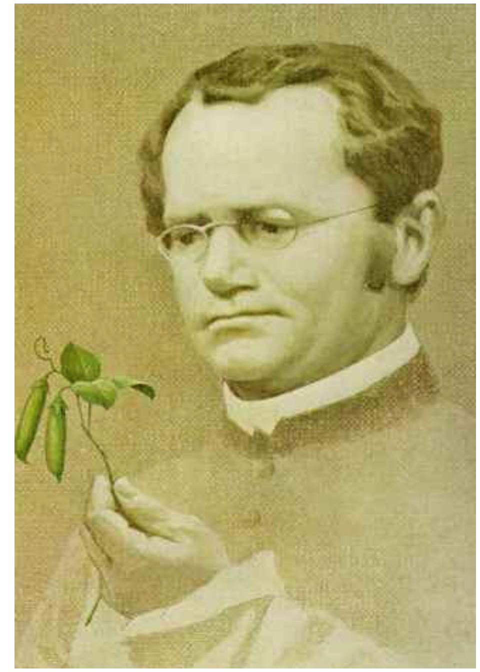
Ch.14 & 15 - Part 1

Genetics & The Work of Mendel

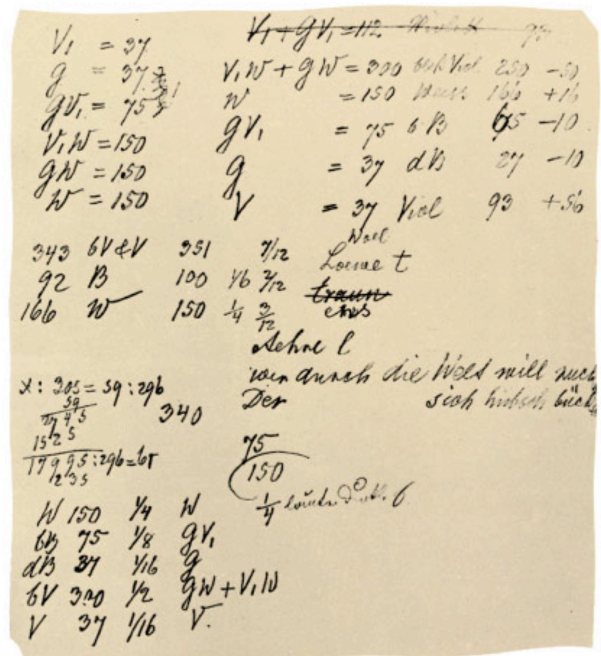
Gregor Mendel

"The father of modern genetics"



Modern genetics began in the mid-1800s in an abbey garden, where a monk named Gregor Mendel documented inheritance in peas which he began breeding around 1857.

- He had studied at the University of Vienna from 1851 to 1853 where he was influenced by
  - a physicist, Christian Doppler, who encouraged experimentation and the application of mathematics to science
  - a botanist, Franz Unger, who aroused Mendel's interest in the causes of variation in plants.
- In the monastery, he carefully planned experiments
- He used quantitative analysis
  - collected data by recording the number of pea plants exhibiting traits he was interested in
    - excellent example of scientific method



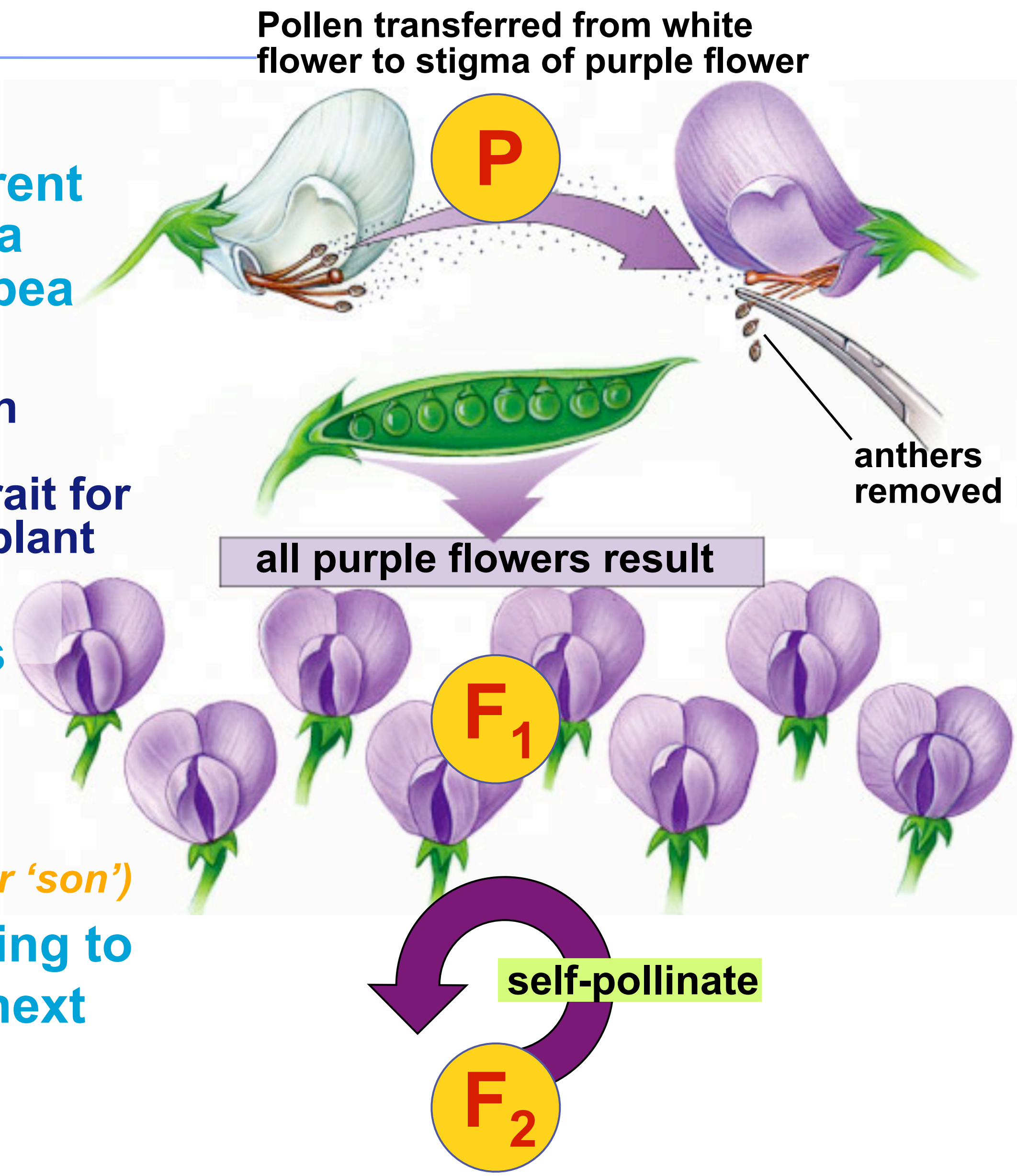
Terminology

- Heritable features that vary among individuals are called characters.
  - Ex: Flower Color
- Each variant for a character is termed a trait
  - Ex: Purple or White

- Breeding pea plants was advantageous for Mendel:
  - Short generation time
  - Large number of offspring per reproductive cycle
  - Easy to pollinate manually so you can control which parents breed to produce offspring
  - Have characters that vary between two distinct traits or alternatives

Mendel's work

- Bred pea plants
  - Cross-pollinated two different true breeding parents (P), a white and a purple flower pea plant
    - These are plants that when allowed to self-pollinate produced only the same trait for a character as the parent plant
      - P generation
  - Raised the resulting seeds & then observed traits in the next generation, the F_1 generation
      - *F stands for filial (Latin for 'son')*
  - Then, he allowed F_1 offspring to self-pollinate & observed next generation (F_2)
      - F_2 generation

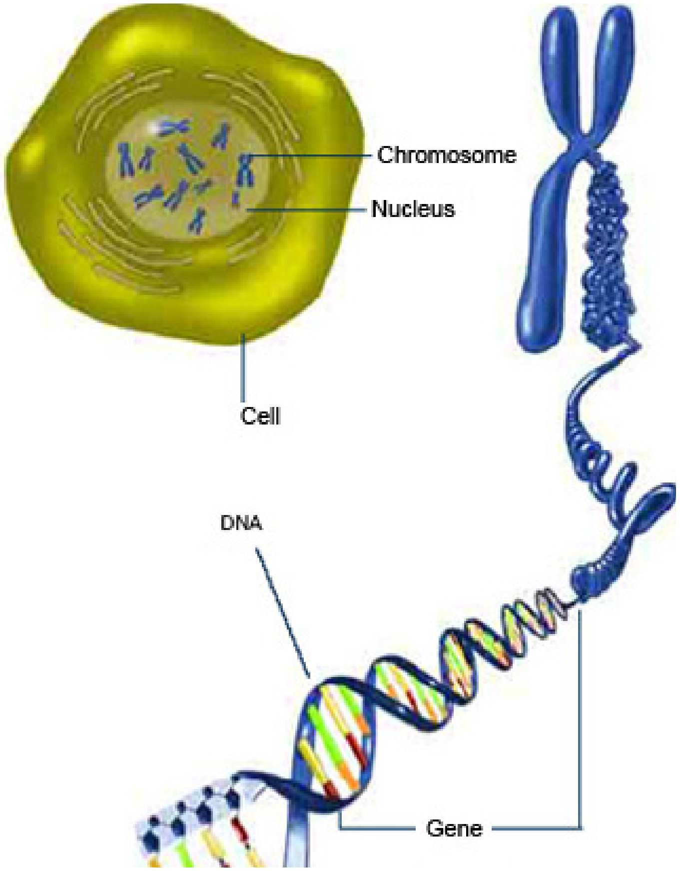


Mendel collected data from the F_2 generation for 7 pea characters, each of which had 2 traits (versions)

Character						F_2 Generation	
	Dominant Form	×	Recessive Form			Dominant:Recessive	Ratio
	Purple flowers	×	White flowers			705:224	3.15:1
	Yellow seeds	×	Green seeds			6022:2001	3.01:1
	Round seeds	×	Wrinkled seeds			5474:1850	2.96:1
	Green pods	×	Yellow pods			428:152	2.82:1
	Inflated pods	×	Constricted pods			882:299	2.95:1
	Axial flowers	×	Terminal flowers			651:207	3.14:1
	Tall plants	×	Dwarf plants			787:277	2.84:1

Hypotheses on Heredity

- In the 1800s it was thought that parental traits blended together in offspring.
  - In the 'Blending' hypothesis,' genetic material contributed by 2 parents mixes like two colors of paint.
    - Problem: Over time, populations should all end up with uniform traits.
- Mendel's experiments showed that this did not occur.
  - Mendel's research gave support to a 'particulate' hypothesis of inheritance.
    - Though Mendel never knew about DNA, today we know that parents pass on discrete heritable units (genes stored in DNA) that retain their separate identities in offspring in an undiluted form.



