General Biology 1
BIO1101
Syllabus & Textbook: http://goo.gl/rvgdrH

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Letter Grade	Numerical Ranges
A	93-100
A-	90-92.9
B+	87-89.9
B	83-86.9
B-	80-82.9
C+	77-79.9
C	70-76.9
D	60-69.9
F	59.9 and below

OER
Lecture:https://openlab.citytech.cuny.edu/bio-oer/page/2/
Lab: https://openlab.citytech.cuny.edu/bio-oer/

Grade Breakdown:
Exams (4): 20% Each
Quizzes: 20% Average

Recap: Lecture 23

1. <u>DNA Compaction/Condensation:</u>

Human genome spans 2 meters, fits into nucleus (5-8 µm diameter)

Active genes **500X** compacted, Inactive mitotic DNA (**10,000X**)

<u>2. Histone Core Complex: Octomer 8 Proteins (2- H2A, H2B, H3, H4)</u>

DNA double helix – 2 nm

Nucleosome "Beads on a string" – 11 nm (~200 bp)

147 bp (1.65 turns) around histone core – 6X compaction

Chromatin fiber – 30 nm (H1) – 40 X compaction

Loops (300 nm) – 500X compaction

Chromatid 700 nm – 10,000 X compaction

<u>3. Chromosome Organization:</u>

A. Chromosome vs chromatid

B. p arm, q arm, centromere

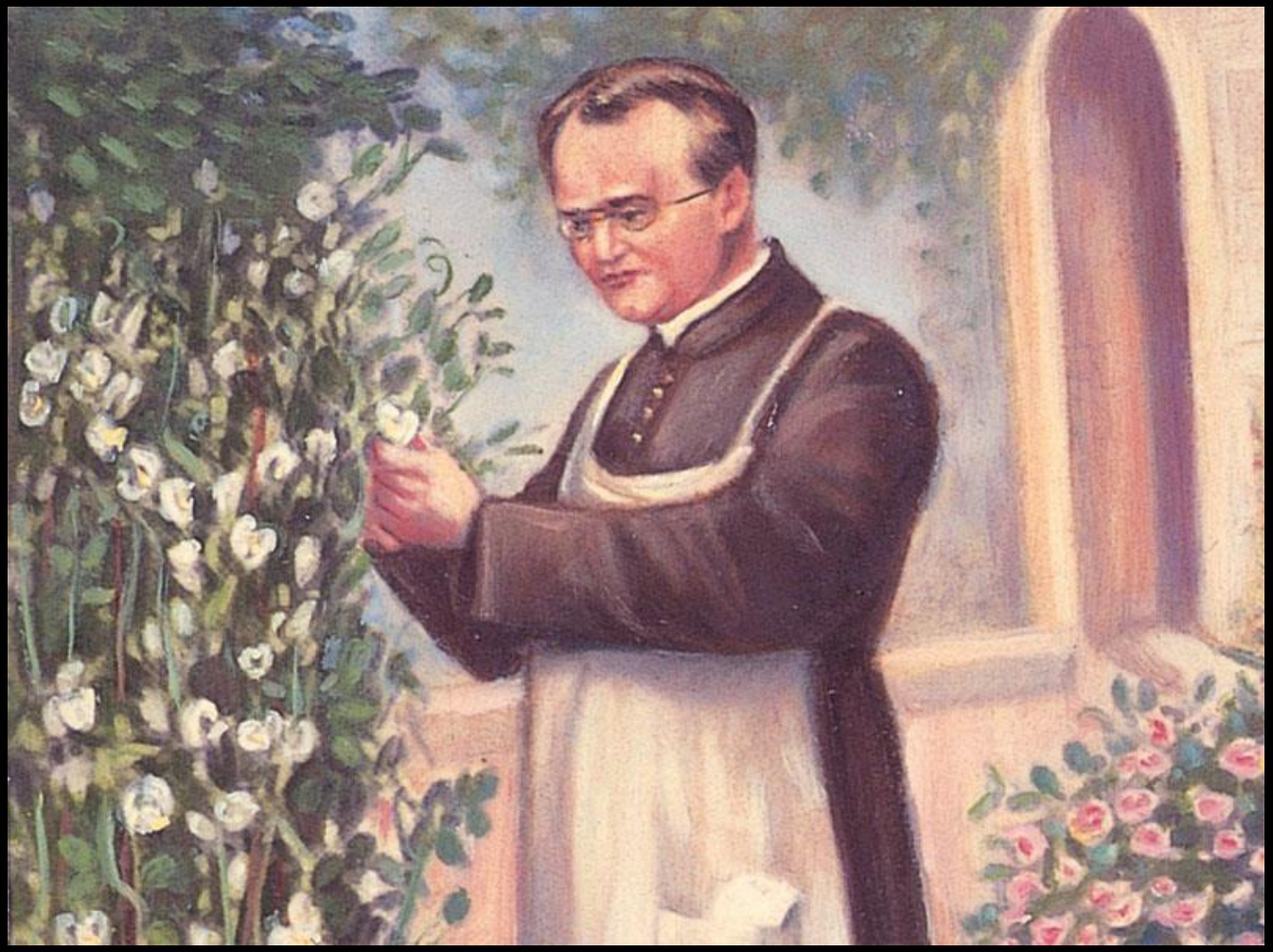
C.Ori (replication), Telomeres (stabilization) TTAGGG

D. Heterochromatin/Euchromatin

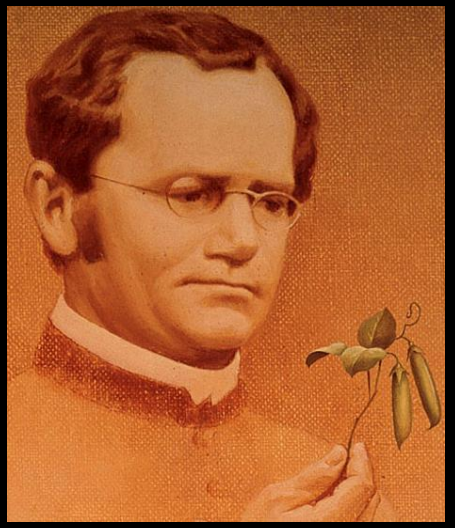
<u>4. Epigenetics</u>

1. Histone Code (M, A, P) – H3
2. X-chromosome inactivation (Barr bodies)

GENETICS



Gregor Mendel



- The “father” of modern Genetics (1850s and 1860s)
- Austrian monk
  - Studied science and mathematics at University of Vienna
  - Conducted breeding experiments with the garden pea *Pisum sativum*
  - Carefully gathered and documented mathematical data from his experiments
- Formulated fundamental **laws of heredity** in early 1860s
  - Had no knowledge of cells or chromosomes
  - Did not have a microscope

Blending Inheritance

- Theories of inheritance in Mendel's time:
  - Based on blending characteristics
  - Parents of contrasting appearance produce offsprings of intermediate appearance

- Mendel's findings were in contrast with this
  - He formulated the particulate theory of inheritance
  - Inheritance involves reshuffling of genes from generation to generation

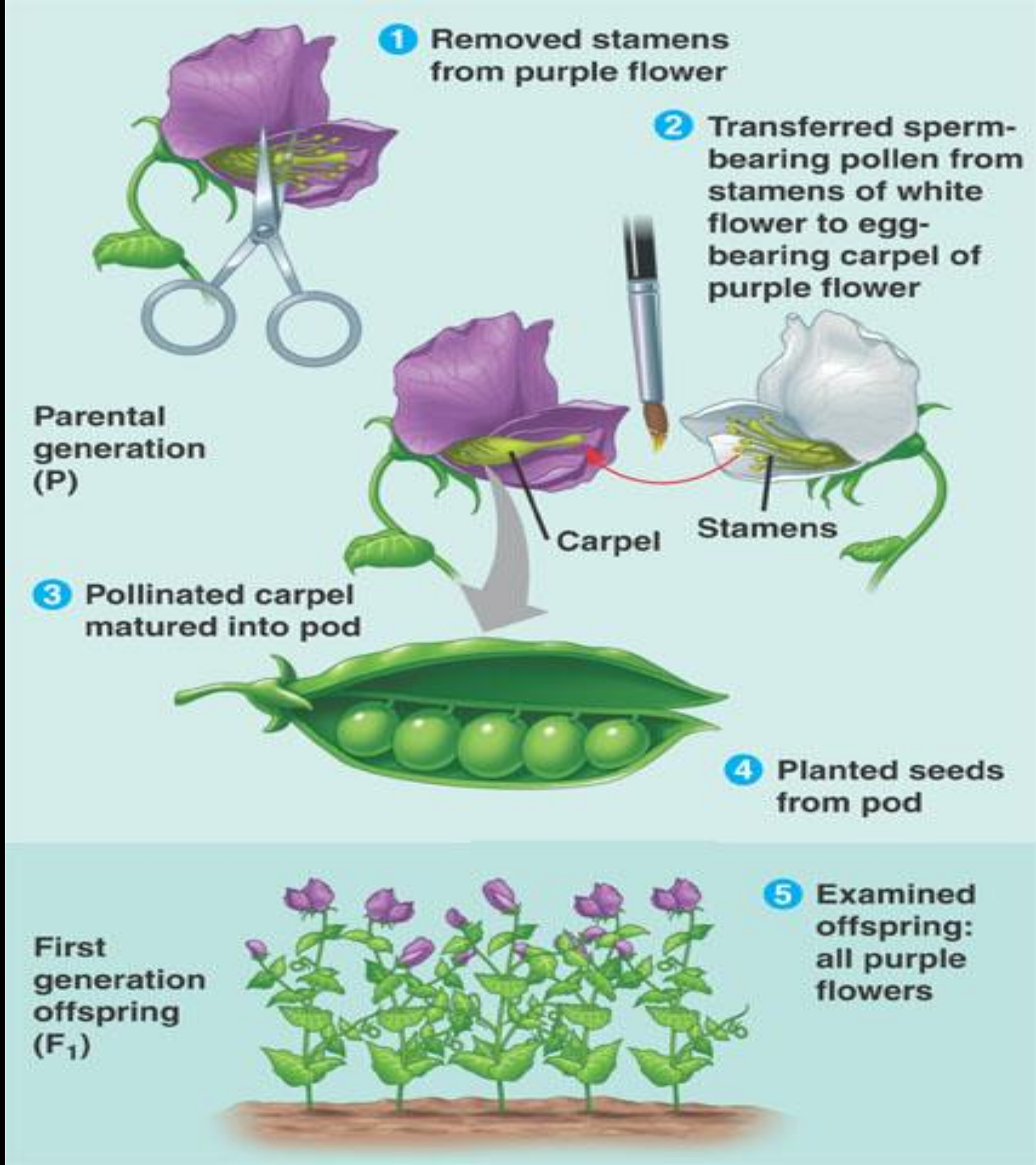
Mendel's experimental procedure

- Studied "heredity" by breeding pea plants
- Studied seven (7) traits and measured how they were passed on, generation by generation
- He envisioned "units" of inheritance, now known as **genes**.
- **Character** – a "heritable" feature that varies among individuals.
  - e.g. flower color, height
- **Trait** – each variant of a character (e.g. purple flowers, tall plants)
- **True-breeding (homozygous)** – plants that self-pollinate have NO variation for a particular trait

