

# 易位 (Translocation)

- 一、易位的基本概念
- 二、易位的减数分裂行为
- 三、易位的遗传学效应
- 四、易位与人类疾病



# 一、易位的基本概念 (Translocation)

Translocation is the movement of a chromosomal segment to **a new location** in the genome

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# 易位的主要类型

- ◆ 单向易位

(simple translocation)

- ◆ 相互易位

(reciprocal translocation)

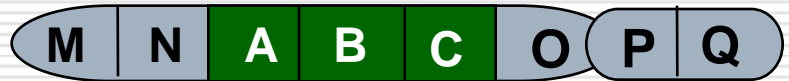
- ◆ 罗伯逊易位

(Robertsonian translocation)





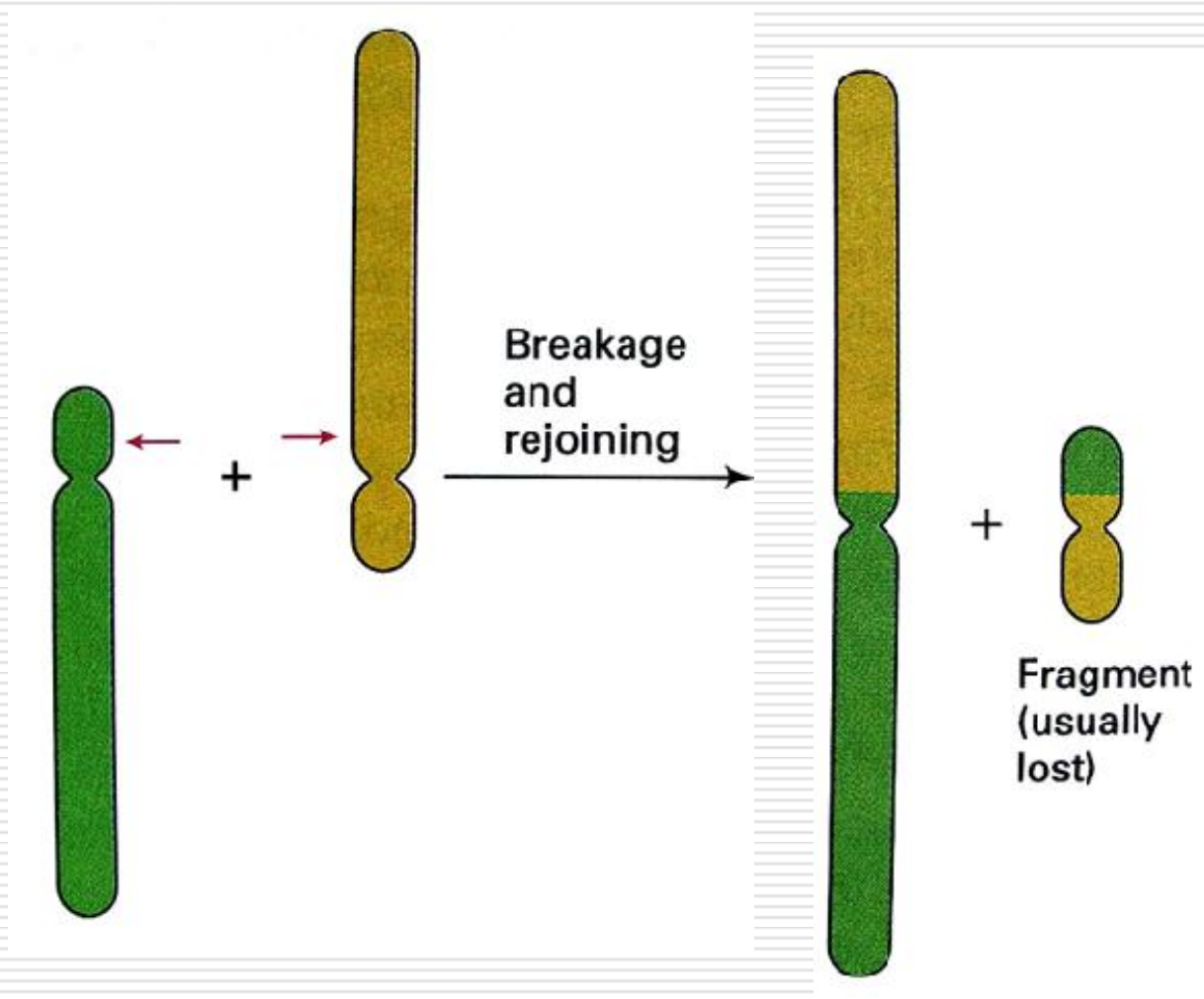
正常染色体



单向易位

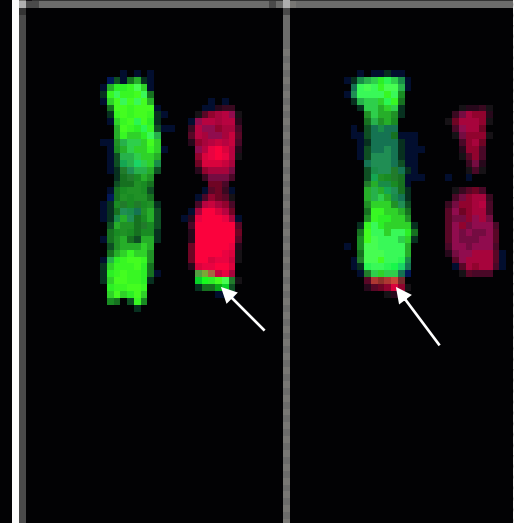
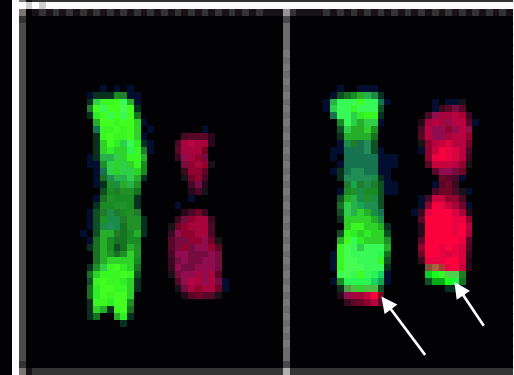
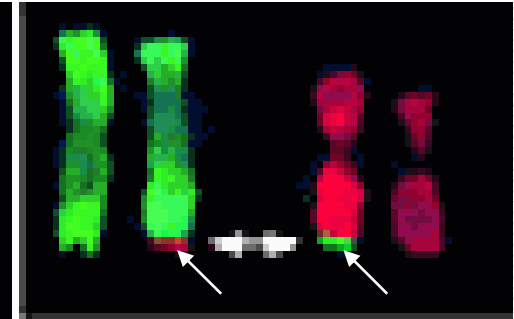
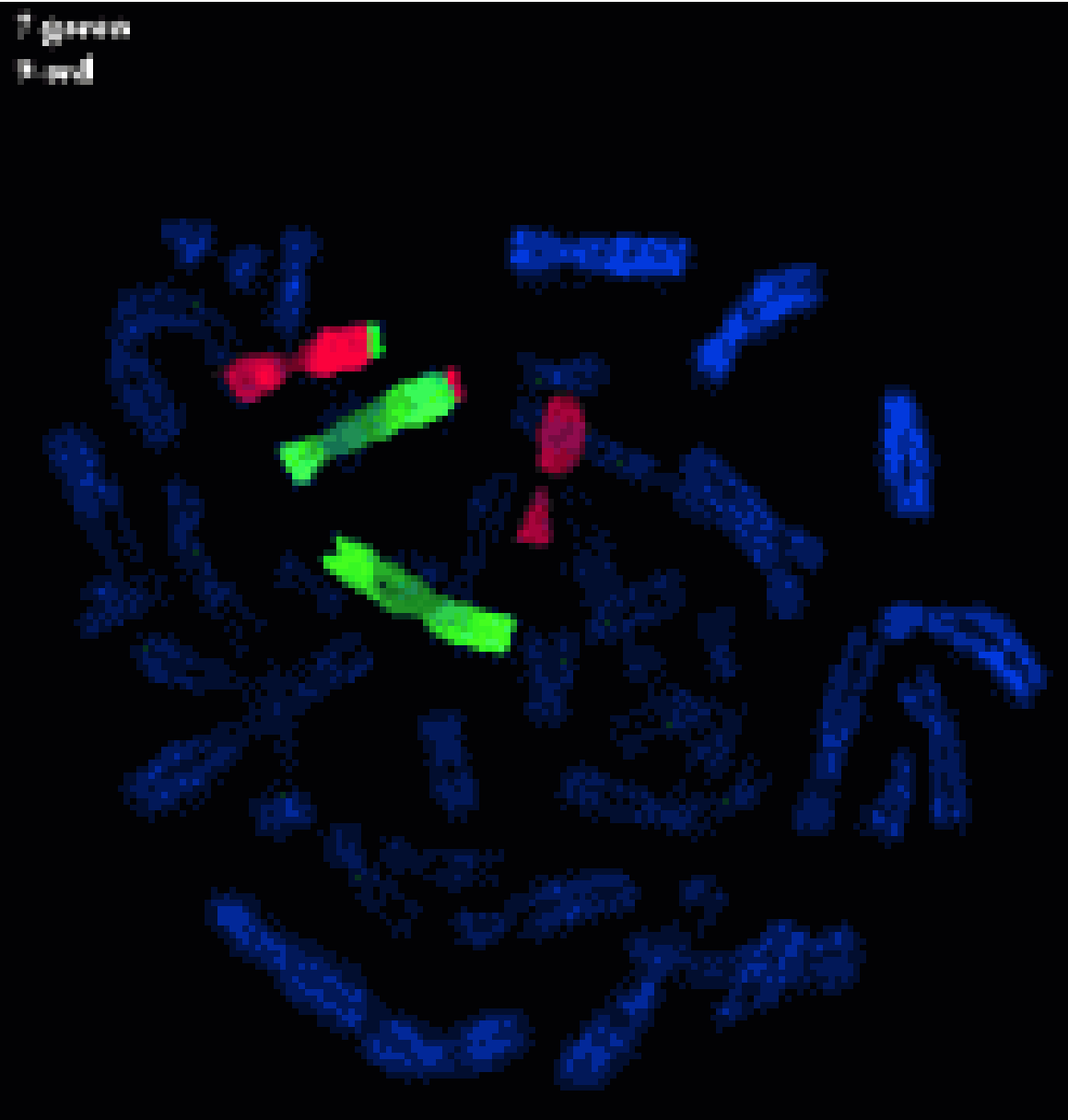


