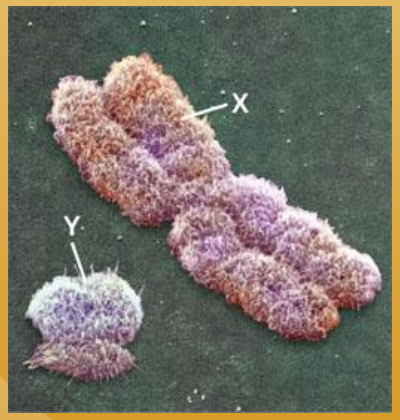
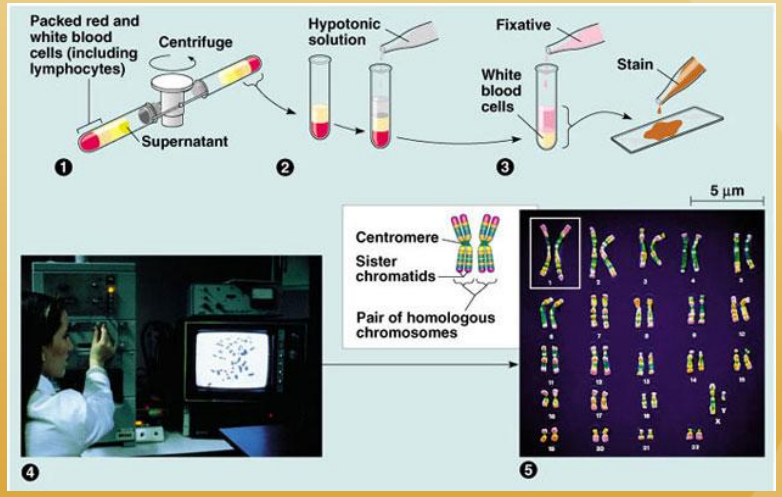
Sex-linked Genetic Disorders & Autosomal Disorders



Packet #15

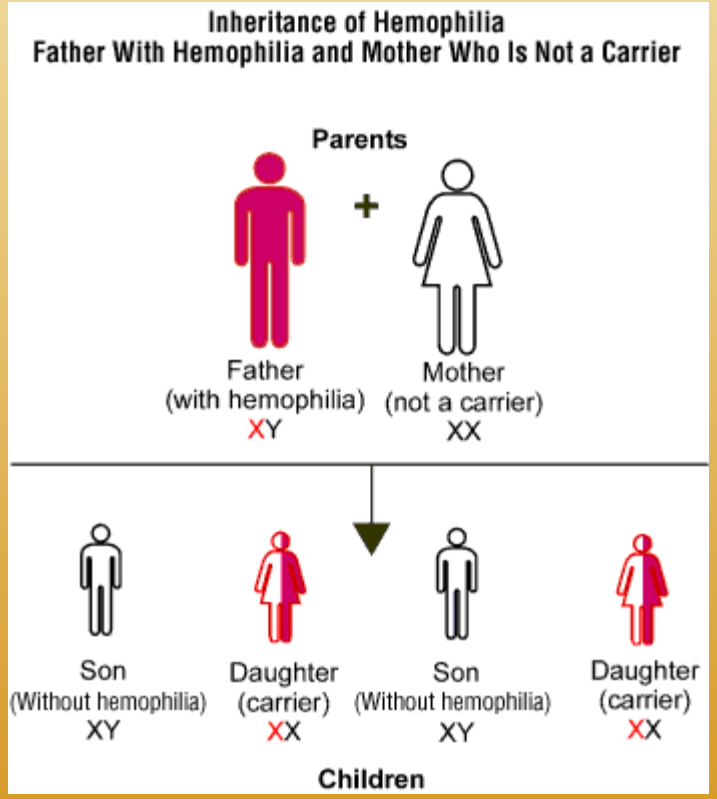
Introduction

- Sex Linked Genetic Disorders
  - Disorders caused by malfunctioning genes on the sex chromosome.
- Autosomal Genetic Disorders
  - Disorders caused by malfunctioning genes on an autosome.