Genes are sections of DNA in Prokaryotes & Eurkaryotes *(genes can be found stored in RNA in some viruses)* **which contain the information for making RNAs & Proteins.**

DIFFERENCE BETWEEN GENE AND DNA

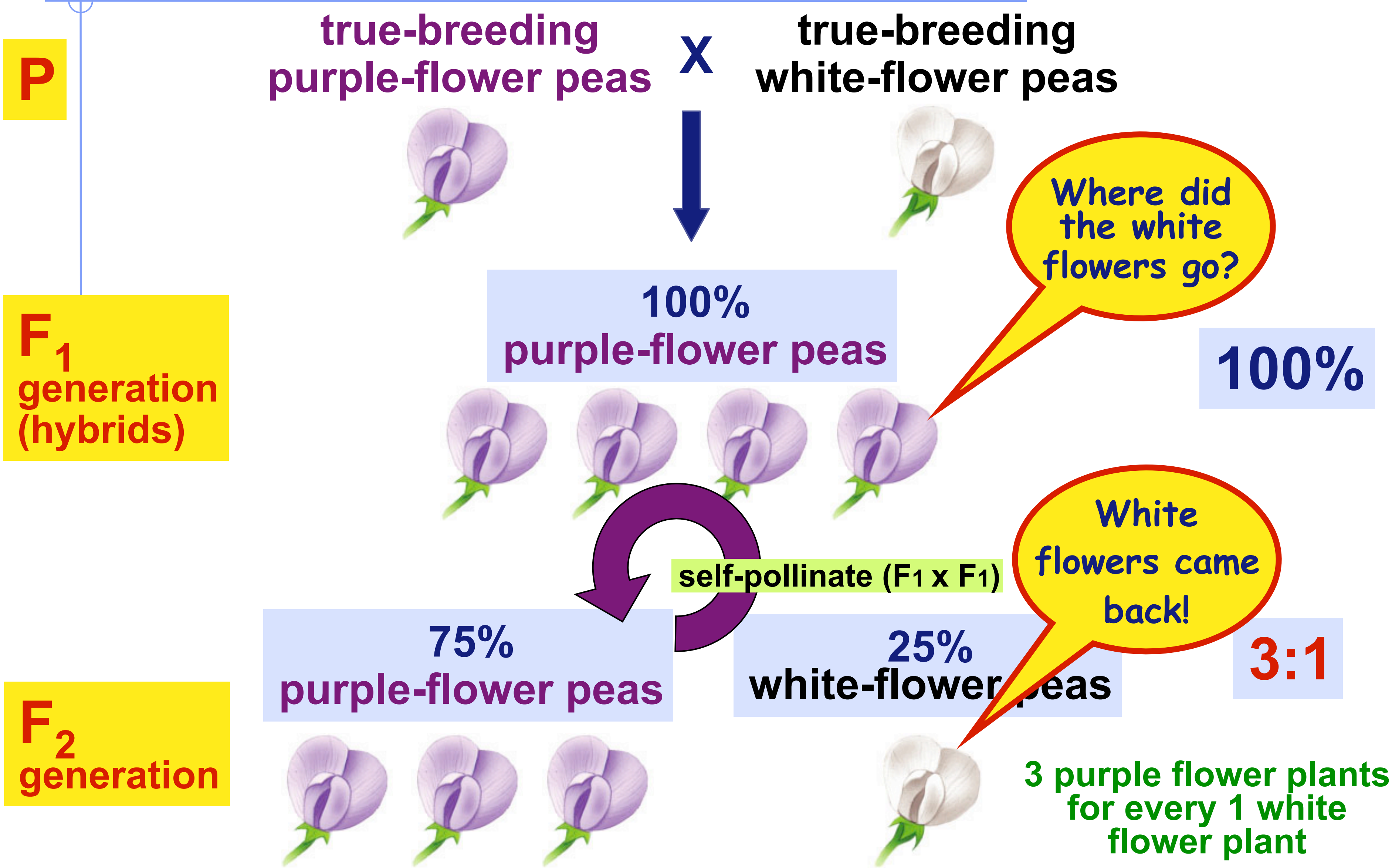
GENE

A GENE IS A SEQUENCE OF DNA OR RNA THAT CODES FOR A MOLECULE THAT HAS A FUNCTION. THE TRANSMISSION OF GENES TO AN ORGANISM'S OFFSPRING IS THE BASIS OF THE INHERITANCE OF PHENOTYPIC TRAITS.

DNA

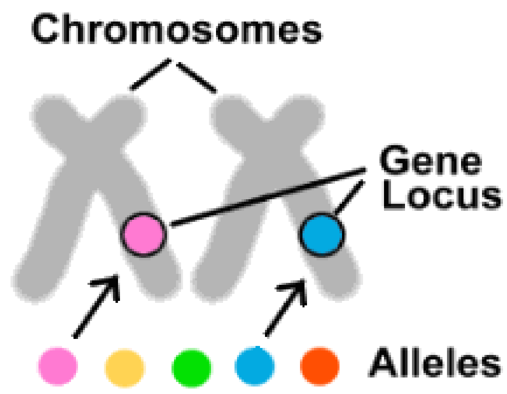
DEOXYRIBONUCLEIC ACID IS A MOLECULE COMPOSED OF TWO CHAINS WHICH COIL AROUND EACH OTHER TO FORM A DOUBLE HELIX, CARRYING THE GENETIC INSTRUCTIONS USED IN THE GROWTH, DEVELOPMENT, FUNCTIONING OF ALL KNOWN LIVING ORGANISMS.

Looking closer at Mendel's work



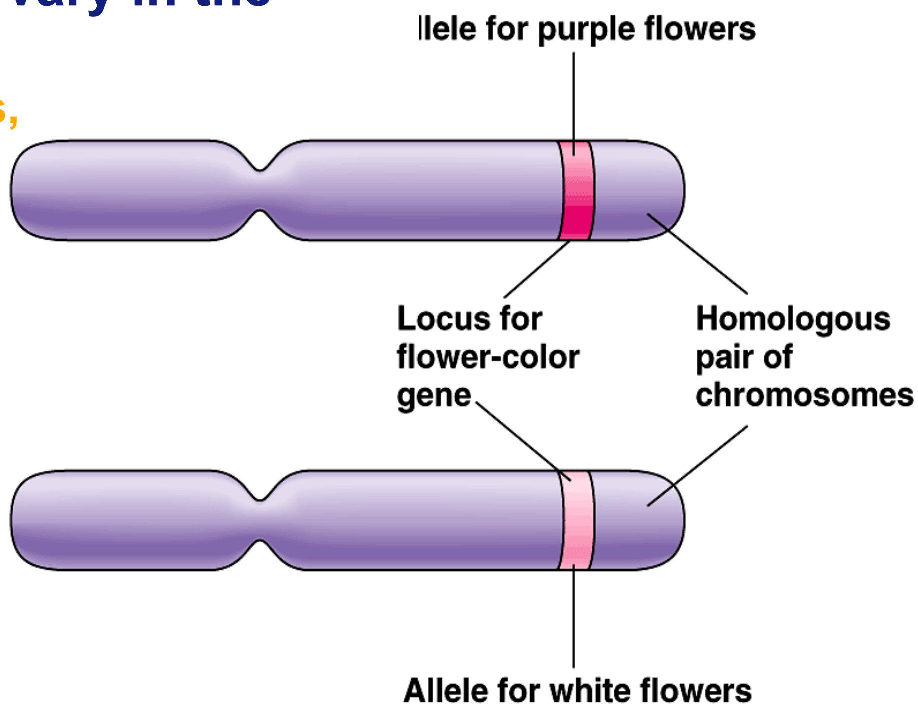
What did Mendel's findings mean?

- Traits come in alternative versions which account for the variations in inherited characters
  - purple vs. white trait for the flower color character
- The location of a gene on a chromosome = locus
- Alternative versions of a gene: alleles
  - There can be many alleles of a gene, but each diploid organism carries either 2 of the same type of allele or 2 different types of alleles
  - Different alleles of a type of gene vary in the sequence of nucleotides
    - Differ in the sequence of As, Ts, Cs, Gs along the DNA double helix of that gene on a chromosome



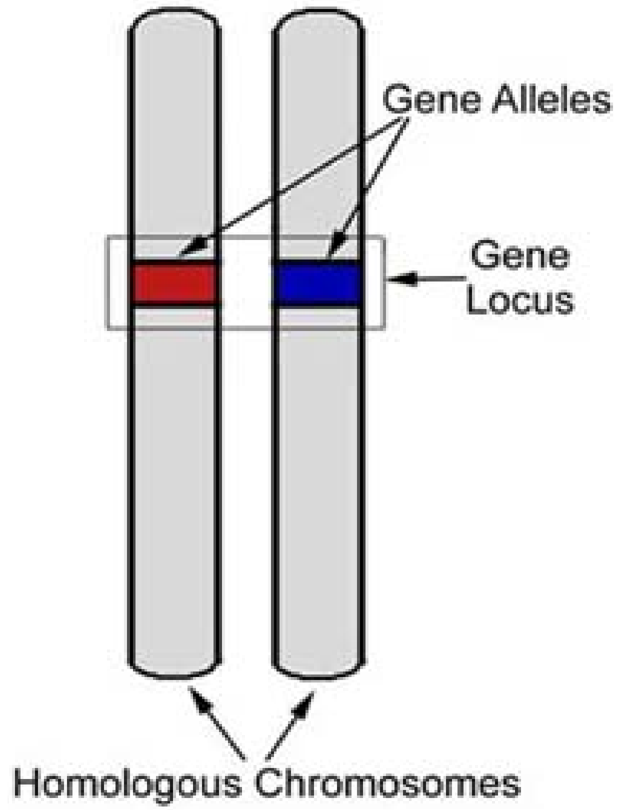
the purple-flower allele & white-flower allele are two DNA variations of a gene found at the flower-color locus

different versions of a gene (different alleles) may be found at same location on two homologous chromosomes



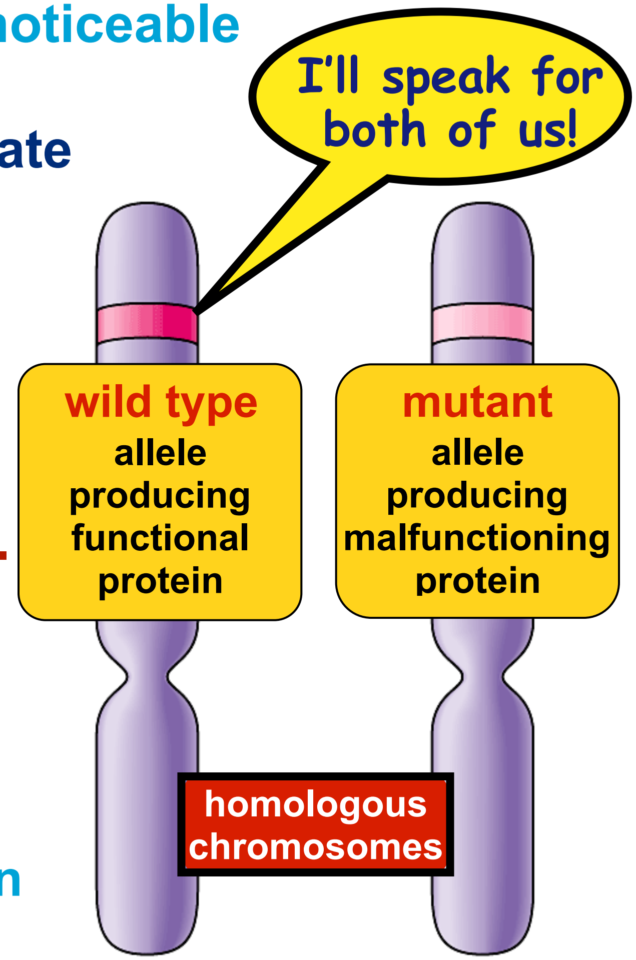
Traits are inherited as discrete units

- **For almost all characteristics, a diploid organism inherits 2 alleles, 1 from each parent**
  - **Humans are diploid organisms**
    - **Inherit 2 sets of chromosomes, 1 from each parent**
      - **Humans have two copies of every gene**
    - **A genetic locus with an allele is represented twice in a diploid cell, once per homolog** ***(the only exception being XY humans, the Y-type DNA molecule missing many of the genes found on the X-type)***
      - **like having 2 editions of encyclopedia**
        - **Encyclopedia Britannica**
        - **Encyclopedia Americana**



What did Mendel's findings mean?

