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 chromosome of the species.

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

Genotype : the complete set of alleles of an individual. The alleles of a living organism's genes are normally shared among all members of the species.

Genome : in the broad sense, the total genetic information of an individual; more specifically, the complete set of genes or coded sequences of genetic material.

An allele: a variant of a DNA sequence located at a specific locus on a chromosome; one of the alternative forms of the same gene. In diploid cells, there are two alleles for each gene (one inherited from each parent, which may be identical or different). In a population, several alleles of a gene may coexist.

Locus: the physical location of a DNA sequence (coding or non-coding) on a chromosome.

Loci: plural of locus.

Homozygous: a diploid cell or individual possessing two identical alleles (AA or aa). It produces only one type of gamete.

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

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

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

Recessive Character: an allele that does not determine the phenotype in the heterozygous state. A trait is recessive if it manifests only in the homozygous state.

Codominant Character: a trait in which different phenotypic versions are all detectable in the heterozygote.

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 written in uppercase (e.g., L, G). It expresses itself in the phenotype when present in a single copy.

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

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**

First Law of Mendel: Law of Uniformity of the First Filial Generation (F1)

- "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 two alleles of a gene pair segregate during gamete formation": individuals of the F2 generation differ from one another. This variation is explained by the segregation of alleles during gametogenesis, so that each gamete carries only one of the two parental alleles.

Third Law of Mendel: Law of Independent Assortment of Alleles

- The Observed phenotypes demonstrate that the segregation of alleles is independent for different gene

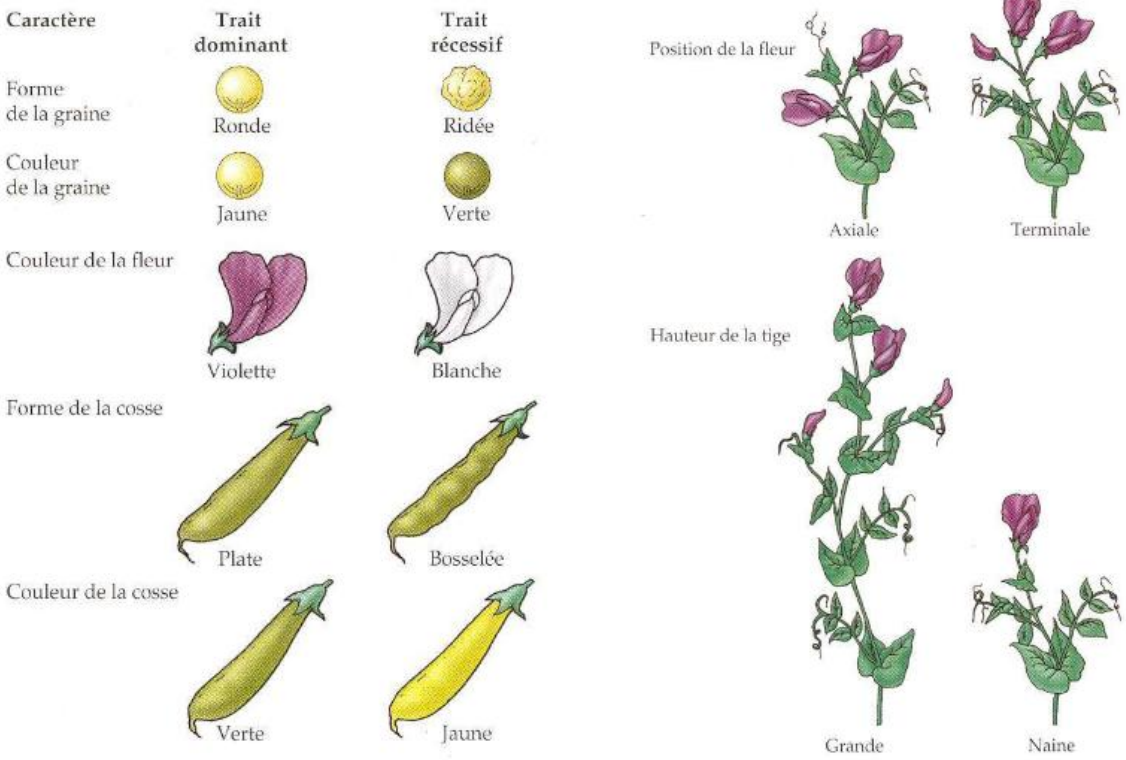
Mendel's Laws

- In a typical experiment, Mendel crossed two contrasting pure-line varieties, a process known as hybridization.
- The pure-line parents constitute the P generation.
- The hybrid offspring of the P generation are called the F1 generation.
- When F1 individuals self-pollinate, they produce the F2 generation.

