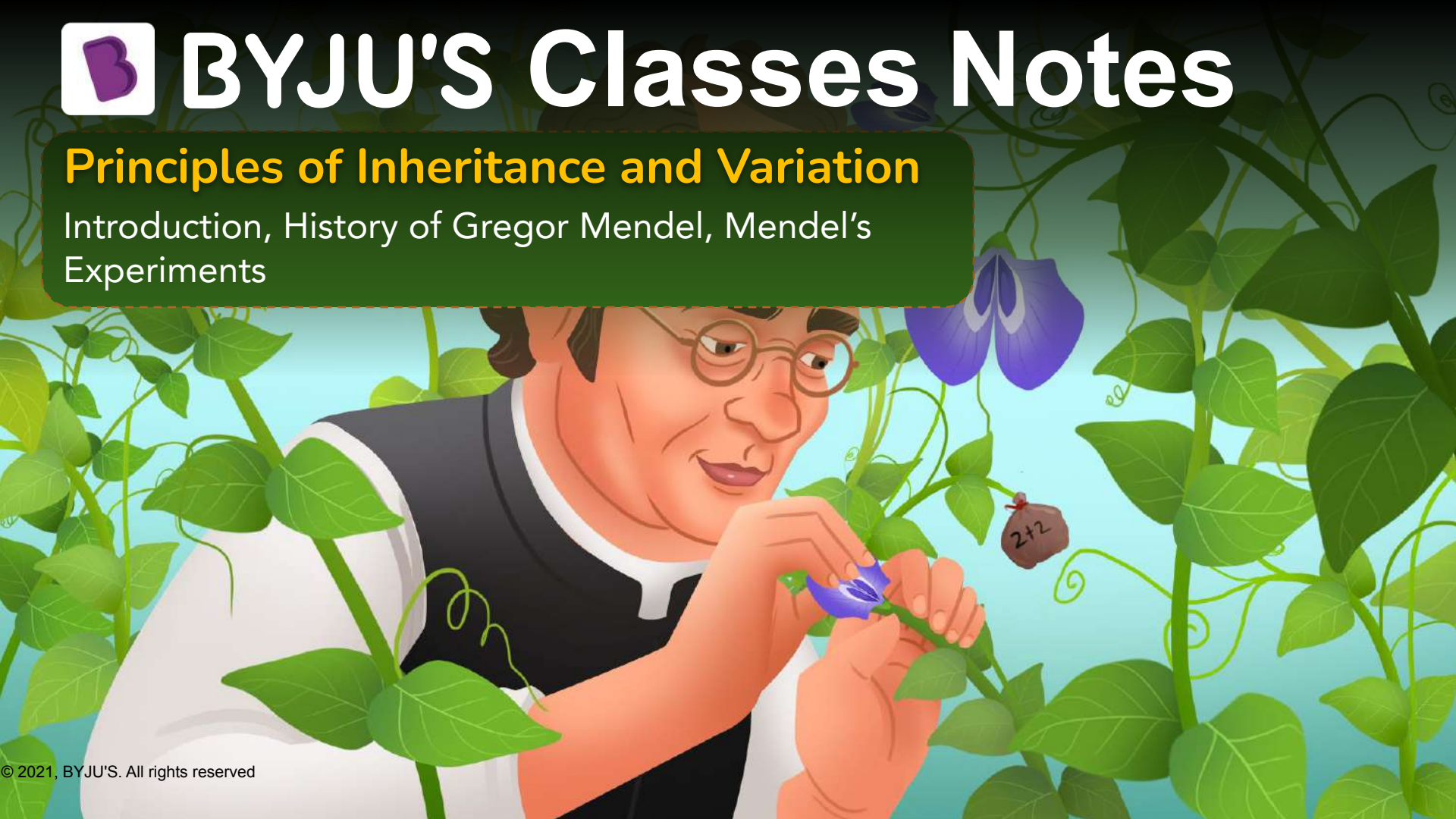
BYJU'S Classes Notes

Principles of Inheritance and Variation

Introduction, History of Gregor Mendel, Mendel's Experiments

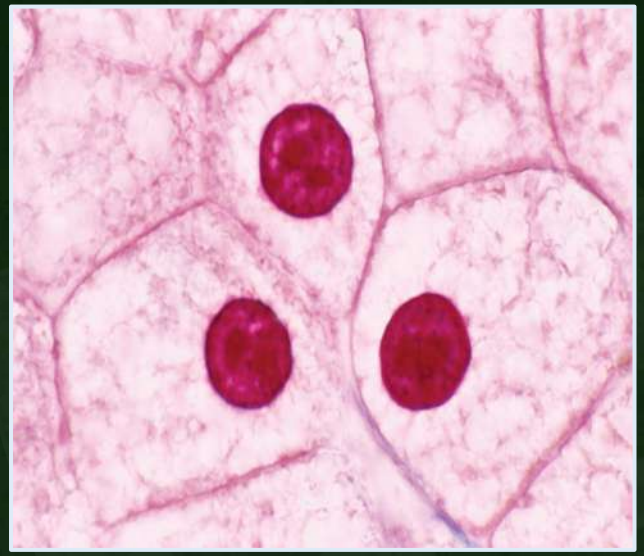


Key Takeaways

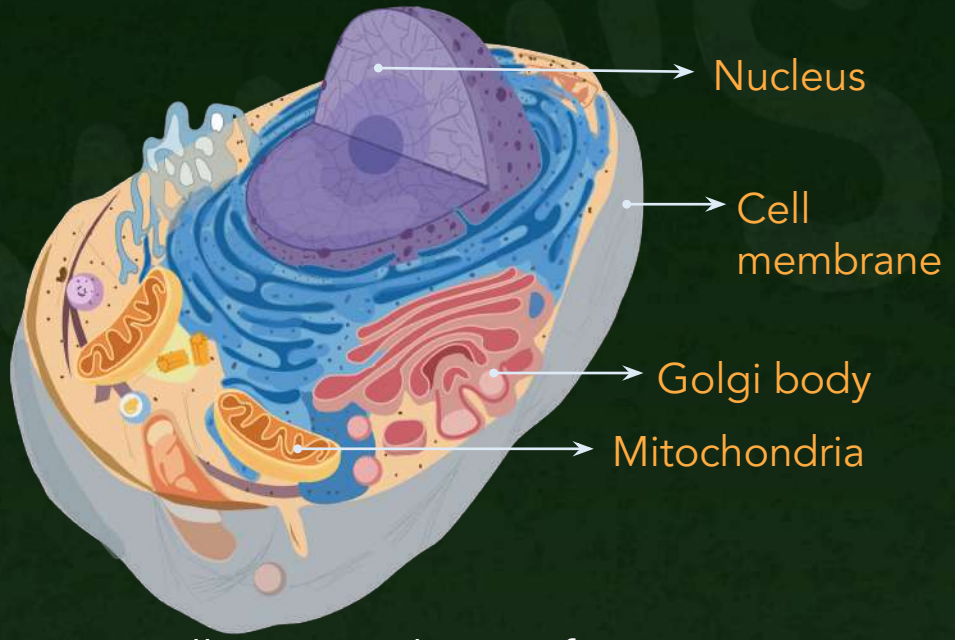
1. Introduction to genetics
2. History of Gregor Mendel
3. Mendel’s experiments
   - Experimental technique

Summary

Recall! Cell - The Fundamental Unit of Life

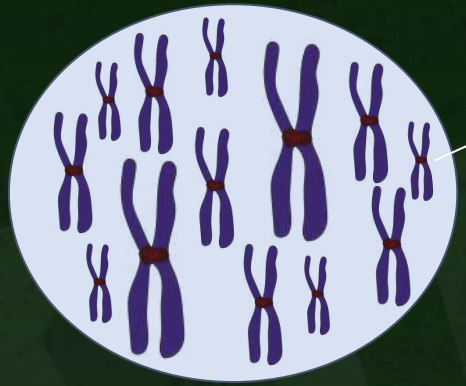


- Every organism is made up of cells.

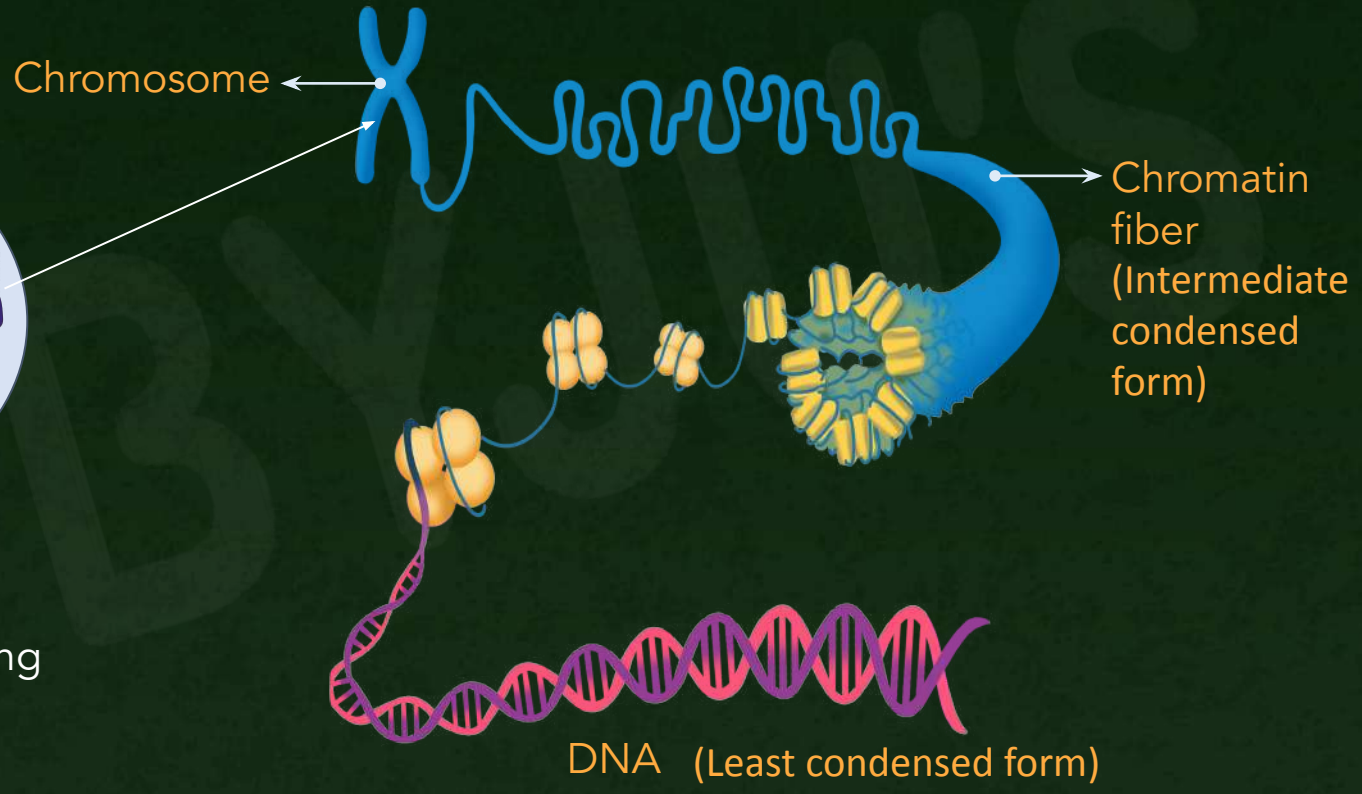


- Cells are made up of organelles.

Recall! Genetic Material

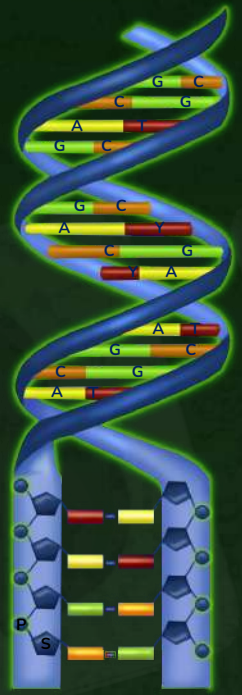


- Genetic material is packed into chromosomes during metaphase of cell division.



Recall! DNA - Sequence of Nucleotides

DNA (Deoxyribonucleic acid)



- DNA is the genetic material that consists all the informed for producing proteins.
- Sequence of nucleotides in DNA codes for specific proteins.

Introduction to Genetics

Gene

- Distinct sequence of nucleotides of DNA that codes for a specific protein
- Physical and functional unit of heredity

Heredity

- Transmission of genes from one generation to another

Inheritance

- The pattern in which the genes are transferred from one generation to another

Genetic Terms

Genetics

- Branch of biology that deals with the study of genes and its inheritance

Character

- A heritable feature that varies among individuals

Trait

- Each variant of a character

What is a gene?

A. A protein

B. A lipid

C. Unit of nuclear membrane

D. Unit of heredity

What is a gene?

A. A protein

B. A lipid

C. Unit of nuclear membrane

D. Unit of heredity

History of Gregor Mendel

- Mendel was born in 1822 to a poor farmer family in Austria. He received agricultural training in childhood and was good in academics.

- Later, he got trained to become a scientist. After completing his studies, he became a monk.

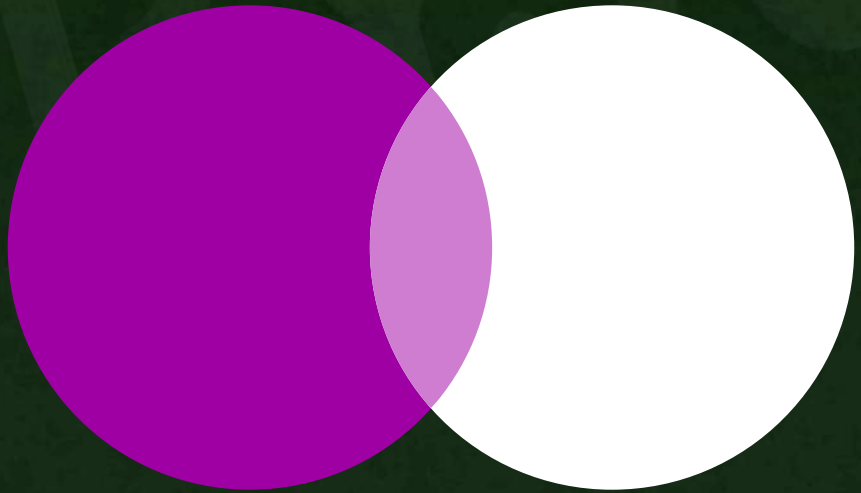
History of Gregor Mendel

- At the age of 21, he joined the monastery, where he got the name Gregor.

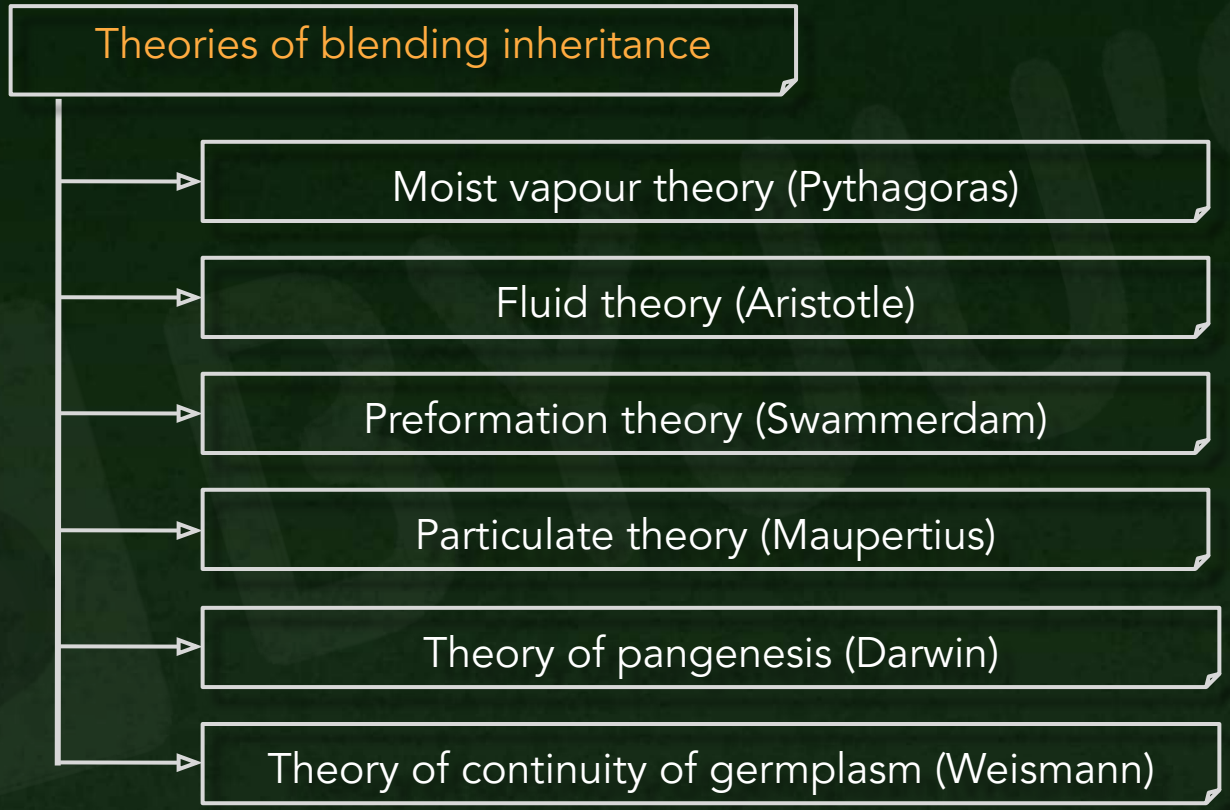
- Mendel spent much of his time in the backyard of the monastery because of his childhood inclination towards plants and agriculture.
- Among all the different kinds of plants, the pea plant caught his attention.

Pre-Mendelian Era – Blending Theory

- During the 1800s, people widely believed in the blending theory. It states that "offspring of 2 parents would always have the average or mix of the traits of the parents".
- For example, mixing of violet and white flower bearing parents would give a pale violet or pink coloured offspring.



Pre-Mendelian Era – Blending Theory



Mendel had doubts about the blending theory. So, to test this, He started to experiment with his garden pea plants.

Mendel's Experiments

Pea plant (*Pisum sativum*) – experimental model plant

- Has life span of one year
- Requires less maintenance and labour
- Multiple plants can be grown in small area
- Availability of pure varieties
- Exhibits a variety of visible characters with contrasting traits
- Self-fertilising plant and hence mating can be controlled easily

Pea Plant – Experimental Model Plant

Different characters of pea plant

- It has chromosome number 2n = 14.
- Mendel originally found that pea plants had a lot of different characters.
- Some characters caught his attention more than the others.

Pea Plant – Experimental Model Plant

Out of so many different characters, Mendel focused on these 7 characters.

Pea Plant – Experimental Model Plant

Tall pea plant Short pea plant

Violet flower

White flower

Axial flower

Terminal flower

Green pod

Yellow pod

Inflated pod

Constricted pod

Yellow seed

Green seed

Round seed

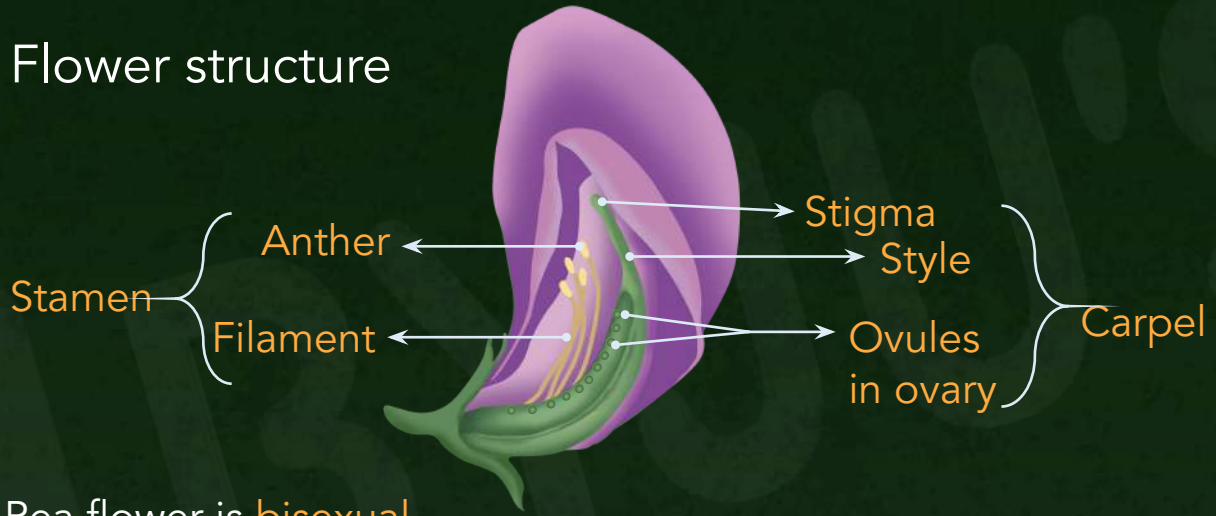
Wrinkled seed

Pea Plant – Experimental Model Plant

Contrasting traits		Character
Tall plant	Short plant	Plant height
Violet flower	White flower	Flower colour
Axial flower	Terminal flower	Position of flower
Green pod	Yellow pod	Pod colour
Inflated pod	Constricted pod	Pod shape
Yellow seed	Green seed	Seed colour
Round seed	Wrinkled seed	Seed shape

Pea Plant- Flower

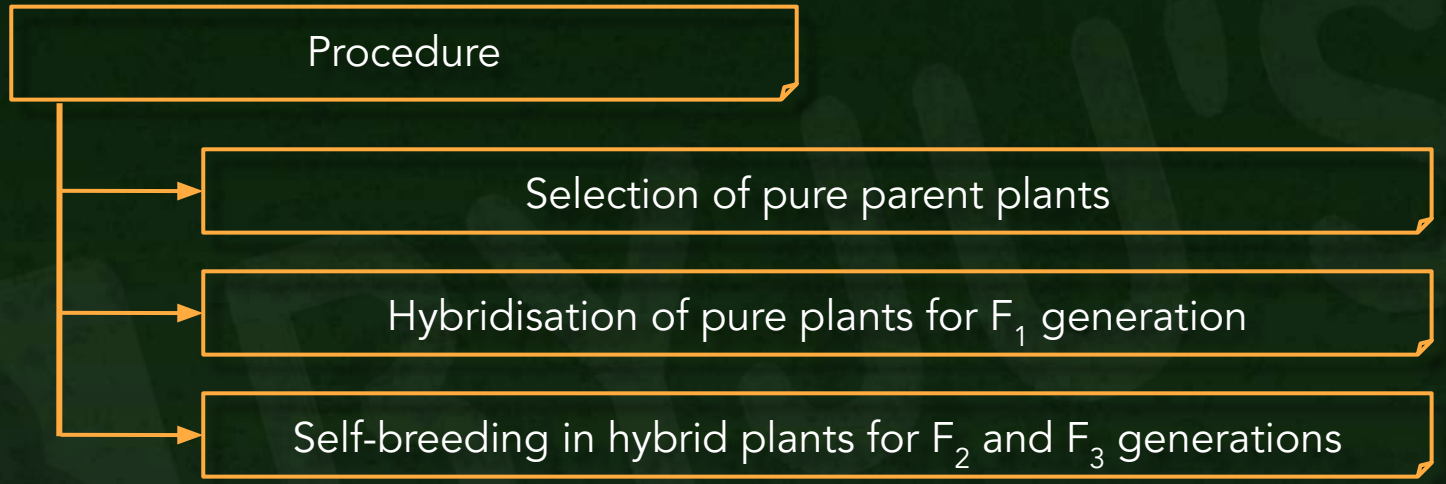
Flower structure



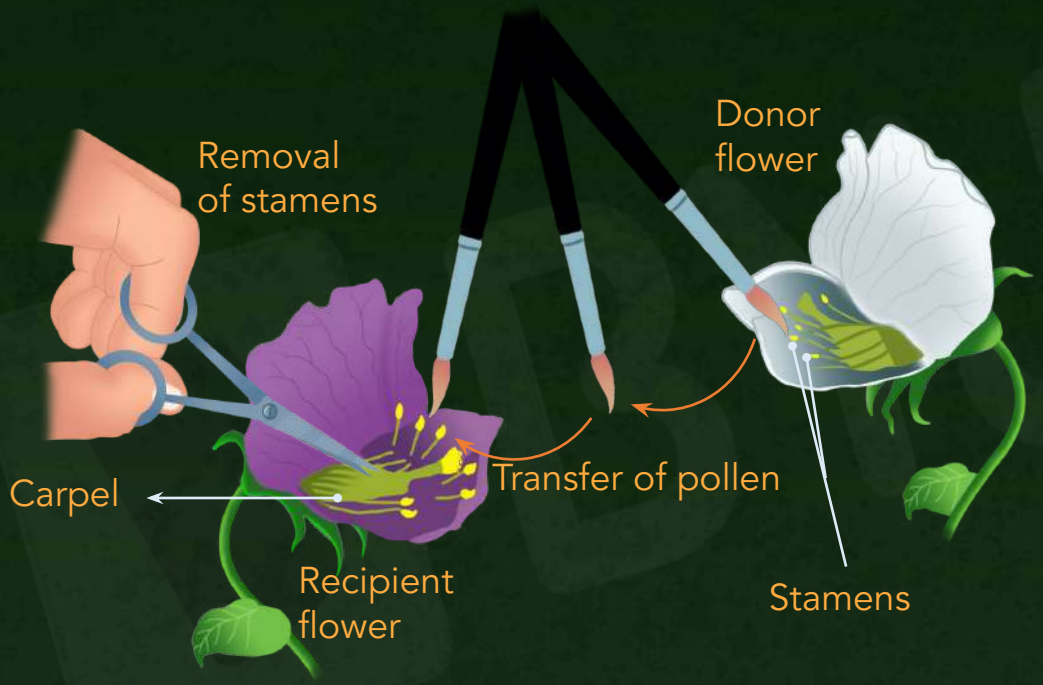
- Pea flower is bisexual.
  - It contains both male and female reproductive structures enclosed in the innermost two petals.
- It is self-fertilising in nature.
- Controlling mating in such flowers is easier because it can avoid contamination from other pollens.


Experimental Technique

Mendel's Experiment



Controlled Mating - Experimental Technique



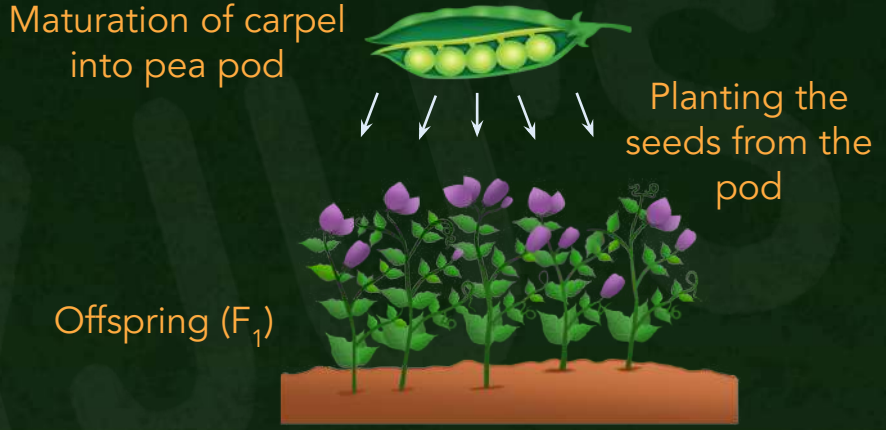
Step 1

- Stamens of the recipient flower were removed making it female flower (emasculation).

Step 2

- Mature pollen grains were collected manually from the donor flower using brush and carefully transferred to the stigma of female recipient flower (cross-pollination).

Controlled Mating - Experimental Technique



Step 3

- These flowers were then covered with transparent bags (bagging) to avoid contamination from any other pollen source.
- It was also labelled (tagging) to avoid any confusion.

Step 4

- After fertilisation, the carpel matured into a pea pod. Mendel collected the seeds formed from this fertilisation.
- These seeds were sown and the plants grown from these were called F_1 offspring.

Monohybrid Experiment

True breed yellow pea plant

X

True breed green pea plant

Flower

Flower

- True breed - Plant which undergoes self fertilisation for several generations, such that their traits remain unchanged.

Monohybrid Experiment

Plants = First filial generation (F_1)

All F_1 generation plants bore yellow coloured peas

This was against the blending theory which suggests that yellow and green colours would blend to give seeds with intermediate colour.

Monohybrid Cross

Mendel continued the experiment further to understand the observations.

Filial 1 generation (F_1)

1) Self pollination of F_1 generation

2) Seeds obtained from self pollination of F_1

3) Seeds planted

4) F_2 or filial 2 generation obtained

Plants = Filial 2 generation (F_2)

- The F_2 generation was found to have both yellow and green pea seeds, in the ratio of 3:1.
- The characteristic which had disappeared in F_1 generation (green coloured seeds) had appeared again in F_2 generation.

Monohybrid Experiment

F_2 generation plants with yellow peas

F_2 generation plants with green peas

3.01 : 1

- He found that around 6,022 plants had yellow peas and 2,001 plants had green peas.
- The ratio coming to 3:1

Which of the following did Mendel consider as characters?

A. Flower colour

B. Terminal flowers

C. Seed shape

D. Both (a) and (c)

Which of the following did Mendel consider as characters?

A. Flower colour

B. Terminal flowers

C. Seed shape

D. Both (a) and (c)

Monohybrid Cross

Cross between plants differing only in one character.

Summary

- Gene
  - Distinct sequence of nucleotides of DNA that codes for a specific protein
  - Physical and functional unit of heredity
- Heredity - Transmission of genes from one generation to another
- Inheritance - The pattern in which the genes are transferred from one generation to another
- Genetics - Branch of biology that deals with the study of genes and its inheritance
- Character - A heritable feature that varies among individuals
- Trait - Each variant of a character

Summary

- Seven contrasting traits of pea plant were studied by Mendel

Tall pea plant Short pea plant

Violet flower

White flower

Axial flower

Terminal flower

Green pod

Yellow pod

Inflated pod

Constricted pod

Yellow seed

Green seed

Round seed

Wrinkled seed

Summary

Controlled mating - experimental technique

1. Stamens of the recipient flower were removed making it a female plant (emasculation).
2. Mature pollen grains were collected manually from the donor flower and transferred to the stigma of female recipient flower (cross-pollination).
3. These flowers were then covered with transparent bags (bagging) to avoid contamination from any other pollen source.
4. It was also labelled (tagging) to avoid any confusion.
5. After fertilisation, the carpel matured into a pea pod. Mendel collected the seeds formed from this fertilisation.
6. These seeds were sown and the plants grown from these were called F1 offspring.

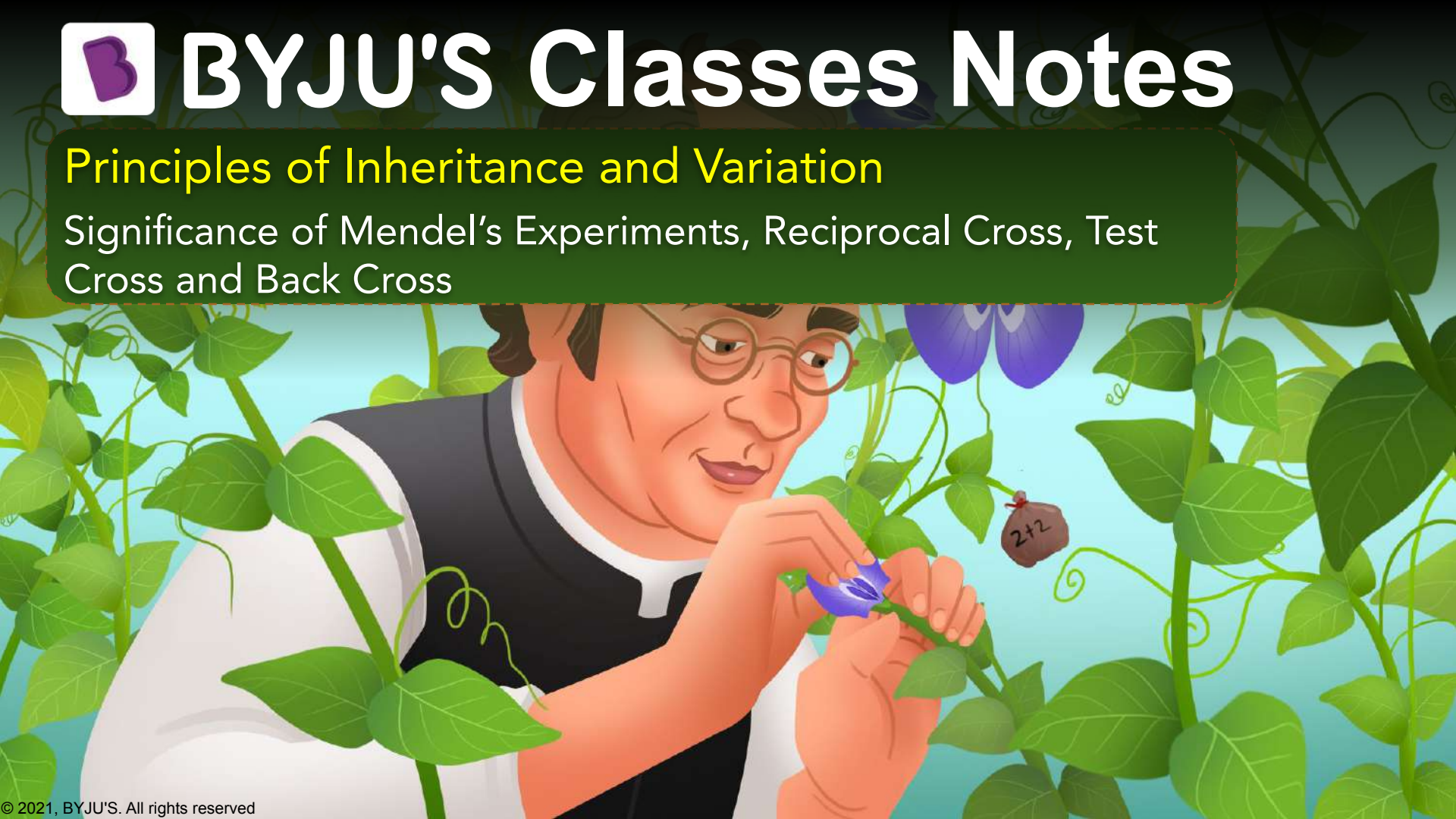
Summary

- Mendel choose pea plant for the following reasons:
  - Has life span of one year
  - Requires less maintenance and labour
  - Multiple plants can be grown in small area
  - Exhibits a variety of visible characters with contrasting traits
  - Self-fertilising plant and hence mating can be controlled easily
- Mendel is known as the "Father of Modern Genetics"

BYJU'S Classes Notes

Principles of Inheritance and Variation

Significance of Mendel's Experiments, Reciprocal Cross, Test Cross and Back Cross



Key Takeaways

1. Significance of Mendel's experiments
2. Reciprocal cross
3. Mendel's observations
4. Mendel's propositions
5. Test cross and back cross

Summary

Recall! Traits

Traits

They are different variants of a character

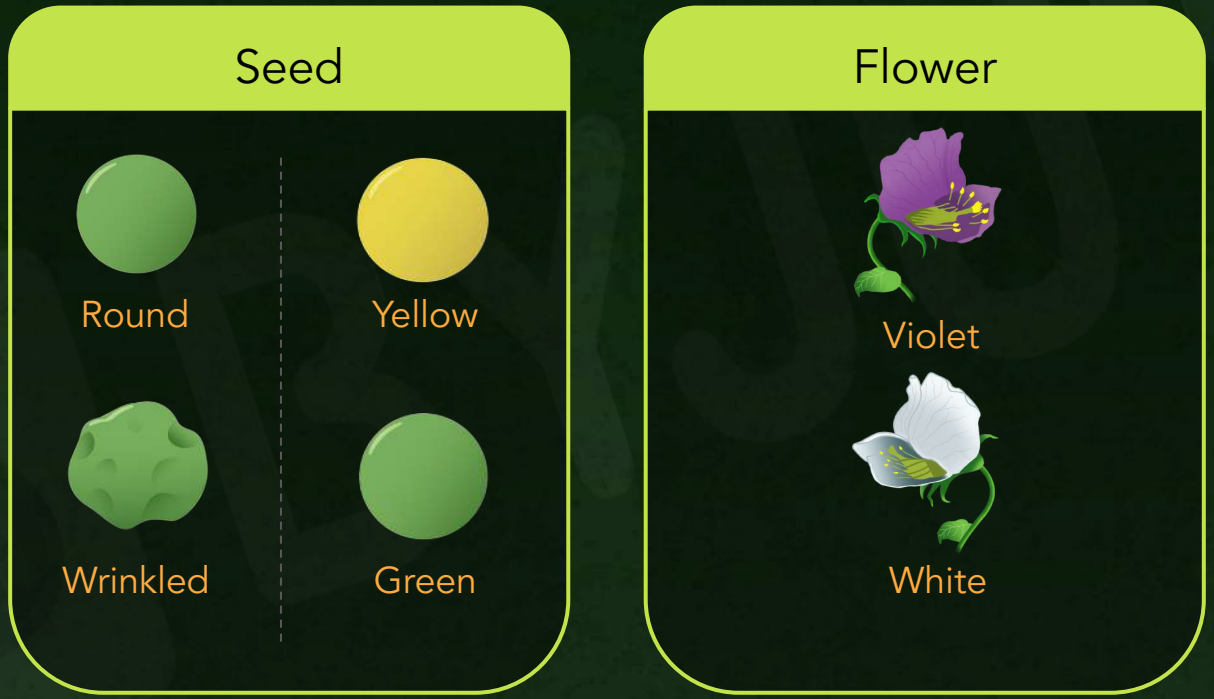
Character Example: Colour of pea seed

Trait Example: Yellow or green coloured pea seeds

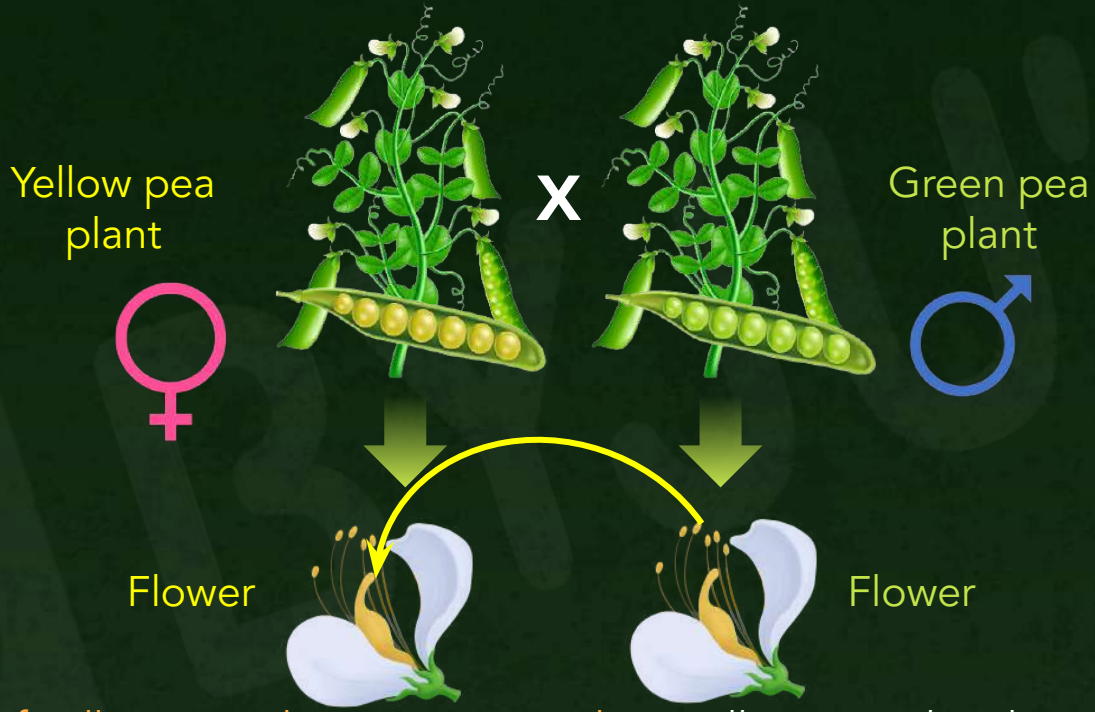
Recall! Contrasting Traits Used by Mendel

Pod		Stem	
 Inflated (Full)	 Green	 Axial flowers	 Long (6 - 7 ft)
 Constricted	 Yellow	 Terminal flowers	 Short (3/4th - 1 ft)

Recall! Contrasting Traits used by Mendel

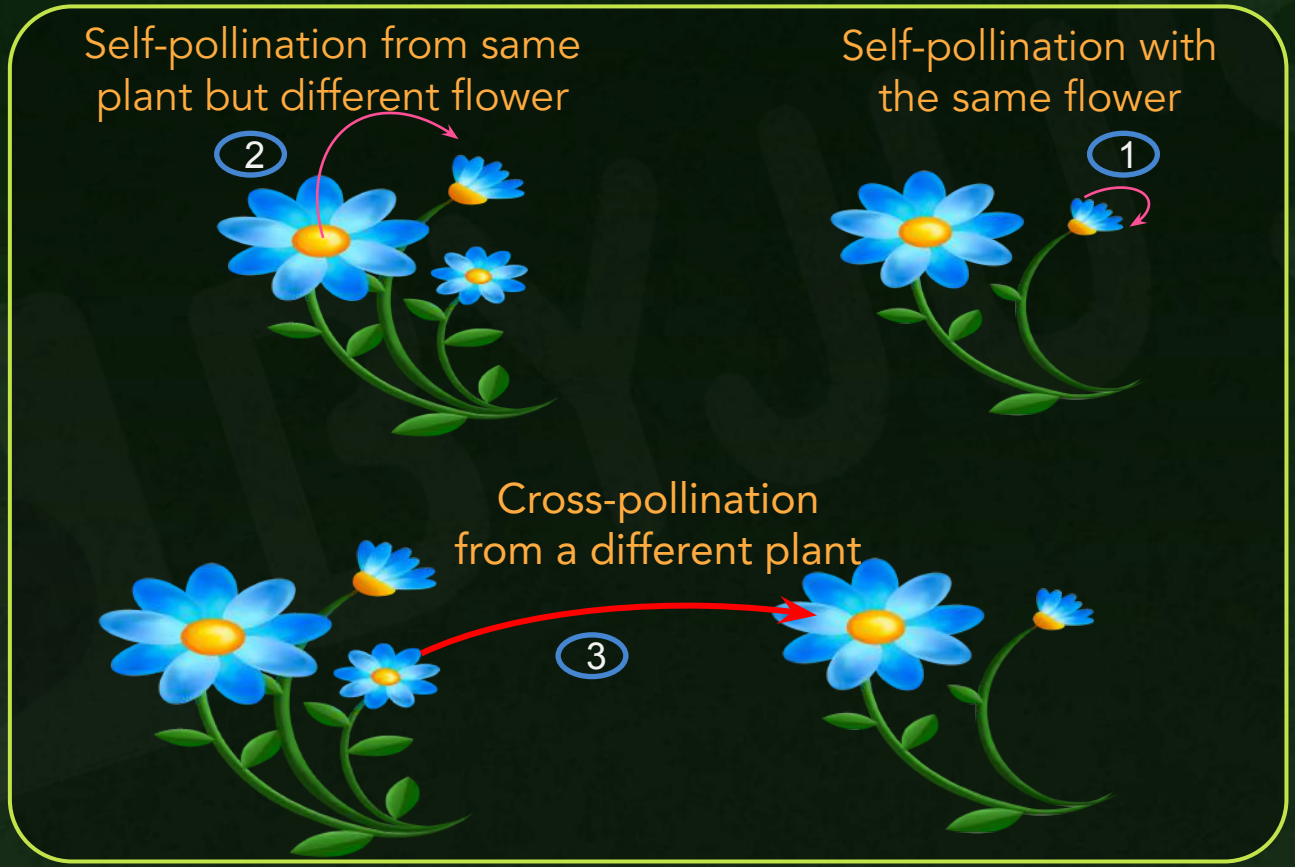


Recall! Procedure for Cross-Pollination



- The stamens of yellow pea plant are removed, so yellow pea plant has stigma only (female parent).
- Then pollens of green pea plant are transferred to the stigma of yellow pea plant, making green pea plant the male parent.

Recall! Self-Pollination and Cross-Pollination

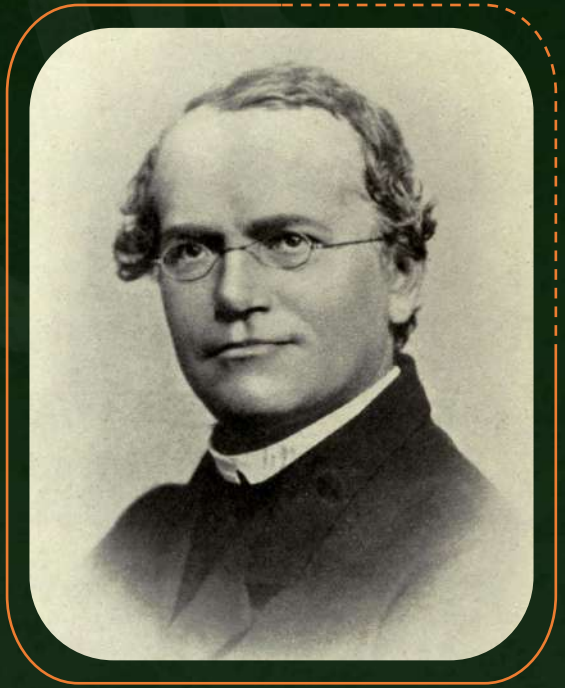


Significance of Mendel's Experiments

- Duration of experiments - 8 consecutive years
- Number of plants experimented – approx. 28,000 plants
- Data set – Minimum three generations for eac of the 7 characters
- Obtained interesting results which were explained mathematically and statistically
- Published his work in 1865
- Concluded that factors are discrete units which are passed from one generation to another

Mendel's Work Was Not Appreciated!

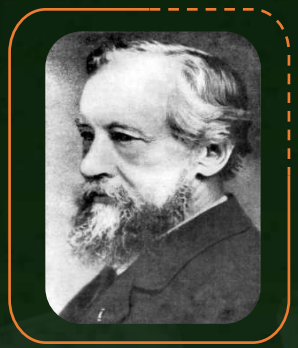
- Mendel's work was not widely publicised because of poor communication.
- Contemporaries believed in 'blending theory' and did not accept his results.
- He was asked to show the physical existence of "factors".
- Usage of statistics and mathematics to explain biological phenomena was unaccepted and misunderstood.
- Limited circulation of the "Proceedings of Brunn Natural Science Society" in which his work was published.



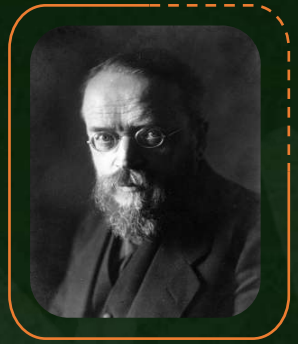
Mendel's Work Was Not Appreciated!

- He failed to reproduce the results on Hawkweed (*Hieracium*) undertaken on the suggestion of Nägeli. It was due to non-availability of pure lines.
- Lack of aggressiveness in his personality was one of the reasons.
- The scientific world was being rocked at that time by Darwin's theory of evolution (Origin of Species, 1859).
- Mendel's concept of stable, unblending, discrete units or factors for various traits did not find acceptance from the contemporaries.

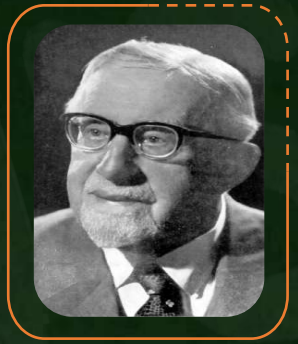
Resurfacing Of Mendel's Work - 1900's



Hugo de Vries



Carl Correns



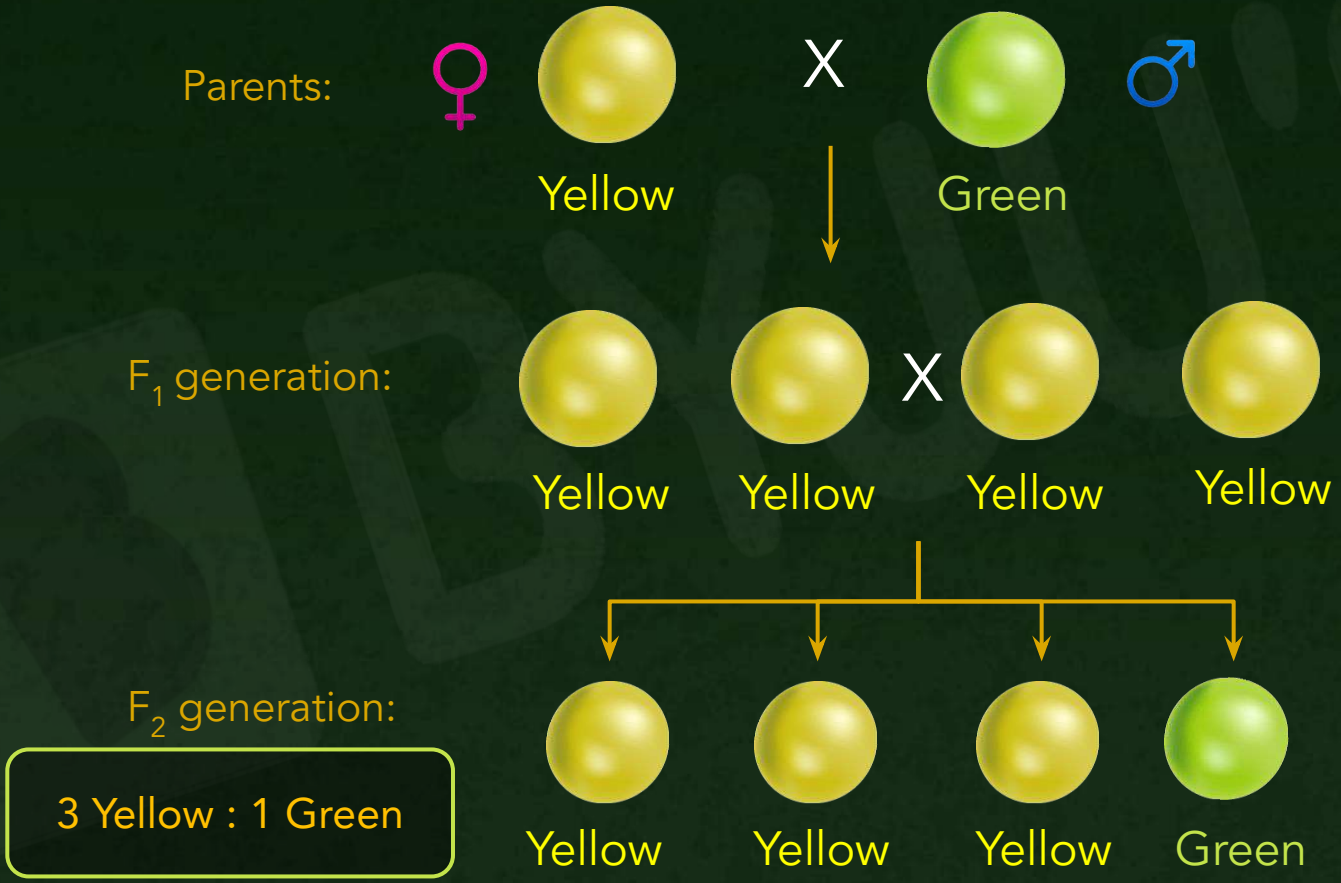
Erich Von Tschermak

- These three scientists independently rediscovered the same concepts as that of Mendel.
- They were unaware of Mendel's work initially, but later acknowledged Mendel as the pioneer.
- Mendel's work resurfaced and got the appropriate recognition.

Mendel - Father of Modern Genetics

- Innovative and unique work by him unravelled the fundamental laws of inheritance.
- He laid the foundation of modern genetics.
- He is known as the "Father of Modern Genetics"

Recall! Monohybrid Cross



Reciprocal Cross

Reciprocal cross

It is a paired cross in which traits of male and female parents are switched.

Reciprocal Cross

- Mendel performed his experiment with yellow and green pea seed-bearing true breeds.
- He first chose yellow pea plant to be female parent and green pea plant to be male parent.
- He performed the cross and obtained F_1 progenies.
- He found that all the F_1 progenies were yellow.
- When F_1 was selfed, he found that F_2 progeny had yellow and green pea plants in the ratio 3:1.

Reciprocal Cross

- Mendel switched the parents. This time he chose green pea plant as female parent and yellow pea plant as male parent. He obtained F_1 and F_2 progenies.
- He observed that regardless of which plant was taken as male and female parent, the progenies he obtained were same.
- All F_1 generation were yellow and F_2 generation had yellow and green pea plants in the ratio 3:1.

Reciprocal Cross

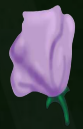
Reciprocal cross yielded same ratio of progeny

F_1 = all yellow

F_2 = yellow : green = 3:1

Mendel's Observations

- One of the two traits disappeared in F1, which reappeared in F2 in the ratio of 3:1.
- Traits were not blended in progeny.
- Mendel performed similar experiments for the other 6 characters, and observed similar pattern of inheritance in them as well.

Seed	Flower	Pod	Stem
 Round	 Violet	 Inflated (Full)	 Axial flowers
 Wrinkled	 White	 Green	 Long (6 - 7 ft)
		 Constricted	 Terminal flowers
		 Yellow	 Short (3/4th - 1 ft)

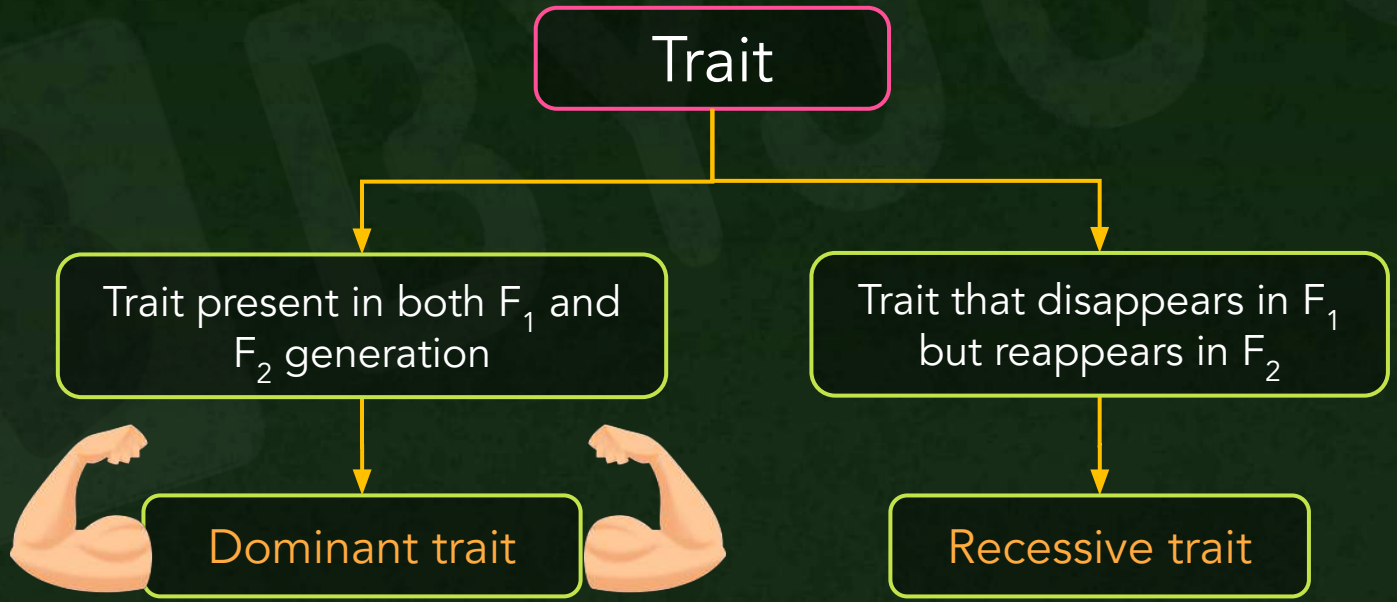
Mendel's Assumptions

- A variety of hypotheses and terms given by Mendel hundreds of years back still hold true.
- Some of the terms used by him have been renamed but the concept is still consistent.

Mendel's terms		Current terms
Character	Example: Colour of pea	Character
Trait	Example: Yellow or green colour	Allele
Factor	Unit of heredity	Gene

Mendel's Propositions

- There are two traits for each character.
- Each one of these traits is represented by factors.
- Each progeny receives one factor from each parent.
- One of these traits is dominant and the other is recessive.



Mendel's Propositions

- In case of cross between, yellow and green coloured pea plants, yellow can be called as dominant trait since it was present in both F_1 and F_2.
- While green is recessive trait since it was absent in F_1 but reappeared in F_2.

Dominants of Other Traits

Dominant	Recessive	Dominant	Recessive
 Round	 Wrinkled	 Violet	 White
Dominant	Recessive	Dominant	Recessive
 Inflated (Full)	 Constricted	 Green pod	 Yellow pod

Dominants of Other Traits

Dominant	Recessive	Dominant	Recessive
			
Axial flower	Terminal flower	Tall plant	Dwarf plant

Did You Know?

Dominant but rare

Recessive but common

- Green pea allele is recessive but today in market, we usually find that most peas are green rather yellow.
- So green pea is recessive yet common, whereas yellow pea is dominant but rare.

Mendel's Propositions

Dominant trait

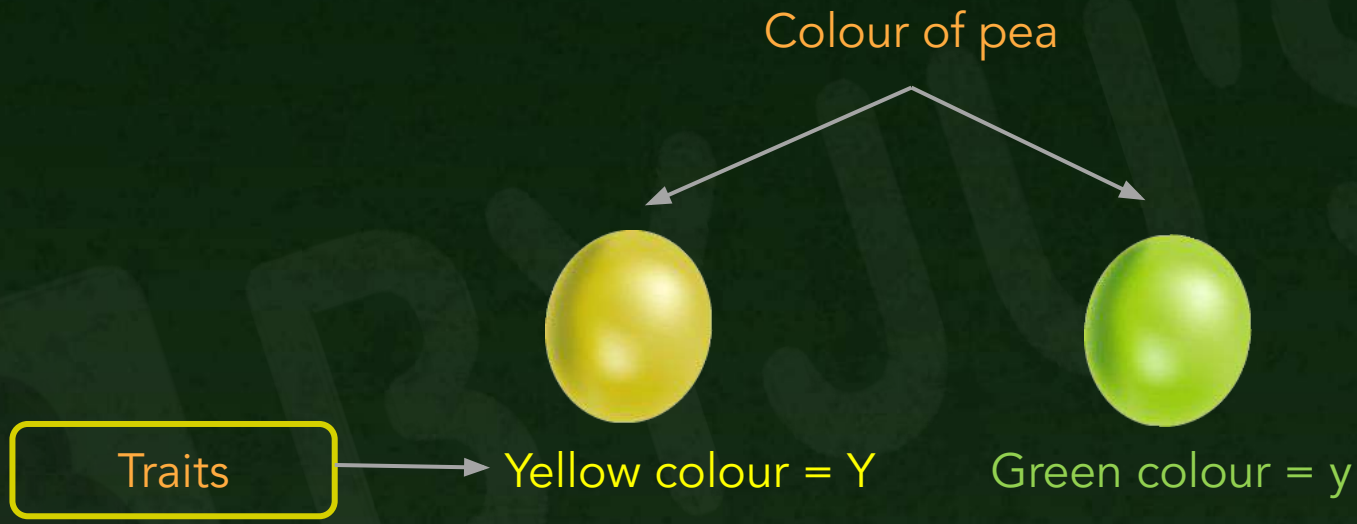
Recessive trait

- Since the recessive trait reappeared in F2 generation, Mendel thought that recessive trait must have been there in F1 as well but its presence was masked by the dominant trait.

Mendel's Propositions

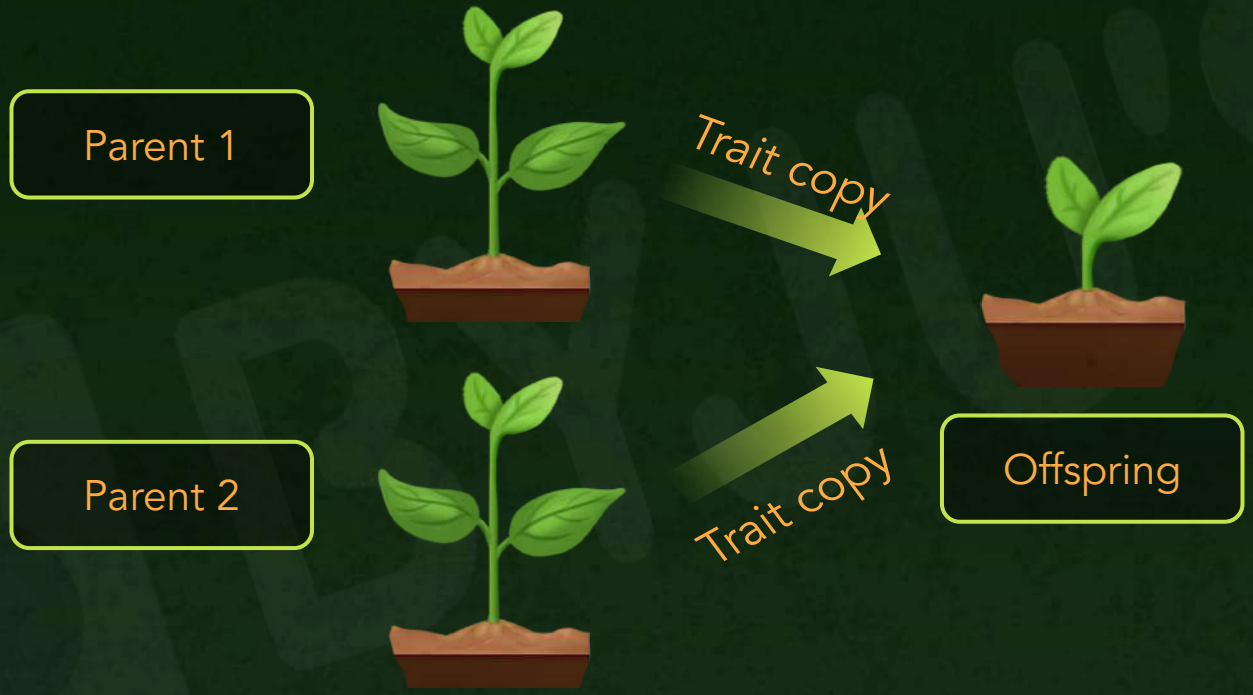
- Hence, he proposed that plants have two copies of each trait.
- For example two copies of traits for colour of pod, two for shape of seed, two for colour of flower etc.

Mendel's Propositions



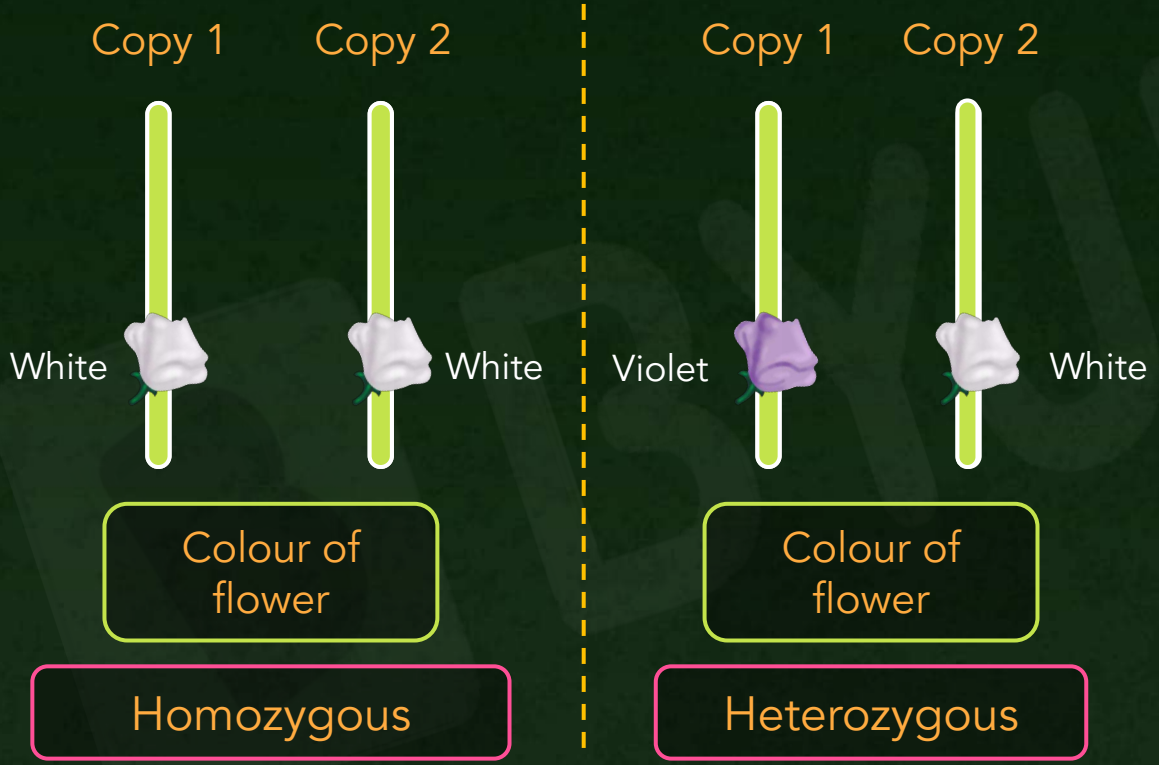
- He gave the contrasting characters certain symbols
  - First letter of character: Capital= Dominant; Small letter = Recessive

Mendel's Propositions



- Each parent contributes one copy of trait to offspring.
- The type of trait copy that the offspring receives, decides the overall appearance of offspring.

Mendel's Propositions



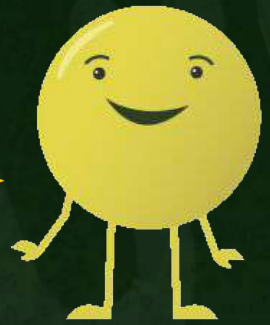
- If both the traits received by offspring are identical, then it is known as homozygous.
- If received copies are different or non identical, then it is known as heterozygous.

Mendel's Propositions

S. No.	Character	Dominant	Recessive	Chromosome No.
1	Stem height	Tall (T)	Dwarf (t)	4
2	Flower colour	Violet (V)	White (v)	1
3	Flower position	Axial (A)	Terminal (a)	4
4	Pod shape	Inflated (I)	Constricted (i)	4
5	Pod colour	Green (G)	Yellow (g)	5
6	Seed shape	Round (R)	Wrinkled (r)	7
7	Seed colour	Yellow (Y)	Green (y)	1

Mendel's Propositions

1 or 2 copies of dominant trait



Yellow colour

- Mendel also hypothesised that, since dominant trait appeared in more ratio, even a single copy of dominant trait will be seen as the observable feature.
- Here, in case of yellow and green pea cross, single or two copies of yellow trait will give yellow peas.

Mendel's Propositions

2 copies of recessive trait

Green colour

- For the recessive trait to be seen, it should be present as two copies.
- Two copies of green traits will give green peas.

Heterozygous condition means

a) both the traits are different

b) both the traits are identical

c) only one trait is present

d) none of the above

Heterozygous condition means

a) both the traits are different

b) both the traits are identical

c) only one trait is present

d) none of the above

Test Cross

- It is a cross performed to determine whether offspring is homozygous or heterozygous dominant by crossing with the recessive parent.
- The dominant character-expressing offspring is crossed with a plant that is homozygous recessive for the same characteristic.
- So, in a test cross for yellow pea plant, yellow pea plant is crossed with homozygous recessive green pea plant.
- The yellow plant maybe YY or Yy (containing two or one copy of the dominant Y trait).

Test Cross - Homozygous

- If all F_1 offsprings are found to be yellow seed pea plants expressing the dominant trait, then the test plant is homozygous dominant YY.

Test Cross - Heterozygous

- Alternatively, if F_1 offspring exhibit a 1:1 ratio of yellow and green seed pea plants, then the test plant is heterozygous Yy.

Test Cross

Heterozygous dominant

X

Homozygous recessive

Violet

White

Violet Violet White White

Ratio of Violet : White = 1:1

Back Cross

Back cross

Cross of an offspring with one of its parent plant

Test cross is a type of back cross

Test cross is the cross between

a) test plant and homozygous recessive parent

b) test plant and dominant parent

c) test plant and any parent

d) none of the above

Test cross is the cross between

a) test plant and homozygous recessive parent

b) test plant and dominant parent

c) test plant and any parent

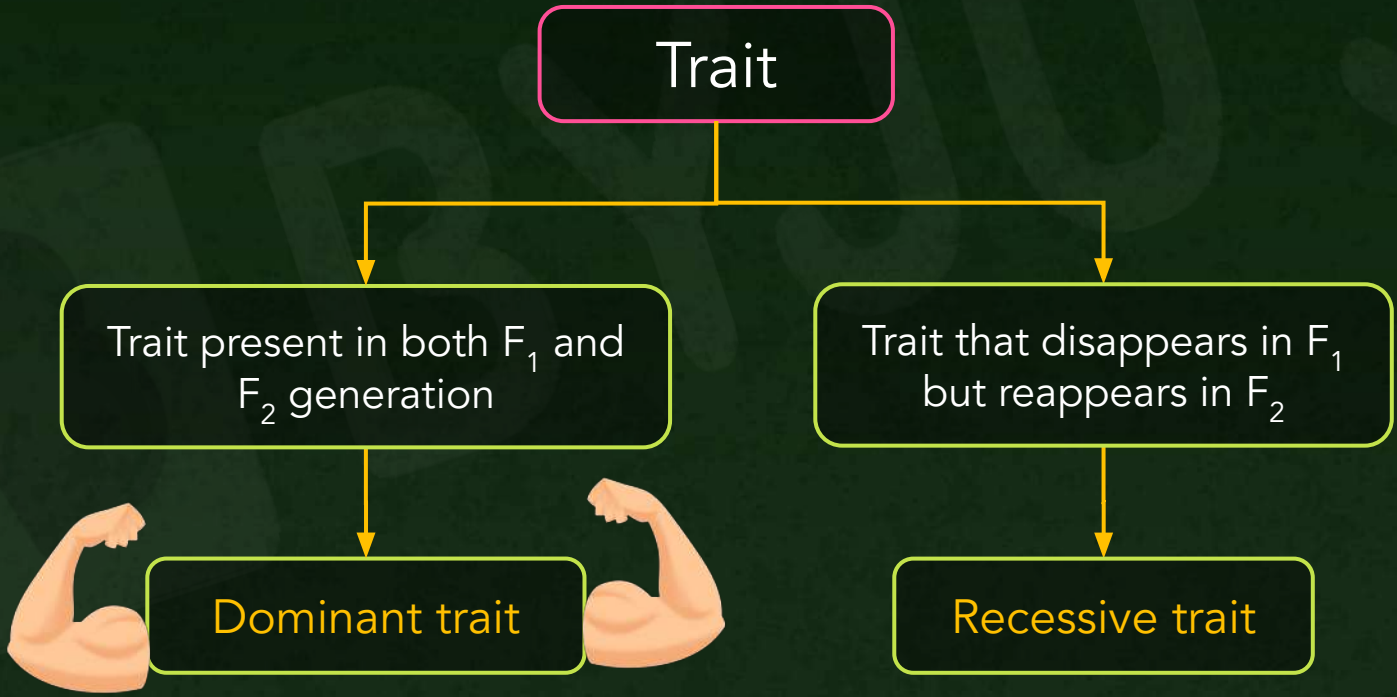
d) none of the above

Summary

Monohybrid cross

Summary

Dominant and recessive trait



Summary

Homozygous and heterozygous

Colour of flower

Homozygous

Both the copies of alleles code for same colour - White

Colour of flower

Heterozygous

Both the copies of alleles code for different colours - White and violet

Summary

Reciprocal, test and back cross

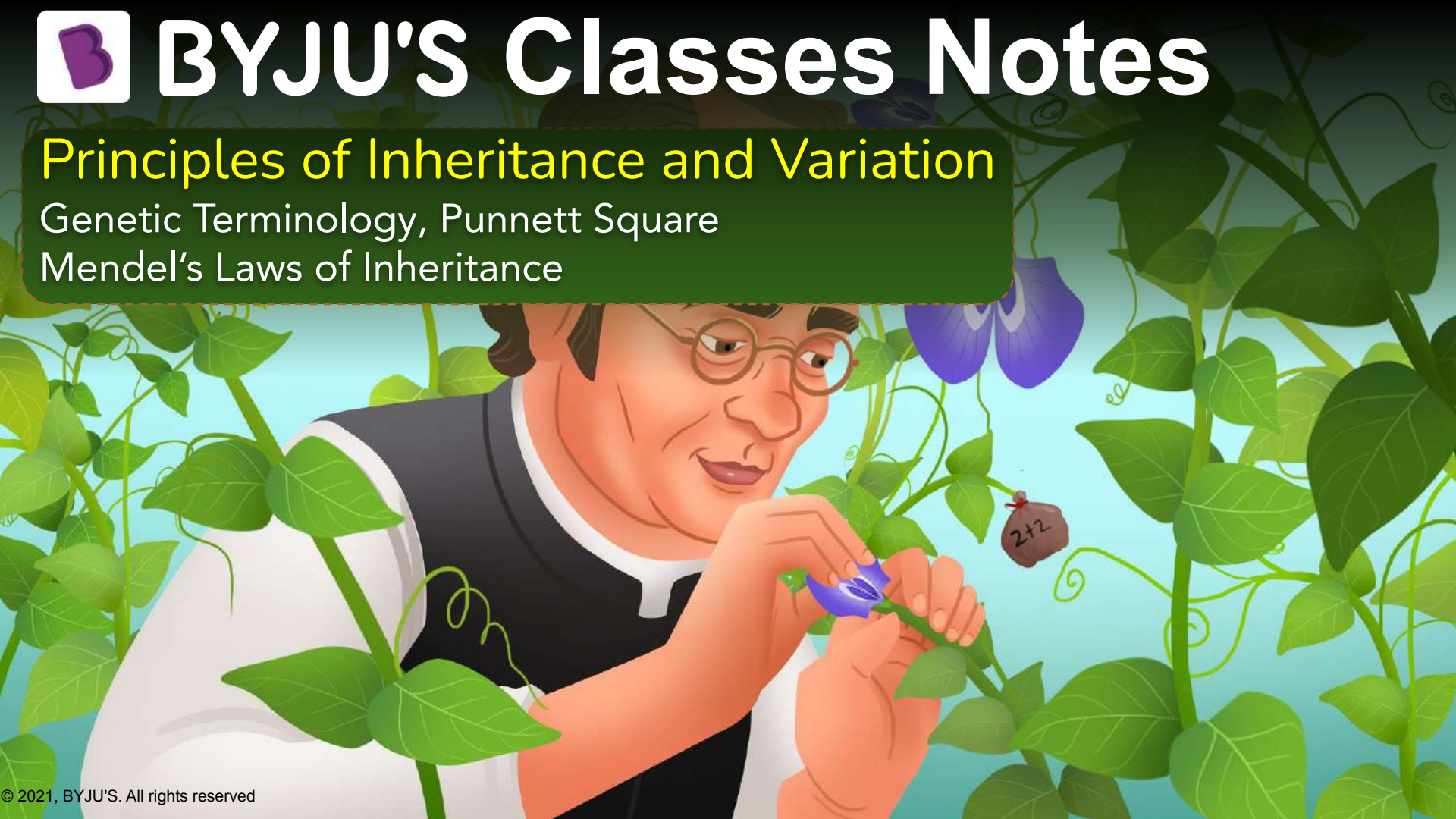
Reciprocal cross	A paired cross in which traits of male and female parents are switched
Test cross	A cross between test plant and homozygous recessive parent
Back cross	Cross of an offspring with one of its parent plant

BYJU'S Classes Notes

Principles of Inheritance and Variation

Genetic Terminology, Punnett Square

Mendel's Laws of Inheritance



Key Takeaways

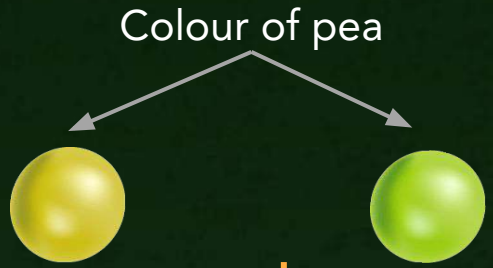
1. Genetic terminology
2. Punnett square
   - Phenotypic and genotypic ratios
   - Test cross and back cross

Mendel’s law of inheritance 3

- Law of dominance
- Law of segregation

Summary

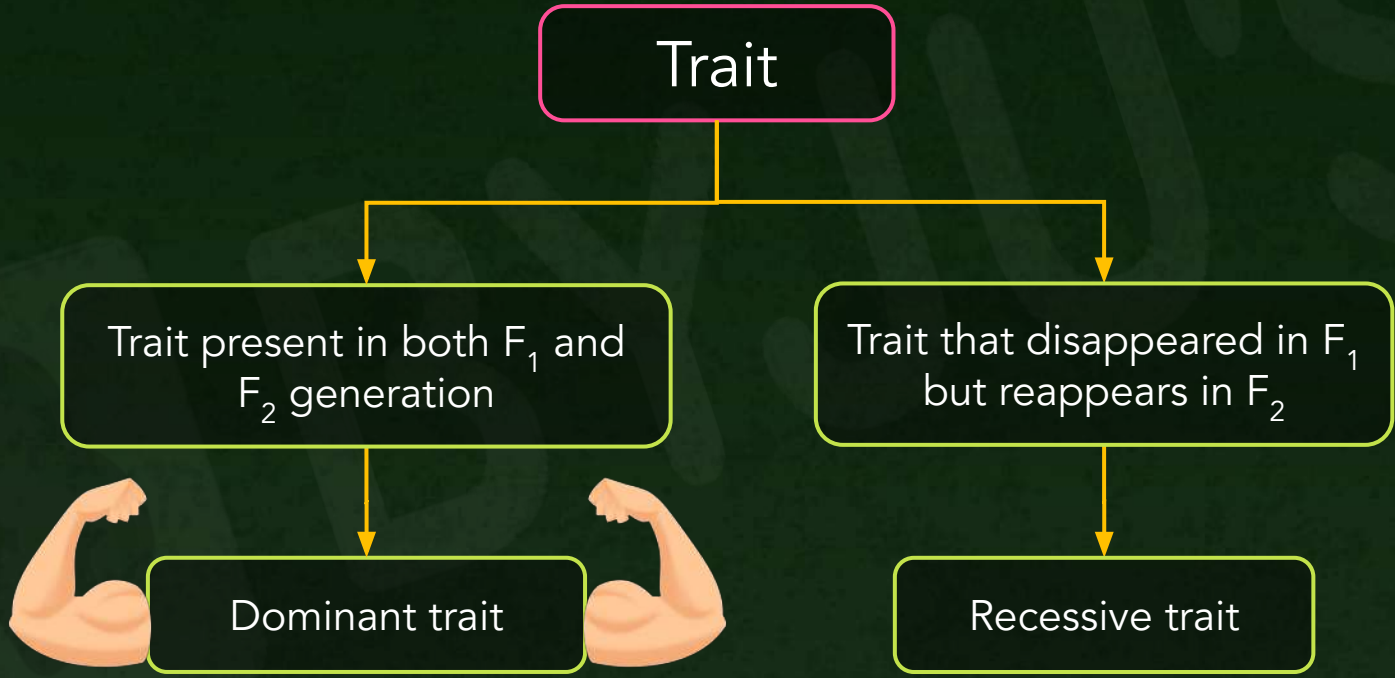
Recall! Allele Representation



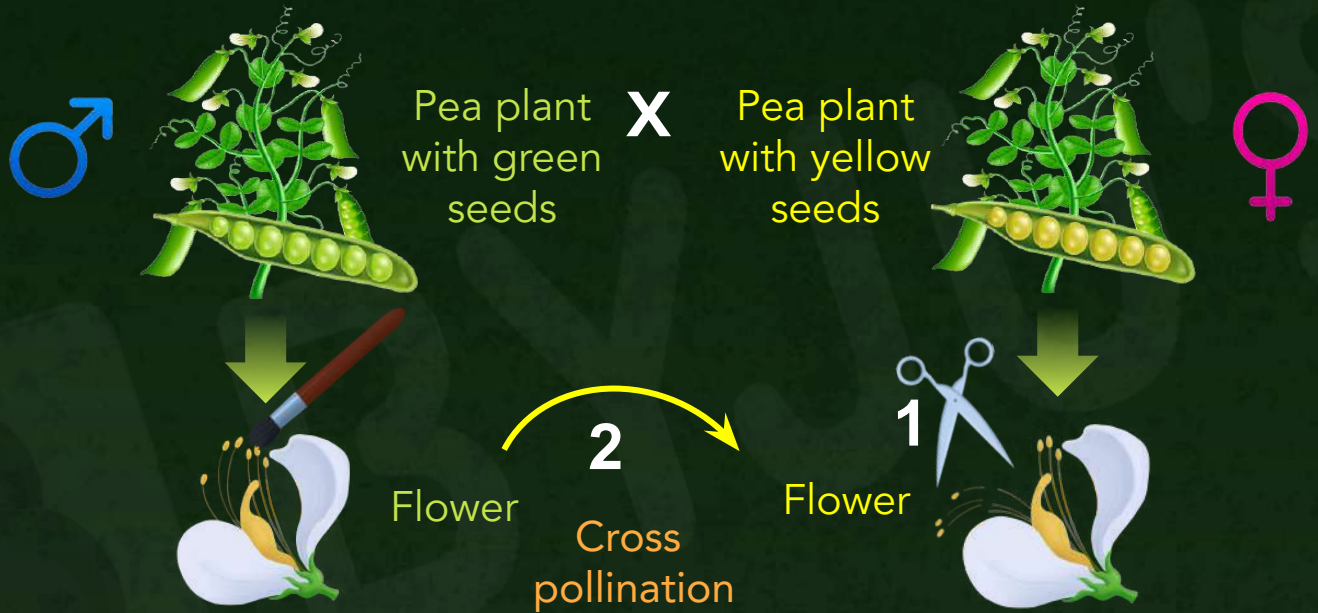
Alleles → Yellow colour = Y Green colour = y

- Every diploid organism has two copies of alleles present for each character.

