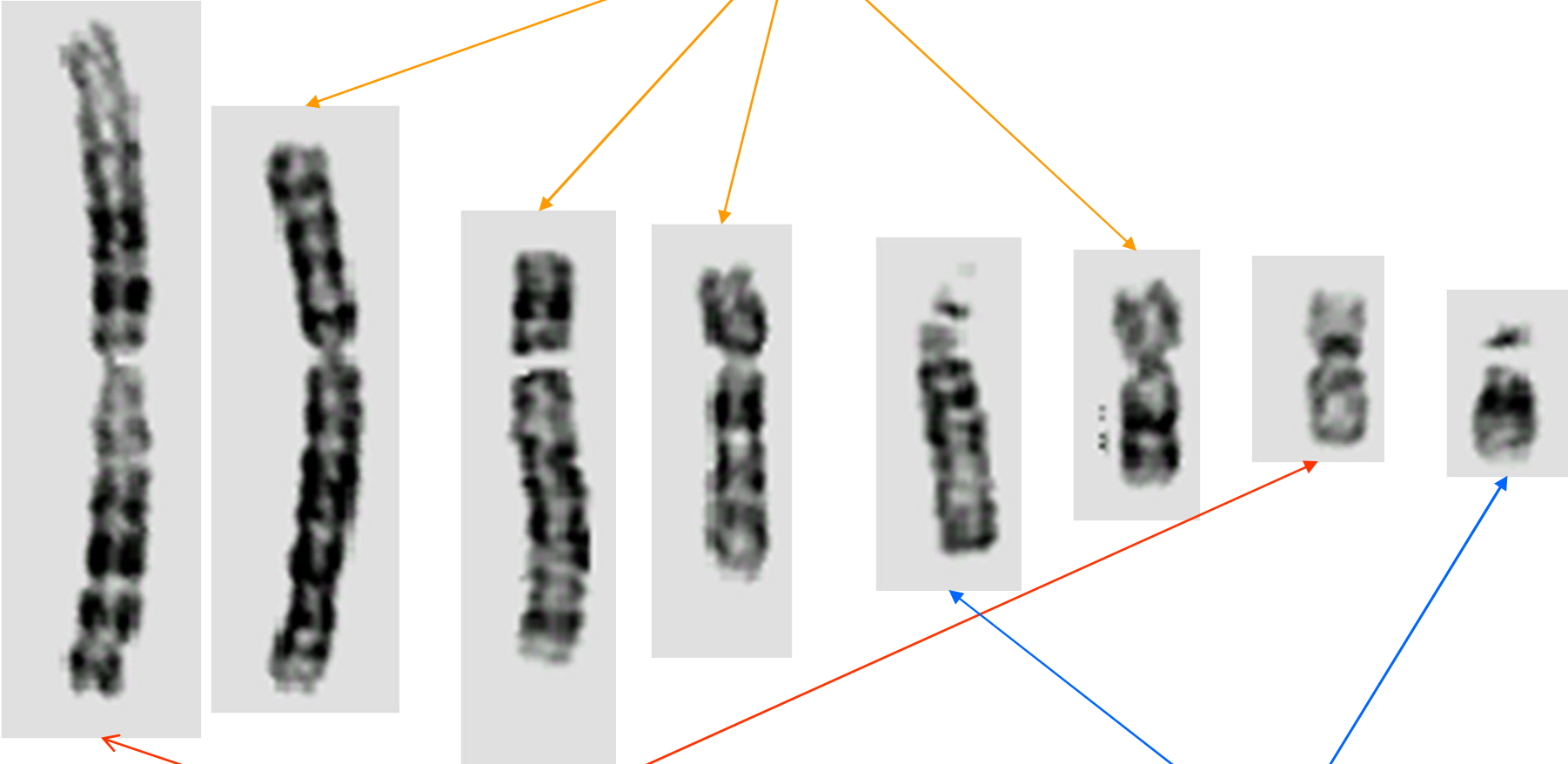


Alterazioni di struttura dei cromosomi

Submetacentric

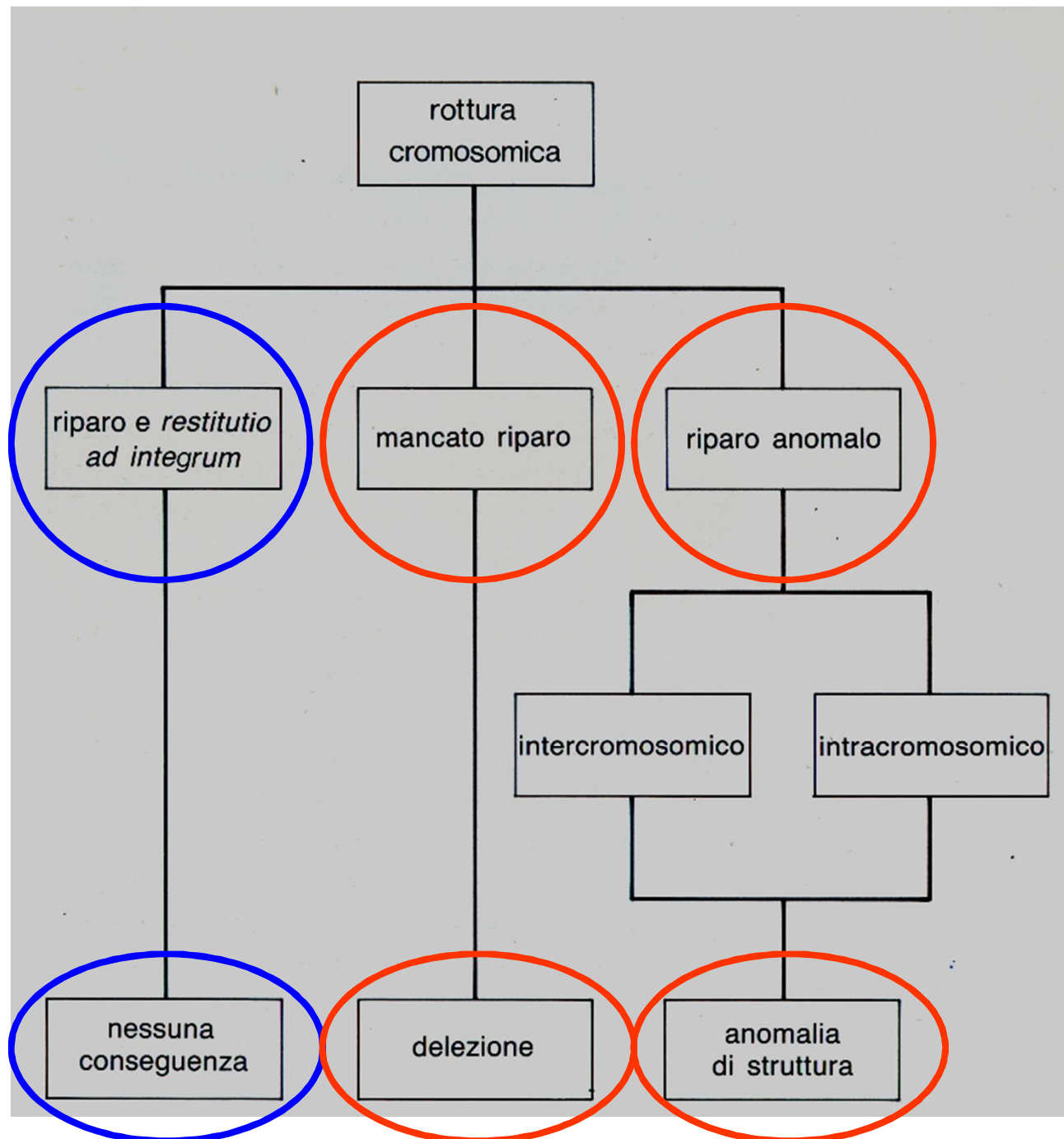


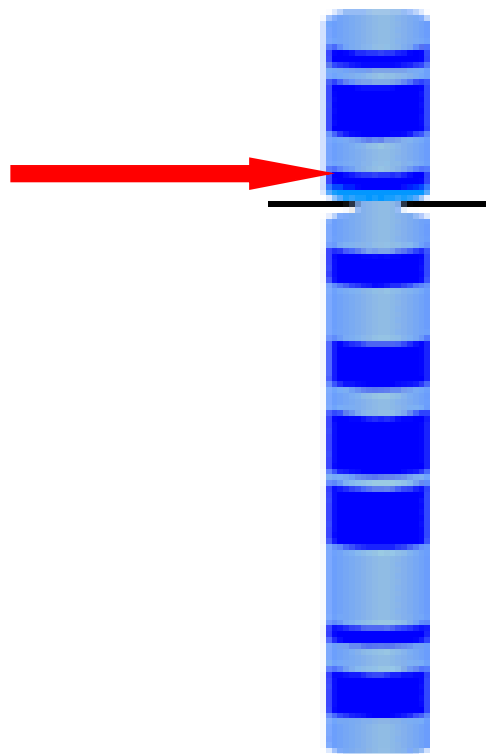
Metacentric

Acrocentric

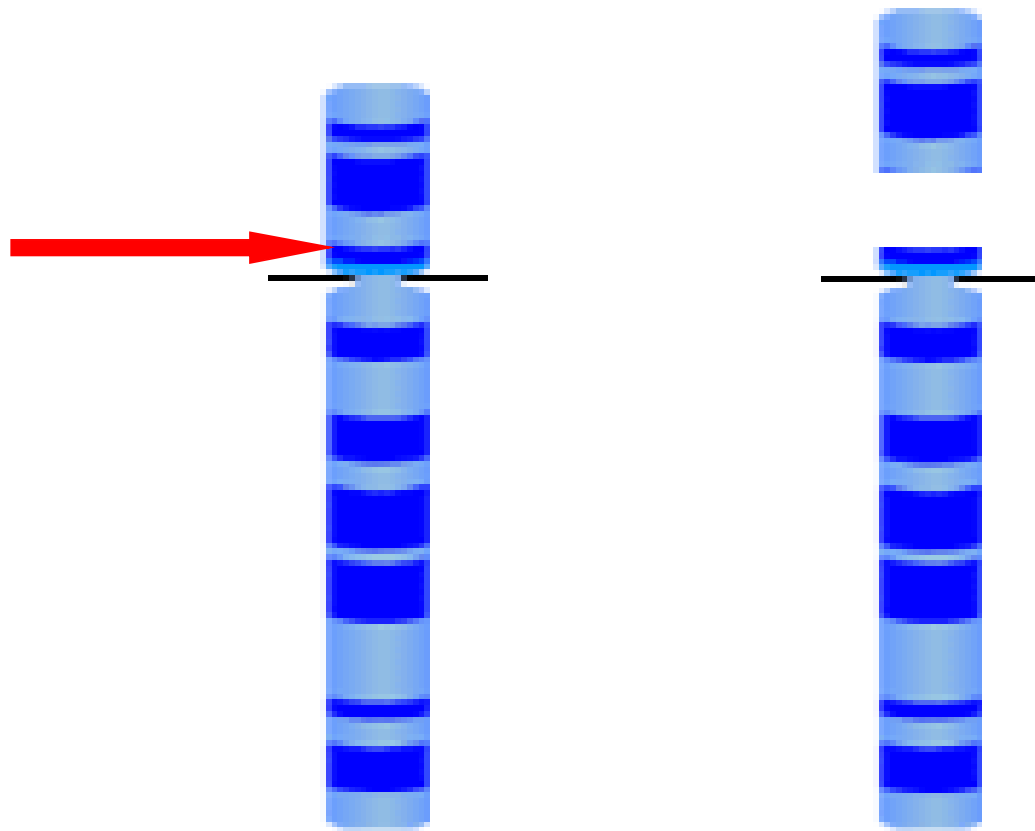
Alterazioni strutturali dei cromosomi

**sono il risultato di rotture
cromosomiche**

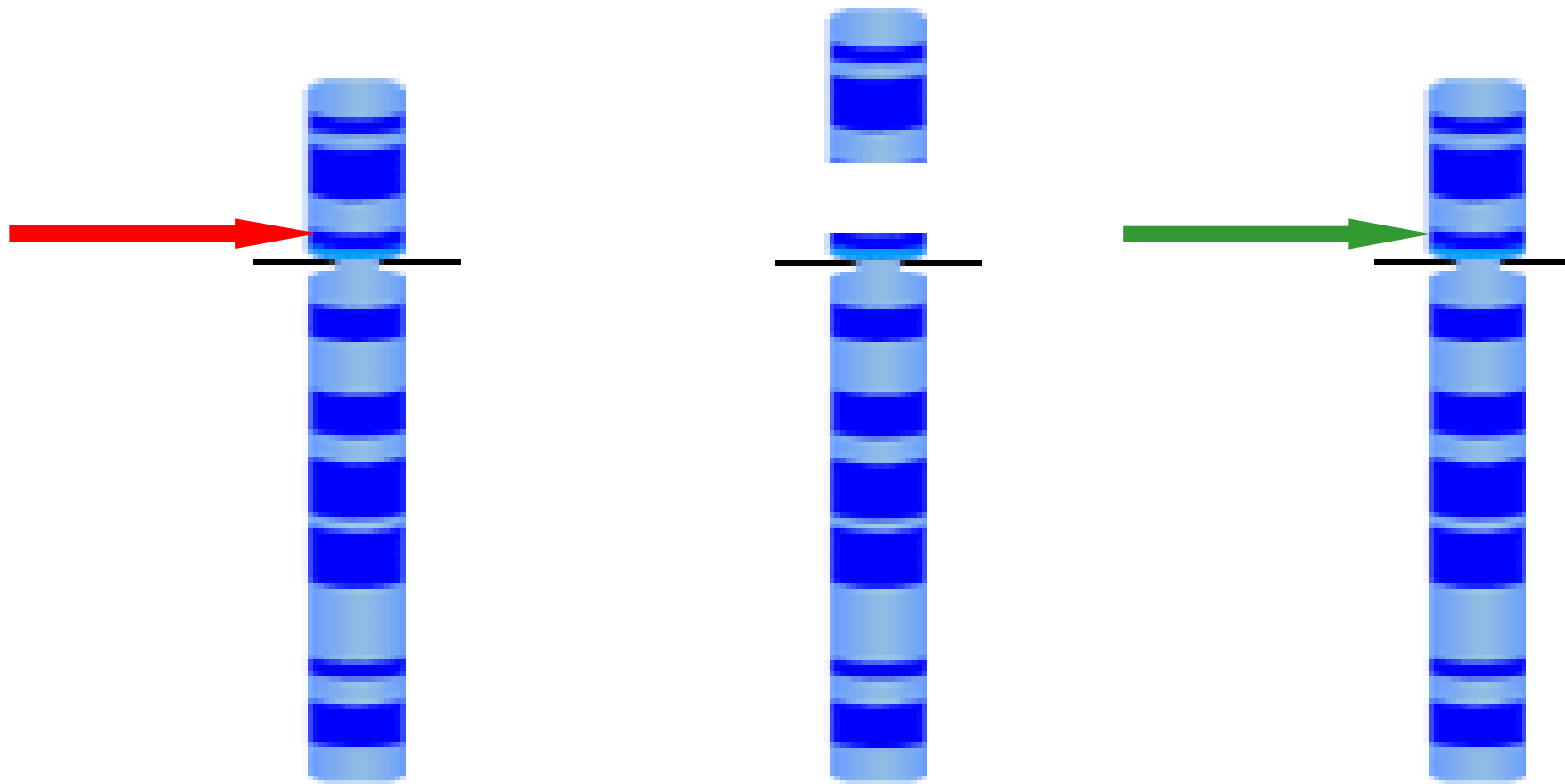




