BASIC PRINCIPALS OF GENETICS

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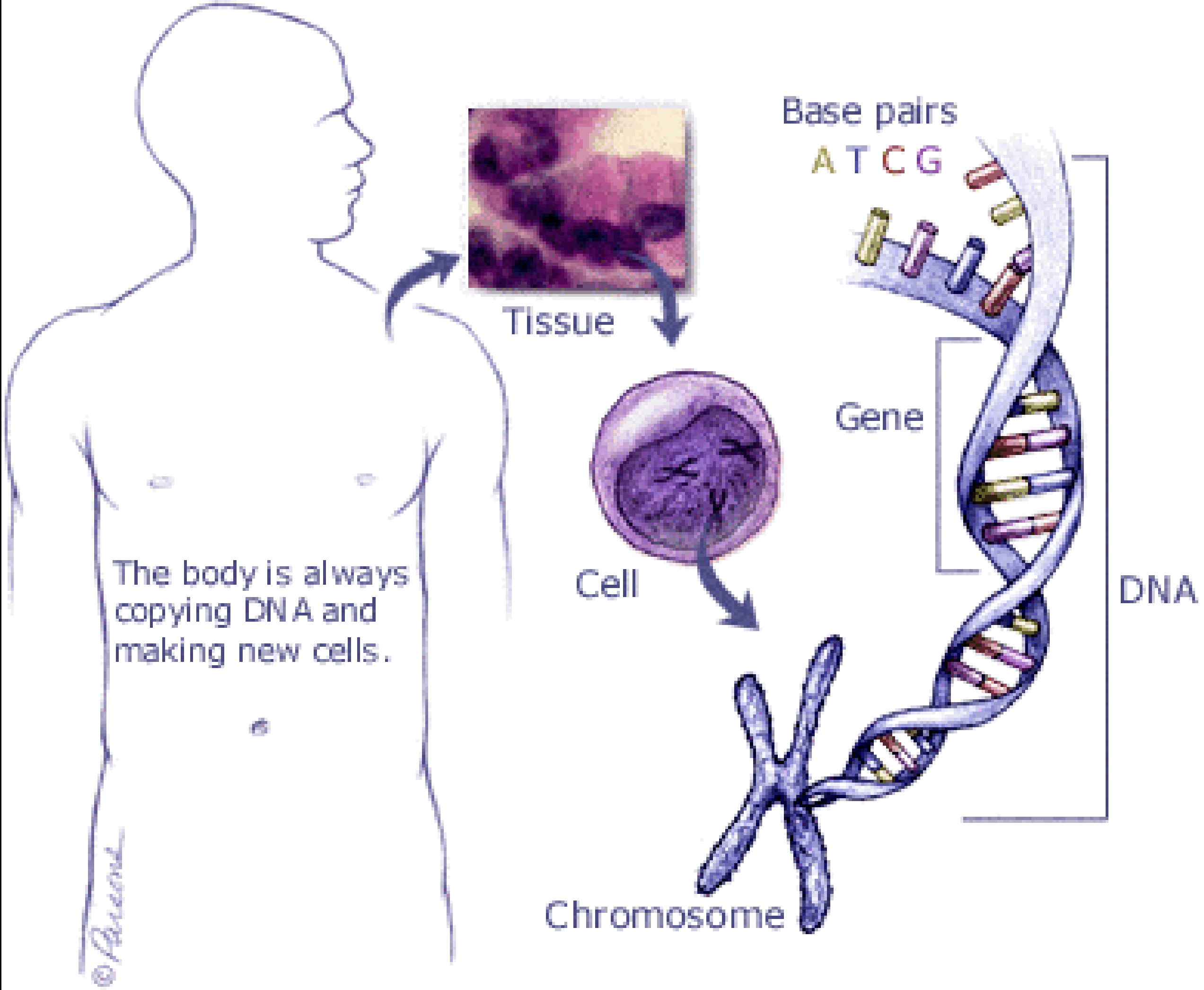
GENETICS

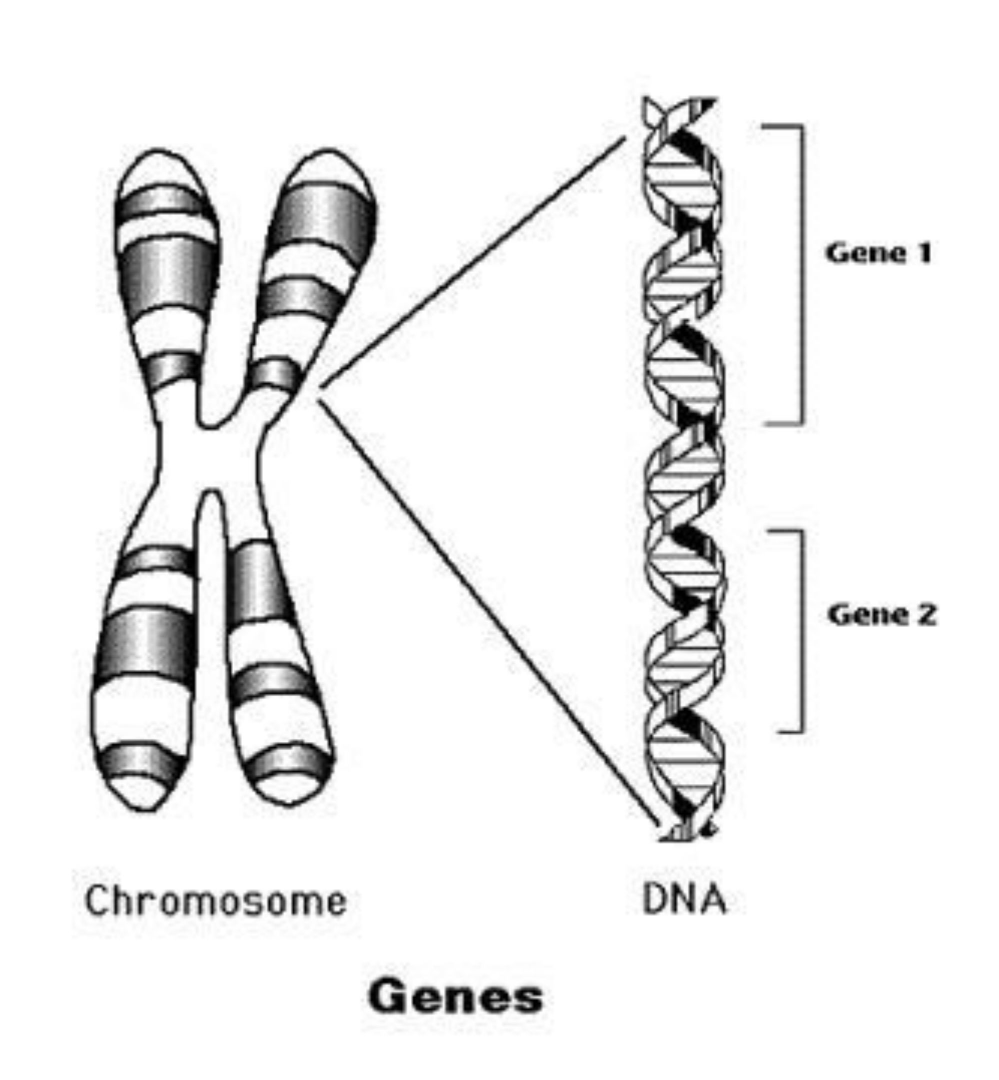
- Genetics is the study of how traits are passed from one generation to the next.
  - Traits are specific characteristics that vary from one individual to another.
    - Examples: hair color, height, intelligence.