Recall! Dominant and Recessive Trait



Recall! Monohybrid Cross

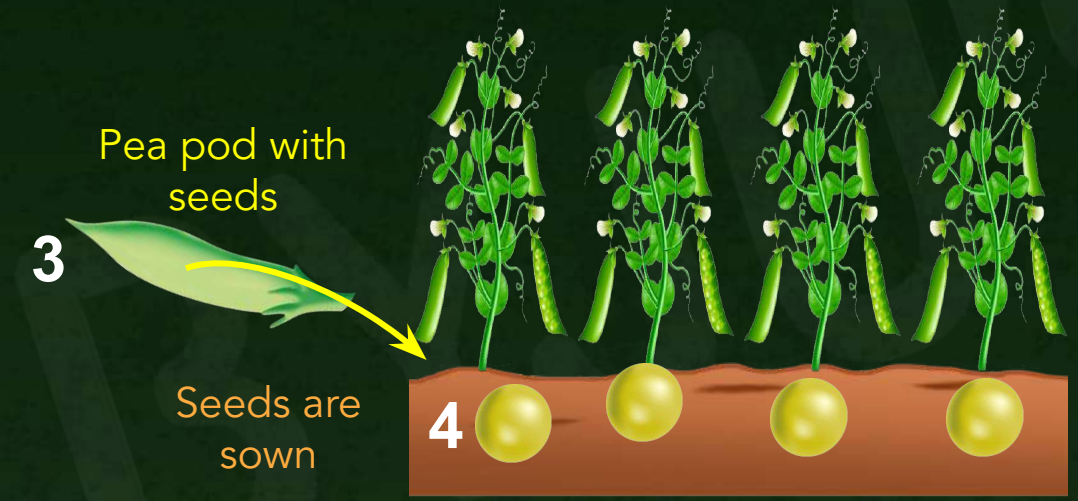


1) Stamens are removed from the yellow seed pea plant (female parent).

2) Pollens are transferred from green seed pea plant (male parent) to yellow seed pea plant's stigma (cross-pollination).

Recall! Monohybrid Cross

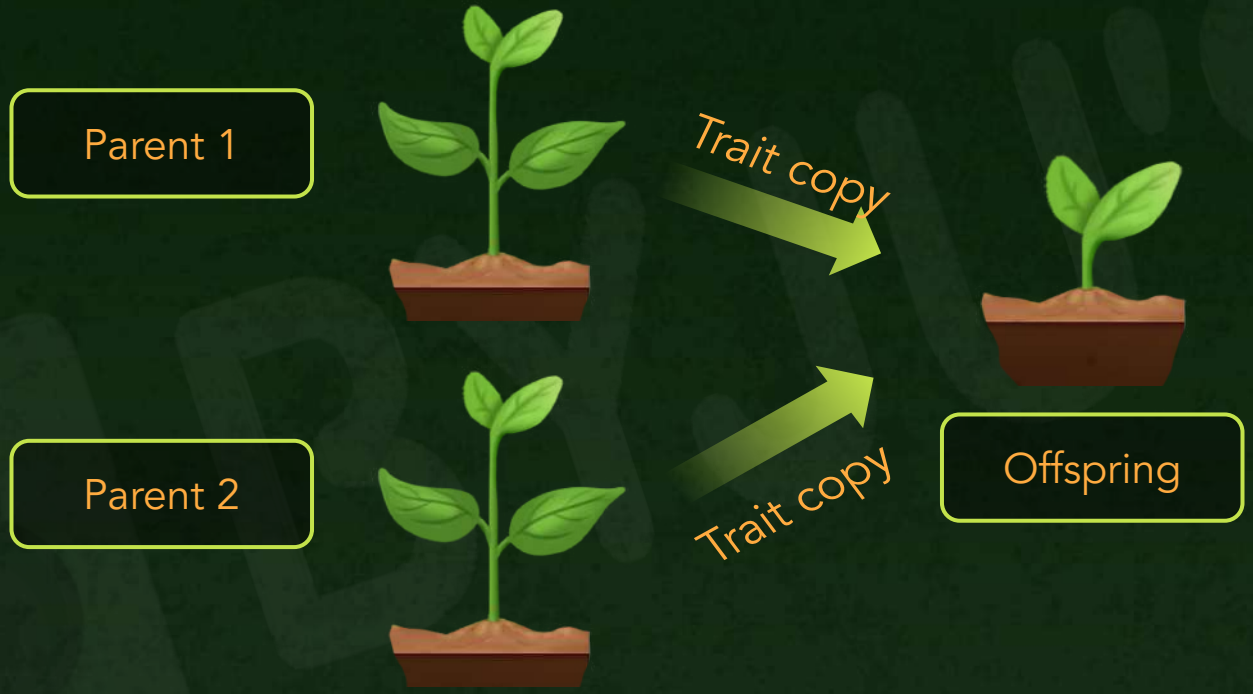
Carpel matures into pod and ovules mature to form seeds



3) Post fertilisation, carpel forms the pod and ovules form the seeds.
4) These pea seeds are sown and allowed to germinate, thus giving the F_1 generation of plants.
5) All the plants of F_1 generation, were found by Mendel, to contain yellow colored seeds.

This was found to be in direct contrast with the blending theory, according to which, the color of the seeds in F_1 generation should have been yellow.

Recall! Mendel's Propositions



- Each parent contributes one copy of trait to offspring.
- The type of trait copy that the offspring receives, decides the overall appearance of offspring.

Recall! Back Cross

Back cross

Cross of an offspring with one of its parent plant

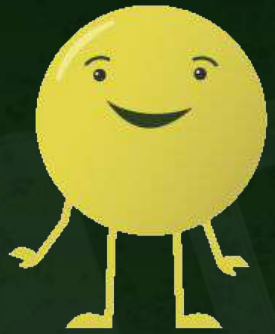
Test cross

Cross of an offspring with the recessive parent plant

Test cross is a type of back cross

Genetic Terminology

Yellow



YY or Yy

Phenotype

The character of an organism which is observable

Genotype

The actual genetic makeup of the organism

Green

yy

Combination of alleles in an organism is

a) its genotype

b) its phenotype

c) factor

d) all of the above

Combination of alleles in an organism is

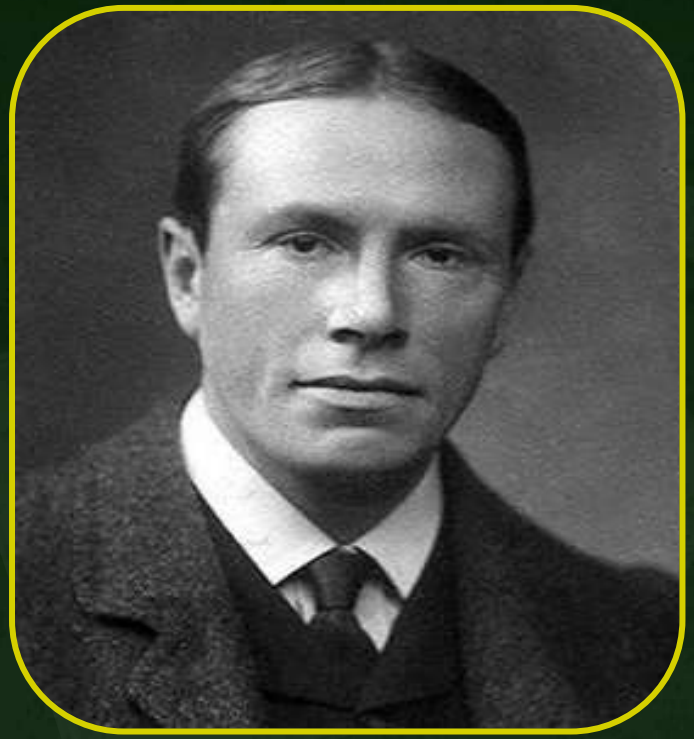
a) its genotype

b) its phenotype

c) factor

d) all of the above

Punnett Square

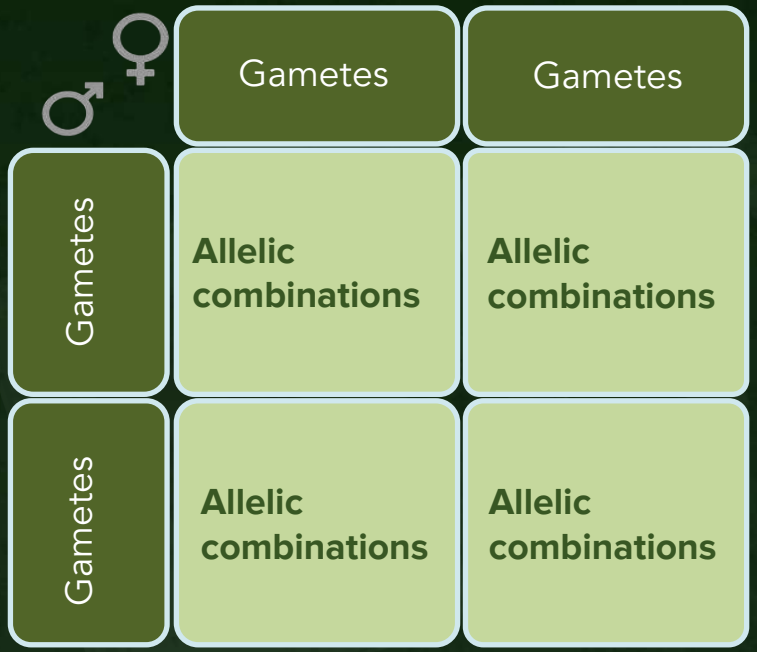


Reginald Crundall Punnett

- It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross.
- Reginald Crundall Punnett, an English geneticist, came up with the Punnett square in 1905.

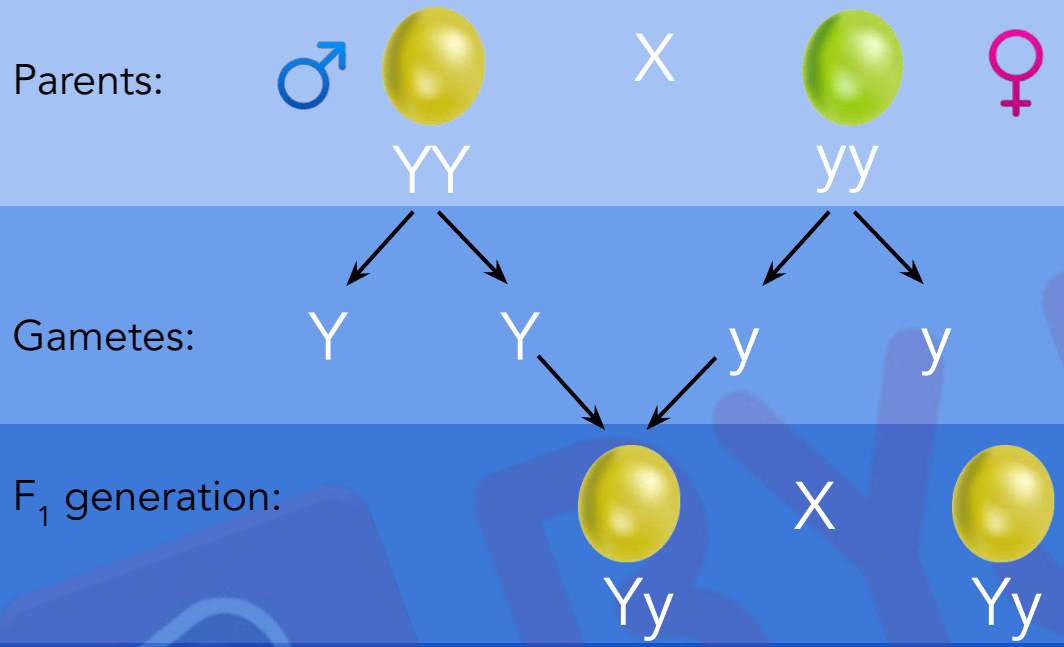
Punnett Square

Creating a Punnett square



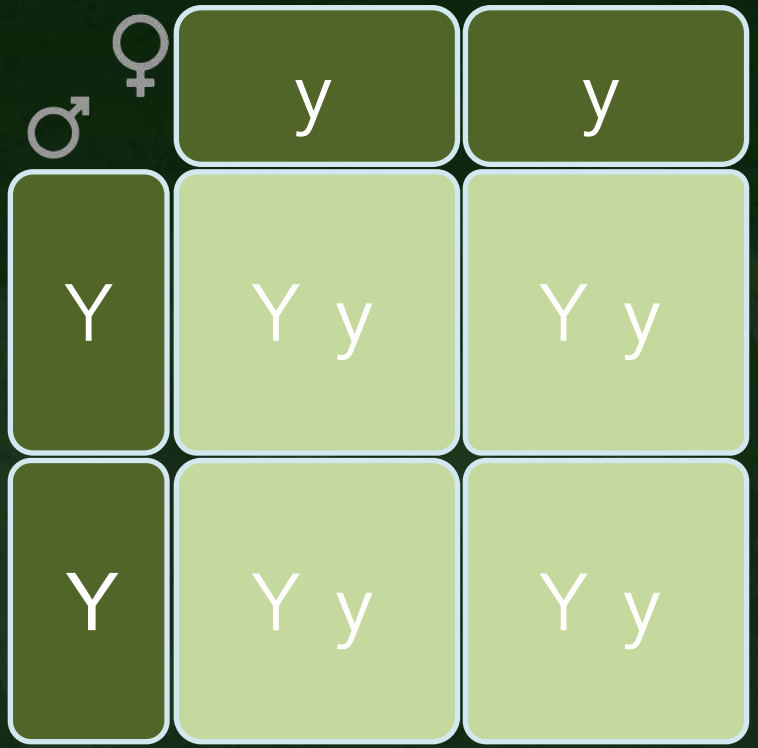
- First step: A checker box is drawn.
  - Hence, Punnett square method is also known as checkerboard method.
- Second step: Possible male and female gametes are written on top row and left column.
- Third step: All possible allelic combinations are then populated in the squares.

Mendel's Experiment



- Gametes from parents (yellow seed and green seed) fuse to form the F_1 generation
- All the F_1 generation seeds hence formed have the same phenotype and genotype
- Phenotype- Yellow seeded plant
- Genotype- Yy

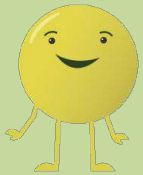
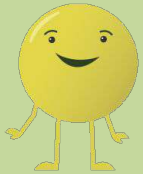
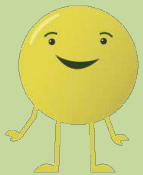
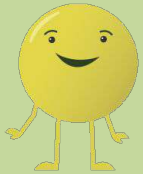
Punnett Square - Monohybrid Cross F_1



Genotype of F_1 generation

100% of F_1 generation has genotype Yy

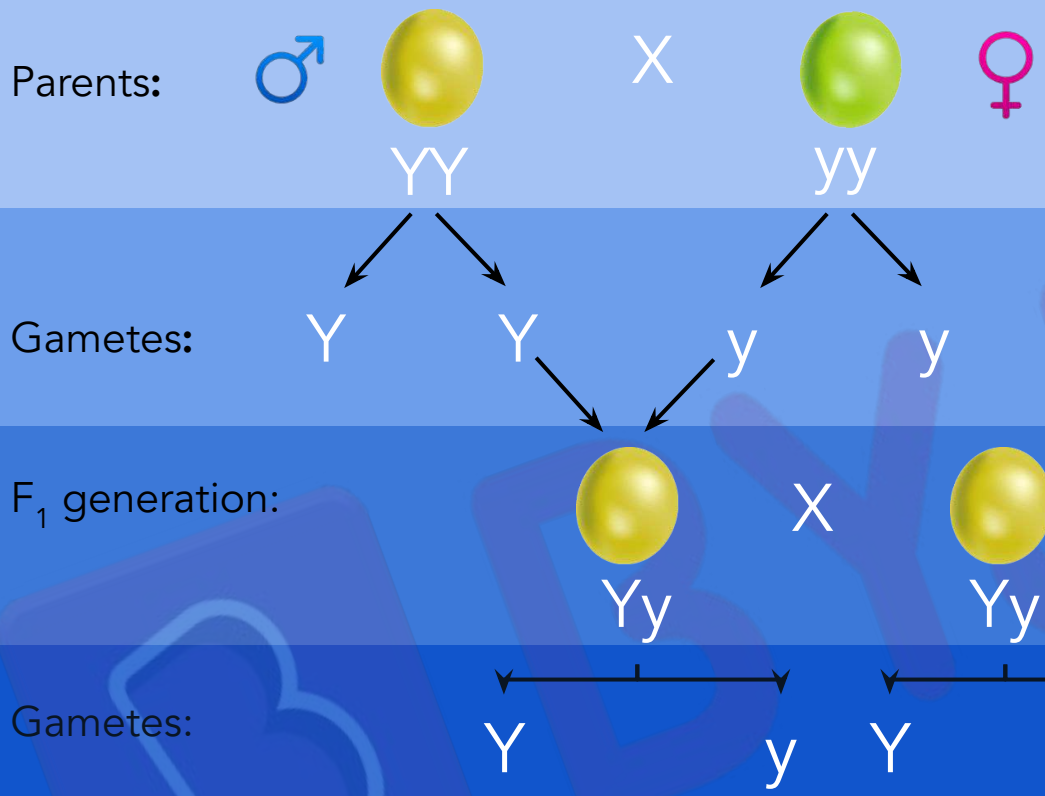
Punnett Square - Monohybrid Cross F_1

♂ \ ♀	y	y
Y		
Y		

Phenotype of F_1 generation

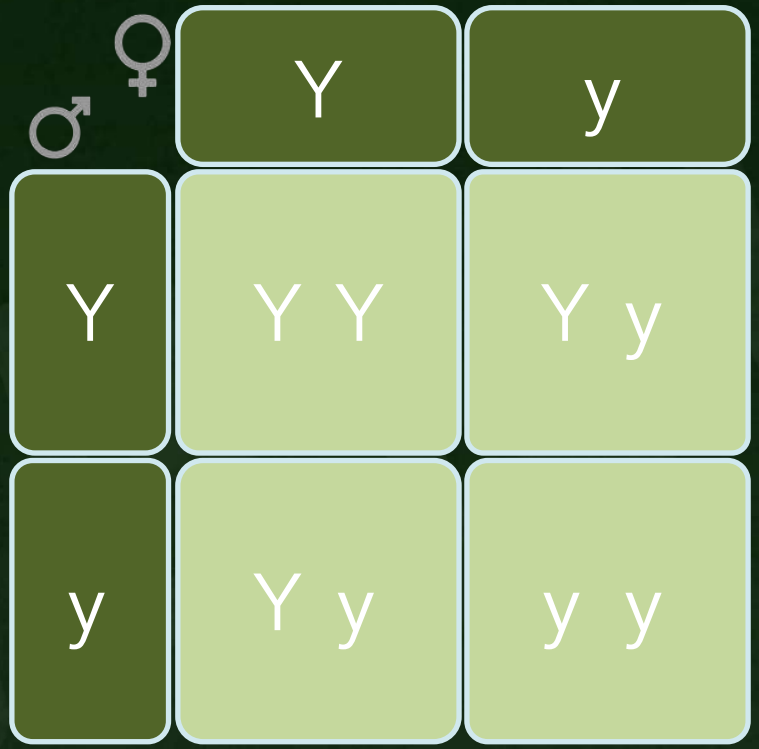
100% of F_1 generation has phenotype yellow peas

Mendel's Experiment

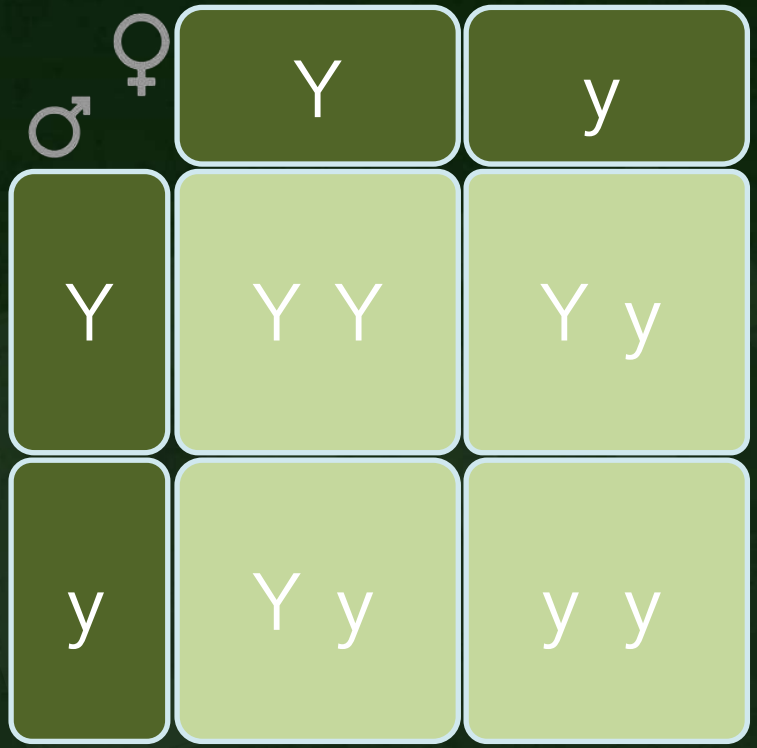


- F_1 generation then undergoes selfing and meiosis results in formation of 4 gametes.
- Further, the F_2 generation is formed.

Punnett Square - Monohybrid Cross F_2



Punnett Square - Monohybrid Cross F_2



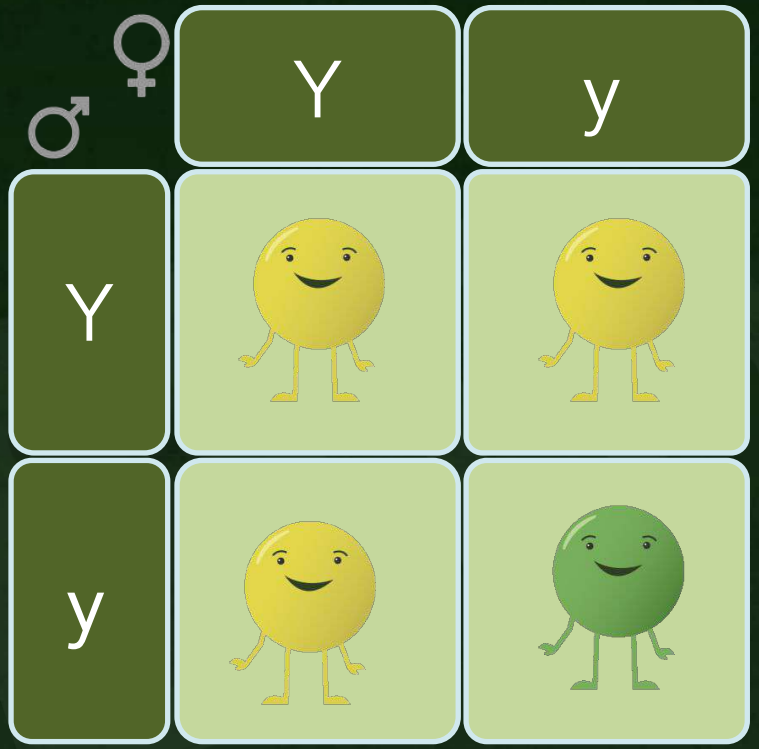
Genotype of F_2 generation

25% of F_2 generation has genotype YY

50% of F_2 generation has genotype Yy

25% of F_2 generation has genotype yy

Punnett Square - Monohybrid Cross F_2

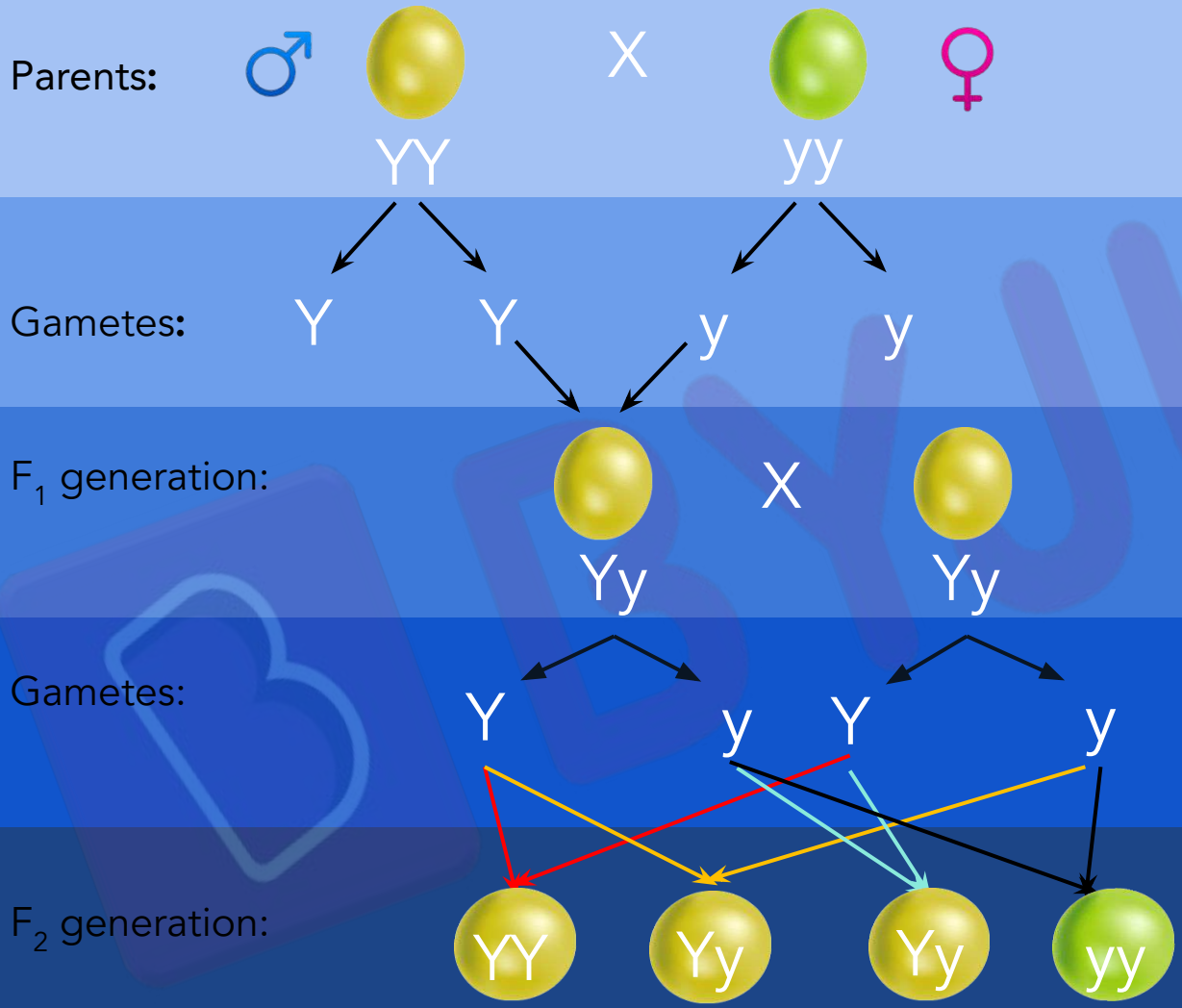


Phenotype of F_2 generation

75% of F_2 generation has phenotype yellow peas

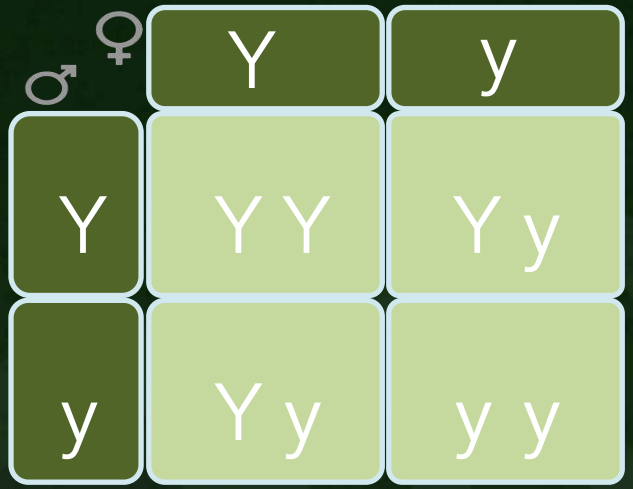
25% of F_2 generation has phenotype green peas

Mendel's experiment



- Finally, the F_2 generation is obtained.
- Phenotype - 3 yellow : 1 green
- Genotype - 1:2:1

Genotypic and Phenotypic Ratio



Genotypic ratio

YY = 1/4

Yy = 1/2

yy = 1/4

Genotypic ratio = $\frac{\text{Observed genotype}}{\text{All possible genotypes}}$

Genotypic ratio → YY : Yy : yy = 1 : 2 : 1

Binomial Expression

- If x and y are the two alleles (Y and y)
- If a and b are their frequency of appearance in offspring (½ Y or ½ y), the equation will be as follows:

$(ax + by)^2$

$= (½\ Y + ½\ y)^2$ (By substitution)

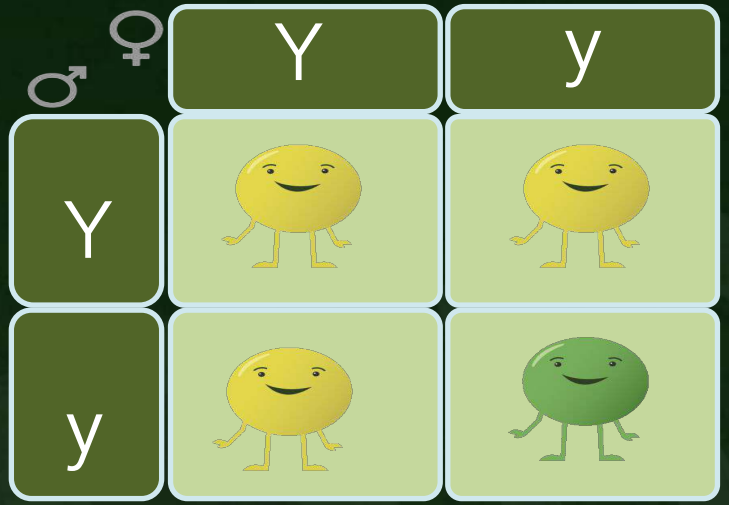
$= (½\ Y + ½\ y)\ X\ (½\ Y + ½\ y)$ (By expansion)

$= ¼\ YY + ½\ Yy + ¼\ yy$ (After solving)

Therefore, final ratio

YY : Yy : yy = 1 : 2 : 1

Phenotypic Ratio



Phenotypic ratio

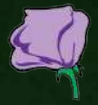
= 3/4

= 1/4

Phenotypic ratio = $\frac{\text{Observed phenotype}}{\text{All possible phenotypes}}$

Phenotypic ratio → Yellow : Green = 3 : 1

Genotypic and Phenotypic Ratio = Universal

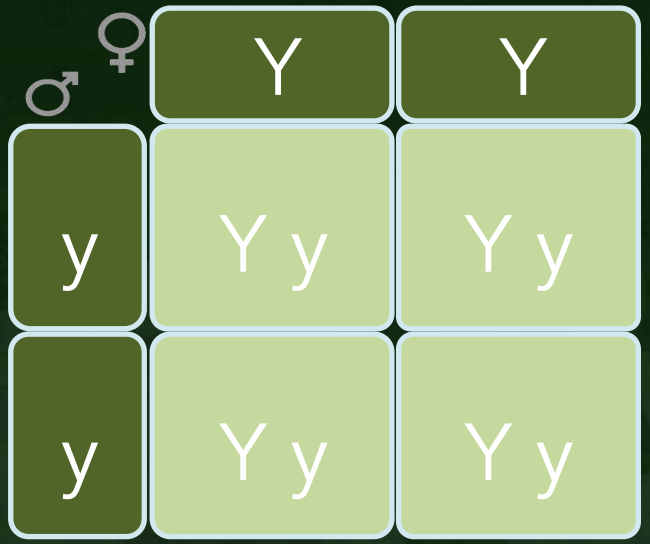
Seed	Flower	Pod		Stem
 Round	 Yellow	 Violet	 Inflated	 Green
 Wrinkled	 Green	 White	 Constricted	 Yellow

 Axial flower	 Tall	
 Terminal flower	 Dwarf	

- Mendel tried out the cross between plants differing in other traits.
- Similar results were obtained with the other traits that he studied: Only one of the parental traits was expressed in the F_1 generation while at the F_2 stage both the traits were expressed in the ratio 3:1.
- The contrasting traits did not show any blending at either F1 or F2 stage.

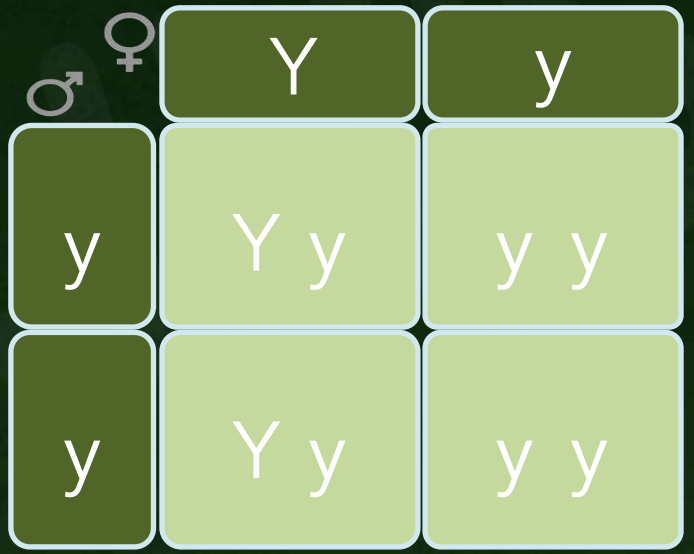
Punnett Square - Test Cross

Homozygous



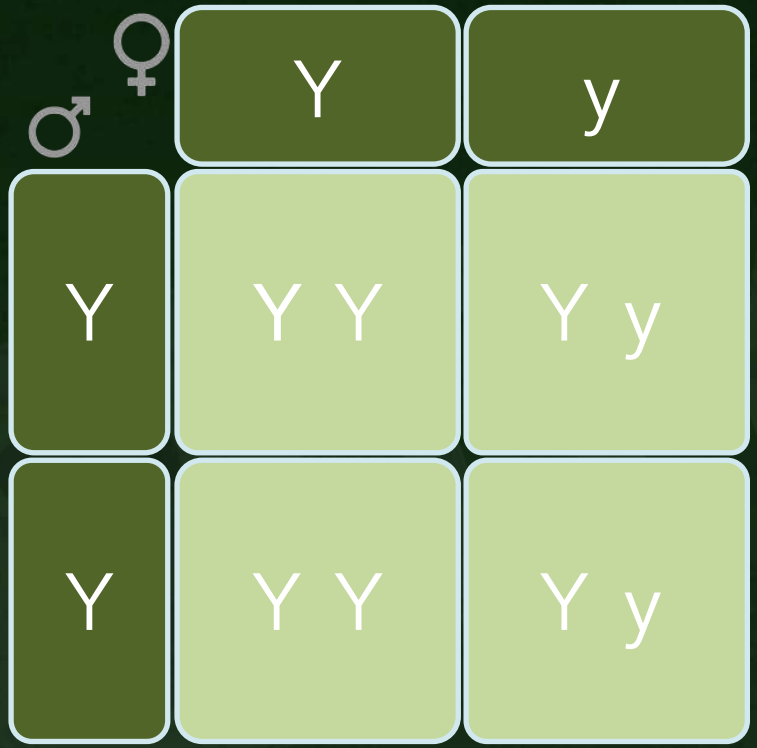
100% offspring are yellow;
Test plant is homozygous dominant

Heterozygous



50% offspring are yellow;
50% offspring are green;
Test plant is heterozygous dominant

Punnett Square - Back Cross

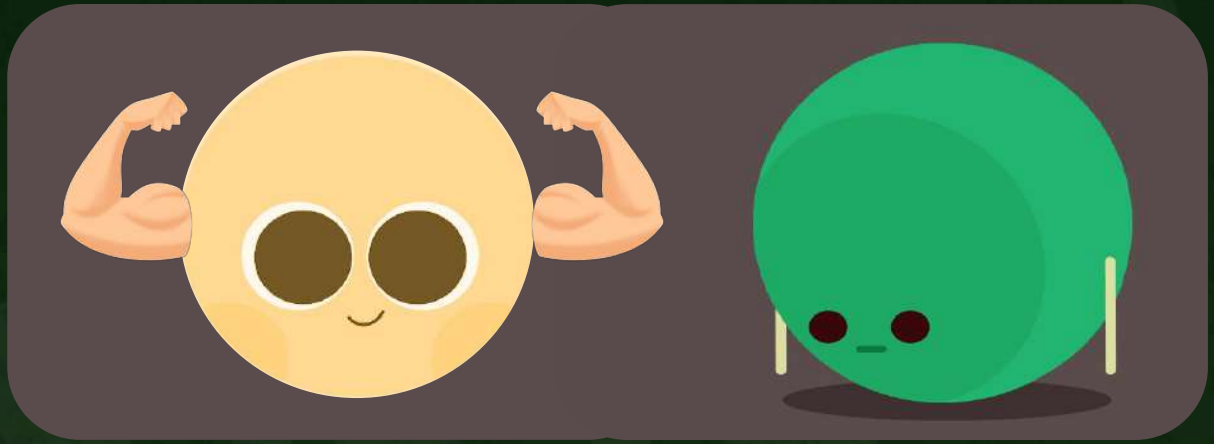


100% offspring are yellow

Law of Dominance

When the alleles are different or in heterozygous condition, then one dominates the other.

Law of Dominance



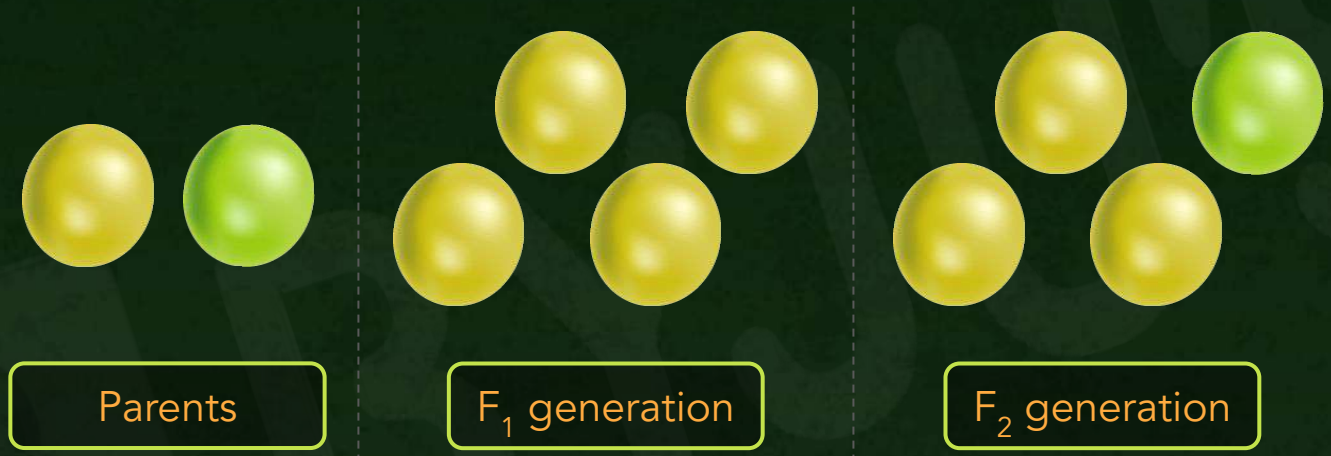
YY or Yy yy

- Allele for yellow colour pea (Y) is dominant over allele for green colour pea (y).
- Hence, all the heterozygous Yy progeny of F_1 were yellow coloured.
- Allele for green colour is recessive and hence it was seen only in F_2 generation when both the alleles were of y (yy genotype) in one of the four progenies.

Law of Segregation

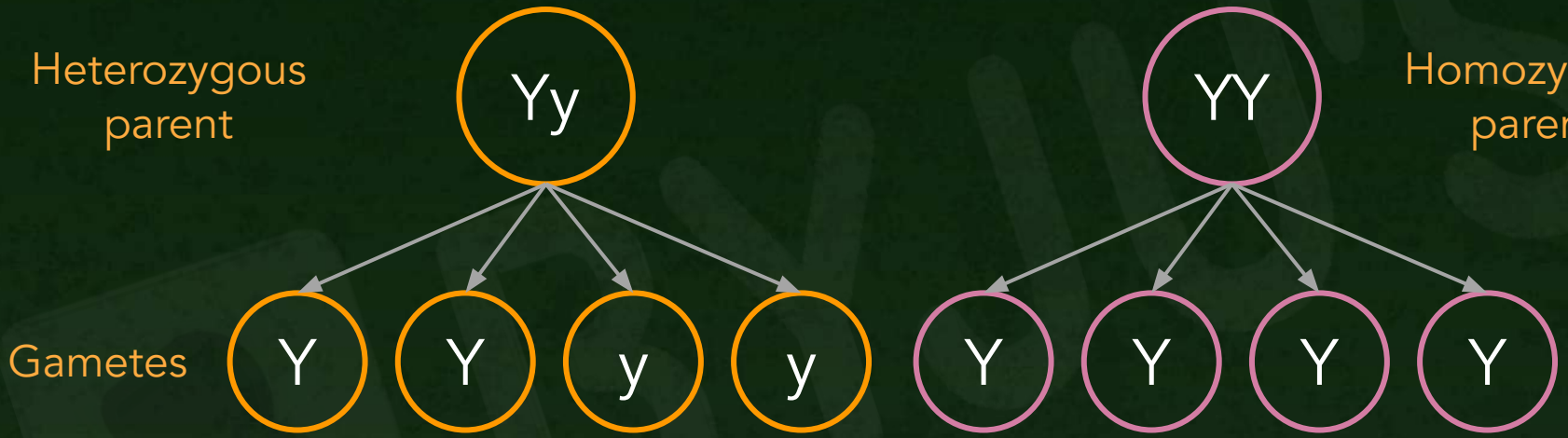
During gamete formation, pair of alleles segregate such that each gamete receives only one allele.

Law of Segregation



- Mendel proposed this law based on the observation that, there was no blending/mixing of alleles seen in any progeny.
- Though the parents contain two alleles before gamete formation, the alleles segregate from each other such that a gamete receives only one of the two alleles.

Law of Segregation



- A homozygous parent produces similar gametes.
- While a heterozygous one produces two kinds of gametes, each having one allele in equal proportion.

According to law of dominance, in a pair of dissimilar alleles

a) one allele dominates the other

b) both the alleles dominate each other

c) one allele masks the other

d) both a and c

According to law of dominance, in a pair of dissimilar alleles

a) one allele dominates the other

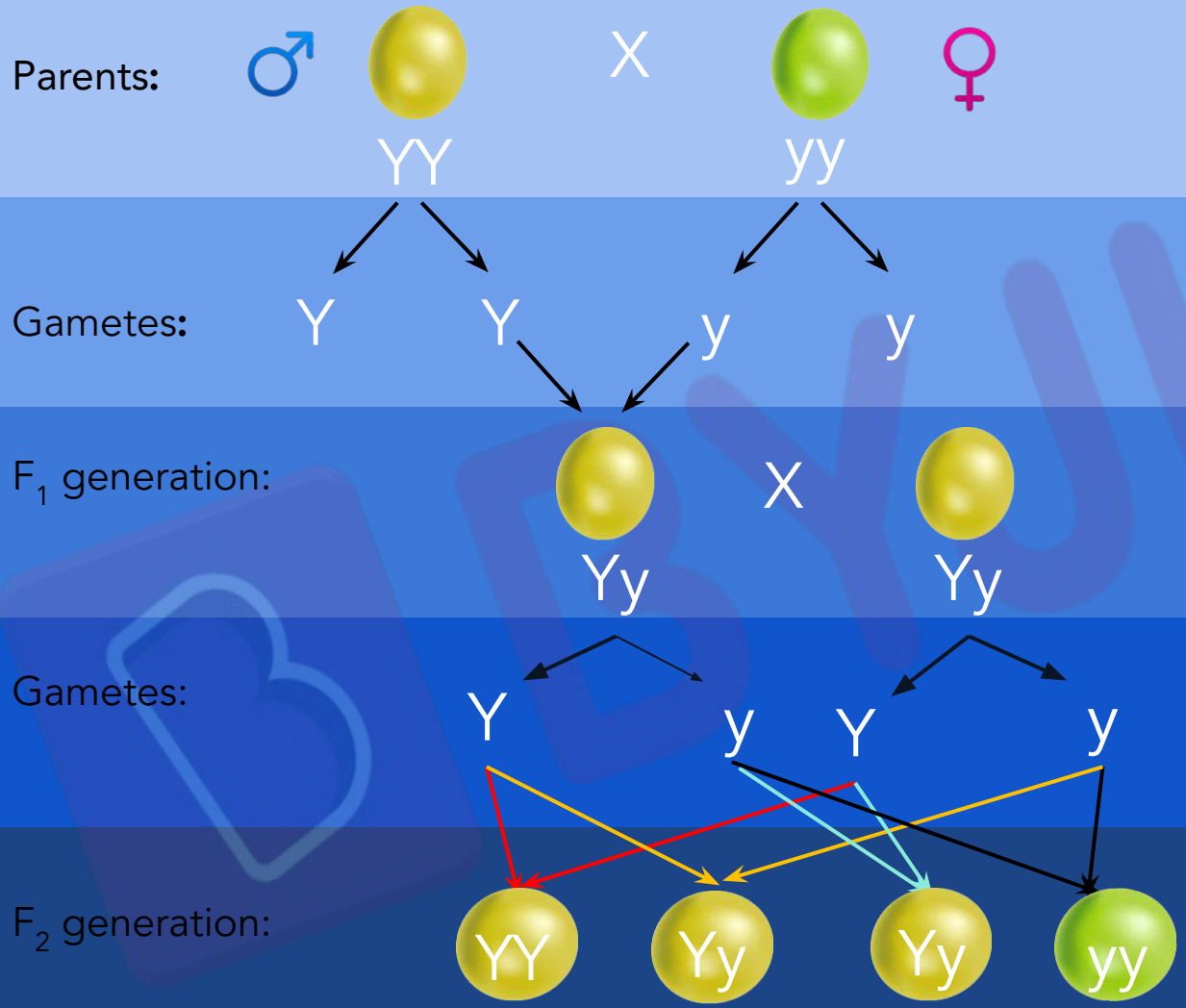
b) both the alleles dominate each other

c) one allele masks the other

d) both a and c

Summary

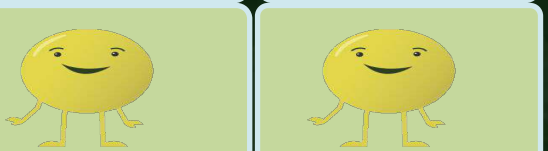
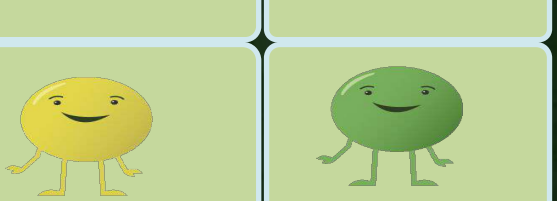
Mendel's experiment



- Finally, the F_2 generation is obtained.
- Phenotype - 3 yellow : 1 green
- Genotype - 1:2:1

Summary

Phenotypic and genotypic ratio

♂ \ ♀	Y	y
Y		
y		

Phenotypic ratio

Yellow : Green = 3 : 1

♂ \ ♀	Y	y
Y	Y Y	Y y
y	Y y	y y

Genotypic ratio

YY : Yy : yy = 1 : 2 : 1

Summary

Punnett square - Monohybrid cross F2

♂ \ ♀	Y	y
Y	 YY	 Yy
y	 Yy	 yy

Summary

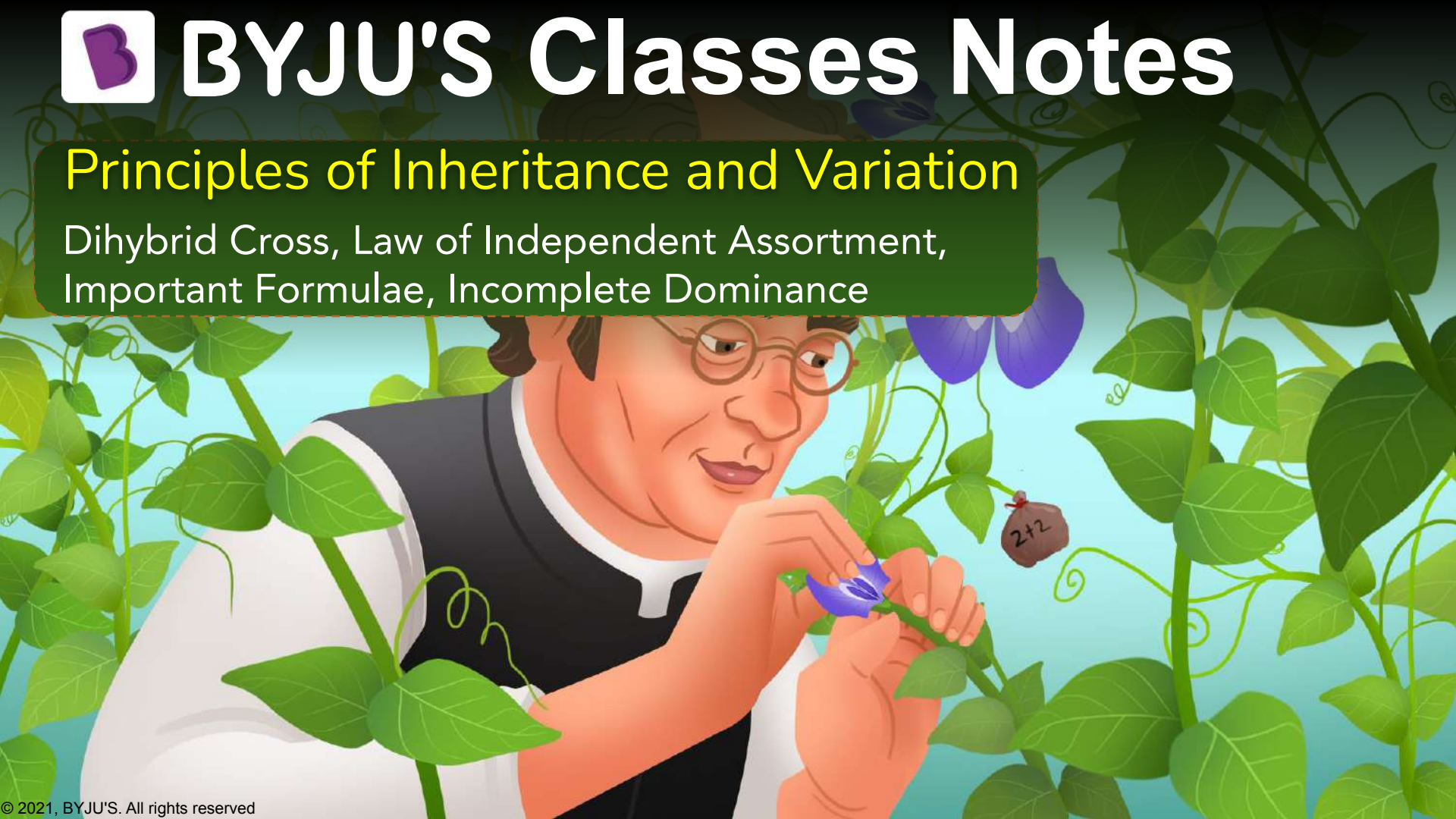
Genetic terms

- Genotype – Combination of alleles
- Phenotype – Observable trait
- Homozygous – Both the alleles are identical
- Heterozygous – Alleles are different
- Dominant – Allele which is expressed as phenotype during heterozygous condition
- Recessive – Allele which is not expressed as phenotype during heterozygous condition

BYJU'S Classes Notes

Principles of Inheritance and Variation

Dihybrid Cross, Law of Independent Assortment, Important Formulae, Incomplete Dominance



Key Takeaways

1. Dihybrid cross
   - Phenotypic ratio
   - Genotypic ratio
2. Law of independent assortment
3. Important formulae
4. Incomplete dominance

Summary

Recall! Contrasting Traits Used by Mendel

Pod


Inflated (Full)


Green


Constricted


Yellow

Seed


Round


Yellow


Wrinkled


Green

Stem


Axial flowers


Tall (6 - 7 ft)


Terminal flowers


Dwarf (3/4 - 1 ft)

Flower


Violet

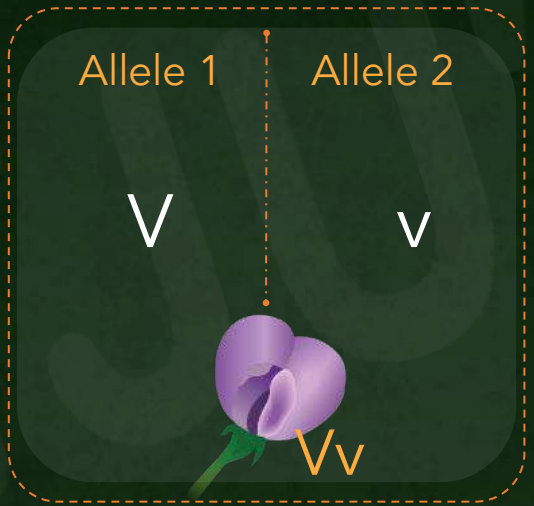

White

Recall! Monohybrid Cross

Colour of flower- White

Homozygous

Both the alleles code for same colour i.e. white in this case



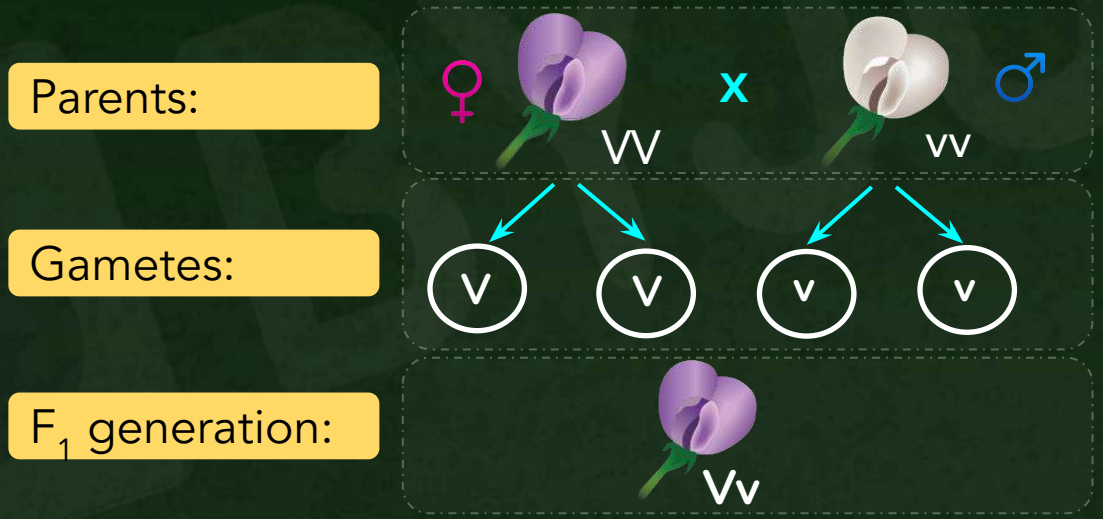
Colour of flower - Purple

Heterozygous

The two alleles code for different colours i.e. white and purple in this case

Recall! Law of Dominance

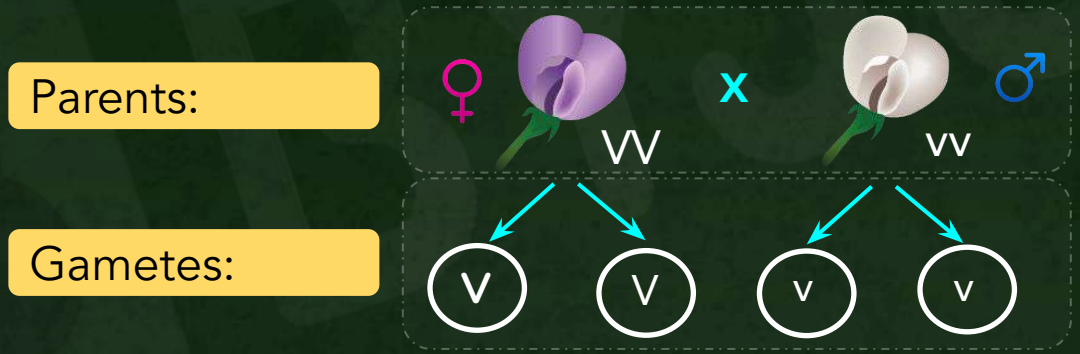
- When the alleles are different or in heterozygous condition, then one allele dominates the other .



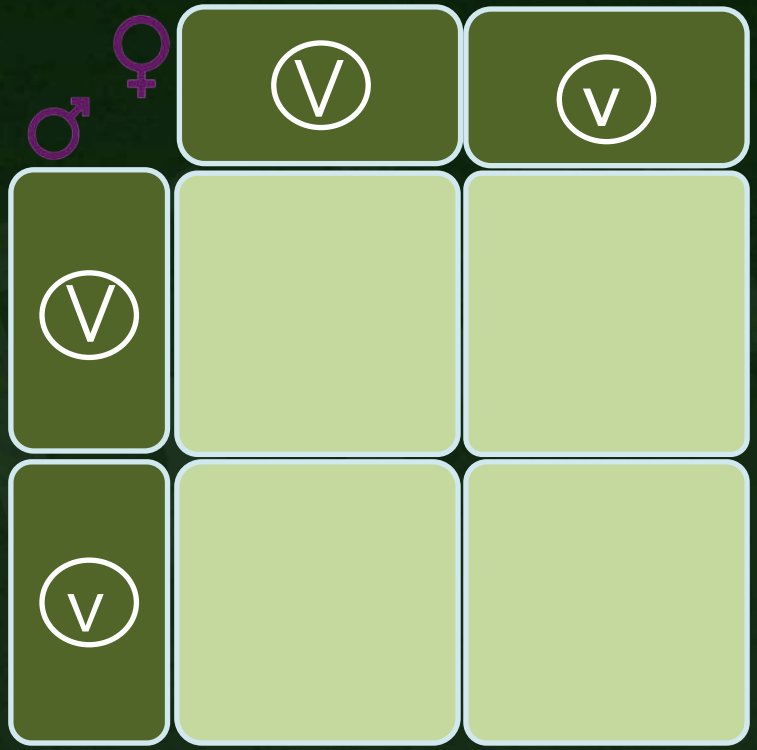
Purple is the dominant allele

Recall! Law of Segregation

- During gamete formation, pair of alleles segregate such that each gamete receives only one allele.



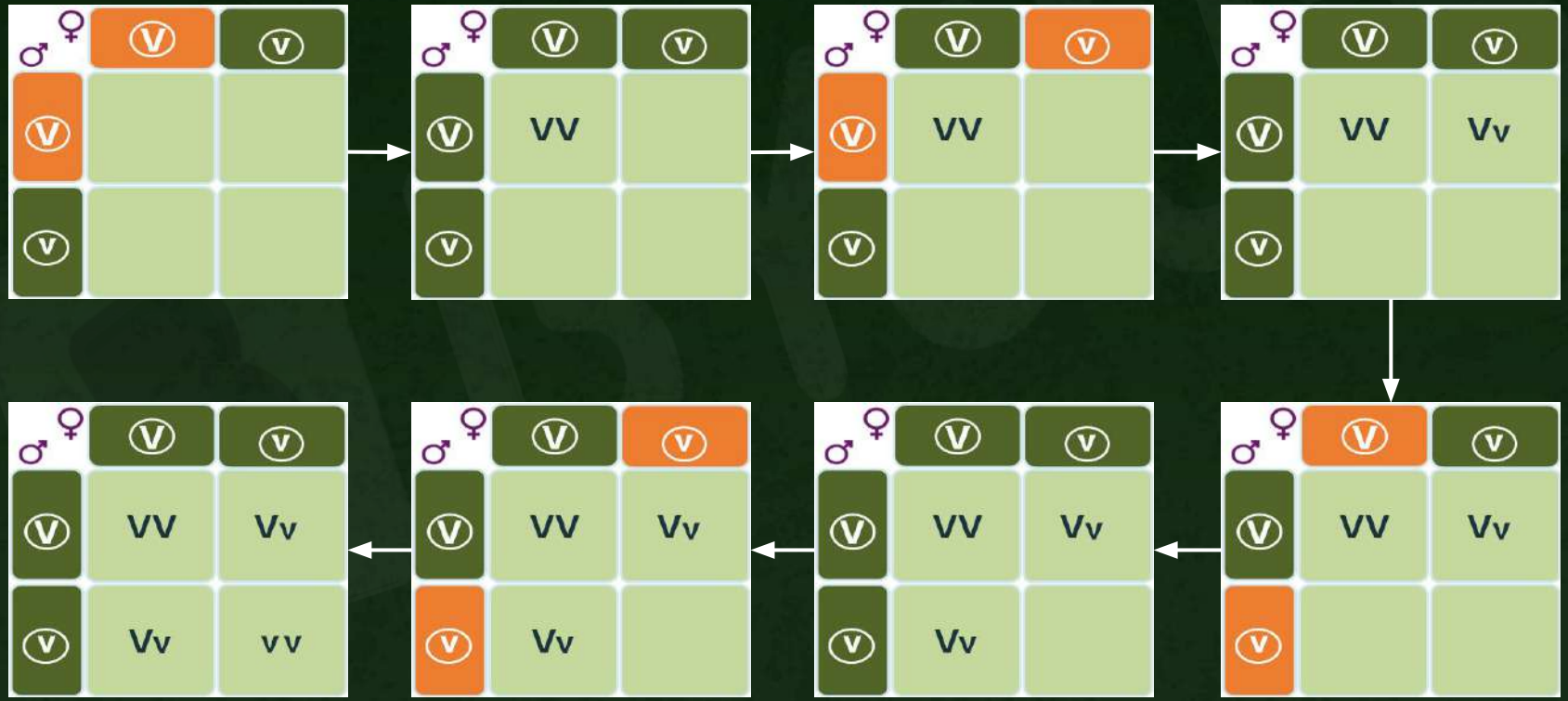
Recall! Punnett Square



- It is a method to depict Mendel's cross using checker boards.

Recall! The Punnett Square Method to Depict Mendel's Cross

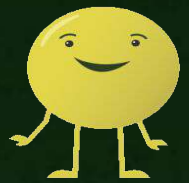
Vv x Vv



Dihybrid Cross

- It is a cross between two individuals with two different observable characters.
- In the monohybrid cross, plants used differed in one character.
- Mendel's next objective was to check whether these laws were applicable for each pair of differing characters when several different characters are united in the hybrid through fertilisation.

Dihybrid Cross

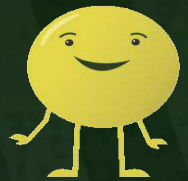


Parental generation

Pea plant with round, yellow seeds

X

Pea plant with wrinkled, green seeds

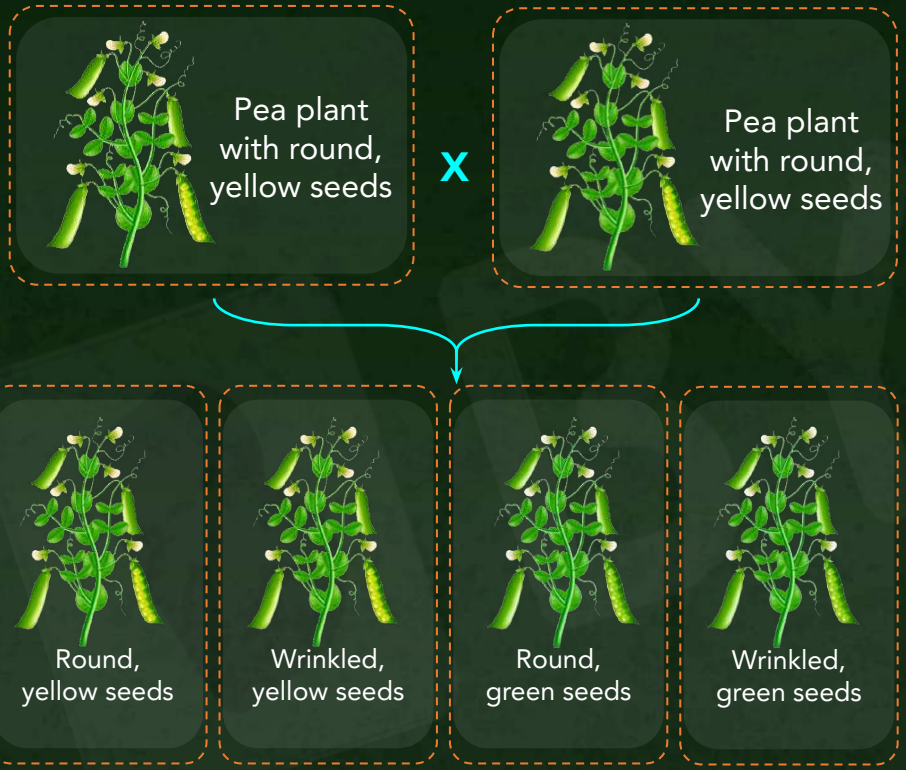


Pea plant with round, yellow seeds

F_1 generation

- Mendel chose pure lines/true breeds as parent plants.
- He chose pea plants producing round yellow, and wrinkled green peas to perform the cross.

Dihybrid Cross

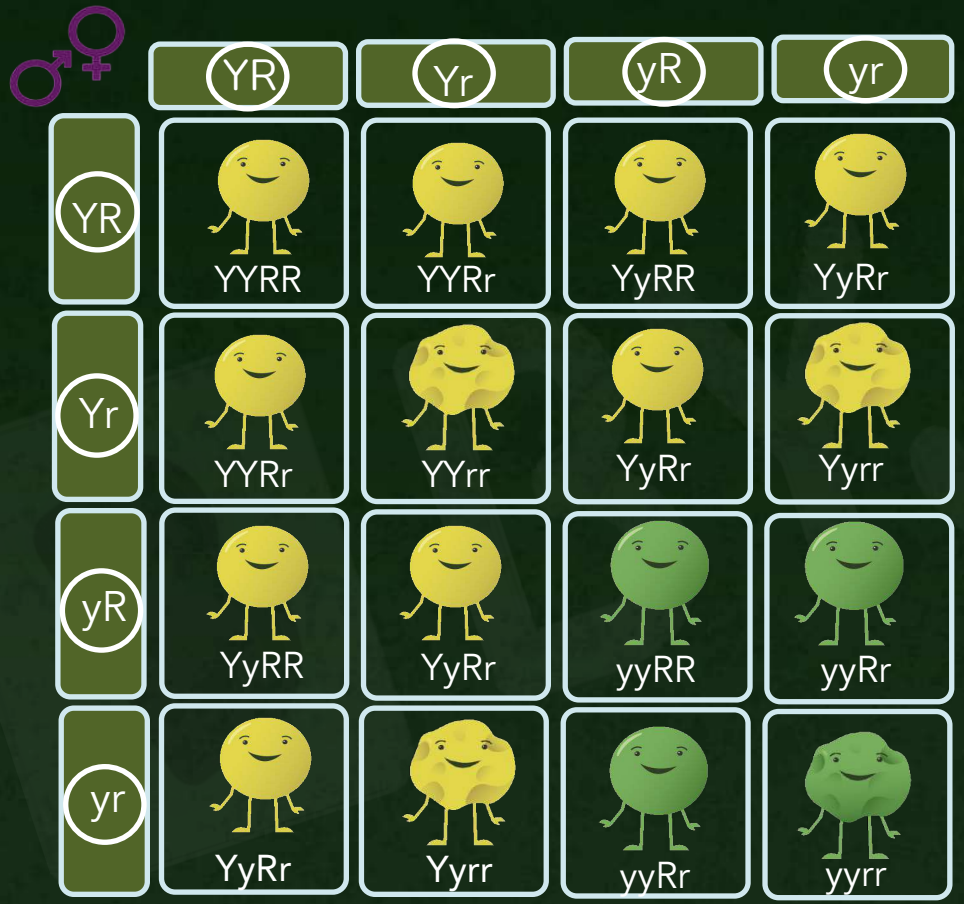


F_1 generation

F_2 generation

- He then self-fertilised the F_1 generation.
- He found that some plants had
  - yellow and round seeds
  - yellow and wrinkled seeds
  - green and round seeds
  - green and wrinkled seeds.

Dihybrid Cross

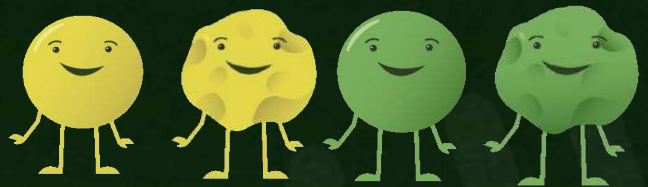


- The F_2 generation of this cross can be easily depicted via Punnett square.
- Parental gametes are written outside the box.
- F_2 generation is tabulated in boxes showing all phenotypes and genotypes.

Dihybrid Cross

F_2 generation

Phenotypic ratio

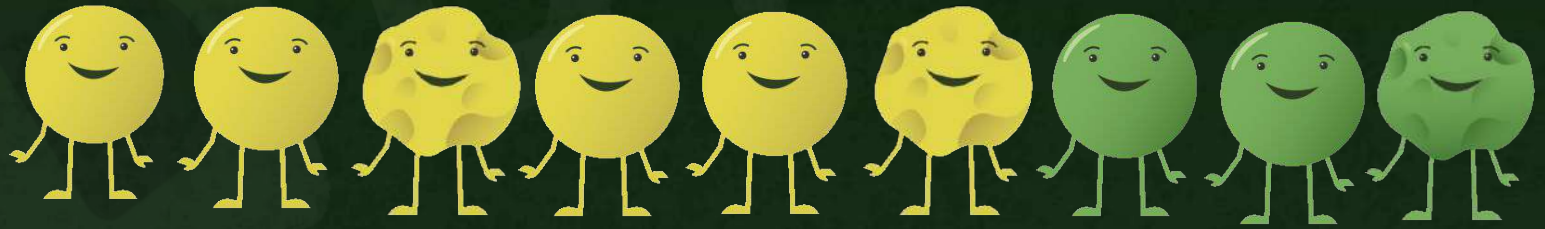


Round, yellow | Wrinkled, yellow | Round, green | Wrinkled, green

9 : 3 : 3 : 1

F_2 generation

Genotypic ratio



YYRR | YYRr | YYrr | YyRR | YyRr | Yyrr | yyRR | yyRr | yyrr

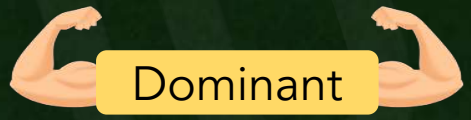
1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

Dihybrid Cross

Inference

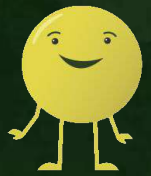
Based on the above observations, it can be concluded:

- Round seed is a dominant trait.
- Wrinkled seed is a recessive trait.



Recessive

- Round and yellow seeds hence form the dominant phenotype.
- Green and wrinkled seeds form the recessive phenotype.

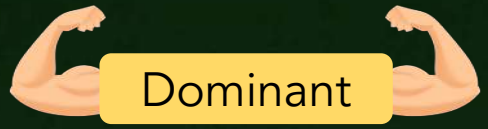


Round, yellow

Green, wrinkled

- Round green seeds, and round yellow seeds are also formed in intermediate proportions.
- This indicates that alleles of different characters are passed onto gametes independently of each other.

Dihybrid Cross



Dominant

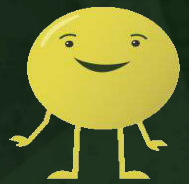
Recessive

Y

R

y

r



YYRR

yyrr

The alleles are represented in form of the letters.

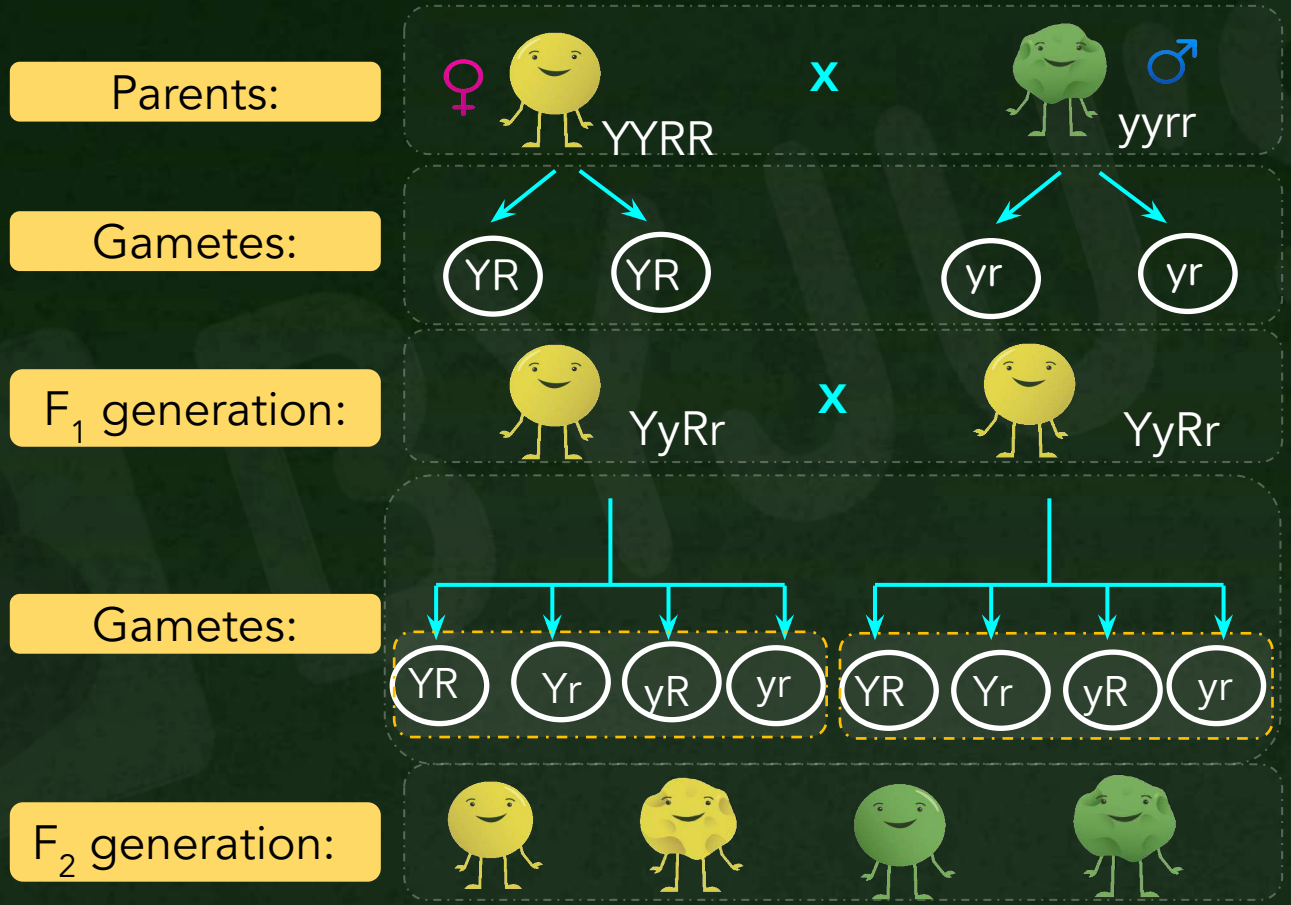
These are the dominant and recessive variants respectively.

Law of Independent Assortment

Based on the observations of dihybrid cross and the inferences drawn, Mendel proposed the law of independent assortment which states that:

"When two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters".

Dihybrid Cross



Dihybrid Cross

- Consider the segregation of one pair of genes R and r.
- 50% of the gametes have the gene R and the other 50% have r.
- Each gamete having either R or r allele should also have the allele Y or y.
- The segregation of 50% R and 50% r is independent of the segregation of 50% Y and 50% y.
- Thus, there are four genotypes of gametes (four types of pollen and four types of eggs).
- The four types are RY, Ry, rY and ry each with a frequency of 25% or 1/4th of the total gametes produced.

A cross was performed between tall plant (dominant, heterozygous trait) with green seeds and dwarf plant with yellow seeds (dominant, heterozygous trait), what is the phenotypic ratio of offspring that are dwarf and yellow?

A 3 : 16

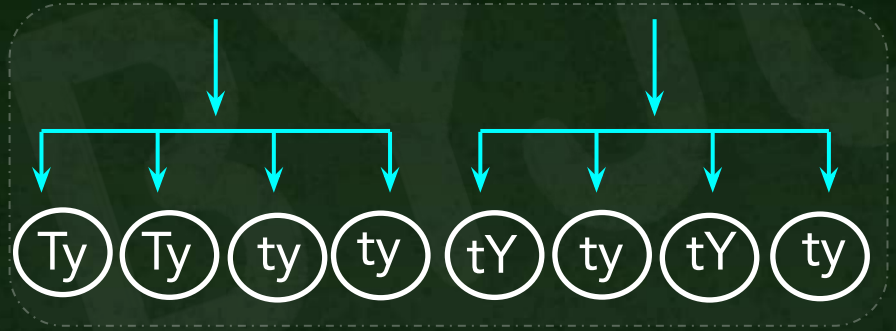
B 5 : 8

C 1 : 4

D 7 : 16

Parents:

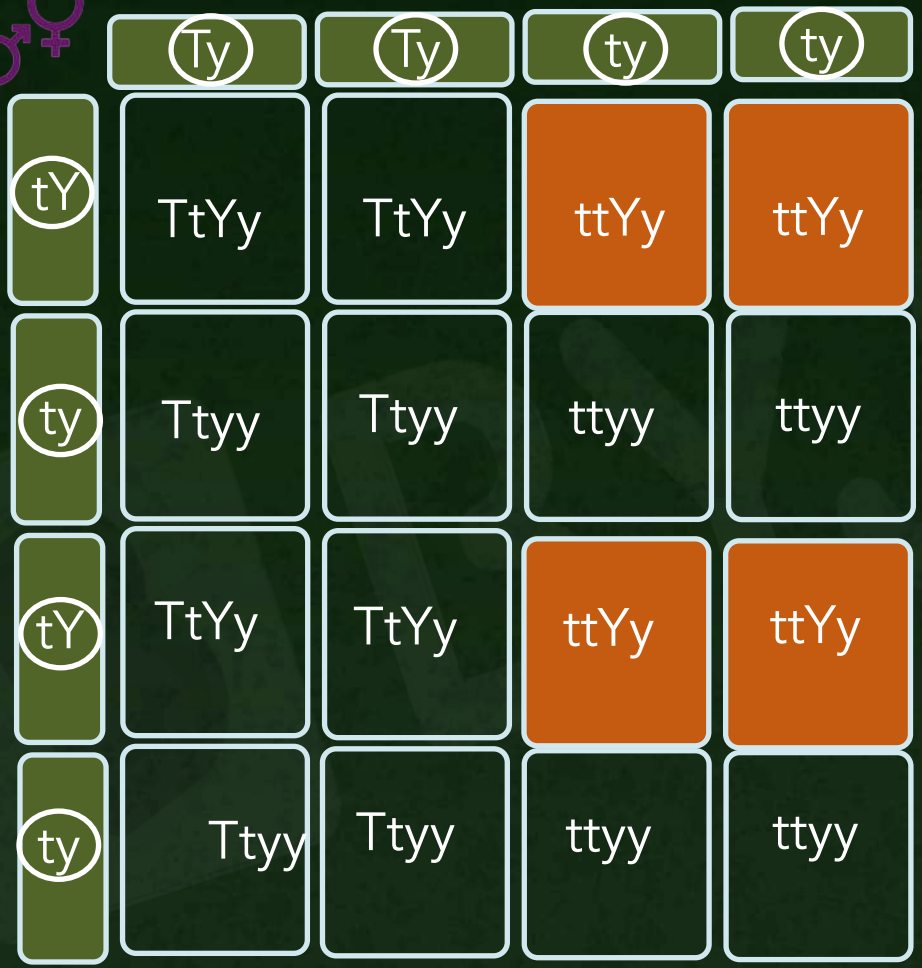
Gametes:



	Ty	Ty	ty	ty
tY	TtYy	TtYy	ttYy	ttYy
ty	Ttyy	Ttyy	ttyy	ttyy
tY	TtYy	TtYy	ttYy	ttYy
ty	Ttyy	Ttyy	ttyy	ttyy

Phenotypic Ratio (dwarf and yellow):

4 : 16



Phenotypic Ratio:

1 : 4

As phenotypic ratio of dwarf and yellow plants is asked, it is 4 : 16 = 1 : 4.

