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The Human Genome: What's 'Race' Got To Do With It? The Implications of Human Genome Research for Minority Health Issues: The Benefits of Genetic Research in Improving Health and Health Care

delivered by

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LECTURE

Introduction

Let me begin by acknowledging the Nova Scotian Black Community for this unprecedented International Symposium on *"Racism and the Black World Response"*. The timeliness of this Symposium is observed in both its commemoration of the *United Nations' Third Decade Against Racism and the UN International Year of Mobilization Against Racism, Racial Discrimination, Xenophobia, and Related Intolerance.* I am truly honored to participate in this historic gathering and sincerely thank Professor Esmeralda Thornhill and other organizers of this Symposium for the invitation and opportunity to come and share thoughts with you this evening on the

implications of human genome research for minority health issues: the potential benefits of genetic research in improving health and health care.

Human Genome Knowledge Defining History

For anyone who does not yet know it, let me state upfront that 2001 is not just another year on the calendar. While it is indeed the beginning of a new millennium, it also marks the beginning of a new two thousand year cycle of knowledge in the earth. Knowledge that challenges each of us, individually and collectively, to think about how we define ourselves, and the relevance of this definition to the health of our bodies, the integrity of our community, and the stability of our world. The year 2001 is a defining moment in human history, characterized by a new knowledge base for meeting the challenges of disease and unraveling the mysteries of life. This is the year when the first draft of a completed sequence of the 3.1 billion parts of the human genome was published in the scientific literature and made known to the public. A time when humankind is gazing inquiringly upon the magnificence of its own genetic blueprint and beginning to exploit the knowledge encoded in the genome to find the genetic causes of diseases and unmask the science of gene genealogy-- the ultimate biological measure of human and population history, and molecular evolution.

During this time I would like to challenge you to think with me about the knowledge gained from the Human Genome Project (HGP) and from research on human genome sequence variation, and the relevance of this knowledge to our understanding of ethnic differences and disparities in disease susceptibilities and expression. The objectives of my presentation are threefold:

1) To define what the human genome is;
2) To describe how new knowledge gained from the HGP is transforming biology and biomedical science in the 21st century, and
3) To discuss why African people must be informed and involved in the genome revolution.

Knowledge gained from the HGP and research on DNA sequence variation reveals, perhaps like no other advance of western science, the inextricable link of biology and identity to health and disease.

Significance of Genomic Knowledge

The significance of the HGP for humankind ultimately must be examined in the light of its impact on public health. Because society will be transformed by the exploitation of new knowledge gained from explorations of the human genome, it is imperative that such knowledge be communicated to *all* communities to ensure democratization of the benefits of genetic

research for disease prevention, health promotion, and the elimination of disparities in health between majority and minority populations. For, informed, educated, and activist communities are the stakeholders who will ultimately determine if the billions of dollars expended in sequencing the human genome will usher in the promises of a new era of human liberation from the tyranny of chronic debilitating diseases and a greater understanding and respect for the place of humankind in the order of Life.

The timeliness of this International Symposium on *Racism and the Black World Response* is underscored by its focus on the history and reality of "how we define ourselves" as critical to any actions geared towards Black Community empowerment, capacity-building, and collective strategizing, The organizers of this Symposium have shown great insight in planning a program that recognizes the sobering implications of how knowledge gained from human genome science is impacting society in general and minority health issues in particular. Because so much of the current attention in human genome research is focused on the technological feats of sequencing, I especially appreciate the recognition and attention that this Symposium directs to community education on genetics research and its relevance to the health of Black people. As stunning and awesome as is the sequencing of 3 billion nucleotides in the human genome, community education in genome science is the most compelling and potentially the most transformative challenge to 21st century science and society. It is the *expectations* of and *accountability* of informed and educated stakeholder communities that will serve as both catalysts and road maps – They will determine whether the billions of research dollars expended in sequencing the human genome will herald an age of freedom from disease, disability, and death due to complex diseases, such as diabetes, cancer, and heart disease - OR whether the HGP will be remembered as the most expensive, self-promoting and exploitative venture in the history of Western science and technology.

