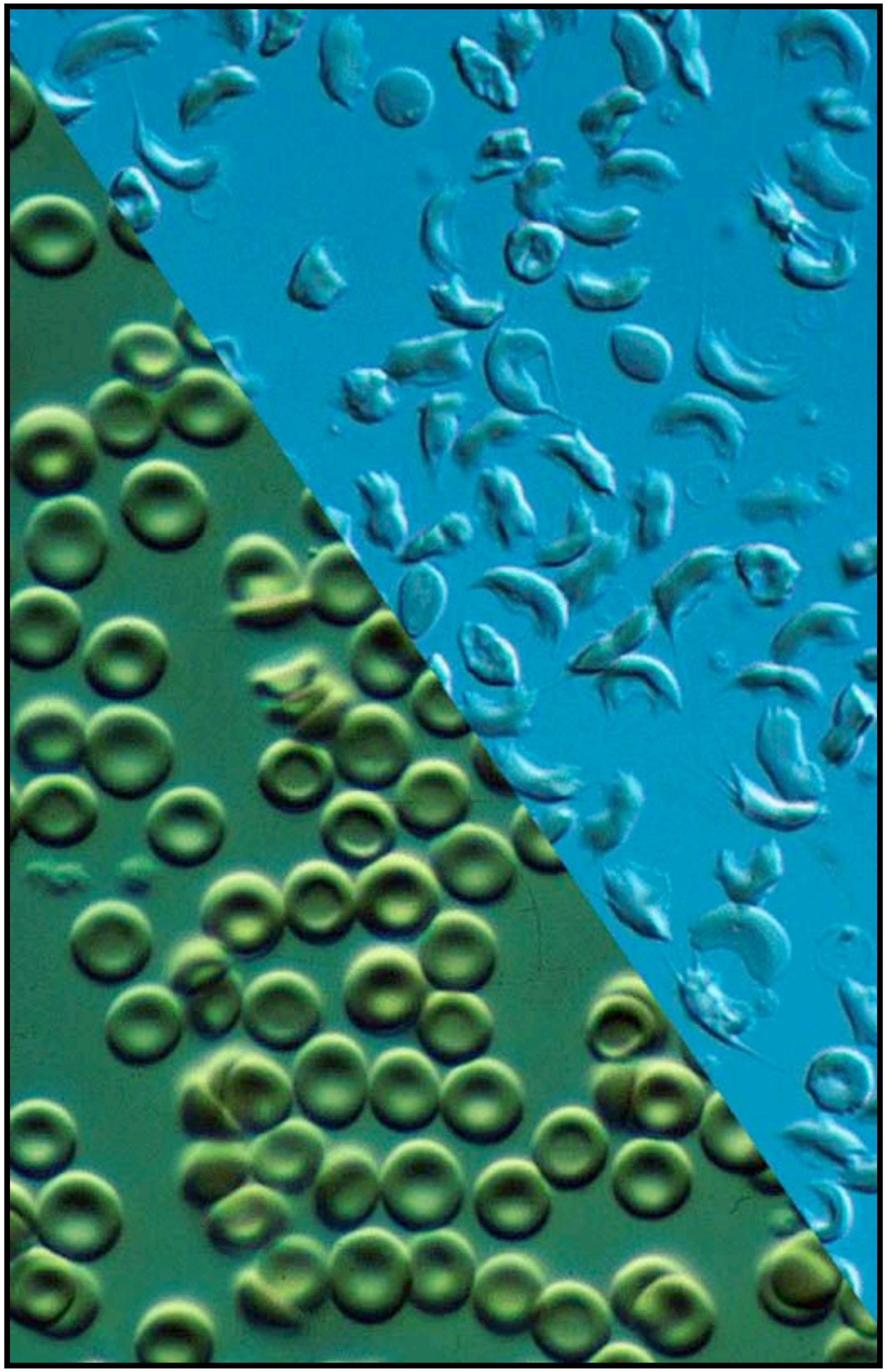
Chapter 4

- ❖Extensions to Mendelian Genetics
- ❖ **Gene Interactions**



Gene Interactions – Extensions to Mendelian Genetics

- Just as different alleles of **1 gene** can interact in complex ways,

- **2 different genes** can also act together to modify a phenotype:

  - 2 genes 1 phenotype (Additive Gene Action)

  Complementation (complementary gene action)

  Epistasis (recessive and dominant)

