

NATIONAL HUMAN GENOME RESEARCH INSTITUTE

NIH Health Disparities Strategic Plan

Fiscal Years 2004-2008



MISSION STATEMENT

The National Human Genome Research Institute (NHGRI) led the National Institutes of Health's (NIH) contribution to the International Human Genome Project, whose primary goal was the sequencing of the human genome. This project was successfully completed in April 2003. Now, the NHGRI's mission is focused on a broad range of studies aimed at understanding the structure and function of the human genome and its role in health, disease, and society.

To that end, NHGRI supports the development of resources and technology that will accelerate genome research and its application to human health. A critical part of NHGRI's mission continues to be the study of the ethical, legal, and social implications (ELSI) of genomic research. NHGRI also supports the training of scientists and clinicians, as well as the dissemination of genomic information to both the general public and to health professionals.

STRATEGY FOR ADDRESSING HEALTH DISPARITIES

The NHGRI Health Disparities Strategic Plan defines currently underrepresented individuals as those individuals from the following groups:

- **individuals from underrepresented racial and ethnic groups;**
- **individuals with disabilities; and**
- **individuals from socially, culturally, or educationally disadvantaged backgrounds.**

NHGRI Research Goal

The National Human Genome Research Institute recognizes that any role that genetics and genomics may play in causing health disparities is limited. It is understood that many of the causes of health disparities do not have a genetic contribution. Thus, it is not the intent of NHGRI to “overemphasize genetics as a major explanatory factor in health disparities” (Sankar P., et al., Genetic Research and Health Disparities, *JAMA* Vol. 291 No. 24 p. 2985-2989, June 23, 2004). Rather, the contributions that the National Human Genome Research Institute can make in addressing and eliminating health disparities in populations affected by health disparities will derive from an integrated approach to the Institute's overall mission to understand further the structure and function of the human genome and its role in health, disease, and society.

The NHGRI Health Disparities Research Plan reflects the overall goals of the NIH. NHGRI's specific research goals in this area are to:

1. Advance our understanding of the development and progression of diseases that contribute to health disparities.
2. Develop a detailed understanding of the heritable variation in the human genome.

3. Develop robust strategies for identifying the genetic contributions to disease and drug response.
4. Use newly acquired knowledge regarding genes and pathways as a basis for the development of new, targeted therapeutic approaches to diseases that contribute to health disparities.
5. Investigate how genetic risk information is conveyed in clinical settings and how that information influences health strategies and behaviors.
6. Advance our understanding of the relationships between genomics, race, and ethnicity, and the consequences of uncovering these relationships.
7. Develop new or improved approaches for detecting and diagnosing the onset of diseases that contribute to health disparities.
8. Advance our understanding of the ethical, legal, and social implications of genome research and its role in understanding the causes of health disparities.

NHGRI Research Capacity Goal

The very nature of genomic and ELSI research requires the involvement of a large cadre of investigators with diverse perspectives and varying scientific interests. Since its inception, NHGRI has made a significant effort to include individuals with a variety of perspectives in all of its activities. As the Institute has grown in size and complexity, the need for inclusion and collaboration has become even more imperative. A number of initiatives have been undertaken on this front, and they continue to evolve. This Strategic Plan is intended to provide a coherent framework for all of NHGRI's activities related to health disparities and allow the Institute to further refine its programs and develop new programs in the most effective way possible. NHGRI's research capacity goals are to:

1. Increase the diversity of those engaged in (including as subjects of) genome research.
2. Expand opportunities in research training and career development for research investigators from populations affected by health disparities, including racial and ethnic minority populations.
3. Increase the number of researchers conducting research focused on genomic and health disparities.
4. Increase representation in peer review of scientists and others from populations affected by health disparities.
5. Promote the development of inter-institutional partnerships between historically research-intensive and historically minority-serving institutions that seek to build the latter's research infrastructure.

NHGRI Community Outreach, Information, and Dissemination: Public Health Education Goal

“Marked health improvements from integrating genomics into individual and public health care depend on the effective education of the public and health professionals about [the] interplay of genetic and environmental factors in health and disease” (Collins F.S., et al. A

vision for the future of genomics research, *Nature*, Vol. 422, No. 6934, April 24, 2003, p. 835-847). The NHGRI will expand its current activities and create new programs to:

1. Provide current information to health care providers, enabling them to enhance the quality of care provided to individuals within populations affected by health disparities.
2. Facilitate the incorporation of relevant health care information into the curricula of medical and allied health professions schools, public health schools, and the continuing education activities of health professionals.
3. Maintain ongoing communication linkages and partnerships with community-based and faith-based organizations, health care associations, foundations, and academic institutions; and foster dialogues with populations affected by health disparities in areas where NHGRI is actively conducting research.
4. Collaborate with public health and health-oriented policy centers to translate research findings into policy documents that can be used by policy groups and other stakeholders to explain new, important discoveries to decision-makers.

