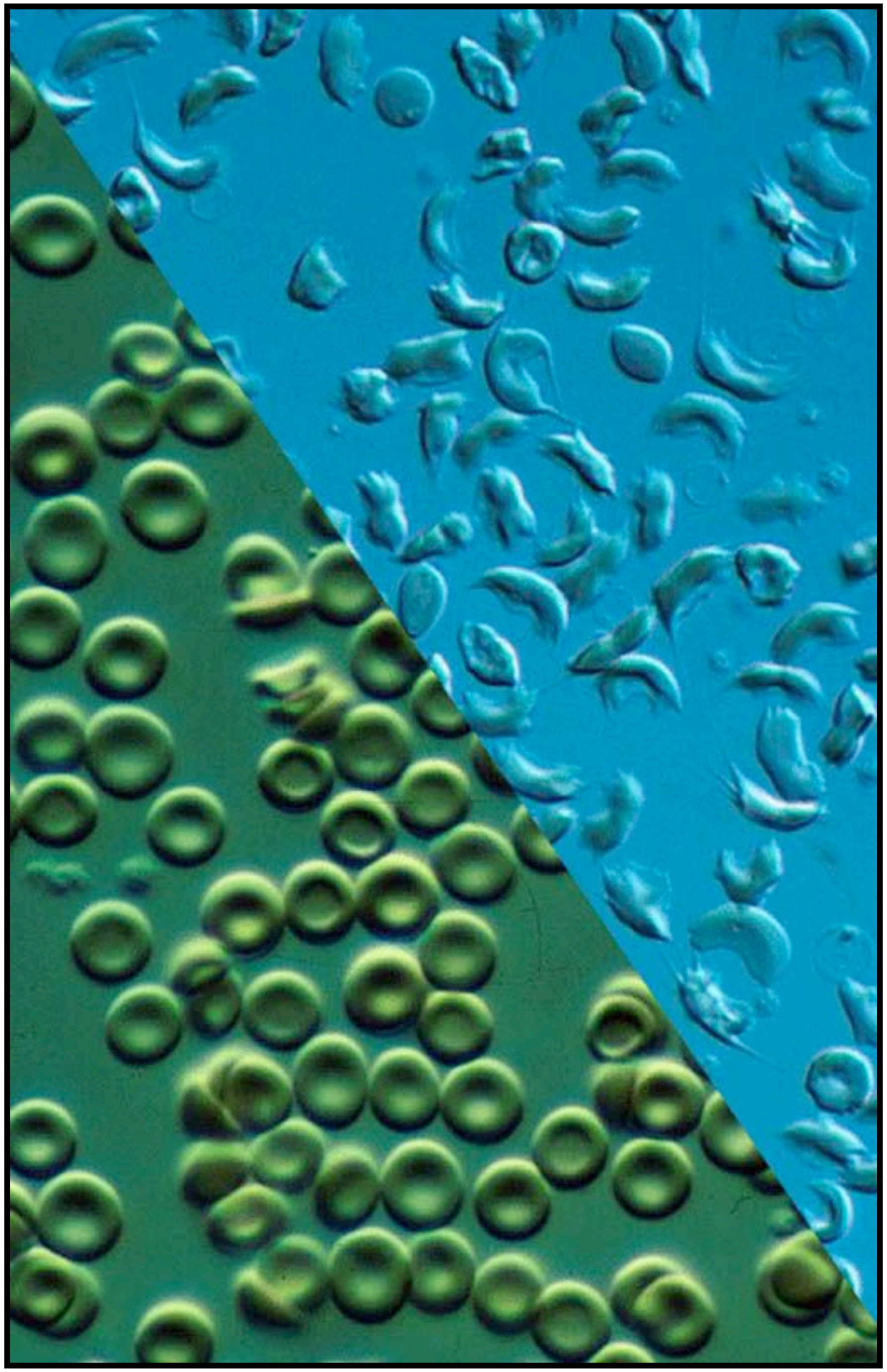
# Chapter 4

- 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)

