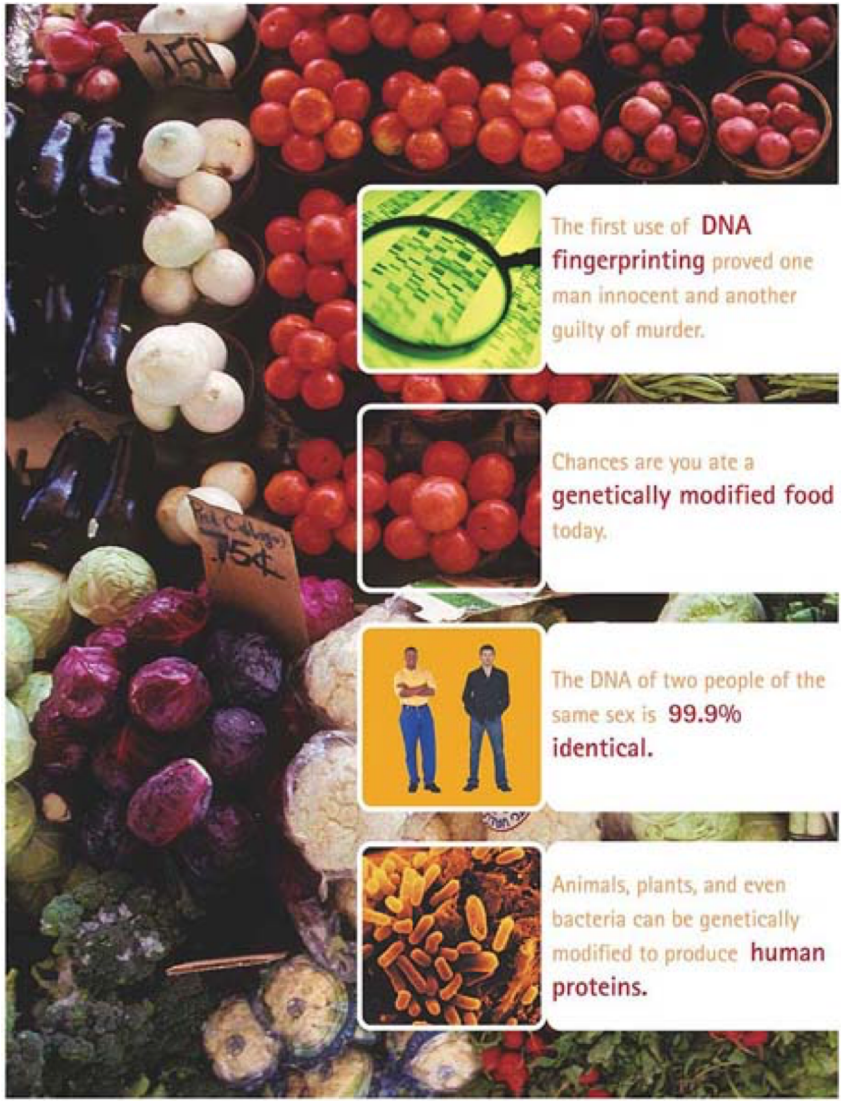
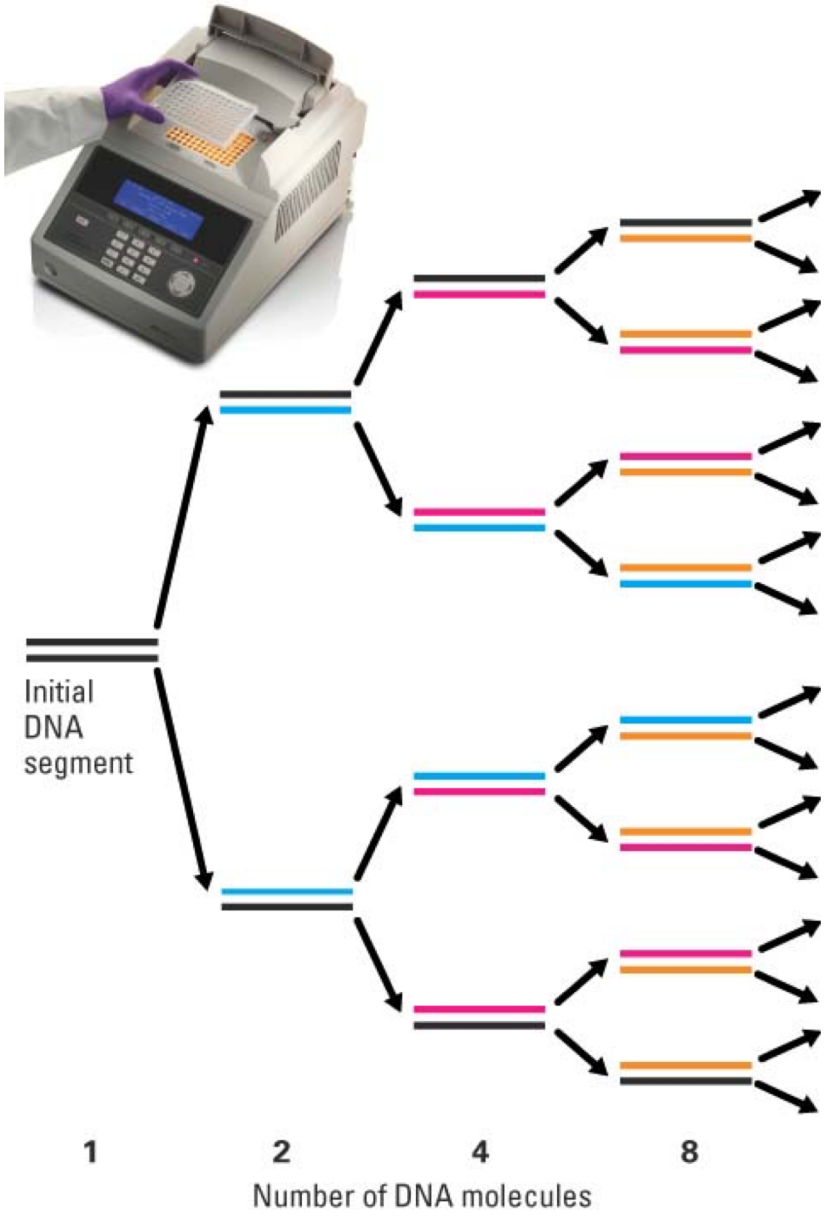
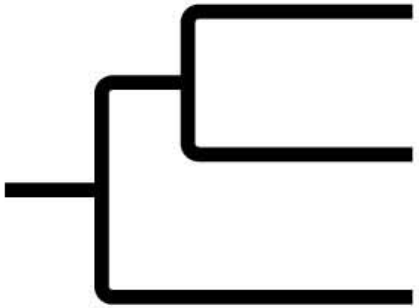
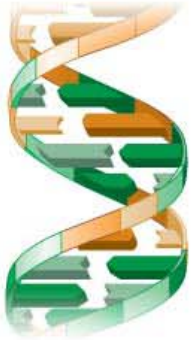
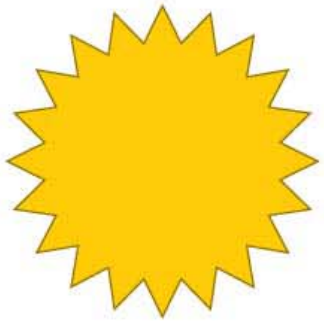
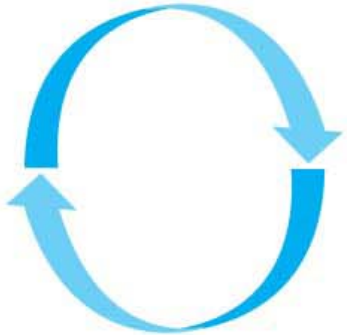
Chapter 12: **DNA Technology**





MAJOR THEMES IN BIOLOGY

Evolution	Structure/ Function	Information Flow	Energy Transformations	Interconnections within Systems
				

DNA replication (

Gene expression (semaine passée)

Gene transmission (cette semaine)

