

May 6, 2014

I hope everyone is enjoying the start of spring, after what has been (for many of us) a long and chilly winter. This is a busy time of year at NHGRI, and so I have no shortage of updates for you. On the programmatic front, I am eager to share information about our recently reconfigured extramural training and mentored career development programs. The new programs expand into genomic medicine, while continuing a focus on basic genome science.

May's The Genomics Landscape features stories about:

- NHGRI's Research Training and Career Development: Genome Science to Genomic Medicine
- NHGRI's \$1000 Genome Technology Development Program
- Incidental Findings in Clinical Genome Sequencing Workshop
- Global Alliance for Genomics and Health

All the best,

Si.

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NHGRI's Research Training and Career Development: Genome Science to Genomic Medicine

The fast-paced nature of genomics provides seemingly endless opportunities to pursue exciting research. While invigorating, this presents challenges when it comes to ensuring the presence of a strong pool of future researchers and providing genomics expertise to individuals at different points in their scientific careers. Providing genomics training is thus an important component of NHGRI's mission. How does NHGRI's Extramural Research Program prioritize its training efforts? How do we ensure that the appropriate expertise is available to the researchers who will solve complex genomic problems and bring genomics to medical care?



For many years, NHGRI has supported training programs in basic genome science and computational biology, areas that were essential to the completion of the Human Genome Project and the genomics efforts that immediately followed. To keep pace with our expanding genomic research interests, we outlined broad expectations for training in NHGRI's 2011 strategic plan, "Charting a course for genomic medicine from base pairs to bedside." So, after a number of planning discussions, NHGRI has designed ways to address these needs, even in the face of constrained budgets.

At a strategic planning workshop for training, held in April 2013, there was broad support for NHGRI to continue its training efforts in basic genome science, with particular emphasis on bioinformatics, biostatistics, and the quantitative and computational sciences. There was also considerable interest in seeing an expansion in the training and mentored career development programs, so as to encompass genomic medicine. The workshop recommended four initiatives that would work synergistically to create such a complete extramural training portfolio for NHGRI. Our recent Program Announcements (outlined below) describe the specific programs that we have designed to modernize that portfolio.

The first is a program to train graduate students and postdoctoral fellows in genomics; the second is a postdoctoral program to train those with doctoral and/or medical degrees in genomic medicine; the third is an individual mentored career development program for those with a doctoral degree and/or medical degree to cross-train in another discipline relevant to genomics; and the fourth is an individual mentored career

NHGRI's \$1000 Genome Technology Development Program



In March, *Nature* published a feature article about the NHGRI DNA sequencing technology development ('\$1000 genome') program. There was also an accompanying editorial in the same issue. These articles accurately describe and credit what has arguably been the most successful technology-development program in the history of NIH (not just NHGRI!). The main article also nicely and appropriately credits NHGRI's Jeff Schloss for his outstanding leadership of the program since its inception. To view the main article, see

<u>nature.com/news/technology-the-1-000-</u> <u>genome-1.14901</u>. To view the accompanying editorial, see

nature.com/news/how-to-get-ahead-1.14891. Additionally, Jeff was recently interviewed by *Techonomy* as the "Government's \$1,000 Genome Man." To read that interview, see techonomy.com/2014/03/talking-governments-1000-genome-man/.

Incidental Findings in Clinical Genome Sequencing Workshop



On February 18-19, NHGRI convened experts to discuss the potential research implications of recommendations and guidelines about the management of incidental findings associated with clinical exome and genome sequencing. The objectives of the workshop were to: (1) discuss key questions and challenges in determining the role (if any) of clinical recommendations in shaping research practices [using the recent <u>American College of Medical</u> <u>Genetics and Genomics (ACMG)</u>

<u>recommendations</u> as a point of departure] and (2) inform NHGRI about the possible development of a normative and scientific research agenda and a policy agenda relating to the return of incidental findings in clinical and development program in genomic medicine for those with a medical degree or M.D.-Ph.D. These four programs are further detailed below:

- Institutional Training Program in Genomic Sciences for Graduate Students and Postdoctoral Fellows (T32) – This program aims to develop leaders in genomic science by expanding the trainees' knowledge and skill set in the quantitative and informational sciences. Cross-training can include clinical discovery work and technology development.
- Institutional Training Program for Postdoctoral Fellows in Genomic Medicine (T32) – This <u>new</u> program targets the development of leaders in genomic medicine. The program will provide clinicians training to gain a skill set and knowledge in genomics. To create this new initiative, we have expanded our previous T32 program to support training in genomic medicine. The program is targeted to M.D. or clinical Ph.D. postdoctoral fellows.
- Individual Mentored Career Award in Genomic Sciences

 (K01) The objective of this program is to develop leaders in genomic sciences by cross-training investigators in other relevant scientific disciplines. For this component of our extramural training portfolio, an emphasis is expected on the quantitative and data sciences.
- Individual Mentored Clinical Scientist Career Award in <u>Genomic Medicine</u> (K08) – The objective of this <u>new</u> program is to develop leaders in genomic medicine who have M.D. or Ph.D. degrees and who wish to pursue careers in genomic medicine research by gaining a skill set and knowledge in genomics, bioinformatics, and the quantitative sciences.