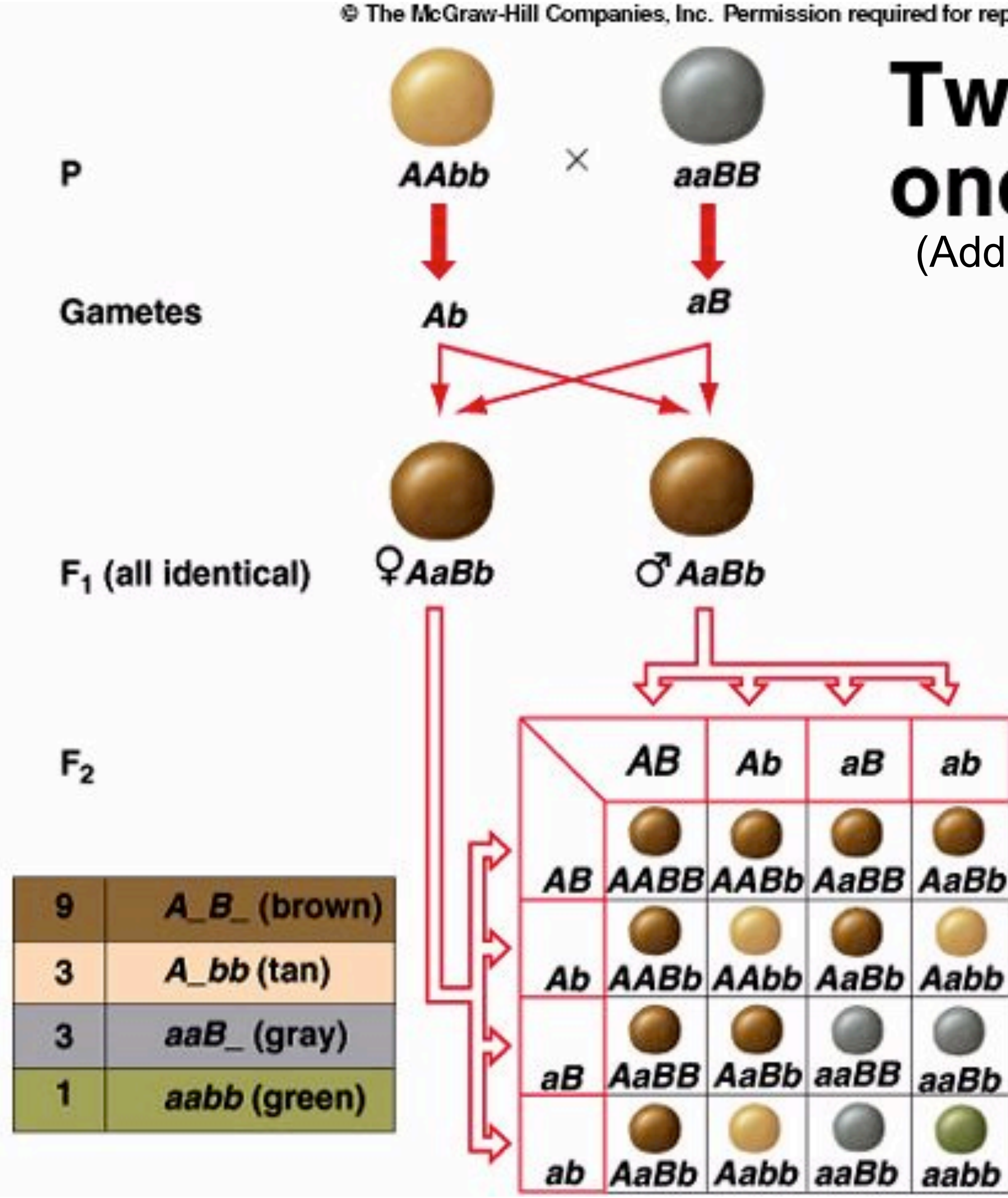
    Redundancy

# 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

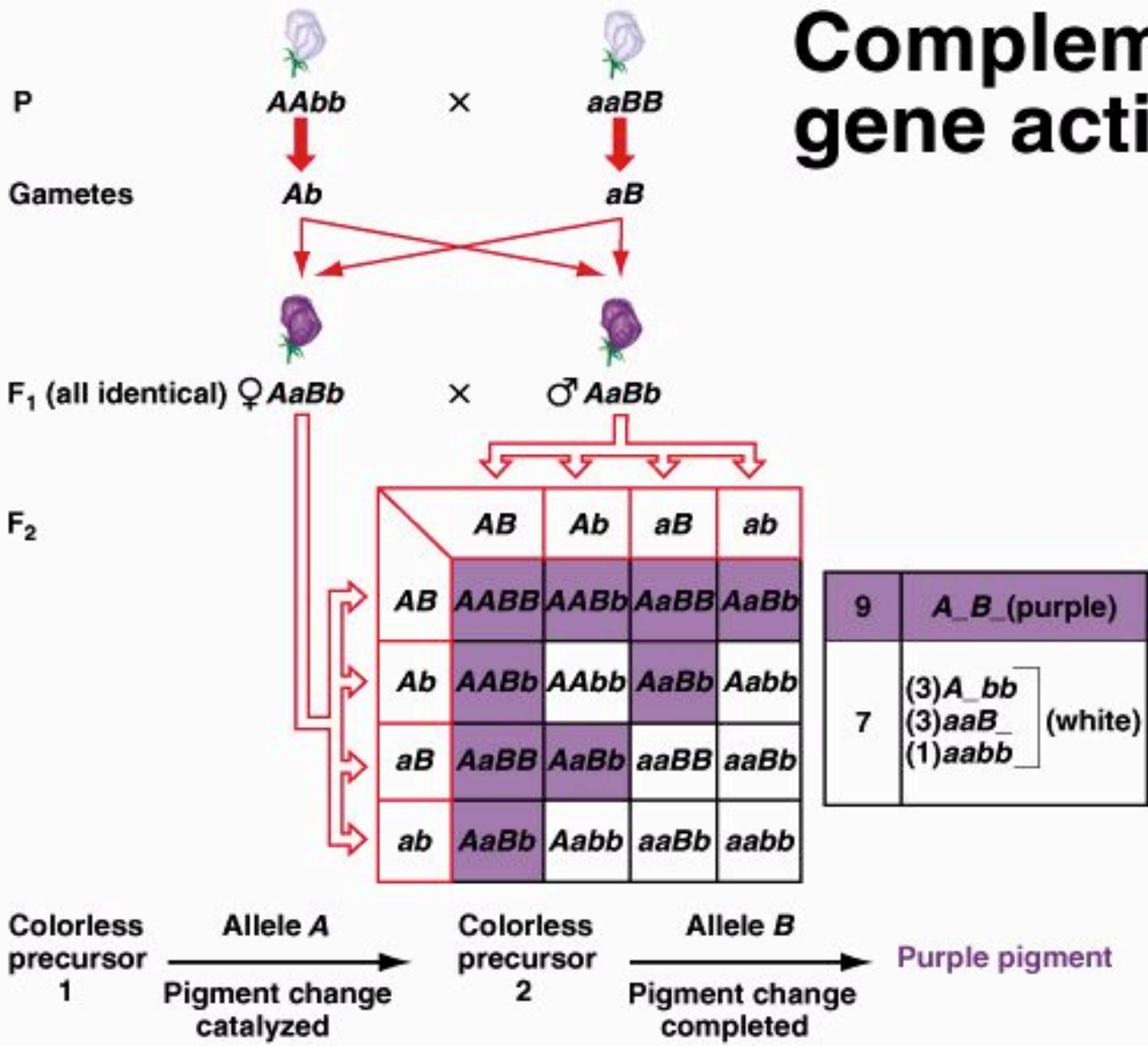
- **Dominance Relationships**:
  - Tan is dominant to green
  - Gray is dominant to green
  - Brown is dominant to gray, green and tan.
  - Tan and Gray are incompletely dominant, giving rise to brown.
- **Genotypic classes**:
  - Brown: A_B_
  - Tan: A_bb
  - Gray: aaB_
  - Green: aabb