- **Some traits mask other traits**
  - If 2 alleles at a locus differ, then the <u>dominant allele</u> is the one which determines the organism's appearance and the <u>recessive allele</u> is the one which has no noticeable effect on appearance.
    - Purple & white flower colors are separate traits that do <u>NOT</u> blend in organisms
      - Purple x White ≠ light purple flowers
    - The purple allele's product <u>MASKED</u> the white allele's product
- **When one type of gene is turned "<u>ON</u>," all copies** *(2 in diploids)* **of that gene turn on.**
  - <u>dominant allele</u>
    - Codes for <u>functional</u> protein
    - Masks the other allele's protein effects
  - <u>recessive allele</u>
    - Either does <u>not</u> make a functional protein or codes for a <u>malfunctioning</u> protein



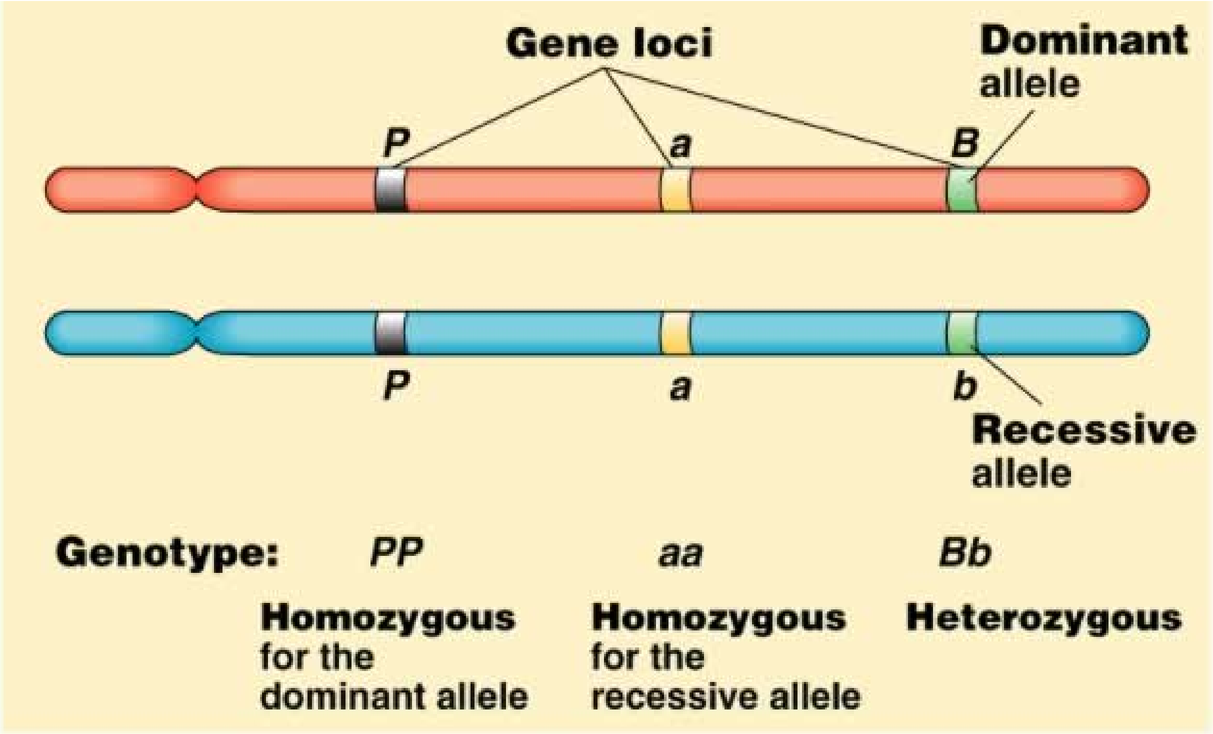
Useful Genetic Vocabulary

Homozygote: Organism that has a pair of identical alleles for a character in question

Heterozygote: Organism that has two different alleles for a gene in question

Genotype: An organisms genetic makeup

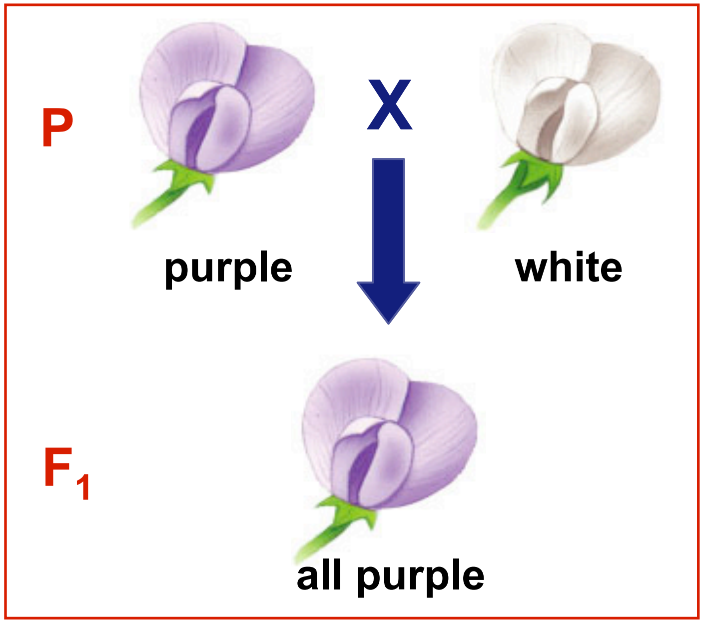
- ✓ Capital letter often designates dominant allele
- ✓ Lower case letter often used to designate recessive allele



Genotype vs. phenotype

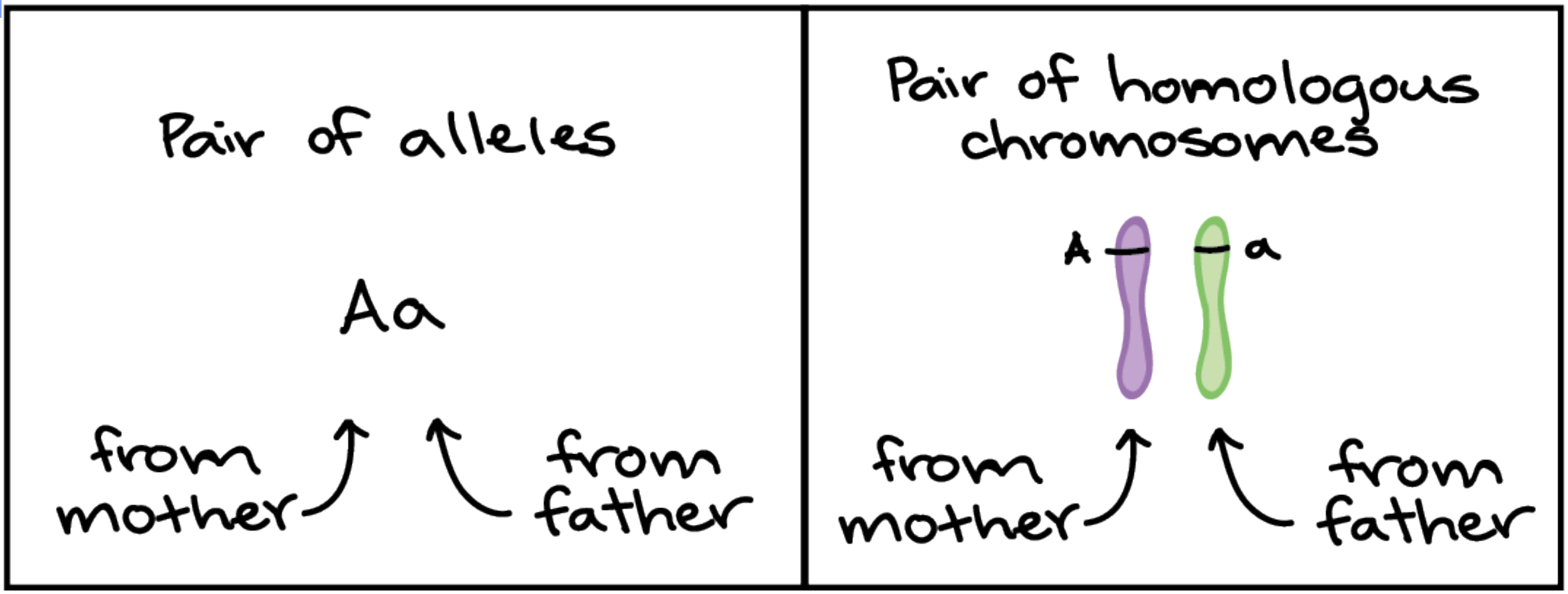
- There exists a difference between how an organism "looks" & its genetics *(the version of DNA it possesses)*
  - In Mendel's crosses, the P generation purple plant differed genetically from the F1 generation purple plant
  - phenotype
    - description of an organism's trait
    - the "physical" effect of gene when on
  - genotype
    - description of an organism's genetic makeup
    - which alleles for genes an organism has in its DNA

Explain Mendel's results using
…dominant & recessive
…phenotype & genotype



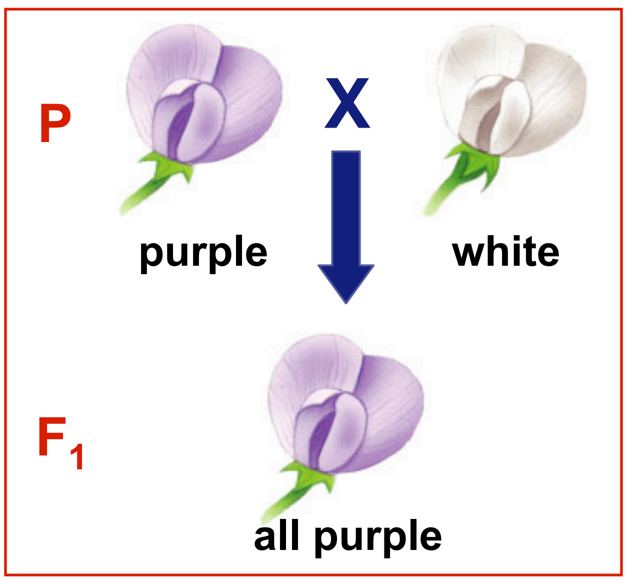
Gametes contain 1 set of chromosomes compared to somatic cells which contain 2 sets

- Chromosomes carry genes and come in matched (homologous) pairs in an organism
- One member of the chromosome pair comes from the mother and one from the father.
  - The members of a homologous pair SEPARATE in meiosis, so each sperm or egg (gamete) receives just one homolog and, therefore, just one copy of each gene on that homolog.



