

**National Human Genome Research Institute  
Five-Year Strategic Plan for Reducing Health Disparities**

**Submitted to  
The National Center for Minority Health and Health Disparities**

**For  
The NIH Comprehensive Strategic Plan and Budget  
To Reduce and Ultimately Eliminate Health Disparities**

## TABLE OF CONTENTS

<b>Table of Contents</b>	<b>2</b>
<b>NHGRI Mission Statement</b>	<b>6</b>
<b>Overview of NHGRI Strategy for Addressing Health Disparities</b>	<b>6</b>
<b>AREA OF EMPHASIS NO. 1: RESEARCH IN HEALTH DISPARITIES</b>	
<b>1A : GENOMIC RESEARCH</b>	<b>9</b>
<b>Rationale and Priority of Objective</b>	
<b>Objective 1A.1:</b>	
<b>Continue and Further Expand Research Collaborations between NHGRI and Howard University Africa America Diabetes Mellitus Study (AADM) and African American Hereditary Prostate Cancer Study Network (AAHPC)</b>	
Action Plan 1A.1	
Performance Measures 1A.1	
Outcome Measures 1A.1	
<b>Objective 1A.2:</b>	
<b>Develop and Conduct “The Barbados Prostate and Breast Cancer Study”</b>	
Action Plan 1A.2	
Performance Measures 1A.2	
Outcome Measures 1A.2	
<b>Objective 1A.3:</b>	
<b>Implement a New Program Called the Intramural Health Disparities Research Award</b>	
Action Plan 1A.3	
Performance Measures 1A.3	
Outcome Measures 1A.3	

**AREA OF EMPHASIS NO. 1: RESEARCH IN HEALTH DISPARITIES  
(continued)**

**1B : ETHICAL, LEGAL AND SOCIAL IMPLICATIONS  
(ELSI) RESEARCH**

**13**

**Rationale and Priority of Objective**

**Objective 1B.1:**

**Study the Ethical, Legal and Social Implications of Genetic Variation Research for Individuals and Diverse Racial and Ethnic Groups (FY1999-FY2002 and FY2002-FY2005)**

Action Plan 1B.1

Performance Measures 1B.1

Outcome Measures 1B.1

**Objective 1B.2:**

**Study Hereditary Hemochromatosis and Iron Overload Disease in Diverse Populations**

Action Plan 1B.2

Performance Measures 1B.2

Outcome Measures 1B.2

**Objective 1B.3.**

**Implement the ELSI/Native American Research Centers for Health (NARCH) Initiative**

Action Plan 1B.3

Performance Measures 1B.3

Outcome Measures 1B.3

**Objective 1B.4:**

**Support ELSI Research addressing Minority Participation in Genetic Research**

Action Plan 1B.4

Performance Measures 1B.4

Outcome Measures 1B.4

**Rationale and Priority of Objectives**

**Objective 2.1:**

**Expand Minority Training Opportunities at Centers of Excellence in Genomic Science (CEGS) and Genome Production Centers**

Action Plan 2.1

Performance Measures 2.1

Outcome Measures 2.1

**Objective 2.2:**

**Continue to Support and Develop the “Genomics Short Course for Faculty at Minority Serving Institutions”**

Action Plan 2.2

Performance Measures 2.2

Outcome Measures 2.2

**Objective 2.3:**

**Through the Minority Supplement Program, Expand Opportunities for Minority Students and Faculty to Participate in Genomic and ELSI Research by Supporting Relevant Research Experiences and Attendance at Workshops and Conferences**

Action Plan 2.3

Performance Measures 2.3

Outcome Measures 2.3

**Objective 2.4:**

**Increase the Number of Minorities Participating in the T32 Training Grant Program**

Action Plan 2.4

Performance Measures 2.4

Outcome Measures 2.4

**Objective 2.5:**

**Increase the Number of Summer Internship Opportunities at NHGRI for Minority Students**

Action Plan 2.5

Performance Measures 2.5

Outcome Measures 2.5

**Objective 2.6:**

**Increase the number of Summer Internship Opportunities for Minority students at the National Human Genome Center at Howard University (NHGC)**

Action Plan 2.6

Performance Measures 2.6

Outcome Measures 2.6

**Objective 2.7:**  
**Increase the Number of Minorities Participating in the NHGRI's  
Visiting Investigator Program**

Action Plan 2.6  
Performance Measures 2.6  
Outcome Measures 2.6

**Objective 2.8:**  
**Increase the Number of ELSI Researchers from Minority  
Communities and Minority-Serving Institutions**

Action Plan 2.7  
Performance Measures 2.7  
Outcome Measures 2.7

**AREA OF EMPHASIS NO. 3: EDUCATION AND OUTREACH**

**23**

**Rationale and Priority of Objectives**

**Objective 3.1:**  
**Develop and Distribute an NHGRI Genome Minority Outreach  
Video For Inclusive Education (Genome MOVIE)**

Action Plan 3.1  
Performance Measures 3.1  
Outcome Measures 3.1

**Objective 3.2 :**  
**Continue to Promote and Distribute the NHGRI K-12  
Education Kit "The Human Genome Project – Exploring  
Our Molecular Selves" to Minority Communities**

Action Plan 3.2  
Performance Measures 3.2  
Outcome Measures 3.2

**Objective 3.3:**  
**Establish a Vital Presence at Minority-focused Conferences**

Action Plan 3.3  
Performance Measures 3.3  
Outcome Measures 3.3

**Objective 3.4:**  
**Continue to Support and Develop an Annual Conference  
About the Human Genome Project for the Public**

