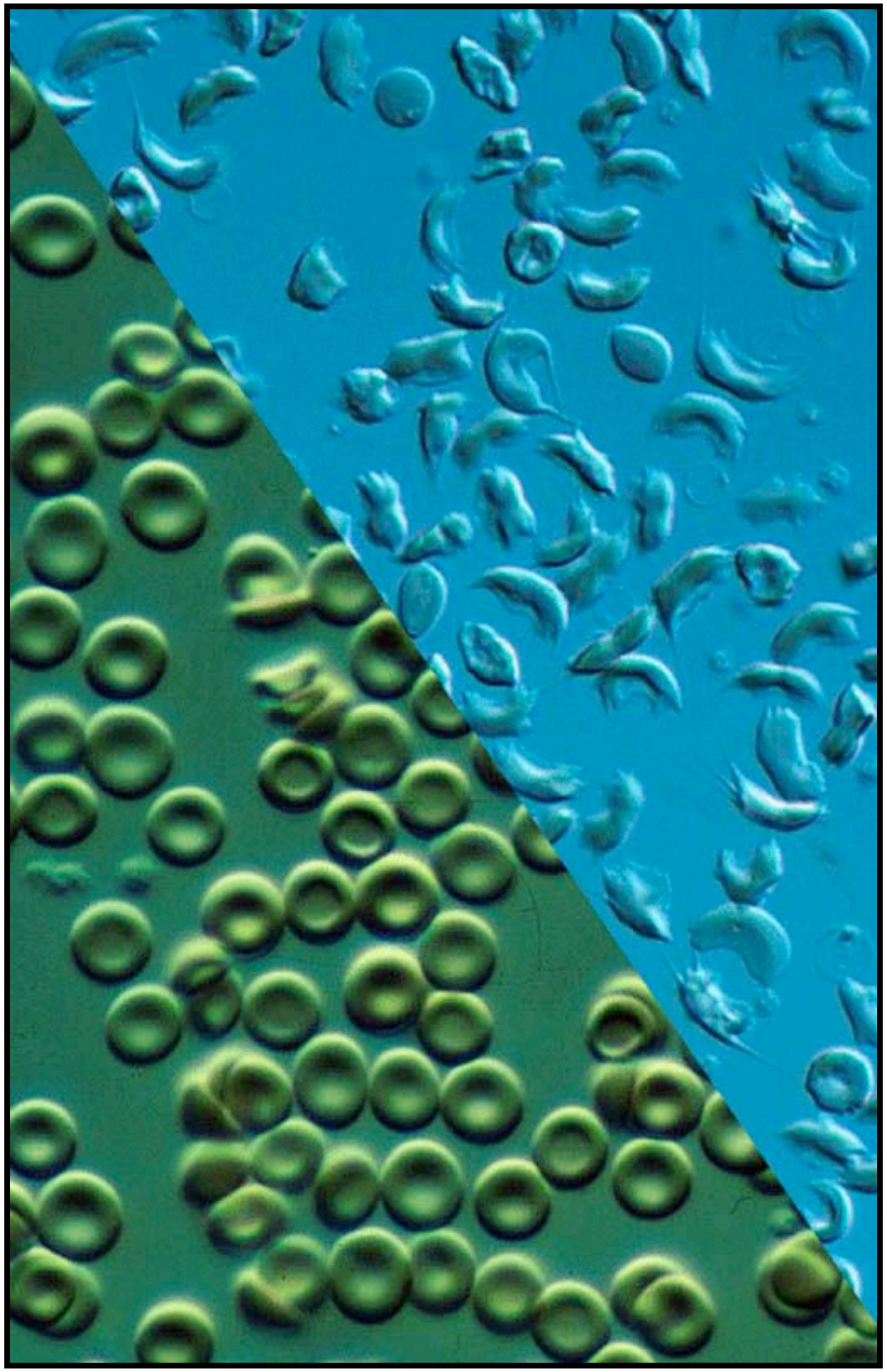
Chapter 4

❖Extensions to Mendelian Genetics

❖ Allele Interactions



INTRODUCTION

- Mendelian inheritance describes inheritance patterns that obey two laws
  - Law of segregation
  - Law of independent assortment

- Simple Mendelian inheritance involves
  - A single gene with two different alleles
  - Alleles display a simple dominant/recessive relationship

INTRODUCTION

- In this chapter we will examine traits that deviate from the simple dominant/recessive relationship

- The inheritance patterns of these traits still obey Mendelian laws
  - However, they are more complex and interesting than Mendel had realized

Variations of Mendelian Phenotypic Ratios

- alleles can interact with each other in complex ways
    - incomplete dominance
    - codominance
    - pleiotropic alleles
- many traits are controlled by more than one gene
    - epistasis
    - redundancy
    - complementation
    - penetrance
    - expressivity
- interactions of genes with the environment

So far we have looked at traits that are clearly dominant or recessive.

