

# Chromosomal Aberrations

# Types of Chromosome

Metacentric:

Telocentric:

Acrocentric:

Sub-metacentric:

# Variations in Chromosome Number and Arrangement

# Terminology

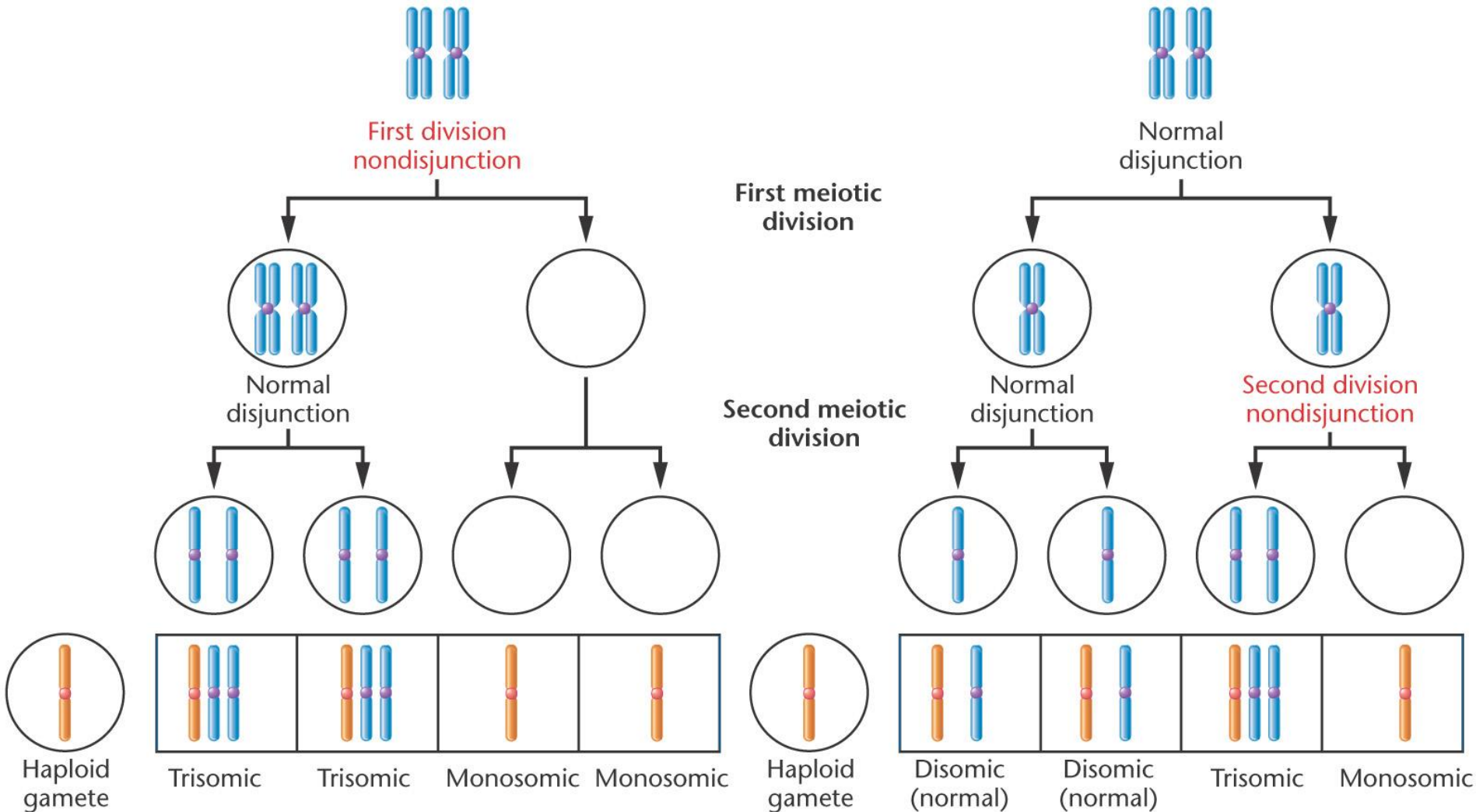
different

**TABLE 8.1****TERMINOLOGY FOR VARIATION  
IN CHROMOSOME NUMBERS**

Term	Explanation
Aneuploidy	$2n \pm x$ chromosomes
Monosomy	$2n - 1$
Trisomy	$2n + 1$
Tetrasomy, pentasomy, etc.	$2n + 2, 2n + 3$ , etc.
Euploidy	Multiples of $n$
Diploidy	$2n$
Polyploidy	$3n, 4n, 5n, \dots$
Triploidy	$3n$
Tetraploidy, pentaploidy, etc.	$4n, 5n$ , etc.
Autopolyploidy	Multiples of the same genome
Allopolyploidy (Amphidiploidy)	Multiples of different genomes

# Aneuploidy

# Nondisjunction



# Nondisjunction In Drosophila



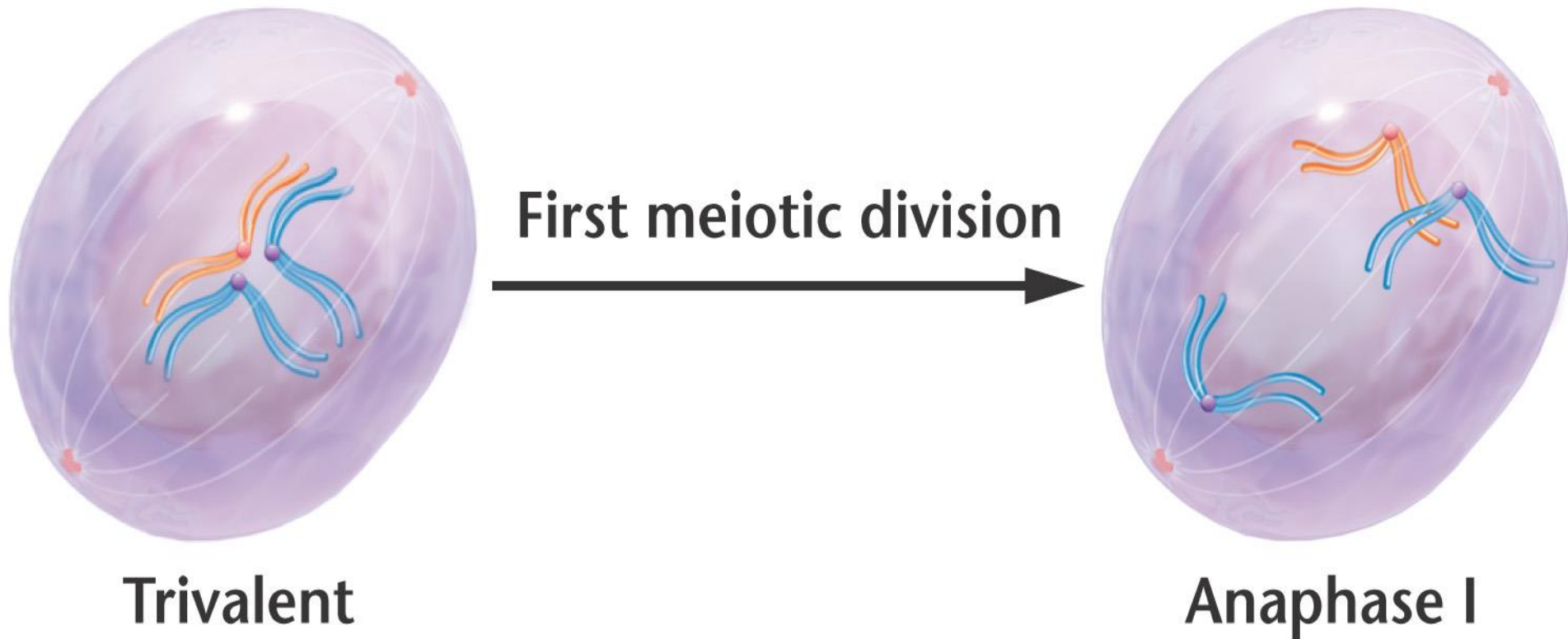
	W	Y
w	Ww (Red eyed female)	wY (White eyed Male)
ww	Www Dies	wwY White eyed female
(-)	W- Red eyed Male (Sterile)	-Y Dies

