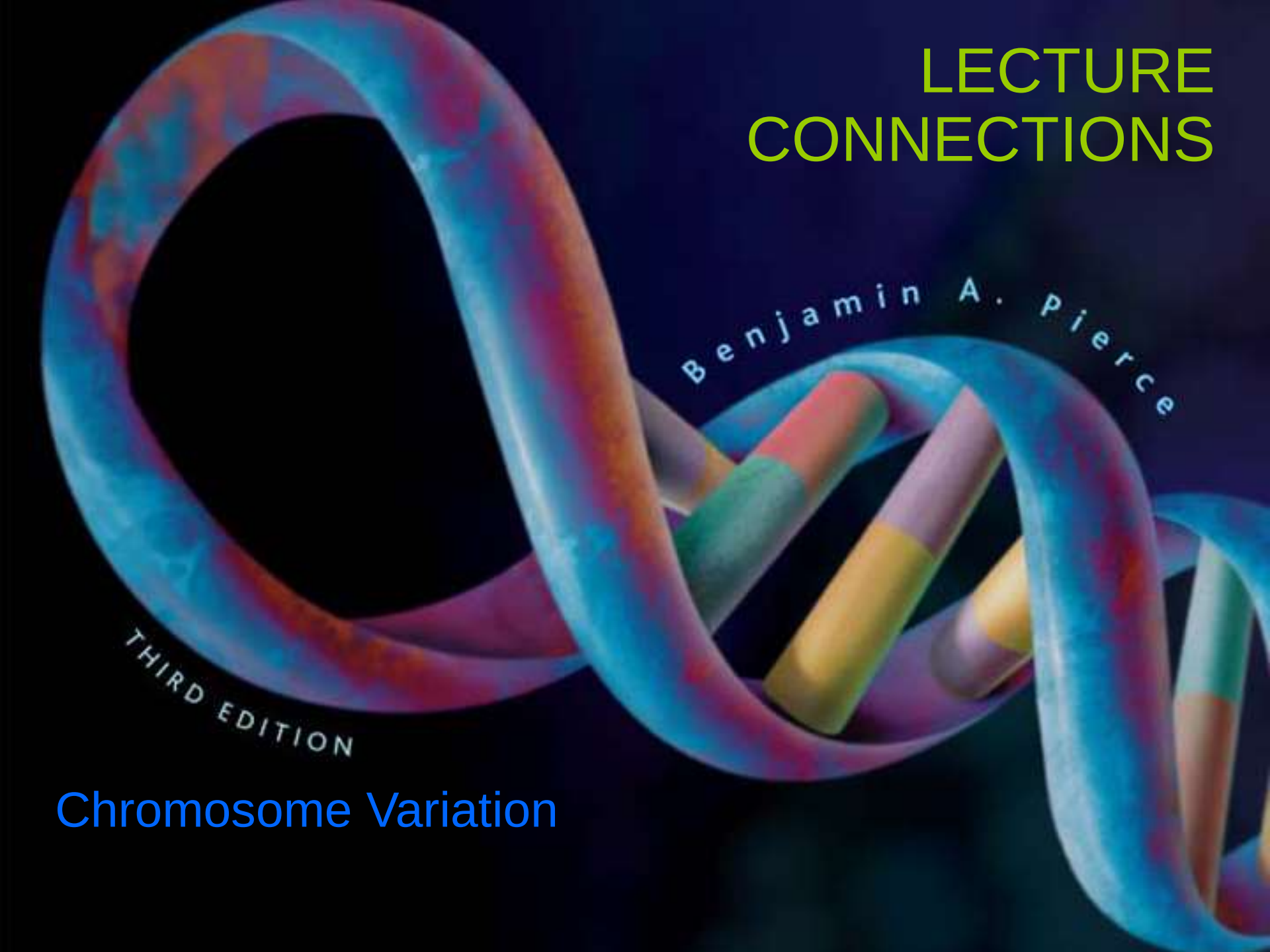
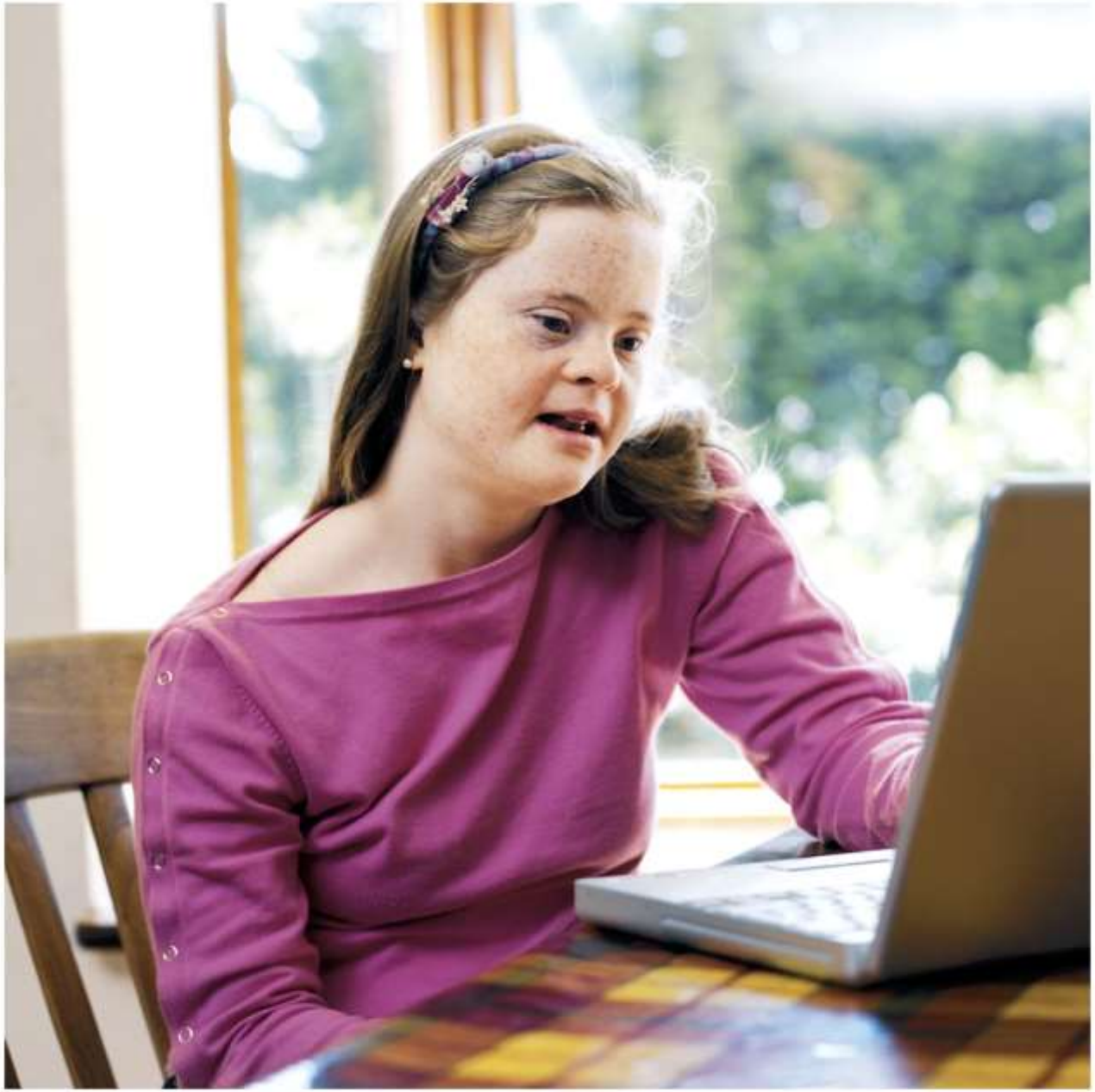

LECTURE
CONNECTIONS
Benjamin A. Pierce
THIRD EDITION
Chromosome Variation

Outline

- Chromosome Mutations Include Rearrangements, Aneuploids, and Polyploids, 238
- Chromosome Rearrangements Alter Chromosome Structure, 240
- Aneuploidy Is an Increase or Decrease in the Number of Individual Chromosomes, 249
- Polyploidy Is the Presence of More than Two Sets of Chromosomes, 255
- Chromosome Variation Plays an Important Role in Evolution, 260



Chapter 9 Opener
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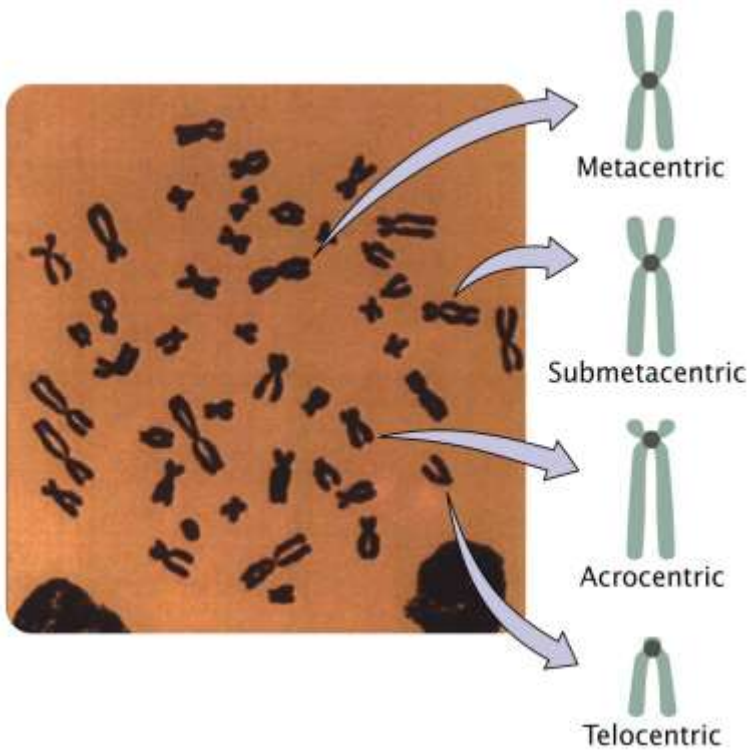
Chromosome Mutations Include Rearrangements, Aneuploids, and Polyploids

- Chromosome Morphology (position of the centromere on the chromosome):
  - Metacentric
  - Submetacentric
  - Acrocentric
  - Telocentric



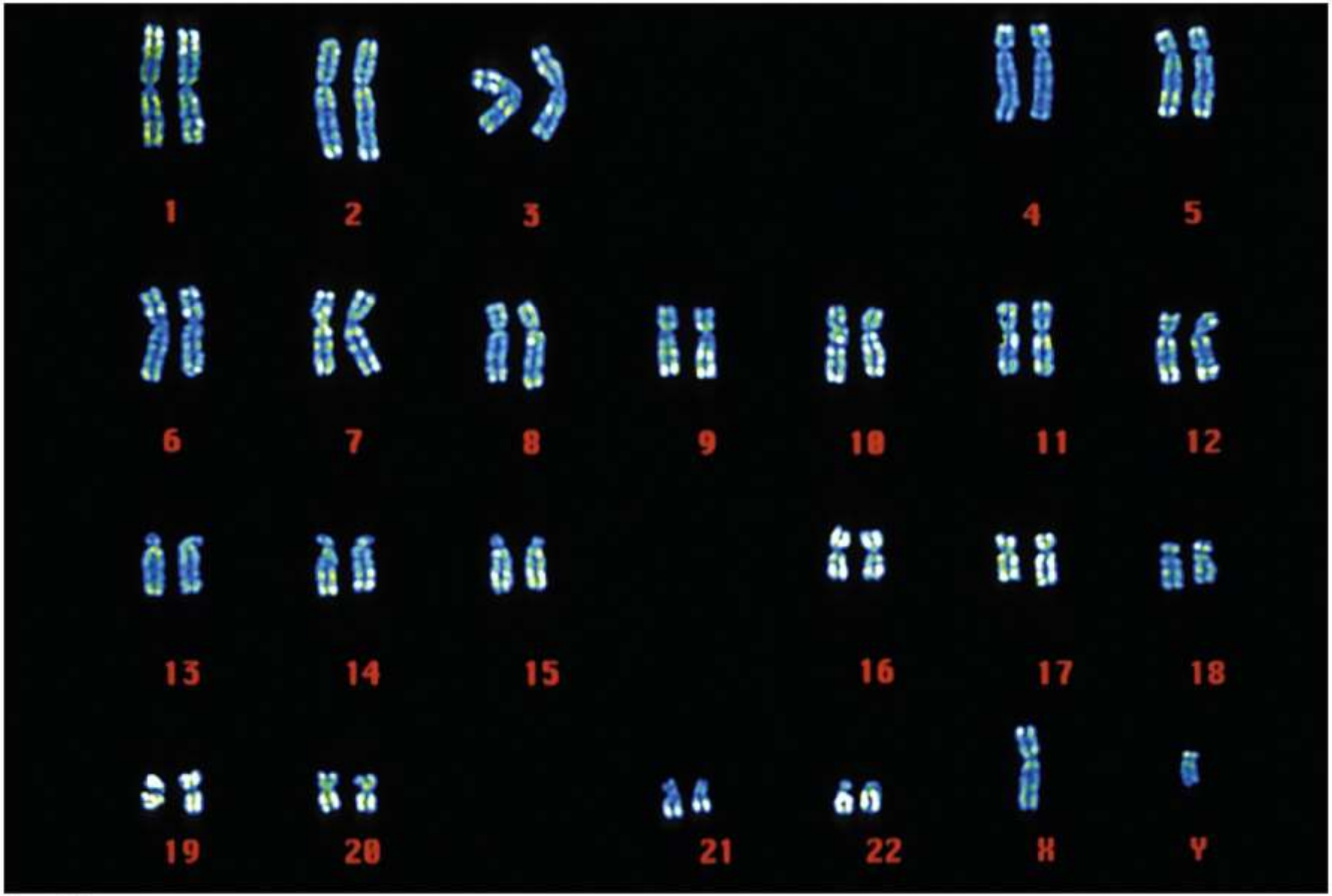


Figure 9-1
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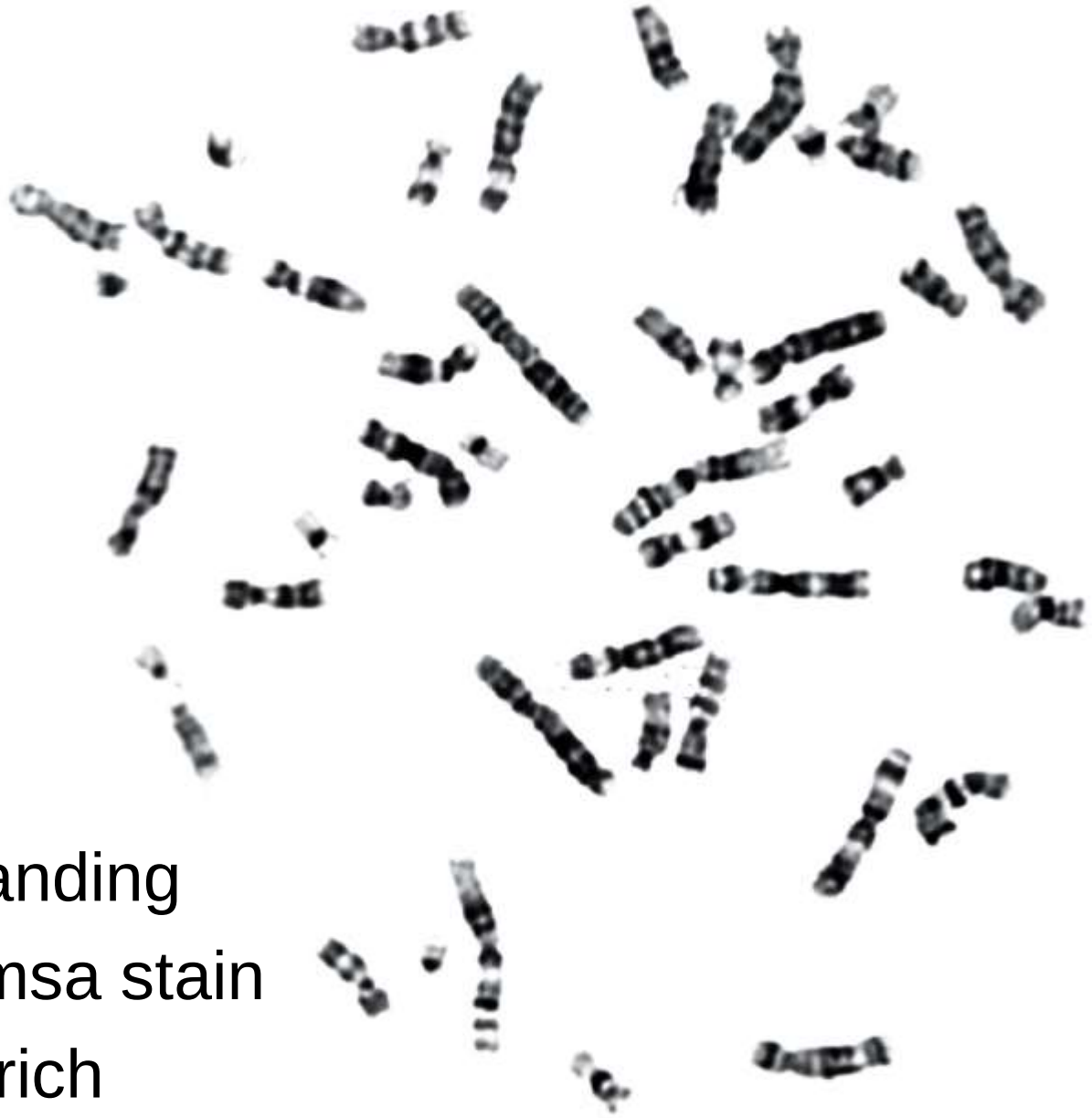
Human Karyotype



G banding

Giemsa stain

A-T rich

Figure 9-2a
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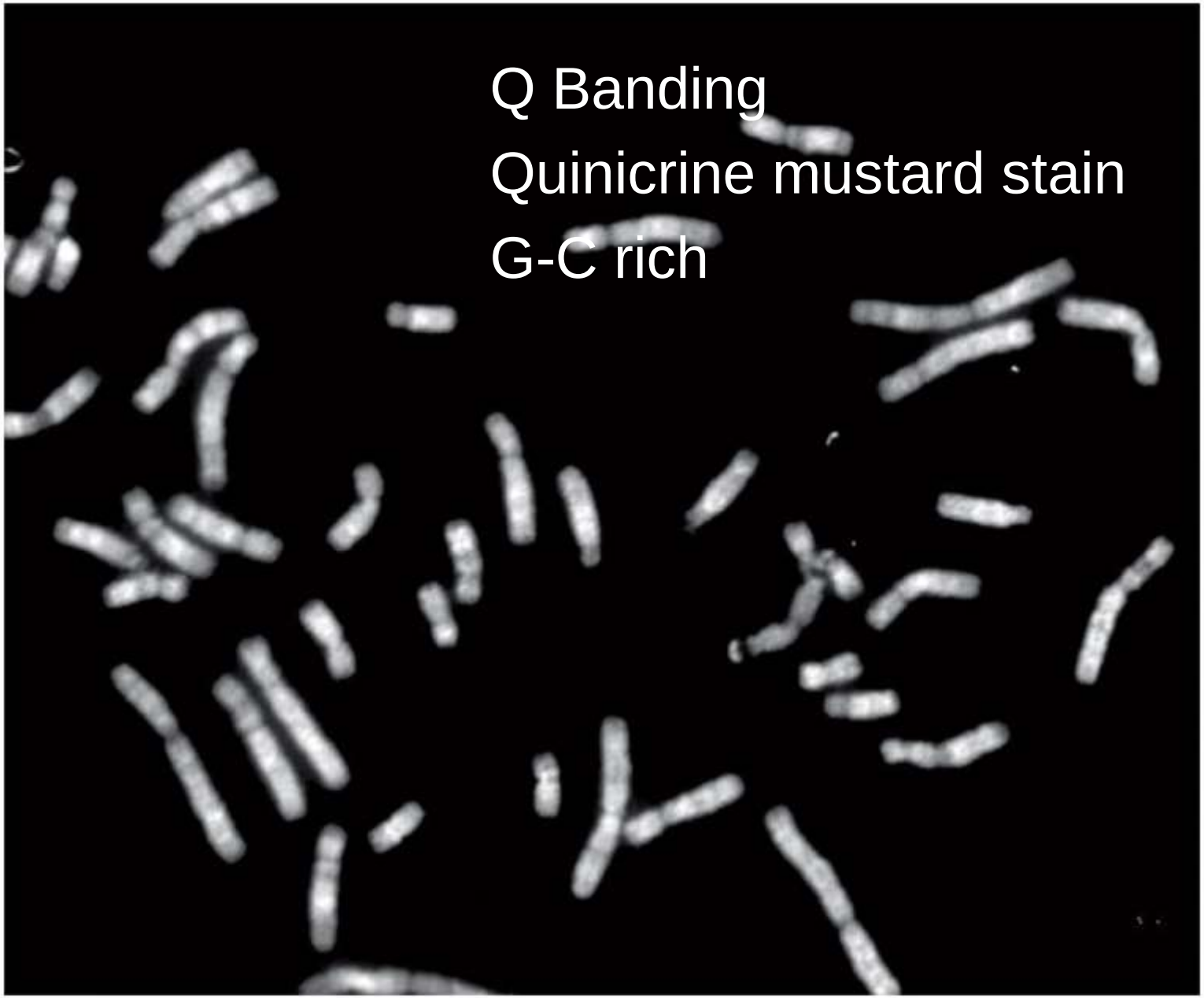

Figure 9-2b
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C Banding centromeres

Figure 9-2c
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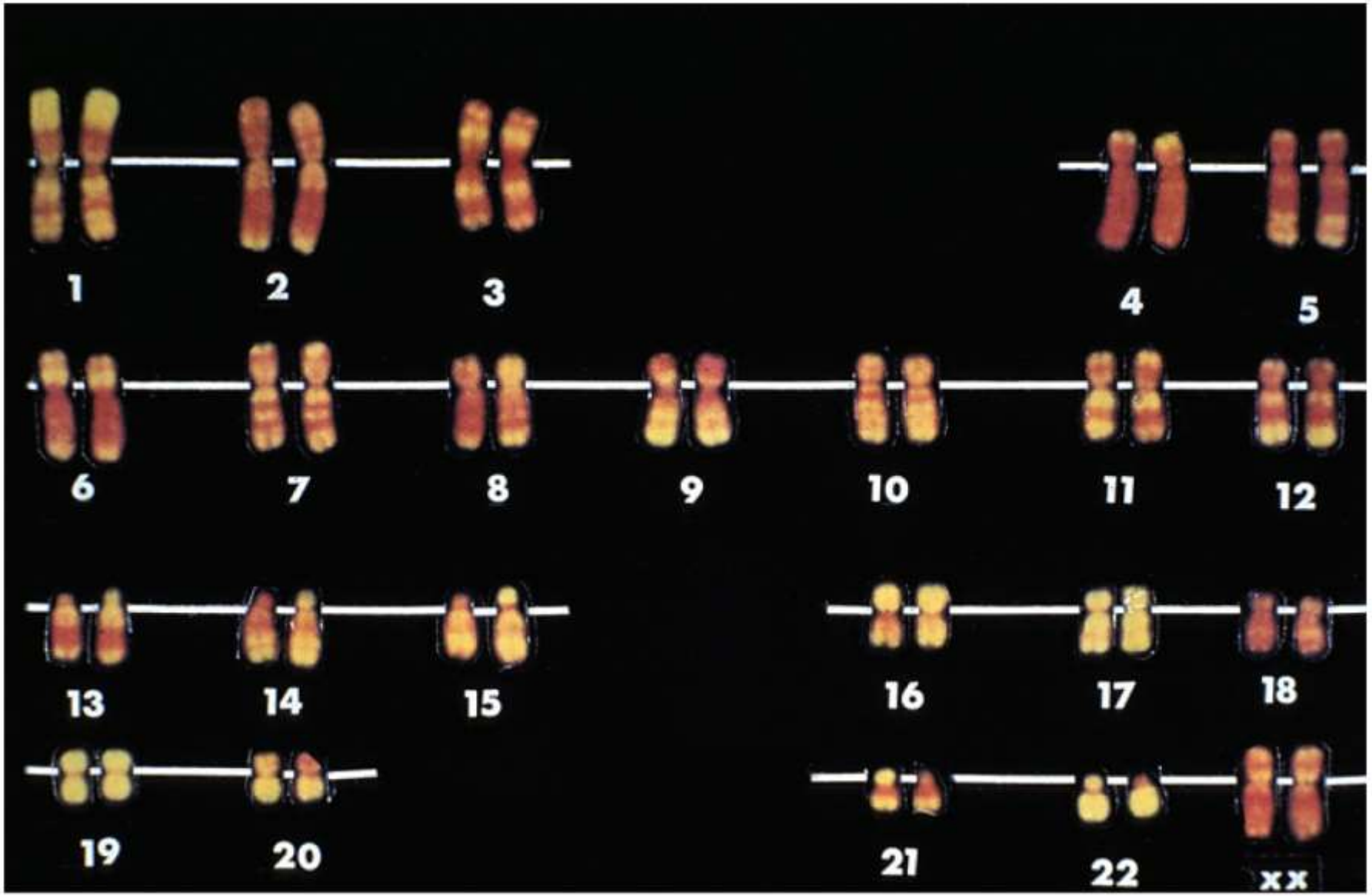