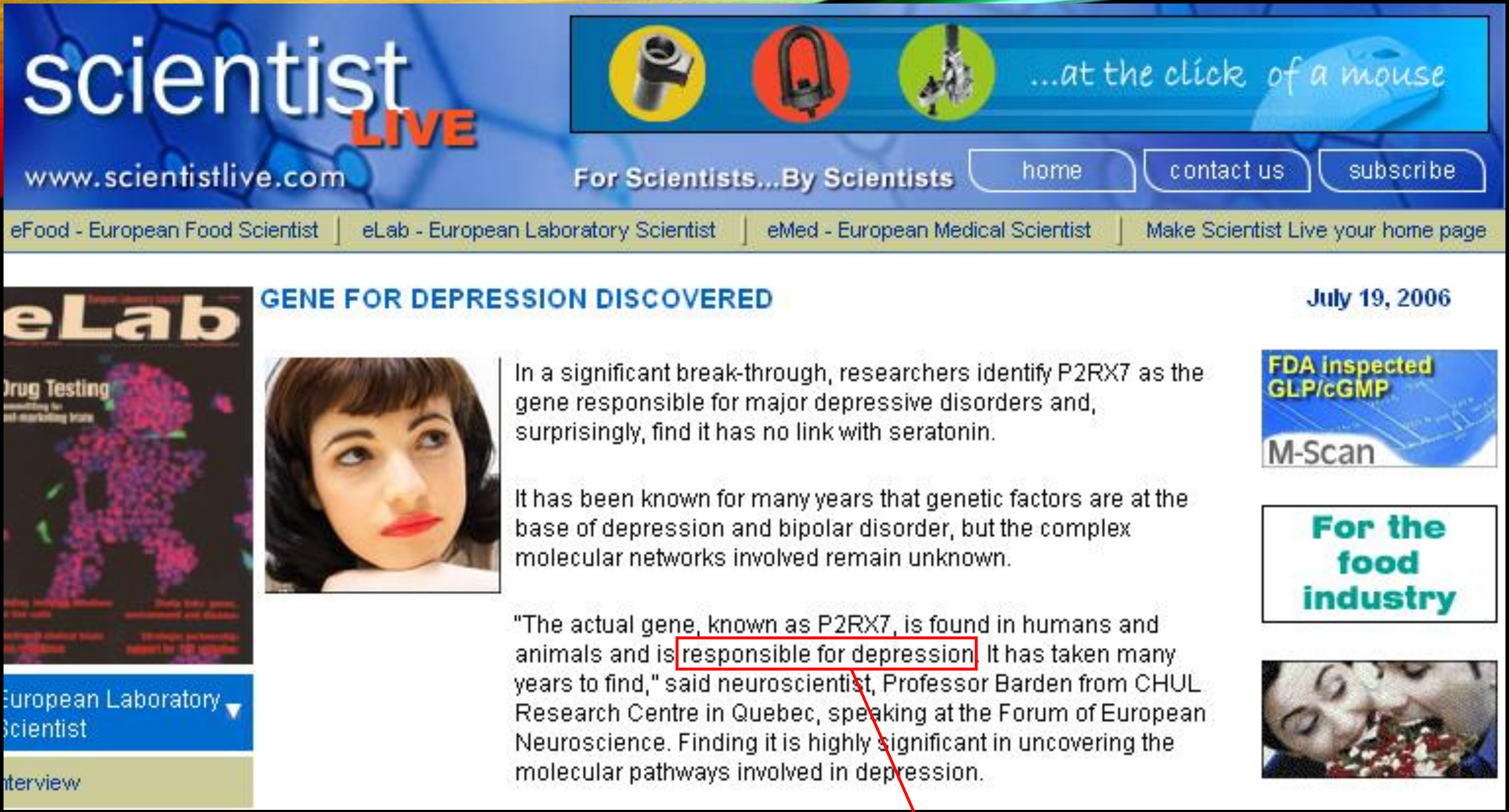
TRAITS

- Genetics – study of how <u>traits</u> are passed from <u>parent</u> to <u>offspring</u>

- Traits are determined by the genes on the chromosomes. A gene is a segment of DNA that determines a trait.







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GENE FOR DEPRESSION DISCOVERED

July 19, 2006

In a significant break-through, researchers identify P2RX7 as the gene responsible for major depressive disorders and, surprisingly, find it has no link with seratonin.

It has been known for many years that genetic factors are at the base of depression and bipolar disorder, but the complex molecular networks involved remain unknown.

"The actual gene, known as P2RX7, is found in humans and animals and is responsible for depression. It has taken many years to find," said neuroscientist, Professor Barden from CHUL Research Centre in Quebec, speaking at the Forum of European Neuroscience. Finding it is highly significant in uncovering the molecular pathways involved in depression.

	Frequency
Case	0.200
Control	0.165

Odds ratio: 1.26

1. **Eye-catching headline of the form "Gene for…"**

2. **Highly qualified factual paragraph**

BBC NEWS WORLD EDITION

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EDITIONS
Change to UK

Wednesday, 4 September, 2002, 00:47 GMT 01:47 UK

Gene for obsessive behaviour

Excessive hand washing is a symptom of OCD

Scientists have uncovered a gene which they believe may play a role in causing obsessive-compulsive disorder (OCD).

They hope that their work will allow earlier identification and treatment of people who are at greatest risk of developing the potentially debilitating condition.

They also hope it will lead to more effective therapies.

See also:

- 20 Dec 00 | Medical notes
  Obsessive-Compulsive Disorder
- 30 May 02 | Health
  Religious 'vulnerable to compulsion'
- 16 Aug 01 | Health
  Writer's cramp 'a sign of obsession'

Internet links:

- Obsessive-Compulsive Foundation
- Obsessive-Compulsive Disorder
- Molecular Psychiatry
- Obsessive Action

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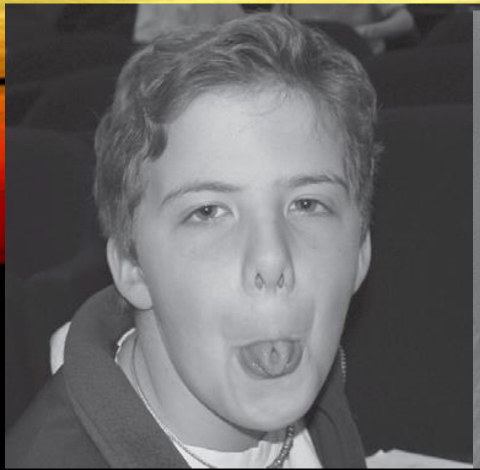
Top Health stories now:

- Heart risk link to big families
- Back pain drug 'may aid

WHAT MAKES YOU UNIQUE?

- Sure, we're all humans, but what makes you different from others in the room.
  - Your talents, interests or dreams?
  - Your personality, looks or clothes?
- Actually, one of the reasons you're unique is because of the genes you inherited from your parents.
  - Your genes make you unique, they have a hidden potential that can make you excel at things others can not.
  - Seek out your potential and you will find it, otherwise it will be wasted.



Tongue roller Non roller

Detached Earlobes

Attached Earlobes

Widow's Peak Straight Hair Line

Dimples

Cleft Chin

LET'S SEE THE CLASS RESULTS..

Let's calculate the frequency of each trait for our class:

$$\frac{\text{Number of students with the trait}}{\text{Number of students in the class}} \times 100 = \_\_\_\_\_\_\_\_\%$$

Example: Hitchhiker's thumb (Class size =30)

$$\frac{5 \times 100}{30} = 17\%$$

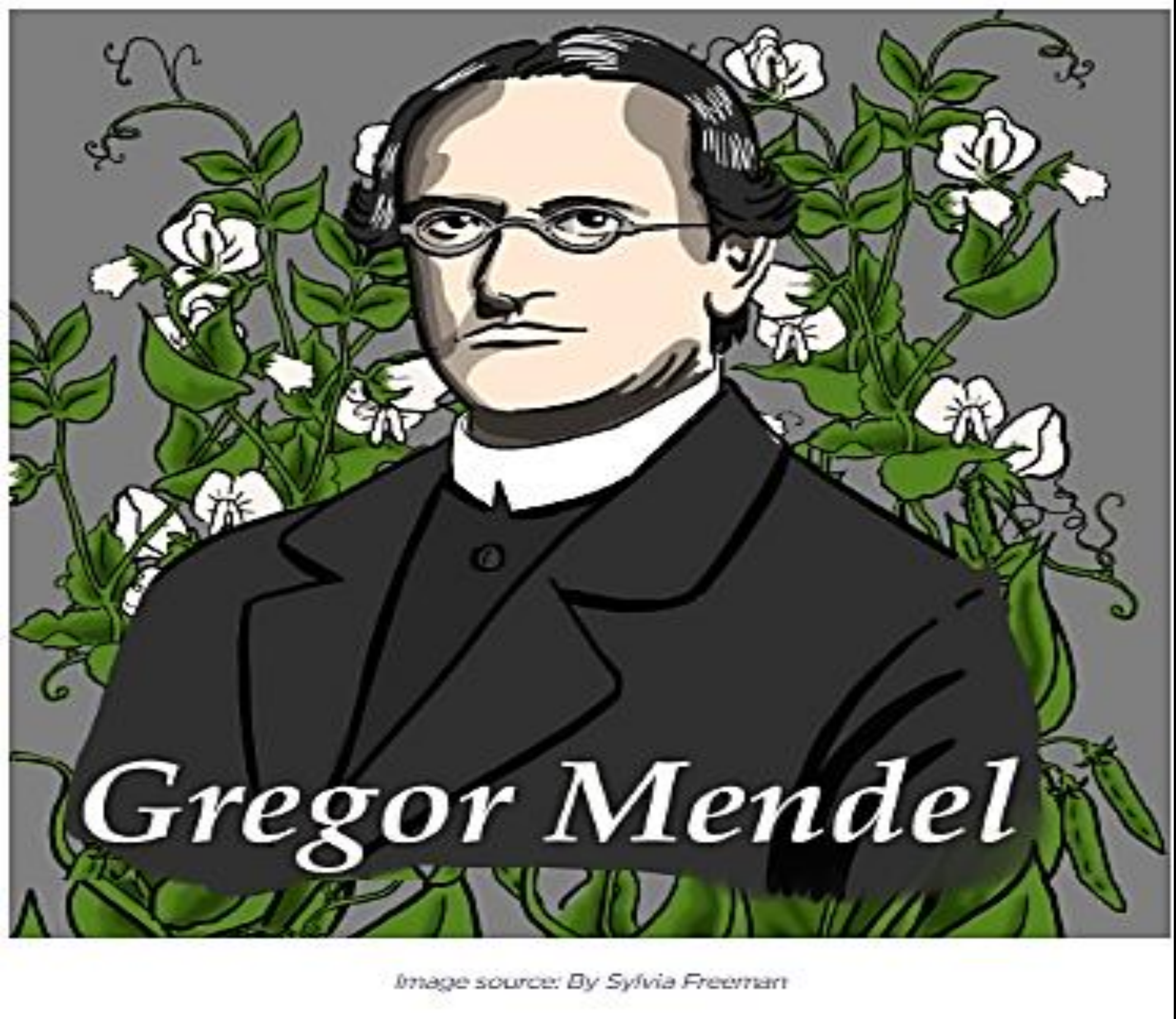
Let's compare the frequency of traits in the classroom population with the frequency in the general population.

