Cell Biology, Genetics and Evolution

SEX LINKAGE

IN DROSOPHILA AND HUMAN

By: Shozab Seemab Khan (PhD Scholar)

Sex Linkage in Drosophila

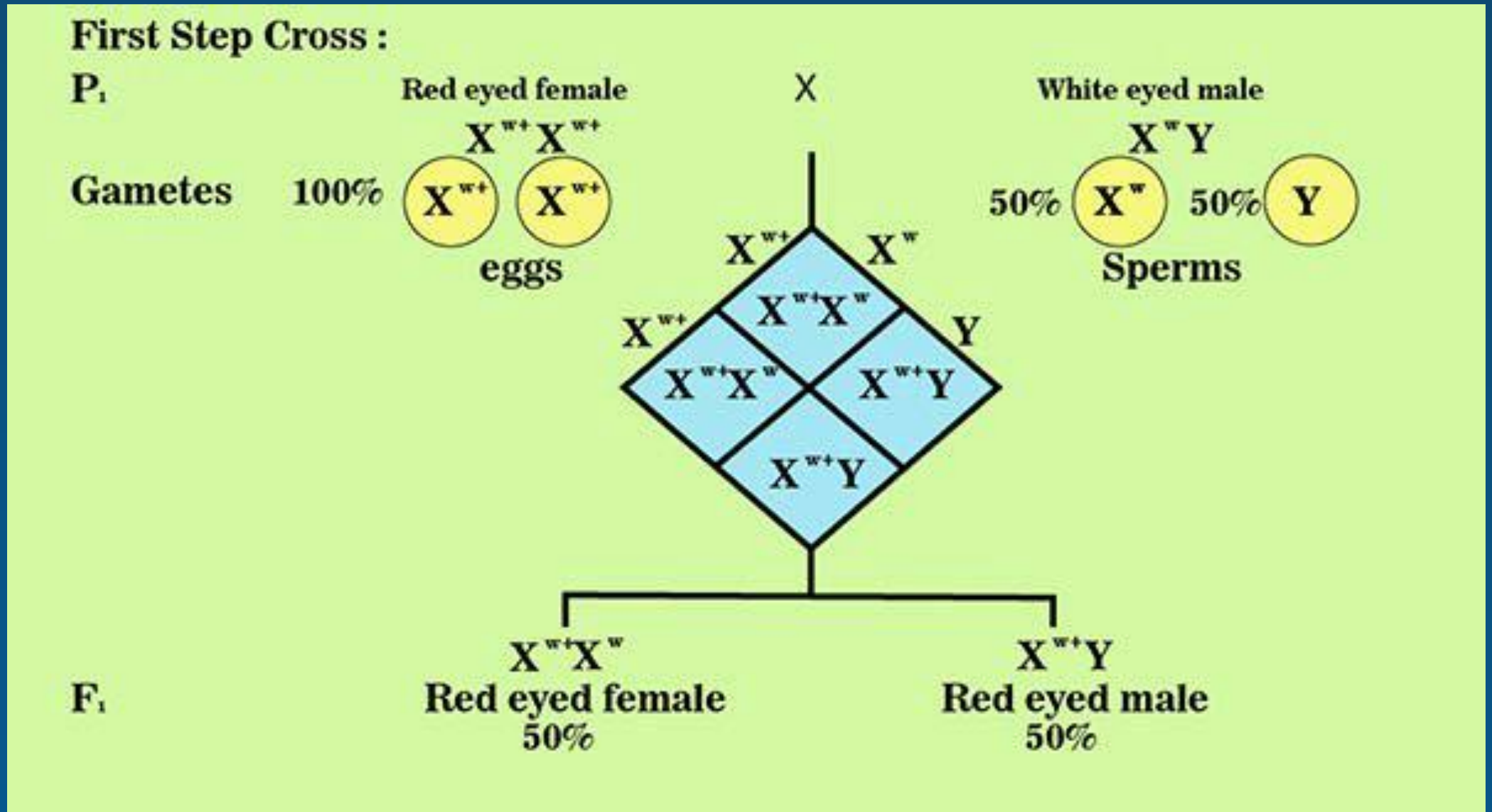
- Thomas Hunt Morgan (1910) provided experimental evidence in support of chromosomal theory of heredity through discovery of sex linkage in fruit fly (*Drosophila melanogaster*).
- Morgan raised cultures of Drosophila flies to study different traits, such as colour of the eye. Normal fruit flies, the wild type, have **bright red eyes**.
- One of his coworkers **Calvin Bridges**, observed an unusual **white eye mutant male fly**.