Cromosoma integro
Si verifica una rottura



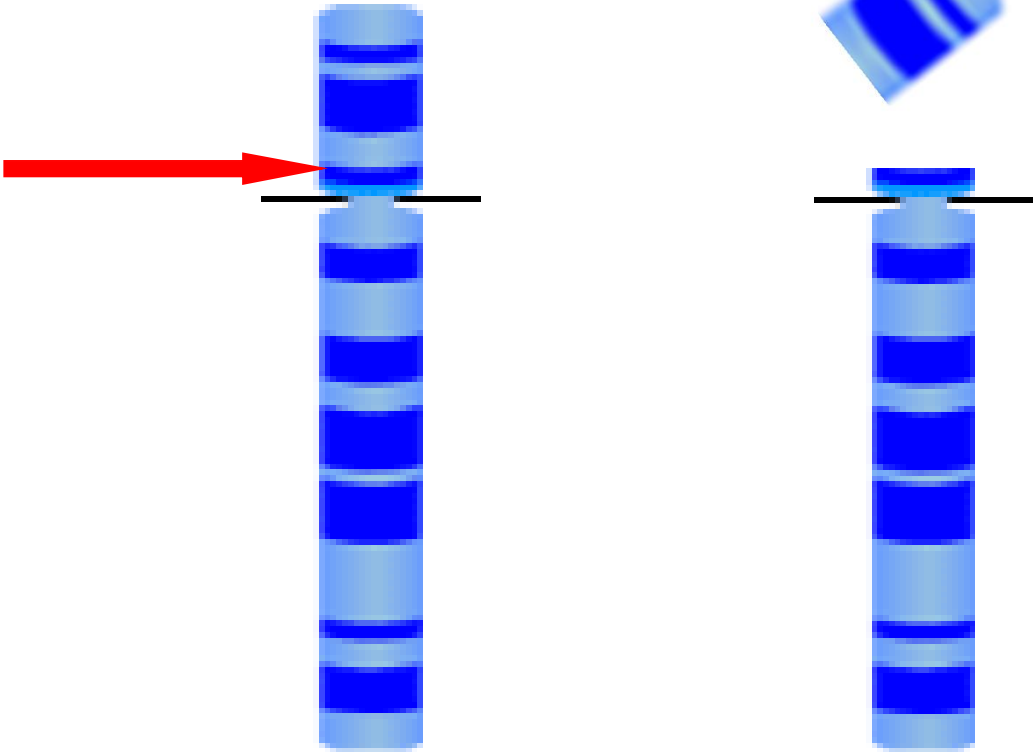
Cromosoma integro
Si verifica una rottura



Cromosoma integro
Si verifica una rottura

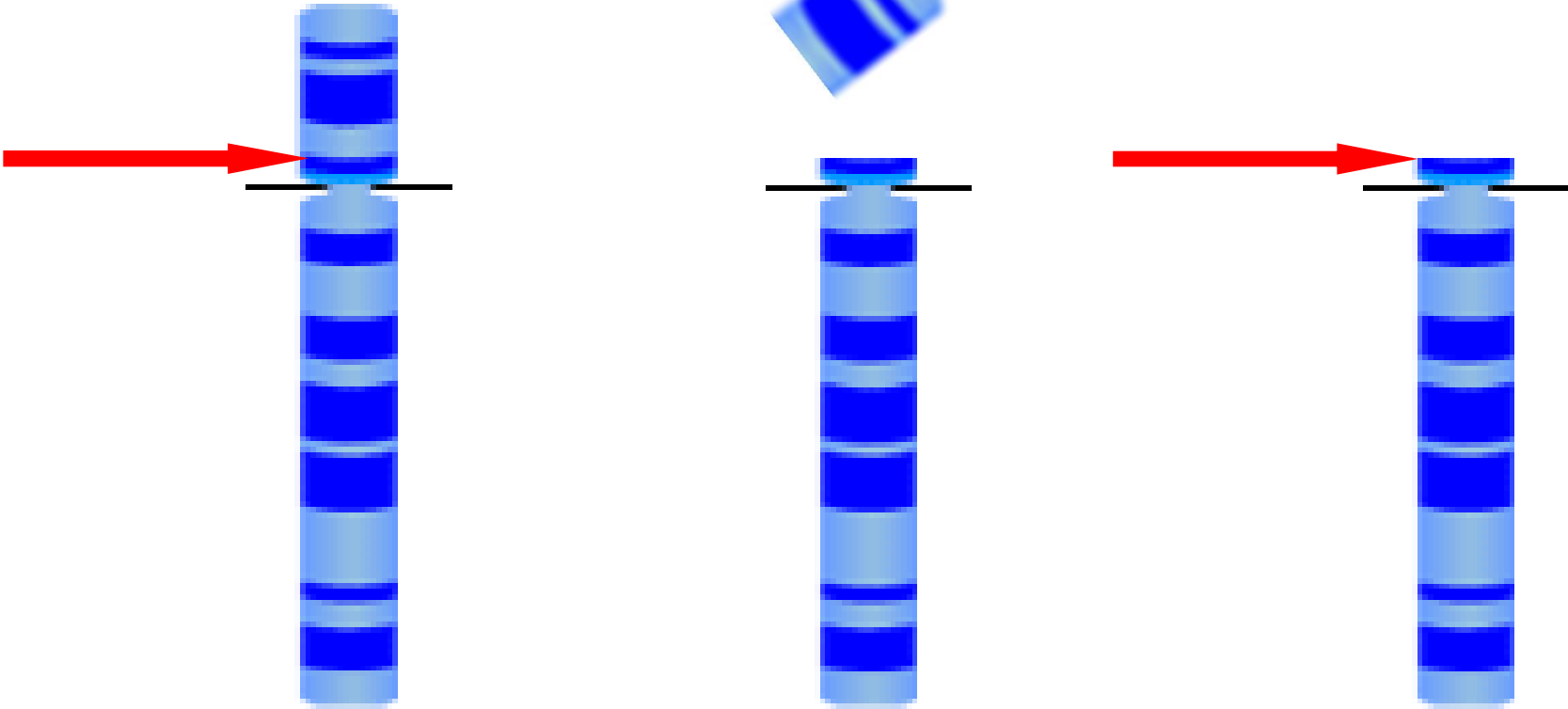
Riparo e
restitutio ad integrum

Cromosoma acentrico: si perde



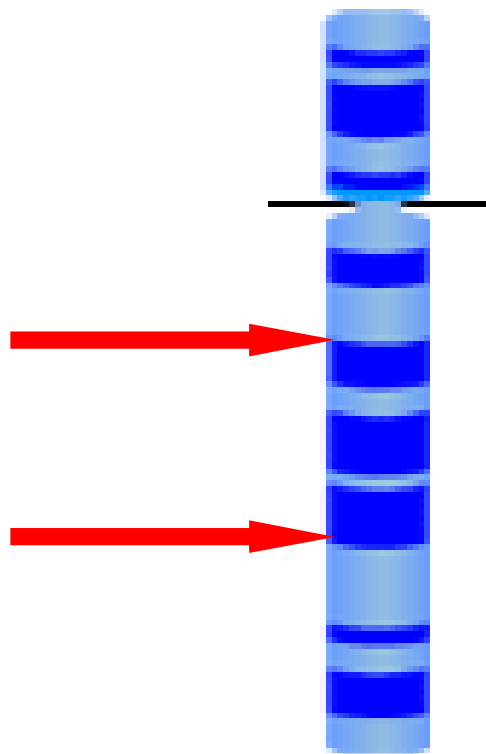
**Cromosoma integro
Si verifica una rottura**