A cross was performed between tall plant (dominant, heterozygous trait) with green seeds and dwarf plant with yellow seeds (dominant, heterozygous trait), what is the phenotypic ratio of offspring that are dwarf and yellow?

A 3 : 16

B 5 : 8

C 1 : 4

D 7 : 16

Important Formulae

Types of gametes = 2^n — For all gametes

Types of phenotypes = 2^n

Types of genotypes = 3^n

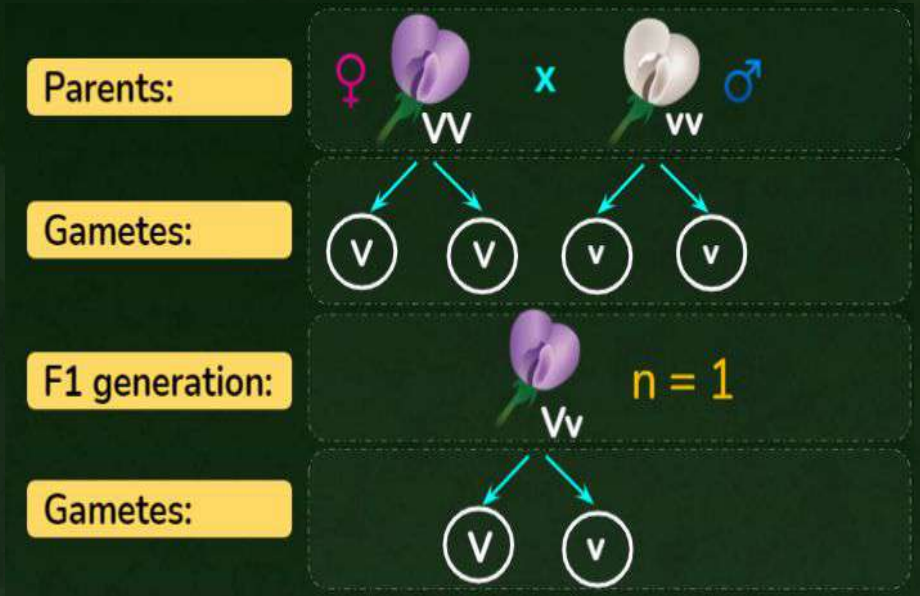
} Only in case of self-fertilisation

where, n = number of heterozygous gene pairs

- These formulae are used to calculate the type and number of gametes, phenotypes and genotypes based on number of heterozygous gene pairs present.

Monohybrid Cross

Types of gametes = 2^n

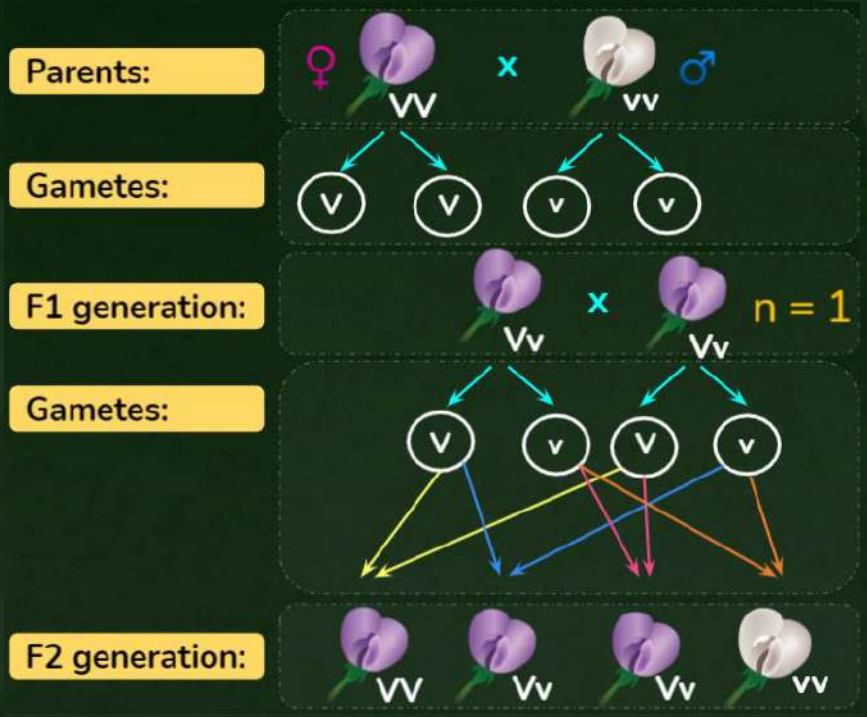


- In parents, no heterozygous pair is seen.
- So, the equation will be 2^0= 1(only 1 type of gamete seen)
- One heterozygous pair is obtained after F_1 generation - Vv
- n=1
- Types of gametes = $2^n = 2^1 = 2$

Types of gametes = 2

Monohybrid Cross

Types of phenotypes = 2^n

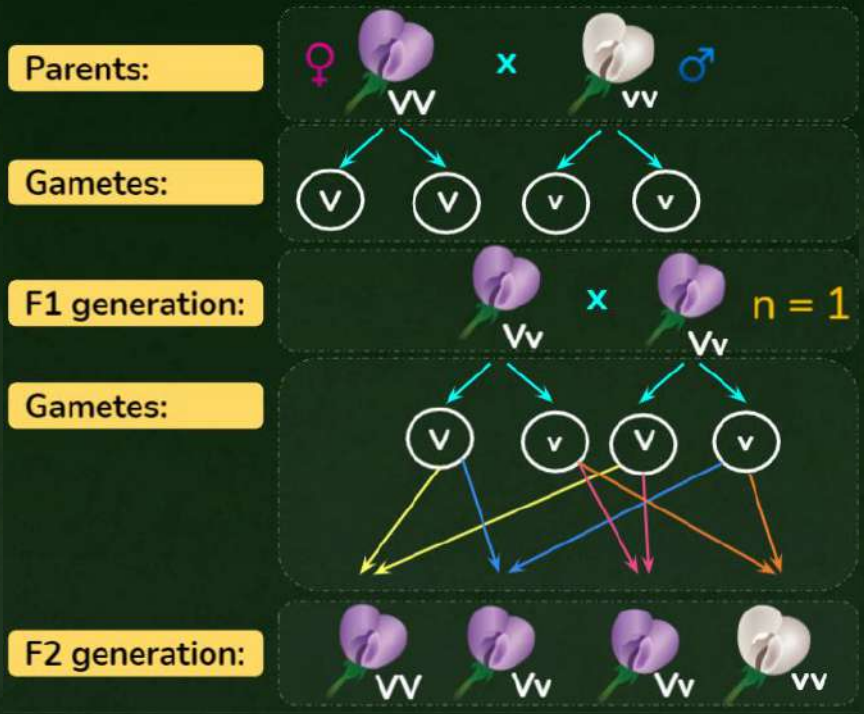


- One heterozygous pair is obtained after F_1 generation - Vv.
- n=1
- $2^n = 2^1 = 2$

Types of phenotypes = 2

Monohybrid Cross

Types of genotypes = 3^n

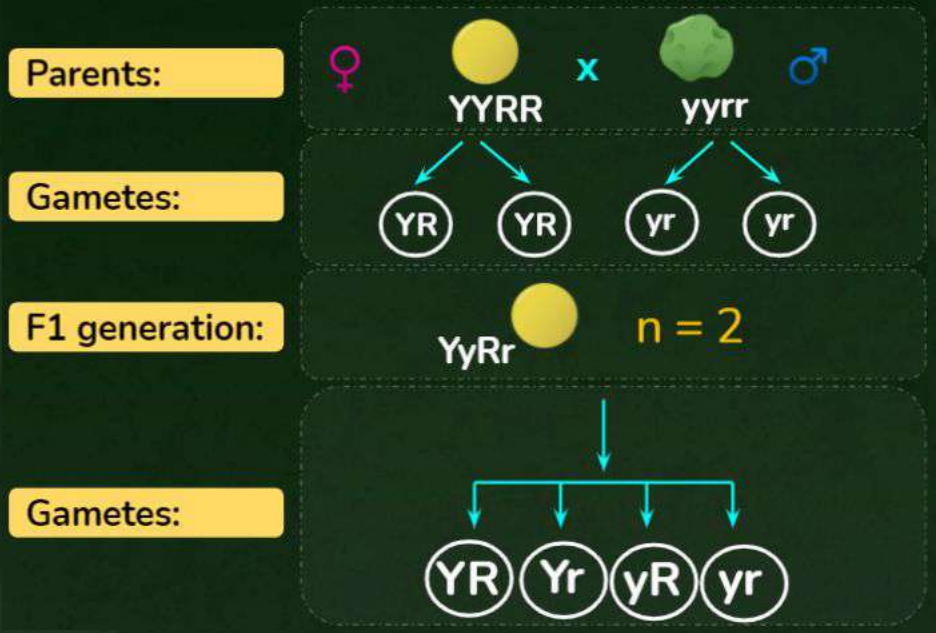


- One heterozygous pair is obtained after F_1 generation (Vv).
- n=1
- $3^n = 3^1 = 3$
- So, the types of genotypes are VV, Vv and vv.

Types of genotypes = 3

Dihybrid Cross

Types of gametes = 2^n

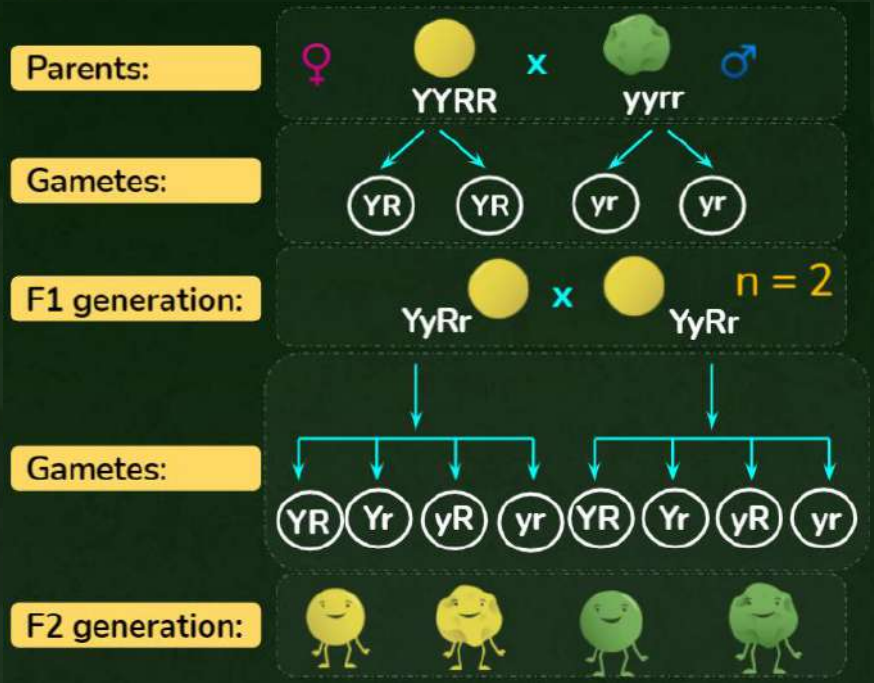


- Two heterozygous gene pairs are obtained after F_1 generation, Yy and Rr.
- $2^n = 2^2 = 4$

Types of gametes = 4

Dihybrid Cross

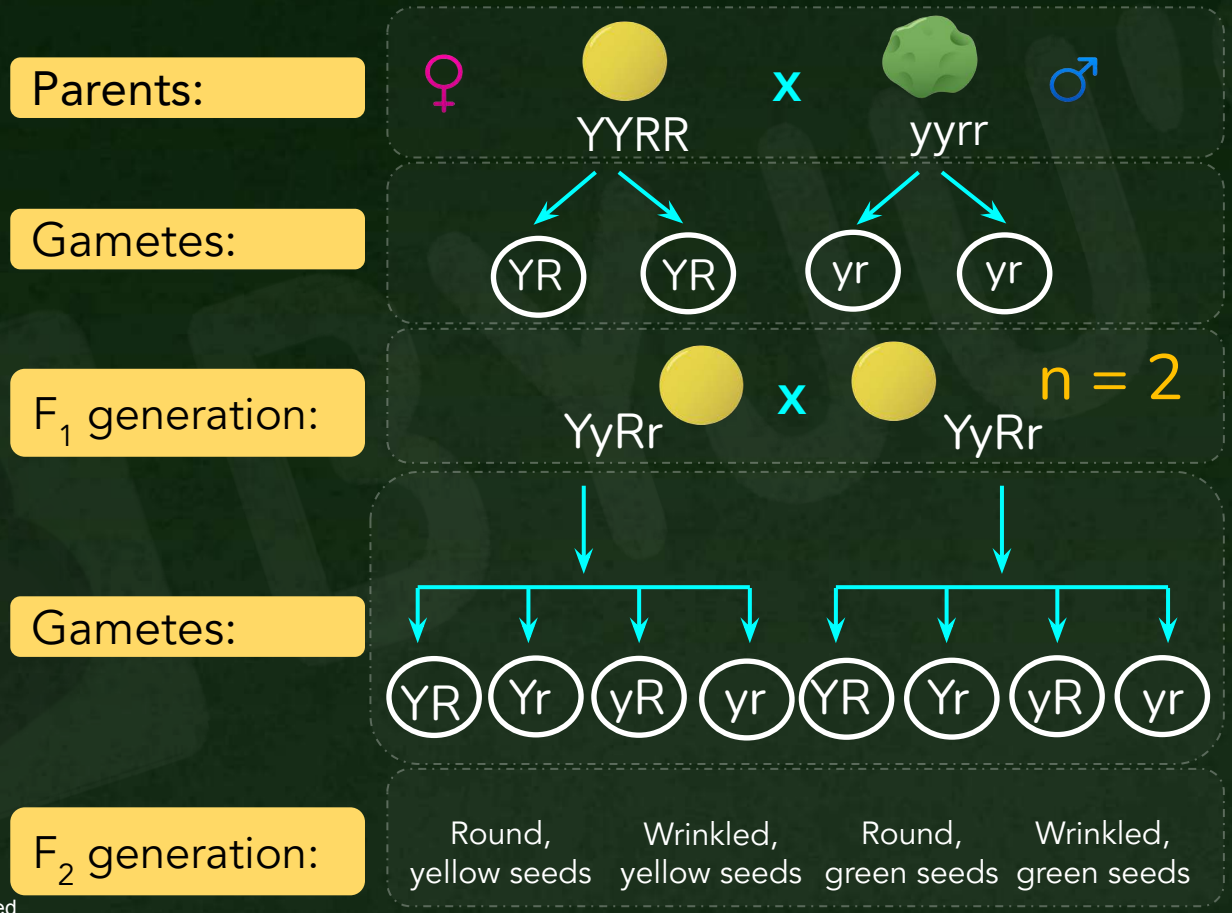
Types of phenotypes = 2^n



- Two heterozygous pairs are obtained after F_1 generation, Yy and Rr.
- $2^n = 2^2 = 4$

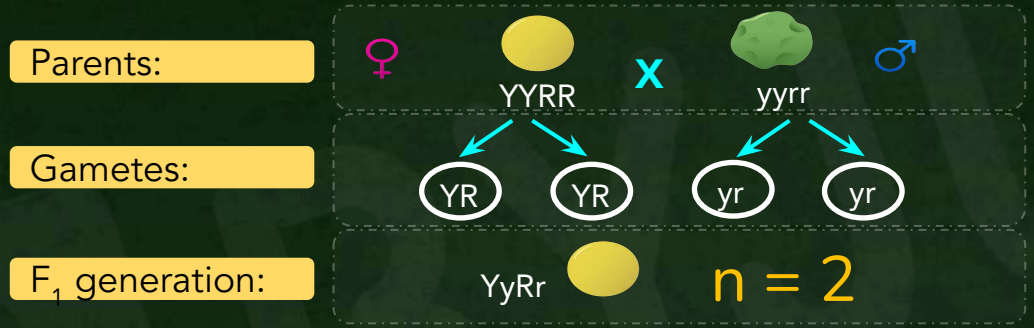
Types of phenotypes = 4

Dihybrid Cross

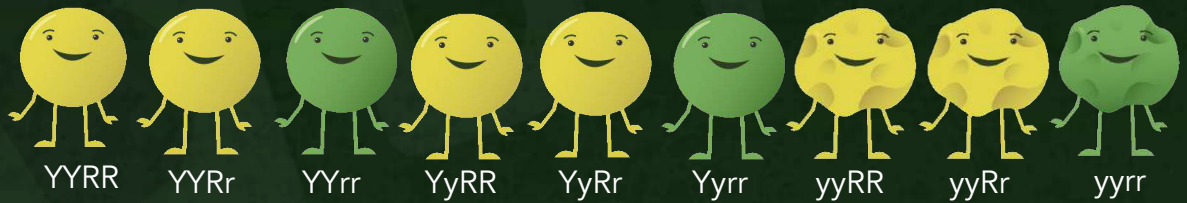


Dihybrid Cross

Types of genotypes = 3^n



- Two heterozygous pairs are obtained after F_1 generation, Yy and Rr.



Types of genotypes = 9

Use the probability method to calculate the number of gametes and genotypes, respectively, in a self-cross of AaBbCc parents.

A 16; 32

B 4; 9

C 8; 27

D 12; 18

Types of gametes = 2^n

Types of genotypes = 3^n

where n = number of heterozygous gene pairs = 3

Aa Bb Cc

Types of gametes = 2^3

Types of genotypes = 3^3

where n = 3

Types of gametes = 8

Types of genotypes = 27

Use the probability method to calculate the number of gametes and genotypes, respectively, in a self-cross of AaBbCc parents.

A. 16; 32

B. 4; 9

C. 8; 27

D. 12; 18

Exceptions to Mendel's Laws

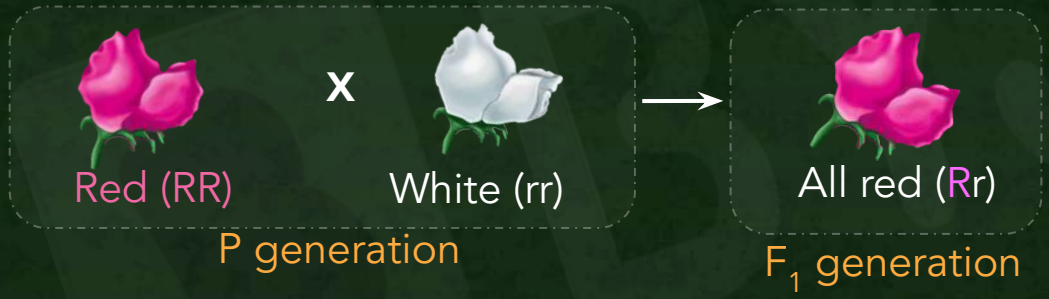
- Incomplete dominance
- Co-dominance
- Multiple alleles

Incomplete Dominance

Let us consider a snapdragon flower. They are:

- red coloured in homozygous dominant condition
- white coloured in homozygous recessive condition

Now, if these two parents are crossed, then according to Mendel's laws, we should get red flowers in F_1 generation.



- Similarly, according to Mendel's laws, F_2 generation should have the phenotypic ratio of 3:1 for red:white flowers, as depicted here.

However, this is not the case in snapdragon.

F_2 generation

♂ \ ♀	R	r
R	RR	Rr
r	Rr	rr

Phenotypic ratio (F_2) -	Red : White 3 : 1
Genotypic ratio (F_2) -	RR : Rr : rr 1 : 2 : 1

Incomplete Dominance

- Instead, in F_1 generation, all flowers obtained are pink in color.
- This is an intermediate phenotype which hints at blending of traits, a theory rejected by Mendel's experimental results.

- Similarly, in F_2 generation, the phenotypic ratio matches the genotypic ratio, where both the original parental type and the F_1 phenotypes appear.

F_2 generation

♂ \ ♀	R	r
R	RR	Rr
r	Rr	rr

Phenotypic ratio (F_2) -	Red	Pink	White
	1	2	1

Genotypic ratio (F_2) -	RR	Rr	rr
	1	2	1

Incomplete Dominance

Incomplete dominance is a form of intermediate inheritance in which one allele for a particular trait is not expressed completely over its paired allele.

Incomplete Dominance

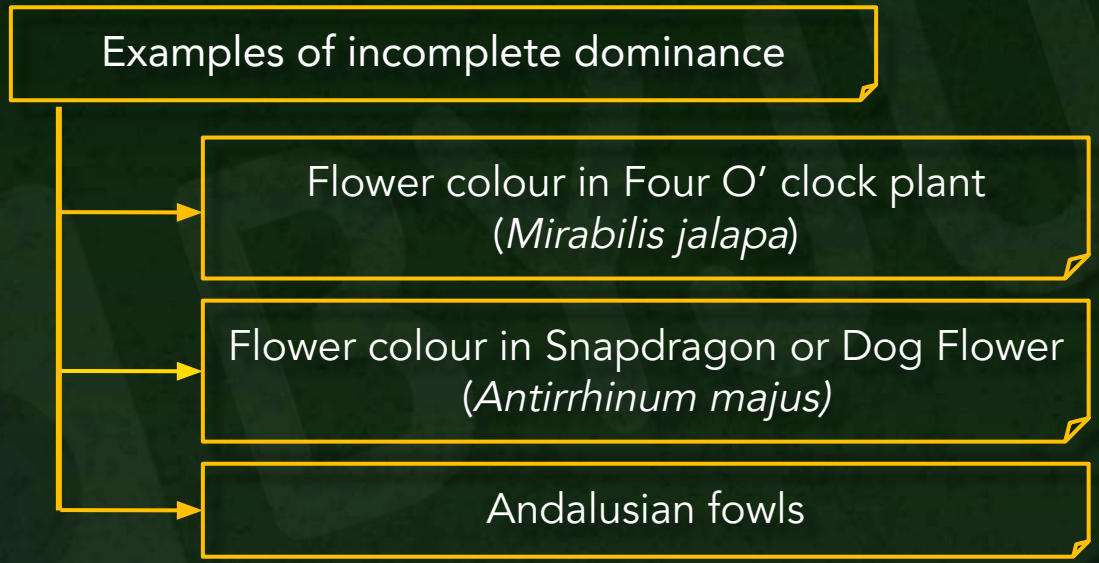
Comparison

	Complete dominance	Incomplete dominance
F_2 generation	Phenotypic ratio	Phenotypic ratio
		
	Red : White	Red : Pink : White
	3 : 1	1 : 2 : 1
	Genotypic ratio	Genotypic ratio
	RR : Rr : rr	RR : Rr : rr
	1 : 2 : 1	1 : 2 : 1

Incomplete Dominance

- It describes the general situation in which the phenotype of a heterozygote is intermediate between the two homozygotes on some quantitative scale of measurement.
- None of the two alleles of a gene is completely dominant over each other.
- When both alleles are present together, a new phenotype is formed.
- New phenotype is intermediate between the independent expression of the two alleles.

Incomplete Dominance



Incomplete Dominance

- German botanist Carl Correns first observed this phenomenon in the color of the four o'clock flower (*Mirabilis jalapa*).
- A homozygous red-flowered four-o'clock plant was crossed to a homozygous white-flowered plant.
- The wild type allele for red flower color is designated C_R and the white allele is C_W.

Incomplete Dominance

Examples

Law of dominance is not applicable in incomplete dominance.

Mirabilis jalapa
(Four o'clock)

Snapdragon / *Antirrhinum majus*
(Dog flower)

Incomplete Dominance

- *Antirrhinum* is a genus of plants commonly known as dragon flowers or snapdragons because of the flowers' fancied resemblance to the face of a dragon that opens and closes its mouth when laterally squeezed.

Incomplete Dominance

- Andalusian fowls have two pure forms, black and white.
- If the two forms are crossed, F1 individuals appear blue coloured due to occurrence of fine alternate black and white stripes on the feathers.
- F2 generation produces three types of fowls— 1 black: 2 blue: 1 white.

Incomplete Dominance

Parents:

Pure black
BB

×

Pure white
bb

F_1 generation:

Blue
Bb

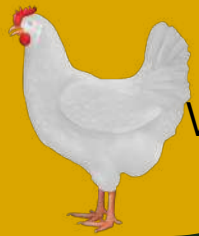
Mating among F_1 generation

F_2 generation:

Black
BB

Blue
Bb

Blue
Bb



White
bb

Summary

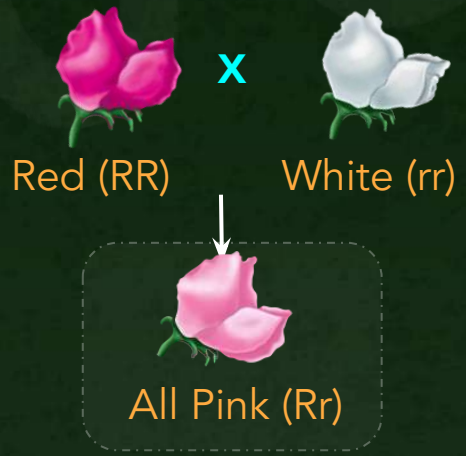
- Dihybrid cross
  - A dihybrid cross is a breeding experiment between two individuals that differ in two different characters.

- Law of independent assortment
  - It states that “during a dihybrid cross (crossing of two pairs of traits), an assortment of each pair of traits is independent of the other”. In other words, during gamete formation, one pair of traits segregates from another pair of traits independently.

Summary

- Incomplete dominance

Incomplete dominance is a form of intermediate inheritance in which one allele for a particular trait is not expressed completely over its paired allele.



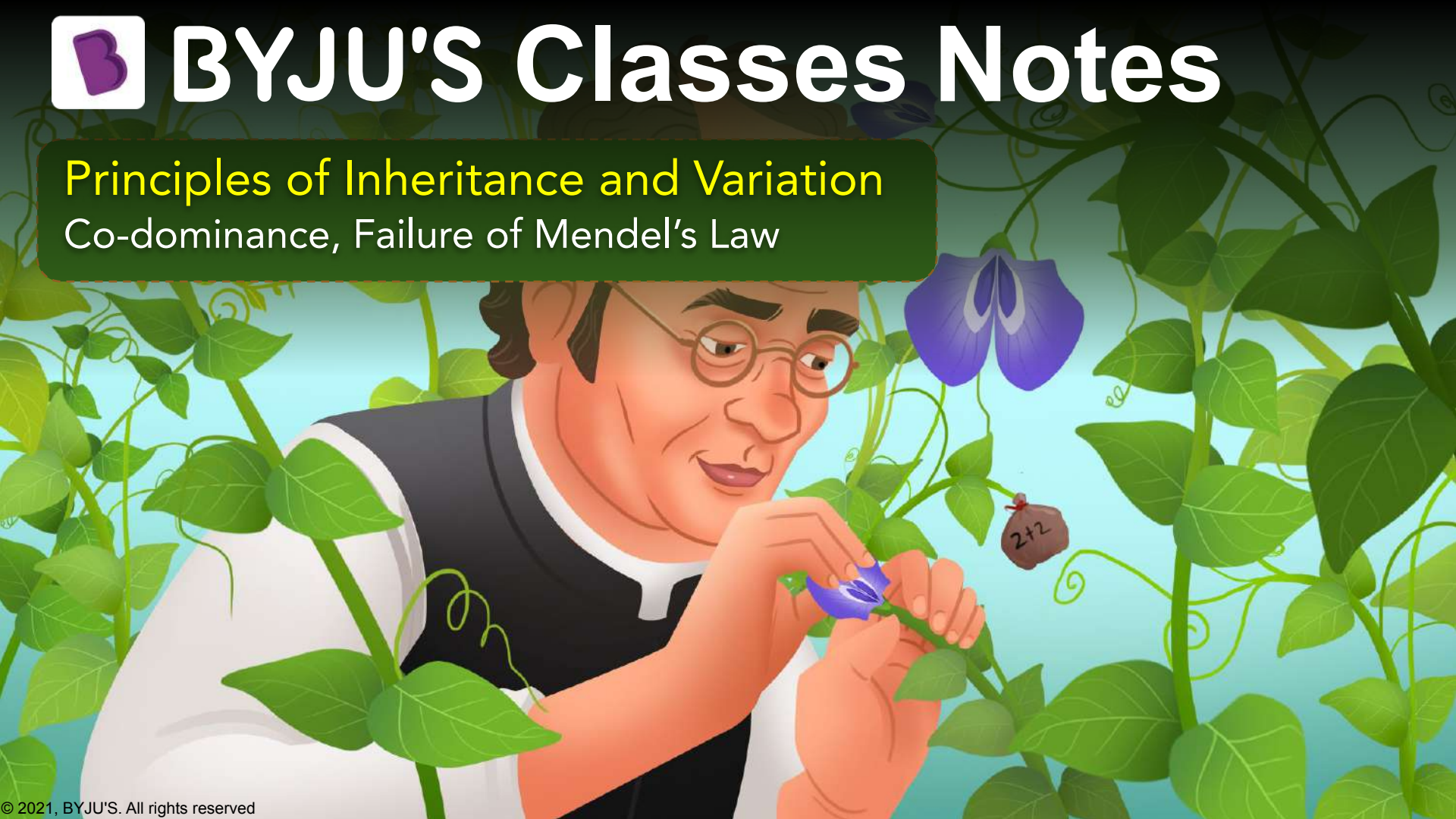
- Important formulae
  - Types of gametes = 2^n
  - Types of phenotypes =2^n
  - Types of genotypes =3^n

where, n= number of heterozygous gene pairs

BYJU'S Classes Notes

Principles of Inheritance and Variation

Co-dominance, Failure of Mendel's Law



Key Takeaways

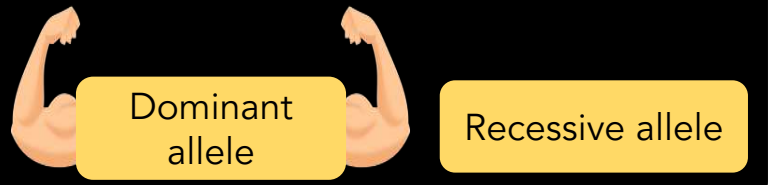
1. Co-dominance
2. The concept of dominance

Summary

Recall: Mendel's Laws of Inheritance

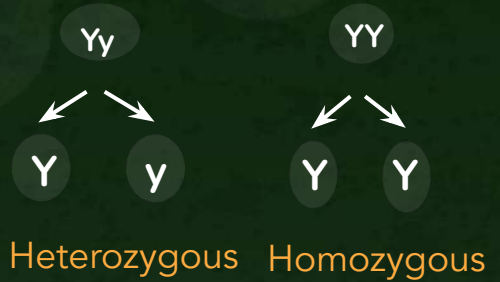
Law of Dominance :

In a dissimilar pair of alleles, one member of the pair dominates (dominant) the other (recessive).



Law of Segregation :

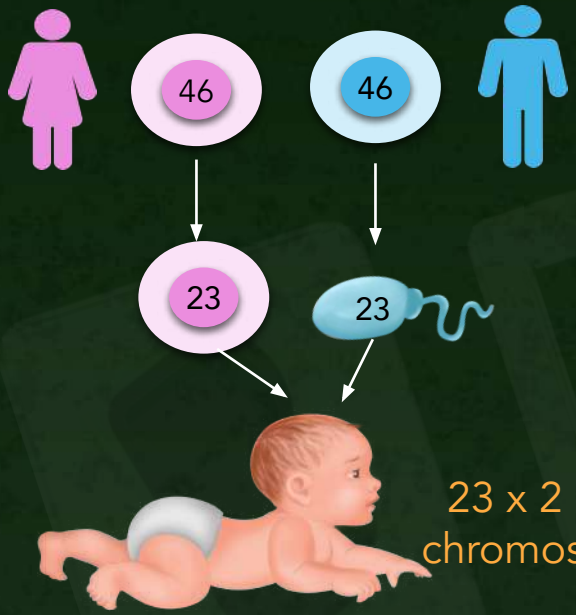
During gamete formation, pair of alleles segregate such that each gamete receives only one allele and alleles don't blend.



Law of independent assortment:

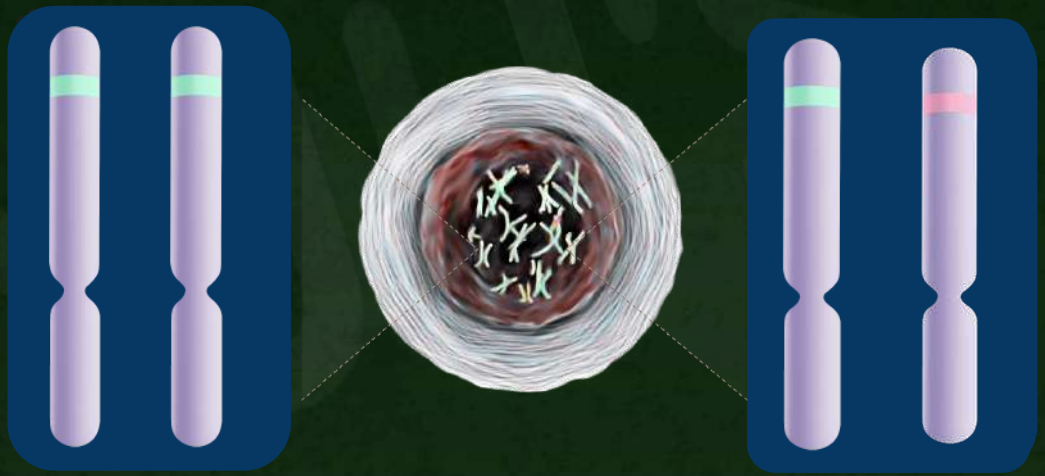
When two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of character.

Recall! Genetic Terms



23 x 2 = 46 chromosomes

Offspring receive 23 chromosomes from each parent. The obtained gene may be dominant or recessive.



Homozygous

Same alleles of the gene on chromosomes

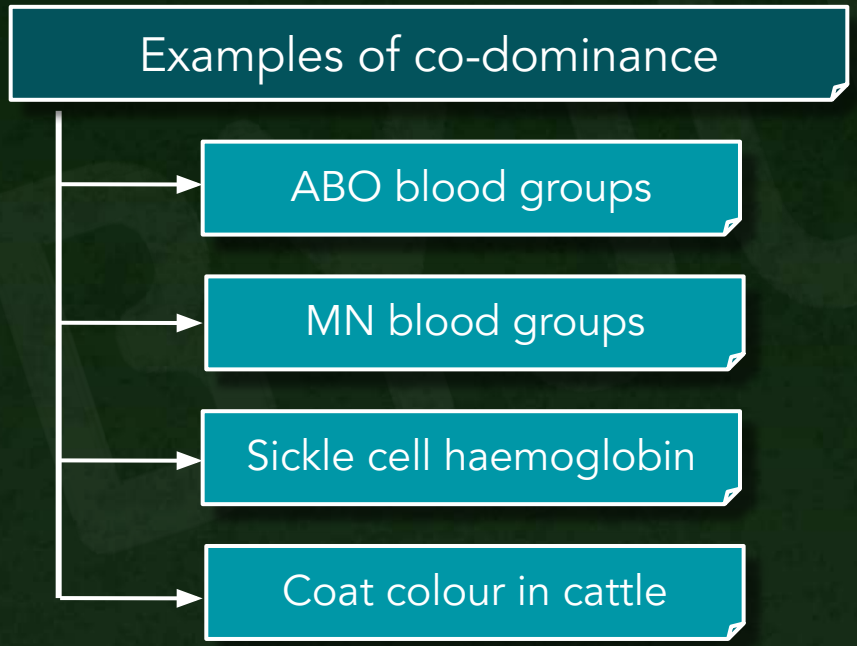
Heterozygous

Different alleles of gene on chromosomes

Co-dominance

- Co-dominance is the type of dominance where the offspring show similarity to both the parents and it is due to independent and equal expression of both alleles.
- Both the alleles which are present together in heterozygous individual but do not show dominant – recessive relationship nor intermediate condition, but express their traits independently are known as co-dominant alleles.
- Co-dominant alleles should not be confused with incomplete dominance.
- Co-dominant inheritance is characterized by distinct expression of the gene products of both alleles.
- For co-dominance to be studied, both products must be phenotypically detectable.
- Co-dominant alleles are shown by the same capital letter with different superscripts. E.g.I^A, I^B, Hb^A, Hb^S.
- Another method is to show them by their own capital alphabets.
- E.g., R (for red hair) and W (for white hair) in cattle.

Co-dominance



Co-dominance

P generation

F_1 generation

- When red and white cattle are crossed, independent characters of both are seen in the offspring (roan).
- The alleles of both parents independently express themselves without blending.
- This is the concept of co-dominance.

F_2 generation

♂ \ ♀	R	W
R	 RR	 RW
W	 RW	 WW

Phenotypic ratio - Red : Roan : White

1 : 2 : 1

Genotypic ratio - RR : RW : WW

1 : 2 : 1

Co-dominance

Examples

Red

White

Roan

Shorthorn cattle

Blood type A

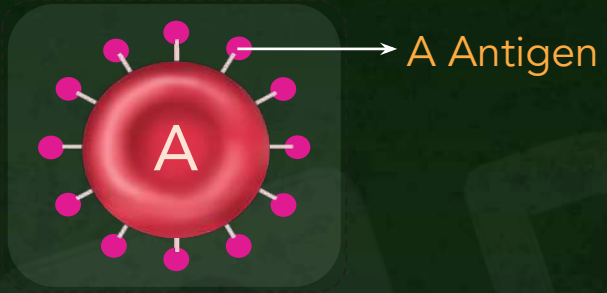
Blood type B

Blood type AB

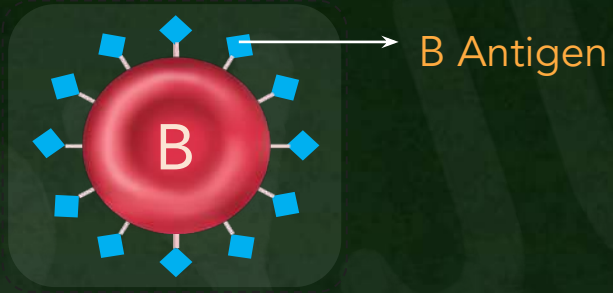
Human ABO blood groups

Co-dominance in Blood Groups

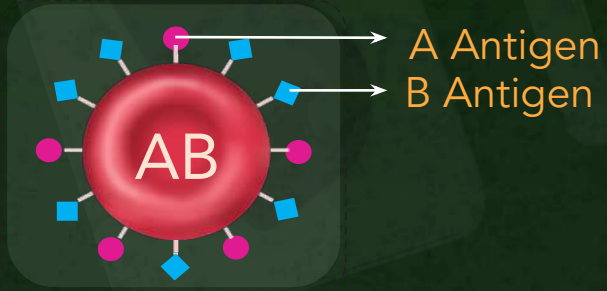
Based on presence or absence of antigens A and B on RBCs, there are four different blood groups.



Blood Group A



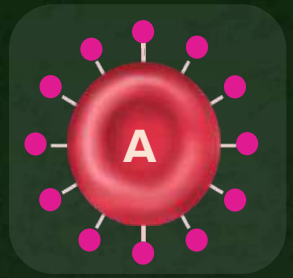
Blood Group B



Blood Group AB

Blood Group O

Allele: I represents the gene responsible for blood groups, whereas the alleles are written in superscript.



Blood group A

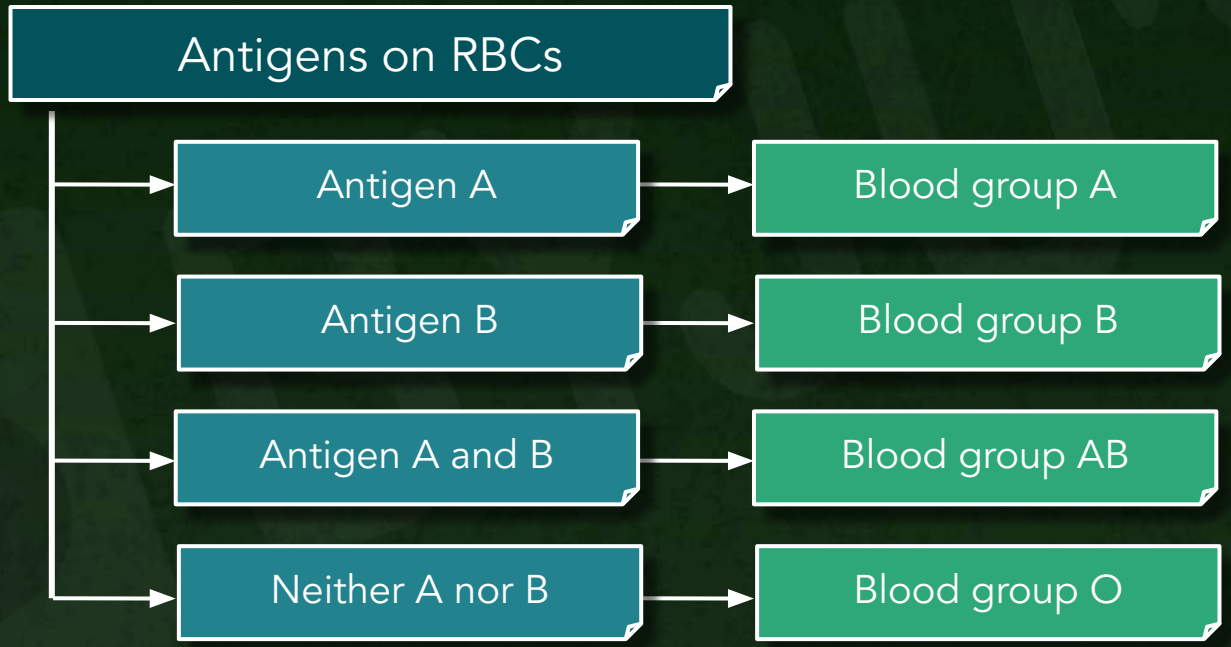
A($I^A I^A$) - Homozygous

A($I^A i$) - Heterozygous

Co-dominance

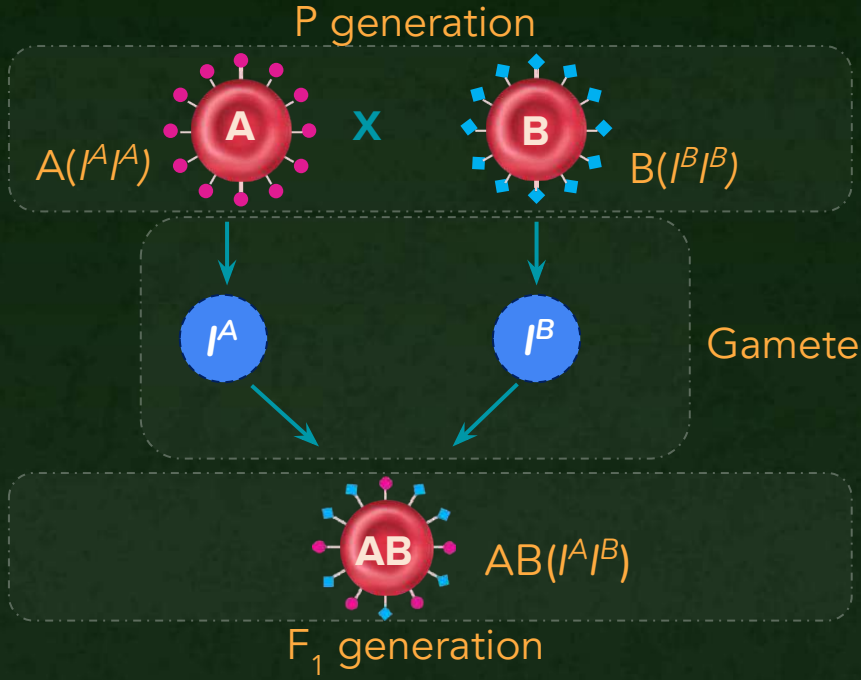
- Three blood groups A, B and O were discovered by Landsteiner while one blood group AB was discovered by de Castello and Sturli.
- ABO grouping is based on the presence or absence of two surface antigens on the RBCs namely A and B.
- Similarly, the plasma of different individuals contain two natural antibodies.

Co-dominance



Co-dominance

I^A and I^B are co-dominant, and hence they create a new blood group AB which has both the antigen A and antigen B.



F_2 generation

♂ \ ♀	I^A	I^B
I^A	A I^AI^A	AB I^AI^B
I^B	AB I^AI^B	B I^BI^B

Phenotypic ratio(F_2) - A : AB : B
1 : 2 : 1

Genotypic ratio(F_2) - I^AI^A : I^AI^B : I^BI^B
1 : 2 : 1

Co-dominance

- Persons with I^AI^B alleles have blood group AB because both I^A and I^B alleles are co-dominant.
- AB blood group is characterised by the presence of both antigen A (from I^A) and antigen B (from I^B) over the surface of RBCs.
- Karl Landsteiner and Philip Levine discovered a glycoprotein molecule found on the surface of RBCs that acts as an antigen.
- In the human population, two forms of this glycoprotein exist, designated M and N; an individual may exhibit either one or both of them.
- The MN system is under the control of a locus found on chromosome 4, with two alleles designated LM and LN.

Co-dominance

Blood group (Phenotype)	Genotype	Antigen present
M	M M	M
N	N N	N
M N	M N	M N

Hypothetical Comparison of Various Inheritance Patterns

Complete dominance

Phenotypic ratio

Red : White

3 : 1

Genotypic ratio

RR : Rr : rr

1 : 2 : 1

Co-dominance

Phenotypic ratio

Red : Roan : White

1 : 2 : 1

Genotypic ratio

RR : RW : WW

1 : 2 : 1

Incomplete dominance

Phenotypic ratio

Red : Pink : White

1 : 2 : 1

Genotypic ratio

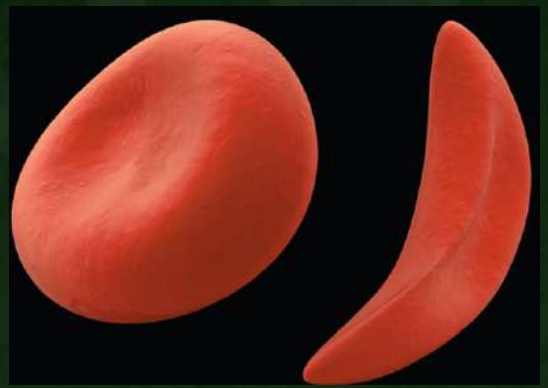
RR : Rr : rr

1 : 2 : 1

Co-dominance: Sickle Cell Haemoglobin

- It is an autosomal recessive disorder in which the affected individual produces an altered form of haemoglobin.
- Most people carry the Hb^A allele and make haemoglobin A.
- Individuals affected with sickle cell anemia are homozygous for the Hb^S allele and produce only haemoglobin S.
- This causes their red blood cells to deform into a sickle shape.

RBCs



Normal Sickle

Co-dominance: Sickle Cell Haemoglobin

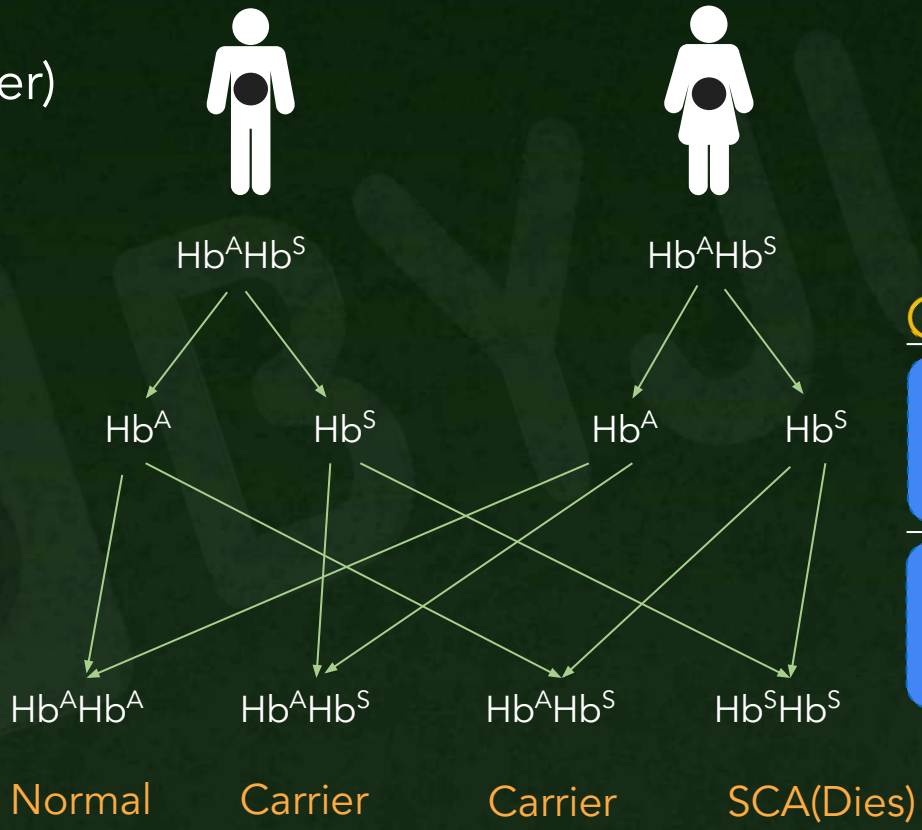
- The sickling phenomenon causes the life span of these cells to be greatly shortened to only a few weeks compared with a normal span of four months, and therefore, anemia occurs.
- Abnormal sickled cells can clog the capillaries at places in the body, leading to localised areas of oxygen depletion.

Co-dominance: Sickle Cell Haemoglobin

- Such an event, called a crisis, causes pain and sometimes tissue and organ damage.
- Kidneys, muscles, joints, brain, gastrointestinal tract, and lungs can be affected.
- For these reasons, the homozygous Hb^SHb^S individual usually has a short life span relative to an individual producing haemoglobin A.

Co-dominance: Sickle Cell Anemia

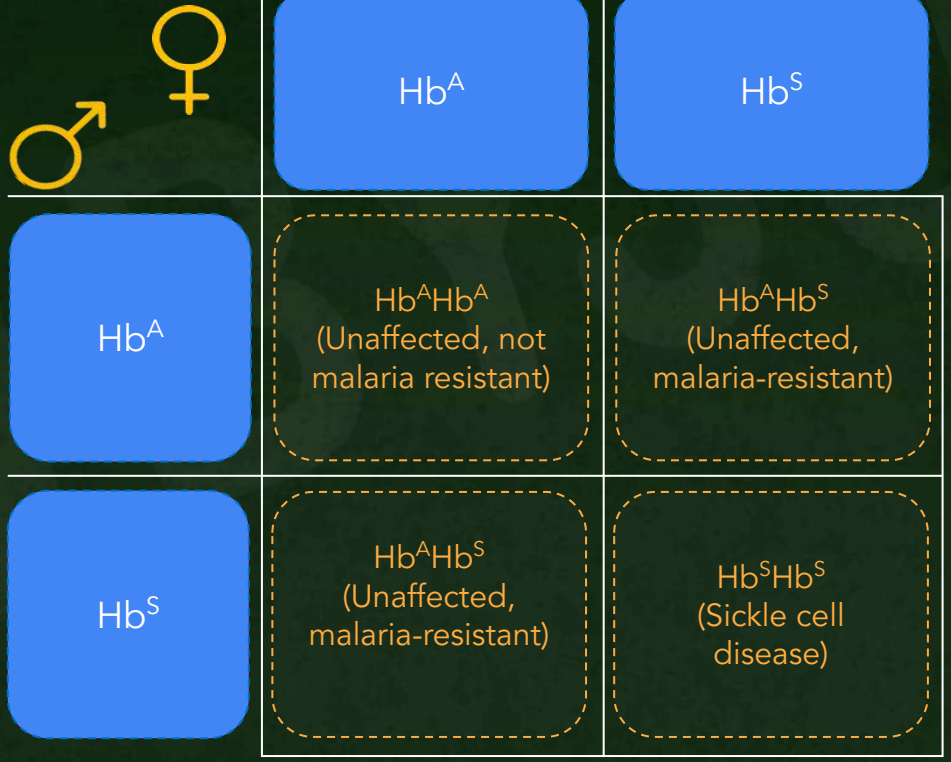
Inheritance
(Carrier x Carrier)



♂ \ ♀	Hb^A	Hb^S
Hb^A	Hb^AHb^A	Hb^AHb^S
Hb^S	Hb^AHb^S	Hb^SHb^S

Co-dominance: Sickle Cell Haemoglobin

Hb^AHb^S X Hb^AHb^S

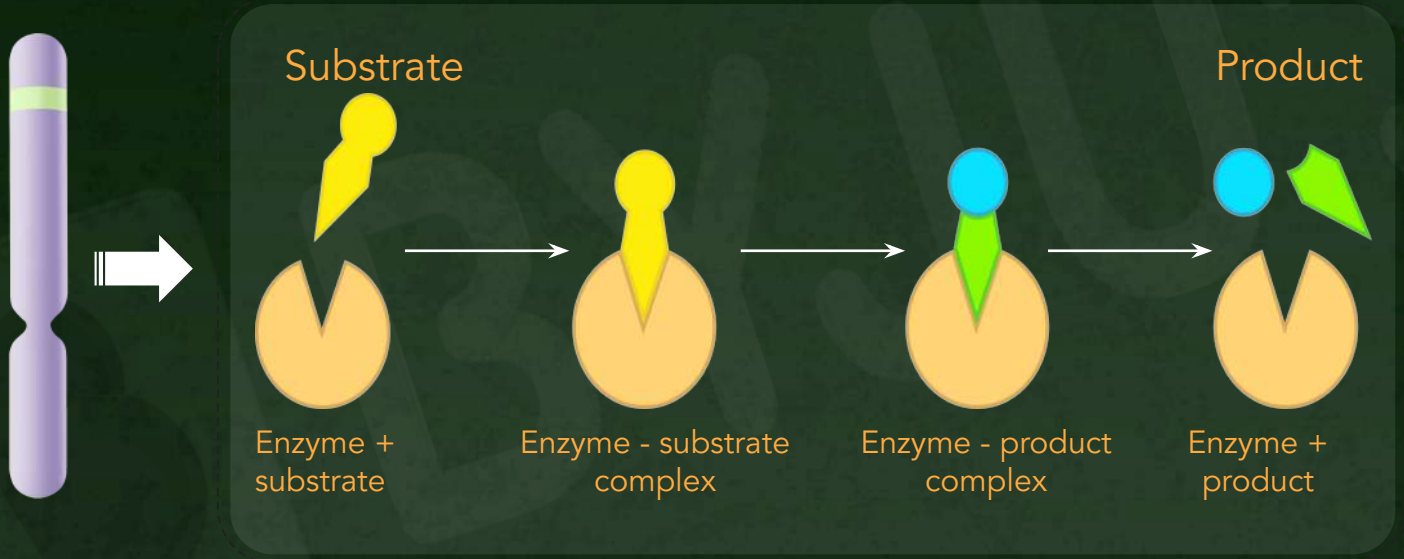


Co-dominance: Sickle Cell Haemoglobin

- Sickle cell allele has been found at a fairly high frequency among human populations that are exposed to malaria.
- The protozoan genus that causes malaria, *Plasmodium*, spends part of its life cycle within the *Anopheles* mosquito and another part within the red blood cells of humans who have been bitten by an infected mosquito.
- Red blood cells of heterozygotes, Hb^AHb^S, are likely to rupture when infected by this parasite, thereby preventing the parasite from propagating.
- People who are heterozygous have better resistance to malaria than do Hb^AHb^A homozygotes.
- In regard to haemoglobin itself, there is co-dominance.
- The alleles Hb^A and Hb^S encode two different forms of haemoglobin that differ by a single amino acid, and both forms are synthesised in the heterozygote.

The Concept of Dominance

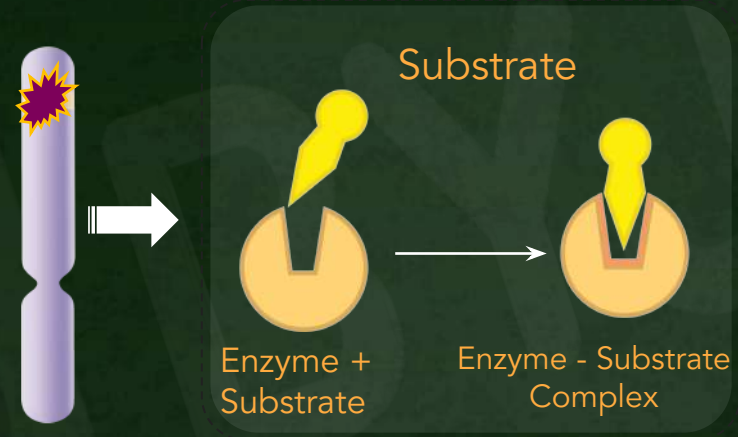
Mendel's laws failed to explain the concepts of incomplete dominance and co-dominance.



For example, a gene contains the information for producing an enzyme. Now there are two copies of a gene i.e., two allelic forms. The normal allele produces the normal enzyme which is needed for the transformation of a substrate S.

The Concept of Dominance

- This allele can change due to some modifications.



Normal/less efficient enzyme

Non-functional enzyme

No enzyme

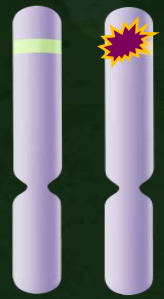
- Can result in mutated allele or modified enzyme or faulty/no product formation.

The Concept of Dominance

- When the modified allele produces a non-functional enzyme or no enzyme, the phenotype may be affected.
- The phenotype/trait will only be dependent on the functioning of the normal allele.
- The unmodified, fully functional allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele.

Dominant allele

- Normal
- Unmodified
- Fully functional
- Original phenotype



Recessive allele

- Modified
- Non-functional

The Concept of Dominance

- Now, in cases of co-dominance or incomplete dominance, the new alleles do not produce useless or non-functional enzyme.
- Instead, they produce an enzyme which actually has the ability to give rise to a new or different trait.
- Hence, when the two different alleles are present together in an individual, one of the two things happen:
  - the physiological process initiated by the different alleles result in a new trait, which can be seen as a blend of the homozygous traits (incomplete dominance).
  - the two alleles are expressed independent of each other and hence both the alleles are expressed together in the phenotype (co-dominance).

Sally bought two potted plants of the same flower but different colour. One was red and another was white. She planted the pots in her garden. After some time, she crossed the red flower plant with the white one. To her surprise, the cross produced flowers of completely different colour; i.e. pink. What do you think is the relation between the red and white colour of the flowers?

A Complete dominance

B Incomplete dominance

C Co-dominance

D Multiple alleles

Sally bought two potted plants of the same flower but different colour. One was red and another was white. She planted the pots in her garden. After some time, she crossed the red flower plant with the white one. To her surprise, the cross produced flowers of completely different colour; i.e. pink. What do you think is the relation between the red and white colour of the flowers?

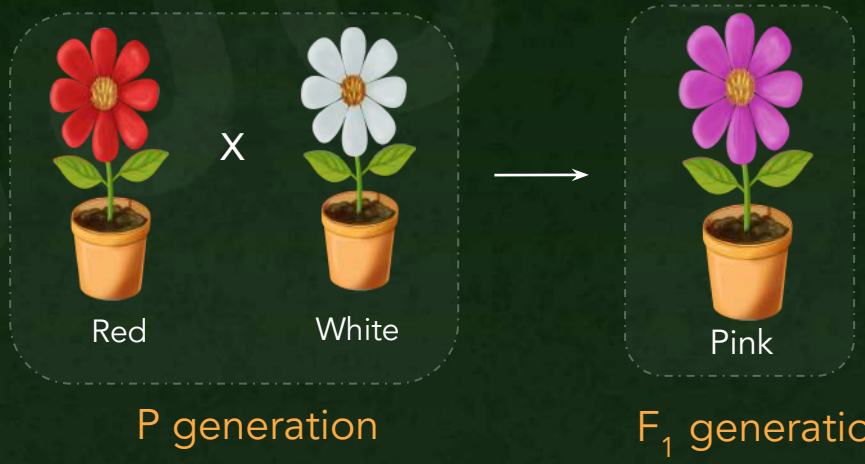
B

A Complete dominance

B Incomplete dominance

C Co-dominance

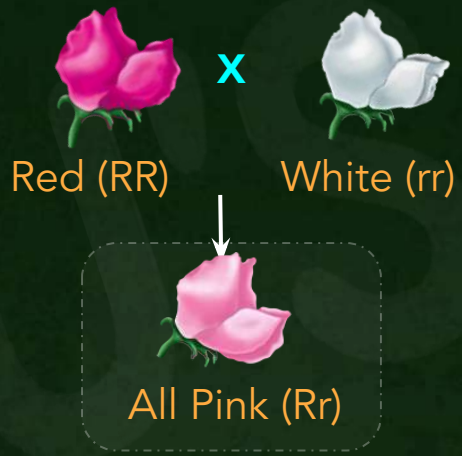
D Multiple alleles



Summary

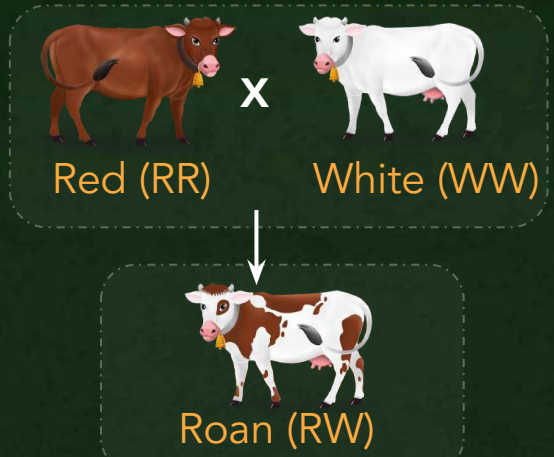
Incomplete dominance

Incomplete dominance is a form of intermediate inheritance in which one allele for a particular trait is not expressed completely over its paired allele.



Co-dominance

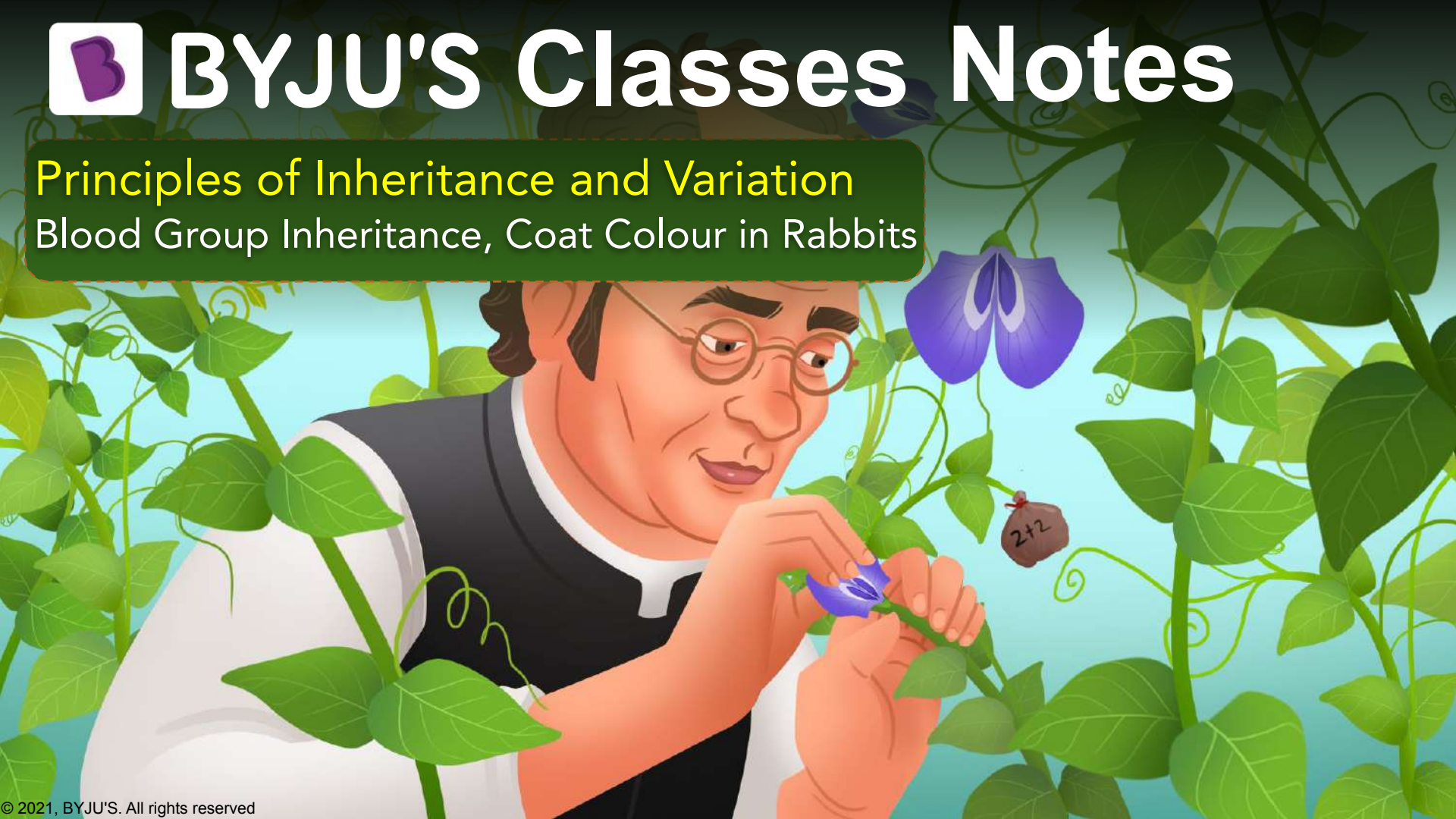
Co-dominance is the type of dominance where the offspring show similarity to both the parents and it is due to independent and equal expression of both the alleles.



BYJU'S Classes Notes

Principles of Inheritance and Variation

Blood Group Inheritance, Coat Colour in Rabbits



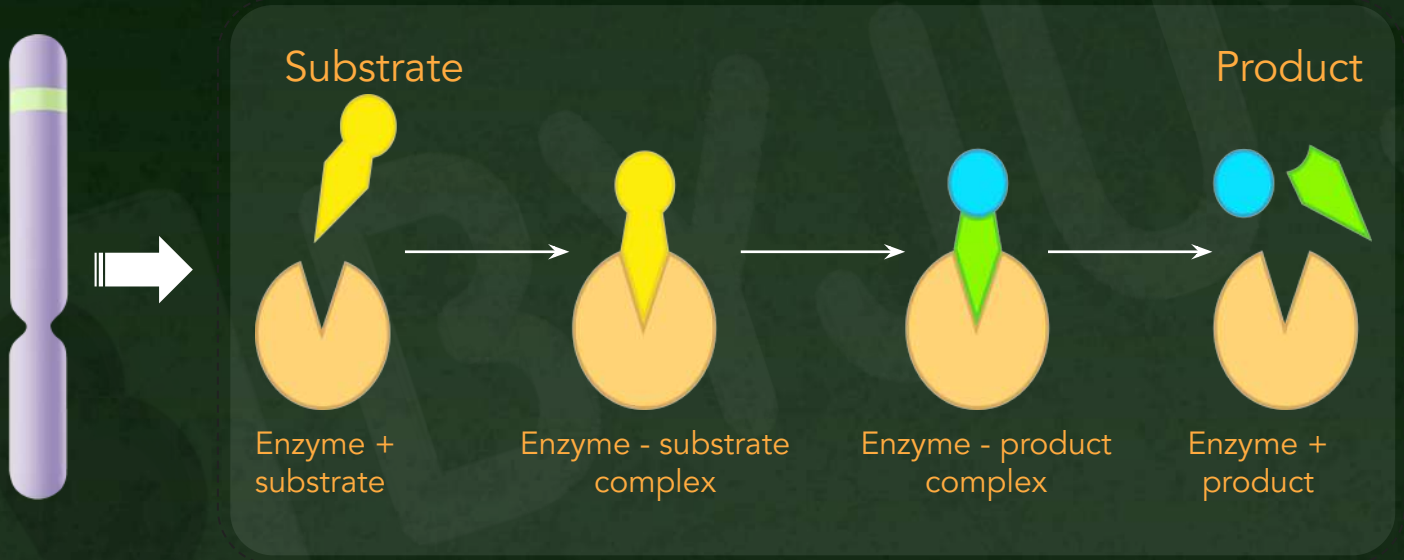
Key Takeaways

1. The concept of dominance
2. Multiple alleles
3. Multiple alleles for blood groups in humans
4. Multiple alleles for coat colour in rabbit

Summary

The Concept of Dominance

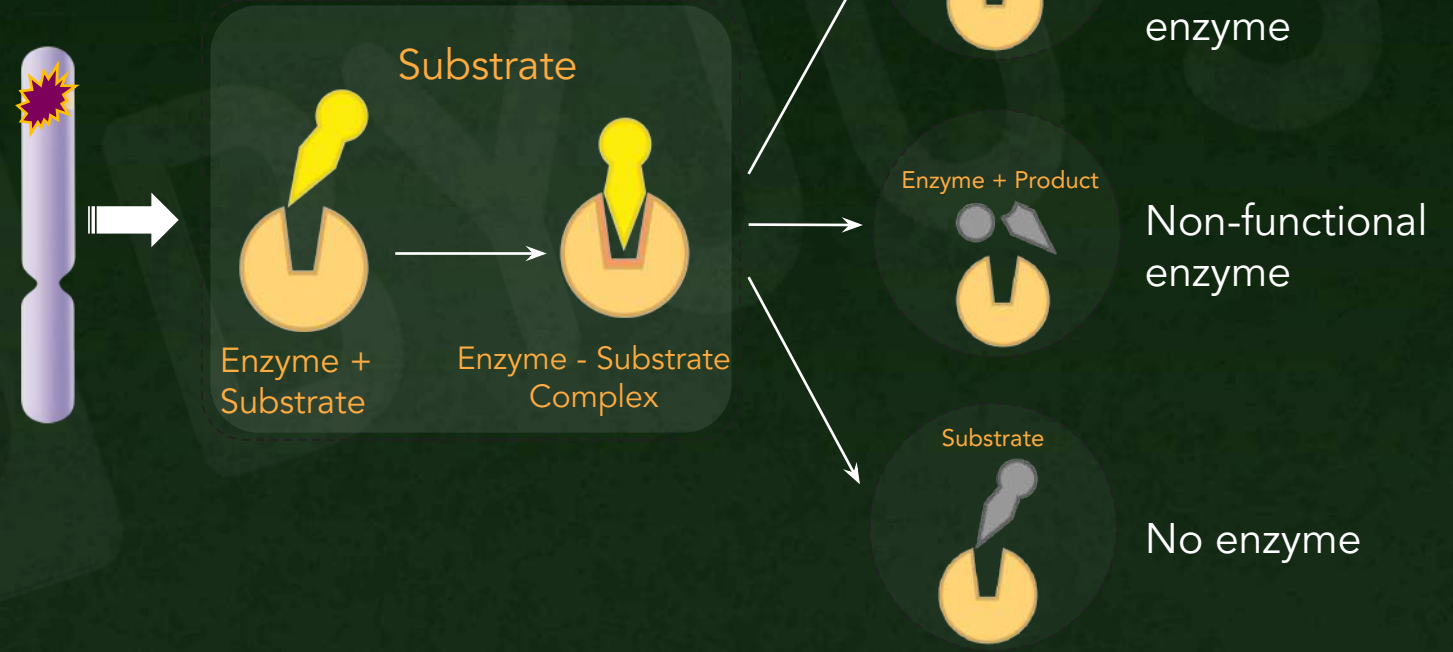
Mendel's law failed to explain the concepts of incomplete dominance and co-dominance.



For example, a gene contains the information for producing an enzyme. Now there are two copies of a gene i.e., two allelic forms. The normal allele produces the normal enzyme which is needed for the transformation of a substrate S.

The Concept of Dominance

This allele can change due to some modifications

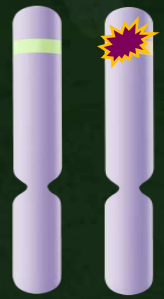


The Concept of Dominance

- When the modified allele produces a non-functional enzyme or no enzyme, the phenotype may be affected.
- The phenotype/trait will only be dependent on the functioning of the normal allele.
- The unmodified, fully functional allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele.
- So, the recessive trait is seen because both the alleles present produce non-functional enzyme or no enzyme at all.

Dominant allele

- Normal
- Unmodified
- Fully functional
- Original phenotype



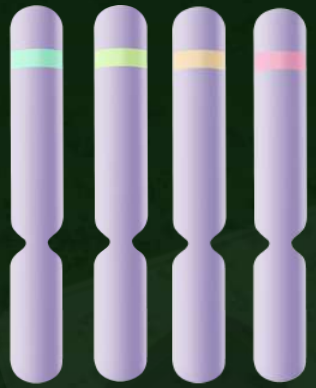
Recessive allele

- Modified
- Non-functional

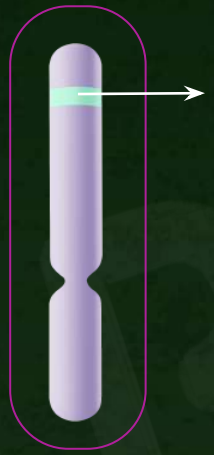
The Concept of Dominance

- Now, in cases of co-dominance or incomplete dominance, the new alleles do not produce useless or non-functional enzyme.
- Instead, they produce an enzyme which actually has the ability to give rise to a new or different trait.
- Hence, when the two different alleles are present together in an individual, one of the two things happen:
  - the physiological process initiated by the different alleles result in a new trait, which can be seen as a blend of the homozygous traits (incomplete dominance).
  - the two alleles are expressed independent of each other and hence both the alleles are expressed together in the phenotype (co-dominance).

Multiple Alleles



More than two alleles of the same genes in a population

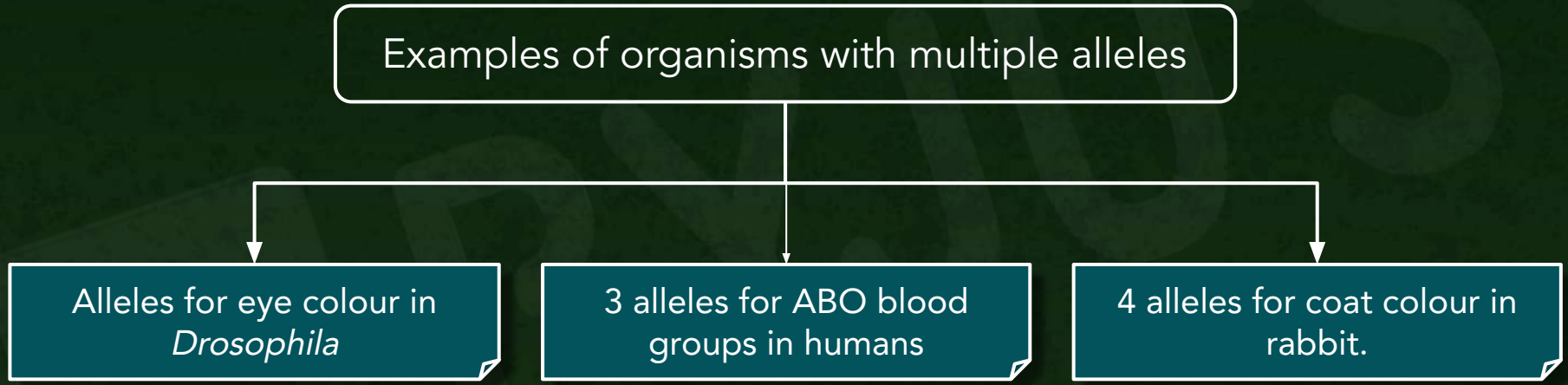


Single chromosome has only one allele

Individual has only two of the alleles

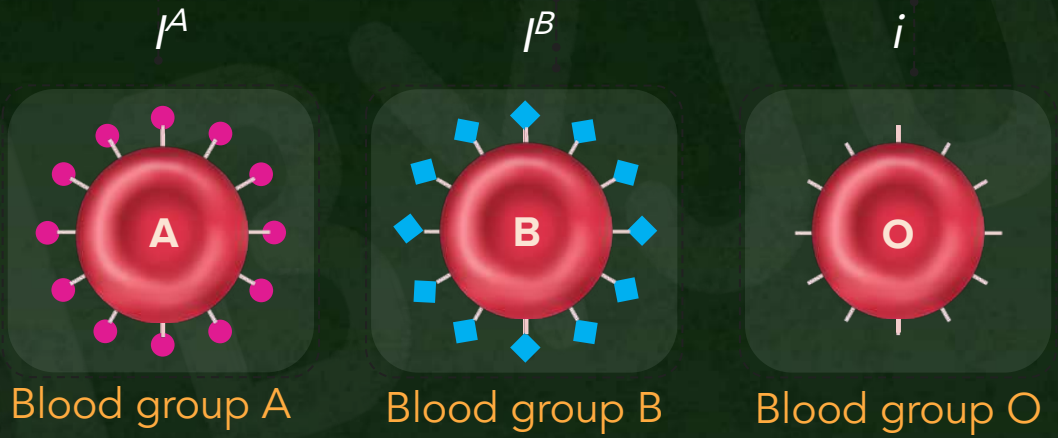
Other alleles are found in other members of the population

Multiple Alleles



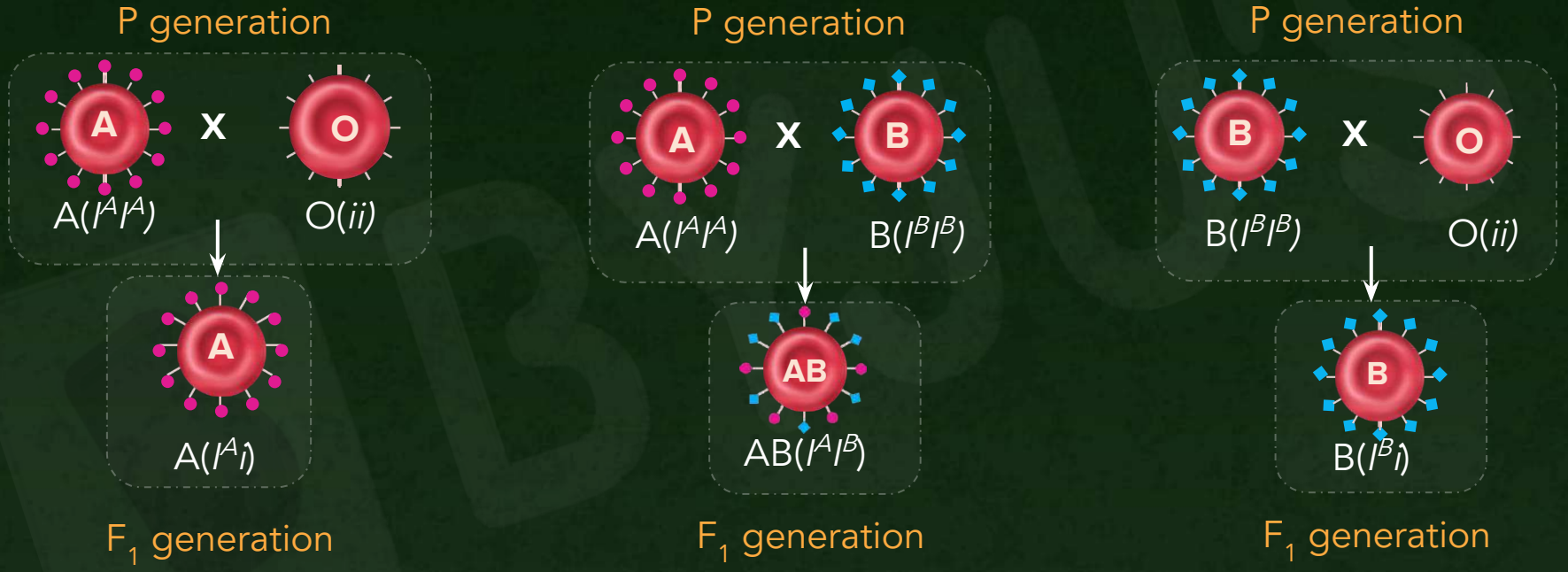
Multiple Alleles for Blood Group in Humans

Gene *I*



Human ABO blood groups

Multiple Alleles for Blood Group in Humans



- When I^A and i are present, only I^A is expressed because *i* does not produce any antigen.
- When I^B and i are present, only I^B is expressed because *i* does not produce any antigen.

Multiple Alleles for Blood Group in Humans

Allele from parent 1	Allele from parent 2	Genotype of offspring	Blood type of offspring	Type of dominance
I^A	I^A	I^AI^A	*A*	-
I^A	I^B	I^AI^B	*AB*	Co-dominance
I^A	*i*	I^Ai	*A*	Dominance
I^B	I^A	I^AI^B	*AB*	Co-dominance
I^B	I^B	I^BI^B	*B*	-
I^B	*i*	I^Bi	*B*	Dominance
i	*i*	*ii*	*O*	-

Human blood groups (ABO)

Phenotype	Genotype
O	*ii*
A	I^AI^A, I^Ai
B	I^BI^B, I^Bi
AB	I^AI^B

Multiple Alleles for Blood Group in Humans

	O	A		B		AB	
	i	i	I^A	i	I^B	I^A	I^B
O	ii **O**	ii O	I^Ai A	ii O	I^Bi B	I^Ai A	I^Bi B
A	ii O	ii O	I^AI^A **A**	ii O	I^Bi B	I^Ai A	I^Bi B
B	ii O	ii O	I^Ai A	ii O	I^BI^B **B**	I^Ai A	I^Bi B
AB	I^Ai A	I^Ai A	I^AI^A A	I^Ai A	I^AI^B AB	I^AI^A A	I^AI^B **AB**

This box marks the unique genotypes found in human blood groups.