	W	Y
ww	Www (Super female dies)	wwY (White eyed female)
Y	WY Red eyed Male	YY Dies
w	Ww Red eyed Female	wY White eyed Male
wY	WwY Red eyed female	wYY White eyed Male

# Monosomy

# Trisomy

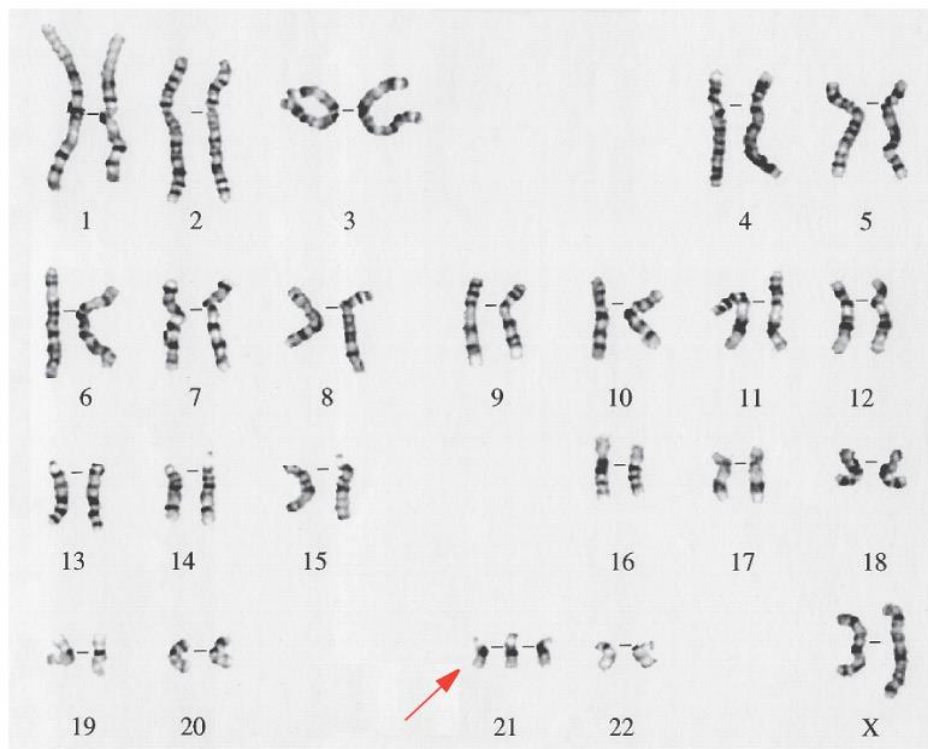
# Trisomy Meiosis



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# Down Syndrome

# Down Syndrome – Trisomy 21



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# Polyploidy in Plants



# Autopolyploidy

# Allopolyploidy

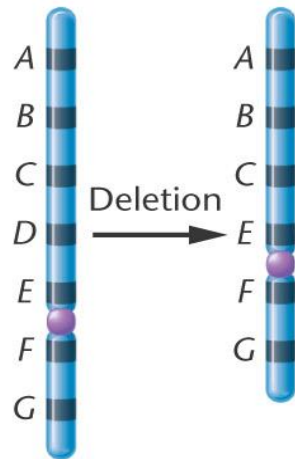
# Allotetraploids



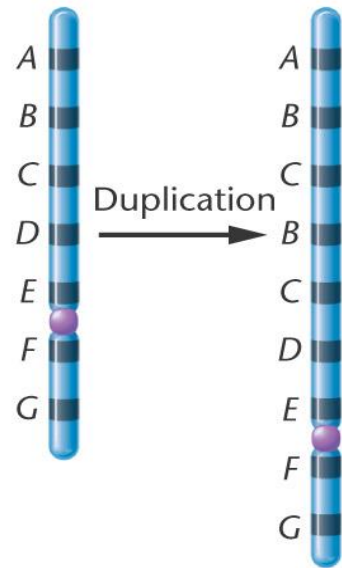
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# Chromosome Rearrangements

