LECTURE 1

Introduction to Mendelian Genetics

What is Genetics?

Genetics is the study of genes, genetic variation, and heredity in organisms

Father of Genetics

Born: Czech Republic

Study: Vienna

Research: Bruno Monastery
(1856-1863)

Concept of Dominance ✓
Law of Segregation ✓
Law of Independent Assortment ✓



How Mendelism (1865) was different from others ?

1. Particulate Inheritance :

Character ⟶ Factor is responsible

- Particulate in nature
- Maintain their separate identity

2. Discontinuous variations — Limited phenotypic class
   - Tall / Dwarf
   - Round / wrinkled

Rediscovery of Mendelian Work – 1900



Courtesy of American Philosophical Society, Curt Stern Papers.
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Carl Correns ✓



Erich von Tschermak ✓

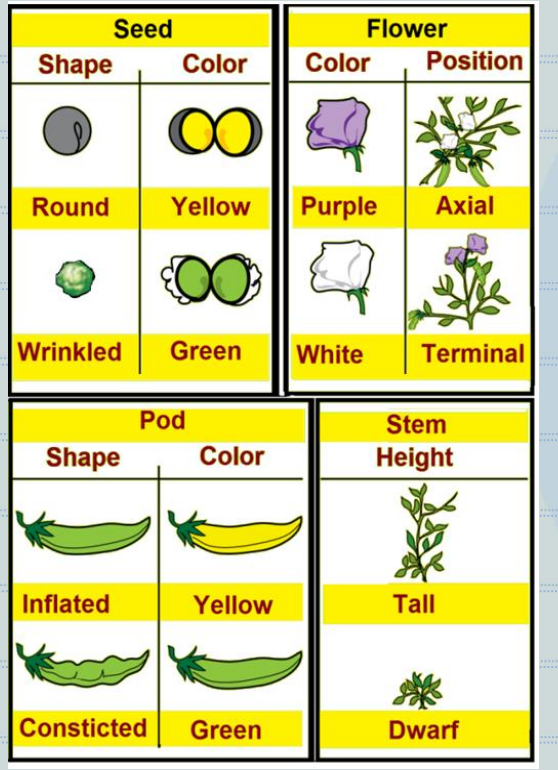


Hugo De Vries, around 1920.
Photo courtesy of
Cold Spring Harbor Laboratory Archives.

Hugo De Vries ✓

Reasons for Mendel's Success?

✓ Pisum sativum
- 7 characters



- Large sampling size.
- Used pure lines of Pea
- Focussed on one or few characters
- Experimental Approach and used Mathematics.
- Confirm his inferences from several generations of plants.

Character

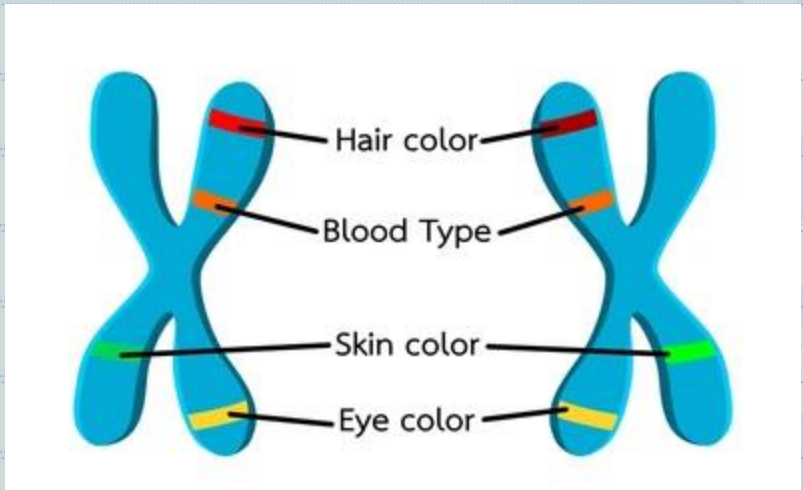
- A heritable feature that can vary between individuals.
- Attribute or feature under investigation

Example: height or eye color.

Character may have two or more traits- observable characteristics

Gene (Mendel's factor):

A "gene" is a unit of heredity that is transferred from parent to offspring and is responsible for the expression of a particular trait or characteristic.