相互易位



罗伯逊易位

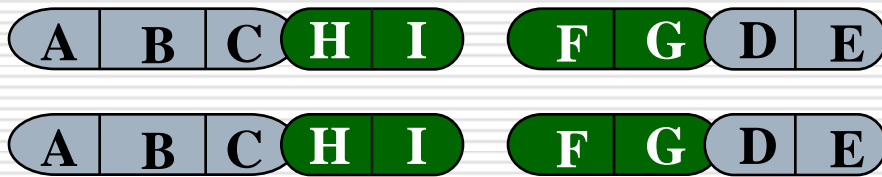
Figure 1  
Panel 1



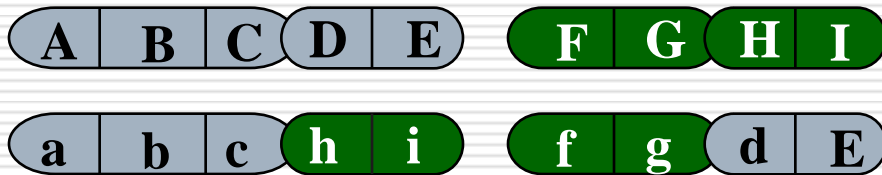
易位区段



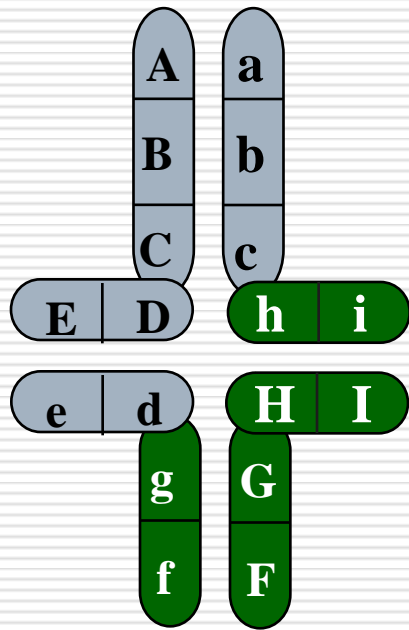
## 二、易位的减数分裂行为



易位纯合体



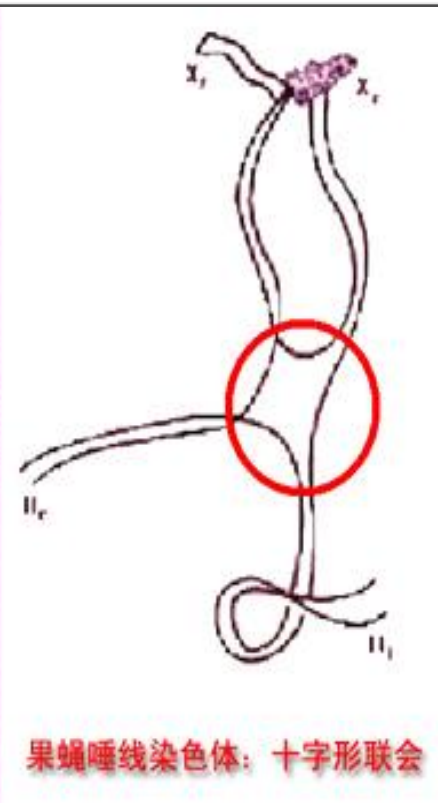
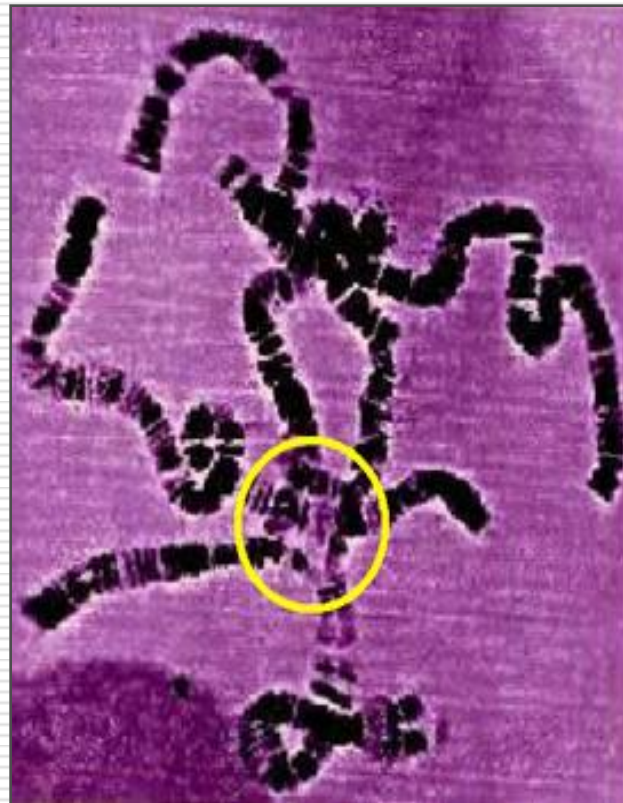
易位杂合体