Chapter 9:
Patterns of Inheritance

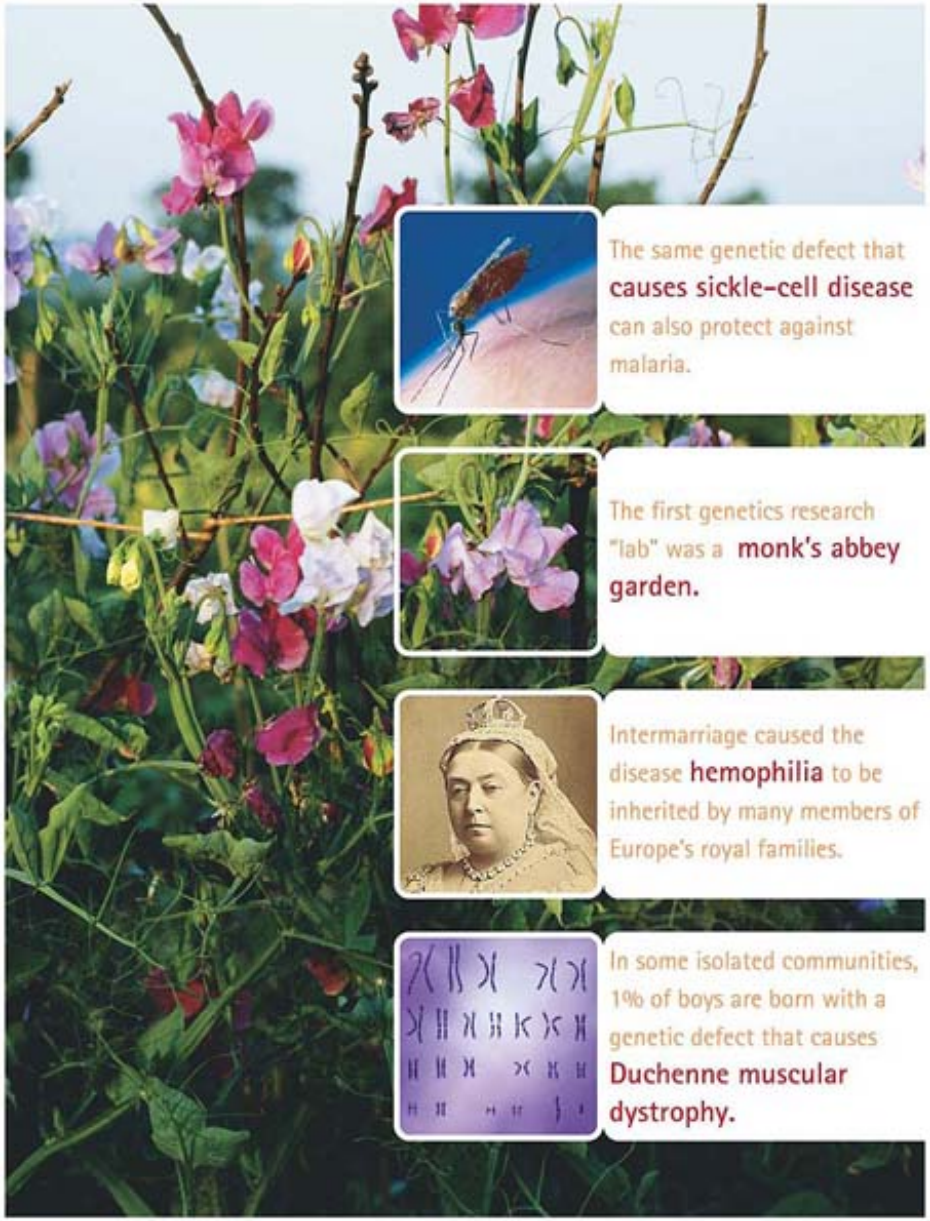
Modes de transmission

- transmission **autosomique**

  ◊ dominante
  ◊ récessive

- transmission **liée au chr. X**

  ◊ dominante (pas dans BIO109)
  ◊ récessive

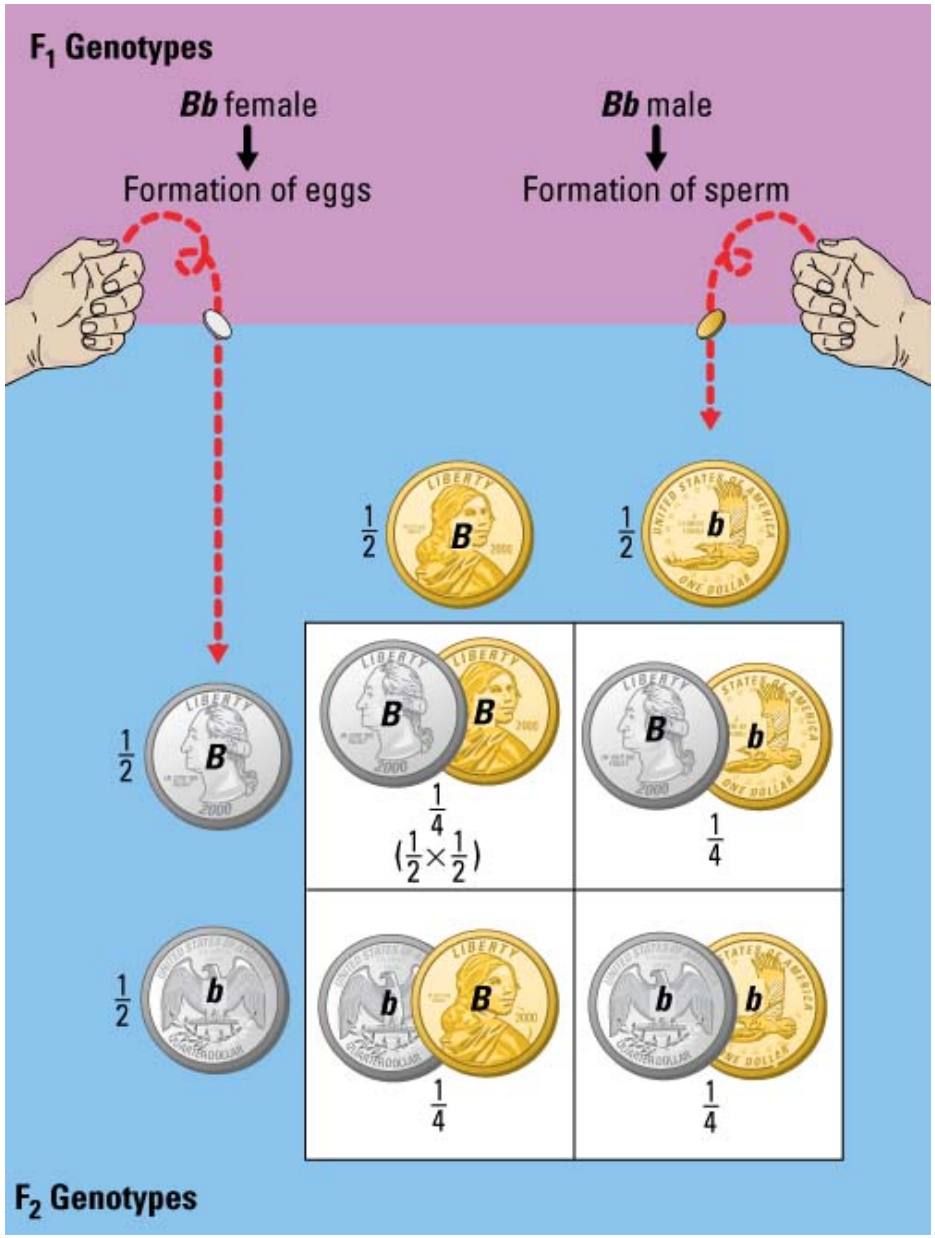


Segregation of alleles and fertilization as chance events.

Essential Biology chapter 9

Tutorial 11.1 parts 1 to 3

part 4 about **probability**



Mendel's laws of inheritance

- studied 7 traits in peas which are determined by **single genes**:
  → color of flower (purple or white)
  → shape of seeds (round or wrinkled)
  → color of seeds (yellow or green)

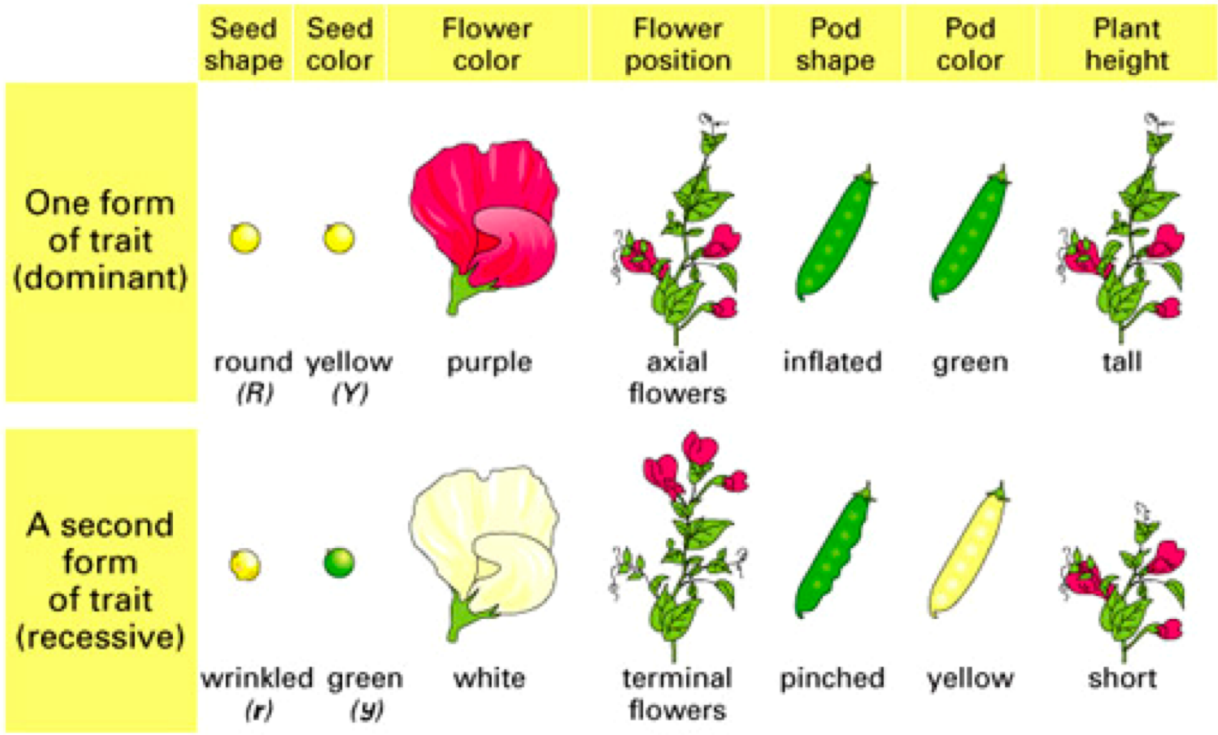


Figure 20-15 **Essential Cell Biology, 2/e. (© 2004 Garland Science)**

Tutorial 11.1 parts 1 to 3

part 4 about **probability**

Mendel's crossing of genetically pure pea plants

Genetically pure = Mendel's words for homozygous

- cross: yellow-seeded x green-seeded:
  100% are yellow seeded in F1

→ inheritance NOT **by blending** of parents' traits

→ NOT uniparental: independent if
  yellow-seeded as pollen or as flower

- self-crossing of F1: in F2: 75% yellow, 25% green

→ "green seeds" is not lost in F1

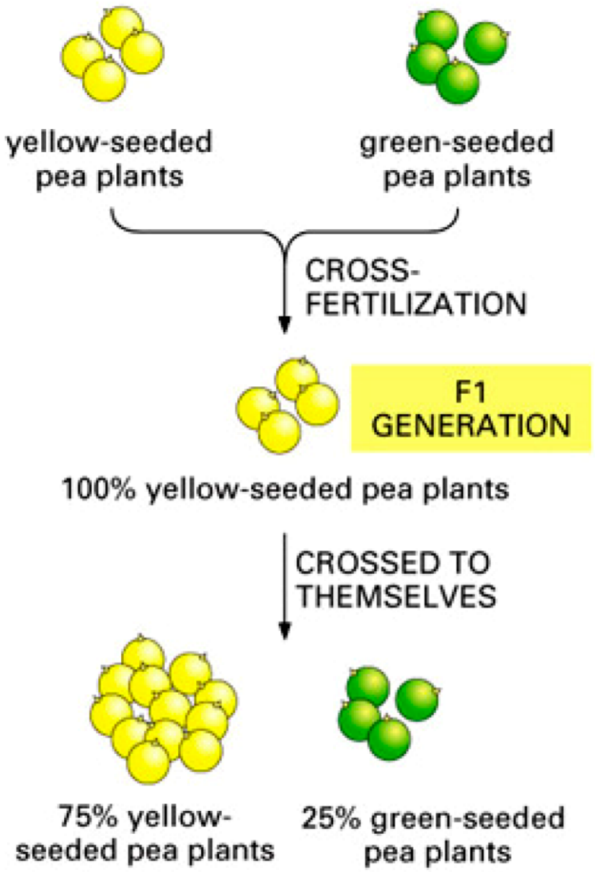
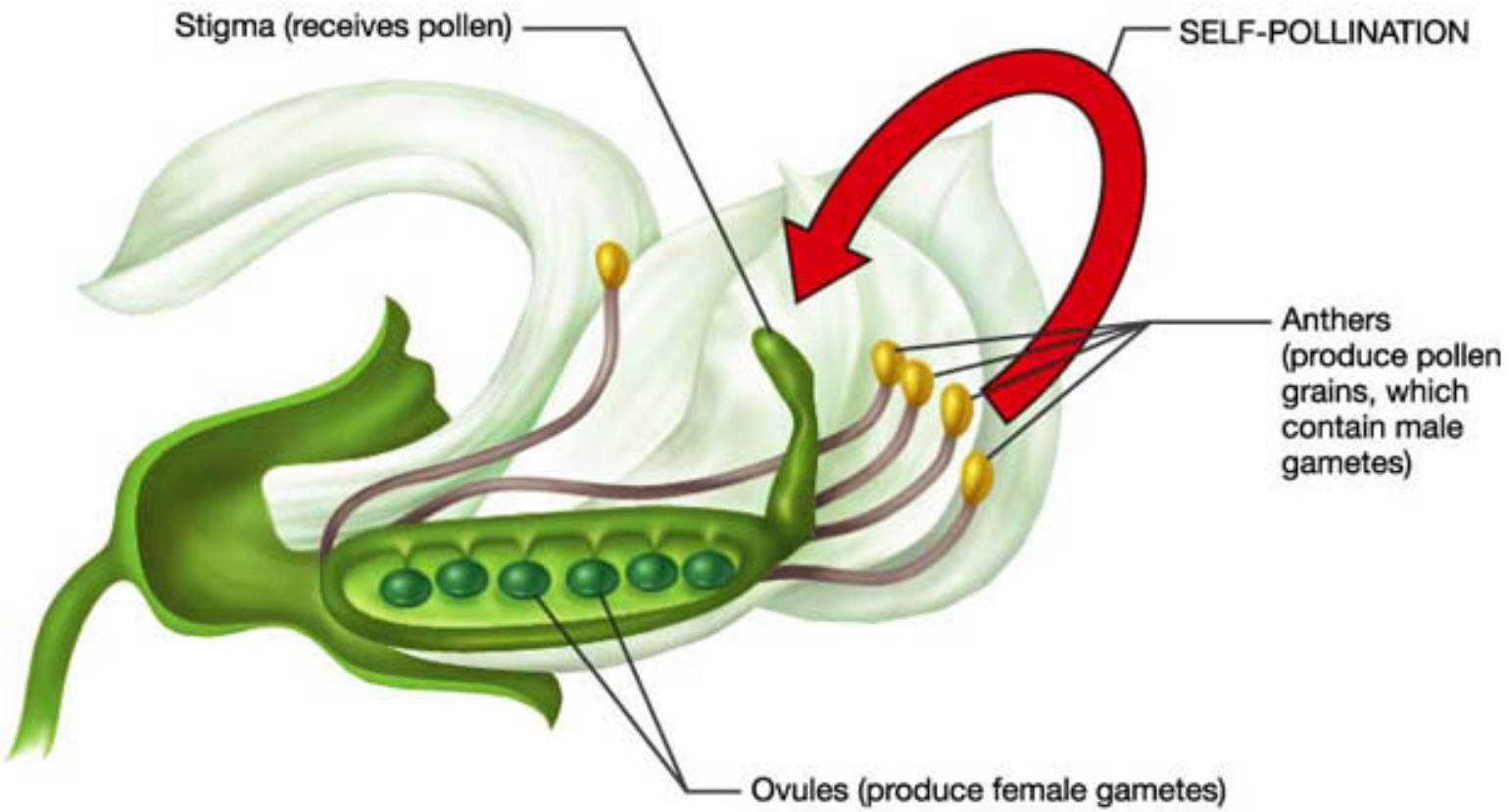


