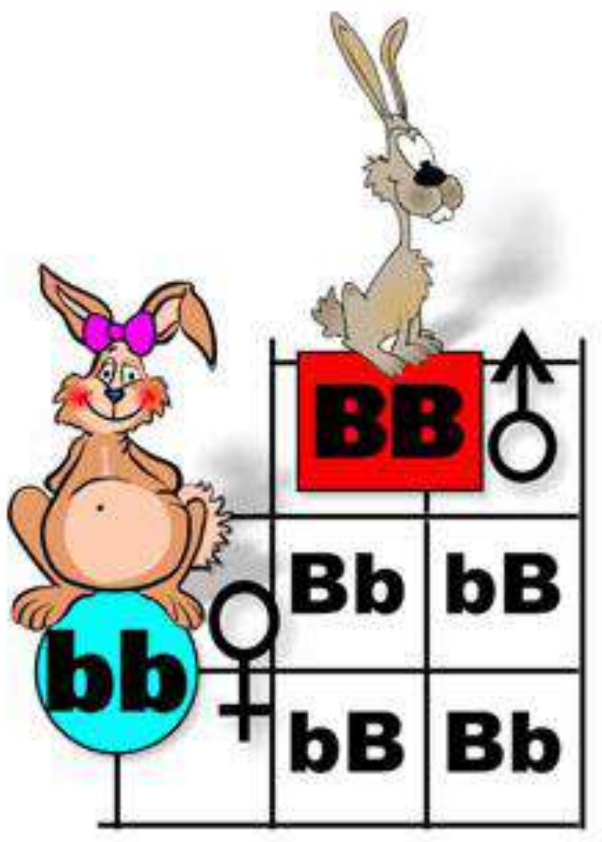
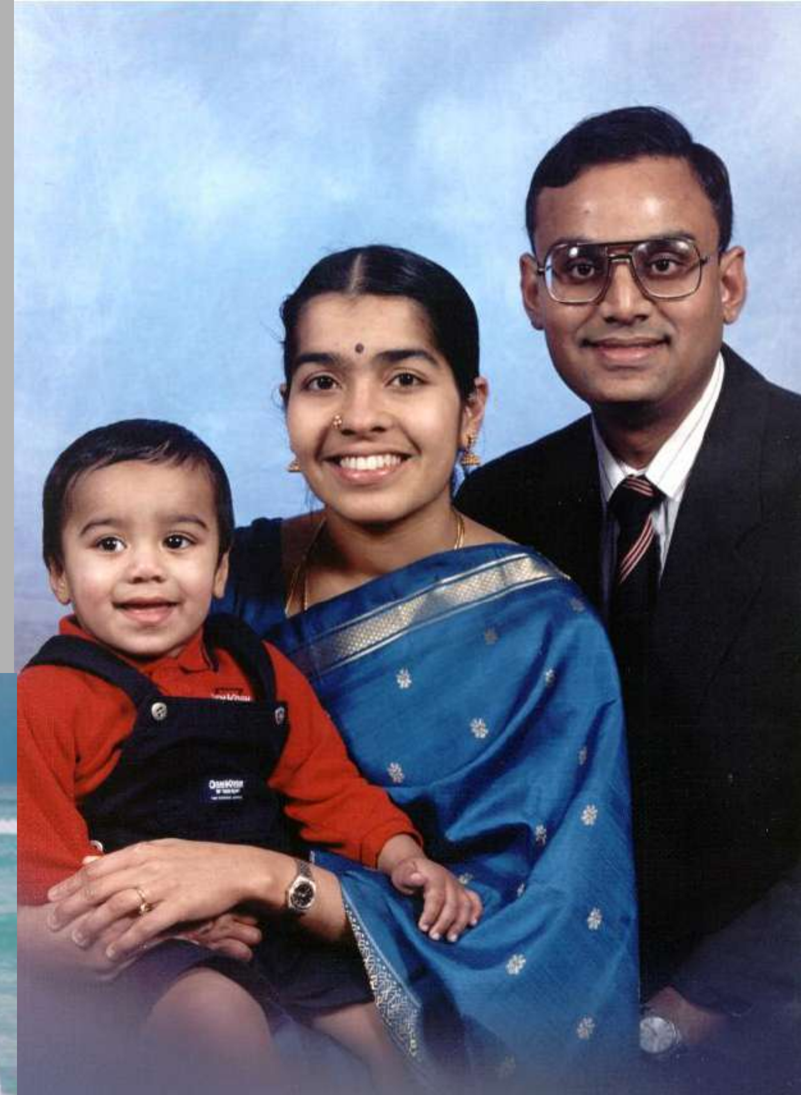
NOTES: Chapter 14, part 1 – Mendelian Genetics!!



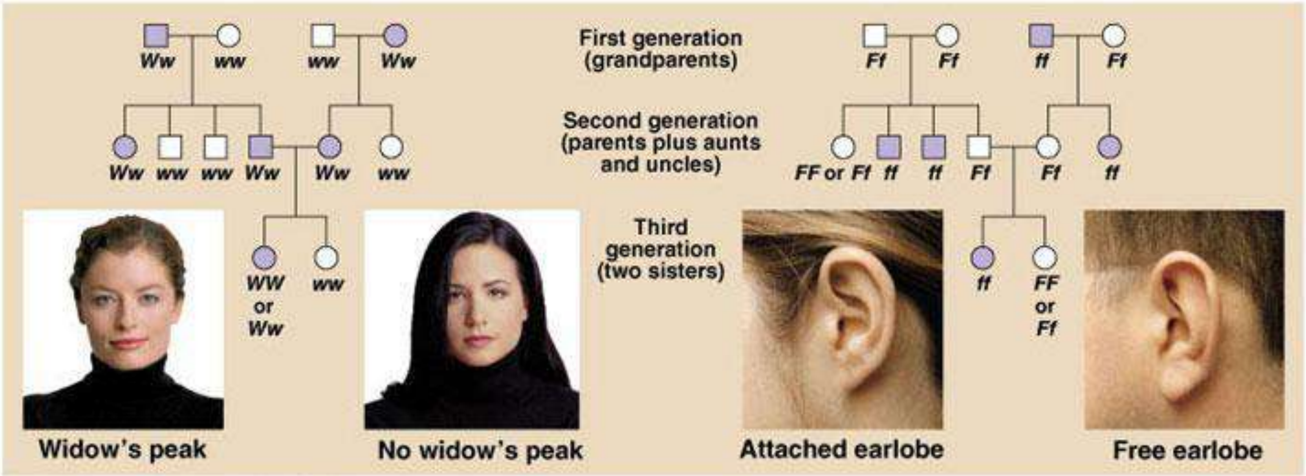
- One possible explanation of heredity is a **“blending” hypothesis:**
  – the idea that genetic material contributed by two parents mixes in a manner analogous to the way blue and yellow paints blend to make green

- An alternative to the blending model is the “particulate” hypothesis of inheritance (the gene idea):
  – parents pass on <u>discrete heritable units</u>, genes



How are traits inherited?

- Trait: <u>some aspect of an organism that can be described or measured</u>



- Gregor Mendel documented a particulate mechanism of inheritance through his experiments with garden peas

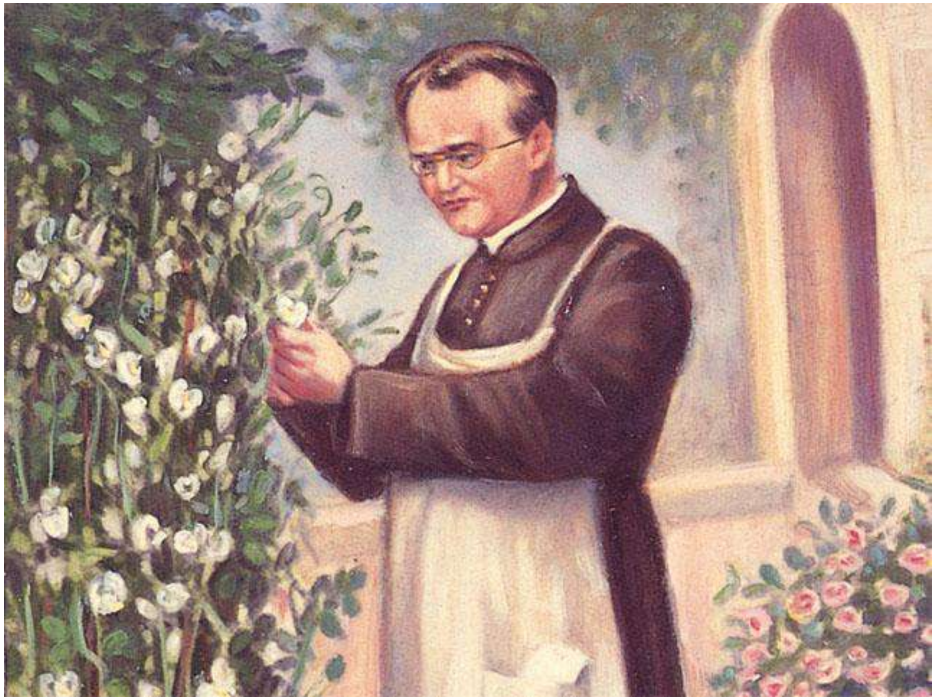
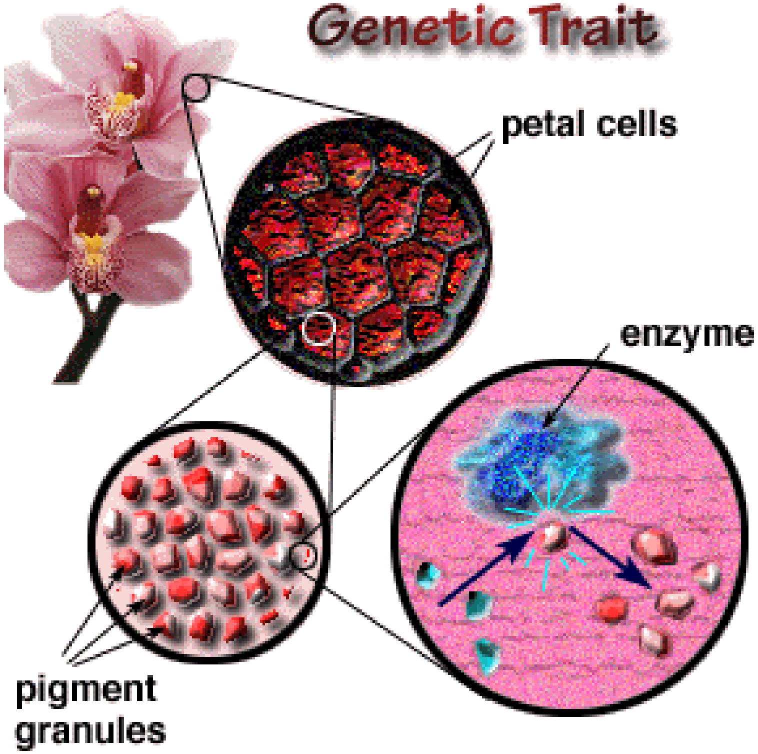


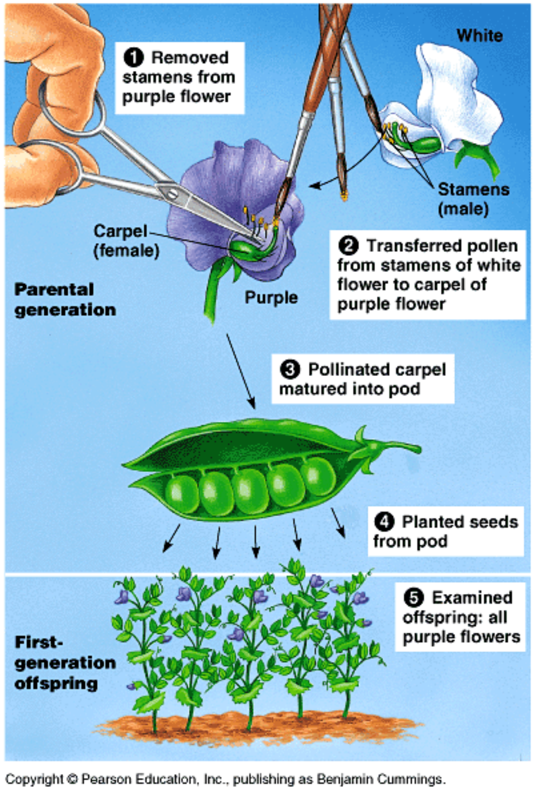
Figure 14.1

- Gregor Mendel, an Austrian monk, applied mathematics to his study of genetics. He chose to study the **garden pea plant** to investigate how traits were passed from generation to generation.



