NIH Director's Perspective And Opportunity for Dialogue

Francis S. Collins, M.D., Ph.D.
Director, National Institutes of Health

NIH Tribal Consultation Advisory Committee Meeting
September 16, 2016

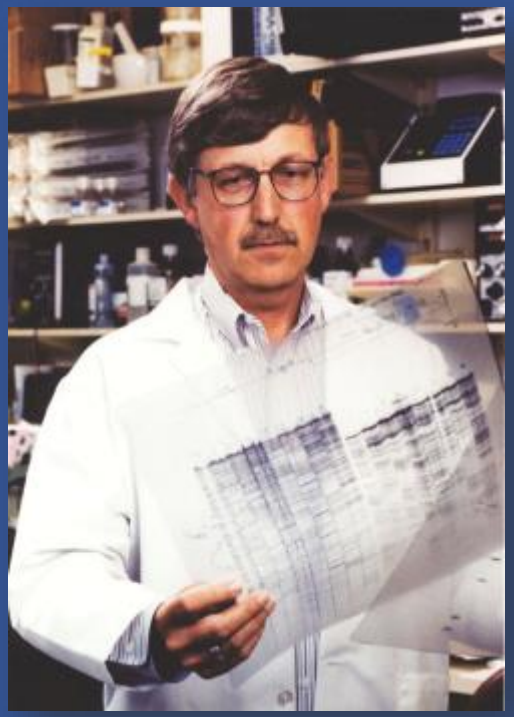
Yale

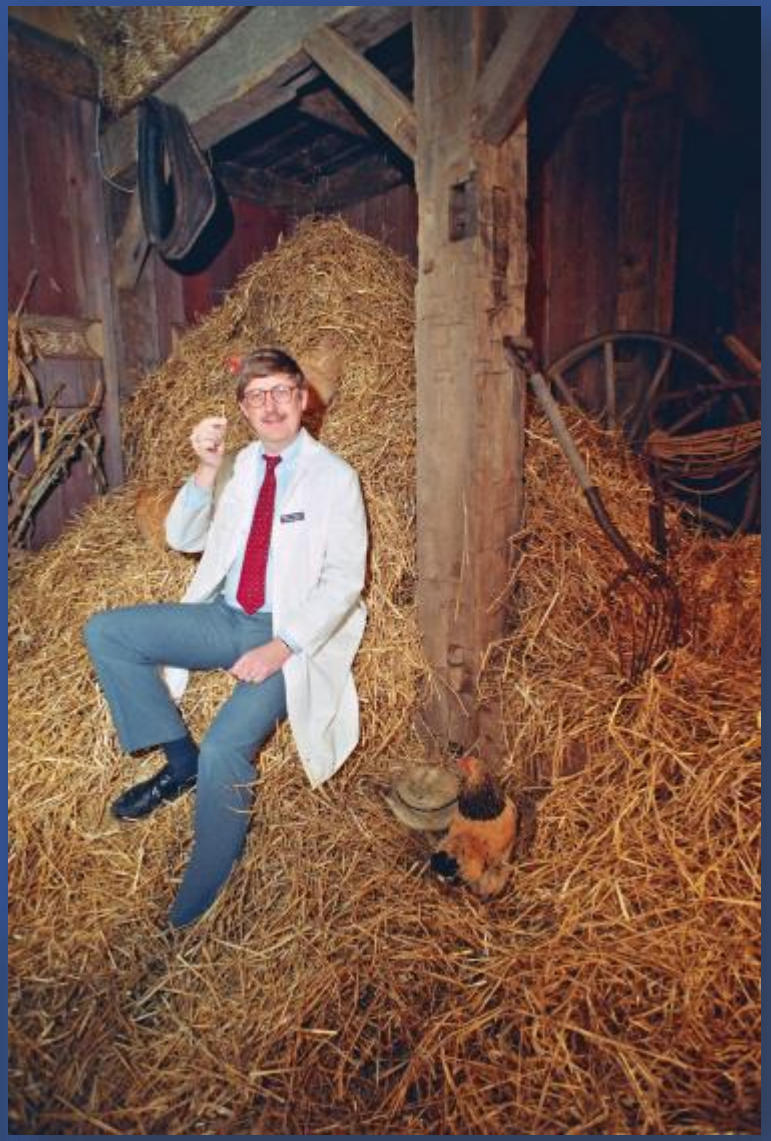
UNC
SCHOOL OF MEDICINE

Yale University
School of Medicine

UNC
SCHOOL OF MEDICINE

“Gene Hunter”

