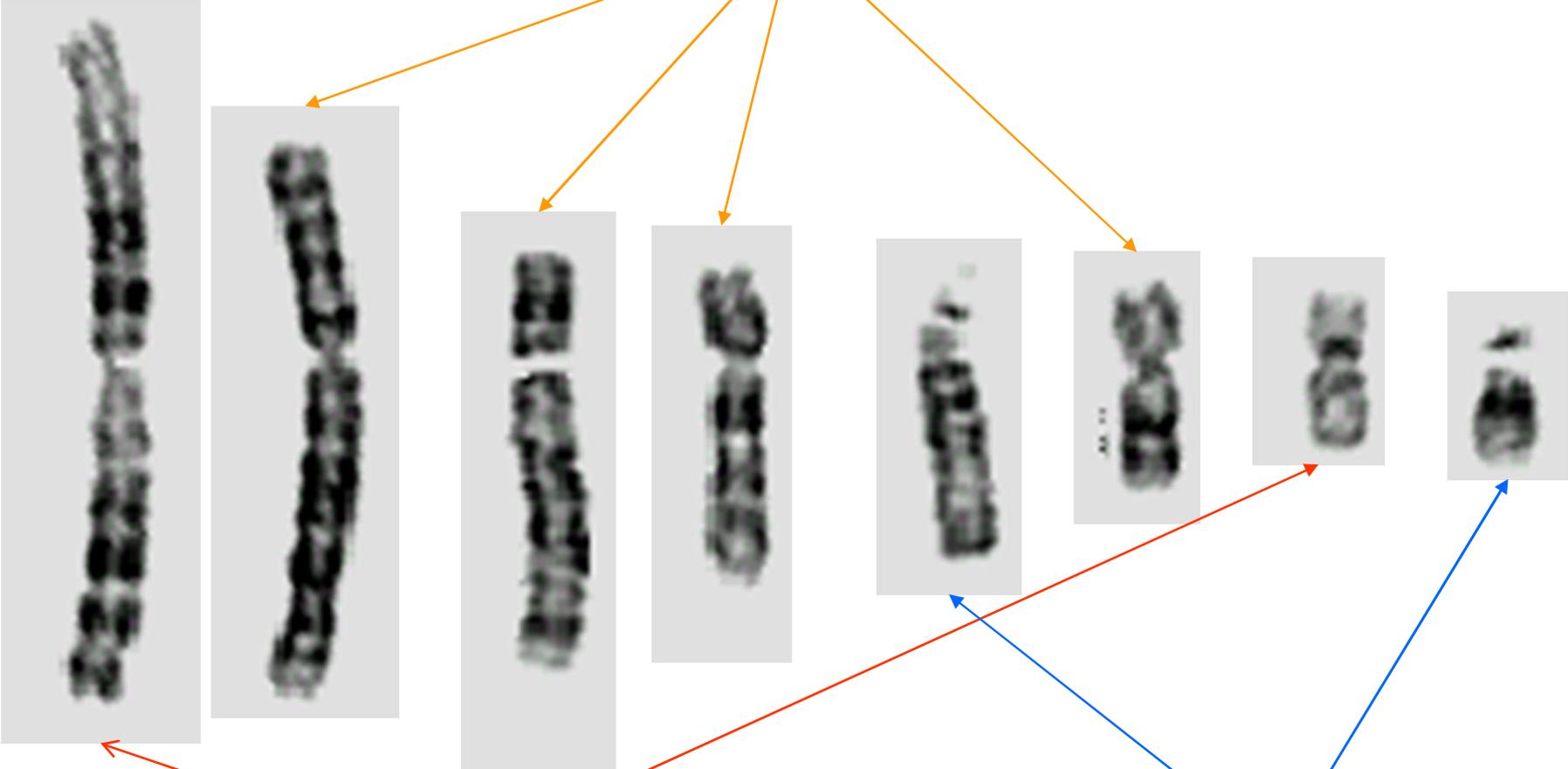


# **Alterazioni di struttura dei cromosomi**

*Submetacentric*

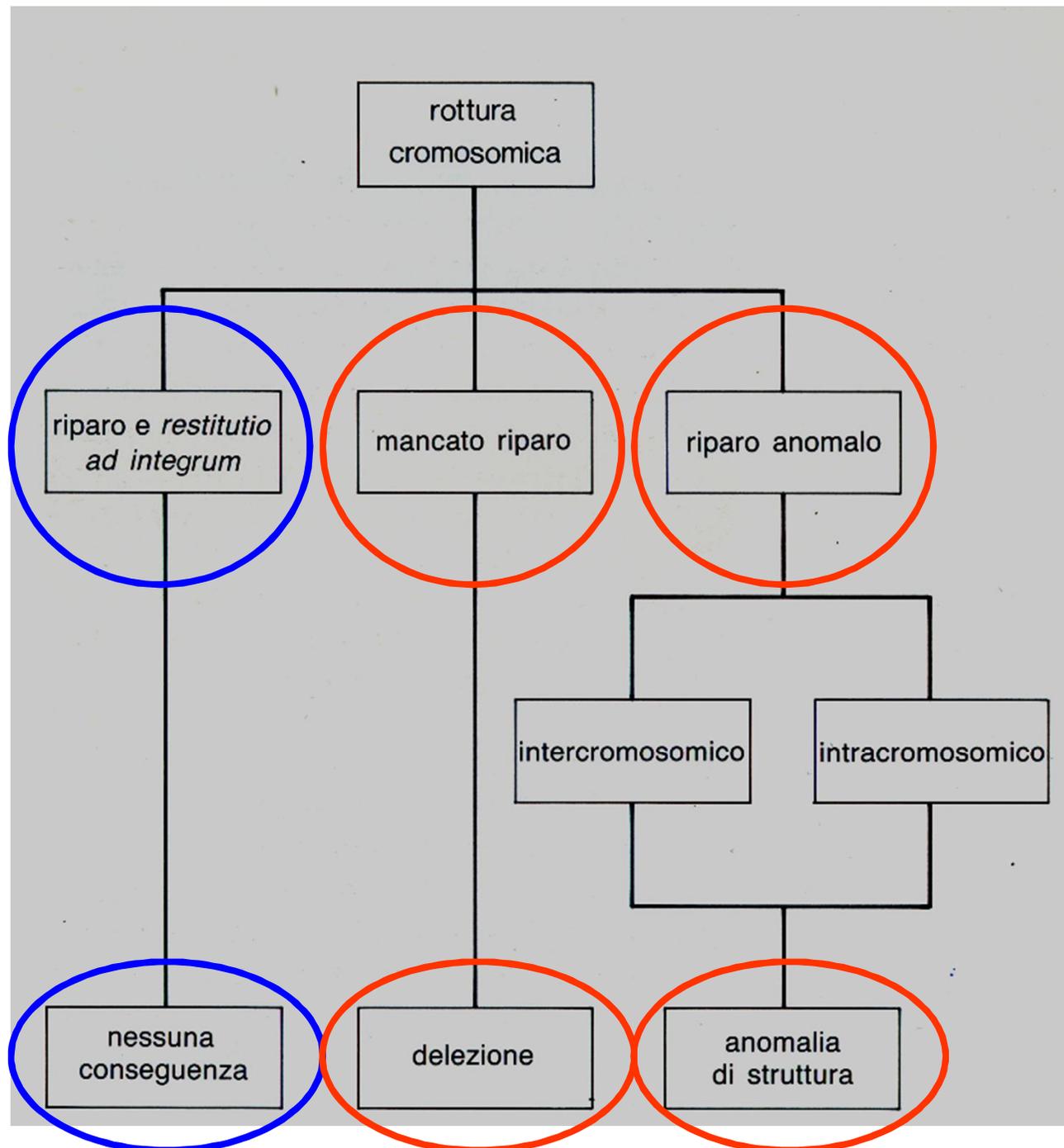


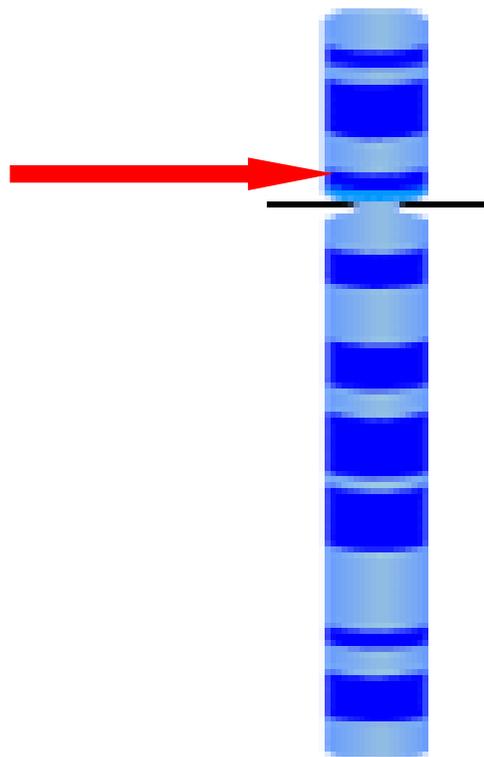
*Metacentric*

*Acrocentric*

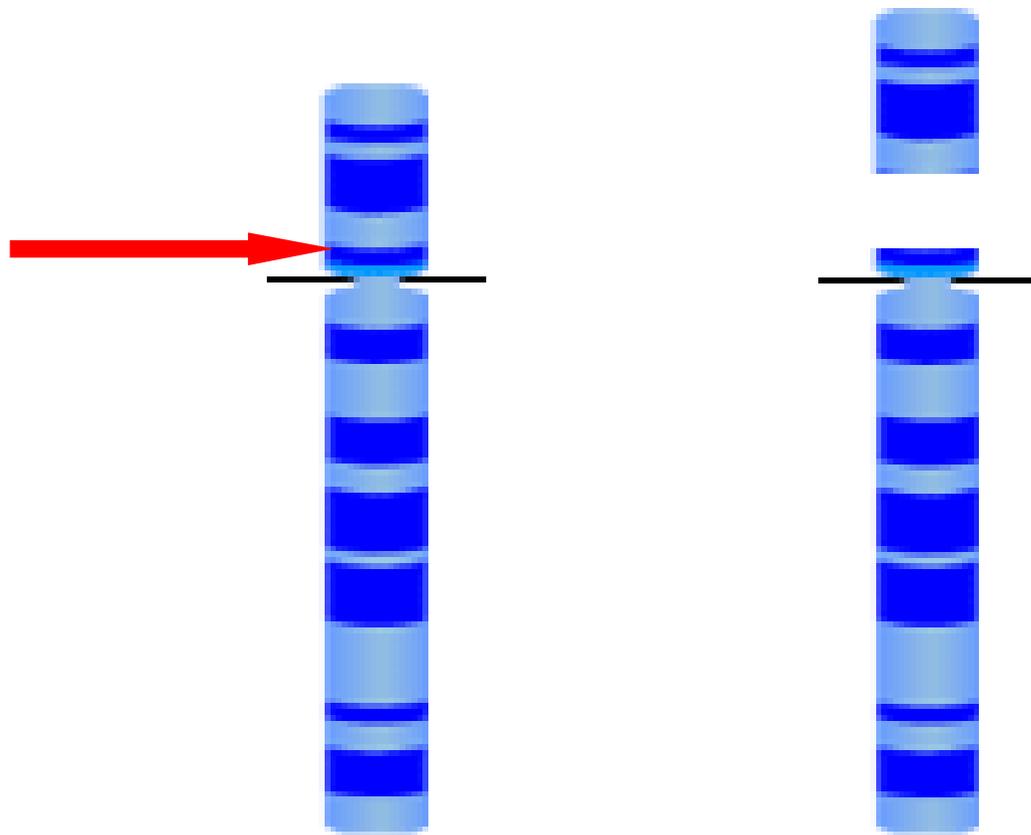
# **Alterazioni strutturali dei cromosomi**

