March 5, 2014

It is now March, and that means one critically important thing – baseball spring training has begun and Opening Day is less than a month away! As my thoughts turn to warmer weather and watching Cardinals’ baseball, I realize that we are nearly halfway through the current fiscal year. This month, I want to give you a glimpse into how the research funds for our Extramural Research Program are distributed among the major areas of emphasis. Also, we say farewell to two NIH icons, Drs. Jane Peterson and Steve Groft – and we honor Dr. Jeff Schloss for his leadership of NHGRI’s highly successful DNA sequencing technology development program. See details below along with other informational items that I hope will be of interest to you.

March’s The Genomics Landscape features stories about:

- NHGRI’s Extramural Research Portfolio – Slicing the Funding Pie
- Jane Peterson, NHGRI Founding Member, Retires
- Featured Presentation at Advances in Genome Biology and Technology Meeting
- Steve Groft, Rare Disease Research Champion, Retires

All the best--- and Go Cardinals!

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The most important task for an Institute’s Extramural Research Program (ERP) is to develop and support a high-quality research portfolio. To this end, NHGRI has undertaken multiple strategic planning efforts, starting with the Human Genome Project and most recently culminating in the publication of “Charting a course for genomic medicine from base pairs to bedside” in 2011. While determining the broad goals for genomics is key for our research agenda, more challenging is making hard decisions about the relative priorities for the various programs that we could fund. Add to that the current challenging budget situation, and we quickly find ourselves facing many difficult choices.

The strategic plan for genomics that NHGRI published in 2011 described an overall broadening of genomics research opportunities, especially those aiming to foster the implementation of genomic medicine. In addition to the historically rich basic genomics research in genome sequencing, genomic variation, and functional genomics, the plan described compelling research into the genomic basis for disease and the implementation of genomics in medical science and healthcare.

The figure above illustrates the alignment of major accomplishments in genomics within each of five major research domains relative to past and future time intervals. In the December 2013 The Genomics Landscape, I described some recent forays into genomic medicine research (aligning under the two right-most domains), including the Implementing GeNomics Into Clinical PractiCes (IGNITE) Network, the Newborn Sequencing In Genomic medicine and public HealTh (NSIGHT) Program, and the Clinical Genome [ClinGen] Resource. These programs are but one way that the NHGRI ERP is diversifying in scope and complexity, while at the same time continuing to support substantial amounts of basic genomics research.

To track the increasing complexity of the ERP research portfolio, we are developing new approaches for coding each of our grants and programs.
(past, present, and future) relative to the components described in the 2011 strategic plan. The figure below summarizes that portfolio analysis for fiscal years 2011 through 2014 (projected). Shown in bar graphs and corresponding pie charts is the distribution of funding across the eight major components of our strategic plan: (a) five research domains: Structure of Genomes, Biology of Genomes, Biology of Disease, Science of Medicine, and Effectiveness of Healthcare; and (b) three cross-cutting areas: Computational Biology, Education and Training, and Genomics and Society.

![NHGRI Extramural Portfolio by Strategic Plan Area](image)

*The 2014 data reflect current estimates.*

From this information, one can see that the bulk of NHGRI’s extramural funds support grants within the Biology of Genomes domain (e.g., the ENCODE project and Functional Analysis Program), Biology of Disease domain (e.g., The Cancer Genome Atlas, Centers for Mendelian Genomics, and the PhenX Toolkit), and Computational Biology area (e.g., model organism databases and iSeq Tools program). A slight trend of decreasing and increasing funds supporting projects in the Structure of Genomes and Effectiveness of Healthcare domains, respectively, is evident, but there are not seismic shifts in the funding across the major domains.

The results of this new portfolio-coding effort will be regularly reviewed by staff and NHGRI advisory groups, aiming to ensure that informed decisions are made with respect to research priorities. We also endeavor to be transparent about such summaries, and plan to provide updated data from time to time in The Genomics Landscape, among other places.

