# 现代生命科学导论C

# Introduction to Life Science C

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杨扬 （1～8周）

## *Lecture 9*

# 遗传学基本定律

# 从这个问题入手：什么决定表型？
（phenotype）

- 表型：具有特定基因型的个体，在一定环境条件下，所表现出来的性状特征的总和。（可遗传）

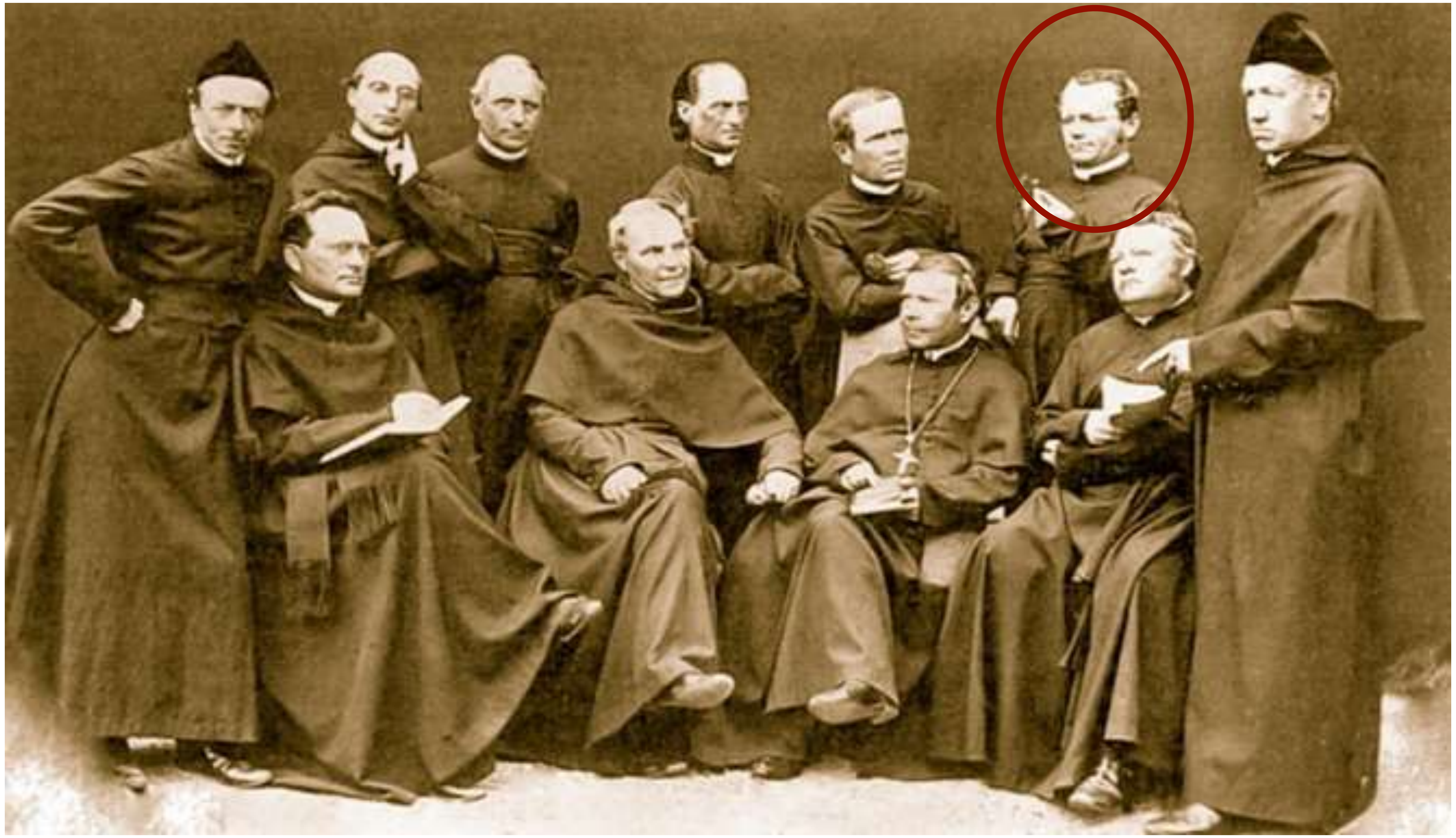
# 从这个问题入手：什么决定表型？

(phenotype)

- 孟德尔遗传学：基因和表型的关系
- 基因的连锁和交换
- 环境对表型的影响

现代遗传学之父：

Gregor Mendel （1822-1884）



# 孟德尔：一个超越时代的天才

Abbey of St Thomas in Brno

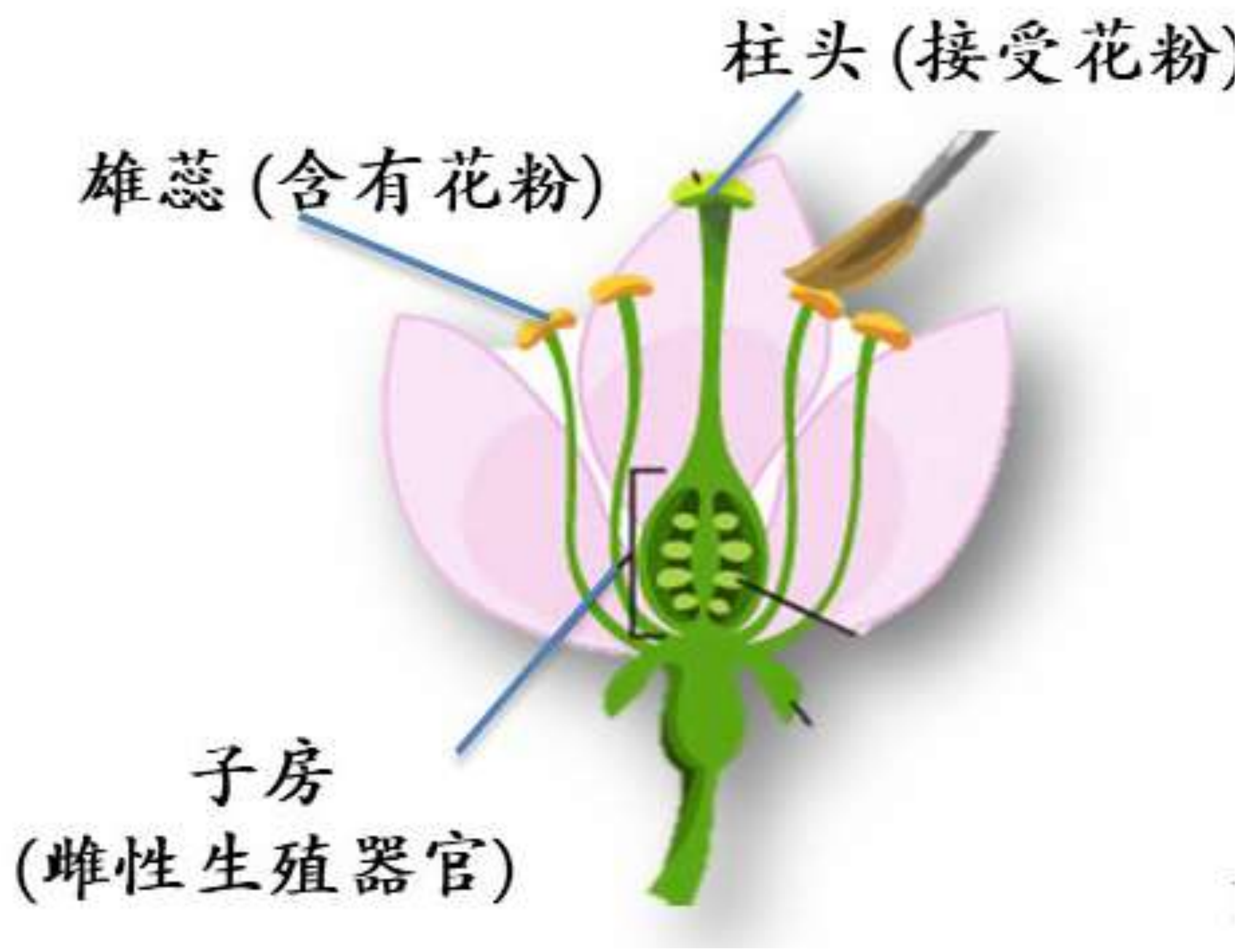
# 孟德尔实验　时代背景


奥匈帝国：纺织业，羊毛工业，以及大力发展动植物育种

维也纳大学：物理学，数学，植物学

# 孟德尔的实验

在众多植物中孟德尔为什么要研究豌豆?



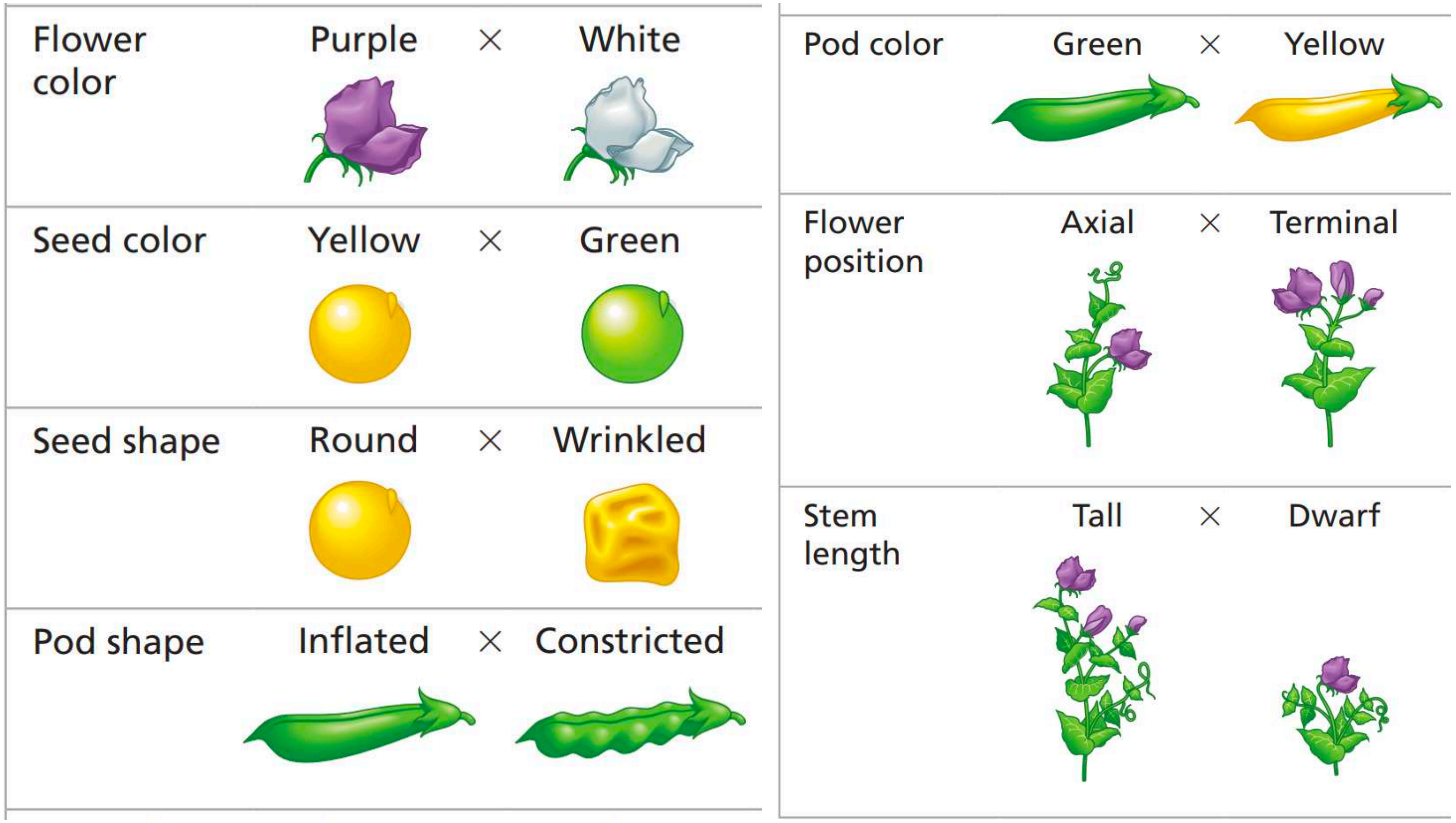

- 自花且闭花授粉，可控
- 籽粒都留在豆荚中
- 有稳定的可以区分的相对性状

(性状：生物体可以遗传的形态特征或生理特征)

# 孟德尔的实验

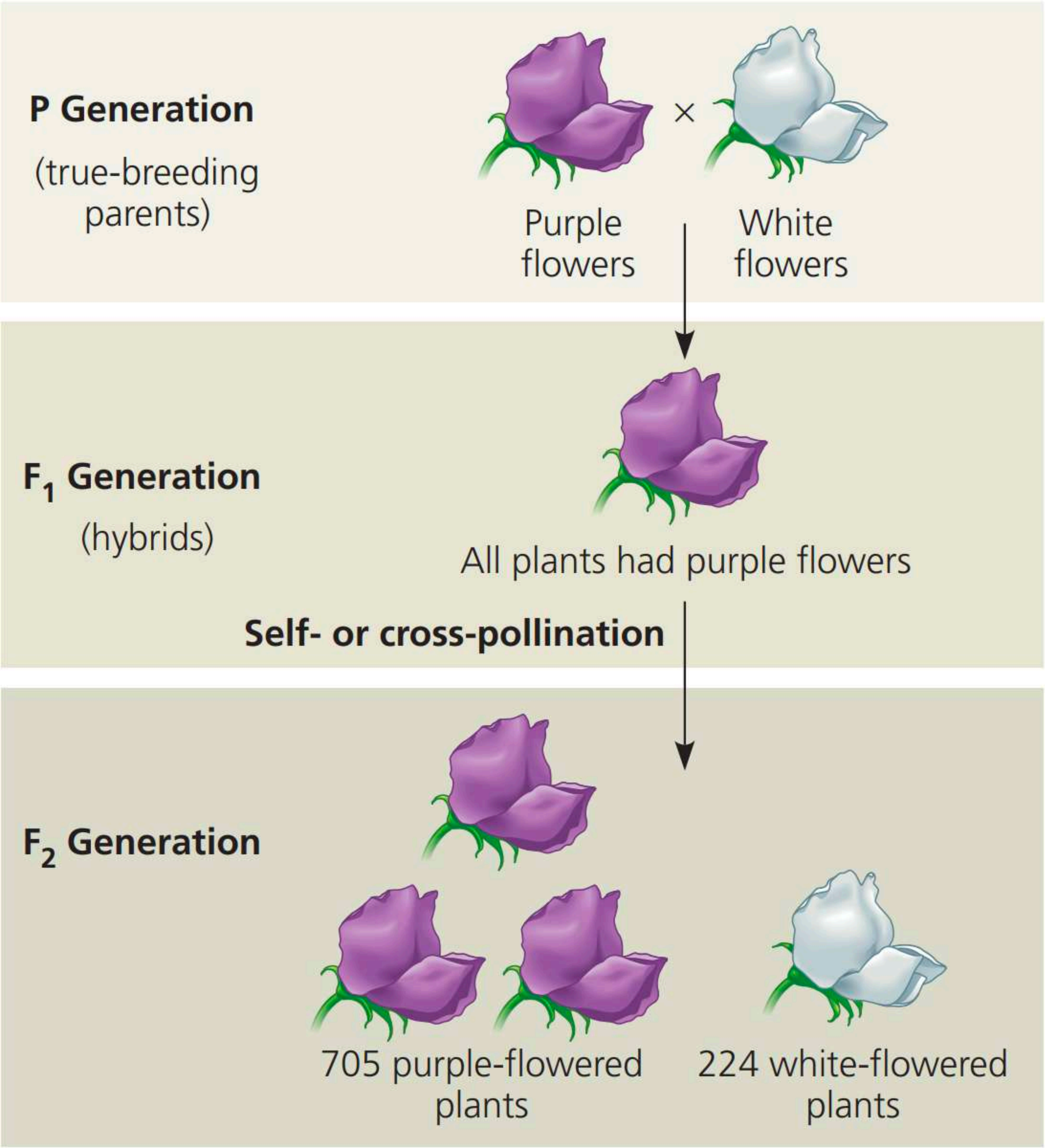
**如何得到这些性状的亲本的?**

- breeding true 纯种品系
- 建立稳定的实验系统：亲代性状必须是稳定不变的




发表的文章中讨论了7种豌豆性状

# 孟德尔的实验



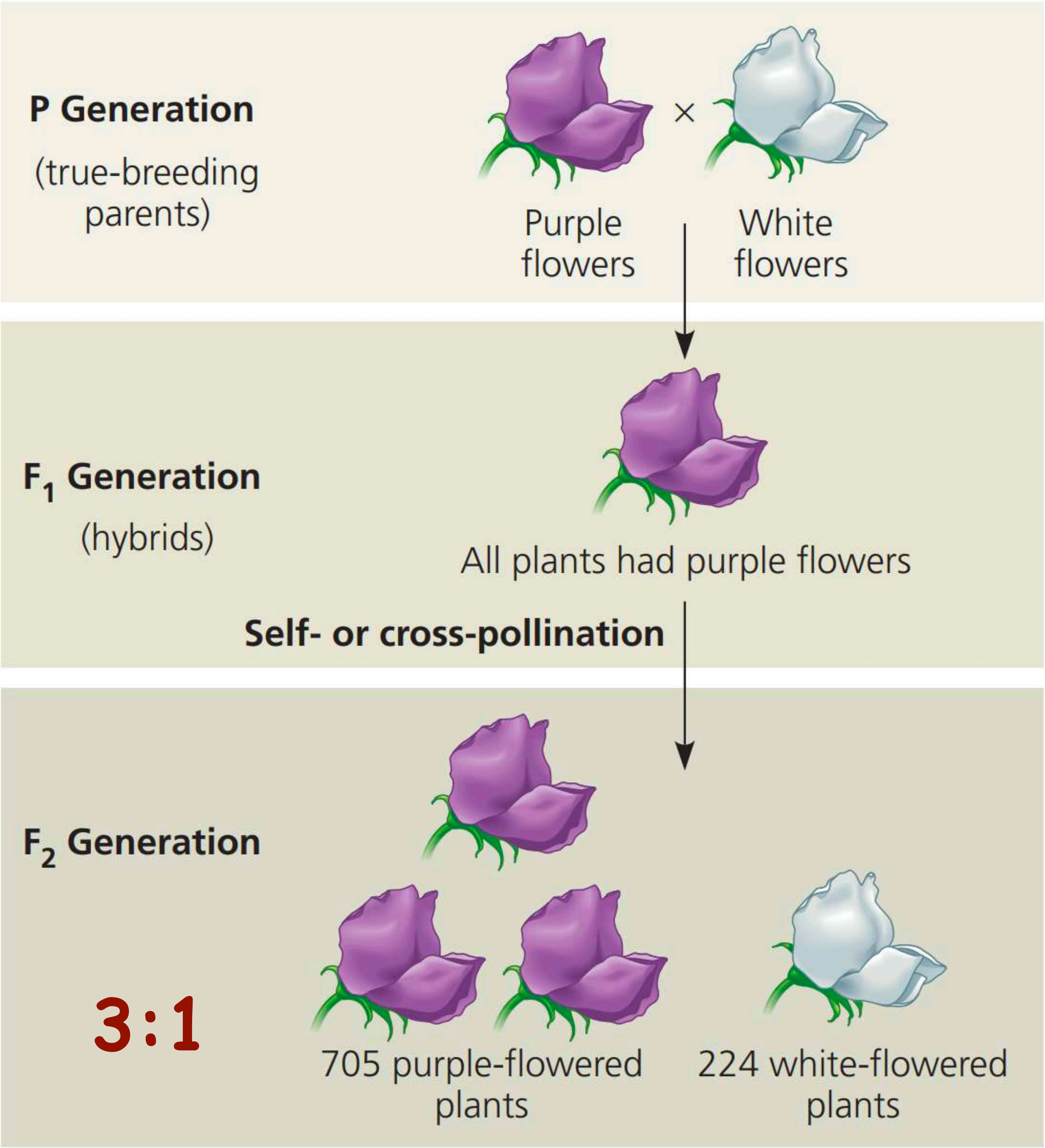

This mating, or *crossing*, of two true-breeding varieties is called **hybridization**. (杂交)

The true-breeding parents are referred to as the **P generation** (parental generation) (亲代), and their hybrid offspring are the **F1 generation** (F1代)

F1 hybrids to self-pollinate produces an **F2 generation** (F2代)

# 孟德尔的实验

## 豌豆性状有显性和隐性之分




Purple flower—— dominant (显性)

White flower—— recessive (隐性)

**Conclusion:** *The "heritable factor"(可遗传因子) for the recessive trait (white flowers) had not been destroyed, deleted, or "blended" in the F1 generation but was merely masked by the presence of the factor for purple flowers, which is the dominant trait.*

# 孟德尔的实验

## F2代性状分离时显性隐性后代之比为3：1

**Table 14.1 The Results of Mendel's $F_1$ Crosses for Seven Characters in Pea Plants**

| Character | Dominant Trait | × | Recessive Trait | $F_2$ Generation Dominant: Recessive | Ratio |
|---|---|---|---|---|---|
| Flower color | Purple  | × | White  | 705:224 | 3.15:1 |
| Seed color | Yellow  | × | Green  | 6,022:2,001 | 3.01:1 |
| Seed shape | Round  | × | Wrinkled  | 5,474:1,850 | 2.96:1 |
| Pod shape | Inflated  | × | Constricted  | 882:299 | 2.95:1 |
| Pod color | Green  | × | Yellow  | 428:152 | 2.82:1 |
| Flower position | Axial  | × | Terminal  | 651:207 | 3.14:1 |
| Stem length | Tall  | × | Dwarf  | 787:277 | 2.84:1 |

