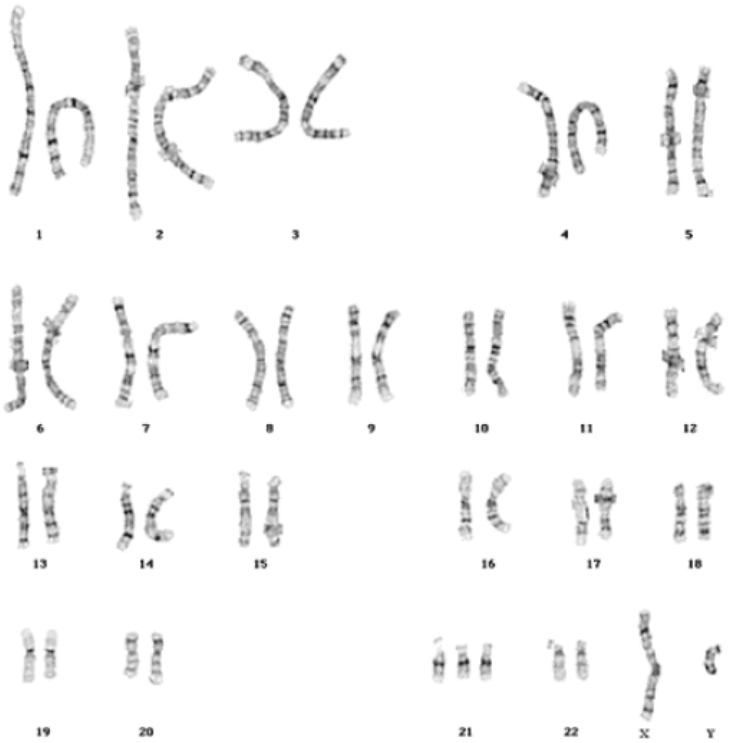
# TEMA 7

## Alteraciones Cromosómicas



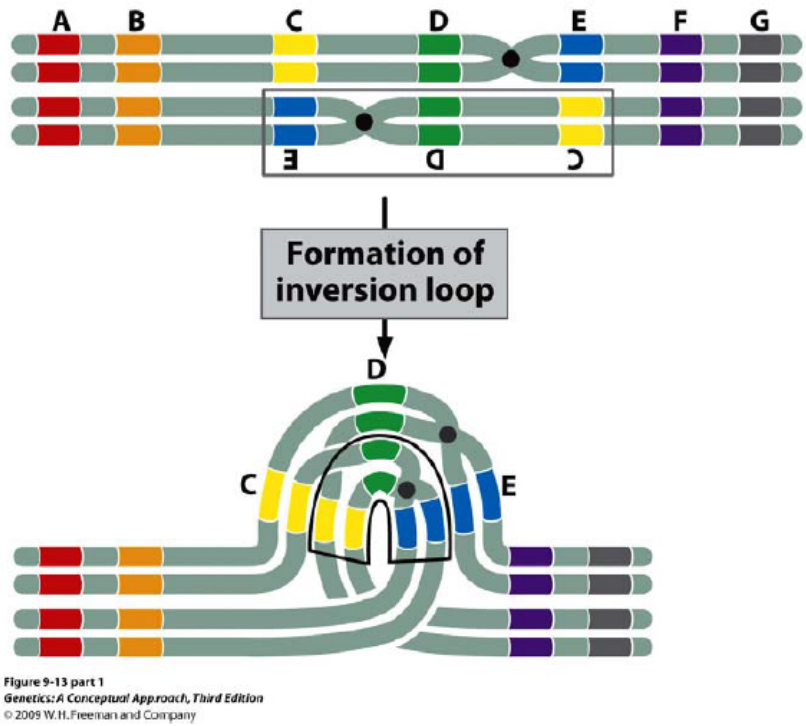




Figure 9-13 part 1
*Genetics: A Conceptual Approach, Third Edition*

# Morfología Cromosómica

Metacentric

Acrocentric

Submetacentric

Telocentric

# Morfología Cromosómica

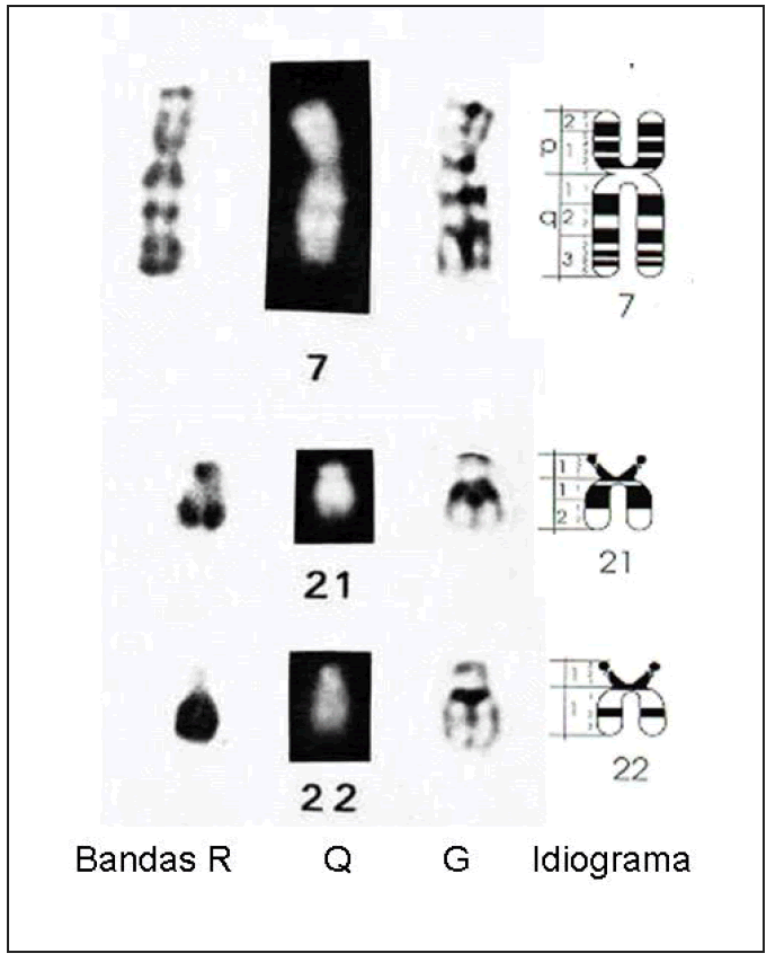
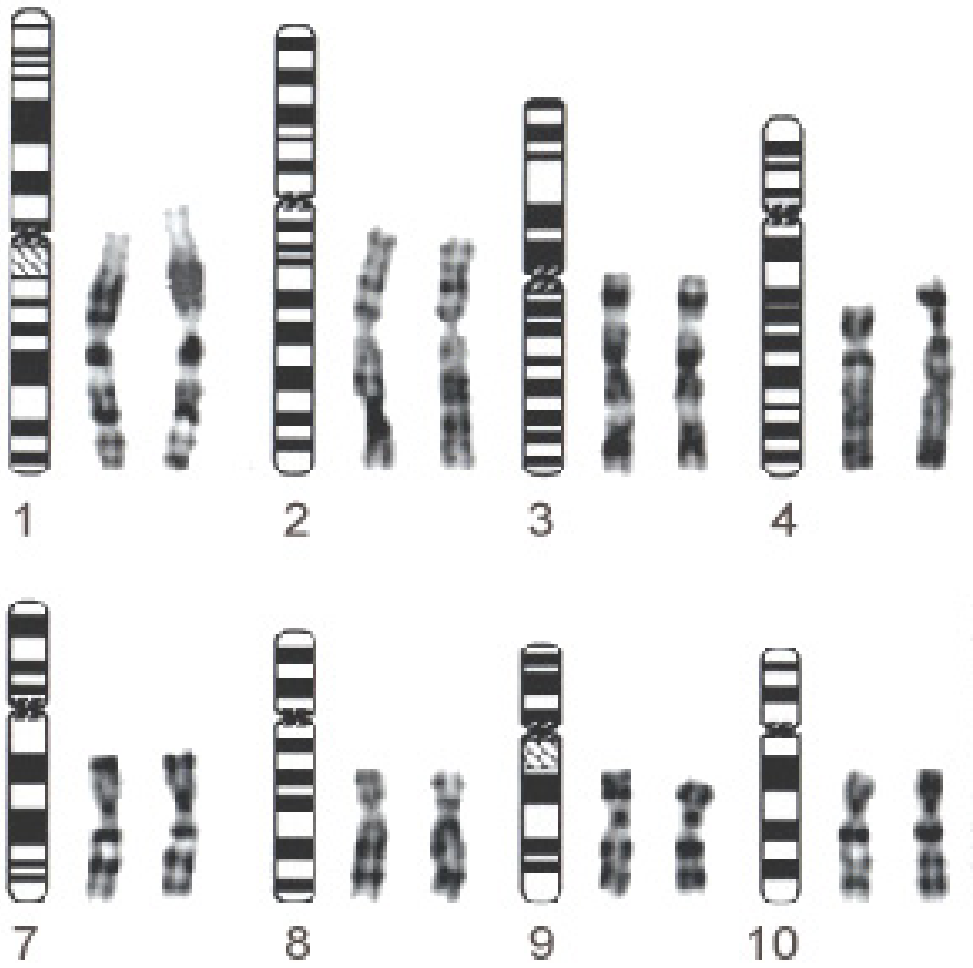
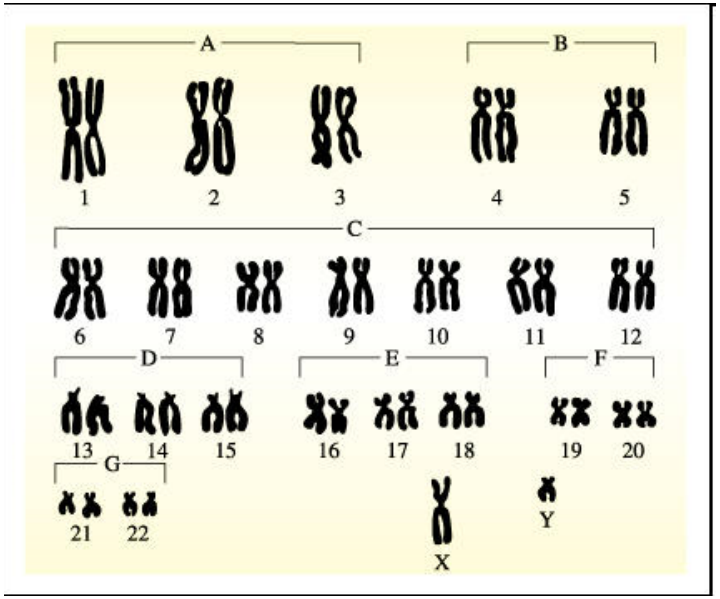



**Figura 2.** *Cromosomas 7, 21 y 22 en bandas R, bandas Q y bandas G, se comparan con un idiograma en bandas G. Se puede ver la diferencia entre el patrón de bandeamiento entre las G y las bandas R, y el mismo patrón entre las bandas G y las bandas Q.*

# Morfología Cromosómica



# Morfología Cromosómica



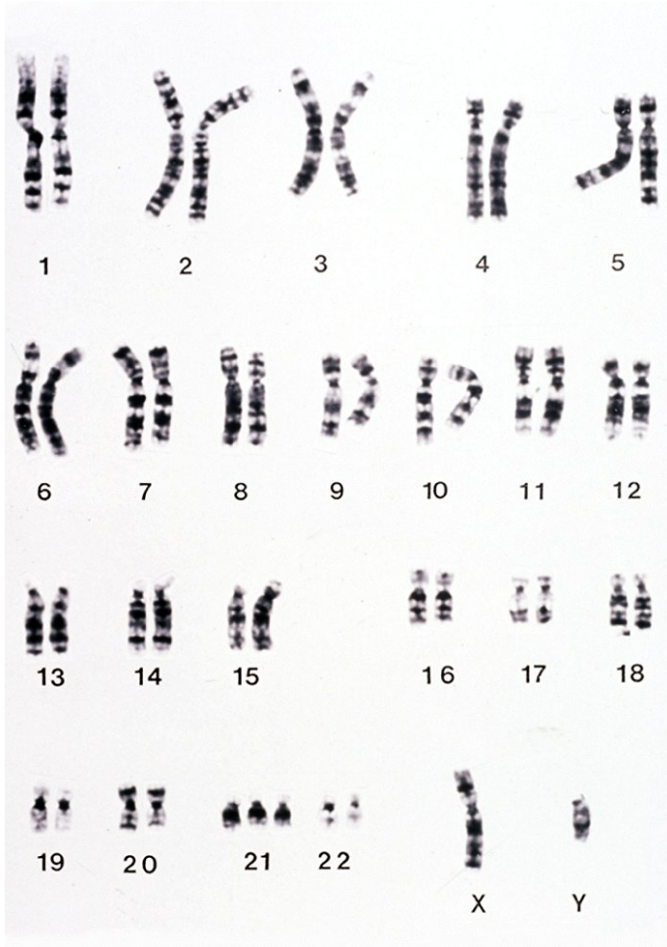




**Figure 9-21**
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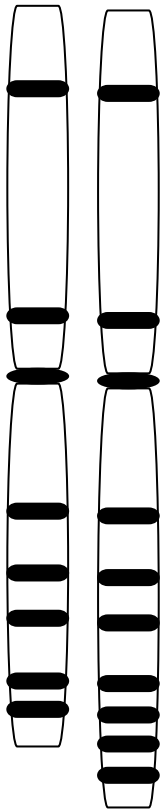
**En tándem, desplazada, directa o invertida.**

# Duplicaciones



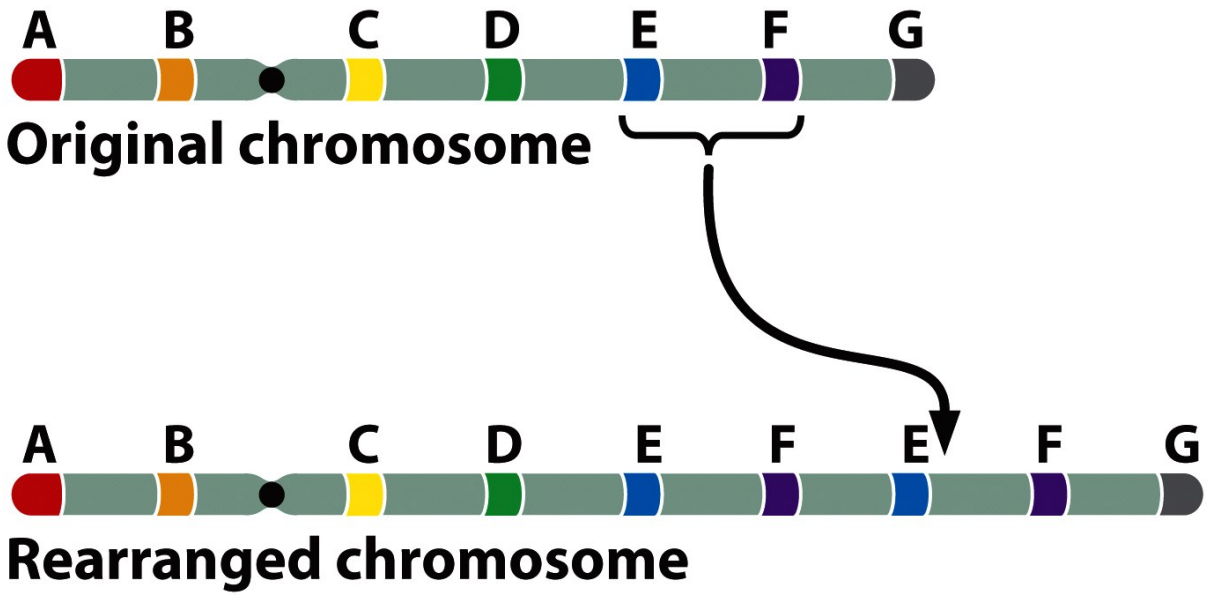



Figure 9-4a
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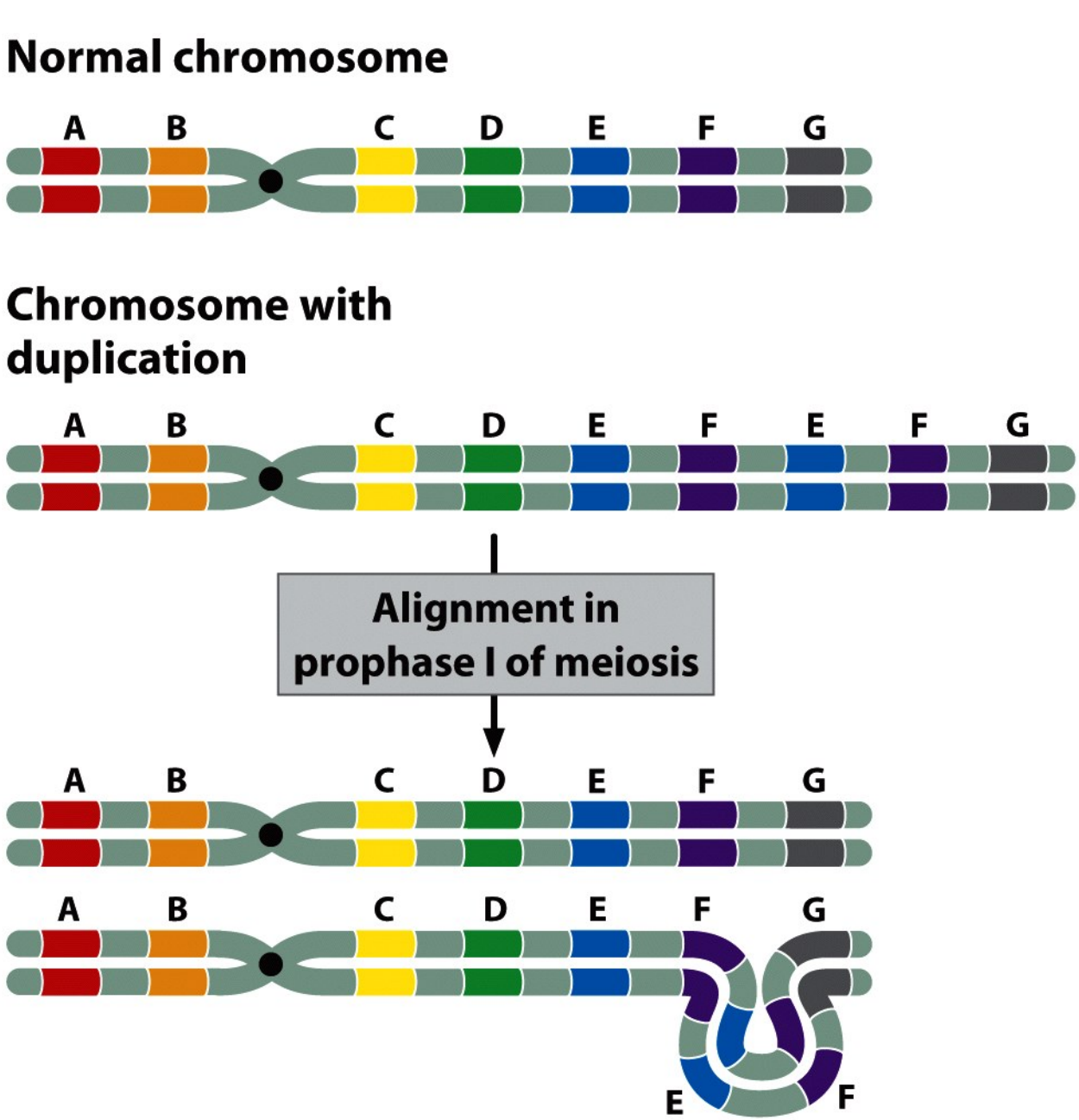