Figure 20-16 Essential Cell Biology, 2/e. (© 2004 Garland Science)

Self-pollination

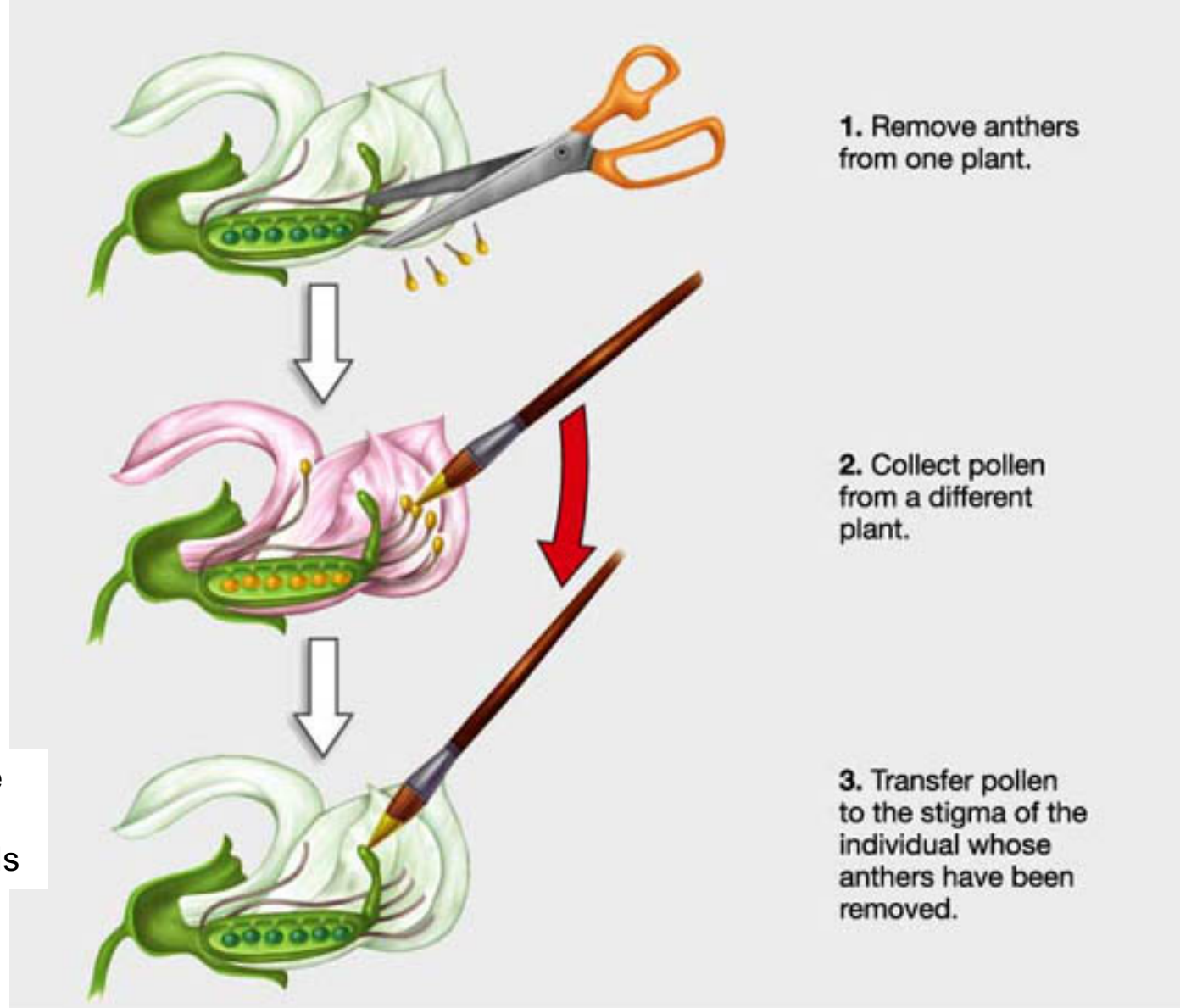
Possible in peas; not possible in all plants



Cross-pollination is time consuming

Cross-pollination

(time consuming)



To observe the phenotype of F1 :
grow plants from the seeds

Mendel suggested that:

→ traits are determined by distinct "hereditary factors" (-> genes)

→ these factors come in different varieties (yellow or green seeds -> alleles)

→ a plant must inherit 2 copies of each factor (one from pollen, one from oocyte)

→ phenotype: the characteristic one observes (seeds are either yellow or green)

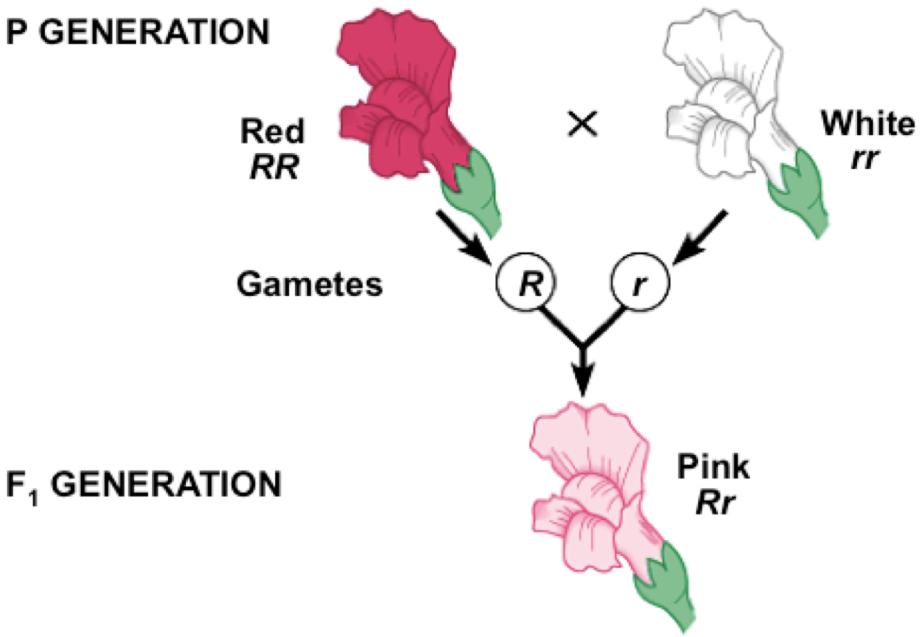
→ genotype: the actual combinations of varieties in the 2 copies

→ homozygous: both copies are of same variety (for yellow or for green)

→ heterozygous: one copy for yellow, one copy for green

→ one variety is **dominant** over the other **recessive** variety

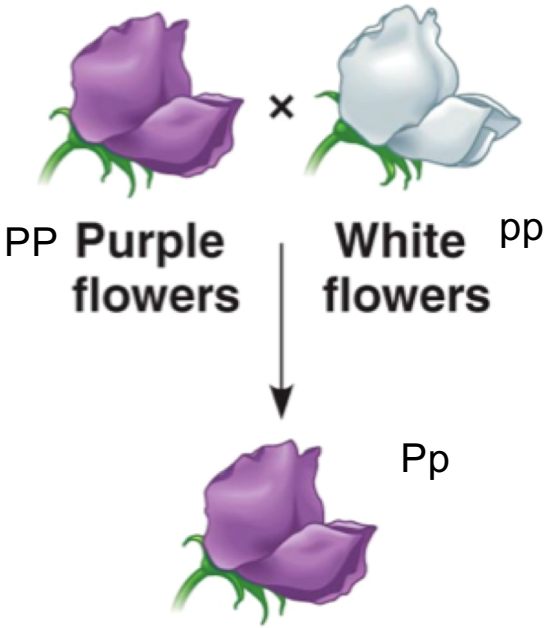
→ Mendel's law of segregation



Incomplete dominance of red over white in snapdragons.

Intermediate phenotype in heterozygotes.

