Chapter 14

Mendel and the Gene Idea

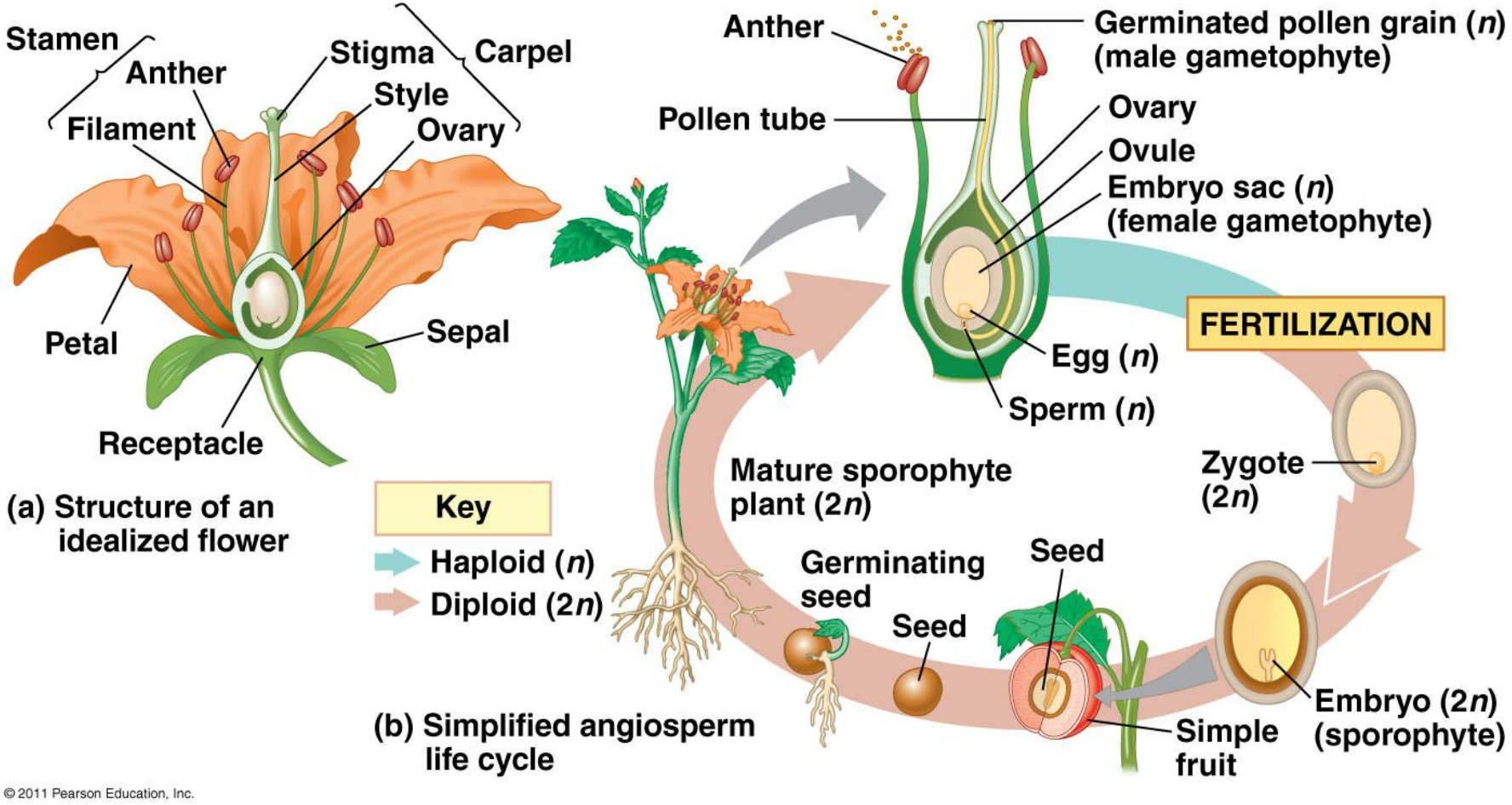
Mendel's Background

- Born in what is now the Czech Republic to a poor farming family
- Moved away from home at 11 to get an education, fought depression several times and had to be sent home
- Joined the monastery and was able to get his education paid for; he studied physics, chemistry, and zoology
- He pursued his scientific investigations and even taught high school

Mendel’s experimental setup

- What organism did he use?
- What made his experiment good?
- What were his controls?

Angiosperm Reproduction



Hybridization

- Mating of 2 true-breeding varieties obtained from self-pollination

P generation (parental)

↓ - did cross-pollination

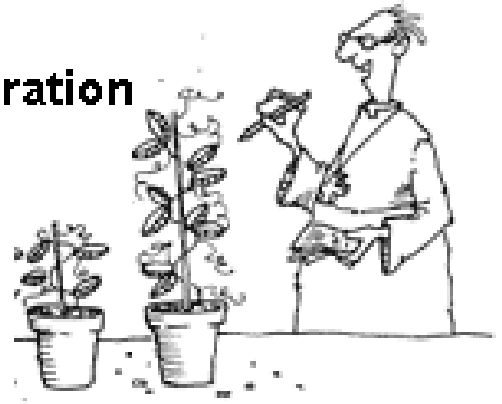
F_1 generation (1^{st} filial)