**Figure 9-5**
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(a)
Wild type
female
$B^+B^+$

*Bar* region

(b)
Heterozygous *Bar*
female
$B^+B$

(c)
Homozygous *Bar*
*female*
***BB***

(d)
Heterozygous
*double Bar*
female
$B^+B^D$

**Figure 9-6**
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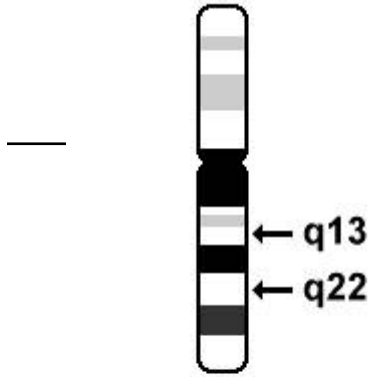
46,XX,del(16)(q13q22)

# Deleciones


**Desequilibrio producto génico, pseudodominancia o haploinsuficiencia**




Cromosoma 16 normal

Cromosoma 16 con deleción

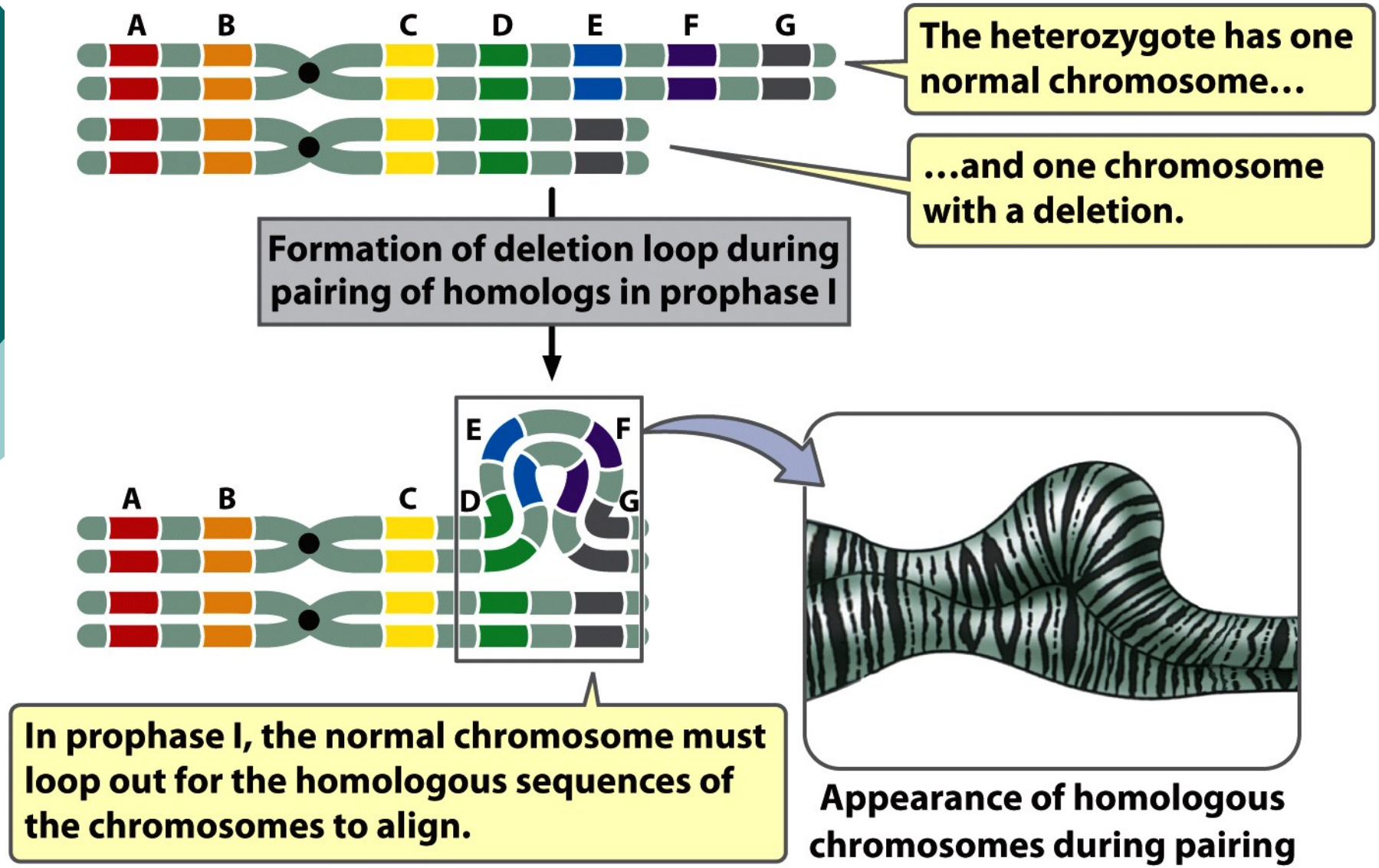



**Figure 9-9**
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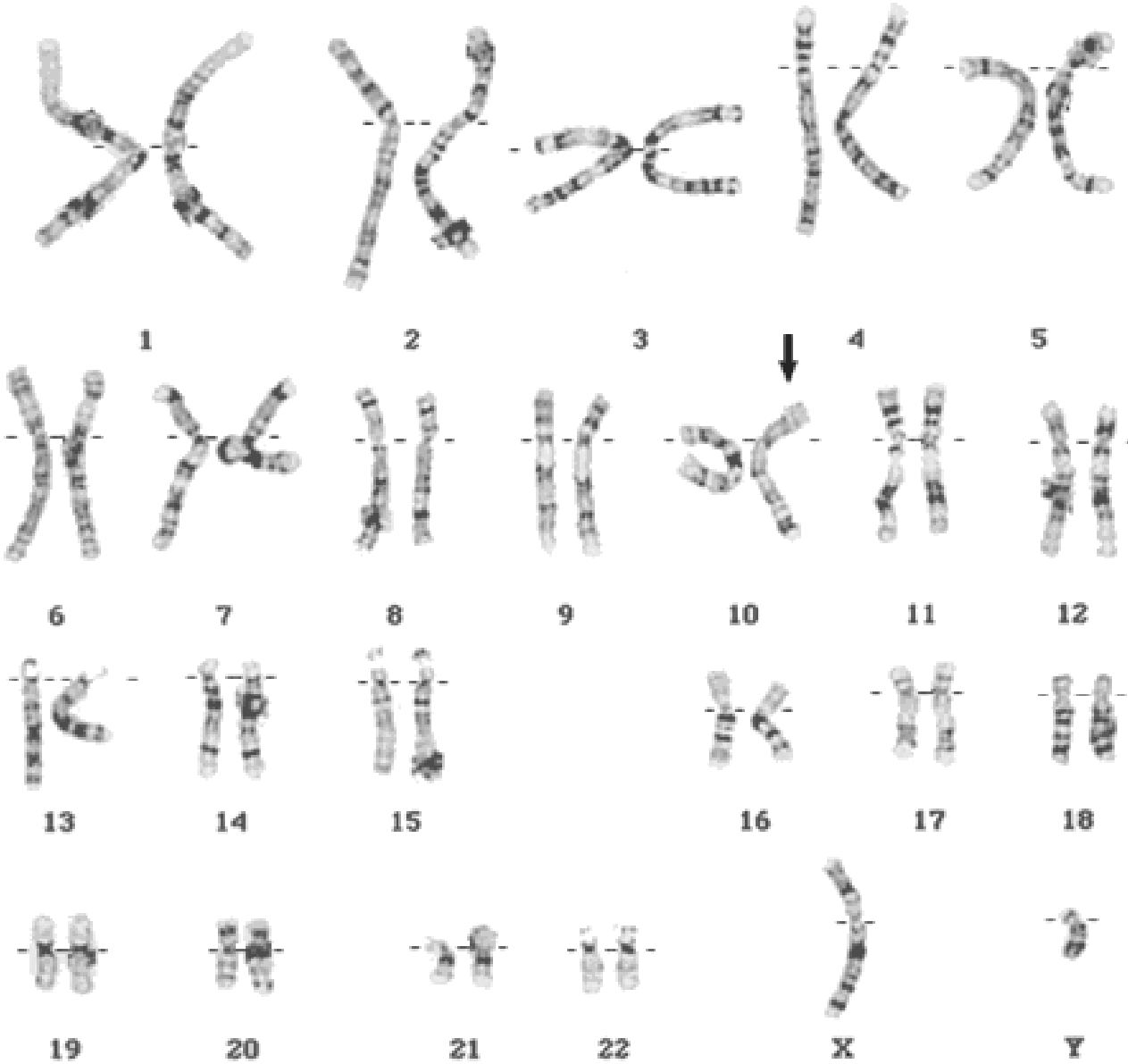
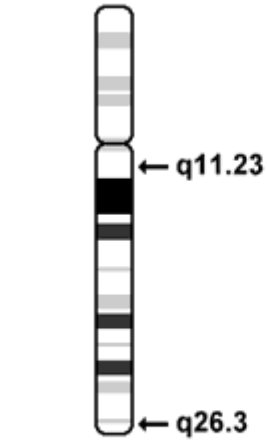
# Efectos Duplicaciones y Deleciones

**Cuadro 9-1 Efectos de algunos reordenamientos cromosómicos humanos**

| Tipo de reordenamiento | Cromosoma | Trastorno | Manifestaciones |
|---|---|---|---|
| Duplicación | 4, brazo corto | — | Cabeza pequeña, cuello corto, nacimiento del cabello bajo, retraso mental y del crecimiento. |
| Duplicación | 4, brazo largo | — | Cabeza pequeña, frente inclinada, anomalía en las manos. |
| Duplicación | 7, brazo largo | — | Desarrollo retardado, asimetría de la cabeza, cuero cabelludo velloso, nariz pequeña, orejas de inserción baja. |
| Duplicación | 9, brazo corto | — | Rostro característico, retraso mental variable, frente alta y ancha, anomalía en las manos. |
| Deleción | 5, brazo corto | Síndrome del *maullido de gato* | Cabeza pequeña, llanto característico, ojos muy separados, cara redonda, retraso mental. |
| Deleción | 4, brazo corto | Síndrome de Wolf-Hirschhorn | Cabeza pequeña con la frente alta, nariz ancha, labio leporino y paladar hendido, retraso mental grave. |
| Deleción | 4, brazo largo | — | Cabeza pequeña, retraso mental leve a moderado, labio leporino y paladar hendido, anomalías en pies y manos. |
| Deleción | 7, brazo largo | síndrome de Williams-Beuren | Características faciales, defectos cardíacos, deficiencia mental. |
| Deleción | 15, brazo largo | Síndrome de Prader-Willi | Dificultades de alimentación en edad temprana, pero con obesidad después del año de edad, retraso mental leve a moderado. |
| Deleción | 18, brazo corto | — | Cara redonda, orejas grandes y de inserción baja, retraso mental leve a moderado. |
| Deleción | 18, brazo largo | — | Forma de la boca característica, manos pequeñas, cabeza pequeña, retraso mental. |

