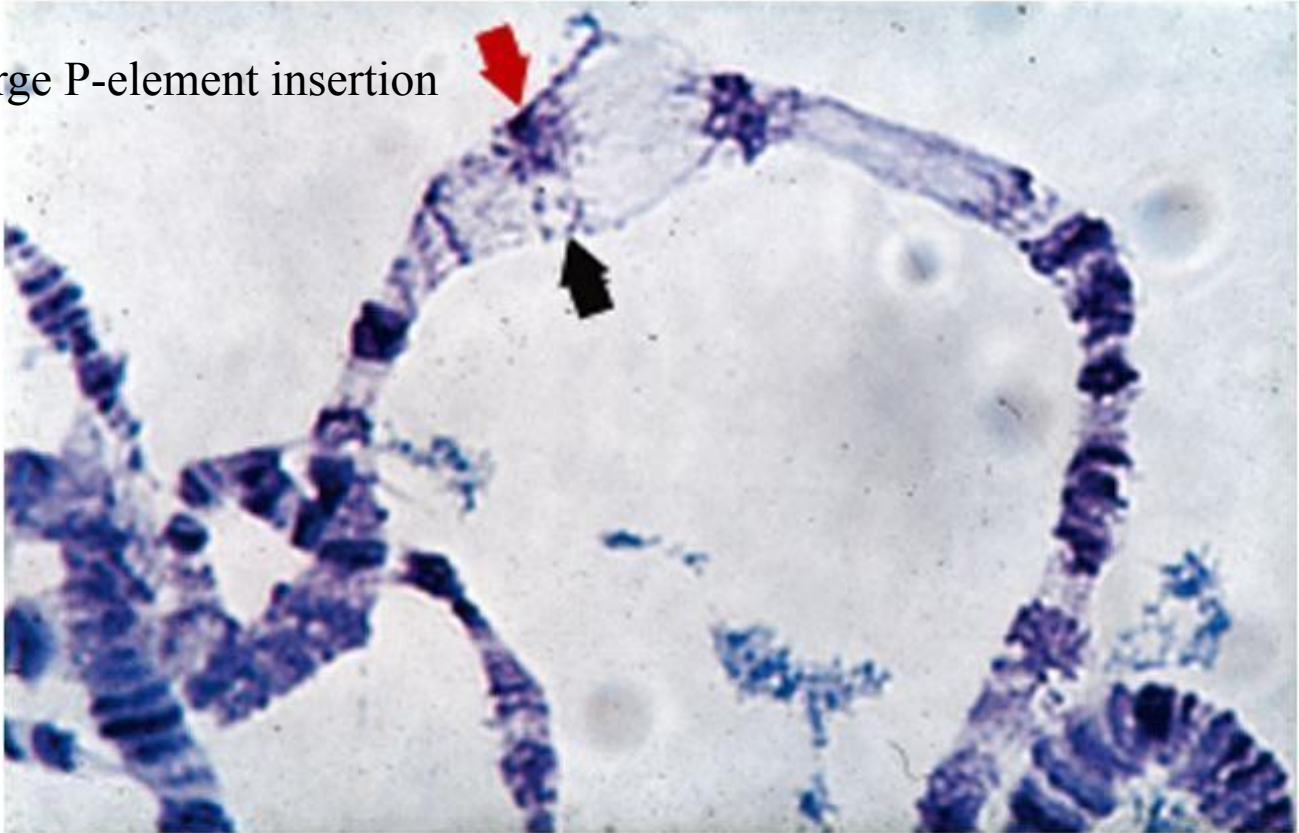
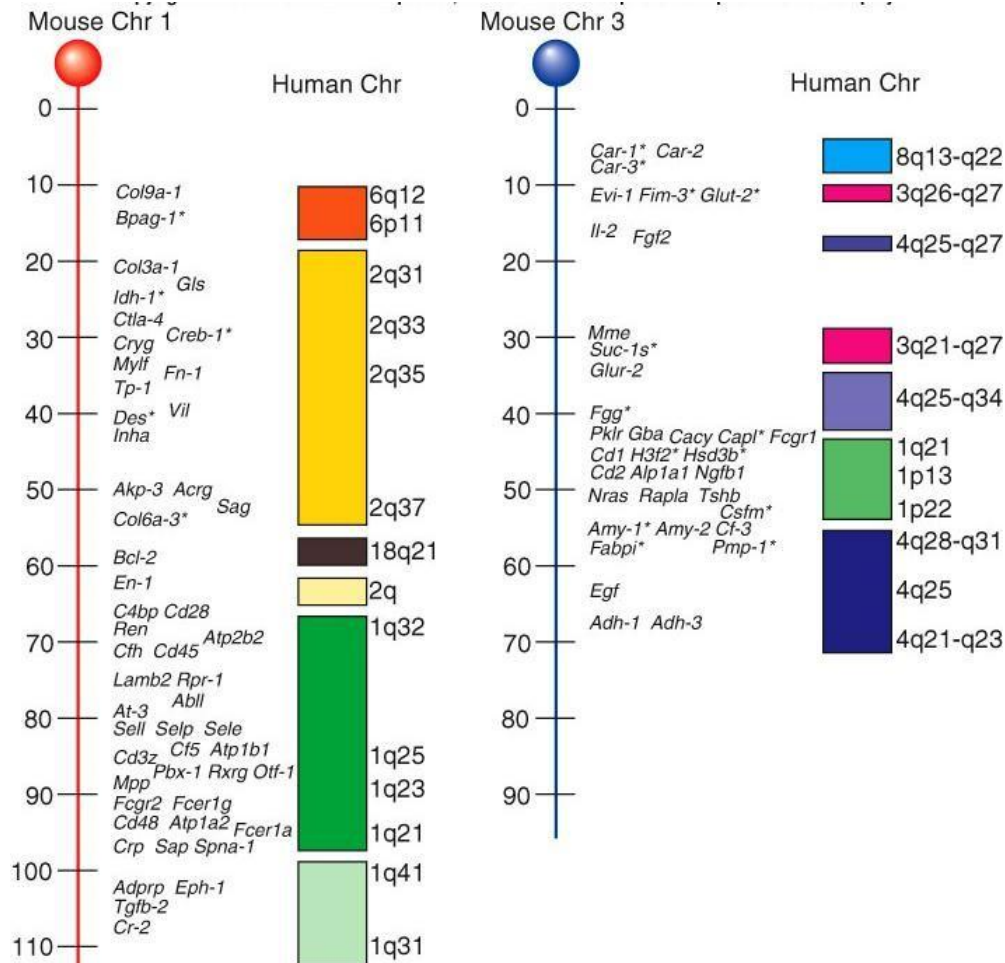


# Kromozom Yapı Değişimleri

Large P-element insertion



# Fare kromozomları insan kromozomlarının 170 parçaya kırılması ile oluşturulabiliyor.



**TABLE 13.1 Chromosomal Rearrangements and Changes in Chromosome Number**

**Chromosomal Rearrangements**

Before

After

**Deletion:** Removal of a segment of DNA



**Duplication:** Increase in the number of copies of a chromosomal region



**Inversion:** Half-circle rotation of a chromosomal region



**Translocations:**

*Nonreciprocal:* Unequal exchanges between nonhomologous chromosomes



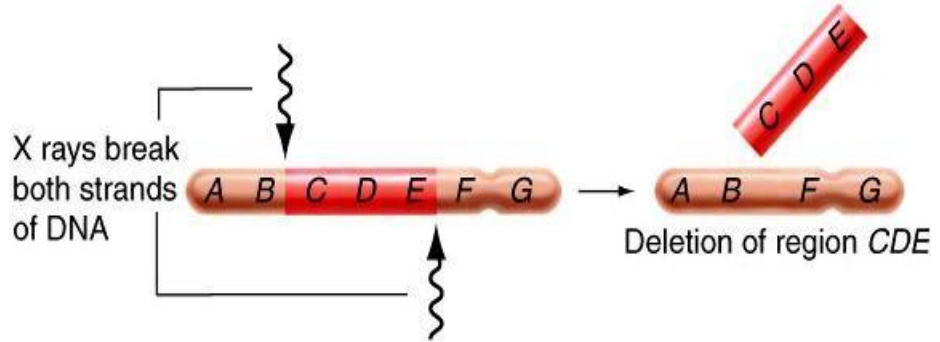
*Reciprocal:* Parts of two nonhomologous chromosomes trade places



**Transposition:** Movement of short DNA segments from one position in the genome to another

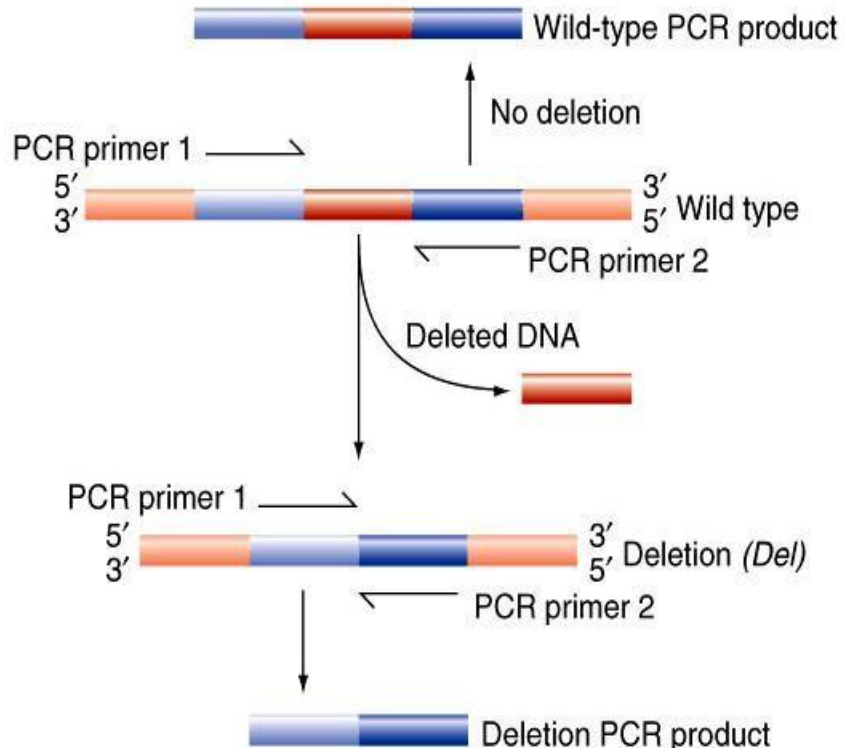


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**(a) DNA breakage may cause deletions**



# Delesyon

**(b) Detecting deletions using PCR**



Delesyon heterozigot halde fenotipi deęiřtirebilir.



**Wild type**  
(two copies of *Notch*<sup>+</sup>)

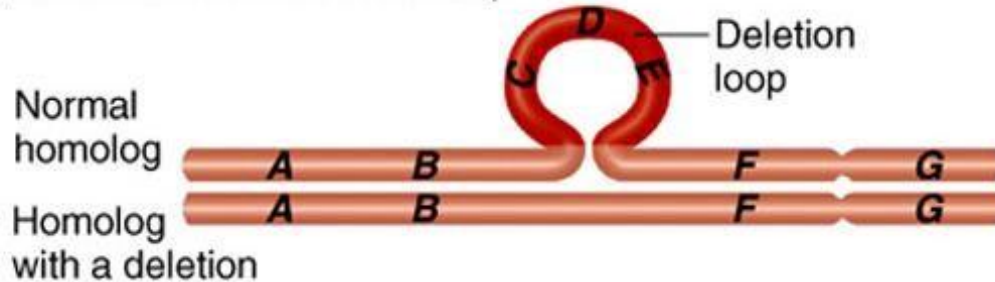


**Del / +**  
(one copy of *Notch*<sup>+</sup>)

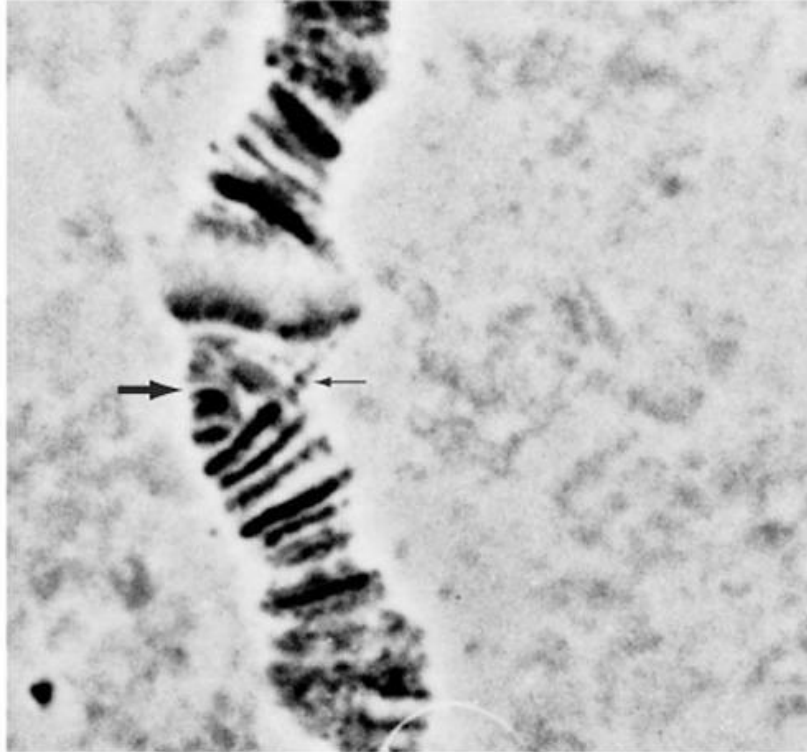
# Delesyonlu kromozomların eşleşmesi

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(a) Formation of a deletion loop