# 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

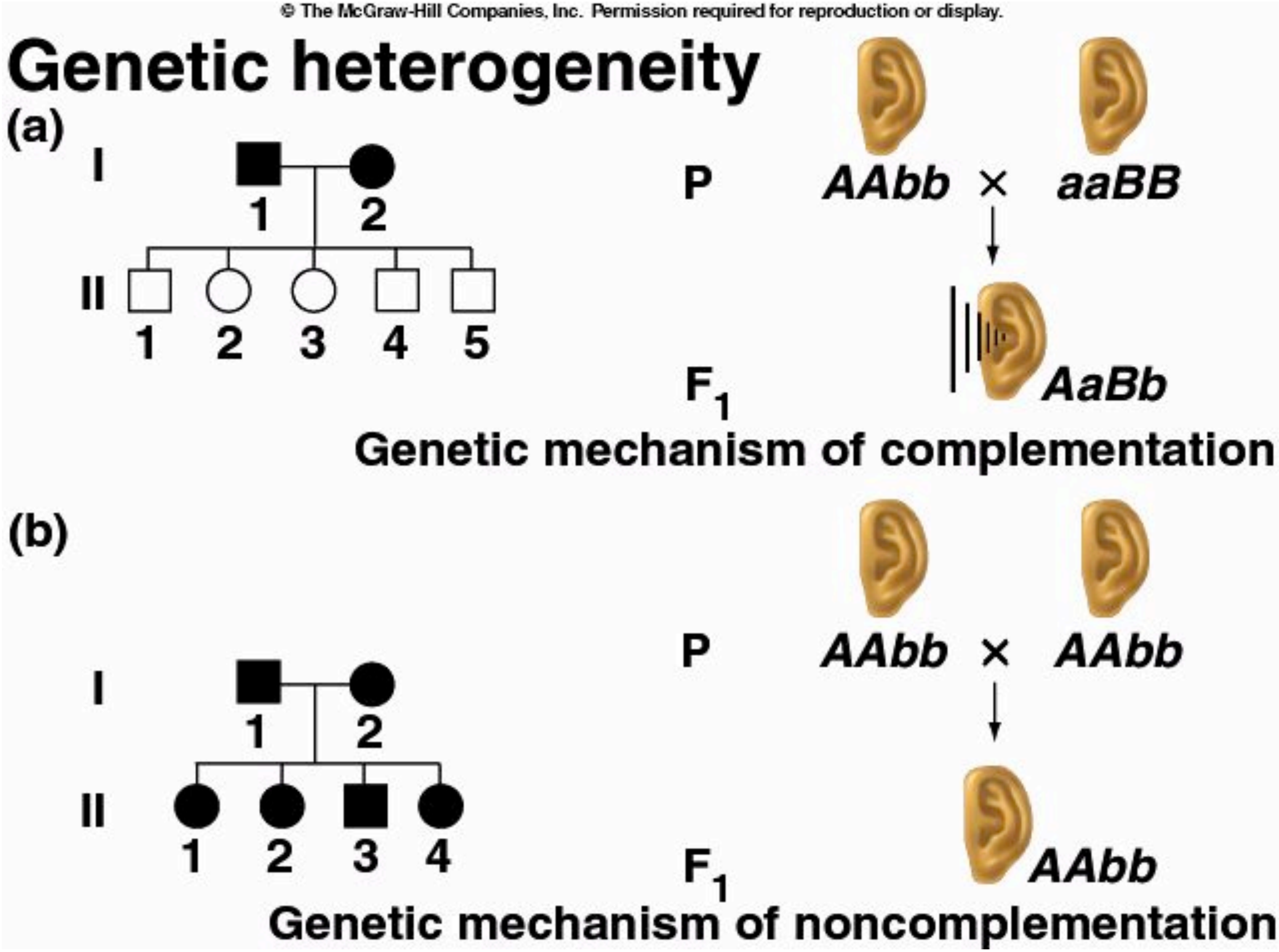
The must have a dominant allele in both genes to result in the purple flower phenotype

# 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*<br>(3)*aaB_*<br>(1)*aabb* (white) |



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Genetic heterogeneity
(a)
I
1
2
II
1 2 3 4 5
P
AAbb × aaBB
F1
AaBb
Genetic mechanism of complementation
(b)
I
1
2
II
1 2 3 4
P
AAbb × AAbb
F1
AAbb
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

