Principles of Inheritance and Variation

Pea Plant

Pisum sativum

Gregor Mendel

Father of Genetics

Gregor Mendel - The Father of genetics

Gregor Mendel, a great Scientist is known as the Father of Genetics.

He observed a lot of variations in pea plants.

Some pea plants were tall, some were short.

Some seeds were yellow and the others were green.

These variations developed curiosity in the mind of Mendel and motivated him to perform a research on pea plants.

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Mendel observed seven pairs of contrasting characters in pea plants.

He performed cross breeding experiments with pea plants by artificial pollination.

He analyzed the results and proposed the Principles of Inheritance of Characters.

This led to the emergence of a new branch of Science known as Genetics.

B. John Ebenezer

Seven Pairs of Contrasting Characters in Pea Plants

Principles of Inheritance

Genetics deals with the inheritance and variation of characters from parents to offsprings.

Inheritance is the process by which characters are transferred from parent to progeny.

Variation is the difference by which progeny differ from their parents.

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Mendel's Findings

Mendel proposed that something was being stably passed on, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as **'factors'.**

Now a days we call them as **genes.**

Gene is therefore are the **units of inheritance.**

Genes which code for a pair of contrasting traits are known as **alleles,** i.e. they are slightly different forms of the same gene.

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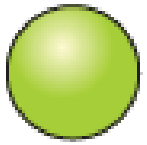
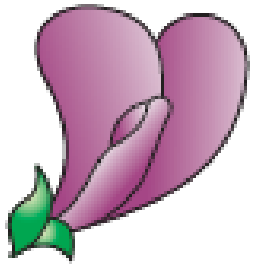
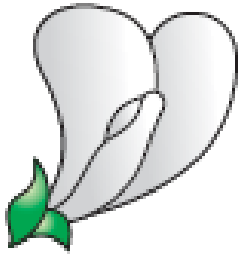
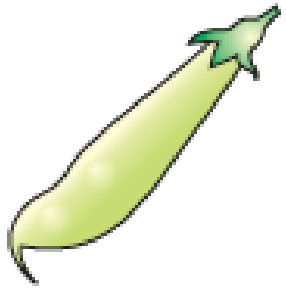
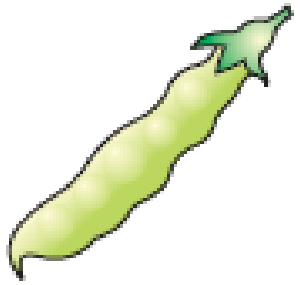
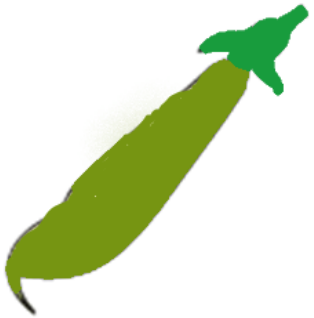
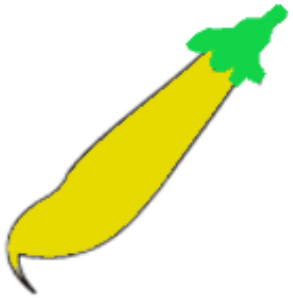
Seven pairs of contrasting characters in pea plant

Character	Dominant	Recessive
Seed shape	Round	Wrinkled
		
Seed Colour	Yellow	Green
		
Flower Colour	Violet	White
		

Seven pairs of contrasting characters in pea plant

Character	Dominant	Recessive
Pod Shape	Full	Constricted
		
Pod Colour	Green	Yellow
		

Seven pairs of contrasting characters in pea plant

Character	Dominant	Recessive
Flower Position	Axial	Terminal
		
Stem Height	Tall	Short
		

Seven pairs of contrasting characters in pea plant

Seed form	Seed color	Pod form	Pod color	Flower position	Seed coat color	Stem length
						
Round (*R*)	Yellow (*Y*)	Inflated (*V*)	Green (*G*)	Axial (*F*) along stem	Gray or gray-brown (*A*)	Tall (*L*)
						
Wrinkled (*r*)	Green (*y*)	Restricted (*v*)	Yellow (*g*)	Terminal (*f*) on top	White (*a*)	Short (*l*)

Pod colour

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Principles of Inheritance

Mendel proposed the three principles of inheritance of characters. They are as follows;

1. Law of Dominance and Recessive.
2. Law of Segregation.
3. Law of Independent Assortment.

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Genes:

A segment of DNA which controls a specific character is called a gene.

Genes are present in chromosomes.

Alleles:

The different forms of a gene are called alleles. ‘Tt’, ‘Rr’, ‘Yy’.

The genotype of Heterozygous Tall character consists of two kinds of genes, capital ‘T’ and small ‘t’ (Tt). These are the alleles.

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Homozygous Genotype:

When both the genes of a particular character are of the same kind, it is called homozygous genotype. Eg. ‘TT’ and ‘tt’.

Heterozygous Genotype:

When both the genes of a particular character are of different kinds, it is called heterozygous genotype. Eg. ‘Tt’, ‘Rr’, and ‘Yy’.

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Phenotypic Ratio:

The ratio obtained based on the visible characters is called Phenotypic Ratio.

Genotypic Ratio:

The ratio obtained based on the genetic combination of alleles is called Genotypic Ratio.

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Each character has more than one forms.

- The character '**Height**' has two forms, one is **tall** and other one is **dwarf**.
- The character **colour of seeds** in pea plant has two forms, one is **yellow**, the other one is **green**.

Each character is controlled by a pair of genes.

- The genes for the character "Yellow" is (YY).
- The genes for the character "Green" is (yy).

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Monohybrid Cross

A cross which involves a single pair of contrasting characters is called a monohybrid cross. For example **Height** of the plants.

Height is a single character. It has two pairs of contrasting characters Tall and Short.

Tall character is controlled by a pair of genes ‘TT’.

Short character is controlled by a pair of genes ‘tt’.

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Dihybrid Cross

A cross which involves two pairs of contrasting characters is called a dihybrid cross. For example **Colour of the seeds and Shape of the seeds** of pea plants.

1. **Colour of the seeds** is a single character. It has a pair of contrasting characters "**Yellow and Green Seeds**".

2. **Shape of the seeds** is a single character. It has a pair of contrasting characters "**Round and Wrinkled Seeds**".

As these two pairs of contrasting characters are considered in a single cross, it is known as a dihybrid cross.

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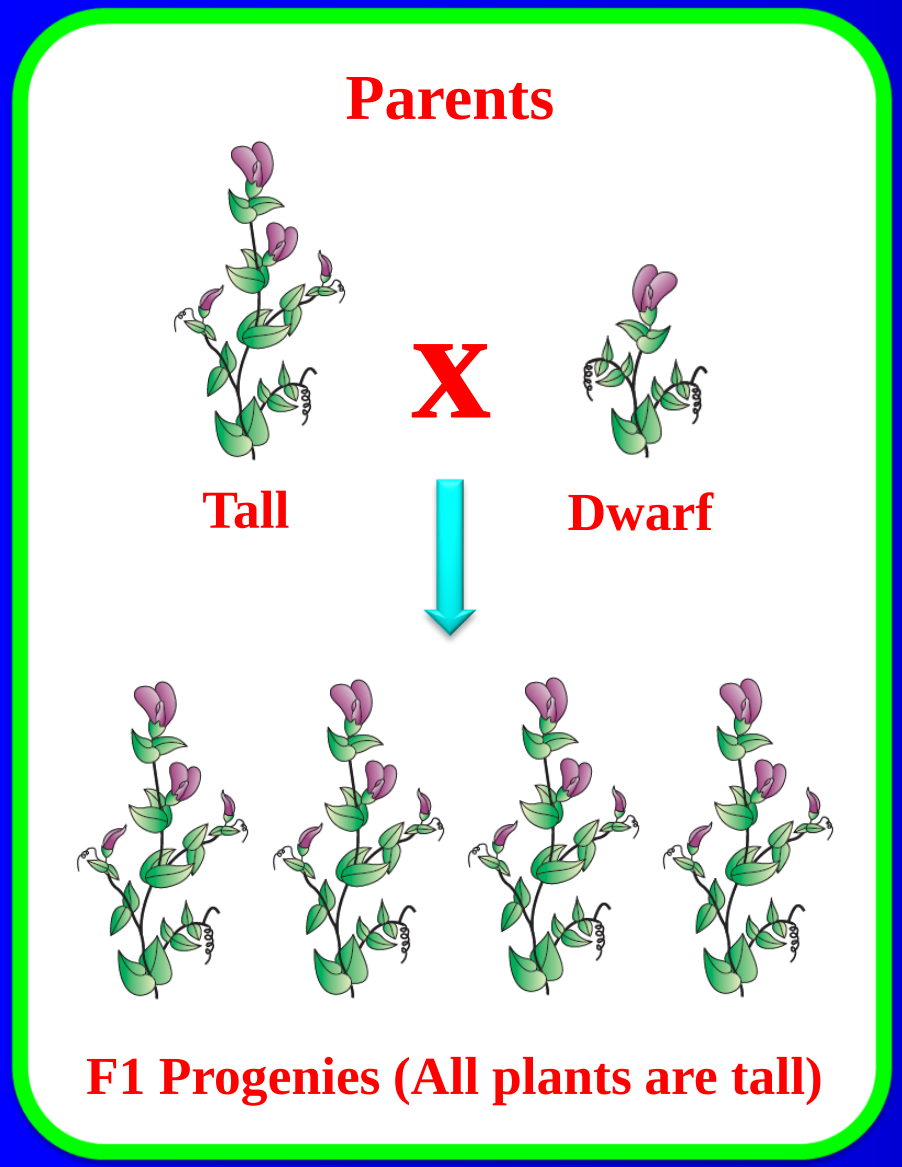
Monohybrid Cross

First Filial Generation - F1



The progenies formed as a result of first cross or parental cross breeding, form the first **filial generation** or F1 generation.

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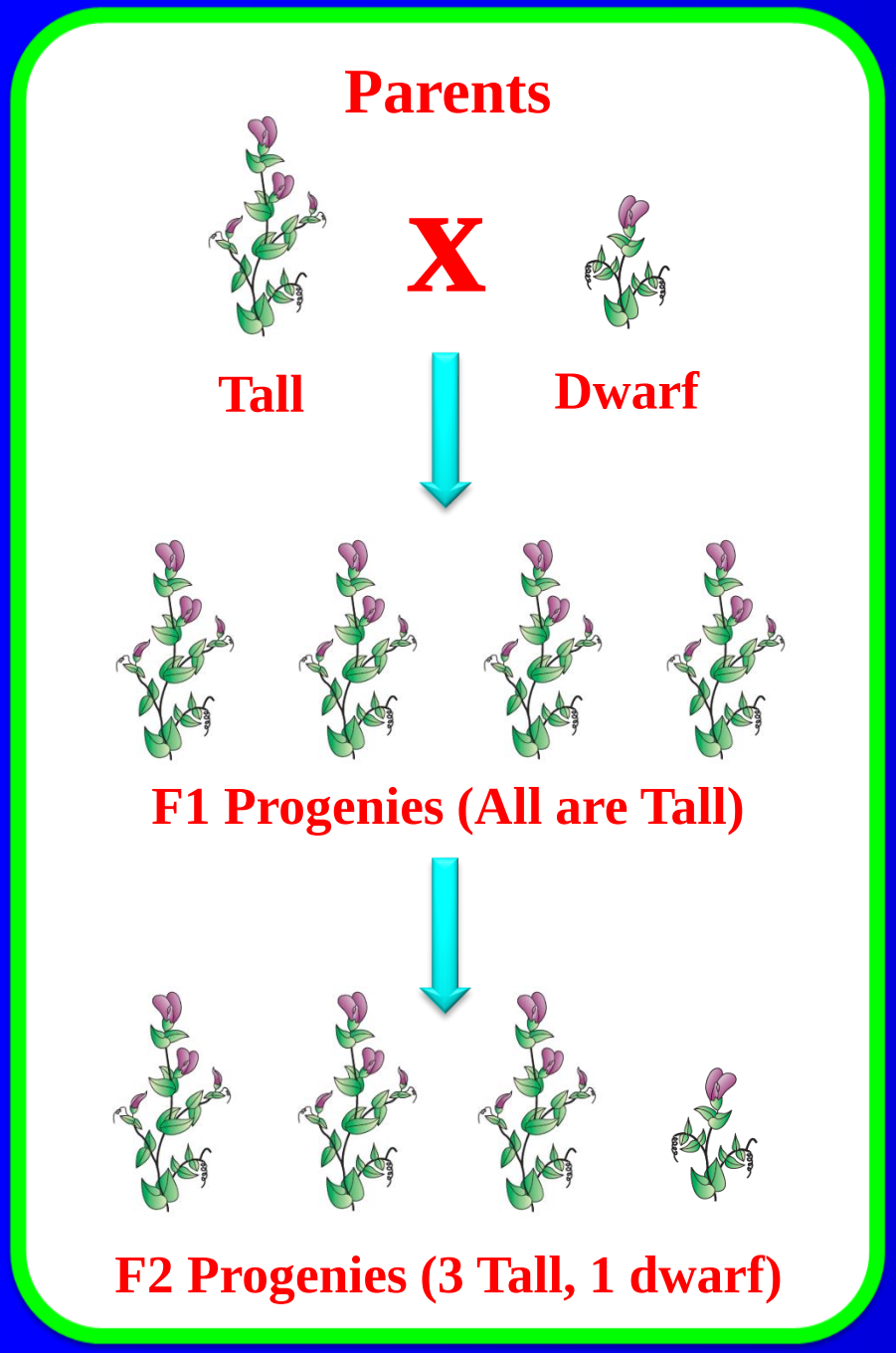
Second Filial Generation - F2



Self Pollination

The progenies formed as a result of self pollination of F1 hybrids form the **Second Filial Generation** or F2 generation.

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Monohybrid Cross

- Mendel selected true bred tall (TT) and dwarf (tt) pea plants.
- Then he crossed these two plants by artificial cross pollination.
- The hybrids produced seeds after fertilization.
- The seeds were sown and they grew into plants.
- These plants represented the first filial generation or F1 generation.
- All the F1 plants were tall.

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Monohybrid Cross

- Then Mendel self-pollinated the F1 plants.
- The F1 hybrids produced seeds after fertilization.
- The seeds were sown and they grew into plants.
- These plants represented the second filial generation or F2 generation.
- He observed that all the plants obtained in the F2 generation were not tall. Instead, one-fourth of the F2 plants were short.
- i.e. Out of four, three plants were tall and one plant was dwarf. (3:1)

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Self-pollination of F1 plants

Based on the experiment, Mendel concluded that the F1 tall plants were not true breeding. i.e. They were not pure tall plants.

They were carrying the traits of both tallness and dwarfness.

They appeared tall because the tall trait is dominant over the dwarf trait.

Dominant Character

The character which appears in F1 is called dominant character.

Recessive Character

The character which is not expressed in F1 is called recessive character.

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Punnett Square

Punnett square is a square diagram that is used to predict the genotypes of a particular cross breeding. It is named after the Scientist Reginald C. Punnett, who devised this approach.

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Monohybrid Cross : First Filial Generation - F1

	Short t	t
Tall T	Tt Tall	Tt Tall
T	Tt Tall	Tt Tall

First Filial Generation - F1

In F1, the progenies consists of one dominant gene, capital ‘T’ and one recessive gene small ‘t’.

The gene capital ‘T’ controls the tall character.

The gene small ‘t’ controls the short character.

Even though gene for the tall character ‘T’ and the gene for the short character ‘t’ are present in the F1 progenies, they appear as tall plants.

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All the progenies of F1 are heterozygously tall plants

	Short t	t
Tall T	Tt Tall	Tt Tall
T	Tt Tall	Tt Tall

First Filial Generation - F1

This happens because the gene capital ‘T’ is a dominant gene and the gene small ‘t’ is a recessive gene.

Dominant gene ‘T’ suppresses the character of the recessive gene ‘t’.

So, only dominant character is expressed and the recessive character is suppressed or hidden.

Hence all the progenies of F1 are heterozygously tall plants.

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Monohybrid Cross : Second Filial Generation - F2

	Tall T	t
Tall T	TT Tall	Tt Tall
t	Tt Tall	tt Short

Second Filial Generation-F2

In F2, three plants are tall and one plant is dwarf. So the phenotypic ratio is 3:1.

But one plant is homozygously tall, having the genotype ‘TT’.

Two plants are heterozygously tall, having the genotype ‘Tt’.

One plant is homozygously short, having the genotype ‘tt’.

Hence the genotypic ratio of F2 is 1:2:1.

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Monohybrid Cross : Second Filial Generation - F2

	Tall T	t
Tall T	TT Tall	Tt Tall
t	Tt Tall	tt Short

Second Filial Generation-F2

The F2 Phenotypic ratio is 3 : 1

The F2 Genotypic ratio is 1 : 2 : 1

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Test Cross

Test Cross

A cross which is done to **determine the genotype** (whether homozygous or heterozygous) of a **dominant individual** is called a test cross.

This is achieved by crossing the individual with a **double recessive parent.**

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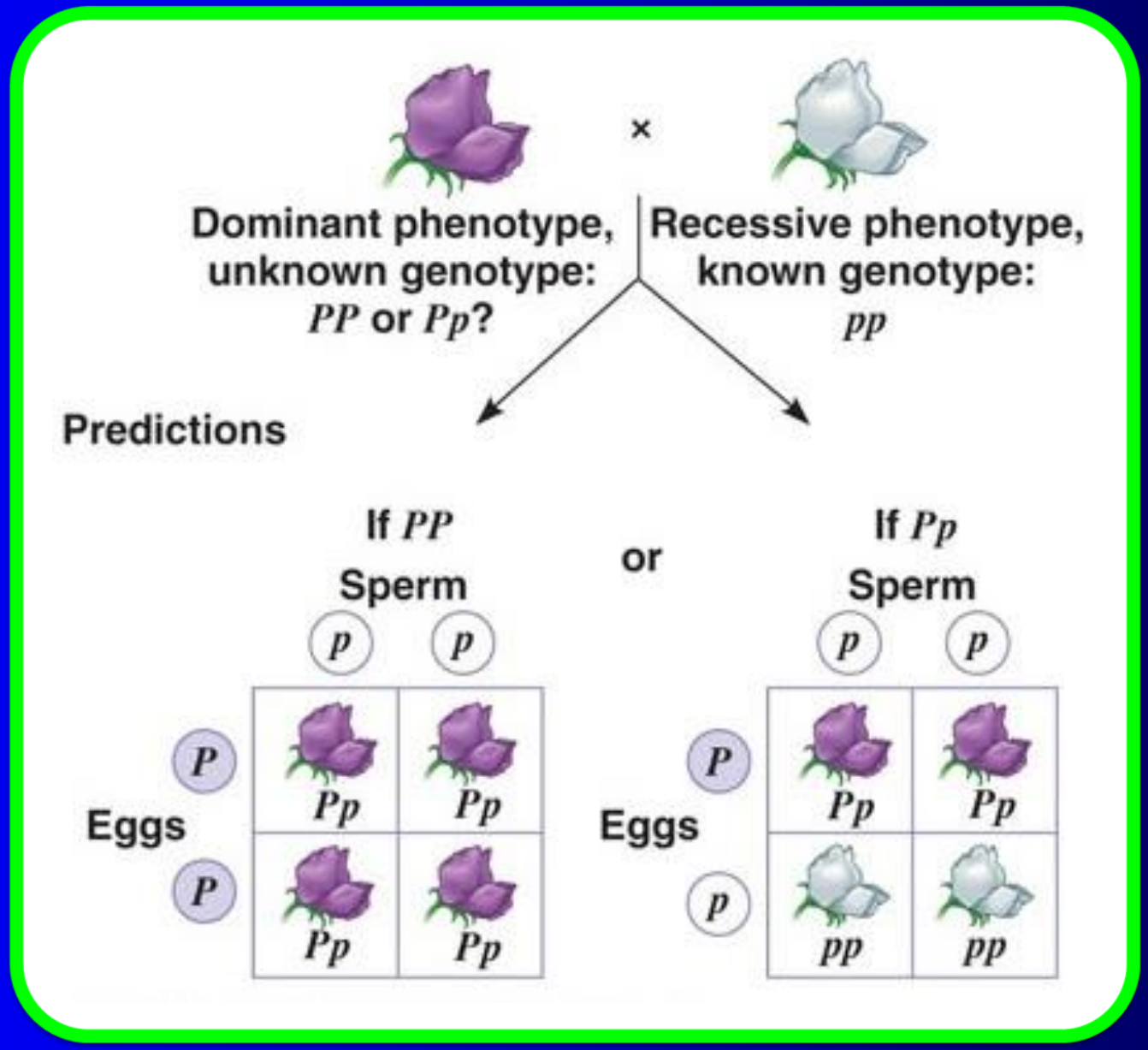
Test Cross



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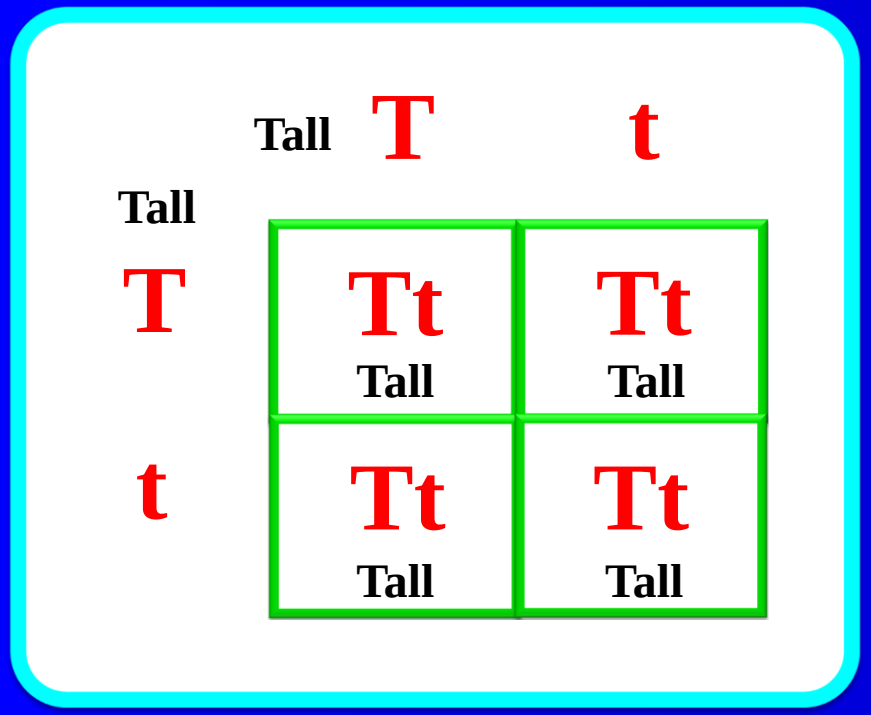
Law of Segregation

Law of Seggregation – Law of purity of gametes



First Filial Generation - F1

Heterozygous parents produce two kinds of gametes each having one allele with equal proportion

The members of allelic pair that **remained together** in the parent, segregate/separate during gamete formation and only one of the alleles enters a gamete.

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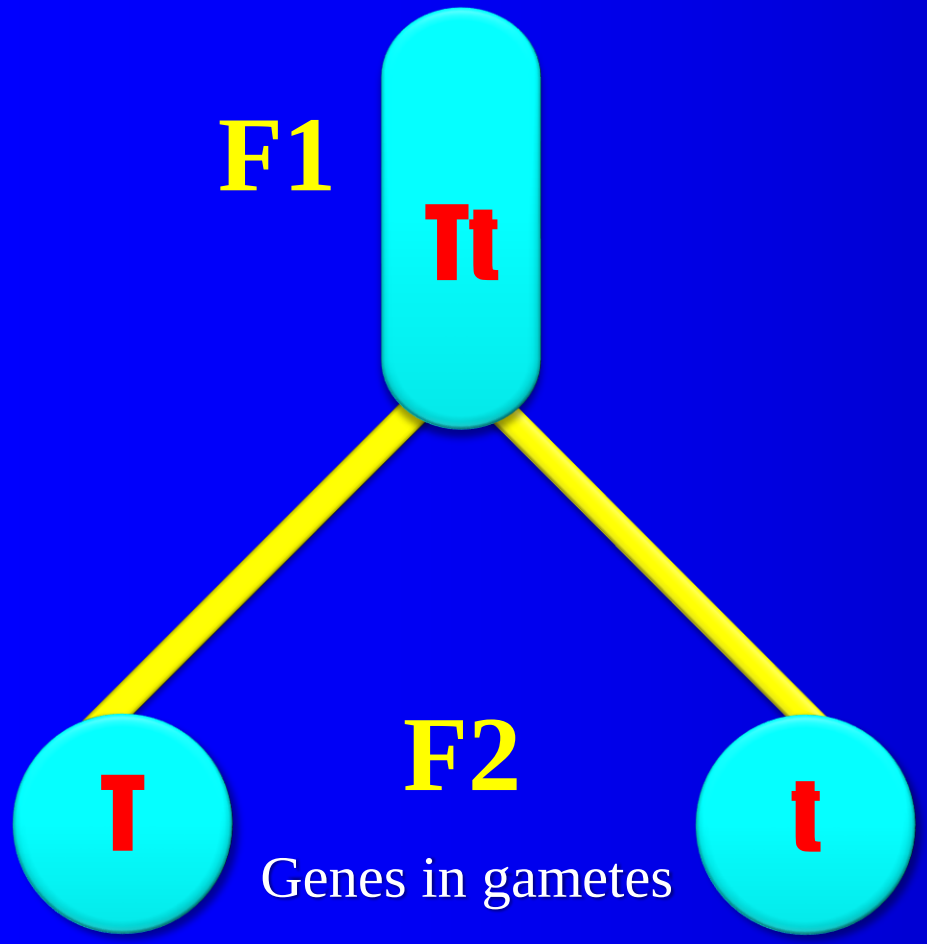
Genes located in the same locus of a chromosome



Second Filial Generation-F2

Law of Segregation:

A pair of genes present in the same locus of a chromosome segregate and pass on to different gametes.

This allows the recessive trait to appear in the F2.

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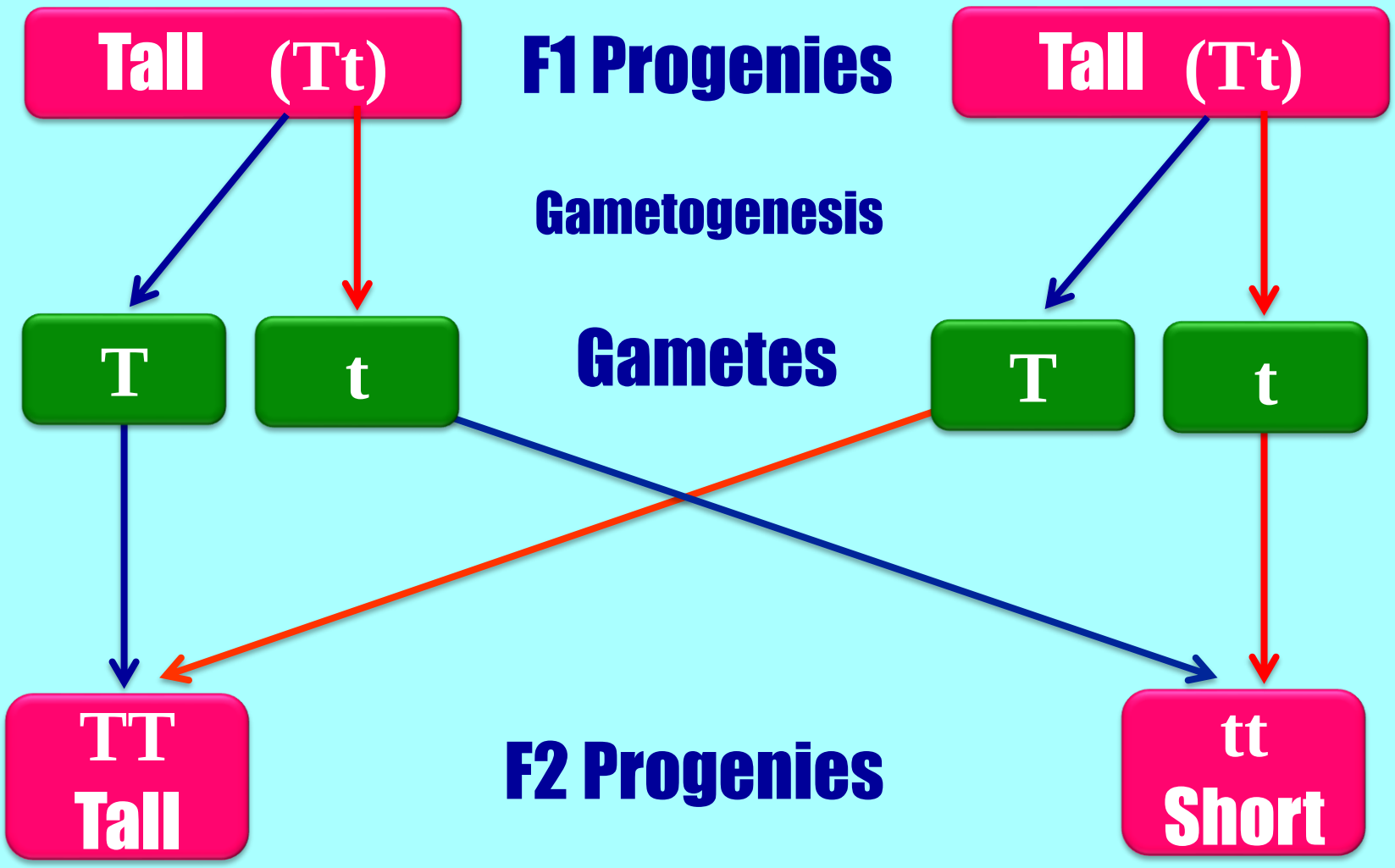
Law of Segregation



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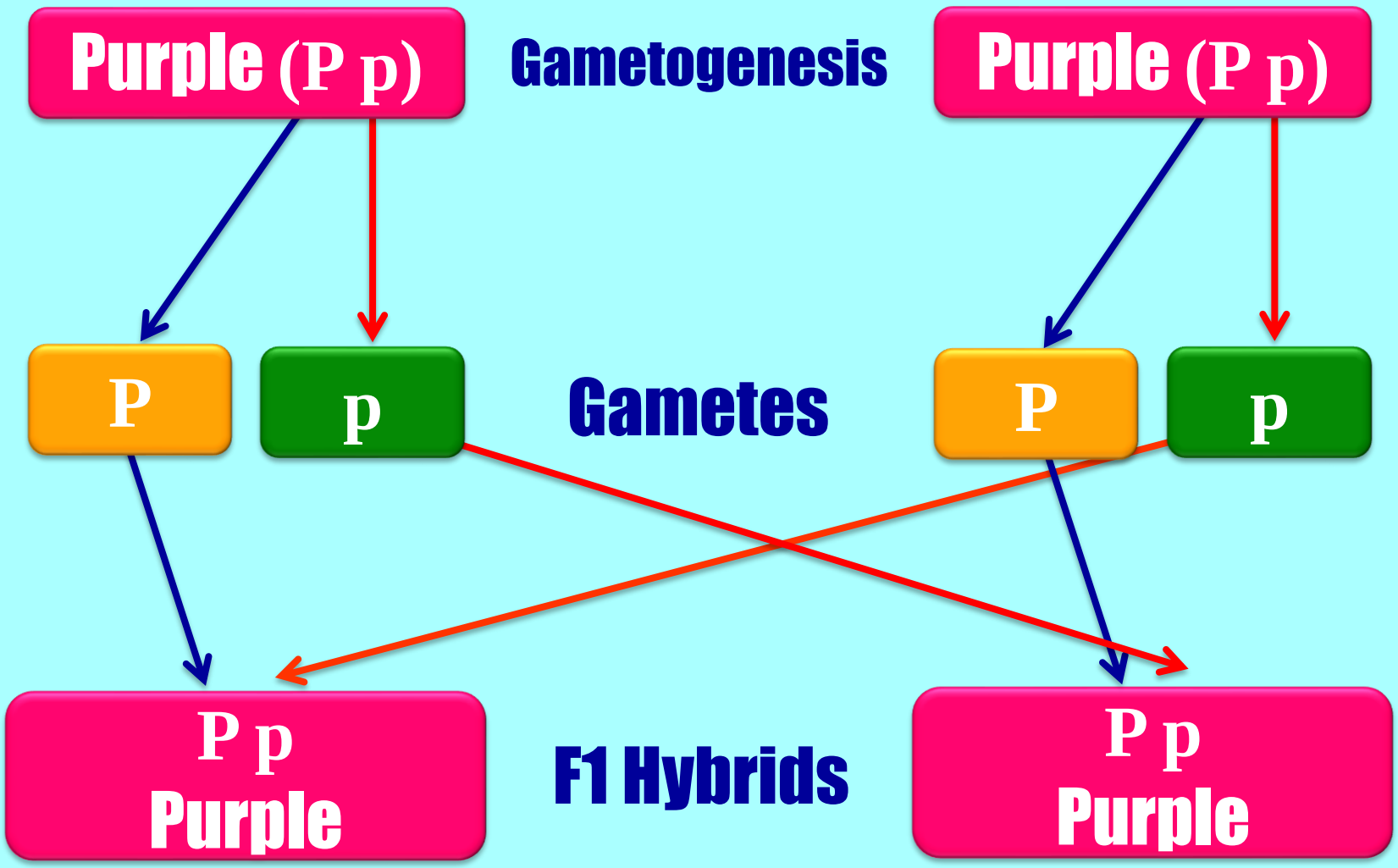
Law of Segregation



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Law of Independent Assortment

Law of Independent Assortment

Based on the observations of **dihybrid cross,** Mendel proposed the **Law of Independent Assortment**.