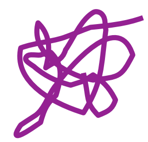
X

purple **white**

F1

purple

precursor → purple pigment



purple pigment forming enzyme

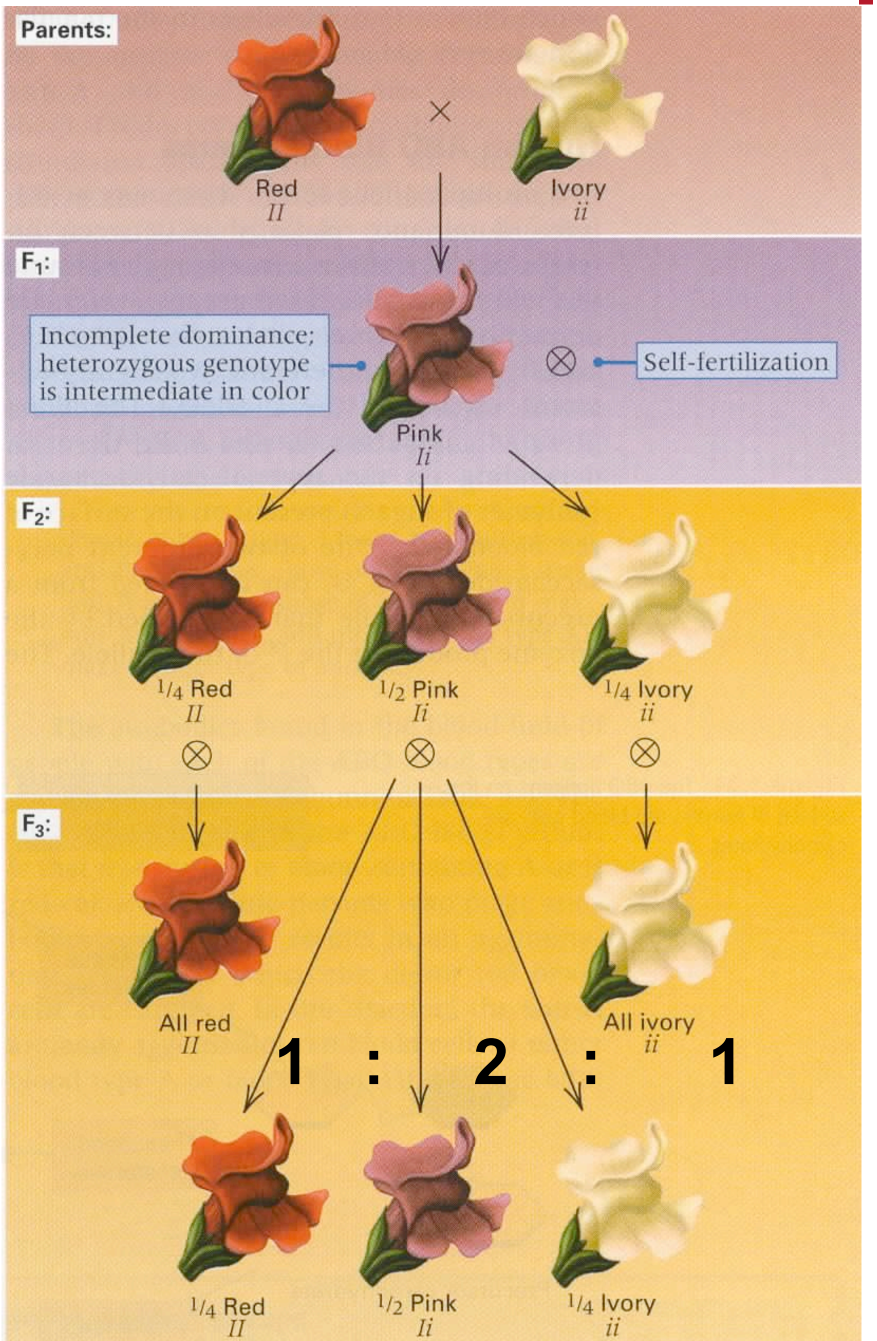
P = dominant allele makes a normal enzyme
p = recessive allele makes a defective one

- But nature is usually more complicated than this.
- Mendel was successful because he chose simple examples to start with (and he had some luck).

Dominance is not always complete

- Crosses between true-breeding strains can produce hybrids with phenotypes different from both parents
  - Incomplete dominance
    - Looks like neither parent
  - Codominance
    - Looks like both parents

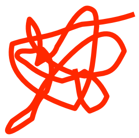
Incomplete Dominance



Incomplete Dominance:
the phenotype of the heterozygous hybrid is usually an intermediate between the homozygous parents.

- Segregation of both genotype and phenotype is 1:2:1.
- Alleles contribute different amounts of functional protein; total amount determines phenotype.

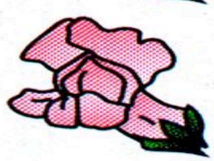
precursor $\longrightarrow$ red pigment



red pigment forming enzyme

A = dominant allele makes a normal enzyme
a = recessive allele makes a defective one

- The amount of red pigment present is dependent on the amount of enzymes present, which is dependent on the number of wildtype copies of the gene. This is a dosage effect.

	Phenotype	Genotype	Amount of gene product
	Red	AA	2x
	Pink	Aa	x
	White	aa	0

Incomplete Dominance

- Example: blooming time in peas.

