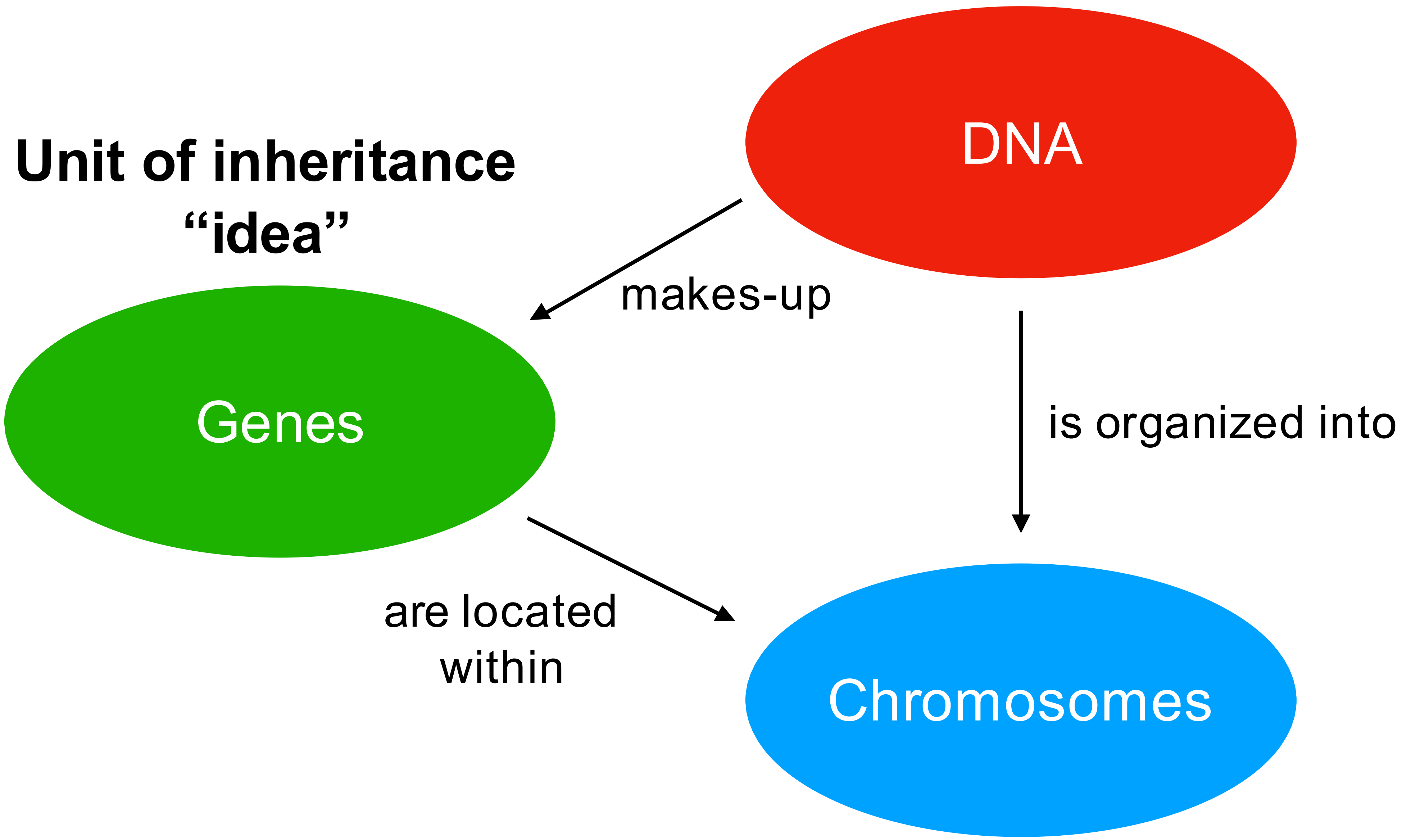
3.4: Mendelian Genetics I

Chapter 12

Learning Objectives

- Explain the principles of segregation
- Understand what alleles are and how they relate to genes and traits
- Explain how genotype and phenotype are different and how they are connected
- Perform monohybrid crosses and use Punnett squares to predict outcomes
- Understand complete dominance and codominance

Genes form the basis of inheritance



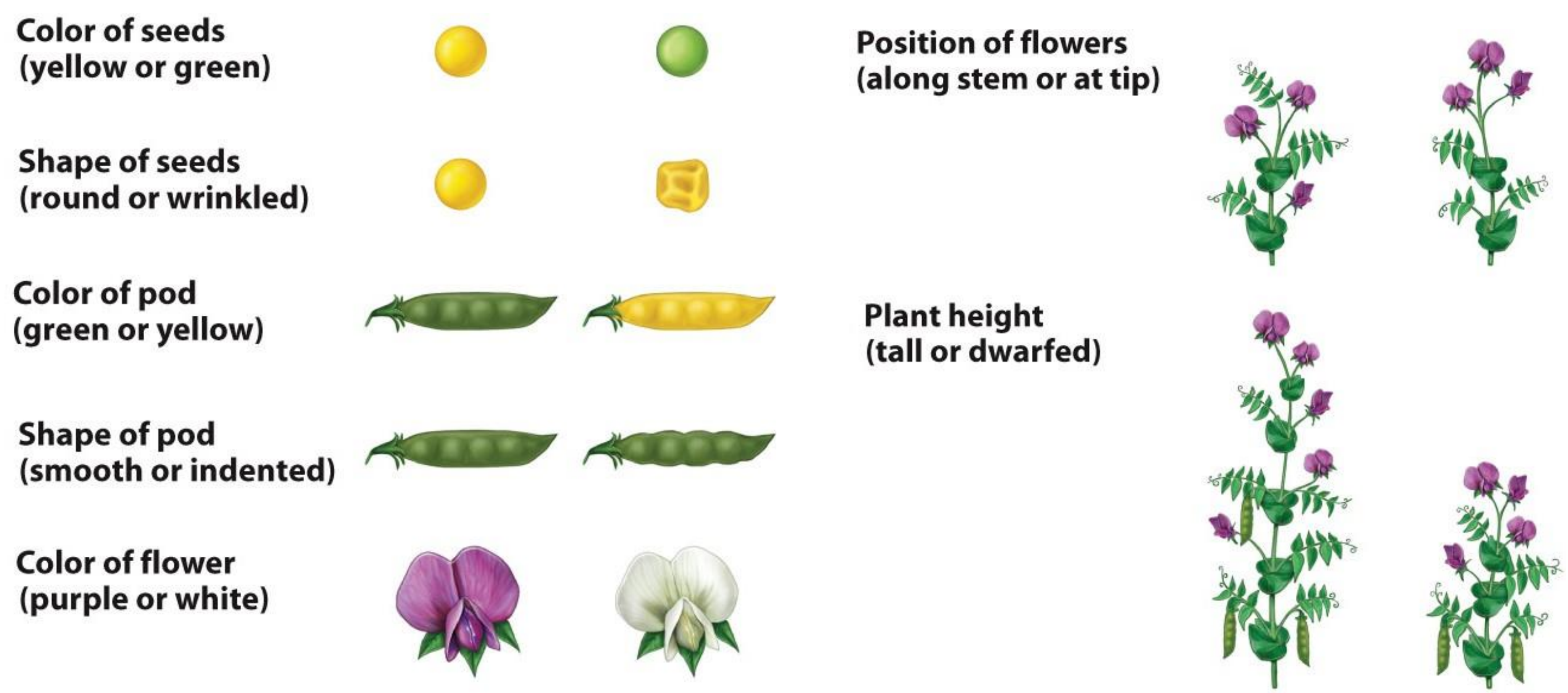
before knowing anything about genes, chromosomes, or DNA principles of inheritance were discovered

~ 170 years ago … by a monk

Gregor Mendel, the father of genetics

1822 Born → **1843** Becomes monk → **1856-1863** Pea experiments → **1866** Publishes work → **1900** Work rediscovered!

The garden pea (*Pisum sativum*)



Morris et al, Biology: How Life Works, 3rd edition (Freeman)

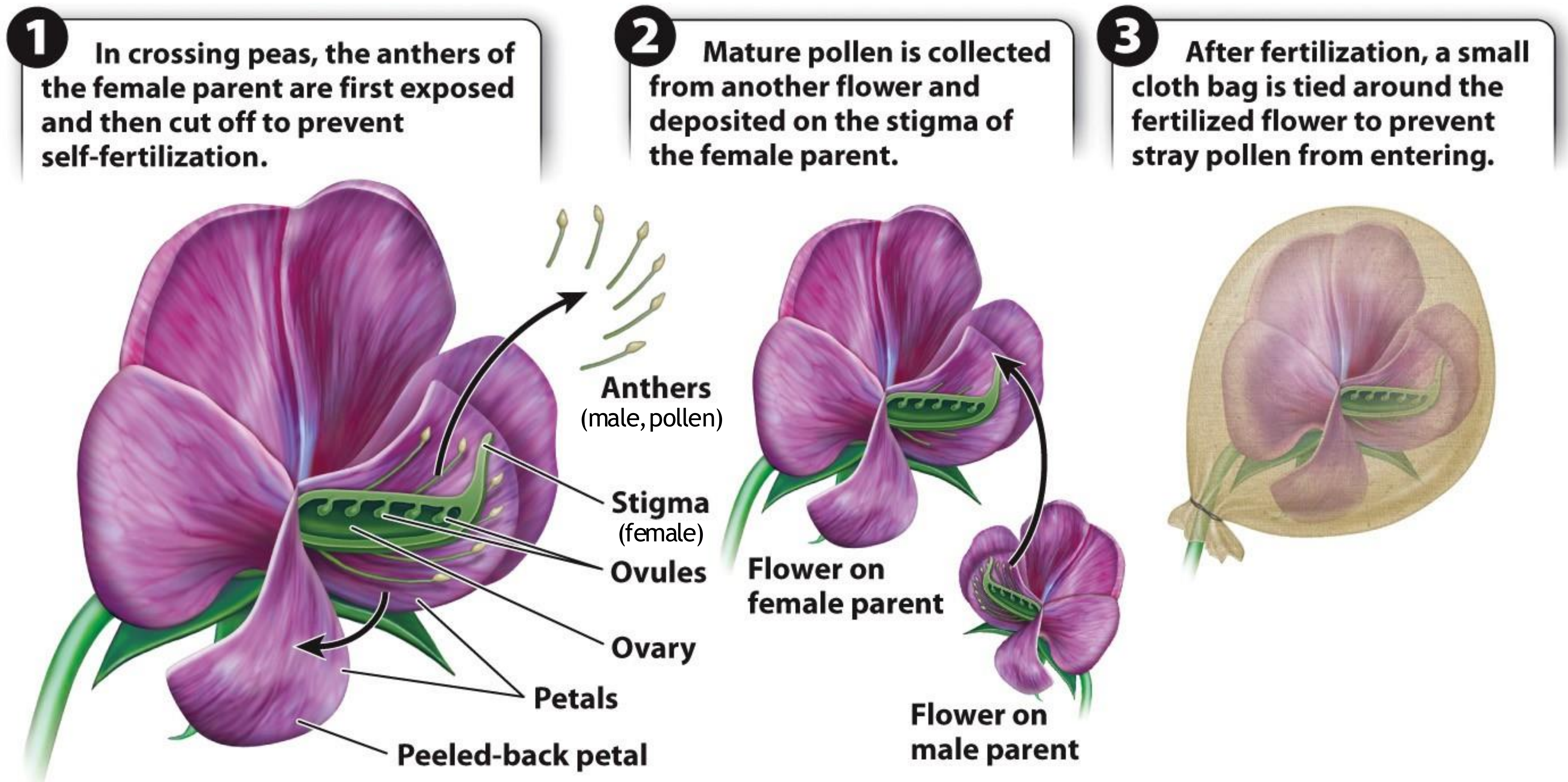
- Fun fact: Mendel failed his teaching certification exam TWICE—but became the father of genetics!
- His work was ignored for 34 years. Why? He was ahead of his time—no one knew about DNA or chromosomes yet!"

Mendel found several varieties that **bred true** for a specific version of a **trait**

- **trait**: characteristic of an organism
- **true breeding**: crosses between individuals with same trait produce identical offspring

Why peas?

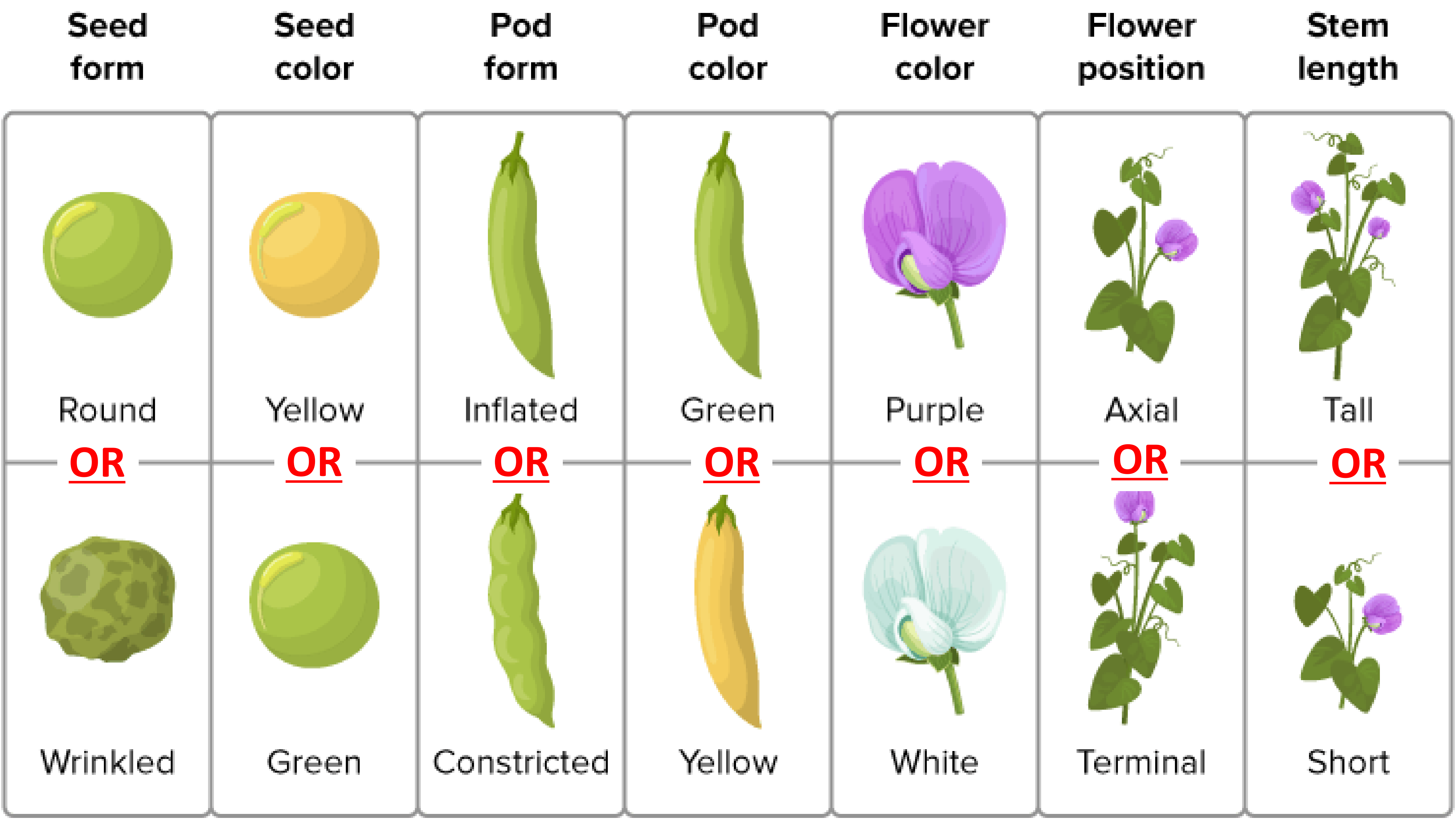
"easy" to cross in a controlled



Feature	Pea Plants	Humans
Generation time	1 growing season	20+ years
Number of offspring	Hundreds per plant	Usually 1-3
Controlled mating	Easy to pollenate	Impossible (and unethical)
Distinct traits	Either/or traits	Most are complex

Even if it were ethical, you'd need to wait 20+ years per generation and could only study a few offspring. Mendel needed thousands of data points. Peas gave him that in just a few years!