Application of Public Comments

In developing this plan, NHGRI has taken into consideration the public comments on the Trans-NIH Strategic Plan for FY 2002-2006 and the input of the academic and research communities published in scientific commentaries. NHGRI's revised plan incorporates these public comments in the following manner:

1. NHGRI seeks to not overemphasize the role of genetics and genomics as the "major explanatory factor in health disparities" (See, JAMA June 23, 2004. Vol. 291. No 24 at 2985-2989).
2. The International Haplotype Map ("HapMap") project includes populations from across the world, initially including populations from Japan, China and Nigeria (Yoruba). These groups were carefully selected as part of an inclusive process involving many members of the HapMap Consortium. Community Advisory Groups were established in Nigeria, Japan, and China, to obtain specific inputs from public members of the involved communities about the design of the work. The HapMap project is also testing the expectation that information gathered from these populations will be broadly applicable to all populations in the world.
3. NHGRI will continue to sponsor and enhance programs aimed at mentoring, educating, and providing grant support to scientists and populations from underrepresented backgrounds who are actively pursuing research careers.
4. NHGRI is promoting partnerships between HBCUs and institutions with significant populations of students from underrepresented backgrounds and research-intensive institutions.
5. NHGRI education and outreach efforts are increasing the information that is provided for individuals whose native language is not English. For example, NHGRI, in collaboration with the Genetic and Rare Disease Information Center, is developing

- new programs to specifically reach Spanish-speaking members of the United States population.
6. In 2003, NHGRI developed a new Branch focusing on Public Education and Community Involvement, whose goal is to promote and direct NHGRI's public information and community outreach activities.
 7. In 2003, NHGRI developed a new Intramural Research Branch focusing on social and behavioral research. This new Branch investigates the social and behavioral factors that facilitate translation of genomic discoveries for health promotion, disease prevention, and health care improvements.

1.0 AREAS OF EMPHASIS IN RESEARCH

1.1 Area of Emphasis One: Genomic Research - To study the genetic factors contributing to diseases that disproportionately affect populations affected by health disparities.

All people share the vast majority of the human genetic information. Indeed, any two individuals share 99.9% of their DNA sequence. Similarly, all populations share most genetic variation. However, the small differences that do exist include variations that have important medical consequences. Thus, while most of human genetics research will apply broadly to all groups of people regardless of which individuals conclusions are based upon, it is also important to study whether specific genetic factors may underlie disparate rates of incidence or patterns of progression of disease when they are observed among different groups. Risk factors for common complex diseases such as prostate cancer and diabetes include both genetic and environmental/behavioral/social factors, but in most cases, the relative contribution of these factors is not known. NHGRI is working with the Office of Behavioral and Social Science Research (OBSSR) to investigate strategies for studying the interplay between social, behavioral, and genetic factors with respect to human health. As mentioned above, NHGRI has also established an Intramural Research Branch specifically focused on these types of questions.

Scientific opportunities to identify associations between sequence variations and incidence of disease have increased in extraordinary ways in the past several years, and they are being actively pursued by laboratories worldwide. An important aspect of these efforts will be the development of the research capacity in Historically Black Colleges and Universities (HBCUs) to conduct research that exploits the tools and information derived from the Human Genome Project, the HapMap project, and similar large-scale studies.

1.1.1 Objective One: Continue and Further Expand Research Collaborations between NHGRI and HBCU's

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. The identification of genes underlying susceptibility to common diseases and, more importantly, a thorough understanding of the function of these genes and their interaction with environmental factors will lead to improved management and treatment of these diseases. This project aims to collect family- and population-based information in a way that maximizes the participation of minority physicians, research scientists, and underrepresented communities.

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 an innovative research collaboration between investigators from Howard University and scientists in NHGRI's Intramural Research Program. The collaboration involves support for projects involving African-Americans affected with diabetes. In FY 1997, NHGRI added hereditary prostate cancer to this set of collaborative projects. In addition, Howard University and NHGRI are serving jointly as research-training sites for African-American scientists involved in these projects. Among the specific 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 May 1, 2001, when The National Human Genome Center at Howard University was formally dedicated. NHGRI will continue to develop similar collaborations with other HBCU's in order to promote genomics-based research.

NHGRI took into consideration the public comments, as described above. This Emphasis Area addresses one of the six major themes: "Improve research infrastructure at minority academic institutions."

1.1.1.2 Performance Measures

- Number of collaborations started with investigators and/or scientists from diverse groups
- Inclusion of individuals from health disparities populations as research participants
- Published research

1.1.1.3 Outcome Measures

- Increased participation of HBCU's and/or investigators from diverse groups in research collaborations with NHGRI Intramural scientists.
- Increased understanding of factors underlying racial and ethnic minority participation (or refusal to participate) in genetic studies

1.1.2 Objective Two: Africa-America Diabetes Mellitus Study (AADM)

1.1.2.1 Action Plan:

Because of the high frequency of environmental risk factors for type 2 diabetes in the African-American population, it is particularly 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 Africa-America Diabetes Mellitus Study (AADM), five sites in West Africa were selected through a peer review process from a total of 24 applications; three of these sites are in Nigeria and two are in Ghana. Because of the 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, 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 successfully met its goal of recruiting 15 affected sibling pairs ("sib-pairs") per site. Based on this experience, a full-scale study was implemented in September 1998; this study successfully met its goal of recruiting 400 affected sib pairs and 200 spouse controls from West Africa by the end of the study period. Genetic analysis of the blood samples was performed at The Center for Inherited Disease Research (CIDR). The services for the AADM study were awarded through a competitive application process. The study has not only yielded high-quality data, but has led to the recruitment of several top-flight scientists to the National Human Genome Center at Howard University. A number of phenotypic measurements were made on the research participants, who have stayed in touch with clinical personnel. Analysis of the genotype and phenotype data has led to the identification of several chromosomal regions that appear to harbor susceptibility genes for diabetes and related traits in this West African population. This work has recently been published (Rotimi CN et al., *Diabetes* 53:838-841, 2004). A substantial grant application is being prepared for submission to NIDDK to support the continuation of this work.

1.1.2.2 Performance Measures:

Fine mapping of regions with evidence for linkage from the genome-wide scan, follow-up measurements, and ascertainment of the status (alive/dead, and any clinical complications) of subjects.

