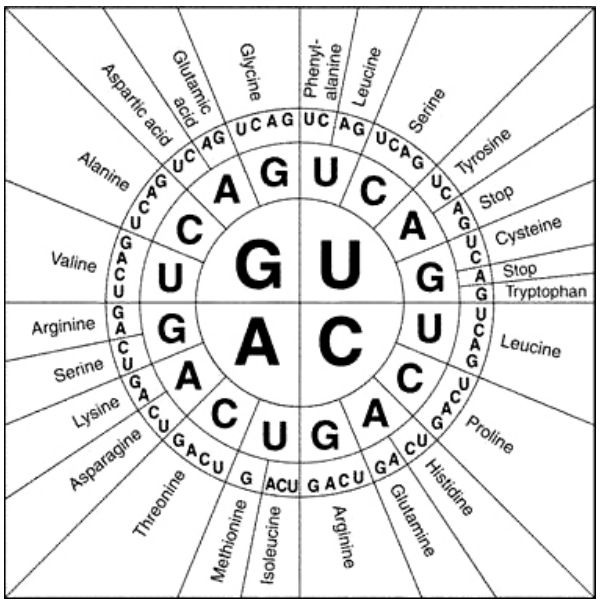
**Biology**
**The Molecular Biology of Mutations: Muscular Dystrophy**

1. The table shows five different mutations of a small section of a gene.

- Use the base-pairing rules to complete the second column.
- Use the genetic code chart to translate the mRNA, and write in the amino acid abbreviations. Highlight stop codons and amino acid changes; use dashes to indicate any amino acids that will be missing as a result of stop codons.
- Identify the type of mutation from among these possibilities:
  - **Substitution / Silent Mutation** → a single base is substituted for another, causing no changes to the amino acid sequence
  - **Substitution / Missense Mutation** → a single base is substituted for another, causing a single amino acid change
  - **Substitution / Nonsense Mutation** → a single base is substituted for another, causing a premature stop codon
  - **Deletion / Frame Shift Mutation** → one or more bases is/are deleted, causing a frame shift which changes multiple amino acids or causes a premature stop codon
  - (more details about these mutations available in this slideshow)




| **DNA** (Template Strand) | **mRNA** codons (Highlight Stop Codons!) | **Amino Acid Sequence in Protein (Highlight Changes, -- if Missing!)** | **Mutation Type** |
|---|---|---|---|
| Original (wild type) DNA = GCA AGT ACC TGA | CGU UCA UGG ACU | arginine serine tryptophan threonine | None |
| Mutation 1 = GC**C** AGT ACC TGA (nucleotide change underlined) | CGG ___ ___ ___ | ___ ___ ___ ___ | _____ |
| Mutation 2 = GCA **C**GT ACC TGA (nucleotide change underlined) | ___ ___ ___ ___ | ___ ___ ___ ___ | _____ |
| Mutation 3 = GCA AGT AC**T** TGA (nucleotide change underlined) | ___ ___ ___ ___ | ___ ___ ___ ___ | _____ |
| Mutation 4 = GA**A GTA CCT GAT** (first C deleted) | ___ ___ ___ ___ | ___ ___ ___ ___ | _____ |
| Mutation 5 = GCA AGT AC**T GAT** (second C deleted) | ___ ___ ___ ___ | ___ ___ ___ ___ | _____ |

2. Explain why deletion of a single nucleotide in a gene generally results in more severe defects in a protein than a substitution mutation that changes a single nucleotide.
Sentence Frames:

_____ results in more severe defects than _____ because _____

Evidence from _____ shows that _____


Modified from an original activity by Dr. Ingrid Waldron, Department of Biology, University of Pennsylvania, 2011

_____ has a different effect than _____ because

3. Muscular Dystrophy (MD) includes several genetic diseases that cause increasing weakness of a person's muscles. Two different types of muscular dystrophy are caused by different types of mutations in the gene for a protein (dystrophin) that helps to prevent muscle cells from dying prematurely.

Duchenne Muscular Dystrophy is more severe. A child with Duchenne MD begins to show symptoms of loss of muscle function by about age 3 and needs to use a wheelchair by about age 10. A person with Duchenne MD typically dies as a young adult, due to failure of the muscles in the internal organs, including the heart.

Becker MD is milder. Symptoms do not begin until age 12 or later, and the person lives into their 40s or 50s.

The table below shows the main kinds of mutations that are responsible for these two types of muscular dystrophy. Complete the second column to indicate which type of muscular dystrophy you think would be caused by each type of mutation.

| Mutation # (From Page 1) | Mutation Type | Type of Muscular Dystrophy |
|---|---|---|
| 1 | | |
| 2 | | |
| 3 | | |
| 4 | | |
| 5 | | |

Explain your reasoning for why you classified each mutation type with a specific type of MD . Use the sentence frames below or write your own sentences

_____ is likely caused by _____ resulting in….

This makes sense because...

The protein _____ function because

_____.

Compare/ contrast signal words:

| | | | | |
|---|---|---|---|---|
| however | while | whereas | on the other hand | |

Extension: search YouTube for one or more videos that chronicle the life of a person with each type of muscular dystrophy: Duchenne and Becker.

Muscular Dystrophy Mutation Activity Self-Evaluation:

| 7-8 | 5-6 | 3-4 | 1-2 |
|---|---|---|---|
| 4 | 3 | 2 | 1 |
| All of the previous plus:<br>❏ Contrast frameshift & point mutations. (2)<br>❏ Support the claim | Students will be able to:<br>❏ Translate & Transcribe the DNA to mRNA and amino acid sequence. (1)<br>❏ Identify silent and missense mutations<br>❏ Identify nonsense mutations due to | Students can sometimes:<br>❏ Translate & Transcribe the DNA to mRNA | Students are not yet able to:<br>❏ Translate & Transcribe the DNA to mRNA |

| | | | |
|---|---|---|---|
| with evidence. (3) | substitution and frame shift<br>❑ Make a claim as to which types of mutations lead to Duchenne and Becker MD | and amino acid sequence. (1) | and amino acid sequence. (1) |

**I would give myself a _____ because _____ .**