



# Addressing Health Disparities through Researching the Human Genome

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**Founder, President, and CEO**

**Whole Genome Science Foundation, Inc.**

**March 12, 2018**

**Clayton Hall Conference Center**

**University of Delaware**

# OBJECTIVES

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- **Define the relationship of Human Genome Research to Health Disparities.**
- **Relate the science of the human genome to the science of health disparities.**
- **Describe how addressing health disparities through researching the human genome is relevant to achieving the USA Public Health Service goals of “Healthy People 2020”.**

# Y2K Signs of the Times

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**MILLENNIUM**

*of the mind*

**CENTURY**

*of consciousness*

**DECADE**

*of discovery/destiny*

**DAY**

*of decision on*

**Human Identity and Purpose**



# Genome Knowledge Revolution

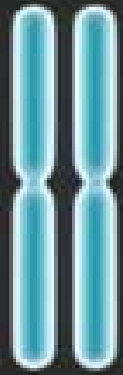
## Big Data to Knowledge (BD2K)

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2001

The beginning of a new cycle of  
**KNOWLEDGE** on the earth about **LIFE**  
and the **SCIENCE** of Human Identity  
and Population Diversity.

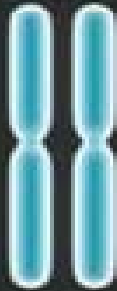
# The Human Genome



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XX

or



XY

# Prologue

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**The human genome forces us to become aware of how we define ourselves, and how ‘who we say we are’ governs our behavior, which is related to the health of our body, the integrity of our communities, and the stability of our world.**

# 3<sup>rd</sup> Millennial Public Health Service (PHS) Initiatives

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## **PHS ‘Healthy People 2010’**

1. The Human Genome Project (2003)
2. International Haplotype Mapping Project (2005)

## **PHS ‘Healthy People 2020’**

3. BRAIN Initiative (2013)
4. Precision Medicine Initiative (2015)
5. 21<sup>st</sup> Century Cures Act (2016)

# Healthy People 2020 Overarching Goals

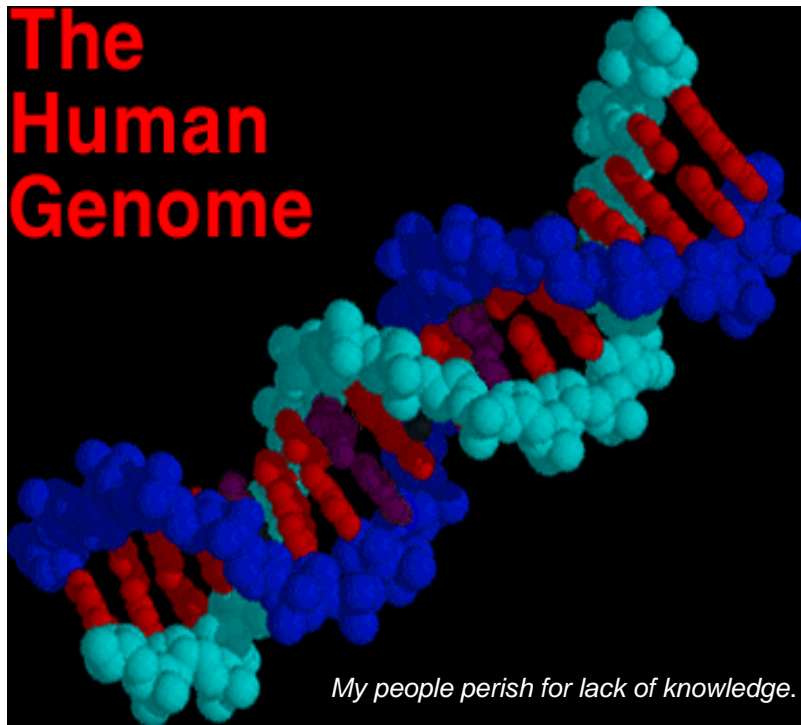
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1. Attain high-quality, longer lives free of preventable disease, disability, injury, and premature death.
2. Achieve health equity, eliminate disparities, and improve the health of all groups.
3. Create social and physical environments that promote good health for all.
4. Promote quality of life, healthy development, and healthy behaviors across all life stages.



# A Living Information and Communication System for LIFE in the 3<sup>rd</sup> Millennium

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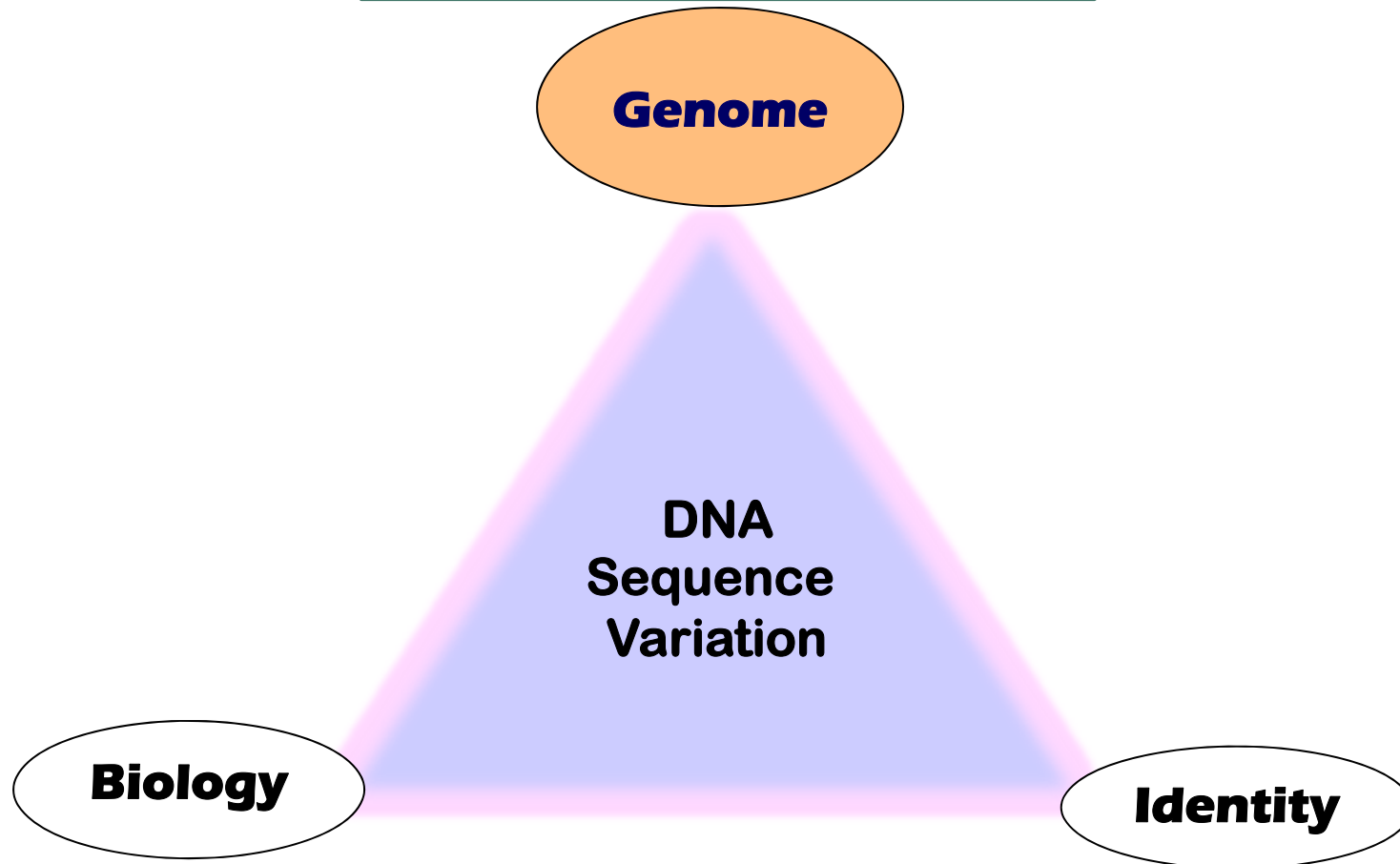


With completion of the human genome project, comes a new KNOWLEDGE System for biology, biomedical and the LIFE sciences- KNOWLEDGE as old as the origins of humanity itself, and as new as the most recent genome and 'omic' discoveries.