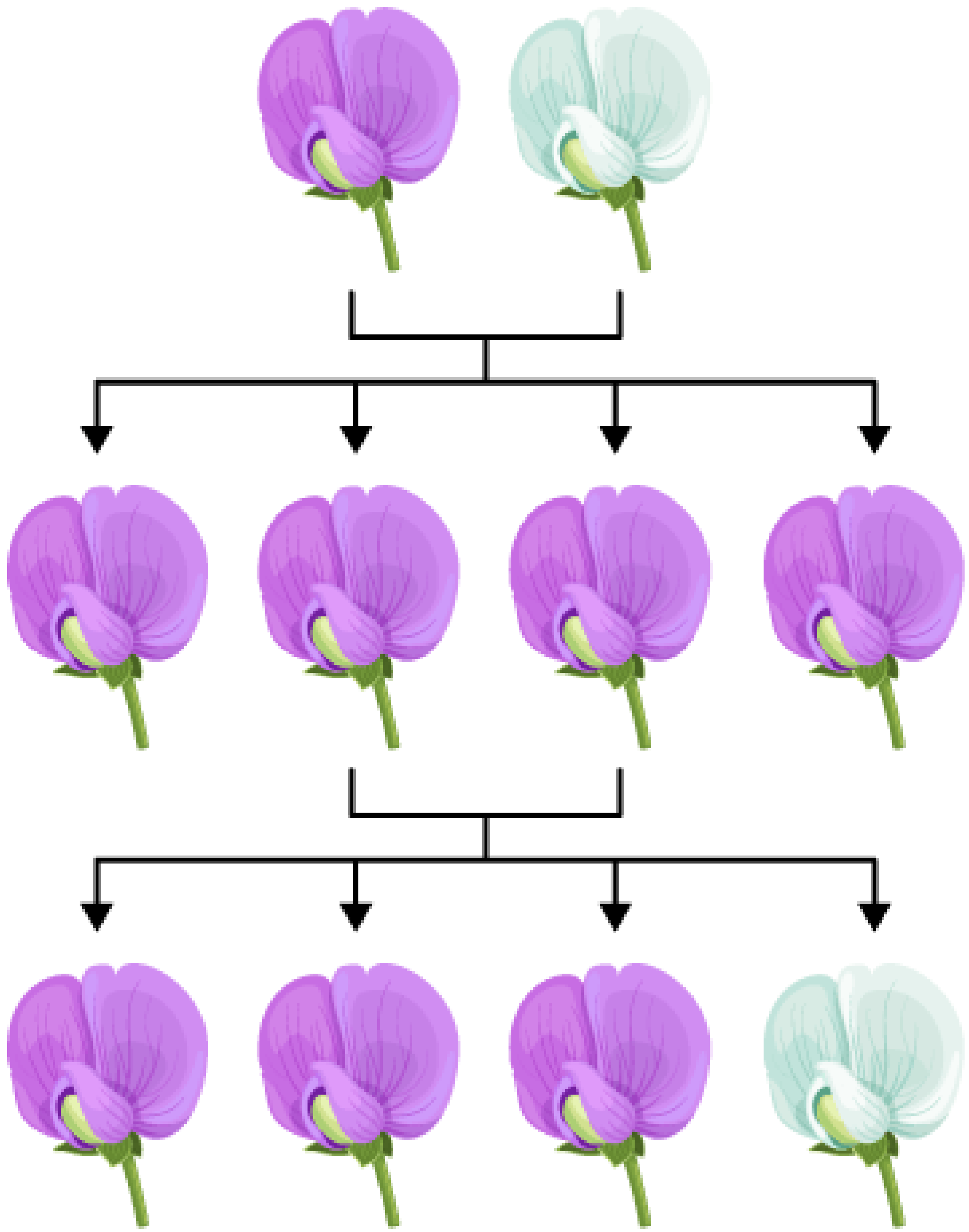
Mendel's Seven Traits: Always Either/Or



Why these traits? They're **discrete** (either/or, no in-between) and controlled by single genes. This made patterns easy to spot!

Simple cross terminology

♂ = male ♀ = female



- **Cross:** cross-pollination (mating)
- **P generation :** parental
- **F_1 (first filial) generation:** offspring of P
- **F_2 (second filial) generation:** offspring of F_1

(from Latin, filia = daughter; filius = son)

Monohybrid crosses (a cross that follows the inheritance of ONE trait)

True breeding: Plants that always produce offspring identical to themselves
- Example: Purple × Purple = Always purple (for many generations)
- Why important: Ensures you know exactly what you're starting with

Mendel's Experimental Approach

STEP 1: Start with true-breeding plants

plants with yellow seeds always made offspring with yellow seeds if self-crossed

plants with green seeds always made offspring with green seeds if self-crossed

ICA Q1: *A gardener has a plant with red flowers. When she lets it self-pollinate, she gets 87 red-flowered offspring and 0 white-flowered offspring. Is this plant true-breeding for red flowers? Explain your reasoning.*

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YES! A true-breeding plant is homozygous (has two identical alleles). When it self-pollinates, it produces only offspring that look like itself.

This plant produced 87 red offspring and 0 white offspring. If it were NOT true-breeding (heterozygous), we'd expect about 25% white flowers (roughly 22 out of 87). Since ALL offspring are red, the parent must be homozygous (like RR) and therefore true-breeding.

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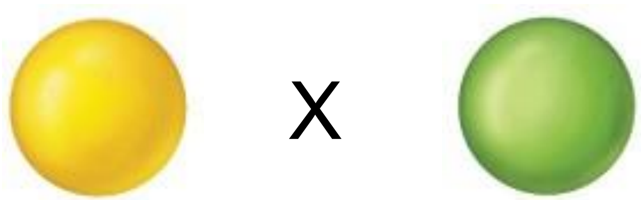
STEP 2: Cross Two Different True-Breeding Line

♀



♂

P generation

↓

?

F1 generation

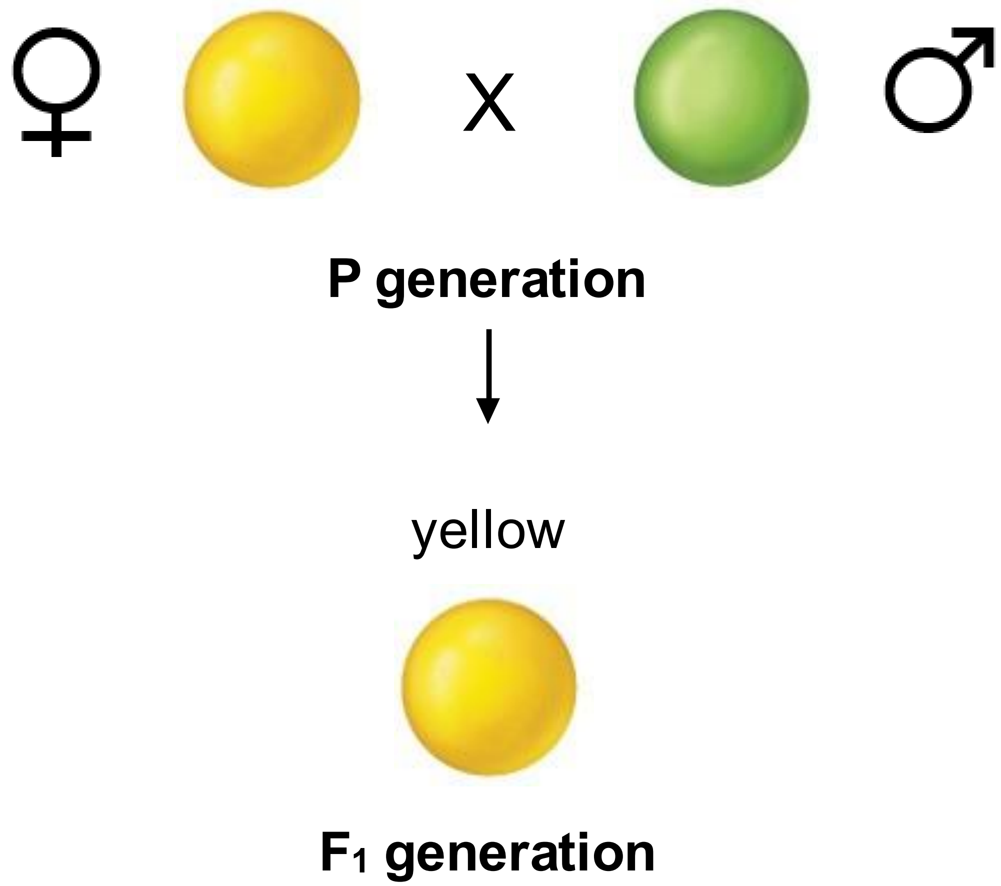
If the parents have different traits, which trait would their offspring inherit?

Before Mendel, three hypotheses existed:

- Ovist - moms phenotype
- Spermist – dads phenotype
- Blending hypothesis – mix of yellow and green

Monohybrid crosses (a cross that follows the inheritance of ONE trait)

What Mendel Actually Found



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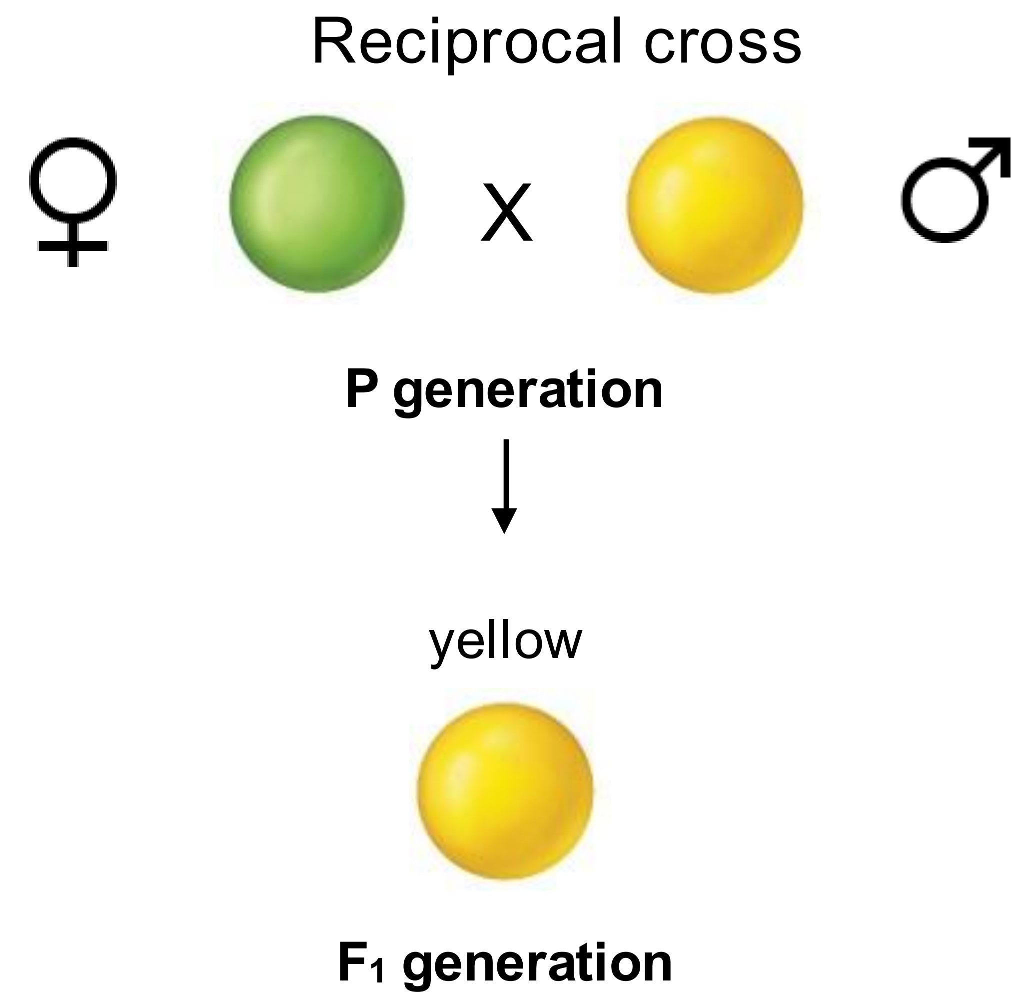
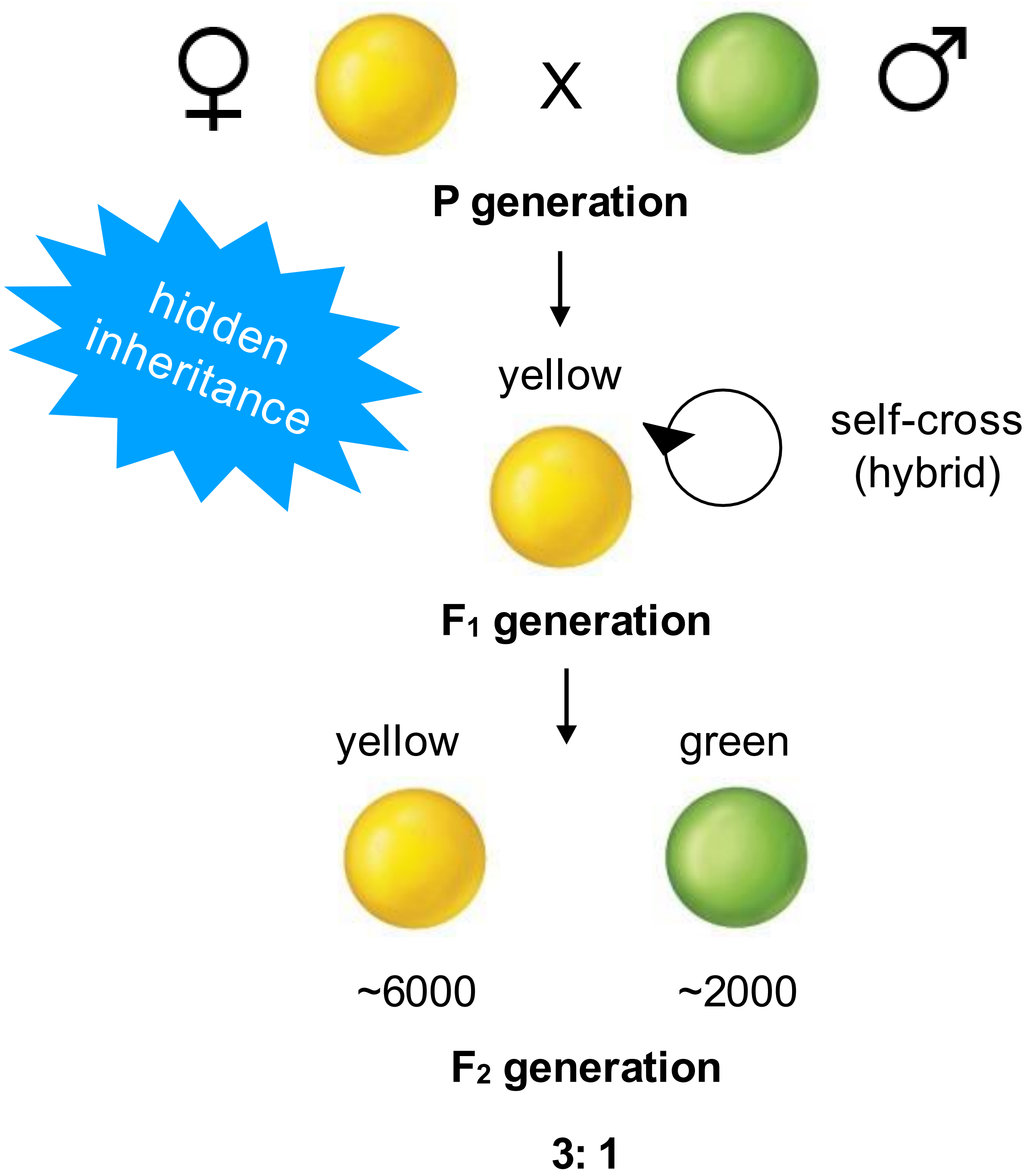