1.1.2.3 Outcome Measures:

- A better understanding of the etiology of type 2 diabetes mellitus in African Americans, meeting NHGRI research goals of 1) advancing our understanding of the development and progression of type 2 diabetes in individuals of African descent, and 2) the development of new or improved approaches for detecting and diagnosing the onset of type 2 diabetes in these individuals

1.1.3 Objective Three: African-American Hereditary Prostate Cancer Study

Network (AAHPC)

1.1.3.1 Action Plan:

The National Human Genome Center at Howard is also coordinating the African-American Hereditary Prostate Cancer Study Network (AAHPC). The study has enrolled 100 families with hereditary prostate cancer in which at least four men, diagnosed at or before 65 years of age, are affected in each family; four other (unaffected) relatives must also be available for study. African-American prostate cancer families fitting this description are almost completely missing from other pedigree collections, despite the higher incidence and lethality of prostate cancer in African-American men. DNA from these families is being studied to see if linkage can be found to a known hereditary prostate cancer location on chromosome 1, as well as whether linkage to other chromosomal locations (or specific genes) exists.

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.

NHGRI took into consideration public comments, as described above. This Objective addresses two of the six major themes: 1) “Improve research infrastructure at minority academic institutions;” and 2) “Expand the scope of research to include cultural, psychological, behavioral, social, gender, environmental and biological factors appearing unique to race.”

1.1.3.2 Performance Measures:

- Scientific publications reporting the progress and results of the research
- Number of faculty, postdoctoral fellows, and graduate students trained while conducting this research

1.1.3.3 Outcome Measures:

A better understanding of genetic risk factors underlying hereditary prostate cancer in African-Americans, meeting NHGRI’s research goals of 1) advancing our understanding of the development and progression of hereditary prostate cancer in individuals of African descent and 2) the development of new or improved approaches for detecting and diagnosing the onset of Increased participation by African-Americans in future research studies (NHGRI research capacity goal to increase the number of participants of diverse ancestry and populations in genome research)

1.2 Area of Emphasis Two: Genomic Research – To include racial and ethnic minority populations and other disparity populations in research on detecting and diagnosing the onset of rare and genetic diseases

1.2.1 Objective One: Advance our understanding of the natural history, underlying causes, and onset of genetic and rare diseases in racial and ethnic minority populations and other disparity populations.

1.2.1.1 Action Plan:

Racial and ethnic minority populations are significantly underrepresented in the research of rare genetic conditions. NHGRI's Intramural Research Program is developing ways in which to bolster the recruitment of individuals from racial and ethnic minority and other disparity populations so that they can participate in basic research efforts aimed at detecting and diagnosing genetic and rare diseases. Intramural research studies that are aggressively seeking to increase the diversity of racial, ethnic, and other disparity populations are:

1. The Attention Deficit Hyperactivity Disorder in Hispanics Study
2. The Severe Immunodeficiencies Study
3. The Genetic Study of Inherited Parkinson's Disease
4. The Identification of Genes Associated with Hermansky-Pudlak Syndrome (HPS) in Hispanic Population Study

1.2.1.2 Performance Measures:

- Develop procedures to encourage carrier screening, beginning with affected relatives
- Recruitment of at-risk family members for screening
- Detection of at-risk couples
- Genotyping, phenotyping, clinical follow-up with participants

1.2.2.3 Outcome Measures:

- Development of new or improved approaches for detecting and diagnosing the onset of genetic disorders in these individuals
- Better understanding of the natural history, causes, and treatment of genetic disorders in racial and ethnic populations
- The investigation of how genetic risk information is conveyed in clinical settings and how that information influences health strategies and behaviors

1.3 Area of Emphasis Three: Ethical, Legal, and Social Implications (ELSI) Research Exploring the Ethical, Legal, and Social Issues (ELSI) Related to Genetics and Health Disparities

Human genetic variation research, especially as it relates to risk factors for common, complex disorders, is leading to increased knowledge regarding how this variation may contribute to the health status of individuals. It is also leading to more knowledge about variation within and among populations, and how this variation may contribute to the aggregate health status of different racial and ethnic groups. The International HapMap Project is an international effort spearheaded by the NHGRI to create a haplotype map of the human genome, a resource that will make it possible to conduct disease gene association studies much more quickly and efficiently than ever before.

While the ultimate goal of studies aimed at relating human genetic variation to disease susceptibility 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. NHGRI, through its Ethical, Legal, and Social Implications (ELSI) Research Program, has implemented two separate initiatives to encourage additional research on the ELSI of genetic variation research for both individuals and diverse population groups.

This Objective addresses one of the representative public comments: “Increase numbers of health disparities populations studied by the NIH.” We have developed studies to not only include but to also increase the numbers of participants from health disparities populations.

1.3.1 Objective One: Study the Ethical, Legal, and Social Implications of Genetic Variation Research for Individuals and Diverse Racial and Ethnic Groups (FY2002-FY2005)

1.3.1.1 Action Plan:

In FY 2002, the ELSI Research Program released a second RFA for studies of the ethical, legal, and social implications of genetic variation research for individuals and diverse racial and ethnic groups. This RFA was designed to solicit proposals for research that further addresses the issues outlined above, building on research funded from 1999-2002. Five new studies were funded under this RFA in FY 2003, most of which are relevant to the issue of health disparities. These new studies are part of a growing consortium of projects sponsored by the ELSI Research Program which directly address these issues. Currently, 26 projects are included in the consortium.

This Objective addresses one of the six major themes in the public comments: “Expand the scope of research to include cultural, psychological, behavioral, social, gender, environmental, and biological factors appearing unique to race.”