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R Banding C-G rich

Types of Chromosome Mutations

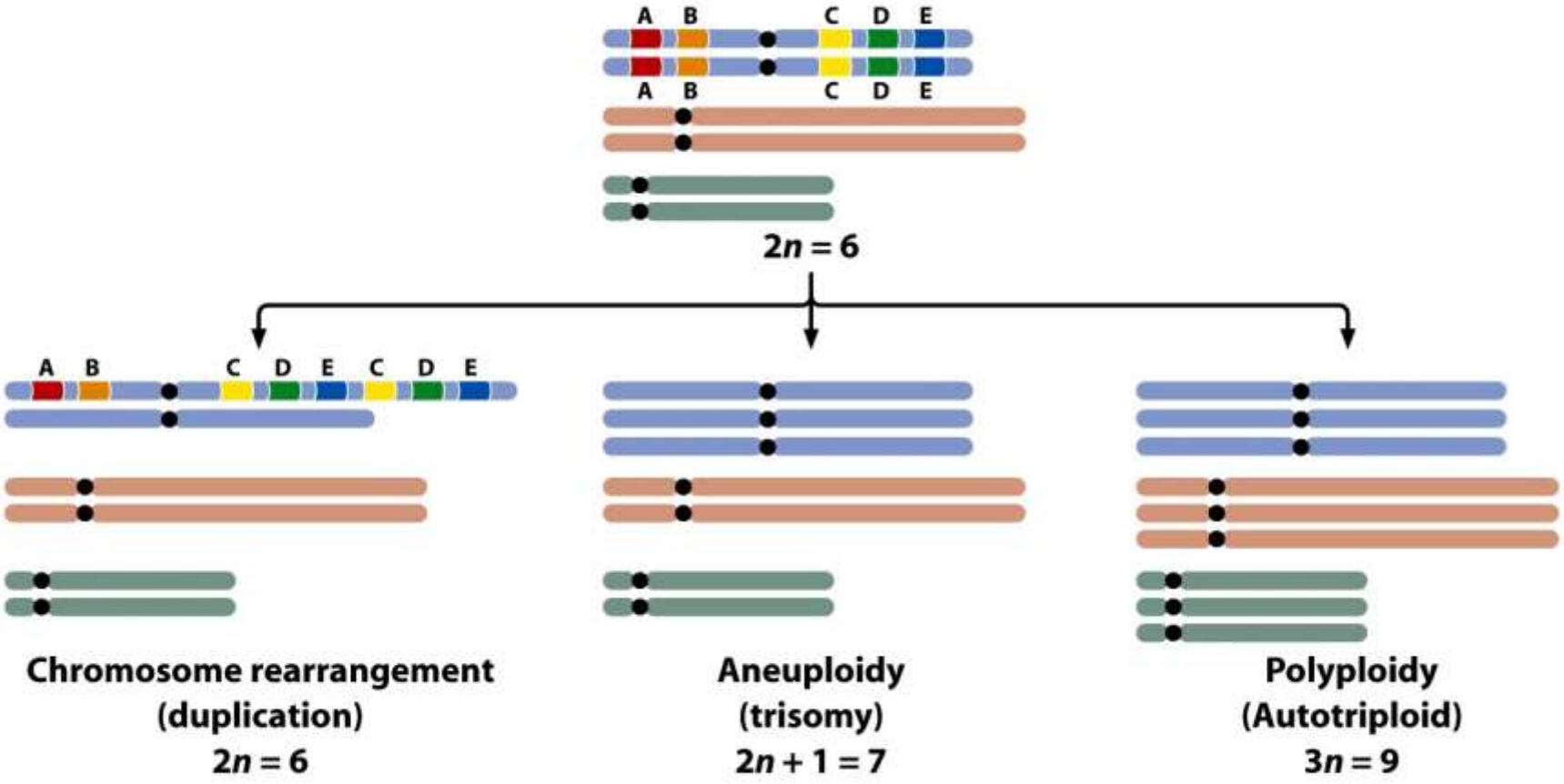

Figure 9-3
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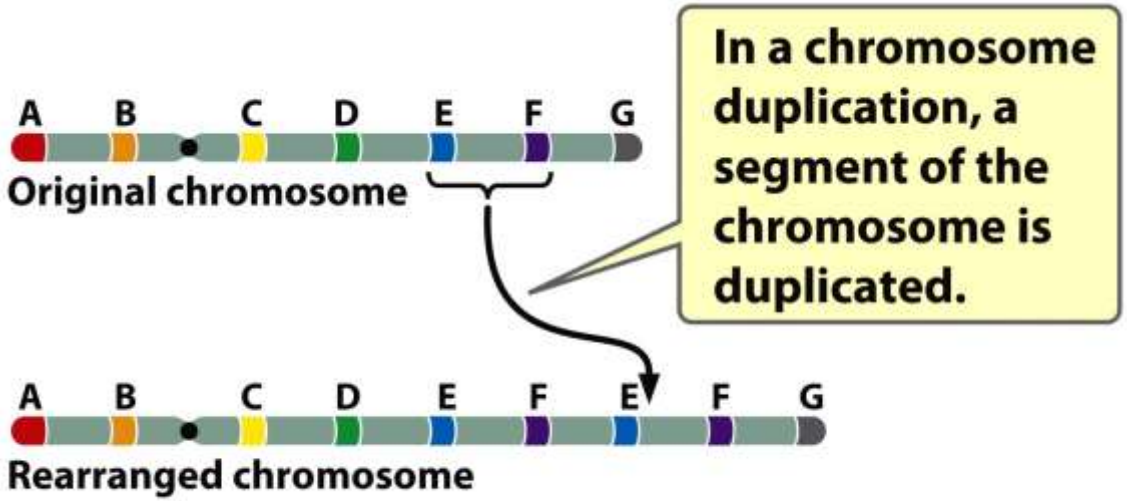
Chromosome Rearrangements Alter Chromosome Structure

- **Duplication**
  - Tandem
  - Reverse
  - Displaced



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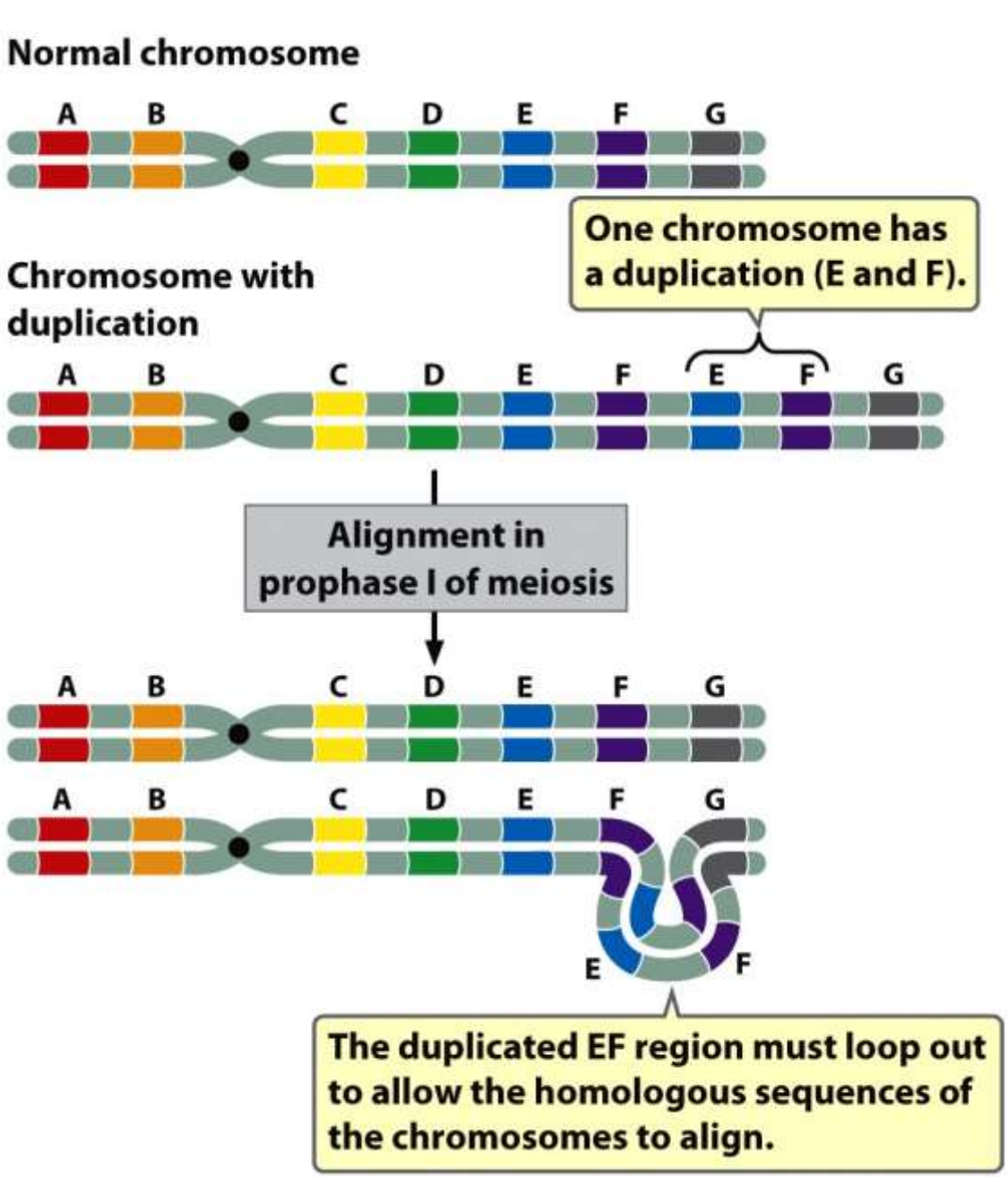

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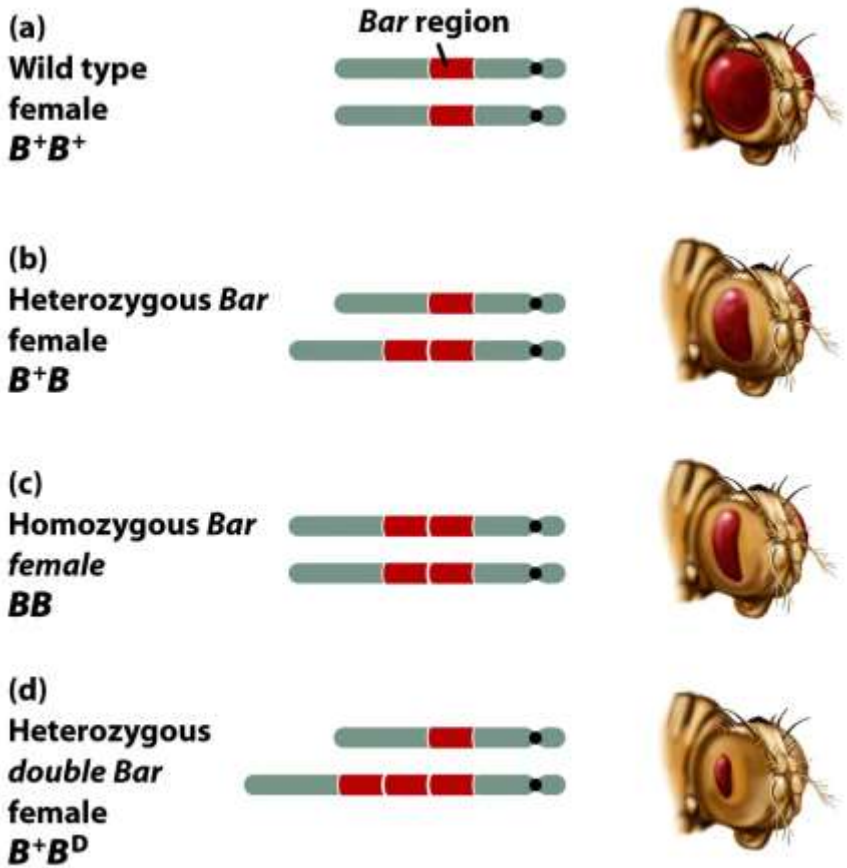
Bar Phenotype



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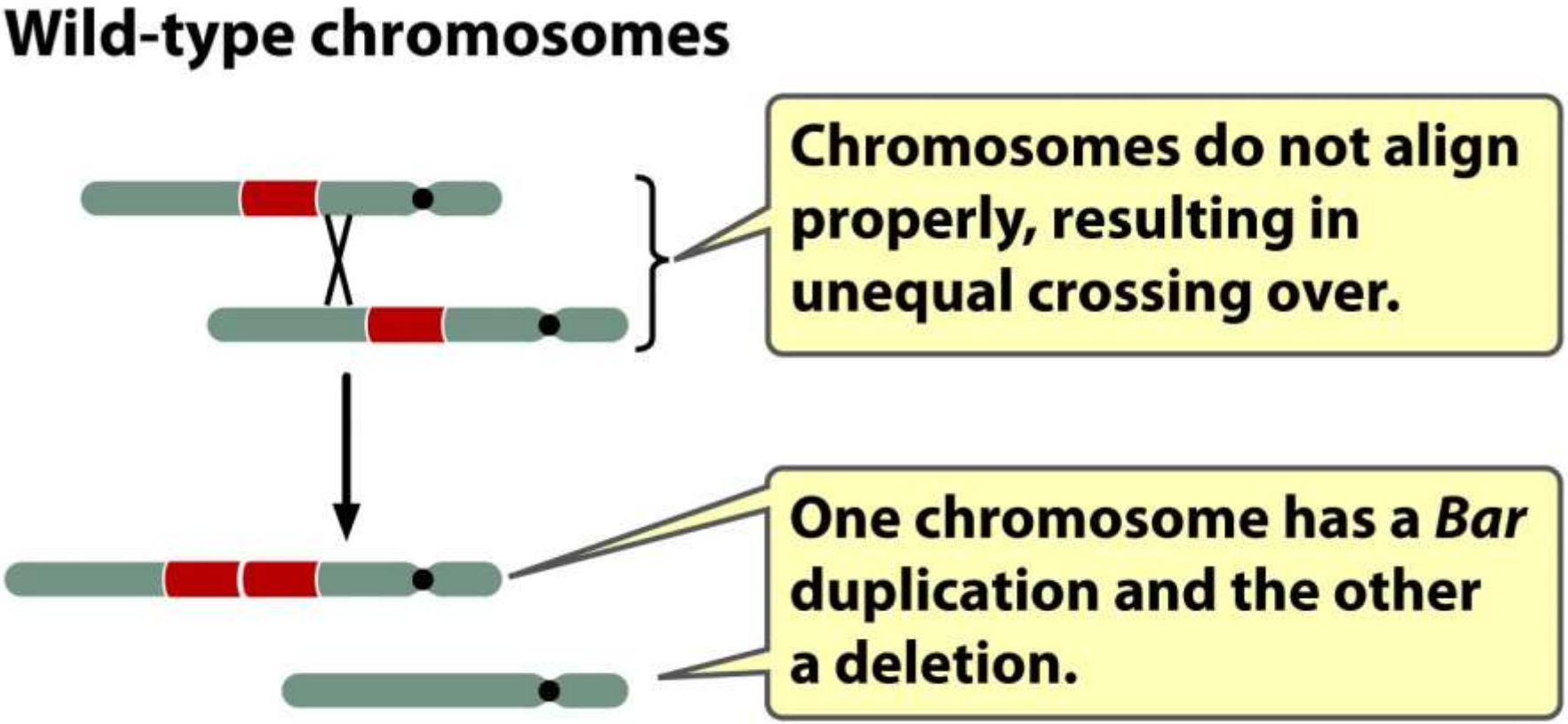

Figure 9-7 part 1
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Bar chromosomes

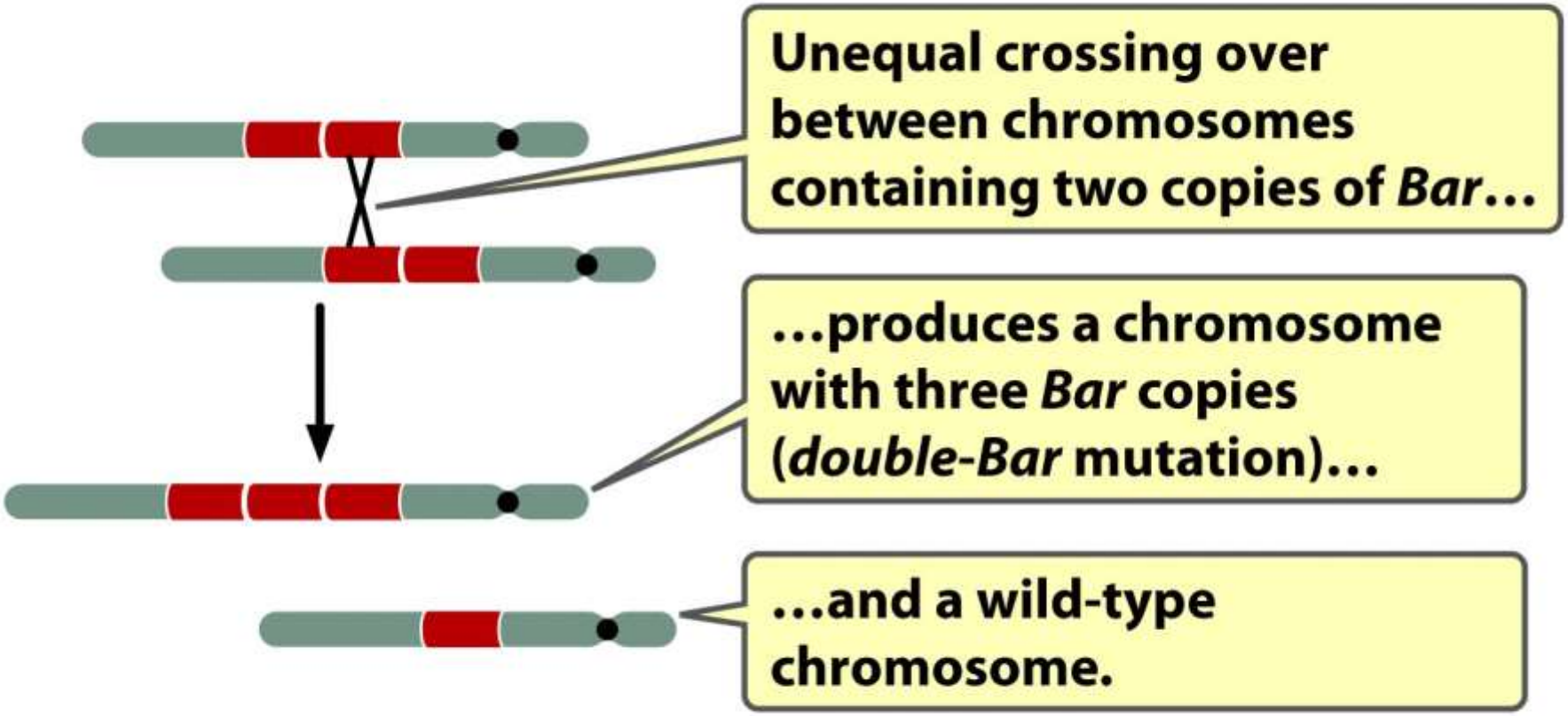

Figure 9-7 part 2
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Unbalanced gene dosage leads to developmental abnormalities