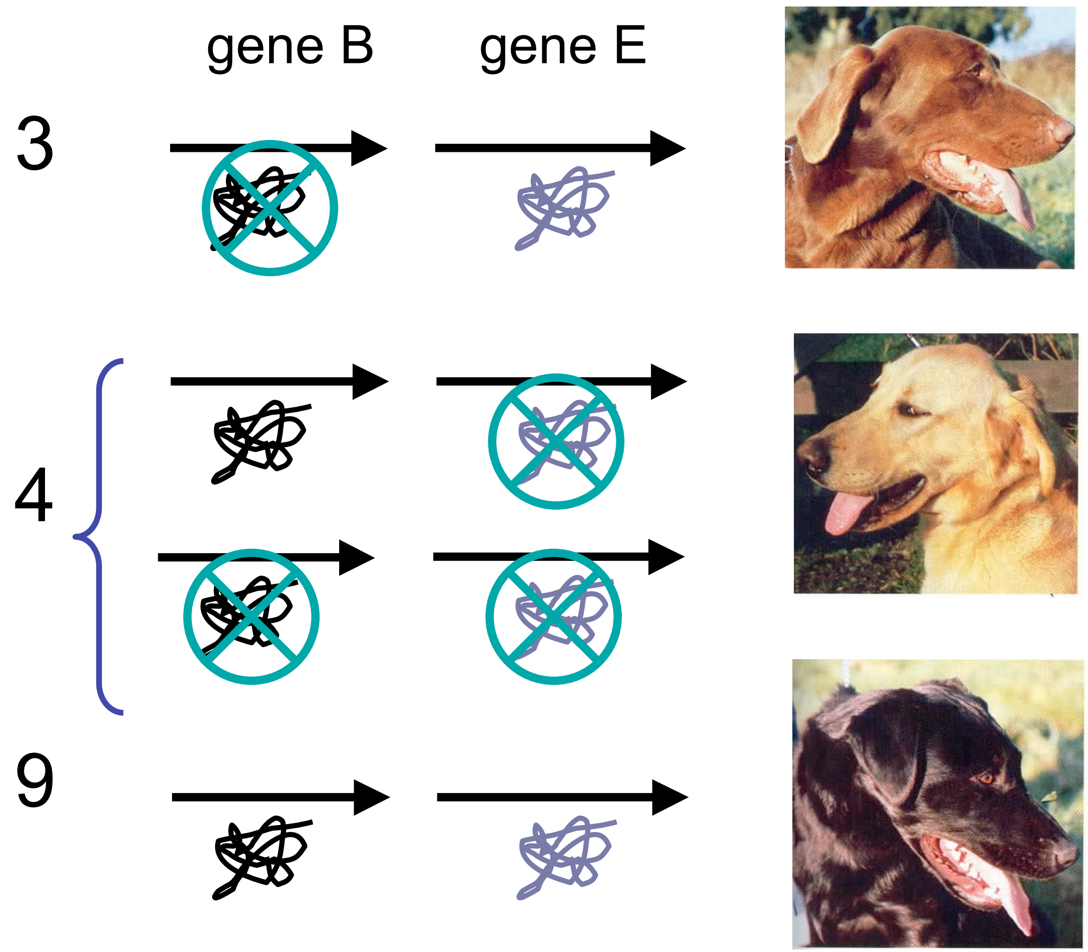
| | BE | Be | bE | be |
|---|---|---|---|---|
| BE | BBEE | BBEe | BbEE | BbEe |
| Be | BBEe | BBee | BbEe | Bbee |
| bE | BbEE | BbEe | bbEE | bbEe |
| be | BbEe | Bbee | bbEe | bbee |

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.