Importance and Potential of the Human Genome

In addressing the topic of *Collective Strategizing* from a health perspective, I want to focus your attention on the potential benefits of genetic research in improving health and health care by underscoring the links between human genome research, self- knowledge, and racial minority health issues. In my opinion, the formal beginning of the United States Human Genome Project in 1990 represents a defining moment in Western science and human history. My reasons for making this bold affirmation are two-fold. First, the HGP, perhaps like no other leading edge of Western science, **challenges scientists to expand, and make more inclusive, the *context* or measure of humanity, in order to better understand the *content* of human biology**. Second, with regard to the human story or history, sequence data emerging from the HGP directly challenges prevailing constructs of human populations which traditionally partition humanity into bounded ethnic and/or racial groups. At this, the dawn of the 21st century, the HGP has extended the probing of biomedical science to the ultimate level of biological identity, that is unique DNA sequence variation. Moreover, exploration of the human genome has introduced new prospects for understanding molecular processes underlying disease and disease

susceptibility. Attention is now focused on DNA sequence variation and the challenges inherent in distinguishing sequence variation of biomedical interest (i.e., mutations) from the tremendous amount of natural variation (i.e., polymorphisms) of biological interest. Because natural variation in the human genome is the ultimate measure of biological relationship, it is a determinant of individual, family, population, and human identity. Studies on DNA sequence analysis already show that populations differ in the frequency of both mutations of biomedical interest and polymorphisms of biological interests. Thus, the population that is used as the reference to map mutations and polymorphisms becomes very relevant to the identification and proper application of information emerging from DNA sequence variation.

Let me direct your attention for a moment to the historic and evolutionary significance of completing the human genome sequence for humankind. I say historic, because completing the human genome sequence marks this moment in history as the ceremonial beginning of a new era of biomedical science, genomic medicine and the paradigm shift in biology to DNA-sequence-based diagnosis and prevention of disease. I say evolutionary because, with the completion of the human genome sequence, comes a new knowledge-base for biology and biomedical science. A knowledge base that is as old as the origins of humanity and yet as new as the most recent gene discovery. This knowledge base connects all life and has the capacity to transform our most basic concepts of self and human identity. Thus, sequencing the human genome is not only applicable to biomedical science in the identification of genes of both clinical and non-clinical interests, but also to more fundamental questions of human identity and integrity. One of the major implications of human genome research for racial minority health issues is its potential impact on how we define ourselves.

The human genome is unique in that it is the fundamental level and expression of life. It contains all the information required for the construction, assembly, and operation of the human body. Thus it is both a type of "*Manufacturer's Handbook"* and "*Owner's Manual"*. Because the genome exists in all nucleated cells of the body and the body is encoded in every genome, the genome and the body are inextricably one. The human genome is not only the most complex information system known to mankind, but also an unfathomable communications system, in which the four-lettered DNA sequence code is translated into "flesh" and dwells among us, as us. As the "Book of Life", the genome contains the record of every human being that ever was and is and will ever be. It encodes both the laws of Life and of Creation. The knowledge contained therein is indeed unique. One wonders if science is not the instrument for revelation of this knowledge in our time. The sequencing of the human genome has shifted the orientation of human knowledge from the outside appearance of things to the inside reality of life expressed at the molecular and cellular, or microcosmic levels. The HGP project also shifts the definition of humankind from a population-based to a DNA sequence-based science. The characterization of DNA sequence variation in the human genome is not only applicable to human biology, but also to human identity.

The most salient feature of human identity at the sequence level is variation. Human genome sequence variation dispels the myth of a majority. At the level of the genome, every genome is unique; **the norm is variation not uniformity**, **and the norm is best defined as a range of**

variation. As medicine becomes increasingly more customized and tailor-made to measure, and designer-driven, a more refined definition of humanity and the individual will be required. It remains to be determined how DNA sequence-based knowledge of self and group identity will impact minority health issues. Biological anthropologists and population geneticists are already mining the rich resources of natural variation in the human genome to reconstruct population history. Although no known biological product is encoded by much of the natural variation in the genome, it is nonetheless transmitted from generation to generation through the genome much like the genes that code for proteins, the functional products of genes. Natural variation in DNA sequences is a very rich source of information on family and population history. The results of research in areas of molecular evolution on gene genealogies in human populations are challenging old ways of characterizing racial and ethnic groups, which traditionally have been based on phenotypic, linguistic, and/or cultural differences. Anthropologists have estimated that less than 1% of the total gene pool code for the phenotypic characteristics widely used in the western world to classify human populations. In other words, the genes for physical appearance, such as skin color, eye color, and hair texture are an extremely small fraction of the approximately 3 billion nucleotides that make-up the human genome. If DNA sequence-based biology is to be science driven, then scientists and the general public must better understand the public health significance of the vastly greater stretches of unexpressed DNA sequence variation. The genome era is also forcing a paradigm shift in biology. A shift that is not just a change, but rather a transformation in the way we define ourselves; the way we see ourselves; the way we see our world, and how we see ourselves in relationship to our world.

The Human Genome and Racial Minority Communities

As an African American woman trained in the discipline of Human Genetics, I am aware of the narrow, Eurocentric context in which much of human biology has heretofore been cast, and of the history of exploitation and exclusionary practices of Western science and biomedical research practised in Communities of Color. As part of the African American Community and a member of the Academy, I am convinced that the active participation of Communities of Color in general, and African people in particular, will be a major factor in whether knowledge gained from sequencing the human genome will contribute to widening the gap or eliminating national and global health disparities between socio-economically and politically advantaged and disadvantaged people. I am therefore committed to realizing the benefits of genetics in public health and to the importance of connecting research, education, practice and community.