# Inversiones



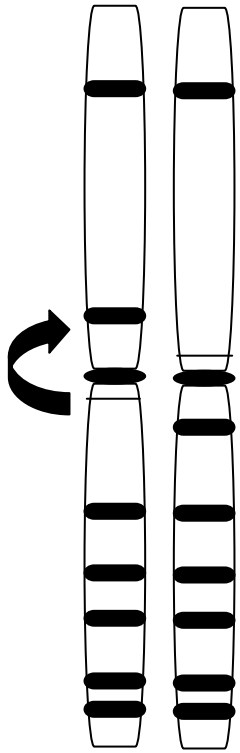




Normal Chromosome 10

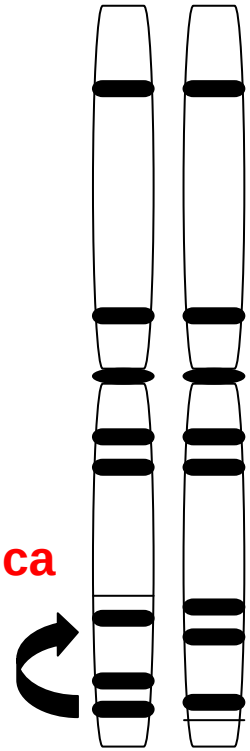
Inverted Chromosome 10

# Inversiones

**Pericéntrica**



**Paracéntrica**



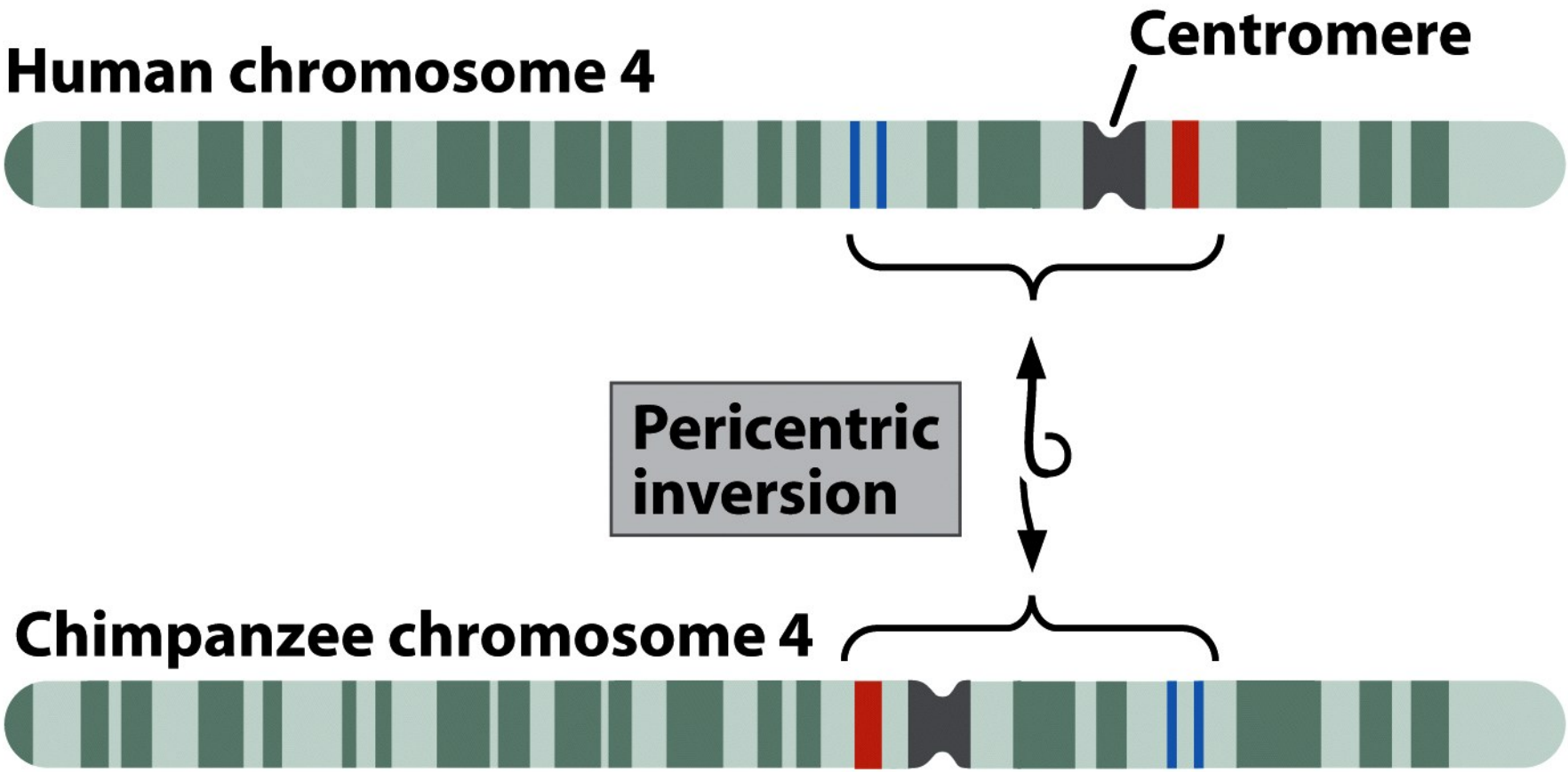



**Figure 9-14**
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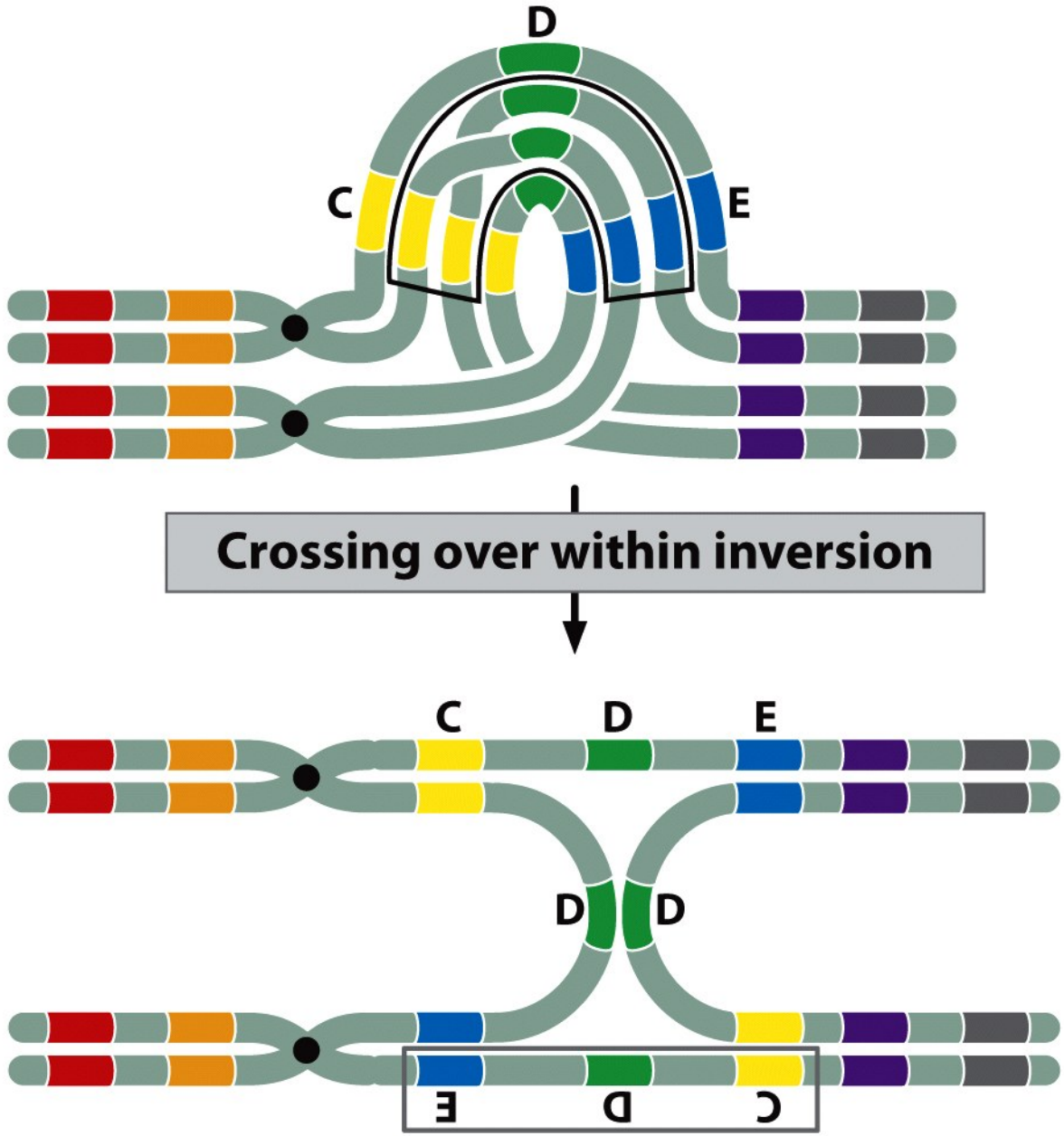



**Figure 9-12 part 2**
***Genetics: A Conceptual Approach, Third Edition***

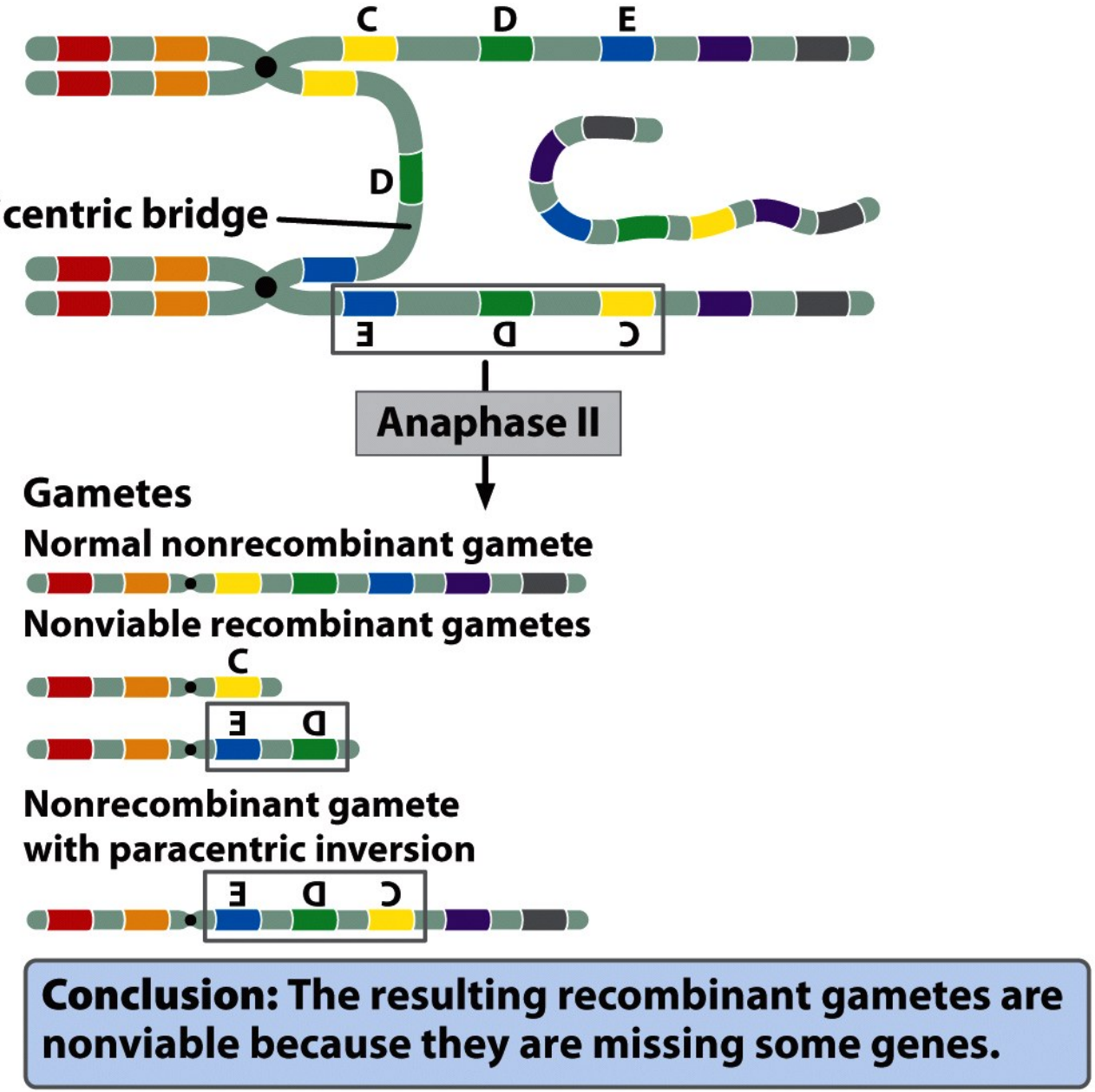
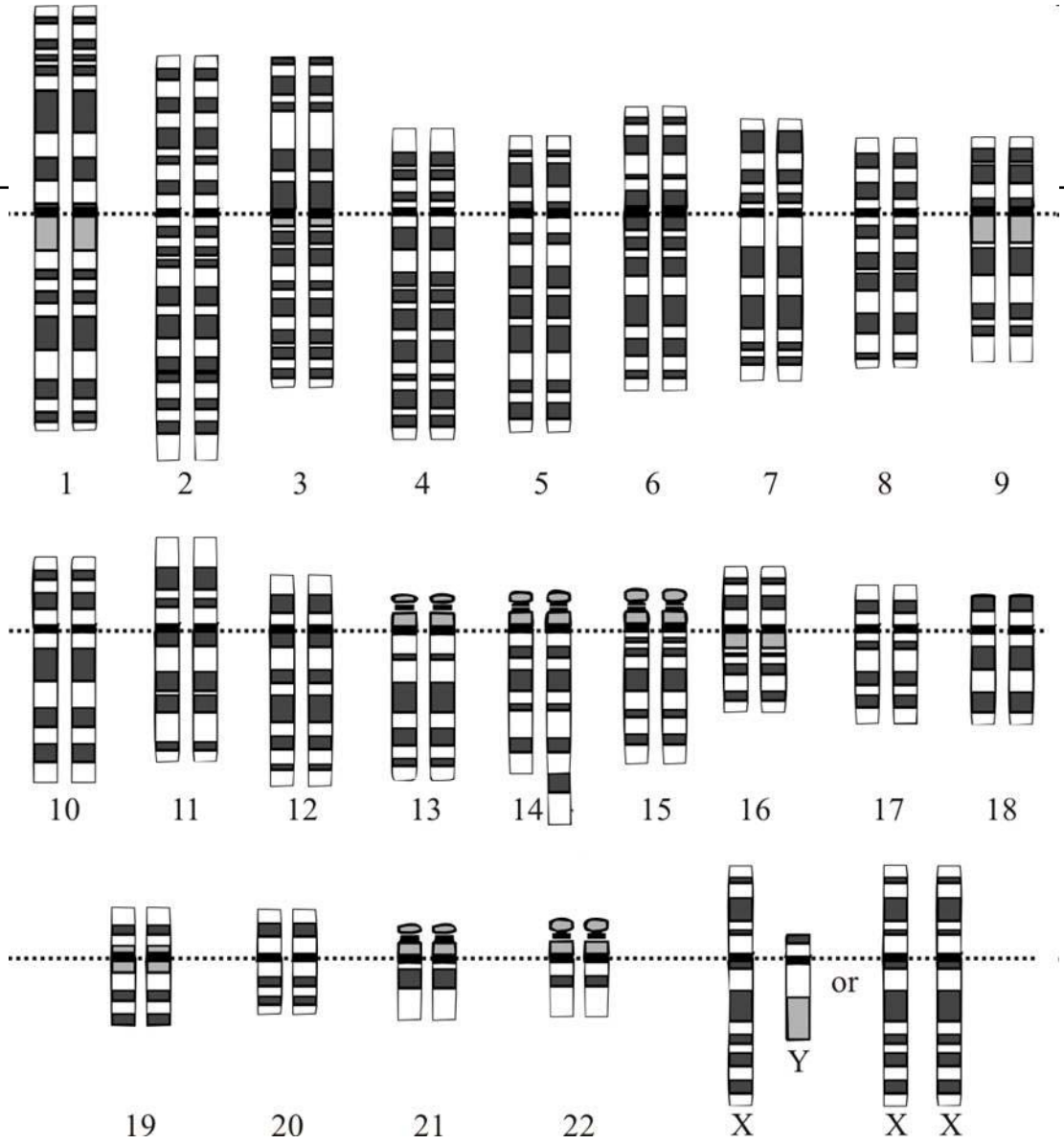



**Figure 9-12 part 4**
***Genetics: A Conceptual Approach, Third Edition***


1
2
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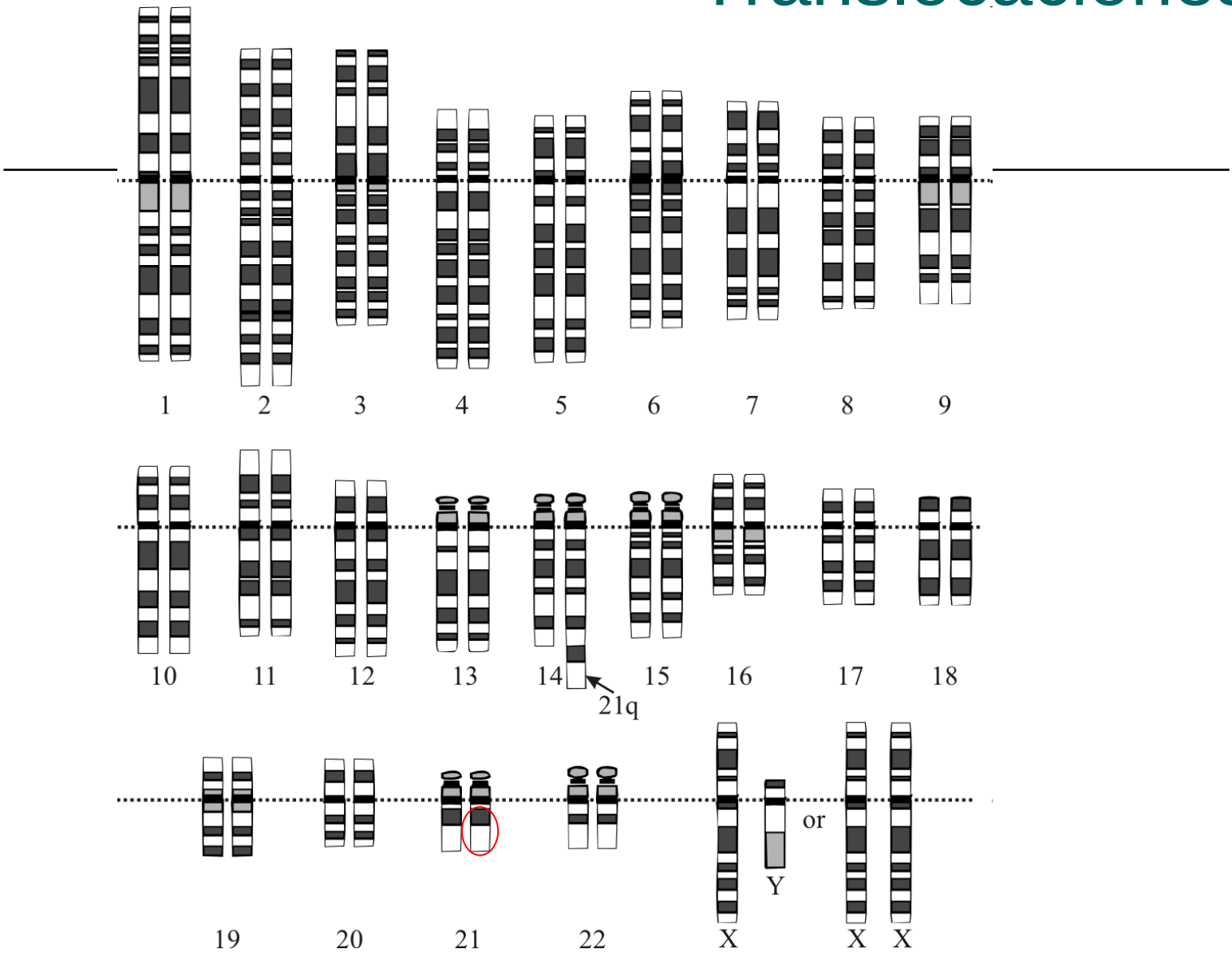
18
19
20
21
22
X
Y
or
X
X

# Translocaciones



# Translocación Robertsoniana

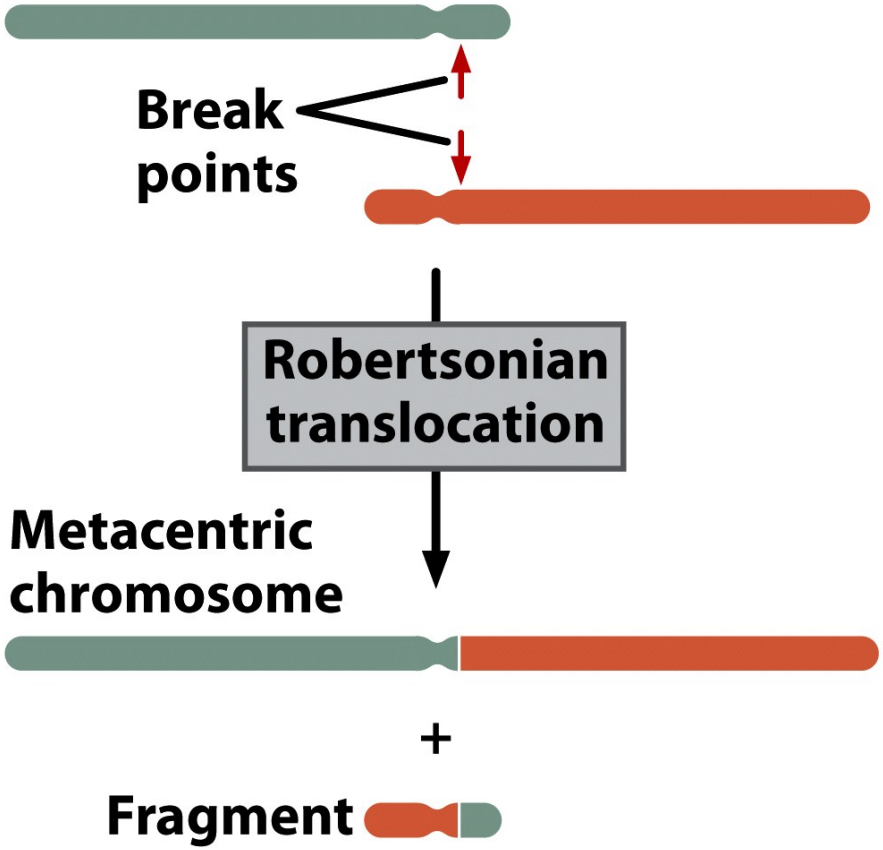



Figure 9-15
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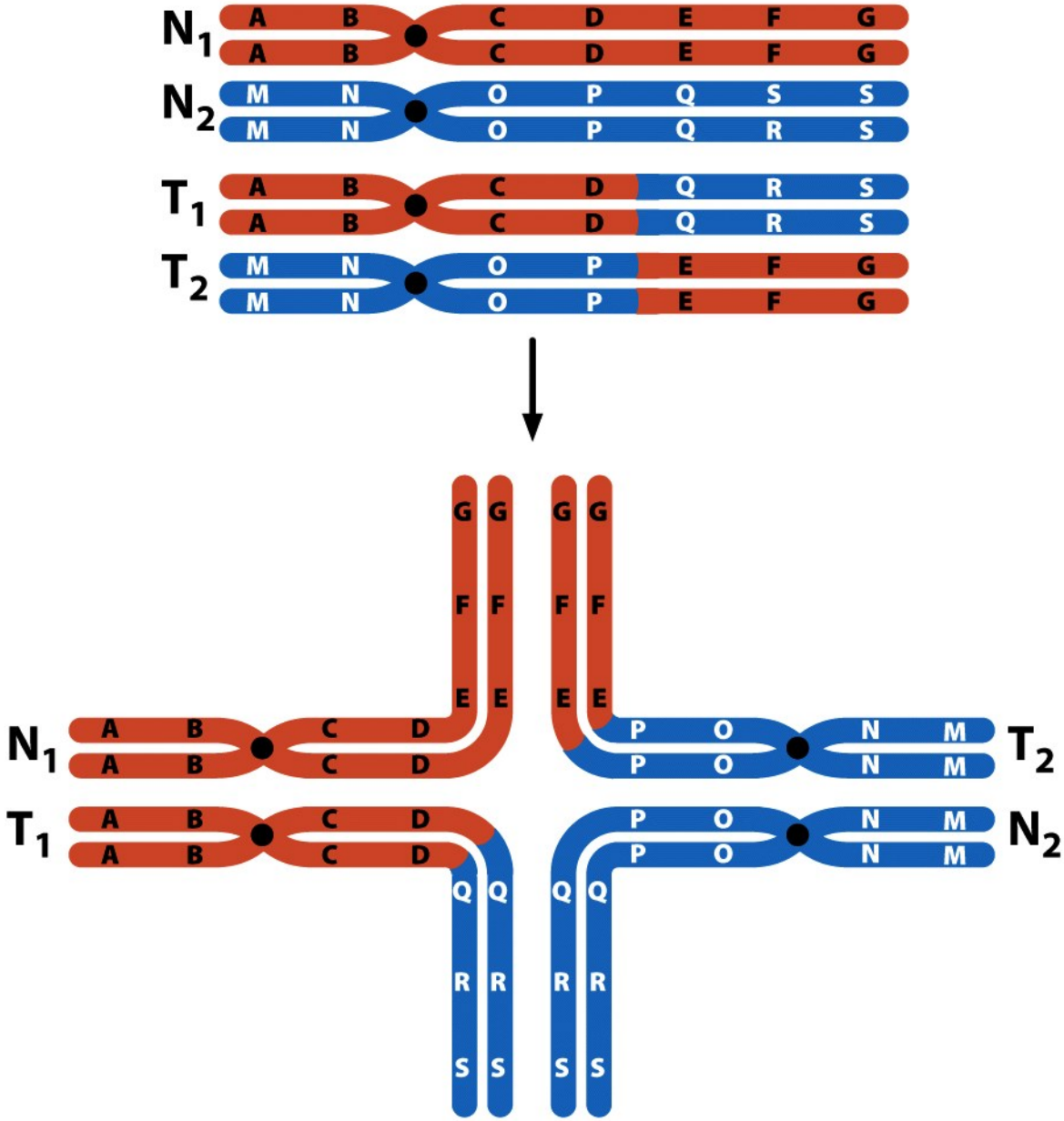