**sono il risultato di rotture  
cromosomiche**

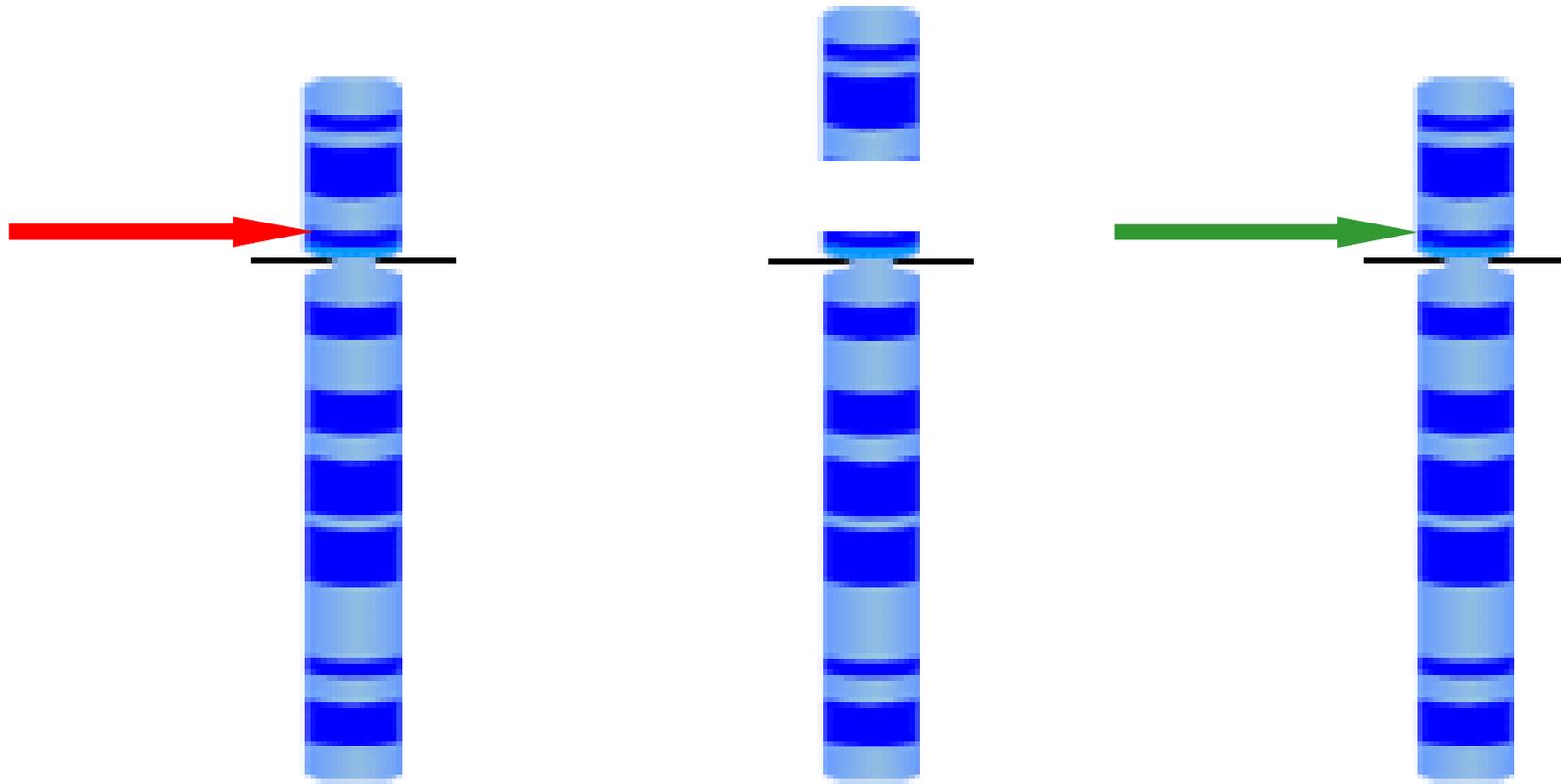




**Cromosoma integro**  
**Si verifica una rottura**



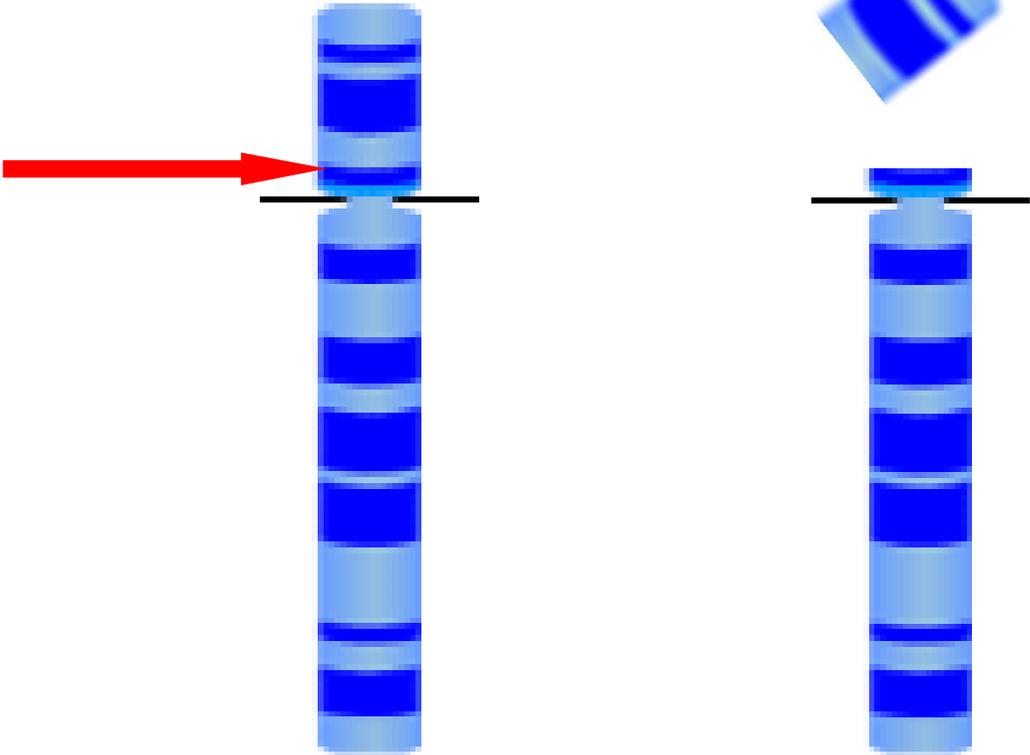
**Cromosoma integro**  
**Si verifica una rottura**



**Cromosoma integro**  
**Si verifica una rottura**

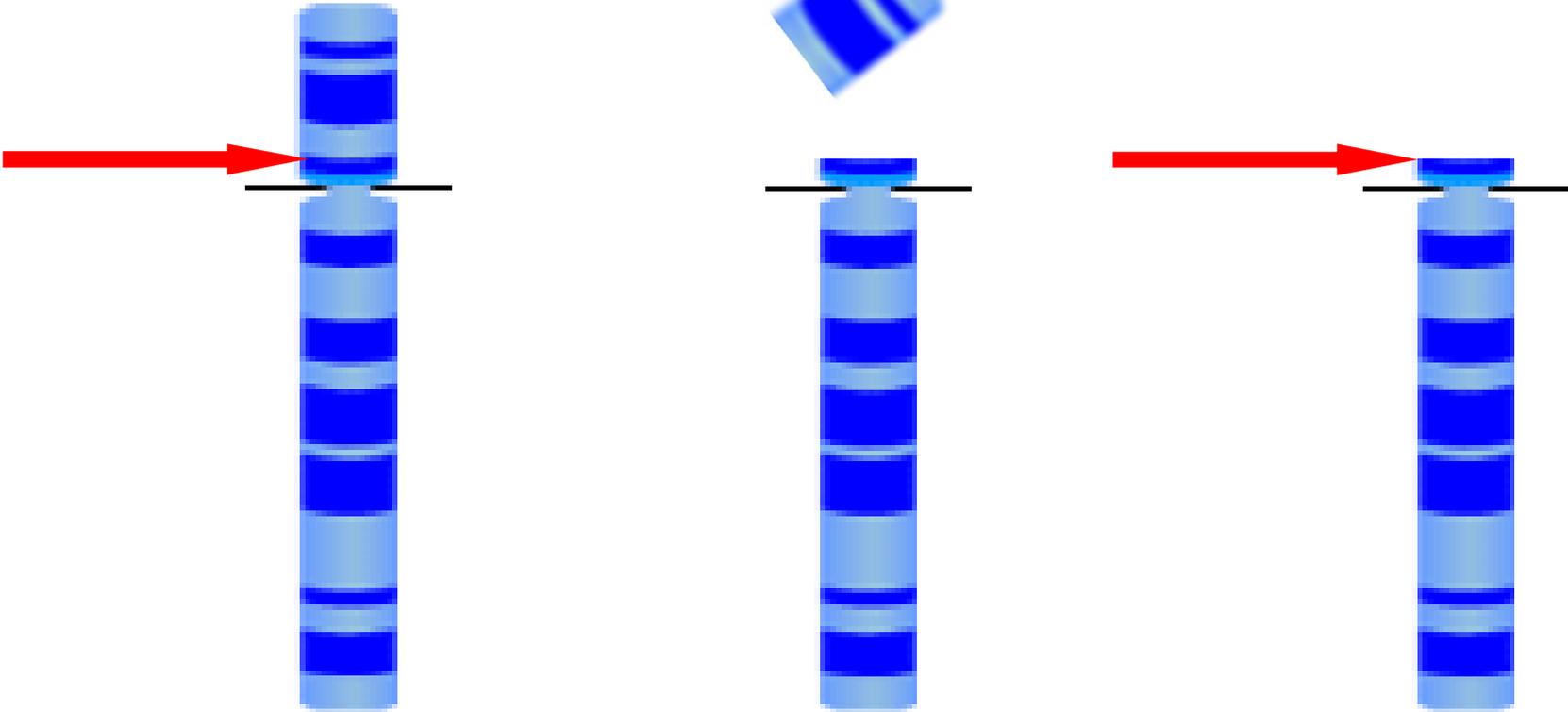
**Riparo e**  
**restitutio ad integrum**

**Cromosoma acentrico: si perde**



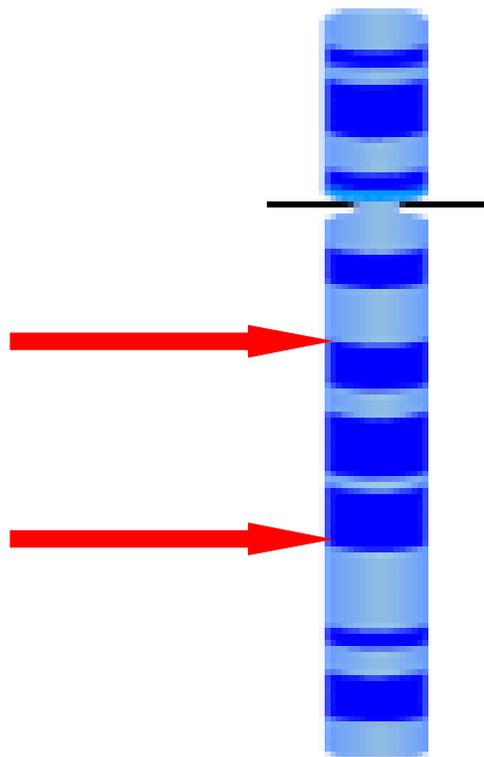
**Cromosoma integro  
Si verifica una rottura**