四重体

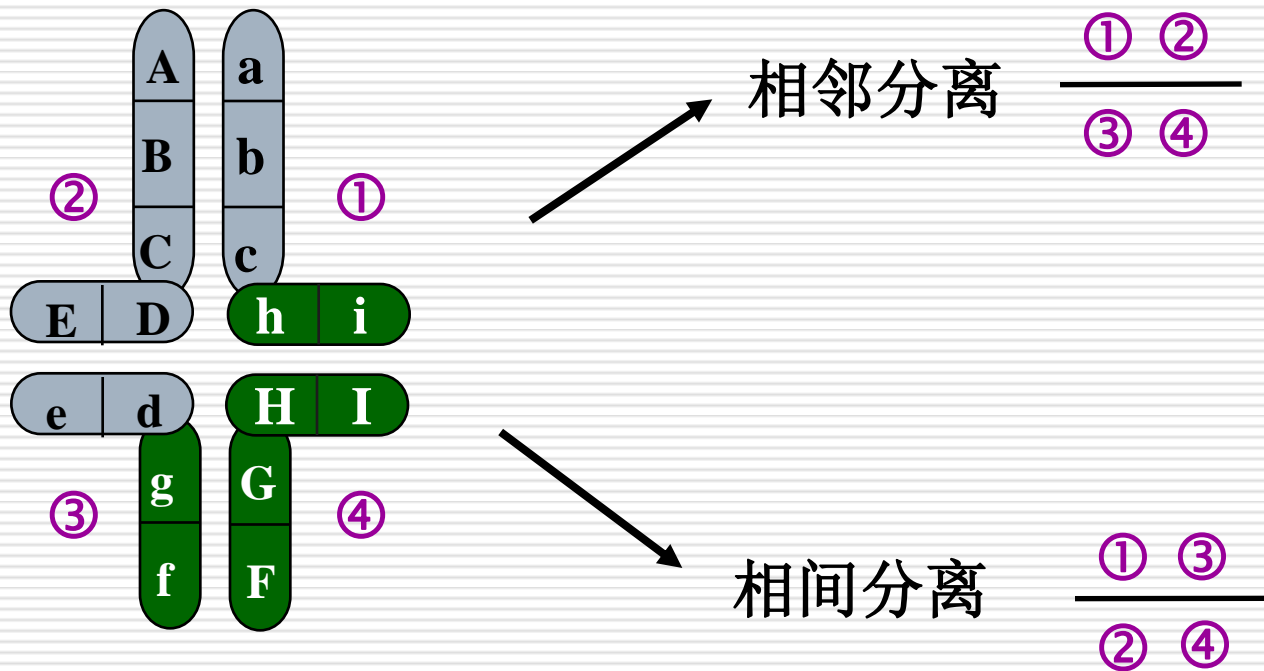
形成“+”结构

减数分裂粗线期



果蝇唾线染色体：十字形联会

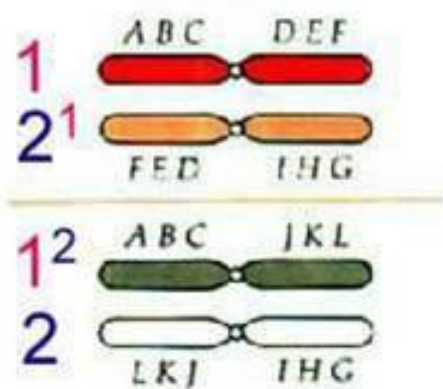
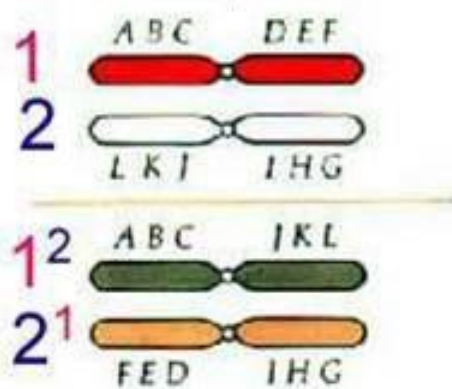
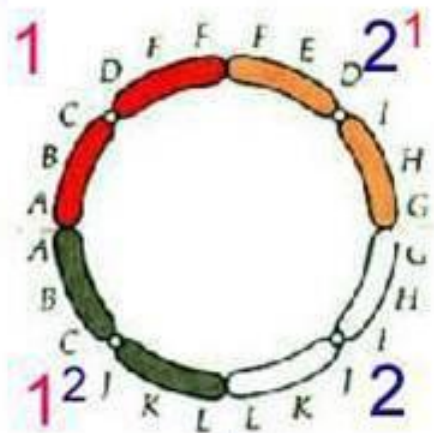
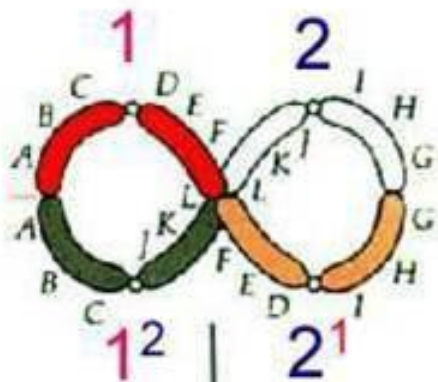
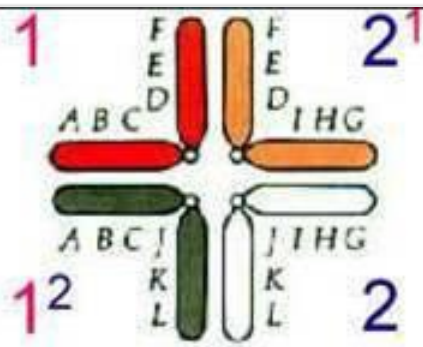


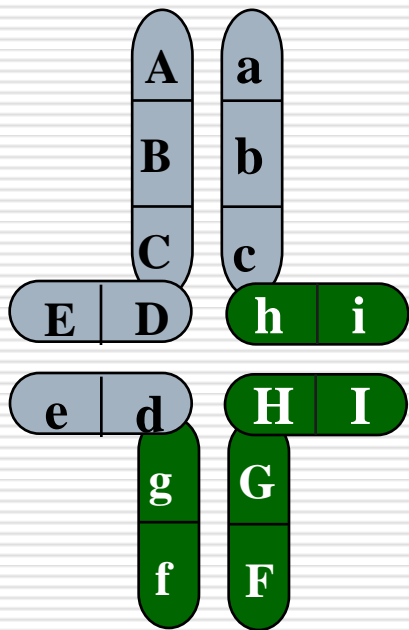



减数分裂前期

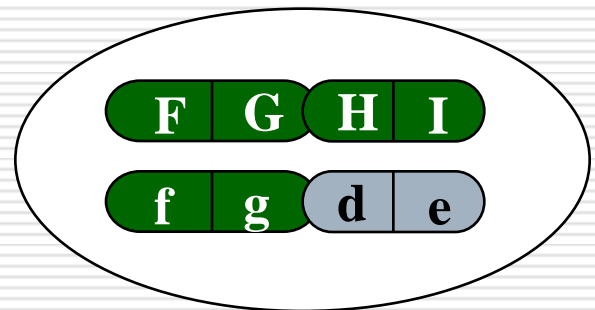
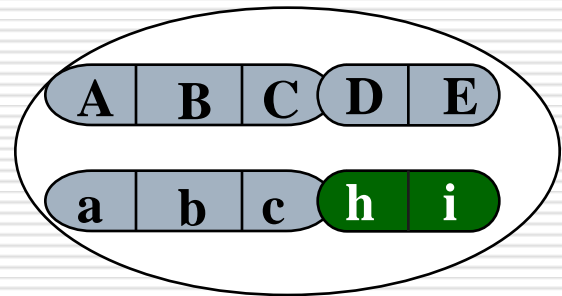


减数分裂后期



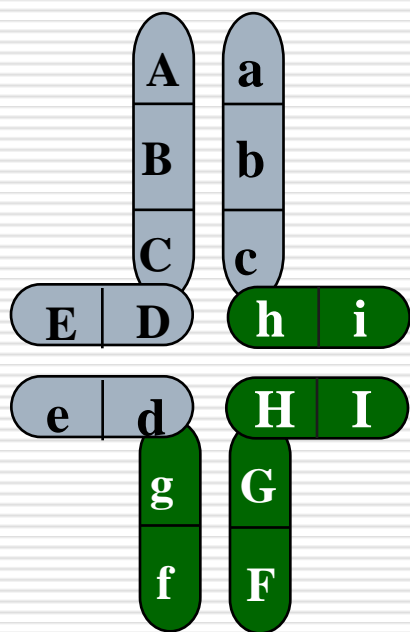


细胞分裂  
  
 配子形成

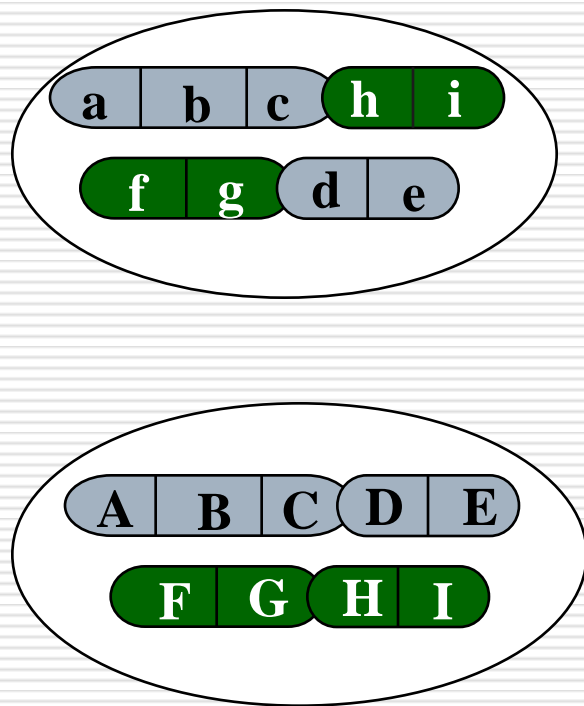


不可育

相邻分离(Adjacent segregation)



