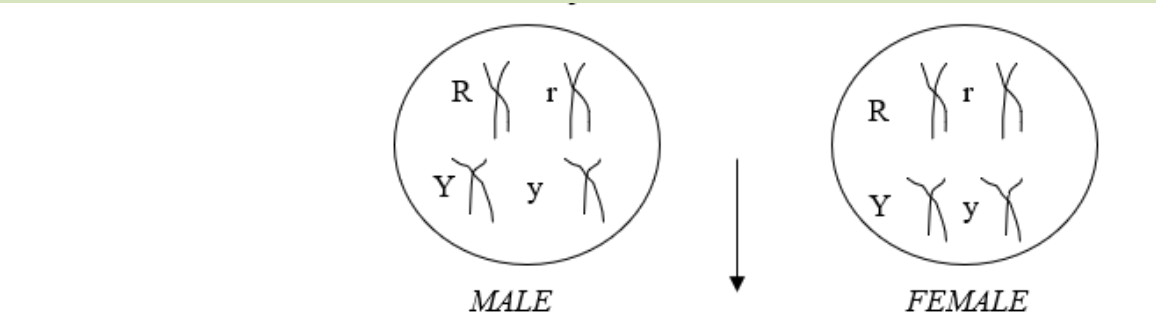
# Go to the "Slide Show" shade above

Click on "Play from Beginning"

