

Chapter 14

Mendel and the Gene Idea

Lectures modified by Garrett Dancik

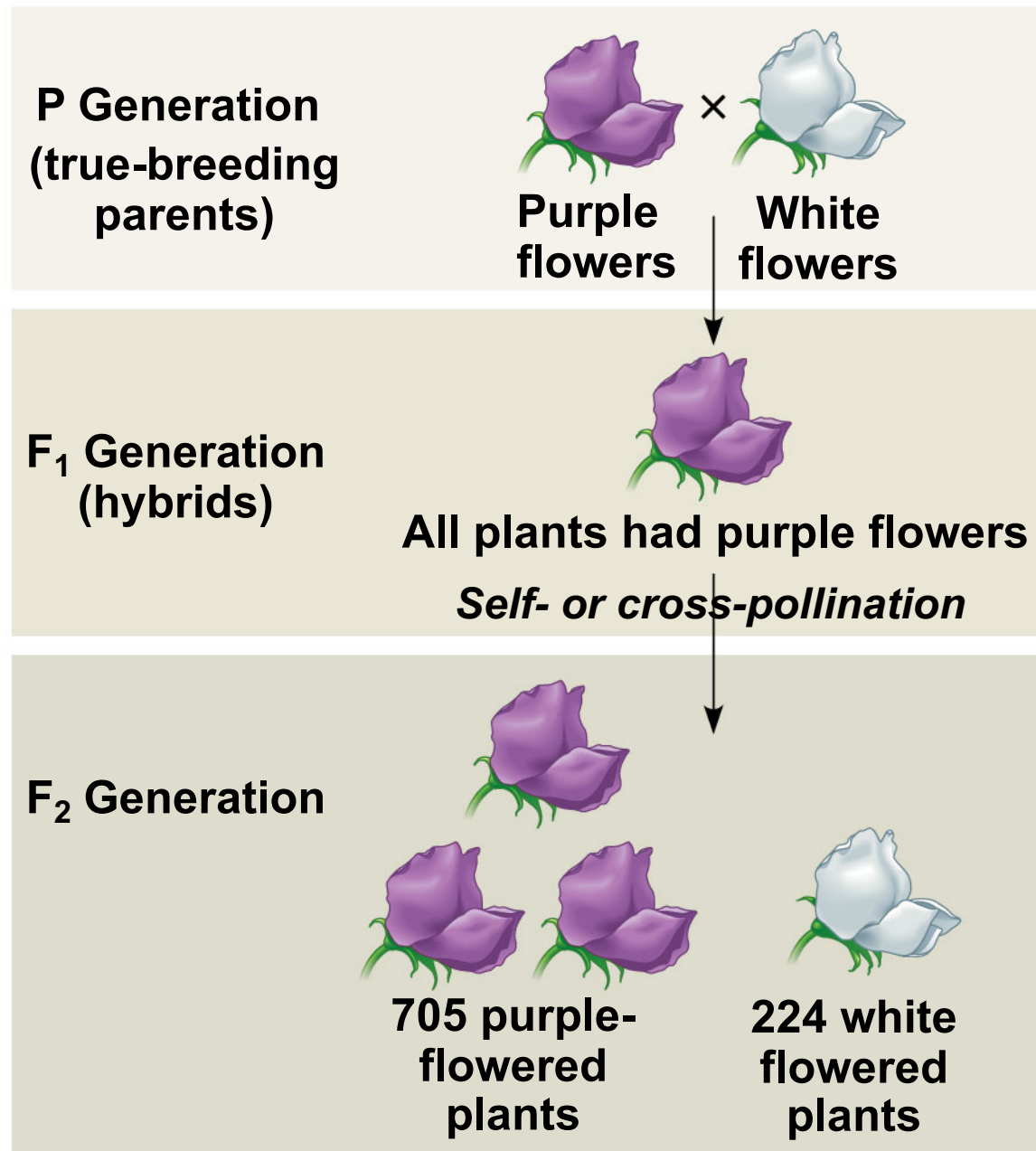
**Lectures by
Erin Barley
Kathleen Fitzpatrick**

Mendel's Experiments

- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments
- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**
- The true-breeding parents are the **P generation**
- The hybrid offspring of the P generation are called the **F₁ generation**
- When F₁ individuals self-pollinate or cross-pollinate with other F₁ hybrids, the **F₂ generation**

