Lecture 1: Mendelian Genetics

Patterns and Principles of Heredity

Outline of Mendelian Genetics

- Mendel's approach to genetic analysis including his experiments and related analytic tools
- Mendel's two explanations (Laws) for the inheritance patterns of pea traits
- Several key concepts and terminology

  (gene, allele, phenotype and genotype, probability, Punnet square, test cross, chi-square test)

Fig. 2.3

The historical puzzle of inheritance

- Artificial selection has been an important practice since before recorded history
  - Domestication of animals
  - Selective breeding of plants
- 19^{th} century – precise techniques for controlled matings in plants and animals to produce desired traits in many of offspring
- Breeders could not explain why traits would sometimes disappear and then reappear in subsequent generations.

Historical theories of inheritance

- One parent contributes most features (e.g., homunculus, N. Hartsoiker, 1694)
- Blending inheritance – parental traits become mixed and forever changed in offspring

Gregor Mendel (1822-1844)

State of genetics in early 1800's

What is inherited?

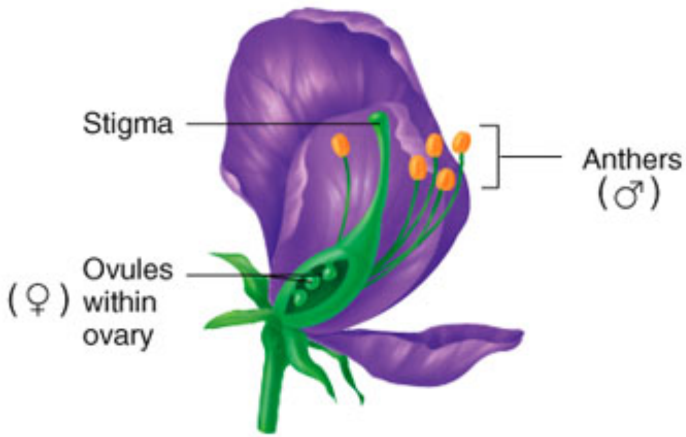
How is it inherited?

What is the role of chance in heredity?

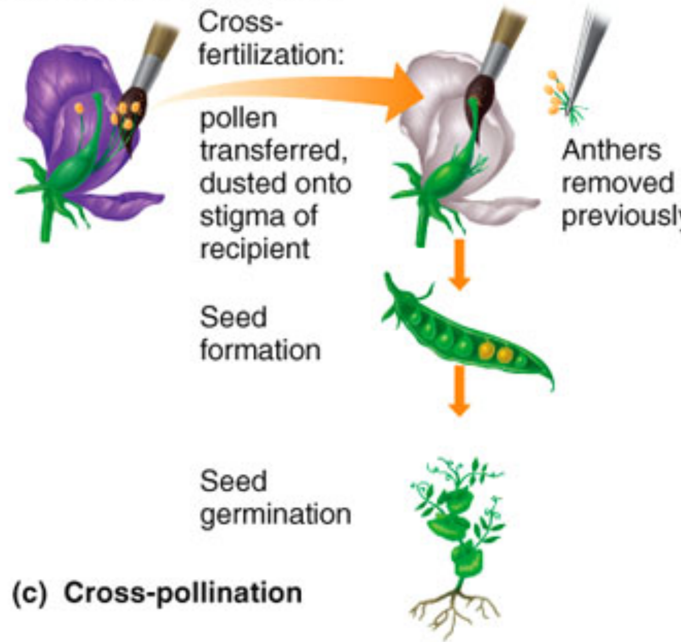
Keys to Mendel's experiments

- The garden pea was an ideal organism
  - Vigorous growth
  - Self fertilization
  - Easy to cross fertilize
  - Produced large number of offspring each generation
- Mendel analyzed traits with discrete alternative forms
  - purple vs. white flowers
  - yellow vs. green peas
  - round vs. wrinkled seeds
  - long vs. short stem length
- Mendel established pure-breeding lines to conduct his experiments