Multiple Alleles for Coat Colour in Rabbit

- Rabbits have four different alleles that determine their coat colours.

Gene C

Agouti

Chinchilla

Himalayan

Albino

Coat colour in rabbits

Multiple Alleles for Coat Colour in Rabbit

- Differences in the various alleles are related to the function of tyrosinase.
- The C allele encodes a fully functional tyrosinase that eventually results in a full brown coat color.
- The chinchilla allele (C^{ch}) is a partial defect in tyrosinase that leads to a slight reduction in black pigment and a greatly diminished amount of orange/yellow pigment, which makes the animal look gray.
- The albino allele, designated c, is a complete loss of tyrosinase, resulting in white color.

Multiple Alleles for Coat Colour in Rabbit

- The himalayan pattern of coat color, determined by the C^h allele, is an example of a temperature-sensitive allele.
- The mutation in this gene has caused a change in the structure of tyrosinase, so it works enzymatically only at low temperature.
- Because of this property, the enzyme functions only in cooler regions of the body, primarily the tail, the paws, and the tips of the nose and ears.

Did You Know?

Multiple alleles

- 15 alleles code for the eye colour in *Drosophila*.
- They show various types of co-dominance and incomplete dominance.

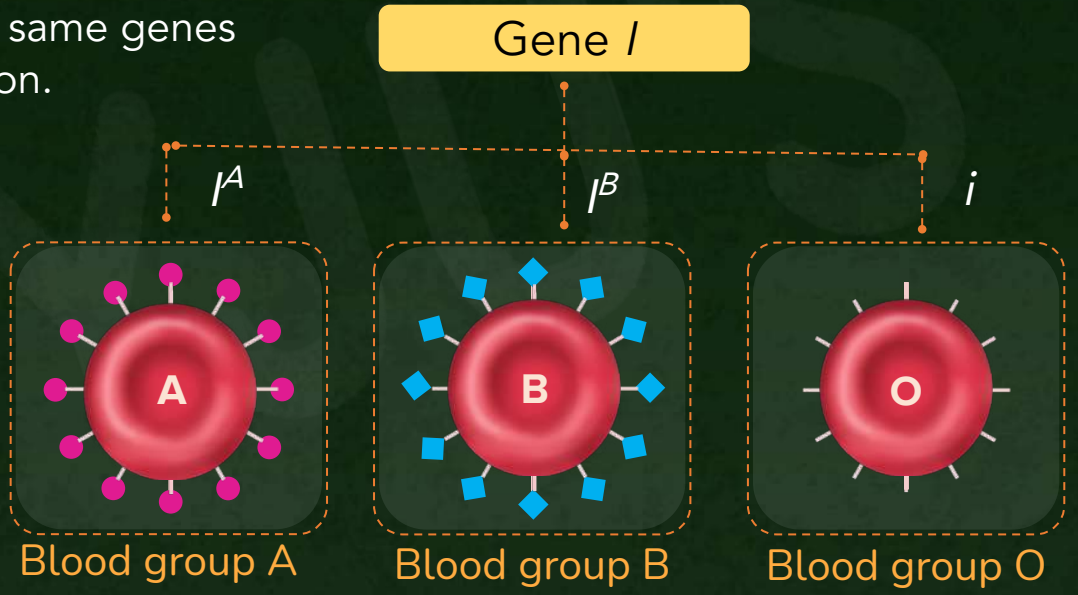
Summary

Multiple alleles

Presence of more than two alleles of the same genes in a population.

Human Blood Groups (ABO)

Phenotype	Genotype
O	ii
A	I^AI^A, I^Ai
B	I^BI^B, I^Bi
AB	I^AI^B



Human ABO blood groups

Summary

- Rabbits have four different alleles that determine their coat colours.

Gene C

Agouti

Chinchilla

Himalayan

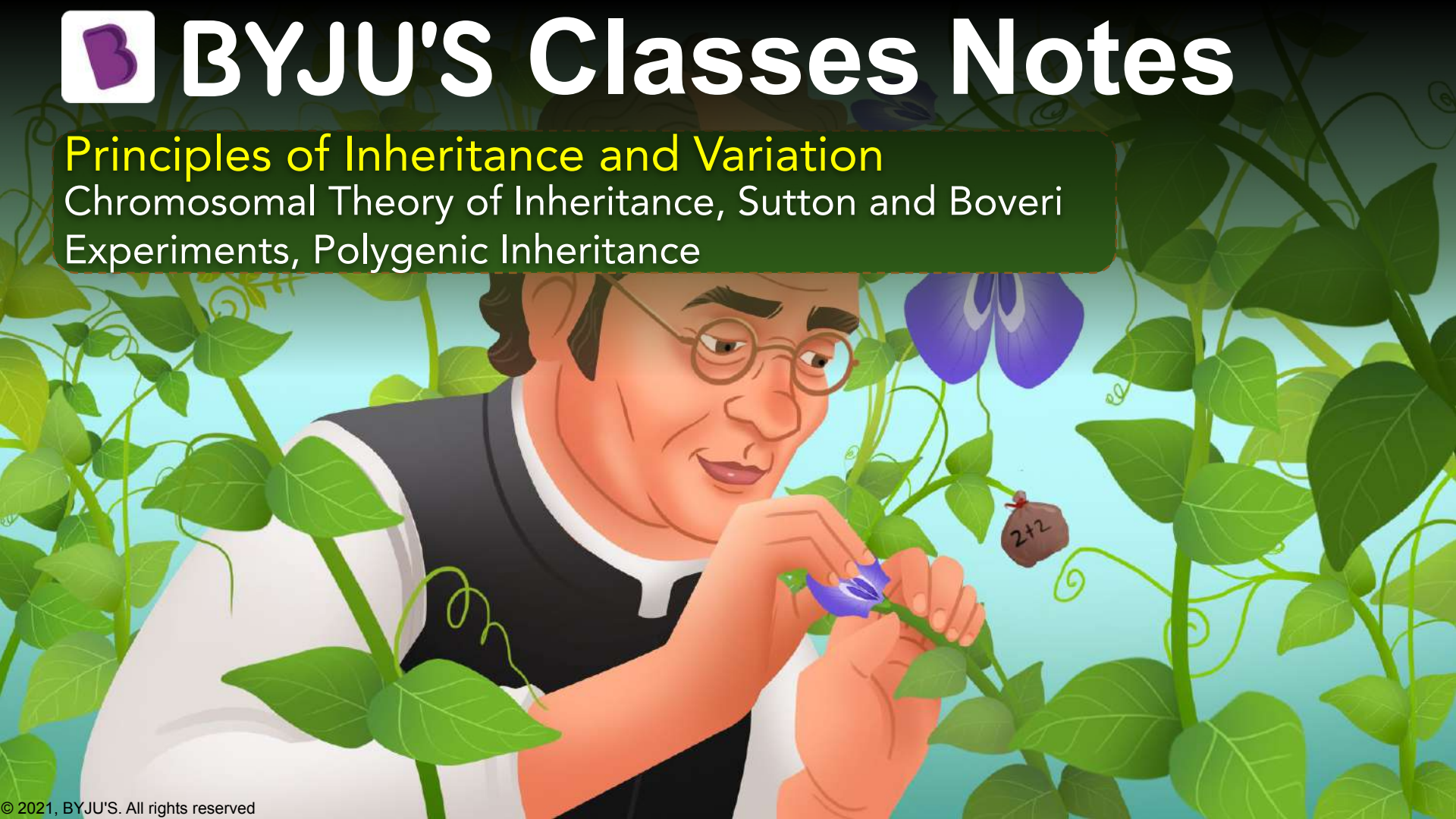
Albino

Coat colour in rabbits

BYJU'S Classes Notes

Principles of Inheritance and Variation

Chromosomal Theory of Inheritance, Sutton and Boveri Experiments, Polygenic Inheritance



Key Takeaways

1. Chromosomal theory of Inheritance
2. Sutton and Boveri Experiments
3. Similarities between chromosomes and factors
4. Polygenic Inheritance

Summary

Recall! Exceptions to Mendel's Laws of Inheritance

01 Incomplete dominance

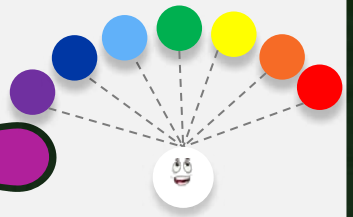
None of the factors/alleles are dominant

Both of the alleles in a heterozygote are expressed independently

02 Co-dominance

03 Multiple alleles

More than two alleles of same gene



A brown female (BB) dog gave birth to a litter of 8 puppies. Half of the puppies are completely brown and half of them have white and brown spots (Bb) on them. If the colour coat of dog shows codominance, what is the genotype of the father dog? Completely brown- BB; spotted- Bb; white-bb.

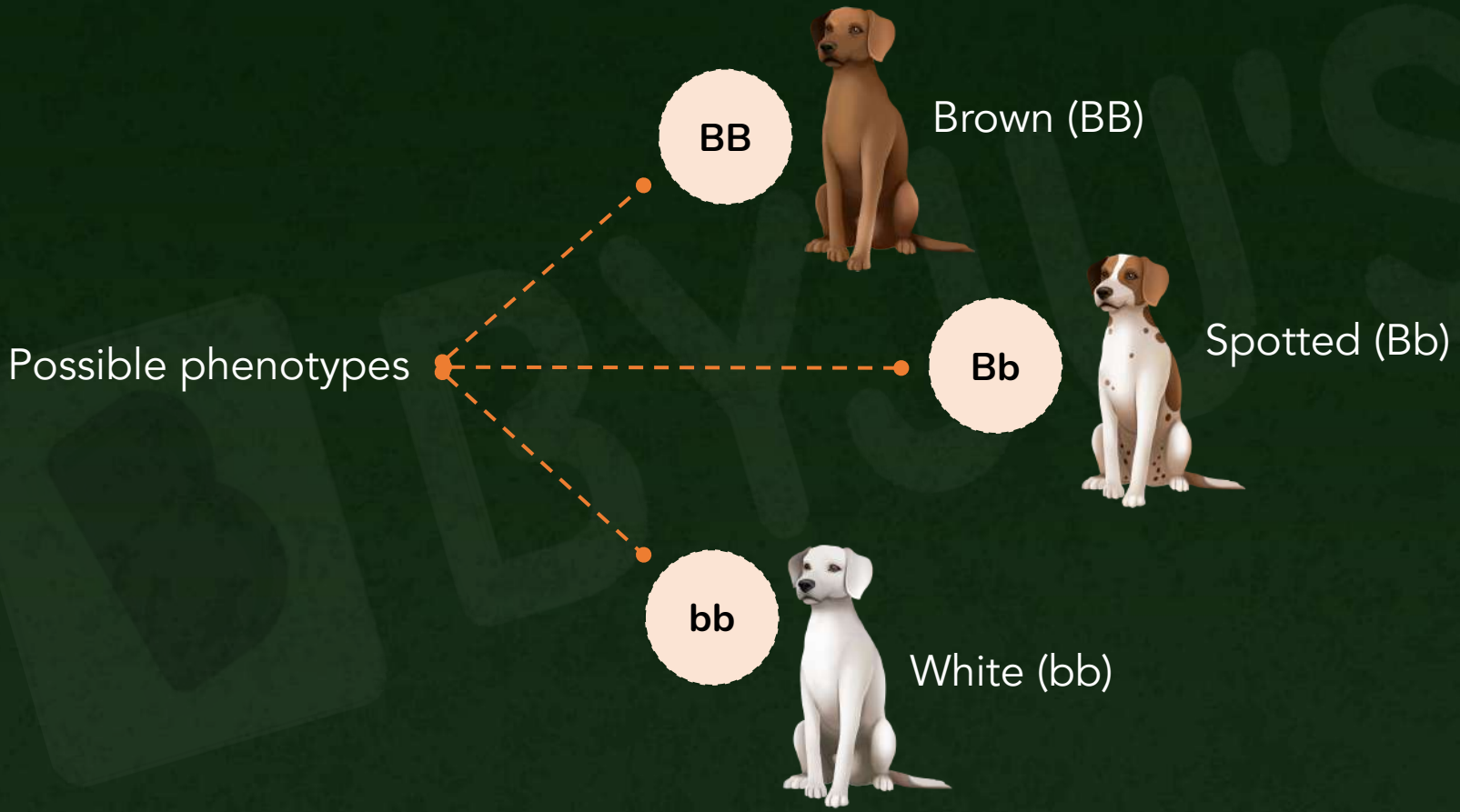
A Bb

B bb

C BB

D Can’t be decided

Discussion



Discussion

P generation:

Brown (BB)

X

Brown (BB)

Gametes:

B

B

F_1 generation:

Brown (BB)

Possibility 1: Brown female and brown male

Discussion

P generation:

Brown (BB)

x

Spotted (Bb)

Possibility 1: Brown female and spotted male

Gametes:

B B b

F_1 generation:

Brown (BB)

Spotted (Bb)

50% brown offspring and 50% spotted offspring

Discussion

P generation:

Brown (BB)

X

White (bb)

Gametes:

B

b

F_1 generation:

Spotted (Bb)

Possibility 3: Brown female and white male

A brown female (BB) dog gave birth to a litter of 8 puppies. Half of the puppies are completely brown and half of them have white and brown spots (Bb) on them. If the colour coat of dog shows codominance, what is the genotype of the father dog? Completely brown- BB; spotted- Bb; white-bb.

A Bb

B bb

C BB

D Can’t be decided

What is the chance that a type O and Type AB couple could produce offspring with type A?

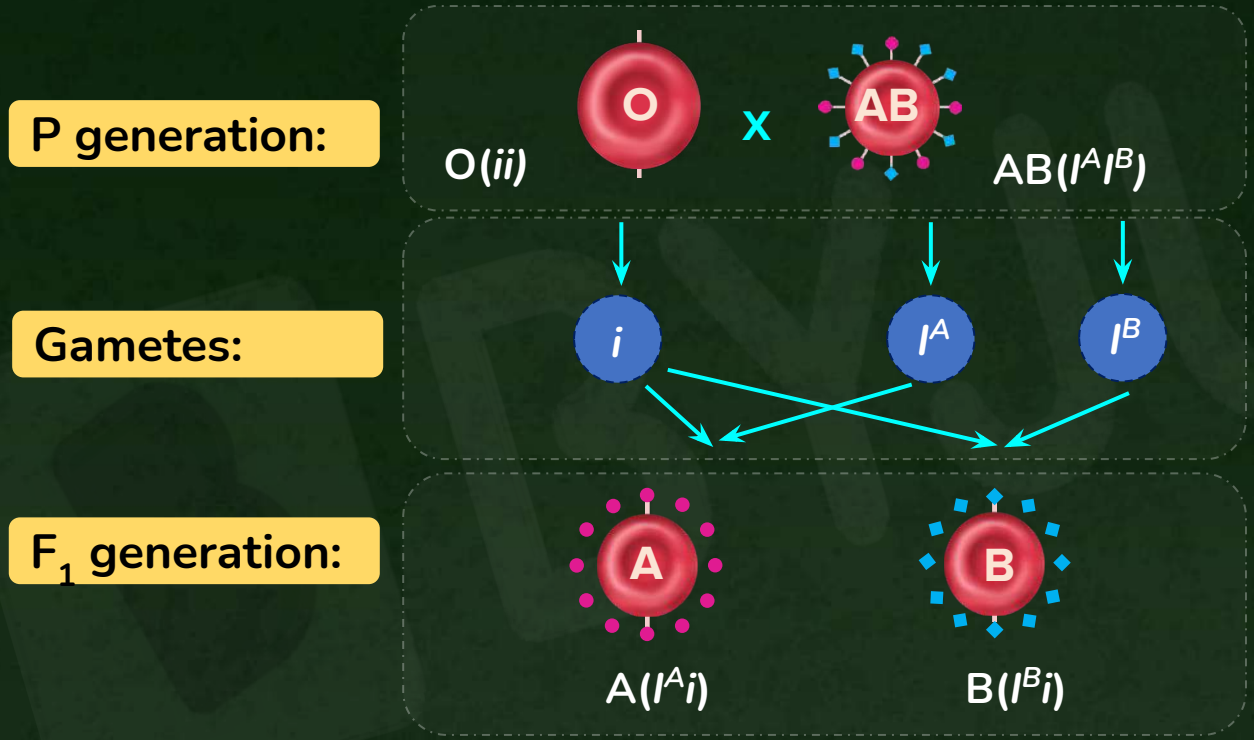
A. 50 %

B. 75 %

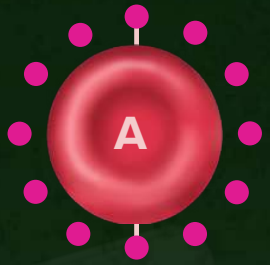
C. 25 %

D. 100 %

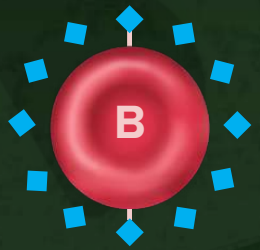
Discussion



Discussion



$A(I^Ai)$



$B(I^Bi)$

$$\text{Percentage/ Chances} = \frac{\text{Desired blood group}}{\text{Total number of possible blood groups}} \times 100$$

$$\text{Percentage/ Chances} = \frac{1}{2} \times 100 = 50\%$$

What is the chance that a type O and Type AB couple could produce offspring with type A?

A. 50 %

B. 75 %

C. 25 %

D. 100 %

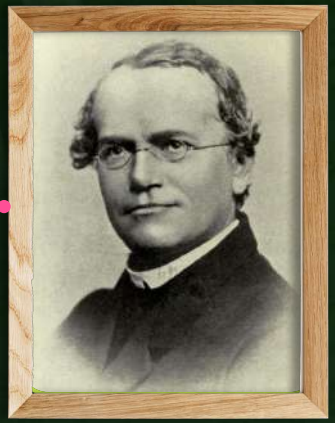
Recall! Mendel's Conclusions

Factors come in pairs

Factors segregate during gamete formation

Mendel's conclusions

Factors segregate independently of each other



Recall! Reasons of Negation of Mendel's Work

No communication

Not widely publicised

Concept of factors

Concept of "factors" not accepted by contemporaries

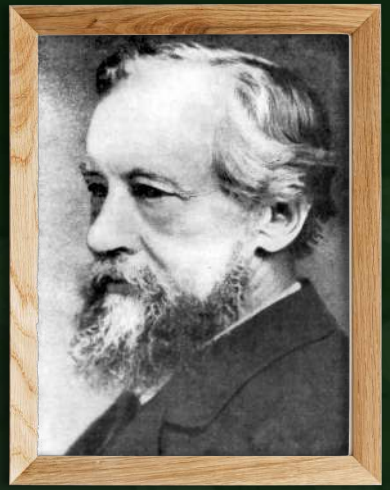
Maths to explain biology

Usage of mathematics unacceptable to many biologists

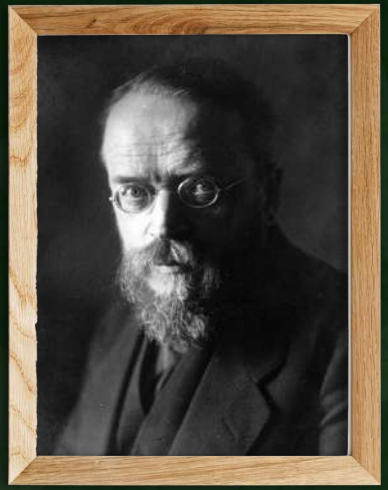
Physical proof

No physical proof for existence of factors

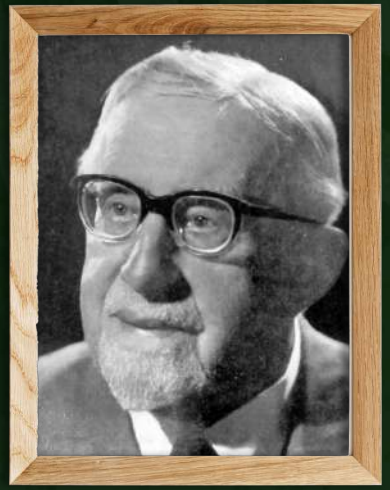
Recall! Resurgence of Genetics



Hugo de Vries



Carl Correns

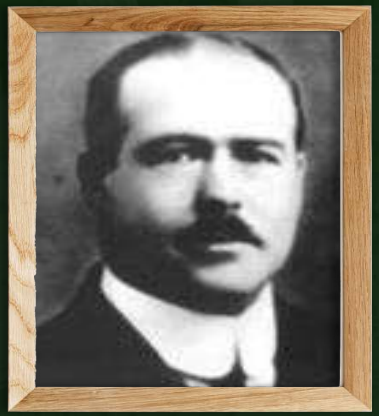


Erich Von Tschermak

These three scientists independently rediscovered the same concepts as that of Mendel.

Chromosomal Theory of Inheritance

- Scientists Walter Sutton and Theodore Boveri independently performed some experiments.
- They were able to carefully observe cell division and discovered that chromosomes separate during cell division and are present in pairs, specifically in meiosis.
- Their conclusions reinforce Mendel's work which are now known as chromosomal theory of inheritance.



Walter Sutton

Theodore Boveri

Sutton and Boveri Experiment – 1

Lubber grasshopper

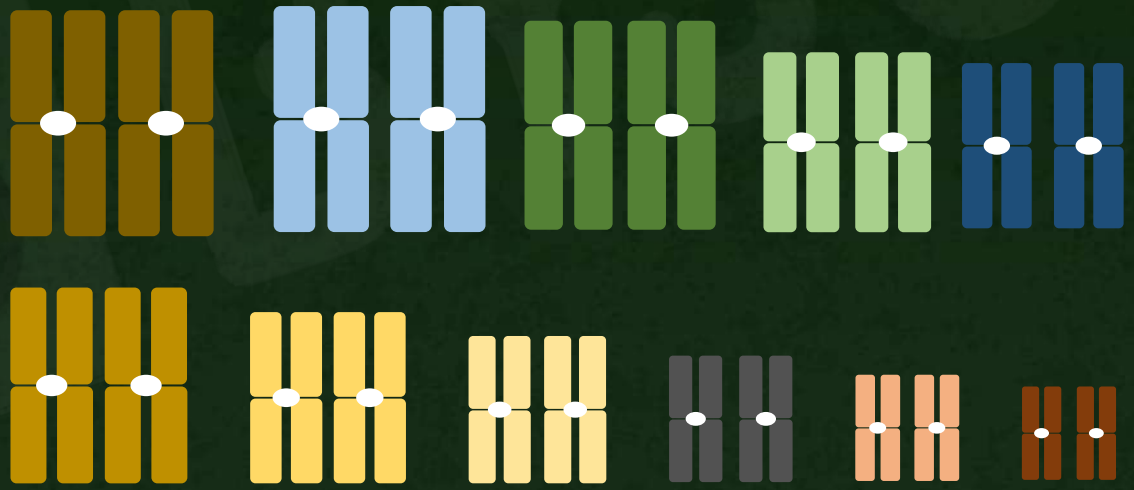
Walter Sutton conducted the experiment on lubber grasshopper.

- He observed the cells of the grasshopper under microscope and observed that it had 22 chromosomes in each cell.
- Initially, all 22 chromosomes appeared quite different from each other.
- But when he started arranging them based on their structure and size, he found a pattern.
- He saw that every chromosome had an identical pair which are now known as homologous pairs.

Sutton and Boveri Experiments - 1

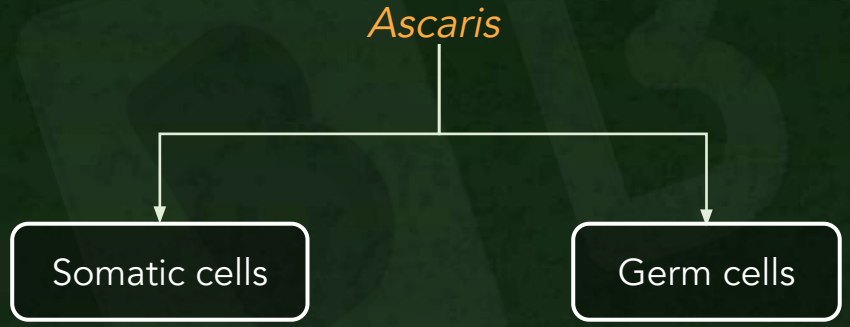
Conclusion:

Every chromosome had an almost identical partner.
So, chromosomes occur in pairs.



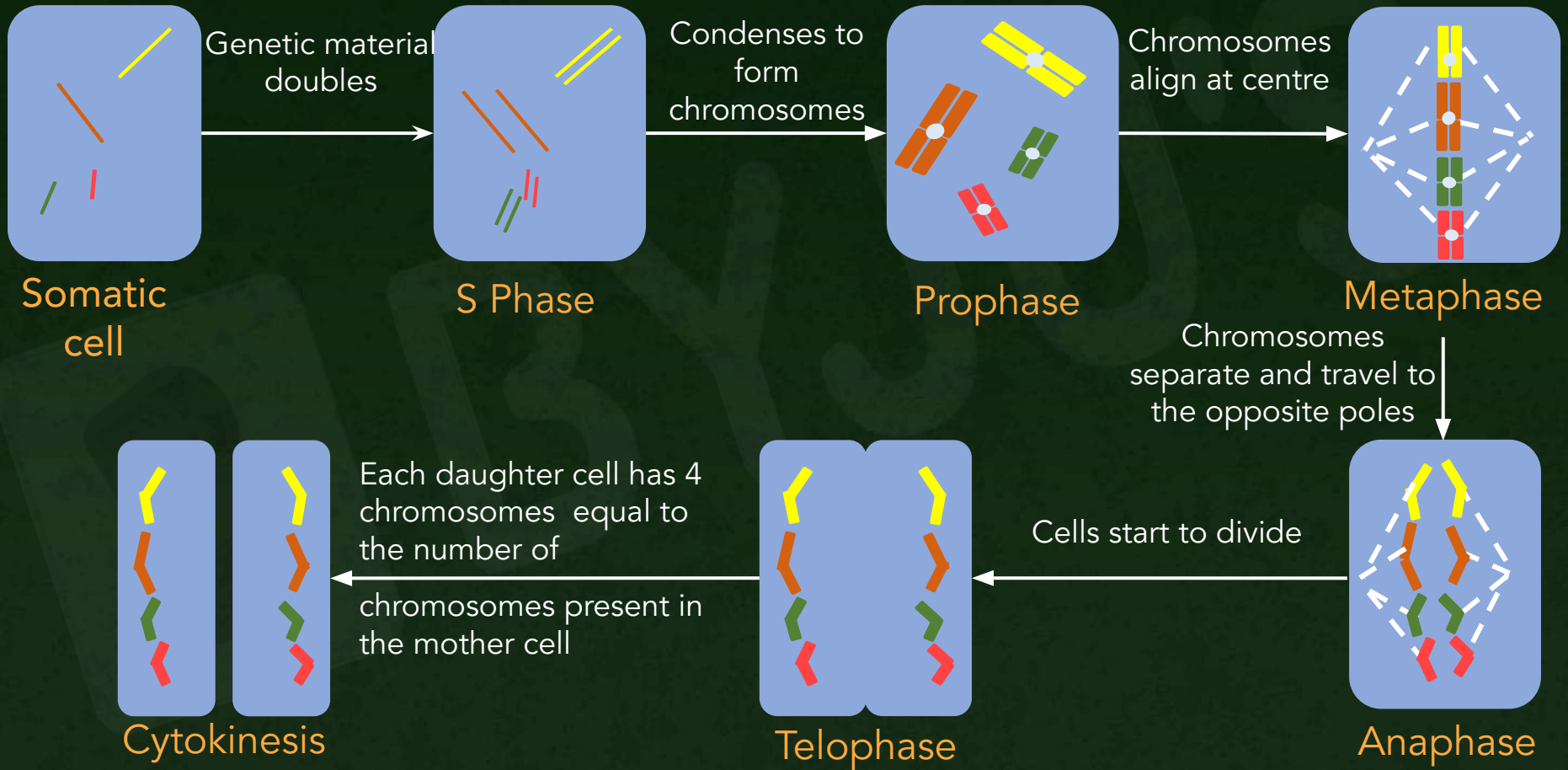
Sutton and Boveri Experiment - 2

Ascaris

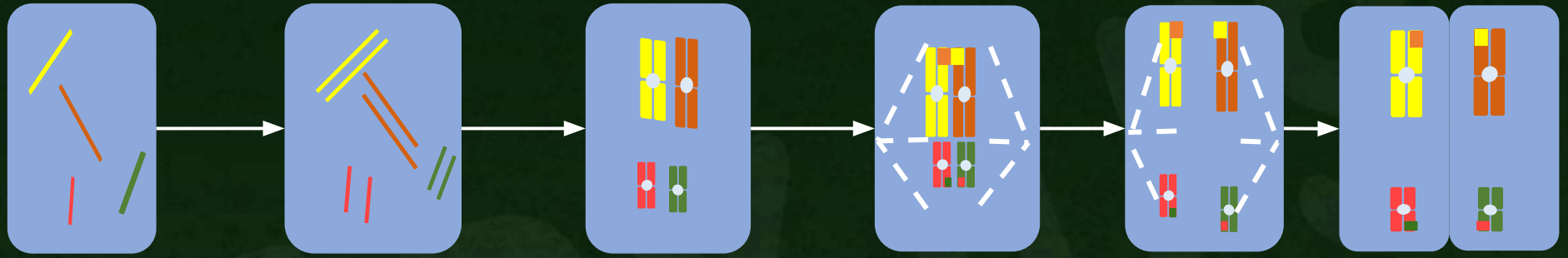


- Boveri worked on *Ascaris* and found that the organism has 4 chromosomes.
- He observed that, there are 2 types of cells present in *Ascaris*.
  - In the present era, they are called somatic cells and the germ cells.
- The way each of these cells divide is different.

Sutton and Boveri Experiments - 2



Sutton and Boveri Experiment - 2



Germ cell

S phase

Genetic material doubles

Prophase I

4 chromosomes are formed and crossing-over occurs between homologous chromosomes

Metaphase I

Homologous pairs of chromosomes align at the equatorial plate

Anaphase I

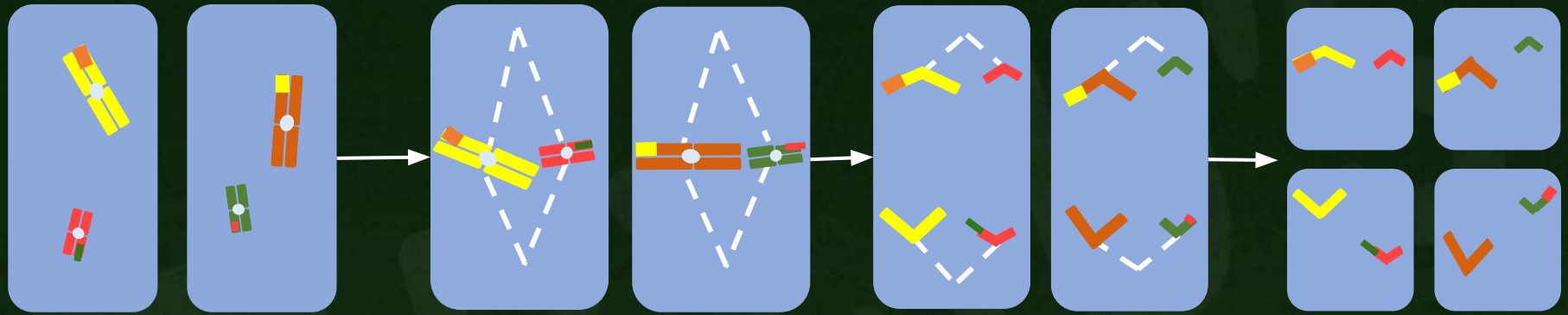
The identical pairs separate and move to opposite poles

Telophase I

Karyokinesis is completed which will be followed by cytokinesis

Sutton and Boveri Experiment - 2

Germ cells



Prophase II

Metaphase II

The chromosomes get arranged at the centre

Anaphase II

Sister chromatids separate and travel to opposite poles

Telophase II and cytokinesis

By the end of the division 4 cells were formed and 2 chromosomes were present in each cell

Sutton and Boveri Experiment - 2

Conclusion:

- Meiosis reduces number of chromosomes in germ cells by half.
- Chromosomes segregate during gamete formation.

Sutton and Boveri Experiment – 3

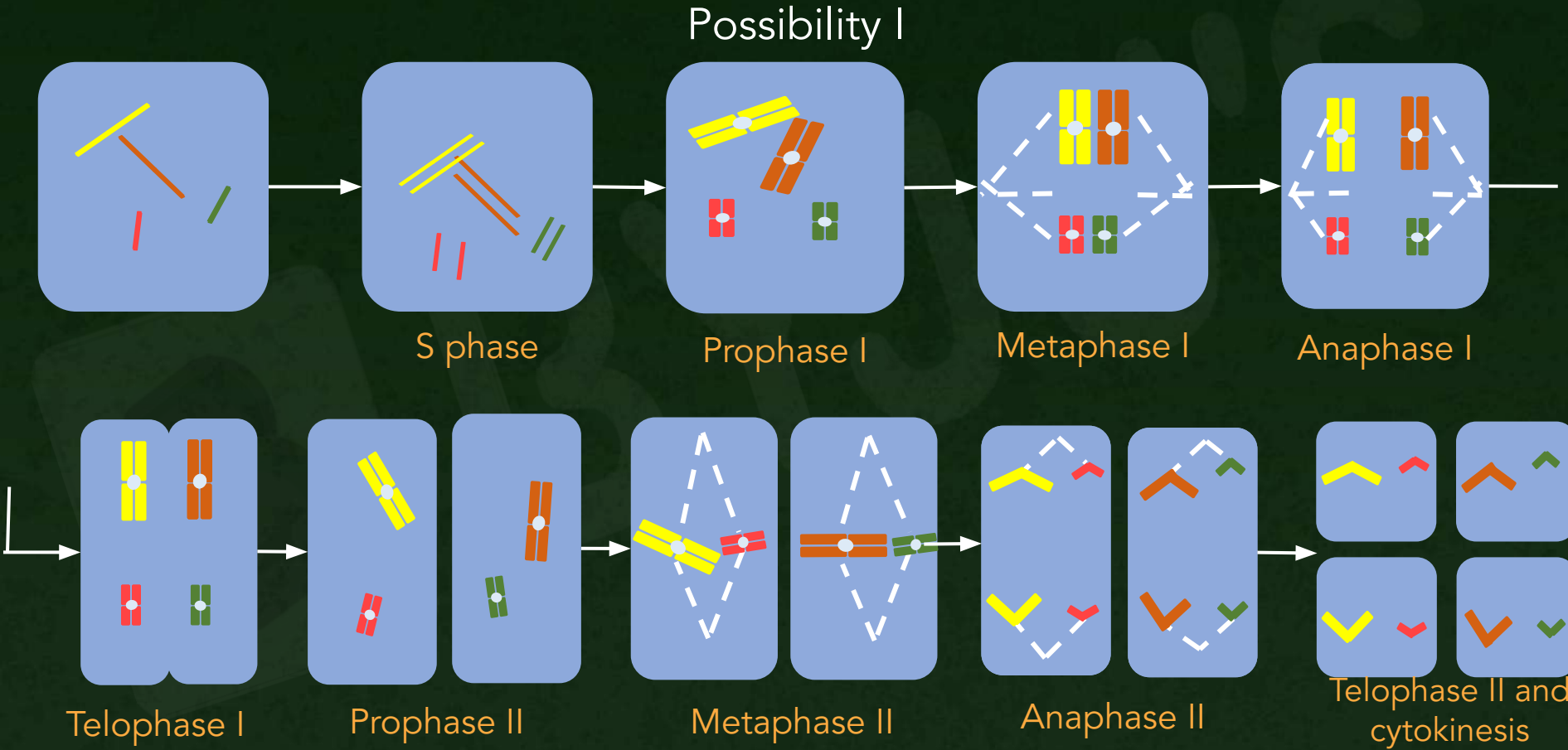
Walter Sutton

Lubber grasshopper

Sutton conducted another experiment on the same lubber grasshopper.

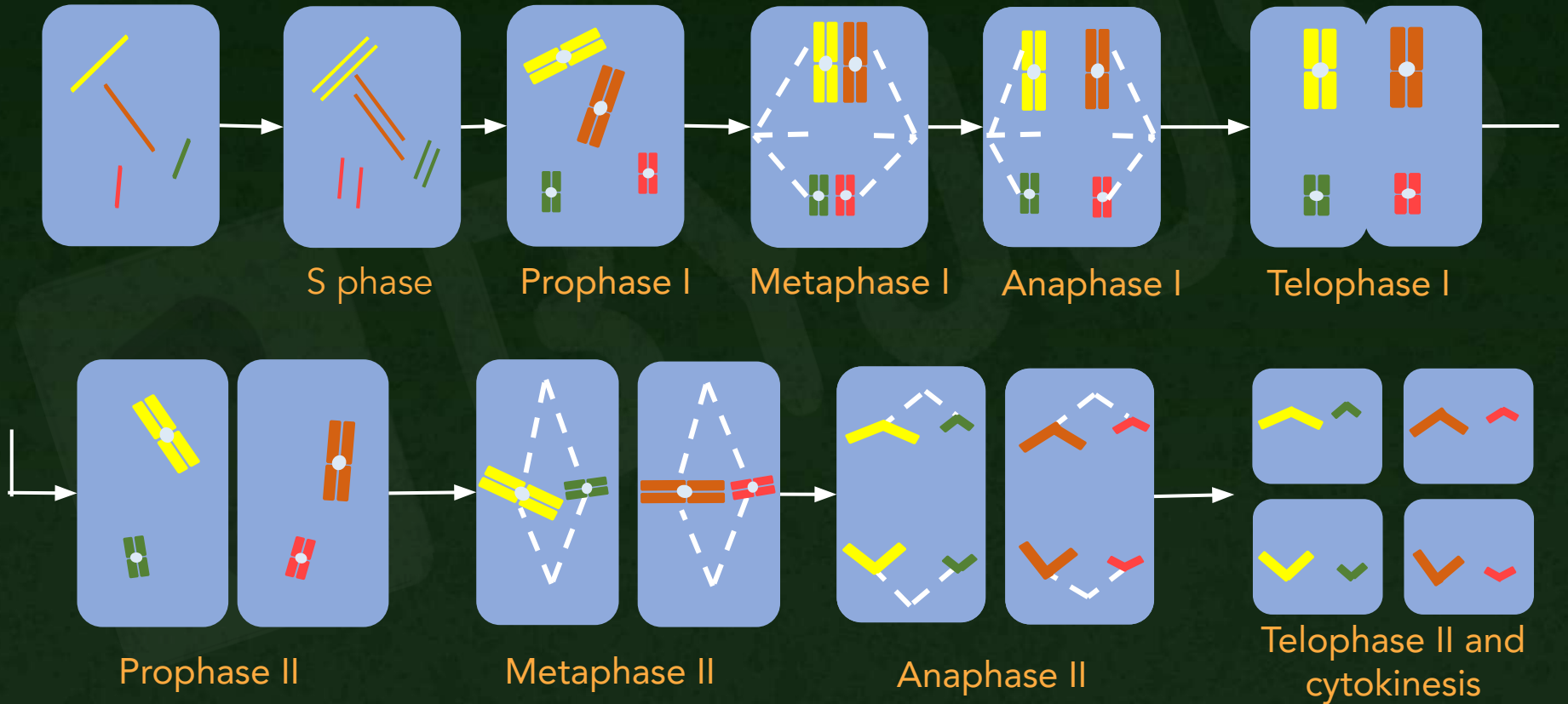
- He had observed that grasshopper had a total of 22 chromosomes in a cell.
- Let us consider only 4 chromosomes.
- With these 2 sets of homologous chromosomes, there are two possibilities for meiosis.

Sutton and Boveri Experiment - 3



Sutton and Boveri Experiment - 3

Possibility II



Sutton and Boveri Experiment - 3

- On comparing both possibilities, we find:
  In case I, yellow and red are together.
  In case II, yellow and green are together.
- Each chromosome was, therefore, independent of others.
- When they were separated into gametes, the set of chromosomes in each daughter cell ended up having a mixture of the parental traits, but not necessarily the same mixture as that of other daughter cells.
- Hence, it can be concluded that during gamete formation, chromosomes in a homologous pair separate independently of other homologous pairs of chromosomes.

Sutton and Boveri Experiment - 3

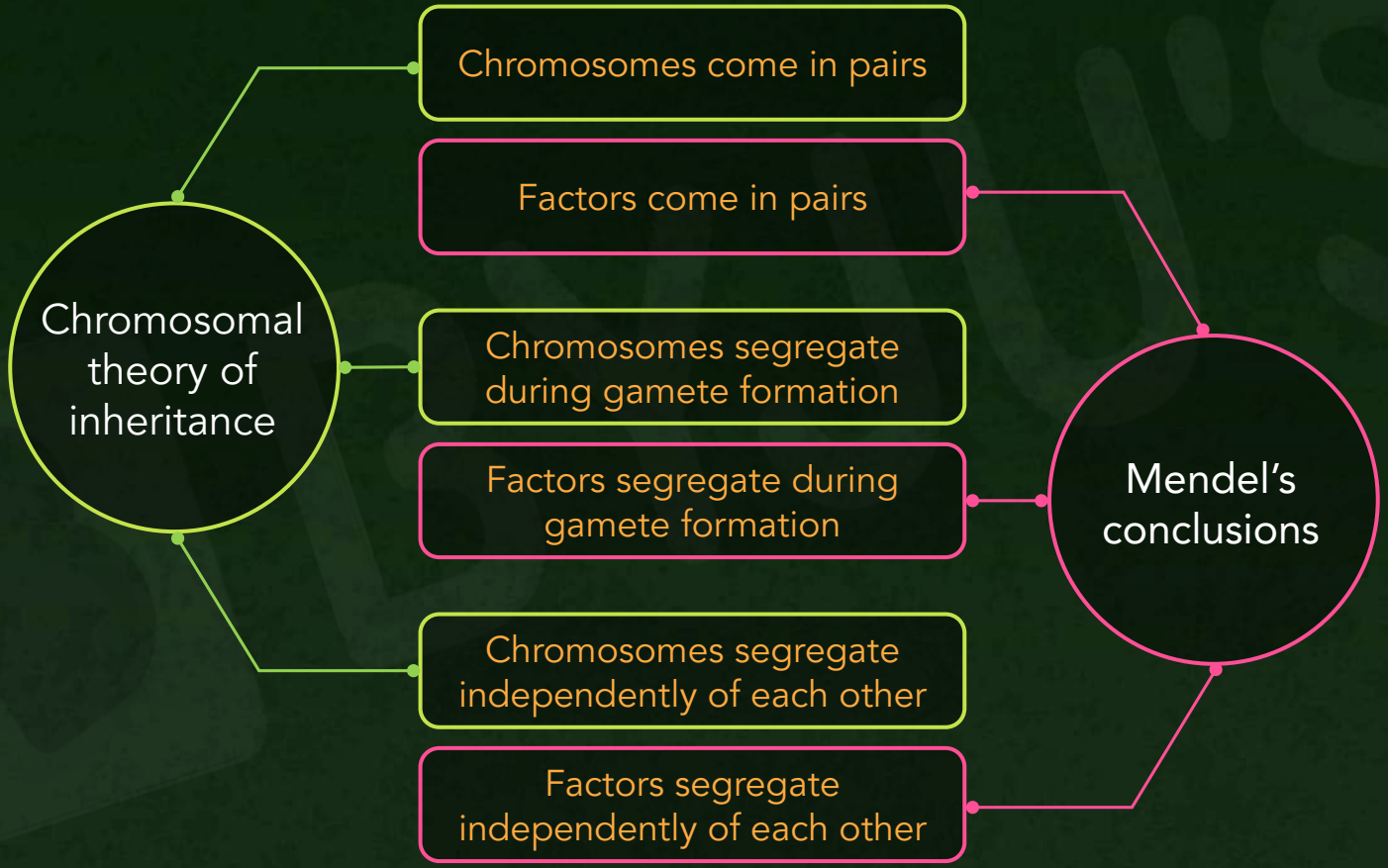
Conclusion:

- During meiosis for gamete formation,
  - The homologous pairs of chromosomes align themselves on metaphase plate randomly and separation occurs in anaphase I.
- Chromosomes in a homologous pair separate independently of other homologous pairs of chromosomes.

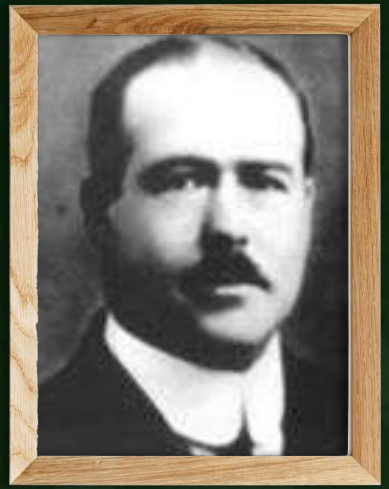
Sutton and Boveri Experiment – 3

Chromosomes somatic cell (2n)	Chromosomes germ cells (n)	Number of possible combinations in germ cell (2^n)
4	2	$2^2 = 4$
6	3	8
10	5	32

Similarities b/w Factors and Chromosomes



Sutton and Boveri Theory of Inheritance

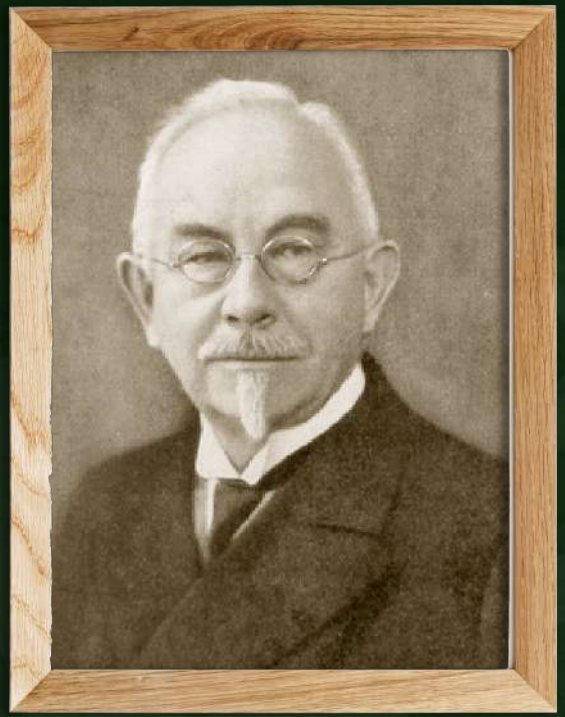


Walter Sutton

Theodore Boveri

- Sutton and Boveri also came up with a hypothesis, based on their experiments:
    - Mendel's factors are present on specific locations of the chromosomes
    - Inheritance of chromosomes = Inheritance of factors

Discovery of Gene



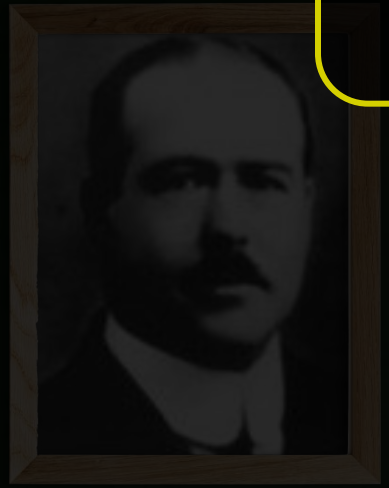
Wilhelm Johannsen

- Coined the term ‘gene’
- Described gene as the fundamental, physical and functional unit of heredity

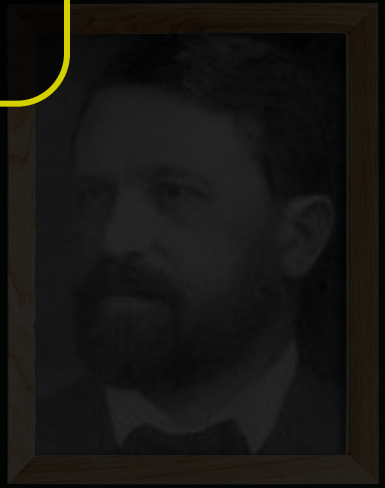
Chromosomal Theory of Inheritance

Genes are found on specific locations of the chromosomes and the behavior of these chromosomes during meiosis can explain the Mendel's laws of inheritance

Factors = Genes



Walter Sutton



Theodor Boveri

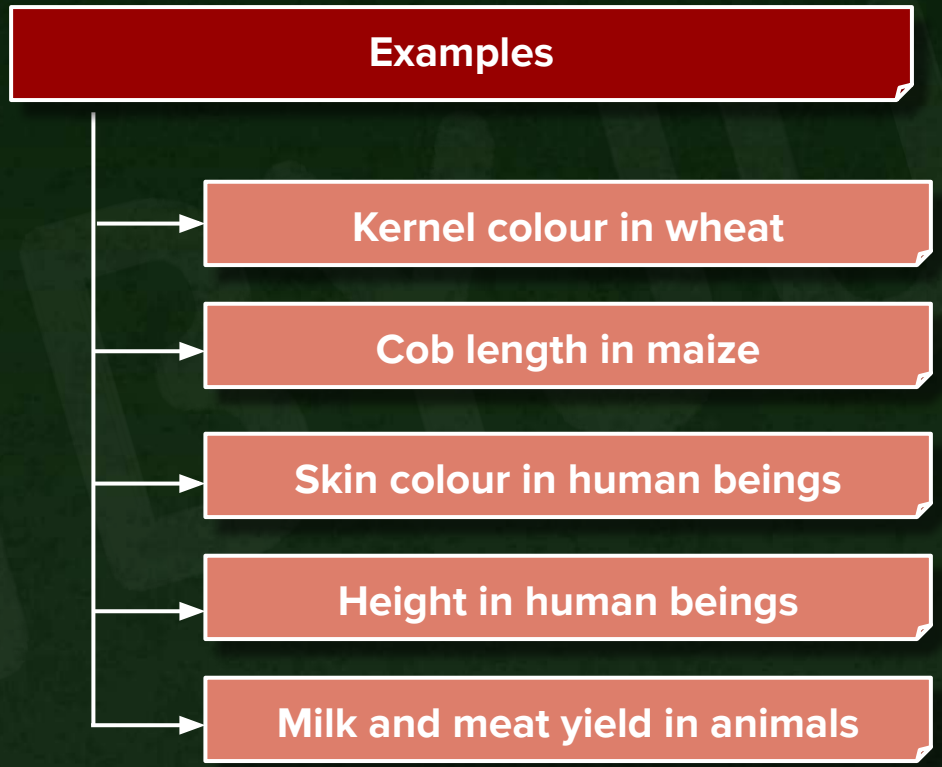
Polygenic Inheritance

- Characters like height and skin color in humans which do not have distinct traits and are spread like gradient are known as polygenic traits.
    - These are controlled by three or more genes.
    - The dominant alleles have a cumulative effect.
    - Each dominant allele expresses a part of the trait and the full trait is expressed only when all the dominant alleles are present.
    - The genes involved in such quantitative inheritance are called polygenes.
    - The inheritance of such type of traits is called polygenic inheritance.

Polygenic Inheritance

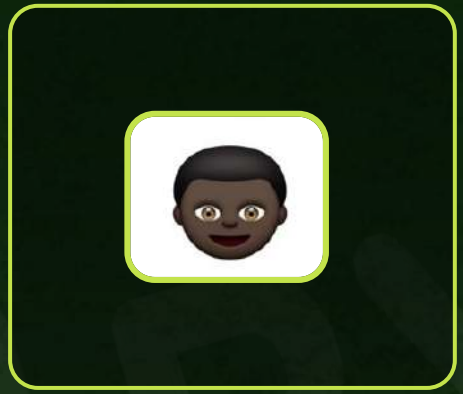
- The traits controlled by quantitative inheritance are sometimes known as metric traits because they can be measured in terms of unit of size, height, weight or number.
- They are also known as multiple factor inheritance.
- It is characterized by the occurrence of intermediate forms (continuous variations) between the parental types.
- A cross between two pure breeding parents does not produce dominant trait of one parent but instead an intermediate trait is exhibited.

Polygenic Inheritance

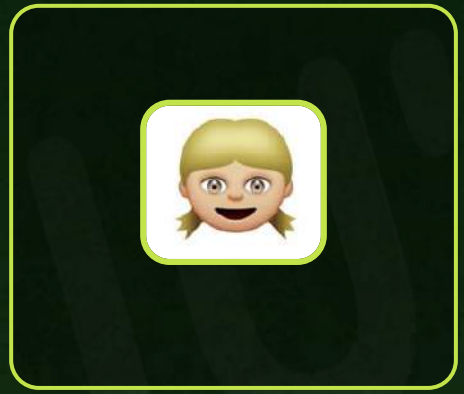


Polygenic Inheritance – Human Skin Colour

AA
BB
CC



X



aa
bb
cc

AABBCC

aabbcc

- Melanin pigments determine skin color.
- Assume that amount of melanin is determined by three genes" A", "B", "C".
- Dark skin tone is dominant gene. The dominant gene is responsible for fixed amount of melanin produced.

- White/very light is the phenotype of recessive alleles.

When a Dominant (black) parent is crossed with the recessive parent, the progeny can show any intermediate phenotype.

Polygenic Inheritance – Human Skin Colour

AABBCC

aabbcc

- The amount of melanin produced is proportional to number of dominant genes.
- This is the reason intermediate phenotypes are seen.
- For example: If the progeny has “aaBbCC”, the skin tone will be intermediate.

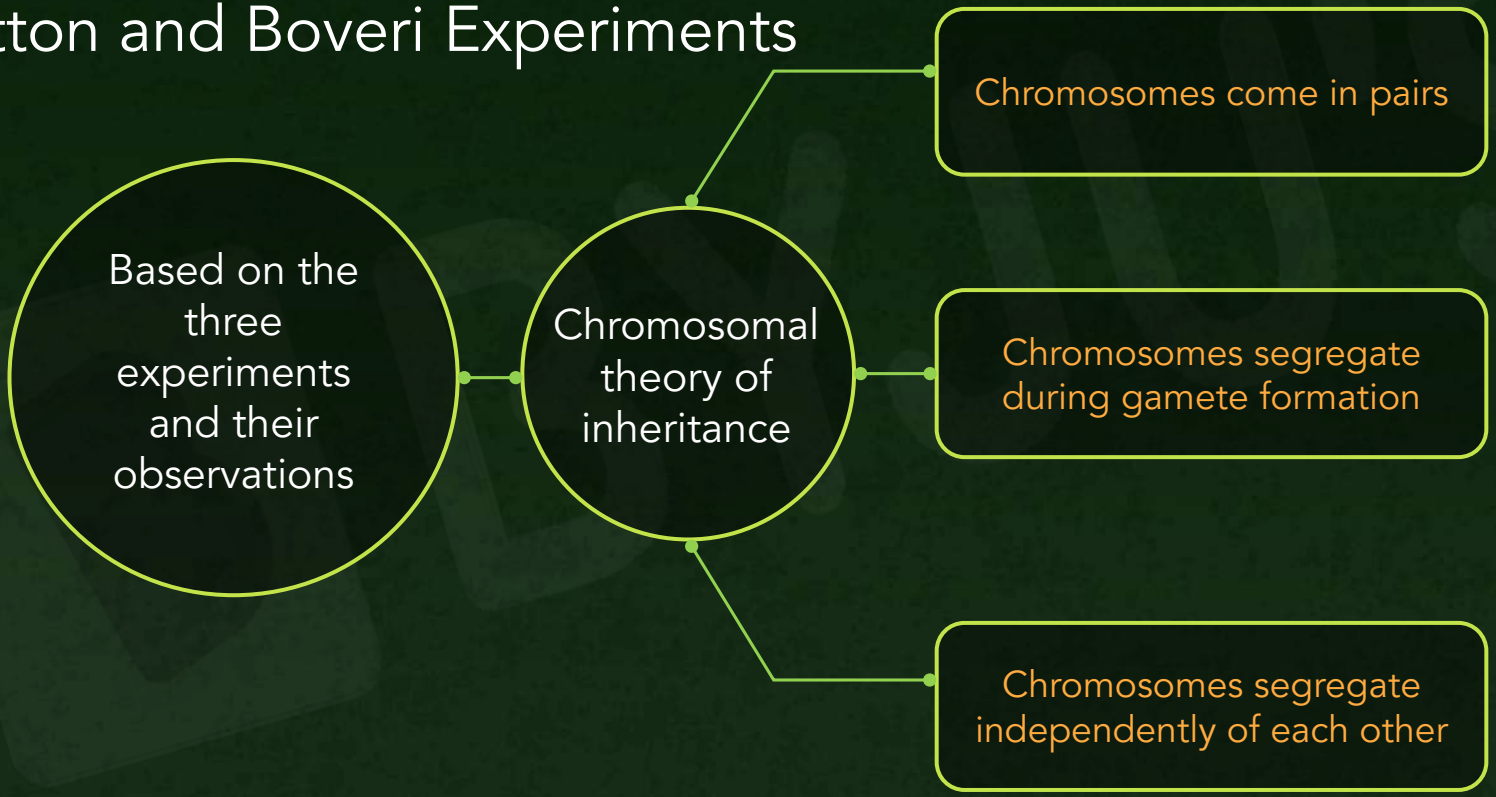
Did You Know?

Emojis for all

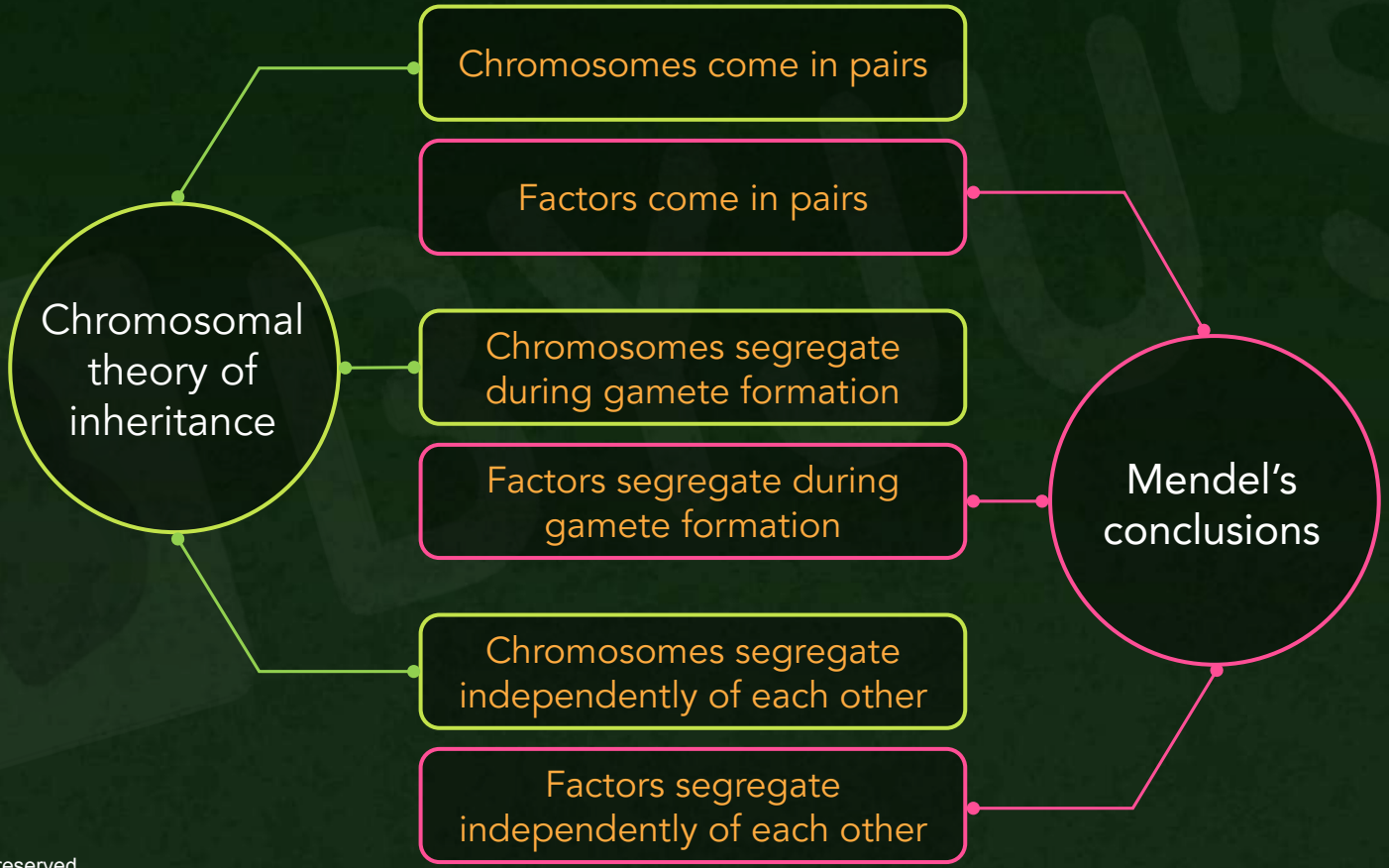
- Racism has no basis in science.
- Apps have included several different skin tones in their emojis.

Summary

Sutton and Boveri Experiments



Summary



Summary

Chromosomal theory of inheritance

Genes are found on specific locations of the chromosomes and the behavior of these chromosomes during meiosis can explain the Mendel's laws of inheritance.

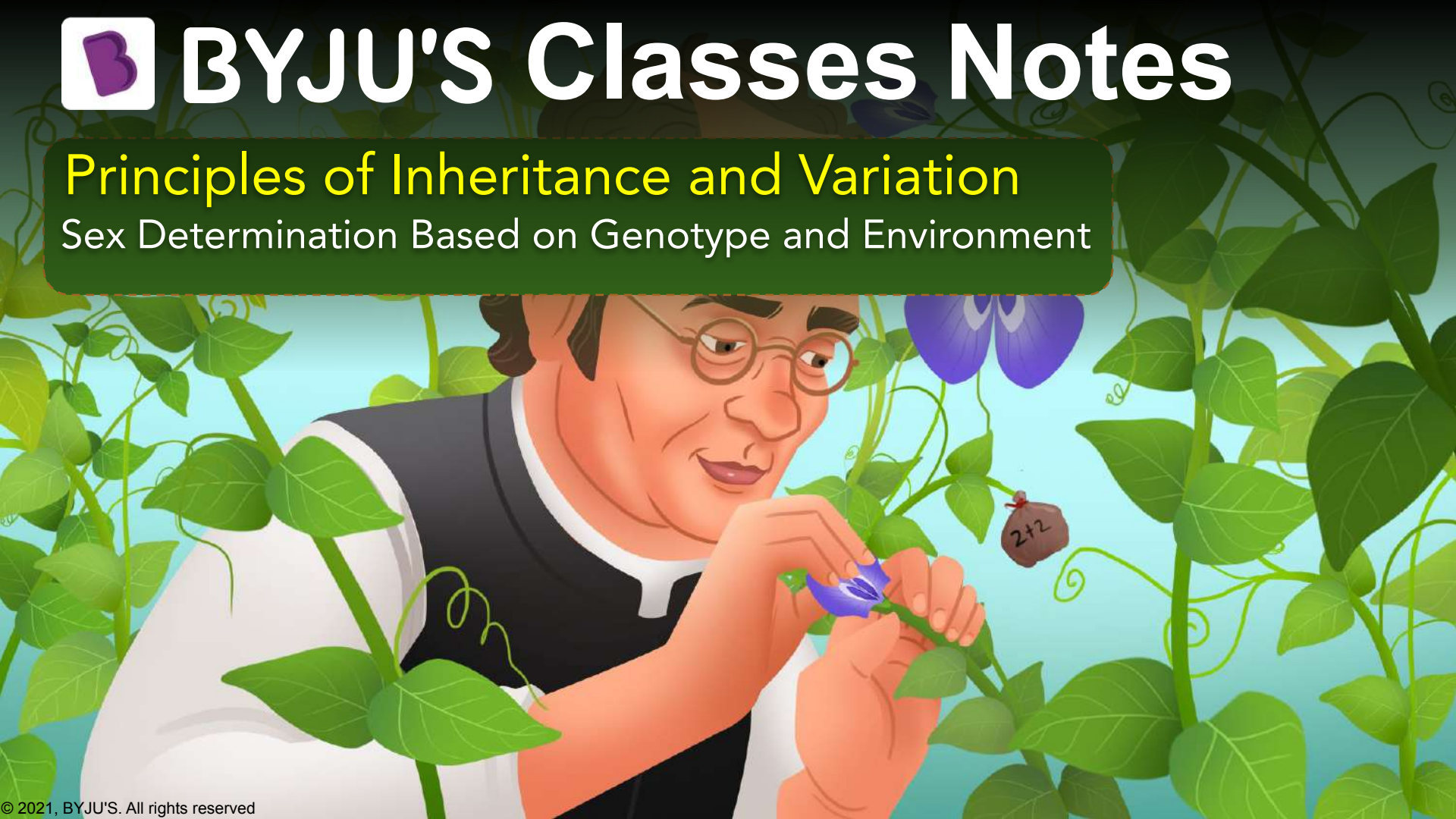
Summary

- Polygenic inheritance
  - Polygenic traits - Traits that do not occur distinctly, and are spread gradiently and controlled by two or more genes. This type of inheritance is called polygenic inheritance.
  - Eg: Skin color in humans

BYJU'S Classes Notes

Principles of Inheritance and Variation

Sex Determination Based on Genotype and Environment



Key Takeaways

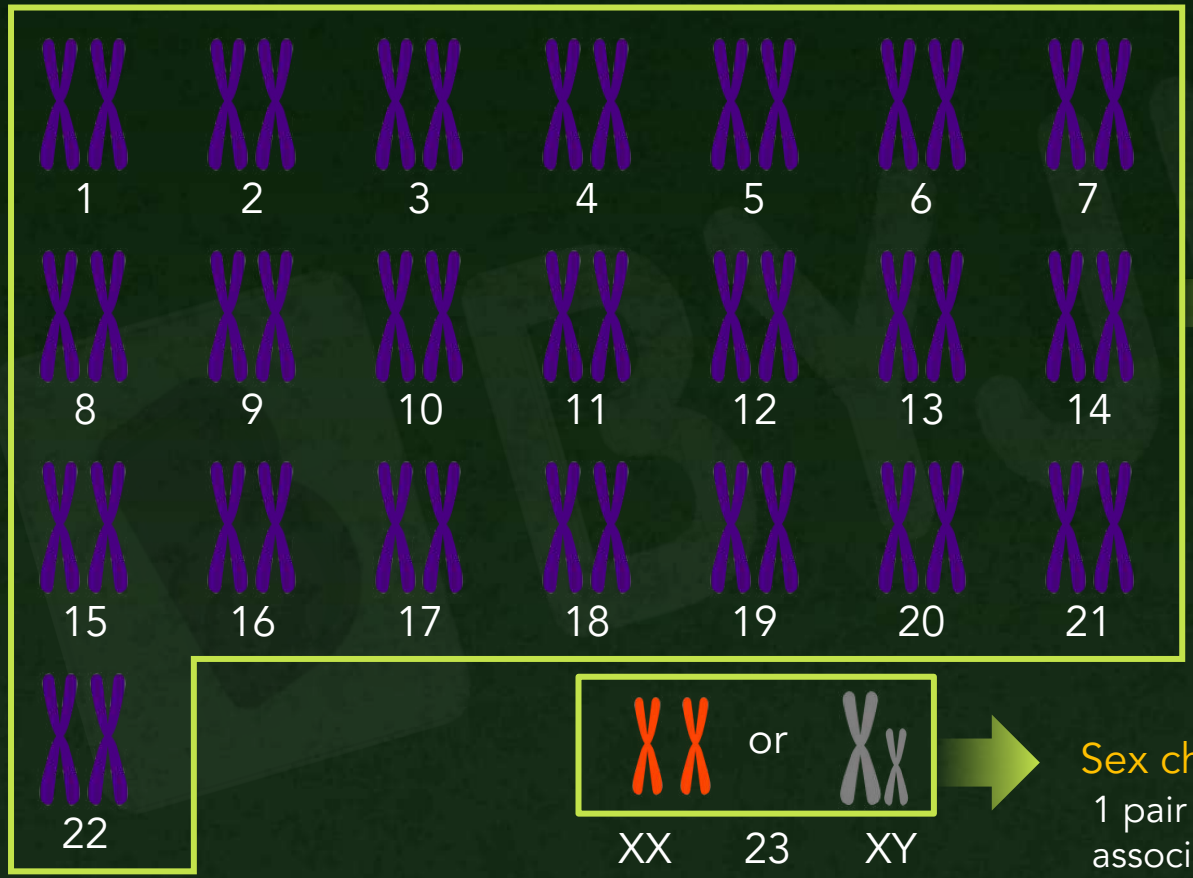
Sex determination

Genotypic sex determination

Environmental sex determination

Summary

Recall! Human Chromosomes



Autosomes

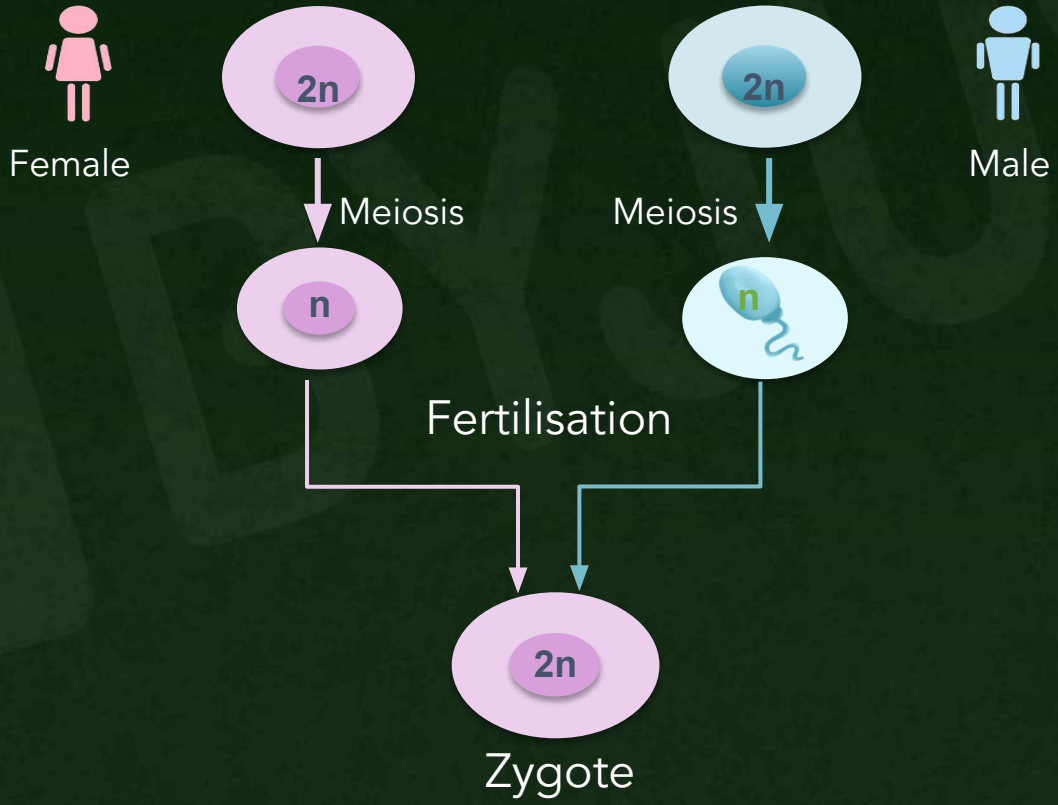
22 pairs of autosomes which are associated with somatic cells.

Sex chromosomes

1 pair of sex chromosomes associated with germ cells.

Recall! Fertilisation

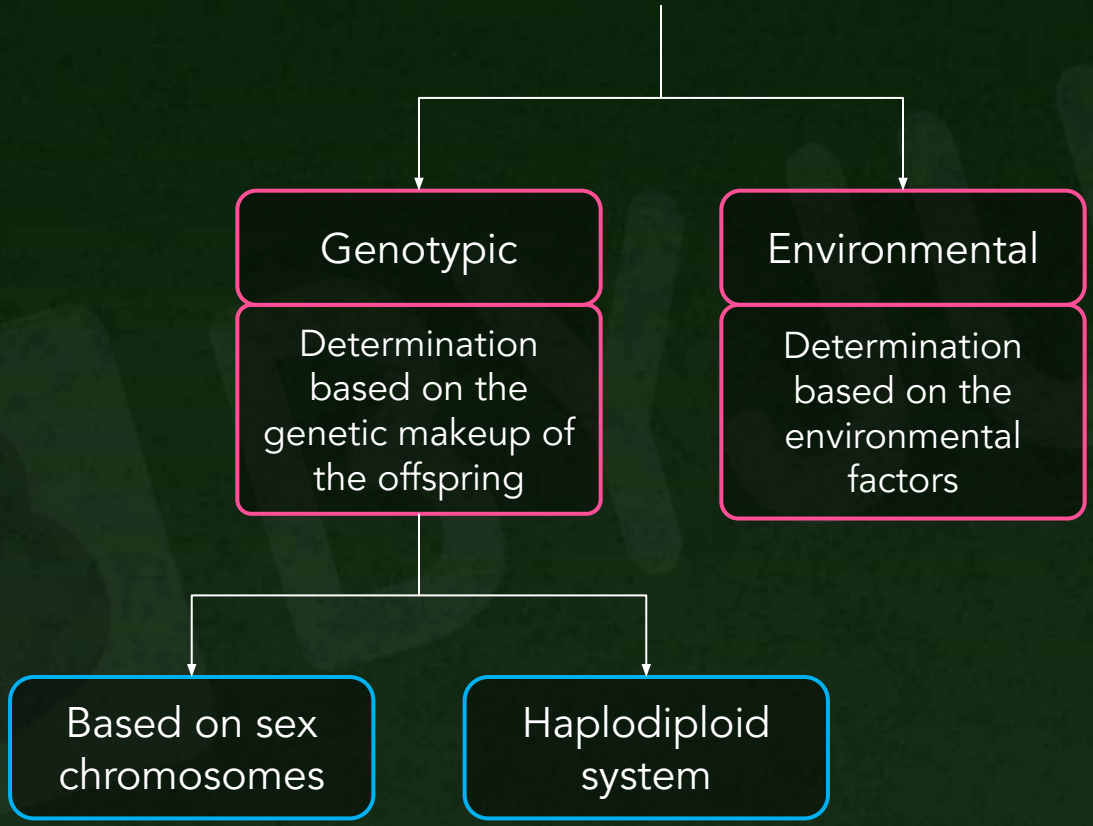
- During fertilisation, gametes fuse to form the zygote which develops into a foetus.



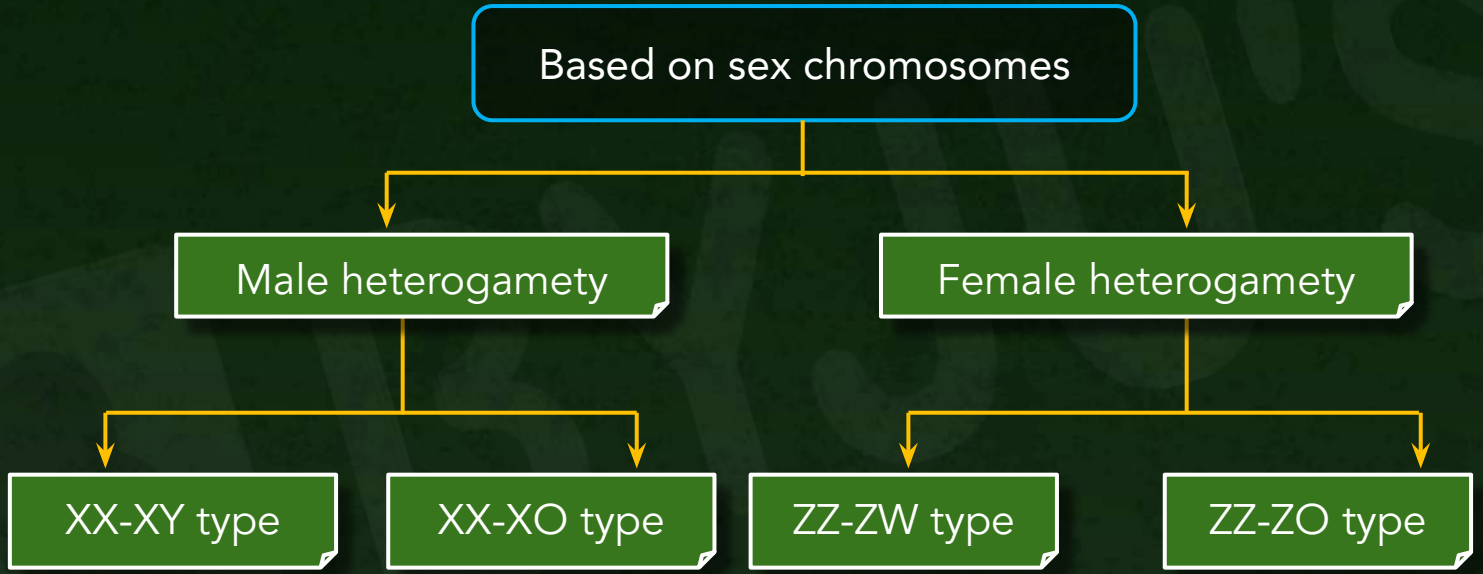
Sex Determination

- Sex determination is a biological mechanism which determines the development of sexual characteristics in an organism.
- It was first studied by Henking (1891) on insects.
- He discovered X chromosome and named it ‘X-body’.
- This lead to development of chromosomal basis of sex determination.

Sex Determination

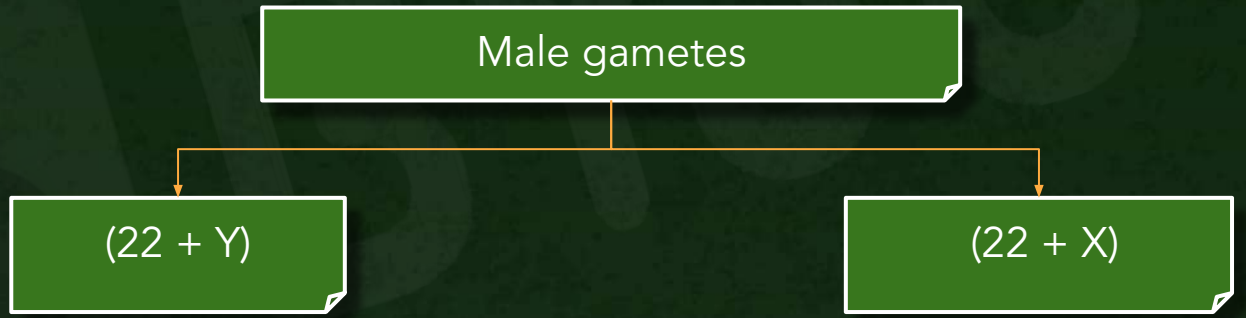


Sex Determination



XX-XY Type (*Lygaeus* Type)

- Males have 2 heteromorphic sex chromosomes i.e. X and Y.
- Females have 2 homomorphic or isomorphic sex chromosomes i.e. X and X.
- Example: Most of the mammals.



- Fertilisation of egg with sperm having X chromosome produces female child (44 + XX).
- Fertilisation of egg with sperm having Y chromosome produces male child (44 + XY).

