

Summary of ELSI Strategic Planning Webinar #1:

Appropriate Use and Implementation of Genomics in Diverse Communities

June 28, 2019 3-5pm ET

The National Human Genome Research Institute (NHGRI) is undergoing a Strategic Planning process. In early 2018, NHGRI staff formed internal working groups tasked with identifying ambitious challenges to address in the next decade of genomics. These draft challenges, which have been informed by stakeholder feedback collected over 18 months, are meant to serve as potential building blocks for the final strategic plan which will be published in October 2020.

The Ethical, Legal, and Social Implications (ELSI) Research Program held the first of two Strategic Planning webinars in June of 2019. The goal of these two-hour webinars was to get direct input on four of the one-page challenges that had been drafted by the working group representing ELSI, education and engagement in the Strategic Plan. Dave Kaufman gave a brief introduction to the goals of the webinar, background on how the webinars fit into the Strategic Planning process, what kind of feedback NHGRI was looking for, and the two challenges that would be discussed during this first webinar. During the webinar, participants were split into two virtual breakout rooms moderated by NHGRI staff to allow more people to voice their opinions. Participants shared concerns and suggestions regarding each challenge in their breakout room. After discussing both challenges, all participants were brought back together to hear brief summaries of the discussions. This written summary is a compilation of comments from both groups on each challenge. Both oral and written comments were provided during the webinar. The version of each challenge that was discussed during the webinar may be found in the attached appendix.

Challenge 1: Ensure that genomics serves individuals, families, communities, and society, while recognizing the promise and limitations of genomics, and the social contexts it operates in (see Appendix for the one-page description of Challenge 1)

Overall Comments

Many participants noted this challenge could be written more broadly, and that it could serve as a core value for the entire strategic plan. The work and issues are not unique to ELSI. An initial concern from many participants was the clarity of this challenge. They noted discord between the title (which focuses on serving everyone who can benefit and adapting to “social contexts”) and the rest of the one-page document which mentions other contexts (healthcare setting, disease being addressed, gene-environment interaction) and implies that benefits to people may vary depending on the contexts they find themselves in. By including these different components, the meaning the challenge is trying to convey is obscured. Clearly defining the various components of interest (e.g. socioeconomic determinants of health and disease; the epidemiology of the disease and populations being addressed; the healthcare environment) and providing examples of how these components might enhance or limit the role of clinical genomics would help clarify the challenge. Specific suggestions included expanding upon what is meant by “environmental context and interactions within clinical care”, specifying key public health and population-related issues such as penetrance, and clarifying what “social context” means and how it might shape the role of clinical genomics. It was also noted that if the scope is limited to medical genomics, it should still include Mendelian disorders.

Concept of Limitations

Participants also explored the concept of limitations raised in the challenge. One participant noted that genomics has transformed biology but has not yet changed medicine and healthcare as much as initially anticipated. There was consensus that the concept of ‘limitations’ needs to be more clearly articulated. Ideas of what this concept could refer to included:

- Factors within healthcare settings that could limit the utility of genomics
- Understanding that genomics is not the answer to all medical issues and exploring those limitations
- Exploring how additional genomic research can help overcome these limits, as well as the perceived value of genomic medicine in the face of actual strengths and

limitations. The value of genomics to communities and individuals could be reassessed periodically, since genomics is so dynamic.

- Considering the appropriate level of investment in genomic medicine in light of cost and personnel trade-offs
- Exploring the value of genomics in multifactorial disease treatment and prevention

Webinar participants discussed how promises and limitations may look different for individuals, families, communities and society. For example, benefits at the individual level may not accrue to populations in public health approaches – the example of minimizing high-false positive rates was raised as a potential challenge. Alternatively, the benefits of a limited genetic test to diagnose an individual might underestimate the benefits of population-based exome or genome screening. Differences should be researched, understood and used in implementing genomics, particularly for public health uses.

Education and Engagement

Several comments were made about how the challenge might strengthen its emphases on education and engagement. It was repeatedly noted that communication in both education and engagement is a two-way street. The important role of bi-directional communication between publics/patients and researchers/care providers should be raised. Communities, healthcare providers, researchers, and IRB members should share the responsibilities of educating one another about genomic medicine and research.

NHGRI should consider existing resources and research, and engage people with diverse expertise about technological approaches to social determinants of health. Clinical genomics will improve when the contexts where it operates are better understood. Establishing clear goals in partnership with stakeholders through engagement and education is a way forward. The role of engagement is further addressed in the second discussion. As the groups transitioned to the second discussion, participants asked about the relationship between the

two challenges, noting that barriers addressed in the second challenge could influence what is needed in this challenge.

