



Program Results Topic Summary: Genetics

Program Results Special Report

Advances in the study of human genes offer promise for alleviating disease and improving human quality of life. Critics worry, however, that these advances could undermine the very meaning of human society.

Since 1991, the Robert Wood Johnson Foundation (RWJF) has funded 32 grants and contracts totaling about \$7 million for projects related to genetics, genetic technologies and genetic services. See the [Appendix](#) for a complete list. Projects related to genetics supported by the Foundation and reported on in Program Results covered the following topics:

- **Ethical and Legal Issues in Genetics, Genetic Technology, Genetic Testing and Genetic Services**—One project aimed to bring individuals and groups together to discuss and research the ethical and legal issues raised by current advances in genetics.
- **Consumer Attitudes and Awareness**—Six projects supported by RWJF have assessed and/or influenced the public's attitudes toward and awareness of genetic technologies.
- **Health Care Professionals: Attitudes, Training and Practice**—Eight projects focused on the effect that advances in human genetics have made to the field of health care, especially by assessing the attitudes of health professionals, and addressing changes to their training and practice to accommodate these advances.

This report provides a road map of RWJF's support for work focused on genetics.

OVERVIEW OF RWJF INTEREST IN GENETICS

Advances in the study of human genes offer promise for alleviating disease and improving human quality of life. Critics worry, however, that these advances could undermine the very meaning of human society. Genetics, the branch of biology that deals with heredity, especially the mechanisms of hereditary transmission and the variation of inherited characteristics among similar or related organisms, is at the root of many of the most heated ethical debates in American society—genetic screening, cloning, gene therapy, genetic privacy and stem cell research.

Since 1991, RWJF has funded 32 grants and contracts totaling \$6.6 million for projects related to genetics, genetic technologies and genetic services. In 2002, RWJF pulled back from making grants focused on genetics as it developed a grantmaking strategy to focus on other defined strategic objectives. Programmaking in this area could become active again in the future, however.

This report provides a road map of RWJF's support for work focused on genetics, providing links to Program Results (where available) on 15 of the projects. It does not cover other RWJF grants on which reports have not been written. For a full list of RWJF grants, see the [Appendix](#).

Supported projects cover a range of topics within the field of genetics and approach the topic from varying points of view. Because the projects were not developed in concert with one another, they vary in the perspective taken toward the ethical issues raised by genetic technologies. Some approach these technologies with circumspection, either by asking the opinion of consumers or professionals toward these technologies, or by bringing together individuals to debate their costs and benefits. Other projects seem to take scientific advances more or less for granted, looking instead at what health professionals need to do to prepare themselves and their colleagues for what seem to be inevitable advances in the field.

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- **Consumer Attitudes and Awareness**—Six projects supported by RWJF have assessed and/or influenced the public's attitudes toward and awareness of genetic technologies.
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ETHICAL AND LEGAL ISSUES IN GENETICS, GENETIC TECHNOLOGY, GENETIC TESTING AND GENETIC SERVICES

Privacy issues are at the forefront of concerns over genetic advances, according to information gleaned by the **Council for Responsible Genetics**. In 2003, (see [Program Results](#) on ID# 047719) the council produced a series of six reports on legislation and court precedents relating to applications of human genetics. In *Genetic Privacy: A Report*

on *Legislation and Legal Precedents in the United States*, the Council for Responsible Genetics concluded:

- Despite more than a decade of legislative activity, the United States continues to lack a broad minimum standard of protection for genetic privacy. As a result, individuals must rely on a mixture of state and federal statutes that regulate access to genetic and other health information by specific entities—such as hospitals, employers and forensic laboratories—and for specific purposes—such as insurance underwriting.
- Resistance from certain stakeholders within the health care system who rely on access to genetic information continues to inhibit privacy reforms. Thus, the genetic records of patients, research subjects and others continue to undergo routine exchange and disclosure with minimal privacy safeguards.
- The fear of discrimination remains a major roadblock both to public participation in genetics research and to the adoption of genomic medicine by physicians and consumers. Surveys and clinical research indicate that the fear of third-party access to genetic records deters many people from undergoing genetic testing and participating in medical research. Without better protection of private information, these fears will likely continue and inhibit the full realization of potential scientific advances.

Quick Facts

About 65 percent of both African Americans and Whites would want a genetic test for a treatable disease.

58 percent of African Americans and 61 percent of Hispanics would want a test for an untreatable disease compared to 43 percent of Whites.

The council also created the Genetics and the Law [website](#), which analyzes and explains the legal and ethical implications of state and federal legislation pertaining to genetics and includes a database of case law and legislation. Topics covered include newborn screening, criminal justice, privacy, employment and health insurance.

RWJF also supported a regional conference in Arizona covering legal and ethical issues related to human genetics.¹

CONSUMER ATTITUDES AND AWARENESS

Asked their opinion of genetic technologies, Americans reveal conflicting attitudes—on the one hand most Americans show an interest in genetic testing, but many are concerned about privacy issues, according to one survey supported by RWJF. To address the lack of understanding of these technologies, Fred Friendly Seminars produced a widely viewed series on the subject for public television.

¹ In 1999, RWJF provided partial support for a regional conference of state health officials, researchers, genetic counselors, Native American health officials and consumers on ethical and pragmatic issues in new advances in genetics. (See [Program Results](#) on ID# 035763.)

Consumer Attitudes

The University of Michigan (see [Program Results](#) on ID#s 030214 and 035311), conducted a national survey in 2000 of Americans' knowledge, attitudes, values and behavior regarding genetic testing. The survey found that race plays a factor in Americans' comfort with genetic technology:

- About 65 percent of both African Americans and Whites would want a genetic test for a *treatable* disease, but larger percentages of African Americans (58%) and Hispanics (61%) would want a test for an *untreatable* disease than would Whites (43%).
- Despite their interest in genetic testing, African Americans' concerns about privacy and lack of confidence in institutions and trust in their doctors may discourage them from seeking such tests. African Americans are more concerned about privacy than Whites; 44 percent strongly disagree that their privacy is well-protected versus 30 percent of Whites. African Americans also express significantly less confidence in institutions than Caucasians and less trust in their doctor to keep information private (51% versus 59% of Whites).

As a result of the survey, the project staff convened a conference on genetics and health disparities.

Discussions at the conference, according to Eleanor Singer, Ph.D., project director, "represented most of the large spectrum of opinion on this contentious issue, ranging from the view that advances in genetics will make health disparities among race-ethnic groups obsolete to the position that the emphasis on genetics serve to rigidify racial and ethnic differences while absolving society of the need for addressing the social and environmental conditions largely responsible for health disparities."²

Public Awareness

In January 2003 approximately 2.3 million viewers tuned in to a three-part television series on genetics produced by **Fred Friendly Seminars** in association with Thirteen/WNET (see [Program Results](#) on ID# 040923). The series, now available on video, was intended to increase public awareness of advances in

Quick Facts

Workable, comprehensive definitions of the terms "genetic information," "genetic testing" and "genetic screening" should be developed, uniformly adopted and widely disseminated.

Since genetic issues involve not only individual patients, but also a patient's family members, research should be conducted into issues concerning consent and notification.

More molecular, environmental, behavioral and epidemiological research is needed to determine the sensitivity and specificity of genetic markers and to measure risk for disease.

² "Genetic Research Stirs Discussion at Two Conferences," The University Record Online [University of Michigan], April 5, 2004, available [online](#).

genetic technologies and issues such as genetic privacy, prenatal screening and genetic engineering. A [website](#), Our Genes/Our Choices, accompanied the series, and provides additional resources for viewers.

RWJF provided partial funding to the **March of Dimes Birth Defects Foundation** for the Genetics Awareness Coalition (see [Program Results](#) on ID# 035750), which met in 1998/1999 to explore potential collaboration among federal agencies and professional groups to develop a campaign to increase public awareness of advances in genetics and the impact of these advances on health and health care. The organizational meetings occurred, but RWJF did not ultimately fund the public awareness campaign.

The 1997 **Sun Valley Forum on National Health** (see [Program Results](#) on ID# 030194) reviewed the implications of advances in genetics for the improvement of health and health care. Participants jointly issued a set of observations and recommendations entitled *The New Genetics: Challenges and Consequences of Therapeutic Developments*.³ Recommendations included:

- Greater interaction between the general public and the scientific community needs to occur to increase public understanding of scientific advances in general, and genetics in particular, as well as of related ethical, legal and social issues.
- Workable, comprehensive definitions of the terms "genetic information," "genetic testing" and "genetic screening" should be developed, uniformly adopted and widely disseminated.
- The general public should receive adequate, comprehensible information about genetic advances. This should occur at all levels, including beginning in elementary school, patient education and targeted education of media, judges, elected officials, businesspersons and other community leaders.
- Societal commitment to, resources devoted to, and activities aimed at disease prevention should be dramatically enhanced.
- Since genetic issues involve not only individual patients, but also a patient's family members, research should be conducted into issues concerning consent and notification.

Quick Facts

Insufficient data on clinical validity and clinical utility were foremost among the aspects of genetic testing that caused physicians to hesitate in using predictive genetic testing.

Concerns about insurance discrimination in the absence of treatments for patients with positive test results also discouraged use.

High cost and perceptions of inconvenience associated with obtaining genetic testing were also barriers to use of genetic testing.

³ Sun Valley Forum on National Health. "The New Genetics: Challenges and Consequences of Therapeutic Developments." A supplement to the *Western Journal of Medicine*, 168(6), 1998.

- More molecular, environmental, behavioral and epidemiological research is needed to determine the sensitivity and specificity of genetic markers and to measure risk for disease.

RWJF also funded two projects meant to increase undergraduates' awareness of genetics issues:

- **Student PUGWASH USA** (see [Program Results](#) on ID#s 020447 and 033226), promoted increased awareness on college campuses of social and ethical aspects of genetics and other technological issues.
- **Stanford University** (see [Program Results](#) on ID# 037556) produced a CD-ROM to teach genomics and genetics to undergraduate science students. Copies are available from [Twisted Ladder Media](#). A version is available for physicians who wish to expand their genetics knowledge; continuing medical education credit is offered.

HEALTH CARE PROFESSIONALS: ATTITUDES, TRAINING AND PRACTICE

In 1998 and 2001/2002, RWJF supported studies of health care professionals and their understanding of and attitudes toward genetics and genetic testing. According to these studies, health professionals have been under-informed about advances in genetics and their application to medical practice. Three projects have addressed improving health professionals' knowledge of genomics and genetic technology, including support for a continuing education program offered online, the publication of a book on the topic and a coalition of health professional education organizations.

Physician and Health Care Leader Attitudes

In 2001 and 2002, the Institute for the Future (see [Program Results](#) on ID#s 040794 and 043843) assessed the knowledge and attitudes of health care leaders and providers about genomic medicine.

- This qualitative study found that nearly all the health care leaders who participated exhibited only rudimentary general knowledge about the human genome and genetic technologies.⁴

The institute also prepared a proposal requesting support for a genetic education initiative aimed at primary care physicians, but RWJF declined to fund it.

⁴ Billings PR, Carlson R, Carlson J, Wilson CO, Cain M, Shorett P and Everett W. "Ready for Genomic Medicine? Perspectives of Healthcare Decision-Makers." *Archives of Internal Medicine*, September 12, 2005.

In 1998, **Johns Hopkins University** (see [Program Results](#) on ID# 033717) interviewed physicians to ascertain the barriers and incentives of the diffusion of genetic services.

Key findings were:

- Insufficient data on clinical validity and clinical utility were foremost among the aspects of genetic testing that caused physicians to hesitate in using predictive genetic testing.
- Concerns about insurance discrimination in the absence of treatments for patients with positive test results also discouraged use.
- High cost and perceptions of inconvenience associated with obtaining genetic testing were also barriers to use of genetic testing.

The main reason physicians who adopted testing did so was interest on the part of the patient.

Health Professional Training and Practice

Cold Spring Harbor Laboratory (see [Program Results](#) on ID# 022602) convened two meetings of genetics researchers and primary care providers in 1994 and 1995 to examine how advances in genetic knowledge affect primary health care.

The meetings resulted in a book, *Toward the 21st Century: Incorporating Genetics Into Primary Health Care*⁵, arguing that genetics will be an increasingly important part of medical practice and that genetics caregivers will be critical. The Foundation distributed 1,600 copies of the book to medical schools, professional organizations, public policy-makers and other leaders.

The project led to three additional grants—one to for a survey of managed care providers, one to start the National Coalition for Health Professional Education in Genetics and one to the Alliance of Genetic Support Groups for research on consumer issues in managed care.

In 1996, **Neil Holtzman, M.D., M.P.H., at Johns Hopkins University** (see [Program Results](#) on ID# 028511), surveyed health maintenance organization (HMO) medical directors to determine the extent of requests and coverage for predictive genetic testing.

⁵ Touchette N, Holtzman NA, Davis JG and Feetham S, with an afterword by Collins FS. *Toward the 21st Century: Incorporating Genetics into Primary Health Care*. Cold Spring Harbor, NY: Cold Spring Harbor Laboratory Press, 1997

Quick Facts

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RWJF awarded a planning grant (ID# 030764) to the **National Coalition for Health Professional Education in Genetics**, which is sponsored jointly by the American Medical Association and the American Nurses Association. The coalition is an interdisciplinary group that brings together 140 organizations to promote health professional education in the area of human genetics.

Funding from RWJF (see [Program Results](#) on ID#s 030764, 037749 and 043547) allowed the coalition to create educational tools and resources to help integrate genetics content into the knowledge base of health professional and students in the health professions. During the funding period, the coalition:

- Became a reliable locus for inquiries and requests for assistance in genetics from the health care field and the community at large.
- Developed the *Core Competencies in Genetics Essential for All Health-Care Professionals*⁷, which defines the knowledge, skills and attitudes that health care professionals should possess to integrate genetics and genomics into mainstream healthcare.
- Produced pre-service and continuing educational resources for health care professionals.

The Alliance of Genetic Support Groups is a national organization representing individuals with genetic disorders and their families. In 1997, the alliance (see [Program Results](#) on ID# 030944) organized a series of meetings to open a dialogue between health care organizations, consumer groups and managed care organizations in order to improve genetic services delivery. The meetings laid the groundwork for improved working relationships between the consumer genetics community and managed care providers.

Two additional projects funded by RWJF created websites to improve the integration of genetic

Quick Facts

The University of Virginia (see Program Results on ID#s 035277 and 040685) developed and pilot tested a Web-based tool for collecting family health history that can enhance communication between primary care providers and individuals about their risks for common diseases with genetic components.

In 2003, the March of Dimes Birth Defects Foundation (see Program Results on ID# 033331) launched Genetics & Your Practice, a website providing practical information and tools for integrating genetics into medical practice.

⁶ Myers MF, Doksum T and Holtzman NA. "Genetic Services for Common Complex Disorders: Surveys of Health Maintenance Organizations and Academic Genetic Centers." *Genetics in Medicine*, 1(6): 272–285, 1999.

⁷ Core Competencies Working Group. "Recommendations of Core Competencies in Genetics for All Health Professionals." *Genetics in Medicine*, 3(2): 155–159, 2001.

services into primary care medical practice.

- **The University of Virginia** (see [Program Results](#) on ID#s 035277 and 040685) developed and pilot tested a [Web-based tool](#) for collecting family health history that can enhance communication between primary care providers and individuals about their risks for common diseases with genetic components.
- In 2003, the **March of Dimes Birth Defects Foundation** (see [Program Results](#) on ID# 033331) launched [Genetics & Your Practice](#), a Web site providing practical information and tools for integrating genetics into medical practice and offers the option of earning continuing medical education credits for completion of the course. Three educational modules outline a practical process for integrating genetics into professional practice:
 - *Genetic testing and screening.* This module provides a step-by-step guide to genetic testing and tools that can help health professionals minimize disease-associated morbidity and mortality.
 - *Family health and social history.* This module provides a time-saving method for taking a family history and a sample family history questionnaire that users can download.
 - *Referral to genetic services.* This module gives practitioners referral information and tools to help them find professional genetic service providers in their local areas.

More than 2,000 health care professionals visited the site during its first 21 months.

APPENDIX: GRANTS BY THE ROBERT WOOD JOHNSON FOUNDATION RELATED TO GENETICS: 1991 TO 2006

Program ID#	Dates	Programee	Program Title	Amount	Program Results (if available)
PC241	January 1991 to December 1991	University of Pennsylvania	Study of the Barriers That May Impede the Transfer of Genetics Technology	\$28,370	NA
020447	August 1992 to December 1993;	Student PUGWASH USA	Student Conferences on Implications of Advances in Medical Technology	\$25,150	Program Results on ID#s 020447 and 033226
022602	February 1994 to July 1995	Cold Spring Harbor Laboratory	Meetings on the Health Care Implications of the Human Genome Project	\$138,691	Program Results on ID# 022602

028511	January 1996 to December 1996	John Hopkins University	Project to Assess Medical Education and Practice Initiatives Related to Genetic Services	\$41,807	Program Results on ID# 028511
030194	December 1996 to November 1997	Sun Valley Forum on National Health	Support for a Conference on the Therapeutic Implications of Advances in Genetic and Molecular Medicine	\$37,500	Program Results on ID# 030194
030214	May 1997 to October 1998	University of Michigan	Developmental Work for a Study of Changes in Public Attitudes Toward Genetic Testing	\$30,664	Program Results on ID#s 030214 and 035311
030764	January 1997 to January 1998	American Nurses Foundation	Establishment of the National Coalition for Health Professional Education in Genetics	\$50,000	Program Results on ID#s 030764, 037749 and 043547
030944	January 1997 to July 1997	Alliance to Genetic Support Groups	Improving the Delivery of Genetic Services in Managed Care	\$34,729	Program Results on ID# 030944
033226	October 1997 to June 1998	Student PUGWASH USA	Student Conference on Science and Social Responsibility	\$50,000	Program Results on ID#s 020447 and 033226
033331	June 2000 to December 2004	March of Dimes Birth Defects Foundation	Expanding and Evaluating the Genetics & Your Practice Curriculum	\$746,363	Program Results on ID# 033331
033717	February 1998 to February 1999	Johns Hopkins University	Pilot Study to Examine Physicians' Attitudes Toward Adoption of New Genetic Technologies	\$38,732	Program Results on ID# 033717
033808	June 1998 to August 1998	Susan Jo Bumagin & Associates	Review and Assessment of a Genetics Education and Demonstration Project	\$20,747	NA
033960	February 1998 to August 1998	American Medical Association	Training Physicians in Advances in Genetics	\$12,893	NA
035277	November 1998 to September 2001	University of Virginia	Helping Primary Care Practitioners Assess Genetic Vulnerability to Common Chronic Diseases	\$1,572,602	Program Results on ID#s 035277 and 040685
035311	June 1999 to October 2004	University of Michigan	Genetic Technology and Health: Knowledge, Attitudes, Values and Behavior	\$474,651	Program Results on ID#s 030214 and 035311
035750	November 1998 to June 1999	March of Dimes Birth Defects Foundation	Support of Meetings on Genetics Awareness for Public Health Practice	\$14,680	Program Results on ID# 035750
037556	May 2000 to April 2004	Stanford University	Expanding the New Genetics: Interactive Courseware for Physicians	\$190,575	Program Results on ID# 037556

037749	December 1999 to April 2001	Foundation for the National Institutes of Health	National Coalition to Educate Health Professionals in Genetics	\$268,589	Program Results on ID#s 030764, 037749 and 043547
040685	February 2001 to May 2002	University of Virginia	Helping Primary Care Practitioners Assess Genetic Vulnerability to Common Chronic Diseases	\$528,854	Program Results on ID#s 035277 and 040685
040794	April 2001 to October 2001	Institute for the Future	Exploring Options to Improve Public and Private Decision-Making About Genetic Information	\$152,818	Program Results on ID# 047719
040923	July 2001 to December 2003	Fred Friendly Seminars	Fred Friendly Seminars Series on the Practical and Ethical Challenge of Advances in Genetic Technology	\$747,300	Program Results on ID# 040923
041614	April 2001 to May 2001	Mountain States Genetics Foundation	Symposium on Cancer Genetics Counseling Issues for Medical Professionals	\$10,000	NA
043547	September 2001 to April 2003	National Coalition to Educate Health Professionals in Genetics	National Coalition to Educate Health Professionals in Genetics	\$415,441	Program Results on ID#s 030764, 037749 and 043547
043843	March 2002 to February 2003	Institute for the Future	Exploring Options to Improve Public and Private Decision-Making About Genetic Information	\$111,666	Program Results on ID#s 043843 and 047719
047719	March 2003 to August 2004	Council for Responsible Genetics	Genetic Information and the Law: Establishing a Comprehensive Database of Essential Cases and Regulations for Public Health	\$129,891	Program Results on ID#s 043843 and 047719
047723	September 2003 to August 2005	Illinois Institute of Technology	Impact of Gene Patents on the Delivery of Health Care Services	\$275,948	NA
047742	March 2003	National Disease Research Interchange	Conference on the Genetics of Rare Disease	\$2,500	NA
048449	September 2003 to August 2004	Cornell University	Gene Therapy Research Experience for a Native American Medical School Candidate	\$264	NA
051150	January 2005 to December 2006	National Coalition for Health Professional Education in Genetics	Web-Based Education and National Forum on Race and Genetics for Health Professionals	\$300,000	NA
051426	July 2004 to	Das Deutsch Center	Educational Intervention	\$280,862	NA

	June 2008	for Special Needs Children	to Reduce the Incidence of Genetic Disease Among Amish families		
051536	September 2004 to June 2005	Columbia University	Potential Modification of the Effect of Alcohol on Breast Cancer Risk by Variation in Genes Involved in Alcohol Metabolism	\$99,171	NA
051538	September 2004 to August 2005	New York University	Gene Therapy Research Experience for a Native American Medical School Candidate	\$14,736	NA