Cromosoma acentrico: si perde

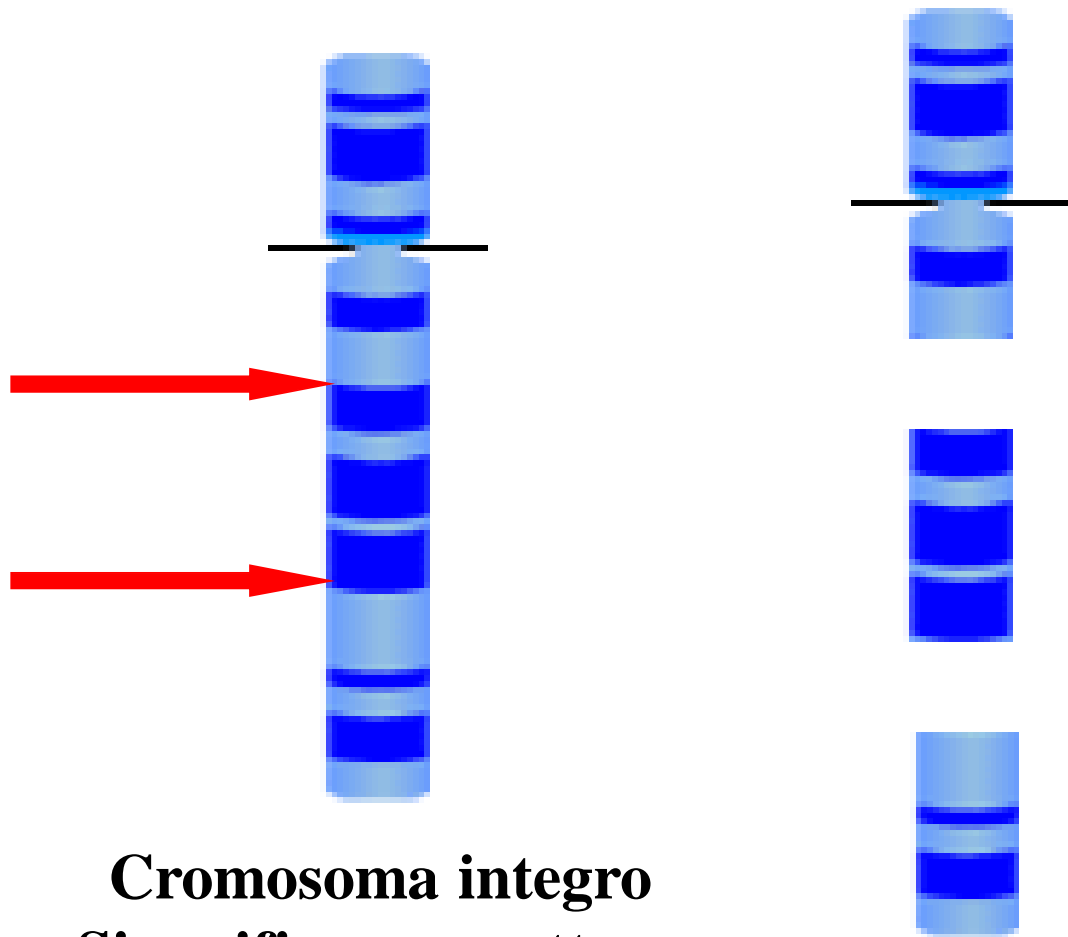


**Cromosoma integro
Si verifica una rottura**

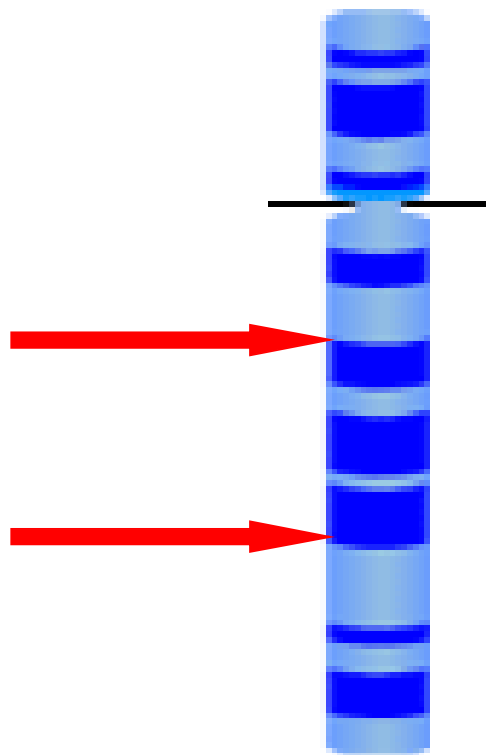
**Cromosoma
deleto**



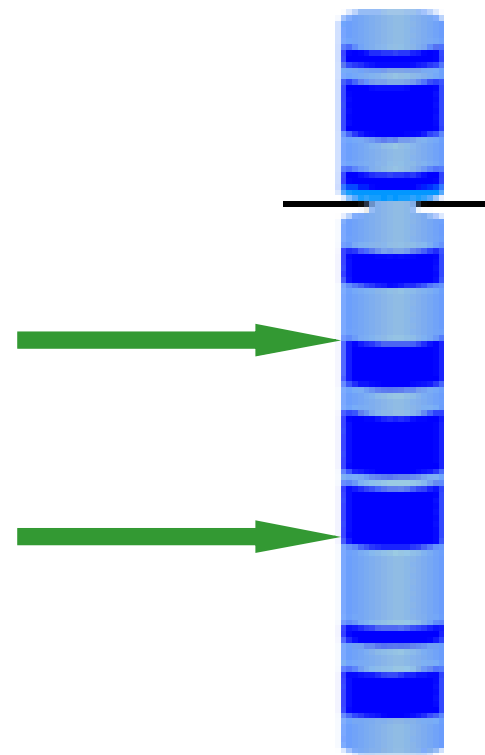
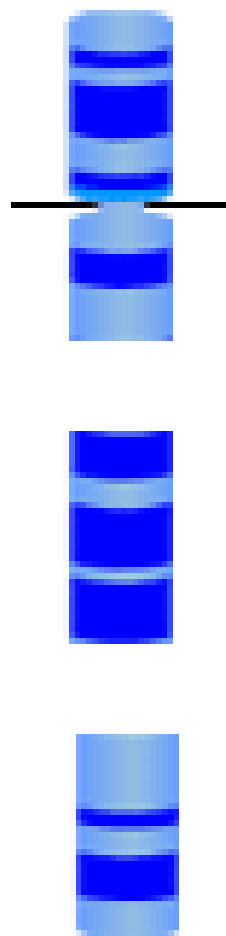
Cromosoma integro
Si verifica una rottura