Challenge 2: Promote equitable access to genomic technologies and recognize the enduring effects of societal inequities (see Appendix for the one-page description of Challenge 2)

Webinar participants discussed various issues of access relating to this challenge. Given that there are access issues to medical technology and research generally, participants asked if NHGRI is trying to create a different or higher standard of access for genetics, is working towards the same standard for access but only in genetics, or addressing healthcare access issues related to genetics from the ground up (i.e. addressing basic issues of inequity).

One person felt the question to be asked is how ELSI and NHGRI can help promote more equitable research practices. Similar questions were raised about the need generally for more diversity among biomedical research participants and whether genomics is different. Where genetics has unique needs regarding inclusion of data, NHGRI should specify them. Some felt that language about inclusion and research participation was out of sync with the overall statement of promoting access to genetic technologies. They recommended clarifying whether the challenge is about access to tools and technologies, research participation, or both.

Several attendees felt that “access” does not capture the challenge accurately. The ELSI program should prioritize partnerships with underrepresented communities and groups to develop research that is needed and acceptable. The determination of “equity” should come from the communities working with the researchers and should not be decided for them. Another participant said that NHGRI should explicitly consider the quality of the resources and technology communities have access to.

A participant recommended that rather than call out stigmatizing research, that section of the challenge could be reframed to emphasize research on factors related to health and resilience in groups under stress.

Broad Reach of Challenge 2

It was noted that equitable access is broader than genomic technology and must be addressed in corners outside of ELSI research. One person said equitable access should be a guiding principle for technology development in general.

One participant suggested using a phrase more proactive than “recognizing” the effect of social inequities. ELSI researchers can focus on the extent to which genomic technologies are not promoting equitable access. While it’s important to recognize enduring issues, coming up with solutions and supporting research that works on deep structural issues that researchers, clinicians and communities face is a separate challenge. Efforts should go beyond recognizing social inequities and aim to impact them in health-positive ways at individual and community levels. A participant cautioned that just addressing past injustices tends to minimize present challenges. NHGRI might want to present a broader range of forward-looking issues in this challenge and encourage actions to address them.

Participants were interested in the idea of developing resources to improve equity across biomedicine. They thought NHGRI could expand on what this toolbox would look like and where support for research on these tools would come from. Some suggested tools addressing privacy issues or mistrust of researchers would work towards equity. Others said an important component is ensuring that materials meet the patients and public where they are instead of assuming ‘scientific literacy.’

Issues and Assumptions Related to Challenge 2

One participant suggested the title be reframed to focus on developing genomic technology to appropriately serve the community and avoid the assumption that the same technologies could be applied to all populations. Similarly, another said there is a need to innovate and adapt genomic technologies to different communities here and internationally, rather than simply promote what already exists.

The title of the challenge appears to assume that communities want genomic technology. It should not be presumed that all communities have the same drive for genomics.

If one focus of this challenge is research participation and development of knowledge, an assumption to be wary of is that greater participation in research will automatically create benefits for individuals or communities who are newly participating. Better research methods alone are insufficient to fix the problems of access.

There was a concern that a goal to create equitable access to genomic technologies might conflict with the goal of the first challenge to research and acknowledge the limitations of genomics in some contexts. Participants suggested using more realistic phrasing in this challenge such as seeing that “benefits of genomic technologies are accessible and address concerns of the community” to help with the contradiction.

Engagement and education

Webinar participants discussed issues of engagement. There needs to be consideration of who is being engaged and/or educated, which is not clear in the current version. Again, it should not be assumed that ELSI researchers are the ones educating communities. NHGRI staff acknowledged that NHGRI is not “the source” of foundational knowledge and that knowledge within communities should be recognized.

Participants suggested putting more emphasis on reciprocal engagement and education, again emphasizing bi-directional flow of information. NHGRI can help train genomic medicine or genomic science researchers in cultural competence and community engagement.

Engagement and education should not always be paired together. Moreover, engagement and education are not interchangeable and just because one is addressed does not mean the other will be. Engagement needs to be integrated early and be part of the whole process, not included at the tail end or as ‘box-checking.’ Legitimate engagement needs to be prioritized to be done well.

Another key element in engaging communities is understanding that there may be more pressing issues than genomics. It will be useful to ask communities to help put genomics in the broader context of healthcare and explore when genomic medicine is a good use of resources. One participant suggested researchers include groups based on healthcare needs (such as mental health) on the list of those to engage. Finally, when the private sector is included as a stakeholder, it is important to consider whether and how to weigh and incorporate their goals.

Participants noted that, while strong on ethics and social issues, the challenges did not address legal issues including those related to discrimination, sovereign Native American nations, and the changing set of data privacy laws.