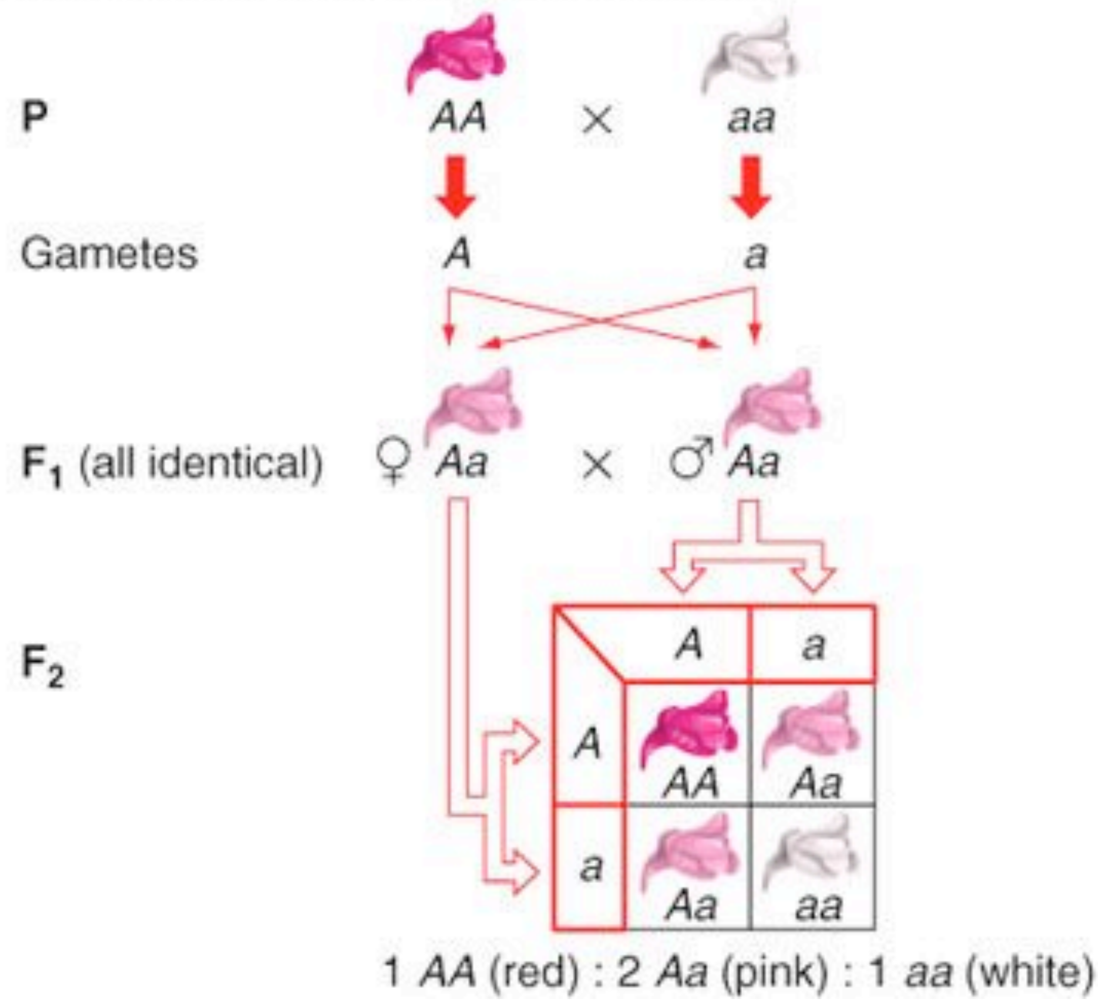
- Example: Flower color.
  - A cross of pure breeding red lowered plants and pure breeding white flowered plants yields plants with pink flowers.
  - Is this blending?
  - Check F2 generation ratios.

- Biochemical basis of incomplete dominance.

- ***Incomplete dominance is when neither of the two traits are seen in the F1 of a cross of two pure breeding traits.***

Incomplete dominance in snapdragons

(b) A Punnett square for incomplete dominance



Incomplete Dominance

- What is the phenotypic and genotypic ratio?
  - Genotype:
    - 1 AA: 2 Aa : 1 aa
  - Phenotype
    - 1 red : 2 pink : 1 white
  - Notice anything?
    - They are the same

- Usually use R and R' or A and A' because neither is dominant

Question

- In cattle, roan coloring is the result of crossing red with white cattle. What crosses would result in a pure line of roan cattle?
  - 1. Red x Red
  - 2. Red x White
  - 3. White x White
  - 4. Roan x Roan
  - 5. None

Codominance

- Both alternative traits can be visualized in the F1.
- Example: spotted lentil plant crossed to a dotted lentil plant.
- F1 hybrids look like both parents.
- Check phenotypic and genotypic ratios of F2.
- ***Codominance is when both alternative traits are expressed in the F1 of a cross of two pure breeding parents.***