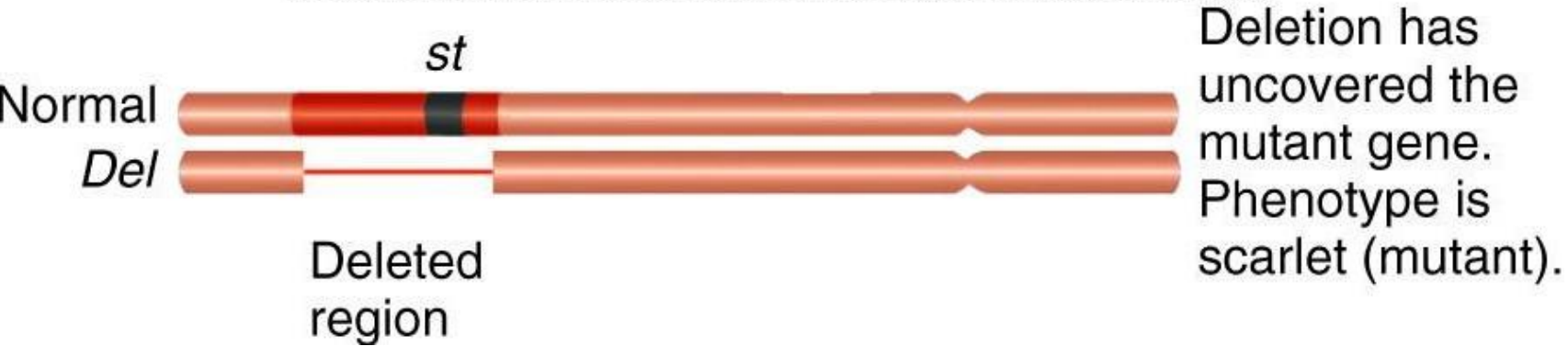


(b) A deletion loop in *Drosophila* polytene chromosomes



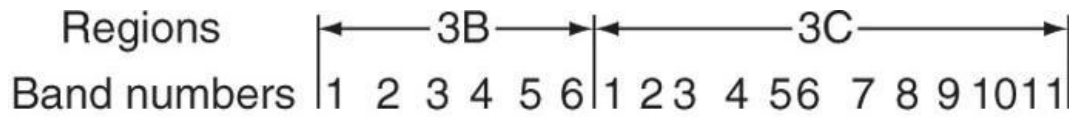
Mayozda eşleşme

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# Genetik haritalamada delesyonların kullanımı



Small segment of banded polytene chromosome



Specific deletions

- Df 258-45*
- Df 67c23*
- Df N8*
- Df 264-32*
- Df 264-33*

Phenotype of mutation/*Del* heterozygote

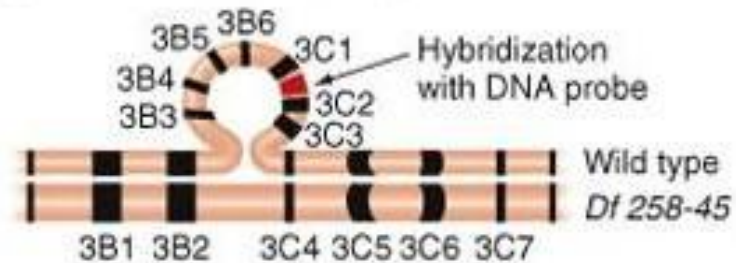
- $w^- rst^+ fa^+$
- $w^- rst^+ fa^+$
- $w^- rst^- fa^-$
- $w^+ rst^- fa^-$
- $w^+ rst^+ fa^-$



Regions of genes

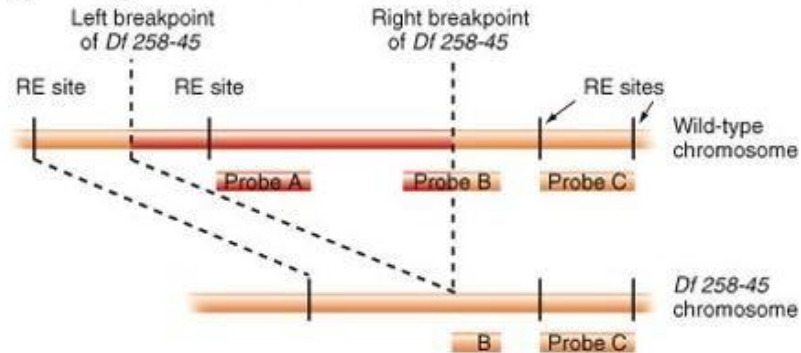
# Delesyon haritalamasında moleküler yöntemler de kullanılabilir (*in situ* melezleme)

(a) Characterizing deletions with *in situ* hybridization.

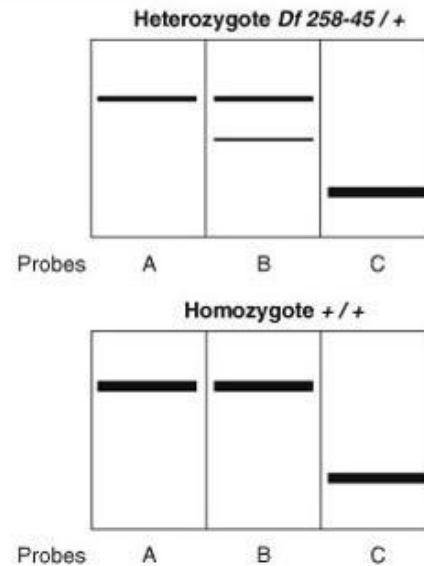


# Delesyon haritalamasında Southern blot

(b) Probing the location of a deletion breakpoint.



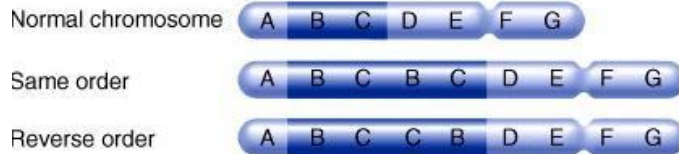
(c) Southern blot results



# Duplikasyonlar

## (a) Types of duplications

### Tandem duplications



### Nontandem (dispersed) duplications



## (c) Different kinds of duplication loops

### Duplicated chromosome



Mayozda Kromozom eşleşmesi

DNA replikasyonu sırasında  
Homolog olmayan krossin over'ler

**(a) Duplication heterozygosity can cause visible phenotypes.**



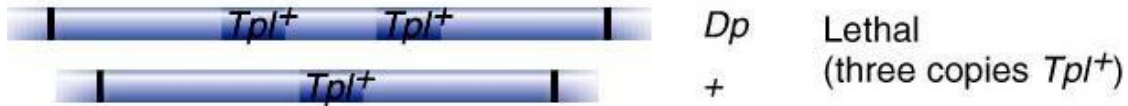
Wild-type wing:  
two copies of *Notch<sup>+</sup>* gene



Aberrant  
wing veins

Three copies of *Notch<sup>+</sup>* gene

(b) For rare genes, survival requires exactly two copies.



## Genotype of X chromosomes



Wild type



Bar



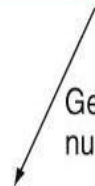
Double-Bar

## Phenotype

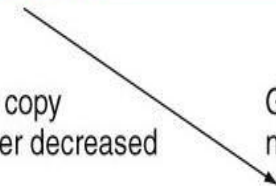
Wild-type eye



## Out-of-register pairing during meiosis in a Bar-eyed female



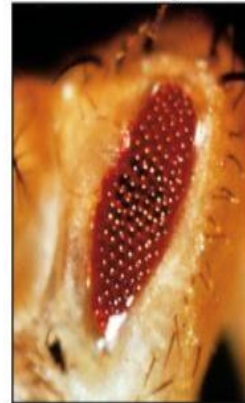
Gene copy number decreased



Gene copy number increased



Double-Bar eye

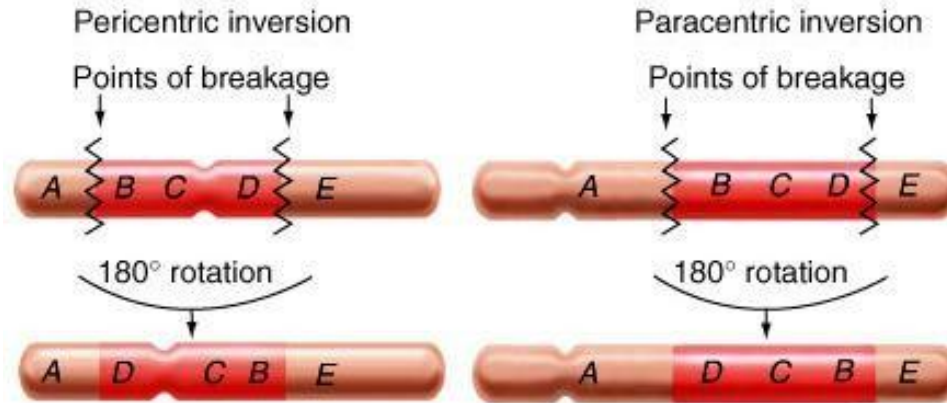


Gen aileleri örn. Hemoglobin genleri  
duplikasyonla oluşur.

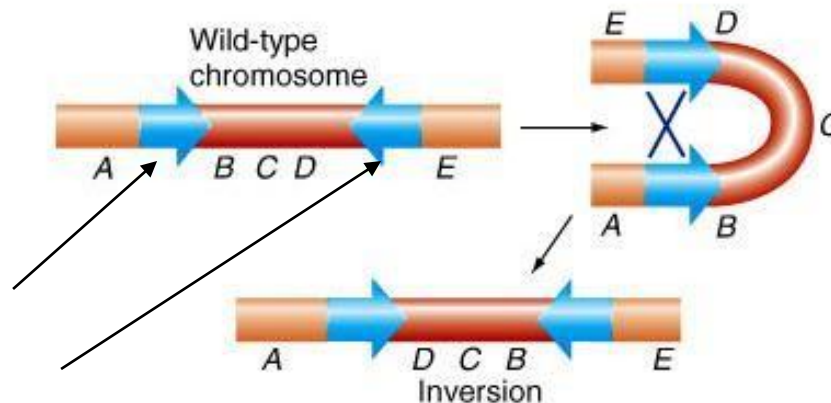


# Inversiyon

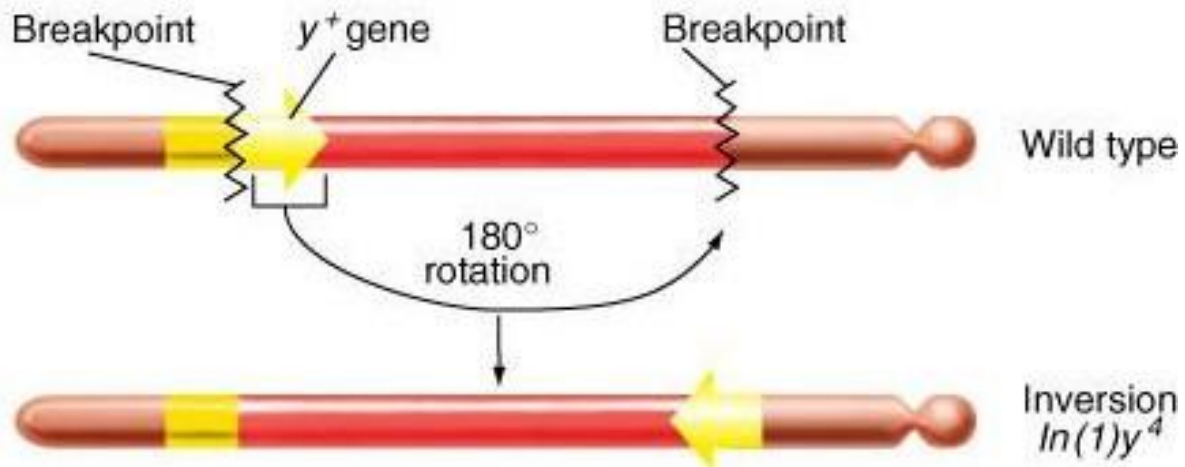
(a) Chromosome breakage can produce inversions.