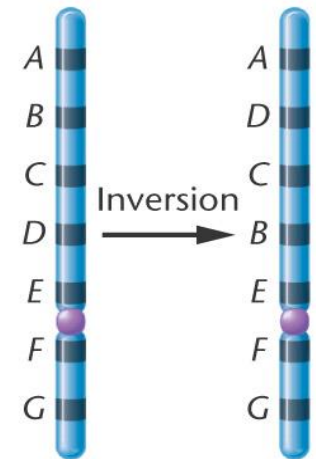
(a) Deletion of *D*



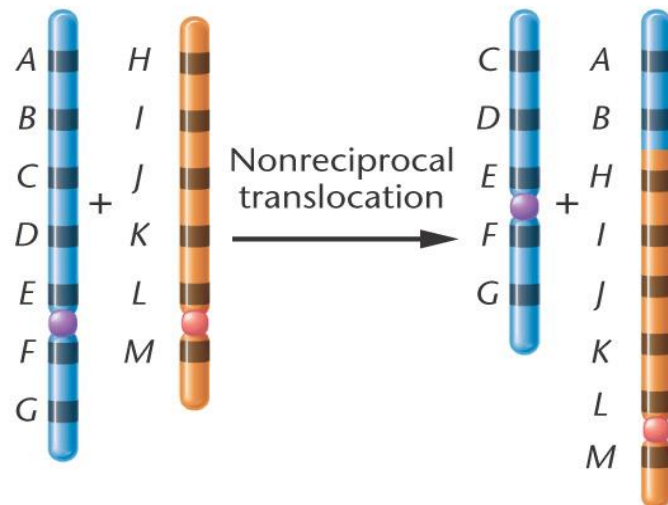
(b) Duplication of *BC*



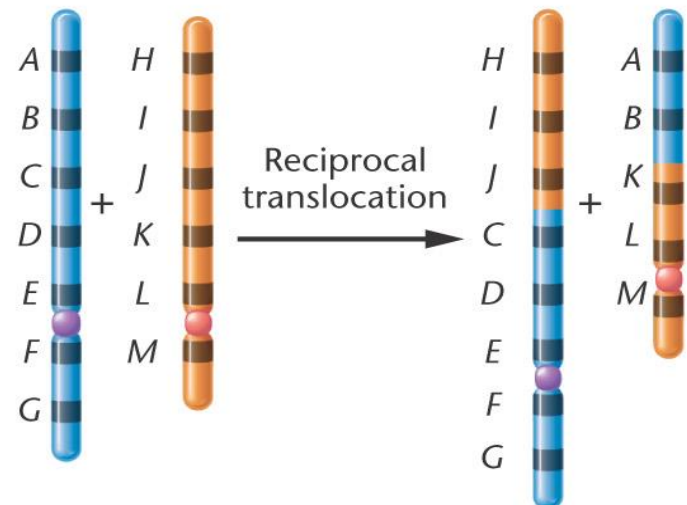
(c) Inversion of *BCD*



(d) Nonreciprocal translocation of *A-B*



(e) Reciprocal translocation of *A-B* and *H-I-J*

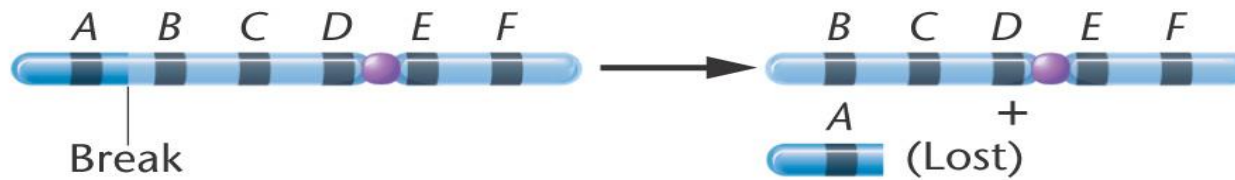


# Consequences of Rearrangements

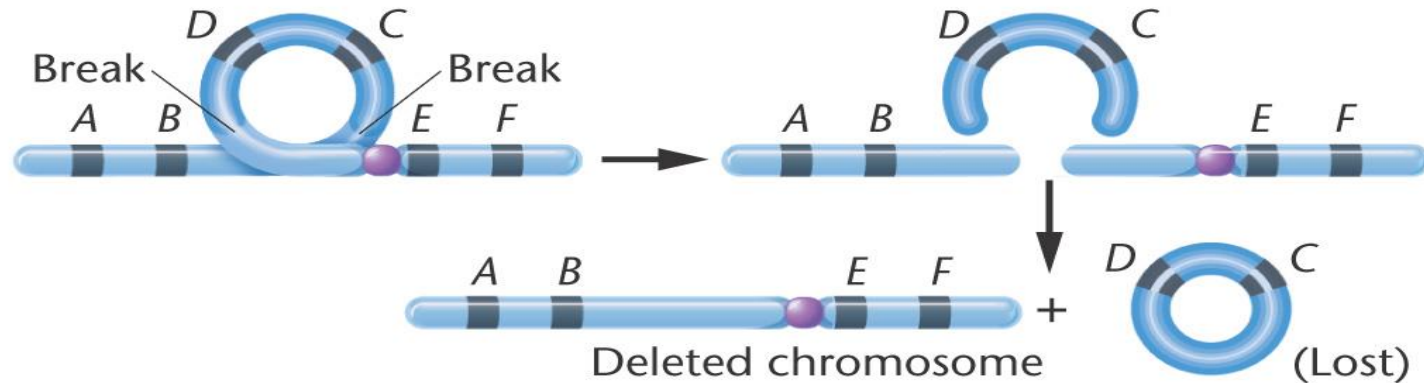
# Deletions



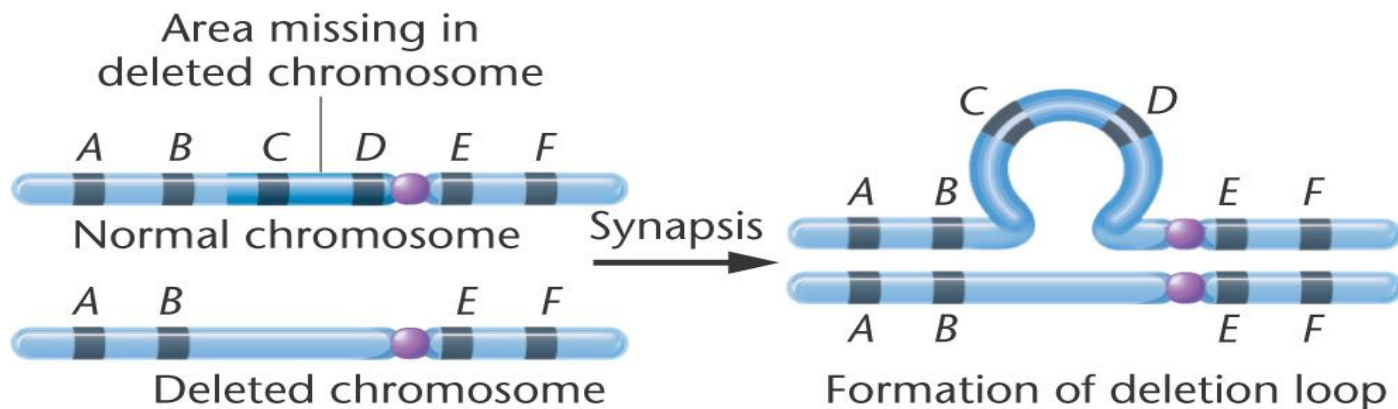