**Timeless Knowledge, Whose Time Has Come.**

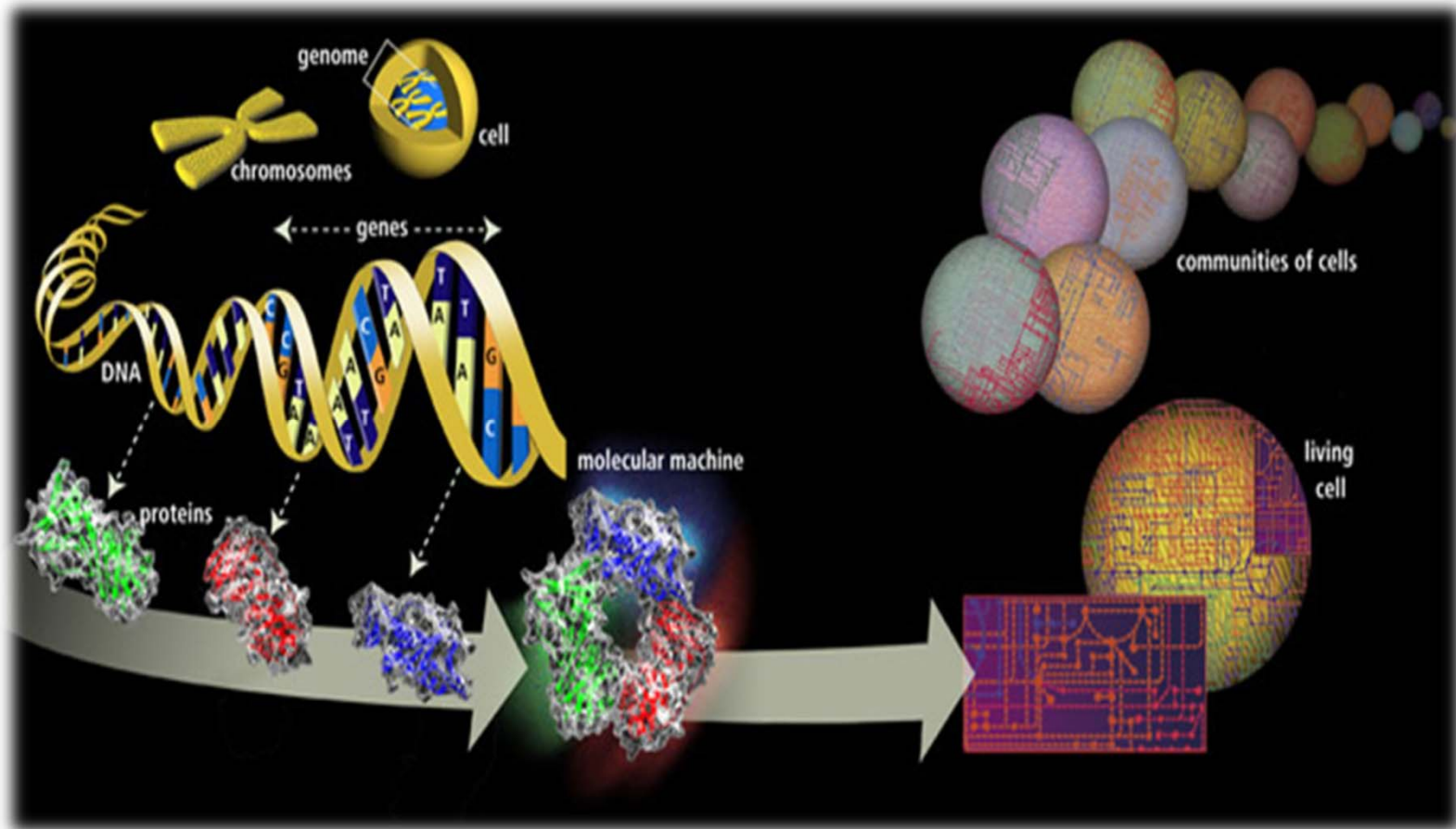
# The Human Genome

It's all about Life



*We use genome variation for gene and self discovery.*

# A Finished Human Genome Sequence from DNA to Life

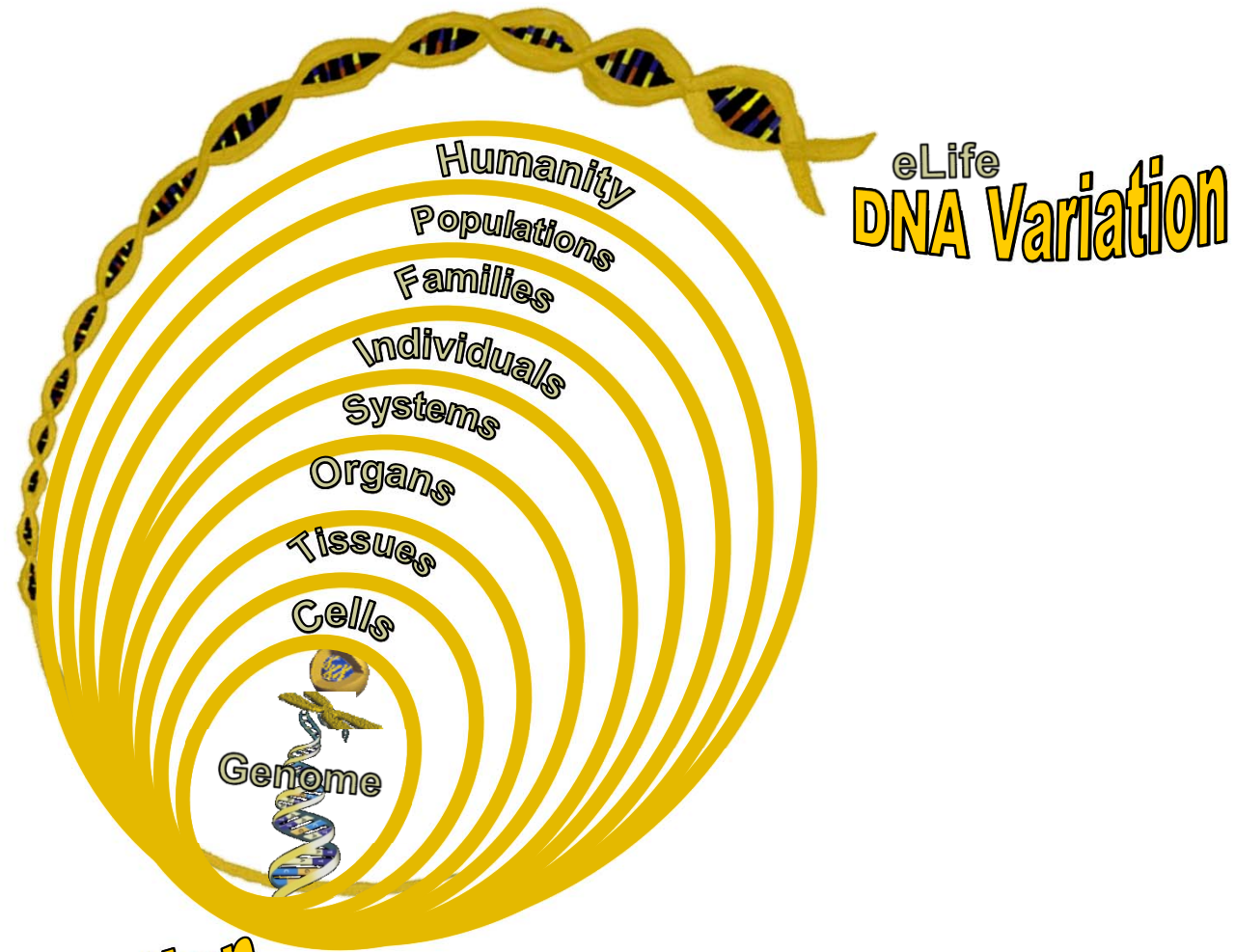


[http://www.ornl.gov/TechResources/Human\\_Genome/primer\\_pic.htm](http://www.ornl.gov/TechResources/Human_Genome/primer_pic.htm)

# Unlocking the Mysteries of Life

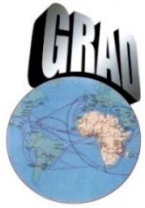
Biological Organization and Complexity