Codominant lentil coat patterns

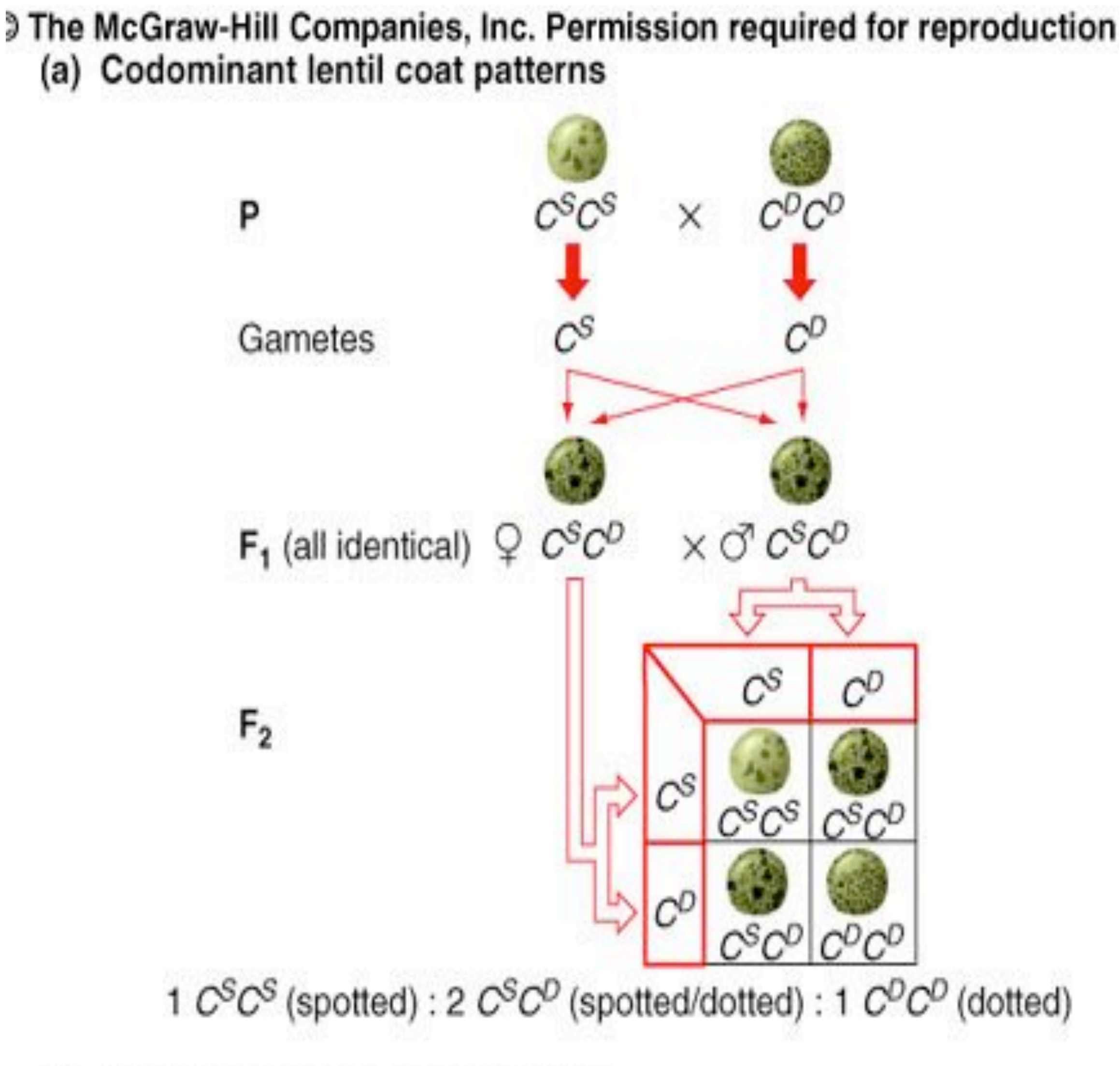


Fig. 3.4a

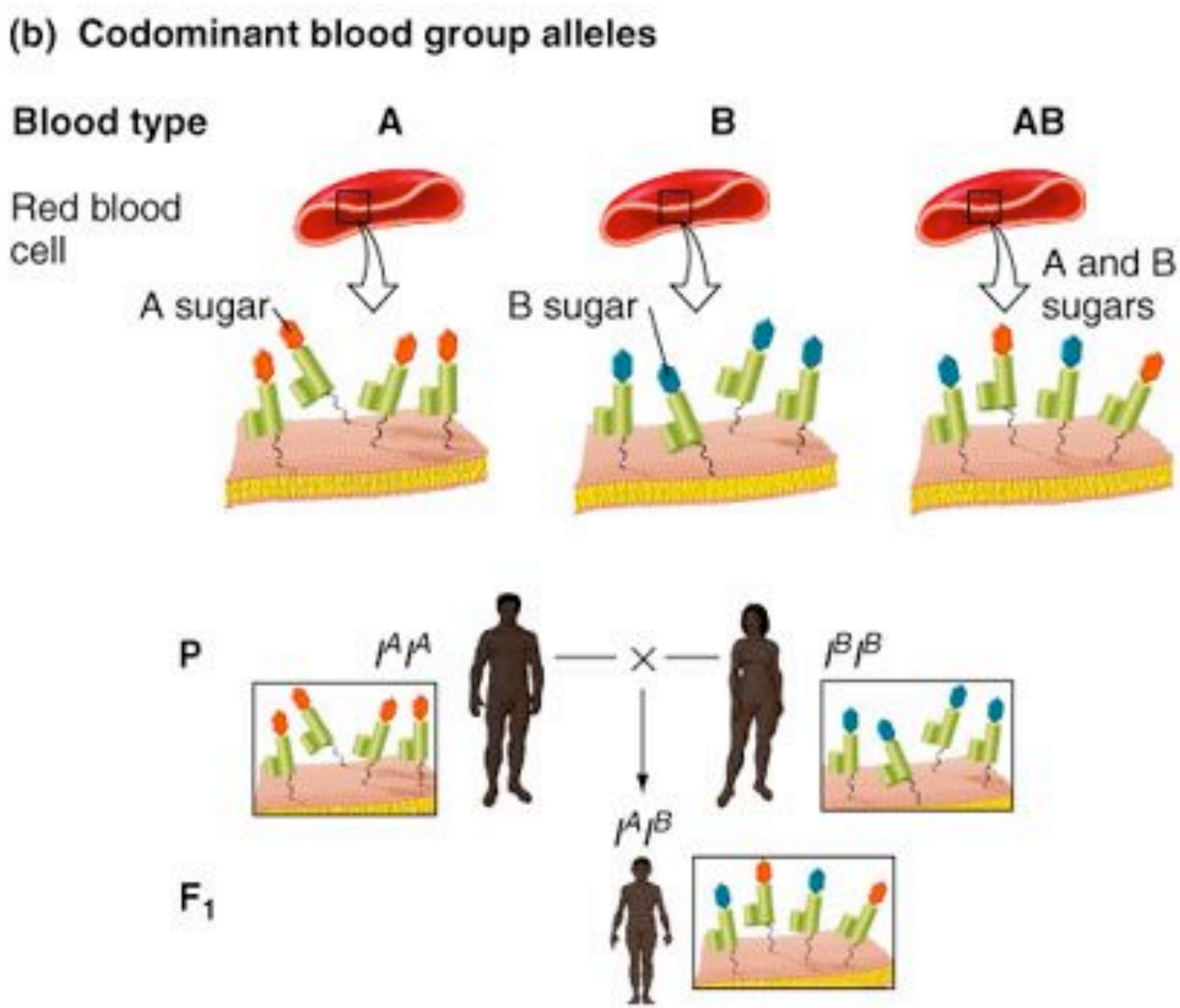
Co-Dominance

- What is the phenotypic and genotypic ratio?
  - Genotype:
    - 1 CsCs: 2 CsCd : 1 CdCd
  - Phenotype
    - 1 spotted : 2 spotted and dotted : 1 dotted
  - Again, notice anything?
    - They are the same
- In this case both alleles expressed

Do variations on dominance relations negate Mendel's law of segregation?