**Figure 9-16 part 1**
***Genetics: A Conceptual Approach, Third Edition***

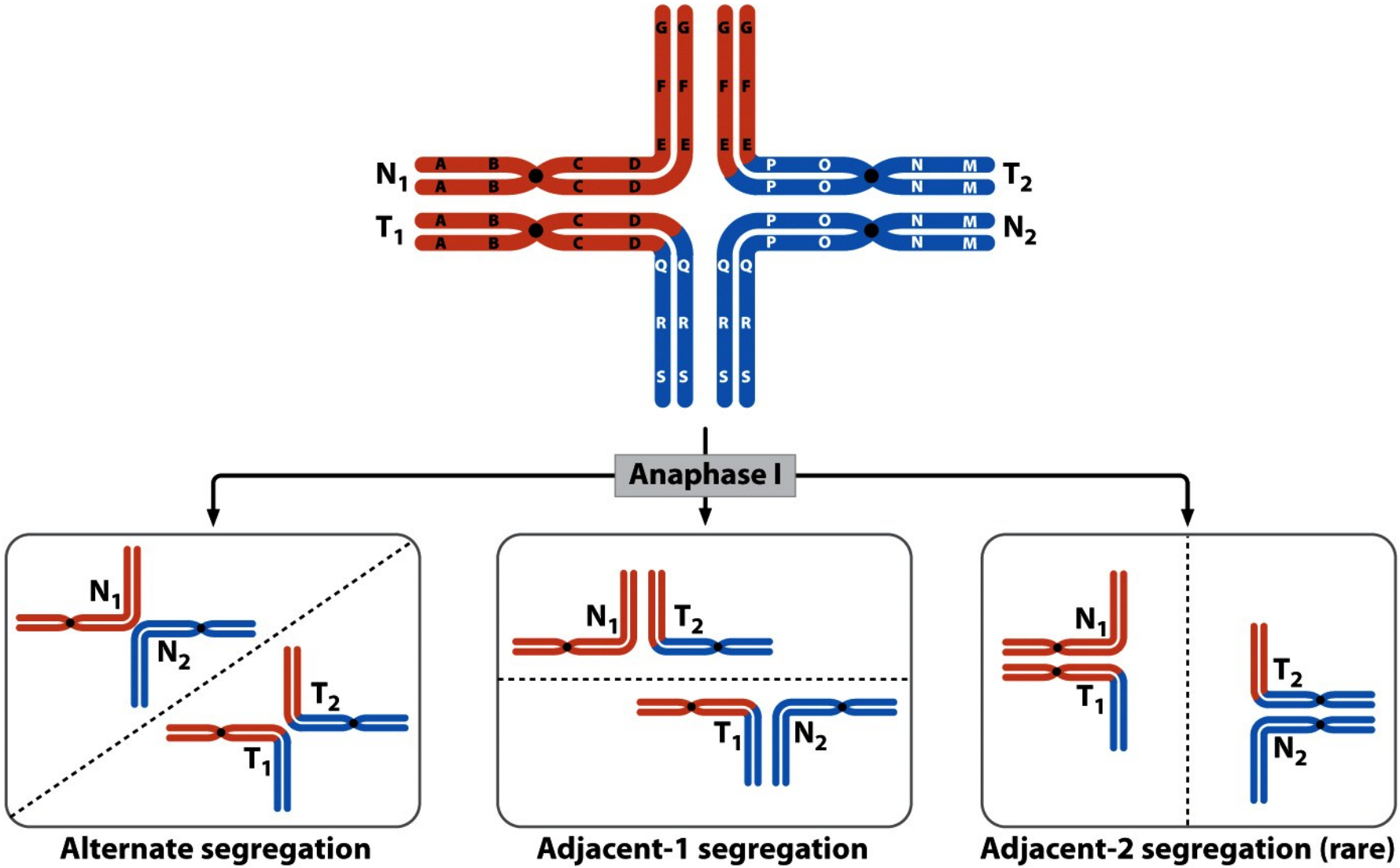


**Figure 9-16 part 2**
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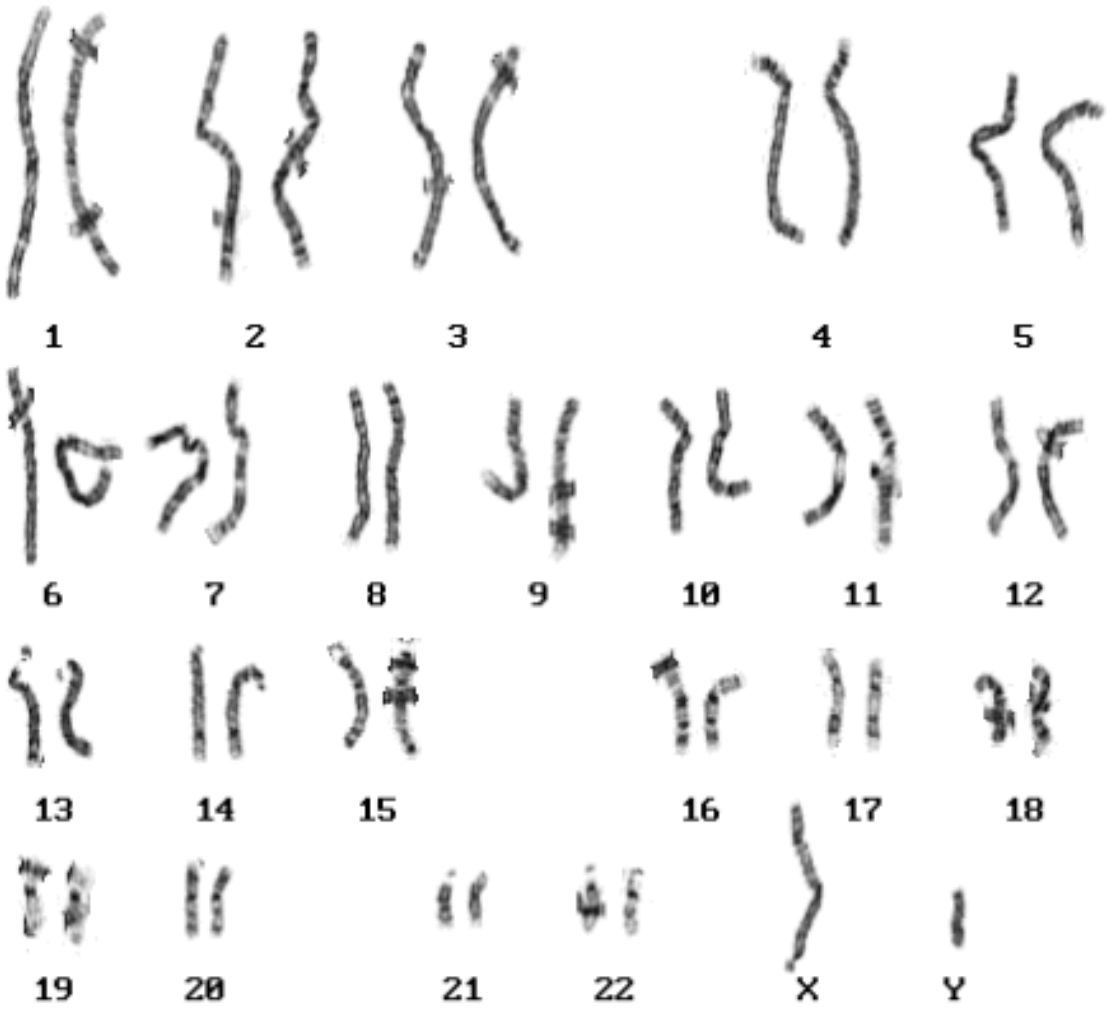
# X Frágil




# Mutaciones Estructurales

- Deleciones
- Duplicaciones
- Inversiones
- Translocaciones

# Mutaciones Numéricas

- Aneuploidías
- Poliploidías

# Aneuploidías

- Nulisomía (2n-2)
- Monosomía (2n-1)
- Trisomía (2n+1)
  - Autosomas
  - Cromosomas Sexuales
- Tetrasomía (2n+2)

# Monosomía

**Síndrome de Turner**

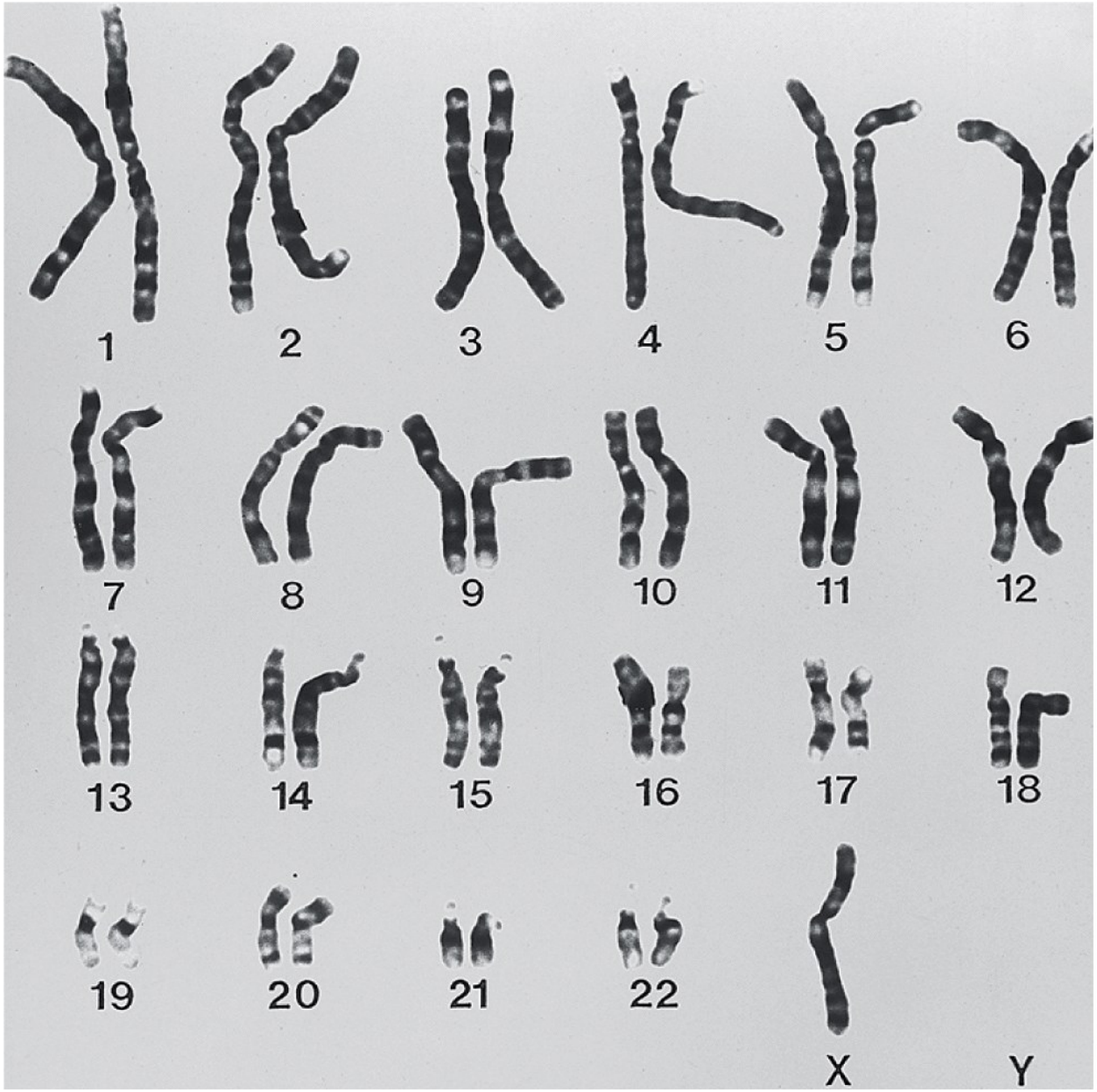



**Figure 4-8**
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Trisomía

Figure 4-9
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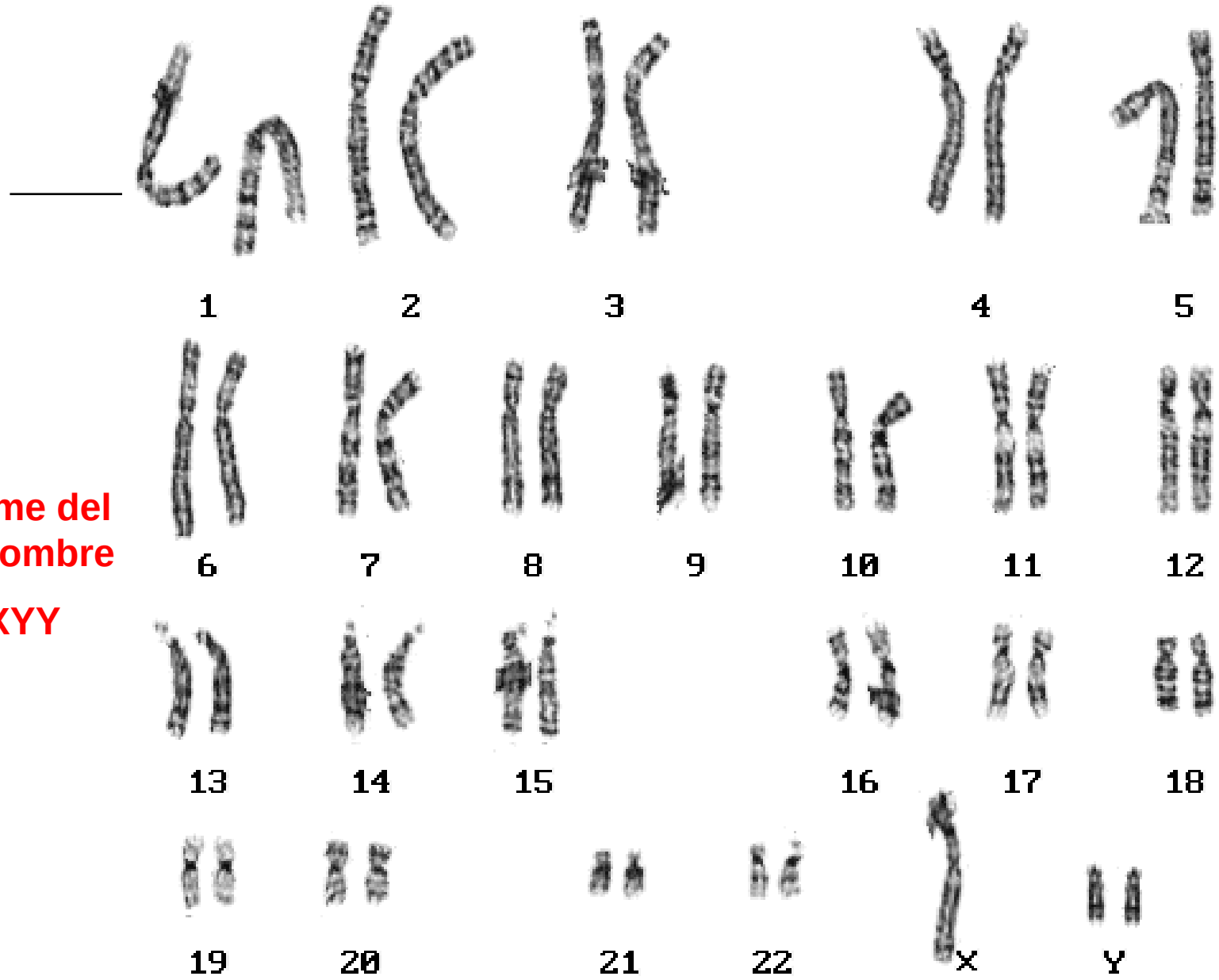
**Síndrome de Klinefelter**