# Chapter 12
# GENES & HEREDITY

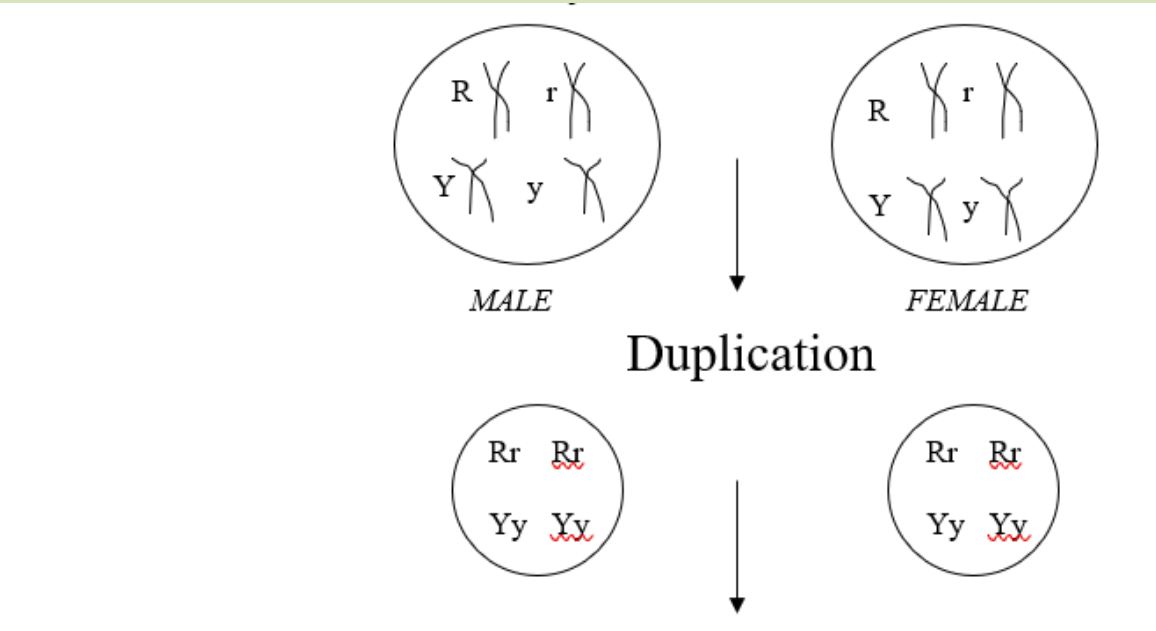
WARM-UP

# Show the Meiotic Division



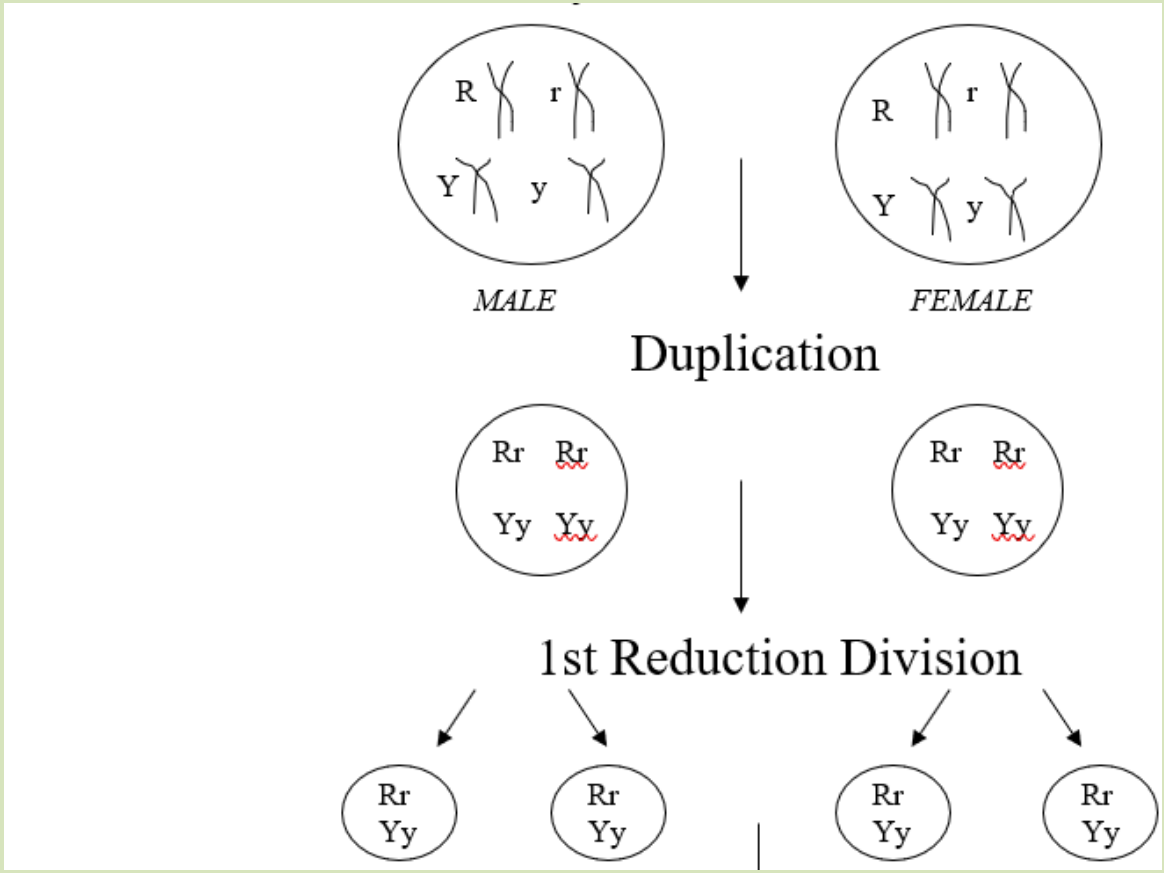
WARM-UP

# Show the Meiotic Division



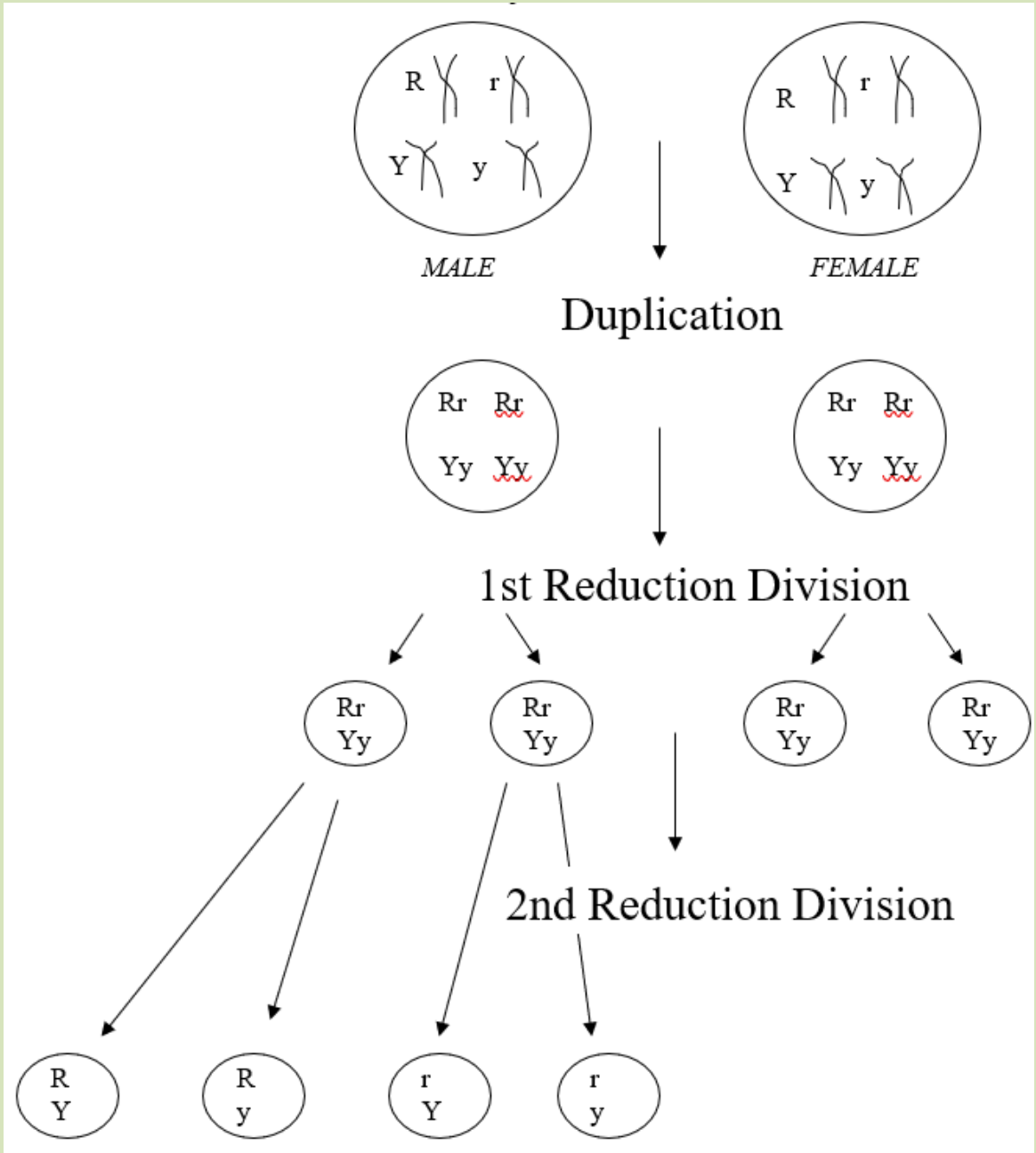
WARM-UP

# Show the Meiotic Division

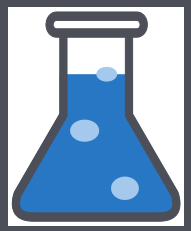


WARM-UP

# Show the Meiotic Division



# Lesson Objectives



By the end of this lesson, you should be able to:

- Define genetics and heredity, distinguishing characteristics from traits, genes, and alleles.
- Describe the history of genetics and Mendel's role.
- Delineate the science of heredity in terms of generations (P, $F_1$, $F_2$), differentiating genotype from phenotype.
- Understand and use the law of segregation to show monohybrid test crosses.
- Understand and use the law of independent assortment to show dihybrid test crosses.

- **Science Practice: Monohybrid Test Cross**

## GENETICS

The scientific study of the transmission of **traits** from parent to offspring through successive generations.

Tongue Roller



Non-roller

Widow's Peak

Attached earlobes (left)

Hitch-hiker's thumb

Mid-digital hair

Dimple in chin

Freckles

Morton's Toe

Genetics is the study of

# HEREDITY

*Transmission of **traits** from one generation to the next.*

# Species Traits

Traits that ALL members of a given species share.

- **humans** – walk erect, highly developed nervous system, opposable thumb, flat fingernail (primate), limited body hair.
- **Eagles** – wing span up to 6 feet; large talons; double fovia in eyes; hooked beak for tearing up prey;
- Affect the entire species in terms of survival vs. extinction.

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# Individual Traits

Traits that distinguish individual members within a given species.

- **humans** – color of eyes, skin, hair; stature (height, weight, bone structure); metabolic rate (thin, fat).
- **Eagles** – Bald Eagle; golden eagle; white-tailed eagle; Sea eagle.
- Normally does not affect the entire species in terms of survival vs. extinction.

# Genetics was founded on Observation (not science)

Traits were thought to be passed from generation to generation through the blood. Hence, the concept of “blood lines” or “blue blood” (royalty) was commonly accepted.

- “Blood Brothers” or Blood Sisters

“Orientals are short”

- Actually only due to dietary shortages (Yao is 7’7” tall in the NBA). Chinese people are as tall as Americans now that nutritional information was discovered.
- Knights of the round table were only 5’4” tall, yet were esteemed, revered for their stature in the medieval times (*again, nutrition was the issue, not genetics*).

# Gregor Mendel

- Austrian monk (1822-1884)
- Called the "Father of Genetics".
- Began the field of Genetics in the 1860s.
- Deduced the Principles of Genetics by breeding garden peas.

# Gregor Mendel

## (1822-1884)

**Entered a Monastery** in Brunn, Czechoslovakia to get an education

- It was *common (for centuries) for people who wanted to be spiritual to pursue monastic life.*
- *Prior to Reformation - mainly priests, nuns in Catholicism or Eastern Orthodoxy.*
- *After Reformation - monks, friars, bishops, reverends, pastors – depending on the religious persuasion … concept of clergy-laity was strong … still is*).

**University of Vienna** – Pursued mathematics and science for 2 years hoping to become a teacher … but he failed the certification exams.

**Became an Abbot in a Monastery**

# The Science of Genetics began in an Abbey Garden

Mendel is famous for his work with **pea plants** while in the monastery.

**Repeatable results**

*Many had previously done studies with plants, but their results were not conclusive or scientific.*

- Mendel choose **definite** and **measurable** hereditary traits.
- Mendel applied mathematics to his observations (objective, not "subjective" or philosophical in nature).
- Mendel took a scientific approach in all his work using a logical sequence with proper methodology.
- Mendel received world-wide recognition in 1901 for his work and scientific approach which he published (the recognition came AFTER HE DIED).

# The Science of Genetics began in an Abbey Garden

- In 1866, Mendel
  - correctly argued that parents pass on to their offspring discrete **"heritable factors"** and
  - stressed that the heritable factors (today called **GENES**) retain their individuality generation after generation.
- A heritable feature that varies among individuals, such as flower color, is called a **CHARACTER**.
- Each variant for a character, such as purple or white flowers, is a **TRAIT**.

| Character | Traits | |
|---|---|---|
| | Dominant | Recessive |
| Flower color |  Purple |  White |
| Flower position |  Axial |  Terminal |
| Seed color |  Yellow |  Green |
| Seed shape |  Round |  Wrinkled |
| Pod shape |  Inflated |  Constricted |
| Pod color |  Green |  Yellow |
| Stem length |  Tall |  Dwarf |

# The Science of Genetics began in an Abbey Garden

- **True-Breeding** varieties result when self-fertilization produces offspring all identical to the parent.
- The offspring of two different varieties are **Hybrids**.
- The cross-fertilization is a hybridization, or **Genetic Cross**.
- True-breeding parental plants are the **P Generation**.
- Hybrid offspring are the **$F_1$ Generation** **(1st "filial" gen.)**.
- A cross of $F_1$ plants produces an **$F_2$ Generation** **(2nd "filial" generation.)**.