↓ - did self-pollination

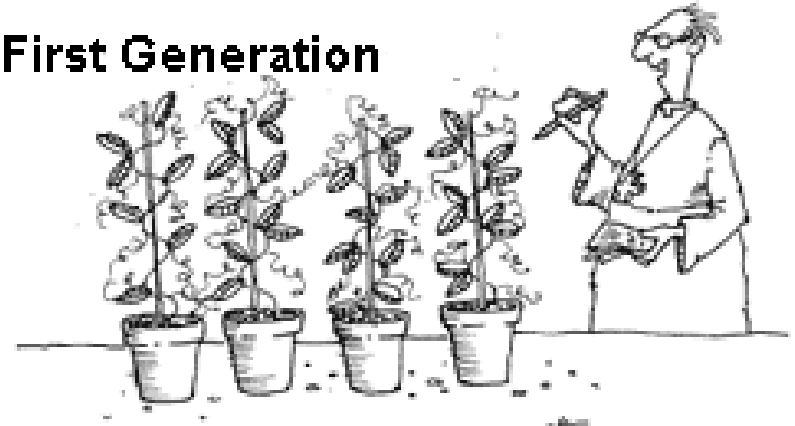
F_2 generation (2^{nd} filial)

Visualizing Mendel's Experements

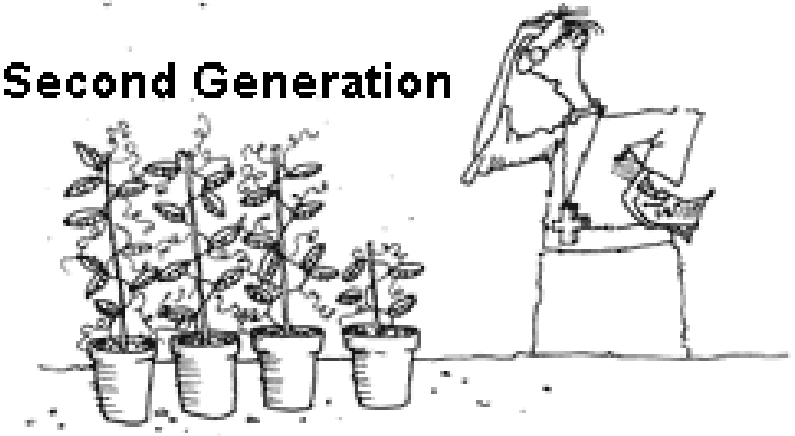
Parent Generation



First Generation



Second Generation



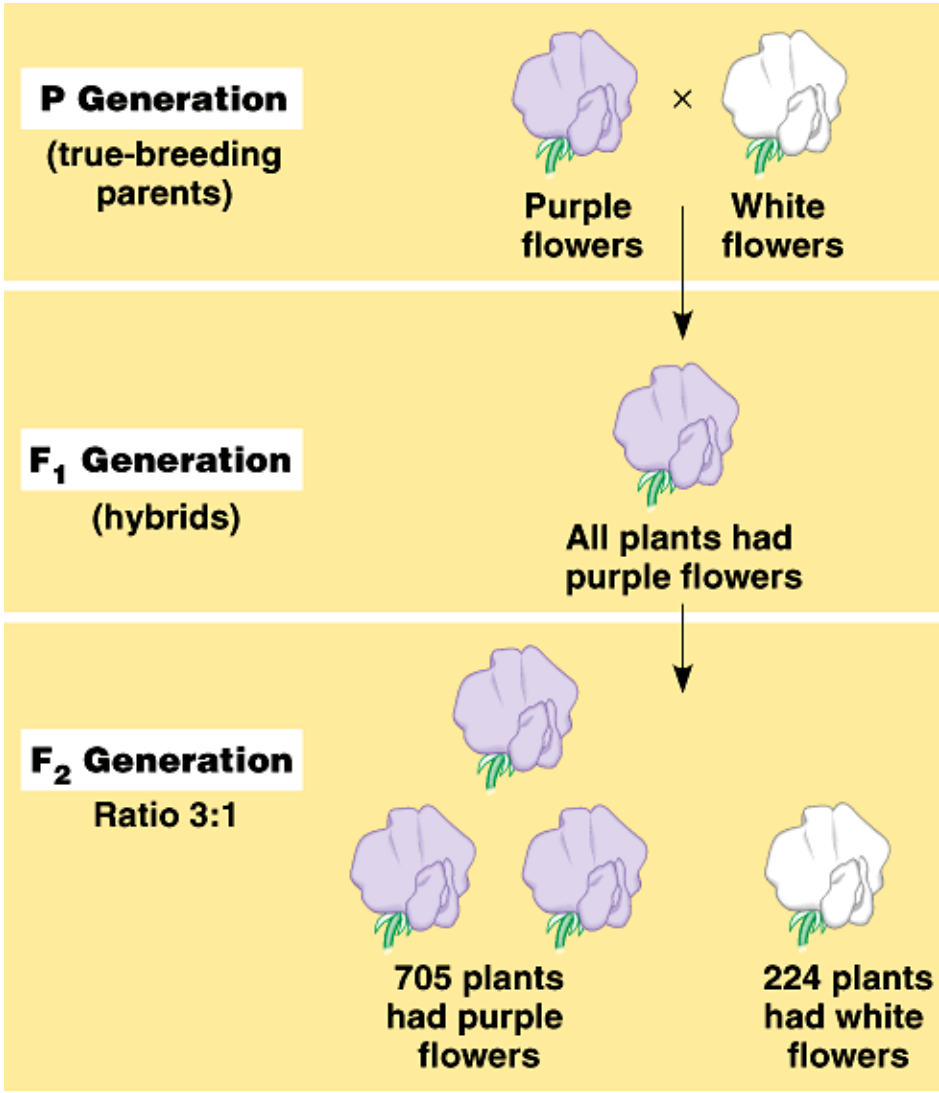
Mendel and The Gene Idea

- Gregor Mendel is known as the "Father of Genetics."
- Mendel's two fundamental principles of heredity are now known as the:
  - *law of segregation*
  - *law of independent assortment*.