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DNA Variation



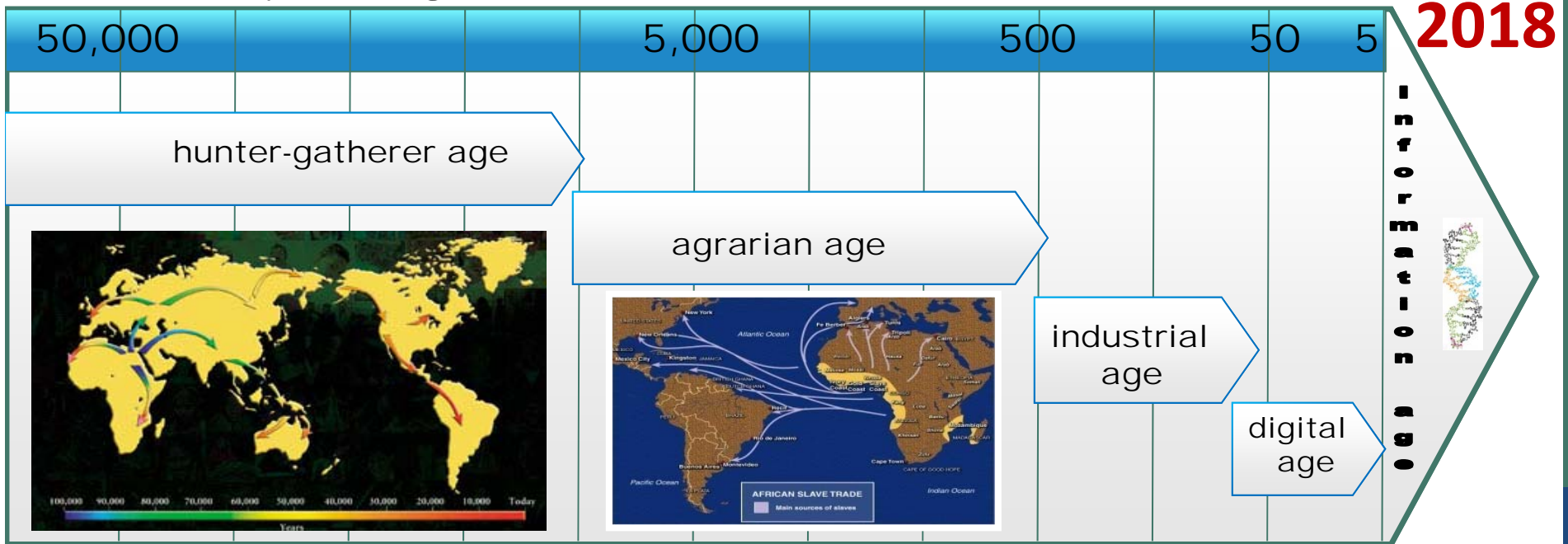


GENOMIC RESEARCH IN  
THE AFRICAN DIASPORA

# The Human Genome Project

The GENOME story of human origins, migrations, adaptations, transformation, and liberation, now unfolded in research on DNA sequence variation.

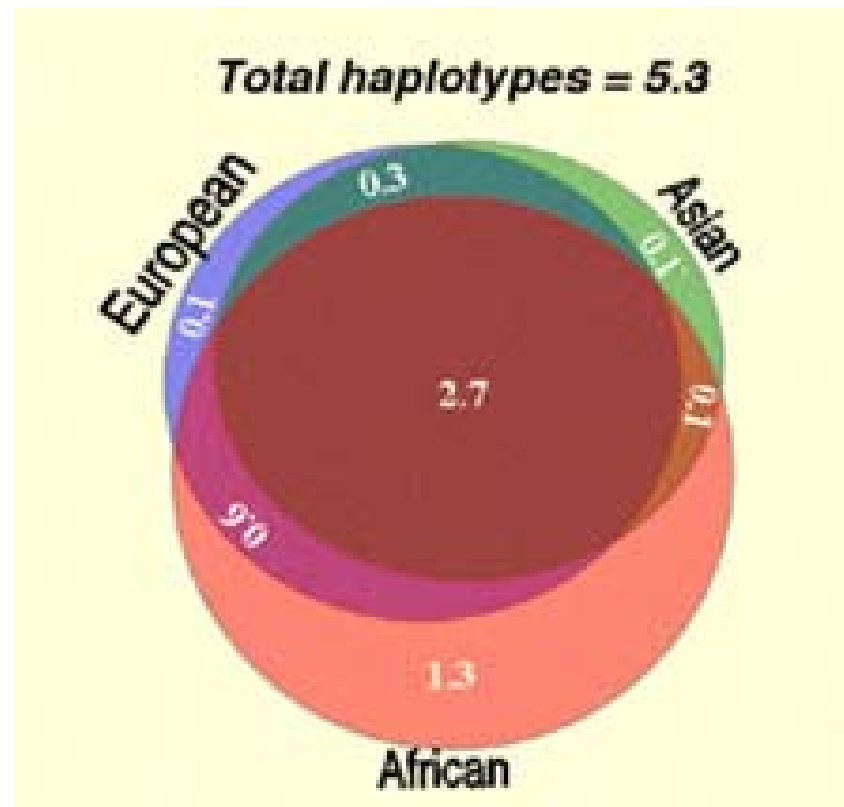
## Timeline (years ago)



At the end of all our exploring, we shall arrive where we began and know the place for the first time.