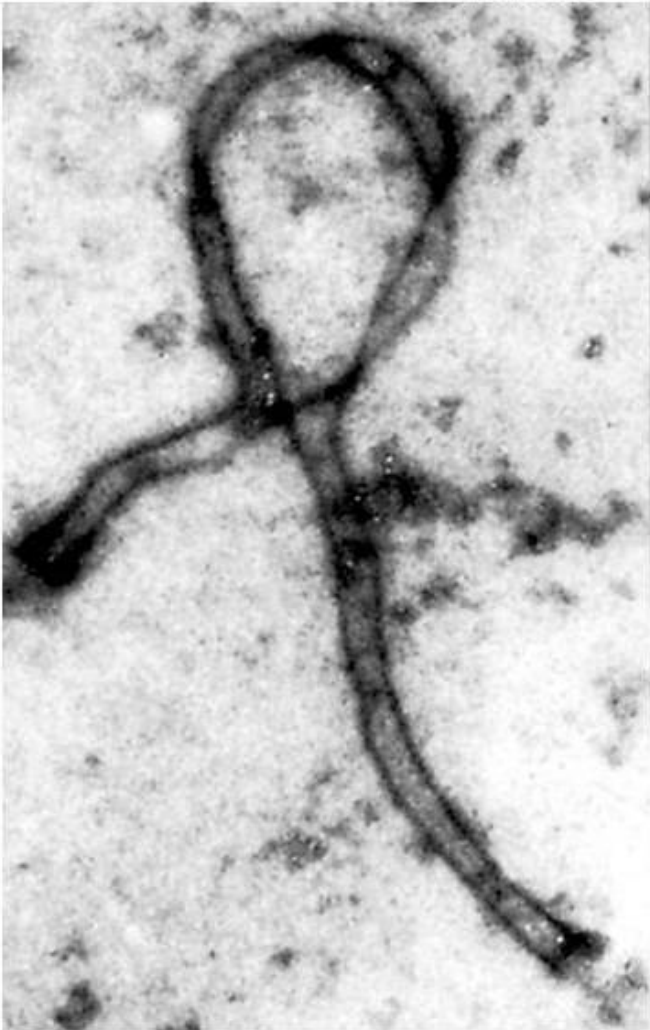
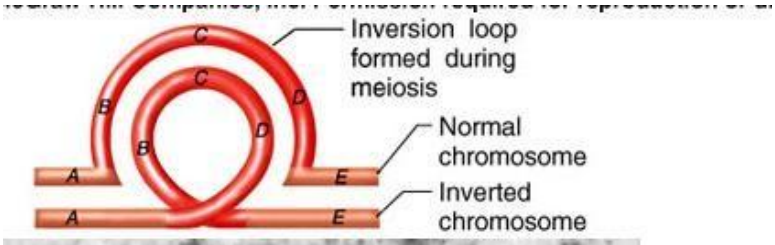
(b) Intrachromosomal recombination can also cause inversions.



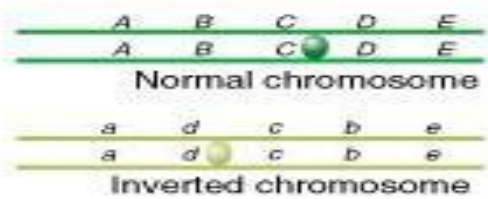
**(c) Inversions can disrupt gene function.**



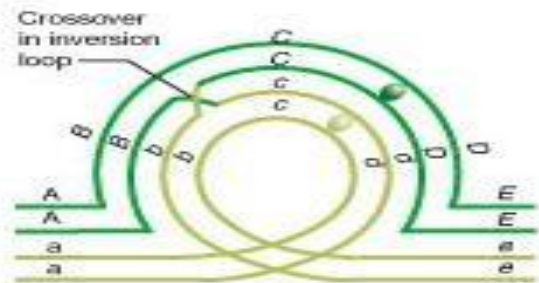
# Inversiyon loop



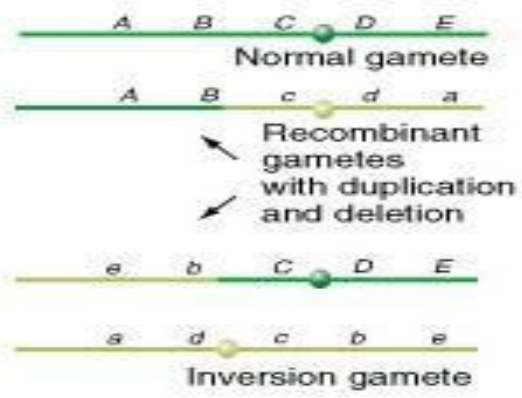
(a) Pericentric inversion heterozygote



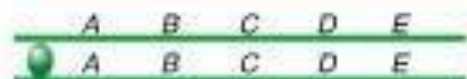
Pairing during meiosis I



Completion of both meiotic divisions



(b) Paracentric inversion heterozygote



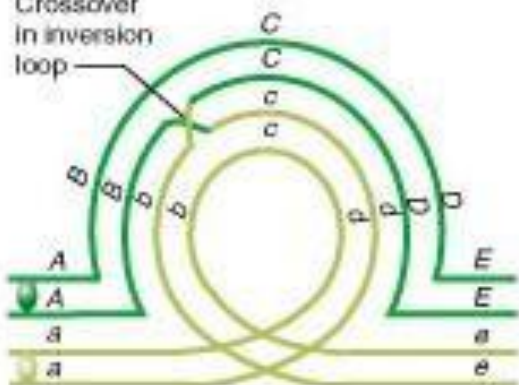
Normal chromosome



Inverted chromosome

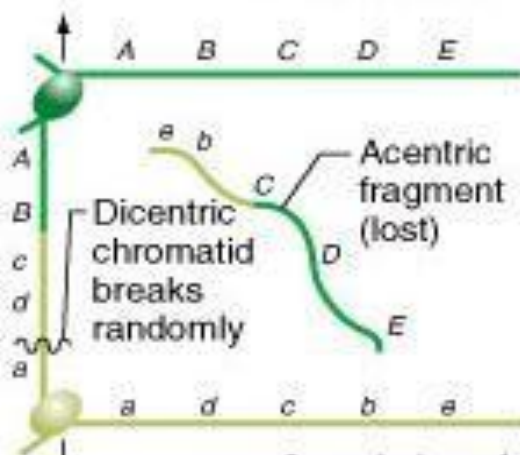
Pairing during meiosis I

Crossover in inversion loop



Chromosome separation during anaphase of meiosis I

Chromosome separation during anaphase of meiosis I



Completion of both meiotic divisions



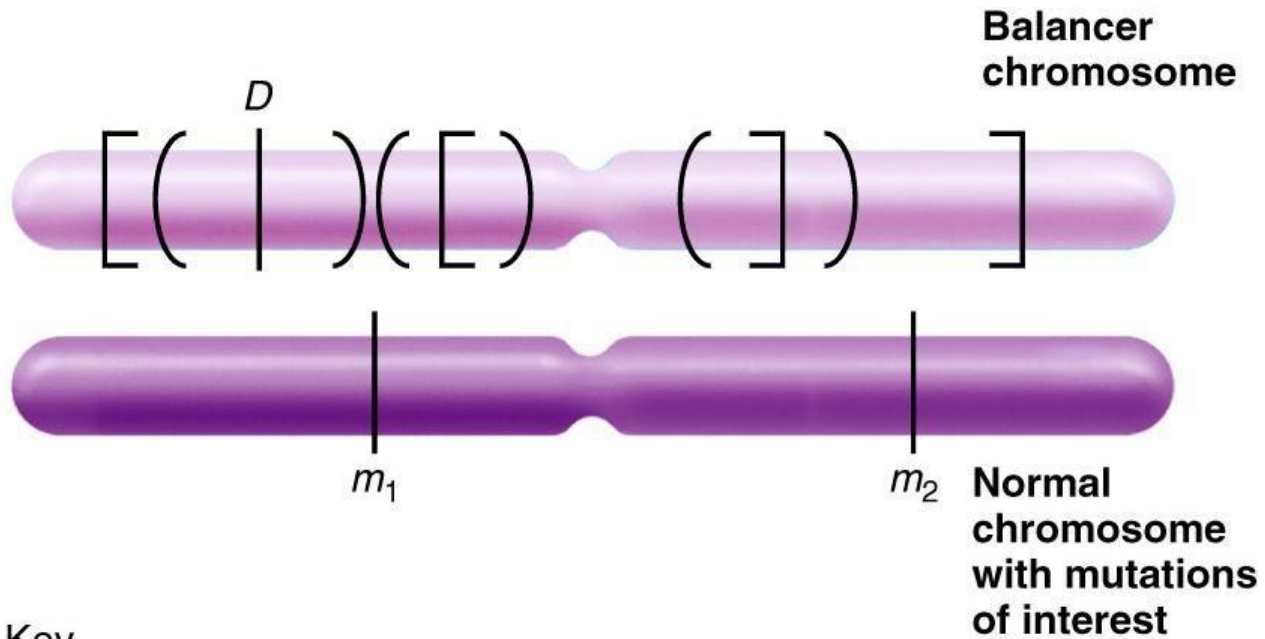
Normal gamete



Broken, deleted products of dicentric chromosome



Inversion gamete



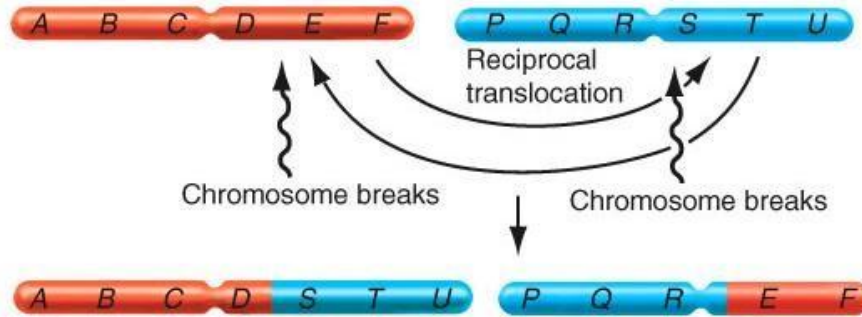
Key

$[ ]$  Breakpoints of pericentric inversions

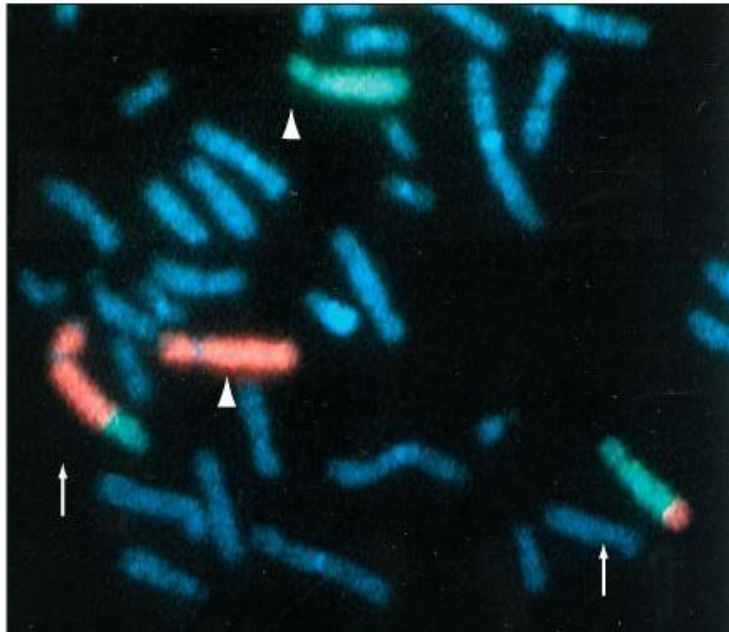
$( )$  Breakpoints of paracentric inversions

# Resiprokal translokasyon

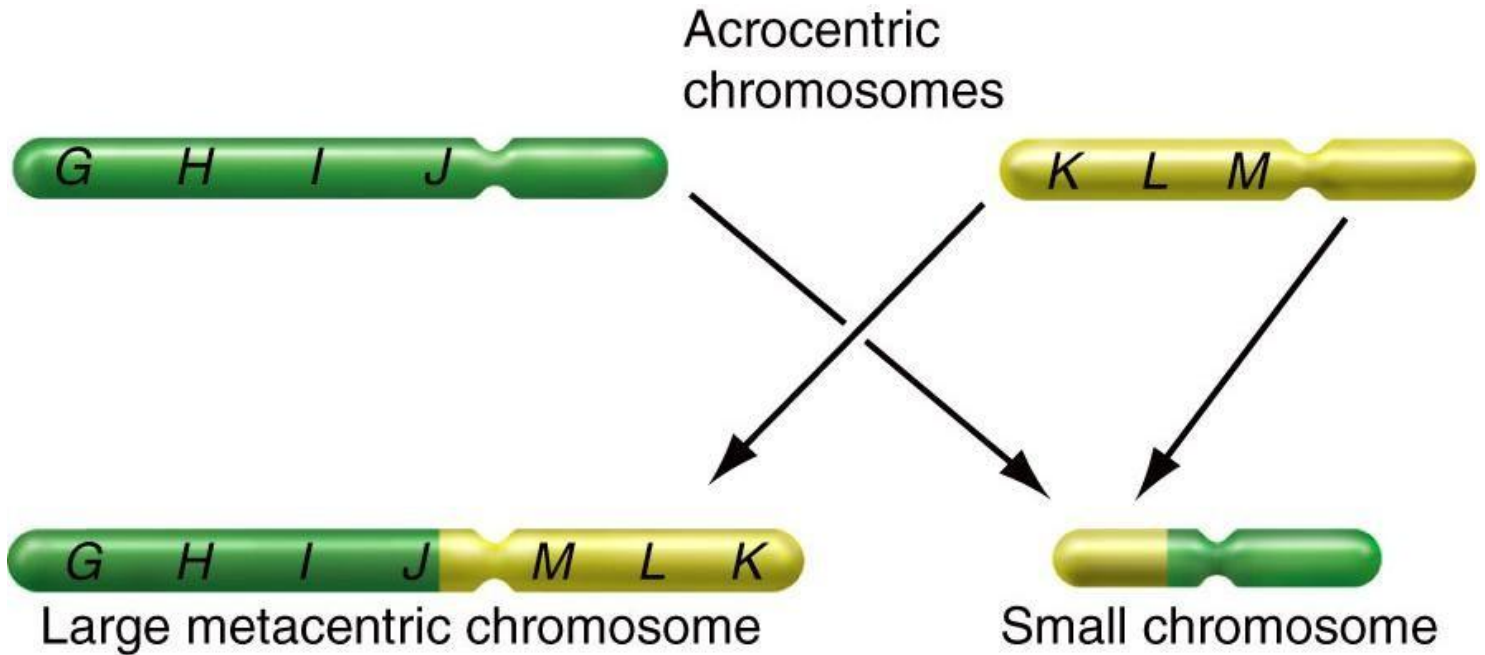
(a) Two chromosome breaks can produce a reciprocal translocation.



