



December 10, 2013

Welcome to the December issue of *The Genomics Landscape*, a monthly email message that aims to disseminate information directly from the NHGRI Director to the broader genomics community and other interested recipients.

Featured in this month's *The Genomics Landscape* is:

- [Jumping into the Deep End of Genomic Medicine](#)
- [FDA Clears Next-Generation Genome Sequencer for Routine Medical Care](#)
- [Celebrating 20 Years of the NHGRI Intramural Research Program](#)
- [Genome Medicine VI Meeting](#)

You received this email, along with others in October and November, because we have identified you as part of the 'NHGRI constituency.' **However, this is the last month that you will receive *The Genomics Landscape* automatically; if you wish to continue receiving this monthly email, you need to 'opt in' through a simple sign-up procedure (see below)...unless you have already done so.** Also below, you will find an email address for sending me suggestions for topics to discuss in the future.

Finally, I would like to wish you and your family a safe and happy holiday season. I look forward to communicating with you in 2014!

All the best,

A handwritten signature in black ink, appearing to read 'Eric'.

To continue receiving *The Genomics Landscape* each month,
you must sign up via the following ListServ:
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To suggest topics for future messages, please send an e-mail to:
NHGRILANDSCAPE@MAIL.NIH.GOV

Jumping into the Deep End of Genomic Medicine

Genomic Medicine: An emerging medical discipline that involves using genomic information about patients as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the other implications of that clinical use.

When NHGRI published its new strategic vision for genomics ([Charting a course for genomic medicine from base pairs to bedside](#)) in 2011, we recognized that the Institute had a lot to learn about the research needed to apply genomics to clinical care. At the same time, it seemed critical that we begin to establish a foundation of research programs that would facilitate the implementation of genomic medicine, so we decided to jump in and start swimming!

The 2011 plan outlined areas for research and development in diagnostics, therapeutics, and clinical informatics [including electronic medical records (EMRs)]. A prototypic project for the latter is our Electronic Medical Records and Genomics ([eMERGE](#)) Network. Launched in 2007, eMERGE aims to integrate genomic information into EMRs for use in clinical care to improve genetic risk assessment, disease prevention, diagnosis, and treatment. A newer effort is our [Clinical Sequence Exploratory Research](#) (CSER) program, a component of NHGRI's [Genome Sequencing Program](#). CSER is investigating the technical, practical, and ethical issues involved in implementing genome sequencing in clinical practice.

Following the publication of the 2011 plan, NHGRI has sponsored a series of [Genomic Medicine Meetings](#) to bring together genomic researchers, clinicians, and others to identify research opportunities, share implementation approaches, facilitate the development of an active research community, and foster collaborations. To date, five such gatherings have helped us to formulate ideas about the research and policy issues relevant to the implementation of genomic medicine. They have also helped our Extramural Research Program formulate plans for new research initiatives.

I would like to highlight three recently launched genomic medicine research programs among our new initiatives: the [Implementing GeNomics Into Clinical PracTicE](#) (IGNITE) Network, the Genomic Sequencing and Newborn Screening Disorders Program, and the Clinical Genome Resource (ClinGen) Program.

The [IGNITE Network](#) aims to enhance the use of genomic medicine by exploring methods for effective implementation, diffusion, and sustainability of genomic medicine in diverse clinical settings. Three pilot projects and one coordinating center are currently funded. The pilot projects address different topics: implementation of genomic risk information for kidney disease in an urban setting, utility of family health history, and using pharmacogenomics information to inform treatment. Each project will incorporate genomic information into EMRs and provide

FDA Clears Next-Generation Genome Sequencer for Routine Medical Care



U.S. Food and Drug Administration
Protecting and Promoting Your Health

In mid-November, the FDA announced that it would allow marketing of four “next generation” genome sequencing devices/applications for clinical use. These instruments, reagents, and test systems allow clinical laboratories to sequence a patient’s DNA as well as enable clinicians to take a comprehensive look at a patient’s genome to diagnose disease or identify the potential cause(s) of symptoms. To read about this FDA clearance, see genome.gov/27555431.