While the alleviation of disease is the prime motivation for the HGP, my emphasis is on the implications of human genome research for racial minority health issues. If health is recognized as “more than” the absence of disease, then human genome research must go beyond a focus on disease to a greater understanding of the “more than” implicit in health. Because an individual’s concept of identity frames his or her reality, I hypothesize that the study of disease in individuals and between groups cannot be uncoupled from an individual’s and/or group’s concept of identity. Studies of DNA sequence variation challenge the truth of perceived and believed links between human identity and biology inculcated in Western culture. The social implications of uncoupling

individual and group identity from biology are enormous. It remains to be determined whether attention to emerging knowledge of DNA sequence variation may effect a paradigm shift in our understanding of individual and group identity. Knowledge gained from the human genome is unique in its capacity to unshackle or liberate science and society from constructs of biology, that are themselves predicated on a very limited and incomplete picture of the human identity. If sickness and disease result from an incomplete and distorted concept of human identity – then it remains to be determined whether wholeness and health would ensue after a more comprehensive construct of biology based on more complete knowledge of the human genome.

It is now, that the world is challenged to close the gap in health status among different segments of humanity. **There is indeed much to be learned, when we set about /start viewing human variation as a gift and not an aberration**. It is noteworthy that knowledge of population differences in profiles of variation in the human genome, coupled with knowledge of the broader spectrum of natural variation in the genome of African peoples, underscores the critical importance of the population reference in human genome research. Understanding the "language of life" encoded in DNA sequence variation is indeed the brave new frontier of whole genome science, genomic medicine, and public health. Genomic research in African peoples offer unique resources for understanding human genome variation. **Because the African American genome brings together the depth and breadth of DNA sequence variation resident in the oldest African populations with more evolutionarily recent profiles of variation found by admixture with Europeans and Native Americans, the African American genome is perhaps the most comprehensive single population resource for exploiting DNA sequence variation in the genetic dissection of complex diseases.**

Genomic Research in the African Diaspora

Let me move towards my conclusion by commenting briefly on Genomic Research in the African Diaspora, commonly referred to as GRAD. This is a concept for human genome research initially proposed by investigators at Howard University contemporaneously with the beginning of the First Five Years (FY 1991-1995) of the U. S. Human Genome Project. GRAD focuses on DNA sequence variation as the foundation of biology and biomedical science. The long-range goal of GRAD is to improve the health status of African people through research on DNA sequence variation and the application of knowledge gained from research to better understand the biomedical significance of gene-based differences already known to exist among populations in immune response to organ transplants, susceptibility to diseases such as diabetes, sensitivity to drugs, cancer, and the influence of environment on health. GRAD provides the research foundation for the newly formed National Human Genome Center at Howard University in Washington, DC, USA. The purpose of this National Center is to bring multicultural perspectives and resources to an understanding of human genome variation and its implications for health and life. Our mission is knowledge driven-- to explore the science of and teach the knowledge about DNA sequence variation in the causality, treatment, and prevention of diseases common in African Americans and other peoples of the African Diaspora. By addressing population variability in the human genome, the NHGC brings a depth perception to

the linear perspective of human biology. The implications of this more enriched construct of human biology in improving health and health care will be determined, not so much *by the science*, as *by the scientists*, and, not by scientists *in isolation* but *in Community*.

The National Human Genome Centre

The NHGC seeks to enhance the quality of life for Americans by improving the health status of ethnic groups and/or populations who bear a disproportionate burden of the disease in our racially stratified, hierarchical society. Research at the NHGC is designed to engage and empower individuals in community to probe questions about human identity and relationships in ways that explore genomic links between constructs and concepts of human identity and the expression of health and disease in individuals and communities. The ethical, legal, and social implications of decisions informed by such knowledge are enormous and will impact public policies at all levels of life. Research at the NHGC forces discussion of public policy issues on the proactive participation of individuals in promoting health of the body and community. Towards this end, the GRAD is an important beginning in bringing African and people of the African Diaspora from diverse disciplinary perspectives together in and with Community, to examine and assess plans geared to procuring comprehensive reference resources for collaborative research on human genome variation. Because African Diasporic populations include the depth and breadth of human DNA sequence variation, these populations provide the most comprehensive resources for exploiting DNA sequence variation in gene mapping and the genetic dissection of complex diseases.