Red eyed female

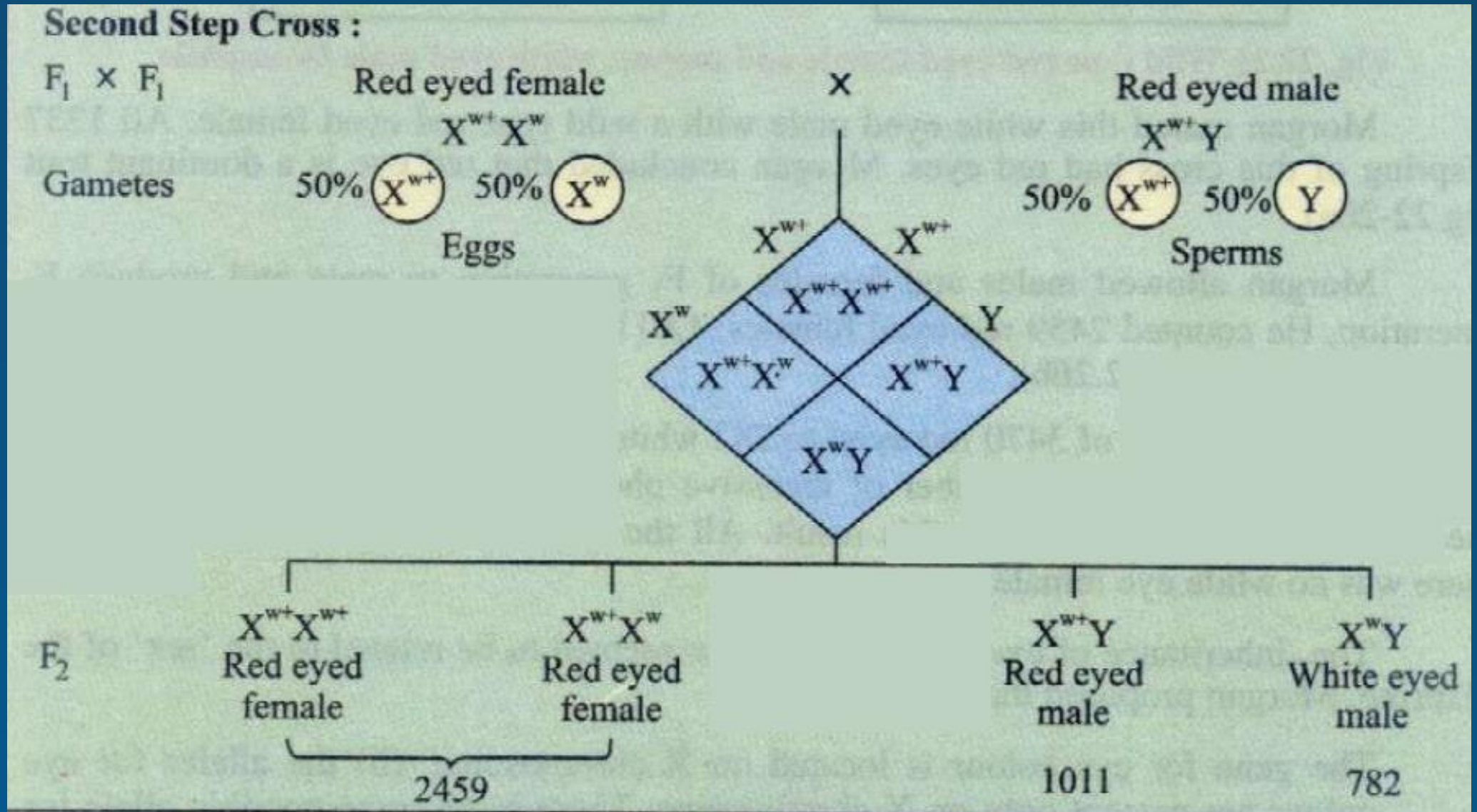
White eyed male

Wild type red eyed female and mutant white eyed male Dorsophilia

- Morgan mated this **white eyed male** with a **wild type red eyed female**. All 1237 offspring of this cross had **red eyes**. Morgan concluded that **red eye** is a dominant trait.



- Morgan allowed males and females of F1 generation to mate and produce F2 generation. He counted 2459 **red-eyed females**, 1,011 **red-eyed males** and 782 **white eye males** among F2.



Sex Linkage in Drosophila

- The proportion of 3470 red eyed to 782 **white eyed flies** did not perfectly fit into Mendelian 3 : 1 ratio.
- The number of recessive phenotype individuals was too small. There was another peculiarity in this result.
- All the **white-eyed flies** were only males.
- There was no **white eye female** in F2 generation.

Sex Linkage in Drosophila

- The inheritance of eye colour some how seemed to be related to the ‘sex’ of the offspring. Morgan proposed that:
- (i) The gene for eye colour is located on X chromosome,
- (ii) Alleles for eye colour are present only on X chromosome. There is no corresponding allele for this trait on Y chromosome.
- Thus even a single recessive allele on X chromosome can express itself in males because Y chromosome is empty for that gene. Males are hemizygous as they carry just one allele on their only X chromosome. Females have two X chromosomes, each carrying an allele of the trait. Females can be homozygous or heterozygous.

