Objective 11: Analyze a simple pedigree

- **Pedigree**—a family history that shows how a trait is inherited over several generations.
- **Carriers**—individuals who are heterozygous for an inherited disorder but do not show symptoms.

**Albinism pedigree**

Horizontal lines indicate matings.
Vertical lines indicate offspring, arranged from left to right in order of their birth.

The colored symbols represent affected individuals:
- □ Male
- ○ Female
- ● Male albino
- ◼ Female albino

**Autosomal or sex-linked?**

- **Autosome**—a chromosome other than an X or Y chromosome.
  - If a trait is autosomal, it will appear in both sexes equally.
- **Sex-linked trait**—a trait whose allele is located on the X chromosome.
  - Most sex-linked traits are recessive.
  - A male who carries a recessive allele on the X will exhibit the condition.

**Analyzing pedigrees**

- **Dominant or recessive?**
  - If the trait is autosomal dominant, every individual with the trait will have a parent with the trait.
  - If the trait is recessive, an individual can have one, two, or neither parent exhibit the trait.

**Analyzing pedigrees**

- **Heterozygous or homozygous?**
  - Individuals with autosomal traits that are heterozygous or homozygous dominant, their phenotype will show the dominant characteristic.
  - Homozygous recessive individuals will show the recessive characteristic.
  - Two individuals who are heterozygous carriers of a recessive mutation will not show the trait, but they can produce children who might.
You Should Now Be Able To:

• When analyzing a pedigree, explain how you can determine if an individual is a carrier for the trait being studied.

Traits influenced by the environment

• An individual’s phenotype often depends on environmental conditions.
  – *hydrangea* flowers range in color from blue to pink, depending on the acidity of the soil
  – fur color of the arctic fox is affected by temperature
    • reddish brown in the summer, white in the winter

More examples of traits influenced by the environment

– Fur color in Siamese cats is influenced by temperature
  • dark fur occurs at locations on its body that are cooler than the normal body temperature
– height in humans is influenced by nutrition
– exposure to the sun alters the color of the skin
– males & females inherit a “baldness” allele, but it is activated by testosterone

Objective 13: Describe how mutations can cause genetic disorders

• *Mutations*—changes in genetic material
  – many are carried by recessive alleles in heterozygous individuals
  – mutations are rare because cells have efficient systems for correcting errors
• *Genetic disorders*—harmful effects produced by inherited mutations

Objective 14: List two genetic disorders, and describe their causes and symptoms

• **Sickle cell anemia**—a defective form of hemoglobin causes many red blood cells (RBCs) to bend into a sickle shape
  – cells rupture easily, resulting in less oxygen being carried by the blood
  – can get stuck in blood vessels, cutting off blood supply to an organ
  – affects 1 out of every 500 African Americans

Sickle cell anemia

• Caused by a recessive allele
• The allele protects heterozygous individuals from the effects of malaria.
  – The sickled RBCs of heterozygous individuals kill the parasite that causes malaria
  – heterozygous individuals still produce enough normal RBCs
Hemophilia & Huntington’s Disease

- **Hemophilia**—a condition that impairs the blood’s ability to clot.
  - Caused by a sex-linked recessive allele

- **Huntington’s Disease**—genetic condition characterized by loss of muscle control, uncontrollable spasms, severe mental illness, and eventually death.
  - Caused by an autosomal dominant allele

Objective 15: Evaluate the benefits of genetic counseling

- **Genetic counseling**—a form of medical guidance that informs people about genetic problems that could affect them or their offspring.
- Therapy may be available to treat a disorder if diagnosed early
  - phenylketonuria (PKU)
- Gene Therapy?

You should now be able to:

- Differentiate between incomplete dominance and codominance
- Identify two examples of traits that are influenced by environmental conditions
- Summarize how a genetic disorder can result from a mutation
- Describe how males inherit hemophilia.
- Explain why a couple might undergo genetic counseling?

- **Cystic Fibrosis**
  - Most common genetic disorder among whites (affects 1-2500 whites).
  - 1 out 22 whites are carriers

- **Phenylketonuria (PKU)**
  - A recessive genetic disorder
  - Body cannot convert the amino acid phenylalanine into tyrosine (lacking the enzyme phenylalanine hydroxylase)
  - Phenylalanine builds up in blood and body tissue.

- **Duchenne Muscular Dystrophy**
  - X-linked genetic disorder (affects 1-3500 boys worldwide)
  - Lack of dystrophin (a large protein required for structural support of cells) causes muscle cells to “explode.”