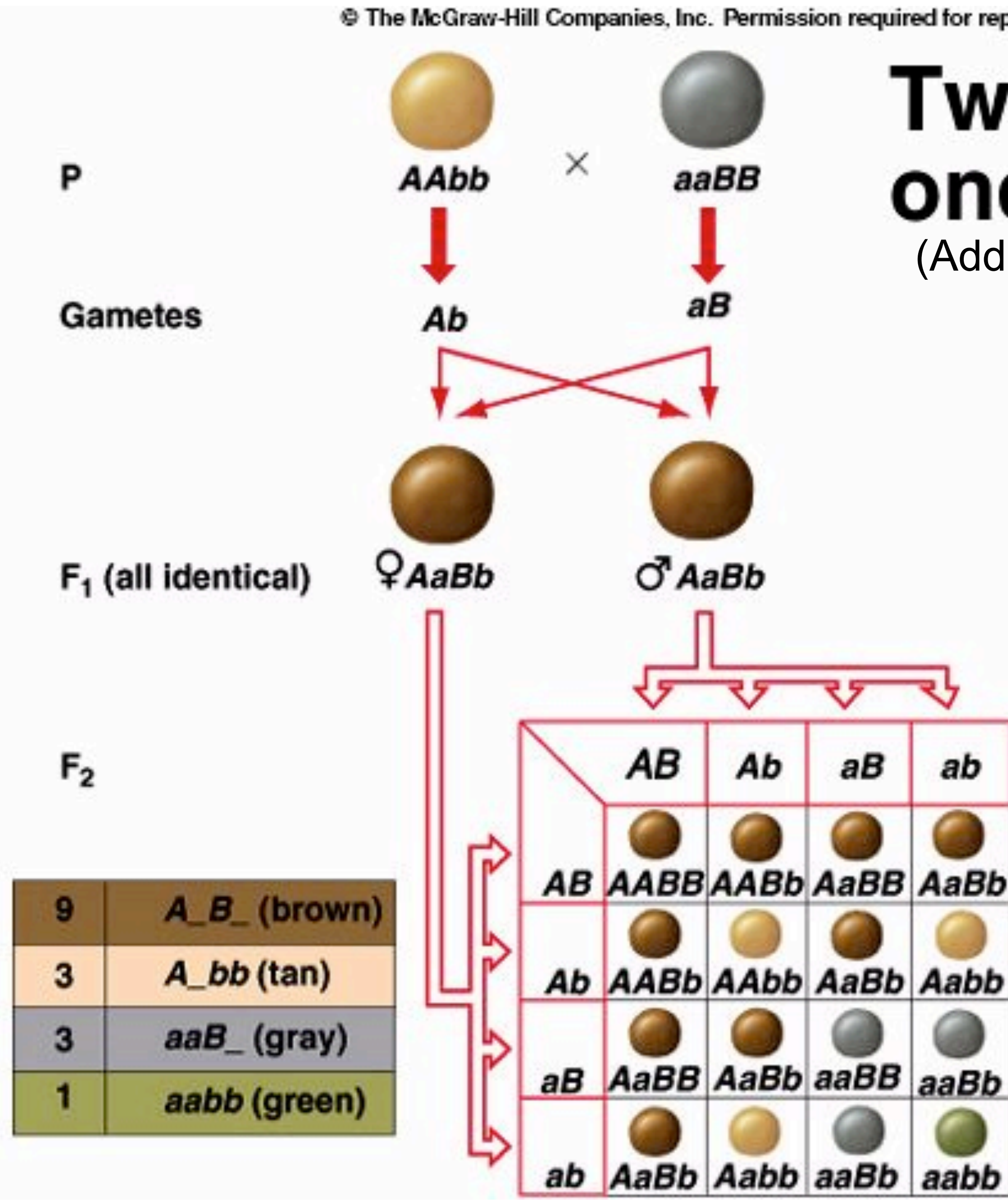
  Redundancy

Multifactorial Inheritance

- Vast majority of traits are determined by multiple factors:
  - genetic as well as environmental.
- Gene interactions between two or more genes
  - Example: Lentil Seed color.
- **F1 all same, F2: 4 different phenotypes**
- **F2 phenotypic ratio is 9:3:3:1**
  - (same as **F2 dihybrids** in Mendel's original crosses).
- Difference:
  - in original crosses: 2 independent traits/phenotypes=2 independent genes;
    - Seed color and seed shape
  - here: multiple phenotypes of 1 trait=2 independent genes
    - Seed color only.