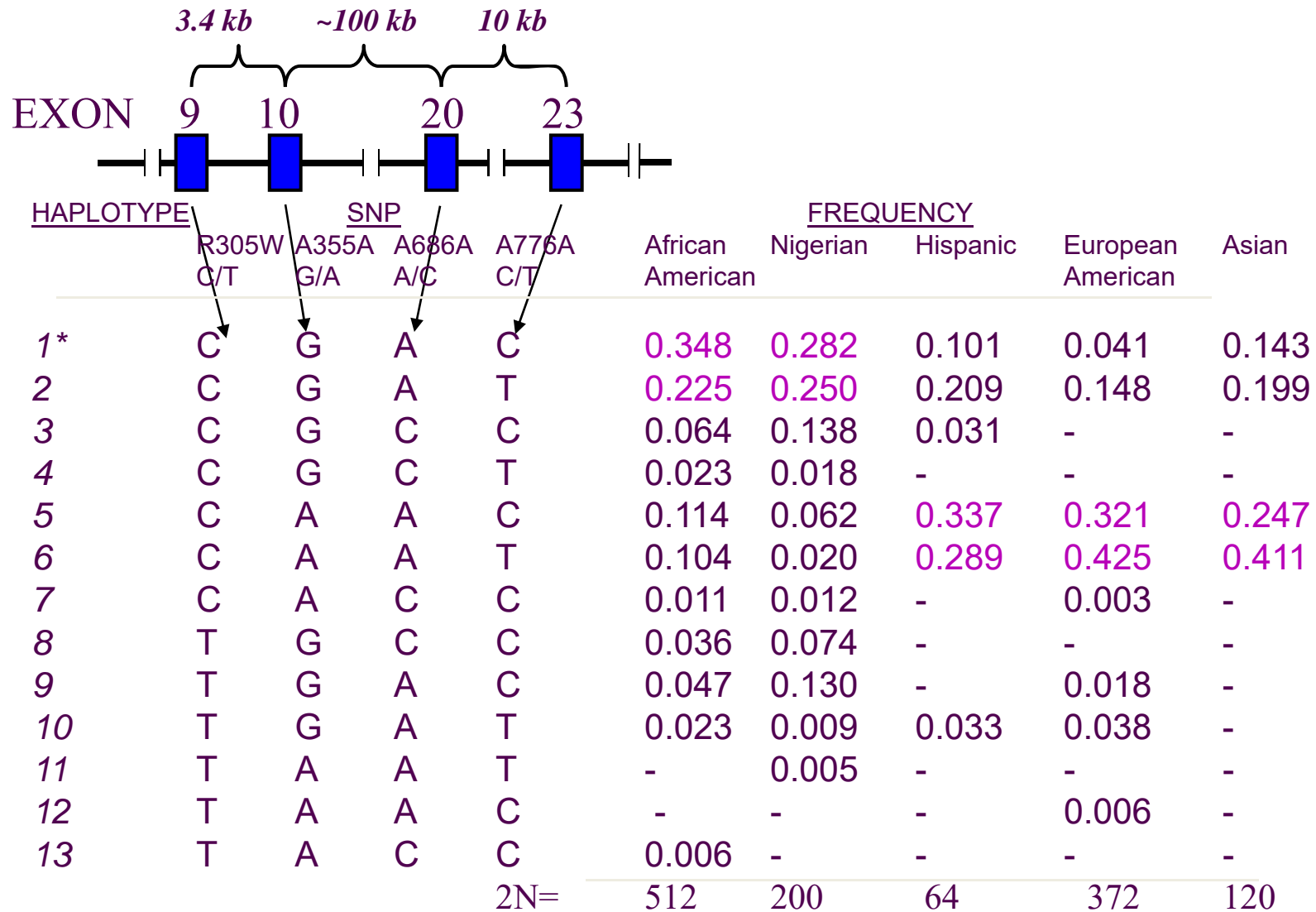
*T.S. Eliot*

# Genetic Variation in Diverse Populations



Gabriel, S. B. *et al.* The structure of haplotype blocks in the human genome. *Science* 296(5576):2225-9, 2002.

# P gene haplotype frequencies



# Individual and Population-based Genome Variation



In **2007**

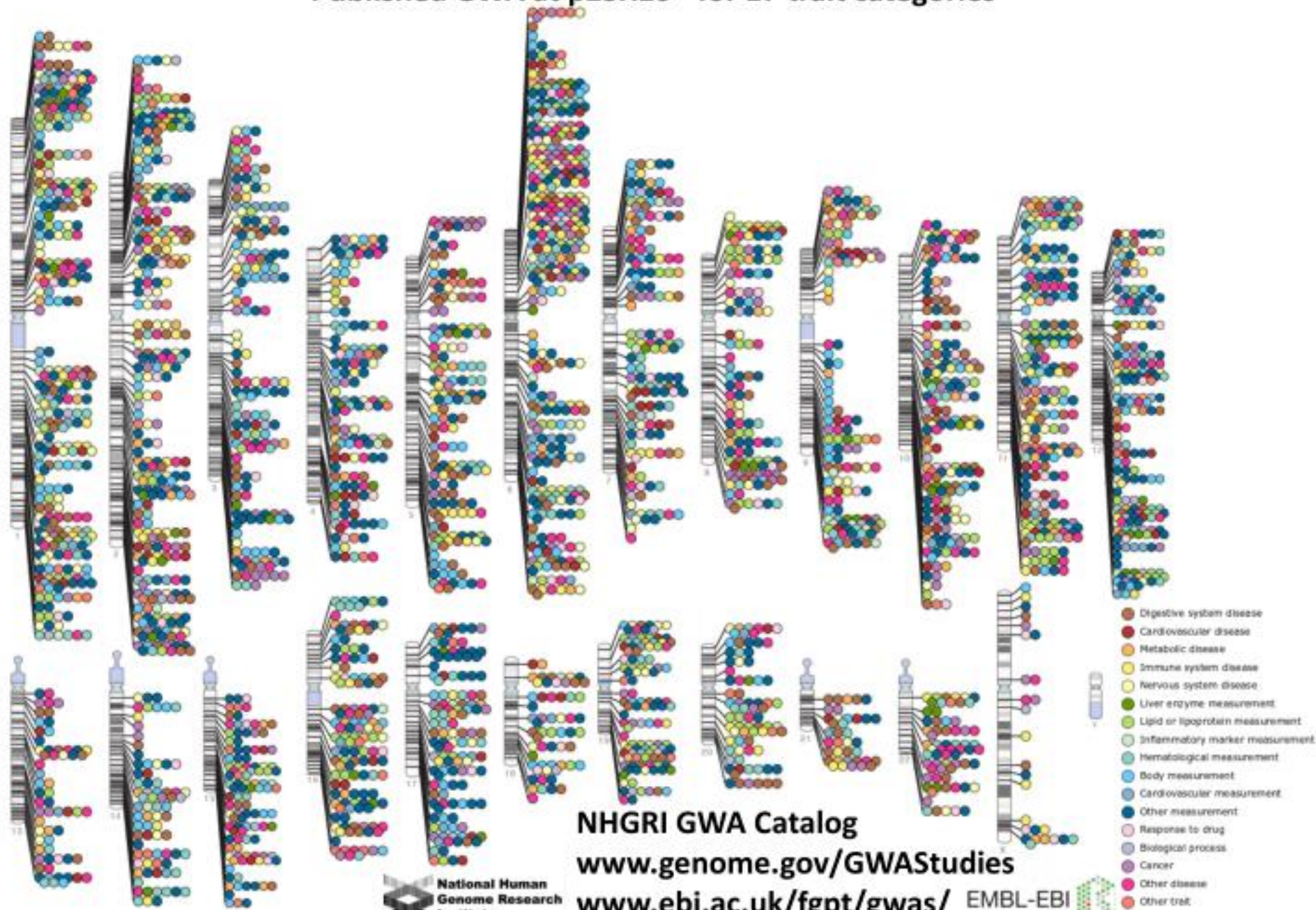
researchers came to appreciate the extent to which our genomes differ from person to person and the implications of this variation for deciphering the genetics of complex diseases and personal traits.