Genotype of Mendel's first cross

- **Can represent alleles as letters**
  - **Flower color alleles → P or *p***
  - **What is the genotype of true-breeding purple-flower peas → PP**
  - **What is the genotype of true-breeding white-flower peas → pp**



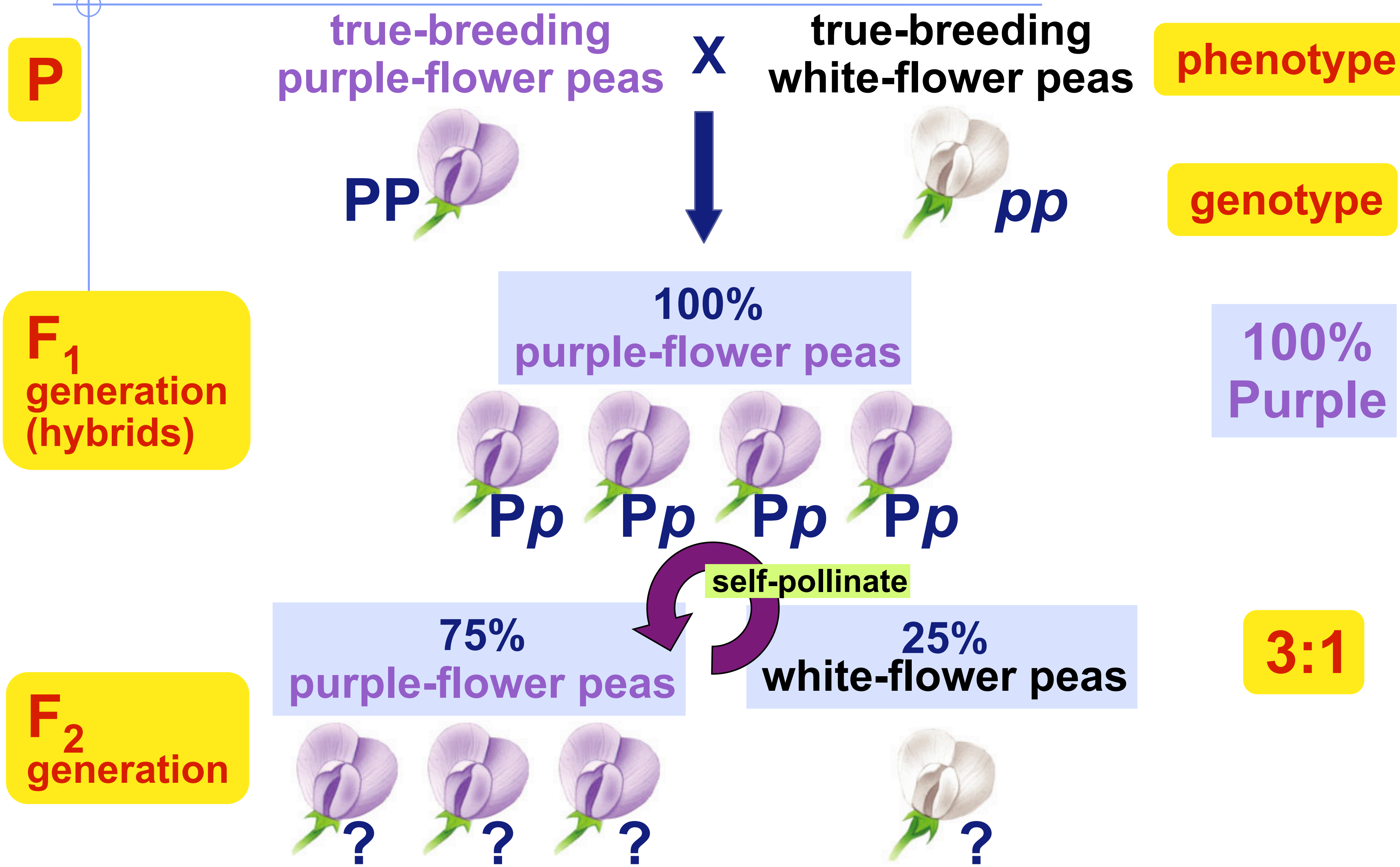
PP x *pp*

↓

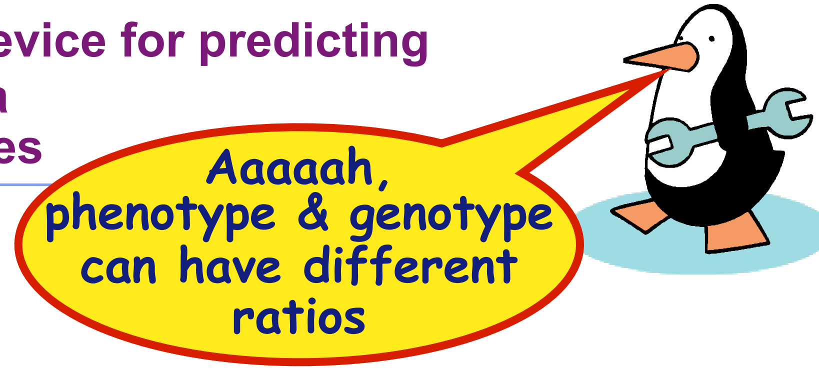
P*p*

F1 purple flower is a hybrid

Looking closer at Mendel's work



Punnett squares - diagrammatic device for predicting the allele composition of offspring from a cross of individuals with known genotypes



F_1 generation (hybrids)

P*p* x P*p*

Male gametes / sperm

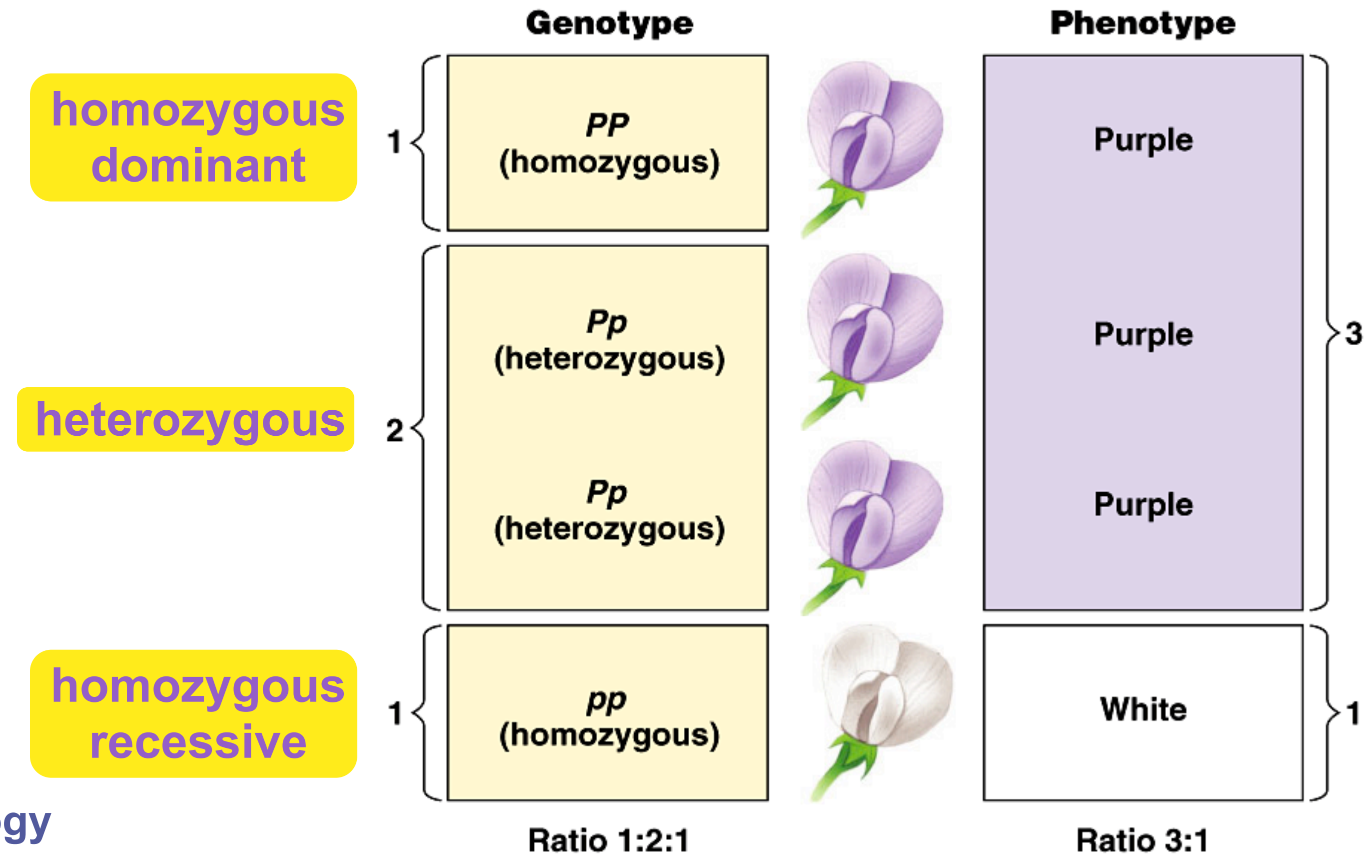
Female gametes / eggs

	P	*p*
P	PP	P*p*
p	P*p*	*pp*

	% genotype	% phenotype
PP	25%	75%
P*p*	50%	
P*p*		
pp	25%	25%
	1:2:1	3:1

Genotypes

- **Homozygous** = *same* alleles = PP & *pp*
- **Heterozygous** = *different* alleles = P*p*



Phenotype vs. genotype

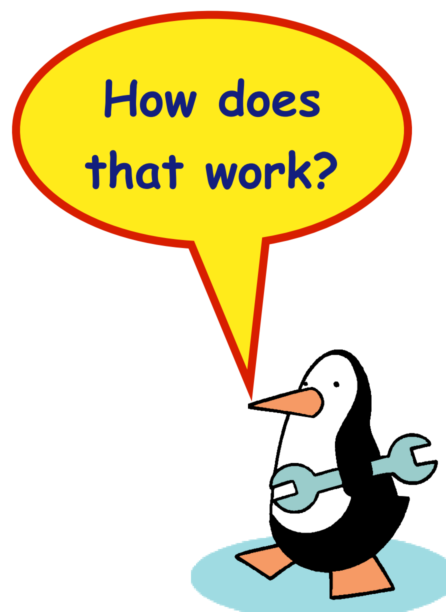
- 2 organisms can have the same phenotype but have different genotypes

	purple	PP	homozygous dominant
	purple	Pp	heterozygous

How do you determine the genotype of an individual with with a dominant phenotype?

Test cross

- Breed the dominant phenotype — "the unknown genotype" — with a <u>homozygous recessive</u> (*pp*) to determine the identity of the unknown allele
  - The genotype of a <u>homozygous recessive</u> is always known!