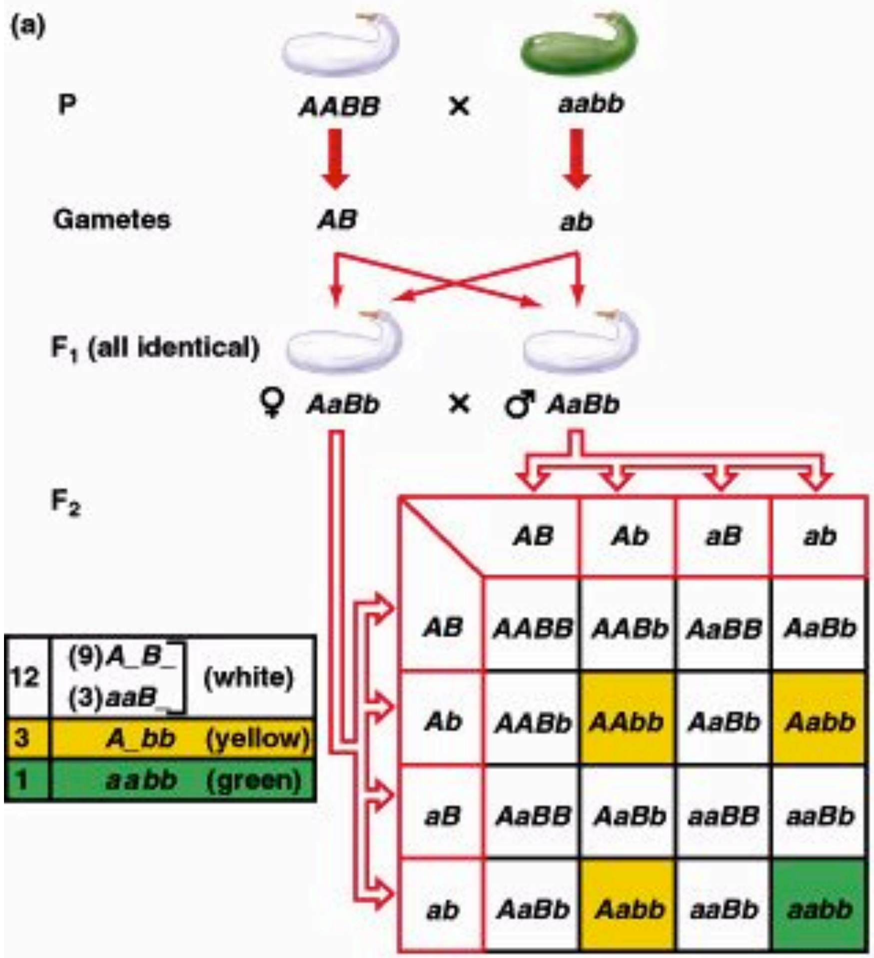
# Dominant Epistasis

- 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

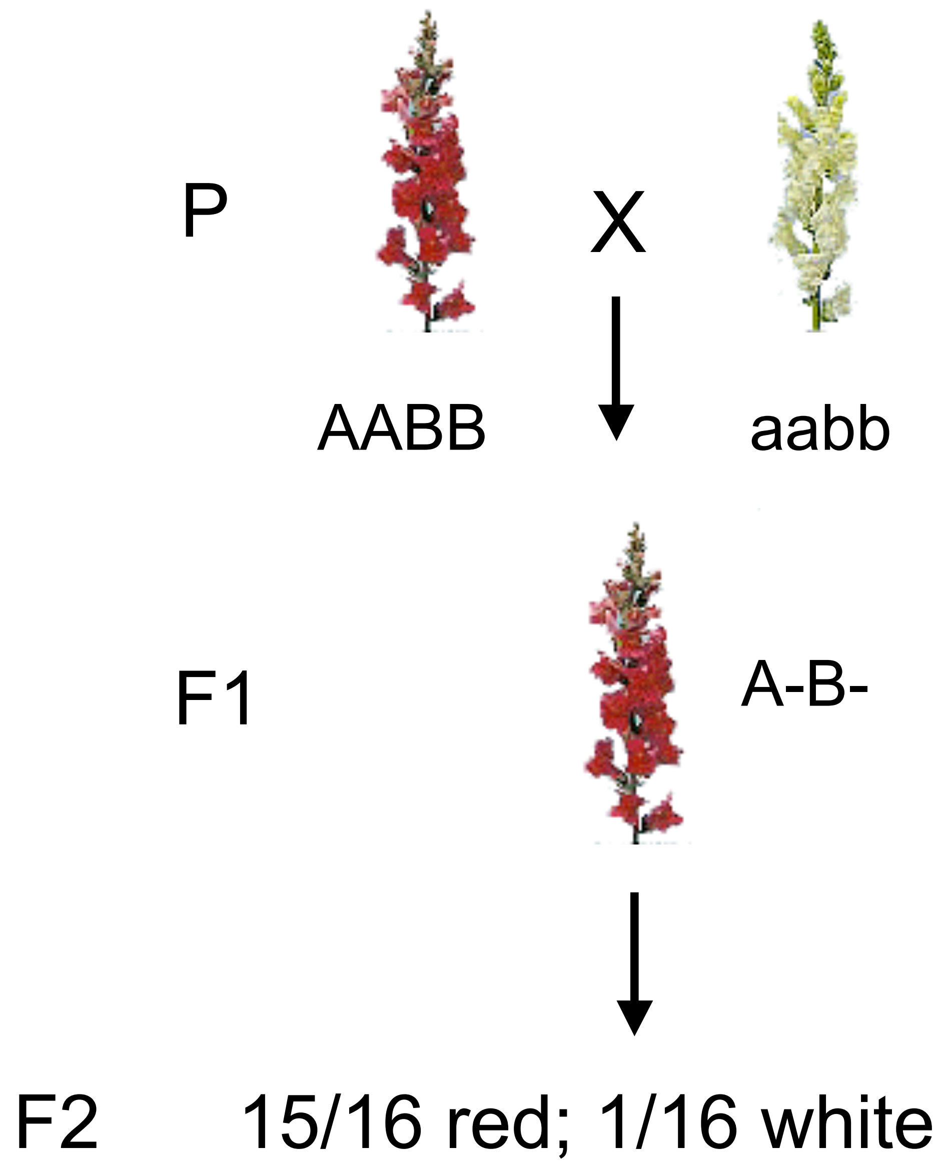
- Example: Summer Squash

# Dominant epistasis



# Redundancy: Duplicate