Fundamentals of Genetics

Gregor Mendel (1822-1884)

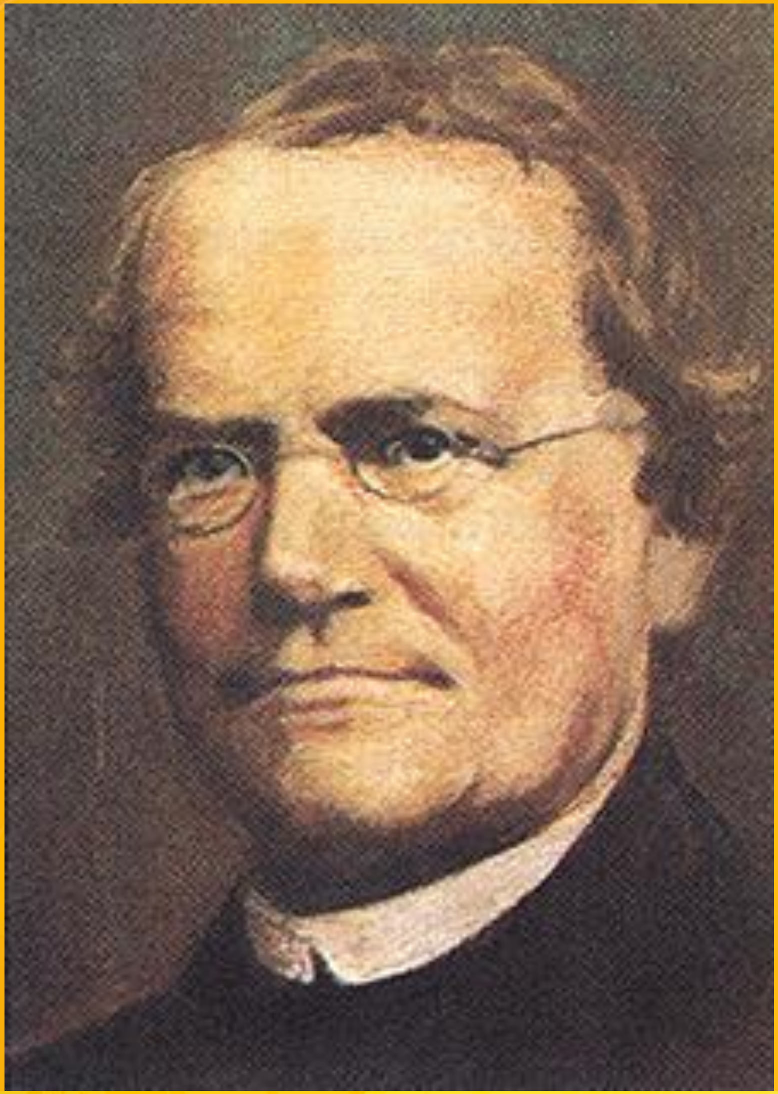
Responsible for the Laws governing Inheritance of Traits

Gregor Johann Mendel

- Austrian monk
- Studied the inheritance of traits in pea plants
- Developed the laws of inheritance
- Mendel's work was not recognized until the turn of the 20th century

Gregor Johann Mendel

- Between 1856 and 1863, Mendel cultivated and tested some 28,000 pea plants
- He found that the plants' offspring retained traits of the parents
- Called the "Father of Genetics"



Site of Gregor Mendel's experimental garden in the Czech Republic

Particulate Inheritance

- Mendel stated that physical traits are inherited as "particles"
- Mendel did not know that the "particles" were actually Chromosomes & DNA

Genetic Terminology

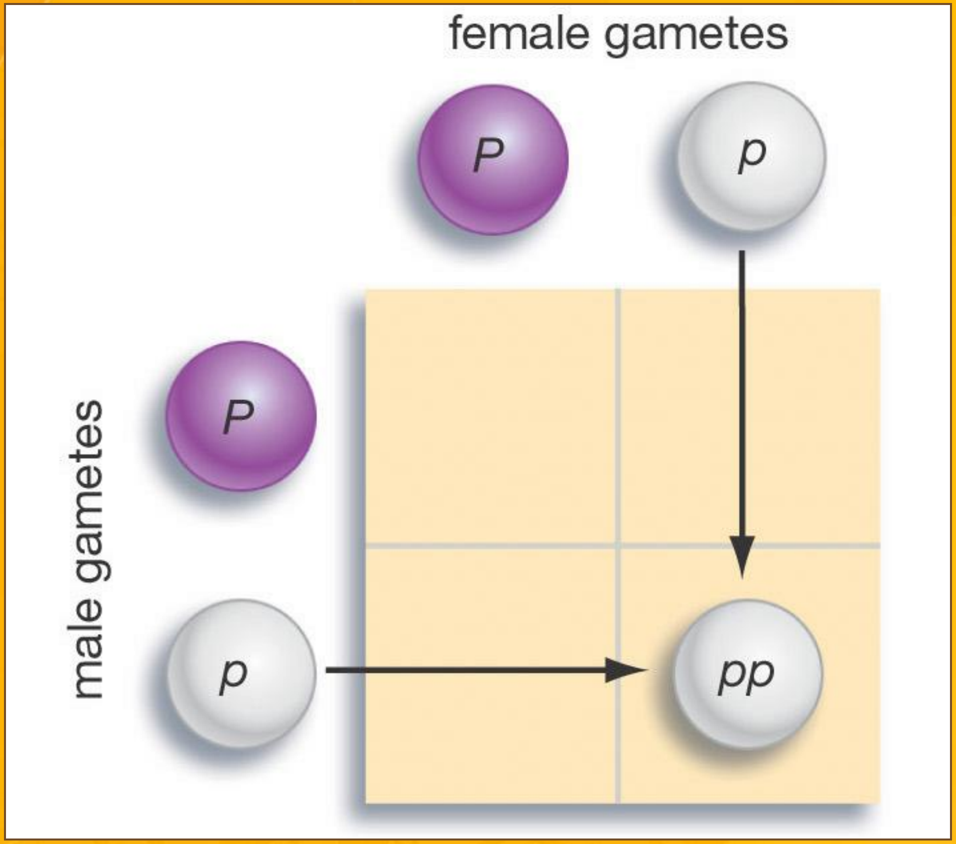
- **Trait** - any characteristic that can be passed from parent to offspring
- **Heredity** - passing of traits from parent to offspring
- **Genetics** - study of heredity

Types of Genetic Crosses

- **Monohybrid cross** - cross involving a single trait
  e.g. flower color
- **Dihybrid cross** - cross involving two traits
  e.g. flower color & plant height

Punnett Square

Used to help solve genetics problems



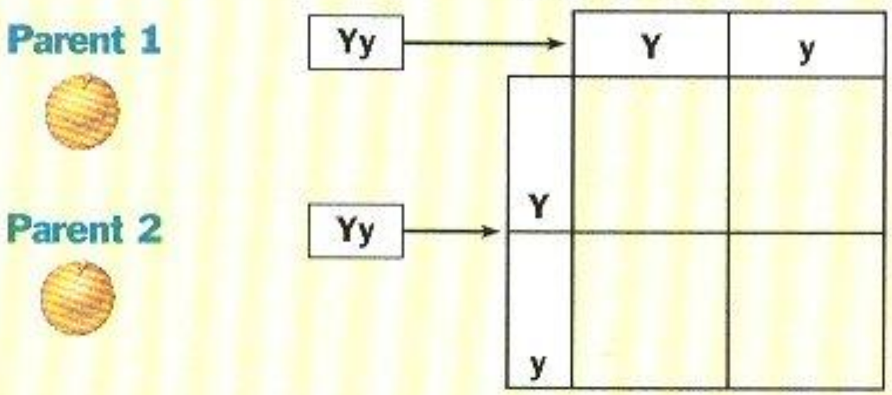
How to Make a Punnett Square

Punnett squares allow geneticists to predict the possible genotypes and phenotypes of offspring.