细胞分裂  
 配子形成



可育

相间分离(alternate segregation)



# 三、易位的遗传学效应

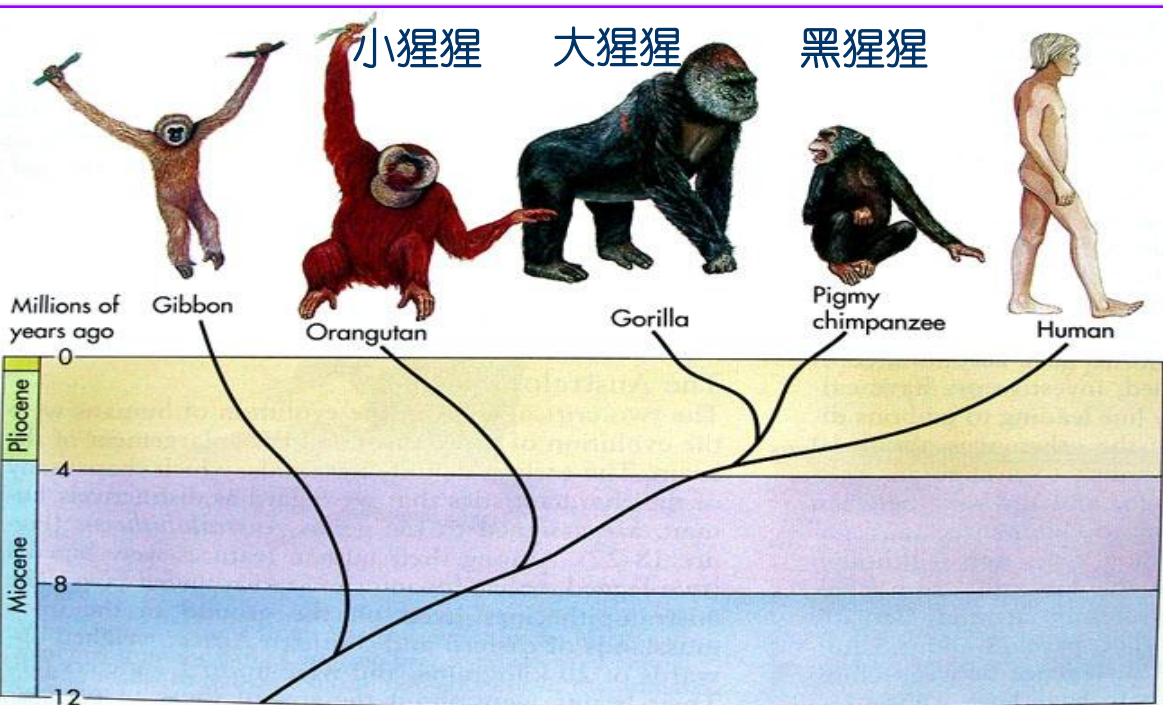
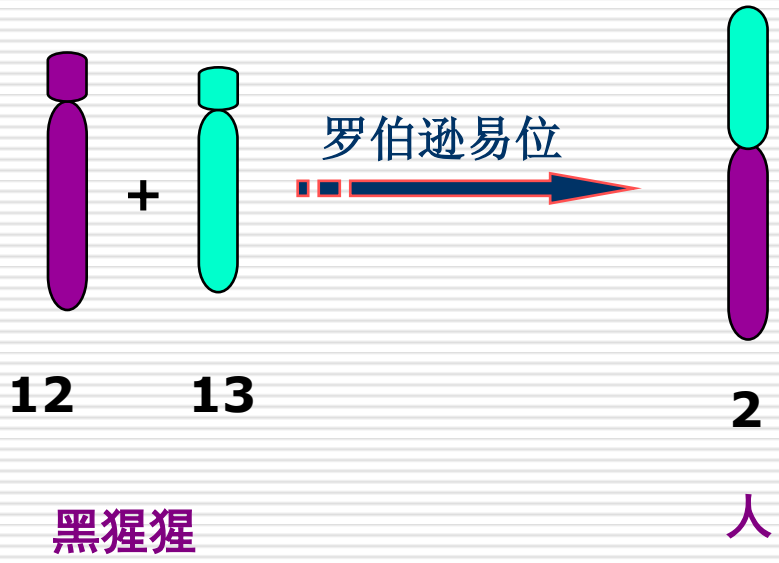
◆ 半不育 (semisterility)

