Lesson 04: Genetics of the diploids

1. General

a. Vocabulary

Haploid : individual having only one set of chromosomes

Diploid : an individual possessing two copies of each of the species' chromosomes.

Phenotype : the set of observable structural and functional characteristics (both physical and chemical) of an organism. It can be quantifiable (e.g., tall height) or qualifiable (e.g., hair color). It is the visible expression of the genotype in a given environment.

Genotype : the set of alleles of an individual. The alleles of the genes of a living organism are normally found in all members of its species.

Genome : in the broad sense, the sum of an individual's genetic information, but more specifically, it is the set of genes or coded information of genetic material.

An allele: a variant of the DNA sequence located at a specific locus in a chromosome; it is one of the alternative forms of the same gene. In a diploid cell, there are two alleles for each gene (one allele from each parent, though they can be identical). In a population, there may be several alleles of a gene.

Locus: the physical site where a DNA sequence (coding or non-coding) is located on a chromosome.

Loci: plural of locus.

Homozygous: diploid cell or individual having 2 identical alleles (AA or aa). It produces only one type of gametes.

Heterozygous: an individual who has two different alleles for the same gene (Aa).

Line pure : a group of individuals presenting similar genetic material (species, offspring, lineage, or variety). A population that is homozygous for nearly all genes.

Dominant Character: an allele that conditions a phenotype in the heterozygous state. A character is dominant if it manifests in the heterozygote.

Recessive Character: an allele that does not condition the phenotype in the heterozygous state. A character is recessive if it only manifests in the homozygote.

Codominant Character: a character whose different phenotypic versions are all detectable.

b. Terminology

- ✓ **Generations:**

– P = parental generation

– F1 = First filial generation, the offspring of generation P

– F2 = 2nd filial generation, the descendants of the F1 generation (same for F3 and so on)

- ✓ **Crossing:**

– Mating between a male and a female individual

c. Conventional writing

Phenotype: written in brackets [ ]

Genotype: written in parentheses ( )

Dominant Character: the first letter of the trait is written in uppercase (e.g., L G). It expresses itself in the phenotype when present in a single version.

Recessive Character: the first letter of the trait is written in lowercase (e.g., vg e). It expresses itself in the phenotype when present in two versions.

d. Gene transmission in diploid organisms (2n)

1) The transmission of a single characteristic (animals presenting a single difference): this is **monohybridism** eg: hair color in mice (wild mice: gray, mutant mice: white)

2) The transmission of two characteristics: this is **dihybridism** eg: in drosophila (length of wings, color of eyes, color of body etc.)

3) The transmission of three or more characteristics: this is **multihybridism**

Mendel's Laws

First Law of Mendel: Law of Uniformity of First Generation Hybrids

- "The first generation of hybrids is homogeneous": All hybrids of the F1 generation are similar to one another (same phenotype and genotype).

Second Law of Mendel: Law of Segregation of Alleles in the F2 Generation

- "The alleles of the same pair segregate during gamete formation": F2 individuals are different from each other. This difference is explained by the segregation of alleles during gamete formation, which are therefore pure: each gamete contains only one of the two alleles.

Third Law of Mendel: Law of Independent Assortment of Alleles

- The observed phenotypes show that the segregation was independent for the different allele pairs.

• In a typical experiment, Mendel paired two contrasting, pure-line varieties, a process called hybridization

• Pure line parents are the P generation

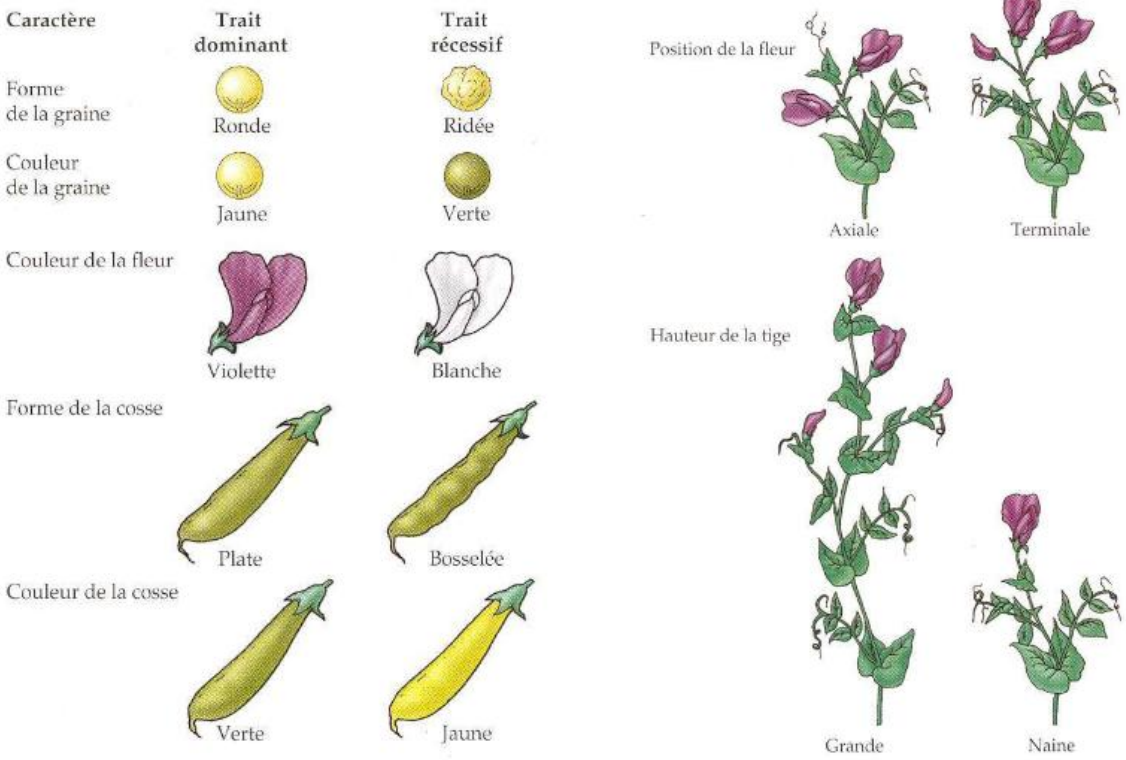
• The hybrid descendants of the P generation are called the F1 generation

