American Society of Bioethics and Humanities (ASBH) NHGRI Strategic Planning Session

October 19, 2018

On Friday, October 19, 2018, a one-hour NHGRI Strategic Planning session was held at the American Society of Bioethics and Humanities annual meeting during the annual ELSI Affinity Group session. Attendance was higher than expected: 57 participants from outside of the NHGRI, including a mix of trainees and junior and senior faculty. Many current grantees attended as well as NHGRI staff members Dave Kaufman, Nicole Lockhart and Ben Berkman.

To maximize the discussion period, introductory remarks were kept brief. Participants were asked not to focus on funding, but instead to generate broad, forward-looking goals for ELSI research and to provide an example of a major milestone that ELSI researchers should achieve toward those goals by 2030. At the participants' request, four general themes were displayed that had arisen in the strategic planning process: disparities and equity; empowered decisionmaking about individual data; data use, consent, and protection; and gene-environment interaction.

Attendees were divided into four groups of roughly 15 each to encourage discussion and participation. Each group had a recorder (three NHGRI staff and one post-doc volunteer) who took notes and reported back the top three to four ideas or themes at the end of the session. Participation was robust, and the group seemed motivated to provide additional ideas and feedback throughout the strategic planning process.

The major ideas and examples of related milestones are recorded below. Feedback from each of the discussion groups has been synthesized and roughly organized by topic. Considerable input was offered on ways to improve the conduct and impact of ELSI research. Multiple groups discussed the need for a broader commitment to ELSI research across NIH to fully realize the benefits of the work. Additional topic categories included the conduct of genomic research, genomic medicine, data privacy and protection, policy, equity, gene-environment interaction, genomics and industry, genomics and identity, and citizen science.

The more detailed summary below is intended to reflect the majority of ideas expressed by attendees. Although it is unlikely that every idea captured here will be part of the final NHGRI Strategic Planning document, these ideas will be extremely helpful for ELSI Program staff to consider as the program moves forward.

Conduct of ELSI Research

Participants stressed the continued importance of foundational, conceptual and normative research to provide theoretical grounding and clarity. Conceptual and normative research is vital in all types of ELSI research. In ELSI research embedded in other genomic studies, more conceptual work could help researchers develop stronger formative bases for empirical work (which is sometimes viewed as being reactive).

Participants felt ELSI research should strengthen efforts to increase the overall impact of ELSI research on real-world issues. As the field of ELSI research evolves, broader multidisciplinary participation may be needed to identify new approaches, insights, and solutions.

Participants were also supportive of more international ELSI research, including both comparative analyses and international partnerships, such as the H3Africa program. At a high level, participants emphasized that ELSI considerations should permeate the framing of the NHGRI Strategic Plan and not be isolated within a single section. Attendees also described the need for ELSI research at other NIH Institutes and Centers and better coordination and dissemination across the NIH.

Conduct of Genomic Research

Attendees encouraged more research partnerships that would continuie to align research, industry, and community goals surrounding genomics. To this end, ELSI work should promote transparency from commerical genomic interests and set policy goals for industry. Effective outreach to commercial entities is needed to bring bioethical standards into commercial environments.

Genomic Medicine Implementation

Genomic medicine must be considered in the context of the broader healthcare system. This includes a more complete understanding and appreciation for real health resource trade-offs that may arise as funds for genomic medicine compete against funds for other medical and public health activities. Development of new models for genomic medicine implementation that consider practical, ethical, and economic factors and do not increase health disparities will be essential. An organized, balanced effort to develop evidentiary standards to bring genomics into practice would represent significant progress towards this end.

Patient and provider education will be vital to genomic medicine implementation. The genomics community should develop appropriate goals and claims to avoid over-inflating the potential of genomics. Patient education should be tailored to specific clinical and social contexts and focus on practical and reasonable genomic interventions. Efforts to improve genomic literacy should demonstrably increase knowledge in patients and providers.

Attendees also stressed the importance of considering gene-environment interactions in a way that broadens the standard conception of environment. Consideration of how to bring economic, social, cultural, and justice factors into context with the broader physical environment is warranted. Gene-environment interactions should be explored as an important factor situating genomics within health care. Such understanding can help the public to put genes into the broader environmental context and understand the relative contribution of both genes and environment. Further research into the ELSI issues surrounding epigenomics (see definition) is also needed.

