February 2, 2016

Much to the chagrin of most adult Washingtonians (I note that children have a very different attitude about this), our seemingly mild winter ended abruptly last weekend with the accumulation of a record-breaking amount of snow. However, the 'Blizzard of 2016' failed to chill my enthusiasm for recent developments with the NHGRI Genome Sequencing Program – whose new phase was announced last month.

Our recent attention has also been focused on the upcoming meeting of the National Advisory Council for Human Genome Research (NACHGR), which begins on Monday, February 8 at 10:00 AM EST. To watch my Director's Report as well as other portions of the meeting's Open Session, visit genome.gov/GenomeTVLive/. See various additional details below, along with other information items that I hope will be of interest to you.

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

- NHGRI Genome Sequencing Program: Blazing the Path Forward in Human Disease Genomics
- New Online eLearning Tool: In and Beyond Africa
- DNA Day 2016 Essay Contest Now Open
- Publication from the IGNITE Network
- NHGRI Welcomes the New Chief Grants Management Officer

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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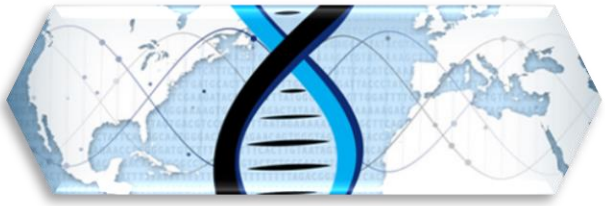
NHGRI Genome Sequencing Program: Blazing the Path Forward in Human Disease Genomics

A few weeks ago, NHGRI renewed its Genome Sequencing Program (GSP), the Institute's largest single grant program that has its origins in the Human Genome Project. Since NHGRI grants are always awarded for a limited number of years at a time, the GSP has experienced a number of 'renewals' – and so this renewal could mistakenly seem like a simple and mindless process. But rest assured, this was not the case!

Genomics is a fast-paced field, with new technologies and approaches being continuously developed. We see the renewal of a program as an opportunity to reformulate and recast our scientific goals to keep up with these developments. So, what is the overarching goal for our renewed GSP? – advancing human disease genomics.

For many years, one of the main goals of the GSP has been to implement new DNA sequencing technologies at larger scale, increase genome-sequencing capacity, and decrease genome-sequencing costs. It was not that long ago when only one human genome sequence was being generated in the world (by the Human Genome Project), and its cost was quite high. Last year, our GSP produced 1.3 Terabases (1,300,000,000,000,000 bases) of DNA sequence (the equivalent of 14,000 human genome sequences), with the associated costs per human genome sequence approaching $1,000. With such a capability in terms of cost-effective data production, it now seems like the right time for the GSP to focus most of its attention on studying human diseases.



Common diseases, such as diabetes and asthma, affect hundreds of millions of people worldwide. Unfortunately, such disorders are quite complicated and require very large studies (involving thousands of affected and unaffected individuals) to identify and tease apart the complex set of genomic and environmental factors contributing to disease. To address this, NHGRI is funding four research centers, the Centers for Common Disease Genomics (CCDGs), for a total of ~$240 million over the next four years. CCDG scientists will be generating genome sequences for close to 200,000 individuals, initially focusing on cardiovascular/metabolic and neuropsychiatric diseases. Through these efforts, CCDG researchers aim to develop better approaches for using genome sequencing to study common diseases more broadly.

The broader GSP will also include a continuation of the Centers for Mendelian Genomics (CMGs), which aim to identify the mutated genes underlying rare inherited (or Mendelian) diseases. Mendelian diseases, named after Gregor Mendel (the founder of modern genetics), are typically transmitted from parents to offspring and tend to be caused by a

New Online eLearning Tool: In and Beyond Africa

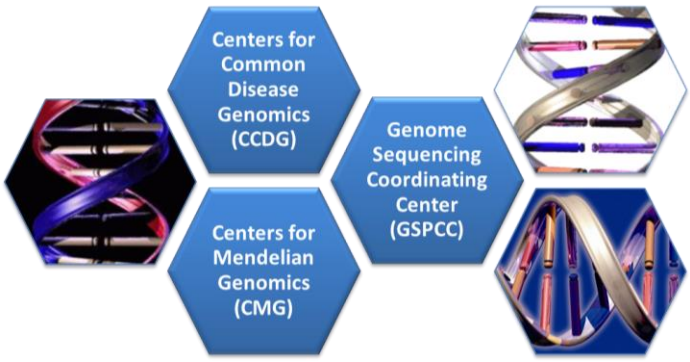
A new online eLearning tool that expands upon one component of the *Genome: Unlocking Life's Code* exhibition was recently released. This interactive tool – "In and Beyond Africa" – allows users to explore migration patterns that influenced human genomic diversity on Earth. The tool contains a timeline that tracks human migration out of Africa from ~200,000 years ago to today, and includes five games to engage people. To learn more, visit unlockinglifescode.org/iaba.

DNA Day 2016 Essay Contest Now Open

The American Society of Human Genetics (ASHG) invites students in grades 9-12 to participate in the 11th Annual DNA Day Essay Contest. Students are asked to choose a currently available genetic test for a condition or disease that does not cause symptoms until adulthood, describe how the test works and the reliability of the test results, and defend or refute the recent recommendation from ASHG's position statement on pediatric genetic testing (as it pertains to that genetic test). For more information on the Annual DNA Day Essay Contest, visit ashg.org/education/dnaday.shtml. For further details on NHGRI's DNA Day 2016 plans, be sure to read next month's issue of *The Genomics Landscape*.

genomic defect in a single gene. Of note, I highlighted recent accomplishments of this four-year program in the August 2015 issue of *The Genomics Landscape*. With ~$40 million of funding over the next four years, the CMGs will continue their work to find the responsible genes, improve gene-discovery methods, coordinate with analogous efforts in other countries, and share resources with the broader scientific community.