• When F1 individuals self -pollinate , the F2 generation is produced.

In total, Mendel studied 7 pairs of lines corresponding to 7 pairs of phenotypes:

Character	**Phenotypes**	
Seed shape	round	wrinkled
Seed color	YELLOW	green
Color of flowers	purple	white
Pod shape	rounded	angular
Color of pods	green	YELLOW
Position of flowers	axial	terminal
Length of stems	long	short

Il présente des traits (caractères) de formes opposées distinctes.



Le matériel de Mendel : des pois de lignée pures

Pourquoi des pois ?

1) Le pois a des caractères faciles à observer

Couleur des fleurs, longueur de la tige, forme des graines …

2) Chaque caractère n'a que 2 formes

« 2 variations »

Fleurs blanches ou violettes, tiges longues ou courtes …

3) La fleur est fermée et donc à l'abri de la pollinisation extérieure

→ contrôle possible de la fécondation

Pourquoi des pois de lignée pure?

Afin d'évaluer le résultat des manipulations qu'il prévoyait faire sur ces lignées lors de leur reproduction.

Comment a-t-il obtenu ses pois de lignée pure?

Il a cultivé des pois durant plusieurs générations et a sélectionné les lignées dont les pois produisaient toujours des plants semblables à eux-mêmes.

Par exemple, des pois à fleurs violettes produisant toujours des pois à fleurs violettes.

2. Monohybridism

- Monohybridism consists of a cross between two parents that differ in only one trait (1 gene). For a given trait:
- If the hybrids exhibit the phenotype of one parent, the trait of that parent is **dominant**, and the trait of the other parent is **recessive**.
- If the hybrids exhibit an intermediate phenotype between the two parents, there is codominance.

- **Example 01: Case of dominance**

A gene responsible for coat coloration in an animal is defined by two alleles (a pair of alleles B/b), each characterized by a phenotype: B produces the dominant black color and b produces the recessive white color.

Allele B dominant : allele who drives to the phenotype **[black]** dominant **(BB or Bb)**

Allele b recessive : allele Who conduit has her phenotype **[white]** uniquely if on East below homozygous form **(bb).**

Crossing between line pure BB x bb

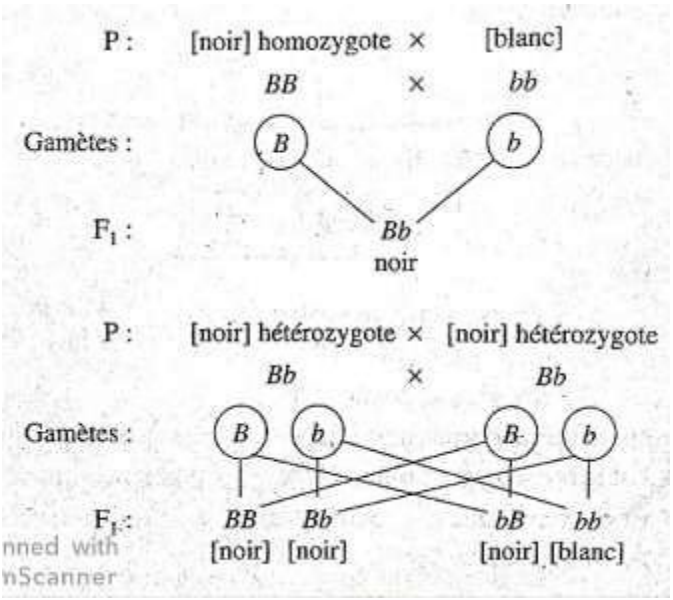
- **Generation F1 :** 100% Bb / 100% phenotype dominant [black]

Generation F2 : ¼ BB, ½ Bb , ¼ bb / ¾ phenotype dominant [black] and ¼ phenotype recessive [white].