Genotypic Sex Determination

Sex determination-Humans

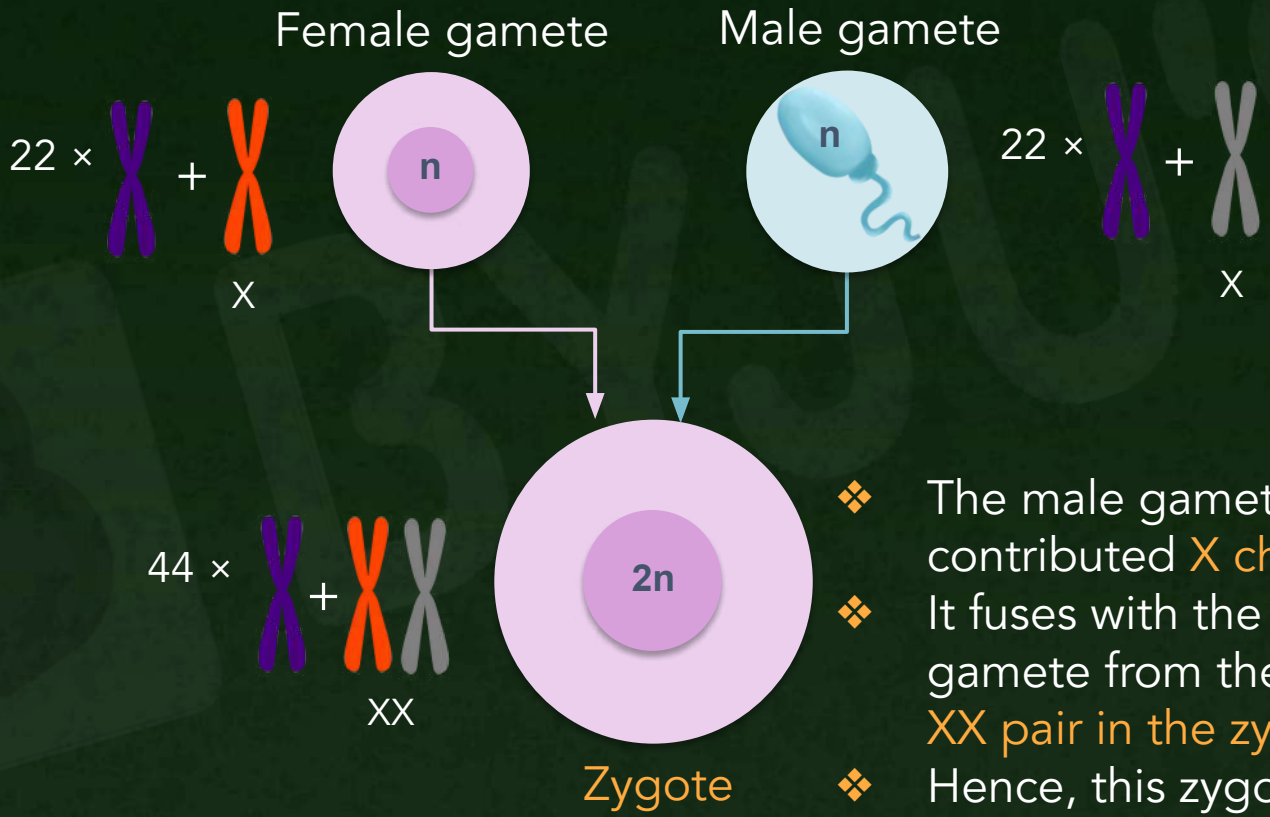
♂ \ ♀	X	X
X	X X	X X
Y	X Y	X Y

50% female

50% male

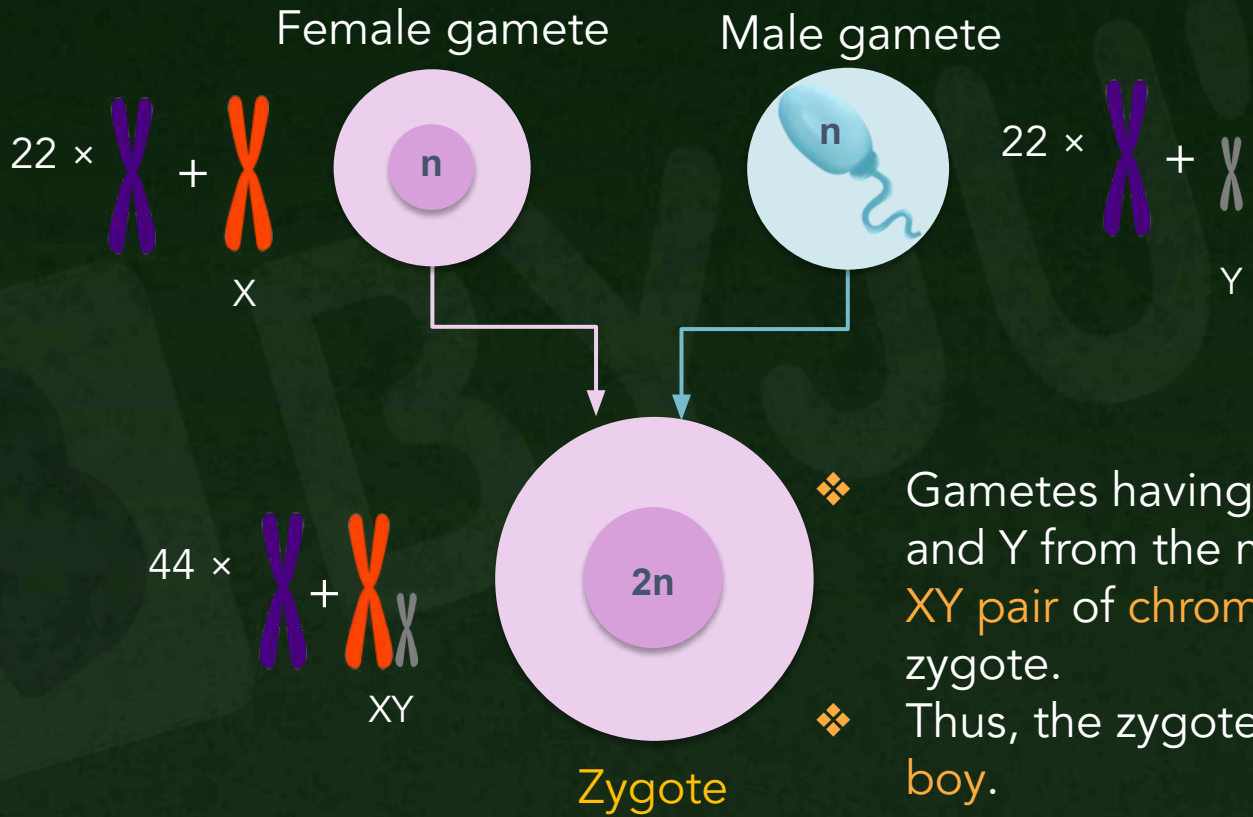
- Female always contributes X as the sex chromosome to the zygote.
- Sex chromosome contributed by the male (X or Y), determines the sex of the offspring.
- It is the male sperm that determines whether offspring will be a girl or a boy, chances being 50% for each.

Sex Determination-Humans



- The male gamete has contributed X chromosome.
- It fuses with the X chromosome gamete from the female to form XX pair in the zygote.
- Hence, this zygote is a girl.

Sex Determination-Humans

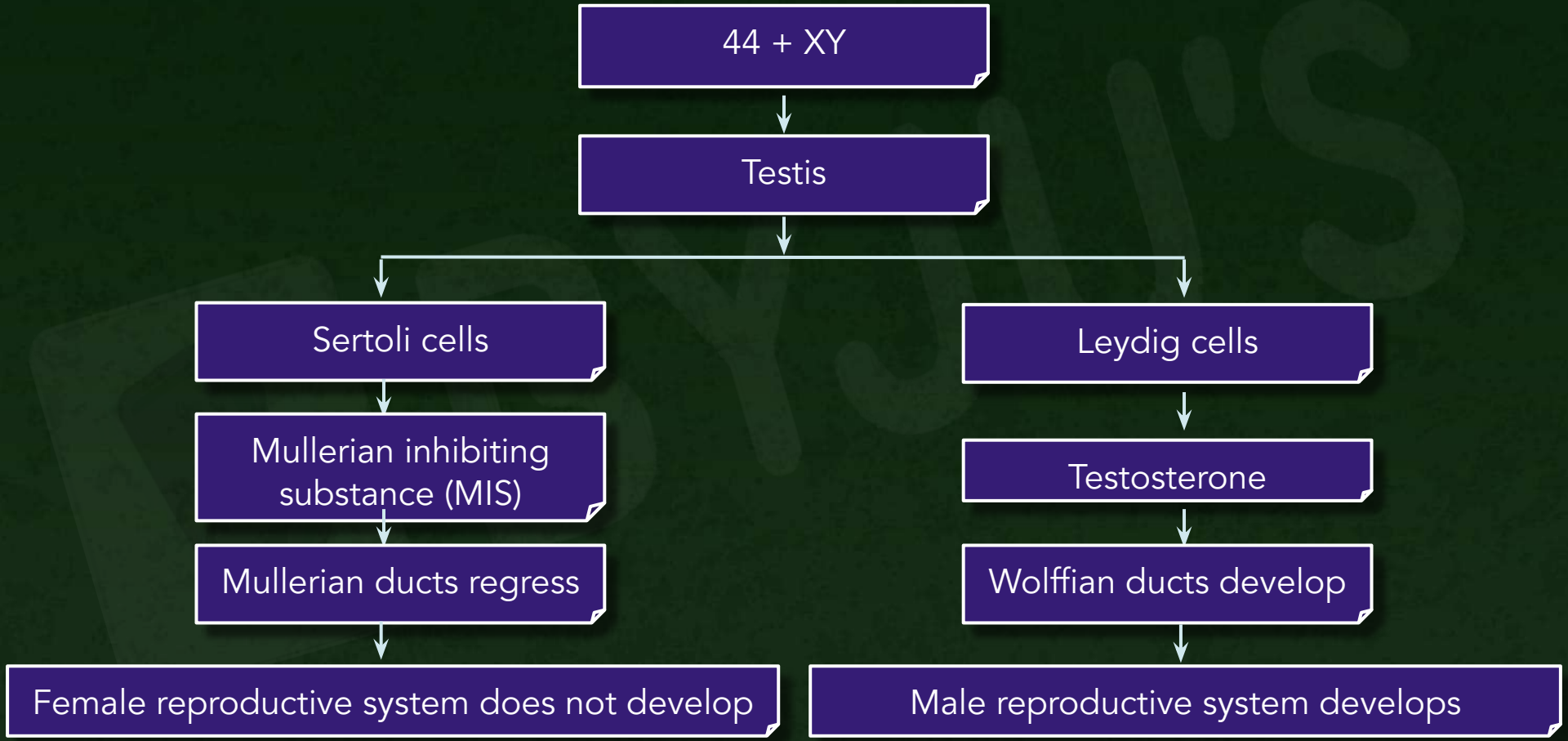


- Gametes having X from the female and Y from the male fuse to give XY pair of chromosomes in the zygote.
- Thus, the zygote is 44+XY i.e a boy.

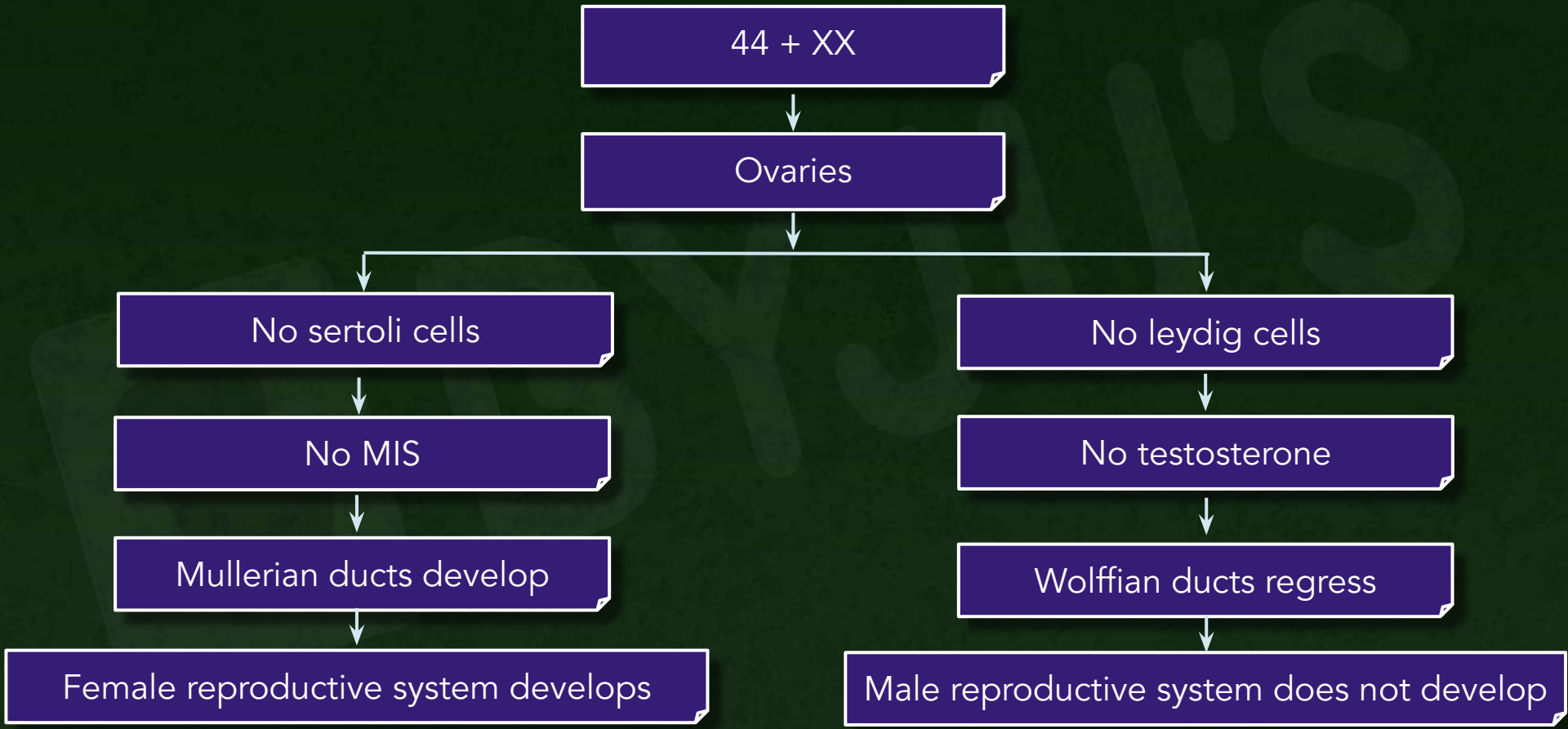
Sex Determination-Humans

- So, it is the father's sperm (that fertilises the egg) which decides whether the baby will be a girl or a boy.
- It is totally a random method, as man has no control over the sex chromosome in his sperm either.
- Nobody should be blamed for giving birth to a child of certain sex.

Sex Determination-Humans

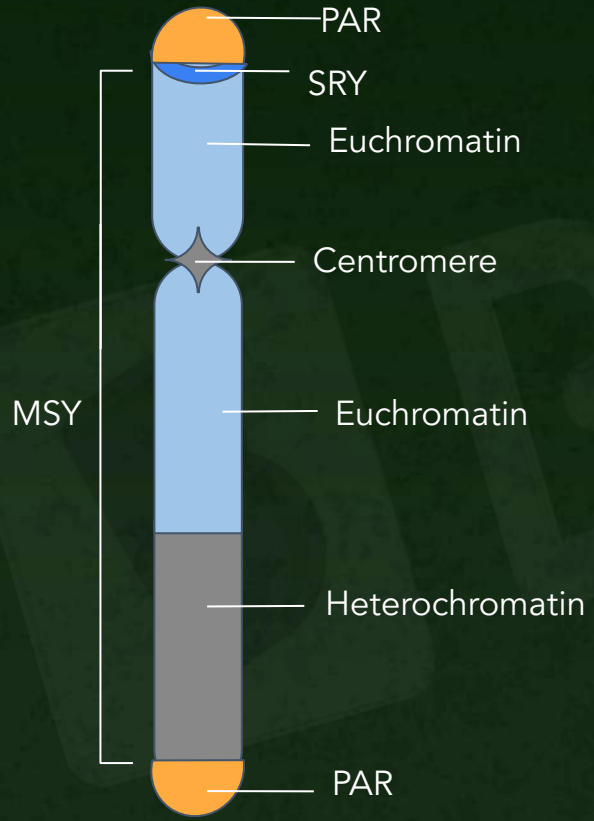


Sex Determination-Humans



Did You Know?

Y chromosome



- The human Y chromosome, was long thought to be mostly blank genetically.
- However, data has indicated that Y chromosome has several genes.
- Y chromosome is the smallest of all the chromosomes.

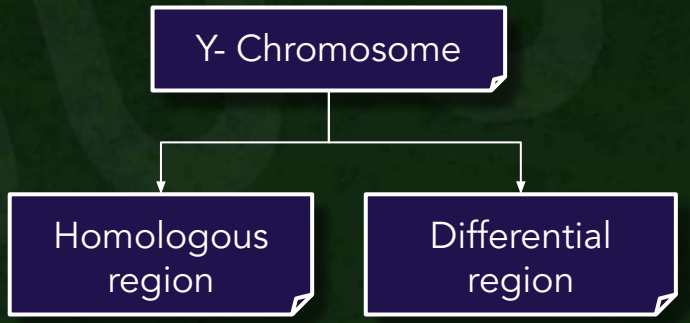
Key: PAR: Pseudoautosomal region

SRY: Sex-determining region Y

MSY: Male-specific region of Y

Y Chromosome: Differential Region

- Differential region of Y chromosome carries only Y- linked genes or holandric genes.
- It has SRY (Sex determining region) which codes for TDF (Testis determining factor).
- TDF is required for the development of male sex.
- In absence of TDF, female sex develops.
- On both ends of the Y chromosome, pseudoautosomal regions (PARs) are present.
- These share homology with regions on the X chromosome and synapse and recombine with it during meiosis.

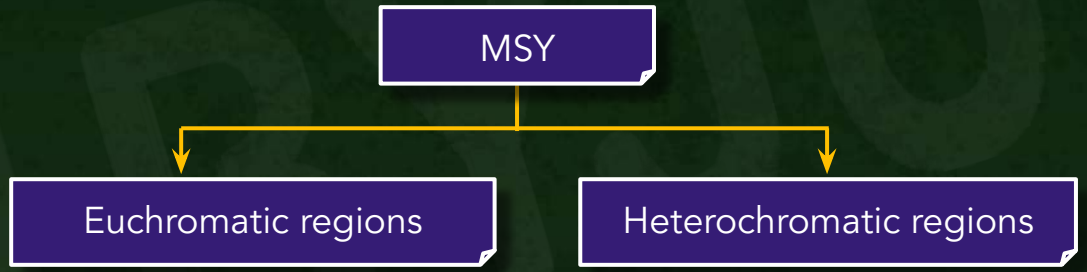


Y Chromosome: Differential Region

- The remainder of the chromosome, does not synapse or recombine with the X chromosome.
- As a result, it was originally referred to as the non-recombining region of the Y (NRY).
- More recently, researchers have designated this region as the male-specific region of the Y (MSY).

Y Chromosome: Differential Region

- The MSY is divided between euchromatic regions, containing functional genes, and heterochromatic regions, lacking genes.

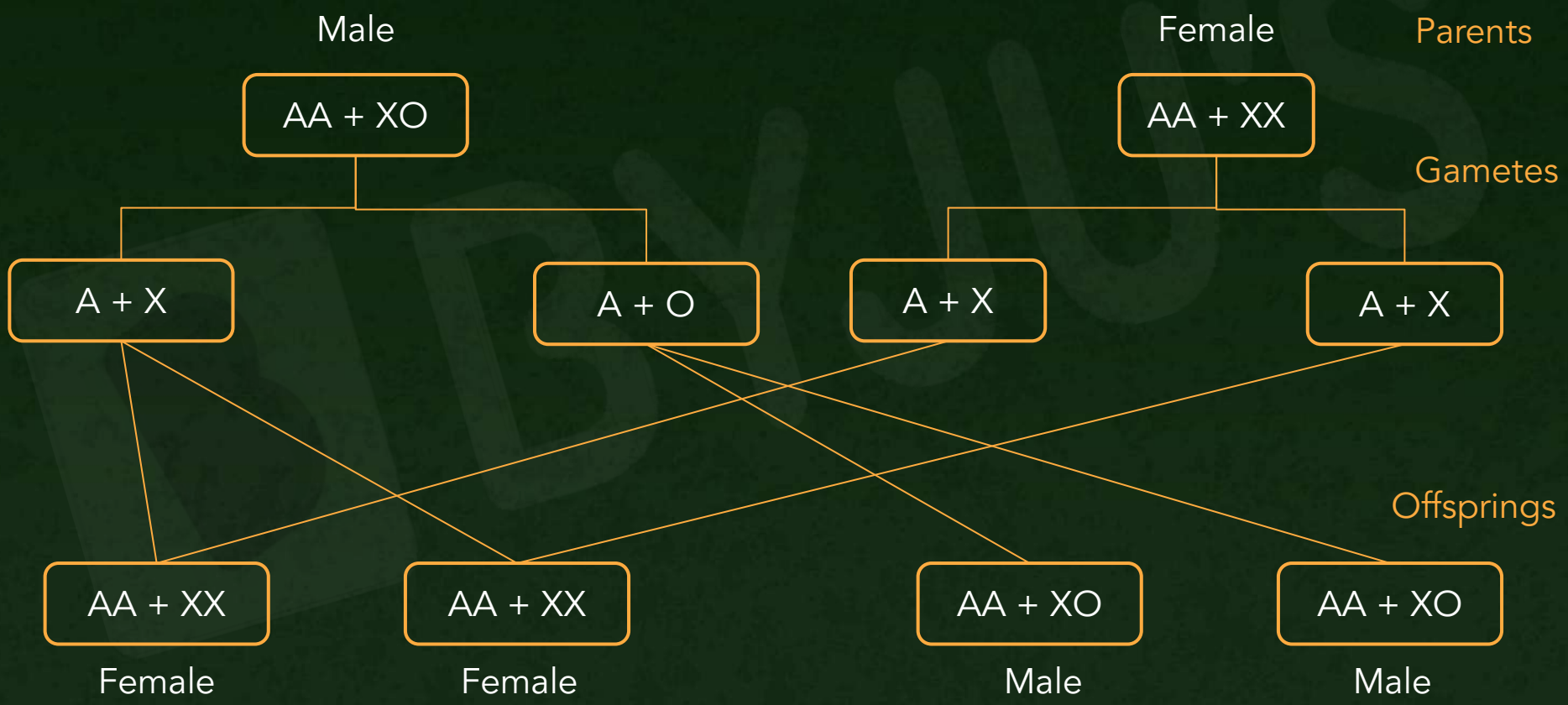


- Within euchromatin, adjacent to the PAR of the short arm of the Y chromosome, is the critical gene that controls male sexual development, the sex determining region Y (SRY).

Sex Determination- XX-XO Type

- In roundworms and some insects (grasshoppers, cockroaches), the females have two sex chromosomes, XX, while the males have only one sex chromosome, X (designated as X0).
- Female gametes are homogametic (A+X).
- Males are heterogametic with half the male gametes carrying X-chromosome (A+X) while the other half being devoid of it (A+ 0).

Sex Determination- XX-XO Type



Sex Determination- ZZ-ZW Type

- In birds and some reptiles both the sexes possess two sex chromosomes.
- The females contain heteromorphic sex chromosomes (AA + ZW) while the males have homomorphic sex chromosomes (AA+ ZZ).
- The females are heterogametic and produce two types of eggs, (A + Z) and (A + W).
- The male gametes or sperms are of one type (A + Z).

Sex Determination - Birds

♂ \ ♀	Z	W
Z	Z Z	Z W
Z	Z Z	Z W

50% Males 50% Females

Sex chromosomes → Z | W

Female → ZW

Male → ZZ

- Birds have a similar system as human but the difference is the female sex chromosomes determines the sex of the offspring.
- The chances of offspring being a male or female is 50% each.

Sex Determination- ZZ-ZO Type

- This type of sex determination occurs in some butterflies and moths.
- Here the females have odd sex chromosome (AA + Z) while the males have two homomorphic sex chromosomes (AA + ZZ).
- Females produce two types of eggs, one with male producing sex chromosome (A + Z) and female forming without the sex chromosome (A + 0).
- The males are homogametic, forming similar types of sperms (A + Z).

Sex Determination - Fruit Fly

No. of X chromosomes : Pair of autosomes

Case 1:

XX : AA

↓

2:2

= 1

Female

Male

Case 2:

XY : AA

↓

1:2

= 0.5

- *Drosophila* has 3 pairs of chromosomes.
- 2 pairs are autosomes and 1 pair is that of sex chromosomes.
- The sex chromosome may be XX or XY.
- Depending upon the number of X chromosomes present, the ratio varies which determines the sex of the *Drosophila*.

Sex Determination - Fruit Fly

Drosophila melanogaster

- Sex chromosomes → X | Y
- Y has no role in sex determination
- Ratio - no. of X chromosomes : pair of autosomes

Sex Determination in Grasshopper

Female → XX Male → XO

♂ \ ♀	X	X
X	X X	X X
O	XO	XO

50% Females

50% Males

- In grasshopper, the males have only one X chromosome, besides the autosomes whereas female has two X chromosomes.
- Eggs fertilized by sperm having X chromosome become females and those fertilized by sperm, that do not have X chromosome become males.

Did You Know?

Fact - It is always a girl!

Desert grassland whiptail lizard

- The desert grassland whiptail lizard (*Aspidoscelis uniparens*), is an all-female species of reptiles in North America.
- It's reproduction process does not need male fertilisation.
- The lizards reproduce by parthenogenesis.

Sex Determination-Haplodiploid System

Worker ♀ | Drone ♂ | Queen ♀

- Honeybees and ants have a hierarchy in their society.
- There is a queen and there are several drones and workers.
- Drones are males whose role is to fertilize a receptive queen.
- Workers are females who are infertile.
- The jobs of worker bees include housekeeping, feeding the all bees and larvae, collecting the pollen and nectar and making the wax.
- Basically, everything in the hive is done by worker bees.

Sex Determination-Haplodiploid System

Based on the number of sets of chromosomes

- Sex determination in honey bees is based on the number of sets of chromosomes an individual receives.
- When a queen mates with the drone, she stores the sperms of the drone.

Sex Determination - Haplodiploid System

There are 2 possible cases

Case 1: When queen needs more workers in the society .

- Queen utilises the sperms and an offspring is formed from the union of a sperm and an egg.
- Zygote develops as female. (Sperms from drone and egg from queen bee.)
- Thus, the females obtained are diploid.

Case 2: When queen needs more drones in the society.

- She does not utilise the sperm and an offspring is formed from the unfertilised egg (Queen bee produces the egg).
- This is called parthenogenesis.
- Offspring obtained is the male drone.
- This means that the males have half the number of chromosomes than that of a female (drones are haploid).

Sex Determination-Haplodiploid System

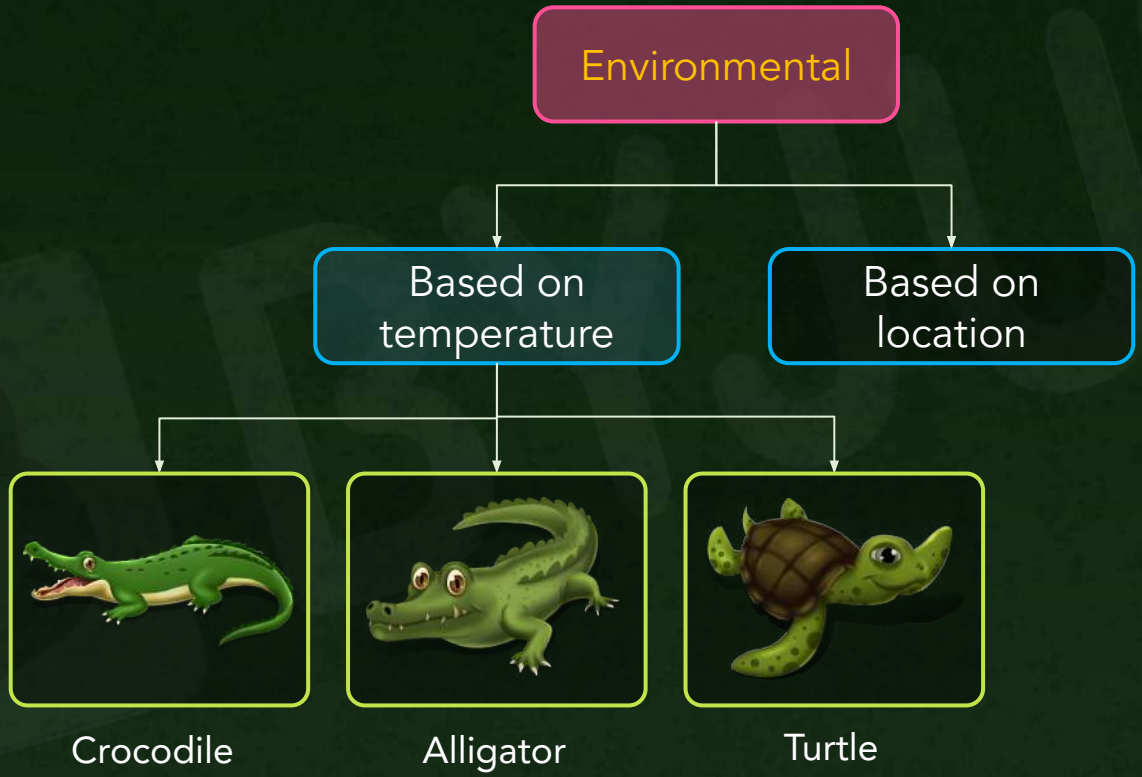
- Based on the number of sets of chromosomes
- Female (Queen/worker) → Diploid (fusion of gametes)
- Male (Drone) → Haploid (unfertilised egg)
- Males produce sperms by mitosis

Worker ♀

Drone ♂

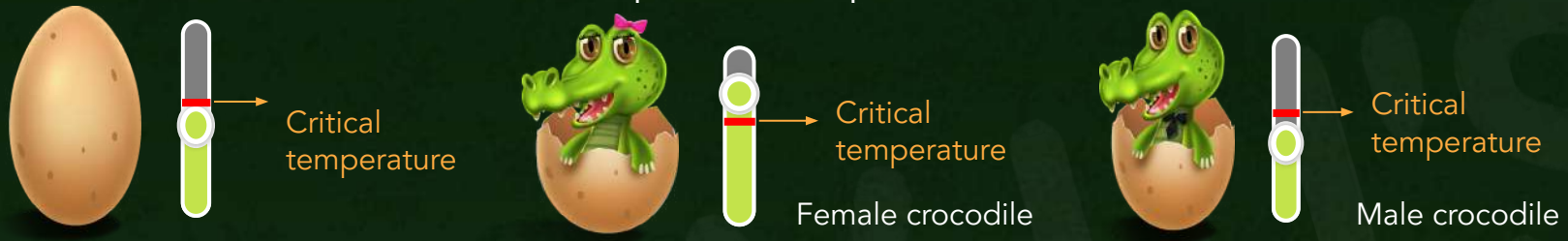
Queen ♀

Sex Determination



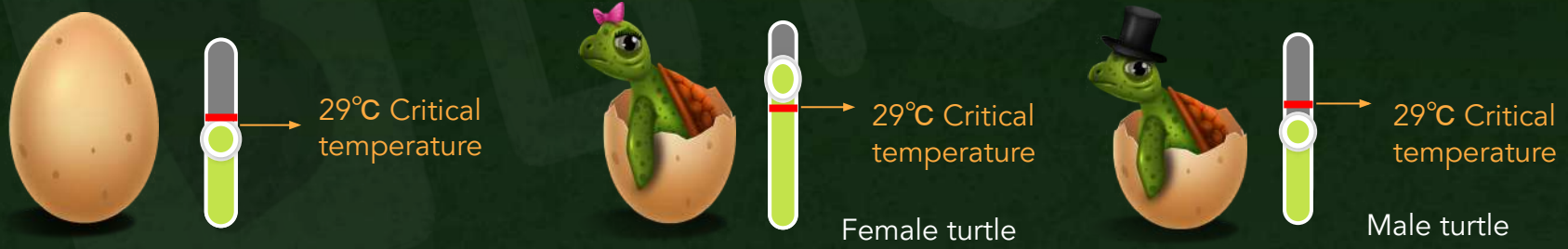
Temperature Dependent–Crocodile & Alligator

Temperature dependent – crocodile



- Sex determination for both crocodiles and alligators is same. If the eggs are subjected to higher temperatures, it is a female and if the eggs are subjected to lower temperatures, it is a male.

Temperature dependent – turtle



- For turtles - If the eggs are subjected to temperatures higher than 29^0C it is a female, and if the eggs are subjected to temperatures lower than 29^0C, it is a male.

Sex Determination

Environmental

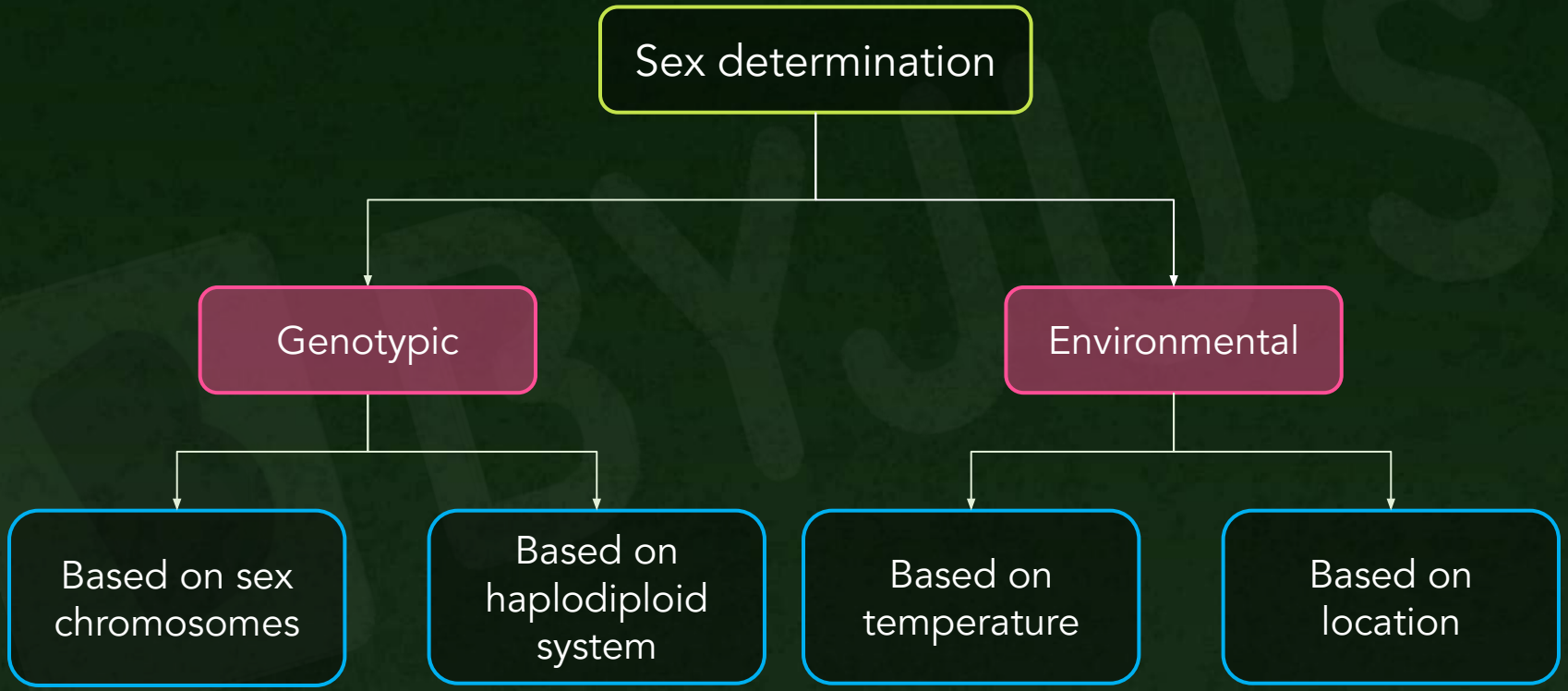
- Based on temperature
- Based on location

Green spoon worm

Based on location of larvae landing

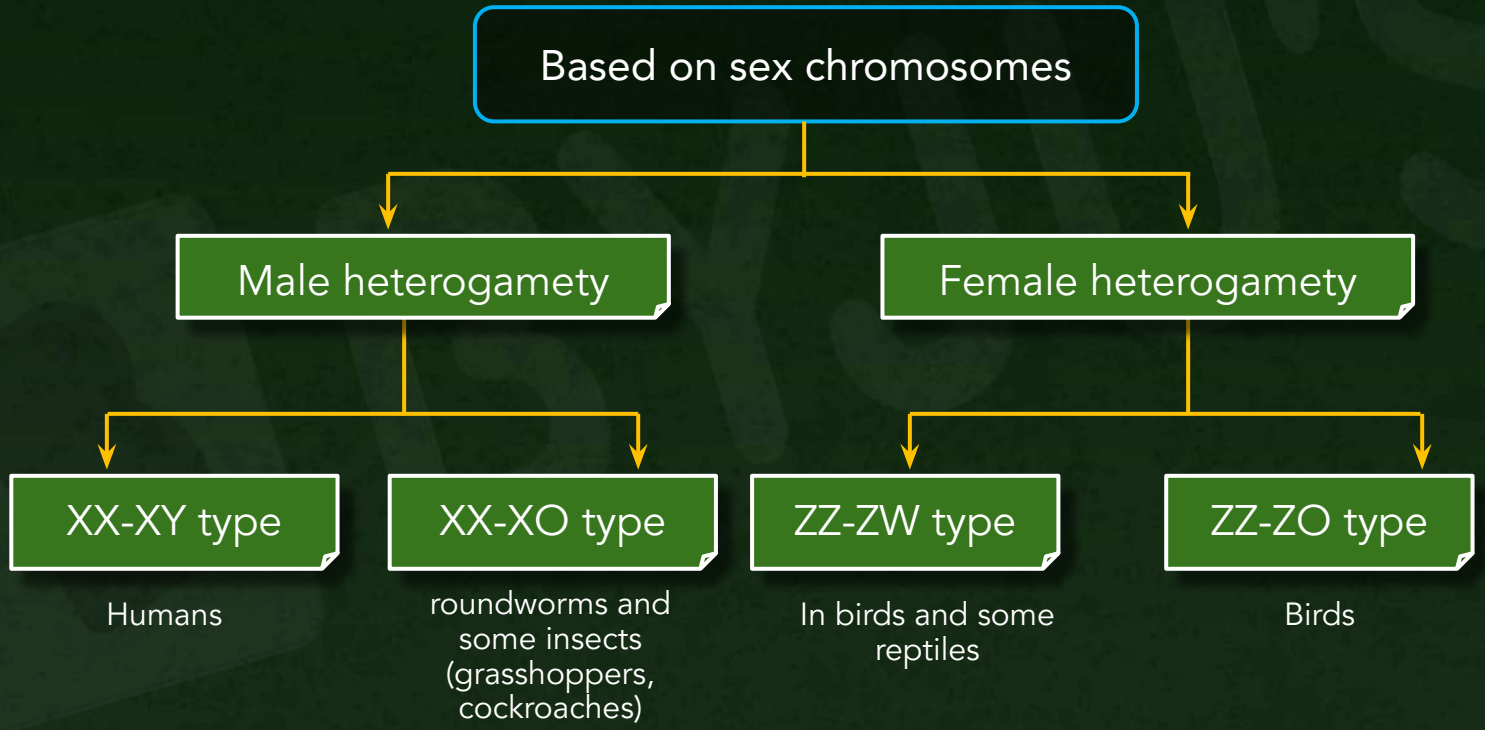
- Larvae lands on ocean floor → Female
- Larvae attached to female's body → Male

Summary



Summary

Genotypic: based on sex chromosomes



Summary

Genotypic: based on haplodiploid system

Worker

Drone

Queen

Case 1: When queen needs more workers in the society .

- Queen utilises the sperms and an offspring is formed from the union of a sperm and an egg.
- Zygote develops as female. (Sperms from drone and egg from queen bee.)
- Thus, the females obtained are diploid.

Case 2: When queen needs more drones in the society.

- She does not utilise the sperm and an offspring is formed from the unfertilised egg (Queen bee produces the egg).
- Offspring obtained is the male drone.
- This means that the males have half the number of chromosomes than that of a female (drones are haploid).

Summary

Environmental : based on temperature

- Sex determination for both crocodiles and alligators is same. If the eggs are subjected to higher temperatures, it is a female and if the eggs are subjected to lower temperatures, it is a male.

- For turtles - If the eggs are subjected to temperatures higher than 29^0C it is a female, and if the eggs are subjected to temperatures lower than 29^0C, it is a male.

Summary

Environmental : based on location

Green spoon worm

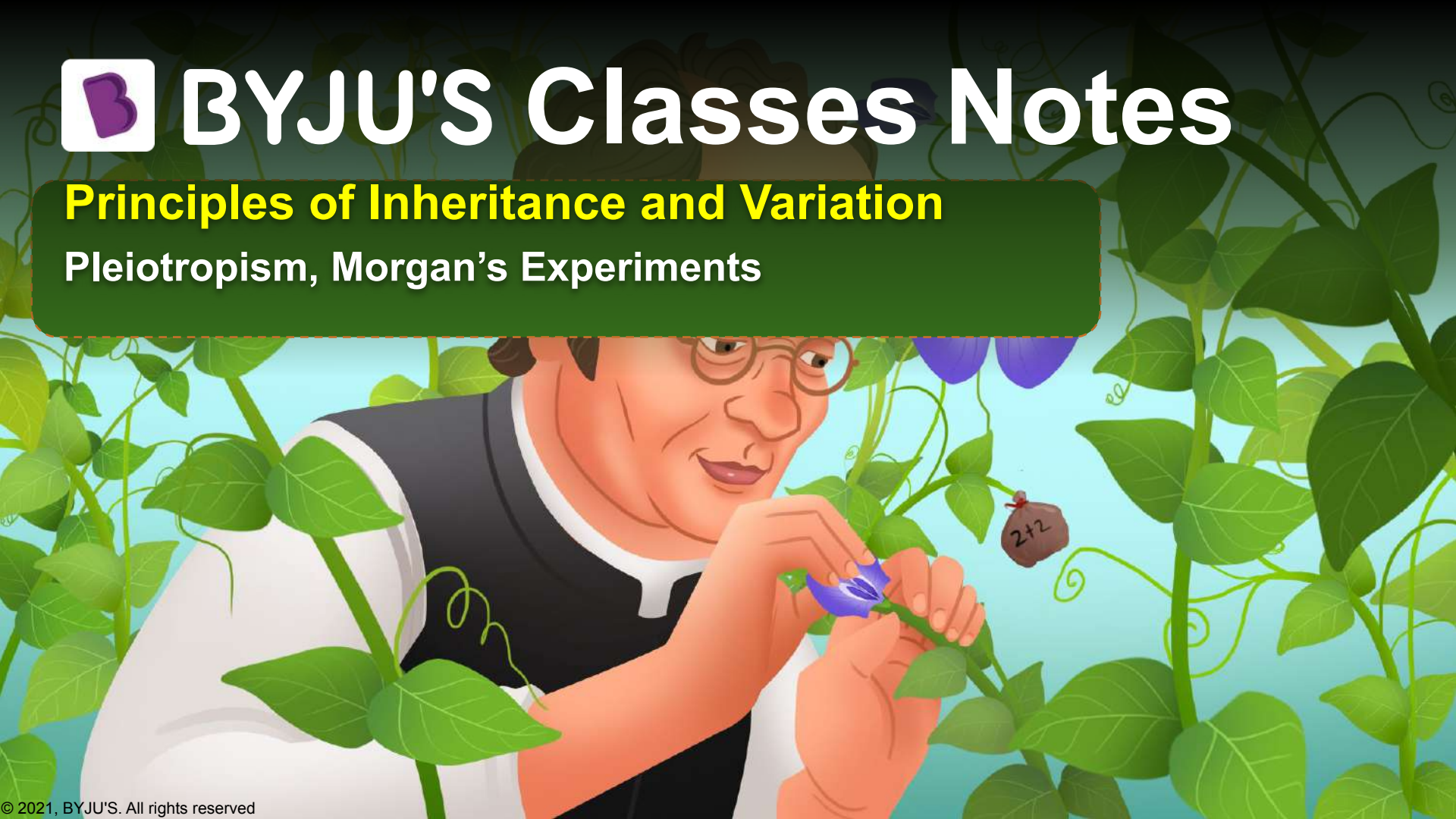
Based on location of larvae landing

- Larvae lands on ocean floor → Female
- Larvae attached to female's body → Male

BYJU'S Classes Notes

Principles of Inheritance and Variation

Pleiotropism, Morgan's Experiments



Key Takeaways

1. **Pleiotropism**
   - Sickle cell anaemia
   - Phenylketonuria
   - Kartagener's syndrome
   - Cystic fibrosis
2. **Morgan's monohybrid cross**

Summary

Pleiotropism

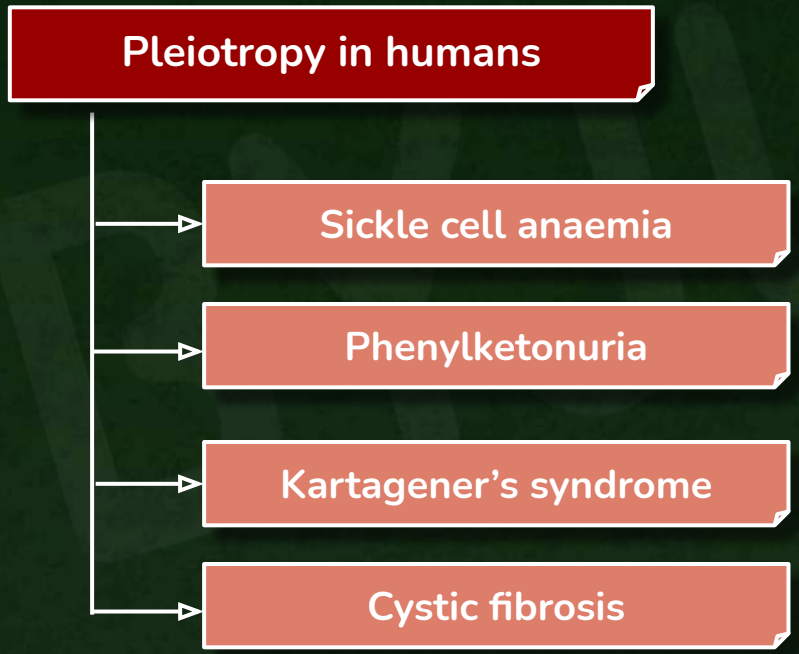
- Single gene exhibit multiple phenotypic expression
- Example: Effect of a gene on metabolic pathway which contributes to different phenotype

Example - Phenylketonuria

- Effected enzyme : Phenylalanine Hydroxylase (Enzyme is either missing or severely reduced.)
- Caused due to single gene mutation.
- So the metabolic pathway associated with the enzyme is disrupted and leads to multiple phenotypes.

Diseased state - phenylketonuria

Pleiotropism

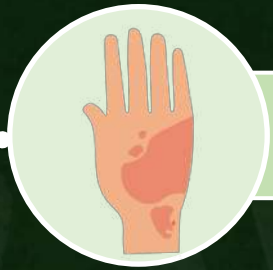


Sickle Cell Anaemia

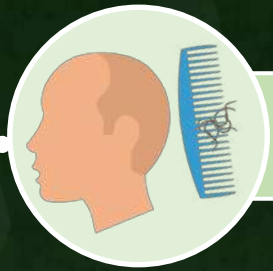
- Single gene exhibit multiple phenotypic expression
- Causative genes alter the type of haemoglobin and also change the form of RBCs.

Pleiotropism - Phenylketonuria

Symptoms of phenylketonuria



Skin pigmentations



Reduction in hair

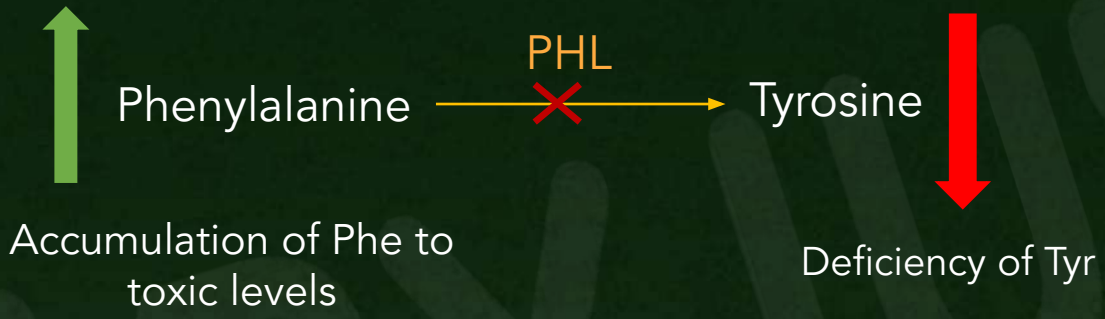
Mental retardation

Phenylketonuria(PKU)

- It is an autosomal recessive metabolic disorder resulting from lack of an enzyme known as phenylalanine hydroxylase (PHL).
- PHL helps to convert phenylalanine into tyrosine.

PHL-Phenylalanine hydroxylase

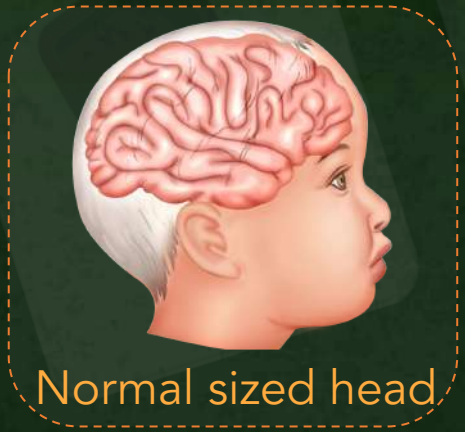
Phenylketonuria



- Phenylketonuria results in accumulation of phenylalanine to toxic levels and the deficiency of tyrosine.
- Tyrosine is required for synthesis of various neurotransmitters.
- Tyrosine deficiency results in reduced brain development and other neurological conditions.
- Excess presence of PHL in cerebrospinal fluid (CSF) causes mental retardation, intellectual disability and mental disorders.

PHL- Phenylalanine hydroxylase

Phenylketonuria

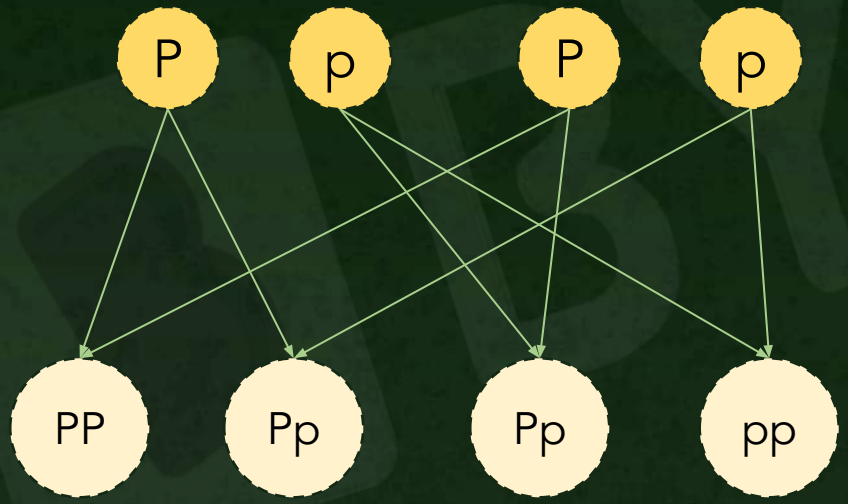


Symptoms

- Small sized head (microcephaly)
- Learning disability and delayed development
- Neurological disorders
- Behavioural, emotional and social problems
- Fatigue

Phenylketonuria(PKU)

Carrier x Carrier



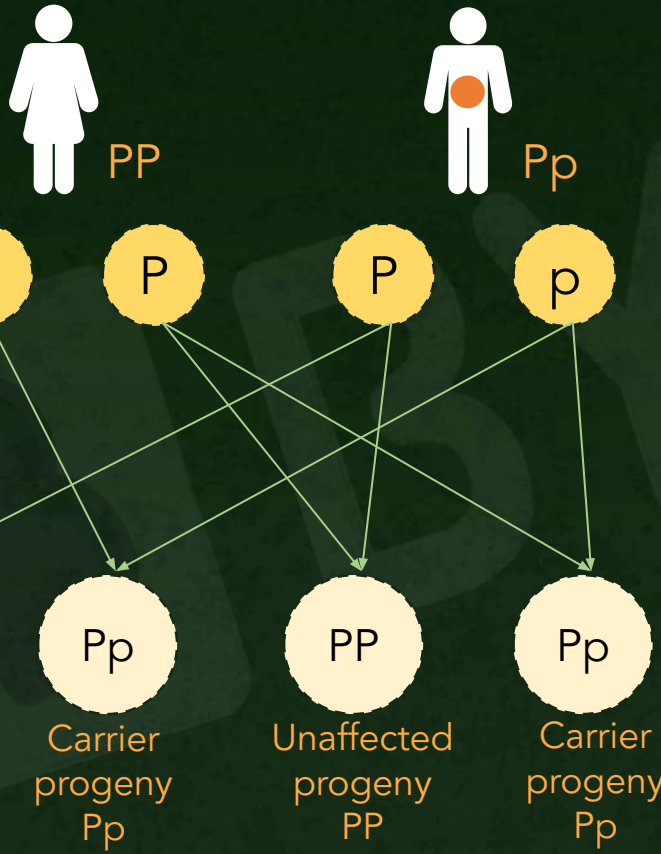
Unaffected progeny PP

Carrier Progeny (50%) Pp

Affected progeny pp

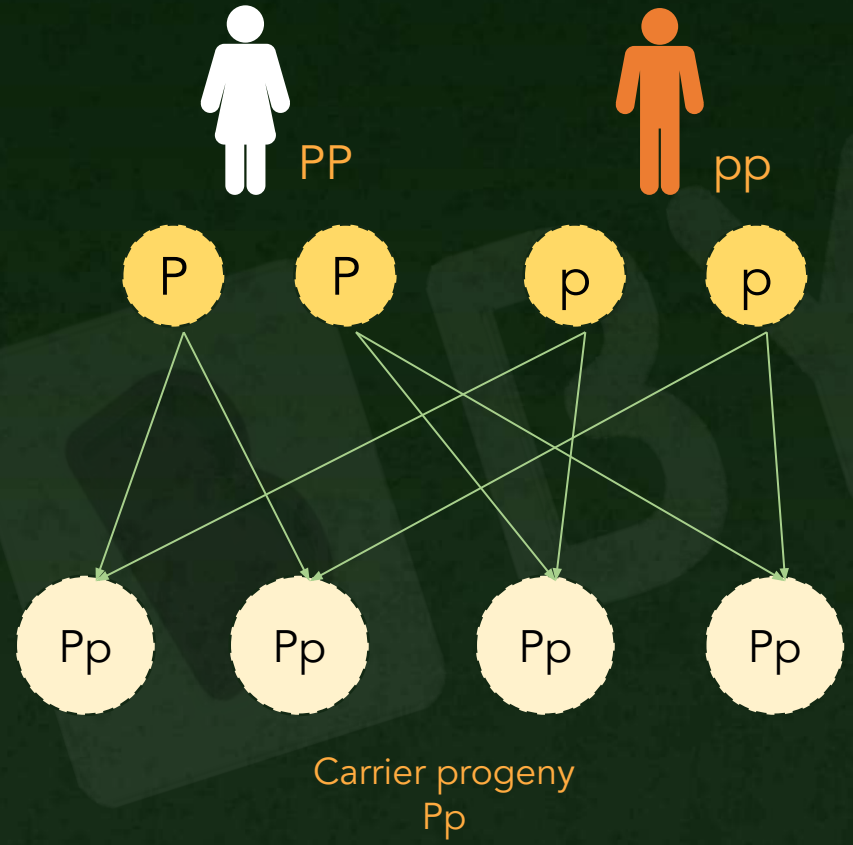
Phenylketonuria

Unaffected x Carrier



Phenylketonuria

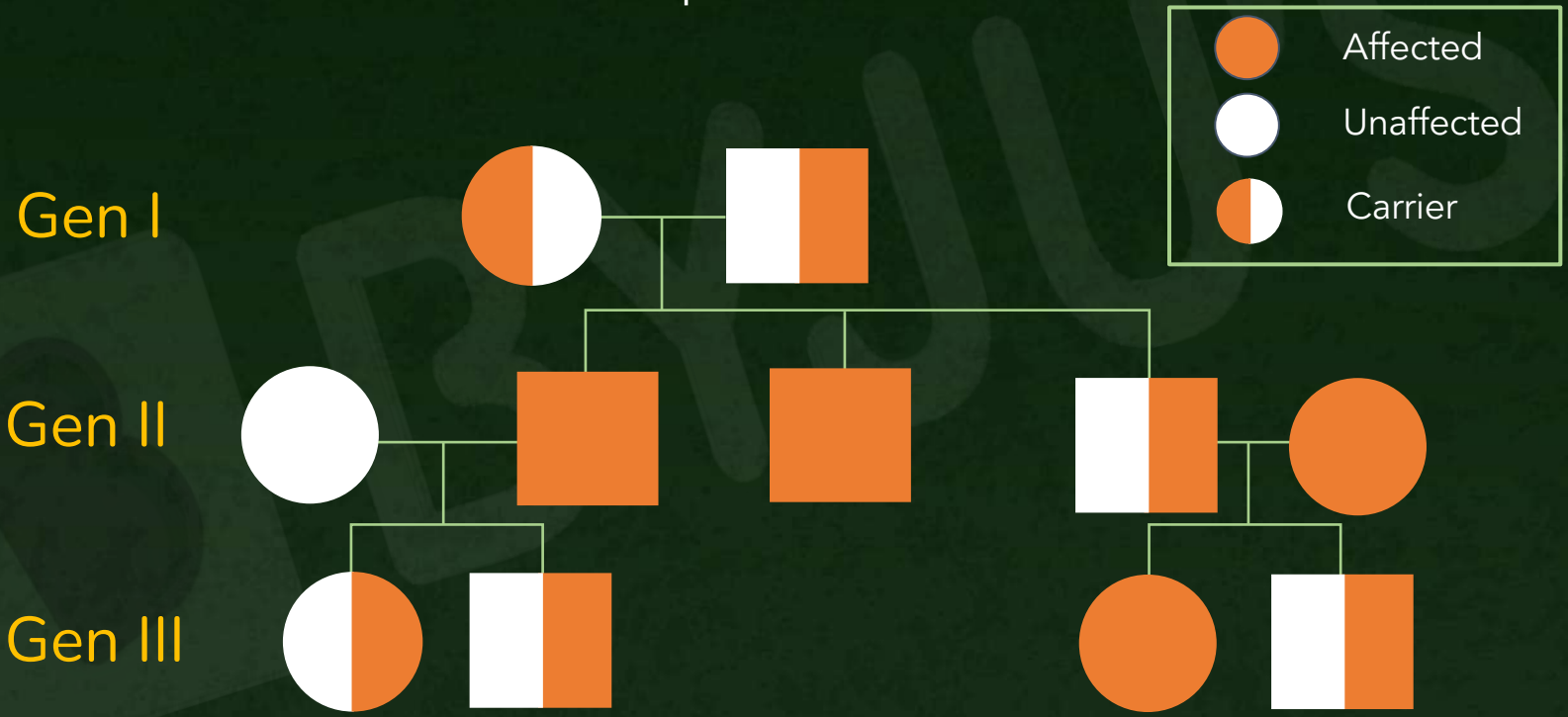
Unaffected x Affected



♀ \ ♂	p	p
P	P p	P p
P	P p	P p

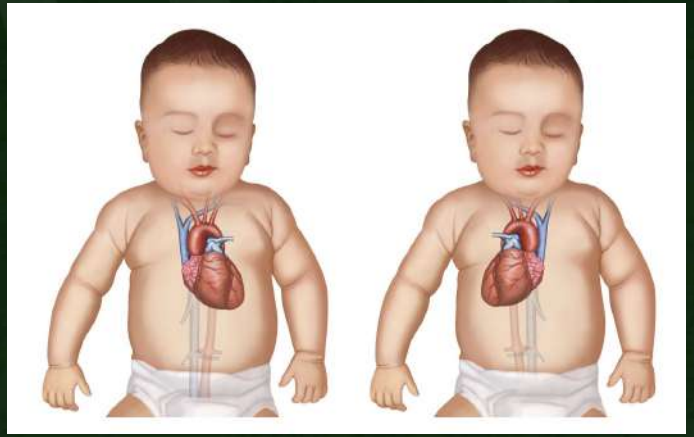
Inheritance Pattern in Phenylketonuria

If both the parents are carriers:



Kartagener's Syndrome

- An abnormal condition inherited as an autosomal recessive trait and characterized by situs inversus.
  - Situs inversus is a condition in which the organs are arranged in a mirror positions of the normal positions.
- There is lateral disposition of the viscera such as of the heart or the liver, abnormalities in the protein structure of cilia and chronic bronchiectasis and sinusitis.



Normal Dextrocardia

Kartagener’s Syndrome

- These patients lack dynein protein in all their ciliated and flagellated cells rendering these structures immotile.
- Males with disease are sterile (have immotile sperm) and are susceptible to bronchial infections (immotile respiratory cilia) and a 50 percent chance of having their heart on the right side of their body.
- These patients lack dynein protein in all their ciliated and flagellated cells rendering these structures immotile.
- Males with the disease are sterile (have immotile sperm) and are susceptible to bronchial infections (immotile respiratory cilia) and a 50 percent chance of having their heart on the right side of their body.

Cystic Fibrosis

- It is a metabolic disorder that is controlled by a single autosomal recessive gene.
- The gene specifies an enzyme that produces a unique glycoprotein responsible for production of mucus with abnormally high viscosity.
- Viscous mucus interferes with the normal functioning of several exocrine glands, including those in the skin (sweat), lungs (mucus), liver and pancreas.
- Abnormally high levels of sodium chloride occur in the sweat, and mucus stagnates in tubules of the lungs, which frequently become infected, giving rise to bronchitis.
- Secreting cells in the liver and the pancreas are damaged, decreasing production of fat-emulsifying agents and digestive enzymes and thus interfering with digestion and absorption of food.

Introduction to TH Morgan and *Drosophila*

Reasons geneticists prefer using *Drosophila* as experimental model:

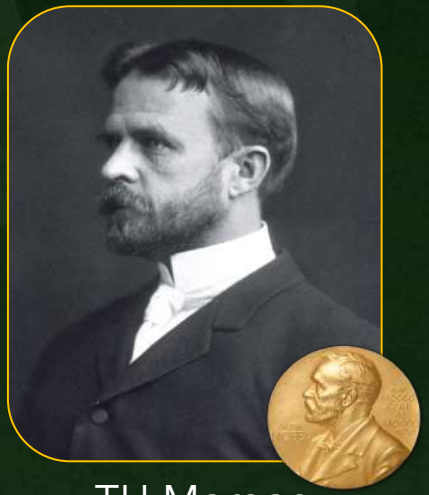
- Has only 8 chromosomes in the nucleus of each cell of its body
- Life cycle of two weeks
- Easily breeds in lab on synthetic media and requires less labour
- Produces large number of progeny on mating
- Male and female sex are different
- Exhibits variation that is visible under low microscope

Fruit fly
(*Drosophila melanogaster*)

Chromosomes of
Drosophila

Introduction to TH Morgan and *Drosophila*

- Thomas Hunt Morgan also used *Drosophila* to study inheritance patterns.
- He received Nobel Prize for explaining the role of chromosomes in heredity via experiments.
- He is also called as the father of experimental genetics.



TH Morgan

Introduction to TH Morgan and *Drosophila*

- For years, Morgan bred fruit flies on agar media and studied their genetics.
- He wanted to induce mutations and obtain a different variety of *Drosophila.*
- After some generations, he found some white eyed fruit fly in the batch, against normally observed red flies.
- He got curious and decided to experiment on the white eyed fruit flies.

Special type

Normal type

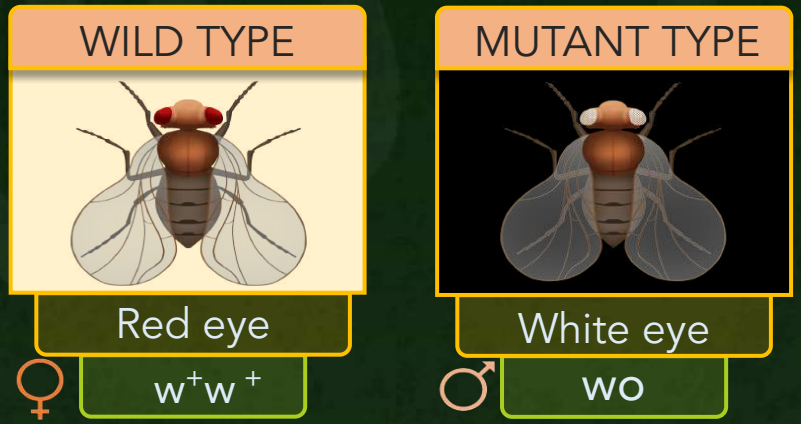
Normal type = Wild type

Special type = Mutant type

Introduction to TH Morgan and *Drosophila*

- To begin with, Morgan decided to involve white eyed *Drosophila* in a monohybrid cross.
- He took a pure type (homozygous) wild type *Drosophila*, which had a red eye.
- He then took the mutant type *Drosophila* which had white eye.
- Then he crossed these two types.

Eye colour



w : codes for white eyed fly
w^+: codes for red eyed fly

Morgan's Monohybrid Cross

Parental generation

F_1 generation

Note: "o" represents the absence of an allele since the character considered is X-linked and not present on Y chromosome.

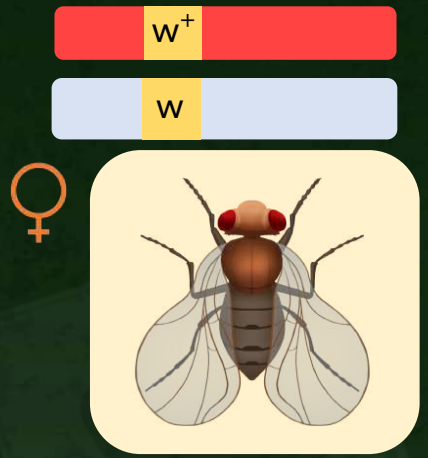
White eyed male (w o)

Red eyed female (w^+w^+)

♂ \ ♀	w^+	w^+
w	w^+w	w^+w
o	w^+o	w^+o

Morgan's Monohybrid Cross

F_1 generation



Red eyed offspring (w^+w)

When X^{w+} egg is fertilized by X^w sperm, the offspring gets w^+w as genotype and red eye phenotype.

- All the offsprings in F_1 generation were having red eyes.
- He concluded that red eye is the dominant trait.
- The phenotype results of F_1 generation were consistent with Mendel's results.
- However, all the progenies didn't have the same genotype as in Mendel's experiment.
- There were two different genotypes in 1:1 ratio.

Red eyed offspring (w^+o)

When X^{w+} egg is fertilized by y sperm which has no allele the offspring gets w^+o as genotype and red eye phenotype.

Morgan's Monohybrid Cross

- Morgan further crossed offspring obtained in F_1 generation.
- In F_2 generation, he found 3 red eyed flies and 1 white eyed fly.

F_1 generation

♂ w^+o × ♀ w^+w

F_2 generation

w^+ w^+ | w^+w | w^+o | wo

3 Red eyed flies : 1 white eyed fly

Phenotypic ratio – 3:1

Red eyed male (w^+o)

Red eyed female (w^+w)

♂ \ ♀	w^+	w
w^+	w^+w^+	w^+w
o	w^+o	wo

Morgan's Monohybrid Cross

Observations and conclusions

- Morgan obtained genotype in the ratio of 1:1:1:1.
- The genotype was different from Mendel's observation.
- However, he obtained the phenotypic ratio of 3:1, the same as Mendel's.

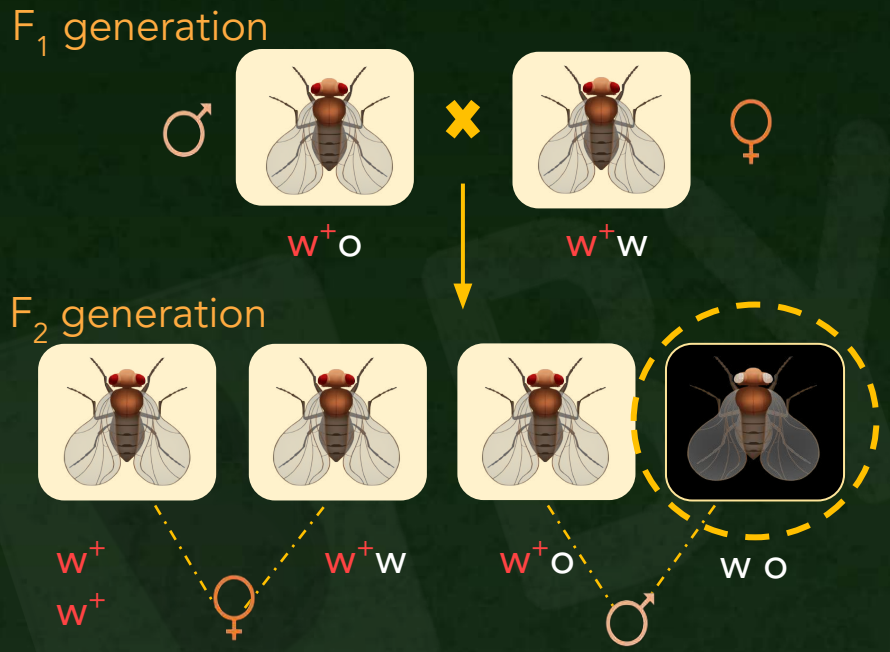
Red eyed male (w^+ o)

Red eyed female (w^+ w)

♂ \ ♀	w^+	w
w^+	w^+w^+	w^+w
o	w^+o	wo

Genotypic ratio – 1:1:1:1

Morgan's Monohybrid Cross



Observations and conclusions

- However, Morgan noticed that every time he repeated this cross, he obtained one white eyed male in F_2 generation but no white eyed female.
- This made him suspect whether the eye color was linked to sex chromosome in *Drosophila*.
- In order to confirm this hypothesis, Morgan crossed heterozygous red-eyed females ($X^{w+}X^{w}$) with white-eyed males ($X^{w}Y$).

Morgan's Monohybrid Cross

Observations and conclusions

- This cross gave a phenotypic ratio of 1:1:1:1 for male white eyed : female white eyed : male red eyed : female red eyed.
- Morgan's hypothesis was proved by this cross since the white eye trait finally appeared in female.
- Morgan concluded that the X-chromosome carried a number of factors.
- For female *Drosophila* to have white eye, each of the eye colour gene has to be ww.
- Only then a female can have white eye. Hence it is a very rare phenomena.

White eyed male (wo)

Red eyed female (w^+w)

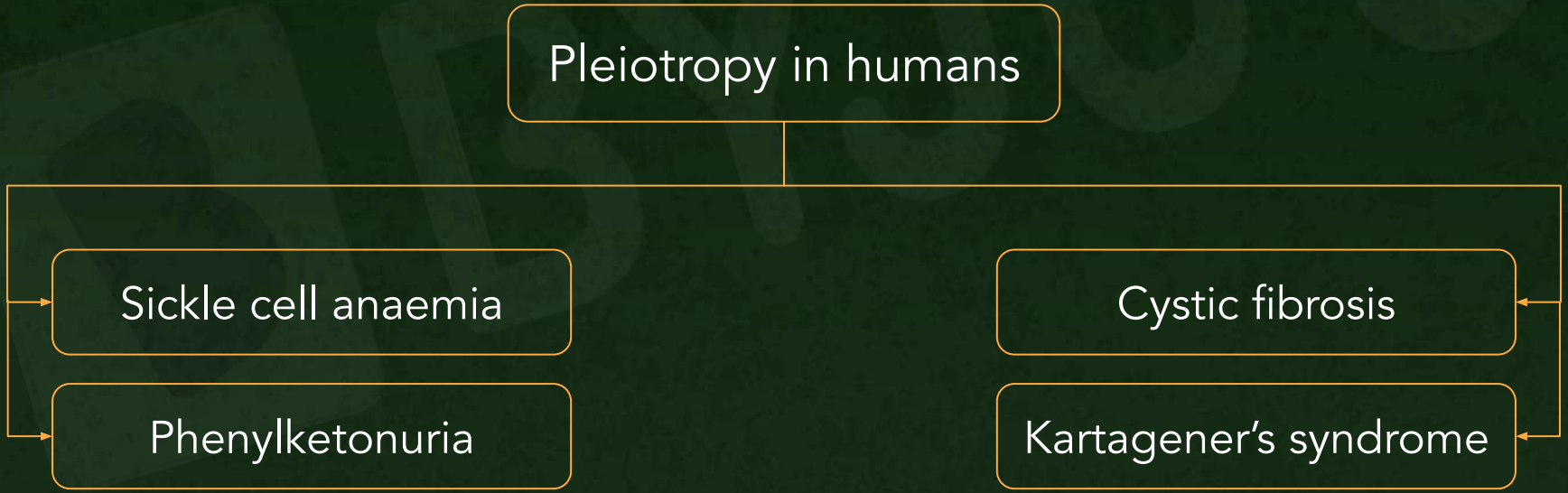
♂ \ ♀	w^+	w
w	w^+w	ww
o	w^+o	wo

Genotypic ratio – 1:1:1:1

Summary

Pleiotropism

It is a phenomena where a single gene shows multiple phenotypic expressions.



Summary

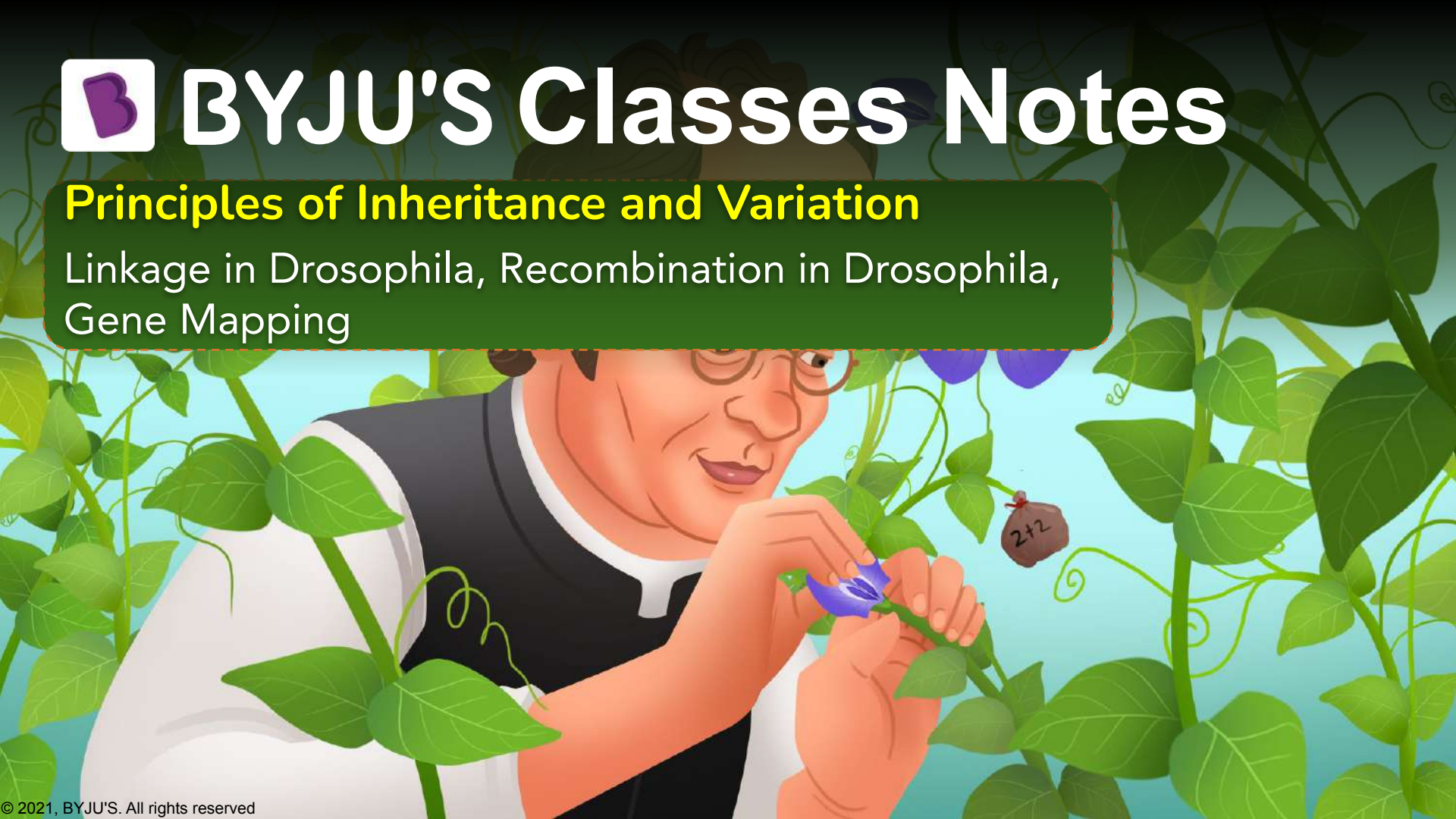
Morgan's monohybrid cross

- Morgan noticed that in his monohybrid cross, he obtained one white eyed male in F_2 generation but no white eyed female.
- This made him suspect whether the eye color was linked to sex chromosome in *Drosophila*.
- In order to confirm this hypothesis, Morgan crossed heterozygous red-eyed females (Xw+Xw) with white-eyed males (XwY).
- This cross gave a phenotypic ratio of 1:1:1:1 for male white eyed : female white eyed : male red eyed : female red eyed.
- Morgan concluded that the X-chromosome carried a number of factors.

BYJU'S Classes Notes

Principles of Inheritance and Variation

Linkage in Drosophila, Recombination in Drosophila, Gene Mapping



Key Takeaways

1. Introduction to TH Morgan and *Drosophila*
2. Morgan’s monohybrid cross
3. Morgan’s dihybrid cross A
4. Morgan’s dihybrid cross B

Summary

Introduction to TH Morgan and *Drosophila*

Reasons geneticists prefer using *Drosophila* as experimental model:

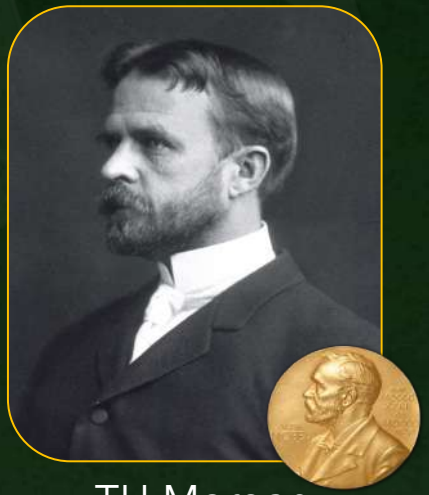
- Has only 8 chromosomes in the nucleus of each cell of its body
- Life cycle of two weeks
- Easily breeds in lab on synthetic media and requires less labour
- Produces large number of progeny on mating
- Male and female sex are different
- Exhibits variation that is visible under low microscope

Fruit fly
(*Drosophila melanogaster*)

Chromosomes of
Drosophila

Introduction to TH Morgan and *Drosophila*

- Thomas Hunt Morgan also used *Drosophila* to study inheritance patterns.
- He received Nobel Prize for explaining the role of chromosomes in heredity via experiments.
- He is also called as the father of experimental genetics.



TH Morgan

Introduction to TH Morgan and *Drosophila*

- For years, Morgan bred fruit flies on agar media and studied their genetics.
- He wanted to induce mutations and obtain a different variety of *Drosophila.*
- After some generations, he found some white eyed fruit fly in the batch, against normally observed red flies.
- He got curious and decided to experiment on the white eyed fruit flies.

Special type

Normal type

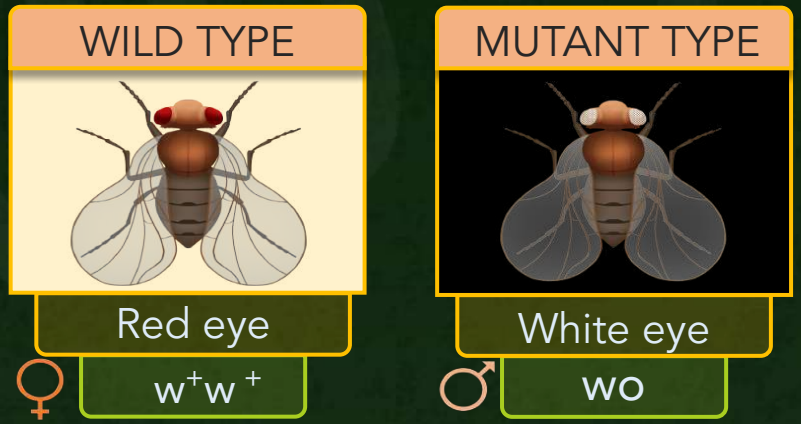
Normal type = Wild type

Special type = Mutant type

Introduction to TH Morgan and *Drosophila*

- To begin with, Morgan decided to involve white eyed *Drosophila* in a monohybrid cross.
- He took a pure type (homozygous) wild type *Drosophila*, which had a red eye.
- He then took the mutant type *Drosophila* which had white eye.
- Then he crossed these two types.

Eye colour



w : codes for white eyed fly
w^+: codes for red eyed fly

Morgan's Monohybrid Cross

Parental generation

♂  ×  ♀

w o

w^+w^+

F_1 generation

w^+w w^+w w^+o w^+o

♀ ♂

Note: "o" represents the absence of an allele since the character considered is X-linked and not present on Y chromosome.

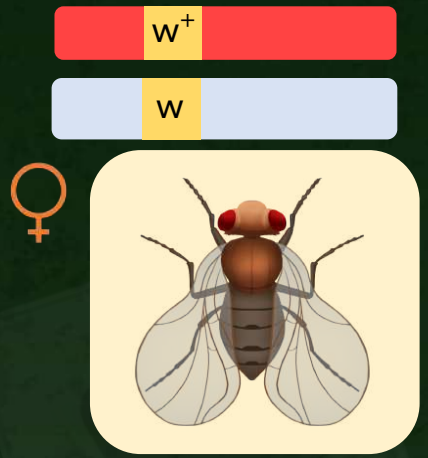
White eyed male (w o)

Red eyed female (w^+w^+)

♂ \ ♀	w^+	w^+
w	w^+w	w^+w
o	w^+o	w^+o

Morgan's Monohybrid Cross

F_1 generation



Red eyed offspring (w^+w)

When X^{w+} egg is fertilized by X^w sperm, the offspring gets w^+w as genotype and red eye phenotype.

- All the offsprings in F_1 generation were having red eyes.
- He concluded that red eye is the dominant trait.
- The phenotype results of F_1 generation were consistent with Mendel's results.
- However, all the progenies didn't have the same genotype as in Mendel's experiment.
- There were two different genotypes in 1:1 ratio.

Red eyed offspring (w^+o)

When X^{w+} egg is fertilized by y sperm which has no allele the offspring gets w^+o as genotype and red eye phenotype.

Morgan's Monohybrid Cross

- Morgan further crossed offspring obtained in F_1 generation.
- In F_2 generation, he found 3 red eyed flies and 1 white eyed fly.

