

Chromosomal Aberrations

Types of Chromosome

Metacentric:

Telocentric:

Acrocentric:

Sub-metacentric:

Variations in Chromosome Number and Arrangement

Terminology

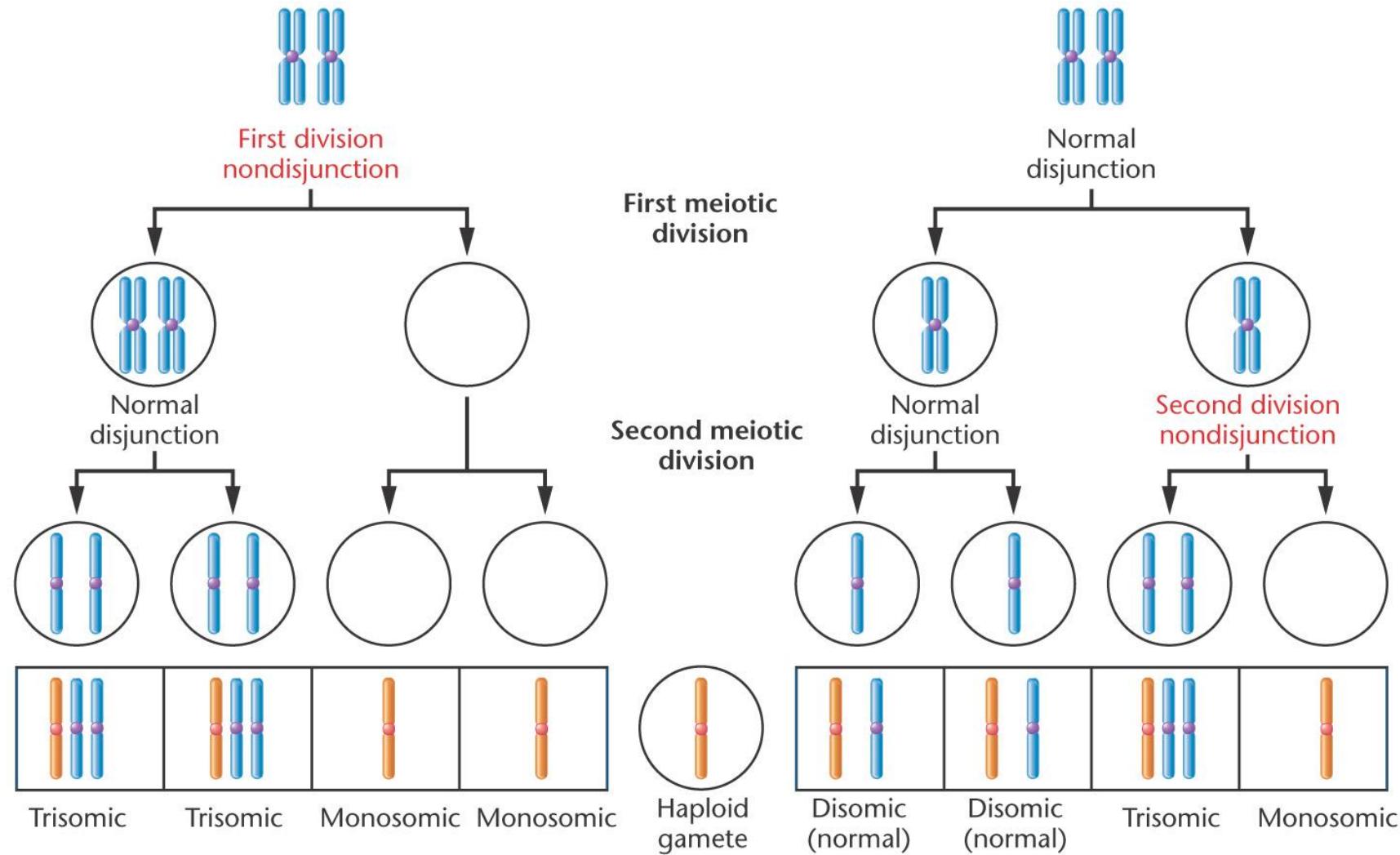
different

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	$2n$
Polyplosity	$3n, 4n, 5n, \dots$
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

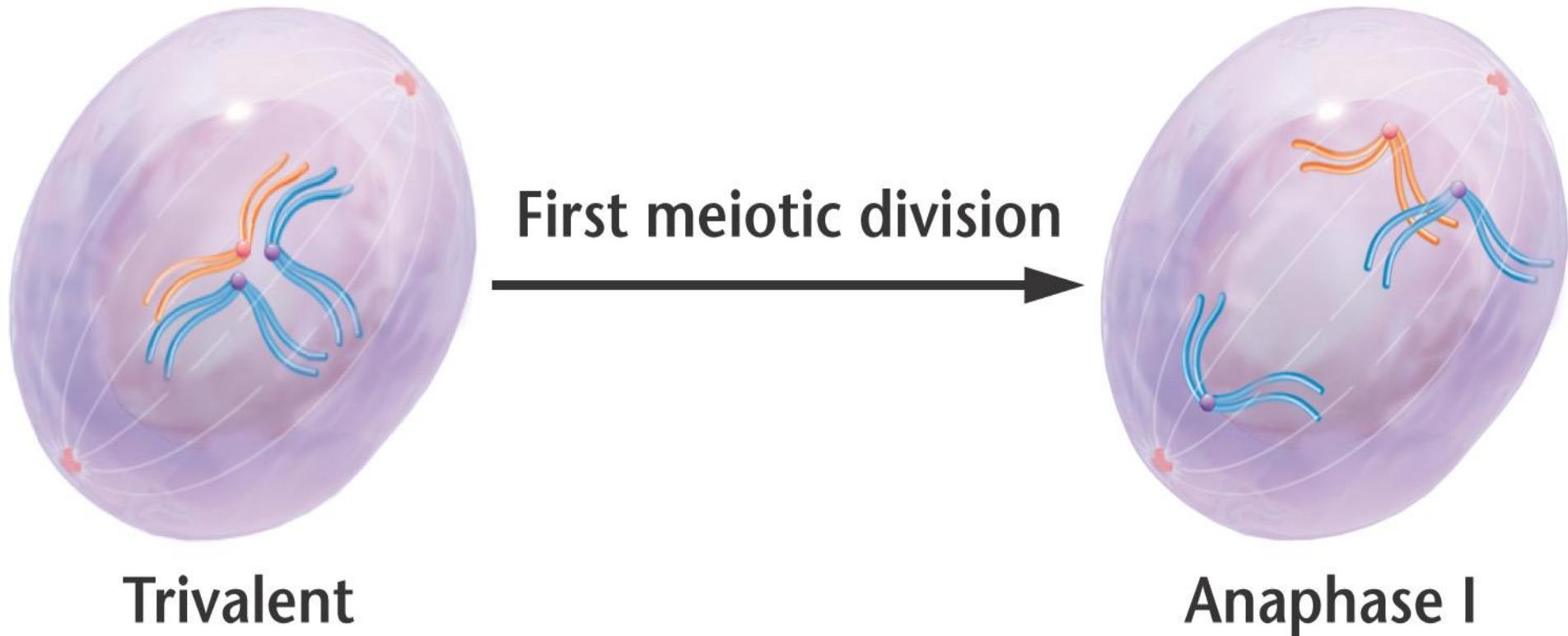
	W	Y
w	Ww (Red eyed female)	wY (White eyed Male)
ww	Www Dies	wwY White eyed female
(-)	W- Red eyed Male (Sterile)	-Y Dies