Celebrating 20 Years of the NHGRI Intramural Research Program



Established in 1993, the NHGRI Intramural Research Program (IRP) was designed to serve as the focal point for genetics and genomics research at NIH and worldwide. The NHGRI IRP is dedicated to utilizing genomics to transform our understanding of biology and to use that information for improving human health. To learn more about the IRP and its 20 remarkable years of research, please visit genome.gov/DIR.

clinical decision support for appropriate interventions or clinical advice. I am excited that this program aims to push the clinical/research boundary further toward the realm of clinical implementation, and I am hopeful that it will provide a solid research base that will contribute to making the incorporation of genomics into medical care a routine event.

Also recently, NHGRI joined the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) in funding four grants under the Genomic Sequencing and Newborn Screening Disorders research program. This program aims to examine the potential of genome sequencing for expanding and improving the care of newborns. A central question for the program is: can sequencing of newborns' genomes provide useful medical information beyond what current newborn screening already provides? Each of the studies includes three components: genomic sequencing and analysis; research related to patient care; and the ethical, legal, and social implications of using genomic information in the newborn period. These studies will be conducted in a number of different populations and environments, including neonatal intensive care patients, healthy newborns, and those with diagnosed conditions (whether included in current newborn screening panels or not). Teams of researchers will work to further the understanding of disorders that appear in newborns and to improve diagnoses and treatments for these diseases using genomic information.

While I am gratified that more and more medical and research centers are sequencing patients' genomes, doctors still struggle to know which genomic variants are relevant to disease and to clinical care. In part, this is due to the lack of a freely available, comprehensive knowledgebase that captures genomic variants, their phenotypic and functional effects, and other relevant clinical information. To address this issue, NHGRI has established a consortium of research groups in collaboration with the National Center for Biotechnology Information's ClinVar database to develop the Clinical Genome Resource (ClinGen), which will facilitate clinical laboratories' submission of disease-associated human genomic variants and related data into [ClinVar](#). Experts within ClinGen will then curate these variants with additional clinical and functional data, and will develop a consensus process to identify clinically relevant variants. The variants and their supporting evidence will be available to clinicians and professional organizations through ClinVar and the ClinGen Resource.

In total, these three new programs represent ~\$11 million of the NHGRI Extramural Research Program's Fiscal Year 2013 budget of ~\$341 million. Importantly, this modest investment has enabled NHGRI to jump into a number of important and exciting areas of genomic medicine implementation in a meaningful way.

For more information on these Genomic Medicine Programs, see:

Implementing GeNomics Into Clinical PracTicE (IGNITE) Network genome.gov/27554185

Genomic Sequencing and Newborn Screening Disorders Program genome.gov/27554919

Clinical Genome Resource (ClinGen) Program nih.gov/news/health/sep2013/nhgri-25.htm

Genomic Medicine VI Meeting



The 6th NHGRI-funded meeting on genomic medicine (aka 'Genomic Medicine VI') will be held on January 8-9 in Washington, D.C. This meeting will bring together international leaders in genomic medicine research and implementation. Tune into [Genome TV](#) to watch the live webcast. For additional information about these meetings, see genome.gov/27549225.



New Funding Opportunities

Analysis of Genome-Wide Gene-Environment (G x E) Interactions (R21)

<http://grants.nih.gov/grants/guide/pa-files/PAR-13-382.html>

BD2K-LINCS-Perturbation Data Coordination and Integration Center (DCIC) (U54)

<http://grants.nih.gov/grants/guide/rfa-files/RFA-HG-14-001.html>

Genome Advance of the Month

To sequence the exome or the genome: that is the question genome.gov/27555511

New Genomics Videos

C-SPAN|Washington Journal interview of Dr. Eric Green “Human Genome Research at the NIH”

<http://www.c-spanvideo.org/program/316589-5>

TEDMED Great Challenges – Genomics and Medicine: Where Promise Meets Clinical Practice youtube.com/watch?v=-VdRMFuB5vo

Practicing Precision Medicine in Cancer Using Genomics youtube.com/watch?v=QF6sSkOU7Ro

EMBO|EMBL Science & Society Conference on Public & Private Health Interview of Dr. Eric Green events.embo.org/science-society-conference/videos.html

Genomics News of Interest

NIH deposits first batch of genomic data for Alzheimer's disease genome.gov/27555537

Researchers identify genomic variant associated with sun sensitivity, freckles genome.gov/27555432

NHGRI spotlights intramural findings at NIH Research Festival genome.gov/27555419

NHGRI funds new CEER grants genome.gov/27555409

Past editions of *The Genomics Landscape* can be accessed at genome.gov/27527308.

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