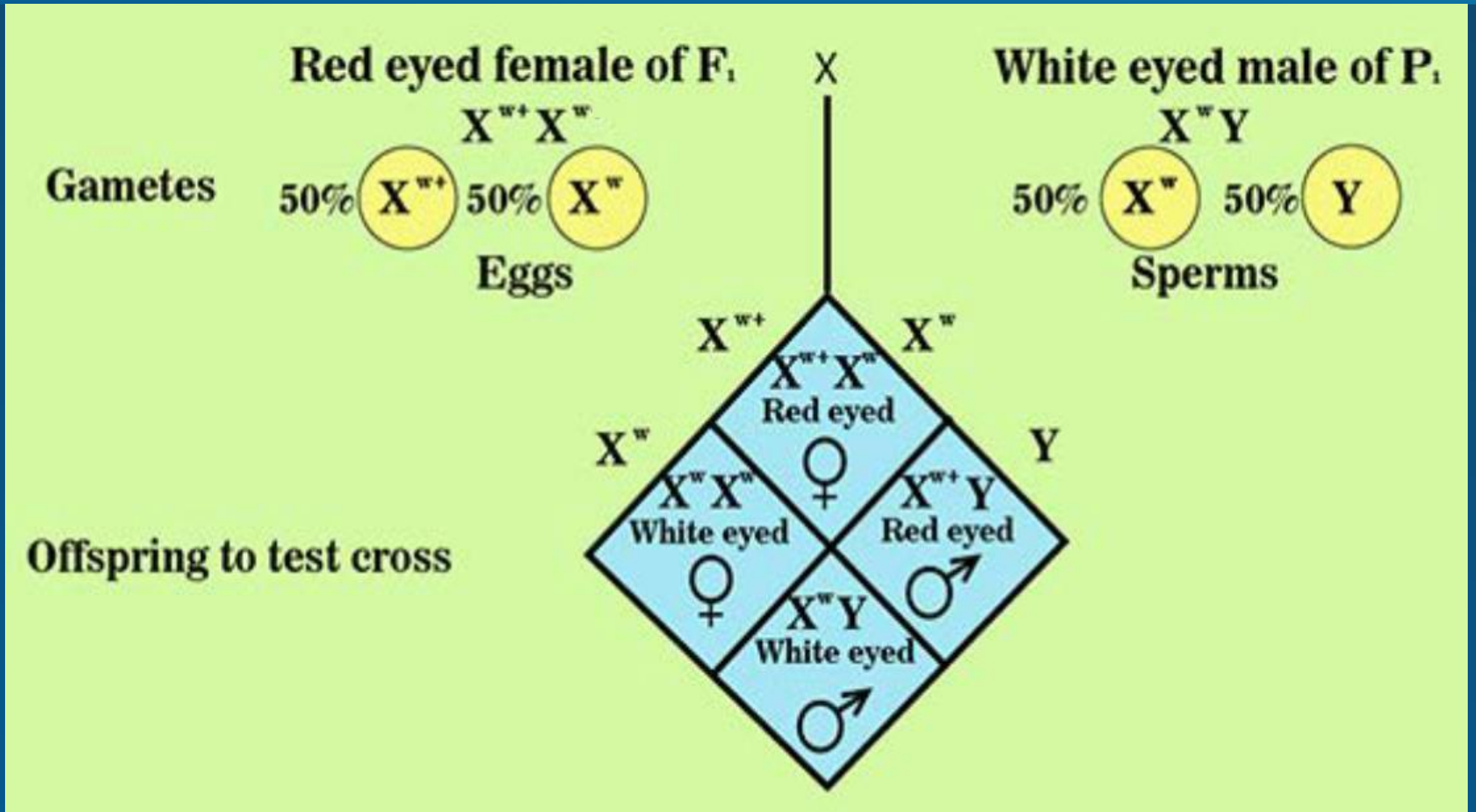
Sex Linkage in Drosophila

- Symbol “w” represents the recessive allele for **white eye**, and “w+” designates its wild type allele for **red eye**. The genotypes of the parents of P1 cross were: **$X^{w+}X^{w+}$ for red eye female**, and **$X^{w}Y$ for the white eye male**.
- Morgan’s hypothesis explained clearly why all the **white eyed flies** in F2 generation were only males.

Test Cross

- Morgan wanted to test his hypothesis. He crossed the P1 white eyed male (X^wY) with one of its own daughters, the red eyed heterozygous female ($X^{w+}X^w$) from F1 generation.
- This test cross produced 129 **red-eyed females**, 132 **red eyed males**, 88 **white-eyed females** and 86 **white eyed males**.
- **White-eyed flies** were less viable than **red-eyed flies**. Half the female offspring in fact had **red eyes** and half had **white**. Similarly half the males had **red eyes** and half had **white**.