	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

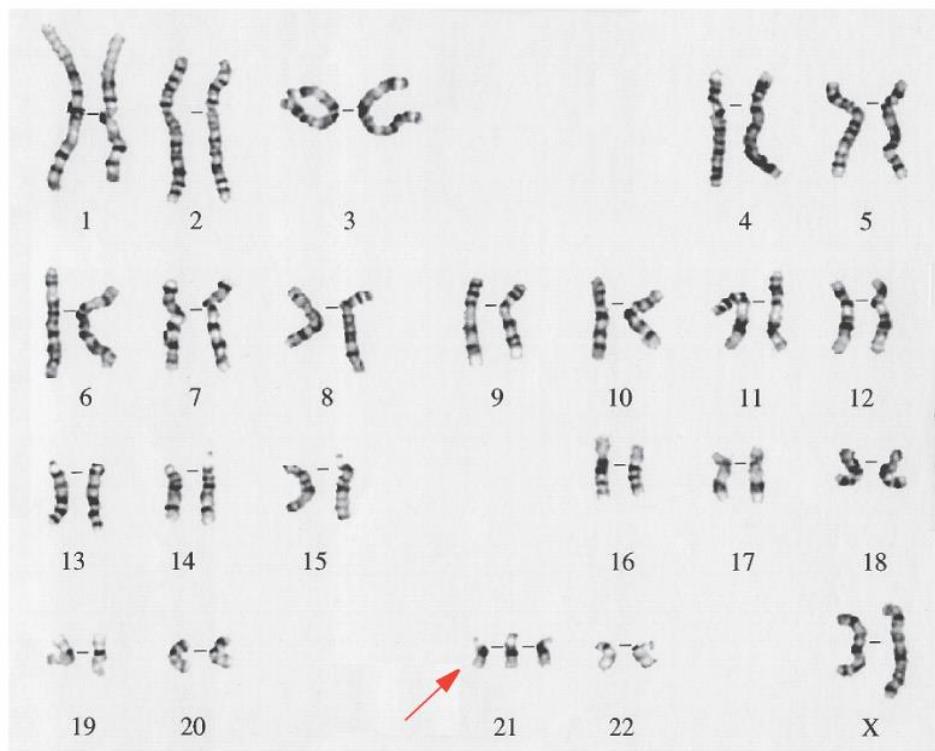
Trisomy Meiosis



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Down Syndrome

Down Syndrome – Trisomy 21



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Polyplody in Plants

Autopolyplody

Allopolyploidy

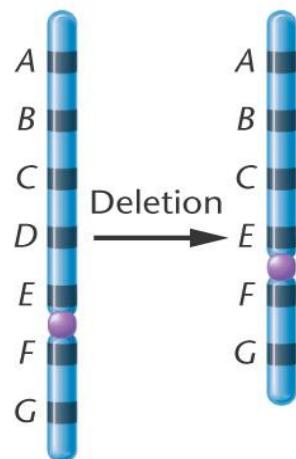
Allotetraploids



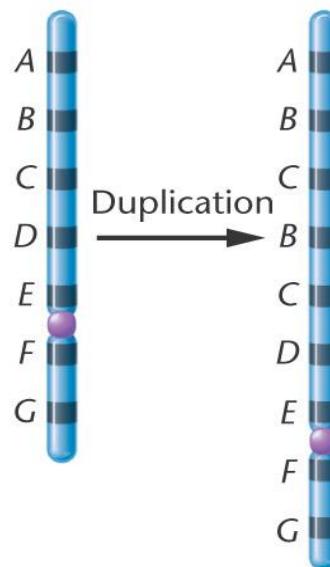
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Chromosome Rearrangements