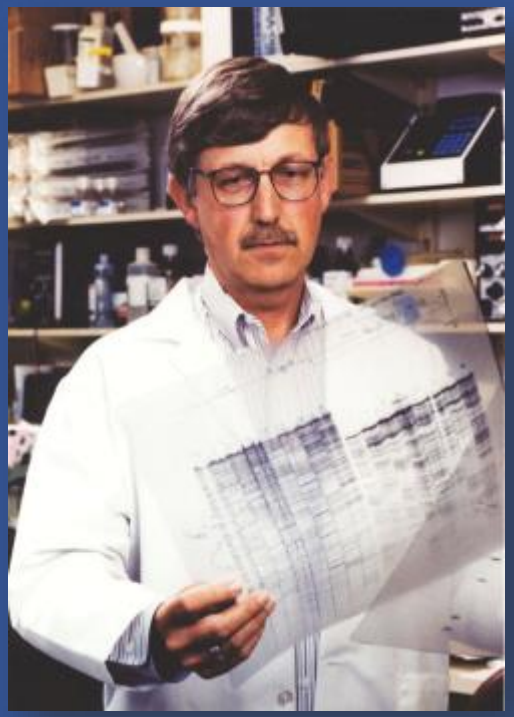


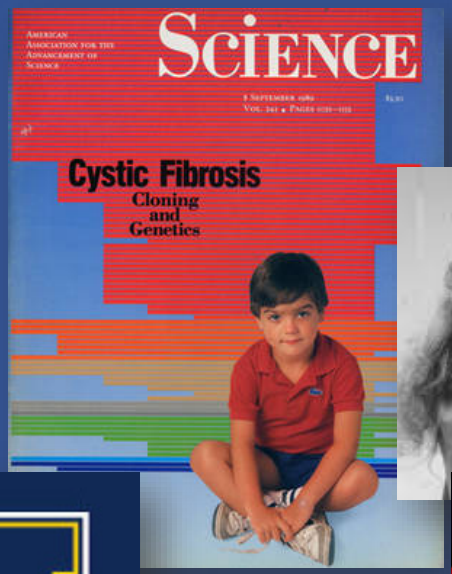


MICHIGAN

“Gene Hunter”

- Cystic fibrosis
- Huntington’s disease
- Neurofibromatosis
- Hutchinson-Gilford Progeria Syndrome





MICHIGAN

Called to Public Service to Lead the Human Genome Project (1993)

The NIH Record

April 27, 1993
Vol. XLV No. 9

'Enormously Pleased To Be Here'

Collins Takes Over Human Genome Project, Institute

By Rich McManus

Celebrated gene chaser Dr. Francis Collins, who in 1989 announced that he and colleagues found the long-sought cystic fibrosis gene and has since codiscovered genes for neurofibromatosis type 1 and Huntington's disease, joined NIH Apr. 3 as director of the National Center for Human Genome Research, which is seeking new status as the National Institute of Genomics and Medical Genetics.

Collins will launch a new intramural science program on campus and has obtained commitments from seven prominent senior investigators who will join NCHGR by fall.

"I am enormously pleased to be here today," said the 43-year-old scientist and physician, who was introduced to the media and NIH colleagues at a Stone House reception on Apr. 7. "I am enormously excited about the Human Genome Project. I can't stop talking about it. It is the single most important scientific endeavor ever mounted by humankind, and it will only happen once in human history.

"By the year 2005, we expect to have the complete sequence of the human genome, as well as genomes of some other organisms. That database will provide grist for the next few centuries of biomedical research. The chance to stand at the helm of that project...is too impossibly wonderful to miss. It is a dream come true."

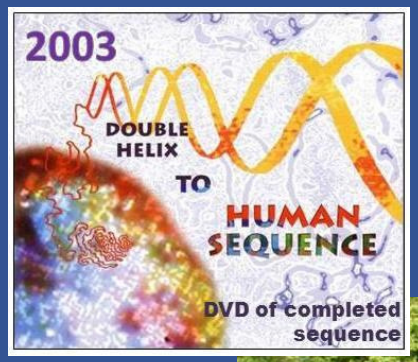
Dr. Francis Collins answers questions at press conference Apr. 7 at Stone House.

Collins, who comes to NIH from the Howard Hughes Medical Institute at the University of Michigan, said his whole life has serendipitously prepared him for the genome post. A native of Staunton, Va., he received his bachelor's degree from the University of Virginia, and M.S. and

(See COLLINS, Page 6)

Leading The Human Genome Project

- 2,500 scientists
- 20 research institutions
- 6 different countries



NATIONAL INSTITUTE OF GENERAL MEDICAL SCIENCES
INDIAN HEALTH SERVICE

AMERICAN INDIAN RESEARCH TRAINING NEEDS MEETING

NATCHER CONFERENCE CENTER
CONFERENCE ROOM E1/E2

AGENDA

August 23, 1999- 8:30am

Traditional Welcome	Richard Harrison, NIDA
Welcome:	Ruth Kirschstein, M.D., Deputy Director, NIH
Introduction	Joan Kauffman
Presentor:	Clifton Poodry

American Indian Research Training Needs Meeting Participants
Natcher Conference Center
August 23-24, 1999

Cleland, Sophia LAKOTA
P.O. Box 452
Lake Andes, SD 57356-0452

Denetclaw, Wilfred, Ph.D. NAVAJO
U.C.S.F. School of Medicine
Department of Anatomy
P.O. Box 0452
San Francisco, CA 94143-0452

Dukepoo, Frank, Ph.D. HOPI-LAGUNA
Northern Arizona University
1625 N. Prairie Way
Flagstaff, AZ 86004

An important mentor for me…

Community Genetics

Frank C. Dukepoo

Department of Biological Sciences,
Northern Arizona University,
Flagstaff, Ariz., USA

Community Genet 1998;1:130–133

Genetic Services in the New Era: Native American Perspectives

Science AAAS

Ancestors of Science, Frank C. Dukepoo

By Next Wave and Science Staff | Sep. 10, 2004 , 8:00 AM

ANCESTORS OF SCIENCE

Frank C. Dukepoo was a geneticist, but he was equally well known for his tireless efforts to improve Native American education. As one of a handful of Native Americans with science doctorates, he was a crusader for scientific and educational change.

Dukepoo was born in 1944 on Arizona's Mohave Indian Reservation to working class parents Eunice, a Laguna, and Anthony Dukepoo, a Hopi. His older brother Freddie, a former lab technician, and his high school counselor, Abraham Lincoln Herm, served as his role models.