Mendel's Experimental, Quantitative Approach

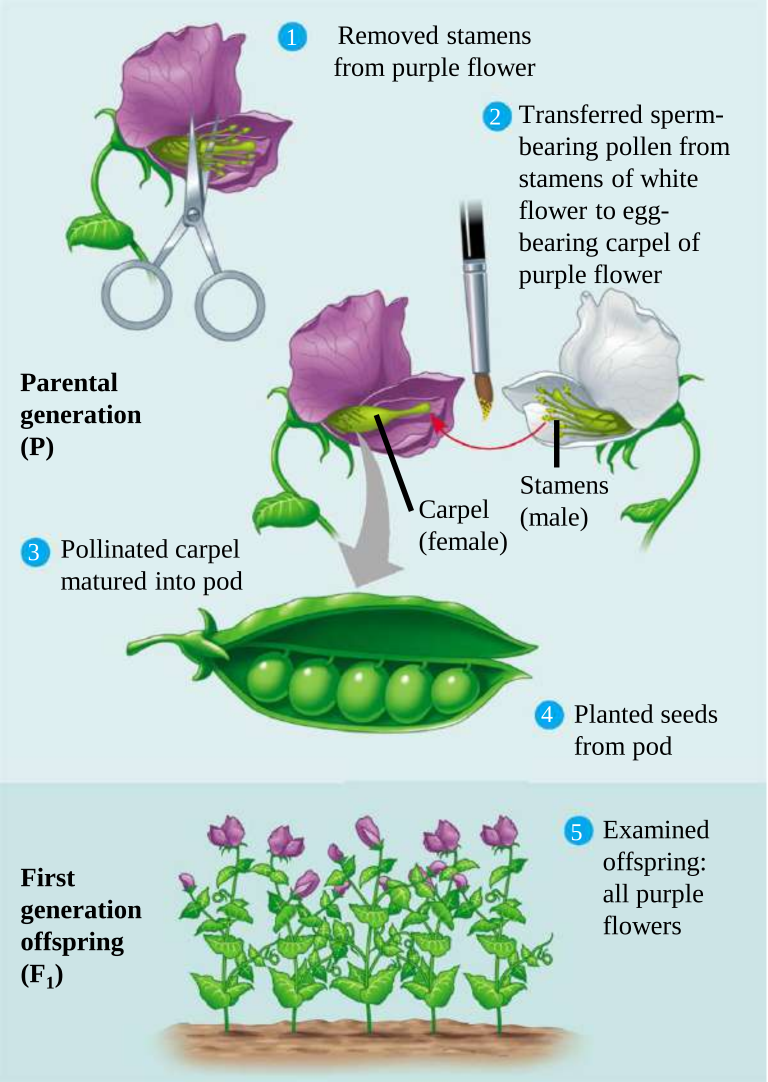
- Mendel chose to work with peas
  - Because they are available in many varieties
  - Because he could strictly control which plants mated with which



• Crossing pea plants

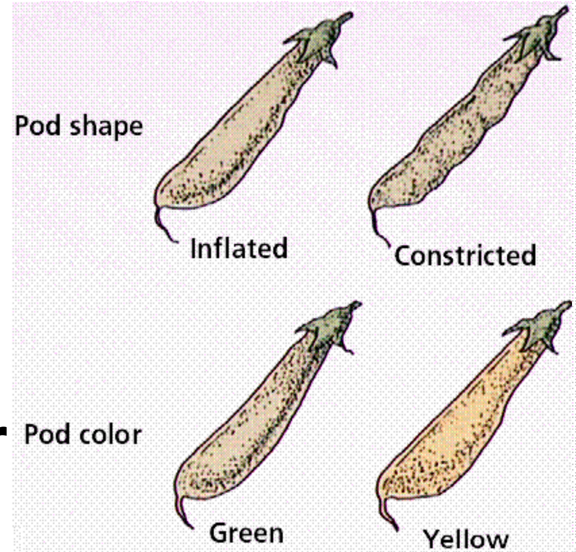
APPLICATION By crossing (mating) two true-breeding varieties of an organism, scientists can study patterns of inheritance. In this example, Mendel crossed pea plants that varied in flower color.

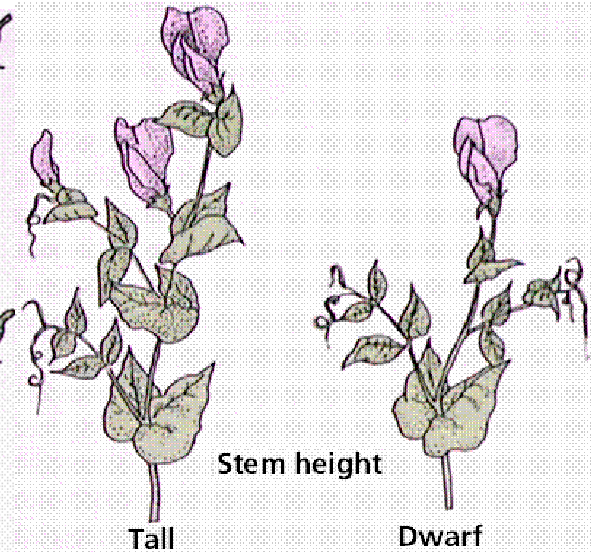
TECHNIQUE

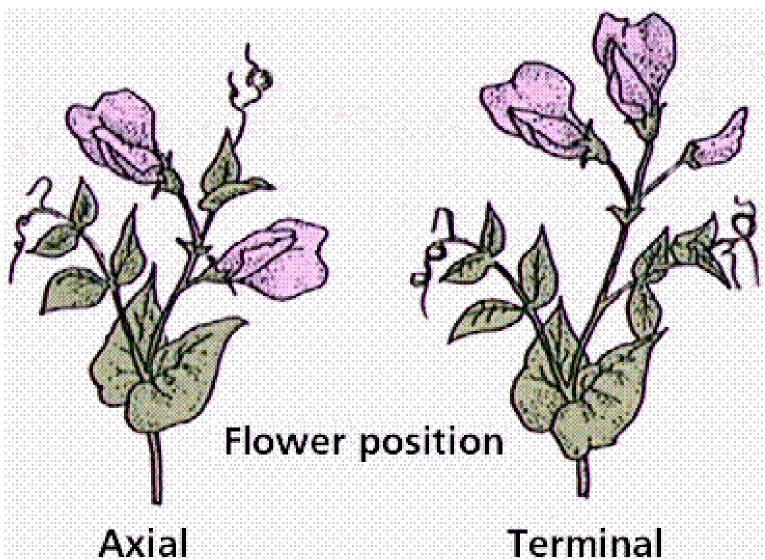


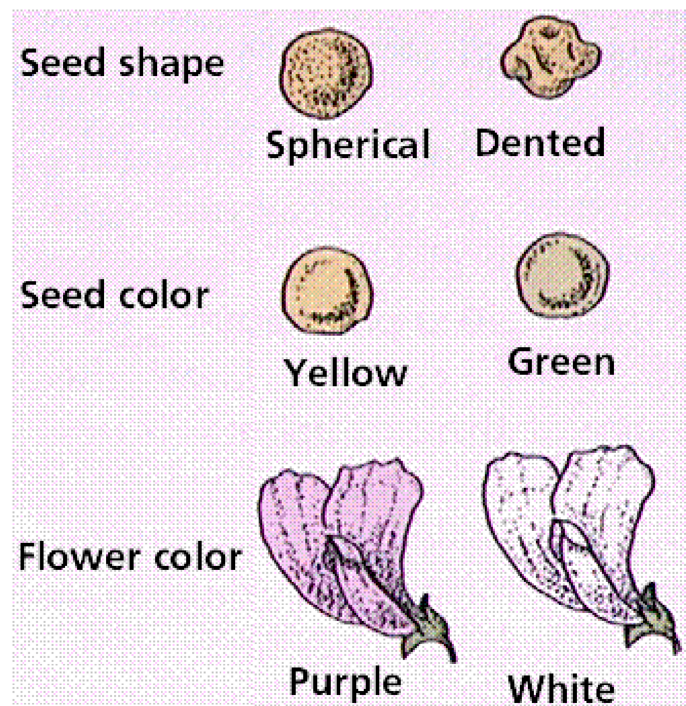
RESULTS When pollen from a white flower fertilizes eggs of a purple flower, the first-generation hybrids all have purple flowers. The result is the same for the reciprocal cross, the transfer of pollen from purple flowers to white flowers.

- Mendel studied 7 different traits:
  - Seed shape
  - Seed color
  - Pod color
  - **Plant height**
  - **Flower color**
  - Pod shape
  - Flower position







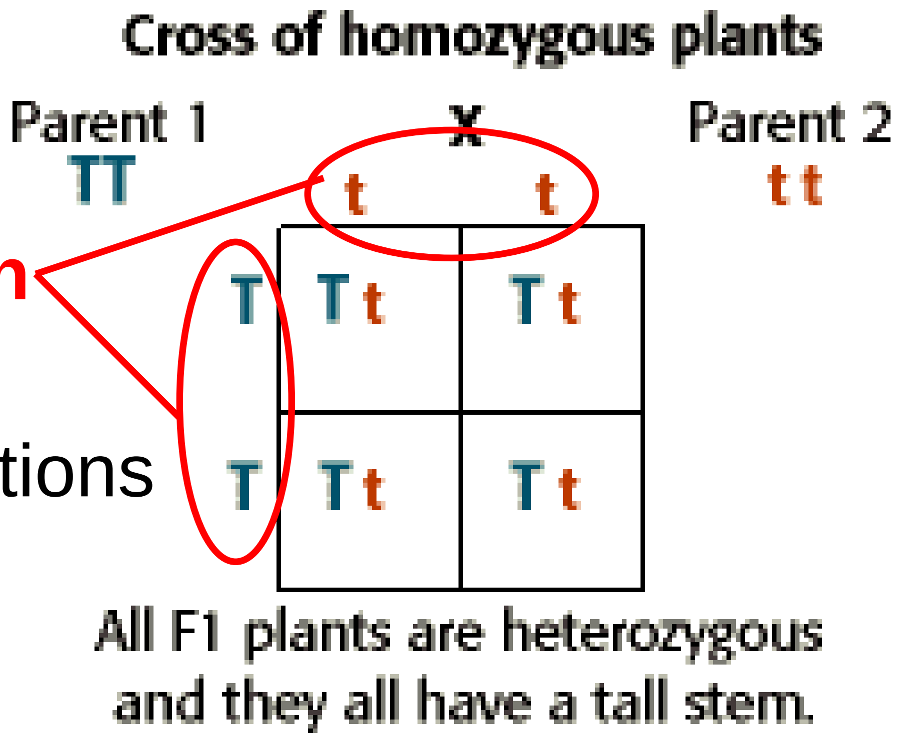


- He chose these traits because they each appeared in <u>2 distinct forms</u>. For example, the plants were either short or tall…there was no intermediate height.

- Mendel crossed ***pure-breeding*** plants with one another.