Figure 14.3-3















EXPERIMENT



- Mendel reasoned that only the purple flower factor was affecting flower color in the F_1 hybrids
- Mendel called the purple flower color a dominant trait and the white flower color a recessive trait
- The factor for white flowers was not diluted or destroyed because it reappeared in the F_2 generation

- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two possible traits
- What Mendel called a “heritable factor” is what we now call a gene

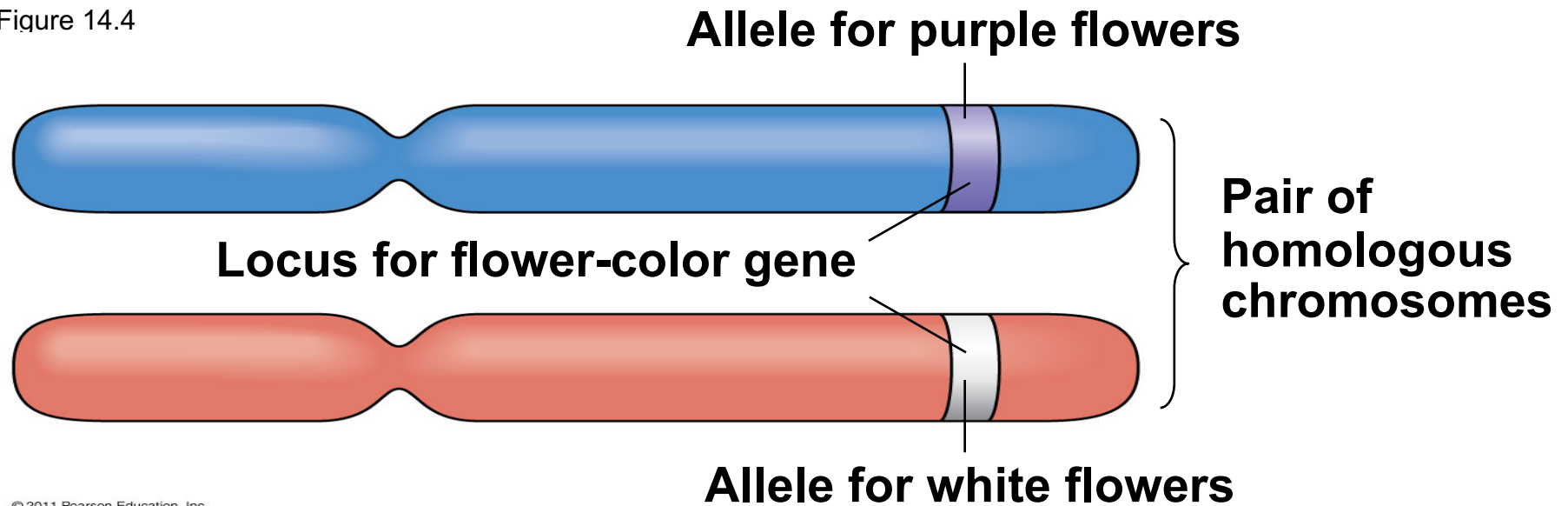
Table 14.1 The Results of Mendel's F₁ Crosses for Seven Characters in Pea Plants

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 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	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

Mendel's Model

- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F_2 offspring
- Four related concepts make up this model
- These concepts can be related to what we now know about genes and chromosomes