Table 1: Frequencies of traits in the general population*

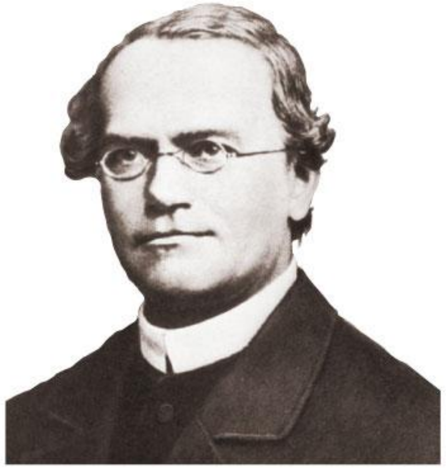
Trait	Frequencies
Gender	Female – 50% Male – 50%
Thumb extension	Straight thumb – 75% Hitchhiker's thumb – 25%
Tongue rolling	Can roll tongue – 70% Can not roll tongue – 30%
Handedness	Right handed – 93% Left handed – 7%
Hand clasping	Left thumb on top – 55% Right thumb on top – 44% No preference – 1%
Color vision	Normal females – almost 100% Colorblind females – less than 1% Normal males – 92% Colorblind males – 8%

requencies for traits are from Online Mendelian Inheritance In Man
ttp://www.ncbi.nlm.nih.gov/omim/).

THE FOUNDER OF MODERN GENETICS FOR HIS LAWS OF INHERITANCE.

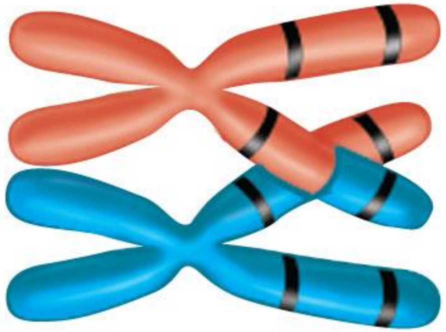


Chapter 1: Big Ideas

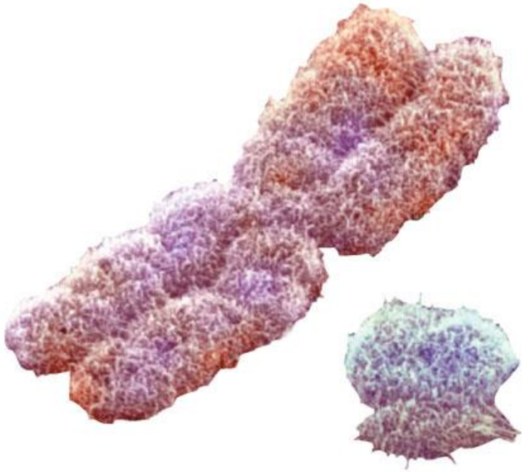


Mendel' s Laws

Variations on Mendel' s Laws



The Chromosomal Basis of Inheritance

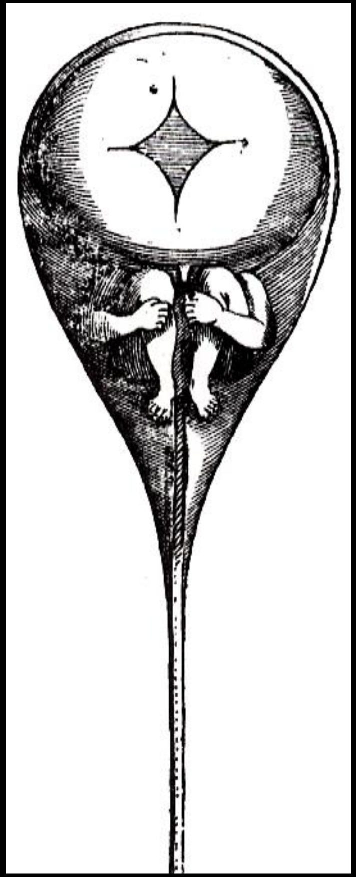


Chromosomes and Sex-Linked Genes

MENDEL'S LAWS

THE SCIENCE OF GENETICS HAS ANCIENT ROOTS

- Pangenesis, proposed around 400 BCE by Hippocrates, was an early explanation for inheritance that suggested that
  - particles called pangenes came from all parts of the organism to be incorporated into eggs or sperm and
  - characteristics acquired during the parents' lifetime could be transferred to the offspring.
- Aristotle rejected pangenesis and argued that instead of particles, the *potential* to produce the traits was inherited.



THE SCIENCE OF GENETICS HAS ANCIENT ROOTS

- The idea that hereditary materials mix in forming offspring, called the blending hypothesis, was
  - suggested in the 19th century by scientists studying plants but
  - later rejected because it did not explain how traits that disappear in one generation can reappear in later generations.

EXPERIMENTAL GENETICS BEGAN IN AN ABBEY GARDEN

- **Heredity** is the transmission of traits from one generation to the next.
- **Genetics** is the scientific study of heredity.
- **Gregor Mendel**
  - began the field of genetics in the 1860s,
  - deduced the principles of genetics by breeding garden peas, and
  - relied upon a background of mathematics, physics, and chemistry.

EXPERIMENTAL 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**.

EXPERIMENTAL 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**.
- A cross of F_1 plants produces an **F_2 generation**.