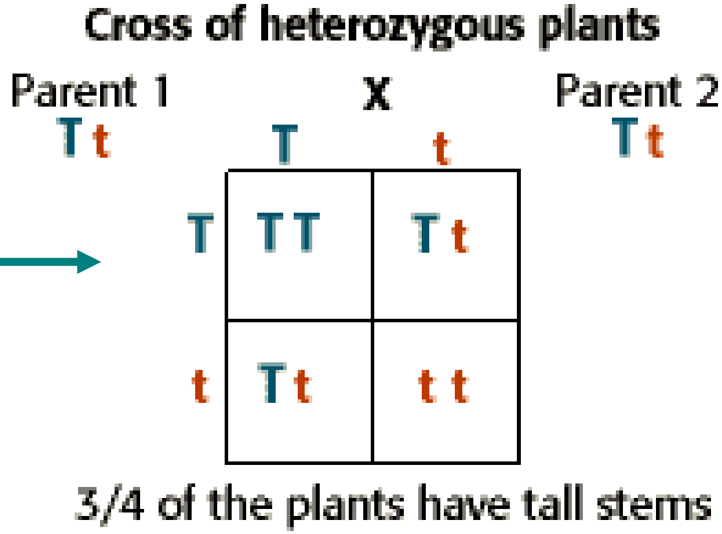
- The result: ***HYBRID*** plants (plants which received half of their genetic information form one type of parent, and the other half from a different type of parent).

- Example:
  - **Parent generation**
  - F_1 generation (possible combinations for offspring are inside square)



F_1 plants self-pollinate and reproduce (Tt x Tt)…
F_2 generation (inside square)

Punnett Square!



– **Dominant trait: <u>TALL STEM HEIGHT</u>**

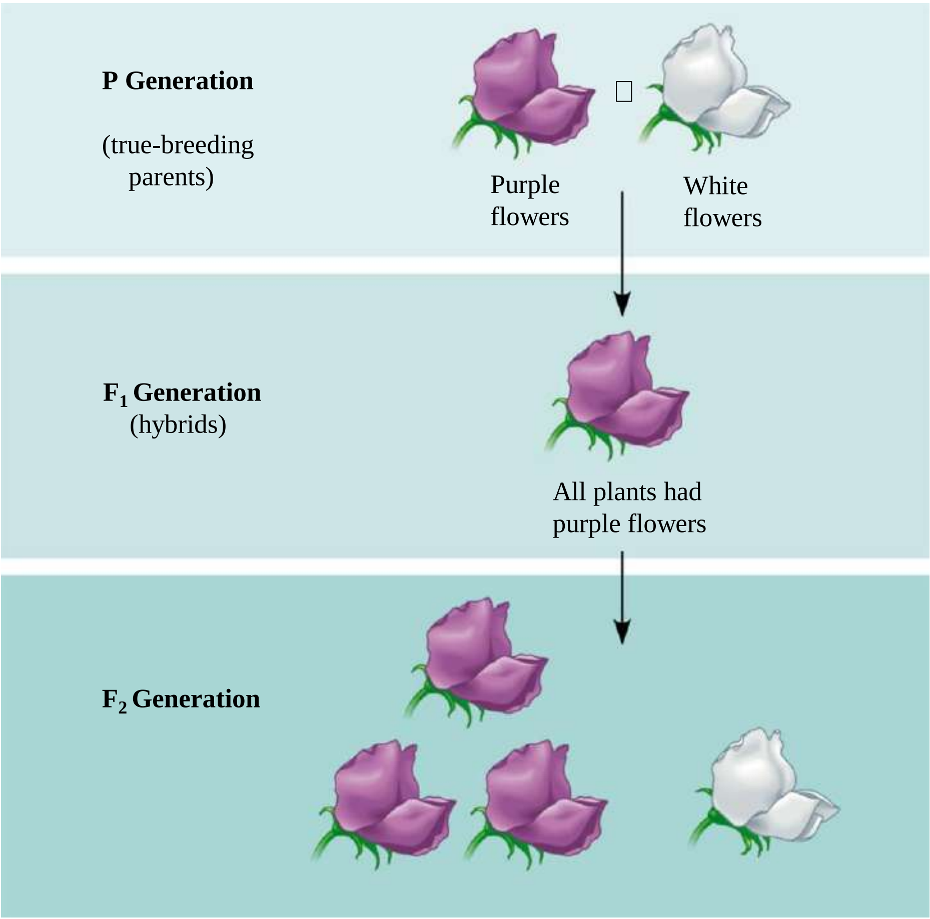
– **Recessive trait: <u>short stem height</u>**

The Law of Segregation

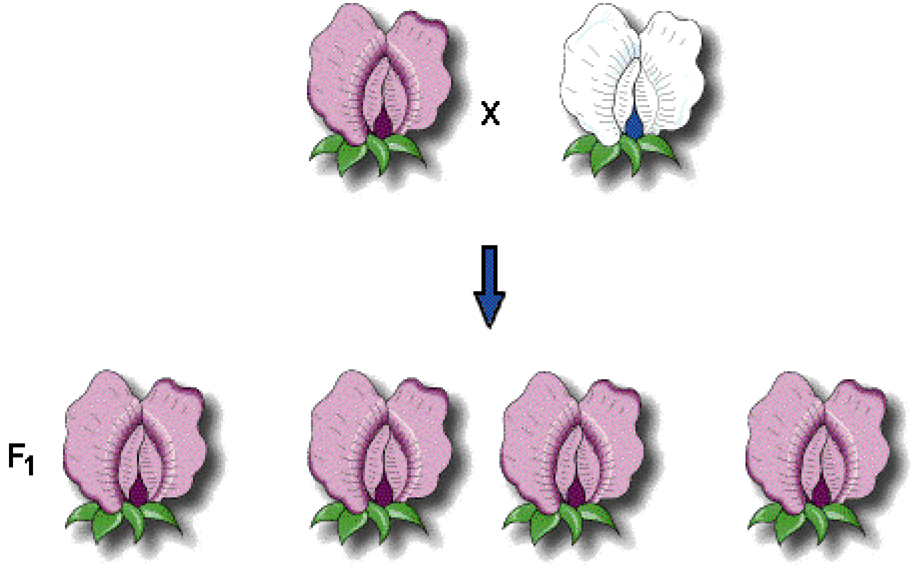
- When Mendel crossed contrasting, true-breeding white and purple flowered pea plants, **all of the offspring were purple**
- When Mendel crossed the F_1 plants, many of the plants had purple flowers, but some had white flowers (**approx. 3:1 ratio**)

EXPERIMENT True-breeding purple-flowered pea plants and white-flowered pea plants were crossed (symbolized by □). The resulting F_1 hybrids were allowed to self-pollinate or were cross-pollinated with other F_1 hybrids. Flower color was then observed in the F_2 generation.

RESULTS Both purple-flowered plants and white-flowered plants appeared in the F_2 generation. In Mendel's experiment, 705 plants had purple flowers, and 224 had white flowers, a ratio of about 3 purple : 1 white.



- Mendel reasoned that
  - In the F_1 plants, only the purple flower "factor" was affecting flower color in these hybrids
  - Purple flower color was **DOMINANT**, and white flower color was **RECESSIVE**



- Mendel observed the same pattern in many other pea plant characters

- His results led him to develop a hypothesis with 4 related ideas:

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

Table 14.1

- First, <u>alternative versions of genes</u> account for variations in inherited characters, which are now called **<u>ALLELES</u>**

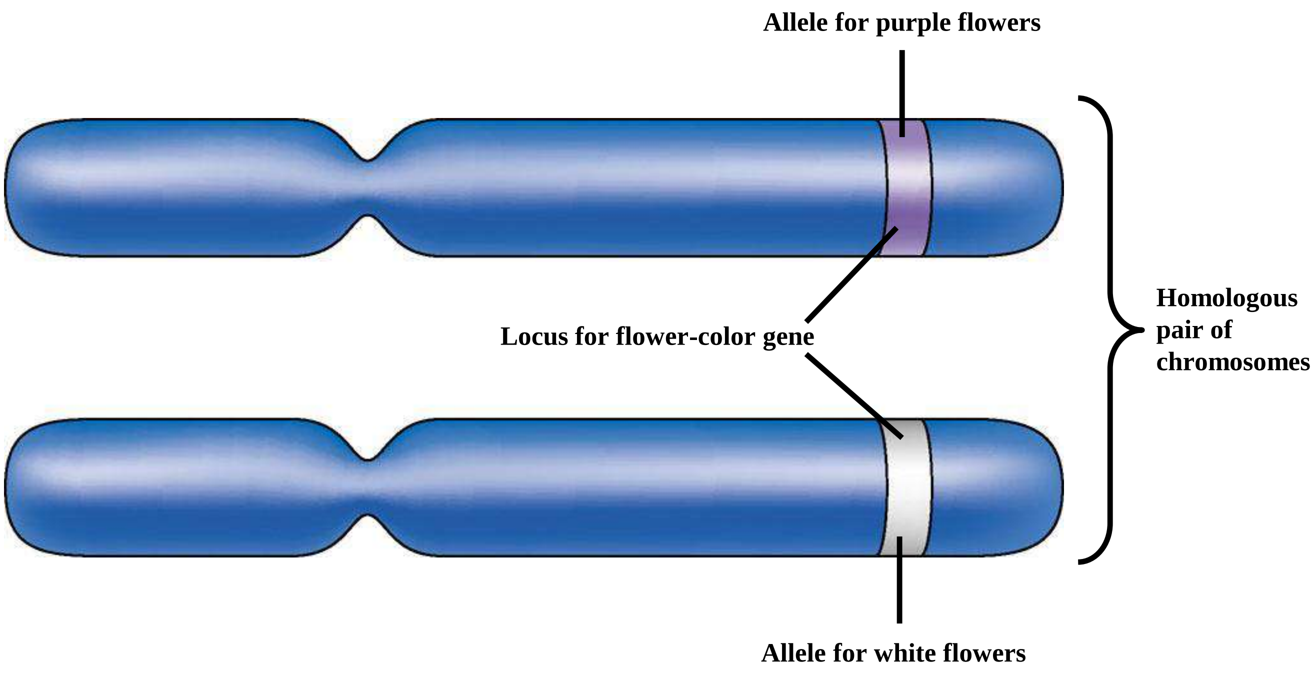
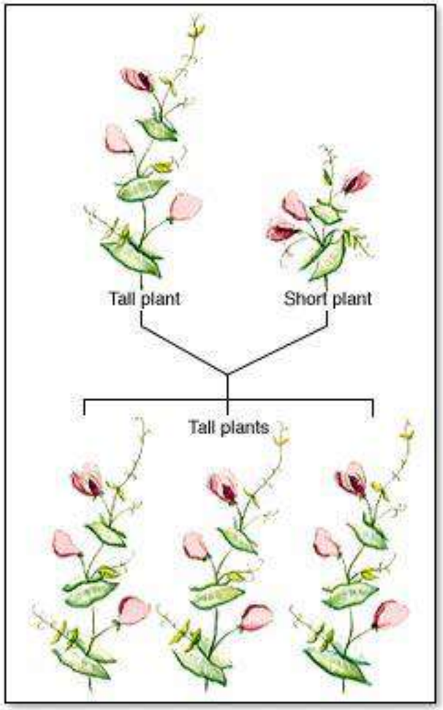
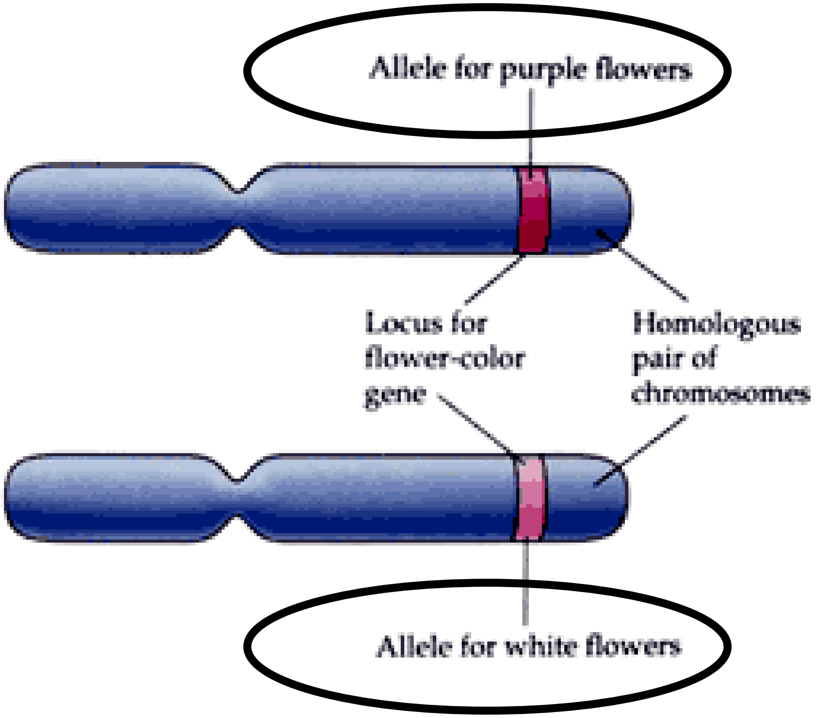


