# Introduction to the ELSI Strategic Planning Webinar: Appropriate Use & Implementation of Genomics in Diverse Communities June 28, 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 June 28th 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.).

**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.

**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.