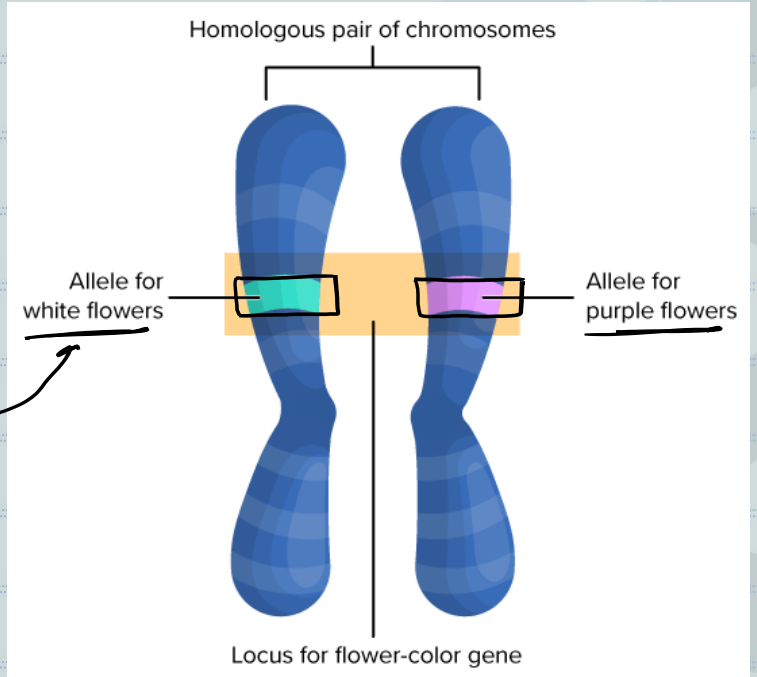


Gene: DNA sequence which is transcribed & contribute to phenotype

Resides on Chromosome

Allele

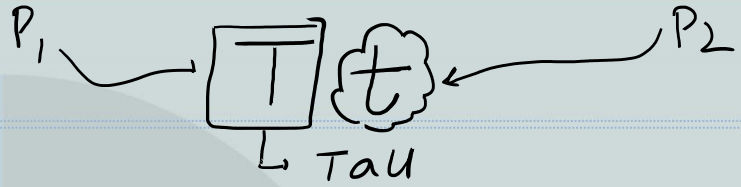
"Alleles" are different forms of the same gene that can result in variations in a trait.



Flower color

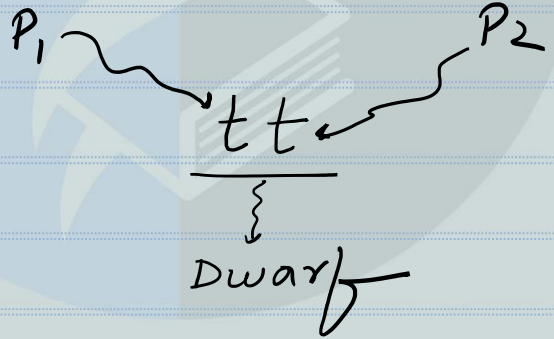
- Alternate form of same gene
- Resides at same locus on homologous chromosome

Dominant and Recessive Allele



A dominant allele produces a dominant phenotype in individuals who have one copy of the allele, which can come from just one parent.

For a recessive allele to produce a recessive phenotype, the individual must have two copies, one from each parent.



Plant Height — 1 gene is Responsible

Genotype → TT, Tt, tt

✓ Genotype

The **genotype** is the genetic constitution of an organism or cell—its molecular makeup.

Combination of alleles that an individual inherits from their parents

2 genes are responsible for a character — A & B

Genotypes : AABB, AaBB, AABb, AaBb, AAbb, aaBB, Aabb, aabb

Phenotype (Trait)

It refers to the observable characteristics or traits of an individual, which result from the interaction between their genotype and the environment.

Phenotype = Genotype + Enviornment

Penetrance

Phenotype = Genotype + Environment
✓ ×

✓ The extent to which a particular gene is expressed in the phenotypes of individuals carrying it.

$A^N A^N$ $A A^D$ $A^D A^D$

A^D : Dominant disorder.

Disease.