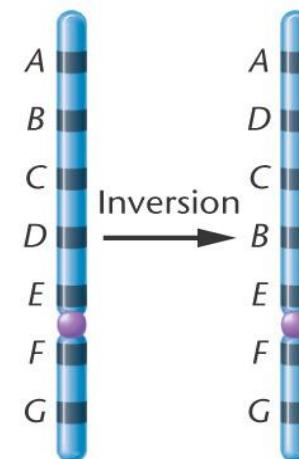
(a) Deletion of D



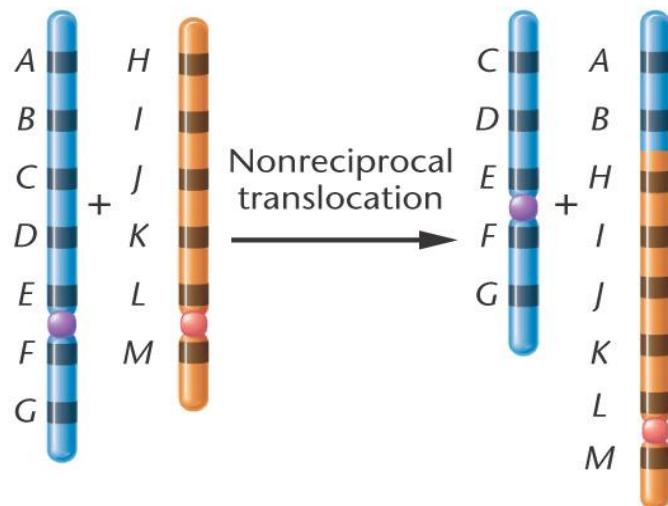
(b) Duplication of BC



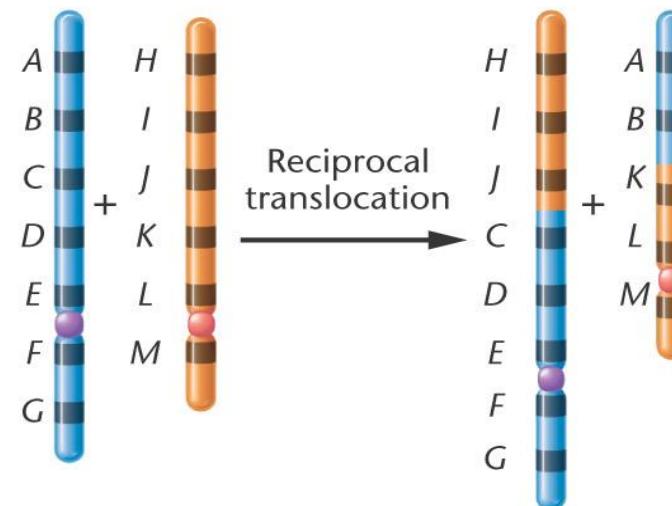
(c) Inversion of BCD



(d) Nonreciprocal translocation of A-B



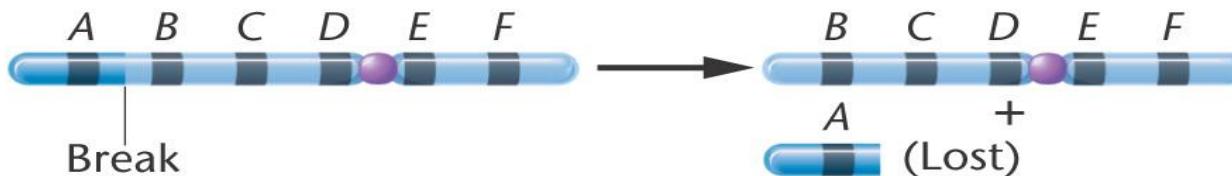
(e) Reciprocal translocation of A-B and H-I-J