One-Trait Inheritance

- Mendel performed **cross-breeding** experiments
  - Used “true-breeding” (homozygous) plants
  - Chose varieties that differed in only one trait (monohybrid cross)
  - Performed reciprocal crosses
    - Parental generation = P
    - First filial generation offspring = F_1
    - Second filial generation offspring = F_2
  - Formulated the **Law of Segregation**
    - **How genes are passed during mating**

Mendel's Monohybrid Crosses: An Example

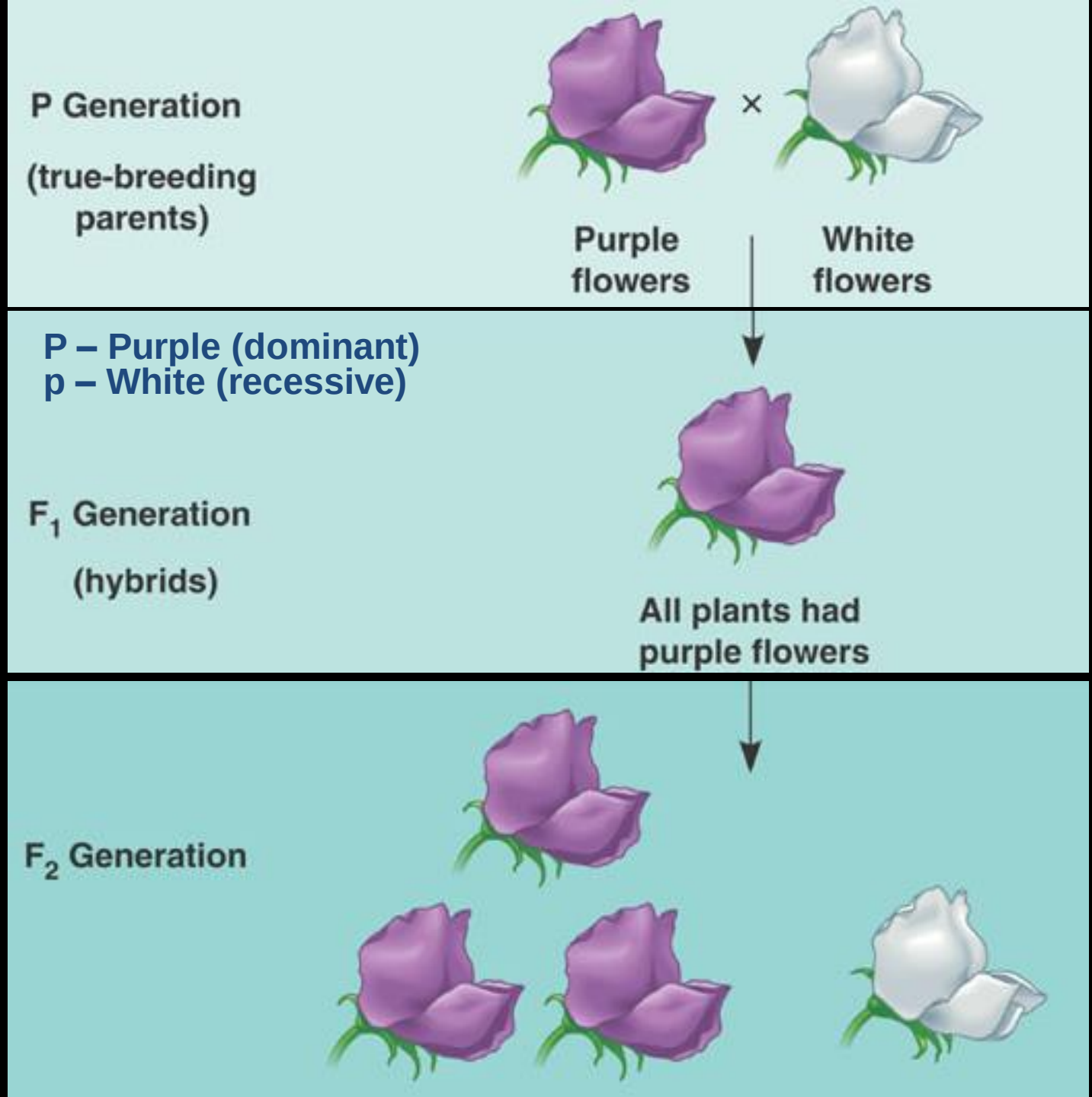


Mendel's Monohybrid Crosses: An Example

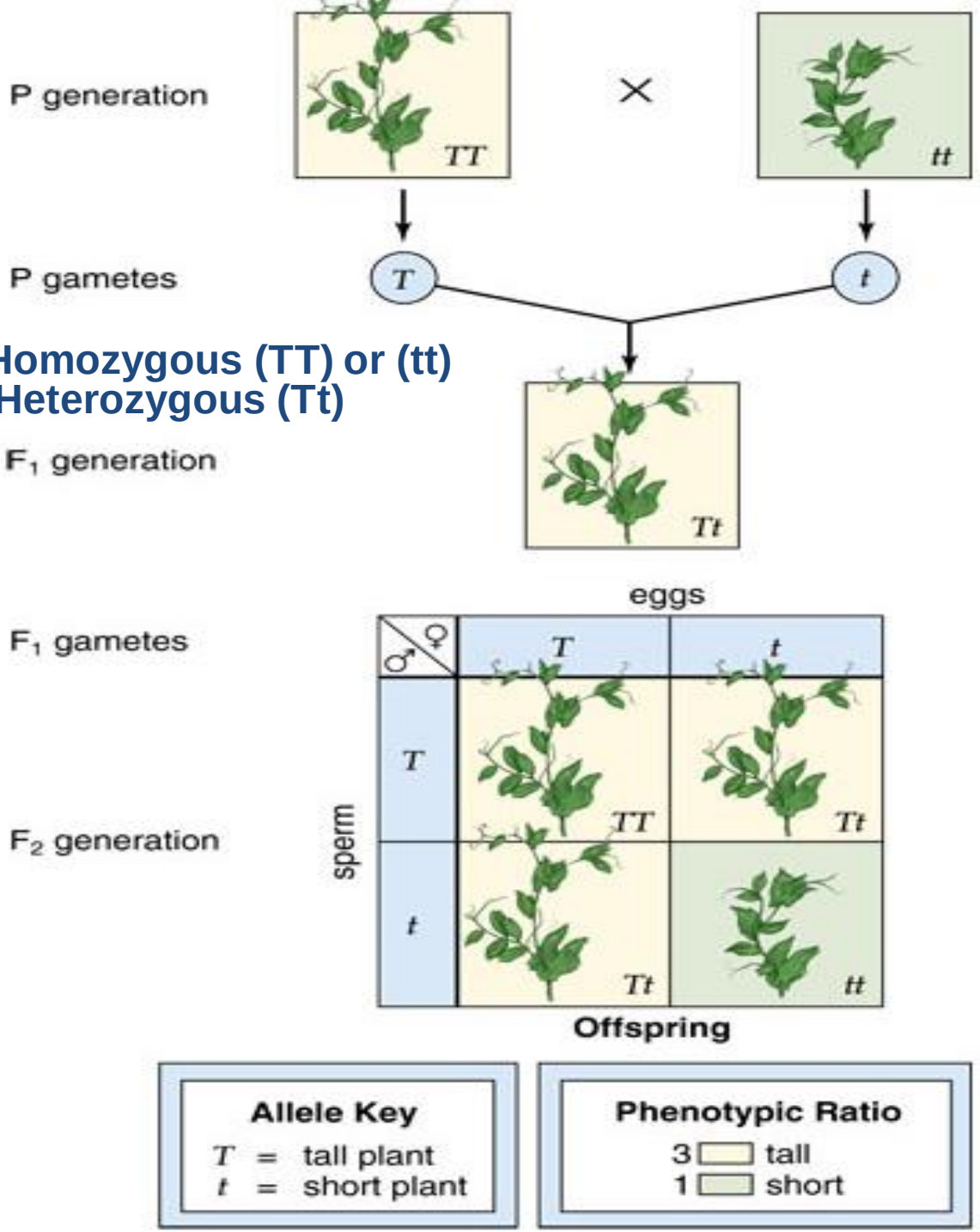
Punnet Square:

1: XX- Female
XY- Male

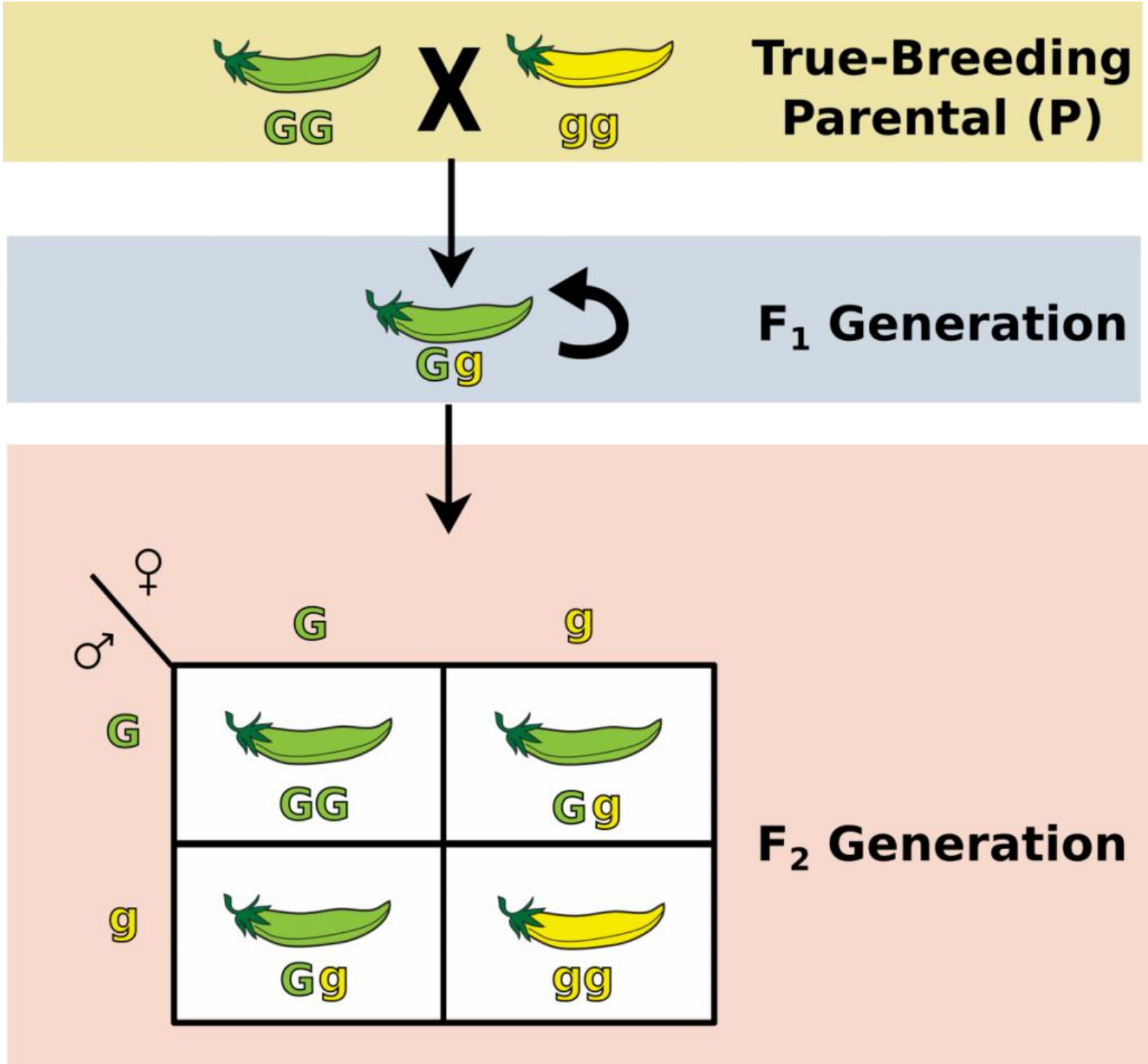
2: Pp X Pp



Mendel's Monohybrid Crosses: An Example



P - Homozygous (TT) or (tt)
F1- Heterozygous (Tt)


GG
X
gg
True-Breeding
Parental (P)
Gg
F1 Generation
♀
♂
G
g
G
g
GG
Gg
Gg
gg
F2 Generation

Garden Pea Traits Studied by Mendel

Trait	Characteristics		F_2 Results*	
	*Dominant	*Recessive	Dominant	Recessive
Stem length	Tall 	Short 	787	277
Pod shape	Inflated 	Constricted 	882	299
Seed shape	Round 	Wrinkled 	5,474	1,850
Seed color	Yellow 	Green 	6,022	2,001
Flower position	Axial 	Terminal 	651	207
Flower color	Purple 	White 	705	224
Pod color	Green 	Yellow 	428	152

*All of these produce approximately a 3:1 ratio. For example, $\frac{787}{277} = \frac{3}{1}$.

b.

Mendel's Monohybrid Crosses: An Example

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant:Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6022:2001	3.01:1
Seed shape	Round 	×	Wrinkled 	5474:1850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

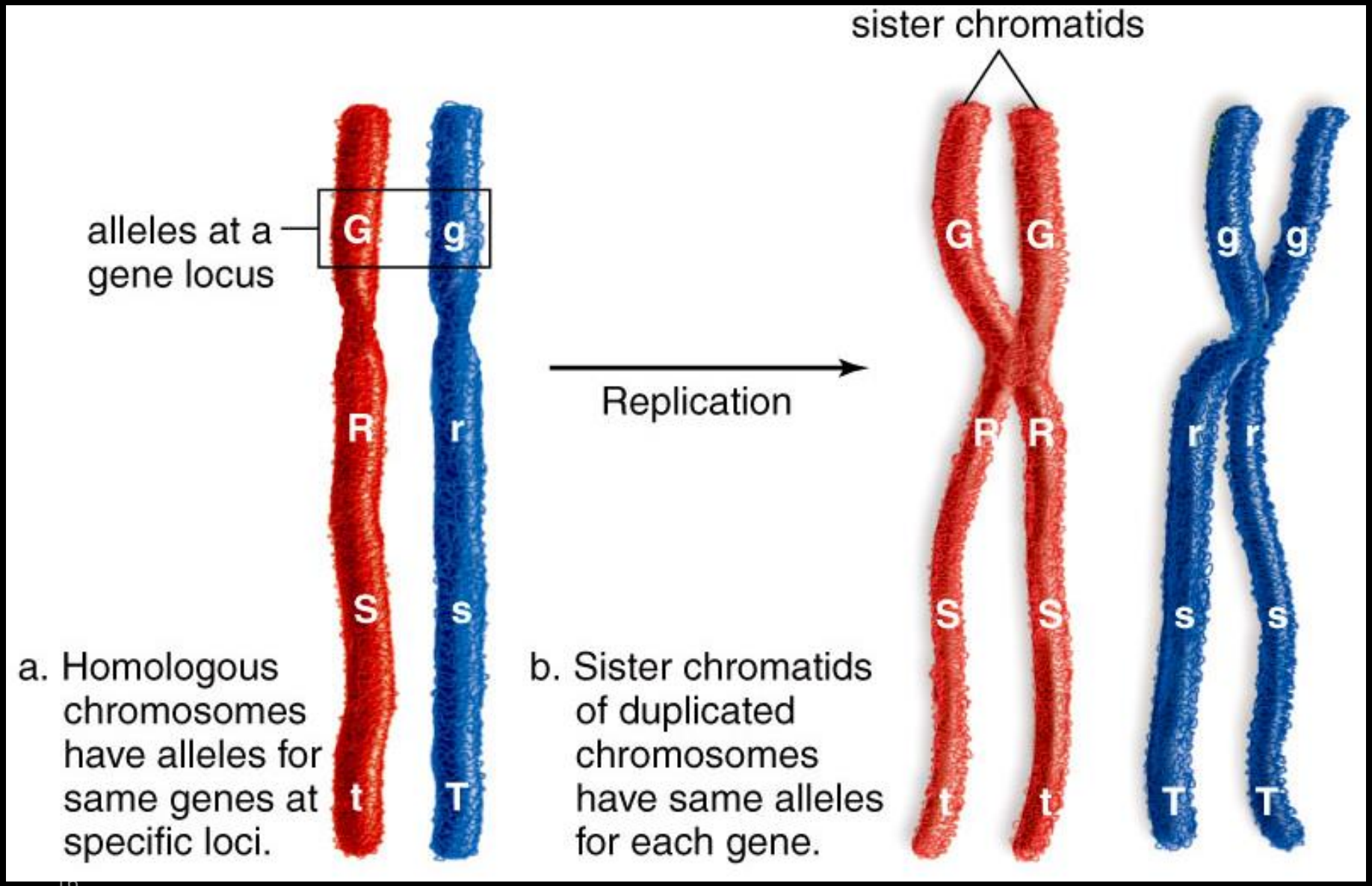
Law of Segregation

- So Mendel guessed that individuals actually carry TWO traits for each character, one from each parent
- But for each character, one trait is **dominant** to the other, so that if you have BOTH, only the dominant one is expressed. The "masked" trait is called **recessive**. In modern language…
- For every **GENE**, an individual has two **ALLELES** (one from each parent)
- If you have two of the SAME alleles, this would be true-breeding. Now called **homozygous (PP)**.
- If you have two DIFFERENT alleles, called **heterozygous (Pp)**, only the dominant one is expressed.