Two genes, one phenotype

(Additive Gene Action)



You can tell this genotype is caused by more than one gene :

- because there are 4 phenotypes not 3 in F2

•1 gene F2 would have 3 phenotypes 1:2:1 ratio

F2 phenotypes

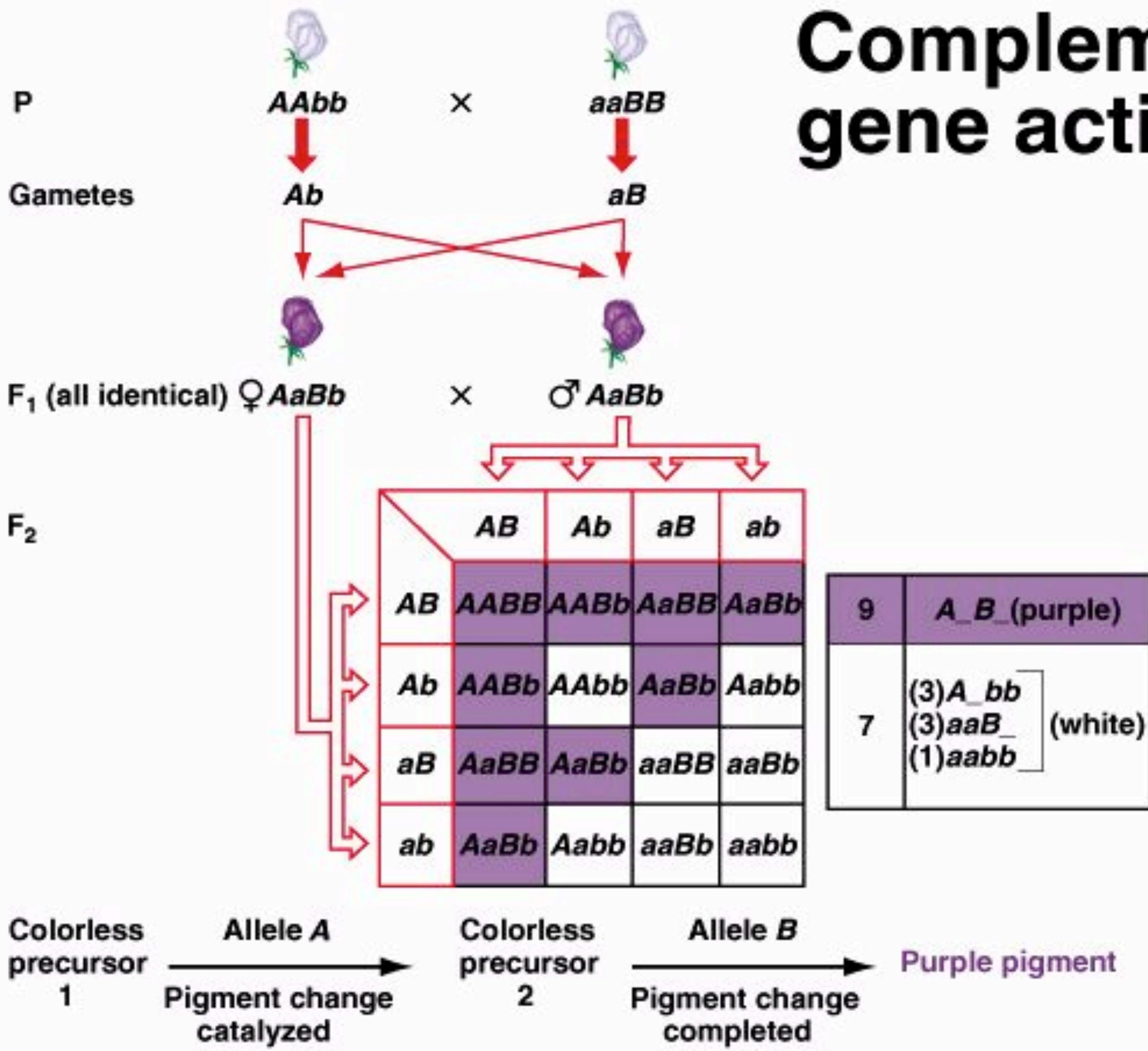
- **Dominance Relationships**:
  - Tan is dominant to green
  - Gray is dominant to green
  - Brown is dominant to gray, green and tan.
  - Tan and Gray are incompletely dominant, giving rise to brown.
- **Genotypic classes**:
  - Brown: A_B_
  - Tan: A_bb
  - Gray: aaB_
  - Green: aabb

Complementary Gene Action

- **Each genotypic class may not always dictate a unique phenotype**
- A pair of genes can often work together to create a specific phenotype. We call this complementary interaction.
- With this type of interaction we see 2 different phenotypes instead of the 4 seen in 2 genes 1 phenotype
- Two or more genotypic classes may display an identical phenotype.
  - Example: Two lines of pure breeding white flowered pea plants falling into different genotypic classes: AAbb & aaBB

