6. Sex chromosomes and sex-linked inheritance

Sex chromosomes are those that determine an individual's sex. In humans and many other organisms, they are distinguished from the non-sex chromosomes, called autosomes. Humans have 23 pairs of chromosomes:

- **Autosomes:** 22 pairs of chromosomes that are identical in both sexes.
- **Gonosomes (sex chromosomes):** one pair of sex chromosomes, the X chromosome and the Y chromosome.

➢ Differences between the X and Y chromosomes

- **X chromosome:** A relatively large chromosome containing about 1,100 genes. These genes are involved in many aspects of human biology, in addition to sex determination.
- **Y chromosome:** Much smaller than the X chromosome, containing about 50–200 genes. Its primary role is male sex determination, particularly through the **SRY gene** (Sex-determining Region Y), which initiates the development of male characteristics in the embryo.

Sex-linked inheritance

Sex-linked inheritance refers to the transmission of genes carried on the sex chromosomes, primarily the X chromosome in humans. Traits or diseases linked to these genes follow a specific inheritance pattern known as **X-linked inheritance** (often X-linked recessive inheritance).

➢ Inheritance of X-linked traits

- **Females (XX):** They can be **homozygous** (two normal alleles or two disease alleles) or **heterozygous** (one normal allele and one disease allele). Females often carry an X-linked recessive disease without expressing symptoms if the defective gene is recessive.

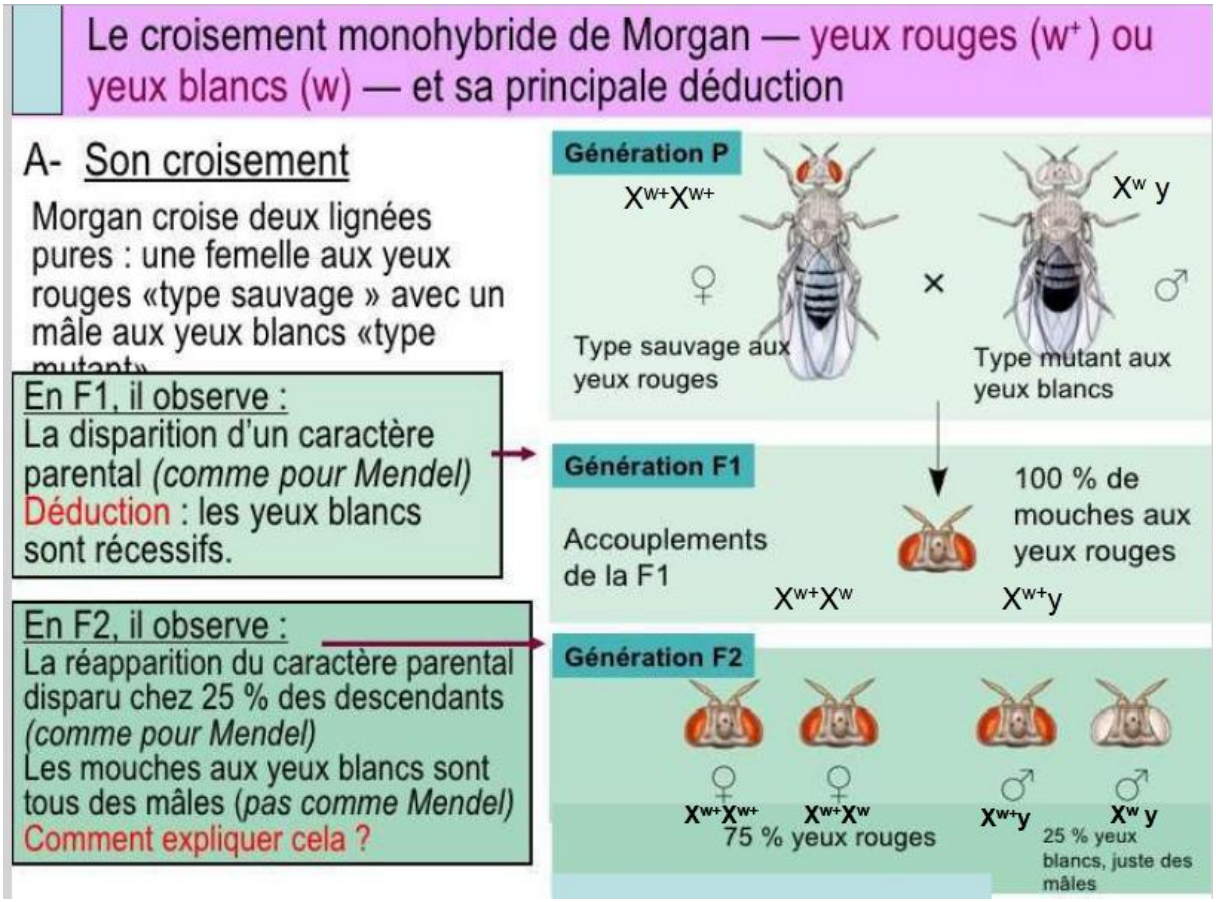
- **Males (XY):** They have only one X chromosome. Therefore, if this chromosome carries a defective gene, they will express the associated trait or disease, since they lack a second X chromosome to compensate.

Examples of sex-related diseases

1. **Hemophilia:** A disorder in which the blood does not clot properly, caused by a defective gene on the X chromosome.
2. **Color blindness:** An inability to distinguish certain colors (most commonly red and green), due to mutations on genes located on the X chromosome.
3. **Duchenne muscular dystrophy:** A severe muscle-wasting disease caused by mutations in the **dystrophin gene** on the X chromosome.

