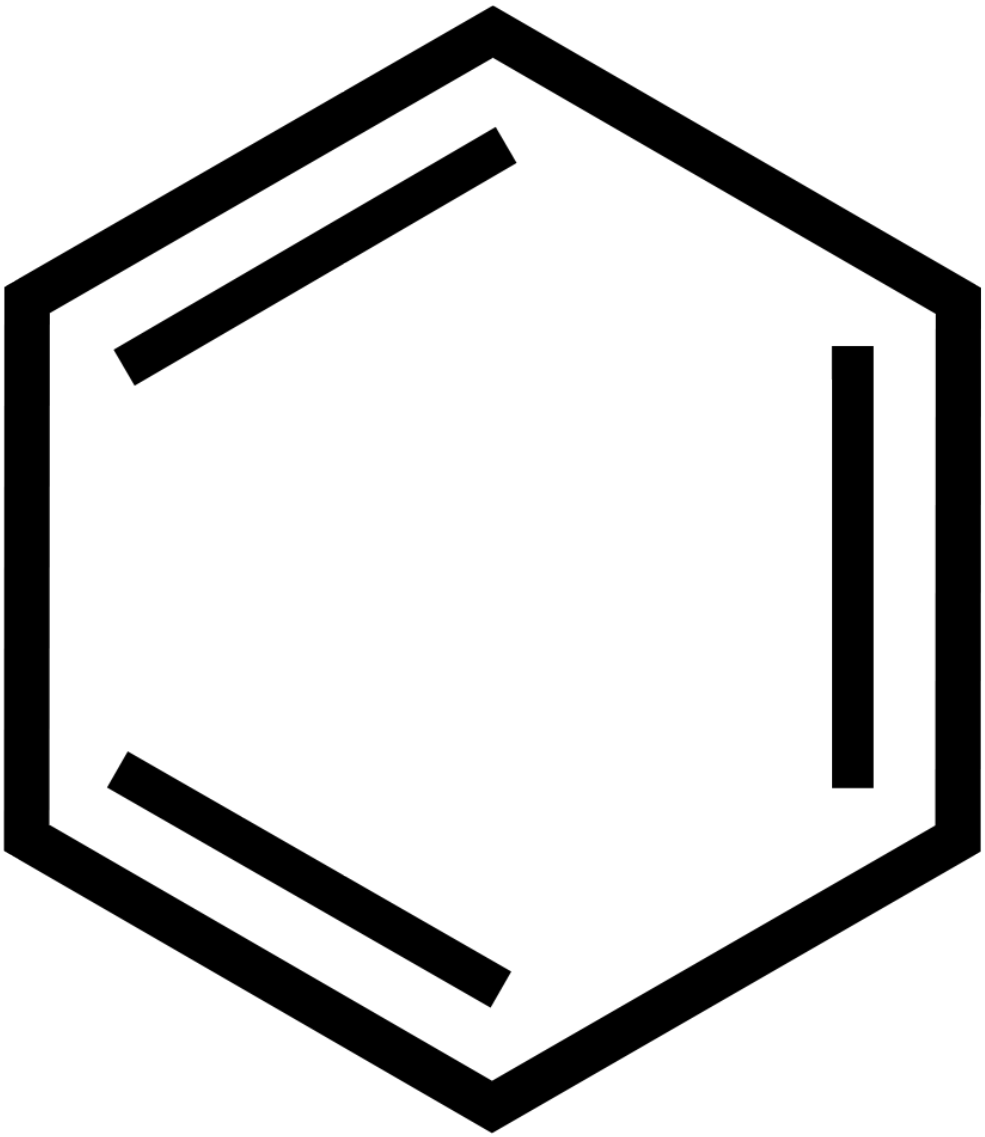

CLUTCH

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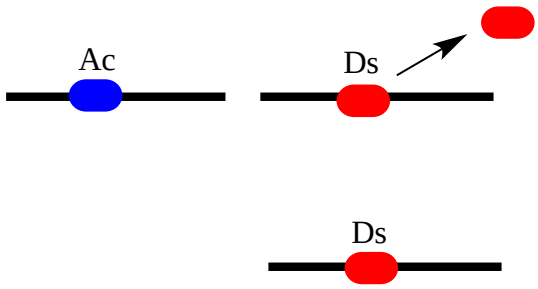
CONCEPT: DISCOVERY OF TRANSPOSABLE ELEMENTS