As a college freshman in 1961 (when Native American college students were virtually nonexistent), Dukepoo experienced rampant racism at Arizona State University (ASU).

Indians, Genes and Genetics:
What Indians Should Know About the New Biotechnology

By Debra Harry and Frank C. Dukepoo

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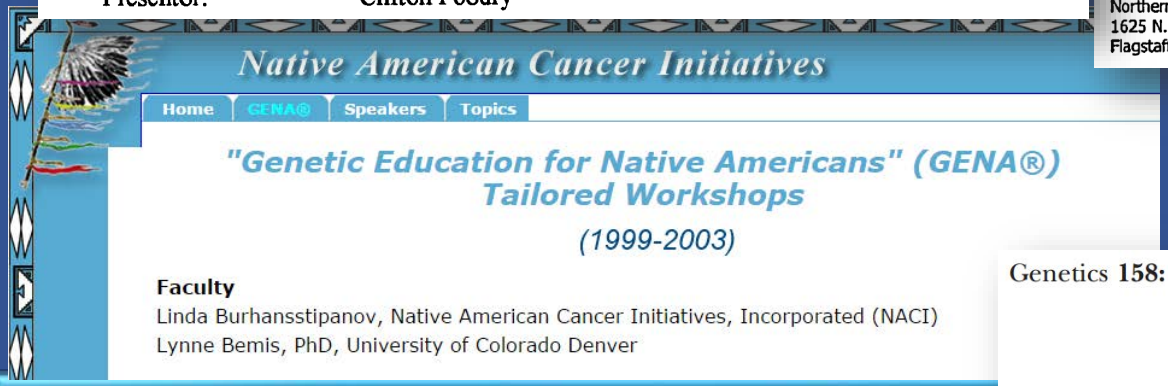

Genetics **158:** 941–948 (July 2001)

Genetics Education

Innovations in Teaching and Learning Genetics
Edited by Patricia J. Pukkila

Development of a Genetics Education Workshop Curriculum for Native American College and University Students

Linda Burhansstipanov,* Lynne Bemis,† Mark Dignan‡ and Frank Dukepoo§,1

"Funded by the ethical, legal, and social implications (ELSI) component of the National Human Genome Research Institute, Genetic Education for Native Americans (GENA) … [has] the long-term goal of providing a balance of scientific and cultural information to increase understanding of genetic research within and among tribes and awareness of careers in genetics."

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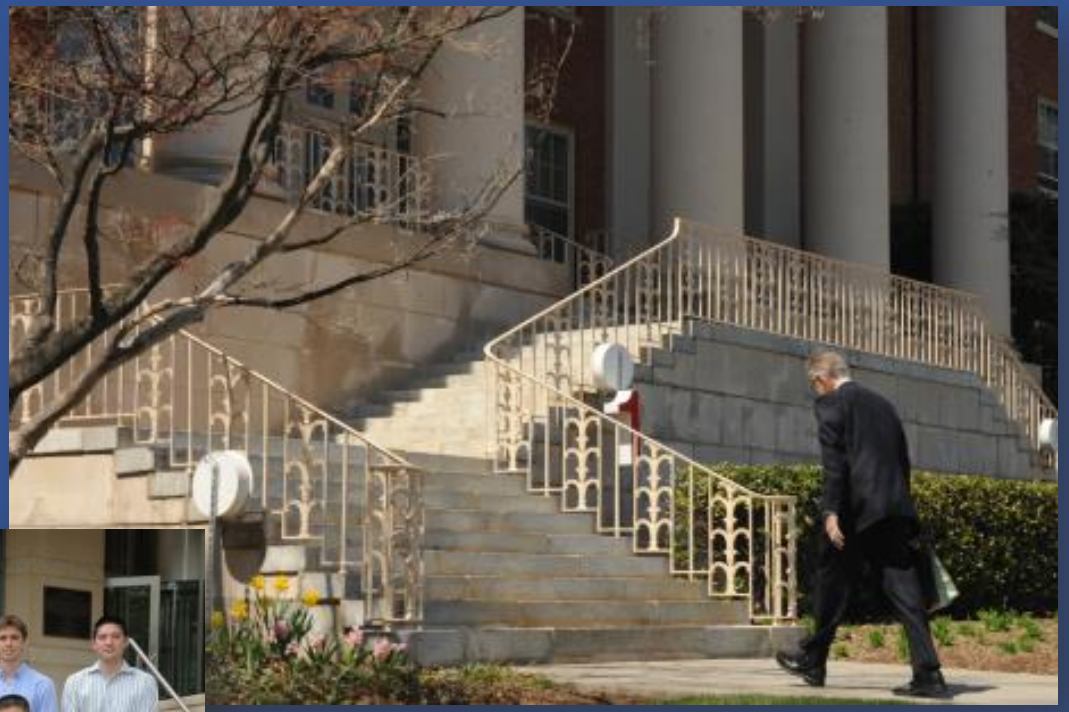
Return to NIH as Director in 2009

