

Summary of Stanford Strategic Planning Town Hall – Establishing a 2020 Vision for Genomics

Tuesday, July 17th, 2018

Overview

On Tuesday, July 17, 2018, the National Human Genome Research Institute (NHGRI) held their second in-person strategic planning town hall at Stanford University in Stanford, California. The goal of this public meeting was to solicit feedback about opportunities for genomics that should be considered as part of NHGRI's strategic planning process. Attendees were invited to give feedback on NHGRI's strategic planning effort and share what they believe are grand challenges in the field of genomics. Approximately 100 individuals from academia and industry attended this event.

The session opened with remarks from Carlos Bustamante, Ph.D., and Dean Lloyd Minor, M.D., of Stanford University, followed by NHGRI Director Eric Green, M.D., Ph.D. Dr. Green explained the context for this town hall and provided information on the plans and timeline for the NHGRI strategic planning process. NHGRI last published a strategic plan in 2011, and the new plan is projected to be published in October 2020. Between now and October 2020, NHGRI will hold town halls and other in-person gatherings across the country to collect input and explore topics to consider in developing a 2020 Vision for Genomics.

Following the introductory remarks, the town hall featured four sessions: 1) clinical, healthcare, and bedside, 2) basic/bench sciences and technology, 3) a panel with industry leaders, and 4) a trainee-only session. There were extensive question and answer discussions with Dr. Green, as well as other NHGRI representatives, including Carolyn Hutter, Ph.D., director of NHGRI's Division of Genome Sciences and Anastasia Wise, Ph.D., program director in NHGRI's Division of Genomic Medicine).

Stakeholders provided input on NHGRI's strategic planning efforts and the future of genomics, which can be categorized into seven concrete areas: data science, technology development, interactions with industry, training, phenotyping and multi-omic data, data standards and emerging areas of emphasis in genomics.

Participants reiterated the need for open and easily accessible genomic data. They expressed frustration with existing databases – that they are poorly curated and missing metadata; that the data from large databases cannot be combined for joint analysis; that there are interoperability issues, even with cloud computing; and that annotation, phenotypic, and other -omic data are frequently absent. Participants pushed for increased data generation, as they feel they do not have key multi-omic, reference, comparative and structural variant data needed for statistical analyses.

Another issue relating to data is the question of data ownership: does it belong to the individual from which it came, the researcher collecting the sample or the consortium that funded its analysis? Several people described the phenomenon of individuals trying to sell their genomic data after getting it commercially sequenced or when applying for a research study, and the ethical and social implications of that transaction. Regardless of these issues, there was unquestioned enthusiasm for NHGRI's plan to continue funding data science.

Technology development – an area highlighted in NHGRI's strategic planning presentation and materials – was also an area that participants unanimously supported. Attendees from industry and academia shared their frustrations of the grant timeline, particularly for technology development and small businesses. They argued that technology changes so rapidly that they must rely on industry more than government funding. A “fast track” for Small Business Innovation Research (SBIR) and technology development grants that would produce innovative and transformative technology was suggested. By doing so, participants argued that NHGRI could improve relationships with industry, stimulate the market and field and increase the prestige of both grant mechanisms.

Increasing support for trainees through NHGRI's training portfolio was a major theme of the meeting. Participants suggested funding more awards for individuals that straddle computational, clinical and basic genomics expertise or programs that will provide a combined computational and clinical education. Trainees expressed concern about the length of training programs, lack of post-doctoral support (compared to the number and level of grants funded by other NIH institutes) and indicated a desire for more explicit training or opportunities in community engagement and bioethics.

Town hall participants advocated for increasing funding and opportunities to collect phenotype and other multi-omic data, in combination with genomic data, for population studies. They did acknowledge the benefits of NHGRI partnering with others rather than taking the lead on those kinds of studies. It was also suggested that patients and disease advocacy groups should be engaged in the collection of phenotype data, discussions on data sharing and what kinds of work they want NHGRI to support going forward.

Many attendees told NHGRI that the institute has the convening power, reputation and position to develop standards in genomics and data science. Several contributors proposed that NHGRI design a new format for variant call files, develop standards in genome annotation and improve reference curation and presentation. Others said that NHGRI should develop reproducibility guidelines for genomics. Incongruities in how clinical genomes are sequenced, annotated and analyzed was a strong factor for why many argued that NHGRI should be involved in these issues.

There were several areas of biology that shareholders proposed NHGRI emphasize in the new strategic plan. Several spoke of an interest in embryonic and neonatal genomics and, more generally, in the genomics of reproductive health. Single-cell

genomics, non-coding genetic elements and structural variants were also classified as high priorities for future activities in genomics. Synthetic or programmable biology was also identified as a field that could be stimulated. One speaker advocated for funding comparative non-human primate and mammalian genomic studies, noting that such studies may have direct implications in human health. Trainees and younger participants suggested that NHGRI embed ethical, legal and social implications (ELSI) teams or ethical components in future programs and consortium, to help eliminate rather than exacerbate health disparities within genomic medicine.

Finally, the Stanford town hall community proposed that NHGRI write a comprehensive review on the state-of-the-art and unsolved problems in genomics. Industry representatives argued that such a document could be helpful in determining the unsolved areas where businesses could contribute to the field. Attendees reiterated that NHGRI should continue to reach out to the non-scientific community during the strategic planning process and other NIH institutes.