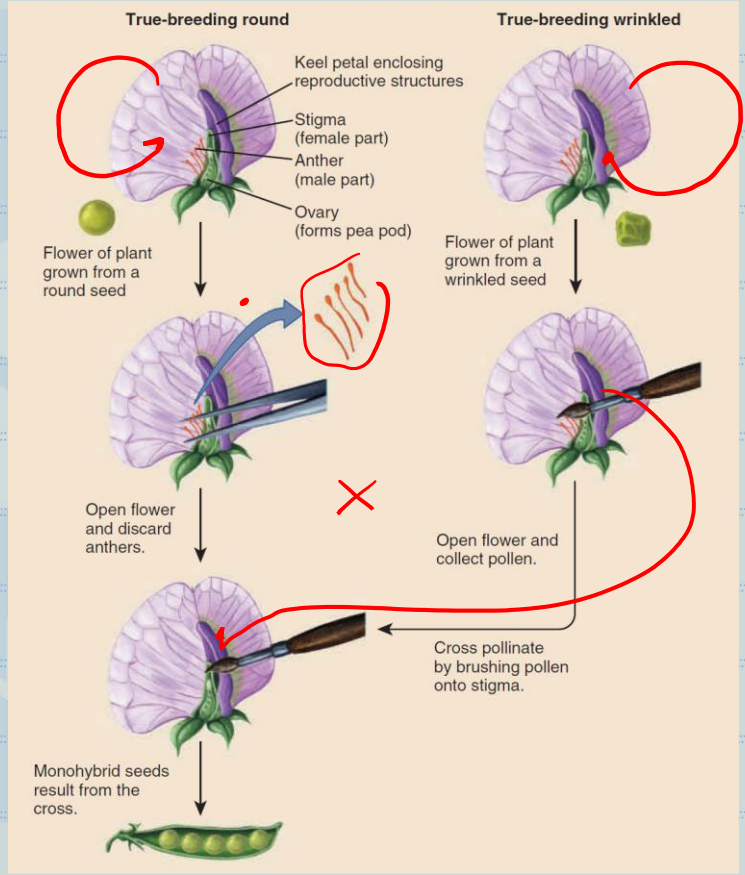
100 Individuals → $A^N A^D$ → 60 show disease symptoms

$$\text{Penetrance (\%)} = \frac{60}{100} \times 100 = 60\%.$$

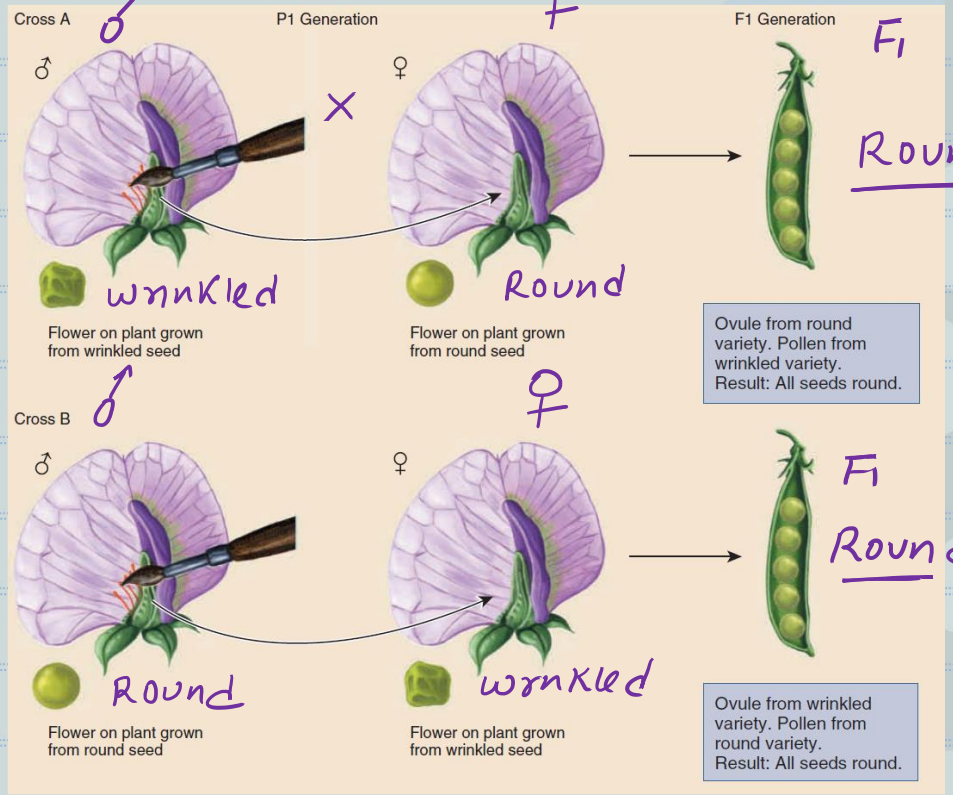
Pollination: Self and Cross

- when same parent behave as male & female

* Different individuals are behaving as male & female parents



Reciprocal cross



Yield equal Results

↓

Mendelian Genetics

↓

Genes resides on autosomes in nucleus and show 100% penetrance regardless of parental origin.