F_1 generation

♂ w^+o × ♀ w^+w

F_2 generation

w^+ w^+ | w^+w | w^+o | wo

3 Red eyed flies : 1 white eyed fly

Phenotypic ratio – 3:1

Red eyed male (w^+o)

Red eyed female (w^+w)

♂ \ ♀	w^+	w
w^+	w^+w^+	w^+w
o	w^+o	wo

Morgan's Monohybrid Cross

Observations and conclusions

- Morgan obtained genotype in the ratio of 1:1:1:1.
- The genotype was different from Mendel's observation.
- However, he obtained the phenotypic ratio of 3:1, the same as Mendel's.

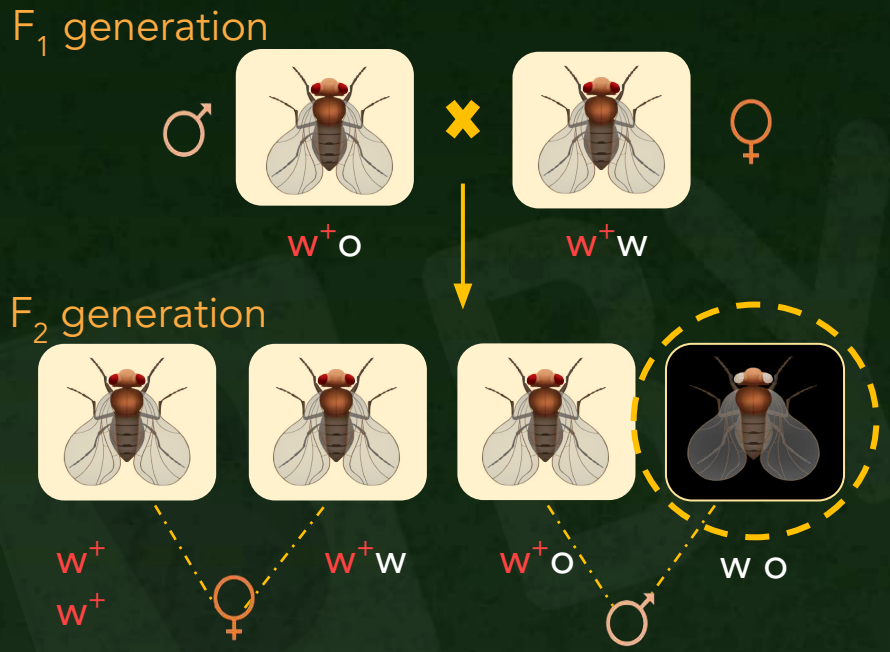
Red eyed male (w^+ o)

Red eyed female (w^+ w)

♂ \ ♀	w^+	w
w^+	w^+w^+	w^+w
o	w^+o	wo

Genotypic ratio – 1:1:1:1

Morgan's Monohybrid Cross



Observations and conclusions

- However, Morgan noticed that every time he repeated this cross, he obtained one white eyed male in F_2 generation but no white eyed female.
- This made him suspect whether the eye color was linked to sex chromosome in *Drosophila*.
- In order to confirm this hypothesis, Morgan crossed heterozygous red-eyed females ($X^{w+}X^{w}$) with white-eyed males ($X^{w}Y$).

Morgan's Monohybrid Cross

Observations and conclusions

- This cross gave a phenotypic ratio of 1:1:1:1 for male white eyed : female white eyed : male red eyed : female red eyed.
- Morgan's hypothesis was proved by this cross since the white eye trait finally appeared in female.
- Morgan concluded that the X-chromosome carried a number of factors.
- For female *Drosophila* to have white eye, each of the eye colour gene has to be ww.
- Only then a female can have white eye. Hence it is a very rare phenomena.

White eyed male (wo)

Red eyed female (w^+w)

♂ \ ♀	w^+	w
w	w^+w	ww
o	w^+o	wo

Genotypic ratio – 1:1:1:1

Idea of Linkage

- Besides Morgan, several other scientists such as Punnett and colleagues were trying to carry forward Mendel's work.
- Punnett and colleagues in their experiments on the sweet pea, noticed that the dihybrid cross phenotypic ratio in F_2 generation deviated majorly from the Mendelian ratio of 9:3:3:1.
- They found that the parental combination of traits tended to appear in higher ratio in F_2 generation and non-parental combination of traits were rare.
- These results along with Morgan's own discovery of linkage of certain genes with sex chromosomes led him to believe that various genes existed on single chromosomes and such genes, which existed on same chromosomes, failed to assort independently.
- Hence, he hypothesized the probability of linkage between genes.

Morgan's Dihybrid Cross A

- Morgan then went on to perform dihybrid cross in the *Drosophila*.
- He crossed a pure breed *Drosophila* with dominant traits with a pure breed *Drosophila* with recessive traits.
- He took characters that he knew were present on same chromosome and hence would show linkage.
- He considered the following two characters:

Character 1 - Body colour

MUTANT TYPE	WILD TYPE
	
Yellow body	Brown body
yy	y^+y^+

Character 2 - Eye colour

MUTANT TYPE	WILD TYPE
	
White eye	Red eye
ww	w^+w^+

Morgan's Dihybrid Cross A

Homozygous recessive parent

♀

Yellow bodied, white eyed

y w

y w

Homozygous dominant parent

♂

Brown bodied, red eyed

y^+ w^+

Male (y^+w^+)

Female (yyww)

♂ \ ♀	yw	yw
y^+w^+	yy^+ww^+ Brown bodied Red eyed	yy^+ww^+ Brown bodied Red eyed
oo	yowo Yellow bodied White eyed	yowo Yellow bodied White eyed

Morgan's Dihybrid Cross A

Note:

- In the above Punnett square, male *Drosophila* gives rise to two gametes, one containing X chromosome and the other containing the Y chromosome.
- However, both the characters in this dihybrid cross are X-linked, that is they are only found on X chromosome.
- Hence, the gamete with Y chromosome doesn't contribute any allele for the two characters.

Morgan's Dihybrid Cross A

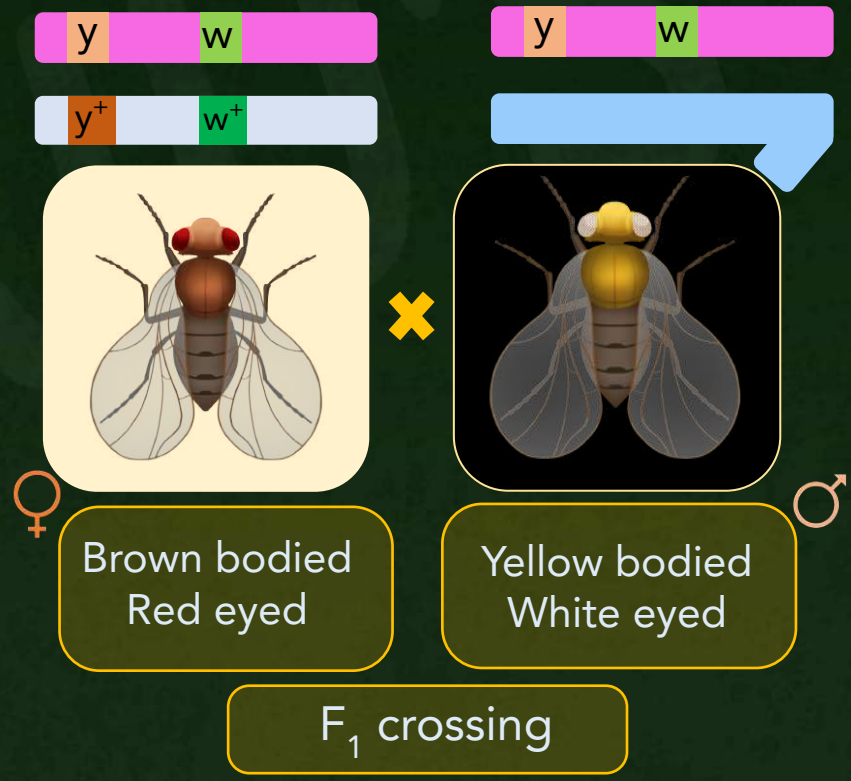
Observations from the results obtained in F_1 generation :

- The results obtained were inconsistent with the results obtained in Mendel's experiments.
- The F_1 generation not only showed the dominant phenotype (as in Mendel's experiments) but also the recessive phenotype.
- This was due to the fact that the characteristics considered are X-linked and the Y chromosome contributed no alleles.
- Hence, expression of recessive phenotype occurred from only one allele obtained from the female parent.

Morgan's Dihybrid Cross A

F_1 generation:

- Morgan then crossed the two different type of progenies obtained in the F_1 generation.
- However, this time, female was the dominant parent while male was the recessive parent.
- The dominant parent was heterozygous and not homozygous.



Morgan's Dihybrid Cross A

F_1 generation gametes:

Since, the gametes are on same chromosomes and hence linked, only these 4 gametes are possible.

Gametes

Morgan's Dihybrid Cross A

F_2 generation (Ideal hypothetical outcome):

- This is the ideal cross outcome that would result if the genes were linked.
- Phenotypic ratio would be 1:1 for Brown bodied red eyed : Yellow bodied white eyes.
- However, this did not happen.

Female (yy^+ww^+)

Male (yowo)

♂ \ ♀	y^+w^+	yw
yw	yy^+ww^+ Brown bodied Red eyed	yyww Yellow bodied White eyed
oo	y^+ow^+o Brown bodied Red eyed	yowo Yellow bodied White eyed

Morgan's Dihybrid Cross A

F_2 generation (Actual outcome):

Yellow bodied White eyed

Brown bodied Red eyed

Parental progenies

Brown bodied White eyed

Yellow bodied Red eyed

Non-parental progenies

- Morgan saw the appearance of non-parental phenotypes and obtained 4 different types of phenotypes as Mendel did in his dihybrid cross experiments.
- Despite the unexpected results, Morgan was convinced that linkage was there because the percentage of parental type progenies was very high compared to non-parental type progenies.
- Individuals with both the recessive traits appeared in equal numbers to individuals with both the dominant traits, in stark contrast to Mendel's F_2 dihybrid ratio.

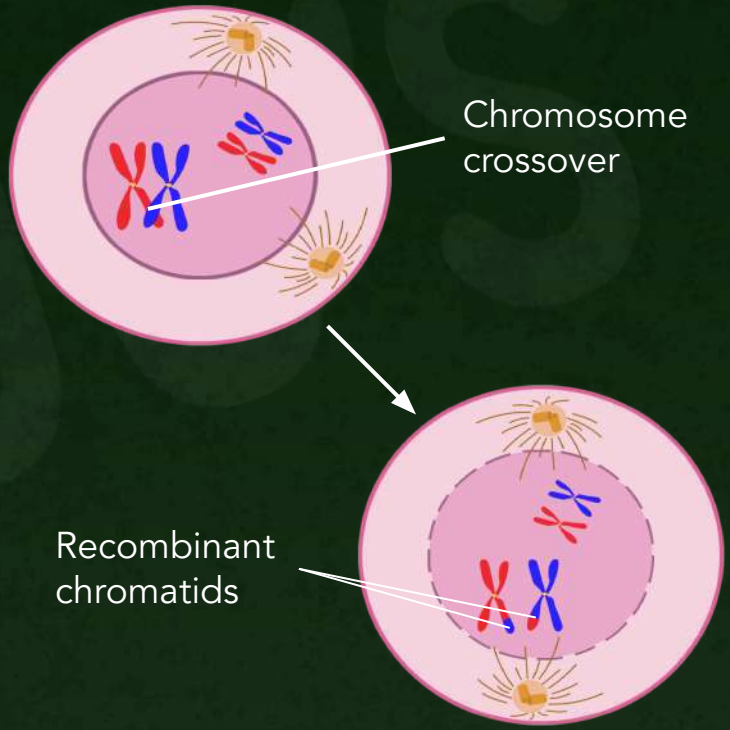
Morgan's Conclusions

Based on all the above observations, Morgan concluded that:

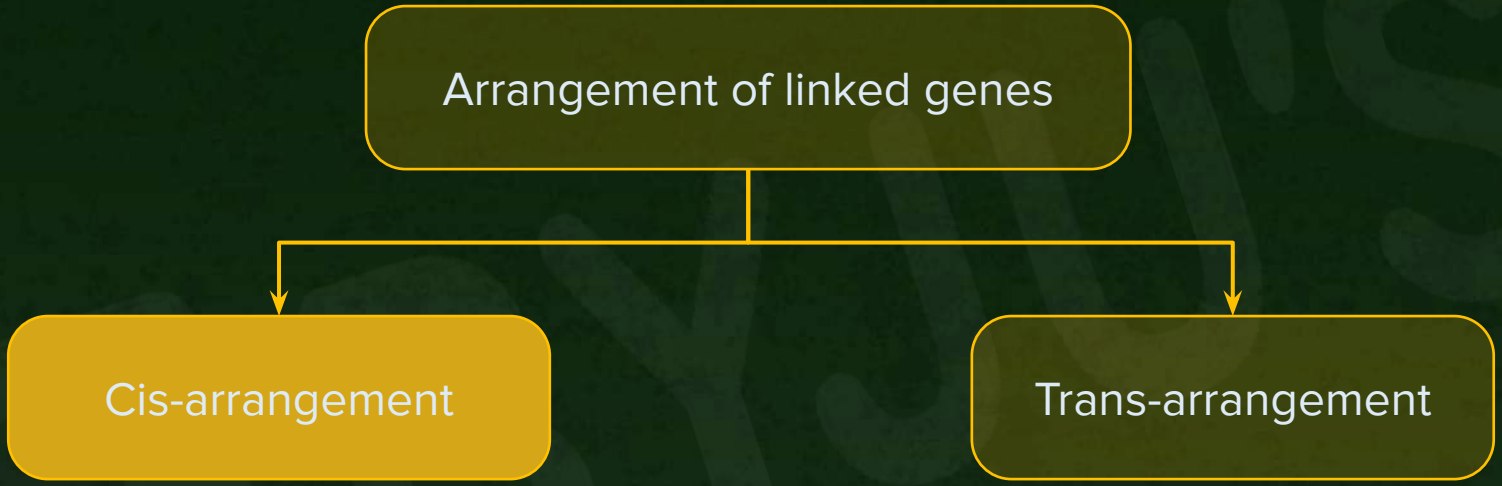
- Higher percentage of individuals with both the recessive traits in F_2 generation was due to the fact they were the parental type.
- Higher percentage of individuals with parental type trait combination was due to linkage between the two genes.
- The presence of non-parental type individuals was due to a phenomenon called recombination.

Linkage

- Linkage is the association of two genes located on the same chromosome.
- Linkage describes the probability of the two genes being inherited together.
- Since genes of both characters that Morgan chose were on the same X chromosome, thus they were inherited together as chromosomes are passed on as single units from one generation to the other.
- Yet, there is a phenomenon of crossing over, first described by Sutton and Boveri, that occurs in prophase, which gave Morgan the clue for presence of the phenomenon of recombination.



Arrangement of Linked Genes



- In this arrangement, dominant alleles of both the linked genes are present on one chromosome and their recessive alleles on its homologous chromosome (AB/ab).
- This arrangement is known as cis-arrangement and genes are said to be in coupling state.

Arrangement of Linked Genes

Arrangement of linked genes

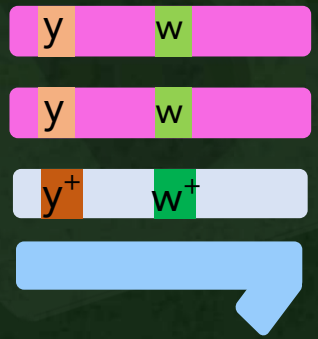
Cis-arrangement

Trans-arrangement

- In this arrangement, dominant allele of one pair and recessive allele of the second pair are present on one chromosome and recessive and dominant alleles respectively on the other homologous chromosome (Ab/aB).
- This arrangement is known as trans-arrangement and genes are said to be in repulsive state.

Recombination

- To explain the occurrence of non-parental type of progenies even in case of a dihybrid cross where the characters considered were on same chromosomes and hence linked, Morgan introduced the concept of recombination.
- Recombination is the phenomenon in which a part of the chromosome is exchanged or crossed over between two homologous chromosomes during prophase.
- Crossing over leads to new alleles or new allelic combination in offspring.
- Hence, even though Morgan expected only 4 different type of gametes from F_1 generation, actually 6 different type of gametes appeared due to recombination.

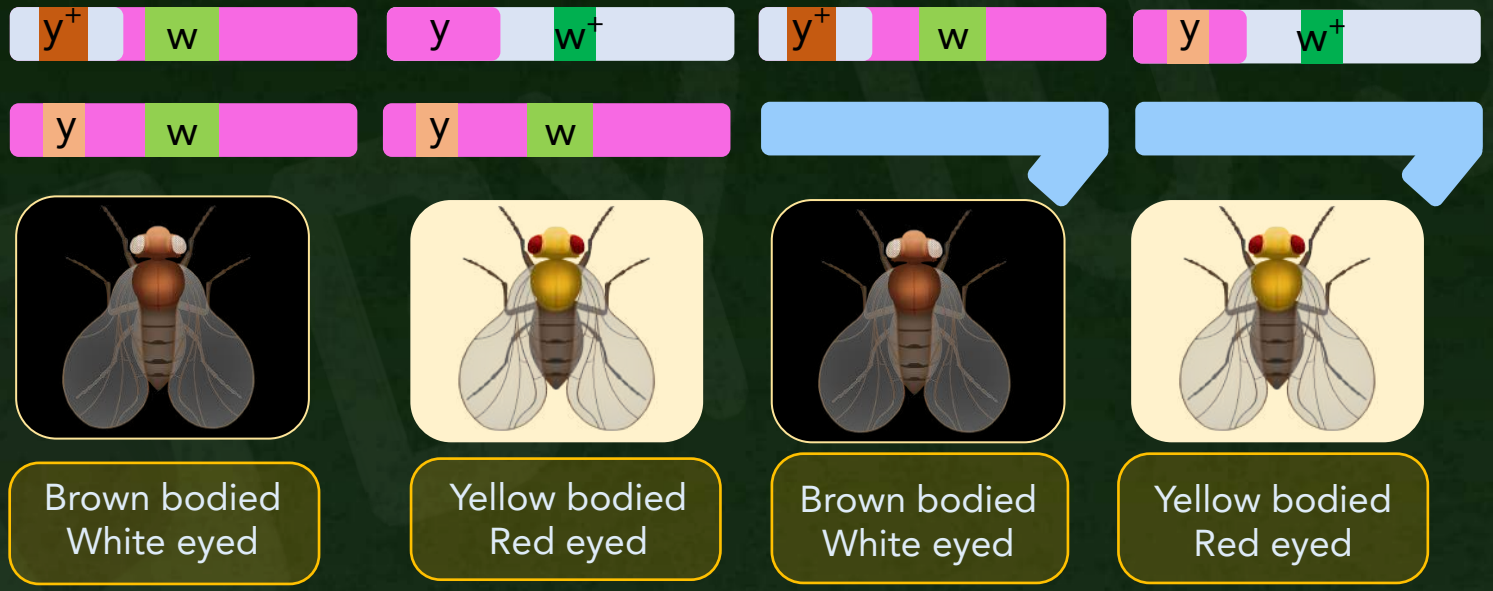


Expected gametes

Gametes that appeared due to recombination

Morgan's Dihybrid Cross A

Non-parental *Drosophila* genotype



Thus, it was because of recombination that non-parental varieties of *Drosophila* were obtained.

Morgan's Dihybrid Cross A

Morgan took his research forward and tried to find out the percentage of parental type offspring and non-parental type of offspring found in the given dihybrid cross

Yellow bodied
White eyed

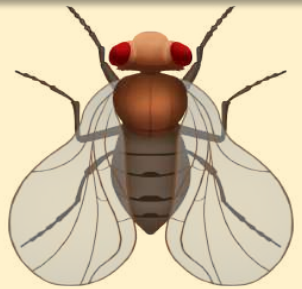
Brown bodied
Red eyed

F_2 generation	Cross A
Parental type	98.7%
Non-Parental type	1.3%

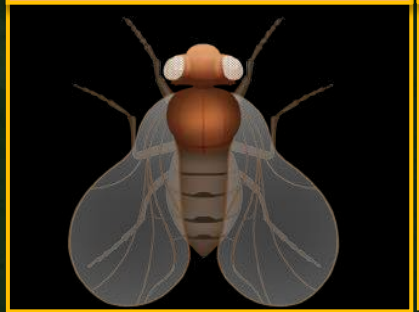
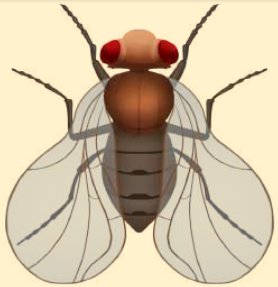
Morgan's Dihybrid Cross B

- Morgan wanted to check if similar ratio of parental and non-parental type would be observed if he changed to a different pair of characteristics, essentially studying a different set of genes simultaneously.
- He took the following two characters for this second dihybrid cross.

Character 1 – Wing size

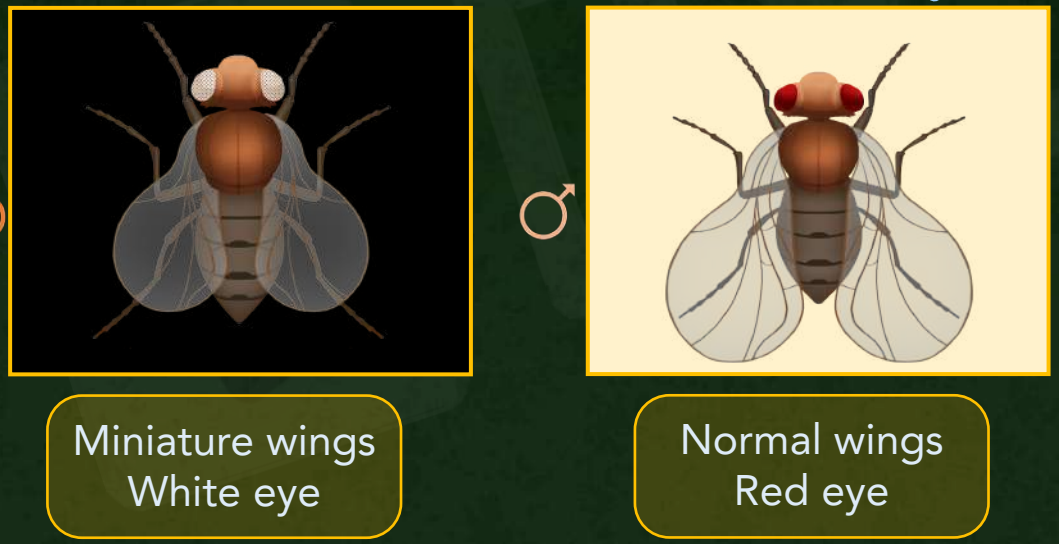
MUTANT TYPE	WILD TYPE
	
Miniature wings	Normal wings
mm	m^+m^+

Character 2 -Eye colour

MUTANT TYPE	WILD TYPE
	
White eye	Red eye
ww	w^+w^+

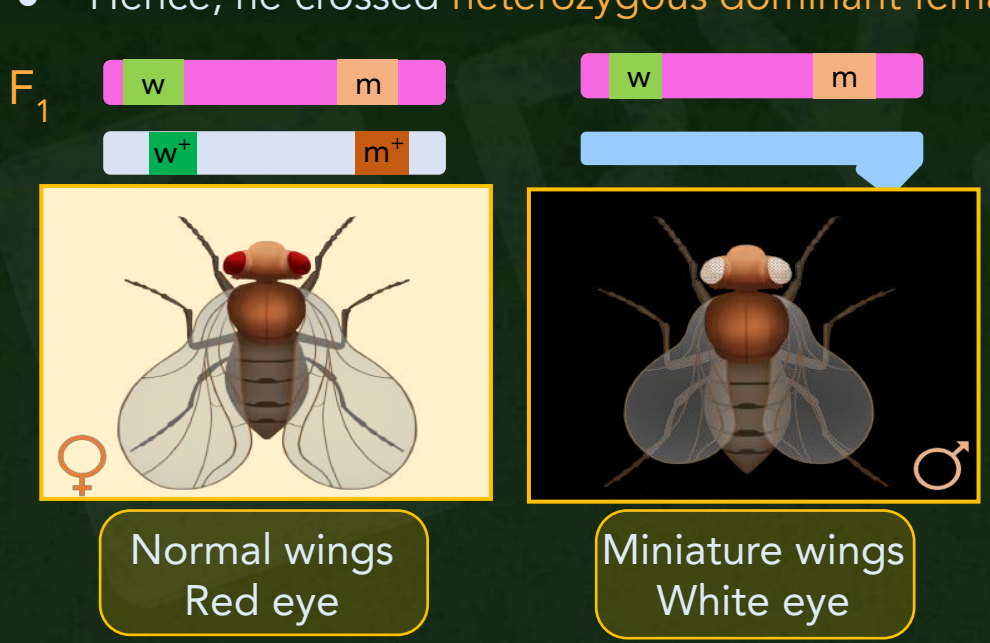
Morgan's Dihybrid Cross B

- Morgan hence crossed homozygous dominant normal wing red eyed male with homozygous recessive miniature wing white eyed female.



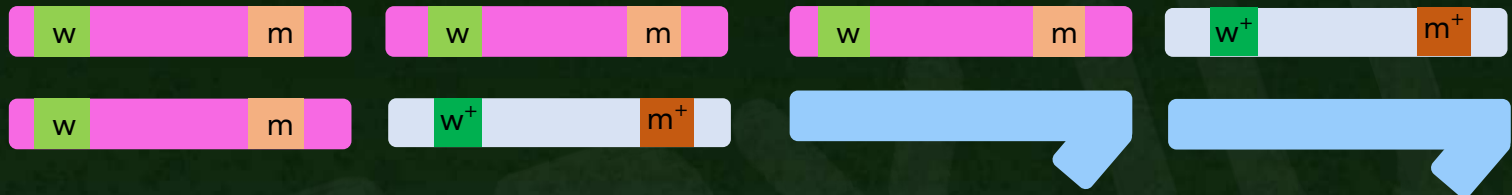
Morgan's Dihybrid Cross B

- Just as in dihybrid cross A, Morgan got the parental type phenotypes in F_1 generation in 1:1 ratio. However, the parent with dominant phenotype was homozygous and not heterozygous.
- In F_1, even though the parental phenotypes were obtained, the individuals were heterozygous.
- Hence, he crossed heterozygous dominant female with recessive male.

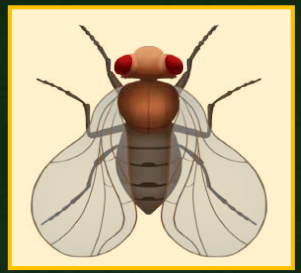


Morgan's Dihybrid Cross B

F_2 generation (Parental type)



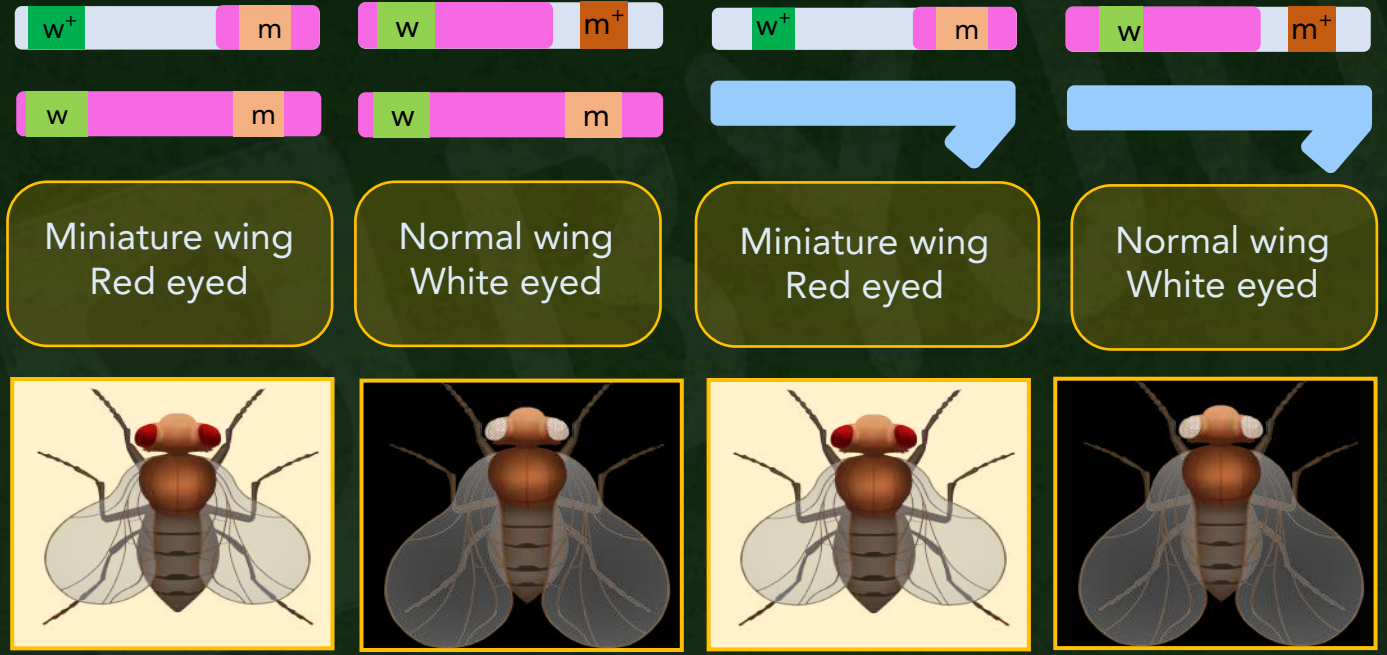
Miniature wings White eye	Normal wings Red eye	Miniature wings White eye	Normal wings Red eye



As expected, he obtained the two parental type phenotypes formed by 4 different genotypes.

Morgan's Dihybrid Cross B

Non-parental *Drosophila* phenotype



Further, he also obtained two non-parental phenotypes formed by 4 different genotypes involving recombinant chromosomes.

Morgan's Dihybrid Cross B

- However, he noticed a massive difference in the percentages of parental and the non-parental phenotypes.
- Non-parental phenotypes were much higher in number in this dihybrid Cross B.
- This result suggested that the linkage between the genes of the two characters considered in this cross was much lower.

F_2 generation	Wing size and eye colour (cross B)
Parental type	62.8%
Non-parental type	37.2%

Linkage and Recombination

Hence, Morgan arrived on the following conclusion:

$$\text{Recombination} \propto \frac{1}{\text{Linkage}}$$

- Recombination is inversely proportional to linkage
- Tighter the linkage - Lesser the chance of recombination
- Lesser the linkage - Higher the chance of recombination

Gene Mapping

- The tool for locating and relatively mapping genes on the chromosomes.
- Gene mapping is also called linkage mapping.
- Gene mapping involves graphical representation of the linear sequence of genes and their relative distances on a chromosome.

Terms associated with gene mapping:

- Unit to measure distance between genes is centiMorgan (cM) or map unit (m.u).
- One map unit corresponds to minimum physical length of chromosome which is required for crossing over.
- Frequency of crossing over is proportional to the distance between genes.

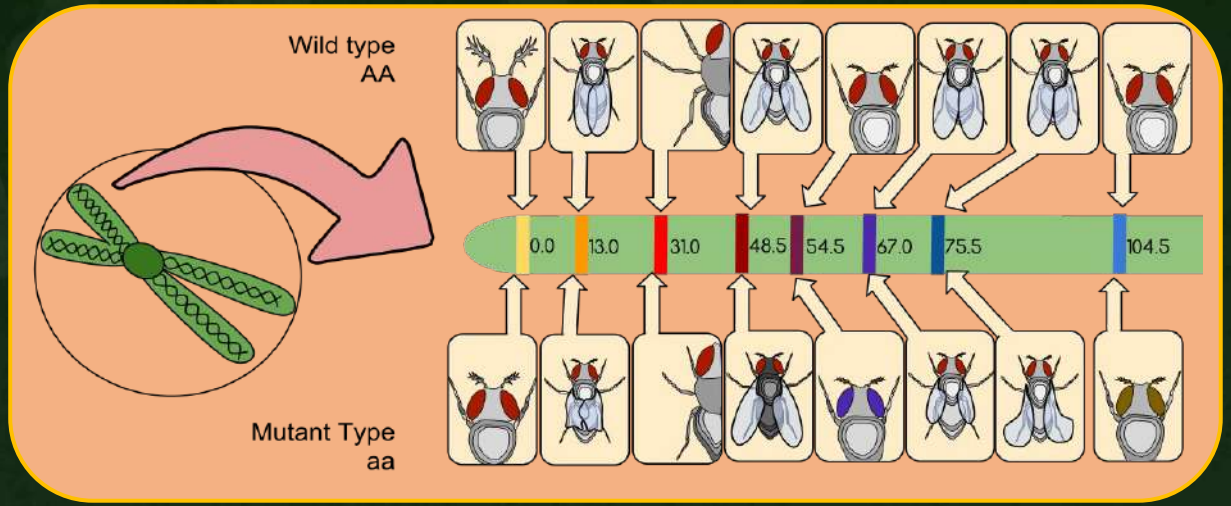
Gene Mapping

- **1 cM = 1% Recombination frequency**
- **% Recombination -** relative distance between genes

$$\text{Recombination Frequency (RF)} = \frac{\text{Recombinants X 100}}{\text{Total number of offspring}}$$

Using all these aspects and calculations, a gene map is constructed.

Representation of gene mapping of a chromosome of *Drosophila*.



Summary

Morgan's monohybrid cross

- Morgan noticed that in his monohybrid cross, he obtained one white eyed male in F_2 generation but no white eyed female.
- This made him suspect whether the eye color was linked to sex chromosome in *Drosophila*.
- In order to confirm this hypothesis, Morgan crossed heterozygous red-eyed females (Xw+Xw) with white-eyed males (XwY).
- This cross gave a phenotypic ratio of 1:1:1:1 for male white eyed : female white eyed : male red eyed : female red eyed.
- Morgan concluded that the X-chromosome carried a number of factors.

Summary

- Morgan further conducted two different types of dihybrid crosses:
    - First one between red eyed, brown bodied *Drosophila* and white eyed, yellow bodied *Drosophila*.
    - Second one between normal wings, red eyed and miniature wing, white eyed *Drosophila*.
    - He discovered that the percentage of parental type individuals was very high (possibly due to linkage) which was inconsistent with Mendel's results.
    - Also, non-parental type individuals appeared despite the genes being present on the same chromosome (possible due to recombination).
    - Consequently, he came up with the theories of linkage and recombination.

Summary

Linkage

- Linkage is the association of two genes located on the same chromosome.
- Linkage describes the probability of the two genes being inherited together.

Recombination

- Recombination is the phenomenon in which a part of the chromosome is exchanged or crossed over between two homologous chromosomes during prophase.
- Crossing over leads to new alleles or new allelic combination in offspring.

Summary

$$\text{Recombination} \propto \frac{1}{\text{Linkage}}$$

- Gene mapping involves graphical representation of the linear sequence of genes and their relative distances on a chromosome.

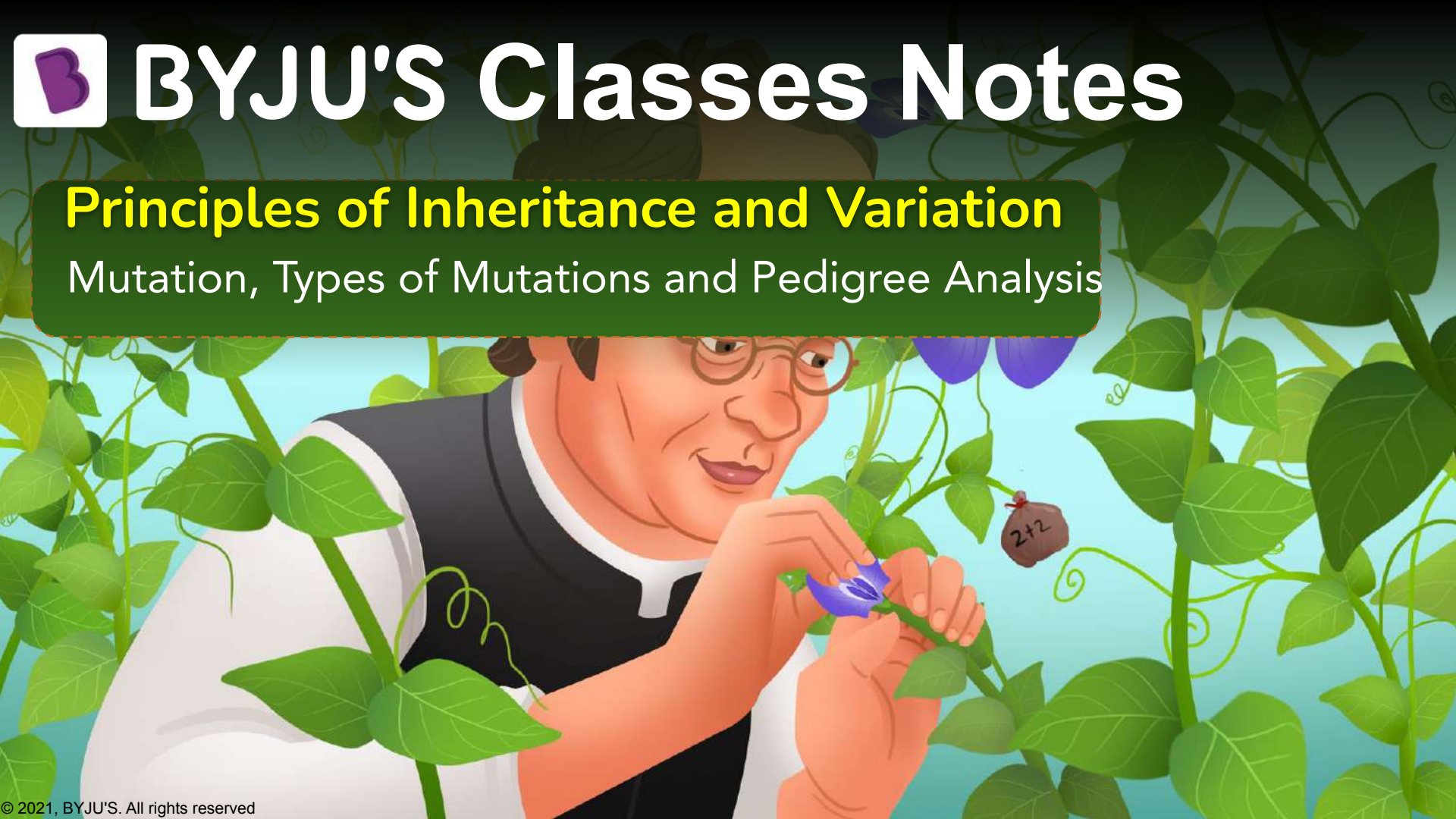
- 1 cM = 1% Recombination frequency
- % Recombination - relative distance between genes

$$\text{Recombination Frequency (RF)} = \frac{\text{Recombinants X 100}}{\text{Total number of offspring}}$$

BYJU'S Classes Notes

Principles of Inheritance and Variation

Mutation, Types of Mutations and Pedigree Analysis



Key Takeaways

1. Gene Mapping
2. Mutation
3. Pedigree analysis

Summary

Gene Mapping

- The tool for locating and relatively mapping genes on the chromosomes.
- Gene mapping is also called linkage mapping.
- Gene mapping involves graphical representation of the linear sequence of genes and their relative distances on a chromosome.

Terms associated with gene mapping:

- Unit to measure distance between genes is centiMorgan (cM) or map unit (m.u).
- One map unit corresponds to minimum physical length of chromosome which is required for crossing over.
- Frequency of crossing over is proportional to the distance between genes.

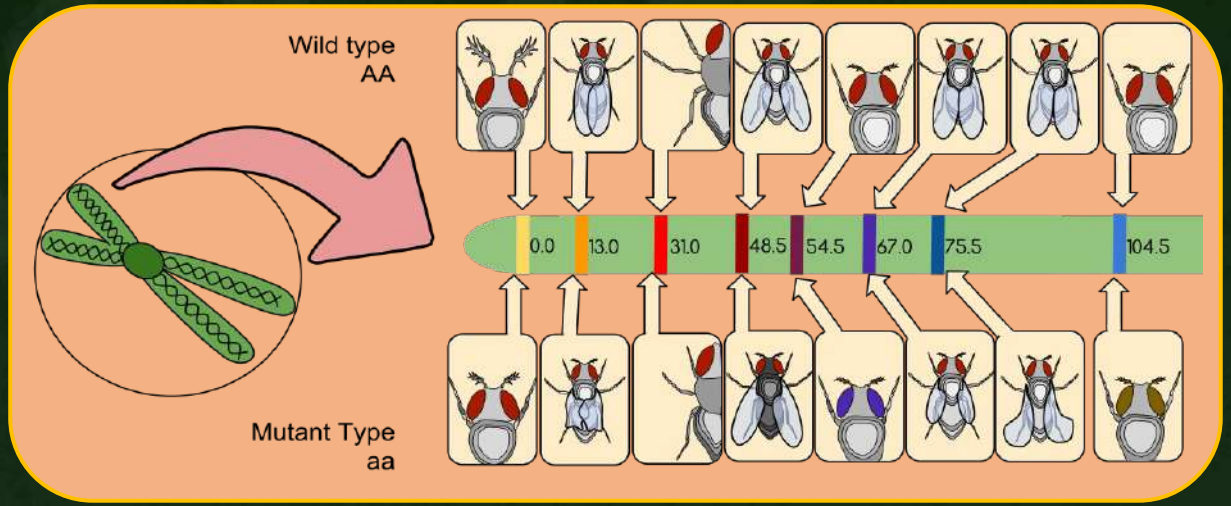
Gene Mapping

- **1 cM = 1% Recombination frequency**
- **% Recombination -** relative distance between genes

$$\text{Recombination Frequency (RF)} = \frac{\text{Recombinants X 100}}{\text{Total number of offspring}}$$

Using all these aspects and calculations, a gene map is constructed.

Representation of gene mapping of a chromosome of *Drosophila*.



Recall! Gene

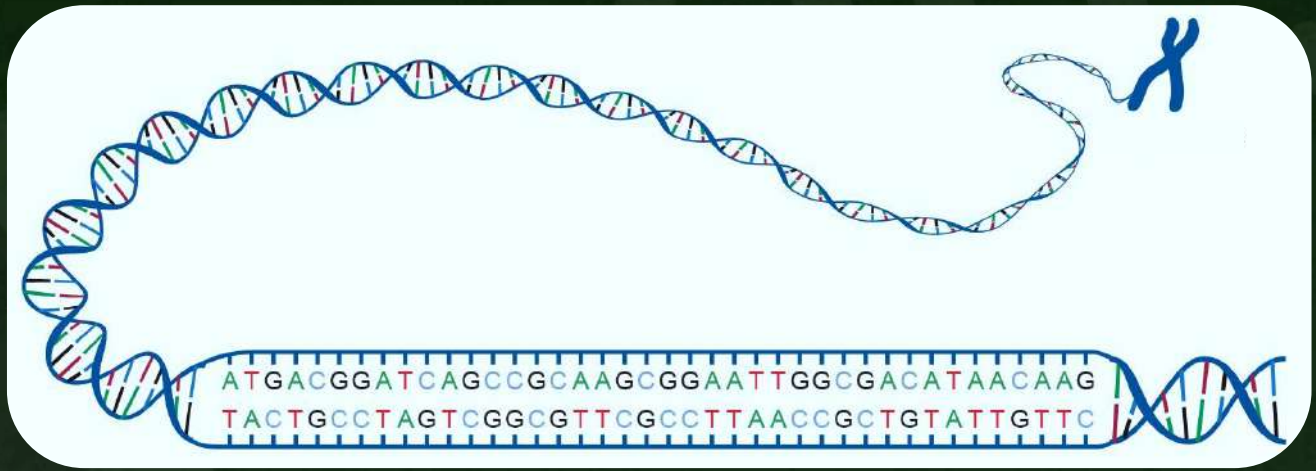
Coding gene

Protein

- Genes code for specific proteins, which have specific functions giving rise to specific characteristics.

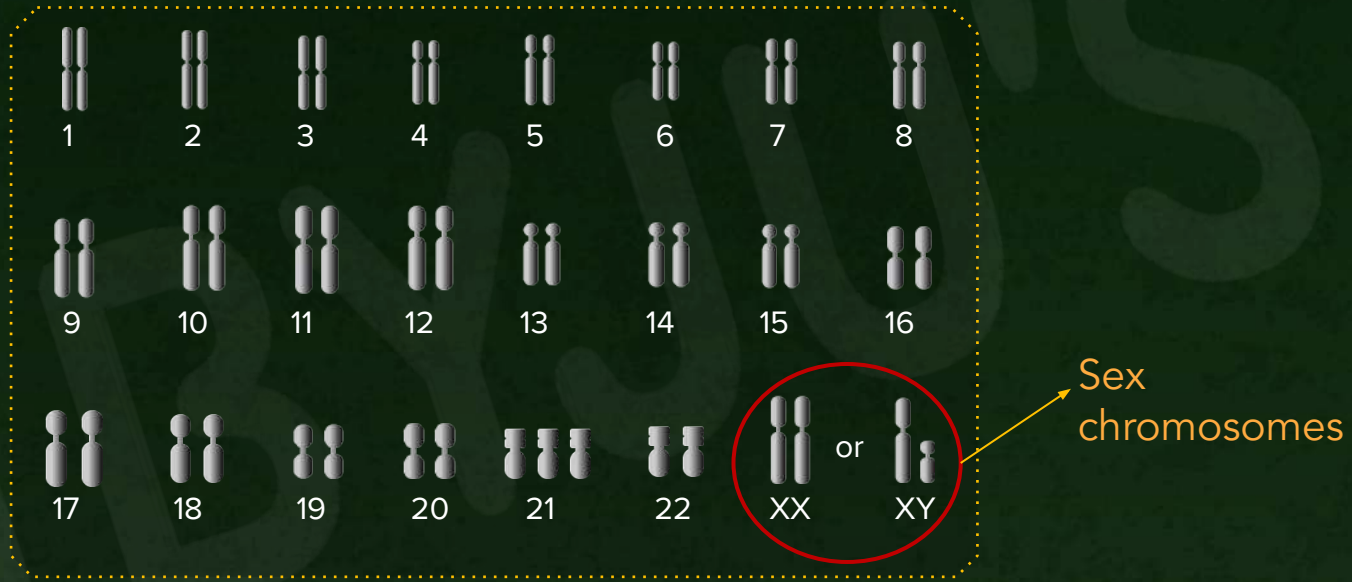
Recall! DNA - Sequence of Nucleotides

DNA (Deoxyribonucleic acid)



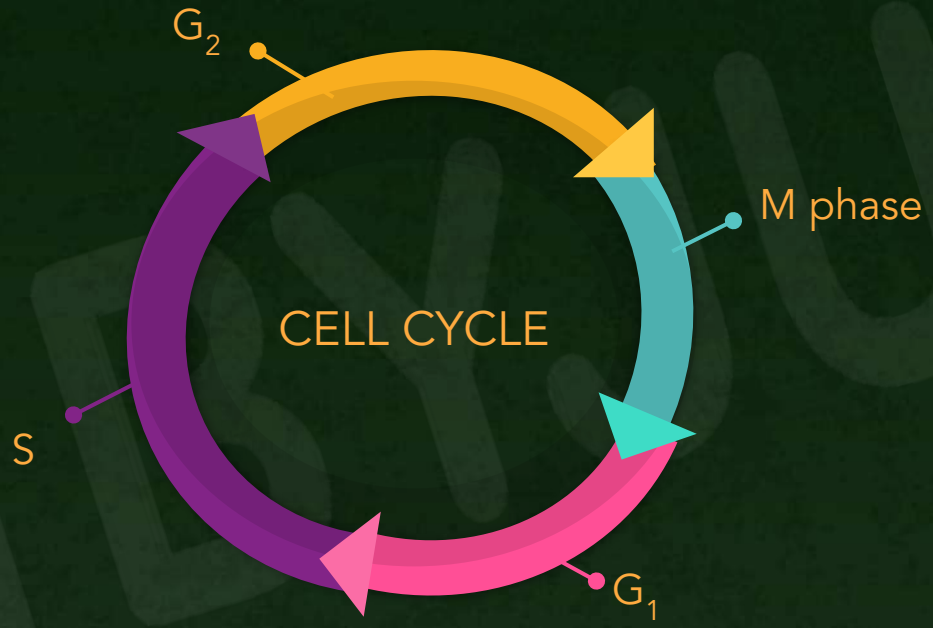
- DNA contains base pairs which carry genetic information

Recall! Human Karyotype



- Out of the 23 pairs of genes in humans, 22 are autosomal chromosomes, and X and Y are sex chromosomes.
- Genes that are inherited via the sex chromosomes are called sex-linked, and the inheritance is called sex-linked inheritance.

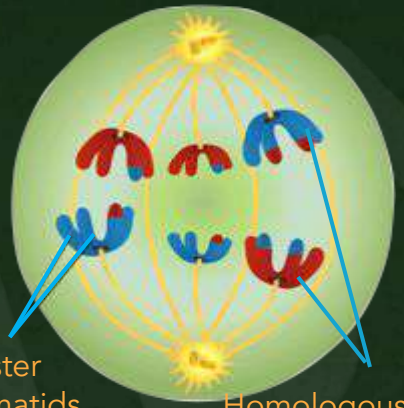
Recall! S-phase of Cell Cycle



- DNA synthesis and duplication takes place during S phase.

Recall! Anaphase of Meiosis

Anaphase I

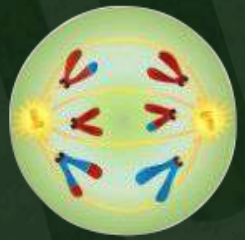


Sister chromatids

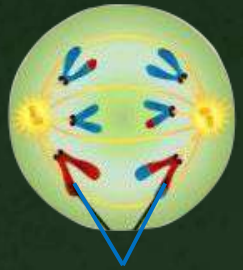
Homologous chromosomes

Homologous chromosomes move to the opposite poles of the cell

Anaphase II



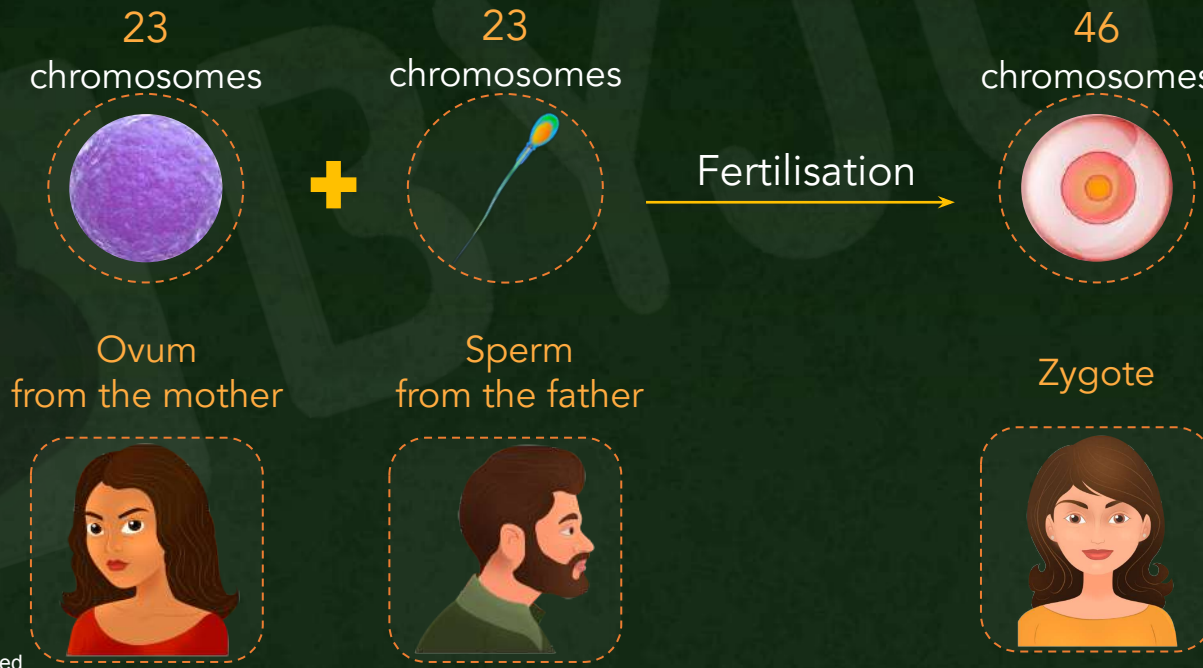
Centromeres divide, chromatids move to the opposite poles of the cells



Sister chromatids separate

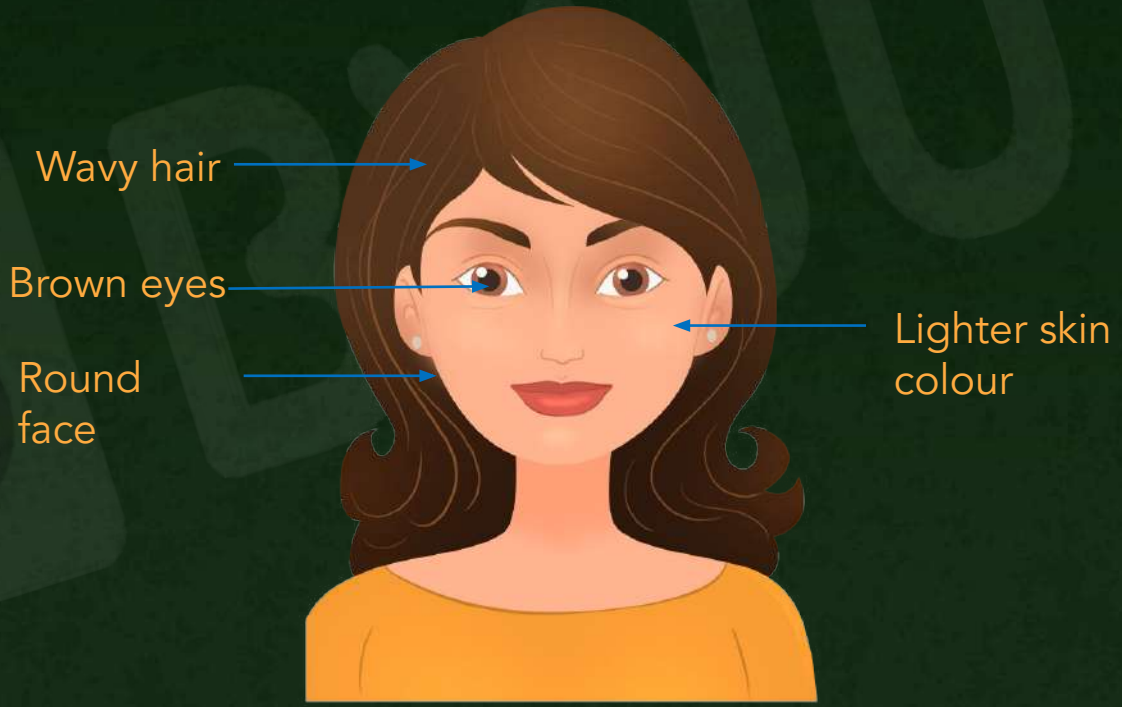
Genotype

- Genotype is the genetic constitution of a living organism.
- It is the sum total of genes inherited from both the parents.

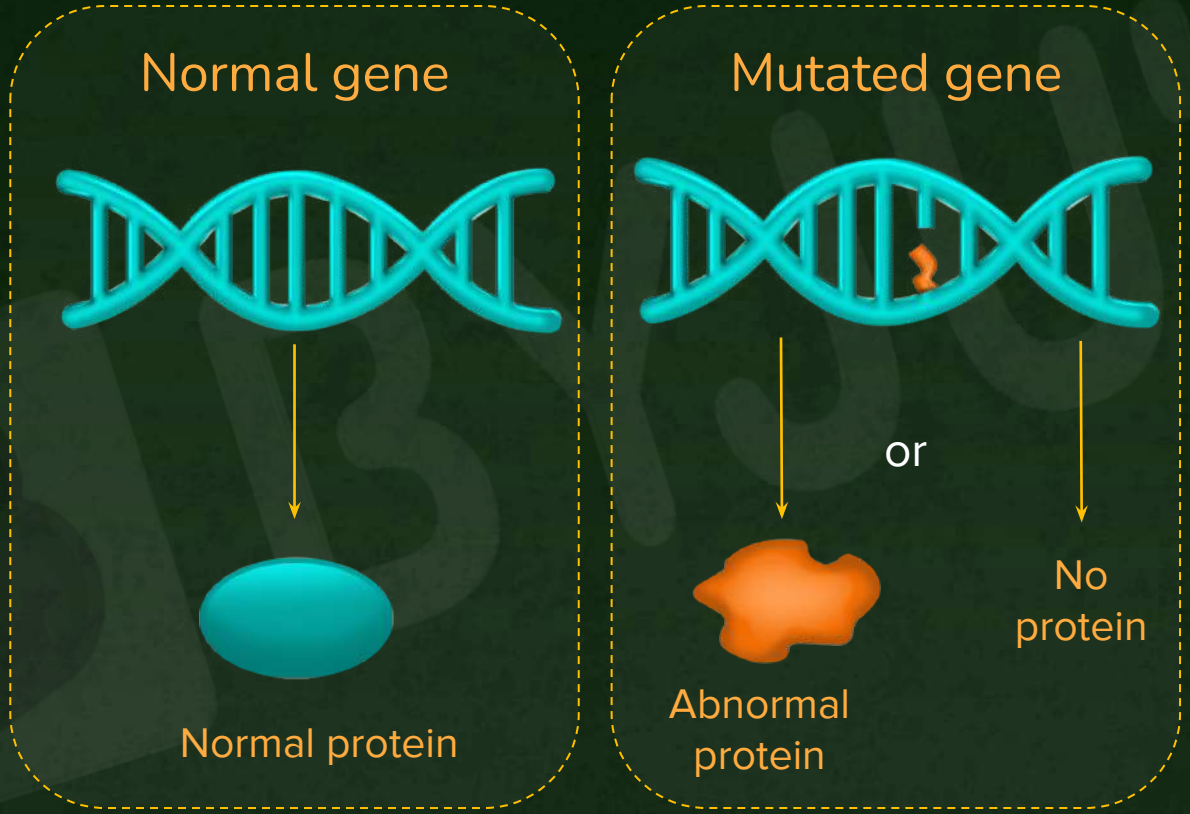


Phenotype

Phenotype can be defined as the observable characteristics of an organism.



Normal v/s Mutated Product

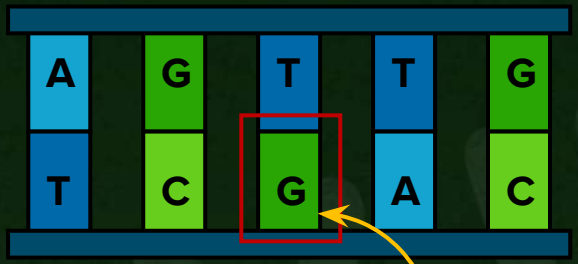


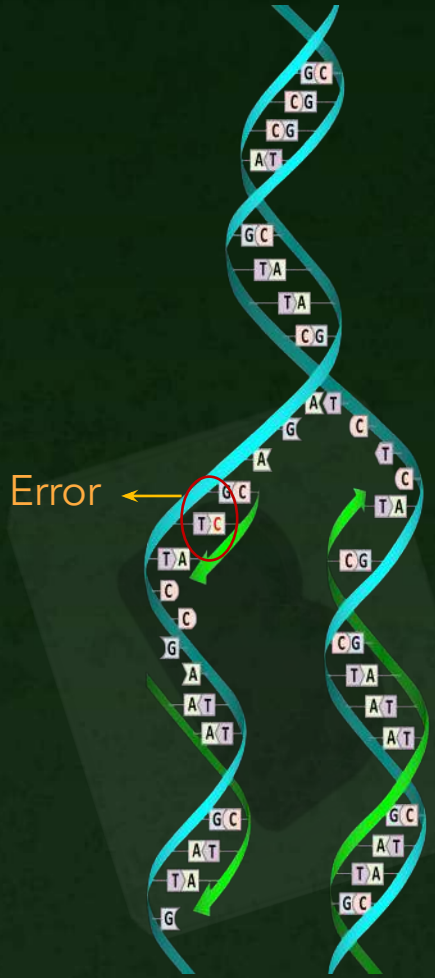
Mutated gene either codes for abnormal protein or does not code for any protein.

Mutation

- Mutation is a phenomenon that results in alteration of DNA sequences and consequently results in changes in the genotype and phenotype.
- Mutations occuring at DNA level are called genetic mutations and those at the chromosomal level are called chromosomal mutations.

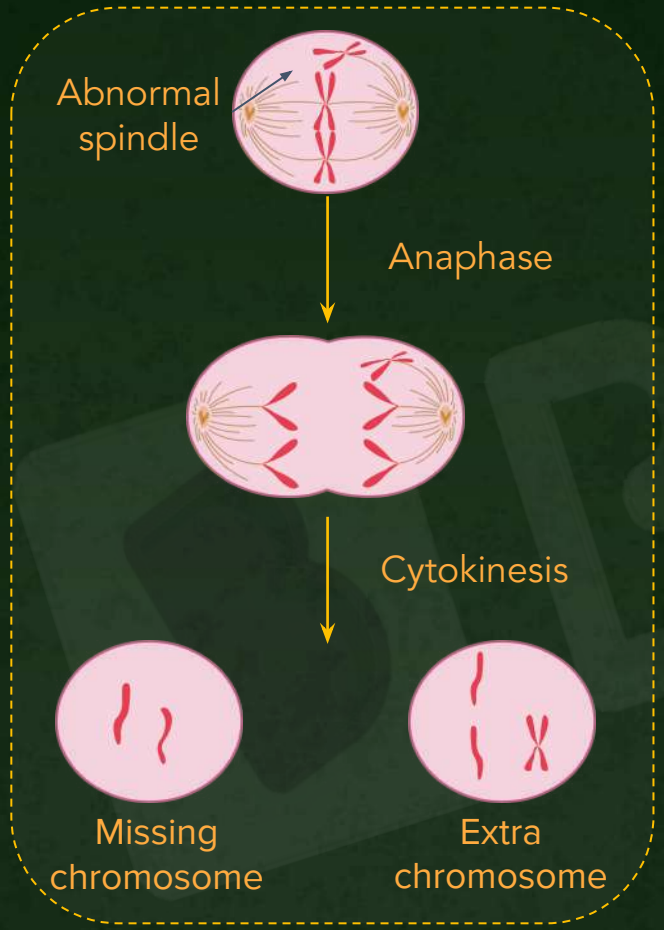
DNA Level Mutation





- Adenine (A) normally binds with Thymine (T) and Guanine (G) with Cytosine (C).
- At the DNA level, a change in the nucleotide base sequence may occur, resulting in mismatched base pairs.
- This is referred to as point mutation.
- These errors can be due to addition of an improper nucleotide, or an extra nucleotide or also because of removal of a nucleotide.
- They ultimately result in the production of an improper protein or no protein or cause no change in the protein produced (silent mutation).

Chromosomal Level Mutation



- Errors can result from improper distribution of chromosomes during cell division.
- This occurrence is called as nondisjunction.
- These errors occur when the duplicated genetic material is getting distributed between the daughter cells, during anaphase.
- Such errors lead to chromosomal mutations, as they occur at the chromosomal level.

Causes of Mutations

Internal

- Natural, spontaneously occurring unpredictable changes in the DNA cause mutations
- Absence of mutagen

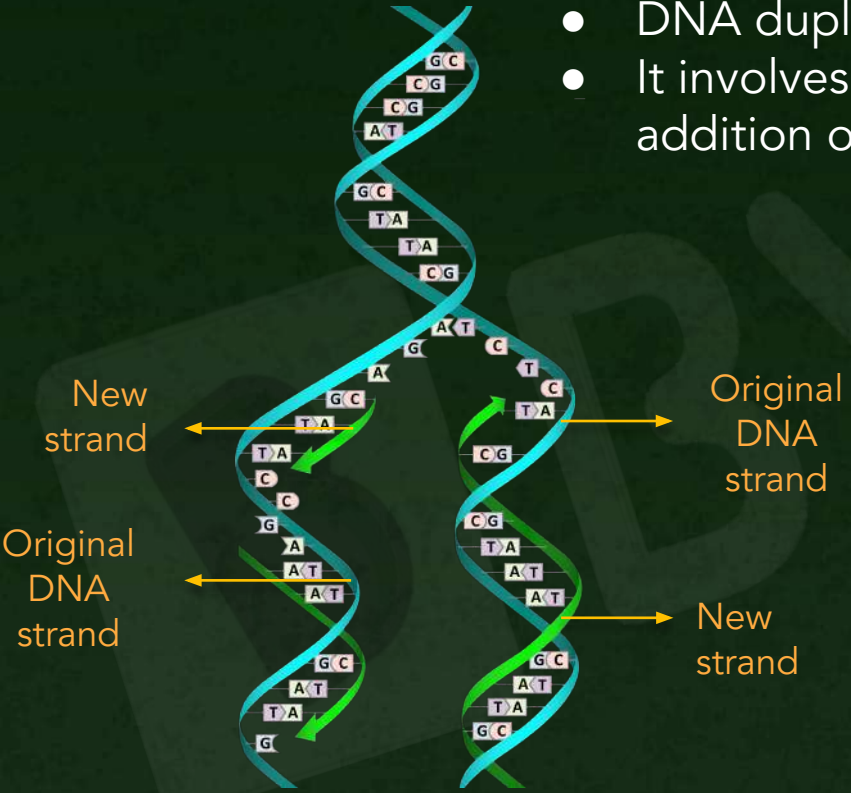
External

- Introduction of changes in the DNA through external agents cause mutations
- Requires presence of mutagen

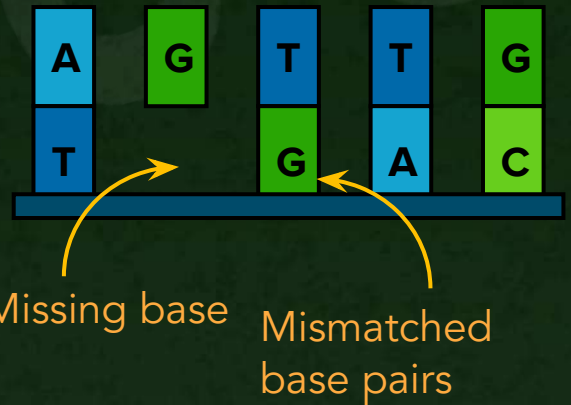
Mutagen is any substance that changes the genetic material of an organism and causes mutation.

S-Phase Problems

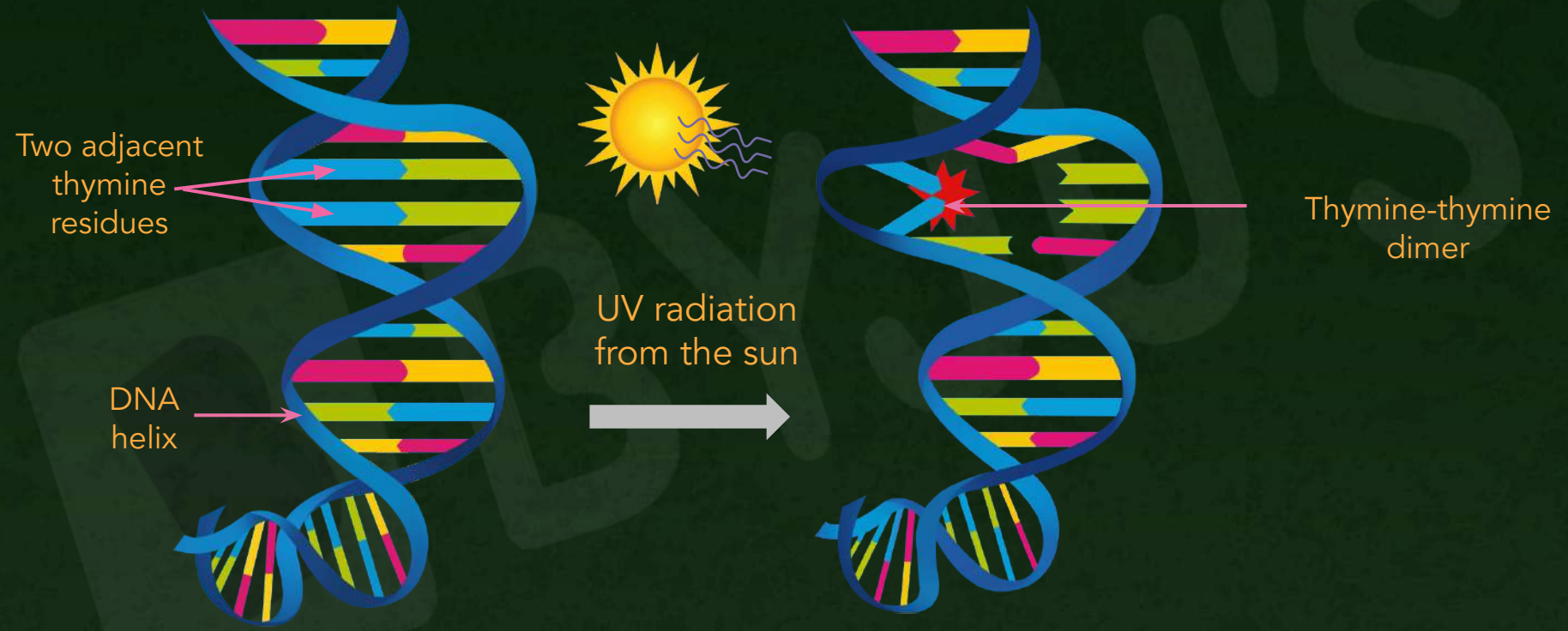
- DNA duplication takes place during S-phase.
- It involves the formation of new strand by the addition of new nucleotides.



DNA Replication



The UV Effect



UV radiation can dimerize the DNA base and hence damaging the DNA structure and arrangement. Thus, the UV radiations result in mutations.

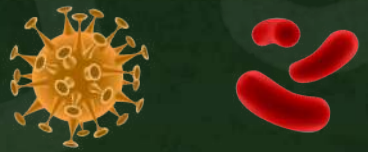
External Agents Causing Mutations

Physical agents

Example : Radiation like UV rays and X-rays

Biological agents

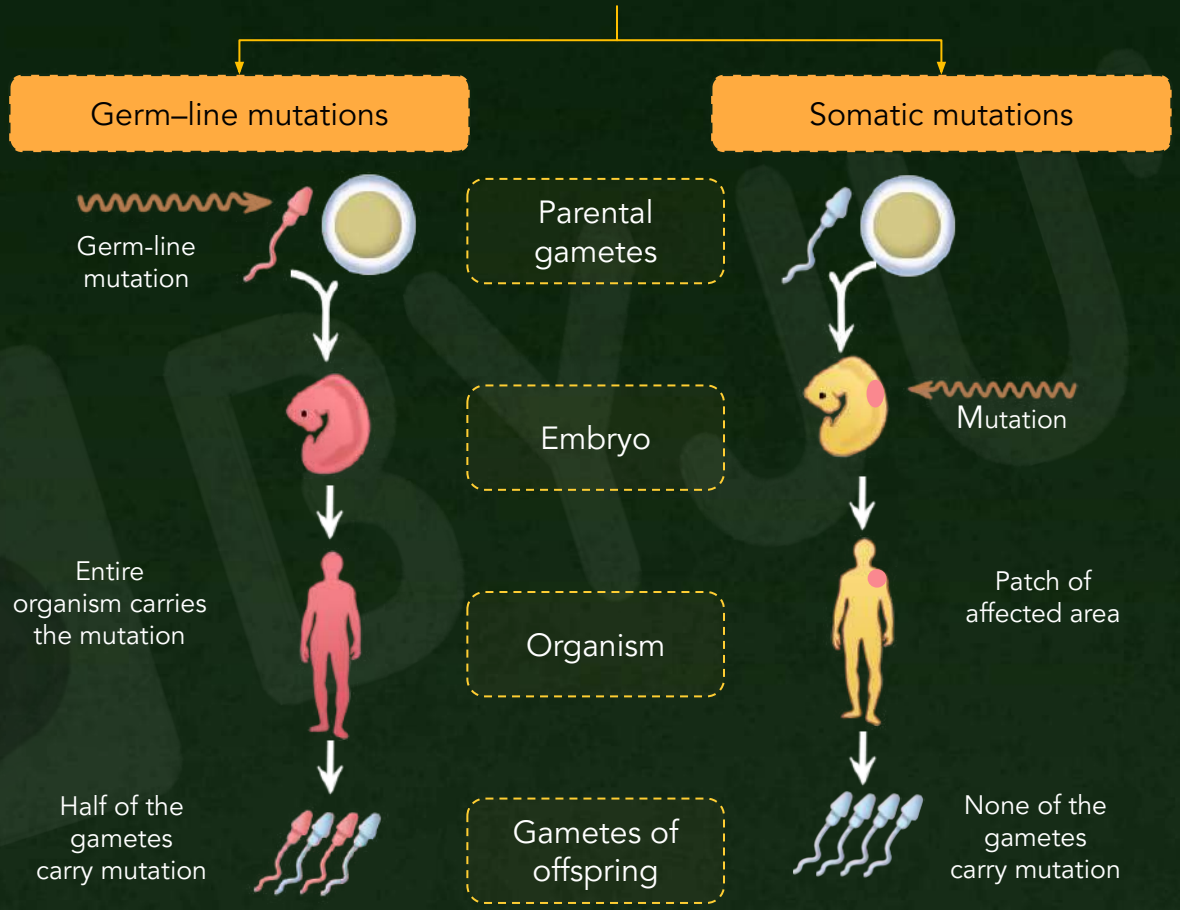
Example : Viruses and bacteria



Chemical agents

Example : Cigarette compounds, benzoyl peroxide

Types of Mutations



Effects of Mutations

1. No change

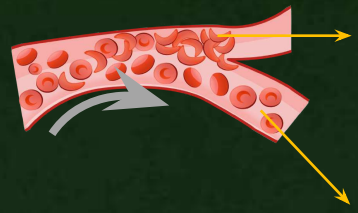
- No noticeable effect on the phenotype
- Causes no change in the amino acid sequence
- Also known as silent mutations

2. Minor change

- Causes minor changes in the phenotype of the organism
- Not dangerous for survival of the organism

3. Major change

- Can have strong negative effects on the phenotype of organism
- Can cause death of the organism
- Also called lethal mutation



Sickle red blood cells, blocking the blood flow

Healthy red blood cell

Sickle cell anaemia

Change in the sequence of nucleotide in DNA is called as

A. mutagen

B. mutation

C. recombination

D. translation

Change in the sequence of nucleotide in DNA is called as

A. mutagen

B. mutation

C. recombination

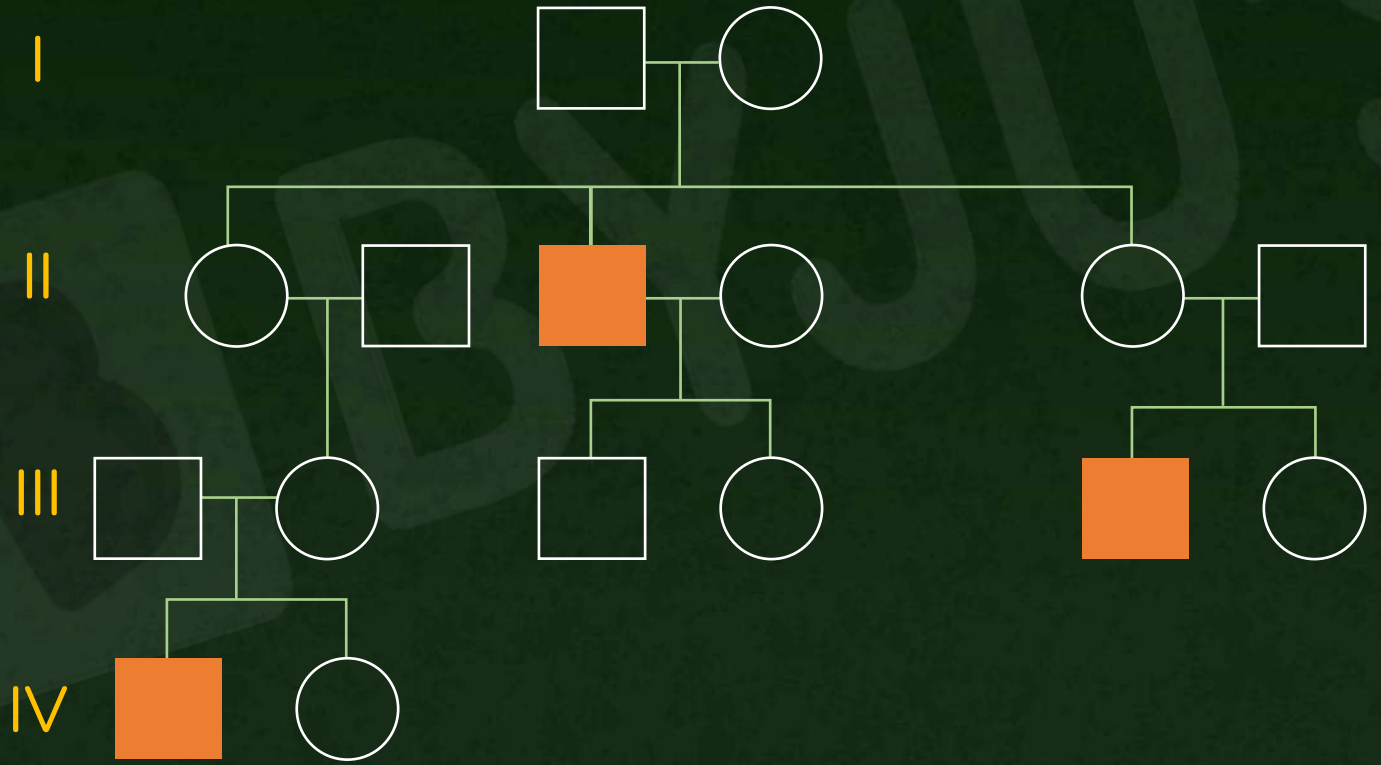
D. translation

Pedigree Analysis

- Pedigree analysis is the study of a particular trait that is inherited from one generation to another.
- It helps to understand the pattern of inheritance for a particular trait.
- It also helps to know whether the trait is dominant or recessive.
- Propositus is the name given to the person/organism for whom the analysis is made.
- The entire family history data of the propositus is mandatory for pedigree analysis.
- The history of the character trait under investigation is mapped by the investigator.
- In the pedigree chart, standard symbols are used.

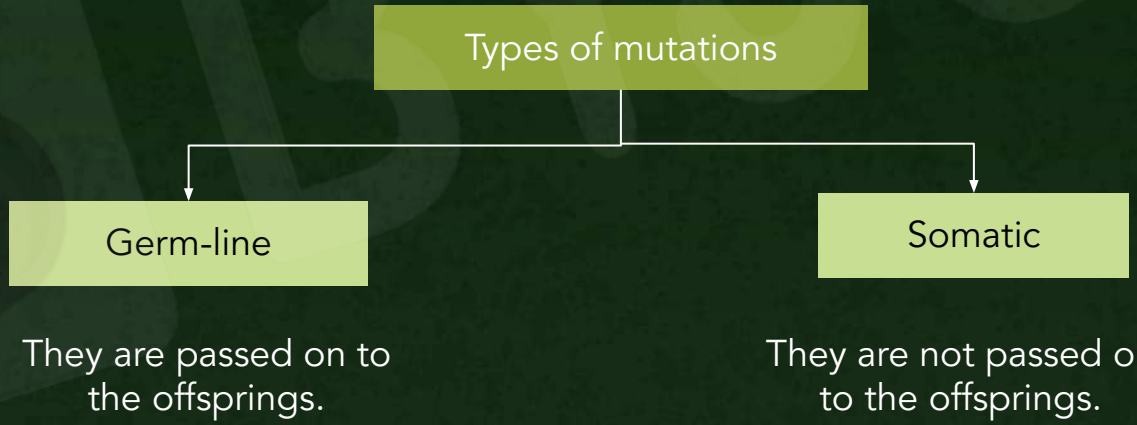
Pedigree Chart

A pedigree chart is a representation of a family chart showing the transmission of a particular trait or disease



Summary

- Mutation
  - A phenomenon that results in alteration of DNA sequences or chromosomes which consequently brings in changes in the genotype and phenotype.