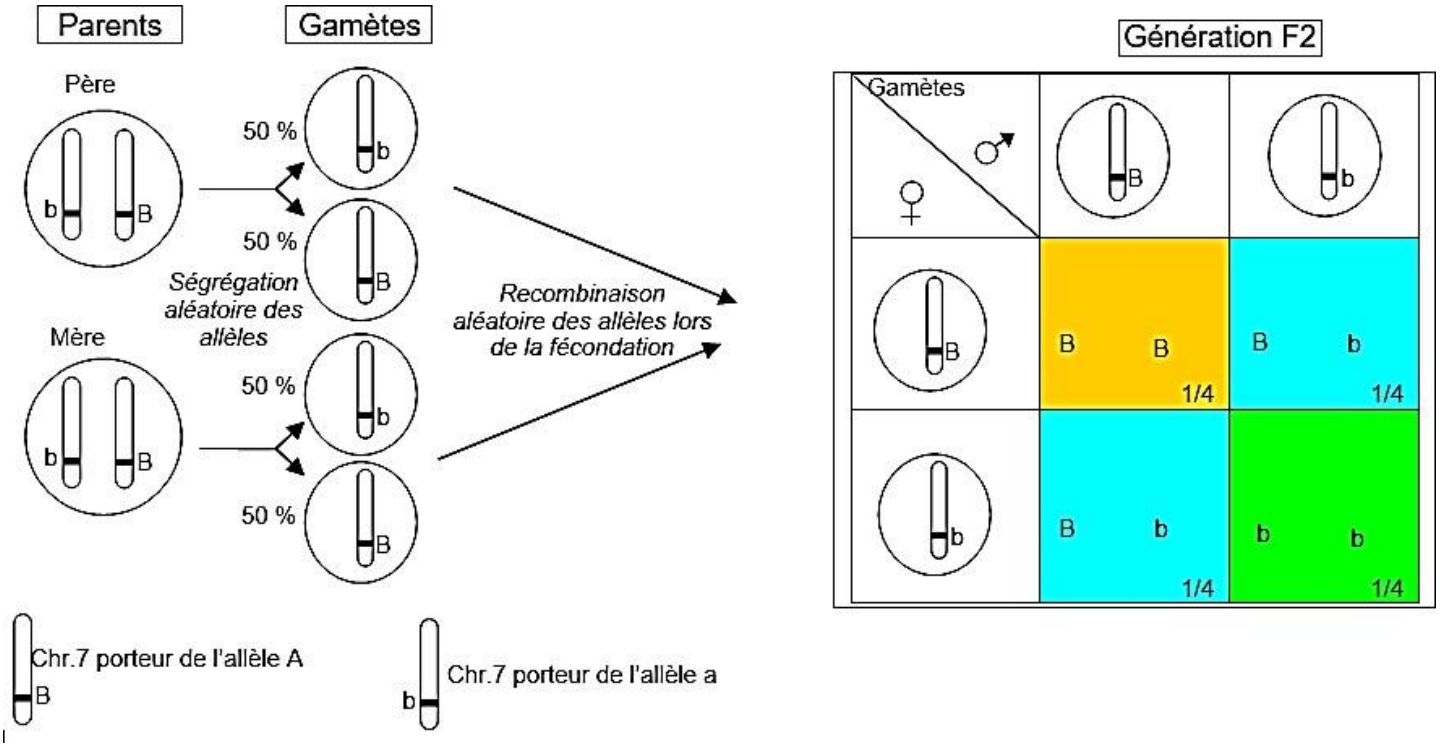
The chessboard of Punnett : method used A painting has double entrance For to show THE genotypes of all possible gametes from each parent.