(b) Chromosome painting reveals a reciprocal translocation.



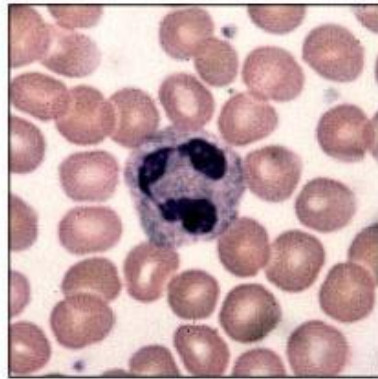
# Robertsonian translokasyonu



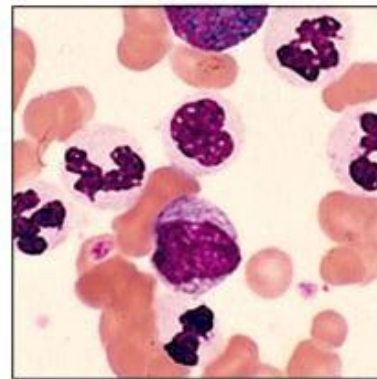
**A Robertsonian translocation**



(a) Leukemia patients have too many white blood cells.

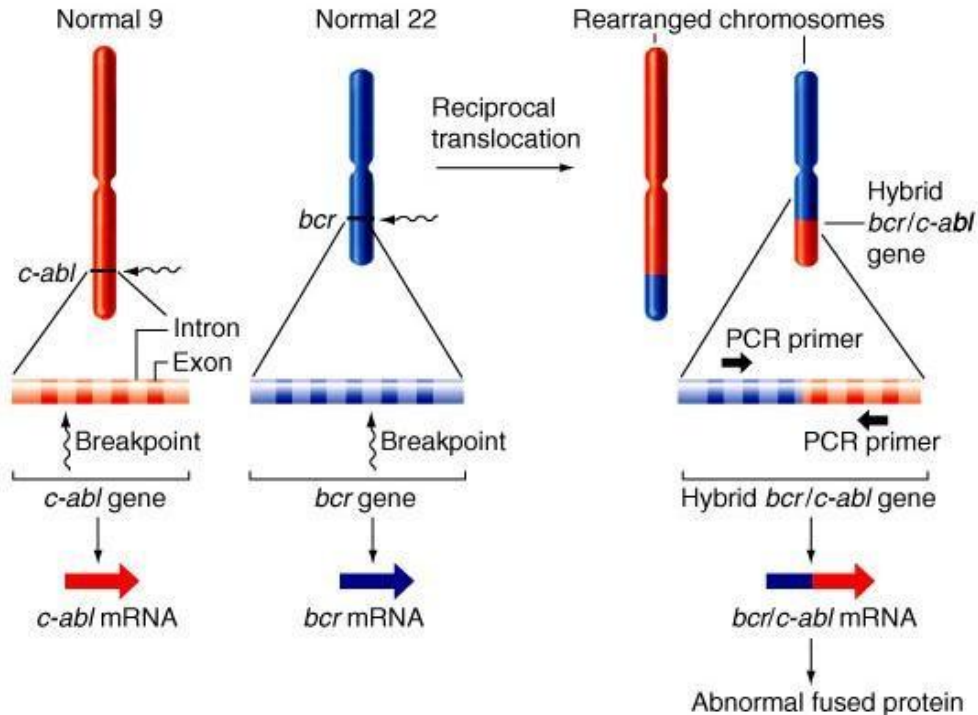


Normal

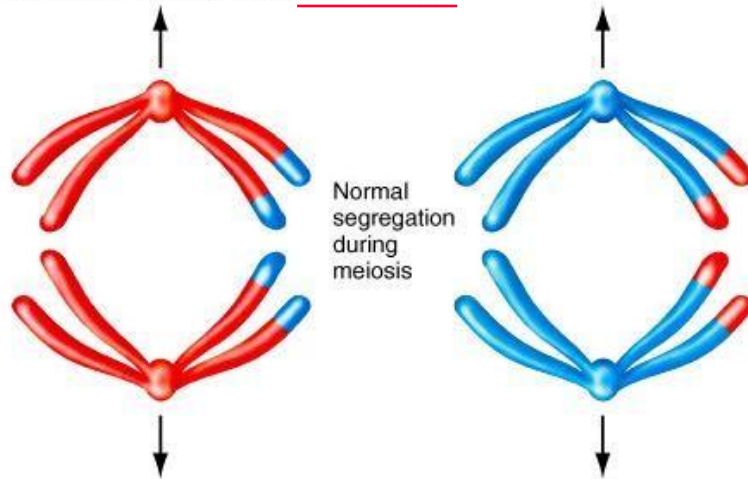


Leukemic

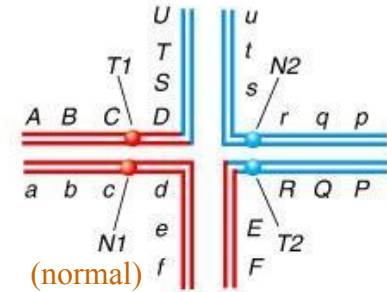
(b) The genetic basis for chronic myelogenous leukemia.



(a) Segregation in a translocation homozygote



(b) Chromosome pairing in a translocation heterozygote



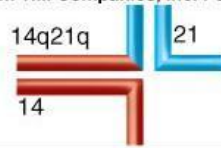
(c) Segregation in a translocation heterozygote

Segregation pattern	Alternate		Adjacent - 1		Adjacent - 2 (less frequent)	
	Balanced $N1 + N2$	Balanced $T1 + T2$	Unbalanced $T1 + N2$	Unbalanced $N1 + T2$	Unbalanced $N1 + T1$	Unbalanced $N2 + T2$
Gametes						
Type of progeny when mated with normal $abcdefpqrstu$ homozygote	$abcdefpqrstu$	$ABCDEF PQRSTU$	(no E, F) None surviving	(no S, T) None surviving	None surviving	None surviving

(d) Semisterility in corn



# Translokasyonlu Down

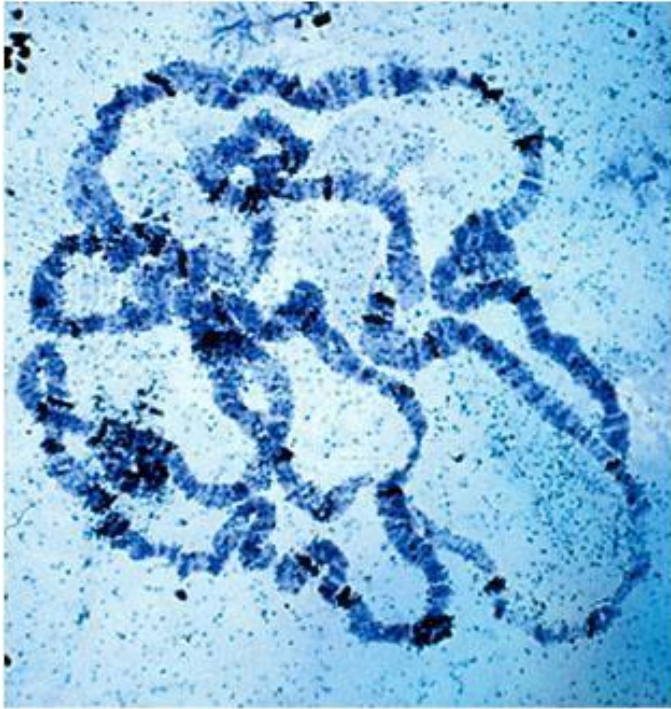


Segregation pattern	Alternate		Adjacent-1	Adjacent-2		
Gametes	<p>14 21</p> <p>Balanced normal</p>	<p>14q21q</p> <p>Balanced Robertsonian translocation</p>	<p>14q21q 21</p> <p>Unbalanced</p>	<p>14</p> <p>Unbalanced</p>	<p>14q21q 14</p> <p>Unbalanced</p>	<p>21</p> <p>Unbalanced</p>
Fertilization with normal gamete	<p>14 21</p> <p>Normal phenotype</p>	<p>14 21</p> <p>Normal phenotype (carrier)</p>	<p>14 14q21q 21</p> <p>Down syndrome</p>	<p>14 21</p> <p>Lethal</p>	<p>14 14q21q 21</p> <p>Lethal</p>	<p>14 21</p> <p>Lethal</p>

# Transposozisyon: Küçük DNA parçalarının hareketi



# *copia* TE

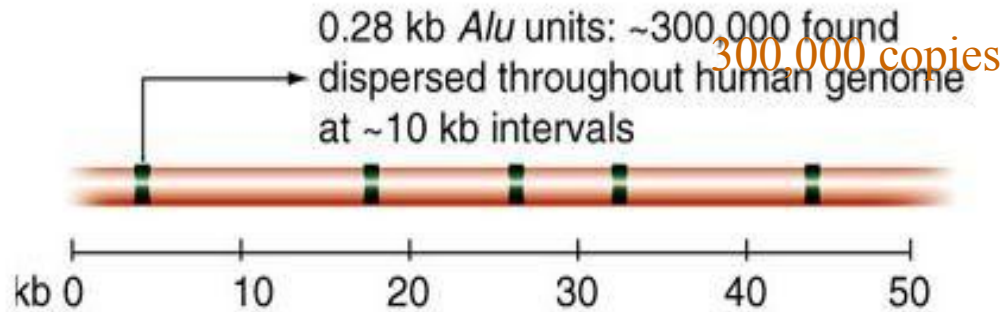


# Transpozabl elementler

Genom içerisinde hareket edebilen DNA parçaları

1. 50-10,000 bp
2. Kopya sayısı 1 den 100e ve 1000e kadar değişir
3. Memelilerde TEs: LINES (L1) ve SINES (Alu)

(a) *Alu* SINEs in the human genome.

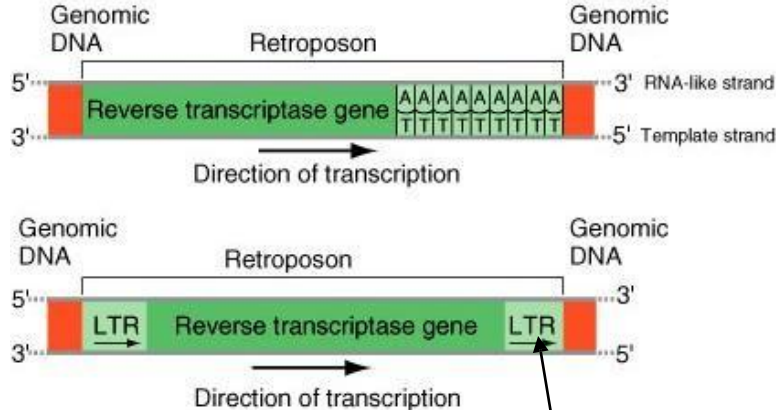


(b) TEs cause mottling in corn.

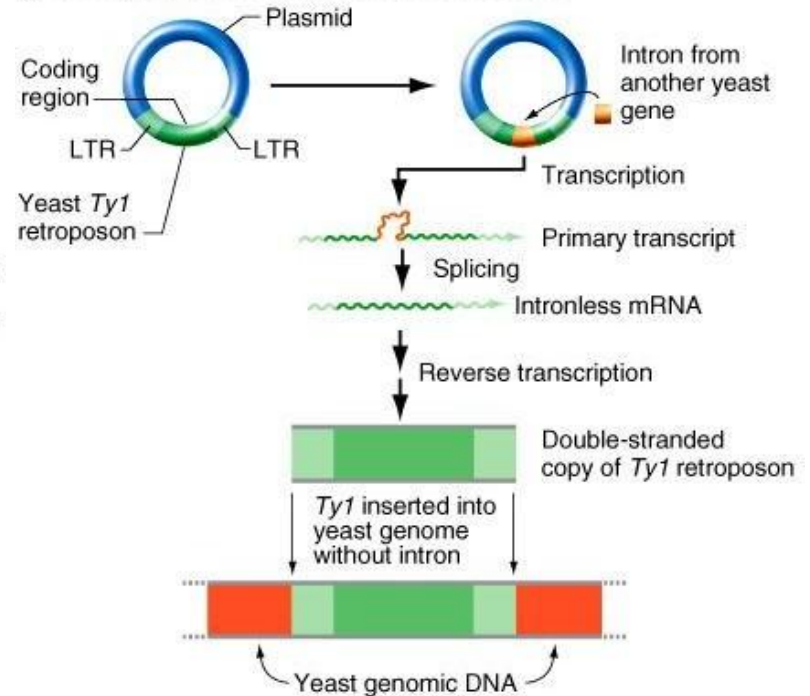


# Retropozonlar: RNA ara moleküllerinin transkripsiyonu ile oluşurlar.

(a) Two kinds of retroposons.