Figure 14.4

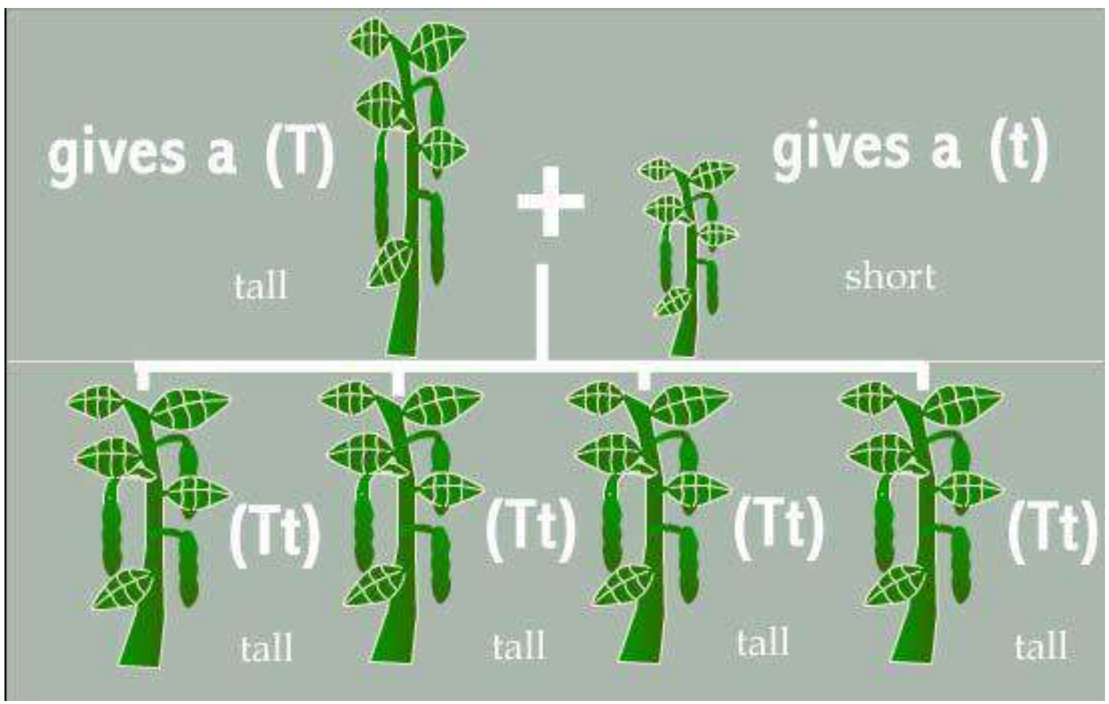
- **Example**: <u>tall (T) and short (t)</u> are alleles of the gene that controls height in pea plants.



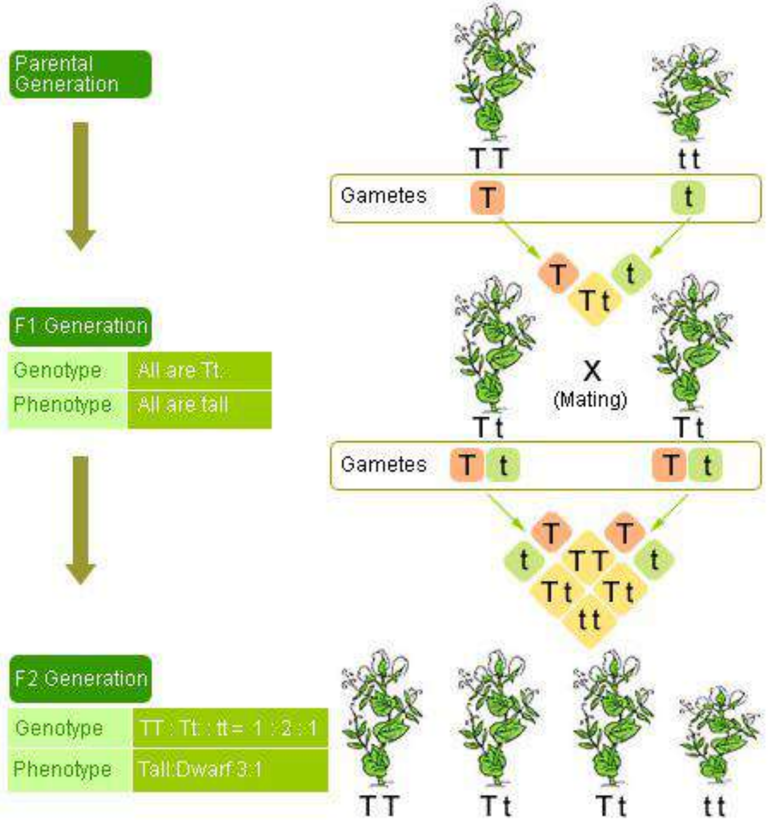


- Second, for each character an organism inherits **<u>two alleles</u>**, <u>one from each parent</u>

  (a genetic locus is actually represented twice)

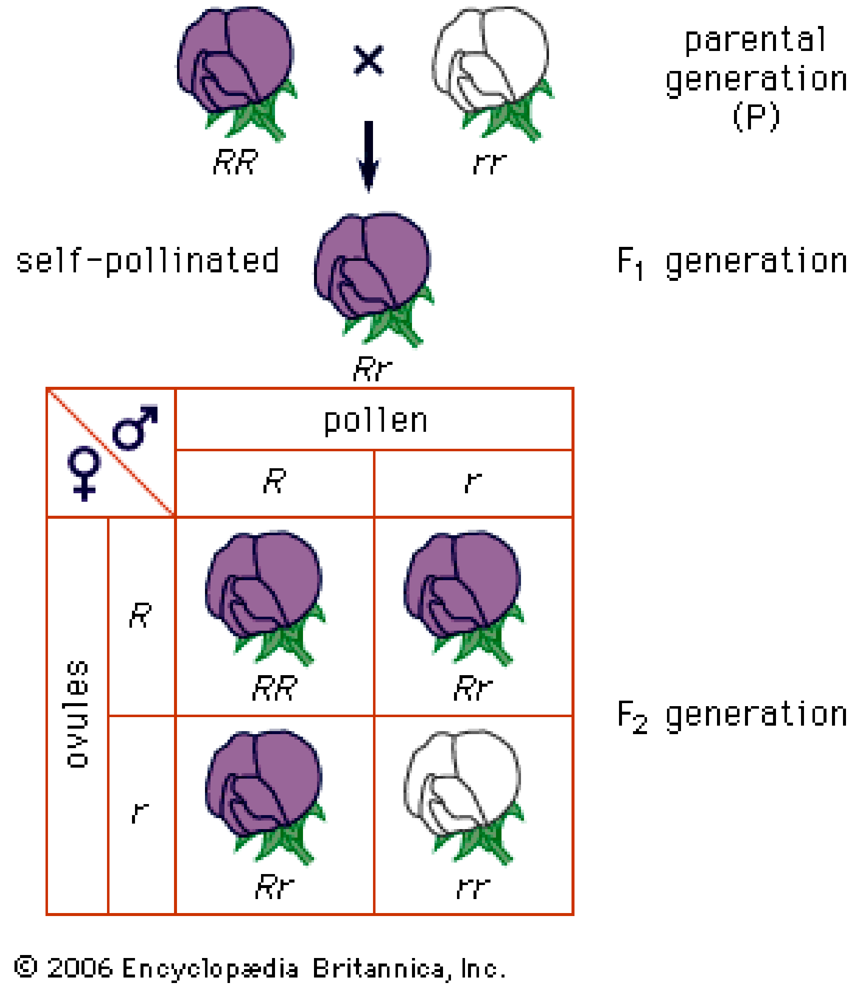


- Third, if the two alleles at a locus differ, then one, the **DOMINANT ALLELE**, <u>determines the organism's appearance</u>;
- The other allele, the **RECESSIVE ALLELE**, has no noticeable effect on the organism's appearance



- Fourth, the **<u>LAW OF SEGREGATION</u>**:

  the two alleles for a heritable character separate (segregate) <u>during gamete formation and end up in different gametes</u>



- Mendel's law of segregation, probability and the Punnett square

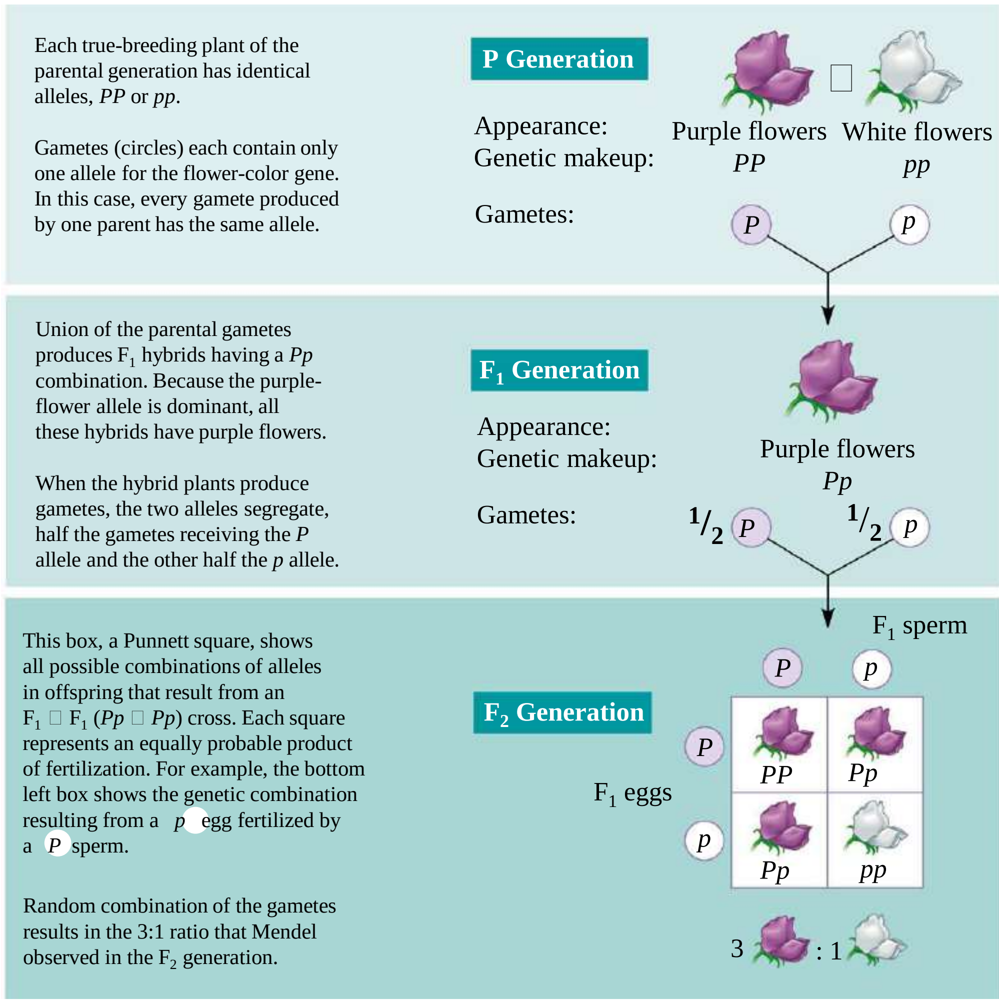


Figure 14.5

Useful Genetic Vocabulary

- An organism that is **HOMOZYGOUS** for a particular gene:

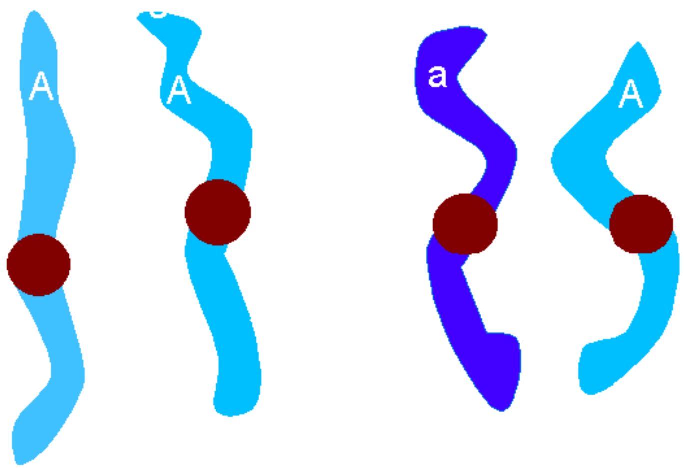
  -has a pair of identical alleles for that gene

  (RR or rr)

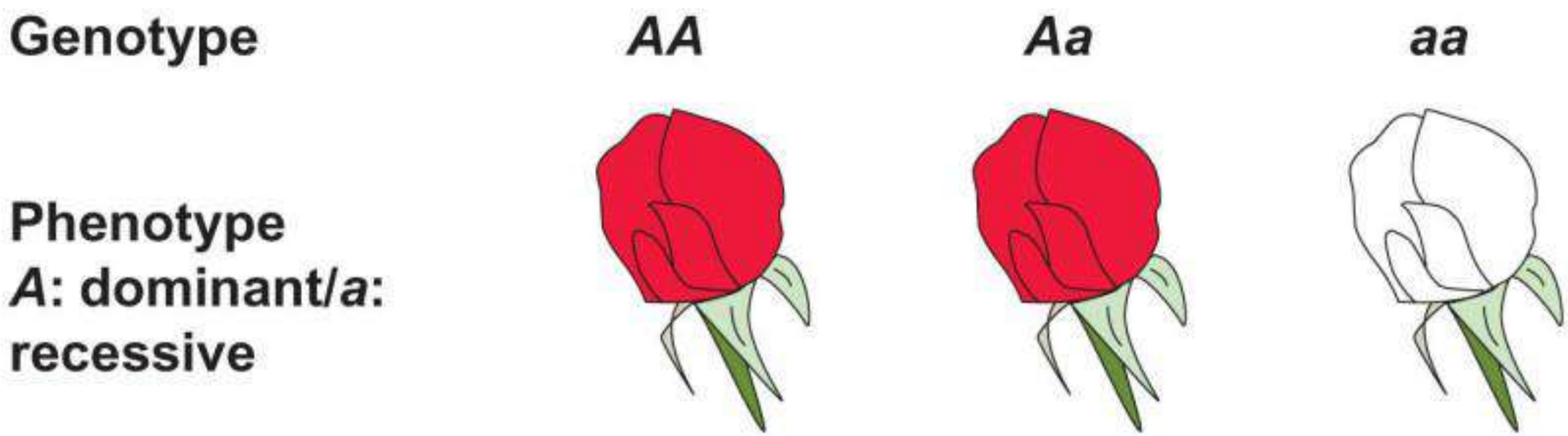
  -exhibits true-breeding

- An organism that is **HETEROZYGOUS** for a particular gene:

  -has a pair of alleles that are different for that gene **(Rr)**



- An organism's **PHENOTYPE is** its physical appearance **(purple flowers)**
- An organism's **GENOTYPE** is its genetic makeup **(PP or Pp)**



- Phenotype versus genotype

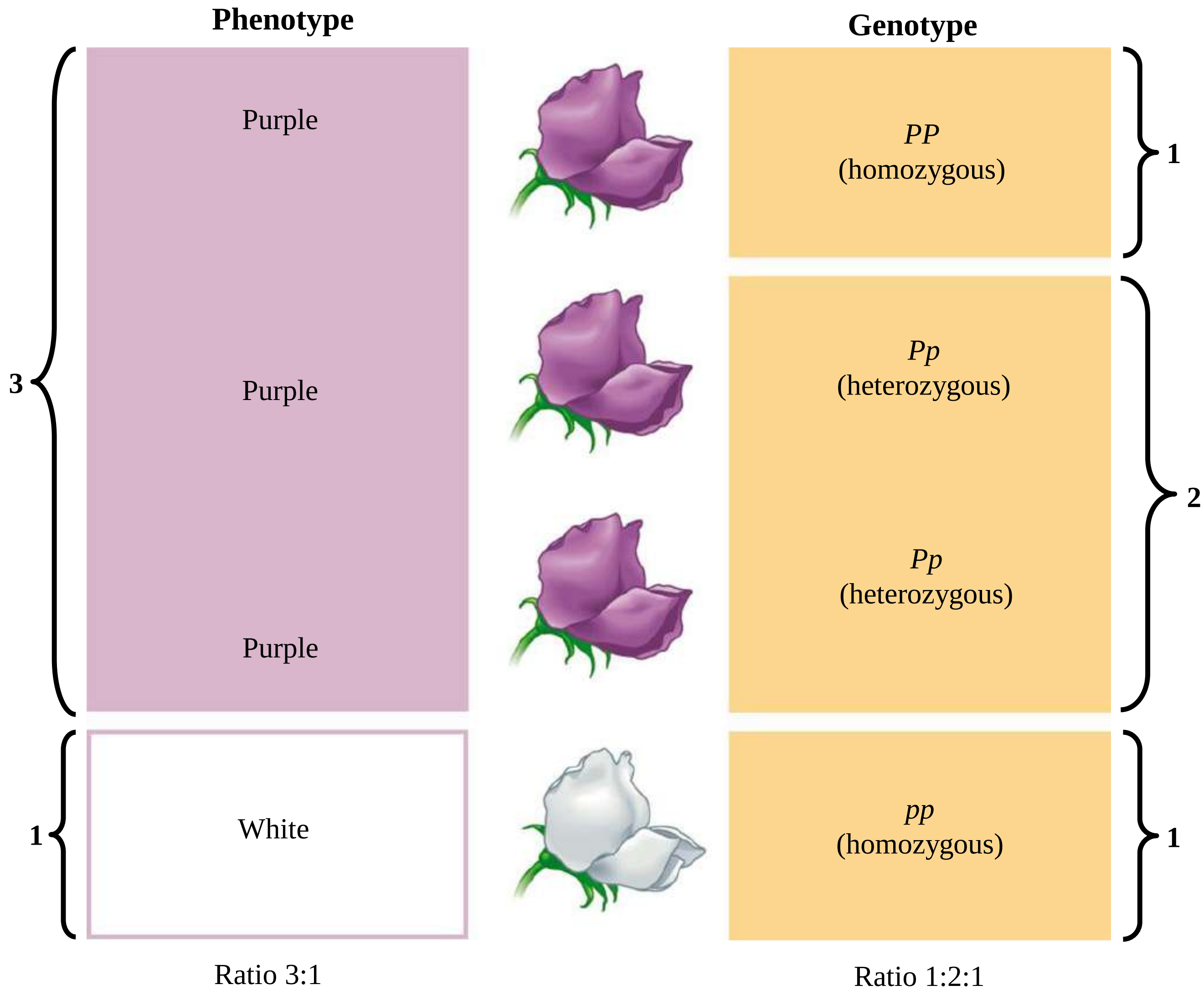
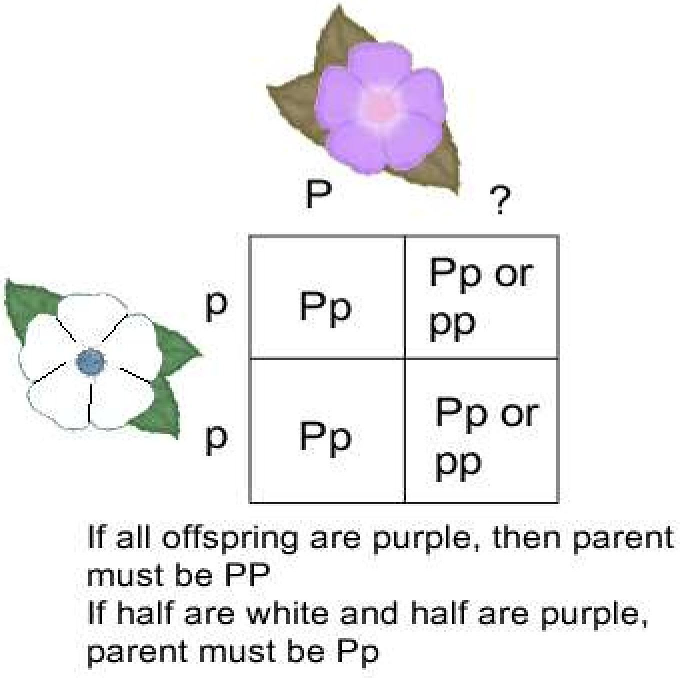


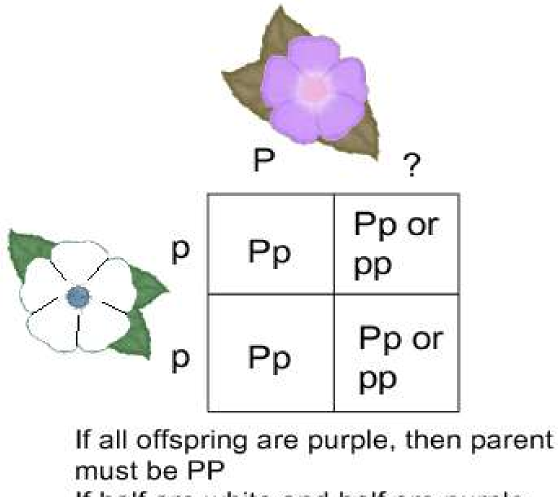
Figure 14.6

The Testcross:

- In pea plants with purple flowers the genotype is not immediately obvious (could be **PP or Pp**)



- a testcross allows us to <u>determine the genotype of an organism with the dominant phenotype</u>, but unknown genotype
- an individual with the dominant phenotype is crossed with an individual that is homozygous recessive for a trait