## The Experiment

**P generation**
**(true-breeding parents)**

Purple flowers × White flowers

**$F_1$ generation**

All plants have purple flowers

Fertilization among $F_1$ plants ($F_1 \times F_1$)

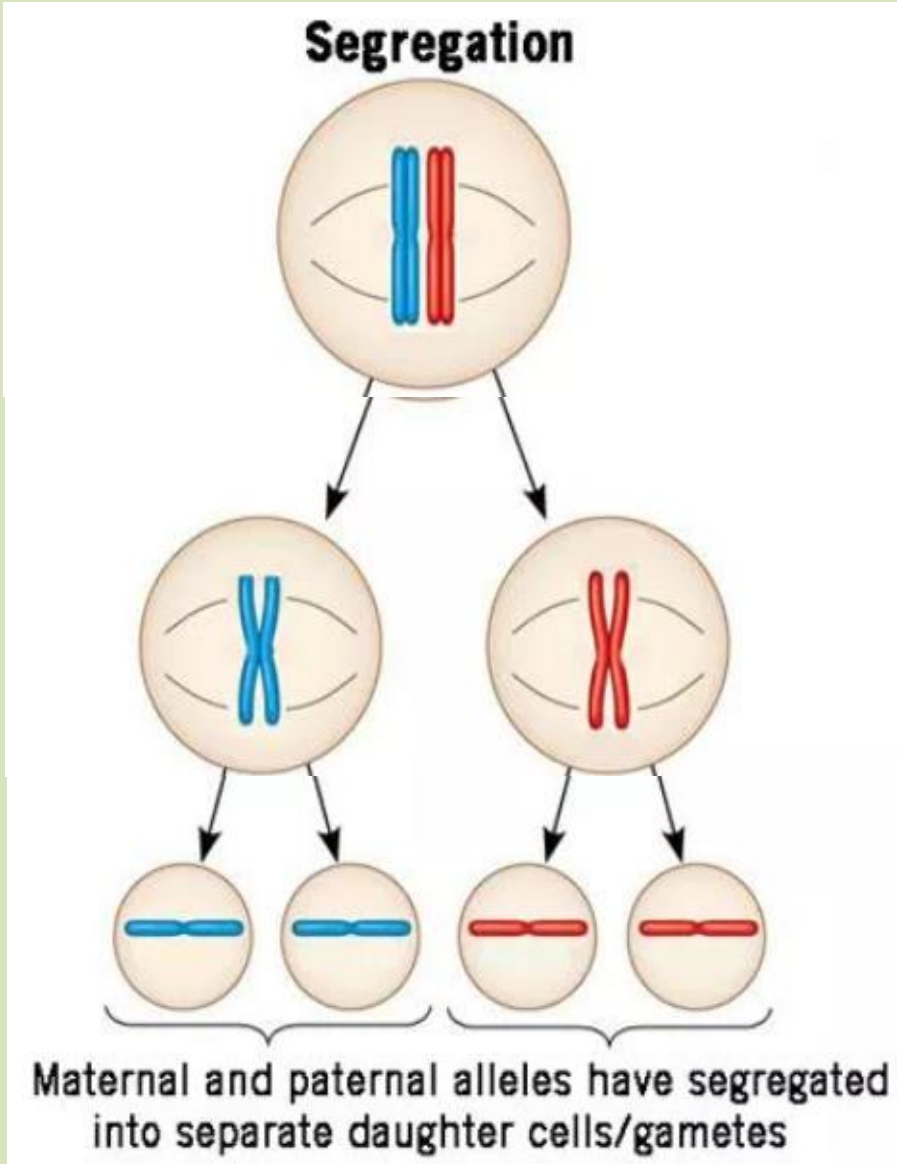
**$F_2$ generation**

$\frac{3}{4}$ of plants have purple flowers

$\frac{1}{4}$ of plants have white flowers

# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

- During fertilization, gametes randomly pair to produce combinations of alleles.
- Two alleles for each trait must separate when gametes are formed.
- A parent passes only one allele for each trait to their offspring.
- This is a random process.



# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

A **sperm** or **egg** carries only one allele for each inherited character…

- Because allele pairs **separate (segregate)** from each other during the production of gametes **(Meiosis).**
- This statement is called the **LAW OF SEGREGATION**.
- The fusion of gametes at **fertilization** creates allele pairs once again.

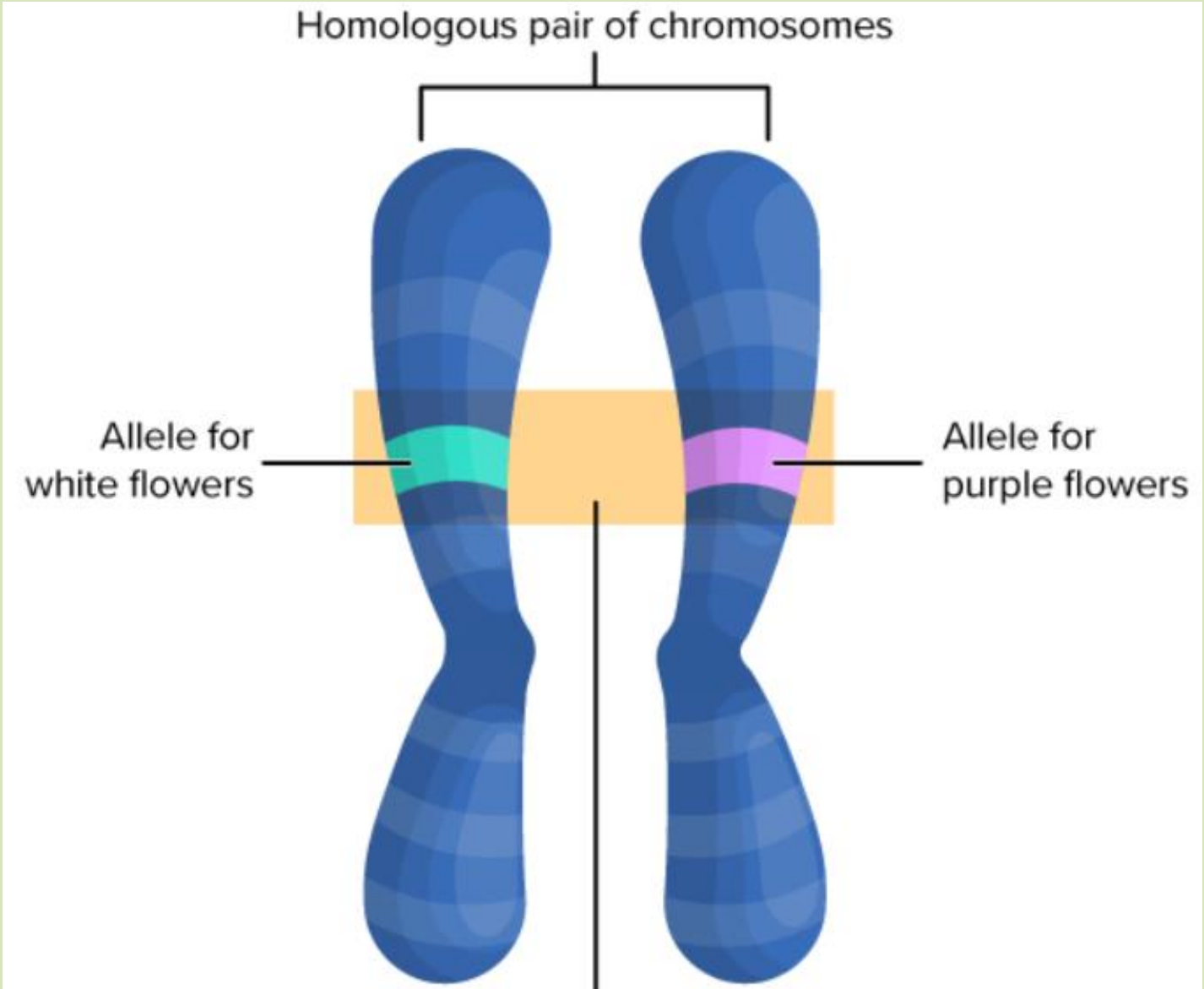
# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

- A cross between two individuals differing in a **single character** is a **Monohybrid Cross**.
- Mendel performed a monohybrid cross between a plant with purple flowers and a plant with white flowers.
  - The **$F_1$ Generation** produced all plants with purple flowers.
  - A **cross of $F_1$ plants** with each other produced an **$F_2$ Generation** with ¾ purple and ¼ white flowers.

# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

- The **all-purple $F_1$ Generation** did not produce white flowers.
- Mendel needed to explain
  - **why** white color seemed to disappear in the $F_1$ generation and
  - **why** white color reappeared in one-quarter of the $F_2$ offspring.

# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character



# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

Mendel developed four hypotheses, described below using modern terminology:

- **Alleles** are **alternative versions of genes** that account for **variations** in inherited characters.
- For **each character**, an organism inherits **two alleles**, **one from each parent**. The alleles can be the same or different.
  - A **Homozygous** genotype has **identical alleles**.
  - A **Heterozygous** genotype has **two different alleles**.

# Mendel's LAW OF DOMINANCE

- For each trait, one gene Allele can mask the expression of the other gene allele.
- If the alleles of an inherited pair differ, then one determines the organism's appearance. The gene allele that masks over another allele is called the **Dominant** allele.
- The other gene allele has no noticeable effect on the organism's appearance and gets masked. This is called the **Recessive Allele**.
- E.g. Tall (T) is the dominant allele while Short (t) is the recessive allele.

# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

The **Phenotype** is the appearance or expression of a trait.