Science 21 December 2007 Vol. 318 no. 5858 pp. 1842-1843



Published Genome-Wide Associations through 12/2012  
 Published GWA at  $p \leq 5 \times 10^{-8}$  for 17 trait categories



NHGRI GWA Catalog

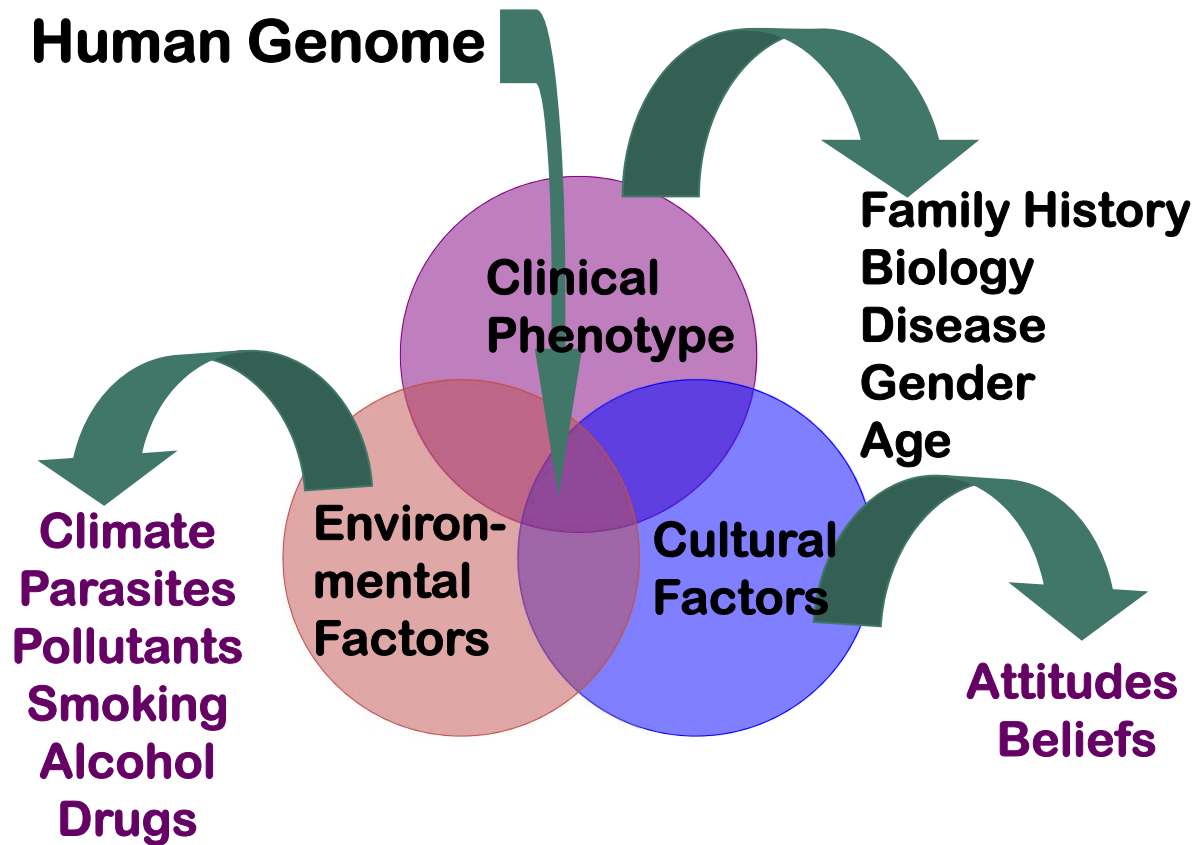
[www.genome.gov/GWASStudies](http://www.genome.gov/GWASStudies)

[www.ebi.ac.uk/fgpt/gwas/](http://www.ebi.ac.uk/fgpt/gwas/)

EMBL-EBI



# Human Genome Variation in Health Disparities



# Health Disparities<sup>1</sup>

Age-adjusted Death Rates by Race and Sex, United States, 1998

Diseases	Male (Per/100,000)		Female (Per/100,000)	
	White	Black	White	Black
Heart Disease	162	232	88	147
Cancer	144	208	104	129
Stroke	25	47	22	37
Diabetes Mellitus	14	29	11	29
Hypertension	2	8	2	7

<sup>1</sup>Source: Nat'l Vital Stat Rep. 2000:48(11);63-64)

# Chances of developing breast cancer for African American women

Age	African American	Nationwide	Caucasian
<b>30-34</b>	33.3	12.6	<b>23.6</b>
<b>40-44</b>	123.9	<b>60.3</b>	119.5
<b>50-54</b>	236.4	129.1	260.2
<b>60-64</b>	297.0	184.3	366.1
<b>70-74</b>	378.2	263.5	483.3
<b>80-84</b>	362.5	304.1	490.9

\*Rates are per 100,000

# Chances of dying from breast cancer for African American women

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Age	African American	Nationwide	Caucasian
<b>30-34</b>	<b>7.7</b>	<b>2.1</b>	<b>3.8</b>
<b>40-44</b>	<b>33.2</b>	<b>10.6</b>	<b>19.7</b>
<b>50-54</b>	<b>70.9</b>	<b>26.5</b>	<b>49.6</b>
<b>60-64</b>	<b>96.5</b>	<b>43.5</b>	<b>81.1</b>
<b>70-74</b>	<b>128.0</b>	<b>66.4</b>	<b>116.7</b>
<b>80-84</b>	<b>162.8</b>	<b>102.7</b>	<b>159.0</b>

\*Rates are per 100,000

\*\*The above information was taken from SEER data and can be found on the Internet

([www.seer.ims.nci.nih.gov/](http://www.seer.ims.nci.nih.gov/)).