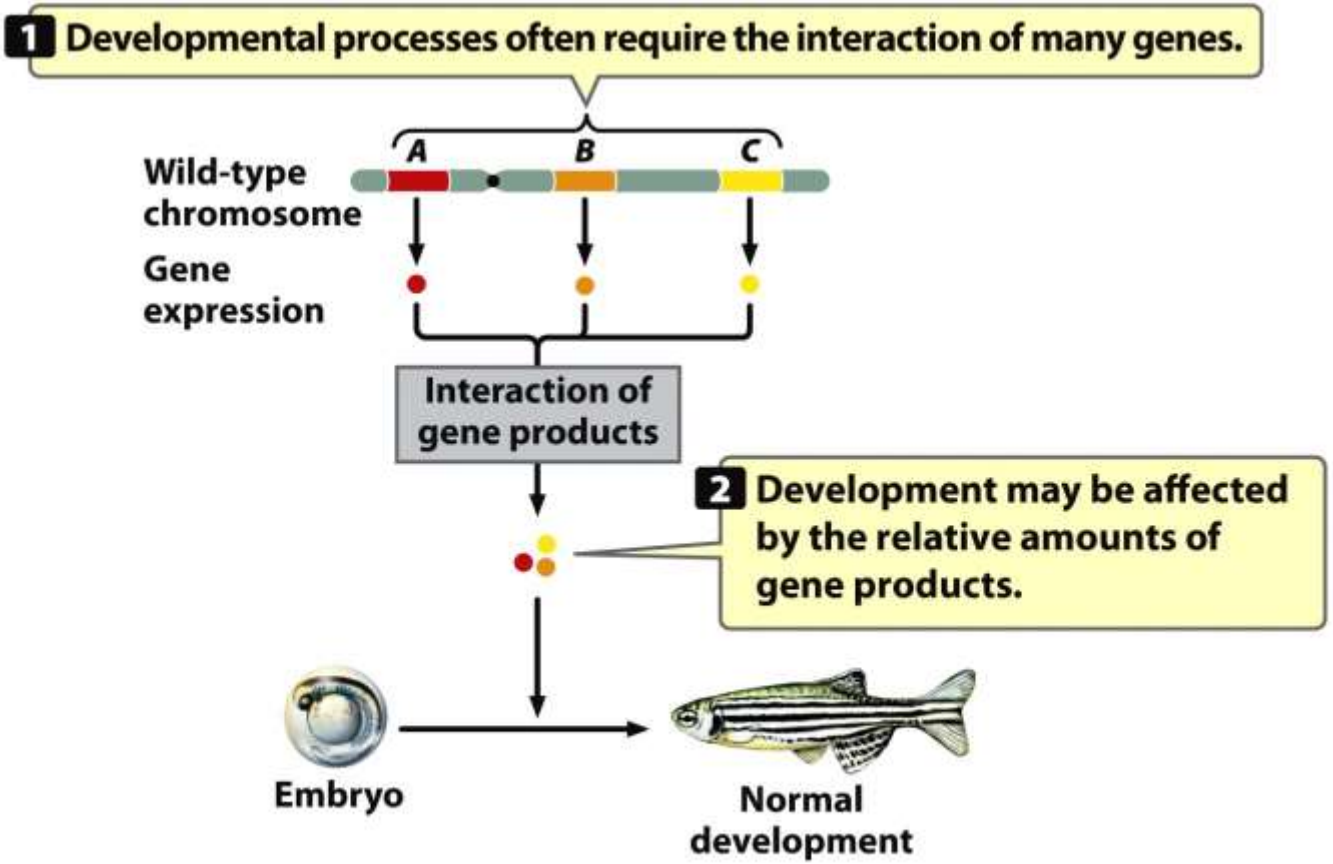

Figure 9-8 part 1
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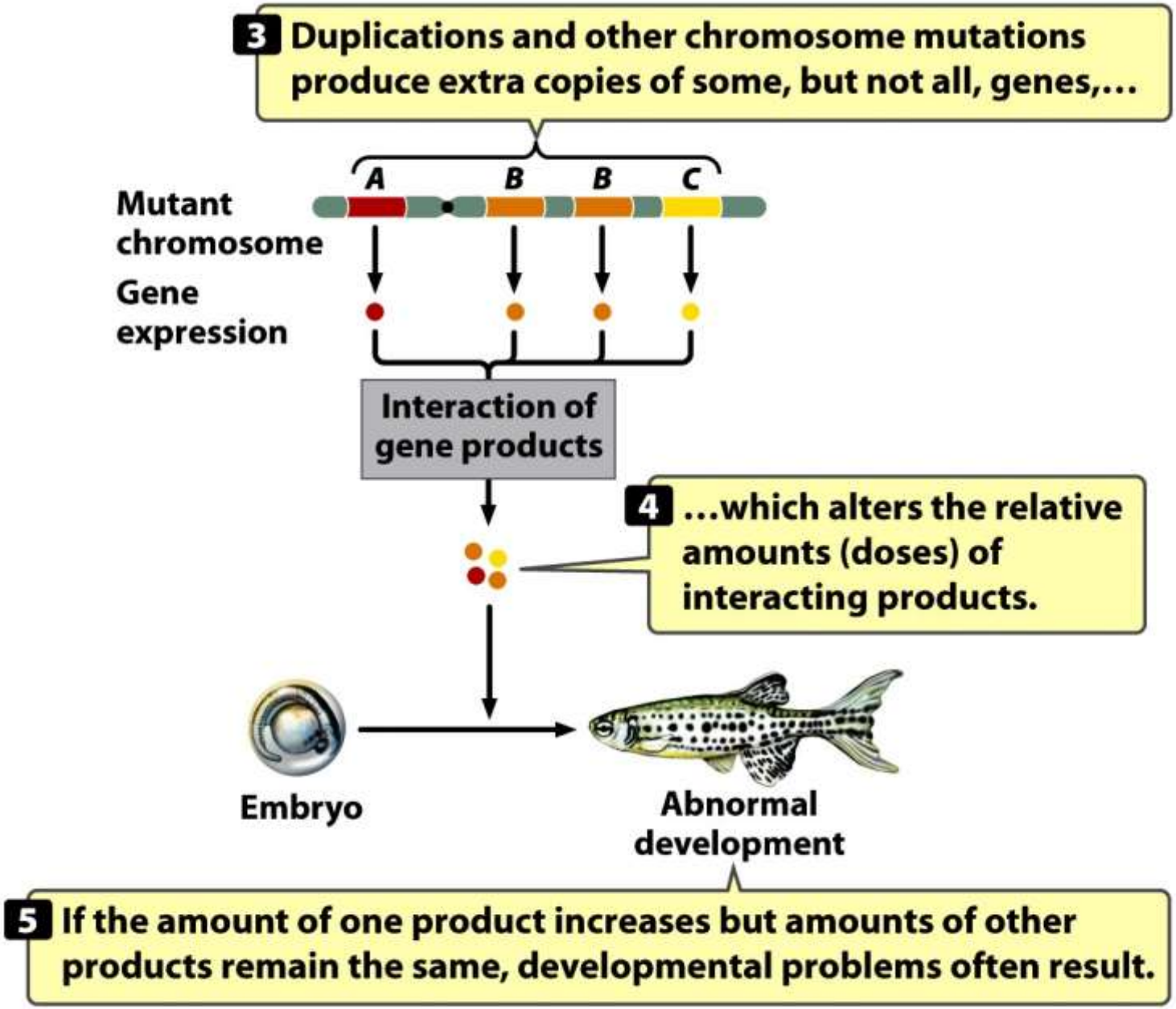

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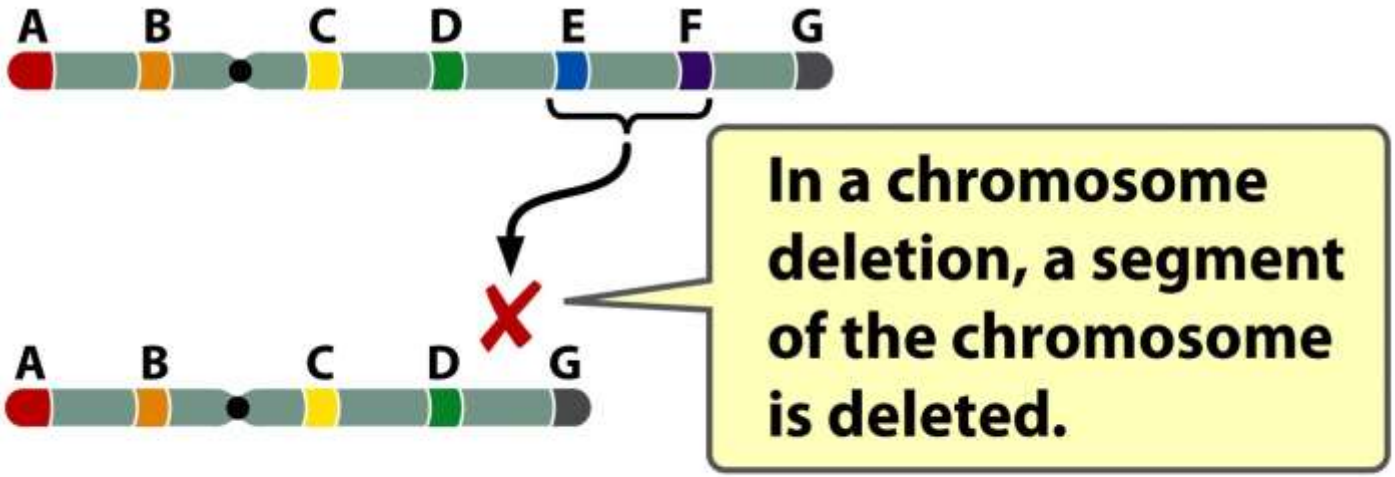
9.2 Chromosome Rearrangements Alter Chromosome Structure

- **Deletions**



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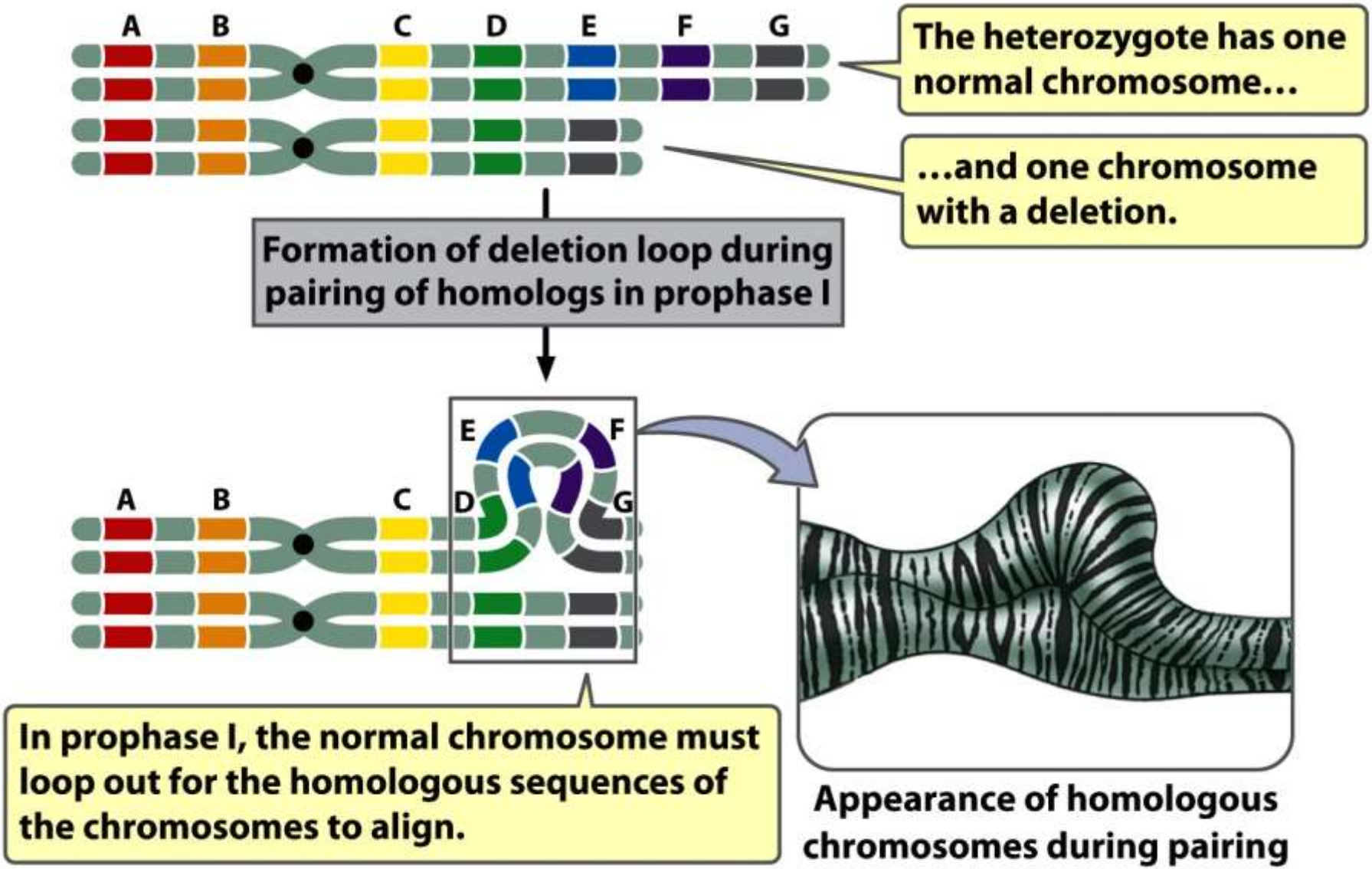

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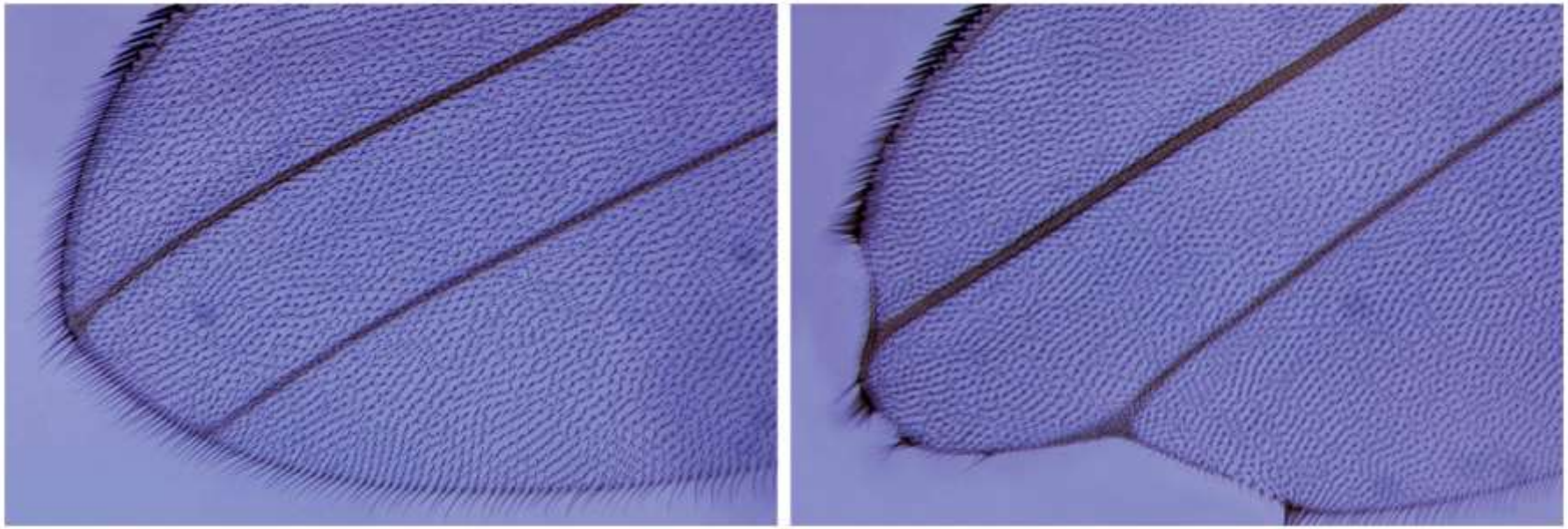

Figure 9-10
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Table 9.1 Effects of some human chromosome rearrangements

Type of Rearrangement	Chromosome	Disorder	Symptoms
Duplication	4, short arm	—	Small head, short neck, low hairline, growth and mental retardation
Duplication	4, long arm	—	Small head, sloping forehead, hand abnormalities
Duplication	7, long arm	—	Delayed development, asymmetry of the head, fuzzy scalp, small nose, low-set ears
Duplication	9, short arm	—	Characteristic face, variable mental retardation, high and broad forehead, hand abnormalities
Deletion	5, short arm	*Cri-du-chat* syndrome	Small head, distinctive cry, widely spaced eyes, round face, mental retardation
Deletion	4, short arm	Wolf–Hirschhorn syndrome	Small head with high forehead, wide nose, cleft lip and palate, severe mental retardation
Deletion	4, long arm	—	Small head, from mild to moderate mental retardation, cleft lip and palate, hand and foot abnormalities
Deletion	7, long arm	Williams–Beuren syndrome	Facial features, heart defects, mental impairment
Deletion	15, long arm	Prader–Willi syndrome	Feeding difficulty at early age, but becoming obese after 1 year of age, from mild to moderate mental retardation
Deletion	18, short arm	—	Round face, large low-set ears, from mild to moderate mental retardation
Deletion	18, long arm	—	Distinctive mouth shape, small hands, small head, mental retardation

Table 9-1
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Chromosome Rearrangements Alter Chromosome Structure

- **Inversion** (depends on the involvement of the centromere in the inversion):
  - Paracentric inversion
  - Pericentric inversion

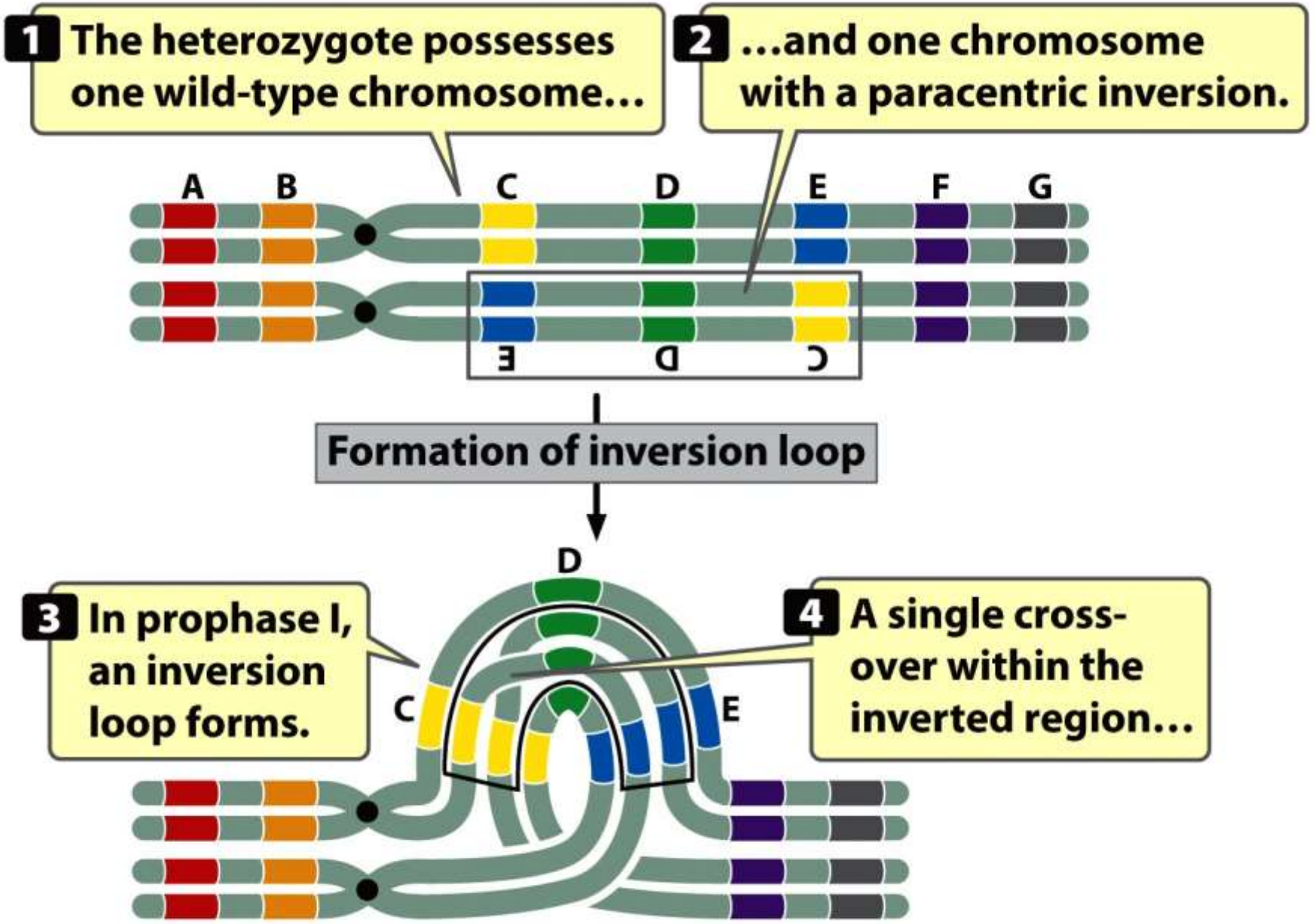

Figure 9-12 part 1
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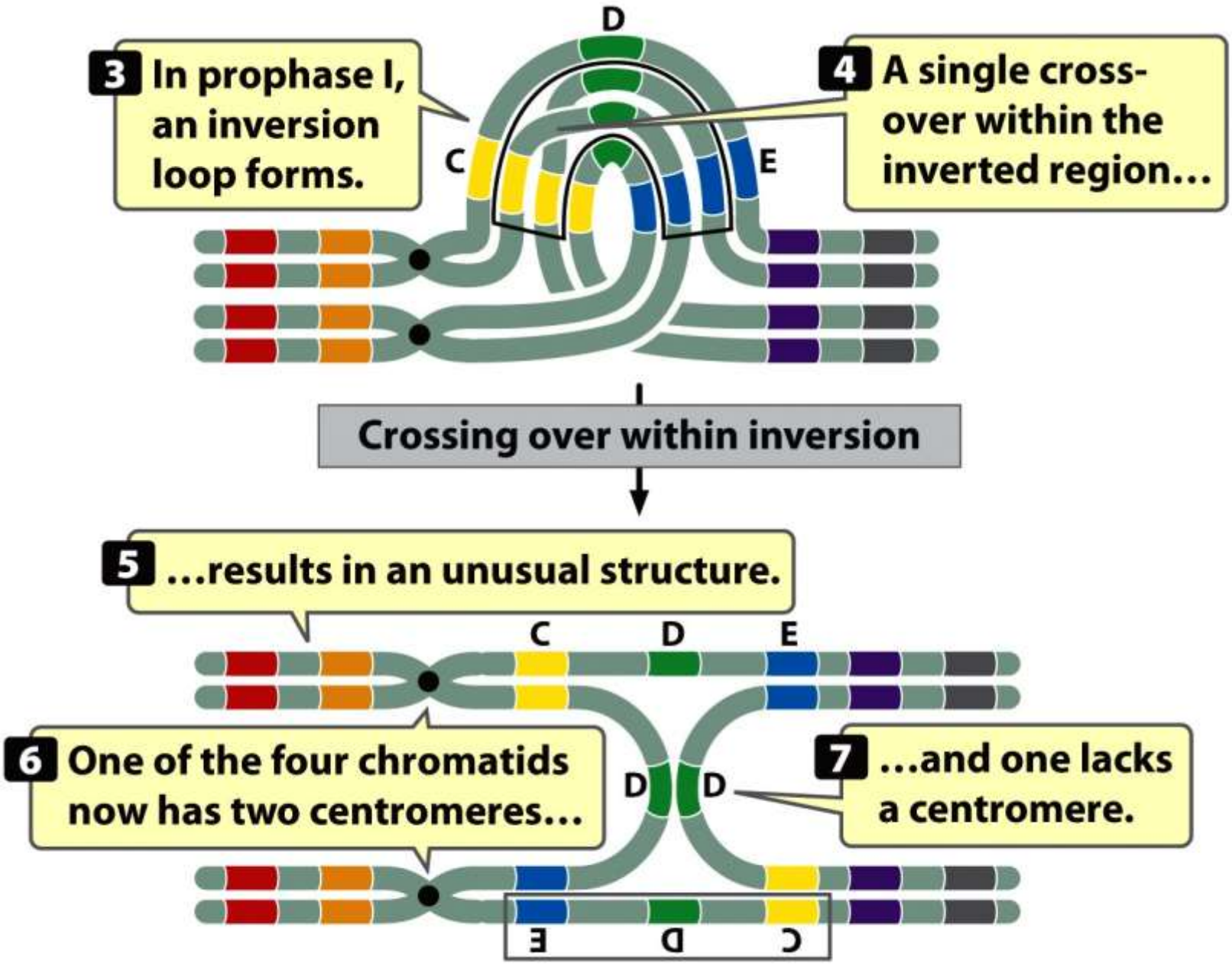