In total, Mendel studied seven pairs of traits, each corresponding to a pair of contrasting 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 refers to a cross between two parents that differ in only one trait (controlled by a single gene).
- If the hybrids display the phenotype of one parent, that parental trait is considered **dominant**, while the other is **recessive**.
- If the hybrids display an intermediate phenotype between the two parents, the inheritance shows **codominance** (or incomplete dominance, depending on the case).

- **Example 01: Case of dominance**

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

- **Dominant allele (B):** an allele that results in the **[black]** phenotype when present in either homozygous (BB) or heterozygous (Bb) condition.
- **Recessive allele (b):** an allele that results in the **[white]** phenotype only when present in the homozygous condition (bb).

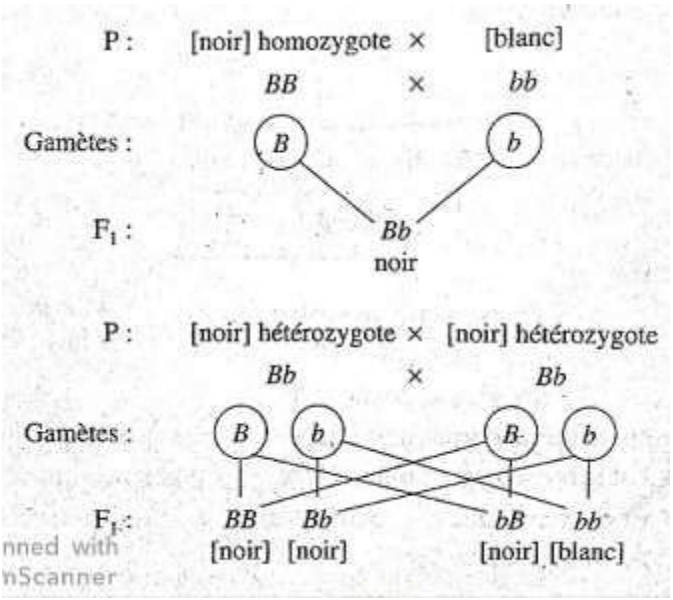
Crossing between line pure BB x bb

- **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].

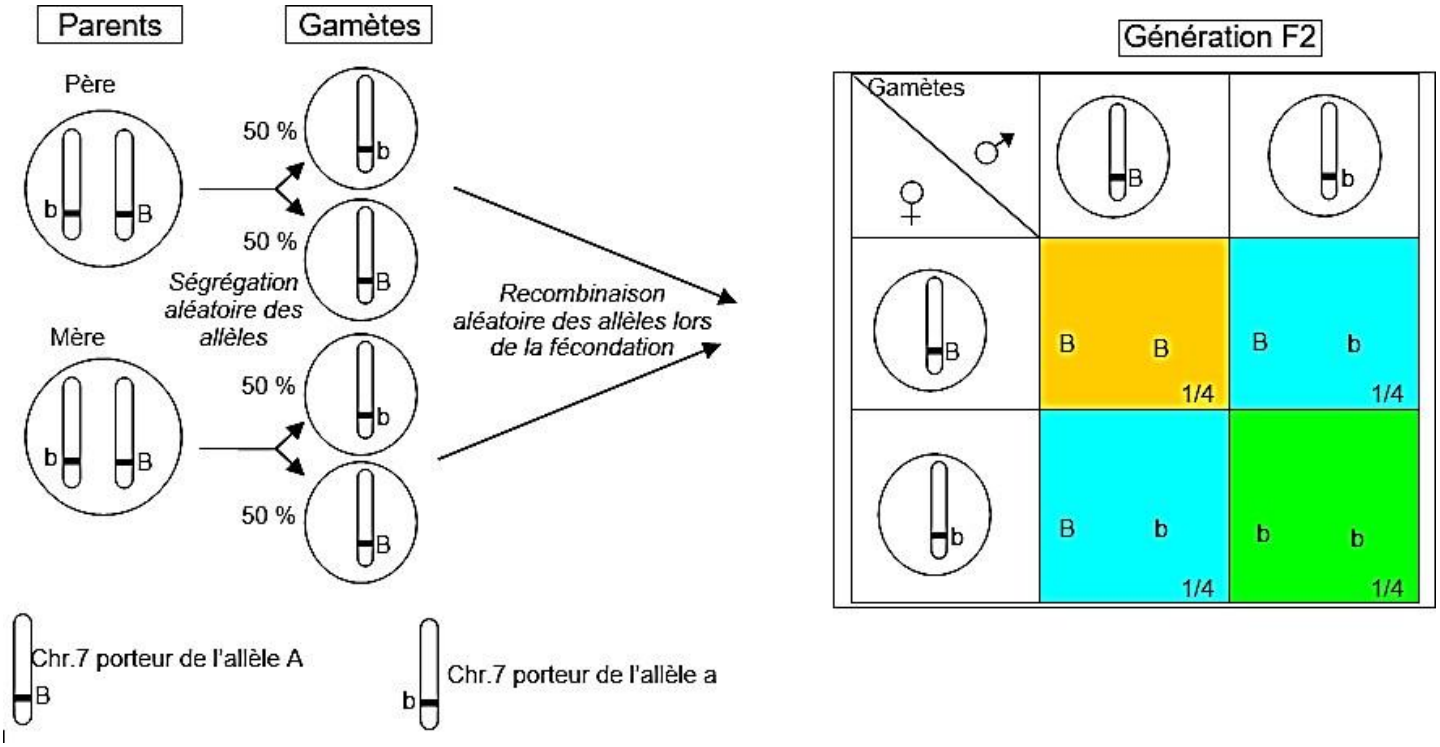
Punnett square: a method that uses a two-dimensional grid to represent the possible combinations of gametes from each parent, showing all potential genotypes of the offspring.