1.3.1.2 Performance Measures:

- Increased number and quality of applications received in response to the 2002 initiative over those received in response to the FY 1999 initiative
- Continue to support a consortium of investigators from supported studies that will meet once a year 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 relevant publications.

1.3.1.3 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

1.3.2 Objective Two: Study Hereditary Hemochromatosis and Iron Overload Disease in Diverse Populations

1.3.2.1 Action Plan:

Iron overload is a common disorder affecting between 1 in 200 – 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 mutations in the HFE gene. The genetic contributions to hh in non-Caucasian populations are not entirely 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 widespread or population-based screening were addressed, especially in non-Caucasian populations.

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. 101,168 adults, of whom 44% are Caucasian, 27% African-American, 13% Asian, 13% Hispanic, 0.7% Pacific Islander, 0.6% Native American and 2% mixed or unidentified by self-identified race/ethnicity have been recruited and screened for these disorders during the course of this five-year study. Data are beginning to be analyzed. The results of this research will enable investigators to answer the important questions identified above, particularly for individuals from minority communities.

This Objective addresses one of the six major themes in the public comments: “Expand the scope of research to include cultural, psychological, behavioral, social, gender, environmental, and biological factors appearing unique to race.”

1.3.2.2 Performance Measures:

- Efforts will be made to assure that research participants from all backgrounds are retained to participate in this study so that comprehensive clinical exams can be carried out to better understand this disorder in individuals, particularly those from minority populations
- In FY 2004-2005, data from this study will be analyzed, with special emphasis on understanding the findings from minority populations
- In FY 2003-2006, scientific publications will report the progress and results of the research, particularly as they relate to iron overload disorders and minority communities

1.3.2.3 Outcome Measures:

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

1.3.3 Objective Three: Develop ELSI Research on Minority Participation in Genetic Research

1.3.3.1 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) have produced (and continue to produce) a unique set of data that will allow investigators in bioethics and the social and behavioral sciences to examine the factors causing African-Americans to consent or refuse to participate in genetic research. Special emphasis will be placed on access to information, informed consent, community attitudes toward genetic research; emphasis will also be placed on the development of 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 increase information available to investigators that will help them to design future genetic research in a way that will more successfully involve minority communities.

This Objective addresses a representative comment to: “Ensure that all communications with the health disparities populations and their subgroups address their needs and perspectives.” NHGRI is interested in supporting programs to increase understanding of factors around participation in genetic studies.

1.3.3.2 Performance Measures:

- Number of collaborations started with minority investigators
- Inclusion of minorities or medically underserved populations as research participants
- Published research

1.3.3.3 Outcome Measures:

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

2.0 AREAS OF EMPHASIS IN RESEARCH CAPACITY

Realizing the promise of genomics-based approaches to biomedical research depends, in large part, on the ability to train a diverse cadre of scientists in both genomic and ELSI aspects of human genome research. The challenges involved in accomplishing this are formidable. Scientists from diverse populations are severely underrepresented in genetics and genomics research and genomic medicine. Ethnicity data from other genetics professional societies reflect similar levels of underrepresentation. To attract individuals from diverse 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.

NHGRI has developed a series of new training initiatives targeted towards both individuals from diverse communities and HBCU's. In late 2001, NHGRI, with approval of its National Advisory Council, implemented its Minority Action Plan. This Plan established an Institute-wide process aimed at increasing the number of individuals from underrepresented groups participating in genomics and ELSI research. The goals for this area of emphasis are: to increase the diversity of investigators participating in genomic research; to increase the diversity of students trained in genomic research approaches; and to expose a greater diversity of students and faculty to genomic research approaches.

This Objective, and others detailed below, addresses one of the six major themes: “Improve research infrastructure at minority academic institutions.” A representative public comment read, “NIH must address the scarcity of qualified minority investigators by ensuring support for underrepresented minorities at every level of the pipeline.”

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

The Centers of Excellence in Genomic Science (CEGS) program will be the centerpiece of NHGRI-supported research in the future. One of the essential features of the CEGS will be their emphasis on the training of new investigators. Given this, these centers have the potential to become ideal sites for the training of individuals from underrepresented minority groups. Each CEGS is required to propose a plan describing what specific training activities it will implement and how it will recruit the relevant trainees. NHGRI staff will encourage the CEGS, as a group, to generate a range of training opportunities focused on underrepresented minorities.

2.1.1.2 Performance Measures:

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

2.1.1.3 Outcome Measures:

- Increased number of minorities trained and proceeding to their next career development stage in the sciences

This Objective addresses such public comments as: “NIH must address the scarcity of qualified minority investigators by ensuring support for underrepresented minorities at every level of the pipeline. Sponsor programs to mentor educate and provide grant support to minorities who pursue health careers opportunities and promote partnerships between minority-serving and research institutions.”

2.1.2 Objective Two: Support Training Opportunities at Centers of Excellence in ELSI Research

2.1.2.1 Action Plan: Centers of Excellence in ELSI Research (CEERs), which will be funded in the summer of 2004, will be the centerpiece of NHGRI ELSI multidisciplinary research in the future. Each Center is required to propose a detailed plan for training new

investigators. As part of this plan, each applicant must include a specific plan to recruit and encourage

The new Centers of Excellence in ELSI Research members of underrepresented minority communities to develop as independent ELSI investigators. This new program has the potential of significantly increasing the number of racial and ethnic minority researchers and researchers from other disparity populations. The CEERS also has the potential of developing a critical mass of investigators focused on the ethical, legal and social implications of genomics research at minority-serving institutions.