◆ 改变连锁关系

◆ 易位与进化



# 易位与进化



# 四、易位与人类疾病

不育

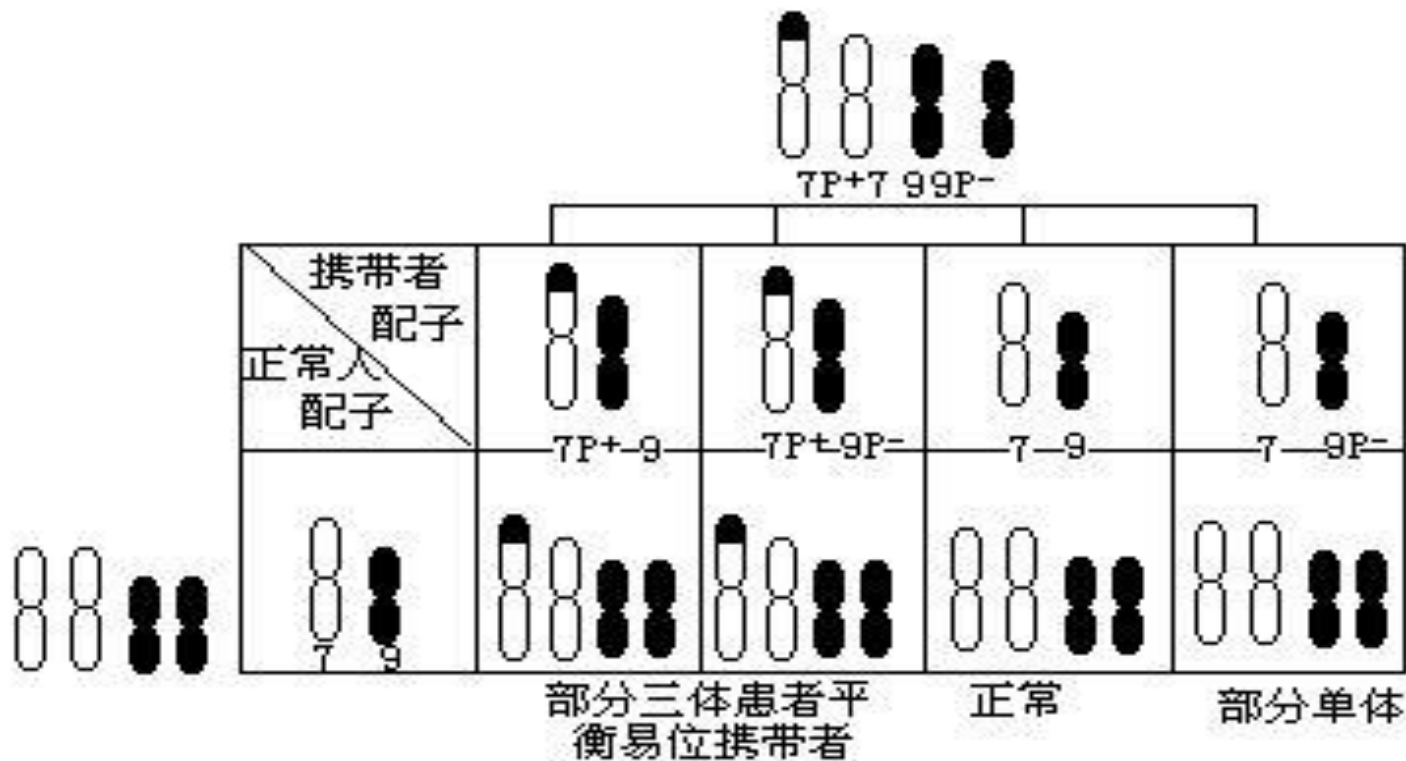
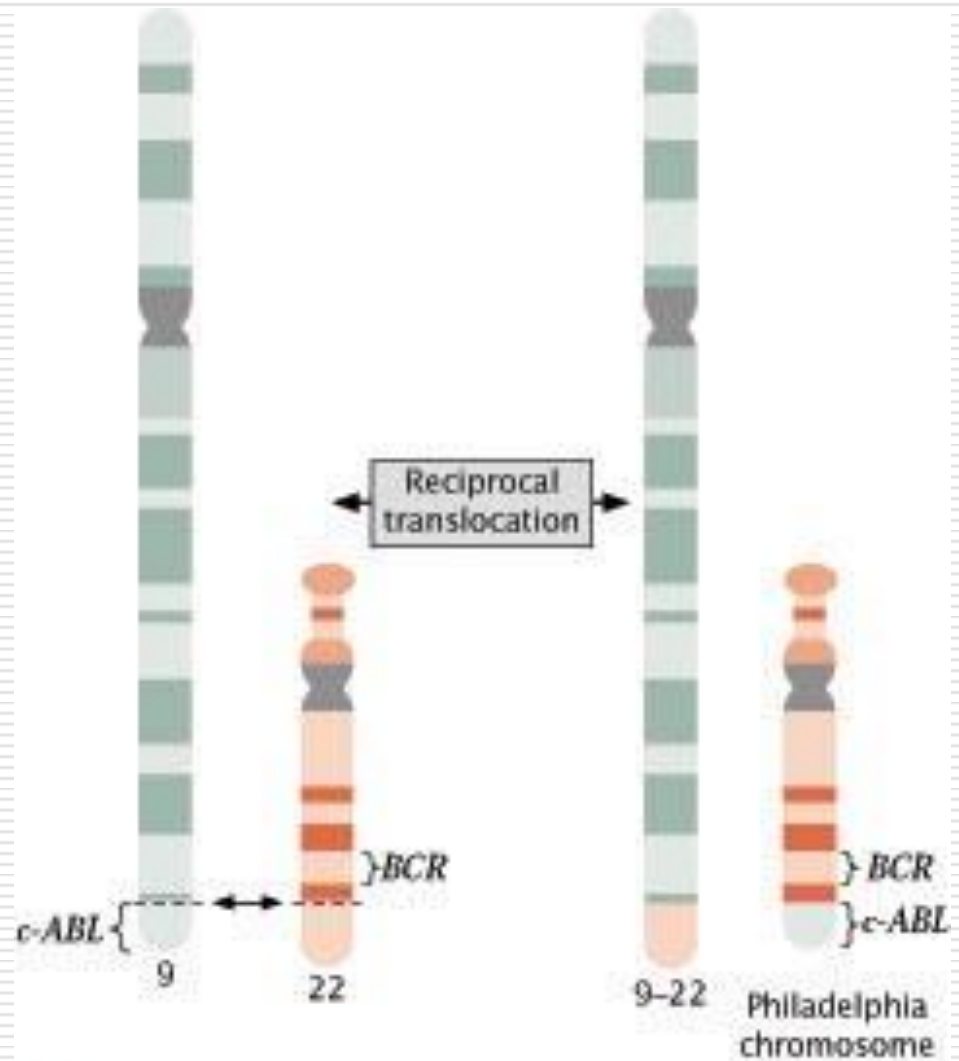