FIGURE 9.2B

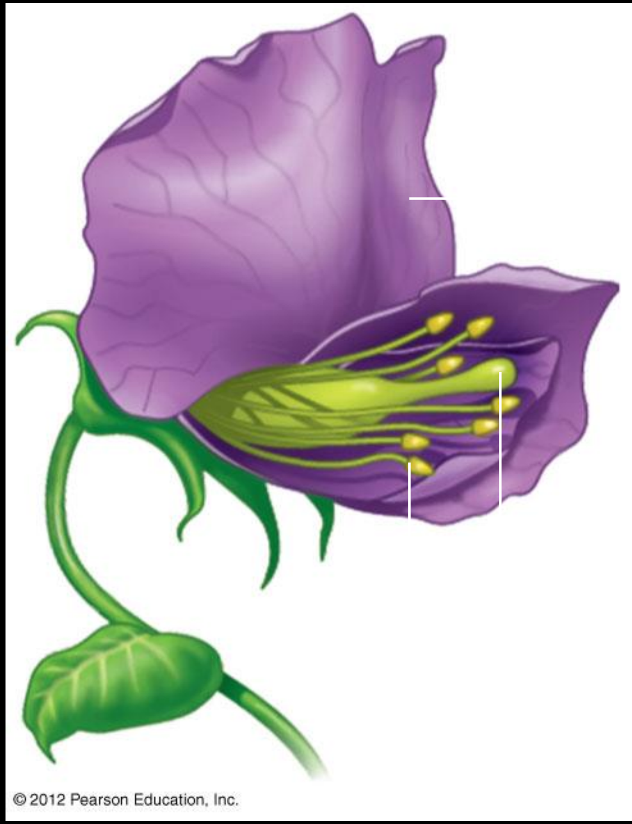


FIGURE 9.2C_S1

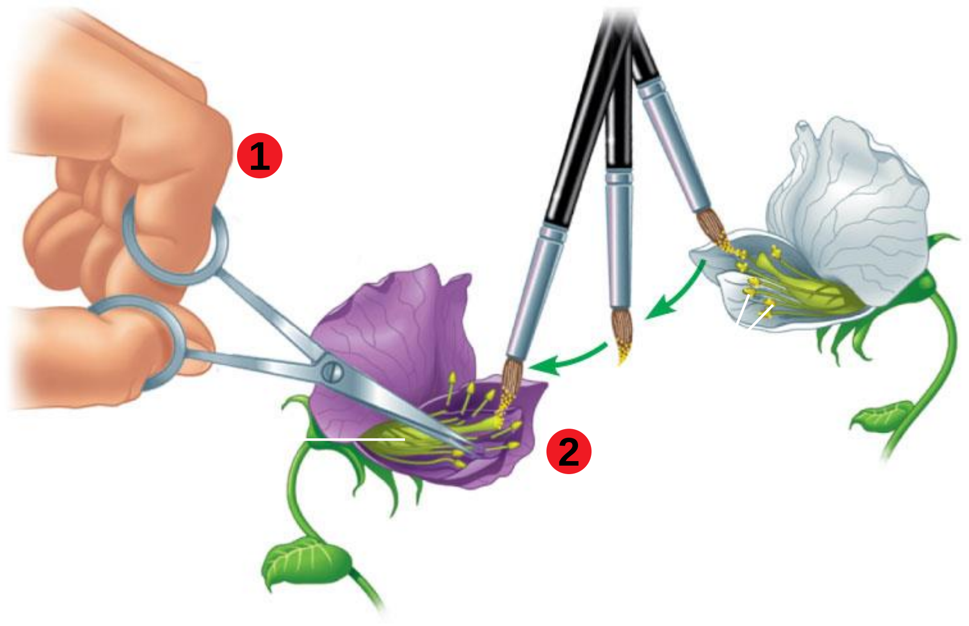


FIGURE 9.2C_S2

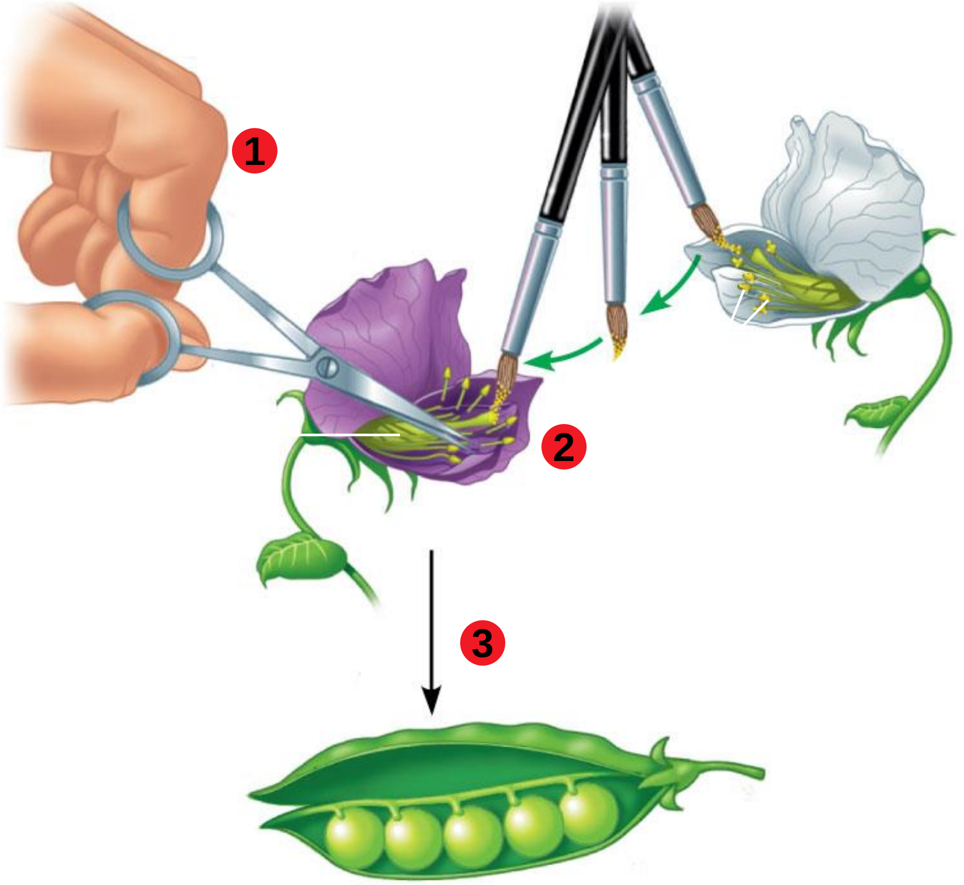
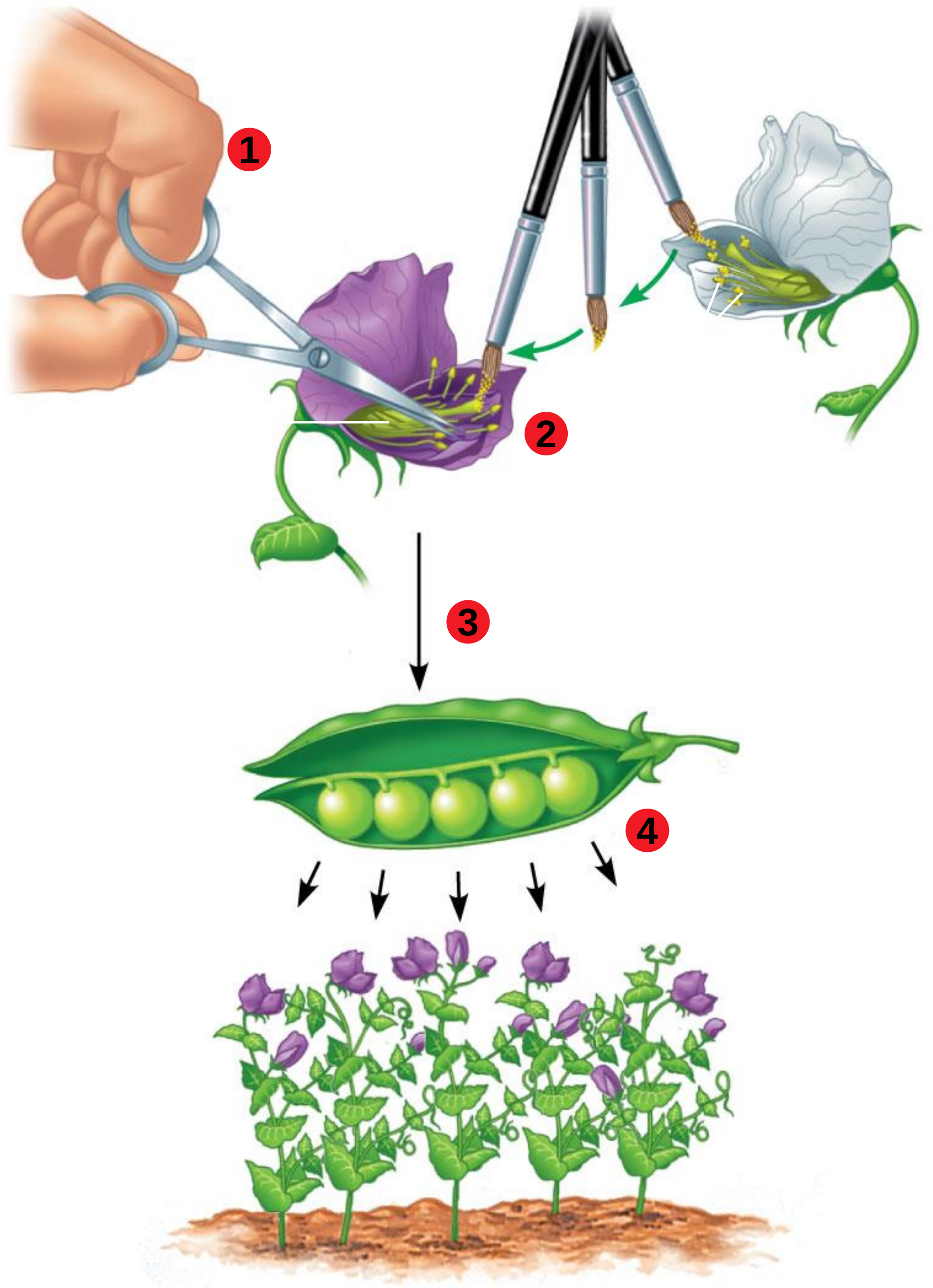


