

March 3, 2015

Featured in last month's *The Genomics Landscape* was the recently announced Precision Medicine Initiative. On February 11 and 12, the first strategic workshop for this Initiative was held on the NIH campus. Experts from a diverse set of fields came together to discuss building a large U.S. research cohort (see <u>nih.gov/precisionmedicine/workshop.htm</u>). In some ways, this was symbolically the first exciting step of many for the Precision Medicine Initiative journey. The videoarchive of the meeting is now available and can be viewed at:

Day 1: videocast.nih.gov/summary.asp?Live=15751&bhcp=1 Day 2: videocast.nih.gov/summary.asp?Live=15753&bhcp=1

In this month's *The Genomics Landscape*, I discuss the importance of supporting genomics research in Africa and describe NHGRI's involvement in some relevant research programs. See various details below, along with other information items that I hope will be of interest to you.

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

- Genomics in Africa
- <u>Request for Information: National Library of Medicine</u>
- eMERGE Publications Highlight Large-Scale Genomics Research
- ASHG-NHGRI Genetics & Public Policy and Genetics & Education Fellowships
- <u>Recruiting a New Chief, Communications and Public Liaison Branch</u>

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 receive The Genomics Landscape, sign up via: <u>list.nih.gov/cgi-bin/wa.exe?A0=NHGRILANDSCAPE</u>~~
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Genomics in Africa

Roughly 100,000 years ago, humans migrated out of Africa and started to populate the globe. Although the human species is now associated with significant phenotypic variation, all humans have a genetic origin in Africa. The genomes of people currently living in Africa are more varied than those of people living anywhere else on Earth. Studying the genomes of Africans thus offers important opportunities to characterize and understand genomic variation. Despite this, Africans are largely underrepresented as both research participants and scientists in genetics and genomics.



To fully capture the genetic and genomic legacy of humanity, it is essential to include representatives of African populations in studies of human genomic variation. It is also critical that the benefits of genomic advances be felt throughout the world by patients, health professionals, researchers, and scientists. Accordingly, NHGRI aims both to increase the use of African populations in genomic studies and to stimulate genomics research in Africa. This is being accomplished in multiple ways, a few of which are highlighted below.

Building on the success of the <u>HapMap Project</u>, the aim of the <u>1000 Genomes Project</u> is to discover and catalog human genomic variants across the world's populations and to support studies relating those variants to health and disease. As a centerpiece of this effort, researchers have now sequenced the genomes of thousands of people in Africa, Europe, East Asia, South Asia, and the Americas. This project represents a collaboration involving researchers from the U.S., U.K., China, and Germany.



Recently, Dr. Charles Rotimi of the NHGRI Intramural Research Program and his colleagues in the <u>African Genome Variation</u> <u>Project</u> published the first comprehensive characterization of Request for Information: National Library of Medicine



The National Library of Medicine (NLM) has been at the forefront of how biomedical data and health information are collected, shared, and analyzed since its founding in 1836. NLM's National Center for Biotechnology Information, GenBank, and numerous other data resources are vitally important to the field of genomics— and, therefore, of great interest to NHGRI.

After serving as NLM Director for over thirty years, Dr. Don Lindberg will be retiring at the end of March. Prior to launching a search for a new NLM Director, NIH is seeking input from stakeholders and the general public to inform the development of a renewed vision for the NLM. Please see the official RFI Guide Notice at

grants.nih.gov/grants/guide/notice-files/NOT-OD-15-067.html for more details on the information being sought and the means by which to submit comments. The deadline for responses is March 13.

eMERGE Publications Highlight Large-Scale Genomics Research



An important goal of NHGRI is to support research that tackles issues related to using genomic information to care for patients. The Electronic Medical Records and Genomics (eMERGE) Network, now in its eighth year, aims to integrate genomic information into electronic medical records (EMRs) to improve genetic risk assessment, disease prevention, diagnosis, and treatment. Recently, members of eMERGE published a series of nearly 20 papers in *Frontiers in Genetics* highlighting their impressive accomplishments to date. Some of the papers explore research methods or offer reviews, while others report new findings. For more information, see <u>genome.gov/27560569</u>. genomic diversity across sub-Saharan Africa. This study, which used data generated by the HapMap and 1000 Genomes Projects, uncovered evidence of how environmental forces (e.g., climate and exposure to infectious agents) have shaped the genomes of Africans and influenced susceptibility to conditions such as malaria, Lassa fever, and trypanosomiasis.

The Human Heredity and Health in Africa (H3Africa) Initiative is jointly funded by the NIH (through the NIH Common Fund) and the U.K. Wellcome Trust. NHGRI plays a major leadership role in the program, which aims to use genomics to study the genetic and environmental determinants of disease in Africa. H3Africa supports studies led by African scientists that use genomic, clinical, and epidemiologic methods to identify hereditary and environmental contributions to the risk of common, noncommunicable disorders (such as heart and kidney disease), as well as communicable diseases (such as tuberculosis).