图9-13 表型正常的7/9易位杂合体可能形成的4型配子。这4型配子与正常配子结合，产生的子代可能4种类型

## 基因异常表达

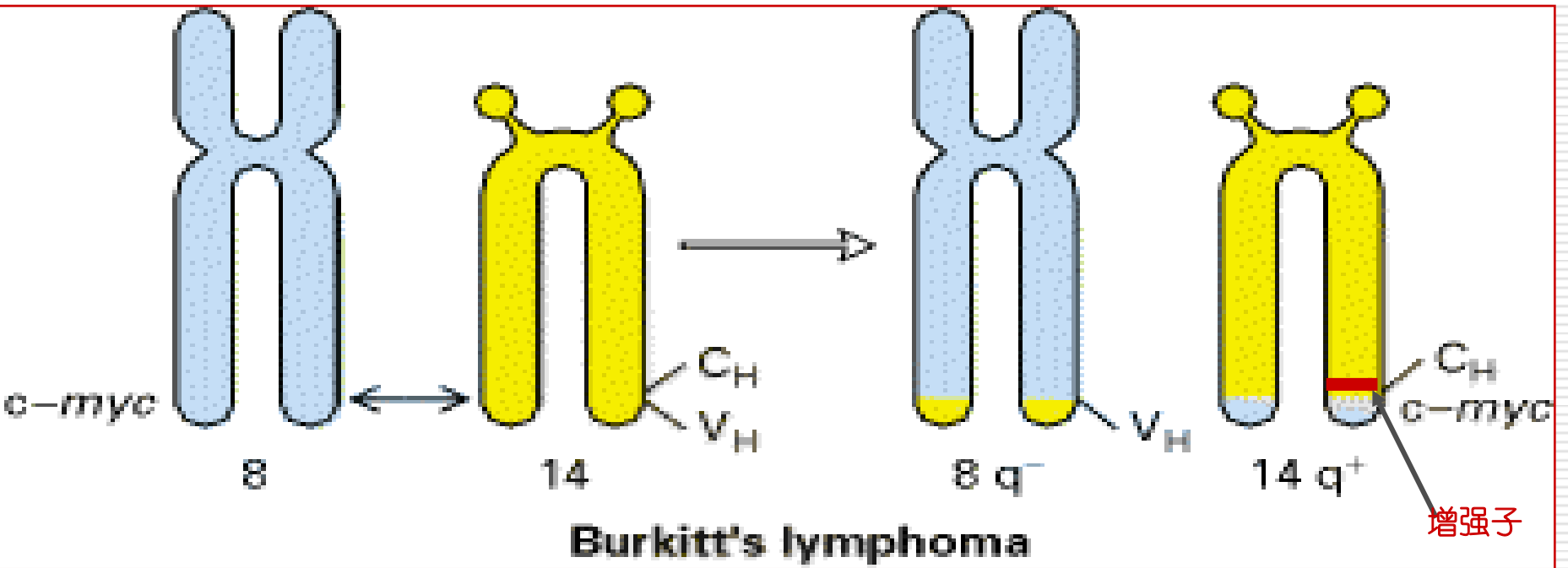
Burkitt's 淋巴瘤

慢性粒细胞白血病

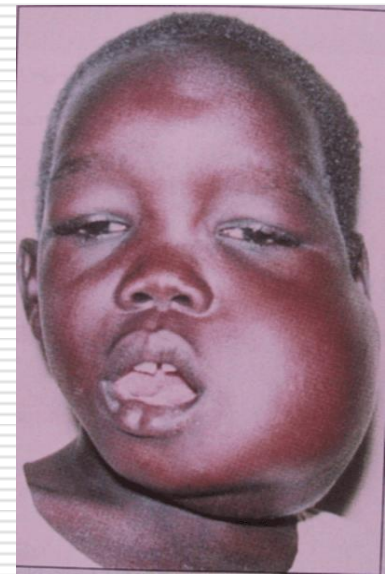


9.31 A reciprocal translocation between chromosomes 9 and 22 causes chronic myeloid leukemia.





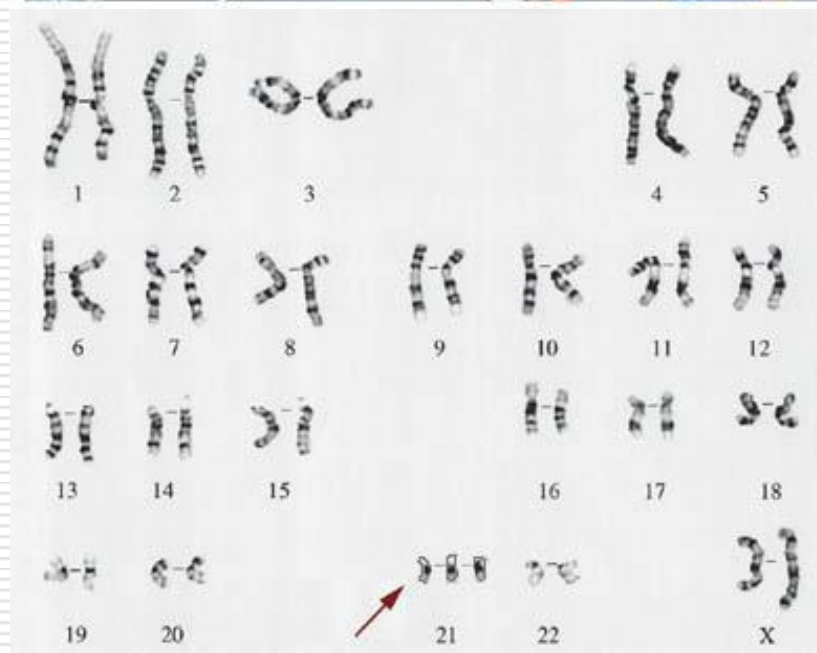
Burkitt's 淋巴瘤中8和14易位, 使*c-myc* 基因被置于抗体基因的增强子附近