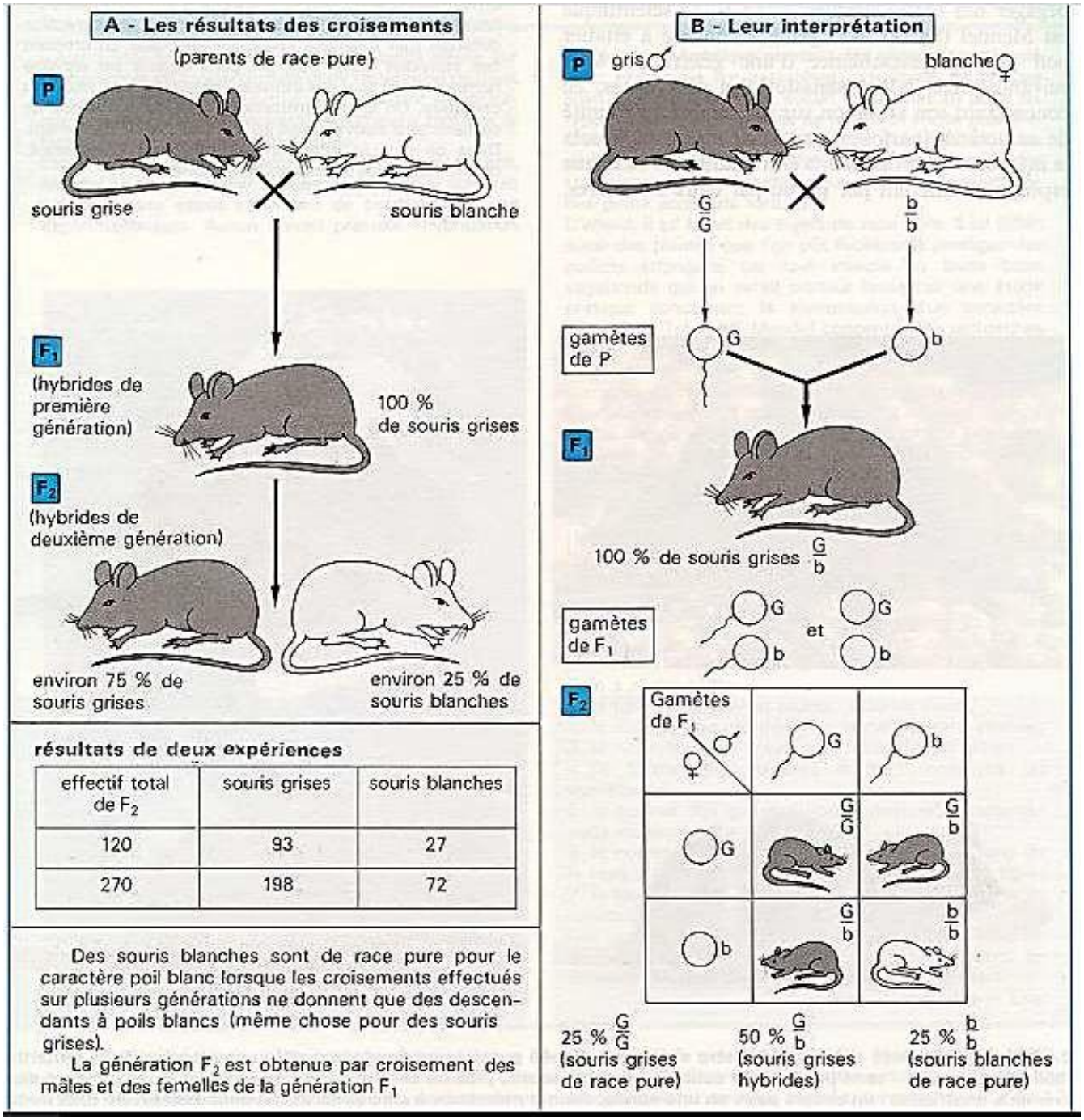
Gamètes	**B**	**b**
B	BB	Bb
b	Bb	bb

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 for coat color in an animal is defined by two alleles (A and B), each associated with a distinct phenotype: allele **A** produces a red coat, allele **B** produces a white coat, and the combination of both alleles (**AB**) results in an intermediate pink phenotype.