This law states that ‘when two pairs of contrasting characters are combined in a hybrid, segregation of **one pair of characters**, is independent of the other pair of characters.

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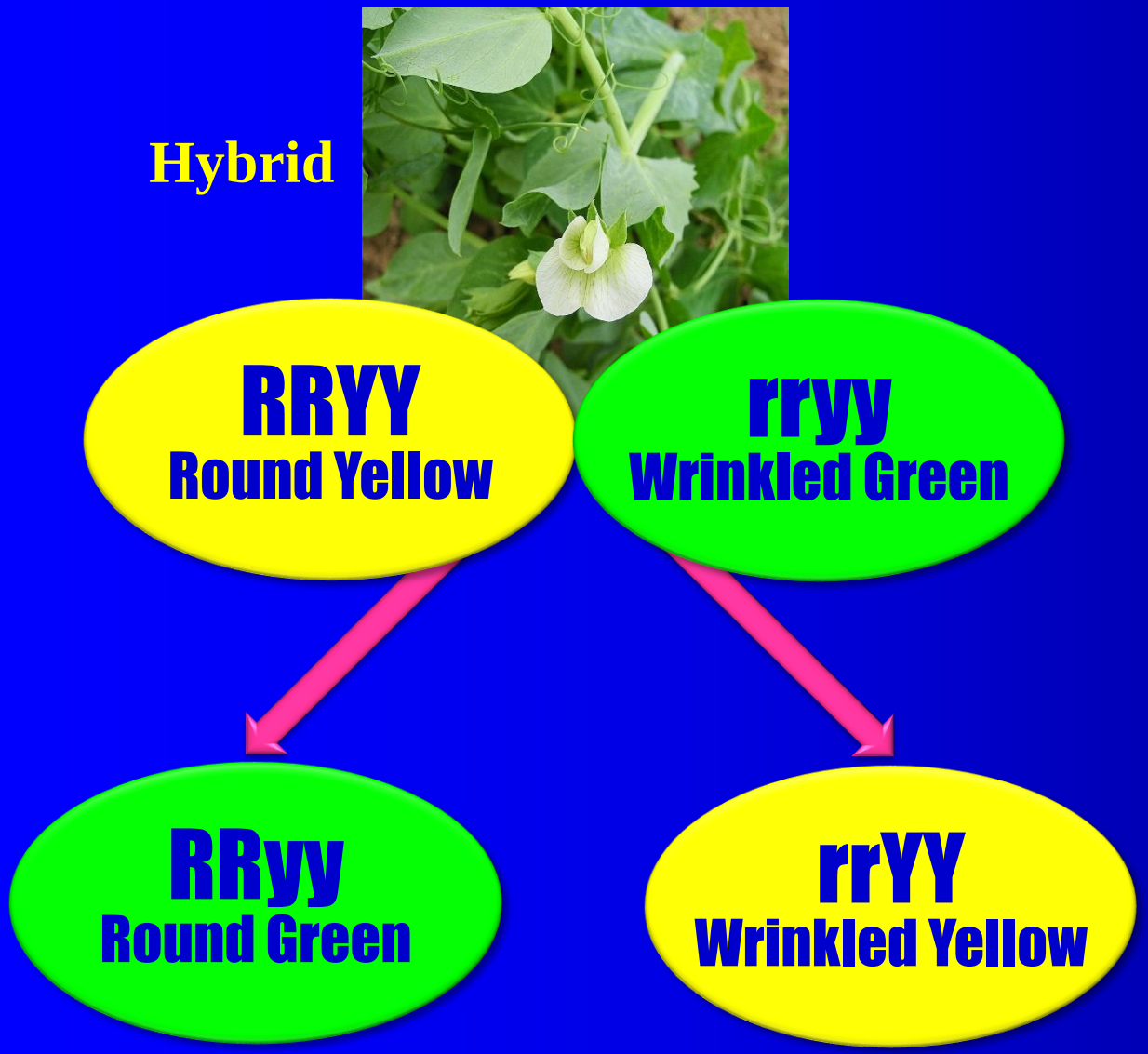
Law of Independent Assortment



When two pairs of contrasting characters are combined

(Round Yellow (RRYY)

and

Wrinkled green (rryy) in a hybrid,

segregation of **one pair of characters**, is independent of the other pair of characters.

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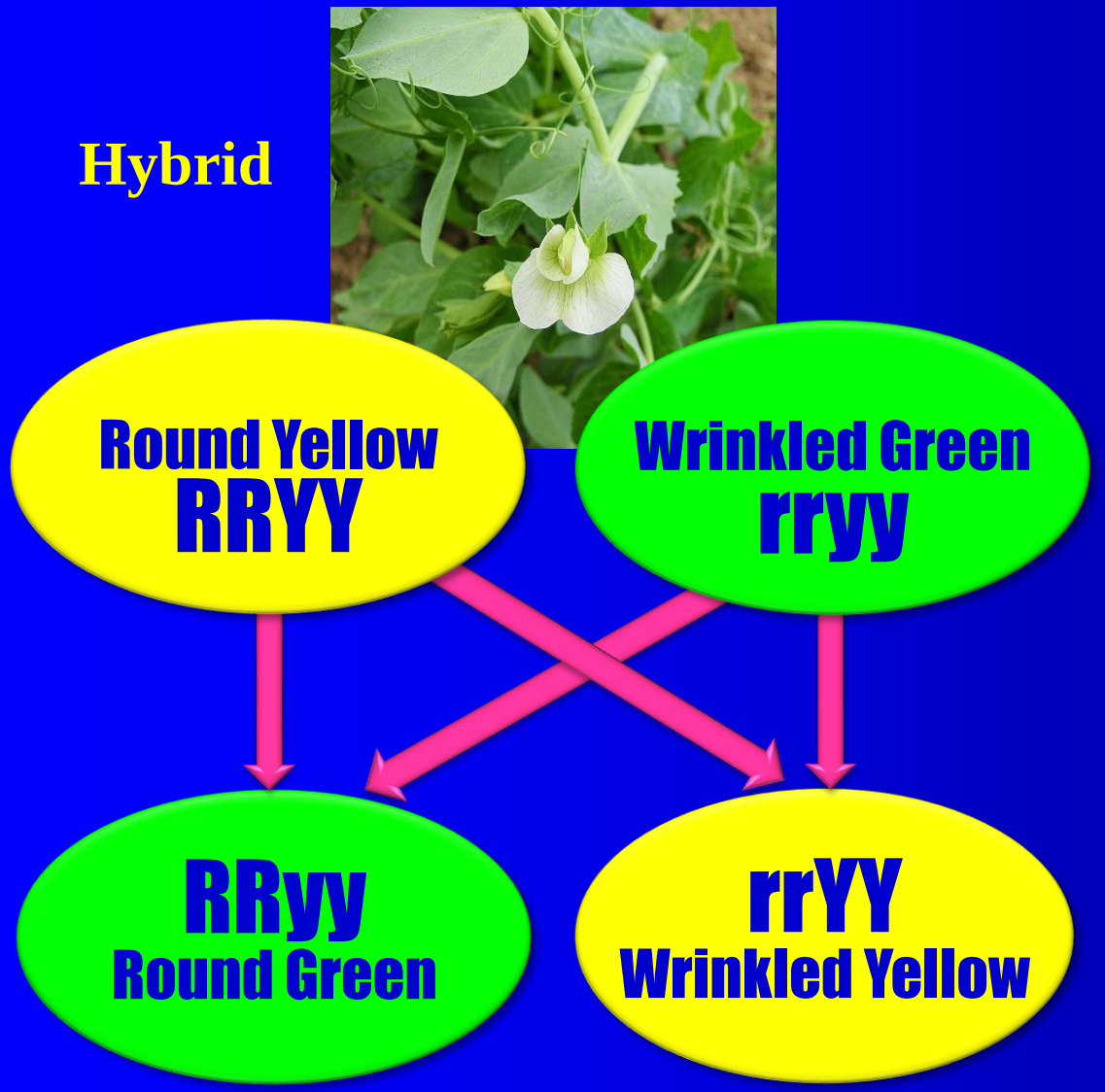
Law of Independent Assortment



When two pairs of contrasting characters are combined

(Round Yellow (RRYY)

and

Wrinkled green (rryy) in a hybrid,

segregation of **one pair of characters**, is independent of the other pair of characters.

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Dihybrid Cross between Pea Plants bearing Round Yellow Seeds and Wrinkled Green Seeds

Dihybrid Cross F1

Round Yellow (RRYY) X **Wrinkled Green (rryy)**

Gametogenesis

RY RY

Gametes

ry ry

RrYy
Round Yellow

F1 Hybrids

RrYy
Round Yellow

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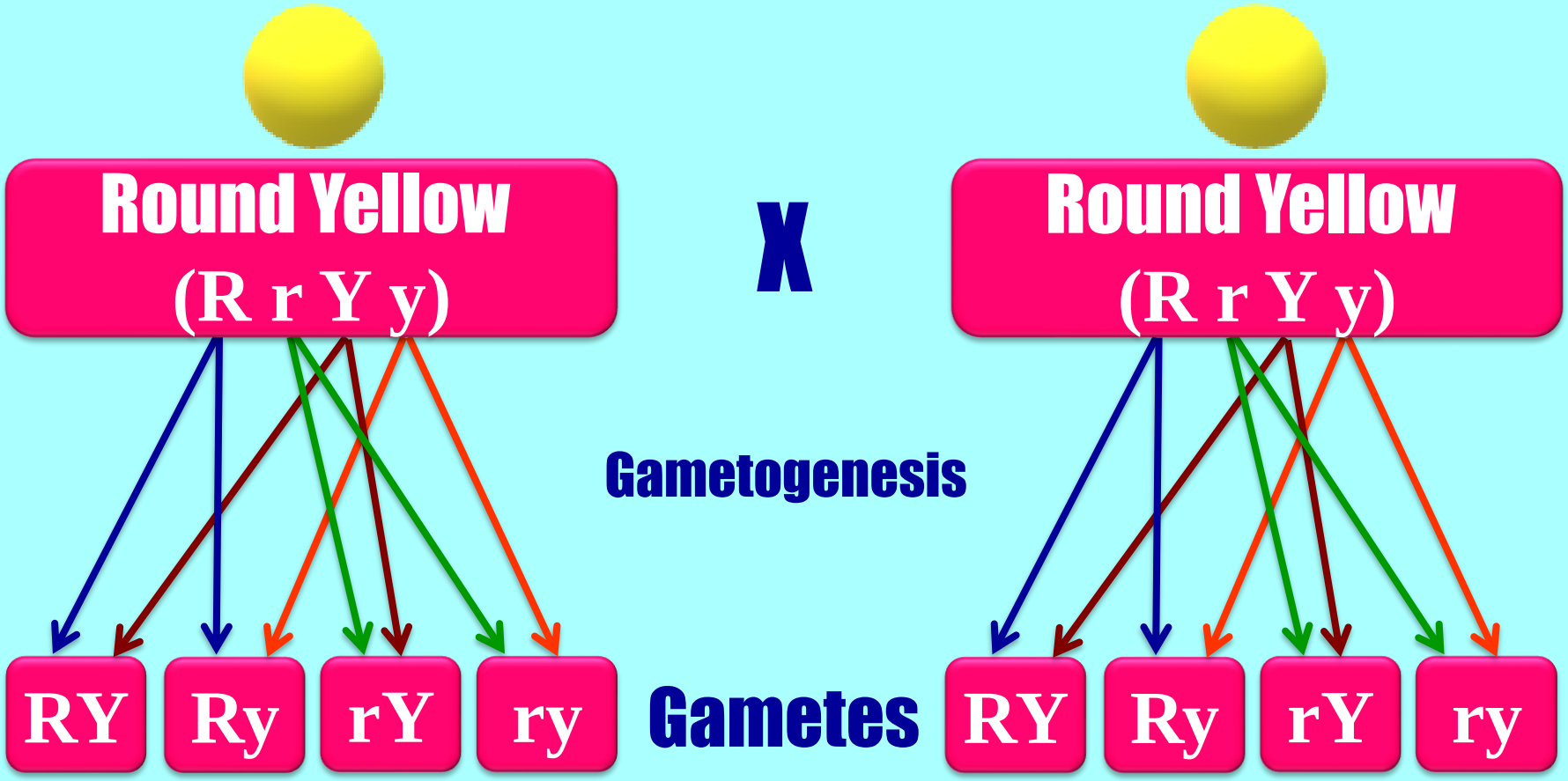
Selfing of F1 Hybrids and Gametogenesis



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The traits of F2 Hybrids of Dihybrid Cross

	RY	Ry	rY	ry
RY	RRYY Round Yellow	RRYy Round Yellow	RrYY Round Yellow	RrYy Round Yellow
Ry	RRYy Round Yellow	RRyy Round Green	RrYy Round Yellow	Rryy Round Green
rY	RrYY Round Yellow	RrYy Round Yellow	rrYY Wrinkled Yellow	rrYy Wrinkled Yellow
ry	RrYy Round Yellow	Rryy Round Green	rrYy Wrinkled Yellow	rryy Wrinkled Green

The traits of F2 hybrids were as follows.

9 **Round yellow seeds**

3 **Round green seeds**

3 **Wrinkled yellow seeds**

1 **Wrinkled green seed**

Hence, the F2 Dihybrid ratio is 9:3:3:1.

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	RY	Ry	rY	ry
RY	RRYY  Round Yellow	RRYy  Round Yellow	RrYY  Round Yellow	RrYy  Round Yellow
Ry	RRYy  Round Yellow	RRyy  Round Green	RrYy  Round Yellow	Rryy  Round Green
rY	RrYY  Round Yellow	RrYy  Round Yellow	rrYY Wrinkled Yellow	rrYy Wrinkled Yellow
ry	RrYy  Round Yellow	Rryy  Round Green	rrYy Wrinkled Yellow	rryy  Wrinkled Green

The F2 Dihybrid Ratio is 9:3:3:1

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Why Mendel's Theory was Unrecognised?

Firstly communication was not easy in those days and his work could not be widely publicized.

Secondly his concept of genes (or factors, in Mendel's word) as stable and discrete (distinct) units that controlled the traits and of the pair of alleles which did not' blend' with each other, was not accepted by his contemporary scientists as an explanation for the apparently continuous variation seen in nature.

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Why Mendel's Theory was Unrecognised?

Thirdly Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.

Finally he could not provide any physical proof for the existence of factors.

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Rediscovery of Mendel's Result

1990 three scientists **(deVries, Correns and von Tschermak)** independently rediscovered Mendel's result on the inheritance of character

Incomplete Dominance

Incomplete Dominance

When a cross between two pure breed is done for one contrasting character, the F1 hybrid phenotype does not resemble either of the two parents and was in between the two, called incomplete dominance.

Inheritance of flower color in the dog flower (snapdragon or Antirrhinum sp.) is a good example of incomplete dominance.

The F2 phenotypic ratio is 1:2:1 instead of 3:1 as Mendelian monohybrid cross.

Genotypic ratio of F2 generation is 1:2:1

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Snapdragon

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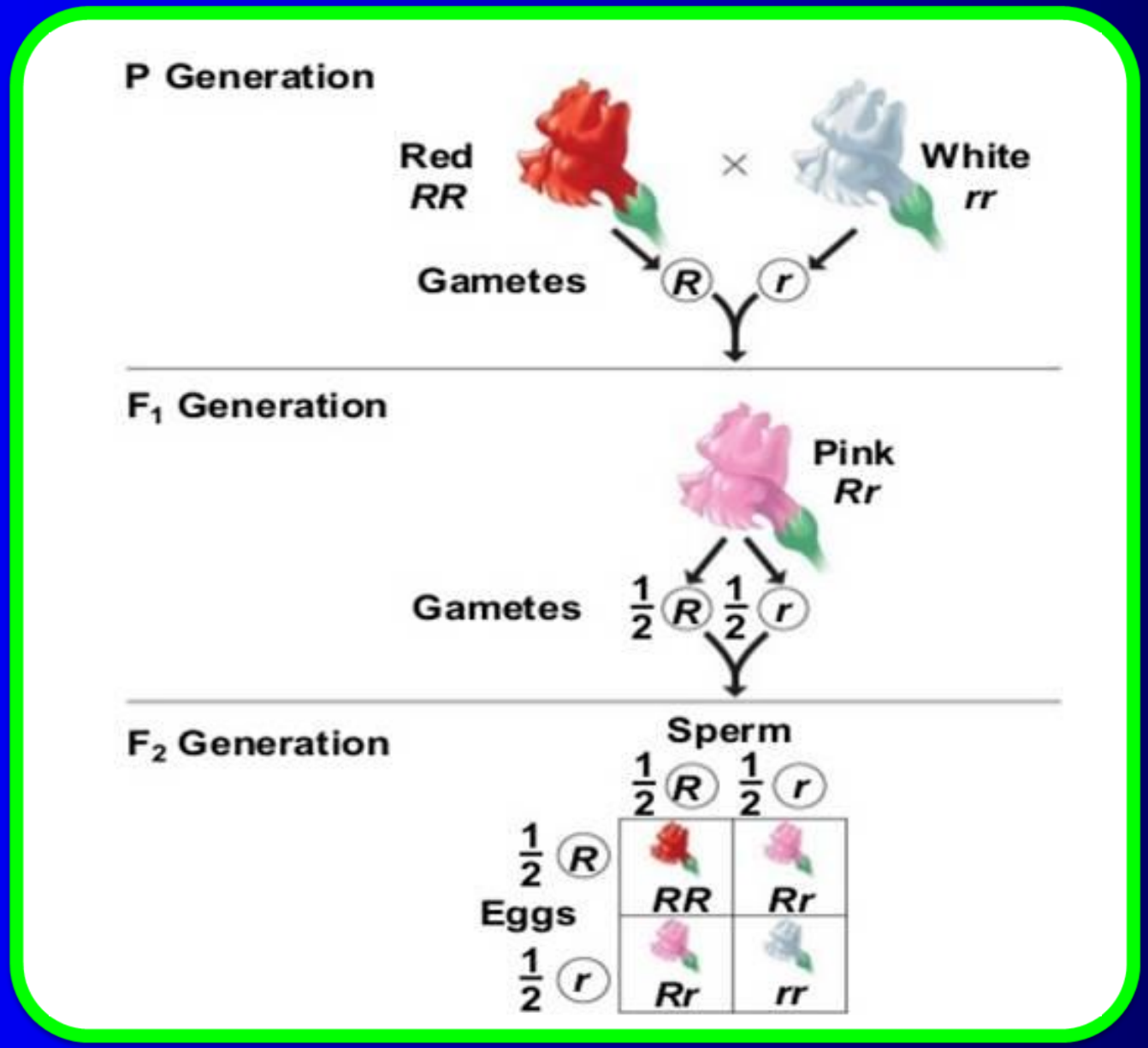
Incomplete Dominance



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Polygenic Inheritance

Polygenic Inheritance

The inheritance of a trait which is controlled by **three or more genes** is called polygenic inheritance.

Example: **Height, skin colour and weight** in human beings.

Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment.

In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive.

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Polygenic Inheritance

To understand this better let us assume that three genes A, B, C control skin colour in human.

The dominant genes **A, B and C** are responsible for **dark skin colour**

The recessive genes **a, b and c** are responsible for **light skin colour.**

The dominant alleles **(AABBCC)** will have the **darkest skin colour.**

The recessive alleles **(aabbcc)** will have the **lightest skin colour.**

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Polygenic Inheritance

As expected the genotype with three dominant alleles and three recessive alleles will have an intermediate skin colour.

In this manner the number of each type of alleles in the genotype would determine the **darkness or lightness of the skin** in an individual.

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Codominance

Codominance

Codominance is a pattern of inheritance in which both the alleles are expressed.

In co-dominance the F1 generation resembles both the parents.

A good example is ABO blood grouping in human beings.

ABO blood groups are controlled by the gene ***I***.

Each person has any **two of the three alleles** of ***I*** gene (IA, IB and i).

The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene.

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Codominance

The alleles ***IA*** and ***IB*** produce a slightly different form of sugar

The allele *i* doesn't produce any sugar.

IA and ***IB*** are completely dominant over ***i***.

When ***IA*** and *i* are present only ***IA*** expresses, because *i* does not produce any sugar.

When ***IB*** and *i* are present ***IB*** expresses.

The *I* designation stands for **isoagglutinogen**, another term for antigen.

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Codominance

But when ***IA*** and ***IB*** are present together they both express their own types of sugars due to co-dominance.

Hence red blood cells have both A and B types of sugars.

Since there are three different alleles, there are six different combinations of these three alleles there are six different genotypes of the human ABO blood types.

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Codominance

The blood group is determined by the presence or absence of one or both the glycoproteins.

Group A has glycoprotein A.

Group B has glycoprotein B.

Group AB has glycoproteins A and B.

Group O has none of the glycoproteins.

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Codominance

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	$i\,i$	O

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Blood Grouping in Human

Antigen in RBC	A	B	AB	O
Antibody in Plasma	Y B	Y A	Y No Antibody	Y AB

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Blood Grouping in Human

Blood Groups	A	B	AB	O
Genotypes	IA IA or IA i	IB IB or IB i	IA IB	i i

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Blood Grouping in Human

Rhesus Factor	Rh	
Blood Type	Positive (+)	Negative (-)

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Pleiotropy

Pleiotropy - Phenylketonuria

A single gene which exhibits multiple phenotypic expression is called **pleiotropic gene.**

Phenylketonuria, a disease in human is an example of pleiotropy.

It is an **autosomal recessive genetic disorder.**

It is caused due to **mutation** in the gene present on chromosome 12 that codes for the enzyme **phenyl alanine hydroxylase.**

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Pleiotropy - Phenylketonuria

A defect in a single gene that codes for the enzyme phenylalanine hydroxylase results in the multiple phenotypes associated with PKU, including mental retardation, eczema, and pigment defects that make affected individuals lighter skinned

Phenotypic Expression:

Mental retardation.

Reduction in hairs.

Reduction in skin pigmentation.

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Pleiotropy

A single gene that controls multiple phenotypic characters is called **pleiotropic gene.**

Starch synthesis in pea seeds is controlled by a single gene.

It has two alleles **B** and **b.**

Starch is synthesized effectively by **BB** dominant genotype and therefore, **large starch grains** are produced.

The **'bb'** recessive genotype has less efficiency hence produce **smaller starch grains**.

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Pleiotropy

A single gene that controls multiple phenotypic characters is called **pleiotropic gene.**

Starch synthesis in pea seeds is controlled by a single gene.

Dominant genotype **BB** produces **large starch grains**.

Recessive genotype **'bb'** produces **smaller starch grains**.

Heterozygous genotype **(Bb)** produces **intermediate starch grains**.

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Pleiotropy

After maturation,

Seeds with dominant genotype **BB** are **round.**

Seeds with recessive genotype **bb** are **wrinkle.**

Seeds with Heterozygous genotype **(Bb)** are round seed.

So, B seems to be dominant allele over b, but the starch grains produced are of intermediate size.

If starch grain size is considered as the phenotype, the alleles show incomplete dominance.

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Chromosomal Theory of Inheritance

Chromosomal Theory of Inheritance

Proposed by **Walter Sutton** and **Theodore Bovery** in 1902.

They studied the chromosome movement during meiosis.

The behavior of chromosomes was parallel to the behavior of genes and used chromosome movement to explain Mendel's laws.

Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the **chromosomal theory of inheritance.**

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Chromosomal Theory of Inheritance

Chromosome and genes are present in pairs in diploid cells.

Homologous chromosomes separate during gamete formation (meiosis)

Fertilization restores the chromosome number to diploid condition.

The chromosomal theory of inheritance claims that, it is the chromosomes that segregate and assort independently.

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Experimental Verification of Chromosomal Theory

Experimental verification of chromosomal theory of inheritance by **Thomas Hunt Morgan** and his colleagues.

Morgan worked with tiny fruit flies ***Drosophila melanogaster.***

Why did Morgan select Drosophila for his study?

Drosophila are **suitable for genetic studies.**

They can be grown on **simple synthetic medium** in the laboratory.

They complete their life cycle in **about two weeks.**

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Experimental Verification of Chromosomal Theory

A single mating could produce **a large number of progeny** flies.

They have **clear differentiation of male and female flies.**

They have **a lot of hereditary variations** which can be seen with low power microscopes.

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Linkage and Recombination

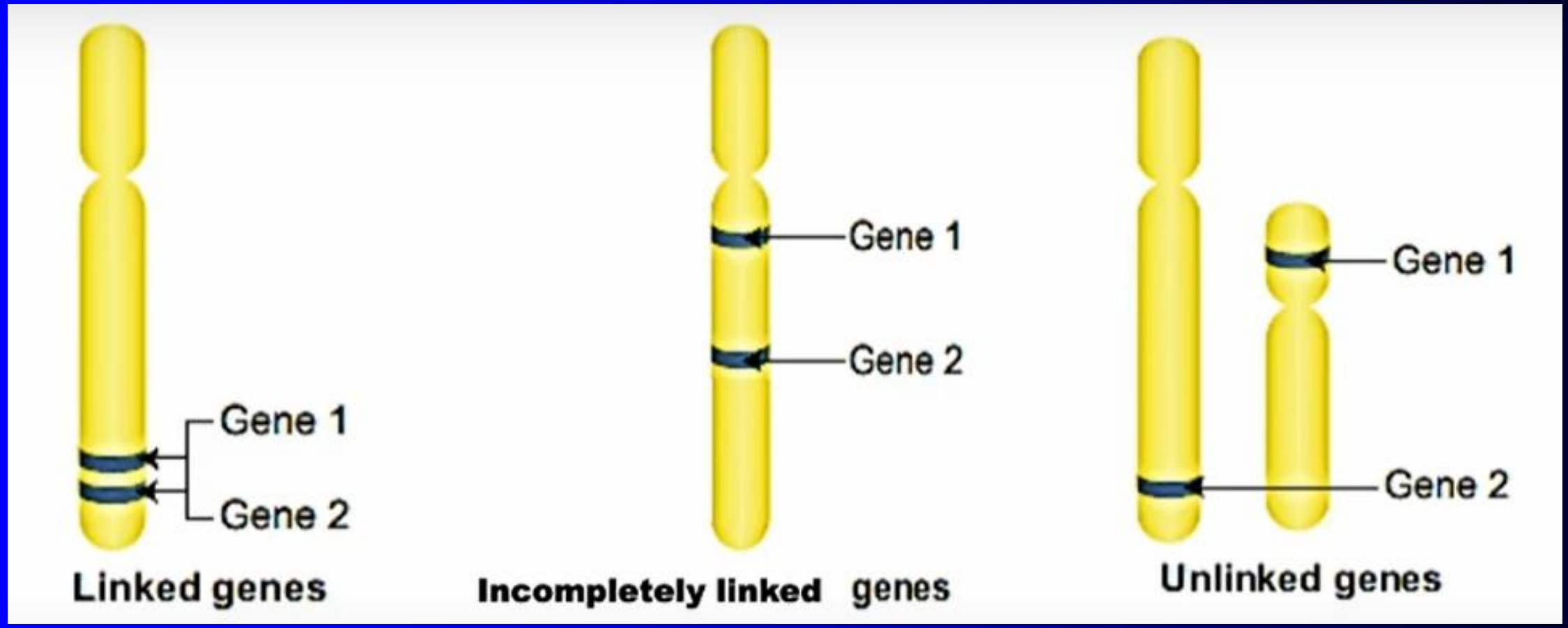
Morgan hybridized **yellow bodied**, **white eyed** females to **brown-bodied**, **red eyed** male and intercrossed their F1 progeny.

He observed that the two genes did not segregate independently of each other and the F2 ratio deviated very significantly from 9:3:3:1 ratio (expected when the two genes are independent).

When two genes in a dihybrid cross, situated on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type.