With the heavy emphasis of the GSP on studying human disease, one might ask about genomic studies of cancer? The NHGRI GSP has extensive experience in using genomics to study cancer through the very successful The Cancer Genome Atlas (TCGA) effort. TCGA is a partnership between NHGRI and the National Cancer Institute (NCI), and will come to completion later this year. The current phase of GSP will shift away from cancer studies in order to focus on other disease areas; meanwhile, NCI is actively and appropriately expanding its cancer genomics programs. The NHGRI GSP will continue working with other NIH partners – for example, the National Heart, Lung, and Blood Institute (NHLBI) will contribute to both the CCDG and CMG components, while the National Eye Institute (NEI) will contribute to the CMG components. Eventually, we anticipate that other NIH institutes and centers will collaborate with the NHGRI GSP in disease areas of interest.



In order to properly support the CCDG and CMG components of the GSP, we have also started a GSP Coordinating Center (GSPCC) funded with ~$4 million over four years. The GSPCC will facilitate interactions among GSP investigators and contribute to data analysis and program outreach. Another way that NHGRI plans to recruit 'new' and creative minds to the GSP is through Genome Sequencing Program Analysis Centers (GSPAC). These groups will be funded later this year to contribute to the analyses of the data generated by the GSP.

Since the early 1990's, I have witnessed the GSP evolve from sequencing the first human genome, to conducting large comparative genomics studies, to generating vast catalogs of human genomic variants, and, most recently, to developing new genomic approaches for studying human disease. Through these efforts, genome sequencing has become a fundamental tool for biomedical research. I am profoundly excited to now watch the next four years of the NHGRI GSP, as it advances our ability to use genome sequencing to unravel the molecular underpinnings of rare and common diseases.

Publication from the IGNITE Network

The NHGRI-funded IGNITE Network recently published its first network-wide publication, entitled "The IGNITE Network: A model for genomic medicine implementation and research." The IGNITE Network was established in 2013, and is comprised of six projects and a coordinating center. The Network supports the development, evaluation, and dissemination of tools designed to facilitate the use of genomic information in healthcare settings. This paper describes the organization, methods, and goals of the entire IGNITE Network and of each individual project.

NHGRI Welcomes the New Chief Grants Management Officer

NIH Grants and Funding

Last month, Ms. Deanna Ingersoll was recruited to NHGRI to become our new Chief Grants Management Officer. Ms. Ingersoll comes to NHGRI from the National Institute of Allergy and Infectious Diseases (NIAID), where she served as a Lead Grants Management Specialist for the past 11 years. In her new role at NHGRI, she will review and award research grants, conduct appropriate audits to ensure compliance with relevant policies, and lead the Grants Administration Branch. NHGRI is excited to welcome Ms. Ingersoll to our Institute!

Spotlight on the Precision Medicine Initiative (PMI)

nih.gov/precisionmedicine

- A video recording of the recent PMI Cohort Program Advisory Panel Meeting is now available.
- The *New York Times* article, "New Guidelines Nudge Doctors to Give Patients Access to Medical Records," provides an overview of the new guidelines issued by the U.S. Department of Health & Human Services on individuals' rights under the Health Insurance Portability and Accountability Act (HIPAA) to access their health information.
- A video recording of the NIH pre-application technical assistance webinar regarding PMI Cohort Program funding opportunities is now available.

NIH/NHGRI News of Interest

Happy New Year … and a Look Back at a Memorable 2015

Pi Day 2016 Schedule and Events

Latest Advances In Gene Editing: The Possibilities And Risks

Taking Control: Learn More About Accessing Your Health Information

What's Next for the National Institutes of Health?

Unlocking Life's Code: January 2016 Newsletter

People Behind the Science Podcast: Eric Green

Register Now for Rare Disease Day at NIH on February 29

Cancer's on the March but 'We've Got Its Number': NIH Director

The National Cancer Moonshot Begins to Take Shape

Genome Advance of the Month

Gene-Editing Technology Harnessed to Protect Plants from Viruses

The "Bunny Ear" Hypothesis: How Defective DNA Looping May Contribute to Cancer

Genomics Research

Researchers Discover Three Glaucoma-Related Genes

Vanderbilt Study Raises Questions about Reporting Incidental Genetic Findings

Schizophrenia's Strongest Known Genetic Risk Deconstructed

Gene Expression Test Aims to Reduce Antibiotic Overuse

Funding News

Legislative Mandates in Effect for FY 2016

Salary Limitation on Grants, Cooperative Agreements, and Contracts

Notice Clarifying SBIR Direct-to-Phase II Eligibility Criteria

Applicant Information Webinar for NHGRI ENCODE RFAs

Fellowship Opportunities

The Genetics & Public Policy Fellowship

The Genetics & Education Fellowship

Upcoming Webinars

Seventy-Sixth Meeting: National Advisory Council for Human Genome Research – February 8, 2016

Funding Opportunities

Expanding the Encyclopedia of DNA Elements (ENCODE) in the Human and Mouse

Characterizing the Functional Elements in the ENCODE Catalog

Computational Analysis of the ENCODE Data

ENCODE Data Coordination Center

ENCODE Data Analysis Center

International Research Ethics Education and Curriculum Development Award

International Bioethics Research Training Program

Genome Sequencing Center for the Gabriella Miller Kids First Pediatric Research Program

NIH Pathway to Independence Award

Administrative Supplements for Research on Sex/Gender Differences

Initiative to Maximize Research Education in Genomics: Courses for Skills Development