Through these programs, NHGRI hopes to bring cross-training opportunities to individuals at different career levels and to support the training of investigators working in both basic genome science and genomic medicine. Creating leaders in both of these research areas is essential to realizing the full potential of genomics, and this new suite of programs promises to move us closer to that realization. For more information about NHGRI's extramural training programs, see genome.gov/10000950. research settings. Workshop attendees represented academia, government, healthcare professionals, institutional review boards, the Presidential Commission for the Study of Bioethical Issues, professional societies, and the UK Biobank. Materials from the workshop, including a summary of the panel presentations and subsequent discussions, are available at genome.gov/27556768.

Global Alliance for Genomics and Health



The Global Alliance for Genomics and Health (GA4GH) is a recently established, international, not-for-profit coalition. Its goal is to formulate standards and a framework for enhanced worldwide data sharing. Over 150 organizations across the world are involved in this new initiative, including research funding agencies, healthcare providers, research institutions, disease advocacy organizations, and private companies. The Steering Committee for GA4GH is led by Dr. David Altshuler (Broad Institute, Harvard Medical School, and Massachusetts General Hospital).

The first face-to-face meeting of the GA4GH took place in March at the Wellcome Trust in the UK, with eight NIH representatives attending. Several working groups presented updates on their progress in areas such as data, security, regulatory and ethical considerations, and clinical data. Issues related to consent and privacy will be examined by the GA4GH, and global standards will be developed in several areas. For more information, visit genomicsandhealth.org.



Genomics Research Findings

NIH-Funded Atlas Details Gene Activity of the Prenatal Human Brain

Experts Decode Germs' DNA to Fight Food Poisoning

Defining Functional DNA Elements in the Human Genome

<u>Genomics' Daunting Challenge: Identifying Variants</u> <u>that Matter</u>

Secrets of a Supercentenarian's Genome

Genomics News of Interest

Centre for Therapeutic Target Validation Launched

PhenomeCentral: A New Matchmaking Web Portal Launched on Rare Disease Day

Questions Remain; Some Rules Should Be Clear

Rescuing US Biomedical Research from its Systemic Flaws

Scientists Embark on Unprecedented Effort to Connect Millions of Patient Medical Records

NHGRI Workshop Spotlights the Connection Between Genomics and Theater

The Ethical, Legal, and Social Implications Program of the National Human Genome Research Institute

NIH News of Interest

NIH Fiscal Year 2015 Budget request

<u>NIH to Jump-Start Genomic Medicine with New</u> <u>Program: Genomic Data Will Play Increased Role in</u> <u>Studies at the NIH Clinical Center</u>

NIH Funding News

Comparing Success Rates, Award Rates, and Funding Rates

New NIH Policy on Resubmission Applications

Maintaining Confidentiality in NIH Peer Review

Announcement of Human Heredity and Health in Africa (H3Africa) Meetings in Cape Town, South Africa – <u>Genome Analysis</u> <u>Session</u> and <u>Cardiovascular Workshop</u>

Notice of NIH HeLa Genome Sequence Data Submission and Access Policy

NIH Funding Opportunities

Genomic Resource Grants for Community Resource Projects (U41)

Ruth L. Kirschstein National Research Service Award (NRSA) Fellowships (Pre-Doctoral, Senior, Post-Doctoral, and Dual-Doctoral)

Early Stage Development of Technologies in Biomedical Computing, Informatics, and Big Data Science (R01, R41/R42, and R43/R44)

Extended Development, Hardening and Dissemination of Technologies in Biomedical Computing, Informatics and Big Data Science (R01)

<u>Predoctoral Training in Biomedical Big Data</u> <u>Science (T32)</u>

Undiagnosed Diseases Gene Function Research (R21)

Genome Advance of the Month

Constructing the First Designer Yeast Chromosome Opens Door to Reengineering Cells

New Genomics Videos

Know Your Family History: Improve Your Health – Michelle Snyder, Sarah von Schuch, and Janine Lewis

The Dog Genome: Shedding Light on Human Disease – Elaine Ostrander

What Exactly is the Human Genome? – Eric Green and Francis Collins

<u>Genome-Scale Sequence Analysis</u> – Tyra Wolfsberg

<u>Regulatory and Epigenetic</u> <u>Landscapes of Mammalian Genomes</u> - Laura Elnitski

Applications of Genomics to Improve Public Health - Colleen McBride

Introduction to Population Genetics -Lynn Jorde

Individualized Care, Genome Analysis are the Future of Medicine – Francis Collins

> NHGRI Employment Opportunity

NHGRI Chief of the Genomic Healthcare Branch

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