Cromosoma integro
Si verifica una rottura

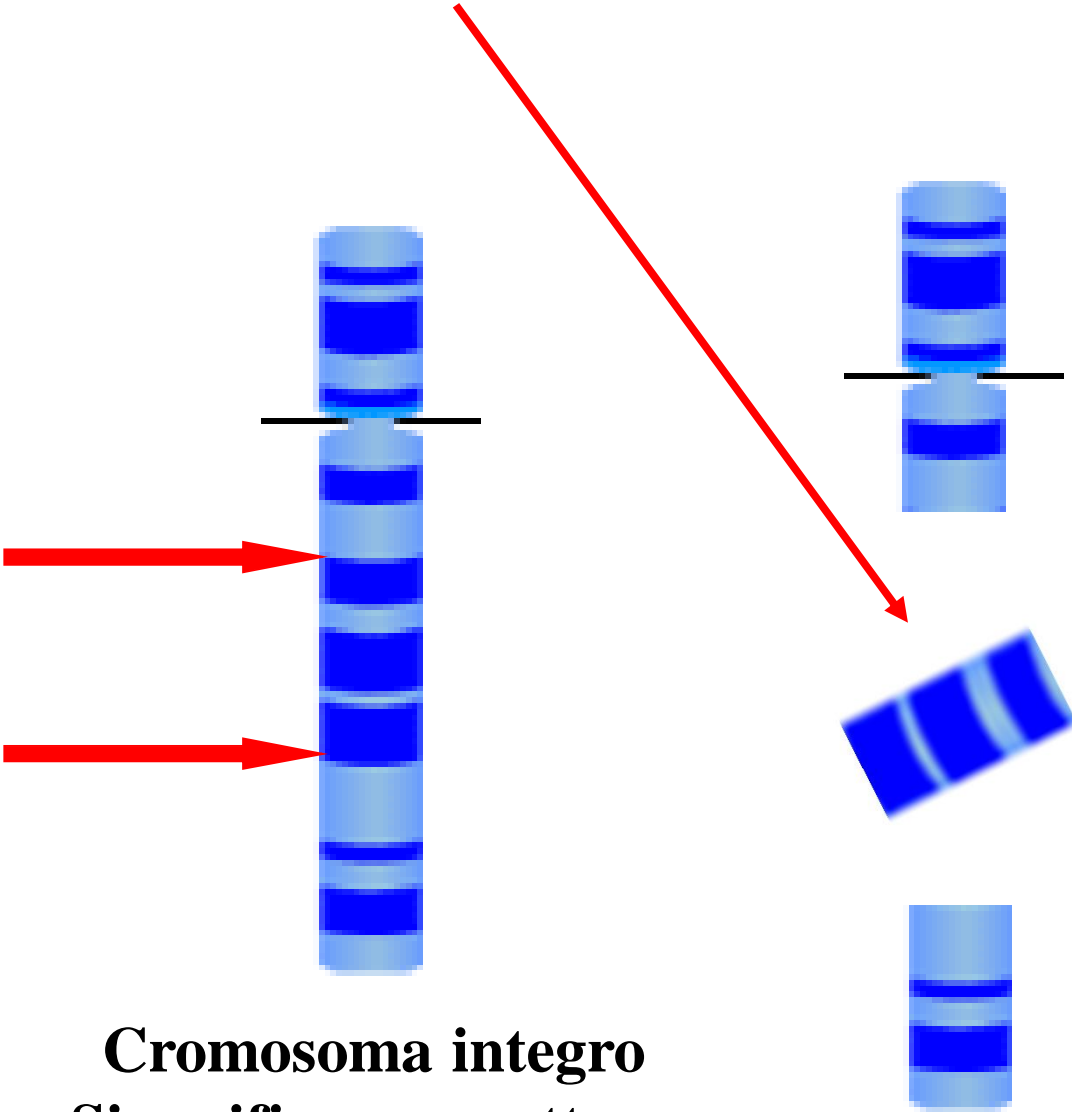


**Cromosoma integro
Si verifica una rottura**



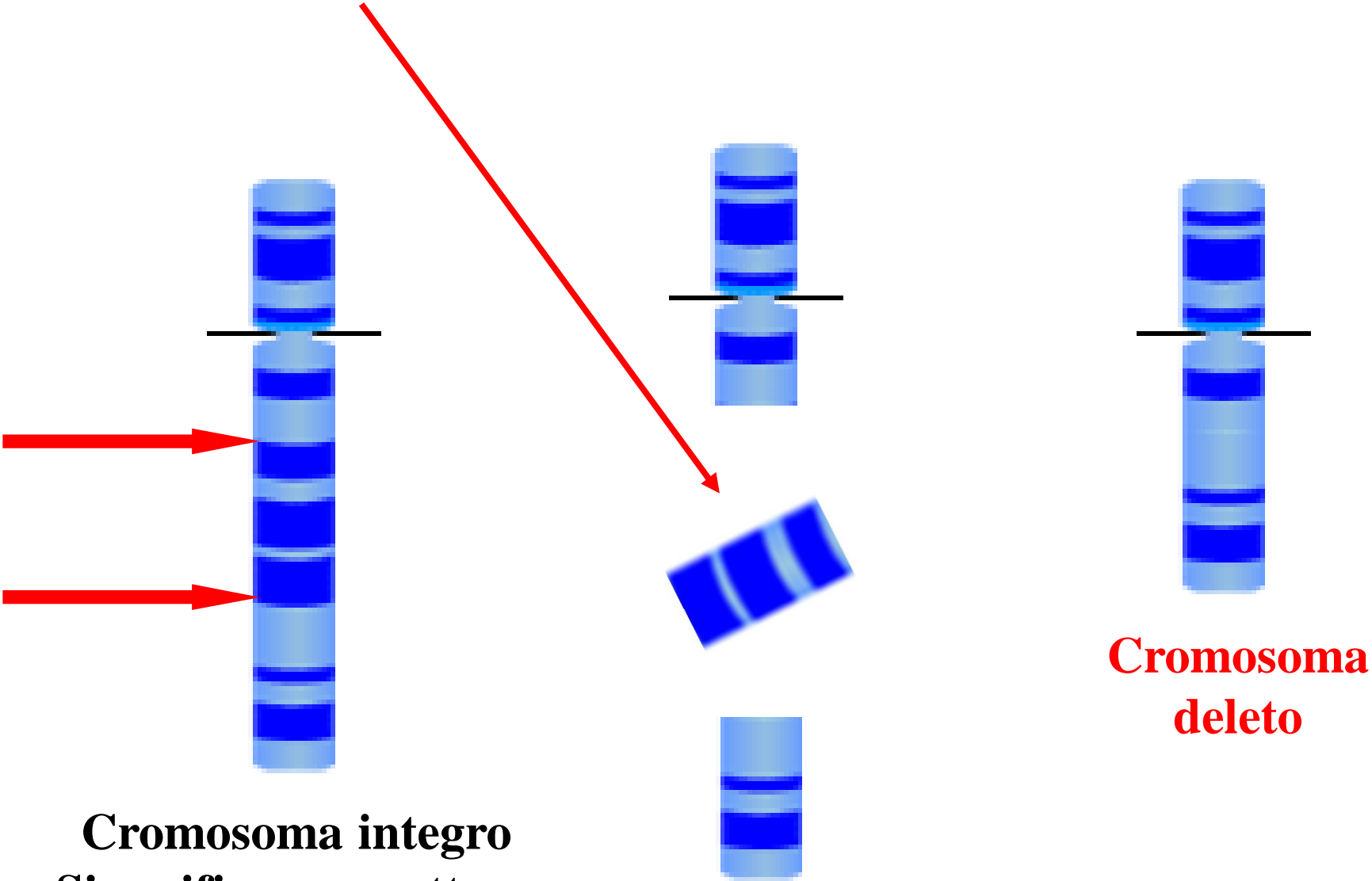
**Riparo e
restitutio ad integrum**

Cromosoma acentrico: si perde



**Cromosoma integro
Si verifica una rottura**

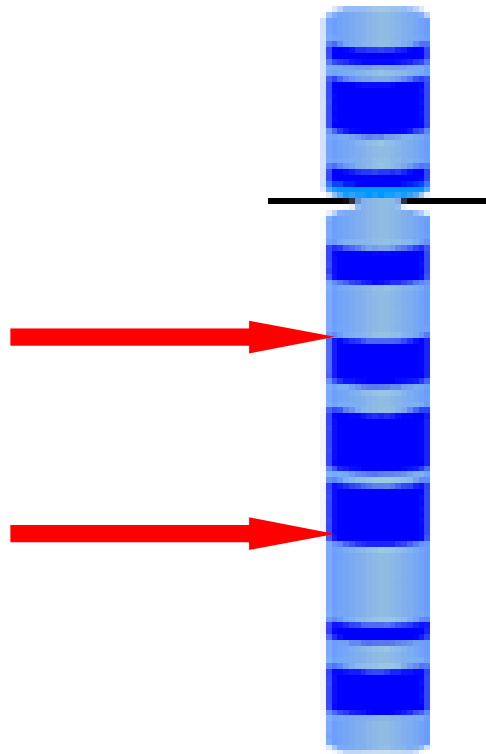
Cromosoma acentrico: si perde



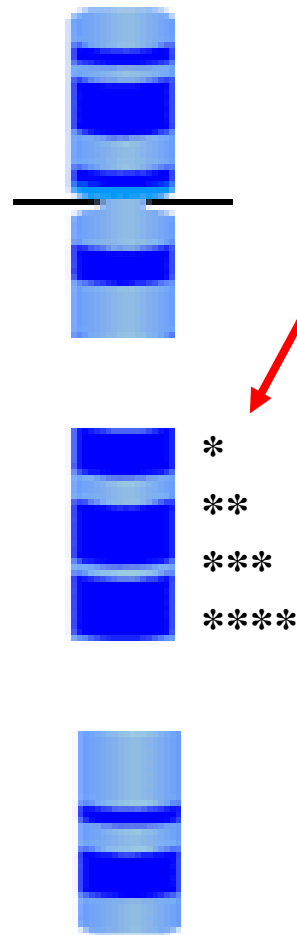
**Cromosoma integro
Si verifica una rottura**

**Cromosoma
deleteo**

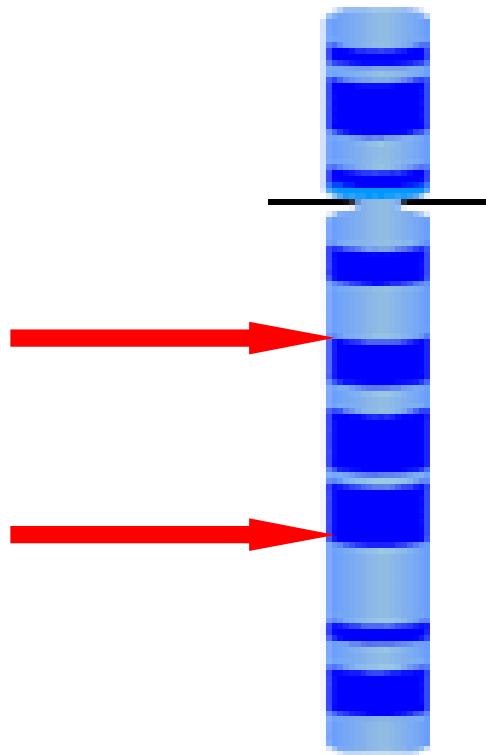
Cromosoma acentrico: RUOTA di 180°



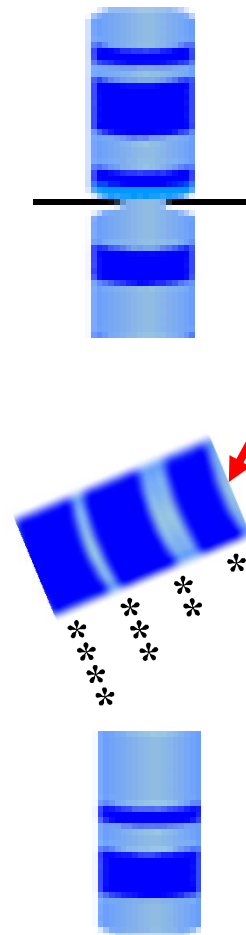
**Cromosoma integro
Si verifica una rottura**