See Tutorial 11.1



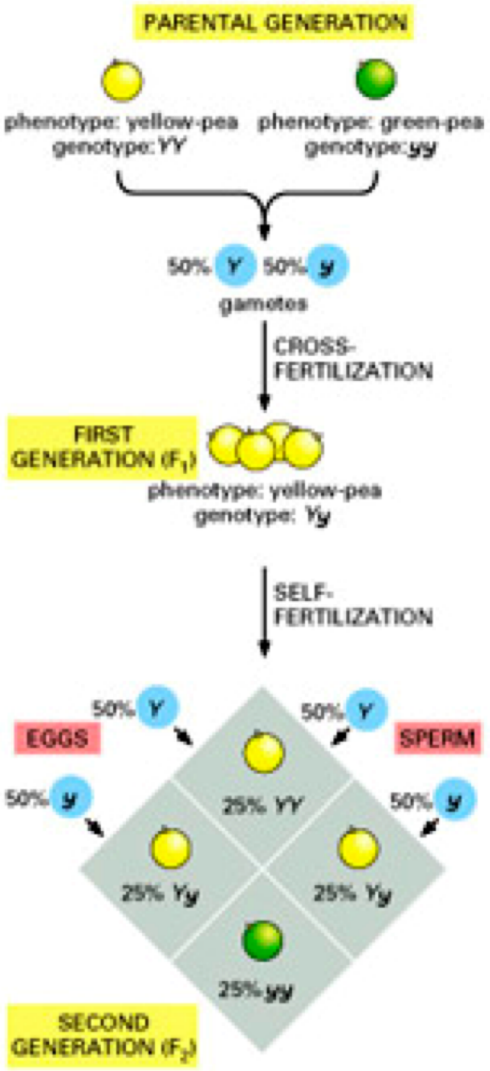
Complete dominance of purple over white in peas.

The concept of incomplete dominance was unknow to Mendel.

Mendel's Law of segregation

- gametes from F1 plants: each only one allele
  → 50% green, 50% yellow
- after self-crossing:
  25% green-green -> green
  25% yellow-yellow -> yellow
  50% yellow-green -> yellow
- 3:1 ratio (phenotype)
- applies to all sexually reproducing organisms!

Monohybrid cross : 1 trait



Mendel's Law of independent assortment

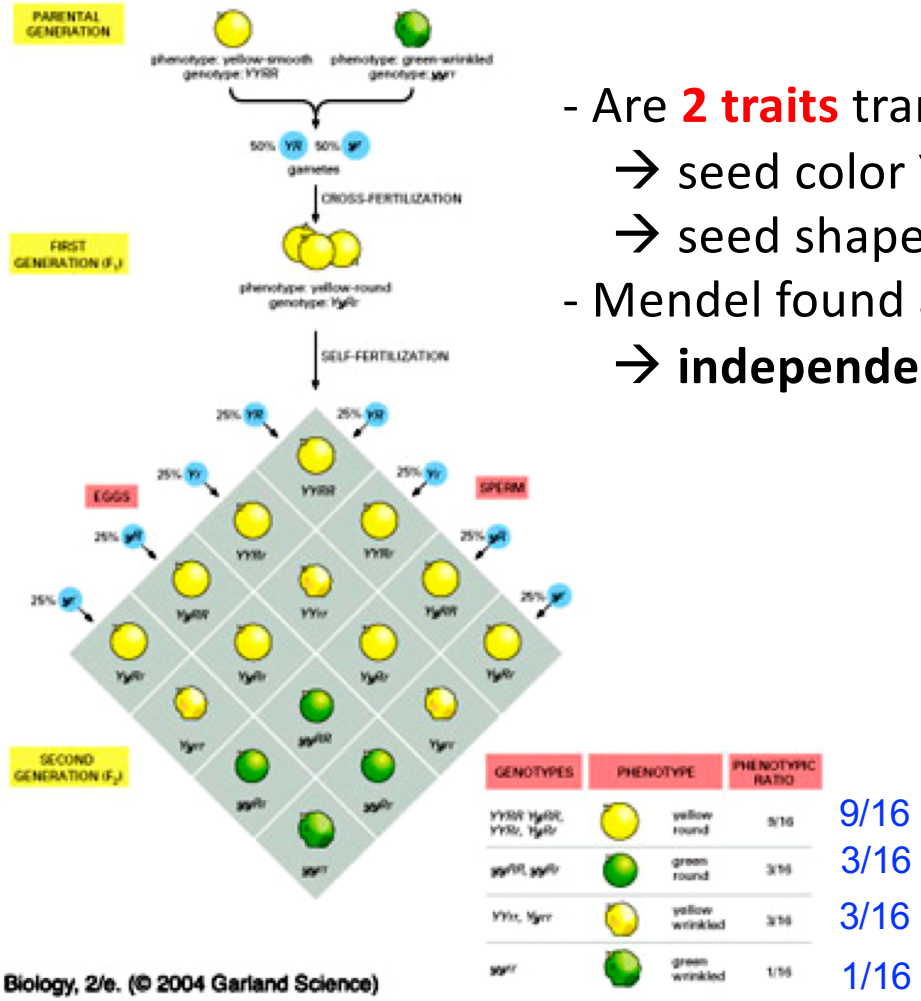


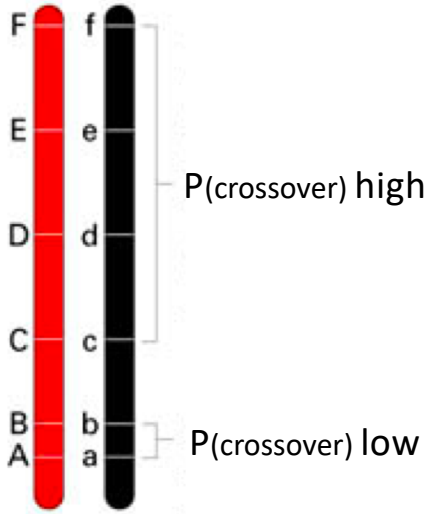
Figure 20-20 **Essential Cell Biology, 2/e. (© 2004 Garland Science)**

- Are **2 traits** transmitted independently from each other?
  - → seed color Y or y (YELLOW or green)
  - → seed shape R or r (SMOOTH or wrinkled)
- Mendel found all 4 combinations; ratio 9:3:3:1
  - → **independent assortment**

Dihybrid cross : 2 traits

Independent assortment of genes

- **independent assortment** of genes, if they are:
  - → on separate chromosomes
  - → on same chromosome but **far apart**
    - → cross-over during meiosis
- linkage studies to identify genes responsible for genetic diseases :
  - → follow segregation of a disease phenotype with marker phenotypes of known location on chromosomes
  - → the "less independent" the assortment, the closer the gene and the marker



All 7 traits studied by Mendel were unlinked.
Linkage was discovered by T. H. Morgan.

Mendelian laws apply to other diploid multi-cellular organisms, including us.

A gene encodes enzyme needed for melanin production.

(tyrosinase)

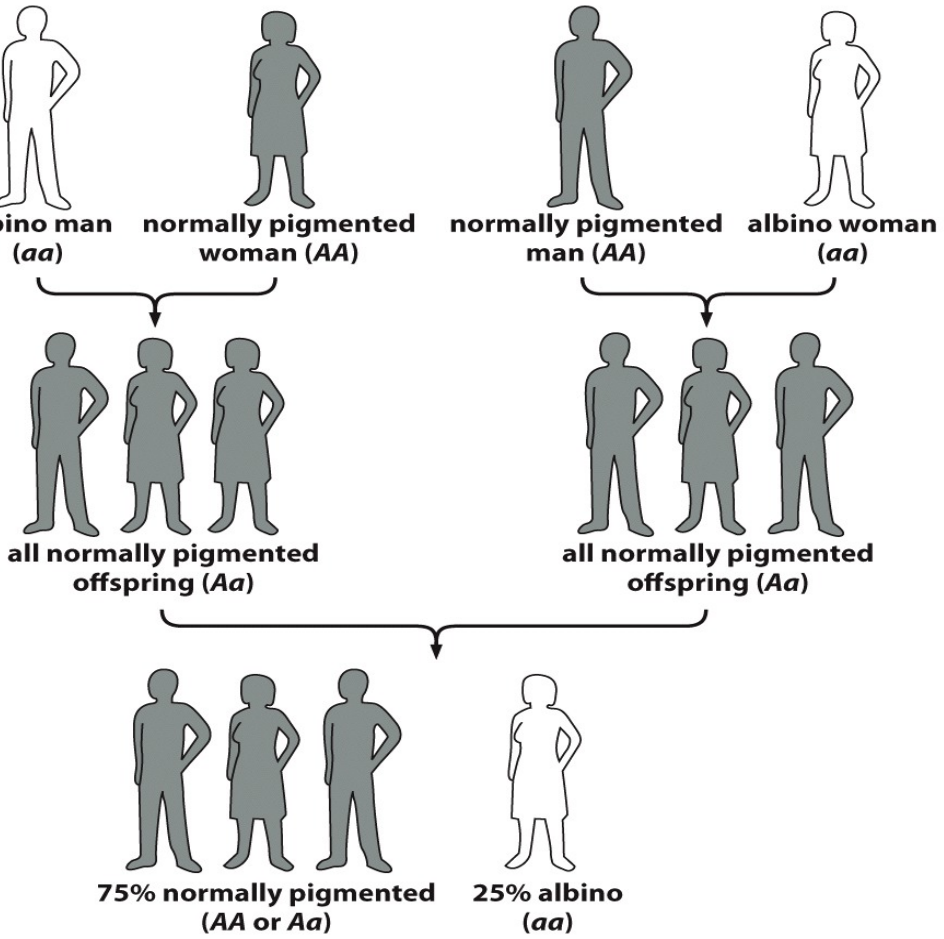


Fig. 19-25 3 : 1

Albinism is an **autosomal recessive** phenotype.

Because A is completely dominant, impossible to know whether he is a
carrier (A/a)
or not (A/A)
without a molecular analysis.

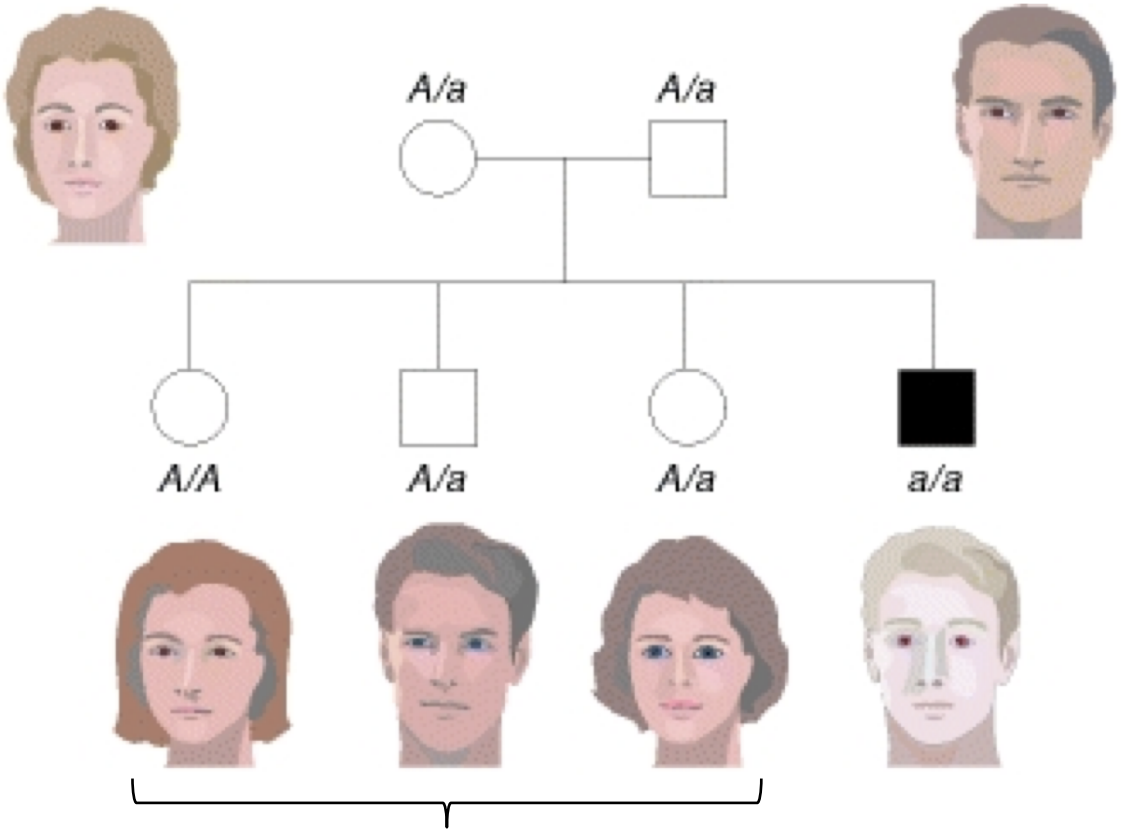
(real family)

Matthew
14 ans

Ben
7 ans

Jessica
11 ans

Albinism : ***complete dominance*** of the normal phenotype.