Back Cross

P_1 (AA) × P_2 (aa)

↓

F_1 - Generation

F_1 (Aa) × aa → Aa : aa

1 : 1

F_1 (Aa) × F_1 (Aa) → AA / Aa

F_1 (aa) × aa → All aa

- Cross of Progeny with any of parents

  $F_1 \times P_1$ (Dominant Phenotype)

  $F_1 \times P_2$ (Recessive Phenotype) ← Test cross

- Test cross is type of back cross where F_1 is crossed with homozygous recessive parent.
- Determine unknown genotype of F_1.

Probability

P = 0 to 1

- Predicts how likely something is to happen.

Probability of Head in coin = 1/2

Tail = 1/2

Boy = 1/2

Girl = 1/2

Dice probability of 1 = 1/6



F_2 : Tall 3/4

Dwarf 1/4.

multiplication
The product rule: More than one event in desired order

	1	2	3	
Coin Toss →	H	T	H	order

$1/2 \times 1/2 \times 1/2 = 1/8$

Sibship of 3 →	B	B	G	order

$1/2 \times 1/2 \times 1/2 = 1/8$

F_2 generation all 3 Tall: T T T

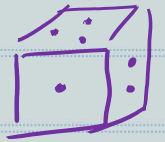
$3/4 \times 3/4 \times 3/4$

$= 27/64.$

The Sum rule: More than one outcome are accepted in same event(s).

Event 1 : Head or Tail

$\frac{1}{2} + \frac{1}{2} = 1$



Throw 1 ⟶ Probability of getting 2 or 6

$P(2) = \frac{1}{6}$ ✓

$P(6) = 1/6$ ✓

$\frac{1}{6} + \frac{1}{6} = \frac{2}{6} = \frac{1}{3}$

Theoretical probability

Expected based on assumption

Empirical probability

• observed after experiment

Theoretical probability is based on the assumption that outcomes have an equal chance of occurring while empirical probability is based on the observations of an experiment.

Toss coin 100 times

Expected 50 Head
50 Tail

$p(H) = 0.5$

$p(T) = 0.5$

• slightly different

Observation → 60 Head
40 Tail

$p(h) = 0.6$

$p(T) = 0.4$

Probability of multiple events = Number of ways X probability of each event

3 X 1/8 = 3/8

Toss coin 3 times — Desired events 2 Head & 1 Tail

Number of ways :

H → H → T
H → T → H
T → H → H

— 3

H, H, T

$\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$

Number of ways in which desired event can be done ?

- Out of 3 child : 2 Boy & 1 Girl
  BBG, BGB, GBB = 3 ways
- Out of 4 toss : 3 Head & 1 Tail
  HHHT, HHTH, HTHH, THHH = 4 ways.
- Out of 3 F_2 plants : 2 Tall & 1 Dwarf
  TTD, TDT, DTT = 3 ways
- Out of 4 toss : 2 Head & 2 Tail
  HHTT, HTTH, TTHH, HTHT, THTH ...

To find the number of ways where 'r' objects can be chosen from 'n' objects and the order is not important.

$$^{n}C_{r} = \frac{n!}{(n-r)!\,r!}$$

$$= \frac{4 \times 3 \times 2 \times 1}{(4-2)!\;2!}$$

$$= \frac{4 \times 3 \times 2 \times 1}{2 \times 1 \times 2 \times 1}$$

$$= 6.$$

n = the number of total items/objects.

r = how many items/object of one type are taken at a time.

Out of 4: 2 Head & 2 Tail

$n = 4$

$r = 2$

Apply your mind

✓ BBBG
✓ BBGB
✓ BGBB
✓ GBBB

4 ways

How many ways it can be done that out of 4 children 3 are boy and one is girl?

