October 6, 2015

This month brought a historic 'odometer moment' for the field of genomics – October 1, 2015, marked the 25th anniversary of the launch of the Human Genome Project. I, for one, cannot believe a quarter-century has now passed since many of us started working on the Project. At the same time, it is truly incredible to think about how far genomics has progressed since that time. I thought the significance of this anniversary warranted making this topic the lead story in this month's *The Genomics Landscape*; in addition, I reflect on this important anniversary in a recent video interview now available on the NHGRI web site.

With this issue, I am also happy to report that *The Genomics Landscape* turns two years old. Even after 24 issues, the number of noteworthy items has not diminished, as evidenced by the other topics featured here; see various details below, along with other information items that I hope will be of interest to you.

Specifically, October's *The Genomics Landscape* features stories about:

- 25th Anniversary of the Launch of the Human Genome Project
- Alan Guttmacher Retires as NICHD Director
- Undiagnosed Diseases Network Patient Gateway Launched
- NHGRI Implementation of the NIH Genomic Data Sharing Policy
- National Library of Medicine Director Search
- NHGRI Awards Functional Variation Grants
- Human Heredity and Health in Africa Public Event and Videocast
- ASHG-NHGRI Genetics & Public Policy and Genetics & Education Fellows

All the best,

Watch here for current and upcoming locations of the Smithsonian-NHGRI exhibition "Genome: Unlocking Life's Code" as it tours North America!

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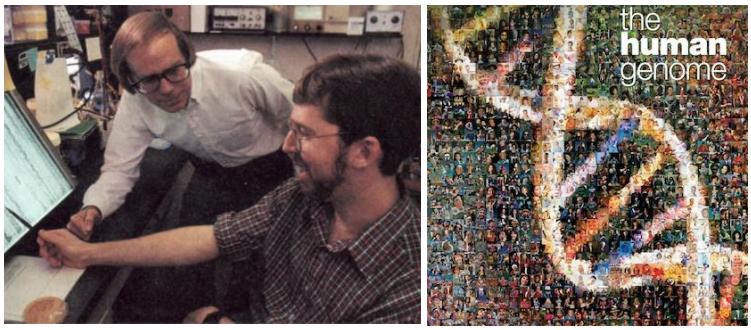
25th Anniversary of the Launch of the Human Genome Project

A quarter-century ago, the NIH teamed up with the U.S. Department of Energy, the Wellcome Trust in the United Kingdom, and scientists from around the world to launch the Human Genome Project (HGP) – a massive, collaborative effort to map and sequence the human genome. October 1, 2015, marked the 25th anniversary of this launch. I find it hard to believe that it has been 25 years since the Project lunged from its starting line!

In 1990, scientists and program administrators set lofty goals to complete the HGP in 15 years and at a cost of ~$1 per base of the human genome. Since the human genome is ~3 billion bases in size, the total cost of the HGP was, therefore, projected to be $3 billion dollars. While hopes were high that this could be achieved, there were many obstacles to overcome. Genetic and physical maps of the human genome needed to be generated. DNA sequencing technologies and strategies needed to be developed. Coordination and robust management practices needed to be put into place. In the beginning, it was unclear to many of the participants whether the HGP could be completed in the projected time or at the projected cost.

At the time of the HGP's launch, I was a postdoctoral fellow in Dr. Maynard Olson's laboratory at Washington University (see picture below), working on the human genome mapping efforts of the HGP. It was the beginning of a new scientific era, and none of us quite envisioned just how quickly genomics would come into its own as a discipline and become so integral to other areas of basic research, let alone translational and clinical research.



On left: Maynard Olson and Eric Green, Washington University, 1990.
On right: Cover art from the 2001 publication describing the draft genome sequence in Nature.

