

May 2, 2017

Last month, NHGRI celebrated the biggest 'holiday' of the year for genomics – DNA Day. Using social media and inperson events, we reached students and life-long learners across the globe and facilitated discussions about genetics and genomics. For example, I enjoyed participating in our Twitter Chat (#DNADay17) that featured NASA astronaut Dr. Kate Rubins and NIH Director Dr. Francis Collins discussing applications of genomics in space exploration. To see the archived Twitter conversation, use the @DNAday handle. We were pleased to hear about other major DNA Day programs around the country, illustrating how this annual event has become an important part of our field.

This month's The Genomics Landscape features stories about:

- <u>NHGRI's Genomic Medicine Working Group Illuminating Medicine's Future</u>
- Inter-Society Coordinating Committee for Practitioner Education in Genomics
- <u>Towards Broader Use of Scientific Preprints</u>
- <u>NHGRI Short Course in Genomics: Nurse, Physician Assistant, and Faculty Track Applications Open</u>

All the best,

Sim.

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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NHGRI's Genomic Medicine Working Group – Illuminating Medicine's Future

Early this month, genomic medicine researchers, clinicians, and other experts will gather in Silver Spring, Maryland as part of the latest edition in a series of meetings centered around the challenges of bringing genomic medicine into routine clinical practice (see the meeting webcast <u>here</u> live on May 2 and 3).

The ten 'Genomic Medicine meetings' held to date are one example of the productive work of the <u>NHGRI Genomic Medicine</u> <u>Working Group</u> (GMWG), a designated working group of the <u>National Advisory Council for Human Genome Research</u>. To help establish the Institute's extramural portfolio of genomic medicine research, the GMWG was constituted shortly after the publication of the Institute's 2011 strategic <u>plan</u>. The group was charged with advising NHGRI about opportunities and challenges in genomic medicine implementation, especially in light of the new, more clinically oriented goals articulated in the 2011 strategic plan.



The theme of this month's <u>'Genomic Medicine 10' Meeting</u> is implementing pharmacogenomics – the study of how genomic differences among individuals influence their response to medications. As with the <u>past nine Genomic Medicine meetings</u>, the goals of this gathering are to review past and ongoing research efforts and to establish what future research is needed to facilitate broader efforts in genomic medicine implementation. More specifically, the Genomic Medicine 10 meeting will survey programs that are currently implementing pharmacogenomics, review the field's latest advances, and develop a strategic framework for implementing pharmacogenomics on a larger scale.

Apart from coordinating Genomic Medicine meetings and, in some cases, overseeing the publication of meeting summaries (e.g., from Genomic Medicine 9, "<u>From Bench to Bedside</u>"), the GMWG also provides more general advice about NHGRI's

Inter-Society Coordinating Committee for Practitioner Education in Genomics



A summary of the most recent meeting of the Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC) is now available. The focus of this meeting was to share needs, interests, and activities among member organizations, and to learn about opportunities for funding provider-education activities. Representatives from member organizations discussed ways to facilitate genomics education. Ideas included the use of professional societies and conferences, local networks, buddy systems, electronic consultation, and continuing medical education (CME). Ideas for creating new ISCC Working Groups were also discussed, along with the challenges of producing clinical genomic reports that are easily understood by nongenomics practitioners. For more information on the ISCC, see genome.gov/27554614/.

Towards Broader Use of Scientific Preprints

NIH encourages the use of preprints

Preprints and other interim research products "can be cited anywhere other research products are cited."

In May 2016, funding agency representatives, preprint server organizations, and researchers met to coordinate efforts for making biomedical research manuscripts (i.e., preprints) more broadly available prior to formal publication. As the pace of biomedical research has accelerated, several 'preprint servers' have been established to facilitate the dissemination of scientific information more rapidly. These have been largely modeled after a preprint server established many years ago by the physics research community (arXiv). The May 2016 meeting was sponsored by <u>ASAPbio</u>, and a genomic medicine research programs. These include a number of named initiatives, such as Clinical Sequencing Exploratory Research (CSER), Implementing Genomics in Practice (IGNITE), Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT), Electronic Medical Records and Genomics (eMERGE), and Clinical Genome Resource (ClinGen), along with the NIH <u>Common Fund's</u> Undiagnosed Diseases Network (UDN).