(a) *Pisum sativum*



(b) Pea flower anatomy



(c) Cross-pollination

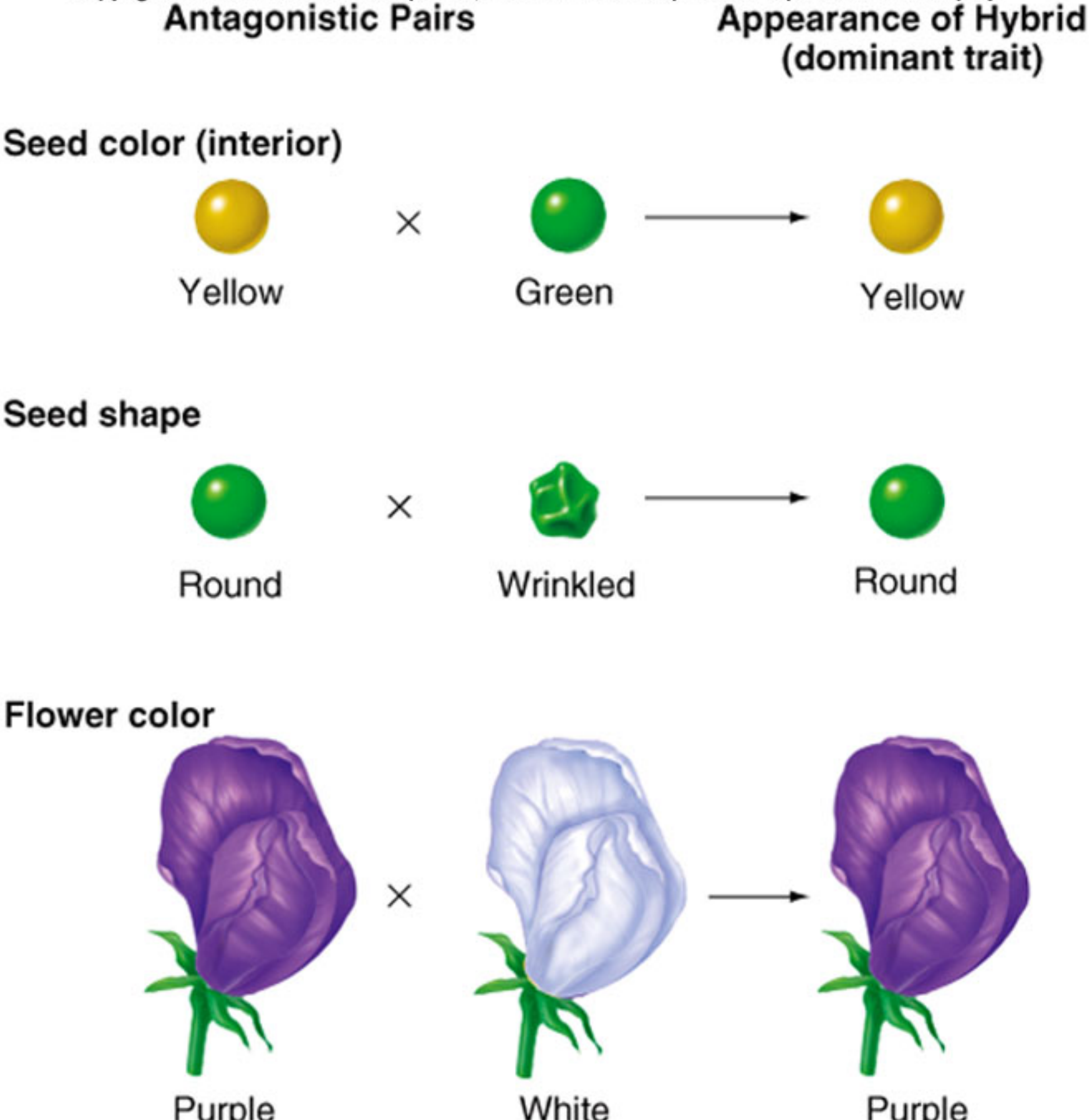