(1) 4
(2) 2
(3) 8
(4) 10

$n = 4$

$r = 3$ or 1

$$= \frac{n!}{(n-r)!\,r!} = \frac{4!}{(4-3)!\,3!} = \frac{4 \times 3 \times 2 \times 1}{1 \times 3 \times 2 \times 1}$$

$$= 4$$

Apply your mind

What is probability that a sibship of 4 children all are girls?

(1) 1/4
(2) 1/32
(3) 1/8
(4) 1/16

g g g g no of ways = 1

$1 \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = 1/16$

Apply your mind

How many ways it can be done that out of 4 children 2 are boy and two are girl?

(1) 6
(2) 12
(3) 8
(4) 10

$${}^{n}C_{r} \qquad n = 4, \quad r = 2$$

$$= \frac{n!}{(n-r)!\,r!} = \frac{4 \times 3 \times 2 \times 1}{(2 \times 1) \cdot (2 \times 1)}$$

$$= 6$$

Apply your mind

$F_2 \longrightarrow$ P(Tall) = 3/4
P(Dwarf) = 1/4

What is probability that a 3 plants picked at random from F2 of monohybrid cross of pea plant - all are tall?

(1) 1/16
(2) 9/64
(3) 27/64
(4) 63/64

no of ways

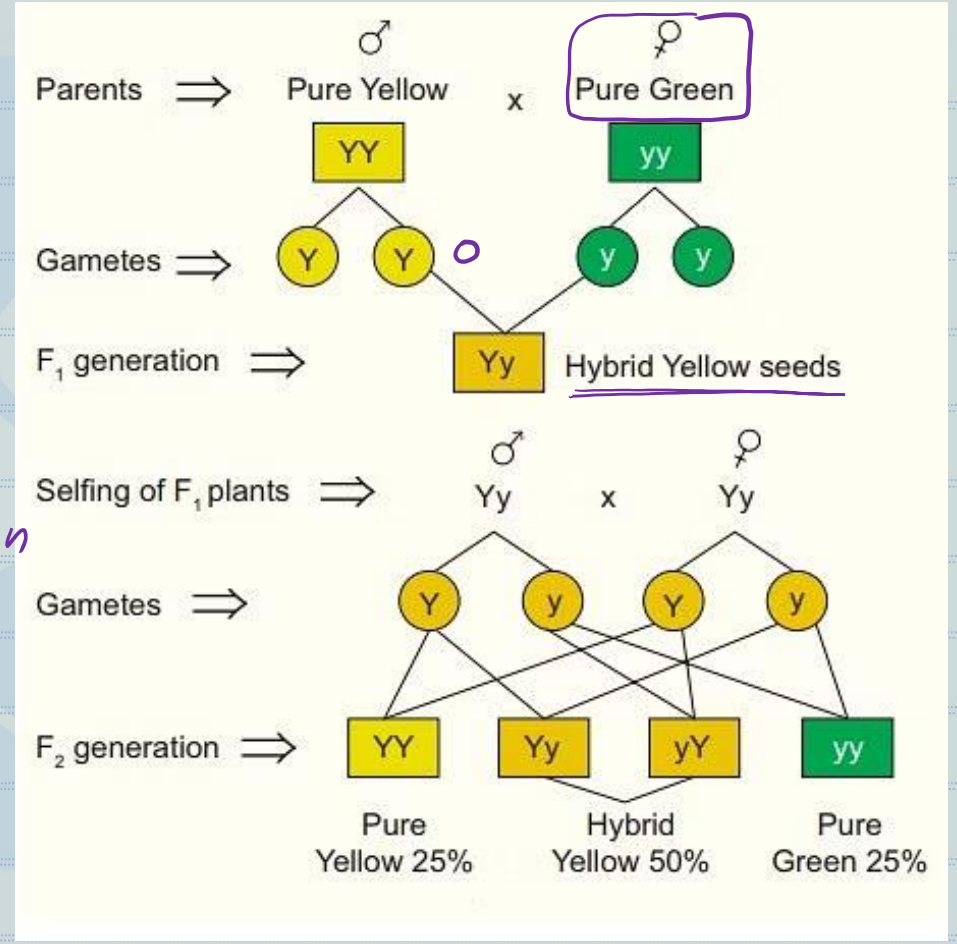
1	2	3
T	T	T

$1 \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} = \frac{27}{64}$

Monohybrid Cross

1. Cross between Parents shows variation is single character
2. Emasculation (female plant) and pollination (male plant)
3. All F1 were similar to one of parent
4. F1 were allowed to selfing
5. F2 Phenotypic ration: 3:1 Yellow : green
6. F2 Genotypic ration: 1:2:1