In this example, both parents are heterozygous for yellow-pea allele (*Yy*).

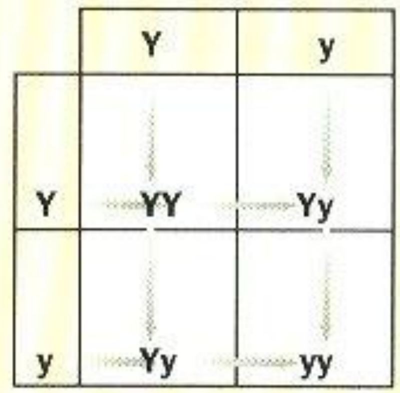
1 Make the grid

Place the alleles of the gametes of one parent along the top of a grid and those of the other parent along the left-hand side.



2 Fill in the grid

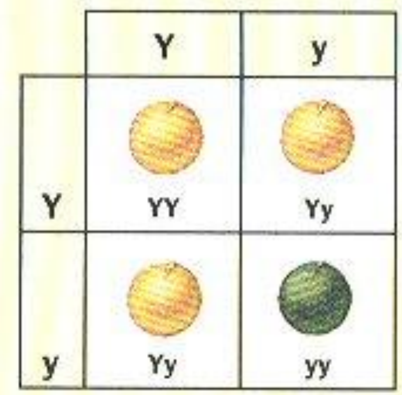
Combine the parent alleles inside the boxes. The letters show the genotypes of the offspring.



The genotype ratio is 1:2:1, meaning 1 *YY*, 2 *Yy*, 1 *yy*.

3 Fill in the offspring

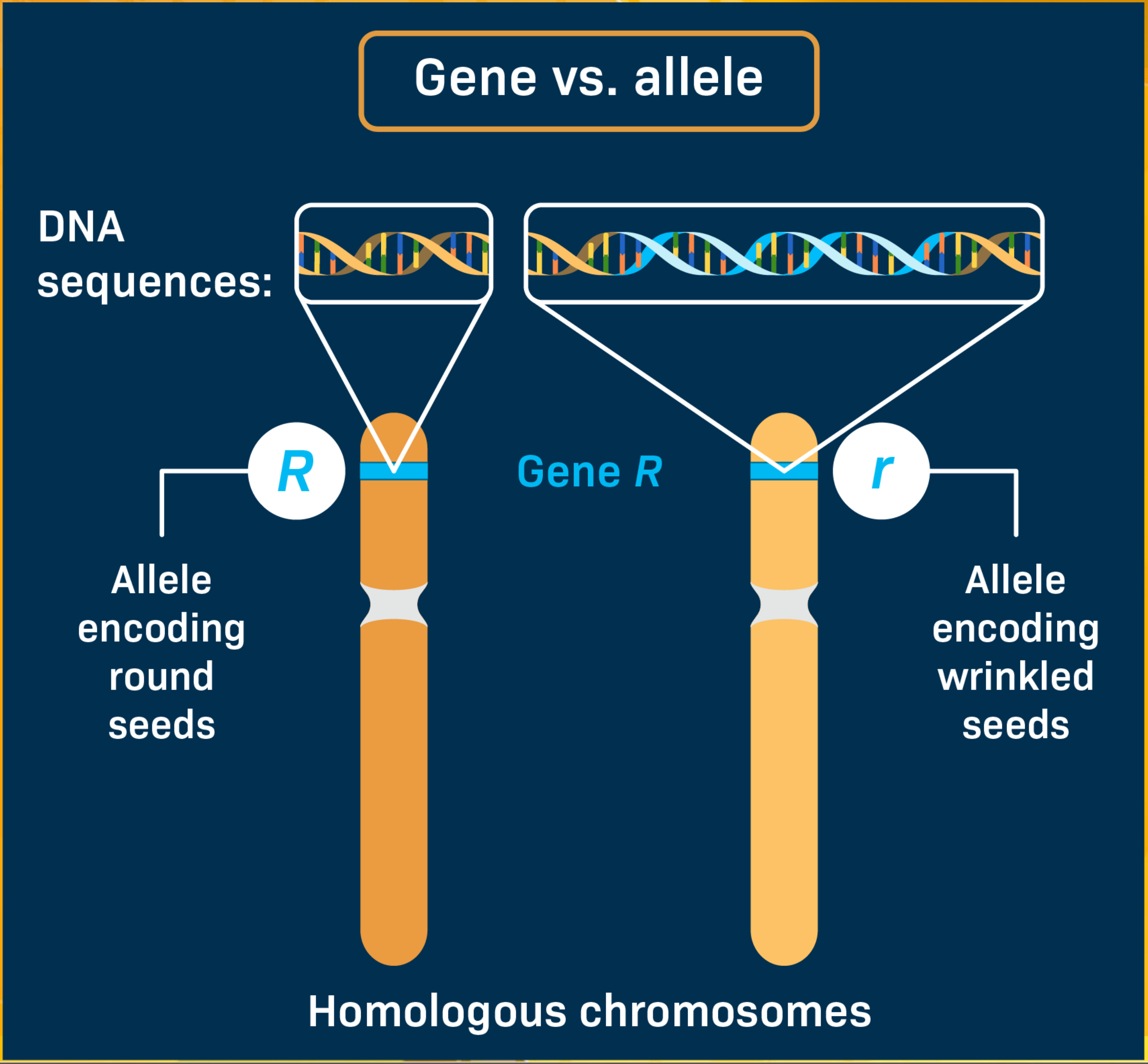
Use the Law of Dominance to determine the phenotypes and phenotype ratio of the offspring.



The phenotype ratio is 3:1, meaning 3 yellow peas to 1 green pea.

Designer "Genes"

- **Alleles** - two forms of a gene (dominant & recessive)
- **Dominant** - stronger of two genes expressed in the hybrid; represented by a capital letter (R)
- **Recessive** - gene that shows up less often in a cross; represented by a lowercase letter (r)


Gene vs. allele
DNA sequences:
R
Gene R
r
Allele encoding round seeds
Allele encoding wrinkled seeds
Homologous chromosomes

More Terminology

- **Genotype** - gene combination for a trait (e.g. RR, Rr, rr)
- **Phenotype** - the physical feature resulting from a genotype (e.g. red, white)

Genotype & Phenotype in Flowers

Genotype of alleles:
R = red flower
r = yellow flower

All genes occur in pairs, so 2 alleles affect a characteristic

Possible combinations are:

Genotypes	RR	Rr	rr
Phenotypes	RED	RED	YELLOW

Genotypes

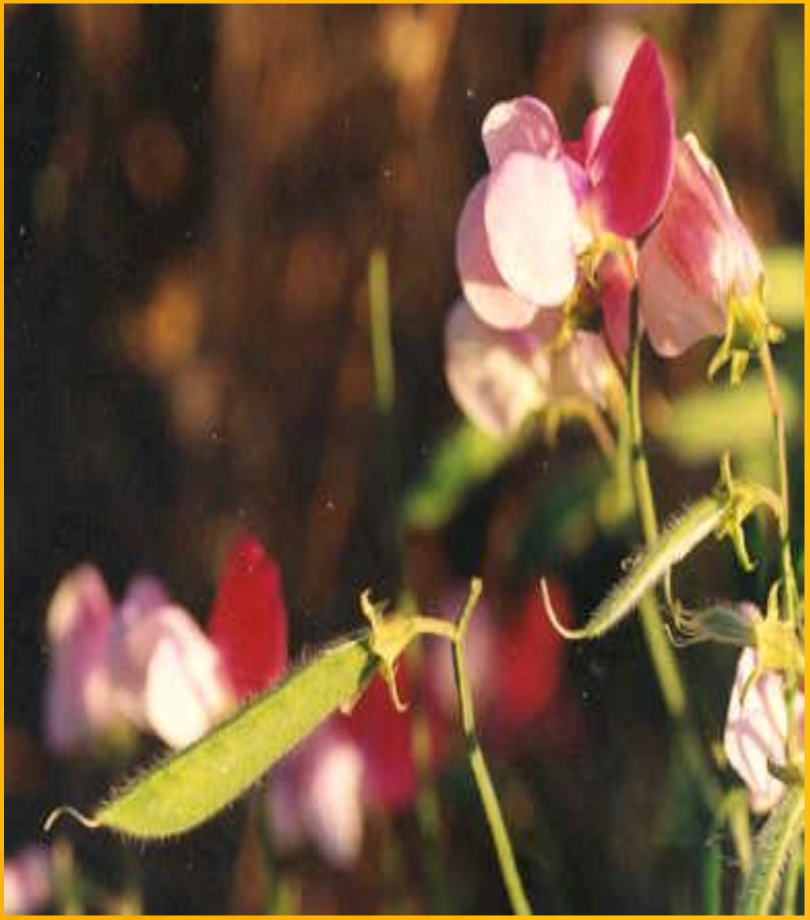
- **Homozygous** genotype - gene combination involving 2 dominant or 2 recessive genes (e.g. RR or rr); also called **pure**
- **Heterozygous** genotype - gene combination of one dominant & one recessive allele (e.g. Rr); also called **hybrid**

Genes and Environment Determine Characteristics

Mendel's Pea Plant Experiments

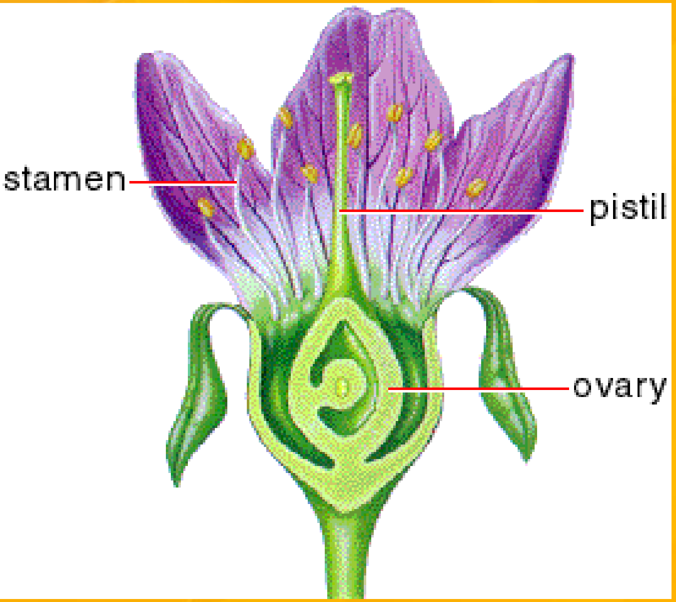
Why peas, *Pisum sativum*?

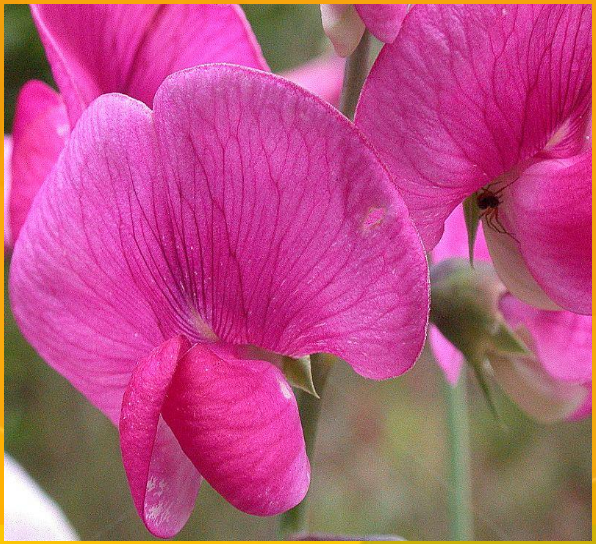
- Can be grown in a small area
- Produce lots of offspring
- Produce pure plants when allowed to self-pollinate several generations
- Can be artificially cross-pollinated



Reproduction in Flowering Plants

- Pollen contains sperm
    - Produced by the stamen
- Ovary contains eggs
    - Found inside the flower





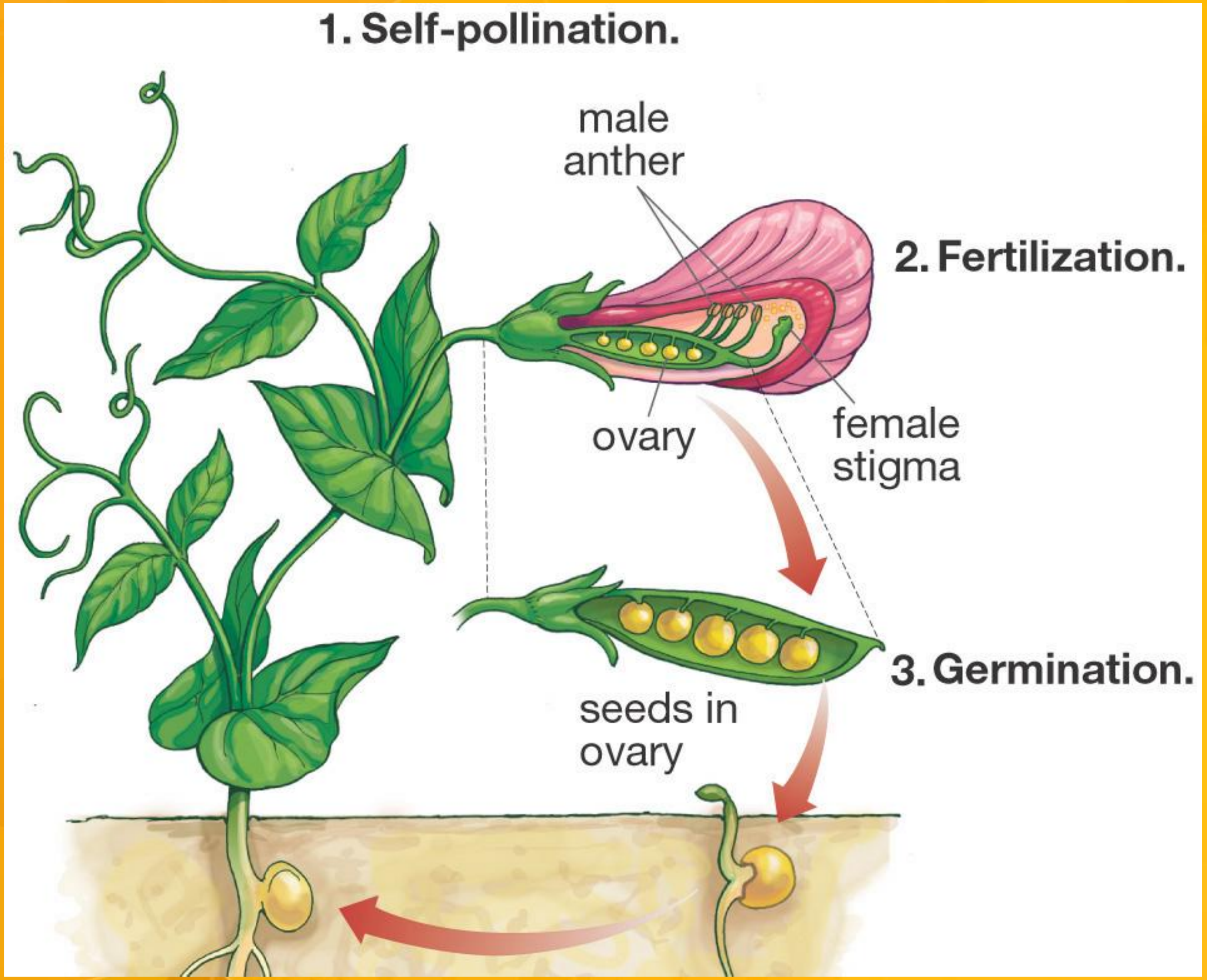
- Pollen carries sperm to the eggs for fertilization
    - *Self-fertilization* can occur in the same flower
    - *Cross-fertilization* can occur between flowers

