November 1, 2016

I do not know about the rest of you, but my most frequent thought for the past couple of months has been "are we there yet?" There seem to be so many things in play right now, including the election, the need to pass a Fiscal Year 2017 federal budget, and the inevitable transition to a new administration. The good news is that we are (hopefully) almost there on all of these fronts. Despite these uncertainties, the pace of genomic advance does not seem to be slowing down, as evidenced by the myriad impressive things that I saw firsthand reported at last month's American Society of Human Genetics meeting in Vancouver.

In this month's issue of *The Genomics Landscape*, I bring to your attention some of the many highlights in genomics and NHGRI programs, including a recent gathering to help developing nations establish capabilities in genetics and genomics and benefit from genomic medicine.

Specifically, November's *The Genomics Landscape* features stories about:

- International Summit on Human Genetics and Genomics
- NHGRI Program Reaches Milestone in Understanding How the Human Genome Works
- Undiagnosed Diseases Network Aims to Ramp Up Its Reach
- 4th ELSI Congress Seeks to Expand the ELSI Universe

All the best,

Eric

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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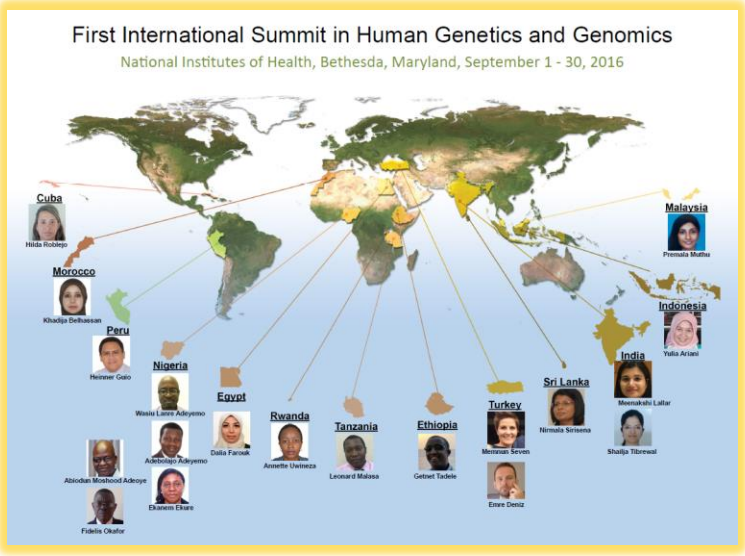
International Summit on Human Genetics and Genomics

In September, NHGRI and NIH debuted a new outreach initiative designed to provide an opportunity to developing nations to build and expand their knowledgebase, infrastructure, and technologies for conducting genetics and genomics research. The first annual International Summit in Human Genetics and Genomics was held on the NIH campus from September 1-30, bringing together international scientists to teach them how to understand the prevalence and basis of genetic diseases in their nations and to facilitate their efforts in addressing public health challenges.

Eight NIH Institutes sponsored participants and helped with the Summit in partnership with the Foundation for the NIH. Coordinated by NHGRI staff under the leadership of Dr. Max Muenke, the Summit's goals included helping attendees to ultimately improve their economies and build healthier nations worldwide by communicating advances in genetics and genomics to the global community; identifying and filling the knowledge gap in genetics and genomics faced by developing nations; and promoting genomic research and medicine through international cooperation and collaboration.



Many countries do not have researchers and healthcare professionals with formal training in genetics or educational programs to create such professionals. Training such scientists and physicians allows for the implementation of genomic medicine and aids in the prevention, diagnosis, and treatment of genetic diseases and congenital birth defects, thereby lessening the burden throughout the world.

The Summit curriculum was aimed at a graduate school level for healthcare professionals, including researchers, physicians, dentists, nurses, and counselors from developing nations. A total of 19 participants came from 13 countries across the globe, including Cuba,

NHGRI Program Reaches Milestone in Understanding How the Human Genome Works

The Reactome knowledgebase, a program funded by NHGRI's Computational Genomics and Data Science program, is making great strides in its goal of annotating every human protein encoded by the human genome. The annotation and release of information describing the 10,000th protein in early October is the culmination of over a decade's worth of work. As the human genome contains around 20,000 genes, this symbolizes the halfway point toward Reactome's goal. Reactome uses sophisticated tools to pull out patterns in large datasets to better understand how biomolecules interact with each other. It relates human proteins, genes, and other biomolecules to the biological pathways and processes in which they are involved. This is one of the many tools that NHGRI supports with the ultimate goal of understanding the genome's role in human health and disease.

Undiagnosed Diseases Network Aims to Ramp Up Its Reach

The Undiagnosed Diseases Network (UDN) is an NIH Common Fund program, with NHGRI serving as the lead Institute. Through a mix of researchers and clinicians working together using advanced DNA sequencing, metabolomics, and model organisms, the UDN aspires to solve the most challenging medical mysteries. The network has extended to seven clinical sites across the country that are able to take on patients that have a medical condition with no known cause. In September 2015, the

Egypt, Ethiopia, India, Indonesia, Malaysia, Morocco, Nigeria, Rwanda, Peru, Sri Lanka, Tanzania, and Turkey. Participants experienced over 50 lectures, field trips to the NIH Intramural Sequencing Center and various outside sites, a bioinformatics workshop, and a patient panel. The Summit encouraged collaborations among participants and NIH investigators, as well as other investigators at nearby institutions. Embassy representatives from the 13 participating countries were invited to attend the concluding talks of the Summit.