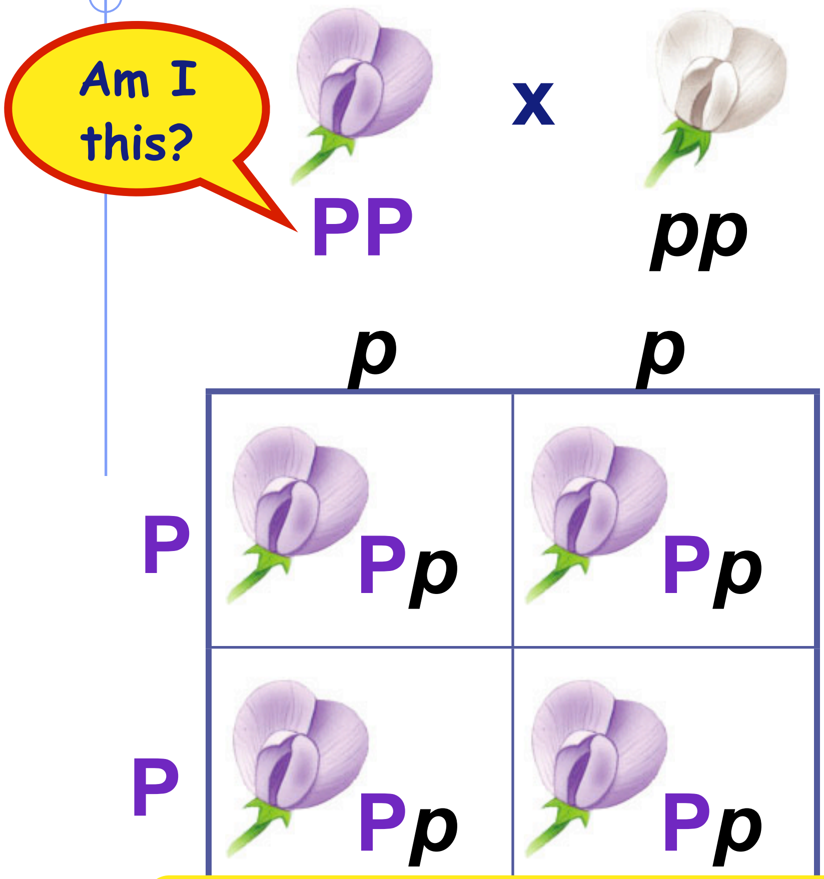


x

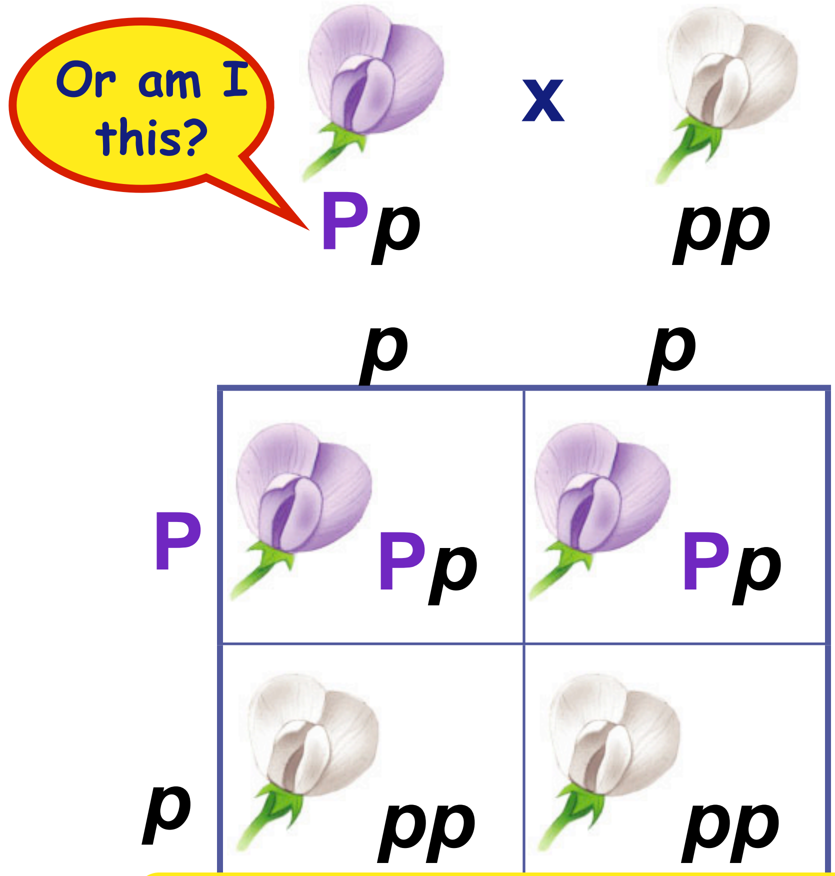
Is this genotype PP or P*p*?

This genotype is definitely *pp*

How does a Test cross work?



If homozygous dominant, expect 100% purple

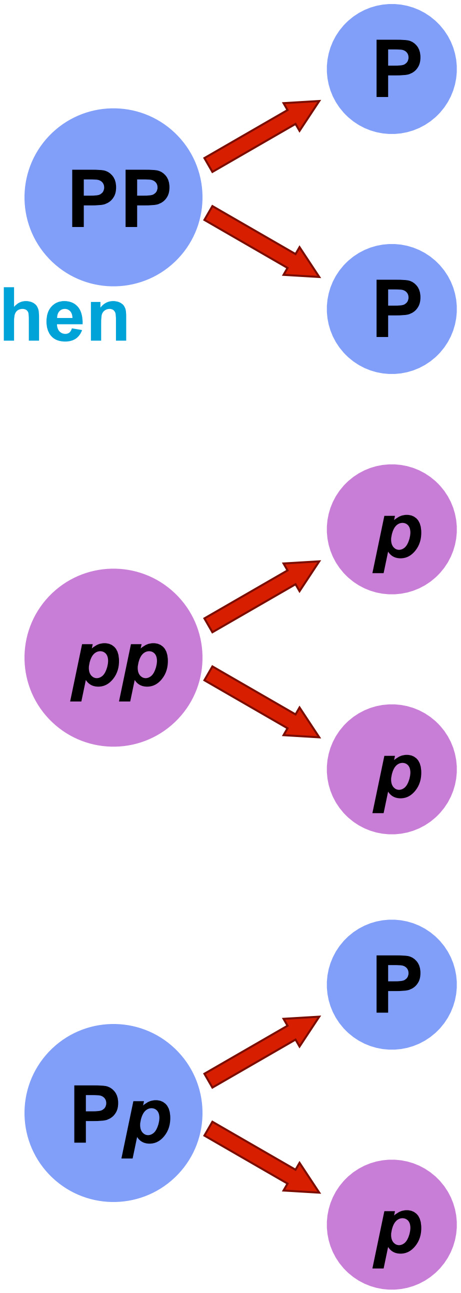


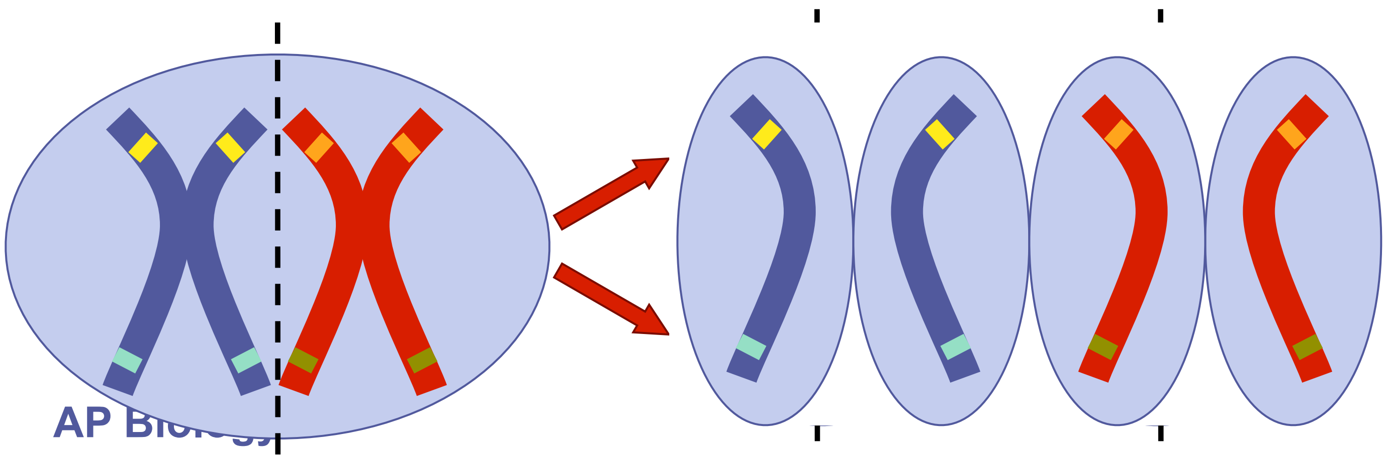
If heterzygous, expect 50% purple:50% white or 1:1

By comparing what you get with what is expected, you can determine which genotype the parent flower started off with.

Mendel's 1st law of heredity

- Law of segregation
  - "traits for a character segregate when gametes are made"
  - during meiosis, alleles segregate
    - homologous chromosomes separate
  - each allele for a trait is packaged into a separate gamete



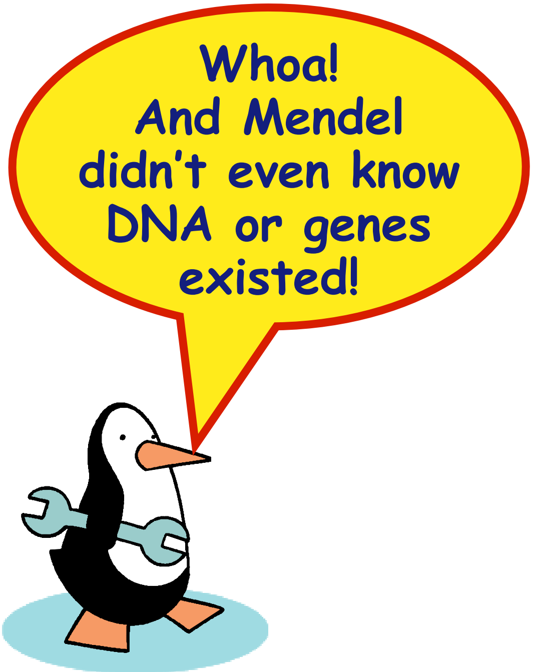


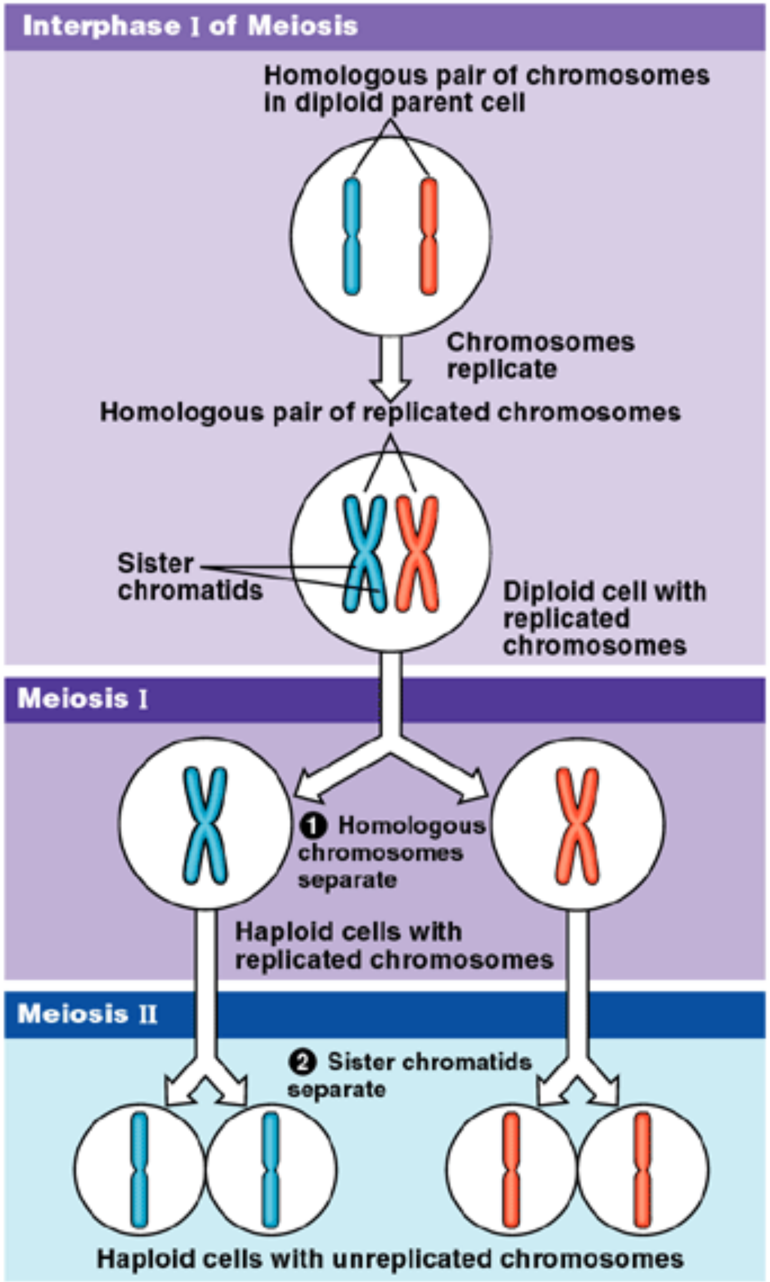
Law of Segregation

- Which stage of meiosis creates the law of segregation?