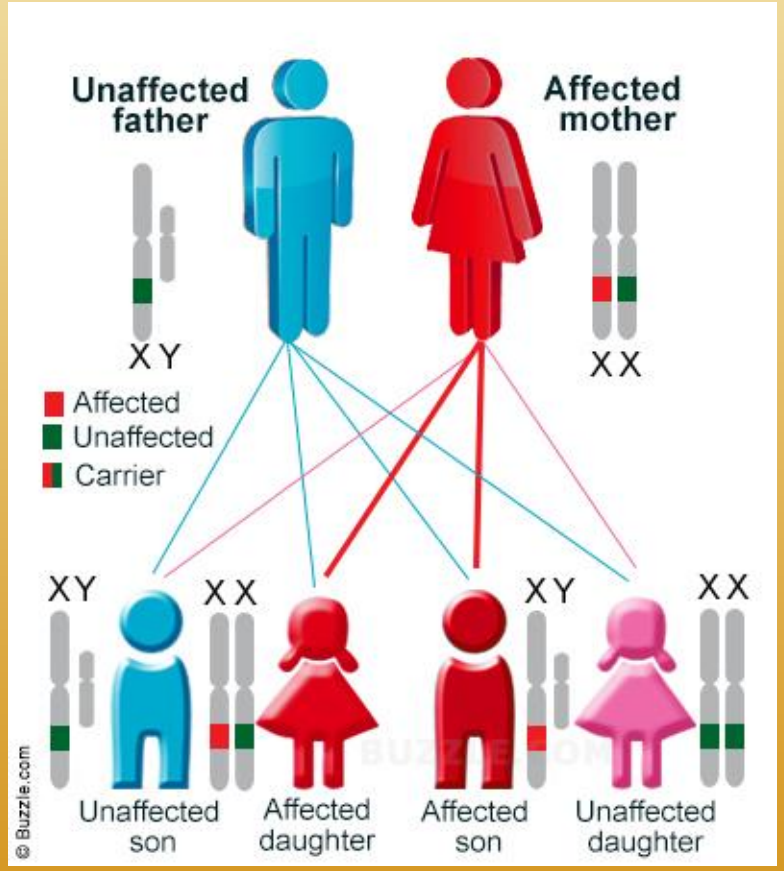
Sex-Linked Disorders

Hemophilia

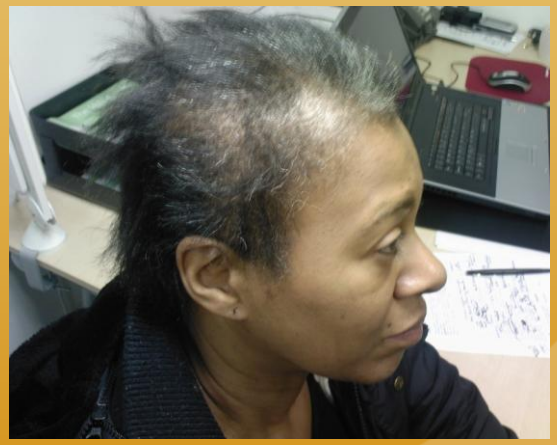


- Hemophilia is an x-linked recessive disorder
  - Males will show this trait if they have the recessive allele on the X chromosome
  - Females will show this trait if they have the recessive allele on both X chromosomes
- Hemophilia does not allow individuals to have the ability to clot their blood.