Action Plan 3.4  
Performance Measures 3.4  
Outcome Measures 3.4

## **National Human Genome Research Institute**

### **Comprehensive Strategic Plan and Budget Submission for the NIH Comprehensive Strategic Plan and Budget To Reduce and Ultimately Eliminate Health Disparities**

#### **National Human Genome Research Institute Mission Statement**

The mission of the National Human Genome Research Institute (NHGRI) is to understand the structure and function of the human genome and the role it plays in health and disease. To that end, the NHGRI supports the Human Genome Project, which is developing tools for improving the effectiveness of human genetics research. NHGRI also supports research that applies the tools produced by the Human Genome Project to studies of specific diseases. The information gained sequencing the human genome will have profound implications for individuals, families, populations, and society. The Ethical, Legal and Social Implications (ELSI) Program was established as an integral part of the Human Genome Project. The ELSI Program was designed to provide a novel approach to scientific research by anticipating, identifying, analyzing, and addressing the ethical, legal and social implications of human genetics research at the same time that the basic scientific issues are being studied.

#### **Overview of the NHGRI Strategy for Addressing Health Disparities**

The very nature of genomic and ELSI research on the human genome demands the inclusion of a cadre of investigators with diverse perspectives and scientific interests. From its inception NHGRI has been concerned about including individuals from various groups in its activities. As the Institute has grown in size and complexity the need for this has become even more imperative and a variety of initiatives have been started and continue to evolve to address this need. This strategic plan brings these diverse activities into a coherent framework that poises the NHGRI to move forward effectively.

In 1998, when NHGRI started an initiative to study human genetic variation, an NHGRI staff group was convened to make recommendations to the Director about how the Institute should proceed to assure that diverse populations were included at all levels of its research programs and other activities. Several goals were identified: to establish meaningful interactions between NHGRI and members of diverse populations; to support research on the ethical, legal, social and health policy issues involved in the use of information about human genetic variation; and to increase the knowledge about genetic variation and its implications among diverse groups, researchers and policy makers. Since that time, the Institute has had discussions with many groups about these goals and has taken a number of steps toward meeting them.

To expand and strengthen these activities further, the NHGRI arranged for Dr. Clifton Poodry, Director of the NIGMS Division of Minority Opportunities in Research, to work with NHGRI on a part time detail. Dr Poodry is providing expert advice about

how NHGRI can move forward more aggressively and effectively to establish training and outreach programs that will develop a genomics and ELSI research community that represents a broad diversity of groups and opinions.

Most recently, in order to address the significant under representation of minorities conducting genomic and ELSI research, NHGRI hosted a workshop in April 2001 to explore new approaches to recruiting minority researchers. A planning committee that included members of the NHGRI staff and the National Advisory Council for Human Genome Research developed the workshop prospectus, participant list and the agenda for the workshop. The workshop was designed "to brainstorm about creative ideas and models for increasing the number of underrepresented minorities pursuing research careers in genomics and related sciences." Invitees included individuals from universities and organizations that had a long history of successfully training underrepresented minorities in research. A number of the initiatives featured in this Strategic Plan were activities recommended by participants in that April 2001 workshop.

A review of the public comments on the October 2000 Strategic Plan to Reduce and Eliminate Health Disparities helped to inform the development of the NHGRI's Strategic Plan. NHGRI's plan places particular emphasis on two of the common themes that clearly emerged from these public comments. First, we are committed to encouraging and also pursuing research partnerships with minority serving institutions in order to build on the technical capabilities for conducting genomic and ELSI research. In this regard, we are especially proud of our long-standing partnership with investigators at Howard University and our contributions to the establishment of the National Human Genome Center at Howard University in May 2001; and more recently of our co-funding of the Vanderbilt-Meharry genetics training grant. Second, NHGRI will continue to build relationships with individuals from minority communities and minority organizations for the purposes of hearing from them about their issues and concerns; for the purposes of information dissemination; and to allow for an opportunity to encourage minority students to pursue careers in genomics and ELSI research.

The NHGRI Strategic Plan for addressing health disparities is organized around the following areas of emphasis, in order of relative priority:

- 1) Research
  - A. Genomic
  - B. Ethical, Legal, and Social Implications (ELSI)
- 2) Training to Build Research Capacity
- 3) Education and Outreach

It is important to recognize that, while we were required to list the relative priority of each area of emphasis, this Strategic Plan is dependent upon the success of objectives in all three areas. Within each area of emphasis, NHGRI has developed a number of objectives to achieve our goals. Each of these objectives in this Plan is presented in priority order within each area of emphasis. NHGRI is committed to carrying out all of

the objectives listed in the report. However, the following projects assume significant co-funding from NCMHD.

<b>Priority for NCMHD co-funding</b>	<b>Area of Emphasis</b>	<b>Objective #</b>	<b>Brief Description</b>
1	Research	1A.1	Howard University and NHGRI Research Collaborations
2	Research	1A.2	Barbados Prostate and Breast Cancer Study
3	Education & Outreach	3.1	Genome Minority Outreach Video for Inclusive Education
4	Research	1A.3	Intramural Health Disparities Research Award

Many of these programs and initiatives are new so that specific performance and outcome measures are challenging to define at this stage. However, whenever possible, specific performance and outcome measures have been included in the specific areas of emphasis. After the first year of experience with some of the new activities, it may be necessary to revisit the measures to determine their appropriateness. It is also important to note that all components of this initiative will be reviewed and evaluated annually by the National Advisory Council for Human Genome Research to assure that the performance and outcome measures are adequately met.