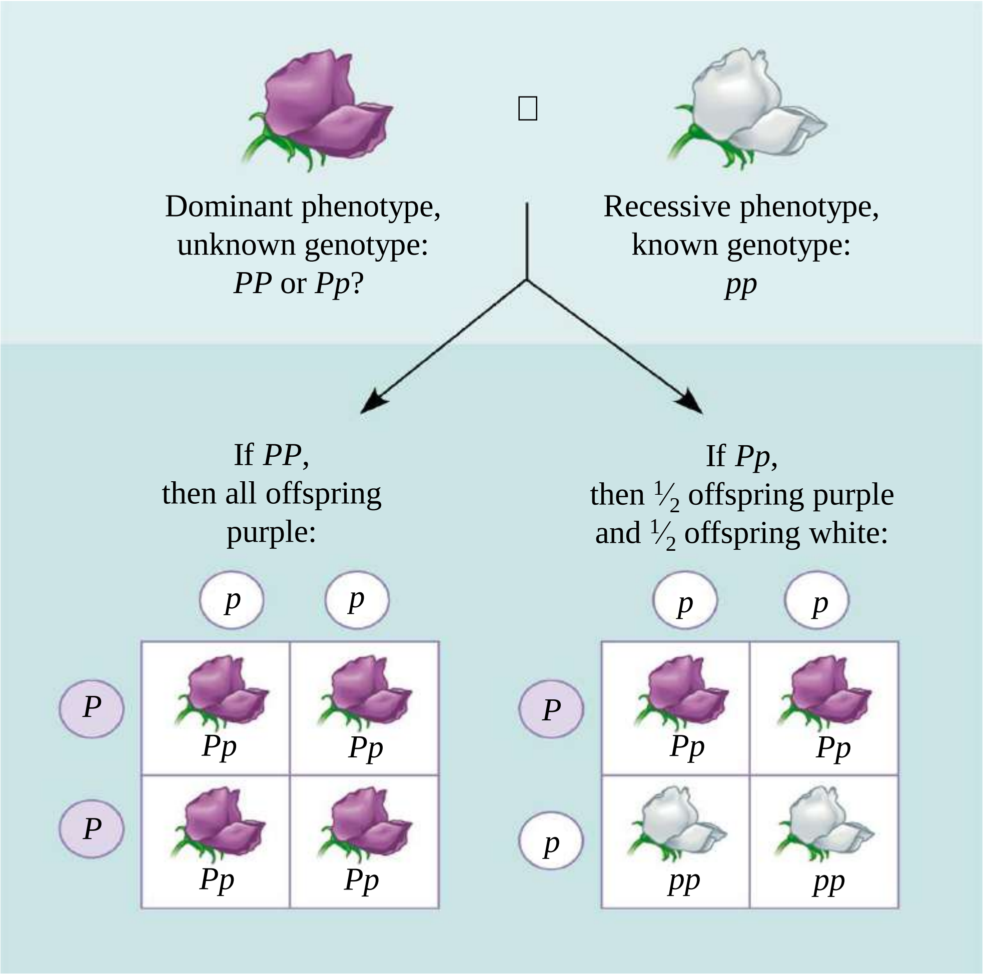


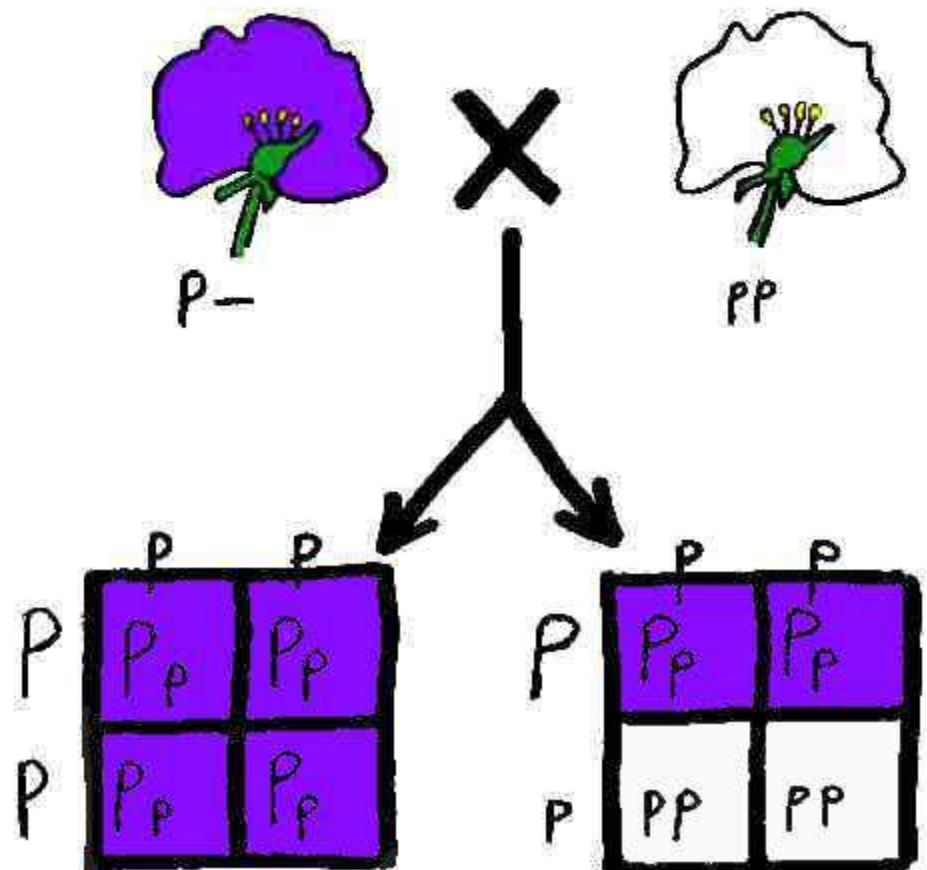
• The testcross:

APPLICATION An organism that exhibits a dominant trait, such as purple flowers in pea plants, can be either homozygous for the dominant allele or heterozygous. To determine the organism's genotype, geneticists can perform a testcross.

TECHNIQUE In a testcross, the individual with the unknown genotype is crossed with a homozygous individual expressing the recessive trait (white flowers in this example). By observing the phenotypes of the offspring resulting from this cross, we can deduce the genotype of the purple-flowered parent.

RESULTS




P–
pp
p
p
P
Pp
Pp
P
Pp
Pp
p
p
P
Pp
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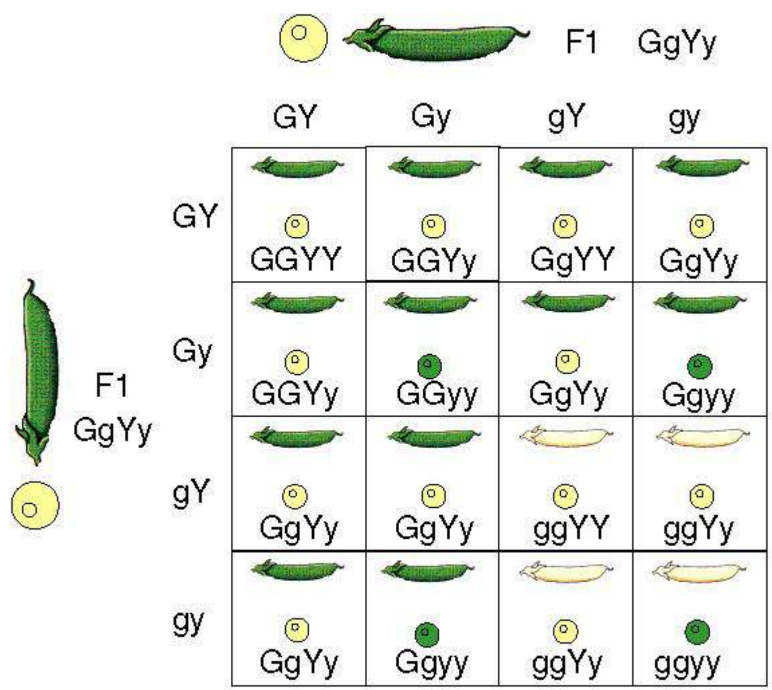
p
pp
pp

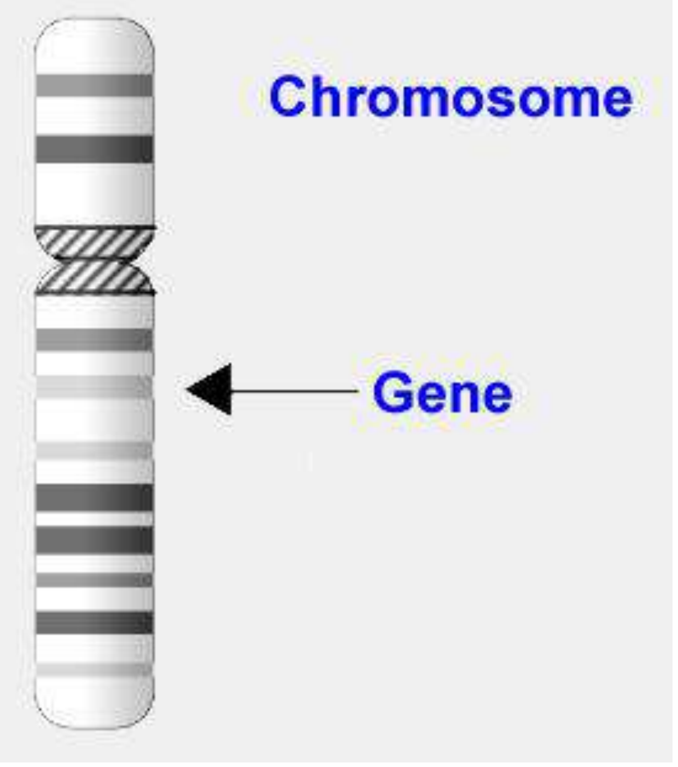
<u>The Law of Independent Assortment</u>

- Mendel derived the law of segregation by following a single trait
- The F_1 offspring produced in this cross were monohybrids, <u>heterozygous for one character</u>

- Mendel identified his second law of inheritance by **following two characters** at the same time
- Crossing two, true-breeding parents differing in two characters produces **DIHYBRIDS** in the F_1 generation, heterozygous for both characters



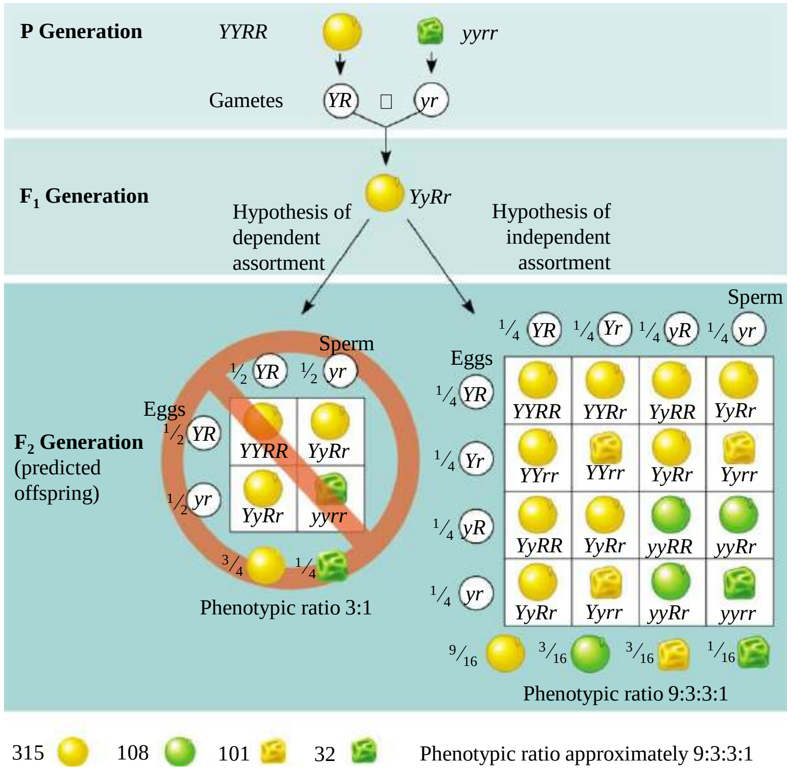
- How are two different characters transmitted from parents to offspring: ***together or independently?***



- A dihybrid cross illustrates the inheritance of two characters
- The result: <u>**4 phenotypes in the F_2 generation**</u>

EXPERIMENT Two true-breeding pea plants—one with yellow-round seeds and the other with green-wrinkled seeds—were crossed, producing dihybrid F_1 plants. Self-pollination of the F_1 dihybrids, which are heterozygous for both characters, produced the F_2 generation. The two hypotheses predict different phenotypic ratios. Note that yellow color (*Y*) and round shape (*R*) are dominant.