FIGURE 9.2C_S3



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

MENDEL'S LAW OF SEGREGATION DESCRIBES THE INHERITANCE OF A SINGLE CHARACTER

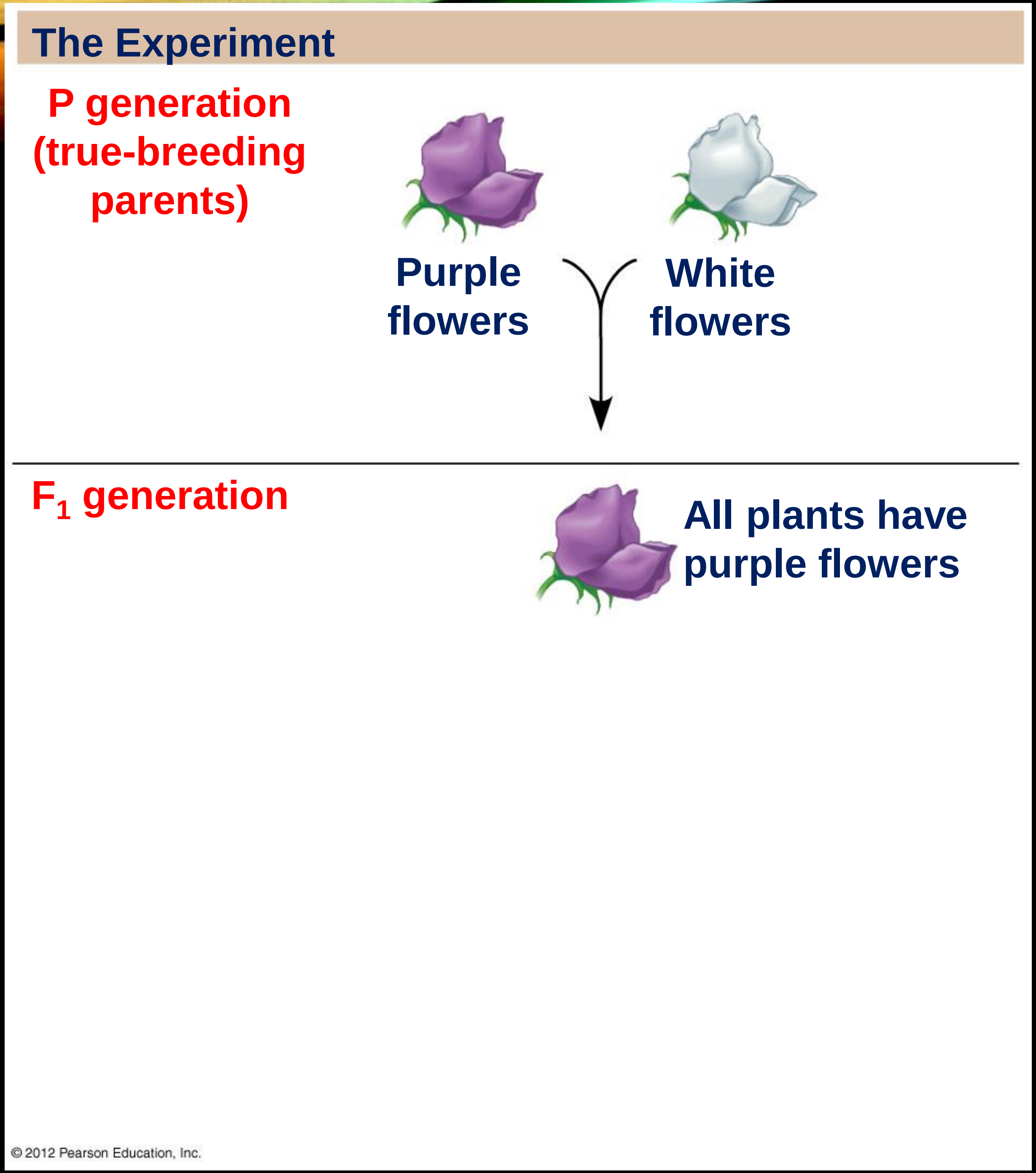
- A cross between two individuals differing in a single character is a **monohybrid cross**.
- Mendel performed a monohybrid cross between a plant with purple flowers and a plant with white flowers.
  - The F_1 generation produced all plants with purple flowers.
  - A cross of F_1 plants with each other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

The Experiment

P generation (true-breeding parents)

Purple flowers

White flowers


The Experiment
P generation (true-breeding parents)
Purple flowers
White flowers
F1 generation
All plants have purple flowers
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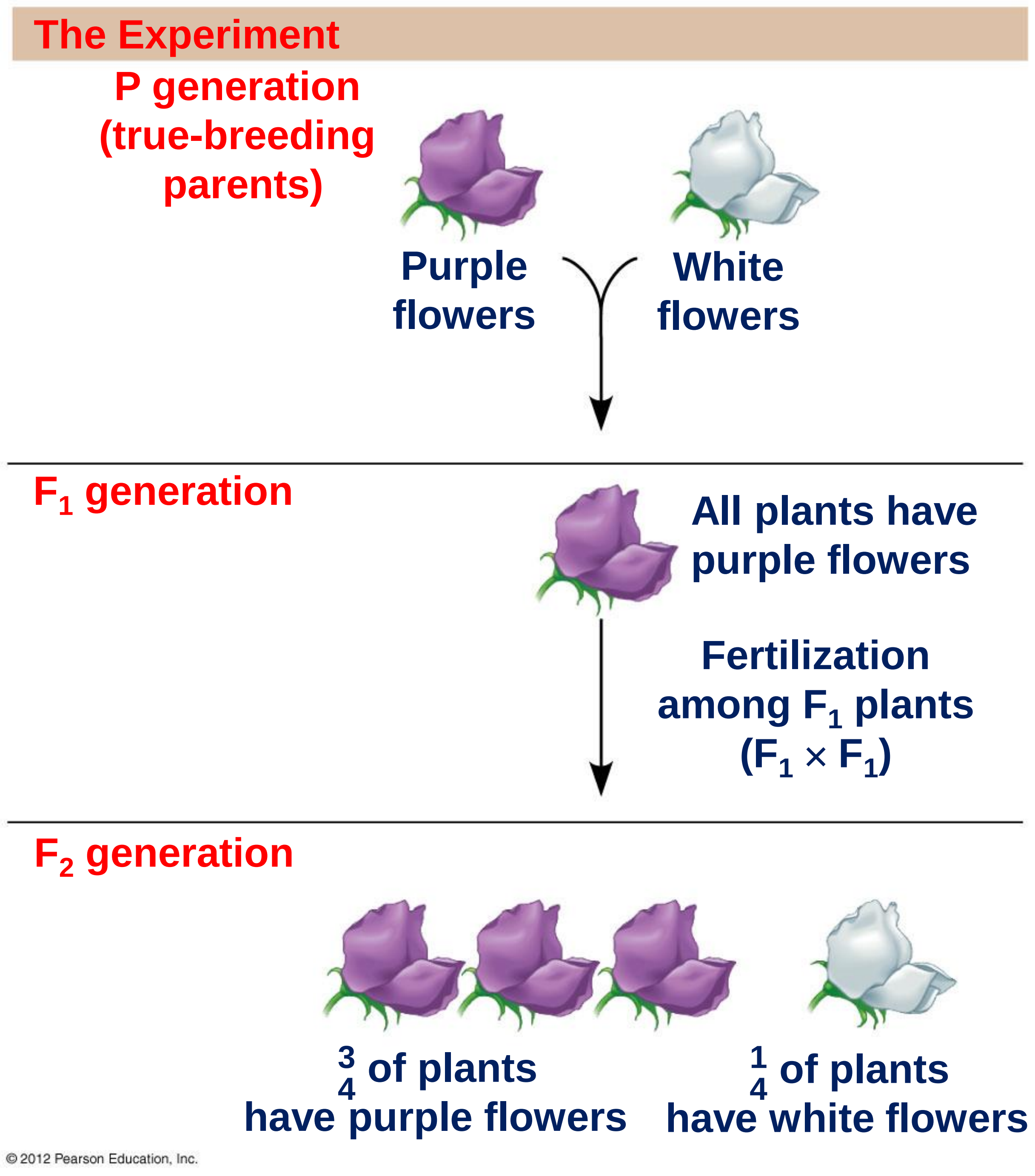
The Experiment
P generation (true-breeding parents)
Purple flowers
White flowers
F_1 generation
All plants have purple flowers
Fertilization among F_1 plants ($F_1 \times F_1$)
F_2 generation
$\frac{3}{4}$ of plants have purple flowers
$\frac{1}{4}$ of plants have white flowers
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MENDEL'S LAW OF SEGREGATION DESCRIBES THE INHERITANCE OF A SINGLE CHARACTER

- The all-purple F_1 generation did not produce light purple flowers, as predicted by the blending hypothesis.
- Mendel needed to explain why
  - white color seemed to disappear in the F_1 generation and
  - white color reappeared in one quarter of the F_2 offspring.
- Mendel observed the same patterns of inheritance for six other pea plant characters.

MENDEL'S LAW OF SEGREGATION DESCRIBES THE INHERITANCE OF A SINGLE CHARACTER

- Mendel developed four hypotheses, described below using modern terminology.
  1. Alleles are alternative versions of genes that account for variations in inherited characters.
  2. For each characteristic, 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 SEGREGATION DESCRIBES THE INHERITANCE OF A SINGLE CHARACTER

3. If the alleles of an inherited pair differ, then one determines the organism's appearance and is called the dominant allele. The other has no noticeable effect on the organism's appearance and is called the recessive allele.

- The phenotype is the appearance or expression of a trait.
- The genotype is the genetic makeup of a trait.
- The same phenotype may be determined by more than one genotype.