Genes

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



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.*

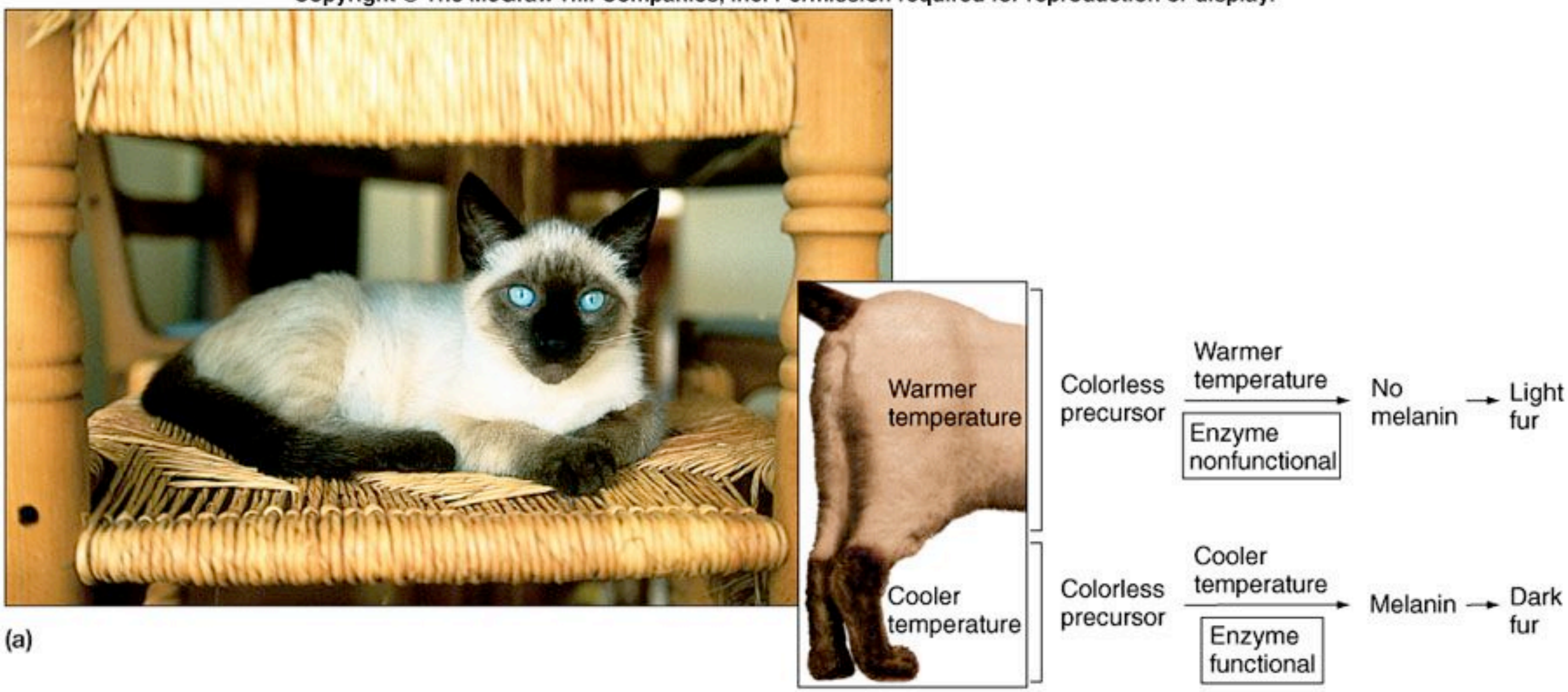
  - Retinoblastoma: some have both eyes affected, some only one.

