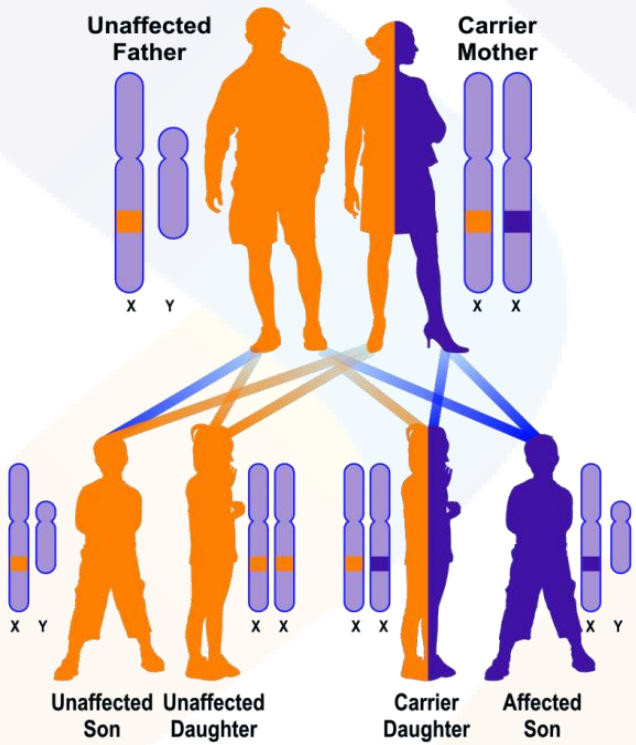
X-linked Recessive, Carrier Mother



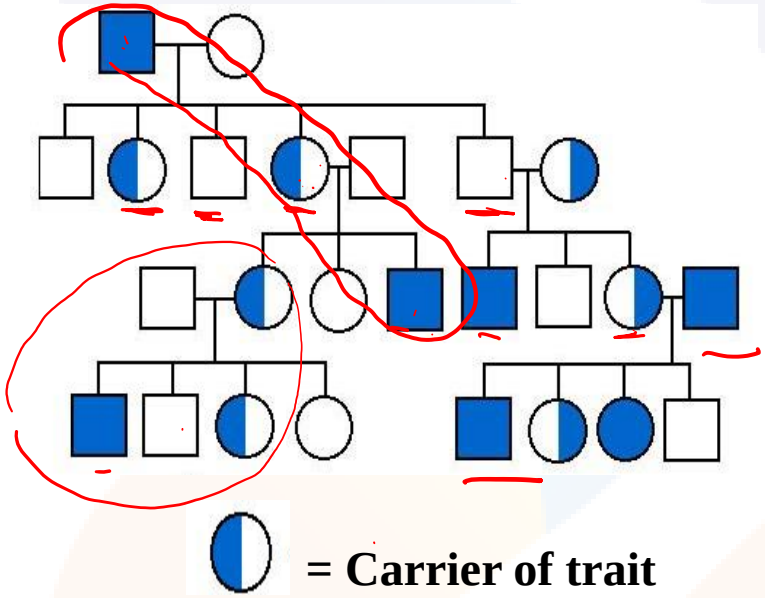
25% **Normal male**

25% **Normal female**

25% **Carrier female**

25% **Colorblind male**

Inheritance of Red-Green color blindness : an X-linked Recessive Trait



Male

Female

Affected male

Affected female

= Carrier of trait

Mating

Parents

Siblings

Males are more affected

Disease gets transferred from mother to son and father to daughter

If generation skipped → Recessive

Never from Father to Son.

Haemophilia

Bleeder's disease

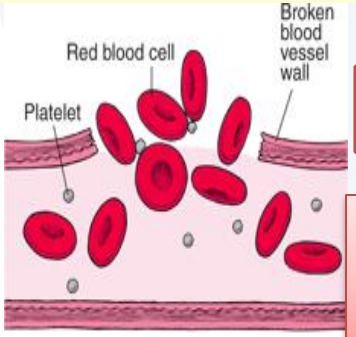
Recessive X–linked genetic disorder

How is it caused??

80% of Haemophilia cases are because of this factor

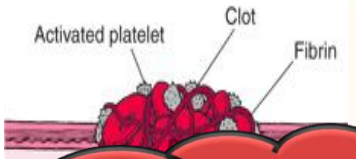
This disorder is due to lack of the functional clotting factor VIII

Haemophilia



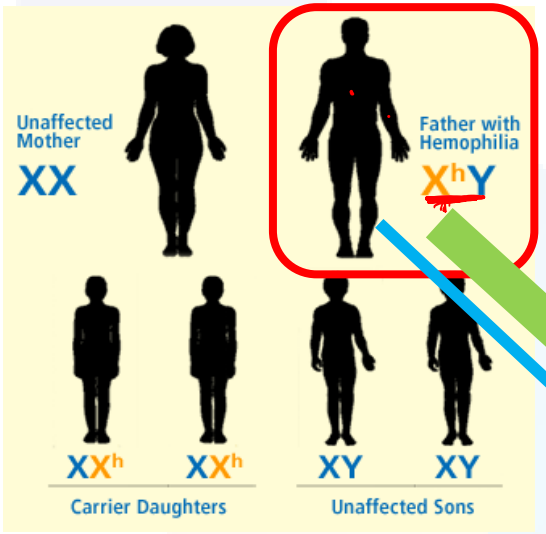
Blood fails to clot or clots very slowly

Recessive gene for haemophilia has deficiency of clotting factors (factor VIII or IX)



Bleeder's disease

Normal blood clotting

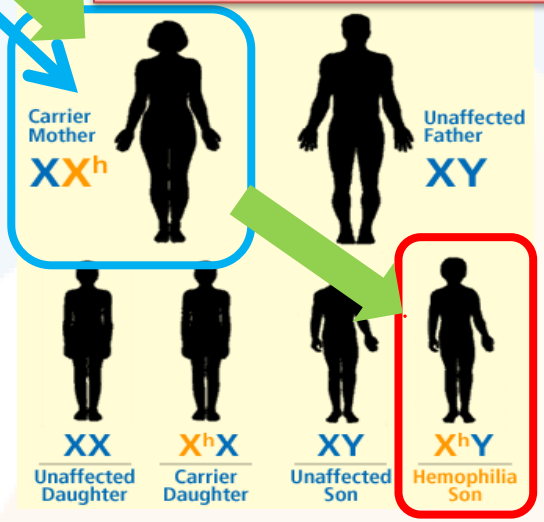


Haemophilic father

Transmits disease

Daughter

Grandson

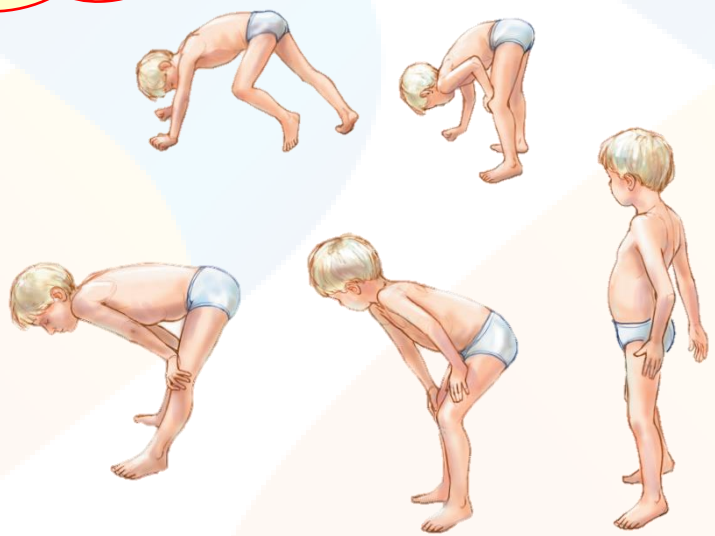


Criss cross inheritance

DUCHENNE MUSCULAR DYSTROPHY

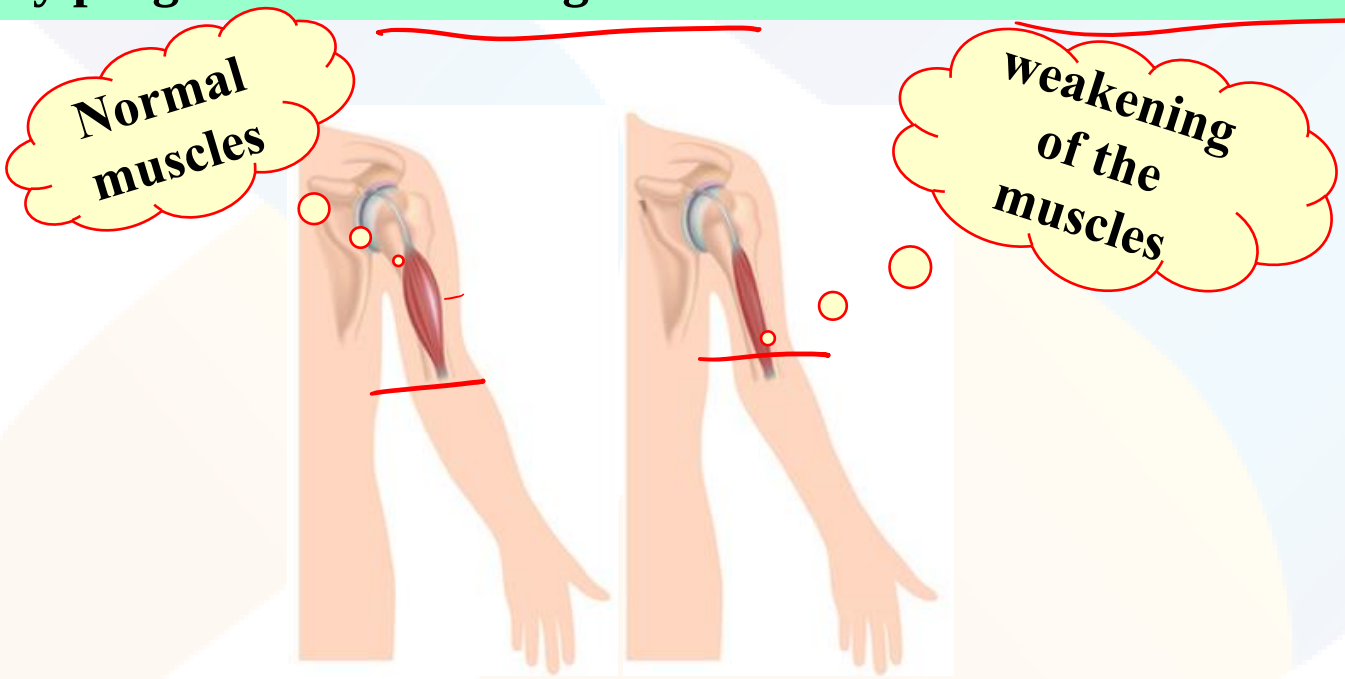
Affects 1 in 3600 boys.

X–linked form of muscular dystrophy.

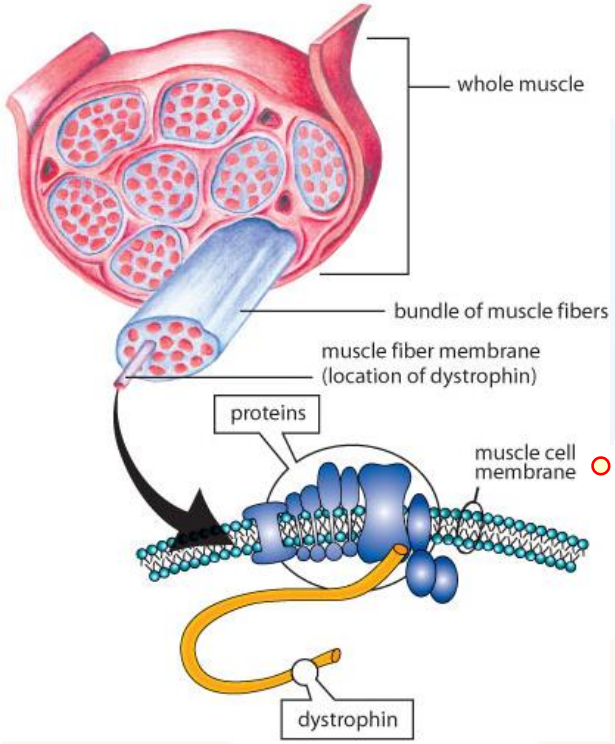


DUCHENNE MUSCULAR DYSTROPHY

By progressive weakening of the muscles and loss of coordination.