MENDEL'S LAW OF SEGREGATION DESCRIBES THE INHERITANCE OF A SINGLE CHARACTER

4. 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. This statement is called the law of segregation.

- Mendel's hypotheses also explain the 3:1 ratio in the F2 generation.
  - The F1 hybrids all have a Pp genotype.
  - A Punnett square shows the four possible combinations of alleles that could occur when these gametes combine.

FIGURE 9.3B_S1

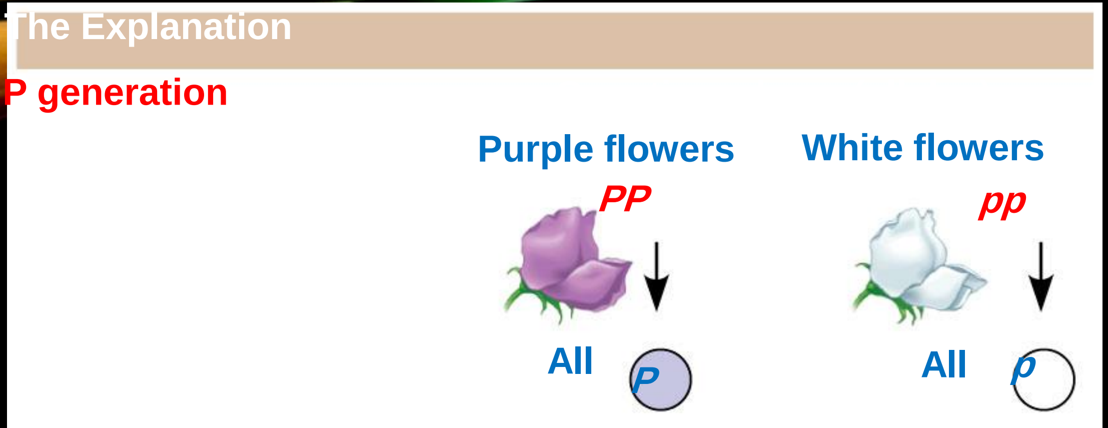


FIGURE 9.3B_S2

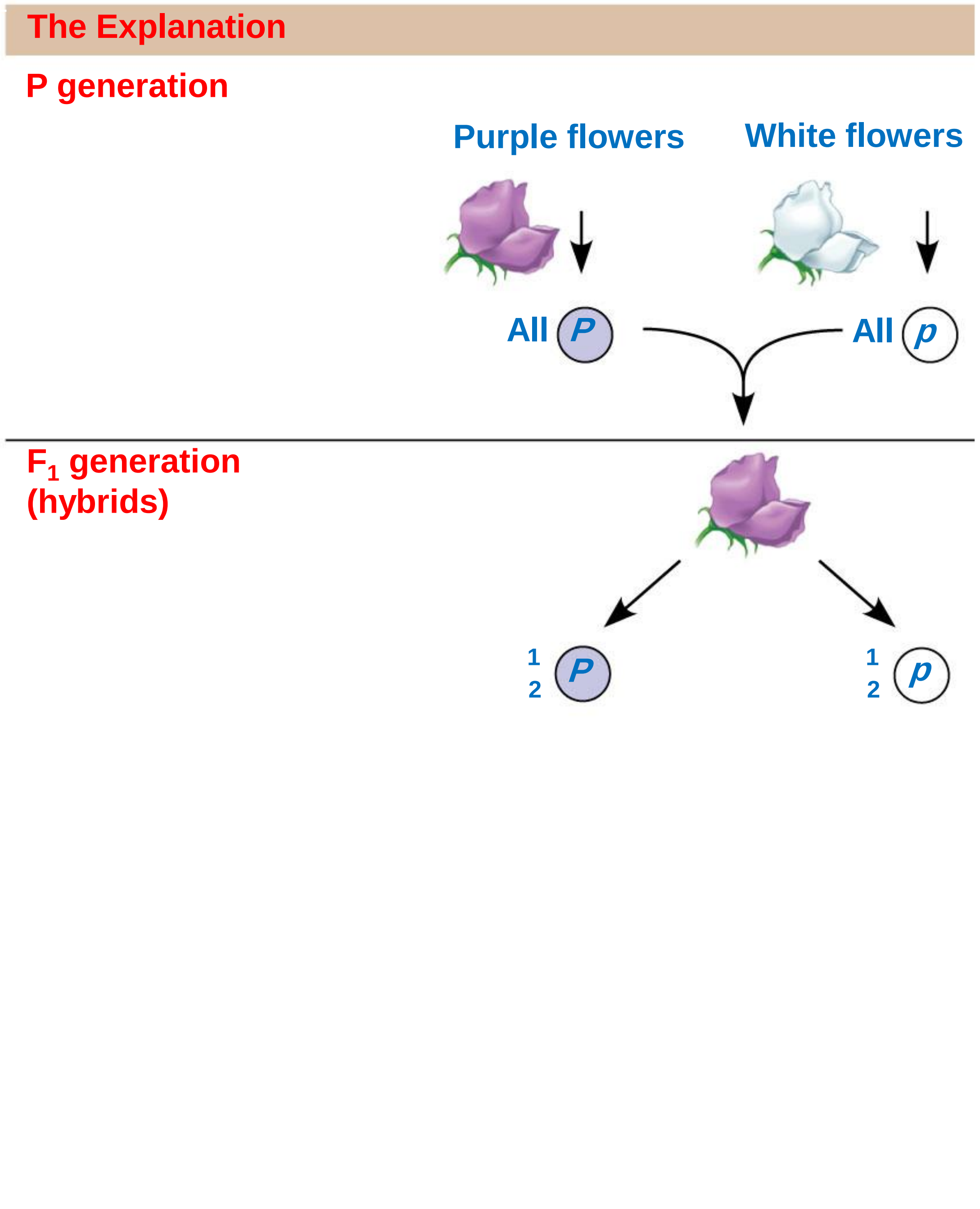
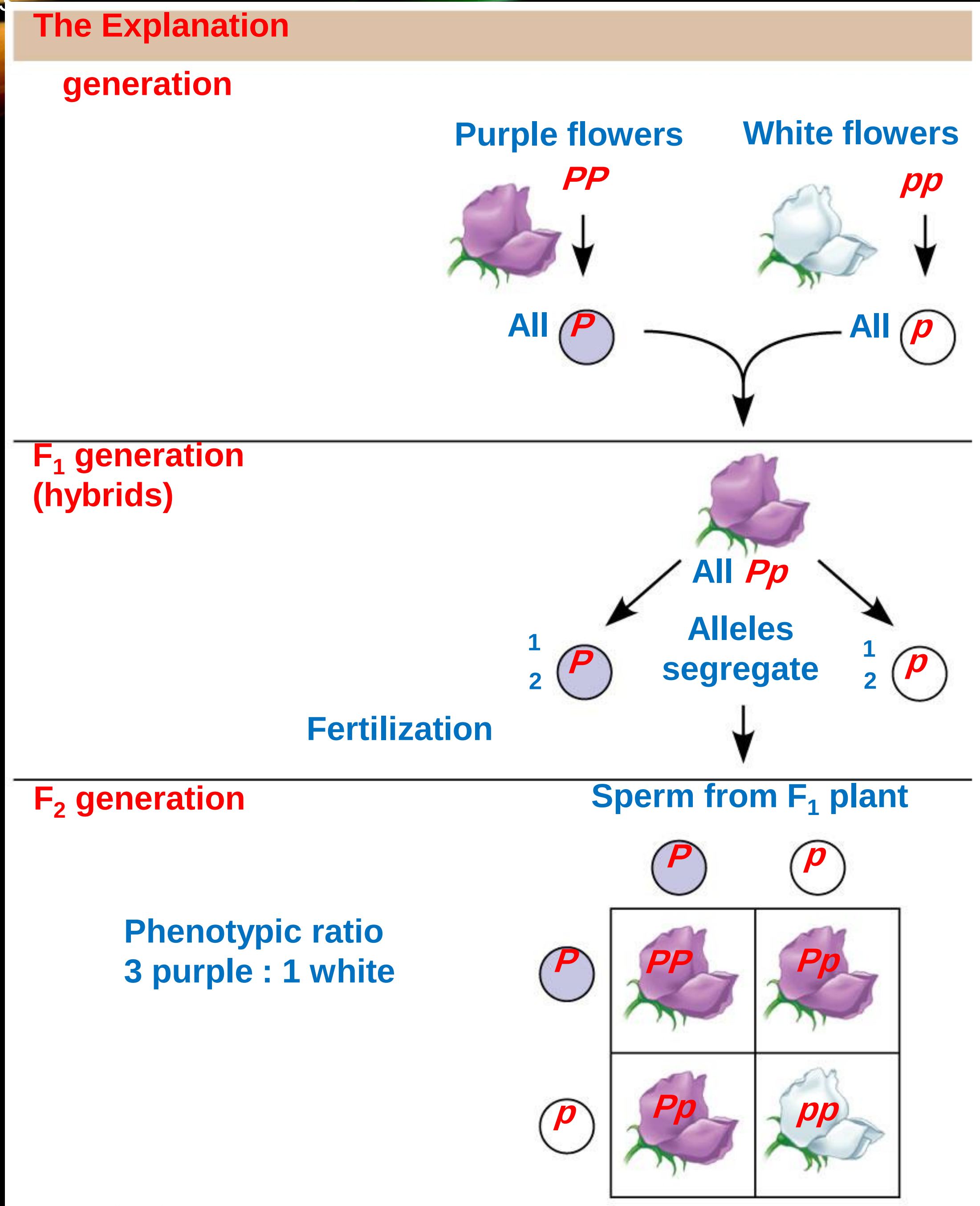
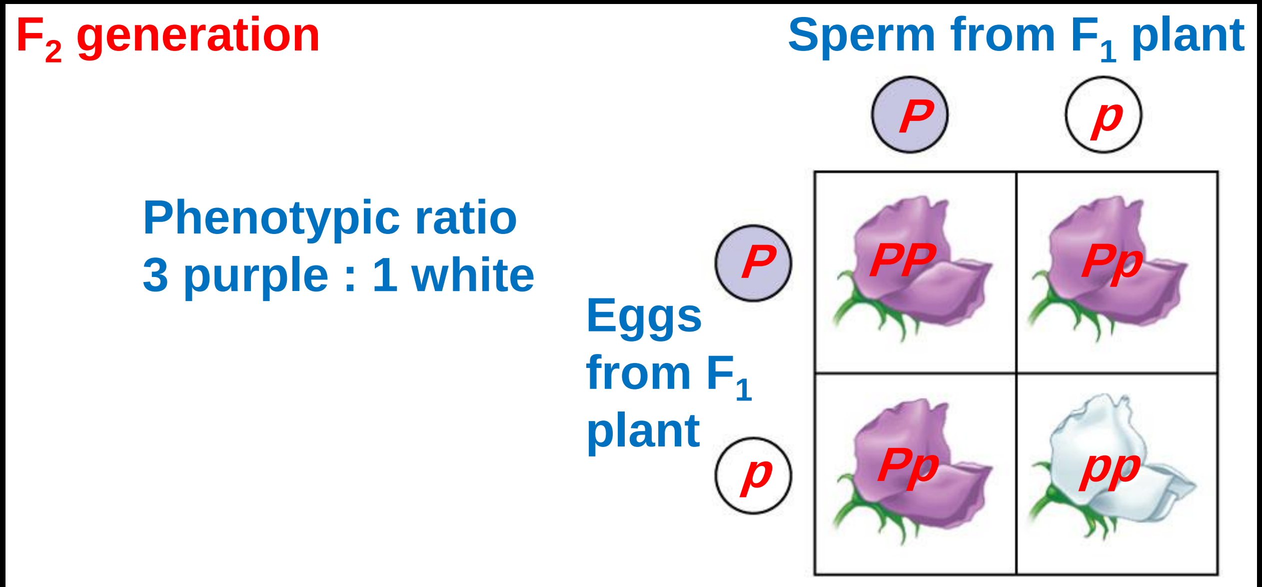