Desired outcomes of the Summit included increasing awareness of the burden that genetic disorders can have on the lives of individuals, their families, and their nations; better communicating that many of these conditions are preventable, diagnosable, treatable, or manageable; and highlighting how the collective effort by governments, educational institutions, clinicians, researchers, support groups, societies, families, and others can reduce the burden of genetic diseases and help improve the quality of life and economies of all countries.

Participants of the first International Summit in Human Genetics and Genomics.

Participants were introduced to technologies such as NextGen DNA sequencing, exome sequencing, and taught how those technologies have a role in research, prevention, diagnosis, and treatment. They also learned about data collection, analysis, interpretation, and submission. They were educated in the role of genetic/genomic counseling and support groups. Information exchange at the Summit was not a one-way street. In addition to learning from NIH staff, participants learned from each other about their experiences in using genetics and genomics technologies. Additional topics included the ethical, legal, and social Implications of generating and using genomic information, precision medicine, and newborn screening.

The current plan is to hold such an annual Summit through 2020, bringing increased knowledge about genetics and genomics to healthcare professionals around the world. For more information, visit genome.gov/27563951/.

network launched a streamlined system, called the UDN Gateway, to facilitate applying to the UDN. The Gateway is an online portal that has currently helped 380 patients gain acceptance into the UDN. It is an additional resource doctors and patients can use to help find a diagnosis, when no other avenues exist. To find out more, see undiagnosed.hms.harvard.edu/apply/.

4th ELSI Congress Seeks to Expand the ELSI Universe

On June 5-7, 2017, researchers and others interested in the Ethical, Legal, and Social Implications (ELSI) of genomics research will gather in Farmington, CT for the 4th ELSI Congress, titled "Genomics and Society – Expanding the ELSI Universe." This quadrennial conference seeks to provide an opportunity for scholars to share and reflect on current research and to envision future directions for ELSI research. The conference will include keynote speakers, plenary panels, workshops, and a wide range of paper, panel, and poster presentations. The organizers hope to engage researchers and students from inside and outside academia, from multiple disciplines and professions, and from the U.S. and other countries. Trainees are encouraged to apply for travel awards. For more information and to submit an abstract or proposal, please visit elsicon2017.org/. Abstracts and proposals are due December 1, 2016.

Spotlight on the Precision Medicine Initiative (PMI)

nih.gov/precisionmedicine

- NIH is changing the name of the PMI Cohort Program to the *All of Us* Research Program.
- PMI *All of Us* Research Program announced the addition of four Healthcare Provider Organizations (HPOs) to its network of HPOs.
- NIH awards additional funding to The Scripps Research Institute for the Participant Technologies Center.
- The PMI All of Us Research Program is launching an online form to gather your input into the development of *All of Us*.

Genomics Research

NIH Scientists Uncover Genetic Explanation for Frustrating Syndrome

Creative Minds: Building the RNA Toolbox

Reactome Announces Annotation and Release of 10,000th Human Protein

Gene Therapy Shows Promise for Treating Niemann-Pick Disease Type C1

NIH Researchers Unveil New Wound-Healing Role for Protein-Folding Gene in Mice

New Videos

IGNITE and Beyond: The Future of Genomic Medicine Implementation

Dr. Collins chats with Astronaut Kate Rubins on the International Space Station

Requests for Information

Including Preprints and Interim Research Products in NIH Applications and Reports

Research Supplement to Promote Workforce Diversity in Small Businesses

Funding Opportunities

Emerging Global Leader Award

International Bioethics Research Training Program

BD2K Support for Meetings of Data Science Related Organizations

In-Depth Phenotyping and Research Using IMPC-Generated Knockout Mouse Strains Exhibiting Embryonic or Perinatal Lethality or Survivability

Animal and Biological Material Resource Centers

Funding Notices

Change in Award Information for "Human Heredity and Health in Africa (H3Africa): Coordinating Center"

Change in Award Information for "Human Heredity and Health in Africa (H3Africa): Ethical, Legal and Societal Issues (ELSI) Research Program"

Optional Electronic Submission Method to Request to Submit an Unsolicited Application That Will Exceed $500,000 in Direct Costs

Upcoming Videocast

Interpretation of Human Genomes and Identification of Impactful Variants Using Biomedical Informatics – November 2, 2016

NIH/NHGRI News of Interest

Unlocking Life's Code: October 2016 Newsletter

NIH Grantee Wins 2016 Nobel Prize in Chemistry

NIH Viewpoint in JAMA about Recent NIH Efforts on Clinical Trials Reform

NIH Nearly Doubles Investment in BRAIN Initiative Research

23andMe, NIH Work to Reduce Health Research Disparities Among African Americans

Research!America to Honor Medical and Health Research Advocacy Leaders

NHGRI Branch Chief, Leslie Biesecker, Elected to the National Academy of Medicine

Centers for Mendelian Genomics 2015 Review Paper Lead Article in *American Journal of Human Genetics* Special Issue

NIH Operates Under a Continuing Resolution