B. John Ebenezer

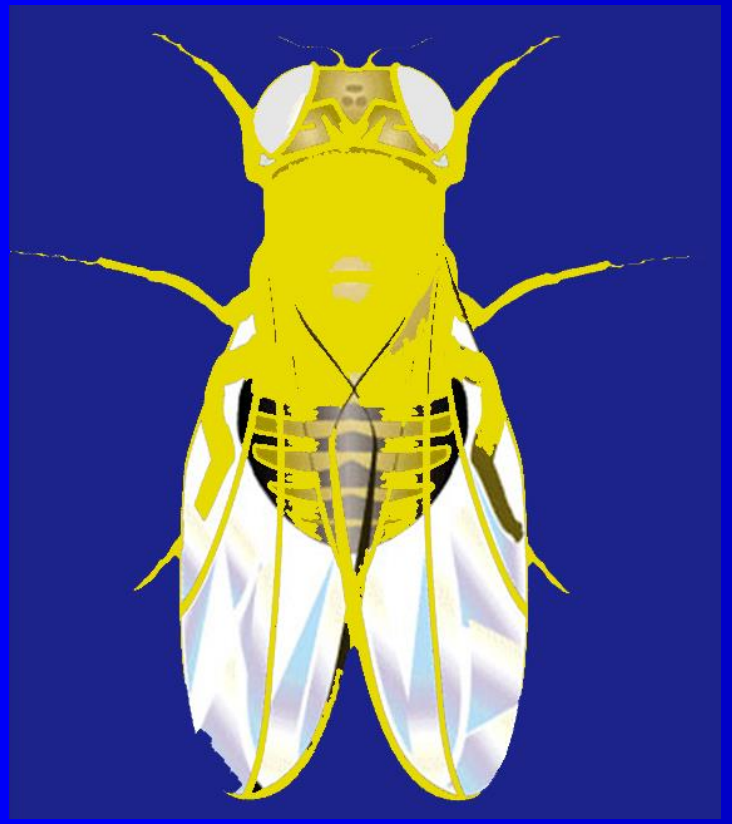
Linkage and Recombination in Drosophila



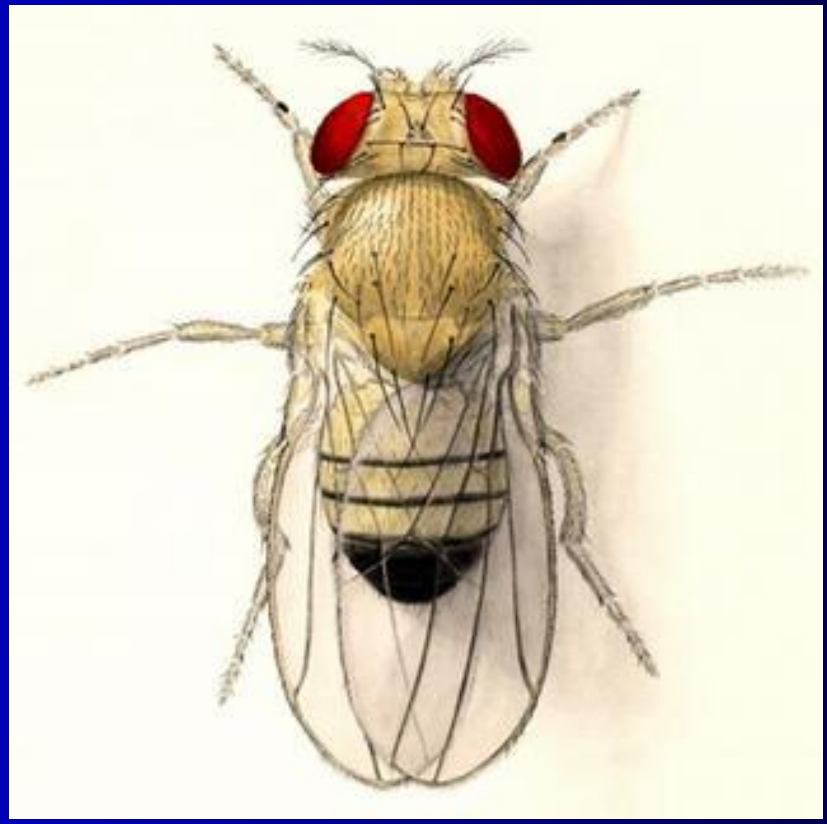
B. John Ebenezer

Linkage and Recombination in Drosophila

Yellow Body
White Eye



Brown Body
Red Eye



B. John Ebenezer

Linkage and Recombination

Brown Body
Red Eyed
Male

Yellow Body
White Eyed
Female

F1 Generation

Brown Body
Red Eyed
Female

Yellow Body
White Eyed
Male

F2 Generation

98.7 % Parental Type

Brown Body
Red Eye

Yellow Body
White Eye

Brown Body
White Eye

Yellow Body
Red Eye

1.3 % Recombinant Type

B. John Ebenezer

Linkage and Recombination

Morgan stated this is due to the physical association or linkage of the two genes and coined the term **linkage.**

Linkage:

Physical association of genes located on the same chromosome is called linkage**.**

Recombination:

The formation of **non-parental** gene combinations leading to the formation of new characters.

B. John Ebenezer

Linkage and Recombination

Morgan's Finding:

Genes which are very **tightly linked** shows very low recombination

Genes which are **loosely linked** shows higher recombination.

The genes for **white eye** and **yellow body** are very tightly linked and shows 1.3 % recombination.

The genes for **white eye** and **miniature wing** are loosely linked and shows 37.2 % recombination.

B. John Ebenezer

Linkage and Recombination

Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and **'mapped'** their position on the chromosome.

The technique of identifying the location of genes and the distance between the genes based on the frequency of recombination is called gene map.

B. John Ebenezer

Linkage and Recombination

	Dominant	Recessive
Eye colour	Red (w+)	White (w)
Wing size	Normal(m+)	Miniature (m)

B. John Ebenezer

Linkage and Recombination

All genes are present on X chromosome (Sex linkage)

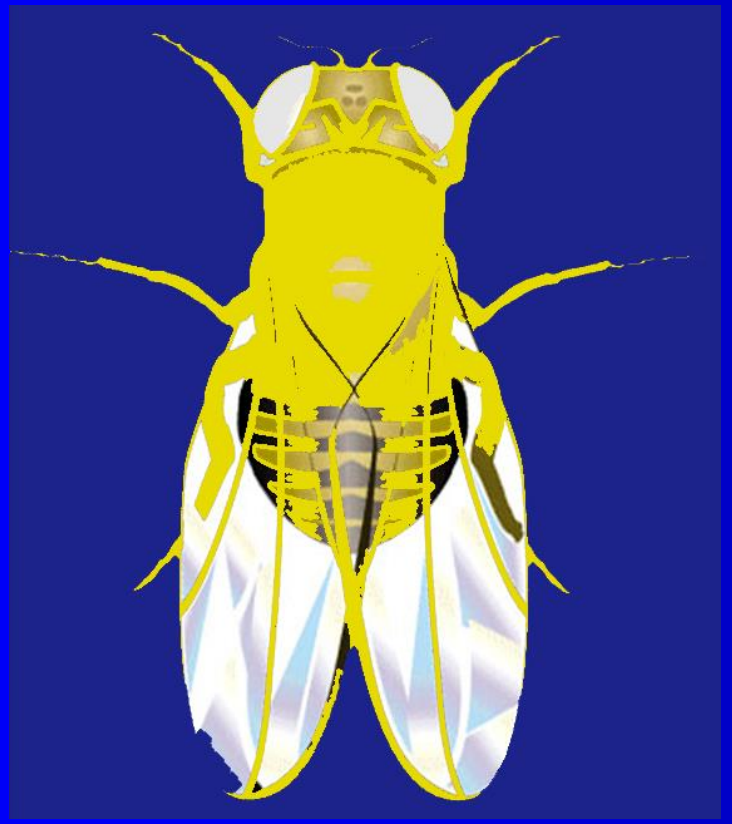
	Dominant	Recessive
Body Colour	Brown (y+)	Yellow (y)
Eye Colour	Red (w+)	White (w)
Wing Size	Normal (m+)	Miniature (m)

Above genes are absent in Y chromosome

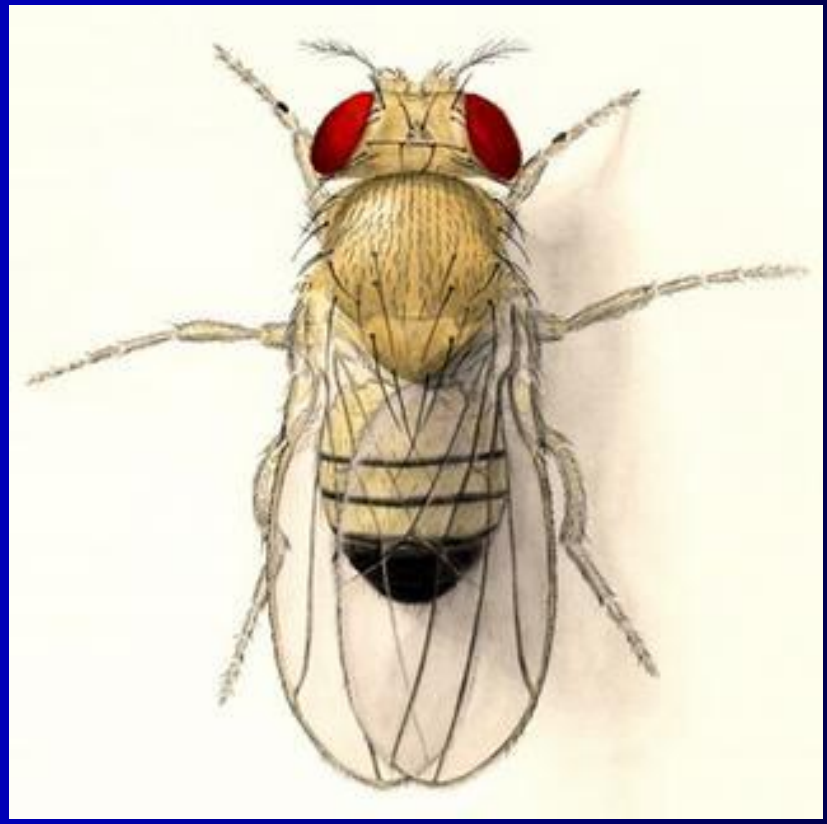
B. John Ebenezer

Linkage and Recombination in Drosophila

Yellow Body
White Eye



Brown Body
Red Eye



B. John Ebenezer

Linkage and Recombination

Brown Body
Red Eyed
Male

Yellow Body
White Eyed
Female

F1 Generation

Brown Body
Red Eyed
Female

Yellow Body
White Eyed
Male

F2 Generation

98.7 % Parental Type

Brown Body
Red Eye

Yellow Body
White Eye

Brown Body
White Eye

Yellow Body
Red Eye

1.3 % Recombinant Type

B. John Ebenezer

Linkage and Recombination

Cross 1

Female Drosophila ♀

y w

X

y w

X

Male Drosophila ♂

y^+ w^+

X

Y

y - Yellow body colour

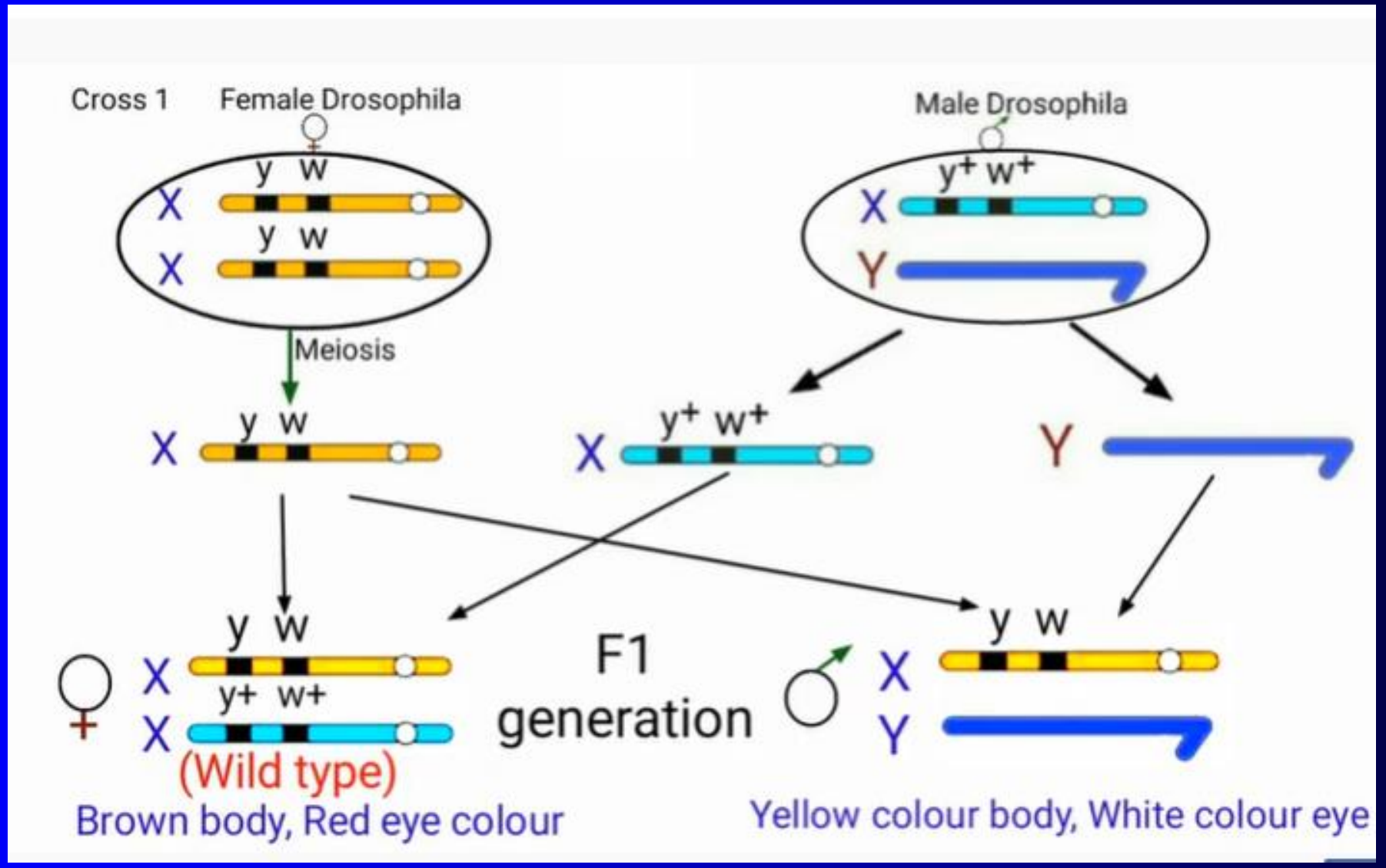
w - White eye colour

y^+ -Brown body colour

w^+ -Red colour eye

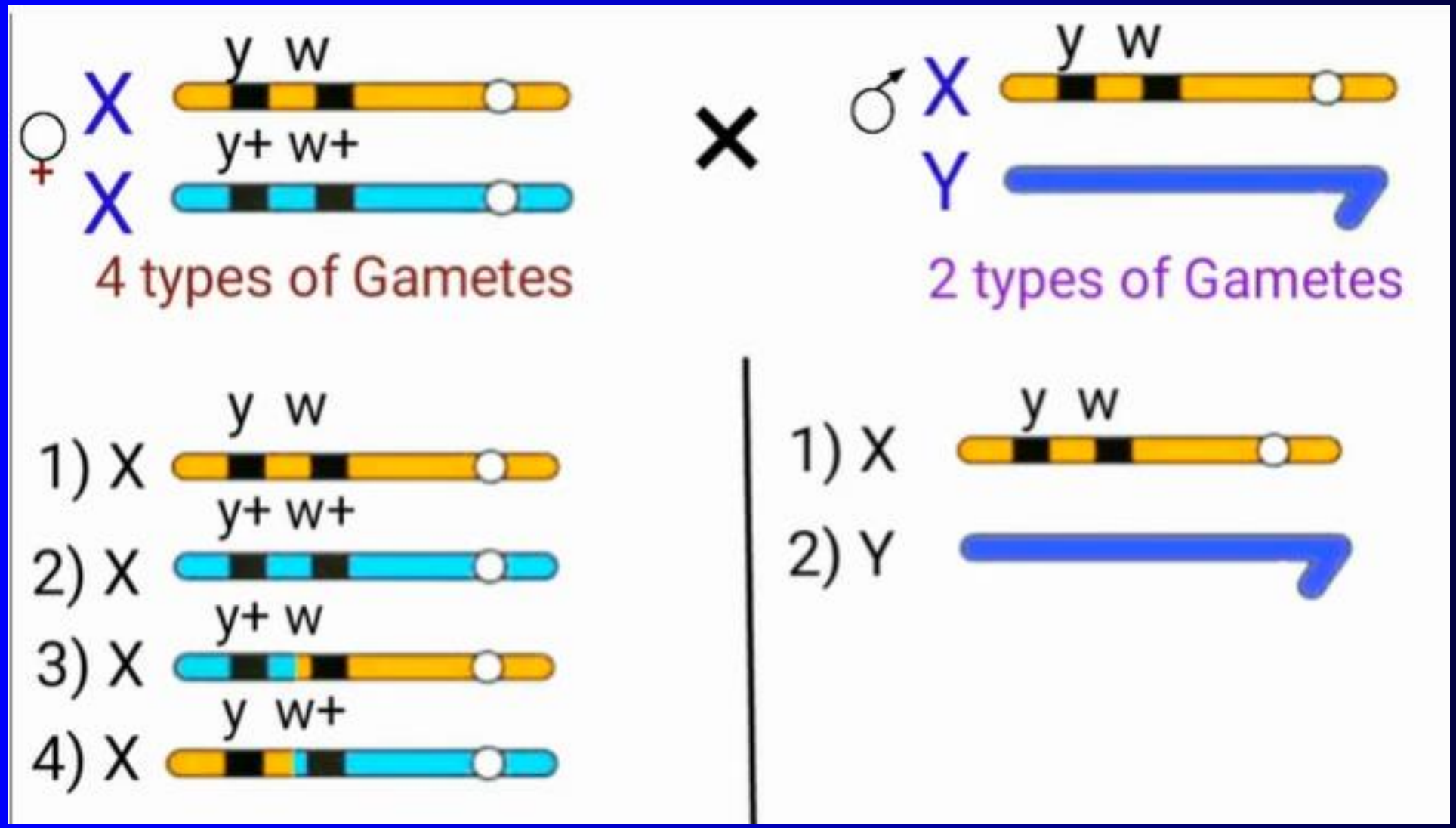
B. John Ebenezer

Linkage and Recombination



B. John Ebenezer

Gametes of F1



B. John Ebenezer

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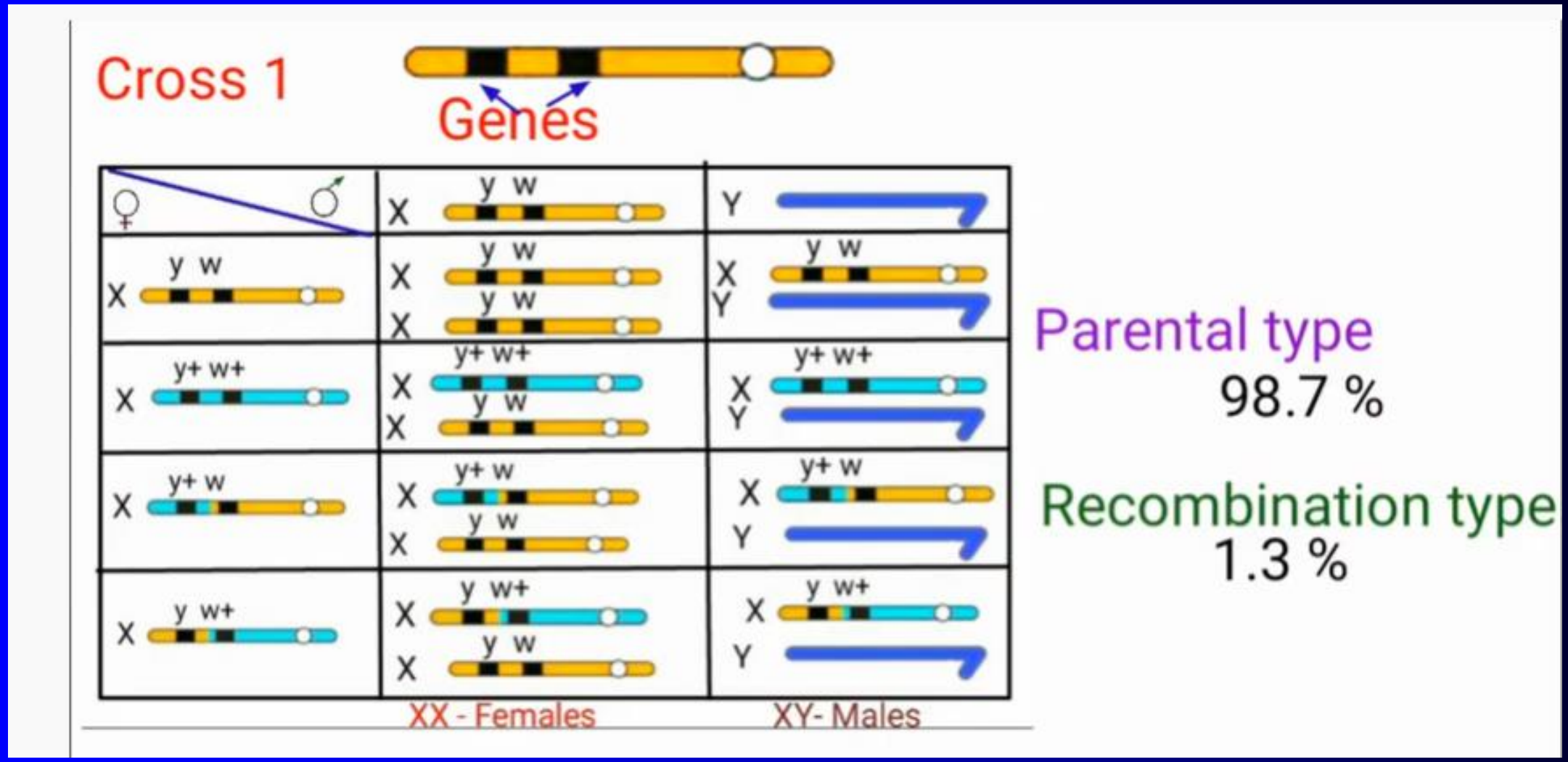
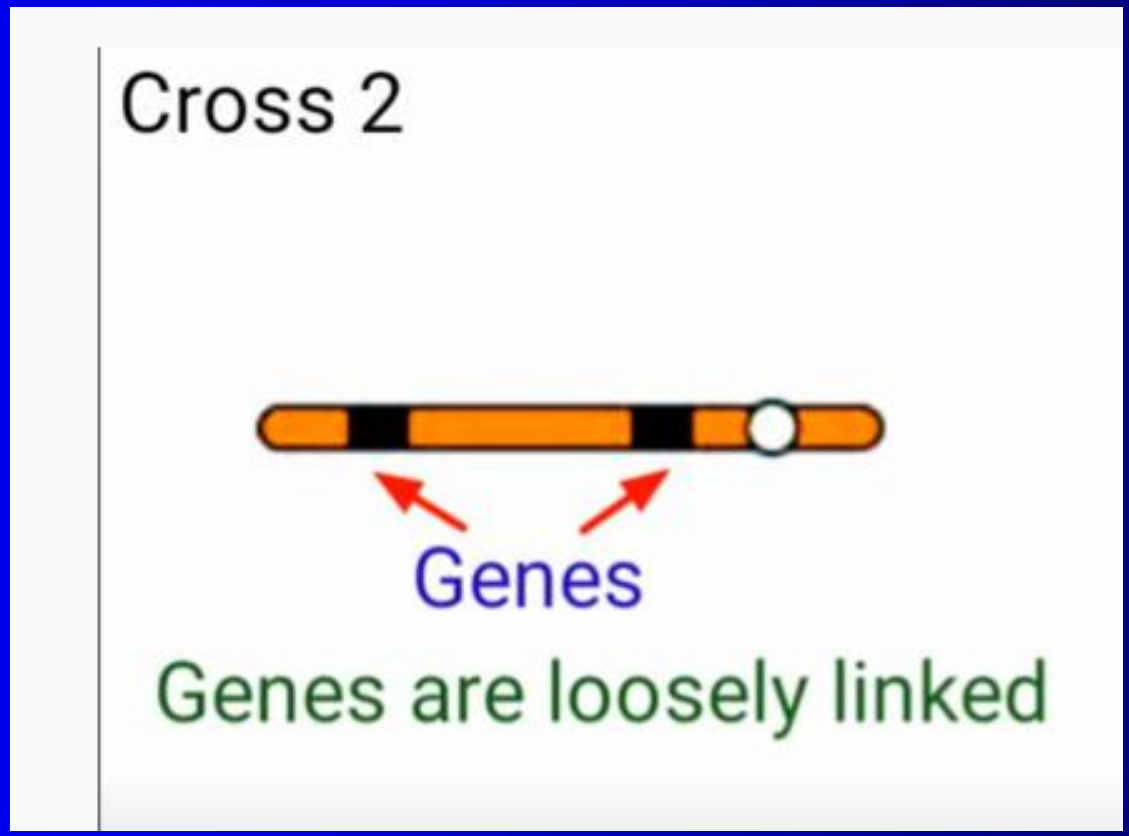


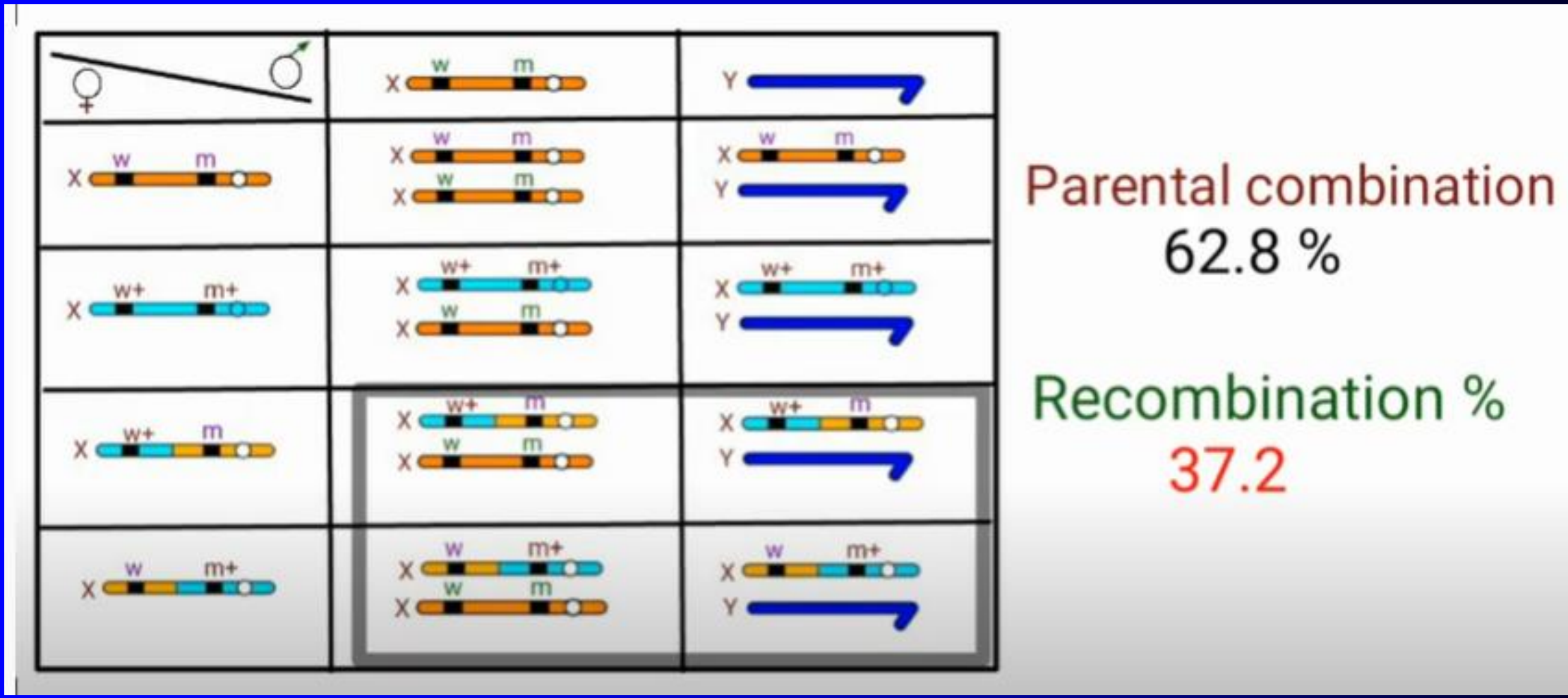
B. John Ebenezer

White eye and Miniature wing



B. John Ebenezer

The genes for **white eye** and **miniature wing** are loosely linked and shows 37.2 % recombination.



B. John Ebenezer

Linkage and Recombination

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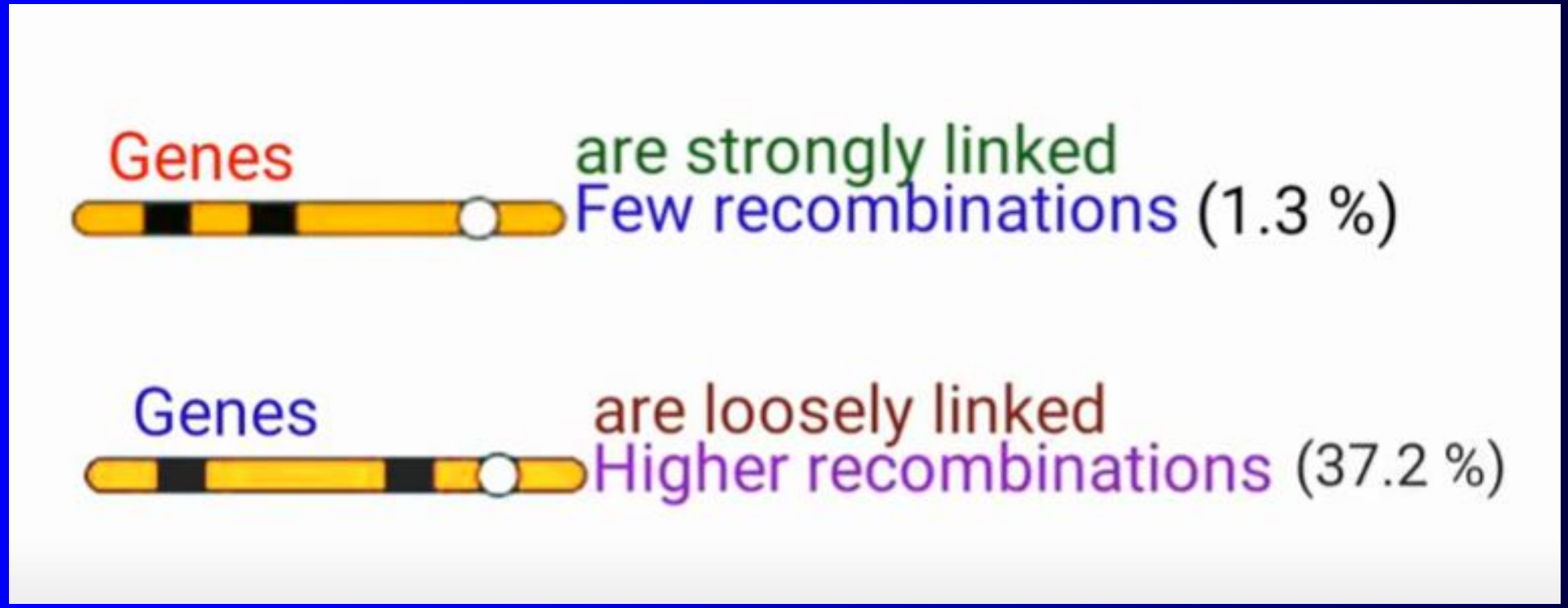
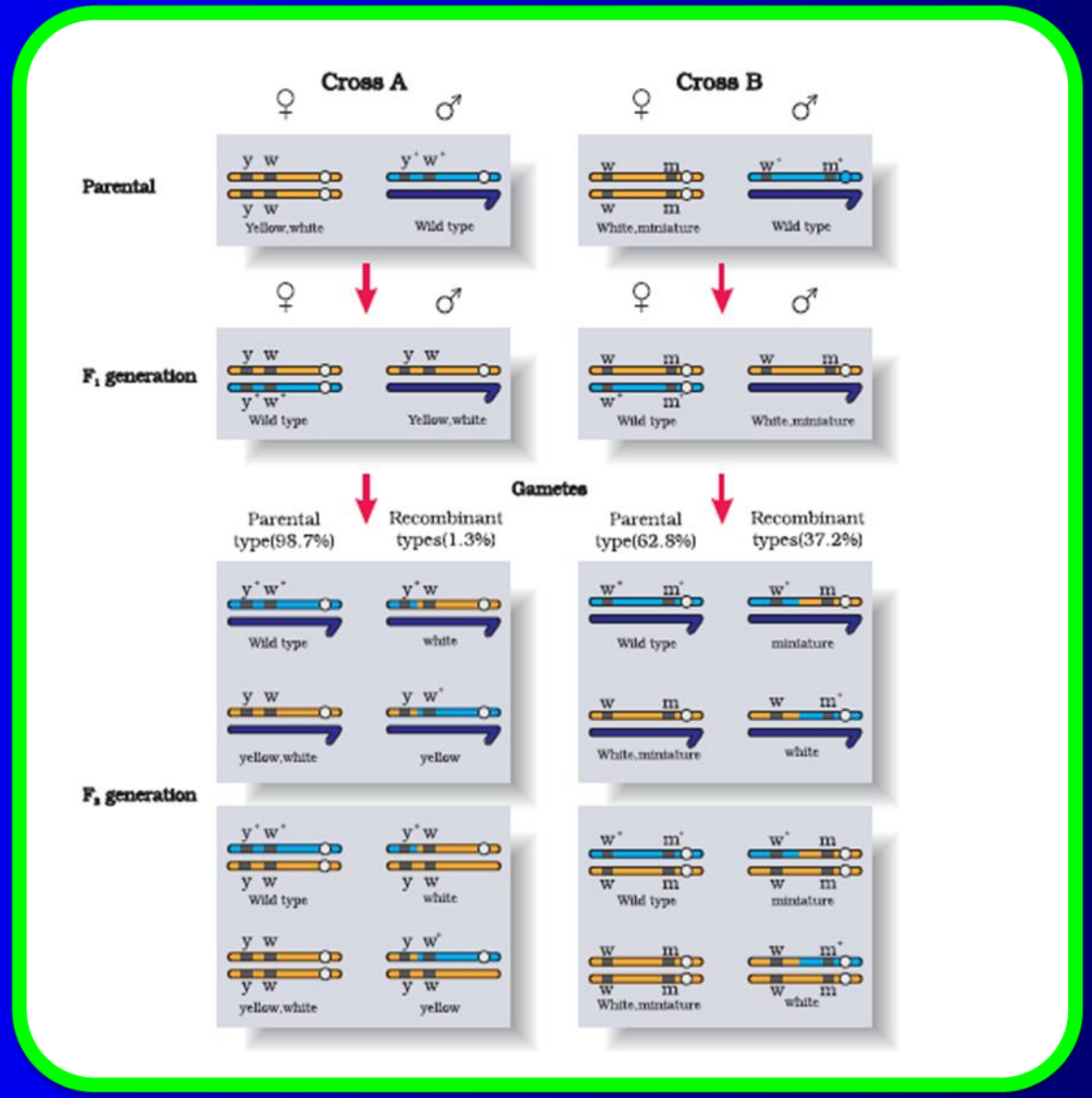
B. John Ebenezer

The genes for **white eye** and **miniature wing** are loosely linked and shows 37.2 % recombination.