2.1.2.2 Performance Measures:

- Number, quality, and innovative nature of programs implemented for increasing the number of minorities exposed to genomics and ELSI research
- Number of minorities participating in these programs

2.1.2.3 Outcome Measures:

- Increased number of minorities trained and proceeding onto careers in ELSI research

2.1.3 Objective Three: Implement a New Program Called “NHGRI Health Disparities Visiting Faculty Program”

2.1.3.1 Action Plan:

Starting in FY 2003-2004, NHGRI established the NHGRI Health Disparities Visiting Faculty Program. The NHGRI Health Disparities Visiting Faculty Program provides researchers focused on genomics and health disparities with the opportunity to spend a 6 to 12 month period at NHGRI. Visiting faculty will work with senior or associate investigators in NHGRI laboratories located in Bethesda, Md. or Baltimore. Visiting faculty will have the opportunity to learn new technologies, develop research collaborations, or conduct sabbatical research. Basic and social science researchers may access NHGRI's laboratories, core facilities, clinics and training programs for study in any area of human genetic disease including the ethical, legal, and social implications of such research. Researchers are expected to share their skills and experience upon return to their home institutions and applications will be evaluated based on this criterion.

This Objective addresses several representative public comments. NHGRI has developed partnerships allowing visiting faculty from diverse and underrepresented institutions to train in the area of genomics and health disparities.

2.1.3.2 Performance Measures:

- Increased number of research projects addressing health disparities in the Division of Intramural Research, NHGRI
- Increased number of visiting minority faculty
- Increased number of publications relevant to issues of health disparities and genomics

2.1.3.3 Outcome Measures:

- Increased understanding of health disparities
- Increased number of publications by minorities as authors and a concomitant increase in understanding of health disparities to meet NHGRI research capacity goals of 1) increased numbers of participants of diverse ancestry and populations in genome research; 2) expansion of opportunities in research training and career development for investigators from diverse and underrepresented communities; and 3) an increase in the number of researchers conducting genomic and health disparities

2.1.4 Objective Four: Continue to Support and Develop the “Current Topics in Genomics Research Short Course” for Faculty and Students at institutions with significantly underserved populations.

The annual Short Course for Faculty is designed to update faculty from diverse institutions on the latest developments in genetic technology, medical genetics, gene therapy, and ethics. The objective of the course is to empower the faculty and build human resources at their home institutions. As part of the Short Course, each faculty member will develop course curricula to use in her or his own courses. The course also assists attendees in incorporating this information into classroom teaching so as to cultivate a diverse population of students interests in genomic research. It also offers information on careers in genetics and grant-writing skills to the participants. All participants spend time visiting NHGRI laboratories in order to experience the latest technologies that are being applied to genetics research first-hand.

Each successful Short Course applicant is asked to select one promising student from the faculty member’s institution to attend the Genome Scholars Program. This program parallels the Short Course and offers a close-up view of careers in genomic research while providing an enhanced mentoring experience. In 2004, 16 faculty members and 16 students will participate in the program.

This Objective addresses several representative public comments. NHGRI has developed partnerships allowing visiting faculty from minority institutions to train in the area of genomics and health disparities.

2.1.4.2 Performance Measures:

- Number of applicants
- Feedback from evaluation forms
- New grantees that participated in the Short Course
- Number of genome scholars that participate in NIH training programs
- Development and use of curriculum

2.1.4.3 Outcome Measures:

- Integration of genomics into curriculum at institutions with a significant numbers from underserved populations.
- Integration of genomics Short Course information into curricula at with a significant numbers from underserved populations.
- Increased the number of students from diverse backgrounds who pursue genomics because of their involvement in the Short Course

2.1.5 Objective Five: Through Minority Supplement Program, Expand the Opportunities for Minority Students and Faculty to Participate in Genomics and ELSI Research by Supporting Relevant Research Experiences and Attendance at Workshops and Conferences

2.1.5.1 Action Plan:

NHGRI, using a NIH-wide mechanism intended to increase the number of individuals from groups underrepresented in biomedical science, has several short- and long-term programs to support minority students and faculty who wish to know more about or pursue genomics and ELSI research. Available through the NRSA pre-doctoral fellowship program and supplemented by NHGRI grants, the program provides an opportunity for individuals to conduct intensive research related to genomics and ELSI. Participants are also encouraged to attend workshops and conferences related to genomics or ELSI. Opportunities are also available for Minority Access to Research Careers Program (MARC) students interested in genomics to be supported by NHGRI under NIGMS MARC initiatives.

This objective takes into consideration the public comments. NHGRI has developed an opportunity for students and faculty to gain exposure into genomic careers.

2.1.5.2 Performance Measures:

- Increased number of students and faculty from diverse populations being trained in genomics and ELSI research
- Fact sheets and a brochure developed for distribution at conferences attended by underrepresented minorities, such as the Society for the Advancement of Chicanos and Native Americans in Science (SACNAS) National Conference, and the Annual Biomedical Research Conference for Minority Students (ABRCMS)
- Plenary sessions or workshops sponsored at professional/scientific meetings that target underrepresented minorities; these will discuss the advances in genomics and the available research and training opportunities
- Meetings attended that are of specific interest to underrepresented minorities
- Web page developed to highlight the Institute's minority activities
- The importance of underrepresented minorities participating in genomics and ELSI research will be emphasized with grantees during the review of their annual research progress

2.1.5.3 Outcomes Measures:

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

2.1.6 Objective Six: Increase the Number of Minorities Participating in the T32 Training Grant Program

NHGRI currently supports 11 T32 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%.

2.1.6.1 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.

This Objective addresses public comments. NHGRI has developed an opportunity for students and faculty to gain exposure into genomic careers.