DUCHENNE MUSCULAR DYSTROPHY

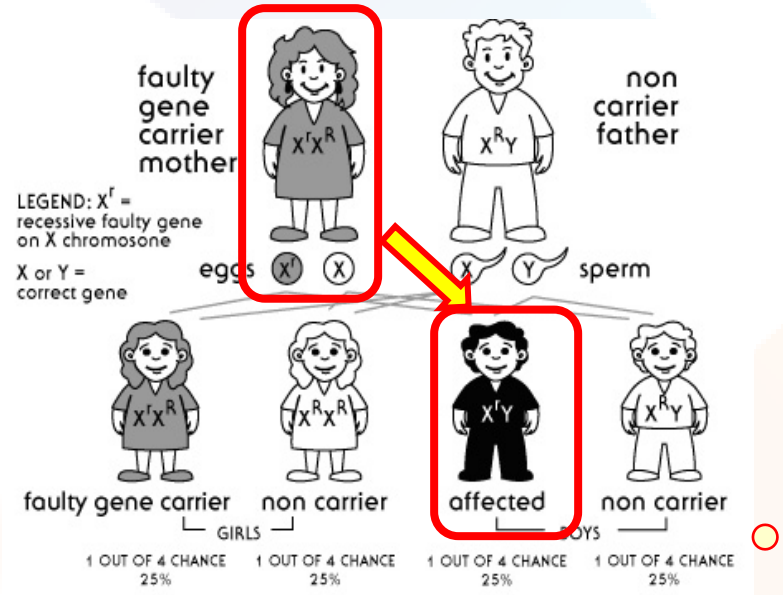


Dystrophin is an important structural component within muscle tissue.

Codes for the protein dystrophin.

DUCHENNE MUSCULAR DYSTROPHY

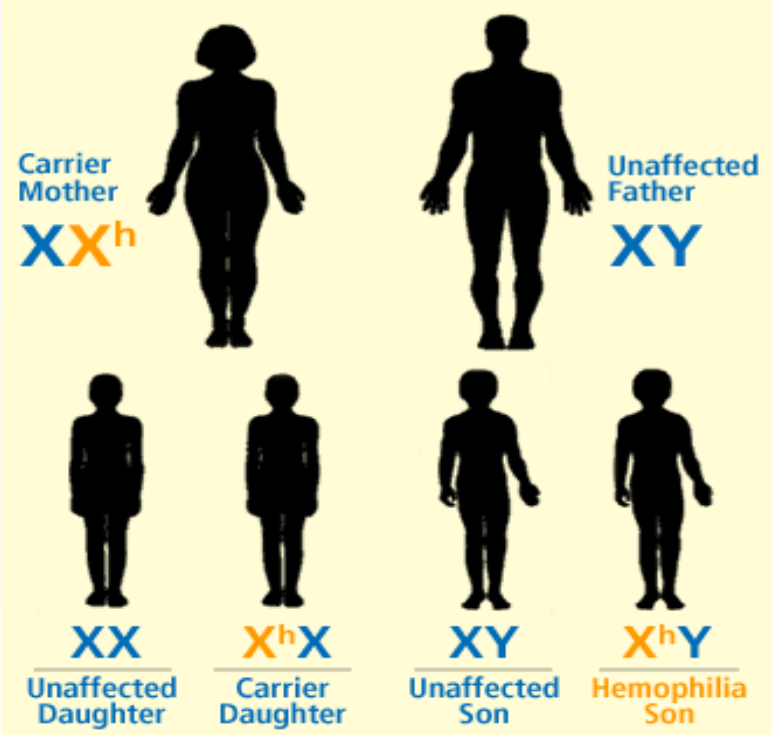
The mother's carrier gene is expected to be affected to half of her male children.



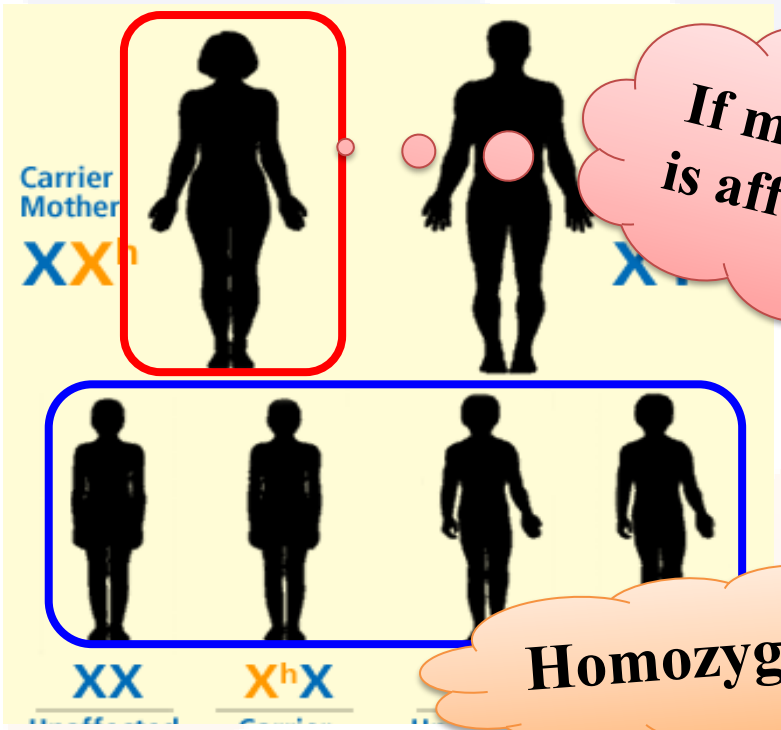
All fo… …ildren … …rier … are expected to be normal.

Whereas

PATTERN OF INHERITANCE (X-LINKED RECESSIVE)



PATTERN OF INHERITANCE



If mother is affected

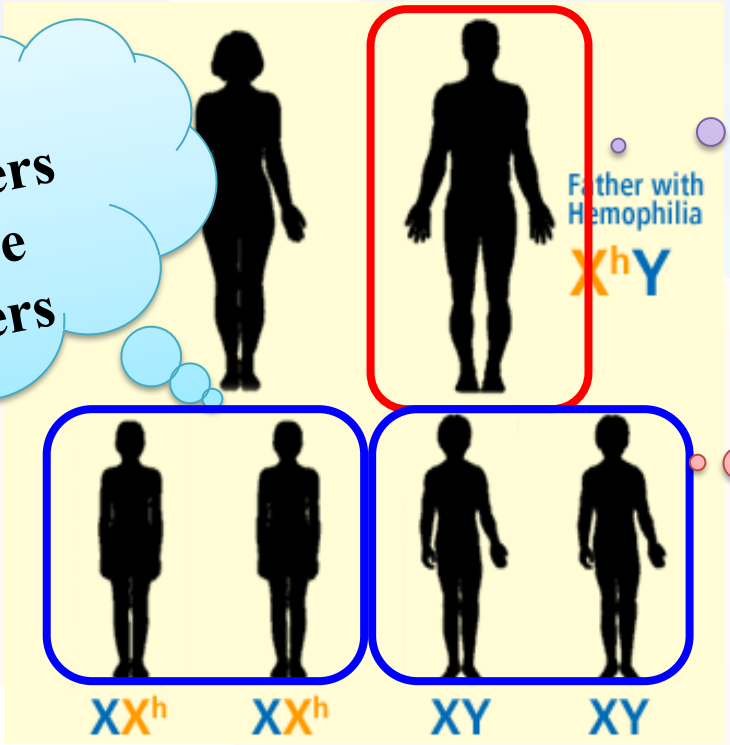
all her children are affected

Homozygous

PATTERN OF INHERITANCE

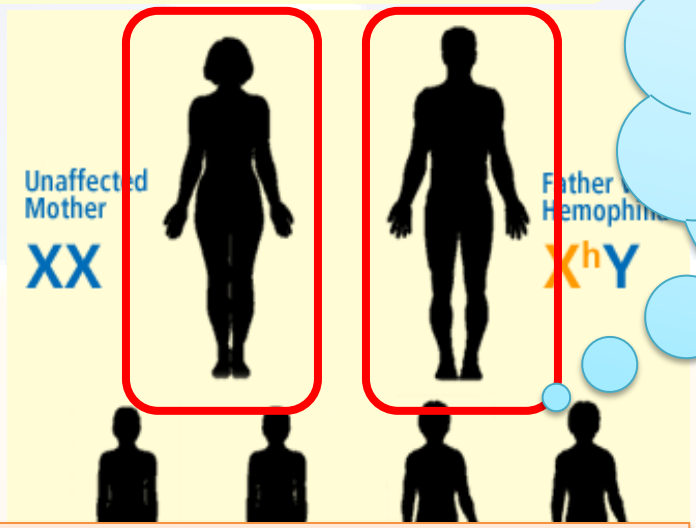
If father is affected

All daughters will be carriers



All the sons are normal

PATTERN OF INHERITANCE



Males are more severely affected than heterozygous females.

Because males do not have a normal copy of the gene to balance the effects of the mutation on their single X-Chromosome.

FOLLICULAR HYPERKERATOSIS

Also called as phrynoderma

X-linked dominant trait

FOLLICULAR HYPERKERATOSIS

Excessive development of keratin in hair follicles

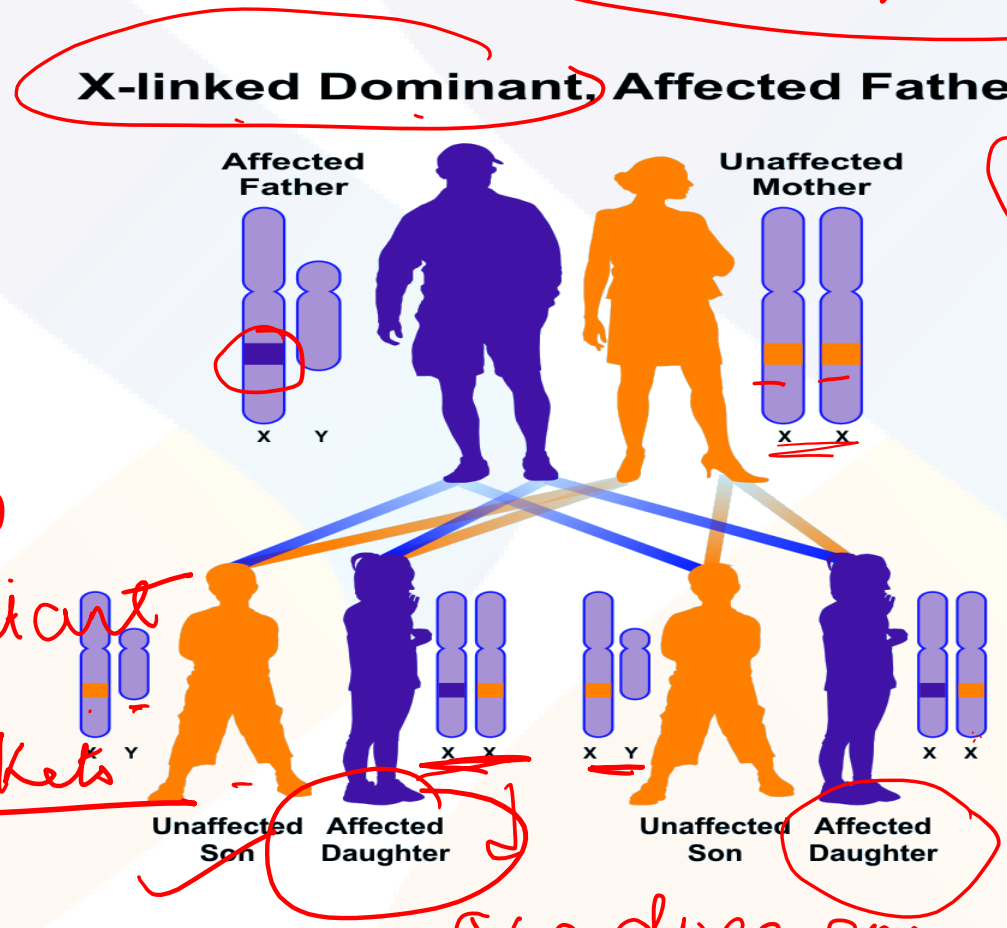
Resulting in rough, cone shaped, elevated papules.

The openings are often closed with a white plug of encrusted sebum.

