CONCEPT: HUMAN GENOME AND MEDICINE

- The **human genome project** sequenced the first human _____-

  □ Identified major components of the human genome

  - Protein coding regions make up only 2% of the genome

    - But, each gene can produce more than one protein

  - **Gene rich** regions are areas that are concentrated with genes

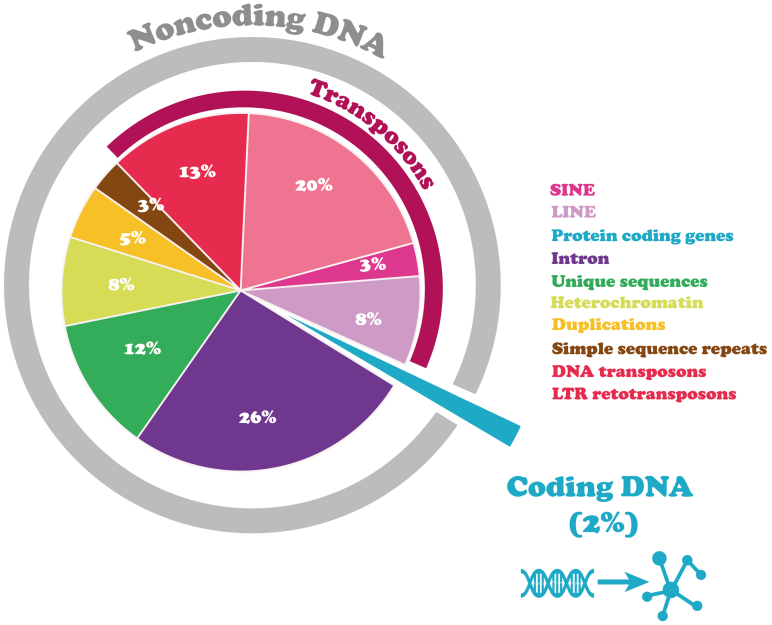
  - **Gene desserts** are regions without any genes

  - It is 99% similar between individuals

    - **Copy number variations** are variations in number of gene copies (due to deletion or insertion)

    - **Single nucleotide polymorphism** are single nucleotide variations that exist between individuals

EXAMPLE: Human Genome Composition



□ Non-coding regions of the genome are just as important as _____ regions

- **ENCODE project** is the encyclopedia of DNA elements

  - Records enhancers, promoters, and other regulatory regions

- **Pseudogenes** are sequences that resemble genes, but are nonfunctional or inactive

Transgenic Organisms and Gene Therapy

● To study human genes, scientists often use transgenic _____

□ **Transgenic organisms** are organisms that contain foreign DNA

- **Gene addition** is when a cloned gene is added to a cell or organism (Ex: glo fish)

- **Gene knockin** is when a gene addition is added to a specific site within a genome

□ Organisms can also be generated with a lack of a gene

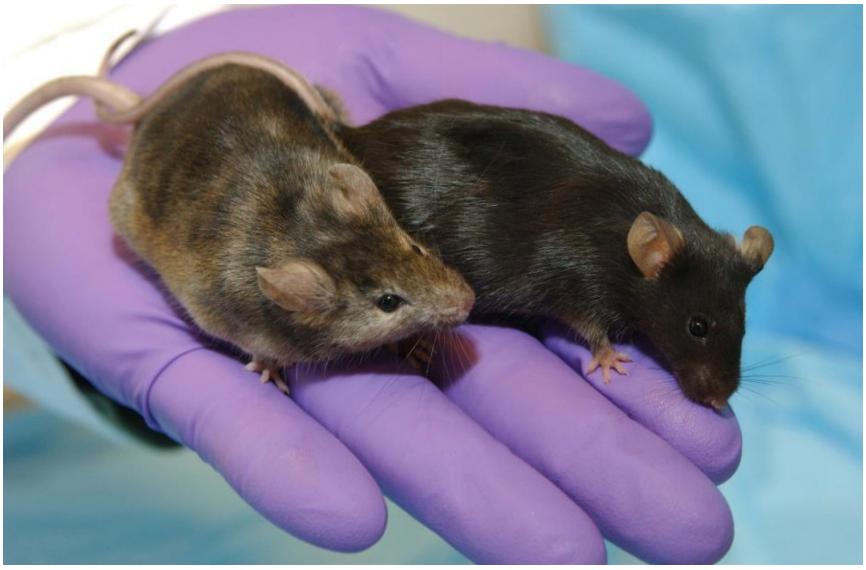
- **Gene knockout** is when the gene is entirely removed or silenced

- **Gene replacement** is when a cloned gene replaces a normal gene in the chromosome

□ These methods create **genetically modified organisms** with altered genomes

- Ex: include developing mice strains of a specific disease, or providing crops with pest resistance

EXAMPLE: Knockout mouse (brown) missing gene for hair growth



● Human gene therapy uses transgenes (foreign DNA) to _____ a disease

□ Personal genomics involves sequencing a diseased individual's DNA and finding a mutant gene

- Then, doctors attempt to add the correct version of the gene back into the cell

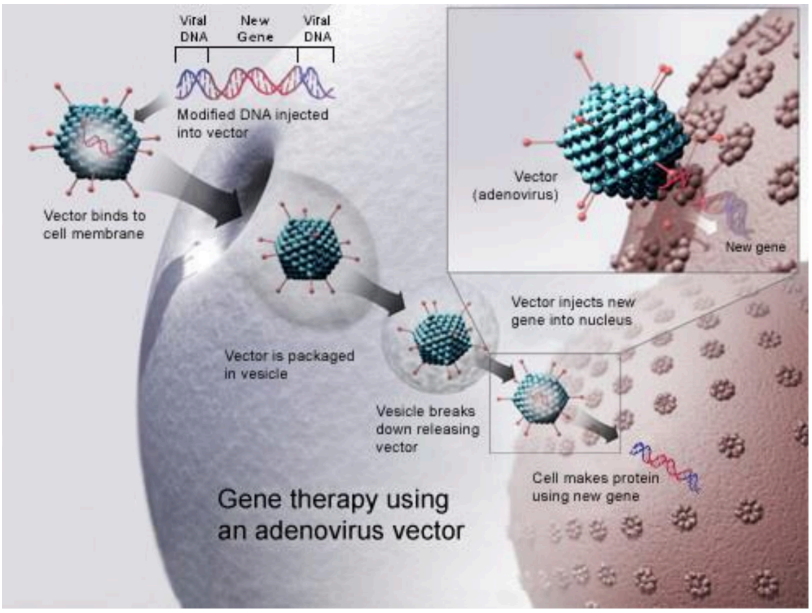
□ Gene therapy involves introducing cloned genes into human cells

- Can uses viruses that inject and incorporate DNA into the human cell

- Can use non-viral methods including liposomes, which are vesicles containing the gene of interest

□ Gene therapy is already being used for diseases like Cystic Fibrosis

EXAMPLE:



PRACTICE:

1. The human genome project discovered that protein coding regions make up what percent of the human genome?
    a. 1%
    b. 2%
    c. 10%
    d. 90%

2. Transgenic organisms are organisms that contain what?
    a. DNA from another organism
    b. RNA from another organism
    c. Protein from another organism

3. The purpose of human gene therapy is to what?
    a. Remove a mutated gene
    b. Remove a mutated RNA
    c. Remove a mutated protein
    d. Add a normal-functioning gene into the organism