Special emphasis will be placed on tracking the outcomes of these new initiatives, both extramurally and intramurally. To this end, NHGRI will work with investigators of Center grants and training grants as well as other grants that receive minority supplements to help them set ambitious, but realistic measurable goals for their proposed minority training activities. These investigators will be asked to provide information about whether they are achieving the goals set, as part of their progress reports each year. If the goals are not met, a remedial plan will be developed for the following year. Investigators will also be required to develop mechanisms to track individuals being trained, as they progress through their training and career paths. This information will be collected and evaluated by NHGRI staff on an annual basis and reported to the National Advisory Council for Human Genome Research.

In FY2002, NHGRI will establish a baseline for the participation of minorities in its extramural training programs. Baseline data will also be collected in the intramural program. This baseline will then be used to establish appropriate goals for future years, so that minority participation in NHGRI programs increases steadily. In addition, an overall evaluation of the entire Strategic Plan will be conducted after 5 years to assess progress towards the ultimate NHGRI goals. A group of advisors, the majority of whom will be from minority populations, will be recruited to assist with this five-year evaluation. The results of this evaluation will be brought to Council for advice on adjustments that need to be made and future directions that the initiative should take.

**Detailed Plan for NHGRI Areas of Emphasis in Health Disparities Research  
for Minority and Underserved Populations (FY 2002-2006)**

**NHGRI AREA OF EMPHASIS 1: RESEARCH IN HEALTH DISPARITIES**

**Area of Emphasis 1A: Genomic Research - To study the genetic factors that contribute to disease that disproportionately affect minority populations an/or are poorly understood in minority populations**

**Rationale and Priority:**

The vast majority of human genetic information is shared by all people. Indeed, any two individuals share 99.9% of their DNA sequence. Similarly, most genetic variation is shared by all racial and ethnic groups. However, the small differences that do exist include many medically important variations. Thus, while much of human genetics research will apply broadly to all groups of people, regardless of which individuals are studied, it is also important to study the genetic factors that may underlie disparate rates of incidence or patterns of progression among different groups. Risk factors for common diseases such as prostate cancer and diabetes will include both genetic and environmental/behavioral/social factors. NHGRI is working with the Office of Behavioral and Social Science Research to investigate strategies for studying interaction among social, behavioral and genetic factors in health.

During recent months, an unprecedented accumulation of information about the human genome and variations therein has been submitted to public databases and more will be added due to the successes of the Human Genome Project. The scientific opportunity to identify associations between sequence variations and incidence of disease is extraordinary and is being pursued by laboratories worldwide. An important aspect of this effort will be the development of the research capacity in minority serving institutions to conduct research that exploits the tools and information derived from the Human Genome Project.

**Objective 1A.1: Continue and Further Expand Research Collaborations between NHGRI and Howard University**

**1A.1 Action Plan:**

Investigators at NHGRI and Howard University have established a collaborative research partnership to identify the genetic and epidemiologic factors that contribute to the disproportionate increase in complex diseases such as cancer and diabetes in African Americans. This project aims to collect family and population-based information in a way that maximizes the participation of minority physicians, research scientists, and the community. The identification of genes underlying susceptibility to common diseases and an understanding of the function of these genes and their

interaction with environmental factors will lead to improved management and treatment of the diseases.

During the past several years the National Center for Minority Health and Health Disparities (formerly the Office of Research on Minority Health) and NHGRI have supported innovative research collaboration between investigators from Howard University and scientists in the intramural research program of the NHGRI. The collaboration involves support for projects involving African Americans affected with diabetes. In FY 1997 the NHGRI added hereditary prostate cancer within this core of collaborative projects. In addition, Howard University and the NHGRI are serving jointly as research-training sites for African Americans involved in these projects. Among the goals of this collaboration was the establishment of a Center at Howard University for collaborative research on genomic analyses of diseases that disproportionately affect African Americans. This goal was realized on 1 May 2001 when The National Human Genome Center at Howard University was formally dedicated.

**Africa America Diabetes Mellitus Study (AADM):** Because of the high frequency of environmental risk factors for type 2 diabetes in the African American population, it is more productive to study genetic risk factors in West Africans, since they are thought by many anthropologists to be the founding population of modern African Americans and have fewer dietary and nutritional confounding variables. To establish recruitment sites for the study, five sites in West Africa were selected through a peer review process from a total of 24 applications, three in Nigeria and two in Ghana. Because of logistical challenges involved in doing a study of this type in West Africa, the study was planned in stages to allow assessment of the sites' ability to recruit appropriate patients and collect blood, urine, and other clinical data, and successfully send the samples and data to the Coordinating Center at Howard University. The one-year pilot project fully met its goal of recruiting 15 affected sib pairs/site. Based on this experience, a full-scale study was implemented in September 1998 with an anticipated total of 400 affected sib pairs and 200 spouse controls from West Africa by the end of the study period. Genetic analysis of the blood samples is being performed at The Center for Inherited Disease Research (CIDR), a centralized facility established in 1996 by eight Institutes at NIH to provide genetic services for investigators seeking to identify genes that contribute to human disease. The services for the AADM study were awarded through a competitive application process. The study has not only started to yield high quality data, but has assisted in the recruitment of several top-flight scientists to the National Human Genome Center at Howard University.