Skin condition is characterized by

Here is female

Tricks



Disease never transfers from father to son

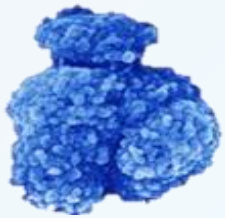
eg Vit D resistant Rickets

(2) All daughters of an affected father will be affected

One dose enough to cause disease

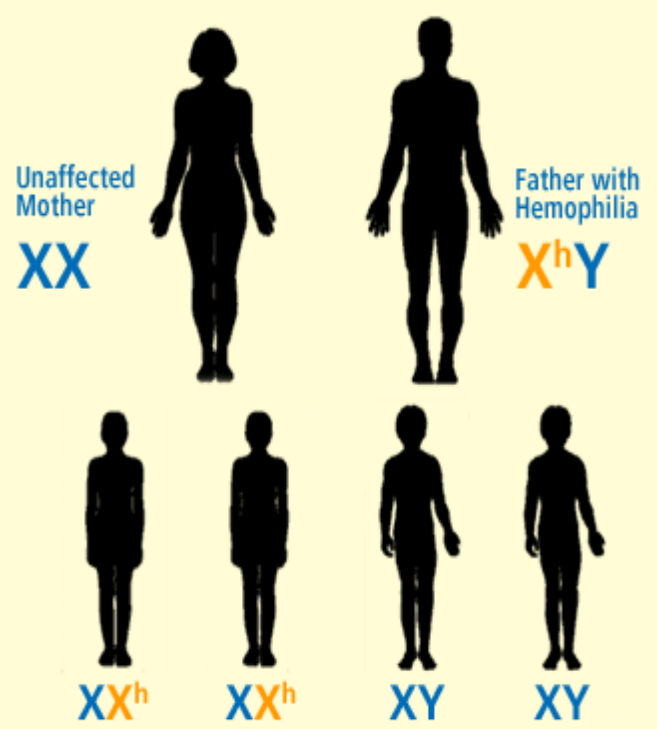
Y-LINKED INHERITANCE

Also called
Holandric
Inheritance



The genes that control Y-linked characters of Y- chromosomes.

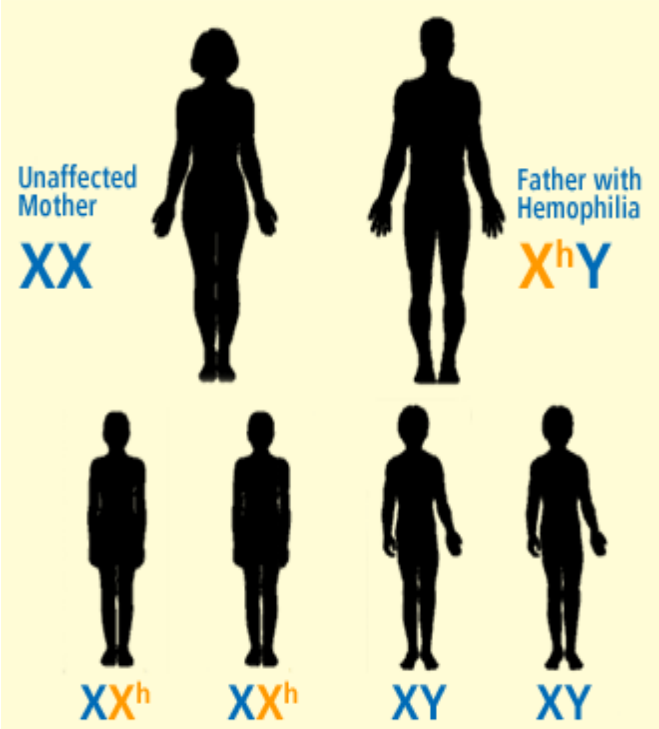
Y-LINKED INHERITANCE



Female offsprings of affected fathers are never affected.

Because females inherit only an X chromosome from their father and never a Y chromosome..

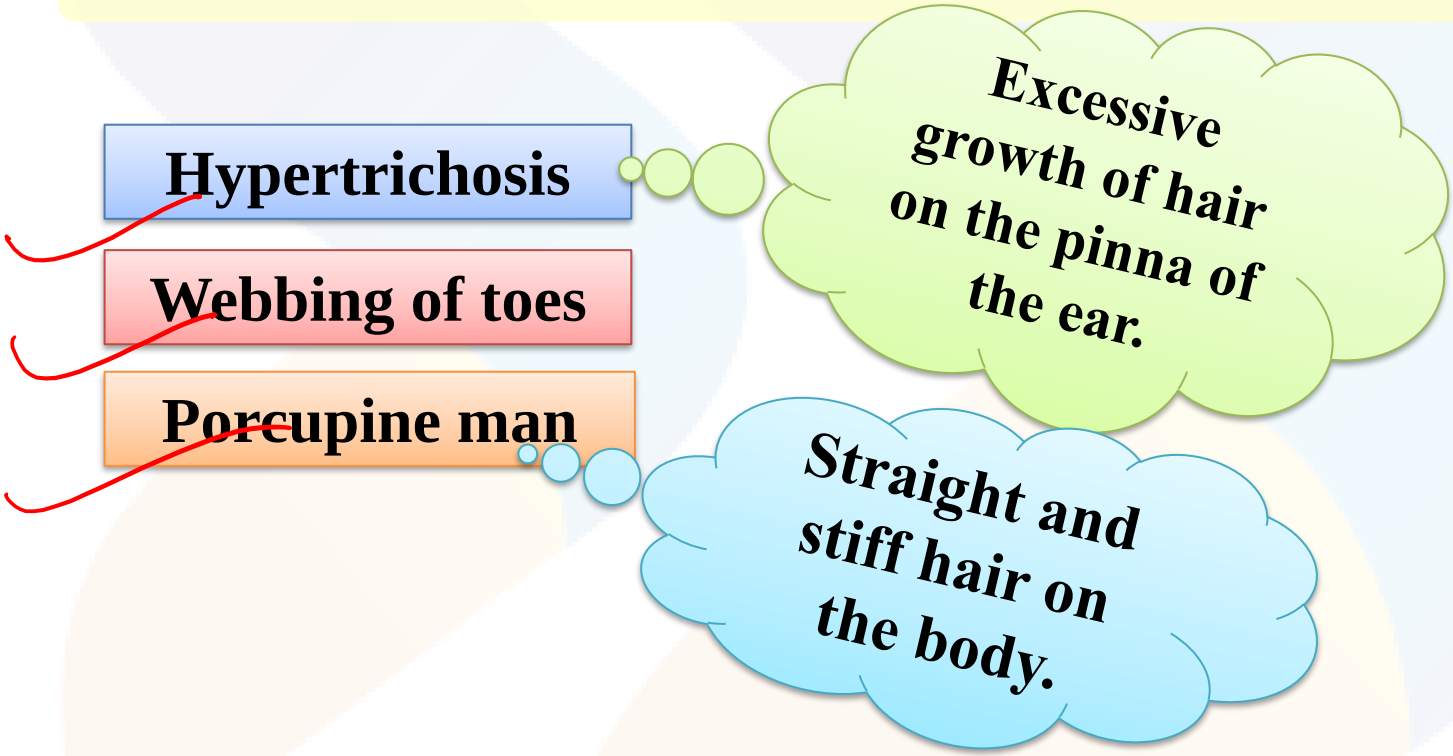
Y-LINKED INHERITANCE



Males are hemizygous for these genes.

SRY gene is on the Y chromosome .

Y-LINKED CHARACTERS IN HUMAN BEING ARE



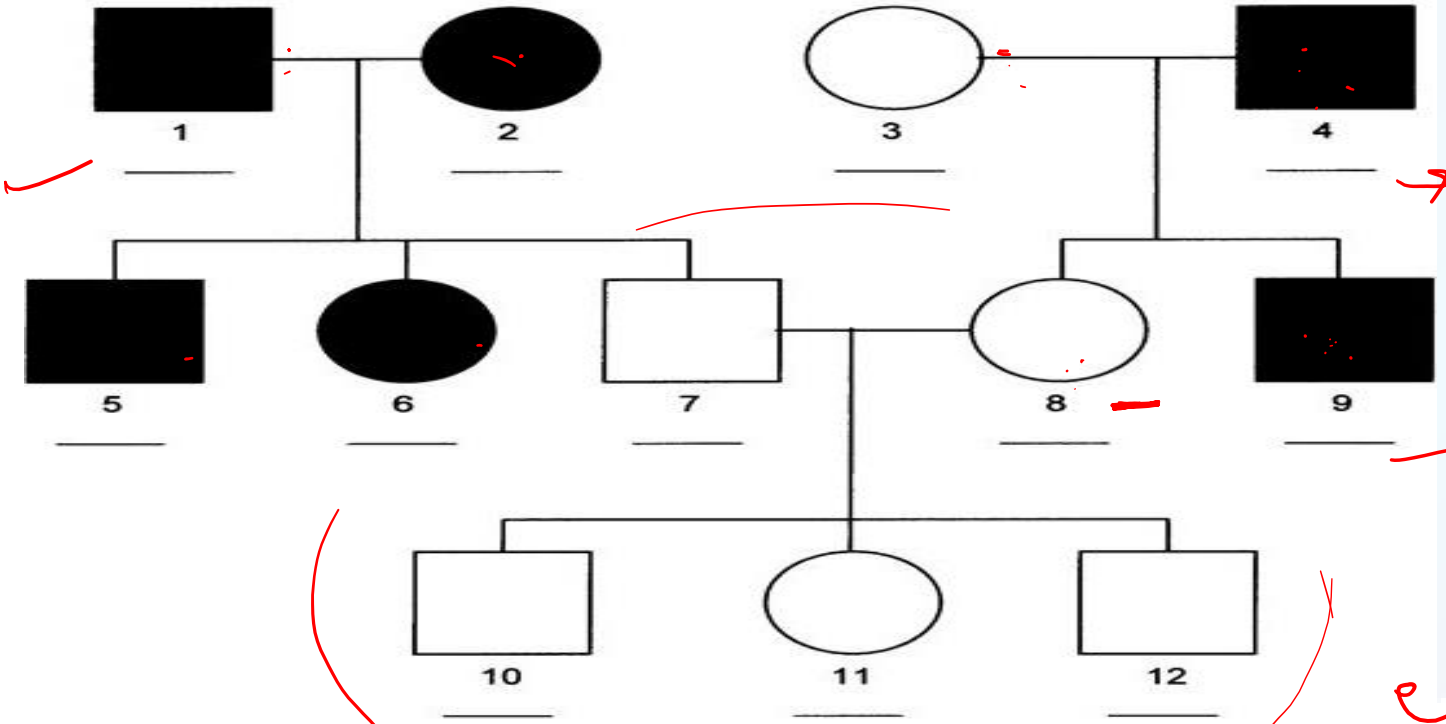
PEDIGREE ANALYSIS

Pedigree analysis helps to work out the possible genotypes from the knowledge of the respective phenotypes.

The possible genetic makeup of a person for a trait can also be known with the help of the pedigree chart.

It helps to study the pattern of inheritance of a dominant or a recessive trait.