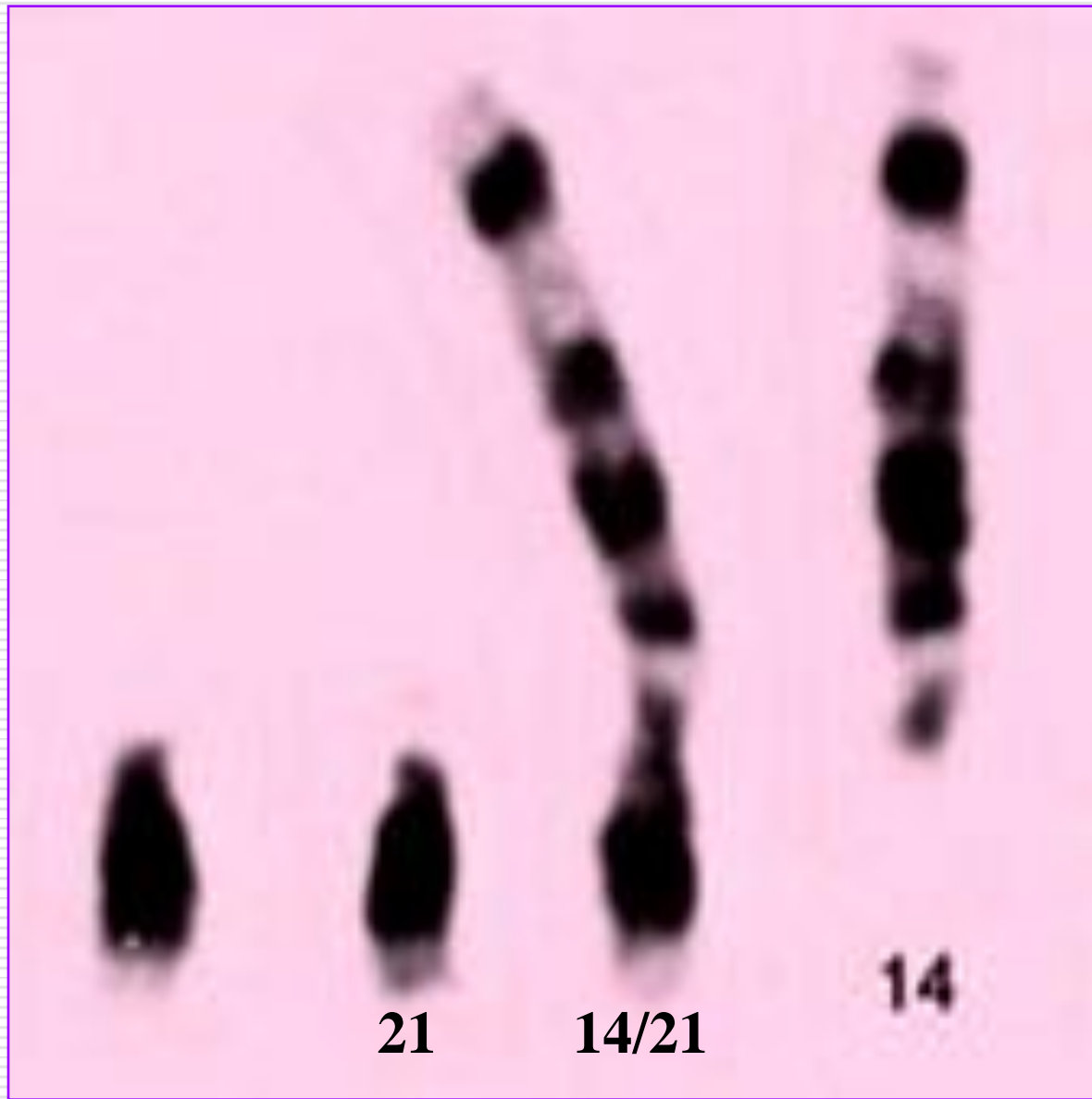


# 家族性染色体病



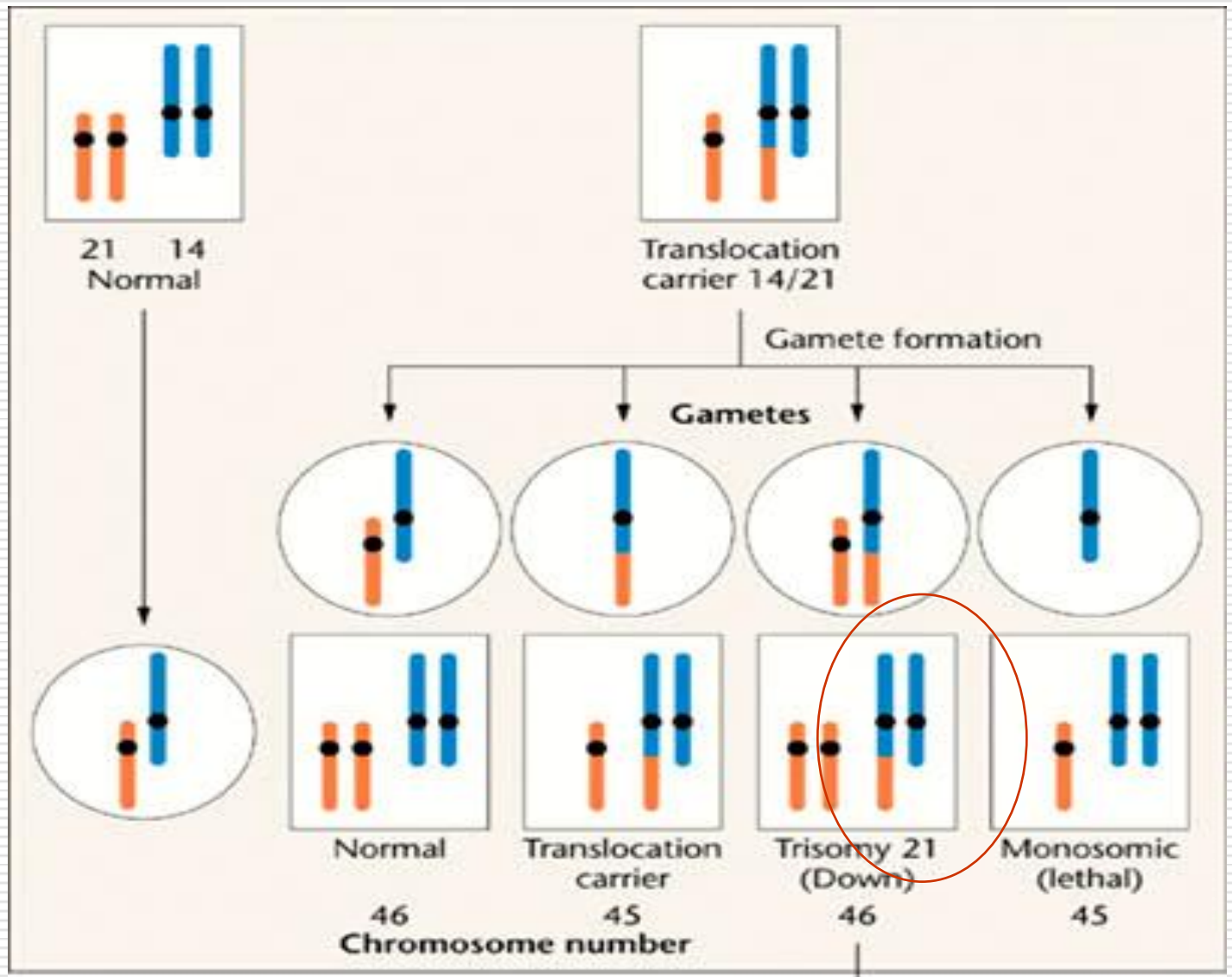
先天愚型  
(Down syndrome)





人类的14/21染色体罗伯逊易位型21三体

# Familial Down syndrome



易位型先天愚型疾病形成过程

谢谢!

下课了