**Cromosoma acentrico: si perde**

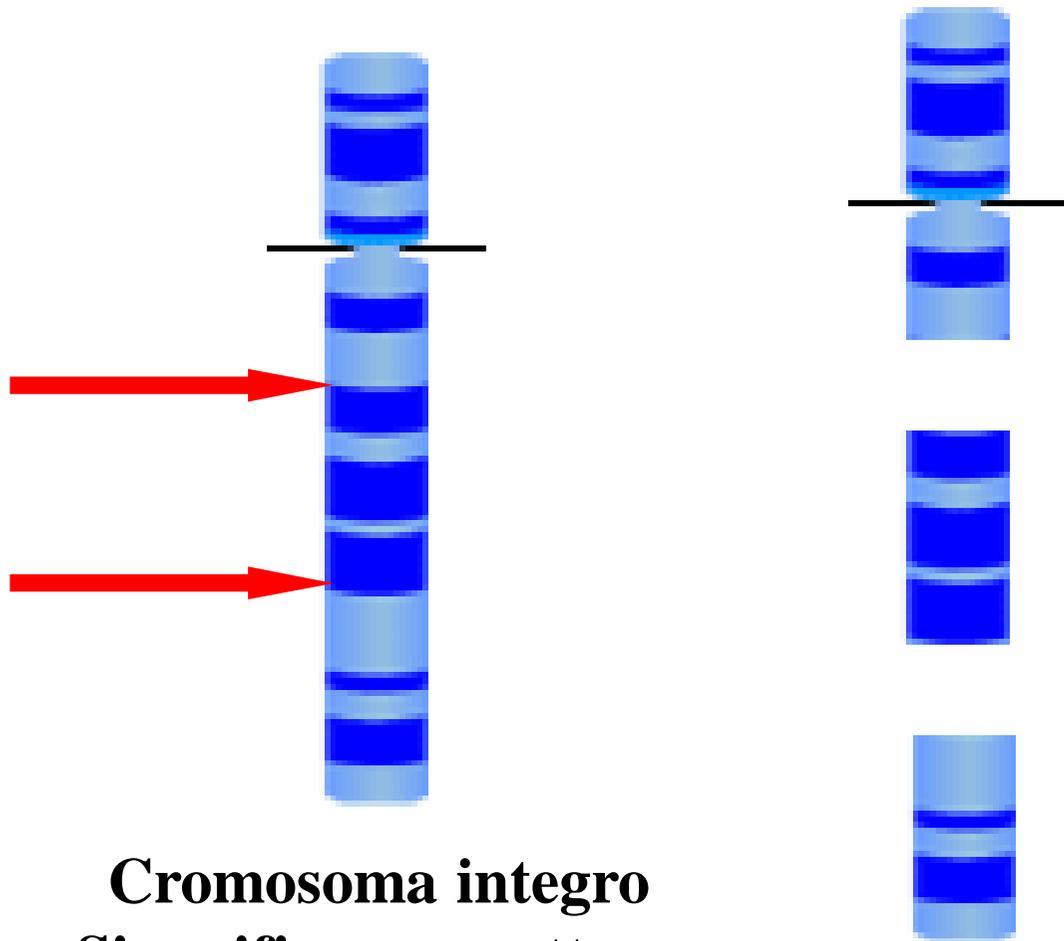


**Cromosoma integro  
Si verifica una rottura**

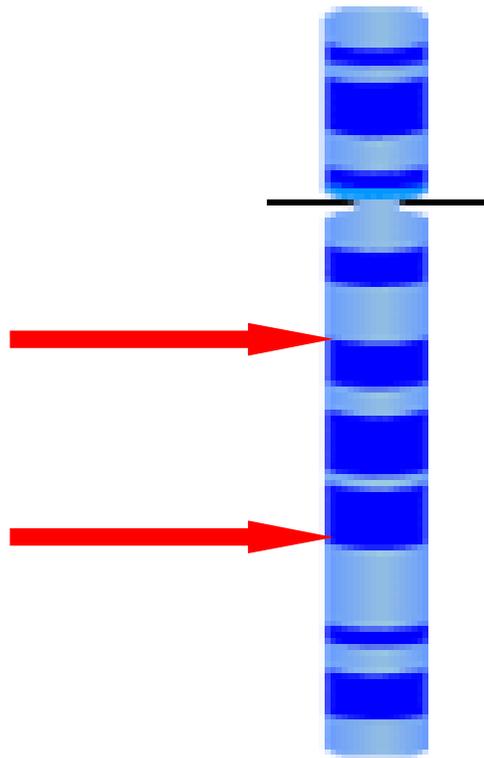
**Cromosoma  
deleto**



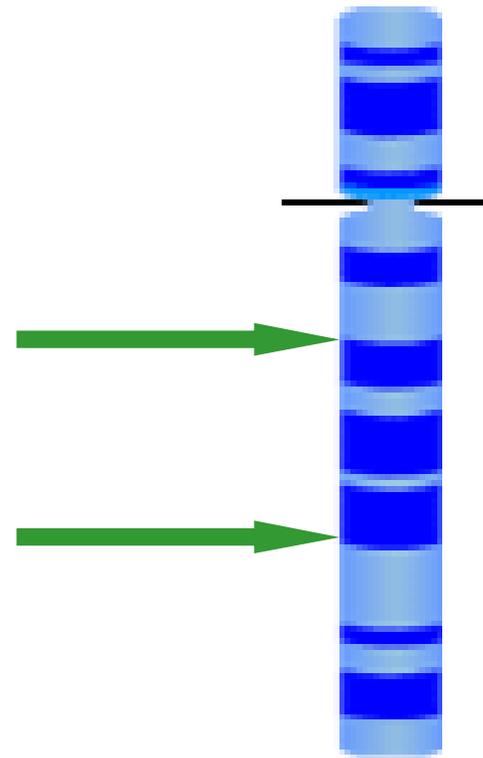
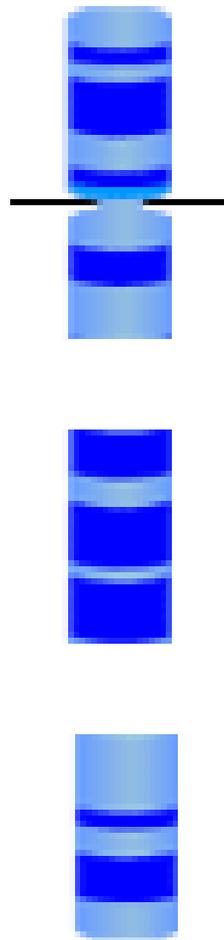
**Cromosoma integro**  
**Si verifica una rottura**



