Introduction to the ELSI Strategic Planning Webinar
Genomic Decision-Making: Power and Identity
July 9, 2019 3 – 5 pm ET

The National Human Genome Research Institute (NHGRI) is undergoing a Strategic Planning process with the goal of publishing the final Strategic Plan in the fall of 2020. Over the past year, the NHGRI has held numerous related engagement events including town halls, meetings and workshops. NHGRI staff have formed internal working groups tasked with identifying and describing ambitious challenges to address in the next decade of genomics. These draft challenges, which have been informed by the feedback received to date, are meant to serve as potential building blocks for the final strategic plan which will be published in October 2020.

The internal working group that focused on ELSI, engagement and education issues in the strategic plan is hosting two distinct webinars to gather feedback on four potential challenges for the field of ELSI research over the next decade. The intent is to ask participants to think broadly about the challenges without regard to NHGRI grant mechanisms, research methods, disciplines, or funding levels. In each webinar, participants will evaluate and advise NHGRI on two related challenges. The first webinar on June 28th will focus on two topics related to the appropriate use and implementation of genomics in diverse communities. The second webinar on July 9th will focus on two topics related to decision-making about genomic data and how genomics influences our understanding of identity.

The draft challenges that follow will be discussed on the July 9th webinar. These challenges are in a nascent stage and will be revisited and revised multiple times before inclusion in the Strategic Plan. For each challenge, we are interested in feedback on whether we have: framed the problem correctly, identified forward-looking areas, made unjustified assumptions, or left key gaps unaddressed. Finally, note that these challenges are not intended to detail how NHGRI might implement the 2020 strategic plan (in terms of specific initiatives, programs, etc.).

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.

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.