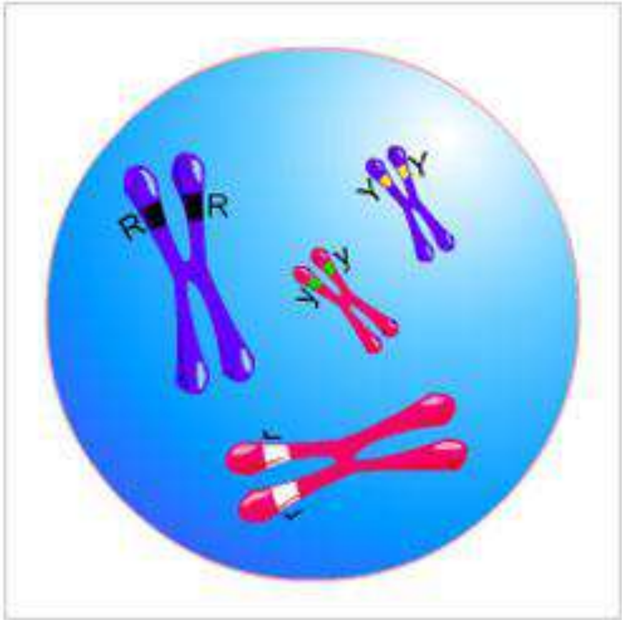
RESULTS



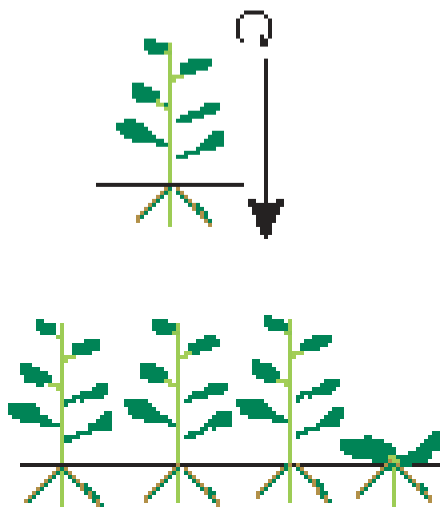
CONCLUSION The results support the hypothesis of independent assortment. The alleles for seed color and seed shape sort into gametes independently of each other.

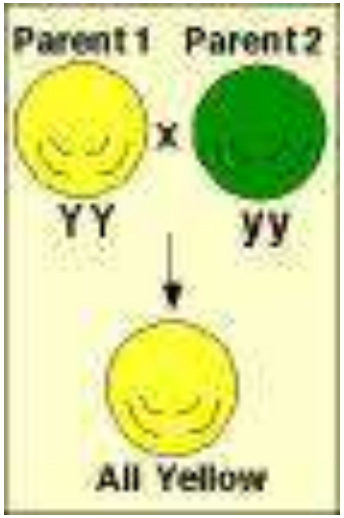
- Using the information from a dihybrid cross, Mendel developed the **<u>LAW OF INDEPENDENT ASSORTMENT</u>**:

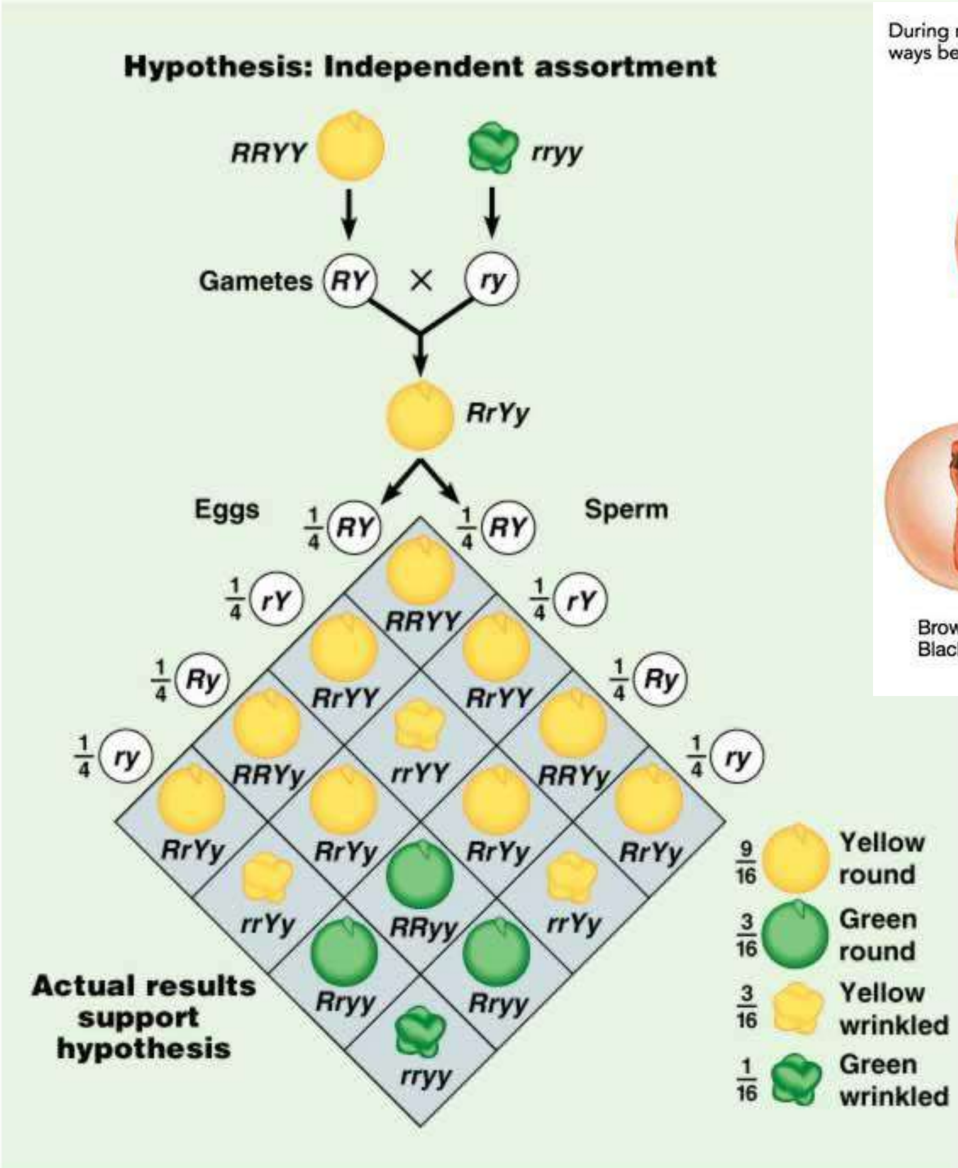
  Each pair of alleles segregates <u>independently during gamete formation</u>

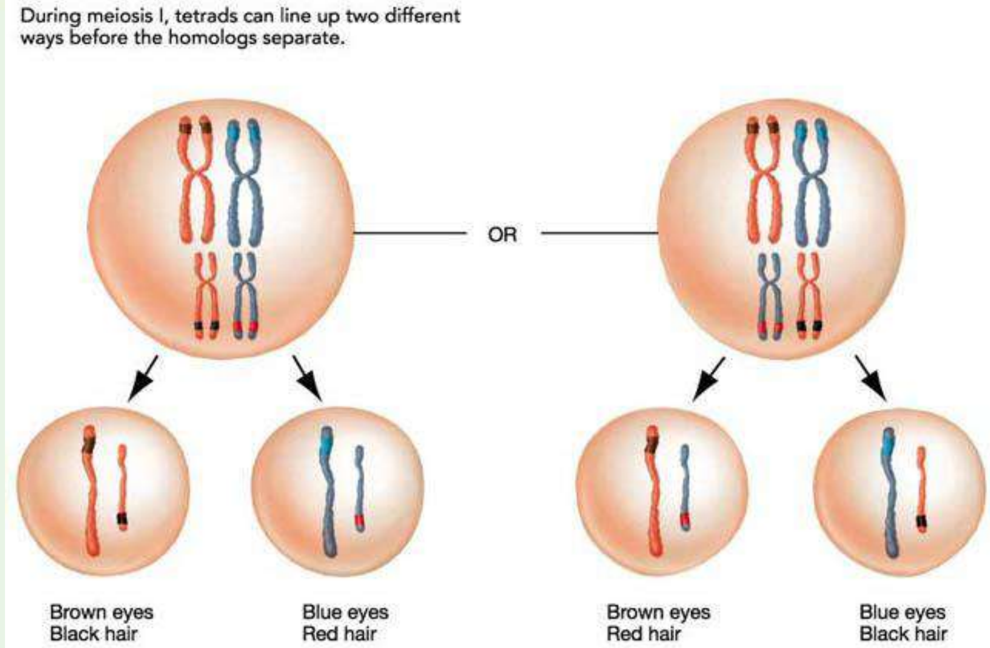


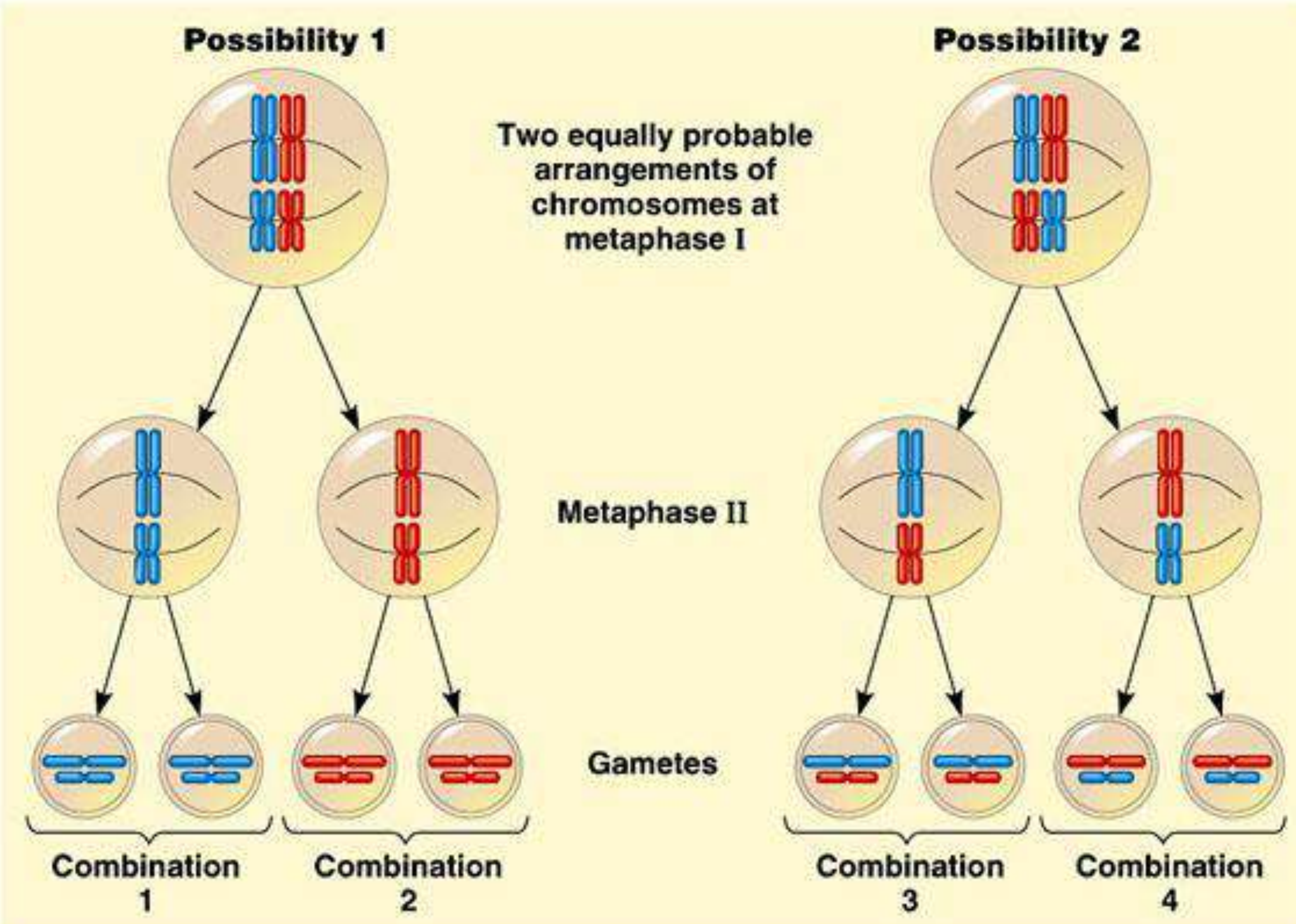
For example, in pea plants, the allele for tallness may be inherited with the allele for yellow seed color, ***or* the allele for green seed color. This is because the separation of the chromosomes during meiosis is random and produces many combinations of chromosomes.