### (a) Origin of terminal deletion



### (b) Origin of intercalary deletion

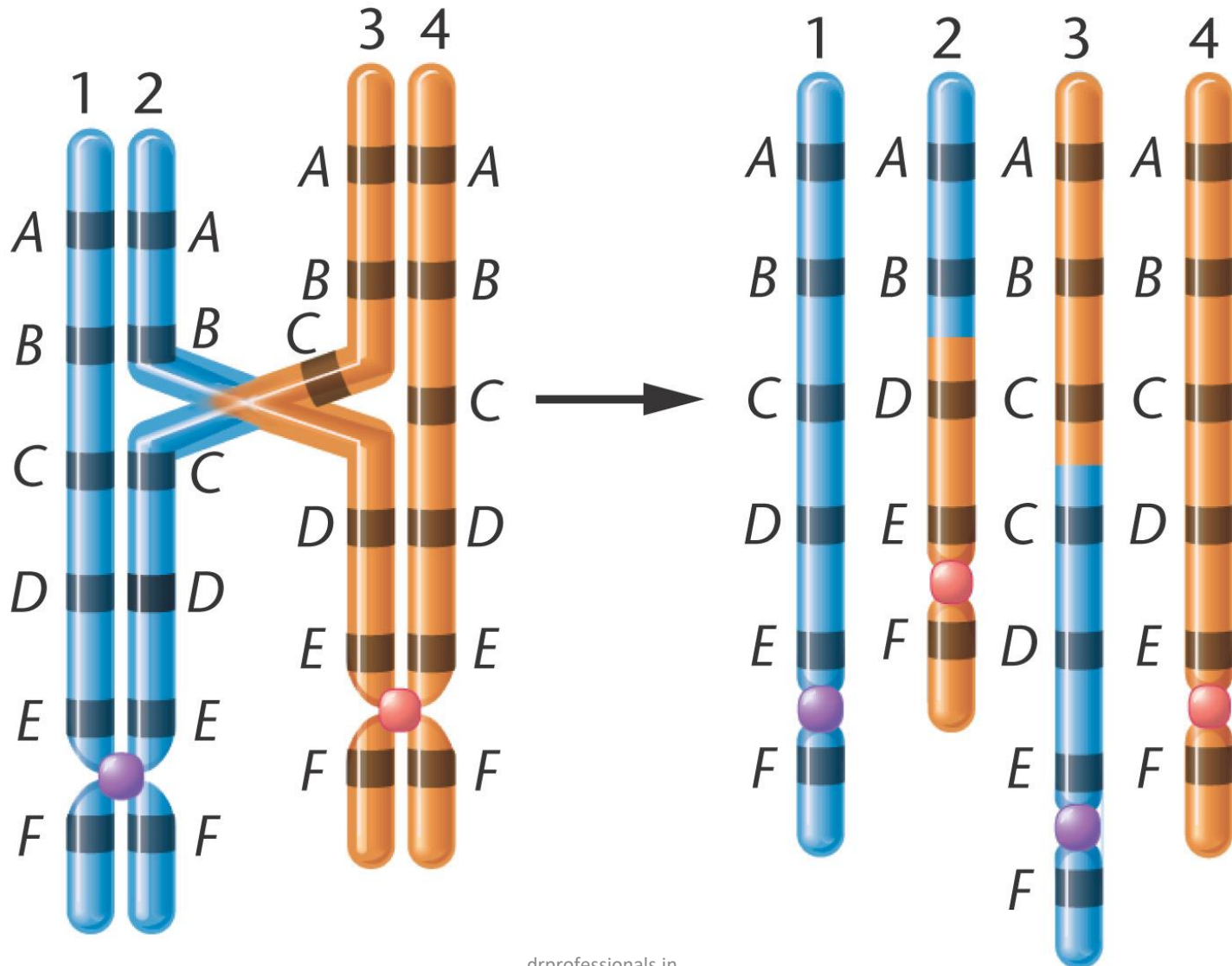


### (c) Formation of deficiency loop





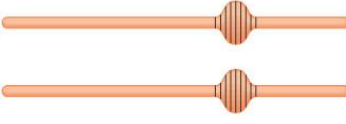

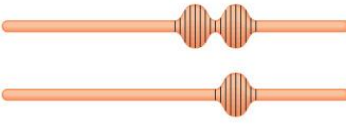

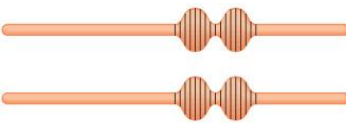

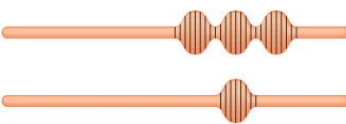
# Duplications

# Unequal Crossing Over



# Position Effects

(a) Genotypes and Phenotypes

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

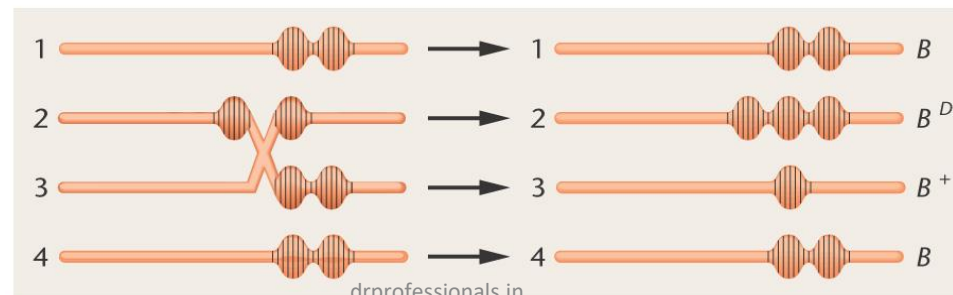


$B^+/B^+$



