December 5, 2017

Over the past few months, I have spoken at several events celebrating genomics and genomic medicine at academic institutions that have invested in these areas. In October, I delivered a keynote address at the 2017 Cleveland Clinic Medical Innovation Summit, which highlighted a number of areas of genomic medicine implementation. Then last month, I gave talks at the University of Connecticut Institute for Systems Genomics 5th Anniversary Symposium and the University of Maryland Baltimore’s Frontiers in Genomics Symposium that commemorated the 10th anniversary of their Institute of Genome Sciences. At each of these events, it was very gratifying to see how major investments in genomics in recent years are paying off for these outstanding institutions.

This month’s The Genomics Landscape features stories about:

- A Responsible Approach to Genomics Research: Investing in Diversity
- New Educational Toolkit to Increase Genomic Literacy Among Physicians
- Electronic Medical Records and Genomics: Scientists Contemplate the Future
- NHGRI’s Les Biesecker Elected 2019 President of ASHG

All the best,

Eva

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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A Responsible Approach to Genomics Research: Investing in Diversity

Large-scale efforts to understand the role of the human genome in health and disease are key for understanding the genomic basis of disease and for establishing how best to implement genomic medicine. However, recent studies have revealed striking gaps in our knowledge of how the genome varies from person to person (or population to population) in both healthy and disease states. Are funding agencies, the research community, and publishers of scientific research doing enough to ensure that the conclusions drawn from genomics research adequately represent the human diversity inherent in the world?

Genomics-based studies, which are increasingly commonplace, are now regularly discovering human genomic variants associated with disease. To fully appreciate such findings, it is important to remember that our complete conceptualization of the ‘human genome’ must account for the genetically and ancestrally diverse individuals that account for the world’s populations. In this way, the inclusion of diverse and underrepresented populations in genomics-based studies is needed to ensure that all benefit from genomic advances.

Understanding the contribution of genomic variation to health and disease has been a major focus of NHGRI’s work for the past three decades. The Institute has demonstrated its commitment through the programmatic support for projects such as Population Architecture using Genetics and Epidemiology (PAGE), Human Health and Heredity in Africa (H3Africa), the Centers for Common Disease Genomics, and the GWAS Catalog. The latter is a joint project with the European Bioinformatics Institute (EMBL-EBI) to curate and catalog the genomic variants associated with human disease.

New Educational Toolkit to Increase Genomic Literacy Among Physicians

The Intersociety Coordinating Committee for Practitioner Education in Genomics (ISCC) recently released a new educational resource focused on implementing training in genomic medicine. With the goal of increasing physicians’ genomic literacy, the Universal Genomics Instructor Handbook and Toolkit promotes a customizable curriculum for physicians and facilitates the ability of educators and healthcare providers to learn how to use genomics in patient care. The handbook and toolkit are freely available online. They include exercises that can be adapted to each medical specialty, and provide instructions for ensuring successful adaptation. For more details, visit genome.gov/27569865.

Electronic Medical Records and Genomics: Scientists Contemplate the Future

In October, NHGRI hosted a program review workshop entitled “eMERGE and Beyond: The Future of Electronic Medicine Records (EMR) and Genomics.” The workshop highlighted accomplishments of the Institute’s Electronic Medical Records and Genomics (eMERGE) Network. The meeting aimed to identify potential future directions for eMERGE and similar programs by outlining research gaps and challenges and by defining future opportunities in genomic medicine research. In addition to presentations from members of the current eMERGE Network, participants heard from
However, a recent analysis published last year by Popejoy and Fullerton in *Nature* using data from the GWAS Catalog found that studies involving individuals of European ancestry made up the vast majority of the participants in genomics research compared with those of non-European ancestry. While this study highlights how far the field has come since the completion of the Human Genome Project, it also identified the need to do more.

To add a voice to this discussion, the NHGRI leadership recently published a perspective in *Nature Reviews Genetics* that outlines actions to be taken by researchers, funding agencies, and journals to ensure that appropriate attention – and more importantly – action is given to this topic. The perspective lays out a variety of ways in which participant diversity can accelerate the subsequent translation of research findings. Specifically, these include performing research that investigates both the genomic and environmental contributors to health disparities, designing studies that recruit diverse participants and underserved populations, making analysis and interpretation resources readily available, and applying the knowledge gained to healthcare systems. Importantly, enhancing the diversity of research participants and investigators could be strengthened in all of these areas.

At the same time, the onus of enhancing diversity cannot rest solely on the shoulders of the research community. Rather, funding agencies, like NIH, should also take steps to support projects that not only include diverse participants and communities, but also fund underrepresented trainees and established researchers to increase the diversity among scientists and clinicians. Finally (and especially relevant to NHGRI’s mission), diversity must be prioritized in genomic medicine research so that all groups and populations benefit from the clinical applications of genomics.

To view the presentations and discussion, tune into GenomeTV.

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**NHGRI’s Les Biesecker Elected 2019 President of ASHG**

Les Biesecker, M.D., has been elected the 2019 President of the American Society of Human Genetics (ASHG), a professional membership organization for human genetics specialists worldwide. Dr. Biesecker is Chief of the Medical Genomics and Metabolic Genetics Branch in NHGRI’s Intramural Research Program, where he studies the relationship of genomic variation to health and disease. Dr. Biesecker leads studies of several rare disorders of development and overgrowth. He also founded ClinSeq® to understand the contributions of rare genomic variants to common disease. Dr. Biesecker will be serving in this role in his personal capacity. To read more, visit genome.gov/27569867/.
Spotlight on the All of Us Research Program
allofus.nih.gov

➢ The All of Us Research Program has partnered with 14 national community groups and healthcare provider associations to raise awareness about participation in the Program.
➢ NIH’s All of Us Research Program has also partnered with the National Library of Medicine to reach communities through local libraries.

Genomics Research

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