**Cromosoma integro**  
**Si verifica una rottura**

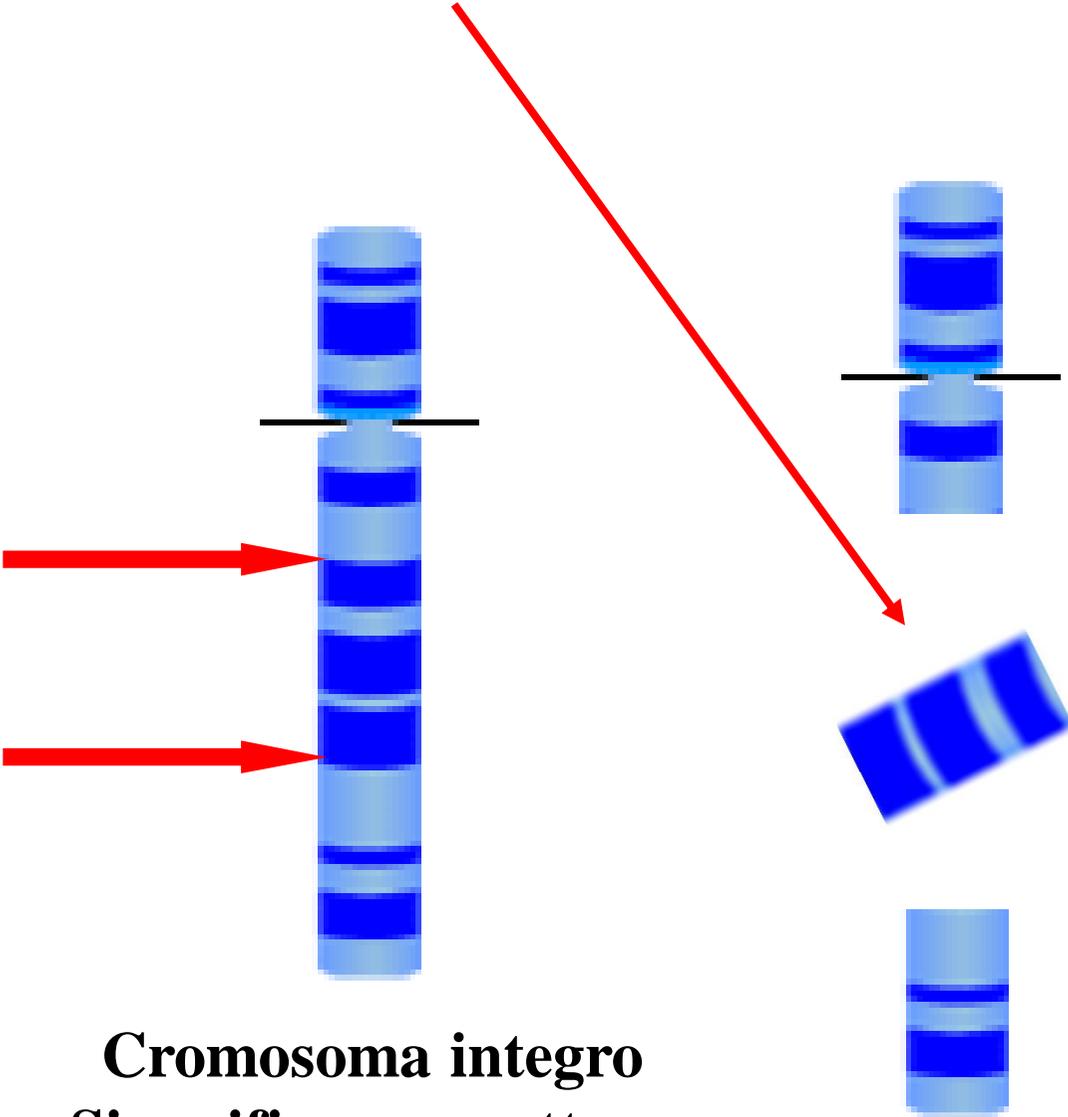


**Cromosoma integro**  
**Si verifica una rottura**



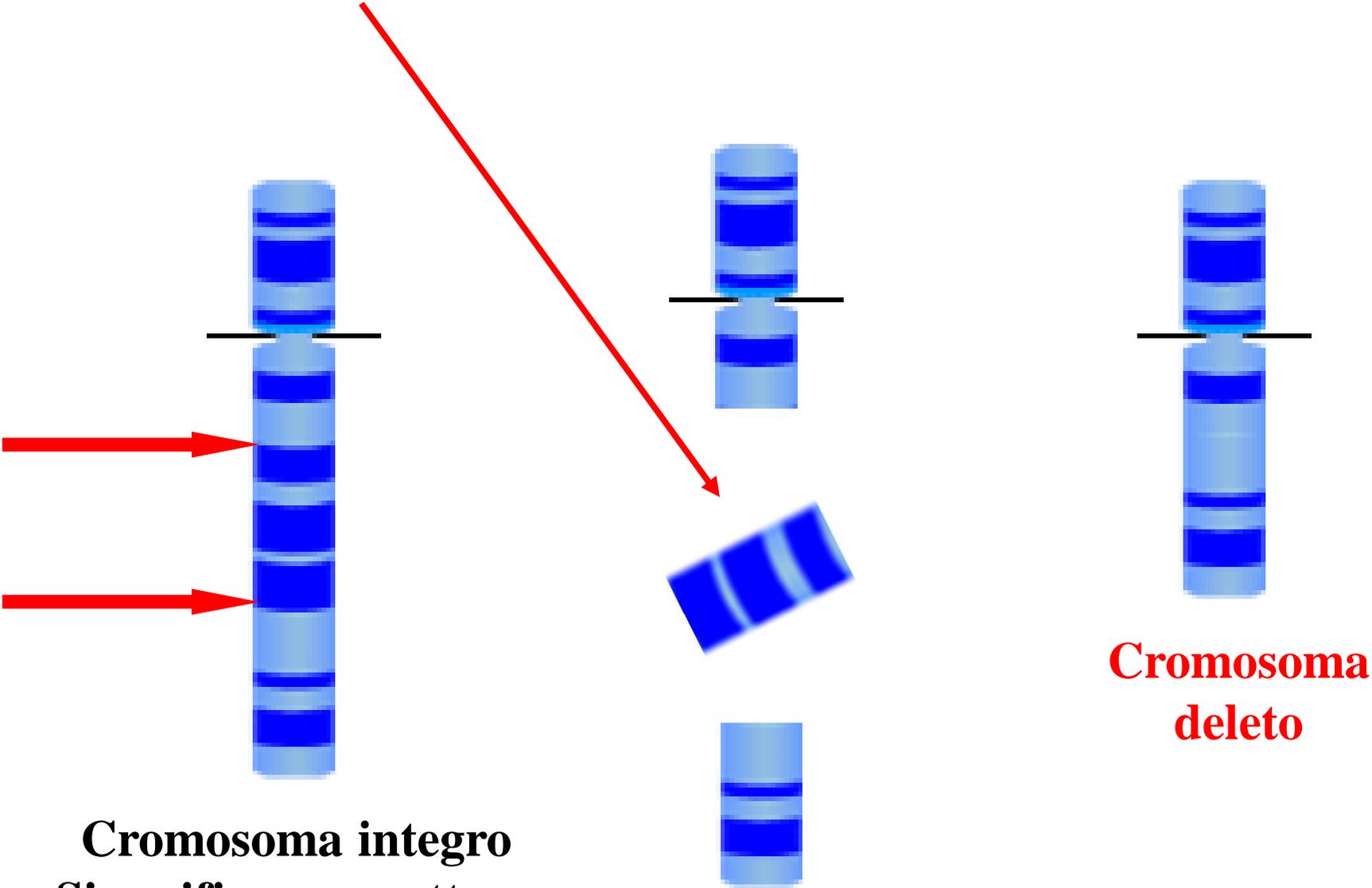
**Riparo e**  
**restitutio ad integrum**

**Cromosoma acentrico: si perde**



**Cromosoma integro  
Si verifica una rottura**

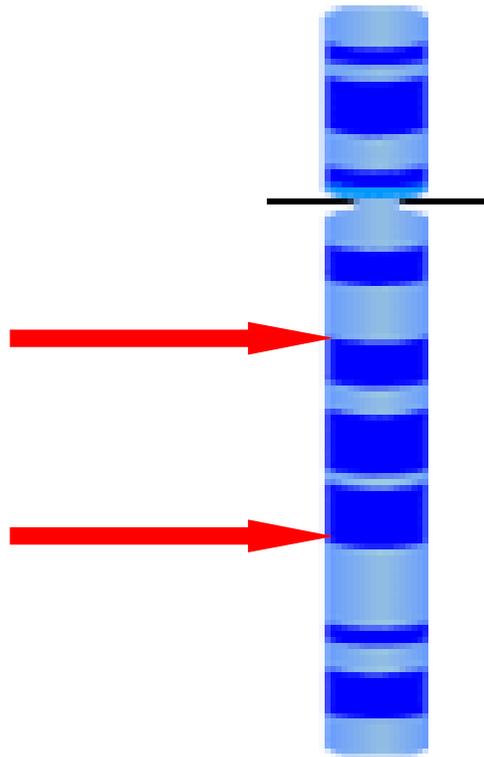
**Cromosoma acentrico: si perde**



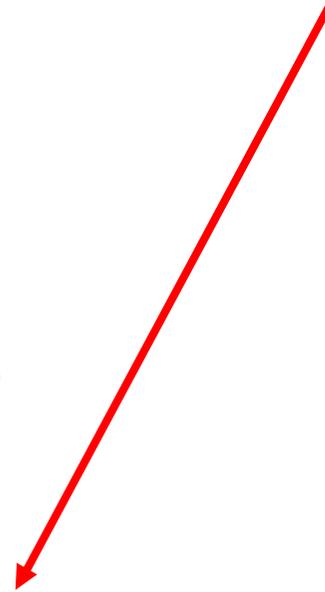
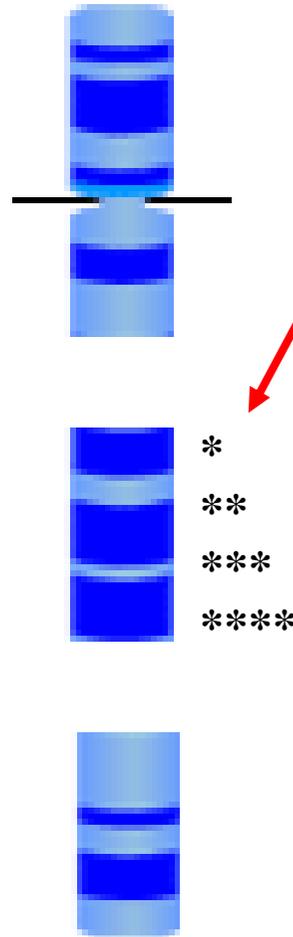
**Cromosoma integro  
Si verifica una rottura**

**Cromosoma  
deleteo**

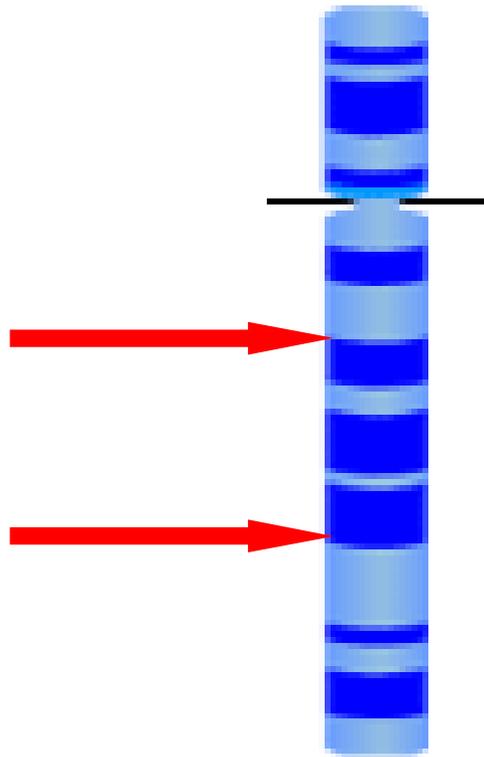
**Cromosoma acentrico: RUOTA di 180°**



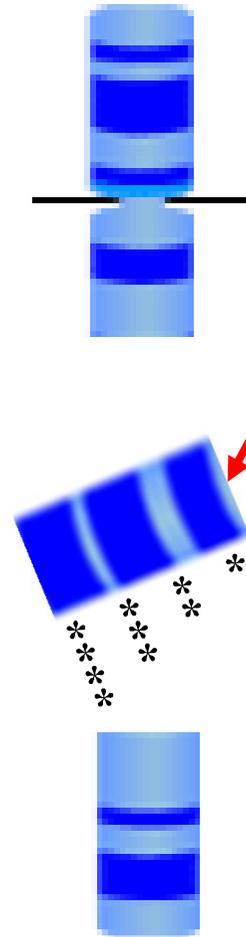
**Cromosoma integro  
Si verifica una rottura**