Modern Genetics View

- Each trait in a pea plant is controlled by two alleles (alternate forms of a gene)
- **Dominant allele** (**capital letter – P)** masks the expression of the **recessive allele (lower-case - p)**
- Alleles occur on a homologous pair of chromosomes at a particular **gene locus**
  - **Homozygous = identical alleles (PP)**
  - **Heterozygous = different alleles (Pp)**

Homologous Chromosomes

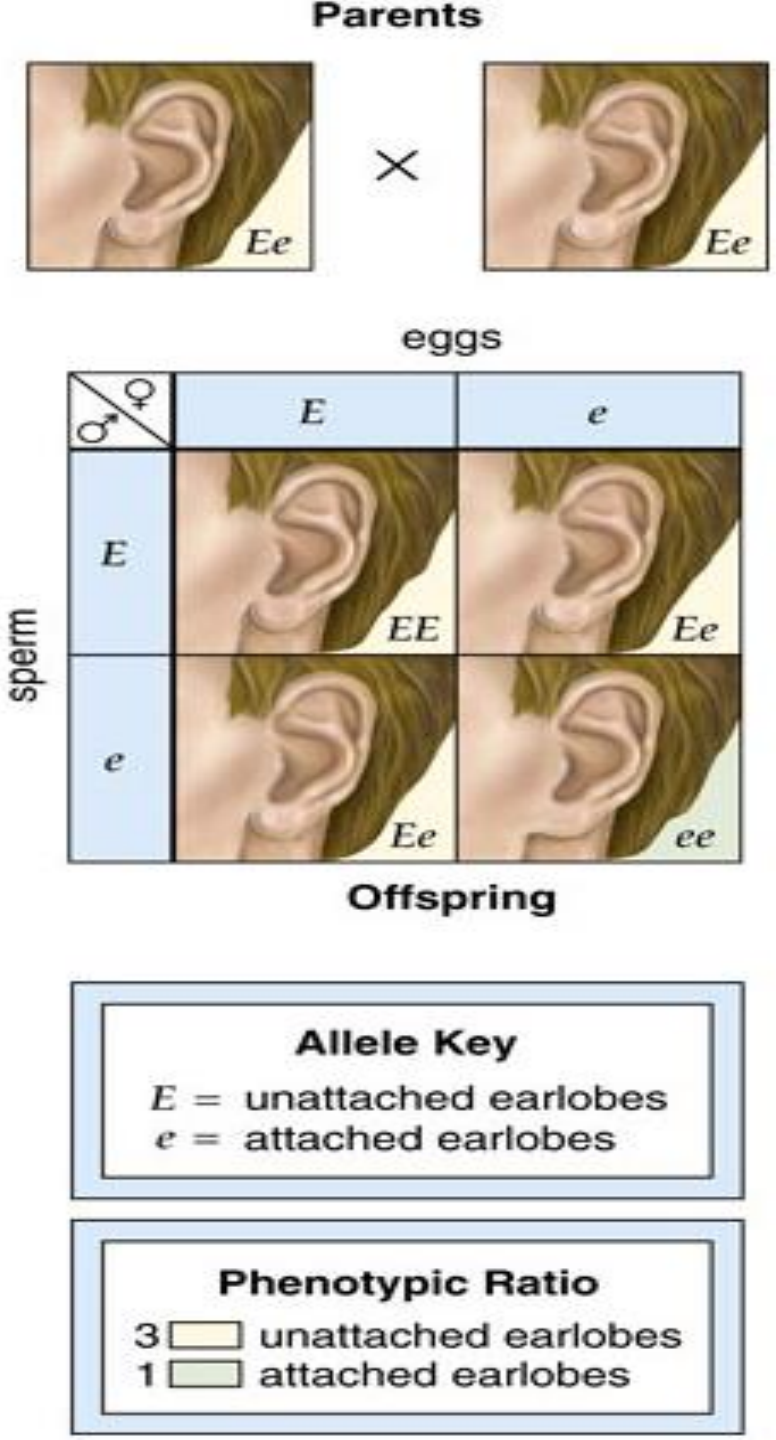


Genotype Versus Phenotype

- **Genotype** (what are the genes)
  - Refers to the two **alleles** an individual has for a specific trait
  - If identical, genotype is homozygous (PP) or (pp)
  - If different, genotype is heterozygous (Pp)
- **Phenotype** (what you see)
  - Refers to the **physical appearance** of the individual
  - (PP) or (Pp) Purple VS (pp) - white

Punnett Square

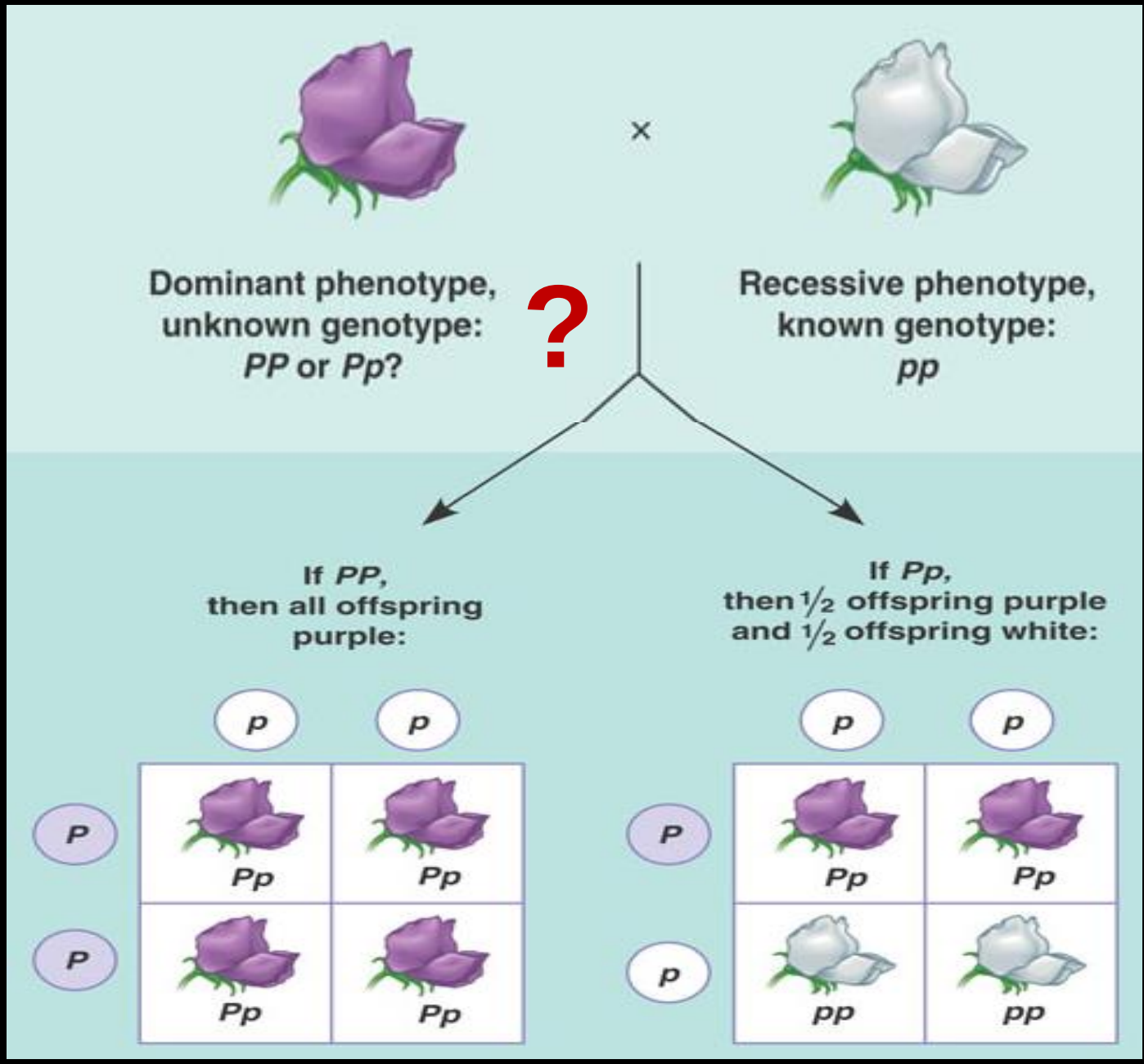
- Table listing all possible genotypes resulting from a cross
  - All possible sperm genotypes are lined up on one side
  - All possible egg genotypes are lined up on the other side
  - Every possible zygote genotypes are placed within the squares



Monohybrid Testcross [Blue vs Green]

- Individuals with **recessive** phenotype always have the **homozygous** recessive genotype (bb)
- However, Individuals with dominant phenotype have indeterminate genotype
  - May be homozygous dominant **(GG)** , or
  - Heterozygous **(Gg)**
- **Test cross determines genotype** of individual having **dominant phenotype (GG) or (Gg)**
  - You simply cross the individual to a pure-breeding (homozygous) recessive (bb)
  - If all offspring are the dominant phenotype (GG) or (Gg), then the unknown must have been homozygous
  - If they are ½ dominant (Gg), ½ recessive (bb), it was heterozygous.