To capture the stories and experiences of some HGP participants, NHGRI and the NHGRI History of Genomics Program will be hosting a special seminar series, "A Quarter Century after the Human Genome Project's Launch: Lessons beyond the Base Pairs," on the NIH campus. We will kick off the

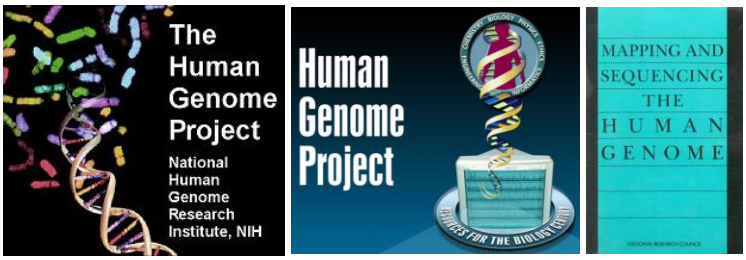
Alan Guttmacher Retires as NICHD Director

Dr. Alan Guttmacher, Director of the *Eunice Kennedy Shriver* National Institute for Child Health and Human Development (NICHD), retired from federal service last month. Dr. Guttmacher came to NIH in 1999 to work at NHGRI, where he served in a number of roles, including Deputy Director and Acting Director. In 2010, after a vigorous search, Alan was named NICHD Director, after which he oversaw the Institute's activities as the focal point at NIH for research in pediatric health and development, maternal health, reproductive health, intellectual and developmental disabilities, and rehabilitation medicine, among other areas. Working with Alan, NHGRI has benefited from numerous interactions with NICHD, including launching the **N**ewborn **S**equencing **I**n **G**enomic medicine and public **H**eal**T**h (NSIGHT) program. His scientific leadership and passion for genomics have been invaluable to NIH, NHGRI, and NICHD, and we wish Alan all the best in his retirement.

Undiagnosed Diseases Network Patient Gateway Launched

In September, the Undiagnosed Diseases Network (UDN) launched its online patient application portal, called the UDN Gateway. The UDN's goal is to improve the level of diagnosis and care for patients with conditions that even skilled physicians have been unable to diagnose despite extensive clinical investigation. The UDN grew out of the success of the Undiagnosed Diseases Program (UDP) at the NIH Clinical Center in Bethesda, Maryland. Since its 2008 launch, the UDP has reviewed more than 3,100 applications and admitted more than 800 patients to the NIH Clinical Center for comprehensive evaluation. The UDN now includes seven clinical sites located across the United States. By the summer of 2017,

series in December with a panel discussion chaired by me, and including former NHGRI Director and current NIH Director, Francis Collins, and former NHGRI Deputy Directors, Elke Jordan and Mark Guyer. Other confirmed speakers for the series include: Maynard Olson, Ewan Birney, Bob Cook-Deegan, Marco Marra, and David Bentley. The lectures and discussions will be videorecorded, and the NHGRI History of Genomics Program will conduct oral history interviews with the presenters and add these to the Institute's growing historical archives.



From left to right: HGP graphic from NHGRI, HGP graphic from U.S. DOE, and cover of the 1988 National Academies report "Mapping and Sequencing the Human Genome".

Part of the HGP's legacy was the fashion in which it paved the way for other 'big biology' projects that followed. Reflecting on this anniversary, I recently co-authored a *Nature* Comment entitled "Twenty-Five Years of Big Biology" with Francis Collins and James Watson, former Directors of NHGRI (or the National Center for Human Genome Research, as we were named at the time Dr. Watson was Director). In that Comment, we describe six key lessons from the HGP beyond the actual genomic data and technologies. These lessons have served to guide many important research projects, including many that are ongoing today. In addition to the written Comment, *Nature* also recently released a 'podcast' interview with me about the HGP's 25th anniversary.

When the HGP started, there was much uncertainty about whether we could really sequence the human genome, let alone do it in 15 years. Being able to sequence a human genome in a matter of days for close to $1,000 seemed like science fiction. Yet this is the reality we are in today! The technological advances that have occurred to make this happen are truly astounding. I am certain that genomics will prove to be as exciting (and impactful) in the next 25 years as it has been over the last 25.