- **The PHYSICAL appearance of an organism – what you see.**
- TALL pea plants, BLONDE hair, BLUE eyes, etc.

- The **Genotype** is the actual genetic makeup of a trait.
  - There are three possible genotypic expressions for every trait:
    - Homozygous Dominant – "pure bred"
    - Heterozygous Dominant – "Hybrid"
      - Both genotypes give the same phenotype (appearance).
      - The offspring give away what the genotype of the parents is.
    - Homozygous Recessive

- The **same phenotype** may be determined by **more than one genotype**.

# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

## Genotype

**GENOTYPE**

The genotype is an organism's genetic information.

**BB**
homozygous dominant

**Bb**
heterozygous

**bb**
homozygous recessive

## Phenotype

**PHENOTYPE**

The phenotype is the set of observable physical traits.

purple

purple

white

# Mendel's LAW OF SEGREGATION describes the Inheritance of a Single Character

- Mendel's hypotheses also explain the **3:1 ratio** in the **$F_2$ Generation**.
  - The **$F_1$ Hybrids** all have a ***Pp* genotype**.
  - A **Punnett Square** shows the four possible combinations of alleles that could occur when these gametes combine **(probability).**

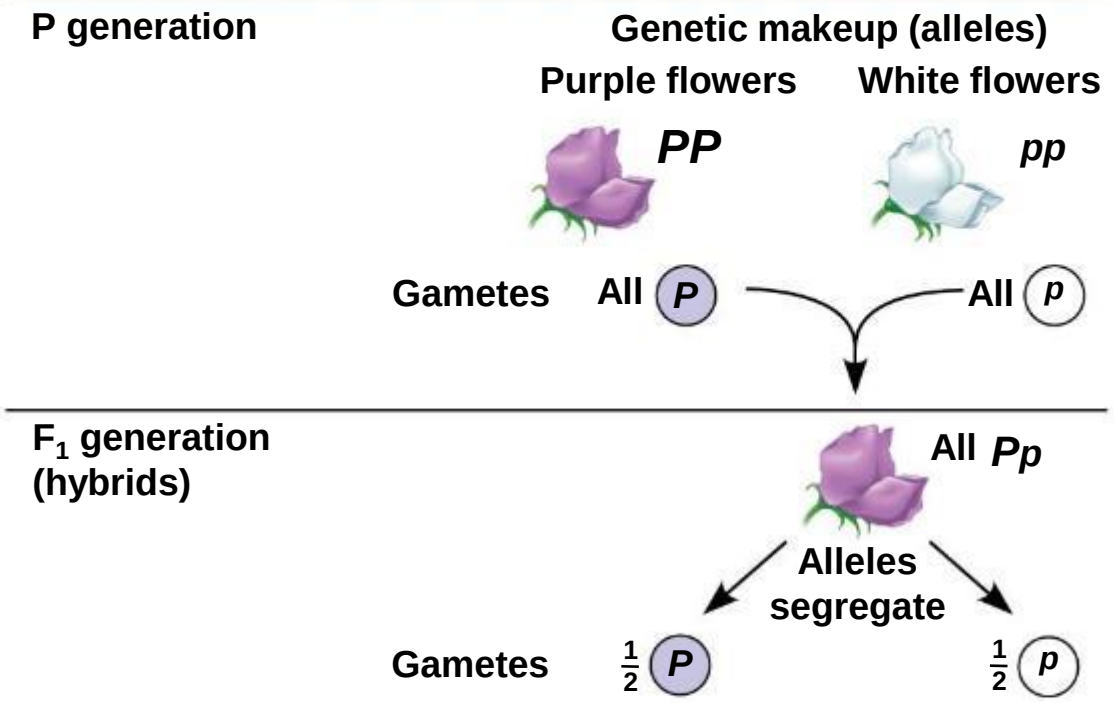
## The Explanation

**P generation**

**Genetic makeup (alleles)**

| Purple flowers | White flowers |
| --- | --- |
|  ***PP*** |  ***pp*** |
| **Gametes** **All** ***P*** | **All** ***p*** |

## The Explanation

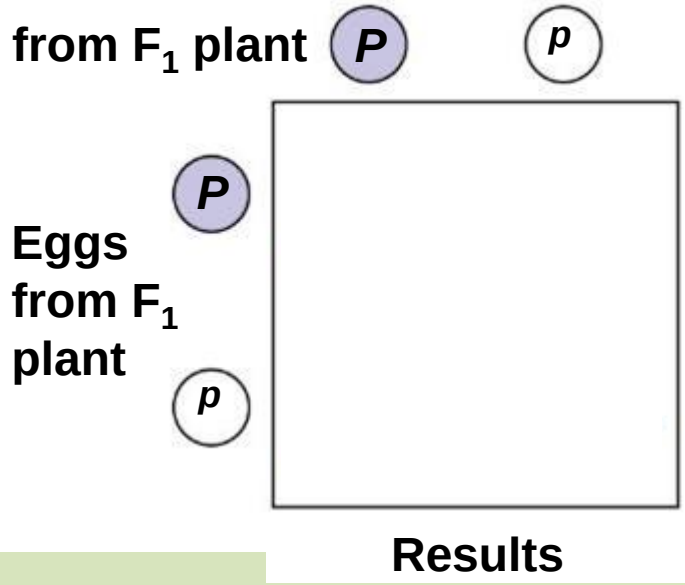


# $F_2$ generation

**Results:**

**Phenotypic ratio**
**3 purple : 1 white**

**Genotypic ratio**
**1 *PP* : 2 *Pp* : 1 *pp***

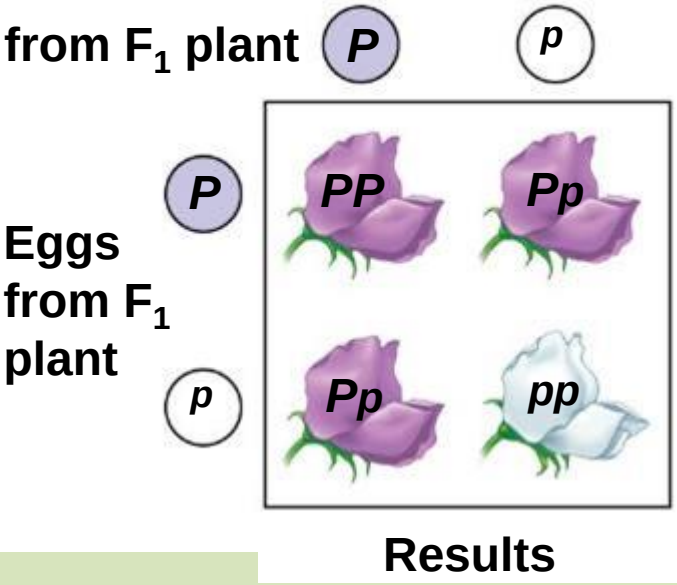


# $F_2$ generation

**Results:**

**Phenotypic ratio**
**3 purple : 1 white**

**Genotypic ratio**
**1 *PP* : 2 *Pp* : 1 *pp***
***25% homozygous dom***
***50% heterozygous dom***
***25% homozygous rec***



## The Explanation

**P generation**

**Genetic makeup (alleles)**

**Purple flowers** **White flowers**

***PP***

***pp***

**Gametes** **All** ***P*** **All** ***p***

**$F_1$ generation (hybrids)**

**All *Pp***

**Alleles segregate**

**Gametes** $\frac{1}{2}$ ***P*** $\frac{1}{2}$ ***p***

**Fertilization**

**$F_2$ generation**

**Results:**

**Sperm from $F_1$ plant** ***P*** ***p***

**Phenotypic ratio**
**3 purple : 1 white**

**Eggs from $F_1$ plant**

***P*** ***p***

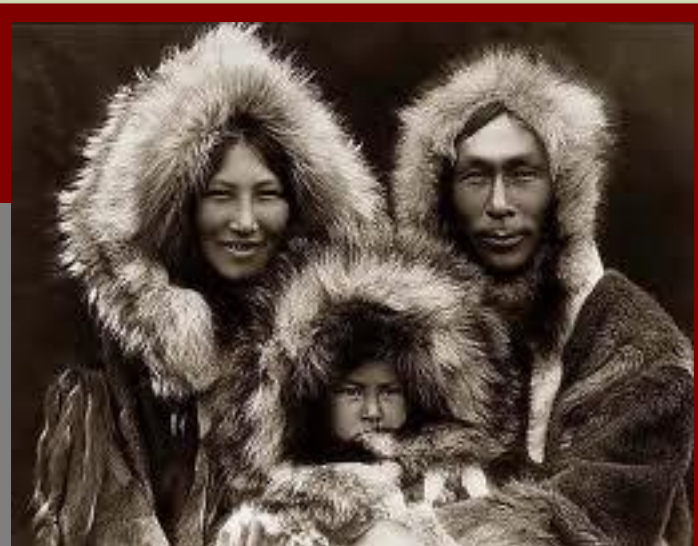
| | |
|---|---|
| ***PP*** | ***Pp*** |
| ***Pp*** | ***pp*** |

**Results**

**Genotypic ratio**
**1 *PP* : 2 *Pp* : 1 *pp***
***25% homozygous dom***
***50% heterozygous dom***
***25% homozygous rec***

If we know the genetic makeup of parents, what type of offspring might they produce?