在圆皮和皱皮的实验中，孟德尔如何得出3:1的结论？

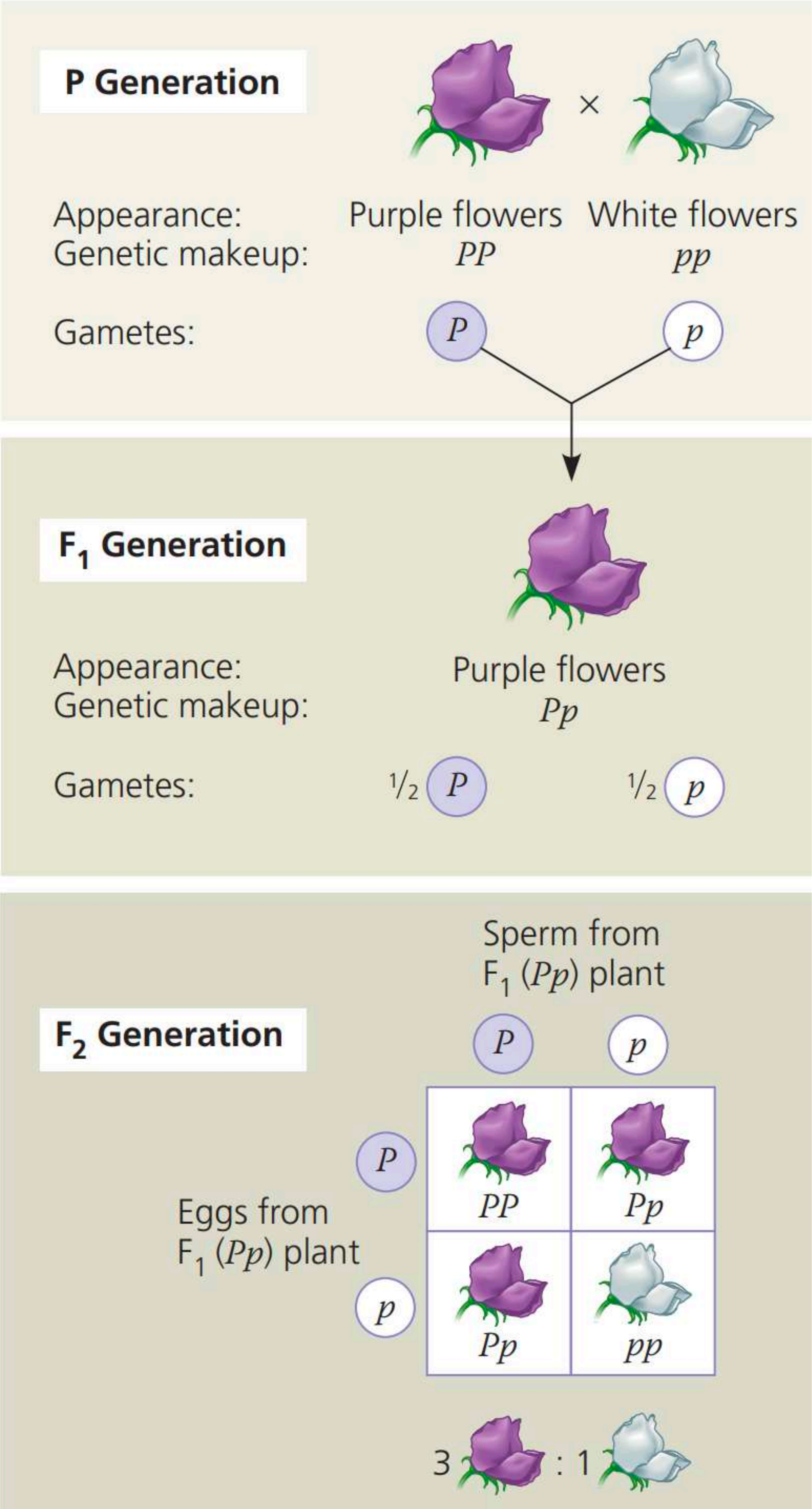
在大量的统计数据中，是否每次实验所得都符合3:1？为什么？

你们认为孟德尔是如何处理的？

在得到大量的统计数据和比例后，孟德尔接下来做的是什么？为什么要这么做？

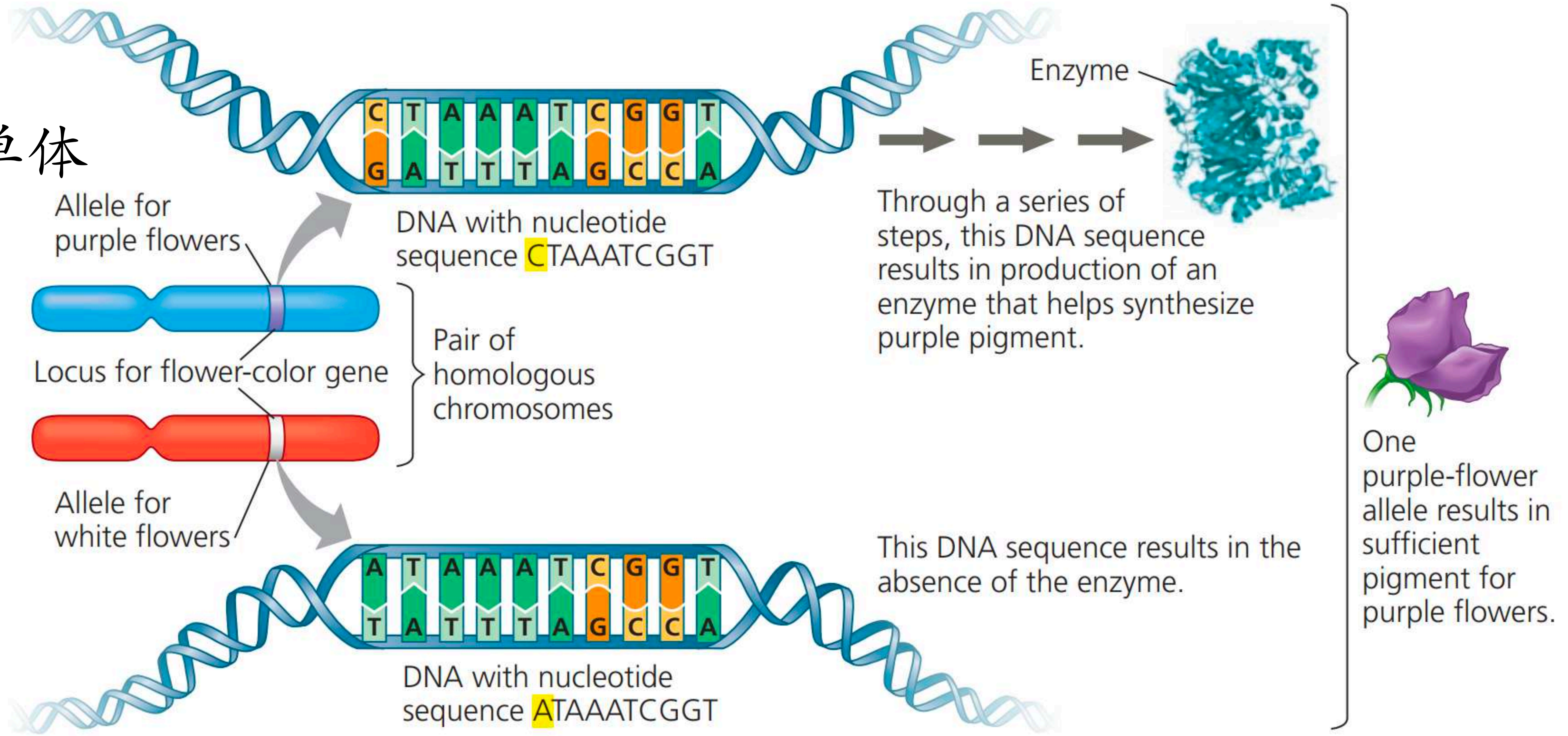
# 孟德尔 建立模型

1. 决定种皮特征的遗传因子有2个不同的版本：R和r，R决定圆皮特征，r决定皱皮特征
2. 每个遗传因子有2个拷贝（等位基因），分别来自父母双方



# Alleles 等位基因, alternative versions of a gene.

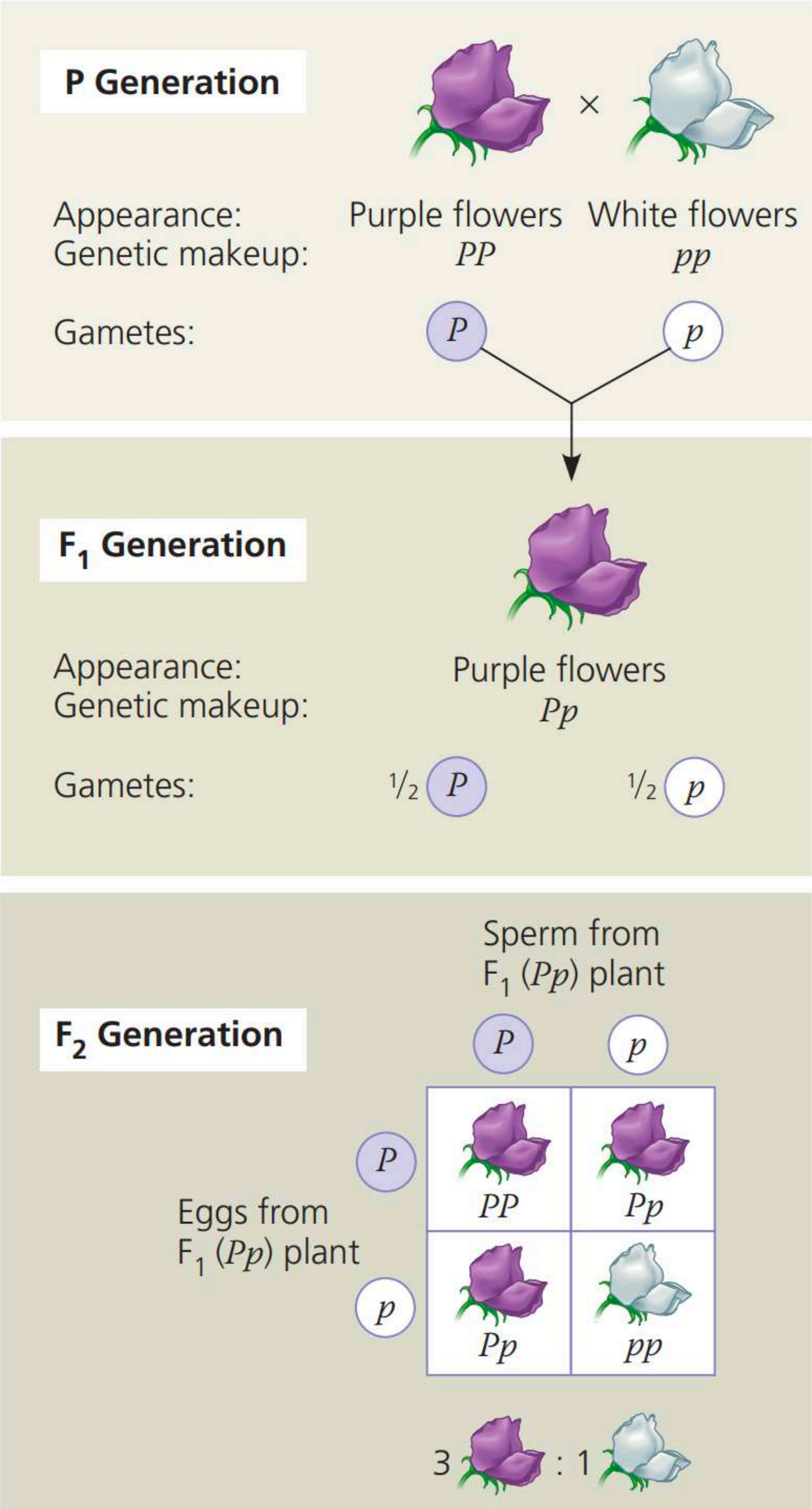
F1代的同源染色单体



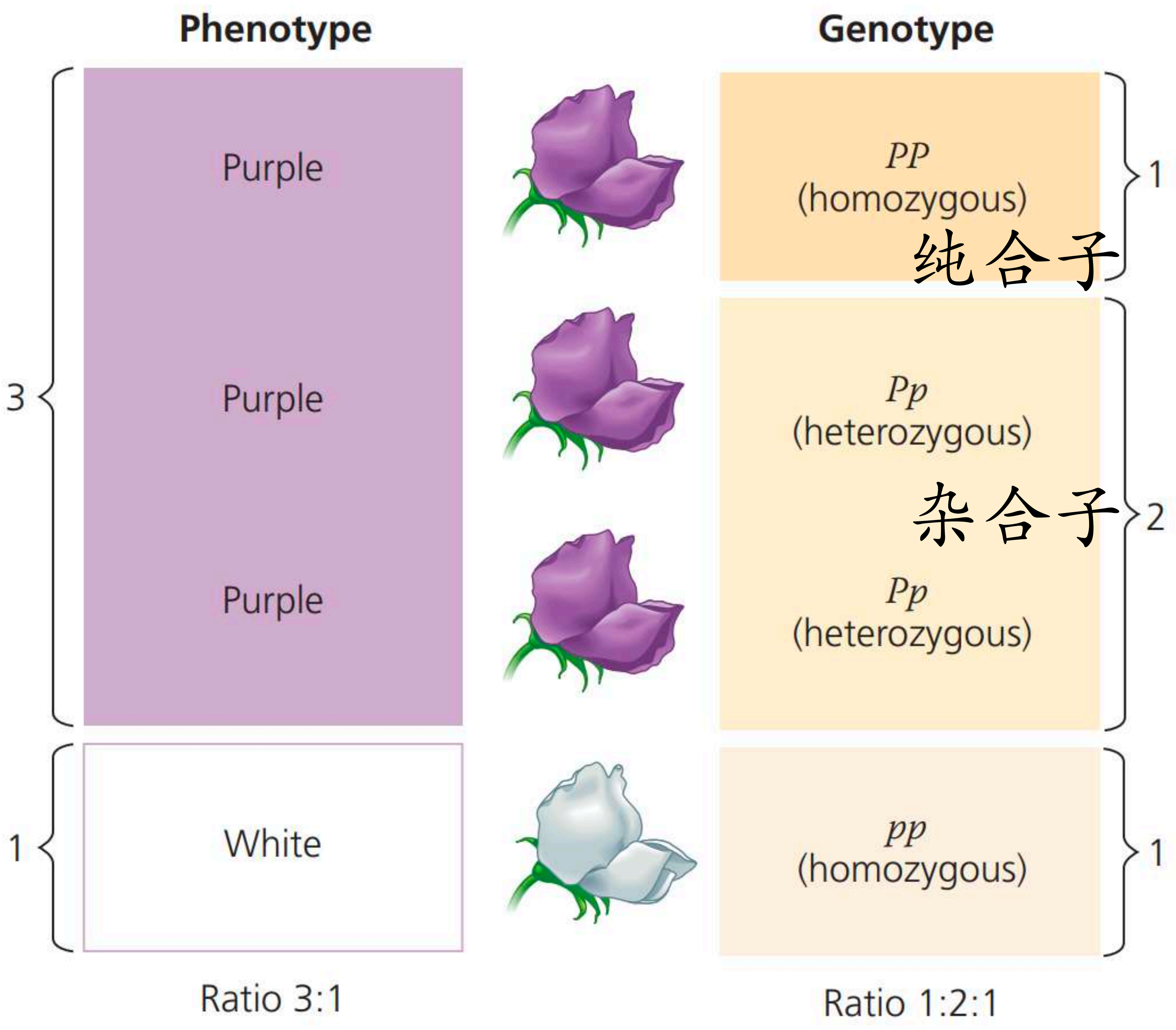
# 孟德尔 建立模型

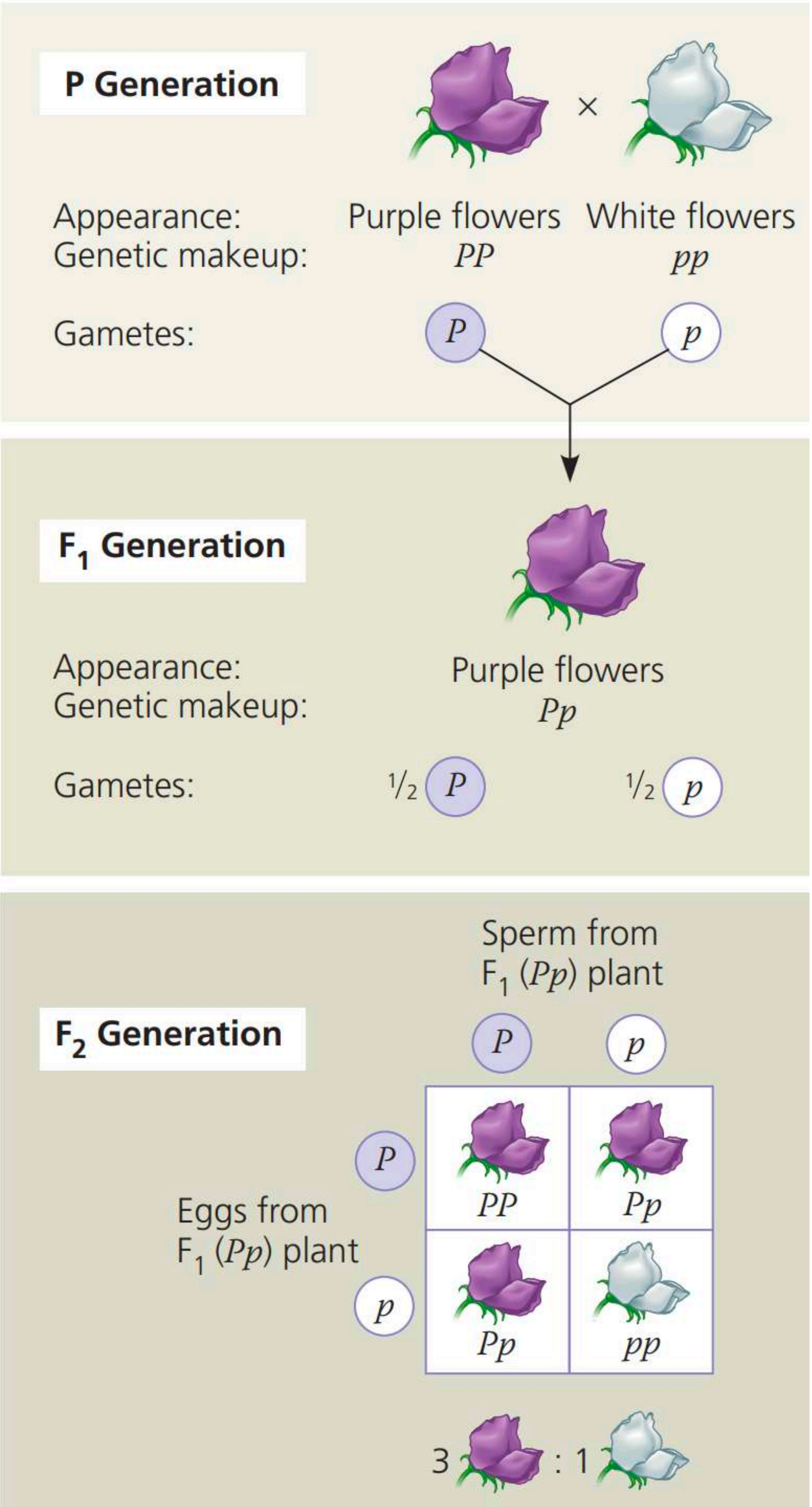
1. 决定种皮特征的遗传因子有2个不同的版本：R和r，R决定圆皮特征，r决定皱皮特征
2. 每个遗传因子有2个拷贝（等位基因），分别来自父母双方
3. 杂合体中显性的决定外在特征，而隐性的作用不明显
4. 在配子形成过程中，决定同一特征的遗传因子的两个拷贝相互分开，分别进入到不同配子中

