November 3, 2015

This month, we celebrate National Family History Day. On Thanksgiving, the U.S. Surgeon General encourages you to talk with your family about your family health history. The "My Family Health Portrait" online tool can help you organize your family health history. You can even print out the compiled information to share with family members and your doctor(s). I hope you will take a moment to talk with your family about this important topic – this month or any time of the year.

In this month's *The Genomics Landscape*, I highlight the 1000 Genomes Project, which came to a close last month. See various details below, along with other information items that I hope will be of interest to you.

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

- 1000 Genomes Project
- TCGA Awarded 2015 Service to America Medals People's Choice Award
- Dr. Tom Insel Departs as NIMH Director
- New NIH Deputy Director for Extramural Research
- New NHGRI Intramural Investigator

All the best,

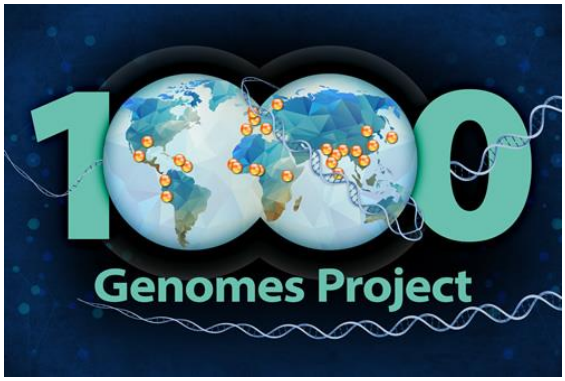
Watch here for current and upcoming locations of the Smithsonian-NHGRI exhibition "Genome: Unlocking Life's Code" as it tours North America!

~To manage your subscription to *The Genomics Landscape*, see: list.nih.gov/cgi-bin/wa.exe?A0=NHGRILANDSCAPE~
~To suggest future topics, send an e-mail to: NHGRILANDSCAPE@MAIL.NIH.GOV~
~To access past editions, see: genome.gov/27541196~

1000 Genomes Project

In October, NHGRI and its international collaborators successfully brought the 1000 Genomes Project to completion. As an important sequel to the Human Genome Project, this impressive effort initially aimed to identify and catalog 95% of the common human genomic variants (specifically, those 'DNA spelling differences' with a frequency of at least 1%). At its conclusion, the 1000 Genomes Project catalogued >99% of all common human genomic variants as well as many rare variants. The resulting resource is proving immensely valuable for studies that relate genomic variation to health and disease.

The international team of scientists working on the 1000 Genomes Project sequenced the genomes of 2,504 people from 26 populations around the world from five ancestral groups (West African, East Asian, South Asian, European, and the Americas). This reflects over 50% more sequenced genomes than originally proposed for the project– a massive overachievement! The resulting data – all of which has been made freely available – represent the world's largest catalog of genomic differences among humans.



Detailed analyses of the resulting >2,500 genome sequences led to the cataloguing of the different classes of genomic variants: single DNA base differences (81 million single-nucleotide polymorphisms, or SNPs), insertions and deletions (3.4 million), and larger structural polymorphisms (68 thousand). In total, the Project identified >84 million variant sites in the human genome.

To document the above accomplishments, the 1000 Genomes Project scientists published two papers in *Nature*: *A global reference for human genetic variation* and *An integrated map of structural variation in 2,504 human genomes*. Importantly, the 1000 Genomes Project Consortium was truly international in nature, including investigators from across the U.S., China, Germany, and the United Kingdom. 1000 Genomes Project funding from NHGRI began in Fiscal Year 2008, and has now concluded. The project was co-funded and co-led by the Wellcome Trust in the United Kingdom.

TCGA Awarded 2015 Service to America Medals People's Choice Award

The Cancer Genome Atlas (TCGA) Project Team, led by Dr. Carolyn Hutter from NHGRI and Dr. Jean Claude Zenklusen from the National Cancer Institute, has been awarded the 2015 Samuel J. Heyman Service to America Medals People's Choice Award. These prestigious awards, nicknamed "the SAMMIES," are presented annually by the nonprofit, nonpartisan Partnership for Public Service to celebrate excellence in federal civil service. The TCGA team analyzed thousands of genome sequences from more than thirty types of cancer, advancing research into the diagnosis, treatment, and prevention of these deadly diseases. For further details, see servicetoamericamedals.org/honorees/view_profile.php?profile=423.

Dr. Tom Insel Departs as NIMH Director

After 13 years as Director of the National Institute of Mental Health (NIMH), Dr. Tom Insel departed NIH on November 1. He will join the Google Life Sciences team at Alphabet (formerly Google) to lead a new effort that will focus on mental health. Throughout his tenure at NIMH, Tom worked closely with NHGRI on Common Fund initiatives such as the Molecular Libraries Program and the Genotype-Tissue Expression Project. He also served as the Acting Director of the National Center for Advancing Translational Sciences until its permanent Director was appointed. Tom is a valued colleague and friend, and we wish him well in the next step of his professional journey! For more information, see nimh.nih.gov/about/dr-tom-insel-to-step-down-as-nimh-director.shtml.

1000 Genomes Project data are proving to be highly valuable for studying both rare and common human diseases. For example, the data help investigators establish the frequency of any genomic variant that they encounter. These data are also powerful for studying the demographics and history of human populations. To aid all such studies, researchers can obtain DNA and cell lines from 1000 Genome Project participants through the NHGRI Sample Repository for Human Genetic Research at the Coriell Institute for Medical Research.

