

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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NHGRI's Extramural Research Portfolio – Slicing the Funding Pie

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 "<u>Charting a course for genomic</u> <u>medicine from base pairs to bedside</u>" 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.



NHGRI's 2011 strategic plan for genomics was published in the issue of Nature shown on the left, with the central organizing figure from that paper shown on the right. Note the five major domains of genomics research activities along the top of that figure.

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 <u>December 2013 The Genomics</u> <u>Landscape</u>, I described some recent forays into genomic medicine research (aligning under the two right-most domains), including the <u>Implementing GeNomics Into Clinical PracTicE (IGNITE) Network</u>, the <u>Newborn Sequencing In Genomic medicine and public HealTh (NSIGHT)</u> <u>Program</u>, and the <u>Clinical Genome (ClinGen) Resource</u>. 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

Jane Peterson, NHGRI Founding Member, Retires



It is truly the end of an era as NHGRI says goodbye to one of its founding staff members - Jane Peterson, Ph.D. After just shy of 25 years at the Institute, Jane officially retired from government service on January 17. Starting later this spring, Jane will become the CEO of the Keystone Symposia. Jane originally came to NHGRI from the National Institute of General Medical Science. She was there at the beginning of the Human Genome Project, helping to see it through its many phases and successful completion. Her scientific acumen, administrative creativity, and 'outside-the-box' thinking benefited the Human Genome Project enormously. After the Project ended, Jane played major leadership roles in establishing a number of successful programs, including the Knockout Mouse Project, The Cancer Genome Atlas, the Human Microbiome Project, and the Human Heredity and Health in Africa Initiative. Congratulations, Jane, on your new gig, and know that you will be missed at NHGRI!

Featured Presentation at Advances in Genome Biology and Technology Meeting



Last month, NHGRI's Jeff Schloss, Ph.D. was honored at the 15th Advances in Genome Biology and Technology (AGBT) meeting. Jeff is the Director of the Division of Genome Sciences within NHGRI's Extramural Research Program. For many years, he has been the Program Director overseeing the Institute's Technology Development Program, where he has skillfully managed a diverse portfolio of grants aiming to develop nucleic acids-related (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.



*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 <u>ENCODE project</u> and <u>Functional Analysis Program</u>), Biology of Disease domain (e.g., <u>The Cancer Genome Atlas</u>, <u>Centers for Mendelian</u> <u>Genomics</u>, and <u>the PhenX Toolkit</u>), and Computational Biology area (e.g., <u>model organism databases</u> and <u>iSeq Tools program</u>). 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



NIH

National Center for Advancing Translational Sciences

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 <u>ncats.nih.gov/news-and-events/features/groft.html</u>.



New Funding Information

NHGRI Funding Policy for Fiscal Year 2014 genome.gov/Pages/Grants/Policies&Guideli nes/2014NHGRIFundingPolicy.pdf

Direct Phase II SBIR Grants to Support Biomedical Technology Development grants.nih.gov/grants/guide/pa-files/PAR-14-088.html

Empirical Research on Ethical Issues Related to Central IRBs and Consent for Research Using Clinical Records and Data (R01) grants.nih.gov/grants/guide/rfa-files/RFA-OD-14-002.html

Development of Software and Analysis Methods for Biomedical Big Data in Targeted Areas of High Need (U01) grants.nih.gov/grants/guide/rfa-files/RFA-HG-14-020.html

Fellowship Opportunities

Apply for NHGRI-ASHG's New Genetics & Education Fellowship – Deadline April 25 ashg.org/pages/education fellowship.shtml

Apply for NHGRI-ASHG's Genetics & Public Policy Fellowship – Deadline April 25 ashg.org/pages/policy_fellowship.shtml

Genome Advance of the Month

Multi-Tasking DNA: Dual-Use Codons in the Human Genome genome.gov/27556096

Genomics News of Interest

The Path to Reading a Newborn's DNA nytimes.com/2014/02/09/business/thepath-to-reading-a-newborns-dnamap.html?_r=0

NHGRI Grantee, Jay Shendure, Featured in NIH Director's Blog <u>directorsblog.nih.gov/2014/02/11/creative-</u> <u>minds-interpreting-your-genome/#more-</u> <u>2617</u>

Team Discovers Genetic Disorder Causing Strokes and Vascular Inflammation in Children <u>genome.gov/27556385</u>

Team Identifies New Genetic Syndrome: Mutations in Gene Involved in Sugar Metabolism Can Lead to Allergy and Immune Disorders <u>niaid.nih.gov/news/newsreleases/2014/Pag</u> <u>es/GeneticSyndrome.aspx</u>

Study Pinpoints Protective Mutations for Type 2 Diabetes broadinstitute.org/news/5570

New Genomics Videos

NHGRI Advisory Council Meeting, February 10, 2014 genome.gov/27552683

2013-2014 Genomics in Medicine Lecture Series youtube.com/playlist?list=PL1ay9ko4A8snwEd0 X5IOYprkiXHgNIcSZ

NIH News of Interest

Ten Drug Companies Form Pact with NIH to Find Paths to New Medicines <u>online.wsj.com/news/articles/SB100014240527</u> 02303519404579353442155924498

NIH Plans to Enhance Reproducibility nature.com/news/policy-nih-plans-to-enhancereproducibility-1.14586

Blazing Trails in Brain Science nytimes.com/2014/02/04/science/blazing-trailsin-brain-science.html?smid=twnytimesscience&seid=auto&_r=0

Hannah Valantine Named NIH's First Chief Officer for Scientific Workforce Diversity <u>nih.gov/news/health/jan2014/od-30.htm</u>

NIH Adds Substantial Set of Genetic, Health Information to Online Database nih.gov/news/health/feb2014/nia-26.htm

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