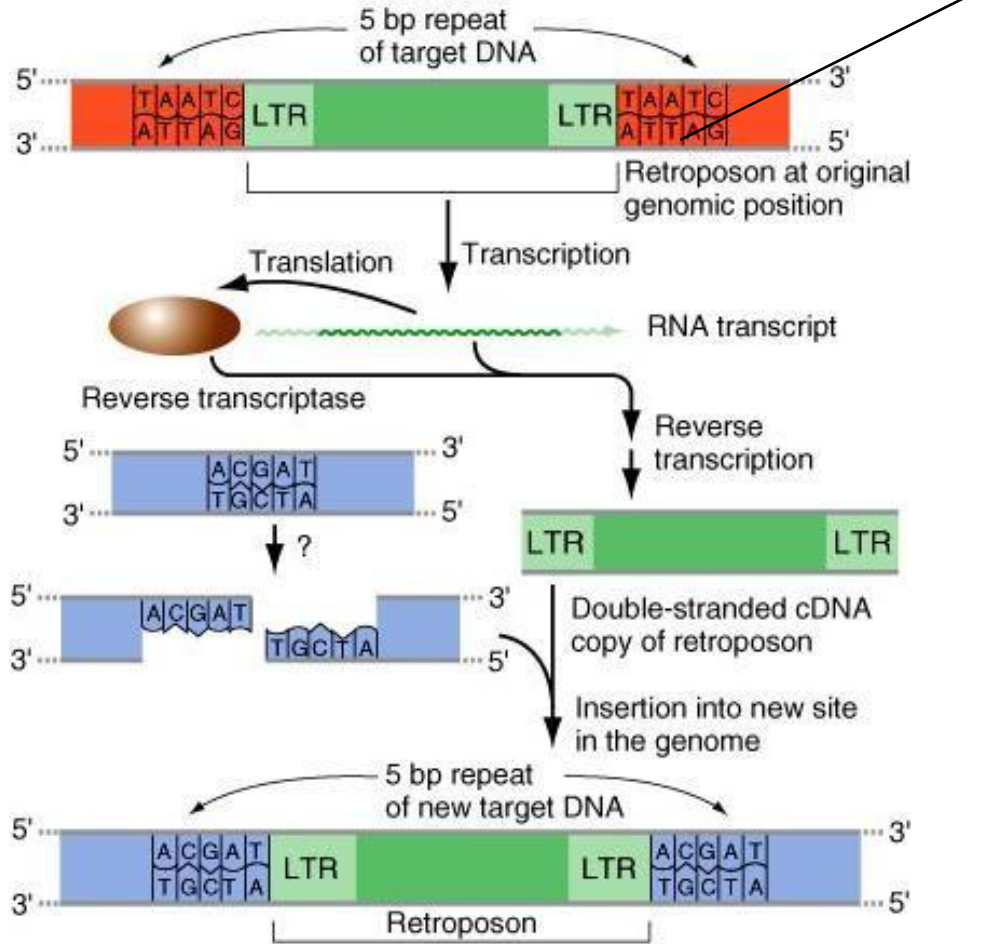
(b) Retroposons move via RNA intermediates.



Retrovirus (RNA tümör virüsü)

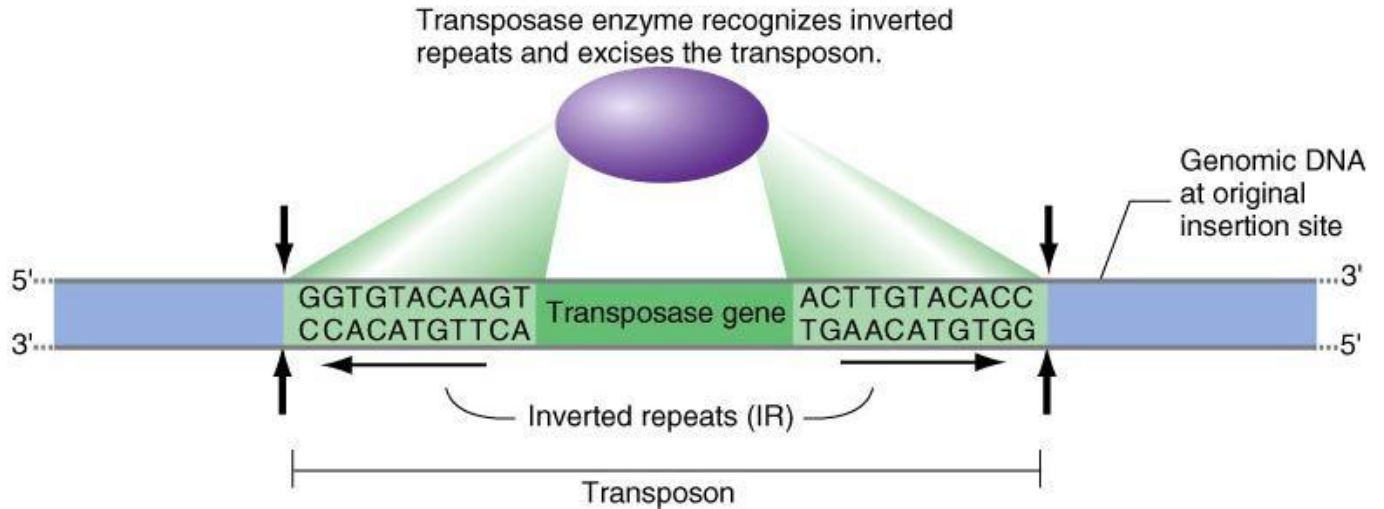


(c) How retroposons move.