B. John Ebenezer

Linkage and Recombination



B. John Ebenezer

Sex Determination

Sex Determination

Henking (1891) traced specific nuclear structure during spermatogenesis of some insects.

50 % of the sperms received this specific structure, whereas 50% of sperms did not receive it.

Henking gave a name to this structure as the **X-body**.

X-body of Henking was later named as X-chromosome.

B. John Ebenezer

Sex Determination in Grasshopper

Sex-determination in grasshopper is **XX-XO type.**

All the eggs bear one ‘X’ chromosome along with autosomes.

50% of sperms have ‘X’ chromosome and the other 50% of sperms do not have ‘X’ chromosome.

The egg fertilized with sperm with ‘X’ chromosome becomes female (22+XX).

The egg fertilized with sperm without ‘X’ chromosome becomes male (22 + XO)

B. John Ebenezer

Sex Determination in Grasshopper

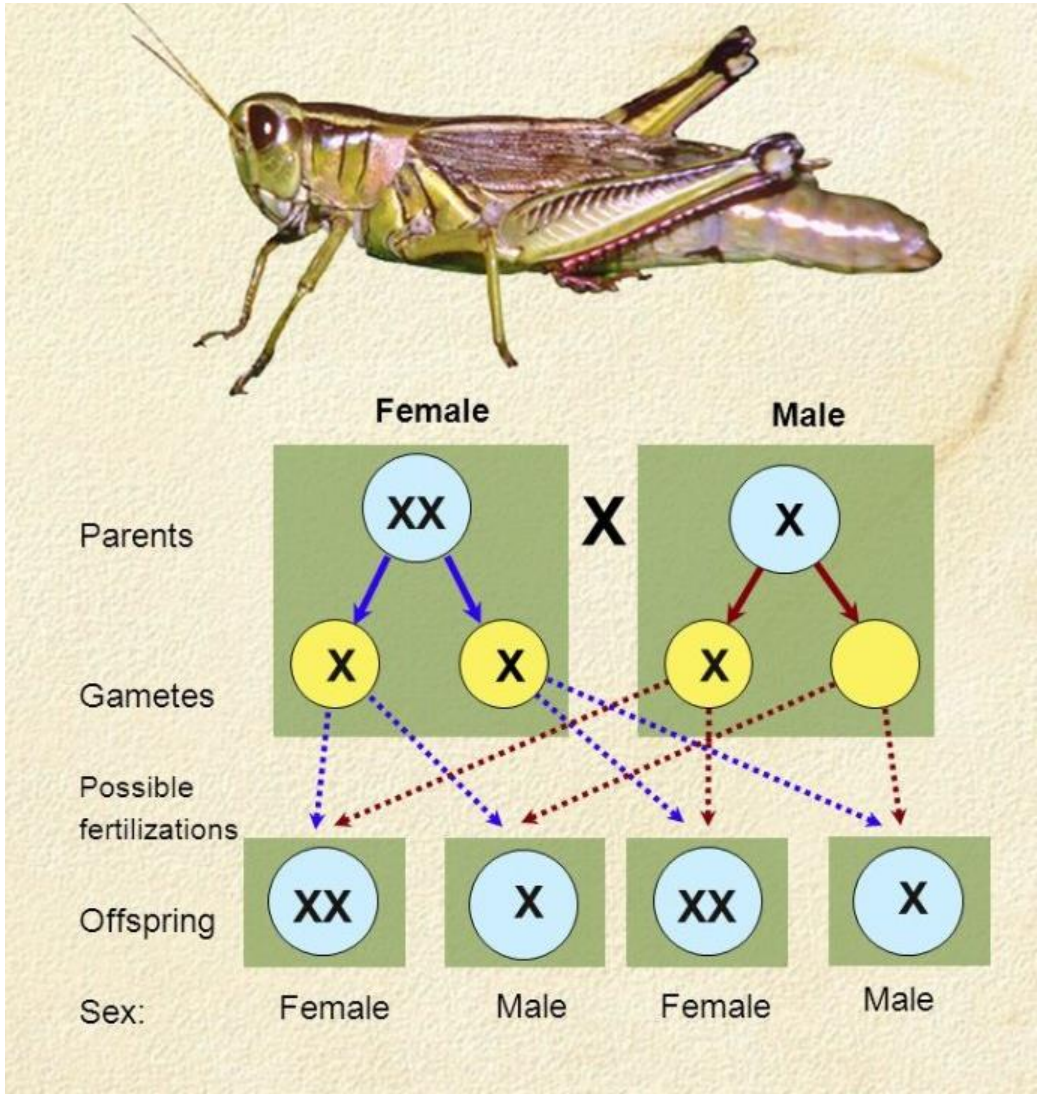
Diploid chromosomal number of female grasshopper is 2n = 24.

Diploid chromosomal number of male grasshopper is 2n = 23.

This is because XX-XO mechanism of sex determination present in grasshoppers.

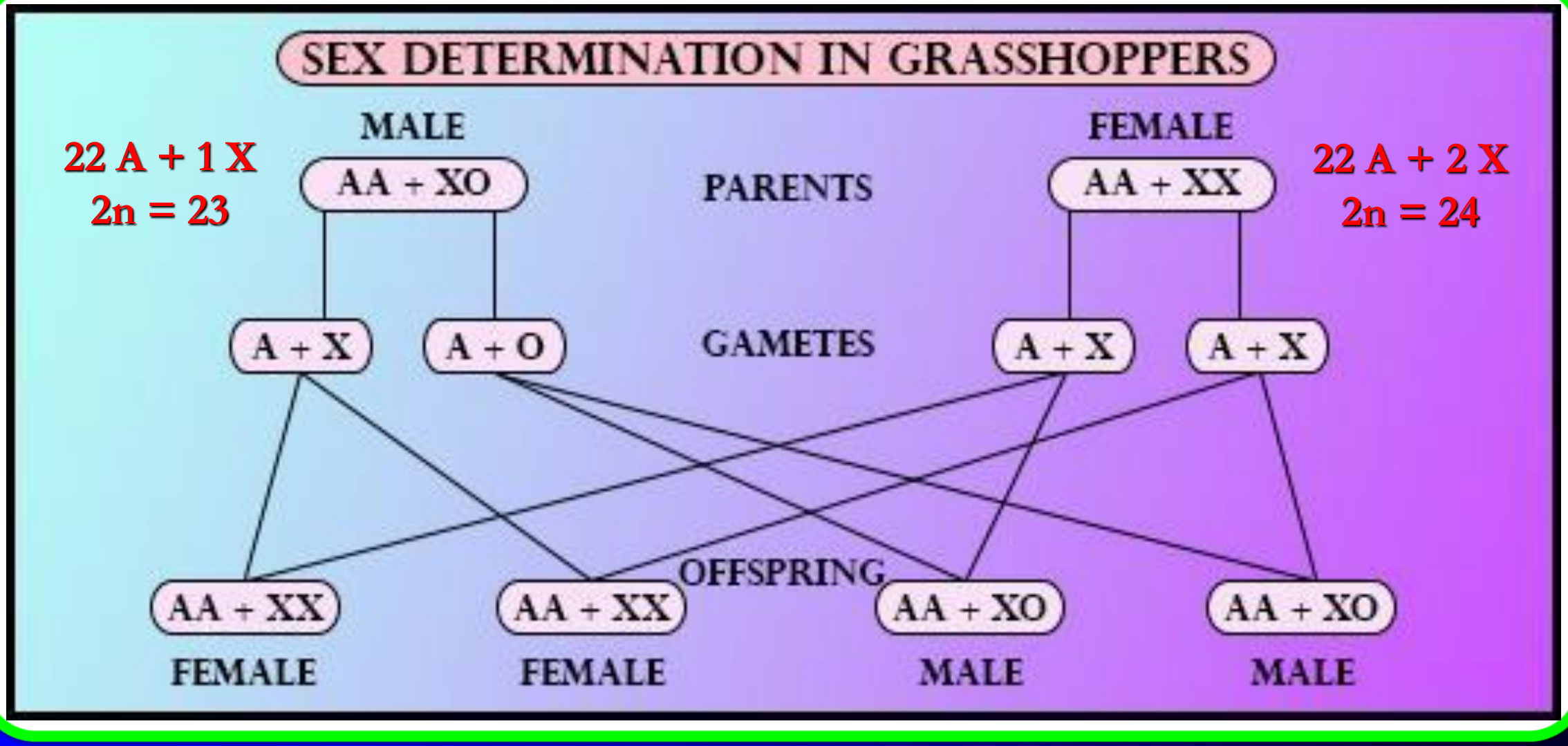
B. John Ebenezer

Sex Determination in Grasshopper



B. John Ebenezer

Sex Determination in Grasshopper



B. John Ebenezer

Sex Determination in Birds

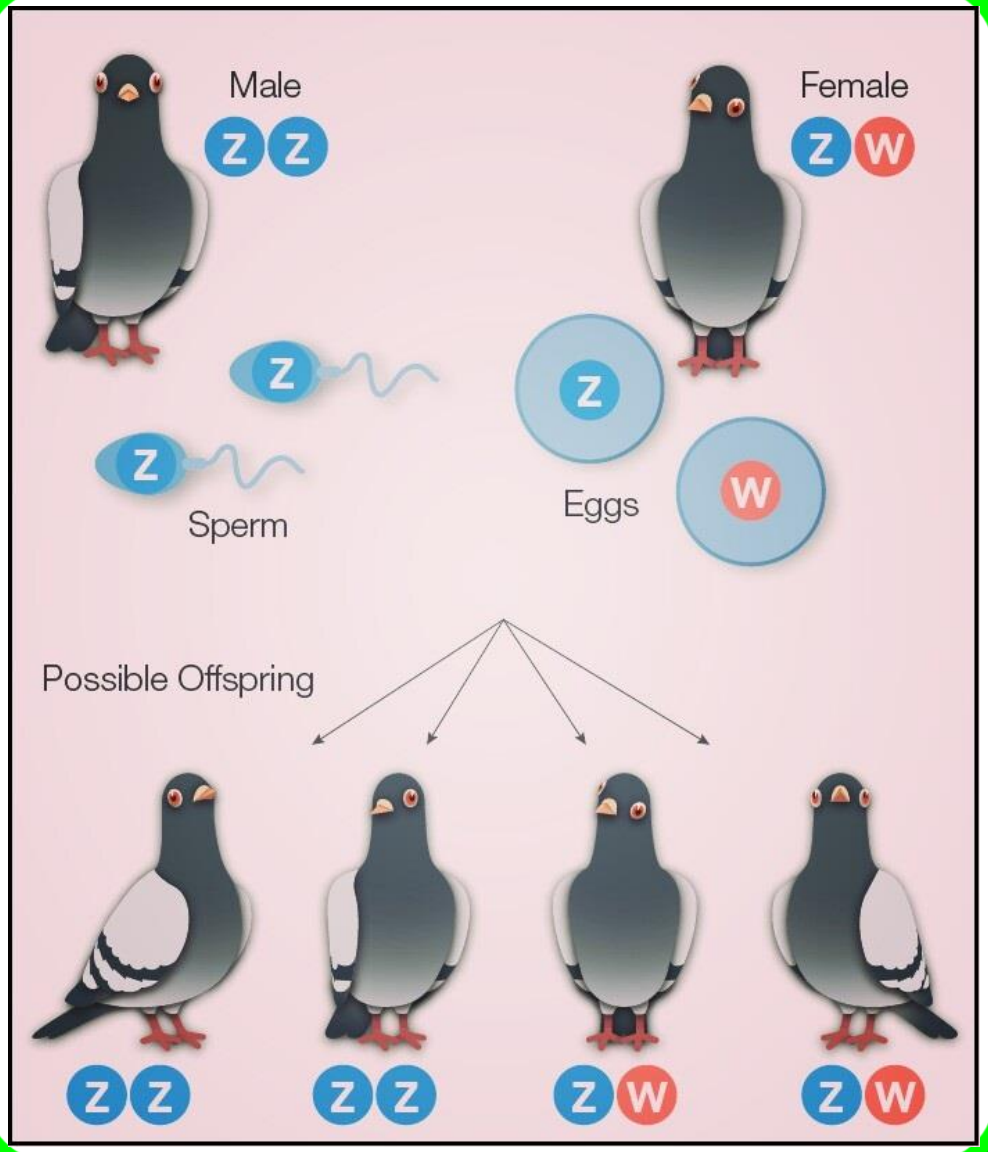
Male birds have two similar sex chromosomes and called **ZZ.**

Female birds have two different sex chromosomes called **Z** and **W.**

Such type of sex determination is called **female heterogamety** and **male homogamety.**

B. John Ebenezer

Sex Determination in Birds



B. John Ebenezer

Sex Determination in Honey Bees

Sex determination in honey bee is based on the number of sets of chromosomes an individual receives.

An offspring formed from the fertilization of a sperm and an egg develop into either queen (female) or worker (female).

An unfertilized egg develops into a male (drone), by means of parthenogenesis.

The males have half the number of chromosomes than that of the females.

B. John Ebenezer

Sex Determination in Honey Bees

The females are diploid having 32 chromosomes and males are haploid having 16 chromosomes.

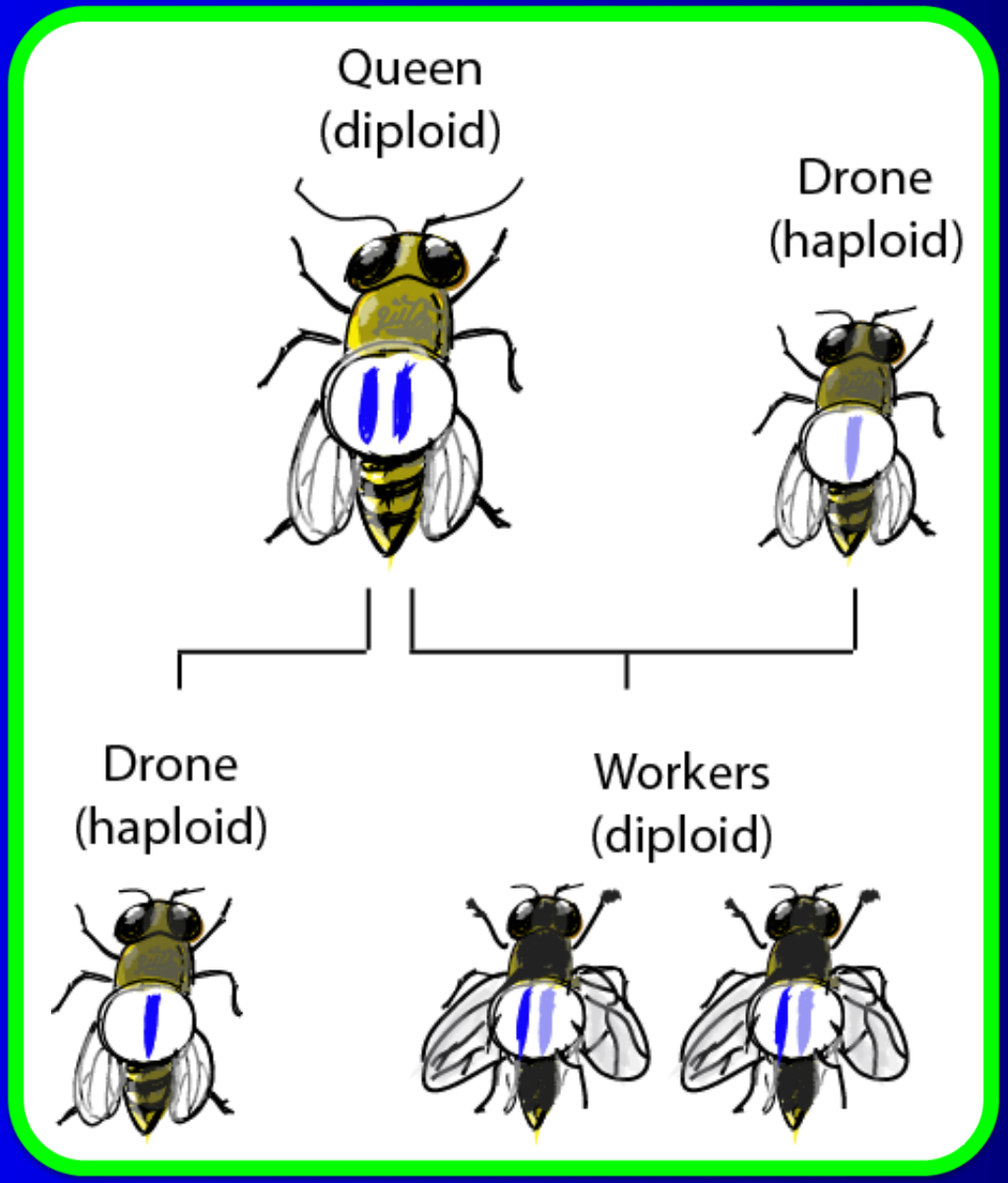
This is called **haplodiploid sex determination**.

Males produce **sperms by mitosis**.

Males do not have father and thus cannot have sons, but have **grandsons.**

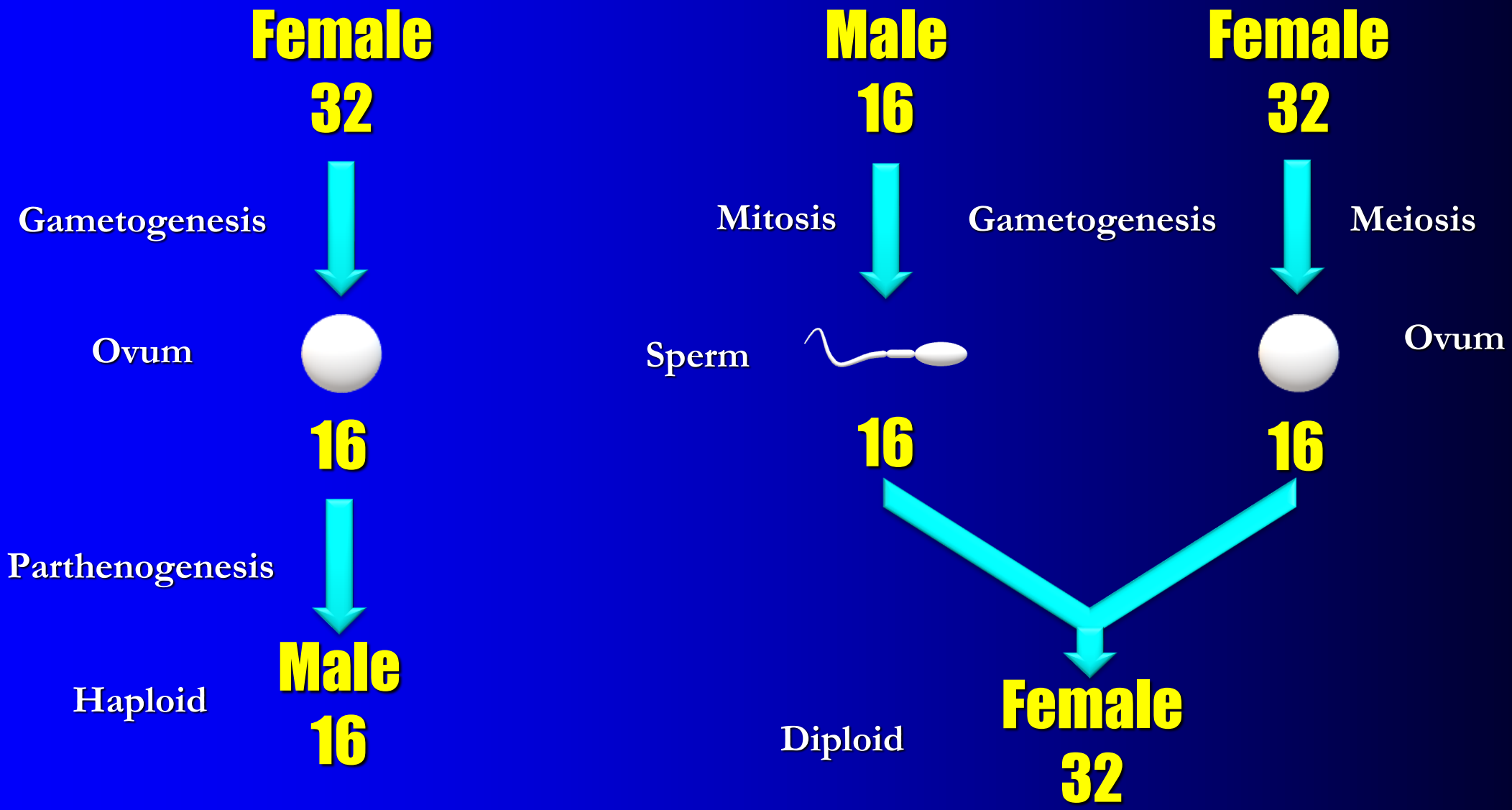
B. John Ebenezer

Sex Determination in Honey Bees



B. John Ebenezer

Sex Determination in Honey Bee



B. John Ebenezer

Sex Determination in Honey Bee

Mother
32

Gametogenesis

16

Parthenogenesis

Male Bee
16

Female Bee
32

Mitosis

Gametogenesis

Meiosis

Haploid

16

16

Daughter (32)

Diploid

Gametogenesis

Haploid

(16)

Parthenogenesis

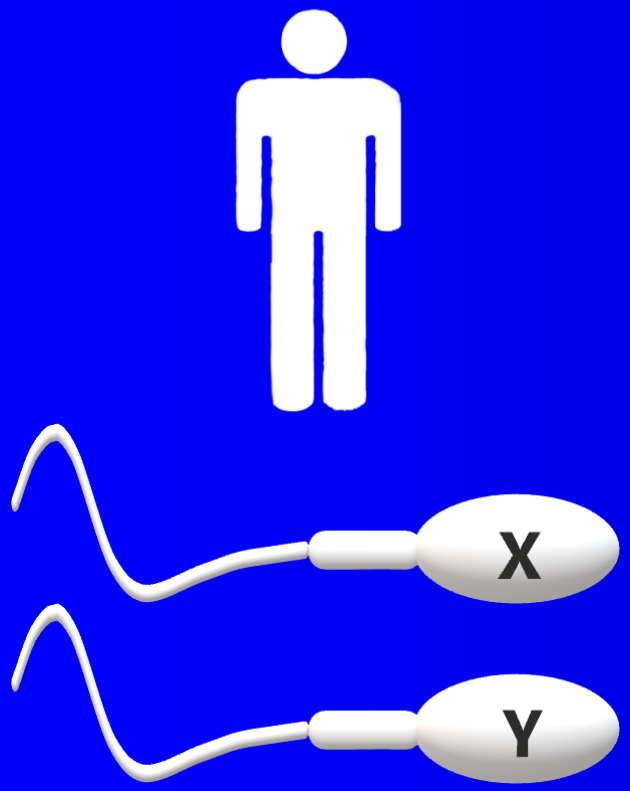
Haploid

Grandson (16)

B. John Ebenezer

Sex Determination in Man

Men are Heterogametic



Men are heterogametic because they produce two different kinds of gametes.

The two kinds of gametes are, sperms with X chromosomes and sperms with Y chromosomes.

50% of the sperms contain X chromosomes and the other 50% of the sperms contain Y chromosomes.

Hence, there is 50% possibility for having male babies and 50% possibility for having female babies.

B. John Ebenezer

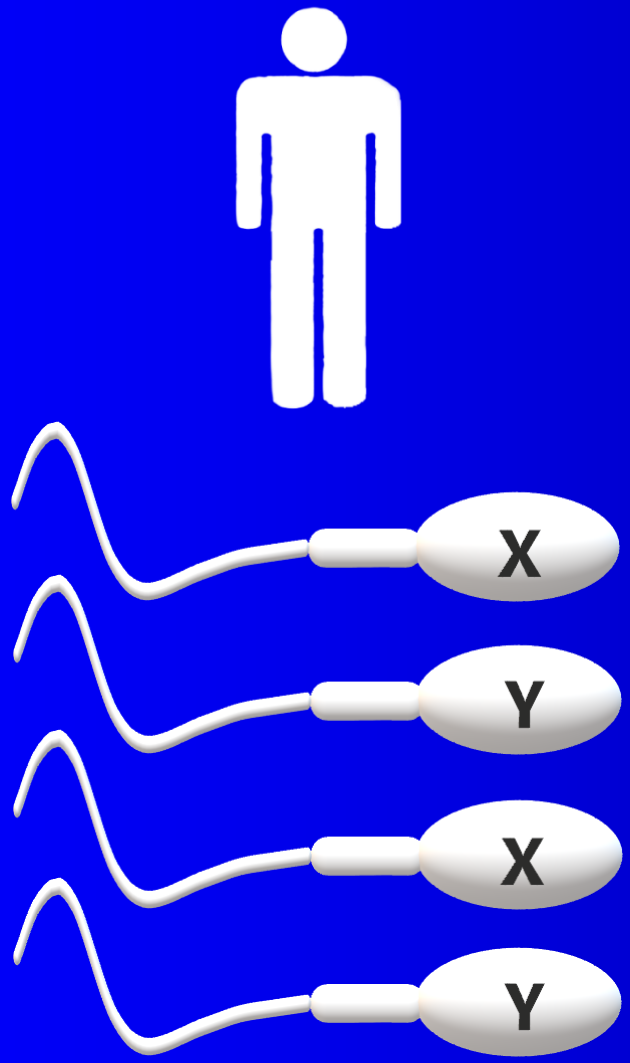
Women are Homogametic

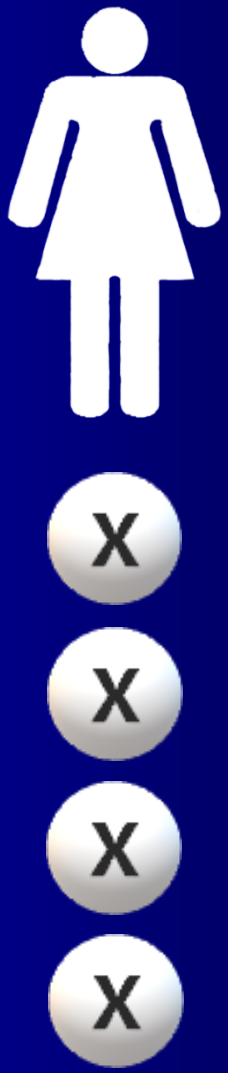
Women are homogametic because all the gametes produced by them are of the same kind.

All the ova produced by women carry only X chromosomes.

B. John Ebenezer

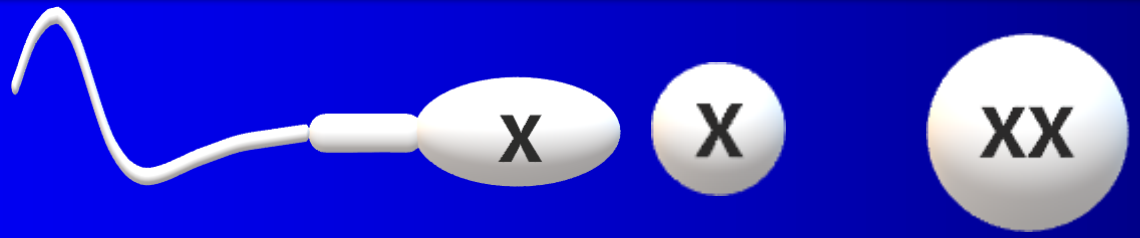
Men are Heterogametic and Women are Homogametic





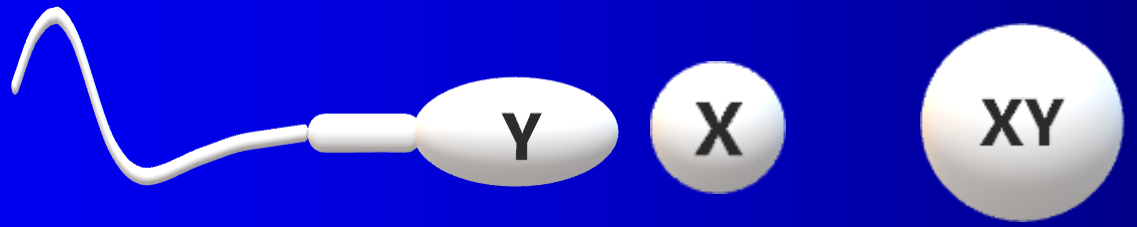
B. John Ebenezer

Father determines the sex of the baby



Female

When a sperm with X Chromosome fuses with the ovum, with X chromosome, the zygote gets XX chromosomes. The zygote with XX chromosomes, develops into a female baby.

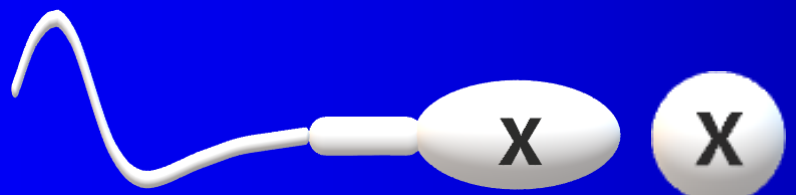


Male

When a sperm with Y Chromosome fuses with the ovum, with X chromosome, the zygote gets XY chromosomes. The zygote with XY chromosomes, develops into a male baby.

