

Summary of African Society of Human Genetics Strategic Planning Town Hall – Establishing a 2020 Vision for Genomics

Thursday, September 20th, 2018

Overview

On September 20, 2018, NHGRI Director Eric Green, M.D., Ph.D., and Program Director Jennifer Troyer, Ph.D., led an NHGRI 2020 strategic planning town hall at the Africa Society for Human Genetics meeting in Kigali, Rwanda. African scientists and ethicists from across the continent gave their input on how NHGRI could be at the “Forefront of Genomics” in both an African and global context. Many of the attendees have received support through the Human Heredity and Health in Africa Program ([H3Africa](#)), which is supported by the NIH Common Fund, the African Academy of Sciences, and the Wellcome Trust. The discussion covered a wide range of topics that NHGRI could address in strategic planning such as technology development, data science, ethics, community engagement, and education. There were also broader conversations on how to empower scientists in Africa.

Technology Development

Attendees of the town hall had many comments on how NHGRI could drive technology development in genetic and genomic science. There was an emphasis on the development of affordable, portable technologies that could be used at the point of care for diagnostics and disease surveillance. Face2Gene and the Oxford Nanopore’s MinION were listed as examples of technologies in this area. Attendees wanted to see NHGRI drive technology development in this area even further, to the point that power sources would not be needed to operate them. In addition, they commented that any technologies that NHGRI helps develop should be environmentally friendly.

Additionally, there was mention of developing new tools that would gather information to integrate with DNA sequence data. Attendees commented that the cost of studying gene-environment interactions is expensive and NHGRI could push technology development in this area to lower the costs. Also, there were calls to create technologies that would make the collection of phenotype data easier and more standardized. Lastly, there were remarks around the development of methods to collect longitudinal data on the timing of epigenetic changes.

Furthermore, attendees praised NHGRI’s contribution in lowering the cost of sequencing technologies, but noted that the cost could be lowered even further. This would allow larger and more diverse cohorts to be sequenced. Thus, they encouraged NHGRI’s continued investment in nucleic acid sequencing technology development.

Data Science

Another large area of discussion during the town hall was around the future of genomic data science. Attendees listed many needs in this area. For instance, they mentioned the need to make genomic data more accessible. There were suggestions to make genomic data easier and more affordable to store as well as having more resources on cloud platforms. Moreover, there were calls to improve the visualization of genomic data so that more people would be able to understand and interpret it.

Many attendees also voiced the need to have more integrative genomic data. There were recommendations to develop graph genomes that would incorporate diverse population data, better tools to analyze admixtures and complex interactions, resources to harmonize pipelines, better ways to assess the clinical actionability of variants, and improve clinical decision support tools that leverage genomic data.

Lastly, attendees stated the need to make genomic data more secure, particularly in disparaged populations where IT infrastructure might not be good enough to protect genomic data. This would help increase participation in large genomic cohort studies.

Ethics and Community Engagement

Among the attendees of the town hall were many specialists in ethics and community engagement. They noted that NHGRI could do much to impact ethics and community engagement of genomic research globally. For instance, attendees suggested that NHGRI should lead the way on how to structure and develop better genomic research consent forms for underprivileged populations. They also emphasized the need for these consents to be easily translatable into languages other than English. Furthermore, attendees suggested that NHGRI could have a large impact on how other countries develop national policies and regulations concerning genomic research. There is a need for a resource of best practices on how to do this. Finally, attendees believed that NHGRI is in a good position to create standards for public engagement between researchers and study participants.

Attendees also brought up the necessity to address ethical issues around the commercial use of genomic information. They thought that NHGRI should play a role in making firm policies to ensure that genomic information benefits patients and will not be used against them.

Education:

A recurring topic that attendees brought up was the need for better educational opportunities for health professionals in genomics and genetics. This includes primary care providers, specialists, and nurse practitioners. They also stressed the need for better tools to convey

genomic information to lay people. Lastly, attendees expressed the need for the training of more genetic counselors.

Empowering Science in Africa:

A general theme of the town hall was the need for the collection of more genomic data from African populations. There was talk of creating a large-scale sequencing program in Africa or something like the All of Us initiative for Africa. This could allow the continent to have a reference graph genome or population reference graphs. However, attendees noted that projects like these would generate large amounts of data that the continent does not have the capacity to analyze. There are certain IT limitations that African scientists face. They are unable to do large cloud computing due to insufficient bandwidth. Thus, attendees recommended putting more investment into IT development in Africa. They also mentioned that giving cloud computing credits to African institutions would be helpful.

Another theme was the desire for African scientists to have the ability to sustain themselves. There were calls for greater training for Africans on grant writing, regional centers of excellence in Africa, and an increased emphasis on international collaborations. Attendees pointed to the many disease areas to which Africans could provide unique contributions such as rare, monogenic, and infectious diseases.