Crossing AA x BB

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

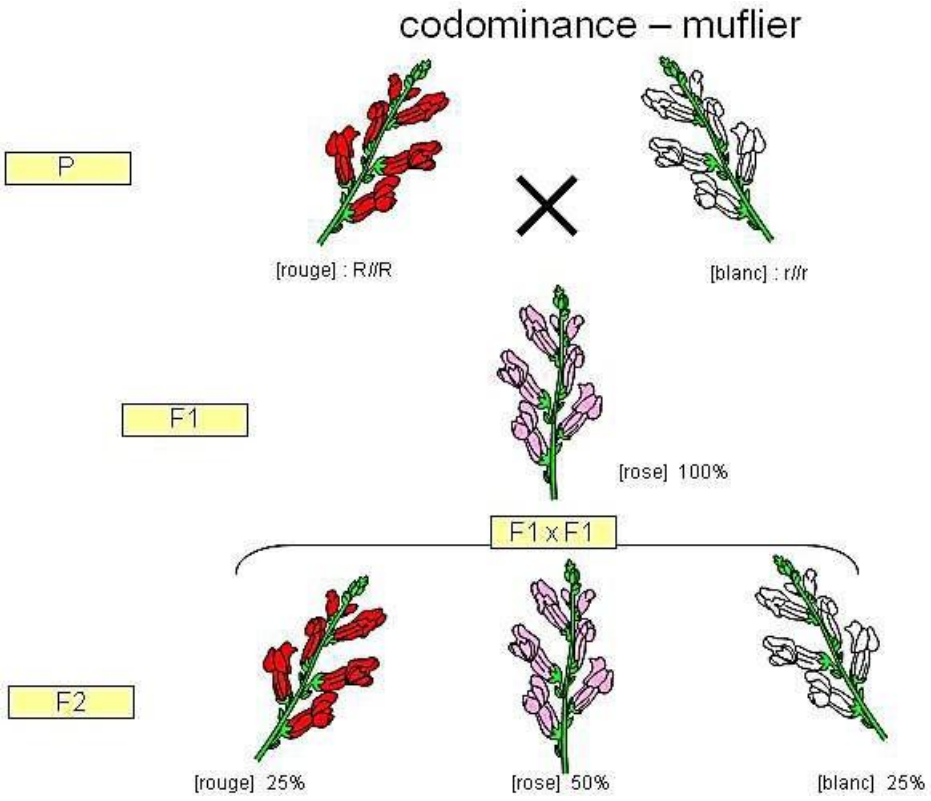
Self-fertilization AB x AB

- **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

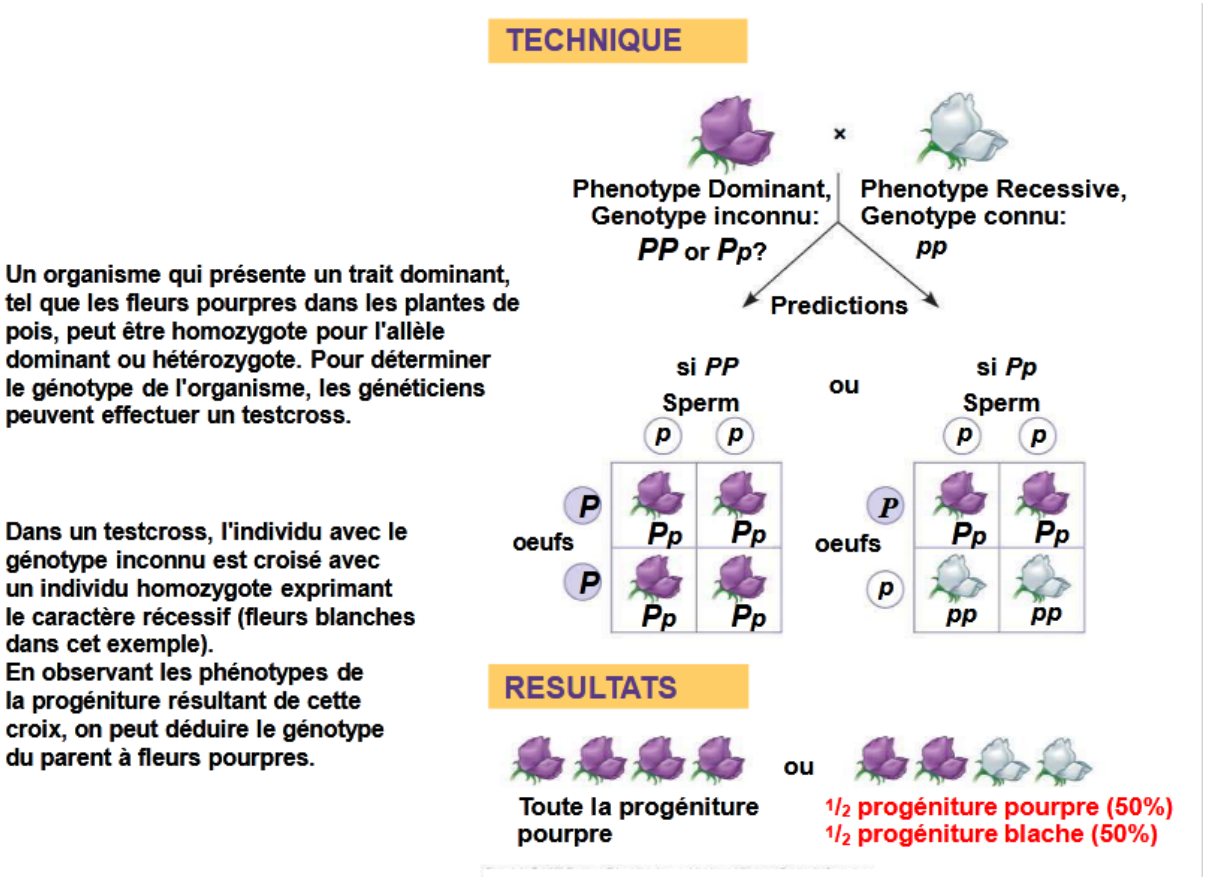
Génotype	**Phénotype**	**Fréquence**
AA	[red]	1/4
AB	[pink]	1/2
BB	[white]	1/4