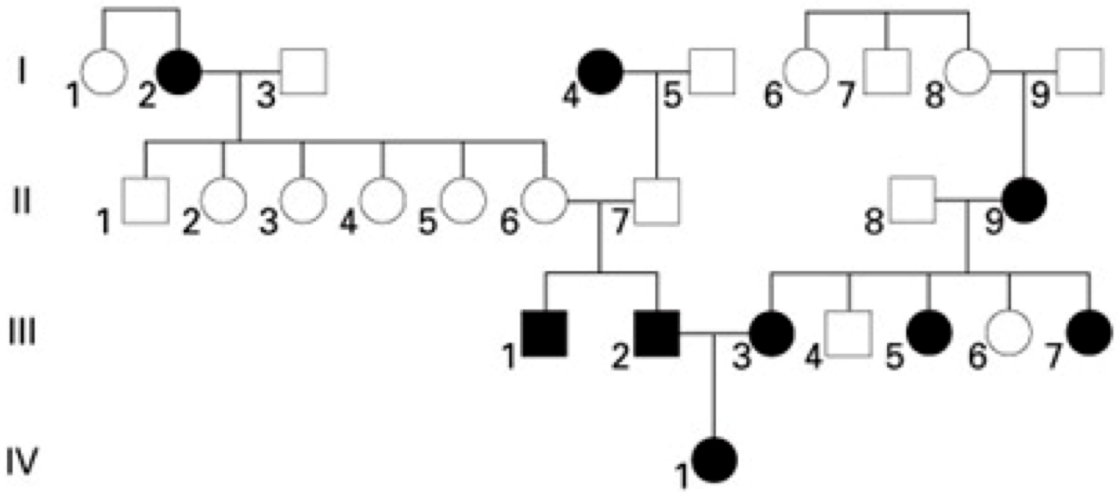
Phenotypes completely identical.

Pedigrees help to follow phenotypes in families

Conventions :

circles:	females
squares:	males
filled:	affected
open:	unaffected

Tutorial 12.2, part 3



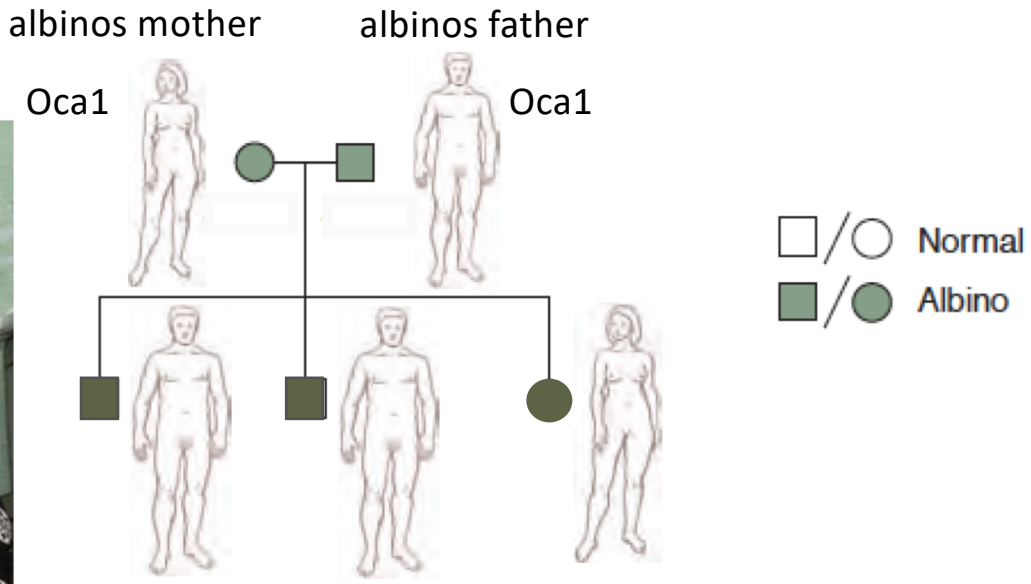
Analysis of this pedigree allows you to find out that the character is transmitted on the mode autosomic recessive.

autosomic i.e. not sex-linked (gene is not on the X chromosome)

recessive : II.6 and II.7 have affected children

Figure 20-19 Essential Cell Biology, 2/e. (© 2004 Garland Science)

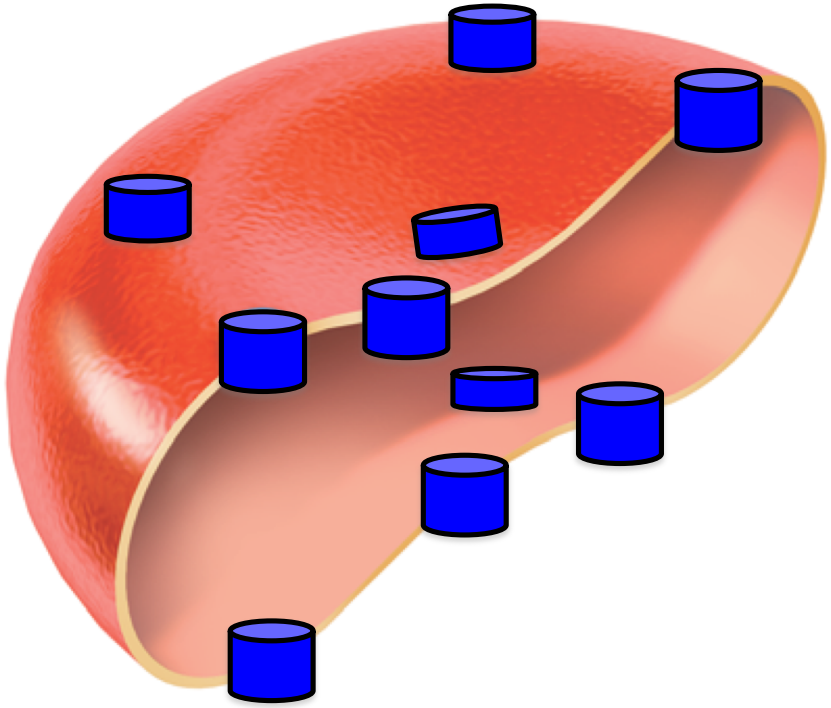
Real life example : the Pullan familly



oca = oculo cutaneous albinism

Globules rouges

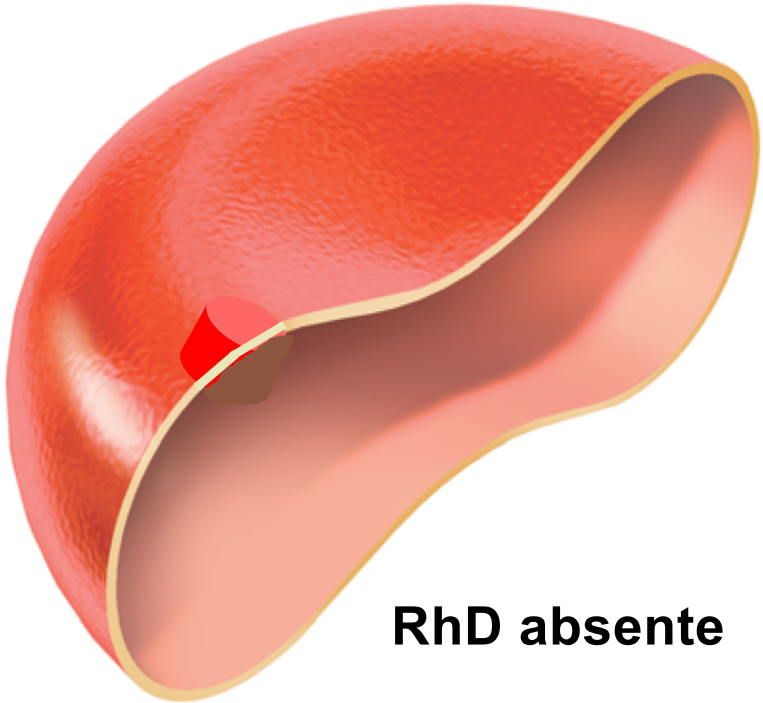
Rhésus positif



RhD

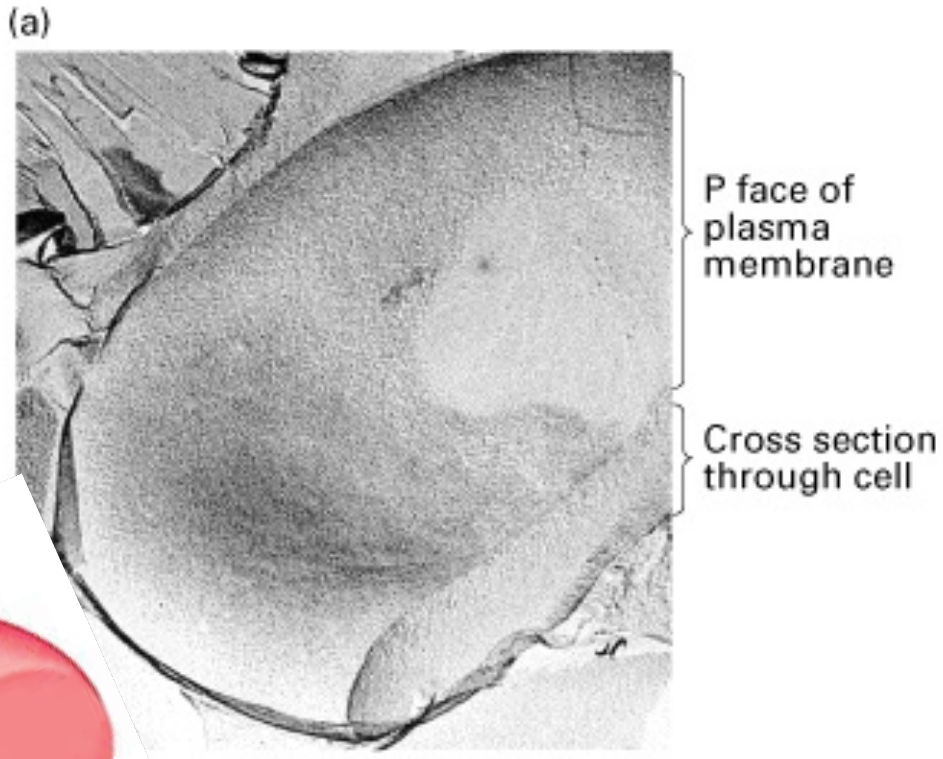
protéine transmembranaire
(transporteur)

