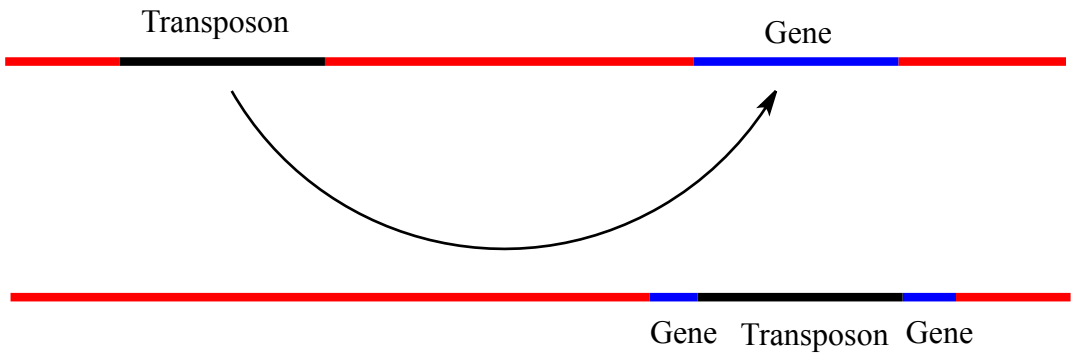
CONCEPT: TRANSPOSONS AND VIRUSES

- **Mobile genetic elements** (jumping genes) are small DNA segments that are found in nearly every cells
  - □ _____ themselves into any DNA sequence, but are unable to leave the cell
    - Make up about 50% of the human genome
    - No function other than the exist (called the "selfish genes")
    - Insert themselves anywhere in the genome
  - □ **Barbara McClintock** discovered them in the 1940s while studying corn
  - □ **DNA transposons** and **Retrotransposons** (RNA based) are the two major families of mobile genetic elements
  - □ Viral genomes, especially *retroviral* genomes, also insert into genome in similar ways as mobile genetic elements

EXAMPLE: Movement of mobile genetic elements



DNA transposons

- **DNA transposons** are mobile genetic elements that move through a _____ intermediate
  - □ Most commonly found in bacteria
    - Eukaryotic DNA transposons have lost their ability to move (3% of human genome)
  - □ Move in the genome by the "cut and paste" method; They are "cut" from one region and "pasted" into another
    - If cut during DNA replication then the transposon can duplicate
  - □ Structure contains inverted repeats of ~50 base pairs flanking a DNA sequence that codes for a *transpose*
    - **Transposase** is the enzyme responsible for _____ the transposon out of the DNA sequence
    - Doesn't lose length when inserting
    - Double strand break repair "pastes" the transposons into a different genomic location

EXAMPLE: Structure of a DNA transposon

DNA Transposon

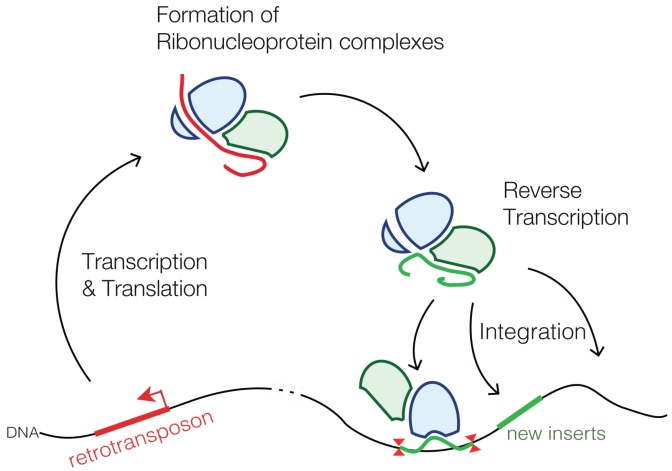
Retrotransposons

● **Retrotransposons** are mobile genetic elements that move through an _____ intermediate

□ They are transcribed into RNA before being processed changed back into DNA for insertion

- **Reverse transcriptase** is the process of changing (reverse transcribing) RNA back into DNA

EXAMPLE: Integration of a retrotransposon into the genome



□ The **Long Terminal Repeat (LTR) retrotransposons** are one class of retrotransposons (8% of human genome)

- Structure consists of direct repeats (250-600 base pairs) that flank a protein coding region

□ **Non-LTR retrotransposons** are a second class of retrotransposons

- **LINES** (Long Interspersed Elements) are _____ commonly found in mammals (6kbp long)

- L1, L2, and L3 are the three classes – Only L1 still transposes

- 21% of human genome

- Structure consists of short direct repeats flanked by two long protein coding regions

- **SINES** (Short Interspersed Elements) are commonly found in mammals (300 bp long)

