# 14.3 Studying the Human Genome

## Lesson Objectives

☐ Summarize the methods of DNA analysis.

☐ State the goals of the Human Genome Project and explain what we have learned so far.

## Lesson Summary

**Manipulating DNA** Since the 1970s, techniques have been developed that allow scientists to cut, separate, and replicate DNA base-by-base. Using these tools, scientists can read the base sequences in DNA from any cell.

► **Restriction enzymes** cut DNA into smaller pieces, called restriction fragments, which are several hundred bases in length. Each restriction enzyme cuts DNA at a different sequence of bases.

► **Gel electrophoresis** separates different-sized DNA fragments by placing them at one end of a porous gel, then applying an electrical voltage. The electrical charge moves the DNA.

► Using dye-labeled nucleotides, scientists can stop replication at any point along a single DNA strand. The fragments can then be separated by size using gel electrophoresis and "read," base-by-base.

**The Human Genome Project** was a 13-year international effort to sequence all 3 billion base pairs in human DNA and identify all human genes. The project was completed in 2003.

► The researchers identified markers in widely separated strands of DNA.

► They used "shotgun sequencing," which uses a computer to match DNA base sequences.

► To identify genes, they found promoters, exons, and other sites on the DNA molecule.

► To locate and identify as many haplotypes (collections of linked single-base differences) in the human population as possible, the International HapMap Project began in 2002.

► The Human Genome Project identified genes associated with many diseases and disorders. From the project came the new science of **bioinformatics**, the creation and use of databases and other computing tools to manage data. Bioinformatics launched **genomics**, the study of whole genomes.

► The human genome project pinpointed genes and associated particular sequences in those genes with numerous diseases and disorders. It also found that the DNA of all humans matches base-for-base at most sites, but can vary at 3 million sites.

► The 1000 Genomes Project, launched in 2008, will catalogue the variation among 1000 people.

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**RJ Alexander**

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