Law of segregation 分离定律



# Phenotype (表型) versus genotype (基因型)



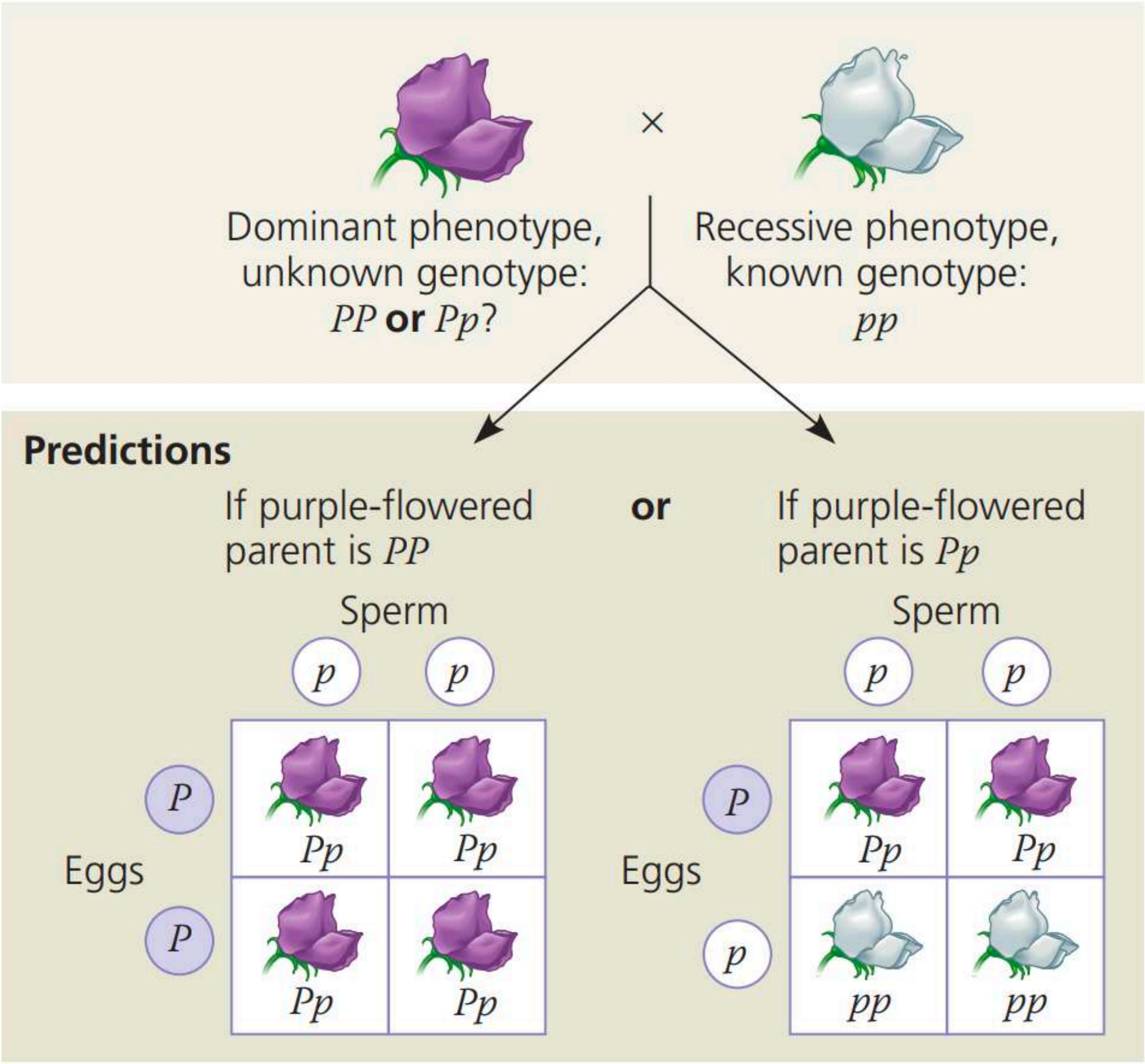




生物体的外观或可观察到的特征，称为其表型

它的基因组成—基因型

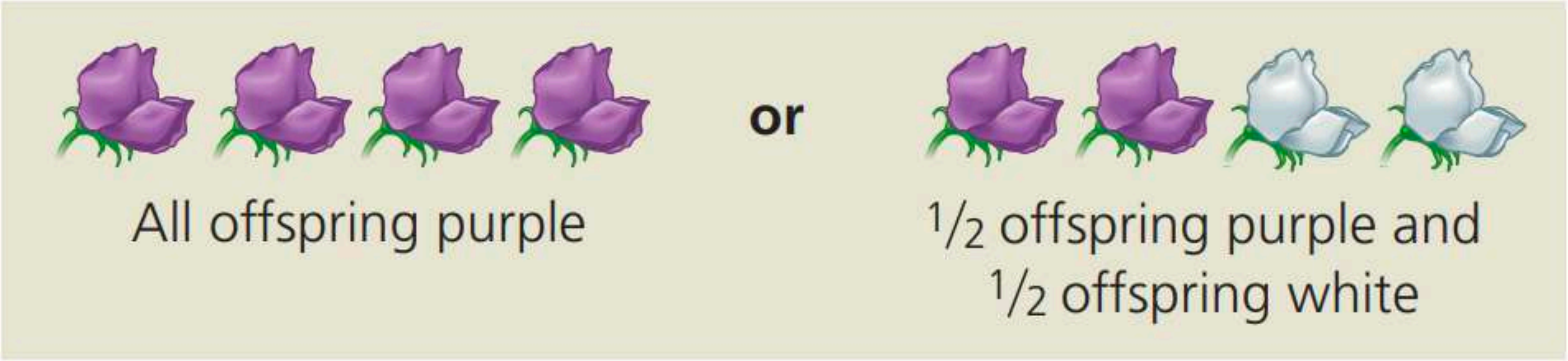
# 孟德尔 建立模型

# The testcross 测交测试




具有未知基因型的个体与表达隐性性状的纯合子个体杂交

Results



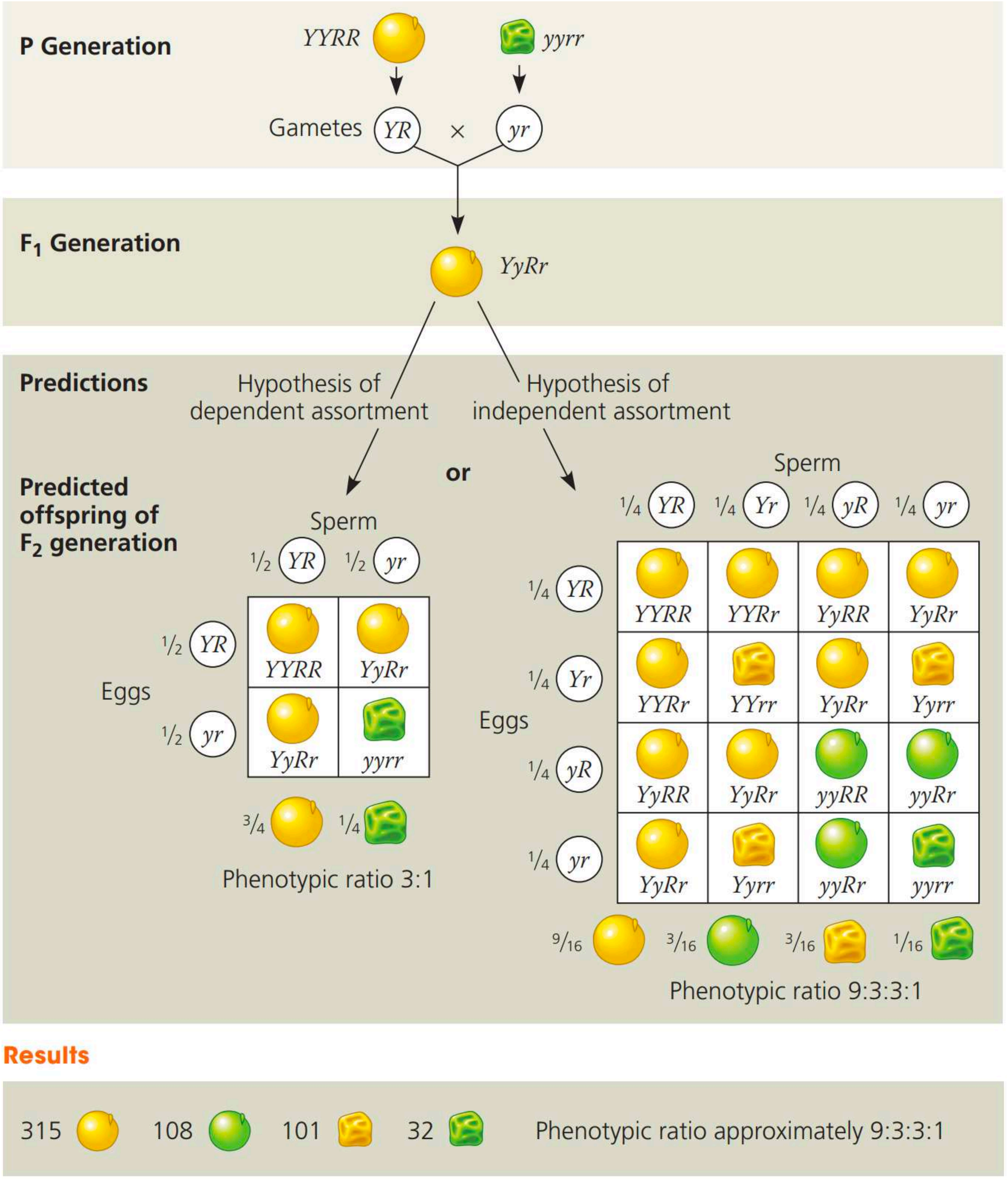
# 单个性状遗传的分离定律（遗传学第一定律）

Law of segregation

一对等位基因在杂合状态下（Aa），互不干预，在形成配子时各自（A或a）完全独立地分离到不同配子中去。

**分离定律的核心问题：等位基因的分离**

适用于：一个同源染色体上的一对等位基因




# 多个性状遗传时的自由组合定律

## Law of Independent Assortment

一个性状的等位基因是依赖还是独立于另一个性状的等位基因分离进入配子？

*Two or more genes assort independently—that is, each pair of alleles segregates independently of each other pair of alleles—during gamete (配子) formation.*

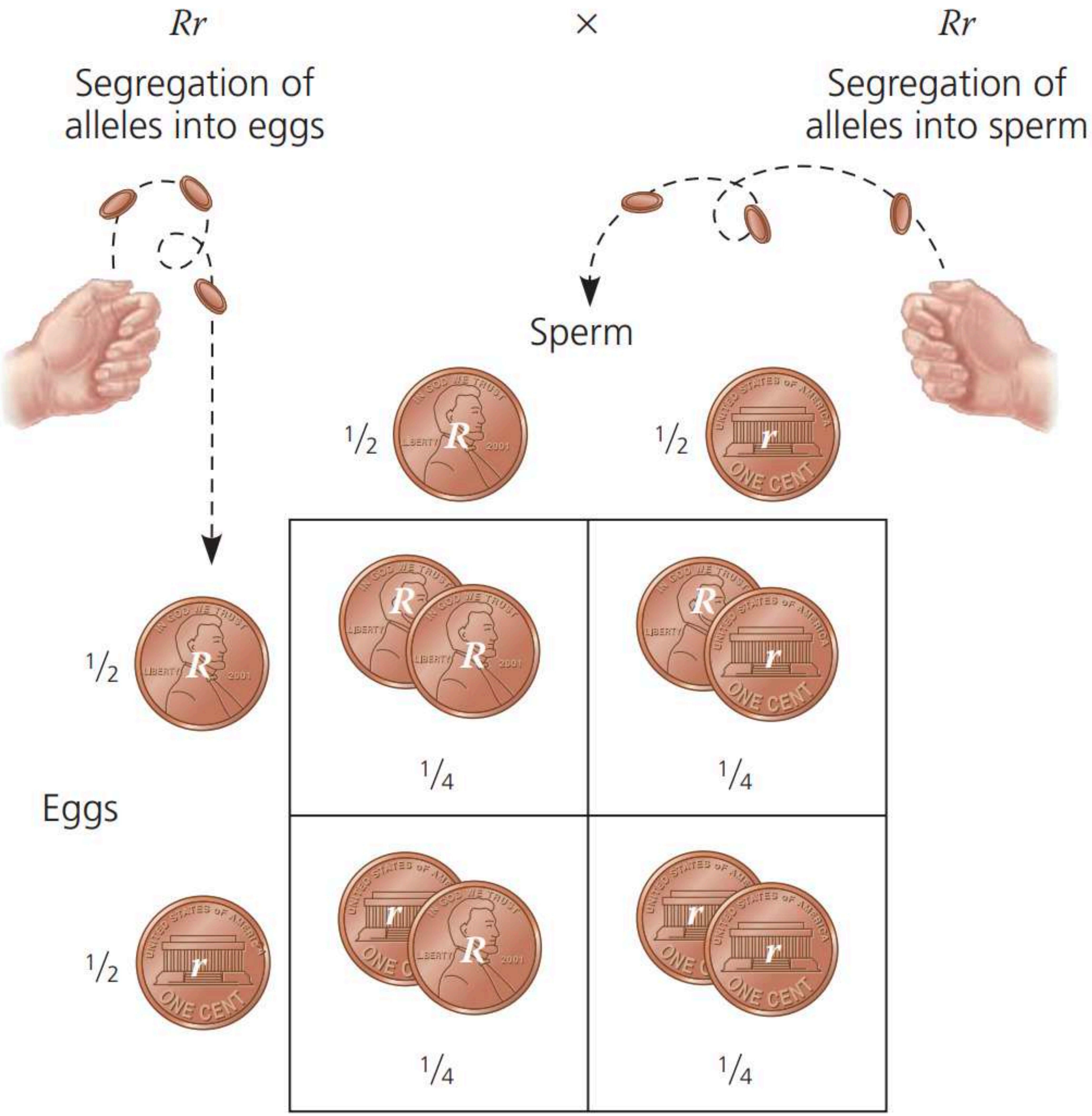
# 自由组合定律

控制两对不同性状的等位基因在配子形成过程中，一对等位基因与另一对等位基因的分离和组合互不干扰。

**自由组合的核心问题：非等位基因的自由组合**

适用于：不连锁基因。位于不同染色体上的等位基因对，
或者适用于同一染色体上相距很远的基因。

# Probability laws (概率论) govern Mendelian inheritance

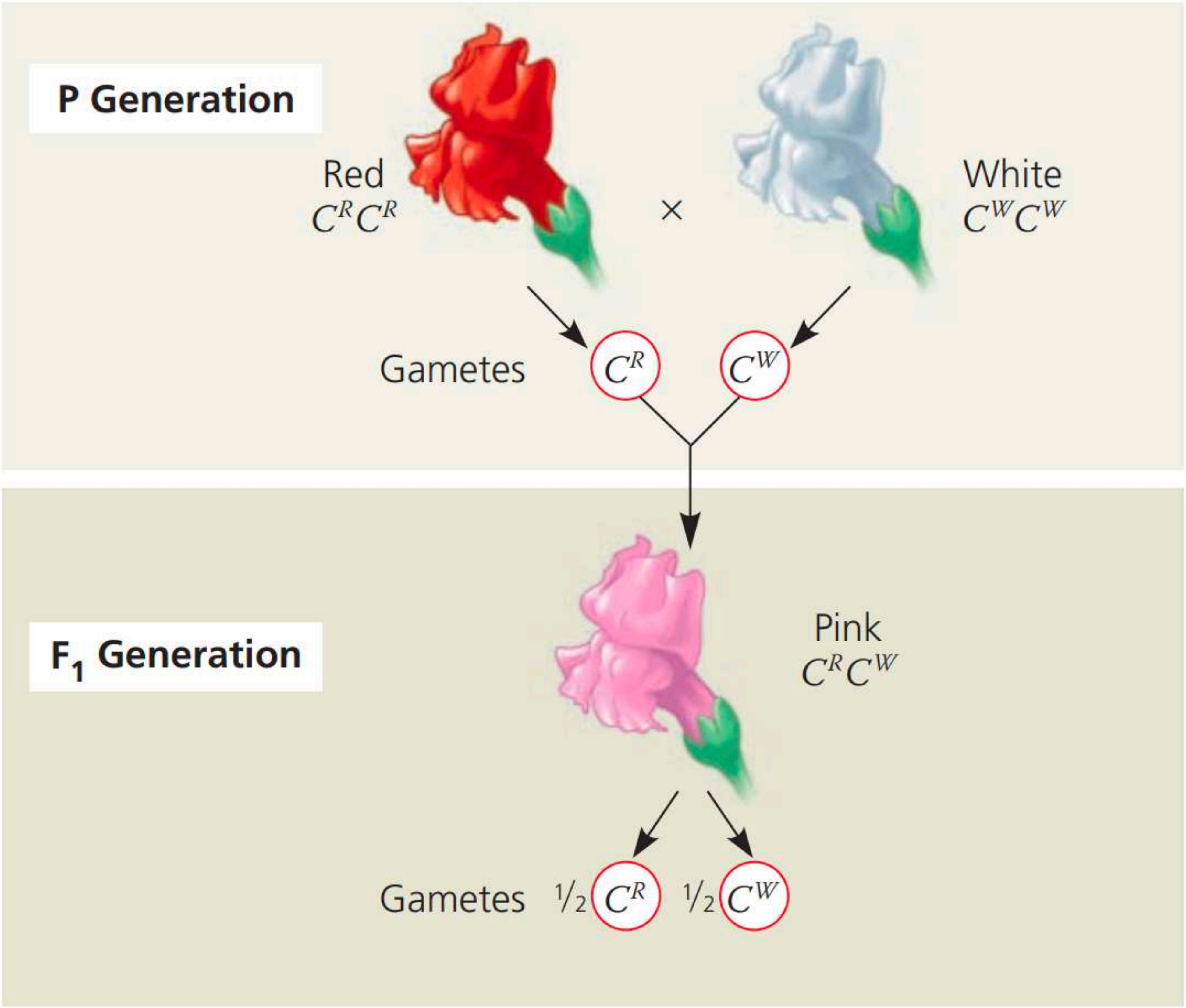


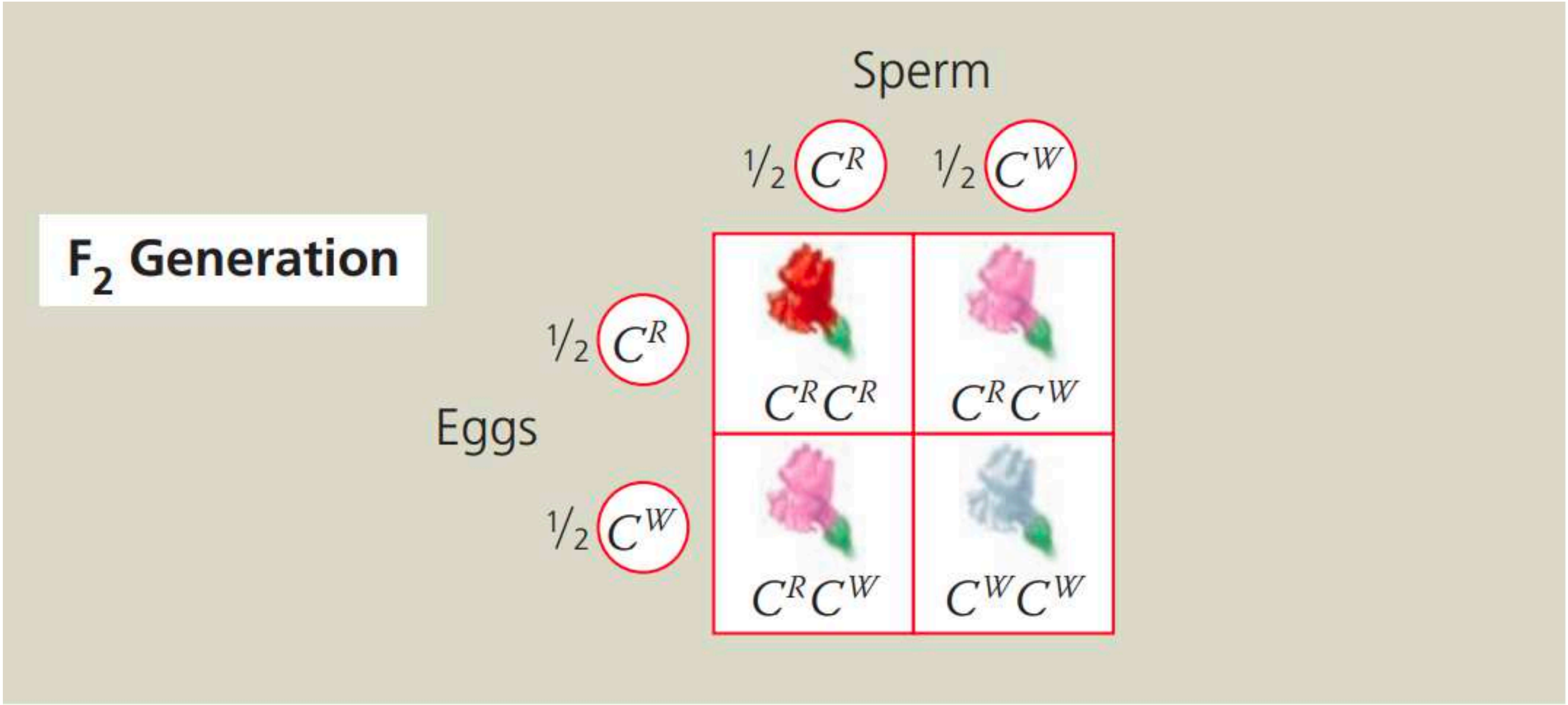

分离定律和自由组合定律：
随机事件，相同概率

应用：育种

# More complex than predicted by simple Mendelian genetics…

当等位基因不完全显性或隐性时，



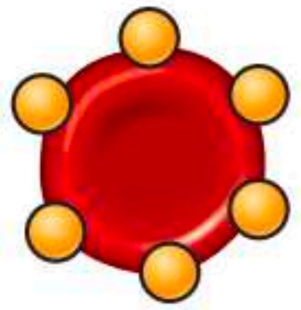

基因型/表型 1:2:1

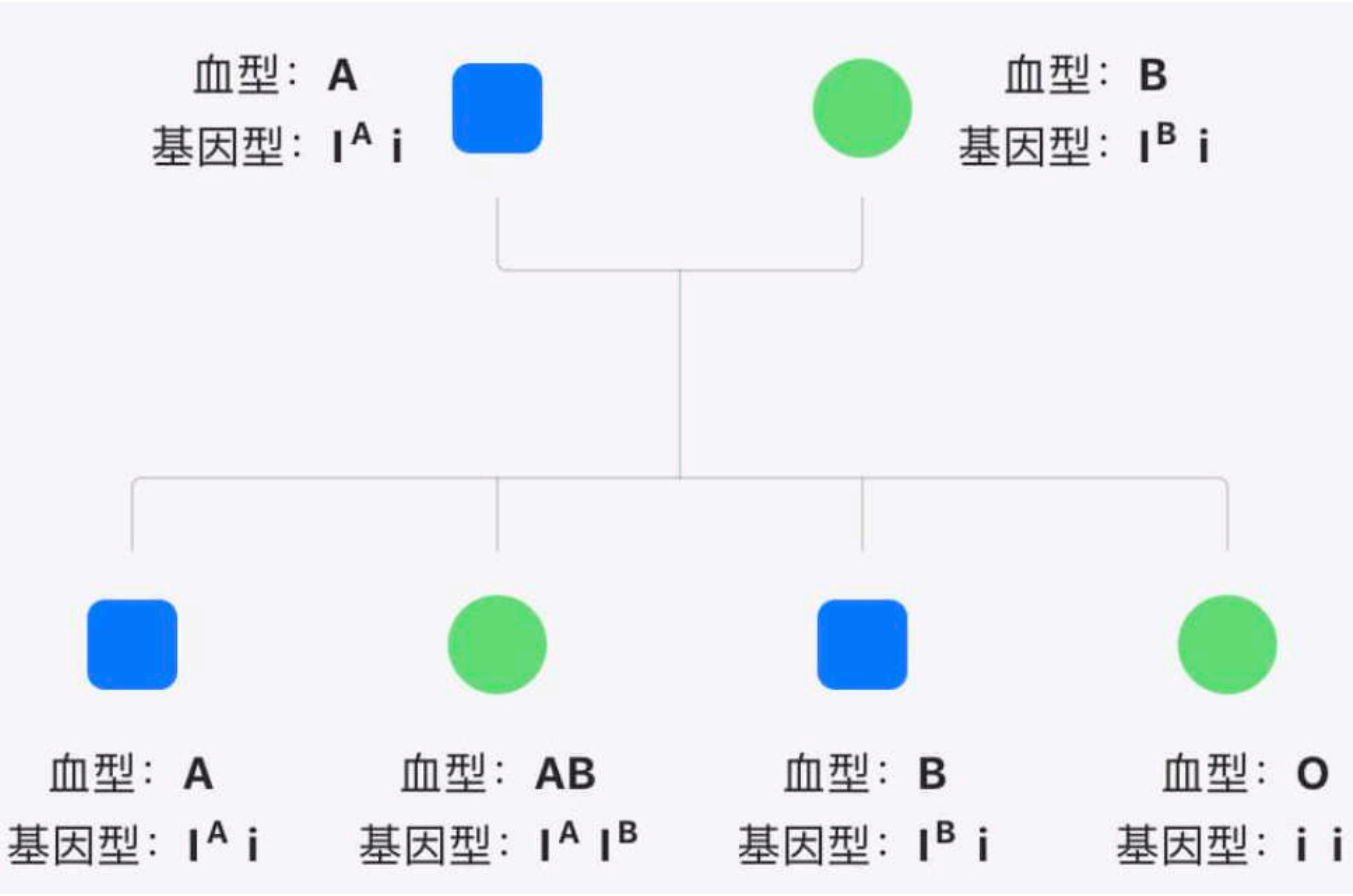
# 当特定基因具有两个以上的等位基因时，

**(a) The three alleles for the ABO blood groups and their carbohydrates.** Each allele codes for an enzyme that may add a specific carbohydrate (designated by the superscript on the allele and shown as a triangle or circle) to red blood cells.