B. John Ebenezer

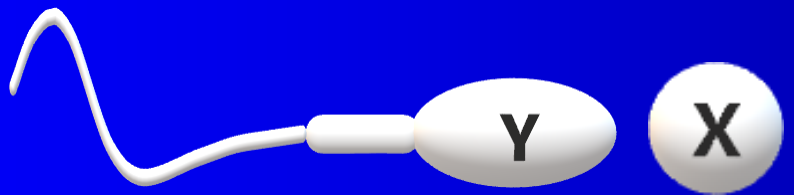
Fertilization



X

XX

Female



X

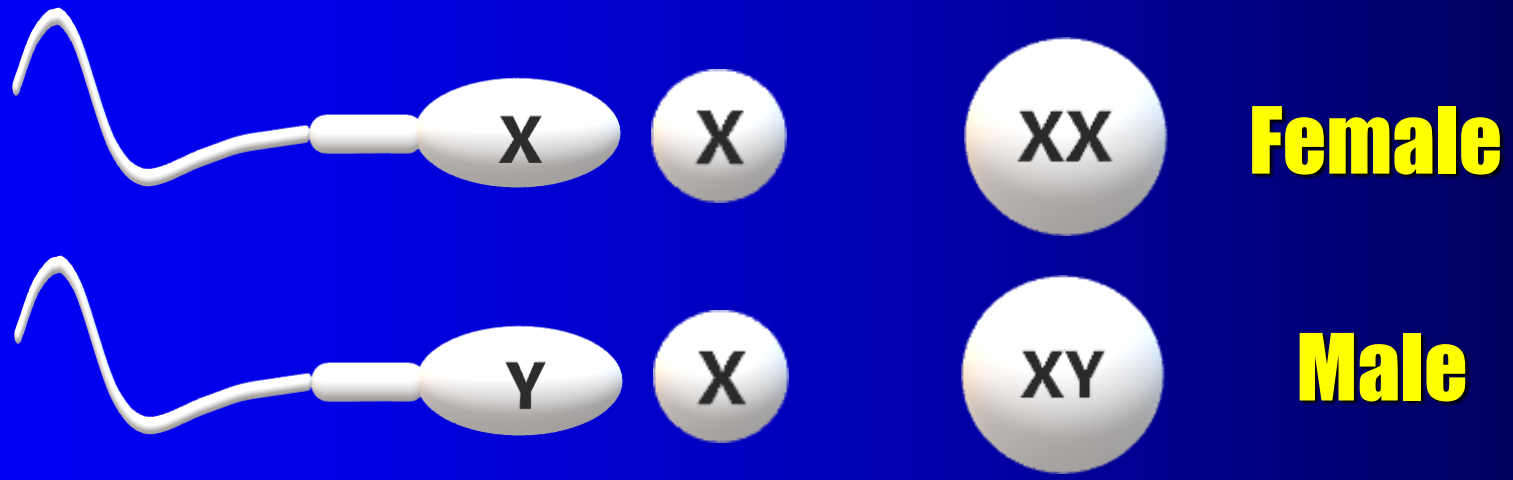
XY

Male

B. John Ebenezer

Sex of the offsprings depends on the type of sperm fusing the ovum.

Hence, it is proved that father determines the sex of the baby, and not the mother.



B. John Ebenezer

Maintenance of a constant Number of Chromosomes

Every species has a constant number of chromosomes.

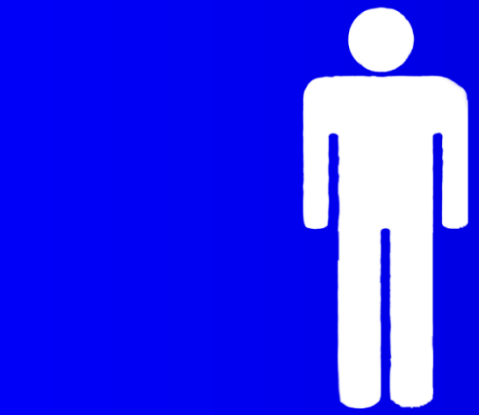
Human beings have 46 chromosomes.

During gametogenesis, the chromosome number is reduced to a half by means of a cell division known as meiosis.

Hence, Sperms and Ova have only 23 chromosomes. (i.e., 22 Autosomes and 1 sex chromosome or Allosome. (22+X, 22+Y)

B. John Ebenezer

Chromosome Number is Reduced to a Half during Gametogenesis



44 + XY

44 + XX

Meiosis

22 + X 22 + Y

22 + X

B. John Ebenezer

Chromosome Number is Maintained during Fertilization

During fertilization, the sperm fuses with ovum to produce zygote.

Zygote consists of 46 chromosomes, because it is the product of fusion between sperm (23) and ovum (23).

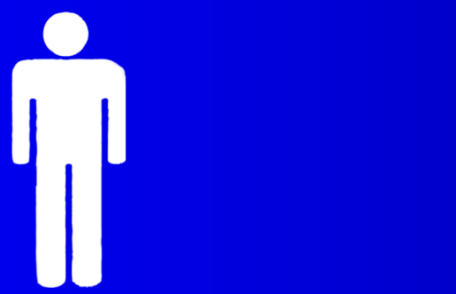
Thus the chromosome number is maintained to 46.

The zygote becomes embryo which gets implanted in the wall of the uterus and develops into a foetus.

The fully matured baby is delivered after 280 days.

B. John Ebenezer

Sex Determination in Human beings



44 + XY

44 + XX

Meiosis

22 + X 22 + Y

22 + X

Chromosome number is reduced to a half during gametogenesis by meiosis and maintained during fertilization

22 + X
22 + X

44 + X X
Female

22 + X
22 + Y

44 + X Y
Male

50% possibility for having male and female babies

Male \ Female	X	X
X	XX	XX
Y	XY	XY

Men produce two kinds of sperms.

50% of the sperms contain X chromosomes and the other 50% of the sperms contain Y chromosomes.

Hence there is 50% possibility for having male babies and 50% possibility for having female babies.

B. John Ebenezer

Mutation

Mutation

The sudden heritable change occurs in the genotype of an organism is called mutation.

Mutation results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism.

Like recombination, mutation also leads to variation in DNA.

B. John Ebenezer

Mutation

Therefore loss (deletions) or gain (insertion/duplication) of a segment of DNA, result in alteration in chromosomes.

Since genes are located on chromosomes, alteration in chromosomes results in abnormalities or aberrations.

Chromosomal aberrations are commonly observed in cancer cells.

B. John Ebenezer

Mutation

Mutation can also arise due to change in a single base pair of DNA. This is known as point mutation.

A classical example of such a mutation is sickle cell anemia.

Deletions and insertions of base pairs of DNA, causes frame-shift mutations.

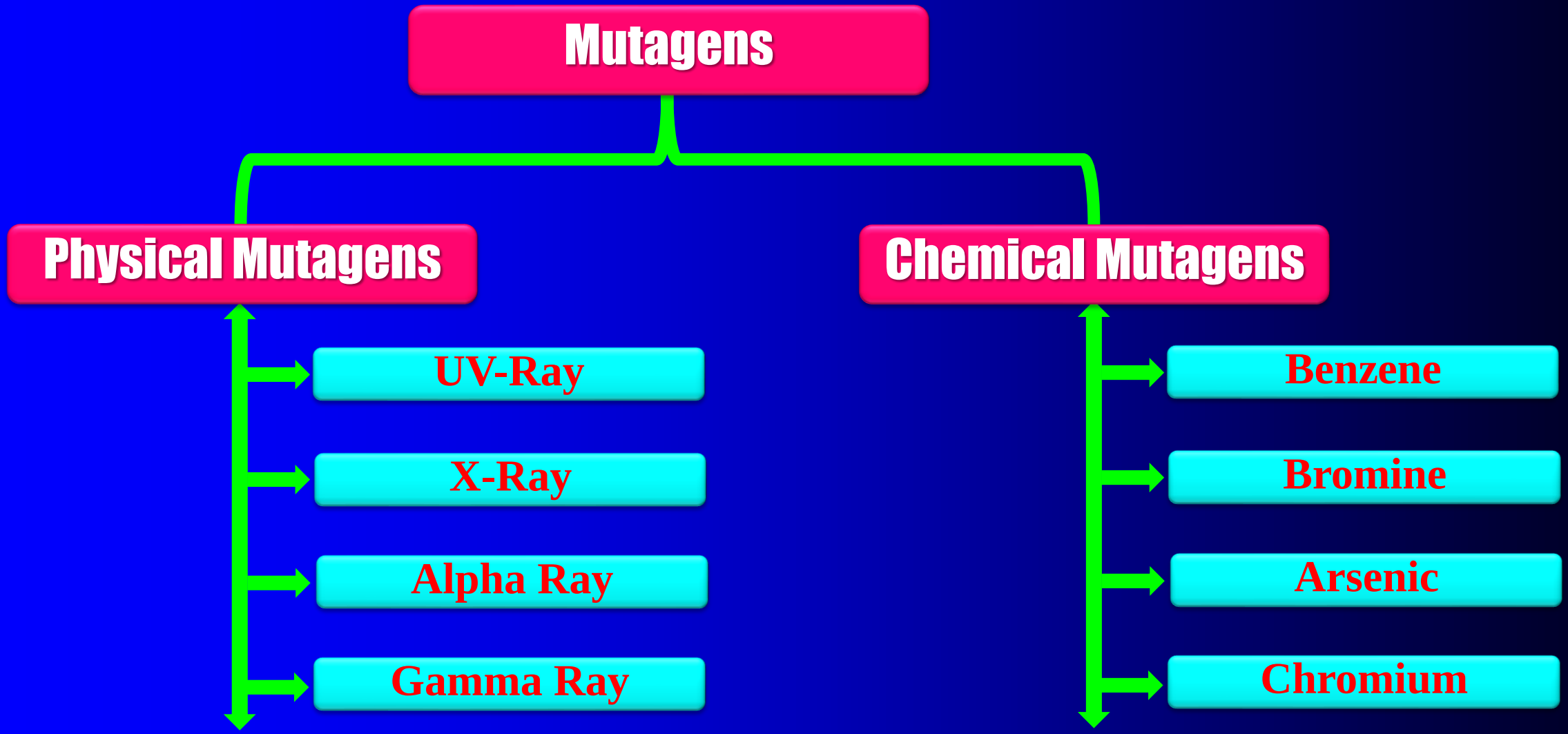
The factors which cause mutation are known as mutagens.

There are physical and chemical mutagens that induce mutations.

UV radiation is a physical mutagen.

B. John Ebenezer

The factors which cause mutation are known as mutagens.



B. John Ebenezer

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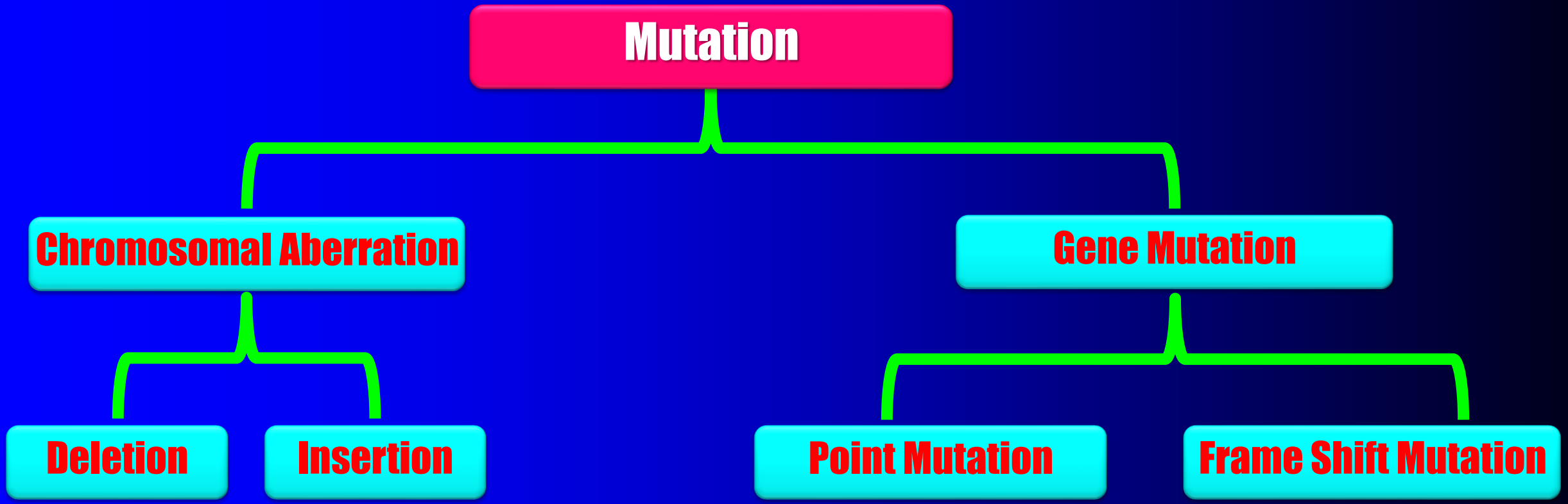
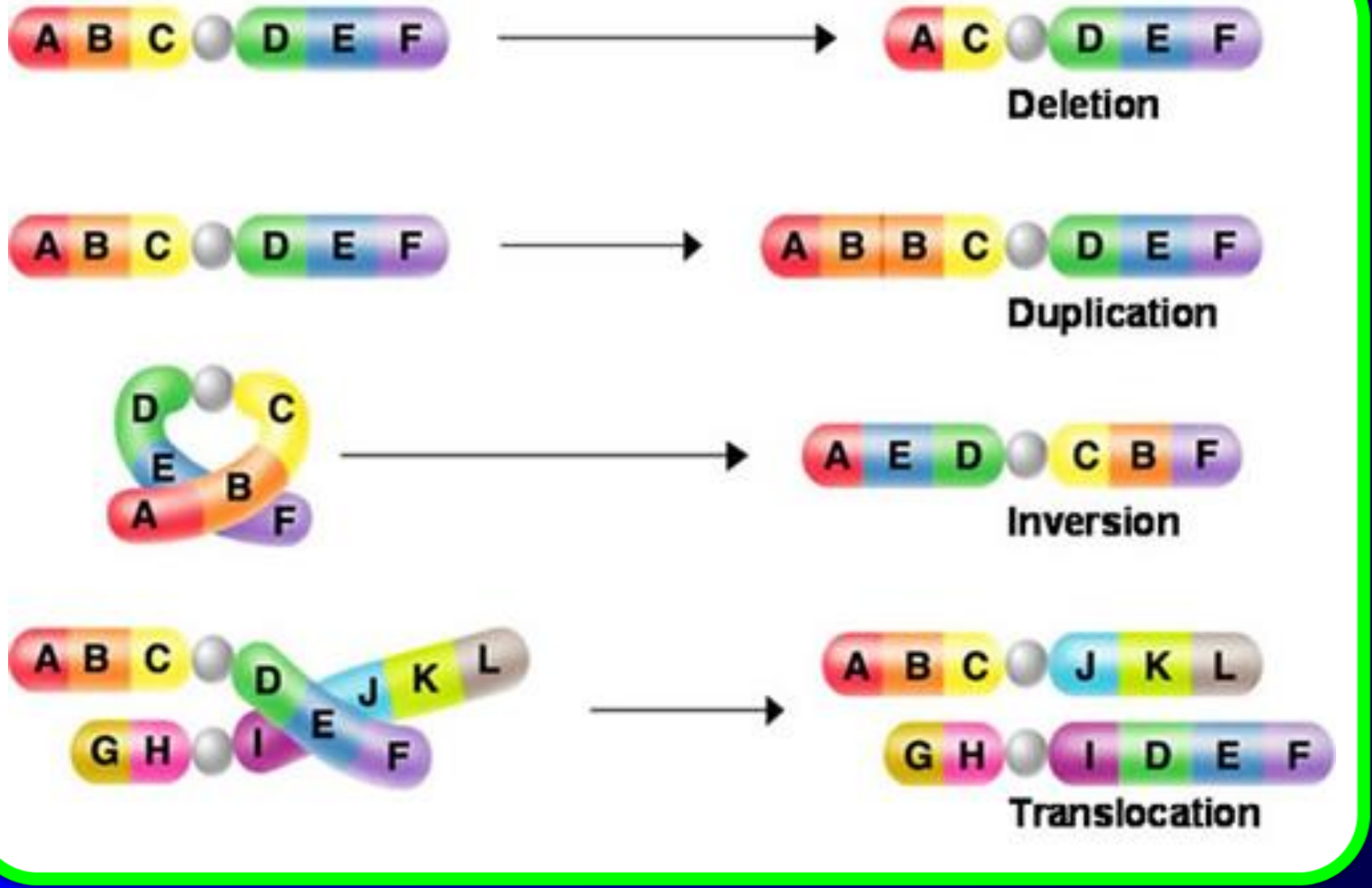


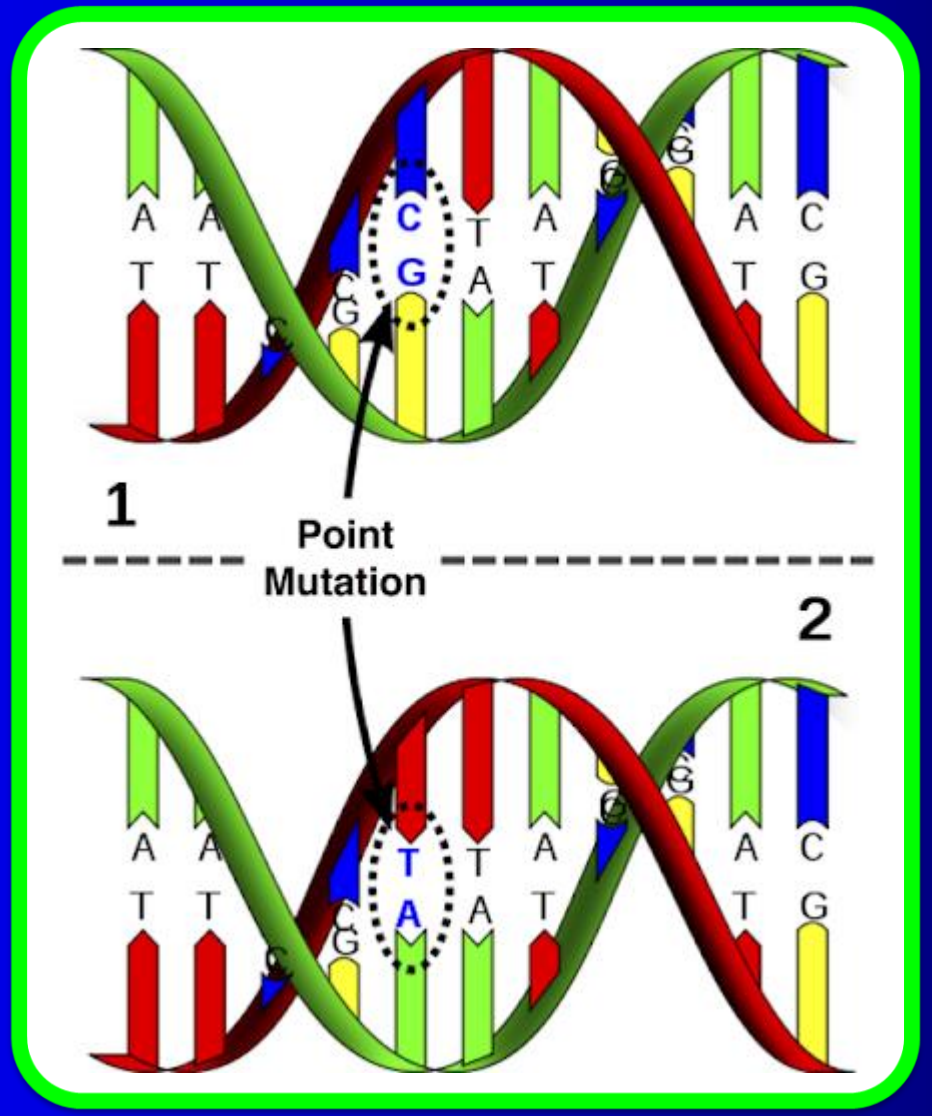
B. John Ebenezer

Chromosomal Aberration



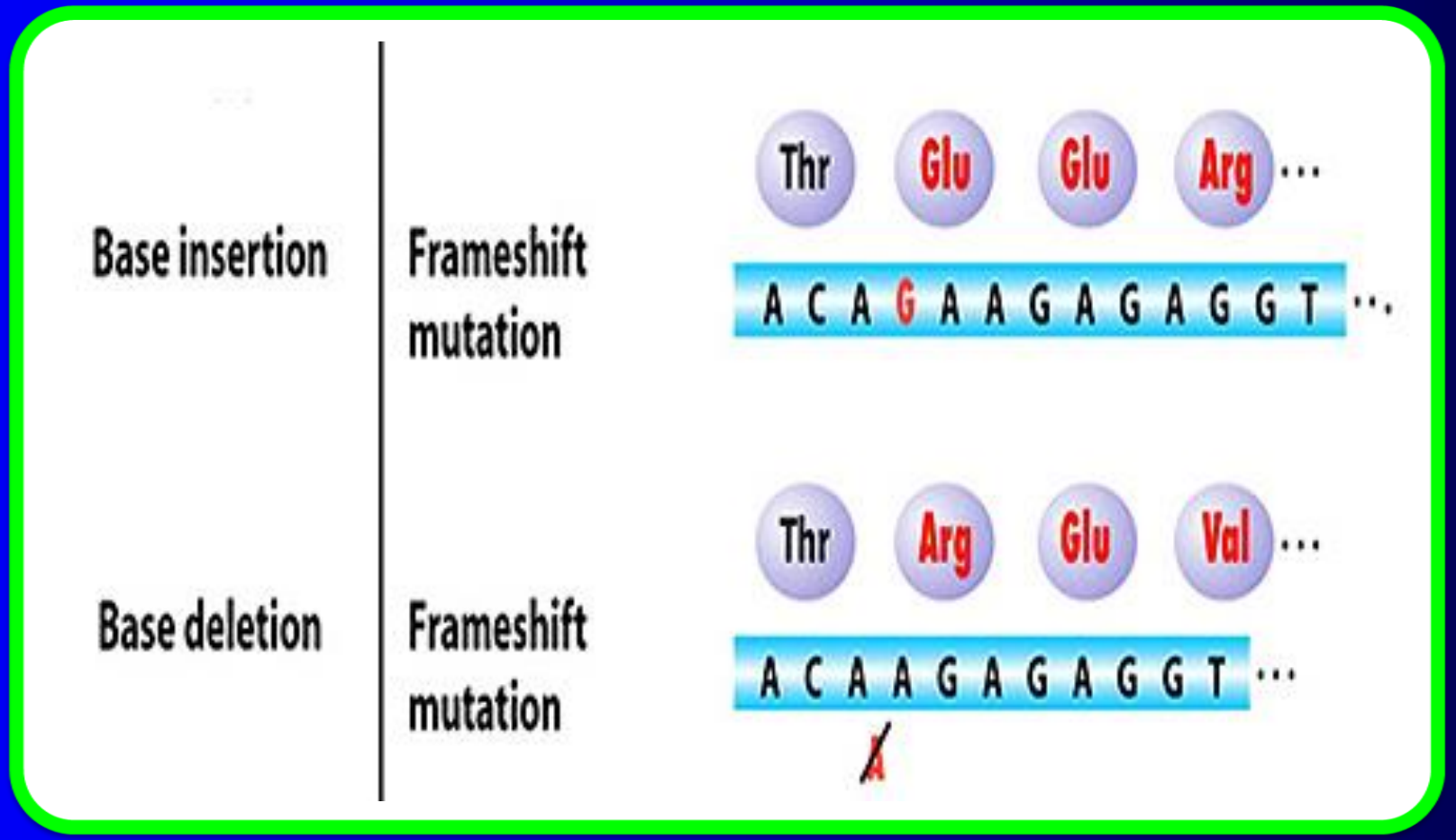
B. John Ebenezer

Point Mutation



B. John Ebenezer

Frame Shift Mutation



B. John Ebenezer

Pedigree Analysis

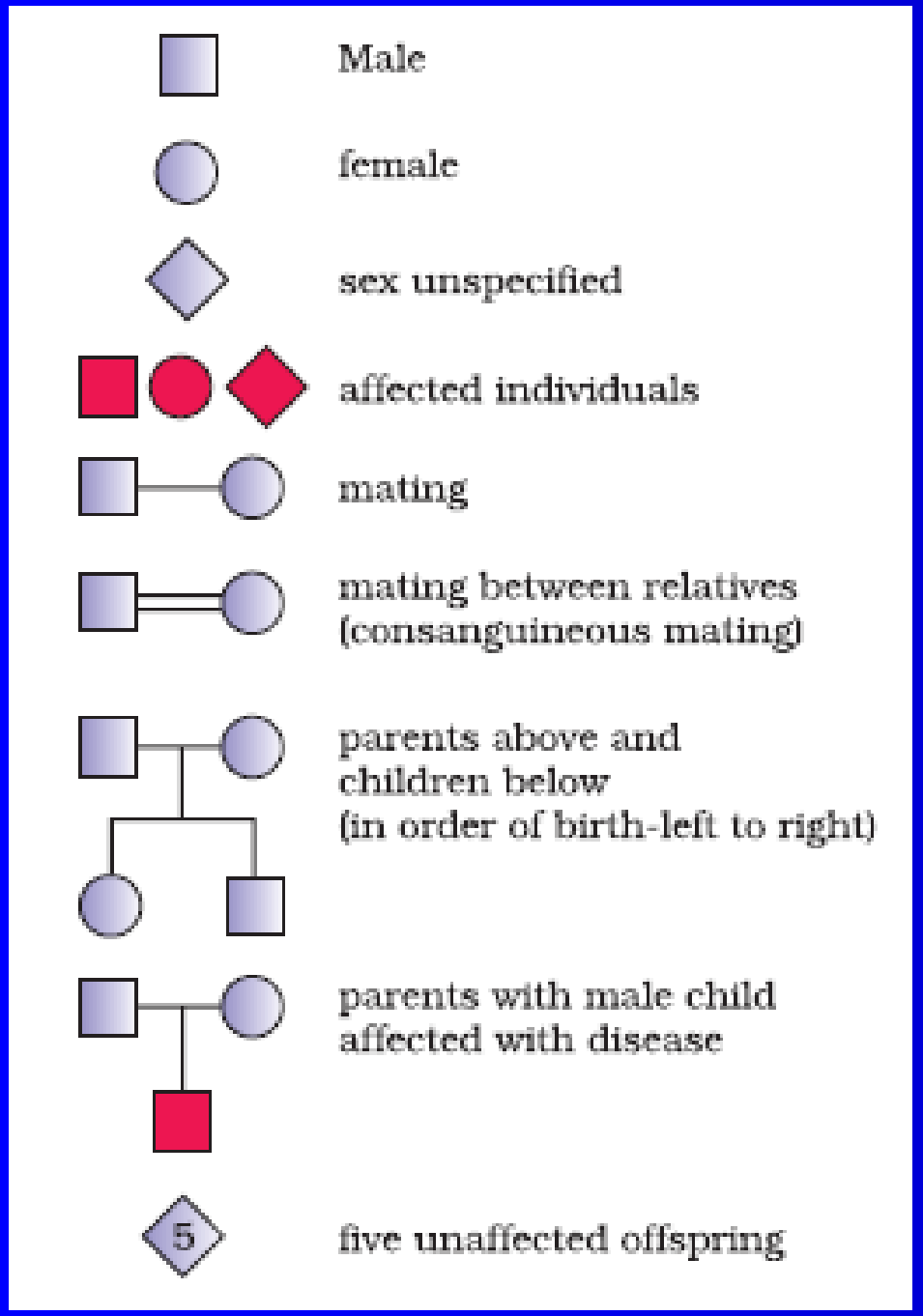
Pedigree:

Pictorial representation of a family history, a family tree that outlines the inheritance of one or more characteristics.

Proband:

The first affected person from whom pedigree analysis starts.

B. John Ebenezer



Symbols used in the human pedigree analysis

Rules of Pedigree

Rules of Pedigree

	Autosomal Dominant	Autosomal Recessive	X-Linked Dominant	X-Linked Recessive
1	Males and females are equally affected **(Autosomal)**	Males and females are equally affected **(Autosomal)**	Males and females are unequally affected **(X-Linked)**	Males and females are unequally affected **(X-Linked)**
2	Trait never skips generations **(Dominant)**	Trait skips generations **(Recessive)**	Trait never skips generations Females are more affected than males **(Dominant)**	Trait skips generations Males are more affected than females **(Recessive)**
3	Both the affected parents can have unaffected children	Unaffected parents (carriers) can have affected children	Affected father will have affected daughters	Unaffected mother (carrier) will have affected sons
4	It is caused by a dominant gene present in an autosome	It is caused by a recessive gene present in an autosome	It is caused by a dominant gene present in an allosome	It is caused by a recessive gene present in an allosome
	Both the parents affected ⬇ Children unaffected	Both the parents unaffected ⬇ Children affected	Affected Father ⬇ Affected Daughters	Unaffected Mother ⬇ Affected Sons
	No carriers	Carriers occur at heterozygous condition	No carriers	Females are carriers at heterozygous condition
	Huntington's disease Familial Hypercholesterolemia	Sickle cell anemia, Cystic fibrosis, Phenylketonuria (PKU)	Hypophatemic Rickets (Vitamin D resistant rickets)	Hemophilia, Colour blindness.