FIGURE 9.3B_S3

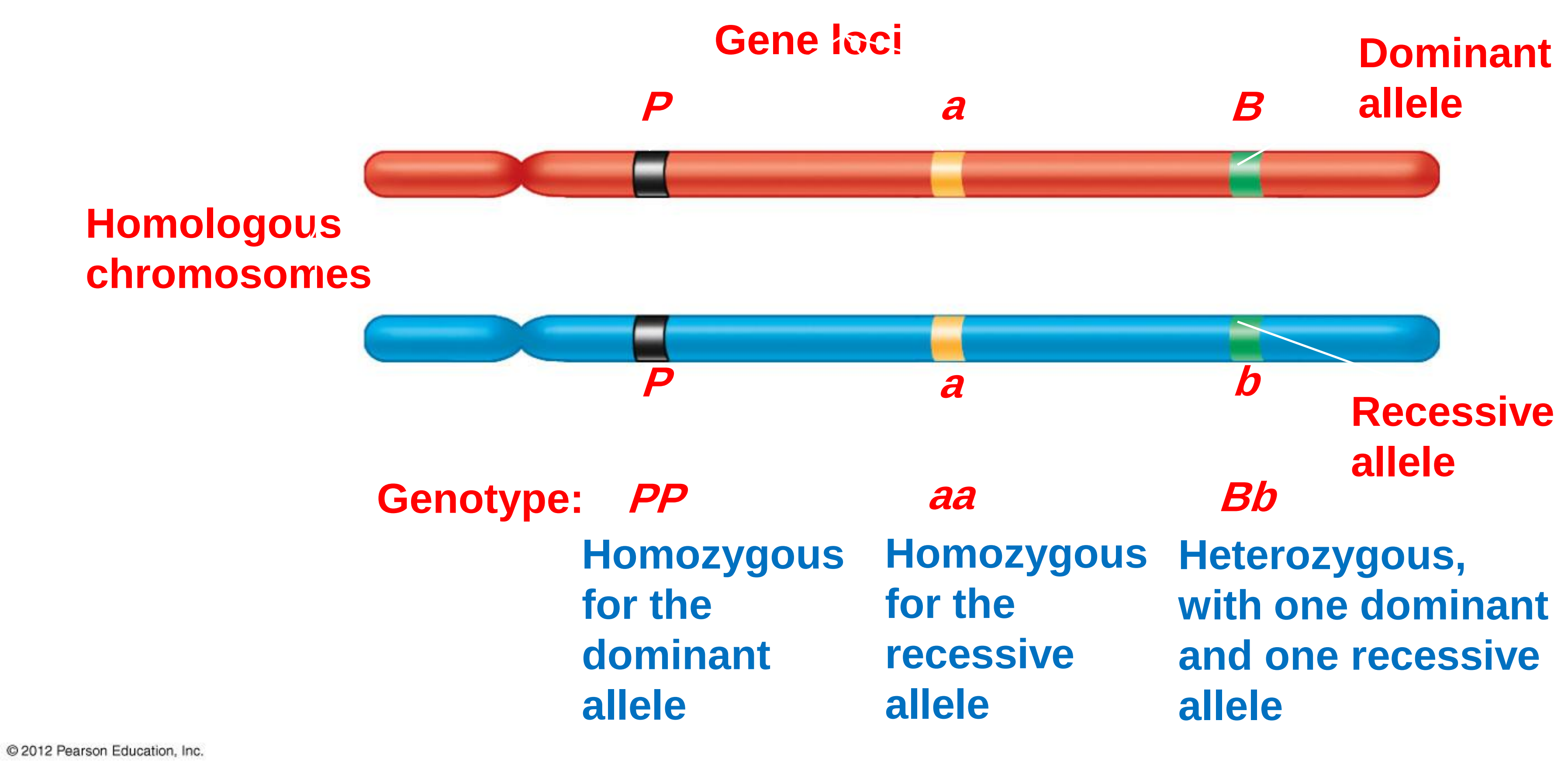




F2 generation
Sperm from F1 plant
P
p
Phenotypic ratio
3 purple : 1 white
Eggs from F1 plant
P
p
PP
Pp
Pp
pp

9.4 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, alleles of a gene reside at the same locus.**
  - **Homozygous individuals have the same allele on both homologues.**
  - **Heterozygous individuals have a different allele on each homologue.**