An important goal of H3Africa is to increase direct funding available to African scientists, so as to foster genomics research on the continent. This is being accomplished through numerous research projects, collaborative centers, and studies focusing on relevant ethical, legal, and societal issues. The development of suitable infrastructure is also essential for establishing a sustainable genomics research base in Africa. H3Africa is funding grants for the collection and banking of research specimens through biorepository research projects, as well as grants for the development of computing capabilities through the H3ABionet. With the increase in available biorepository space within Africa, it is hoped that biosamples will be stored and catalogued properly to increase the availability of appropriate materials for large-scale genomics research.

I am excited to see what the future holds as African scientists and citizens become more engaged in genomics research. For more information about H3Africa, see genome.gov/27542964. For more information about the African Genome Variation Project publication, see genome.gov/27559699. Of further interest, two articles on the state of science funding in Africa were recently published in *The Scientist*: see "Funding Research in Africa" by Paula Park and "Opinion: 'On Funding Research in <u>Africa'</u>" by Francis Collins and Jeremy Farrar.

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



THE AMERICAN SOCIETY **OF HUMAN GENETICS**

NHGRI and the American Society of Human Genetics (ASHG) are now accepting applications for the 2015 Genetics & Public Policy Fellowship and the 2015 Genetics & Education Fellowship. The Genetics & Public Policy Fellowship is designed to be a bridge for genetics professionals wishing to transition to a policy career. The Genetics & Education Fellowship is designed for genetics professionals with an advanced degree who are early in their careers and interested in developing their expertise in genomic literacy efforts, science education policy, and program development. NHGRI and ASHG are accepting applications for both fellowships until April 24, 2015.

Recruiting a New Chief, Communications and **Public Liaison Branch**



NHGRI is currently recruiting a new Chief of the Communications and Public Liaison Branch (CPLB), part of the Institute's Division of Policy, Communications, and Education (DPCE). The CPLB Chief serves as the overall 'communications' director' for NHGRI, working closely with the DPCE Director and NHGRI Director in dealing with all aspects of the Institute's communications programs. Specifically, the individual is expected to manage and lead communications and public relations activities that are fundamental to communicating genomics research advances as well as Institute goals, policies, programs, and accomplishments. This individual also works in close collaboration with many other parts of NHGRI and NIH. For further information about this important position, see genome.gov/27560456.



Genomics Research

Evolution of Marine Mammals to Water Life Converges in Some Genes

NIH Researchers Reveal Link Between Powerful Gene Regulatory Elements and Autoimmune Diseases

NIH-Supported Researchers Map Epigenome of More Than 100 Tissue and Cell Types

Genetic Studies Yield New Insights into Obesity

DNA Sequencer the Size of a Mobile Phone

<u>New Insights into 3D Genome</u> Organization and Genetic Variability

Predicting Cancers' Cell of Origin

eMERGE Publications Highlight Large-Scale Genomics Research Using Electronic Medical Records

Upcoming Webcast/Webinar

Future Opportunities for ENCODE and Beyond – March 10-11, 2015

Webinar for New NHGRI Postdoctoral Training Programs in Genomic Medicine Research – March 18, 2015

New Videos

National Advisory Council for Human Genome Research – February 9, 2015

Precision Medicine Initiative: Building a Large U.S. Research Cohort – February 11-12, 2015 – <u>Day 1</u> and <u>Day 2</u>

Genome Advance of the Month

<u>CRISPR Probes the Inner Workings of the</u> <u>Genome in Real Time</u>

NIH News of Interest

HHS and NIH Take Steps to Enhance Transparency of Clinical Trial Results

Interview with Dr. Francis Collins on What to Expect from the Precision Medicine Initiative

NIH Announces \$41.5 Million in Funding for Human Placenta Project

NIH Public Access Plan for Increasing Access to Scientific Publications and Digital Scientific Data from NIH Funded Scientific Research

Funding News

NIH Interim General Grant Conditions Implementing New HHS Grants Regulations (Uniform Guidance)

<u>Clarification of Language in RFA-HG-15-</u> 001: Centers for Common Disease <u>Genomics</u>

National Heart, Lung, and Blood Institute Participation in RFA-HG-15-002: Centers for Mendelian Genomics

<u>Clarification for RFA-HG-14-008: Courses</u> <u>for Skills Development in Biomedical Big</u> <u>Data Science</u>

Correction to RFA-MD-15-005: NIH Big Data to Knowledge Enhancing Diversity in Biomedical Data Science

Funding Opportunities

Pathway to Independence K99/R00 Awards

Administrative Supplements to Extend the Scope and Reach of LINCS Datasets

High-End Instrumentation Grant Program

NIH Requests for Feedback

Extension of the Public Comment Period for NOT-OD-15-019: Draft NIH Policy on Dissemination of NIH-Funded Clinical Trial Information

Input into the Deliberations of the Advisory Committee to the NIH Director Working Group on the National Library of Medicine

Input on Sustaining Biomedical Data Repositories

NHLBI Whole Genome Sequencing Project

Strategic Plan for the Office of Research Infrastructure Programs: Office of Science Education/Science Education Partnership

Strategic Plan for the Office of Research Infrastructure Programs: Division of Comparative Medicine and Division of Construction and Instruments Programs

Sustaining the Biomedical Workforce and a Potential Emeritus Award for Senior Researchers

Past editions of *The Genomics Landscape* can be accessed at genome.gov/27541196.

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