Rhésus négatif



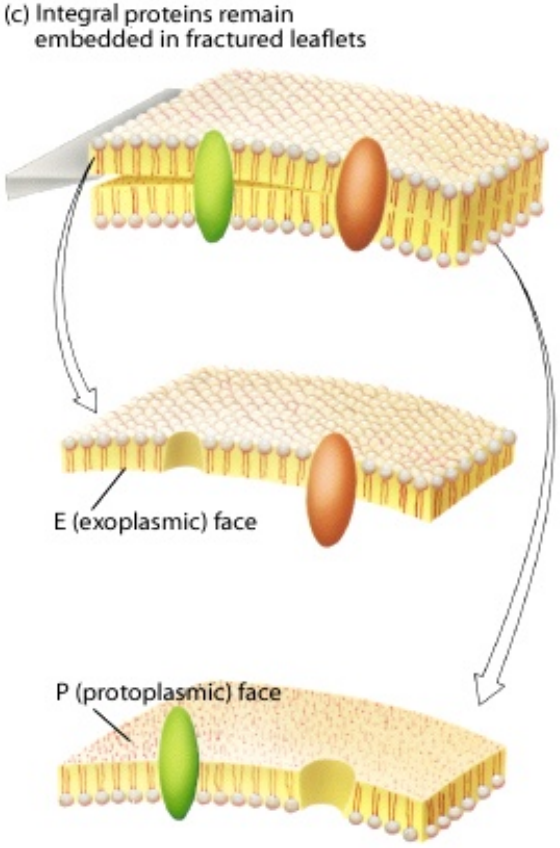
RhD absente

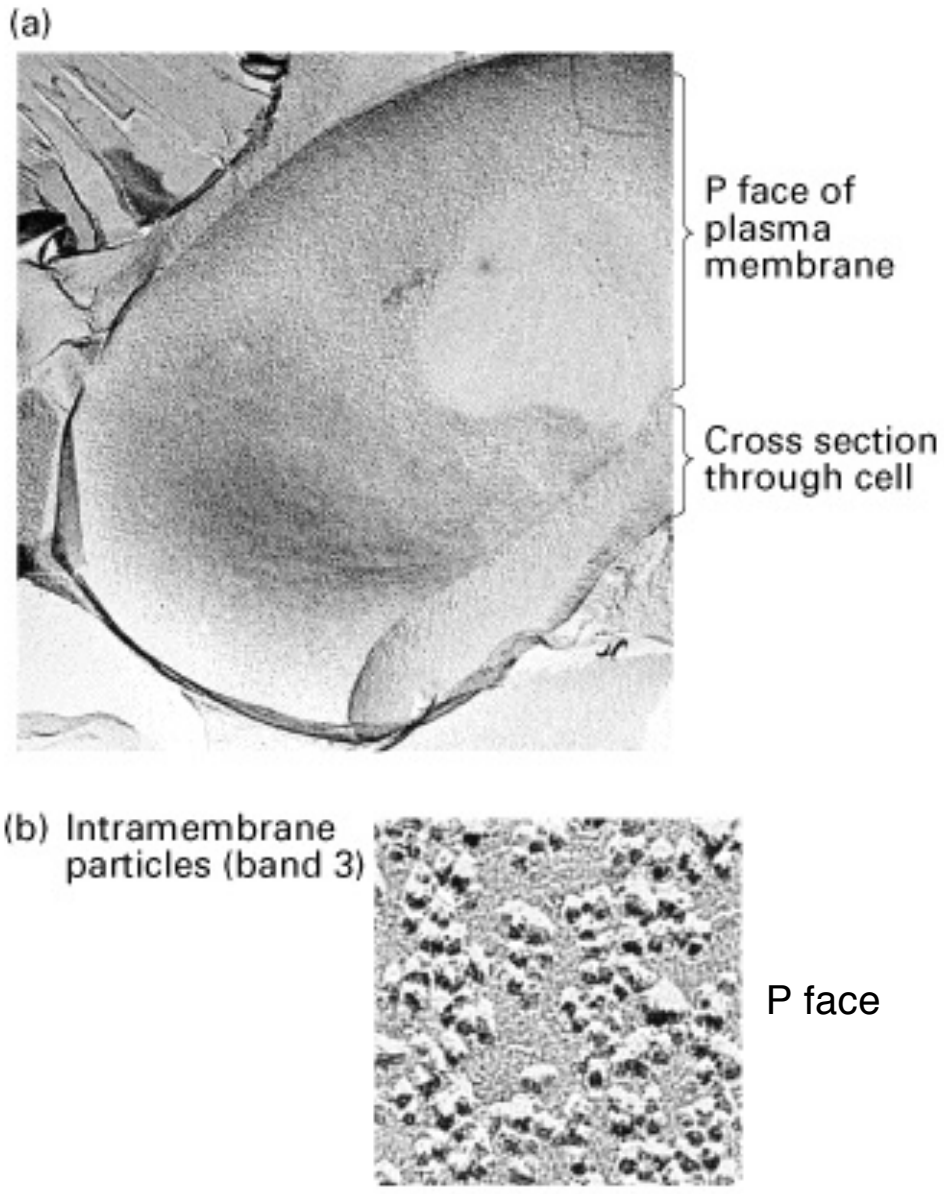
Cryofracture d'un globule rouge :



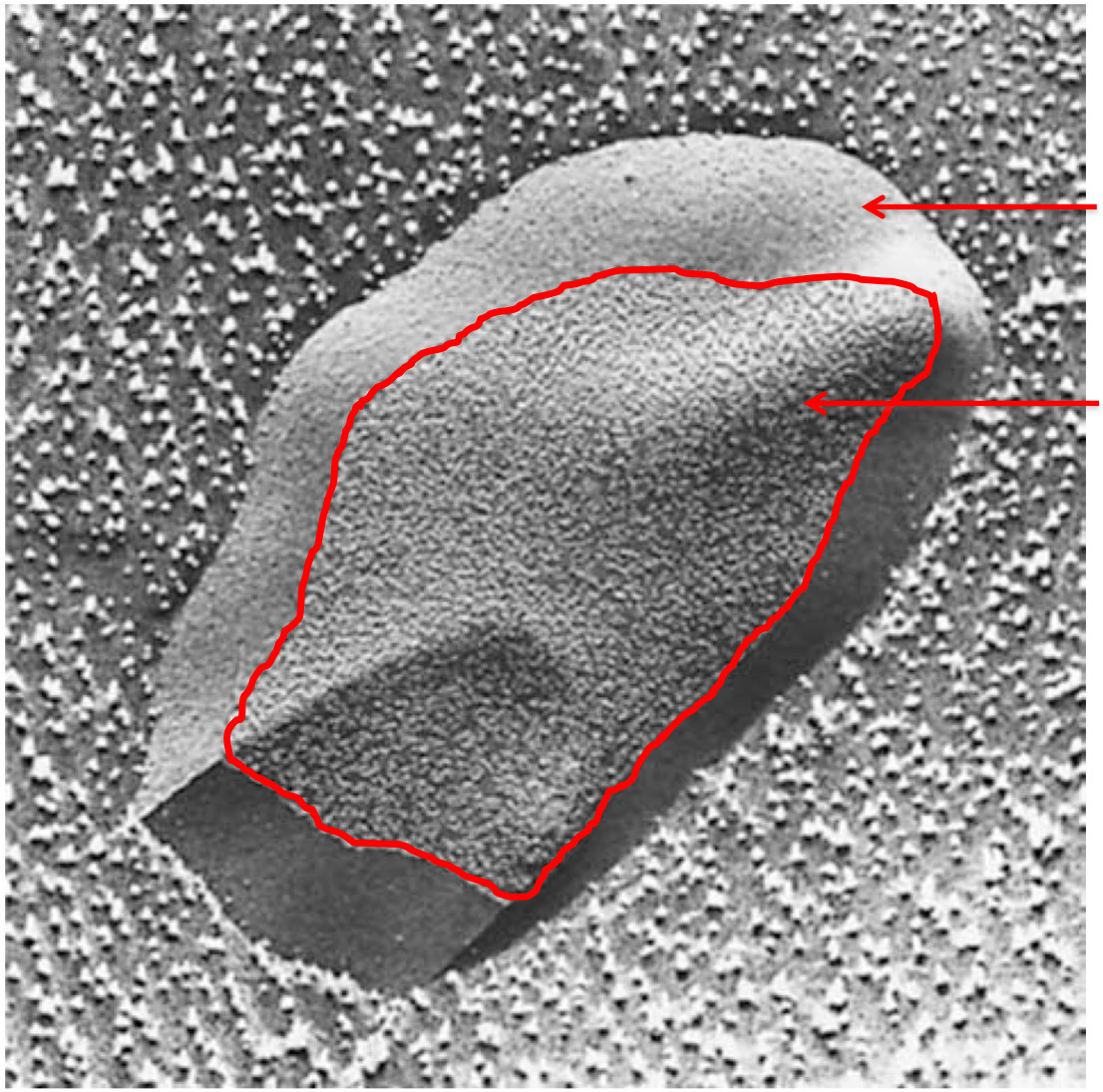
Les protéines transmembranaires deviennent visibles après l'enlèvement du feuillet externe de la membrane.

Cryofracture d'un globule rouge :





Cryofracture d'un globule rouge :

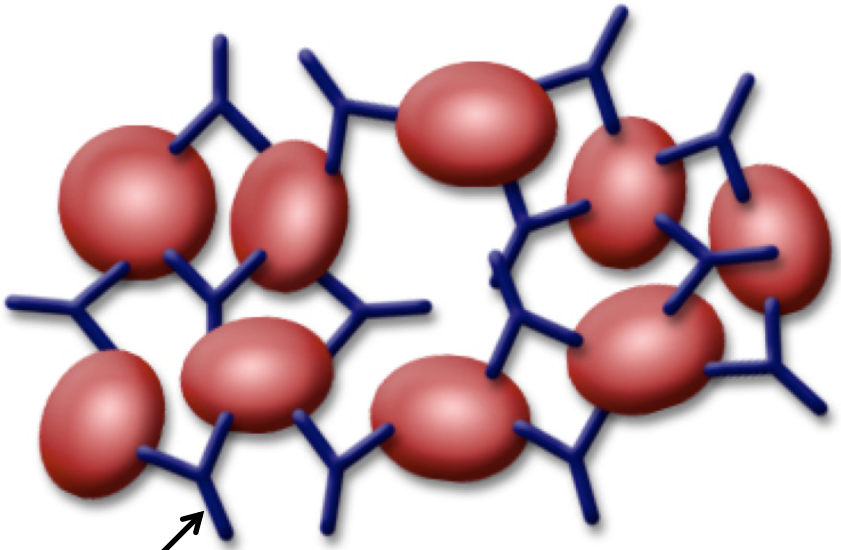


surface externe lisse

face P
protéines transmembranaires visibles.