What is the **probability** of producing different types of offspring?



# Probability

The likelihood that a particular event will occur.

## ACTIVITY: Coin Toss

- e.g. coin flips flip a coin 10 times per person in the class and total the heads vs. the tails (~200 trials)
- The probability comes out ~50% heads and 50% tails for many trials
- Probability is NOT a law and may not always give expected results
- *E.g. After four girls, what is the probability of having another girl?*

# R. C. Punnett Mathematician

Use of Punnett Squares and even Genetics itself is a method of studying the probability or chance of various offspring.

# Punnett Square

A Punnett square is a diagram showing the allele combinations that might result from a genetic cross between two parents.

It is a "Test Cross" or Back Cross to determine if alleles are homozygous or heterozygous.


- Mendel began his experiments using true-breeding parents.
- **He soon discovered that the tall trait was dominant over the dwarf trait.**
- Let's cross a true-breeding tall pea plant [genotype TT] to a true-breeding dwarf pea plant [genotype tt].

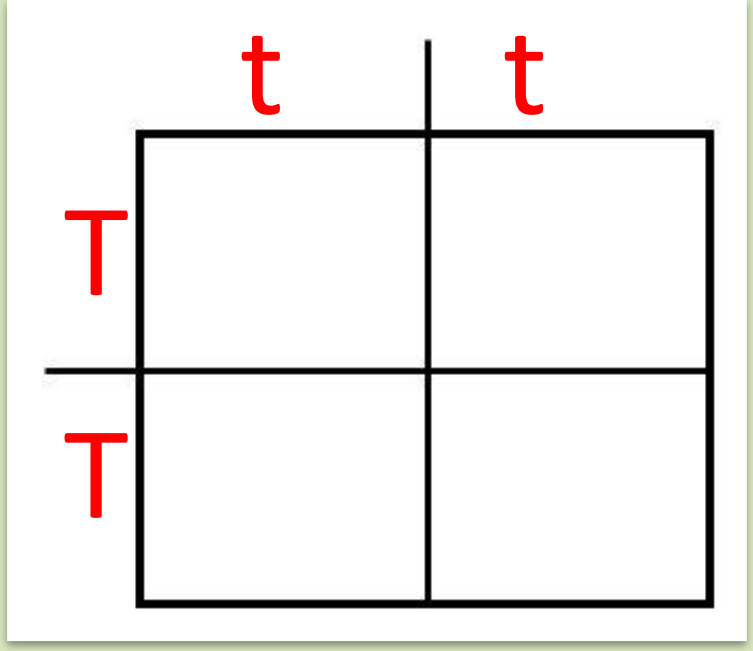
# Punnett Square

A Punnett square is a diagram showing the allele combinations that might result from a genetic cross between two parents.

It is a "Test Cross" or Back Cross to determine if alleles are homozygous or heterozygous.

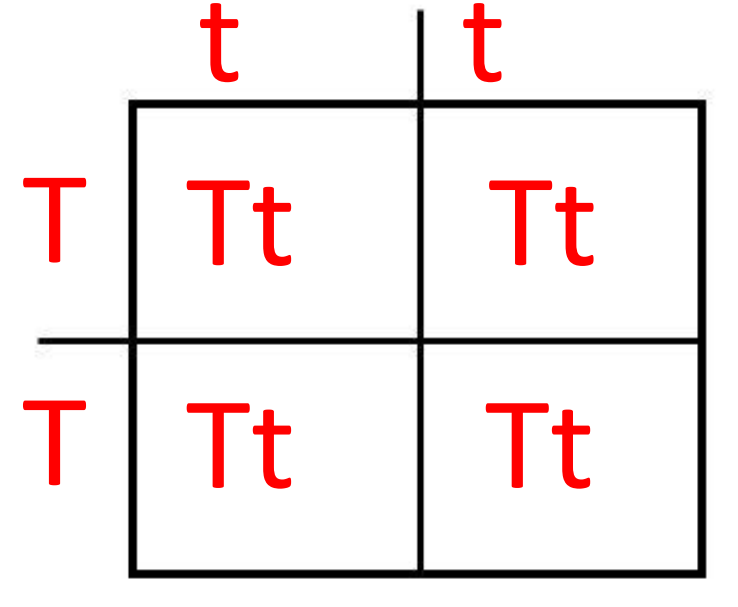
Place the alleles of the first parent on the top of the square.

Place the alleles for the second parent on the left of the square.



# Punnett Square

Fill in the squares to show all the possible combinations of alleles that the offspring might inherit.



# $F_1$ Generation

***offspring of P generation***

- What are the different kinds of offspring that can be formed from parent cells?
- What is the probability for each trait being expressed phenotypically and genotypically?
- The $F_1$ Generation is the first “Filial” generation of the parents.

P Generation

Sperm + egg → zygote

TT tt → $F_1$





**$F_1$ Generation**

*Phenotype*:

? Tall

*Genotype*:

? Heterozygous

# $F_1$ Generation

*offspring of P generation*

- What are the different kinds of offspring that can be formed from parent cells?
- What is the probability for each trait being expressed phenotypically and genotypically?
- The $F_1$ Generation is the first "Filial" generation of the parents.

P Generation

Sperm + egg → zygote

TT tt → $F_1$

| | t | t |
|---|---|---|
| T | Tt | Tt |
| T | Tt | Tt |

→

**$F_1$ Generation**

*Phenotype*:

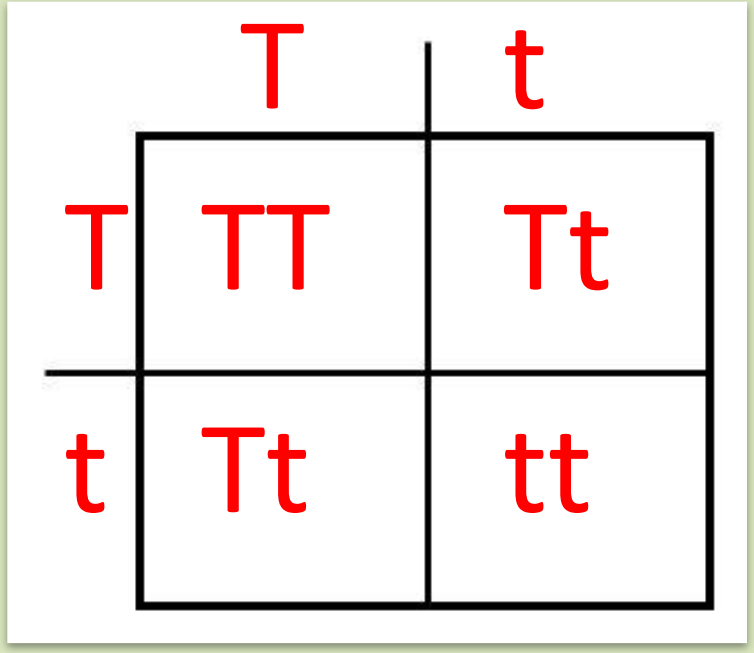
100% Tall

*Genotype*:

100% Heterozygous

# Punnett Square

Use the law of segregation to perform the Punnett square of the $F_1$ generation (both $F_1$ parents are Tt).



# $F_2$ Generation

## *offspring of $F_1$ generation*

- Mendel assumed that an equal number of male and female gametes are produced that could contribute to fertilization (zygote).
- Mendel also assumed that gametes combine at RANDOM.

