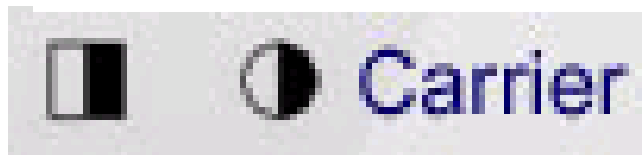
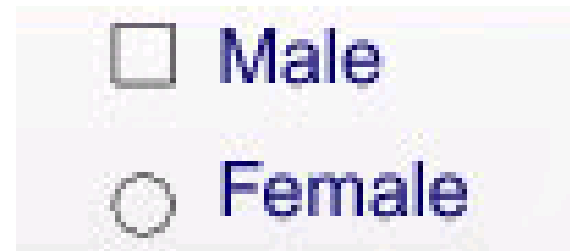


Human Genetics

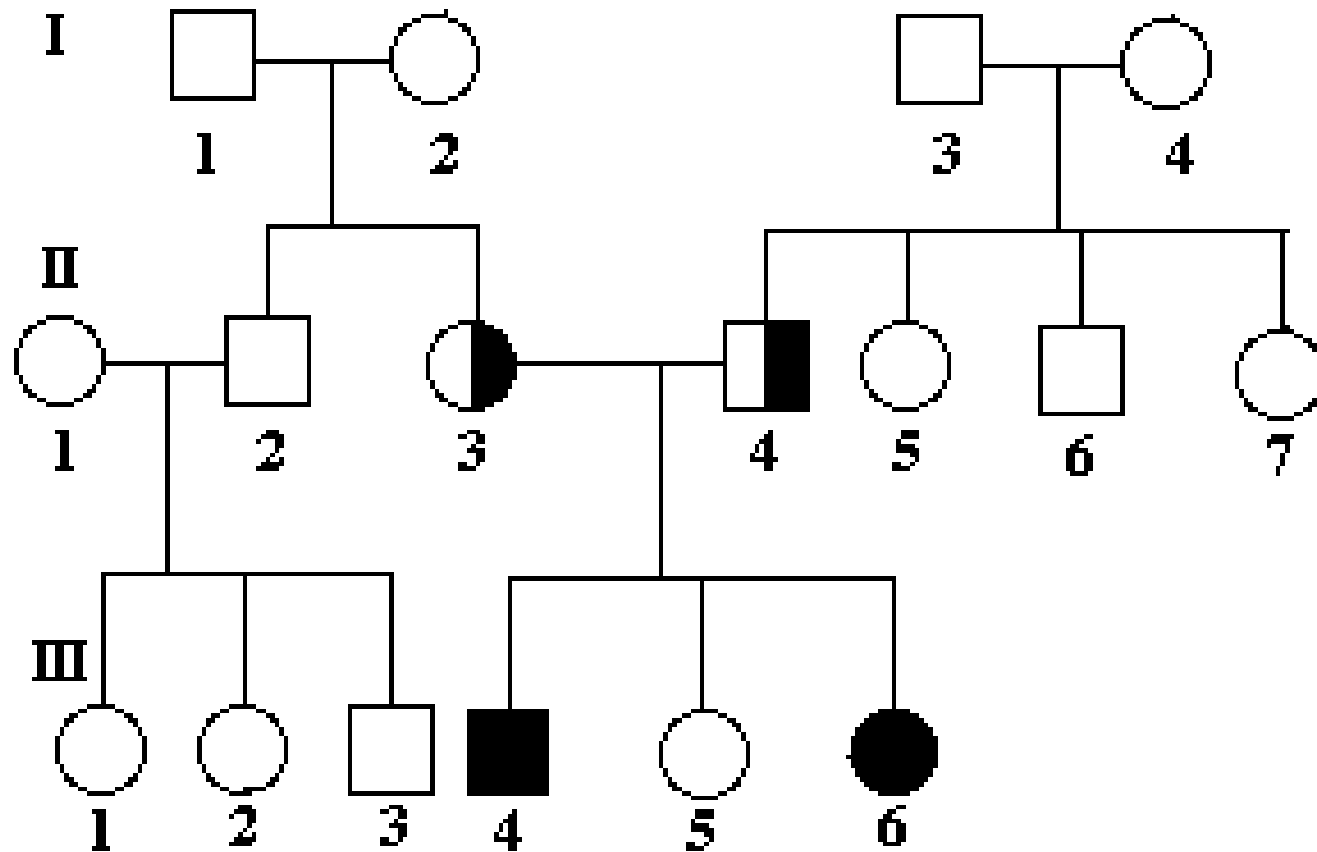


Pedigree

- Graphic representation of genetic inheritance
- Family tree

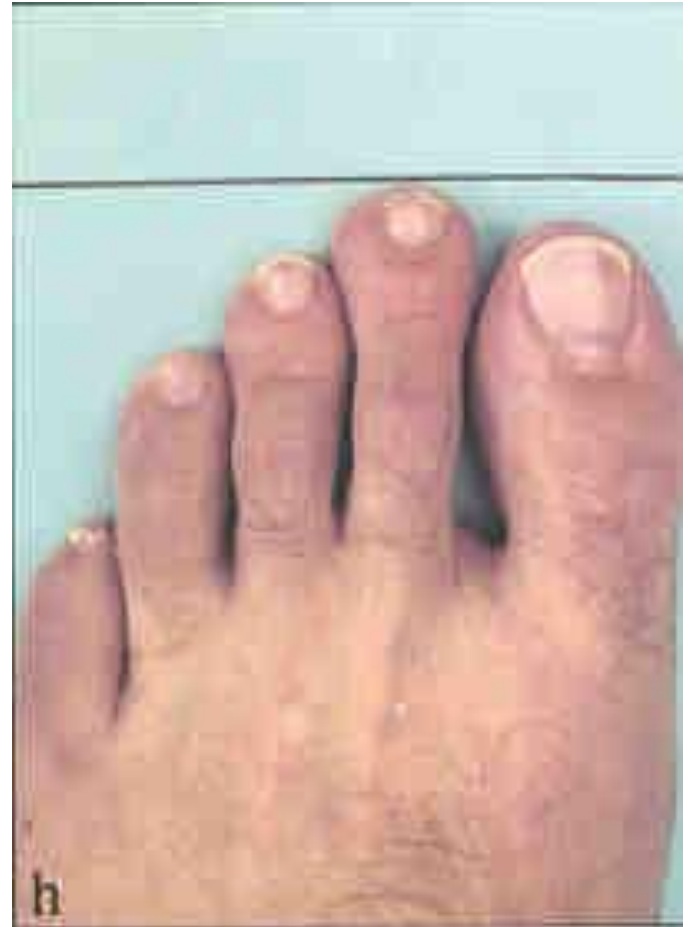


Pedigree



Recessive Traits

- Attached earlobe
- Hitchhikers thumb
- 2nd toe larger than big toe



Recessive Disorders

- Cystic Fibrosis
 - Abnormal amounts of mucus in lungs
 - Abnormal cilia in lungs
 - Death by age 20
 - 1 in 20 white Americans are carriers



Recessive Disorders

- Albinism
 - Lack of pigmentation



Recessive Disorders

- Tay Sachs

- Absence of enzyme that breaks down lipids
- Lipids build up in the brain causing brain damage
- Death by age 5
- Amish and Ashkenazic Jews

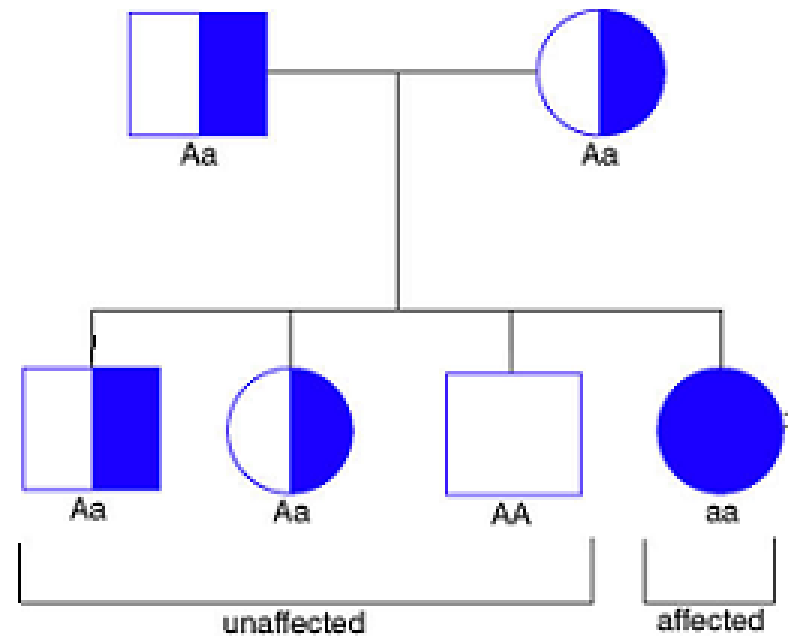


Recessive Disorders

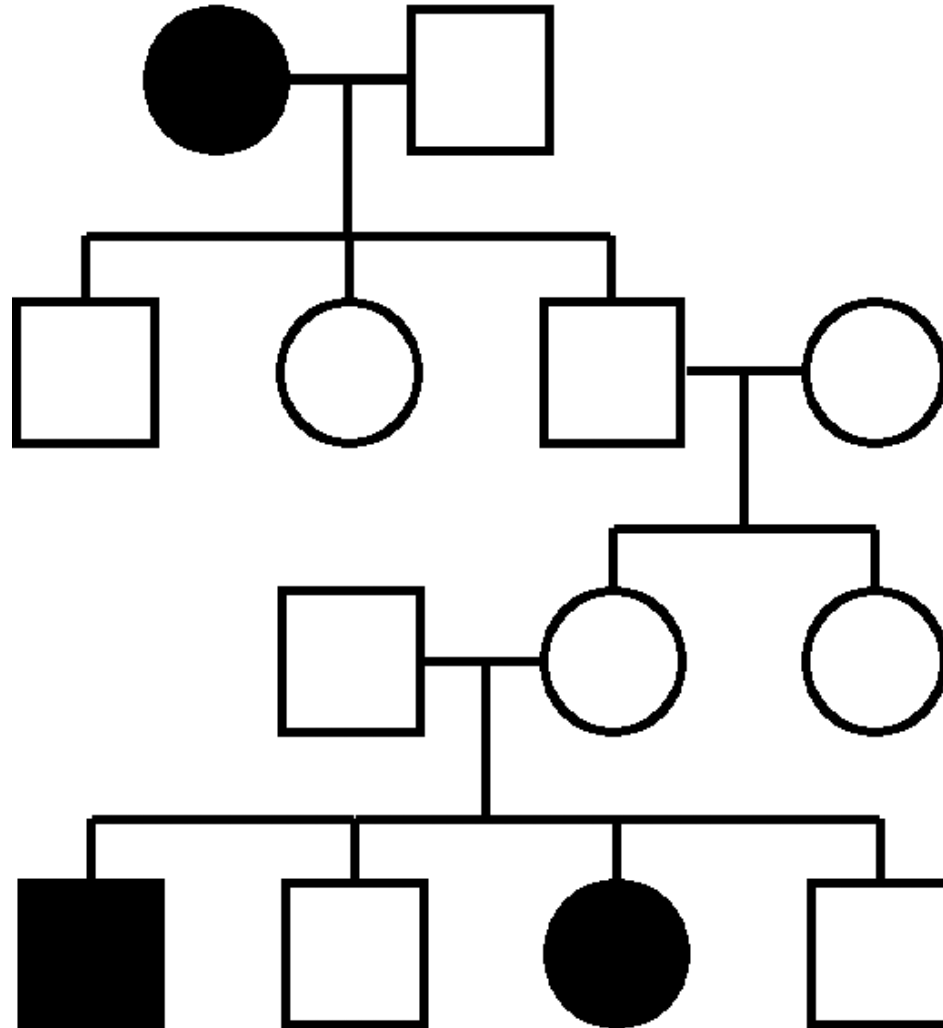
- Phenylketonuria-PKU
 - Absence of enzyme that breaks down phenylalanine
 - Phenylalanine builds up causing brain damage
 - Treated with diet



Recessive Pedigree

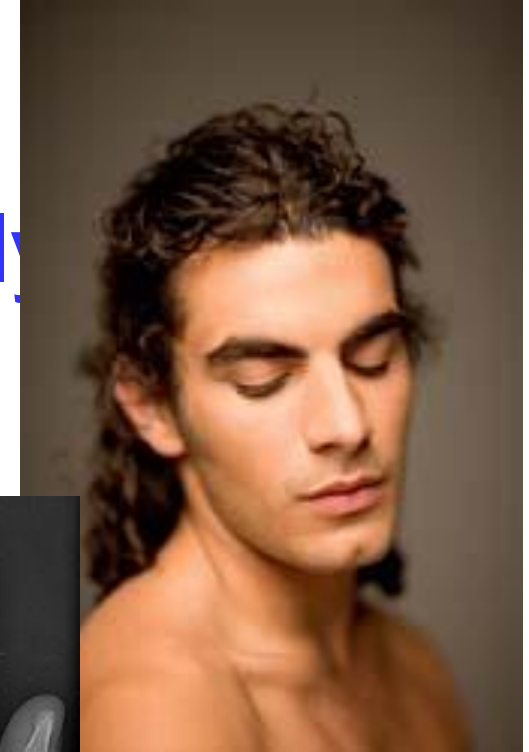


Recessive Pedigree



Dominant Traits

- Widows peak, free earlobes, dimples, cleft chin, polydactyly

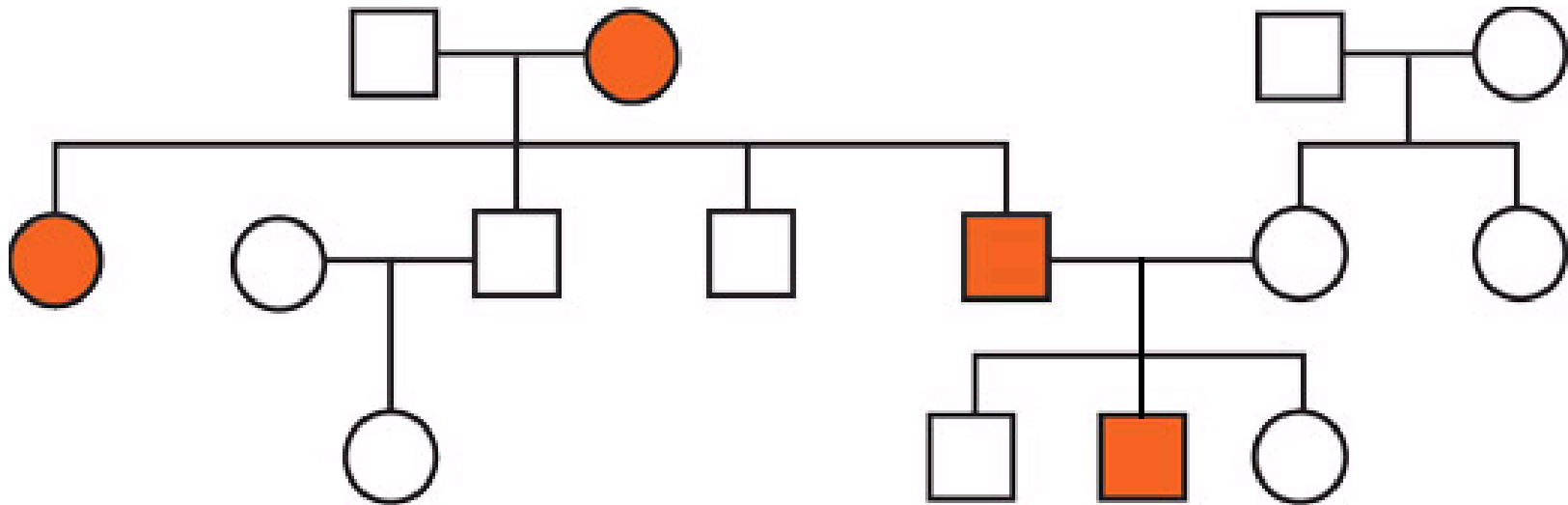


Dominant Disorders

- Huntington's Disease
 - Fatal degeneration of brain
 - Onset of symptoms age 30-50
 - After having children



Dominant Pedigree



□ Male

○ Female

■ Male with HD

● Female with HD

Dominant Disorders

- Achondroplasia
 - Most common type of dwarfism
 - 85% new mutations from normal parents
 - Genotypes
 - dd: Normal
 - Dd: Dwarf
 - DD does not survive

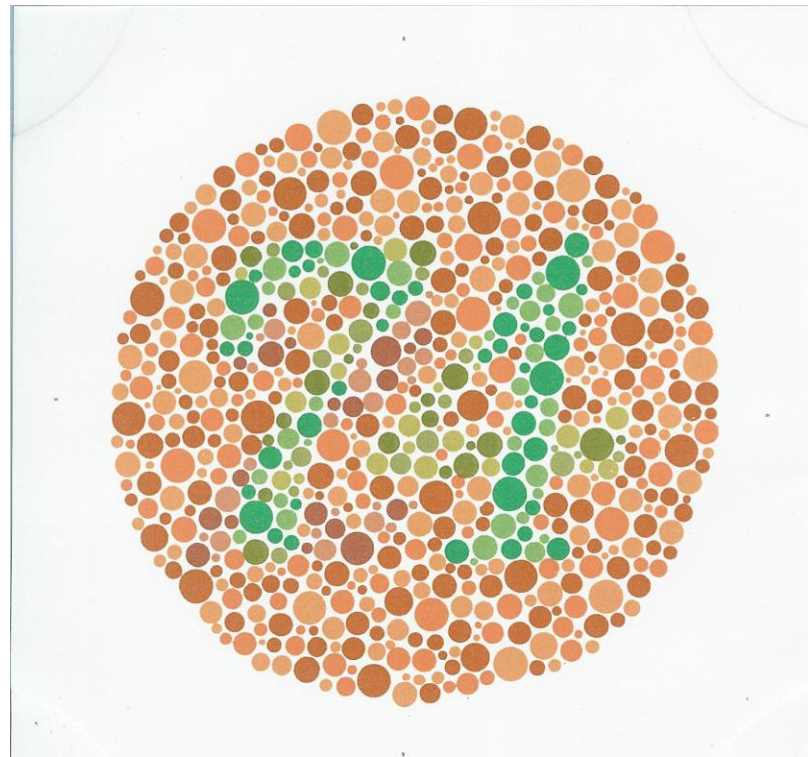
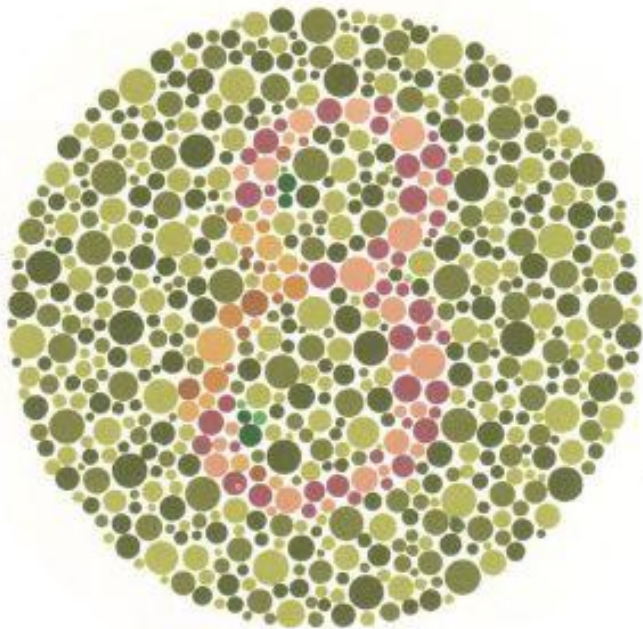


Sex- Linked Disorders

- Defective genes on X chromosome
- Carrier mother passes defective gene to son
- Affected males pass defective gene to daughter (carrier)
- Affected females are rare

Sex- Linked Disorders

- Red-green colorblindness
 - Defective red and green light receptors in eye



Colorblindness Genotypes

- $X^R X^R$: Normal Female
- $X^R X^r$: Carrier Female
- $X^r X^r$: Colorblind Female
- $X^R Y$: Normal Male
- $X^r Y$: Colorblind Male
- No way to have a carrier male

Sex- Linked Disorders

- Hemophilia
 - Absence of blood clotting enzyme



Codominant Disorders

- Sickle-cell anemia
 - 1 in 12 African Americans are carriers
 - Genotypes
 - HH Normal Hemoglobin
 - HS both normal and defective hemoglobin
 - Sickle-cell Trait
 - Few or no symptoms



Codominant Disorders

- SS all hemoglobin is defective
 - Red blood cells become sickle shaped
 - Carry less oxygen



