Teacher Notes for
The Molecular Biology of Mutations and Muscular Dystrophy

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In this discussion/worksheet activity students explore the effects of different types of point mutations and deletion mutations and analyze the reasons why deletion mutations generally have more severe effects than point mutations. Students use their understanding of the molecular biology of mutations to analyze the genetic basis for the differences in severity of two types of muscular dystrophy. To maximize student participation and learning, you may want to have your students complete the questions individually or in pairs and then have a whole class discussion.

To answer the questions in this activity, students need to understand the genetic code and the processes of transcription and translation. To help your students learn this needed background, I recommend the hands-on simulation activity, "From Gene to Protein - Transcription and Translation" (available at http://serendip.brynmawr.edu/sci_edu/waldron/#trans).

Teaching Points

- A mutation is a permanent change in the DNA of a gene.
- A change in a single nucleotide is a point mutation. Some point mutations result in a change in a single amino acid in the polypeptide produced by transcription and translation of the gene. Other point mutations change a codon to another codon for the same amino acid, resulting in no change in the amino acid sequence of the polypeptide. In other cases, a point mutation results in a stop codon which terminates translation and can result in a drastically shortened, nonfunctional protein.
- If a deletion mutation results in the deletion of one nucleotide from the mRNA molecule or the deletion of any other number of nucleotides that is not a multiple of 3, this causes a frameshift during translation of the mRNA. Every codon after this frameshift is changed, which usually results in the production of a nonfunctional protein. In contrast, a deletion mutation that results in the deletion of three nucleotides or a multiple of three nucleotides from the mRNA molecule will have less severe consequences since there is no frameshift and the subsequent codons will not be affected. This same frameshift rule applies to the effects of insertion mutations.
- Most cases of the more severe Duchenne muscular dystrophy result from frameshift deletion mutations or point mutations that produce a stop codon, while the milder Becker muscular dystrophy results from deletion mutations that do not cause a frameshift.

Suggestions for Discussing Student Handout Questions, with Biology Background

1- 2. The effects of the various mutations should be related to the students' understanding of the process of translation (including the effects of stop codons and the effects of shifts in the reading frame for an mRNA molecule in the ribosome).

The first mutation shown is typically described as a silent mutation since there is no change in the amino acid sequence in the polypeptide. However, recent research has shown that some so-called silent mutations are harmful because they affect RNA folding and thus the rate of breakdown of the RNA transcript or because they serve as a better or worse marker of the end of an exon.

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[1] These teacher notes, the student handout for this activity, and links to additional activities are available at http://serendip.brynmawr.edu/exchange/bioactivities.