Some Examples of Mendelian Disorders

Mendelian Disorders

Autosomal Dominant

- Myotonic dystrophia
- Huntington's disease

Autosomal Recessive

- Sickle Cell Anemia
- Thalassemia
- Cystic Fibrosis
- Phenylketonuria (PKU)

X-Linked Dominant

- Hypophatemic Rickets (Vitamin D Resistant Rickets)

X-Linked Recessive

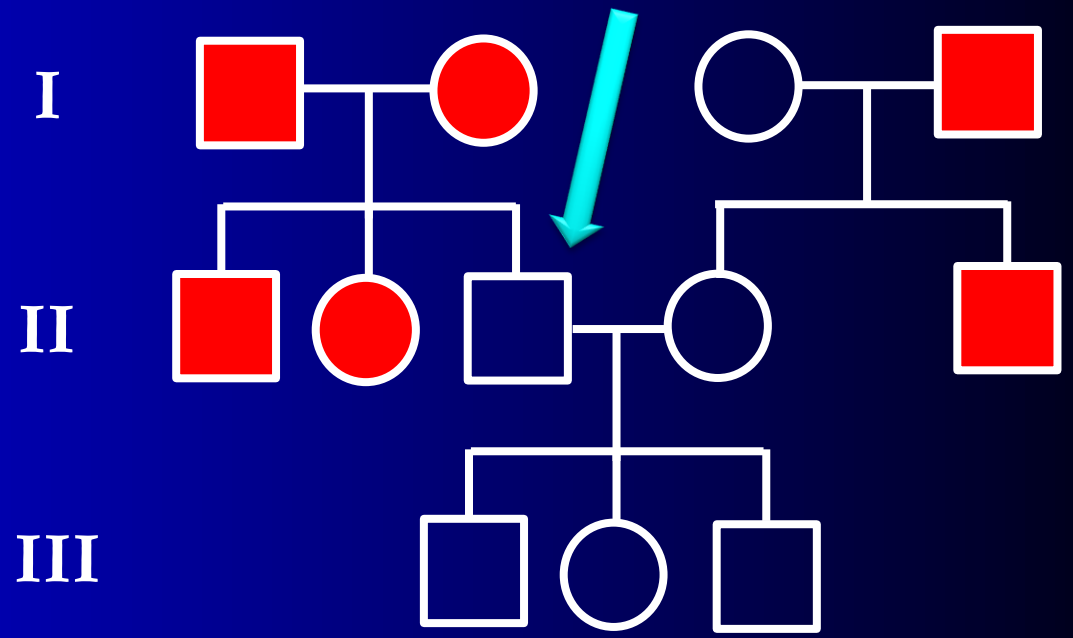
- Haemophilia
- Colour Blindness

B. John Ebenezer

Autosomal Dominant Inheritance

Autosomal Dominant Trait

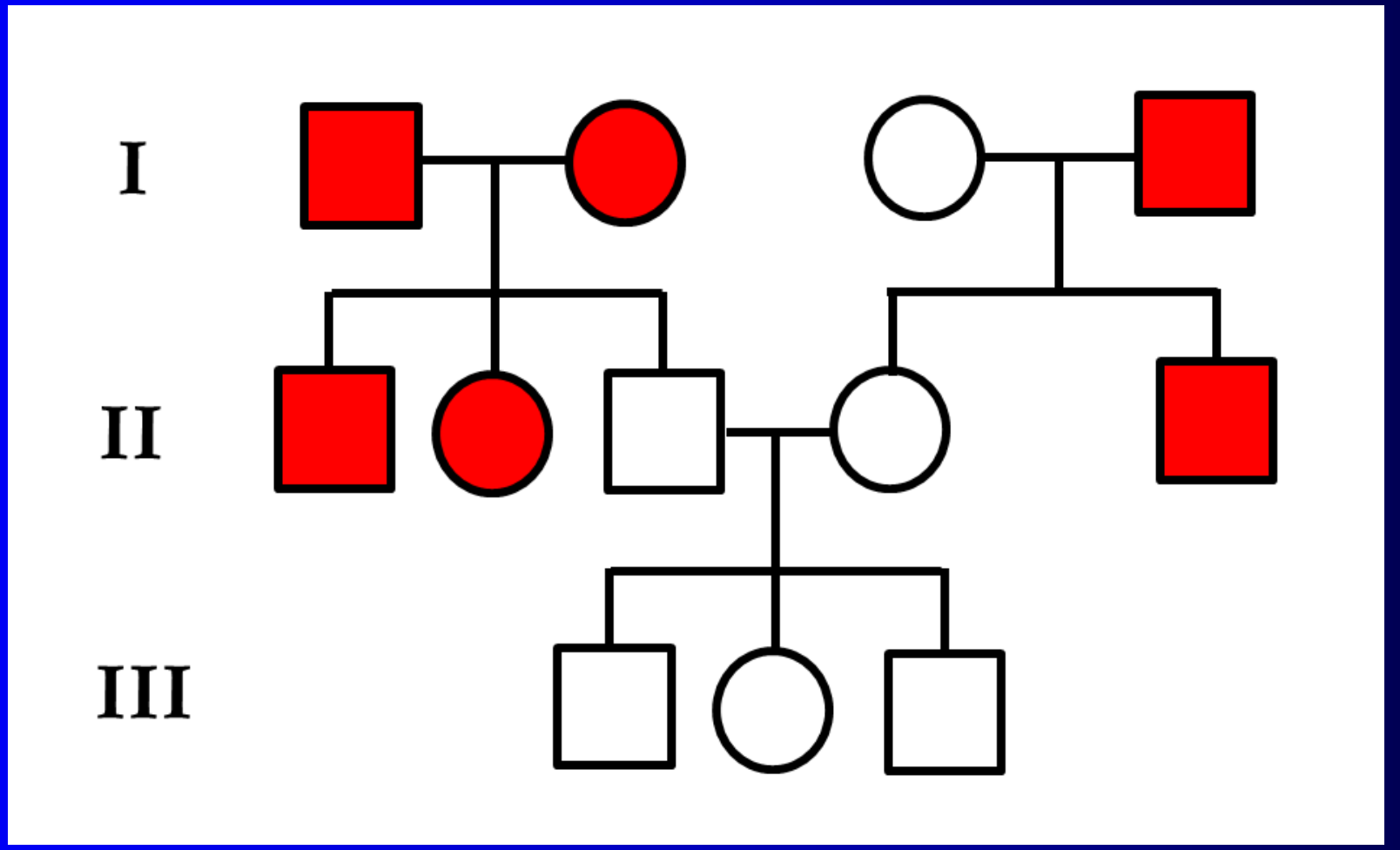
- Males and females are equally affected.
- It does not skip the generation.
- Affected parents can have unaffected children. (both affected father and mother)



Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek

B. John Ebenezer

Autosomal Dominant Trait

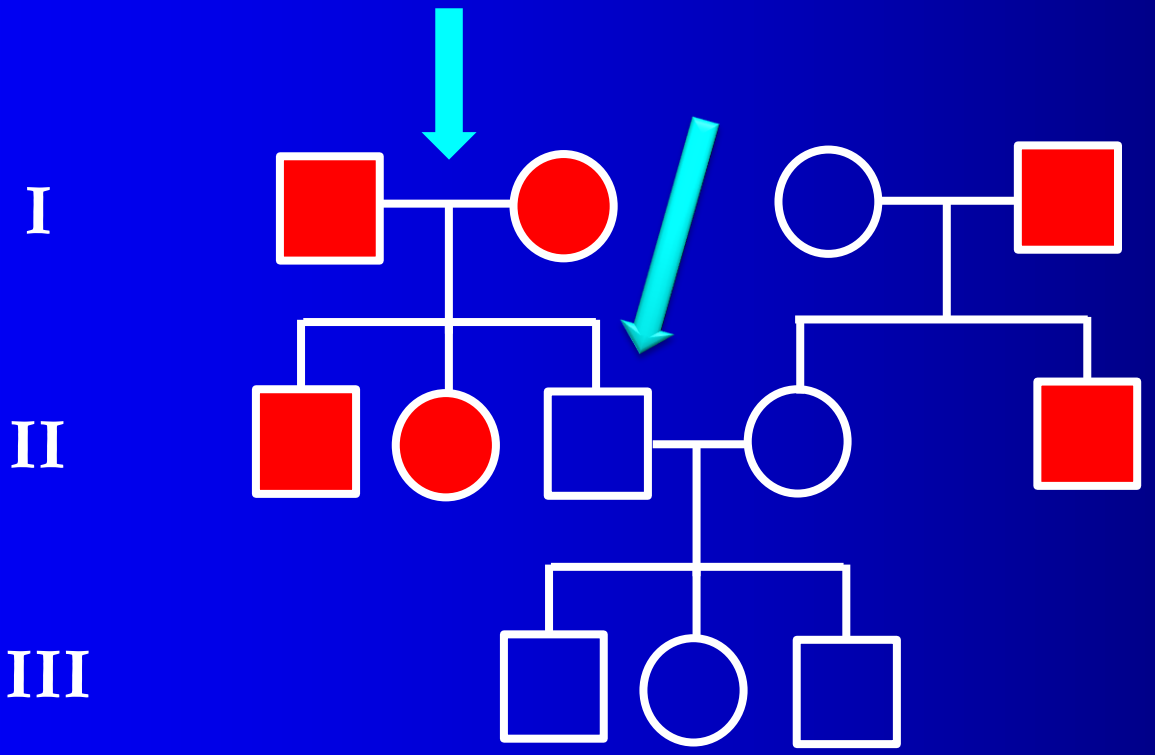


Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek

B. John Ebenezer

Autosomal Dominant Inheritance

Both the parents are affected but one of the sons is unaffected



Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek

B. John Ebenezer

Autosomal Dominant Inheritance

The gene is present in any one of the autosomes and the normal allele is recessive (a) to its mutant allele (A).

Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek.

B. John Ebenezer

Autosomal Dominant Trait

Autosomal dominant inheritance is due to mutation in Gene ‘a’

Genotype of affected individual: Aa or AA.

Genotype of unaffected or normal individual: aa

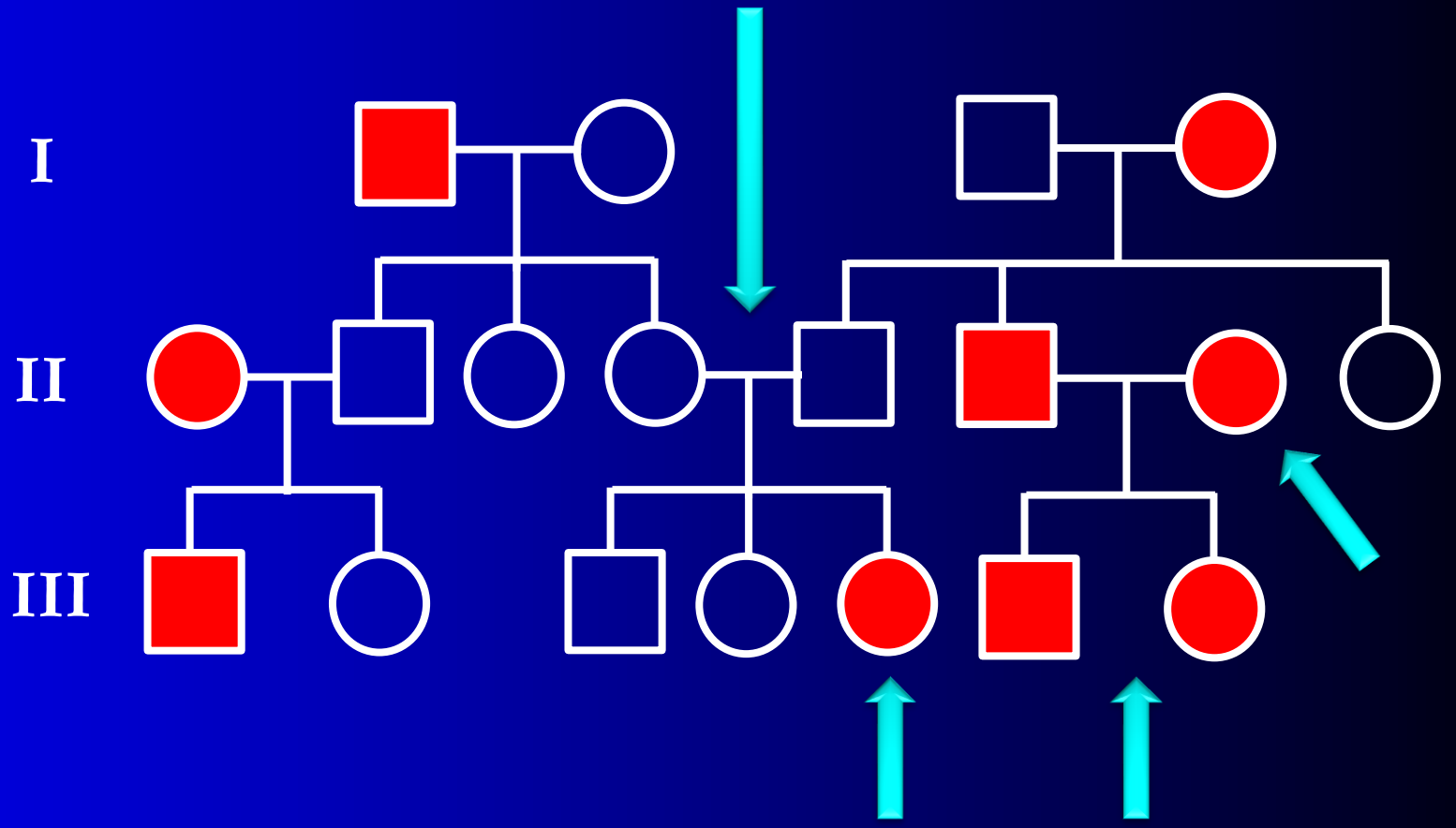
Affected offspring has at least one affected parent

B. John Ebenezer

Autosomal Recessive Inheritance

Autosomal Recessive Inheritance

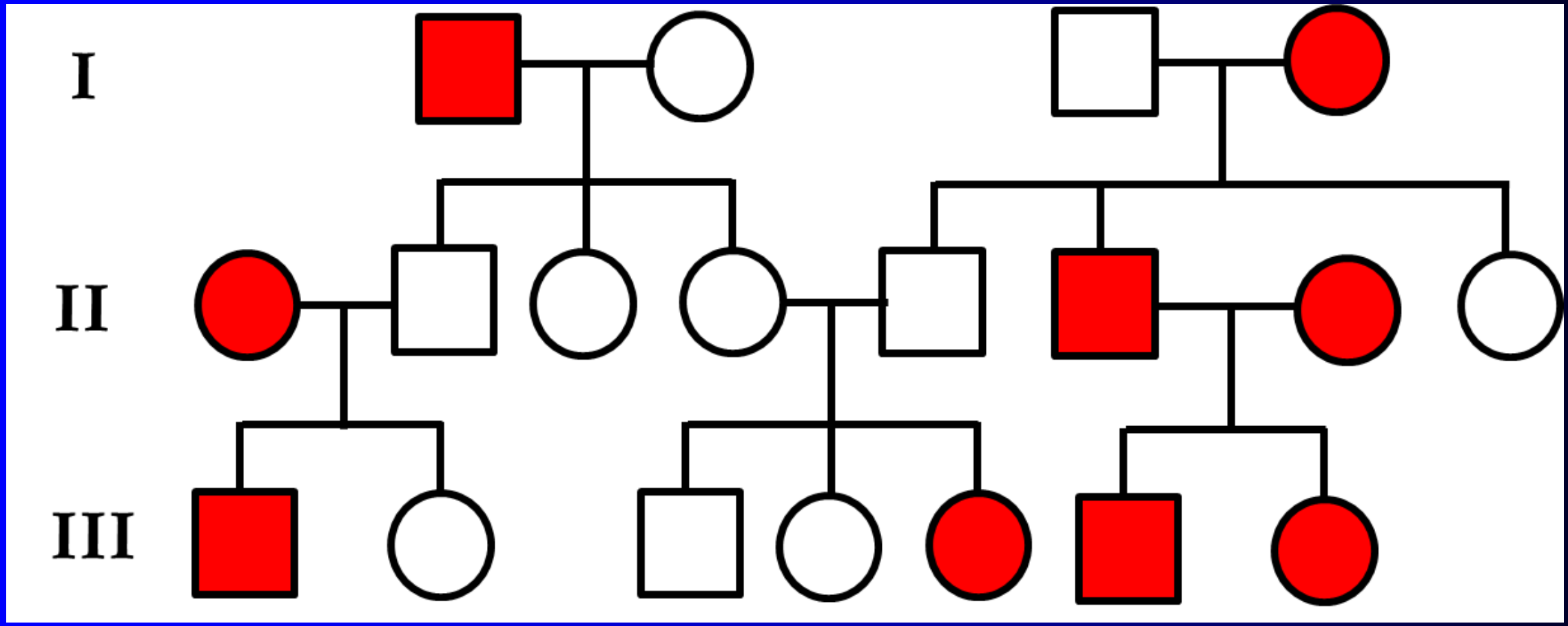
- Males and females are equally affected.
- Trait skips the generations.
- Both the parents are unaffected but the child is affected.
- If both the parents were affected, all the children would be affected.
- Traits are often found in pedigrees with consanguineous marriages.



Examples: Sickle Cell Anemia, Cystic Fibrosis, Phenylketonuria (PKU)

B. John Ebenezer

Autosomal Recessive Inheritance

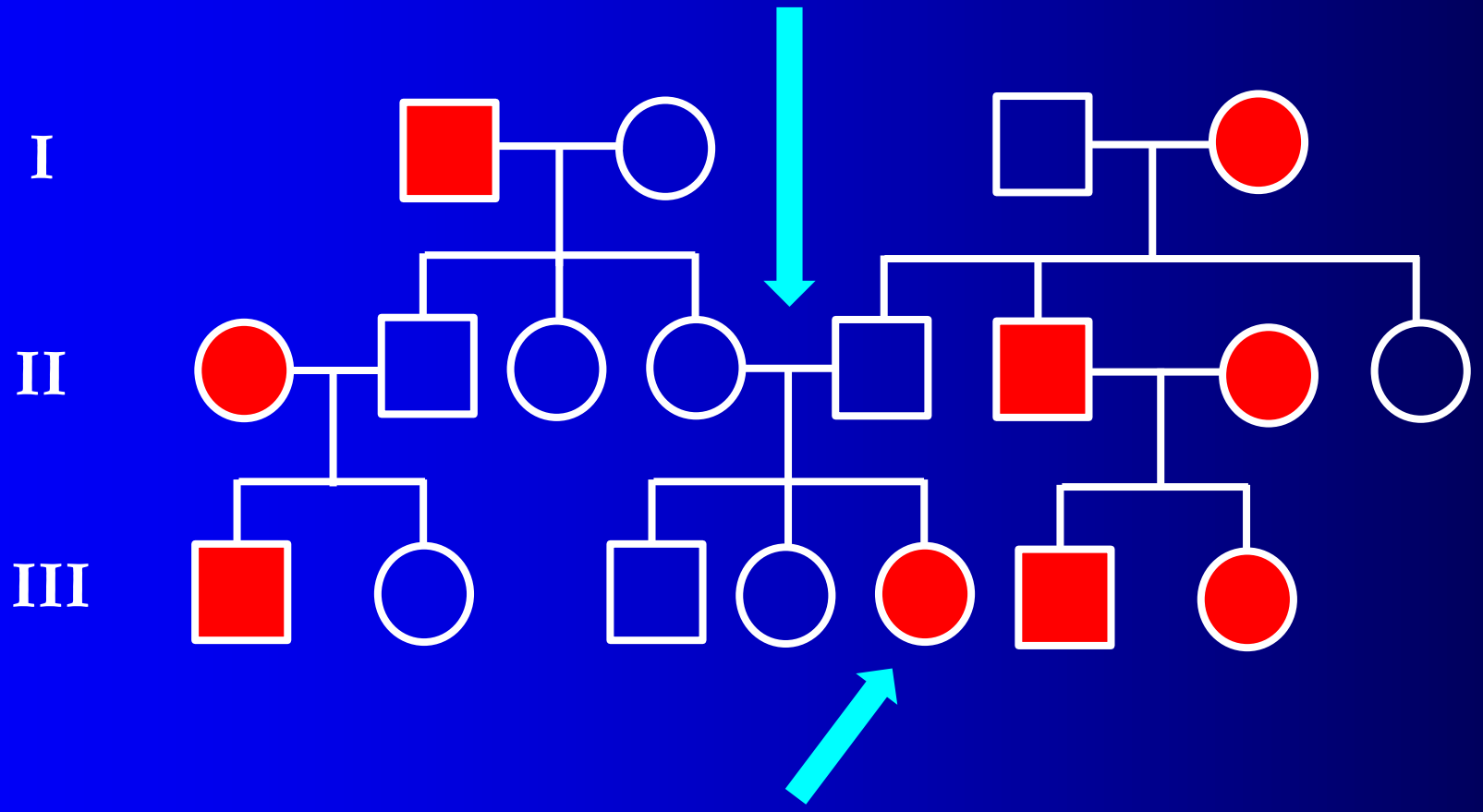


Examples: Sickle Cell Anemia, Cystic Fibrosis, Phenylketonuria (PKU)

B. John Ebenezer

Autosomal Recessive Inheritance

Both the parents are unaffected but the child is affected



B. John Ebenezer

Autosomal Recessive Trait

The mutant allele is recessive (a) to its wild type allele (A).

Genotype of affected individual: aa.

Genotype of unaffected or normal individual: AA or Aa

Affected offspring gets the disease causing allele from both parents

i.e. parents are at least heterozygous for the disease.

Examples: Sickle cell anemia, cystic fibrosis, phenylketonuria (PKU)

B. John Ebenezer

Autosomal Recessive Inheritance

When a normal person marries an affected individual, if all the children were normal, it indicates that the **normal parent is homozygously dominant.**

When a normal person marries an affected individual, if one or more of the children were affected, it indicates that the **normal parent is heterozygously dominant.**

Approximately half of the children are affected.

B. John Ebenezer

Sex Linked Dominant Inheritance

Sex Linked Dominant Inheritance

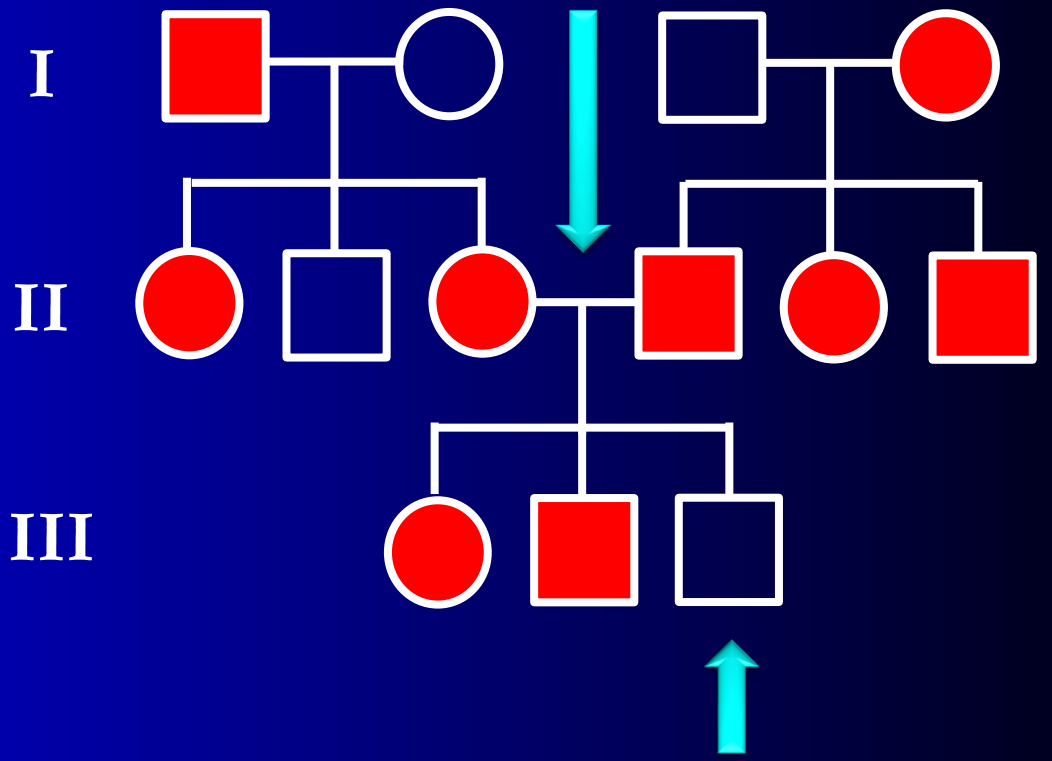
Females are more affected.

Trait never skips generations

Affected parents can have unaffected children.

All daughters of an affected father will be affected.

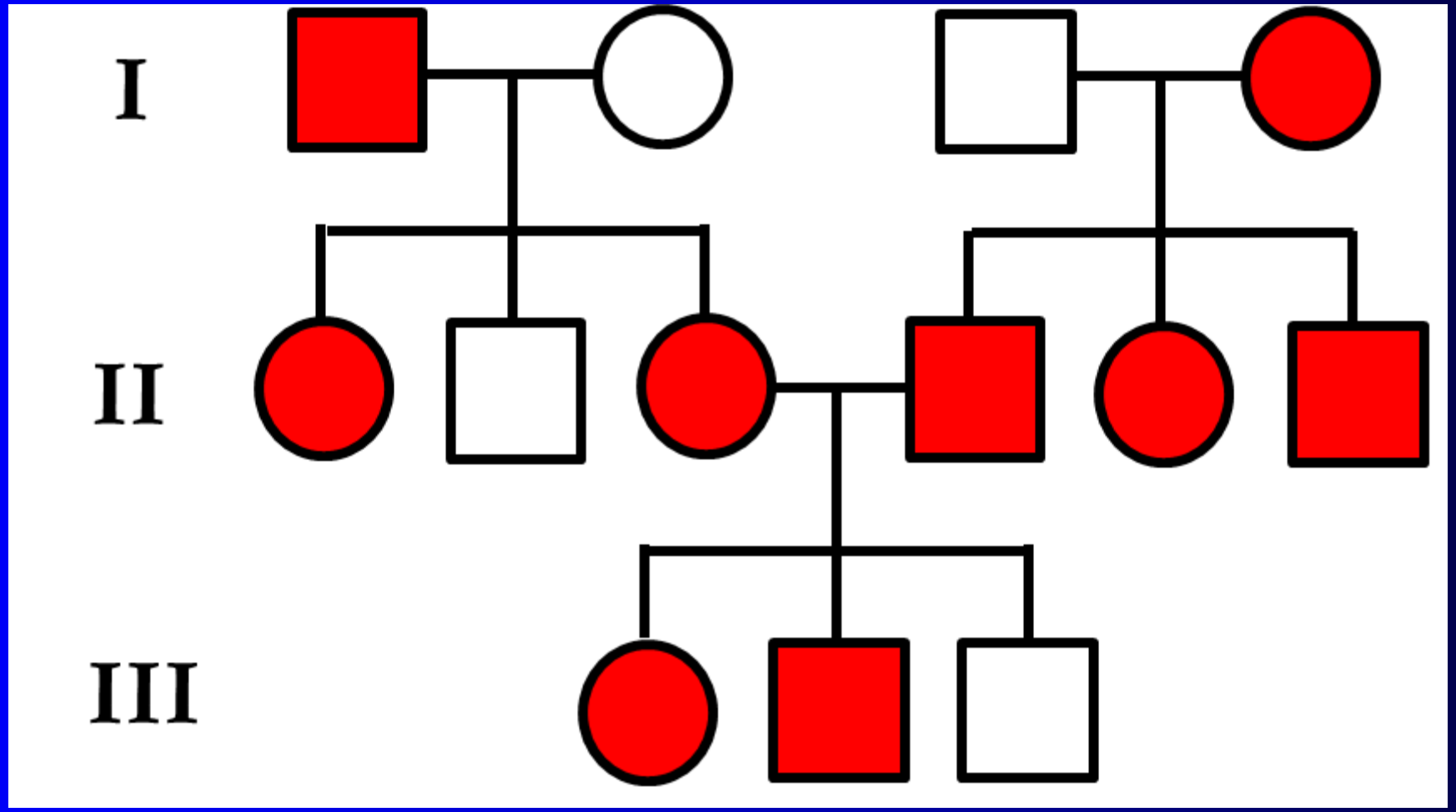
Disease transfers from father to daughter and from mother to both sons and daughters.



Example: Hypophatemic Rickets (Vitamin D Resistant Rickets)

B. John Ebenezer

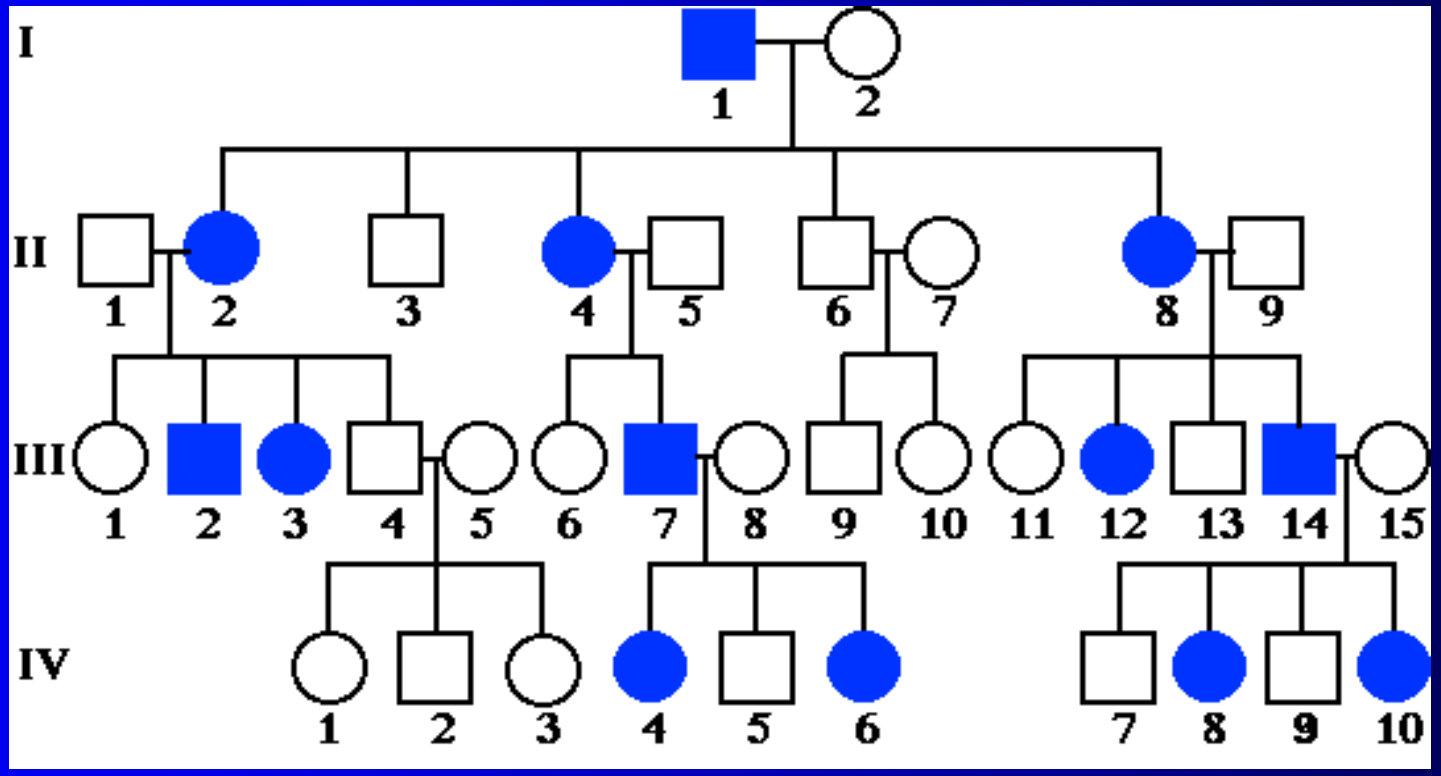
Sex Linked Dominant Inheritance



Example: Hypophatemic Rickets (Vitamin D Resistant Rickets)

B. John Ebenezer

Sex Linked Dominant Inheritance



B. John Ebenezer

Sex Linked Dominant Inheritance

For a male child to be affected the mother must be affected.

For a female child to be affected, either father or mother must be affected. (Any one of the parents must be affected)

Approximately half of the sons of an affected mother are affected, showing mother is heterozygous.

Example: Hypophatemic Rickets (Vitamin D Resistant Rickets)

B. John Ebenezer

Sex Linked Dominant Trait

Genotype of affected males: X^AY.

Genotype of affected females: X^AX^A or X^AX^a

Genotype of normal male: X^a Y.

Genotype of normal female: X^a X^a

Affected father transmits trait to all the daughters but not to sons.

Affected mother (if heterozygous) passes the trait to 50% of her daughters and 50% of her sons.

B. John Ebenezer

Sex Linked Recessive Inheritance

Sex Linked Recessive Inheritance

Male are more affected than females.

Trait skips generations

Disease transfers from

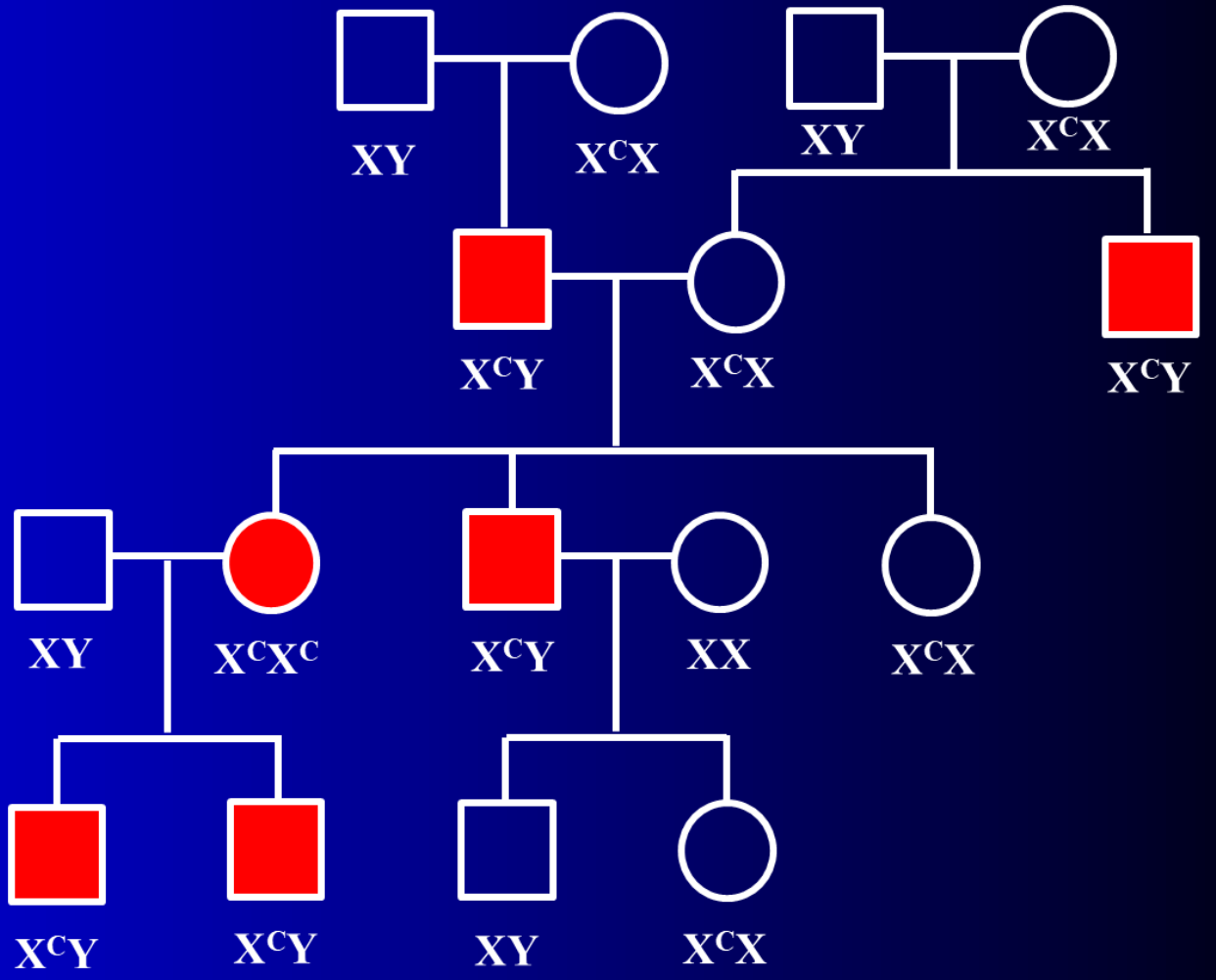
Mother to son and

Father to daughter. (crisscross inheritance)

All the sons of an affected mother will be affected. (Mother to sons)

Disease never transfers from father to son.