Test-Cross

Test cross: a cross between an individual with an unknown genotype and a homozygous recessive tester individual.

- The tester individual produces only one type of gamete and therefore does not influence the phenotypes observed in the offspring.
- The phenotypes and their proportions depend solely on the gametes produced by the individual with the unknown genotype.
- The objective is to determine the genotype of the individual with the dominant phenotype, which may be either homozygous dominant or heterozygous



Un organisme qui présente un trait dominant, tel que les fleurs pourpres dans les plantes de pois, peut être homozygote pour l'allèle dominant ou hétérozygote. Pour déterminer le génotype de l'organisme, les généticiens peuvent effectuer un testcross.

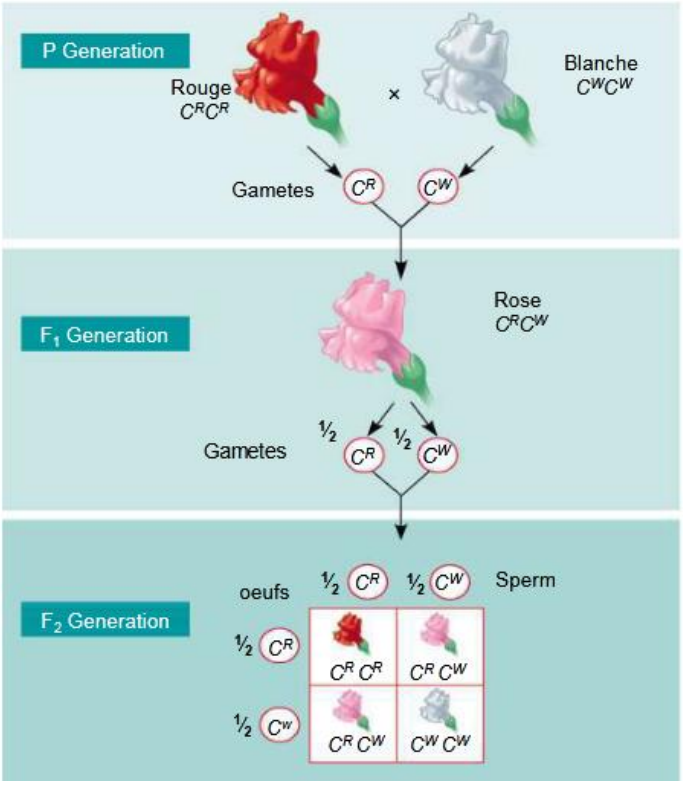
Dans un testcross, l'individu avec le génotype inconnu est croisé avec un individu homozygote exprimant le caractère récessif (fleurs blanches dans cet exemple).
En observant les phénotypes de la progéniture résultant de cette croix, on peut déduire le génotype du parent à fleurs pourpres.

- **Exceptions to Mendel's Laws**

• Incomplete dominance
• Codominance
• Multiple alleles
• polygenic traits
• Environmental effects on gene expression
• Penetration
• Mosaicism germinal
• Sex-linked transmission
• Linked genes
• Epistasis
• 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 or mask the phenotypic expression of a gene at another locus.
- For example, in mice and many other mammals, coat color is determined by the interaction of two genes.
- One gene determines pigment color, with alleles ***B*** for black and ***b*** for brown.
- The other gene, with alleles ***C*** (color) and ***c*** (no color), determines whether the pigment is deposited in the hair.