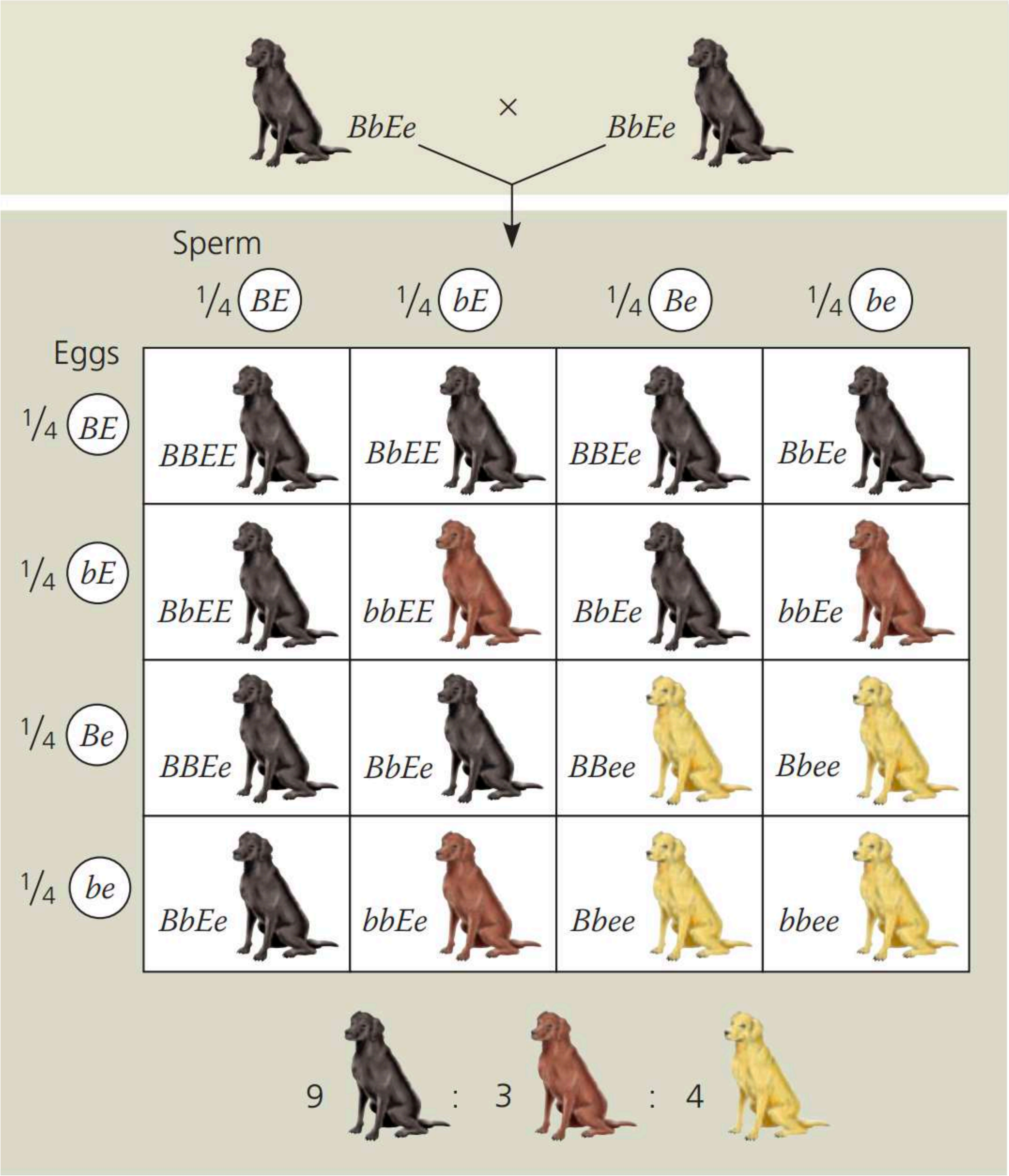
| **Allele** | $I^A$ | $I^B$ | $i$ |
|---|---|---|---|
| **Carbohydrate** | A △ | B ○ | none |

**(b) Blood group genotypes and phenotypes.** There are six possible genotypes, resulting in four different phenotypes.

| **Genotype** | $I^AI^A$ or $I^Ai$ | $I^BI^B$ or $I^Bi$ | $I^AI^B$ | $ii$ |
|---|---|---|---|---|
| **Red blood cell appearance** |  |  |  |  |
| **Phenotype (blood group)** | A | B | AB | O |



当特定基因具有两个以上的等位基因时，




Epistasis: where one gene affects the phenotype of another because the two gene products interact

B：黑色毛，显性

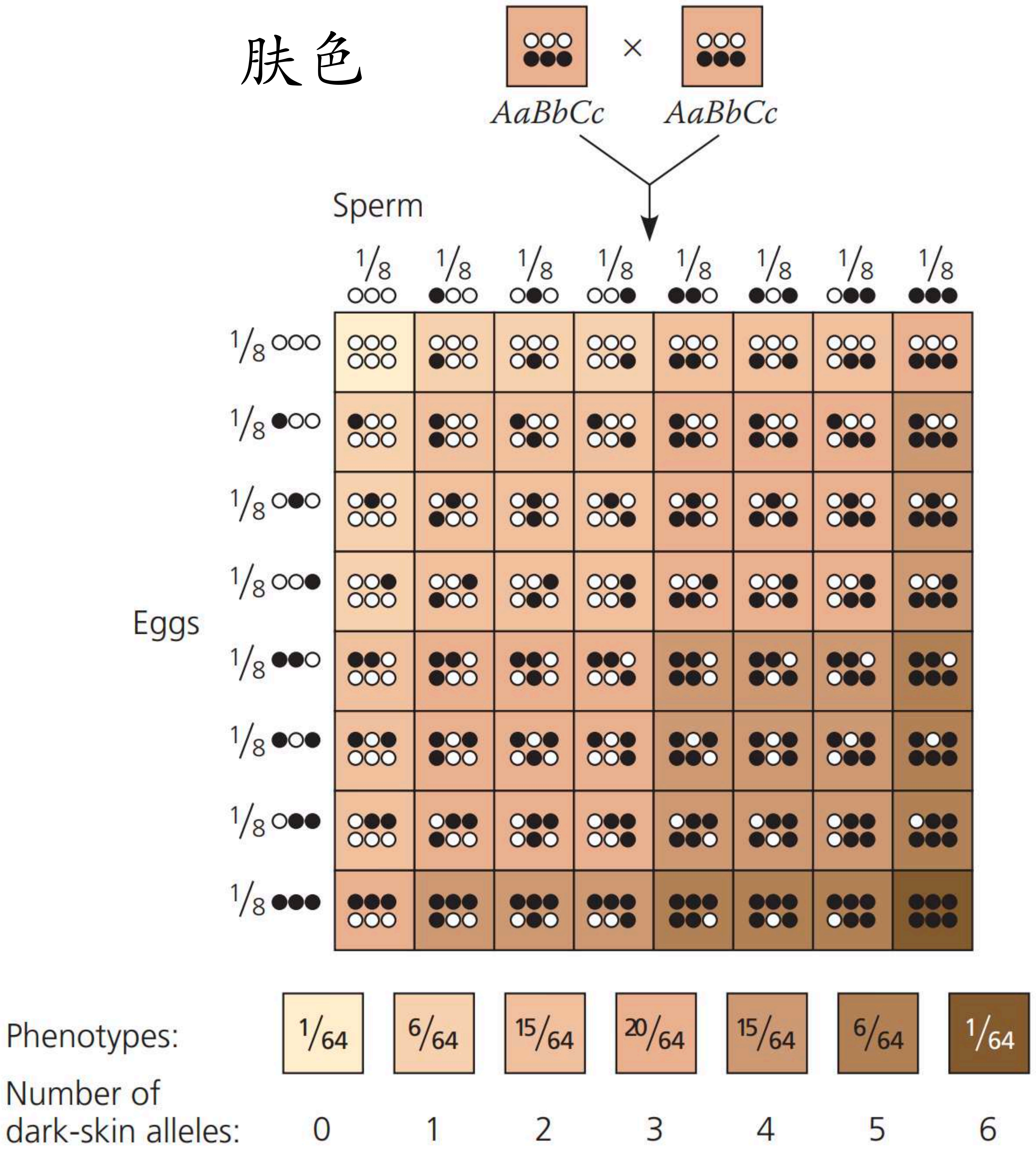
b：棕色毛，隐性

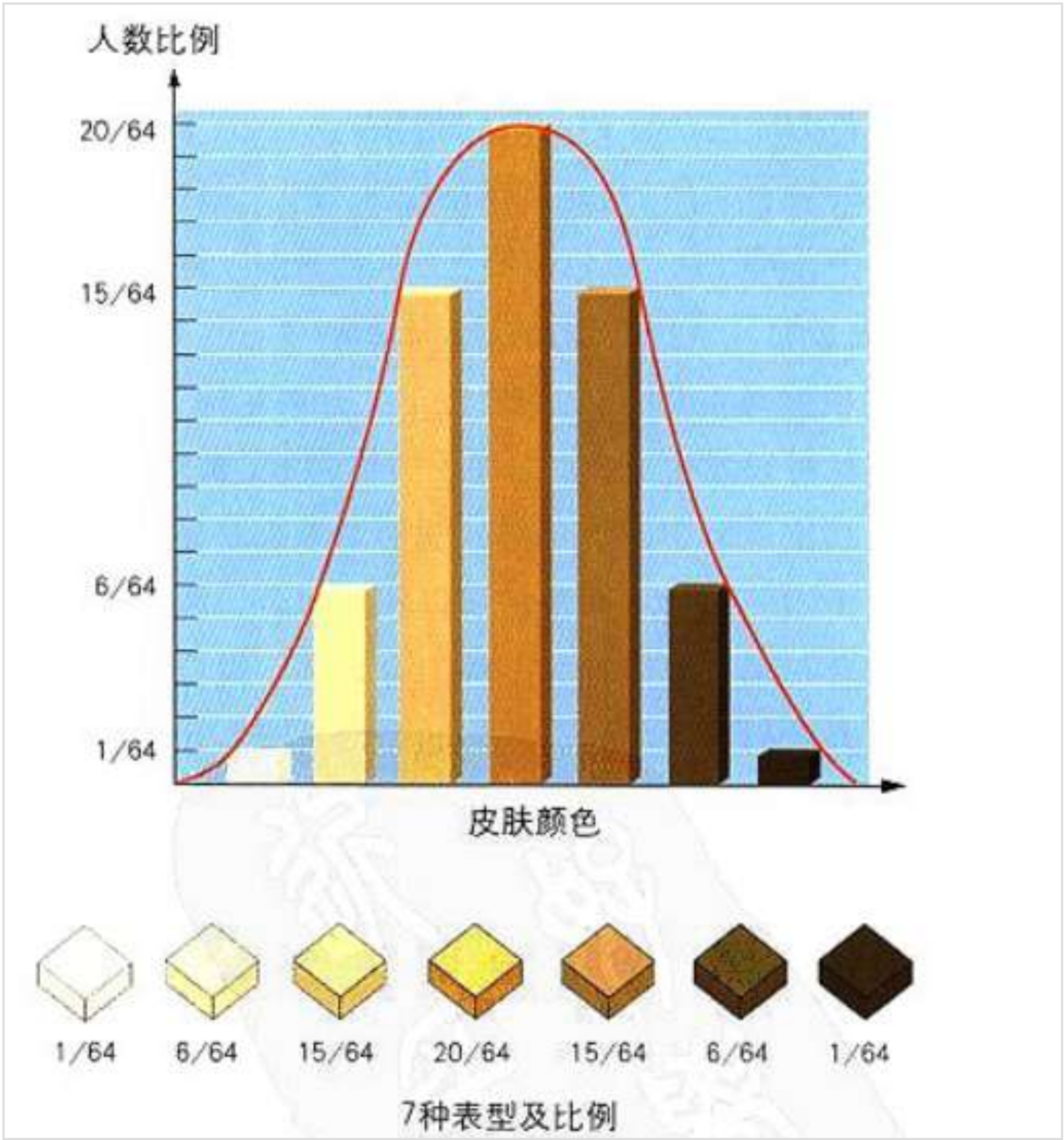
E：色素沉积，显性

e：隐性，显示黄色

当特定基因具有两个以上的等位基因时，

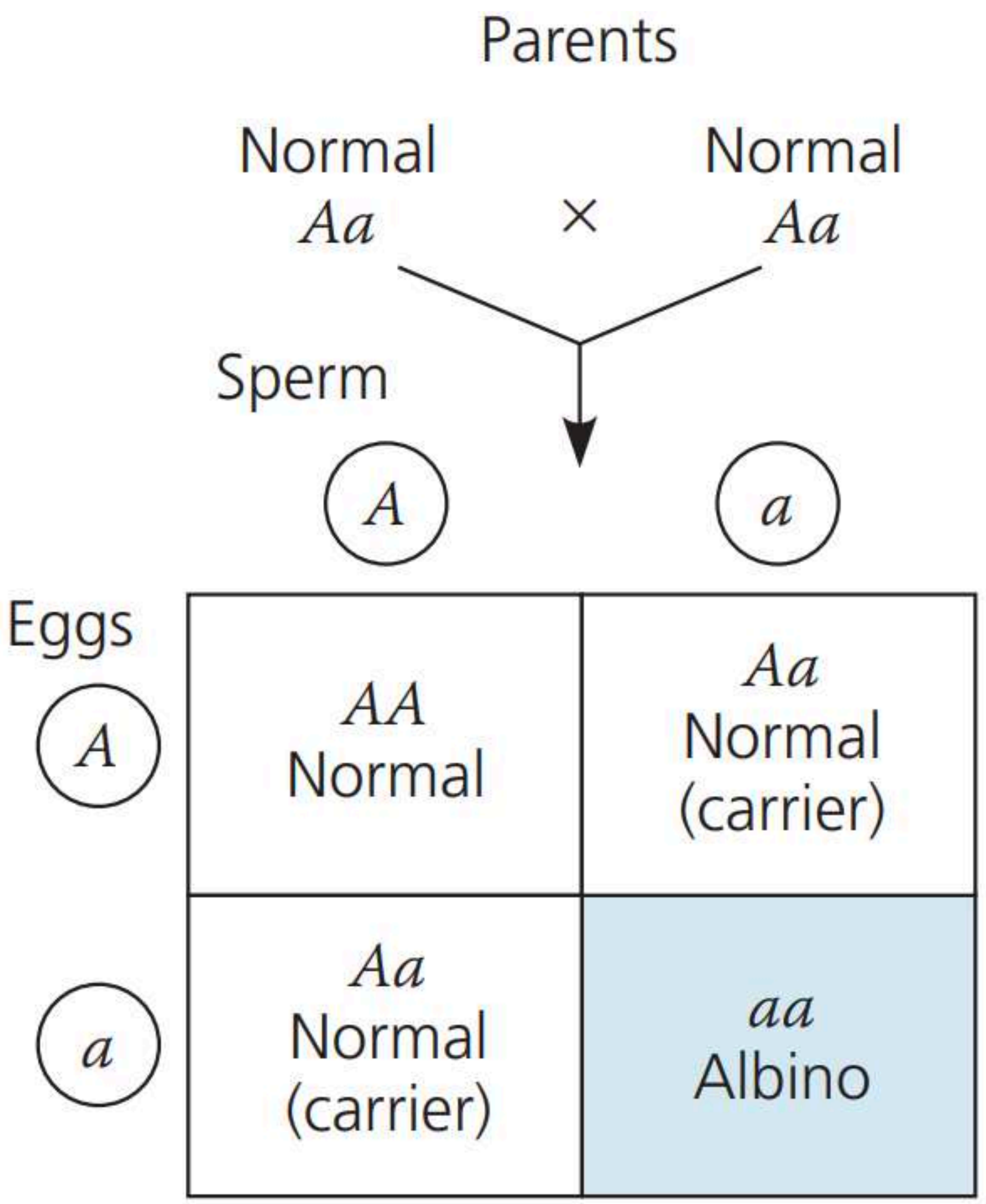
肤色




基因型 AaBbCc 和 AABbcc 对皮肤黑度的遗传贡献相同（三个单位）。 AaBbCc 杂合子之间的交配可能导致七种肤色表型

# 隐性遗传病




Albinism 白化病

Most recessive homozygotes are born to parents who are **carriers** of the disorder but themselves have a normal phenotype

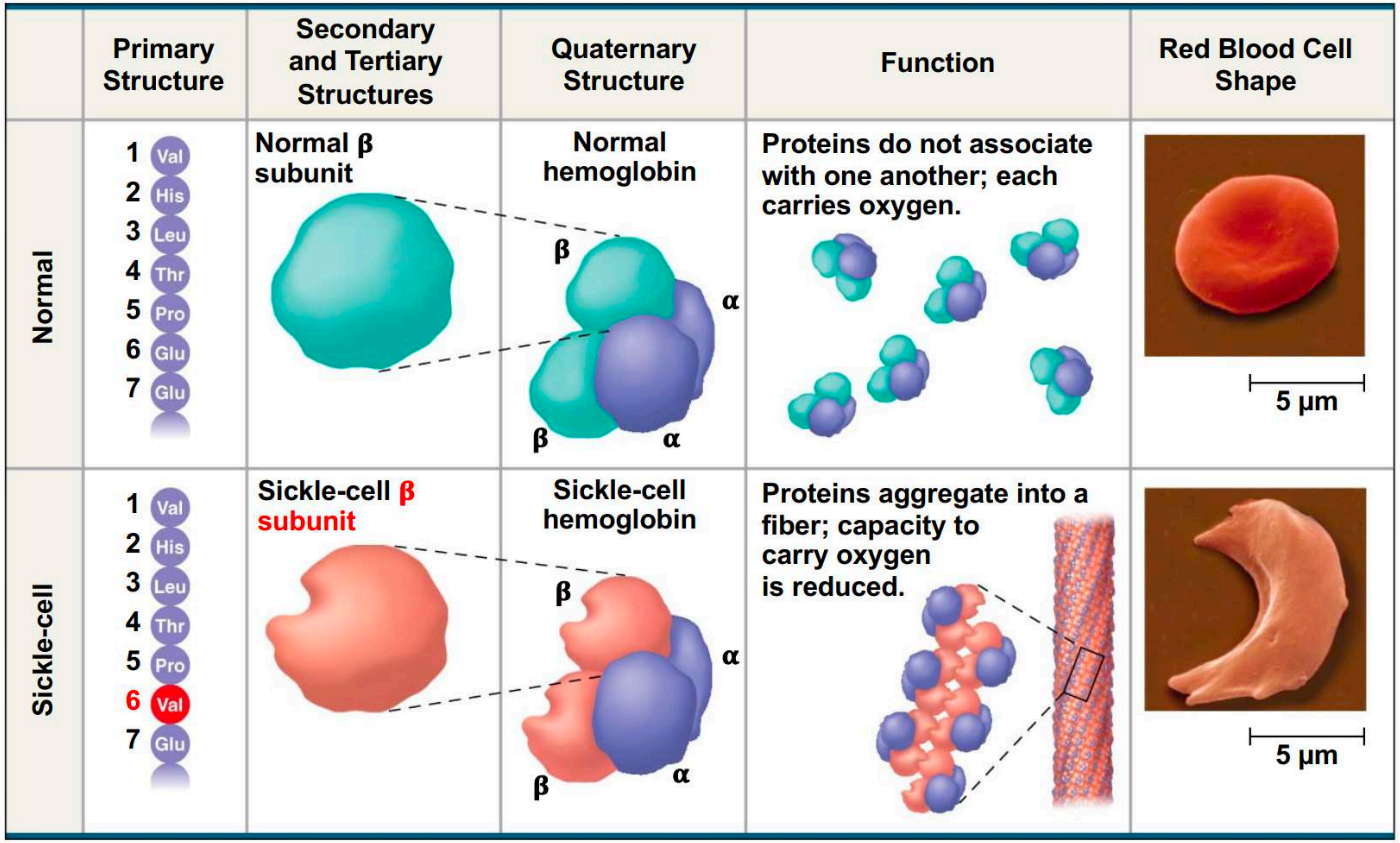
# 隐性遗传病

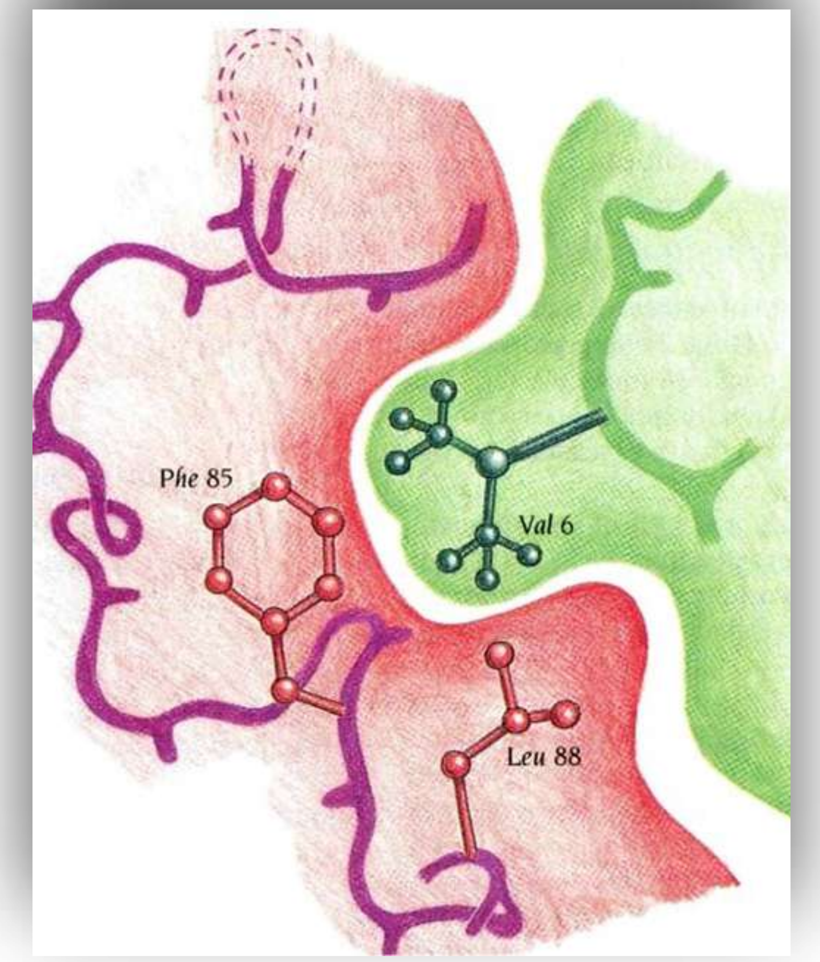
镰刀形细胞贫血症

AA

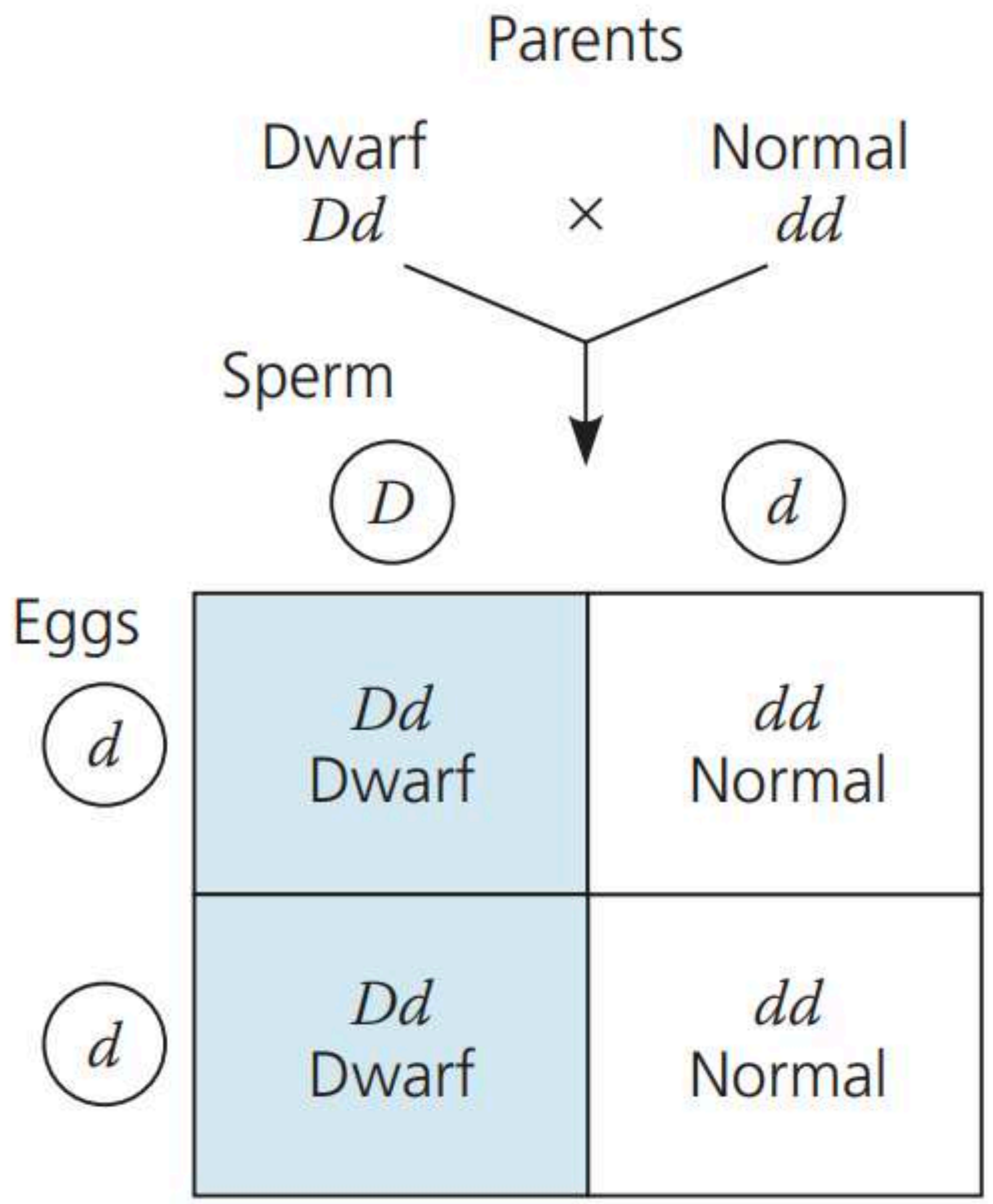
谷氨酸(Glu)
被缬氨酸
(Val)代替

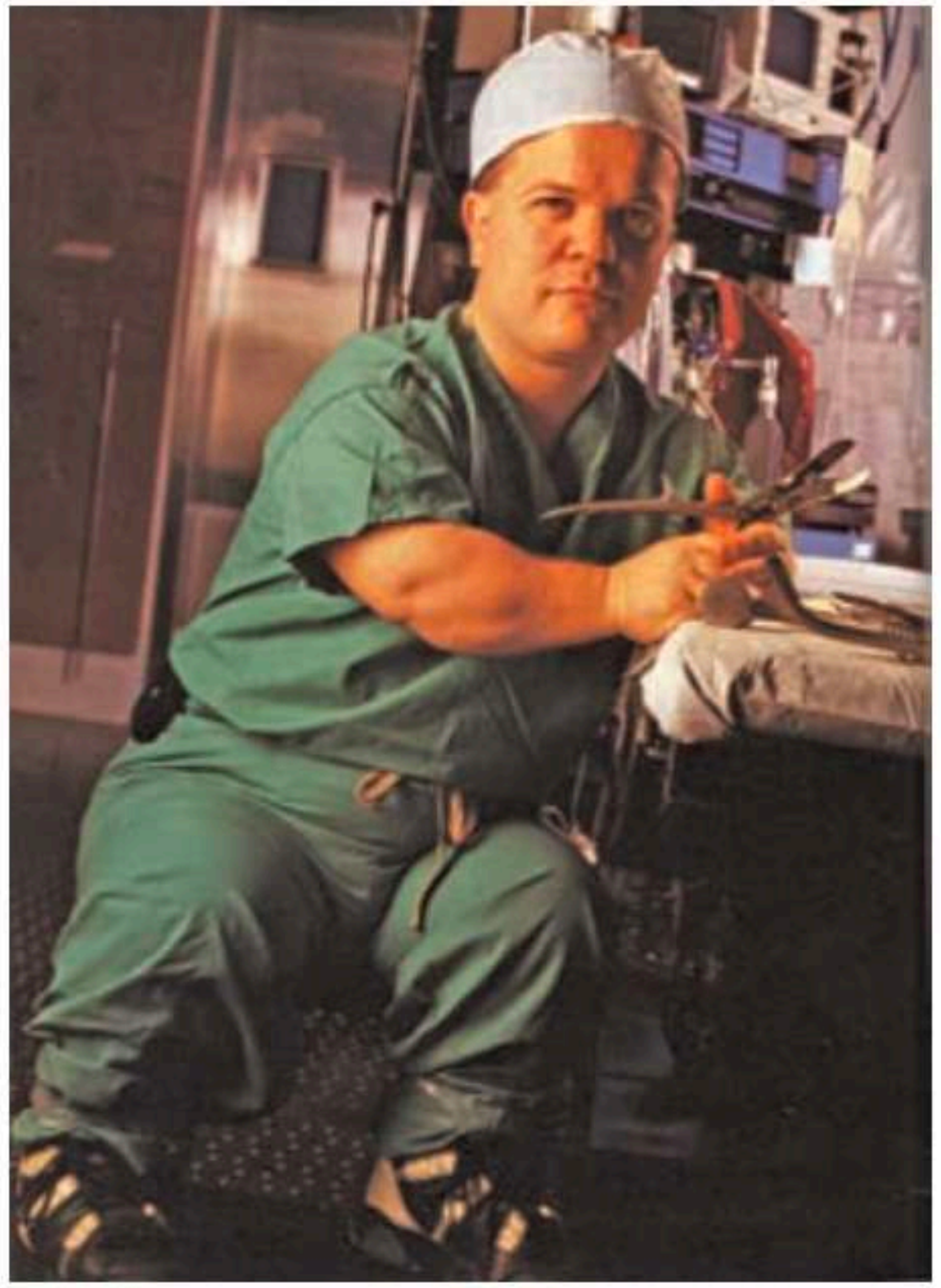
aa



# 显性遗传病



Achondroplasia 侏儒症

Dominant allele can be **rare** as well in the population.