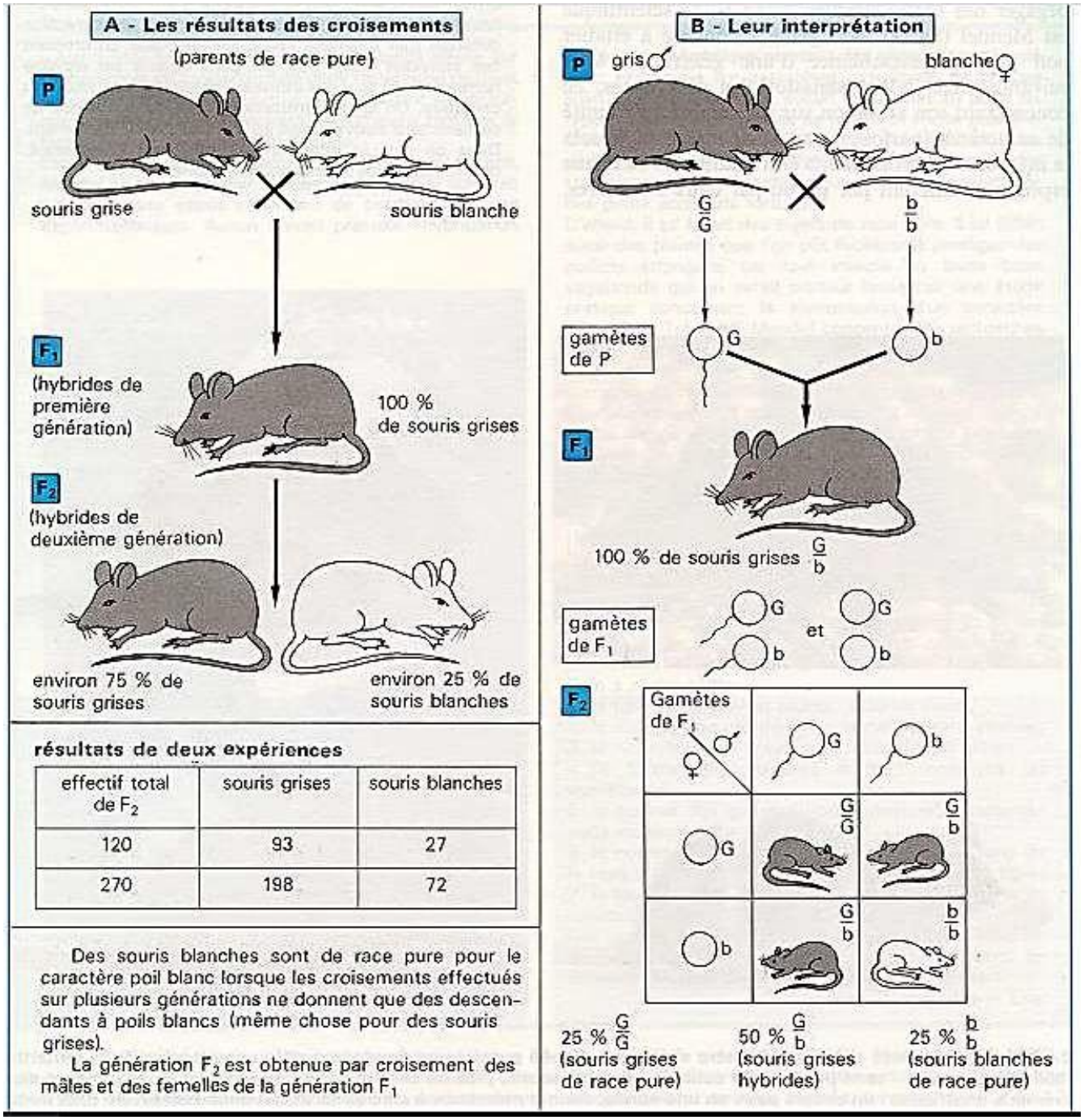
Gamètes	**B**	**b**
B	BB	Bb
b	Bb	bb

Génotype	**Phénotype**	**Fréquence**
BB (homozygote dominant)	[noir]	1/4
Bb (hétérozygote)	[noir]	1/2
bb (homozygote récessif)	[blanche]	1/4



Explanation of F2 :





résultats de deux expériences

effectif total de F_2	souris grises	souris blanches
120	93	27
270	198	72

Des souris blanches sont de race pure pour le caractère poil blanc lorsque les croisements effectués sur plusieurs générations ne donnent que des descendants à poils blancs (même chose pour des souris grises).

La génération F_2 est obtenue par croisement des mâles et des femelles de la génération F_1

Figure: Example of a case of dominance in monohybridism
(http://www.jpboseret.eu/biologie)

- **Example 02 : Case of codominance**

A gene responsible of coat coloration of coat of a animal East defined by 2 alleles (A couple A/B allele), each characterized by a phenotype: A produces the red color and B the white color, and the assembly of the two AB alleles produces an intermediate pink phenotype.

Crossing AA x BB

- **Generation F1 :** 100% AB / 100% phenotype intermediate [pink]

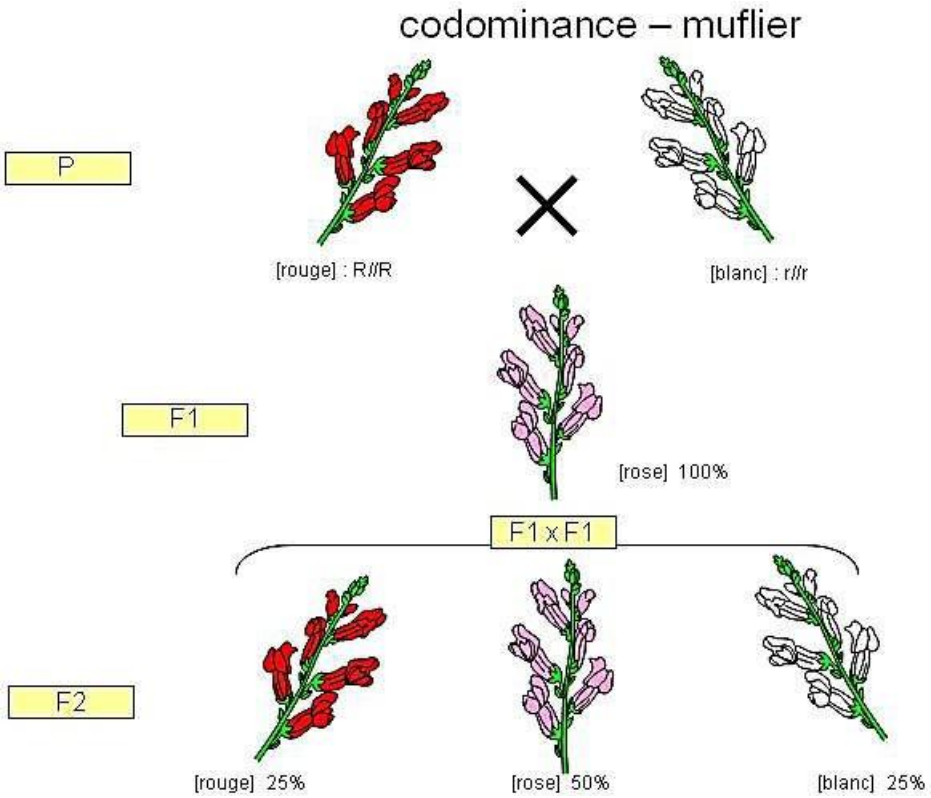
Self-fertilization AB x AB

- **Generation F2 :** ¼ AA, ½ AB, ¼ BB

¼ phenotype [red] and ½ phenotype intermediate [pink] and ¼ phenotype [white]

