June 7, 2016

Last month, I had the honor of giving the Commencement Address at the St. Louis University School of Medicine’s graduation ceremony. I was delighted to have the opportunity to speak to a group of nascent physicians about the roles that they will likely play as young physicians in making genomic medicine a reality. The experience gave me the chance to reflect on how much genomics has changed the biomedical landscape in the 10,500-plus days since my medical school graduation – it is truly remarkable!

In this issue of The Genomics Landscape, we feature some exciting developments with NHGRI’s Clinical Sequencing Exploratory Research Program. This month’s issue also highlights the newly selected Director of the National Library of Medicine, recently funded studies on the ethical, legal and social implications of genomic information, the wrapping up of a seminar series commemorating the 25th anniversary of the launch of the Human Genome Project, and the availability of online videos for a recent set of educational lectures on genome analysis.

Specifically, June’s The Genomics Landscape features stories about:

- Clinical Sequencing: Beyond Exploration
- New Director, National Library of Medicine Selected
- New Centers of Excellence in Ethical, Legal, and Social Implications Research (CEER)
- 25th Anniversary of Human Genome Project’s Launch: Seminar Series Ends
- Current Topics in Genome Analysis Videos Online

All the best,

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Clinical Sequencing: Beyond Exploration

Often, the advances made in technologies for genome sequencing are cited as a pivotal achievement that will change medical practice in important and productive ways. In fact, I am often a person making such claims, and I stand by them. But that is not to say that I think the path forward will be easy – rather, there are many issues that need to be explored in bringing genome sequencing into medicine. NHGRI has a number of major research programs investigating those issues, a key one being the Clinical Sequencing Exploratory Research (CSER; pronounced ‘caesar’) Program.

In 2011, NHGRI launched CSER with co-funding from the National Cancer Institute. Two years later, additional research projects and a Coordinating Center were added, and nine projects formerly comprising the NHGRI-funded Return of Results Consortium and the NHGRI Intramural ClinSeq project were brought into the fold. This impressive national consortium (comprising clinicians, scientists, bioinformaticians, economists, legal scholars, and ethicists) explores the use of genome sequencing to improve patient care.

CSER is capitalizing on NHGRI’s long-standing experience in genome sequencing and analysis to explore the feasibility and best uses of genomic information for medical care. CSER’s efforts have helped to guide the development and sharing of genomic approaches that work in clinical settings. CSER investigators have also made progress in exploring the ethical, legal, and psychosocial implications of bringing genomic data into patient decision-making processes.

In its first five years, CSER has made important contributions in identifying major opportunities and challenges with clinical genome sequencing. By recruiting over 5,700 research participants and by studying the interactions among patients, physicians, and clinical laboratories, CSER is helping define what we do and do not know about the clinical utility of genomic information (e.g., which genomic information is really useful to the patient and healthcare providers). CSER researchers have helped develop ‘best practice’ guidelines about how to increase consistency of genomic variant interpretation, how to disclose research findings in pediatric settings, how to display genomic information in electronic health records, and how to have doctors share tumor genome-sequencing results that may also be heritable. Even with these significant contributions, CSER’s work is not done.

To examine the remaining challenges, NHGRI held a program planning meeting last year (“Integrating Genomic Sequencing into Clinical Care: CSER and Beyond”) and issued a Request for Information (“Maximizing Impact of a Potential Future Program in Clinical Sequencing”). Both of these sought input from the research community about future research needs in the area of clinical genome sequencing. That input helped us to formulate a plan for a next phase of CSER, which will be called “Clinical Sequencing Evidence-Generating Research (CSER2).”
CSER2 aims to identify and analyze important consensus measures in 10,000 participants, thereby generating evidence about the utility of genome sequencing for clinical care. In doing so, CSER2 will strive to generate best practices that can be readily implemented, including for patients of diverse ancestries and socioeconomic situations. The program will investigate the most important interactions among patients, family members, health practitioners, and clinical laboratories that influence the use of clinical genome sequencing, as well as identify and address real-world barriers to integrating genomics into a wide range of healthcare systems.

CSER2 aims to improve how we collect and communicate information about a patient’s health as findings from genome sequencing are generated, including understanding the impact of these findings on health-related outcomes. CSER2 will develop and test approaches to support clinical decision-making with genomic data, so that healthcare professionals have the tools for making the best clinical decisions about their patient and, potentially, their family members.

CSER2 also plans to engage a variety of stakeholders (including professional societies, insurance payers, and regulatory agencies) for examining relevant genomic issues related to clinical practice, policies, regulations, and healthcare reimbursement. As with the initial CSER Program, CSER2 will have an ethical, legal, and social implications (ELSI) research component, in this case studying issues that arise uniquely in diverse populations (and in healthcare settings outside of academic medical centers) and those that are relevant to health disparities.

NHGRI, along with the National Cancer Institute and the National Institute on Minority Health and Health Disparities, plans to fund 3-6 clinical research sites to pursue the main CSER2 goals, 3-6 clinical sites with a focus on increasing the diversity of participants, and a coordinating center. In addition, NHGRI plans to fund 5-7 investigator-initiated novel research projects that address the general CSER2 goals. These CSER2 grants are expected to be awarded next year.

To access the CSER2 Requests for Applications, see Clinical Sites, Clinical Sites with Enhanced Diversity, Coordinating Center, and Investigator-Initiated Projects. Applications for each of these components are due by August 5, 2016. For more information about CSER, see genome.gov/27546194/ and cser-consortium.org/.

Grants will fund research investigating the use of genomic information in the prevention and treatment of infectious diseases; genomic information privacy; communication about prenatal and newborn genomic testing results; and the impact of genomics in American Indian and Alaskan Native communities. For more information, see genome.gov/27565088.

25th Anniversary of Human Genome Project’s Launch: Seminar Series Ends

The NHGRI seminar series commemorating the 25th anniversary of the launch of the Human Genome Project, “A Quarter Century after the Human Genome Project’s Launch: Lessons Beyond the Base Pairs,” has come to an end. Since December 2015, the series has featured a panel discussion with former NHGRI Deputy Directors Drs. Elke Jordan and Mark Guyer, followed by lectures by Drs. Maynard Olson, Ewan Birney, Bob Cook-Deegan, Marco Marra, and David Bentley. To access video recordings from this series, see GenomeTV.

Current Topics in Genome Analysis Videos Online

Every two years, NHGRI hosts a lecture series entitled Current Topics in Genome Analysis. The series of ~14 lectures covers a wide array of cutting-edge topics in genomics, aiming for an audience at the level of first-year graduate students. Videos of all the lectures from the 2016 series are now available at youtube.com/playlist?list=PL1ay9ko4A8skYqhrA41ND27lHtebS0LY. For more information about this lecture series, see genome.gov/ctga2016/.
### Spotlight on the Precision Medicine Initiative (PMI)

- NIH Funds Biobank to Support the PMI Cohort Program: NIH will award $142 million over five years to the Mayo Clinic in Rochester, Minnesota, to establish the world’s largest research-cohort biobank for the PMI Cohort Program.
- The PMI has issued a vacancy announcement for the Chief Technology Officer for the PMI Cohort Program.
- Tune into the videocast of the PMI Cohort Program Advisory Panel Meeting on June 14.

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