Figure 14.4



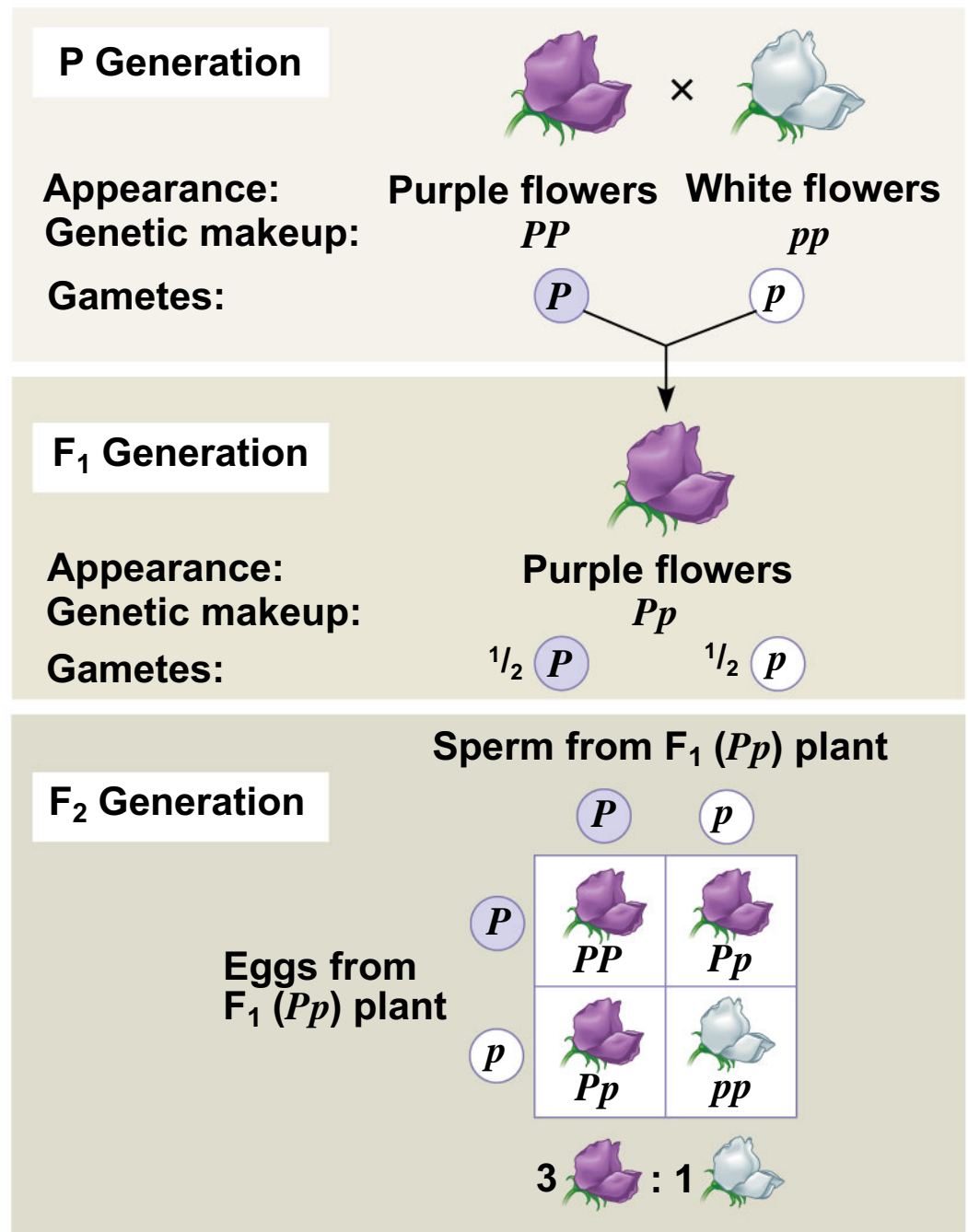
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Mendel's model

1. Alternative versions (alleles) of hereditary factors (genes) account for variability in inherited traits
2. An organism inherits two alleles, one from each parent
3. If two alleles differ, then the trait is determined by the *dominant* allele; the *recessive* allele has no effect on appearance
4. the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes (this is now known as the **law of segregation**)
5. Alleles for separate genes are passed independently of each other (this is now known as the law of **independent assortment** – more on this later)

Figure 14.5-3

- A capital letter denotes the dominant allele while lowercase denotes a recessive allele
- The possible combinations of sperm and egg can be shown using a **Punnett square**, a diagram for predicting the results of a genetic cross between individuals of known genetic makeup



Useful Genetic Vocabulary

- An organism with two identical alleles for a character is said to be **homozygous** for the gene controlling that character
- An organism that has two different alleles for a gene is said to be **heterozygous** for the gene controlling that character
- We distinguish between an organism's **phenotype**, or physical appearance, and its **genotype**, or genetic makeup