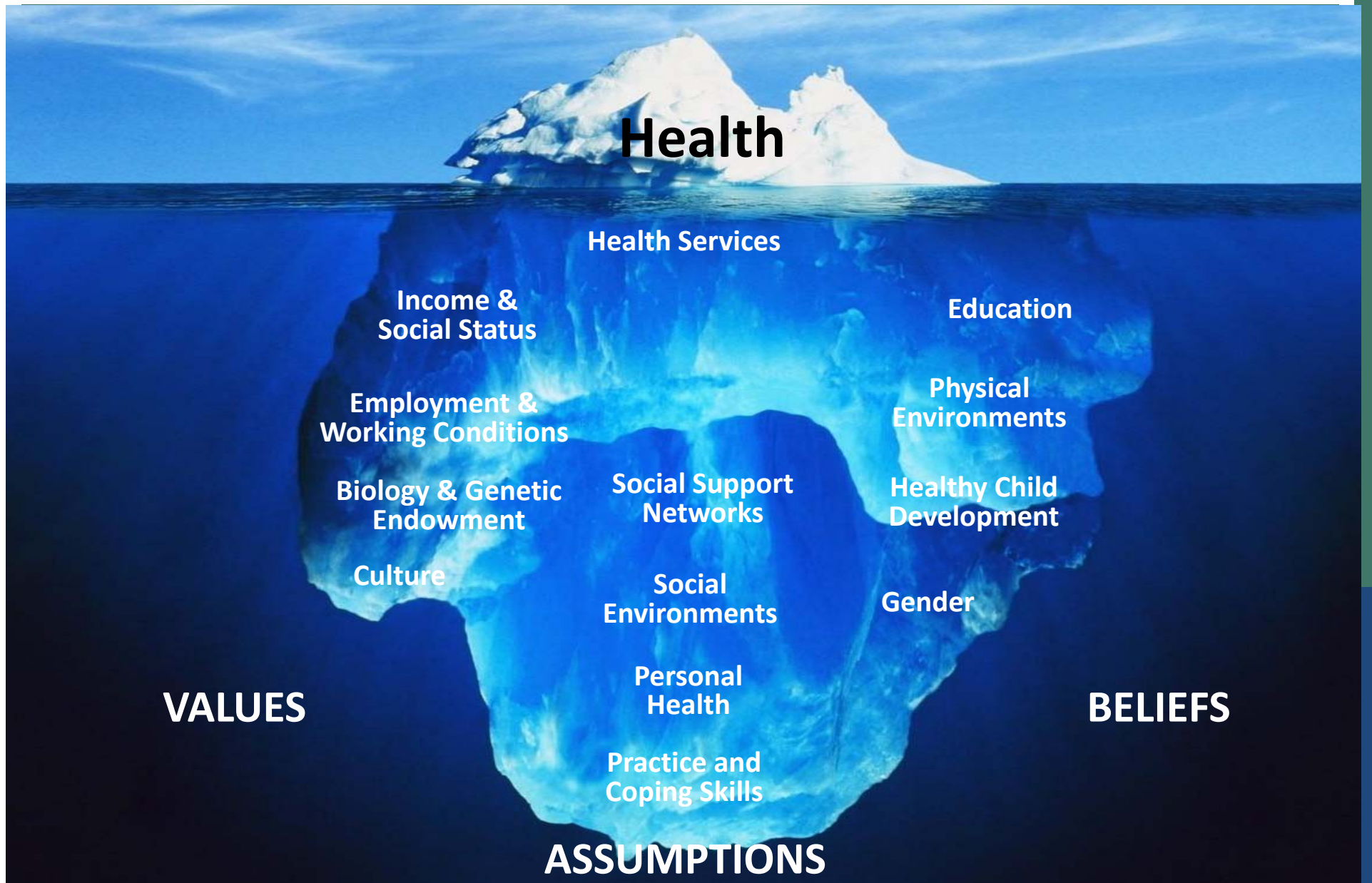
# Social Determinants of Health

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- The social determinants of health are the conditions in which people are born, grow, live, work and age. These circumstances are shaped by the distribution of money, power and resources at global, national and local levels.
- The social determinants of health are mostly responsible for health inequities - the unfair and avoidable differences in health status seen within and between countries.

[http://www.who.int/social\\_determinants/sdh\\_definition/en/](http://www.who.int/social_determinants/sdh_definition/en/)

# Can Social Determinants of Health Be Mapped to the Human Genome?



# A Major Determinant in Health Disparities is BEHAVIOR

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- What is the link between genes and behavior?
- How does our genotype interact with our environment to produce behavior?
- What methods are available to investigate these questions?
- What are the societal (social) and philosophical (ethical) implications of discovering such links?



# Clinical Genome Sequencing

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- The application of genomic information in individual health care is inevitable.
- Examples of how whole-exome or whole-genome information may be used in clinical diagnosis and decision making are no longer rare.
- Commercial and academic molecular laboratories all over the world are beginning to offer whole-exome or whole genome as a clinical genome service.

# BRAIN Initiative

Brain Research through Advancing Innovative Neurotechnologies

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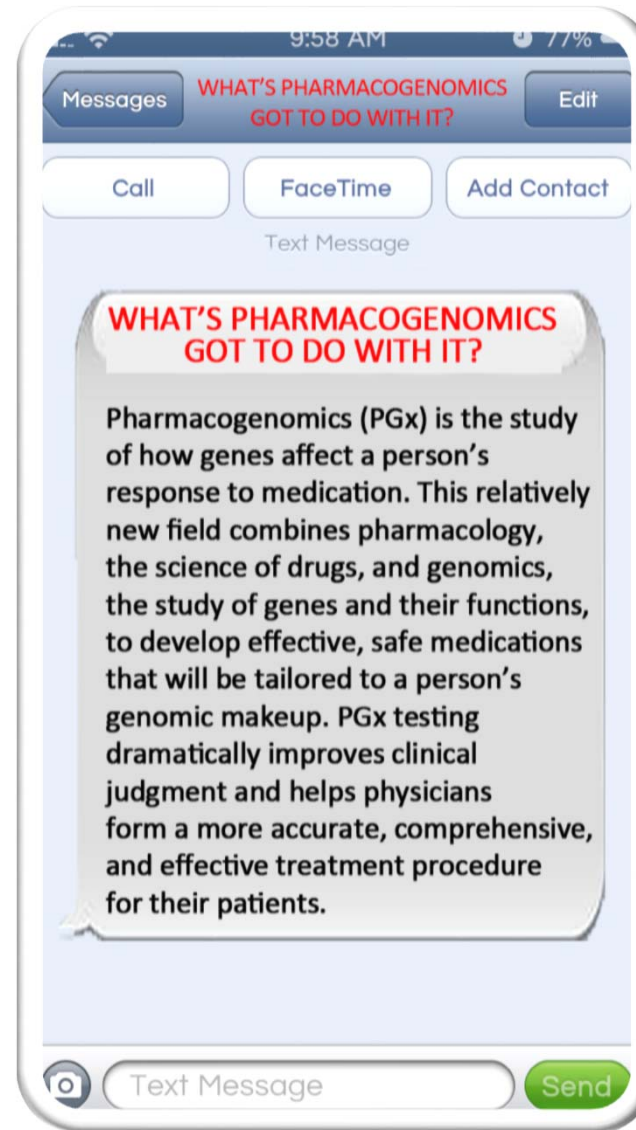
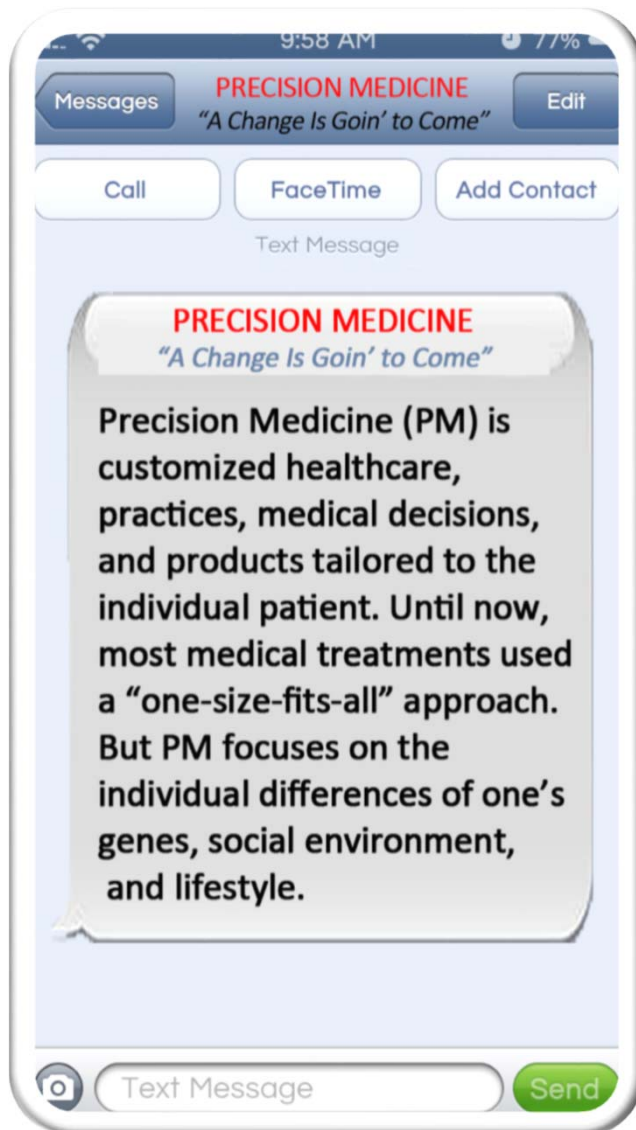
The BRAIN Initiative extends beyond the mapping of the brain and bridges scales that span from atoms to thoughts and **behavior**, linking what is known about single cells and subcellular activities in the brain to whole brain function leading to complex behavior.