- **Barbara McClintock** discovered transposable elements in the 1940s
  - □ **Transposable elements** are small DNA segments that can "jump" throughout the genome
    - They are found in nearly every organism
  - □ McClintock studied chromosomal _____ in maize
    - Chromosome 9 tended to break often in the same exact spot
    - **Ds** (disassociation) factor is located at the area of the break
      - **Non-autonomous element** cannot move without assistance from other element
    - **Ac** (activator) is an unlinked factor that controlled the breakage at the Ds location
      - **Autonomous element** can move without assistance
      - Ac was impossible to map

EXAMPLE:



Ds can move only if Ac is present

□ McClintock found that maize kernel color was an **unstable phenotype** that could change after development

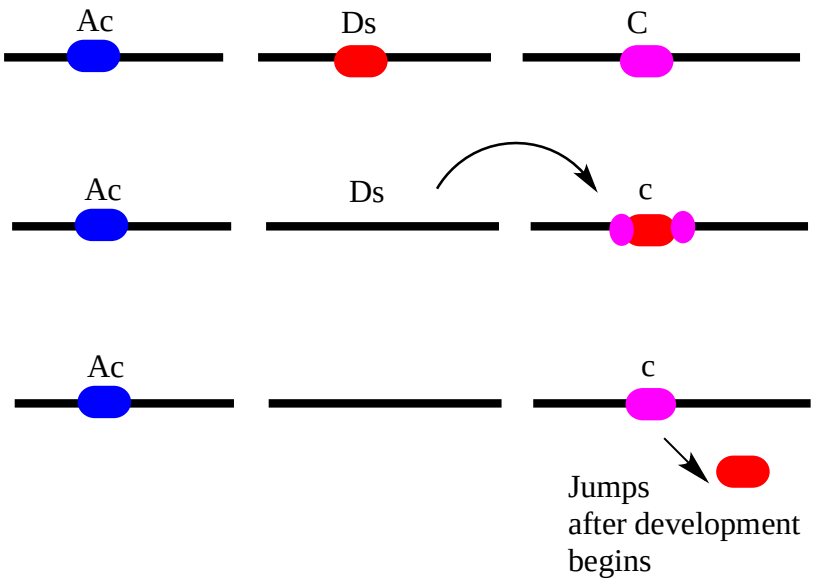
- The C gene controlled corn kernel color

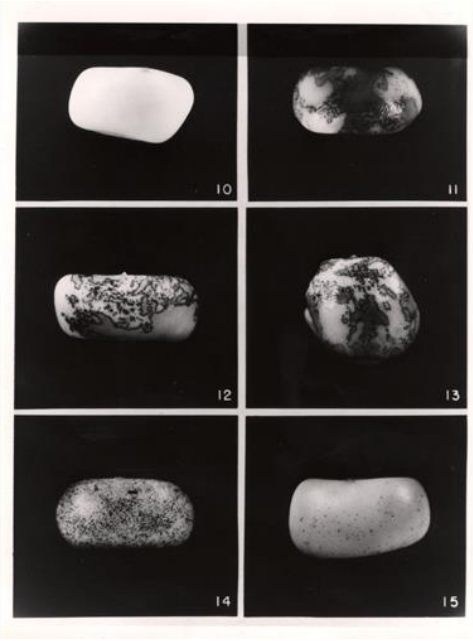
  - Purple had genotype C/-

  - White had genotype c/c

  - White with purple spots had genotype is ???