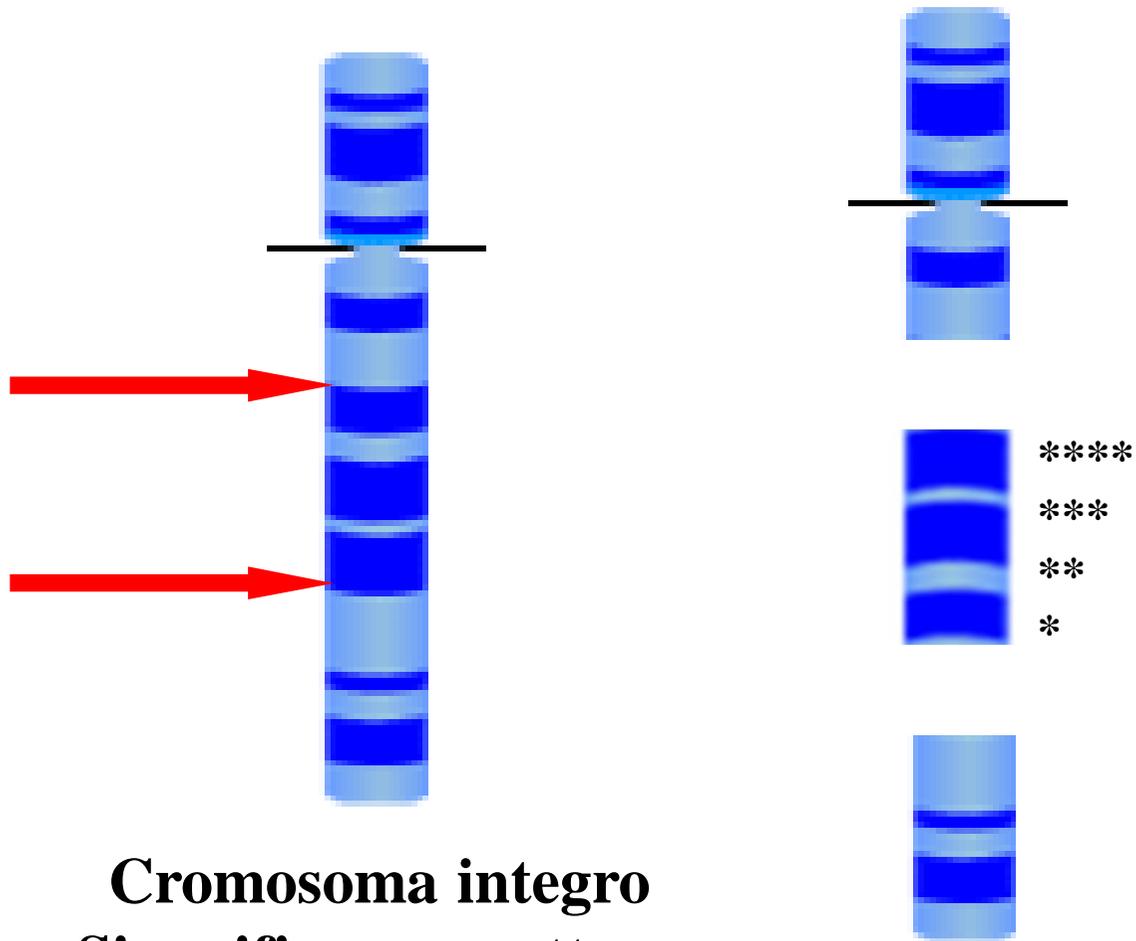


**Cromosoma acentrico: RUOTA di 180°**

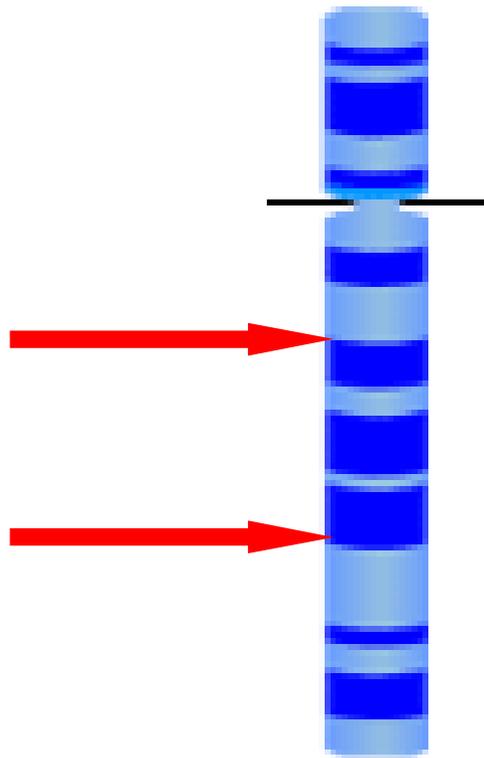


**Cromosoma integro  
Si verifica una rottura**

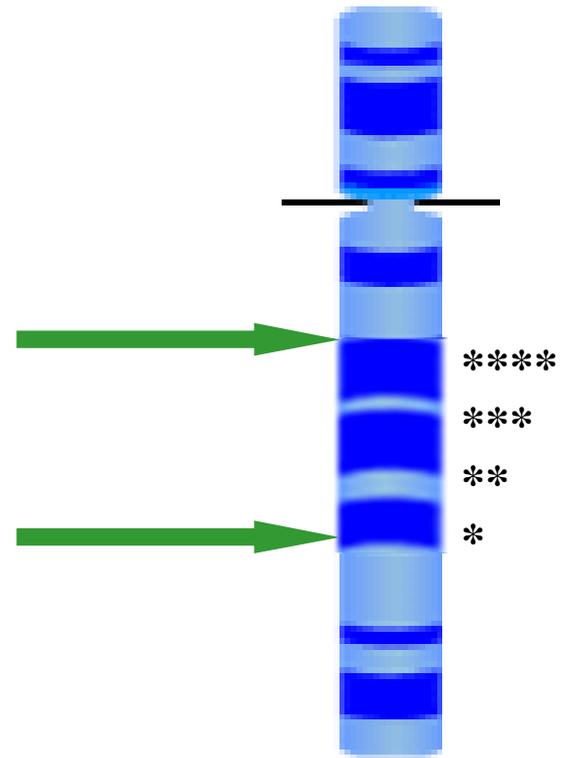
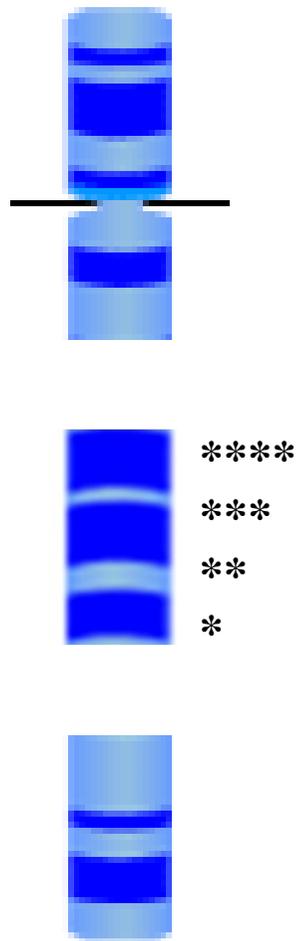




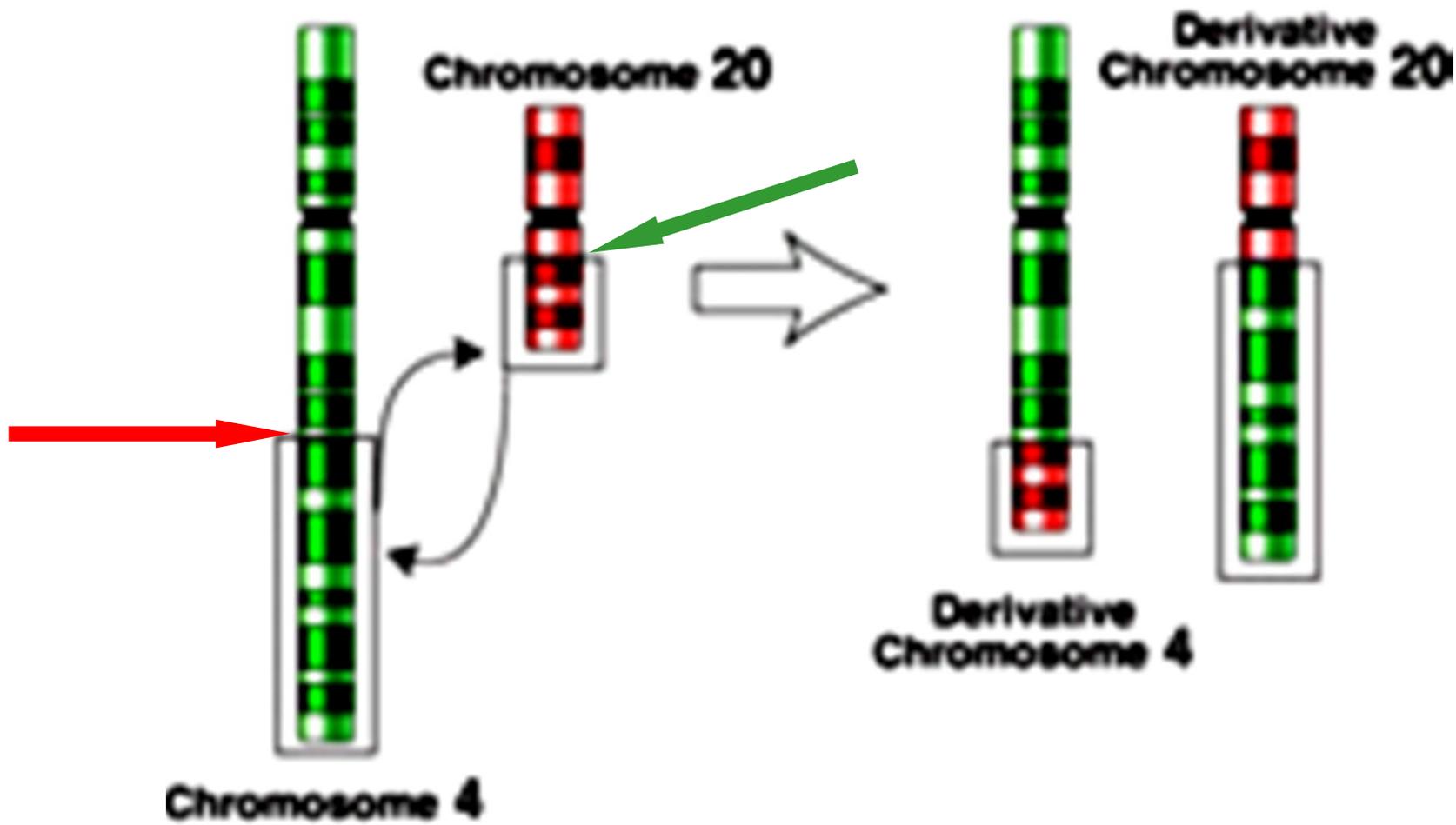
**Cromosoma integro**  
**Si verifica una rottura**

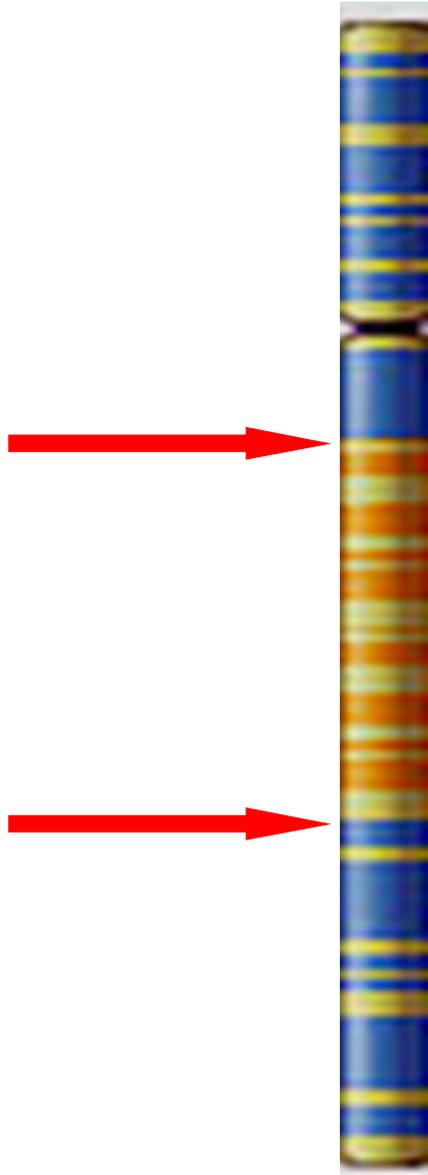
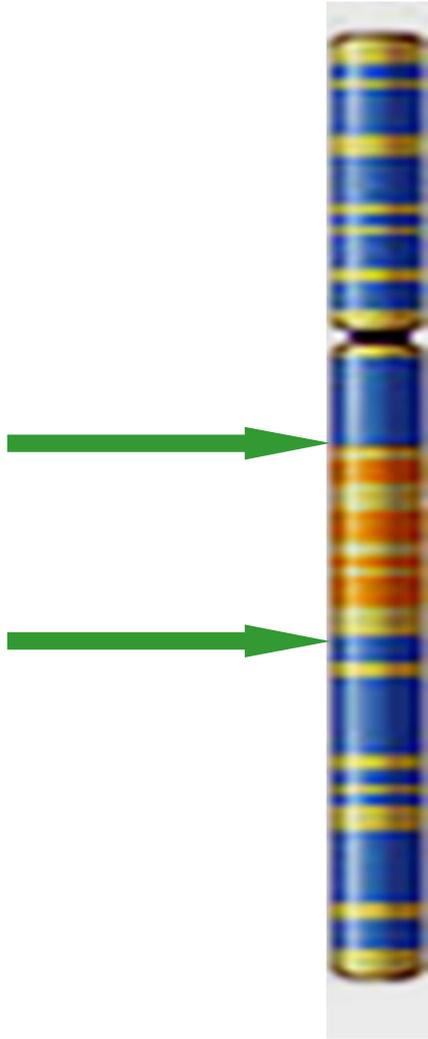