Mendel's Experimental Methods

- Mendel hand-pollinated flowers using a paintbrush
  - He could snip the stamens to prevent self-pollination
- He traced traits through the several generations

How Mendel Began

Mendel produced **pure** strains by allowing the plants to **self-pollinate** for several generations

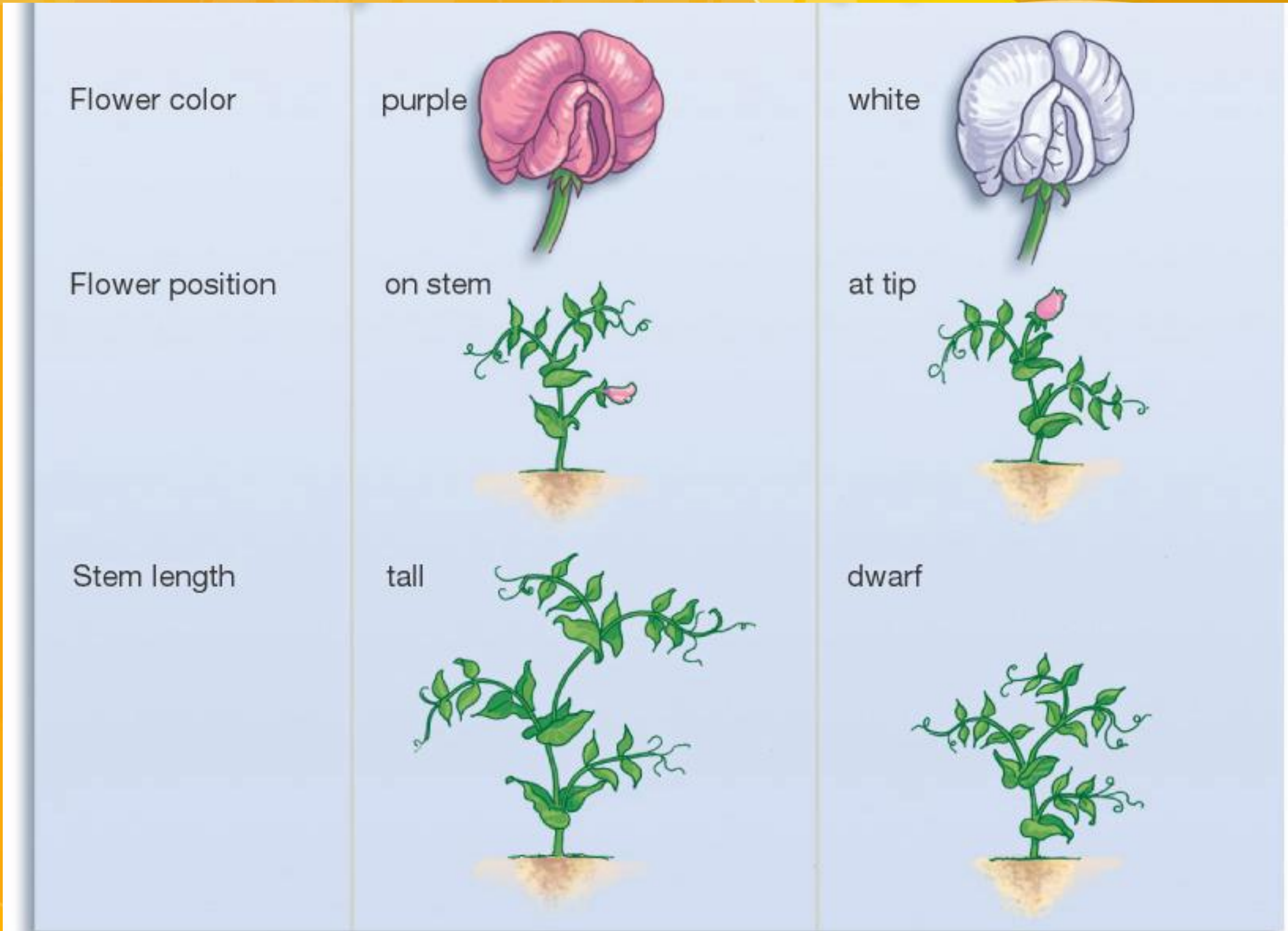


Eight Pea Plant Traits

Seed shape --- Round (R) or Wrinkled (r)
Seed Color ---- Yellow (Y) or Green (y)
Pod Shape --- Smooth (S) or wrinkled (s)
Pod Color --- Green (G) or Yellow (g)
Seed Coat Color ---Gray (G) or White (g)
Flower position---Axial (A) or Terminal (a)
Plant Height --- Tall (T) or Short (t)
Flower color --- Purple (P) or white (p)

Table 11.1 Pea-Plant Characters Studied by Mendel

Character studied	Dominant trait	Recessive trait
Seed shape	smooth 	wrinkled 
Seed color	yellow 	green 
Pod shape	inflated 	wrinkled 
Pod color	green 	yellow 


Flower color
purple
white
Flower position
on stem
at tip
Stem length
tall
dwarf

Mendel's Experimental Results

Table 11.2 Ratios of Dominant to Recessive in Mendel's Plants

Dominant trait	Recessive trait	Ratio of dominant to recessive in F_2 generation
Smooth seed	Wrinkled seed	2.96:1 (5,474 smooth, 1,850 wrinkled)
Yellow seed	Green seed	3.01:1 (6,022 yellow, 2,001 green)
Inflated pod	Wrinkled pod	2.95:1 (882 inflated, 299 wrinkled)
Green pod	Yellow pod	2.82:1 (428 green, 152 yellow)
Purple flower	White flower	3.14:1 (705 purple, 224 white)
Flower on stem	Flower at tip	3.14:1 (651 along stem, 207 at tip)
Tall stem	Dwarf stem	2.84:1 (787 tall plants, 277 dwarfs)
	Average ratio, all traits:	**3:1**

Did the observed ratio match the theoretical ratio?