EXAMPLE:





PRACTICE:

1. In Barbara McClintock's study of corn, which of the following kernel phenotypes did she find was due to transposable elements?
    a. Purple kernel
    b. Blue kernel
    c. White kernel with purple spots
    d. Blue kernel with purple spots

2. A non-autonomous element is a chromosomal element that can what?
    a. Move without assistance
    b. Move with assistance
    c. Remain in the same place
    d. Lock a transposon in place

CONCEPT: TRANSPOSABLE ELEMENTS IN PROKARYOTES

● Prokaryotes have two _____ of transposable elements

□ **Insertion sequence (IS) elements** which are short bacterial DNA sequences that jump around genome

- **Transposase** is the protein required for IS movement

- IS contains two main structural features

- Each end contains an **inverted repeated sequence (IR)** required for mobility

- A transposase gene sits between the IRs

EXAMPLE: Inverted Repeat

TTACG n n n n n n n n n n n CG TAA
AATGC n n n n n n n n n n n G CATT

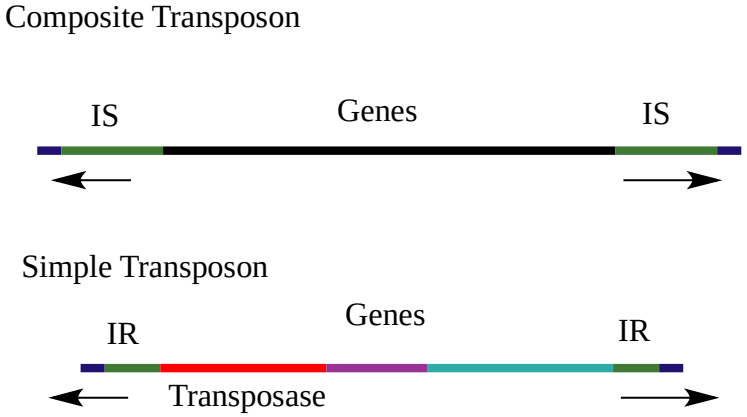
□ **Transposons** are longer DNA sequences that _____ around the genome. There are two types

- **Composite transposons** are flanked by two IS that encode for transposase. Genes sit between IS's

- **Simple transposons** are flanked by IR which surround genes that include transposase

□ Drug resistant genes are transferred between bacterial R plasmids via transposons

EXAMPLE:



□ Transposons transpose in _____- main ways

- **Replicative transposition** is when the transposon is copied and the new copy travels to a new location

  - "Copy and paste" method

- **Conservative transposition** is when the transposon is cut out and moved to new location

  - "Cut and paste" method

EXAMPLE:

Replicative Transposition

Conservative Transposition

PRACTICE:

1. Transposase is a protein that is responsible for what?
    a. Excising a transposable element from a donor site
    b. Insert a transposable element into a target site
    c. Degrading a transposable element that is harming the host genome
    d. Amplifying a transposable element

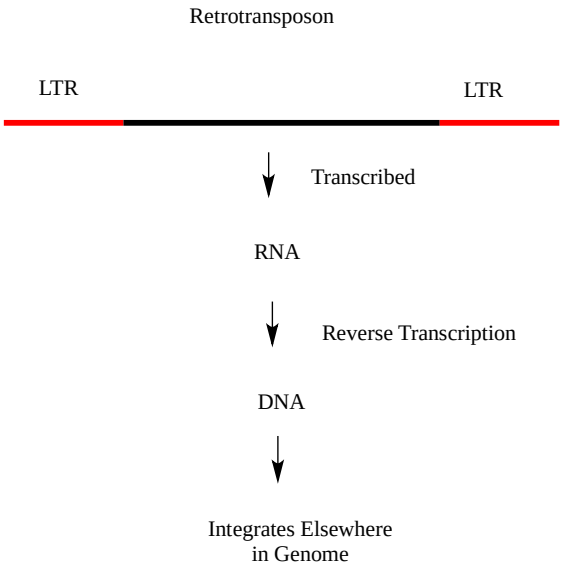
2. Which of the following transposons do not encode for the transposase enzyme?
    a. Insertion sequence elements
    b. Composite transposons
    c. Simple transposons

3. Which of the following sequences is an example of an inverted repeat sequence that would surround one strand of an insertion sequence element?
    a. 5’ AATCG CGATT
    b. 5’ AATCG AATCG
    c. 5’ AATCG TTAGC
    d. 5’ AATCG GCTAA

CONCEPT: TRANSPOSABLE ELEMENTS IN EUKARYOTES

- Eukaryotes have two _____ of transposable elements
  - □ **Retrotransposons (class I elements)** use an RNA intermediate to jump
    - Often, these comes from RNA viruses (retroviruses) that use ssRNA as their genetic material
    - *Reverse transcriptase* transcribes RNA into DNA
    - The **provirus** is the DNA that integrates into the genome
    - Ex: **Long-terminal repeat (LTR) retrotransposons** have long repeats on each end
      - Use "copy and paste" method to transpose
  - □ **DNA transposons (class II elements)** use DNA to jump