Metaphase 1

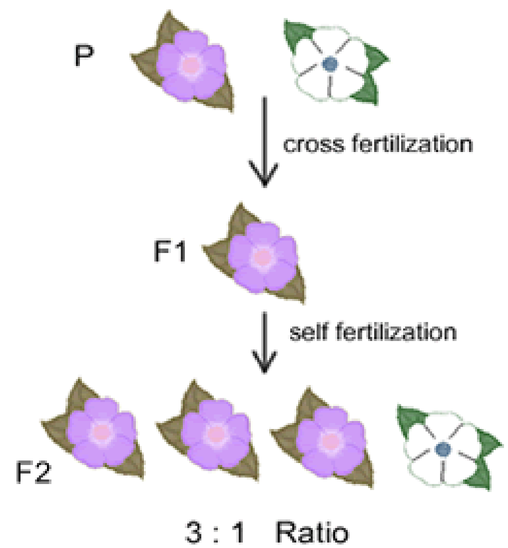
Anaphase 1





Monohybrid crosses

- Some of Mendel's experiments followed the inheritance of <u>single characters</u>
  - Ex: ONLY flower color or ONLY seed color
    - Such crossed are called <u>monohybrid crosses</u>
    - F_1 progeny heterozygous for a character are called <u>monohybrids</u>
      - Ex: Genotype *Pp*

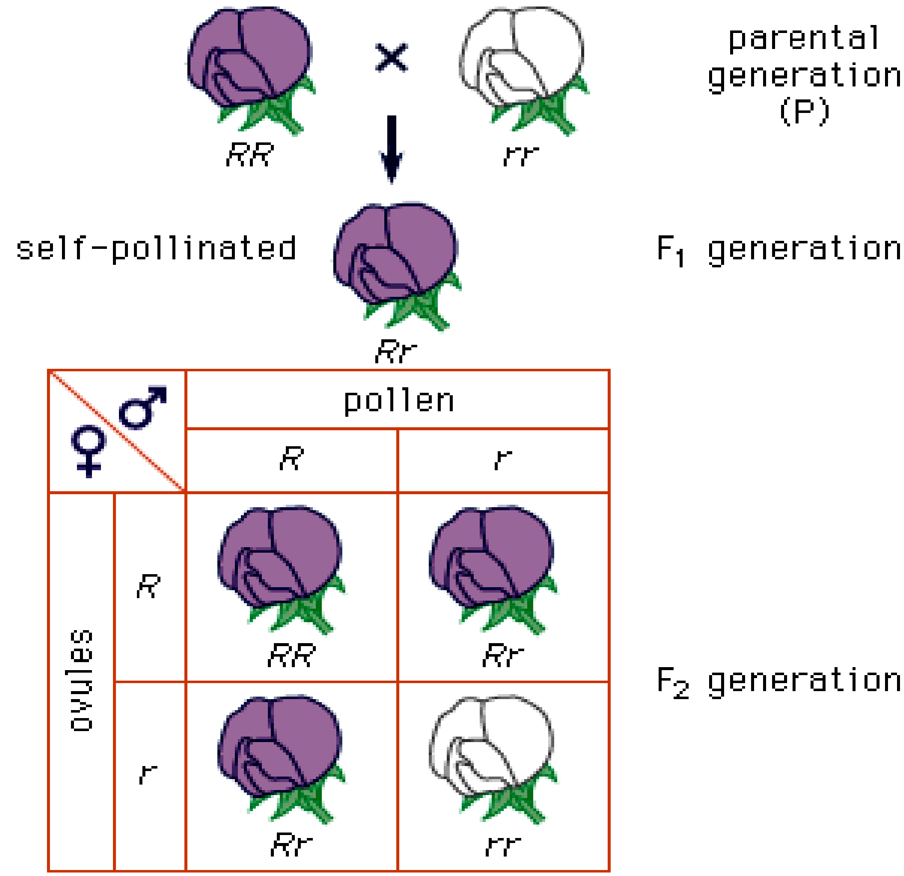


With monohybrid crosses we saw:

Mendel's 1st Law of Heredity

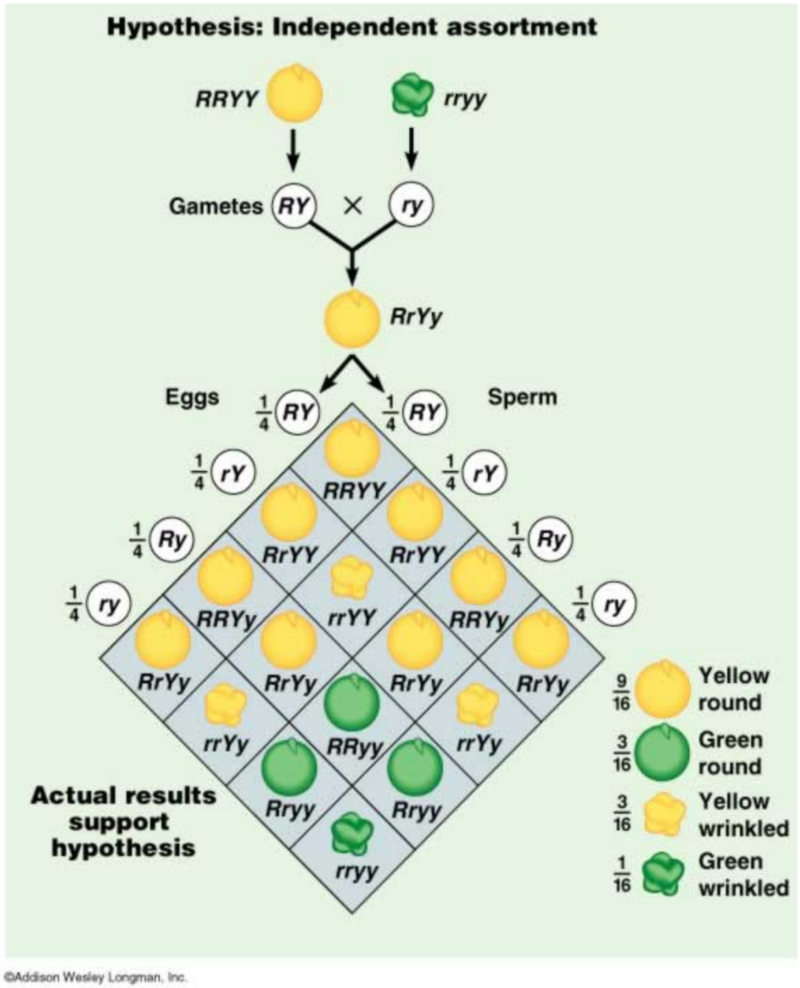
Law of Segregation:

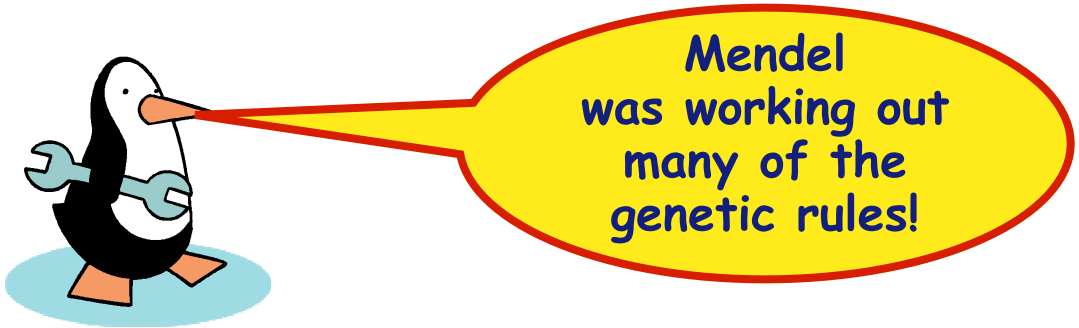
Two alleles for the same heritable character segregate (separate) during gamete formation and end up in different gametes



Dihybrid crosses

- Other of Mendel's experiments followed the inheritance of <u>2 different characters</u> <u>at ONCE</u>
  - Ex: Seed color <u>and</u> seed shape
    - These crosses are termed <u>dihybrid</u> crosses
    - Individuals heterozygous for two characters are called <u>dihybrids</u>
      - <u>Ex: Genotype *YyRr*</u>





Dihybrid cross

P

true-breeding yellow, round peas — **YYRR**

Y = yellow
R = round

x

true-breeding green, wrinkled peas — **yyrr**

y = green
r = wrinkled

F_1 generation (hybrids)

yellow, round peas — **YyRr**

100%

self-pollinate

F_2 generation

9/16 yellow round peas

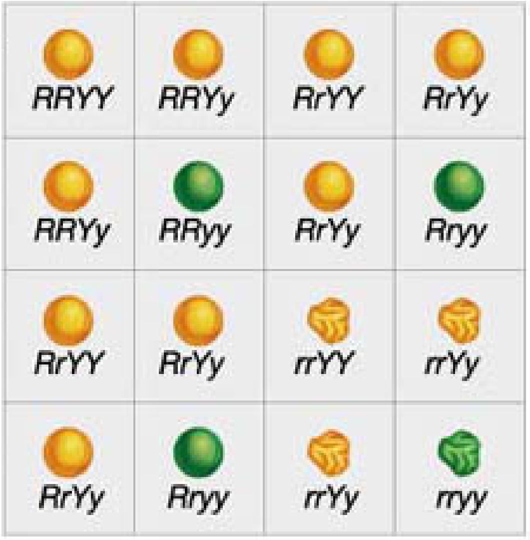
3/16 green round peas

3/16 yellow wrinkled peas

1/16 green wrinkled peas

Phenotypic ratio
9:3:3:1

FYI



Dominant alleles do NOT interact physically with a recessive allele in a heterozygote, subduing the recessive allele.

Alleles on homologous chromosomes are physically independent of one another.

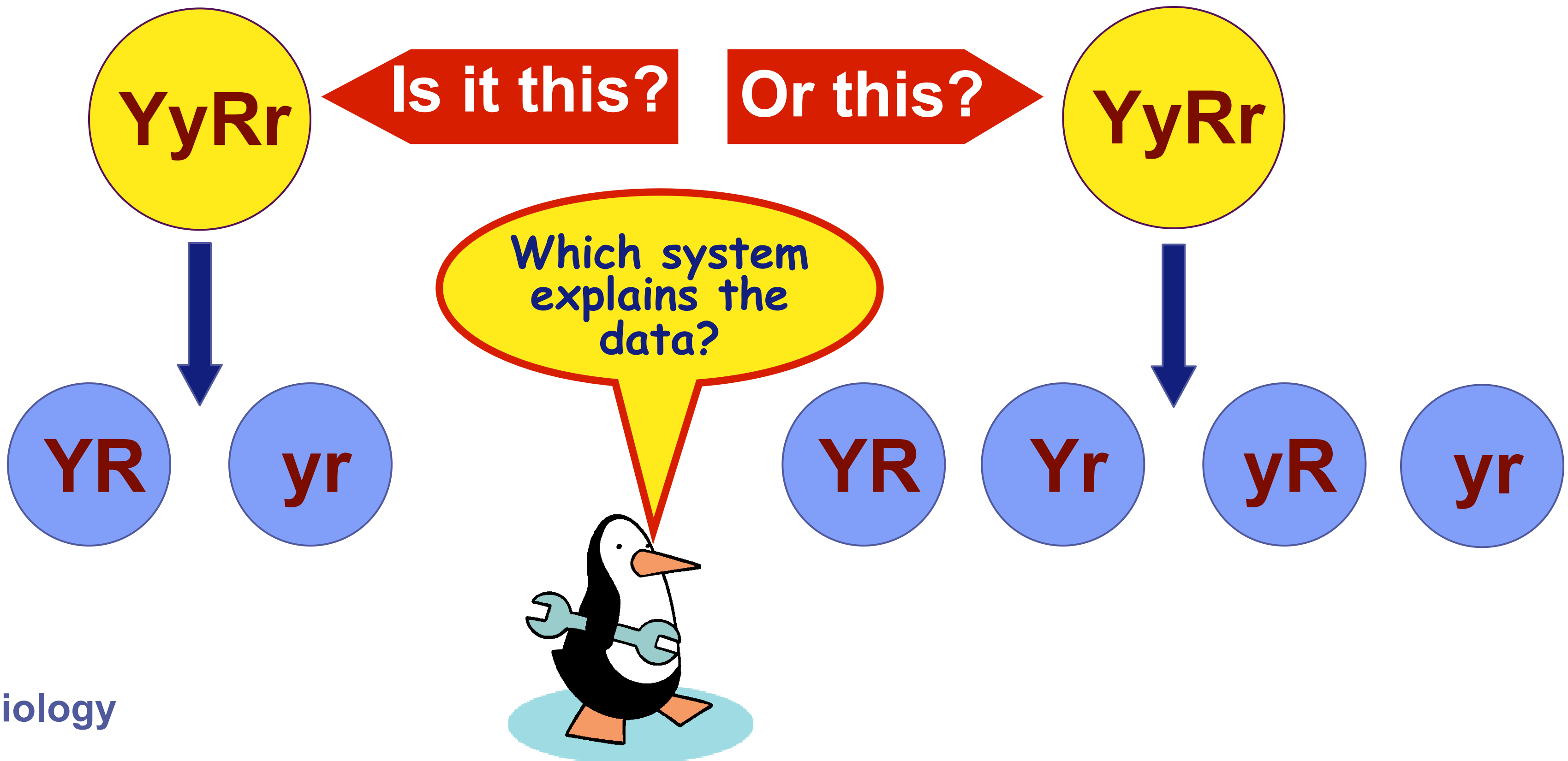
In this F1 generation the reason why Yellow is considered the dominant allele is because it causes the phenotype we see in the heterozygote.

