Name _____ Date _____ Class _____

Real-World Biology: **Analysis**

CHAPTER 12

Mending Mutations

You might know someone who has asthma, arthritis, cystic fibrosis, or sickle-cell disease. These are diseases that are caused by genetic mutations. In recent years, scientists at the Human Genome Project have determined that there are more than 30,000 genes in the 46 human chromosomes. Each gene is a segment of DNA that codes for a specific protein such as eye color. Researchers are now making progress in developing methods of gene therapy for mutations. The first type of therapy developed was technology that corrected a genetic disorder by replacing the defective gene with copies of healthy ones. Scientists continue to refine the technology with improved methods for therapy. They are also identifying additional specific genes that control disorders such as cancer, diabetes, and Alzheimer's disease.

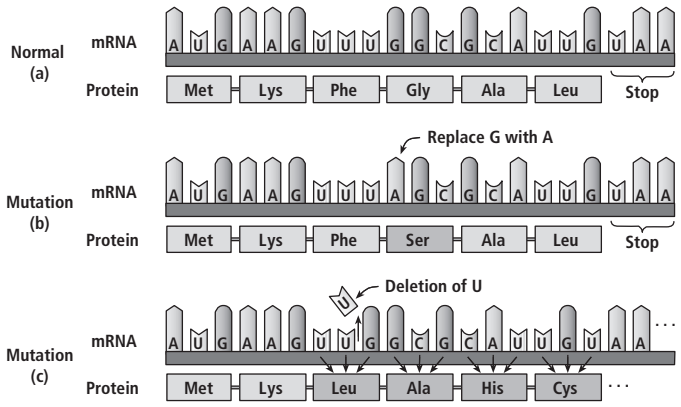

Figure 1

Part A: Finding the Mutation

A team of scientists in Finland has been working for more than ten years on what might be called "the genetics of wheezing." Their work can be best described as a series of steps. Their first step linked increased susceptibility to asthma to a 20-million-base area of Chromosome 7. Their second step in hunting the gene was to collect 900 blood samples from both healthy and asthmatic individuals in families afflicted with asthma. Their third step was to search for large sequences of DNA called haplotypes; these might predispose people to disease. Among the Finns, seven haplotypes were found in the stretch of Chromosome 7. Three of the seven haplotypes were present in more than 50 percent of the asthma patients. They were present in 30 percent of the healthy people. The presence of these haplotypes increased the risk of asthma up to 2.5 times the normal rate.

Analyze and Conclude

*Use **Figure 1** to respond to each question.*

1. **Propose** The team has now associated three haplotypes with high risk for asthma. The next step is to find the gene or genes involved. How can that be done?

2. **Explain** Before a genetic disorder can be corrected, it is necessary to identify the defective gene that is causing the disorder. What type of mutation is illustrated in diagram (b)? How is the protein produced in (b) different from the protein produced by the normal gene (a)?

3. **Apply** What type of mutation is illustrated in diagram (c)? What would have to be done to correct the defective gene?

Part B: Can mutations be mended?

Cystic fibrosis (CF) is an inherited disease caused by an abnormal protein that does not allow the passage of chloride ions into and out of certain cells. As a result, those cells produce thick, sticky mucus and other secretions that clog the lungs and digestive tract. The abnormal gene that causes CF was discovered in 1989. In 1990, researchers corrected CF—in lab dishes—by adding normal copies of the gene into cells. During the following 15 years, clinical trials showed that normal genes can be transferred to CF airways and temporarily improve lung function. The CF gene therapy approach has been to add new, therapeutic genes into cells.

A new technique was used on a mutation that causes one type of disease, severe combined immune deficiency. The mutation was corrected in lab-grown cells. The researchers designed a human-made protein to grab onto the mutated gene and slice through it in the vicinity of the mutation. The break in the gene then triggered the cell's own repair process. The cell copied a normal version of the gene and used the copy to replace a region of the cell's gene that included the mutation. About 7 percent of the lab-grown cells had their mutation fully corrected.

Analyze and Conclude

Respond to each question and statement.

1. **Explain** why CF scientists state that any CF gene technology treatment might need to be repeated periodically to be effective.

2. **Analyze** Have researchers found a cure for severe combined immune deficiency? Explain.

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