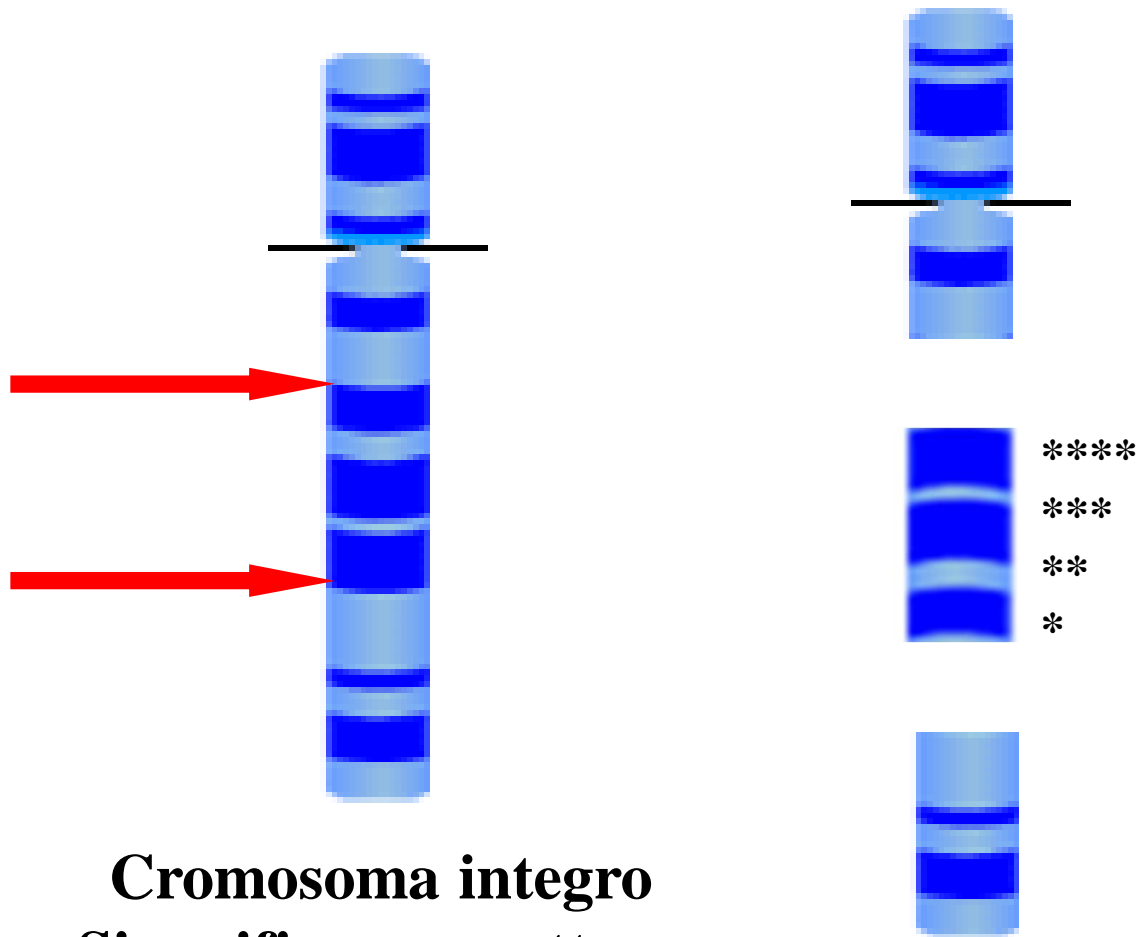


Cromosoma acentrico: RUOTA di 180°

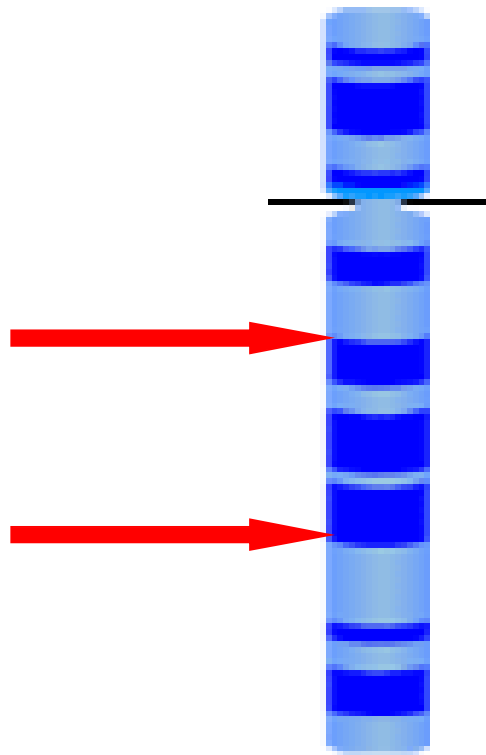


**Cromosoma integro
Si verifica una rottura**

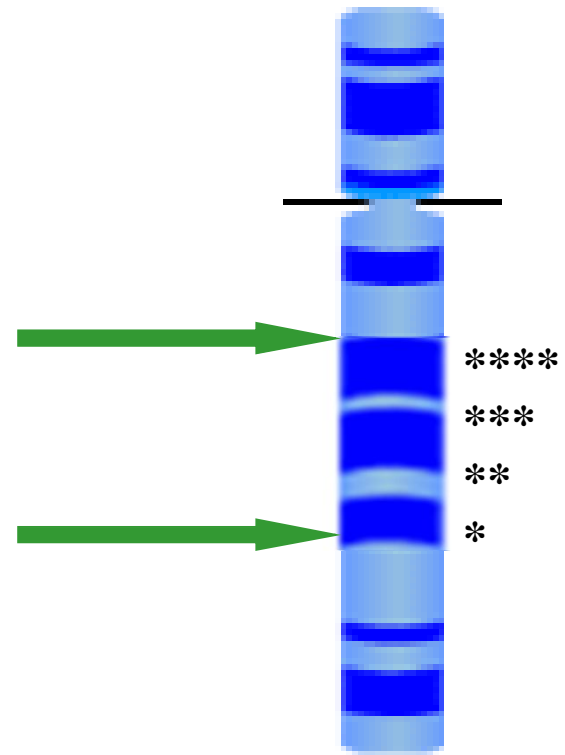
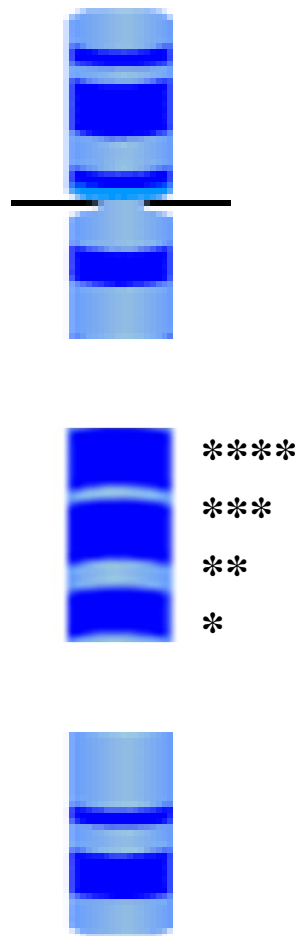




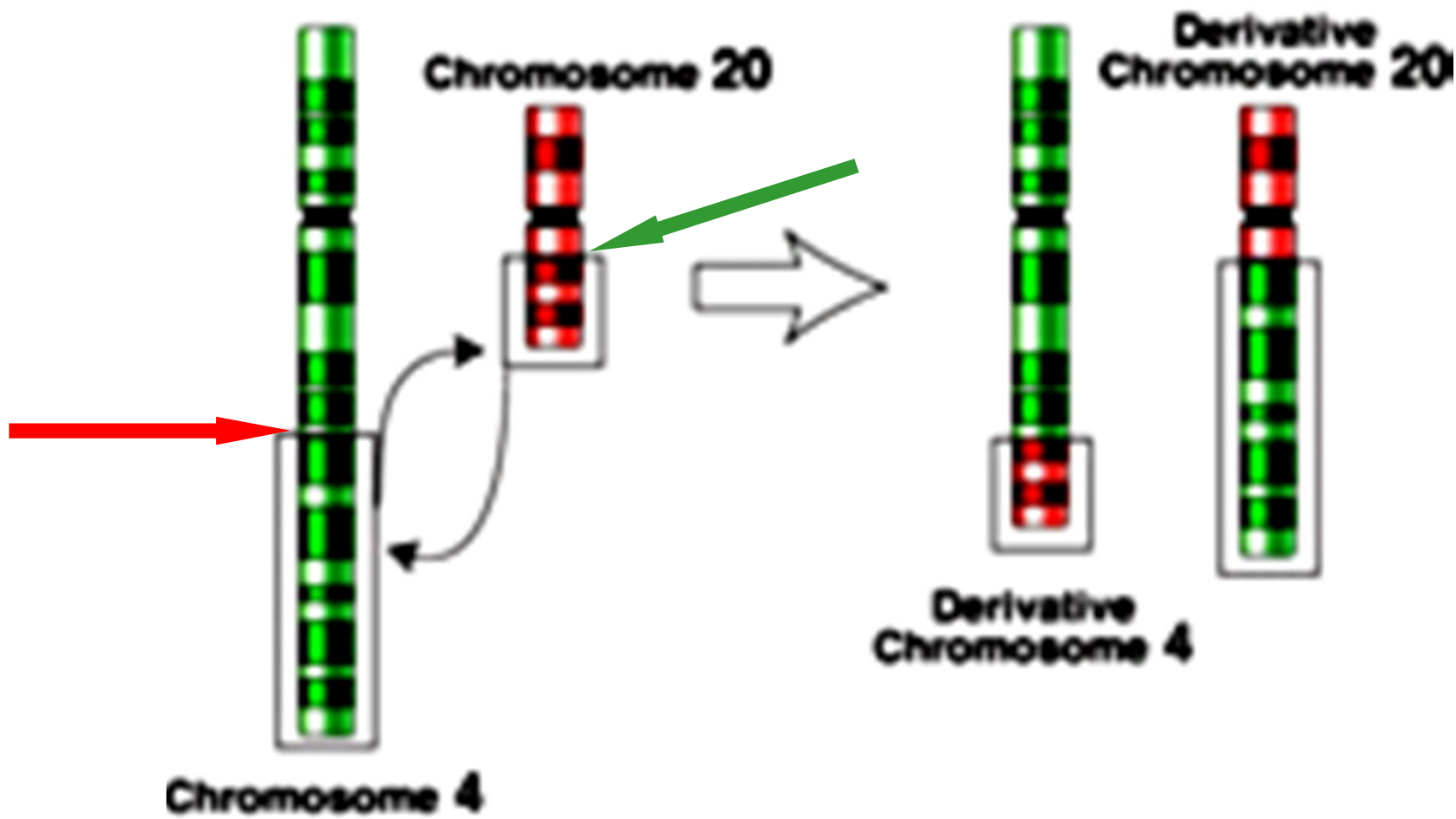
Cromosoma integro
Si verifica una rottura

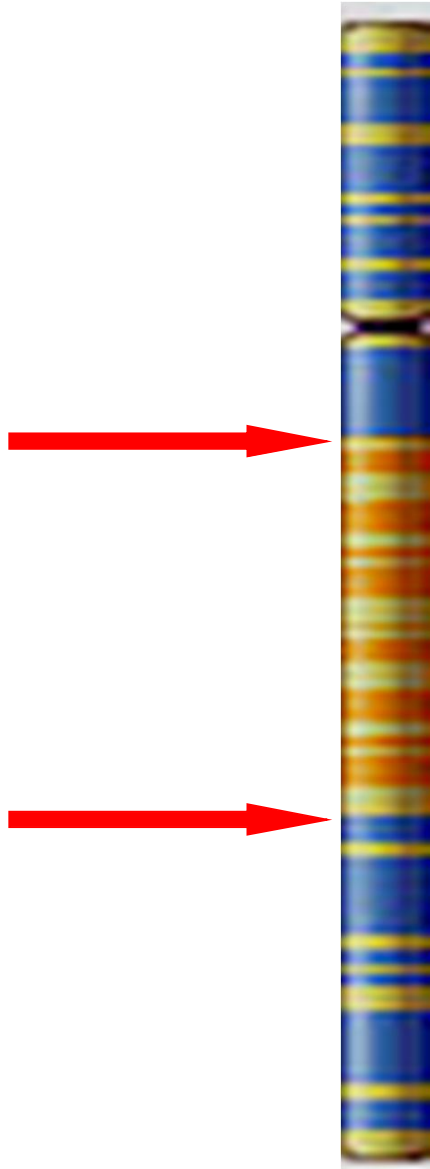
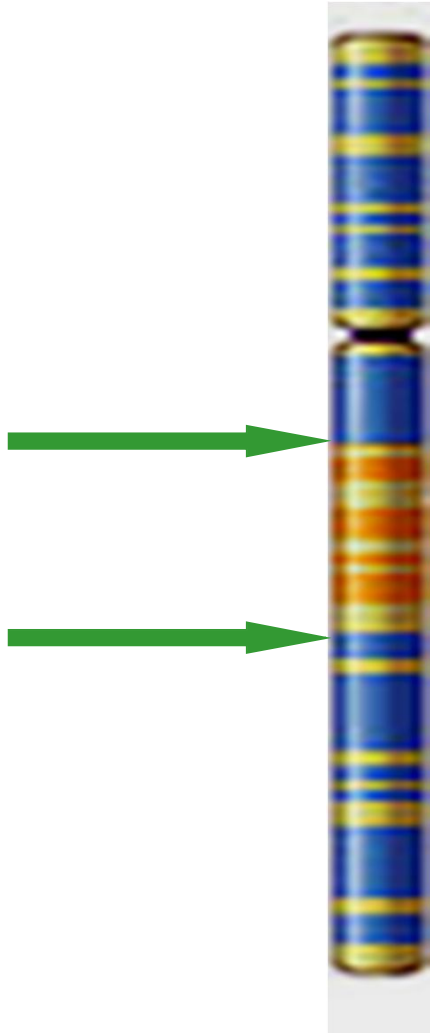