Wrinkled seeds in pea plants with two copies of the recessive allele are due to the accumulation of monosaccharides in the seed because the allele codes for a defective enzyme that would convert the monosaccharides into polysaccharide starch. With high solute concentration, excess water enters the seed and when they are dried, they wrinkle up.

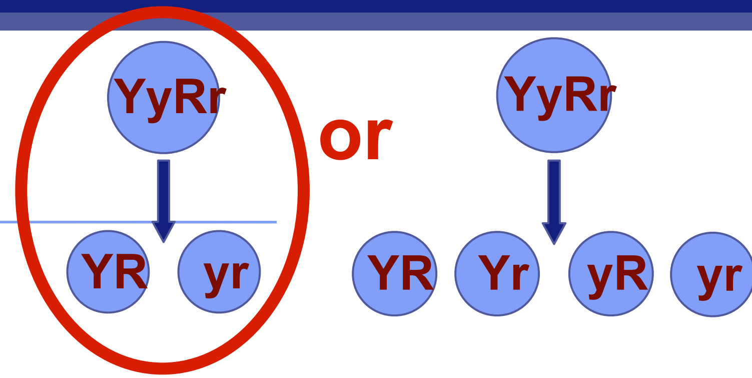
Both homozygous dominants and heterozygotes produce enough enzyme to convert the monosaccharides into starch so water does not diffuse inward by osmosis. When these seeds dry they remain smooth.

What's going on here?

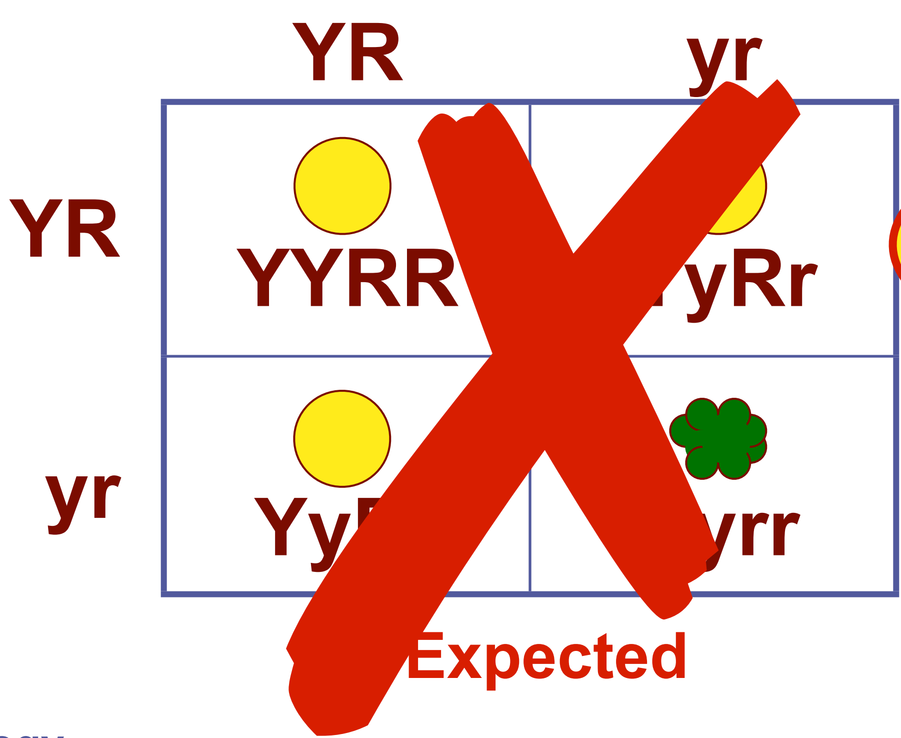
- If <u>different genes</u> are on different chromosomes…
  - How do they assort in the gametes?
    - <u>Together</u> or <u>independently</u>?

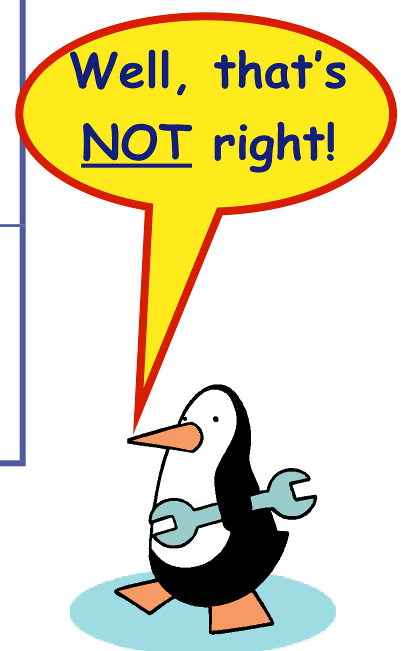


Is this the way it works?



YyRr x YyRr

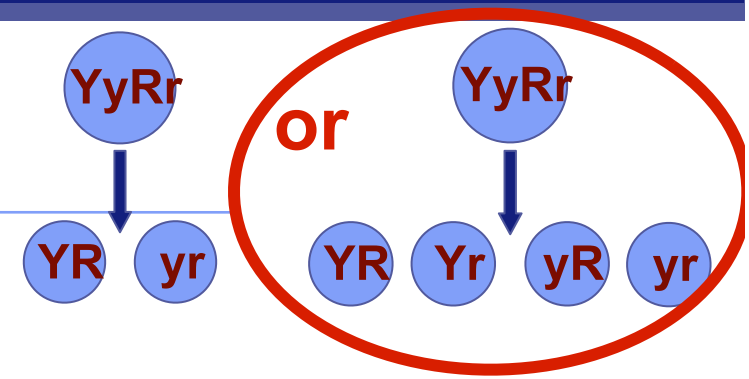




Observed

- 9/16 yellow round
- 3/16 green round
- 3/16 yellow wrinkled
- 1/16 green wrinkled

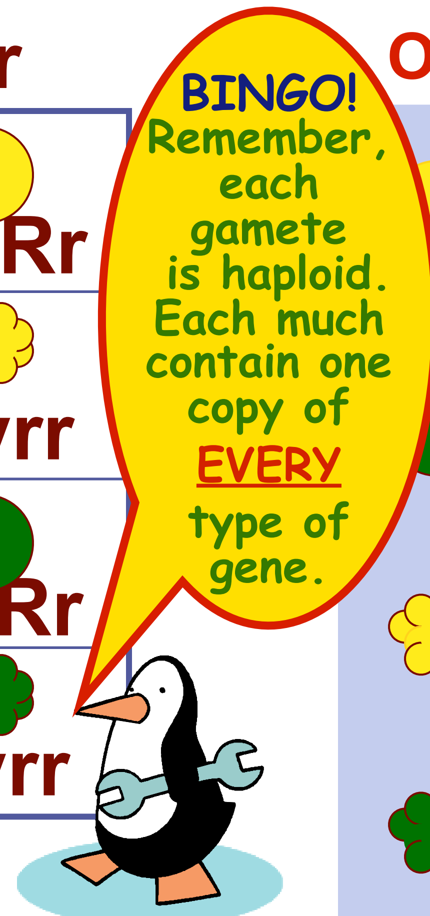
Dihybrid cross



YyRr x YyRr

	YR	Yr	yR	yr
YR	YYRR	YYRr	YyR	YyRr
Yr	YYRr	YYrr	yRr	Yyrr
yR	YyRR	Y	yyRR	yyRr
yr	YyRr	Yyrr	yyRr	yyrr

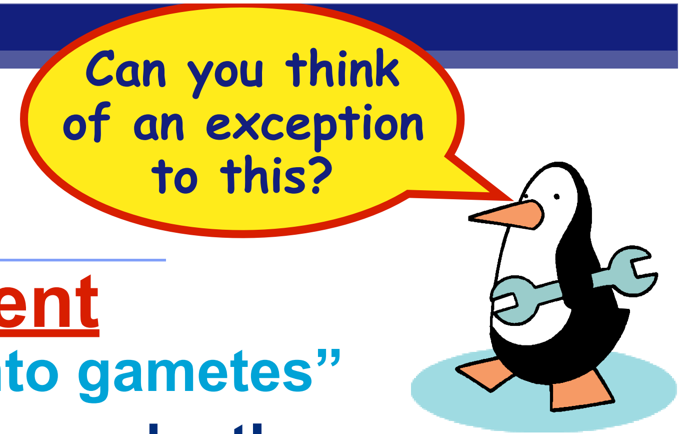
Expected



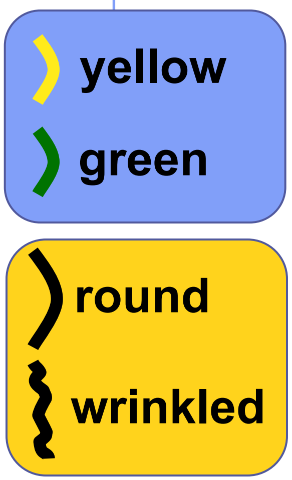
Observed

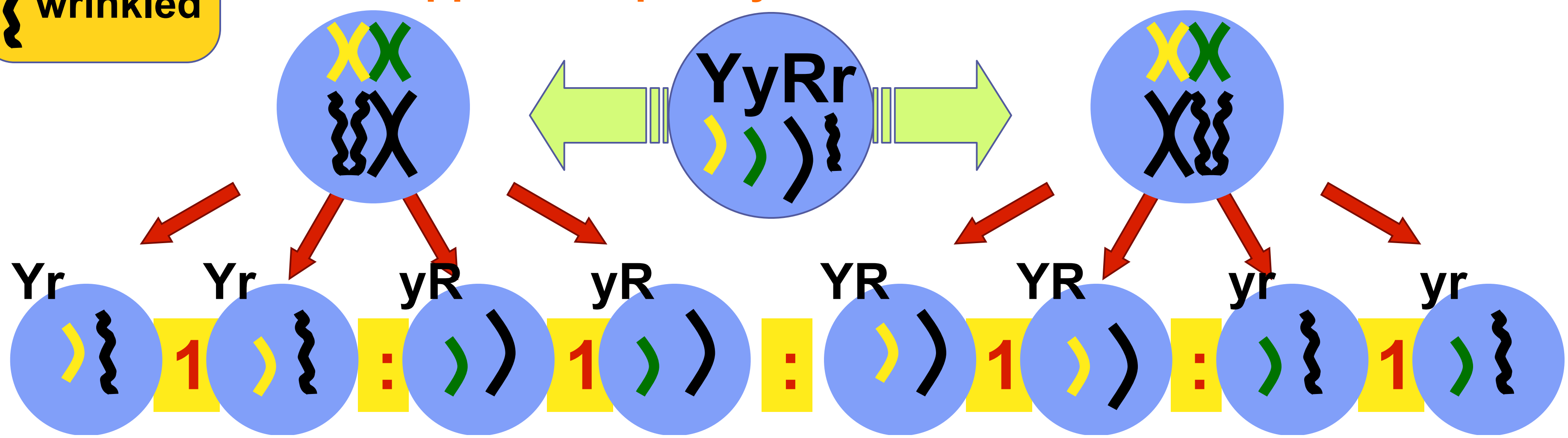
- 9/16 yellow round
- 3/16 green round
- 3/16 yellow wrinkled
- 1/16 green wrinkled