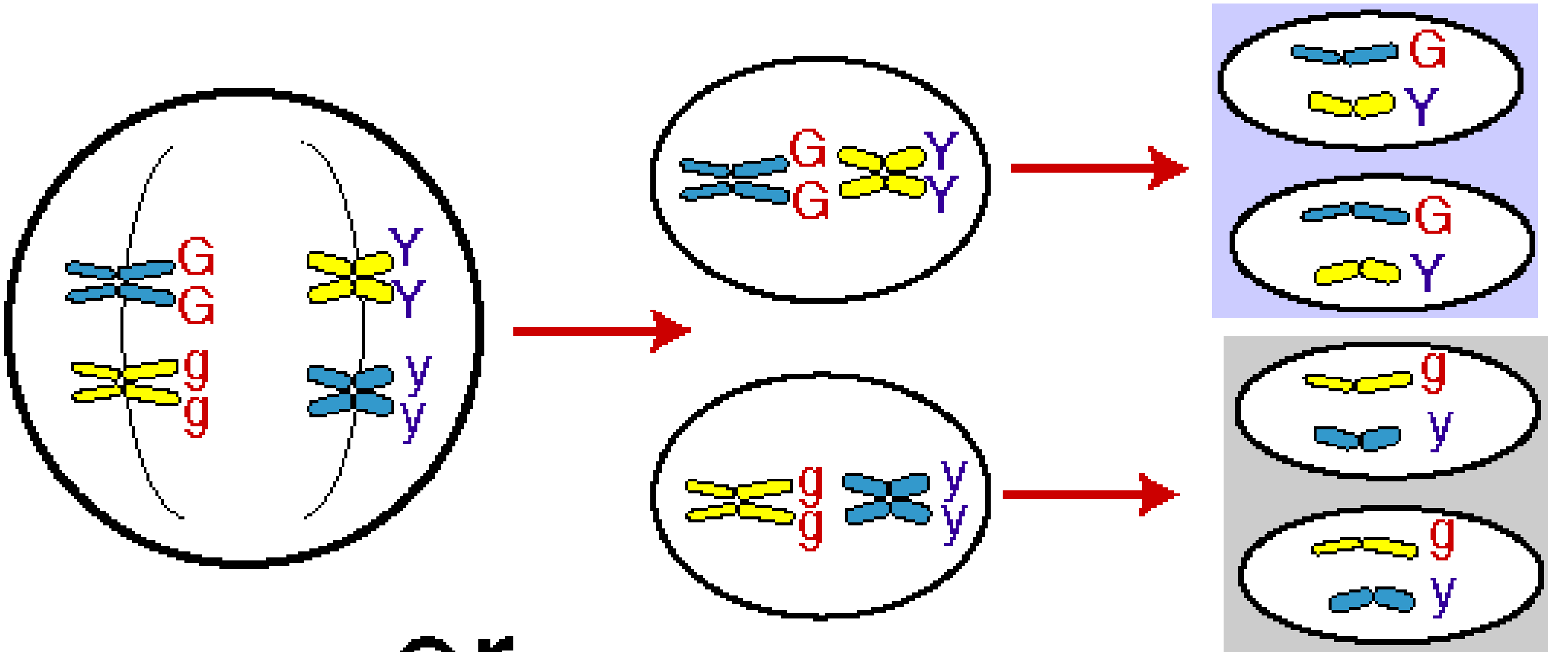




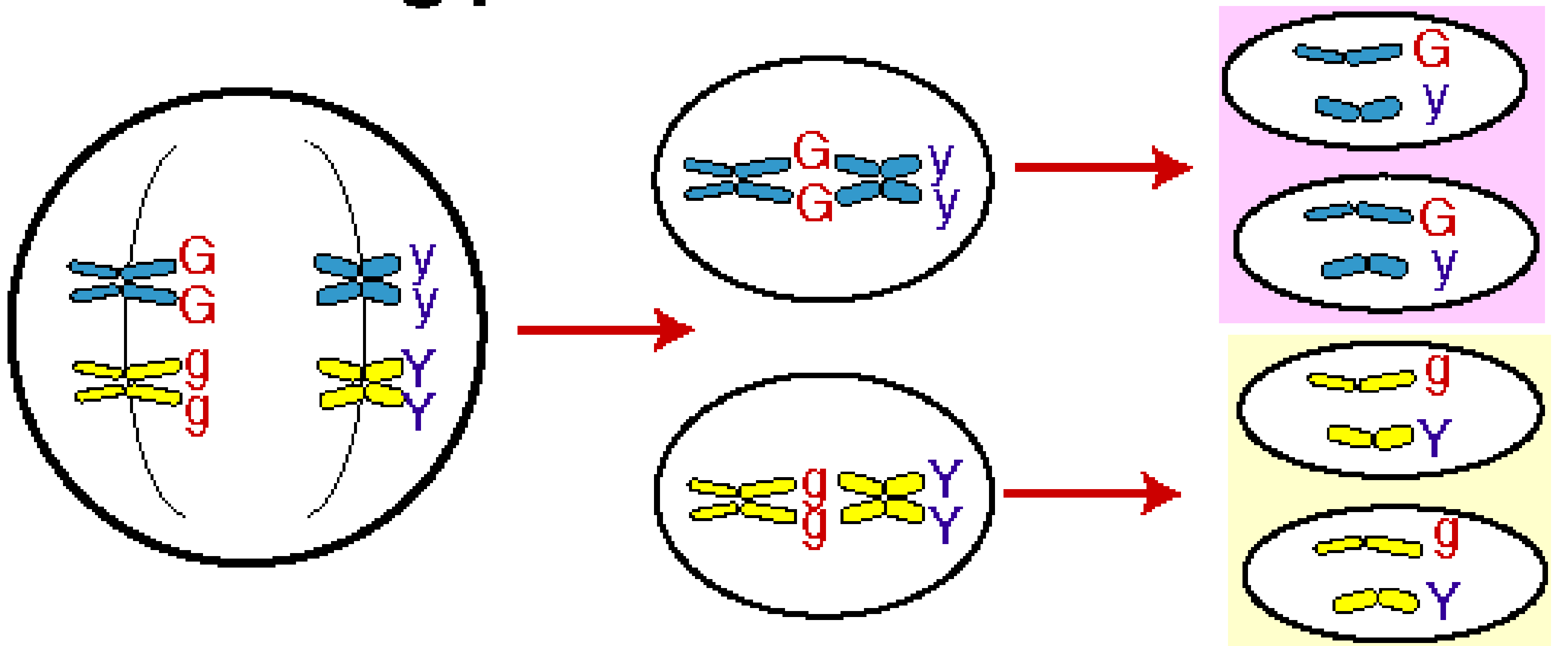

Hypothesis: Independent assortment
RRYY
rryy
Gametes
RY
×
ry
RrYy
Eggs
Sperm
1/4 RY
1/4 rY
1/4 Ry
1/4 ry
RRYY
RrYY
RrYY
RRYy
rrYY
RRYy
RrYy
RrYy
RrYy
RrYy
rrYy
RRyy
rrYy
Rryy
Rryy
rryy
9/16 Yellow round
3/16 Green round
3/16 Yellow wrinkled
1/16 Green wrinkled
Actual results support hypothesis


During meiosis I, tetrads can line up two different ways before the homologs separate.
OR
Brown eyes
Black hair
Blue eyes
Red hair
Brown eyes
Red hair
Blue eyes
Black hair


Possibility 1
Possibility 2
Two equally probable arrangements of chromosomes at metaphase I
Metaphase II
Gametes
Combination 1
Combination 2
Combination 3
Combination 4



or



<u>Laws of Probability & Genetics (14.2)</u>

- **multiplication rule**: to determine the prob. that 2 or more independent events will occur together, we multiply the prob of 1 event by the prob of the other event
- **example**: the prob of 2 coins both coming up "heads" is: <u>**½ x ½ = ¼**</u>

MONOHYBRID CROSS:

- Cystic fibrosis is an autosomal recessive disorder. What is the probability that a couple who are both carriers of this disease will have a child who has the disorder?

MONOHYBRID CROSS:

- Cystic fibrosis is an autosomal recessive disorder. What is the probability that a couple who are both carriers of this disease will have a child who has the disorder?

	R	r
R	**RR**	**Rr**
r	**Rr**	**rr**

¼ chance child will have the disorder

<u>DIHYBRID CROSS:</u>

- Two plants, heterozygous for purple flowers (Pp) and heterozygous for tall stems (Tt) are crossed. What fraction of their offspring will have white flowers and tall stems?

<u>CROSS: PpTt x PpTt</u>

*****each plant can produce the following gametes: <u>PT, Pt, pT, pt</u>***

CROSS: PpTt x PpTt

	PT	**Pt**	**pT**	**pt**
PT	PPTT	PPTt	PpTT	PpTt
Pt	PPTt	PPtt	PpTt	Pptt
pT	PpTT	PpTt	ppTT	ppTt
pt	PpTt	Pptt	ppTt	pptt

CROSS: <u>PpTt x PpTt</u>

	PT	**Pt**	**pT**	**pt**
PT	PPTT Purple, tall	PPTt Purple, tall	PpTT Purple, tall	PpTt Purple, tall
Pt	PPTt Purple, tall	PPtt Purple, short	PpTt Purple, tall	Pptt Purple, short
pT	PpTT Purple, tall	PpTt Purple, tall	ppTT white, tall	ppTt white, tall
pt	PpTt Purple, tall	Pptt Purple, short	ppTt white, tall	pptt White, short

CROSS: PpTt x PpTt

	PT	**Pt**	**pT**	**pt**
PT	PPTT Purple, tall	PPTt Purple, tall	PpTT Purple, tall	PpTt Purple, tall
Pt	PPTt Purple, tall	PPtt Purple, short	PpTt Purple, tall	Pptt Purple, short
pT	PpTT Purple, tall	PpTt Purple, tall	ppTT white, tall	ppTt white, tall
pt	PpTt Purple, tall	Pptt Purple, short	ppTt white, tall	pptt White, short

DIHYBRID CROSS:

- Two plants, heterozygous for purple flowers (Pp) and heterozygous for tall stems (Tt) are crossed. What fraction of their offspring will have white flowers and tall stems?

3/16 or 18.75%

Solving Complex Genetics Problems with Probability:

- Consider the cross:

PpYyRr x Ppyyrr

- What fraction of the offspring from this cross will exhibit the recessive phenotype for all 3 traits? **(ppyyrr)**

Solving Complex Genetics Problems with Probability:

- Consider the cross:

  PpYyRr x Ppyyrr

- Consider each gene separately (P, Y, R)
- Pp x Pp — ***what fraction of offspring will be "pp"?***
- **¼ !!**

Solving Complex Genetics Problems with Probability:

- Consider the cross:

  PpYyRr x Ppyyrr

- Yy x yy   ***what fraction of offspring will be "yy"?***

- **½ !!**

<u>Solving Complex Genetics Problems with Probability:</u>

- Consider the cross:

PpYyRr x Ppyyrr

- Rr x rr ***what fraction of offspring will be "rr"?***

- **<u>½ !!</u>**

<u>Solving Complex Genetics Problems with Probability:</u>

- Consider the cross:

  PpYyRr x Ppyyrr

- SO, what fraction of the offspring from this cross will exhibit the recessive phenotype for all 3 traits? (ppyyrr)
- **<u>$\frac{1}{4}$ x $\frac{1}{2}$ x $\frac{1}{2}$ = 1/16 or 6.25%</u>**