**Cromosoma integro
Si verifica una rottura**



**Cromosoma
Con Inversione**





**Cromosoma con duplicazione
di un segmento**

Anomalie cromosomiche di struttura

Traslocazioni

Bilanciate

Inversioni

Delezioni

Sbilanciate

Duplicazioni

Anomalie cromosomiche di struttura

Bilanciate

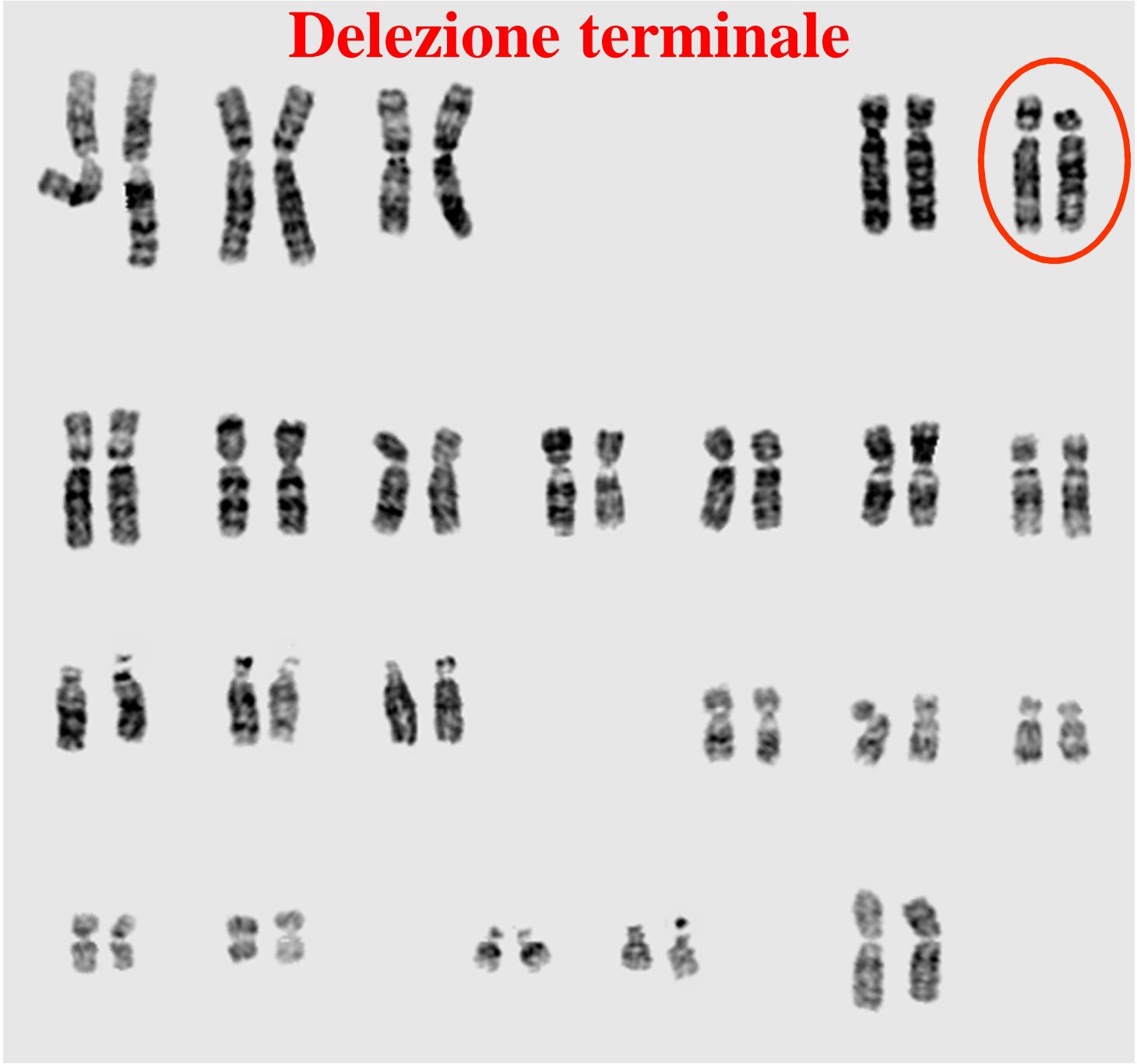
(nella maggioranza dei casi)

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.



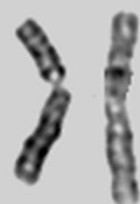
Principali sindromi da microdelezione

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%

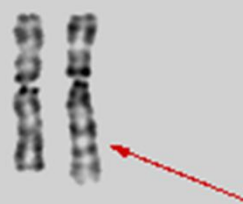
Traslocazioni



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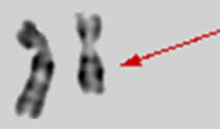
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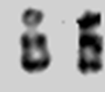
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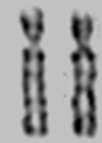
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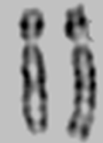
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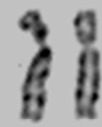
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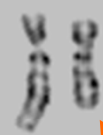
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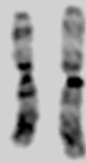
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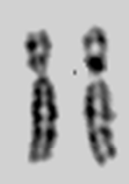
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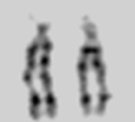
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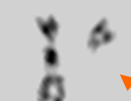
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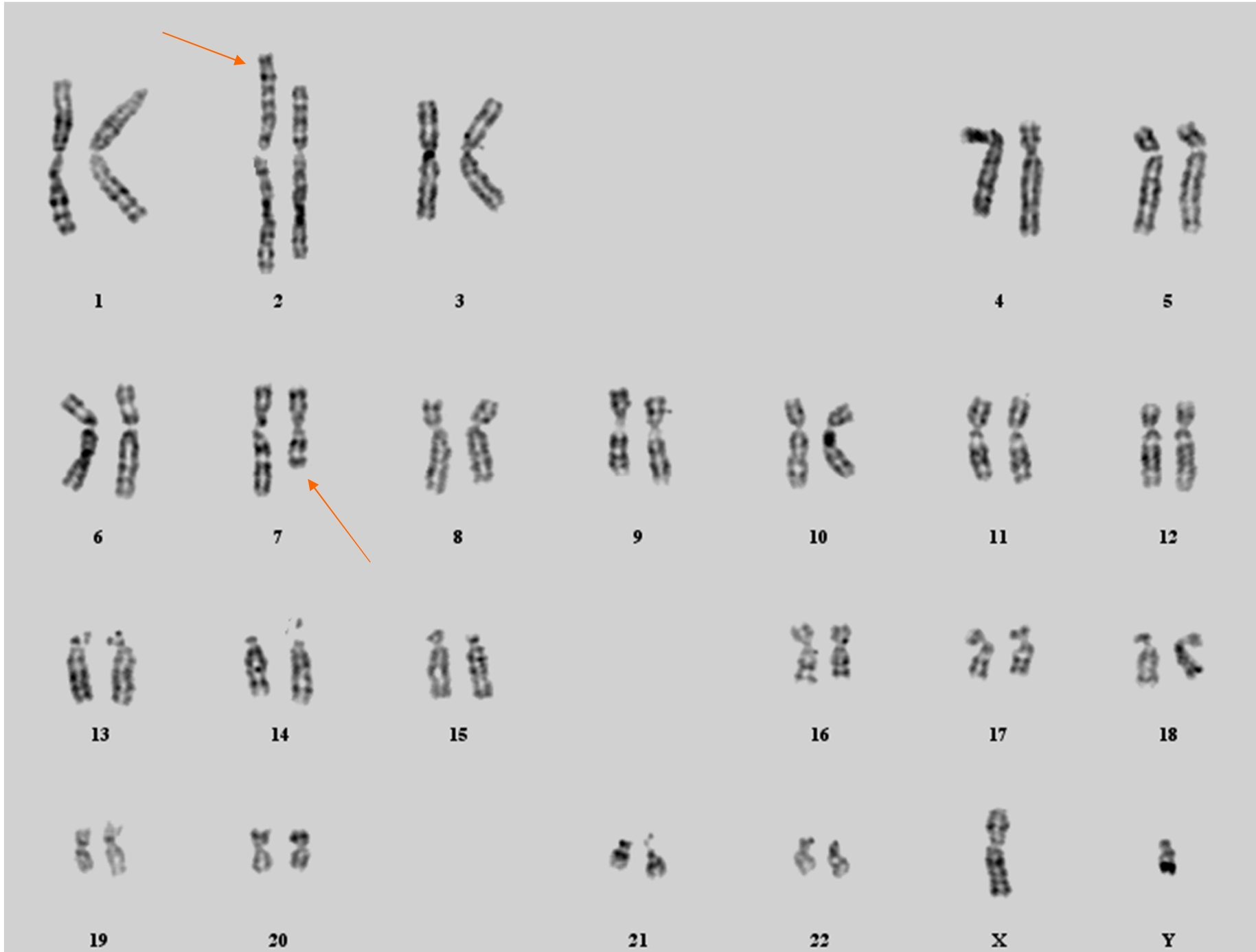
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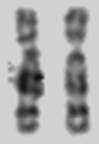
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Traslocazioni robertsoniane



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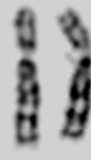
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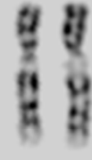
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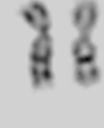
14



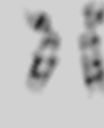
15



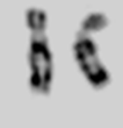
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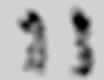
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Inversione paracentrica



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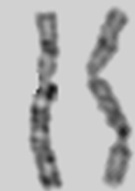
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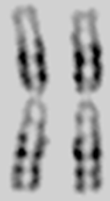


Inv. Par.



Normale

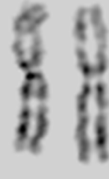
Inversione pericentrica



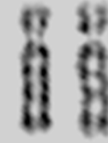
1



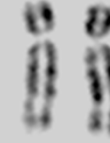
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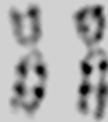
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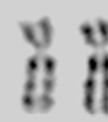
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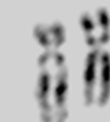
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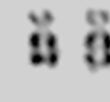
15



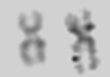
16



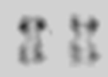
17



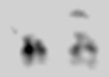
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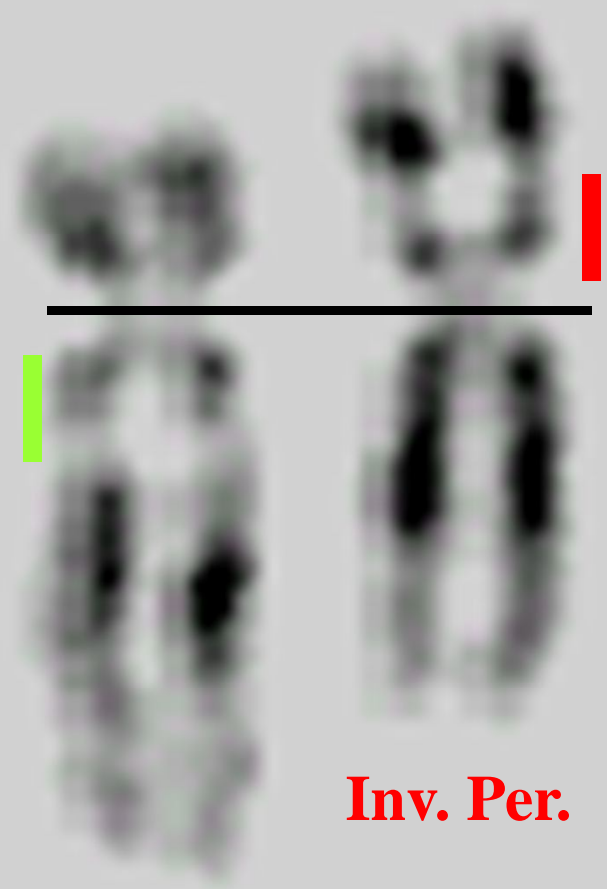
22



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Normale

Inv. Per.