The must have a dominant allele in both genes to result in the purple flower phenotype

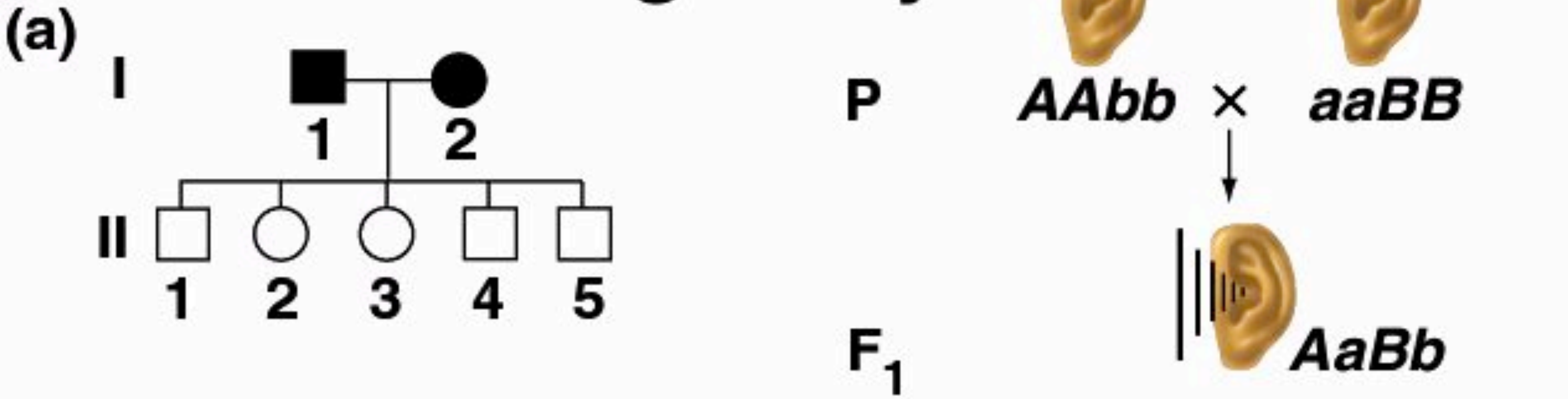
Complementary gene action



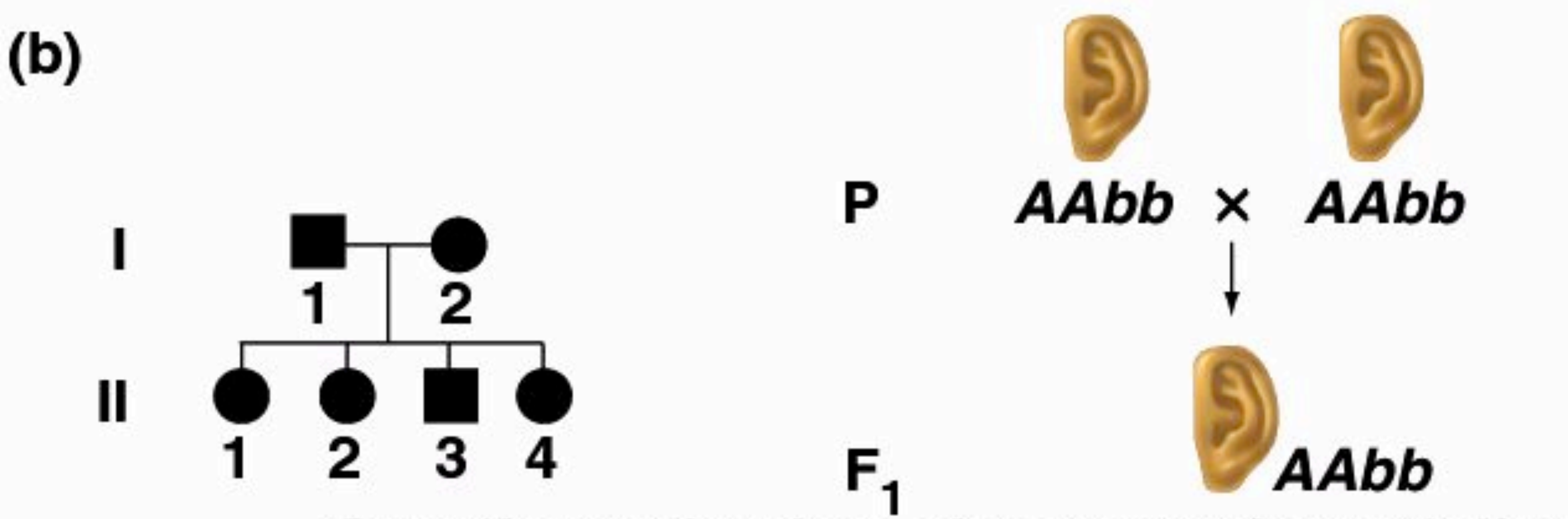
	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