Gene loci
Dominant allele
P
a
B
Homologous chromosomes
P
a
b
Recessive allele
Genotype:
PP
aa
Bb
Homozygous for the dominant allele
Homozygous for the recessive allele
Heterozygous, with one dominant and one recessive allele
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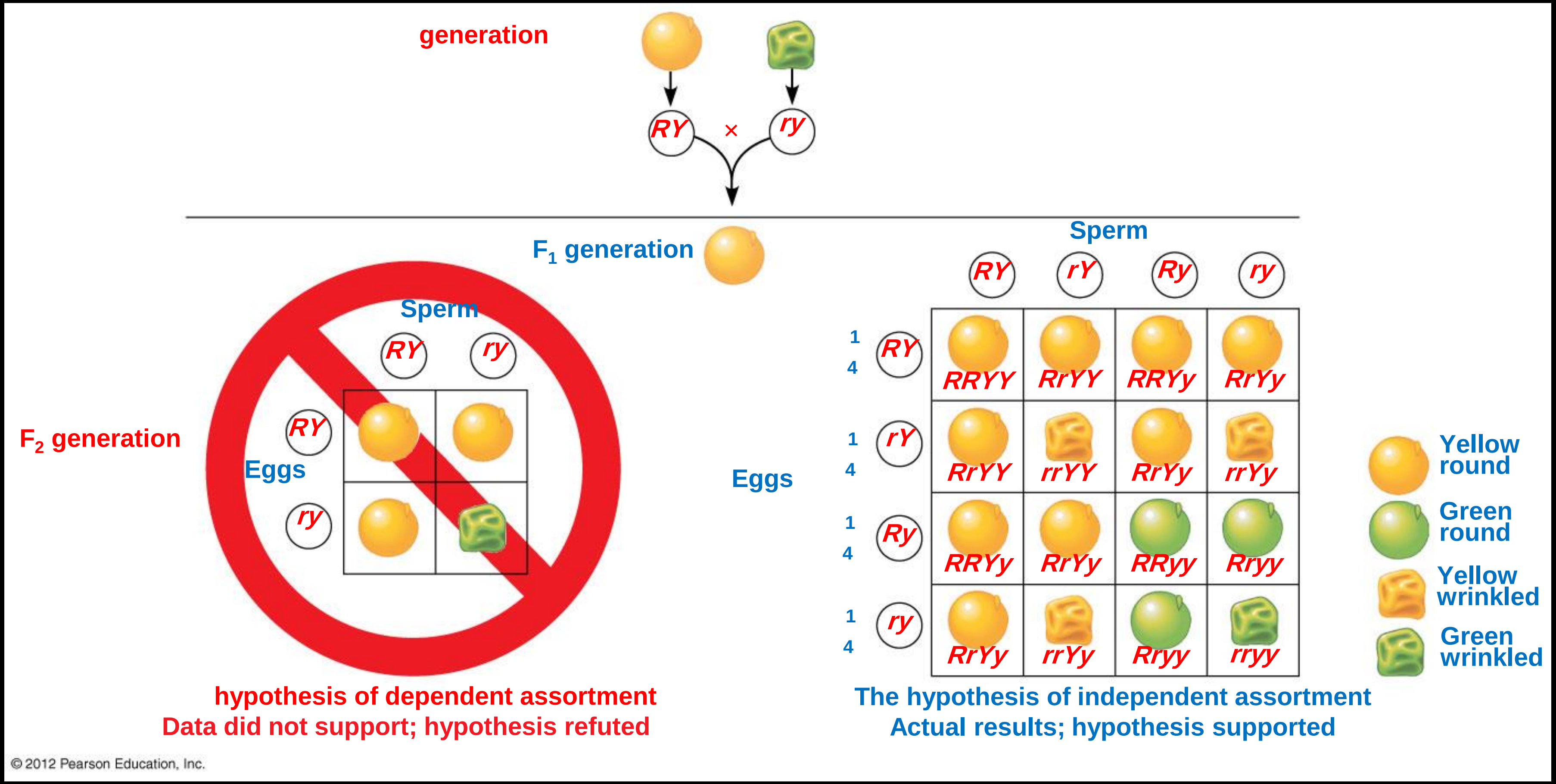
9.4 HOMOLOGOUS CHROMOSOMES BEAR THE ALLELES FOR EACH CHARACTER

- 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
  - F1 generation: all plants with round yellow seeds
  - F2 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

FIGURE 9.5A



generation
RY
×
ry
F1 generation
F2 generation
Sperm
RY
ry
Eggs
RY
ry
hypothesis of dependent assortment
Data did not support; hypothesis refuted
Sperm
RY
rY
Ry
ry
Eggs
1
4
RRYY
RrYY
RRYy
RrYy
RrYY
rrYY
RrYy
rrYy
RRYy
RrYy
RRyy
Rryy
RrYy
rrYy
Rryy
rryy
Yellow round
Green round
Yellow wrinkled
Green wrinkled
The hypothesis of independent assortment
Actual results; hypothesis supported
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MENDEL'S THREE LAWS OF INHERITANCE:

PRINCIPLE OF MENDELIAN INHERITANCE

Law of Segregation

The two alleles for each gene are placed in different gametes.

Law of Independent Assortment

The inheritance of one gene doesn't affect the inheritance of any other gene.

Law of Dominance

When two different alleles are present, only one is dominant and will be expressed.

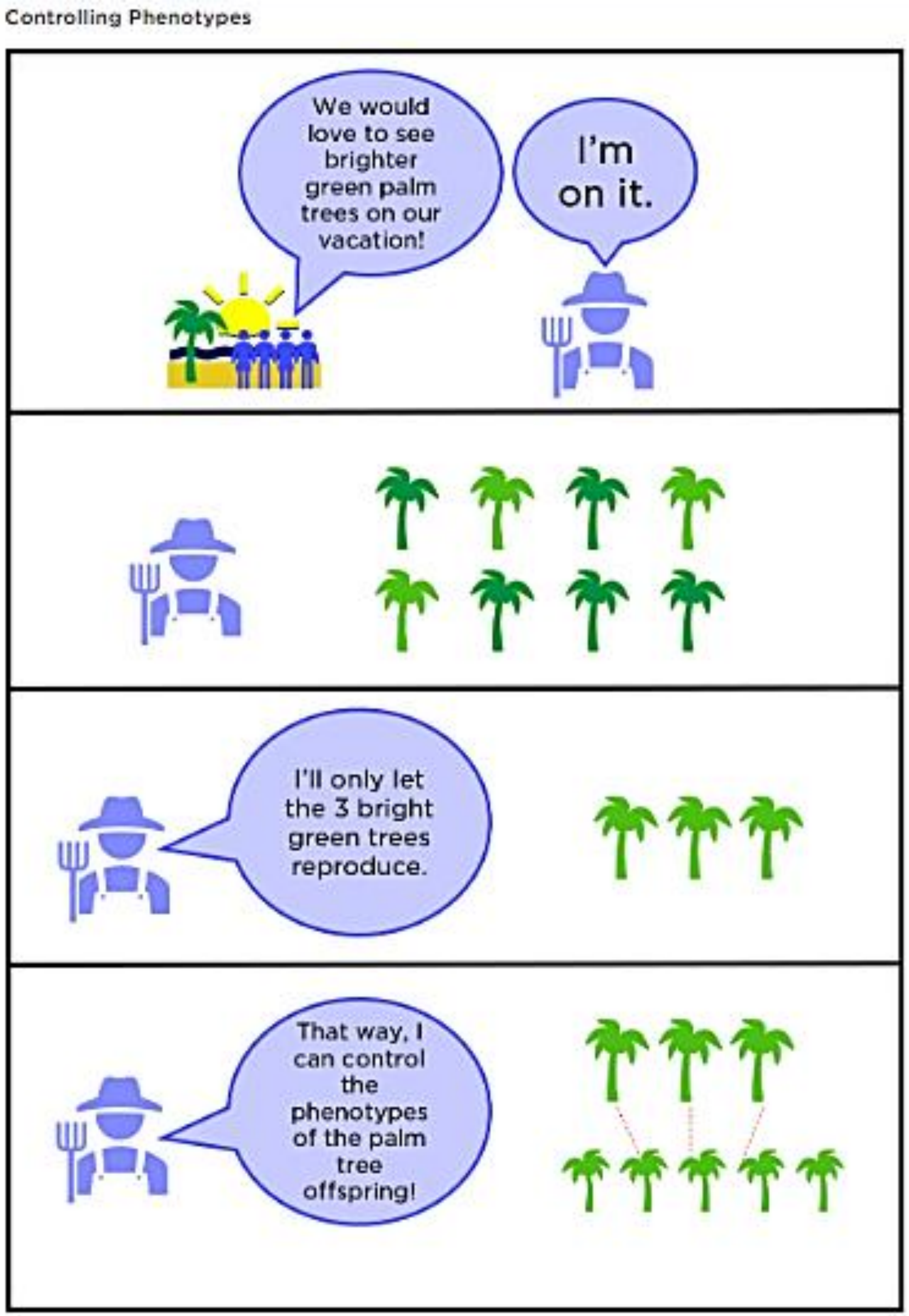
Mendel's Experiments and Laws: Dominance, Separation, and Independence

Mendel the monk's pea plant experiments paved the way for his Law of Independent Assortment. So, what did he do? What did he learn? And what did we learn from him?

1. **The Law of Dominance**: After crossing two pea plants with opposite traits and getting offspring that all looked identical to one another and to one parent, Mendel came up with a new question. Might those offspring still have the ability to pass on the other parent's traits? He crossed two of the offspring together and—aha—he found that some of these new offspring did look like the other grandparent. This information paved the way for our understanding of dominant alleles and recessive alleles today.

2. **The Law of Segregation**: When Mendel saw that those second-generation pea plants could still pass on the traits of their grandparents, he wondered how. He hypothesized that the things (alleles) that carry traits end up separating from each other when they're being passed on. In other words, Mendel said that alleles are separated in the parent to be passed on to the offspring. That way each kid gets one allele from each parent.

3. **The Law of Independent Assortment**: This is where Mendel worked on his dihybrid crosses. Mendel did some math and statistics here and found that a specific phenotypic ratio was present when you crossed pea plants and looked at two traits at once. Because of the math, he was able to hypothesize that if those genes didn't sort independently, you wouldn't see



WHAT'S SELECTIVE BREEDING?