2.1.6.2 Performance Measures:

- A brochure with information about NHGRI's supported T32 training grant programs was 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 and ABRCMS, or where active minority committees exist, such as the American Society of Cell Biology, and professional/scientific meetings

2.1.6.3 Outcomes Measures:

- Increased number of underrepresented minorities enrolled on T32 training grants

2.1.7 Objective Seven: Increase the Number of Summer Internship Opportunities for Minority Students

2.1.7.1 Action Plan:

NHGRI hosts summer internships in intramural laboratories. While only a limited number of summer students can be hosted in DIR laboratories, the fraction of minority students availing themselves of this opportunity has been relatively low.

NHGRI/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. These efforts include the development of printed marketing materials specifically written for minority students. Research Investigators giving seminars at HBCUs and institutions with a diverse population of students also take time at the end of their seminars to inform students in the audience about research opportunities available at both NHGRI and NIH and encourage interested students to contact the NIH Office of Education for further information.

This objective addresses public comments. NHGRI has developed an opportunity for students and faculty to gain exposure into research careers.

2.1.7.2 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

2.1.7.3 Outcome Measures:

- **Increased number of minority students in the summer internship program going on to research careers**

2.1.8 Objective Eight: Increase the Number of ELSI Researchers from Minority Communities and Minority-Serving Institutions

Very few ELSI researchers who are currently funded (or who were funded in the past) are from minority communities. The ELSI program has been increasing its efforts to reach out to potential 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 institutions with significant numbers of students from diverse communities through the application process.

This objective addresses public comments: “NIH must continue to promote the development of inter-institutional partnerships between research-intensive institutions and minority-serving institutions.” NHGRI has developed a program to increase researchers from minority communities.

2.1.8.2 Performance Measures:

- Efforts to provide information about funding opportunities to members of minority communities
- 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

2.1.8.3 Outcome Measures:

- Increased number of ELSI researchers from minority communities and institutions with diverse populations of students

2.1.9 Objective Nine: Expand the Capacity of Institutions to Conduct Research on Sickle Cell Disease

2.1.9.1 Action Plan:

The National Institutes of Health hosted a conference entitled "New Directions for Sickle Cell Therapy in the Genome Era" in Bethesda, Maryland in November 2003. The aim of this conference was to consider how new genomics tools and techniques might be applied both to more fully-understand the biology of sickle cell disease and to develop more effective therapeutic and preventive strategies for the disease. Following the meeting, staff from eight NIH Institutes and Centers formed the Trans-NIH Sickle Cell Disease Therapies Working Group. The working group developed a number of initiatives for consideration for implementation over the course of the next several years. NHGRI took the lead in developing several of these initiatives: facilitating the discovery of small molecules that can be used in basic biological and biomedical studies of sickle cell disease and to translate basic research findings into novel therapeutics in sickle cell disease; training sickle cell researchers in genomics; and establishing a LISTSERV to apprise the research community of funding opportunities relevant to sickle cell disease.

2.1.9.2 Performance Measures:

- Establish and maintain the sickle cell LISTSERV
- Communicate its existence to the community
- Develop a RFA for training
- Develop a RFA for small molecules

2.1.9.3 Outcome Measures:

- LISTSERV operational
- Number of interested researchers subscribed to the sickle cell LISTSERV and number of posts made to the list
- RFAs published
- Training grant awards made in 2005
- Research grants awarded for making small molecules assays robust in 2005

3.0 AREAS OF EMPHASIS IN COMMUNITY OUTREACH, INFORMATION DISSEMINATION, AND PUBLIC HEALTH EDUCATION

The broader inclusion of racial and ethnic minorities and other disparity populations in biomedical research is imperative if it is to be applied to improve the health of all citizens. Inclusion is of particular importance to 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. Generating public policy positions aimed at minimizing the potential misuses of genetic information must also be addressed in a timely fashion. To be ultimately successful, the improvements in research and health care must reach everyone. This success will depend on participation of individuals from *all* communities.

We hope to take advantage of the momentum resulting from increased visibility of genomics and genetics research with the completion of the Human Genome Project in 2003. This is an opportune time to redouble our community outreach, information dissemination, and public education efforts in order to encourage individuals from racial and ethnic minority populations and other disparity populations to actively participate in the genetics revolution – as scientists, clinicians, research participants, and *active contributors* in deliberations of ethics and public policy.

This objective addresses public comments: “NIH must continue to promote the development of inter-institutional partnerships between research-intensive institutions and minority-serving institutions.” NHGRI has developed a program to increase researchers from minority communities. NHGRI has reviewed the public comments and recognizes the themes to “...use racially, culturally sensitive and appropriate communication while strengthening the capacity of minority communities.” To address this concern, we have taken significant measures in information diffusion and dissemination.

3.1.1 Objective One: Facilitate the incorporation of science-based information into the curricula of K-12 schools

3.1.1.1 Action Plan:

NHGRI has developed resources to facilitate the incorporation of science-based information into high school curricula, with targeted dissemination to racial and ethnic minority and other disparity communities. “The Human Genome Project - Exploring our Molecular Selves,” a grade 9-12 education resource, was originally produced by NHGRI in February 2001. In 2002, NHGRI reformatted this education kit, making the materials available for download on its Web site (<http://www.genome.gov>), thus providing expanded access to students and teachers across the country. A second revised edition of the kit will be developed in FY 2005. This kit provides unprecedented access to cutting-edge information about the Human Genome Project and genomics, geared specifically for use by life science educators. The kit has been widely distributed and enthusiastically received.

NHGRI is creating a multimedia interactive education resource to communicate the excitement and opportunities in genomic research and genomic careers to high school and college students, including students from racial and ethnic minority populations and other disparity populations. The goal of this DVD is to expose students to genetics and genomics careers, with a specific aim to expose racial and ethnic minority, rural and low SES students. This DVD would fill a critical need for resources in the NHGRI public education program, which is a key component of our strategy to increase research capacity.