- ~~Ovist~~
- ~~Spermist~~
- ~~Blending~~

Something else is going on!

Where did the "green factor" go?

Monohybrid crosses (a cross that follows the inheritance of ONE trait)



STEP 3: Let F_1 Self-Pollinate

- $F_1 \times F_1 = F_2$ generation (Second Filial generation)
- This is where the magic happens—hidden traits reappear!
- The "green factor" didn't disappear, it was hidden.

STEP 4: COUNT Everything

- Mendel counted thousands of plants
- He found mathematical ratios: 3:1 patterns
- This proved inheritance follows rules, not randomness

Yellow is "dominant" over green ("recessive")

Monohybrid crosses (a cross that follows the inheritance of ONE trait)

ICA Q2: Mendel counted thousands of pea plants. Why was it important to count so many instead of just 10 or 20? Hint: Think about what would happen if you flipped a coin only 4 times vs. 400 times.

All 3:1

A e Results of Mendel's Garden Pea Hybridizations

Characteristic	Contrasting P_0 Traits	F_1 Offspring Traits	F_2 Offspring Traits	F_2 Trait Ratios
Flower color	Violet vs. white	100 percent violet	705 violet 224 white	3.15:1
Flower position	Axial vs. terminal	100 percent axial	651 axial 207 terminal	3.14:1
Plant height	Tall vs. dwarf	100 percent tall	787 tall 277 dwarf	2.84:1
Seed texture	Round vs. wrinkled	100 percent round	5,474 round 1,850 wrinkled	2.96:1
Seed color	Yellow vs. green	100 percent yellow	6,022 yellow 2,001 green	3.01:1
Pea pod texture	Inflated vs. constricted	100 percent inflated	882 inflated 299 constricted	2.95:1
Pea pod color	Green vs. yellow	100 percent green	428 green 152 yellow	2.82:1

Monohybrid crosses (a cross that follows the inheritance of ONE trait)

ICA Q2:
Large sample sizes give more accurate results.

With only 10-20 plants, random chance could easily skew the results. You might get 15 tall and 5 short just by luck, even if the true ratio should be 3:1.

With thousands of plants, random variations balance out and the observed ratio gets much closer to the true genetic ratio (like 3:1 or 9:3:3:1).

The coin flip analogy:
•Flip 4 times: You might get 4 heads (100%) just by chance
•Flip 400 times: You'll get very close to 50% heads, 50% tails
Same principle - larger samples = more reliable patterns.

Mendel's explanation

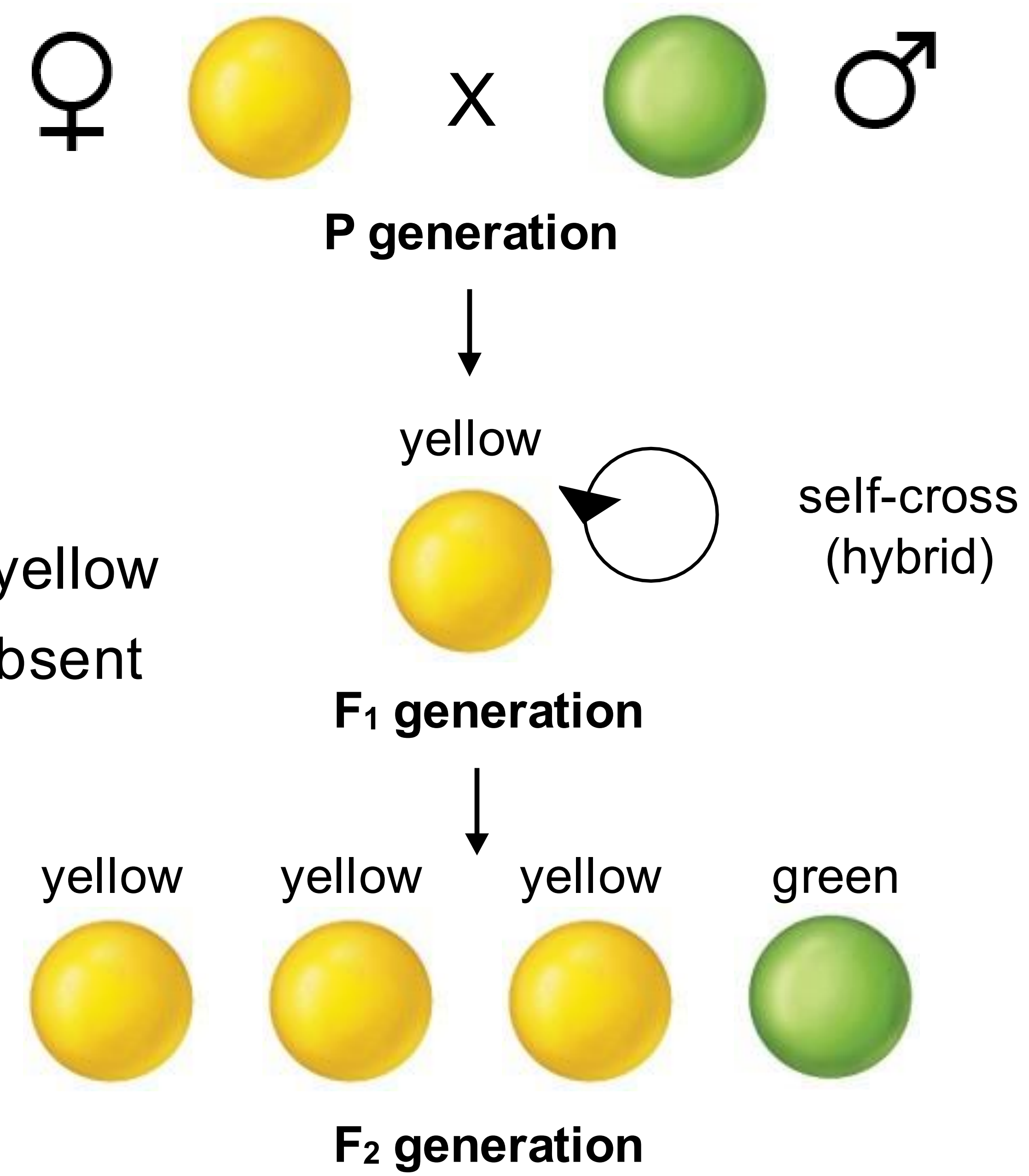
Mendel's Hypothesis:

1. Each parent has **TWO** hereditary units (we now call these **alleles – Y vs y**)
2. These alleles are **different forms of a gene**
3. During reproduction (aka meiosis), alleles **segregate** (separate)
4. Each parent passes on **only ONE** allele to offspring

Uppercase Y = **dominant** allele, when *Y* present = yellow

Lowercase y = **recessive** allele, green when *Y* is absent

DNA (The entire library)
↓
Chromosome (A book in the library)
↓
Gene (A recipe/chapter in the book)
↓
Allele (Chocolate vs. vanilla version of the recipe)
↓
Trait (The cake you actually see/eat)



ICA Q3: For each F2 offspring, predict what alleles each pea has. Each pea should have 2 alleles. Some peas may have multiple options.

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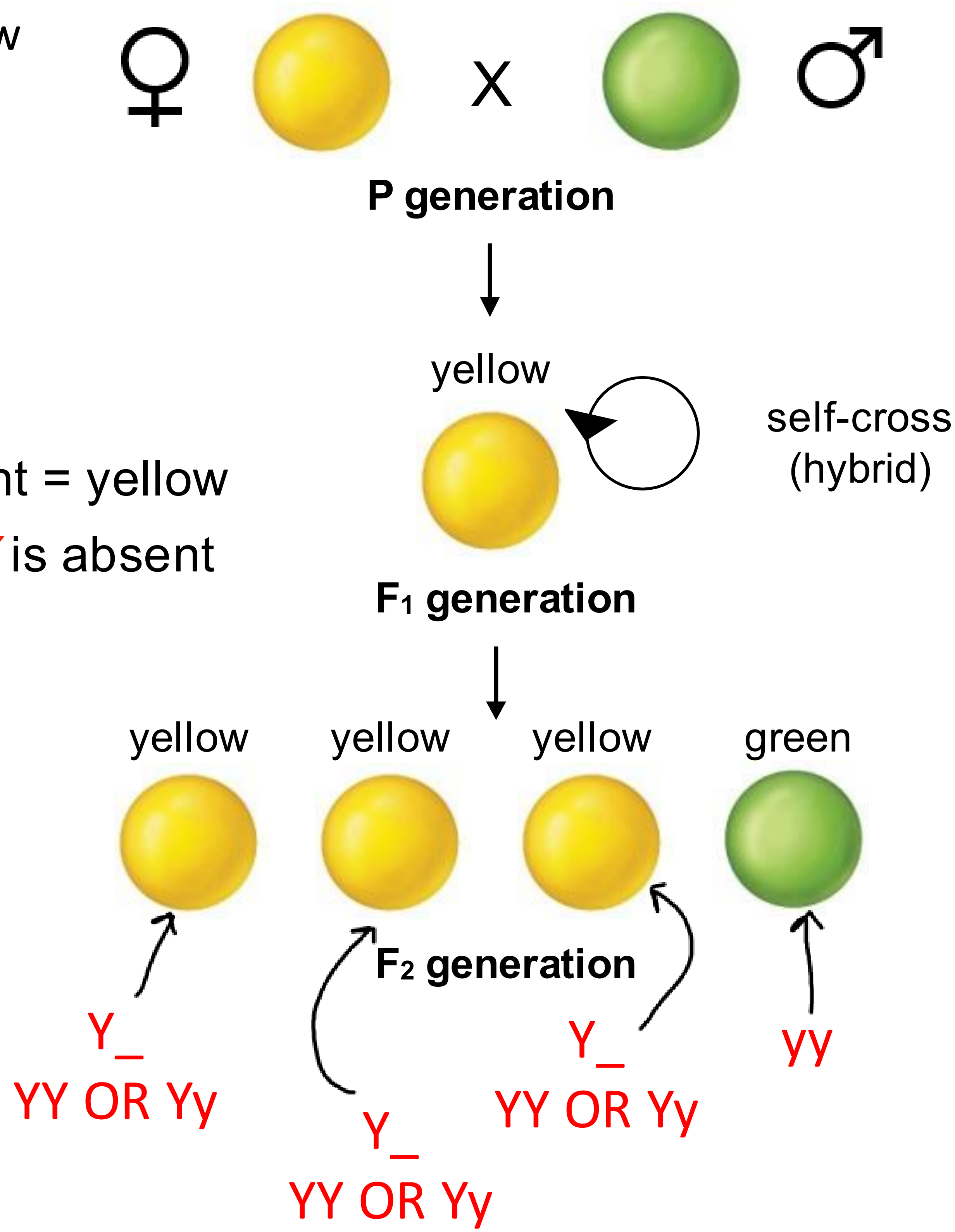

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Mendel's explanation

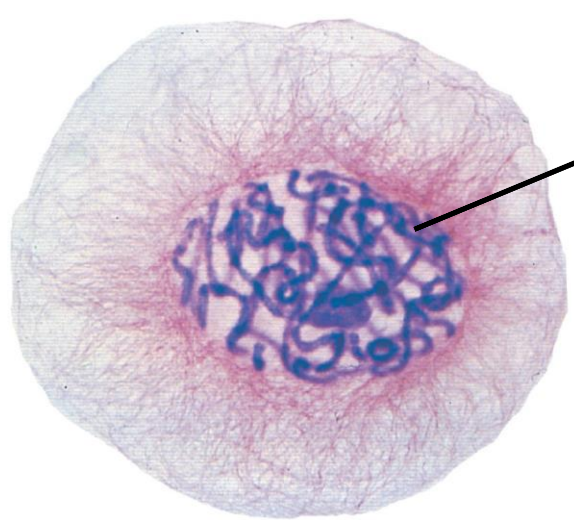
In peas (and humans), each parent has TWO heredity units (alleles)