**Cromosoma integro  
Si verifica una rottura**



**Cromosoma  
Con Inversione**





**Cromosoma con duplicazione  
di un segmento**

# Anomalie cromosomiche di struttura

**Traslocazioni**

*Bilanciate*

**Inversioni**

**Delezioni**

*Sbilanciate*

**Duplicazioni**

# Anomalie cromosomiche di struttura

## Bilanciate

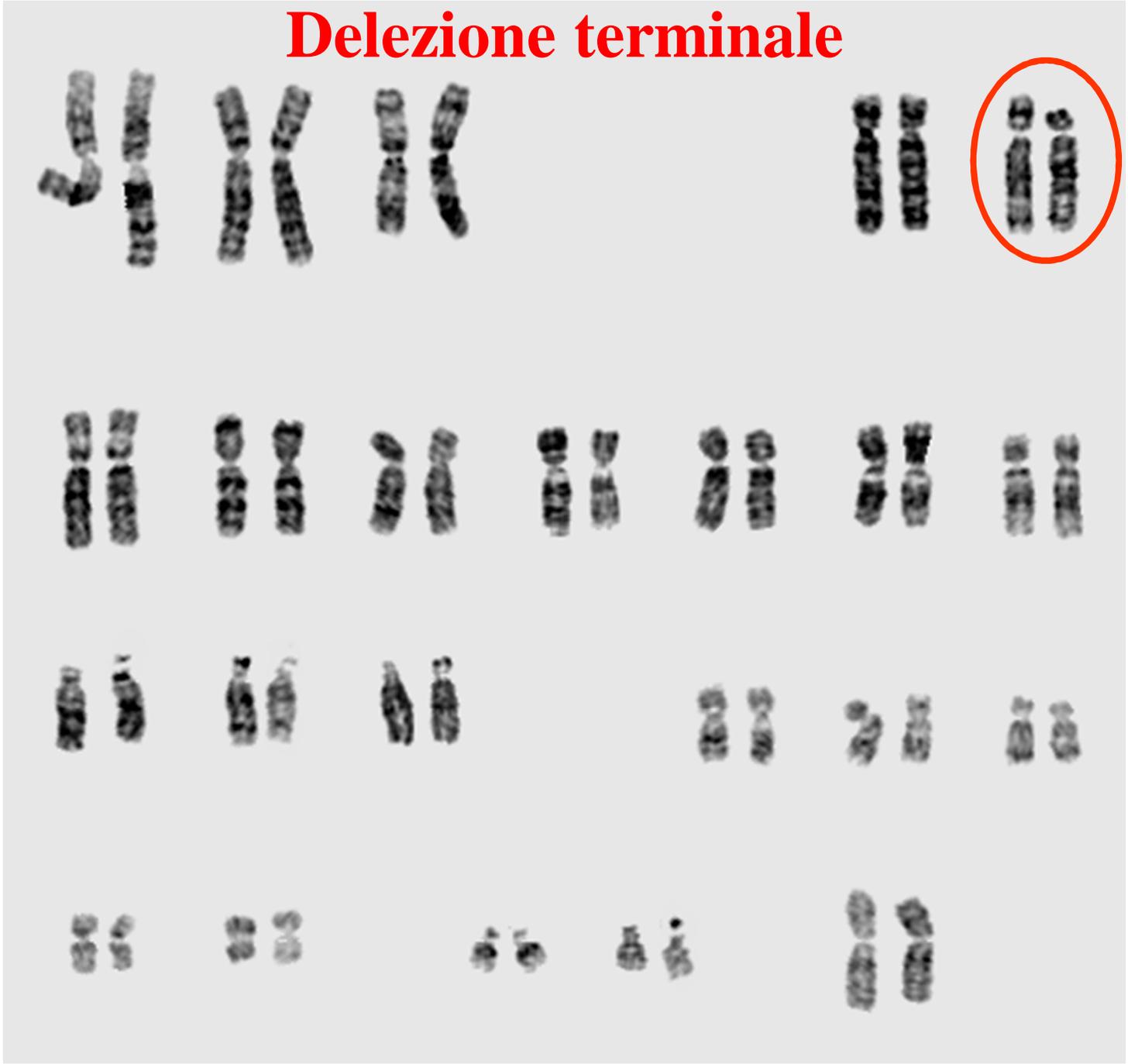
*(nella maggioranza dei casi)*

**non sono correlate ad un fenotipo anomalo**

## Sbilanciate

**sono correlate ad un fenotipo anomalo**  
**(malformazioni e/o ritardo mentale)**

# Delezione terminale



❌ Impossibile visualizzare l'immagine.



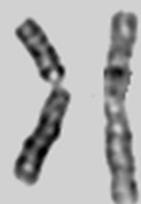
# Principali sindromi da microdelezione

SINDROME	LOCALIZZAZIONE CROMOSOMICA	SOGGETTI CON MICRODELEZIONE
Prader Willi/Angelman	15q11.13	70%
Williams	7q11.23	90%
DiGeorge/Velocardiofaciale	22q11.2	75%
Smith-Magenis	17p11.2	95%
Miller-Dieker	17p13.3	90%

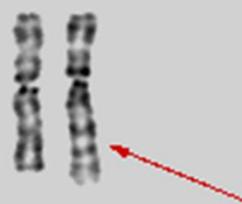
# Traslocazioni



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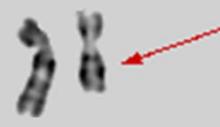
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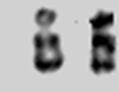
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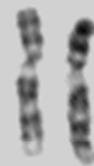
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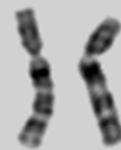
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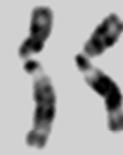
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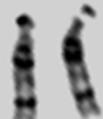
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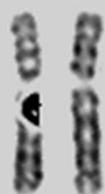
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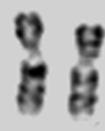
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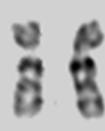
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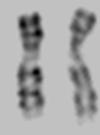
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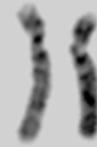
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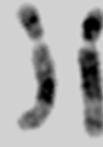
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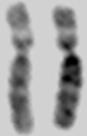
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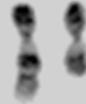
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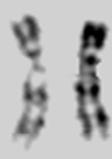
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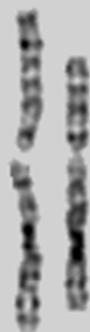
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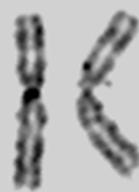
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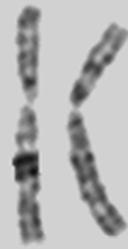
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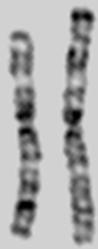
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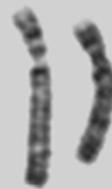
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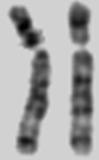
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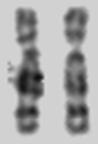
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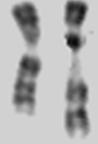
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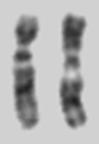
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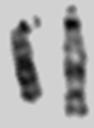
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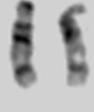
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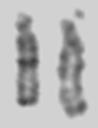
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# Traslocazioni robertsoniane



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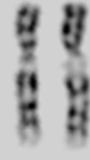
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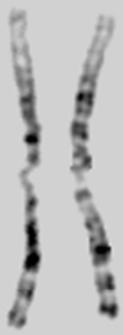
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# Inversione paracentrica



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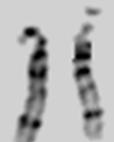
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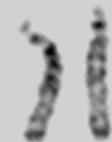
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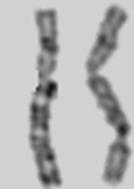
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**Inv. Par.**



**Normale**

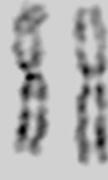
# Inversione pericentrica



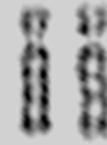
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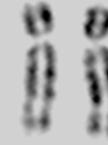
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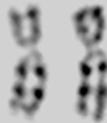
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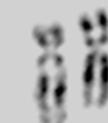
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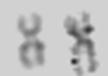
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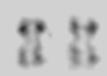
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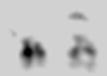
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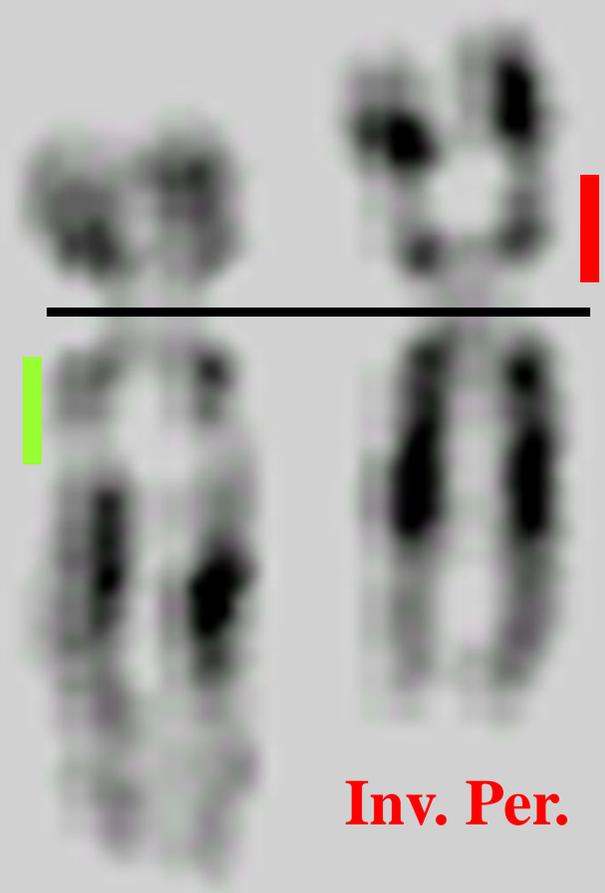
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Normale

Inv. Per.