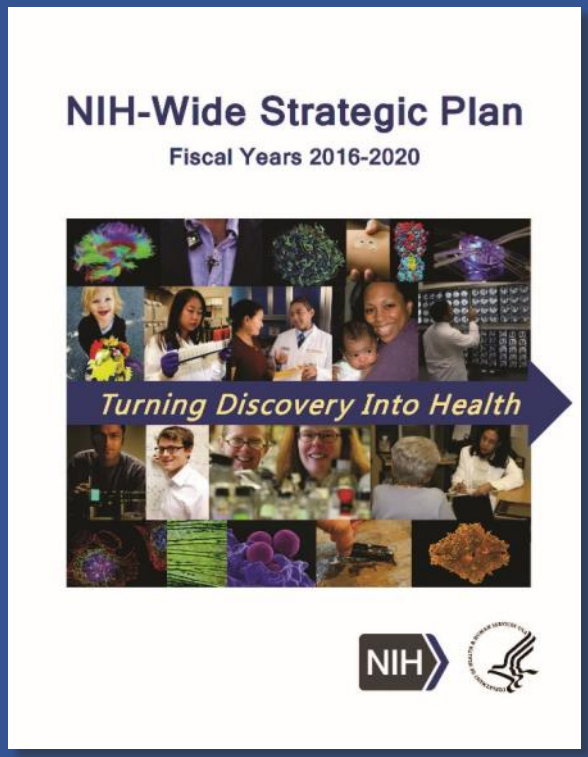
Still a Lab Principal Investigator



NIH: Steward of Medical and Behavioral Research for the United States

"Science in pursuit of fundamental knowledge about the nature and behavior of living systems and the application of that knowledge to extend healthy life and reduce illness and disability."

NIH Strategic Planning



Overview

- Mission of NIH
- Unique moment of opportunity in biomedical research
- Current NIH-supported research landscape
- Constraints confronting the community in the face of lost purchasing power

Objective 1: Advance Opportunities in Biomedical Research

Fundamental Science

- Foundation for progress
- Consequences often unpredictable
- Technology leaps catalyze advances
- Data science increases impact/efficiency

Health Promotion/Disease Prevention

- Importance of studying healthy individuals
- Advances in early diagnosis/detection
- Evidence-based reduction of health disparities

Treatments/Cures

- Opportunities based on molecular knowledge
- Breakdown of traditional disease boundaries
- Breakthroughs need partnerships, often come from unexpected directions
- Advances in clinical methods stimulate progress

Objective 2: Set Priorities

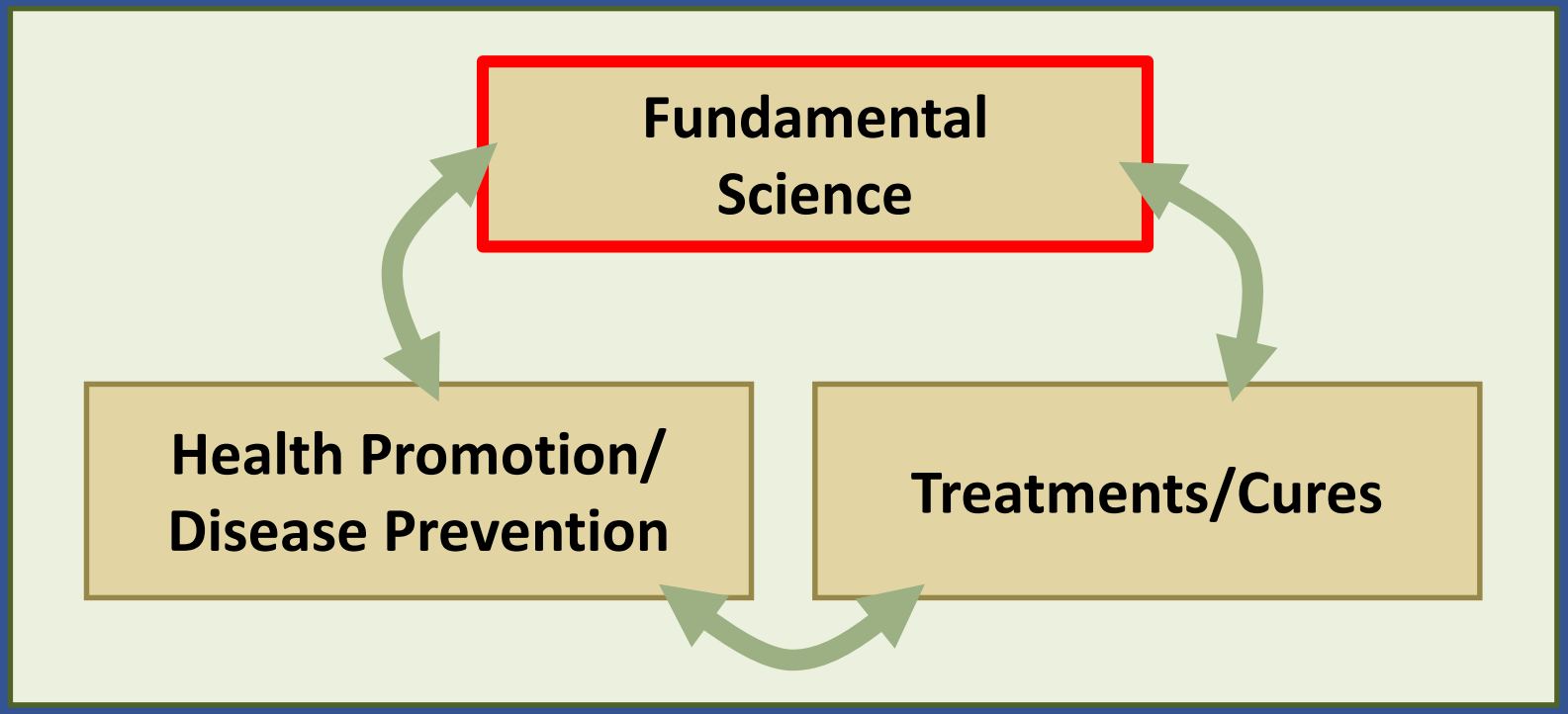
- Incorporate disease burden as important, but not sole factor
- Foster scientific opportunity; need for nimbleness
- Advance research opportunities presented by rare diseases
- Consider value of permanently eradicating a pandemic risk