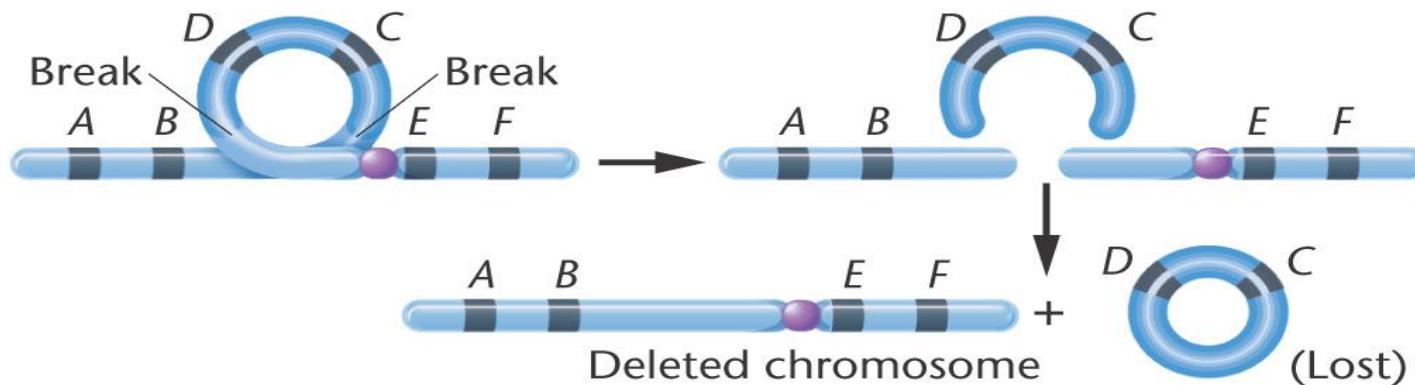
Consequences of Rearrangements

Deletions

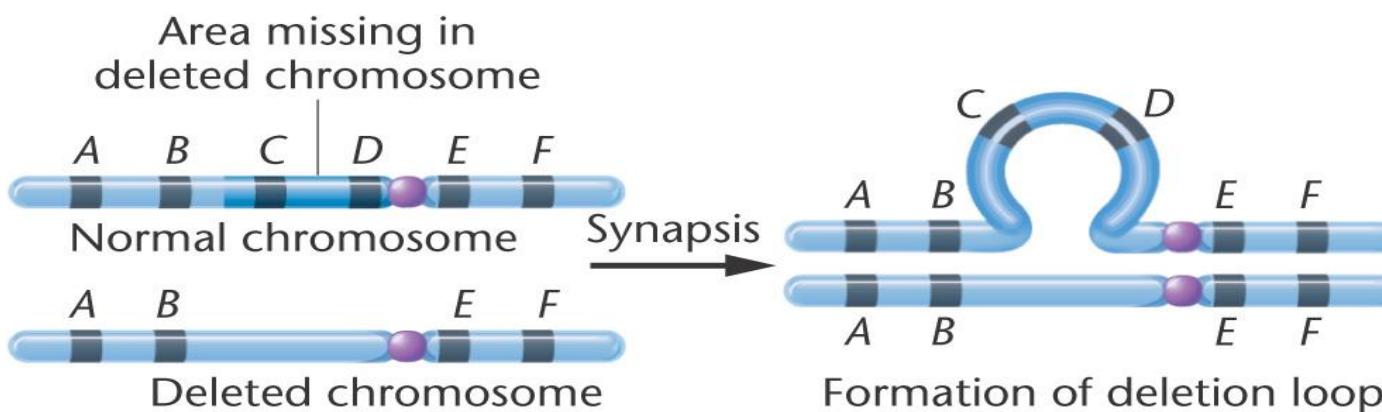
(a) Origin of terminal deletion



(b) Origin of intercalary deletion

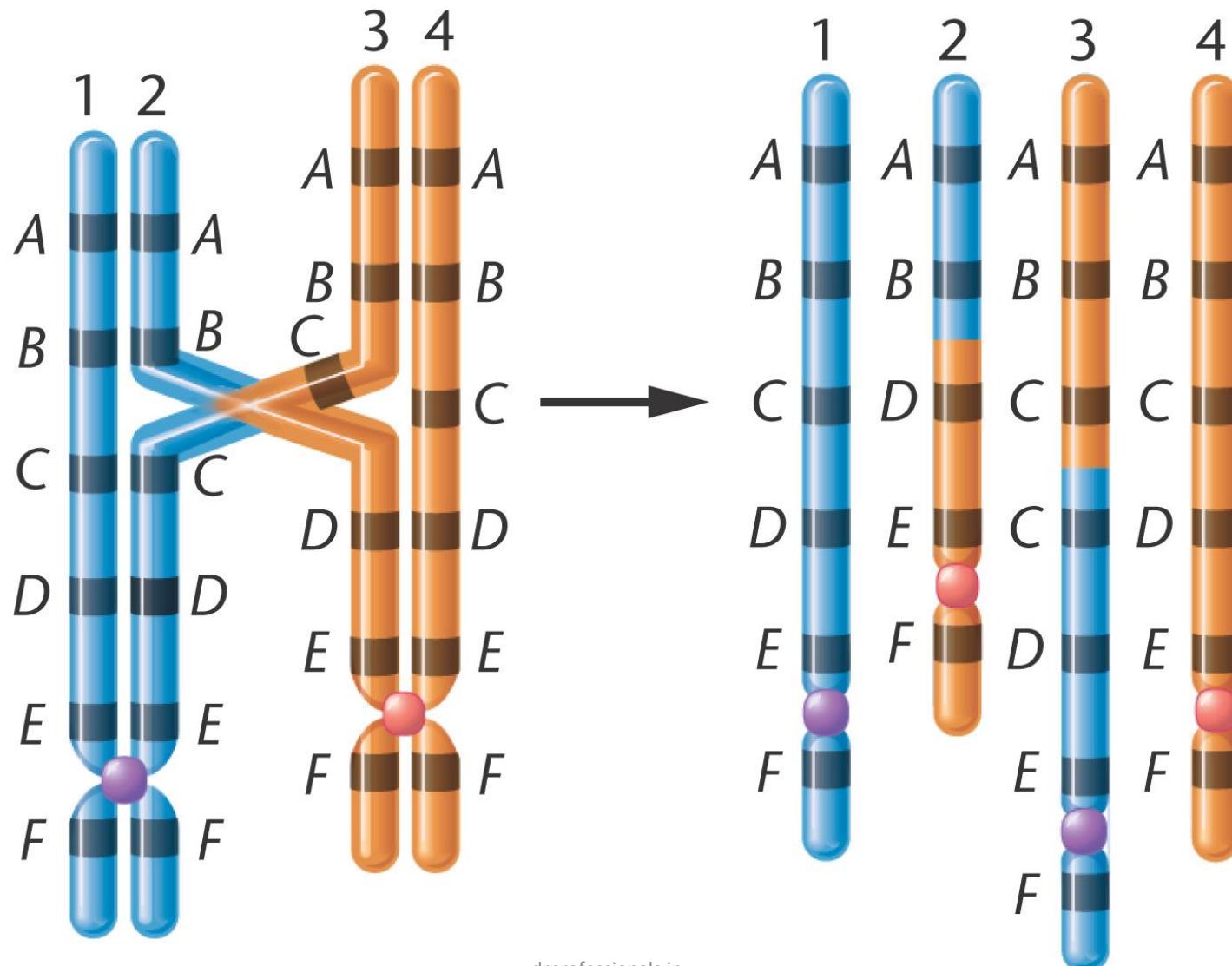


(c) Formation of deficiency loop



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		 

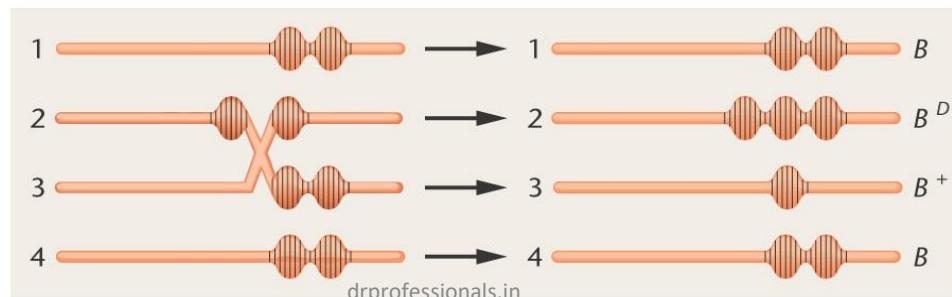