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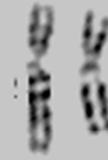
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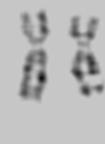
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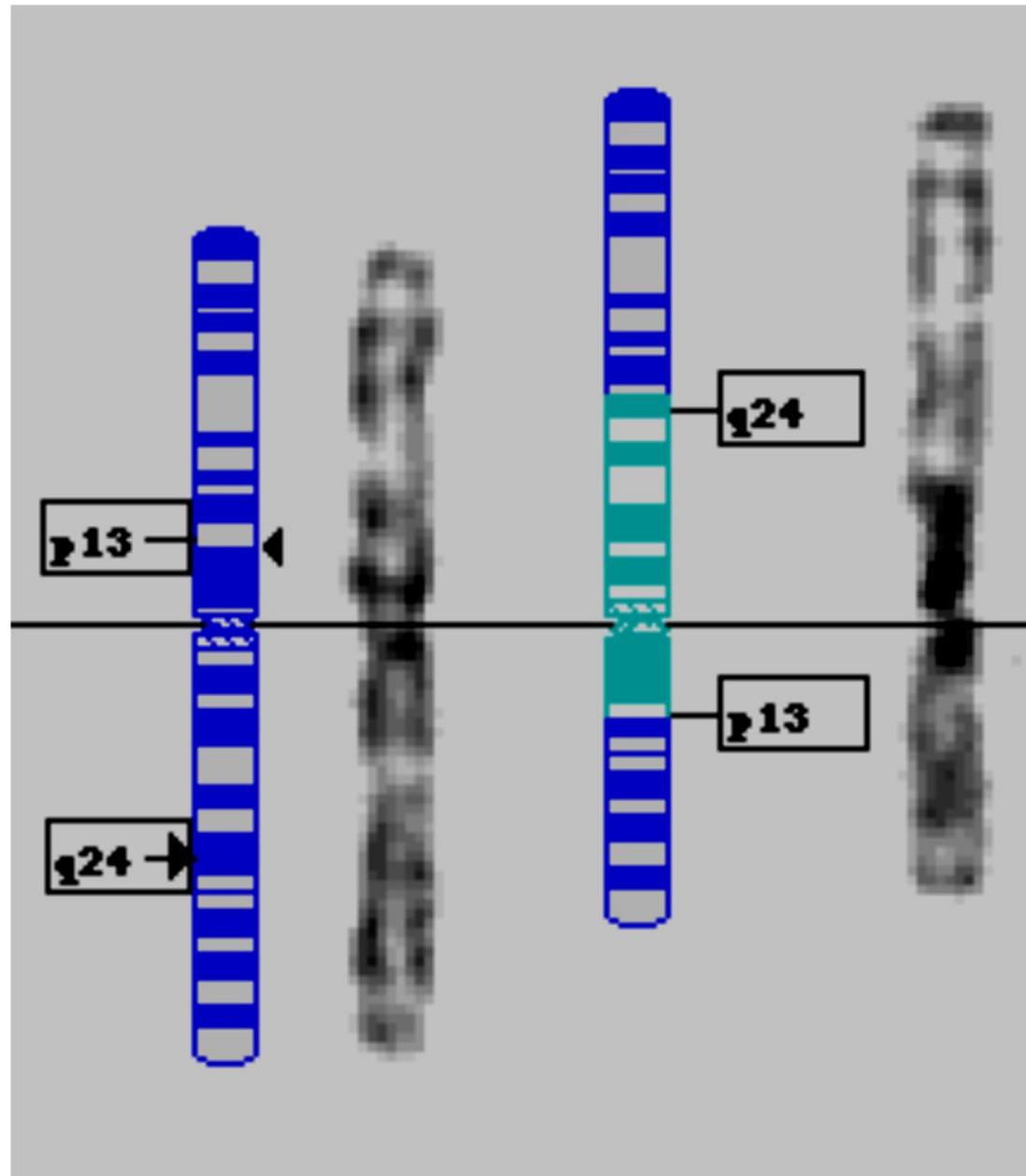
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# Inversione pericentrica eterocromatina



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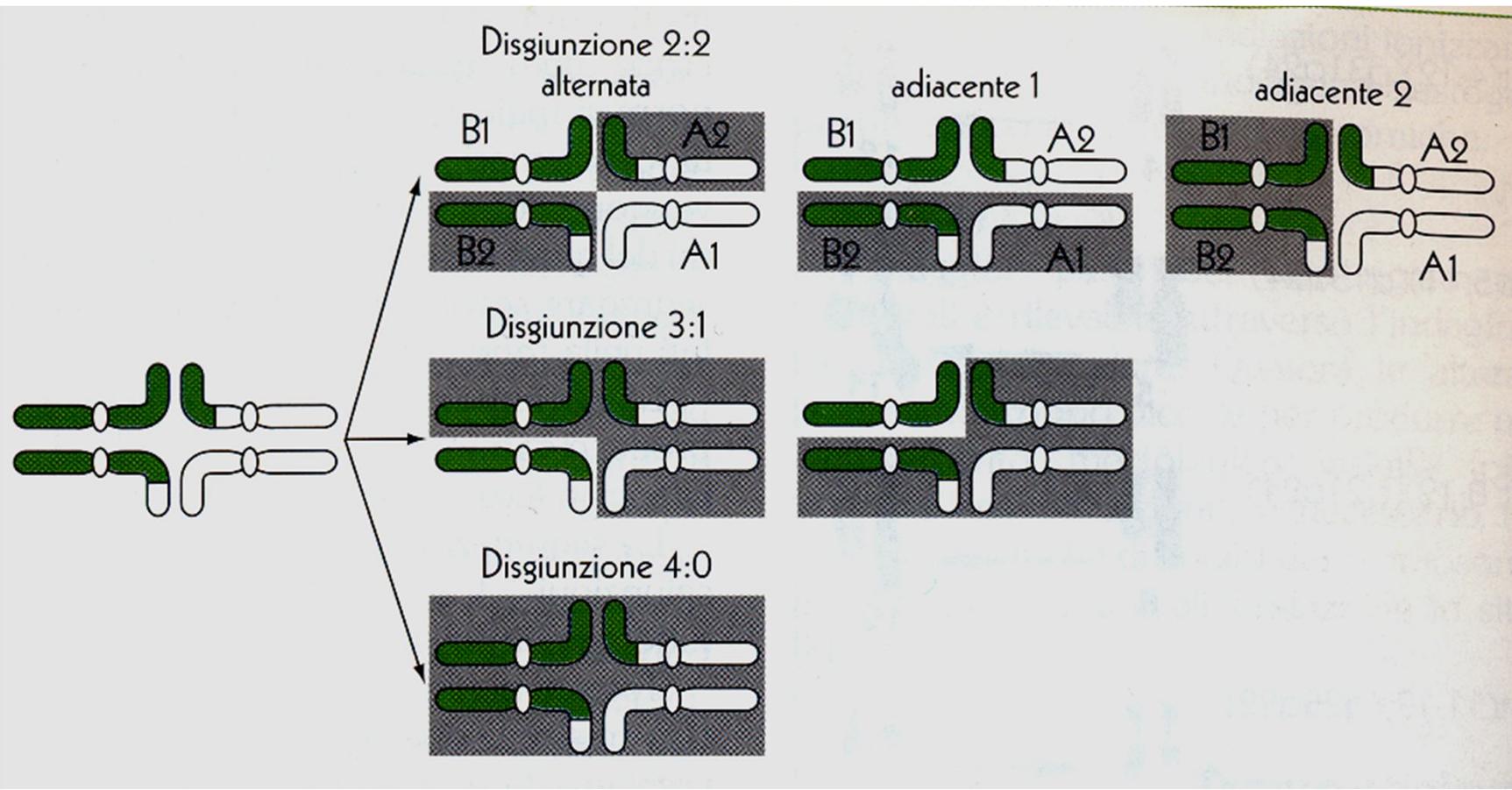
Y