EXAMPLE:



Drosophila P Element

● *Drosophila* P element was one of the first eukaryotic _____ identified

□ The P element is a transposon that can severely disrupt the genome

- Strains of flies with this transposon are called P strains

□ If you mate a male P strain with a female M strain (without P element):

- **Hybrid dysgenesis** defines the multiple serious defects of the offspring (mutations, sterility, breakage)

□ If you mate a female P strain with a male M strain (without P element):

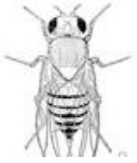
- You get normal offspring

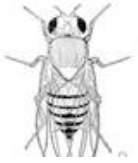
□ Why? The egg in the female P strain can suppress the P element transposons

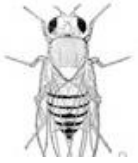
EXAMPLE:

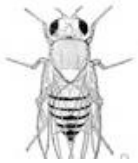
Male P Female M X Male M Female P

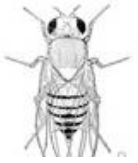
X











Hybrid dysgenesis
(mutated, sterile flies)

Normal Offspring

Human Transposable Elements

● Humans also contain _____ transposable elements

□ **Short interspersed nuclear elements (SINEs)** are one common class of retrotransposons

- ***Alu*** is the most common SINE in humans (300,000 copies)

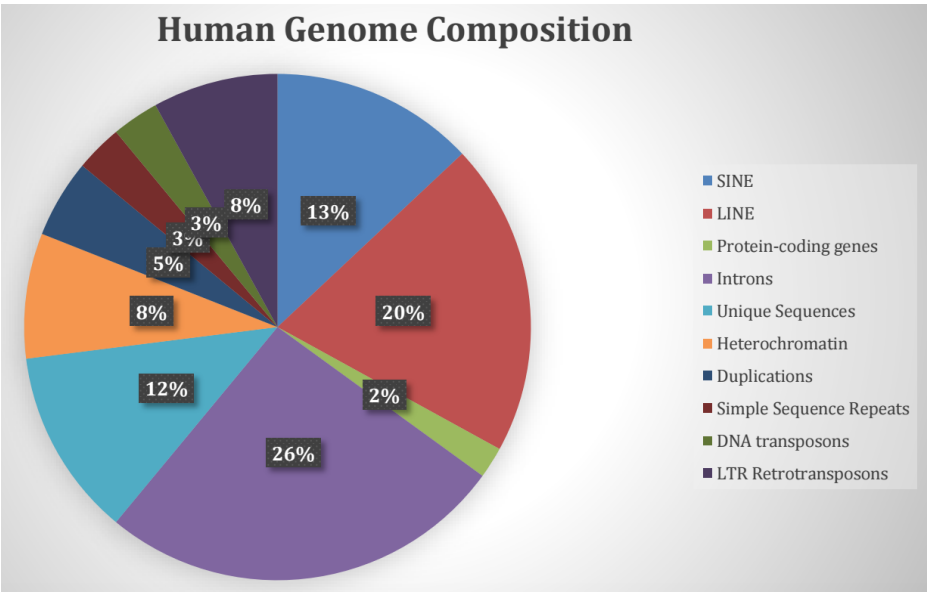
□ **Long interspersed nuclear elements (LINEs)** are a second common class of retrotransposons

- ***L1*** is the most common LINE in humans (20,000 copies)

□ Although most human transposable elements do not move, some still do and can cause disease

- When they do just most do in **safe havens**, which are non-gene regions safe for genes to jump (introns)

EXAMPLE:



● Transposons have a great impact on genome evolution

□ They can cause gene mutations by inserting inside of a gene or a gene regulatory region

□ They can cause chromosomal rearrangements

□ They can relocate genes to new regions

PRACTICE:

1. Which of the following is true regarding reverse transcriptase?
    a. It is required for transposition of DNA transposons
    b. It synthesizes DNA from RNA
    c. It is encoded within long-terminal repeats of retrotransposons
    d. The provirus uses it to insert into the genome