B^+/B^+



B/B^+

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

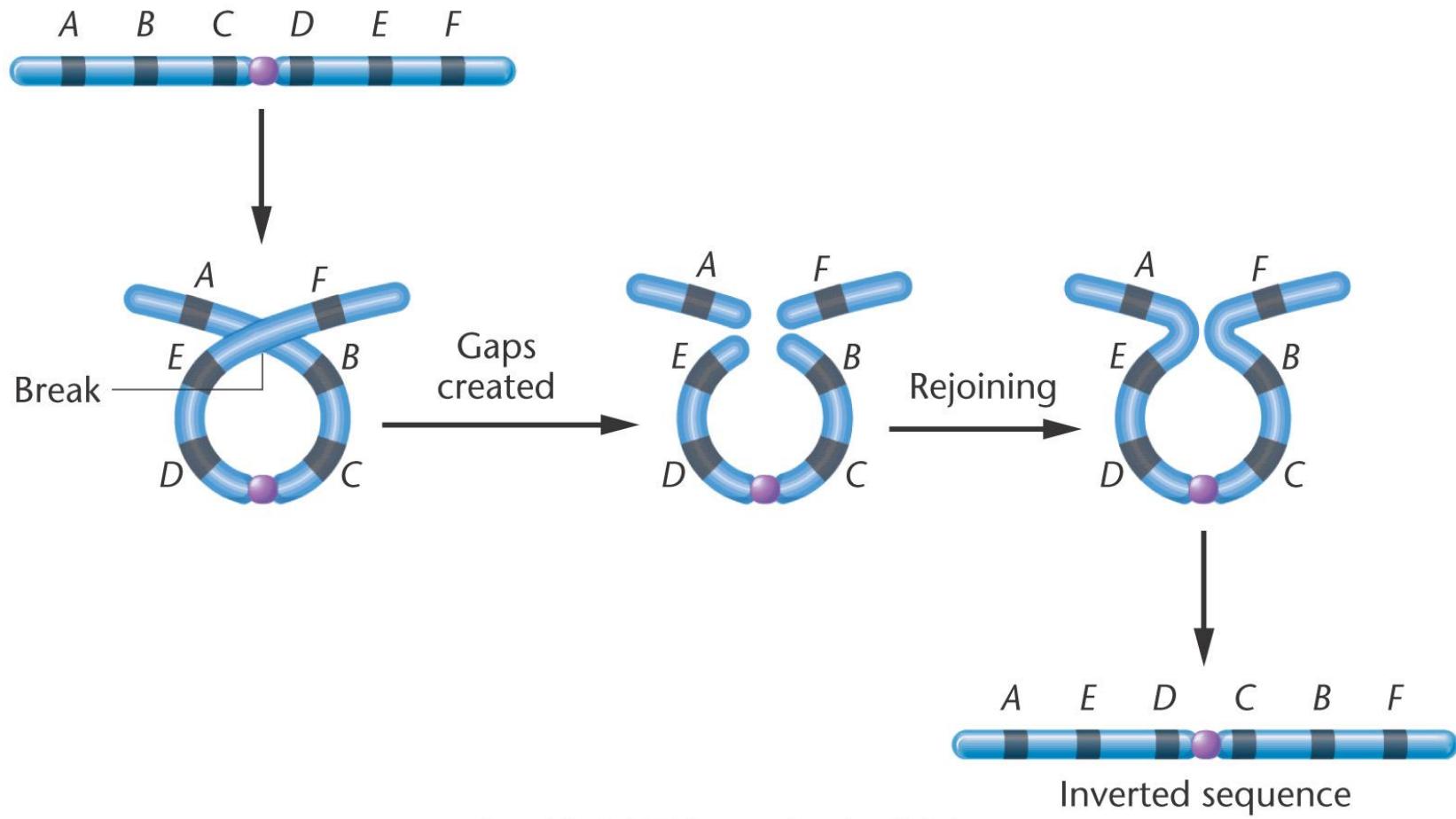


B/B

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

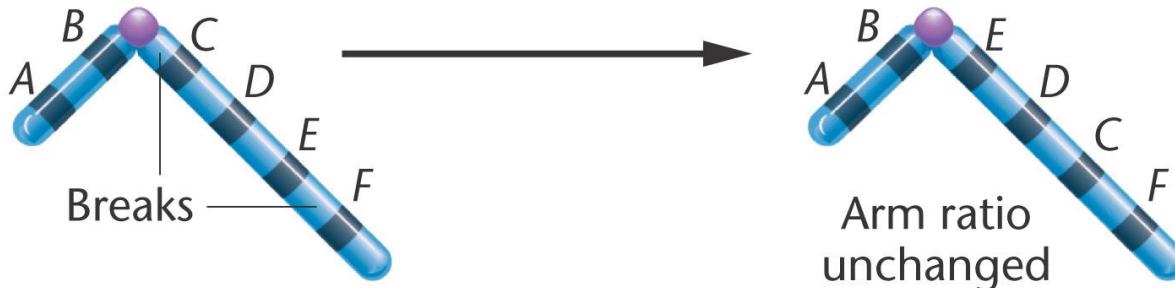
Chromosomal Inversions



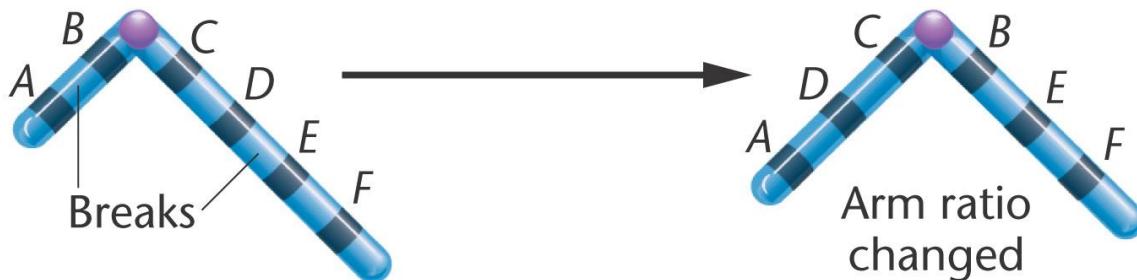
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Chromosomal Inversions

