Summary of ELSI Strategic Planning Webinar #2:

Genomic Decision Making: Power and Identity

July 9, 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 to identify 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 strategic plan which will be published in October 2020.

The Ethical, Legal, and Social Implications (ELSI) Research Program held the second of two Strategic Planning webinars in July 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 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: Empower people to make well-informed decisions about genomic data access, use, sharing and protection (see Appendix for the one-page description of Challenge 1)

Generally, webinar attendees felt that this challenge would benefit from greater specificity and clarity conveying the main goal to be addressed and how that might be accomplished. Discussion about the meaning of phrases like “people”, “well-informed”, and “empowered” guided much of the conversation. Commenters noted ambiguity in whether this challenge seeks 1) to encourage research about the public’s comprehension and use of their genomic information, 2) to implement educational training on genomic and data topics, and/or 3) to incorporate public perspectives into improved frameworks for data stewardship.

Attendees felt the phrasing of the challenge overrepresented the public’s responsibility to ensure their data is used responsibly and protected. Commenters said the challenge should have more language about institutional responsibility to protect the public’s data. Attendees pointed out that even a “well-informed” public could be powerless to use or protect their data if institutions do not offer the public meaningful choices. This sentiment was underscored for vulnerable populations, who might have limited or no choice in matters regarding their genomic information. Participants suggested the challenge call upon institutions using genomic data to create data stewardship systems that support people’s decisions about their information. Some attendees were concerned about the ability to truly inform people so they can make decisions to protect their data.

When asked whether this challenge is novel, many attendees felt the challenge did not address new concerns, but still appreciated that recent changes in the overall context of genomics and its uses make this challenge timely. Participants noted that improving public understanding of genomics and biomedical data use has always been a goal of the NHGRI, but increased efforts to collect, share, interpret, and use genomic data, has brought this goal to the fore. Commenters recommended improving the language of this challenge by identifying and stating its urgency.

Some considered the goal of achieving genomic literacy among the public too broad to tackle considering how fast the field is moving and felt it would be a misdirected goal. Instead, they

suggested NHGRI encourage institutions who collect and share genomic data to use lay-friendly language and provide meaningful and supportive decision tools. Research could be used to inform and evaluate these efforts. Studies of publics, researchers and clinicians who make decisions about genomic data would help us understand how those decisions are made and inform useful tool development. Engagement work could also help create more equitable, bidirectional relationships between those providing, using and sharing data.

In addition, there was recognition that needs and desires regarding genomic data will vary among different communities and individuals, and that it would be inappropriate to assume a one-size-fits-all approach to what constitutes a "well-informed" decision-maker. This prompted some to suggest alternatives to the term "well-informed", such as "personalized", and recommend an approach that encourages critical thinking, as opposed to the more passive act of ensuring people are "informed" about data decisions.

Some commenters felt the challenge focuses too narrowly to the impact genomic medicine has on individuals, pointing out that data can have implications for families and communities. Tribal communities were referenced as a concrete example where decisions are sometimes made at a group level, as opposed to the individual level. Mentioning these kinds of issues and the need to address them could improve the importance and relevance of the challenge.

Finally, some commented that this challenge works off of an underlying assumption that the public will choose to engage in genomic technologies and fails to mention any protections for those who chose not to engage.

Challenge 2: Track and shape genomics influences on concepts of health and wellness, identity, family and community (see Appendix for the one-page description of Challenge 2)

Generally, attendees appreciated the importance and timeliness of this topic. Commenters emphasized that genetic information can be appropriated for naturalistic or deterministic

arguments that can be damaging to individuals and communities. Feedback to improve the challenge revolved around increasing language about inclusivity, explaining potential harms more thoroughly, and bringing public engagement to the fore.

Attendees encouraged the NHGRI to include more language about how genomics may influence concepts of social groups and group membership. Multiple commenters noted a lack of mention of concepts of ability/disability, which they felt was essential to the challenge. Like the first challenge discussed, attendees emphasized that the types and magnitude of impact genomics has will likely vary by community. These commenters felt that input from diverse communities (e.g. disease-patient groups, tribal communities, other underrepresented minorities) could inform the content and implementation of the challenge. Commenters also recognized that the impact of genomic information could extend beyond individuals to systems. For example, genomic information could potentially be used or misused by lawyers, social workers, health care systems, and judges to define an individual's health, wellness, identity, family, or community.

One attendee raised examples of genomics already being used in these ways, and many agreed that examining and evaluating actual uses and their downstream effects should be part of the challenge. Attendees strongly felt that to improve the challenge, NHGRI should expand the scope to assess genomics' current uses and influences on actual health, wellness, law, and identity, and should not be limited to examining related sociocultural concepts. As a corollary, participants said the NHGRI should be prepared to examine existing and potential harms of emerging uses. Language such as "track and shape" to describe the NHGRI's role were judged to be too imprecise, and attendees suggested more active, precise phrases such as "constructively inform", "illuminate", and "communicate". While some suggested the NHGRI should have a strong role in anticipating potential harms, others felt it would be more appropriate for the NHGRI to encourage discourse. Overall, attendees said that transparency would be essential and the NHGRI should include clear language about its roles in the challenge. Some attendees suggested including a point about efforts to disseminate ELSI

research findings to the general public, which could help dispel misconceptions about what kinds of conclusions can be drawn from genomic information.