YY : Yy : yy
1/4 2/4 1/4

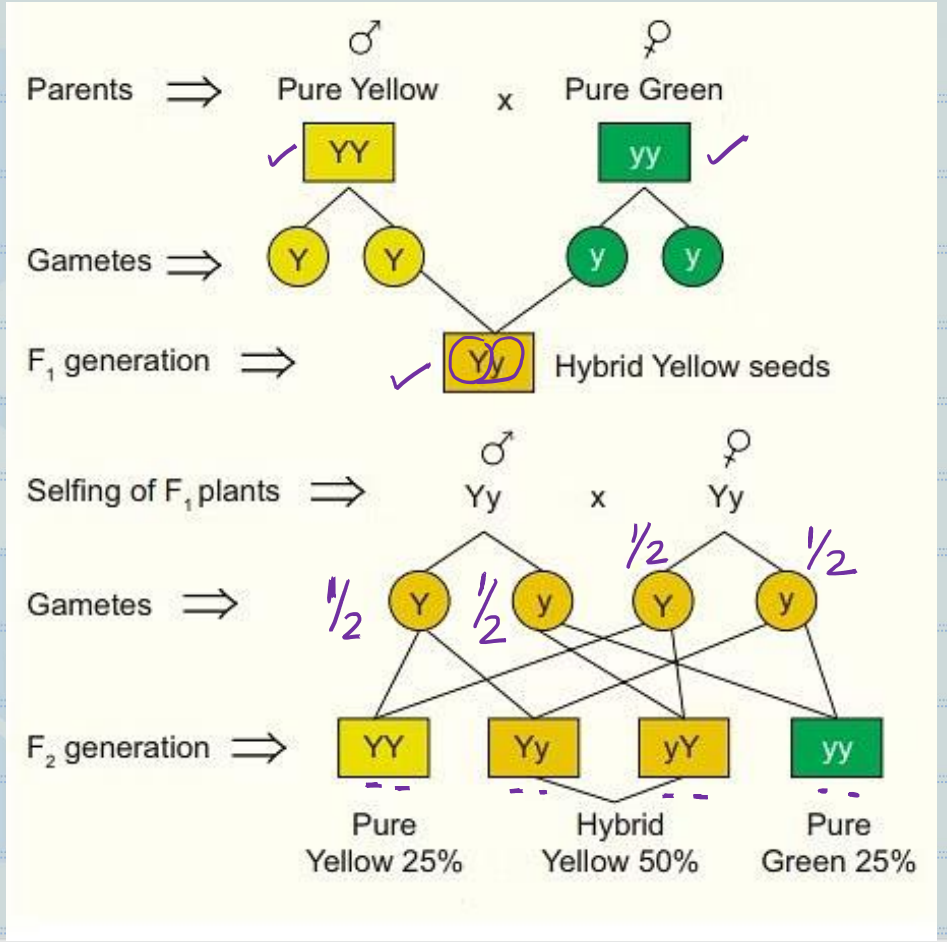


Conclusion monohybrid Cross

1. Each character is controlled by at least 2 factors
2. F1 progeny must inherit one factor from each parent
3. Two factors must separate during gamete formation by equal probability

	Y 1/2	y 1/2
Y 1/2	YY 1/4	Yy 1/4
y 1/2	Yy 1/4	yy 1/4

YY : 1/4 Yy 2/4 yy : 1/4



Concept of Dominance : Single allele is sufficient to provide dominant phenotype.

