

January 9, 2018

Happy New Year!

The arrival of 2018 brings great anticipation about what this year might bring in terms of amazing genomic advances. Before staring too long into your crystal ball, you might find it interesting to look back at 2017 by checking out the top scientific advances assembled by the journals <u>Science</u> and <u>Nature</u>. While <u>Science's Breakthrough of the Year for 2017</u> is the cosmic merger of two neutron stars, the runners-up include three relevant to genomics: "pinpoint gene editing," "a cancer drug's broad swipe," and "gene therapy triumph." Nature's list of <u>Science Events that Shaped the Year</u> honors other genomic breakthroughs, collectively calling them a "genetics bonanza." Truly outstanding genomic accomplishments for 2017 – now let's see what 2018 brings us!

This month's The Genomics Landscape features stories about:

- The NIH Intramural Sequencing Center
- Two Reddit "Ask Me Anything" Sessions Tackle Gene Editing and ELSI
- <u>Congressional Committee Hearings Address Genomic Issues</u>
- Jumpstart a Career in Science Policy or Education with a ASHG-NHGRI Fellowship

All the best,

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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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# **The NIH Intramural Sequencing Center**

The NIH Intramural Sequencing Center (NISC) sits on the top floor of a research building within an office-park area of Rockville, Maryland – a few miles from the main NIH campus. This unassuming location houses the Center's state-of-the-art, high-throughput DNA sequencing facility. In a rather modest start in 1997 under the leadership of its Founding Director Eric Green, NISC began DNA sequencing with six ABI 377 instruments in lab space borrowed from another NIH institute. The fundamental mission for NISC has remained essentially unchanged in >20 years: provide contemporary genomesequencing capabilities for intramural researchers at NHGRI and other NIH institutes/centers. Under the current leadership of Director Jim Mullikin, Ph.D., and Deputy Director Jim Thomas, Ph.D., NISC remains true to that mission, regularly generating large amounts of genome-sequence data and analyses for numerous NHGRI and NIH investigators who are pursuing highly diverse research projects.



Despite its modest overall size (compared to the very large genome-sequencing centers), NISC takes pride in maintaining a broad repertoire of technical capabilities. This includes the ability to analyze genomes, epigenomes, and transcriptomes. The Center also aims to stay at the cutting edge of technology advances. The most interesting pivot points in NISC's history occurred with the switch in the late 1990s from slab-gel-based to capillary-based DNA sequencing and then the transition in the mid-2000s from Sanger-based to 'next generation' DNA sequencing platforms.

While keeping pace with such rapidly changing technologies, NISC has collaborated with individual NHGRI and NIH intramural investigators on specific research projects, and also participated in larger research consortia. For example, NISC contributed to the early efforts in sequencing the mouse genome, the most commonly used mammalian <u>model</u> in biomedical research. This foray into comparative genomics prompted NISC to establish a larger comparative vertebrate sequencing project (affectionately referred to as "ZooSeq"), in which sequence data were generated and compared for the same genomic regions in multiple organisms. The latter included important contributions of comparative genomic data to the early phase of NHGRI's <u>ENCyclopedia of DNA Elements (ENCODE) Project</u>. Later, NISC Two Reddit "Ask Me Anything" Sessions Tackle Gene Editing and ELSI



Program directors from NHGRI's Extramural Research Program will soon team up with Jennifer Doudna's laboratory at the University of California-Berkeley to host a Reddit AMA on her ground-breaking work on CRISPR-Cas9. Dr. Doudna has recently received a grant award as part of NHGRI's Centers of Excellence in Genomic Science (CEGS) program. The AMA will take place on Thursday, January 18 from 1:00 to 3:00 pm ET. Then on Monday, January 29 from 11:00 am to 1:00 pm ET, program directors from NHGRI's Ethical, Legal, and Social Implications Research (ELSI) Program will answer questions from the Reddit community. They plan to discuss the work that NHGRI funds on ELSI topics that are wide-reaching and important in genomics research, including the ethical considerations of genome sequencing in newborns and regulatory issues in directto-consumer genomic testing. But, as per usual, you can ask them anything! To see the Q&A sessions, check out the Science Reddit page.

## Congressional Committee Hearings Address Genomic Issues



Recently, the U.S. Senate and House of Representatives held several hearings that included topics relevant to NHGRI. NIH Director Francis Collins testified in hearings about the 21<sup>st</sup> Century Cures Act, which were held to celebrate the one-year anniversary of the Act's passage and to gauge progress on its implementation. Various genomics-related topics were discussed, including rare disease research and the *All of Us* Research Program. Video of the <u>House of Representatives</u> and <u>Senate</u> hearings are available online; the written <u>testimony</u> of Dr. Collins is also became involved in NIH Common Fund's <u>Human Microbiome</u> <u>Project</u>, in which genome sequencing was used to study the microbial communities that live in and on our bodies.



Over the years, NISC has expanded its genome-sequencing program to include medically-oriented projects that aim to understand the role of genomic variants in human health and disease. One such study, <u>ClinSeq</u>, has been exploring the medical, molecular, and bioinformatic challenges associated with sequencing individual participants' genomes in a clinical research setting.