Figure 9-12 part 2
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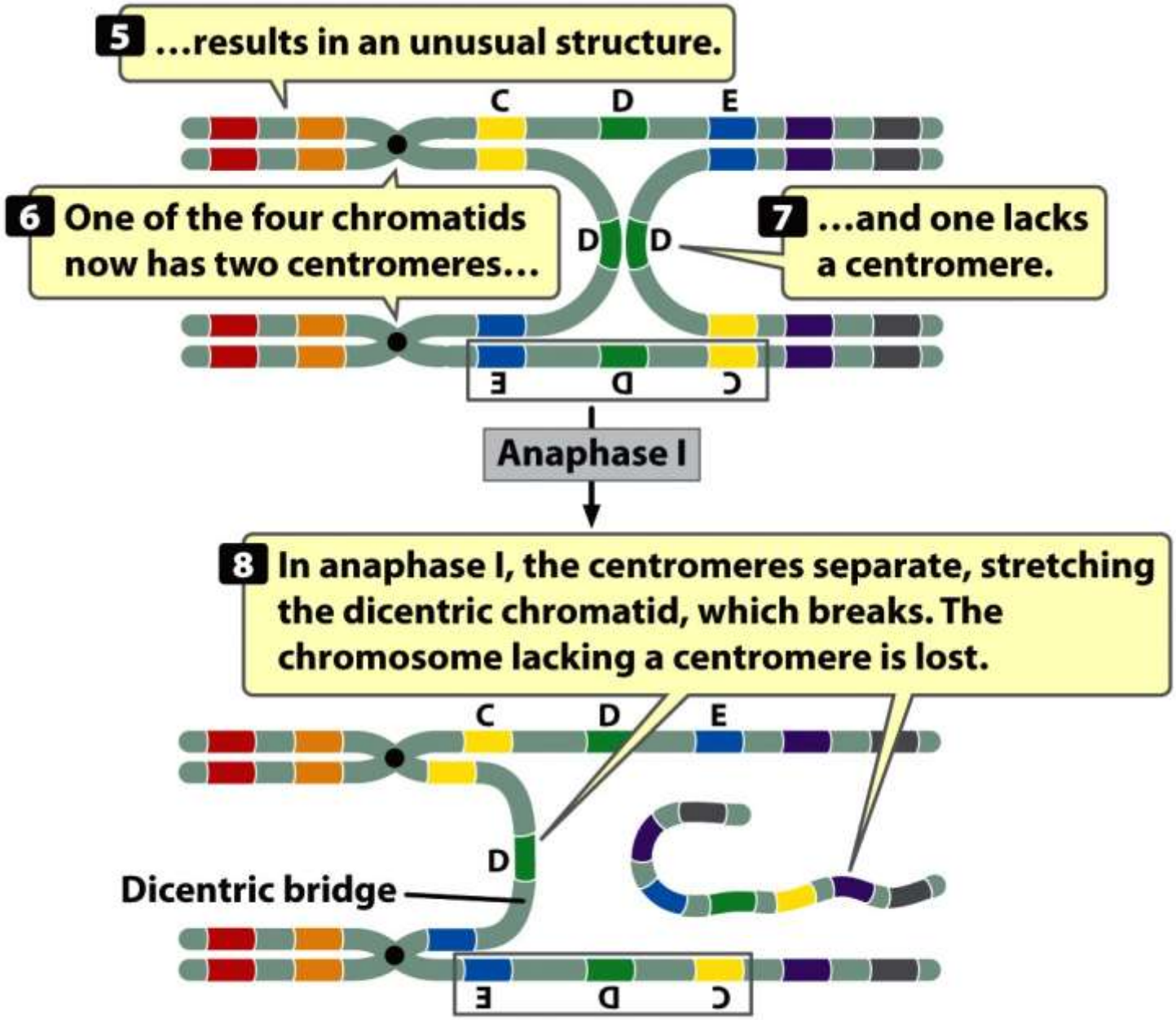

Figure 9-12 part 3
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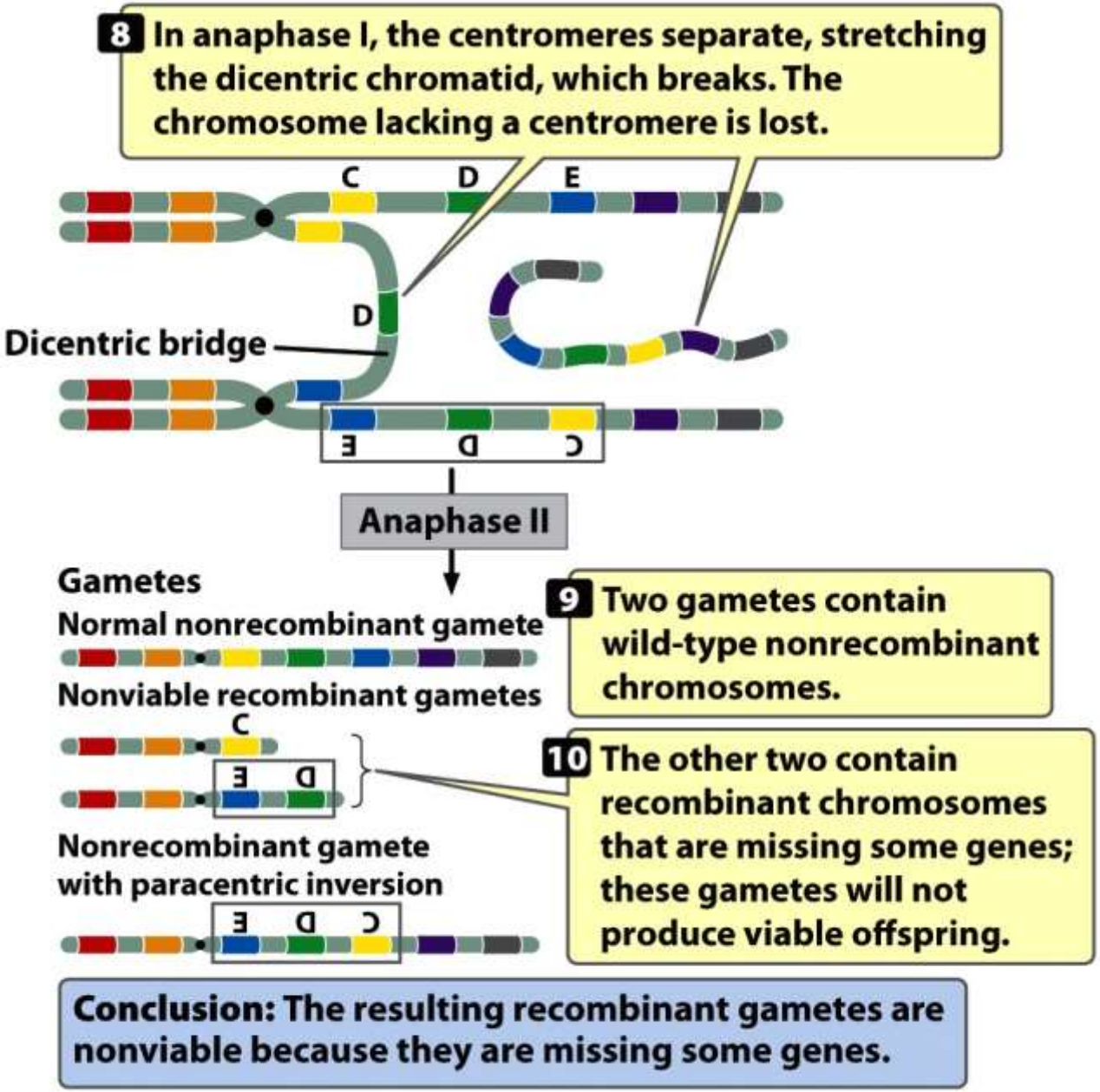

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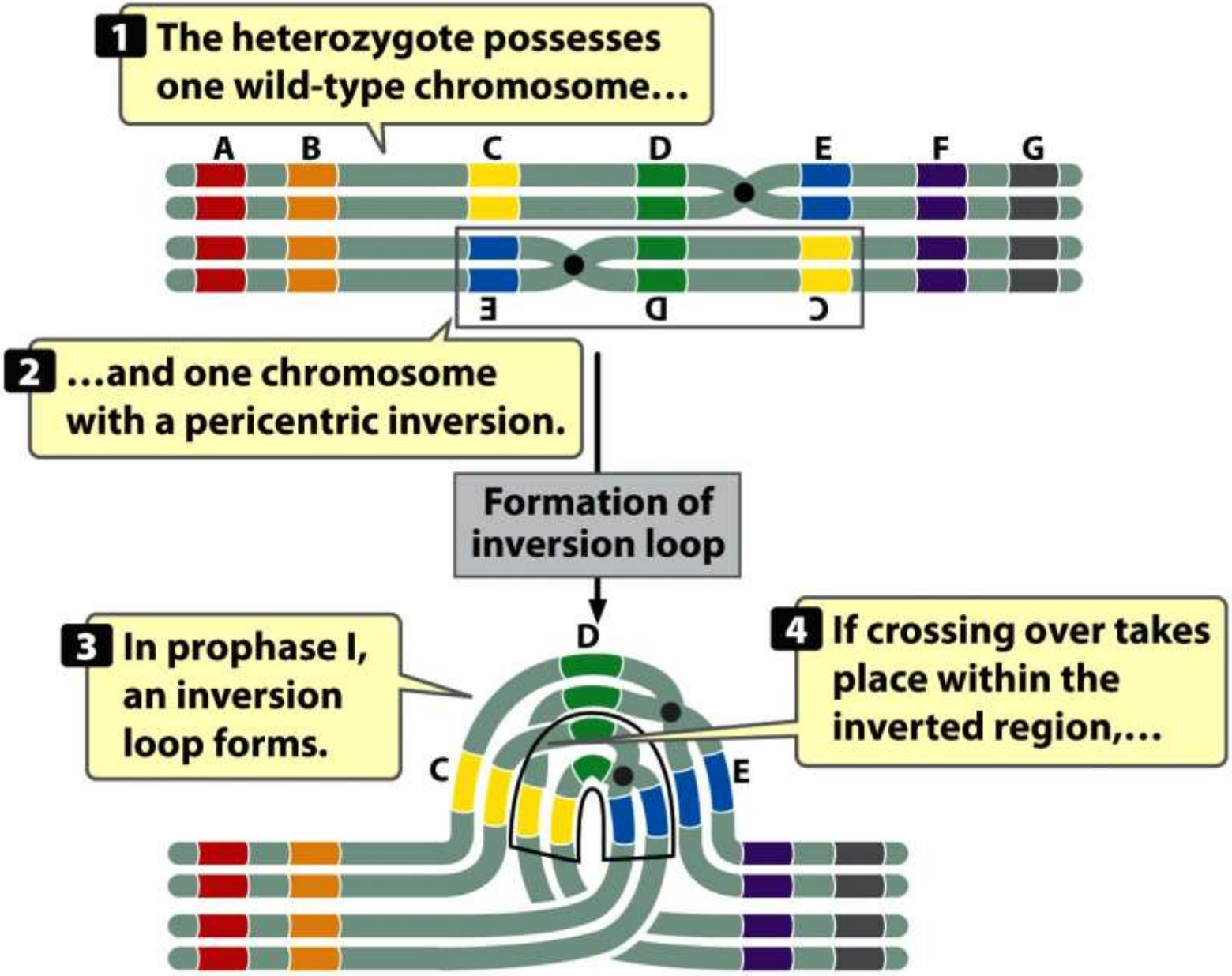

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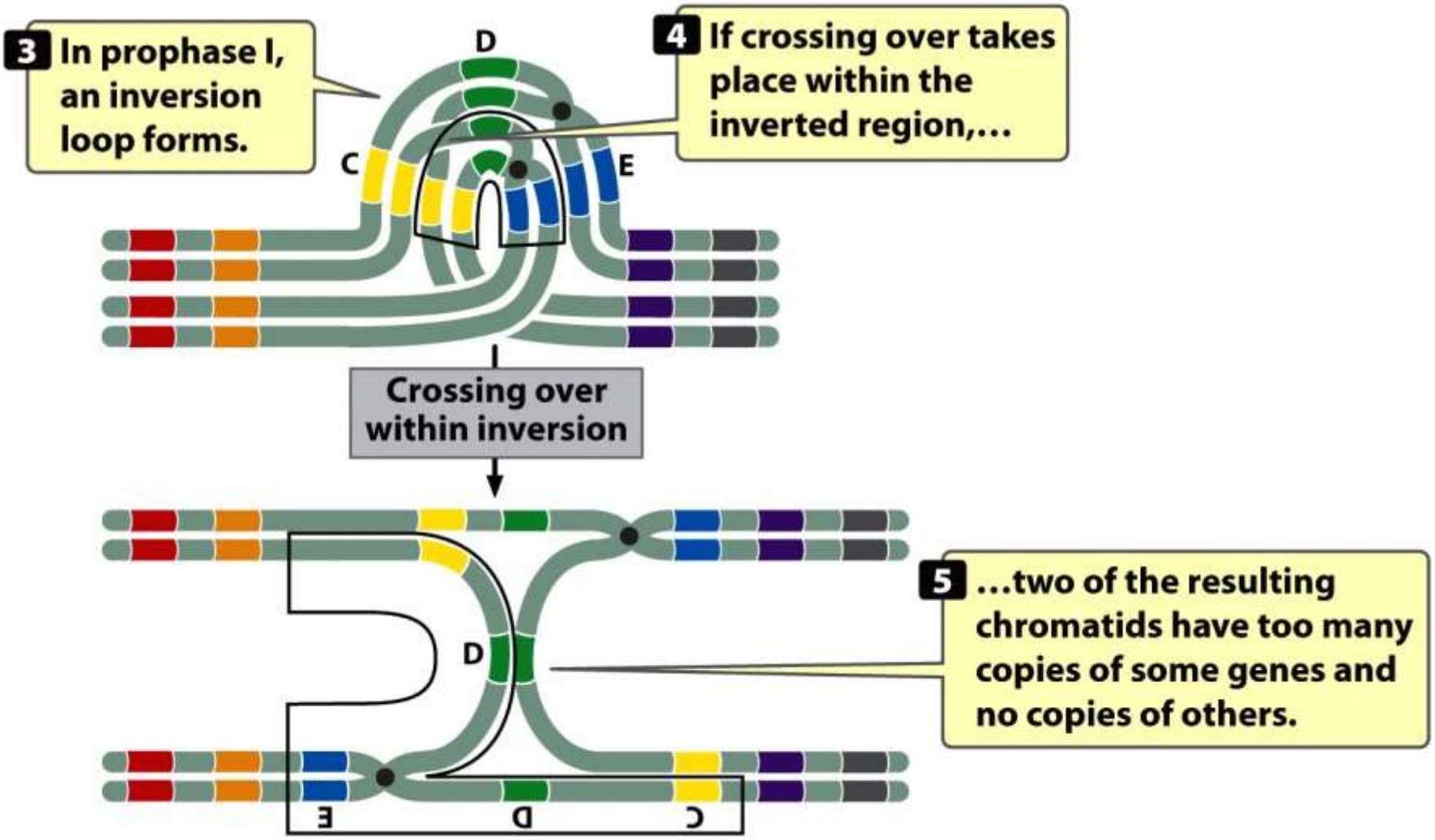

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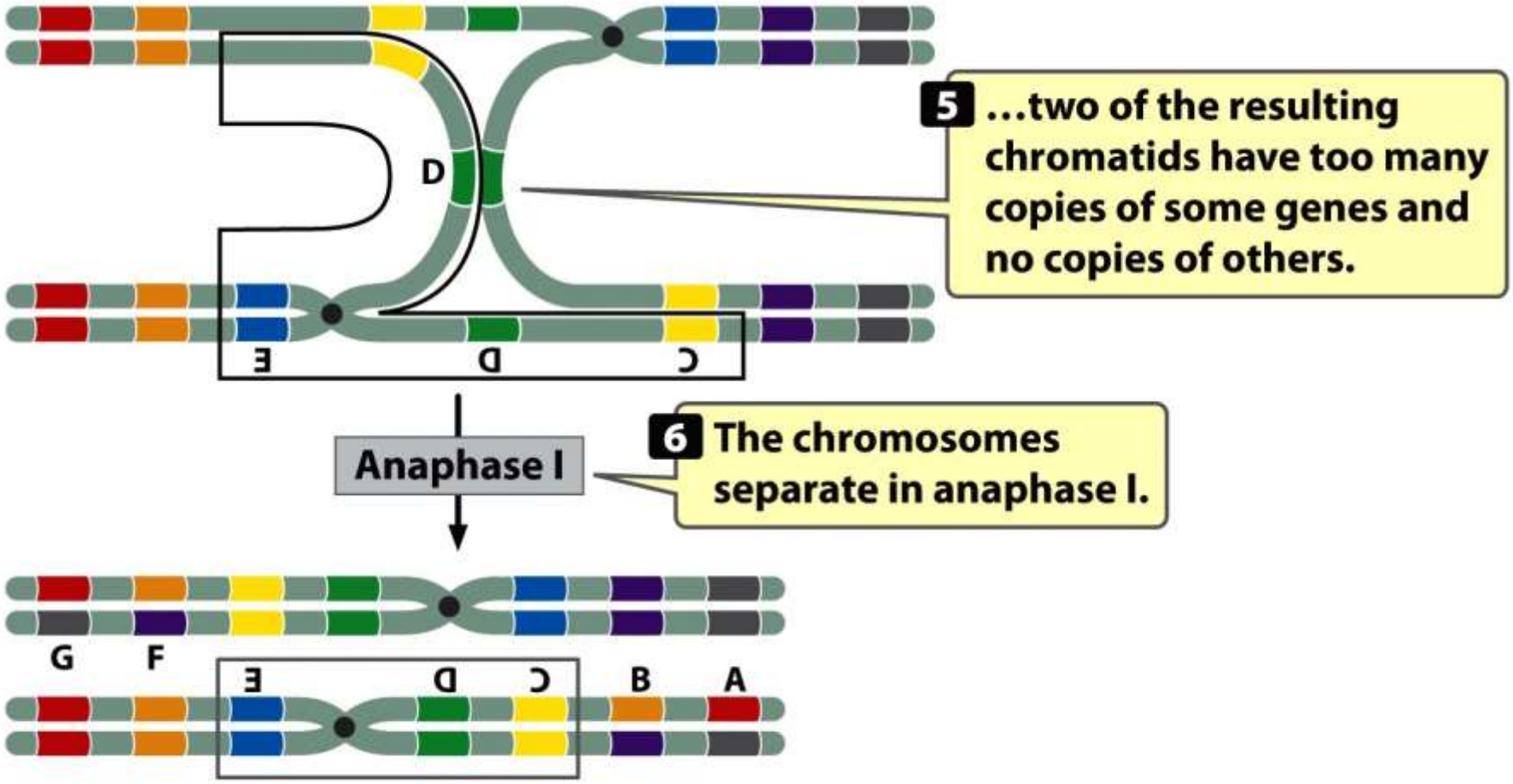

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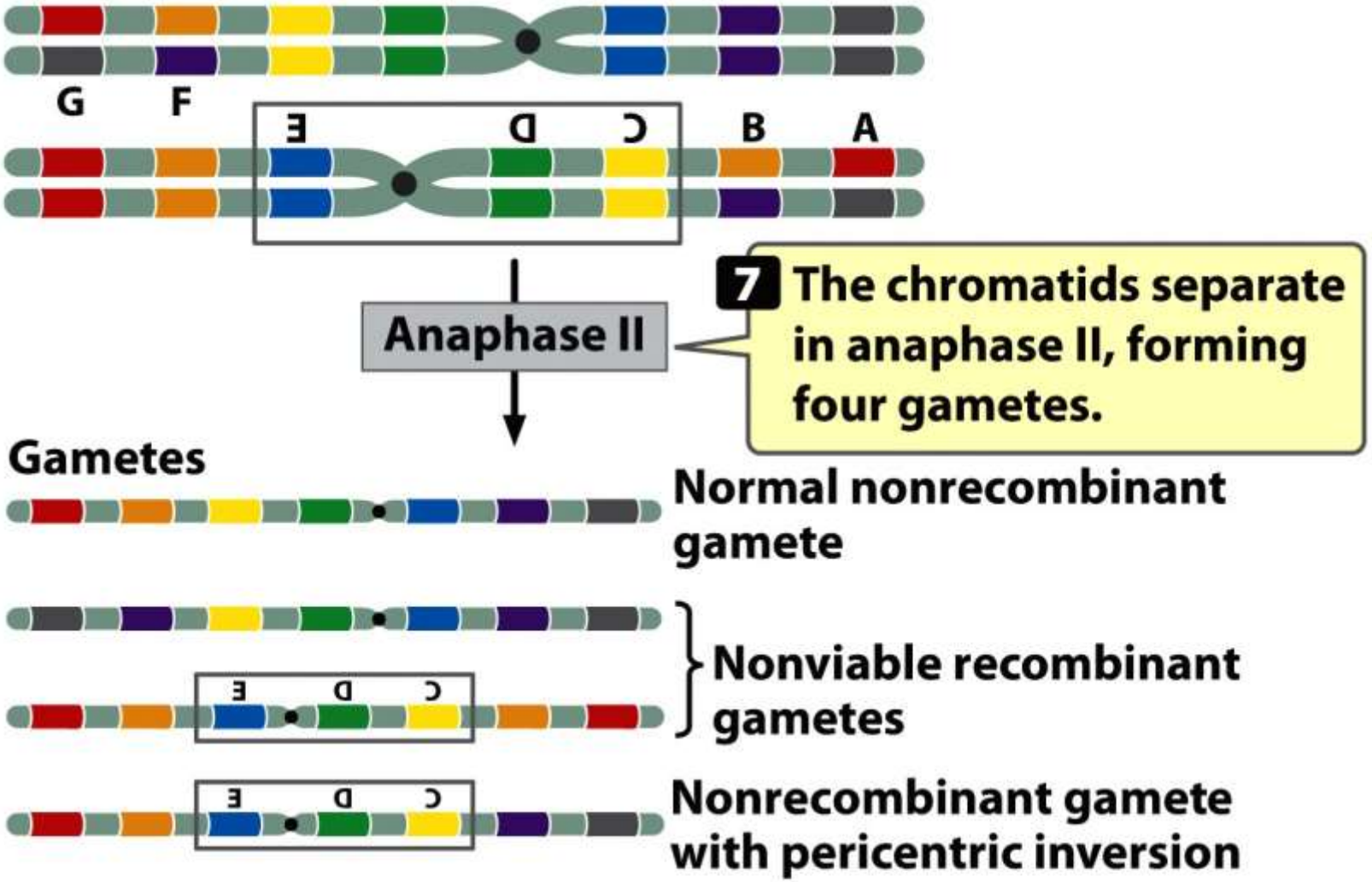

Conclusion: Recombinant gametes are nonviable because genes are either missing or present in too many copies.

