6.2 Mendelian Genetics: When the Role of Genes Is Clear

Gregor Mendel

- Determined how traits were inherited
- The “father of genetics”
- Used pea plants and analyzed traits of parents and offspring

Essay 6-1 figure 1a Biology: Science for Life, 2/e

Mendelian Genetics

- Mendelian genetics
  - the pattern of inheritance described by Mendel
  - for single genes with distinct alleles

- Sometimes inheritance is not so straightforward
  - In humans it is rarely this straightforward…..

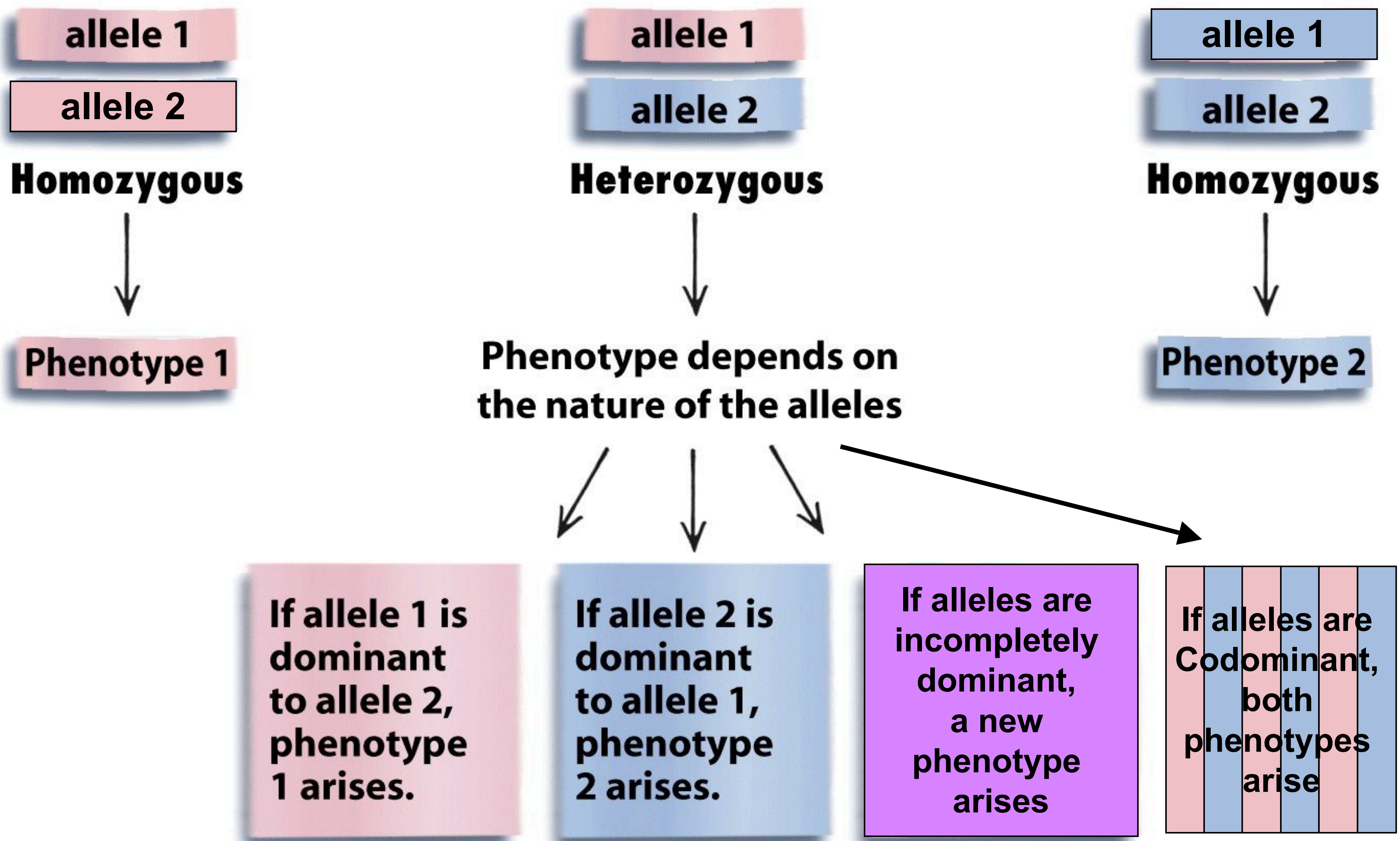
Medelian Definitions

- **Genotype** – combination of alleles
  - **homozygous**: two of the same allele
  - **heterozygous**: two different alleles
- **Phenotype**
  - the physical outcome of the genotype
  - depends on nature of alleles
    - Dominance or recessiveness, etc

Mendelian Definitions

- **Dominant** alleles
  - Can mask a recessive allele
  - Always shows its phenotype
  - Capital letters are used
    - For example: T for tall
- **Recessive** alleles
  - Can be masked by a dominant allele
  - Only show phenotype if both alleles are recessive
  - lower case letters are used
    - For example: t for short
- **Incompletely dominant** alleles
  - Alleles produce an intermediate phenotype
  - Phenotype does not resemble either allele
    - White allele and Purple allele = pink phenotype
- **Codominant** alleles
  - Both alleles are fully expressed
  - Phenotype resembles both alleles
    - Spotted allele and striped allele = spotted and striped phenotype

Mendelian Definitions



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Genetic Diseases in Humans

- Most alleles do not cause diseases in humans
- There are some diseases that are genetic and result from a single gene:
  - Recessive, such as cystic fibrosis
    - Need 2 alleles to have disease
  - Dominant, such as Huntington's Disease
    - Need only one allele to have disease
  - Codominant, such as sickle-cell anemia
    - One allele causes mild disease
    - Two alleles causes severe disease

Genetic Diseases: Cystic Fibrosis

- Affects 1 in 2500 individuals of European-decent
- Recessive condition:
  - individuals with disease have 2 copies of cystic fibrosis allele
- Diseased individuals produce mutant nonfunctioning proteins
  - Normal protein transports chloride ion in and out of cells in lungs and other organs
  - Mutant protein does not
    - Results in a thick mucus layer that is difficult to clear out of lungs and interferes with absorption of nutrients in intestines
    - Can result in suffocation and/or malnutrition and starvation
- Carriers
  - have one cystic fibrosis allele and one normal
  - but do not have cystic fibrosis
  - Appear phenotypically normal
- Carriers can pass the disease allele to children

Huntington Disease

- Dominant condition
- Fatal condition
- Only one Huntington’s allele needed
- Produces abnormal protein that clumps up in cell nuclei
  - Especially nerve cells in the brain
  - Results in progressive brain malfunction and death

Sickle-Cell Anemia

- Codominant
  - both alleles are expressed
- One allele codes for normal hemoglobin and the other codes for sickle-cell hemoglobin

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Sickle-Cell Anemia

- If you have two normal hemoglobin alleles
  - you do not have the disease
- If you have two sickle-cell hemoglobin alleles
  - you have sickle-cell disease
  - You cannot carry oxygen efficiently
  - Can suffocate under low oxygen conditions
  - Red blood cells form a sickle shape and clog arteries
  - Can cause blood clots and death
- If you have one of each, you have sickle cell trait
  - Some of your red blood cells will sickle under low oxygen conditions
  - Some of your red blood cells will clog up arteries
  - Can cause blood clots and death

Why do we care????

- You could be a carrier of a lethal disease and not know it
  - You could pass this allele to your children

- You can “see your future” if you know your genotype…

- Genetics is Nerdy Fun!

How do we know our genotypes?

- Understanding Dominance and Recessive relationships of certain alleles
- Usually need to know phenotypes of parents
- We can use Punnett squares to predict offspring phenotypes
- Punnet Squares use possible gametes from parents to predict possible offspring

Punnett Squares: Single Gene Traits

- A parent who is heterozygous for a trait
  - Pp can produce two possible gametes
  - P   or p
- A parent who is homozygous for a trait
  - PP can only produce one kind of gamete
  - P
- The possible gametes are listed along the top and side of the square
- The predicted offspring genotypes are filled in the center boxes of the square
- Punnet squares can be used to predict possibilities of inheriting genetic diseases

Punnett Square of Complete Dominance

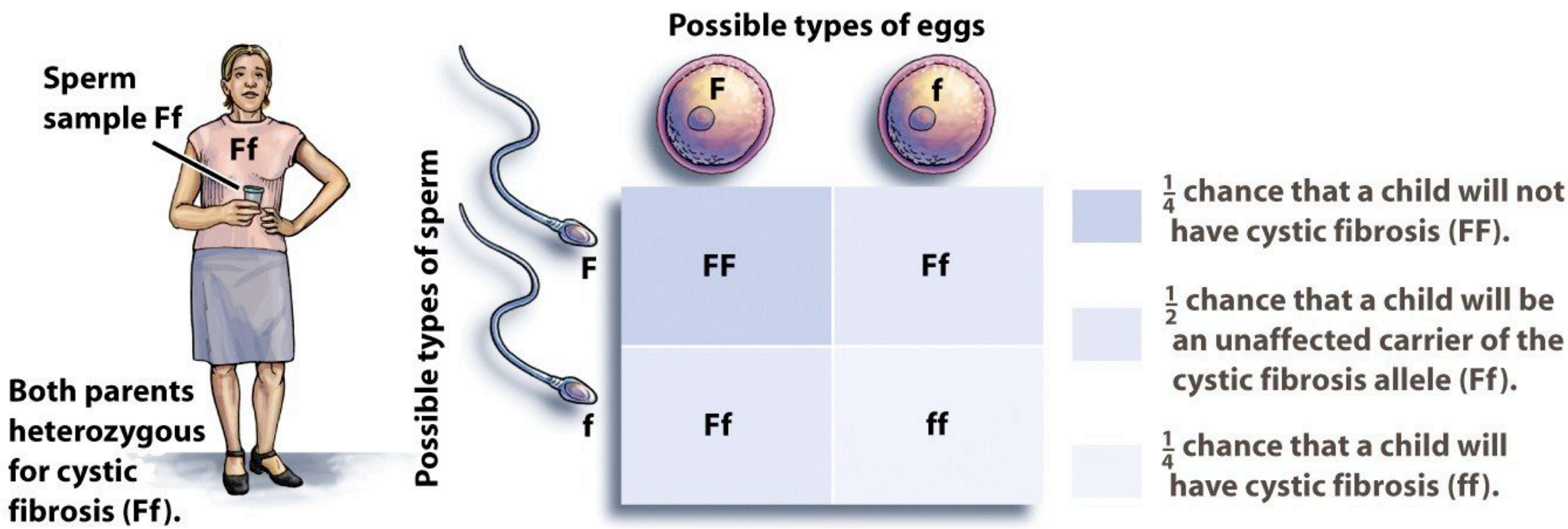
PP= purple
Pp= purple
pp= white

PURPLE (PP)

WHITE (pp)

	P	P
p	Pp	Pp
p	Pp	Pp

How do we know our genotypes?
Use Punnet Squares!



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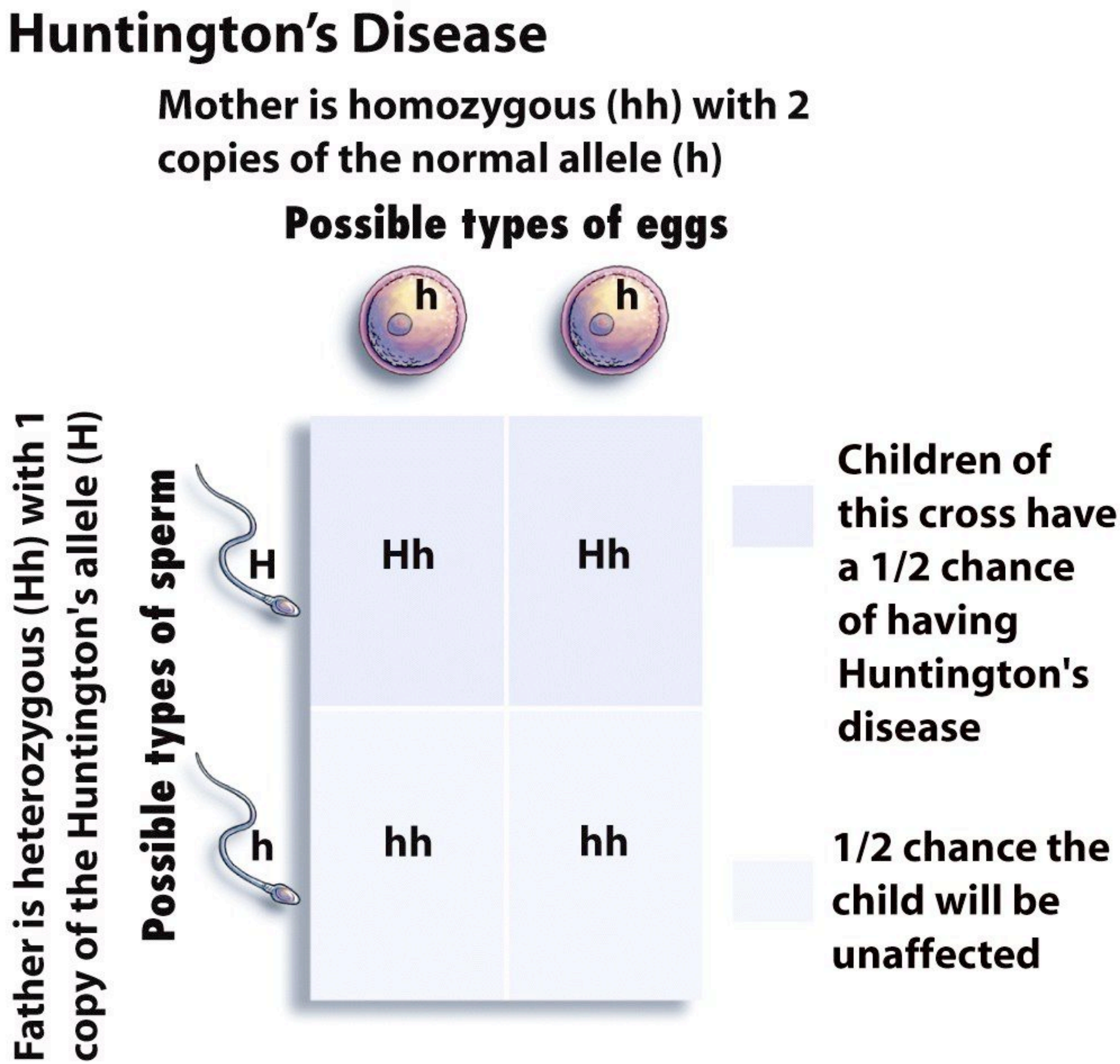
- These are probabilities for each individual offspring
- For every fertilization event
  - there is a 25% chance of having a cystic fibrosis diseased offspring



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Sickle-cell anemia

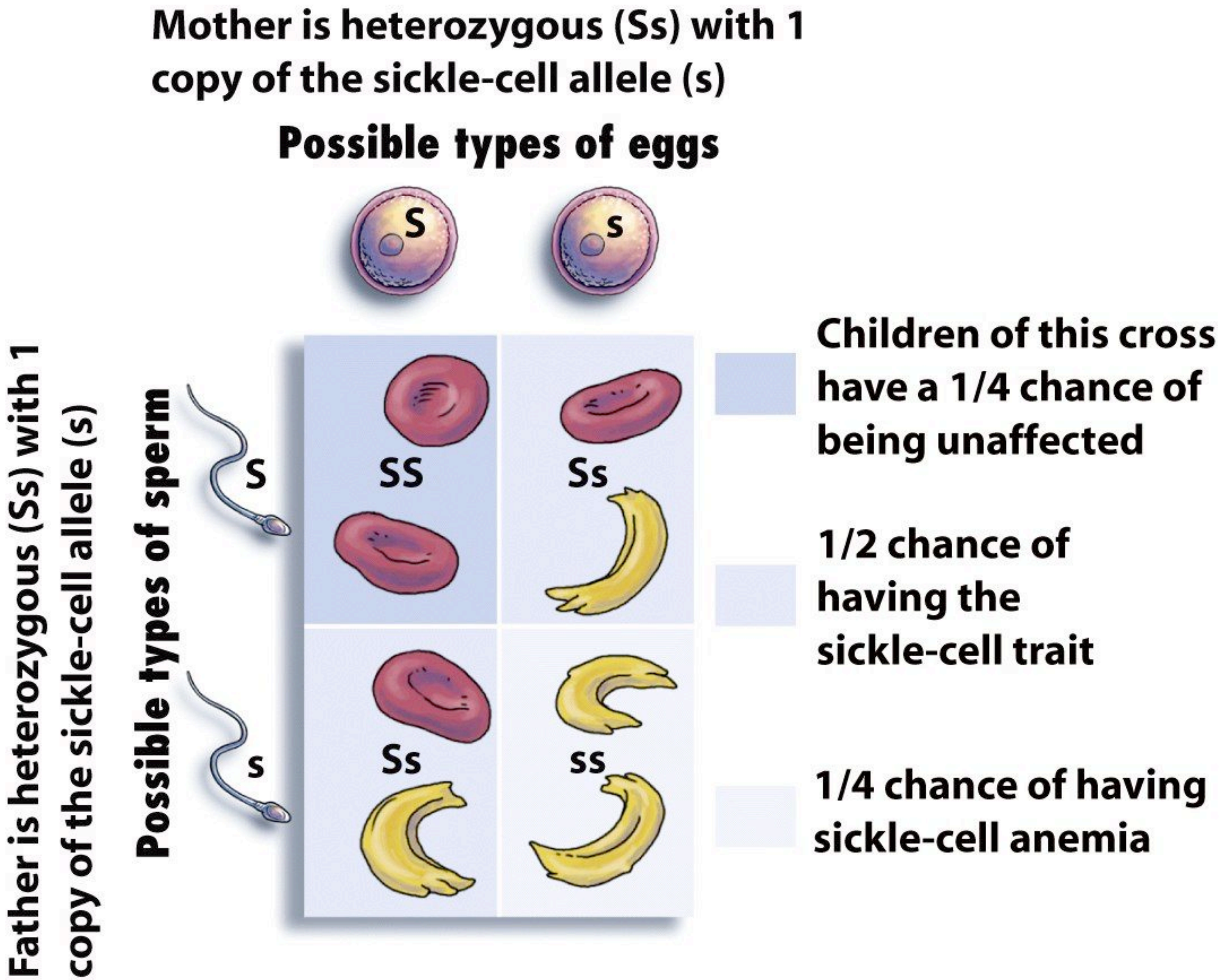
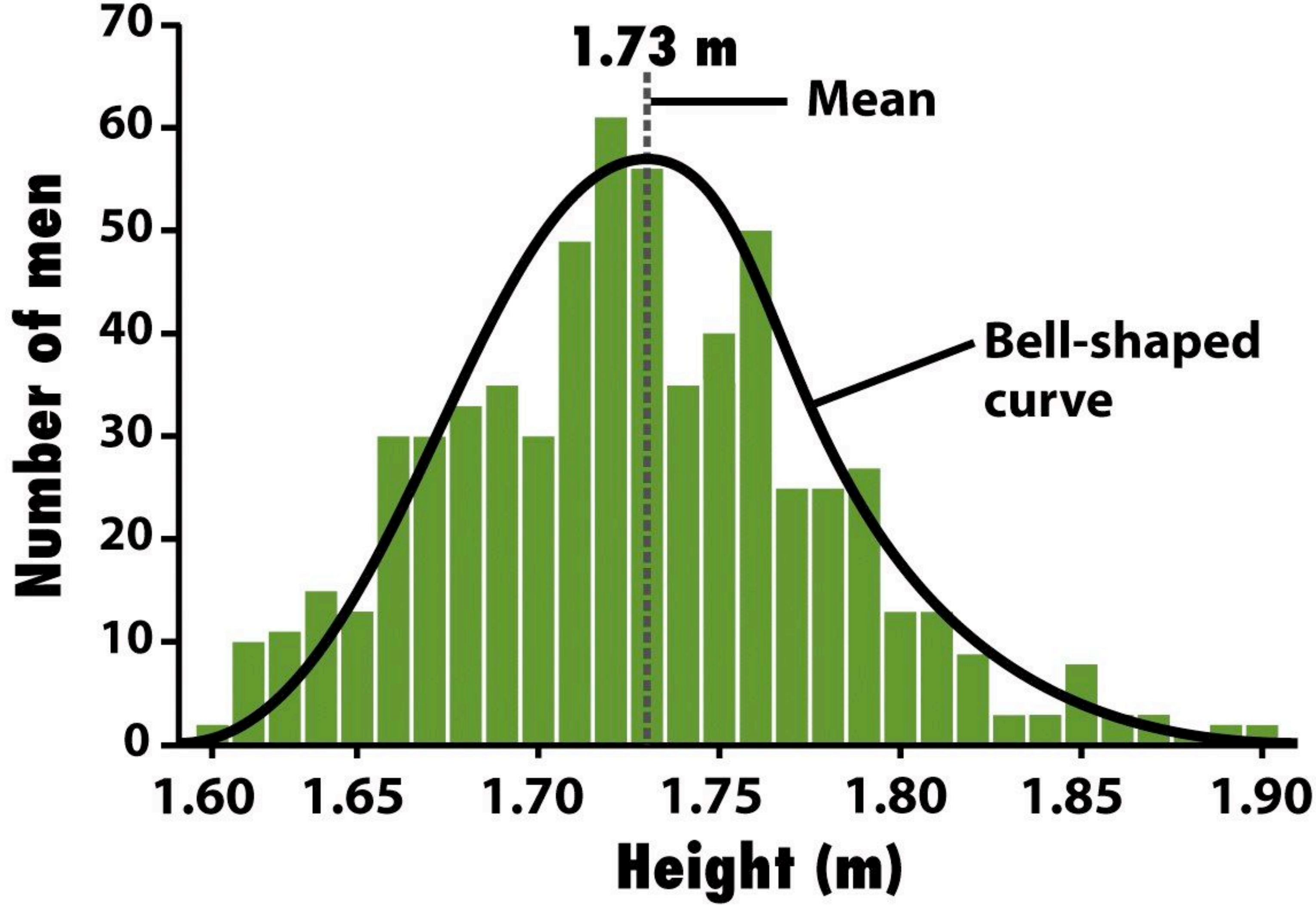


Figure 6-11b Biology: Science for Life, 2/e

6.3 Quantitative Genetics

- Quantitative traits include weight, musical ability, susceptibility to cancer, intelligence, and height…
- The environment plays a role
  - Many genes involved in a person’s maximum height
  - Diet and nutrition determine whether that person reaches the maximum genetically possible
- **Quantitative traits** show **continuous variation**
  - we can see a large range of phenotypes in the population
- **Variance** is the amount of variation in a population

Normal distribution of student height in one college class.



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Variance describes the variability around the mean.

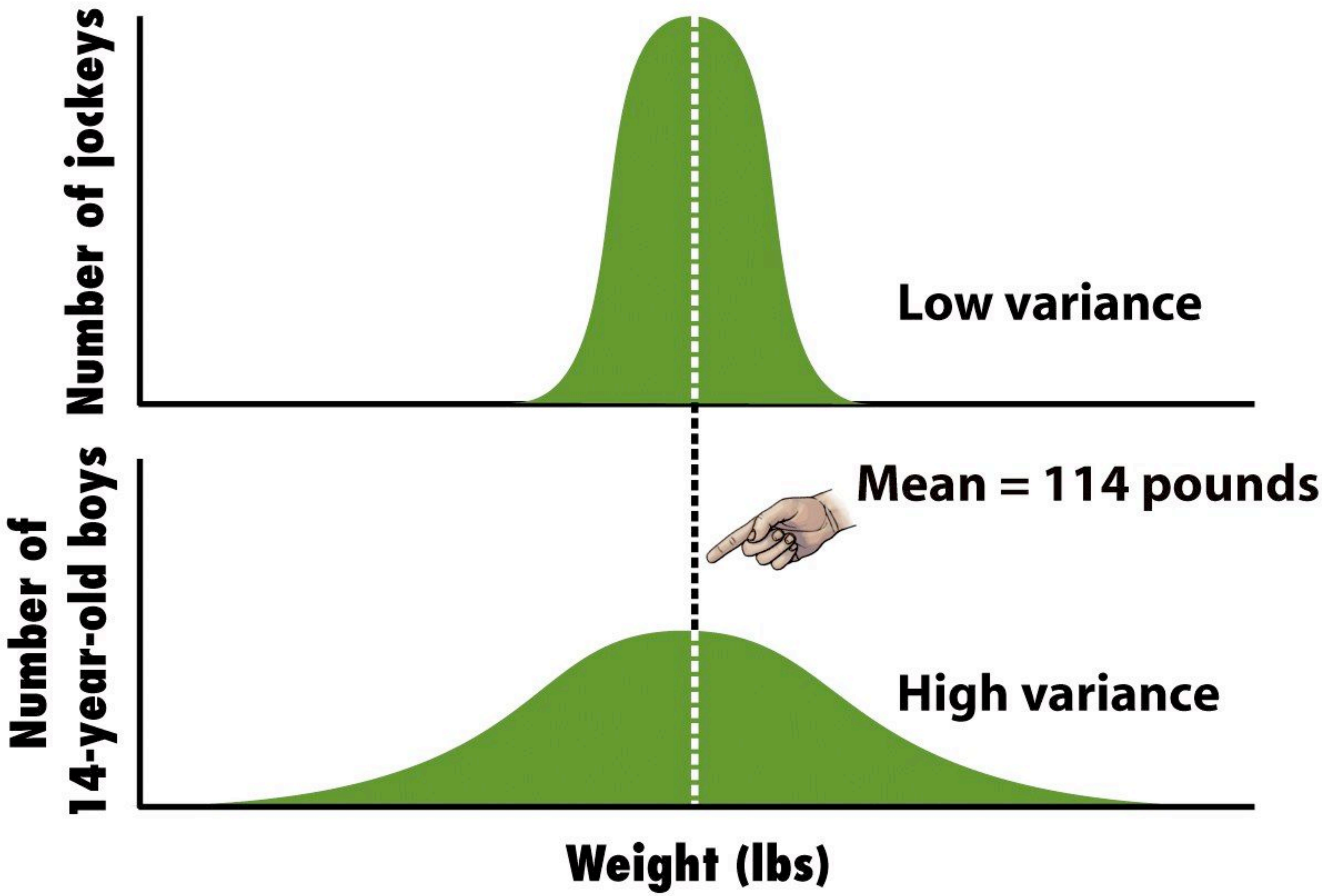

Figure 6-13b Biology: Science for Life, 2/e

Why Traits Are Quantitative

- **Polygenic traits**
  - those traits influence by more than one gene

- Eye color is a polygenic trait
  - There are two genes:
    - pigment
    - Distribution

- This produces a range of eye colors

Why Traits Are Quantitative

- Environment can affect phenotypes
- Identical twins have the same genotypes but may not have exactly the same appearance…

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Why Traits Are Quantitative

- Skin color is affected by both genes and environment…

Genes

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Environment

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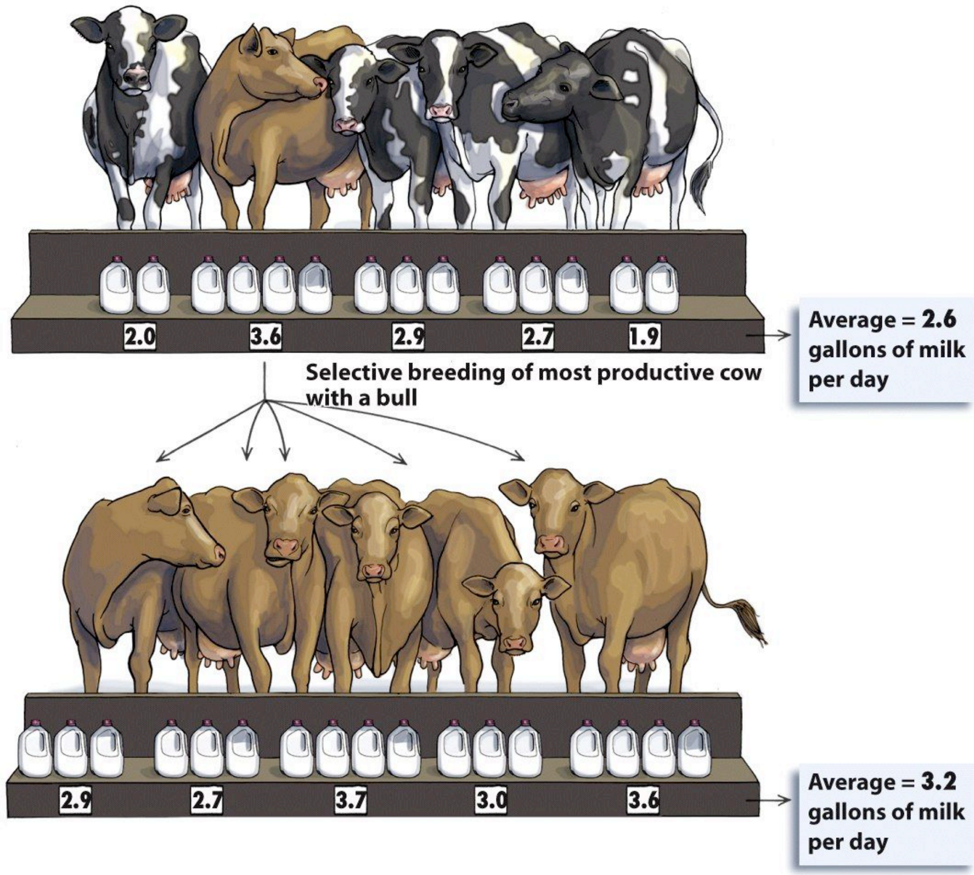
Using Heritability to Analyze Inheritance

- Inheritance patterns for these quantitative traits are difficult to understand

- Researchers use plants and domestic animals to study **heritability**
  - a measure of the relative importance of genes in determining variation in quantitative traits among individuals

Using Heritability to Analyze Inheritance

- **Artificial selection**:
  - controlling the reproduction of organisms to achieve desired offspring



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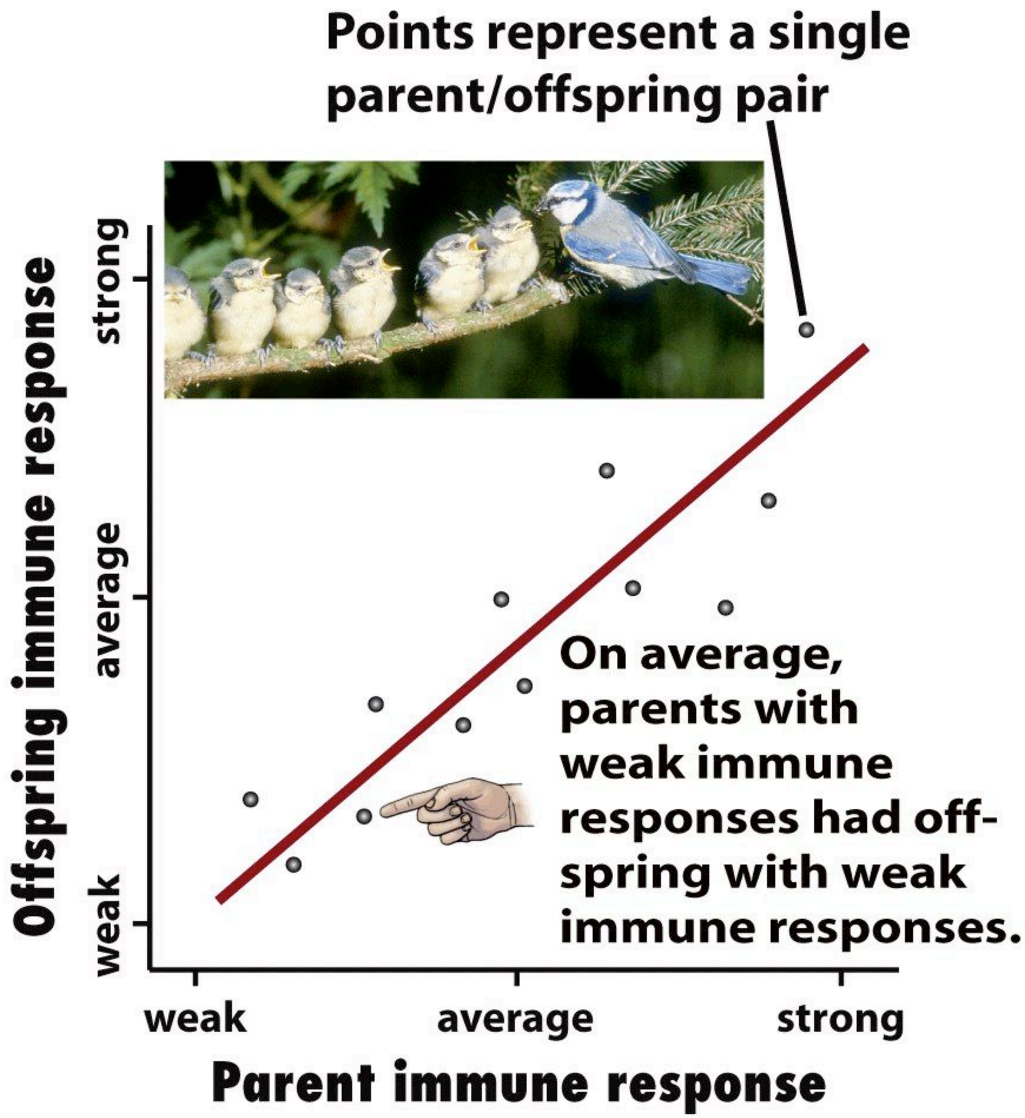
Correlations between Parents and Children

- Using animals we can perform experiments to correlate parental traits with traits in offspring
- Vaccinate parent and offspring in a bird population with Tetnus vaccine
- Look for correlation between parents and offspring in ability to produce anti-tetanus proteins
  - ability to produce an immune response to the vaccine

Correlations between Parents and Children



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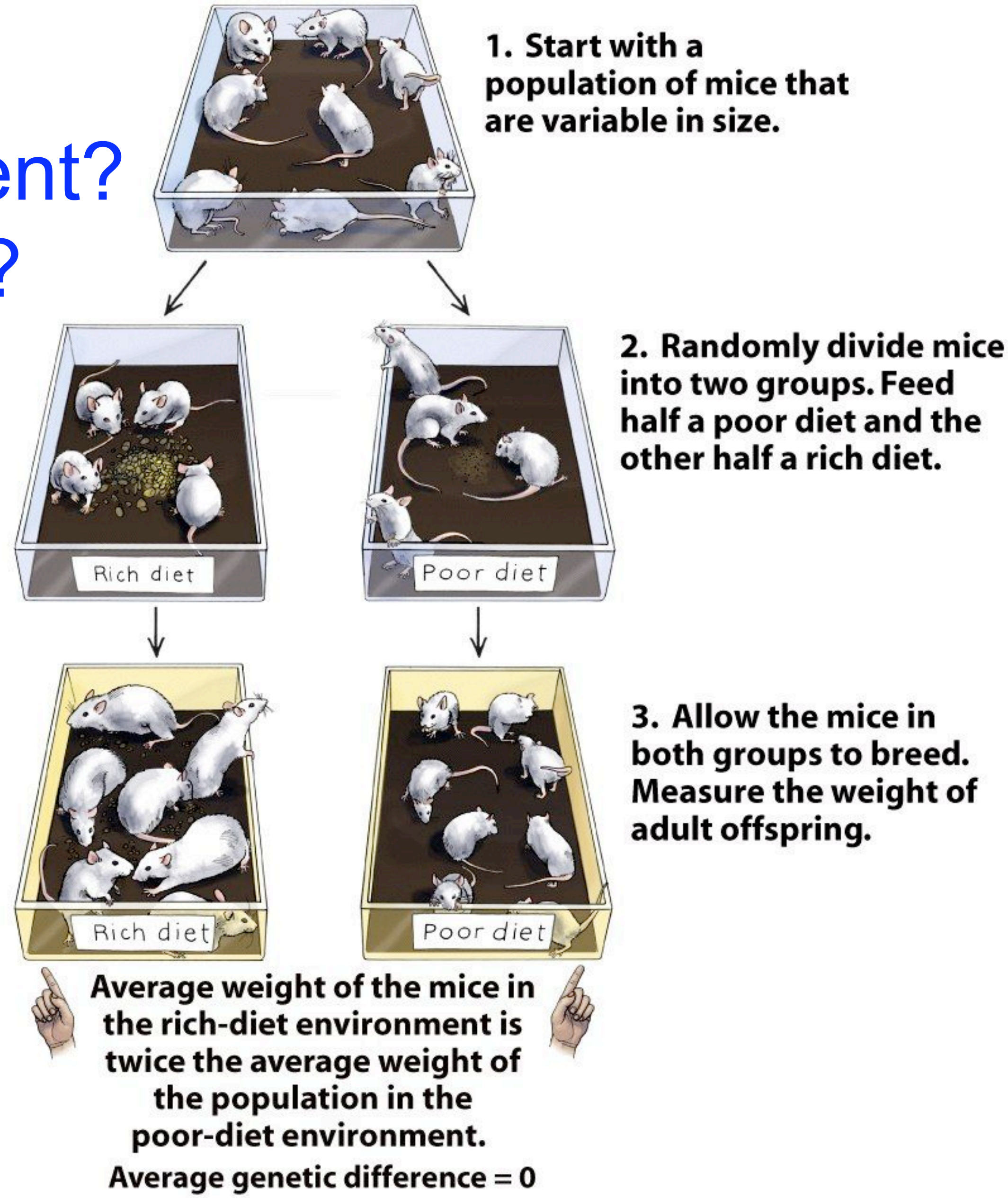
Genes or Environment? Nature or Nuture?

- Highly heritable traits can respond to environmental change

- Traits can be both highly heritable <u>and</u> strongly influenced by the environment
  - examples
    - mouse weight
    - rat intelligence

Genes or Environment?
Nature or Nuture?

Weight of mice is highly dependent on environment



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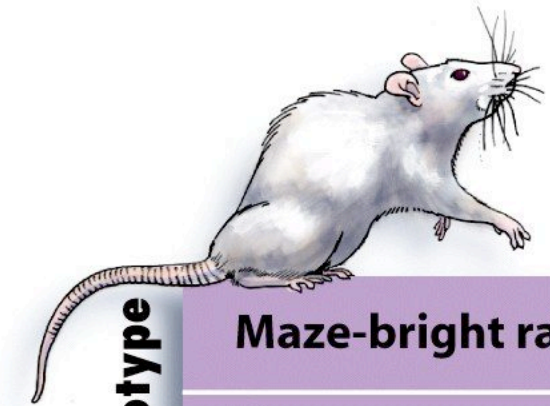
Genes or Environment?
Nature or Nuture?



Phenotype	Environment in which rats were raised		
	Normal environment	Restricted environment	Enriched environment
Maze-bright rats	115	170	112
Maze-dull rats	165	170	122

- **Normal environment:** Maze-dull rats made more mistakes than maze-bright rats when running a maze.
- **Restricted environment:** Both groups made the same number of mistakes when running a maze.
- **Enriched environment:** Both groups made fewer mistakes when running the maze. The maze-dull rats improved the most compared to the normal environment.

Table 6-2 Biology: Science for Life, 2/e