To access a brief history of the Human Genome Project (HGP), see genome.gov/12011239.

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each new clinical site will accept about 50 patients per year. For more information, see genome.gov/27562471.

NHGRI Implementation of the NIH Genomic Data Sharing Policy

NHGRI recently released information about how the Institute will implement the new NIH Genomic Data Sharing (GDS) Policy and related requirements. NHGRI is committed to ensuring that we remain a leader in implementing the GDS Policy within both our Extramural and Intramural Research Programs. For more information, see genome.gov/27562511. Additionally, if you are interested in receiving email notifications from NIH regarding changes and updates to the NIH Genomic Data Sharing (GDS) website, please sign up for the Genomic Data Sharing Listserv.

National Library of Medicine Director Search

A search has been launched to identify the next Director of the National Library of Medicine (NLM) at NIH. This senior position focuses on the direction and management of this critically important component of NIH, a new vision for which was recently established by a Working Group of the Advisory Committee to the NIH Director. Applications will be reviewed starting October 20, and will be accepted until the position is filled. For more information, see jobs.nih.gov/vacancies/executive/nlm_director.htm.

NHGRI Awards Functional Variation Grants

NHGRI, along with the National Cancer Institute, has awarded six new grants for developing new computational approaches to identify genomic variants that influence disease susceptibility or other traits. In the coming years, many whole-genome sequences will be generated, revealing millions of genomic variants. A major unsolved problem is that we do not yet know how to interpret most genomic variants, especially in non-coding regions of the genome (regions that do not directly code for proteins). In order to understand how a genomic variant could contribute to health or disease, scientists need to first identify which genomic variants actually influence biological processes. The goal of this set of new grants is to develop scientific approaches that integrate data from many sources to determine the functional consequences of individual genomic variants. Such data would narrow the set of genomic variants that might be relevant to a disease. The proposed approaches will use information from transcripts, transcription factor binding, nucleosomes, enhancers, DNA shape, conservation, and phenotypes. The funded grants will study autism, cancer, bipolar disorder, type 2 diabetes, and age-related macular degeneration. For more information, see genome.gov/27562555.

Human Heredity and Health in Africa Public Event and Videocast

As part of its 7th Consortium Meeting, the Human Heredity and Health in Africa (H3Africa) program is hosting a public event and videocast on the main campus of the NIH on October 13 beginning at 9:30 AM. The program includes presentations from NIH Director Dr. Francis Collins, Professor Dame Kay Davies of Oxford University, and Dr. Jim Anderson, Director of the NIH Division of Program Coordination, Planning, and Strategic Initiatives (which oversees the NIH Common Fund). To attend the NIH event in person, please register at goo.gl/forms/HCNG7r5ibf. To join the videocast, see videocast.nih.gov.

ASHG-NHGRI Genetics & Public Policy and Genetics & Education Fellows

Each year, NHGRI's Division of Policy, Communications, and Education partners with the American Society of Human Genetics in sponsoring two fellowships – the well-established Genetics and Public Policy Fellowship and the recently launched Genetics and Education Fellowship. The Genetics and Public Policy Fellowship provides the fellow an opportunity to gain valuable policy experience at the Institute, at ASHG, and in the U.S. Congress. The Genetics and Education Fellowship provides a genetics professional the opportunity to receive training and experience in preparation for a career in genetics/genomics education. This year's Genetics and Public Policy fellow is Caroline Young, M.S., and the Genetics and Education fellow is Julie Nadel, Ph.D. For details about the new fellows, visit genome.gov/27562481.

On left: Caroline Young.
On right: Julie Nadel.