Finally, the GMWG has also developed a <u>curated resource</u> of research publications related to genomic medicine in areas such as clinical implementation, pharmacogenomics, oncology, genome sequencing, disease-based findings, and policy. Overall, the GMWG represents an excellent example of how NHGRI can partner with members of the genomics community to think critically and strategically in tackling difficult problems and in developing an ever-improving research agenda.



summary document from that meeting was recently released ("<u>Principles for Establishing a</u> <u>Central Service for Preprints: a statement from a</u> <u>consortium of funders</u>"). The described principles pertain to governance, community support, open access, scholarly standards, leveraging existing resources, software, ease of use, sustainability, and cost effectiveness. Of particular relevance, NIH has now issued a <u>notice</u> encouraging the use of preprints and describing reporting requirements for preprint and other interim research products.

NHGRI Short Course in Genomics: Nurse, Physician Assistant, and Faculty Track Applications Open

National Human Genome Research Institute



Applications are now being accepted for the 2017 NHGRI Short Course in Genomics: Nurse, Physician Assistant, and Faculty Track. This track is for practicing nurses and physician assistants as well as for the educators of these practitioners. The Short Course will be held on the NIH campus starting in early August. During the first two days, participants will listen to lectures on genomics across the healthcare curriculum and the challenges of educating professionals about genomics. The last two days, participants will focus on the use of available resources and the implementation of successful academic and clinical strategies. Applications are due May 10. To apply, please see:

genome.gov/shortcourse/healthprofessionals/.

Spotlight on the All of Us Research Program



allofus.nih.gov

Precision Medicine Initiative aims to probe the 'dark matter' in genetic data: STAT posts a <u>question-and-</u> <u>answer interview</u> with Eric Dishman, director of the National Institutes of Health's *All of Us* Research Program.

NIH & NHGRI News

FDA Allows Marketing of First Direct-to-Consumer Tests that Provide Genetic Risk Information for Certain Conditions

Johns Hopkins Center for Inherited Disease Research Receives \$213 Million of New Funding

Upcoming Videocast

Eightieth Meeting: National Advisory Council for Human Genome Research – May 8, 2016

New Videos

Contemporary Clinical Medicine: Great Teachers Evolution, Biology and Rare Mendelian Phenotype – David Valle

Genomics Research

<u>Creative Minds: A New Mechanism for</u> <u>Epigenetics?</u>

NHLBI Stem Cell Consortium Provides New Insights into Genetics of Heart Disease, Other Conditions

Study Finds Genetic Basis for Drug Response in Childhood Absence Epilepsy

Gene Silencing Shows Promise for Treating Two Fatal Neurological Disorders

<u>NIH Researchers Trace Origin of Blood-Brain</u> <u>Barrier 'Sentry Cells'</u>

Notable Accomplishments in Genomic Medicine

Missing Genes Point to Possible Drug Targets

Researchers Begin to Understand ADHD Genes with Help from Affected Families

Rare "Knockout" Gene Mutations in Humans Help Scientists Determine Gene Function

RNA Sequencing Applied as a Tool to Solve Patients' Diagnostic Mysteries

Funding Opportunities

Integrative Computational Biology for Analysis of NHLBI TOPMed Data

2018 NIH Director's Pioneer Awards

2018 NIH Director's New Innovator Awards

2018 NIH Director's Transformative Research Awards

2018 NIH Director's Early Independence Awards

Secondary Analysis and Integration of Existing Data to Elucidate the Genetic Architecture of Cancer Risk and Related Outcomes – <u>R01 & R21</u>

Funding News

Reporting Preprints and Other Interim Research Products