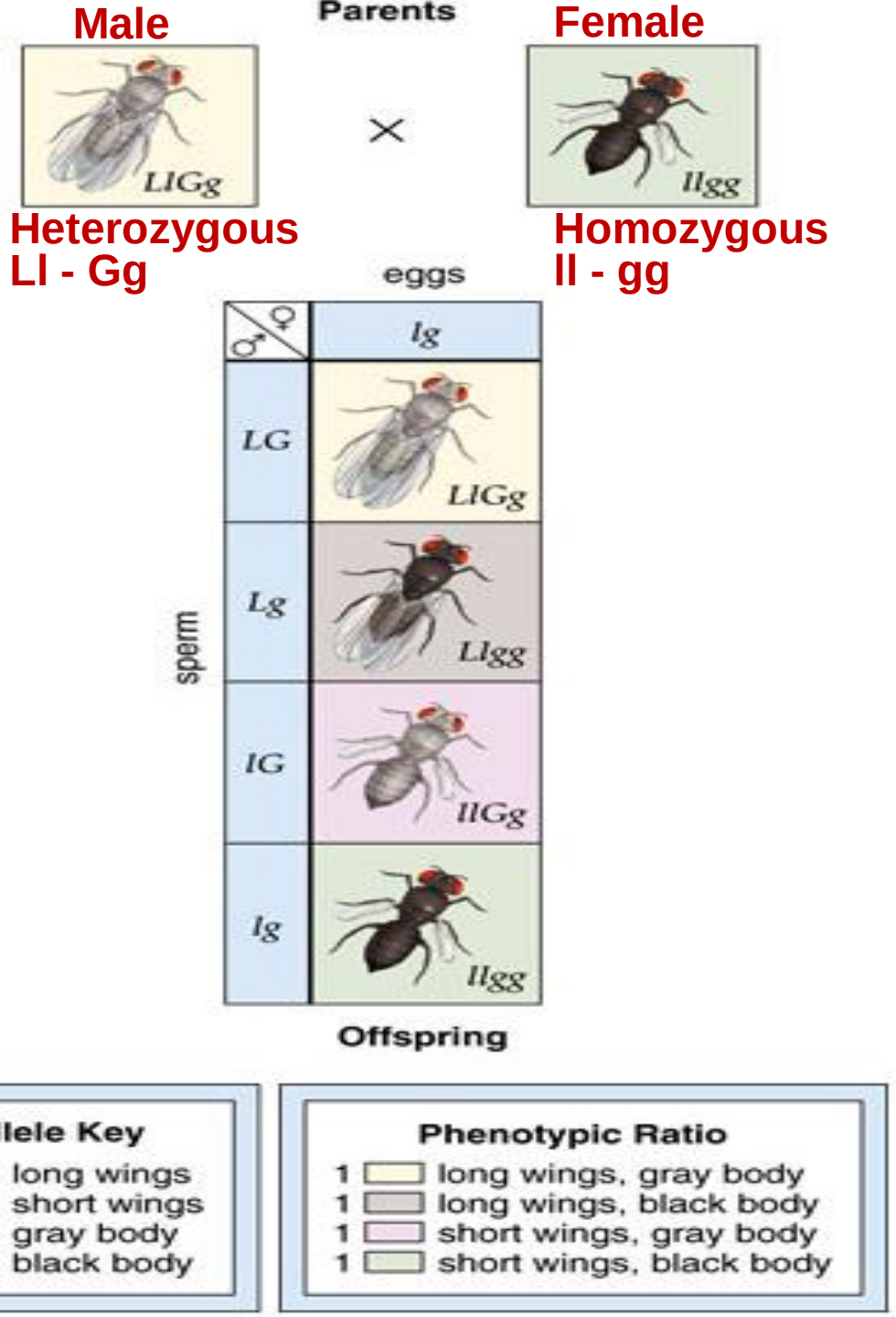
One-Trait Test Cross

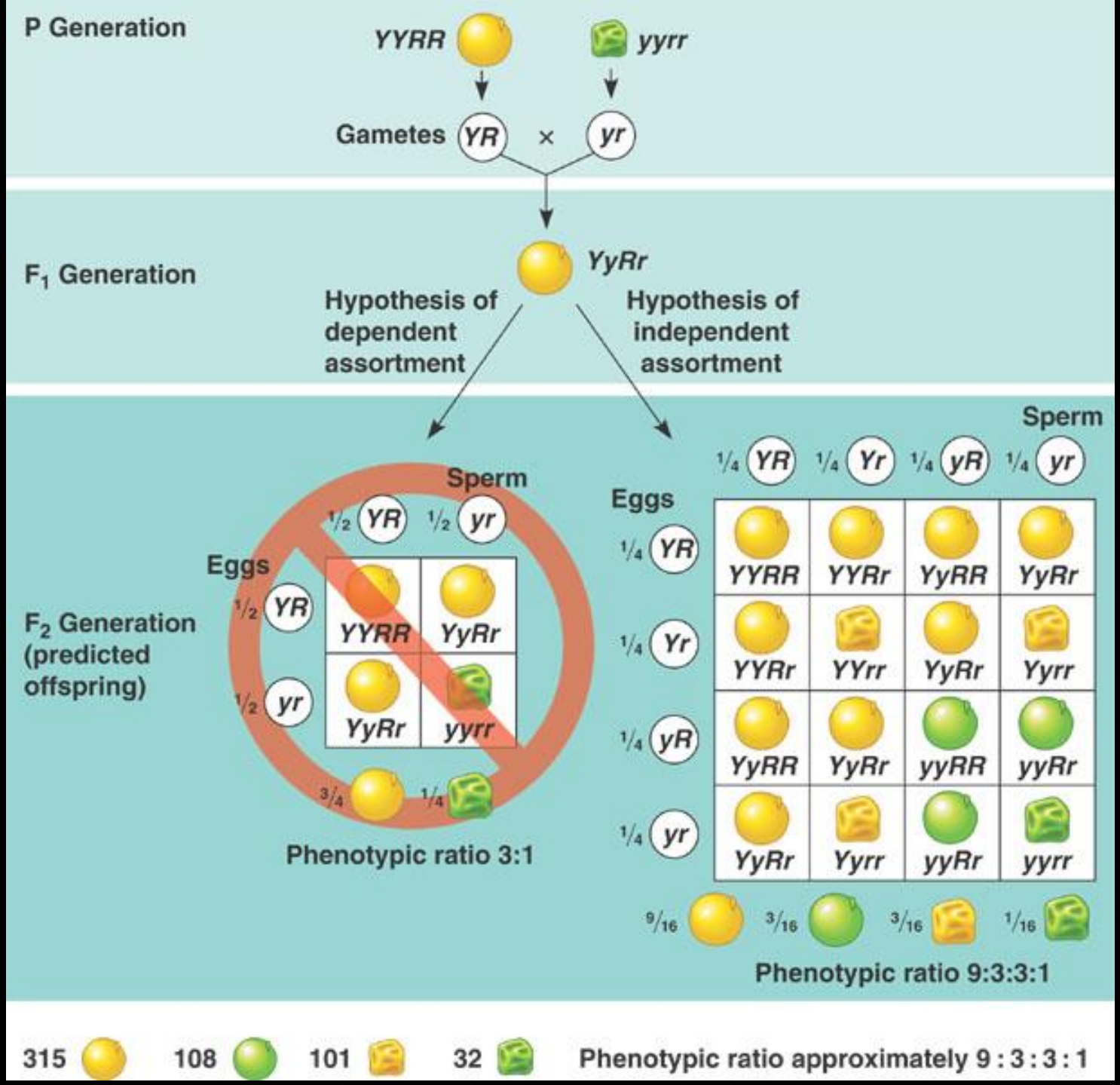


Two-Trait Inheritance

- Dihybrid cross uses true-breeding plants differing in **two traits (TtGg)**
  - Observed phenotypes among F_2 plants
  - Formulated **Law of Independent Assortment**
    - During gamete formation, each pair of alleles will segregate INDEPENDENTLY of all other alleles
    - All possible combinations of alleles can occur in the gametes

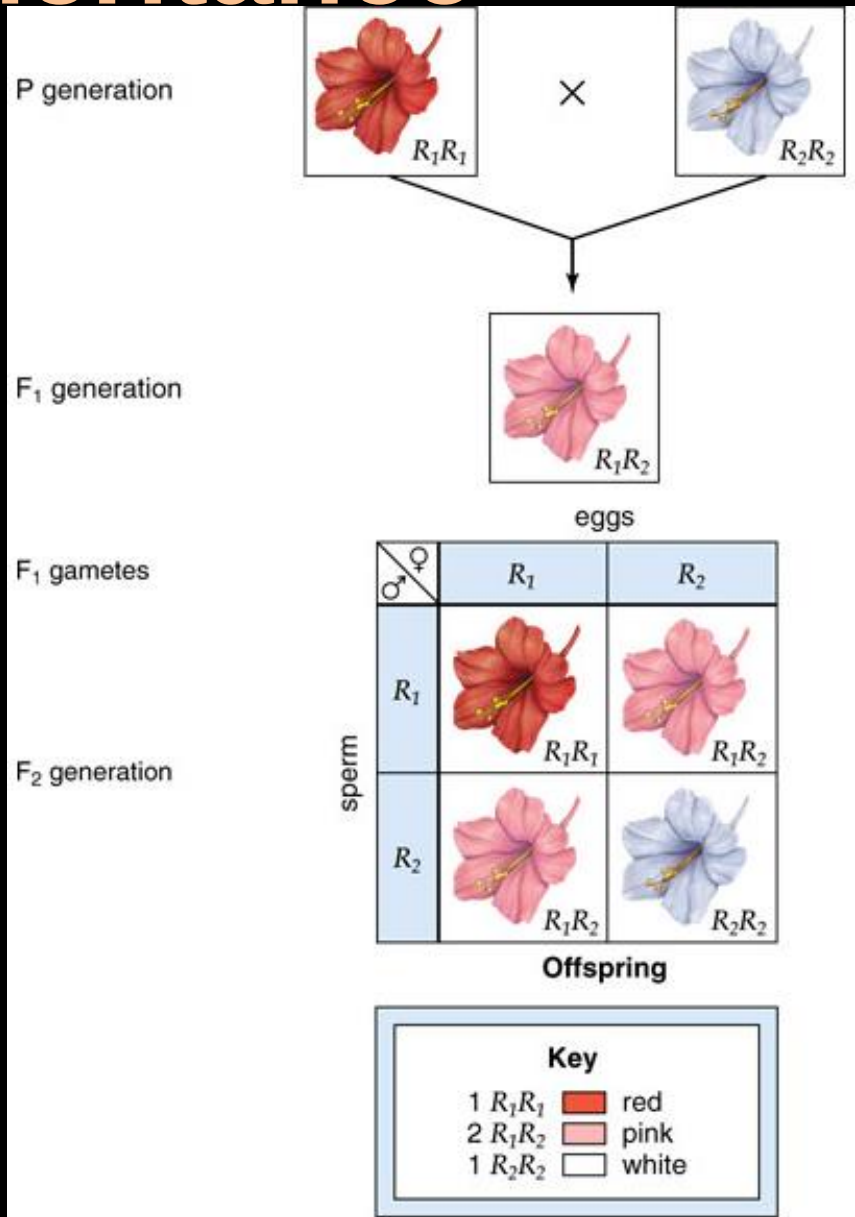
Two-Trait Test Cross




P Generation
YYRR
yyrr
Gametes
YR
×
yr
F1 Generation
YyRr
Hypothesis of dependent assortment
Hypothesis of independent assortment
F2 Generation (predicted offspring)
Sperm
1/2 YR
1/2 yr
Eggs
1/2 YR
1/2 yr
YYRR
YyRr
YyRr
yyrr
3/4
1/4
Phenotypic ratio 3:1
Sperm
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
Eggs
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
YYRR
YYRr
YyRR
YyRr
YYRr
YYrr
YyRr
Yyrr
YyRR
YyRr
yyRR
yyRr
YyRr
Yyrr
yyRr
yyrr
9/16
3/16
3/16
1/16
Phenotypic ratio 9:3:3:1
315
108
101
32
Phenotypic ratio approximately 9 : 3 : 3 : 1