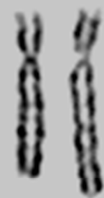
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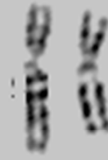
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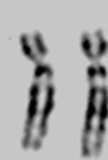
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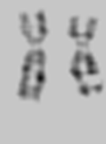
13



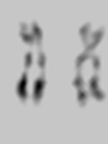
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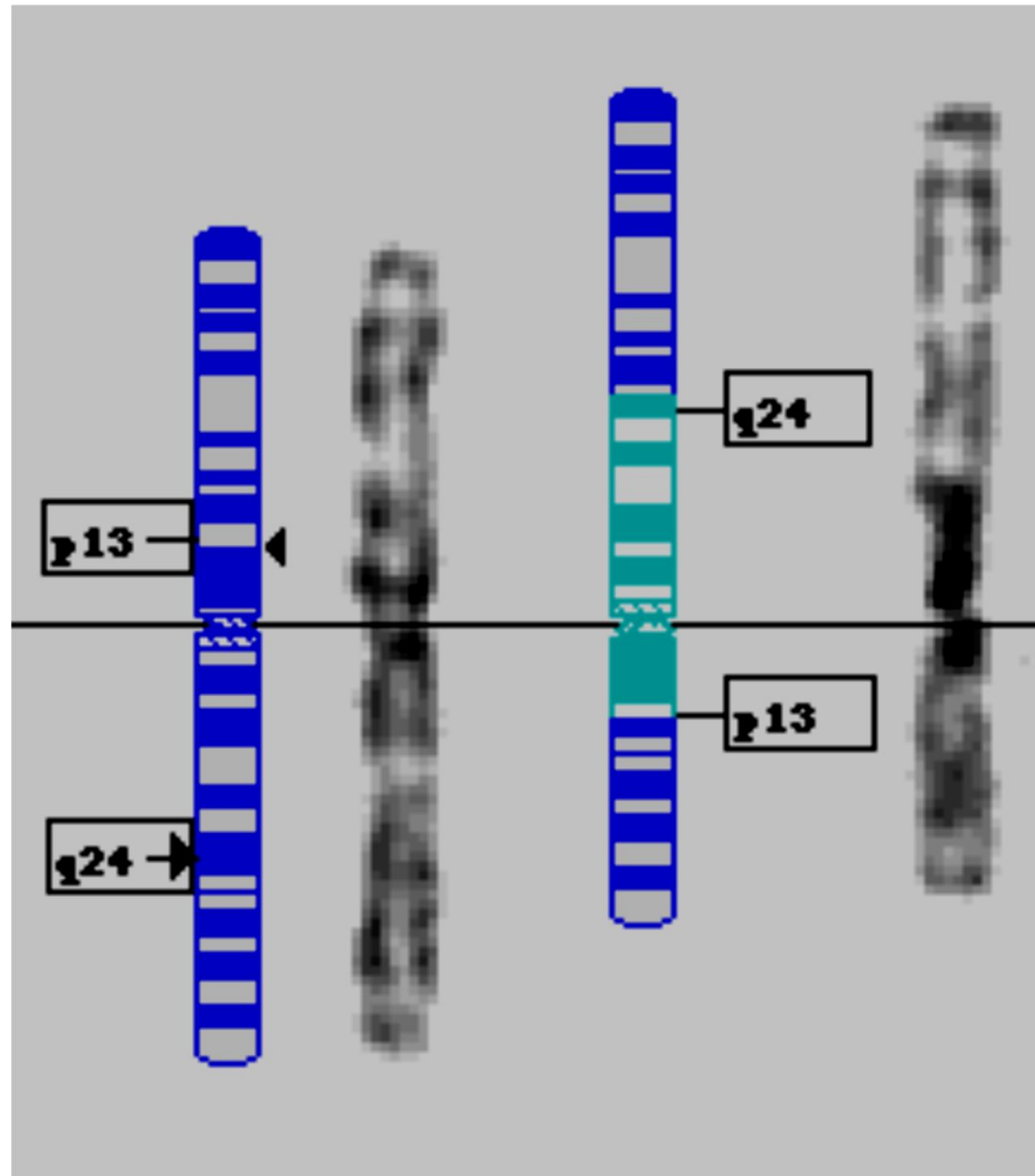
22



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Inversione pericentrica eterocromatina



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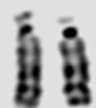
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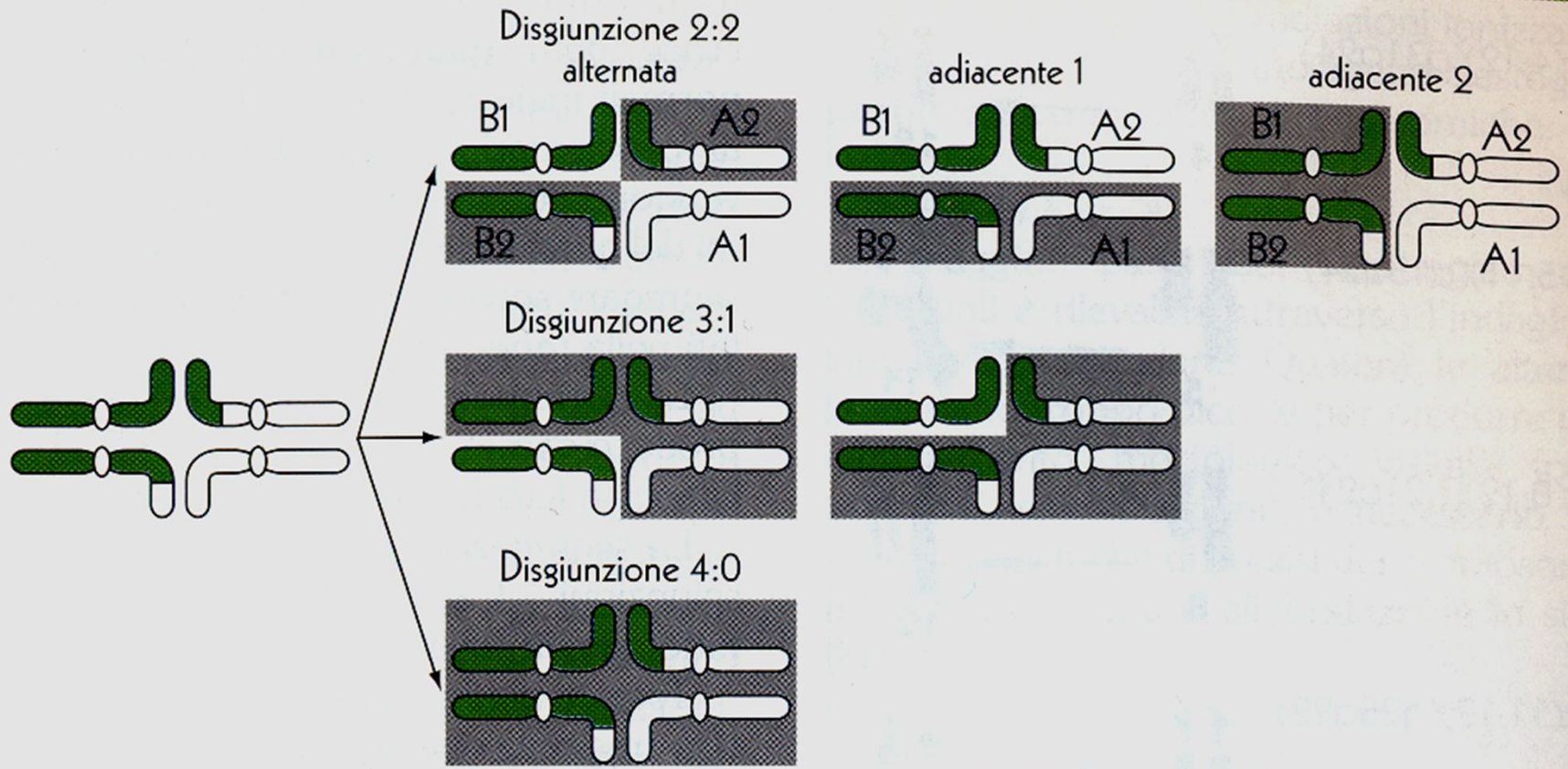
Y

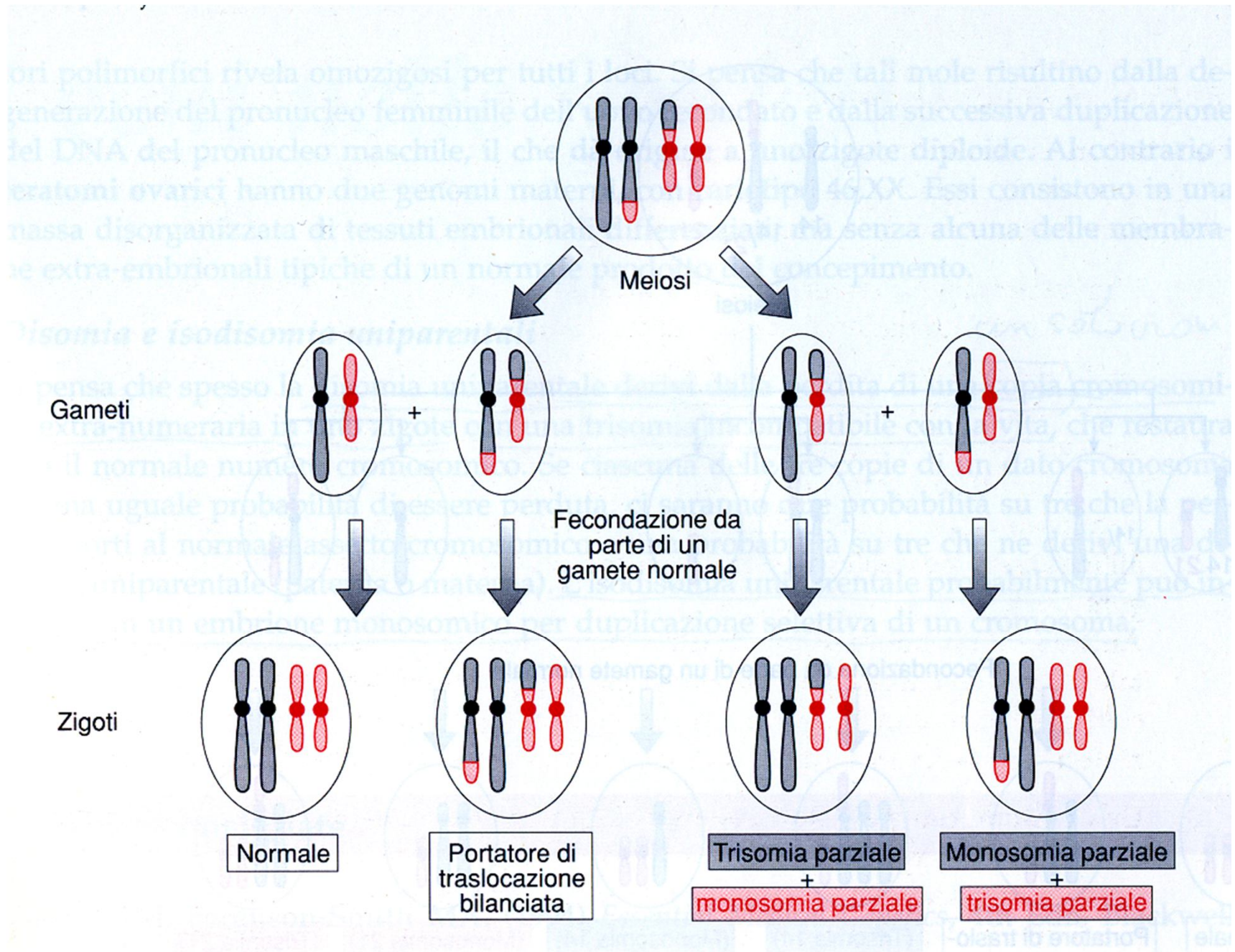
Handwritten text, possibly a signature or name, with a small orange mark above it.