Gamètes	**A**	**B**
A	AA	AB
B	AB	BB

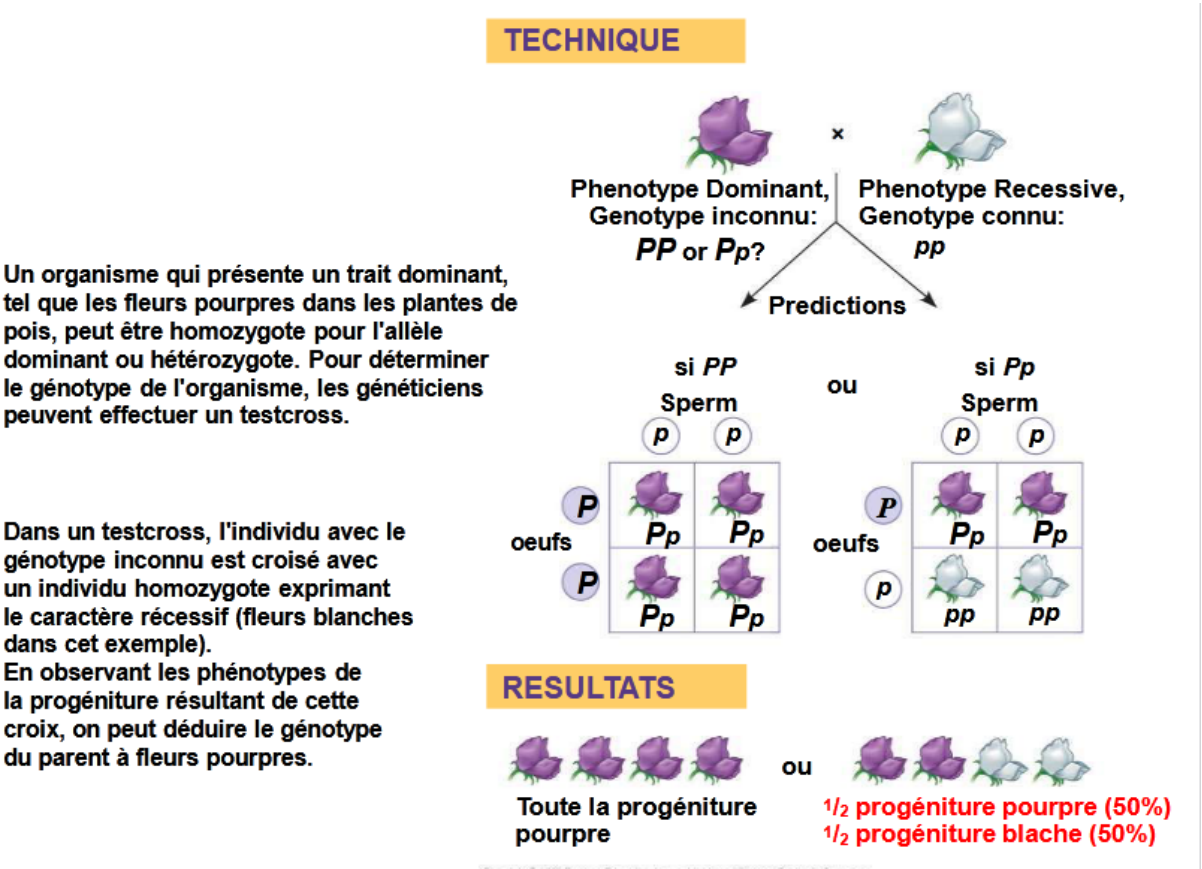
Génotype	**Phénotype**	**Fréquence**
AA	[red]	1/4
AB	[pink]	1/2
BB	[white]	1/4



Test-Cross

The **test-cross** is a cross between an individual with an unknown genotype and a homozygous recessive tester individual.

- The tester individual only produces one type of gamete and does not influence the phenotypes that appear in the offspring of the cross.
- The phenotypes and their proportions depend only on the gametes produced by the individual with the unknown genotype.
- The goal is to determine the genotype of the individual with the dominant phenotype, which

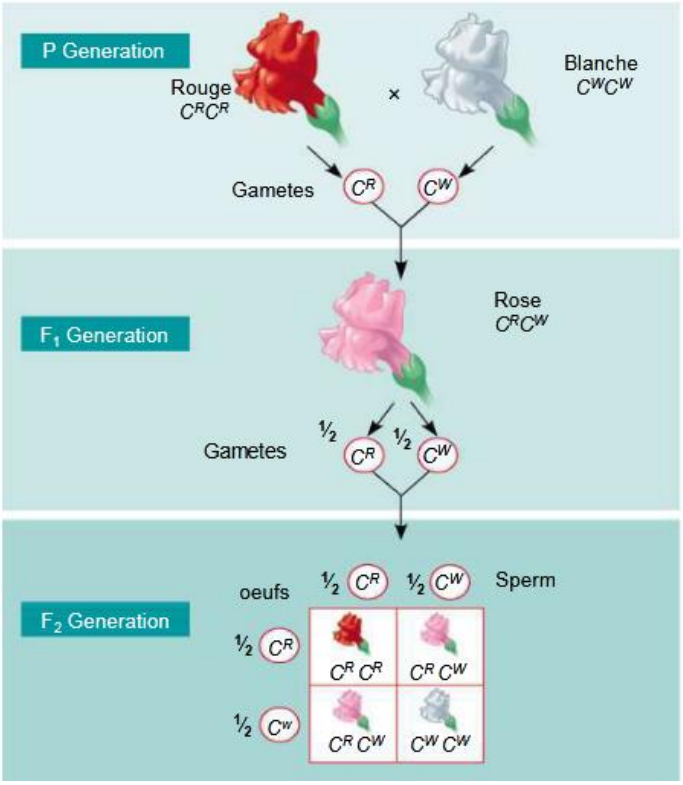


- **Exceptions to Mendel's Laws**

• Incomplete dominance • Sex-linked transmission
• Codominance • Linked genes
• Multiple alleles • Epistasis
• polygenic traits
• Environmental effects on gene expression
• Penetration
• Mosaicism germinal
• Pleiotropy