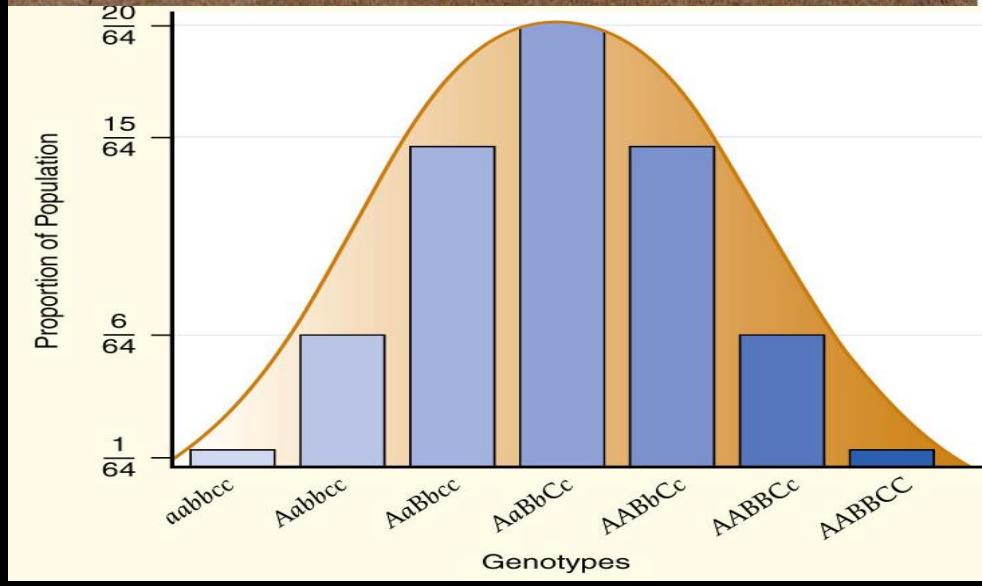
Incomplete Dominance -- Blending Inheritance

- Heterozygote has phenotype intermediate between that of either homozygote
  - Homozygous red has red phenotype
  - Homozygous white has white phenotype
  - Heterozygote has pink (intermediate) phenotype
- Phenotype reveals genotype without test cross



Polygenic Inheritance

- Occurs when a trait is governed by two or more genes having different alleles
- Each dominant allele has a quantitative effect on the phenotype
- These effects are additive
- Result in continuous variation of phenotypes



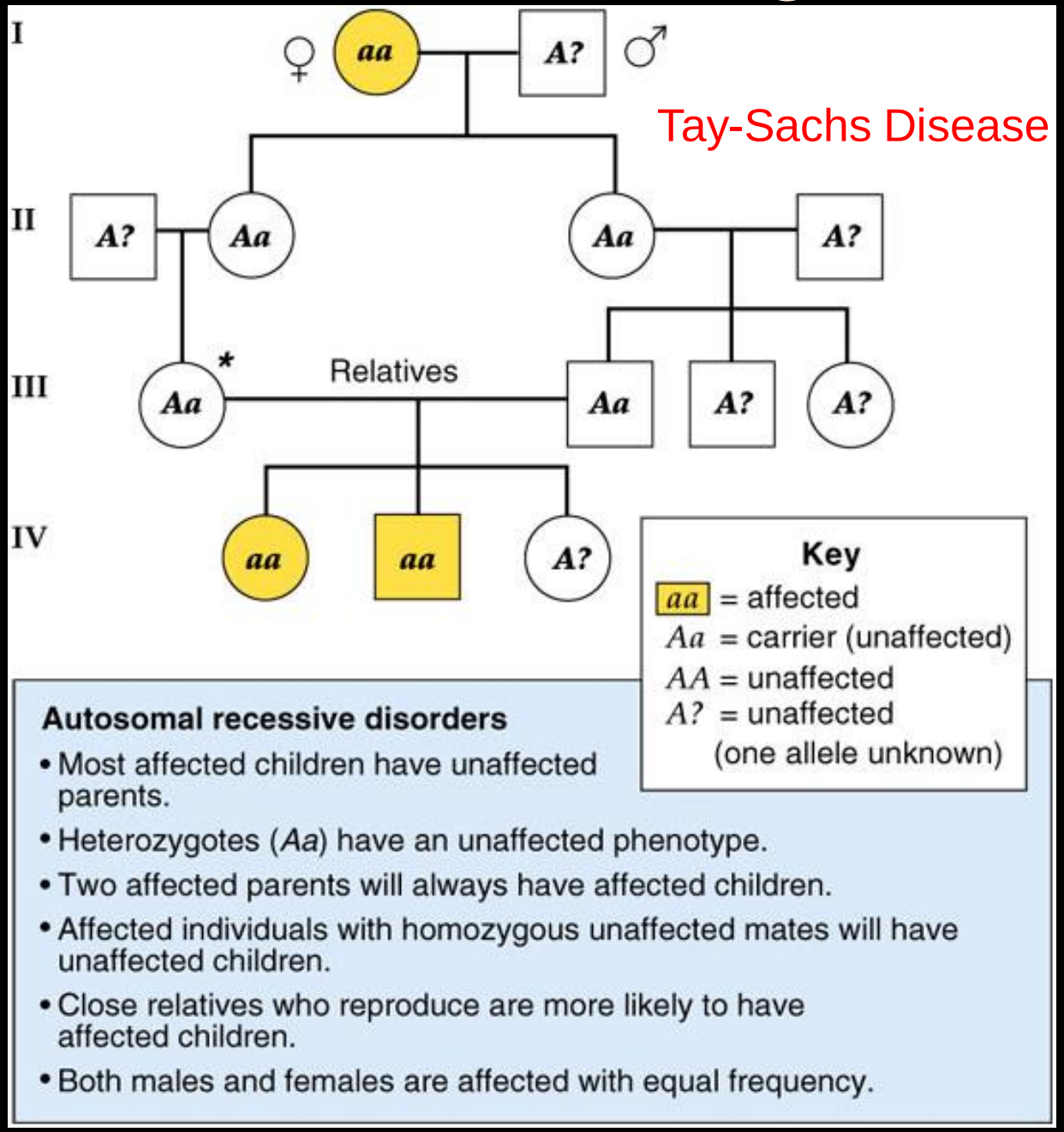
Nature vs. Nurture

- Environment can influence the expression of genes, so phenotype is not always strictly dependent on genotype
  - E.g., identical twins have the same genes, but are NOT perfectly identical Any Ideas How?
- Genes often establish a **norm of reaction**, which is a range of possible phenotypes for a given genotype.
- So “nature” and “nurture” cooperate to establish phenotypes

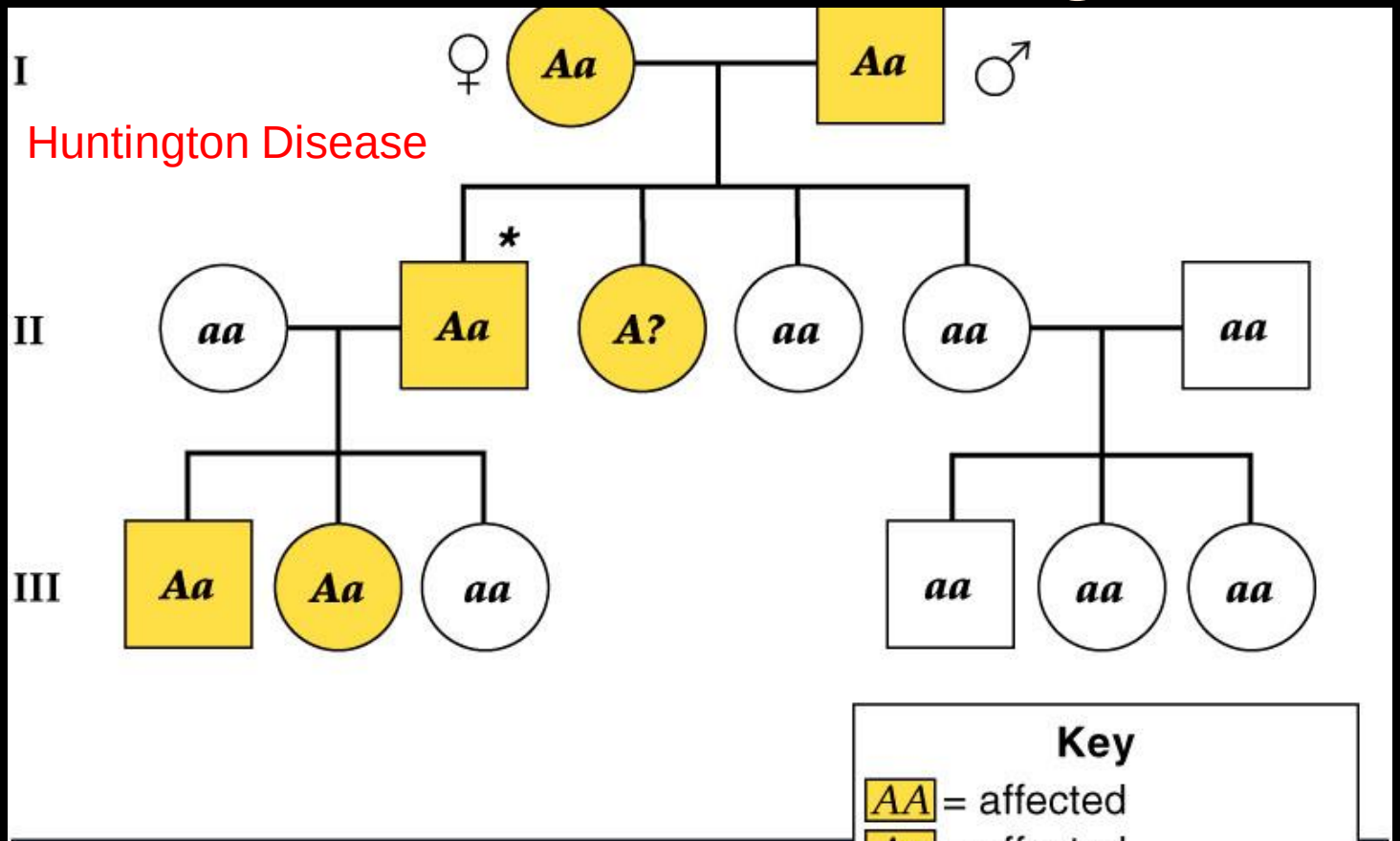
Human Genetic Disorders

- **Autosome** - Any chromosome other than a sex chromosome
- Genetic disorders caused by genes on autosomes are called **autosomal disorders**
  - Some genetic disorders are autosomal dominant
    - An individual with AA has the disorder
    - An individual with Aa has the disorder
    - An individual with aa does NOT have disorder
  - Other genetic disorders are autosomal recessive
    - An individual with AA does NOT have disorder
    - An individual with Aa does NOT have disorder, but is a carrier
    - An individual with aa DOES have the disorder