The 1000 Genomes Project is a stellar example of gifted scientific minds from across the globe coming together to create a valuable resource for the biomedical research community. The two recent *Nature* papers nicely showcase this resource, which is now actively used worldwide to advance understanding about the role of genomic variants in health and disease.

All 1000 Genomes Project data are available at 1000genomes.org. For the recent press release regarding the two *Nature* papers, see genome.gov/27562711. NHGRI's 1000 Genomes web site can be found at genome.gov/27528684.

New NIH Deputy Director for Extramural Research

NIH National Institutes of Health
Office of Extramural Research

Dr. Michael Lauer has been selected as the new NIH Deputy Director for Extramural Research. Dr. Lauer is a board-certified cardiologist who joined NIH in 2007 as a division director at the National Heart, Lung, and Blood Institute. I have worked closely with him on several trans-NIH initiatives, and I look forward to NHGRI interacting with him more broadly in this critical new capacity. For more information and the full announcement, visit nih.gov/about/director/20150928-statement-michael-lauer.htm.

New NHGRI Intramural Investigator

NHGRI is pleased to welcome Dr. Adam Phillippy as the newest Investigator with its Intramural Research Program. Dr. Phillippy joins NHGRI from the National Biodefense Analysis and Countermeasures Center in Frederick, MD, where he founded the bioinformatics group and led the development of new, single-molecule-based methods for microbial forensics and outbreak investigation. He received his Ph.D. in computer science from the University of Maryland under Dr. Steven Salzberg's supervision, where he investigated nucleotide probe and sequencing-based methods for pathogen detection. Dr. Phillippy's laboratory is now part of the NHGRI Computational and Statistical Genomics Branch. His NHGRI research will focus on the design and application of efficient algorithms for the analysis of large-scale genomic data, relevant to *de novo* genome sequencing, variant detection, outbreak detection, and metagenomics. For more information on the NHGRI Intramural Research Program, visit genome.gov/dir.

Spotlight on the Precision Medicine Initiative (PMI)

nih.gov/precisionmedicine

- NIH Guide Notice: Precision Medicine Initiative Cohort Program Recommendations Issued and Accepted
- The White House has posted a series of stories featuring people who have benefited from precision medicine in various ways.
- To receive regular updates from the White House on ways to become involved with the Precision Medicine Initiative, visit their PMI page.

Genomics Research

Gene Therapy Staves Off Blindness from Retinitis Pigmentosa in Canine Model

Scientists Develop Genetic Blueprint of Inner Ear Cell Development

Nuclear Transport Problems Linked to ALS and FTD

Researchers Identify Potential Alternative to CRISPR-Cas Genome Editing Tools

Genomic Analysis Paves Way for Personalized Treatment of Invasive Lobular Carcinoma

The Autism Science Foundation Launches the Autism Sisters Project, to Search for Autisms' "Female Protective Effect"

A Newly-Discovered Tumor Suppressor Gene Affects Melanoma Survival

NIH Researchers Link Single Gene Variation to Obesity

Genome Advance of the Month

Women with Inherited Breast Cancer Risks Face Numerous Challenges

Upcoming Webcasts/Webinars

Common Rule Webinar Series

What You Need to Know About NIH Application Submission and Review

New Genomics Videos

Integrating Genomic Sequencing into Clinical Care: CSER and Beyond

Funding Opportunities

Novel Nucleic Acid Sequencing Technology Development (Direct to Phase II R44)

Novel Genomic Technology Development (R01, R21, R43/R44 and Direct to Phase II SBIR R44)

Limited Competition: Knockout Mouse Phenotyping Project Database (UM1)

Limited Competition: Knockout Mouse Production and Phenotyping Project (UM1)

2016 NIH Director's Early Independence Awards

New Prize Competition Seeks Innovative Ideas to Advance Open Science

Notice of Intent to Publish Funding Opportunity Announcements

Functional Element Mapping Centers (UM1)

Functional Element Characterization Centers (UM1)

Computational Analysis Research Projects (U01)

ENCODE Data Coordination and Analysis Center (U24)

Funding News

ASSIST Now an Option for Small Business Grant Applications

NIH & AHRQ Announce Upcoming Changes to Policies, Instructions and Forms for 2016 Grant Applications

Implementing Rigor and Transparency in NIH & AHRQ Research Grant Applications and Career Development Award Applications

NIH News of Interest

NIH Operates Under a Continuing Resolution

Dr. Collins Testifies Before the Senate on Investing in a Healthier Future

International Diabetes Research Knowledge Portal Opens to Public, Scientists

Knowledge for the World: Genetics Database Celebrates Golden Anniversary

Analysis of Public Comments on NIH-Wide Strategic Plan Request for Information

Genome: Unlocking Life's Code October Newsletter

NIH Announces Common Fund 2015 High-Risk, High-Reward Research Awardees

Request for Comments

Public Comment Sought: Revised Federal Policy for the Protection of Human Subjects ('Common Rule')–Closes December 7, 2015

NCATS Seeks Input into Strategic Planning Process

Research Terms and Conditions Available for Public Comment

Input on Validation Assays for Affinity Reagents Generated by the NIH Common Fund Protein Capture Reagents Program

Request for Information (RFI): Undiagnosed Diseases Research