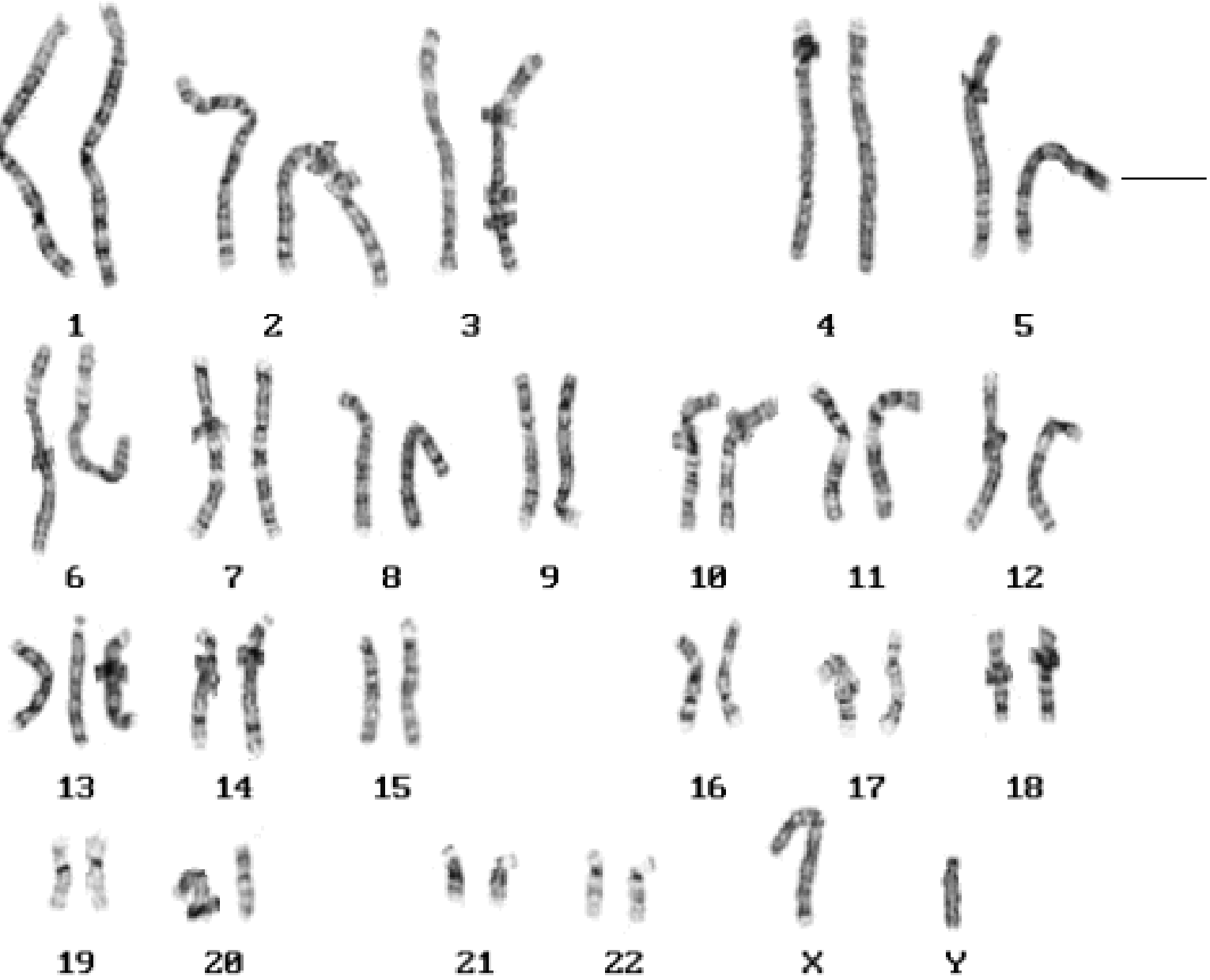
**Síndrome del Superhombre**

**47, XYY**



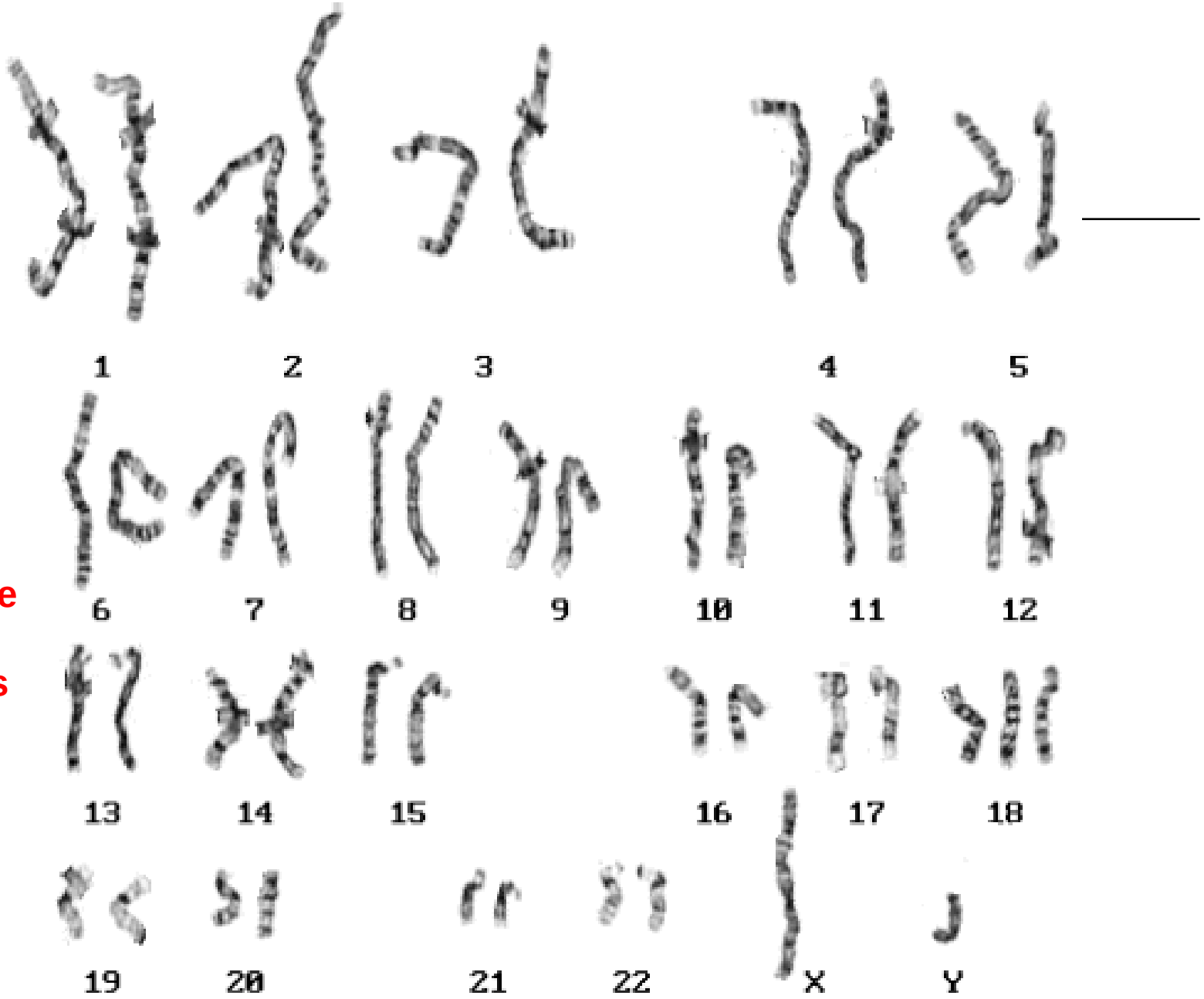
**Síndrome de Patau**

**47, +13**



**Síndrome de Edwards**

**47, +18**



# Síndrome de Down

**47, +21**

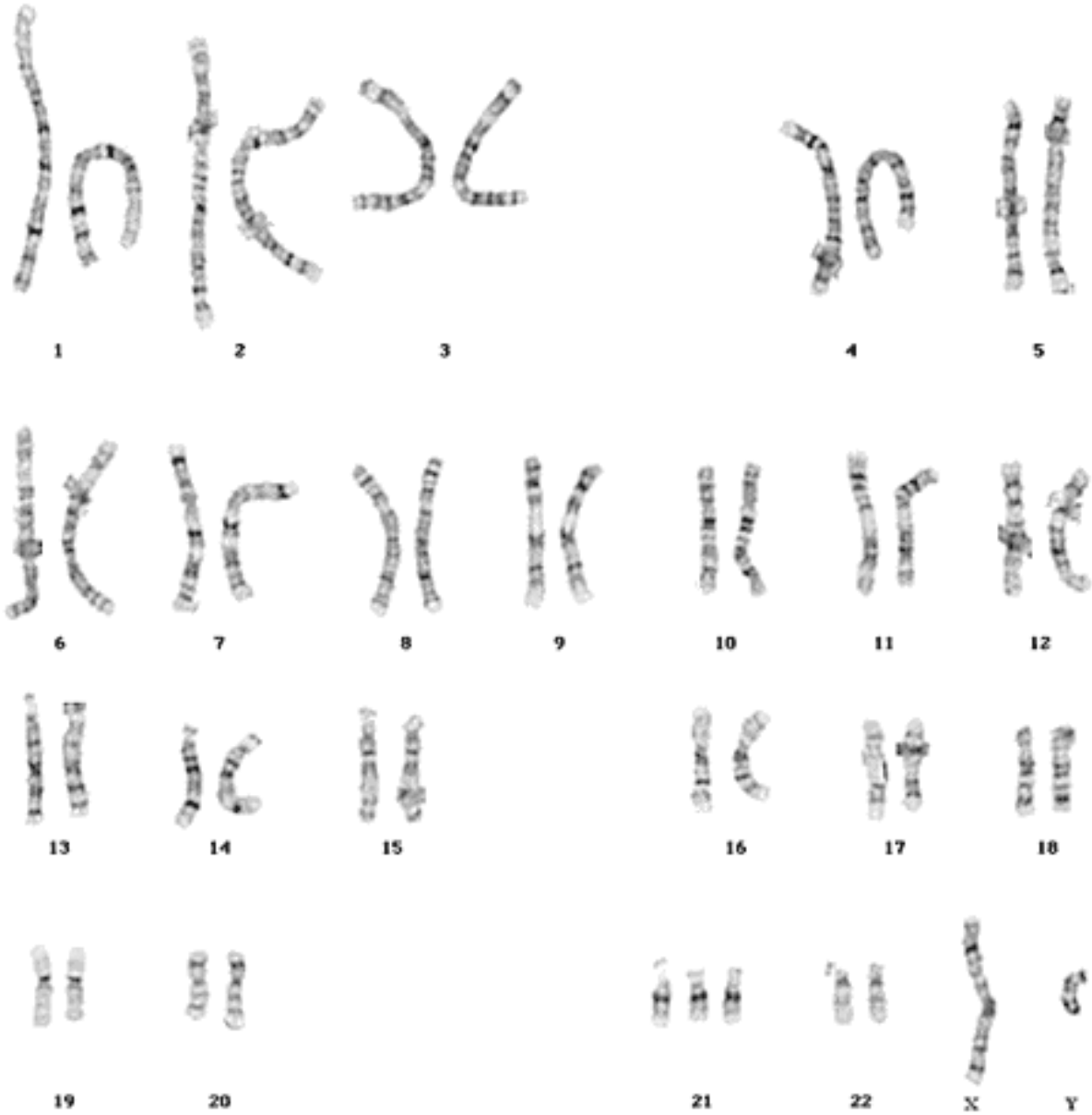



**Figure 9-22**
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# No disyunción en Meiosis I

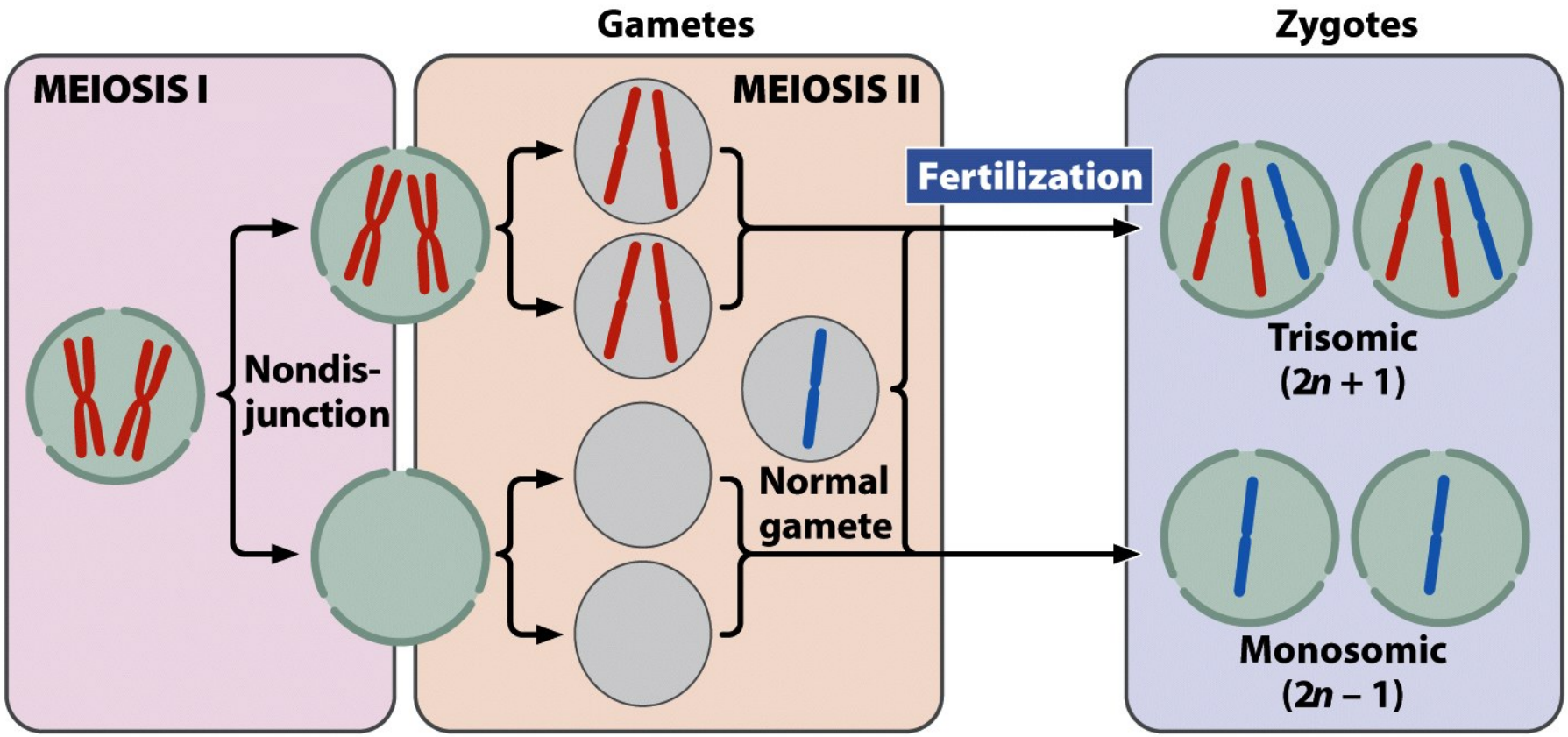


**Figure 9-19a**
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# No disyunción en Meiosis II

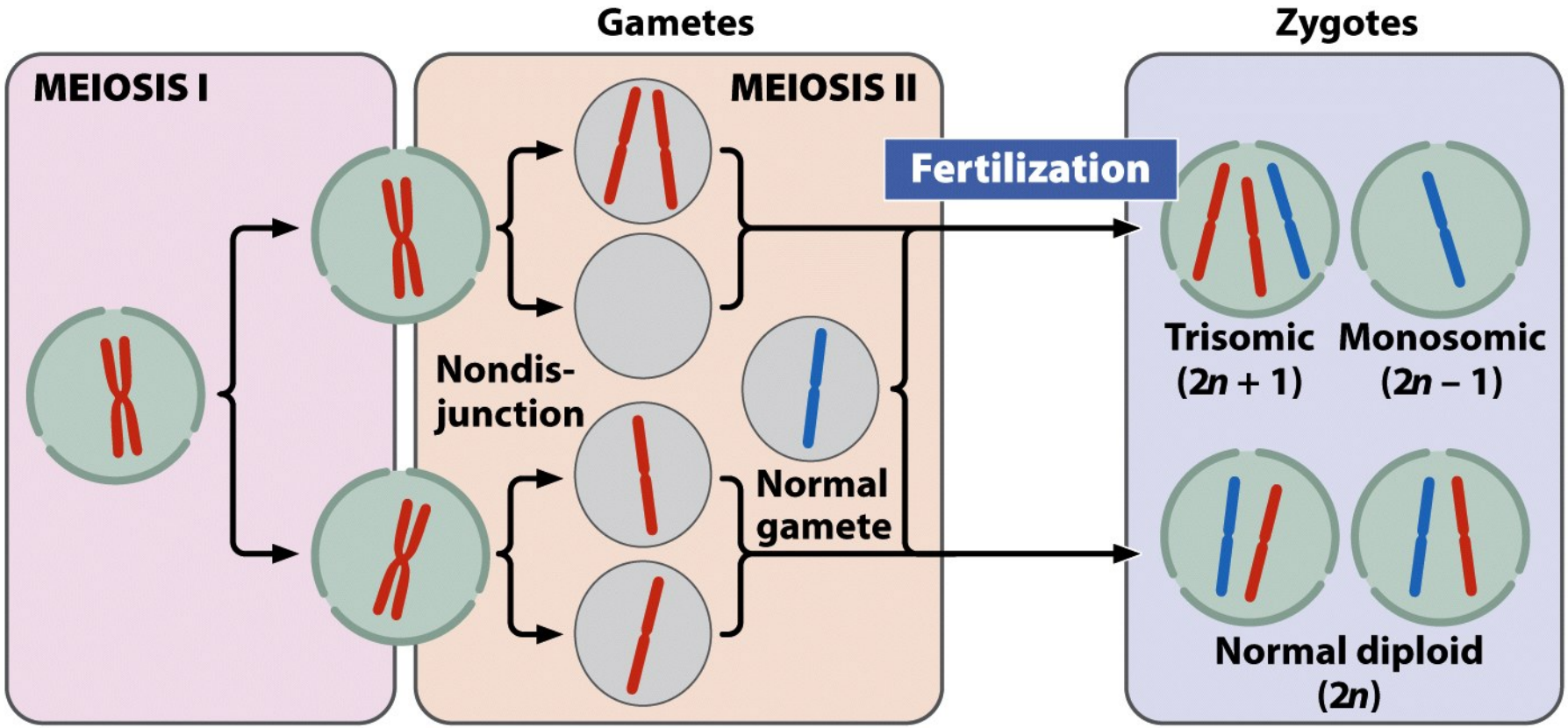


Figure 9-19b
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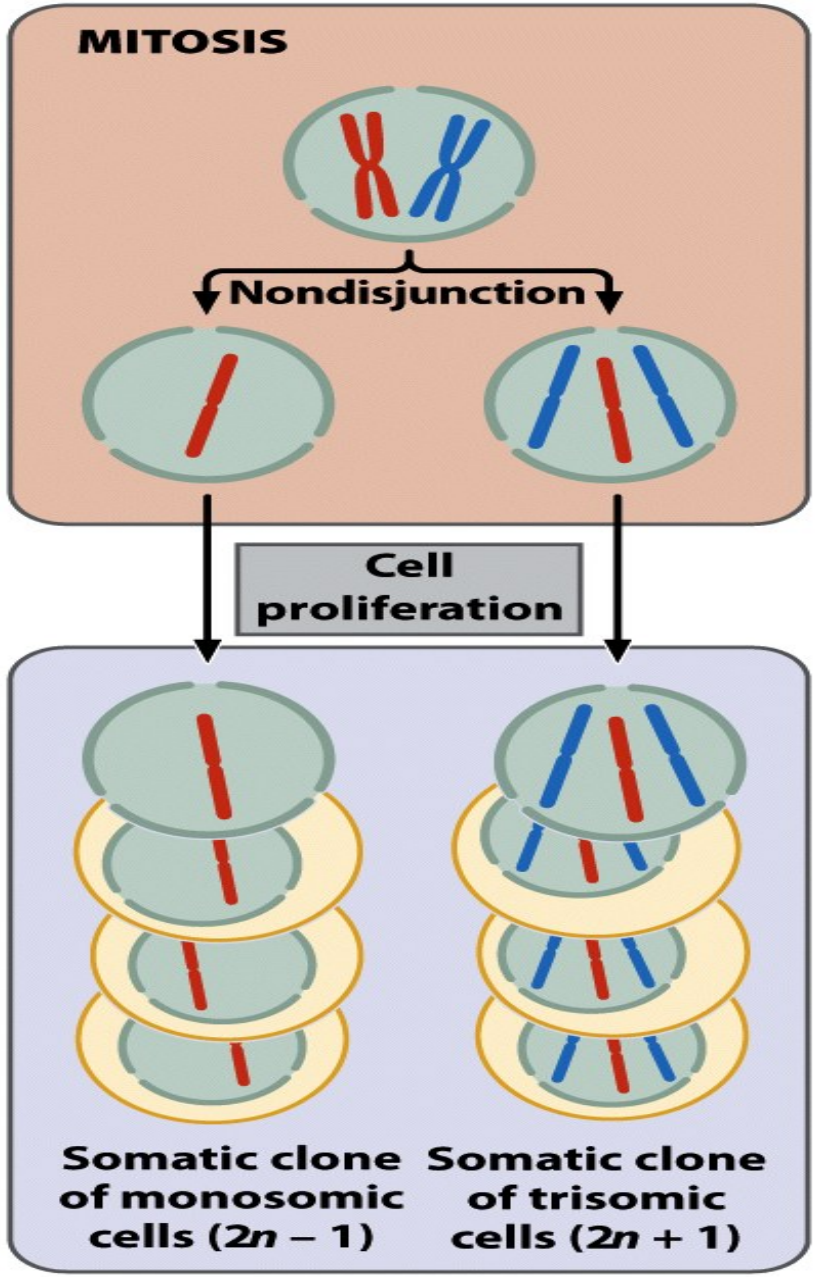