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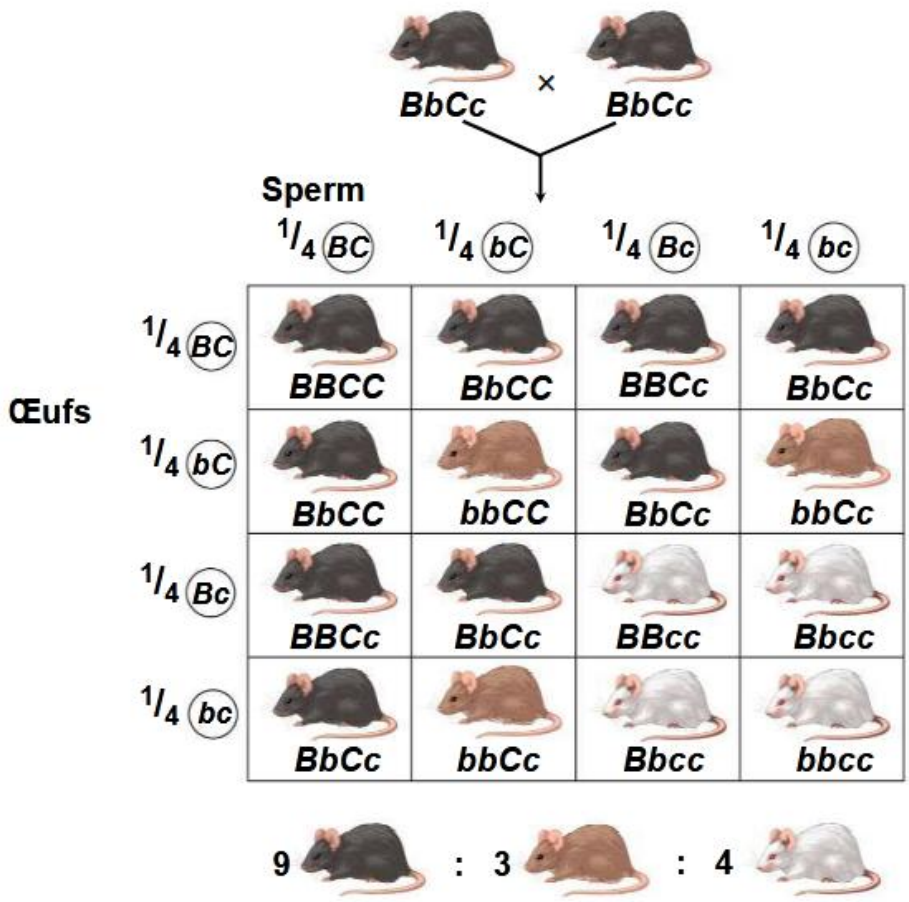

Pleiotropy

Pleiotropy occurs when a single gene influences more than one trait.

- For example, in Labradors, the gene locus that controls the deposition of black pigment in the hair also affects the color of the nose, lips, and eyes.

Pedigree Analysis

- A pedigree (family tree) is a diagram that represents the relationships between parents and children across generations.
- The inheritance of particular traits can be traced and analyzed using pedigrees.
- Pedigrees can also be used to predict the traits of future offspring.
- The rules of multiplication and addition can be applied to calculate the probability of specific phenotypes.