AUTOSOMAL DOMINANT



→ Does not skip generations

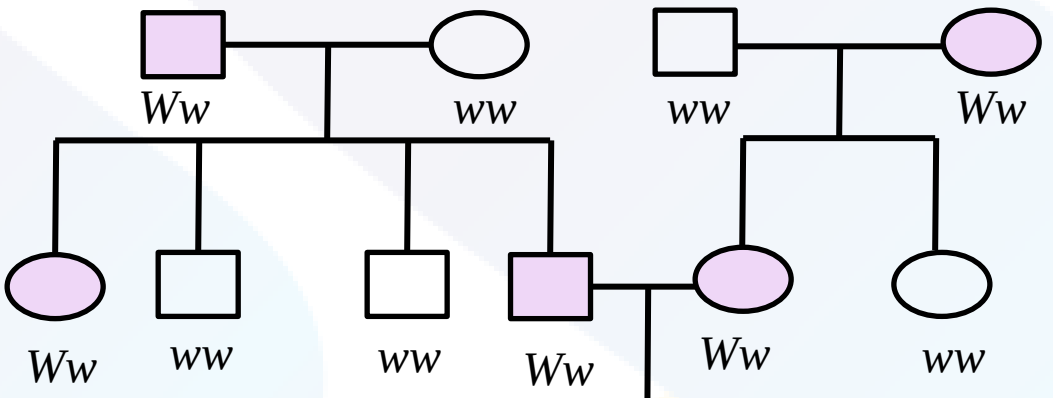
→ Affected parents can have unaffected children

eg Huntington's disease

1st generation (grand parents)

2nd generation (parents, aunts, and uncles)

3rd generation (two sisters)



Widow's peak

No widow's peak

AUTOSOMAL RECESSIVE

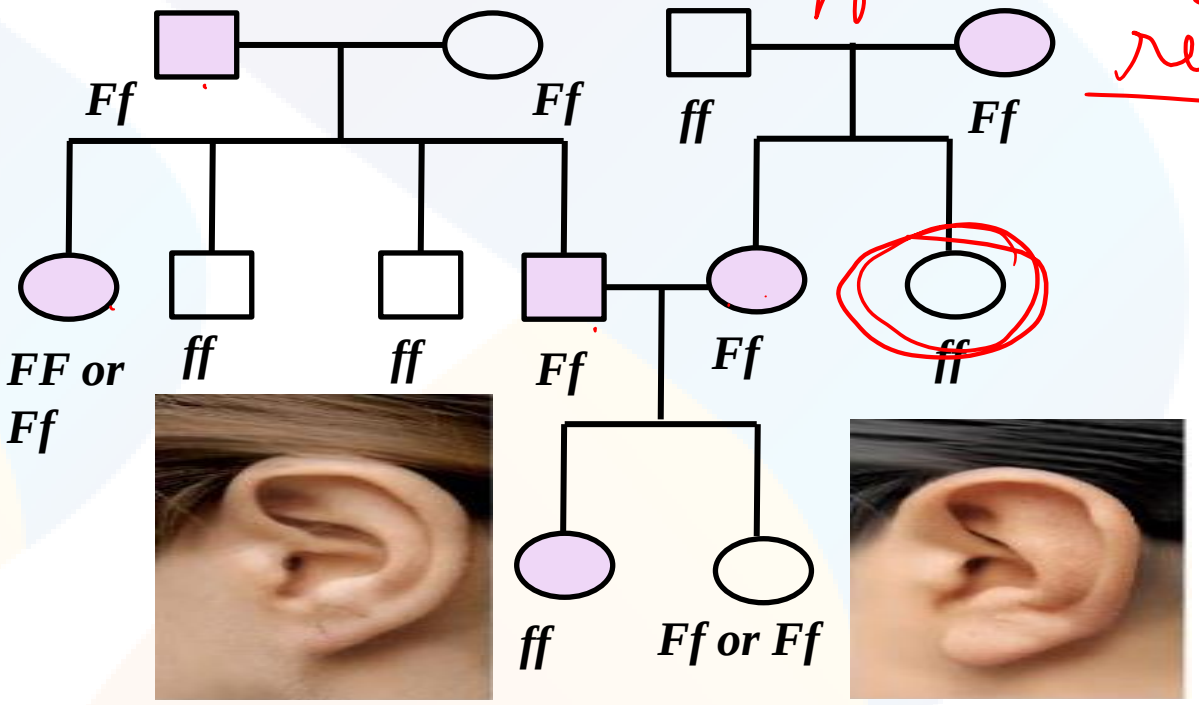
skips generation

unaffected parent can have affected children

1st generation (grand parents)

2nd generation (parents, aunts, and uncles)

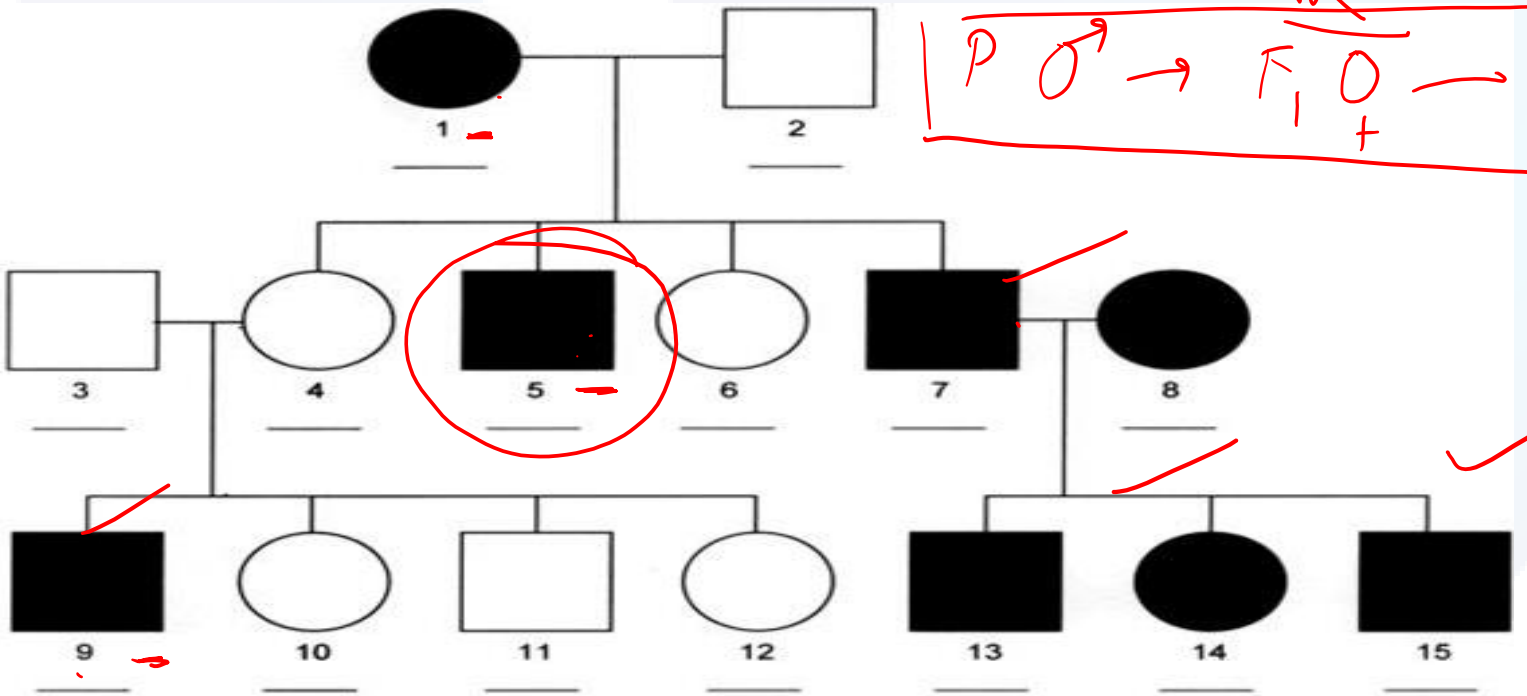
3rd generation (two sisters)



Attached earlobe

Free earlobe

X-LINKED RECESSIVE



Sex linked Inheritance

Humans have 22 pairs of autosomes and 1 pair of sex chromosome, (XX in females) and (XY in males).

X chromosomes may contains alleles for Haemophilia, Colorblind, baldness, Duchenne muscular dystrophy.

These X linked recessive disorder predominantly affects males.

Females are homologous to sex chromosomes: XX, while Males are heterologous to sex chromosomes: XY.

Y chromosomes is much smaller then X, males are hemizygous to X chromosome.

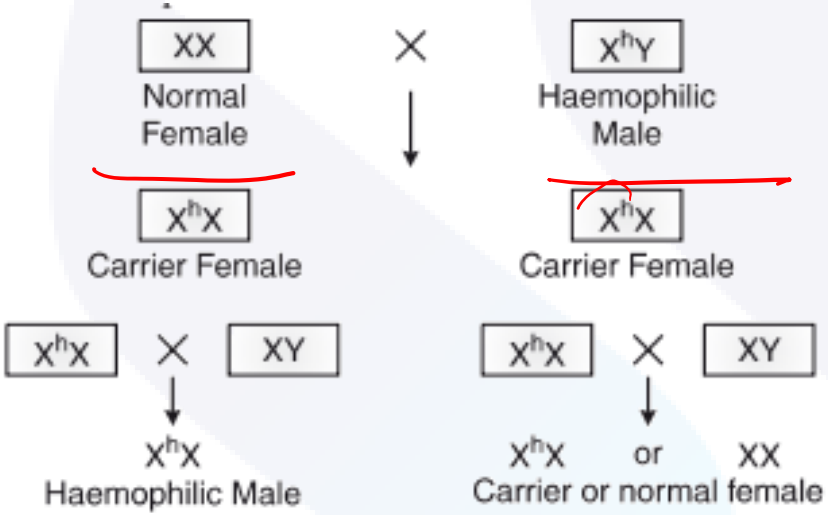
Haemophilia

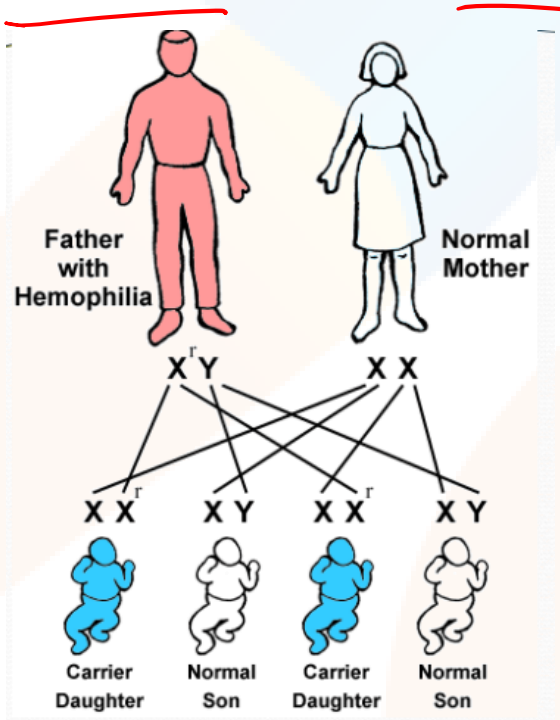
Hemophilia is a blood disorder where the blood doesn't clot properly. A minor cut can cause serious injury.

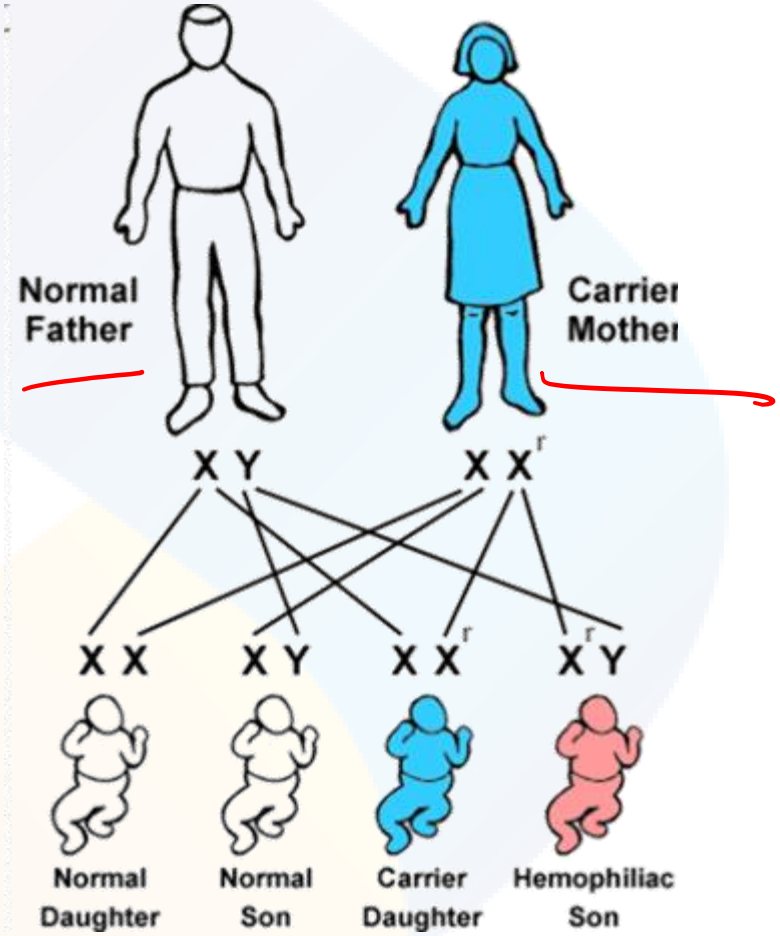
The gene for hemophilia is found on X chromosome, and it is of recessive type.

Males are more likely to get hemophilia because they are hemizygous. Females are carrier in heterozygous condition.