$B/B^+$

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

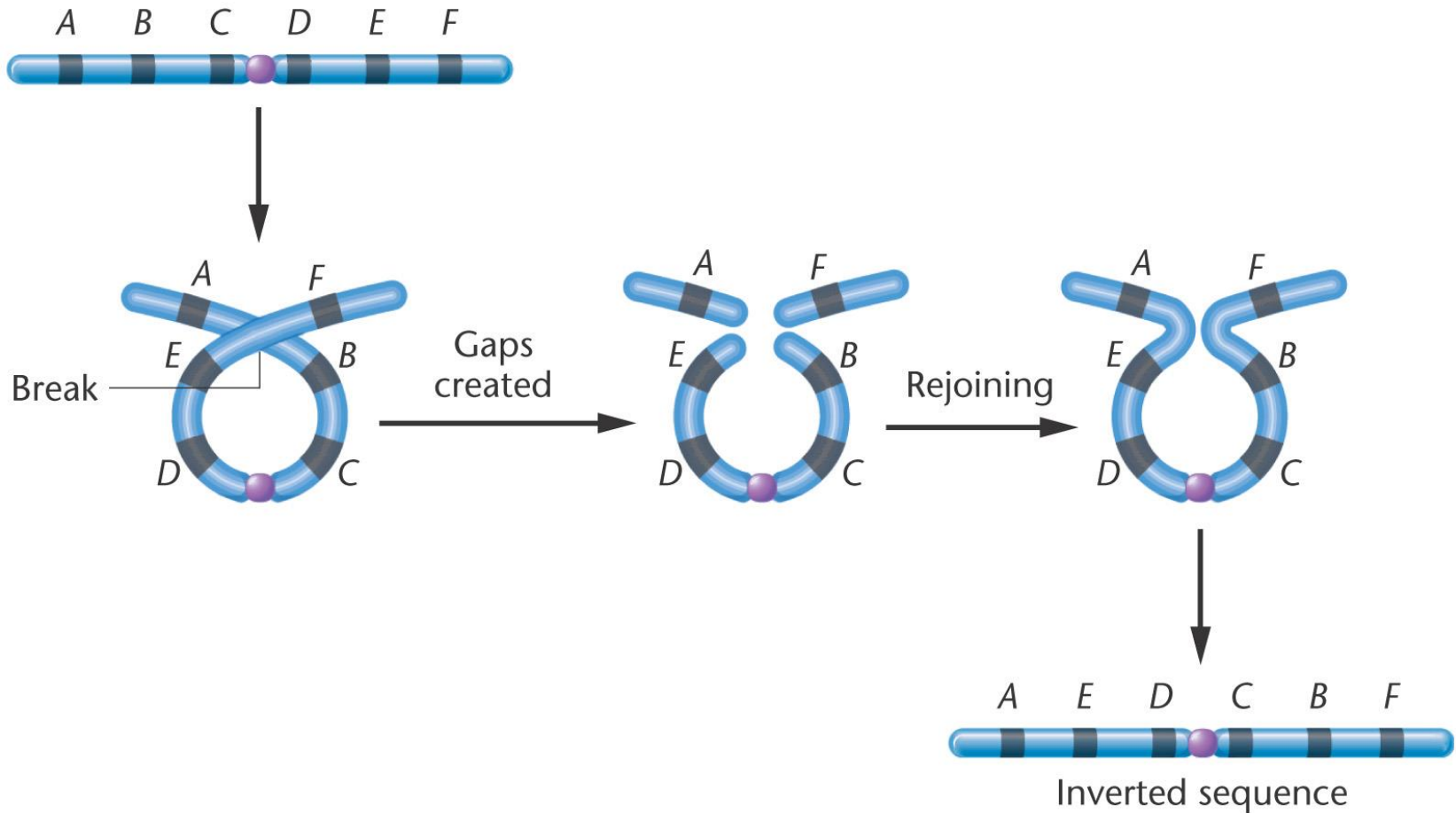


$B/B$

## Position Effect –Bar Eye condition in Drosophila

X Chromosome	Phenotype
B / B	Normal
BB / B	Heterozygous
BB / BB	Homozygous Bar Eye
BBB / B	Heterozygous Ultra Bar
BBB / BBB	Homozygous Ultra Bar

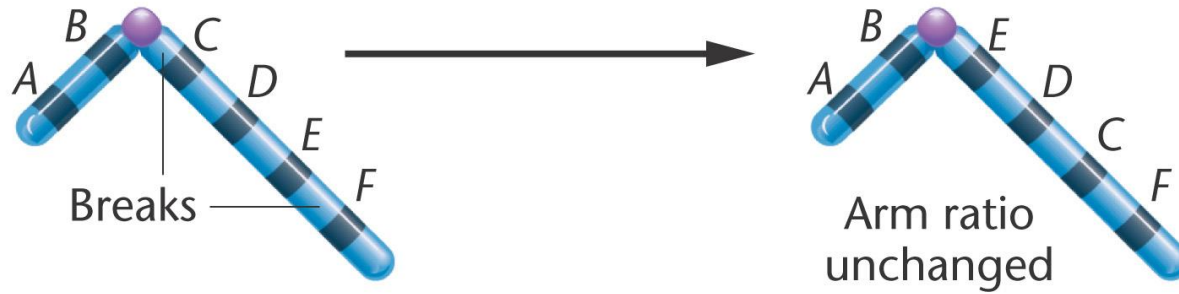
# Chromosomal Inversions



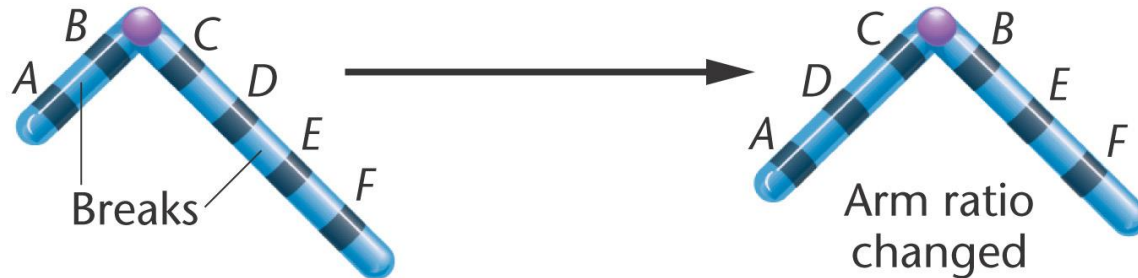
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# Chromosomal Inversions

