June 2, 2015

Two things will 'heat up' over the next month or so: the weather with the start of summer, and some important strategic planning efforts by NHGRI and NIH!

On June 8-9, 2015, an important meeting, entitled *Genomic Medicine 8: NHGRI's Genomic Medicine Programs*, will be held in Rockville, Maryland. The Genomic Medicine 8 meeting will convene leadership from NIH and external groups to examine the NHGRI genomic medicine research portfolio in light of evolving scientific knowledge and emerging opportunities. The meeting will be videocast live (genome.gov/GenomeTVLive/) and later videoarchived on our GenomeTV channel of YouTube. In addition, the Precision Medicine Initiative Working Group of the NIH Advisory Committee to the Director will hold a public workshop on participants and community engagement on July 1-2, 2015. This workshop will also be videocast live (videocast.nih.gov/) and later videoarchived (nih.gov/precisionmedicine). Also see the PMI Spotlight below for further details.

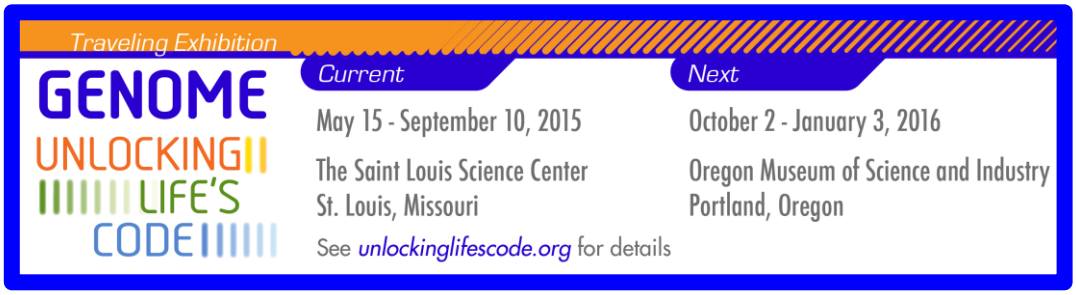
In this month's *The Genomics Landscape*, I highlight the recent publications from the Genotype-Tissue Expression (GTEx) Project. See various details below, along with other information items that I hope will be of interest to you.

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

- Genotype-Tissue Expression (GTEx) Project Reports Research Results
- Clinical Genome Resource (ClinGen) Marker Paper
- Genomics and Health Disparities Lecture Series
- LabTV
- NIH Clinical Center Genomics Collaboration

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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Genotype-Tissue Expression (GTEx) Project Reports Research Results

Some day in the not-too-distant future, many people will have their genomes sequenced as part of their routine medical care. It is the hope of many that such genomic information will lead to improved approaches for preventative medicine and personalized treatments. To realize such a future, a detailed knowledge of the role that individual DNA variants play in genome function is needed. An exciting research endeavor that aims to generate that knowledge is the Genotype-Tissue Expression (GTEx) Project.

Funded since 2010, GTEx is an NIH Common Fund project that aims to provide an information resource about the interplay between genomic variation (i.e., DNA differences among people) and gene activity (i.e., gene expression). Specifically, GTEx researchers study the relationship between particular genomic variants and the expression of genes in different human tissues. Examining how genomic variants influence gene activity will help researchers understand the role that these variants play in common diseases, such as cancer, heart disease, and diabetes.

GTEx scientists are examining more than 30 tissue types collected from deceased donors and organ/tissue transplant patients, creating a robust gene-expression atlas for multiple human tissues. So far, GTEx has collected and analyzed biospecimens from over 800 donors, and is expected to reach its goal of 900 by the end of summer 2015. To help ensure that the consent process and other aspects of the project effectively address the concerns and expectations of the study's participants, GTEx includes an Ethical, Legal, and Social Implications (ELSI) research component.

Last month, GTEx researchers published a major paper in *Science*, examining gene-expression data collected from 1,641 samples across 43 tissues from 175 individuals; these data were generated as part of the pilot phase of GTEx. They looked for

Clinical Genome Resource (ClinGen) Marker Paper

Genomic scientists have made great advances in discovering variants across the human genome. However, there is a lack of accurate, publicly available clinical and health-related information about the role that those variants play in health and disease. To address this, the Clinical Genome Resource (ClinGen) aims to collect human trait and health-related information about genomic variants, develop an approach to identify clinically relevant variants, and establish a knowledgebase about the identified variants. Bringing together this information is essential for realizing the promise of genomic medicine to improve medical care. ClinGen recently published a 'marker paper' describing its overall goals, consortium structure, methodologies, quality standards, data release plans, and progress to date. For additional information, please see genome.gov/27561707.

Genomics and Health Disparities Lecture Series

On May 27, 2015, NHGRI held the inaugural lecture of a new *Genomics and Health Disparities Lecture Series* that aims to explore the role of genomics in achieving health equality. Co-sponsored by several NIH Institutes and the Office of Minority Health at the Food and Drug Administration, the lecture series was established to enhance discussions about how genomics research and technology can affect health disparities. Topics will range from basic science to translational research. The inaugural lecture featured Dr. Carlos Bustamante, who spoke about "Opportunities and Challenges for Health Disparities Research in the Personal Genome Era." For further details, visit genome.gov/27561525.

links between variants at specific genomic locations and the level of activity of genes in particular tissues, finding both that many variants affected activity in all tissues and that some variants affect activity in only one tissue. Two companion papers were also published in *Science*. One examined the effects of protein-truncating variants on gene activity; the other looked for gene-activity patterns associated with age, gender, and other factors.

To date, no project has analyzed genomic variation and gene expression in as many tissues in such a large population. GTEx has established a database and a tissue bank that can be used by researchers around the world for future studies. The data generated link genomic variants associated with disease to gene activity, bringing us one step closer to understanding the biology underlying important human diseases.