The list of NISC's research highlights is long and impressive – citing a few examples helps to illustrate the breadth of its scientific contributions. In 2011, NISC researchers were part of a group that was awarded a AAAS Prize for the <u>analysis</u> of the Neanderthal genome. In 2012, NISC's sequencing and analysis efforts helped scientists and clinicians sort out the <u>mystery</u> of a bacterial outbreak in the NIH Clinical Center. In 2013, NISC contributed to the first genomic <u>survey</u> of the fungal diversity in human skin. In 2017, NISC contributed to a <u>study</u> that discovered new regions of the human genome linked to skin color variation.

NIH Intramural Sequencing Center

NISC's <u>list of publications</u> illustrates the diversity of projects in which it has been involved. Often not emphasized in such publications, though, is NISC's continued commitment to implement new technologies and methods, to improve the quality and efficiency of its DNA-sequencing pipelines, and to continually advance its data-analysis capabilities. In 2017 alone, NISC generated over 90 trillion bases of DNA sequence data. This continual forward-evolution has served NISC well since its founding, and points to a style that will keep the Center valuable for NHGRI and NIH going forward. available. In addition, the Senate held a <u>hearing</u> entitled "Gene Editing Technology: Innovation and Impact," which included a lengthy discussion about research progress and ethical and regulatory issues associated with the CRISPR technology.

### Jumpstart a Career in Science Policy or Education with a ASHG-NHGRI Fellowship



NHGRI and the American Society for Human Genetics (ASHG) are committed to strengthening the workforce with people who can bring genomics to classrooms and the political system. Towards that end, the two organizations sponsor 16-month Genetics & **Public Policy and Genetics & Education** Fellowships that prepare scientists for new and exciting careers in public policy and education, respectively. The Genetics & Public Policy Fellowship was created for genetics professionals to gain valuable policy experience at NHGRI, at ASHG, and in the U.S. **Congress. The Genetics & Education** Fellowship is geared for genetics professionals interested in developing their expertise in genomic literacy programs, science education policy, and education program development. Applications for both 2018 fellowships will open in late January, with more information available on ASHG's website. To read about the current fellows, see genome.gov/27570341/.



## Spotlight on the All of Us Research Program



allofus.nih.gov

### **Genomics Research**

Gene-Based Zika Vaccine is Safe and Immunogenic in Healthy Adults

Adding Letters to the DNA Alphabet

<u>To Sleep or Not:</u> <u>Researchers Explore</u> <u>Complex Genetic</u> <u>Network Behind Sleep</u> Duration

Mouse Model for Rare Genetic Disease Advances Understanding of Parkinson's

Primate Genomics Study Reveals Clues into AIDS Resistance

<u>Cellular Barcoding Helps</u> <u>Scientists Understand the</u> <u>Behavior of Stem Cells</u>

<u>Notable</u> <u>Accomplishments in</u> <u>Genomic Medicine</u>

- To help identify new features for the All of Us platform that would support research on health topics and advance precision medicine, the All of Us Research Program <u>invites</u> submission of important research questions that the program could consider addressing.
- The Genomics Working Group of the All of Us Research Program Advisory Panel recently <u>released a report</u> entitled "Considerations Toward a Comprehensive Genomics Strategy."

## Funding Opportunities

Mentored Research Scientist Development Awards – <u>Clinical</u> <u>Trial Required</u> & <u>Clinical Trial Not Allowed</u>

Mentored Clinical Scientist Research Career Development Awards – <u>Clinical Trial Required</u> & <u>Clinical Trial Not Allowed</u>

Ruth L. Kirschstein National Research Service Award (NRSA) Institutional Research Training Grant

NIH Pathway to Independence Awards – <u>Clinical Trial Required</u> & <u>Clinical Trial Not Allowed</u>

<u>Transformative Technology Development for the Human</u> <u>Biomolecular Atlas Program</u>

Tissue Mapping Centers for the Human Biomolecular Atlas Program

Innovation Corps (I-Corps<sup>™</sup>) at NIH Program for NIH and CDC Translational Research

#### **Funding News**

NIH Grant Closeout Resources for Grantees

<u>NIH Policy and Guidelines on the Inclusion of Individuals</u> Across the Lifespan as Participants in Research

Requirement for Electronic Submission of Research Supplements to Promote Diversity in Health-Related Research

#### NIH & NHGRI News

Law Students Explore Genomics at NIH

Amazing Things that <u>NIH-Funded Research</u> <u>Makes Possible –</u> <u>Podcast</u>

RFI on the 2019–2023 Strategic Plan for the Office of Disease Prevention, NIH

Data with Destiny: A Debrief of an NIH-NSF Workshop

<u>NIH Lifts Funding Pause</u> on Gain-of-Function <u>Research</u>

Dr. Collins Discusses Advances in Gene Therapy with the Associated Press

<u>Unlocking Life's Code:</u> <u>December 2017</u> <u>Newsletter</u>