Spotlight on the Precision Medicine Initiative (PMI)

nih.gov/precisionmedicine

- During a public teleconference on September 17, the Precision Medicine Initiative Working Group of the Advisory Committee to the NIH Director unveiled its report, entitled "The Precision Medicine Initiative Cohort Program – Building a Research Foundation for 21st Century Medicine." The report makes recommendations about the establishment of the U.S. National Research Cohort component of the Precision Medicine Initiative. It was approved unanimously, and accepted by the NIH Director, Dr. Francis Collins. The full text of the report can be found here.
- A search will soon be launched for a Director of the Precision Medicine Initiative Cohort Program. Until the new Director is identified, Dr. Josephine Briggs, Director of the National Center for Complementary and Integrative Health at NIH, will serve as Interim Director. Dr. Briggs will work with leadership from other NIH Institutes and Centers on the early launch of the cohort program.
- On September 22, a PMI Twitter Chat took place to discuss the recently released report, and on September 25, a stakeholders briefing was held. To see the results of the chat, visit here and to watch the briefing, visit here.

Genomics Research

Investigating Why Cancer Comes Back

An Ancient Tumor in Dogs Might Teach New Tricks about Cancer in People

Who Should Decide? The Complex Ethics of Pediatric Genome Sequencing

Diet, Exercise, Smoking Habits and Genes Interact to Affect AMD Risk

A New Single-Molecule Tool to Observe Enzymes at Work

New Hope for Undiagnosed, Critically Ill Newborns at Rady Children's

Scientists Create World's Largest Catalog of Human Genomic Variation

Scientists Test New Gene Therapy for Vision Loss from a Mitochondrial Disease

Genome Advance of the Month

Gene-Editing Technology Uncovers Genetic Link to Infertility

Awards & Recognition

Thomas A. Waldmann Award for Excellence in Human Immunology 2015 – Dan Kastner

Sigma Theta Tau Honor Society of Nursing: International Awards for Nursing Excellence Best of Publication 2015 – Jean Jenkins

University of Texas Alumnus Recognized for Leukemia Research – Paul Liu

Funding Opportunities

A Data Analysis and Coordinating Center for Research Training and Career Development Activities

Novel Nucleic Acid Sequencing Technology Development (R21, R01, and R43/R44)

Funding News

Notice on NIH Research Involving Introduction of Human Pluripotent Cells into Non-Human Vertebrate Animal Pre-Gastrulation Embryos

Participation of Additional NIH Components in "Emerging Global Leader Award"

New Genomics Videos

Advice from the Genetic and Rare Diseases Information Center

UDN Begins Accepting Patient Applications

National Advisory Council for Human Genome Research – September 2015

Genetics/Genomics Competency Center (G2C2) Overview

The Impact of the Human Genome Project, 25 Years from its Launch

1000 Genomes Project Scientists Create World's Largest Catalog of Human Genomic Variation

Upcoming Webcasts

Human Heredity and Health in Africa Project Presentations – October 13, 2015

NIH & NHGRI News of Interest

Kaufmann Appointed Head of NCATS' Office of Rare Diseases Research

Genome Unlocking Life's Code: Timeline of Ancient DNA

NIH Framework Points the Way Forward for Building National, Large-Scale Research Cohort, a Key Component of the President's Precision Medicine Initiative

NIH Addresses the Science of Diversity

NIH Commits $36M to Train Junior Faculty in Africa

Dr. Eric Green Discusses the Work of NHGRI and His Perspective on Genomics

Grants to Help Identify Variants in the Genome's Regulatory Regions that Affect Disease Risk

Precision Medicine: Much More Than Just Genetics

Undiagnosed Diseases Network Launches Online Application Portal

NHGRI Welcomes 2015 ASHG/NHGRI Education and Public Policy Fellows

NHGRI Researchers to Present Talks, Posters at ASHG Annual Meeting

NIH Common Fund Launches Four Programs Designed to Take Aim at Gaps in Biomedical Research

United States Patent and Trademark Office Report on Confirmatory Genetic Diagnostic Test Activity