Technologies for Detecting Heritable Mutations in Human Beings

September 1986

NTIS order #PB87-140158
Foreword

Ensuring the health of future generations of children is of obvious importance to American society. Heritable mutations, permanent changes in the genetic material that can be passed on to succeeding generations, are the cause of a large but currently unquantifiable share of embryonic and fetal loss, disease, disability, and early death in the United States today. The methods now available to study heritable mutations, however, offer relatively little information about the kinds of mutations that can occur, their frequency, or their causes. Recent advances in molecular genetics have opened the door to new and innovative technologies that may offer a great deal more information about DNA. It may soon be possible to characterize mutations precisely, to measure their frequency, and perhaps also to associate particular mutations with exposures to specific mutagenic influences. While some of the new technologies are still on the drawing board, they are developing quickly and several of them may become available for wide-scale use in the next 5 to 10 years.

The Senate Committee on Veterans’ Affairs, the House Committee on Science and Technology, and the House Committee on Energy and Commerce requested that OTA assess the available information about current and proposed means for detecting heritable mutations and on the likelihood and potential impact of such technological advances. These committees have wrestled with the problems of determining whether past exposures to potential mutagens have affected the health of Americans, and of framing reasonable public health laws, given current knowledge and technologies. This report summarizes OTA’s findings as they relate to these issues.

An advisory panel, chaired by Arno G. Motulsky, provided guidance and assistance during the assessment. The OTA Health Program Advisory Committee, OTA staff, and scientific and policy experts from the private sector, academia, and the Federal Government provided information during the assessment and reviewed drafts of the report. We thank all who assisted us. As with all OTA reports, the content of the assessment is the sole responsibility of OTA and does not necessarily constitute the consensus or endorsement of the advisory panel or the Technology Assessment Board. Key OTA staff involved in the assessment were Michael Gough, Julie Ostrowsky, and Hellen Gelband.
Advisory Panel for Technologies for Detecting Heritable Mutations in Human Beings

Arno G. Motulsky, Panel Chair
Center for Inherited Diseases
University of Washington School of Medicine

Richard J. Albertini
Department of Medicine
University of Vermont
College of Medicine

Michael S. Baram
School of Medicine and Public Health
Boston University

Charles R. Cantor
Department of Human Genetics and Development
Columbia University College of Physicians and Surgeons

Dale Hattis
Center for Policy Alternatives
Massachusetts Institute of Technology

Ernest B. Hook
Bureau of Maternal and Child Health
New York State Department of Health

Alfred G. Knudson, Jr.
Institute for Cancer Research
Fox Chase Cancer Center

Nan M. Laird
Department of Biological Statistics
Harvard University
School of Public Health

Mortimer L. Mendelssohn
Lawrence Livermore National Laboratory

Jeffrey H. Miller
Department of Biology
University of California at Los Angeles

James V. Neel
Department of Human Genetics
University of Michigan School of Medicine

Norton Nelson
Department of Environmental Medicine
New York University School of Medicine

Mark L. Pearson
E.I. du Pent de Nemours & Co.

Richard K. Riegelman
Department of Medicine
George Washington University School of Medicine

Liane B. Russell
Oak Ridge National Laboratory

Richard B. Setlow
Brookhaven National Laboratory

William J. Schull
The University of Texas Health Science Center

William G. Thilly
Department of Nutrition and Food Science
Massachusetts Institute of Technology

Richard M. Myers, Special Consultant
University of California at San Francisco

NOTE: OTA appreciates and is grateful for the valuable assistance and thoughtful critiques provided by the advisory panel members. The panel does not, however, necessarily approve, disapprove, or endorse this report. OTA assumes full responsibility for the report and the accuracy of its contents.
OTA Project Staff—Technologies for Detecting Heritable Mutations in Human Beings

Roger C. Herdman, Assistant Director, OTA
Health and Life Sciences Division

Clyde J. Behney, Health Program Manager

Michael Gough, Project Director
Julia T. Ostrowsky, Analyst
Hellen Gelband, Analyst

Other Contributing Staff
Cheryl M. Corsaro, Analyst
Virginia Cwalina, Administrative Assistant
Carol Ann Guntow, Secretary/Word Processor Specialist
Diann G. Hohenthaner, Word Processor/P. C. Specialist
Eric Passaglia, Senior Distribution Specialist

Contractors
Elbert Branscomb, Lawrence Livermore National Laboratory
Neal Cariello, Massachusetts Institute of Technology
Leonard Lerman, Genetics Institute
Harvey Mohrenweiser, University of Michigan Medical School
Richard M. Myers, University of California at San Francisco
Janice A. Nicklas, University of Vermont School of Medicine
Maynard Olson, Washington University Medical School
Cassandra Smith, Columbia University College of Physicians and Surgeons
L.H.T. Van der Ploeg, Columbia University College of Physicians and Surgeons
Diane K. Wagener, Washington, DC

"On detail from the National Institutes of Health, April-June 1985."