The Law of Independent Assortment

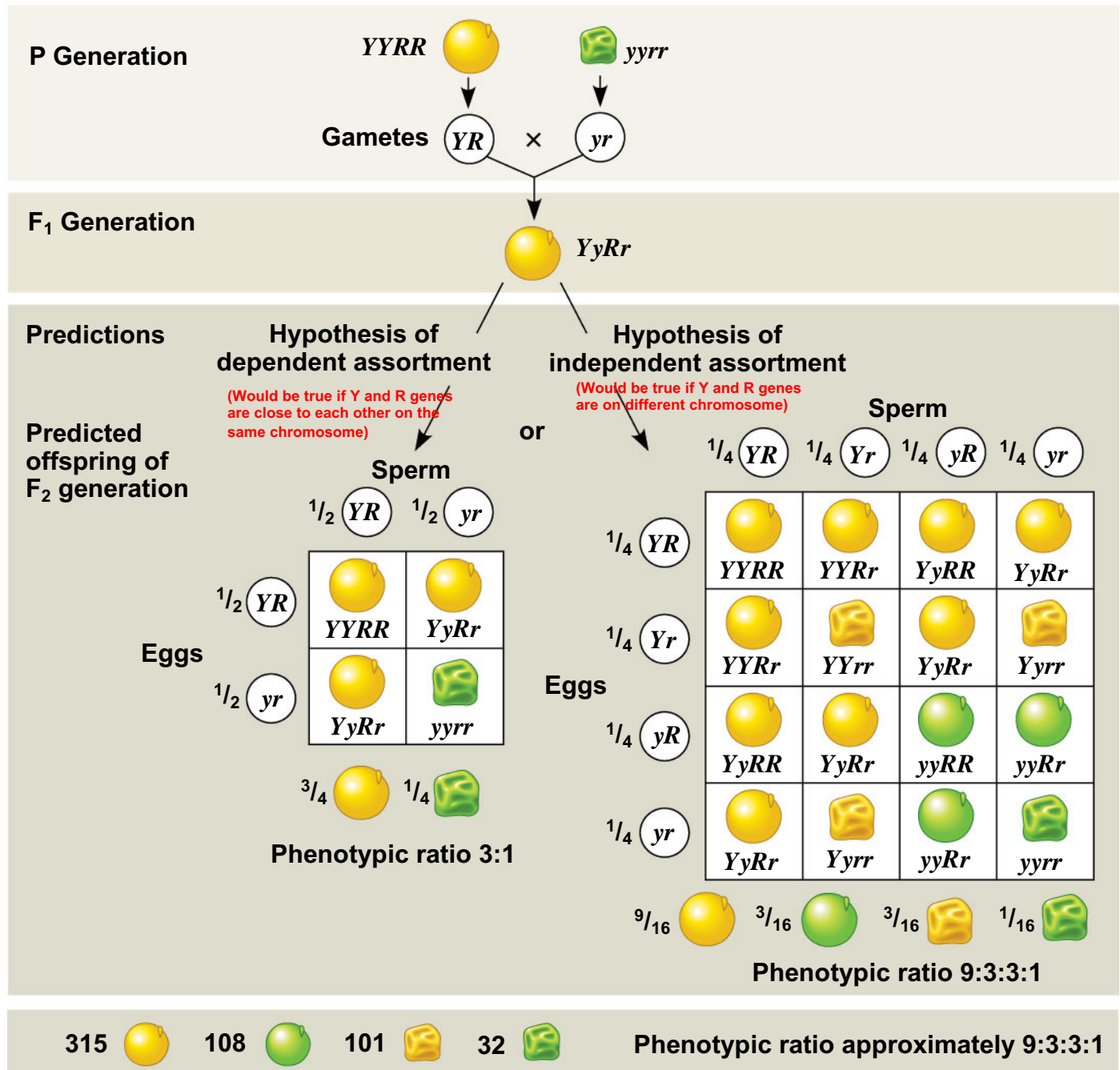
- Mendel derived the law of segregation by following a single character
- The F_1 offspring produced in this cross were **monohybrids**, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a **monohybrid cross**

Question

- Suppose that purple flowers that are heterozygous are crossed with white flowers that are homozygous recessive for flower color.
 - (a) What is the genotypic ratio of the offspring?
 - (b) What is the phenotypic ratio of the offspring?
 - (c) What is the probability that a child flower is purple?