Figure 9-13 part 4
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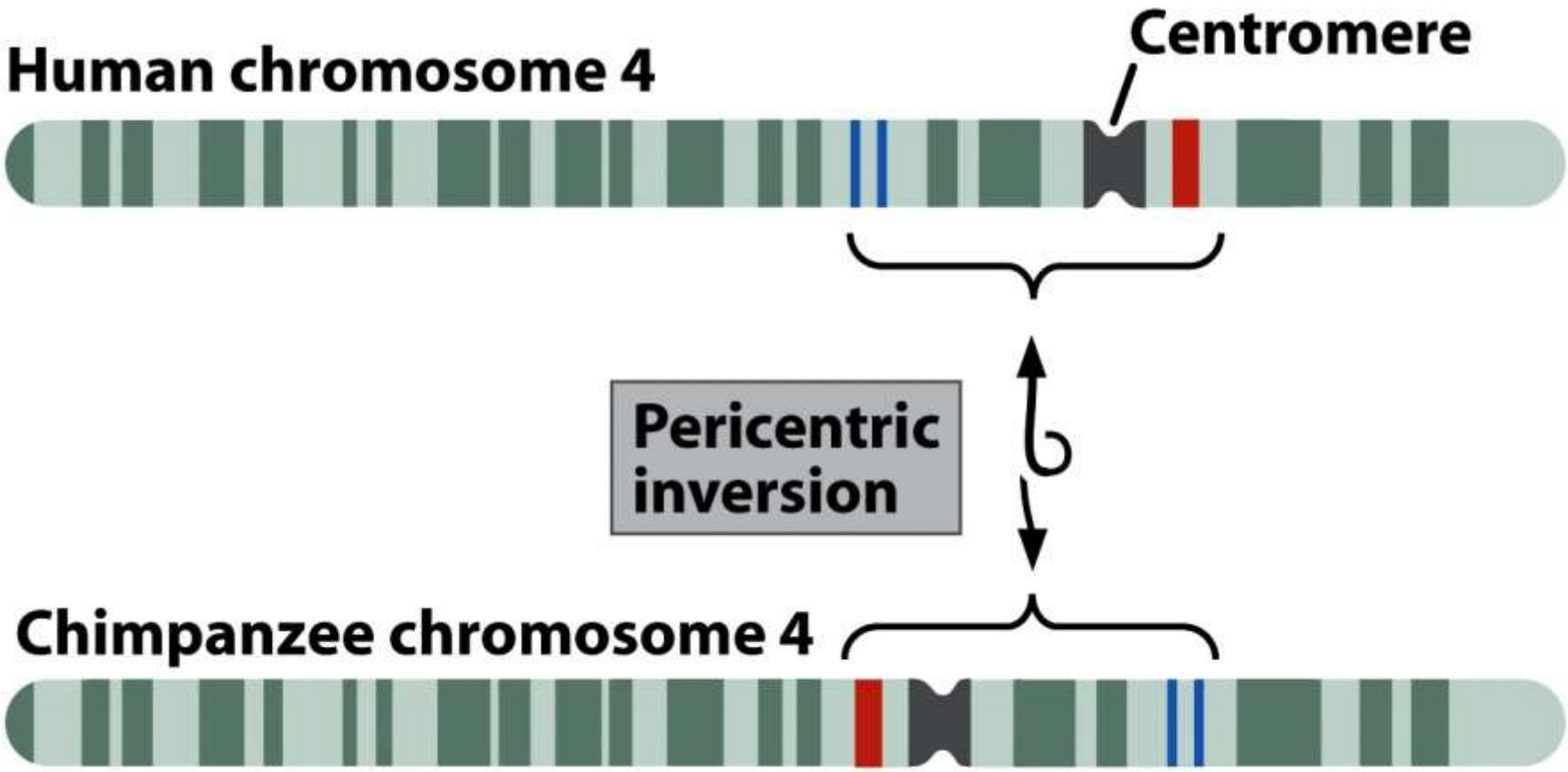

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Translocation

- Nonreciprocal translocation
- Reciprocal translocation
- Robertsonian translocation

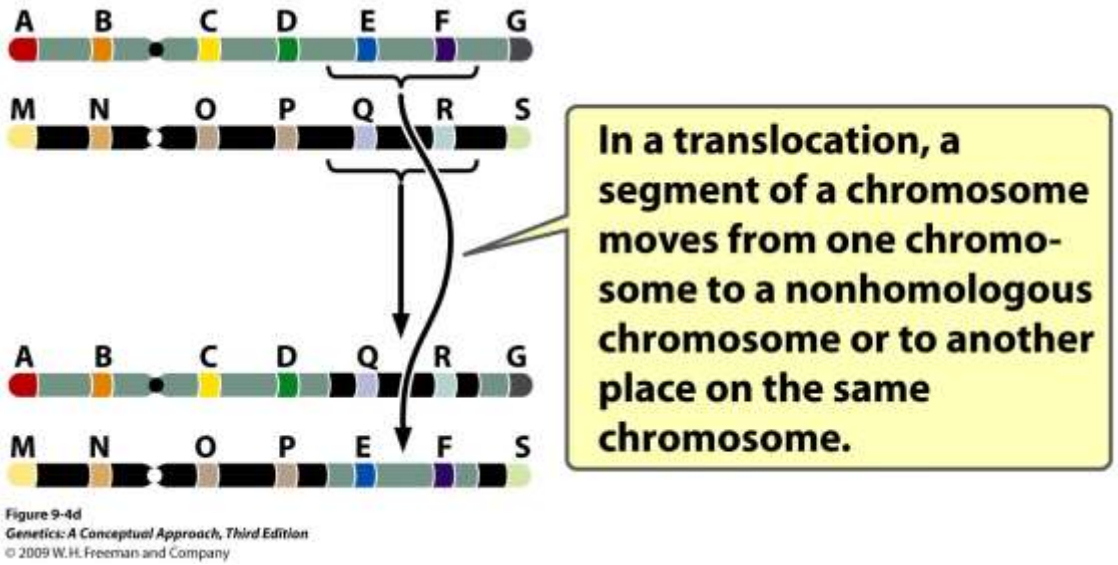

Figure 9-4d
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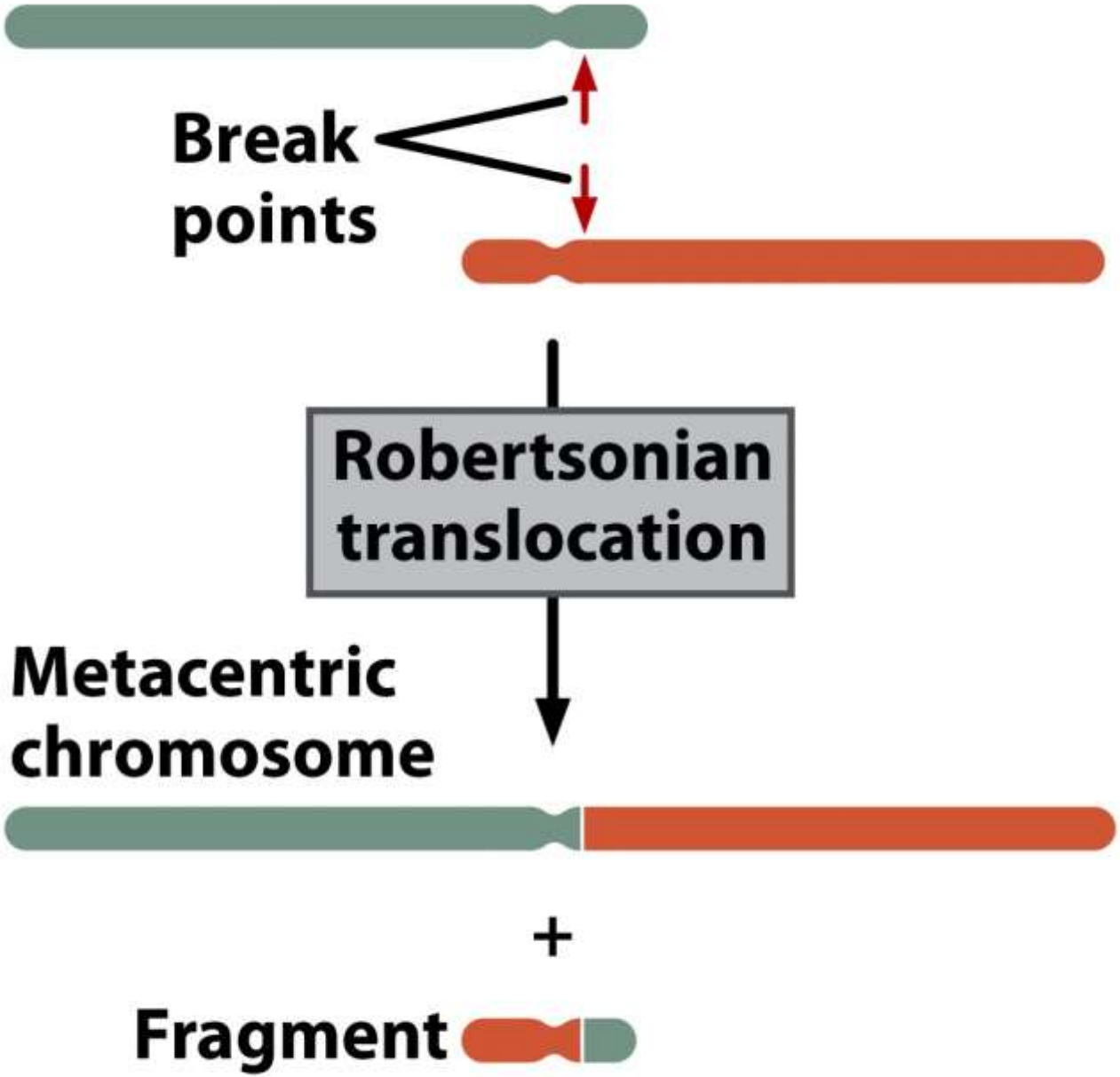

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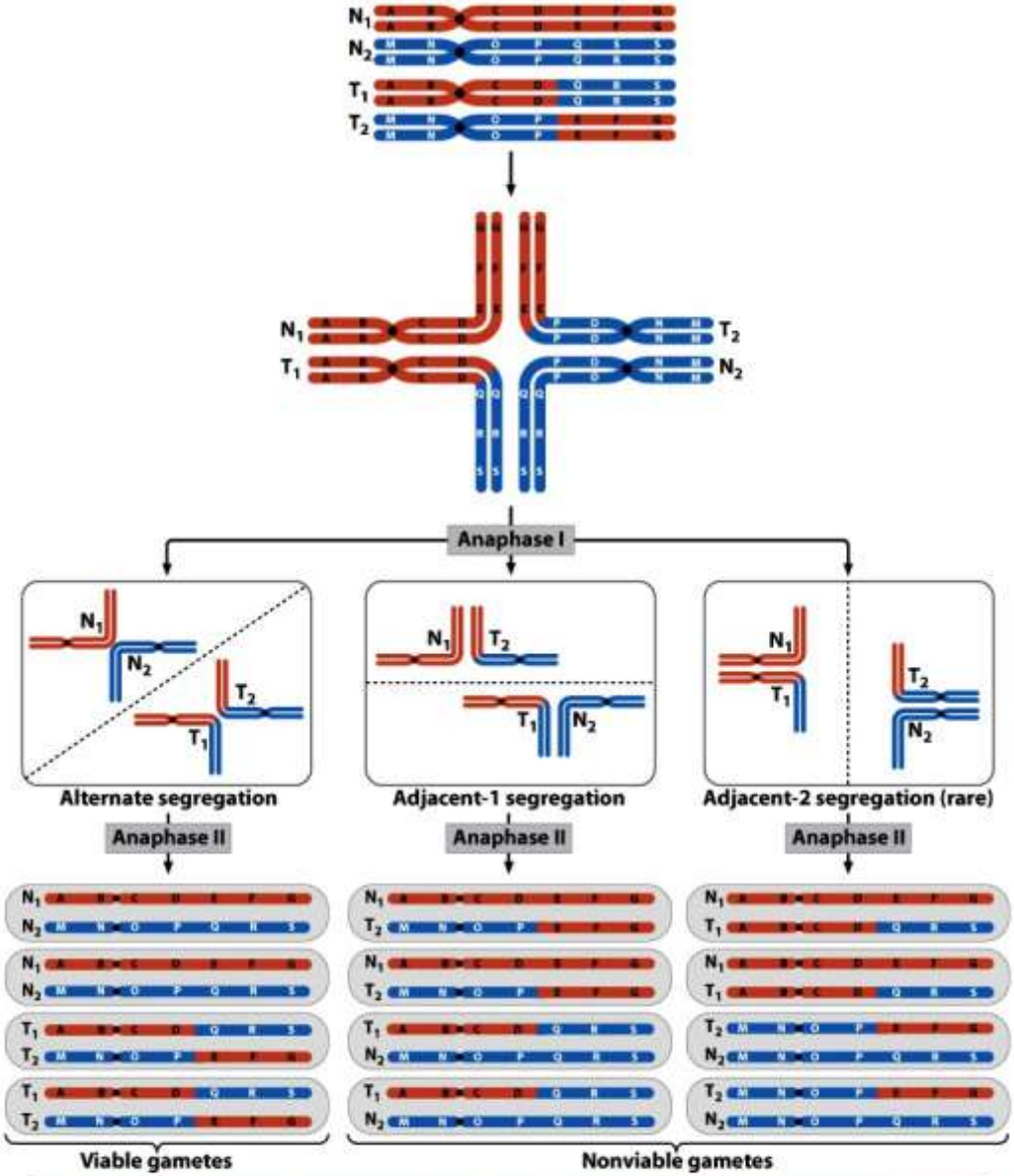

Figure 9-16

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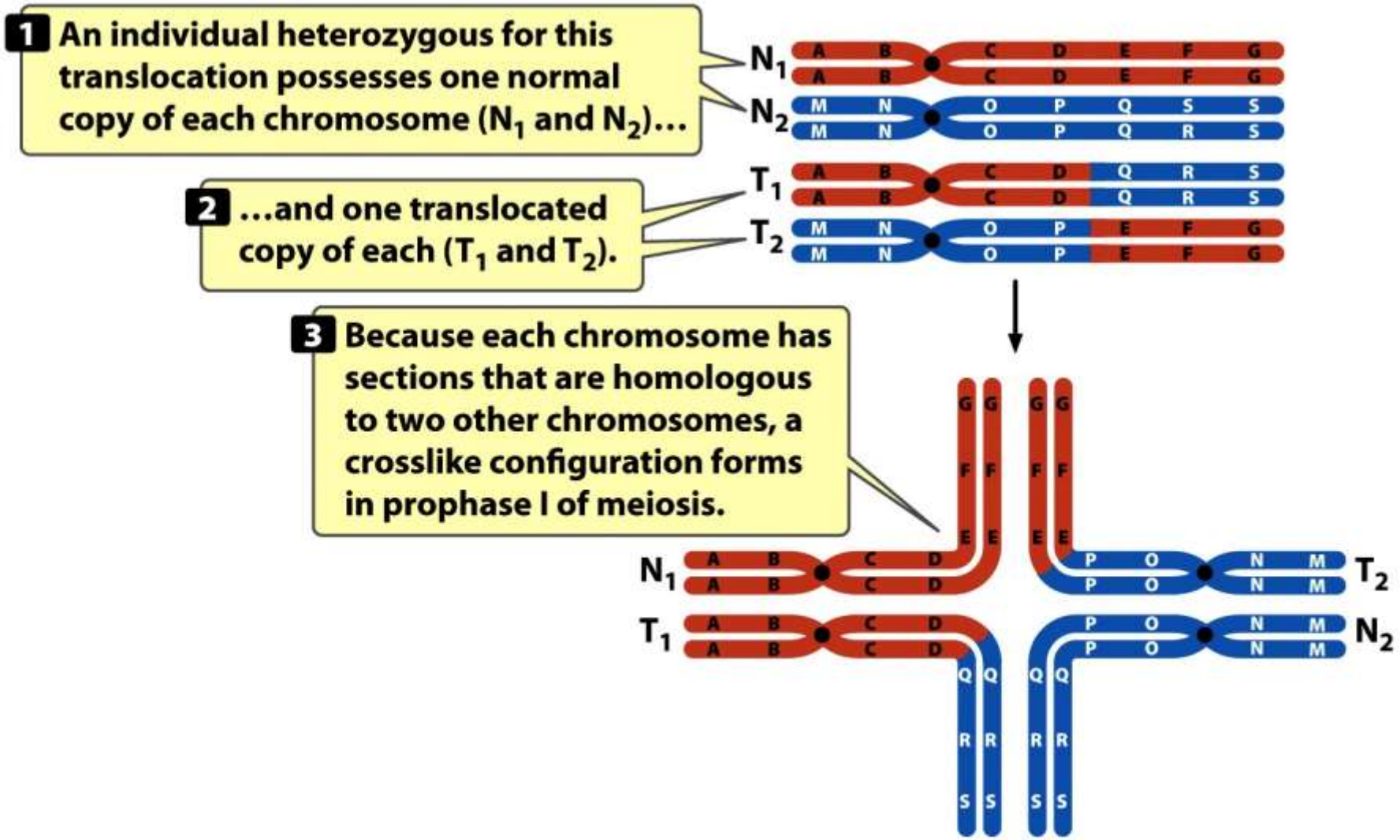

Figure 9-16 part 1
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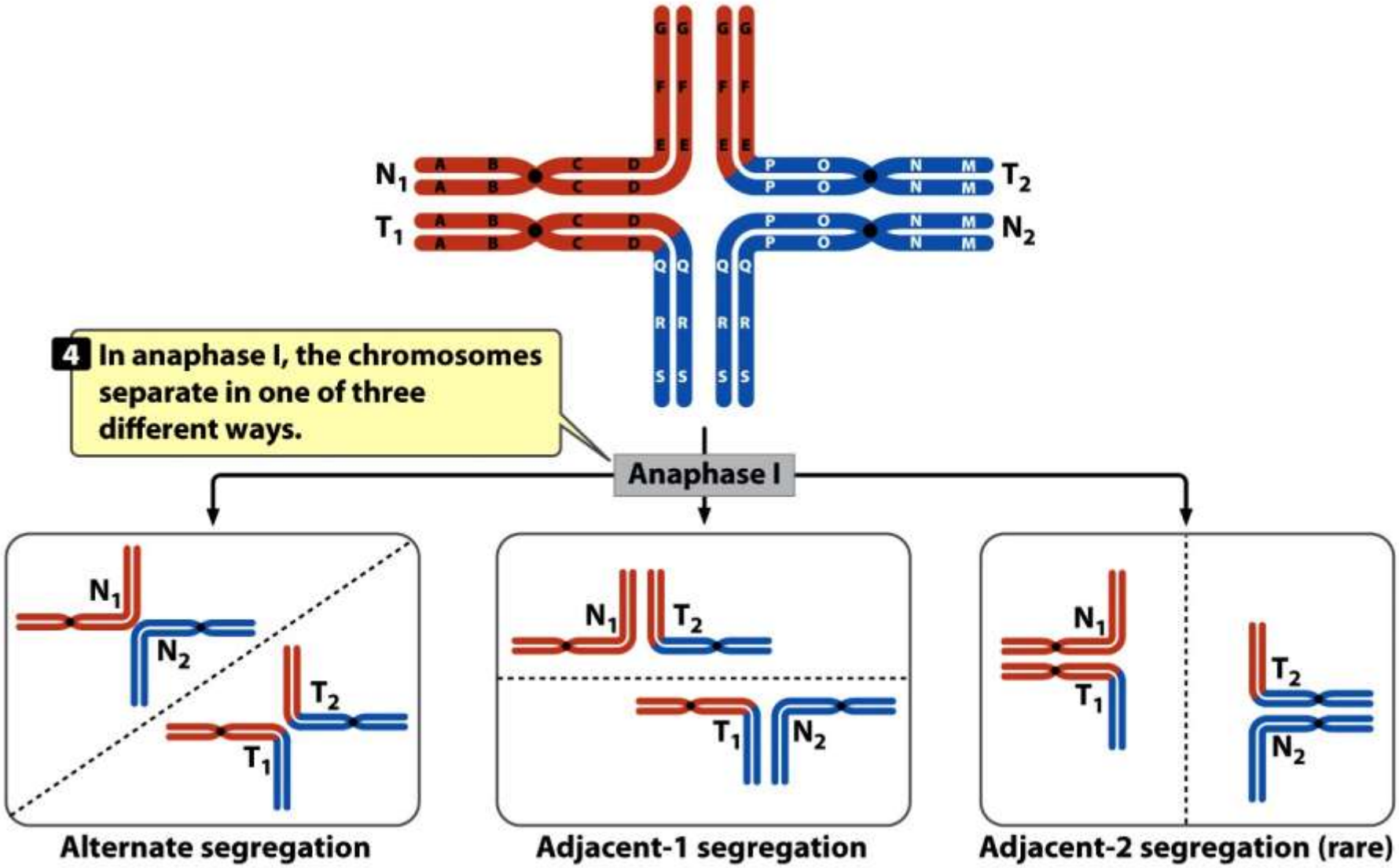

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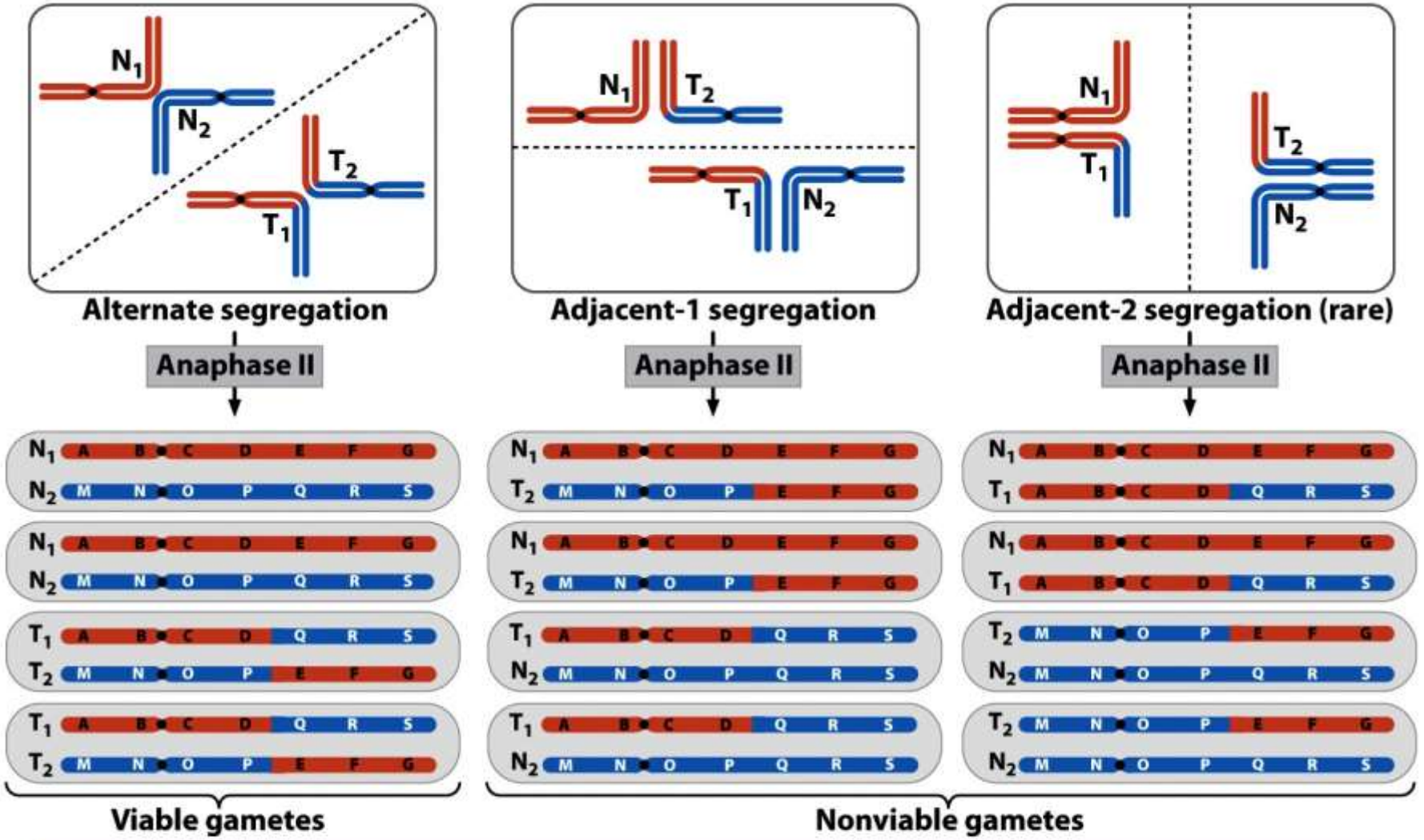

Conclusion: Gametes resulting from adjacent-I and adjacent-2 segregation are nonviable because some genes are present in two copies, whereas others are missing.