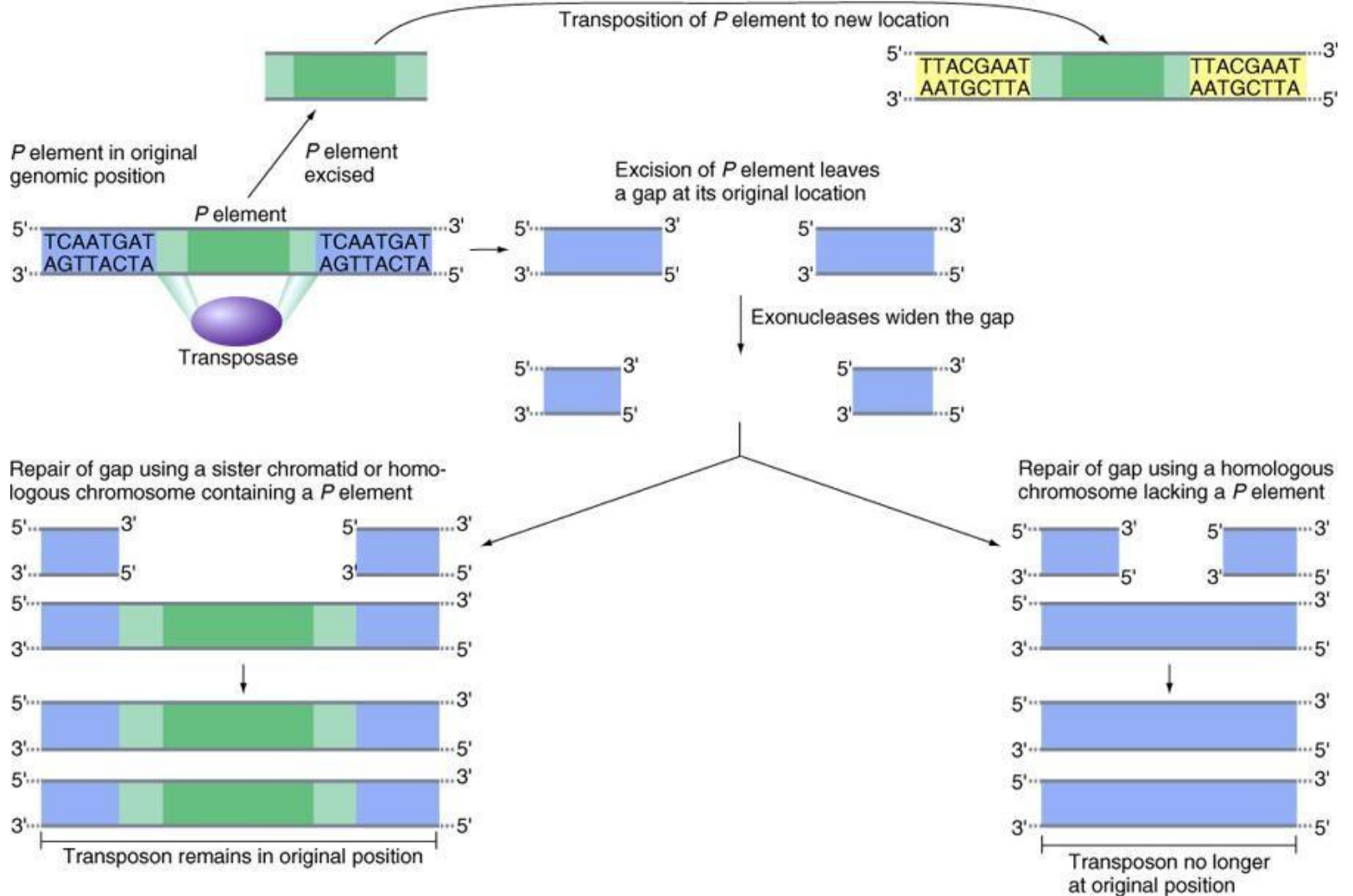


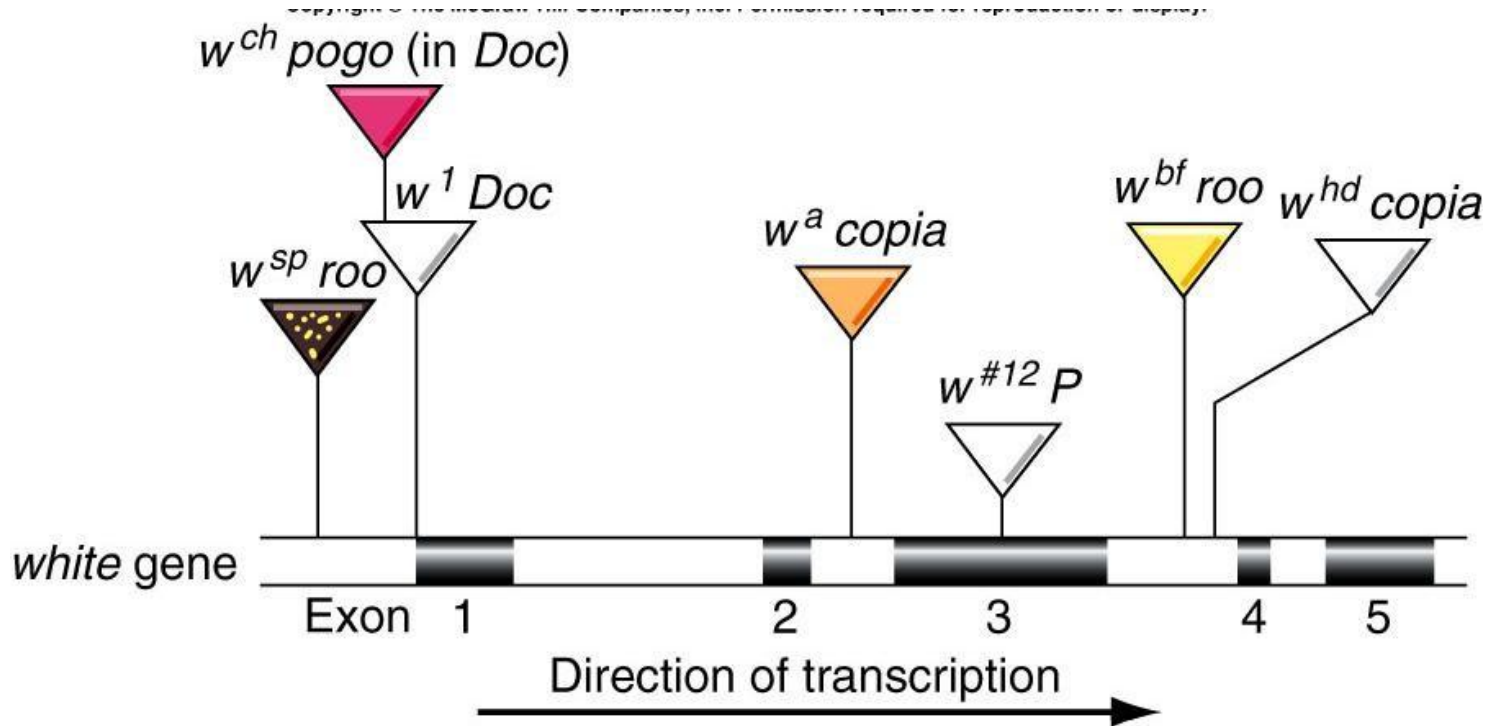
# Transpozonlar

(a) Transposon structure

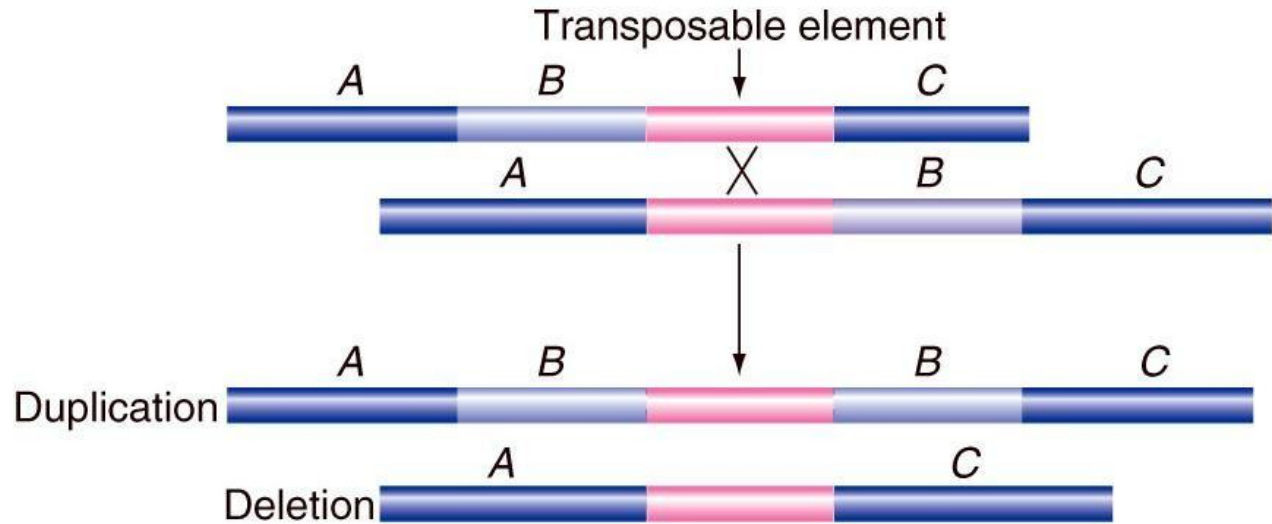


**(b) How P element transposons move**

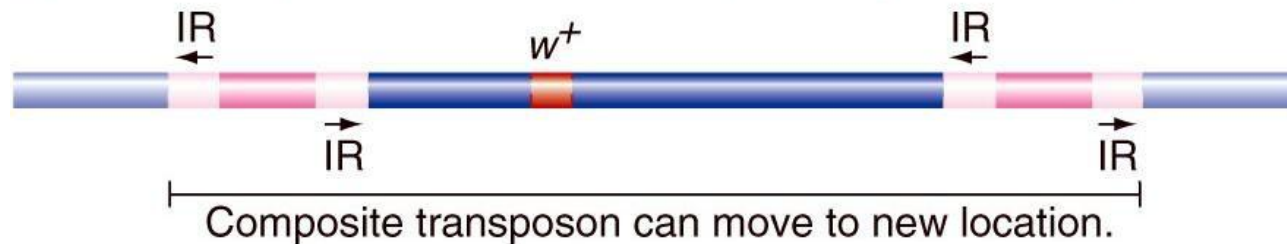




**(a) Unequal crossing-over between TEs.**



**(b) Two transposons can form a large, composite transposon.**



**TABLE 13.1 Chromosomal Rearrangements and Changes in Chromosome Number**

**Changes in Chromosome Number**

**Euploidy:** Cells that contain only complete sets of chromosomes

*Diploidy* ( $2x$ ): Two copies of each homolog



*Monoploidy* ( $x$ ): One copy of each homolog



**Polyploidy:** More than the normal diploid number of chromosome sets

*Triploidy* ( $3x$ ): Three copies of each homolog



*Tetraploidy* ( $4x$ ): Four copies of each homolog



**Aneuploidy:** Loss or gain of one or more chromosomes producing a chromosome number that is not an exact multiple of the haploid number

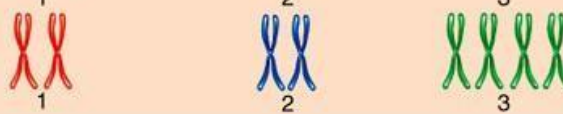
*Monosomy* ( $2n - 1$ )



*Trisomy* ( $2n + 1$ )



*Tetrasomy* ( $2n + 2$ )



XY  
Normal ♂

X Y



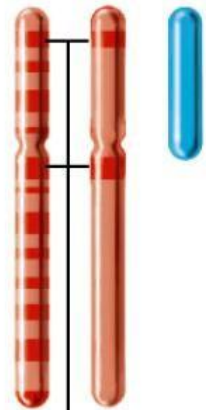
XX  
Normal ♀

X X



XXY  
Klinefelter ♂

X X Y

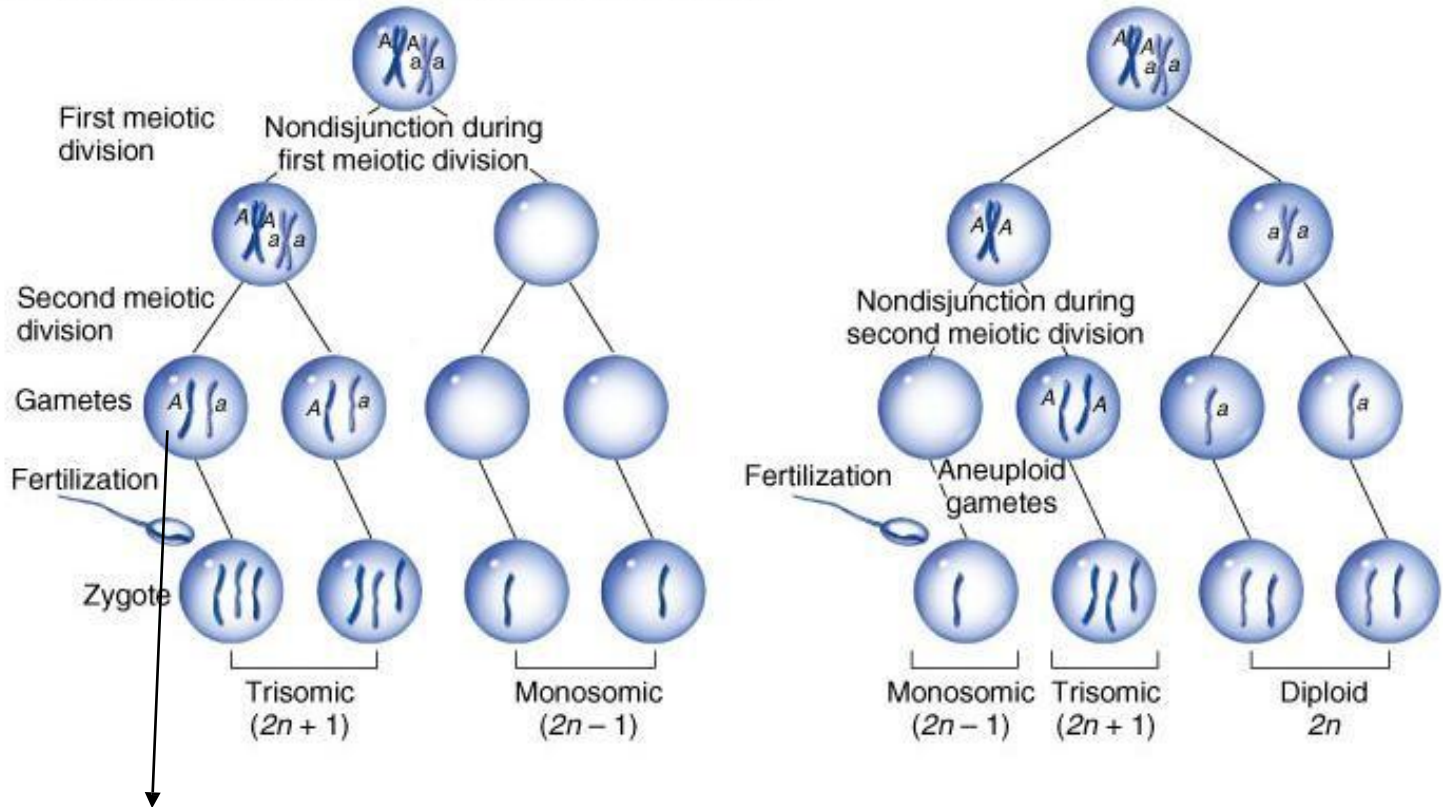


■ Active genes

Some X genes expressed  
at twice the level of normal  
males

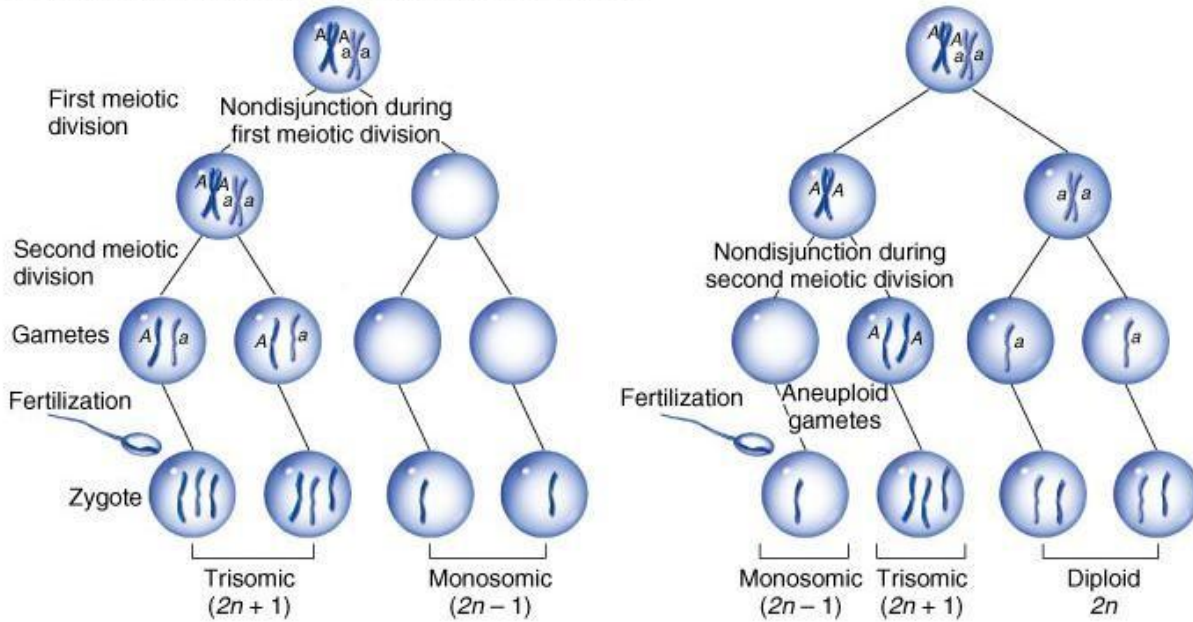
# Nondisjunction in meiotic recombination results in aneuploidy

(a) Nondisjunction can occur during either meiotic division.

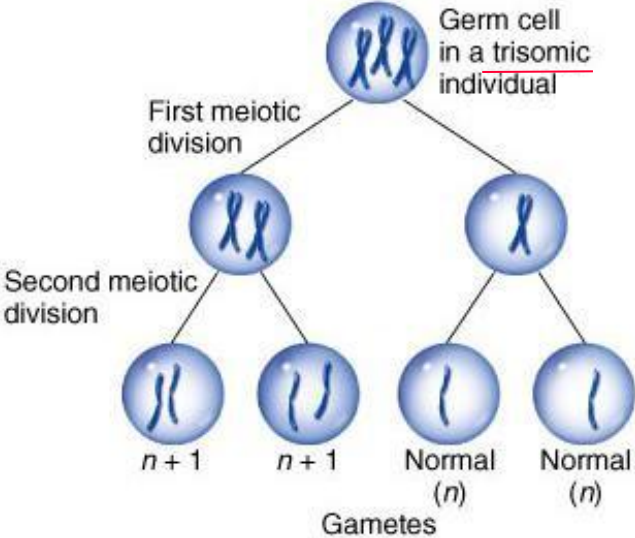
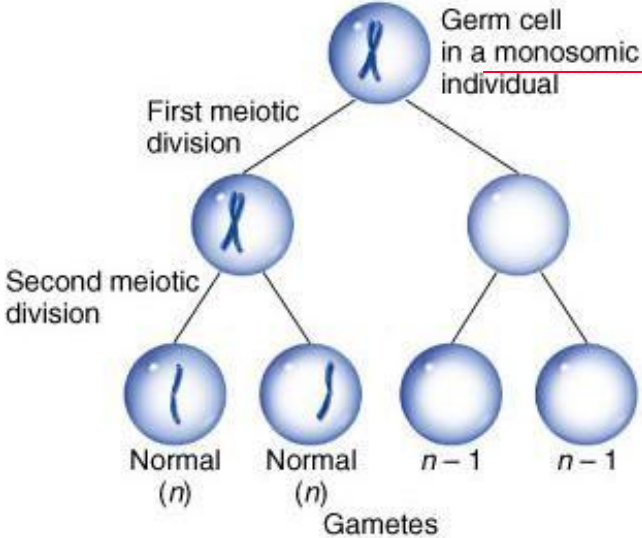




**(a) Nondisjunction can occur during either meiotic division.**



**(b) Aneuploids beget aneuploid progeny.**



**(a) Mitotic nondisjunction**

Diploid cell at metaphase



Mitosis



Nondisjunction of one set of sister chromatids at anaphase produces aneuploid daughter cells.



Trisomic cell



Monosomic cell

**(b) Mitotic chromosome loss**

Diploid cell at metaphase



Mitosis



Lagging chromatid at anaphase is lost. Aneuploid daughter cell produced.