- 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
- A **dihybrid cross**, a cross between F_1 dihybrids, can determine whether two characters are transmitted to offspring as a package or independently

Figure 14.8

EXPERIMENT

- Using a dihybrid cross, Mendel developed the **law of independent assortment**
- The law of independent assortment states that each pair of alleles segregates independently of every other pair of alleles during gamete formation (sperm/egg cells of a YyRr individual will contain *either* YR or yR)
- Strictly speaking, this law applies only to genes on different, nonhomologous chromosomes or those far apart on the same chromosome
- Genes located near each other on the same chromosome tend to be inherited together

Concept 14.3: Inheritance patterns are often more complex than predicted by simple Mendelian genetics


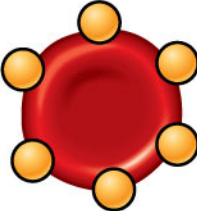
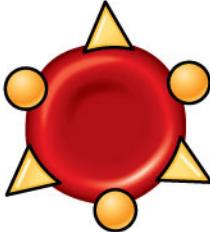

- The relationship between genotype and phenotype is rarely as simple as in the pea plant characters Mendel studied
- Many heritable characters are not determined by only one gene with two alleles
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance
- For example, ABO blood typing is determined by three alleles and is an example of *codominance*

Figure 14.11

(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	$I^A I^A$ or $I^A i$	$I^B I^B$ or $I^B i$	$I^A I^B$	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

Nature and Nurture: The Environmental Impact on Phenotype

- Example: hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity



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Genetic Testing and Counseling

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease

Additional complexities of inheritance

- Most genetic traits are not as simple as the ones that Mendel studied.
- Incomplete dominance, codominance, multiple alleles, pleiotropy, epistasis, and polygenic inheritance are described on the remaining slides

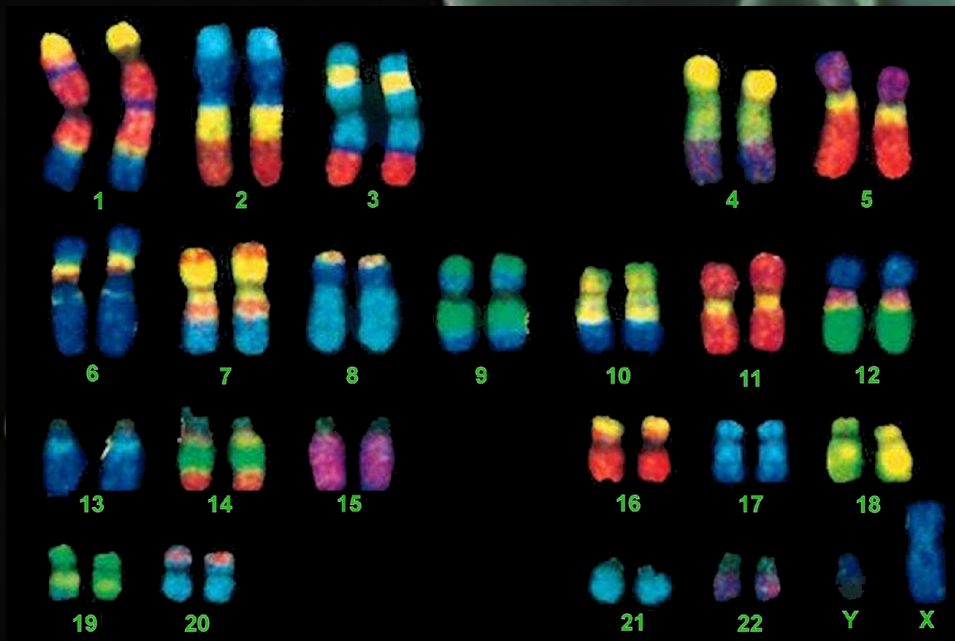
Figure 14.UN03

Relationship among alleles of a single gene	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homozygous dominant	PP  Pp 
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	   C^RC^R C^RC^W C^WC^W
Codominance	Both phenotypes expressed in heterozygotes	I^AI^B 
Multiple alleles	In the whole population, some genes have more than two alleles	ABO blood group alleles I^A , I^B , i
Pleiotropy	One gene is able to affect multiple phenotypic characters	Sickle-cell disease

