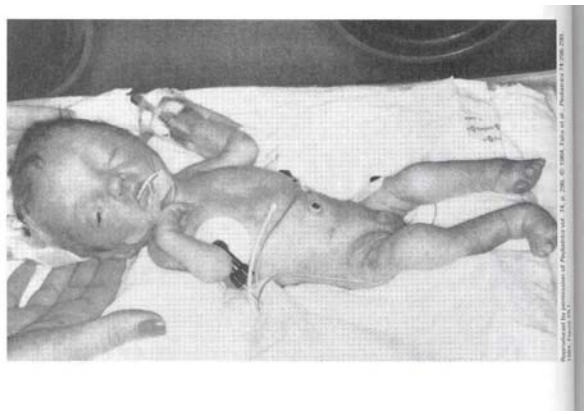


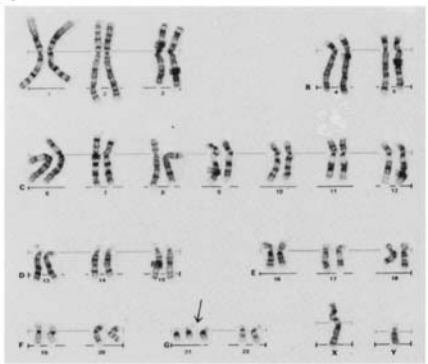
FIGURE 6.14 An infant who has trisomy 18 and a trisomy 18 karyotype.

FIGURE 6.10 A triploid infant, showing the characteristic enlarged head.



Monosomy & Trisomy

a)

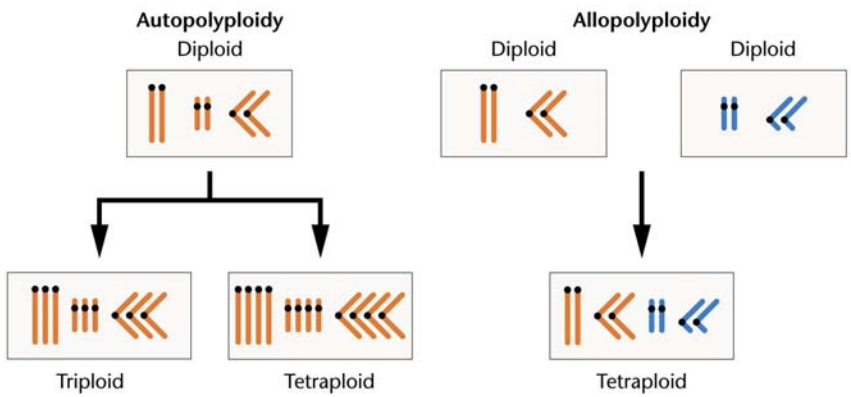


b)

Table 8.2 Relationship Between Age of Mother and Risk of Trisomy-21

Age of Mother	Risk of Trisomy-21 in Child
16–26	7.7/10,000
27–34	4/10,000
35–39	29/10,000
40–44	100/10,000
45–47	333/10,000
All mothers combined	14.3/10,000

Polyploidy



Multiples of same genome

Multiples of Different genomes