F_2

YY	1/4	⟶ Yellow	1/4	⟶ (3/4)	Yellow
Yy	2/4	⟶ Yellow	2/4		
yy	1/4	⟶ Green	1/4	⟶ (1/4)	Green

Genotypic Ratio : 1 : 2 : 1

Phenotypic Ratio : 3 : 1

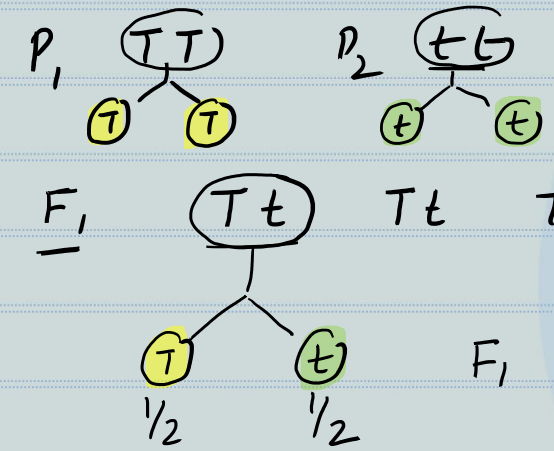
T t ⟶ Tall

R r ⟶ Round

Y y ⟶ Yellow

Law of Segregation :

For every character 2 factors are responsible and they separate during gamete formation by equal Probability.



Tt Tt Tt

- Purity of gametes

F_1 Tt × F_1 Tt

↓

F_2 TT : Tt : tt

1/4 2/4 1/4

Genotypic Ratio

1 : 2 : 1

F_1 Tt × tt (P_1)

↓

Tt : tt

1/2 1/2

Genotypic Ratio

1 : 1

Law of segregation:

- Universal Law
- Applies to all sexually reproducing organism

Proof:

F_2 Genotypic Ratio : 1 : 2 : 1

F_2 Test cross Ratio : 1 : 1

What is probability that a 4 plants picked at random from F2 of monohybrid cross of pea plant – 3 are tall and 1 is dwarf?

(1) 1/16
(2) 9/64
(3) 27/64 ✓
(4) 27/256

3 Tall, 1 Dwarf

[number of ways = 4
[Probability of one of way = TTTD

$\frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} = \frac{27}{256}$

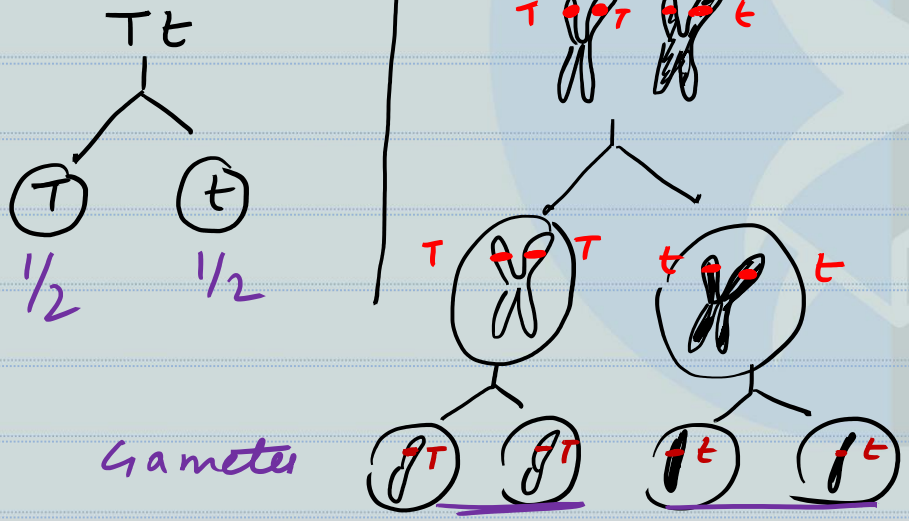
$n C_r = \frac{4!}{(4-3)!\,3!} = \frac{4 \times 3 \times 2 \times 1}{1 \times 3 \times 2 \times 1} = 4$

$4 \times \frac{27}{256} = \frac{27}{64}$

Physical Basis of inheritance

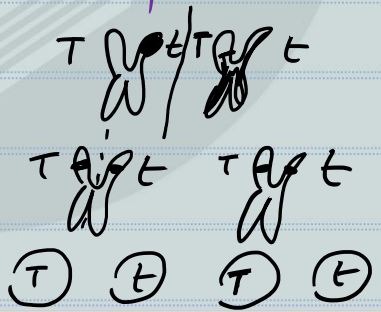
- If there is no crossing over, mendel factor separate during meiosis-I
- If there is crossing over mendel factor (factor) separate during meiosis II

Walter Sutton and Theodar Boveri: The genes are found in the chromosomes



Meiosis-I : Homologous Chromosomes separates

Meiosis II : Sister Chromatids separates



HAPPY LEARNING

THANKS

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