The genomic careers multimedia education product will be in development throughout FY 2004-2005 and is targeted for distribution and promotion in FY 2005-2006. Evaluation of the product will be conducted in FY 2006-2007.

This objective addresses public comments: “Sponsor programs to mentor, educate and provide grant support to minorities who pursue health careers opportunities and promote partnerships between minority-serving and research institutions.” NHGRI has developed an opportunity for students and faculty to gain exposure to genetic and genomic concepts and into genomic research careers.

3.1.1.2 Performance Measures:

- Development of a product that captures the excitement of genomics research and transmits that excitement to its target audience
- Dissemination to high schools and colleges, including targeted dissemination plan to schools with significant percentage of racial and ethnic minority populations and other disparity populations
- Dissemination to racial and ethnic minority schools and communities
- Dissemination to low SES and rural schools and communities
- Increased number of community interactions with the education resources

3.1.1.3 Outcome Measures:

- Evaluation of the quality and effectiveness of the resource
- Increased number of students interested in genomic careers, particularly students from racial and ethnic minority and disparity populations
- Increased utilization of the education kit by racial and ethnic minority schools and communities
- Increased utilization of education kit by low SES and rural schools and communities
- Increased knowledge of genomics research in minority schools and communities

3.1.2 Objective Two: Establish a Vital Presence at Disparity Populations-Focused Conferences

NHGRI continues to enhance its presence at conferences targeted toward minorities by hosting a visible and inviting exhibit booth, organizing genomics symposia, compiling an attractive brochure that highlights training opportunities in genomics, and hosting roundtables or hospitality suites so that students and faculty have a chance to speak with staff from the various training programs of NHGRI. NHGRI will actively seek out opportunities to give presentations to groups such as the Society for Advancement of Chicanos and Native Americans in Science, the National Association of Hispanic Nurses, the Association of American Indian Physicians, the National Medical Association, the National Hispanic Medical Association, the National Black Nurses Association, the Intercultural Cancer Coalition, and other organizations serving the communities appropriate for ELSI research.

NHGRI developed an outreach exhibit plan in FY 2004 to increase and document its outreach activities. NHGRI will continue to increase its level of participation in conferences targeted to disparity populations.

This objective addresses public comments: "...ensure that all communications with health disparities populations and their subgroups addresses their needs and perspectives." NHGRI has developed a strategy to inform and educate disparate populations.

3.1.2.2 Performance Measures:

- Increased number of conferences with an active NHGRI exhibit booth

- Increased number of interactions with racial and ethnic minority students and faculty, students and faculty at minority-serving institutions, students from low SES background and rural communities
- Increased number of genomic symposia/presentations at targeted conferences
- Increased number of students and faculty that participate in NHGRI training activities because of information provided by NHGRI staff at conferences

3.1.2.3 Outcome Measures:

- Increased knowledge of NHGRI training and research programs by racial and ethnic minority students
- Increased participation in NHGRI training and research programs by racial and ethnic minority students
- Increased knowledge of NHGRI training and research programs by low SES and rural students
- Increased participation in NHGRI training and research programs by low SES and rural students

3.1.3 Objective Three: Continue to Support and Develop an Annual Conference about the Genomics for the Public

3.1.3.1 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 participate. NHGRI will make every effort to increase minority participation in the NHGRI Annual Genomics 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, providing them with an opportunity to become informed about all aspects of human genome research. Beginning in 2005, Consumer Day will be held in a different region of the country each year to broaden the national scope and reach of this activity.

This objective addresses public comments: “ensure that all communications with health disparities populations and their subgroups addresses their needs and perspectives.” NHGRI has developed a strategy to inform and educate disparate populations.

NHGRI, in collaboration with community-based partners, are developing models of engaging racial and ethnic minority and other disparity communities to participate in the Consumer Day program. These community based models include collaborations with faith-based and other community-based organizations.

3.1.3.2 Performance Measures:

- Attendance by members of racial and ethnic minority communities at the Annual Conference
- Attendance by racial and ethnic minority students from local schools at the Annual Conference
- Attendance by members of low SES and rural communities at the Annual Conference
- Attendance by low SES and rural students at the Annual Conference
- Conference evaluation forms from attendees to measure impact of these conferences
- Development of additional collaborations with NHGRI Centers and grantees to reach communities around the country

3.1.3.3 Outcome Measures:

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

3.1.4 Objective Four: Genetic and Rare Disease Information Center

3.1.4.1 Action Plan:

NHGRI and the Office of Rare Disease (ORD) established the Genetic and Rare Diseases Information Center (GARD) to provide accurate, reliable, and current information on genetic and rare diseases to patients, families, health care professionals, and biomedical researchers. NHGRI and ORD will develop an outreach plan to provide increased access to the Center by individuals within racial and ethnic minority populations and other disparity populations. NHGRI and ORD will implement the plan in FY 2005-2008.

This objective addresses public comments: “ensure that all communications with health disparities populations and their subgroups addresses their needs and perspectives.” NHGRI has developed a strategy to inform and educate disparate populations. A public comment read, “...distribute NIH resources equitably across all population groups.” NHGRI has developed a partnership to all populations informed about rare genetics issues.

