Genetic Disorder Notes Week 34

Genetic Disorders

C-notes

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

C-notes

Sickle-Cell Anemia
Caused by a recessive allele
It is a homozygous recessive disorder.
<u>Affected</u> have a homozygous genotype (rr)
<u>Carriers</u> have a heterozygous genotype (Rr) but they are NOT affected

Genetic Disorders

C-notes

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)

Genetic Disorders

C-notes

- **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

C-notes

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

C-notes

- 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

Albinism
Also a homozygous recessive disorder
Affected have a (aa) genotype

Genetic Disorders

C-notes

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

Genetic Disorders

Down's Syndrome
Caused by one extra chromosome in the 21st pair

Genetic Disorders

C-notes

Down's Syndrome

- Caused by non-disjunction of the 21st chromosome.
- This means that the individual has a trisomy (3 – 2lst chromosomes).

Genetic Disorders

C-notes

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