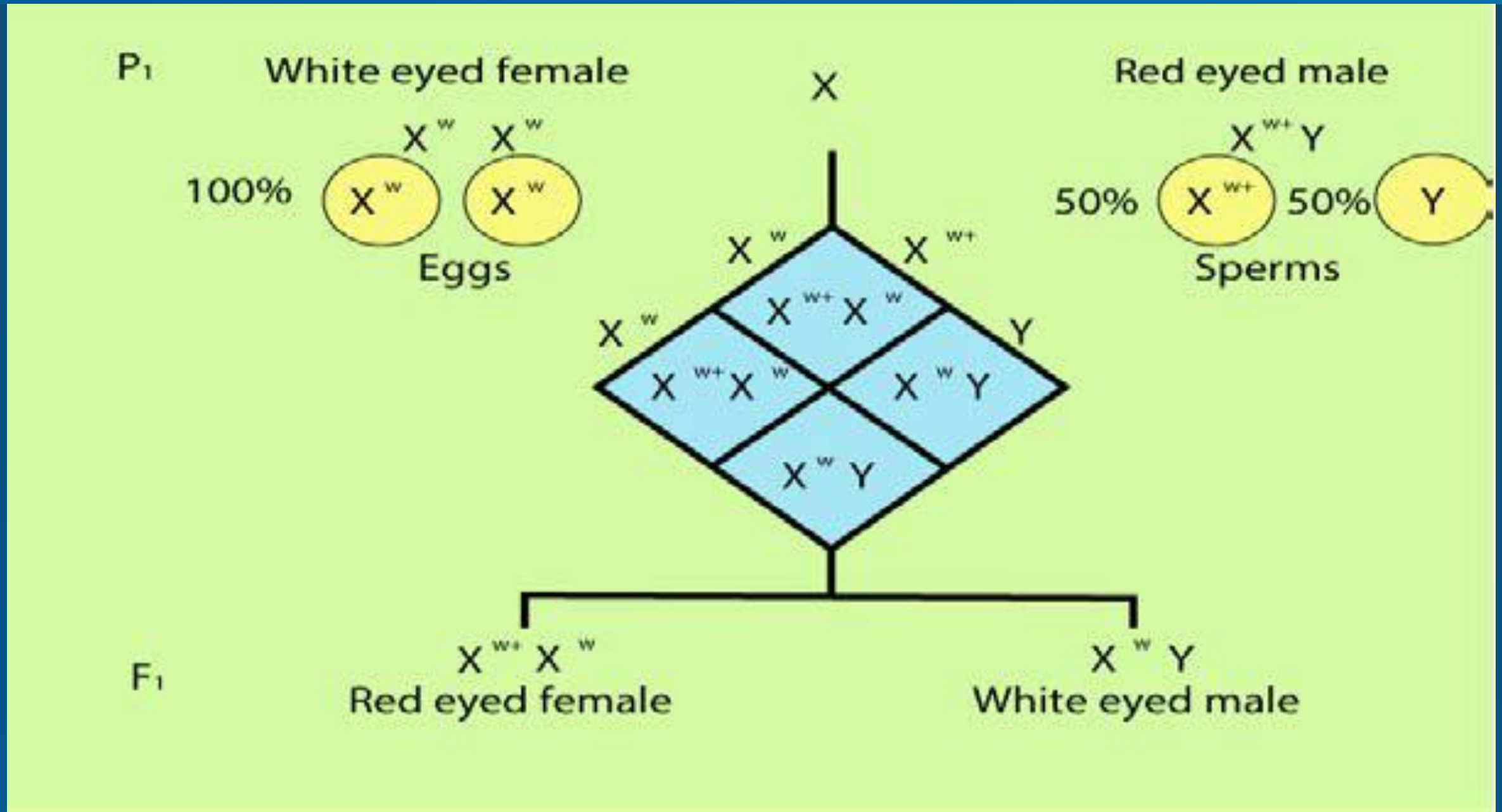
Test Cross


Red eyed female of F1
X
White eyed male of P1
X^w+X^w
X^wY
Gametes
50%
X^w+
50%
X^w
50%
X^w
50%
Y
Eggs
Sperms
X^w+
X^w
X^w+X^w
Red eyed
X^w
Y
X^wX^w
White eyed
X^w+Y
Red eyed
X^wY
White eyed
Offspring to test cross

Reciprocal cross as a confirmatory test

- Appearance of **white eyed female** provided an opportunity for a further confirmatory test. Morgan mated a **white eyed female (X^wX^w)** with a **red-eyed male ($X^{w+}Y$)**. **All female offspring had red eyes**, and **all male offspring had white eyes**.
- Then these **F1 red eyed females** and **white eyed males** were mated to produce F2. Half of the F2 **females had red eyes**, **half had white**. Similarly half of the F2 **males had red eyes** and **half had white**.

Test Cross


P1
White eyed female
X
Red eyed male
X w X w
X w+ Y
100%
X w
X w
Eggs
50%
X w+
50%
Y
Sperms
X w
X w+
X w+ X w
X w
Y
X w+ X w
X w Y
X w Y
F1
X w+ X w
Red eyed female
X w Y
White eyed male

X-Linked Trait

- A trait whose gene is present on X chromosome is called X - linked trait. X - linked traits are commonly referred as sex-linked traits.
- A gene present only on X chromosome, having no counterpart on Y chromosome, is called X - linked gene.