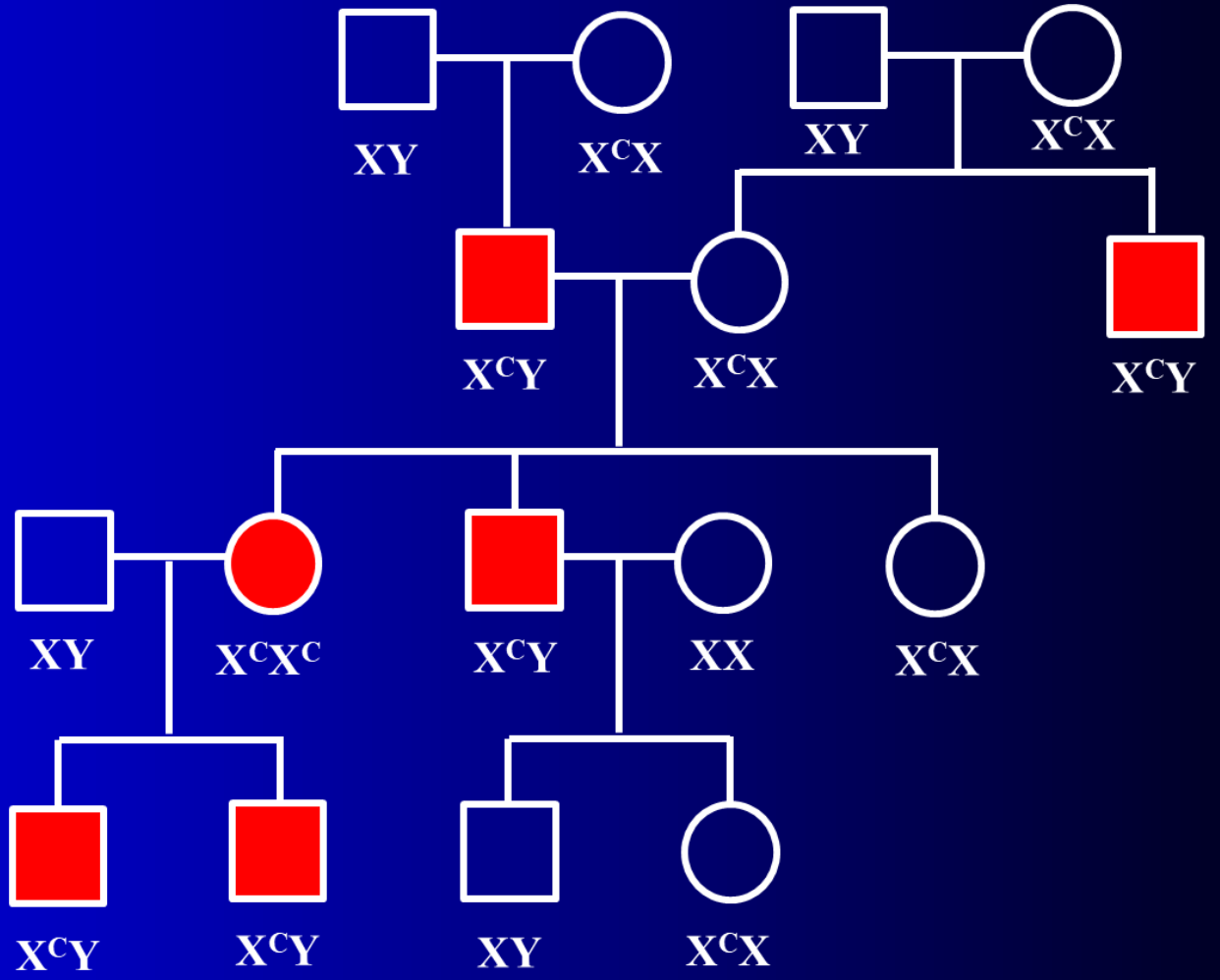
Examples: Hemophilia, Colour Blindness



Unaffected mother (carrier) will have affected sons

Sex Linked Recessive Trait

- This trait is rare in pedigree.
- Males are more often affected than females.
- Unaffected parents can have affected children.
- Affected father does not pass to his sons but passes to all daughters.
- Affected mother passes the trait to all her sons.



B. John Ebenezer

Sex Linked Recessive Trait

Genotype of affected males: $X^a Y$

Genotype of affected females: $X^a X^a$

Genotype of normal male: $X^A Y$

Genotype of normal female: $X^A X^a$ or $X^A X^A$

Affected mother transmits the trait to all the sons.

Carrier mother transmits the trait to 50% of her sons.

Examples: Hemophilia, Colour Blindness.

B. John Ebenezer

Sex Linked Recessive Inheritance

For a **male child** to be affected the **mother must be affected** or a carrier.

Usually affected sons are born to unaffected mother. (Carrier)

All the sons of an affected mother must be affected.

For a female child to be affected, the **father must be affected** and the **mother may be either affected or a carrier.**

Approximately half of the sons of **carrier mother are affected.**

B. John Ebenezer

Mendelian Disorders

Mendelian Disorder

The genetic disorder occurs in humans, caused due to alterations in one gene or in the genome is known as Mendelian Disorder.

Types of Mendelian disorders:

Autosomal dominant.
Autosomal recessive.
Sex-linked dominant.
Sex-linked recessive.

B. John Ebenezer

Some Examples of Mendelian Disorders

Mendelian Disorders

Autosomal Dominant

- Myotonic dystrophia
- Huntington's disease

Autosomal Recessive

- Sickle Cell Anemia
- Thalassemia
- Cystic Fibrosis
- Phenylketonuria (PKU)

X-Linked Dominant

- Hypophatemic Rickets (Vitamin D Resistant Rickets)

X-Linked Recessive

- Haemophilia
- Colour Blindness

B. John Ebenezer

Sickle Cell Anaemia

Sickle cell Anaemia - Autosomal Recessive Disorder

Sickle Cell Anaemia is an **Autosomal Recessive Disorder.**

Sickle cell disease is caused by a mutation in the hemoglobin-Beta gene found on **chromosome 11.**

The disease is controlled by a single pair of allele, HbA and HbS.

Out of the three possible genotypes only homozygous individuals for HbS (HbS HbS) show the diseased phenotype.

Heterozygous (HbA HbS) individuals appear unaffected but they are carriers of the disease.

B. John Ebenezer

Sickle cell Anaemia - Autosomal Recessive Disorder

Normal Hemoglobin is denoted as Hemoglobin-A.

Sickle Cell Hemoglobin is denoted as Hemoglobin-S.

There is 50 per cent probability for the transmission of the mutant gene to the progeny.

This disease can be transmitted from parents to the offspring when both the partners are carriers for the gene (or heterozygous).

B. John Ebenezer

Sickle cell Anaemia - Autosomal Recessive Disorder

The defect is caused due to the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule.

The substitution of amino acid in the globin protein is caused due to the single base substitution at the sixth codon of the beta globin gene from **GAG to GUG**.

The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.

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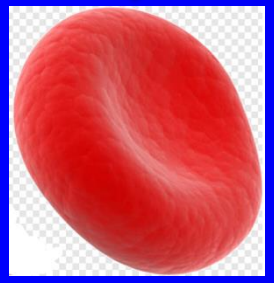
Sickle Cell Anaemia - Autosomal Recessive Disorder

SCA is controlled by two kinds of genes HbA, HbS

HbA HbA → Controls → 

Biconcave Disc Shaped RBC

Normal RBC

HbS HbS → Controls → 

Sickle Shaped RBC

Defective RBC

Heterozygous Genotypes

HbA HbS → Causes → **Unaffected**

Carrier Individual

Homozygous Genotypes

HbS HbS → Causes → **Affected**

Anaemic Individual

SCA is caused by mutation in the Hemoglobin-Beta gene found on **chromosome 11.**

Glutamic Acid → Substituted by → **Valine**

HbS Peptide

Leads to

Sickle Cell Anaemia

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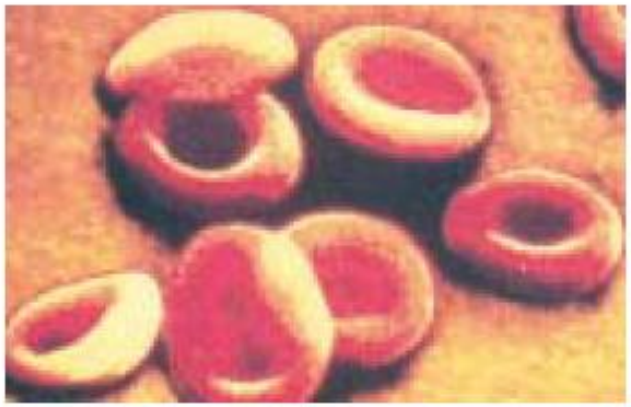
Sickle cell Anaemia - Autosomal Recessive Disorder



Normal Hb (A)gene ---CTC---
---GAG---

mRNA ---GAG---

Val 1 — His 2 — Leu 3 — Thr 4 — Pro 5 — Glu 6 — Glu 7

HbA peptide

Sickle-cell Hb(S) gene ---CAC---
---GTG---

mRNA ---GUG---

Val 1 — His 2 — Leu 3 — Thr 4 — Pro 5 — Val 6 — Glu 7

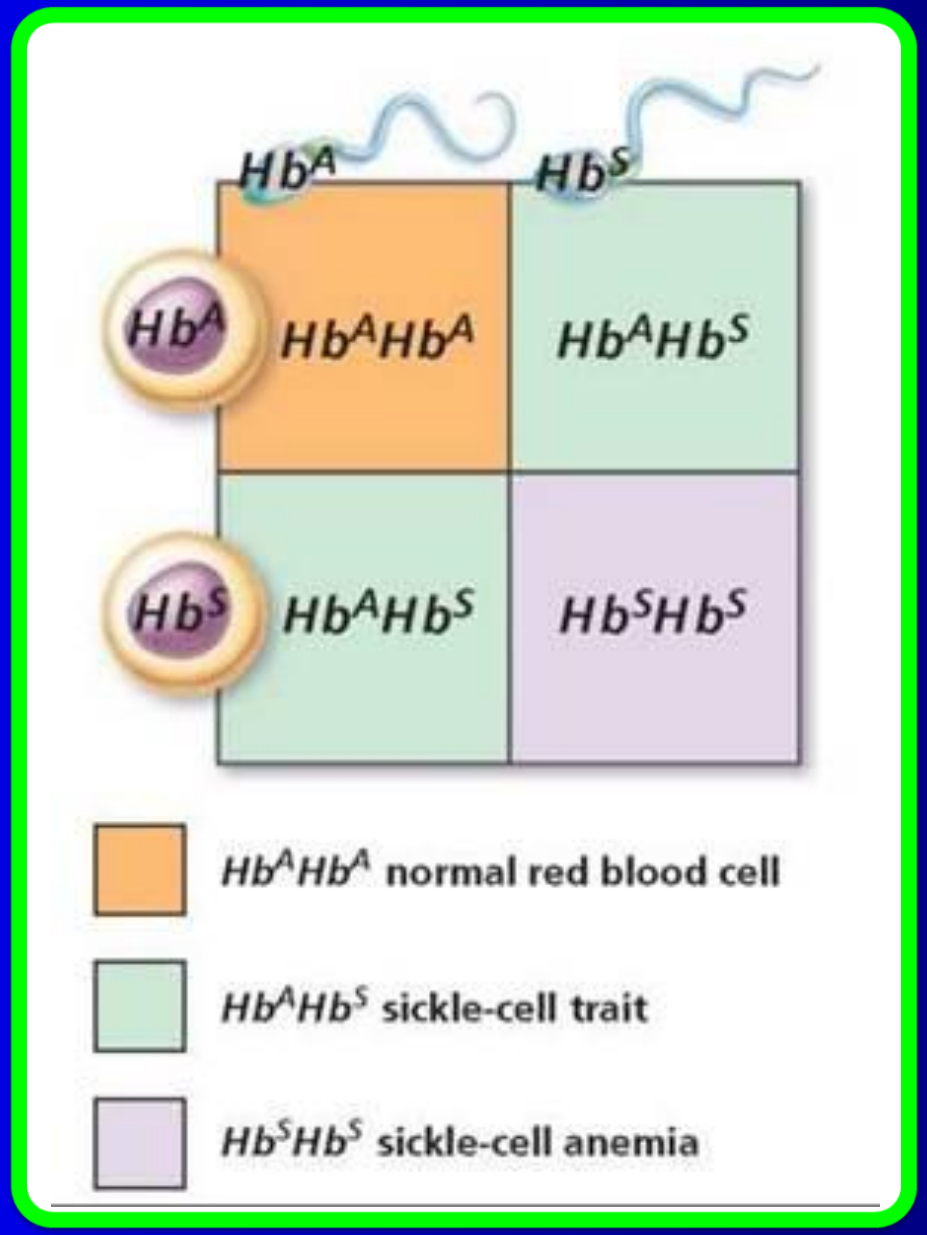
HbS peptide

B. John Ebenezer

Sickle Cell RBC and Normal RBC

B. John Ebenezer

Sickle cell Anaemia - Autosomal Recessive Disorder



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Thalassemia

Thalassemia - Autosomal Recessive Disorder

Thalassemia is an **Autosomal Recessive Disorder.**

The disorder results in **large numbers of red blood cells being destroyed,** which leads to anemia.

Thalassemia occurs when there is a defect in a gene that controls the production of alpha or beta chains of haemoglobin.

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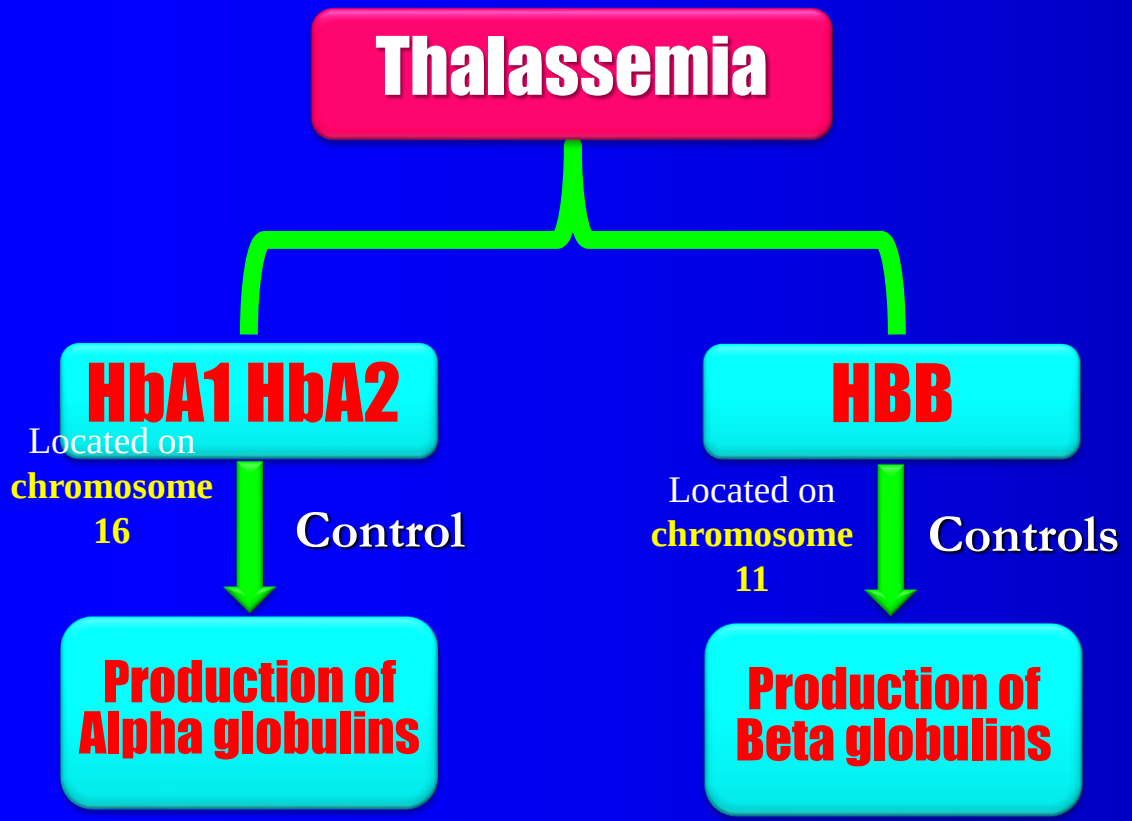
Thalassemia - Autosomal Recessive Disorder



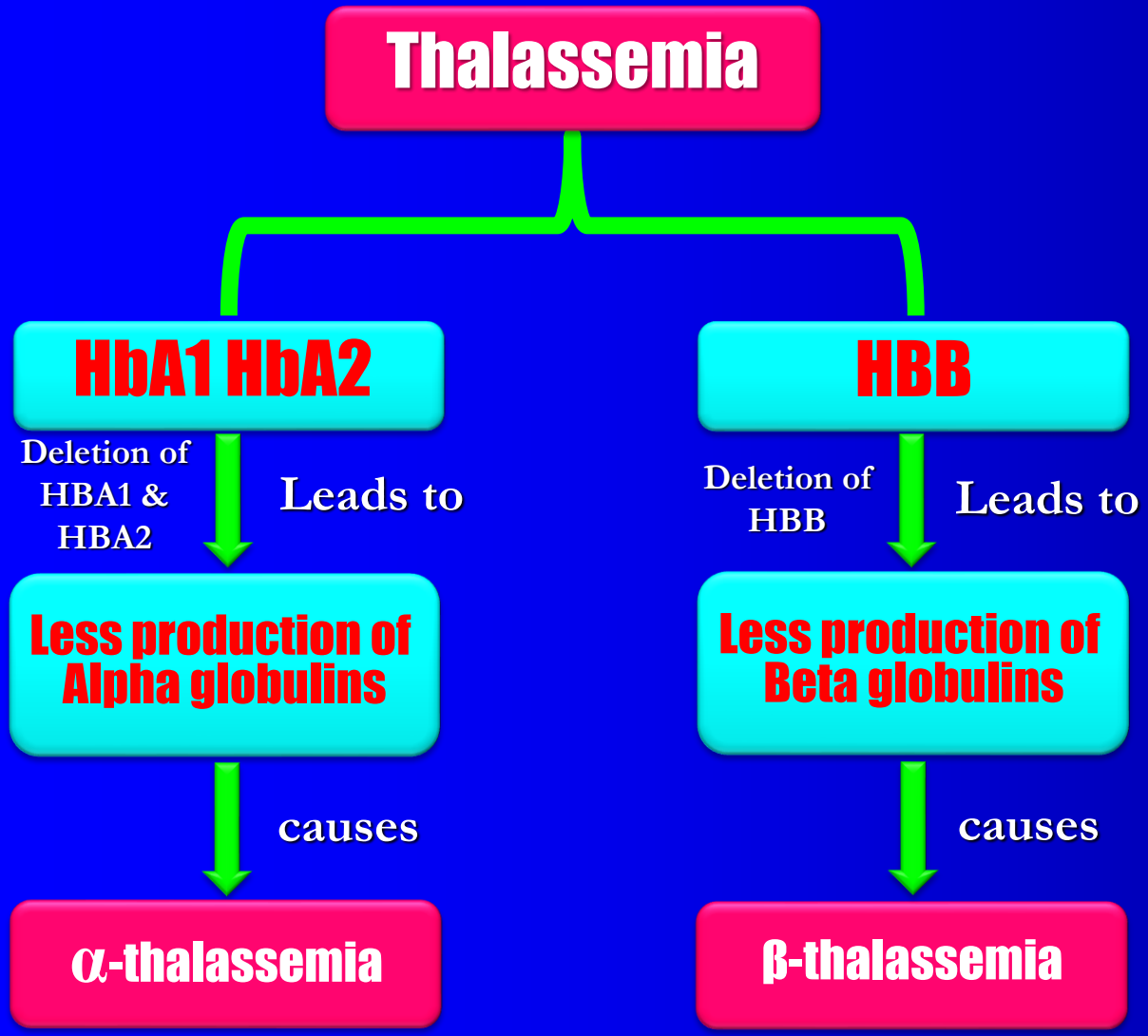
Adult hemoglobin is composed of two alpha (α) and two beta (β) polypeptide chains.

There are two copies of the hemoglobin alpha gene (*HBA1* and *HBA2*), which each encode an α-chain, are located on **chromosome 16.**

The hemoglobin beta gene (*HBB*) encodes the β-chain is located on **chromosome 11.**

B. John Ebenezer

Thalassemia - Autosomal Recessive Disorder



Deletions of genes *HBA1* and/or *HBA2* leads to less production of alpha globulins and causes α-thalassemia.

Deletions of *HBB* leads to less production of beta globulins and causes β-thalassemia.

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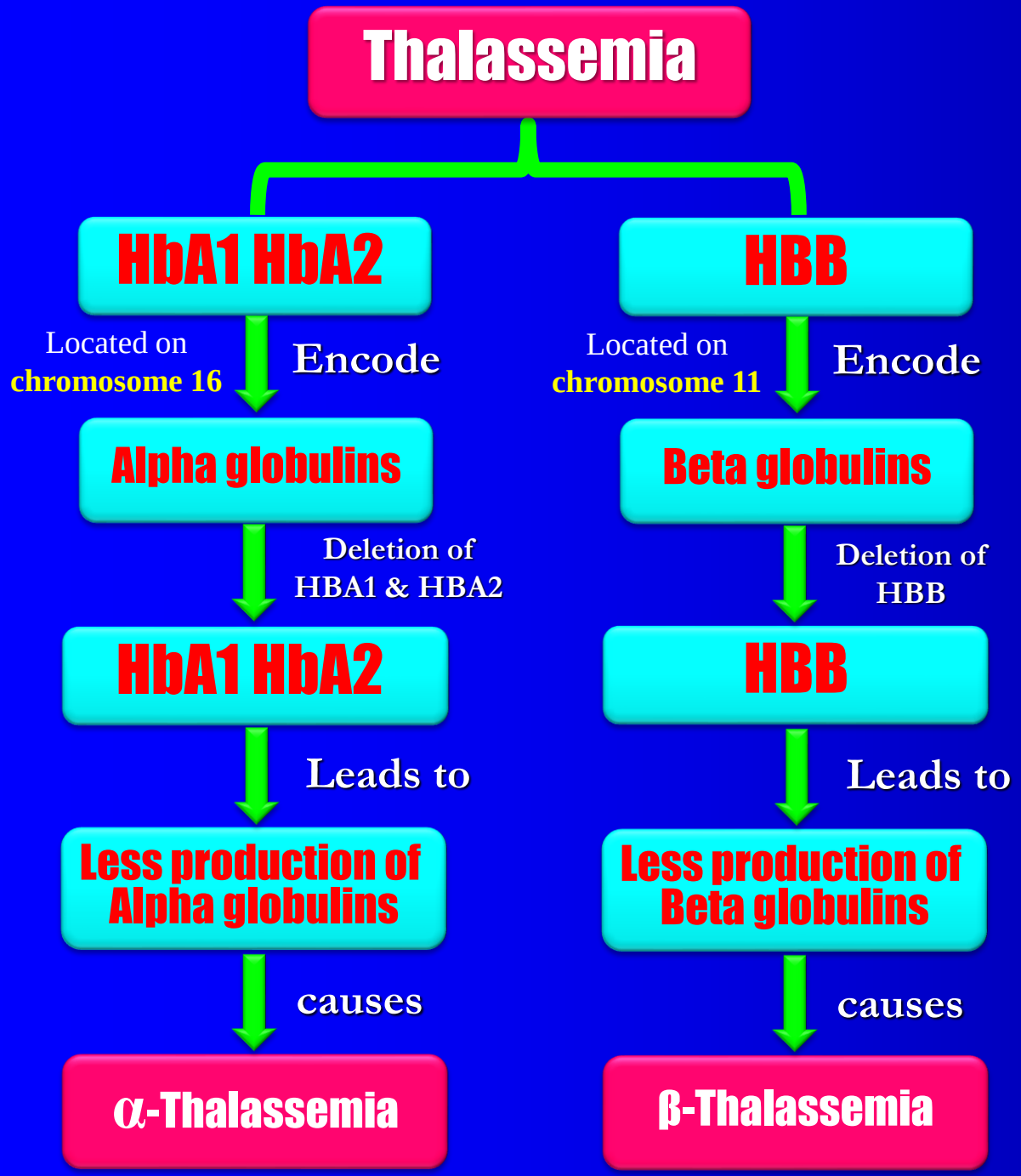

Thalassemia - Autosomal Recessive Disorder

Alpha thalassemia occurs when a gene related to the alpha globin protein is missing or mutated.

Beta thalassemia occurs when a gene defects affect the production of the beta globin protein.

Symptoms:

Facial bone deformities, abdominal swelling, dark urine.

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Thalassemia - Autosomal Recessive Disorder

Sickle-cell anaemia is a qualitative problem of synthesising abnormal haemoglobin.

Thalassemia is a quantitative problem of synthesising too few globin molecules.

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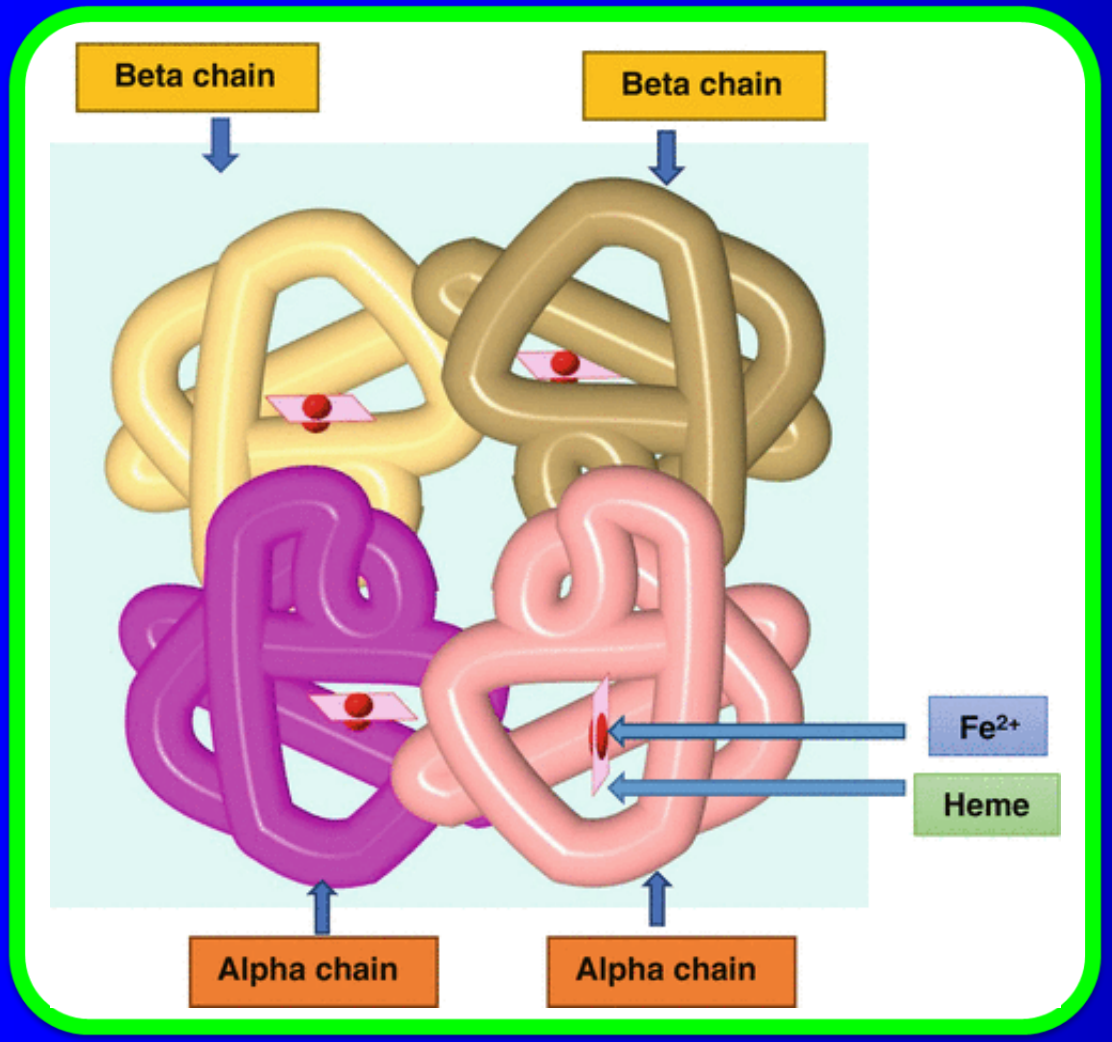
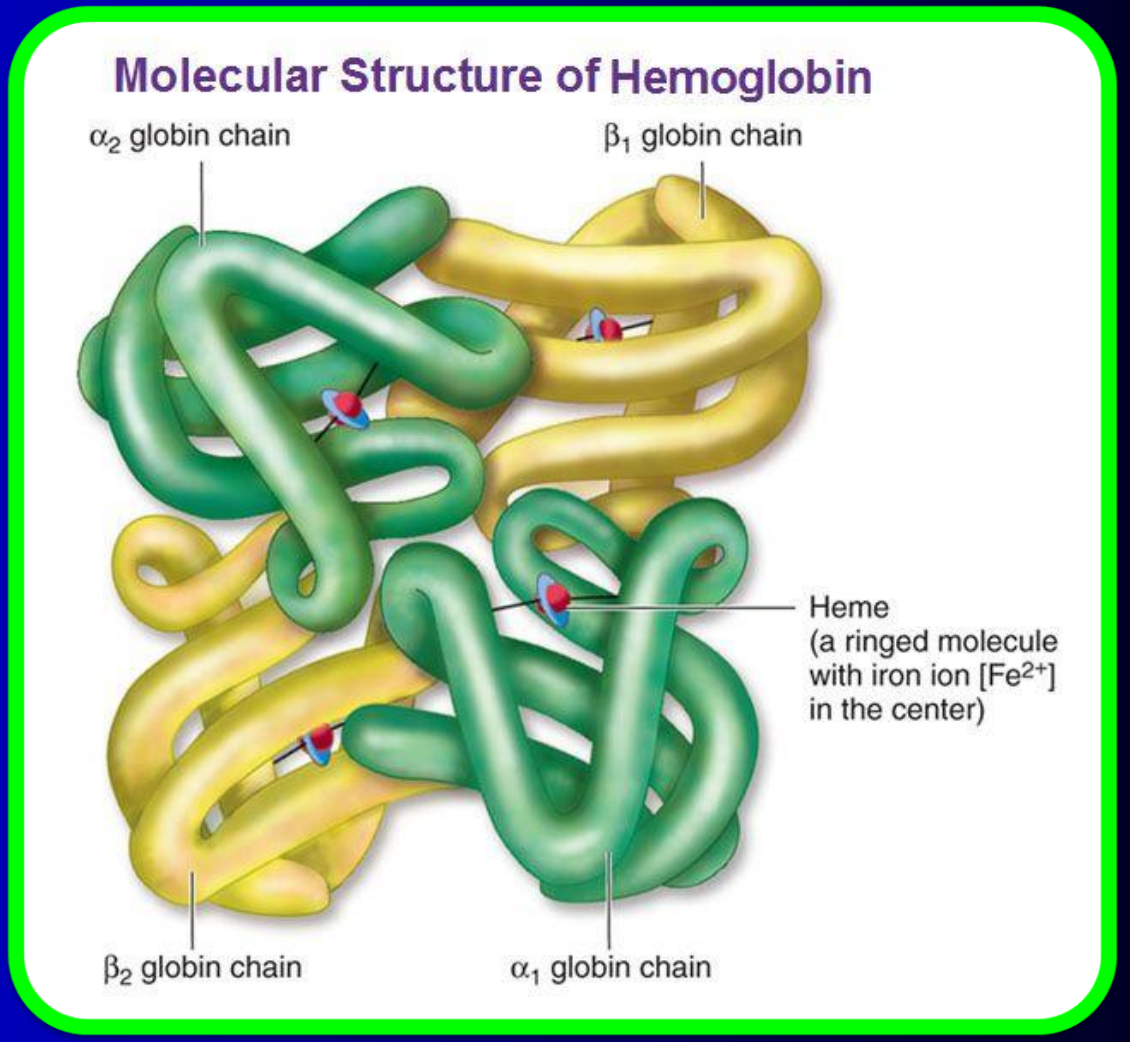
Thalassemia - Autosomal Recessive Disorder





B. John Ebenezer

Phenylketonuria

Phenylketonuria - Autosomal Recessive Disorder

Phenylketonuria is an **Autosomal Recessive Disorder** leads to an inborn error of metabolism.