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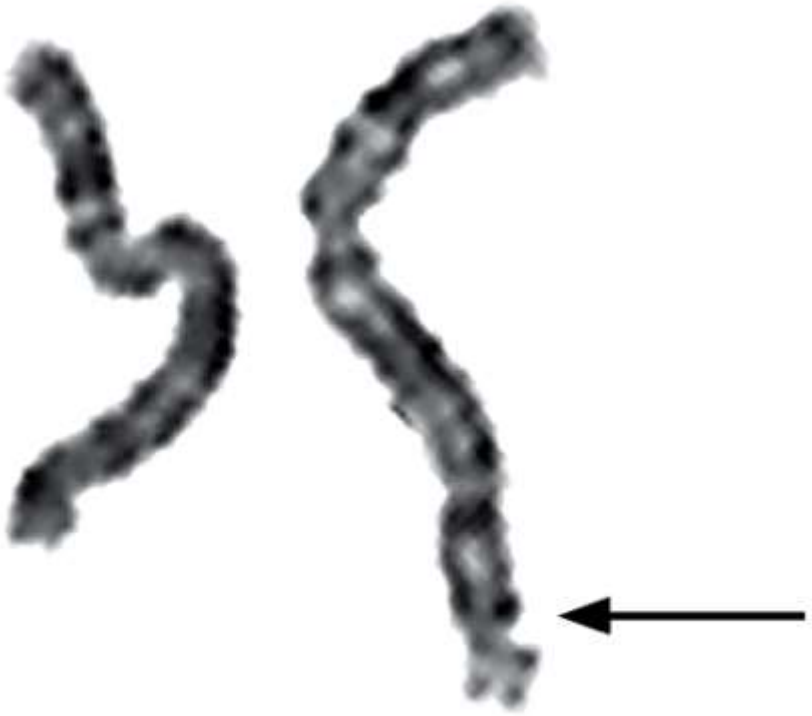
Fragile Sites



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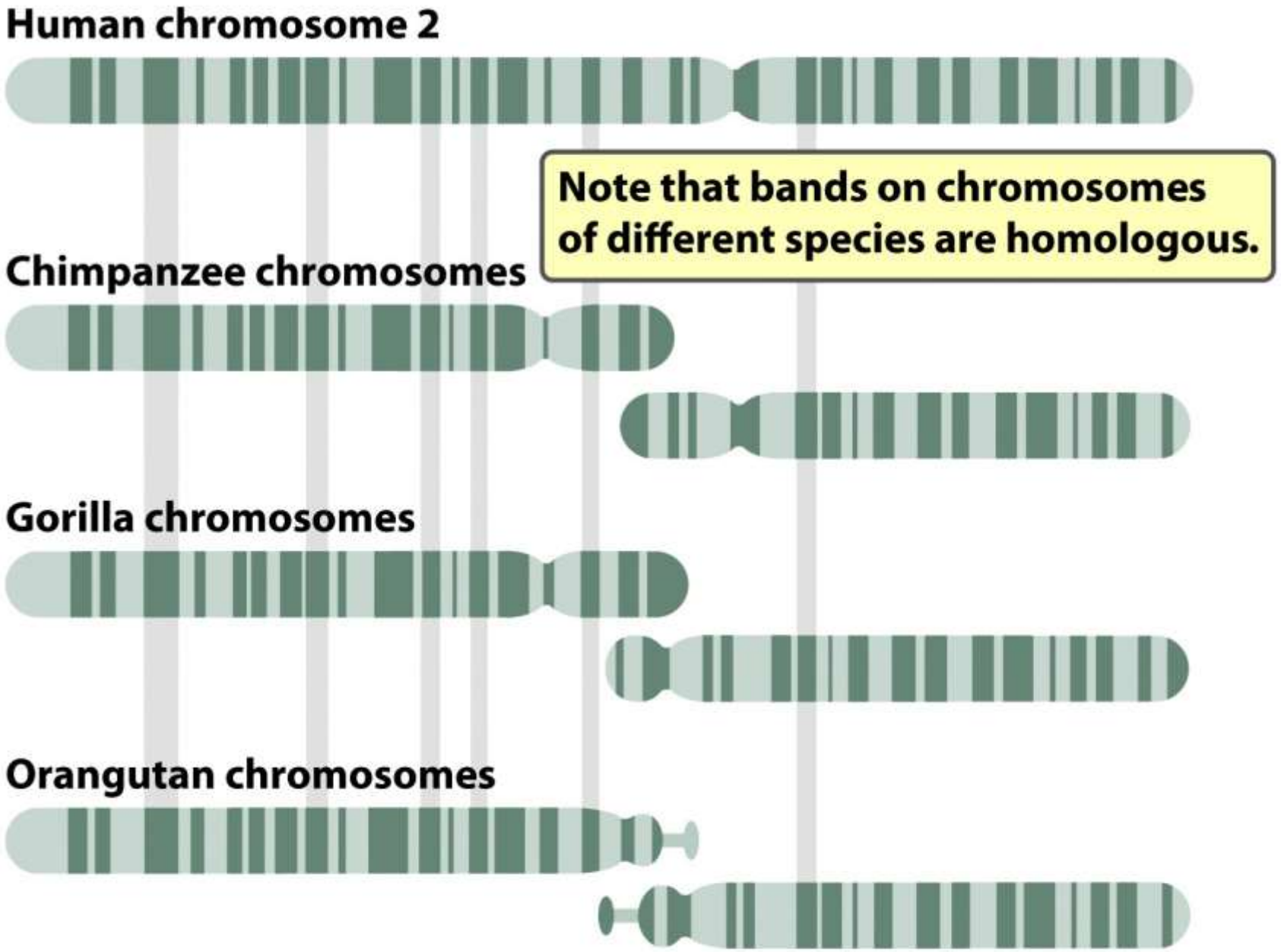

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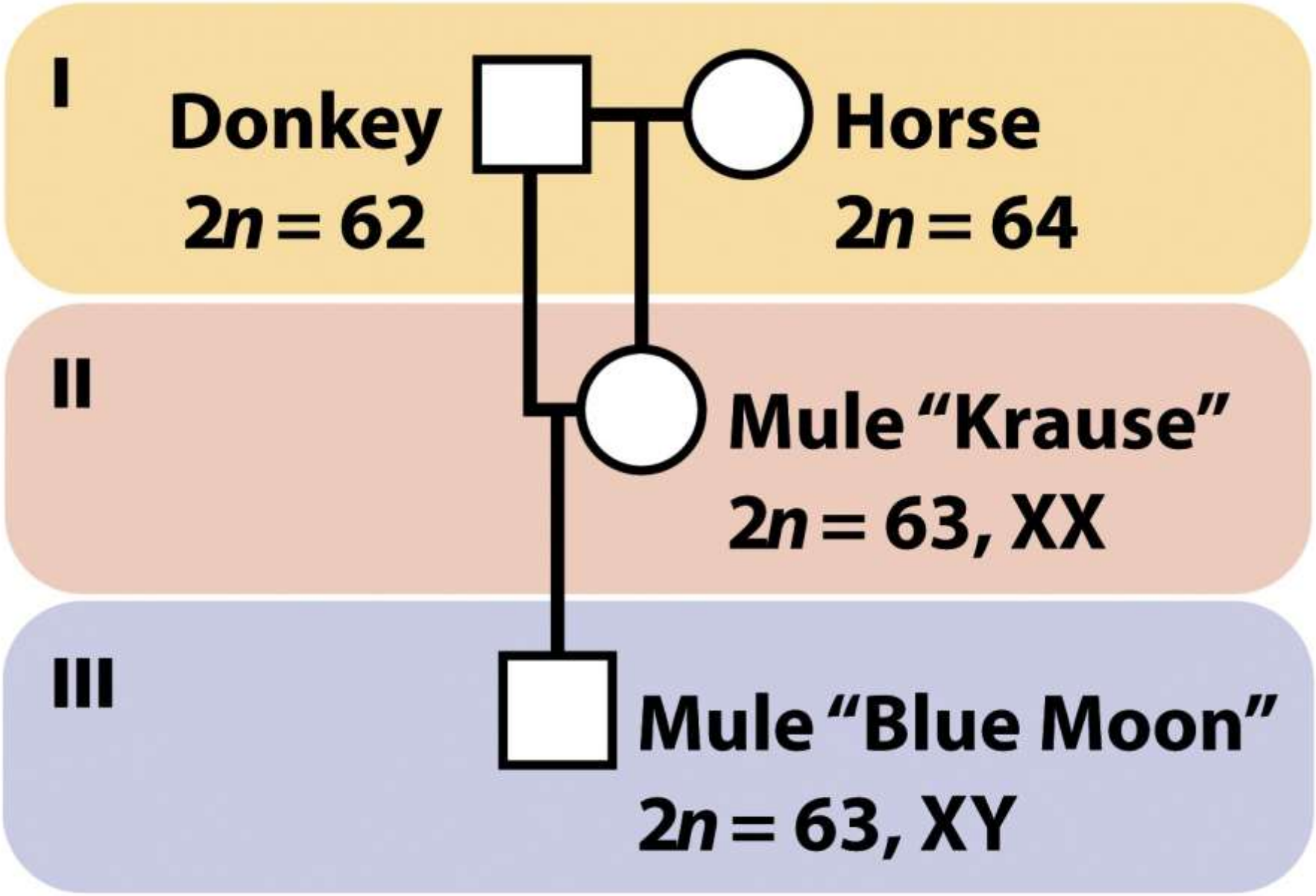
Aneuploidy



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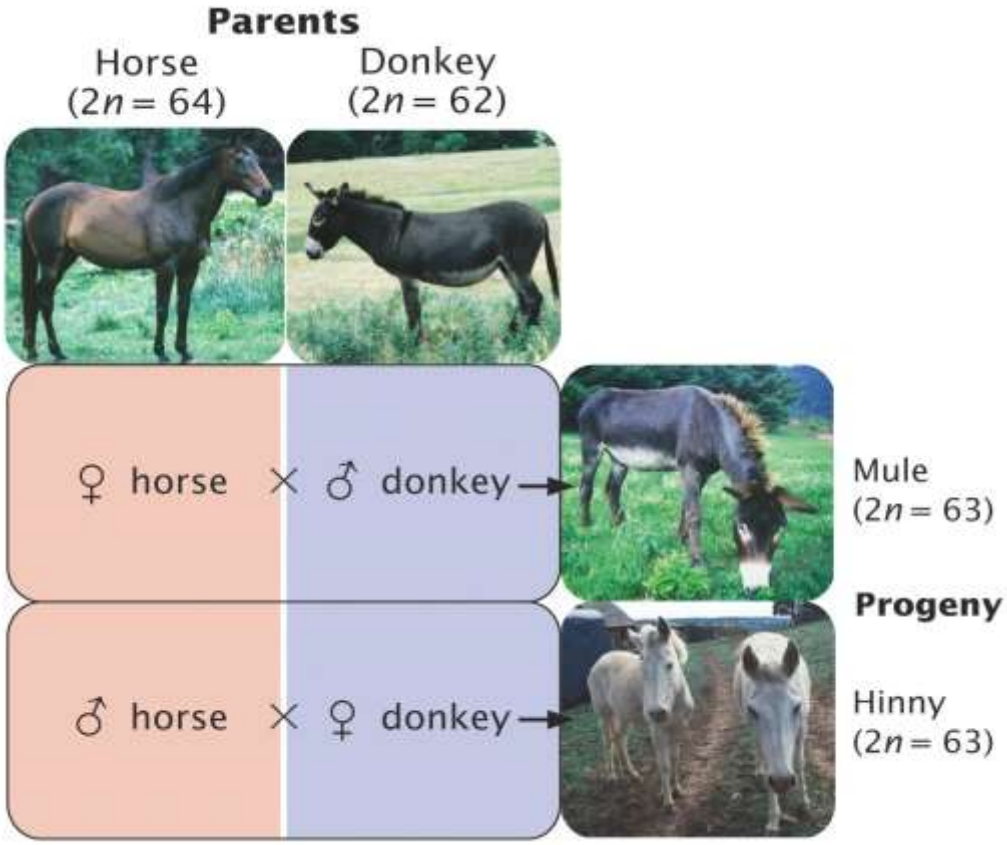
Once in a Blue Moon

Crosses Between Horses & Donkeys



9.3 Aneuploidy Is an Increase or Decrease in the Number of Individual Chromosomes

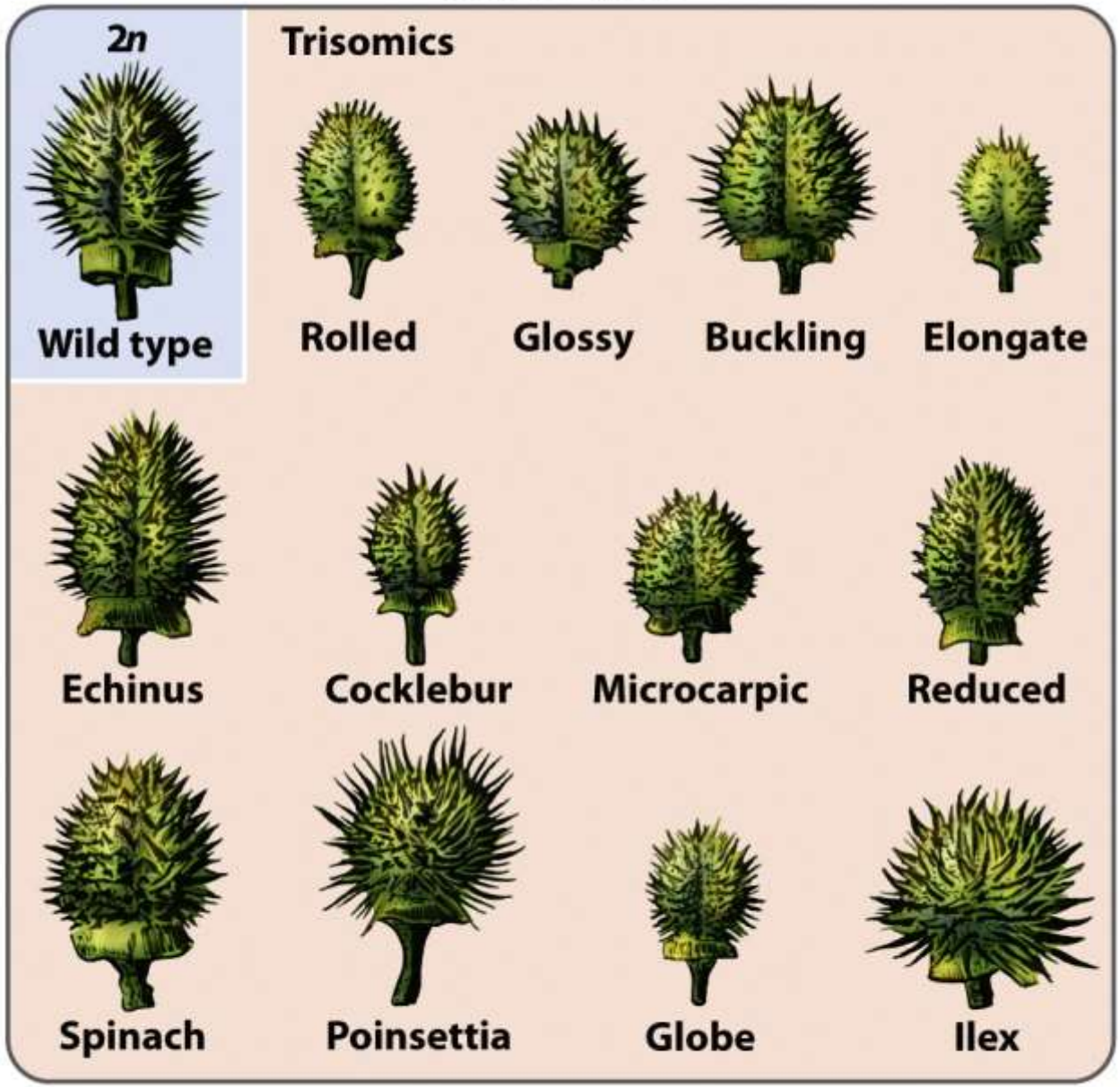
- **Causes of Aneuploidy:**
  - Deletion of centromere during mitosis and meiosis
  - Robertsonian translocation
  - Nondisjunction during meiosis and mitosis

Types of Aneuploidy

- **Nullisomy:** loss of both members of a homologous pair of chromosomes. $2n - 2$
- **Monosomy:** loss of a single chromosome. $2n - 1$
- **Trisomy:** gain of a single chromosome. $2n + 1$
- **Tetrasomy:** gain of two homologous chromosomes. $2n + 2$

Effects of Aneuploidy in Plants

Seed cases



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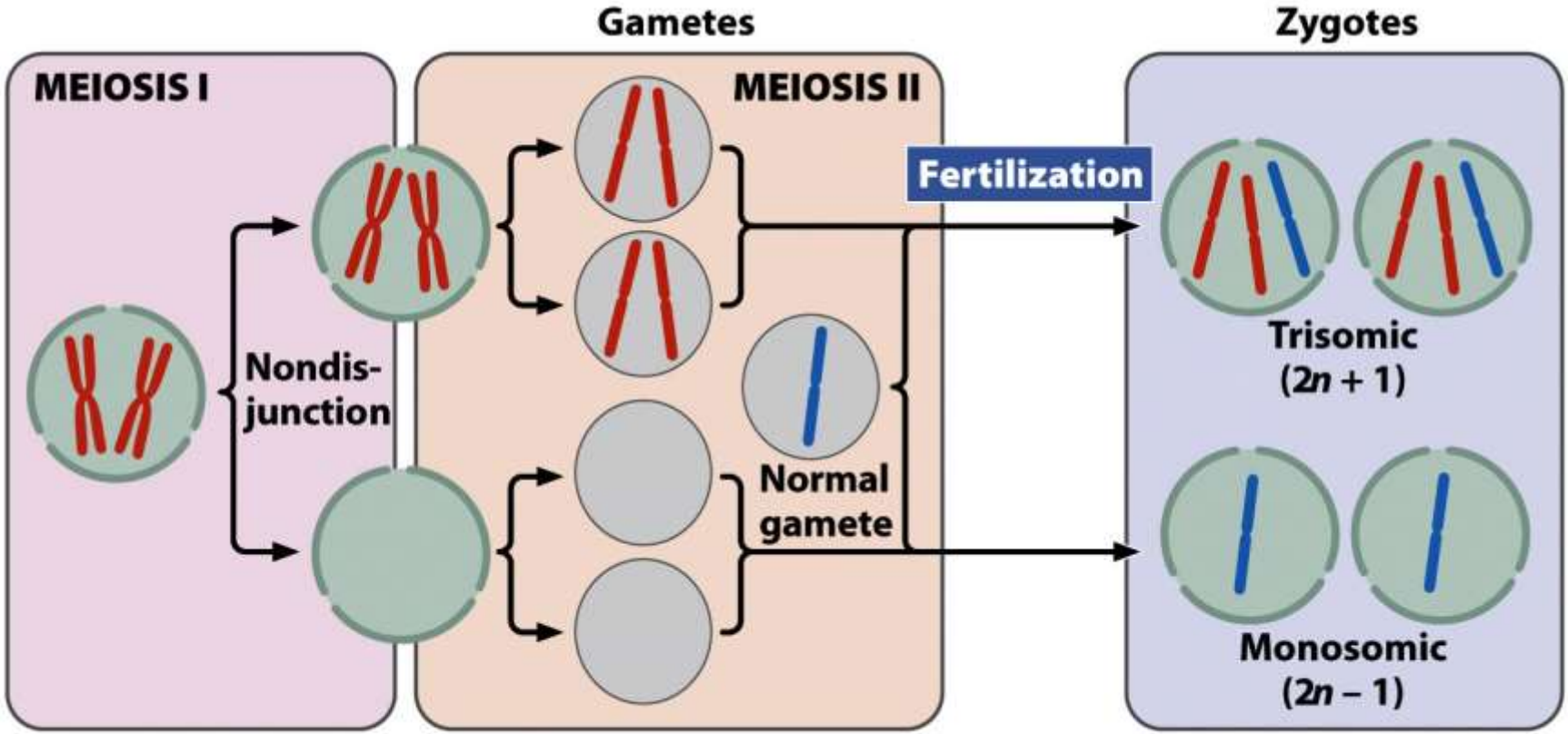
Effects of Aneuploidy

- Drastically altered phenotype
- Due to gene dosage
- Exception: X chromosome in mammals

Production: Nondisjunction in Meiosis I



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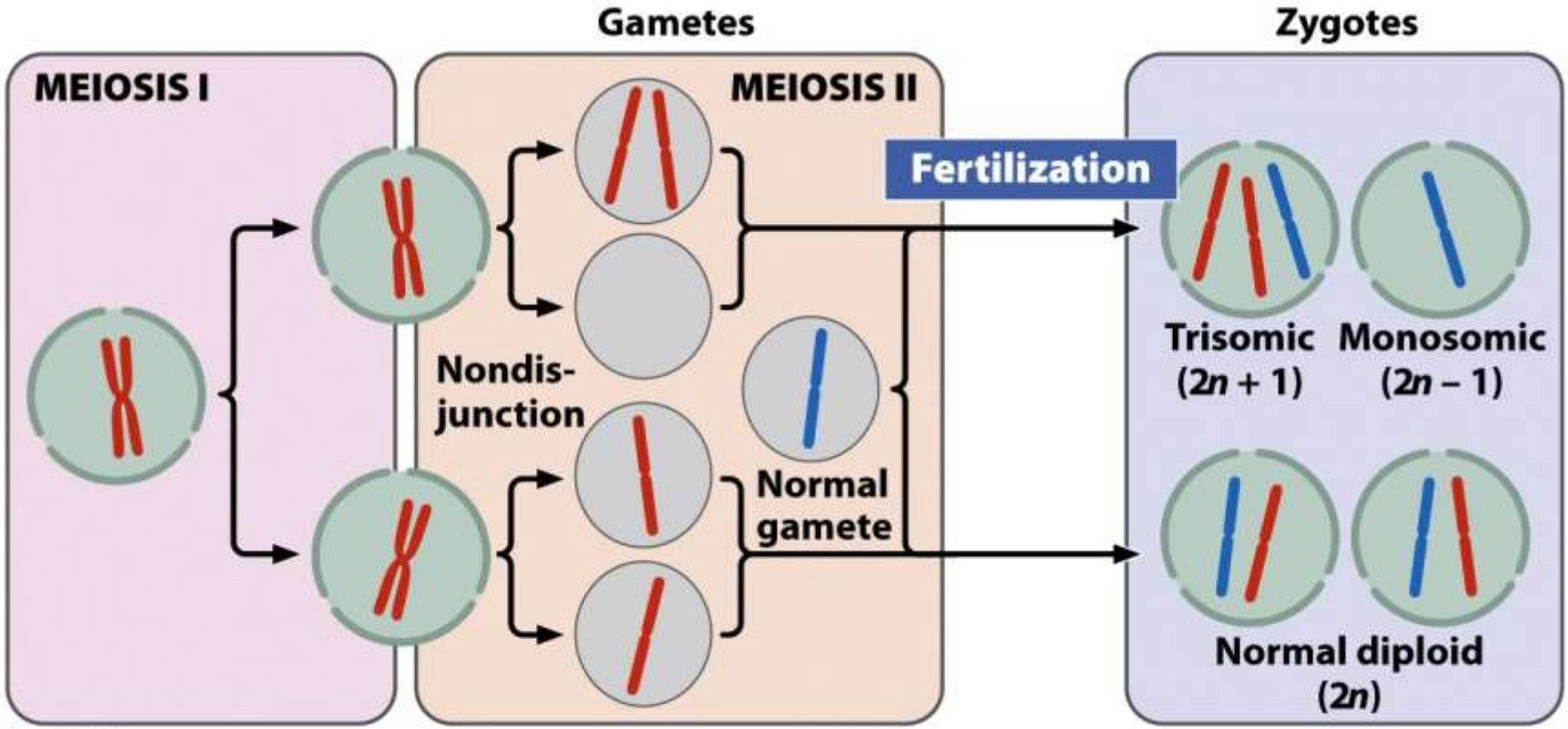
Production: Nondisjunction in Meiosis II



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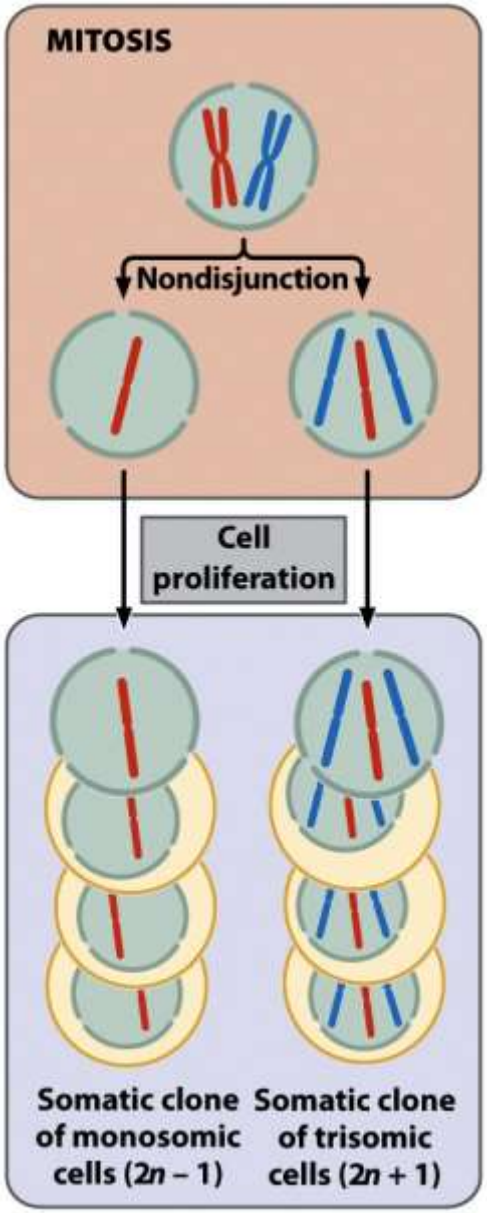

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Production: Nondisjunction in Mitosis

Effects of Aneuploidy in Humans

- **Autosomal aneuploids:**
  - Trisomy 21 – Down syndrome
    - Primary Down syndrome, 75% random nondisjunction in egg formation
    - Familial Down syndrome, Robertsonian translocation between chromosomes 14 and 21

Effects of Aneuploidy in Humans

- **Autosomal aneuploids:**
  - Trisomy 18 – Edward syndrome, 1/8000 live births
  - Trisomy 13 – Patau syndrome, 1/15,000 live births
  - Trisomy 8 – 1/25,000 ~ 1/50,000 live births
  - Why is there a drastic decrease in frequency of these trisomic syndromes from chromosome 18 to chromosome 8?