Mendel's 2nd law of heredity



- **Law of <u>independent assortment</u>**
  - "Characters sort independently into gametes"
  - Each pair of alleles segregate independently of each other pair of alleles during gamete formation
    - different genes separate into gametes <u>INDEPENDENTLY</u>
  - non-homologous chromosomes (different tetrads) align independently
  - gamete types produced in equal amounts YR = Yr = yR = yr
    - only true for genes on <u>separate chromosomes</u> or on same chromosome <u>AND</u> so <u>far apart that crossing over happens frequently</u>



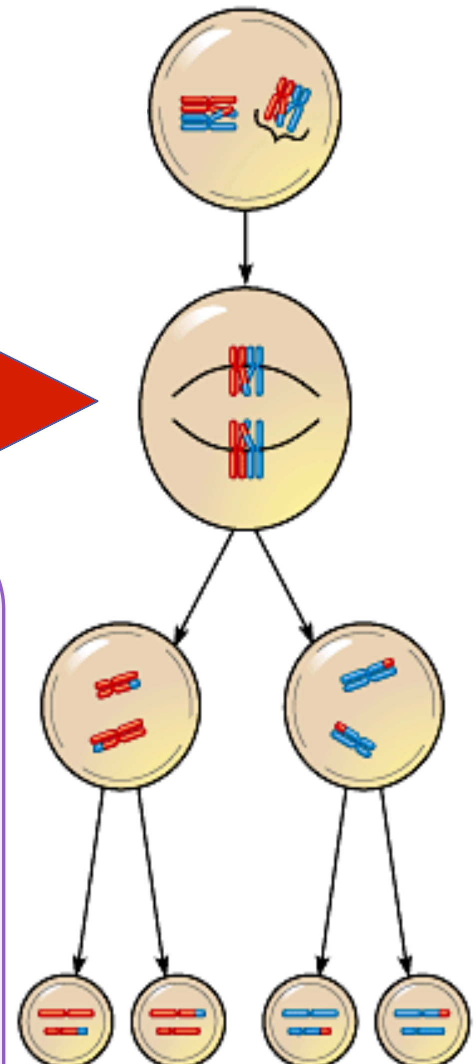


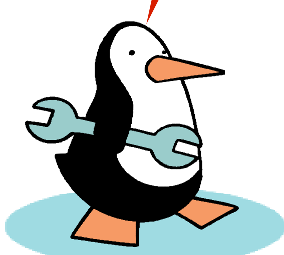
Law of Independent Assortment

- **Which stage of meiosis creates the law of independent assortment?**

Remember Mendel didn't even know DNA —or genes— existed!

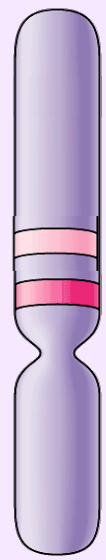
Metaphase 1





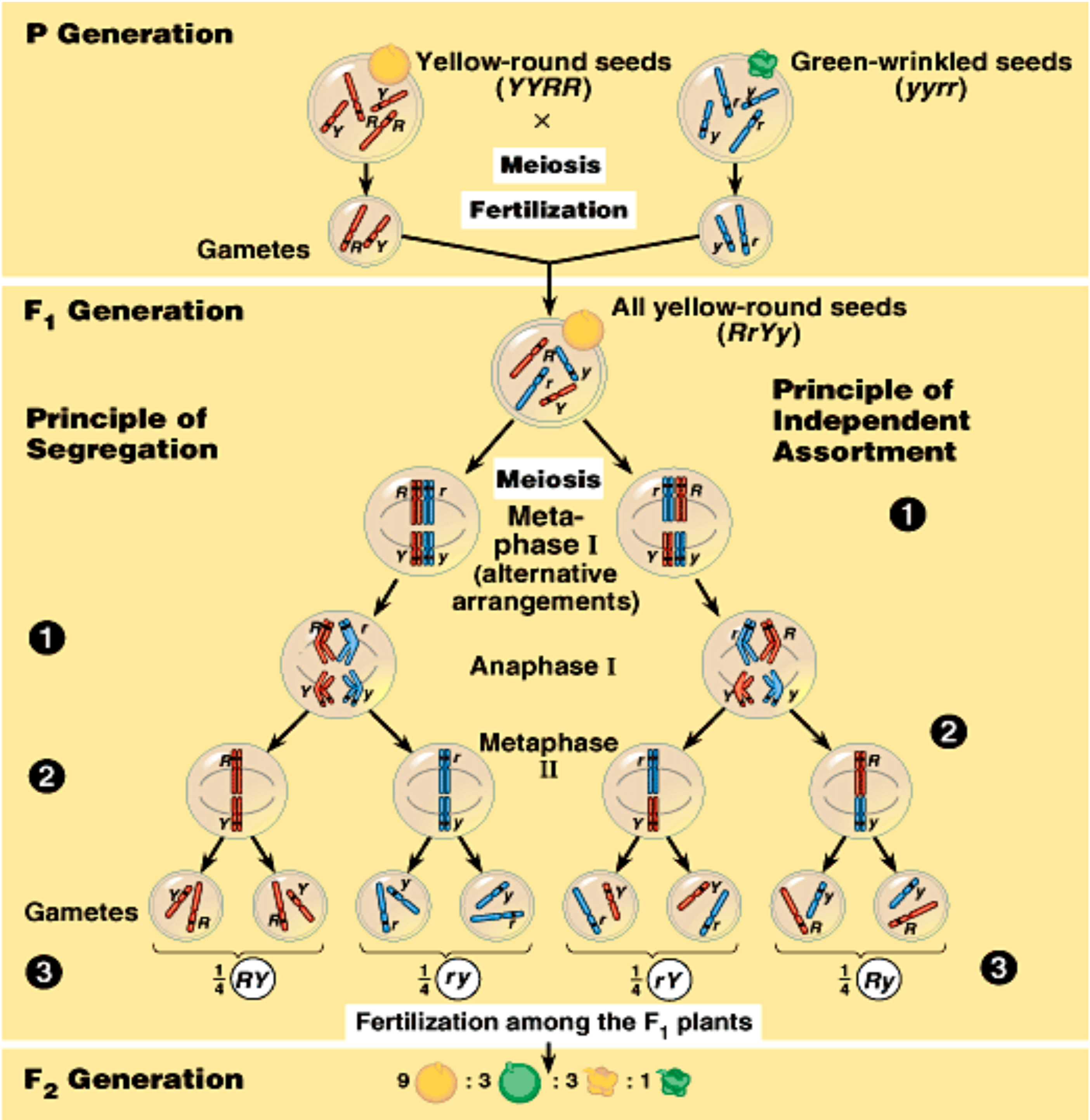
EXCEPTION:

- If genes are on same chromosome & close together
  - will usually be inherited together
  - rarely crossover separates them
  - Genes considered "linked"



The chromosomal basis of Mendel's laws…

Trace the genetic events through meiosis, gamete formation & fertilization to offspring



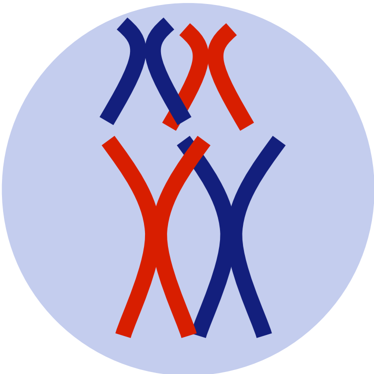
Review: Mendel's laws of heredity

- Law of segregation
  - highlighted in monohybrid cross
    - Follows a single trait (gene)
  - each allele segregates into separate gametes
    - established by Metaphase 1 and Anaphase 1
- Law of independent assortment
  - highlighted in dihybrid (or more) cross
    - Follows 2 or more characters (>1 genes)
  - Distant genes on separate chromosomes (or far apart on same chromosome)

    assort into gametes independently
    - established by Metaphase 1

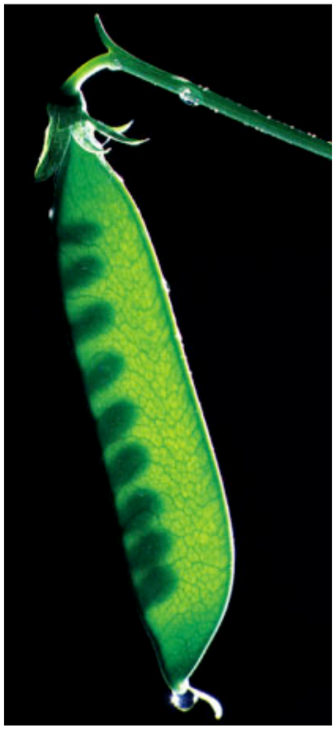
EXCEPTION
- linked genes

metaphase1



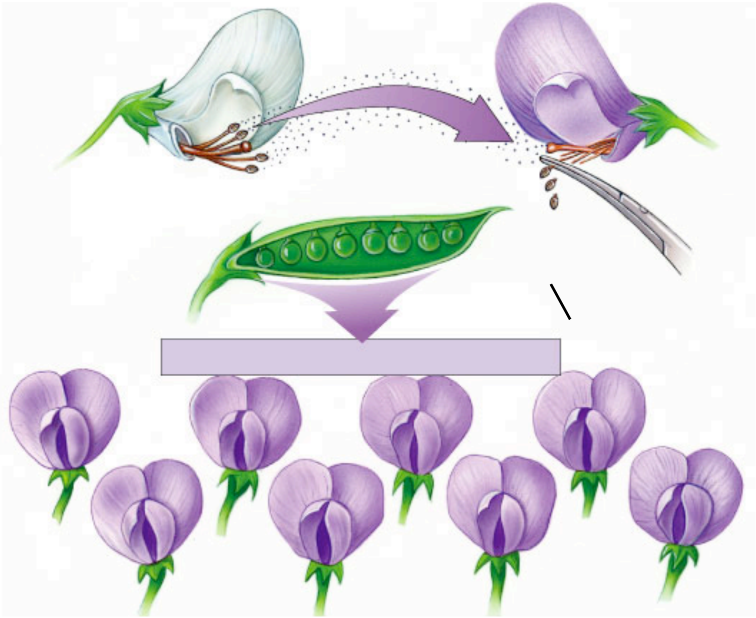
Mendel chose peas wisely

- Pea plants are good for genetic research
  - available in many varieties with distinct heritable features with different variations
    - flower color, seed color, seed shape, etc.
  - Mendel had strict control over which plants mated with which
    - each pea plant has male & female structures
    - pea plants can self-fertilize
    - Mendel could also cross-pollinate plants: moving pollen from one plant to another



Mendel chose peas luckily

- Pea plants are good for genetic research
  - relatively simple genetically
    - most characters are controlled by a single gene with each gene having only 2 alleles,
      - one allele was always completely dominant over the other



Gregor Mendel

Any Questions??