Objective 3: Enhance Stewardship

- Recruit/retain outstanding research workforce
- Enhance workforce diversity
- Encourage innovation
- Optimize approaches to inform funding decisions
- Enhance impact through partnerships
- Ensure rigor and reproducibility
- Reduce administrative burden

Objective 4: Excel as a Federal Science Agency by Managing for Results

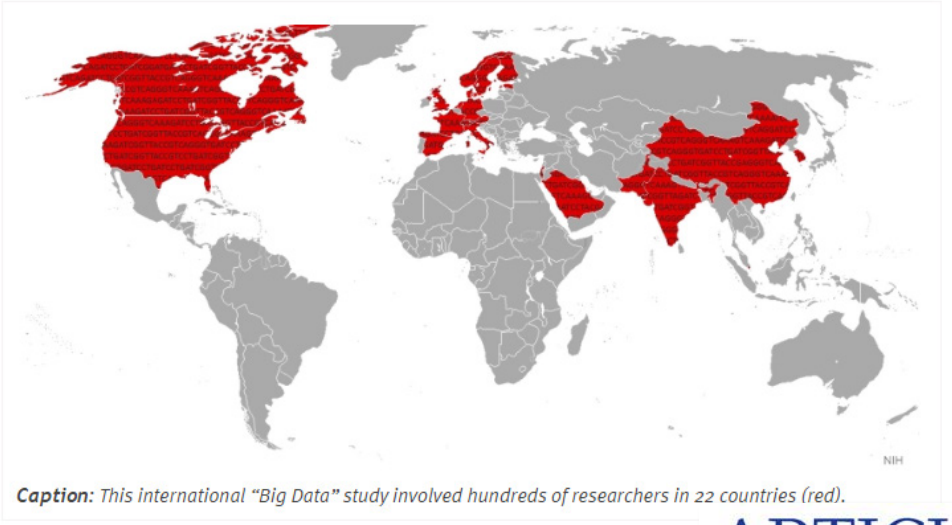
Advancing Opportunities in Biomedical Research



NIH DIRECTOR'S BLOG

International "Big Data" Study Offers Fresh Insights into T2D

Posted on July 12, 2016 by Dr. Francis Collins



Caption: *This international "Big Data" study involved hundreds of researchers in 22 countries (red).*

It's estimated that about 10 percent of the world's population either has
will develop the disease during their lives [1]. Type 2 diabetes (formerly
happens when the body doesn't produce or use insulin properly, causing g