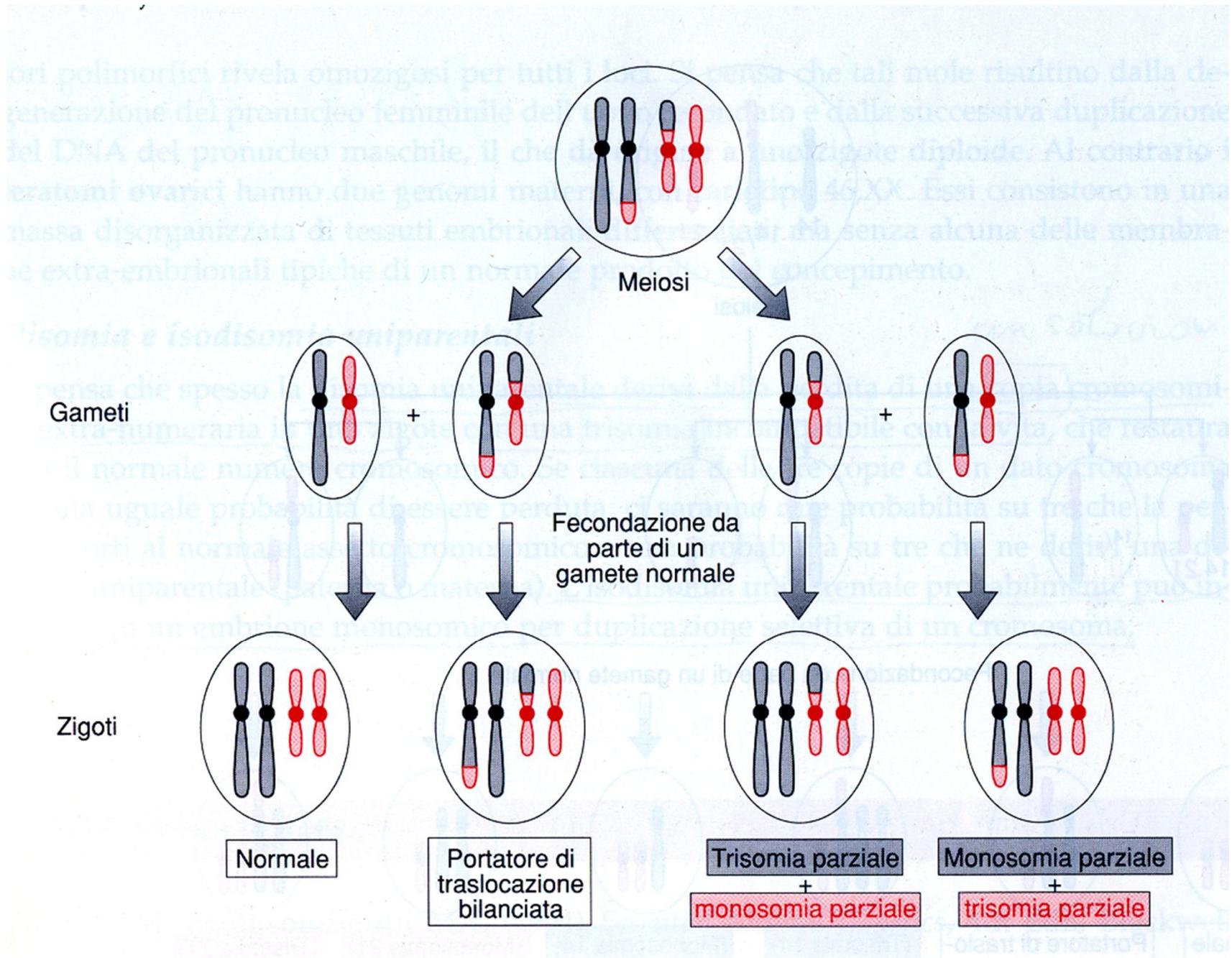


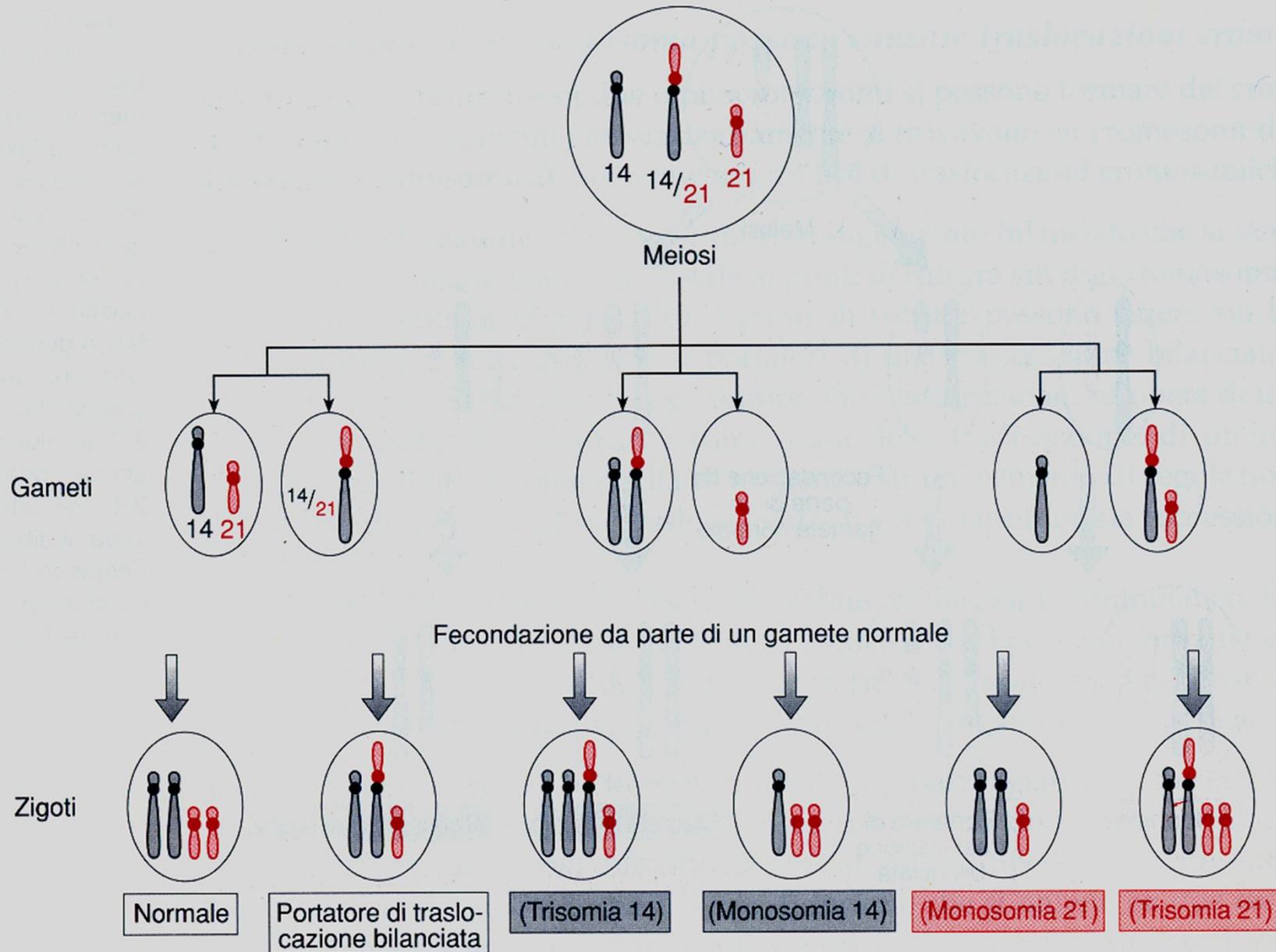
# **Anomalie cromosomiche bilanciate?**

**Il problema non si pone per il  
soggetto portatore,**

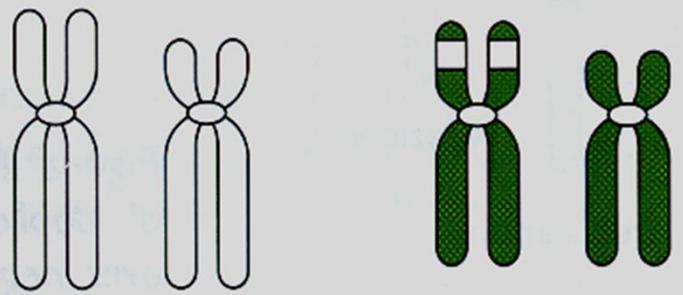
**ma riguarda la sua discendenza...**



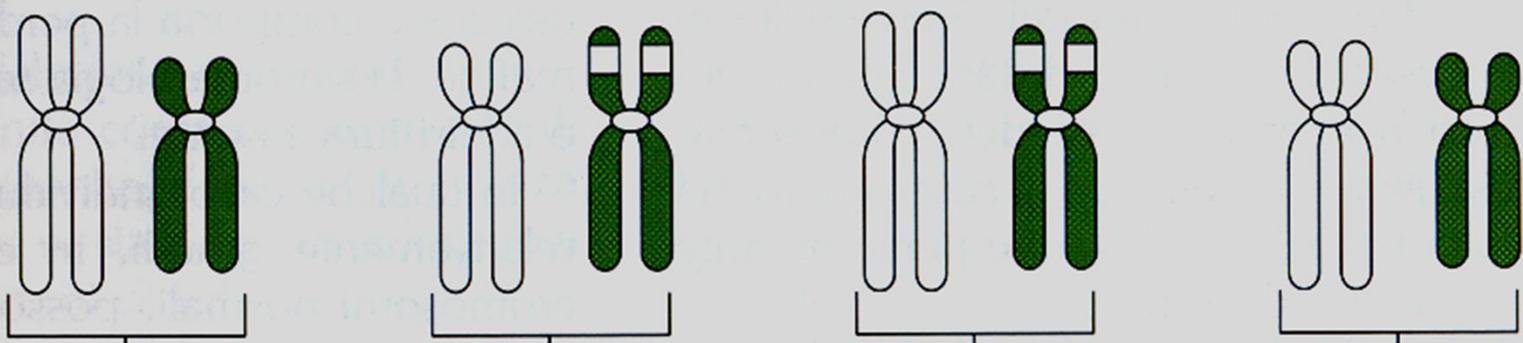




Eterozigote  
per inserzione



Possibili  
gameti



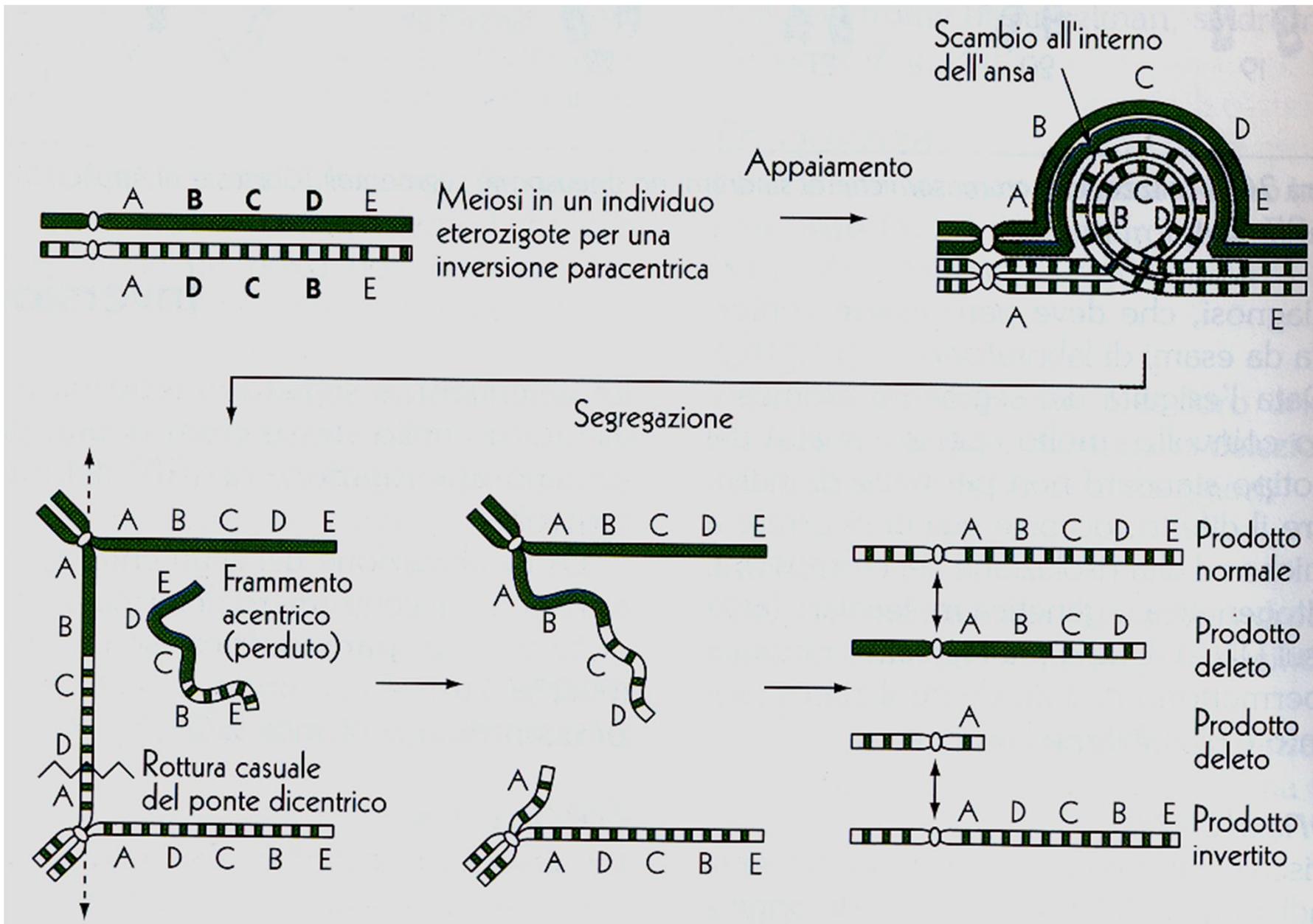
Zigoti

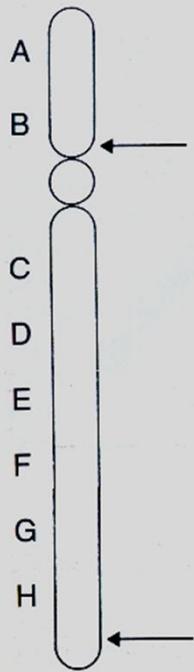
Normale

Eterozigote  
bilanciato

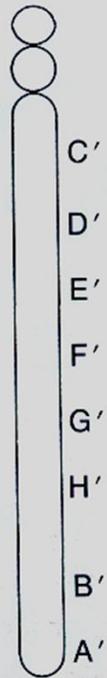
Con  
duplicazione

Con  
delezione

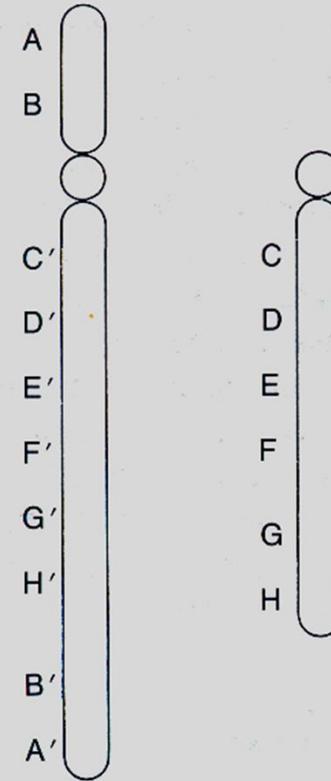
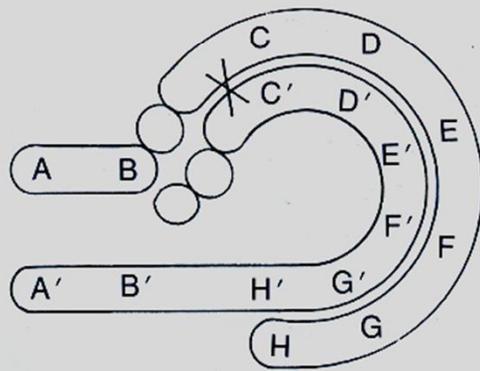




4



inv (4)



dup (4) (AB), del (4) (AB)  
prodotti della ricombinazione