

# Genetic Disorder Notes Week 34

# Genetic Disorders

Any disorder that is caused by malfunctions in genes that are passed on or with the chromosomes.

Examples:

Sickle-cell Anemia

Tay -Sachs

Hemophilia

Albinism

Down's Syndrome

Dwarfism

# Genetic Disorders

## **Sickle-Cell Anemia**

**Caused by a recessive allele**

**It is a homozygous recessive disorder.**

**Affected have a homozygous genotype (rr)**

**Carriers have a heterozygous genotype (Rr) but they are NOT affected**

**Sickle-Cell Anemia**  
**Effects:**  
**Extreme joint pain,**  
**blockages to**  
**capillaries**

# Genetic Disorders

## Tay-Sachs

Also a homozygous recessive disorder.

Affected have a genotype (tt)

Carriers have a heterozygous genotype (Tt)

- **Tay-Sachs Disease**
  - **Cause**
    - homozygous inherited autosomal recessives
  - **Effects (Lack enzyme to break down fatty acids)**
    - Fatty acids accumulate in the brain and effect central nervous system

# Genetic Disorders

## Hemophilia

Is a sex chromosome linked disorder

Transmitted through mother

Affected Boy has  $X^h Y$

genotype

Carriers have a heterozygous

genotype  $X^h X$

# Genetic Disorders



- Hemophilia
  - Cause
    - Inherited recessive allele on the X chromosome
    - Male to male inheritance is impossible
  - Effects
    - Blood is not able to clot normally
    - Minor cuts could possibly lead to bleeding to death



# Genetic Disorders

**C-notes**

Albinism

Also a homozygous  
recessive disorder

Affected have a (aa)  
genotype

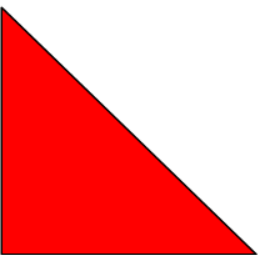
# Genetic Disorders

- Albinism
  - Cause
    - homozygous inherited autosomal recessives
  - Effects
    - lack the enzyme to produce melanin
    - or enzyme that produces melanin is not functional

# Genetic Disorders

**C-notes**

Down's Syndrome  
Caused by one extra  
chromosome in the  
21<sup>st</sup> pair



# Genetic Disorders

## Down's Syndrome

- Caused by non-disjunction of the 21<sup>st</sup> chromosome.
- This means that the individual has a trisomy (3 – 21st chromosomes).

- Dwarfism
  - Cause
    - Inherited dominant allele
  - Effects
    - Stunted growth throughout lifespan