Pattern of Inheritance of X-Linked Trait

- Sex-linked inheritance follows a very specific pattern.
- As a son inherits his X chromosome only from his mother, and a daughter gets an X chromosome from each parent.
- X- linked trait passes in a crisscross fashion from maternal grandfather (P1) through his daughter (F1) to the grandson (F2).
- It never passes direct from father to son because a son inherits only Y chromosome from father.

Y-Linked Traits and its Inheritance

- Y chromosome is not completely inert. It does carry a few genes which have no counterpart on X chromosome. Such genes are called Y-Linked genes and their traits are called Y-linked traits e.g. SRY gene on Y chromosome of man determines maleness.
- Y - linked traits are found only in males. These traits directly pass through Y chromosome from father to son only. As females do not normally inherit Y chromosome, such traits can not pass to them.

Genetics of Color Blindness

- Normal trichromatic colour vision is based on three different kinds of cone cells in the retina, each sensitive to only one of the three primary colours, red, green or blue.
- Each type of cone cell has specific light absorbing proteins called opsins. The genes for red and green opsins are on X chromosome, while the gene for blue opsin is present on autosome 7.
- Mutations in opsin genes cause three types of colour-blindness.

Genetics of Color Blindness

- A dichromat can perceive two primary colours but is unable to perceive the one whose opsins are missing due to mutation.
- **Protanopia** is red blindness, **deuteranopia** is green blindness, while **tritanopia** is blue blindness.
- Some people can detect red and green but with altered perception of the relative shades of these colours.
- They have abnormal but still partially functional opsins.
- They are **protanomalous** and **deuteranomalous** for red and green weakness respectively.