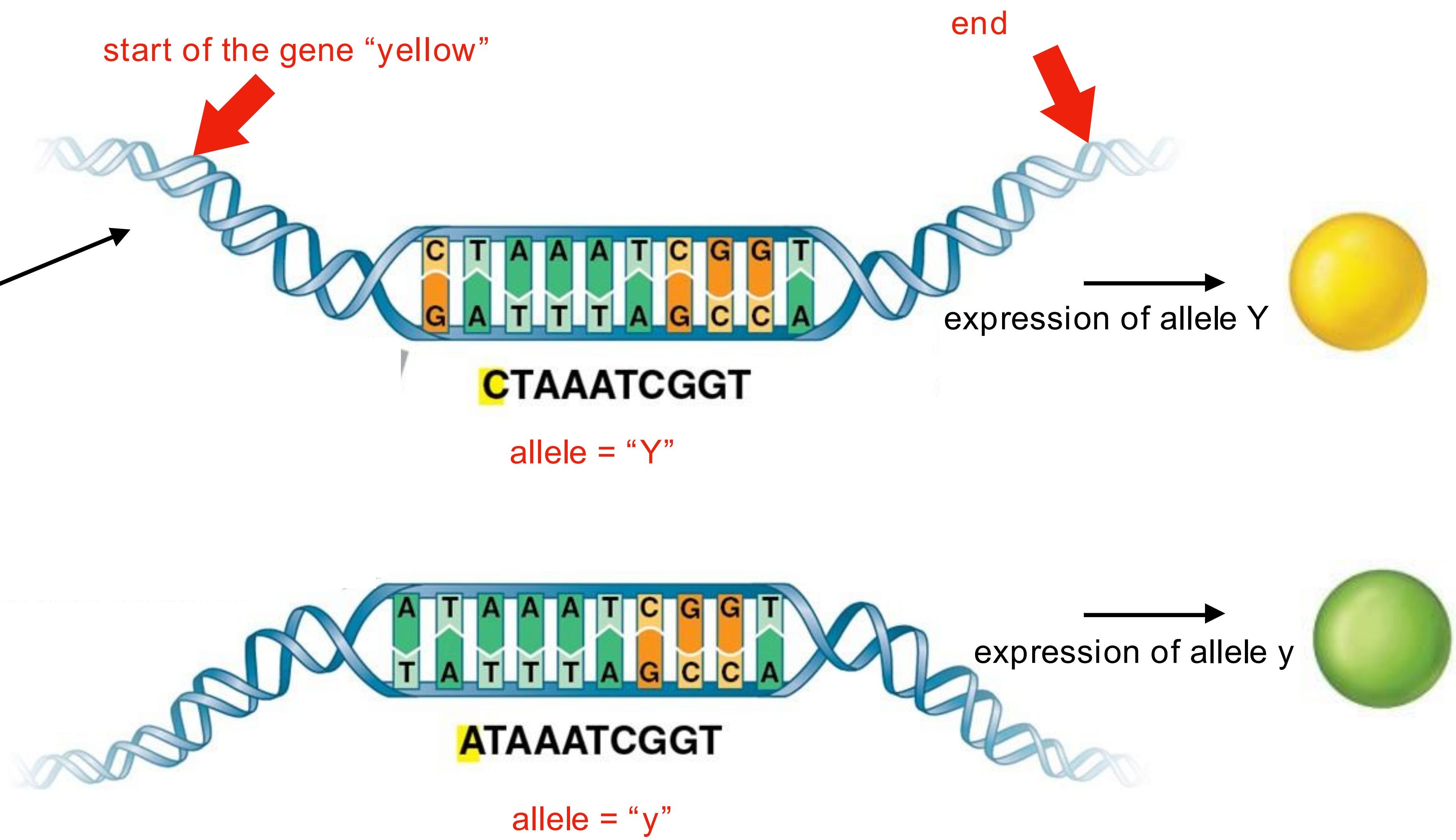
"**alleles**" = different forms of a gene

gene for seed color has 2 alleles: "*Y*" and "*y*"

there can be many versions of a gene that have small differences in their sequence

inside the nucleus of a cell, DNA is organized into linear chromosomes during M-phase of the cell cycle





Genes DON'T change—you always have an 'eye color gene.' But ALLELES can differ—you might have the blue version or the brown version!

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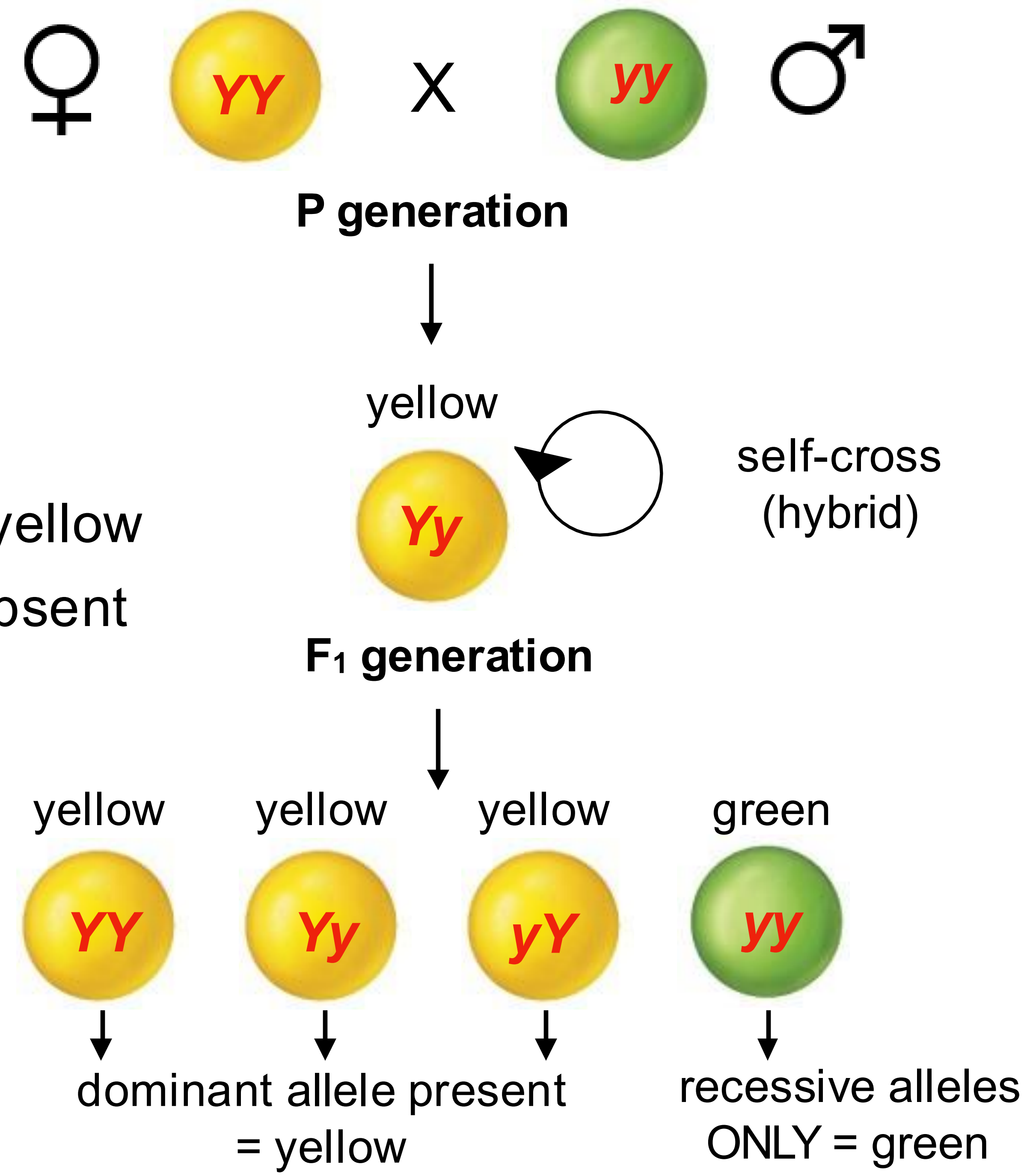


Uppercase Y = **dominant** allele, when *Y* present = yellow

Lowercase y = **recessive** allele, green when *Y* is absent

When you have true breeding organisms, they are “**homozygous**” = both alleles are the same (one from each parent)

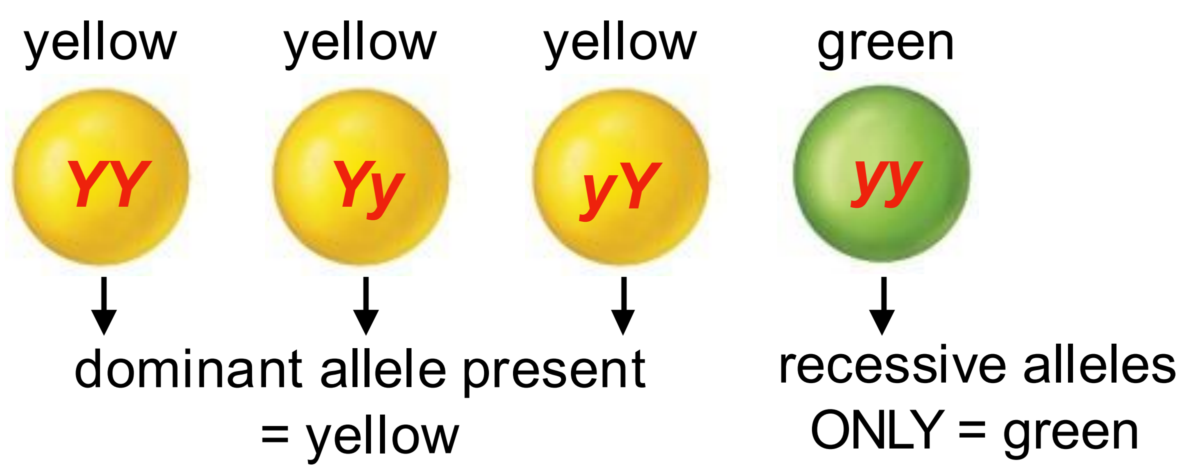
The F_1 hybrids are “**heterozygous**” = have 2 different alleles (one from each parent)
F_1s will express the dominant trait because they have the dominant alleles

In the F_2 generation, the alleles segregate and combine randomly so that 4 combinations are possible

3: 1

ICA Q4: Look at the 4 offspring and label each with one of the following: homozygous dominant, homozygous recessive, OR heterozygous.

ICA Q4:



YY = Homozygous dominant
Yy = Heterozygous
yy = Homozygous recessive

Dominant vs. Recessive: Who Wins?

Think of alleles like speakers

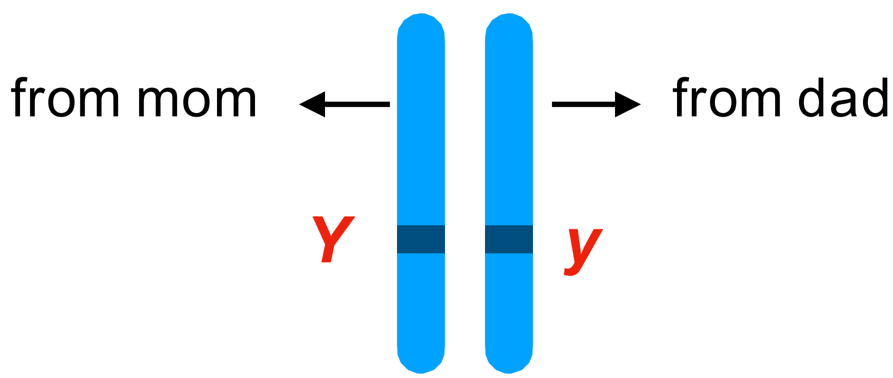
YY = LOUD (2 working speakers)
↓
LOUD SOUND = Yellow seeds

Yy = LOUD (1 working, 1 broken)
↓
Still LOUD enough! = Yellow seeds

yy = Silent (2 broken speakers)
↓
Quiet = Green seeds

Mendel didn't know about chromosomes

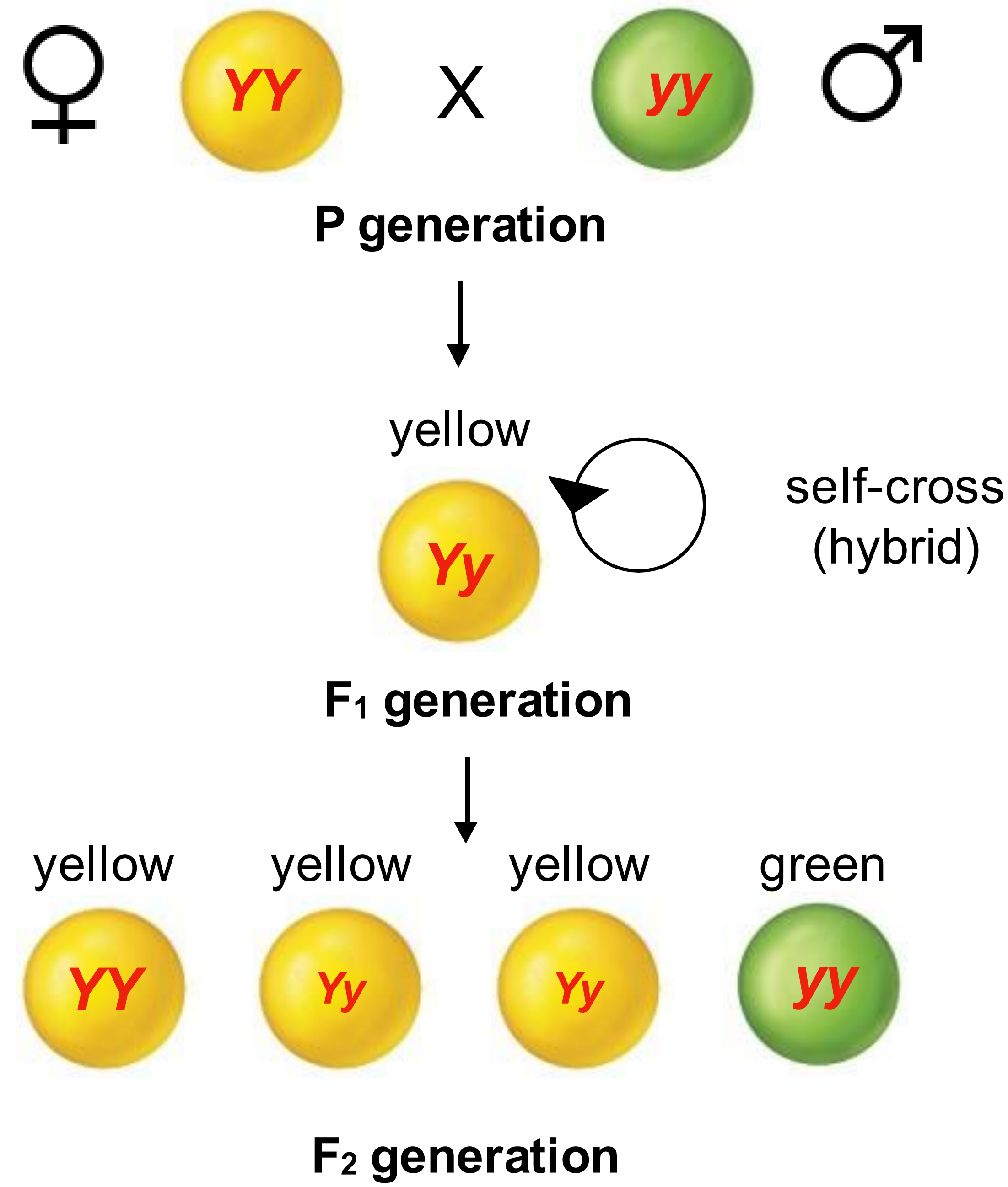
diploid: 2 copies of every chromosome (1 paternal, 1 maternal)