- Dominance relations affect phenotype and have no bearing on the segregation of alleles
- Alleles still segregate randomly
- Gene products control expression of phenotypes differently
- Mendel's law of segregation still applies
- Interpretation of phenotype/genotype relation is more complex

Codominant blood group alleles



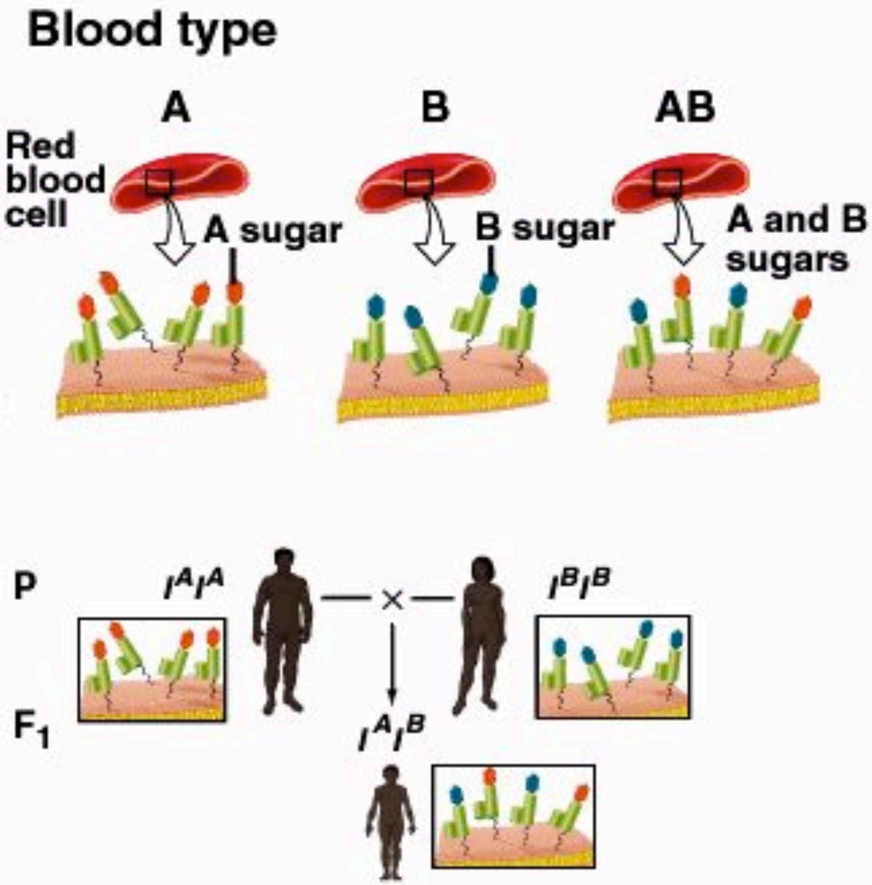
Multiple Alleles

- A gene can have more than two alleles
  - How many do we inherit?
    - Just two…..but can pull from more than two in the gene pool
- Back to blood type
  - How many are there?
  - Three alleles
    - IA
    - IB
    - *i*

ABO Blood Types in Humans

- Three different alleles: I^A, I^B and *i*.
- There are 4 different blood group phenotypes: A, B, AB and O.
- Each pair has a different dominance relationship.
- Biochemical Basis:
  - Complex membrane anchored molecules that cause a variation in the structure of the sugar polymer on the cell surface.
  - Three possibilities: Sugar type A, B or none.

ABO blood types



Genotypes	Corresponding Phenotypes: Type(s) of Molecule on Cell
I^AI^A I^Ai	A
I^BI^B I^Bi	B
I^AI^B	AB
ii	O

ABO Antigens

- **Type A individuals have the A antigen on their RBCs, and anti-B antibodies in their blood.**
  - **Their genotype is I^A/I^A or I^A/i.**
- **Type B individuals have the B antige**
  - **n on their RBCs, and anti-A antibodies in their blood. Their genotype is I^B/I^B or I^B/i.**
- **Type AB individuals have both the A and the B antigen on their RBCs, and neither anti-A nor anti-B antibodies in their blood.**
  - **Their genotype is I^A/I^B.**
- **Type O individuals have neither the A nor the B antigen on their RBCs, and both anti-A and anti-B antibodies in their blood.**
  - **Their genotype is i/i.**

Question

- Which blood type is the universal donor?
  - 1) A
  - 2) B
  - 3) AB
  - 4) O

Question

- Which blood type is the universal acceptor?
  - 1) A
  - 2) B
  - 3) AB
  - 4) O

Question

- In a paternity suit, the child in question has blood type A and the mother is type AB, which of the following men could not possibly be the child's father? A father with type ________ blood.
  - 1. A
  - 2. B
  - 3. AB
  - 4. O
  - 5. All of the above could possibly be the father.