9	A_B_ (purple)
7	(3)A_bb (3)aaB_ (1)aabb (white)

Genetic heterogeneity



Genetic mechanism of complementation



Genetic mechanism of noncomplementation

Epistasis

- One gene’s allele masks the phenotype of the other gene’s alleles.
- Four genotypic classes produce fewer than four phenotypes.
- Different types of epistasis:
- **Recessive epistasis**: *when the recessive allele of one gene masks the effects of either allele of the second gene.*
- **Dominant epistasis:** *when the dominant allele of one gene masks the effects of either allele of the second gene.*

Recessive Epistasis

- Example 1: Coat color of Labrador retriever
- Example 2: ABO blood groups: Bombay phenotype.
- Phenotypic ratios are 9:3:4 in F2.

Coat-Color Inheritance in Labrador Retrievers

P

golden

X

black

F1

black

Recessive Epistasis:

a recessive mutation in one gene masks the phenotypic effects of another

F1

x

Appears like incomplete dominance because some of the progeny look like neither parent, but the ratio is wrong.

F2

9 : 3 : 4

Dihybrid Cross:

BbEe  x  BbEe

F2

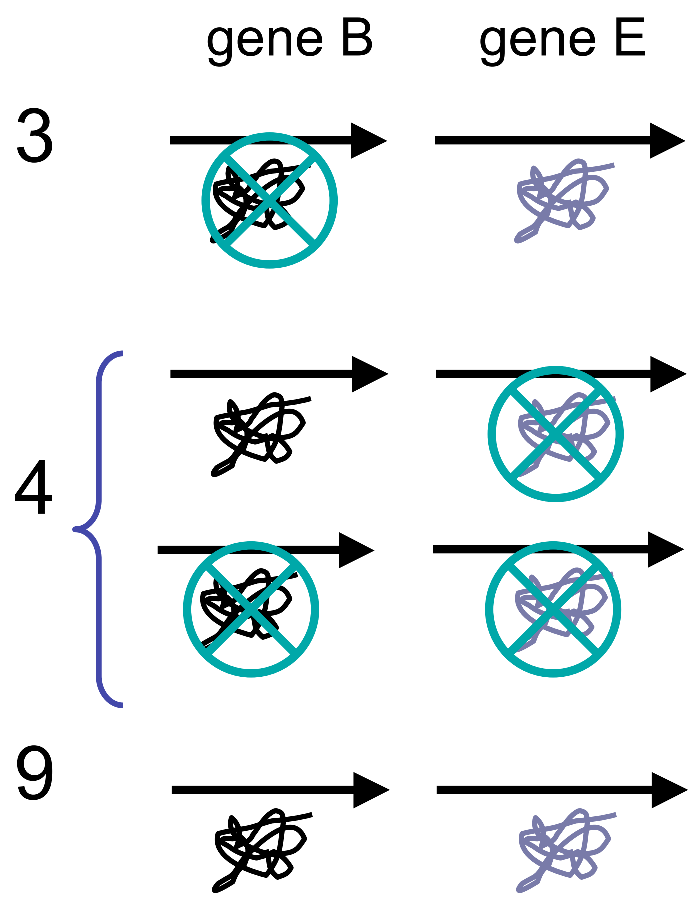
	BE	Be	bE	be
BE	BBEE	BBEe	BbEE	BbEe
Be	BBEe	BBee	BbEe	Bbee
bE	BbEE	BbEe	bbEE	bbEe
be	BbEe	Bbee	bbEe	bbee

9 black: 3 brown: 4 golden

(9 B-E-: 3 bbE-: 3 B-ee: 1 bbee)

Molecular Explanation

Pigment production (B) and subsequent incorporation (E) into the hair shaft are controlled by two separate genes. To be black, both genes must function. Mutations in B (b) lead to brown pigment. Mutations in E (e) lead to no pigment in coat.



Recessive Epistasis

- Two genes involved in coat color determination.
- Gene B determines whether black (B) or brown (bb) pigment is produced.
- Gene E determines if pigment is deposited in hair
  - golden retrievers (ee) make either black (B-) or brown (bb) pigment (look at noses)… but not in fur
- The recessive allele is epistatic to (stands over) other genes when homozygous -- hence the name “recessive epistasis”
- Phenotypes do not segregate according to Mendelian ratios (the phenotypic ratios are modified Mendelian ratios).
- epistasis - (Greek, to stand upon or stop) the differential phenotypic expression of a genotype at one locus caused by the genotype at another, non allelic, locus. A mutation that exerts its expression by canceling the expression of the alleles of another gene.