Mendel's Model

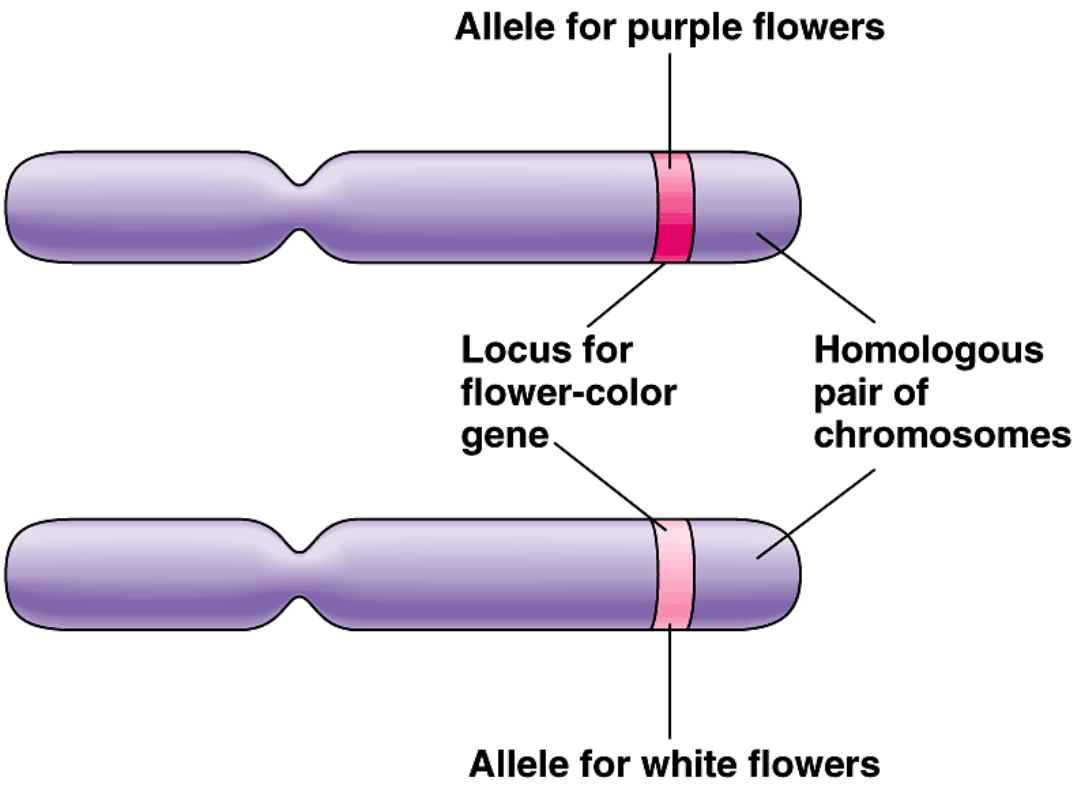
- Mendel knew that the white trait did not disappear in the F_1 generation

- *Alleles* – alternative versions of genes account for variations in inherited characters



Mendel's Model

- Each gene is located at a specific *locus* on a specific chromosome.
- The DNA at that locus, however, can vary somewhat in its sequence of nucleotides and hence in its information content.



Mendel's Model

- For each character, an organism inherits two alleles, one from each parent. *(Wasn't aware of the role of chromosomes at this time)*
  - *Dominant allele* – fully expressed in organism's appearance
  - *Recessive allele* – no noticeable effect on the organism's appearance.

