Genetic Screening and Gene Therapy

**KEY CONCEPT** Genetics provides a basis for new medical treatments.

**MAIN IDEAS**
- Genetic screening can detect genetic disorders.
- Gene therapy is the replacement of faulty genes.

**Connect to Your World**
Anyone could be a carrier of a genetic disorder. Genetic screening is used to help people figure out whether they are at risk for passing on that disorder. If they are at risk, what do they do? Do they not have children? Do they have children and hope that a child does not get the disorder? What would you do?

**MAIN IDEA**

**Genetic screening can detect genetic disorders.**

Every one of us carries alleles that produce defective proteins. Usually, these genes do not affect us in a significant way because we have other alleles that make up for the deficiency. But about 10 percent of people will find themselves dealing with an illness related to their genes at some point in their lives.

**Genetic screening** is the process of testing DNA to determine a person's risk of having or passing on a genetic disorder. Recall that genetic screening often involves both pedigree analysis and DNA tests. Because our knowledge of the human genome is still limited, it is not yet possible to test for every possible defect. Often, genetic screening is used to look for specific genes or proteins that indicate a particular disorder. Some tests can detect genes that are related to an increased risk of developing a disease, such as a gene called BRCA1 that has been linked to breast cancer. There are also tests for about 900 genetic disorders, including cystic fibrosis and Duchenne's muscular dystrophy (DMD). In DMD, it is quite easy to see differences in DNA tests between people with and without the disorder, as shown in Figure 6.1.

Genetic screening can help save lives. It can also lead to some difficult choices. Suppose a person has a family history of cancer and is tested for a gene that may lead to an increased risk of cancer. Is that information helpful or harmful? If a person has a chance of being a carrier of a genetic disorder, should screening be required? As genetic screening becomes more common, more questions like these will need to be answered.

**Infer** Why might genetic screening raise ethical concerns about privacy?
Gene therapy is the replacement of faulty genes.

A defective part in a car or in a computer can be easily replaced. If someone has a faulty gene that causes a disorder, is it possible to replace the gene? The goal of gene therapy is to do exactly that. Gene therapy is the replacement of a defective or missing gene, or the addition of a new gene, into a person’s genome to treat a disease.

For any type of gene therapy to work, researchers such as Dr. Betty Pace, shown in Figure 6.2, must first get the new gene into the correct cells of a patient’s body. Once in the body, the gene has to become a part of the cells’ DNA. One method of gene therapy that scientists have tried is to take a sample of bone marrow stem cells and “infect” them with a virus that has been genetically engineered with the new gene. Then the stem cells are put back into the patient’s bone marrow. Because they are stem cells, they divide and make more blood cells with the gene.

The first successful trial of gene therapy took place in 1990. The treatment was used on two children with a genetic autoimmune disorder, and the children are now adults leading normal lives. However, much of gene therapy is still experimental. For example, researchers are studying several different methods to treat cancer with gene therapy. One experimental approach involves inserting a gene that stimulates a person’s immune system to attack cancer cells. Another method is to insert “suicide” genes into cancer cells. These genes activate a drug inside those cells so that only the cancer cells are killed.

Gene therapy has many technical challenges. First, the correct gene has to be added to the correct cells. And even after researchers have figured out how to transfer the desired gene, the gene’s expression has to be regulated so that it does not make too much or too little protein. Scientists must also determine if the new gene will affect other genes. The many trials have produced few long-lasting positive results. But because of its great potential, research on gene therapy continues.

Synthesize How does gene therapy rely on genetic screening?