Dominance Incomplete

- Aucun des deux allèles n'est dominant et les individus hétérozygotes ont un phénotype intermédiaire
- Par example, au Japan les plantes “Four o’clock”, avec un allèle rouge et un allèle blanc ont des fleurs roses:



Codominance

Example of system blood human ABO :

[A] : A/A = Pour **ce** gène les deux allèles présents sont identiques, l'individu est **homozygote pour ce gène.**
OU A/O = Pour **ce** gène les deux allèles sont différents, l'individu est **hétérozygote pour ce gène.**

individu **homozygote** pour le gène considéré	2 allèles **identiques**
individu **hétérozygote** pour le gène considéré	2 allèles **différents**

A est **dominant** sur O. **O est récessif.**
[B] : B/B (homozygote) OU B/O (hétérozygote) : B est dominant sur O. O est récessif.
[O] : O/O (homozygote) seule possibilité car O est récessif.
[AB] : A/B (hétérozygote), les deux allèles s'expriment. A et B sont codominants.

Relation de dominance/récessivité	L'allèle A **est dominant** sur l'allèle O = l'allèle O est **récessif par rapport** à l'allèle A.	Génotype : (A/O) (A/A)	Phénotype : [A]
	L'allèle B est **dominant** sur l'allèle O = l'allèle O est **récessif par rapport** à l'allèle B.	Génotype : (B/O) (B/B)	Phénotype : [B]
Relation de codominance	Les allèles A et B sont **codominants**.	Génotype : (A/B)	Phénotype : [AB]

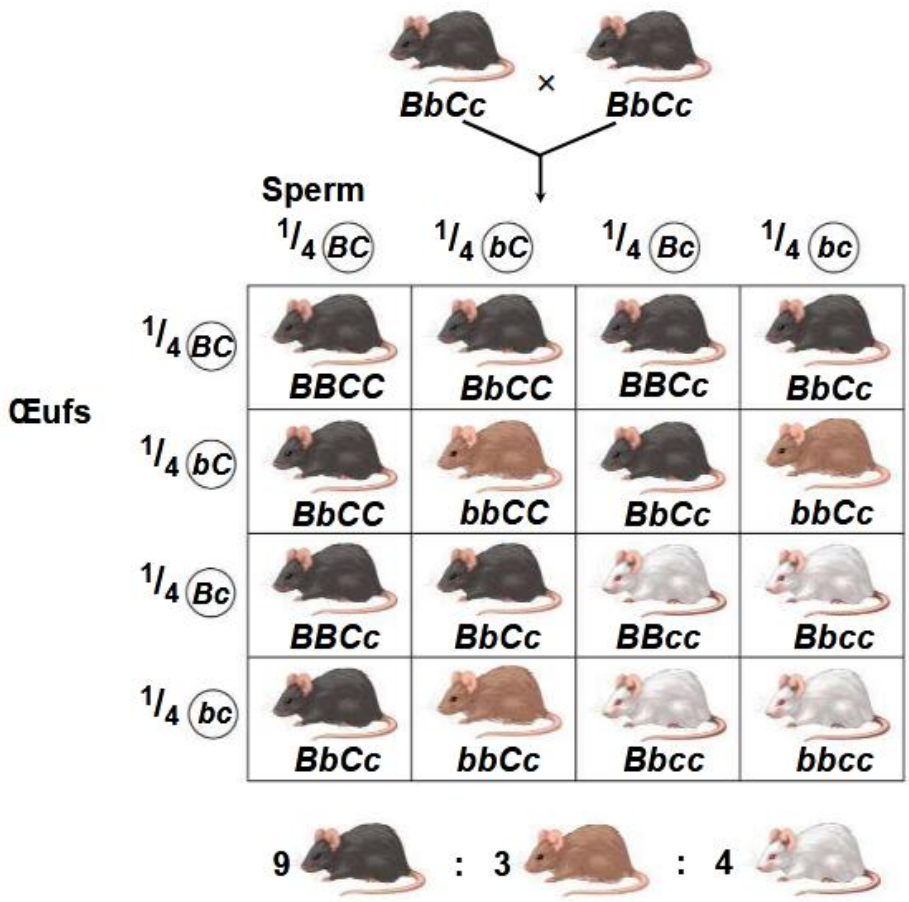
Epistasis

• In epistasis, a gene at one locus can alter the phenotypic expression of another different gene at a second locus

• For example, in mice and many other mammals, coat color depends on two genes

• A gene determines the color of the pigment (with alleles B for black and b for brown)

• The other gene (with alleles C for color and c for no color) determines whether the pigment will be deposited in the hair.



Pleiotropy

• This is when a single gene affects more than one trait.

• For example, in Labradors the gene locus that controls how black pigment is deposited in the hair can also affect the color of the nose, lips and eyes.