The theoretical or expected ratio of plants producing round or wrinkled seeds is **3 round :1 wrinkled**

Mendel's observed ratio was 2.96:1

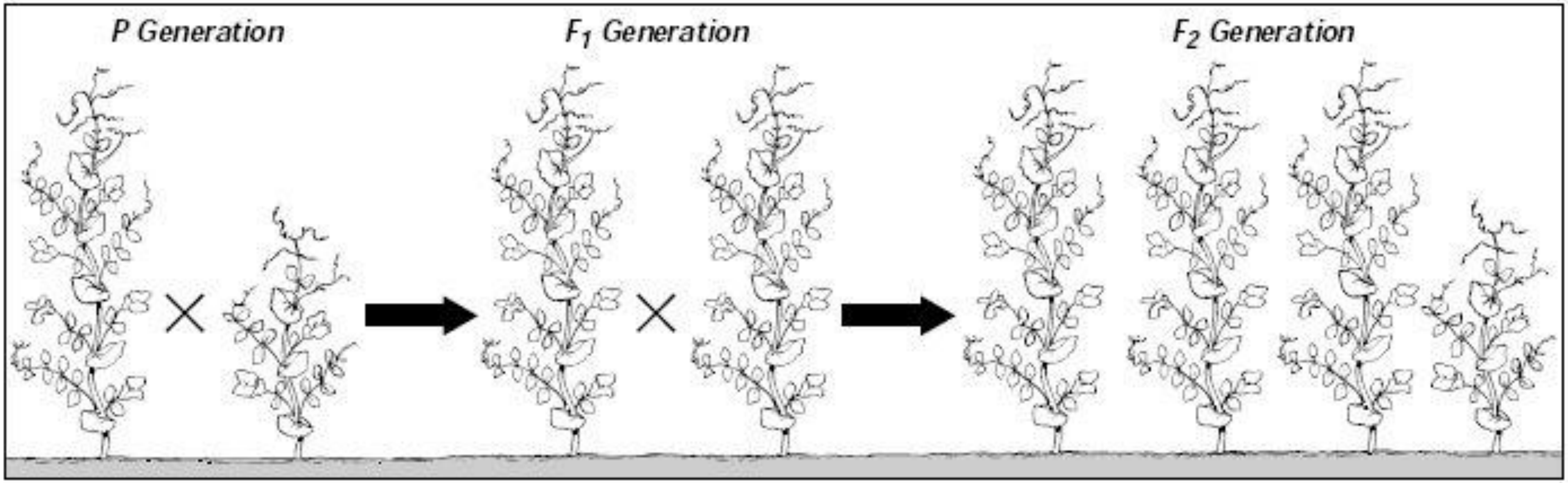
The discrepancy is due to **statistical error**

The **larger the sample** the more nearly the results approximate to the theoretical ratio

Generation "Gap"

- Parental P_1 Generation = the parental generation in a breeding experiment.
- F_1 generation = the first-generation offspring in a breeding experiment. (1st filial generation)
  - From breeding individuals from the P_1 generation
- F_2 generation = the second-generation offspring in a breeding experiment. (2nd filial generation)
  - From breeding individuals from the F_1 generation

Following the Generations



Cross 2 Pure Plants	Results in all Hybrids	Cross 2 Hybrids get 3 Tall & 1 Short
TT x tt	Tt	TT, Tt, tt

Monohybrid Crosses

P_1 Monohybrid Cross

Trait: Seed Shape

Alleles: R – Round r – Wrinkled

Cross: Round seeds x Wrinkled seeds

RR x rr

	r	r
R	Rr	Rr
R	Rr	Rr

Genotype: Rr

Phenotype: Round

Genotypic Ratio: All alike

Phenotypic Ratio: All alike

P_1 Monohybrid Cross Review

- Homozygous dominant x Homozygous recessive
- Offspring all Heterozygous (hybrids)
- Offspring called F_1 generation
- Genotypic & Phenotypic ratio is ALL ALIKE

F_1 Monohybrid Cross

Trait: Seed Shape

Alleles: R - Round r - Wrinkled

Cross: Round seeds x Round seeds

Rr x Rr

	R	r
R	RR	Rr
r	Rr	rr

Genotype: RR, Rr, rr

Phenotype: Round & wrinkled

G.Ratio: 1:2:1

P.Ratio: 3:1

F_1 Monohybrid Cross Review

- Heterozygous x heterozygous
- Offspring:
  25% Homozygous dominant RR
  50% Heterozygous Rr
  25% Homozygous Recessive rr
- Offspring called F_2 generation
- Genotypic ratio is 1:2:1
- Phenotypic Ratio is 3:1

What Do the Peas Look Like?

…And Now the Test Cross

- Mendel then crossed a pure & a hybrid from his F_2 generation
- This is known as an F_2 or test cross
- There are two possible testcrosses:
  - Homozygous dominant x Hybrid
  - Homozygous recessive x Hybrid

F_2 Monohybrid Cross (1^{st})

Trait: Seed Shape

Alleles: R – Round   r – Wrinkled

Cross: Round seeds x Round seeds

RR x Rr

	R	r
R	RR	Rr
R	RR	Rr

Genotype: RR, Rr

Phenotype: Round

Genotypic Ratio: 1:1

Phenotypic Ratio: All alike

F_2 Monohybrid Cross (2nd)

Trait: Seed Shape

Alleles: R – Round   r – Wrinkled

Cross: Wrinkled seeds x Round seeds

rr x Rr

	R	r
r	Rr	rr
r	Rr	rr

Genotype: Rr, rr

Phenotype: Round & Wrinkled

G. Ratio: 1:1

P.Ratio: 1:1

F_2 Monohybrid Cross Review

- Homozygous x heterozygous(hybrid)
- Offspring:
  50% Homozygous RR or rr
  50% Heterozygous Rr
- Phenotypic Ratio is 1:1
- Called Test Cross because the offspring have SAME genotype as parents

Practice Your Crosses

Work the P_1, F_1, and both F_2 Crosses for each of the other Seven Pea Plant Traits

Mendel's Laws

Results of Monohybrid Crosses

- Inheritable factors or genes are responsible for all heritable characteristics
- Phenotype is based on Genotype
- Each trait is based on two genes, one from the mother and the other from the father
- True-breeding individuals are homozygous (both alleles) are the same

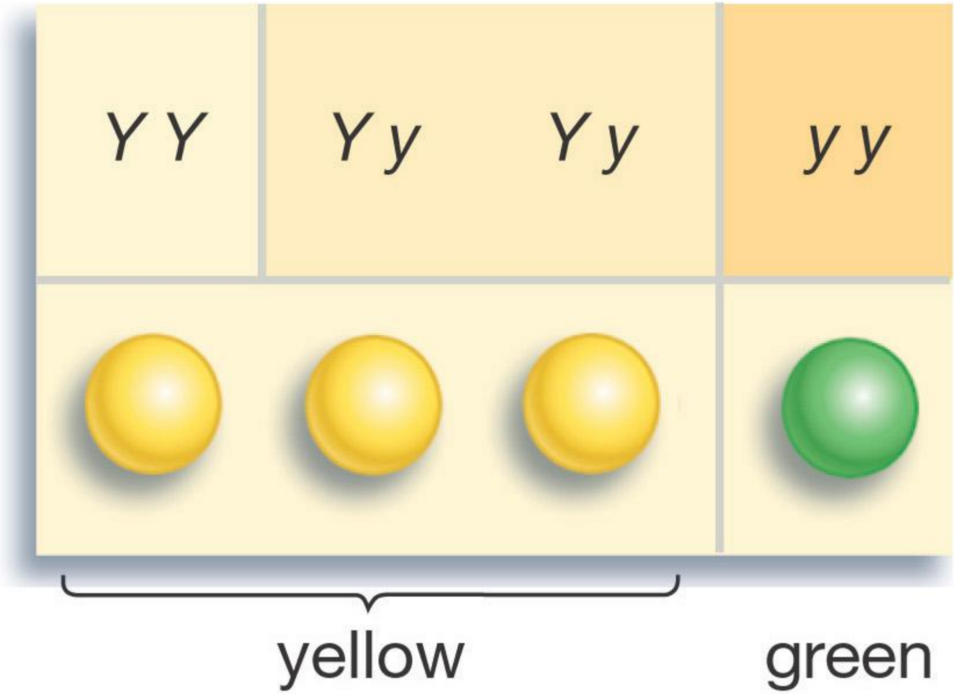
Law of Dominance

In a cross of parents that are **pure for contrasting traits**, only one form of the trait will appear in the next generation.

All the offspring will be heterozygous and express only the **dominant trait**.

RR x rr yields all Rr (round seeds)

Law of Dominance



Three genotypes yield . . .