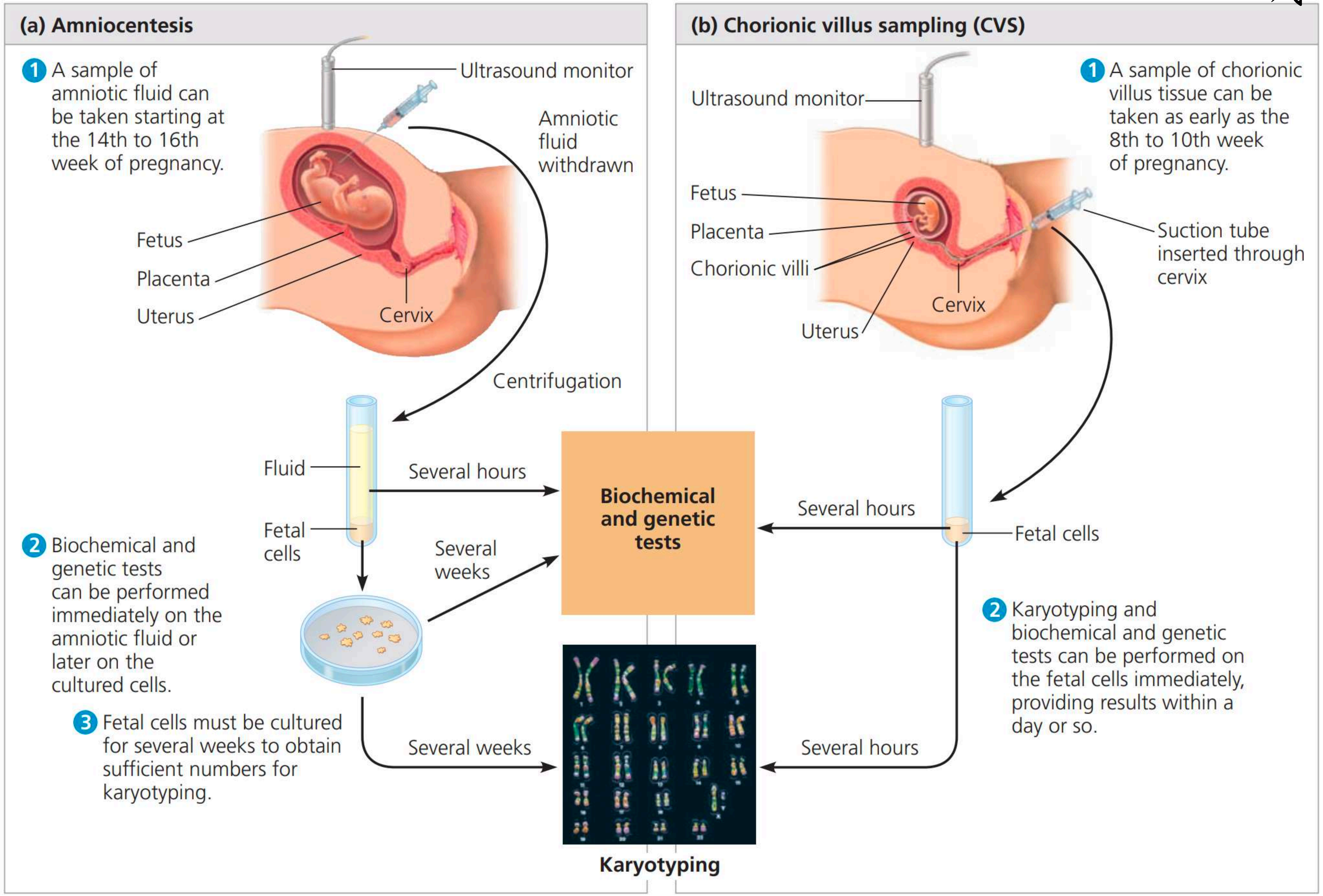
Dominant allele associated with lethal disease are less likely to be passed on the future generations than recessive alleles.

Huntington’s disease is a late onset dominant inherited disorder, and its current identification requires genetic testing.

# Genetic Testing

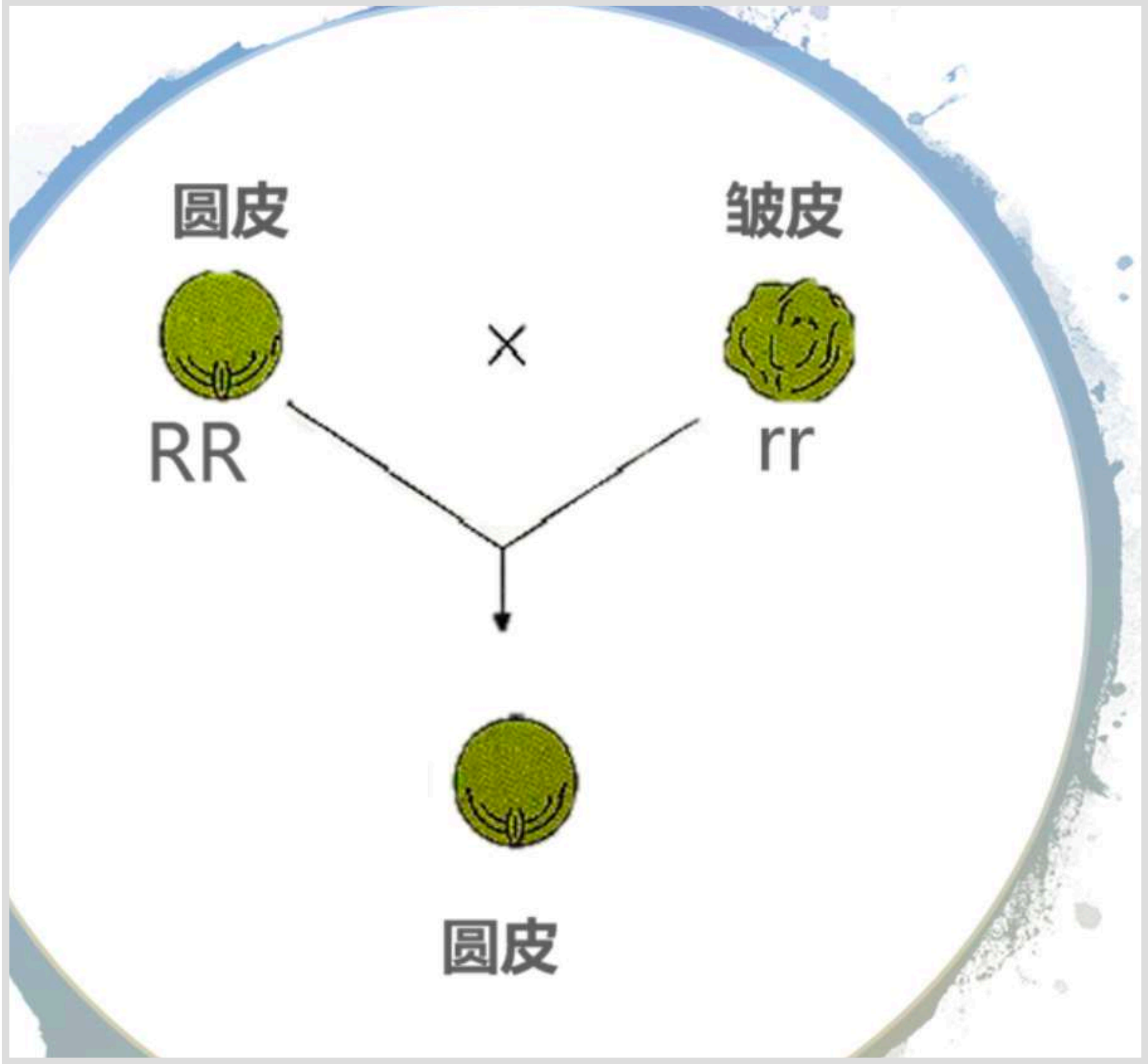
羊膜穿刺术

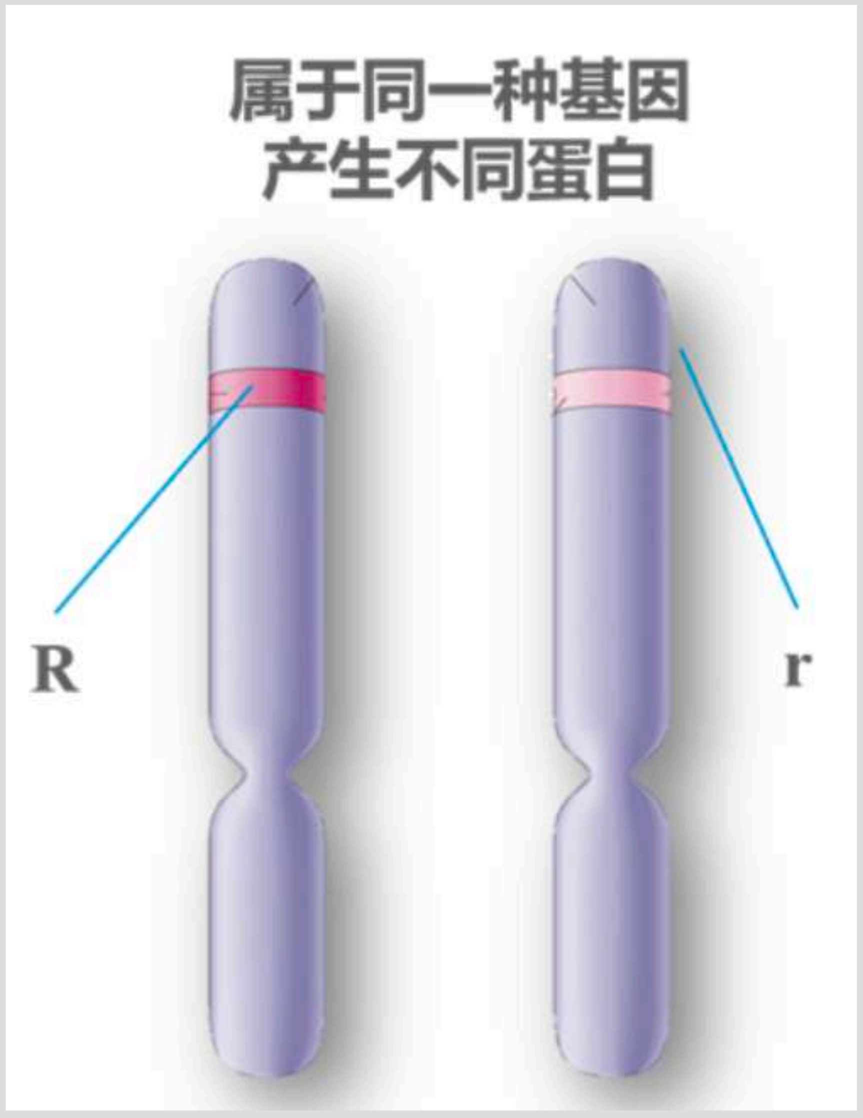
绒毛膜绒毛取样



# 概念

- 性状 (Traits)
- 表型 （Phenotype)
- 基因 （Gene)
- 等位基因 （Allele)
- 基因型 (Genotype)
- 纯合子 (Homozygote)
- 杂合子 (Heterozygote)
- 显性 (Dominant)
- 隐性 (Recessive)

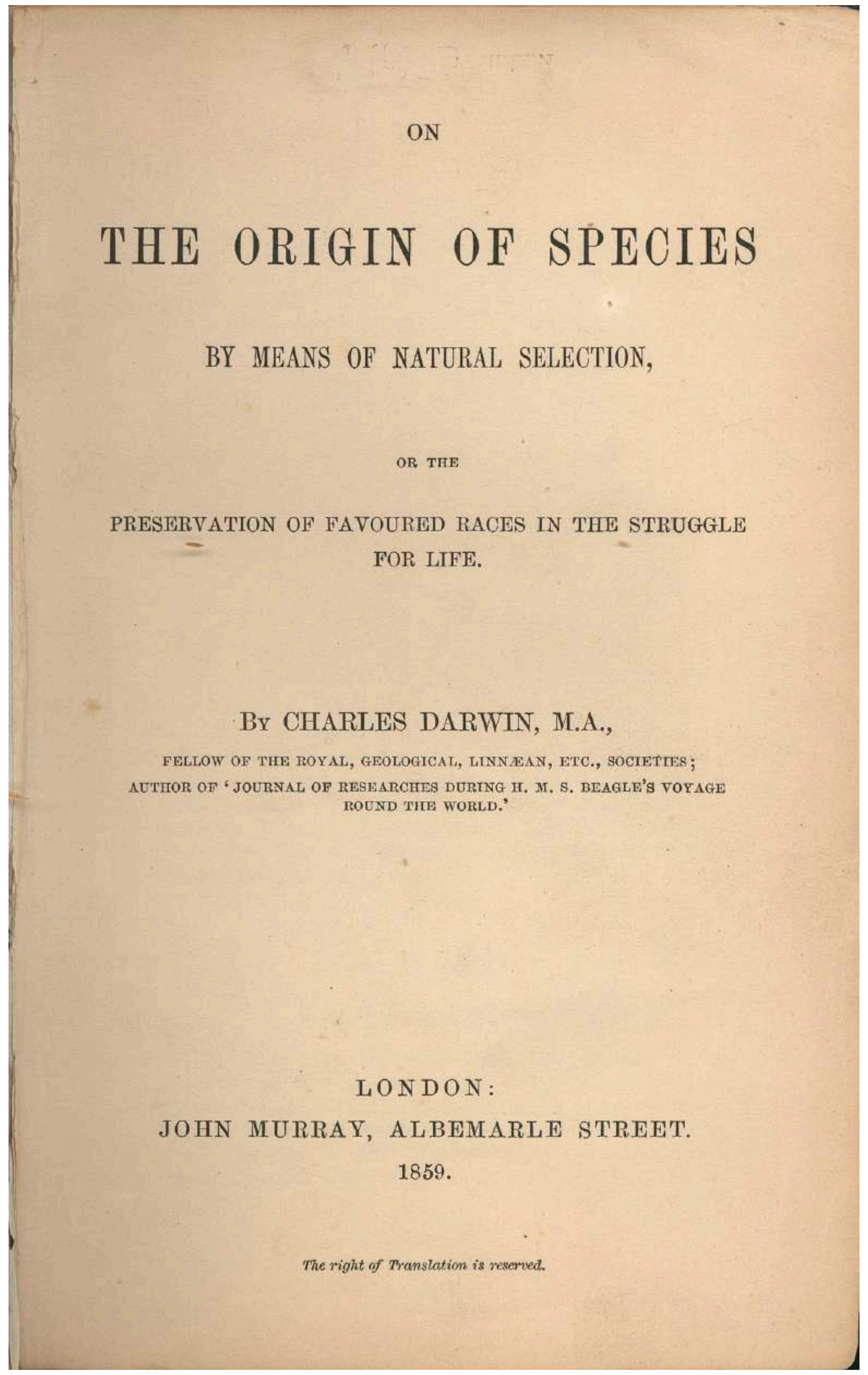
# Summary

孟德尔的遗传学实验：发现基因的存在和遗传学两大定律

基因和性状间的关系

显性和隐性遗传病

# 绝代双雄，未曾相逢


ON

THE ORIGIN OF SPECIES

BY MEANS OF NATURAL SELECTION,

OR THE

PRESERVATION OF FAVOURED RACES IN THE STRUGGLE FOR LIFE.

By CHARLES DARWIN, M.A.,

FELLOW OF THE ROYAL, GEOLOGICAL, LINNÆAN, ETC., SOCIETIES;
AUTHOR OF 'JOURNAL OF RESEARCHES DURING H. M. S. BEAGLE'S VOYAGE ROUND THE WORLD.'

LONDON:
JOHN MURRAY, ALBEMARLE STREET.
1859.

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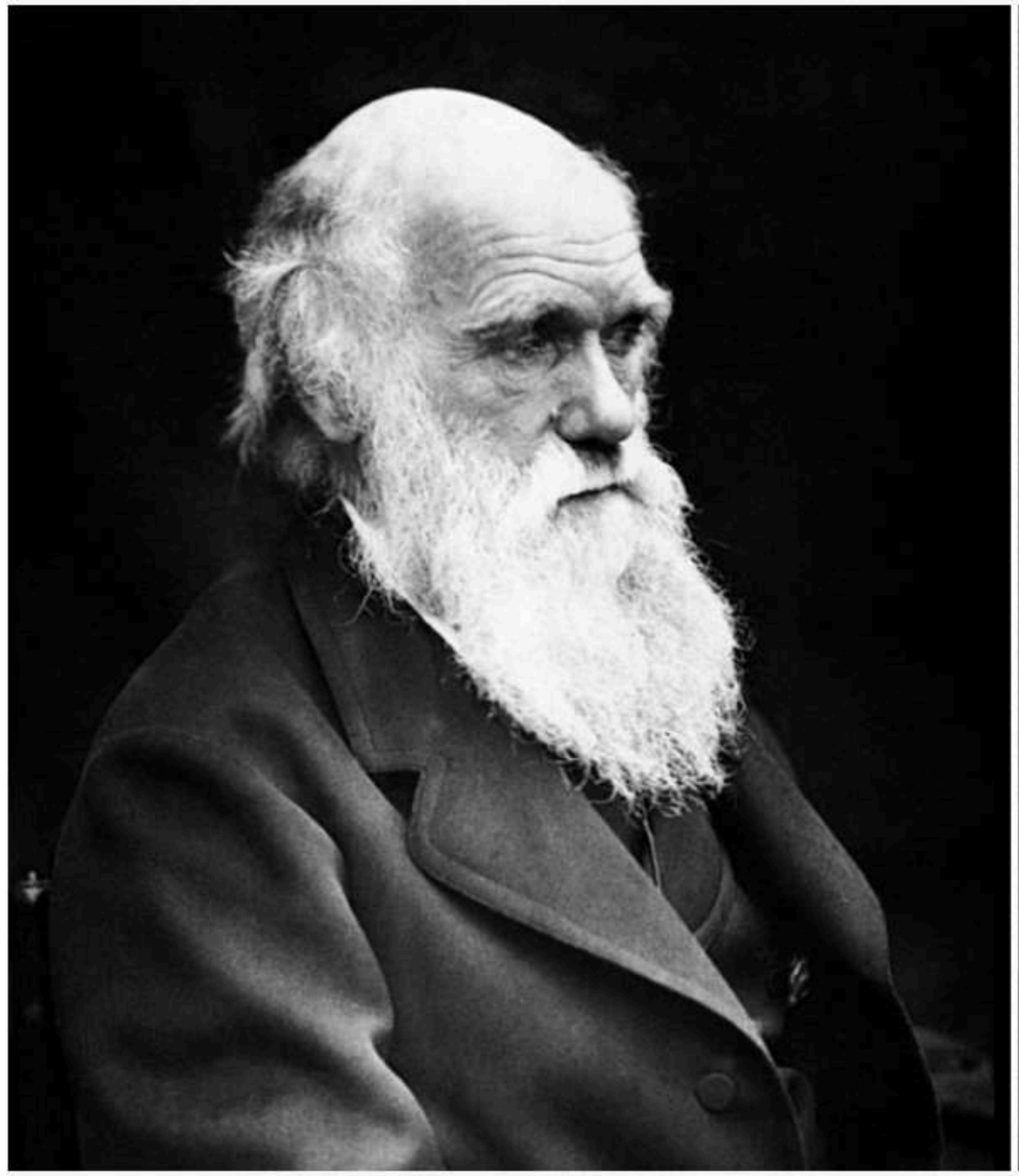
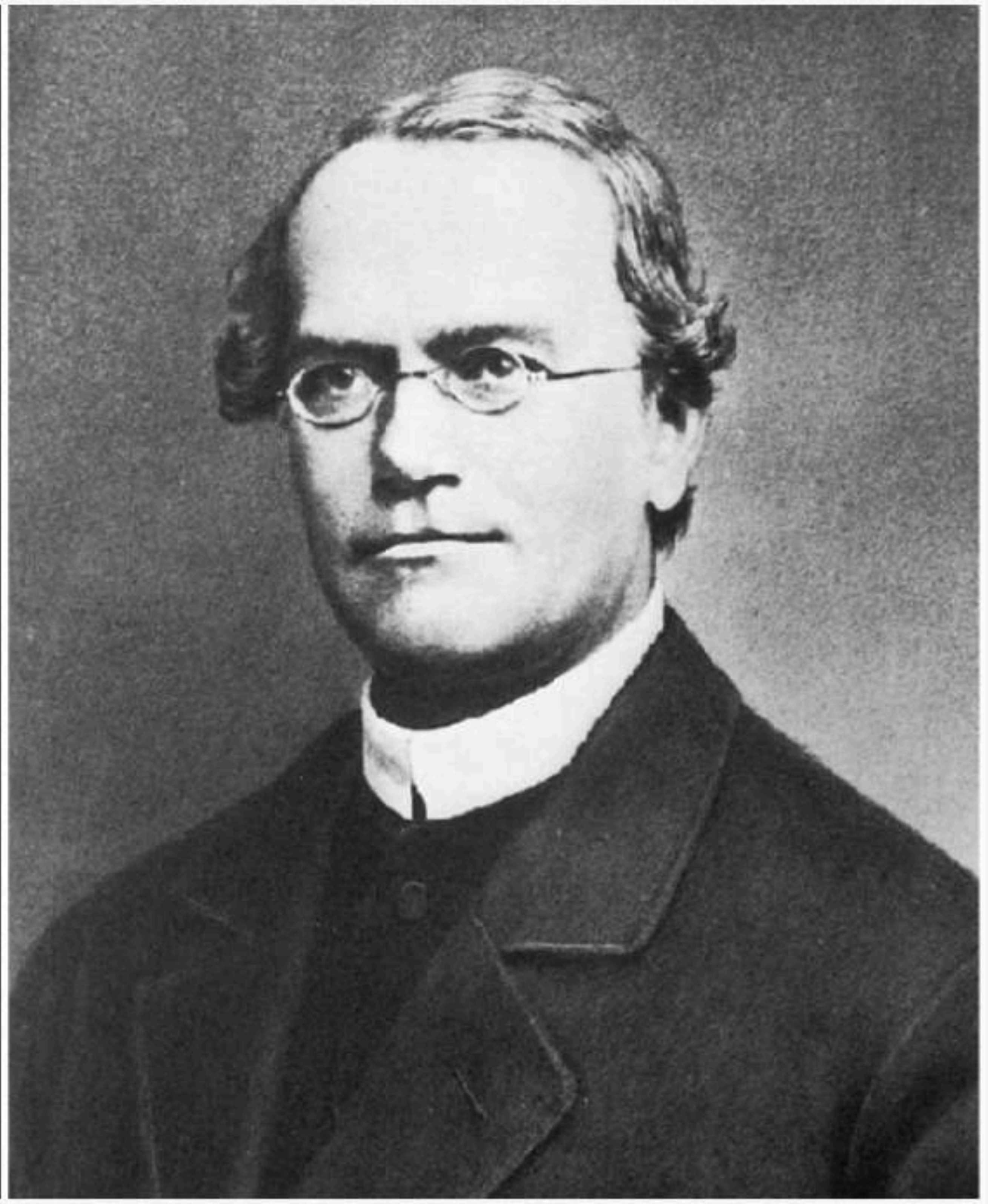
《物种起源》