Inheritance of Hemophilia



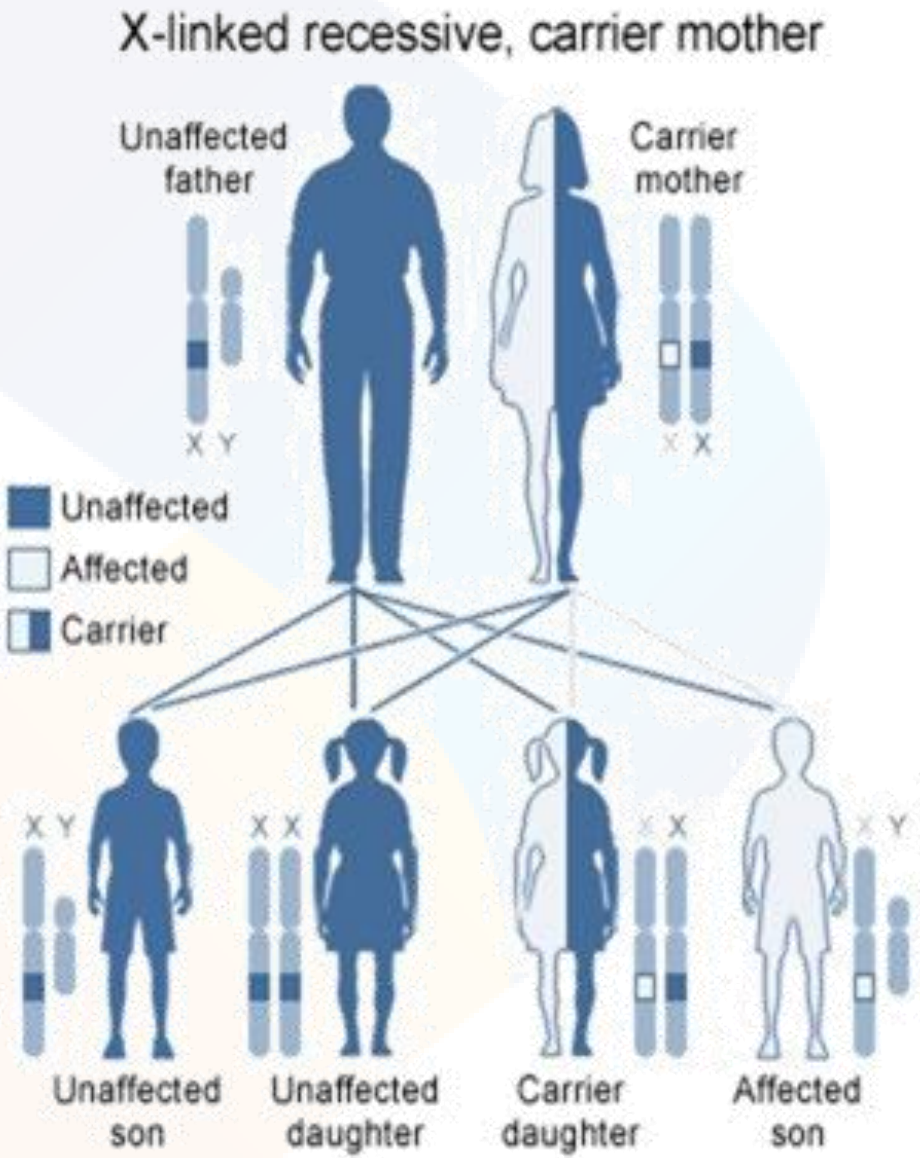




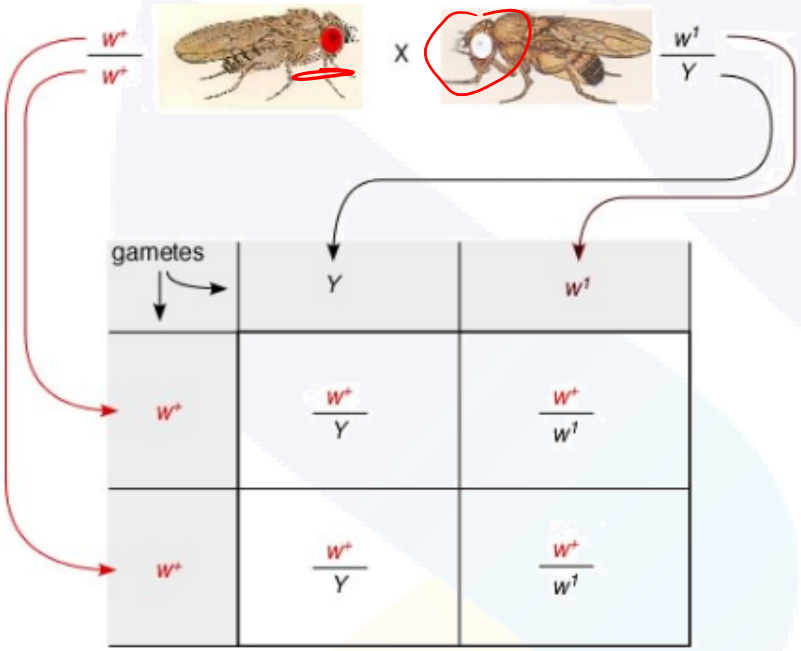
Colorblindness

Colorblindness also a sex-linked recessive disorder, its gene found on X-chromosome and recessive in nature.

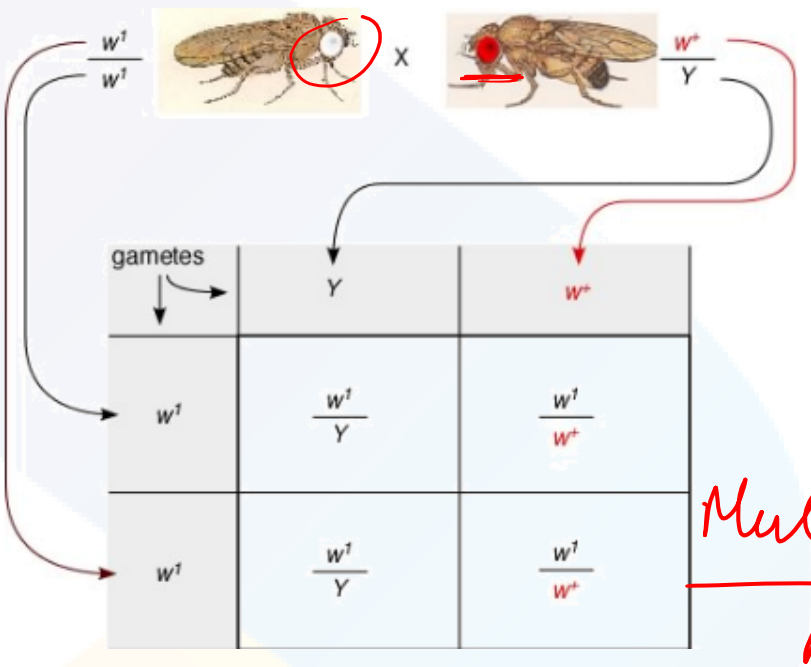
Dominant gene control the production of three different cone cells while a recessive gene only produce two type of cone cells. A colorblind person is not able to distinguish between red and green color.



Eye Color Inheritance in Drosophila

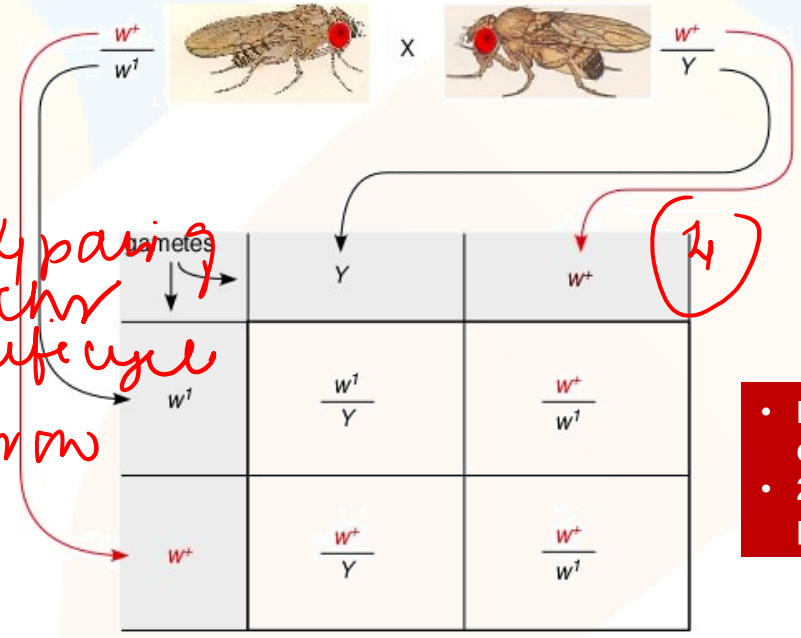


- Red eyed female crossed with white eye male.
- All progenies will show red color.



Mutant

- White eyed female crossed with red eyed male.
- All males will be with white eye.



1. Drosophila 4 pairs chr
2. 2 week lifecycle
3. easily grow

4

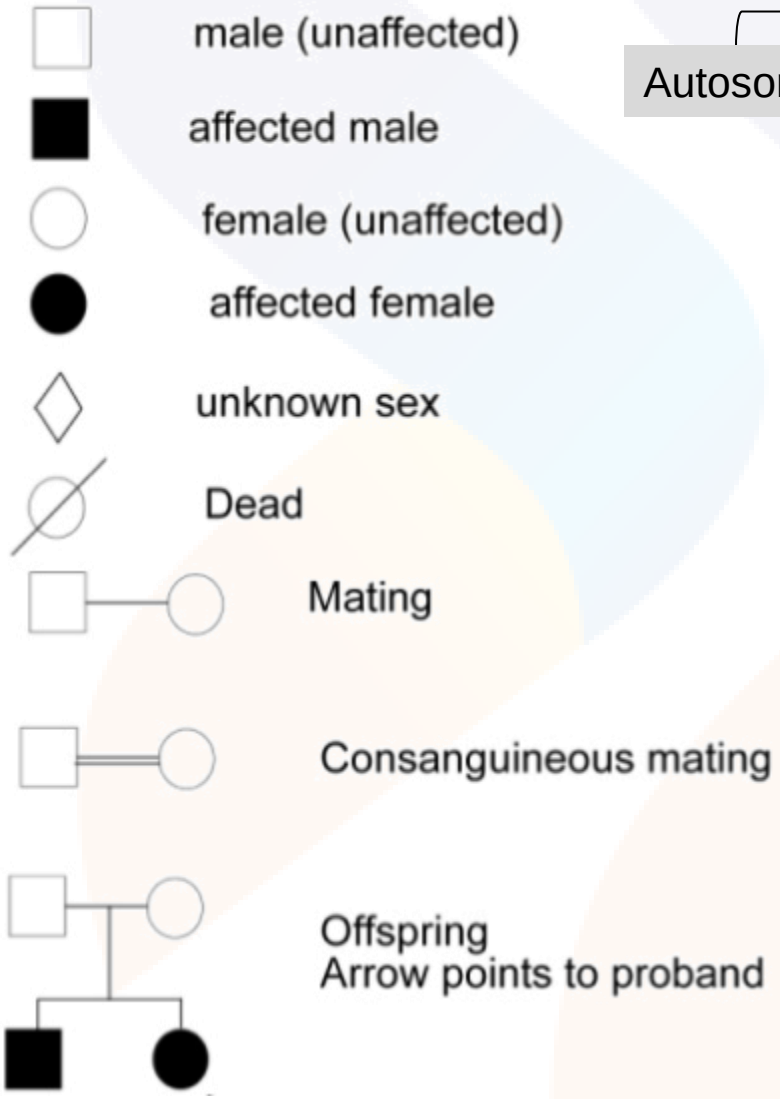
♂ & ♀ diff

- Red eyed female (carrier) crossed with red eyed male.
- 25% chances that the male progeny with white color eye.

Pedigree Analysis - Introduction

Used to determine mode of inheritance (recessive / dominant) in humans.

Symbols used in Pedigree analysis.



