Inheritance Patterns and Human Genetics

Chapter Twelve

Why study inheritance and genetics?

- Biologists have discovered many human disorders are genetic, which means they can be inherited from parent to offspring
- If we know which genes cause particular disorders, maybe we can alter or control these genes to prevent disorders becoming expressed in our children.

Cystic Fibrosis (CF)

- Cystic fibrosis is a genetic disease that causes persistent lung infections and limits a person's ability to breathe over time.
- Jeff Pinard was diagnosed with cystic fibrosis at a young age after another family member was diagnosed.
- In college, Pinard discovered the gene responsible for cystic fibrosis

Gene Mutations

- Cystic fibrosis results from a gene mutation. There are a few categories of gene mutations
    - Point Mutation: the substitution, addition, or removal of a single nucleotide in a single gene on a chromosome
    - Frameshift Mutation: occurs during transcription

Pedigrees

- Geneticists can study human genetic traits and trace genetic diseases by constructing pedigrees.
- A pedigree is a diagram that shows how a trait is inherited over several generations.
- Pedigrees can help biologists study patterns of inheritance, which is the expression of genes over generations.

Carriers

- Pedigrees can help determine which individuals are heterozygous and homozygous.
- This can help geneticists determine if some individuals are carriers of a disease.
- A carrier is an individual that has one copy of a recessive allele, but does not have the disease.