Figure 9-19c
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# No disyunción en Mitosis

# Poliploidías

- Alto porcentaje de plantas
- Algunos animales (invertebrados, peces, anfibios y reptiles)

**Autopoliploidía** **Alopoliploidía**

# Autopoliploidía

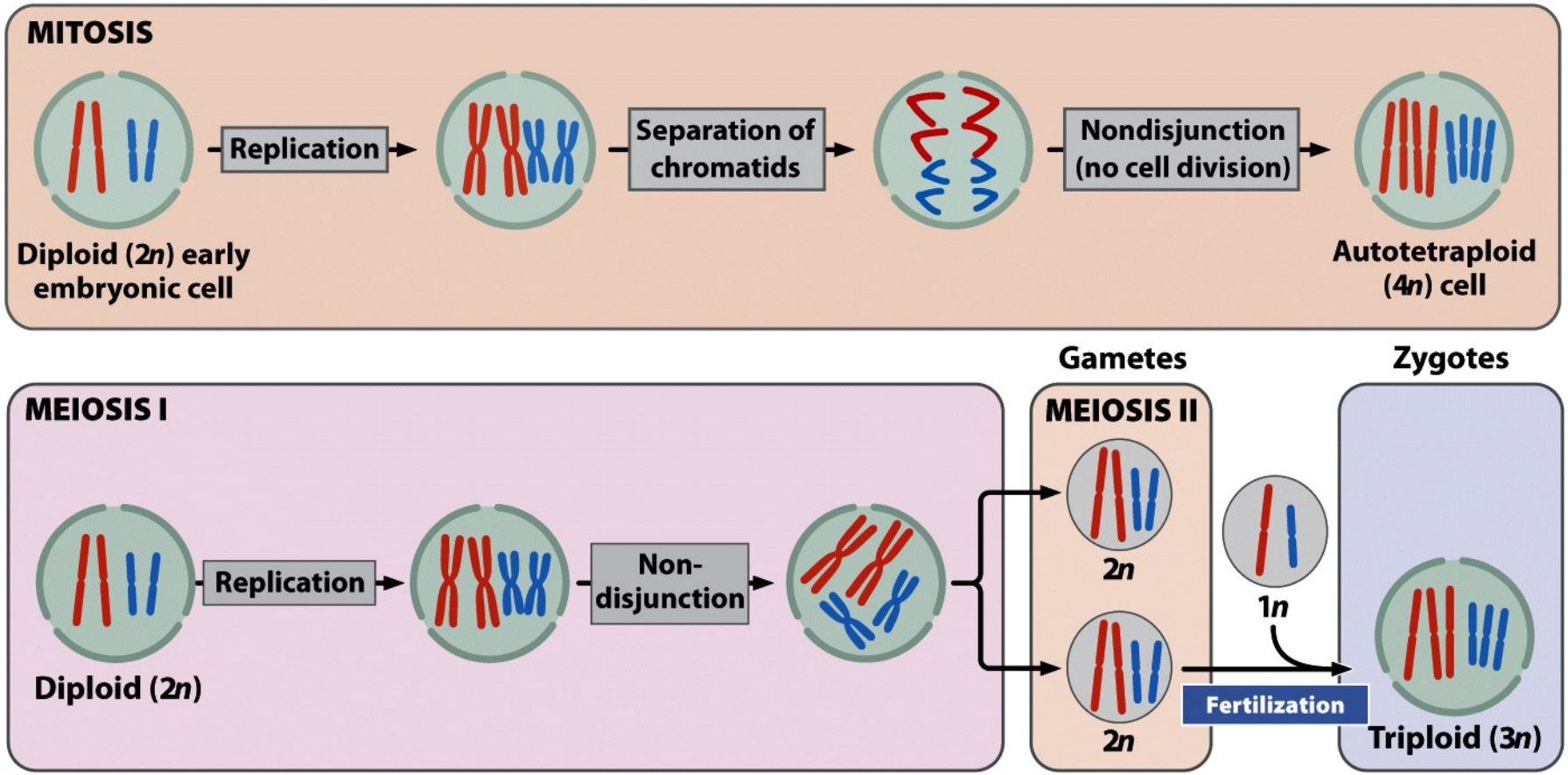


Figure 9-26b
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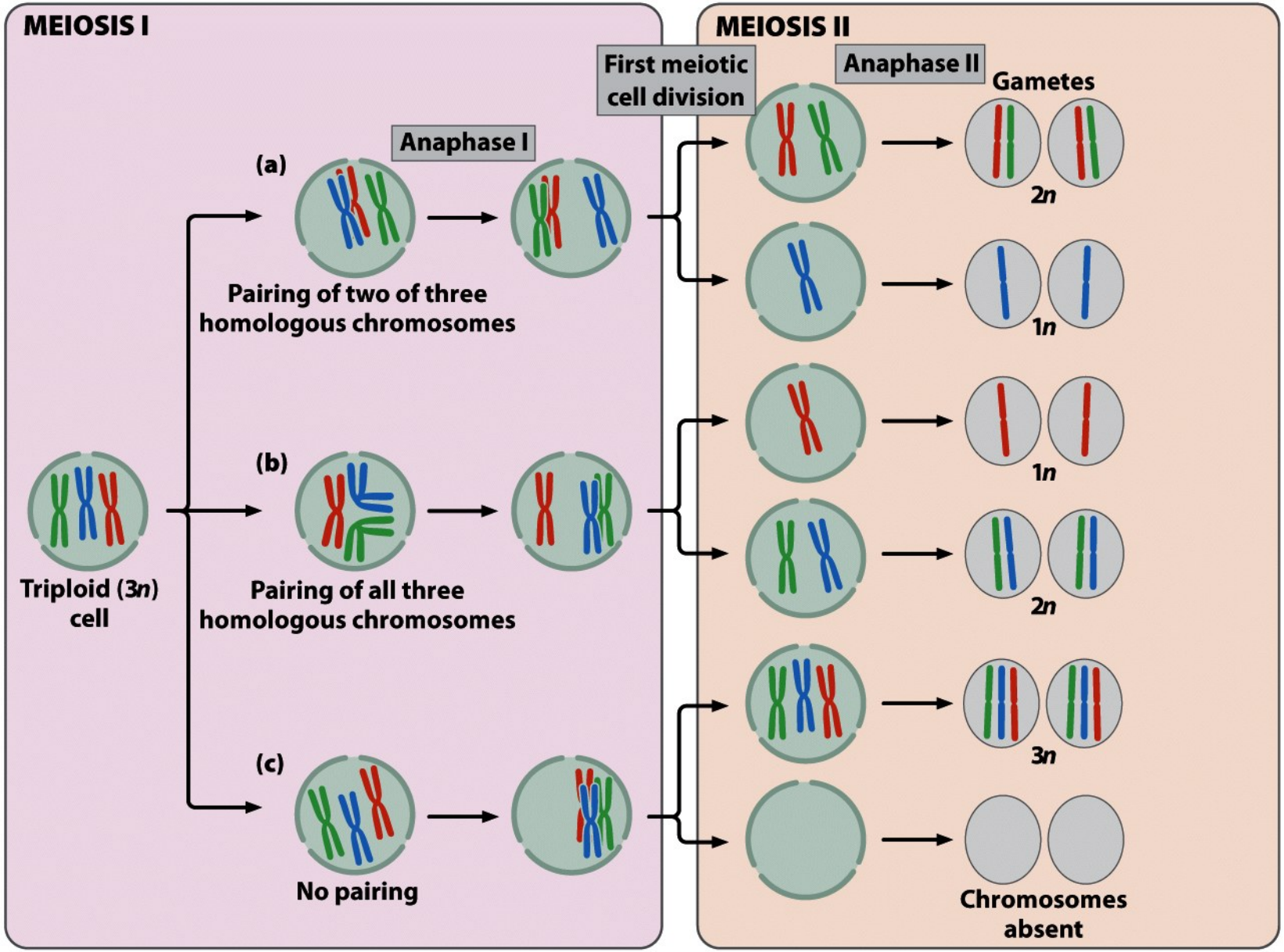


**Figure 9-27**
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# Uso de los Triploides

# Uso de los Triploides

# Alopoliploidía

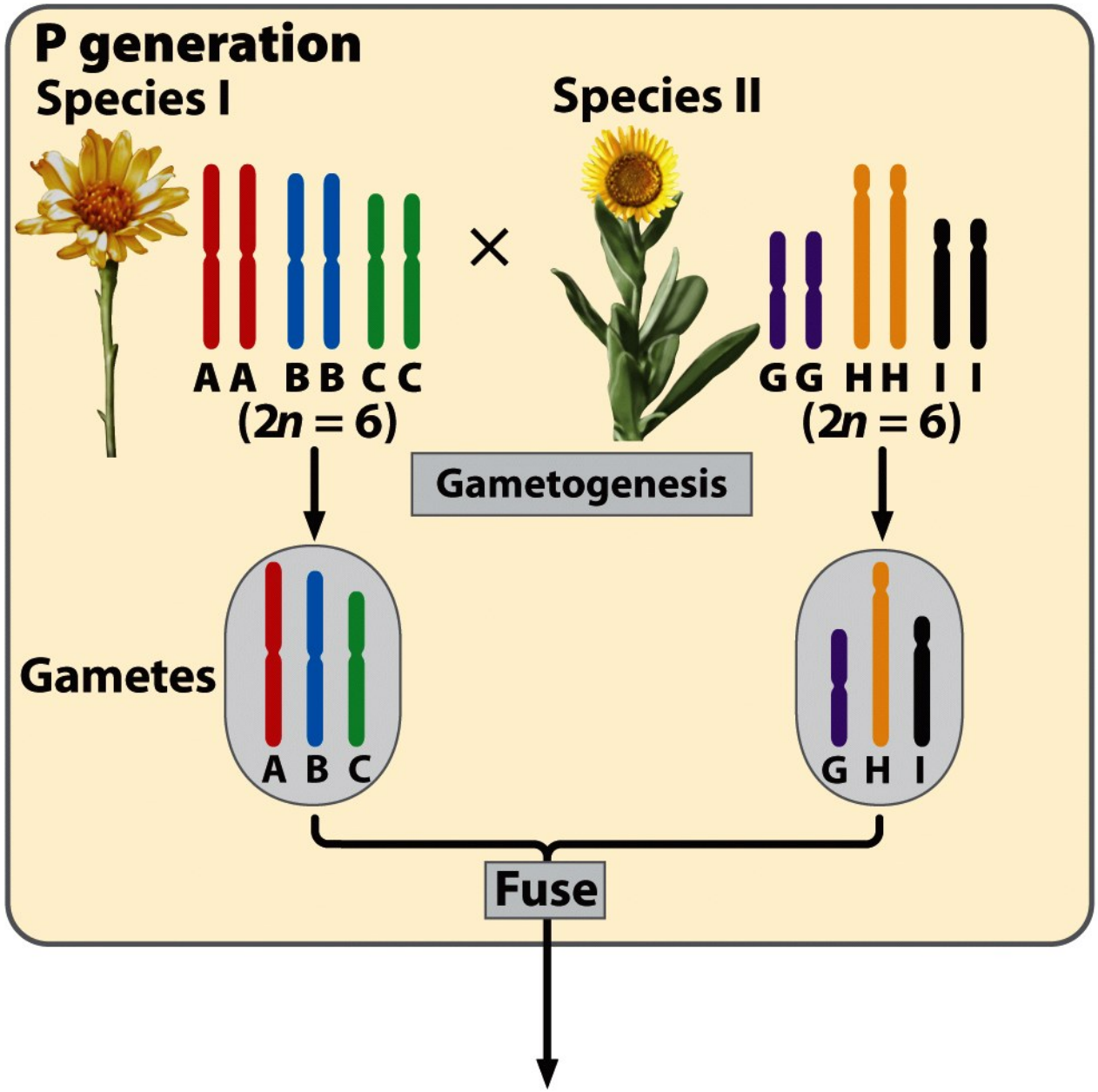


Figure 9-28 part 1
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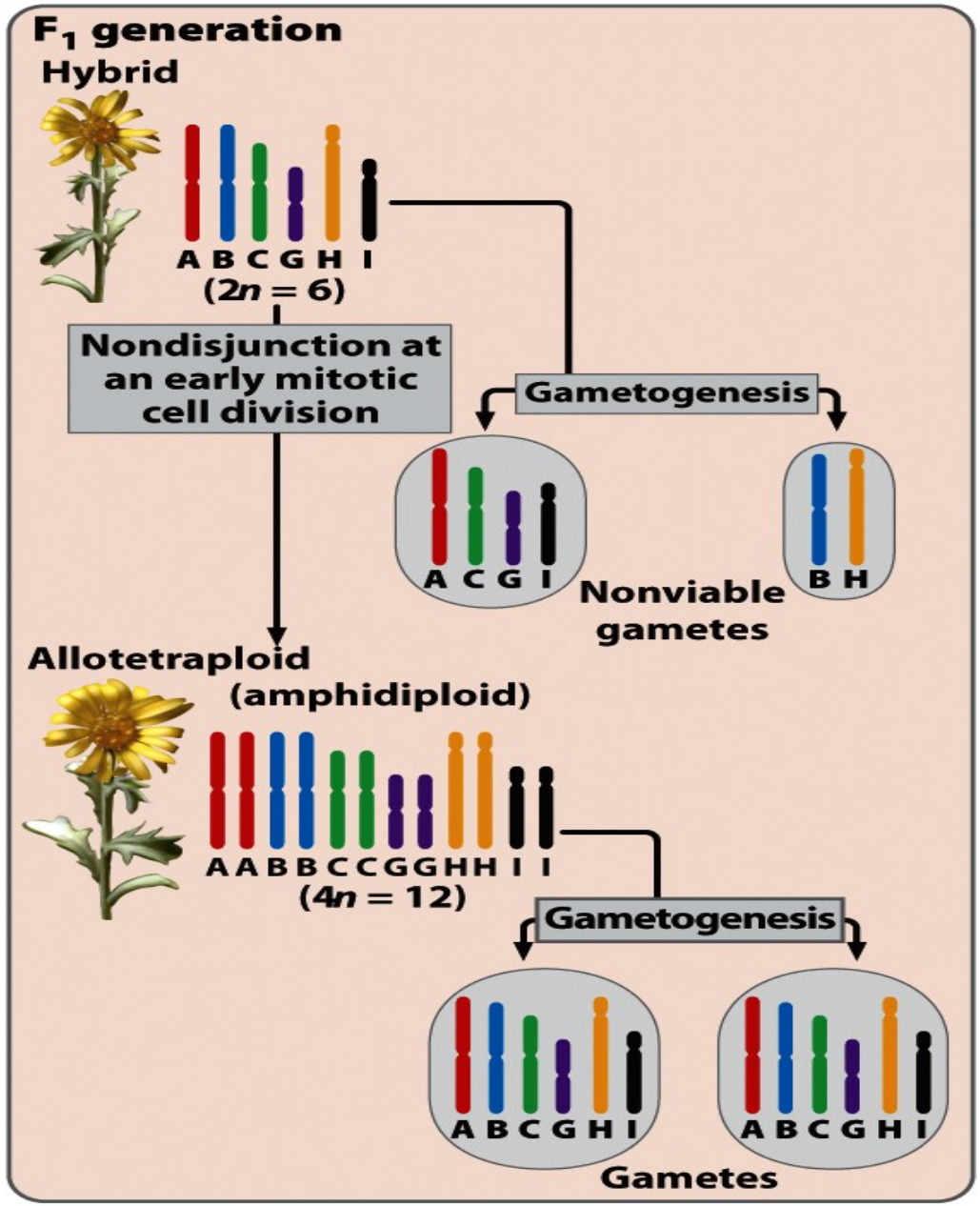
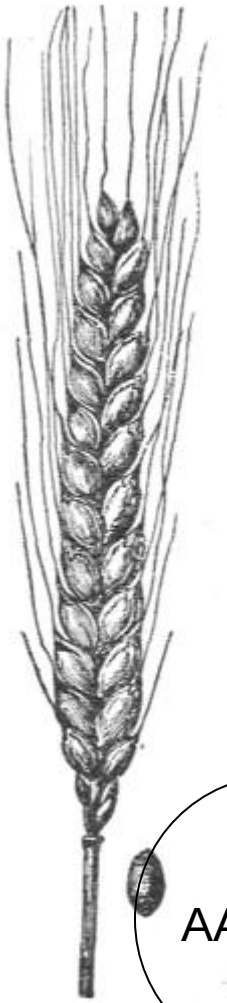



**Figure 9-28 part 2**
***Genetics: A Conceptual Approach, Third Edition***



*Triticum monococcum*
2n=14

AA

BB

*Aegilops sp.* 2n=14

A x B

AB

*Híbrido interespecífico*
$2n=n_1+n_2=14$

*Duplicación Natural*

Trigo Duro 2n=28

AABB

DD

*Aegilops squarrosa*
2n=14

AB x D

ABD

*Híbrido interespecífico*
$2n=n_1+n_2+n_3=21$

*Duplicación Natural*

AABBDD

Trigos Harineros
(hexaploides)