- **Inheritance**
  - Autosomal
  - X chromosome linked
  - Y chromosome linked

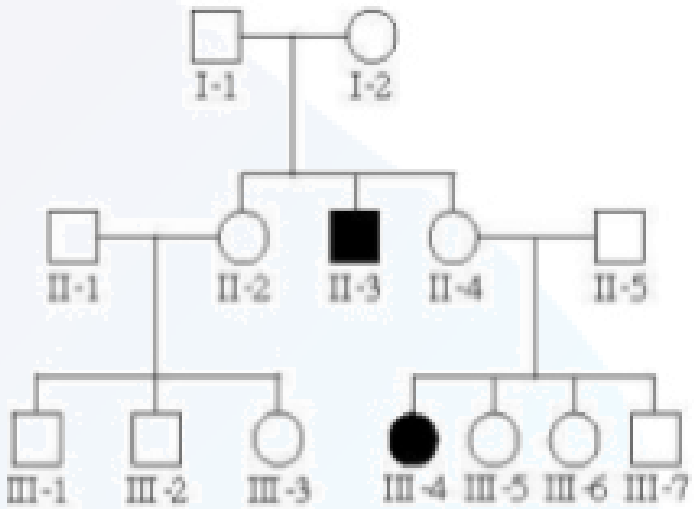
- **Autosomal**
  - Dominant: Huntington's disease
  - Recessive: PKU, Ty-Sachs albinism

- **X chromosome linked**
  - Dominant: hypophosphatemia
  - Recessive: Colorblindness, Haemophilia

- **Y chromosome linked**
  - Dominant:
  - Recessive:

Autosomal linked Inheritance

Autosomal Recessive Inheritance

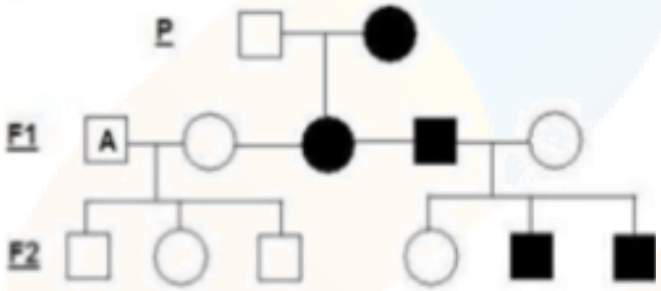


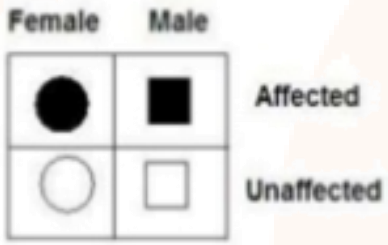
These are very rare, traits often skip generations

Traits affect male and female equally.

Diseases: Cystic fibrosis, sickle cell anemia, phenylketonuria (PKU), Tay-sachs disease.

Autosomal Dominant Inheritance





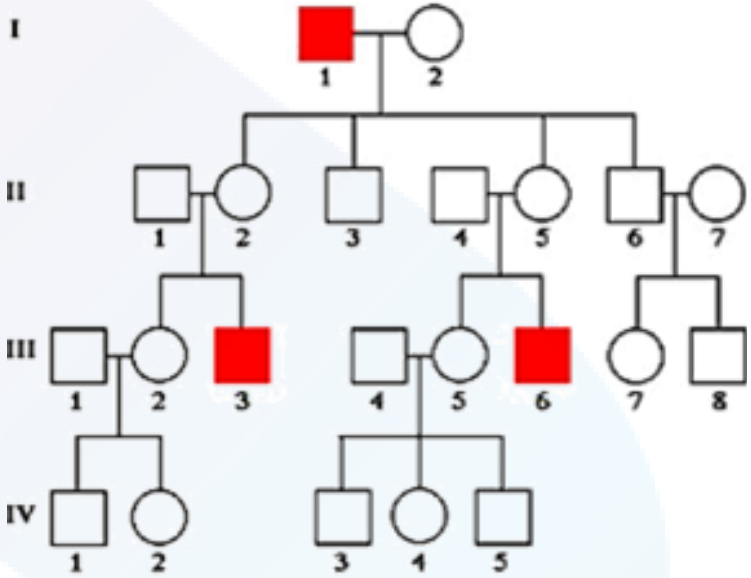
Trait is found in every generation.

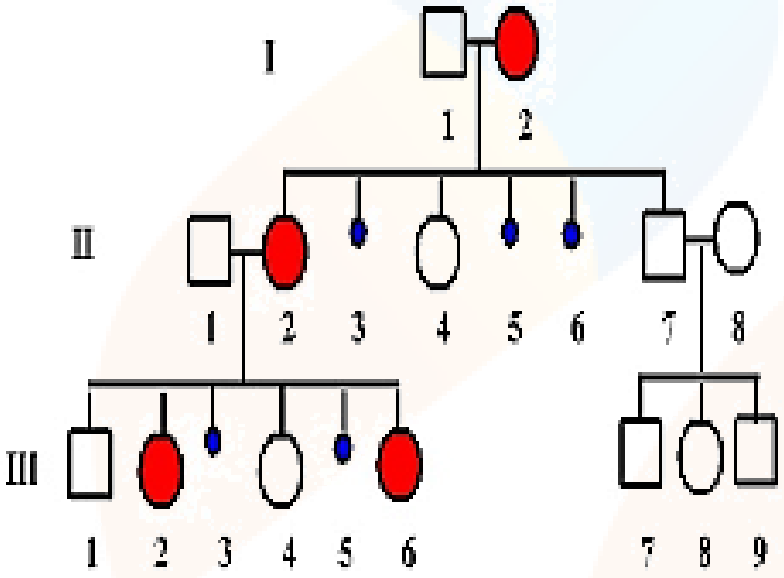
Affected individual also transmit the trait to about ½ of their children (regardless of sex).

Disease: achondroplasia (a skeleton disorder causing dwarfism), Huntington's disease.

X- linked Recessive and Dominant Pedigrees

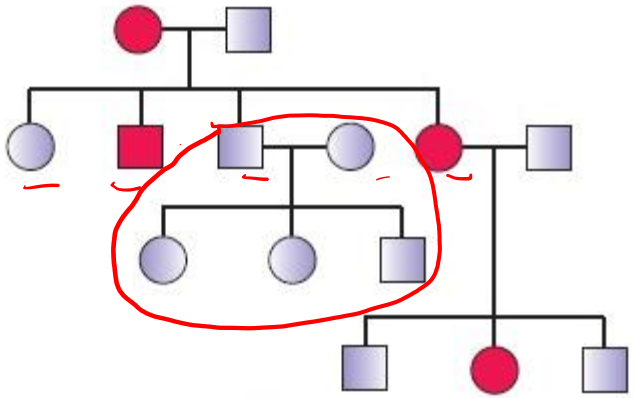
- Trait is rare in pedigree
- Trait skips generations
- Affected fathers DO NOT pass to their sons
- Males are more often affected than females
- Females are carriers (passed from mom to son)





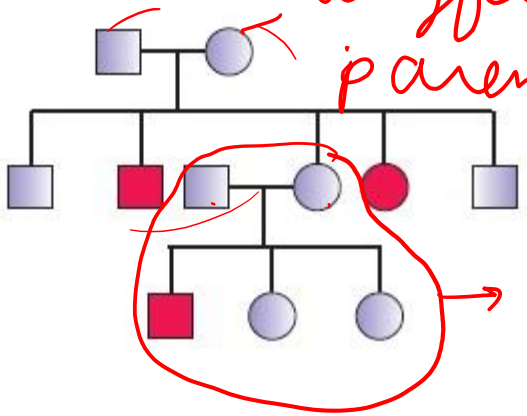
Ex. X – linked rickets (bone lesions)

- Trait is common in pedigree
- Affected fathers pass to ALL of their daughters
- Males and females are equally likely to be affected
- X - linked dominant diseases are extremely unusual
- Often, they are lethal (before birth) in males and only seen in females ex. incontinentia pigmenti (skin lesions) ked rickets (bone lesions)



(a)

Autosomal Dominant



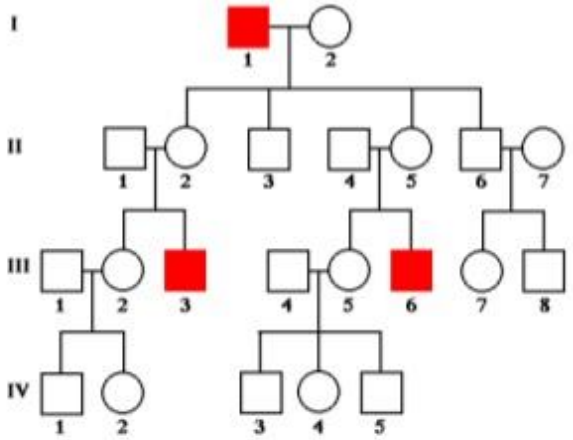
(b)

unaffected parent can have affected child

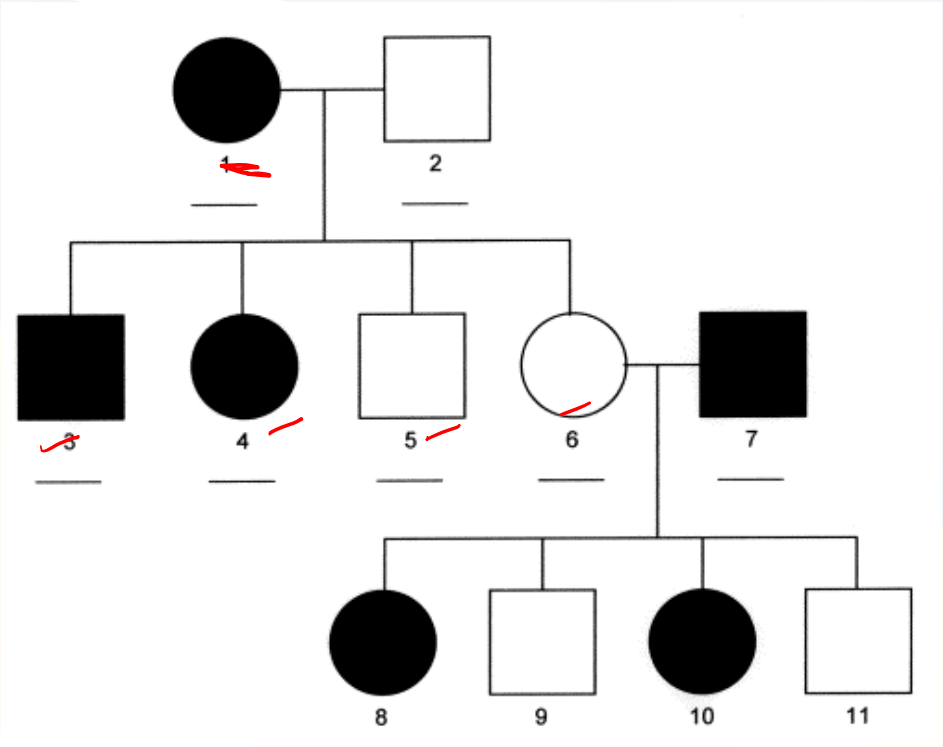
Autosomal Recessive

X-linked recessive pedigrees

- Trait is rare in pedigree
- Trait skips generations
- Affected fathers DO NOT pass to their sons
- Males are more often affected than females
- Females are carriers (passed from mom to son)



QUES

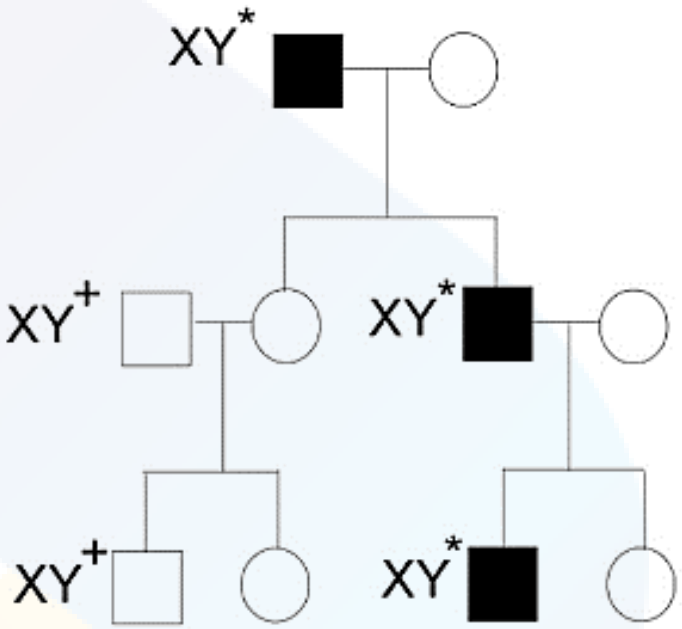


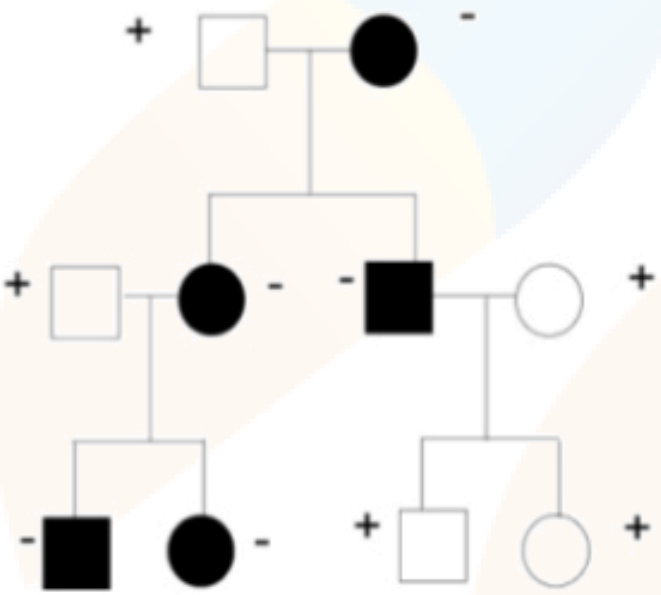
Autosomal

Dominant

Y- Linked Inheritance and Mitochondrial Genes

- Traits on the Y chromosome are only found in males, never in females.
- The father's traits are passed to all sons.
- Dominance is irrelevant: there is only 1 copy of each Y-linked gene (hemizygous).