**African American Hereditary Prostate Cancer Study Network (AAHPC):** The National Human Genome Center at Howard is also coordinating a linkage study of hereditary prostate cancer, the African American Hereditary Prostate Cancer Study Network (AAHPC). The initial aim is to enroll 100 families with prostate cancer in which at least four men, diagnosed at or before 65 years of age, are affected in each family and there are four other (unaffected) relatives available for study. African

American prostate cancer families of this description are almost completely missing from other pedigree collections, despite the higher incidence and higher lethality of prostate cancer in African American men. Through a competitive review, the AAHPC study network has funded seven centers (Detroit, Chicago, New York, Atlanta, Houston, Columbia, and Washington, DC). For most of these, the Principal Investigator is an African American urologist. Community acceptance and participation has been good. Blood samples and clinical data are sent from recruitment sites around the country to the Center for DNA extraction. Over 70 families have already been identified and samples collected. DNA from these families are being studied to see if linkage can be found to a known hereditary prostate cancer location on chromosome 1 as well as whether other linkages exist.

As the first large-scale genetic study of African Americans conducted almost entirely by African American clinical investigators, the AAHPC study has provided a foundation and productive environment for the exploration of all aspects of the involvement of African Americans in genetic research.

### **1A.1: Performance Measures**

- Number of families that have been recruited to participate in the AADM and AAHPC studies
- Scientific publications reporting the progress and results of the research
- Number of faculty, postdoctoral fellows and graduate students trained while conducting this research

### **1A.1 Outcome Measures:**

- A better understanding of genetic risk factors underlying Type II diabetes and hereditary prostate cancer in African Americans.

### **Objective 1A.2: Develop and Conduct “The Barbados Prostate and Breast Cancer Study”**

#### **1A.2 Action Plan:**

The island of Barbados, West Indies, has a relatively homogenous, predominantly black population originating from similar West African regions as the US African American population. The people of Barbados represent an ideal population for genetic-based studies of diseases prevalent in populations of West African ancestry. Prostate and breast cancer are the most common cancers in Barbados. Prostate cancer is substantially more common in African American, and especially Afro-Caribbean men, than in Caucasian or Asian men. Breast cancer, while not more common in blacks than whites, leads to higher mortality in black populations.

NHGRI proposes to complete a national population-based case-control epidemiological study of all incident prostate and breast cancer cases with age/gender matched population

controls in the country over a 4-year period. A familial study component is also planned, given the possibility of common genetic susceptibility in these diseases. Cases will be identified from the only hospital in Barbados and from local physicians, building on the cohorts already studied in earlier NIH-supported studies, and with the previously established cooperation of the Barbados Ministry of Health.

This study will examine a broad range of risk factors and known and novel gene loci with state-of-the-art analytical tools, resulting in a comprehensive approach to the study of prostate and breast cancer. The overall goals will be to identify specific factors involved in genetic susceptibility for these cancers in Barbados, which can then be used to increase overall understanding of disease in this and other populations. Specific aims of the research include:

- Developing procedures for identifying and recruiting participants for the Barbados Prostate and Breast Cancer Study.
- Developing and testing protocols and data collection forms to be used in the Barbados Prostate and Breast Cancer Study.
- Evaluating procedures to determine patterns of incidence and mortality outcomes for persons with prostate and breast cancer, in order to establish guidelines for the development of a Cancer Register.

This study will be a cooperative effort by the University of the West Indies, the State University of New York (SUNY) at Stony Brook, and the National Human Genome Research Institute. Given previous experiences of involved investigators, broad case and control participation is expected, with positive implications for this study's success.

#### **1A.2 Performance Measures:**

- Recruitment of minority families to participate in the Barbados Prostate and Breast Cancer Study
- Scientific publications that report the progress and results of the research
- Number of faculty, postdoctoral fellows, and graduate students trained during the course of conducting this research

#### **1A.2 Outcome Measures:**

- Identification and better understanding of genetic and other risk factors for prostate and breast cancer in populations of African descent.
- Documentation of patterns of incidence, morbidity and mortality for these cancers.

#### **Objective 1A.3: Implement a New Program Called Intramural Health Disparities Research Award**

### **1A.3 Action Plan:**

Starting in FY 2002, the intramural staff plans to launch an Intramural Health Disparities Research Award. The goals of this award will be to increase the number of projects focusing on health disparities, to increase the number of minority post-doctoral fellows and students conducting research in the intramural program, and to encourage interdisciplinary collaborations. Supplementary funding will be offered to investigators who wish to expand their research programs in this area.

### **1A.3 Performance Measures:**

- Increased number of research projects addressing health disparities in the Division of Intramural Research at NHGRI
- Increased number of minority trainees
- Increased number of publications relevant to issues of health disparities

### **1A.3 Outcome Measures:**

- Increased number of publications with minorities as authors
- Increased understanding of health disparities

### **Area of Emphasis 1B: Ethical, Legal, and Social Implications (ELSI) Research-To explore the ethical, legal and social issues (ELSI) related to genetics and health disparities.**

#### **Rationale and Priority:**

Human genetic variation research, especially as it relates to risk factors for common, complex disorders, is leading to increased knowledge regarding variation among individuals and how this variation may contribute to the health status of individuals. It is also leading to more knowledge about variation within and among different racial and ethnic groups (to the extent that such groups can reasonably be identified) and how this variation may contribute to the aggregate health status of those groups. The NHGRI's new initiative to develop a haplotype map of the human genome will make it possible to conduct disease gene association studies more quickly and efficiently than ever before, resulting in an even more rapid proliferation of this new information. Information regarding variation within and among groups, in particular, will increasingly be generated, because the map will facilitate the conduct of association studies in selected populations where certain diseases are more or less prevalent.

While the ultimate goal of studies aimed at relating human genetic variation to disease risk is the improvement of human health, concerns have been raised that the findings of some genetic variation research may be misunderstood. Concerns have also been raised that such findings, if interpreted incorrectly and misused, will exacerbate, rather than ameliorate, already-existing health disparities among racial, ethnic, and socio-economic groups. The NHGRI, through its Ethical, Legal, and Social Implications (ELSI) Research Program,

proposes a new initiative to encourage additional research on the ELSI implications of genetic variation research for both individuals and for diverse population groups.