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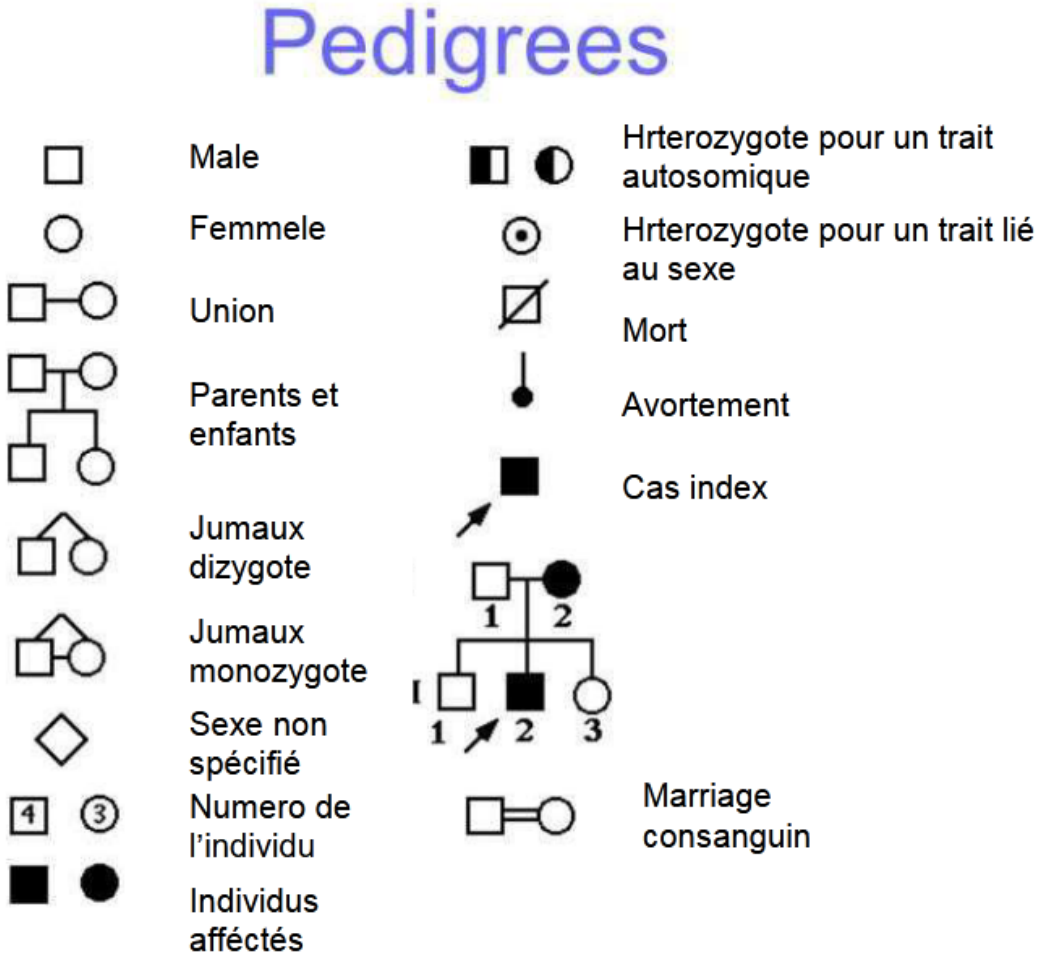
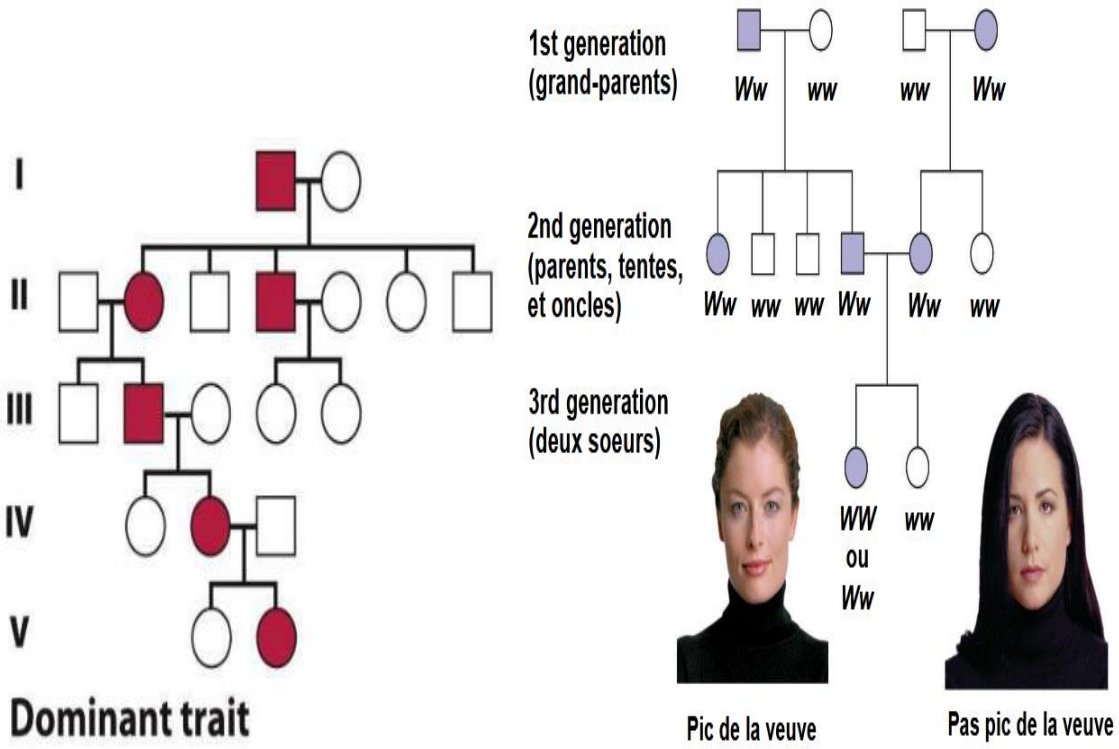


Transmission of a dominant trait

• 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.
- Such parents are heterozygous for the recessive allele and are referred to as ***carriers***.
- Approximately one-quarter of the children of two carriers are expected to be affected.
- Affected individuals are ***homozygous recessive*** for the allele.