For this to work, each parent passes on only 1 chromosome

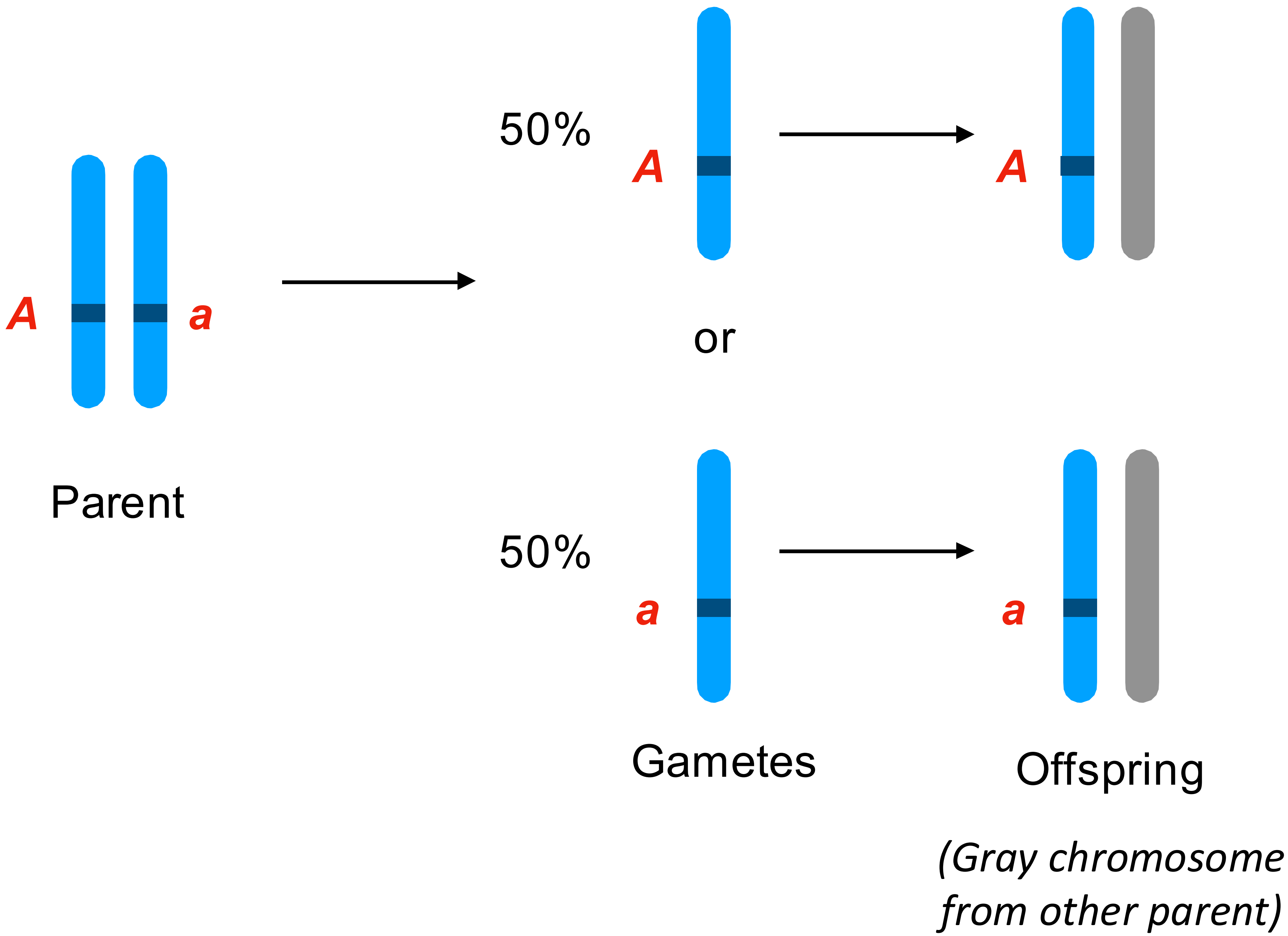
This is true from what we know about meiosis!!

- To mate, parents produce "gametes" (sperm/egg or pollen/ovule)
- Gametes have 1/2 the number of chromosomes
    - How?? MEIOSIS!

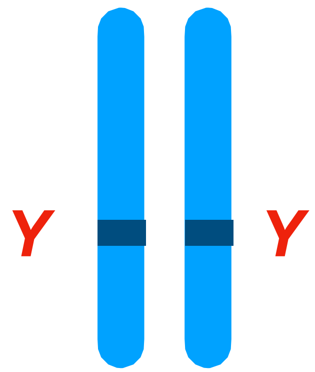
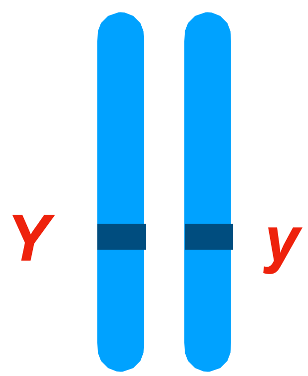
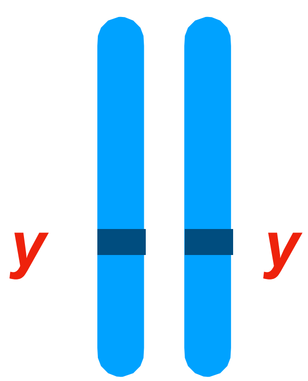


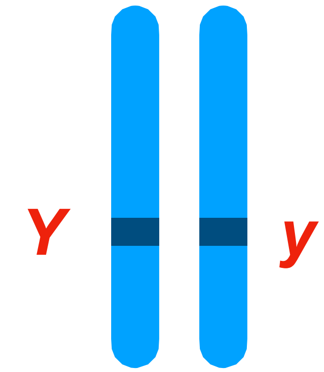
Mendel's 1st rule of inheritance: The law of segregation

- Individuals carry 2 hereditary units (alleles of genes) that control each trait
- Alleles are segregated into separate gametes



For every trait, there are “genotypes” and “phenotypes”

Genotype	Phenotype
	1: yellow
	2: yellow
	3: green



every individual has a genotype for every trait

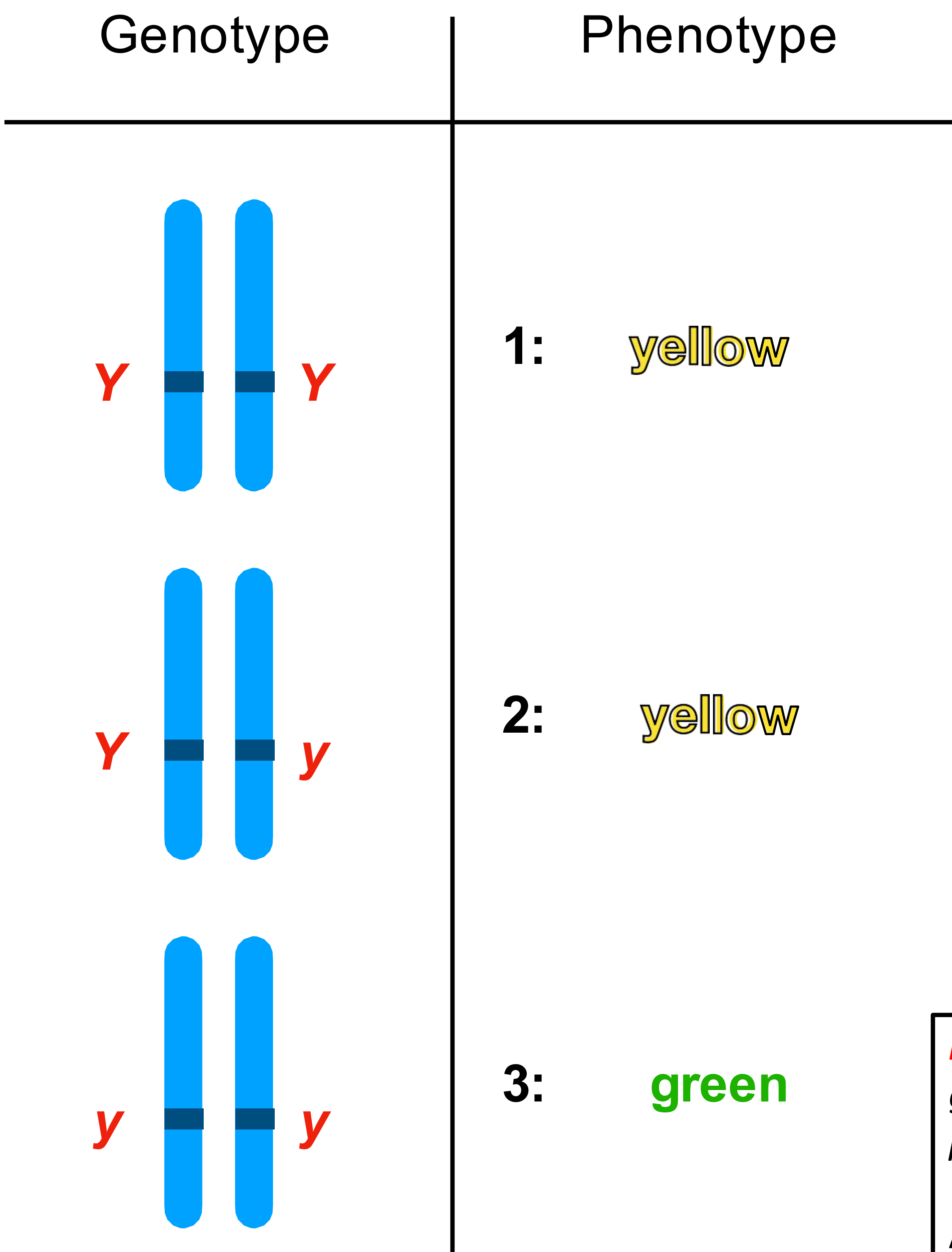
genotypes are the result of 2 alleles at a time

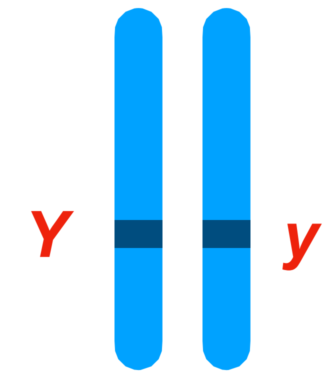
the phenotype is the “appearance”, which depends on the genotype

ICA Q5: *Two yellow pea plants are crossed and produce some green offspring. What can you conclude about the parent plants' genotypes?*

A) Both parents must be YY
B) Both parents must be Yy
C) One parent is YY, one is Yy
D) Both parents must be yy

For every trait, there are "genotypes" and "phenotypes"





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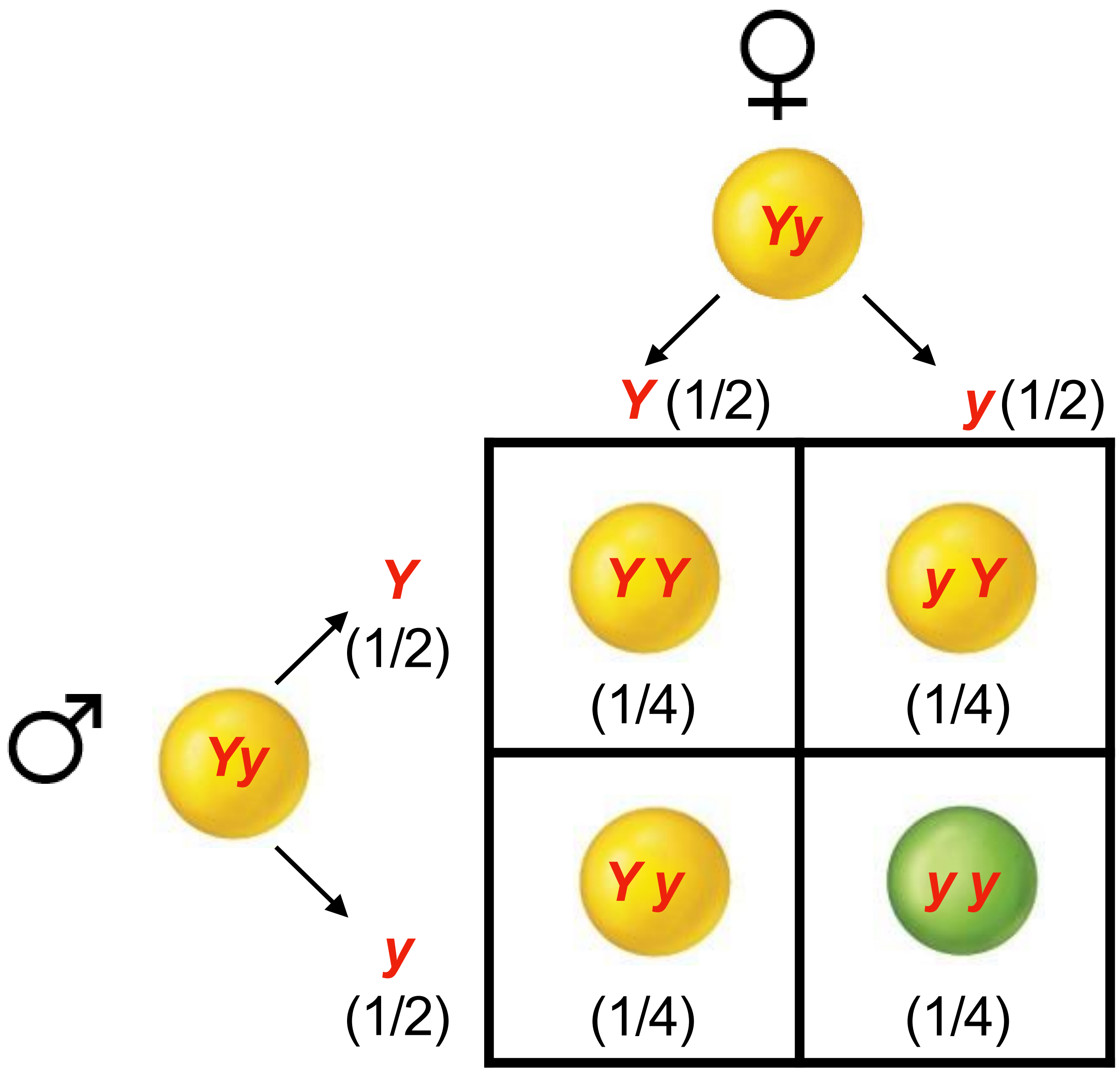
Punnett squares: visualize crosses



3 genotypes, 2 phenotypes

YY, Yy, yy

yellow
green

genotypic ratio = 1 : 2 : 1
25% = *YY*
50% = *Yy*
25% = *yy*

phenotypic ratio = 3 : 1
75% = yellow
25% = green

A Punnett Square is like a multiplication table for genetics—it shows ALL possible offspring combinations!