Data Privacy, Protection and Use

As the uses of genomic data evolve, concepts of data privacy and protection will continue to require examination and evaluation. Human subject protections and consent frameworks could evolve to accommodate new uses of genomics data or, alternatively, the data uses could yield to evolving protection practices and frameworks. Defining practical and normative boundaries and policies to honor the terms of consent in a decentralized data environment is a considerable challenge as data can easily become disconnected from its provenance. Success in this area could include clear mechanisms for people to control or change how their data are being used, even if they have previously provided broad consent or permission.

Data hacking and re-identifiability and the fear of such events are pressing concerns in the broader research community and the public. Attendees expressed concern about the potential roll back of existing protections (e.g., the Genetic Information Non-discrimination Act, Health Insurance Portability and Accountability Act, etc.). One approach could be to depoliticize or codify key protections in local institutions to potentially limit the impact of changing political priorities.

Participants also discussed the need to understand potentially new uses of genomic data in a variety of settings. Attendees described the need to explore our relationships with personal and family data and its relevance to institutions, including social group membership and the accompanying benefits (e.g., access to insurance or access to medicine based on pharmacogenomic testing). Relatedly, there is an opportunity to learn how genomic data could be used collectively outside of academic research setting. For example, exploring how patient community groups use their own data to solve problems (e.g. embarking on self-funded research, locating additional participants, or identifying beneficial coping or treatment strategies) might provide specific examples of success.

Policy

Similar to the call for continued normative and conceptual research, attendees also stressed the need to revisit foundational legal research. More work is required to better understand and improve the use of genomic information within the legal system. Impending regulatory changes, like the revised Common Rule, require increased clarity on whether and how genomic data should be considered identifiable. Policy makers should try to understand and align researcher and public goals and increase transparency and the use of desirable practices. Such alignment should help resolve approaches to perennial issues like consent for biobanking or reconciling competing interests among clinical care, industry, and the public.

Equity

As at other strategic planning sessions, the importance of increasing diversity and equity throughout the field of genomics was emphasized by all groups. The clinical, research, and ELSI workforce must become more inclusive of individuals of different races, socioeconomic status, sexual and gender minorities, and the disabled.

In the research realm, participants described several potential areas to measure successful increases in equity. They included robust and diverse participation in public datasets and biobanks and mitigation of the trend towards the use of biology to reinforce racial stereotypes and stigmas. Participants echoed sentiments expressed at the Morehouse School of Medicine town hall, that we move away from continually stigmatizing underrepresented participants and patients, and instead focus on how we can benefit from and leverage their participation in genomic research. For example, studying the genetics of resiliency in populations that have endured enormous stress could shift how research questions are framed. The meeting participants expressed that NHGRI's focus should be on understanding research participants' priorities and addressing problems.

In the clinic, all tests should be applicable and interpretable for everyone and available in a variety of appropriate healthcare settings. This simple, critical point was emphasized several times.

Finally, it was suggested that NHGRI show leadership in addressing several sensitive topics including: eugenics and the feeling that medical genomics is striving for "perfection"; the role of commercial forces in shaping how genomics is adopted into society; the impact of genomics on reproductive rights, and decision-making; and researchers' fears about tackling or even talking about race and genomics.

Conclusion

This is a summary of the views expressed by ELSI Affinity group attendees, not NHGRI. Because it was the first public 2020 Strategic Planning session to focus solely on ELSI issues, the ASBH session was kept broad to generate both novel ideas and enthusiasm for the strategic planning effort. The 60-minute session gathered the voices of many participants in the room and produced several ideas, goals, points to consider, and examples of steps to accomplish them.

Future ELSI Strategic Planning efforts will explore ELSI research challenges in greater depth to clearly define their scope and measures of success. Attendees at the ASBH session expressed willingness to contribute to future planning work. Staff are formulating plans for webinars and examining options for a follow-up meeting, possibly at the 2019 ASBH annual meeting. Because staff noted that some trainees seemed reluctant to contribute, NHGRI will plan a separate webinar to seek input directly from ELSI trainees.