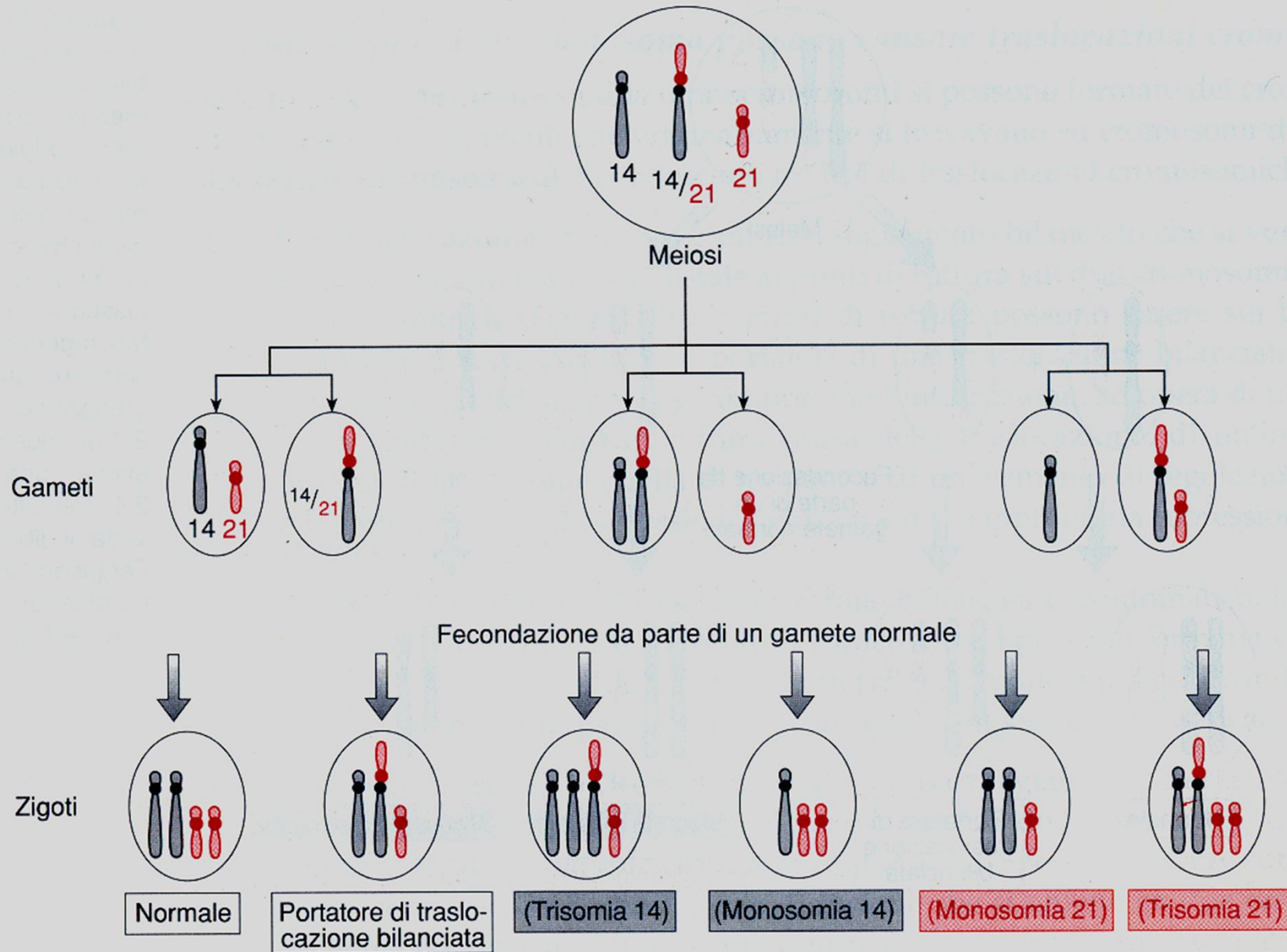
Anomalie cromosomiche bilanciate?

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

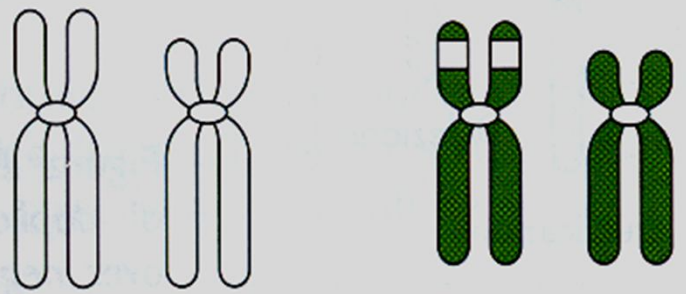
ma riguarda la sua discendenza...



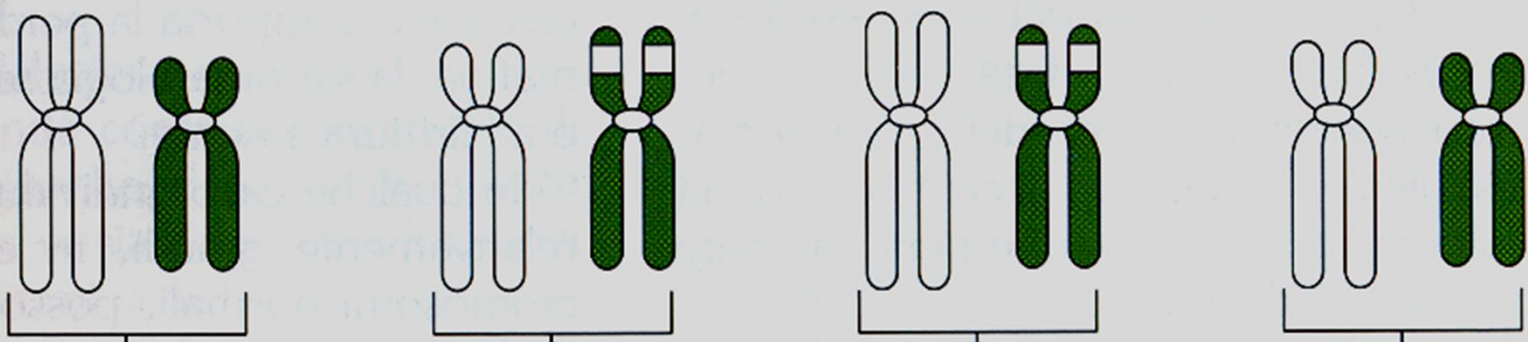




Eterozigote
per inserzione



Possibili
gameti



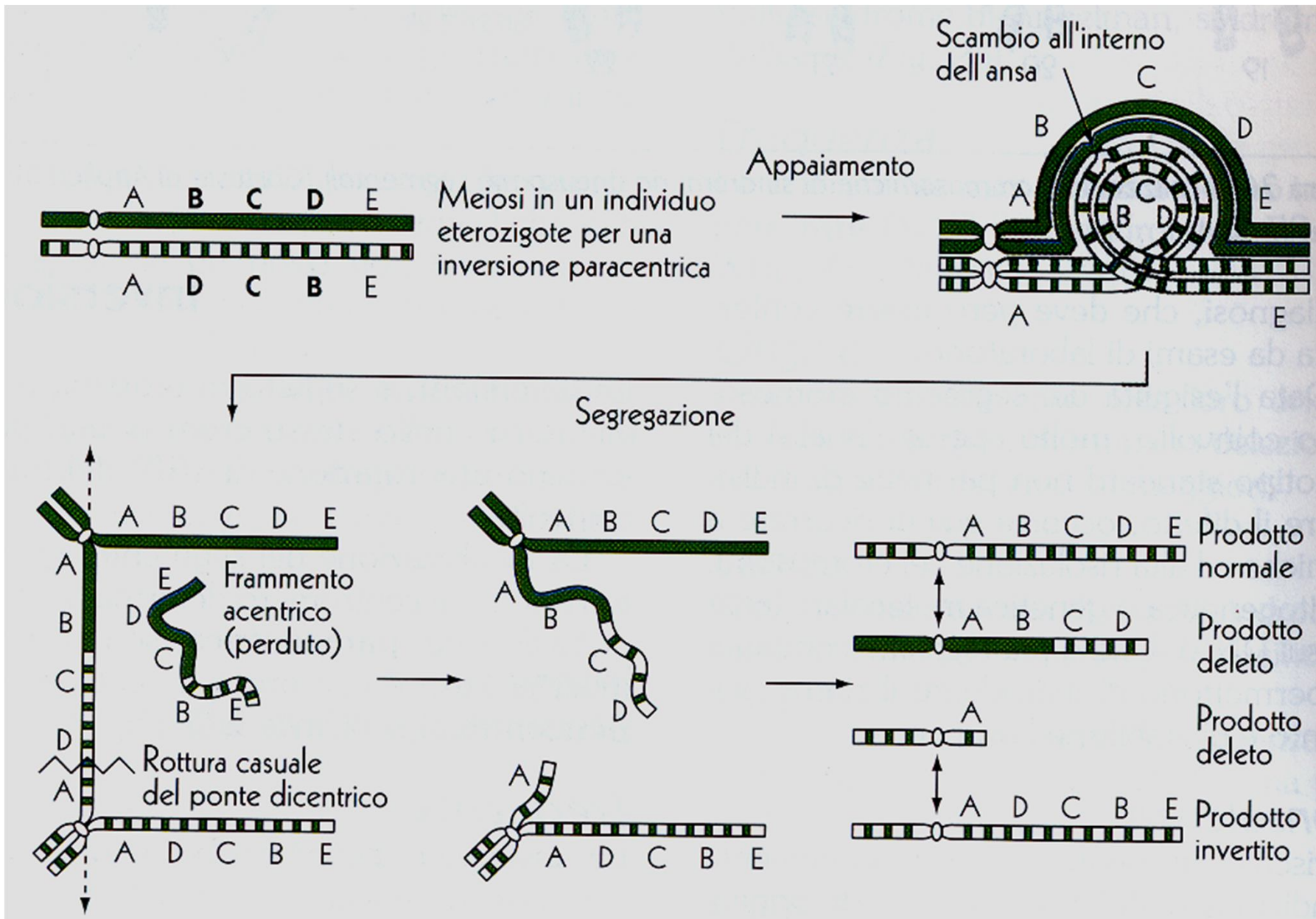
Zigoti

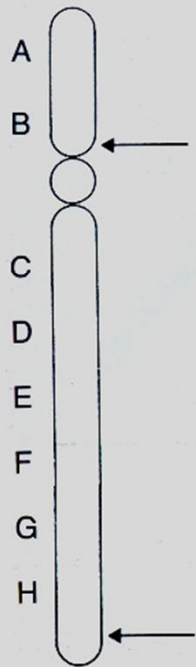
Normale

Eterozigote
bilanciato

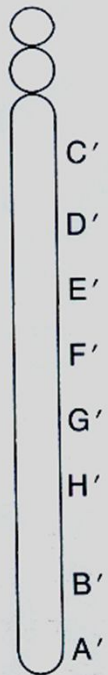
Con
duplicazione

Con
delezione

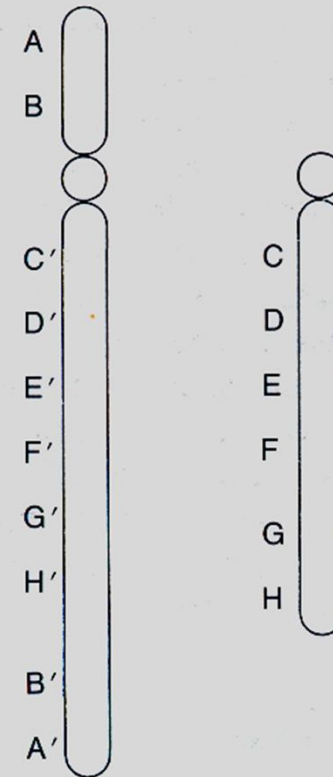
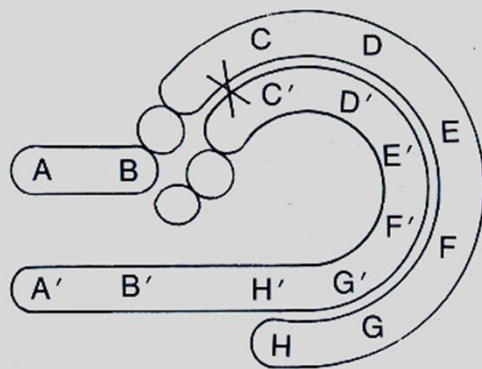




4



inv(4)



dup(4)(AB), del(4)(AB)

prodotti della ricombinazione