# Alopoliploidía en *Brassica*

**Mostaza negra**

BB
n=8

**Berza**

*Brassica nigra*

**Mostaza parda**

BBCC
n=9+8

AABB
n=10+8

*Brassica carinata*

*Brassica juncea*

**Coliflor, brocoli, col verde**

CC
n=9

AACC
n=10+9

AA
n=10

*Brassica olarecea*

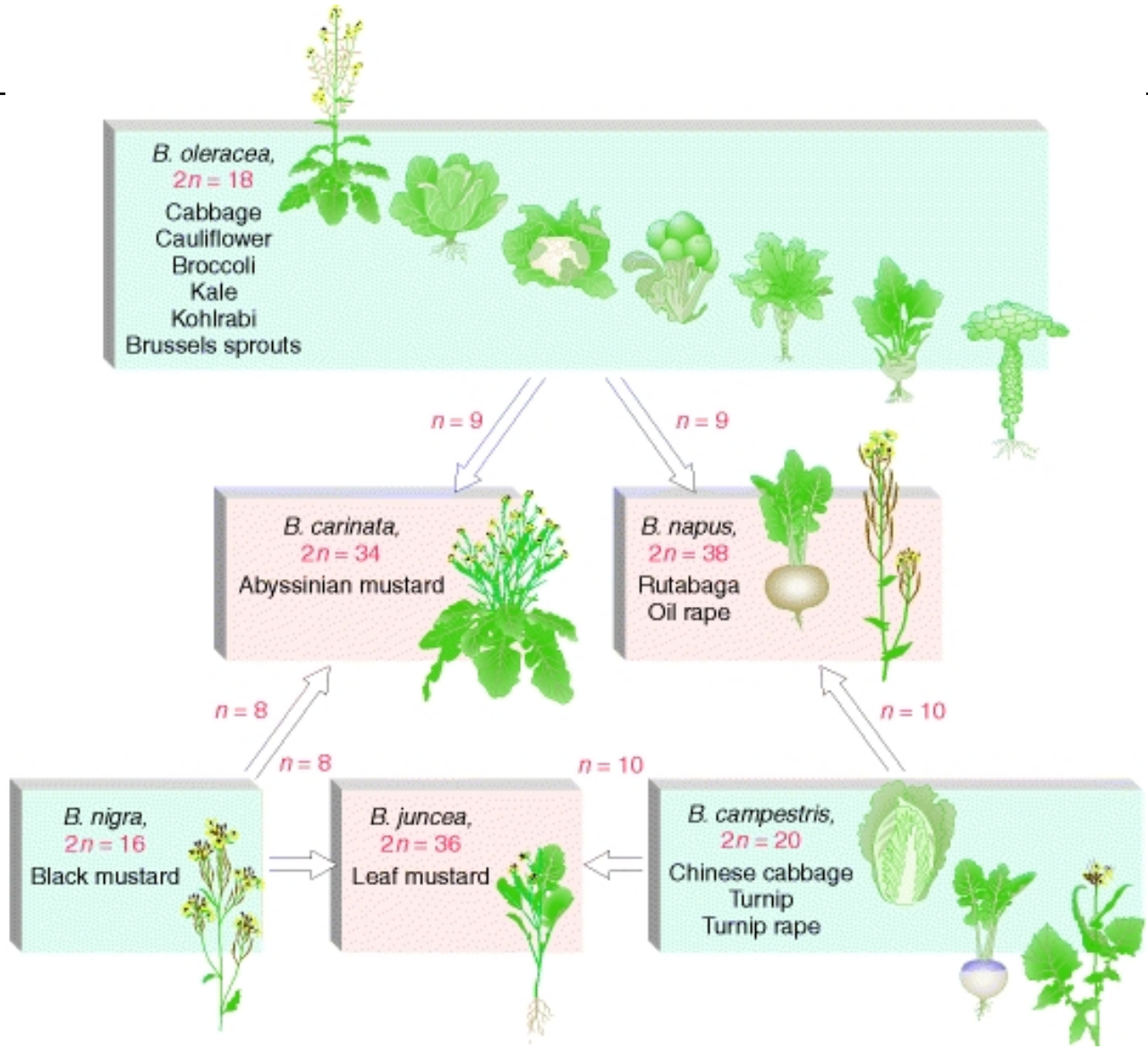
*Brassica napus*

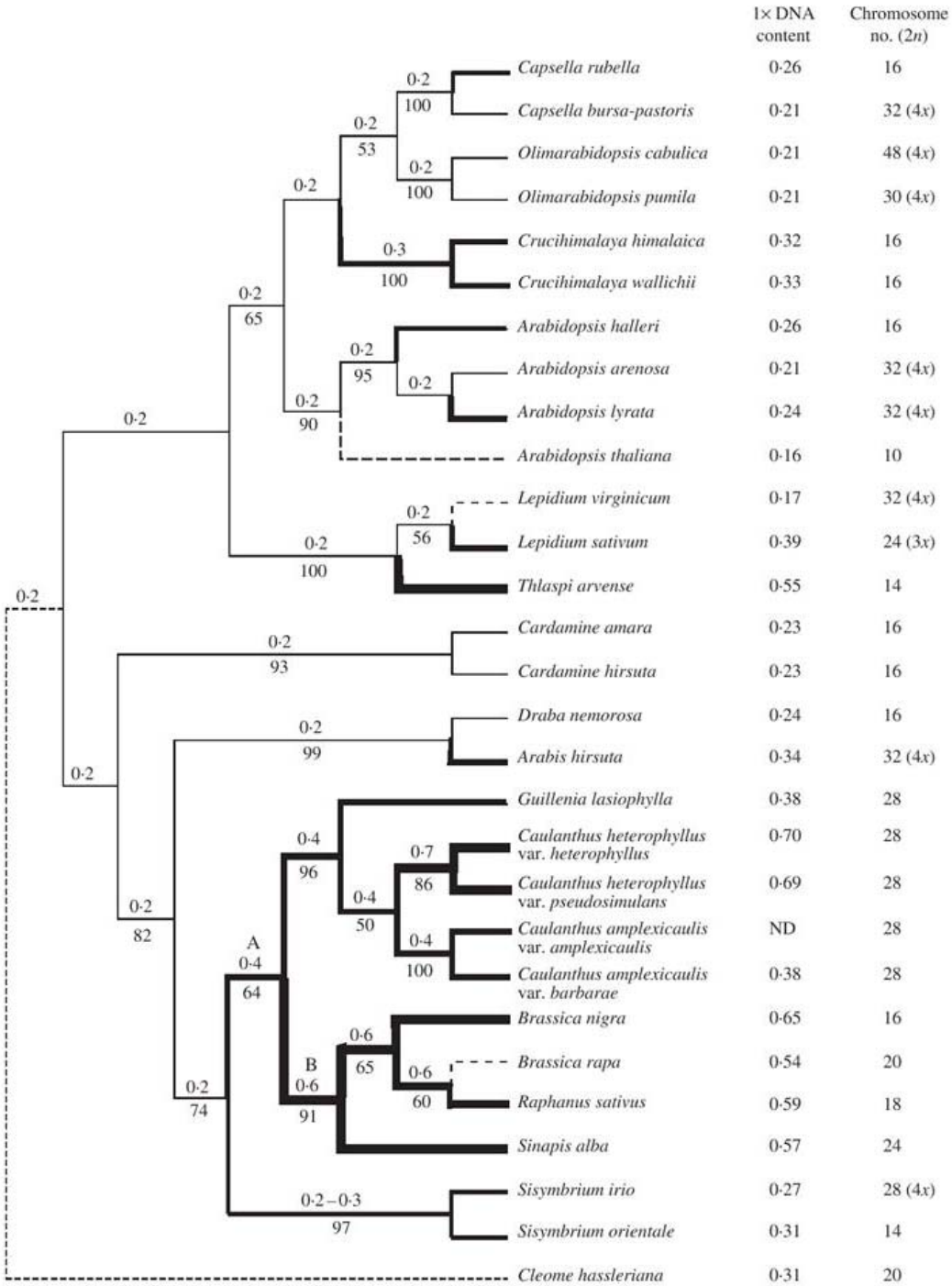
*Brassica rapa*

**Col china, nabo**

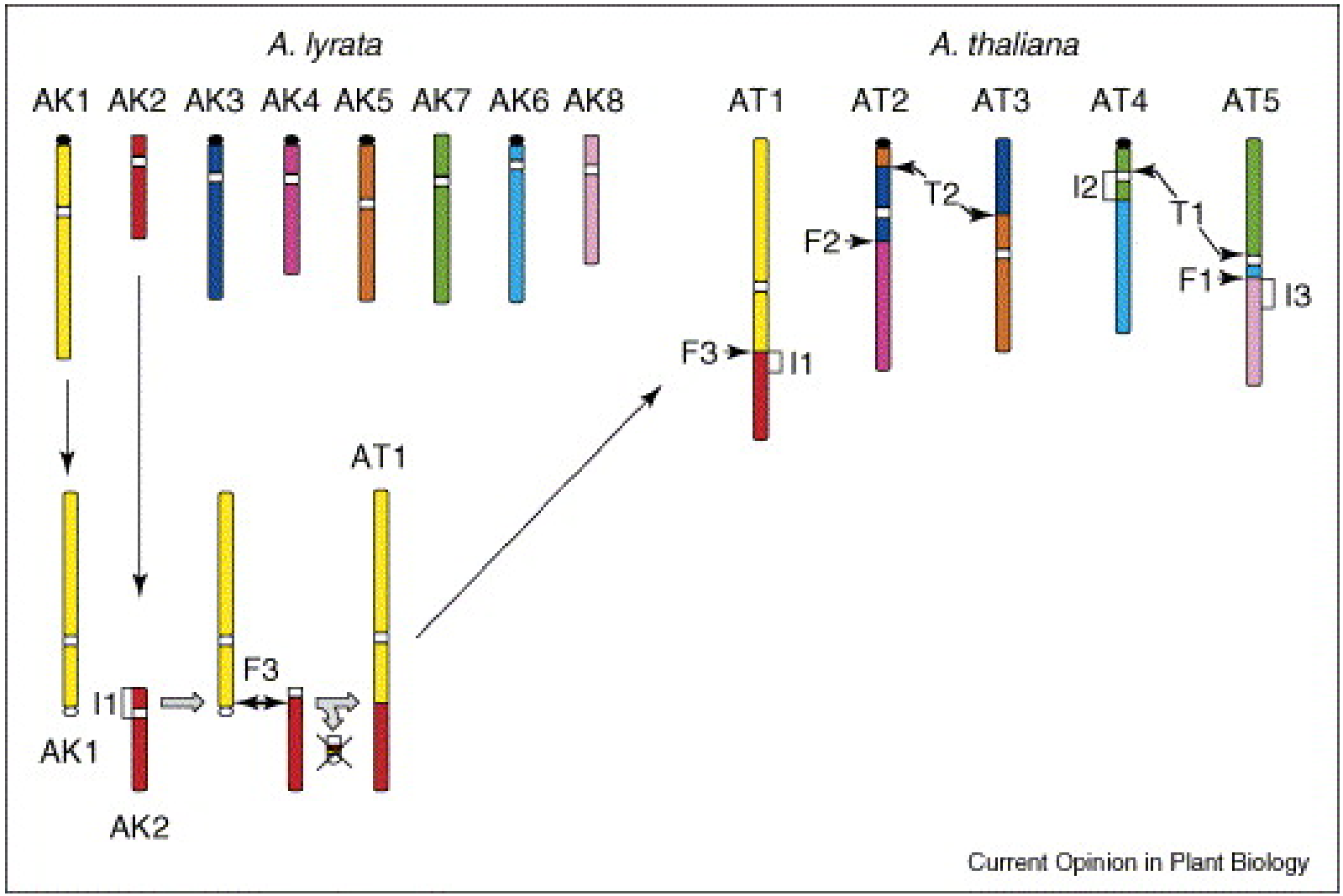
**Colza**

# Evolución Cromosómica en *Brassica*




1× DNA content
Chromosome no. (2n)
Capsella rubella 0·26 16
Capsella bursa-pastoris 0·21 32 (4x)
Olimarabidopsis cabulica 0·21 48 (4x)
Olimarabidopsis pumila 0·21 30 (4x)
Crucihimalaya himalaica 0·32 16
Crucihimalaya wallichii 0·33 16
Arabidopsis halleri 0·26 16
Arabidopsis arenosa 0·21 32 (4x)
Arabidopsis lyrata 0·24 32 (4x)
Arabidopsis thaliana 0·16 10
Lepidium virginicum 0·17 32 (4x)
Lepidium sativum 0·39 24 (3x)
Thlaspi arvense 0·55 14
Cardamine amara 0·23 16
Cardamine hirsuta 0·23 16
Draba nemorosa 0·24 16
Arabis hirsuta 0·34 32 (4x)
Guillenia lasiophylla 0·38 28
Caulanthus heterophyllus var. heterophyllus 0·70 28
Caulanthus heterophyllus var. pseudosimulans 0·69 28
Caulanthus amplexicaulis var. amplexicaulis ND 28
Caulanthus amplexicaulis var. barbarae 0·38 28
Brassica nigra 0·65 16
Brassica rapa 0·54 20
Raphanus sativus 0·59 18
Sinapis alba 0·57 24
Sisymbrium irio 0·27 28 (4x)
Sisymbrium orientale 0·31 14
Cleome hassleriana 0·31 20
A
B

# Evolución Cromosómica
# *Arabidopsis*



# Evolución Cromosómica *Cervidae*

**2n=70** 68,XY A + X acrocéntricos, Y submetacéntrico pequeño

**Hydropotinae** *Hydropotes inermis* ciervo de agua (Viejo mundo, China)

**Odocoloileinae** *Mazama gouazoubira* Ciervo marrón (Nuevo mundo)

**Muntiacinae**

**2n=46h/47m, 48**
*Elaphodus cephalophus*

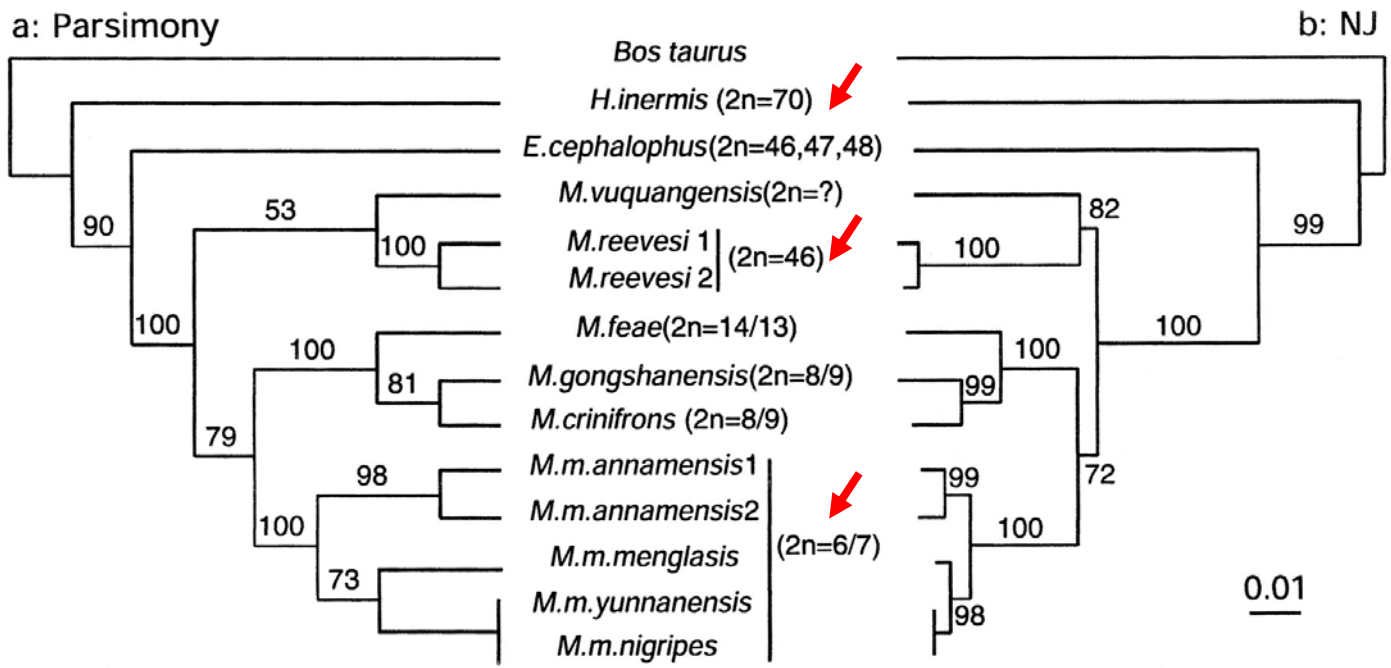
**2n=46**
*Muntiacus reevesi* muntjak de la China

**2n=13h/14m**
*Muntiacus feae*

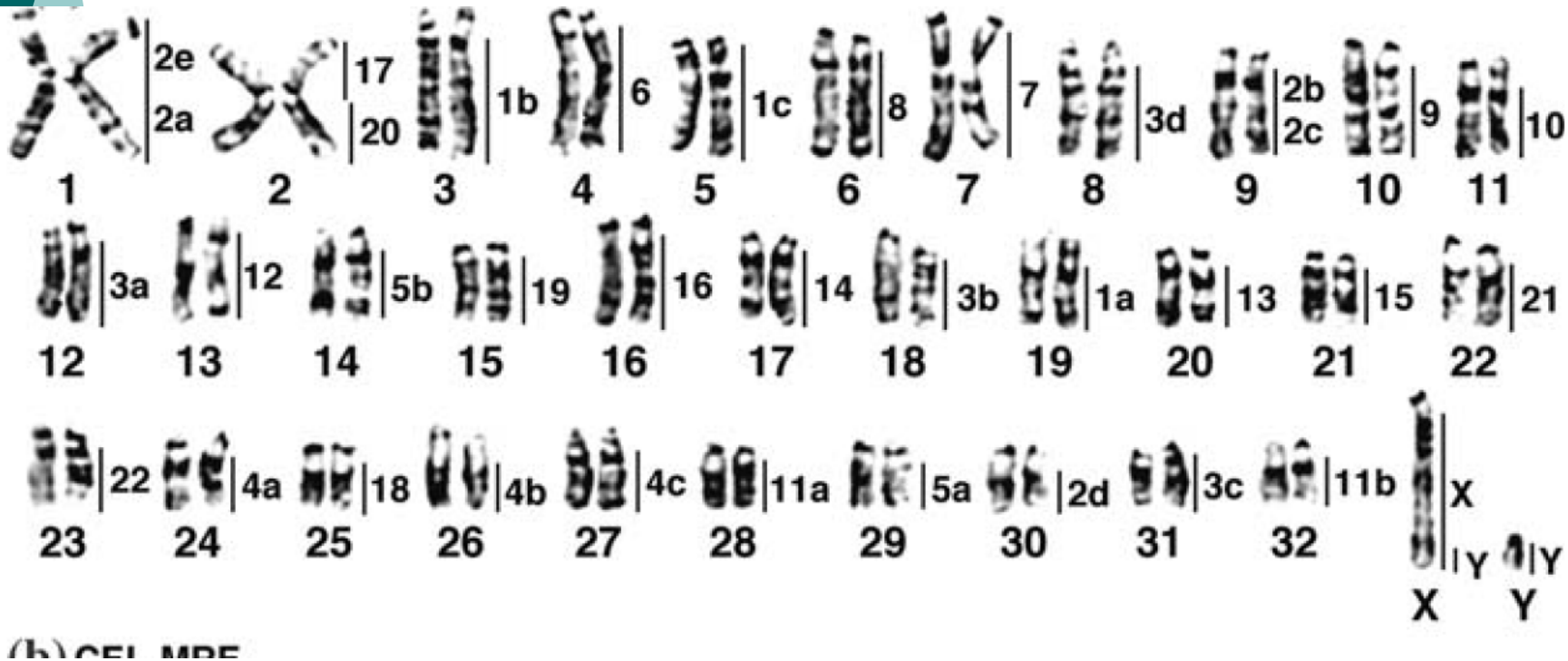
**2n=8h/9m**
*Muntiacus muntjak muntjak*
*Muntiacus crinifrons*
*Muntiacus gongshanensis* China