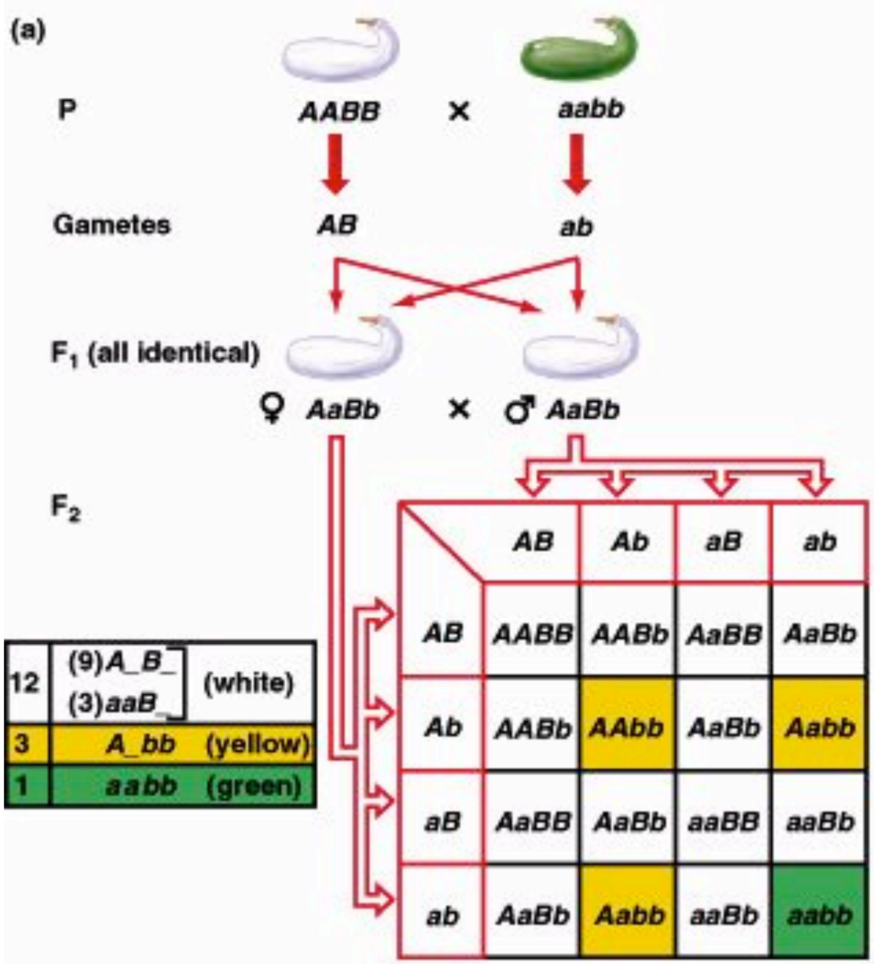
Dominant Epistasis

- caused by the dominant allele of one gene, masking the action of either allele of the other gene.

- Ratio is 12:3:1 instaed f 9:3:3:1

- Example: Summer Squash

Dominant epistasis



Redundancy: Duplicate Genes

Petal color in snapdragons - if Mendel had used snap dragons for his experiments, he wouldn't be famous!

P  X 

AABB aabb

F1  A-B-

F2 15/16 red; 1/16 white

A-B-  X  A-B-

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

15/16 A-B- ➡ red; 1/16 aabb ⇨ white

Whenever a dominant gene is present, the trait is expressed.
One allele is sufficient to produce the pigment.

Hints for figuring out gene interactions:

Look at the F2 phenotypic ratios!!

- If one gene is involved in the trait, then the monohybrid phenotypic ratio is:
  3:1 or 1:2:1 or 2:1

- If two genes are involved in the trait, then the dihybrid phenotypic ratio is:
  9:3:3:1 or some permutation (9:4:3 or 9:7 or 12:3:1)
  → The 1/16 class is always the double homozygous recessive.
  → Look for internal 3:1 ratios, which will indicate dominance/recessive relationships for alleles within a gene.