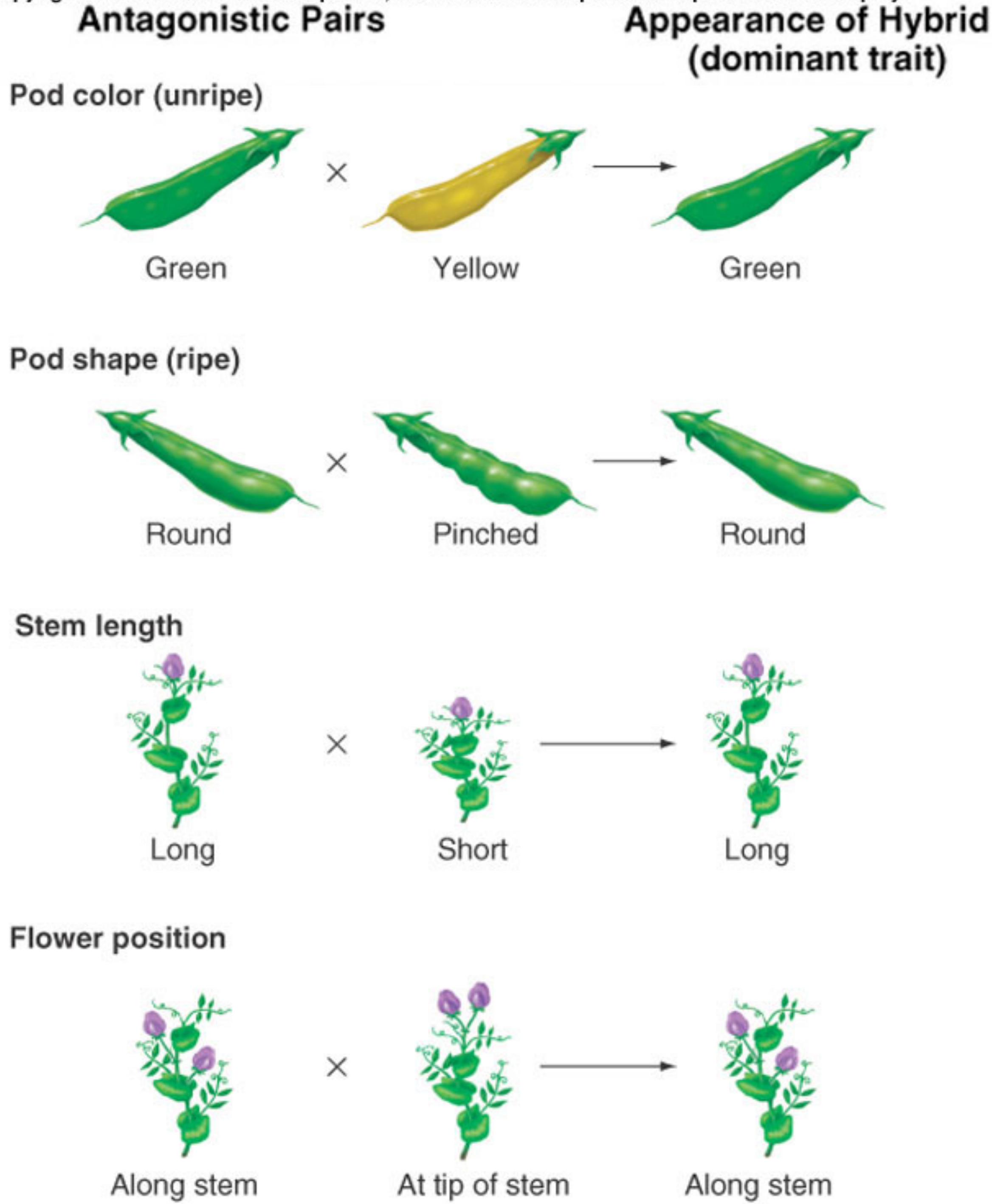
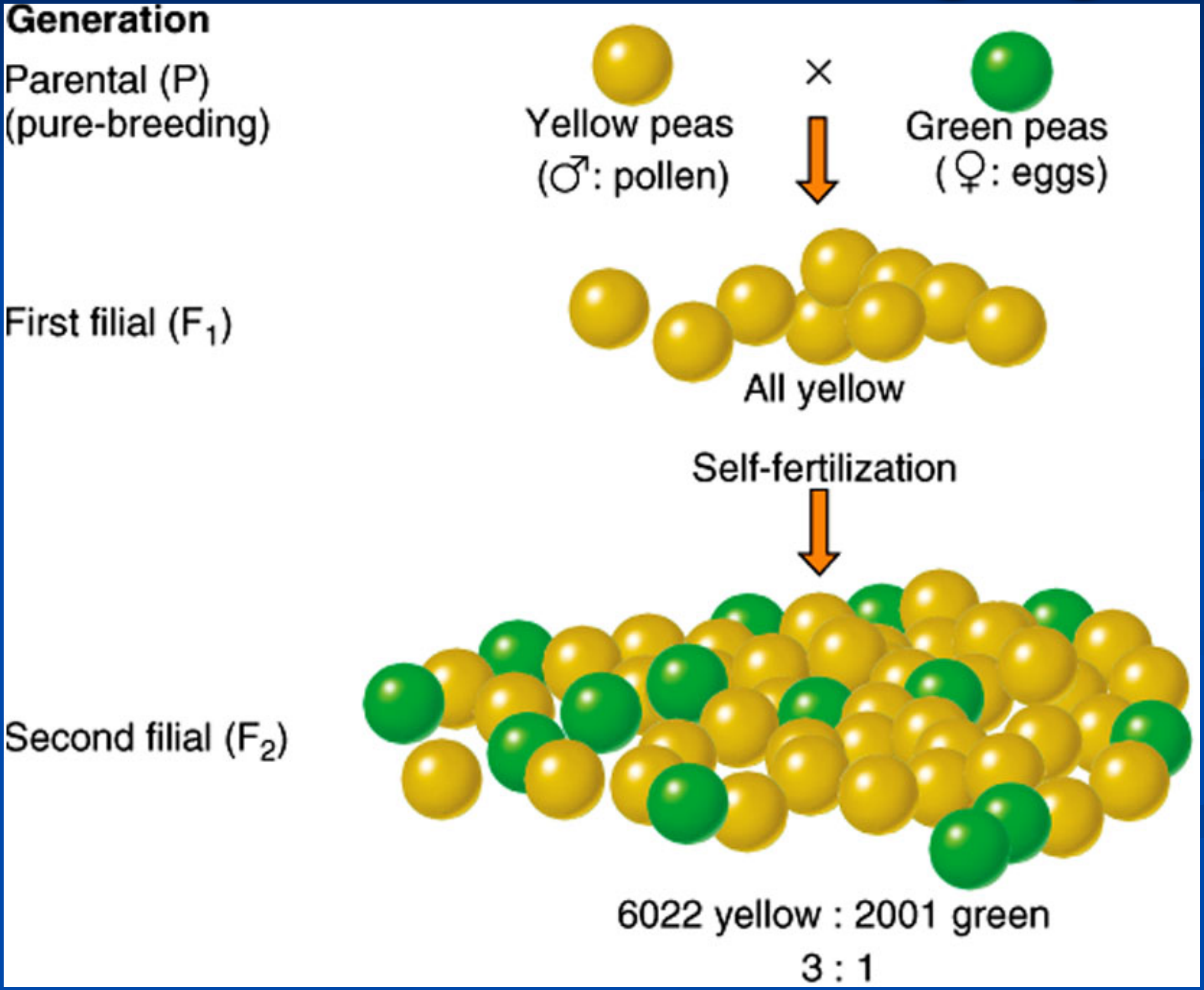