Autosomal Recessive Pedigree Chart



Autosomal Dominant Pedigree Chart

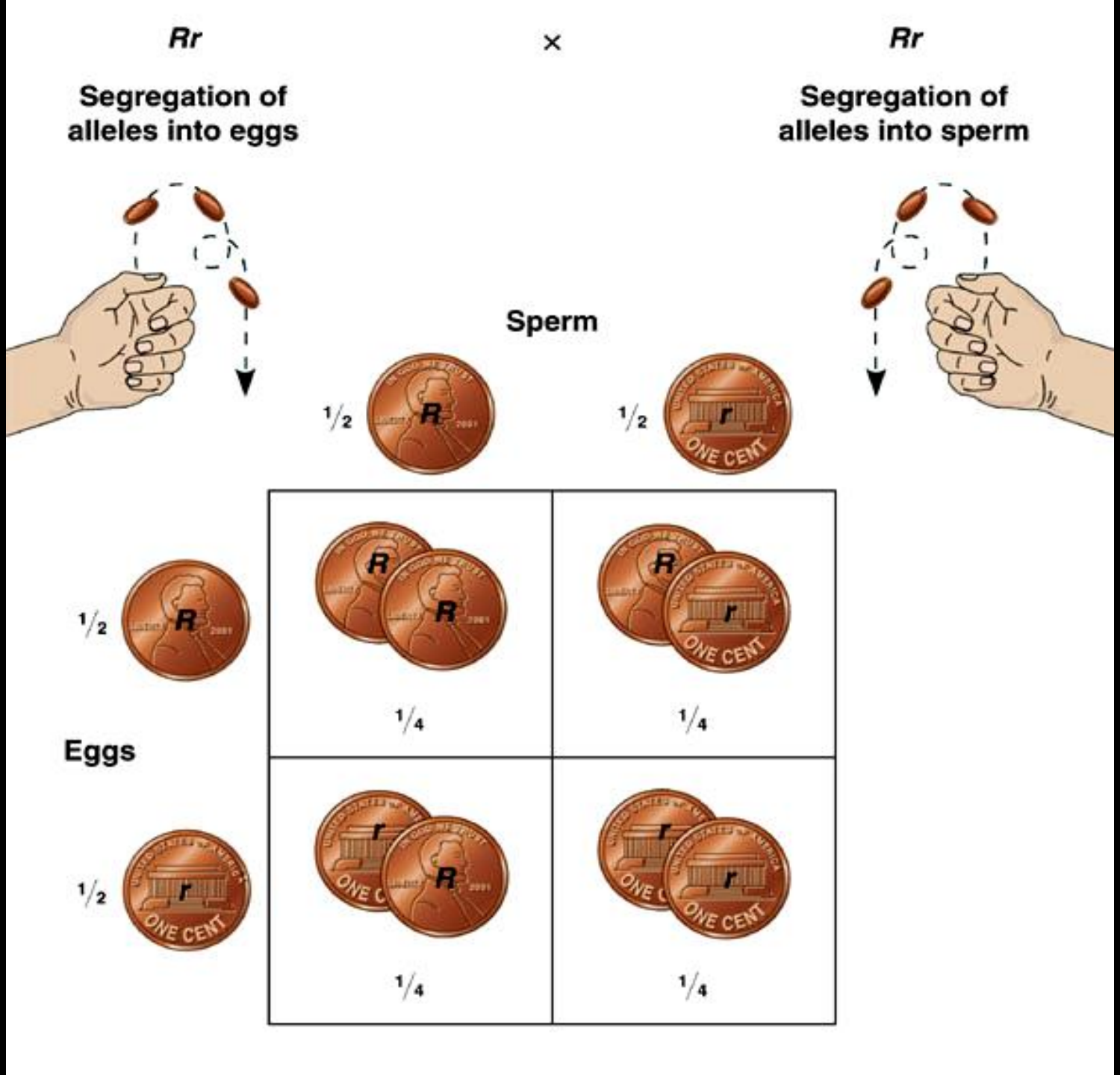


Autosomal dominant disorders

- Affected children will usually have an affected parent.
- Heterozygotes (*Aa*) are affected.
- Two affected parents can produce an unaffected unaffected child.
- Two unaffected parents will not have affected children.
- Both males and females are affected with equal frequency.

Probability

- Assortment of alleles into gametes is random
- Fertilization (which gametes) is random
- Probability = number of ways a certain event could occur divided by the total number of possible outcomes
  - If probability = 1, the event is CERTAIN to happen
  - If probability = 0, the event CANNOT happen
  - Everything in between is a fraction (or percent)
  - Example = flipping a coin: prob. of heads = ½, prob. of tails = ½
  - The probability of all outcomes adds up to 1


Rr
×
Rr
Segregation of alleles into eggs
Segregation of alleles into sperm
Sperm
1/2
1/2
Eggs
1/2
1/2
1/4
1/4
1/4
1/4

Rules for Calculating Probability

So you don't have to use a Punnett square every time…

Rule of addition – If an outcome can occur two different ways, the two probabilities are ADDED to get the final probability

Usually involves an "OR" statement

Rule of Multiplication – The overall probability of two independent events occurring together equals the PRODUCT (multiplication) of their individual probabilities

Usually involves an "AND" statement

Terminology

- Pleiotropy
  - A gene that affects more than one characteristic of an individual (multiple phenotypic effects)
  - Sickle-cell (incomplete dominance)
- Codominance
  - More than one allele is fully expressed
  - ABO blood type (multiple allelic traits)
- Epistasis
  - A gene at one locus interferes with the expression of a gene at a different locus
  - Human skin color (polygenic inheritance)

Autosomal Recessive Disorders

- Tay-Sachs Disease
  - Progressive deterioration of psychomotor functions
- Cystic Fibrosis
  - Mucus in bronchial tubes and pancreatic ducts is particularly thick and viscous
- Phenylketonuria (PKU)
  - Lack enzyme for normal metabolism of phenylalanine

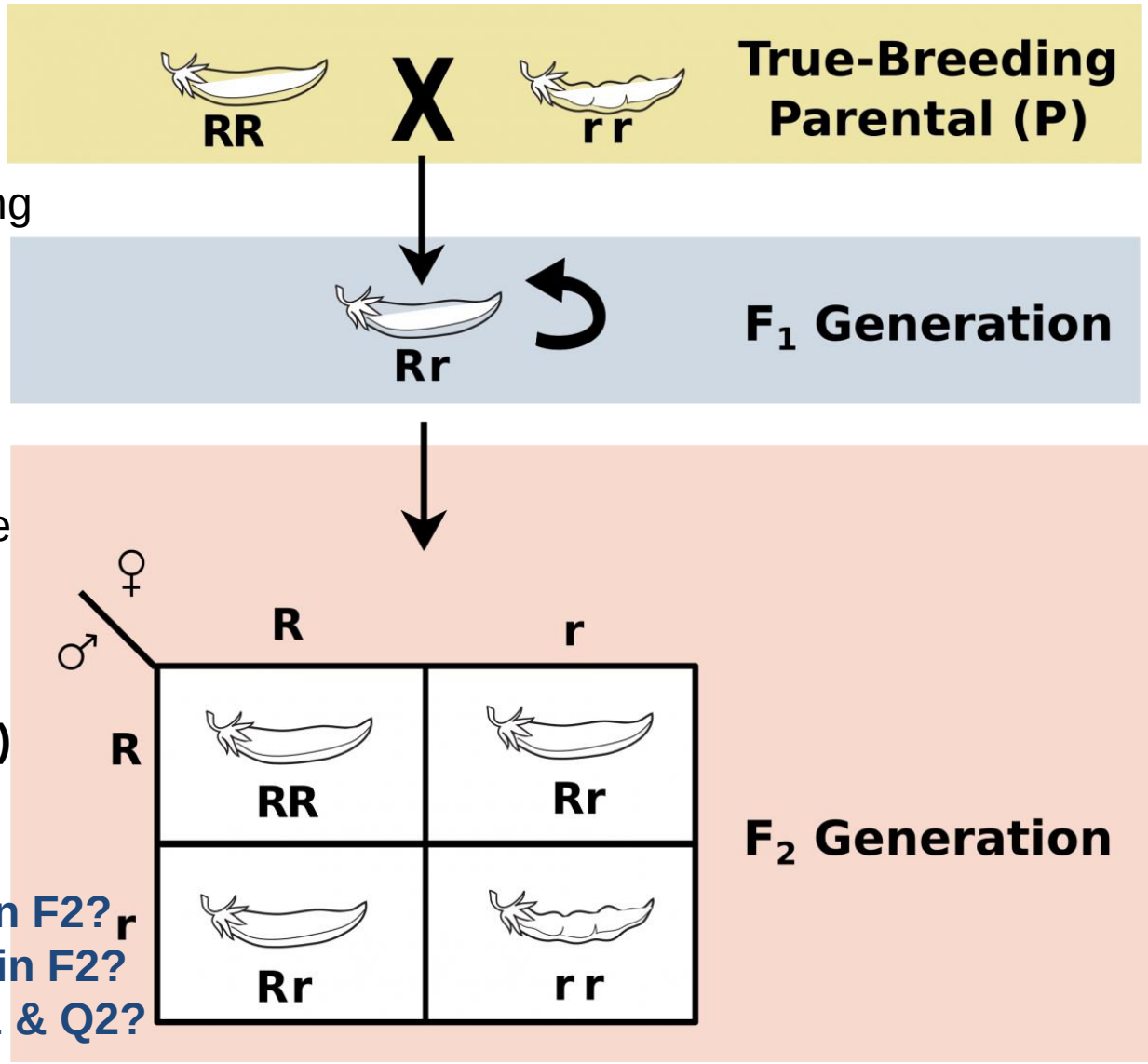
Autosomal Dominant Disorders

- Neurofibromatosis
  - Tan or dark spots develop on skin and darken
  - Small, benign tumors may arise from fibrous nerve coverings
- Huntington Disease
  - Neurological disorder
  - Progressive degeneration of brain cells
    - Severe muscle spasms
    - Personality disorders