- Mitochondria are only inherited from the mother.
- If a female has a mitochondrial trait, all her offspring inherit it.
- If a male has a mitochondrial trait none of his offspring inherit it.

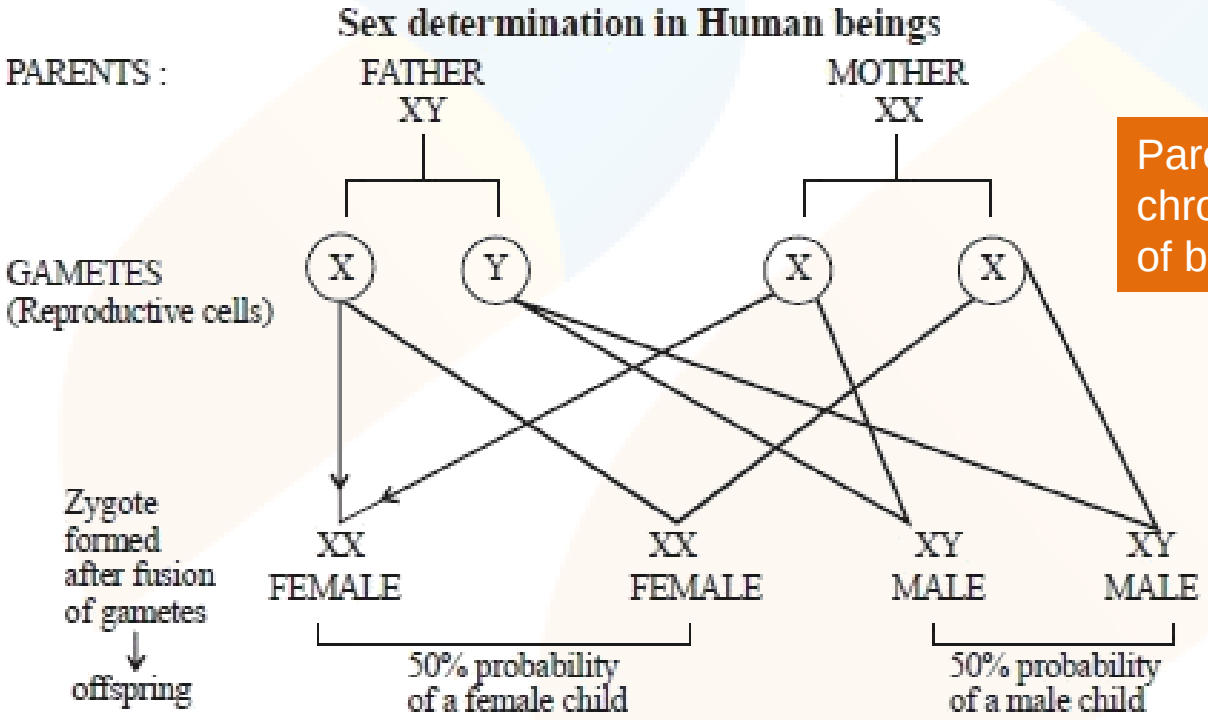
Sex Determination

Henking in 1891 observed a trace of specific nuclear structure in few insects and called this as x body. Later it was called as X chromosome and remaining are as autosomes.

In humans and other organisms XY types of sex determination is seen

In some insects like Drosophila XO type of sex determination is present.

In birds ZW type of sex determination is present, it also show female heterogamy.



Parent with heterologous chromosomes decide the sex of baby.

MALE HETEROGAMETY

Male Sex determination → Heterogametic condition

XX – XO type

XX – XY type

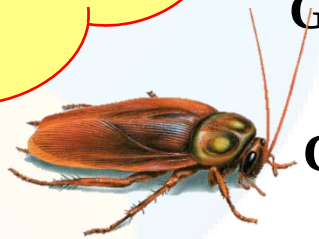
Males are Heterogametic i.e., Males produce dissimilar gametes and females are Homogametic.

XX - XO type of heterogametic sex determination

XX - XO type of heterogametic sex determination

Observed in grass hoppers

Grass hoppers

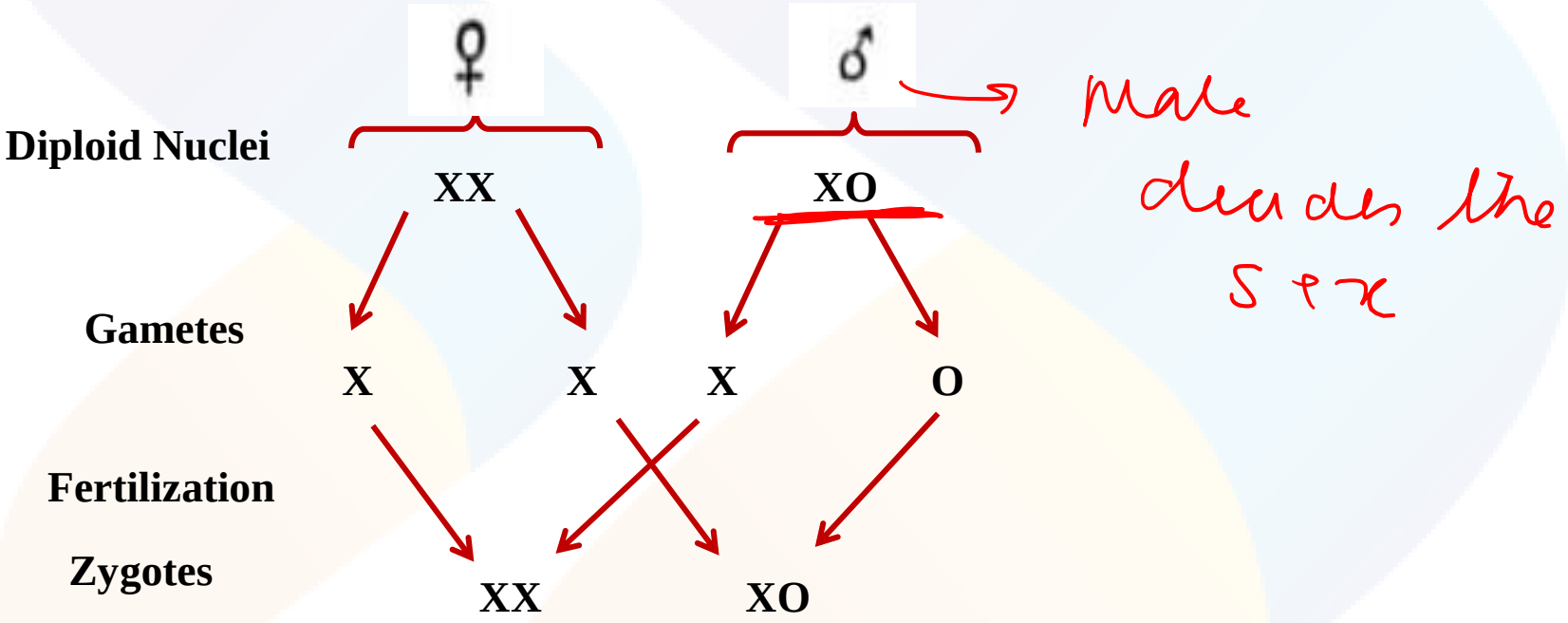


Cockroaches

Bugs

Male sex is determined by the Unpaired X – chromosome.

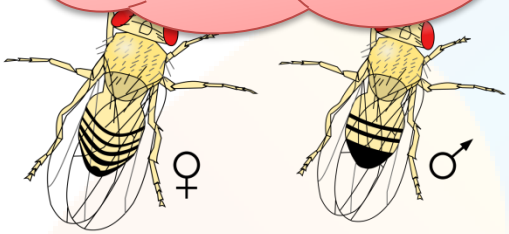
XX – XO TYPE OF HETEROGAMETIC SEX DETERMINATION

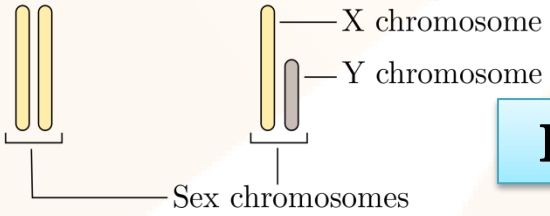


Male decides the Sex

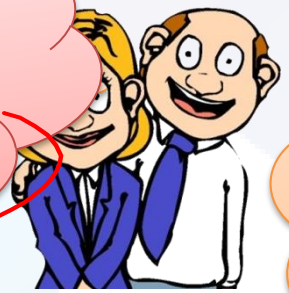
XX - XY type of male heterogametic sex determination

In human beings and some insects such as Drosophila





Drosophila



Females and males produce same number of chromosomes

female is heterogamelw

ZO – ZZ type (Female heterogamety)

