

# Five-Year Research Goals of the U.S. Human Genome Project

October 1, 1993, to September 30, 1998 (FY 1994 through FY 1998)\*

*Major events in the U.S. Human Genome Project, including progress made toward these goals, are charted in a timeline on pp. ii-iii.*

## Genetic Mapping

- Complete the 2- to 5-cM map by 1995.
- Develop technology for rapid genotyping.
- Develop markers that are easier to use.
- Develop new mapping technologies.

## Physical Mapping

- Complete a sequence tagged site (STS) map of the human genome at a resolution of 100 kb.

## DNA Sequencing

- Develop efficient approaches to sequencing one- to several-megabase regions of DNA of high biological interest.
- Develop technology for high-throughput sequencing, focusing on systems integration of all steps from template preparation to data analysis.
- Build up a sequencing capacity to allow sequencing at a collective rate of 50 Mb per year by the end of the period. This rate should result in an aggregate of 80 Mb of DNA sequence completed by the end of FY 1998.

## Gene Identification

- Develop efficient methods for identifying genes and for placement of known genes on physical maps or sequenced DNA.

## Technology Development

- Substantially expand support of innovative technological developments as well as improvements in current technology for DNA sequencing and for meeting the needs of the Human Genome Project as a whole.

## Model Organisms

- Finish an STS map of the mouse genome at a 300-kb resolution.
- Finish the sequence of the *Escherichia coli* and *Saccharomyces cerevisiae* genomes by 1998 or earlier.
- Continue sequencing *Caenorhabditis elegans* and *Drosophila melanogaster* genomes with the aim of bringing *C. elegans* to near completion by 1998.
- Sequence selected segments of mouse DNA side by side with corresponding human DNA in areas of high biological interest.

## Informatics

- Continue to create, develop, and operate databases and database tools for easy access to data, including effective tools and standards for data exchange and links among databases.
- Consolidate, distribute, and continue to develop effective software for large-scale genome projects.
- Continue to develop tools for comparing and interpreting genome information.

## Ethical, Legal, and Social Implications

- Continue to identify and define issues and develop policy options to address them.
- Develop and disseminate policy options regarding genetic testing services with potential widespread use.
- Foster greater acceptance of human genetic variation.
- Enhance and expand public and professional education that is sensitive to sociocultural and psychological issues.

## Training

- Continue to encourage training of scientists in interdisciplinary sciences related to genome research.

## Technology Transfer

- Encourage and enhance technology transfer both into and out of centers of genome research.

## Outreach

- Cooperate with those who would establish distribution centers for genome materials.
- Share all information and materials within 6 months of their development. This should be accomplished by submission of information to public databases or repositories, or both, where appropriate.

\*Original 1990 goals were revised in 1993 due to rapid progress. A second revision was being developed at press time.