Dominance series

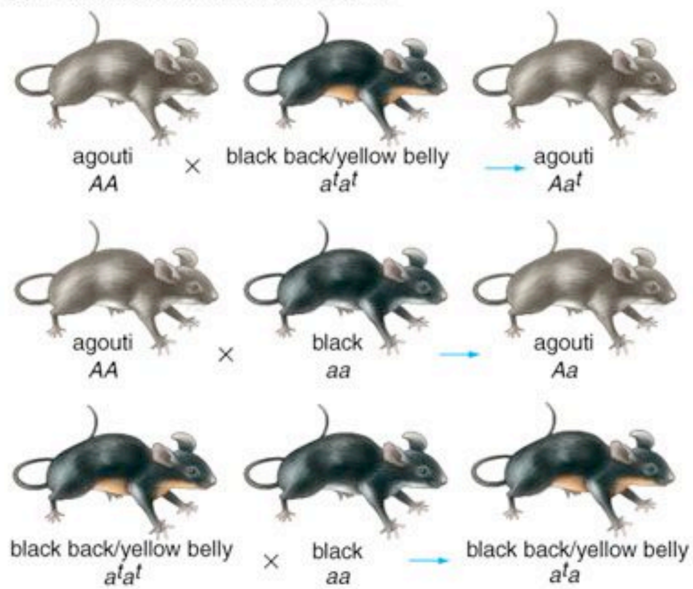
- Alleles are listed in order of most dominant to most recessive
- Establishes dominance relationships between multiple alleles
- Crosses between trre breeding lines allow arrangements
- Interbreeding F1 hybrids results in 3:1 ratios
  - Says alleles are in fact alleles of the same gene

(a) ***Mus musculus*** **(house mouse) coat colors**

(b) Alleles of the *agouti* gene

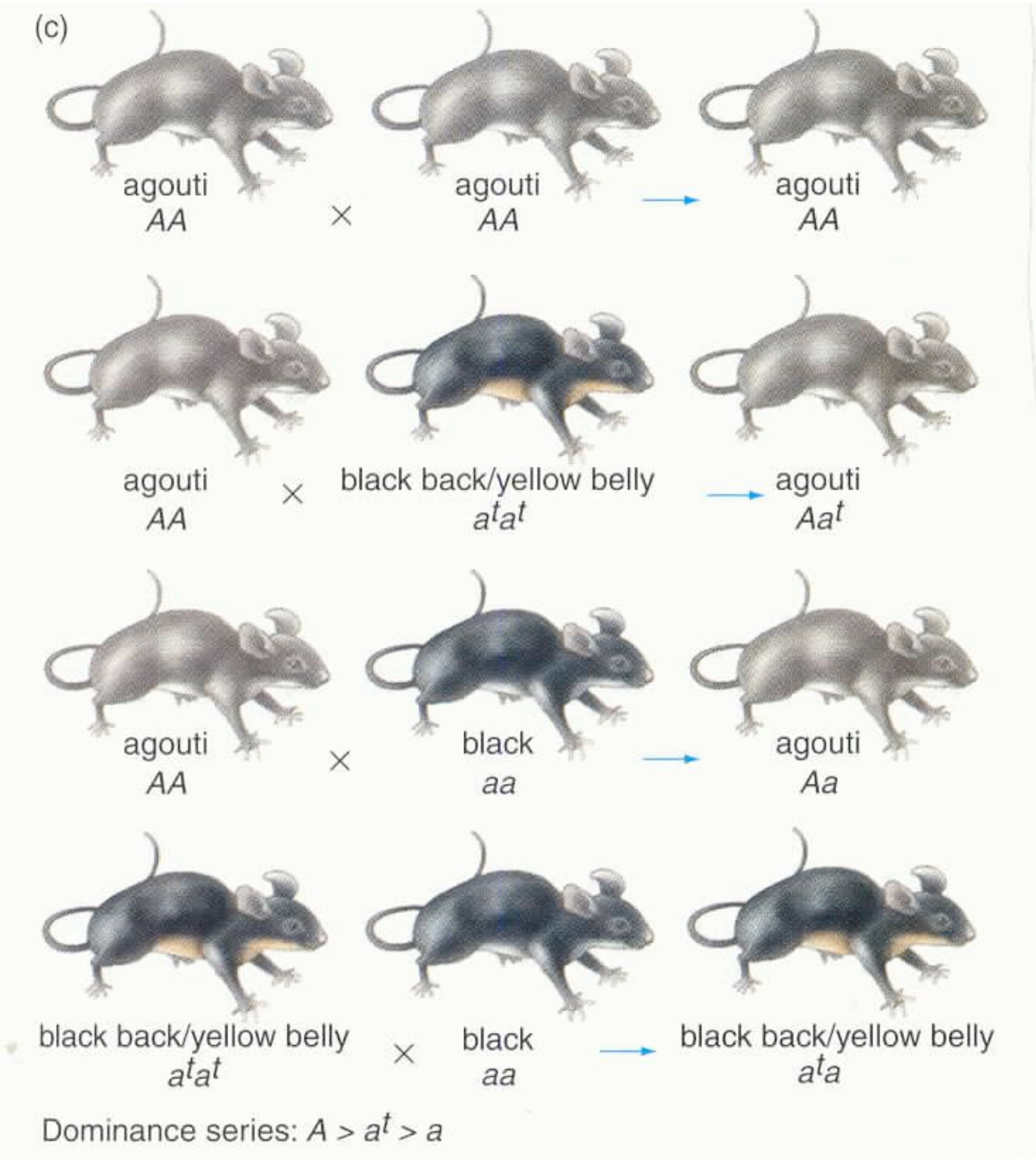
Genotype	Phenotype
$A-$	agouti
a^ta^t	black/yellow
aa	black

(c) Evidence for a dominance series



Dominance series: $A > a^t > a$

Dominance Series of Multiple Alleles



- There are multiple alleles for the gene that codes for coat color in mice.
- Using pair wise crosses of mice homozygous for each of the sixteen agouti alleles one, can construct a dominance series.
- The dominance series describes a hierarchical relationship between the various alleles of a single gene.

