National Human Genome Research Institute

Summary of NHGRI's Strategic Planning Session
Advanced Genomic Technology Development Meeting
Northeastern University, Boston, June 1, 2018

NHGRI staff provided a brief overview of NHGRI's new strategic planning initiative launched in February 2018. Staff summarized the 2020 NHGRI Strategic Plan vision and the institute and the Division of Genome Sciences' missions. They solicited audience feedback regarding bold new technologies that are needed to advance the field of genomics in 10 years and the science these technologies would enable.

In response, the audience said that NHGRI should develop technologies to assay the full human genome, telomere to telomere, including repetitive and unconserved regions, as well as all base modifications. For this to be achieved, the audience suggested that NHGRI develop new approaches to cover parts of the genome that short-read length technologies are not sufficiently determining. These data could be combined with proximity and location assays to create a complete and full dimensional physical view of the genome; allow genomics researchers to phase variants; see DNA-DNA proximity and spatial distribution of RNA and proteins; and better understand the chemical environment and phase transitions.

The audience emphasized the potential clinical applications of current and emerging technologies. They noted a lag between genomic technologies used in research and technologies that are disseminated to the clinic. The audience advocated for the development of clinically feasible and robust assays that are cost-effective and consistent with current pathology samples. The clinical assays alone, however, will not help in better informing diagnoses, unless there is an improved understanding of the genetic basis of disease, and until the gap between genetics and disease is bridged, they said. They suggested that there are opportunities for the private sector. Other ICs are developing biomarkers and NHGRI could help them develop expertise. Participants agreed that the field needs new tools and technologies for use in research settings that will enable understanding of the continuum between the biology of genomes and clinical endpoints.

The audience suggested the development of tools to improve tracking gene-environment interactions, combinatoric gene interactions and sample preparation methods for both the research and clinical communities. The field needs new tools to monitor genomics in living organisms without using disruptive methods and to enable the collection of longitudinal data.

Participants saw significant promise in "programmable biology," including learning how to manipulate and program systems that can record genomic and environmental history, inform computational inferences and, ultimately, understanding of genomics. Programmable biology efforts, they said, might lead to a better understanding of the underlying principles of natural biology, as well as to the development of new tools. The

group recognized that to develop an integrative understanding of genomics, it was important to learn big picture, large scale, insights from masses of details. Convening groups of people with a variety of expertise would also be important to solving genomics problems.

Finally, participants discussed the idea of democratizing genomic information. The audience supported giving individuals control over their genomic information and choices based on knowledge. They suggested empowering citizen scientists to use phone apps and computer software, for example, to mine genomic data, along with other data types. Their contributions would take place in an ecosystem powered by data sharing and open science (including access to resources and databases). Such an ecosystem would reinforce and accelerate advances in the field. They also shared ideas about engaging the community through contests, collaborative problem-solving, and various patient-centric or direct-to-consumer models.