# Precision Medicine

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**A**fter decades of research, we are poised to enter a new era of medical practice where detailed genetic and other molecular information about a patient's disease is routinely used to deploy effective, patient-specific remedies to treat it. We are about to enter the era of precision medicine.

# Precision Medicine and Pharmacogenomics



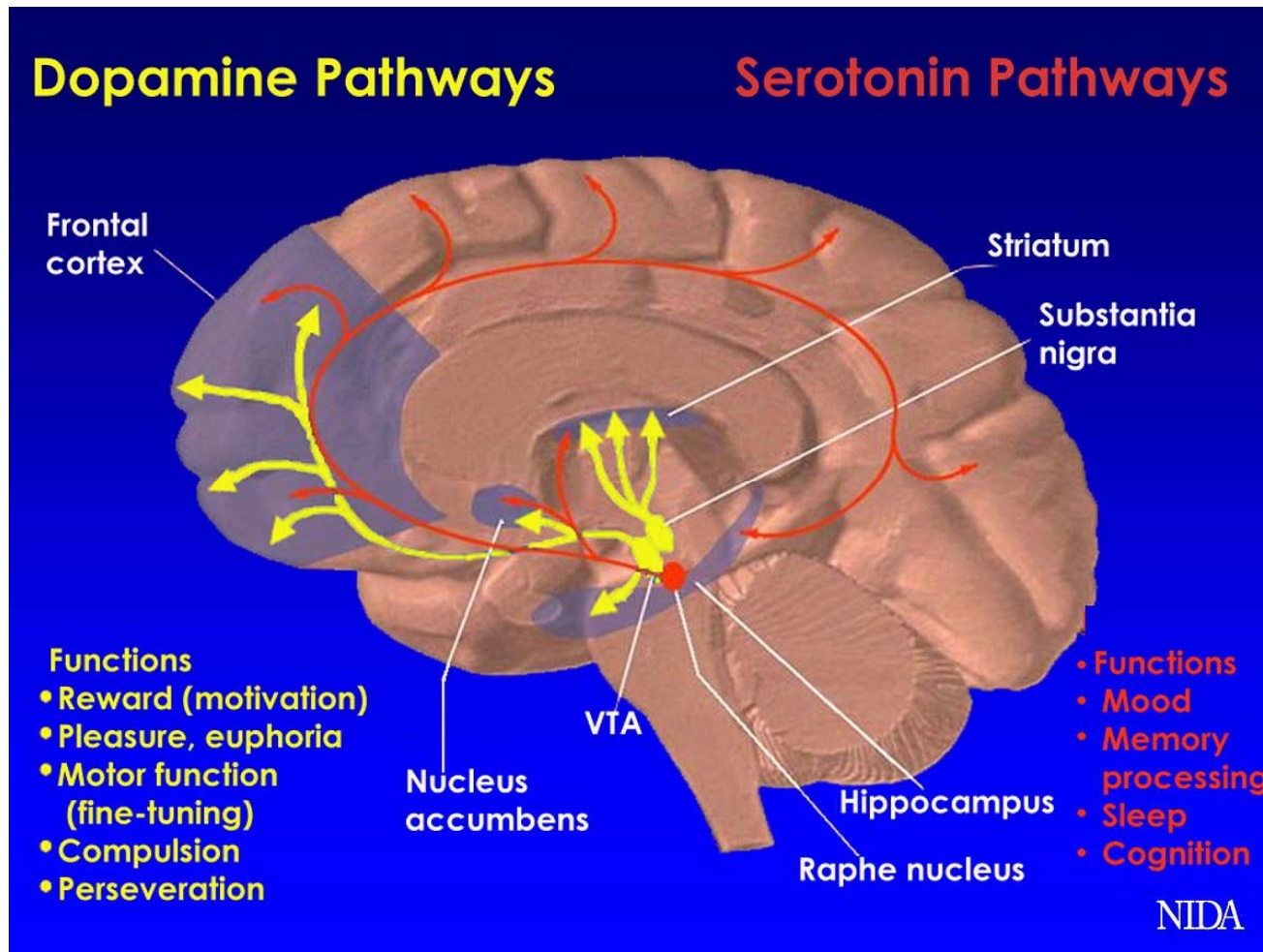
# Pharmacogenomics

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**To fulfill the promise of targeted interventions, clinical trials and observational epidemiologic studies are needed to assess:**

- prevalence of relevant genotypes in the population,
- how drug response varies among individuals with different genotypes,
- whether and to what degree environmental factors interact with genetic factors to influence drug response.