- **'s monohybrid cross :**

Crossing for a sex-linked trait Red (w+) or white (w) eye trait



Morgan's hypothesis to explain his results: The Mendelian trait(gene) for eye color is carried on the X chromosome and has no equivalent on the Y chromosome. (Sex-linked gene).

- **Heredity influenced by sex:**

• **Sex-influenced inheritance** occurs when a trait is controlled by autosomal genes that are expressed differently in males and females. The genes are present in both sexes, but their expression is influenced by stimulator or inhibitor factors linked to the sex chromosomes.

Example : In cattle, bulls carry genes that control milk production. They transmit these genes to their female offspring, but the trait is expressed only in females, while neither bulls nor their male offspring can express it.

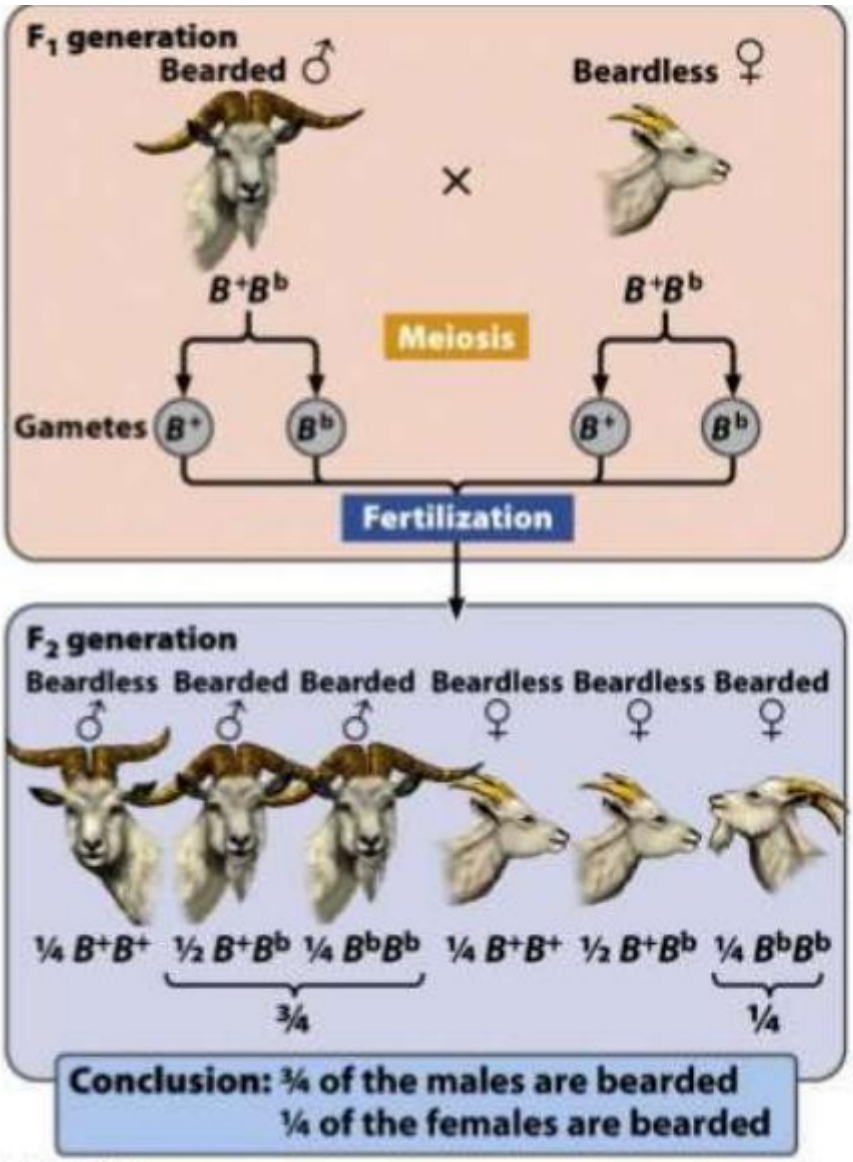


Figure 5-12b
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➢ **X-linked hereditary diseases due to a recessive allele**

- In this mode of inheritance, the disease-causing allele behaves like a recessive trait. Heterozygous females are usually unaffected but can transmit the allele; they are referred to as **carriers**.

- The disease most often manifests in males (XY), who have only one copy of the allele (hemizygous individuals).

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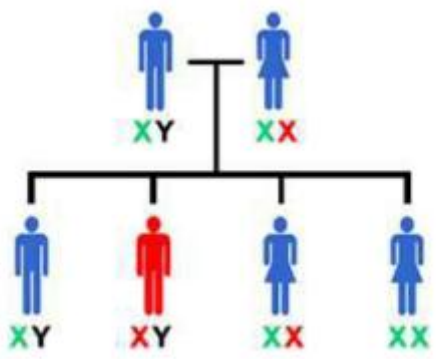

- Color Blindness: Color blindness is when someone has the inability to distinguish the differences between certain colors.

Vue normale

Vue par un daltonien

d = color blind D = normal Dd = carrier

XDXD = normal woman XDXd = surrogate woman

XdXd = affected woman XDY = normal man

XdY = affected man

Une mère dont la vision est normale mais conductrice et un père daltonien peuvent avoir :

Père	x	Mère
X^dY		X^DX^d

a) Un garçon daltonien

b) Une fille daltonienne

La dystrophie musculaire de Duchenne: