Sex-linked Genetic Disorders & Autosomal Disorders

Packet #15
Introduction

- **Sex Linked Genetic Disorders**
  - Disorders caused by malfunctioning genes on the sex chromosome.

- **Autosomal Genetic Disorders**
  - Disorders caused by malfunctioning genes on an autosome.
Hemophilia

- Hemophilia is an x-linked recessive disorder
  - Males will show this trait if they have the recessive allele on the X chromosome
  - Females will show this trait if they have the recessive allele on both X chromosomes
- Hemophilia does not allow individuals to have the ability to clot their blood.
Baldness

- Baldness is an x-linked dominant disorder
  - \( X^B X^b \)
    - This female will not go bald due to lack of testosterone
  - \( X^B X^B \)
    - This individual will start to lose her hair in the future
Autosomal Disorders
Huntington Disease

- Produced by a single dominant allele
- No symptoms appear until 30’s and 40’s
- Symptoms
  - Uncontrollable body movements
  - Degeneration of the nervous system
- Usually fatal 10-20 years after onset of symptoms

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<th>Father</th>
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<th>Huntington's Disease</th>
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Affected child (Hh) = 2/4 = 50%
Carried child (hh) = 2/4 = 50%
There are no carriers
Sickle Cell Anemia I

- Sickle cell is an **autosomal recessive disorder**.

- The mutated allele, Hb\textsuperscript{s}, causes a change in polypeptides found in hemoglobin
  - Hemoglobin is the protein that carries oxygen in red blood cells

- Remember, sickle cell is an example of overdominance (heterozygote advantage)
Sickle Cell Anemia II

- \( \text{Hb}^A\text{Hb}^A \)
  - Homozygous dominant
  - Individuals with this genotype have normal red blood cells but are not resistant to malaria.
- Malaria
  - Caused by the protist *Plasmodium falciparum*
  - Carried by *Anopheles* mosquito.
Sickle Cell Anemia III

- Hb$^A$Hb$^S$
  - Heterozygous
  - Individuals with this genotype have normal red blood cells but are partially resistant to malaria.
  - The protist *Plasmodium falciparum* spends time within red blood cells during their reproductive cycle.
  - When they enter red blood cells of an individual that has a heterozygote genotype for sickle cell, the cell is most likely to rupture—killing the protist.
Sickle Cell Anemia IV

- $Hb^AHb^S$
  - Heterozygous
  - In places of the world where malaria is prevalent, the sickle cell allele $Hb^S$ is found in higher percentages.
  - Even though the genotype $Hb^SHb^S$ condition is detrimental, the survival of the heterozygotes ($Hb^AHb^S$) within places of malaria makes sense—overdominance (heterozygote advantage).
  - They are less prone to malaria and do not have the severe affects of those suffering with sickle cell anemia.
Sickle Cell Anemia V

- **Hb^S**Hb^S
  - Homozygous recessive
  - Red blood cells have a sickle shape.
  - Sickle-shaped red blood cells.
  - Sickle cells have a shortened life span of a few weeks compared to normal cells which should be months.
  - Sickle cells can clog capillaries causing localized oxygen depletion.
Sickle Cell Anemia VI

- Symptoms
  - Fatigue (feeling tired)
  - Paleness
  - Jaundice (Yellowing of the skin and eyes)
  - Shortness of breath
Review
Review

Genetic Disorders

- Malfunctioning Genes
  - Sex Linked Disorders
    - Hemophilia
  - Autosomal Disorders
    - Huntington Disease
    - Sickle Cell Anemia
    - Baldness