6.2 Mutations

Mutation: a change in the genetic code

- Can have a positive, neutral or negative effect
- Caused by:
    - Environmental _____ (radiation, chemicals)
    - Errors during cell _____.
- Types of mutations
    1. _____ mutations
    2. _____ mutations

Point mutations

Point mutation: a small-scale change in the nitrogenous base sequence of DNA;

- Base-pair _____ - nucleotide is replaced by a different nucleotide
- _____ - nucleotide added
- _____ - nucleotide is eliminated
- Point mutations are read by _____ and result in the production of altered _____.

Chromosome Mutations

Chromosome mutation: an error that involves an entire chromosome or a large part of a chromosome.

- Having too few or too many chromosomes due to improper separation of chromosomes or sister chromatids
    - E.g. Down syndrome, Edwards syndrome, etc.
- Large scale deletions, insertions or inversions of portions of chromosomes

Inheriting Mutations

- Mutations that occur in the _____ cells are not passed down to offspring
- Mutations that occur in the sex cells (gametes) are passed down
    - Offspring will have the mutation in _____ cell of its body
    - Offspring will pass it down to next generation
    - **Autosomal** _____ **mutations** could be expressed in every generation
    - **Autosomal recessive mutations** may not be expressed for many generations and _____ later on

Sickle Cell Anemia (SCA)

- A harmful and _____ mutation.
    - Disadvantage: affects function of haemoglobin; rbc changes shape (C-shape) when exposed to low oxygen levels
    - Advantages: Protects against malaria
- Substitution mutation: adenine nucleotide substituted for a _____ nucleotide
    - Results in the production of different proteins (altered function)

Lactose Intolerance & Tolerance

- People who are lactose intolerant do not produce _____ (lactase) to break down lactose
- Most people (75%) are lactose intolerant (worldwide)
- Lactose _____ people have a mutation in chromosome 2!
- Genetic tests can help you determine if you are lactose intolerant

Mutations - spontaneous or induced?

- **Spontaneous Mutation:** A mutation that occurs in _____ or by accident (randomly)
    - A result of a copy error in replication for mitosis or meiosis
- **Induced Mutation:** A mutation that occurs because of exposure to an outside factor/ mutagen
    - Examples: radiation, cigarette smoke

Antibiotic Resistance

- describes strains of bacteria (_____) that are no longer susceptible to the effects of antibiotics
- **Normal mechanism of antibiotics:** attach to specific bacterial cell wall causing bacterium contents to _____ out - bacteria dies.
- Problem: a mutation occurs in the bacterial DNA that helps prevent destruction of the cell wall. Resistant genes may be passed down to the next generation.

Jumping Genes

- _____ McClintock discovered that an organism's genome is not static.
- Transposon: a specific segment of DNA that can move along or between chromosomes.
- Transposition: the process of moving a gene sequence from one part of the chromosome to another.

Hemophilia

- Previously learned: hemophilia is an X-linked genetic disorder
- In rare cases, it can also be caused by a transposon if it inserts itself into a normal blood factor VIII gene
    - Males: one X chromosome has to be affected
    - Females: two X chromosomes have to be affected

Microarray

- **Microarray:** a small membrane or glass slide that has been coated in a predictable and organized manner with a _____ sequence
    - Can test hundreds or thousands of DNA fragments and determine if an individual has a specific genetic disorder (genetic screening)
    - Principle: complementary sequences will bind to each other indicating presence of disease

Homework - Page 239 #2-5

6.3 Genomes

The Human Genome Project

- There are 20,000 to 25,000 genes in the human genome
- Human DNA base sequences (order of A, C, T, G) have been analyzed
    - 3 billion base pairs in our DNA
    - The order of the bases offers lots of information

Human Genome

- Human Genome: the sequence of DNA nitrogenous bases found on the 23 sets of chromosomes in humans
- The human genome is composed of both coding and non-coding DNA
    - **Coding DNA** (2% of human genome) - Regions of the DNA that code for genes and proteins
    - **Non-coding DNA** (98% of human genome) - Regions that do not code for proteins. They have other functions in the cell - considered _____ DNA
        - Mutations here are not a concern

Comparison of numbers of genes

- The water _____ has the largest genome followed by the newt (salamander).
- This doesn't necessarily correspond to the number of coding genes.

Functional Genomics

- Functional genomics is used to determine the function or roles of genes in an organism.
- **Model organism:** an organism that can be used to study biological functions of another organism due to genetic _____.
- _____ are model organisms for humans
- Similarities between _____and types of genes
- **Knockout mice:** A protein function Knockout mouse defines an animal model in which one or more nucleotides are mutated in a way that the protein loses its function.

DNA Banks

- DNA information is considered personal (unless under investigation)
- A DNA bank is a database of DNA sequences (stored samples of DNA); the sequences can be from plants, animals or humans
- Can be used for screening and genetic testing
- Can be used to store DNA of endangered species

DNA Fingerprinting

- Prove guilt and innocence, paternity tests, determining ancestry, identifying individuals in unmarked graves, to prosecute hunters or fishers who hunt or fish certain animals out of season, to determine the true source of animal meat.

Homework - Page 244 #1-6 & 8