Paracentric inversion



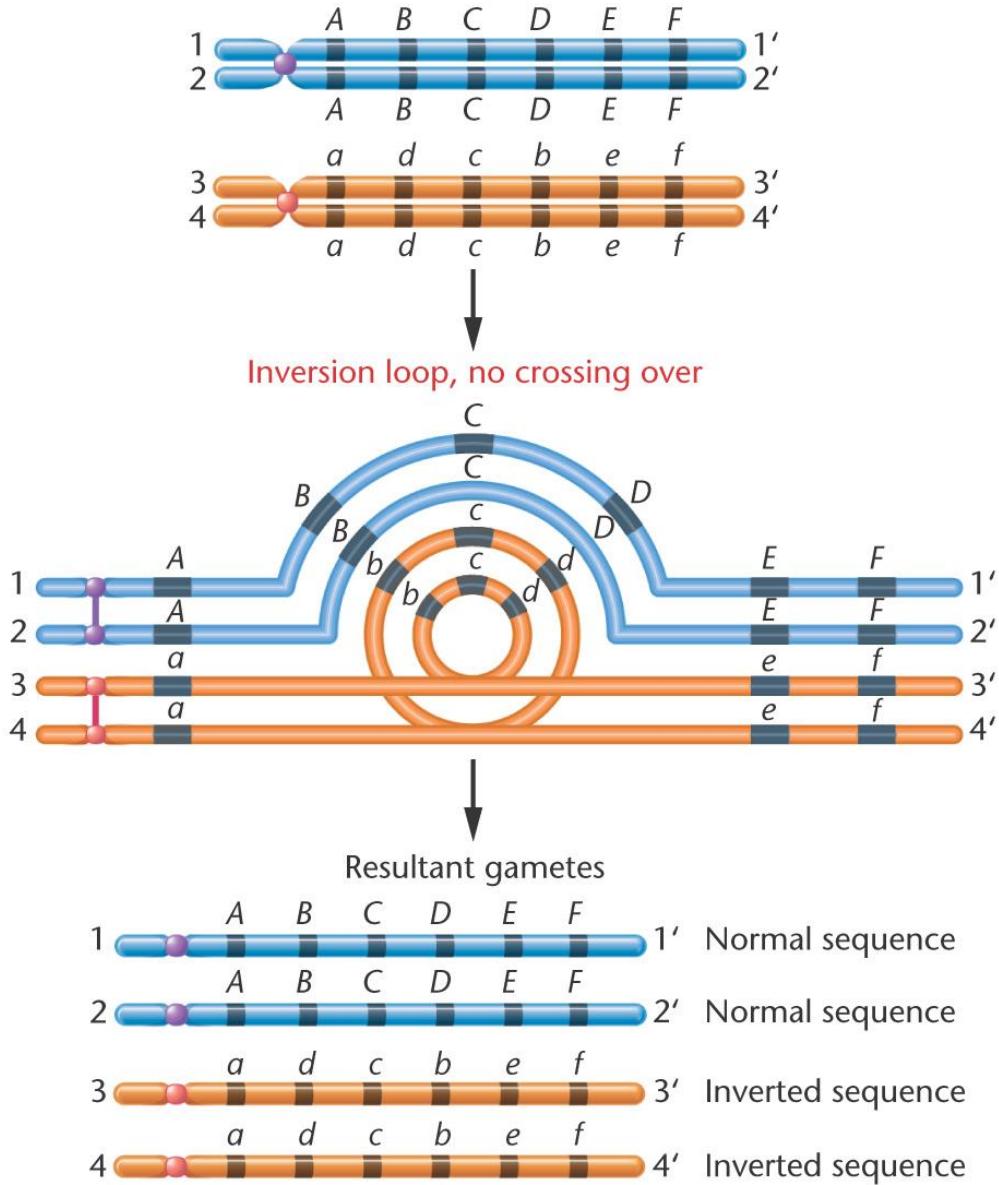
Pericentric inversion



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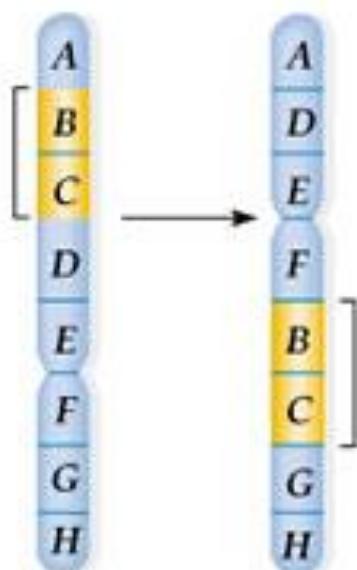
Inversions and Gametogenesis

Paracentric inversion heterozygote

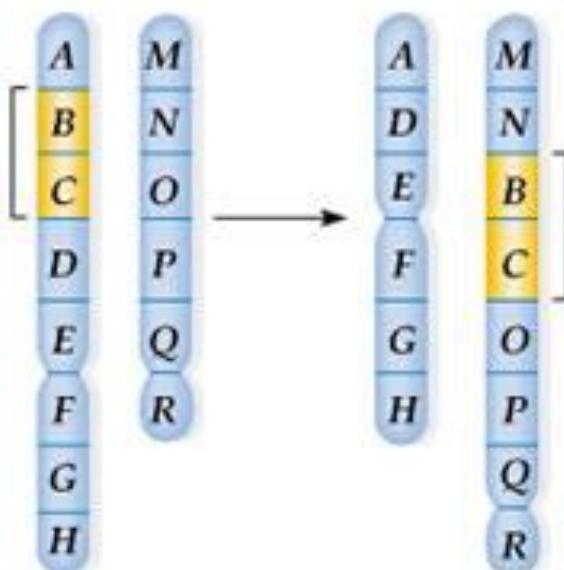


Translocations

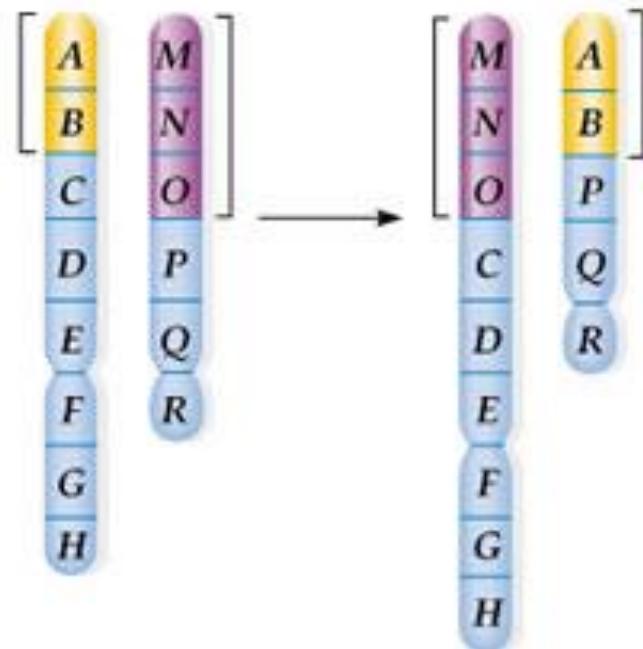
a) Nonreciprocal intrachromosomal translocation



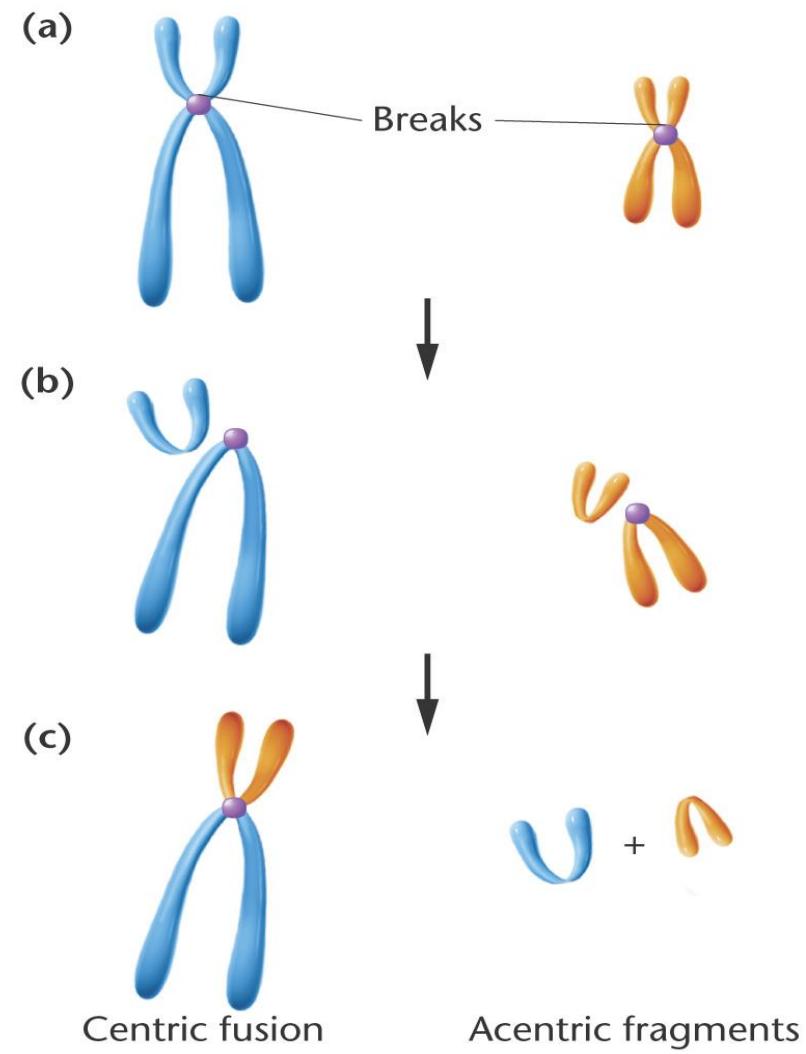
b) Nonreciprocal interchromosomal translocation