Antagonistic Pairs
Appearance of Hybrid
(dominant trait)
Seed color (interior)
Yellow
×
Green
Yellow
Seed shape
Round
×
Wrinkled
Round
Flower color
×
Purple
White
Purple

Fig. 2.8b


Antagonistic Pairs
Appearance of Hybrid
(dominant trait)
Pod color (unripe)
Green
Yellow
Green
Pod shape (ripe)
Round
Pinched
Round
Stem length
Long
Short
Long
Flower position
Along stem
At tip of stem
Along stem

Monohybrid crosses reveal units of inheritance and Law of Segregation



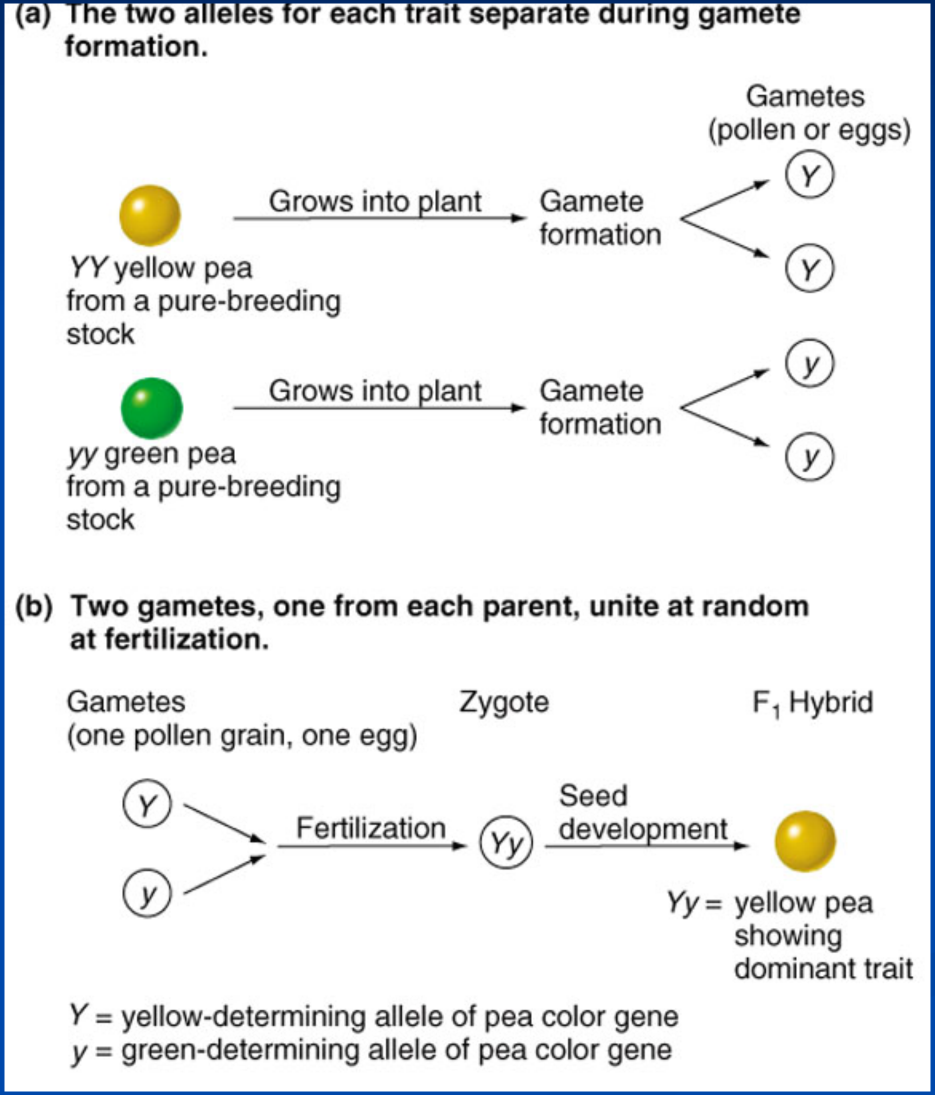
Traits have dominant and recessive forms

- Disappearance of traits in F1 generation and reappearance in the F2 generation disproves the hypothesis that traits blend
- Trait must have two forms
- One form must be hidden when plants with each trait are interbred
- Trait that appears in F1 is dominant
- Trait that is hidden in F1 is recessive

Alternative forms of traits (genes) are alleles

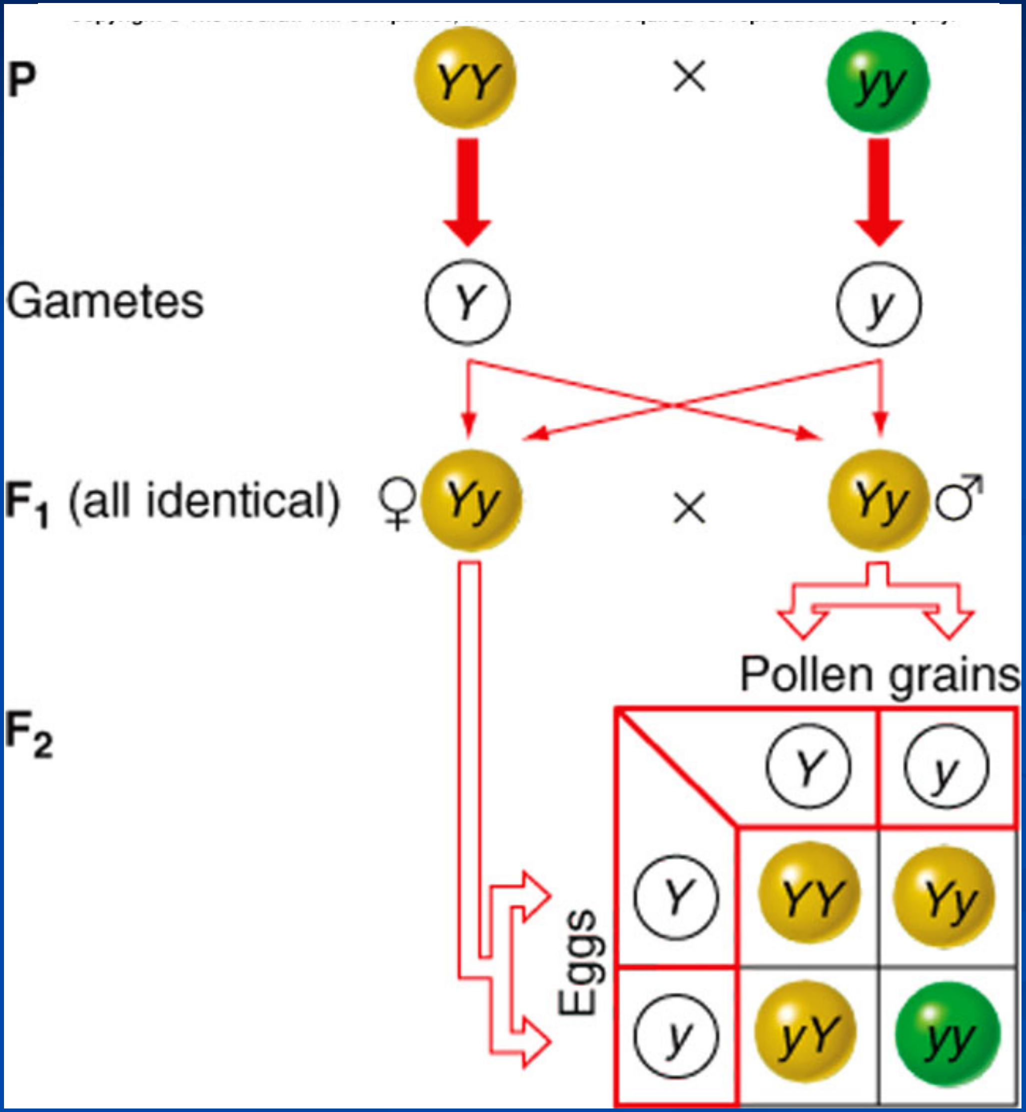
- Each trait carries two copies of a unit of inheritance, one inherited from the mother and the other from the father
- Alternative forms of traits are called alleles