Step-by-step construction

STEP 1: Write Parent Genotypes

- Parent 1: YY
- Parent 2: Yy
- Cross: YY× Yy

STEP 2: Determine Gametes Each Parent Makes

- Parent 1 (YY) can make: Y or Y (50% each, both Y – 100% Y)
- Parent 2 (Yy) can make: Y or y (50% each)

STEP 3: Draw the Grid

- 2×2 grid for monohybrid cross (one trait, 2 alleles)

STEP 4: Label the Grid

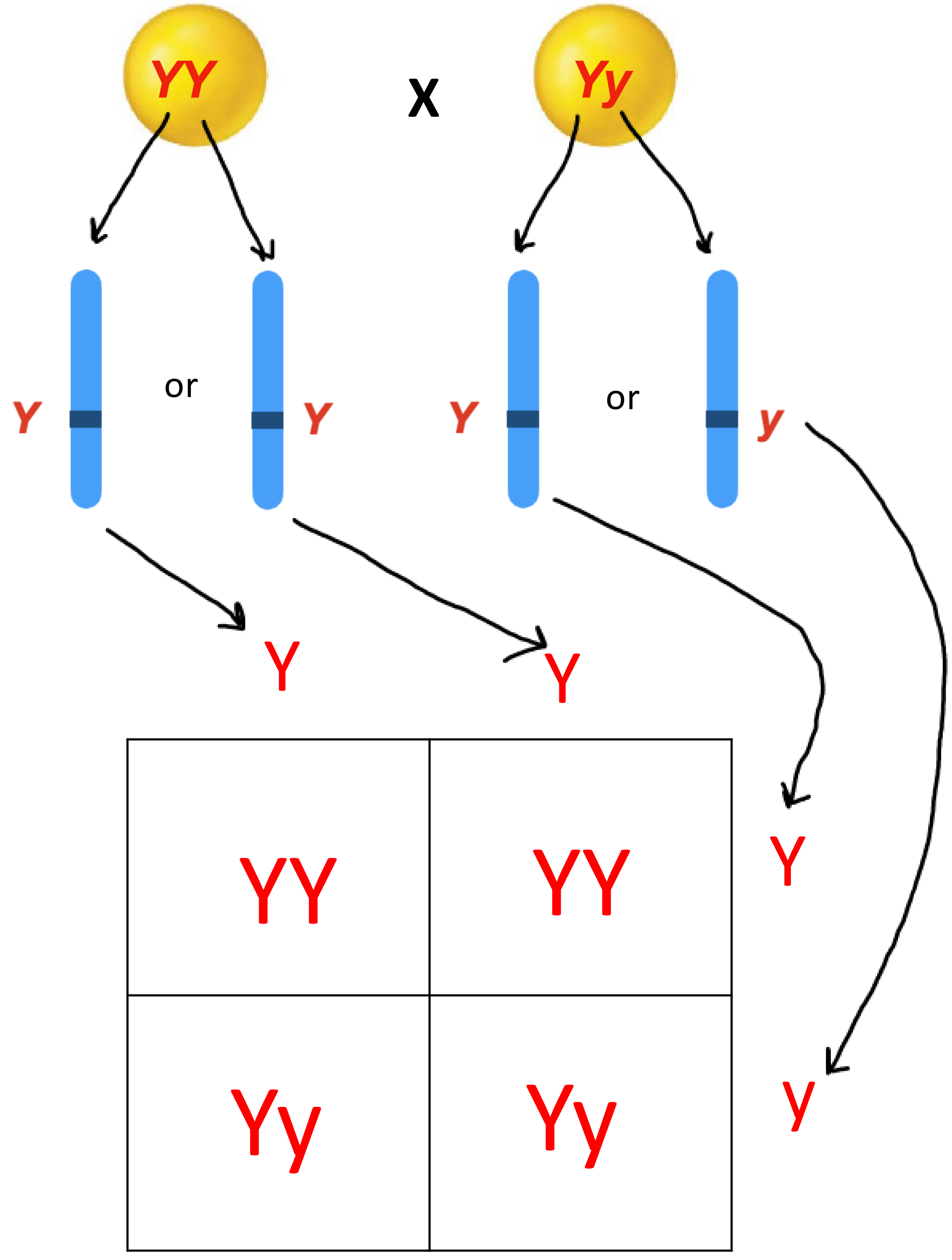
- Put Parent 1's gametes across the TOP
- Put Parent 2's gametes down the SIDE

STEP 5: Fill in the Boxes

- Combine the gametes (like multiplying)—whatever's on top joins with whatever's on the side!

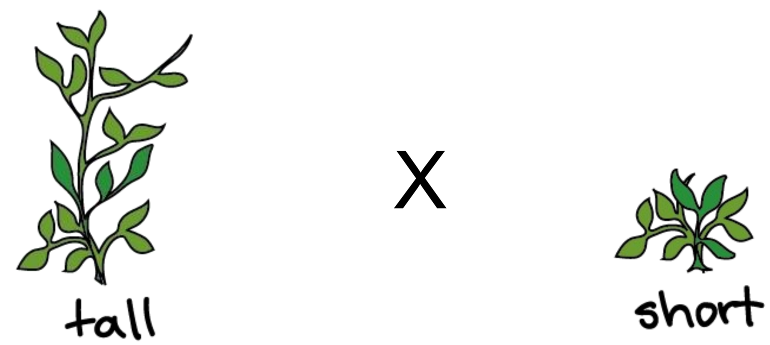
STEP 6: Count the Results

- Each box = one possible offspring
- In this example, 50% YY, 50% Yy
- 100% yellow



ICA Q6: Try it yourself, use a Punnett square to answer the following

<u>The cross:</u>

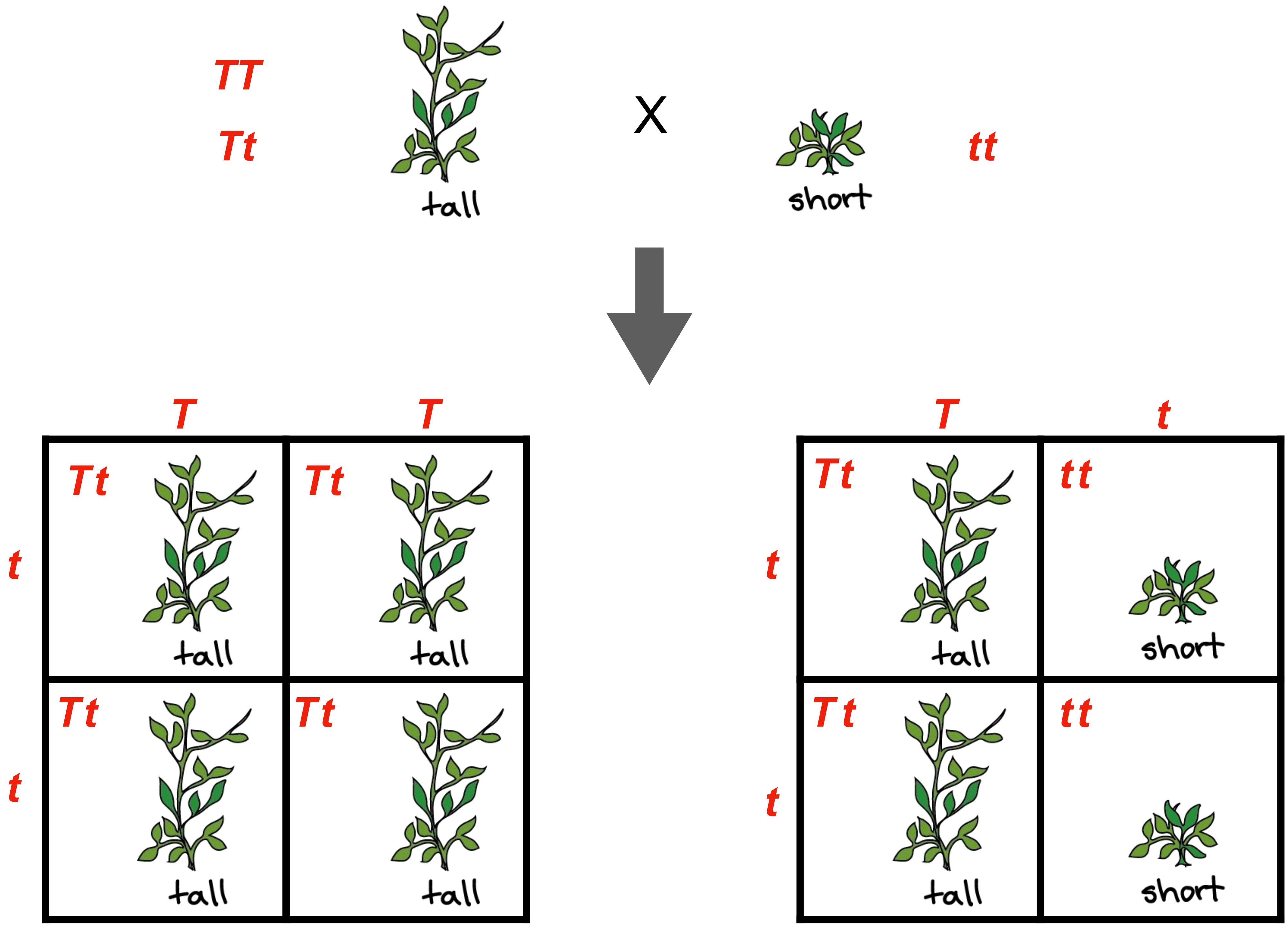


What are the possible genotypes for each parent?
(assume height is controlled by one gene with two alleles: *T* and *t*, where *T* (tall) is dominant to *t* (short))

What is the phenotypic ratio?

What is the genotypic ratio?

Try it yourself, use a Punnett square to answer the following



Phenotypic ratio:100% tall
Genotypic ration: 100% Tt

Phenotypic ratio: 50% tall, 50% short or 2:2 or 1:1
Genotypic ratio: 50& Tt, 50% tt or 2:2 or 1:1

Common Punnett Square Mistakes to Avoid

Mistake 1: Putting the same allele in every gamete box

- Remember: Each box represents a DIFFERENT possible offspring

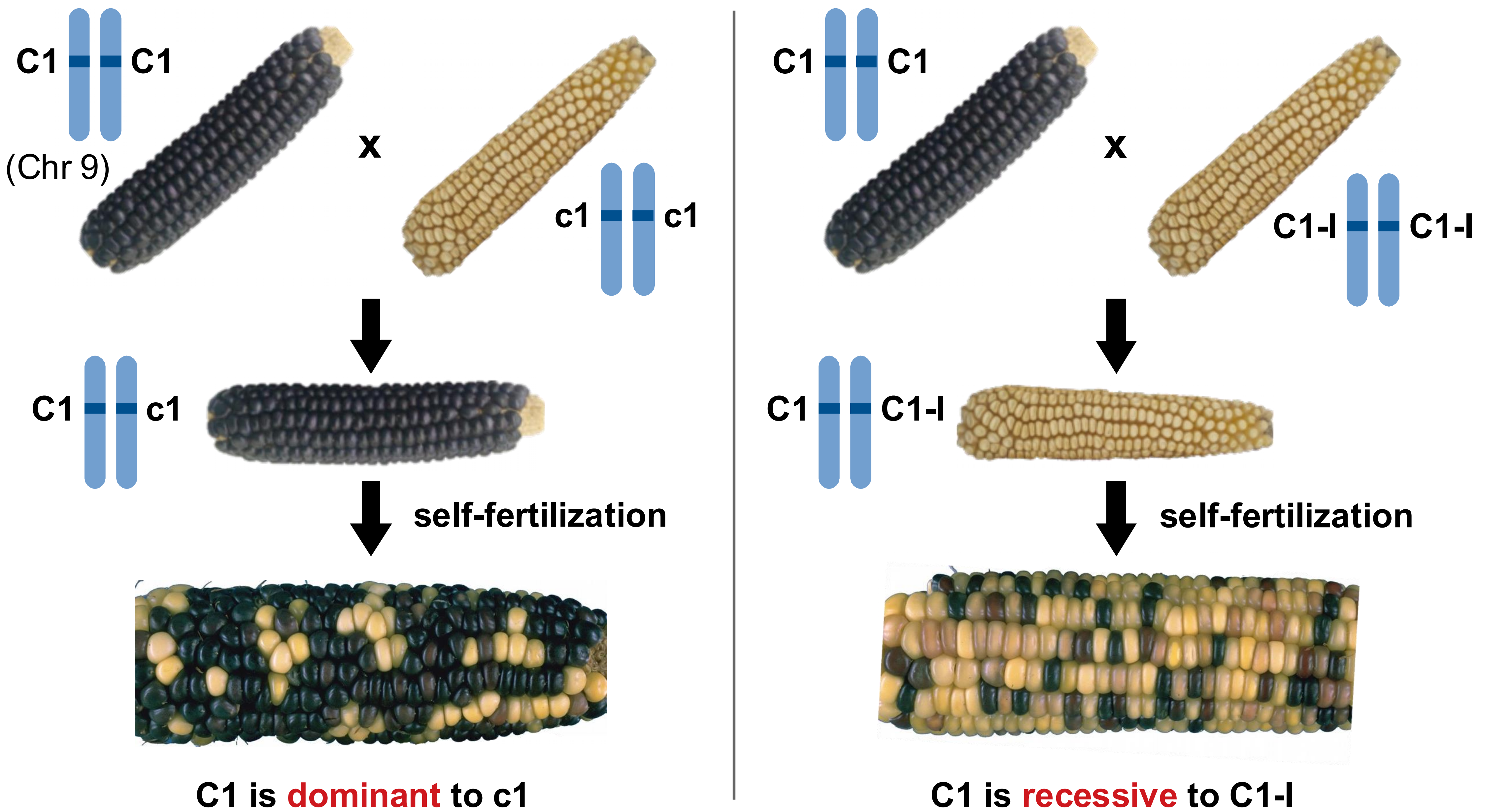
Mistake 2: Forgetting that each parent contributes only ONE allele

- Gametes are haploid (one allele per gene)

Mistake 3: Confusing genotypic and phenotypic ratios

- Genotypic: Counts allele combinations (YY, Yy, yy)
- Phenotypic: Counts appearances (yellow, green)

Dominance is not inherent



Dominance/recessiveness is a relationship between 2 alleles

Also: traits can be controlled by MULTIPLE alleles of the same gene, not just two

What makes an allele dominant?