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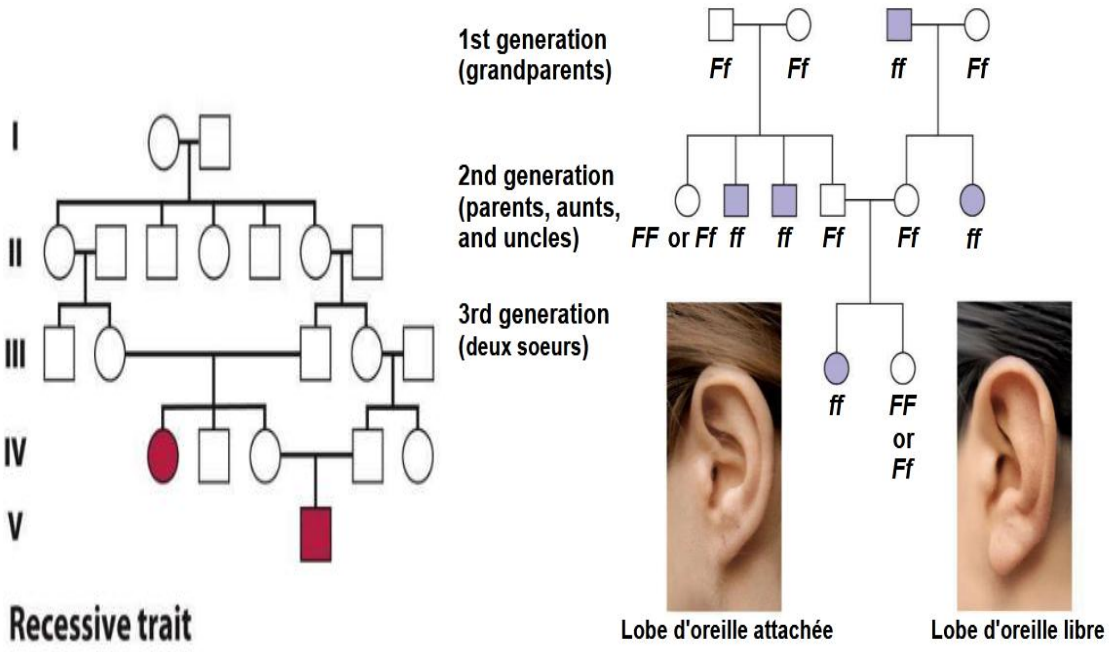

Autosomal recessive diseases

- Many genetic diseases are inherited in a recessive manner.
- Recessive hereditary diseases manifest only in individuals who are **homozygous** for the disease-causing allele.
- Asymptomatic carriers (also called healthy carriers) are heterozygous individuals who carry the recessive allele but remain phenotypically normal.
- When both parents are asymptomatic carriers, there is a one-in-four (1/4) probability that their child will be affected.
- If the disease-causing recessive allele is rare, the probability of two carriers mating is low.
- Consanguinity (marriage between close relatives) increases the likelihood that both parents carry the same rare allele.
- Most societies and cultures have laws or taboos that prohibit marriages between close relatives.

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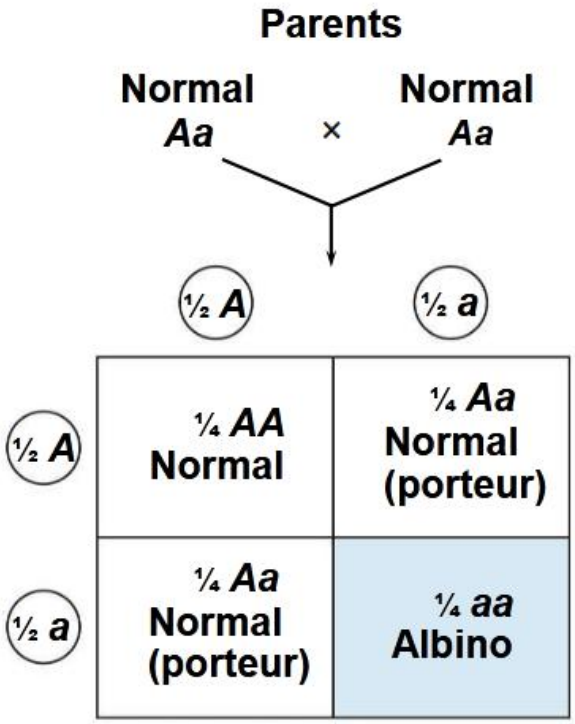
L'albinisme est une affection récessive caractérisée par un manque de pigmentation de la peau et des cheveux



- **dominant diseases**

- Some human genetic diseases are caused by ***dominant alleles.***

- Dominant alleles that cause lethal diseases are rare and often arise through new mutations.

- Dominantly inherited diseases occur in individuals who are ***heterozygous*** for the disease-causing allele, as well as in those who are ***homozygous*** (often presenting a more severe form of the disease).

- If one parent is heterozygous for the disease-causing allele, there is a one-in-two (1/2) probability that their child will be affected.

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