Law of Segregation



- Two alleles for each trait separate (segregate) during gamete formation, and then unite at random, one from each parent, at fertilization

The Punnet Square



Rules of Probability

Independent events - probability of two events occurring together

Coin toss (Penny and Nickel : Head and Tail)
What is the probability that **both** P **and** N will have Head?
Solution = determine probability of each and **multiply** them together. (= 1/2 x 1/2 = 1/4)

Mutually exclusive events - probability of one or another event occurring.

What is the probability of at least one coin has Head?
Solution = determine the probability of each and **add** them together. (= HT : HH : TH : TT = 1/4:1/4:1/4:1/4)
= 1/4+1/4+1/4= 3/4

Probability and Mendel's Results

- Cross Yy x Yy pea plants.
  - Chance of Y sperm uniting with a Y egg
    - ½ chance of sperm with Y allele
    - ½ chance of egg with Y allele
    - Chance of Y and Y uniting = ½ x ½ = ¼
  - Chance of Yy offspring
    - ½ chance of sperm with y allele and egg with Y allele
    - ½ chance of sperm with Y allele and egg with y allele
    - Chance of Yy (½ x ½) + (½ x ½) = 2/4, or 1/2

What about the 3:1 ratio of F2 offspring?

Pollen / Egg

Egg \ Pollen	1/2 Y	1/2 y
1/2 Y	1/4 Y Y	1/4 Y y
1/2 y	1/4 y Y	1/4 y y

F2 ratio of each type

= 1/4 YY : 1/4 Yy or yY : 1/4 yy
= 1/4 YY : 1/2 Yy : 1/4 yy
= 1 : 2 : 1

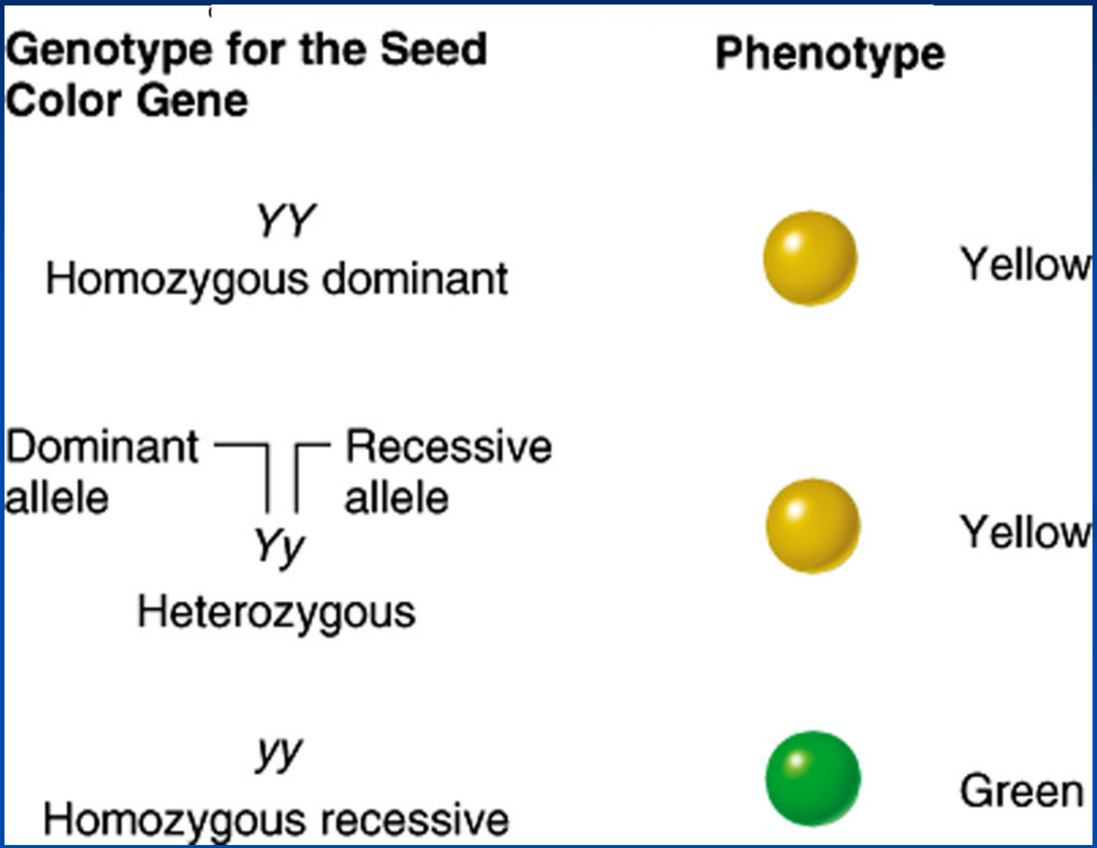
F2 ratio of Yellow vs Green

= 1/4 YY : 1/2 Yy : 1/4 yy
= 3/4 Y_ : 1/4 yy
= 3 : 1

Genotypes and Phenotypes

- Phenotype – observable characteristic of an organism
- Genotype – pair of alleles present in an individual
- Homozygous – two alleles of trait are the same (YY or yy)
- Heterozygous – two alleles of trait are different (Yy)