- Most common is *Alu* element, which still transposes in human genome

- 13% of human genome (10% of SINES are *Alu* sites in humans)

- Most lack protein coding region, and depend on other mobile elements to provide proteins

EXAMPLE: Structure of LINEs

Viruses as Mobile Genetic Elements

● **Viruses** are mobile entities that can _____ their genome into a host cells genome

□ Contain a protein coat surrounding a small amount of genetic information (few – 100s of genes)

- Can be DNA or RNA based

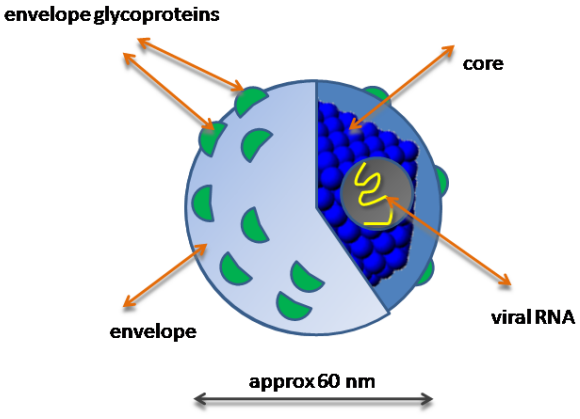
- Disease causing agents

□ Bacteriophages are viruses that infect bacteria and insert their genome into the host bacterium

□ **Retroviruses** store their genetic information as RNA and use it as a _____ to produce DNA

- Reverse transcriptase is typically encoded in the viral genome

EXAMPLE: Structure of a retrovirus



Structure of Hepatitis C Virus

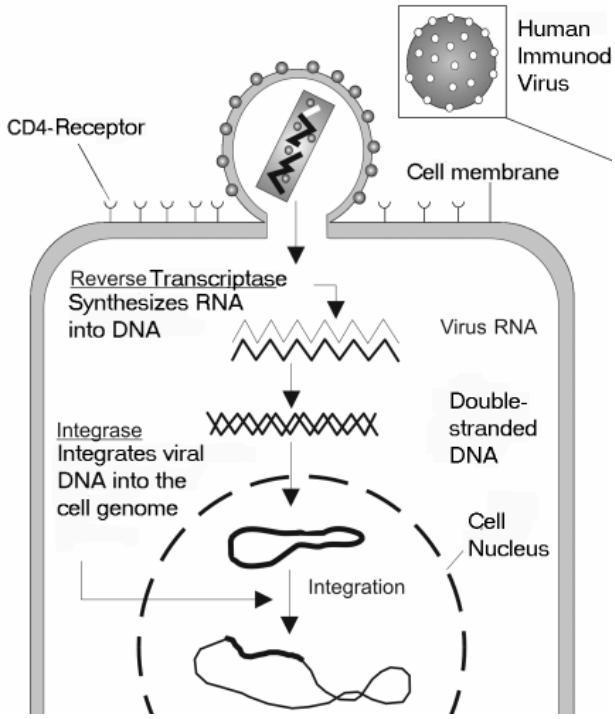
□ Viral genome is generally _____ into the host genome

- **Integrase** is the enzyme responsible for integrating the viral DNA into the host genome

- Does lose length when inserting

- Double strand break repair “pastes” the transposons into a different genomic location

EXAMPLE: Genomic integration of the HIV genome into a host cell DNA



Mobile Genetic Elements in Evolution

● Mobile genetic elements play a major role in _____

□ Mobile genetic elements cause mutations with their jumps

- Bacteria: every 10^5 cell division

  - Can confer antibiotic resistance

- *Drosophila:* 50% of spontaneous mutations

- Mice: has caused about 10% of total mouse mutations

- Humans: 1 in 1000 mutations (0.1%-0.2% of all mutations)

- Can cause disease- Hemophilia

□ Occasionally they can carry additional portions of the genome with them when they move

□ Some important proteins were thought to have evolved from transposable element sequences

- *Transcription factors* bind to DNA and control gene expression

- *Telomerase* is thought to have evolved from a reverse transcriptase encoded by an ancient transposon

□ They can effect expression of genes and proteins

□ They can be inherited

PRACTICE:

1. Which of the following is not considered a mobile genetic element?
    a. DNA transposons
    b. SINEs
    c. Transposase
    d. Retrotransposons

2. Which of the following transposons still jumps in the human genome?
    a. L1 LINE
    b. L2 LINE
    c. L3 LINE

3. What is the name of the enzyme responsible for allowing the transposon to jump within the genome?
    a. Reverse Transcriptase
    b. Transposase
    c. Transposonase
    d. Integrase

4. True or False: Some transposons encode for a protein.
    a. True
    b. False