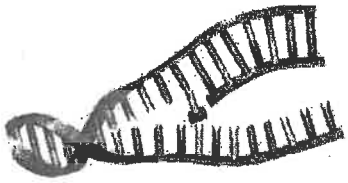
Mutations Worksheet - Deletion, Insertion & Substitution



There are several types of mutations:

- **DELETION** (a base is lost/deleted)
- **INSERTION** (an extra base is added/inserted)
  - --- Deletion & insertion may cause what's called a **FRAMESHIFT** mutation, meaning the reading "frame" changes, thus changing the amino acid sequence from this point forward
- **SUBSTITUTION** (one base is substituted for another)
  - --- If a substitution **changes** the amino acid, it's called a **MISSENSE** mutation
  - --- If a substitution **does not change** the amino acid, it's called a **SILENT** mutation
  - --- If a substitution **changes the amino acid to a "stop,"** it's called a **NONSENSE** mutation

Complete the boxes below. Classify each as **Deletion, Insertion** or **Substitution** AND as either **frameshift, missense, silent** or **nonsense** (Hint: Deletion & Insertion will always be frameshift).

Original DNA Sequence: T A C A C C T T G G C G A C G A C T

mRNA Sequence: _____

Amino Acid Sequence:

Mutated DNA Sequence #1: **T A C A T C T T G G C G A C G A C T**

What's the mRNA sequence? (Circle the change) _____

What will be the amino acid sequence? _____

Will there likely be effects? _____

What kind of mutation is this?

Mutated DNA Sequence #2: **T A C G A C C T T G G C G A C G A C T**

What's the mRNA sequence? (Circle the change) _____

What will be the amino acid sequence? _____

Will there likely be effects? _____

What kind of mutation is this?

Mutated DNA Sequence #3: **T A C A C C T T A G C G A C G A C T**

What's the mRNA sequence? (Circle the change) _____

What will be the amino acid sequence? _____

Will there likely be effects? _____

What kind of mutation is this?

Mutated DNA Sequence #4: **T A C A C C T T G G C G A C T A C T**

What's the mRNA sequence? (Circle the change) _____

What will be the amino acid sequence? _____

Will there likely be effects? _____

What kind of mutation is this?

Name_____

Karyotyping Activity

Period:_____

In this activity, you will use a computer model to look at chromosomes and prepare a karyotype. You will diagnose patients for abnormalities and learn the correct notation for characterizing karyotypes.
Go to **www.biology.arizona.edu**
Click on **Karyotyping** under human biology.

Introduction:

1. What causes a dark band on the chromosome? _____
_____
2. What 3 things are used for comparison to pair chromosomes? _____
_____
_____
3. What is a centromere? _____

Patient Histories:

*Click on Patient Histories. You will be completing a karyotype for Patient A, B & C.
*Match the chromosome to its homolog. After all the matches are completed correctly you will analyze your patient. (Scroll down to view your completed karyotype).

Patient A (Click on the link to "Complete Patient A's Karyotype")

4. What is patient A's history (summarize) _____
5. How many total chromosomes are in your karyotype - count them _____
6. The last set of chromosomes is the sex chromosomes, if you have two large chromosomes, your patient is XX (female), one large and one small indicates and XY (male) . What sex chromosomes does your patient have _____
7. Which chromosome set has an extra + _____
5. What diagnosis would you give this patient (what disease)? _____

Patient B – (Click on the link to go to Patient B and repeat the above process)

6. What is Patient B's history (summarize) _____
7. Finish the notation for this patient's karyotype : 47 X _____
8. What is the diagnosis? _____

Patient C – (Click on the link to go to Patient C and repeat the above process.)

9. Write out the correct notation for this karyotype. _____
10. What is the diagnosis? _____