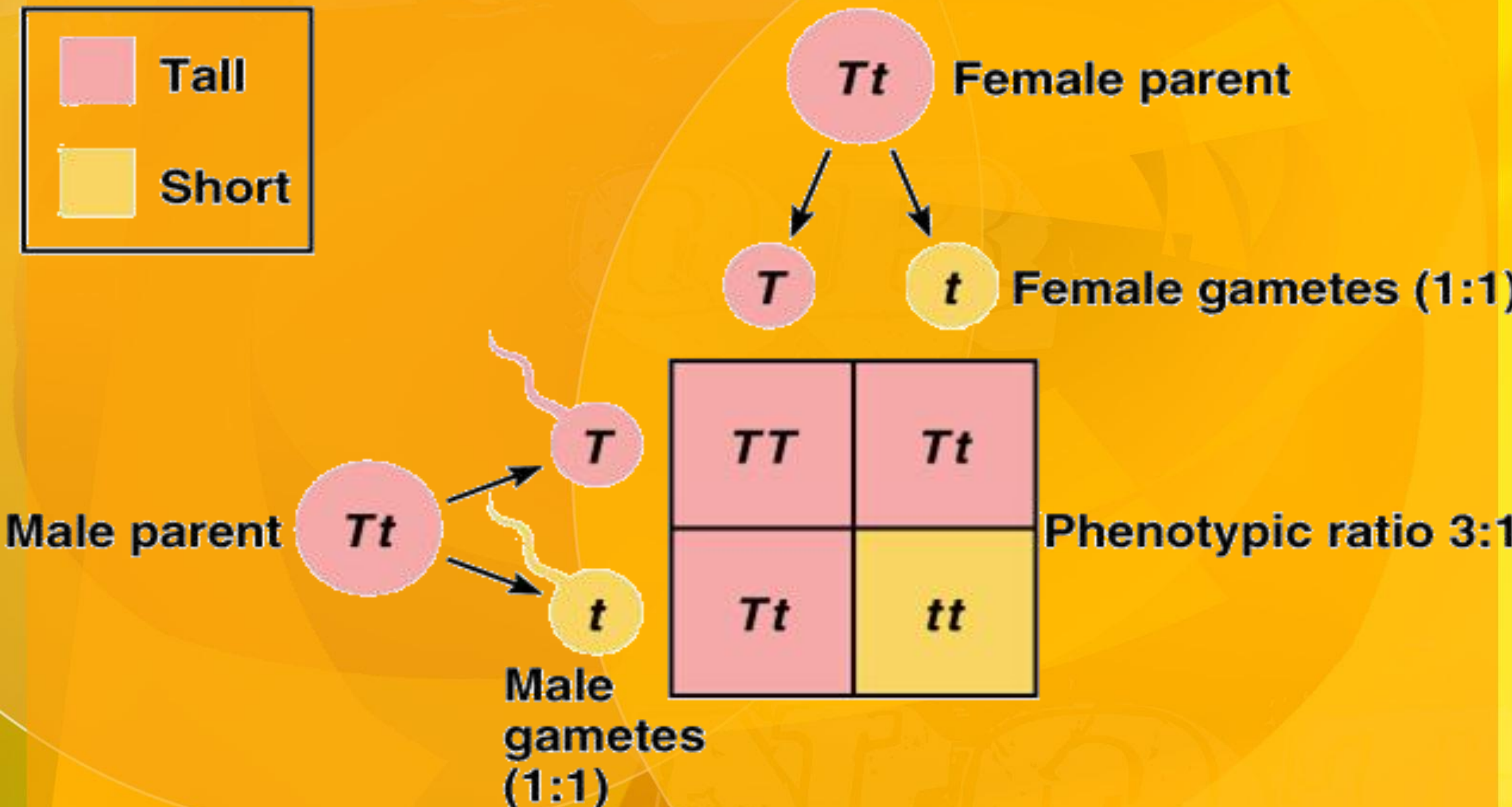
two phenotypes.

Law of Segregation

During the formation of gametes (eggs or sperm), the two alleles responsible for a trait separate from each other.

Alleles for a trait are then "recombined" at fertilization, producing the genotype for the traits of the offspring.

Applying the Law of Segregation



Law of Independent Assortment

- Alleles for *different* traits are distributed to sex cells (& offspring) independently of one another.
- This law can be illustrated using *dihybrid crosses*.

Dihybrid Cross

A breeding experiment that tracks the inheritance of two traits.

Mendel's "Law of Independent Assortment"

a. Each pair of alleles segregates independently during gamete formation

b. Formula: 2^n (n = # of heterozygotes)

Question:

How many gametes will be produced for the following allele arrangements?

Remember: 2^n (n = # of heterozygotes)

1. RrYy

2. AaBbCCDd

3. MmNnOoPPQQRrssTtQq

Answer:

1. RrYy: $2^n = 2^2 = 4$ gametes

   RY Ry rY ry

2. AaBbCCDd: $2^n = 2^3 = 8$ gametes

   ABCD ABCd AbCD AbCd

   aBCD aBCd abCD abCD

3. MmNnOoPPQQRrssTtQq: $2^n = 2^6 = 64$ gametes

Dihybrid Cross

Traits: Seed shape & Seed color

Alleles: R round
r wrinkled
Y yellow
y green

RrYy x RrYy

RY Ry rY ry

RY Ry rY ry

All possible gamete combinations

Dihybrid Cross

	RY	Ry	rY	ry
RY				
Ry				
rY				
ry				

Dihybrid Cross

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

Round/Yellow: 9

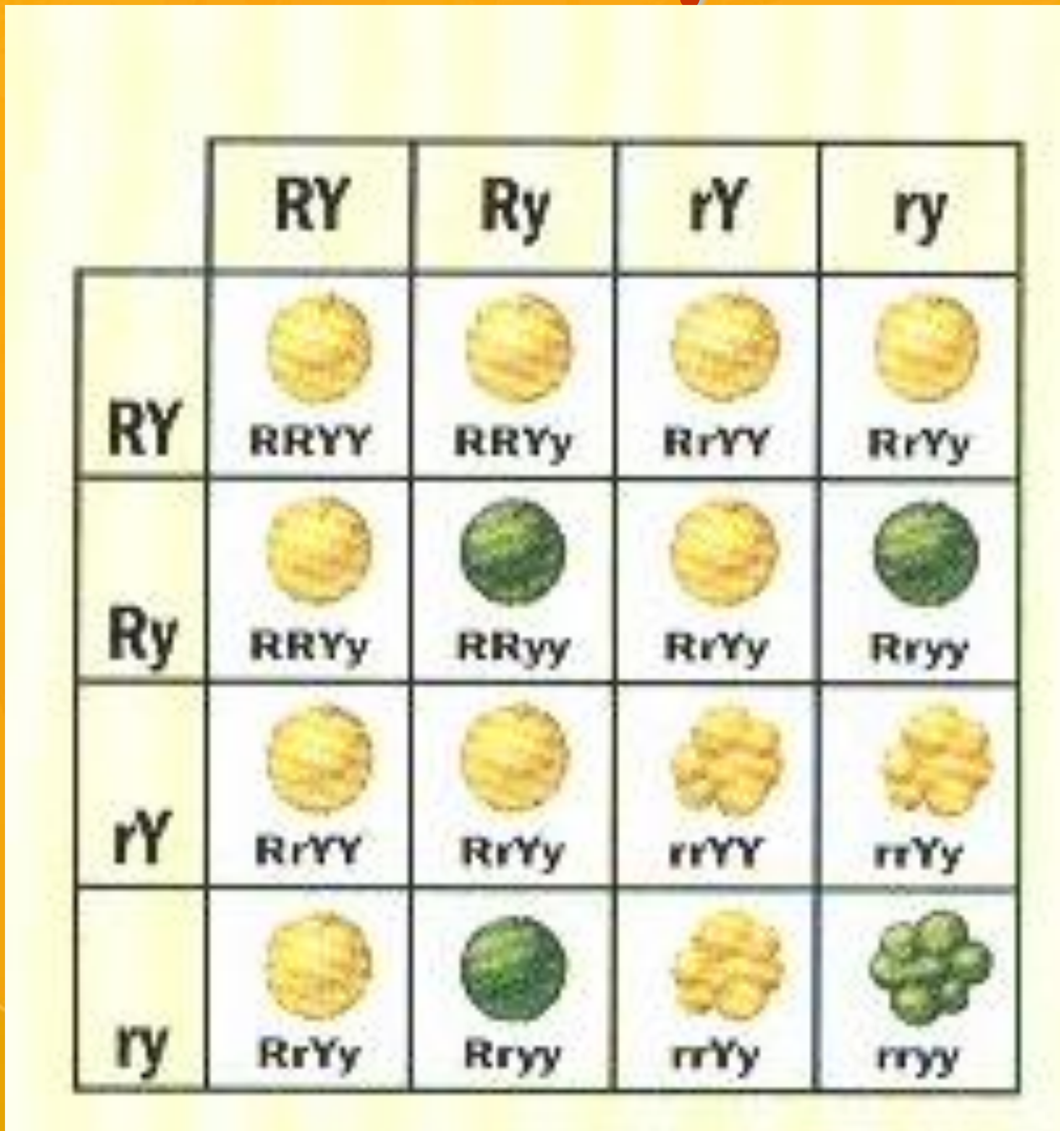
Round/green: 3

wrinkled/Yellow: 3

wrinkled/green: 1

9:3:3:1 phenotypic ratio

Dihybrid Cross



Round/Yellow: 9
Round/green: 3
wrinkled/Yellow: 3
wrinkled/green: 1

9:3:3:1

Test Cross

A mating between an individual of unknown genotype and a homozygous recessive individual.

Example: bbC__ x bbcc

BB = brown eyes

Bb = brown eyes

bb = blue eyes

CC = curly hair

Cc = curly hair

cc = straight hair

	bC	b___
bc		

Test Cross

Possible results:

	bC	b_C_
bc	bbCc	bbCc

or

	bC	b_c_
bc	bbCc	bbcc

Summary of Mendel's laws

LAW	PARENT CROSS	OFFSPRING
DOMINANCE	TT x tt tall x short	100% Tt tall
SEGREGATION	Tt x Tt tall x tall	75% tall 25% short
INDEPENDENT ASSORTMENT	RrGg x RrGg round & green x round & green	9/16 round seeds & green pods 3/16 round seeds & yellow pods 3/16 wrinkled seeds & green pods 1/16 wrinkled seeds & yellow pods

Incomplete Dominance and Codominance

Incomplete Dominance

F1 hybrids have an appearance somewhat **in between** the **phenotypes** of the two parental varieties.

Example: snapdragons (flower)

red (RR) x white (rr)

	r	r
R		
R		

RR = red flower

rr = white flower

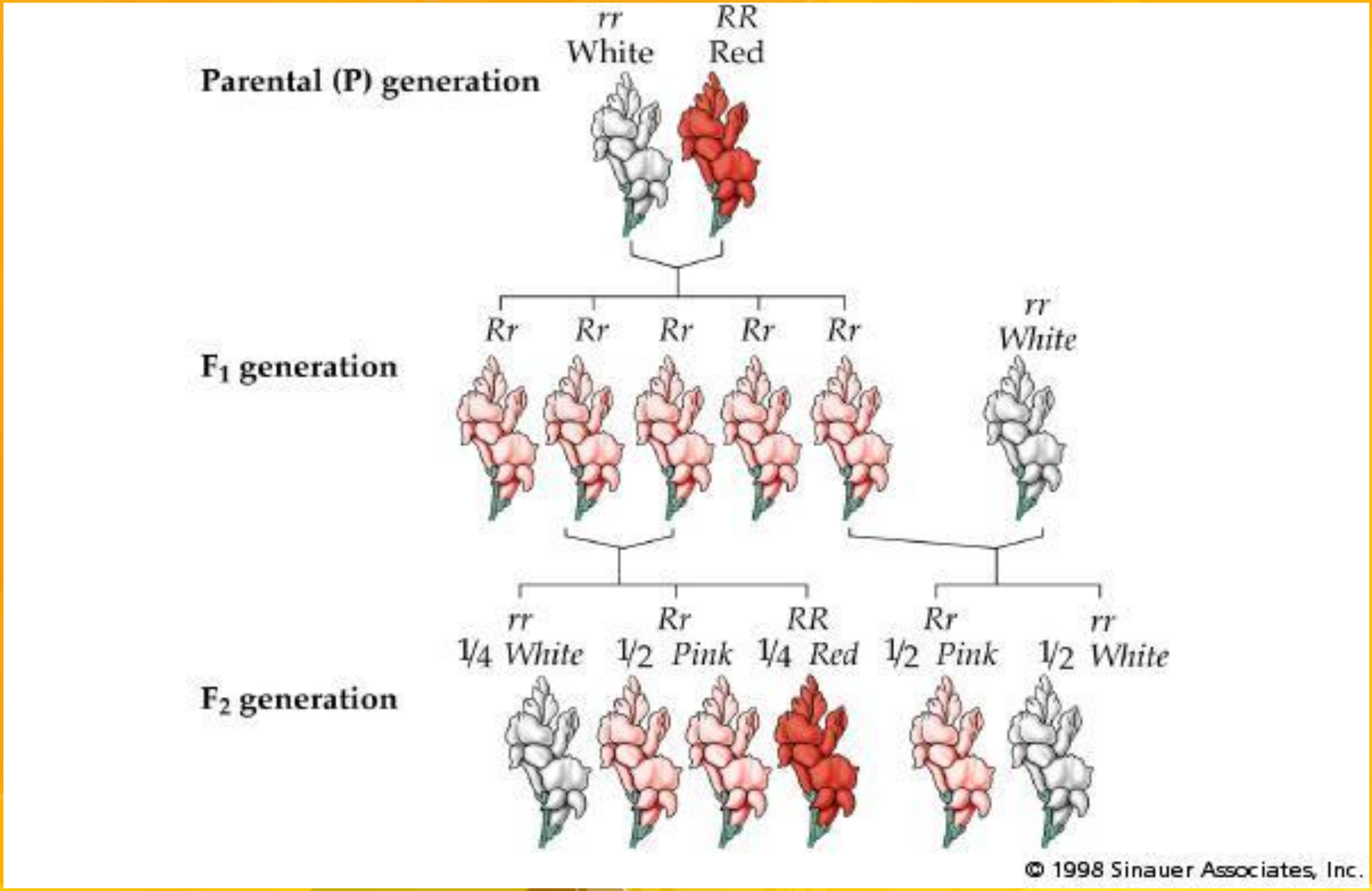
Incomplete Dominance

		r
R	Rr	Rr
R	Rr	Rr

produces the F_1 generation

All Rr = pink (heterozygous pink)

Incomplete Dominance



Codominance

Two alleles are expressed (multiple alleles) in heterozygous individuals.