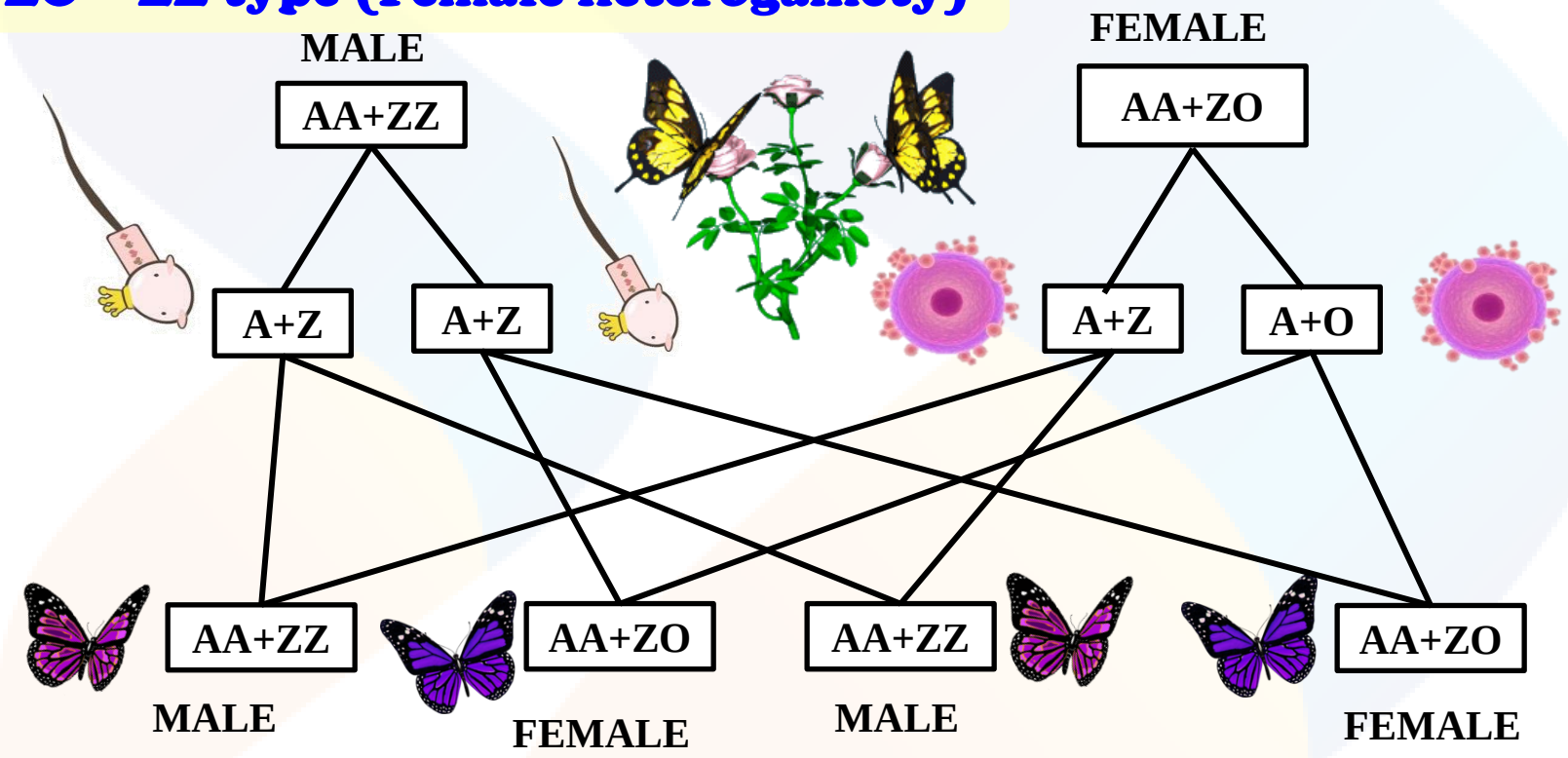
Female Heterogametic

AAZO

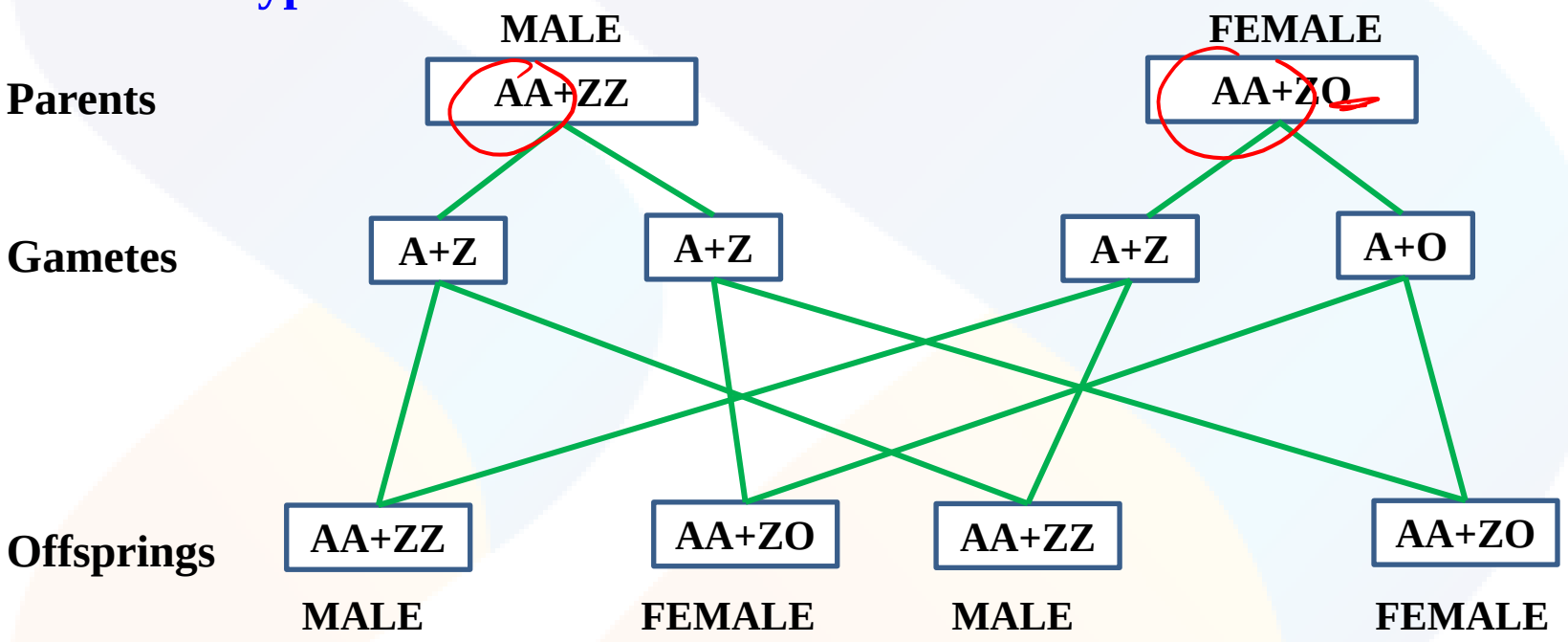
Two types of ova

Females produce ova with Z chromosome and ova without sex chromosome.

ZO – ZZ type (Female heterogamety)



ZO – ZZ type


MALE
FEMALE
Parents
AA+ZZ
AA+ZO
Gametes
A+Z
A+Z
A+Z
A+O
Offsprings
AA+ZZ
AA+ZO
AA+ZZ
AA+ZO
MALE
FEMALE
MALE
FEMALE

+ female heterogamete

ZW – ZZ type(Female heterogamety)

Male Homogametic

AAZZ

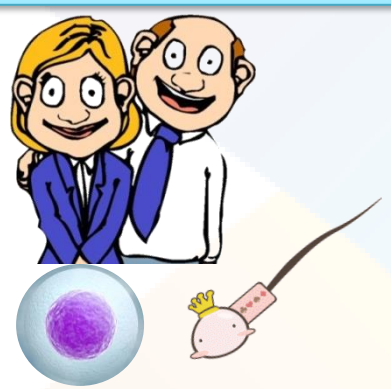
This method is found in birds, reptiles, some fishes.

Similar gametes

Males produce sperms with Z chromosome

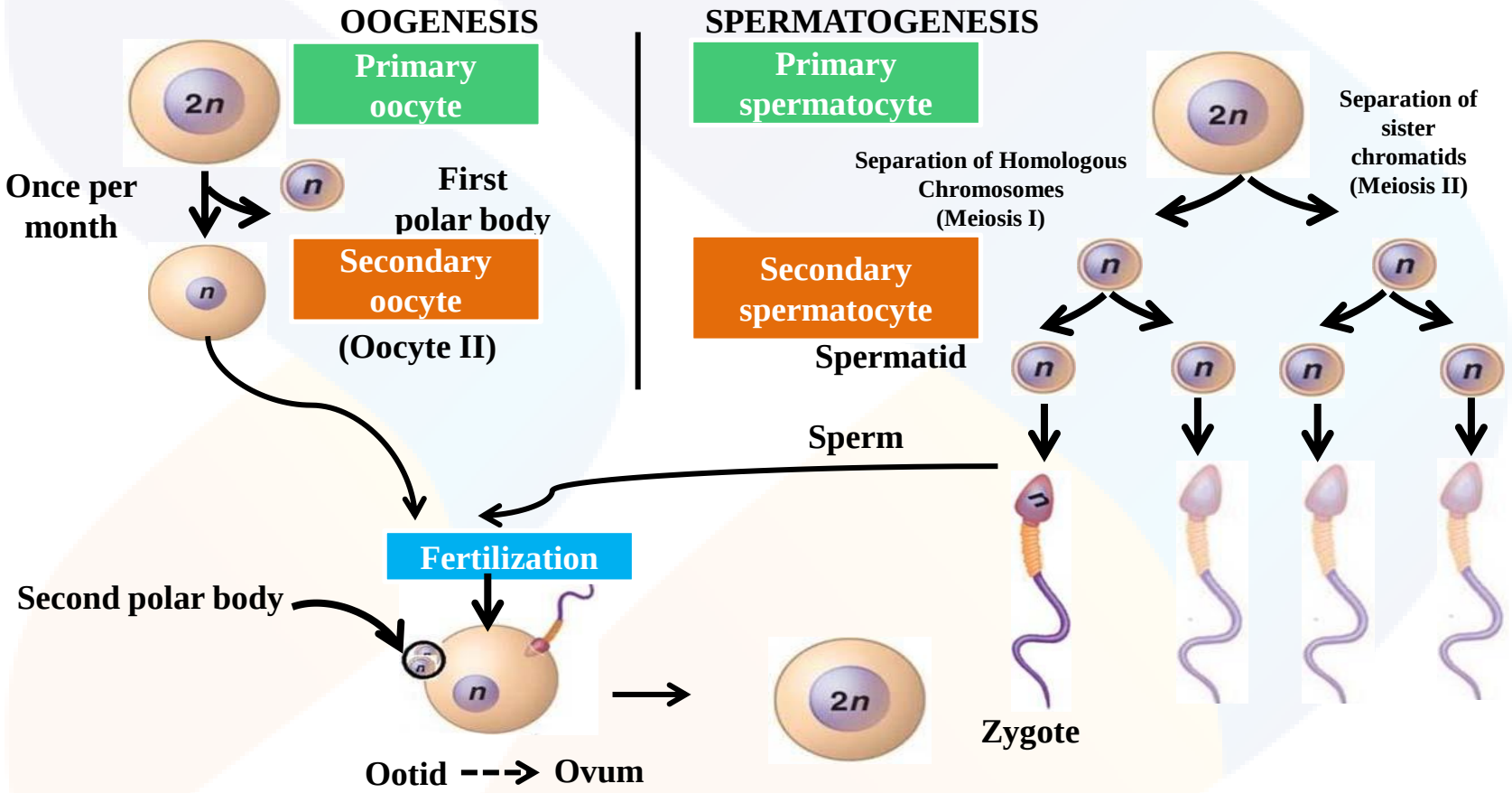
SEX DETERMINATION IN HUMANS

The sex of the child is determined by the genetic make up of sperm.



Because in each pregnancy, there is always 50% probability of either a male or a female child.

It is unfortunate that in our society women are blamed for producing female children and have been ostracised and ill-treated because of this false notion


OOGENESIS
SPERMATOGENESIS
Primary oocyte
Primary spermatocyte
2n
Once per month
n
First polar body
Secondary oocyte
(Oocyte II)
Separation of Homologous Chromosomes (Meiosis I)
Separation of sister chromatids (Meiosis II)
Secondary spermatocyte
Spermatid
Sperm
Fertilization
Second polar body
Ootid
Ovum
Zygote

TYPES OF SEX DTERMINATION

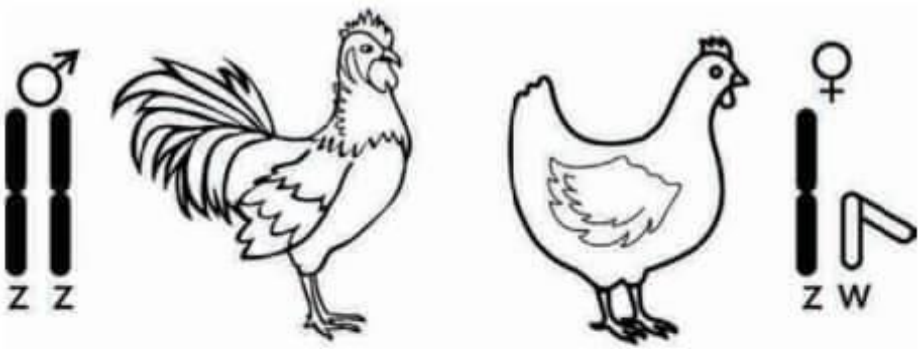
Sex determination in Various Organisms

	Genotype of female	Genotype of male
1. Insects	XX	XO
2. *Drosophila*	XX	XY
3.Birds,fish and butterflies	WZ	WW
4. Humans and other mammals	XX	XY

* female heterogametic

imp for Neet

SEX DTERMINATION IN BIRDS



Male Bird
AA + ZZ

X

Female Bird
AA + ZW

A + Z (sperm) A + Z (sperm)

A + W (ova) A + Z (ova)

	A + Z	A + W
A + Z	AA + ZZ (Male)	AA + ZW (Female)
A + Z	AA + ZZ (Male)	AA + ZW (Female)

ZW type of sex determination as seen in several birds.

→ determines the sex