## Paracentric inversion



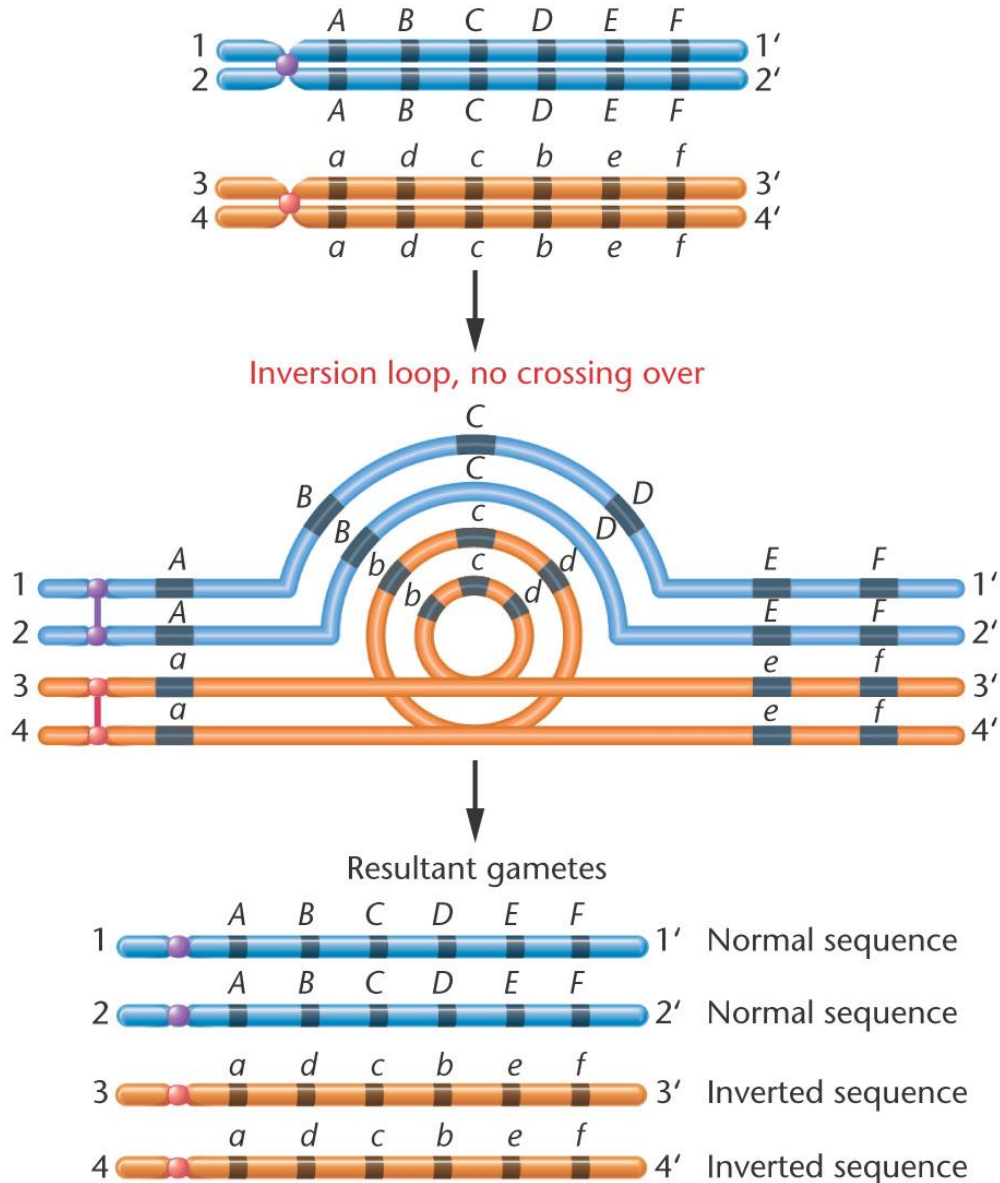
## Pericentric inversion



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# Inversions and Gametogenesis

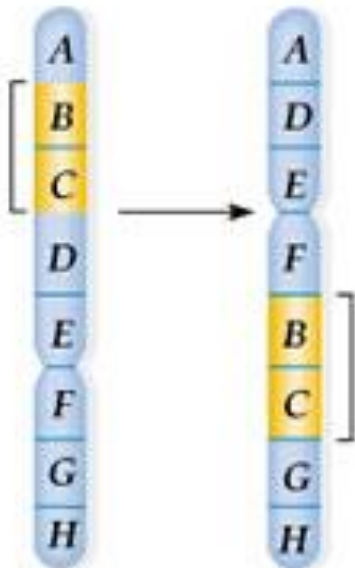
Paracentric inversion heterozygote



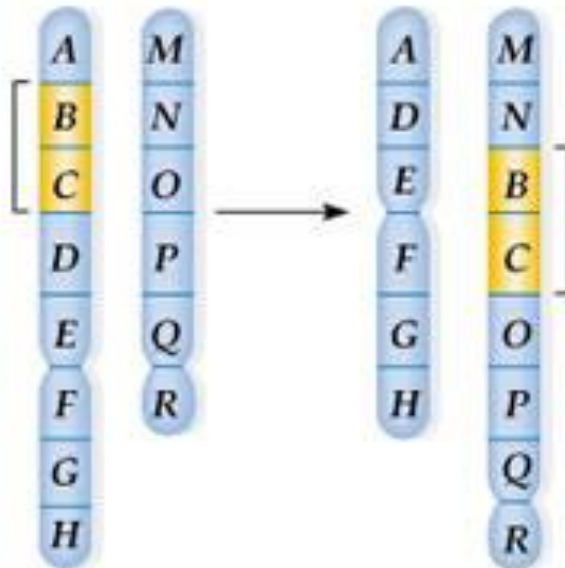


# Translocations

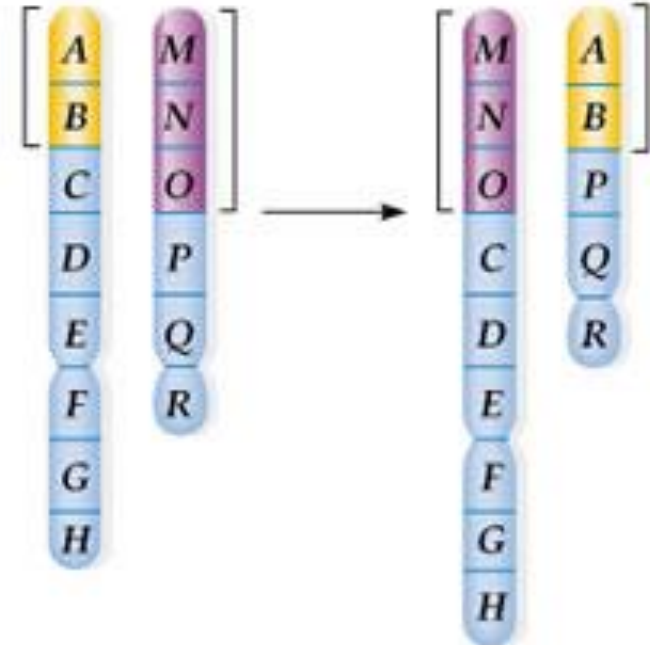
a) Nonreciprocal intrachromosomal translocation



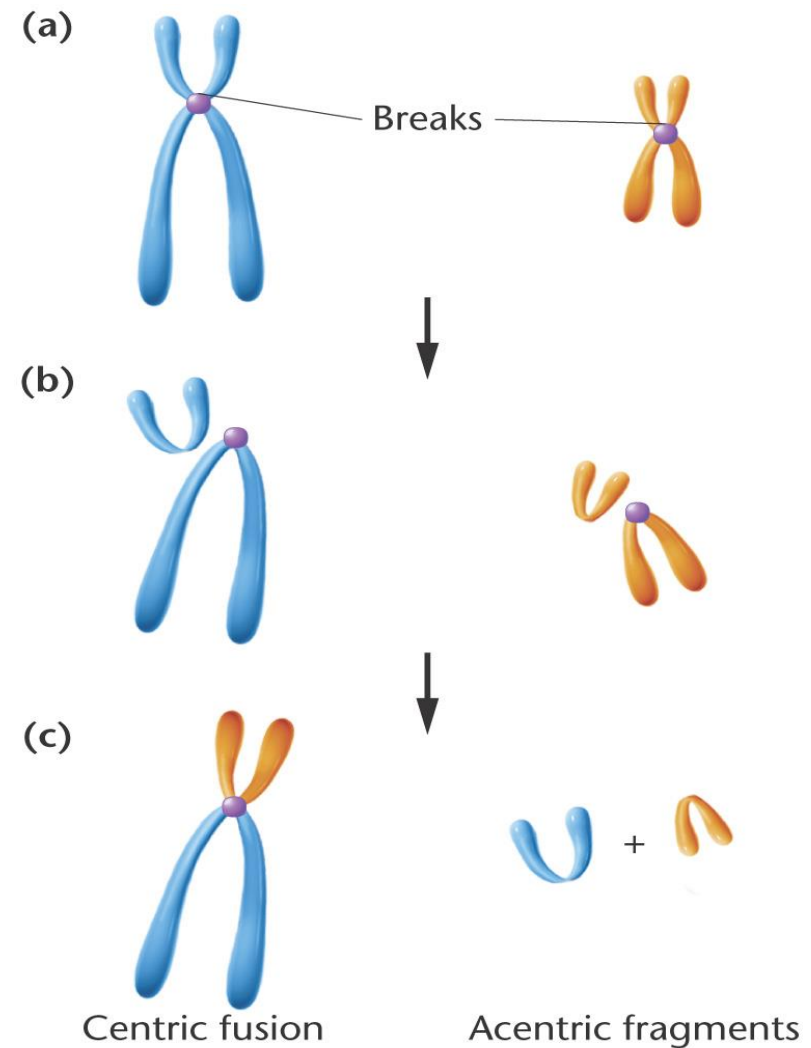
b) Nonreciprocal interchromosomal translocation