ARTICLE

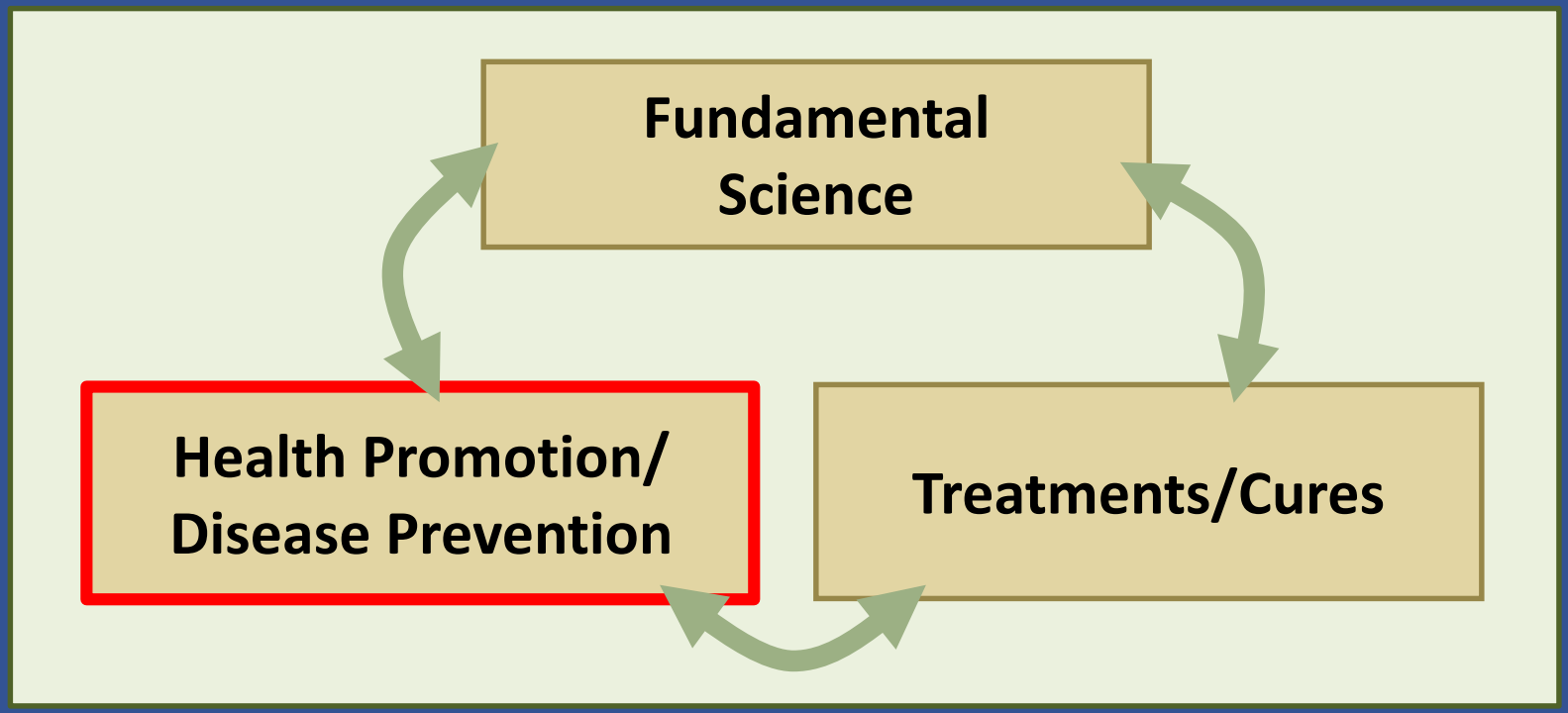
4 AUGUST 2016 | VOL 536 | NATURE

The genetic architecture of type 2 diabetes

A list of authors and affiliations appears in the online version of the paper

The genetic architecture of common traits, including the number, frequency, and effect sizes of inherited variants that contribute to individual risk, has been long debated. Genome-wide association studies have identified scores of common variants associated with type 2 diabetes, but in aggregate, these explain only a fraction of the heritability of this disease. Here, to test the hypothesis that lower-frequency variants explain much of the remainder, the GoT2D and T2D-GENES consortia performed whole-genome sequencing in 2,657 European individuals with and without diabetes, and exome sequencing in 12,940 individuals from five ancestry groups. To increase statistical power, we expanded the sample size via genotyping and imputation in a further 111,548 subjects. Variants associated with type 2 diabetes after sequencing were overwhelmingly common and most fell within regions previously identified by genome-wide association studies. Comprehensive enumeration of sequence variation is necessary to identify functional alleles that provide important clues to disease pathophysiology, but large-scale sequencing does not support the idea that lower-frequency variants have a major role in predisposition to type 2 diabetes.

Advancing Opportunities in Biomedical Research



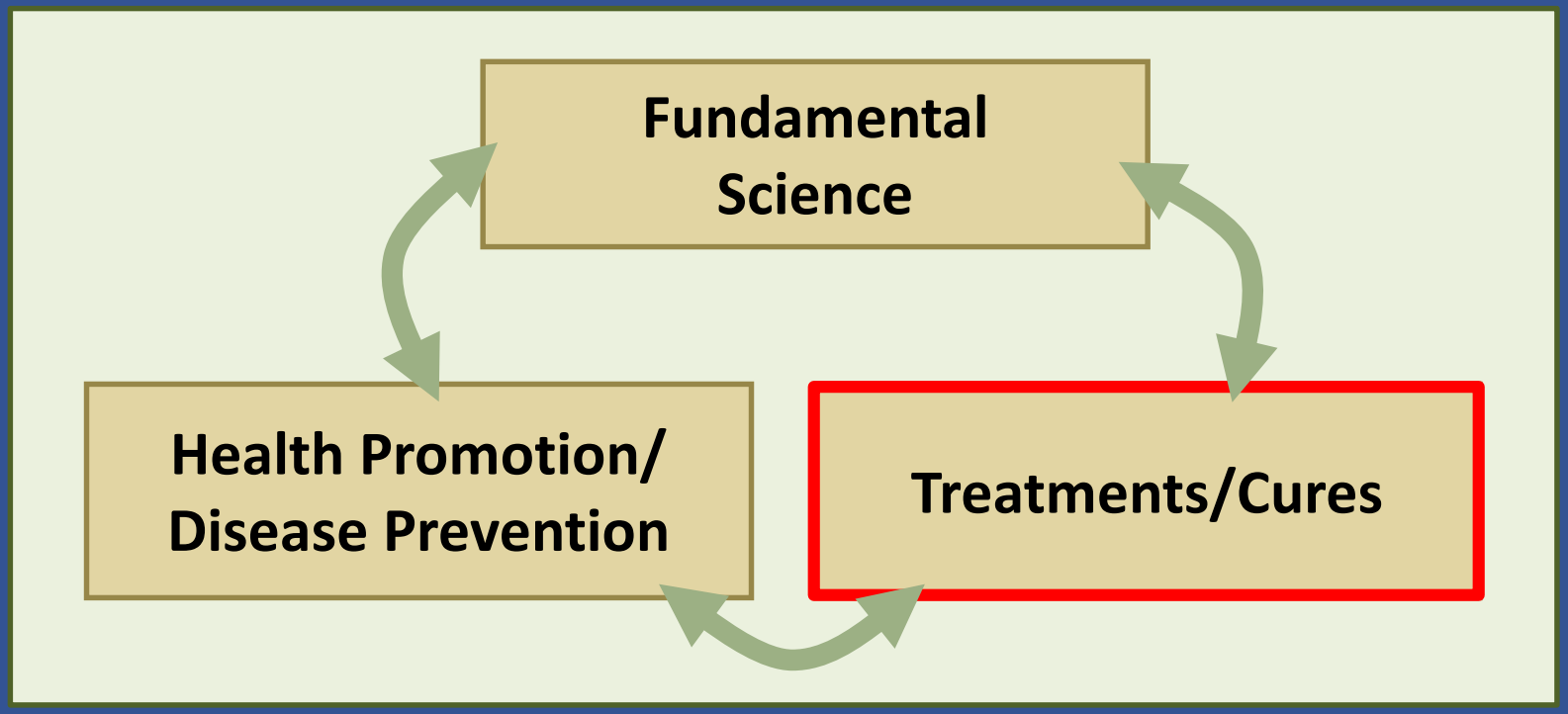
Intervention Research to Improve Native American Health (IRINAH)