Hints for figuring out gene interactions:

- **2 Genes 1 Phenotype (Additive Gene Action):** You can tell this genotype is caused by more than one gene because there are 4 phenotypes not 3 in F2 (9:3:3:1)
  - 1 gene F2 would have 3 phenotypes 1:2:1 ratio
- **Complementary Gene Action**: one good copy of each gene is needed for expression of the final phenotype
  - 9:7 ratio
- **Epistasis**: one gene can mask the effect of another gene
  - 9:3:4 ratio for recessive epistasis
  - 12:3:1 ratio for dominant epistasis
- **Duplicate genes**: only double mutant has mutant phenotype
  - 15:1 ratio

variations on Mendelian inheritance

Gene interaction	Inheritance pattern	A-/B-	A-/bb	aa/B-	aabb	ratio
Additive	Each genotype results in a unique phenotype	9	3	3	1	9:3:3:1
Complementary	At least one dominant allele from each of two genes needed for phenotype	9	3	3	1	9:7
Recessive Epistasis	Homozyous recessive genotype at one locus masks expression at second locus	9	3	3	1	9:3:4
Dominant Epistasis	Dominant allele at one locus masks expression at second locus	9	3	3	1	12:3:1
Duplicate Genes	One dominant allele from either of twogenes needed for phenotype	9	3	3	1	15:1

Sample Problem

true breeding brown dogs **X** true breeding white dogs

F1 = all white

F2 = 118 white 12

32 black 3

10 brown 1

- Find the genotypes of the dogs in each class:

What is the ratio?

How many genes? 2

What is the ratio of white to colored dogs? 12:4 = 3:1

This means that white is dominant to colored so let's call one gene: W= white w=colored

F2 = 118 white
32 black
10 brown

What is the ratio of black to brown dogs? 3 : 1

So black must be dominant to brown. So we will call the second gene: B=black and b=brown

What class of dogs are the double recessive homozygotes and what is their genotype?

Brown - wwbb

What is the genotype of the black dogs?

Must be wwB-

What are the genotypes of the white dogs?

W_ B_ and W_bb

- This is an example of dominant epistasis (white).

Same Genotype may produce different Phenotypes

- **Penetrance:** Genotype does not necessarily define phenotype. *The proportion of individuals with a given genotype express the phenotype determines penetrance.*

- 100% penetrance = all individuals show phenotype.

- 50% penetrance = half the individuals show phenotype.
  - Example: retinoblastoma: only 75% individuals affected.

- **Expressivity**: *the degree or intensity with which a particular genotype is expressed in a phenotype in a given individual.*

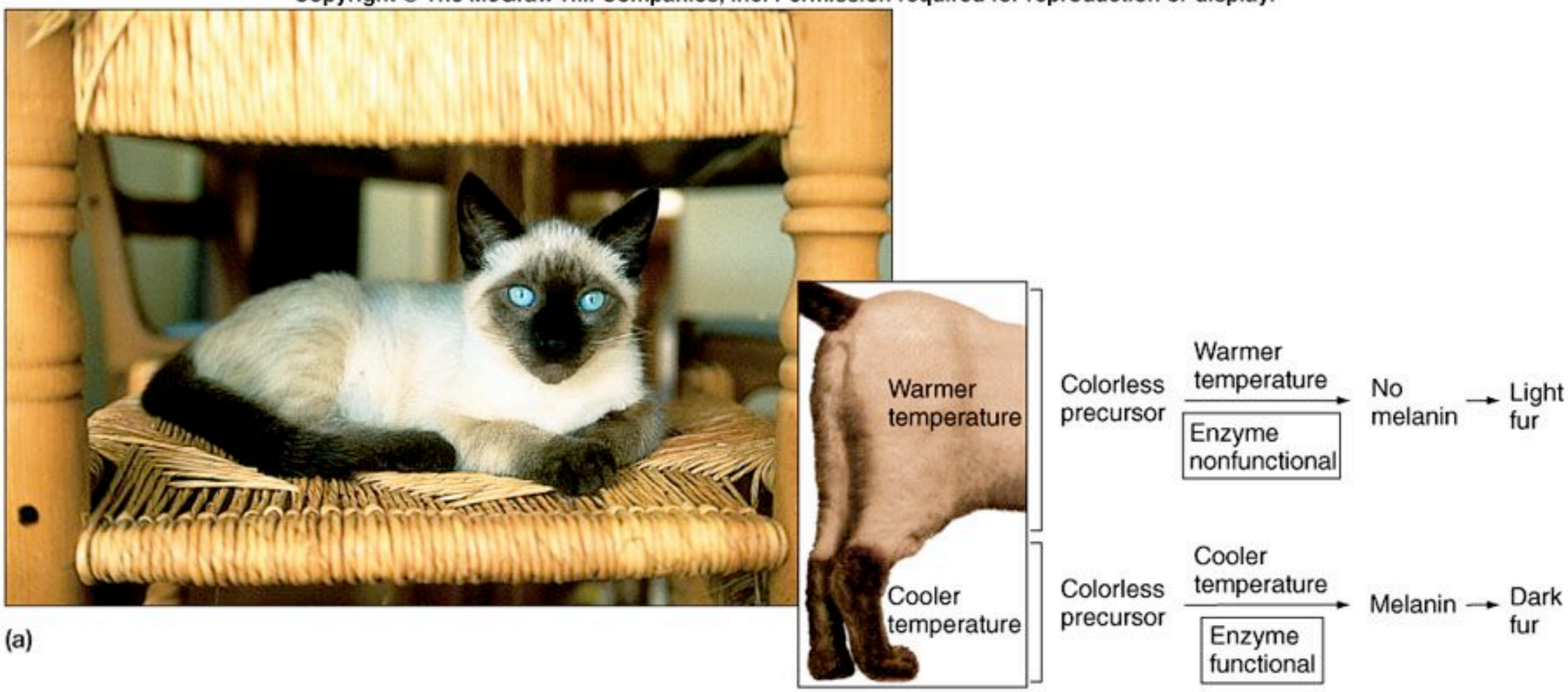
  - Retinoblastoma: some have both eyes affected, some only one.