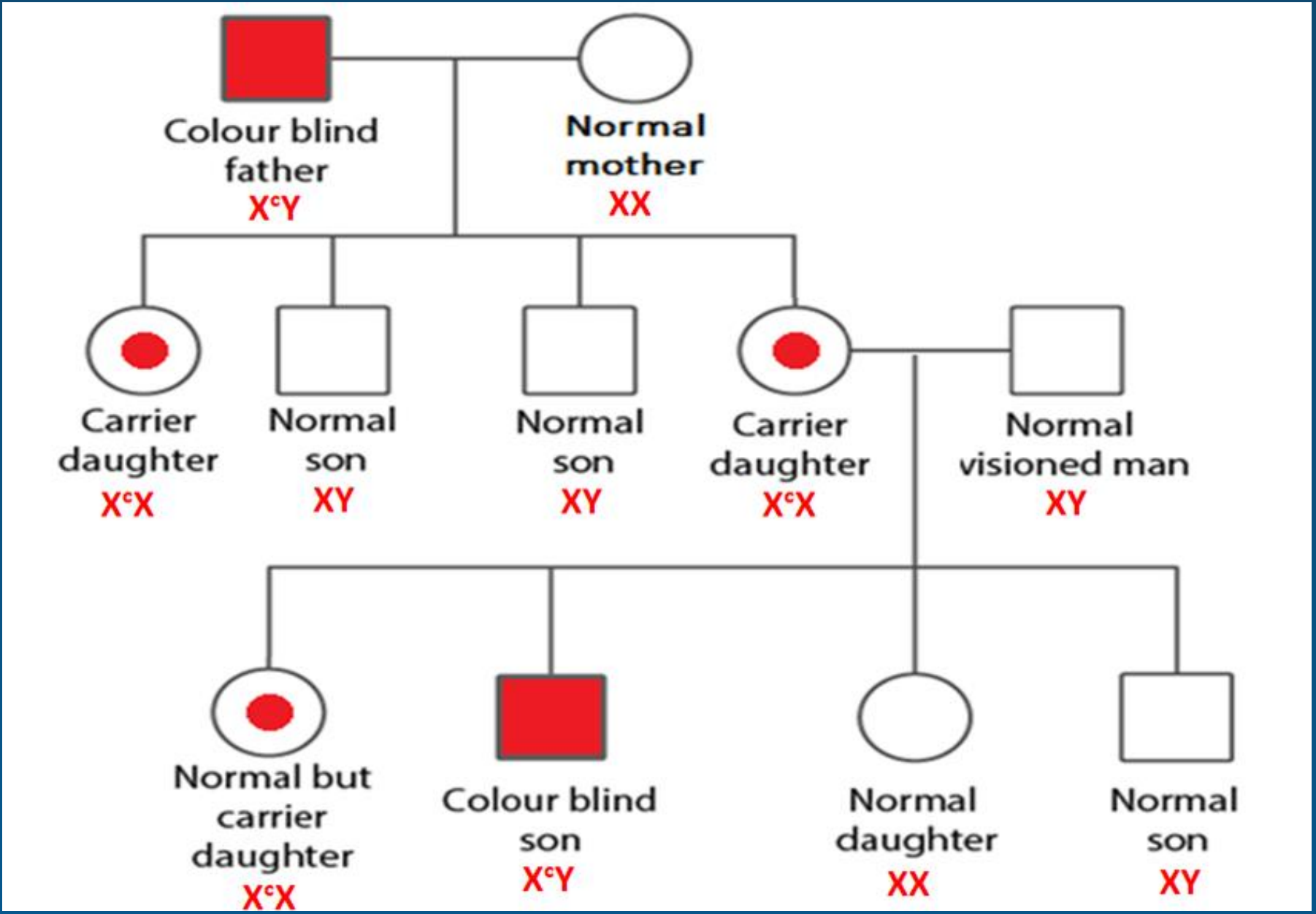
Genetics of Color Blindness

- A monochromat can perceive one colour. Monochromacy is true colour-blindness.
- Blue cone monochromacy is an X – linked recessive trait in which both red and green cone cells are absent. That is why it is also called **red - green colour-blindness**.
- It is a common hereditary disease. Like any sex - linked recessive trait, it also zigzags from maternal grandfather through a carrier daughter to a grandson. It never passes direct from father to son.
- This type of colour blindness is more common in men than women, because chances for a male to be affected by it are much more than a female.

Marriage between colour-blind man and normal visioned woman

- When colour-blind man marries with a normal visioned woman, then they will produce normal visioned male and female individuals in F1.
- The marriage between a F1 normal visioned woman and normal visioned male will produce two normal visioned female, one normal visioned male and one colour-blind male in F2.