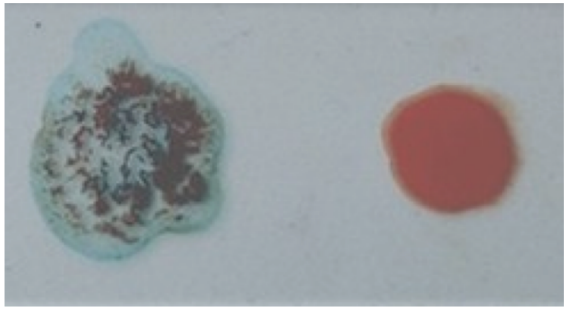
Agglutination

(hémagglutination)

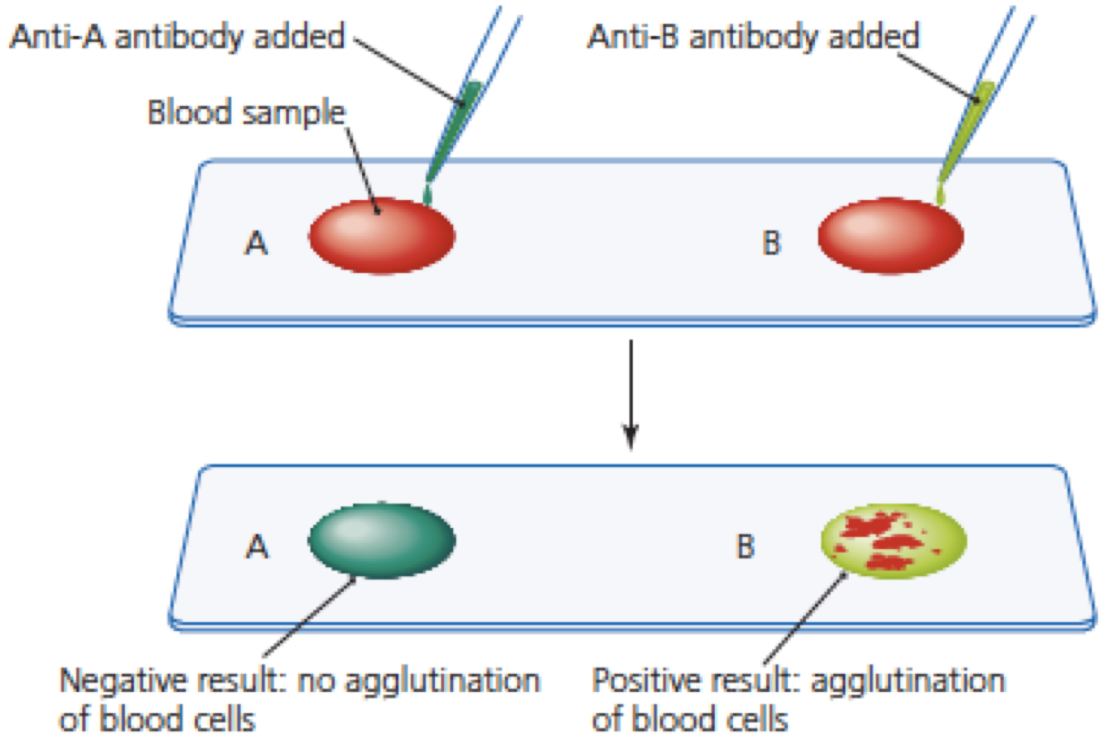


Les anticorps sont divalents

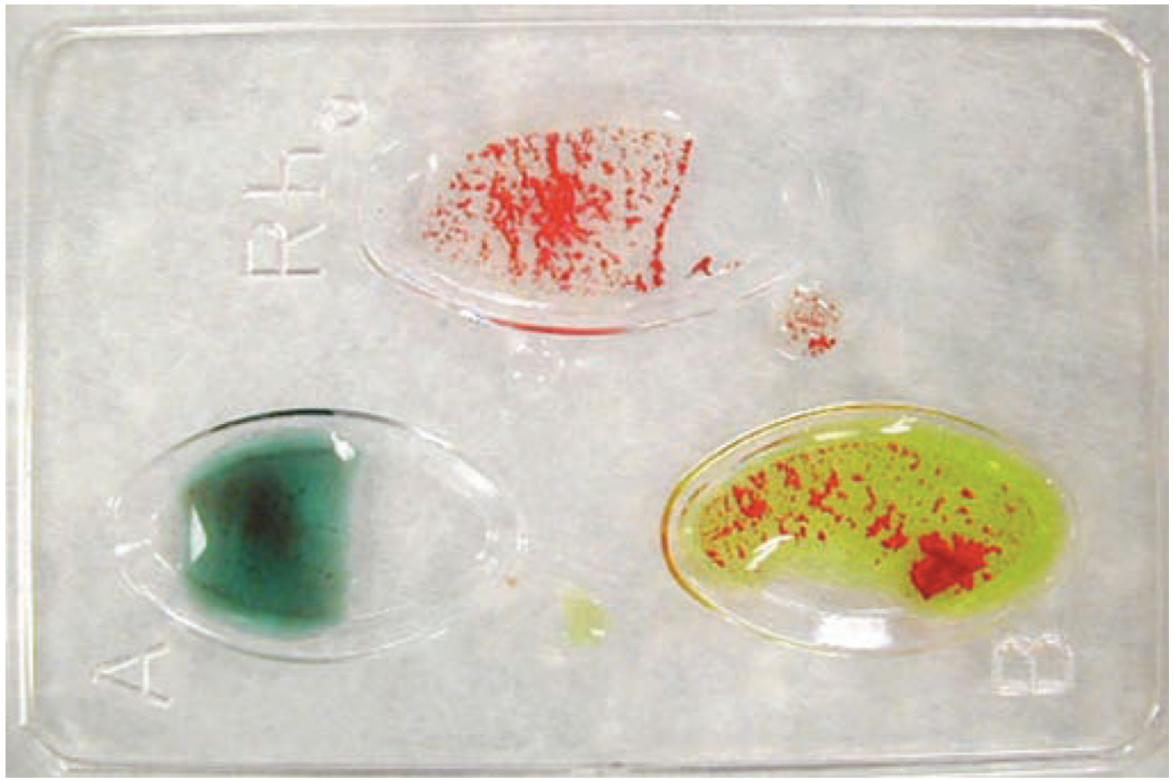
(lient deux antigènes identiques)