Chapter 15

The Chromosomal Basis of Inheritance

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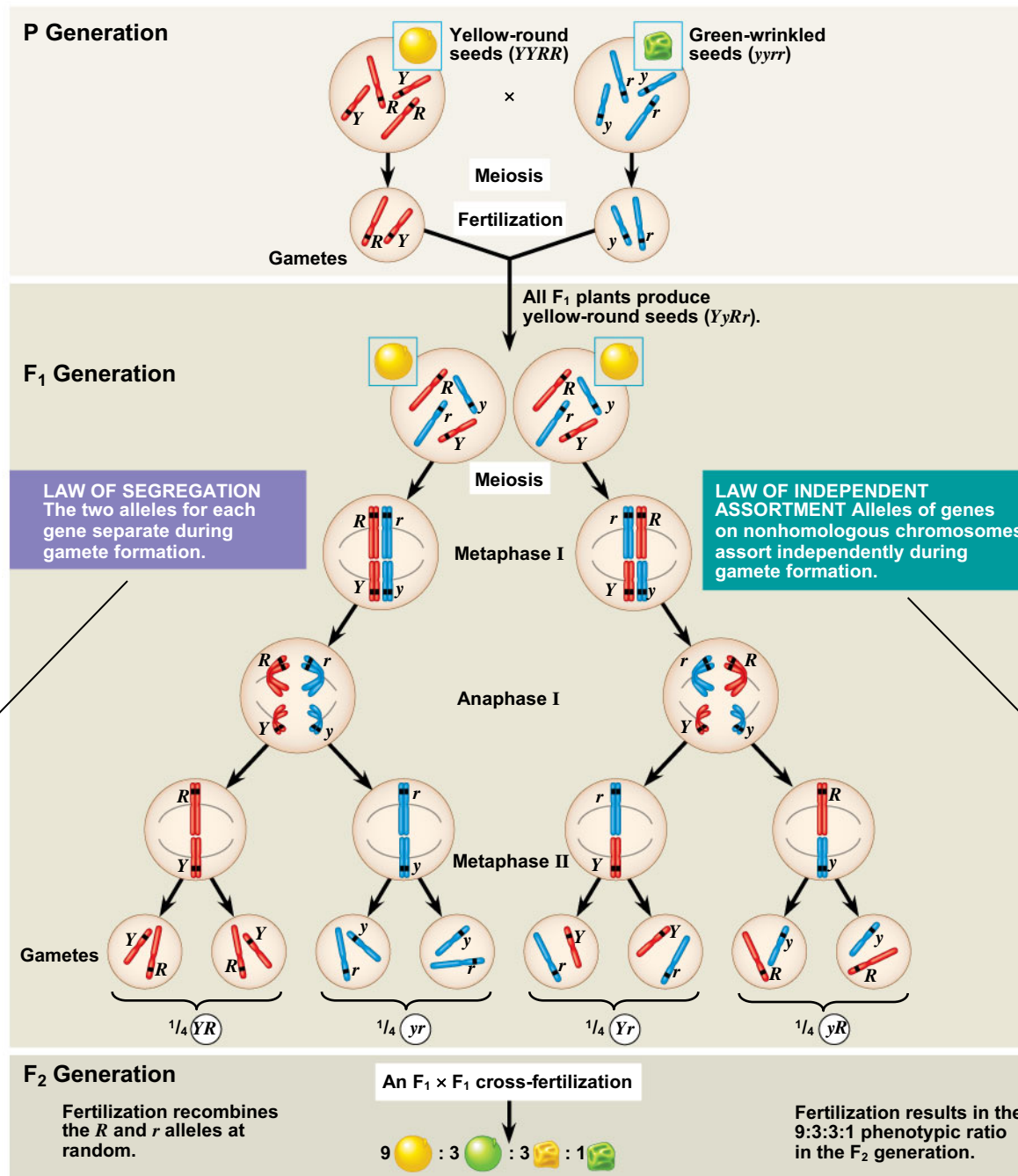


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Concept 15.1: Mendelian inheritance has its physical basis in the behavior of chromosomes

- The **chromosome theory of inheritance** states:
 - Mendelian genes have specific loci (positions) on chromosomes
 - Chromosomes undergo segregation and independent assortment
- The behavior of chromosomes during meiosis (cell division leading to the production of gametes) can account for Mendel's laws of segregation and independent assortment

Figure 15.2



For each gene, gamete contains only one allele (e.g., Y or y, not Yy)

Each gamete randomly contains a combination of alleles from each gene (YR, yR, Yr, yr)

Correlating Behavior of a Gene's Alleles with Behavior of a Chromosome Pair

- In one experiment, Morgan mated male flies with white eyes (mutant) with female flies with red eyes (wild type)
 - The F_1 generation all had red eyes
 - The F_2 generation showed the 3:1 red:white eye ratio, but only males had white eyes
- Morgan determined that the white-eyed mutant allele must be located on the X chromosome
- Morgan's finding supported the chromosome theory of inheritance