1859



达尔文

(1809-1882)



孟德尔　遗传定律1866

(1822-1884)

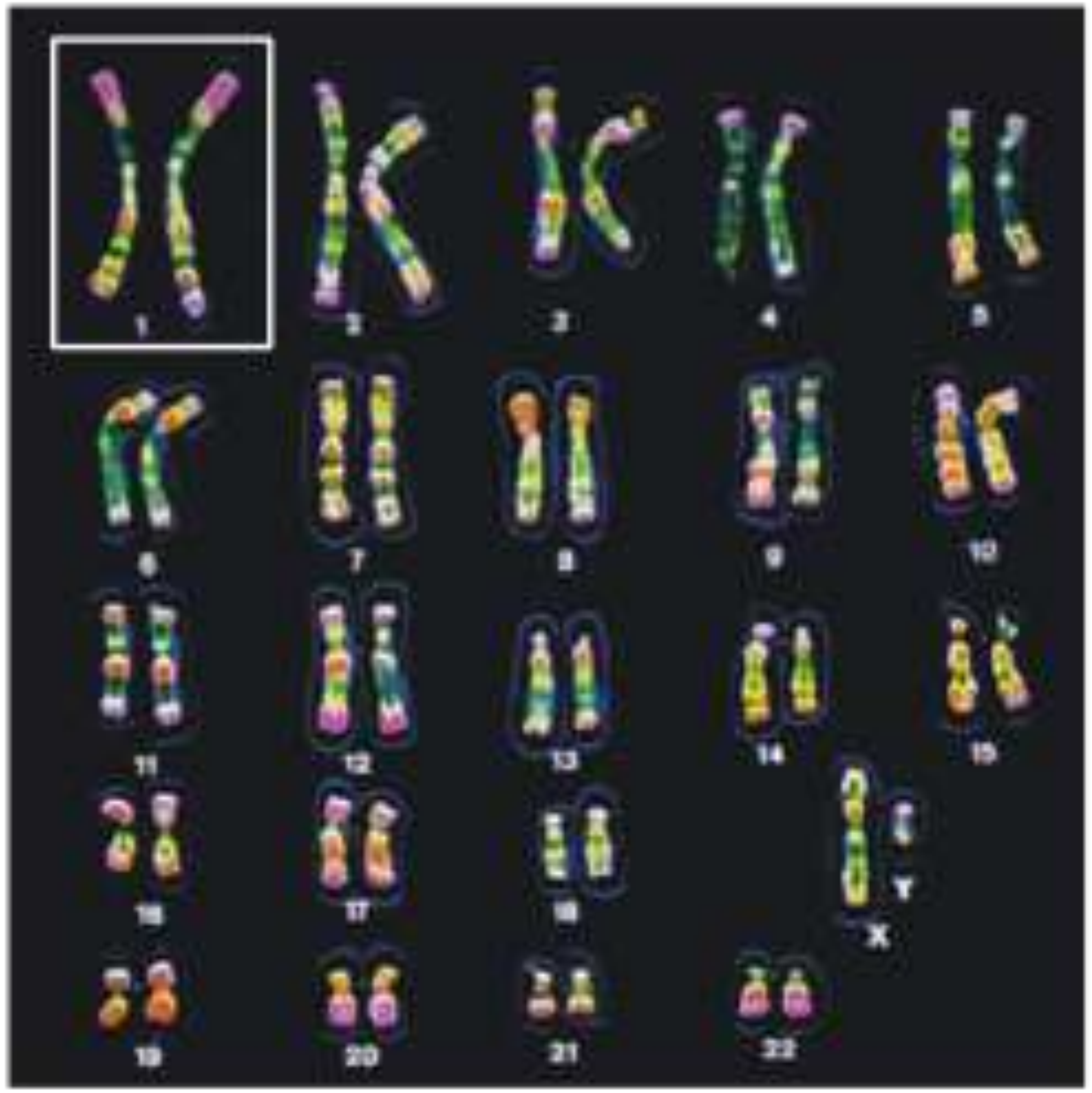
现代达尔文主义=达尔文主义+孟德尔主义

# Genetic Linkage

# （基因连锁和交换定律）

Thomas Hunt Morgan，1909

# 孟德尔遗传学定律的物质基础



Karyotype

染色体组型

# The chromosomal basis of Mendel's laws

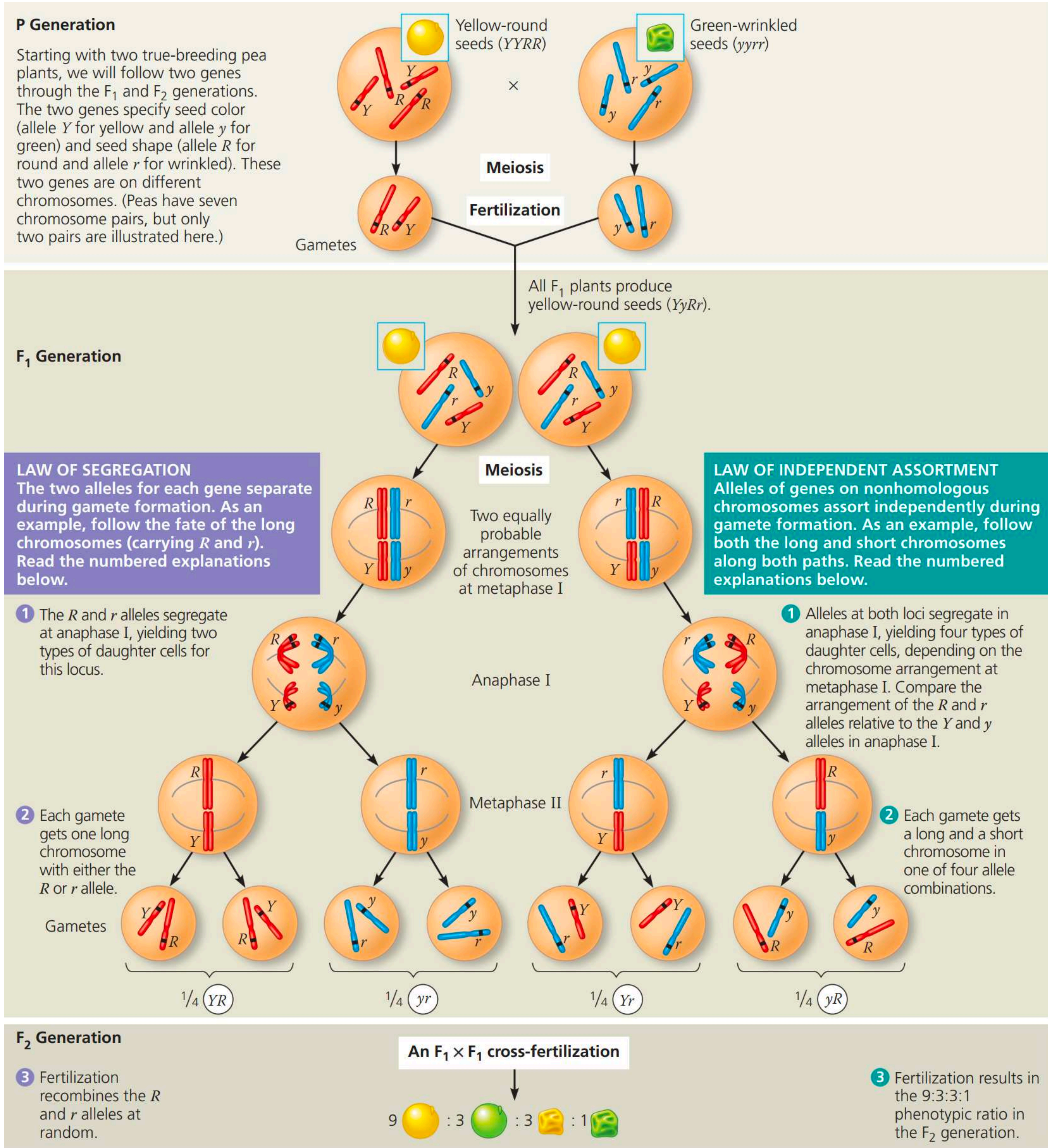
R/r, Y/y位于不同的染色体上

等位基因R与r在减数分裂后期分离

同源染色体分离

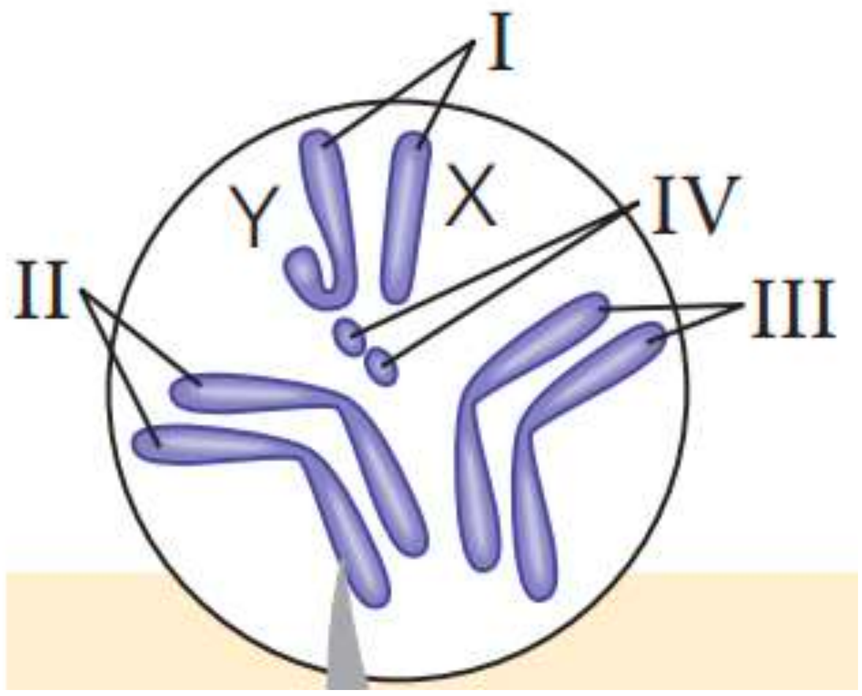
非同源染色体自由组合

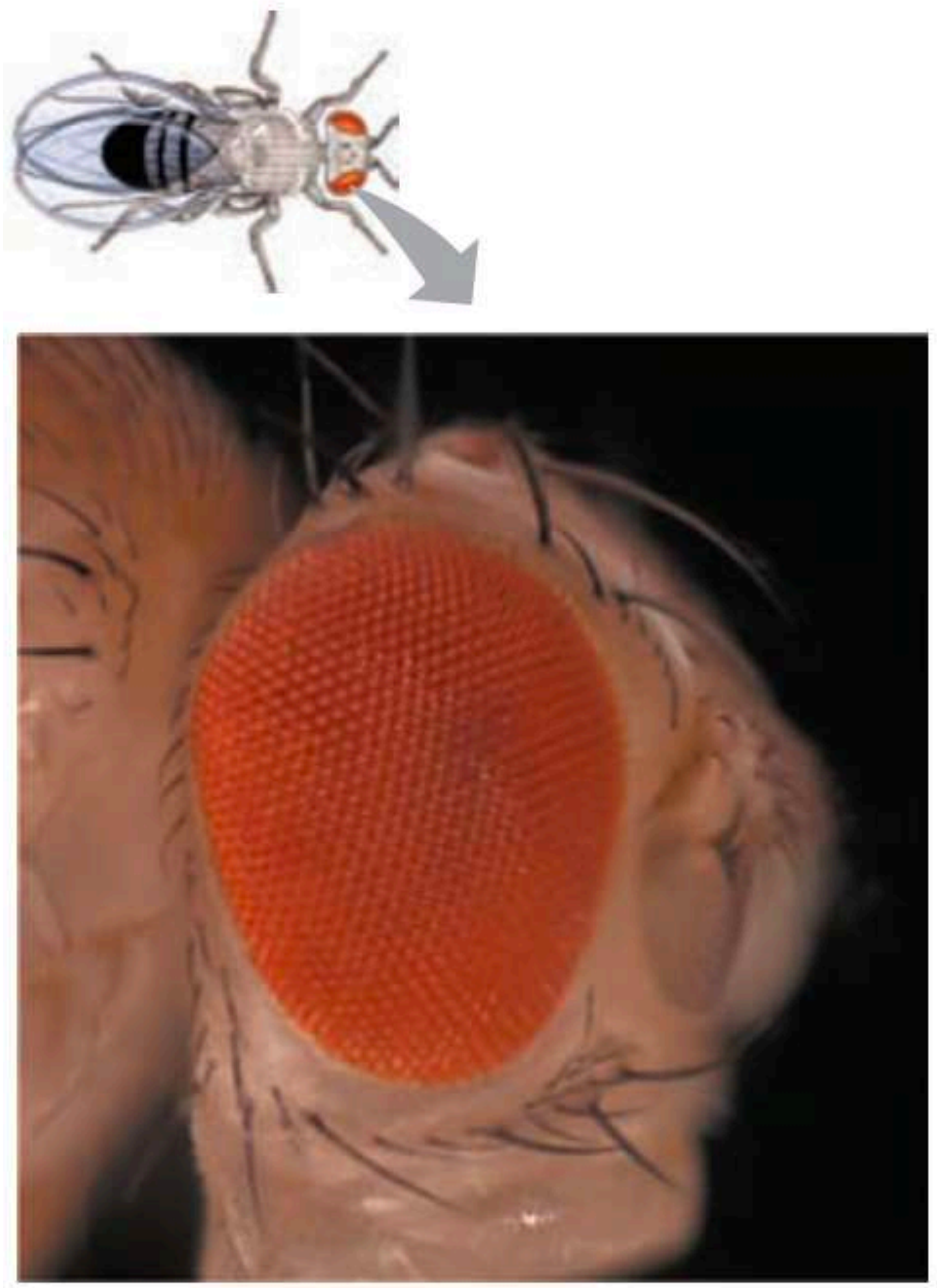
配子



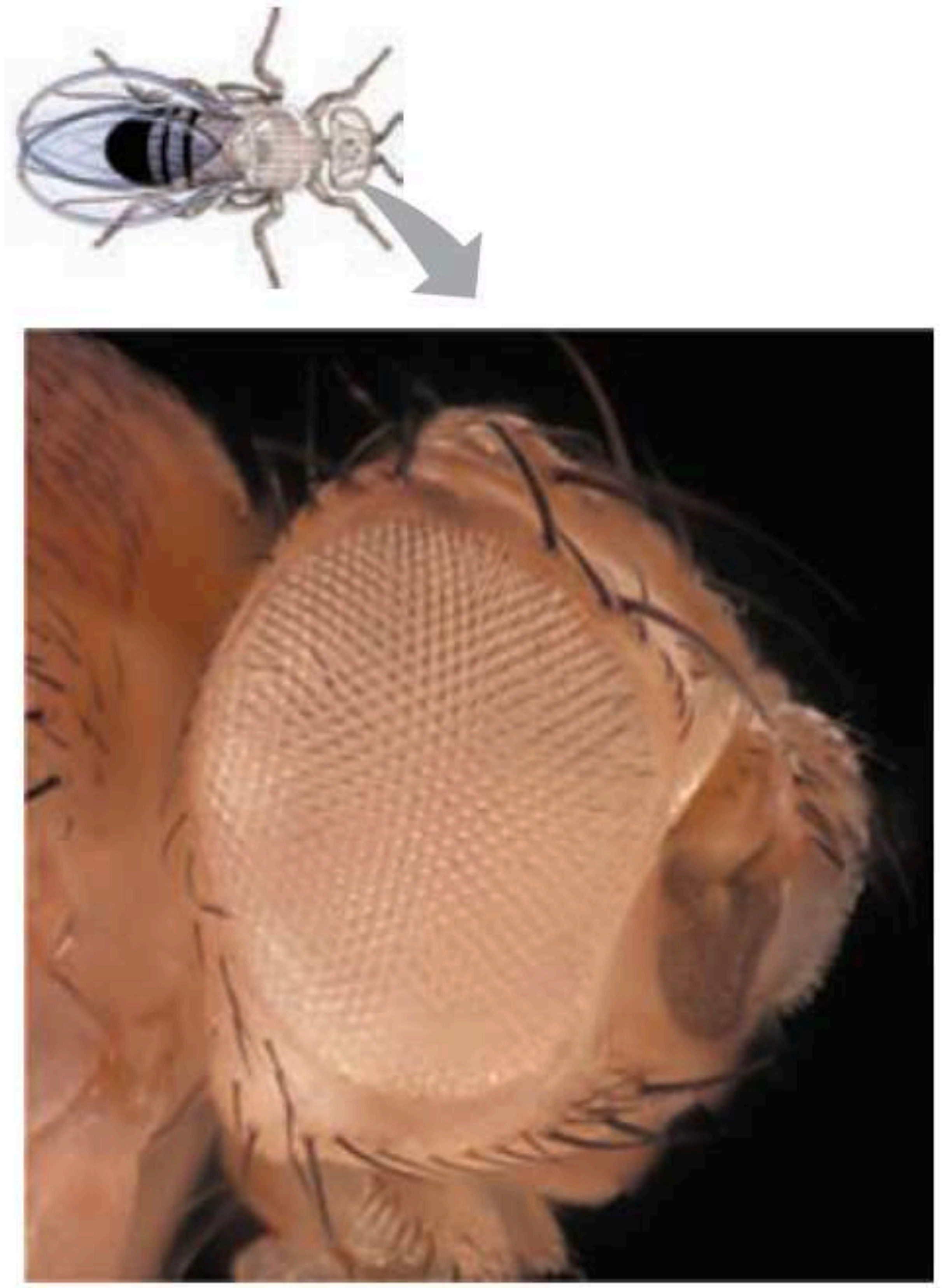
# 摩尔根的实验

实验动物： 果蝇



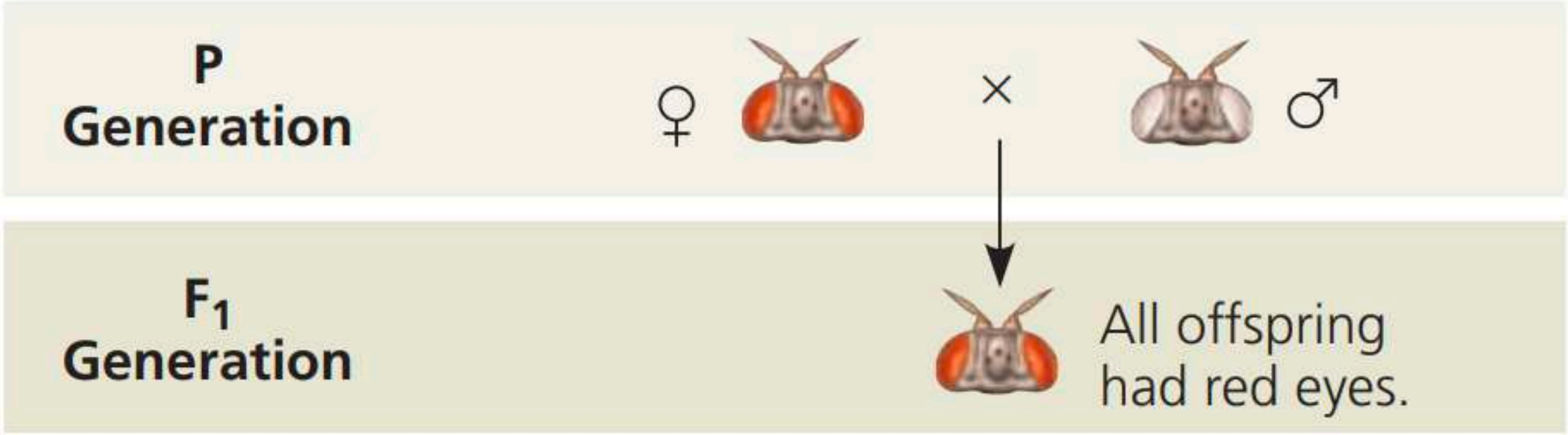