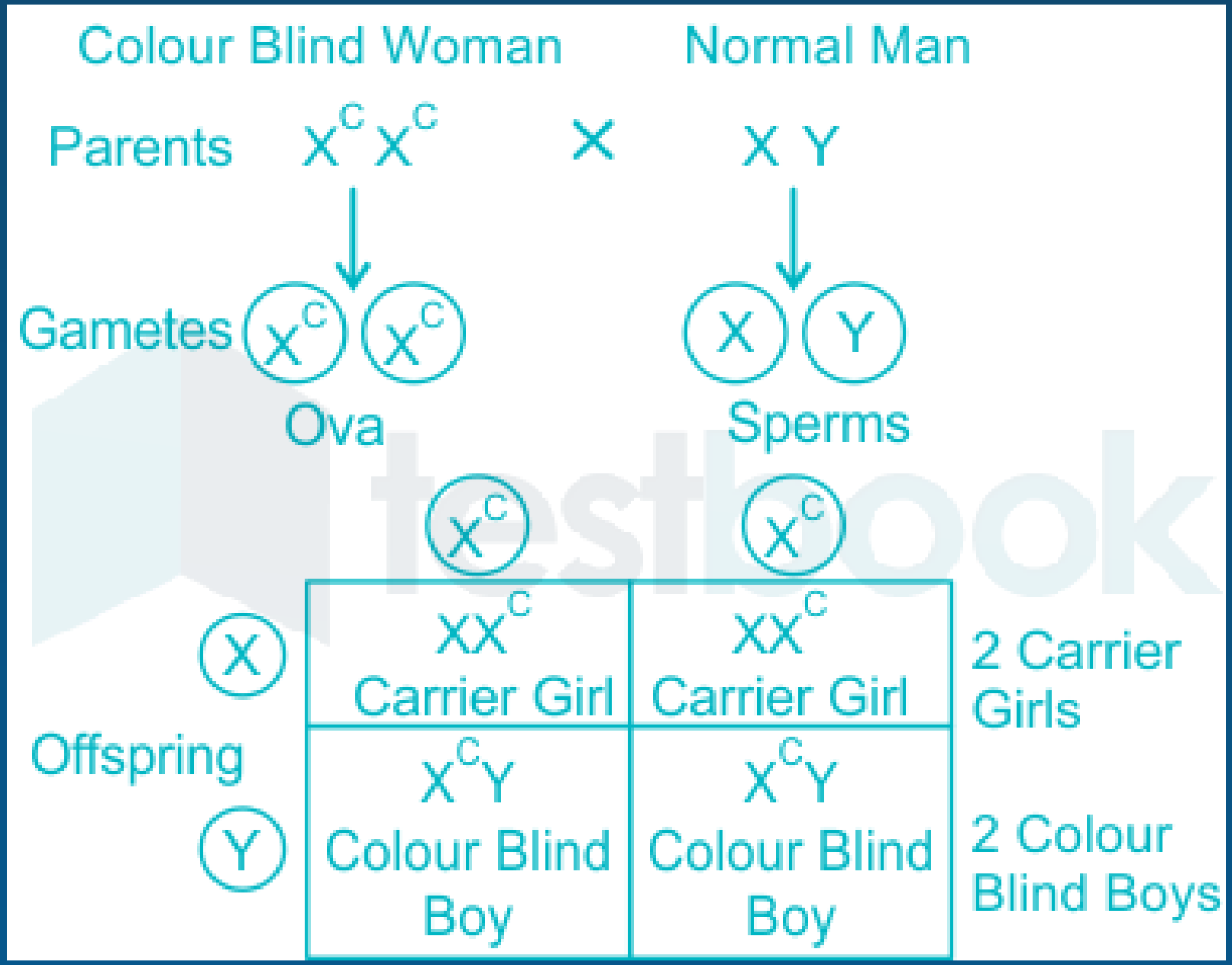
Pedigree of Color Blindness



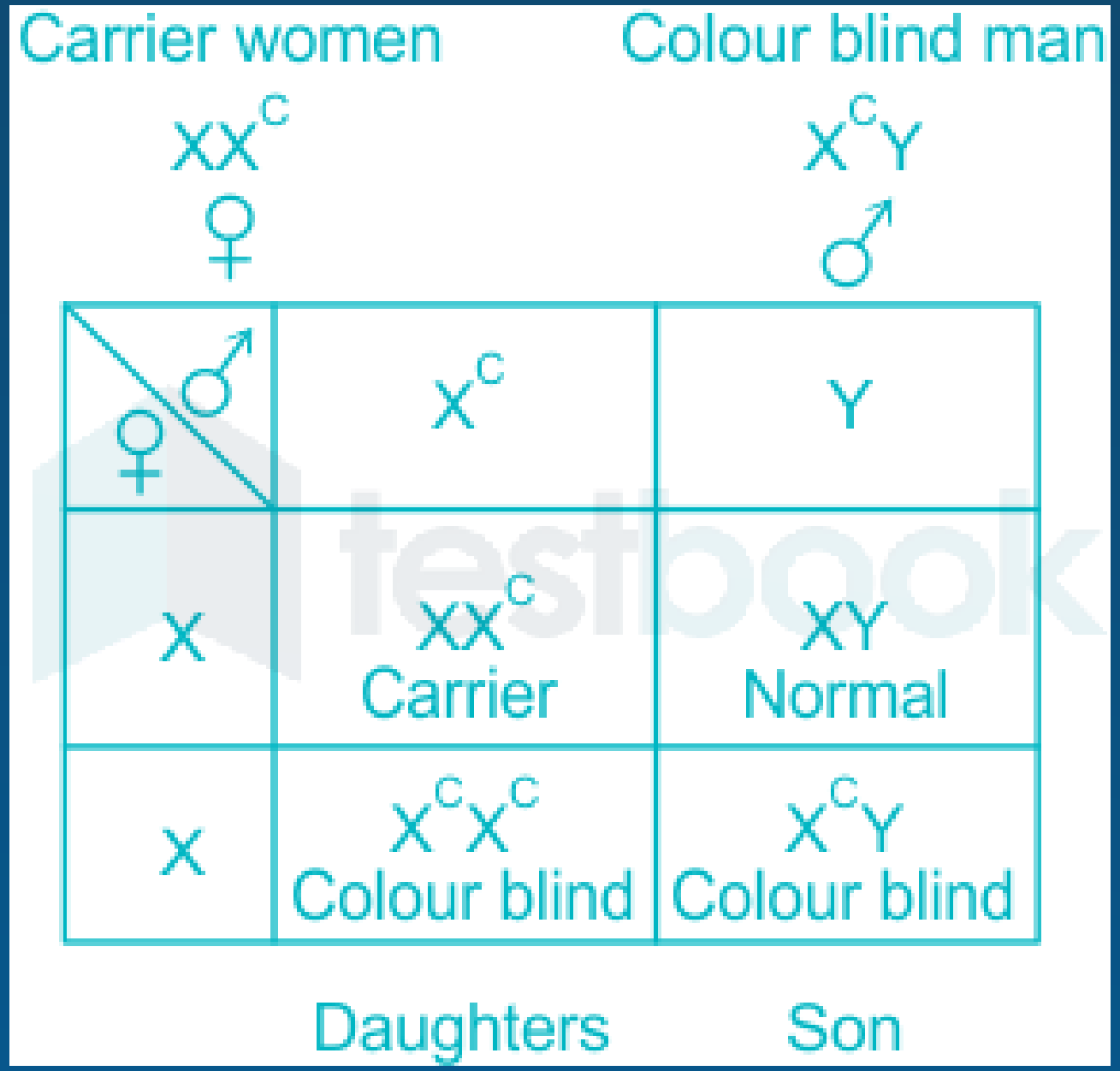
Marriage between normal visioned male and colour-blind female

- If a woman is colour blind and she happens to marry a normal visioned male, then all F1 sons will be colour blind and daughters will be normal visioned (carriers).
- Because male receives one X-linked recessive gene for colour-blindness from colour-blind mother.
- The daughter receives one X-linked dominant gene for normal vision from father and one X-linked recessive gene for colour-blindness from the mother.
- The F1 brother and sisters, if inbred or married they will produce in F2 a colour-blind homozygous daughter, a normal visioned heterozygous daughter, a normal visioned son and a colour blind son.