Figure 15.4

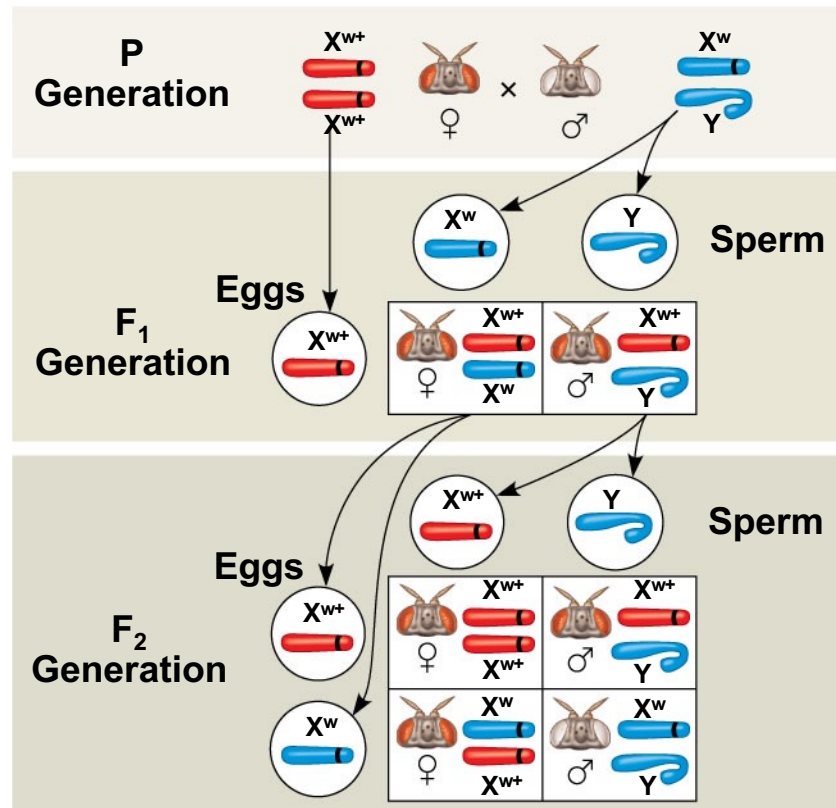
EXPERIMENT



RESULTS



CONCLUSION



XX – female fly
XY – male fly

w^+ - red eyes (wild type)
 w – white eyes (mutant)

Now we use notation to indicate that the eye color allele is on the X chromosome:

X^{w+} - an X chromosome with the red eye allele

Concept 15.2: Sex-linked genes exhibit unique patterns of inheritance

- In humans and some other animals, there is a chromosomal basis of sex determination
- Females are XX, and males are XY*
- Each ovum contains an X chromosome, while a sperm may contain either an X or a Y chromosome
- Other animals have different methods of sex determination

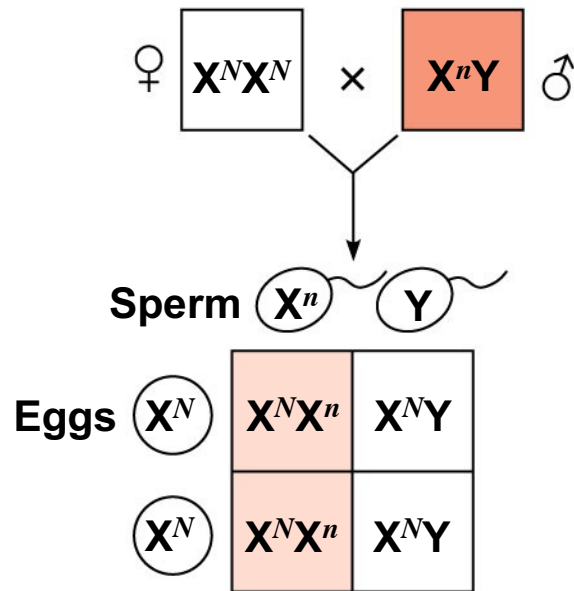
*Actually, it's more complicated than that,

<https://www.sciencealert.com/a-baby-s-sex-is-about-more-than-just-its-x-and-y-chromosomes-new-research-reveals>

- A gene that is located on either sex chromosome is called a **sex-linked gene**
- Genes on the Y chromosome are called Y-linked genes; there are few of these
- Genes on the X chromosome are called **X-linked genes**