Baldness

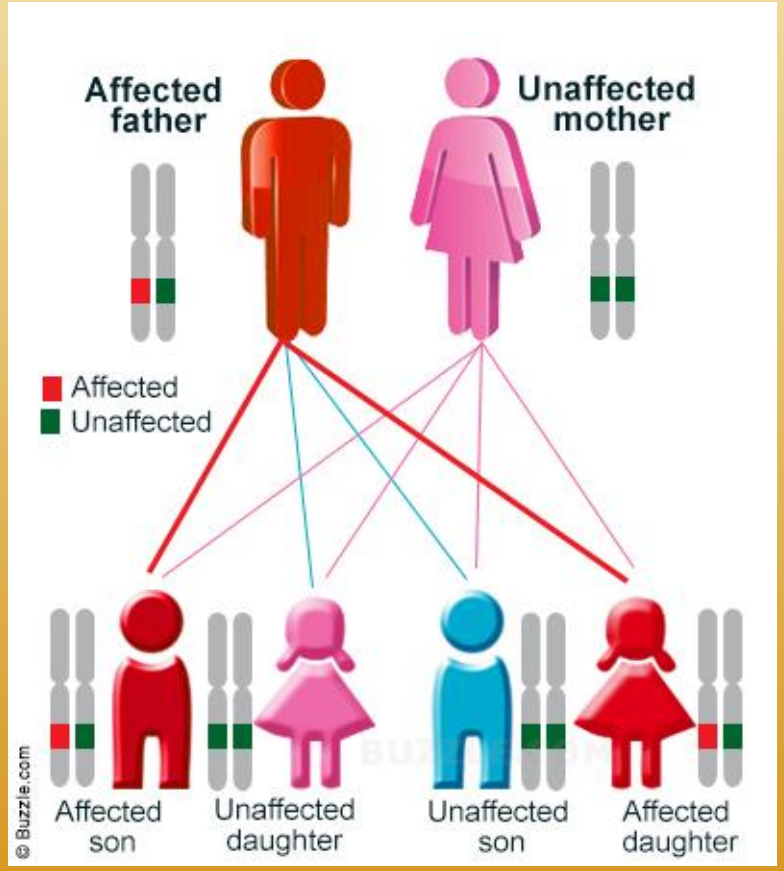


- Baldness is an x-linked dominant disorder
  - **X^BX^b**
    - **This female will not go bald due to lack of testosterone**
  - X^BX^B
    - This individual will start to lose her hair in the future



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

Huntingdon's Disease

Father \ Mother	H	h
h	Hh	hh
h	Hh	hh

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^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

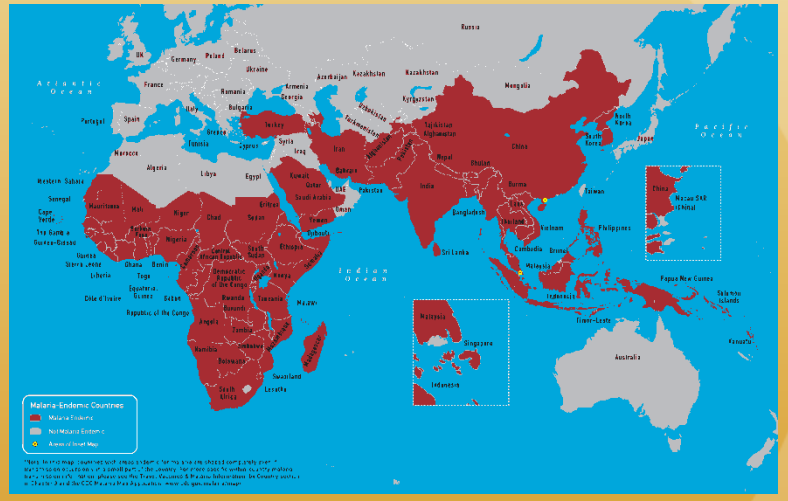
- Hb^AHb^A
  - Homozygous dominant
  - Individuals with this genotype have normal red blood cells but are **not** resistant to malaria.
  - Malaria
    - Caused by the protist ***Plasmodium falciparum***
      - Carried by ***Anopheles*** mosquito.