Monosomic cell



Normal diploid cell

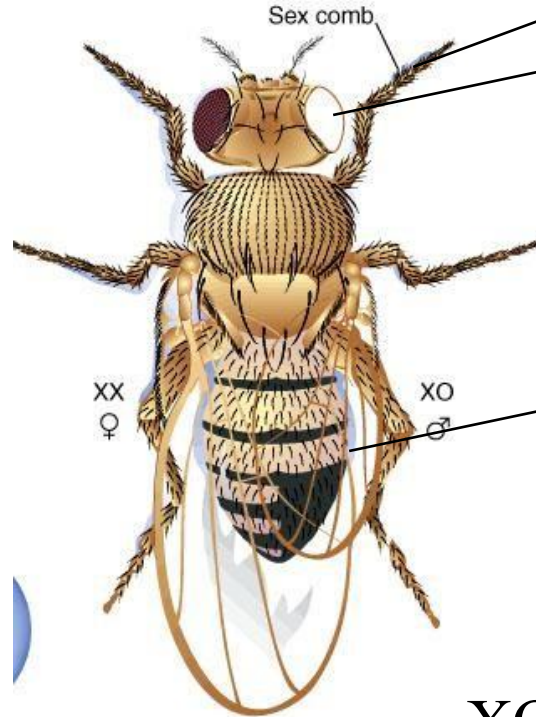
**(c) A**



reproduction or display.  
c) A gynandromorph

Male-specific

White eye



XX  
♀

XO  
♂

Minute wing

XX

XO

$X^{w+m+} X^{w-m-}$

$X^{w-m-} O$

## Changes in Chromosome Number

**Euploidy:** Cells that contain only complete sets of chromosomes

*Diploidy* ( $2x$ ): Two copies of each homolog

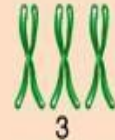
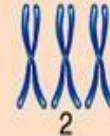


*Monoploidy* ( $x$ ): One copy of each homolog

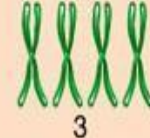
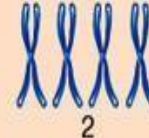
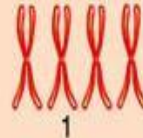


**Polyploidy:** More than the normal diploid number of chromosome sets

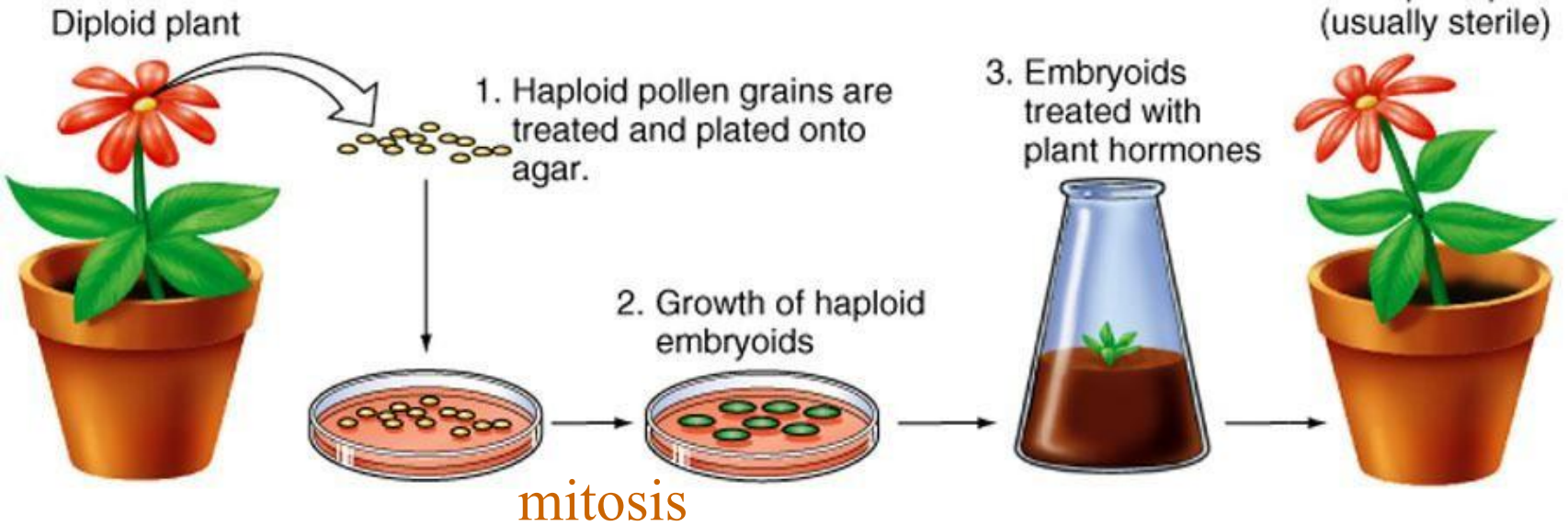
*Triploidy* ( $3x$ ): Three copies of each homolog



*Tetraploidy* ( $4x$ ): Four copies of each homolog



### (a) How to create a monoploid plant



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**(b) Using monoploid plants to select for herbicide resistance**

1. Monoploid plant sensitive to selective agent



2. Cell walls of somatic cells removed; cells exposed to mutagen; plated on agar containing selective agent



3. Cells with resistance mutation grow into resistant embryoids.



4. Resistant monoploid (sterile)



5. Treatment of somatic cells with colchicine

6. Cells become diploid

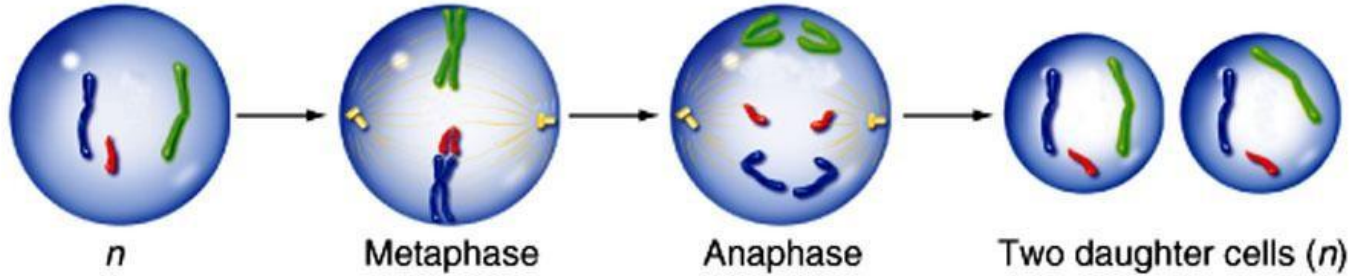


7. Cells can be grown into diploid homozygous resistant plant (fertile).

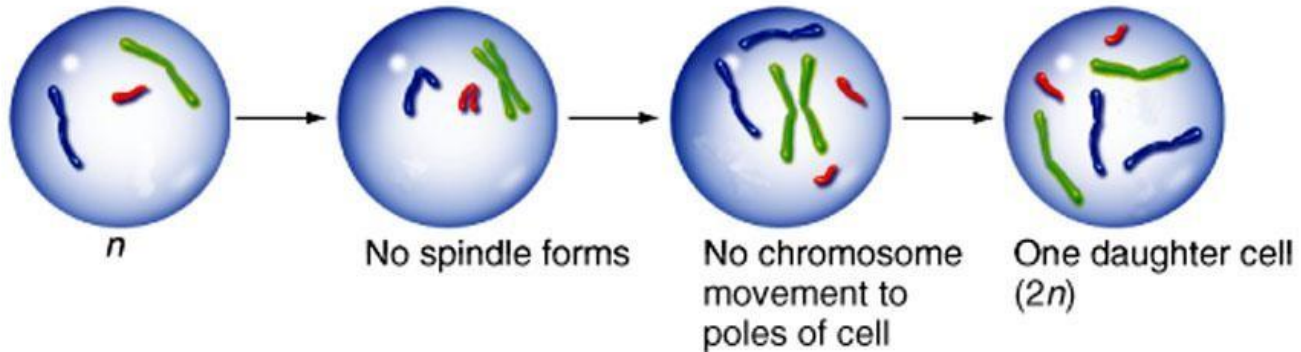


### (c) Using colchicine to double chromosome numbers

Normal mitosis

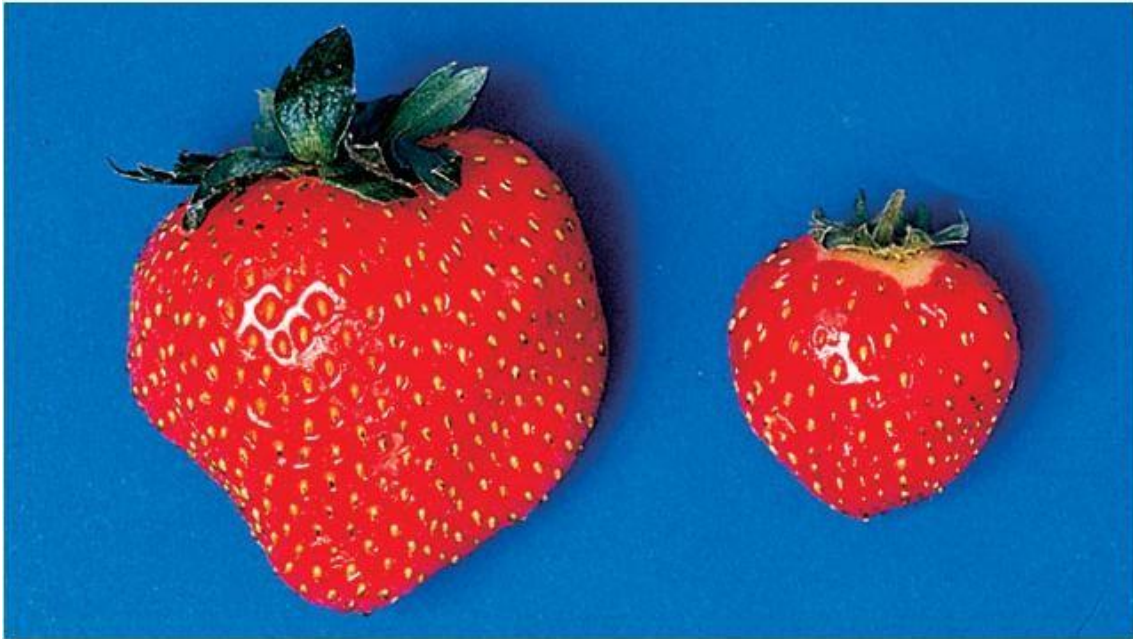


Mitosis with colchicine treatment



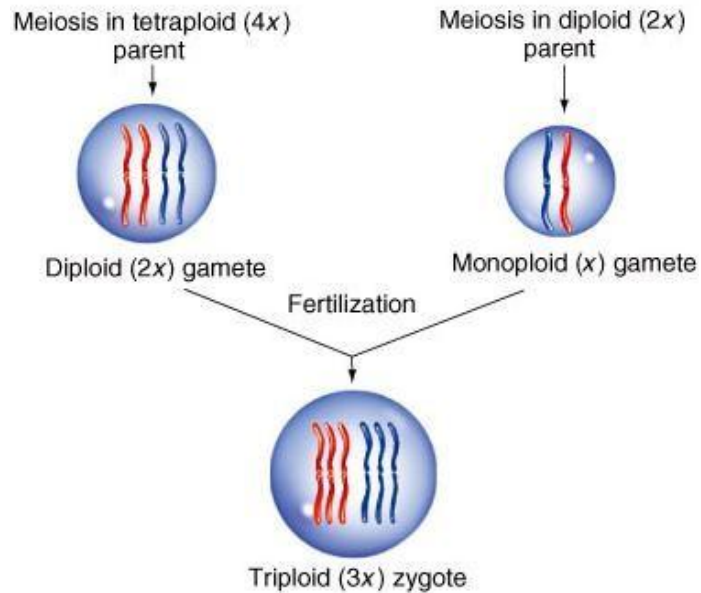


8X

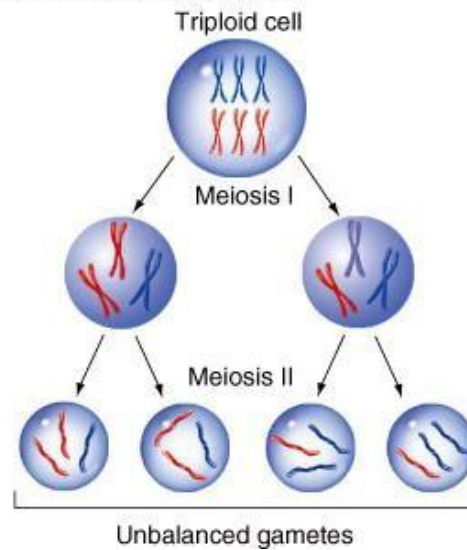


2X

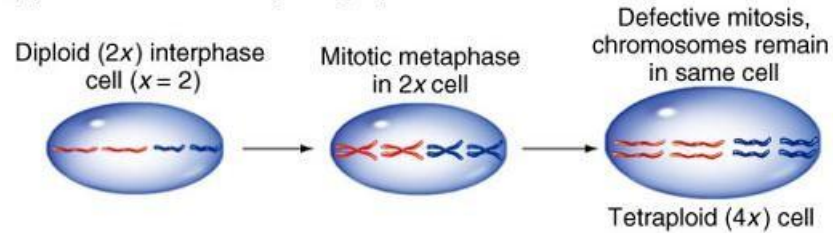
**(a) Formation of a triploid organism**