# 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.)

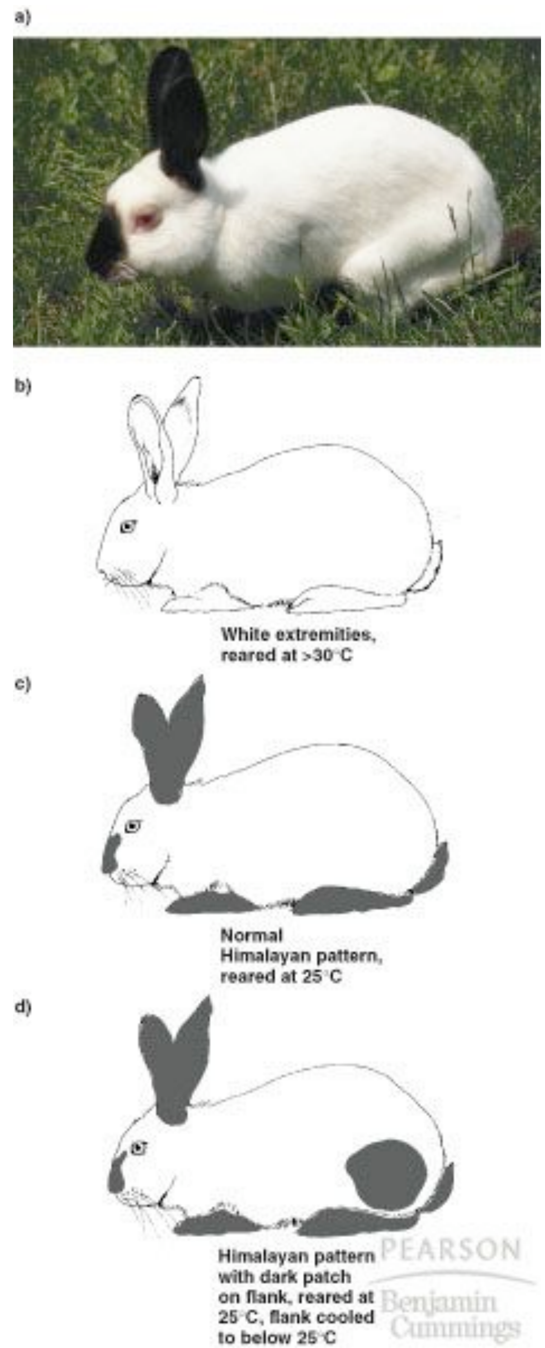
**Modifying environment**:
The environmental influence of a genotype on the phenotype= phenocopy






(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!!