It is caused due to **mutation** in the gene present on **chromosome 12** that codes for the enzyme **phenyl alanine hydroxylase.**

The affected individual lacks the enzyme **phenyl alanine hydroxylase** that converts the amino acid phenylalanine into tyrosine.

As a result of this phenylalanine is accumulated and converted into **phenylpyruvic acid** and **other derivatives**.

B. John Ebenezer

Phenylketonuria - Autosomal Recessive Disorder

Gene (PAH)

Located on **Chromosome-12**

Controls Production of

Phenylalanine hydroxylase

Phenylalanine —(**Phenylalanine Hydroxylase** converts)→ **Tyrosine**

Phenylalanine

In the absence of enzyme Converted to

Phenyl Pyruvic Acid + **Other Derivatives**

These accumulate in the

Brain

Leads to

Mental Retardation

B. John Ebenezer

Pleiotropy - Phenylketonuria

Accumulation of **phenylalanine, phenylpyruvic acid** and **other derivatives** in brain results in mental retardation.

These are also excreted through urine because of its poor absorption by kidney.

Symptoms:

Mental retardation.

Reduction in hairs.

Reduction in skin pigmentation.

B. John Ebenezer

Cystic Fibrosis

Cystic Fibrosis - Autosomal Recessive Disorder

Cystic Fibrosis is an **Autosomal Recessive Disorder.**

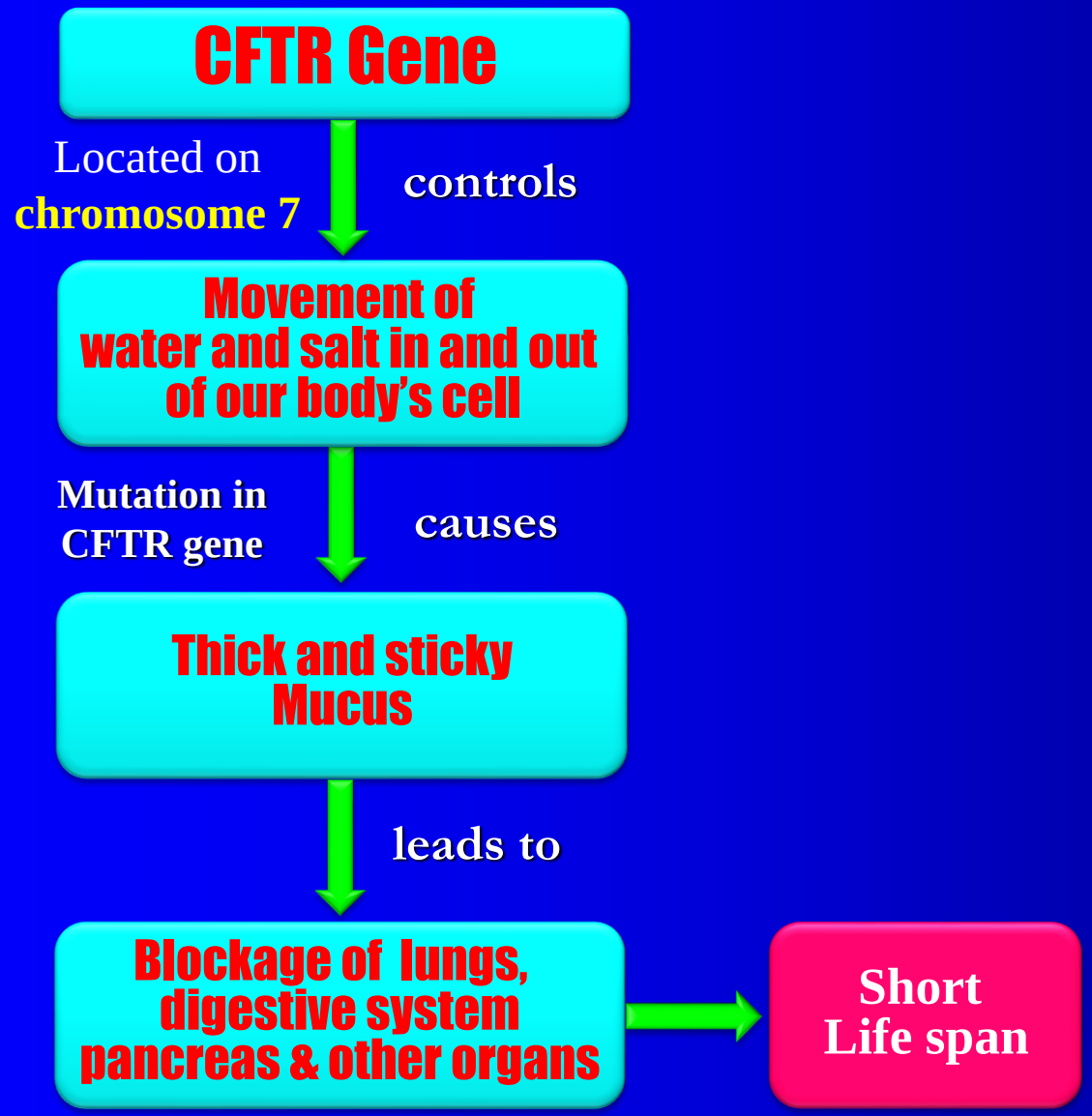
It is caused due to **mutation of a gene** that controls the **movement of water and salt** in and out of the cells.

This disease affects the **lungs and the digestive system** and the body produces **thick and sticky mucus** that blocks the lungs and pancreas.

People suffering from this disorder have a very short life-span.

B. John Ebenezer

Cystic Fibrosis - Autosomal Recssive Disorder



Cystic Fibrosis is an **Autosomal Recessive Disorder.**

It is caused due to **mutation of a cystic fibrosis transmembrane conductance regulator** gene or CFTR gene controls the **movement of water and salt** in and out of the cells.

This disease affects the **lungs and the digestive system** and the body produces **thick and sticky mucus** that blocks the lungs and pancreas.

People suffering from this disorder have a very short life-span.

B. John Ebenezer

Cystic Fibrosis - Autosomal Recessive Disorder

B. John Ebenezer

Haemophilia

Haemophilia - Sex Linked Recessive Disorder

Haemophilia is a **Sex Linked Recessive Disorder.**

This disease is caused due to a **single defective protein** that is a part of the cascade of proteins involved in the clotting of blood.

This results in **non-stop bleeding** even if a small cut occurs in the affected individuals.

B. John Ebenezer

Haemophilia - Sex Linked Recessive Disorder

Blood clotting is controlled by F8 and F9 genes located on X chromosome.

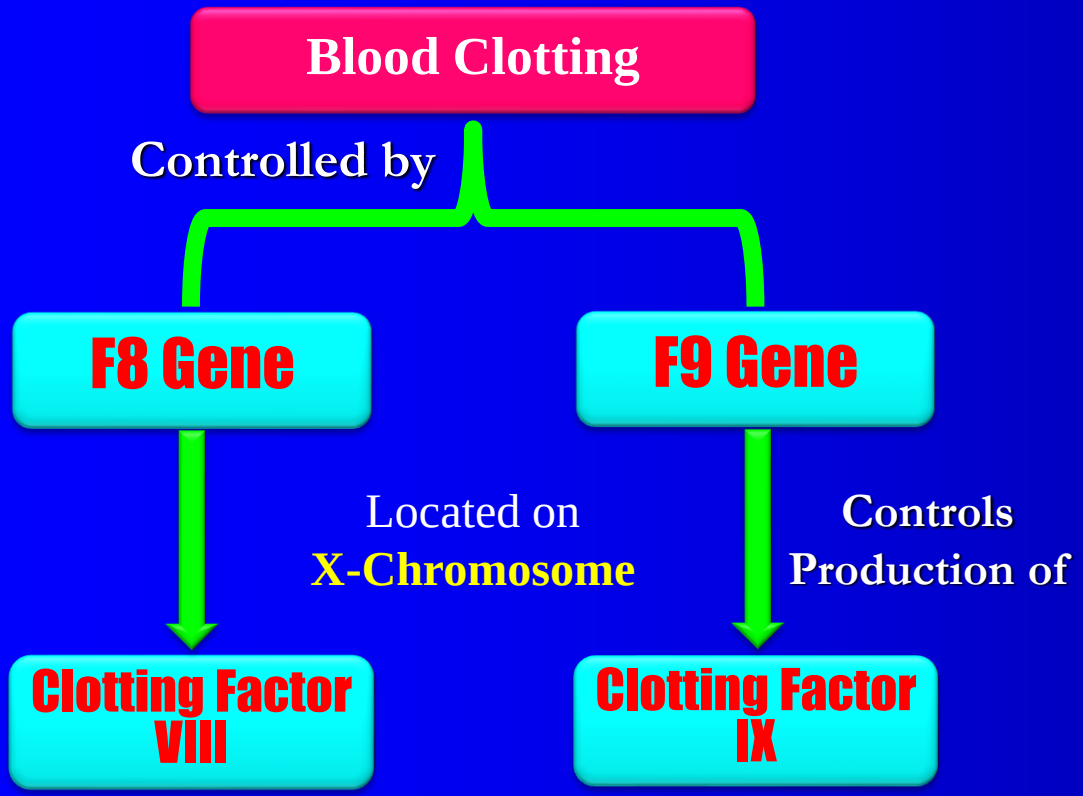
The F8 gene produces blood clotting factor VIII and the F9 gene produces blood clotting factor IX.

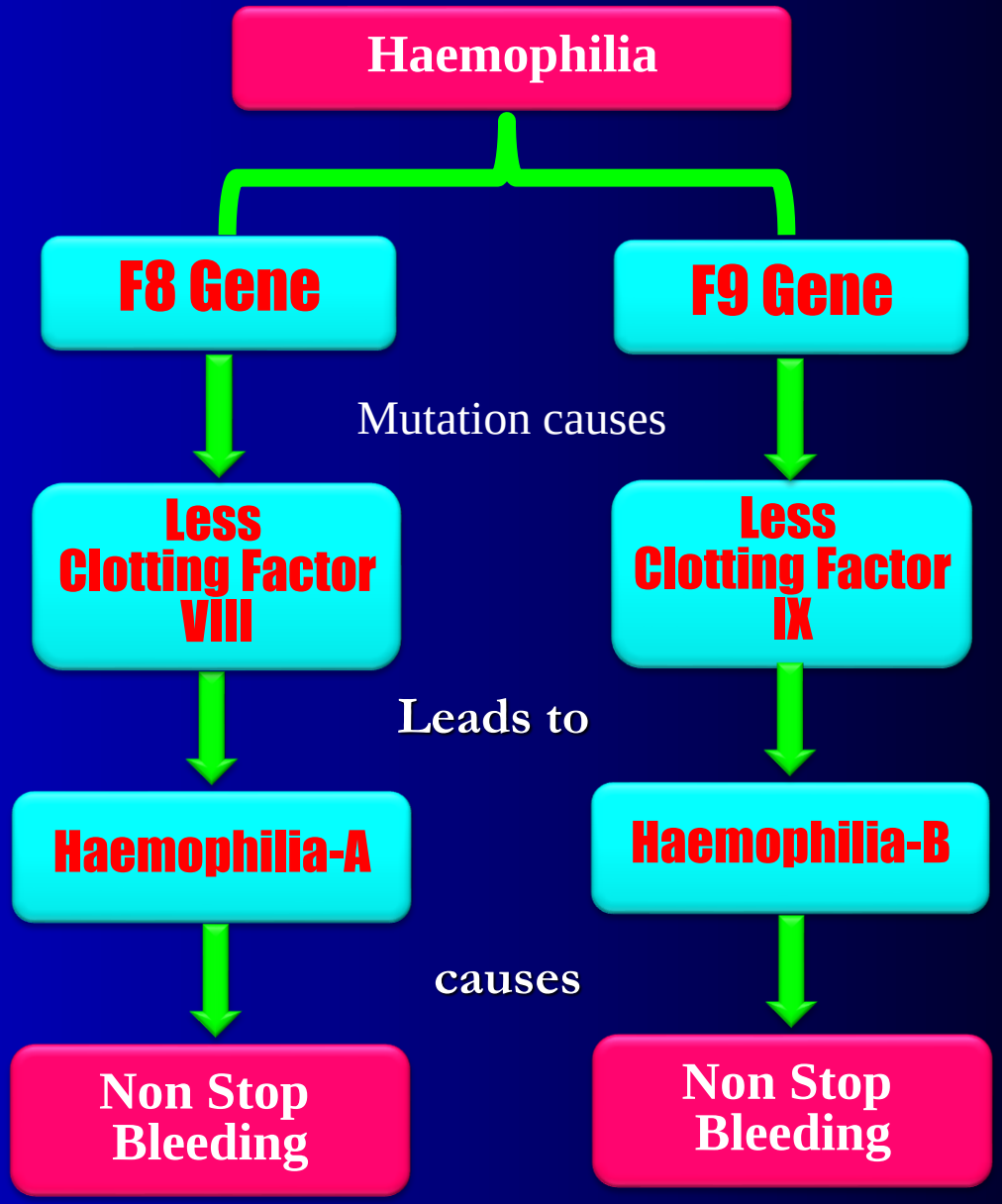
They cause blood clotting during injury.

Mutation in these genes affect the production of blood clotting factors, leading to haemophilia.

B. John Ebenezer

Haemophilia
Sex Linked Recessive Disorder





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Haemophilia - Sex Linked Recessive Disorder

It transmits from **unaffected carrier mother to the sons**.

The possibility of a female becoming a haemophilic is extremely rare because mother should be at least a carrier and the father should be haemophilic.

The family pedigree of Queen Victoria shows a number of haemophilic descendents as she was a carrier of the disease.

B. John Ebenezer

Haemophilia - Sex Linked Recessive Disorder

Normal Male \ Carrier Female	X^H	X^h
X^H	X^HX^H Normal Female	X^HX^h Carrier Female
Y	X^HY Normal Male	X^hY Haemophilic Male

Normal Female 25%

Carrier Female 25%

Normal Male 25%

Haemophilic Male 25%

B. John Ebenezer

Colour Blindness

Colour Blindness - Sex Linked Recessive Disorder

The gene OPN1 located on **X chromosome** control the production of cone pigments and cause colour vision of eyes.

Mutation in OPN1 gene **affect the production of cone pigments**, leading to colour blindness.

B. John Ebenezer

Colour Blindness
Sex Linked Recessive Disorder

Control of Production of Pigments of Cone Cells

Gene (OPN1)

Located on X-Chromosome

Controls Production of

Pigments of Cone cells

Leads to

Colour Vision of Eyes

Cause of Colur Blindness

Colour Blindness

Gene (OPN1)

Mutation causes

No production of Pigments

Leads to

Colour Blindness

Could not see Red & Green colours

B. John Ebenezer

Colour Blindness - Sex Linked Recessive Disorder

Colour blindness is a **Sex Linked Recessive Disorder** caused due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour.

This defect is due to mutation in certain genes present in the X chromosome.

It occurs in about **8 per cent of males** and only about **0.4 per cent of females.**

B. John Ebenezer

Colour Blindness - Sex Linked Recessive Disorder

This is because the genes that lead to red-green colour blindness are on the X chromosome.

Males have only one X chromosome and females have two.

There is 50% possibility for the son of a carrier mother being colour blind.

B. John Ebenezer

Colour Blindness - Sex Linked Recessive Disorder

The mother is not herself colourblind because the gene is recessive.

That means that its effect is suppressed by her matching dominant normal gene.

A daughter will not normally be colour blind, unless her mother is a carrier and her father is colour blind.

B. John Ebenezer

Colour Blindness - Sex Linked Recessive Disorder

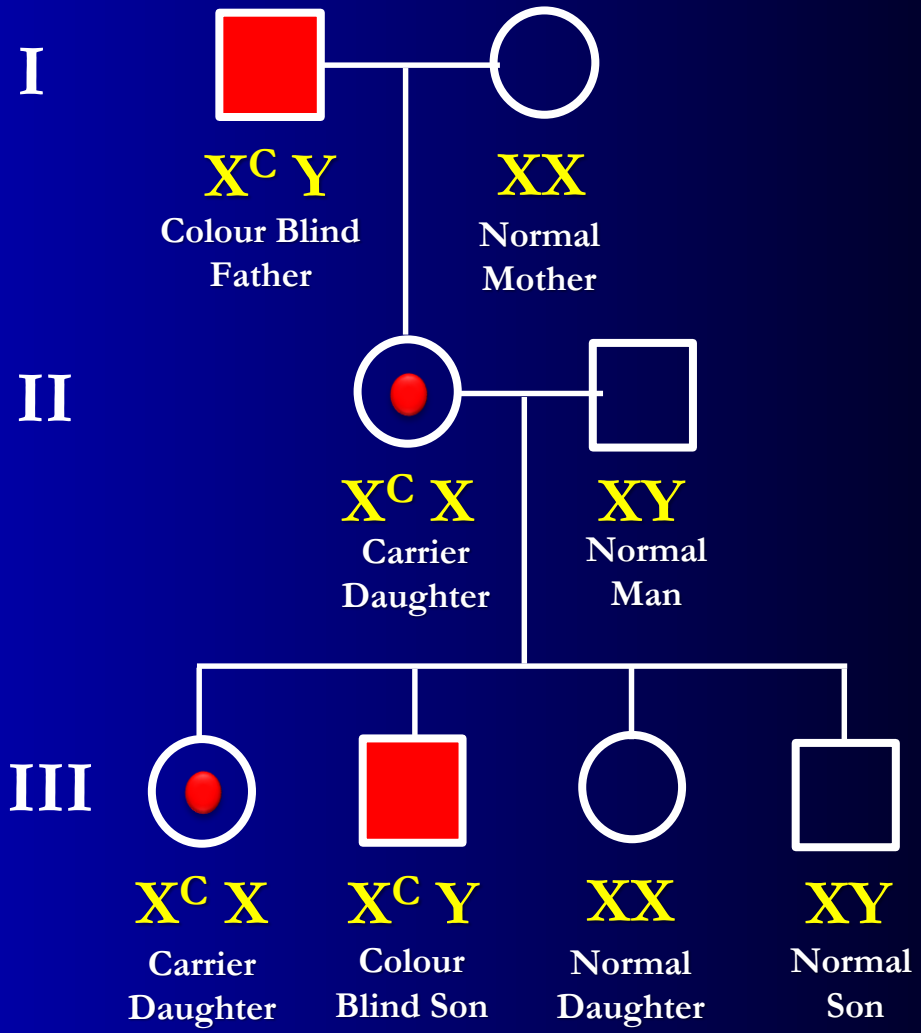
A normal visioned woman, whose father is colour blind, marries a normal visioned man. What would be the probability of her sons and daughters to be colour blind? Explain with the help of a pedigree chart.

B. John Ebenezer

Colour Blindness

Normal Male \ Carrier Female	X^C	X
X	X^CX Carrier Female	XX Normal Female
Y	X^CY Colour blind Male	XY Normal Male

Normal Female 50%
Normal Male 25%
Colour blind Male 25%



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Colour Blind Male and Normal Female

Colour blind Male	Normal Female: X	Normal Female: X
X^C	X^CX Carrier Female	X^CX Carrier Female
Y	XY Normal Male	XY Normal Male

Carrier Female 50%

Normal Male 50%

B. John Ebenezer

Colour Blind Male and Carrier Female

Colour blind Male \ Carrier Female	X^C	X
X^C	X^CX^C Colour blind Female	X^CX Carrier Female
Y	X^CY Colour blind Male	XY Normal Male

Colour blind Female 25%

Carrier Females 25%

Colour blind Male 25%

Normal Male 25%

B. John Ebenezer

Normal Male and Carrier Female

Normal Male \ Carrier Female	X^C	X
X	X^CX Carrier Female	XX Normal Female
Y	X^CY Colour blind Male	XY Normal Male

Carrier Female 25%

Normal Female 25%

Colour blind Male 25%

Normal Male 25%

OR

Normal Female 50%

Normal Male 25%

Colour blind Male 25%

B. John Ebenezer

Colour Blind Male and Colour Blind Female

Colour blind Male \ Colour blind Female	X^C	X^C
X^C	X^CX^C Colour blind Female	X^CX^C Colour blind Female
Y	X^CY Colour blind Male	X^CY Colour blind Male

Colour blind female 50%

Colour blind male 50%

B. John Ebenezer

Colour Blindness

Colour blindness is the inability of an individual to differentiate between red and green colours.

Genotype of colour blindness:

Colour blindness is a recessive X-linked trait.

Males have only one X chromosome and show colour blindness due to the presence of a single recessive allele ($X^{C}Y$).

B. John Ebenezer

Colour Blindness

Females have 2X chromosomes.

They are affected by colour blindness only in homozygous condition.

Females are affected only when father is affected and mother is at least a carrier (Only when both the X chromosomes carry the recessive alleles X^CX^C).

Females act as carriers in heterozygous condition (X^CX).

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Colour Blindness

Among the sons 50% of sons will be colour blind and 50% of sons will be normal visioned.

Among the daughters 50% of daughters will be normal visioned and 50% of daughters will be carrier.

B. John Ebenezer

Chromosomal Disorder

Chromosomal Disorder

The chromosomal disorders are caused due to absence or excess or abnormal arrangement of one or more chromosomes.

Failure of segregation of chromatids during cell division results in the gain or loss of a chromosome, called **aneuploidy**.

Down's syndrome results in the gain of extra copy of chromosome 21.

B. John Ebenezer

Chromosomal Disorder

Turner's syndrome is caused due to loss of an X chromosome in human females.

Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism and, this phenomenon is known as **polyploidy**.

This condition is often seen in plants.

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Chromosomal Disorder

The total number of chromosome of a normal human being is 46 (23 pairs).

Out of these 22 pairs are autosomes and one pair of chromosomes are sex chromosome.

Sometimes either an additional copy of a chromosome is included in an individual or an individual may lack one of any one pair of chromosomes.

B. John Ebenezer

Chromosomal Disorder

These situations are known as trisomy or monosomy of a chromosome, respectively.

Such a situation leads to very serious consequences in the individual.

Down's syndrome, Turner's syndrome, Klinefelter's syndrome are common examples of chromosomal disorders.

B. John Ebenezer

Chromosomal Disorder

Monosomy:

Deletion of a chromosome from a pair of chromosomes is known as monosomy. (2n-1)

Trisomy:

Addition of a chromosome to a pair of chromosomes is known as trisomy (2n+1).

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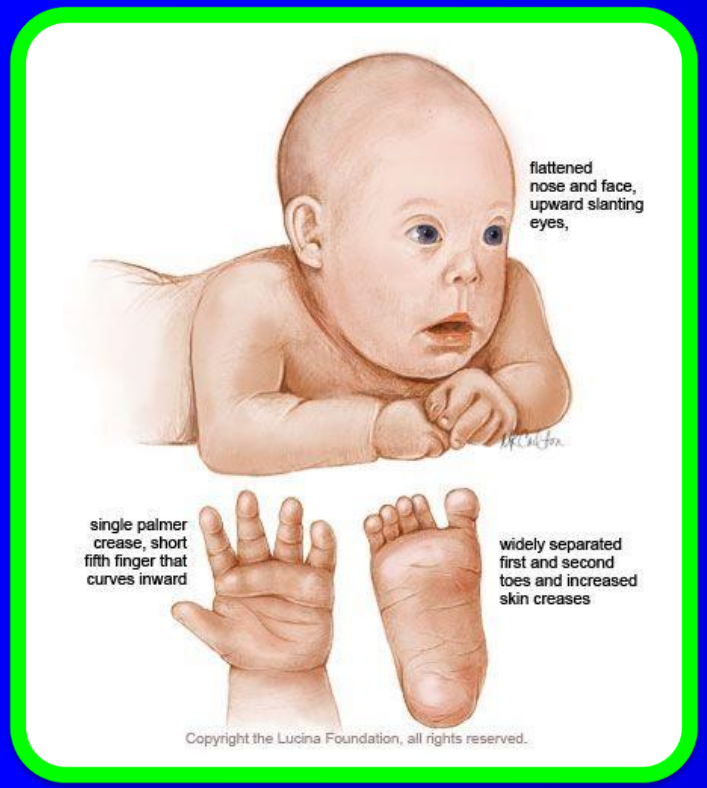
Down's Syndrome

Down's Syndrome

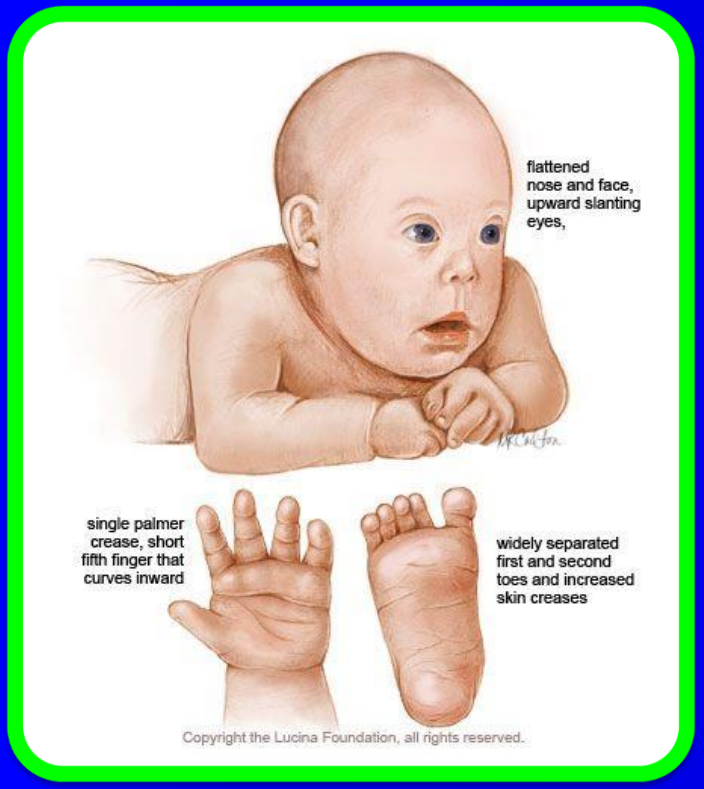


Down's Syndrome is caused due to presence of an additional copy of the chromosome number 21**(Trisomy of 21).**

This disorder was first described by **Langdon Down (1866).**

B. John Ebenezer

Down's Syndrome



Features of Down's Syndrome:

Short stature with small round head.

Furrowed tongue.

Partially opened mouth.

Broad palm with characteristic palm crease.

Physical, psychomotor and mental retardation.

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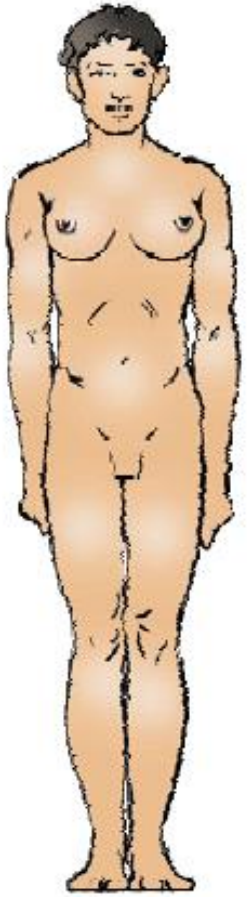
Klinefelter's Syndrome

Klinefelter's Syndrome



Klinefelter's syndrome is caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, (44+XXY).

They are **males** with **feminine** characters.

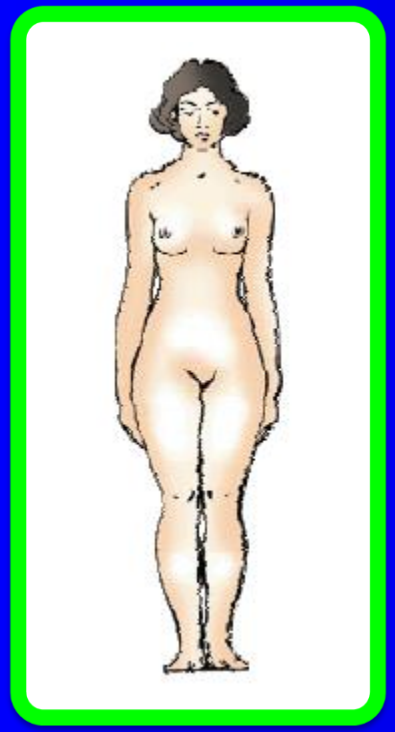
They have breast (Male breast - Gynaecomastia)

They are sterile.

B. John Ebenezer

Turner's Syndrome

Turner's Syndrome



Turner's syndrome is caused due to the absence of one of the X-chromosomes. 45 (44 + X0).

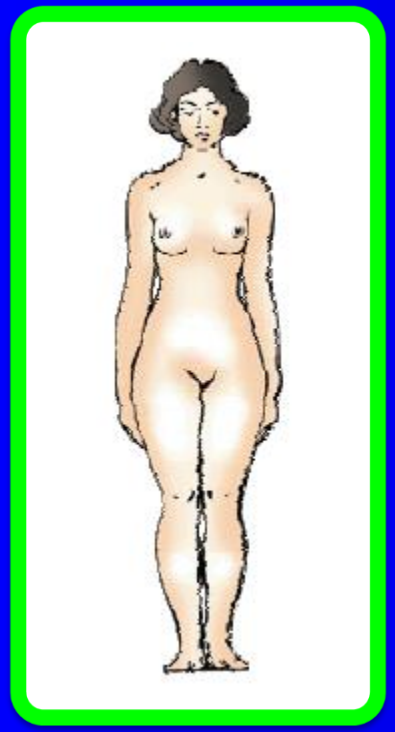
They are **females** but sterile.

They have rudimentary ovaries.

They lack secondary sexual characters.

B. John Ebenezer

Turner's Syndrome



Turner's syndrome is caused due to the absence of one of the X-chromosomes. 45 (44 + X0).

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God Bless You!