2. Mendelian Genetics: Breeding

https://openlab.citytech.cuny.edu/bio1-oer/genetics/6/

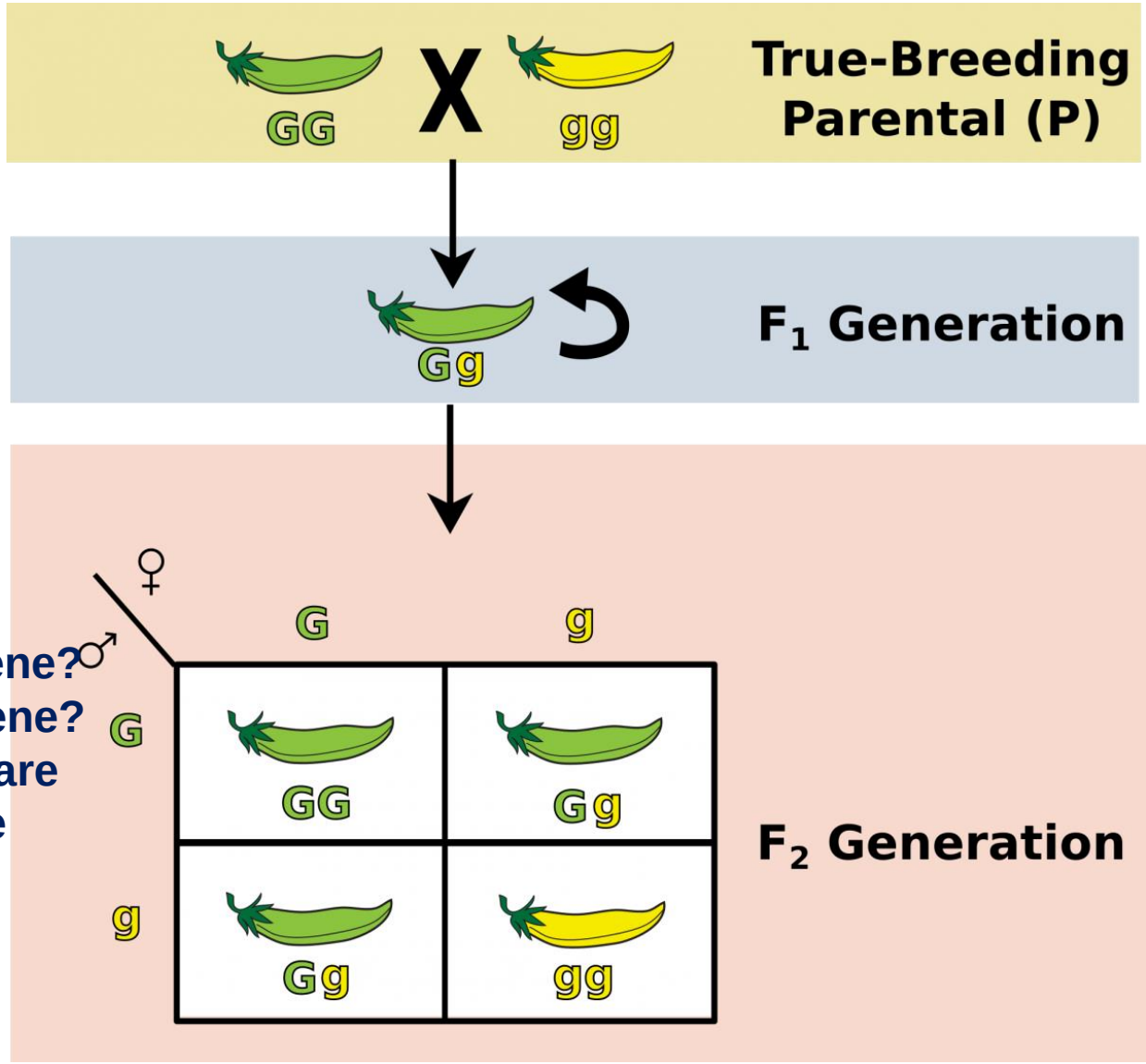
1. The **Punnett square** is a diagram that is used to predict an outcome of a particular cross or breeding experiment.
2. It is named after Reginald C. **Punnett**, who devised the approach.
3. The diagram is used by biologists to determine the probability of an offspring having a particular genotype.
4. **Homozygous (RR) or (rr)**
5. **Heterozygous (Rr)**



Q1. How many Homozygous in F2?
Q2. How many Heterozygous in F2?
Q3. What are they for each Q1 & Q2?

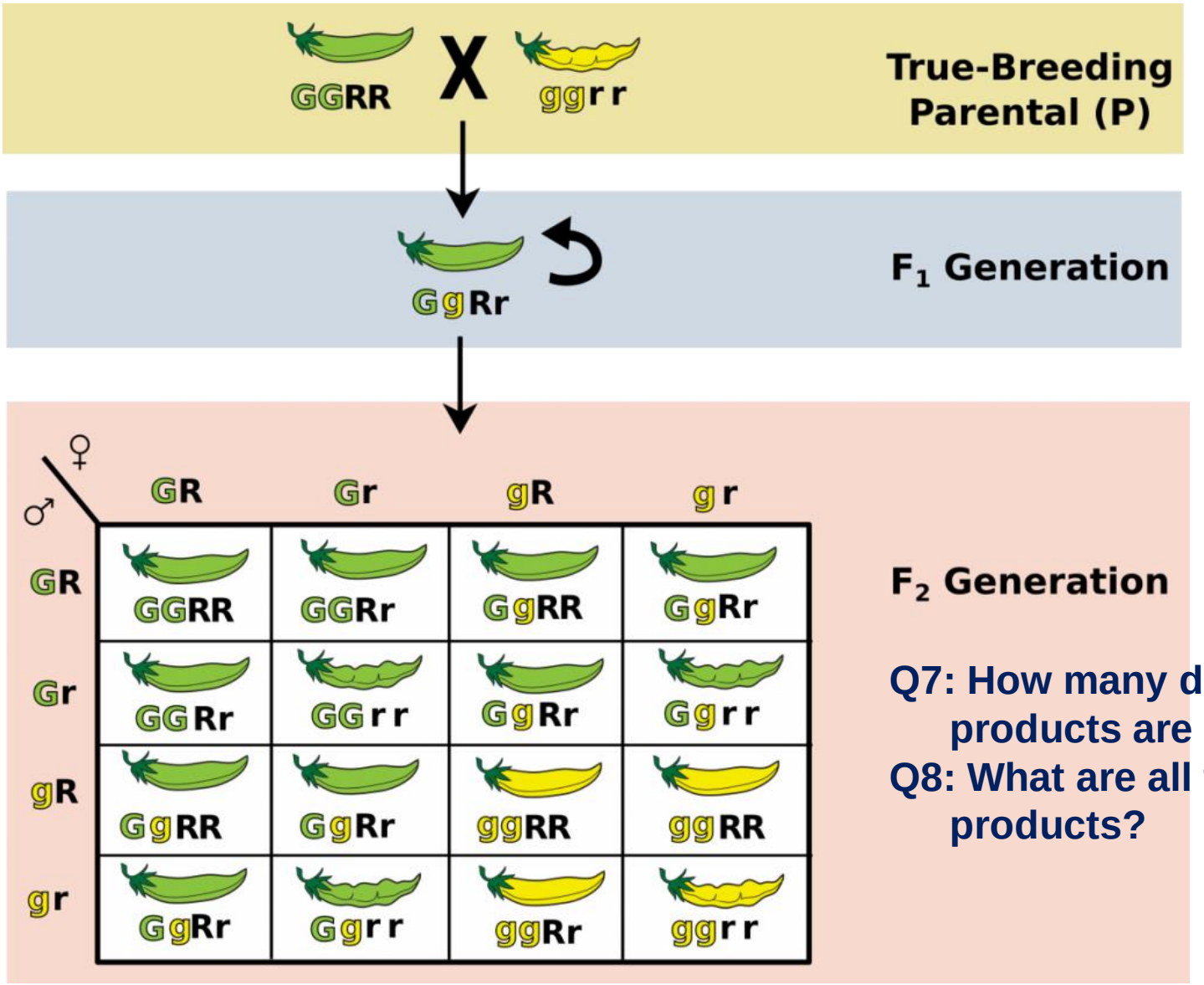
Punnett Square: OneTrait Cross

Dominant (G) versus
Recessive (g) genes



Q4: What is a dominant gene?
Q5: What is a recessive gene?
Q6: In this example, what are the dominant/recessive genes?

Punnett Square: The Two Trait Cross (Dihybrid Cross)



Q7: How many different products are there?

Q8: What are all the different products?

Probability Practice:

Consider this cross: Rr x Rr

- Think about the gametes that each could produce and assign a fraction to each
- Fertilization occurs. What percentage would be…
- RR ? Rr ? rr ?
- What fraction would be the dominant phenotype?

Consider this cross: Tt x tt

- What fraction would be tt?
- What fraction would be the recessive phenotype?

Consider this cross: GgEe x GgEE

- What fraction would be ggEE?
- What fraction would be GgEe?
- What fraction would be ggee?