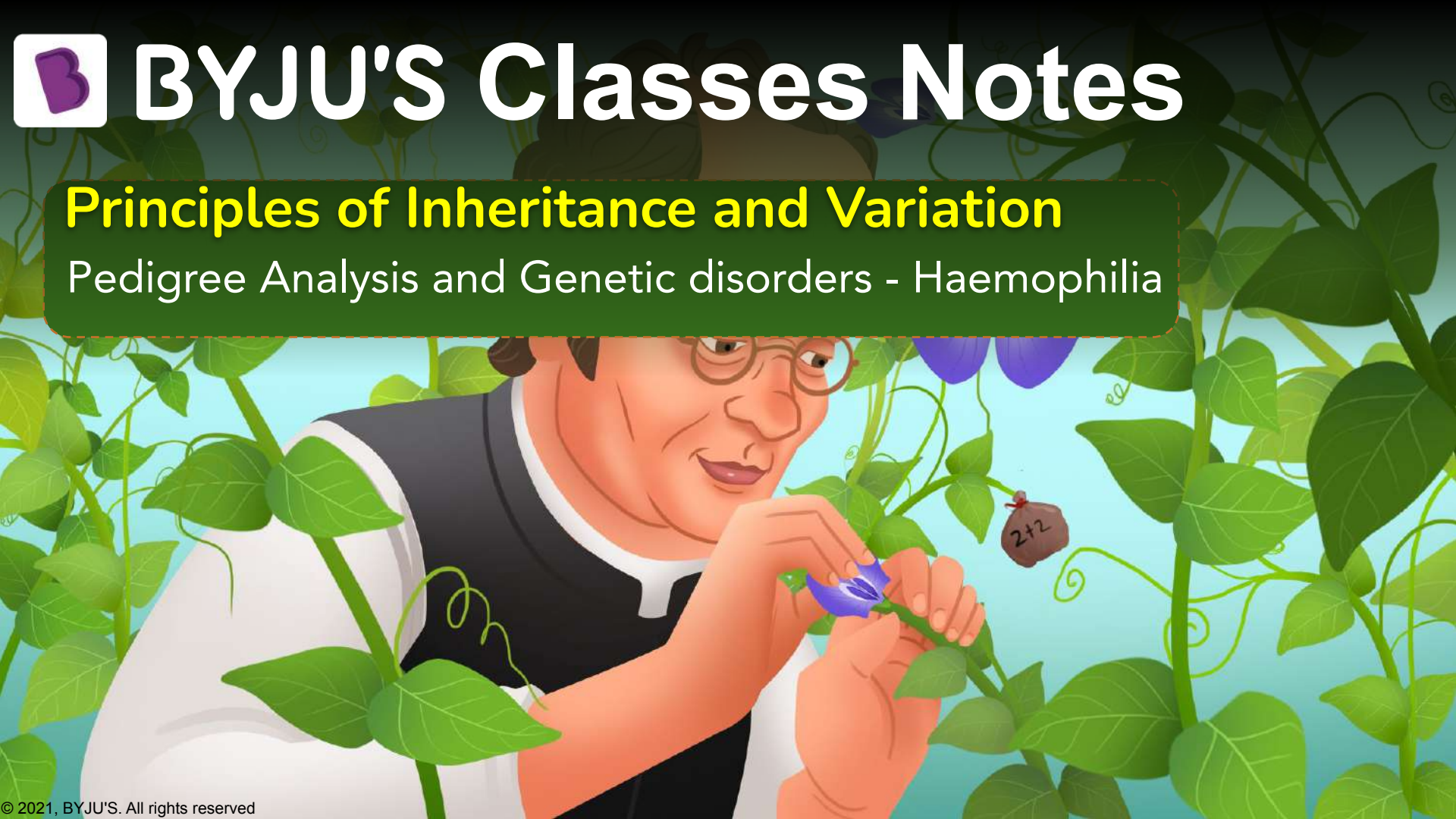
Summary

- Pedigree analysis
  - It is the study of a particular trait that is inherited from one generation to another.
  - It helps to understand the pattern of inheritance for a particular trait, and also to know whether the trait is dominant or recessive.

BYJU'S Classes Notes

Principles of Inheritance and Variation

Pedigree Analysis and Genetic disorders - Haemophilia

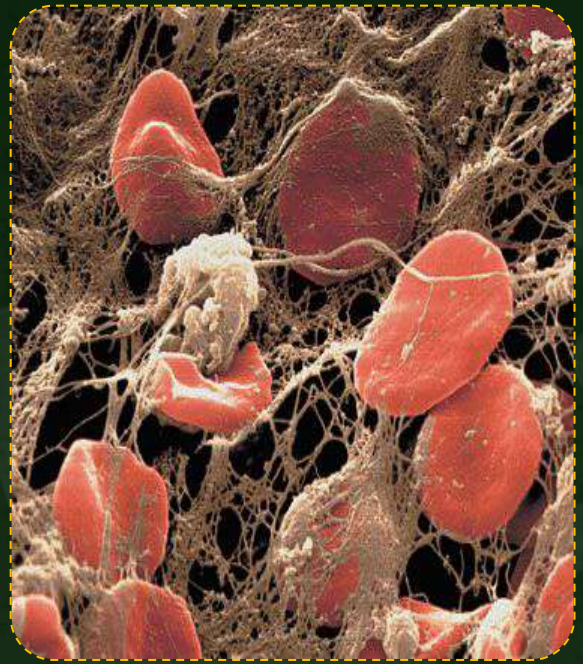


Key Takeaways

1. Pedigree analysis
2. Genetic disorders
3. Mendelian disorders
   - Haemophilia

Summary

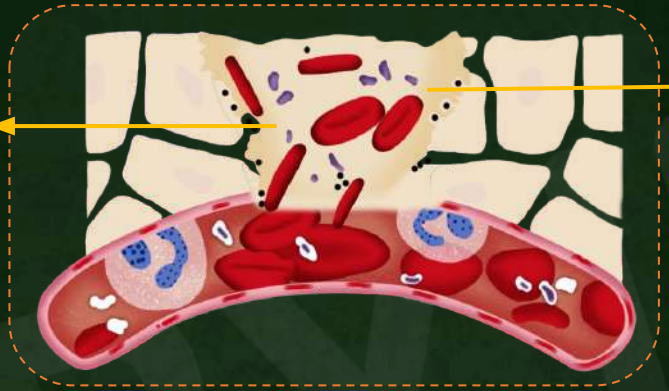
Recall! Blood Clotting



- Fibrin
- Platelets
- Clotting factors
- Blood cells

Blood clotting ingredients

Recall! Blood Clotting



Damaged tissue outside vessel

Platelets in damaged blood vessel

Cascade of clotting factors

Cascade of clotting factors

Ca^{2+}

Ca^{2+}

Prothrombin activator

Prothrombin

Ca^{2+}

Thrombin

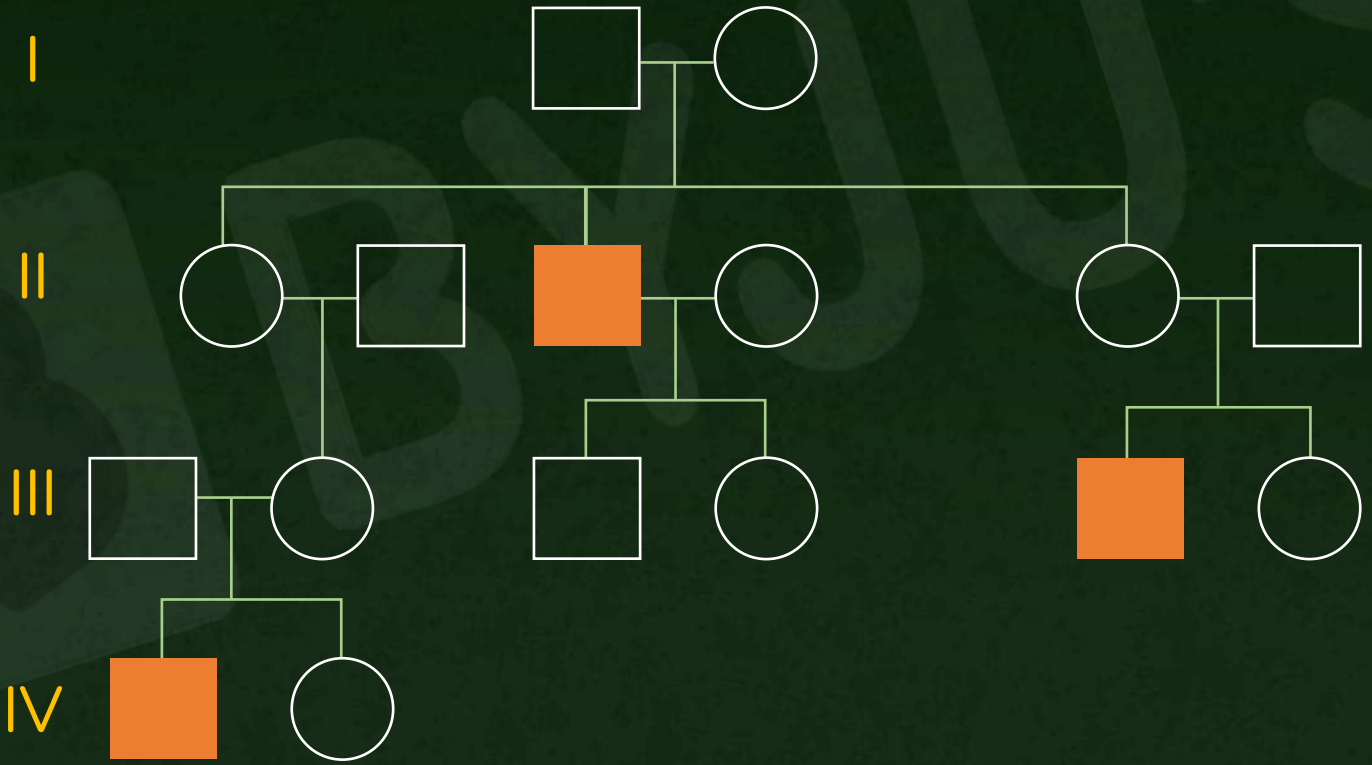
Fibrinogen

Fibrin

Stable clot

Pedigree Analysis

A pedigree chart is a representation of a family chart showing the transmission of a particular trait or disease.



Did You Know?

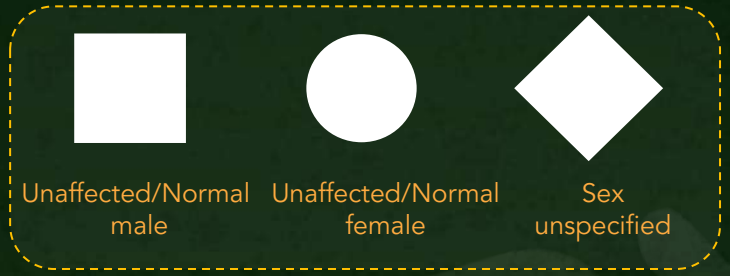
- Pedigree is derived from the French phrase "pied de gru" which means foot of a crane.

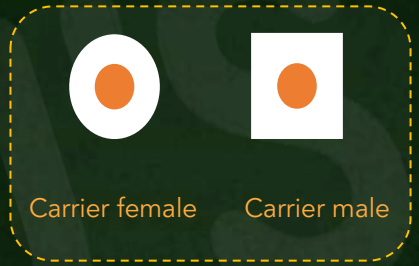
- The lines and the split lines used in the chart resemble the thin leg and tiny foot of the crane.

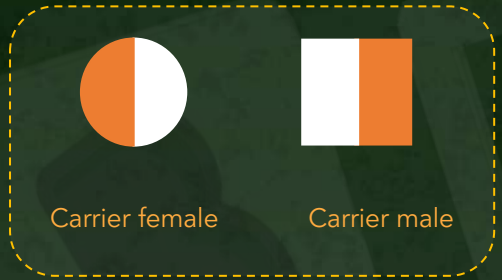
Uses of Pedigree Analysis

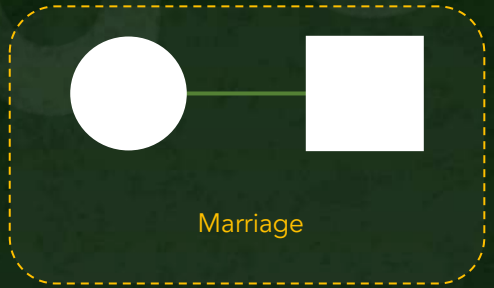
- Allows better understanding of transmission of genes within a family
- Can help in predicting the occurrence of a disease prevalent in the family
- Useful in genetic counselling

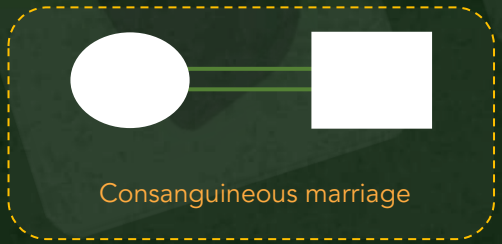
Understanding the Symbols

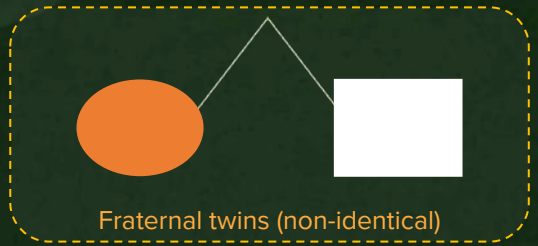


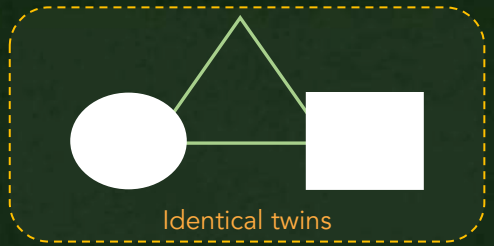






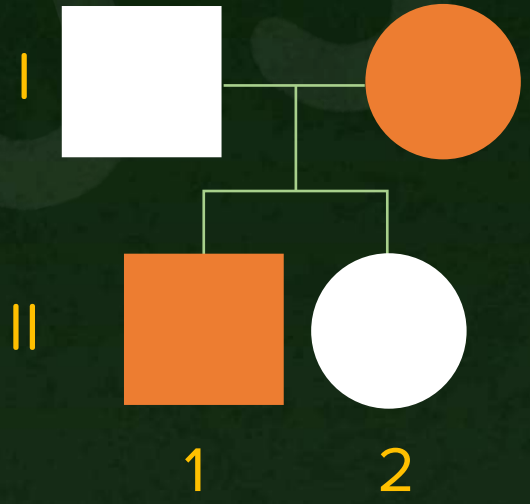




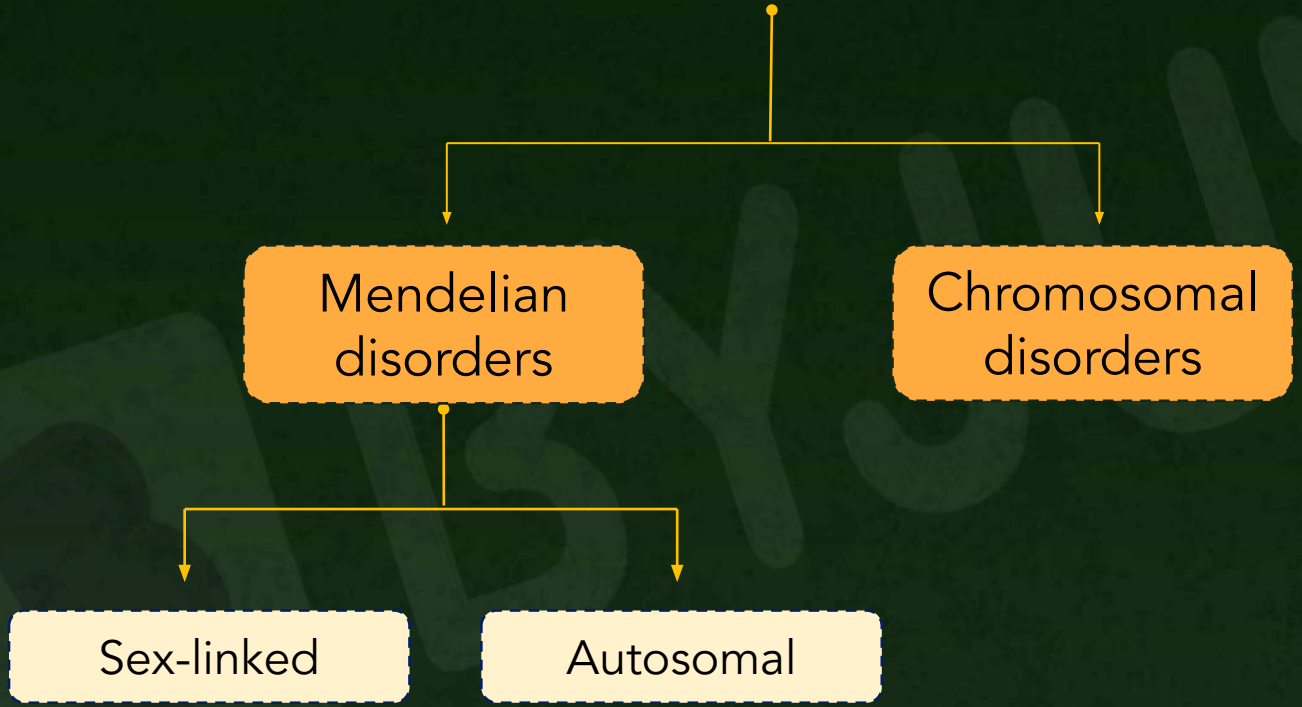


Understanding the Symbols

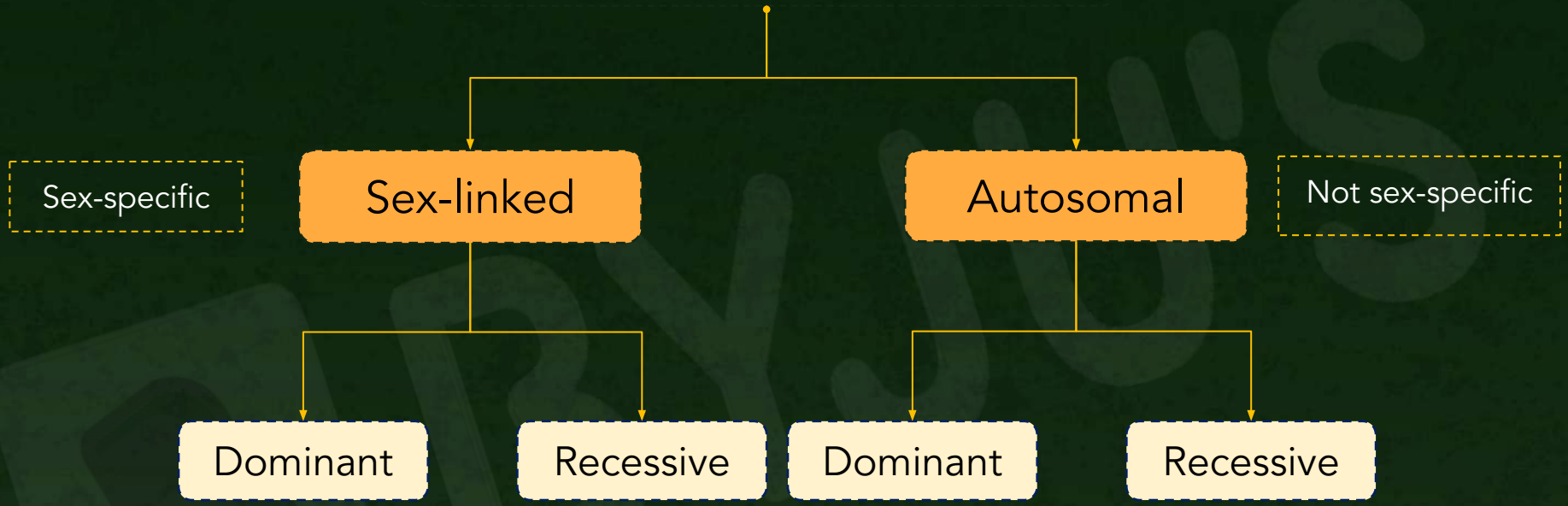
- Parents and 1 boy and 1 girl (in order of birth)
- Roman number - Generation
- Arabic number - No. of individual children



Genetic Disorders



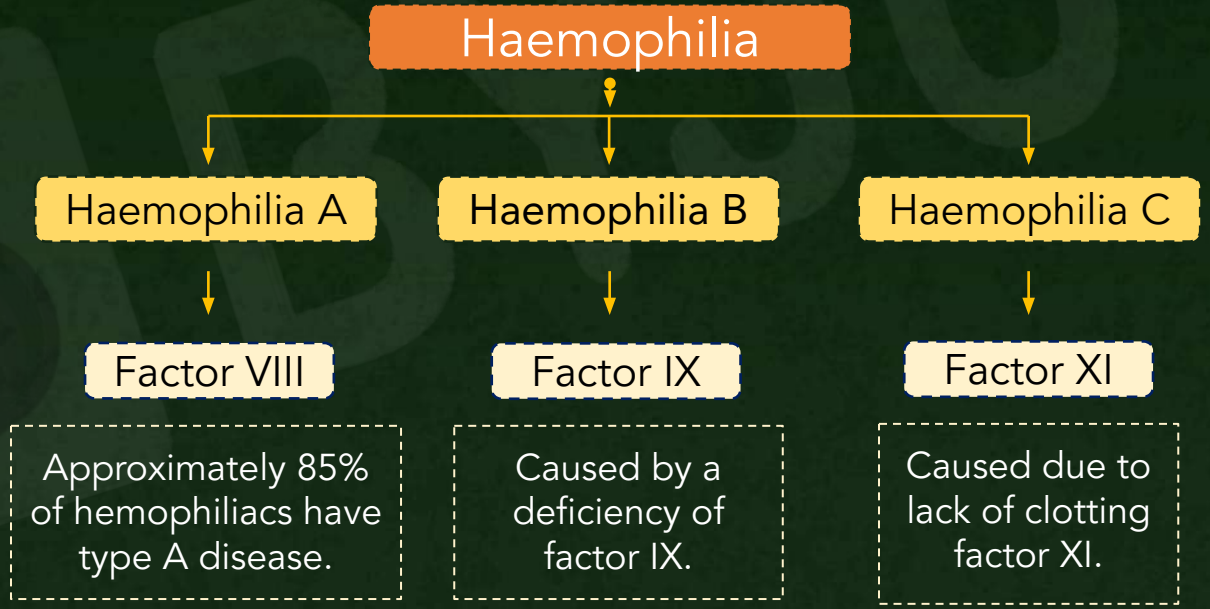
Mendelian Disorders



- Mendelian disorders are mainly determined by alteration or mutation in a single gene.
- The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis.

Haemophilia

- Haemophilia is an X- linked, inherited genetic disorder that impairs the body's ability to make clots.
- A person suffering from haemophilia either has low supply of one of the factors needed to clot blood or completely lacks those factors.
- Two common factors that affect blood clotting are factor VIII and factor IX.



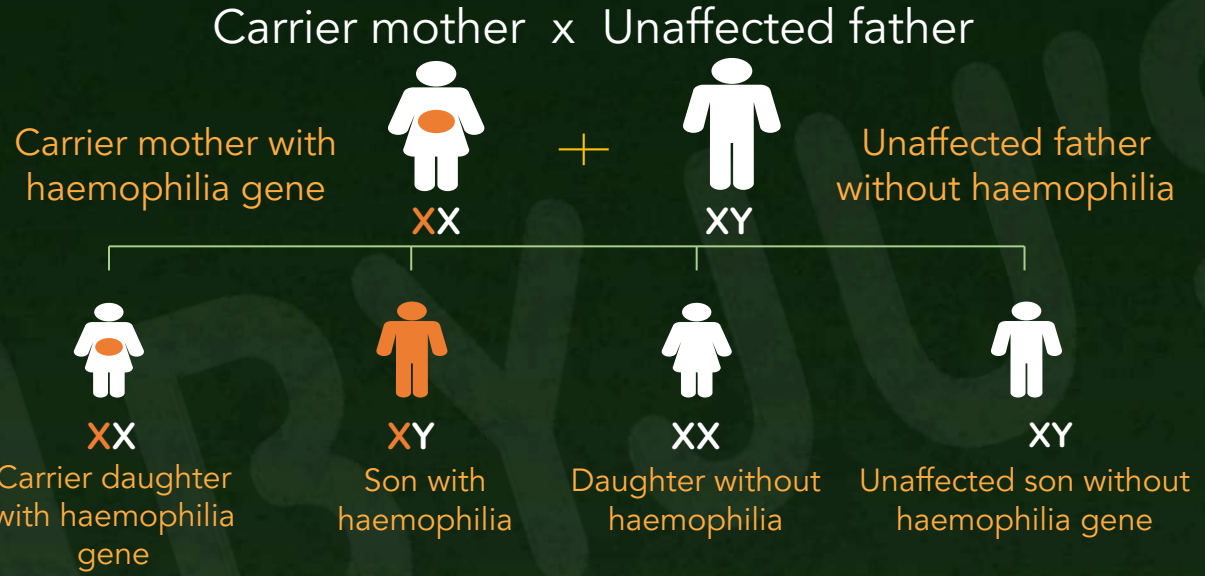
Haemophilia

- It is sex-linked disease which is also known as bleeder's disease.
- The patient will continue to bleed even from a minor cut.
- It is due to the presence of a recessive sex linked gene 'h', carried by X-chromosome.
- A female becomes haemophilic only when both its X chromosomes carry the gene (X^hX^h).
- Such females generally die before birth because the combination of these two recessive alleles is lethal.
- A female having only one allele for haemophilia (XXh) appears normal.
- Such females are known as carriers.
- In case of males, a single gene for the defect is able to express itself as the Y-chromosome is devoid of any corresponding allele (XhY).

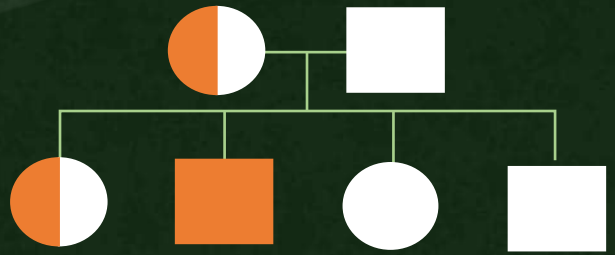
Haemophilia

- Haemophilia is also called Royal disease as it has been quite common in the royal families of europe.
- The disease spread to them through the children of Queen Victoria.
- The ancestors of the queen did not possess the disease.
- It appears that the gene for haemophilia developed either in the germ cells of her father or herself through mutation.
- Being sex-linked, the gene for haemophilia shows criss-cross inheritance.
- Its frequency is 1 in 7000 in human males and 1 in 10000000 in females.

Haemophilia

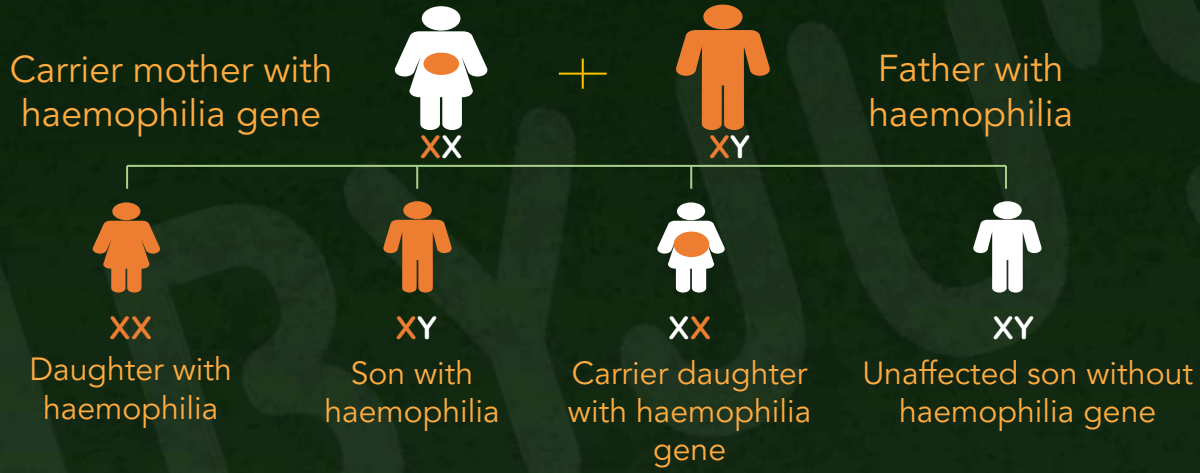


Pedigree chart

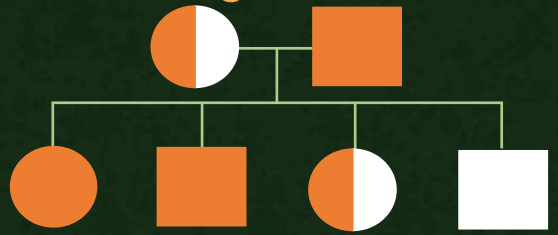


Haemophilia

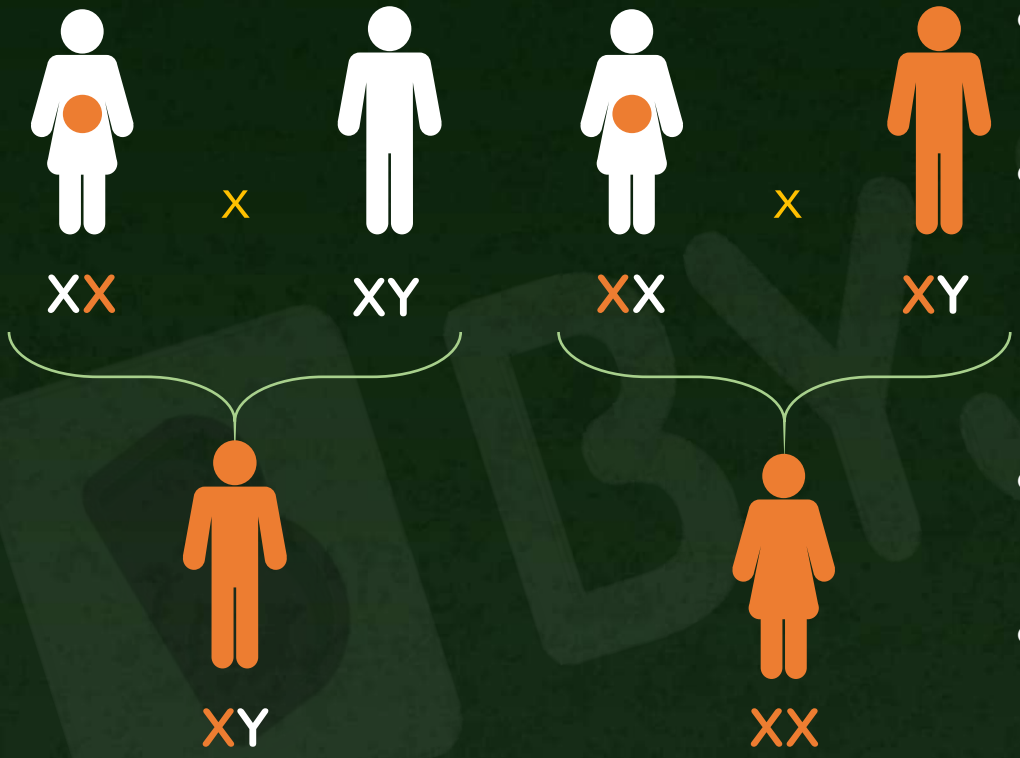
Carrier mother x Affected father



Pedigree chart



Haemophilia



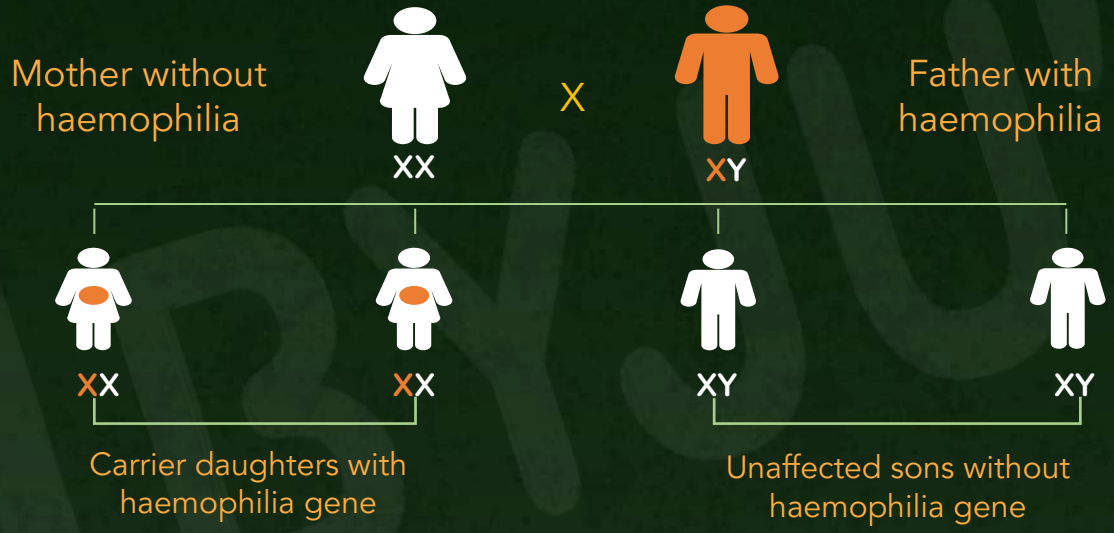
Possibility when mother is carrier and father is unaffected

Possibility when mother is carrier and father is affected

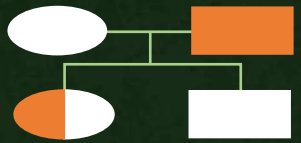
- The heterozygous female (carrier) for haemophilia may transmit it to the sons.
- The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic.
- Males are more likely to get this disease than females.
- The family pedigree of Queen Victoria shows a number of haemophilic descendents as she was a carrier of the disease.

Haemophilia

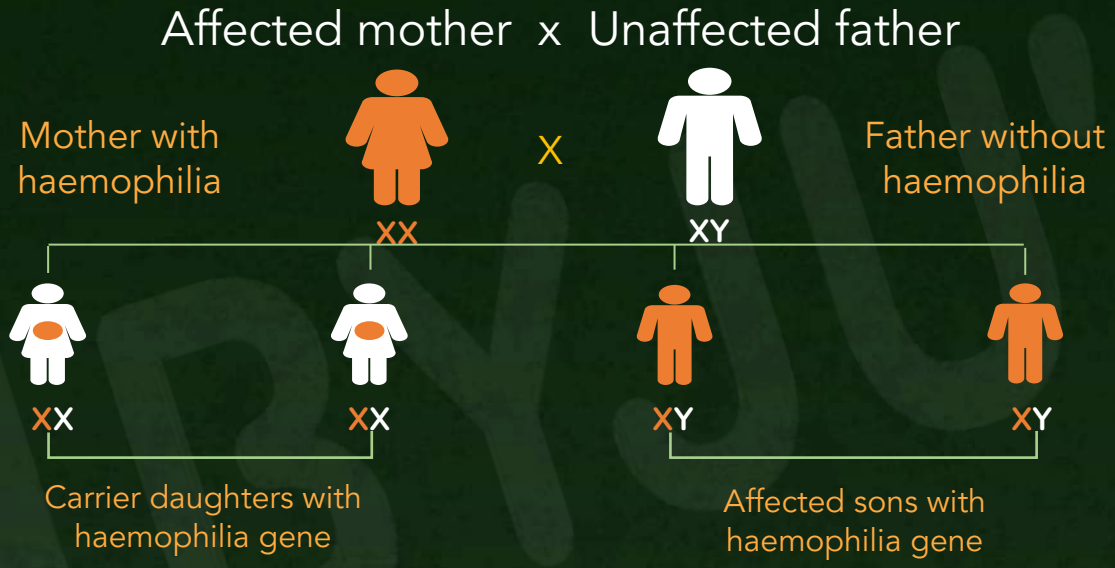
Unaffected mother x Affected father



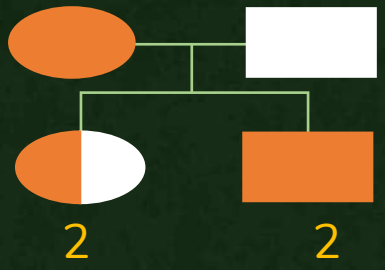
Pedigree chart



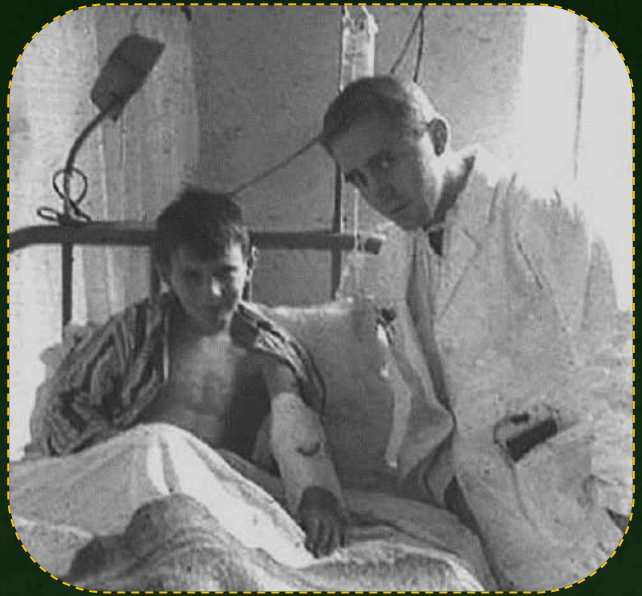
Haemophilia



Pedigree chart



Did You Know?



- Haemophilia B is also known as Christmas disease.
- The disorder was first reported in 1952 in a 5-yr old patient with the name of Stephen Christmas.

Summary

Understanding the symbols

Unaffected/Normal male

Unaffected/Normal female

Sex unspecified

Affected female

Affected male

Carrier female

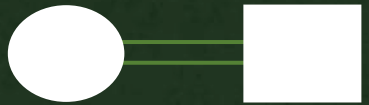
Carrier male

Carrier female

Carrier male

Deceased female and male

Marriage

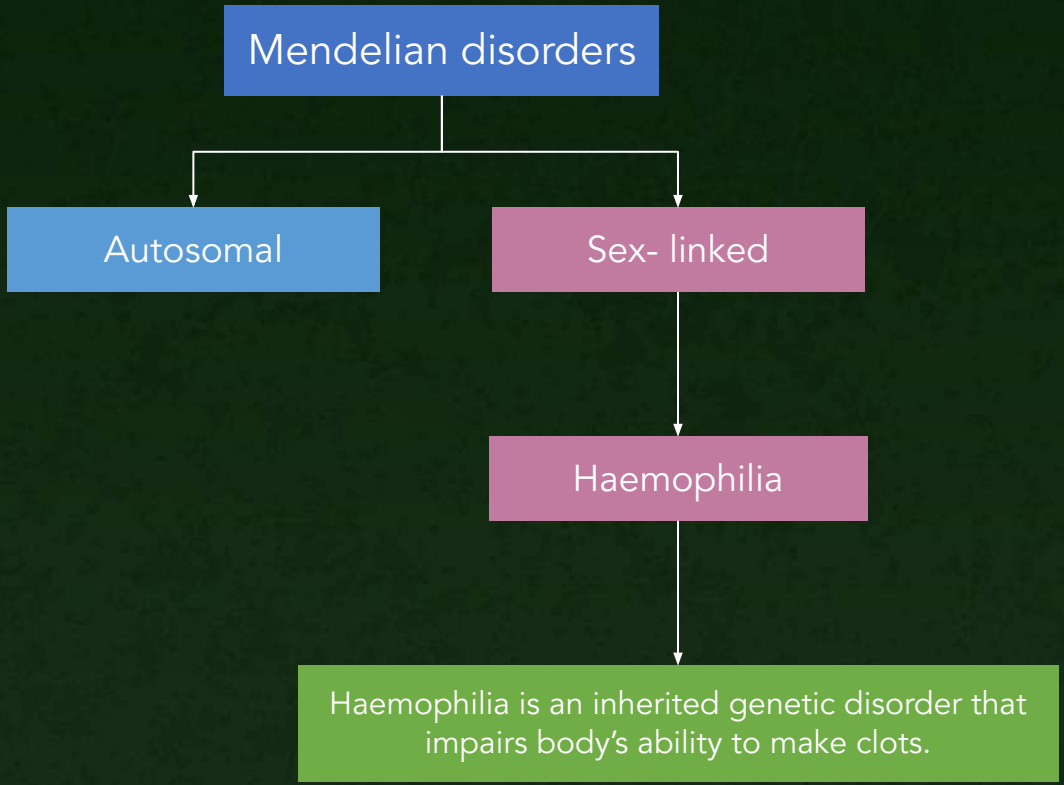


Consanguineous marriage

Fraternal twins (non-identical)

Identical twins

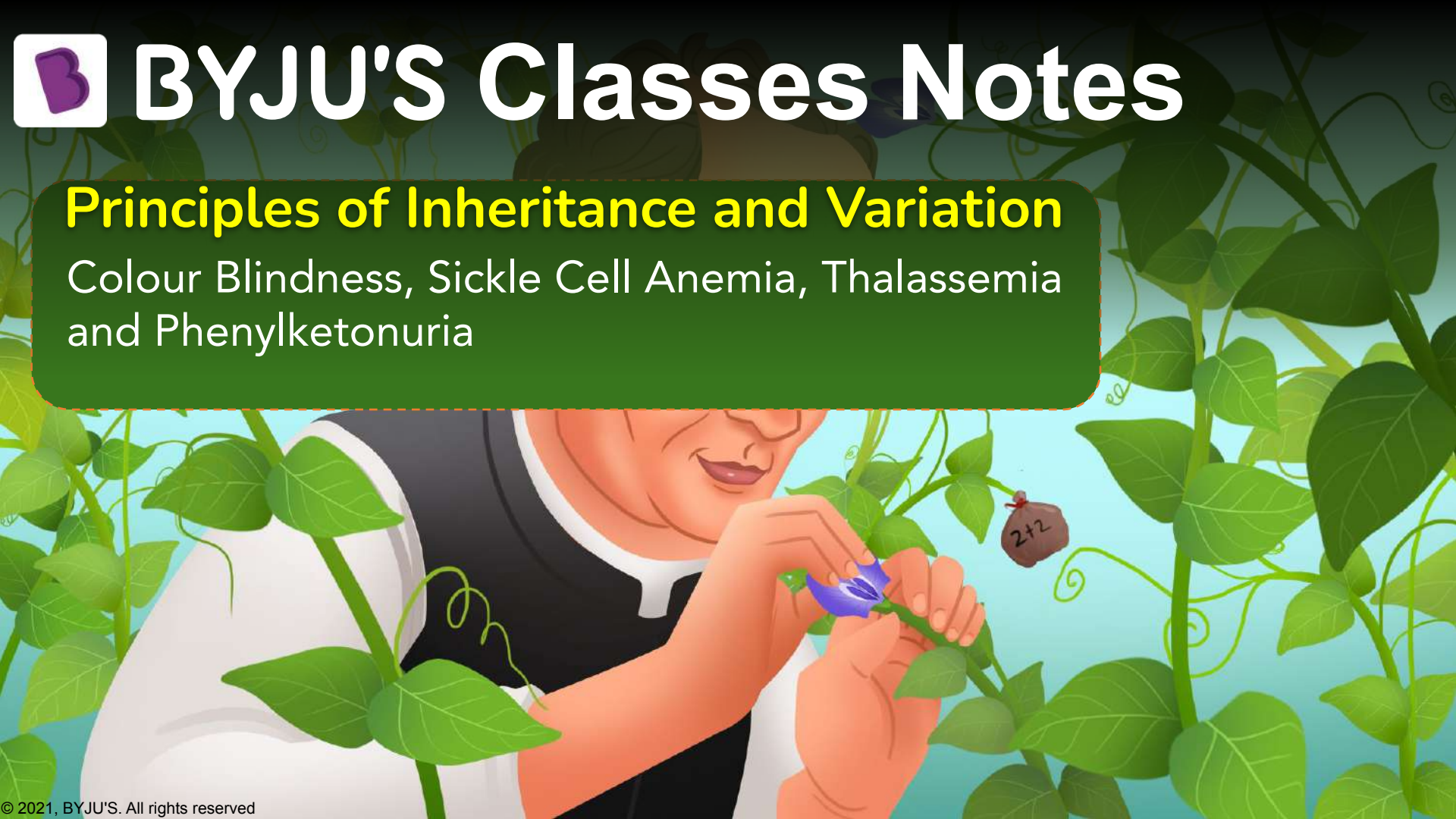
Summary



BYJU'S Classes Notes

Principles of Inheritance and Variation

Colour Blindness, Sickle Cell Anemia, Thalassemia and Phenylketonuria



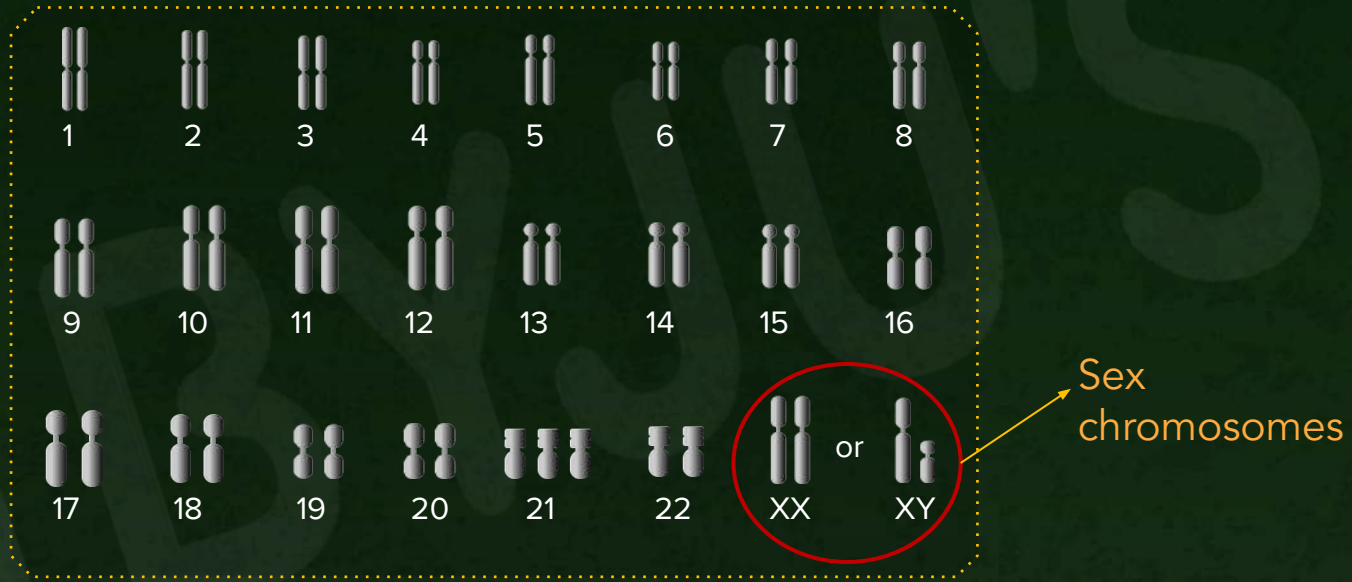
Key Takeaways

Mendelian disorders

- Colour blindness
- Sickle cell anemia
- Thalassemia
- Phenylketonuria

Summary

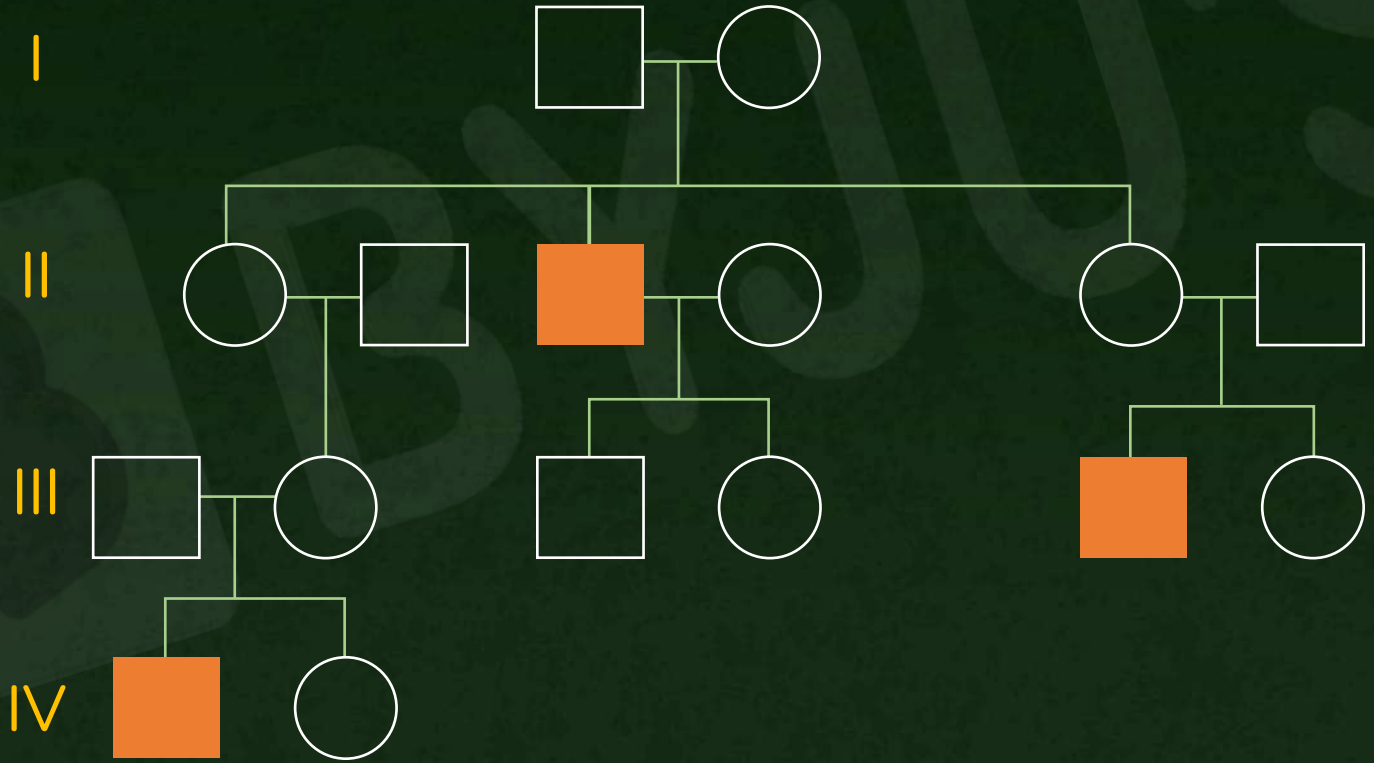
Recall! Human Karyotype



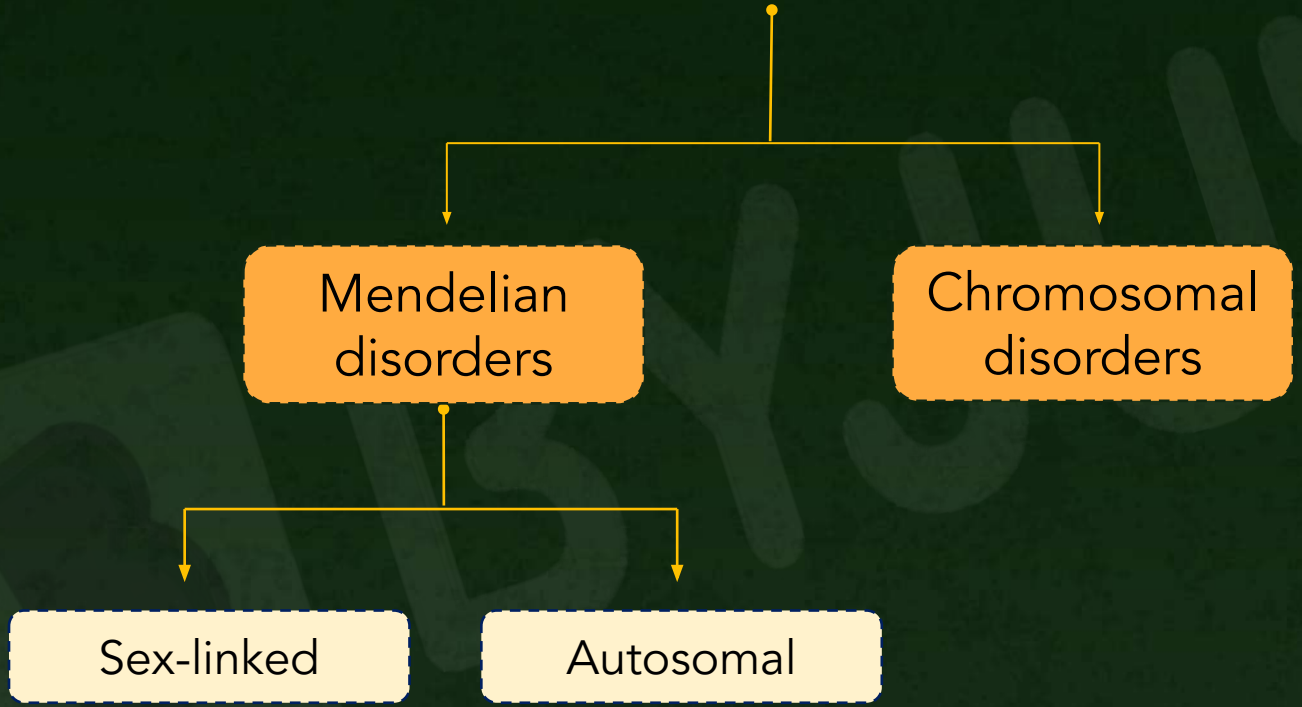
- Out of the 23 pairs of chromosomes in humans, 22 are autosomal chromosomes, and X and Y are sex chromosomes.
- Genes that are inherited via the sex chromosomes are called sex-linked, and the inheritance is called sex-linked inheritance.

Recall! Pedigree Chart

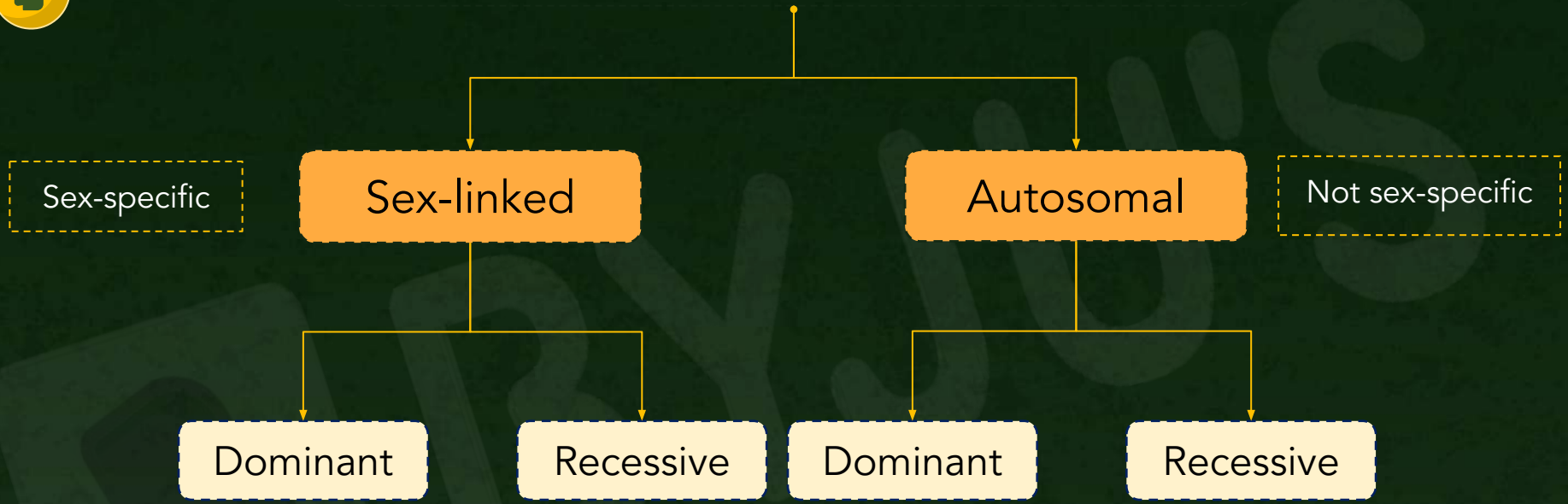
A pedigree chart is a representation of a family chart showing the transmission of a particular trait or disease.



Recall! Genetic Disorders



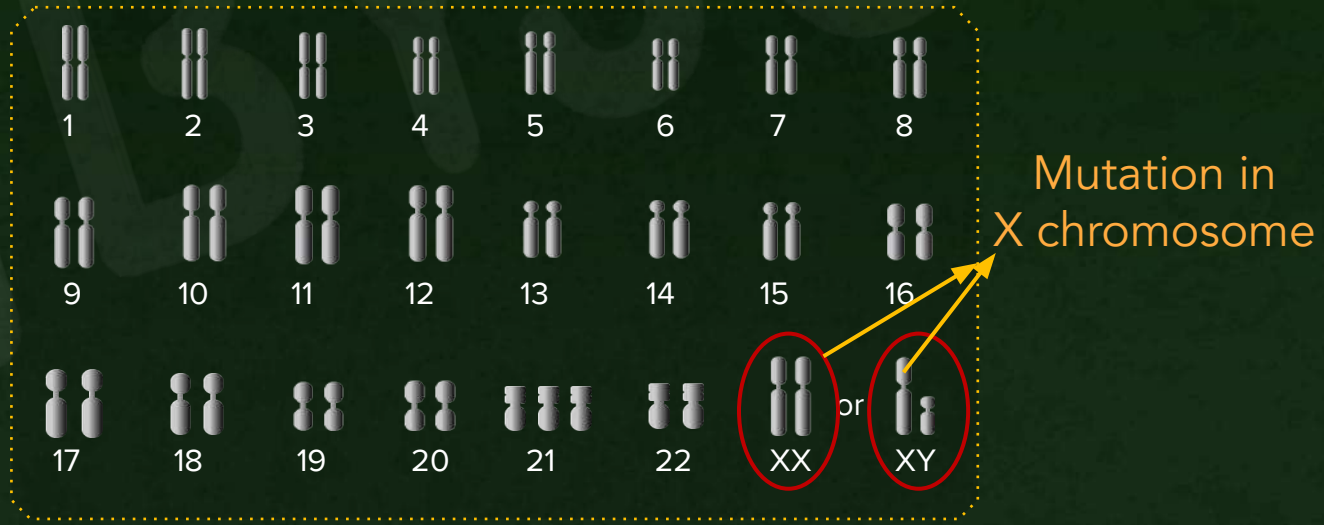
Recall! Mendelian Disorders



- Mendelian disorders are mainly determined by alteration or mutation in a single gene.
- The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis.

Colour Blindness

- Sex-linked recessive disorder due to defect in either red or green cones of eye resulting in failure to discriminate between red and green colour.
- Results from mutation in the X chromosome.
- Females are usually carriers, whereas males are more commonly affected by the disorder.



Colour Blindness

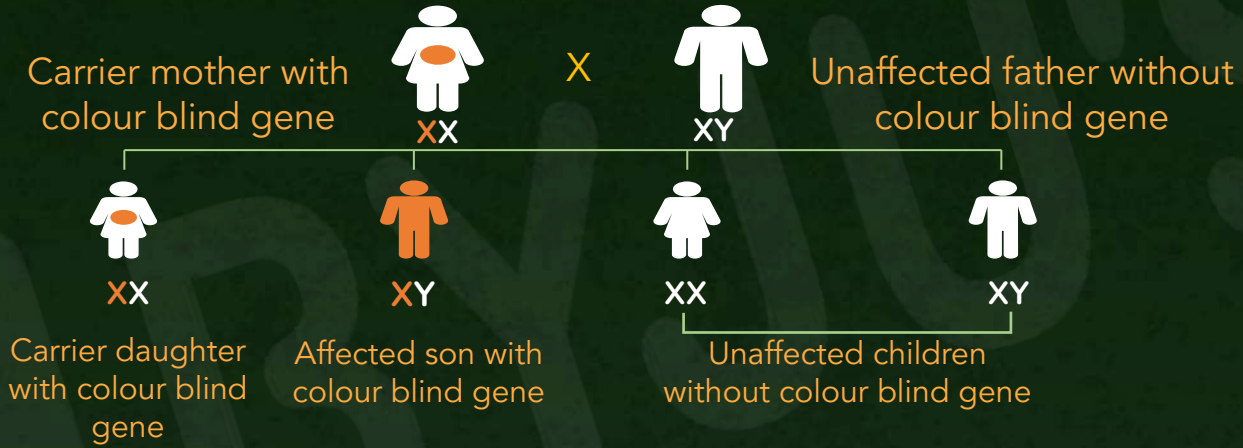
- Vision is, however, not affected and the colour blind person can lead a normal life.
- The gene for the normal vision is dominant.
- The normal gene and its recessive allele are carried by X-chromosomes.
- In females, colour blindness appears only when both the sex chromosomes carry the recessive gene ($X^c X^c$).

Colour Blindness

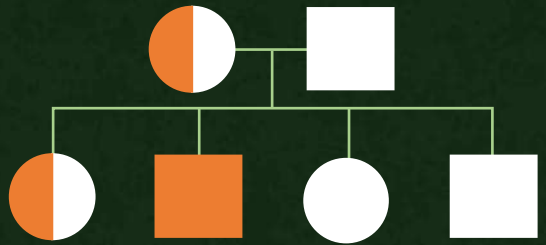
- Females are usually carriers.
- Males are at higher risk of getting colour blindness as they have only 1 X-chromosome whereas females have 2.

Colour Blindness

Carrier mother x Unaffected father



Pedigree chart



Colour Blindness

Carrier mother x Affected father

Carrier mother with colour blind gene

XX

X

XY

Affected father with colour blind gene

XX

Affected daughter with colour blind gene

XY

Affected son with colour blind gene

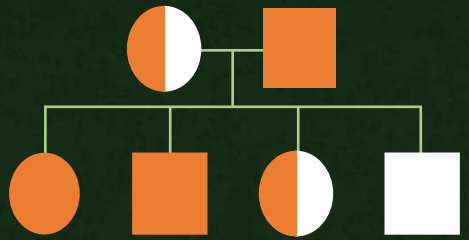
XX

Carrier daughter with colour blind gene

XY

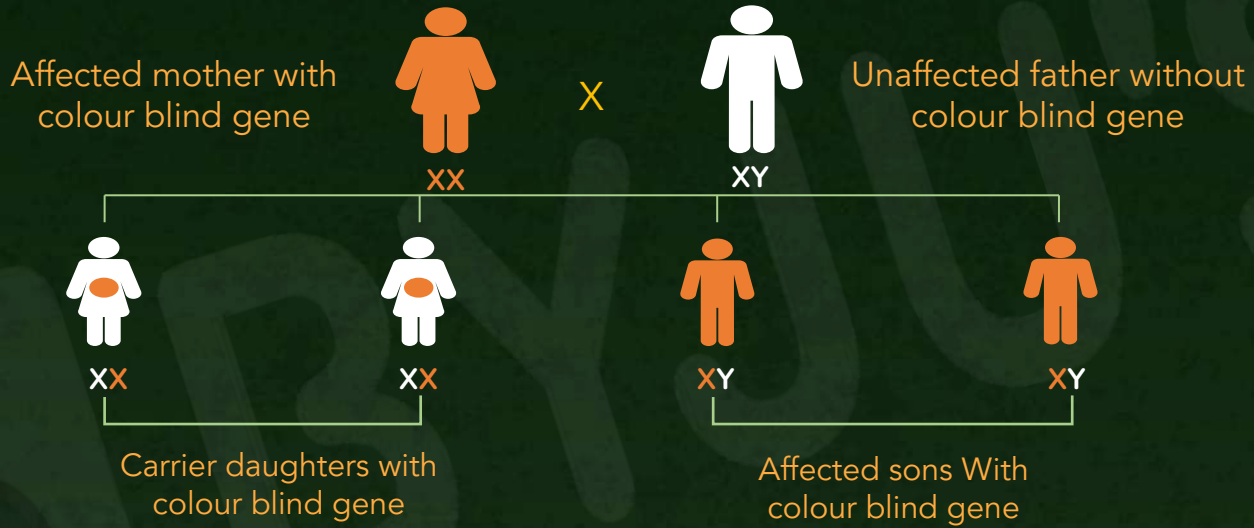
Unaffected son without colour blind gene

Pedigree chart

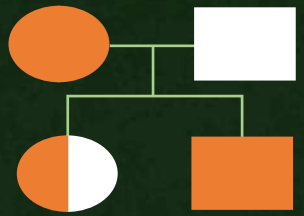


Colour Blindness

Affected mother x Unaffected father

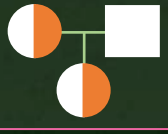


Pedigree chart

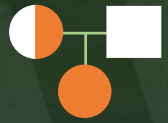


Which of the following figures represent parents with a female child affected with a disease?

A



B



C

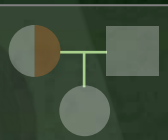
D

Which of the following figures represent parents with a female child affected with a disease?

A

B

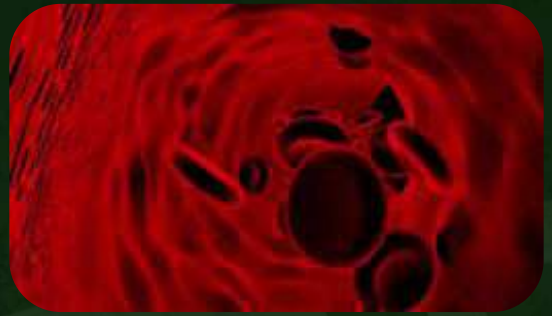
C



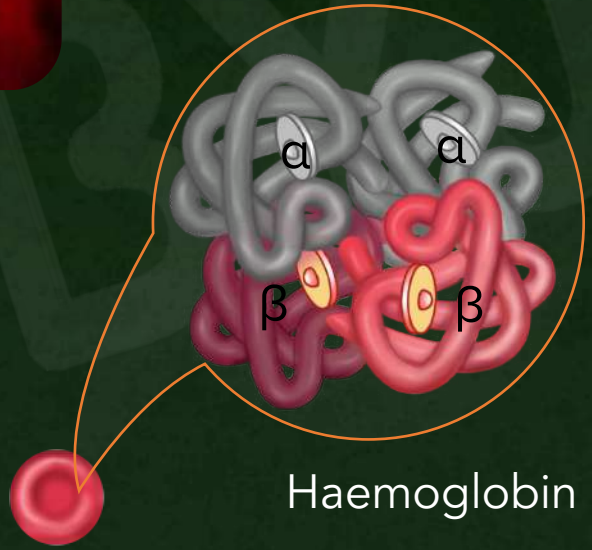
D

Sickle Cell Anemia (SCA)

Haemoglobin - The oxygen transporter



- Haemoglobin (Hb) present in the RBCs are important for carrying O_2 and CO_2 across the body.
- Hb is a protein that has 2 alpha globin chains and 2 beta globin chains.



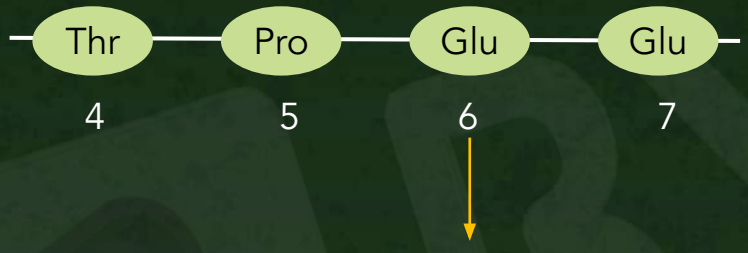
Sickle Cell Anemia (SCA)

- Sickle cell anemia is an inherited red blood cell disorder in which there aren't enough healthy red blood cells to carry oxygen throughout the body.
- Affected person has abnormal haemoglobin.
- It is an autosomal recessive disorder.
- A mutation in the Hb gene causes abnormal Hb synthesis resulting in the formation of sickle shaped cells.
- This change is caused by a single change of amino acid in the Hb protein.
- Glutamic acid in the 6th position of the beta globin chain is replaced by Valine, which causes change in Hb and thereby in RBCs, from biconcave to sickle shaped.
- This results in aggregation of RBCs in the blood capillaries leading to blockage of arteries.
- Sickle shaped RBCs also get destroyed faster than normal RBCs, resulting in severe anemia.

Sickle Cell Anemia (SCA)

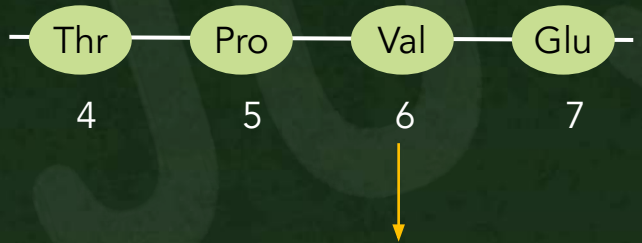
Normal RBCs vs Sickled RBCs

Normal amino acid sequence



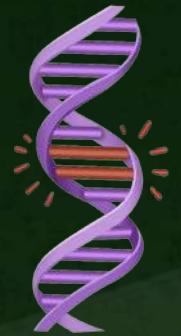
Normal red blood cells

Single change in amino acid sequence

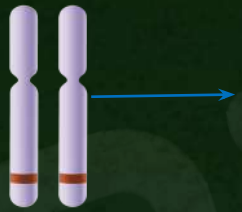


Sickle shaped red blood cells

Sickle Cell Anaemia (SCA)

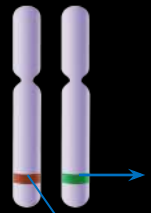


Hemoglobin gene



Mutation

Abnormal adult hemoglobin (HbS)



Normal hemoglobin A gene (HbA)

Sickle trait (HbS)

Hb^AHb^A

Normal

Hb^AHb^S

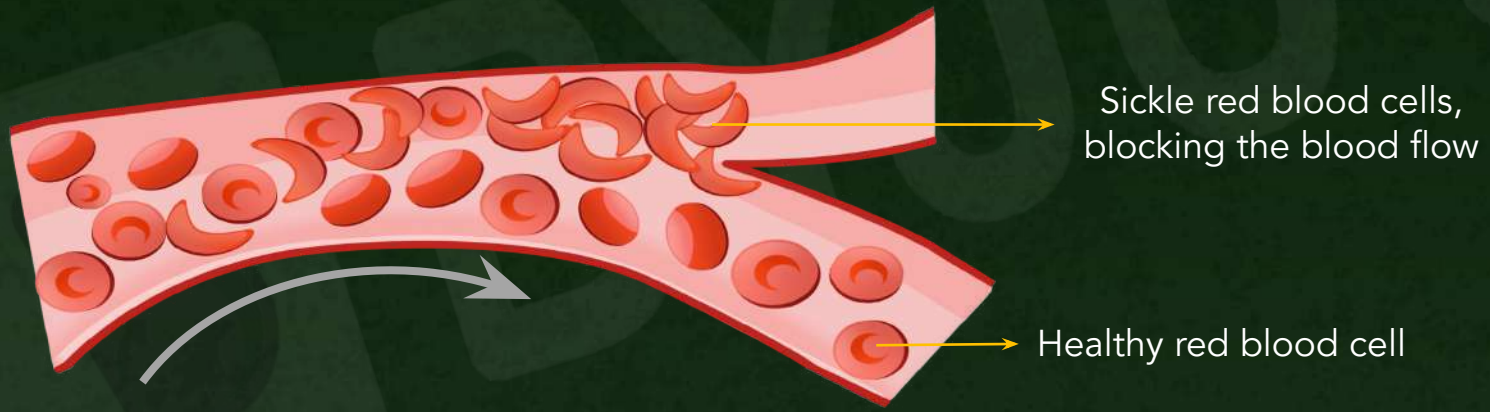
Carrier

Hb^SHb^S

SCA

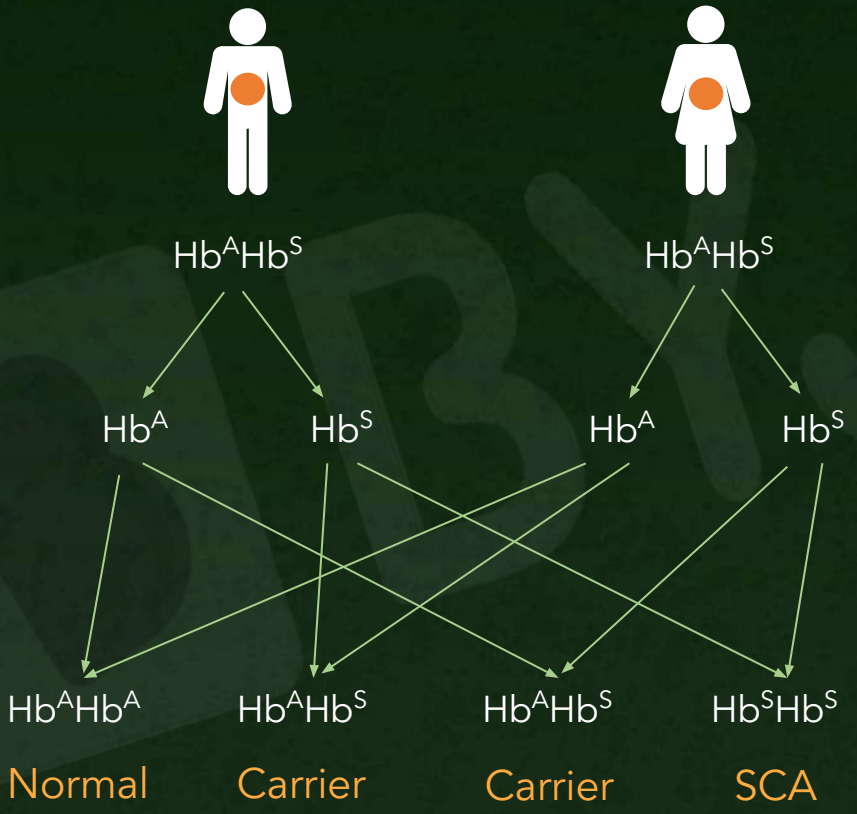
Sickle Cell Anemia (SCA)

Restricted and normal blood flow



Sickle Cell Anemia (SCA)

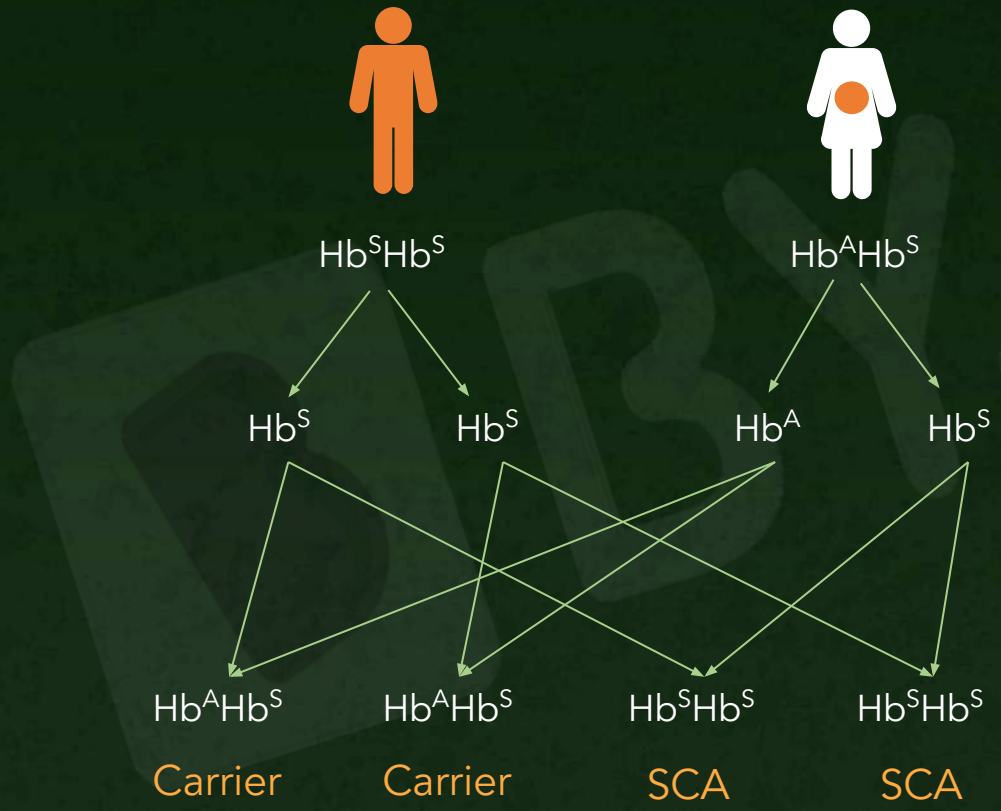
Inheritance (Carrier x Carrier)



♂ \ ♀	Hb^A	Hb^S
Hb^A	Hb^AHb^A	Hb^AHb^S
Hb^S	Hb^AHb^S	Hb^SHb^S

Sickle Cell Anemia (SCA)

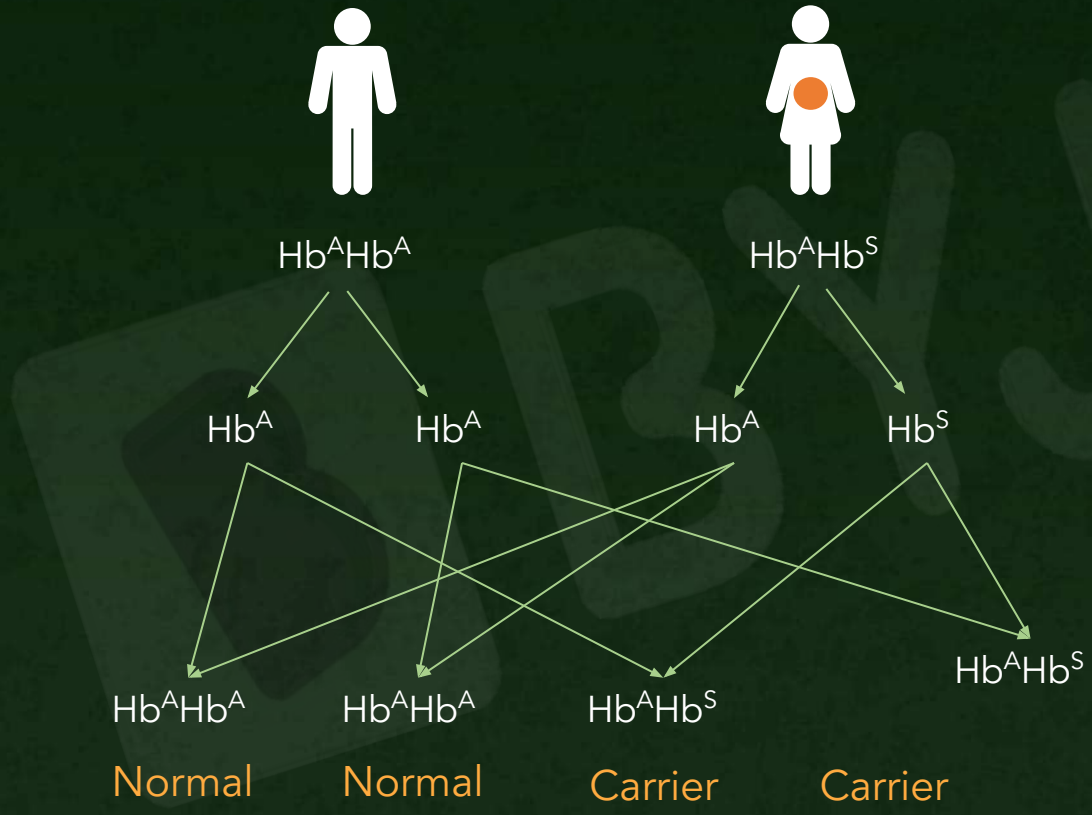
Inheritance (Affected x Carrier)

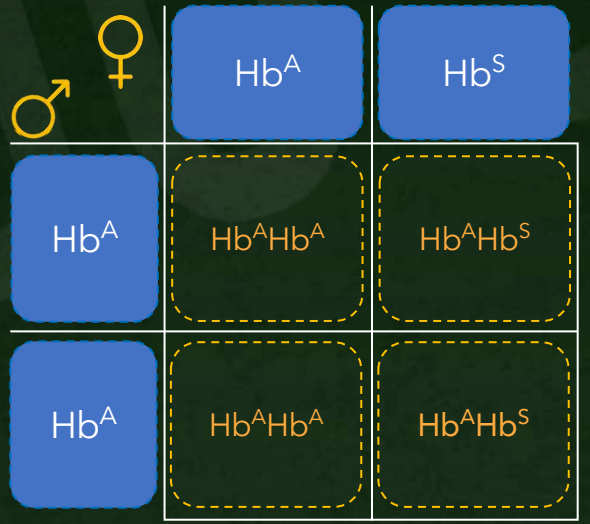


♂ \ ♀	Hb^A	Hb^S
Hb^S	Hb^AHb^S	Hb^SHb^S
Hb^S	Hb^AHb^S	Hb^SHb^S

Sickle Cell Anemia (SCA)

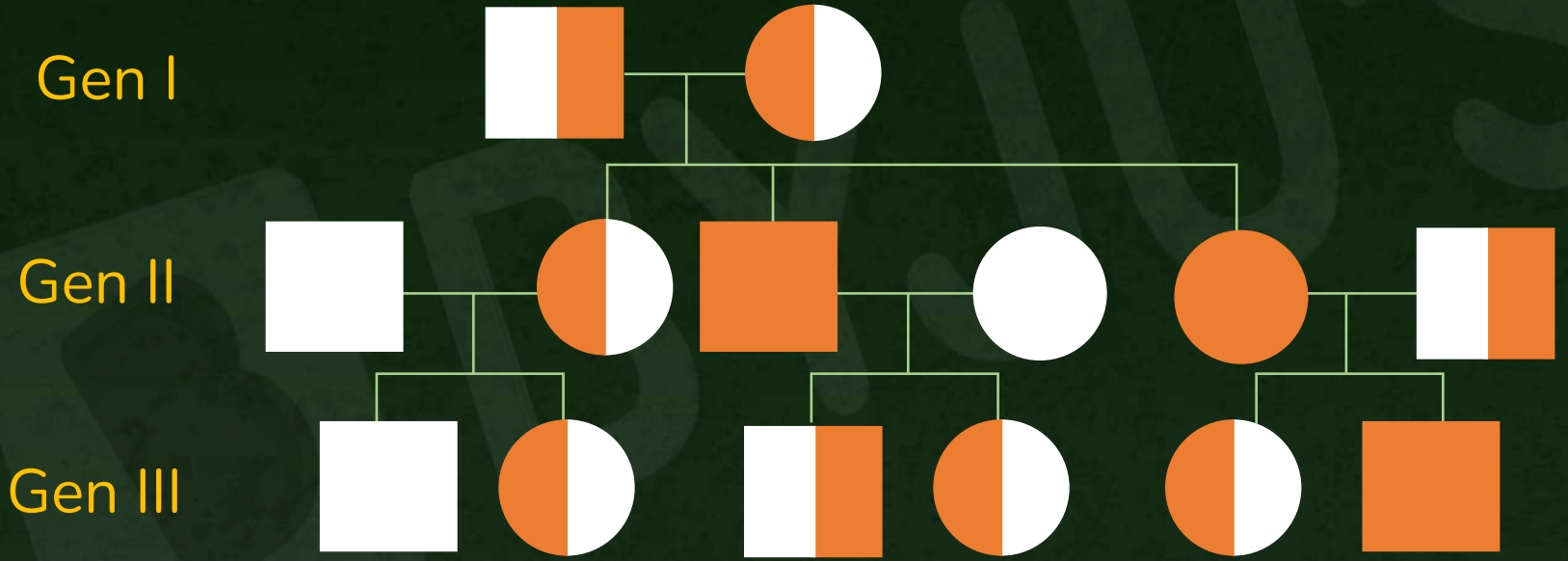
Inheritance (Unaffected x Carrier)





Inheritance Pattern in Sickle Cell Anemia

If both the parents are carriers:



Thalassemia

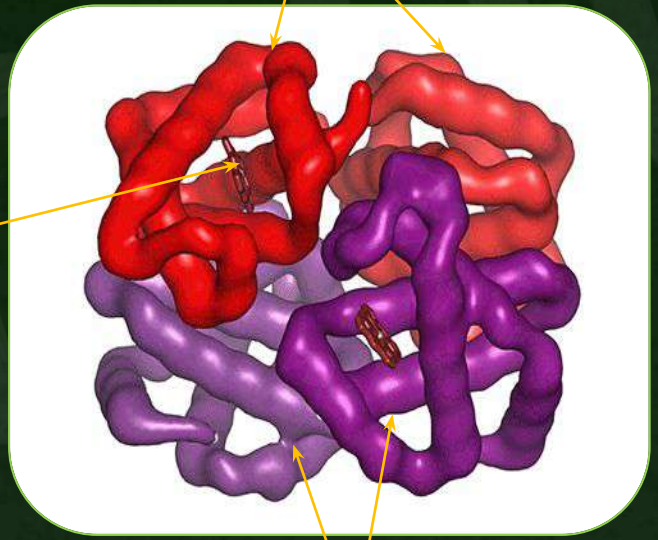
- It is a rare inherited autosomal blood disorder resulting from inadequate Hb synthesis.
- It results in the extreme destruction of red blood cells that leads to anaemia.
- If any of the parents have thalassemia, the baby is likely to develop a disease called “thalassemia minor”.
- If both the parents suffer from this disease, the child is more likely to get the disease.
- It was discovered by Cooley but the term was given by Whipple and Bradford after its prevalence in mediterranean region.
- Thalassemia is autosomal recessive blood disease which appears in children of two unaffected carriers (heterozygous parents).
- The defect can occur due to mutation or deletion of the genes controlling the formation of globin chains (commonly α and β) of haemoglobin.
- Imbalanced synthesis of globin chains of haemoglobin causes anaemia.