Genotypes versus phenotypes



Yy × Yy

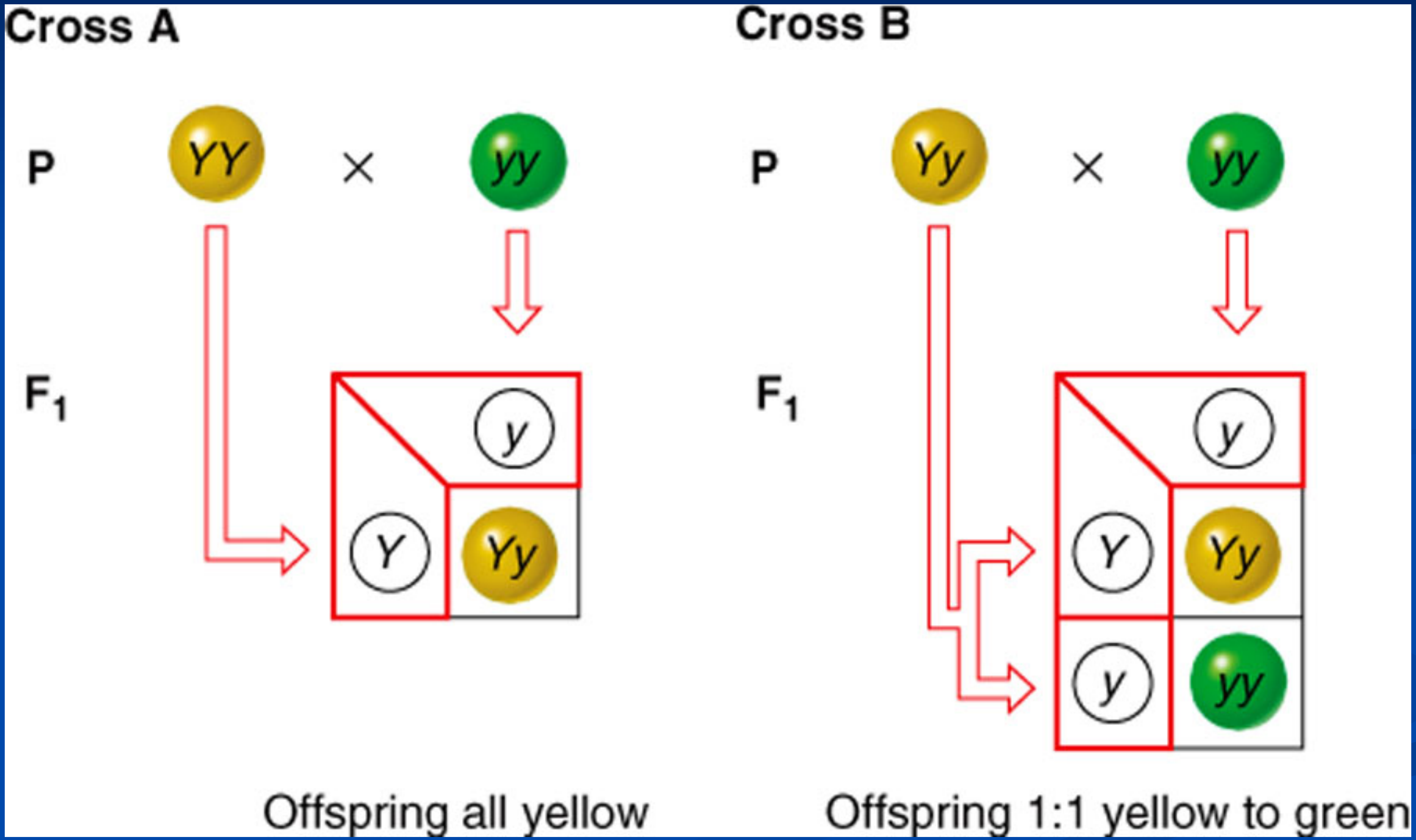
1 : 2 : 1

YY : Yy : yy

3 : 1

Yellow : green

Test cross reveals unknown genotype



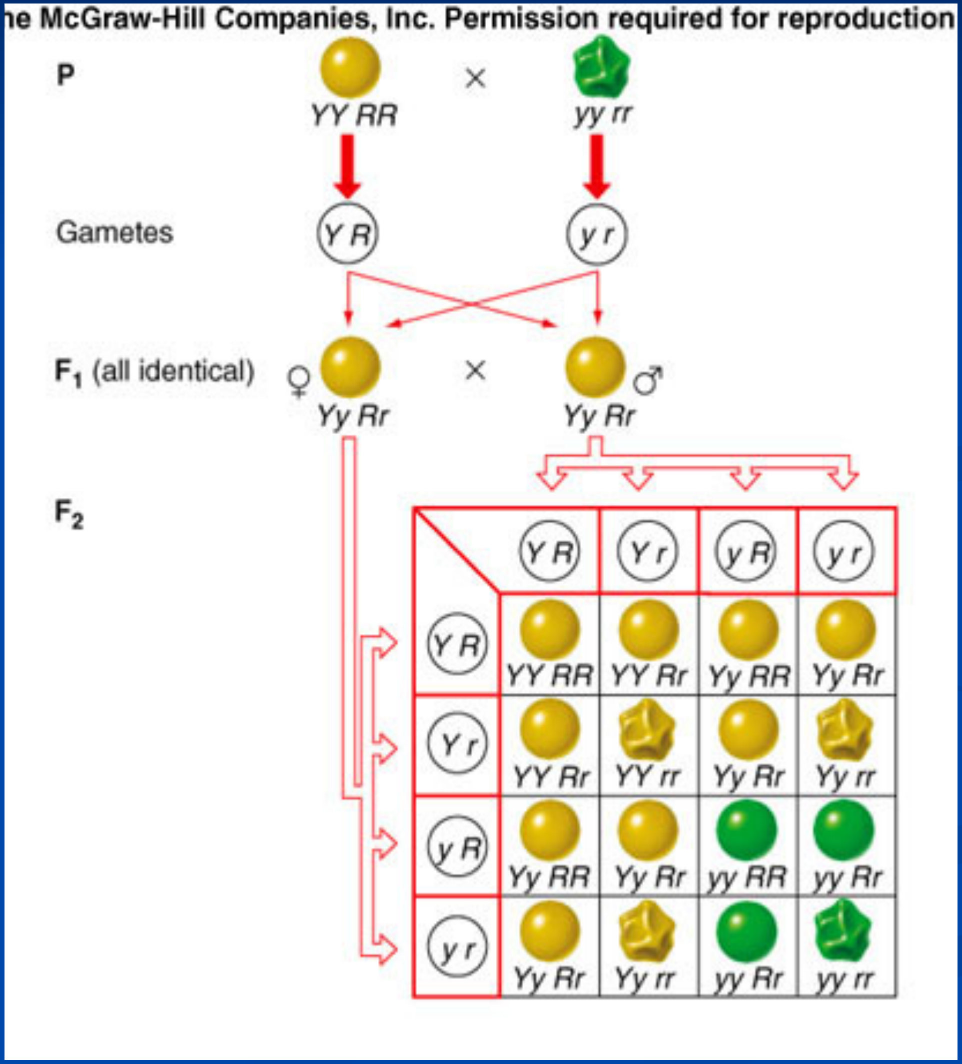
Dihybrid crosses reveal the law of independent assortment

- A dihybrid is an individual that is heterozygous at two genes
- Mendel designed experiments to determine if two genes segregate independently of one another in dihybrids
- First constructed true breeding lines for both traits, crossed them to produce dihybrid offspring, and examined the F2 for parental or recombinant types (new combinations not present in the parents)