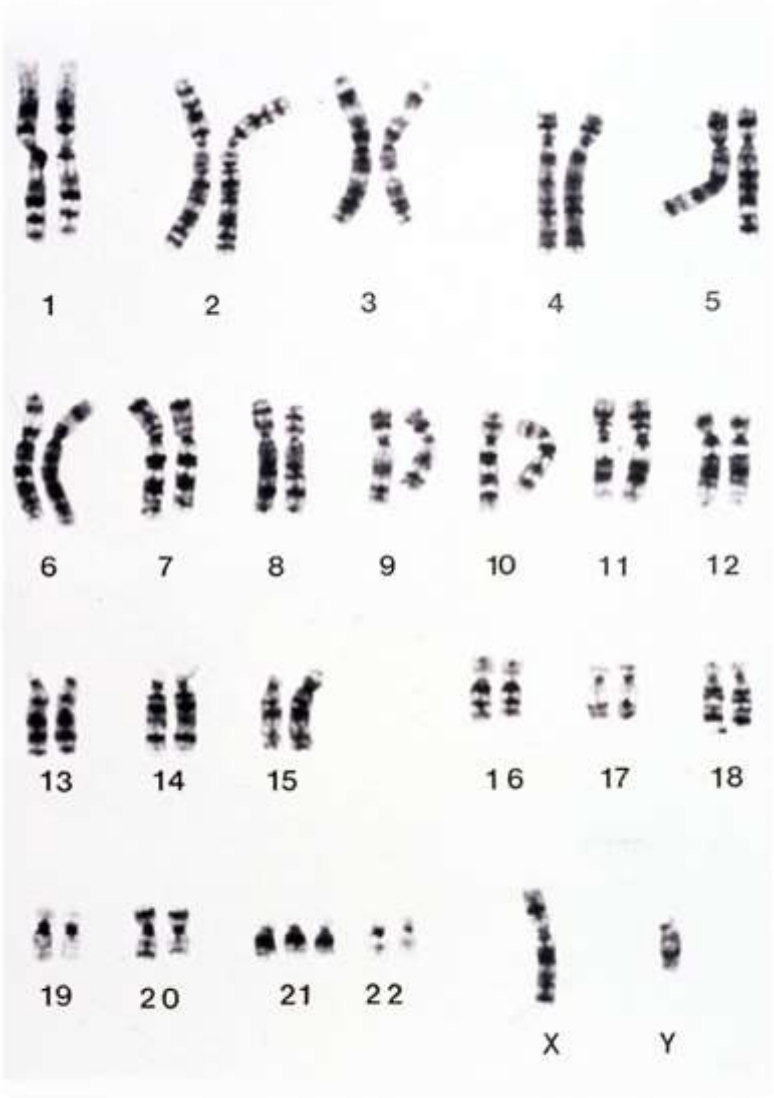
Effects of Aneuploidy in Humans

- **Autosomal aneuploids:**
  - Aneuploidy and maternal age
  - Possible interpretation of this connection
- **Uniparental disomy:** Both chromosomes are inherited from the same parent.
  - Mosaicism and nondisjunction in mitotic division

Effects of Aneuploidy in Humans

- **Sex-chromosome aneuploids:**
  - Turner syndrome. XO
  - Klinefelter sydrome. XXY

Primary Down Syndrome



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Familial Down Syndrome

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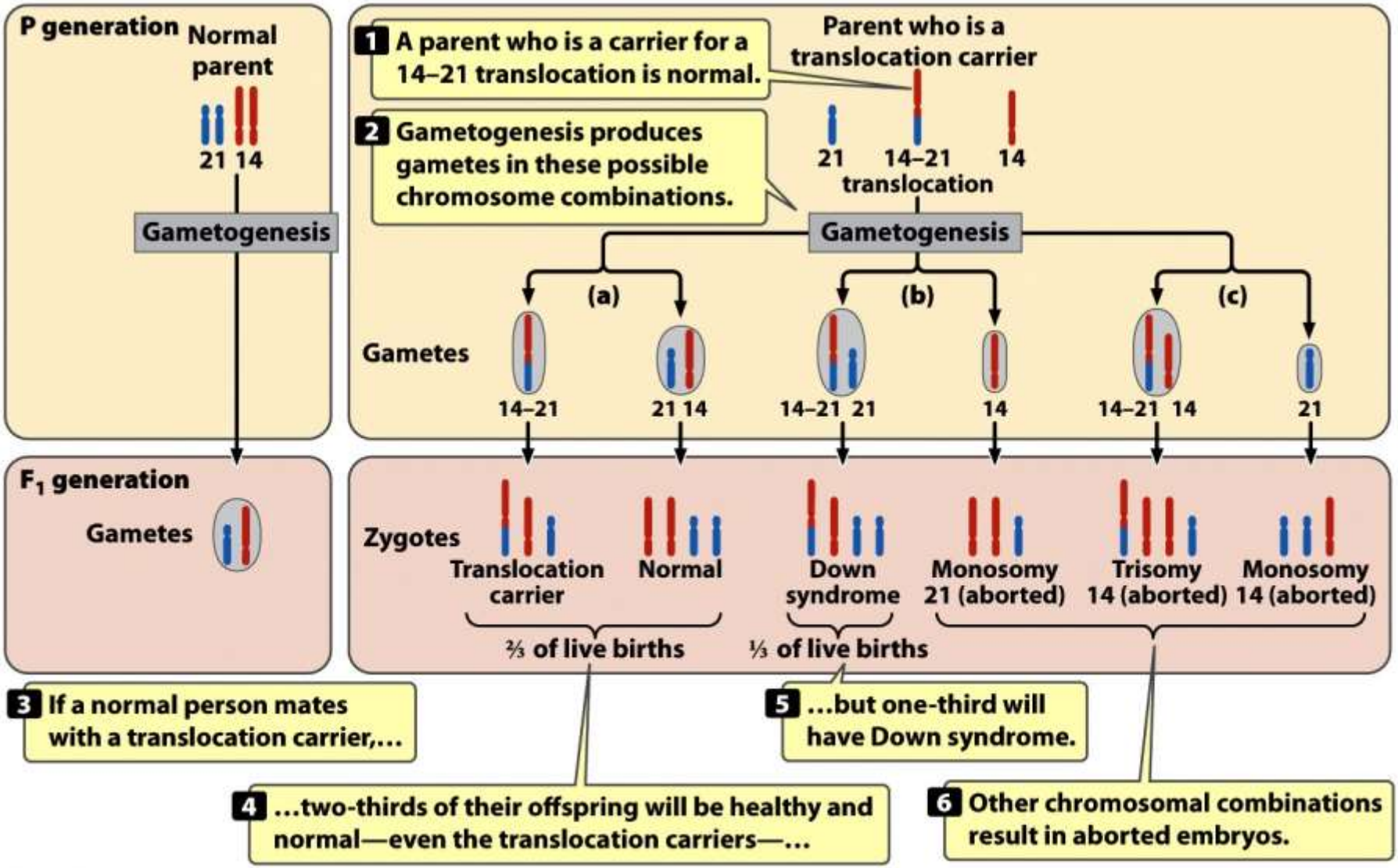

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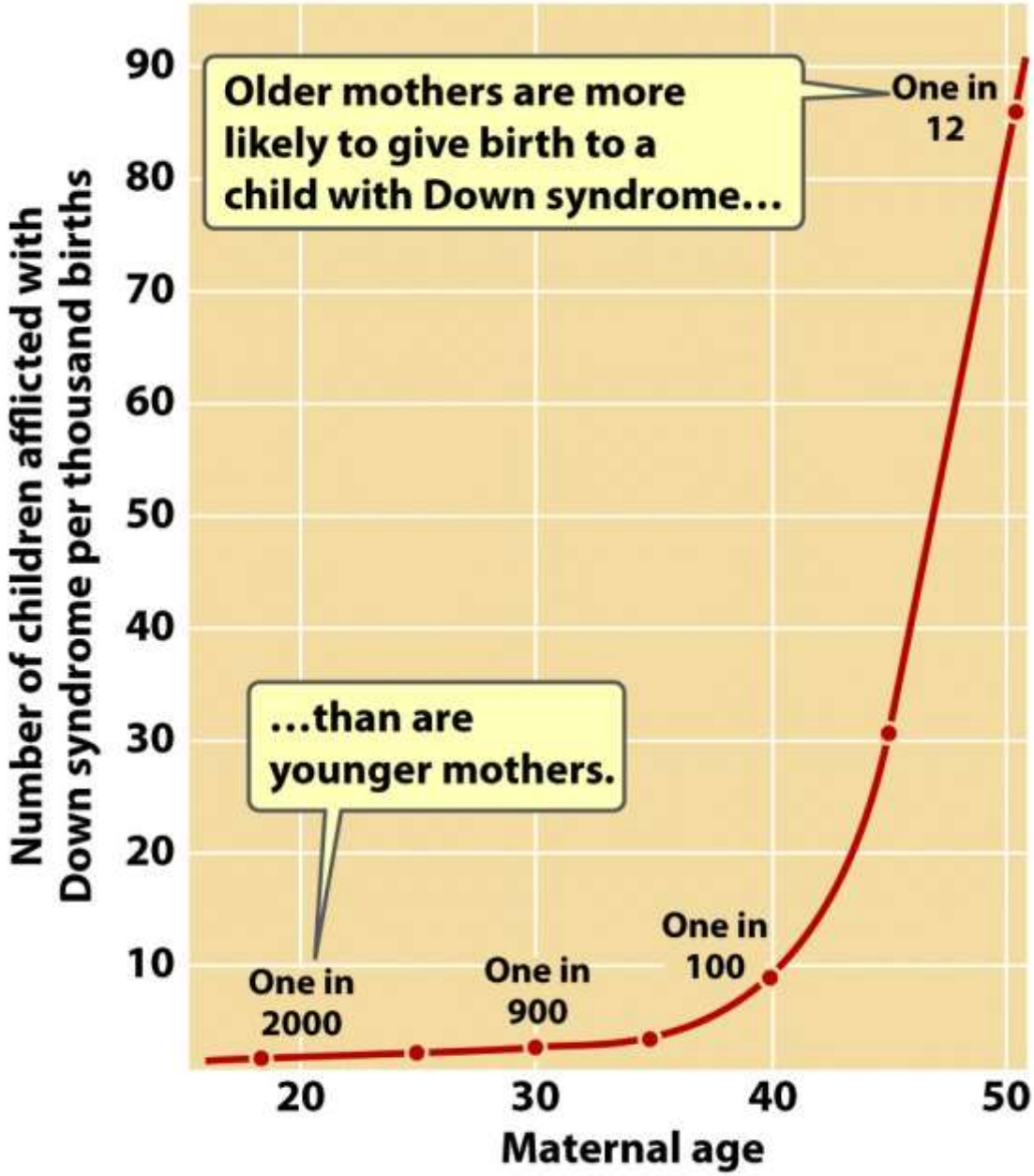

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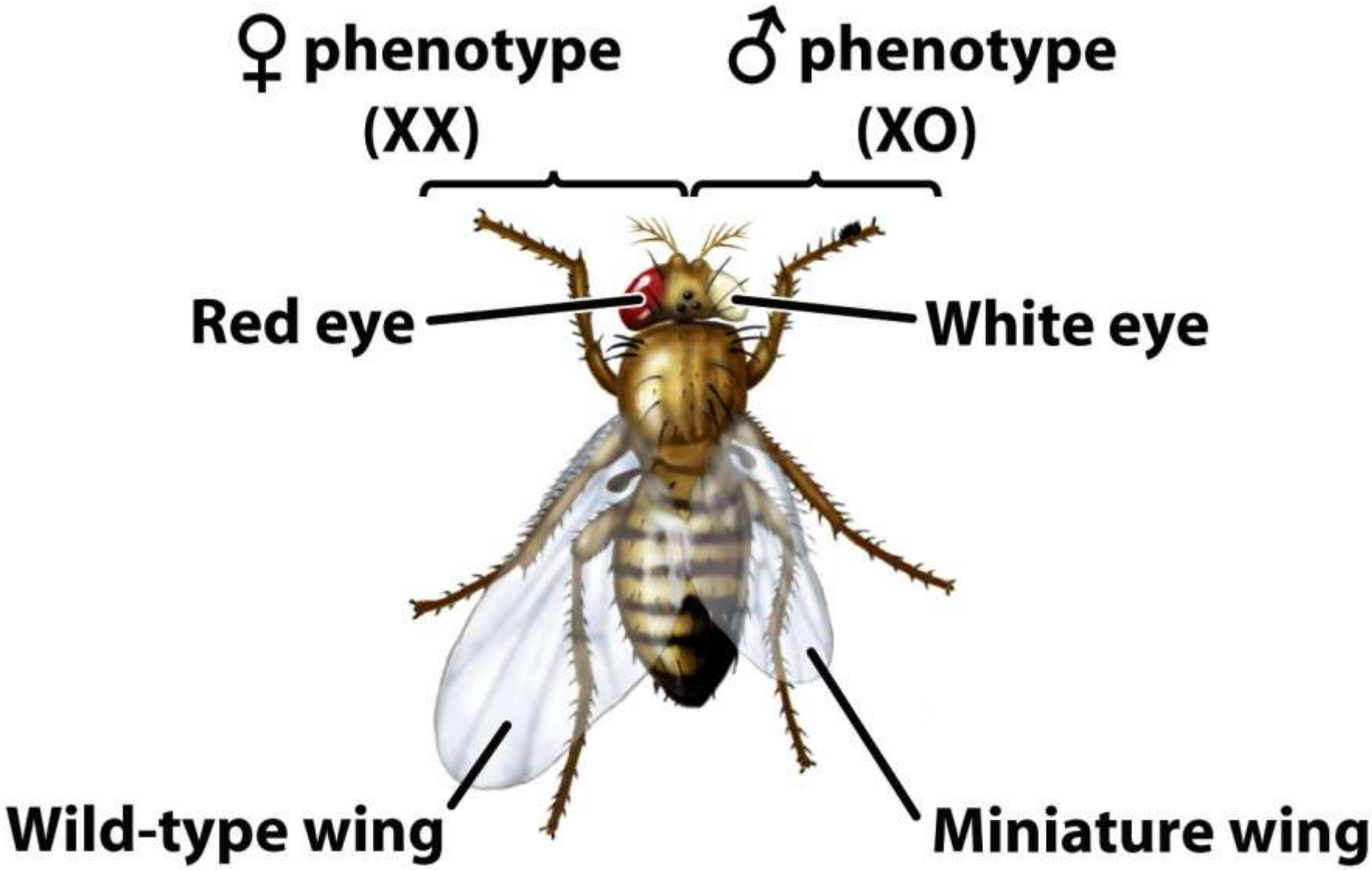

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Polyploidy

Polyploidy Is the Presence of More than Two Sets of Chromosomes

- **Autopolyploidy:**

  From single species

- **Allopolyploidy:**

  From two species

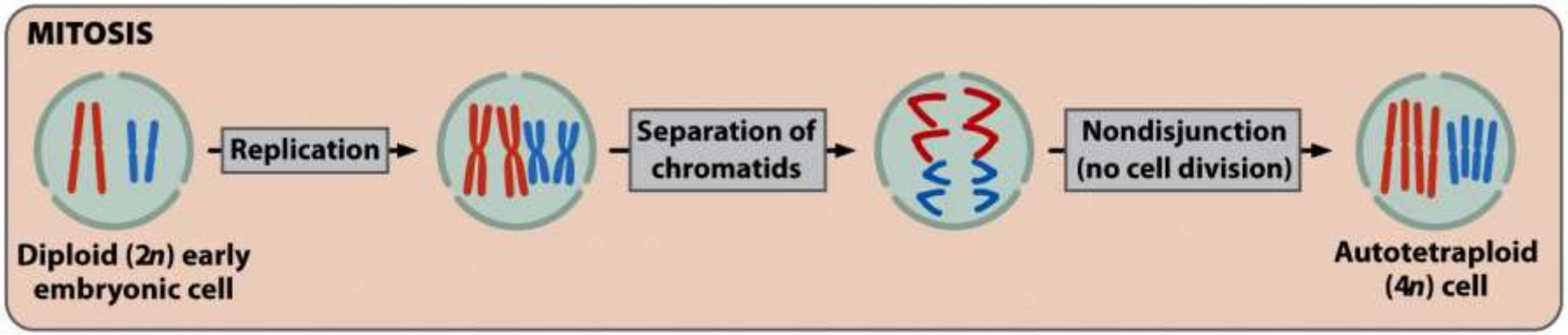

Figure 9-26a
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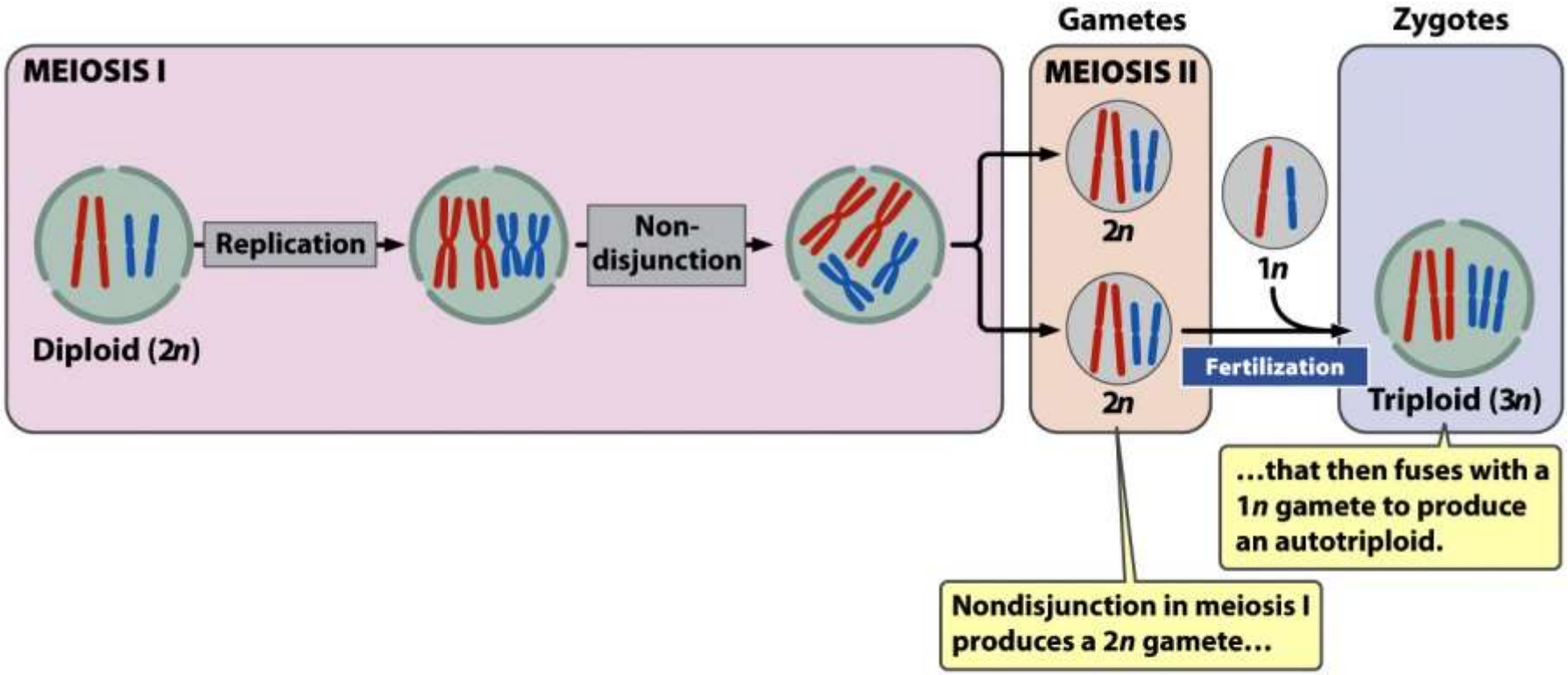