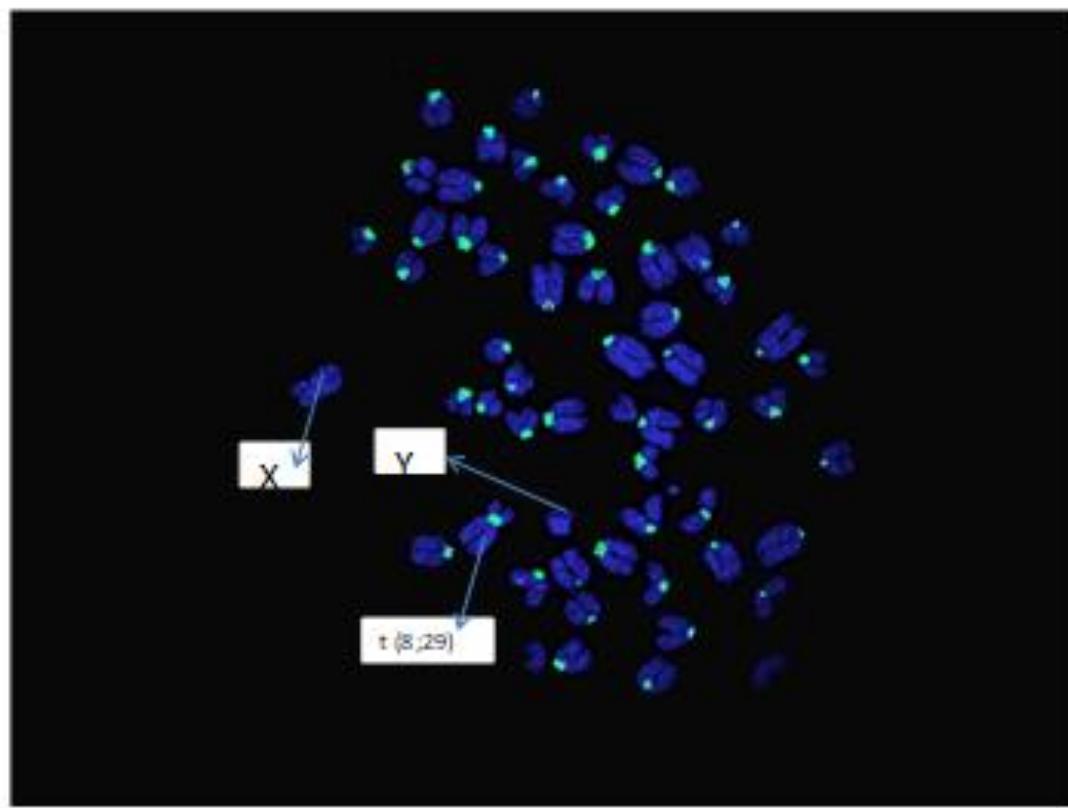
c) Reciprocal interchromosomal translocation



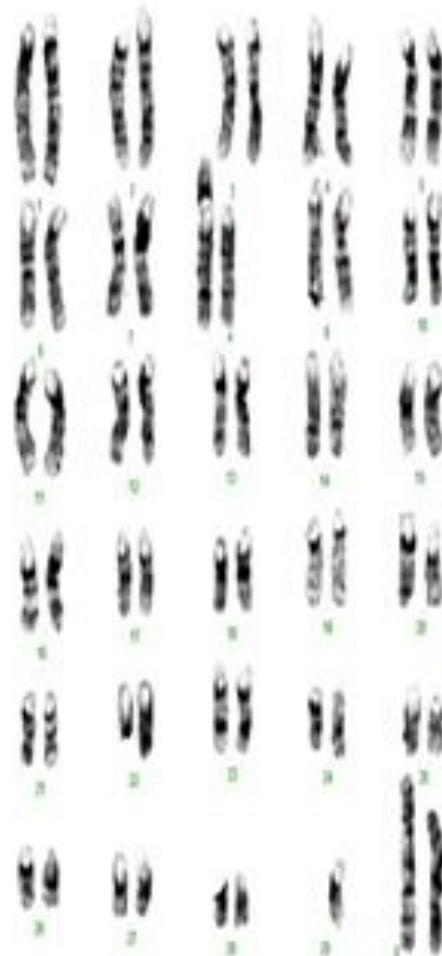
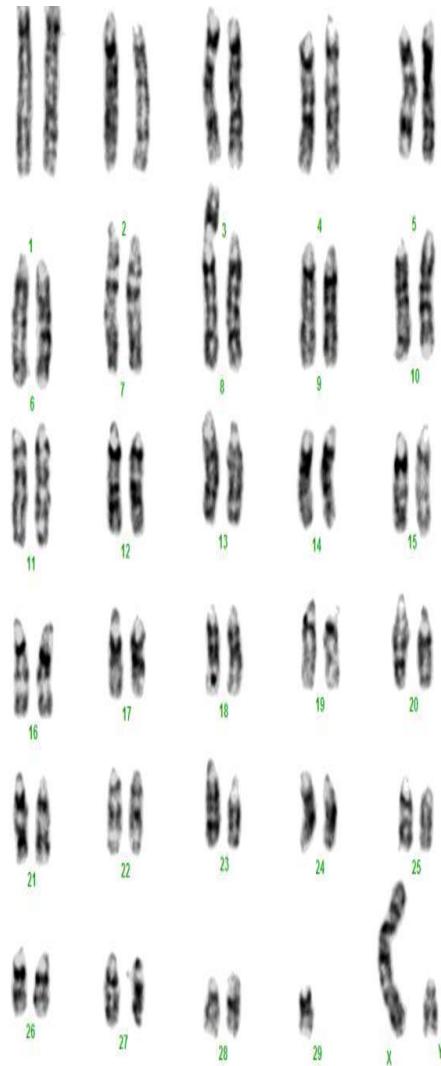
Familial Down Syndrome



Fluorescent In-situ Hybridization

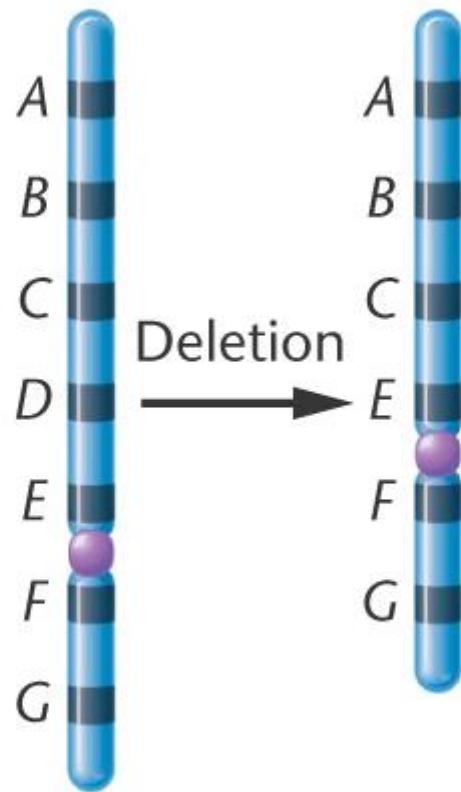


GTG-banded Karyotype of native male cattle showing 8;29 Robertsonian translocation