Thalassemia

Haemoglobin (Hb)

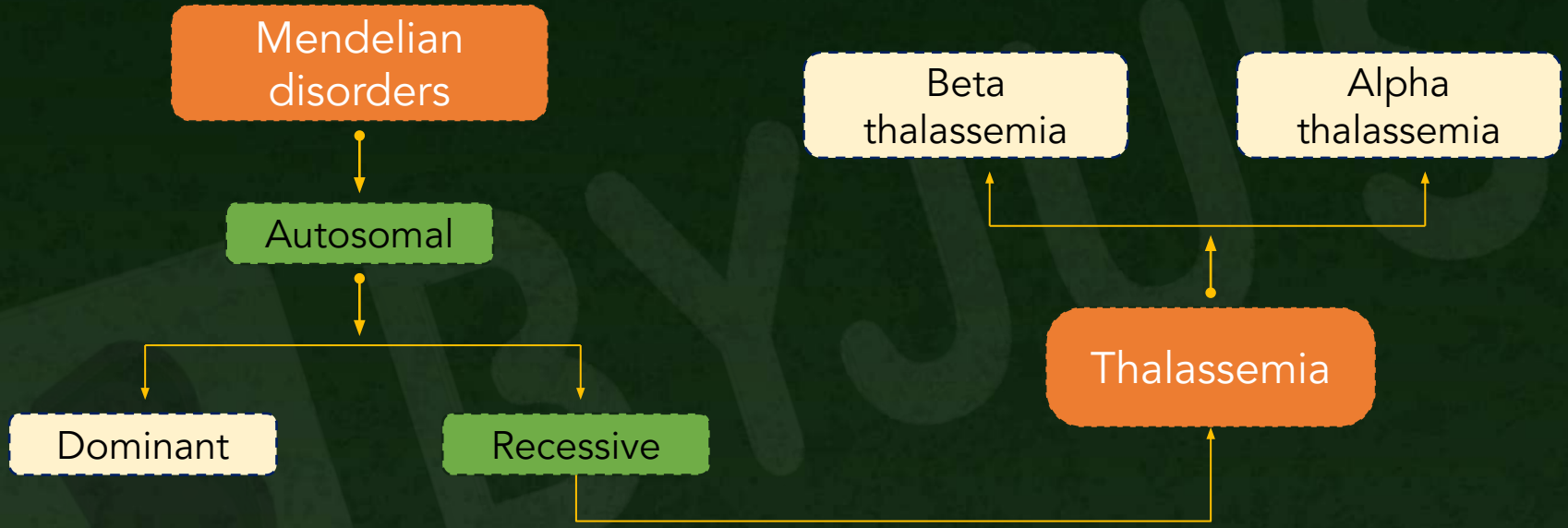
α globin chains (each with 141 amino acids)

Heme group with Fe in the centre around a porphyrin ring



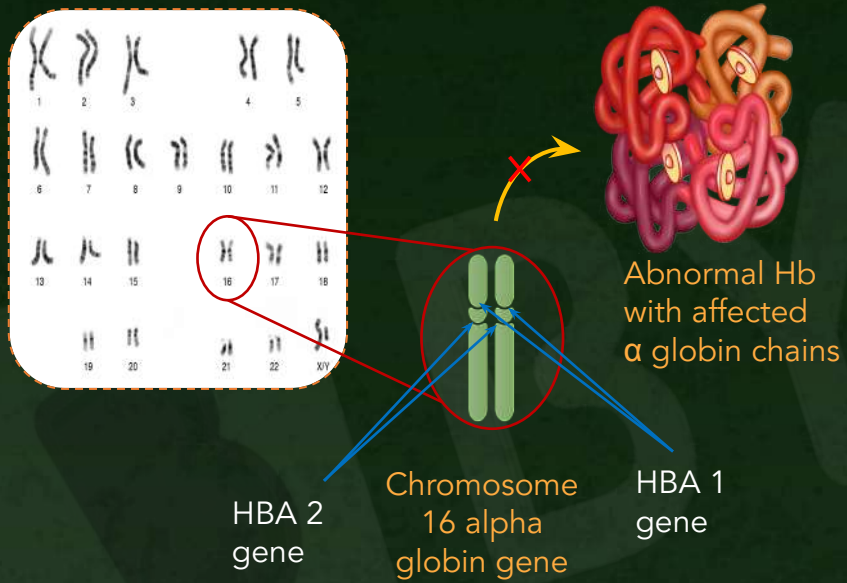
β globin chains (each with 146 amino acids)

Thalassemia



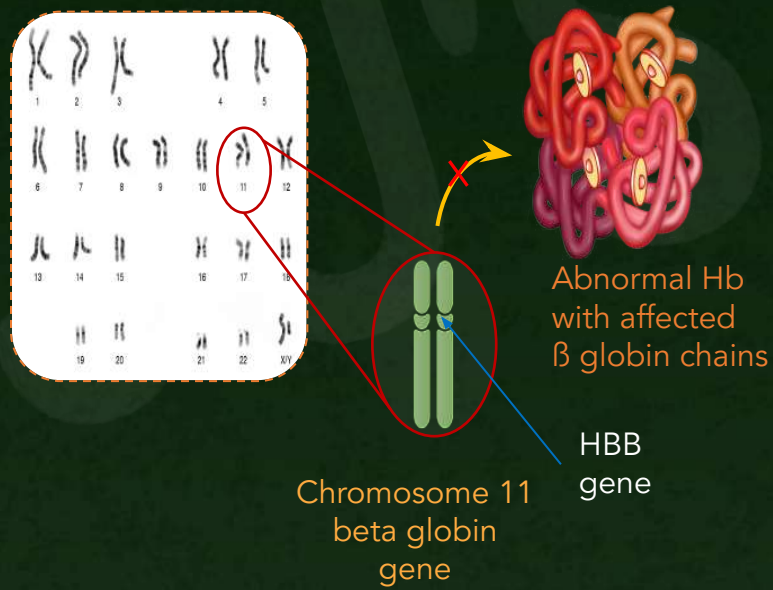
Thalassemia

Alpha thalassemia



- Alpha thalassemia is caused due to defect in 2 closely linked genes HBA1 and HBA2 on chromosome 16 of each parent.

Beta thalassemia



- Beta thalassemia is controlled by a single gene HBB on chromosome 11 of each parent and occurs due to mutation of one or both the alleles of the gene.

Thalassemia

Alpha thalassemia

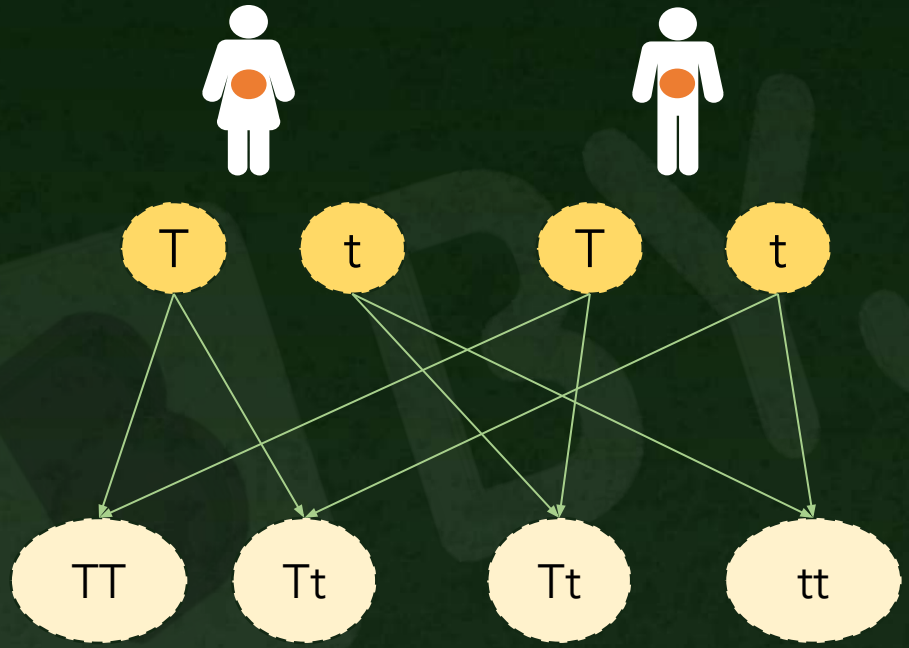
- It is caused by the defective formation of α-globin.
- It is controlled by two genes present on chromosome 16, HBA1 and HBA2 with a total of four alleles.
- Persons with one defective allele are silent carriers while two defective alleles produce α-thalassemia minor.

Beta thalassemia

- There is decreased synthesis of β-globin in this case.
- The defect is due to alleles of HBB gene present on chromosome 11.
- Persons with one defective allele suffer from thalassemia minor with larger number of microcytic erythrocytes and lesser amount of haemoglobin.
- Persons with both the defective alleles suffer from Cooley's anaemia or thalassemia major.

Thalassemia

Carrier x Carrier



♂ \ ♀	T	t
T	T T	T t
t	T t	t t

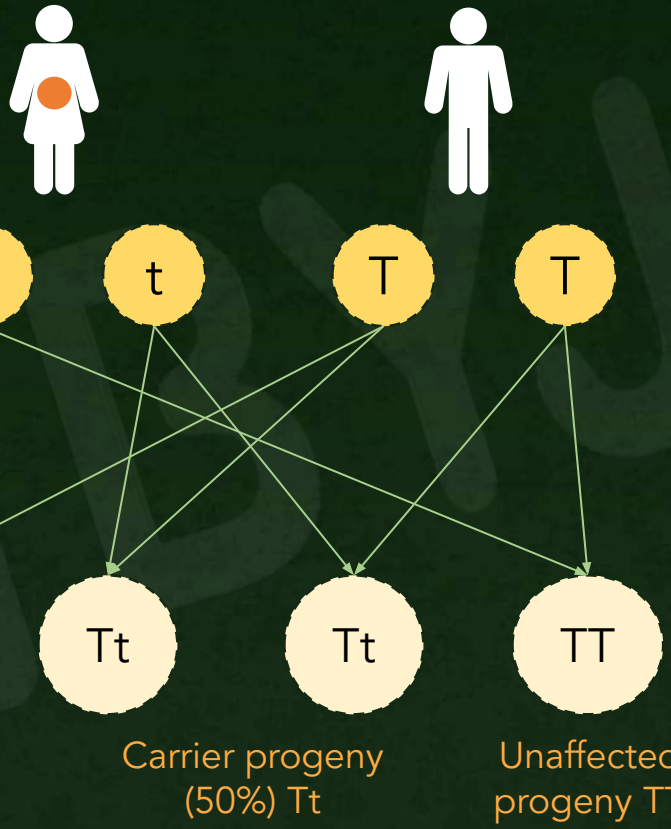
Unaffected progeny TT

Carrier progeny (50%) Tt

Affected progeny tt

Thalassemia

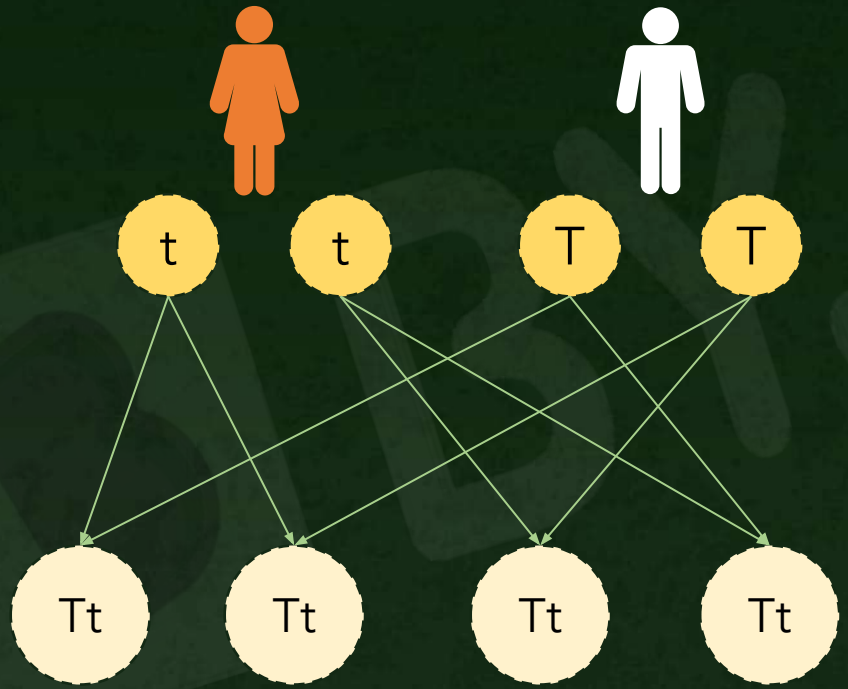
Carrier x Unaffected



♂ \ ♀	T	t
T	T T	T t
T	T T	T t

Thalassemia

Affected x Unaffected

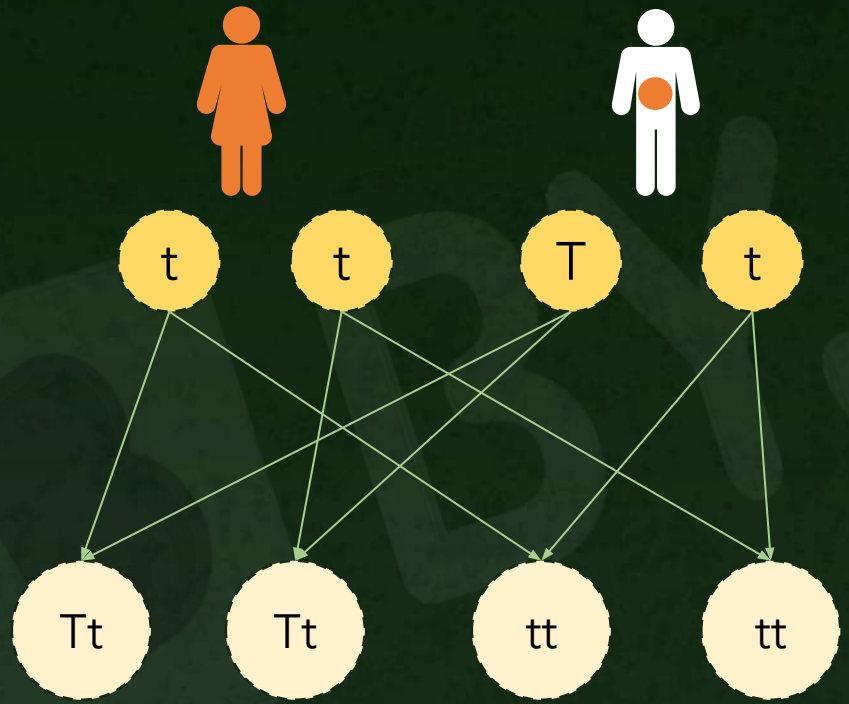


Carrier progeny
Tt

♂ \ ♀	t	t
T	T t	T t
T	T t	T t

Thalasemia

Affected x Carrier



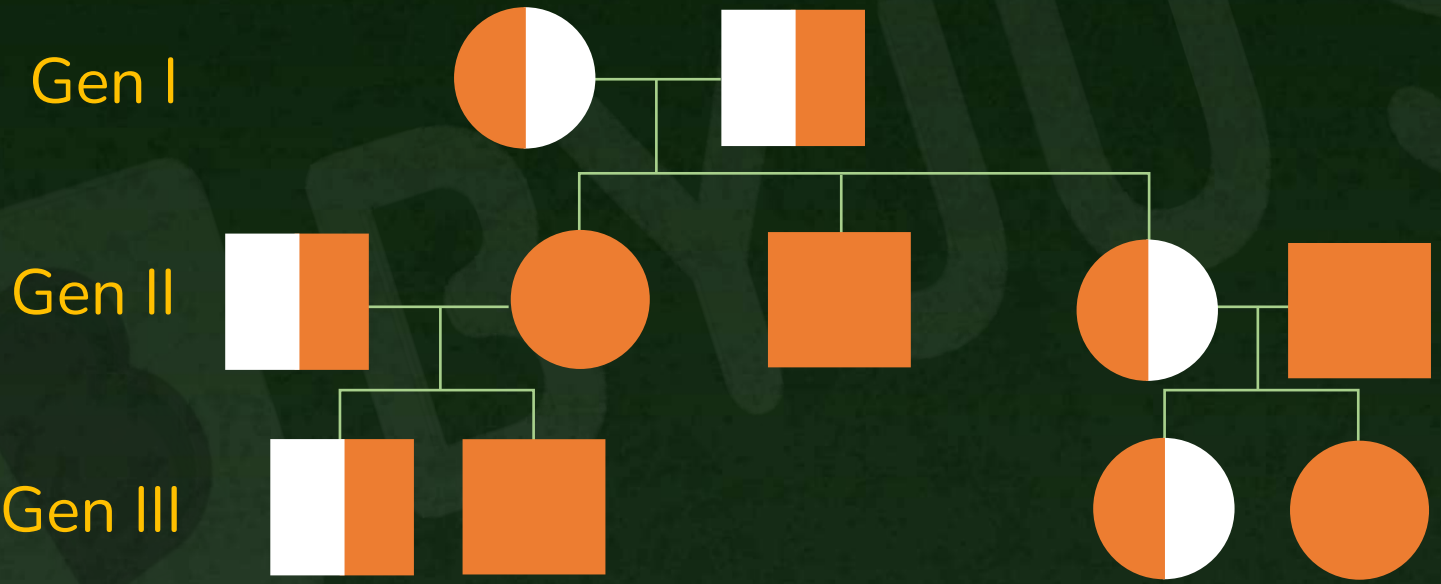
Carrier progeny
Tt

Affected progeny
tt

♂ \ ♀	t	t
T	T t	T t
t	t t	t t

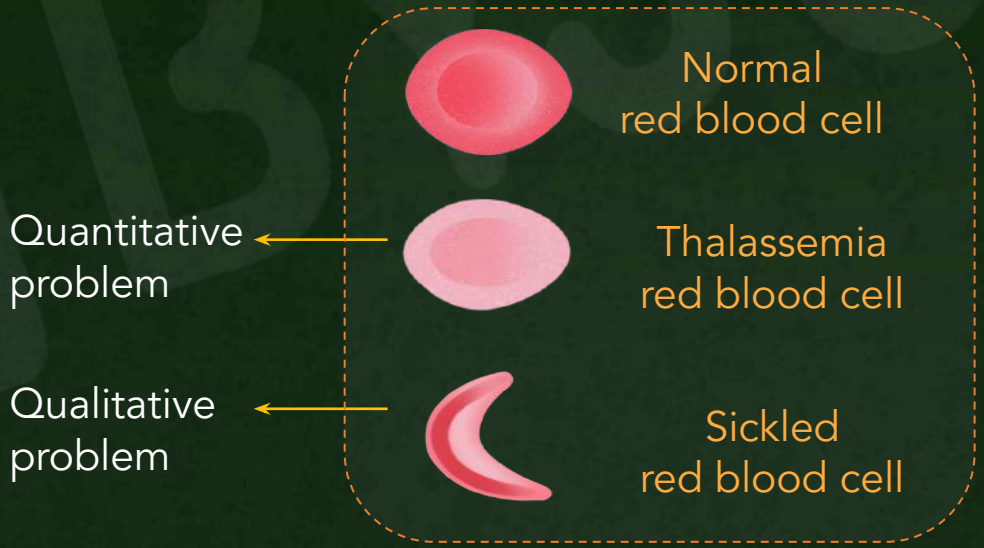
Inheritance Pattern in Thalassemia

If both the parents are carriers:



Abnormal forms of RBCs

- Thalassemia
  - A quantitative problem of synthesizing too few globin molecules.
- Sickle cell anemia
  - A qualitative problem of synthesising an incorrectly functioning globin.

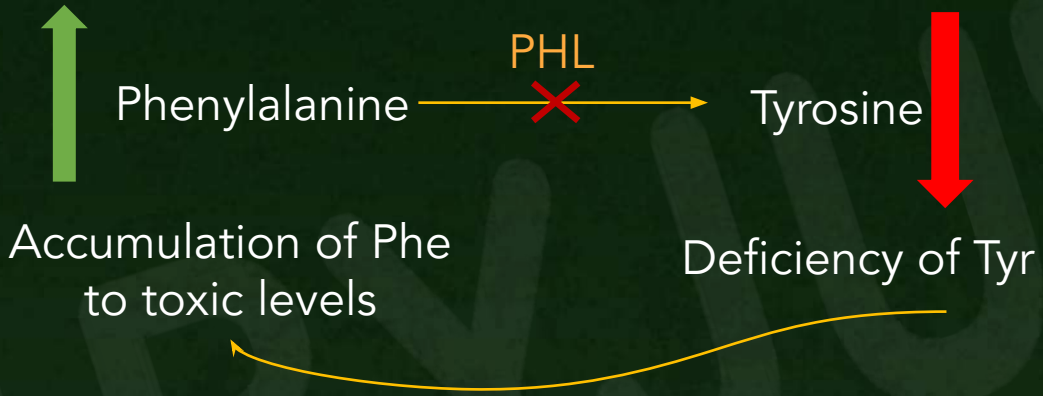


Phenylketonuria(PKU)

- It is an autosomal recessive metabolic disorder resulting from lack of an enzyme known as phenylalanine hydroxylase (PHL).
- PHL helps to convert phenylalanine into tyrosine.

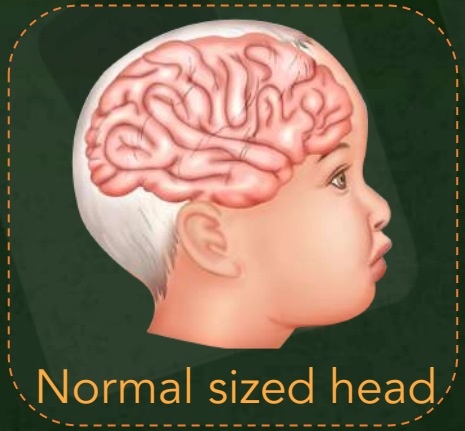
PHL - Phenylalanine hydroxylase

Phenylketonuria (PKU)



- Phenylketonuria results in accumulation of phenylalanine to toxic levels and deficiency of tyrosine.
- Tyrosine is required for synthesis of various neurotransmitters.
- Tyrosine deficiency results in reduced brain development and other neurological conditions.
- Excess presence of Phe(Phenylalanine) in cerebrospinal fluid (CSF) causes mental retardation, intellectual disability and mental disorders.

Phenylketonuria (PKU)

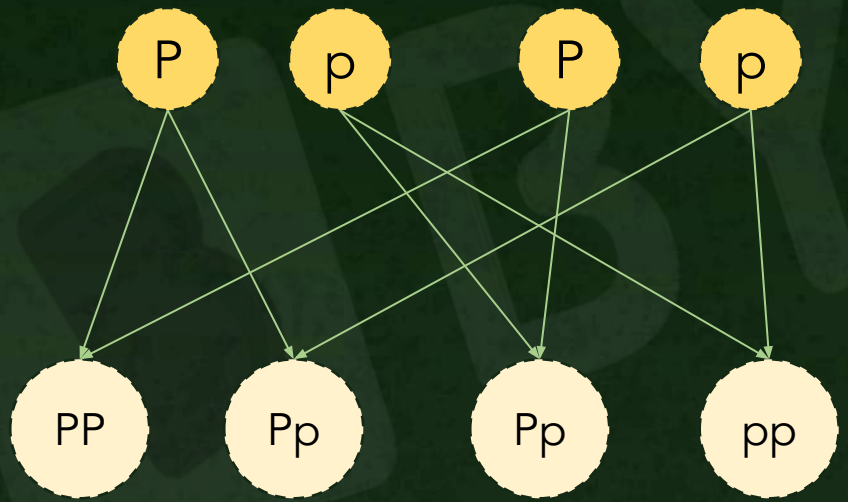


Symptoms

- Small sized head (microcephaly)
- Learning disability and delayed development
- Neurological disorders
- Behavioural, emotional and social problems
- Fatigue

Phenylketonuria(PKU)

Carrier x Carrier



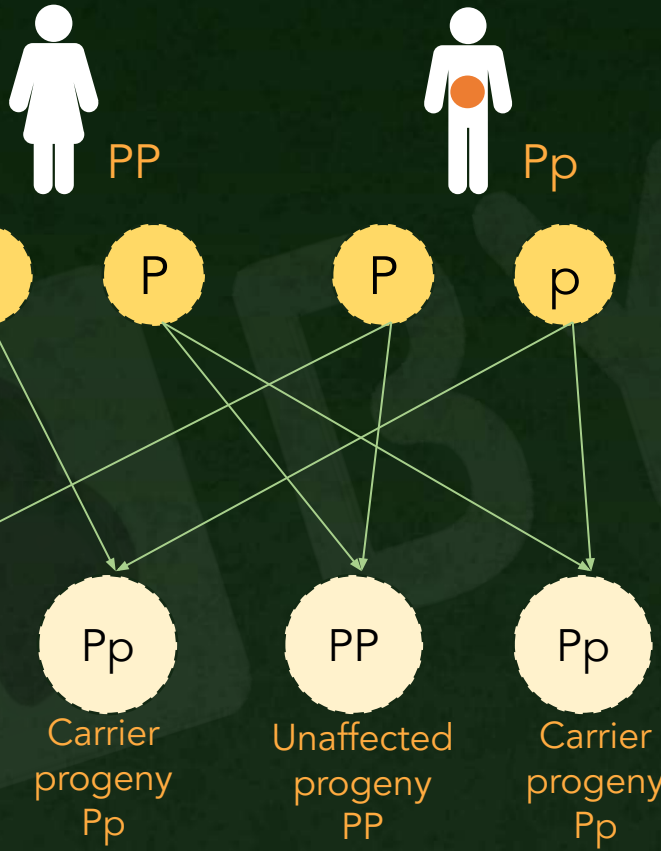
Unaffected progeny PP

Carrier Progeny (50%) Pp

Affected progeny pp

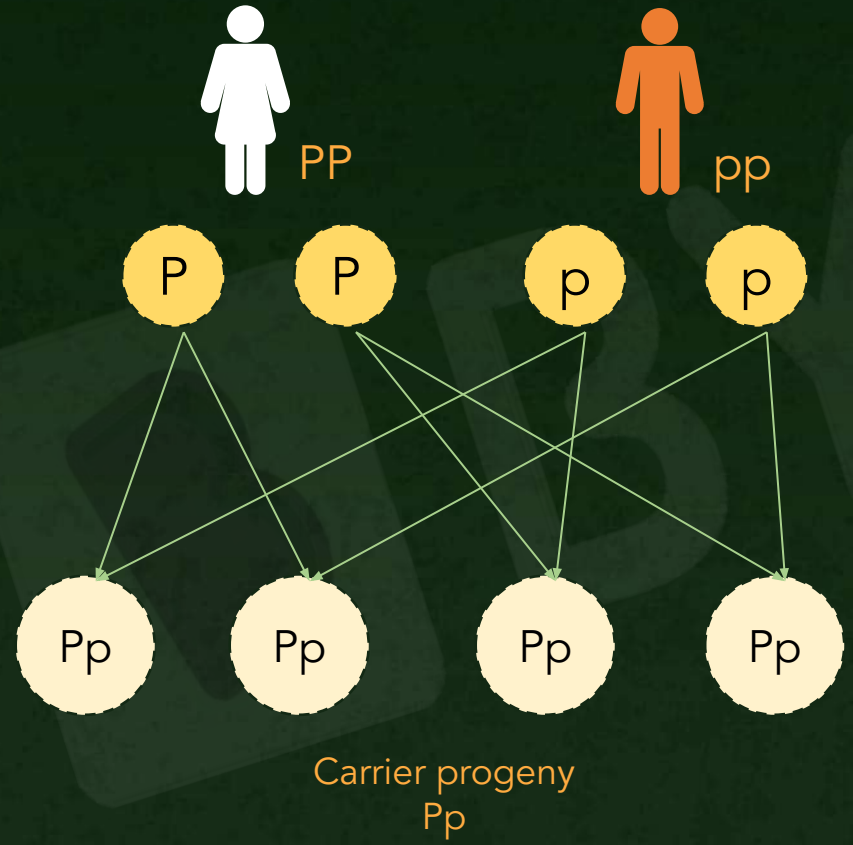
Phenylketonuria (PKU)

Unaffected x Carrier



Phenylketonuria (PKU)

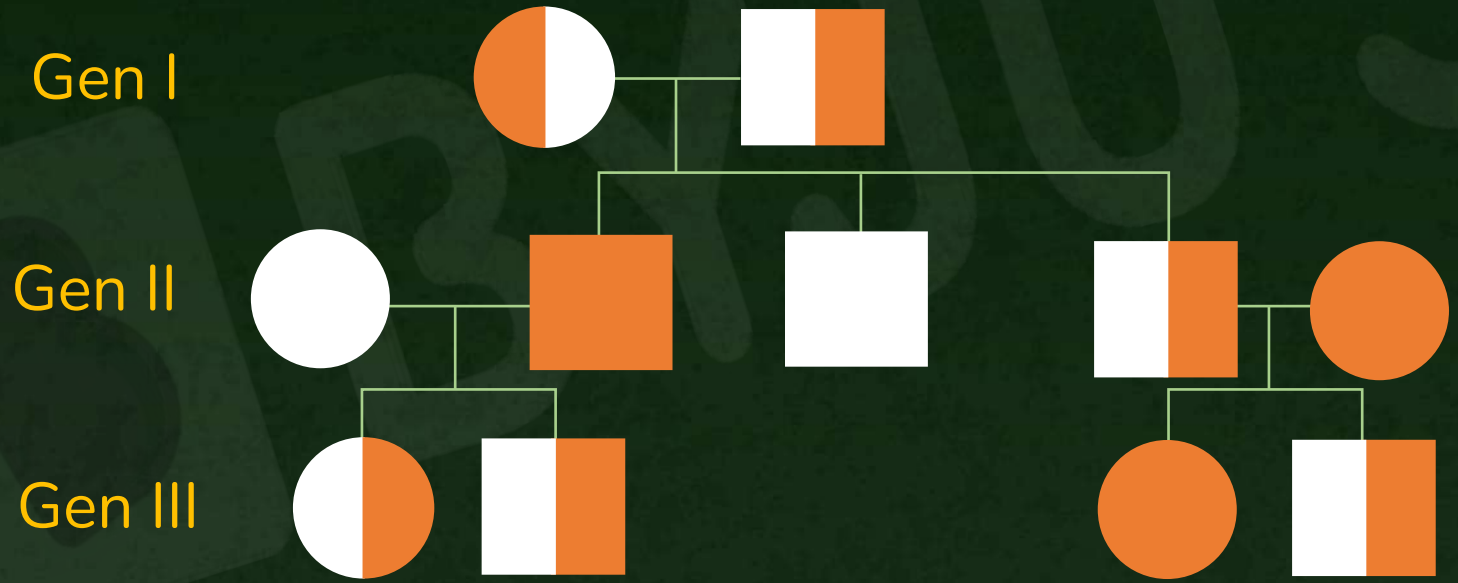
Unaffected x Affected



♀ \ ♂	p	p
P	P p	P p
P	P p	P p

Inheritance Pattern in Phenylketonuria

If both the parents are carriers:



Summary

- Pedigree analysis
  - It is the study of a particular trait that is inherited from one generation to another.
  - It helps to understand the pattern of inheritance for a particular trait, and also to know whether the trait is dominant or recessive.

Summary

Understanding the symbols

Unaffected/Normal male

Unaffected/Normal female

Sex unspecified

Affected female

Affected male

Carrier female

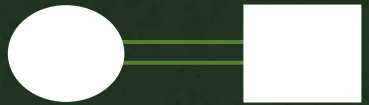
Carrier male

Carrier female

Carrier male

Deceased female and male

Marriage

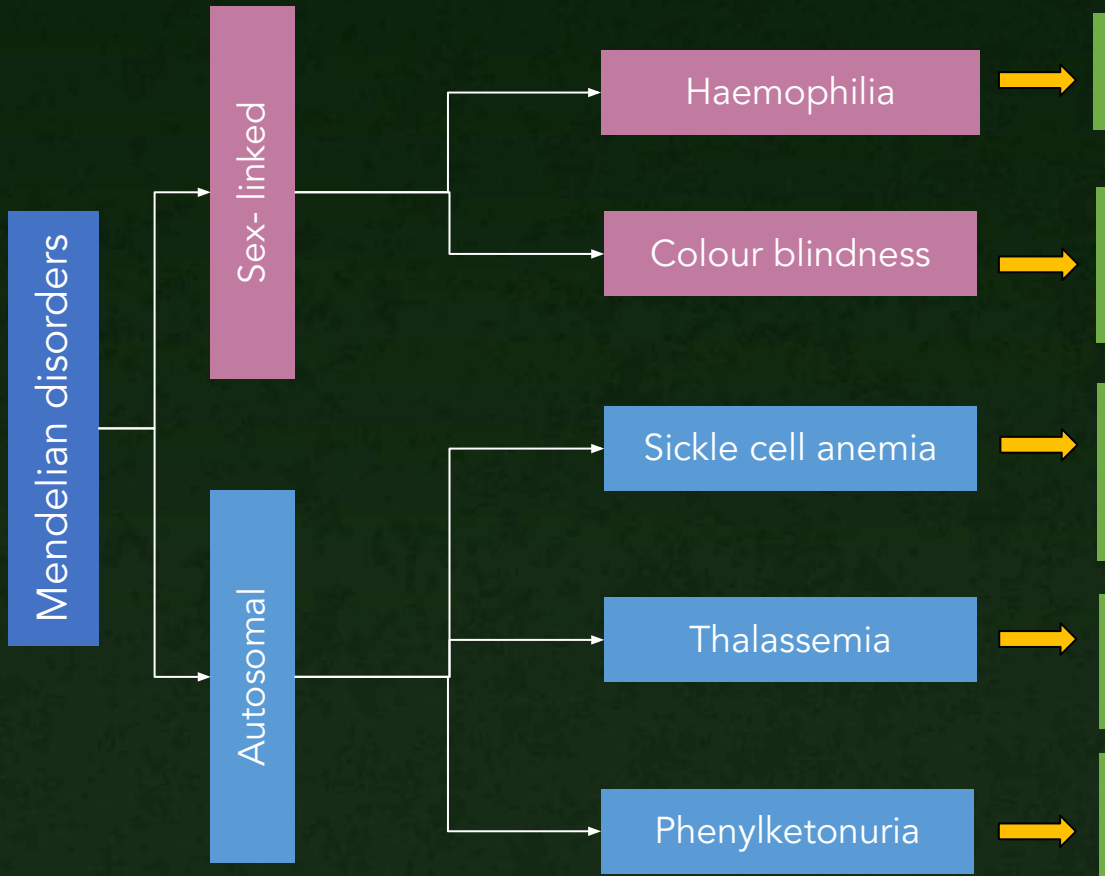


Consanguineous marriage

Fraternal twins (non-identical)

Identical twins

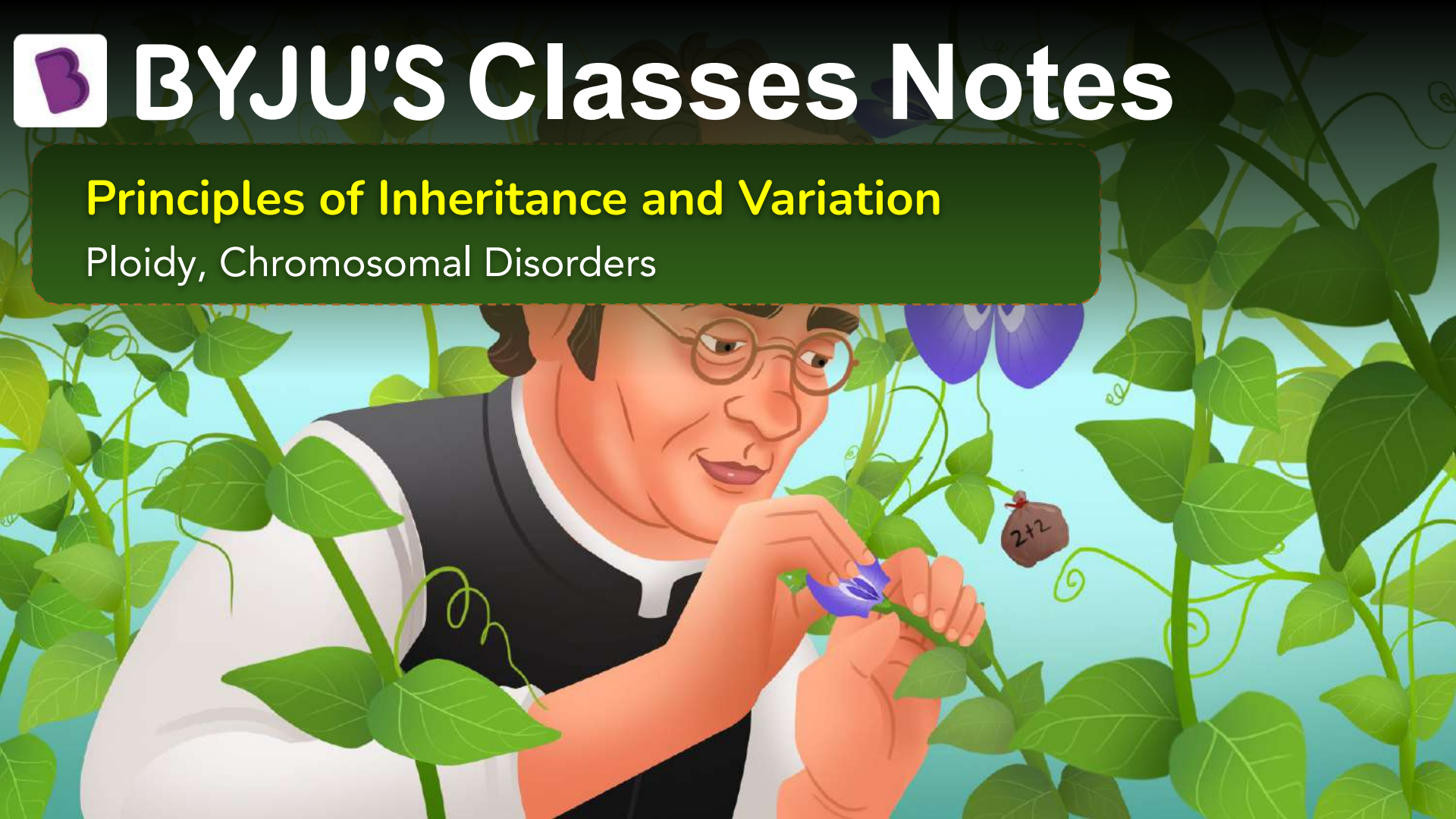
Summary



BYJU'S Classes Notes

Principles of Inheritance and Variation

Ploidy, Chromosomal Disorders



Key Takeaways

Ploidy

1

Euploidy

Aneuploidy

2

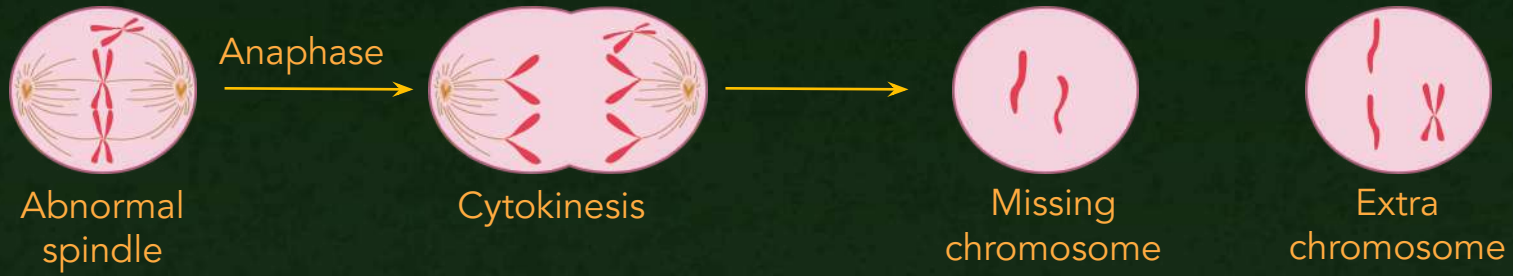
Chromosomal disorders

Types of disorders

Summary

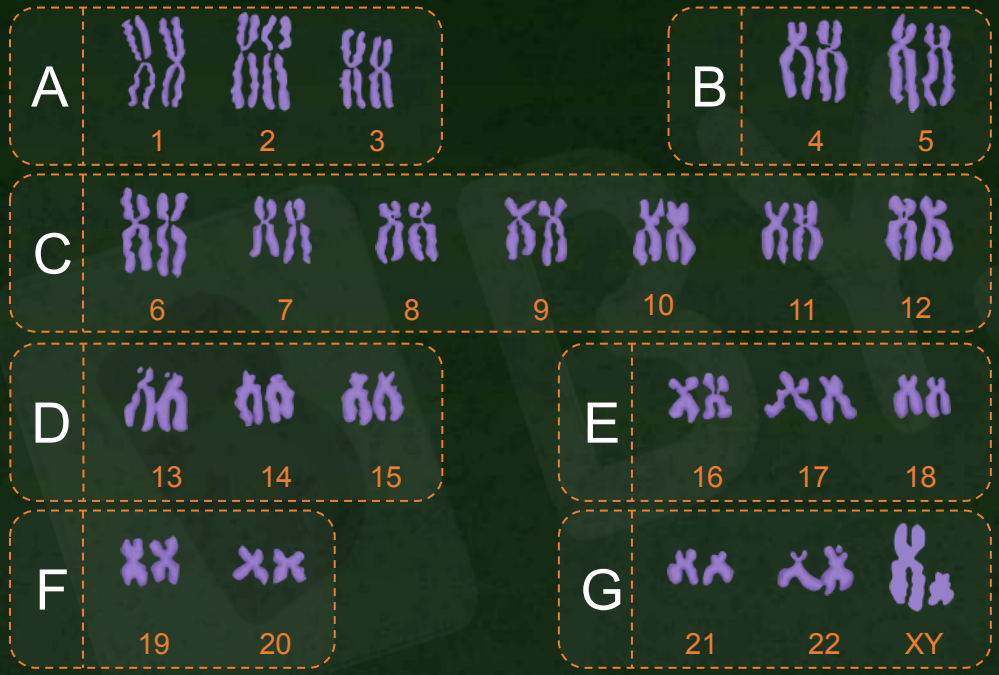
Chromosomal Aberrations

- Errors in replication can result from improper distribution of chromosomes during cell division. This occurrence is called as non-disjunction of chromosomes.
- This changes the ploidy of the cell.



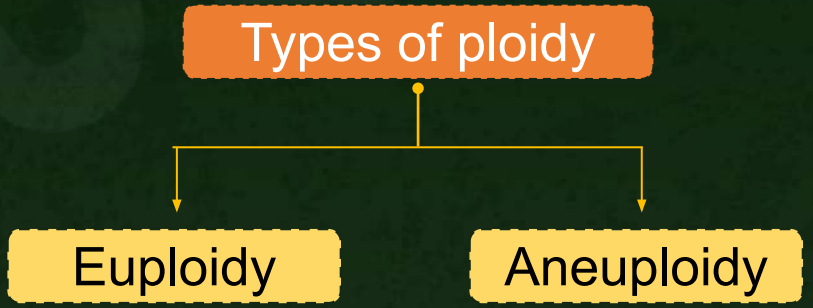
Ploidy

It is the number of chromosomes or sets of chromosomes present in a cell.



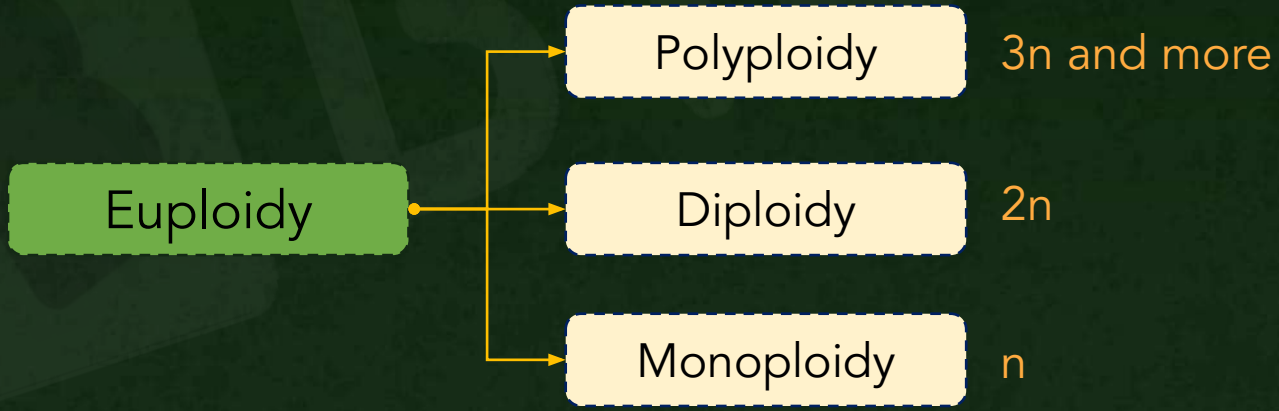
Human karyotype – Male

The ploidy of human body cell is diploidy.



Euploidy

- A single set of chromosomes in a cell is also known as basic set.
- Euploidy is a condition in which chromosomes in the cell are in multiples.
- Based on how many sets of chromosomes are present, there are different types of euploidy and it is denoted as n, 2n, 3n and so on.



Types of Euploidy

Monoploidy

- The condition in which the cell has a single set of chromosome.
- The cell or organism is called monoploid.
- Denoted as : n

Diploidy

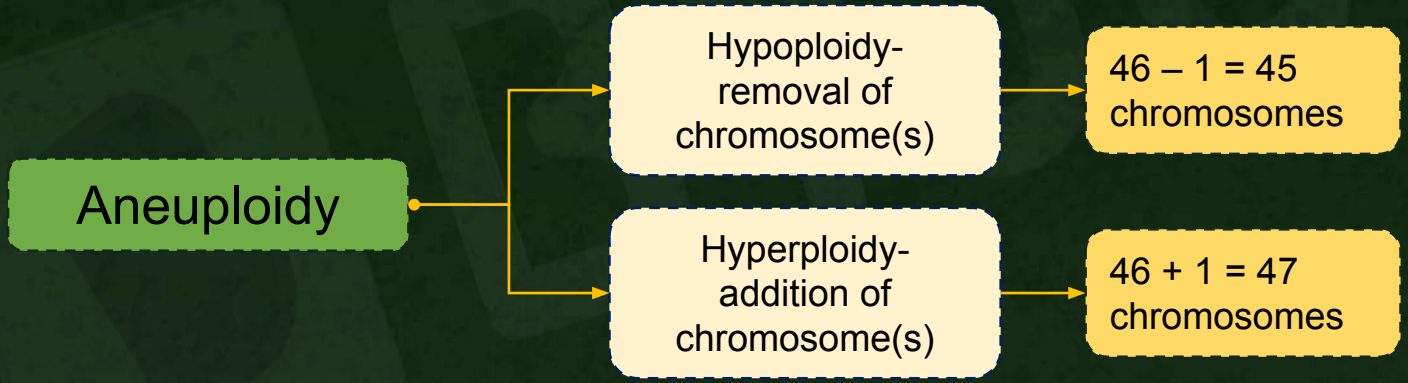
- The condition in which the cell has two sets of chromosomes.
- The cell or organism is called diploid.
- Denoted as : 2n

Polyploidy

- The condition in which the cell has multiple sets of chromosomes.
- The cell or organism is called polyploid.
- Denoted as : 3n, 4n, etc.

Aneuploidy

- In the case of aneuploidy, there is addition or removal of chromosomes and not a set of chromosome.
- Example - Klinefelter syndrome in human beings.



Types of Aneuploidy

Hypoploidy [removal of chromosome(s)]	
No. of chromosomes removed	Type of hypoploidy
- 1	Monosomy (2n – 1)
- 2	Nullisomy (2n – 2)

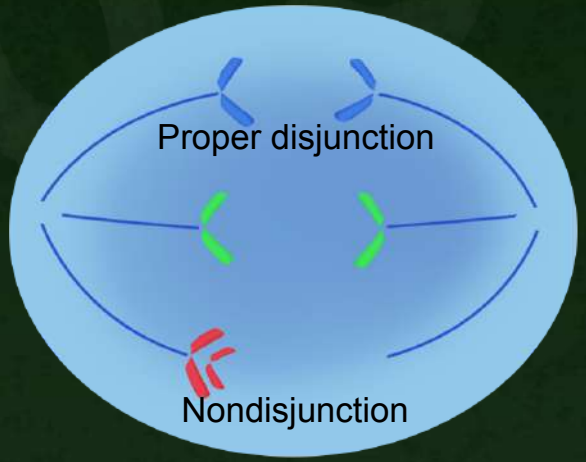
- If one chromosome is removed it is called monosomy.
- If two chromosomes are removed it is called nullisomy.

Hyperploidy [addition of chromosome(s)]	
No. of chromosomes added	Type of hyperploidy
+ 1	Trisomy (2n + 1)
+ 2	Tetrasomy (2n + 2)

- If one chromosome is added, then the type of hyperploidy is trisomy.
- If two chromosomes are added, then the type of hyperploidy is tetrasomy.

Chromosomal Disorders

- Abnormal number of chromosomes in human cells results in chromosomal disorders.
- Caused by either addition or removal of chromosome(s).
- Can occur due to nondisjunction of chromosomes (unable to split properly) during cell division.
- This error occurs during the anaphase of the cell division.



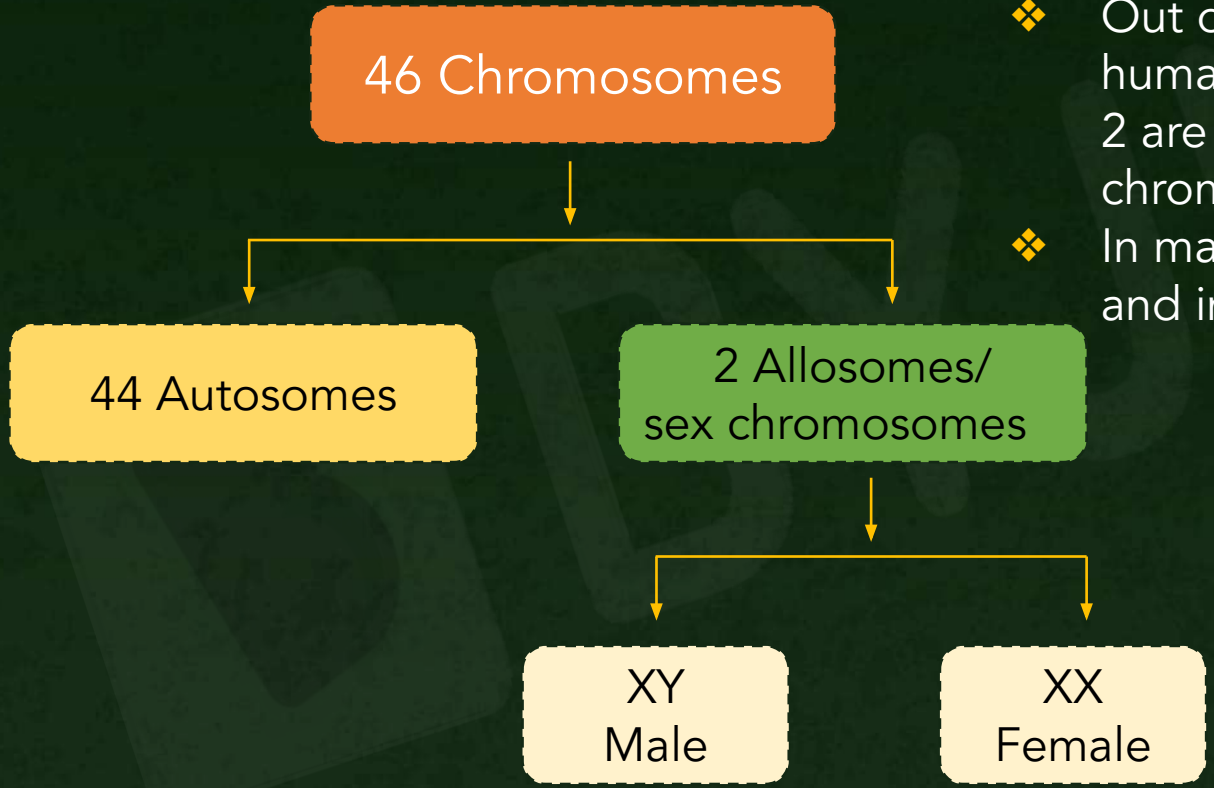
Anaphase

Types of Chromosomal Disorders

Type of aneuploidy	Nullisomy- removal of two chromosomes (2n-2)
Total number of chromosomes	44
Disorders	Human beings with nullisomy condition don't survive

Type of aneuploidy	Monosomy removal of one chromosome (2n-1)
Total number of chromosomes	45
Disorders	Turner's syndrome, Cri-du-chat syndrome

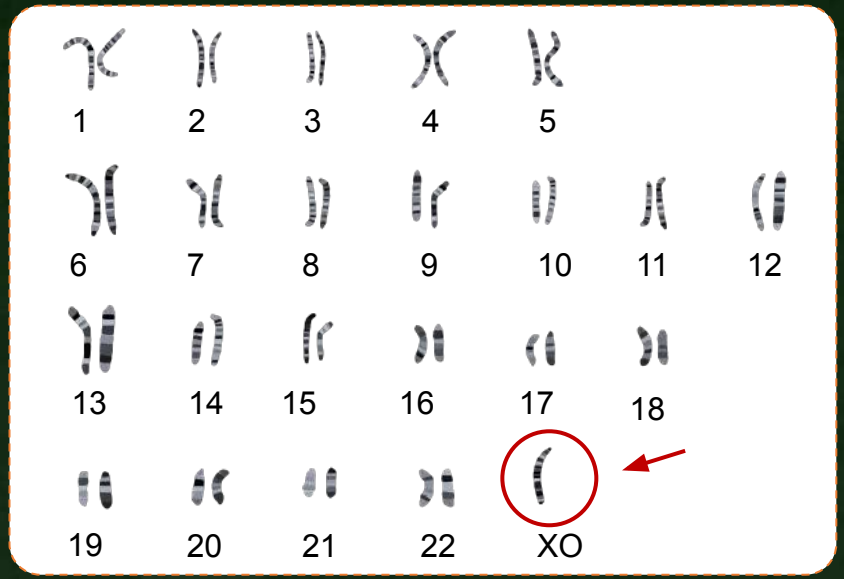
Recall! Autosomes Vs Sex Chromosomes



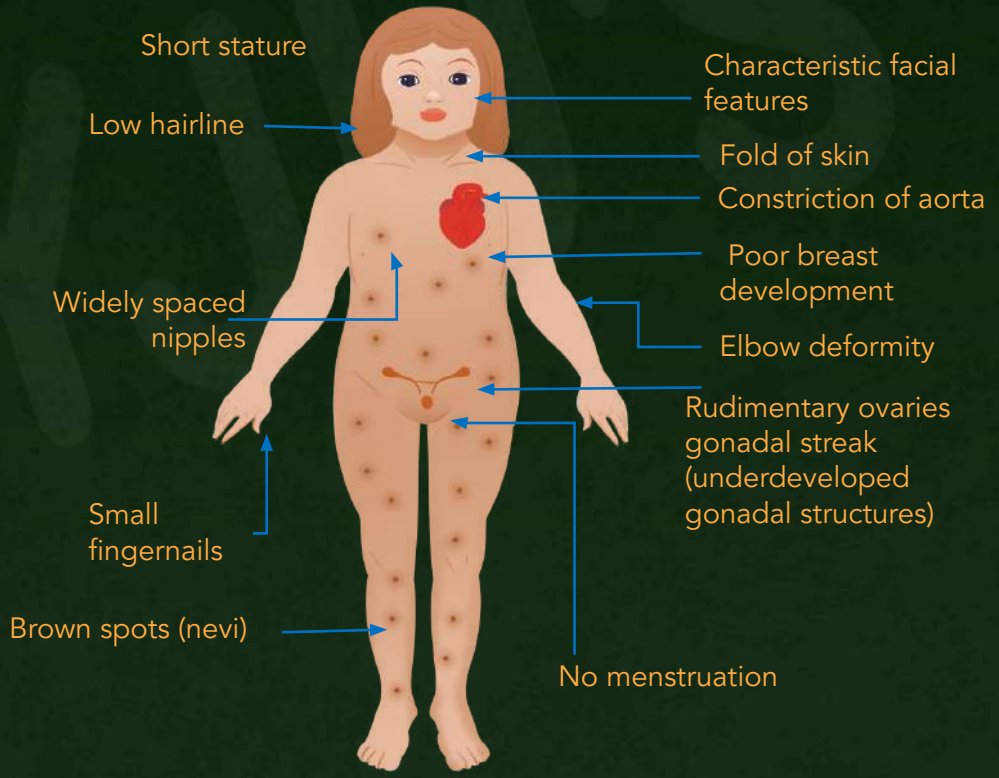
- Out of the 46 chromosomes that humans have, 44 are autosomes and 2 are allosomes or sex chromosomes.
- In males, sex chromosomes are XY and in females they are XX.

Turner's Syndrome

- Removal of one X chromosome from the sex chromosomes
- Occurs in females
- Chromosome composition: 44A+XO

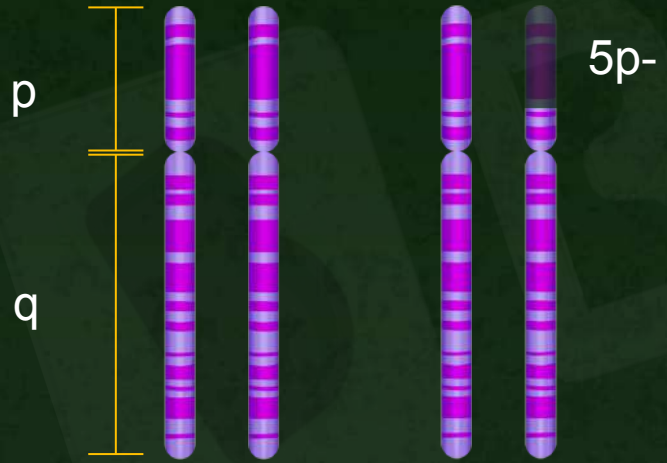


Turner's syndrome karyotype



Cri-du-chat Syndrome

- Infant's cry is high pitched
- Sounds like a cat's cry



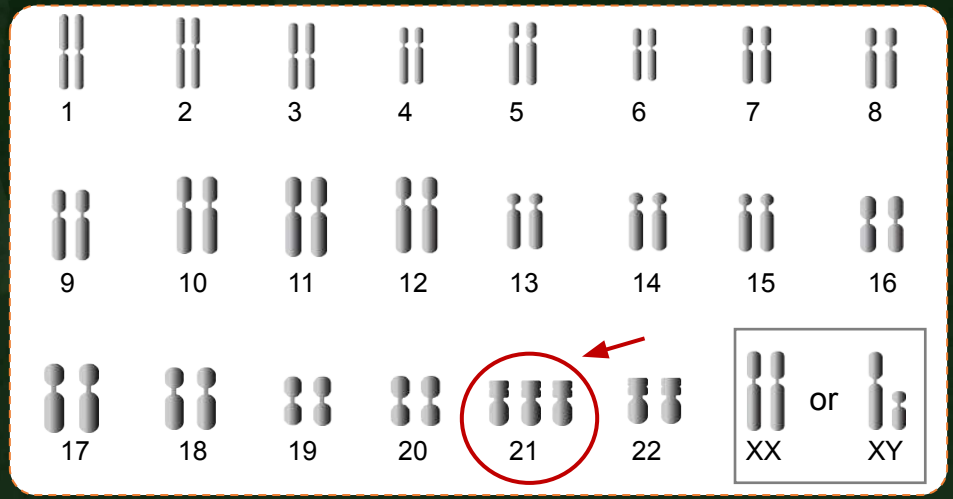
5p-syndrome =
Cri-du-chat syndrome

- It caused due to the deletion of a part of the 5th chromosome.
- Since only a part of chromosome is deleted and not the full chromosome, this type of aneuploidy is called partial monosomy.

Down's Syndrome

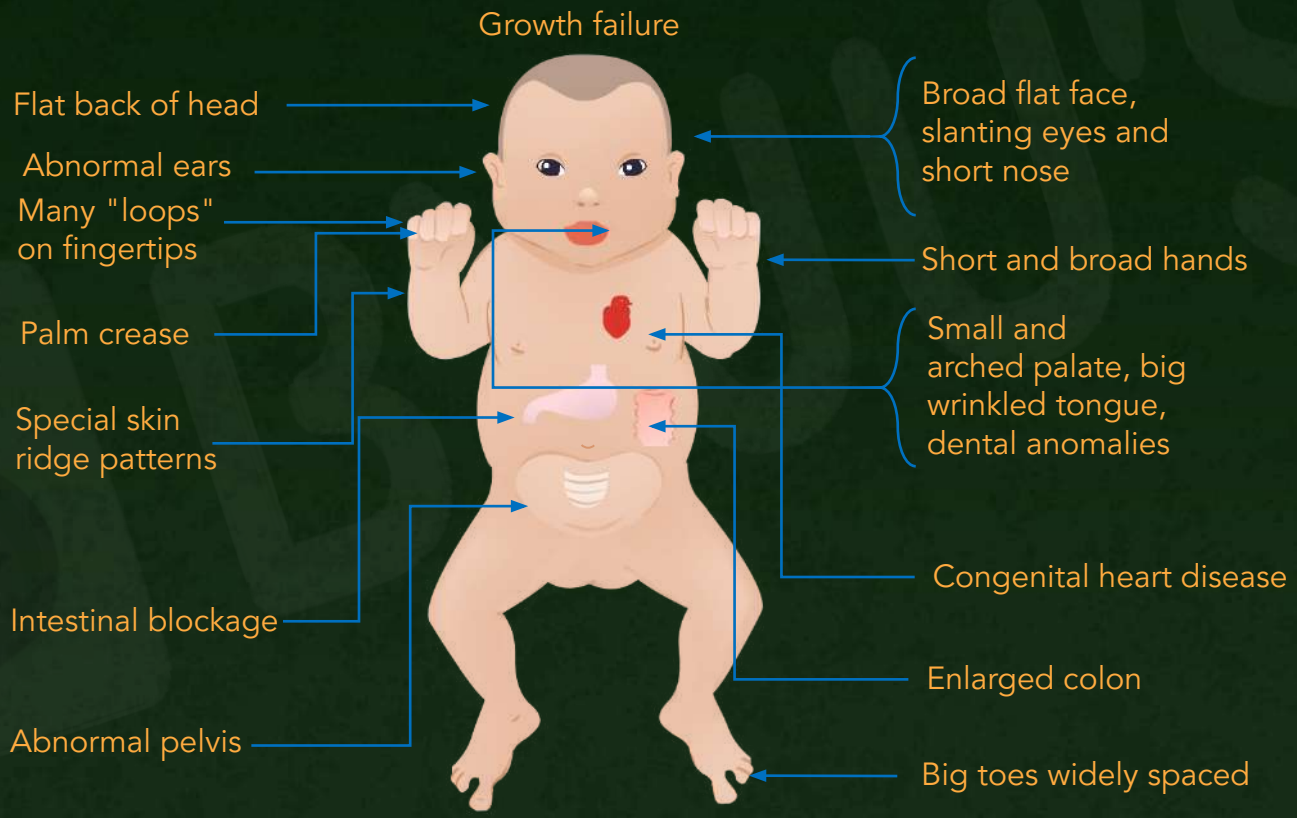
- Trisomy of 21st chromosome
- Occurs both in females and males

Type of aneuploidy	Trisomy- Addition of one chromosome (2n+1)
Total number of chromosomes	47



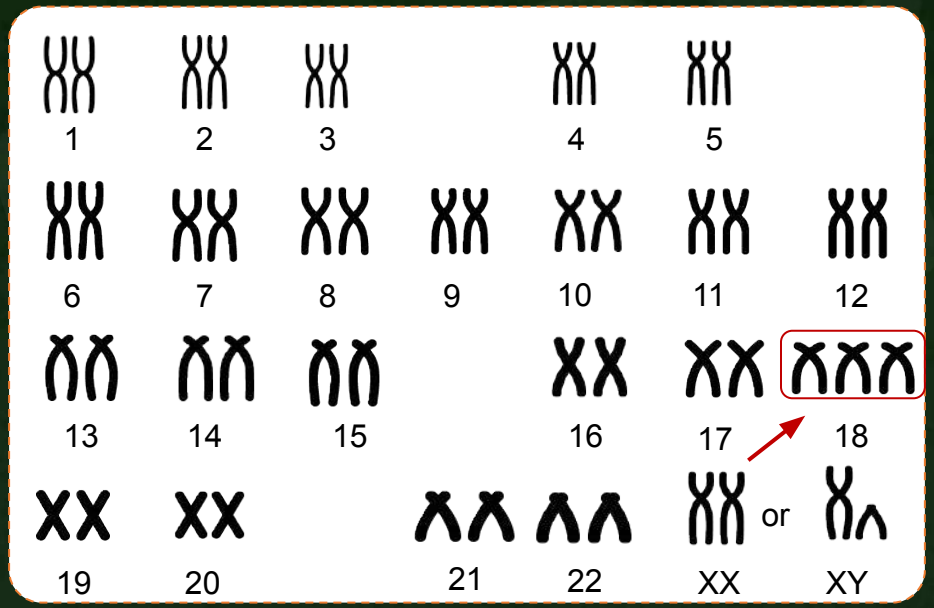
An extra chromosome in the 21st chromosome results in Down's syndrome

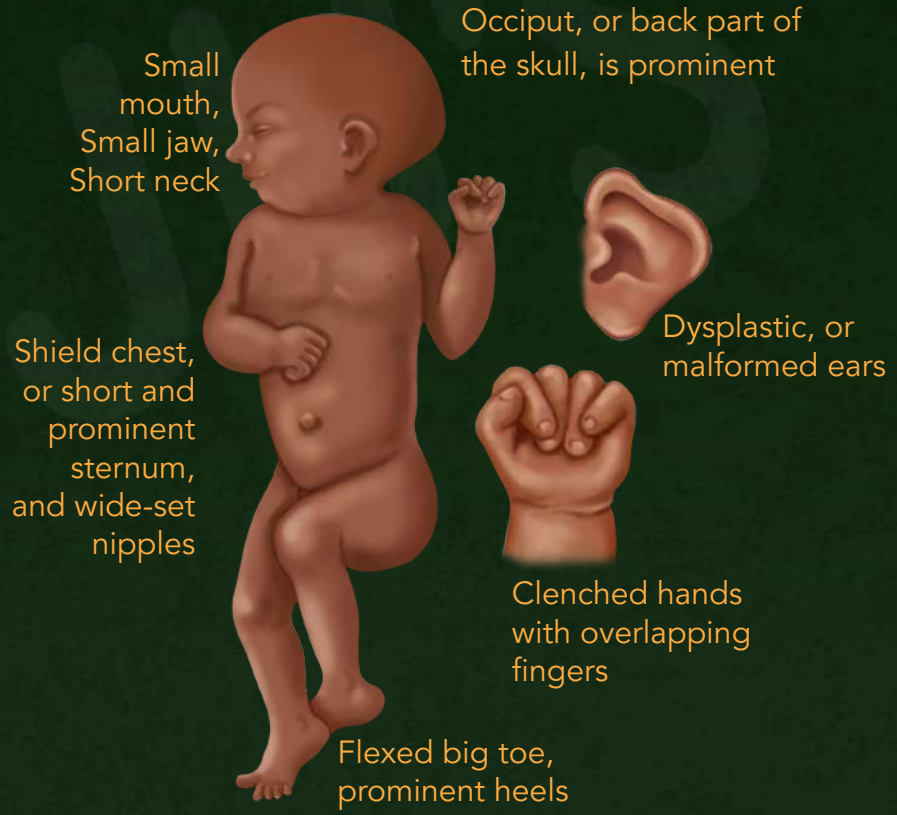
Down's Syndrome



Edward's Syndrome

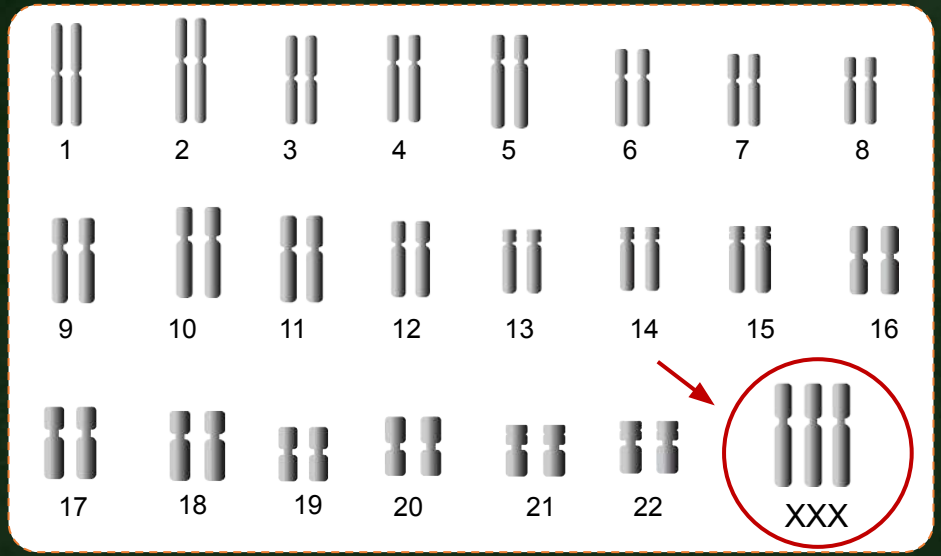
- Trisomy of 18th chromosome
- Occurs in both males and females





Triple X Syndrome/Super Female

- Extra X chromosome in sex chromosomes
- Takes place only in females
- Chromosomal composition: 44A+XXX

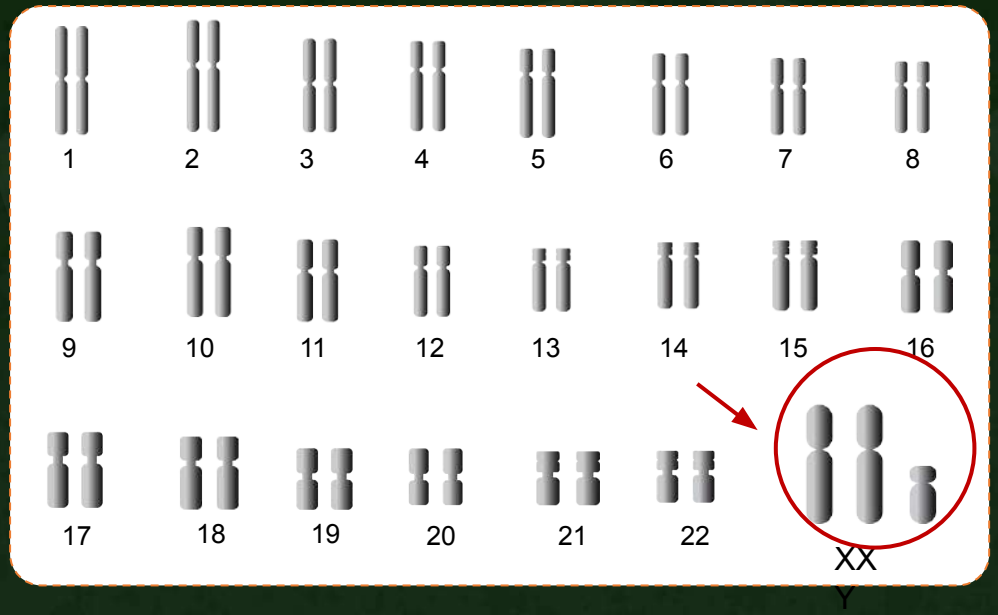


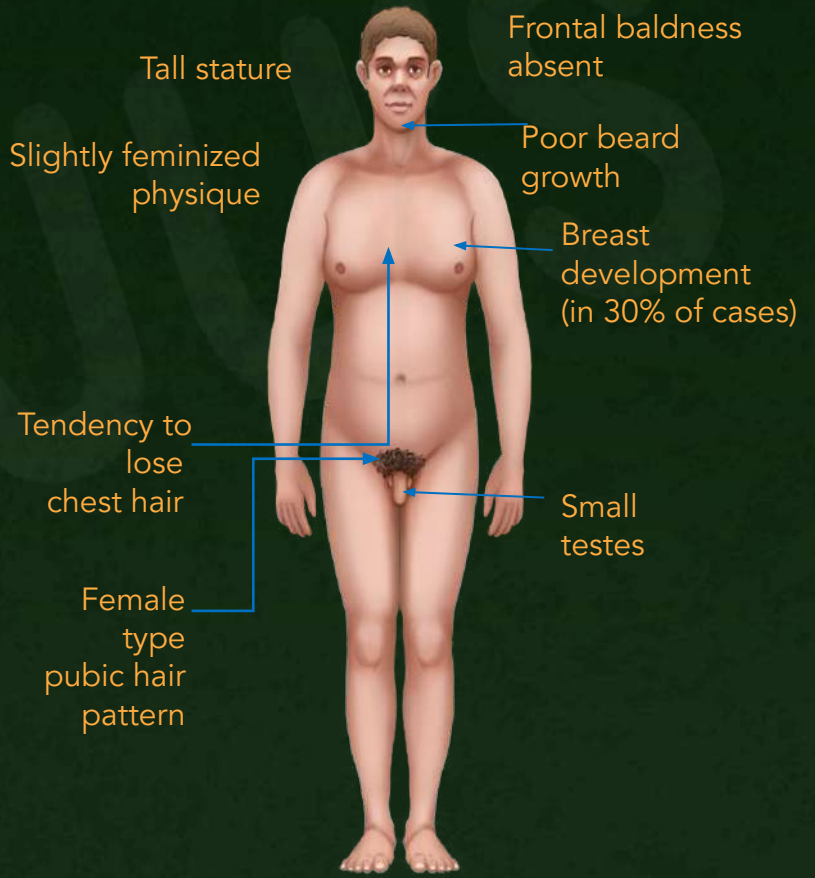
Features

- Delayed motor development
- Delayed speech
- Low IQ
- Wide set eyes

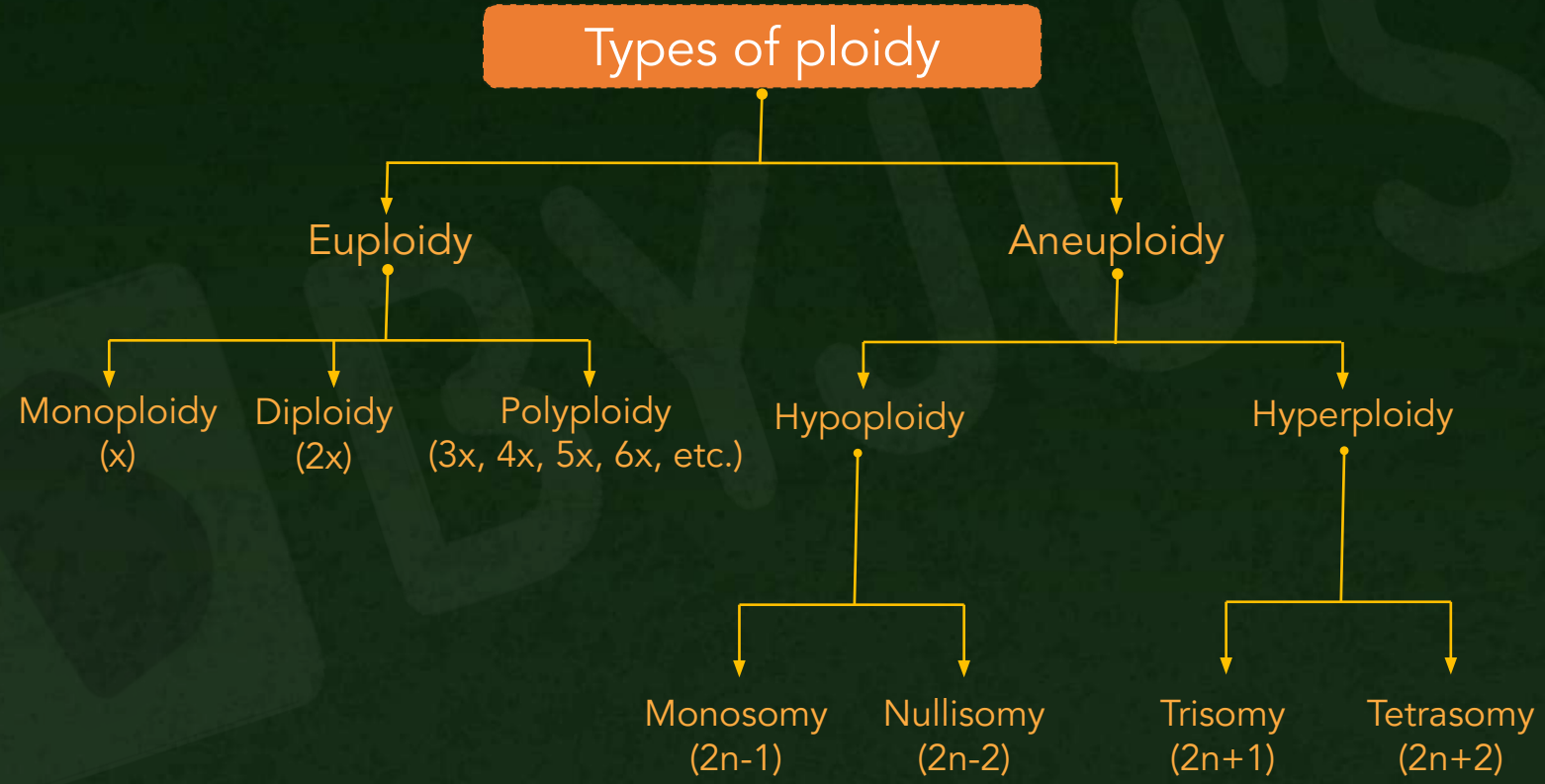
Klinefelter's Syndrome

- Extra X chromosome in sex chromosomes
- Occurs only in males
- Chromosomal composition: 44A+XXY





Summary



Summary

Chromosomal disorders

Turner's syndrome

Affected female lacks one X chromosome

Down's syndrome

Affected female has an extra X chromosome

Cri-du-chat syndrome

Affected infants have high pitched cry

Edward's syndrome

Trisomy of 18^{th} chromosome

Triple X syndrome

Affected female has an extra X chromosome

Klinefelter's syndrome

Affected male has an extra X chromosome