Results of Mendel's dihybrid crosses

- F2 generation contained both parental types and recombinant types
- Alleles of genes assort independently, and can thus appear in any combination in the offspring

Dihybrid cross shows parental and recombinant types



Dihybrid cross produces a predictable ratio of phenotypes

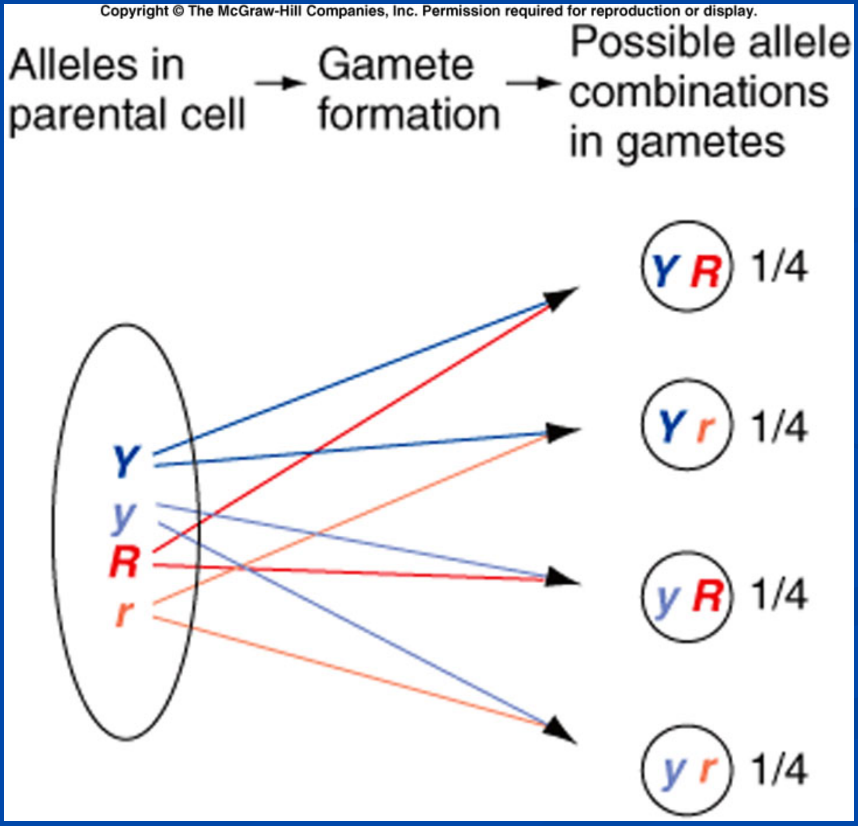
Type	Genotype	Phenotype	Number	Phenotypic ratio
Parental	*Y– R–*	yellow round	315	9/16
Recombinant	*yy R–*	green round	108	3/16
Recombinant	*Y– rr*	yellow wrinkled	101	3/16
Parental	*yy rr*	green wrinkled	32	1/16