3.1.4.2 Performance Measures:

- Awareness by racial and ethnic minority communities of GARD
- Awareness by low SES and rural communities of GARD
- Promotion of GARD services at professional and public conferences
- Provide the latest research-based information about genetic and rare disease to health professionals serving racial and ethnic minority communities
- Provide the latest research-based information about genetic and rare diseases to health professionals serving low SES and rural communities

3.1.4.3 Outcome Measures:

- Increased utilization of the Information Center by racial and ethnic minority patients and families
- Increased utilization of the Information Center by low SES and rural patients and families
- Increased utilization of the Information Center by health care professionals serving racial and ethnic minority patients
- Increased utilization of the Information Center by health care professionals serving low SES and rural patients and families

3.1.5 Objective Five: Family History Public Education Initiative

3.1.5.1 Action Plan:

Family History Day is an initiative whose objectives are to increase the American public's awareness of the importance of family history, to provide accessible methods for easily obtaining an accurate family history, and to increase health professionals' use of family histories in disease prevention and health promotion. A Web-based tool is being designed that will enable users to chart a pedigree and to print out a copy for themselves and their physician; the tool will be made available in FY 2004.

Thanksgiving Day 2004 will serve as the inaugural National Family History Day. In a phased approach, this year's initiative will focus on increasing awareness of the importance of family history and will lay a foundation to make this an effective annual national campaign. A component of the Annual National Family History Day Initiative is to reach the diverse communities of the country. Community- and faith-based organizations will be engaged in the initiative, helping to develop models for reaching racial and ethnic minority and disparity communities.

This objective addresses public comments: "Ensure that all communications with health disparities populations and their subgroups addresses their needs and perspectives." NHGRI has developed a strategy to inform and educate disparate populations.

3.1.5.2 Performance Measures:

- Increased utilization by racial and ethnic minority patients and families of Web-based or printed family history tool
- Increased utilization by low SES and rural patients and families
- Increased utilization by health care professionals serving racial and ethnic of Web-based or printed family history tool
- Increased utilization by health care professionals of the patient's family history in care of patient within racial and ethnic minority populations and other disparity populations

3.1.5.3 Outcome Measures:

- Evaluation of the utilization of the Web by the public
- Increased awareness of family medical history as a preventative health tool
- Increased overall preventative health

3.1.6 Objective Six: DNA Day Public Education Programs

3.1.6.1 Action Plan:

NHGRI seeks to facilitate the incorporation of science-based information about genetics and genomics in a high school outreach and information dissemination program. NHGRI, with partners the American Society of Human Genetics, the Genetic Alliance, and the National Society of Genetic Counselors, embarked in 2004 on a national "DNA Day"- a high school outreach program that took place on April 30, 2004. The DNA Day program began in 2003, coinciding with the completion of human genome sequencing; this was seen as an opportunity to have a "teachable moment," given the large amount of press attention given to the Human Genome Project at that time.

Faculty, staff and postdoctoral fellows from all three NHGRI divisions visited high schools across the country to talk to students about genomic science and how it will influence the future of biology, medicine, and society. The DNA Day Ambassador program provides an opportunity for students, specifically including those from underrepresented and disadvantaged populations, to get excited about careers in science and increase their awareness of how rapid changes in the areas of genetics and genomics will affect their everyday lives. A complete list of schools visited in 2004 can be found at <http://www.nhgri.nih.gov/11511624>. Ambassadors have been encouraged to stay in contact with the schools they visited so that the teachers at these schools have a "personal contact" through which they can continue to obtain up-to-date information about genomics.

The goals of this project were to: 1) excite students about careers in genetics and genomics by exposing them to genetic concepts, genomic researchers, and health professionals; 2) update and/or introduce educators to NHGRI's education resources; 3) foster interactions

between NHGRI investigators, trainees, and staff with the broader public community through schools; and 4) expand NHGRI's collaborations with consumer organizations and professional associations.

3.1.6.2 Performance Measures:

- Awareness by racial and ethnic minority students of genetics and genomic concepts
- Awareness by low SES and rural communities of genetics and genomic concepts
- Number of schools with that student body is predominantly racial and ethnic minority or other disparity populations that participate in the DNA Day program

This objective addresses public comments: "Ensure that all communications with health disparities populations and their subgroups addresses their needs and perspectives." NHGRI has developed a strategy to inform and educate disparate populations.

3.1.6.3 Outcome Measures:

- Evaluation of the teachers, students and the ambassadors of the benefits of the DNA program
- Evaluation of teachers, students and the ambassadors of the impact of the DNA Day program on the student body
- Increased knowledge and excitement about the Human Genome Project
- Increased interaction of NHGRI investigators with the community

3.1.7 Objective Seven: Community Based Outreach and Dissemination Demonstration Projects

3.1.7.1 Action Plan:

The success of genomics-based tools to improve the health of all depends in part on communicating advances in genomic and ELSI research to all communities and on empowering 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: 1) to foster development of policy, education, and outreach activities relevant to increasing the involvement of diverse communities in all aspects of genetic research; 2) to reduce health disparities; and 3) to integrate new genetic technologies into health care in underserved communities. NHGRI will develop models of partnership with community-based organizations to foster dialogue on genomics and biomedical research and the dissemination of latest research-based information on genetic disease to communities.

This objective addresses public comments: “Ensure that all communications with health disparities populations and their subgroups addresses their needs and perspectives.” NHGRI has developed a strategy to inform and educate disparate populations. A public comment read, “NIH must support research and promote information dissemination through partnership with academic institutions and communities so that research results will reach communities.”

3.1.7.2 Performance Measures:

- Develop model community engagement programs in racial and ethnic minority communities
- Develop model community engagement programs in low SES and rural communities

3.1.7.3 Outcome Measures:

- Evaluation of the programs and the communities knowledge about genomics
- Increased involvement of diverse communities in subjects pertaining to genetic research
- Increased integration of genetic technologies into underserved communities