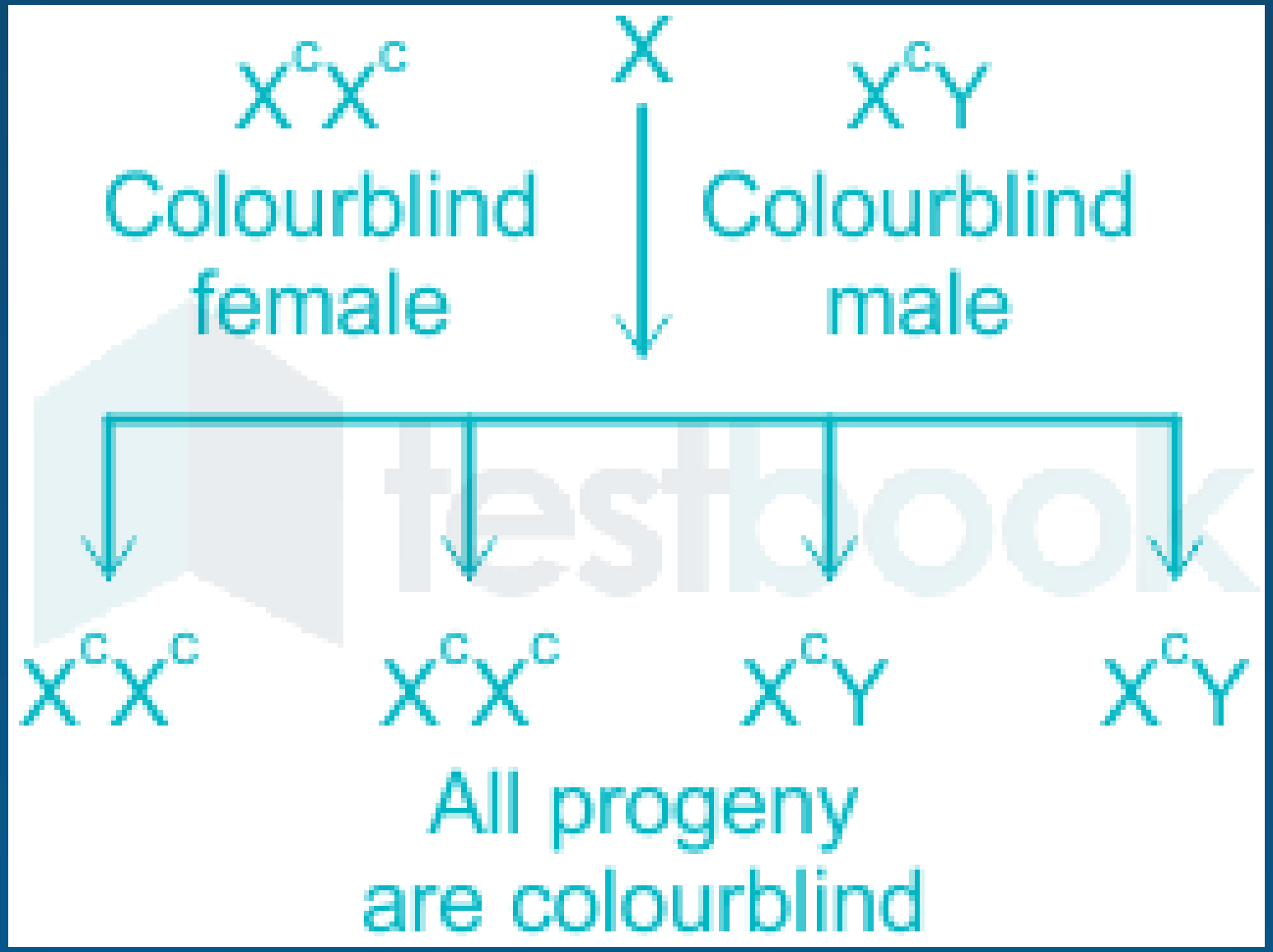
Pedigree of Color Blindness



Pedigree of Color Blindness



Pedigree of Color Blindness



Sex Limited Trait

- A sex-limited trait is limited to only one sex due to anatomical differences. Such trait affects a structure or function of the body present in only males or only females.
- These trails may be controlled by sex-linked or autosomal genes.
- Genes for milk yield in dairy cattle affect only cows.
- Similarly beard growth in humans is limited to men. A woman does not grow a beard herself but she can pass the genes specifying heavy beard growth to her sons.

Sex Influenced Trait

- Sex influenced trait occurs in both males and females but it is more common in one sex. It is controlled by an allele that is expressed as dominant in one sex but recessive in the other. This difference in expression is due to hormonal difference between the sexes.
- Pattern baldness is a sex influenced trait. Many more men than women are bald. It is inherited as an autosomal dominant trait in males but as an autosomal recessive trait in females. A heterozygous male is bald but a heterozygous female is not. A woman can be bald only when she is homozygous recessive.


Thank
You!