Example: blood type

1. type A = I^AI^A or I^Ai
2. type B = I^BI^B or I^Bi
3. type AB = I^AI^B
4. type O = ii

Codominance Problem

Example: homozygous male Type B (I^BI^B)

x

heterozygous female Type A (I^Ai)

	I^A	i
I^B	I^AI^B	I^Bi
I^B	I^AI^B	I^Bi

$1/2 = I^AI^B$

$1/2 = I^Bi$

Another Codominance Problem

- **Example:** male Type O (ii)

  x

  female type AB (I^AI^B)

	I^A	I^B
i	I^Ai	I^Bi
i	I^Ai	I^Bi

1/2 = I^Ai

1/2 = I^Bi

Codominance

Question:

If a boy has a blood type O and his sister has blood type AB, **what are the genotypes and phenotypes of their parents?**

boy - type O (ii) X girl - type AB (I^AI^B)

Codominance

Answer:

	I^A	i
I^B	I^AI^B	
i		ii

Parents:

genotypes = I^Ai and I^Bi

phenotypes = A and B

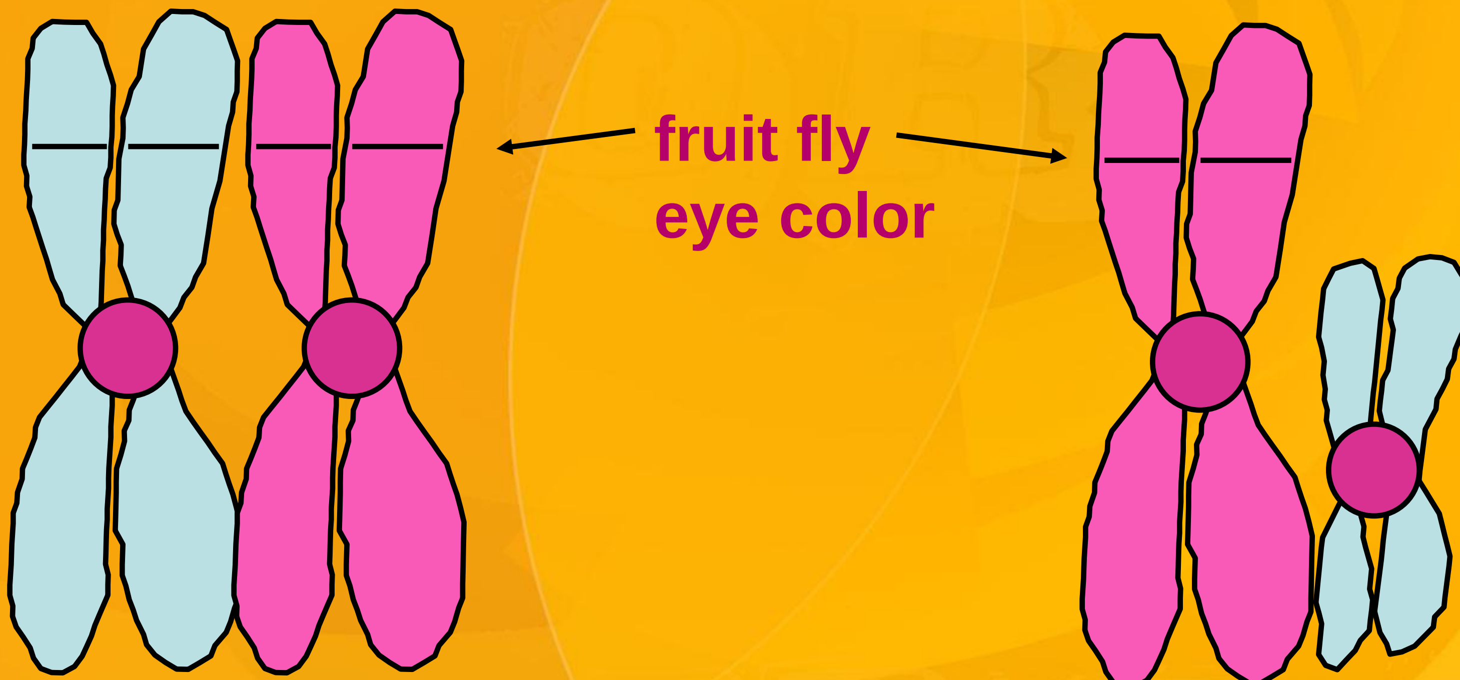
Sex-linked Traits

- Traits (genes) located on the sex chromosomes
- Sex chromosomes are X and Y
- XX genotype for females
- XY genotype for males
- Many sex-linked traits carried on X chromosome

Sex-linked Traits

Example: Eye color in fruit flies

Sex Chromosomes



XX chromosome - female

Xy chromosome - male

Sex-linked Trait Problem

Example: Eye color in fruit flies

(red-eyed male) x (white-eyed female)

X^rY x X^rX^r

Remember: the Y chromosome in males does not carry traits.

RR = red eyed

Rr = red eyed

rr = white eyed

Xy = male

XX = female

	X^r	X^r
X^r		
Y		

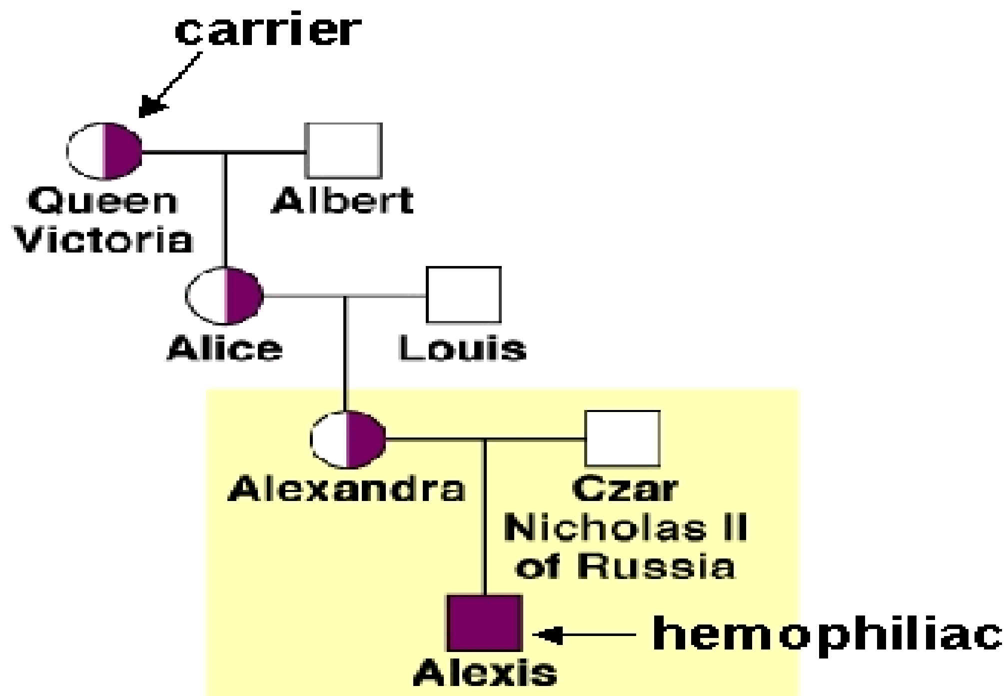
Sex-linked Trait Solution:

	X^r	X^r
X^R	$X^R X^r$	$X^R X^r$
Y	$X^r Y$	$X^r Y$

50% red eyed
female
50% white eyed
male

Female Carriers

In a sex-linked trait (like hemophilia), women are carriers, and men have the phenotype more often.



Genetic Practice Problems

	T	t
T	TT	Tt
t	Tt	tt

Breed the P_1 generation

tall (TT) x dwarf (tt) pea plants

	t	t
T		
T		

Solution:

tall (TT) vs. dwarf (tt) pea plants

	t	t
T	Tt	Tt
T	Tt	Tt

produces the
F_1 generation

All Tt = tall
(heterozygous tall)

Breed the F_1 generation

tall (Tt) vs. tall (Tt) pea plants

	T	t
T		
t		

Solution:

tall (Tt) x tall (Tt) pea plants

	T	t
T	TT	Tt
t	Tt	tt

produces the F_2 generation

1/4 (25%) = TT
1/2 (50%) = Tt
1/4 (25%) = tt

1:2:1 genotype
3:1 phenotype