Law of Segregation

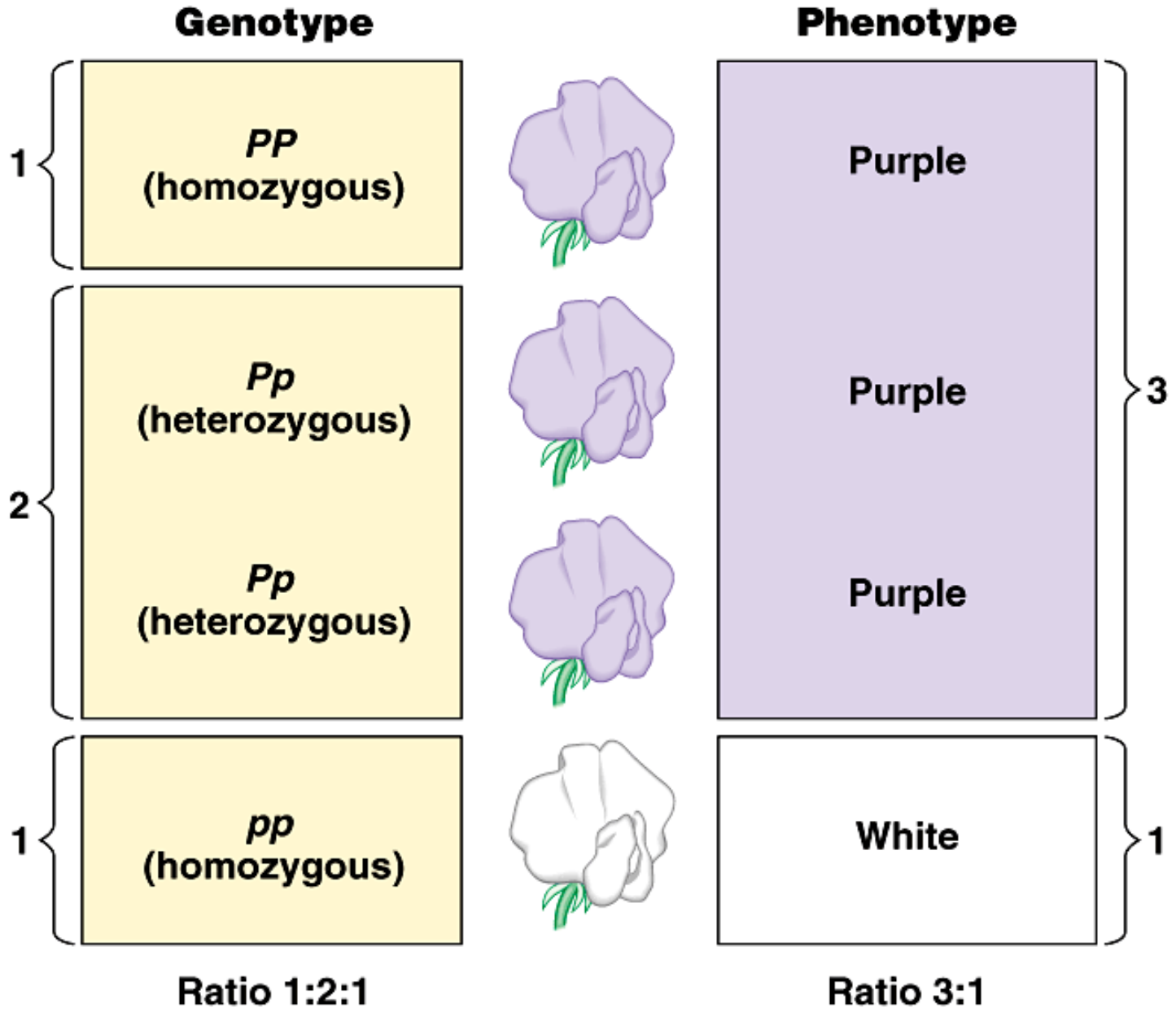
- **States: The two alleles for each character segregate or separate during gamete production (meiosis)**
  - Sister chromatids separate and so do the alleles of the genes on the chromosomes

Some Useful Genetic Vocabulary

- *Homozygous* – an organism having a pair of identical alleles for a character (PP or pp)
- *Heterozygous* – an organism having two different alleles for a gene (Pp)

- *Phenotype* – an organism's traits or physical appearance (purple or white flowers)
- *Genotype* – an organism's genetic makeup (PP, pp, or Pp)

Genotype vs. Phenotype



Law of Independent Assortment

- **States: Each pair of alleles segregates independently of other pairs of alleles during gamete formation**

- In pea plants, flower color is independent of seed color, is independent of seed-shape character, etc.

Law of Independent Assortment

- This law only applies to genes (allele pairs) that are located on non-homologous chromosomes.

- Genes located near each other on the same chromosome tend to be inherited together – linked genes

Punnett Squares

- Chart to show the probability of certain traits occurring in offspring
  - *Monohybrid cross* – mating parents with one characteristic
    - BB X bb
  - *Dihybrid cross* – mating parents with two characteristics
    - BBRR X bbrr

Monohybrid Cross

- Write out all the monohybrid cross results using the following:
  - B = brown eyes
  - b = blue eyes

- Be sure to include the genotypic and phenotypic ratios or percentages

Dihybrid Cross

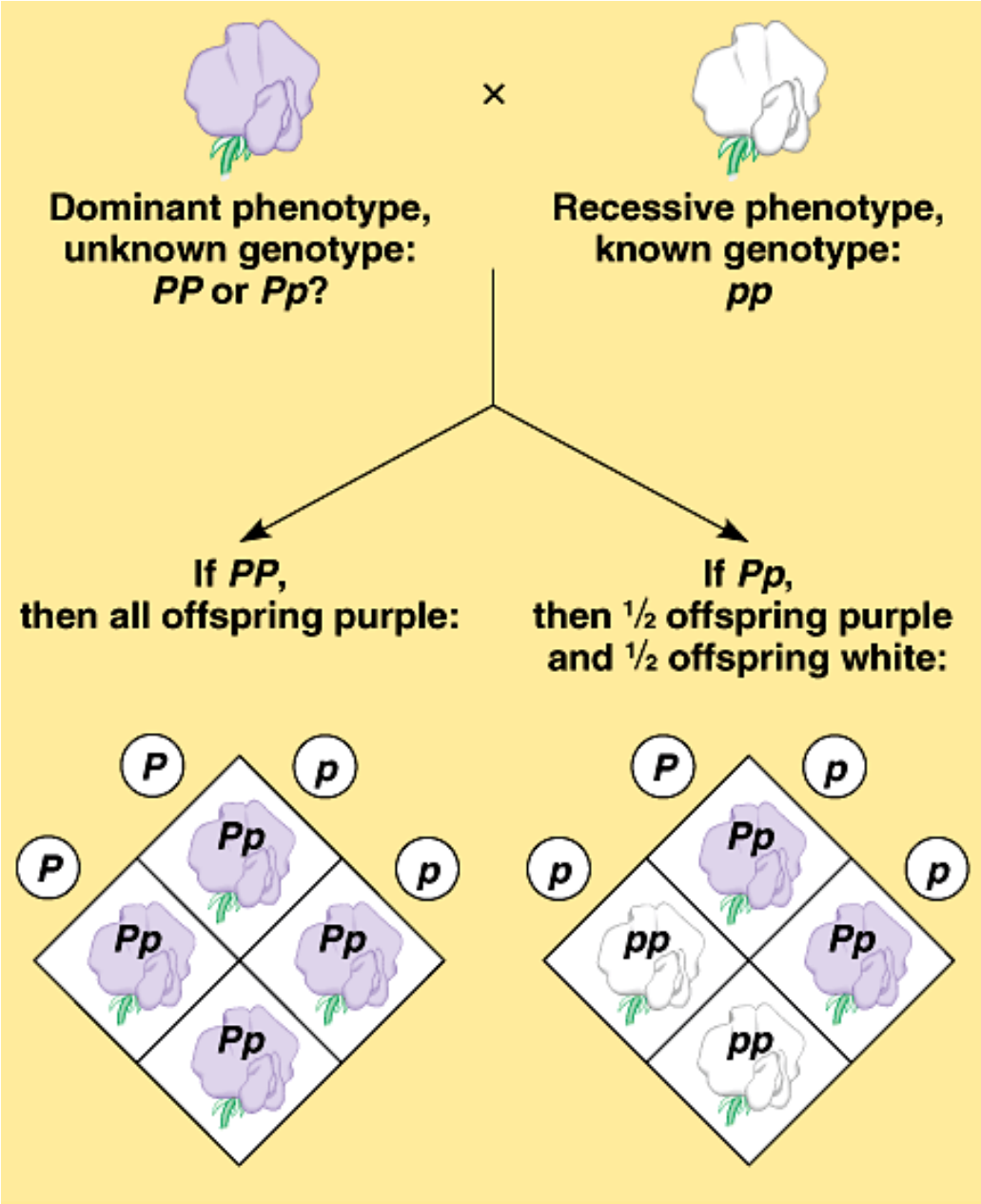
- Complete the following 2 crosses using
  - B = brown eyes
  - b = blue eyes
  - T= tall
  - t = short

- Parent 1: Homozygous dominant for both traits crossed with Parent 2: Homozygous recessive for both traits
- Parent 1: Heterozygous for both traits crossed with Parent 2: Heterozygous for both traits

Testcross

- How can we determine if a pea plant with purple flowers is PP or Pp?

Testcross


×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
If PP, then all offspring purple:
If Pp, then ½ offspring purple and ½ offspring white:
P
p
P
p
Pp
Pp
Pp
Pp
P
p
p
p
Pp
pp
Pp
pp

Laws of Probability

- Multiplication Rule:
  - To determine the probability of two or more independent events occurring together, multiply the probability of one event by the probability of another event
  - Ex) Two coins landing on heads = ½ X ½ = ¼

Laws of Probability

- Addition Rule:
  - To determine the probability that any one of two or more mutually exclusive events will occur is calculated by adding together their individual probabilities
  - Ex) F2 heterozygote from two heterozygous parents = ¼ + ¼ = ½
    - Dominant from egg and recessive from sperm OR Recessive from egg and dominant from sperm

Probability Problems

- Cross BbRr X BBrr
  - What is the probability of getting BBRr?

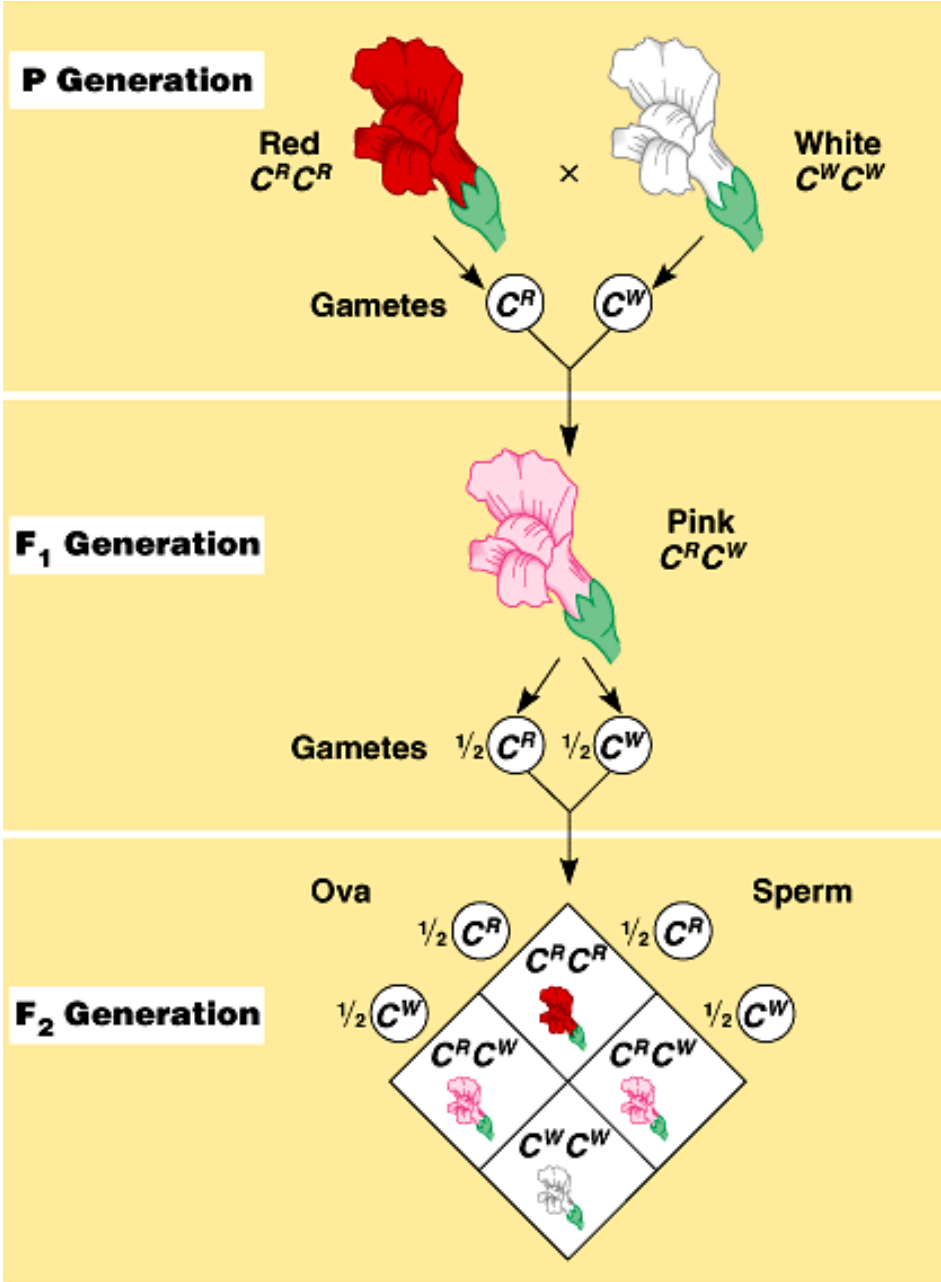
- Cross DdTT X ddtt
  - What is the probability of getting at least one recessive trait?

Probability Problems

- Cross PpYyRr X Ppyyrr
- What is the probability of getting PPyyRr?
- What is the probability of getting offspring that were heterozygous for all traits or homozygous recessive for all traits?

Incomplete Dominance

- The F_1 hybrids have an appearance somewhere in between the phenotypes of the two parental varieties.
- Example: Snapdragons



Codominance

- Two alleles affect the phenotype in separate, distinguishable ways
- Example: Blue Roan Horse
  - Horse has a mixture of black and white hair, giving the horse a bluish appearance, rather than a gray color.

- Example: Andalusian Chicken
  - Has an equal number of black and white feathers, resulting in a bluish appearance.

Dominant/Recessive Relationships

1. Range from complete dominance through various degrees of incomplete dominance, to codominance.
2. Reflect mechanisms by which specific alleles are expressed in the phenotype and do not involve the ability of one allele to subdue another at the level of the DNA.
3. They do no determine or correlate with the relative abundance of alleles in a population.

Relationship Depends on Level of Phenotype Examined

- Organismal level – Tay-Sachs allele is recessive
- Biochemical level – Activity level of the enzyme is intermediate (incomplete dominance) because half the normal activity is enough to not express the Tay-Sachs disease
- Molecular level – heterozygous individuals produce equal amounts of the normal and dysfunctional enzymes (codominance)

- All this to say that human genetics is VERY complex!!

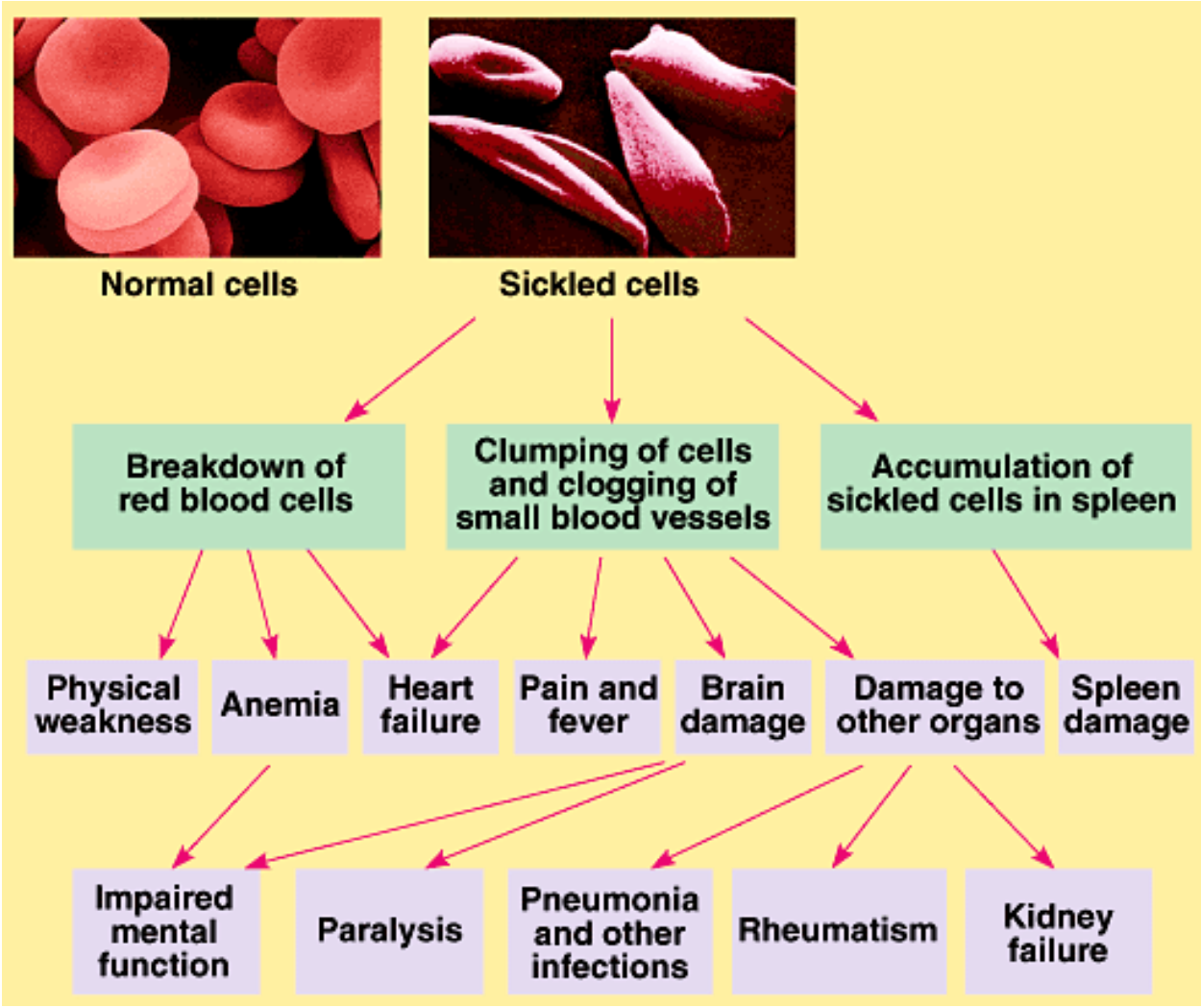
Multiple Alleles

- Genes that exist in populations in more than two allelic forms.
- ABO blood groups in humans
  - Blood types can be A, B, AB, or O
  - The letters refer to two carbohydrates that may be found on the surface of red blood cells.
    - I = represents the enzyme that attaches the carbohydrate to the cell
  - Blood cells may have one substance or the other (type A or B), both (type AB), or none (type O).

Pleiotropy

- The ability of a gene to affect an organism in many ways

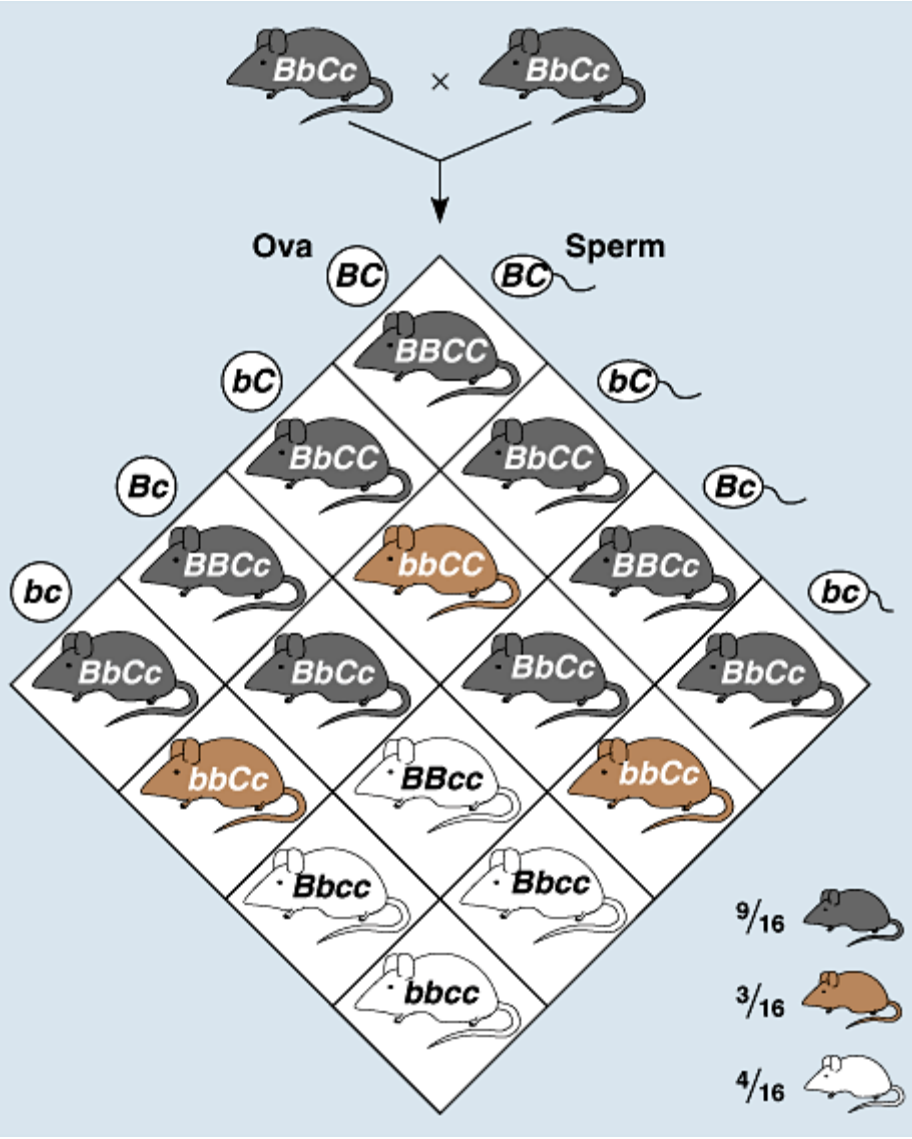
- Example: sickle cell anemia can cause spleen damage, heart failure, weakness, etc.