Ratio of yellow (dominant) to green (recessive) = 12:4 or 3:1

Ratio of round (dominant) to wrinkled (recessive) = 12:4 or 3:1

The law of independent assortment

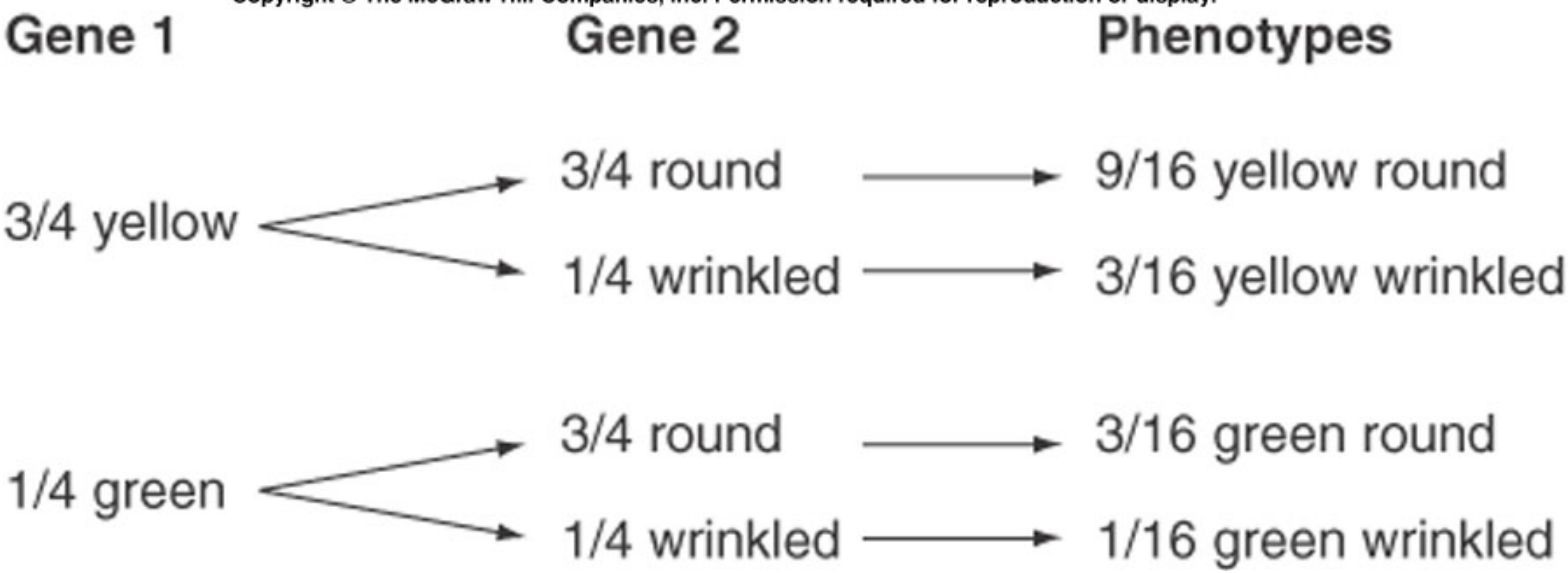
- During gamete formation different pairs of alleles segregate independently of each other



The ratio and numbers of different phenotype category:
(use the branching-diagram calculation method)

each of two traits has two alleles, so 2 X 2 = 4 different categories

the ratio of different categories = 9/16 : 3/16 : 3/16 : 1/16



How many different genotypes for the dihybrid F2 offspring?

each of two traits has three types, so 3 X 3 = 9 different categories

[YY, Yy, yy] x [RR, Rr, rr] = 3 x 3 = 9

The Punnet sqare method initially gives you 4 x 4 = 16 categories, but, if you look very carefully, there are redundant categories.

What about the numbers of different phenotype and genotype category, in case dealing with **more than 3 traits** with each having two alleles?

each of three traits has two alleles, so 2 x 2 x 2 = 8 different phenotypes

each trait has three different genotypes [AA, Aa, aa] x [BB, Bb, bb] x [CC, Cc, cc] = 3 x 3 x 3 = 27

No of different phenotypes = 2^n (n= No of traits or genes)

No of different genotypes = 3^n

Laws of probability for multiple genes

P RRYYTTSS X rryyttss

gametes RYTS ryts

F1 RrYyTtSs X RrYyTtSs

gametes

RYTS	RYTs	RYtS	RYts
RyTS	RyTs	RytS	Ryts
rYTS	rYTs	rYts	rYTS
ryTs	rYtS	rYts	ryts

RYTS	RYTs	RYtS	RYts
RyTS	RyTs	RytS	Ryts
rYTS	rYTs	rYts	rYTS
ryTs	rYtS	rYts	ryts

F2 What is the ratio of different genotypes and phenotypes?

P RRYYTTSS × rryyttss

F1 RrYyTtSs × RrYyTtSs

What is the probability of obtaining the genotype RrYyTtss?

Rr × Rr	Yy X Yy	Tt × Tt	Ss × Ss
1RR:2Rr:1rr	1YY:2Yy:1yy	1TT:2Tt:1tt	1SS:2Ss:1ss
2/4 Rr	2/4 Yy	2/4 Tt	1/4 ss

Probability of obtaining individual with Rr **and** Yy **and** Tt **and** ss.

$2/4 \times 2/4 \times 2/4 \times 1/4 = 8/256$ (or 1/32)

Loci Assort Independently! Look at each locus independently.

P RRYYTTSS × rryyttss

F1 RrYyTtSs × RrYyTtSs

What is the probability of obtaining a completely homozygous genotype?

Genotype could be RRYYTTSS **or** rryyttss

Rr × Rr	Yy × Yy	Tt × Tt	Ss × Ss
1RR:2Rr:1rr	1YY:2Yy:1yy	1TT:2Tt:1tt	1SS:2Ss:1ss
1/4 RR 1/4 rr	1/4 YY 1/4 yy	1/4 TT 1/4 tt	1/4 SS 1/4 ss

$(1/4 \times 1/4 \times 1/4 \times 1/4) + (1/4 \times 1/4 \times 1/4 \times 1/4) = 2/256$

Homework assignment for the discussion session of Aug 29st , Sept 1st

-Chapter 2

#1, #5, #7, #11, #15, #17, #18, #25, #26, #27