$F_1$ Generation
Sperm + egg → zygote
Tt Tt → $F_2$

| | T | t |
|---|---|---|
| T | TT | Tt |
| t | Tt | tt |

→

**$F_2$ Generation**

*Phenotype*:

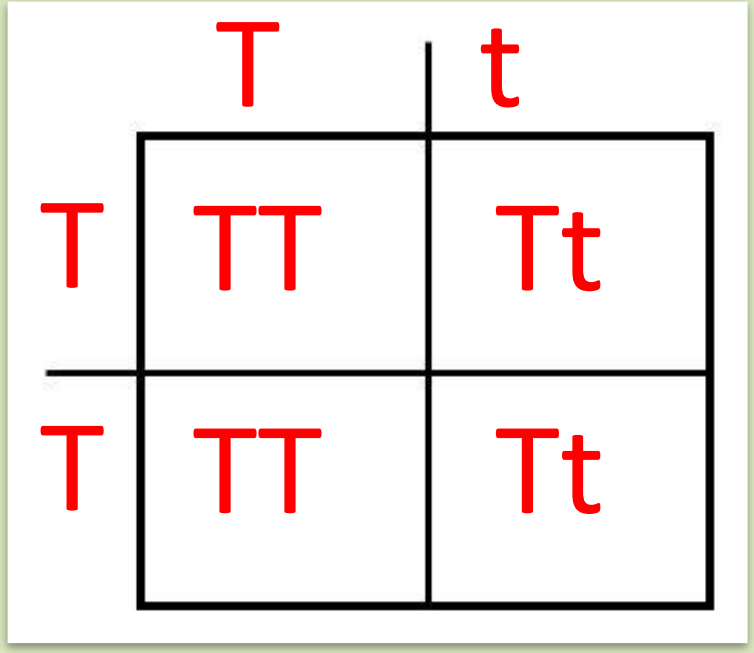
75% Tall
25% Short

*Genotype*:

25% Homozygous Tall
50% Heterozygous Tall
25% Homozygous Short

# Using Probability and Punnett Squares to Work Genetics Problems

## Punnett Square




1. The **alleles** of the first parent will be placed across the **top** of the square.
2. The **alleles** of the second parent will be placed along the **left side** of the square.
3. The possible gene combinations of the offspring will be placed inside the squares.
4. **Letters** represent the **alleles**.
5. Capital letters represent **Dominant** alleles.
6. Lower case letters represent **Recessive** alleles.

- In the original Test Cross (TT with tt), the dwarf trait disappeared.
- Mendel allowed the $F_1$ generation to self-pollinate.
- Show this cross using the Punnett square below.

TRY IT

What is the genotype of each parent?

Set up the Punnett Square.

Give the results.

- In the original Test Cross (TT with tt), the dwarf trait disappeared.
- Mendel allowed the $F_1$ generation to self-pollinate.
- Show this cross using the Punnett square below.

TRY IT

## The genotype of each parent? Tt x Tt

| | T | t |
|---|---|---|
| **T** | TT | Tt |
| **t** | Tt | tt |

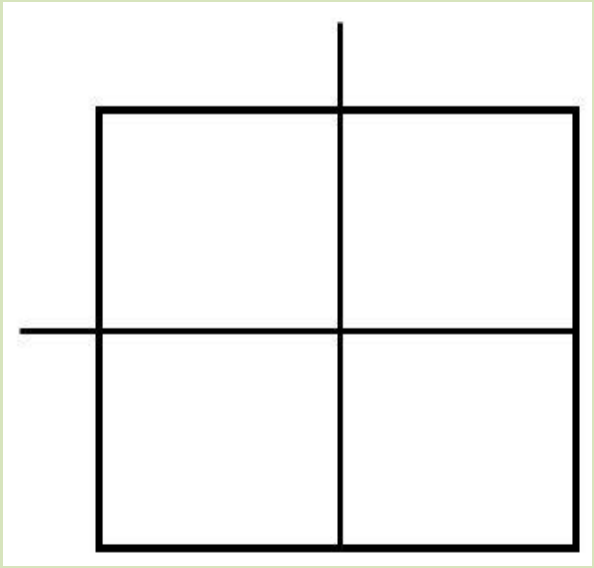
| Genotypes | Phenotypes |
|---|---|
| 1/4 TT | ¾ Tall |
| 2/4 Tt | |
| 1/4 tt | ¼ dwarf |

Having dimples is dominant over the absence of dimples. Cross a heterozygous dimpled man with a woman who does not have dimples. Show all work in the Punnett square and summarize your findings in the table

TRY IT

What is the genotype of the man?

What is the genotype of the woman?



| Genotypes | Phenotypes |
| --- | --- |
| | |

Having dimples is dominant over the absence of dimples. Cross a heterozygous dimpled man with a woman who does not have dimples. Show all work in the Punnett square and summarize your findings in the table

TRY IT

What is the genotype of the man? Dd

What is the genotype of the woman? dd

| | D | d |
|---|---|---|
| d | Dd | dd |
| d | Dd | dd |

| Genotypes | Phenotypes |
|---|---|
| 2/4 Dd<br>2/4 dd | 2/4 dimples<br>2/4 no dimples |

In dogs, the allele for short hair (B) is dominant over the allele for long hair (b). Two short haired dogs have a litter of puppies. Some of the puppies have short hair and some of the puppies have long hair.

What are the genotypes of the parents?

If the litter of puppies contained 12 pups, how many would you expect to have short hair?

How many would you expect to have long hair?

In dogs, the allele for short hair (B) is dominant over the allele for long hair (b). Two short haired dogs have a litter of puppies. Some of the puppies have short hair and some of the puppies have long hair.

## The genotypes of the parents?

Bb and Bb

| | B | b |
|---|---|---|
| B | BB | Bb |
| b | Bb | bb |

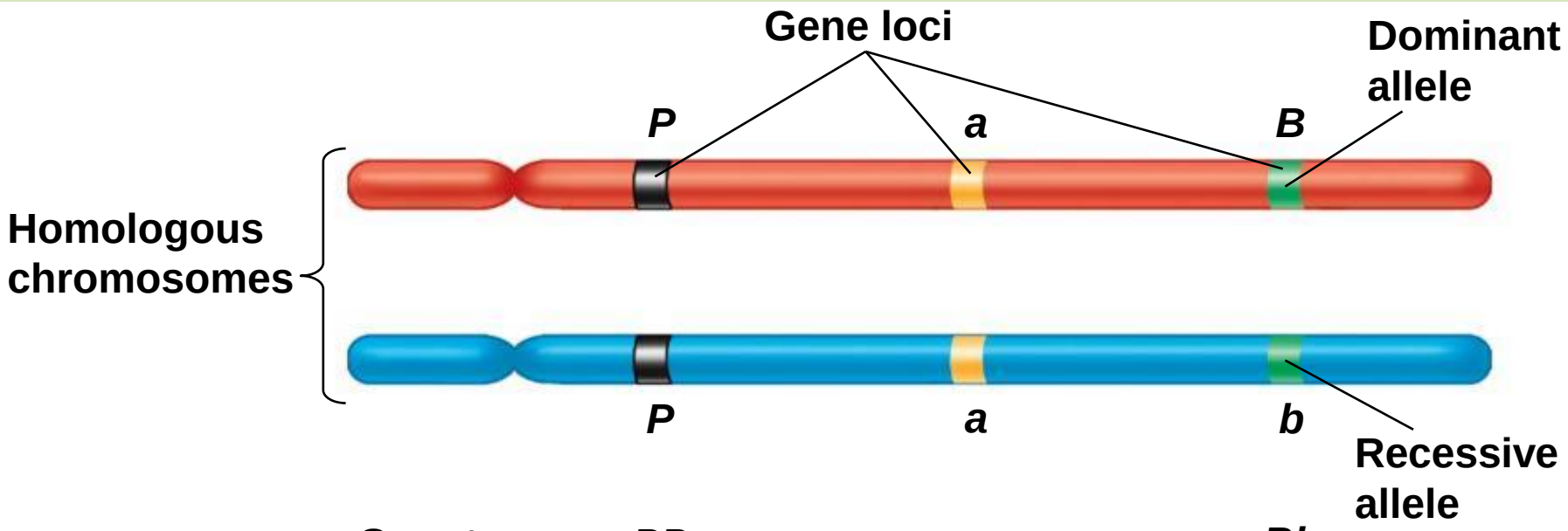
| Genotypes | Phenotypes |
|---|---|
| 1/4 BB<br>2/4 Bb<br>1/4 bb | ¾ short hair<br>¼ long hair |

If the litter of puppies contained 12 pups, how many would you expect to have short hair? ¾ of the 12 should have short hair. ¾ of 12 = 9 pups

How many would you expect to have long hair? ¼ of 12 = 3 pups

# Homologous Chromosomes bear the Alleles for Each Character

- A **LOCUS** (plural, *loci*) is the specific location of a **gene** along a **chromosome**.
- For a pair of **homologous chromosomes** (**homologs**), alleles of a gene reside at the same locus.
  - **Homozygous** individuals have the **same allele on both homologs**.
  - **Heterozygous** individuals have a **different allele on each homolog**.