Pleiotropy

- One gene may contribute towards several visible characteristics.
  - E.g: coat color in mice
- In addition to coat color, a specific allele at this locus also causes lethality.
- ***The phenomenon of a single gene determining multiple distinct traits is known as pleiotropy.***
- Pleiotropy may be the result of a single gene product (protein) giving rise to multiple functional characteristics.
  - Examples:
  - Recessive lethal alleles
  - Sickle Cell Syndrome

The A^Y Allele Is Pleiotropic

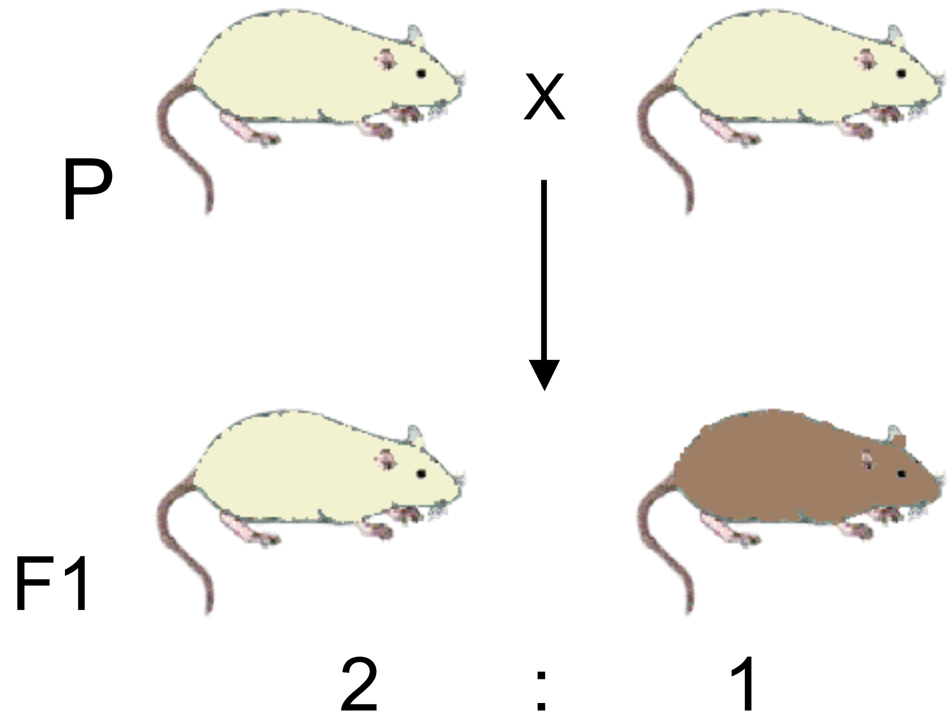
<u>Pleiotropic</u>: multiple physical effects (phenotypes) caused by a single altered gene or pair of altered genes.

A^Y: mutation, yellow coat

A^Y/A mice are yellow, obese, and develop tumors and type 2 diabetes

A/A: wild-type agouti (brown)