Appendix

Draft Challenges Discussed at ELSI Strategic Planning Webinar #1:

Appropriate Use & Implementation of Genomics in Diverse Communities

June 28, 2019 3 – 5 pm ET

The draft challenges that follow were discussed on the June 28th ELSI Strategic Planning Webinar. These challenges were in a nascent stage at that time and have since been revised based on feedback from the ELSI Webinars and additional feedback from researchers in the ELSI and genomics communities. The version of the challenges provided below is no longer current, but is provided as context for the webinar summary.

Challenge 1: Ensure that genomics serves individuals, families, communities, and society, while recognizing the promise and limitations of genomics, and the social contexts it operates in

I. Context and Significance

Interest is growing in the contributions of gene-environment interactions to health and disease. Multi-omic datasets fused with deep phenotype records and environmental exposure data are in demand. To influence common multifactorial diseases and take on public health challenges, genomics must be deployed in an expanding range of social and disease contexts. Situating clinical genomics in a given social context (e.g. health care system, community, family) means accounting for the salient epidemiological, social and cultural factors contributing to health and disease. The balance between the benefits of genomic medicine and the necessary diversion of health resources away from other priorities should be considered in pragmatic program design. We must also understand how genomics contributes to the multifactorial etiology, prevention and treatment of a given disease. Planning and assessment of new genomic medicine applications must consider the environment surrounding both patients and disease and be transparent with patients and providers about the power and limitations of genomics.

II. Barriers

Clinical genomics is early in its transition towards prevention and public health approaches, and most research to date has occurred in a limited number of controlled contexts. The value of genomics will vary across communities; engagement work in populations and disease groups could identify community values and priorities but can be time consuming and expensive. Several challenges exist in communicating genomic findings to patients and providers. The limitations of genomic findings are not well understood; genomic risks are often presented in isolation, ignoring other risk factors and health concerns.

III. Why is this at the forefront of genomics and within NHGRI's mission?

NHGRI has been catalytic in moving genomic medicine forward. To effectively take on complex common diseases, an approach that includes social and environmental factors is critical.

IV: What is needed?

Research and engagement to identify societal and community goals for genomic applications can inform the development of programs suited to contextual factors and needs. An organized, balanced effort to develop normative, evidentiary and economic standards to bring genomics into practice is critical. Attention to the language used to talk about genomics and genomic risk can help put genomic findings into broader health and prevention contexts. Measurably improving genomic education for health professionals about the medical relevance of genomic findings is paramount. Reciprocal engagement and education of genomicists by health professionals and community

members can help us deliver clear, practical genomic interventions that are germane to patients, families and communities.

Challenge 2: Promote equitable access to genomic technologies and recognize the enduring effects of societal inequities

I. Context and Significance

The current genomic evidence base represents participants primarily of European ancestry and mainly from urban areas close to major academic research institutions, thereby limiting the applicability of those studies to other populations. While there are powerful and well-established scientific justifications for the inclusion of underrepresented and underserved populations, equally powerful is the moral obligation to seek social justice through inclusion of individuals from diverse populations.

II. Barriers

Truly equal access to genomic technologies will require not only recruitment of more diverse populations, but also recognition of the varied needs, concerns, and motivations held by these groups. Concerns related to issues such as privacy, discrimination, historical abuses, mistrust of research and access to the resulting benefits of genomic medicine, must be acknowledged and wherever possible, addressed. In addition, structural norms, provider attitudes and organizational barriers that result in translational inequities must be confronted.

III. Why is this at the forefront of genomics and within NHGRI's mission?

As genomics undergoes a new phase of growth, NHGRI must work towards inclusive and equitable science and healthcare. NHGRI's leadership in genomic data resources and ELSI research provides unique expertise and opportunities to make real progress. While other NIH Institutes and Centers are pursuing greater diversity within their scientific areas of interest, NHGRI is positioned to create foundational tools and resources for improving equity that could have a broad impact across biomedicine.

IV: What is needed?

Promoting equal access to genomic research and healthcare is a difficult challenge that will require commitment, time, and effort from the entire genomics community. Known issues such as mistrust and historical abuse must be addressed through meaningful, sustained engagement by researchers, policy makers, and health workers. Communities should be engaged prior to genomics research or implementation to understand their preferences, goals, and concerns. Researchers should cease stigmatizing underrepresented participants and patients, and instead focus on promoters of health and factors that associate with resilience and overcoming adversity. More educational resources are needed to improve scientific literacy of the public and healthcare professionals, including community health workers. Finally, ELSI research is needed to study implications within and across diverse populations to continually improve and iterate on efforts in genomic science and medicine.