Groupage ABO par hémagglutination



Groupage ABO par hémagglutination



Des colorants sont ajoutés pour que les erreurs soient remarquées

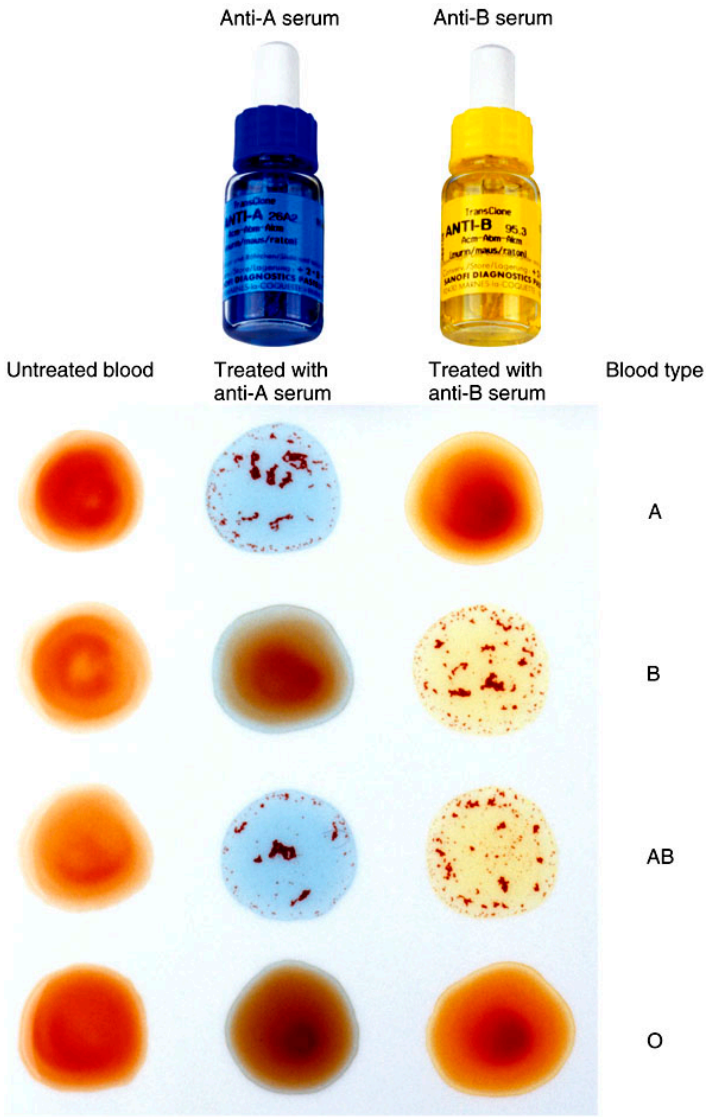


Figure 19.14 Tortora - PAP 12/e

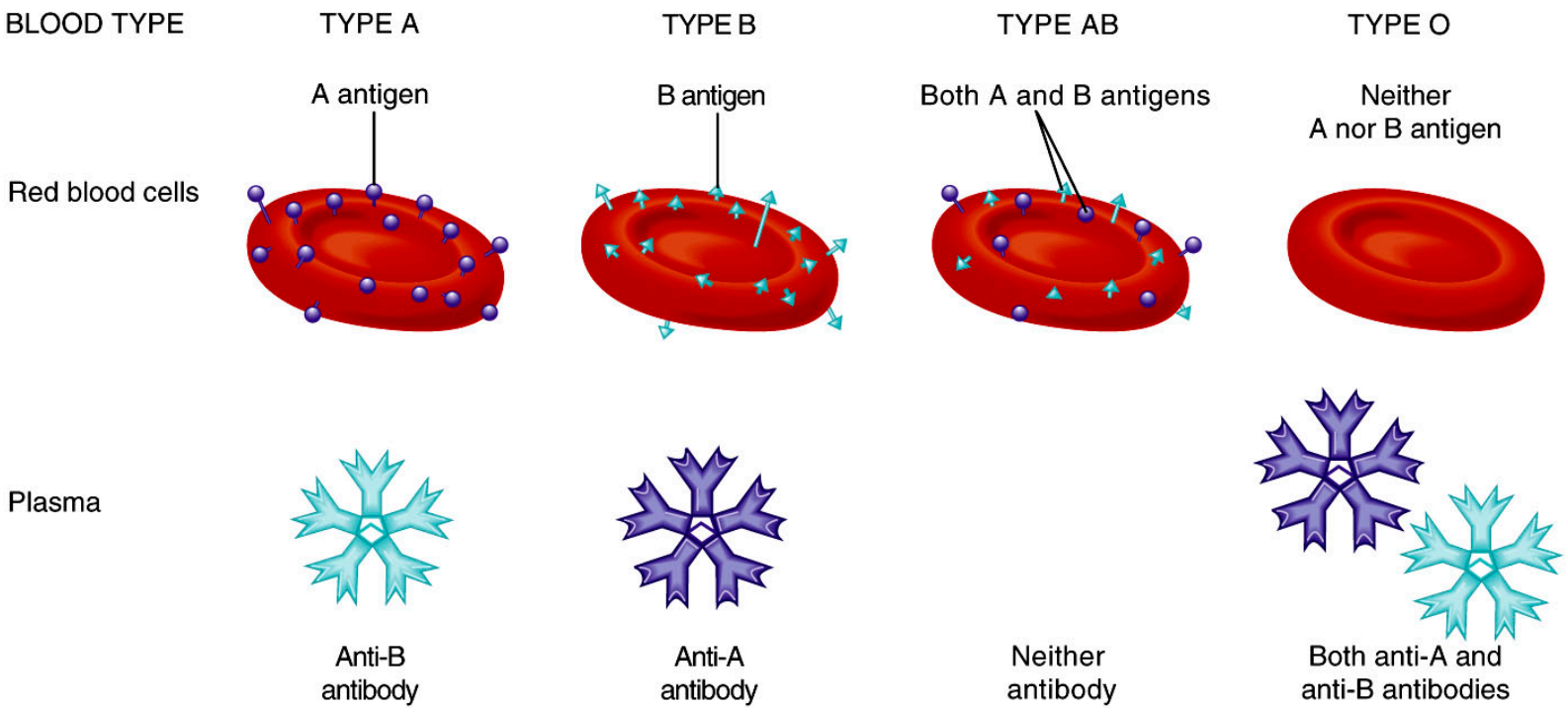
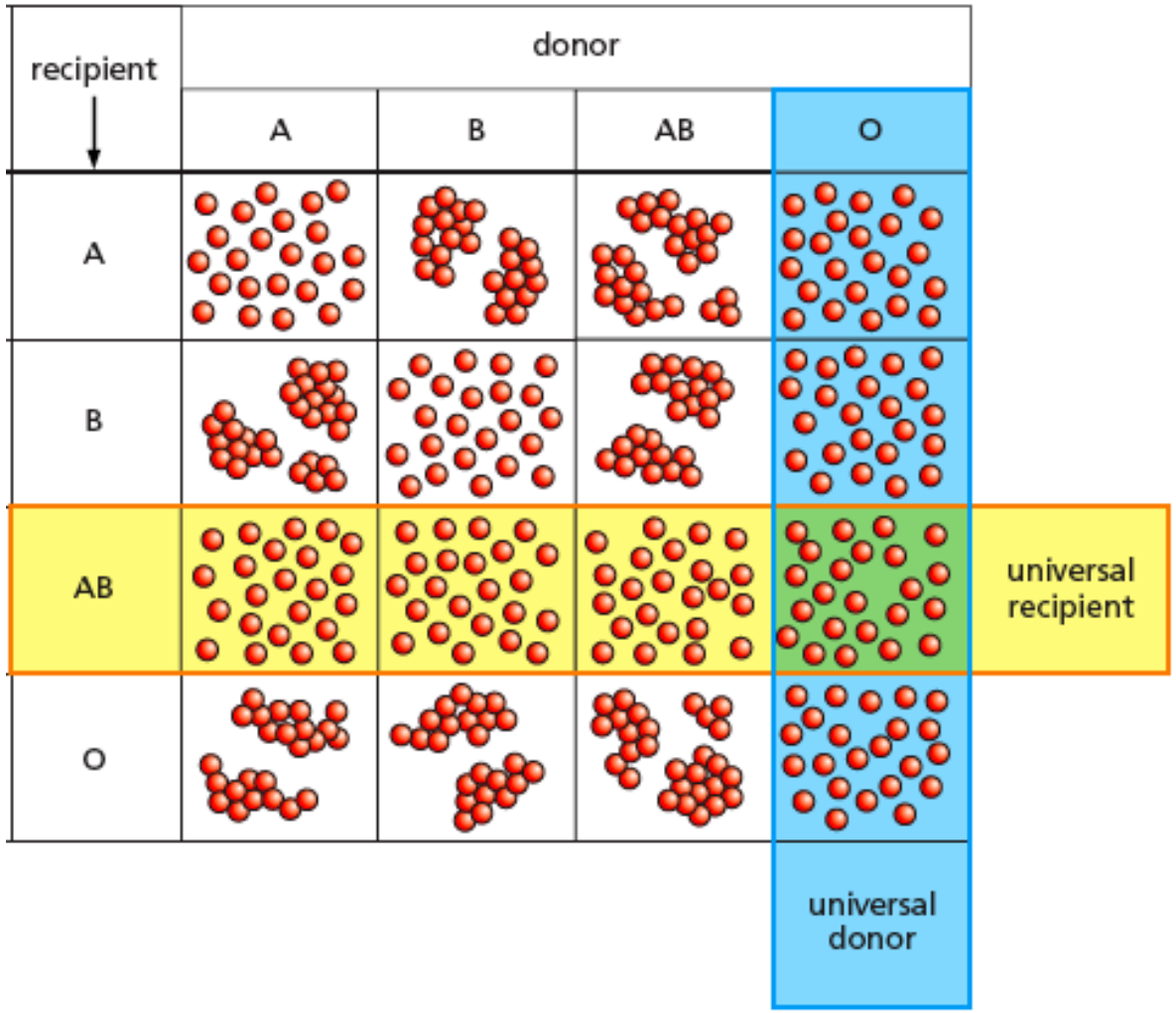
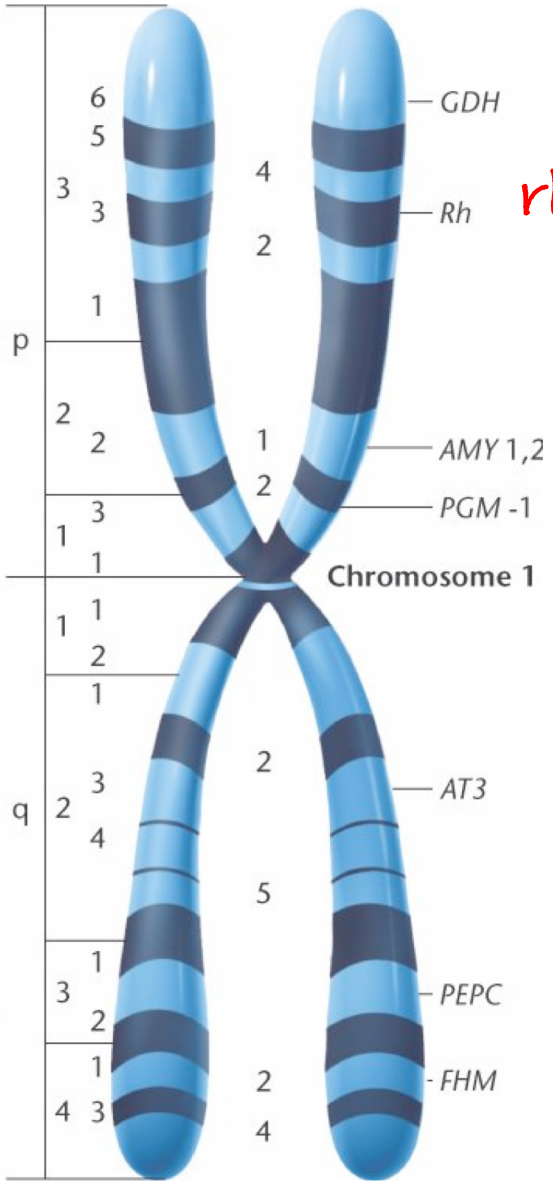


Figure 19.12 Tortora - PAP 12/e

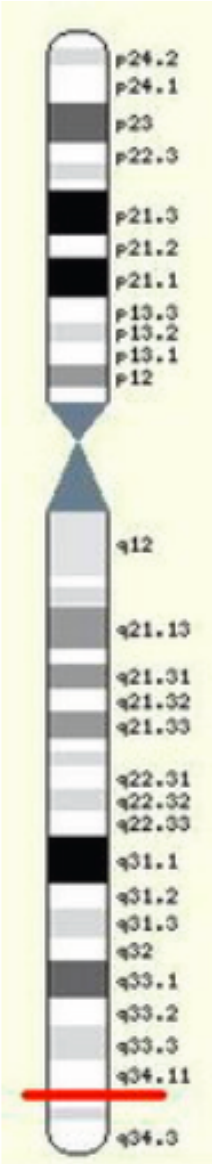

recipient
donor
A
B
AB
O
A
B
AB
O
universal recipient
universal donor

Chromosome 1
(humain)

bras court



rhésus



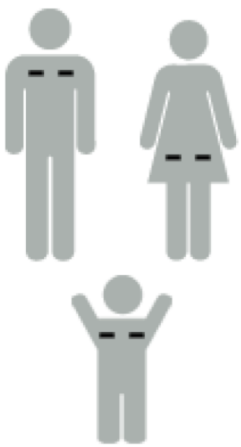
The A B O locus

Chromosome 9

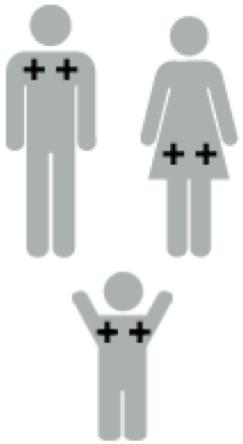
The gene encodes a glycosyl transferase :

3 allels

Transmission héréditaire : le phénotype Rhésus négatif est récessif



100%



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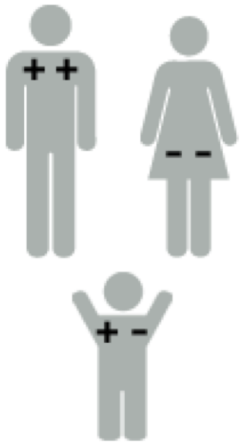
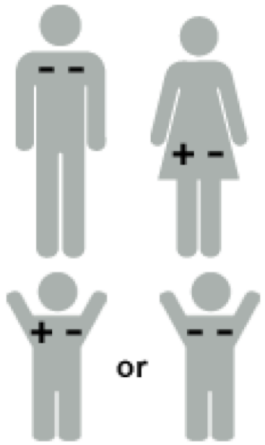


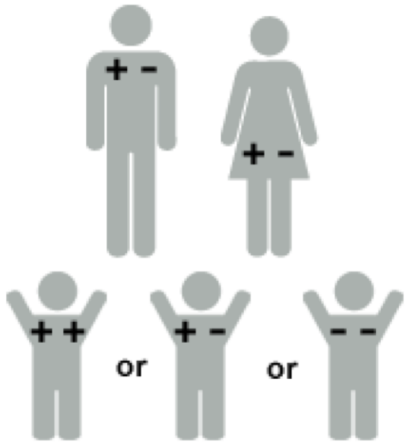
100%

génétique mendélienne

Transmission héréditaire : le phénotype Rhésus négatif est récessif



50% 50%



25% 50% 25%

génétique mendélienne