Using our pea examples:

Dominant allele (Y): Produces a FUNCTIONAL enzyme that makes yellow pigment
Recessive allele (y): Broken gene—makes NO enzyme

Key: You only need ONE working copy to make yellow pigment!

Genotype → Enzymes working → Pigment made? → Seed color
YY → ✓✓ (both work) → YES (lots!) → Yellow
Yy → ✓✗ (one works) → YES (enough) → Yellow
yy → ✗✗ (none work) → NO → Green

MYTH: Dominant = more common or 'better'
TRUTH: Dominant just means it shows up with one copy.
****Many genetic diseases are actually dominant!

There are degrees of dominance

*Mendel's pea plants all followed **complete dominance** (one allele completely masks the other). But not all traits work this way!*

Complete dominance

- The pea examples we have been looking at have been complete dominance
- Y is COMPLETELY dominant over y

Incomplete dominance

- Alleles blend together

Codominance

- Both alleles show up in the phenotype

Complete dominance

PP
Pp

P allele → produces purple pigment

has 1 non-functional allele, but because the P allele is making pigment, flower is still purple

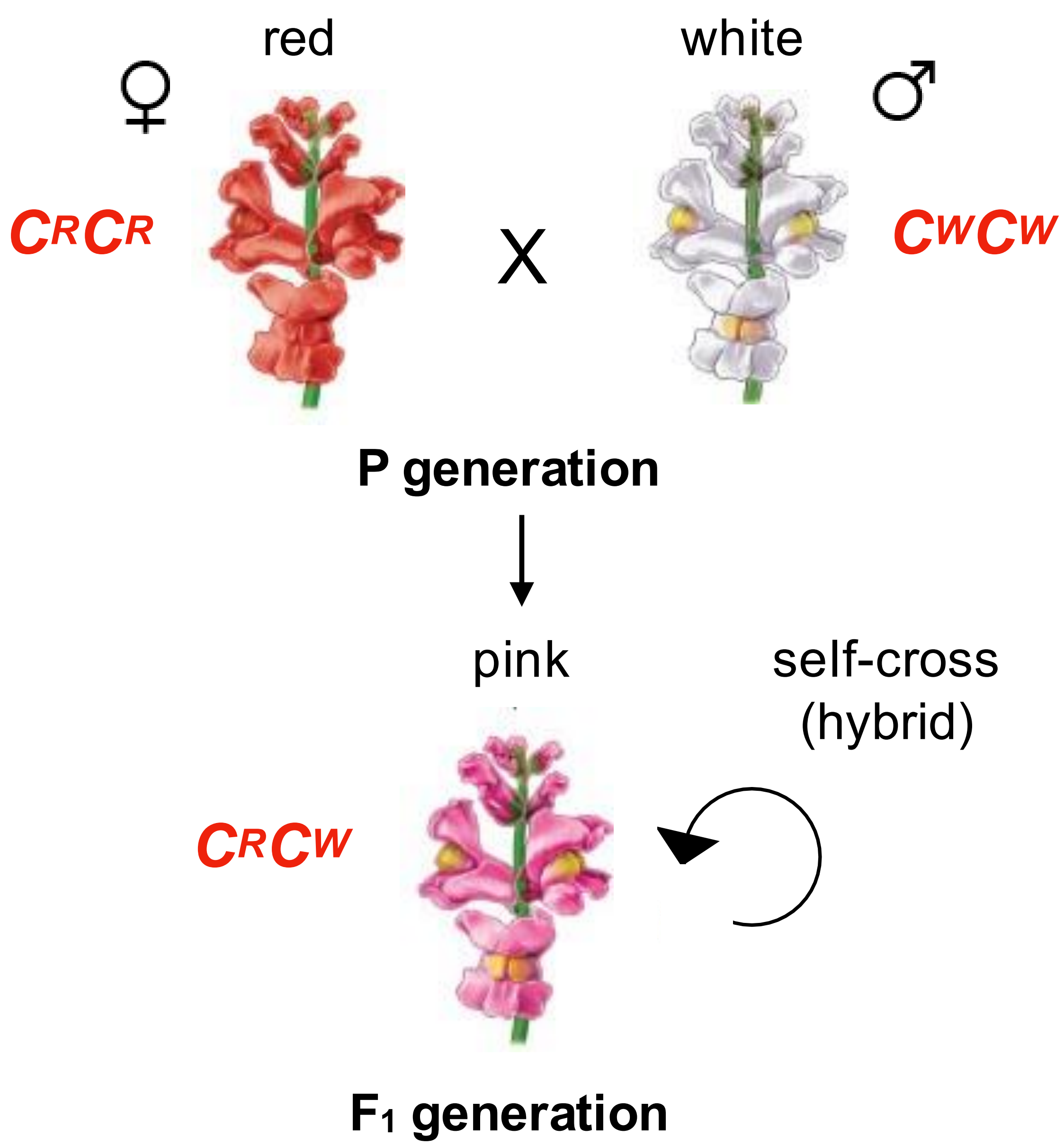
pp

p allele → non-functional gene → no pigment

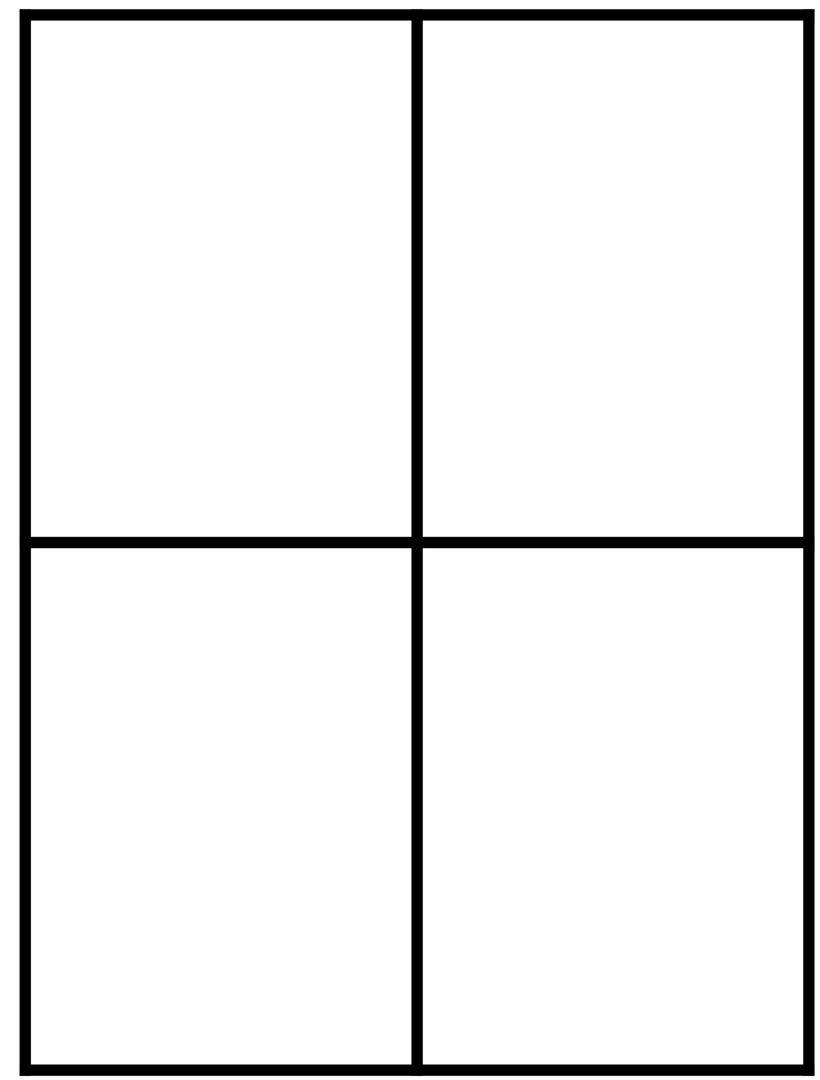
PP → 2 working enzymes → LOTS of purple pigment → Purple flower
Pp → 1 working enzyme → ENOUGH purple pigment → Purple flower
pp → 0 working enzymes → NO purple pigment → White flower

Incomplete dominance

With complete dominance: Yy looks like YY (one allele wins)
With incomplete dominance: C^R C^w looks DIFFERENT from both parents (blend!)



ICA Q7



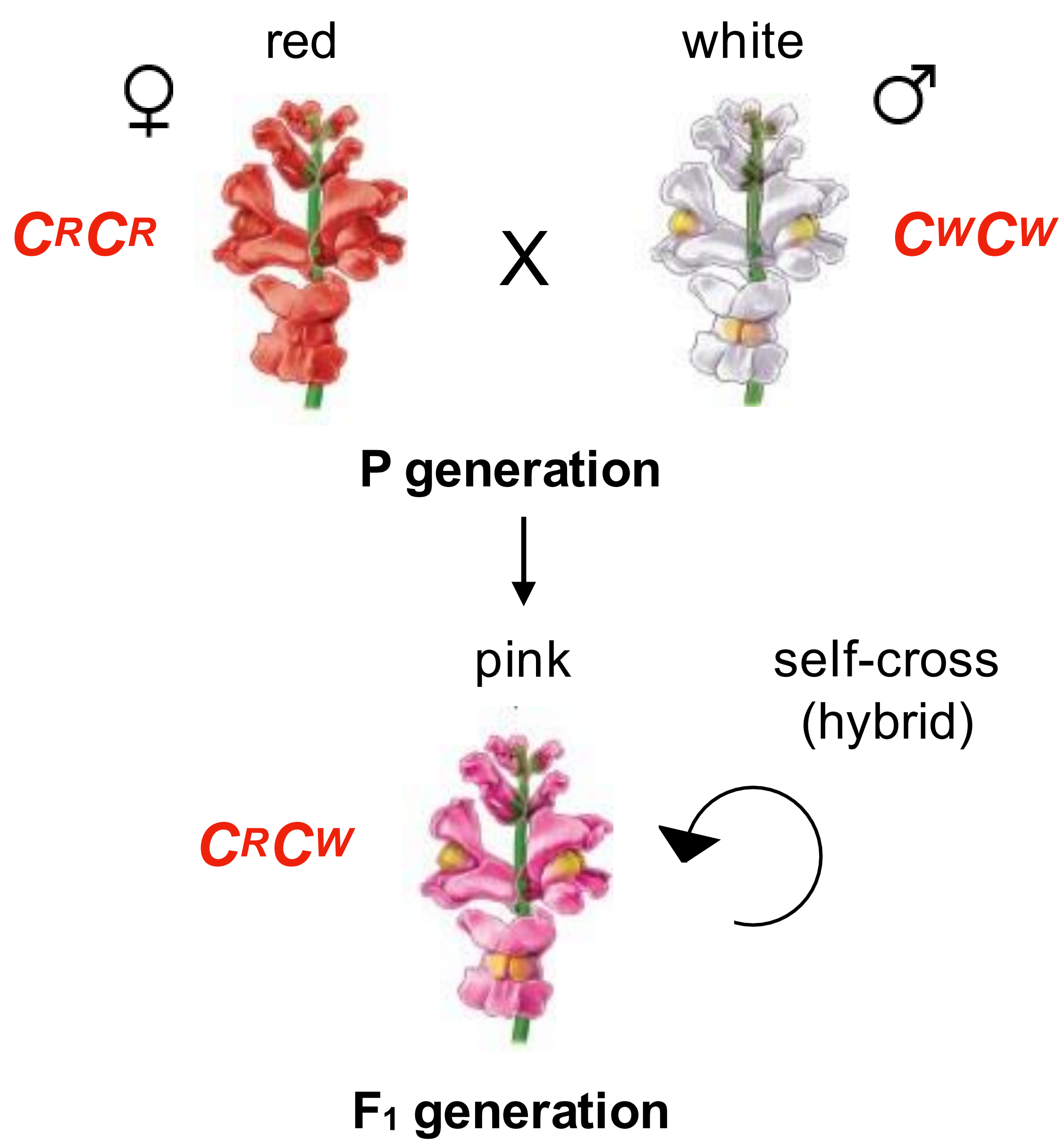
C^R C^R → Makes LOTS of red pigment → **Red**

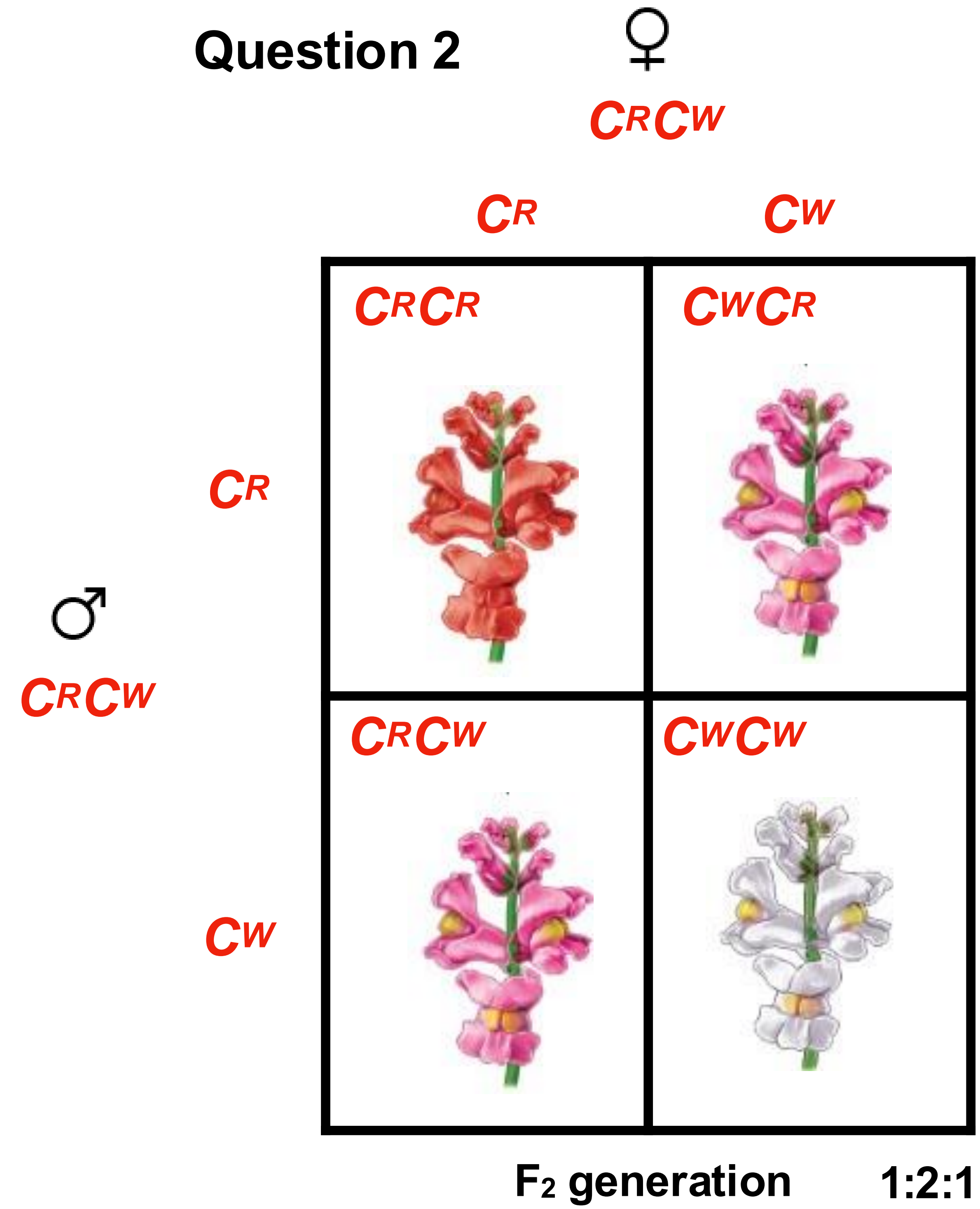
C^R C^W → Makes SOME red pigment (only one copy of C^R) → Makes SOME white pigment → Diluted red = **Pink**

C^W C^W → Makes NO red pigment → Makes LOTS of white pigment → White

Incomplete dominance

With complete dominance: Yy looks like YY (one allele wins)
With incomplete dominance: $C^R C^w$ looks DIFFERENT from both parents (blend!)