野生型 W+

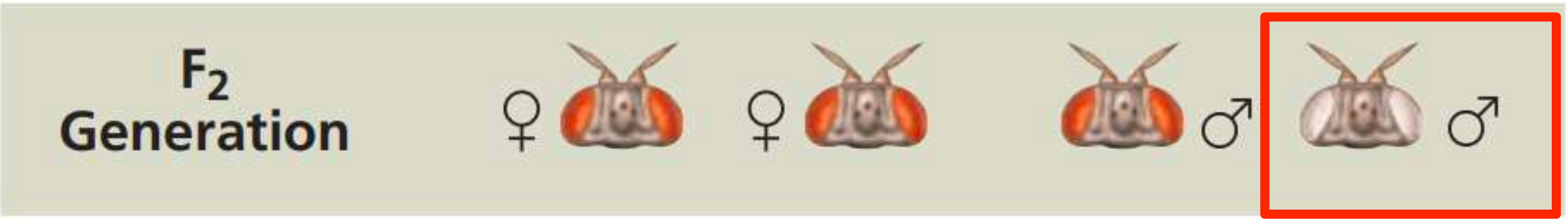


突变型W

# 摩尔根的实验

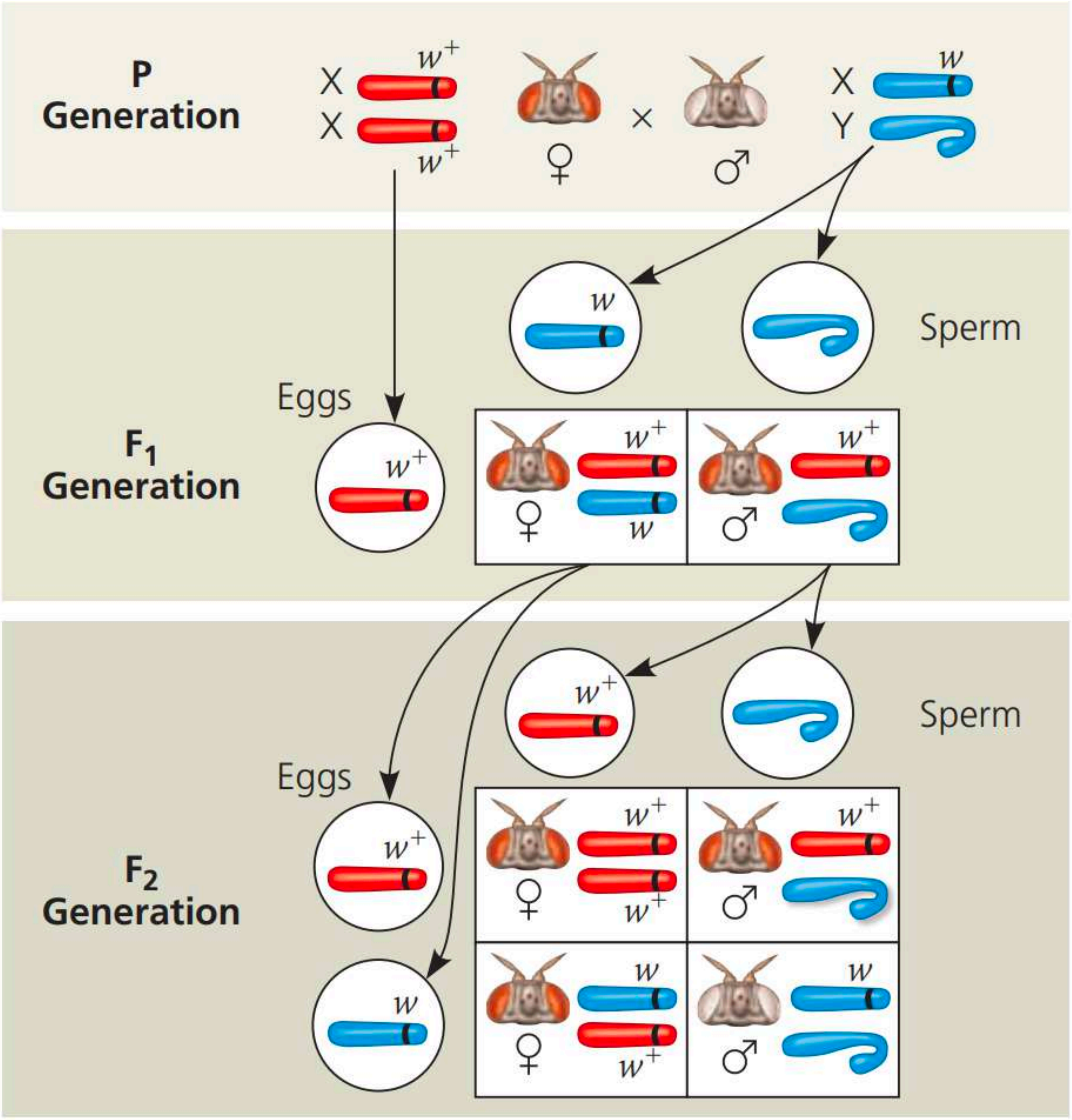



F1红眼雌性 x F1红眼雄性



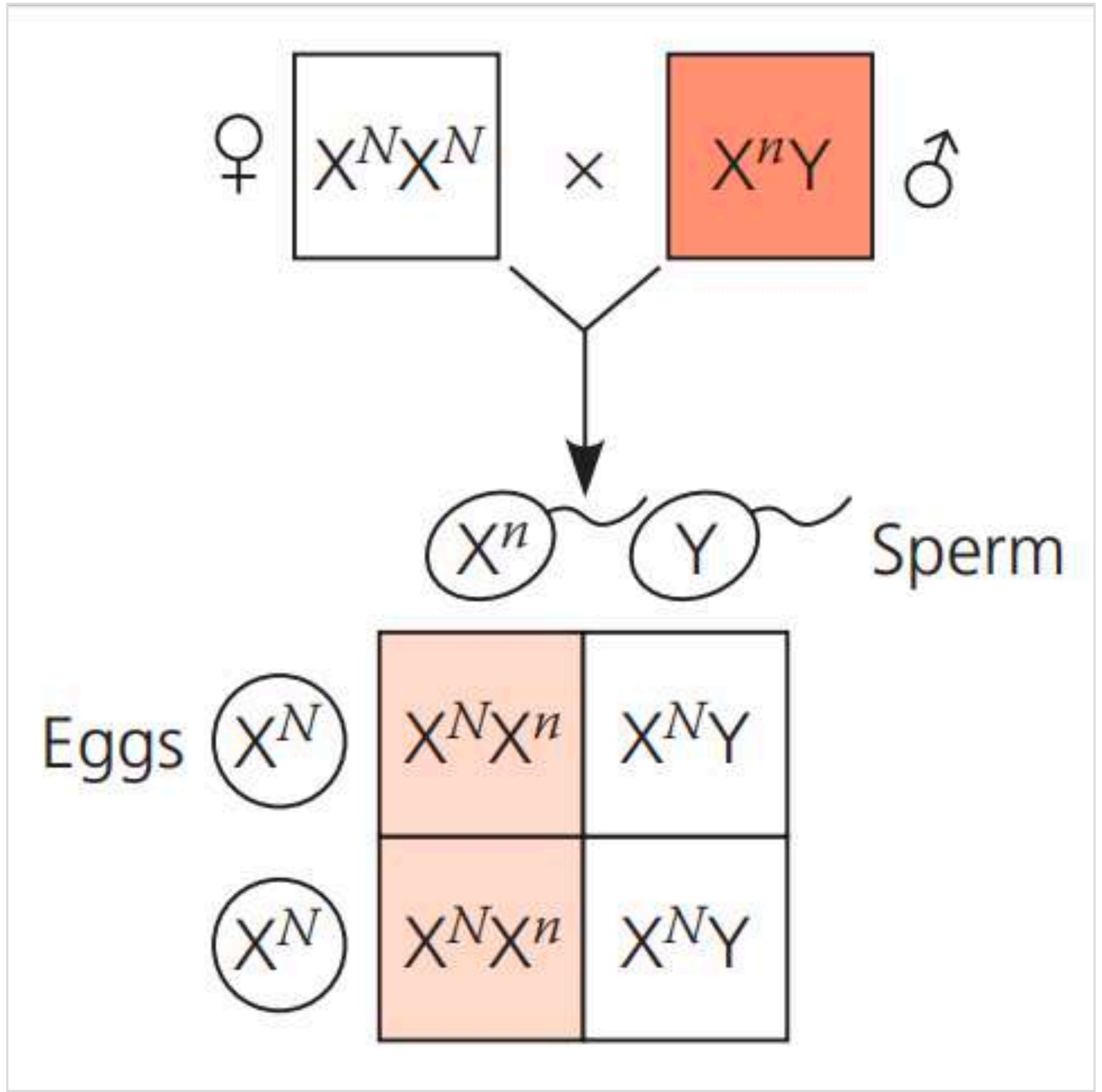

3:1

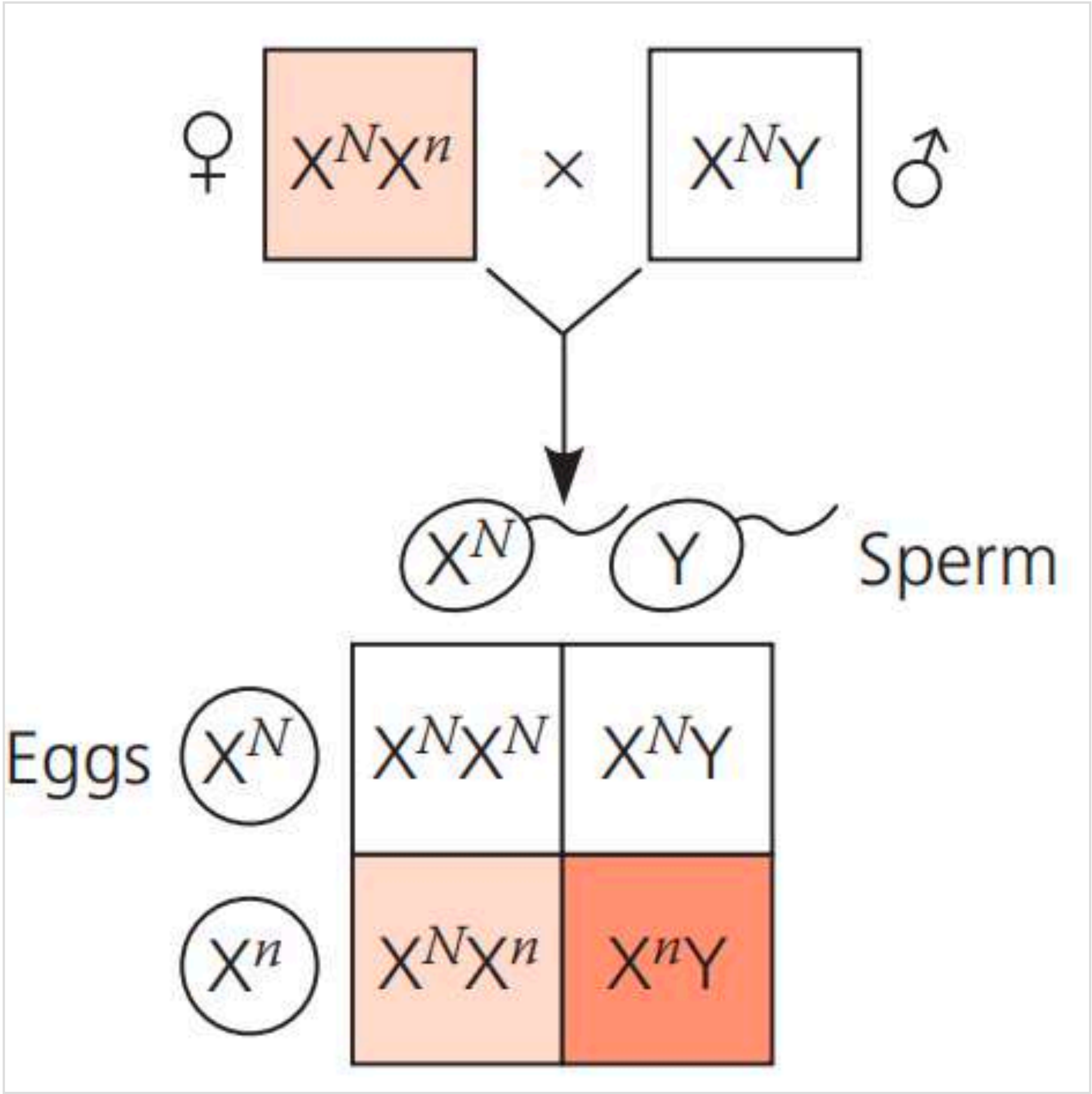
特定基因携带在特定染色体上

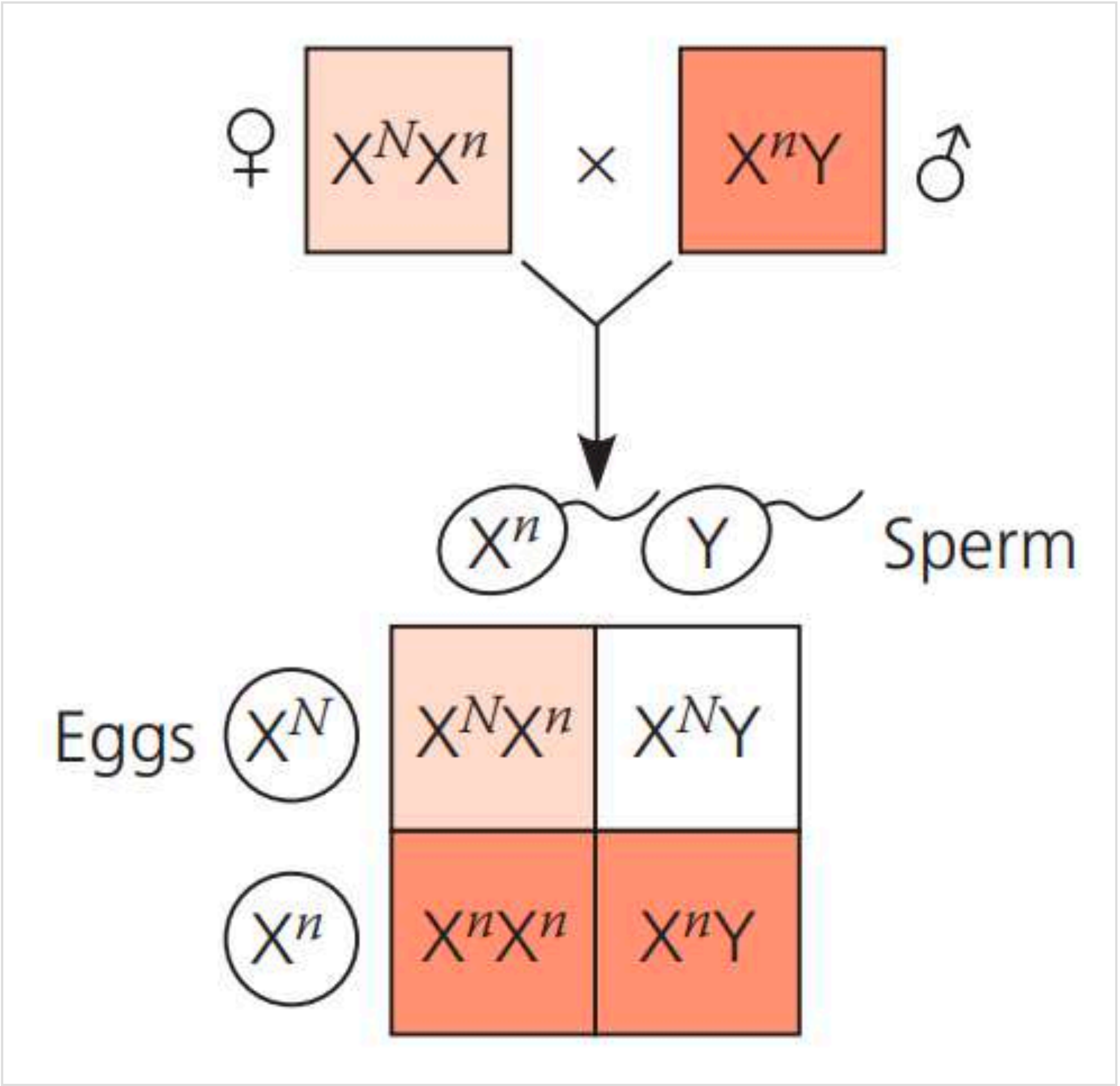


# Inheritance of X-Linked Genes X连锁基因的遗传

位于任一性染色体上的基因称为性连锁基因



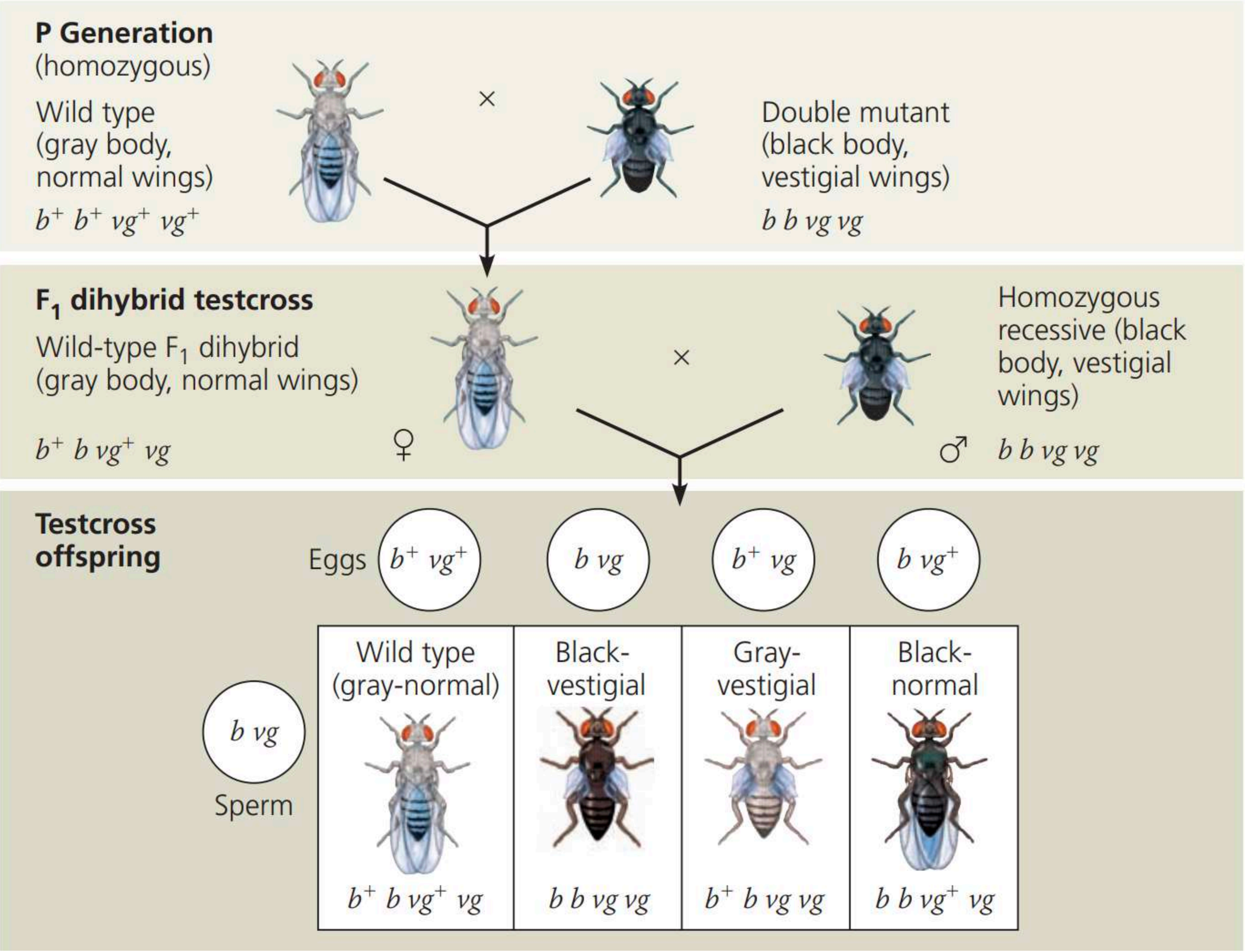



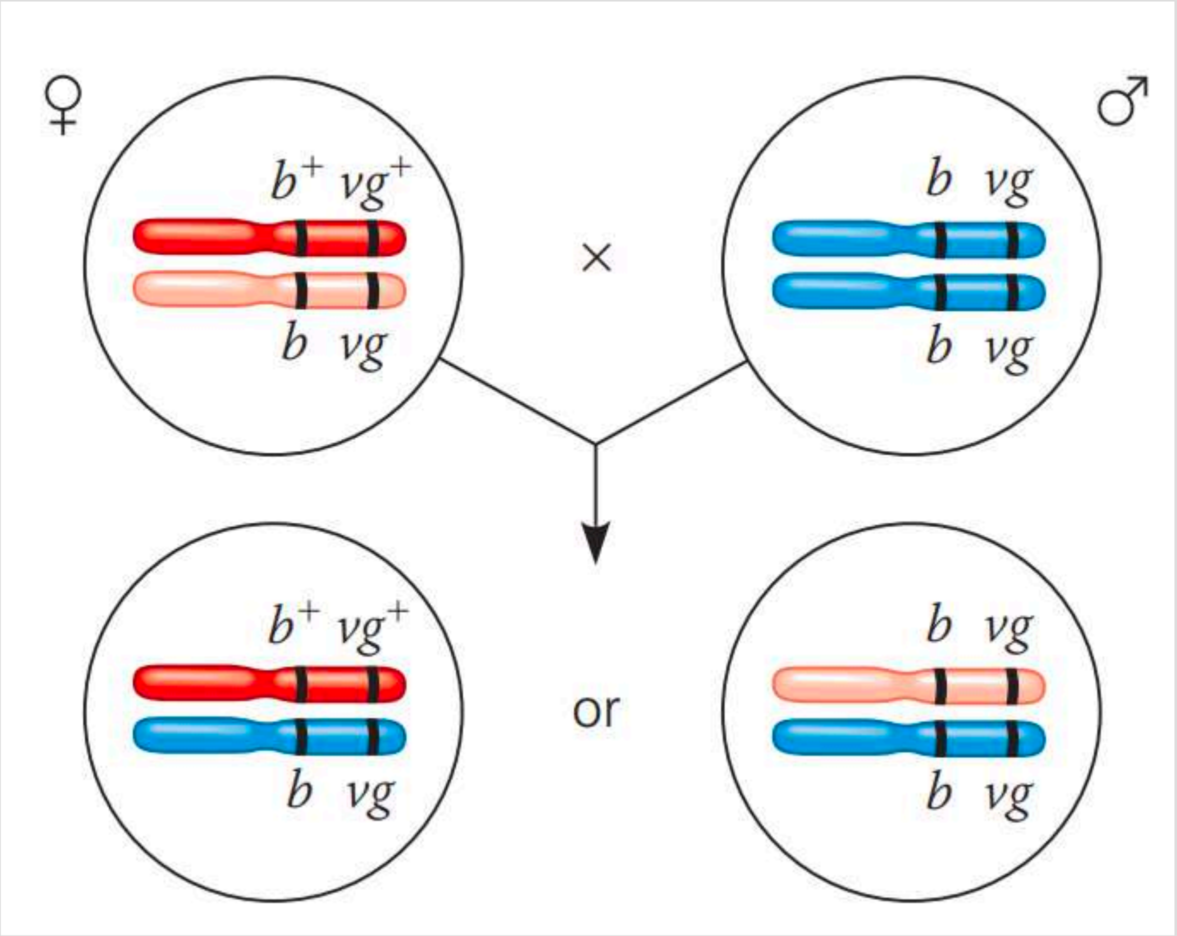

例子：红绿色盲

Linked genes (连锁基因) tend to be inherited together because they are located near each other on the same chromosome