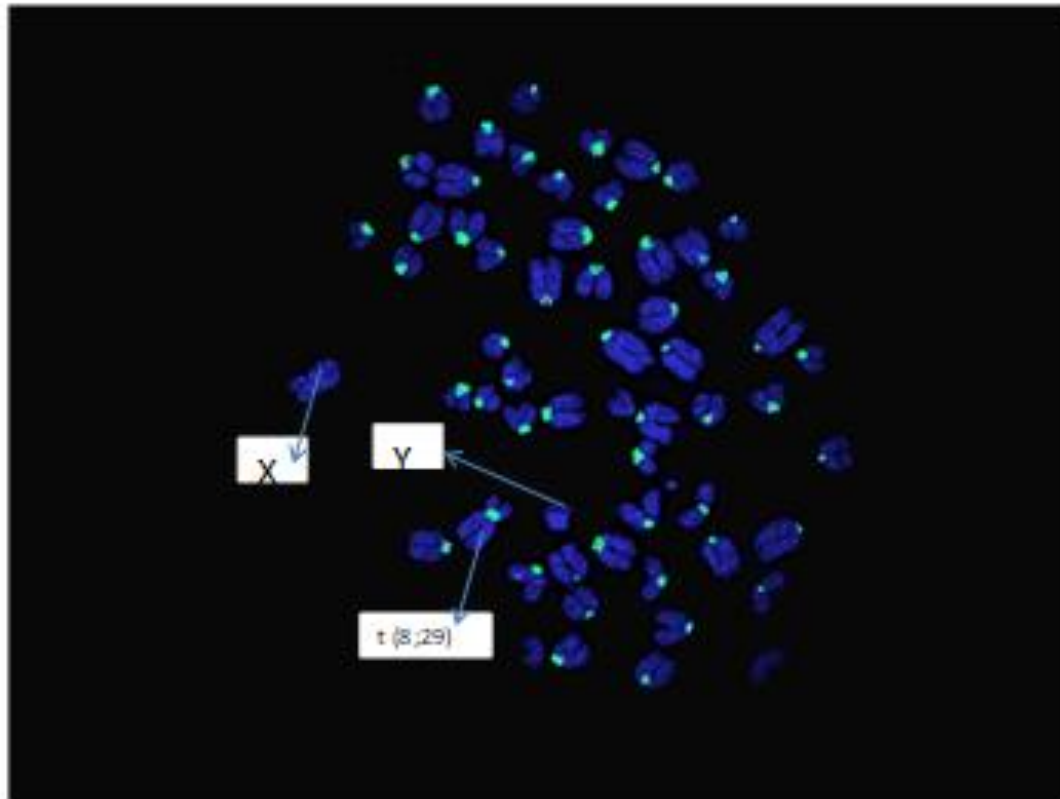
c) Reciprocal interchromosomal translocation



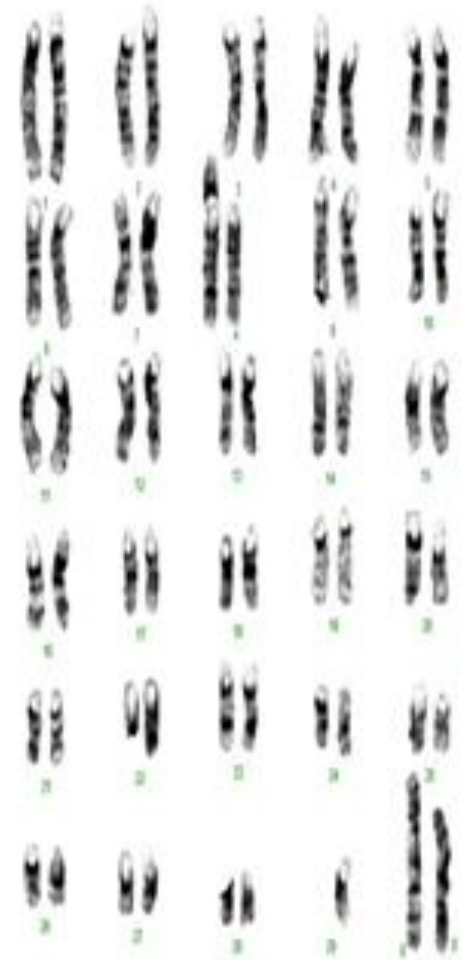
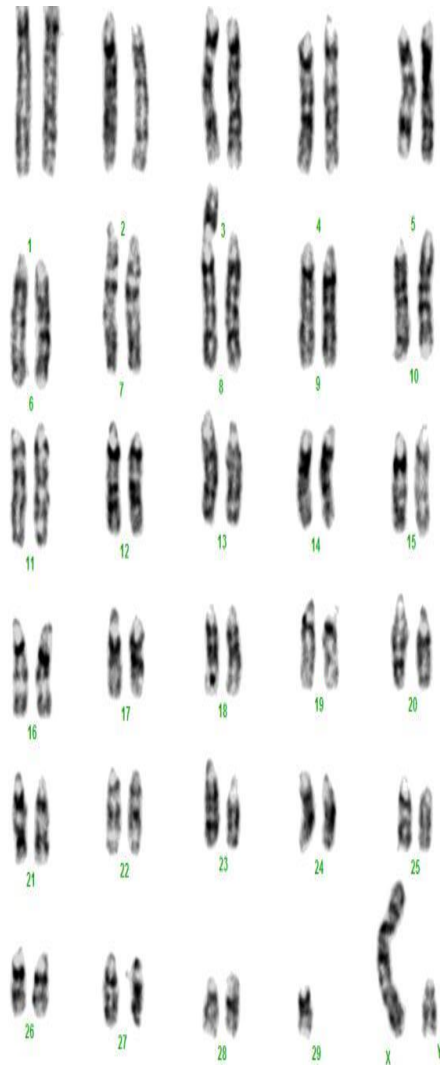
# Familial Down Syndrome