Modifier Effects

- **Modifier Genes**: *they have a subtle, secondary effect which alters the phenotypes produced by the primary genes.*
  - E.G. Tail length in mice. The mutant allele *t* causes a shortening of the tail. Not all short tails are of the same length: another gene affects the actual length. (Variable expressivity).
- **Modifying environment**: The environment may influence the effect of a genotype on the phenotype.
  - E.G.: Siamese cats: temperature dependent color of coat. Color shows up only in extremities, where the temp is lower (enzyme for pigment formation is active only at lower temp.)

Modifying environment:

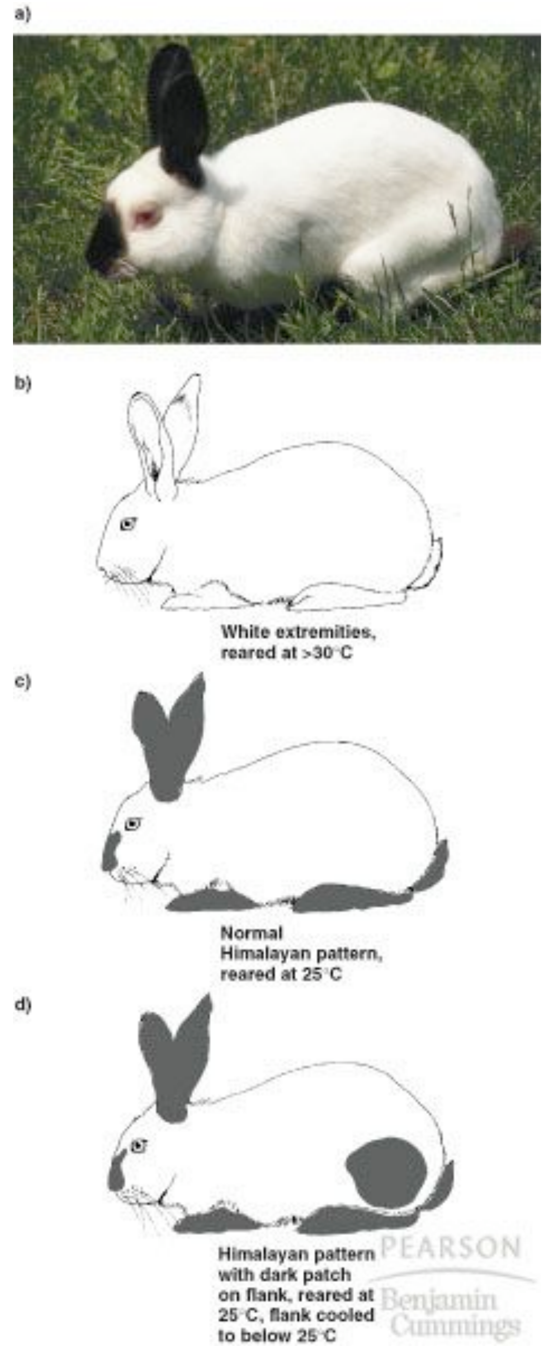
The environmental influence of a genotype on the phenotype= phenocopy



(a)

(b)

Modifying environment:
The environmental influence
of a genotype on the phenotype=
phenocopy



Homework Problems

– Chapter 4

– # 15, 16, 19, 26

- DON'T forget to take the online QUIZ!!