Conclusion – A Scientific Revolution in Knowledge

Let me conclude by stating that humanity is in the midst of a scientific revolution in knowledge about human history and biology, knowledge that shifts the focus from the phenotype or external appearance of things, to the genotype, the internal view of life. One of the most sobering lessons of the human genome is that our knowledge of human biology and potential is distorted by extreme limitations. It is as if our expressed desires to heal our bodies, our Communities, and our world cannot be achieved at our present level and state of knowledge. We need a paradigm shift, a different system of explanations, a whole new way of thinking about who we are and our relationship to ourselves and to each other.

In some ways, our times are akin to the days when humankind thought that the Earth was the center of the universe. The truth of this thinking was confirmed by anyone on Earth who could look into the heavens and see the sun rise in the Eastern sky and make its way across the heavenly orb to settle down in the Western sky. This was plain for all to see, the sun revolved around the Earth and Earth was indeed the center of the universe. With developments in optics, powerful telescopes were made which increased humankind's capacity to peer into the heavens and study the movement of the sun and other objects in the sky brought into view with the telescope. Astronomers studying the movement of objects in the sky soon recognized inconsistencies in their calculations when the position of Earth was placed in the center of

objects in the sky. Galileo, a 16th century astronomer and physicist, is credited with using science to show that inconsistencies in the calculations of the movement of objects in the sky could be resolved if, instead of placing earth in the center, the sun is positioned in the middle with the Earth revolving around it. To say that Earth was not the center of the universe and the sun occupied this central place was, at that time, considered heresy by many in the Church, the Authorities on Truth.

Nonetheless, the facts supporting the heliocentric construct of the world held center stage until advances in space travel allowed humankind, once again, to extend the boundaries of our knowledge and we began to scan the outer limits of our solar system, with science now revealing new facts and data about the design of the world and raising questions about the place of humankind in it. It should be noted that a more complete knowledge of Earth's orbit around the sun was essential and fundamental to humankind's growth and unfolding capacity to plot trajectories that later allowed us to fulfill our dream of space travel from the Earth to the Moon. The increase in knowledge brought by travel to the outer limits of Earth's solar system once again challenged humankind's view of our solar system as the only sun in the heavens with its orbiting planets and their orbiting moons. A more complete and enhanced knowledge of the heavens revealed that Earth's solar system is just one of many galaxies all with their suns, planets; and it moves in orderly, clockwork fashion through space. We now know that not only does our galaxy not exist alone in the universe, but rather, that at the extremes of observation, it appears as if new galaxies are being continually born and that the universe is alive and unlimited!

The impact of knowledge that our galaxy is not alone in the heavens and that humankind may not be alone in the universe has challenged our thinking about who we are as humans and the relationship of how we define ourselves to the stability of our world. In the last millennium, humankind explored space, the limits of our perceived outer world. Now, at the dawn of a new millennium and the complete sequencing of the human genome, humankind is poised on the thresholds, ready to explore and discover *the limits of our perceived inner world*. Representing the fundamental knowledge- base of biology (i.e., the science of life), could it be that such knowledge provides fresh perspective now for novel paradigm shifts in our knowledge of life and our place and relationship in it? Could it be that more complete knowledge about knowledge is required for humankind to fulfill our desires for health and healing of our body? Is life so constructed that there is a relationship between what we think about and what we create? If knowledge is structured in awareness, what relationship does our increasing awareness of our *outer* and *inner world* have to the desires of our hearts? The year 2001 is defined as the end of the beginning of a new era in science, ushered in by the revelation of more complete knowledge on the construction and operation of the human body. We can only wonder if such knowledge will provide the foundation for better understanding, health, and elimination of diseases of body and mind.

Consider for a moment if racism is cast or characterized as an infectious and deadly disease that kills oppressor and oppressed alike, a disease that is rooted in erroneous thinking and false beliefs about who and what is the true nature of humankind's inherited identity and inheritance….How then might racism be properly characterized as a "crime against humanity"? And, if crime is defined as a violation of the law, racism then is guilty of being an egregious violation of the Law of Life that causes oppressor and oppressed alike to live so far below the

Truth of identity that the crime itself is punishable by death. Can a more complete knowledge of human identity revealed from the inner perspective of the human genome change humankind's awareness and thinking about our body and its place in the Order of Life? Could it be that this International Symposium on *Racism and the Black World Response* is demonstrable of and fundamental to a new way of thinking and believing about who we are as humankind? Is the cure to racism rooted in this more complete knowledge of the Truth of human identity and potential? If sickness and disease are inextricably tied to disease and death, is healing and health also linked to knowledge and thinking that are in harmony with the Law of Life? Do we need a whole new paradigm shift in our understanding of the relationship of how we define ourselves to the health of our bodies, integrity of our Communities, and stability of our world? Will 2001 be the year that the mind of humankind was changed and humanity redefined itself in the Order of LIFE?