

This is Part 1 of a two-part report published in 1997 to reflect research and progress in the U.S. Department of Energy (DOE) Human Genome Program from 1994 through 1996, with specified updates made just before publication. Part 1 is the program overview and report on progress, and Part 2 consists of 1996 research abstracts.

Print copies of Parts 1 and 2 and subsequent reports on DOE genome research are available upon request from the Human Genome Management Information System (HGMIS); Oak Ridge National Laboratory; 1060 Commerce Park; Oak Ridge, TN 37830 (423/576-6669, Fax: /574-9888, bkq@ornl.gov).

Electronic versions are accessible via the DOE and HGMIS Web sites below.

- http://www.er.doe.gov/production/ober/hug_top.html
- <http://www.ornl.gov/hgmis/research.html>

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COVER

Detailed chromosome descriptions, together with other biological resources, software, and instrumentation generated in the first 7 years of the DOE Human Genome Program (HGP), are enabling researchers to begin focusing on their most challenging goal: Determining the sequence of DNA subunits (the bases A, T, C, and G) found in the 24 different human chromosomes. Differences in DNA sequence underlie much of life's diversity.

The cover depicts the progress of human genome research, beginning with a microscopic view of a duplicated chromosome (top). Genome researchers begin with a very small chromosomal fragment (asterisk), using enzymes to cut it into the smaller pieces (red bars) required for DNA sequencing. Automated technology determines the DNA sequence of all or part of each fragment (graph with color-coded peaks).

Another HGP goal is to identify the estimated 70,000 to 100,000 genes, which account for only about 5% of human DNA. Computer analysis of DNA sequences is one way investigators identify gene features in DNA sequences (solid line with tick marks). In a living cell, individual gene segments from DNA molecules are assembled into short-lived intermediary molecules (short red line), and the information is translated by the cell's machinery into three-dimensional proteins (black globular structure at bottom). All organisms are made up largely of proteins that provide the structural components and specialized enzymes required by cells and tissues.

Public resources and technologies arising from the HGP and other genome efforts worldwide are laying the foundation for future explorations into the functions of each protein encoded by the genes. This research, which also will investigate how proteins work together in systems and pathways and react to external cues, will extend far into the future.