**Objective 1B.1: Study the Ethical, Legal and Social Implications of Genetic Variation Research for Individuals and Diverse Racial and Ethnic Groups (FY1999-2002 and FY2002-FY2005)**

**1B.1 Action Plan:**

In FY 1999 the ELSI program issued a Request for Applications (RFA) to solicit applications for studies to identify, explore and address the ELSI issues that arise in the course of research on human DNA sequence variation and in the use of the information that may result from this research. Particular emphasis was placed on studies that explore: 1) how research on human genetic variation can be conducted in an ethically and culturally sensitive way; 2) how the information that results from this research will interact with current concepts of race and ethnicity; 3) how cultural and socioeconomic factors influence the interpretation and use of this information; and 4) how this information may influence access to and use of genetic health services by various groups. Five studies were funded under this RFA in FY 2000, all of which are relevant to the issue of health disparities. Bringing together investigators from these projects and three other related projects, the ELSI research program has organized a consortium of supported studies and the principal investigators meet once or twice a year. These meetings allow researchers to compare findings on issues common to all the projects, reduce duplication of effort, and promote sharing of information.

To build on this beginning, in FY 2002, the ELSI Research Program will release a second RFA for studies of the ethical, legal and social implications of genetic variation research for individuals and diverse racial and ethnic group. This RFA will be designed to elicit research that further addresses the issues outlined above, building on the research funded from 1999-2002.

The timeline for this new initiative is FY 2002 through 2005.

**1B.1 Performance Measures:**

- Increased number and quality of applications received in response to this initiative over those received in response to the FY 1999 initiative.
- Organization of a consortium of investigators from supported studies that will meet once a year. These meetings will allow researchers to compare findings on issues common to all projects, reduce duplication of effort, and promote sharing of information.
- Assessment on an annual basis of the quantity and quality of published research resulting from this initiative, the number of minority grantees, and the number of individuals from diverse racial and ethnic groups included as research participants.
- Increased number of publications.

### **1B.1 Outcome Measures:**

- Establishment of a body of scholarship that informs researchers on the ELSI implications of genetic variation research
- Establishment of a body of scholarship on how minority communities can be involved in genetic research

### **Objective 1B.2: Study Hereditary Hemochromatosis and Iron Overload Disease in Diverse Populations**

#### **1B.2 Action Plan:**

Iron overload is a common disorder affecting between 1 in 200 and 1 in 400 individuals. One of the major causes of iron overload is a genetic disorder known as hereditary hemochromatosis (hh). It has been estimated that one in every 400 Caucasians has hh due to genetic mutations in their HFE genes. The genetic contributions to hh in non-Caucasian populations are not at all clear. In 1997, a panel of experts concluded that it would be premature to consider widespread genetic screening for hh until some important questions about prevalence, penetrance, genotype/phenotype correlations and the psychosocial impact of wide-spread or population-based screening were addressed.

In order to answer these questions, NHGRI is collaborating with the National Heart, Lung, and Blood Institute (NHLBI) to fund a large multi-center study designed to study iron overload and hh in diverse populations. Five field centers (including Howard University), a coordinating center, and a central laboratory have been funded to carry out this project. It is expected that 100,000 participants will be recruited and screened for these disorders during the course of this five-year study, including 45,000-50,000 individuals from minority communities. This will enable the researchers to answer important questions identified above, particularly for individuals from minority communities.

Recruitment of participants began in February 2001. At the end of FY2001, approximately 20,000 individuals had been recruited for the initial screen (about 50% were from minority communities).

#### **1B.2 Performance Measures:**

- In FY2002, another 50,000 individuals will be recruited for this study (45-50% of these individuals will be from minority communities);
- In FY2003, the final 25,000 individuals will be recruited for this study, making 100,000 total participants (45-50% of these individuals will be from minority communities);
- In FY2004, data will be analyzed from this study;
- In FY 2003-6, scientific publications will report the progress and results of the research, particularly as they relate to iron overload disorders and hh in minority communities.

### **1B.2 Outcome Measures:**

- Increased understanding of the genetic factors underlying hh in diverse populations and the impact of this knowledge for these populations.

### **Objective 1B.3: Implement ELSI/ Native American Research Centers for Health (NARCH) Initiative**

#### **1B.3 Action Plan:**

The NHGRI has partnered with NIGMS and the Indian Health Service to support research into the ethical, legal and social implications of genetic research as part of the new Native American Research Centers for Health (NARCH) initiative. This initiative supports partnerships between American Indian or Alaska Native tribes or of tribal-based organizations such as the National Indian Health Board and Area Health Boards, and institutions that conduct intensive academic-level biomedical and behavioral research.

#### **1B.3 Performance Measures:**

- Number of participants recruited
- Number of projects on ELSI issues
- Scientific publications that report the progress and results of the research

#### **1B.3 Outcome Measures:**

- American Indians and Alaska Natives develop expertise and participate in research on the ELSI implications of genetic and genomic research.

### **Objective 1B.4: Develop ELSI Research on Minority Participation in Genetic Research**

#### **1B.4 Action Plan:**

The NHGRI will support the analysis of ethical, legal and social issues affecting African American participation in genetic research. Historically, African Americans have not participated in genetic research at the same level as members of other racial or ethnic groups. The research collaborations between NHGRI and the National Human Genome Center at Howard University on diseases that disproportionately affect African-Americans (see Objective 1A.1) offer a unique set of data that will allow investigators in bioethics and the social sciences to examine the factors leading African-Americans to either agree or refuse to participate in genetic research. Special emphasis will be placed on access to information, informed consent, community attitudes toward genetic research, and methods to optimize informed decision making regarding participation in genetic research and use of the knowledge gained through this research. It is hoped that the research supported by

this initiative will serve as a model of ELSI research that will be critical to the design of future genetic research involving minority communities.