**Genotype:** ***PP*** ***aa*** ***Bb***

**Homozygous for the dominant allele**

**Homozygous for the recessive allele**

**Heterozygous, with one dominant and one recessive allele**

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

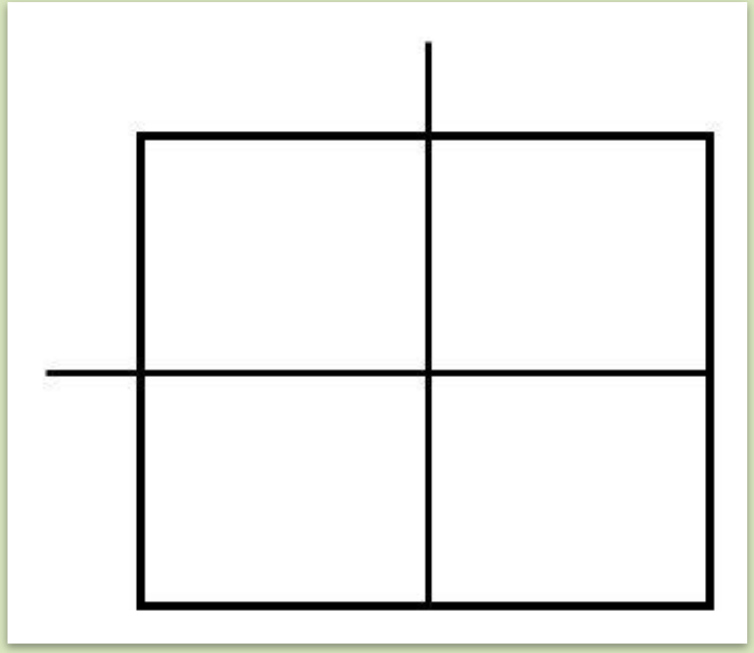
- Involves TWO traits and their inheritance.
- **The LAW of INDEPENDENT ASSORTMENT**
  - **Each trait is inherited separately or independently – they do not influence one another in heredity.**
  - **Each trait (allele) is found on a different chromosome.**
- Mendel studied crosses between the Pea Plants that simultaneously differed in TWO characteristics.
  - **He crossed Round, yellow seeds (RRYY) with wrinkled, green seeds (rryy) and found that their offspring expressed completely different characteristics.**

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

**Traits: Seed shape** and **Seed color**

**P generation**: RRYY x rryy

Round yellow Wrinkled green



QUICK CHECK

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

**Traits:** **Seed shape** and **Seed color**

**P generation:** RRYY (Round yellow) x rryy (Wrinkled green)

QUICK CHECK

| | ry | ry |
|---|---|---|
| RY | RrYy | RrYy |
| RY | RrYy | RrYy |

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

**Traits: Seed shape** and **Seed color**

**P generation**: RRYY (Round yellow) x rryy (Wrinkled green)

| Gametes | RY | RY |
|---|---|---|
| ry | RrYy<br>Round yellow | RrYy<br>Round yellow |
| ry | RrYy<br>Round yellow | RrYy<br>Round yellow |

**$F_1$ Generation**

*Phenotype*:

100% Round Yellow

*Genotype*:

100% Heterozygous for Round, Yellow

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

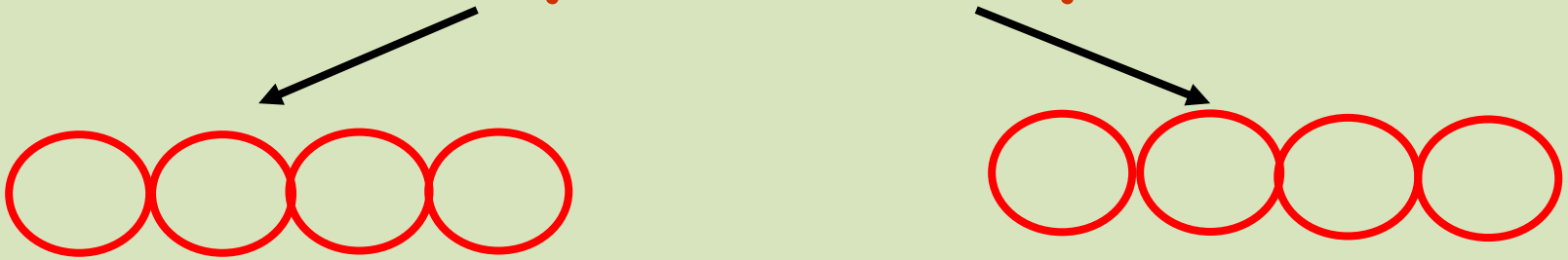
**Traits:** Seed shape and Seed color

**Alleles:** R round r wrinkled

Y yellow y green

## $F_1$ generation

RrYy x RrYy



All possible gamete combinations?

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

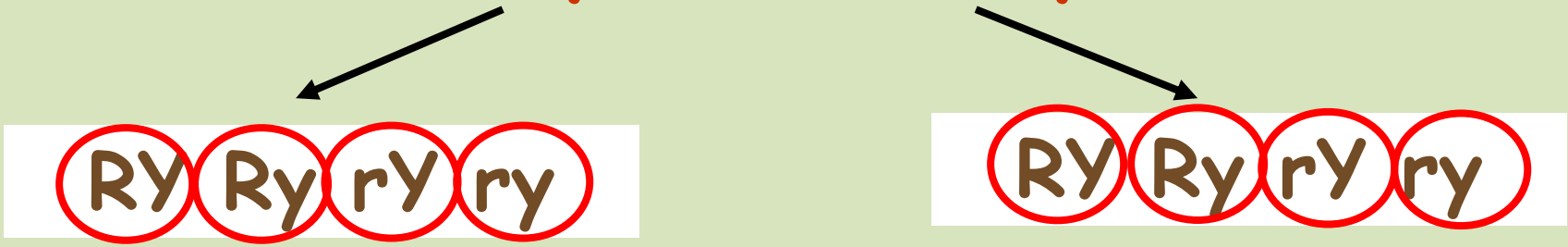
**Traits:** Seed shape and Seed color

**Alleles:** R round r wrinkled

Y yellow y green

## $F_1$ generation

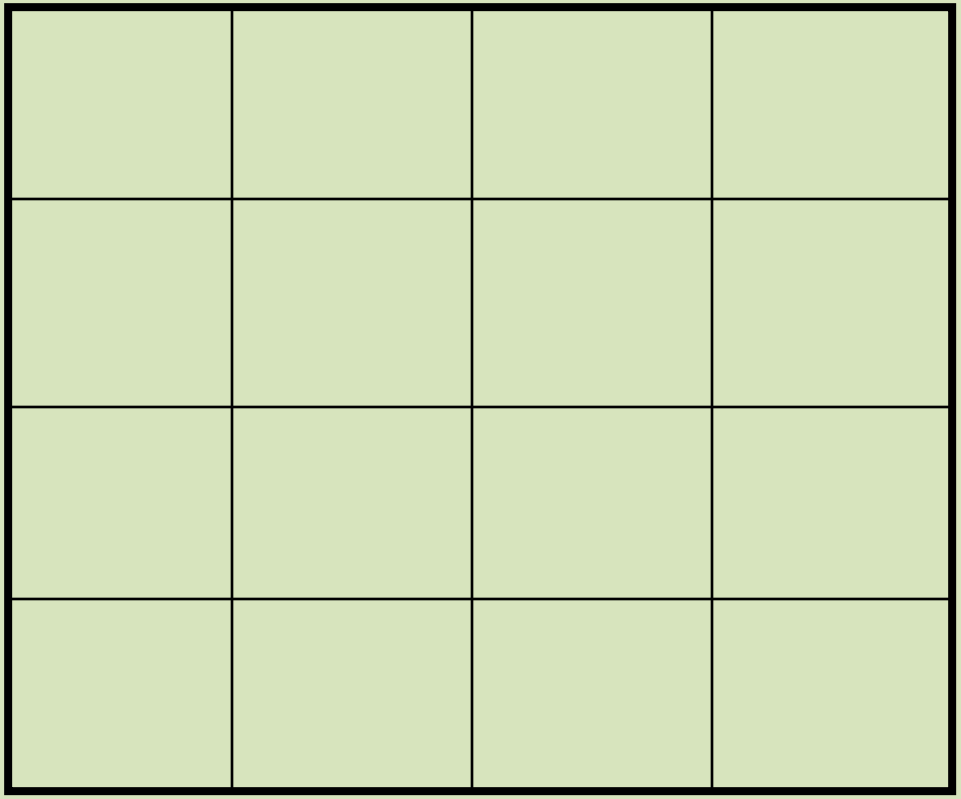
RrYy x RrYy




All possible gamete combinations

- ✓ When two traits are being considered (dihybrid cross), the Punnett square will need 16 squares.
- ✓ Each parent will pass one allele of each gene pair to the offspring. **RrYy x RrYy**