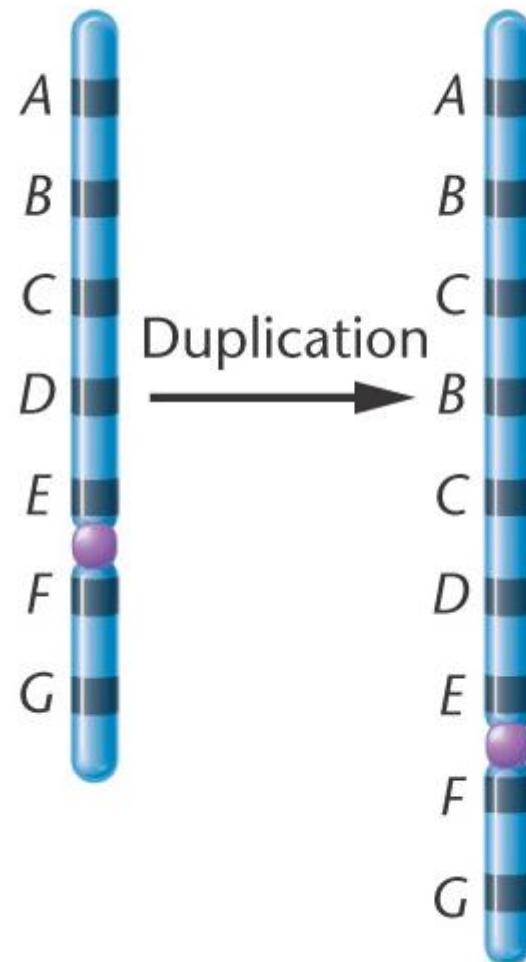


GTG-banded Karyotype of native female cattle showing 8;29
Robertsonian translocation

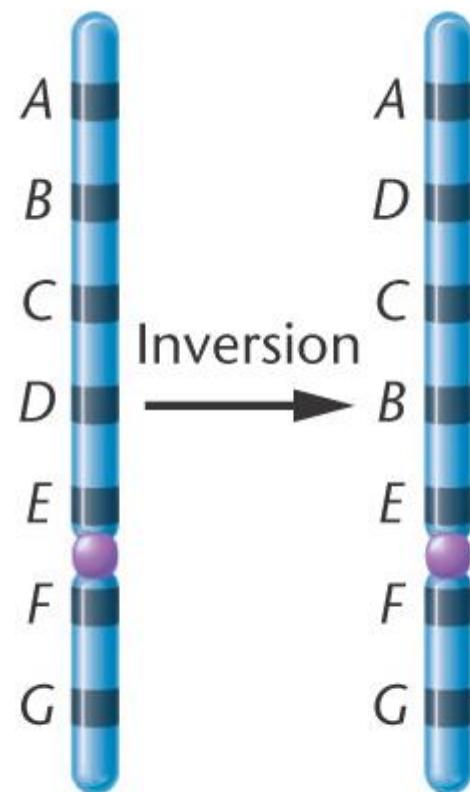
(a) Deletion of D



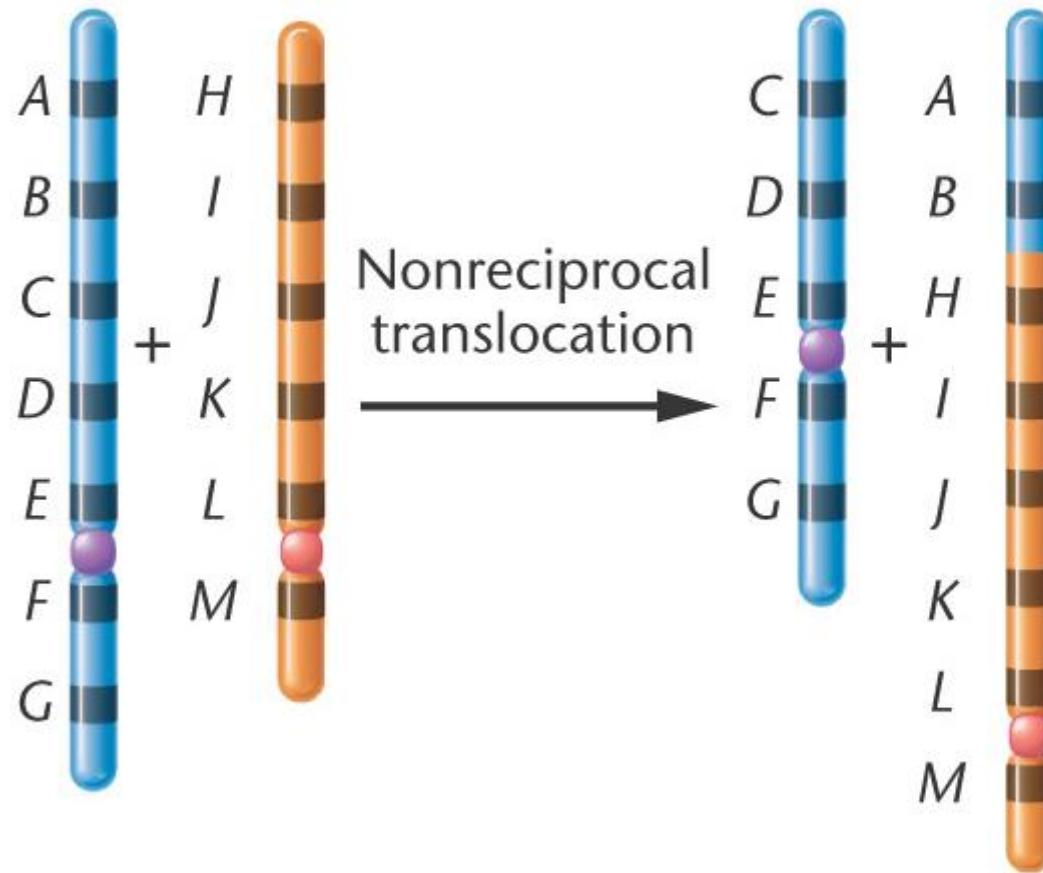
(b) Duplication of BC



(c) Inversion of *BCD*



(d) Nonreciprocal translocation of A–B



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