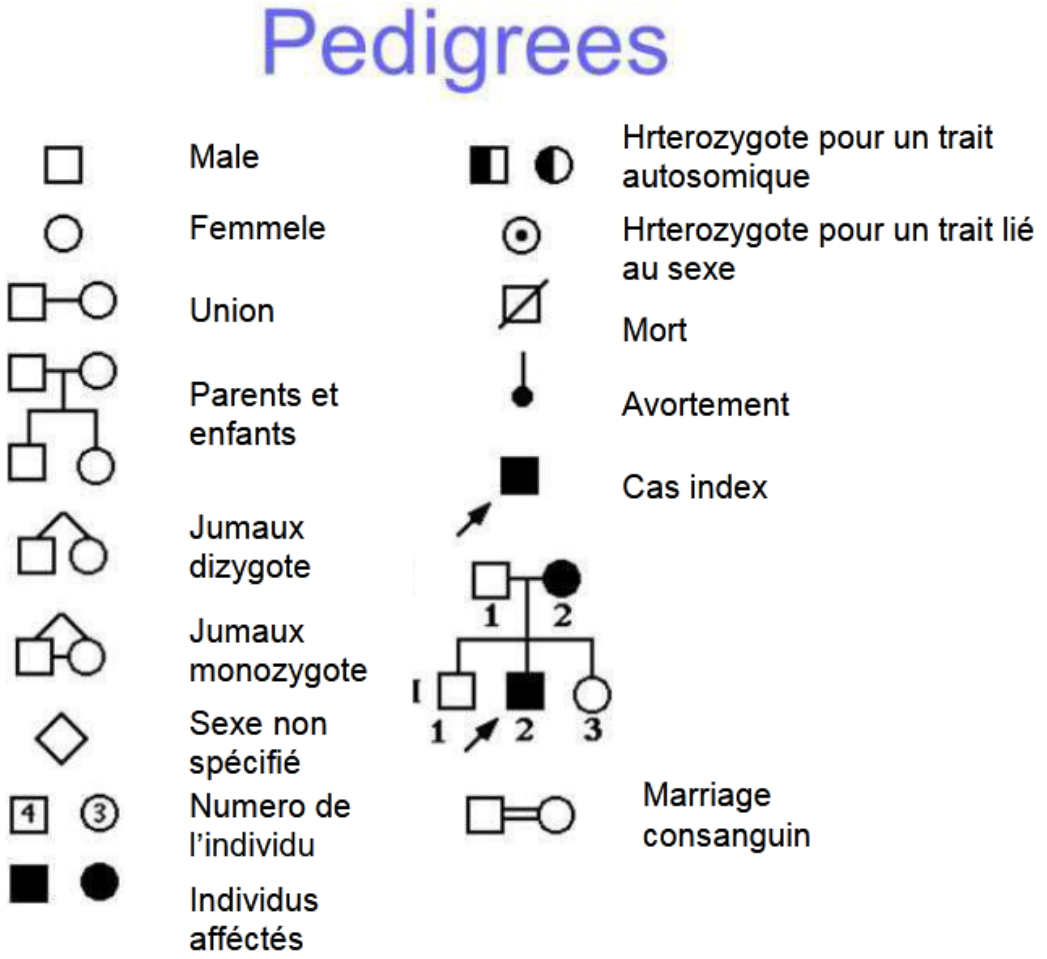
Pedigree Analysis

• A family tree (pedigrees) is a tree that describes the relationships of parents and children across generations.

• The inheritance of particular traits can be traced and described using pedigrees

• Pedigrees can also be used to make predictions about future offspring

• We can use the rules of multiplication and addition to predict the probability of specific phenotypes.

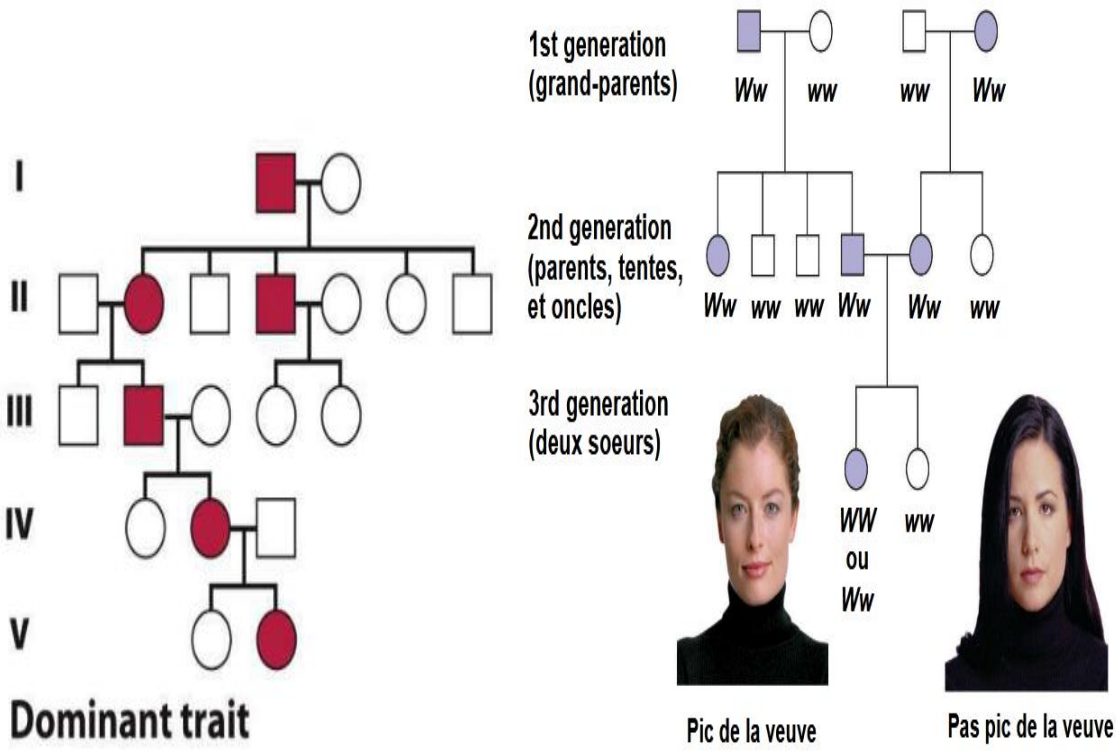


Transmission of a dominant trait

• Every individual who carries the dominant allele manifests the trait.