**1B.4 Performance Measures:**

- Number of projects started
- Number of minority investigators involved
- Number of minority trainees participating
- Inclusion of minorities as research participants
- Published research

**1B.4 Outcome Measures:**

- Increased understanding of factors underlying African-American participation or refusal to participate in genetic studies.

**NHGRI AREA OF EMPHASIS 2 – TRAINING TO BUILD RESEARCH CAPACITY**

**Rationale and Priority:**

The success of the Human Genome Project depends in part on the ability to train a diverse cadre of scientists in genomic and ELSI approaches to human genome research. The challenges are formidable. The representation of minority individuals in genetics and genomics research and clinical medicine is tragically low. Recent statistics reveal that of 4,810 U.S. based members of the American Society of Human Genetics, 0.6% are American Indian, Eskimo or Aleut; 1.1% are African American, and 0.7% are Hispanic. Ethnicity data from other genetics professional societies reflect similar levels of underrepresentation. To attract individuals from minority communities to this field, interest and excitement in genomics and genetics needs to be engendered early. Furthermore, the initiatives must aim to encompass all stages of the career ladder from student to professional, with opportunities for training at every stage.

The NHGRI has developed a series of new training initiatives targeted toward both minority individuals and minority serving institutions. The goals for this area of emphasis are: to increase the diversity of investigators participating in genomic approaches to research; to increase the diversity of students trained in genomic approaches to research; and to expose a greater diversity of students and faculty to genomic approaches to research.

**Objective 2.1: Expand Training Opportunities at Centers of Excellence in Genomic Science (CEGS) and Genome Production Centers**

### **2.1 Action Plan:**

The new Centers of Excellence in Genomic Science (CEGS) centers will be the centerpiece of NHGRI supported research in the future and will have training of new investigators as an essential component. They are therefore ideal sites for the training of individuals from underrepresented minorities as well. Each CEGS will be asked to propose what specific training activities they will implement and how they will recruit the relevant trainees. Staff will encourage the CEGS as a group to generate a range of training opportunities focused on underrepresented minorities. The requirement for this training component will be made explicit in the revised program announcement that was issued in Summer 2001. The Centers will be expected to have an average of 10% of their trainees from underrepresented minority populations.

### **2.1 Performance Measures:**

- Number of programs implemented for increasing the number of minorities exposed to genomics research
- Number of minorities participating in these programs

### **2.1 Outcome Measures:**

- Increased number of minorities trained and proceeding onto research careers

### **Objective 2.2: Continue to Support and Develop the “Genomics Short Course for Faculty at Minority Serving Institutions”**

### **2.2 Action Plan:**

The annual Short Course for Faculty at Minority Institutions is designed to update faculty from institutions with substantial minority enrollment on the latest developments in genetic technology, medical genetics, gene therapy and ethics. The course also assists attendees in incorporating this information into classroom teaching to cultivate minority student interest in genome research, and offers information on careers in genetics and grant writing skills. Participants visit NHGRI laboratories and experience first-hand the latest technologies that are being applied to genetics research. In 2001, over 30 faculty members participated in the program.

### **2.2 Performance Measures:**

- Number of applicants
- Number of attendees
- Feedback from evaluation forms

### **2.2 Outcome Measures:**

- Integration of genomics into curriculum at minority serving institutions
- Integration of Short Course information into curriculum at minority serving institutions

- The importance of underrepresented minorities participating in genomics and ELSI research emphasized with grantees during the review of their annual research progress.

### **2.3 Outcomes Measures:**

- Increased number of underrepresented minorities taking courses in genomics and/or pursuing genomic or ELSI research.

### **Objective 2.4: Increase the Number of Minorities Participating in the T32 Training Grant Program.**

The NHGRI currently supports eleven training grants; three of these are currently in a no-cost extension. The average percentage of underrepresented minorities on all training grants ranges between 5% and 10%.

### **2.4 Action Plan:**

To increase the number of underrepresented minorities participating on T32 training grants, program directors will be made aware that recruitment and appointment of underrepresented minorities on training grants must be a high priority in order for the program to continue. The NHGRI grantees meeting scheduled for the fall of 2001 should provide program directors with additional information about resources that are available to them. A goal of 10% minority trainees enrolled over the life of the training grant has been set.

### **2.4 Performance Measures:**

- A brochure with information about NHGRI's supported T32 training grant programs distributed at conferences attended by underrepresented minorities, such as SACNAS and ABRCMS.
- Training directors attend and make presentations about their research and training grant programs at scientific/professional meetings that are of specific interest to underrepresented minorities, such as SACNAS, ABRCMS, or where active minority committees exist, such as American Society of Cell Biology, and professional/scientific meetings.
- An average of 10% of trainees on board from minority populations within the next three years. Eventually, the percentage should rise to the percentage of minorities in the baccalaureate population.

### **2.4 Outcomes Measures:**

- Number of underrepresented minorities who will pursue careers in genomic or ELSI research.

**Objective 2.5: Increase the Number of Summer Internship Opportunities for Minority Students**

**2.5 Action Plan:**

NHGRI hosts summer internships in Intramural laboratories. In previous summers, the participation of minority students has been relatively low.

