

Section 2: DNA Mutations: Substitutions, Insertions, and Deletions

Learning Objective: Differentiate between substitution, insertion, and deletion gene mutations, and analyze why frameshift mutations often have severe effects on proteins.

Key Vocabulary: mutation, point mutation, substitution, insertion, deletion, frameshift mutation, codon, reading frame.

A **mutation** is any change in the nucleotide sequence of DNA. Mutations can occur spontaneously during DNA replication errors in the S phase, or from environmental factors called mutagens (such as UV radiation).

There are three primary types of single-gene point mutations:

- **Substitution:** One nucleotide base is swapped for a different base. This may change a single amino acid or cause no change at all (silent mutation).
- **Insertion:** One or more extra base pairs are added into the DNA strand.
- **Deletion:** One or more base pairs are removed from the DNA strand.

Substitution 

Insertion 

Deletion 



Frameshift Alert: Because ribosomes read mRNA in three-base words called **codons**, adding or deleting bases shifts the entire **reading frame**, altering every codon downstream!

Compare each mutated DNA sequence to the original reference strand. Identify the mutation type (**Substitution**, **Deletion**, or **Insertion**) Write **Yes** or **No** if it shifts the frame. **SPELLING WILL COUNT!**

Original Reference Strand: TAC - GGT - CAA

Mutated Strand 1: TAC - GAT - CAA

Type: _____ Frameshift? _____

Mutated Strand 3: TAC - GTC - AA

Type: _____ Frameshift? _____

Mutated Strand 2: TAC - GGT - CAA - T

Type: _____ Frameshift? _____

Mutated Strand 4: TAT - GGT - CAA

Type: _____ Frameshift? _____

2. The sentence below is an analogy for the genetic code, where each three-letter word represents a three-base codon:

Original Sentence: THE BIG RED CAT ATE THE WET RAT

a) Rewrite the sentence showing a **substitution** of the letter 'H' with 'O' in the first word:

b) Rewrite the sentence showing a **deletion** of the letter 'B' in the second word, preserving the strict three-letter reading frame:

c) Which of the two mutated sentences above makes more sense overall? How does this analogy explain why frameshifts are usually more damaging than substitutions?

Explain here...

Section 3: From DNA to Chromosomes

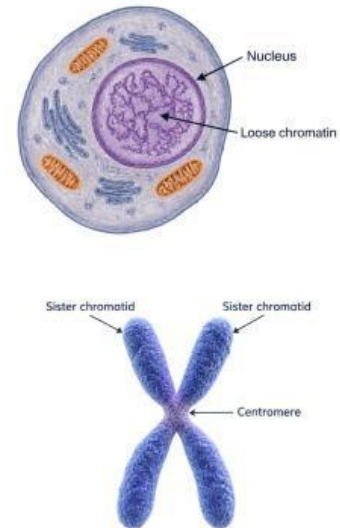
Learning Objective: Explain that genes are sections of DNA and describe how DNA is tightly packaged into chromosomes inside the cell nucleus.

Key Vocabulary: gene, DNA, nucleus, histone, chromatin, chromosome, sister chromatids, centromere.

In eukaryotic cells, the nucleus contains DNA. A **gene** is a section of DNA that carries instructions, and each DNA molecule contains many genes.

The cell winds DNA around spool-like proteins called **histones**. During Interphase, DNA is generally less tightly packed as **chromatin** than it is in a dividing chromosome. This open arrangement lets enzymes and other cell machinery reach DNA. The DNA is what instructs ALL cell processes. DNA must also be accessible for **replication** during S phase and for DNA repair.

Before a cell divides, it copies its DNA and packs the chromatin more tightly into visible **chromosomes** inside the nucleus. Each chromosome contains many genes along its DNA. After DNA is copied, a duplicated chromosome has two identical copies called **sister chromatids**, joined at a region called the **centromere**.



2. Examine the provided diagram of a duplicated chromosome. Use the terms in the bank below to identify and define the key structural features:

Word Bank: Sister Chromatid • Centromere • Histone Protein • Chromatin

a) Name the structure that holds two identical copies of a replicated chromosome together:

b) Name one individual copy of the duplicated chromosome: _____

c) Describe the genetic relationship between the two sister chromatids joined at a single centromere. Are their DNA sequences identical, similar, or completely different?

3. Compare loose chromatin with tightly condensed chromosomes by answering the prompts below:

Why must DNA remain in the loose, uncoiled **chromatin** form during the cell's daily life (Interphase)?
What cellular processes require access to uncoiled DNA?

A. DNA is tightly packed so the cell can read genes and repair DNA more quickly.

B. Loose chromatin allows cell machinery to access DNA for transcription, DNA replication, and DNA repair.

C. Chromatin must remain loose only so chromosomes can move through the cytoplasm during Interphase.

D. DNA stays uncoiled because the cell does not use its genetic information during Interphase.

5. Use the diagram to label each numbered structure. Use each word-bank term once.

Word Bank: chromosomes • proteins • DNA • genes • nucleus