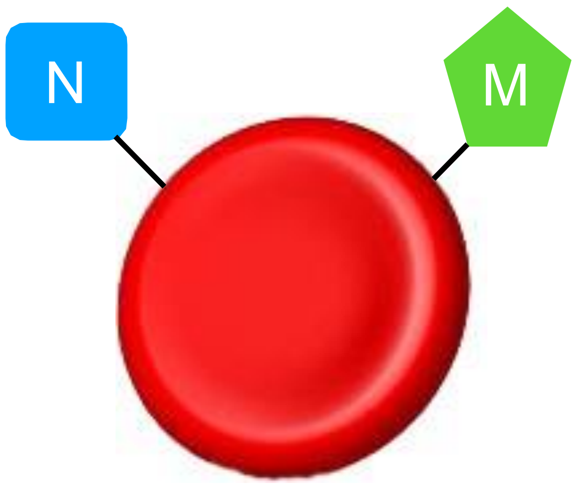




1:2:1 phenotypic ratio (every genotype looks different!)

Codominance

both alleles for the same characteristic are **simultaneously** expressed

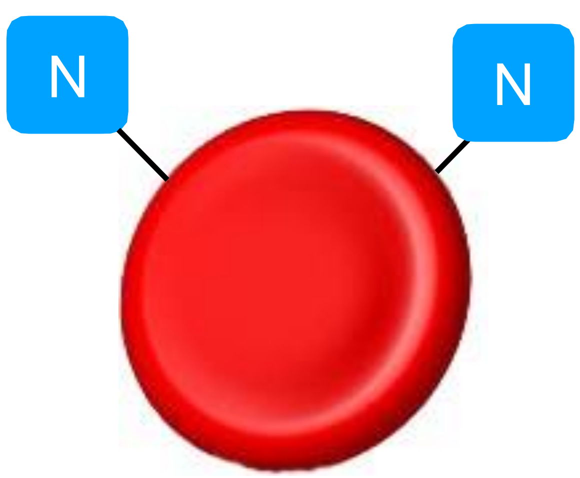


MNS blood group system - a classification of human blood based on the presence of different chemicals found on red blood cells

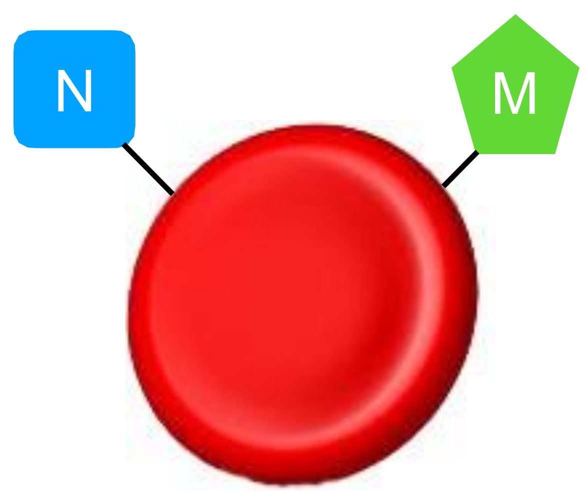
One gene → N allele (L^N)
→ M allele (L^M)

these alleles are codominant

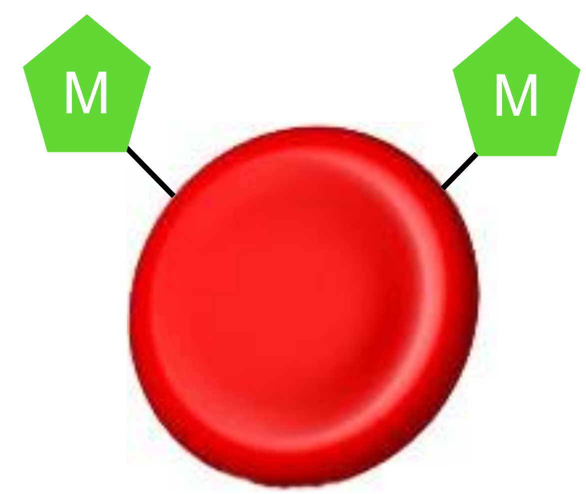
$L^N L^N$



$L^N L^M$



$L^M L^M$



$L^M L^M$ → M markers only on red blood cells
$L^M L^N$ → BOTH M and N markers on red blood cells (both expressed!)
$L^N L^N$ → N markers only on red blood cells

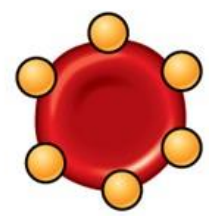
There can be multiple alleles for any gene

So far: Each gene had TWO alleles (Y/y, C^R/C^W)
ABO blood: ONE gene, but THREE alleles: I^A, I^B, i

(a) The three alleles for the ABO blood groups and their carbohydrates

Allele	I^A	I^B	i
Carbohydrate	A △	B ○	none

(b) Blood group genotypes and phenotypes

Genotype	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell with surface carbohydrates				
Phenotype (blood group)	A	B	AB	O

what is the relationship between I^A and i alleles?

what is the relationship between I^A and I^B alleles?

ICA Q8

There can be multiple alleles for any gene

So far: Each gene had TWO alleles (Y/y, C^R/C^W)
ABO blood: ONE gene, but THREE alleles: I^A, I^B, i

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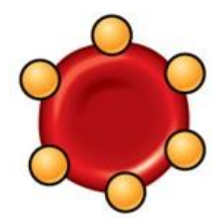
Red blood cell with surface carbohydrates				
Phenotype (blood group)	A	B	AB	O

what is the relationship between I^A and i alleles?

I^A is completely dominant to i

what is the relationship between I^A and I^B alleles?

I^A and I^B are codominant

Question 3

Can You Distinguish These Patterns?

ICA Q9: A cross between two pink flowers produces 15 red, 34 pink, and 16 white offspring. This is an example of: Hint: think about ratios AND phenotype.

A) Complete dominance
B) Incomplete dominance
C) Codominance
D) Multiple alleles

Can You Distinguish These Patterns?

ICA Q9: A cross between two pink flowers produces 15 red, 34 pink, and 16 white offspring. This is an example of: Hint: think about ratios AND phenotype.

A) Complete dominance
B) Incomplete dominance
C) Codominance
D) Multiple alleles

The 1:2:1 ratio and intermediate pink phenotype indicate incomplete dominance

- **3:1 = complete dominance**
- **Codominance = shows BOTH alleles --> pink + pink = pink (in this case)**
- **Multiple allele = means more than 2 alleles exist in the gene pool, but this 1:2:1 ratio only requires 2 alleles (one for red, one for white) showing incomplete dominance. Simple ratio = a simple answer (NOT multiple alleles)**

Common Misconceptions

Misconception #1: "Dominant = More Common"

TRUTH: Dominance describes how alleles interact, NOT their frequency

Example: Polydactyly (extra fingers/toes)

- Caused by a dominant allele of the Gli3 gene
- Occurs in only ~1 out of 400 births
- Very rare, even though it's dominant!

Misconception #2: "Dominant = Better/Normal"

TRUTH: Many genetic diseases are caused by dominant alleles!

Examples of Dominant Genetic Disorders:

- Huntington's disease
- Achondroplasia (a form of dwarfism)
- Marfan syndrome

Common Misconceptions

Misconception #3: "Genes Change, Alleles Don't"
TRUTH: It's exactly the opposite!

Correct Understanding:

- **Gene** = The location/function - stays constant
  Example: "Everyone has an eye color gene"
- **Allele** = The version - can differ
  Example: "Some people have the blue allele, others have the brown allele"

Misconception #4: "Genotype Directly Determines Phenotype"
MORE ACCURATE MODEL:
Genotype → (gene expression in specific environment) → Phenotype

Factors that influence phenotype:

- Which genes are turned on/off
- Environmental conditions
- Random developmental variation
- Interactions with other genes

Example: Himalayan rabbits

- Genotype codes for enzyme that makes dark pigment
- Enzyme only works at cool temperatures
- Result: Dark fur on ears, nose, feet (cool) but white fur on body (warm)

Why This Matters: Clinical Applications

1. Genetic Counseling
- Predict risk of inheriting genetic disorders
- Example: Cystic fibrosis (recessive) - both parents must be carriers

2. Paternity Testing
- Uses multiple genetic markers
- Example: Child with type O blood cannot have a type AB parent

3. Understanding Disease Inheritance
- Dominant disorders: Affected person usually has affected parent
- Recessive disorders: Can "skip generations"

ICA q10: **Case Study:** A couple comes for genetic counseling. Both have normal pigmentation, but each has one parent with albinism (a recessive condition).

Part A: What are the genotypes of:
- *The couple?*
- *Their parents?*

Part B: What is the probability this couple will have a child with albinism?

ICA q10: Case Study: A couple comes for genetic counseling. Both have normal pigmentation, but each has one parent with albinism (a recessive condition).

Part A: What are the genotypes of:

Let's use: A = normal pigmentation, a = albinism

The couple: Both are **Aa** (heterozygous)

They have normal pigmentation, so they must have at least one A
Each has one parent with albinism (aa), so they must have inherited one a allele

Their parents:
The albino parents: **aa**
The normal parents: **AA** or **Aa** (we can't tell which, but doesn't matter for the problem)

ICA q10: **Case Study:** A couple comes for genetic counseling. Both have normal pigmentation, but each has one parent with albinism (a recessive condition).

Part B: What is the probability this couple will have a child with albinism?

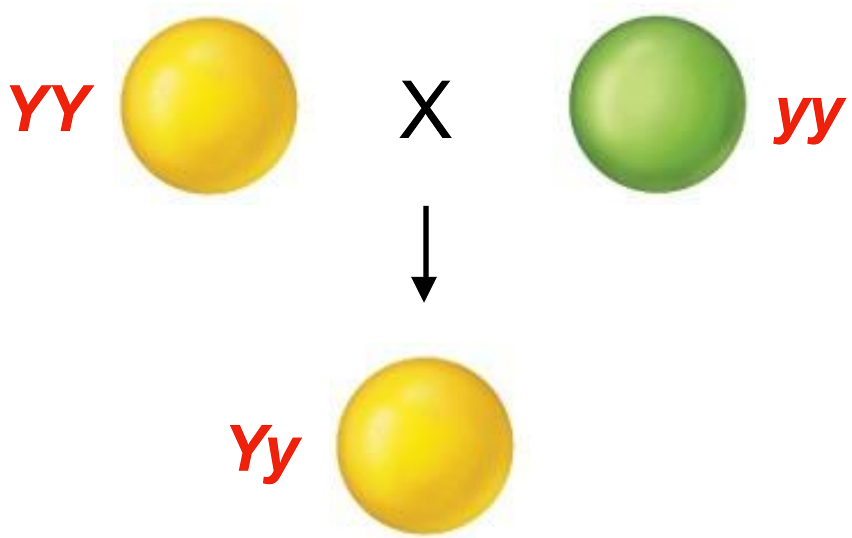
Cross: Aa × Aa
Possible offspring:
AA (normal)
Aa (normal)
Aa (normal)
aa (albinism)
Probability = 1/4 or 25%

Conclusions

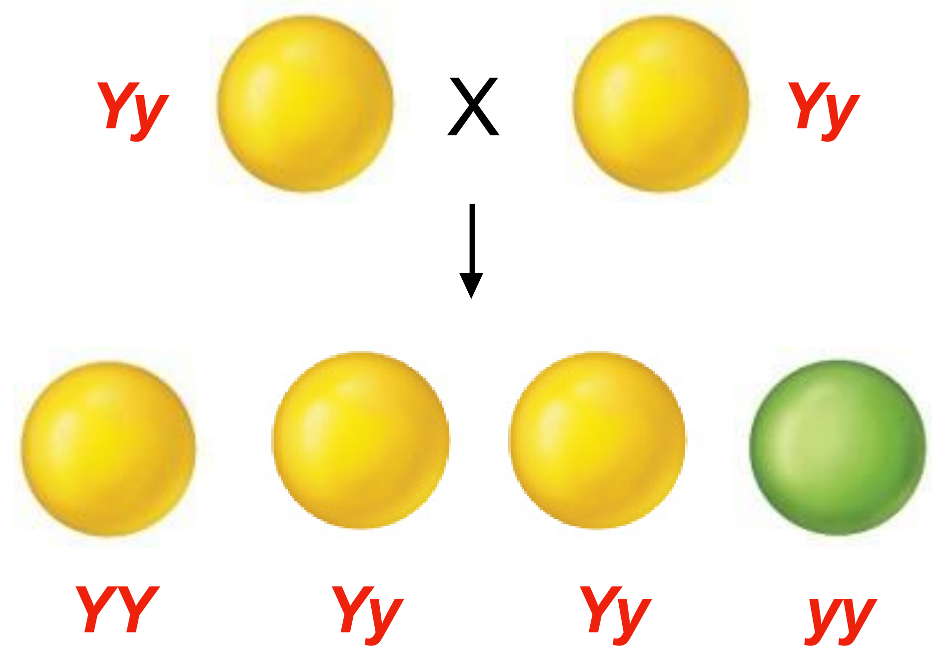
1. There's no magic to inheritance:
   Traits are inherited based on segregation of alleles

2. Alleles can be dominant, recessive, incomplete dominant, or codominant
   The phenotype you see is the consequence of the relationship between the alleles in the genotype

3. Every gene can have multiple alleles, not just two



even if you express the dominant trait,
you could be carrying a recessive allele



the recessive trait can re-emerge,
even if the parents express the dominant trait

Announcements

Problem set 3A due TODAY
Problem set 3B will be available today

Exam 3 is coming up quickly – Monday, November 10th