Sickle Cell Anemia III

- Hb^AHb^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.**

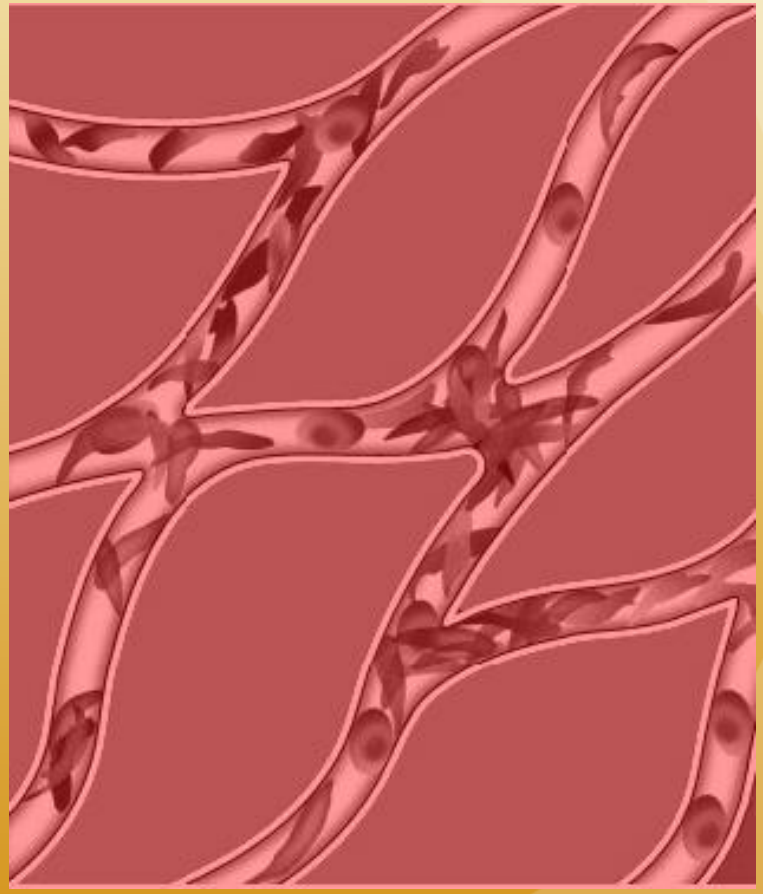
Sickle Cell Anemia IV

- 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.



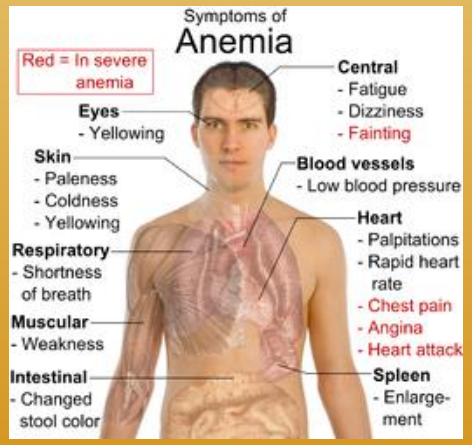
Sickle Cell Anemia V

- $Hb^{S}Hb^{S}$
  - 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**.



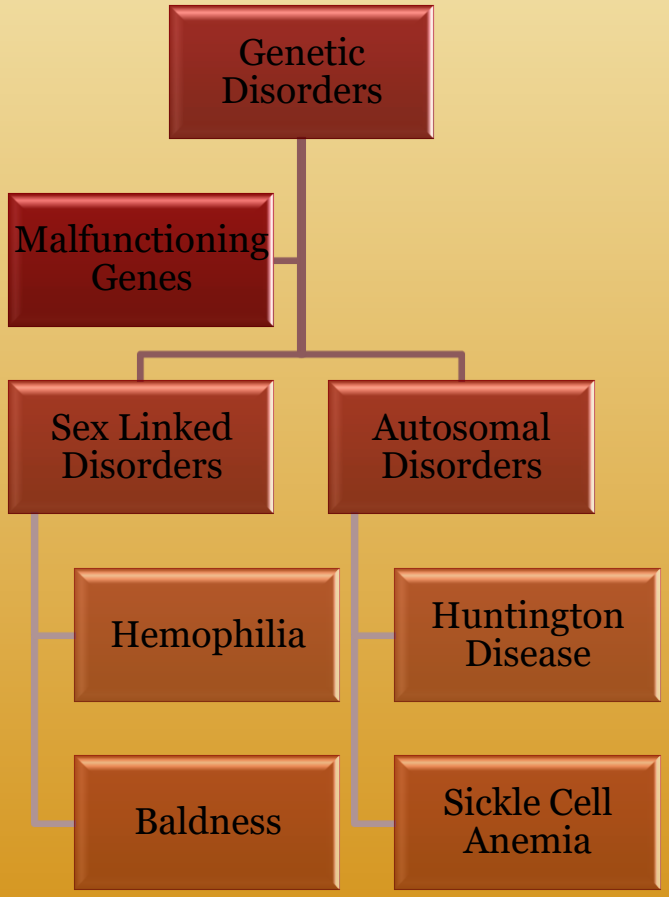
Sickle Cell Anemia VI

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



Review

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