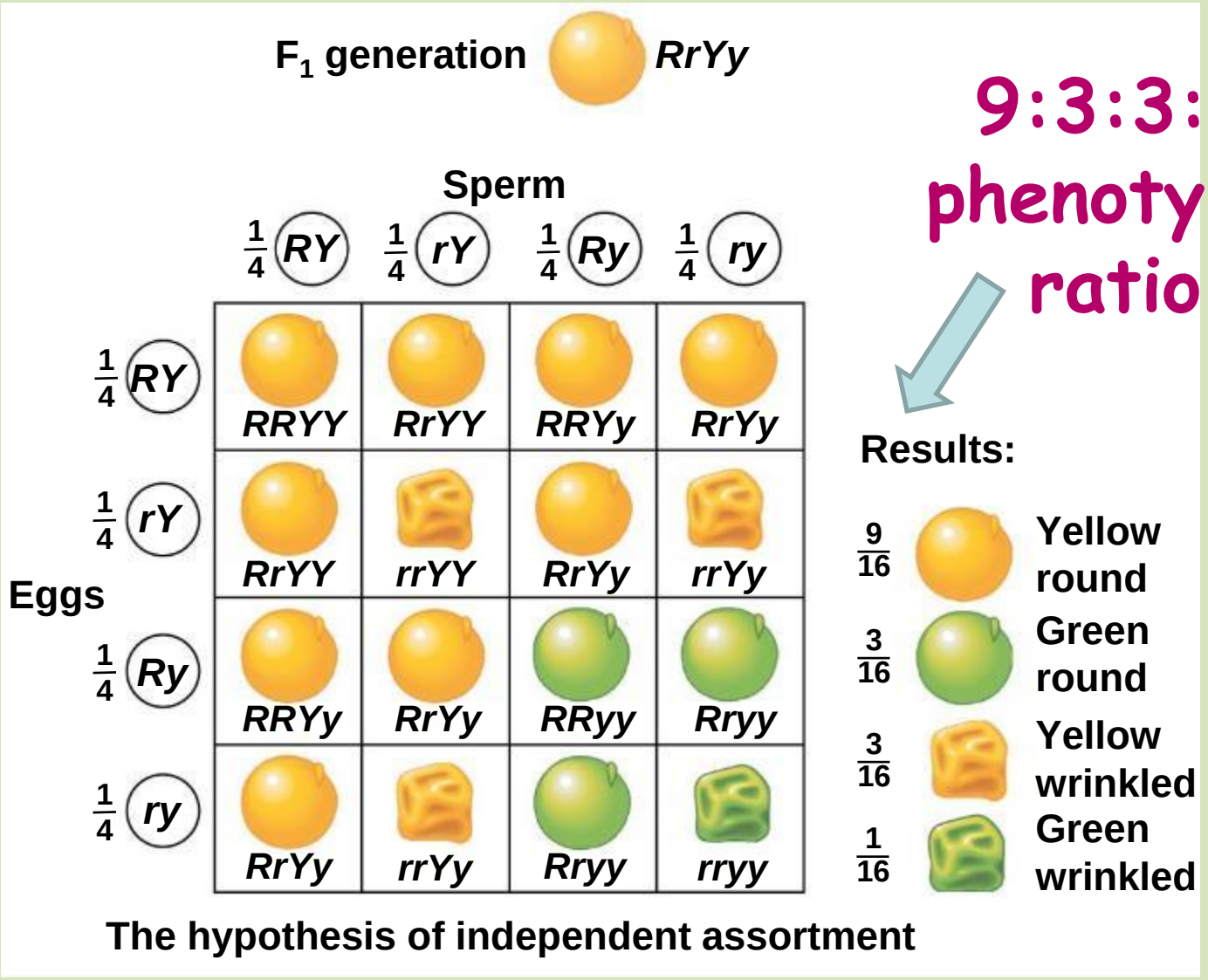
TRY IT



- ✓ When two traits are being considered (dihybrid cross), the Punnett square will need 16 squares.
- ✓ Each parent will pass one allele of each gene pair to the offspring.

TRY IT

| | RY | rY | Ry | ry |
|---|---|---|---|---|
| RY | RRYY | RrYY | RRYy | RrYy |
| rY | RrYY | rrYY | RrYy | rrYy |
| Ry | RRYY | RrYy | RRyy | Rryy |
| ry | RrYy | rrYy | Rryy | rryy |


F1 generation
RrYy
Sperm
1/4 RY
1/4 rY
1/4 Ry
1/4 ry
Eggs
RRYY
RrYY
RRYy
RrYy
rrYY
rrYy
RRyy
Rryy
rryy
9:3:3:1 phenotypic ratio
Results:
9/16 Yellow round
3/16 Green round
3/16 Yellow wrinkled
1/16 Green wrinkled
The hypothesis of independent assortment

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

**$F_1$ generation:** RrYy (Round yellow) x RrYy (Round Yellow)

| Gametes | RY | Ry | rY | ry |
|---|---|---|---|---|
| **RY** | RRYY<br>Round yellow | RRYy<br>Round yellow | RrYY<br>Round yellow | RrYy<br>Round yellow |
| **Ry** | RRYy<br>Round yellow | Rryy<br>Round green | RrYy<br>Round yellow | Rryy<br>Round green |
| **rY** | RrYy<br>Round yellow | RrYy<br>Round yellow | rrYY<br>wrinkled yellow | rrYy<br>wrinkled yellow |
| **ry** | RrYy<br>Round yellow | Rryy<br>Round green | rrYy<br>wrinkled yellow | rryy<br>wrinkled green |

**$F_2$ Generation**

*Predictions*

| | |
|---|---|
| 9 | Two dominant traits |
| 3 | 1 dominant & 1 recessive trait |
| 3 | 1 dominant & 1 recessive trait |
| 1 | Two recessive traits |

**$F_2$ Generation**

*Actual results*

| | |
|---|---|
| 315 | Round Yellow seeds |
| 101 | Wrinkled Yellow seeds |
| 108 | Round Green seeds |
| 32 | Wrinkled Green seeds |

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

- A **Dihybrid Cross** is a mating of parental varieties that differ in two characters.
- Mendel performed the following dihybrid cross with the following results:
  - **P Generation**: round yellow seeds × wrinkled green seeds
  - **$F_1$ Generation**: all plants with round yellow seeds
  - **$F_2$ Generation**:
    - **9/16** had round yellow seeds
    - **3/16** had wrinkled yellow seeds
    - **3/16** had round green seeds
    - **1/16** had wrinkled green seeds

# The LAW OF INDEPENDENT ASSORTMENT is revealed by tracking Two Characters at once

Mendel needed to explain why the $F_2$ offspring

- had new nonparental combinations of traits and
- had a **9:3:3:1 phenotypic ratio**.

Mendel

- suggested that the **inheritance of one character has no effect on the inheritance of another**.
- suggested that the dihybrid cross is the equivalent to two monohybrid crosses.
- established the **LAW of INDEPENDENT ASSORTMENT**.

# SUMMARY OF MENDEL'S PRINCIPLES


- **Mendel's Principles** form the basis of **Modern Genetics** include the following:

1. The inheritance of traits is determined by individual units known as ____.
2. ____ are passed from parent to offspring.
3. Each gene has two or more forms called ____.
4. Some alleles are ____, while other alleles are ____.
5. Each parent has ____ alleles for a particular trait that they inherited from their parents. They will pass ____ allele to their offspring when the alleles are segregated into ____.
6. The alleles for one trait segregate ____ of the alleles for another trait.

# SUMMARY OF MENDEL'S PRINCIPLES


- **Mendel's Principles** form the basis of **Modern Genetics** include the following:

1. The inheritance of traits is determined by individual units known as **genes.**
2. **Genes** are passed from parent to offspring.
3. Each gene has two or more forms called **alleles**.
4. Some alleles are **dominant**, while other alleles are **recessive**.
5. Each parent has **two** alleles for a particular trait that they inherited from their parents. They will pass **one** allele to their offspring when the alleles are segregated into **gametes**.
6. The alleles for one trait segregate **independently** of the alleles for another trait.