- IRINAH: founded in 2011 to develop, adapt, test effectiveness of health promotion, disease prevention interventions in AI/AN populations
  - Network of principal investigators and their partners
  - Supported by multiple NIH Institutes and Centers
- Mission advanced by consecutive Program Announcements – one (PAR-14-260) is currently taking applications
- Funded projects include:
  - "Residential Wood Smoke Interventions Improving Health in Native American Populations" (Curtis Noonan, Univ. Montana)
  - "Diet Intervention for Hypertension: Adaptation and Dissemination to Native Communities" (Valarie Blue Bird Jernigan, Univ. Oklahoma)

IRINAH
Intervention Research to Improve Native American Health

IRINAH
spring 2016 projects update

Advancing Opportunities in Biomedical Research



Intramural NIDDK Phoenix Epidemiology and Clinical Research Branch (PECRB)

- 1963: NIDDK and Indian Health Service researchers discovered high rate of type 2 diabetes (T2D) in Gila River Indian (Pima) Community
  - Founded PECRB
- Today, in multiple sites, PECRB studies etiology of obesity, diabetes – especially among American Indians
  - Conducts epidemiological, genetic, physiological research
- American Indian participants have been invaluable partners

THE NIH CATALYST — A PUBLICATION ABOUT NIH INTRAMURAL RESEARCH

NATIONAL INSTITUTES OF HEALTH — JANUARY-FEBRUARY 2013

NIDDK Research in Clinic in Arizona Aims to Lessen Health Disparities

BY AMY F. REITER, NIDDK

that the burden of type 2 diabetes is great in that community as well as in other Native American communities in and near Phoenix."

The close-knit community may enable better research. "We are conducting the Family Investigation pathy in Diabetes (FIND) to genetic factors that contribute kidney disease," said PECRB Robert Nelson. "In this proto-

Leslie J. Baier,[1] Yunhua Li Muller,[1] Maria Sara Remedi,[2] Michael Traurig,[1] Paolo Piaggi,[1] Gregory Wiessner,[1] Ke Huang,[1] Alyssa Stacy,[1] Sayuko Kobes,[1] Jonathan Krakoff,[1] Peter H. Bennett,[1] Robert G. Nelson,[1] William C. Knowler,[1] Robert L. Hanson,[1] Colin G. Nichols,[2] and Clifton Bogardus[1]

***ABCC8* R1420H Loss-of-Function Variant in a Southwest American Indian Community: Association With Increased Birth Weight and Doubled Risk of Type 2 Diabetes**

Diabetes 2015;64:4322–4332 | DOI: 10.2337/db15-0459

J Clin Endocrinol Metab, April 2016

Metabolic Risk Factors and Type 2 Diabetes Incidence in American Indian Children

Kevin M. Wheelock, Madhumita Sinha, William C. Knowler, Robert G. Nelson, Gudeta D. Fufaa, and Robert L. Hanson

Centers for American Indian and Alaska Native Health Center for Diabetes Translational Research (CAIANDTR)

- Seeks to increase scientific knowledge about diabetes prevention, management interventions proven effective in clinical and community settings
  - Goal: improve diabetes-related health of AI/AN individuals
- Recent project assessed the Special Diabetes Program for Indians-Diabetes Prevention Demonstration Project
  - Affected dietary, activity changes; factors affecting retention, participation
  - How outcomes are affected by psychosocial, socioeconomic factors

colorado school of public health
UNIVERSITY OF COLORADO
COLORADO STATE UNIVERSITY
UNIVERSITY OF NORTHERN COLORADO

National Cancer Moonshot

- Announced by President Obama, 2016 State of the Union
  - Vice President Biden appointed to lead
- Proposed $755M in FY17 for NIH and FDA