The DIR is implementing new measures in an effort to increase the level of participation by minority students. These measures are primarily focused on outreach and recruitment efforts.

**2.5 Performance Measures**

- Outreach efforts to increase application submissions by minority students.
- The number of applications submitted by minority students
- The number of minority students participating in the summer student program

**2.5 Outcome Measures:**

- The number of minority students going on to graduate studies in science

**Objective 2.6: Increase the number of Summer Internship Opportunities for Minority students at the National Human Genome Center at Howard University (NHGC)**

**2.6 Action Plan:**

The DIR and the NHGC have been actively involved in major collaborations involving health disparities research. (see section 1.A.1)

In an effort to increase the participation of minority students in health disparity research, NHGRI has initiated a summer student program at the NHGC. 10 students participated during the inaugural year of the program. The number has been increased to 24 for FY02.

**2.6 Performance Measures:**

- Outreach efforts to increase application submissions by minority students.
- The number of applications submitted by minority students
- The number of minority students participating in the program at Howard.

**2.6 Outcome Measures:**

- The number of minority students going on to graduate studies in science.

**Objective 2.7: Increase the Number of Minorities Participating in the NHGRI's Visiting Investigator Program**

**2.7 Action Plan:**

The Visiting Investigator Program (VIP) is designed to provide tenured or tenure-track faculty-level investigators from outside the NIH with the opportunity to spend six- to 12-months in the NHGRI laboratories, accomplishing significant research objectives. Visiting investigators can use their stay to learn new technologies, develop research collaborations, or pursue sabbatical research projects. Basic, clinical, and social scientists may access the Institute's laboratories, core facilities, and training programs for study in any area of human genetic disease, including the ethical, legal and social implications of genetic research. The purpose of this program is to increase the number of minority scientists involved in genomic research.

**2.7 Performance Measures:**

- The number of inquiries and applications to the VIP program by minority investigators.

**2.7 Outcome Measures:**

- The number of minority investigators participating in the program.

**Objective 2.8: Increase the Number of ELSI Researchers from Minority Communities and Minority-Serving Institutions**

**2.8 Action Plan:**

Very few ELSI researchers currently funded (or funded in the past) are from minority communities. The ELSI program plans to increase its efforts to reach out to minority researchers and researchers at minority-serving institutions, in order to inform them about ELSI Research funding opportunities. In addition, ELSI staff will organize and provide technical assistance seminars and workshops for minority communities on grant writing. Further, ELSI staff will provide assistance to minority investigators and investigators from minority-serving institutions through the application process.

**2.8 Performance Measures:**

- Efforts to provide information about funding opportunities to members of minority communities (e.g. through mailings to minority institutions, attendance at minority meetings such as the Society for the Advancement of Chicanos and Native Americans in Science (SACNAS) and the Annual Biomedical Research Conference for Minority Students (ABRMRS))
- Number of seminars and technical assistance workshops for minority communities on grant writing

- Percent of ELSI grant applications that come from individuals from minority communities or from researchers at minority-serving institutions and are successful.
- Number of ELSI research papers published by researchers from minority communities or researchers from minority serving institutions.

### **2.8 Outcome Measures:**

- Increased number of ELSI researchers from minority communities and minority-serving institutions

## **NHGRI AREA OF EMPHASIS 3 – EDUCATION AND OUTREACH**

### **Rationale and Priority:**

The broader inclusion of minorities in biomedical research is imperative if we are to see it applied to improve the health of all citizens. Inclusion is of particular importance for NHGRI, given the far-reaching implications of genetic information and technologies for both individuals and groups. There is a sense of urgency in the recruitment of individuals to meet the monumental challenges of translating human genome sequence information to items of societal value. The initial sequence and analysis of the human genome represents a significant milestone in science, yet formidable challenges remain in translating this genetic sequence information into clinically beneficial diagnostics and therapeutics and in generating public policy to minimize the potential misuses of genetic information. To be ultimately successful, the improvements in research and health care must reach everyone. This success will depend on participation by individuals from diverse communities.

We hope to take advantage of the momentum resulting from increased visibility of the Human Genome Project since the publication of the draft human genome sequence in February 2001. This is an opportune time to redouble our education and outreach efforts in order to encourage individuals from historically underrepresented communities to actively participate in the genetics revolution – as scientists, clinicians, research participants, and active contributors in deliberations of ethics and public policy.

The success of the Human Genome Project depends in part on the ability to communicate advances in genomic and ELSI research to all communities and to empower them to utilize new genetic information and technologies to maximize benefits and avoid potential misuses. In essence, the goals for this area of emphasis are: To foster development of policy, education and outreach activities relevant to increasing the involvement of diverse communities in all aspects of genetic research, to reducing health disparities, and to integrating new genetic technologies into health care in underserved communities.

### **Objective 3. 1: Develop and Distribute an NHGRI Genome Minority Outreach Video for Inclusive Education (Genome MOVIE)**

#### **3.1 Action Plan:**

NHGRI is creating a video to communicate the excitement and opportunities in genomic research to underrepresented minorities. The goals of this video are to aid in the recruitment of individuals from historically underrepresented communities to become active participants in genomics and genetics at all levels, to reflect the diversity of the target audience, to provide compelling role models, and to serve as a resource for anyone providing outreach and education to minority and underrepresented communities.

The aim of the video is to effectively communicate NHGRI's excitement about the promise of genomics and genetics that will in turn generate an excitement and motivation to participate in one of the greatest scientific and societal challenges of our time. The successful production of this video would fill a critical need for resources in the NHGRI minority outreach program that is a key component of our strategy to address health disparities.