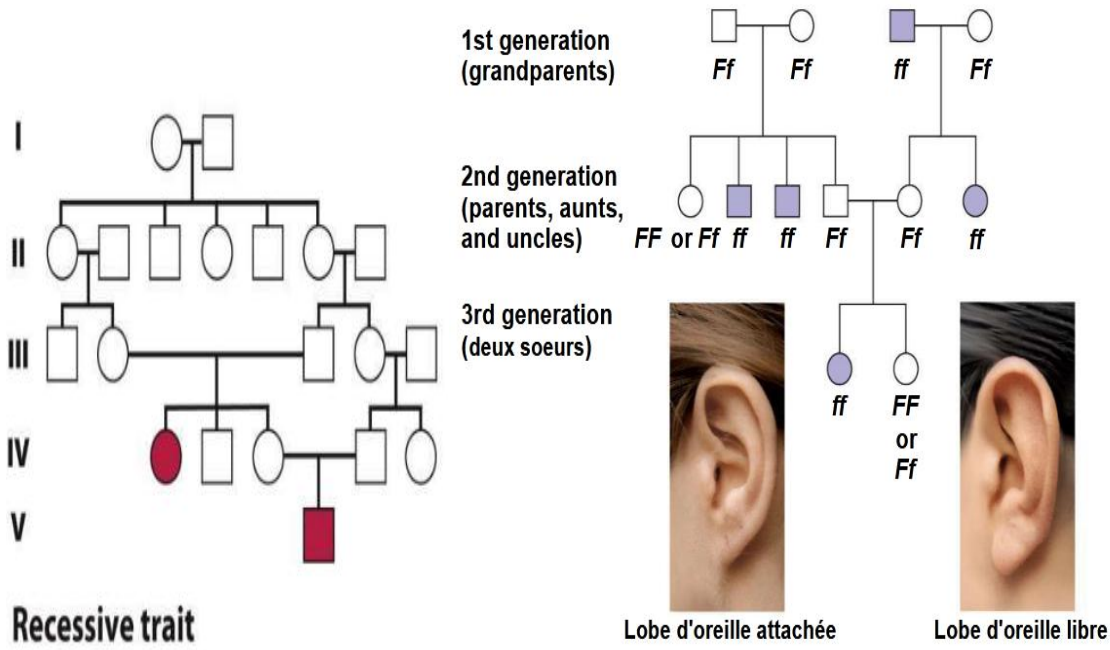
• Each affected person must have at least one affected parent.

• Most individuals showing the trait are heterozygous, and they have a ½ chance of passing the trait on to their children.



Transmission of a recessive trait

- Recessive traits may appear in individuals whose parents are not affected.
- Parents are heterozygous for the recessive allele and are called carriers
- About 1/4 of carriers' children are affected
- Affected individuals are homozygous for the trait.



Autosomal recessive diseases

• Many genetic diseases are inherited recessively

• Recessive hereditary diseases only manifest in individuals who are homozygous for the disease allele.

• Asymptomatic carriers (or healthy transmitters or drivers) are heterozygous individuals who carry the recessive allele (in the heterozygous state) but who are phenotypically normal.

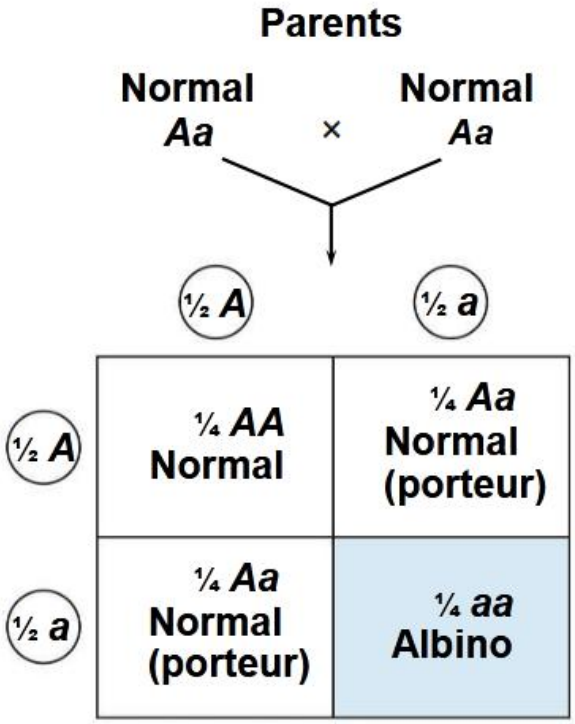
• If two parents are asymptomatic carriers, they have a one in four (1/4) chance of having a sick child.

• If a recessive allele that causes a disease is rare, then the probability of two carriers mating is low

• Consanguinity (i.e., marriage between close relatives) increases the risk of mating between two carriers of the same rare allele

• Most societies and cultures have laws or taboos against marriages between close relatives.

L'albinisme est une affection récessive caractérisée par un manque de pigmentation de la peau et des cheveux



dominant diseases

• Some human genetic diseases are caused by dominant alleles

• Dominant alleles that cause fatal disease are rare and often occur as a result of mutations

• Dominantly inherited diseases occur in individuals homozygous for the morbid allele (severe form of the disease) and in individuals heterozygous for the morbid allele

• If one of the two parents is heterozygous for the morbid allele, there is a one in two (1/2) chance of having a sick child.

L'achondroplasie est une forme de nanisme causée par un allèle dominant rare