# Reward Circuitry of the Brain

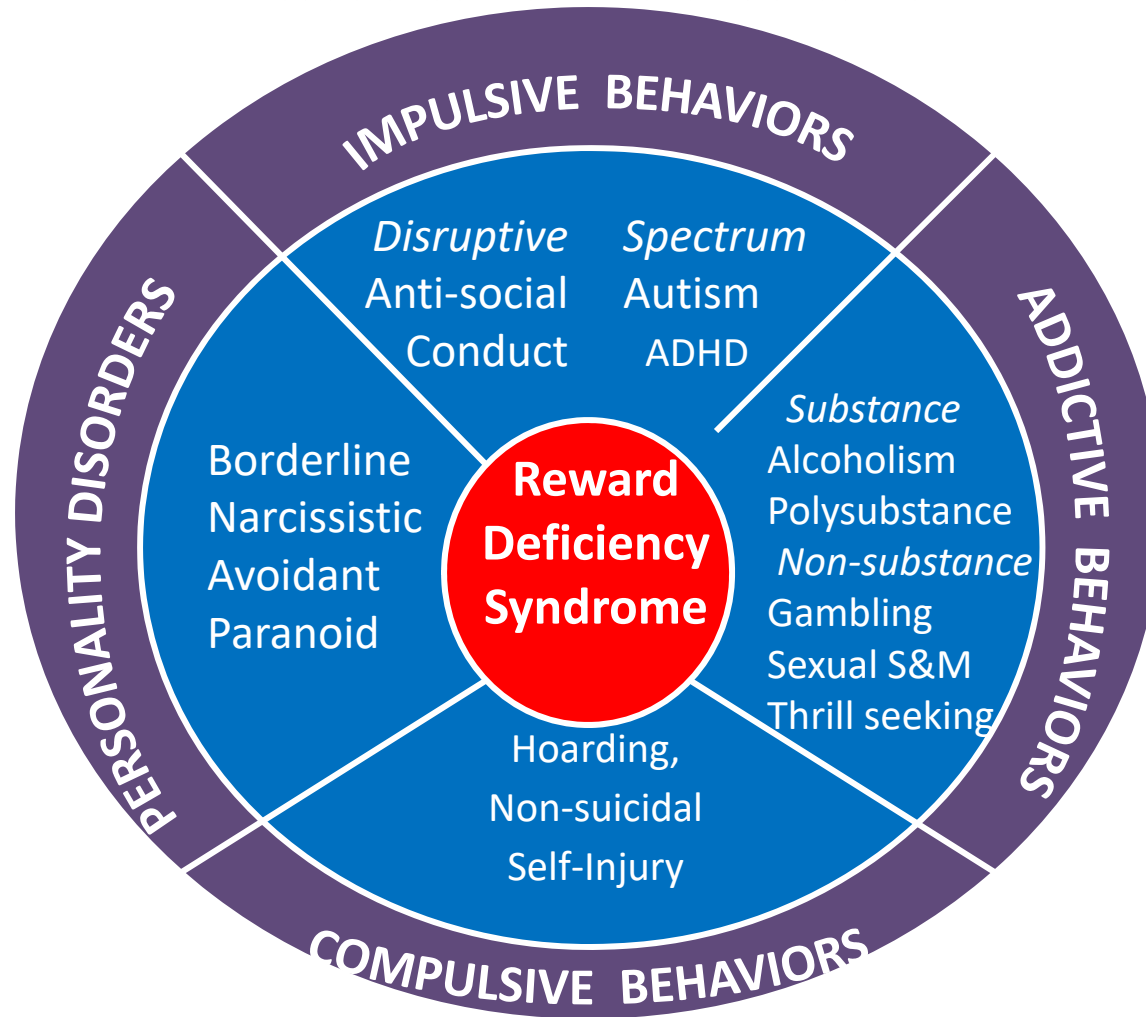


# Polymorphic Reward Gene Variants

*Epigenetic Expression*

**Hypodopaminergic State or Trait**

*Associated Behavioral Outcome*



Blum, K., et al. (2014) Hypothesizing "Reward" Gene Polymorphisms May Predict High Rates of Injury and Addiction in the Workforce: A Nutrient and Electrotherapeutic Based Solution. *Health*, 6:2261-2285.

# Opioid Use Disorder

## Genome Variation

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African-Americans and Hispanics express the dopamine receptor DRD2A1 allele at double the frequency of European-Americans.

In preliminary pharmacogenomics testing in a cohort of African American heroin users reveal a significant 85% frequency of the CYP3A4 \*1B allele, the variant which confers higher metabolism of the treatment drug, buprenorphine, but only a frequency of 13% in European Americans.

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Ettienne E *et.al.* Pharmacogenomics-Guided Policy in Opioid Use Disorder (OUD) Management: An Ethnically-Diverse Case-Based Approach. Addictive Behaviors Reports 6:8-14, 2017.

Kenneth Blum, *et.al.* (2017) Buprenorphine and Naloxone combinations and Dopamine. Current Psychopharmacology, Vol. 0, No. 01.



# NHGC Biophysics Research & Interdisciplinary Development Group (BRIDG)

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James Lindesay, PhD

**To explore the biophysical underpinnings of common variation in the genome to better understand the functional aspects of natural variation (i.e. polymorphisms using first principles of thermodynamics and statistical physics (genodynamics)).**



**Research focus on genome variation expressed in human identity and through population diversity- a natural probe for interrogating the biology and science of health and disease.**

$$E=MC^2$$

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Like everything in the universe, the human genome, and the biology it encodes, is made of energy.

The significant problems we have cannot be solved at the same level of thinking with which we created them.

Albert Einstein

# 2018

# Happy New Year

The Human Genome is a dynamic information and communication system that encodes both systematic and creative aspects of being.



**THE TIME IS ALWAYS NOW**  
 HOWARD UNIVERSITY  
 SESQUICENTENNIAL  
 1867 - 2017

**JANUARY**

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**FEBRUARY**

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From the  
*Biophysics Research and Interdisciplinary  
 Development Group (BRIDG)*

**“Truth is the ultimate theoretical construct for all science.”**

*Georgia M. Dunston, PhD*

**MARCH**

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**APRIL**

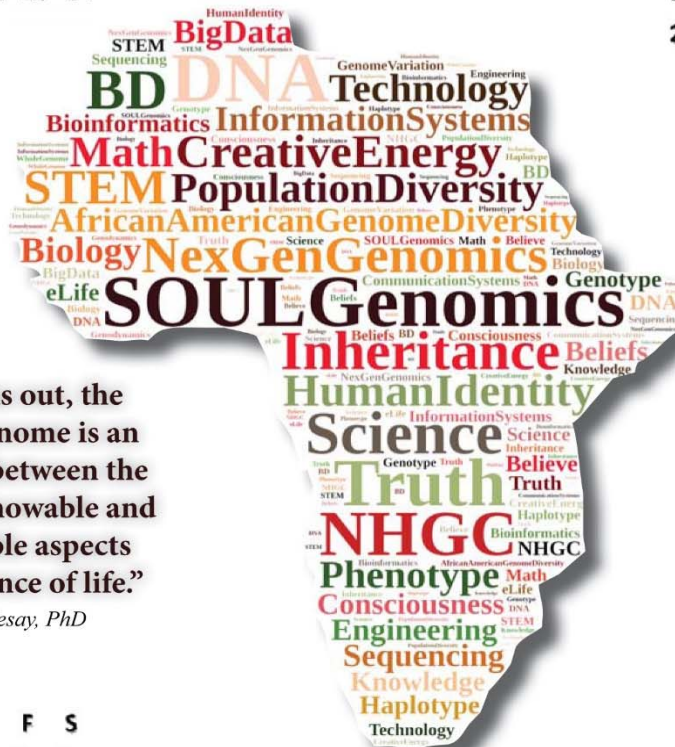
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**MAY**

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**“As it turns out, the human genome is an interface between the known, knowable and unknowable aspects of the science of life.”**

*James Lindesay, PhD*



**“Quantum biology has driven scientists to re-examine long-held assumptions of origins, purpose, and identity.”**

*Philip Kurian, PhD*

**JULY**

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**JUNE**

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**AUGUST**

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**SEPTEMBER**

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**OCTOBER**

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The (Re)union of SCIENCE and SPIRITUALITY in Human Identity and Population Diversity in Health and Disease



**NOVEMBER**

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**DECEMBER**

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# Genomics-Driven Paradigm Shift in Reality

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## FROM

Biochemical  
Molecule  
Physical  
Natural  
Body  
Translation  
Classical  
Disease, Death  
and dying

## TO

Biophysical  
Energy  
Mental  
Supernatural  
Soul  
Transformation  
Quantum  
Health, Life  
and living

# Summary

## ADVANCES IN HUMAN GENOME SCIENCE

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### Offer Unprecedented

**Medical opportunity**

**Commercial promise**

**Ethical danger**

**Social challenge**

# Future Directions

## COMMUNITY-ACADEMIC PARTNERSHIPS IN TRANSLATIONAL RESEARCH

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For conducting research at the interface of genetics, neuroscience, drug abuse, and behavior---

- How can academic research centers, community health service providers, and communities collaborate in using translational research to eliminate health disparities and optimize health equity for *'All of Us'*?

# Healthy People 2020

Genomics  
and Culture

Education &  
Training

Inclusion in  
Clinical Trials

Increase Participation



# The 21<sup>st</sup> Century Cures Act

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The bill contains \$4.8 billion in spending over 10 years for new research at the National Institutes of Health, including:

- **\$1.8 billion for the cancer research “moonshot”** championed by Vice President Joe Biden.
- **\$1.56 billion for the BRAIN Initiative**, a project to create new technologies that will allow for comprehensive mapping of the human brain.
- **\$1.4 billion for the Precision Medicine Initiative**, a project supported by Obama to collect genetic data on one million American volunteers that will be used to help develop new treatments.
- States will receive grants worth **\$1 billion** over the next two years **for drug abuse prevention and treatment programs.**