灰色的身体
正常大小的翅膀

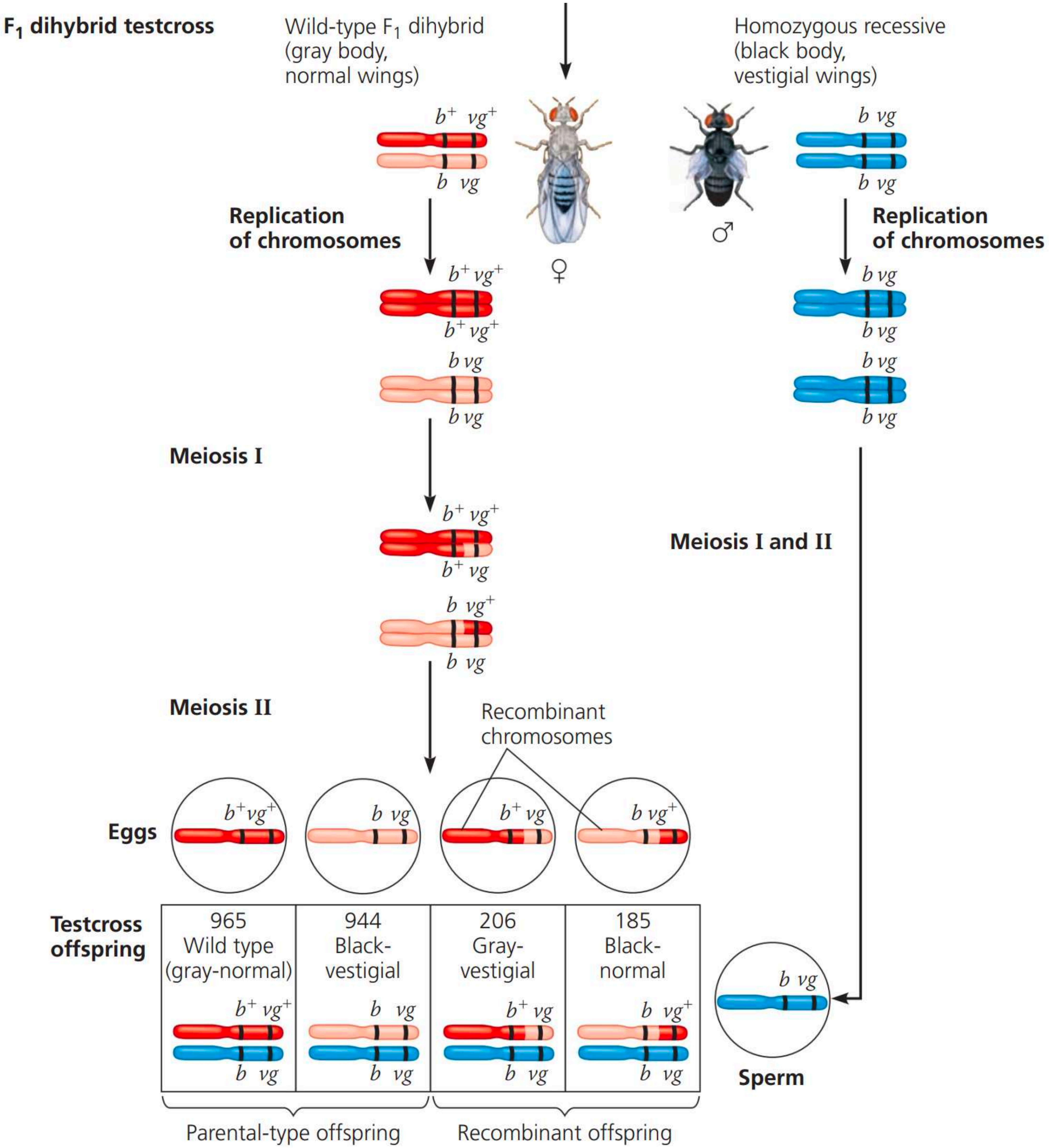



黑色的身体
退化翅膀

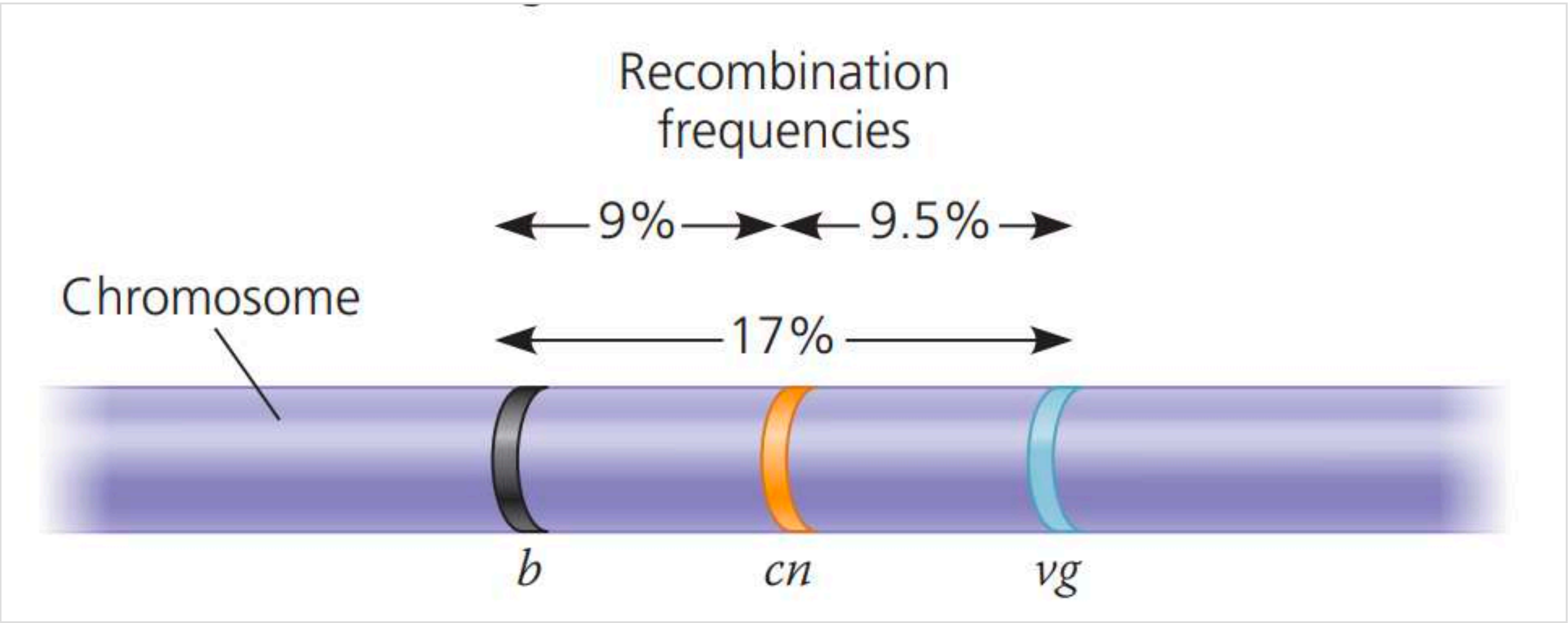



| | | | | | | | |
|---|---|---|---|---|---|---|---|
| Predicted ratio if genes are located on different chromosomes: | 1 | : | 1 | : | 1 | : | 1 |
| Predicted ratio if genes are located on the same chromosome *and* parental alleles are always inherited together: | 1 | : | 1 | : | 0 | : | 0 |
| Data from Morgan's experiment: | 965 | : | 944 | : | 206 | : | 185 |

# Chromosomal basis for recombination of linked genes

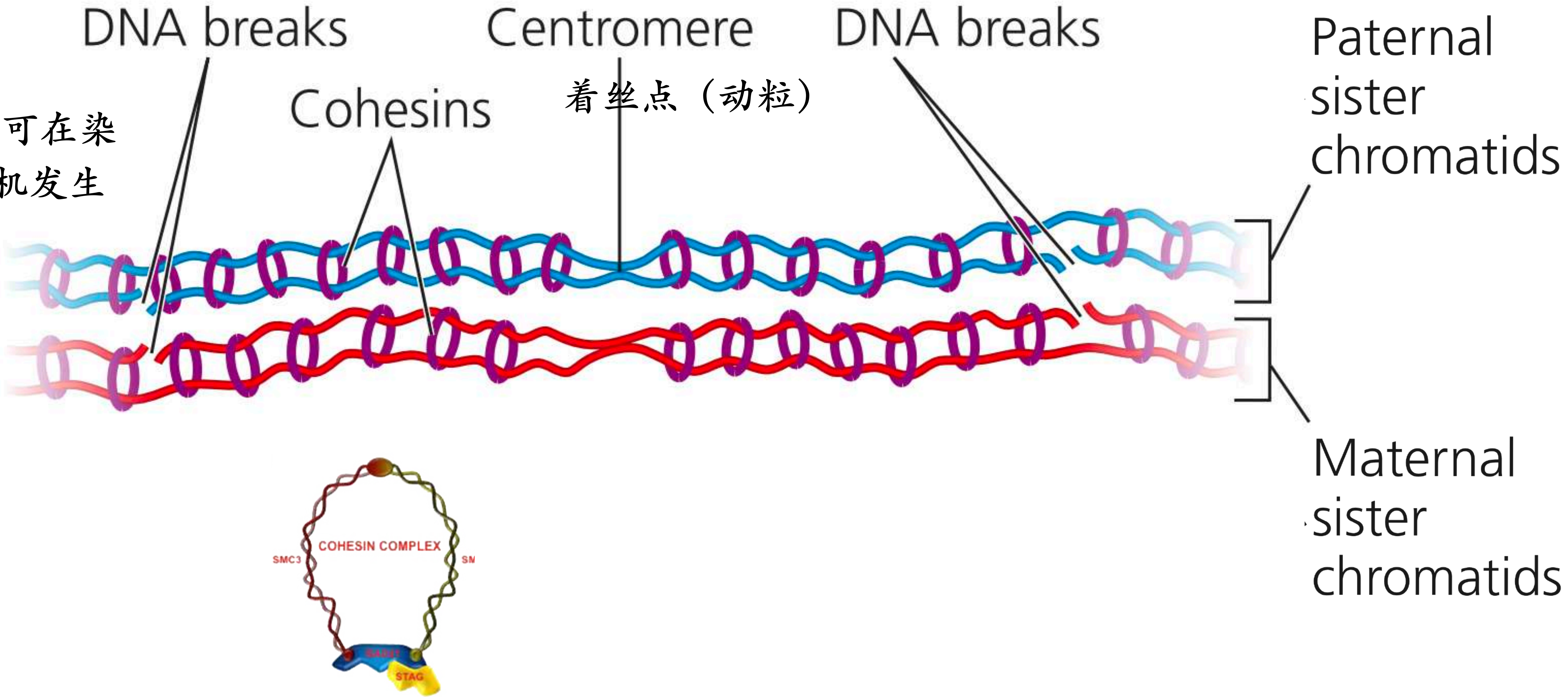


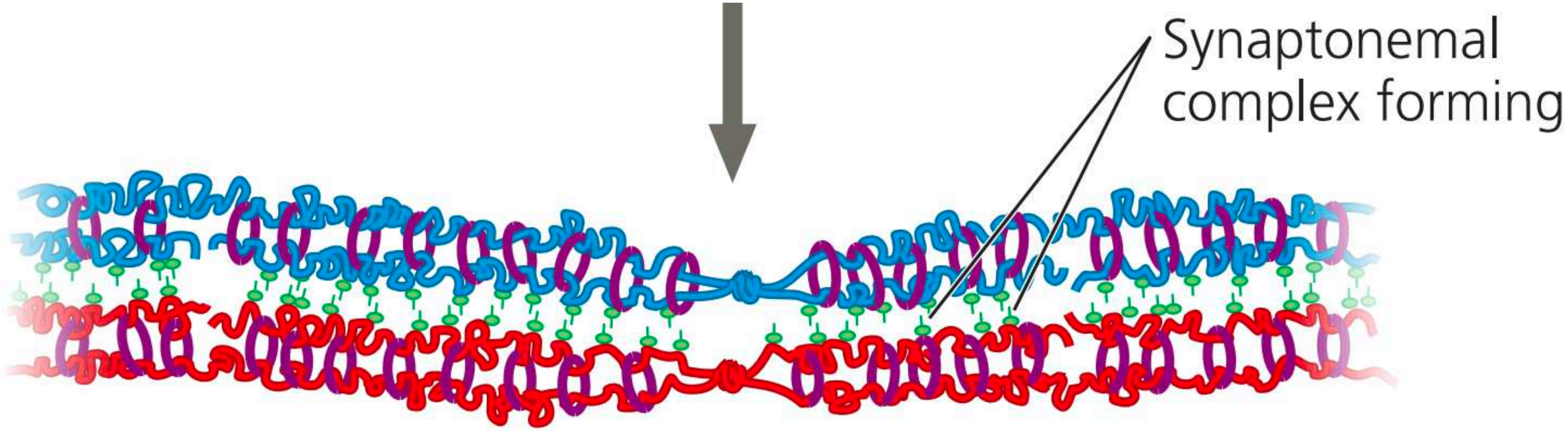

**Crossing over**



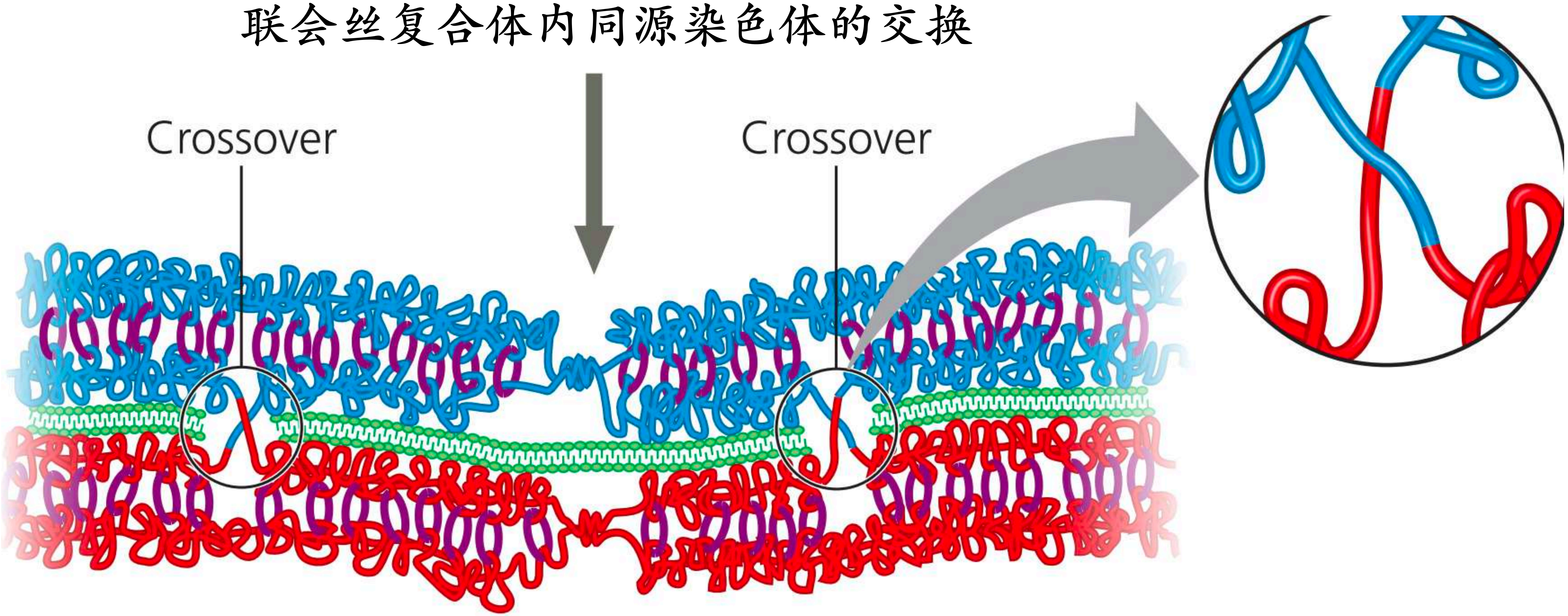
# 基因遗传连锁和交换现象的生物学基础

DNA断裂可在染色体上随机发生

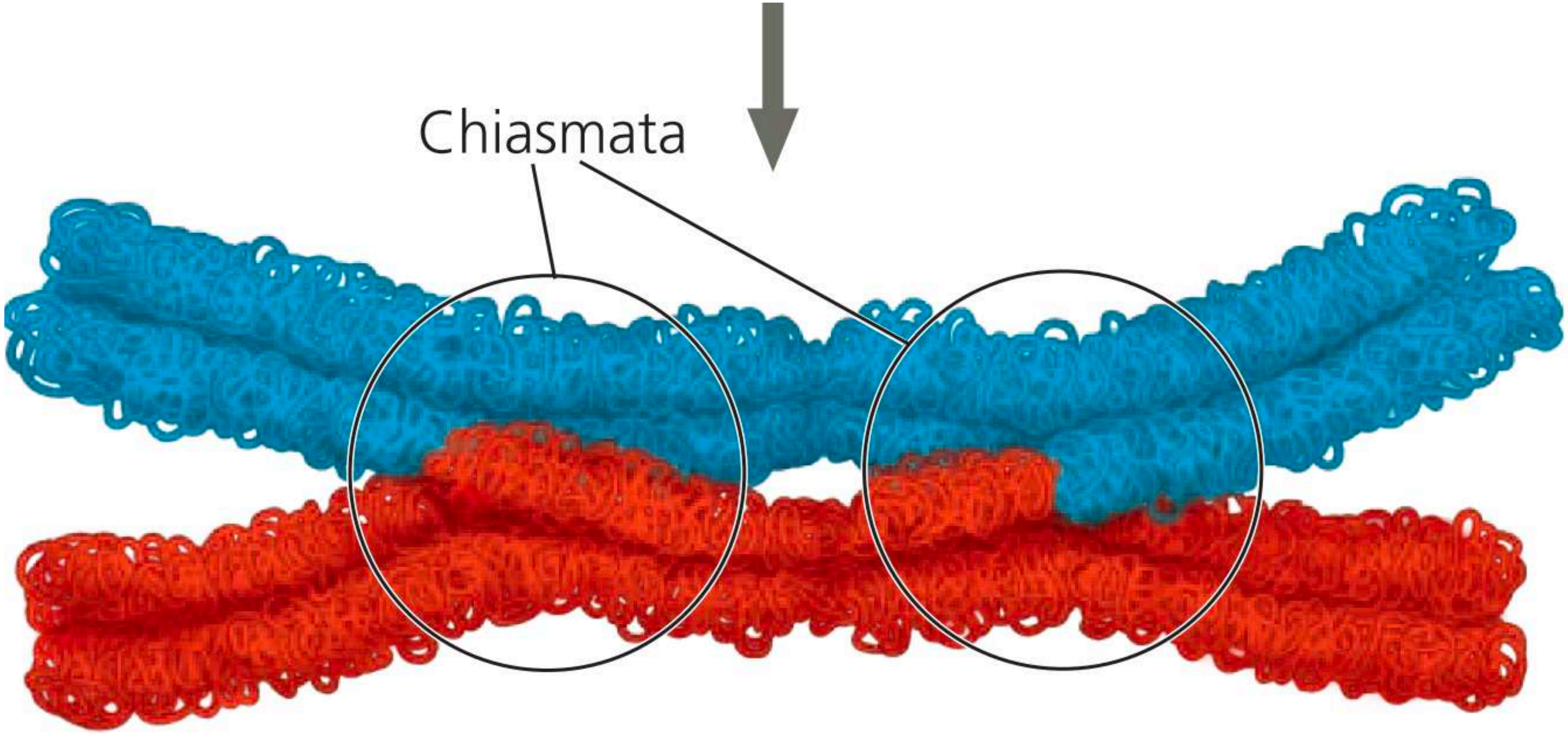





联会丝复合体内同源染色体的交换

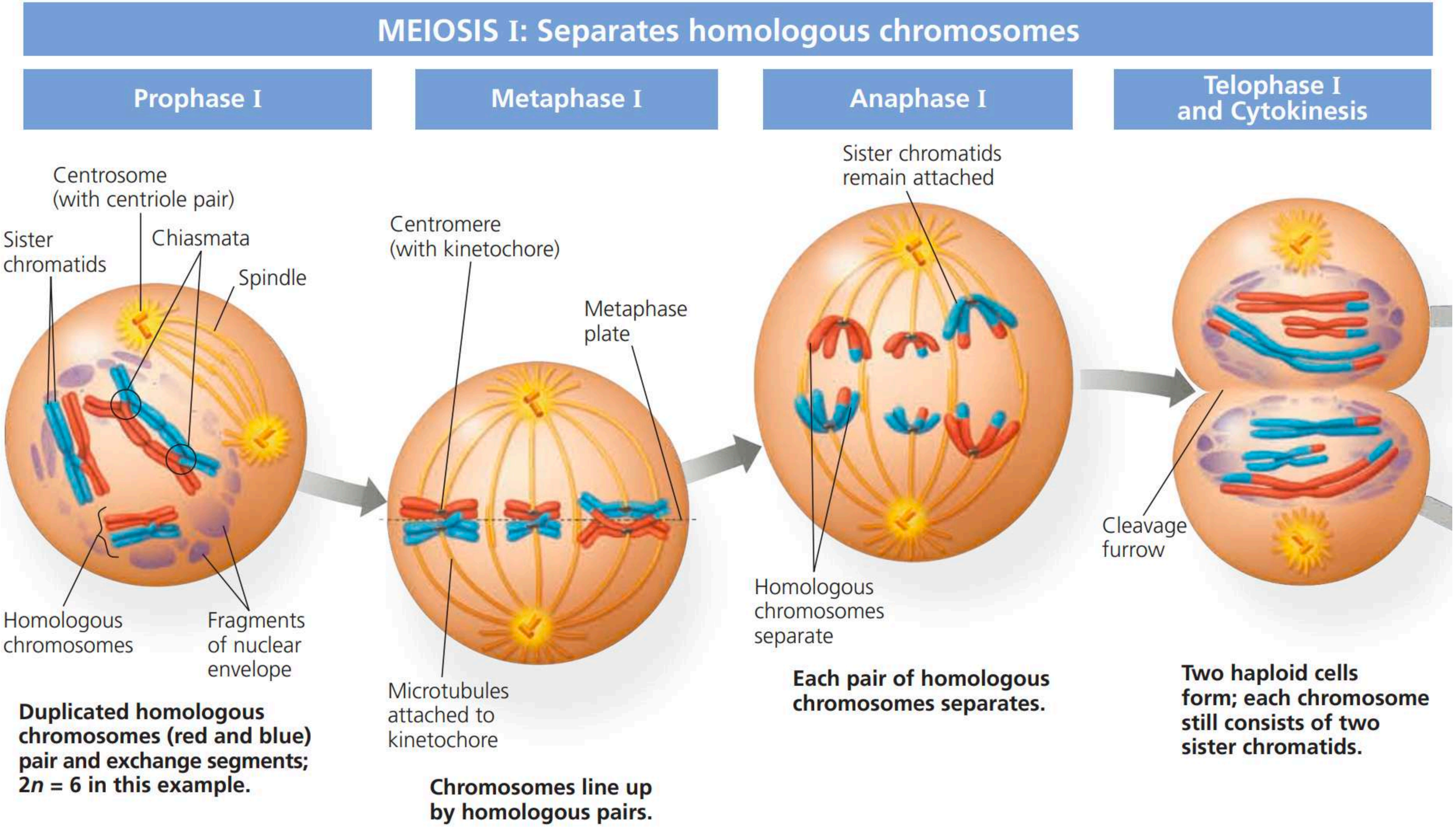


# 四分体的形成




Chiasmata (chiasma for singular) where crossovers have occurred

At least one crossover per chromosome must occur in order for the homologous pair to stay together as it moves to the metaphase I plate.



# 基因连锁和交换定律

生殖细胞形成过程中，位于同一染色体上的基因是连锁在一起，作为一个单位进行传递，称为连锁律。在生殖细胞形成时，一对同源染色体上的不同对等位基因之间可以发生交换，称为交换律与互换律。

适用于：位于同源染色体上的非等位基因。

# 环境对表型的影响

# Summary

遗传学三大基本定律

基因分离定律
基因自由组合定律
基因遗传的连锁与交换定律

伴性遗传

孟德尔

摩尔根