To read the 2011 NHGRI strategic plan, see genome.gov/Pages/About/Planning/2011NHGRIStrategicPlan.pdf

To see more about funded programs and projects coded by strategic plan areas, see genome.gov/27534285.

 technologies – in particular, DNA sequencing technologies. To highlight the incredible achievements of NHGRI grantees in this area, Jeff submitted a poster abstract to the AGBT meeting on the history of technology development at NHGRI and our $1000 genome effort. For the first time in the history of the meeting, the organizers elected to elevate his poster abstract to a talk – not just any talk, but a featured presentation to kick off the meeting’s plenary session on technology development! The title of Jeff’s talk was “Ambitious Goals, Concerted Efforts, Conscientious Collaborations – 10 Years Hence.” This honor speaks to the true nature of Jeff’s impact and accomplishments over the past 18 years. For more information on NHGRI’s Technology Development Program, see genome.gov/10000368.

**Steve Groft, Rare Disease Research Champion, Retires**

Long-time rare disease research champion Stephen Groft, Pharm.D., Director of the Office of Rare Diseases Research (ORDR) within the National Center for Advancing Translational Sciences, retired on February 8. During his 20+ years as ORDR Director, Steve worked with legislators, regulators, researchers, pharmaceutical representatives, patients, families, and patient advocacy groups to create an environment fostering support, communication, research, and development of treatments for rare and orphan diseases. Steve and his colleagues collaborated with NHGRI to establish the Genetic and Rare Diseases Information Center and Therapeutics for Rare and Neglected Diseases Program as well as the Undiagnosed Diseases Program. His passion, commitment, and leadership leave a remarkable legacy.

For more information, see ncats.nih.gov/news-and-events/features/groft.html.
**NHGRI Funding Policy for Fiscal Year 2014**

NHGRI Advisory Council Meeting, February 10, 2014 [genome.gov/27552683](http://genome.gov/27552683)

2013-2014 Genomics in Medicine Lecture Series [youtube.com/playlist?list=PL1ay9ko4A8snwEd0X5IOYprkiXHqNtcSZ](https://youtube.com/playlist?list=PL1ay9ko4A8snwEd0X5IOYprkiXHqNtcSZ)

**Genome Advance of the Month**

Multi-Tasking DNA: Dual-Use Codons in the Human Genome [genome.gov/27556096](http://genome.gov/27556096)

The Path to Reading a Newborn’s DNA [nytimes.com/2014/02/09/business/the-path-to-reading-a-newborns-dna-map.html?_r=0](http://nytimes.com/2014/02/09/business/the-path-to-reading-a-newborns-dna-map.html?_r=0)

NHGRI Grantee, Jay Shendure, Featured in NIH Director’s Blog [directorsblog.nih.gov/2014/02/11/creative-minds-interpreting-your-genome/#more-2617](http://directorsblog.nih.gov/2014/02/11/creative-minds-interpreting-your-genome/#more-2617)

**Genomics News of Interest**


**Fellowship Opportunities**


**New Genomics Videos**

Team Discovers Genetic Disorder Causing Strokes and Vascular Inflammation in Children [genome.gov/27556385](http://genome.gov/27556385)

Team Identifies New Genetic Syndrome: Mutations in Gene Involved in Sugar Metabolism Can Lead to Allergy and Immune Disorders [niaid.nih.gov/news/newsreleases/2014/Pages/GeneticSyndrome.aspx](http://niaid.nih.gov/news/newsreleases/2014/Pages/GeneticSyndrome.aspx)

Study Pinpoints Protective Mutations for Type 2 Diabetes [broadinstitute.org/news/5570](http://broadinstitute.org/news/5570)

**NHGRI News of Interest**

Ten Drug Companies Form Pact with NIH to Find Paths to New Medicines [online.wsj.com/news/articles/SB10001424052702303519404579353442155924498](http://online.wsj.com/news/articles/SB10001424052702303519404579353442155924498)


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