**2n=6h/7m**
*Muntiacus muntjak vaginalis* muntjak de la India

# Evolución Cromosómica *Cervidae*



# *Cervus nippon* (2n=66)



Cariotipo de *Muntiacus reevesi*

*China*

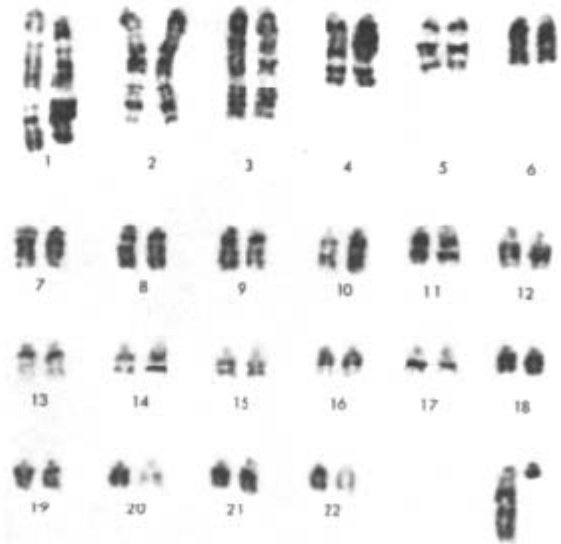
2n=46

44 + XY

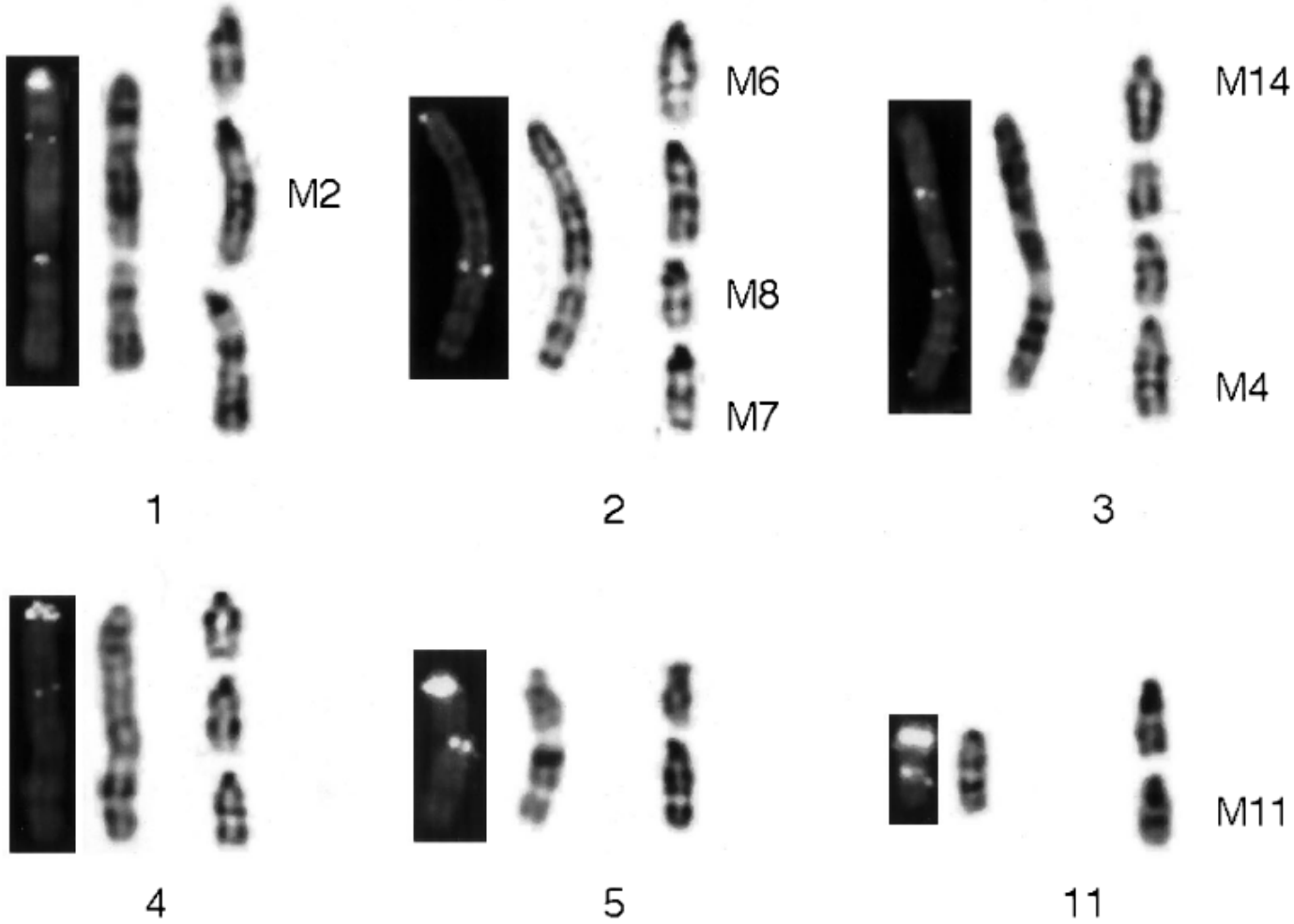
Acrocéntricos

Y pequeño

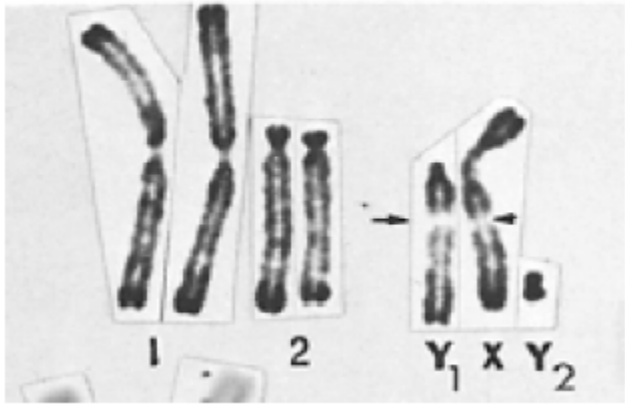
submetacéntrico

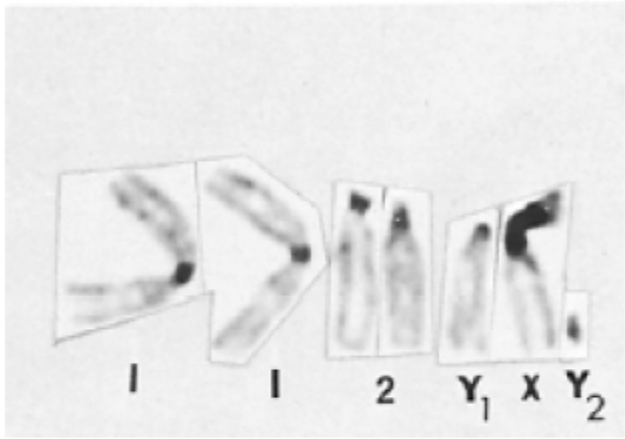


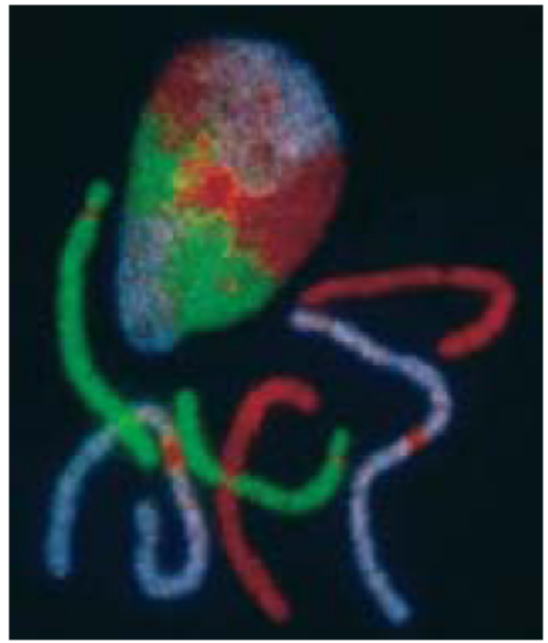
**Los cromosomas**
***Muntiacus reevesi* se originaron por 12 fusiones en tandem**
**implicando 18 cromosomas de**
**un cariotipo cervido ancestral**
**El resto de los cromosomas permanecieron intactos**



# Cariotipo de *Muntiacus muntjak vaginalis* India



2n =7 macho 4 + $XY_1Y_2$

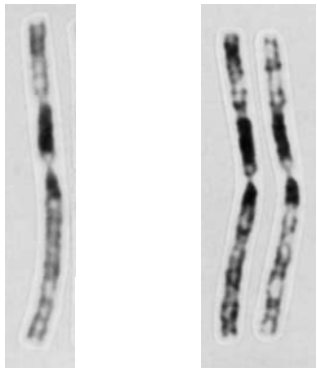
2n = 6 hembra 4 + XX

# Los números cromosómicos extremos en hormigas

***Myrmecia pilosula***

**n = 1**

**n=1**



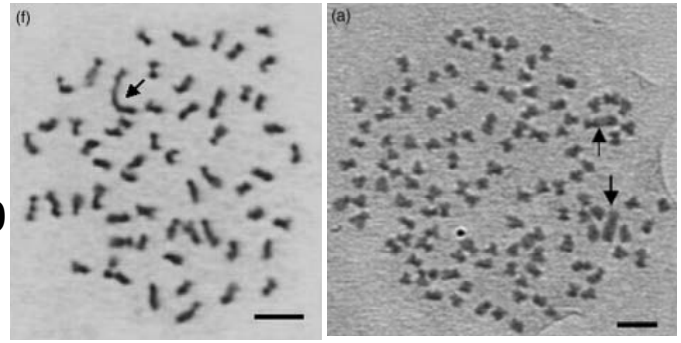
**2n=2**

Crosland MWJ & Crozier RH 1986: Science 231: 1278

***Dinoponera lucida***

**n = 60**

**n=60**




**2n=120**

Mariano et al. 2008: Insect Conservation and Diversity 1: 161-168.

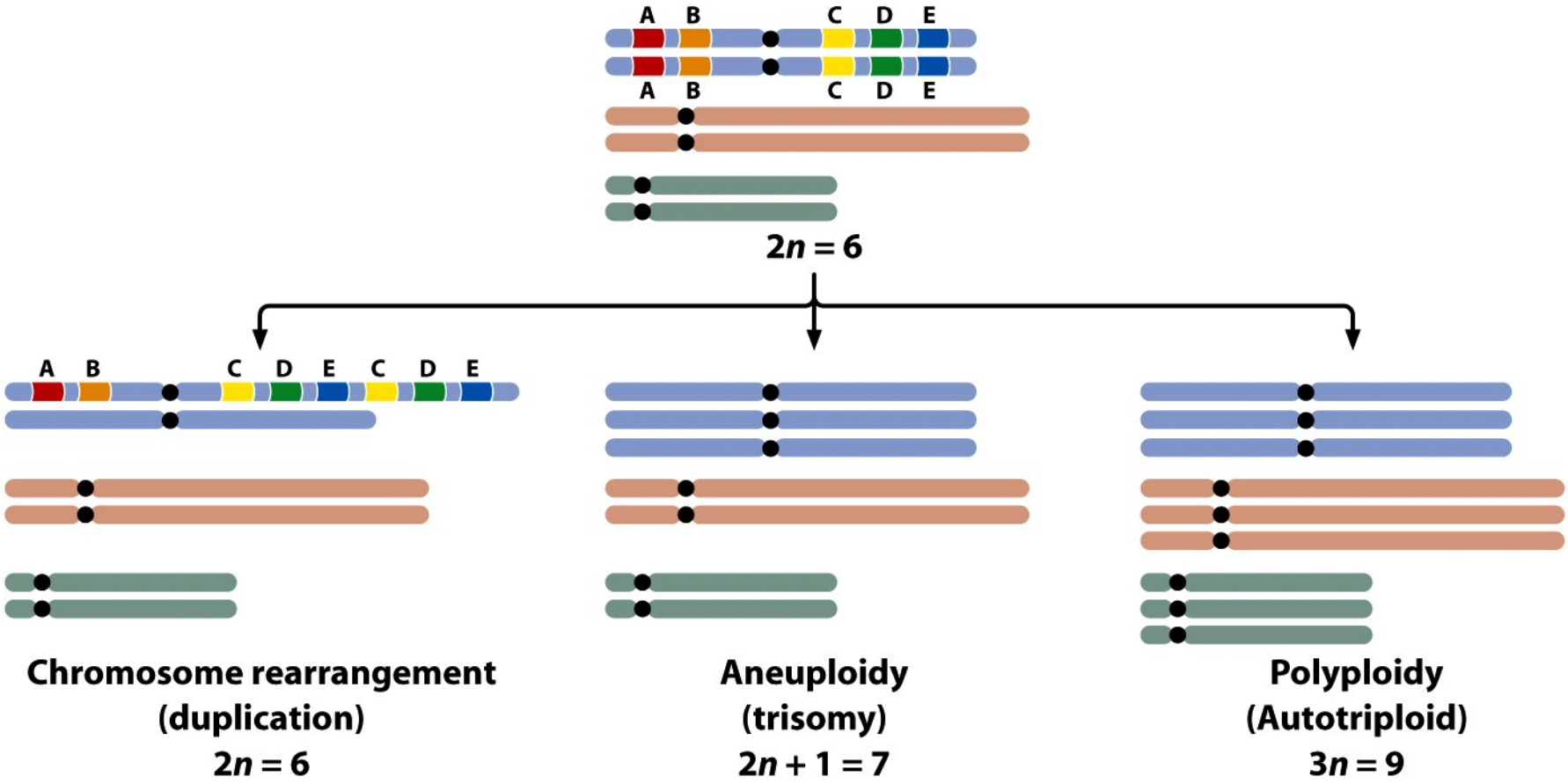
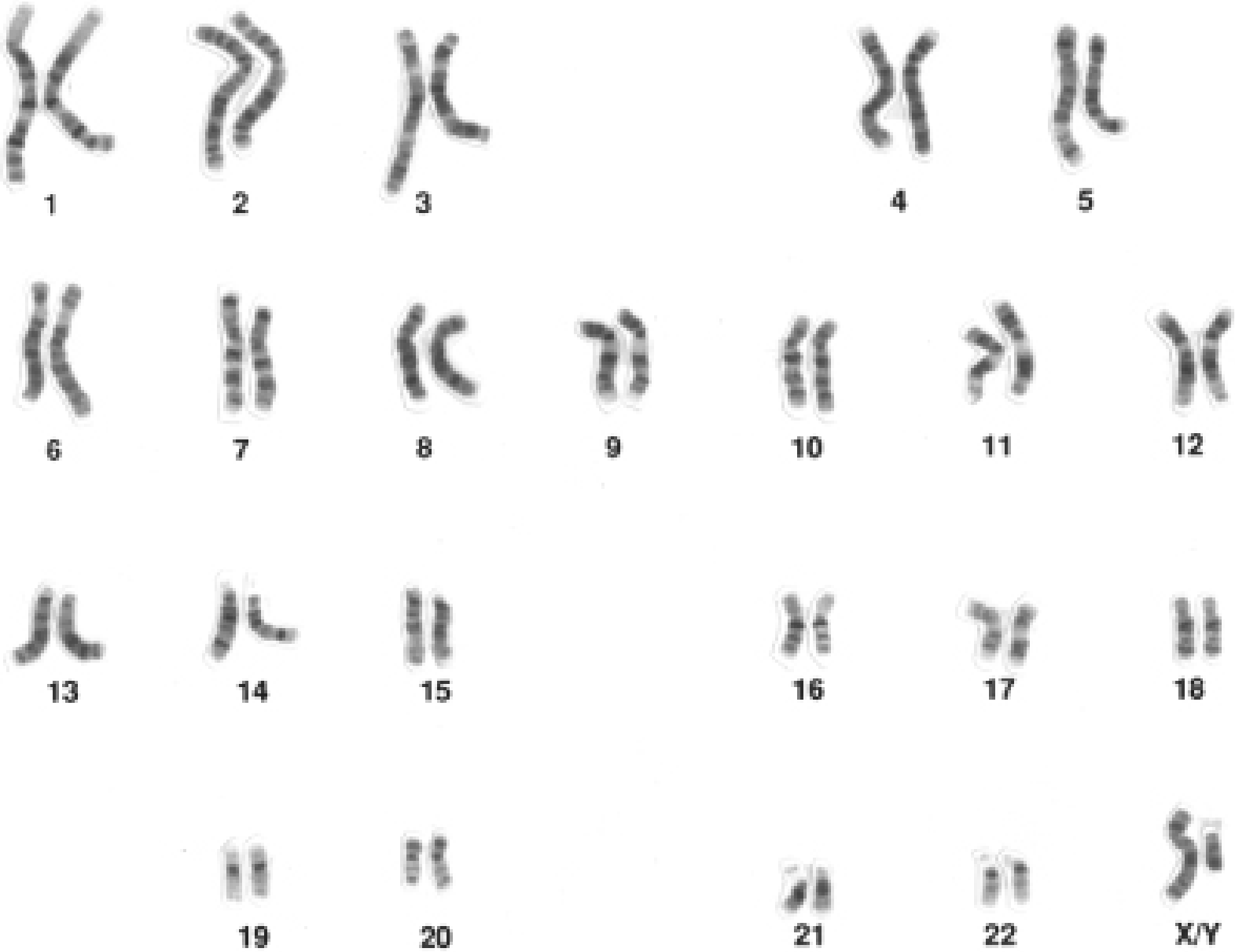



**Figure 9-3**
***Genetics: A Conceptual Approach, Third Edition***


1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X/Y

# ¿Es “mala” la Mutación?

- Provoca muchos desórdenes genéticos
- Fuente de Variabilidad Genética
  - Evolución
  - Herramienta Análisis Genético
    - Marcadores Moleculares
    - Análisis de Mutantes