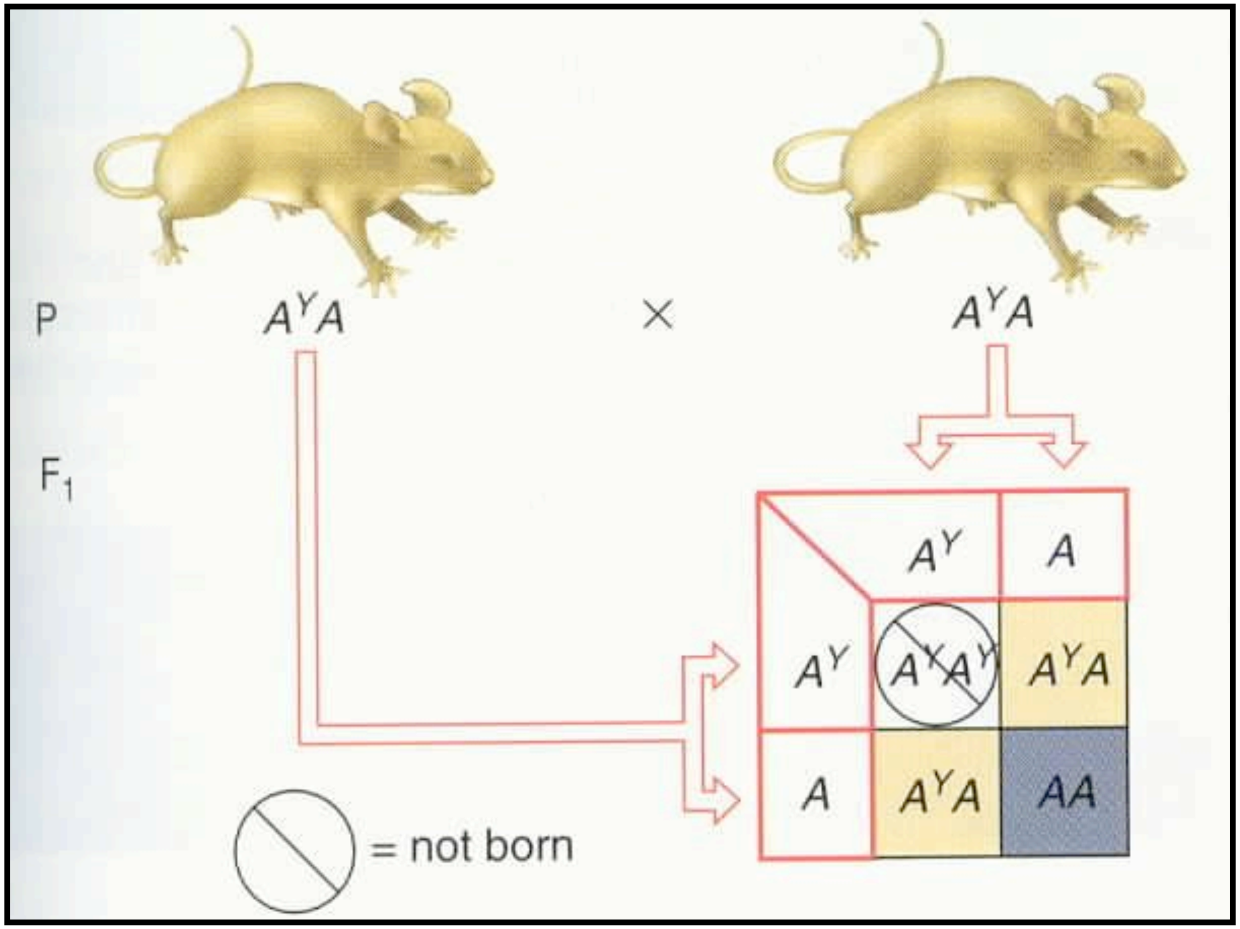
A^Y/A^Y die *in utero*



always get 2 yellow : 1 wild-type

pure breeding yellow mice can never be obtained

Genetic Explanation



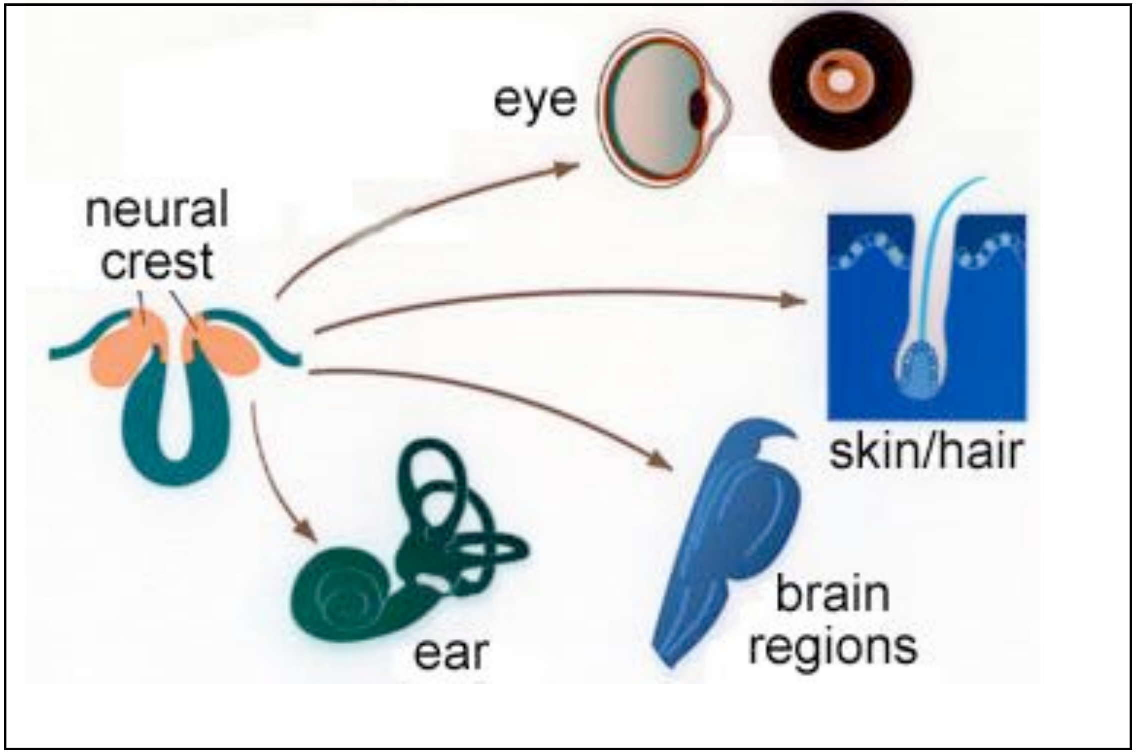
- Homozygous A^Y/A^Y mice die in utero and are never observed; it is a lethal phenotype.
- Yellow mice are heterozygotes.
- Homozygous recessive (wild type) mice are agouti.

- A^Y allele is <u>dominant</u> over the A normal allele for coat color
- but is <u>recessive</u> for lethality.
- Dominance depends on what trait you look at.

Another example: White coat color and blue eyes in dogs and cats is often co-inherited with deafness. Pleiotropic genes have mutant phenotypes that disrupt more than one process, even seemingly unrelated ones, such as pigment formation and ear development.

Pleiotropy

Pleiotropy



- Hereditary congenital deafness in dogs (and cats) is
  - associated with defects in neural crest cells
    - which are precursors for both
      - melanocytes
        - required for pigmentation of the coat and eyes
      - and structures of the inner ear.

Conclusions:

- Alleles of a single gene can interact with other alleles of the same gene or with the environment.

- When heterozygous offspring look like one parent but not the other -
    - complete dominance, dominance series.

- When heterozygotes show a phenotype unlike that of either parent -
    - incomplete dominance.

- When heterozygotes show characteristics of both parents
    - co-dominance.

Question

- When the same gene is related to respiratory problems and sterility, it can be described as:
  - 1. pleiotropy
  - 2. co-dominance
  - 3. Incomplete dominance
  - 4. Complete dominance

Summary

TABLE 3.1 For Traits Determined by One Gene: Extensions to Mendel's Analysis Explain Alterations of the 3:1 Monohybrid Ratio

What Mendel Described	Extension	Extension's Effect on Heterozygous Phenotype	Extension's Effect on Ratios Resulting from an $F_1 \times F_1$ Cross
Complete dominance	Incomplete dominance Codominance	Unlike either homozygote	Phenotypes coincide with genotypes in a ratio of 1:2:1
Two alleles	Multiple alleles	Multiplicity of phenotypes	A series of 3:1 ratios
All alleles are equally viable	Recessive lethal alleles	No effect	2:1 instead of 3:1
One gene determines one trait	Pleiotropy: one gene influences several traits	Several traits affected in different ways, depending on dominance relations	Different ratios, depending on dominance relations for each affected trait

Homework Problems

- Chapter 4
- # 1, 4, 6, 7, 8, 9, 10, 11, 12, 13, 14

- DON'T forget to submit the online iActivity
  - "Charlie Chaplin"