## Fluorescent In-situ Hybridization

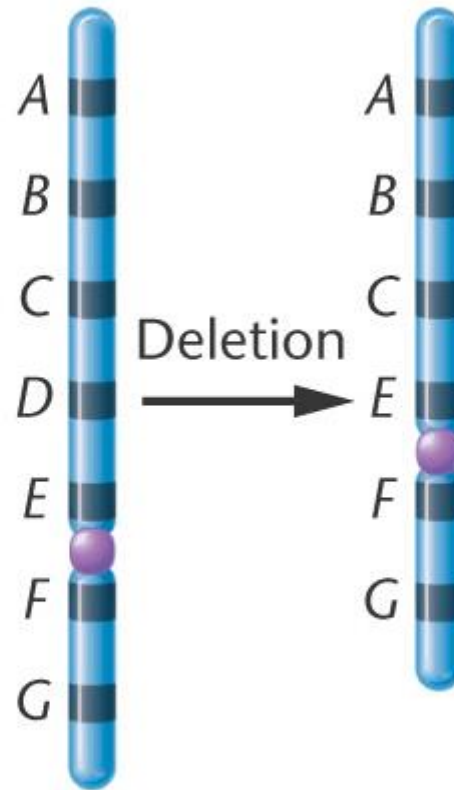


GTG- banded Karyotype of native male cattle showing 8;29 Robertsonian translocation

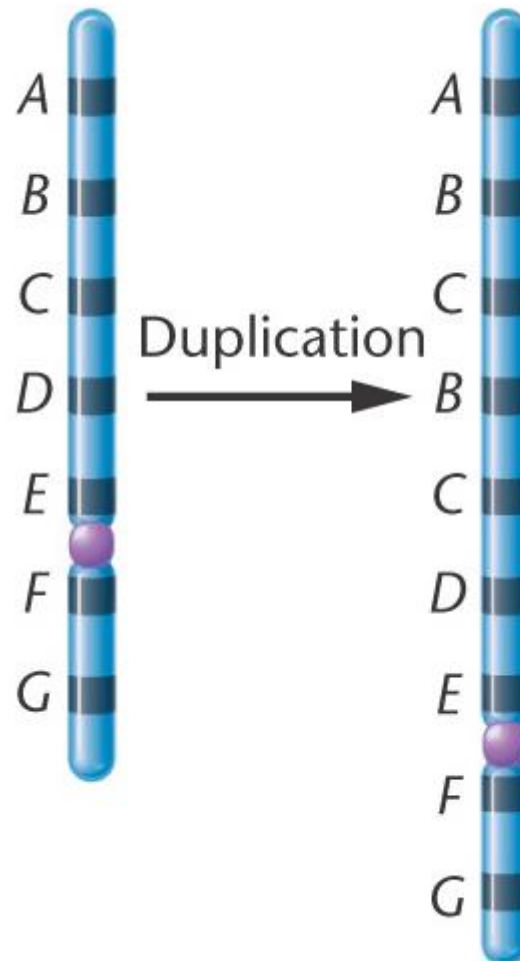


GTG- banded Karyotype of native female cattle showing 8;29 Robertsonian translocation

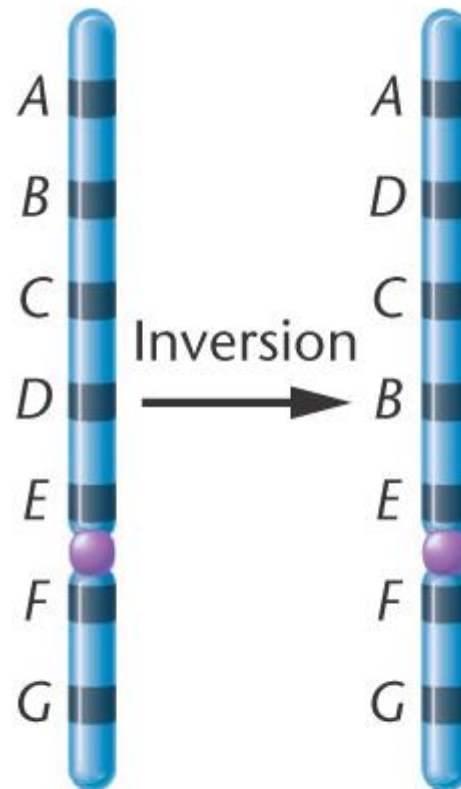
### (a) Deletion of *D*



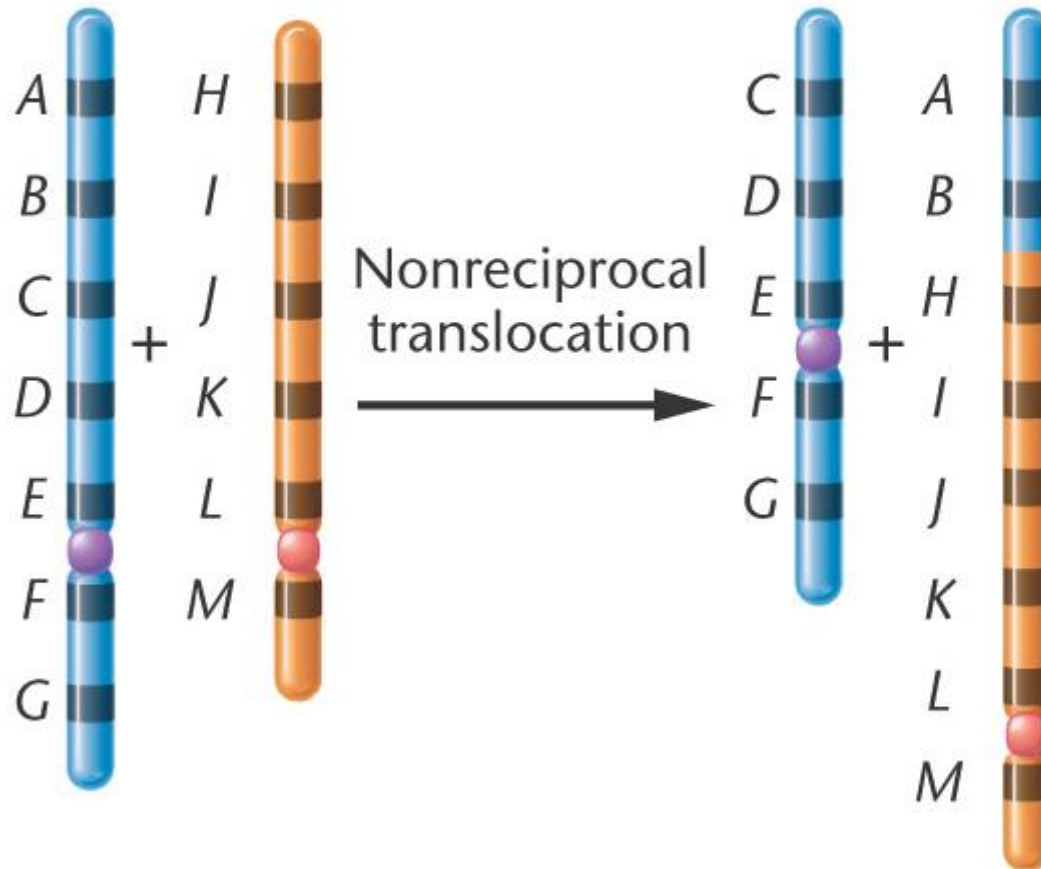
## (b) Duplication of *BC*



### (c) Inversion of *BCD*



**(d) Nonreciprocal translocation of A–B**





(e) Reciprocal translocation of *A-B* and *H-I-J*