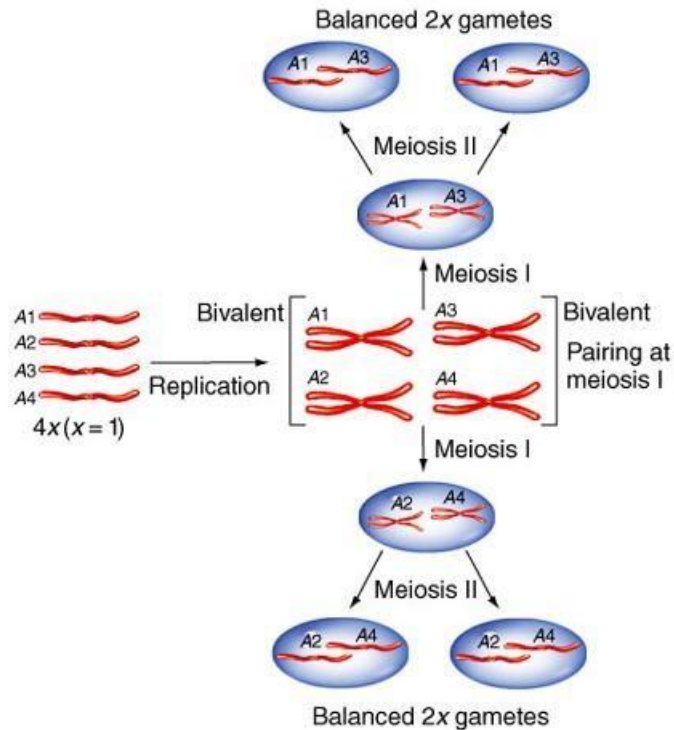
**(b) Meiosis in a triploid organism**



**(a) Generation of tetraploid (4x) cells**



**(b) Pairing of chromosomes as bivalents**

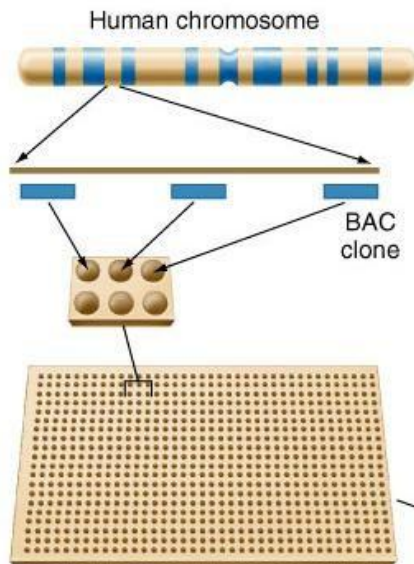


(c) Gametes formed by  $A A a a$  tetraploids

Chromosomes	Pairing	Gametes produced by random spindle attachment
1. $\text{---} \cdot A$ 2. $\text{---} \cdot A$ 3. $\text{---} \cdot a$ 4. $\text{---} \cdot a$	$  \begin{array}{cc}  \uparrow A & \uparrow a \\  \text{1} \text{---} \cdot A & \text{3} \text{---} \cdot a \\  \downarrow A & \downarrow a \\  \text{2} \text{---} \cdot A & \text{4} \text{---} \cdot a  \end{array}  $ or $  \begin{array}{cc}  \uparrow A & \uparrow a \\  \text{1} \text{---} \cdot A & \text{4} \text{---} \cdot a \\  \downarrow A & \downarrow a \\  \text{2} \text{---} \cdot A & \text{3} \text{---} \cdot a  \end{array}  $	$1 + 3 A a$ $1 + 4 A a$ $2 + 4 A a$ or $2 + 3 A a$
	$  \begin{array}{cc}  \uparrow A & \uparrow A \\  \text{1} \text{---} \cdot A & \text{2} \text{---} \cdot A \\  \downarrow a & \downarrow a \\  \text{3} \text{---} \cdot a & \text{4} \text{---} \cdot a  \end{array}  $ or $  \begin{array}{cc}  \uparrow A & \uparrow a \\  \text{1} \text{---} \cdot A & \text{4} \text{---} \cdot a \\  \downarrow a & \downarrow A \\  \text{3} \text{---} \cdot a & \text{2} \text{---} \cdot A  \end{array}  $	$1 + 2 A A$ $1 + 4 A a$ $3 + 4 a a$ or $2 + 3 A a$
	$  \begin{array}{cc}  \uparrow A & \uparrow A \\  \text{1} \text{---} \cdot A & \text{2} \text{---} \cdot A \\  \downarrow a & \downarrow a \\  \text{4} \text{---} \cdot a & \text{3} \text{---} \cdot a  \end{array}  $ or $  \begin{array}{cc}  \uparrow A & \uparrow a \\  \text{1} \text{---} \cdot A & \text{3} \text{---} \cdot a \\  \downarrow a & \downarrow A \\  \text{4} \text{---} \cdot a & \text{2} \text{---} \cdot A  \end{array}  $	$1 + 2 A A$ $1 + 3 A a$ $3 + 4 a a$ or $2 + 4 A a$

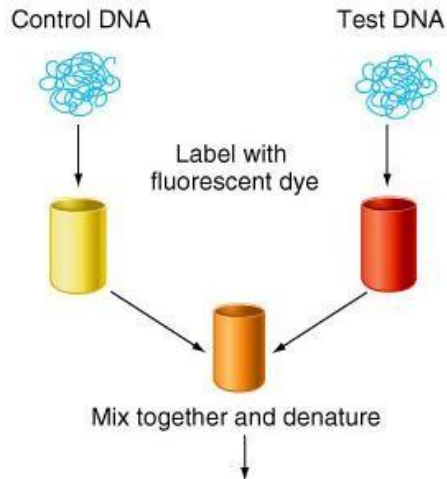
Total:  
 $8A a : 2A A : 2a a = 4A a : 1A A : 1a a$

**(a) Prepare microarray.**



Microarray with ordered series of BAC clones across the entire human genome

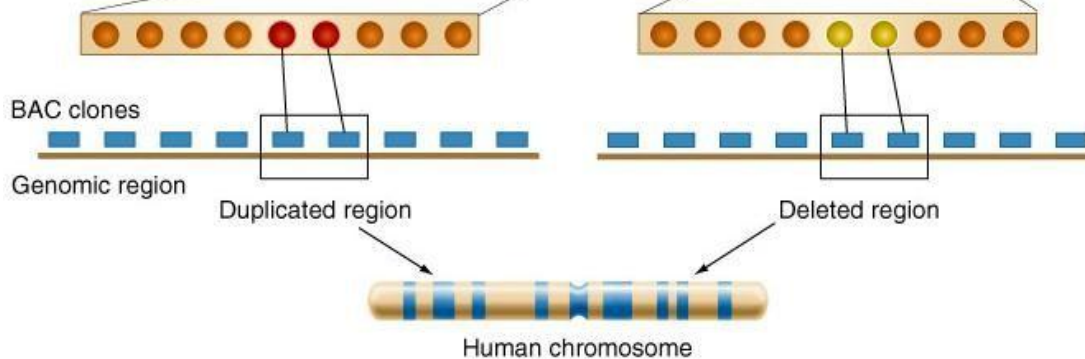
**(b) Prepare genomic DNA samples.**



**(c) Incubate microarray with combined samples.**



**(d) Examples of results with duplicated or deleted genomic regions.**

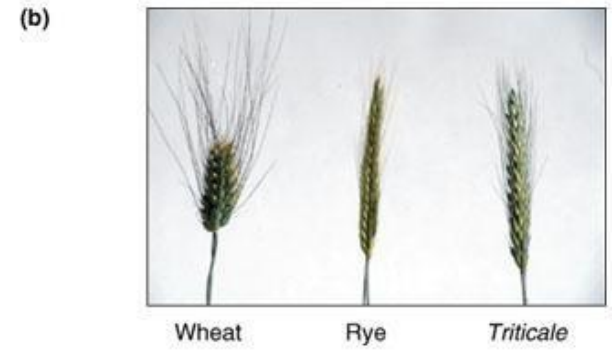
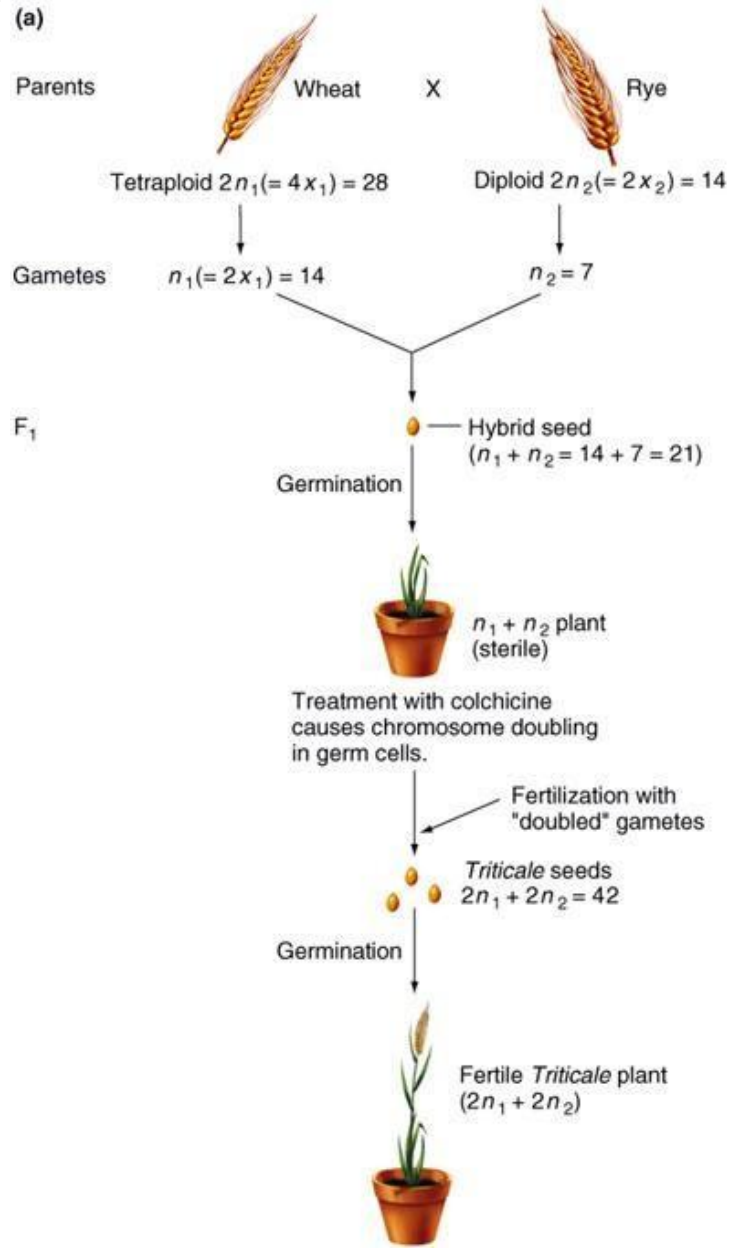


**TABLE 13.2** Aneuploidy in the Human Population

Chromosomes	Syndrome	Frequency at Birth
Autosomes		
Trisomic 21	Down	1/700
Trisomic 13	Patau	1/5,000
Trisomic 18	Edwards	1/10,000
Sex chromosomes, females		
XO, monosomic	Turner	1/5,000
XXX, trisomic	X inactivation Limited fertility	1/700
XXXX, tetrasomic		
XXXXX, pentasomic		
Sex chromosomes, males		
YYY, trisomic	Normal	1/10,000
XXYY, tetrasomic	Klinefelter	1/500
XXXY, tetrasomic		
XXXXY, pentasomic		
XXXXXY, hexasomic		

About 0.4% of all babies born have a detectable chromosomal abnormality that generates a detrimental phenotype.

Fig. 13.34



# “B” Kromozomları

“Accessory”, “supernumerary”, “additional” kromozomlar, “selfish genetic elements”

Normal kromozom yapısı “A”

## “B” kromozomları

- küçük
- Bitkiler ve hayvanlar aleminde çeşitli sayılarda
  - 1300 den fazla bitkide
  - 500 hayvanda
  - çeşitli mantarlarda
- genetik olarak inaktif, gelişim için elzem değil, çok az bazen de hiç fenotipik
- Yaşamsal değil, bir tür içerisinde bazı bireylerde oluşur
- Gametogenez sırasında veya sonrasında oluşur



Bazı organizmalarda:

-heterokromatik görünümde (deeply staining)

-light staining with telomeric and centromeric region of heterochromatin

Çavdar, mısır; akrosentrik

Parazitik eşek arısında; submetasentrik

*Allium cernuum*; metasentrik

Her hücrede bulunmaz

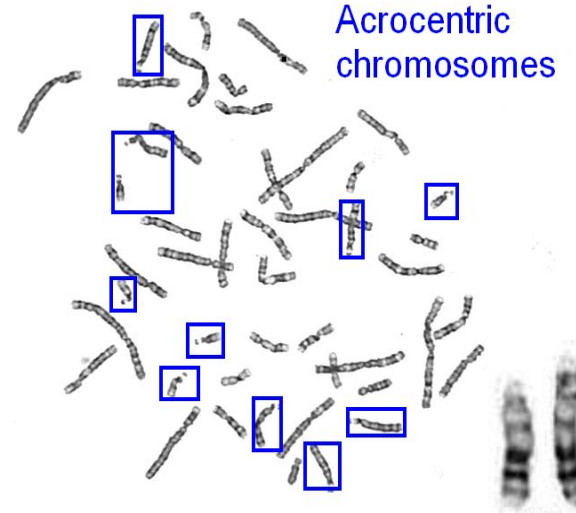
Mitotik aktif somatik ve germ hücrelerinde rastlanır

*Triticum tripsacoides* : B kromozomu kök ucu hücrelerinde yok, fakat present polen anahücrelerinde var

*Crepis capillaris* :

2 B kök ucu vegetatif hücrelerde

4 B Germ hücrelerinde



-işlevsel rRNA genleri(NOR lokus)taşırılar

-A kromozomlarının eşleşmesini

-Kiasma oluşumunu

-genetik rekombinasyonu etkilerler

**- Gen anlatımı DNA metilasyonu ile ilişkilidir.**