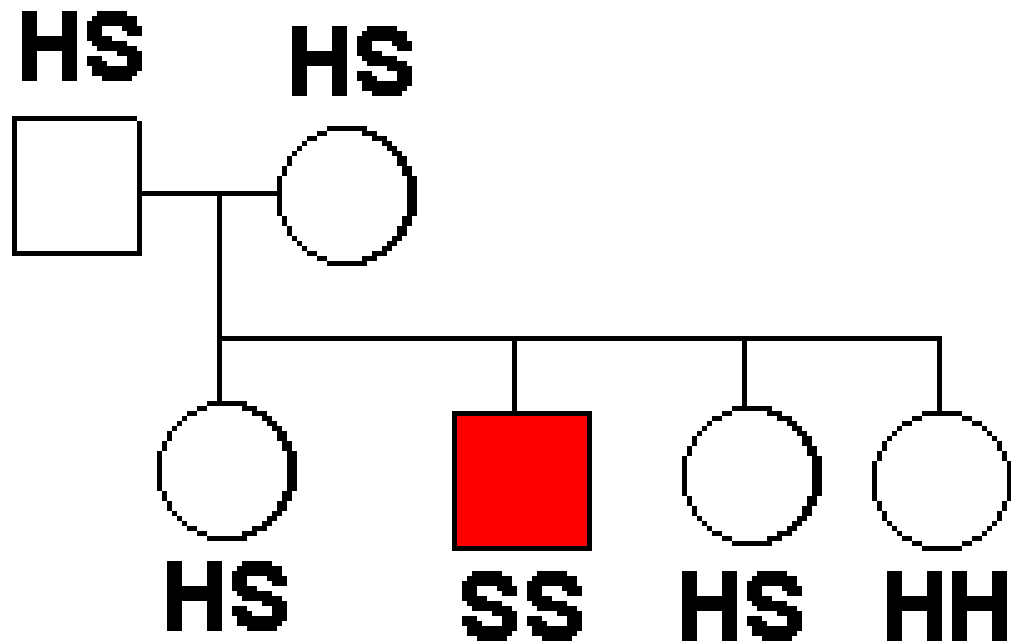
Codominant Disorders

– Beneficial mutation

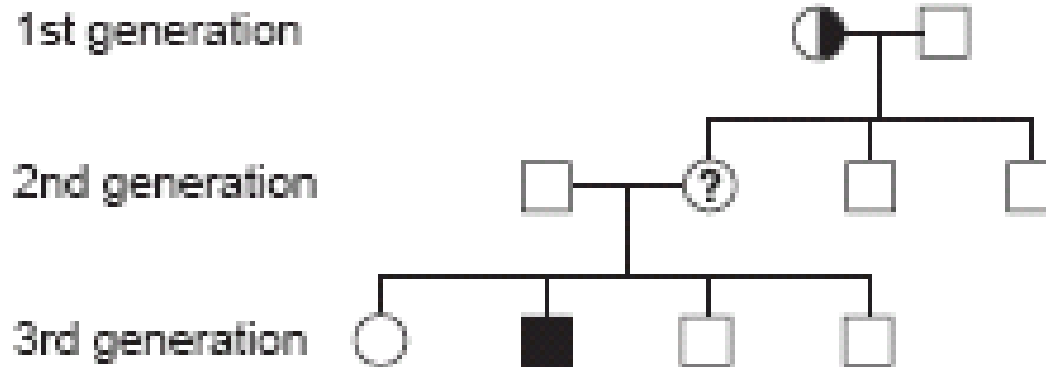
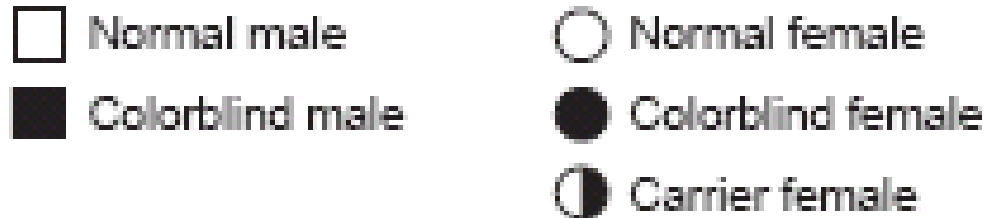
- HS and SS are immune to malaria
- HH is not immune



Sickle-Cell Pedigree



Pedigree Practice



**Pedigree Chart Tracing Sex-linked
Colorblindness in a Family**