In order to insure that the content and presentation of the NHGRI video meet the needs of the communities to which it is targeted we will work with educators and individuals with expertise in the development of culturally competent educational materials for diverse communities. These experts will help shape the stories, images and sounds needed to capture the enthusiasm of the intended audiences as well as the help develop the most effective strategies to promote, disseminate and evaluate the Genome MOVIE.

The Genome MOVIE will be in development throughout FY2002 and is targeted for distribution and promotion in FY 2003. Evaluation of the product will be conducted through FY2003 and FY 2004.

#### **3.1 Performance Measures:**

- Development of a product that captures the excitement of the Human Genome Project and translates it to its target audience
- Dissemination to minority schools and communities
- Utilization by minority schools and communities
- Evaluation of the quality and effectiveness of the video

#### **3.1 Outcome Measures:**

- Increased number of members of underrepresented communities as active participants in genomics and genetics at all levels

**Objective 3.2: Continue to Promote and Distribute the NHGRI K-12 Education Kit “The Human Genome Project - Exploring our Molecular Selves” to Minority Communities**

**3.2 Action Plan:**

An educational kit –“The Human Genome Project - Exploring our Molecular Selves”- has been produced by NHGRI in connection with the publication of the initial working draft sequence of the human genome in February 2001. This kit provides unprecedented access to cutting-edge information about the Human Genome Project for life sciences education to minority schools and communities. The kit has been widely distributed and enthusiastically received. Staff will continue to actively inform teachers and administrators at schools with significant minority enrollment about the availability of the web-based kit (all 60,000 mail ordered copies of the kit have been distributed). Staff will also evaluate the distribution, use and value of the kit in schools with significant minority enrollment and assess means of better reaching these schools and develop additional materials that address their specific needs. In order to assist with integration of the educational materials into schools, NHGRI is coordinating the establishment of a mentorship network with student and faculty members of the American Society for Human Genetics and others from the scientific community.

The results of the evaluation of the education kit will be used to help encourage the use of the resource, to update and improve the materials, and to help make decisions about producing and distributing similar resources.

**3.2 Performance Measures:**

- Dissemination to minority schools and communities
- Wider access to the education kit through reformatting the CD-ROM and making the materials available for download on the web.
- Effectiveness of the mentorship network in helping local high school classrooms use the education kit
- Evaluation of the quality and effectiveness of the education kit

**3.2 Outcome Measures:**

- Increased number of community interactions involving the education kit
- Increased utilization of the education kit by minority schools and communities

**Objective 3.3: Establish a Vital Presence at Minority-focused Conferences**

**3.3 Action Plan:**

NHGRI will establish a presence at conferences targeted toward minorities (e.g. Society for Advancement of Chicanos and Native Americans in Science National Conference, Annual Biomedical Research Conference for Minority Students) by hosting a visible and active

exhibit booth; organizing genomics symposia; compiling an attractive brochure that highlights opportunities for minority students in genomics; and hosting roundtables or hospitality suites so that students have a chance to talk with staff from the various training programs of NHGRI. NHGRI will actively seek out opportunities to give presentations to groups such as Zeta Phi Beta, the National Association of Hispanic Nurses, the Association of American Indian Physicians, the National Medical Association, the Intercultural Cancer Coalition, and other organizations serving the communities appropriate for ELSI research. NHGRI will participate in at least three conferences per year aimed at minority or underrepresented communities.

### **3.3 Performance Measures:**

- Increased number of conferences with an active NHGRI exhibit booth
- Increased number of interactions with minority staff and students and/or students and faculty at minority serving institutions
- Increased number of genomic symposia/presentations at minority focused conferences
- Increased number of students and faculty that participate in NHGRI training activities because of information provided by NHGRI staff at conferences.

### **3.3 Outcome Measures:**

- Increased knowledge of NHGRI training and research programs by minorities.
- Increased participation in NHGRI training and research programs by minorities.

### **Objective 3.4: Continue to Support and Develop an Annual Conference about the Human Genome Project for the Public**

#### **3.4 Action Plan:**

The social, ethical and policy challenges posed by genomics can only be addressed justly and equitably by ensuring that all segments of the population are able to actively participate. NHGRI will make every effort to increase minority participation in the NHGRI Annual Human Genome Project information conferences (also known as “Consumer Day”), which were initiated in the fall of 1999. The goal of the annual conferences is to engage and empower the public and members of voluntary health organizations to become informed about and engaged in all aspects of the Human Genome Project. Minority institutions and organizations will actively be sought as partners in the planning process to develop a program that will attract minority participation. For the Fall 2001 conference, NHGRI will partner with minority organizations and institutions in the DC area (Zeta Phi Beta Sorority - National Education Foundation, the National Human Genome Center at Howard University, and the Family Life Center Foundation at Shiloh Baptist Church) with a goal of developing a program that can be replicated across the country, ideally by NHGRI grantees in partnership with their local minority institutions and organizations. In future years, similar partnerships will be developed with other minority communities.

### **3.4 Performance Measures:**

- Attendance by members of minority communities
- Attendance by minority students from local schools
- Conference evaluation forms from attendees to measure impact of these conferences
- Development of additional Human Genome Project information conferences for communities around the country

### **3.4 Outcome Measures:**

- Increased participation in NHGRI activities by minorities.
- Increased linkages with minority organizations and institutions

NHGRI Health Disparities Budget  
(Dollars in Millions)

Institute / Center	FY 2002			FY 2003		
	Research	Infrastructure	Outreach	Research	Infrastructure	Outreach
NHGRI	\$12.40	\$1.80	\$0.60	\$13.20	\$2.40	\$0.40