Epistasis

- A gene at one locus alters the phenotypic expression of a gene at a second locus.
- In mice – *B* = black fur, *b* = brown fur

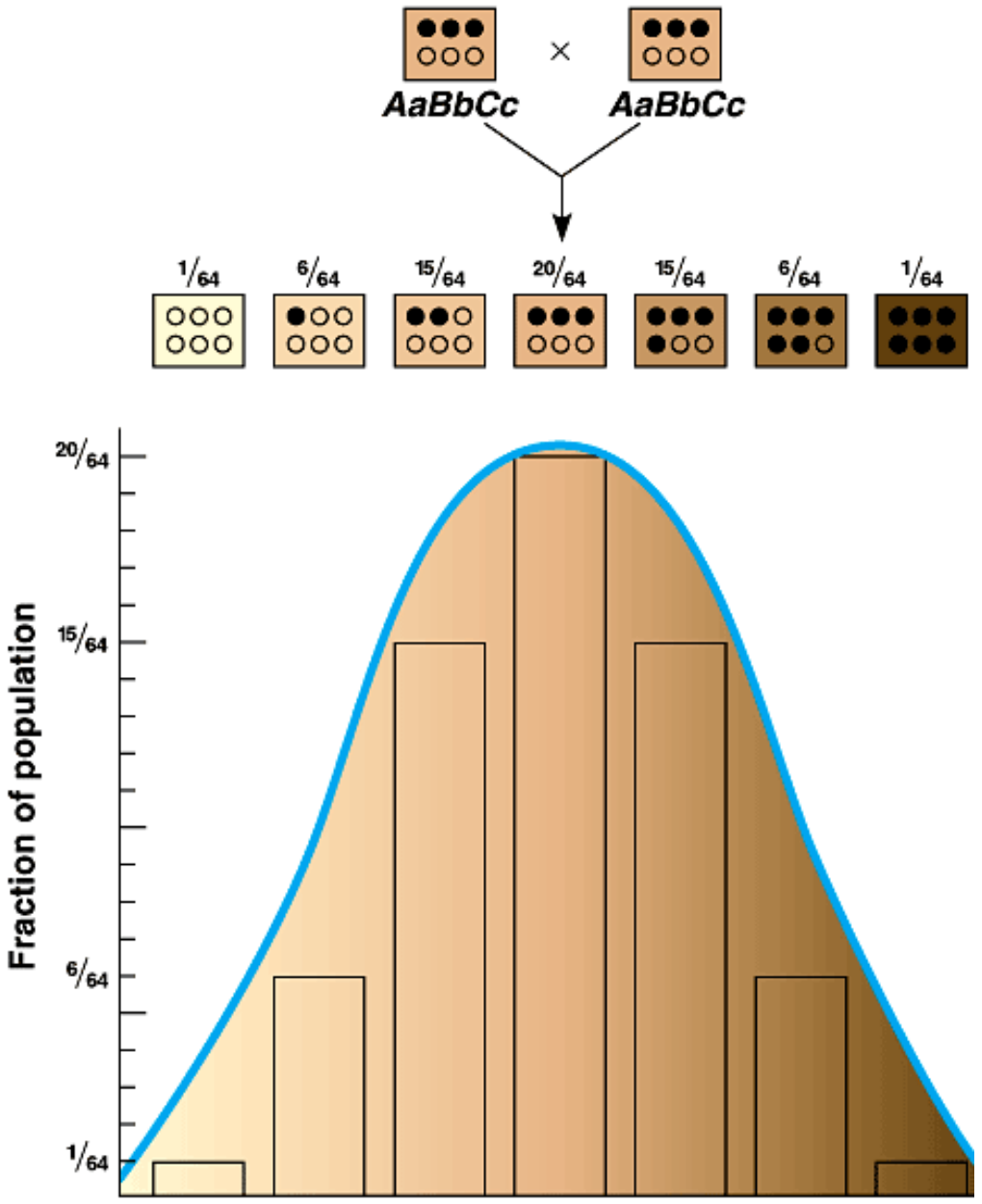
  C is a dominant allele that determines if pigment is deposited in the hair. If a mouse inherits *cc*, it will be albino.



Polygenic Inheritance

- An additive effect of two or more genes on a single phenotypic character.
- Varies in a population along a continuum
- Example – human skin color and height
  - Human skin color has at least 3 separately inherited genes

Polygenic Inheritance



Nature and Nurture: The Environmental Impact on Phenotype

- Indicates a range of phenotypic possibilities determined by a specific genotype that can be altered by environmental factors.
- This range is called *norm of reaction multifactorial* – many factors, both genetic and environmental, collectively influence phenotype.

Disorders

- Recessive inheritance – cystic fibrosis, sickle cell, consanguineous matings (incest)
- Dominant inheritance – achondroplasia (form of dwarfism), Huntington's disease
  - People with a dominant lethal disorder more than likely will not survive to reproduce, but people with a recessive lethal disorder can be a carrier and pass on the allele to offspring
- Multifactorial inheritance – has a genetic and environmental component
  - Ex) heart disease, diabetes, cancer, alcoholism, bipolar