2. Which of the following elements is a transposable element in *Drosophila*?
    a. *Ds* element
    b. *Alu* element
    c. P element
    d. *Ac* element

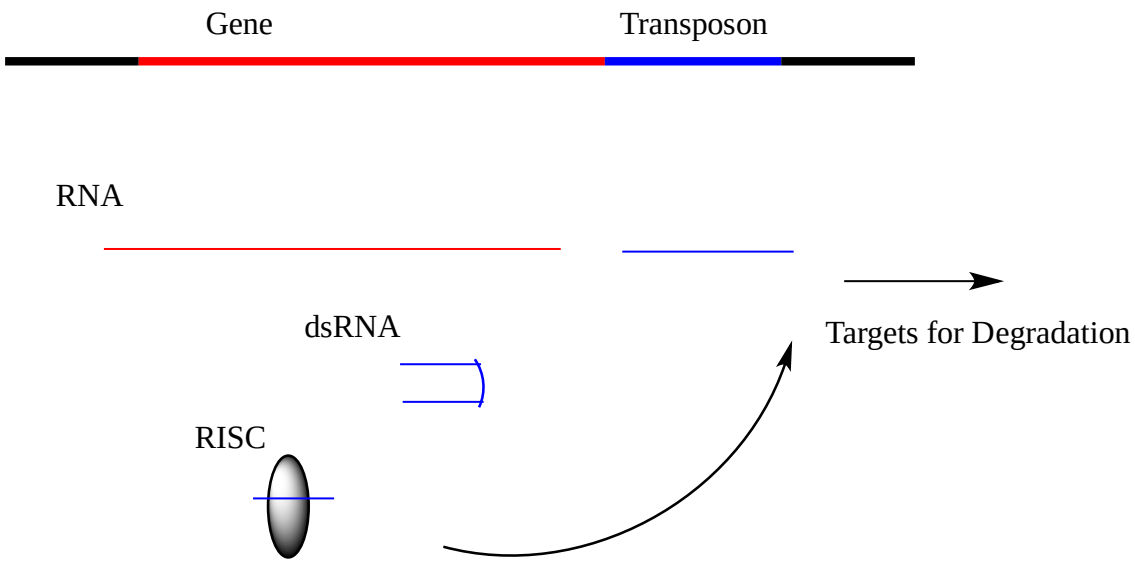
3. Which of the following is an example of a safe haven for transposon movement?
    a. Intron
    b. Exon
    c. Promoter
    d. Enhancer

4. Which of the following would occur in an *Alu* element jumped into the AG splice site of a human gene?
    a. Splicing would occur, and the protein would be unaffected
    b. Translation would not occur on any part of the mRNA
    c. Splicing would not occur and the protein would be altered
    d. Chaperone proteins would correct the damage, and the protein would be unaffected

CONCEPT: REGULATION OF TRANSPOSON MOVEMENT

- Regulation of transposon movement is not well understood
  - □ But, scientists have found that _____ RNAs can silence and stop transposon movement
  - □ The Tc1 transposon in *C. elegans* is present in somatic and germ cells, but only expressed in somatic cells
    - Tc1 is transcribed as part of other genes
    - Tc1 RNA contains repeated sequences that cause it to fold upon itself
    - The cell recognizes dsRNA and causes DICER and RISC to process it (From RNA interference)
    - When RISC binds the Tc1 processed RNA it targets it to degrade other Tc1 transcripts

EXAMPLE:



- □ **piRNA** helps regulate transposons in _____
  - **Pi-clusters** are >100kb of DNA that contain transposons that are transcribed
  - These long RNA transcripts are processed and complexed with *argonaute*
  - This complex travels around the cell degrading other transposons
- □ **crRNA** helps regulate transposons and viral DNA in bacteria
  - RNA from transposons or viruses are captured by **CRISPR**, which targets them for degradation

PRACTICE:

1. Which of the following molecules are known to be able to regulate transposon movement?
    a. DNA
    b. RNA
    c. Protein

2. Which type of RNA is known to regulate transposon movement in *C. elegans*?
    a. Tc1 RNA
    b. piRNA
    c. miRNA
    d. crRNA