GTEx is a prototypic NIH Common Fund project, in that no one NIH institute or center would have likely pursued the project alone. The NIH Common Fund aims to support projects that are of interest to many NIH institutes and centers, as well as their grantees. The GTEx project is funded through the NIH Common Fund and managed by the NIH Office of the Director in partnership with NHGRI, the National Institute of Mental Health (NIMH), the National Heart, Lung, and Blood Institute (NHLBI), the National Cancer Institute (NCI), the National Center for Biotechnology Information (NCBI), the National Institute on Drug Abuse (NIDA), and the National Institute of Neurological Disorders and Stroke (NINDS).

For more information about the recent GTEx publications, see nih.gov/news/health/may2015/nhgri-07.htm. To access GTEx data, see gtexportal.org/home/. Lastly, GTEx recently held a symposium at the NIH in Bethesda, Maryland to showcase its accomplishments. To view a recording of the two-day symposium, "GTEx Symposium: All Things Considered – Biospecimens, 'Omics Data, and Ethical Issues", see videocast.nih.gov/summary.asp?Live=16342&bhcp=1 and videocast.nih.gov/summary.asp?Live=16346&bhcp=1.

LabTV

NIH has partnered with LabTV to produce a series of videos featuring NIH scientists and trainees. LabTV was founded by Jay Walker, the curator and chairman of TEDMED. These videos are being posted online, with the goal of fostering students' interest in pursuing scientific research careers. Personal stories from bright scientific minds describe their paths to success and the challenges along the way, providing models for students as they begin their own professional journeys. Already among the many NIH stories featured on LabTV are four NHGRI scientist profiles. New videos are added monthly. For further information, please see genome.gov/27561075.

NIH Clinical Center Genomics Collaboration

Researchers from NHGRI, the Whitehead Institute, and the NIH Clinical Center have teamed up for a unique collaboration. Their project, entitled "The Next Era of Y-Chromosome Research: Beyond the Reproductive Tract," is part of a new U01 program called "Opportunities for Collaborative Research at the NIH Clinical Center." Lead investigators Max Muenke of NHGRI and David Page of the Whitehead Institute are examining the impact of single-copy genes on the male-specific region of the Y chromosome beyond the reproductive tract, which remains a scientific and medical mystery. They will evaluate patients with naturally occurring Y-chromosome anomalies, employing systematic multi-system phenotyping available at the NIH Clinical Center. These patients will also undergo multi-level molecular phenotyping in the Page laboratory to detect perturbations resulting from the disruption of broadly expressed genes on the male-specific region of the Y chromosome. To read more about the new program that made this innovative collaboration possible, see genome.gov/27557011.

Spotlight on the Precision Medicine Initiative

- On April 28-29, 2015, experts and members of the public met to discuss unique scientific opportunities for the Precision Medicine Initiative's national research cohort. The workshop summary and video can be viewed here: nih.gov/precisionmedicine/workshop-20150428.htm. On May 28-29, 2015, there was a public workshop on cohorts and electronic health records. Video for that workshop can be found here: nih.gov/precisionmedicine/workshop-20150528.htm.
- The White House is looking for a Champion of Change for Precision Medicine. For further details and to submit a nomination, visit whitehouse.gov/blog/2015/05/18/nominate-white-house-champion-change-precision-medicine.
- An NIH collection of news articles related to the Precision Medicine Initiative is available at nih.gov/precisionmedicine/inthenews.htm.
- On July 1-2, 2015, tune in live to watch the Precision Medicine Initiative's public workshop on participants and community engagement at videocast.nih.gov/.
- The NIH just released a Request for Information on Community Engagement and Health Disparities for the Precision Medicine Initiative. It is open for responses until June 19. Please see grants.nih.gov/grants/guide/notice-files/NOT-OD-15-107.html.

Genome Advance of the Month

Scientists Discover Topical Insights into the Effects of Sun Exposure on Skin

Upcoming Webcast

Genomic Medicine 8: NHGRI's Genomic Medicine Portfolio

New Videos

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

GTEx Symposium: All Things Considered – Biospecimens, 'Omics Data, and Ethical Issues – May 20 (Day 1) & 21 (Day 2), 2015

David Page – Lost in Translation: Do Males and Females Read their Genomes Differently

Carlos Bustamante – Opportunities and Challenges for Health Disparities Research in the Personal Genome Era

Genomics Research

Genomic Knowledge is Power in the Fight Against Obesity

GTEx Findings Reveal New Insights into How DNA Differences Influence Gene Activity, Disease Susceptibility

Scientists Create Mice with a Major Genetic Cause of ALS and FTD

Hitting the Right Target? Lab Studies Suggest Epigenetic Drug May Fight Childhood Brain Cancer

Creative Minds: Building a Better Electronic Health Record

ClinGen Setting Standards for When Genes and their Variants Matter in Disease

Congressional Action Needed to Optimize Regulation of Genomic Tests

New Platforms Genetically Barcode Tens of Thousands of Cells at a Time

Single-Cell Analysis Hits its Stride

U.S. Introduces New DNA Standard for Ensuring Accuracy of Genetic Tests

NIH News of Interest

A Note on Genome Editing

Work of Cancer Genome Atlas Leads to Breakthroughs in Cancer Diagnosis, Treatment

NCI-MATCH Trial Will Link Targeted Cancer Drugs to Gene Abnormalities

Funding & Resource Opportunities

Discovery of the Genetic Basis of Structural Birth Defects and of Childhood Cancers: Gabriella Miller Kids First Pediatric Research Program

New Educational Resources

What Do You Think? An Interactive Learning Tool

Human Identity Lesson Plan