Figure 9-26b
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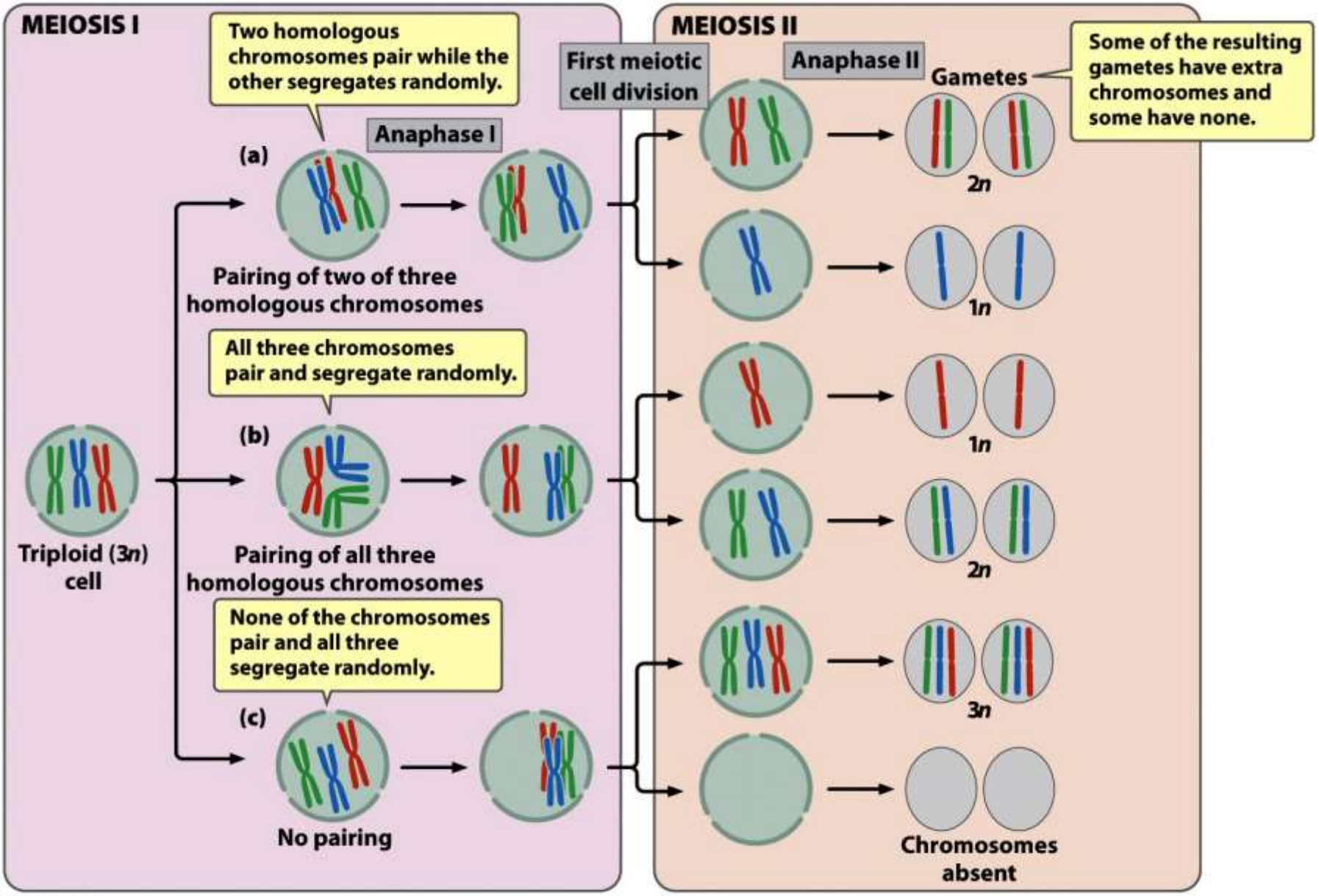

Figure 9-27
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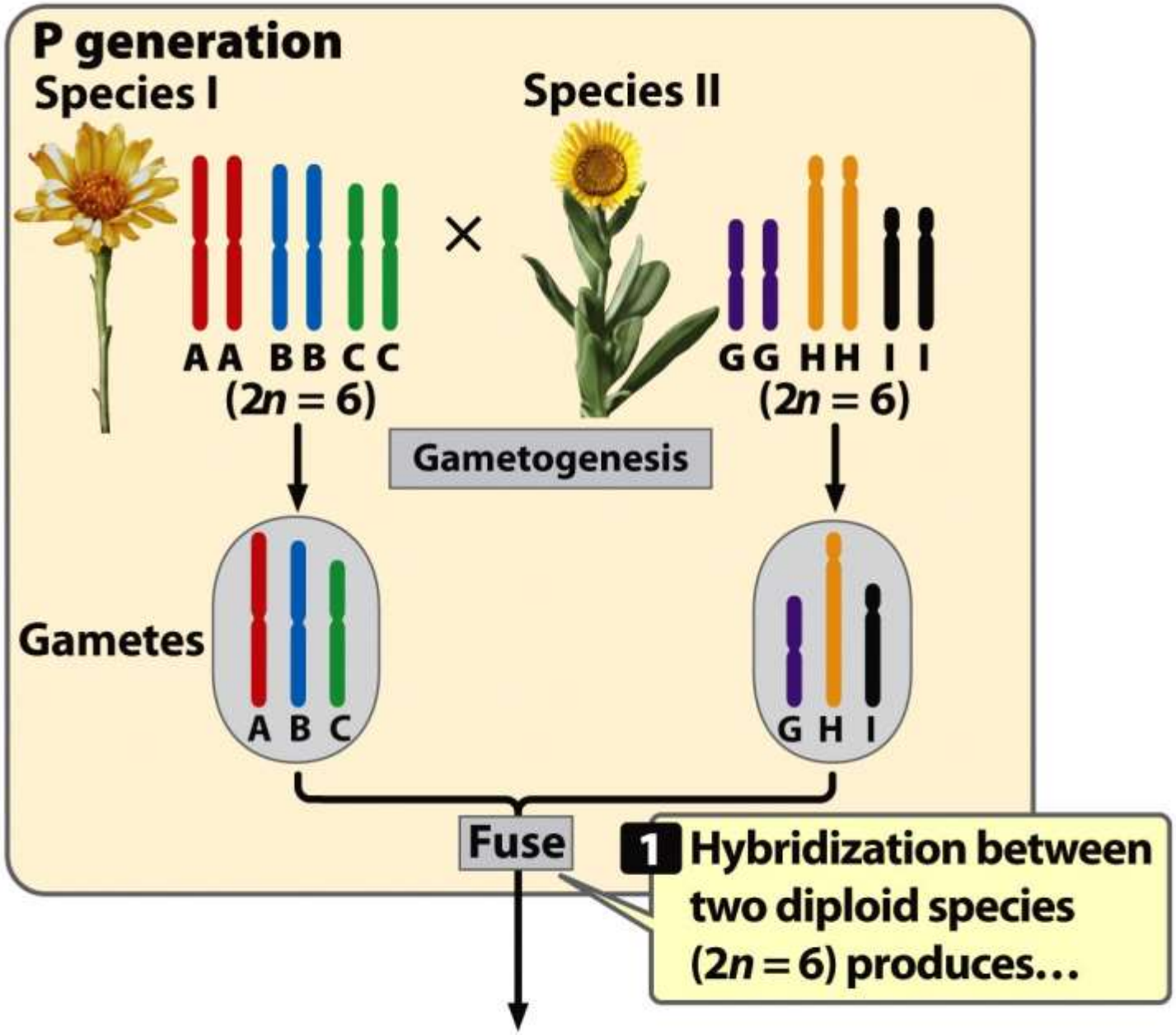

Figure 9-28 part 1
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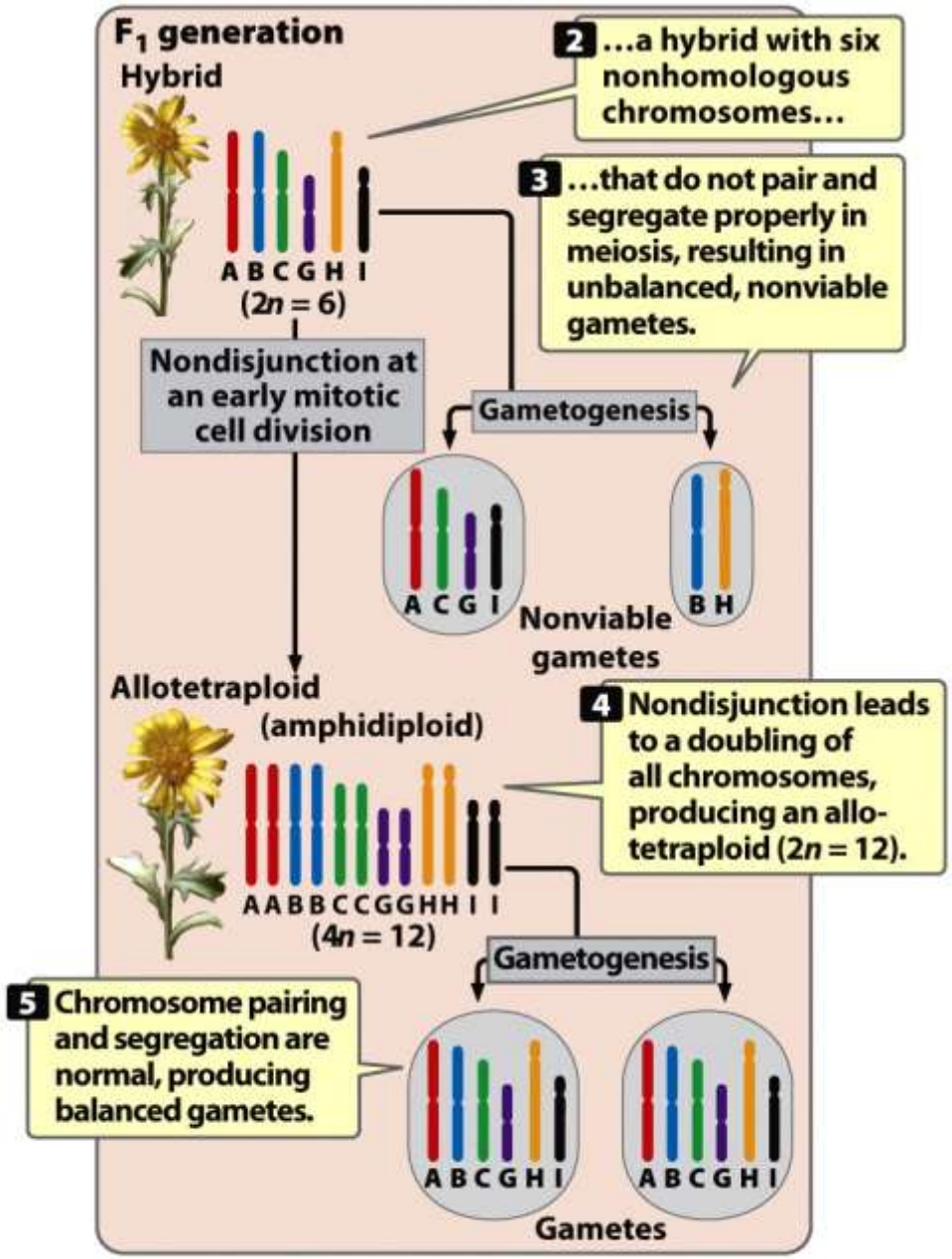

Figure 9-28 part 2

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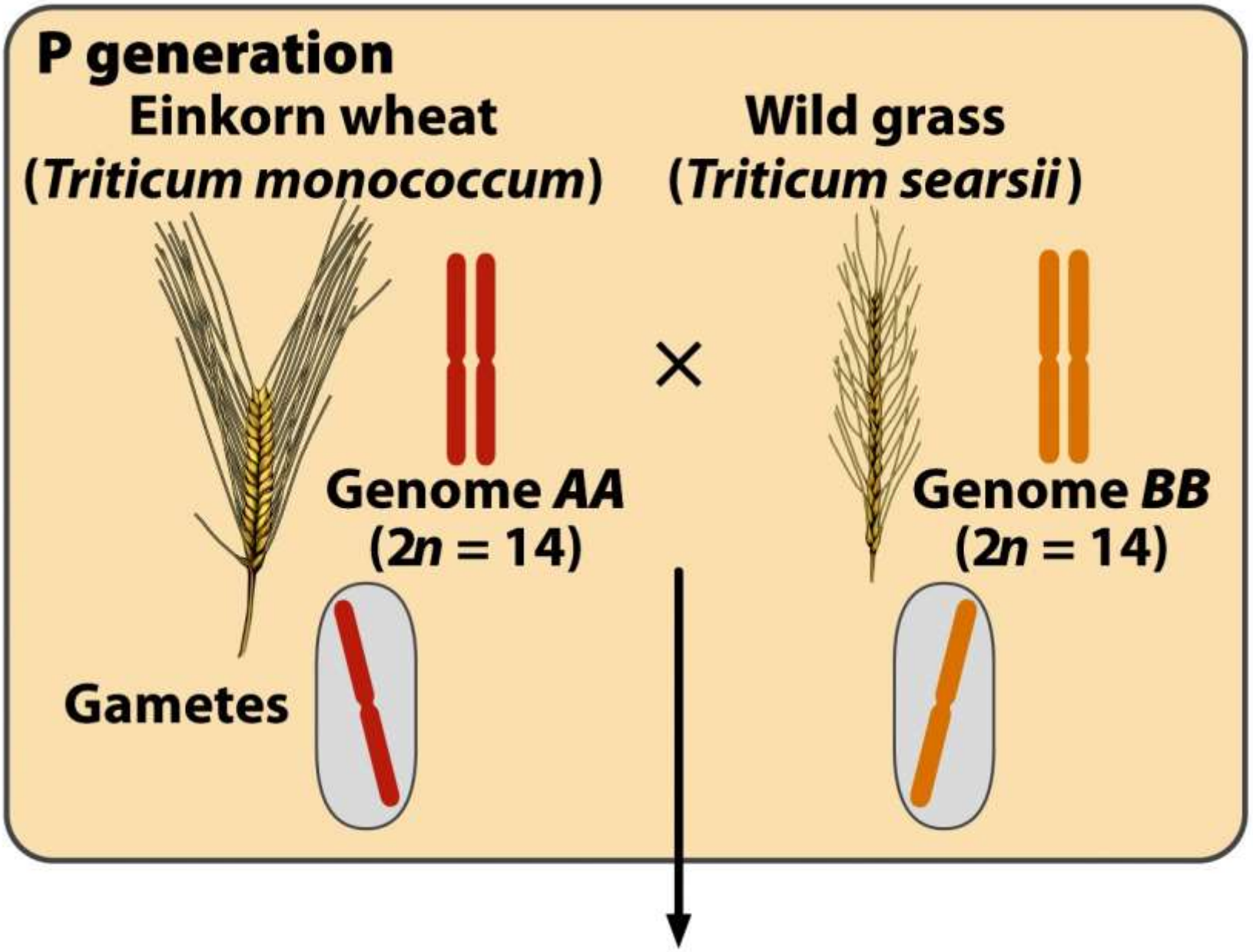

Figure 9-29 part 1
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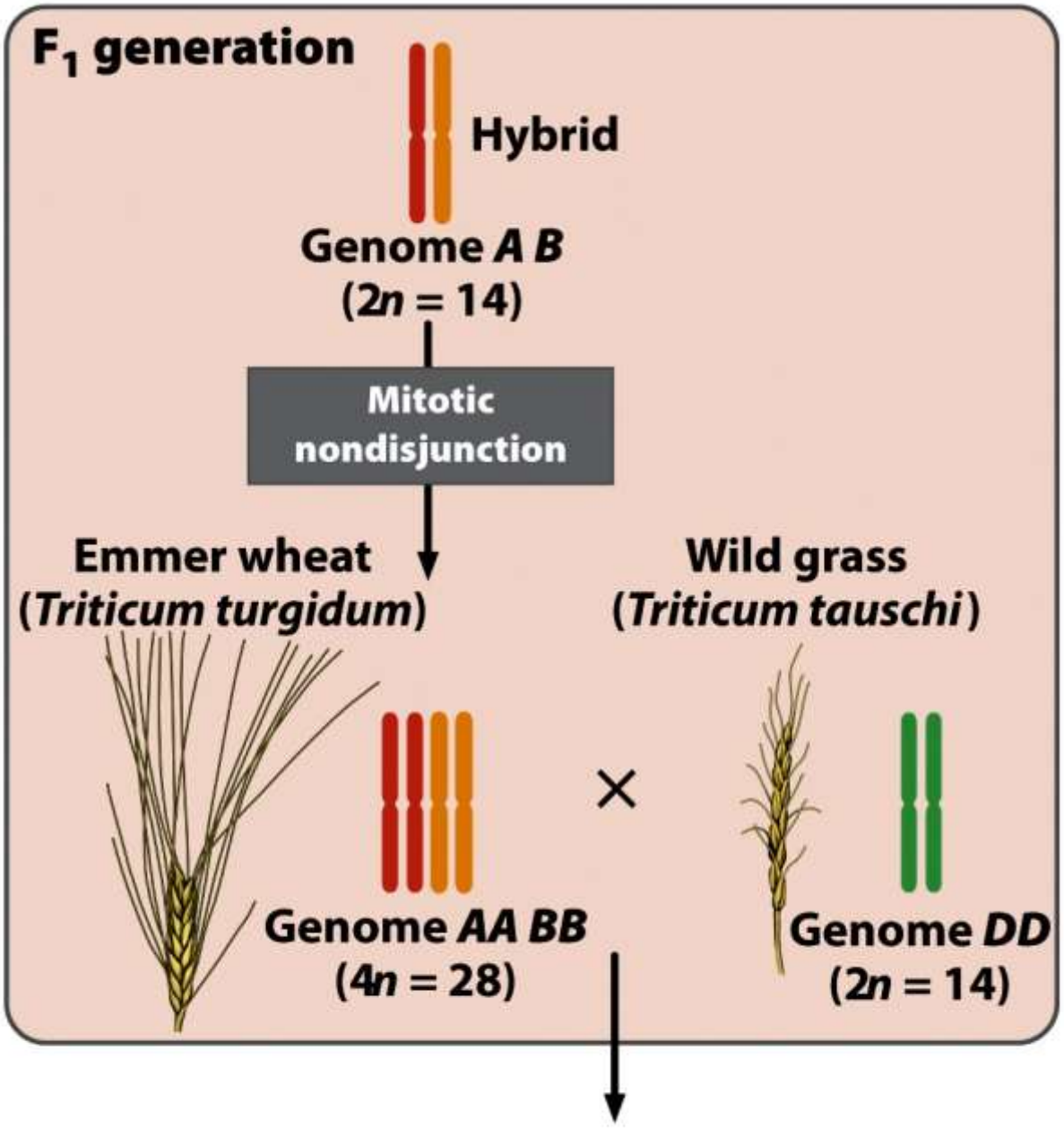

Figure 9-29 part 2
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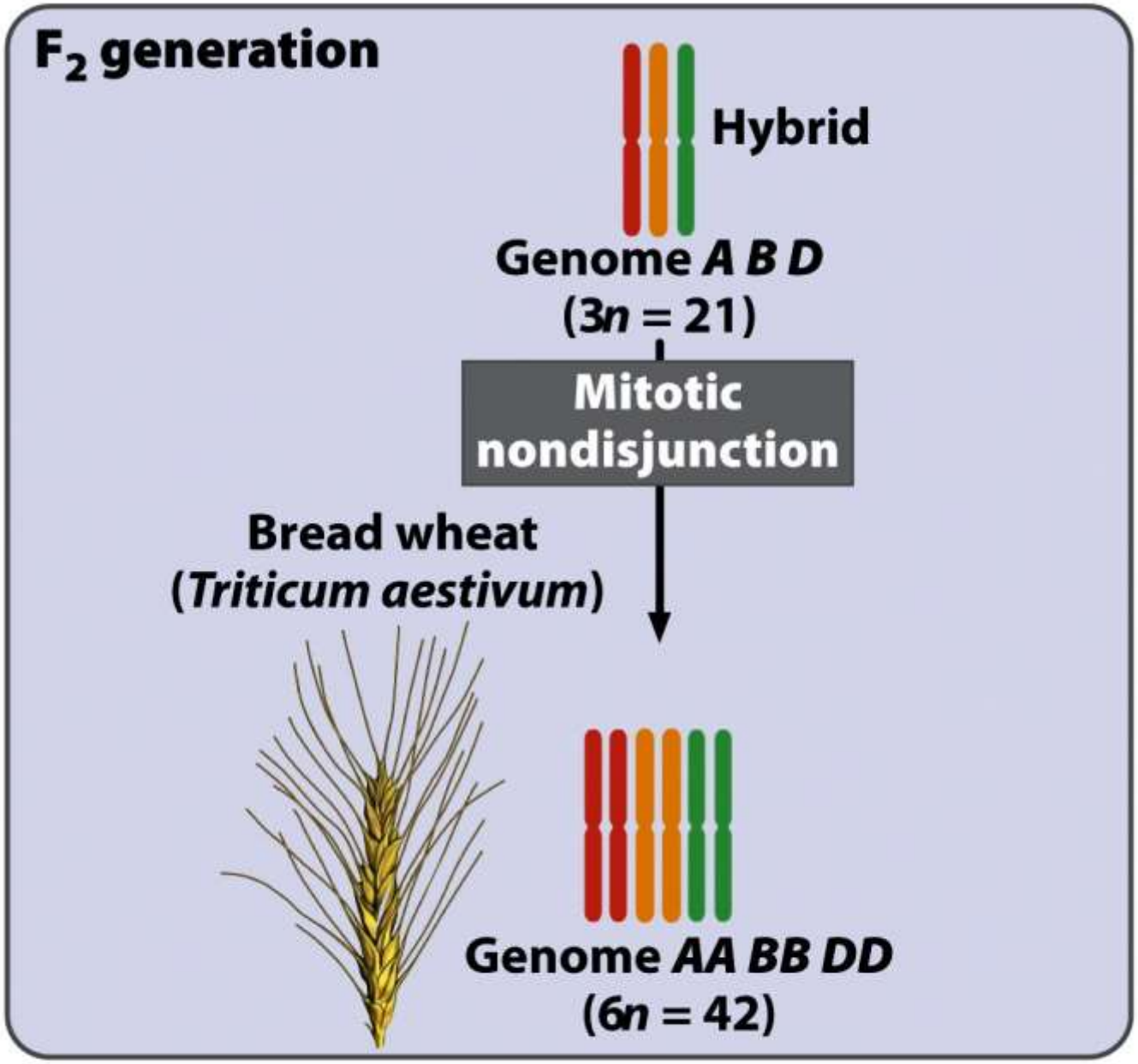

Figure 9-29 part 3
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The Significance of Polyploidy

Table 9.2 Examples of polyploid crop plants

Plant	Type of Polyploidy	Ploidy	Chromosome Number
Potato	Autopolyploid	4*n*	48
Banana	Autopolyploid	3*n*	33
Peanut	Autopolyploid	4*n*	40
Sweet potato	Autopolyploid	6*n*	90
Tobacco	Allopolyploid	4*n*	48
Cotton	Allopolyploid	4*n*	52
Wheat	Allopolyploid	6*n*	42
Oats	Allopolyploid	6*n*	42
Sugar cane	Allopolyploid	8*n*	80
Strawberry	Allopolyploid	8*n*	56

Source: After F. C. Elliot, *Plant Breeding and Cytogenetics* (New York: McGraw-Hill, 1958).

Table 9-2
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Table 9.3 Different types of chromosome mutations

Chromosome Mutation	Definition
Chromosome rearrangement	Change in chromosome structure
Chromosome duplication	Duplication of a chromosome segment
Chromosome deletion	Deletion of a chromosome segment
Inversion	Chromosome segment inverted 180 degrees
Paracentric inversion	Inversion that does not include the centromere in the inverted region
Pericentric inversion	Inversion that includes the centromere in the inverted region
Translocation	Movement of a chromosome segment to a nonhomologous chromosome or to another region of the same chromosome
Nonreciprocal translocation	Movement of a chromosome segment to a nonhomologous chromosome or to another region of the same chromosome without reciprocal exchange
Reciprocal translocation	Exchange between segments of nonhomologous chromosomes or between regions of the same chromosome
Aneuploidy	Change in number of individual chromosomes
Nullisomy	Loss of both members of a homologous pair
Monosomy	Loss of one member of a homologous pair
Trisomy	Gain of one chromosome, resulting in three homologous chromosomes
Tetrasomy	Gain of two homologous chromosomes, resulting in four homologous chromosomes
Polyploidy	Addition of entire chromosome sets
Autopolyploidy	Polyploidy in which extra chromosome sets are derived from the same species
Allopolyploidy	Polyploidy in which extra chromosome sets are derived from two or more species

Table 9-3
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Chromosome Variation Plays an Important Role in Evolution

- New and extra copies of genes give rise to new functions.
- New and extra sets of genes may give rise to new species.

Summary