Finally, a few attendees commented on the role of direct-to-consumer (DTC) companies in this challenge. Attendees felt it was important to emphasize that *all* forms of genomic data including those ordered directly by consumers could have an influence on concepts of health, wellness, identity, family, and community (not just data coming from clinical- and research-directed efforts). They also noted that the NHGRI could acknowledge its bias given past and current collaborations with DTC companies.

Appendix

Draft Challenges Discussed ELSI Strategic Planning Webinar #2

Genomic Decision-Making: Power and Identity

July 9, 2019 3 – 5 pm ET

The draft challenges that follow were discussed on the July 9th 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: Empower people to make well-informed decisions about genomic data access, use, sharing and protection

I. Context and Significance

We expect large growth in the numbers of people, families and communities that have genomic sequence information available to them. Genomic data merged with phenotypic data have already become a currency with significant value to many. While ownership of data may be distributed or transferred, the decisions to use data or transfer ownership should be shared. NHGRI can strive to ensure that members of society have the knowledge to make informed choices about the uses of their genomic data, that those choices comport with their goals and values, and that individuals can access, understand, and utilize their own data, if desired and benefit from it.

II. Barriers

Decision-making about the use and flow of genomic and health data is a multi-dimensional problem involving several use-cases and a range of associated goals and values. More than one person may need to make decisions about an individual's data, which could involve choices about access, use, sharing and protection of the data. Values affecting these decisions range from promoting science to minimizing discrimination. One person's decisions may also involve and/or impact family members and communities. Informing these complex decisions requires that we increase genomic literacy and numeracy, a fundamental problem where one solution will not fit all.

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

Developing complex multi-omic datasets that contain rich phenotypic and covariate data has been deemed essential across the NHGRI Strategic Plan. As we promote, collect, use and share these datasets to advance the forefront of genomics, NHGRI will be uniquely positioned to engage and educate individuals and communities to empower the use and control of their own data. Bringing the unique lens of ELSI research to inform these efforts and engaging various communities to understand the spectrum of perspectives will ensure that decision-making frameworks can be developed, assessed and adapted for a diverse set of people, families and communities.

IV: What is needed?

First, we must demonstrably improve genomic literacy and numeracy, paying attention to underserved and under-resourced communities. We must engage with stakeholders and communities to identify the goals and values that underlie decisions about genomic data. Based on that work and bi-directional learning, we must develop a set of tools that will inform decisions, acknowledge and validate concerns and support the choices made. In addition, resources that facilitate understanding and visualization should be available for individuals who choose to use them. ELSI research will complement education and engagement to identify and address cultural and social factors influencing decision-making and assess and iterate on the work to develop engaging, effective tools.

Challenge 2: Track and shape genomics' influences on concepts of health and wellness, identity, family and community

I. Context and Significance

Millions of people have participated in Direct-to-Consumer (DTC) genetic ancestry and health tests. These numbers will grow as ancestry companies offer more health related results. At the same time, a growing number of large association studies examining social and behavioral traits are being published. Traditional modes of communication and social media platforms are amplifying the connections being made between genomics and concepts of personality, health, disease, race, ethnicity and identity. Genomic information may be viewed as a window to greater self-knowledge and self-determination, a path to family and group membership, or a call to join others with similar variants to power new research. It may also be seen as determining the health, disease or personality of an individual or community. Genomic data may be conflated with social constructs of race and ethnicity. They may be viewed as markers of imperfection, a means of discrimination, or the basis for excluding individuals from social or political groups. Understanding these evolving interpretations of the meaning and power of genomic data and anticipating downstream effects on cultural norms and institutions can help shape education, engagement, clinical implementation and research efforts in response.

II. Barriers

Controlling interpretations of the social significance of genomic information is not within the purview of the NHGRI. Even if it were, the myriad sources that contribute to these interpretations are not within our control. It could be argued that efforts by NHGRI to amplify some interpretations and counter others might be biased by the institute's need to promote the value of research it supports. NHGRI's work on this challenge might focus on ensuring that social interpretations cleave to the science and promote transparency about the values we aim to propagate in genomics.

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

NHGRI's ELSI program has historically led work in this area and should continue to do so as genomics roles in society continue to grow and evolve. Additionally, as a leading communicator in the field, NHGRI should consider its own role in shaping concepts of health and wellness, identity, family and community.

IV: What is needed?

At a minimum, NHGRI should support research by independent scholars on the influence genomics is having on these social constructs, to assess the roles NHGRI, industry and others have in shaping them, and to anticipate downstream implications. While some interpretations may accurately reflect the genomic science, others may misunderstand or deliberately misrepresent the field. In the latter case, research can uncover the basis of these misinterpretations. If they are found to be unethical, unwarranted or in conflict with values we hope to instill in genomics, robust engagement and education about the science and its limits could be considered.