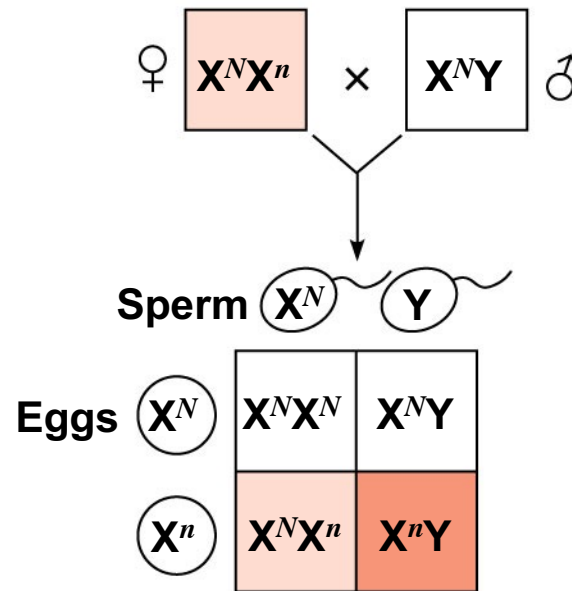
- X-linked genes follow specific patterns of inheritance
- For a recessive X-linked trait to be expressed
 - A female needs two copies of the allele (homozygous)
 - A male needs only one copy of the allele (hemizygous)
- X-linked recessive disorders are much more common in males than in females

Inheritance of X-linked recessive traits

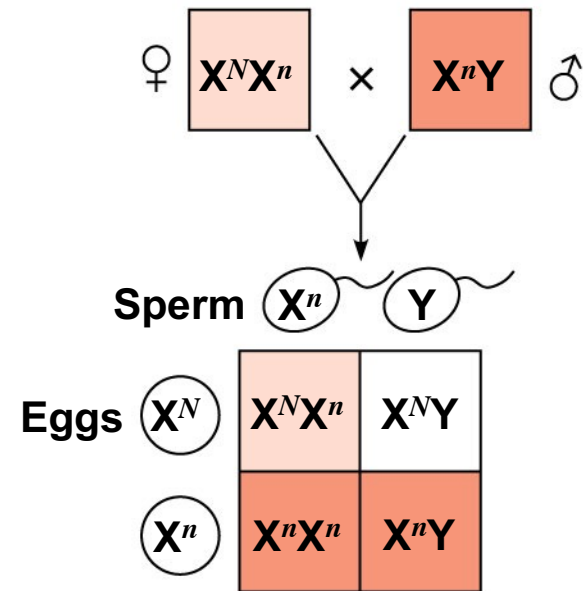


(a)

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(b)



(c)

- Some disorders caused by recessive alleles on the X chromosome in humans
 - Color blindness (mostly X-linked)
 - Duchenne muscular dystrophy
 - Hemophilia

X Inactivation in Female Mammals

- In mammalian females, one of the two X chromosomes in each cell is randomly inactivated during embryonic development
- The inactive X condenses into a **Barr body**
- If a female is heterozygous for a particular gene located on the X chromosome, she will be a mosaic for that character

Figure 15.8

Early embryo:

X chromosomes

**Allele for
orange fur**

**Allele for
black fur**

**Cell division and
X chromosome
inactivation**

**Two cell
populations
in adult cat:**

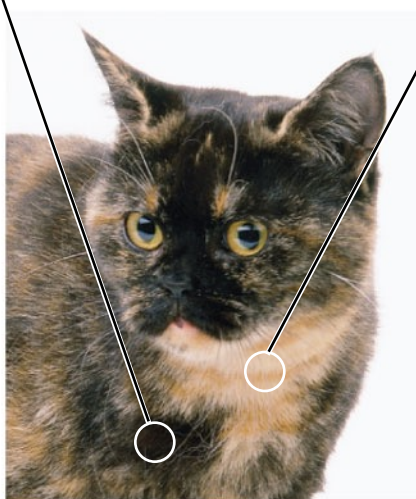
Active X

Inactive X

Active X

Black fur

Orange fur



Recombination of Linked Genes: Crossing Over

- Morgan discovered that genes can be linked, but the linkage was incomplete, because some recombinant phenotypes were observed
- He proposed that some process must occasionally break the physical connection between genes on the same chromosome
- That mechanism was the **crossing over** of homologous chromosomes

- Genes that are far apart on the same chromosome can have a recombination frequency near 50%
 - This is due to crossing over:
 - <https://www.youtube.com/watch?v=pdJUvagZjYA>
- Such genes are physically linked, but genetically unlinked, and behave as if found on different chromosomes