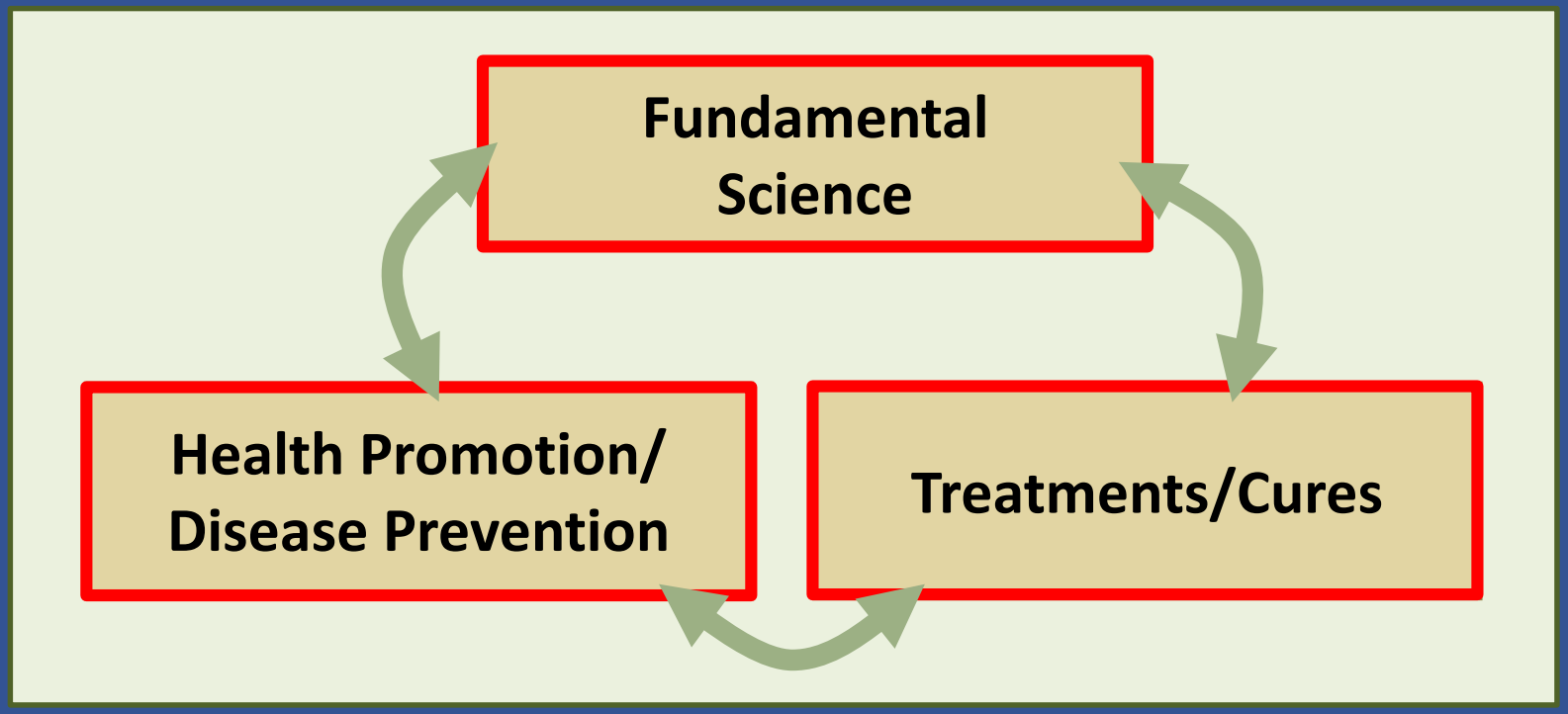
Cancer Moonshot's Blue Ribbon Panel
10 Recommendations

Areas include:

- Cancer immunotherapy clinical trials network
- Therapeutic target identification to overcome drug resistance
- Development of new enabling cancer technologies
- Prevention and early detection: implementation of evidence-based approaches

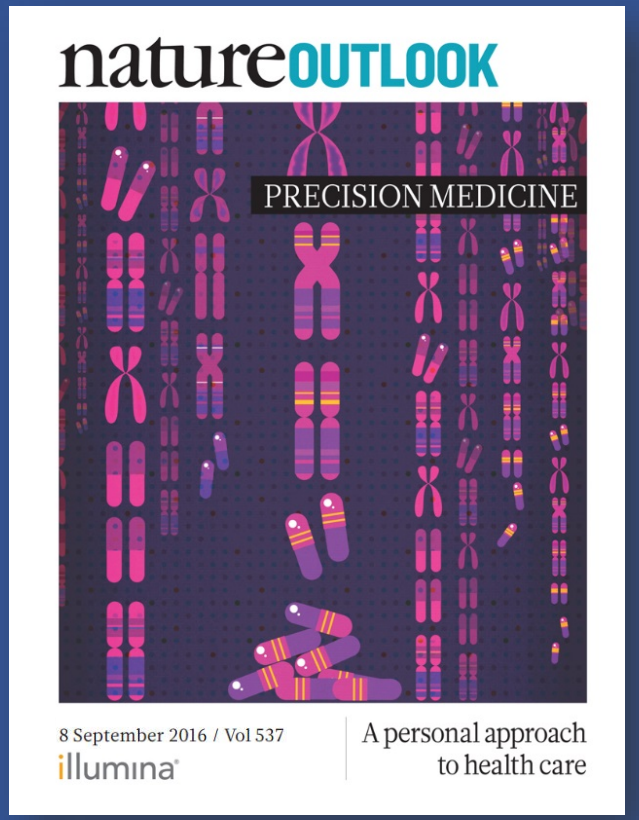
Advances "would prevent additional cancer cases and unnecessary deaths in the general population, populations that experience persistent cancer disparities (e.g., low-income, minority, rural, and other underserved populations), as well as populations with familial cancer risk…."

Advancing Opportunities in Biomedical Research



Precision Medicine

- An emerging approach for disease treatment and prevention that takes into account individual variability in lifestyle, environment, and genes
- A radical shift in how each of us can receive the best care possible based on our unique makeup



The Precision Medicine Initiative® Cohort Program

- Cornerstone of larger NIH-led PMI
- One of the most ambitious research projects in history
  - In size and scope
- One million or more volunteers
  - Reflecting the broad diversity of the U.S.
- Opportunities for volunteers to provide data on an ongoing basis
- Launch to happen in phases
  - Estimated 3–4 years to reach one million
- Data shared freely and quickly to inform a variety of research studies

The scope of biomedical research supported through and at the NIH is wide, and we're confident that, thanks to the talented staff and scientists that work there, we'll one day find cures for diseases…. Ensuring this efficient basic biomedical research base and supporting the next generation of researchers is critical to pave the way for these long-term advancements.

— Congressman Tom Cole




NIH… Turning Discovery Into Health

directorsblog.nih.gov @NIHDirector