DOE/ER-0713 (Part 2)

HUMAN GENOME PROGRAM REPORT

Part 2, 1996 Research Abstracts

Date Published: November 1997

Prepared for the
U.S. Department of Energy
Office of Energy Research
Office of Biological and Environmental Research
Germantown, MD 20874-1290

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managed by
Lockheed Martin Energy Research Corporation
for the
U.S. Department of Energy
Under Contract DE-AC05-96OR22464

Preface

More than a decade ago, the Office of Health and Environmental Research (OHER) of the U.S. Department of Energy (DOE) struck a bold course in launching its Human Genome Initiative, convinced that its mission would be well served by a comprehensive picture of the human genome. Organizers recognized that the information the project would generate—both technological and genetic—would contribute not only to a new understanding of human biology and the effects of energy technologies but also to a host of practical applications in the biotechnology industry and in the arenas of agriculture and environmental protection.

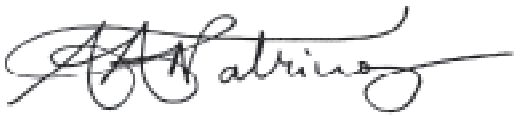
Today, the project's value appears beyond doubt as worldwide participation contributes toward the goals of determining the human genome's complete sequence by 2005 and elucidating the genome structure of several model organisms as well. This report summarizes the content and progress of the DOE Human Genome Program (HGP). Descriptive research summaries, along with information on program history, goals, management, and current research highlights, provide a comprehensive view of the DOE program.

Last year marked an early transition to the third and final phase of the U.S. Human Genome Project as pilot programs to refine large-scale sequencing strategies and resources were funded by DOE and the National Institutes of Health, the two sponsoring U.S. agencies. The human genome centers at Lawrence Berkeley National Laboratory, Lawrence Livermore National Laboratory, and Los Alamos National Laboratory had been serving as the core of DOE multidisciplinary HGP research, which requires extensive contributions from biologists, engineers, chemists, computer scientists, and mathematicians. These team efforts were complemented by those at other DOE-supported laboratories and about 60 universities, research organizations, companies, and foreign institutions. Now, to focus DOE's considerable resources on meeting the challenges of large-scale sequencing, the sequencing efforts of the three genome centers have been integrated into the Joint Genome Institute. The institute will continue to bring together research from other DOE-supported laboratories. Work in other critical areas continues to develop the resources and technologies needed for production sequencing; computational approaches to data management and interpretation (called informatics); and an exploration of the important ethical, legal, and social issues arising from use of the generated data, particularly regarding the privacy and confidentiality of genetic information.

Insights, technologies, and infrastructure emerging from the Human Genome Project are catalyzing a biological revolution. Health-related biotechnology is already a success story—and is still far from reaching its potential. Other applications are likely to beget similar successes in coming decades; among these are several of great importance to DOE. We can look to improvements in waste control and an exciting era of environmental bioremediation, we will see new approaches to improving energy efficiency, and we can hope for dramatic strides toward meeting the fuel demands of the future.

In 1997 OHER, renamed the Office of Biological and Environmental Research (OBER), is celebrating 50 years of conducting research to exploit the boundless promise of energy technologies while exploring their consequences to the public's health and the environment. The DOE Human Genome Program and a related spin-off project, the Microbial Genome Program, are major components of the Biological and Environmental Research Program of OBER.

DOE OBER is proud of its contributions to the Human Genome Project and welcomes general or scientific inquiries concerning its genome programs. Announcements soliciting research applications appear in *Federal Register*, *Science, Human Genome News,* and other publications. The deadline for formal applications is generally midsummer for awards to be made the next year, and submission of preproposals in areas of potential interest is strongly encouraged. Further information may be obtained by contacting the program office or visiting the DOE home page (301/903-6488, Fax: -8521, *genome@oer.doe.gov*, URL: *http://www.er.doe.gov/production/ober/hug_top.html*).



Office of Biological and Environmental Research
U.S. Department of Energy
November 3, 1997

Foreword

The research abstracts in this section were funded in FY 1996 by the DOE Office of Health and Environmental Research, which was renamed Office of Biological and Environmental Research in 1997.

These unedited abstracts were contributed by DOE Human Genome Program grantees and contractors. Names of principal investigators are in bold print. Submitted in 1996, contact information is for the first person named unless another investigator is designated as contact person. Principal investigators of research projects described by abstracts in this section are listed under their respective subject categories, and an index of all investigators named in the abstracts is given at the end of this report.

Part 1 of this report contains narratives that represent DOE Human Genome Program research in large, multidisciplinary projects. As a convenience to the reader, these narratives are reprinted (without graphics) as an appendix to this volume, Part 2. The projects represent work at the Joint Genome Institute (p. 72), Lawrence Livermore National Laboratory Human Genome Center (p. 73), Los Alamos National Laboratory Center for Human Genome Studies (p. 77), Lawrence Berkeley National Laboratory Human Genome Center (p. 81), University of Washington Genome Center (p. 85), Genome Database (p. 87), and National Center for Genome Resources (p. 91). Only the contact persons for these organizations are listed in the Index to Principal and Coinvestigators. More information on research carried out in these projects can be found on their listed Web sites.

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Project Categories and Principal Investigators

*Russian scientists designated by an asterisk received small emergency grants following December 1992 site reviews by David Galas (formerly DOE Office of Health and Environmental Research, which was renamed Office of Biological and Environmental Research in 1997), Raymond Gesteland (University of Utah), and Elbert Branscomb (Lawrence Livermore National Laboratory).