Executive Summary
The National Human Genome Research Institute (NHGRI) virtually hosted their thirteenth Genomic Medicine meeting (GM XIII) titled “Developing a Clinical Genomic Informatics Research Agenda.” The meeting’s goal was to develop a research strategy on using genomic-based clinical informatics tools and resources (GBCITR) to improve the detection, treatment, and reporting of genetic disorders in clinical settings. The objectives of the meeting included:

- to define the current status of genomic-based clinical informatics and related knowledge gaps;
- to determine facilitators and barriers that affect the development and deployment of genomic-based clinical informatics resources and research needed to address them;
- to identify research needed to improve how genomic-based clinical informatics resources impact the patient and clinical decision-making processes.

Six sessions were presented and discussed with experts from government agencies, the private sector, and academia in genomic medicine and clinical informatics to address the meeting's objectives. The titles of the five sessions were:

- Making the Case for a Clinical Genomic Informatics Research Strategy
- Need for Research in Advanced Technologies (AT) to Support Genomic Medicine
- Researching the Stakeholder Perspective: Enablers and barriers that affect the integration of genomic-based clinical informatics resources in the health care system
- Defining Research Agenda that Addresses the Process for Developing Genomic-Based Clinical Informatics Resources
- Genomics in a Fragmented Healthcare Environment
- A Genomic Based Informatics Research Strategy

Before the meeting, GMXIII invitees were asked to complete a survey to assess important considerations needed to develop a genomic-based clinical informatics research strategy. The survey results were shared with invitees and presented by Dr. Marc Williams, co-chair of the GMXIII meeting. The meeting also included a presentation by the NHGRI Director, Dr. Eric Green, on the Institute’s recently published “Strategic Vision for improving human health at the Forefront of Genomics.” Dr. Green’s presentation highlighted the future research priorities and opportunities in human genomics relevant to NHGRI’s mission. An overview of the meeting is described in the “Synopsis” section of this executive summary.
Synopsis:
Session One: Making The Case for A Clinical Genomic Informatics Research Strategy
Session One highlighted the current state of GBCITR efforts in reducing barriers to implement and advance genomic medicine. The highlights from this session included:

- Identifying elements from the survey and the Technical Desiderata where significant progress has been made and which ones still require additional research support;
- A need to ensure that the development and implementation of GBCITR is done in a manner that includes equitable representation from diverse and underserved populations;
- Reporting outcomes from GBCITR efforts that capture data regarding both the benefits and harms in their clinical decision support use to improve mitigation approaches.

NHGRI should consider supporting research focused on the development and implementation of GBCITR that addresses the Technical Desiderata elements that have not yet been addressed in a manner that includes equitable representation of diverse populations.

Session Two: Need For Research In Advanced Technologies (AT) To Support Genomic Medicine
Session Two focused on identifying areas of support in developing and implementing AT to advance genomic medicine. For this session, AT included, but was not limited to, technology for improving healthcare both within and outside of the traditional patient visits that support genomic medicine. Recommendations from this session included:

- Investing in research that advances a patient-centered approach in the development and implementation of AT;
- Research in AT conducted by the genomic community should complement efforts in the private sector;
- Supporting research that generates outcomes that can be used to inform the business model of AT in a manner that promotes open-source development and attracts a broad range of stakeholders in understanding more about the incentives to implement these tools and resources.

NHGRI should consider developing a multidisciplinary consortium to create sustainable and scalable infrastructures to support genomic data collection and use. Such a consortium should address key issues such as interconnectedness, a networked ecosystem, security, and privacy concerns while promoting research, empowering patients, and promoting diversity.

Session Three: Researching The Stakeholder Perspective: enablers and barriers that affect the integration of genomic-based clinical informatics resources in the healthcare system
Session Three aimed to describe the necessary partnerships, collaborations, and processes needed to generate a sustainable GBCITR research strategy. This session also highlighted a need for research investments in genomic-based clinical workflows to support clinical decision support. Recommendations from this session included:

- Research efforts should incentivize collaborations in the development, implementation, and maintenance of a learning health system for genomics;
• A GBCITR strategy should incorporate an educational and policy research component that focuses on reducing barriers and improving knowledge for patients and providers;
• Research should focus on the development, implementation, and maintenance of genomic-based workflows that:
  o Diminish burdens for primary care providers
  o Tap into other healthcare workers and engage patients
  o Support innovation beyond typical clinical decision support reliance on alerts and reminders.

NHGRI should consider supporting the development and implementation of genomic-based workflows (including diverse methods for educating providers and patients) into EHRs to improve genomic medicine implementation. These efforts could be conducted as 1-3 year “sprints” instead of a more traditional 4-5 year project plan. Such sprints could help address the rapid pace at which workflows are being developed to support genomic medicine.

**Session Four: Defining A Research Agenda That Addresses The Process For Developing Genomic-Based Clinical Informatics Resources**

Session Four covered the role health information technologies play in advancing research in the field of genomic medicine. This session also included a discussion about the role GBCITR play in precision oncology and evidence curation. Recommendations from this session included research support to:

• Improve the interfaces between EHR’s Health Level Seven (HL7) and Fast Healthcare Interoperability Resources (FHIR) and laboratory information systems (LIMS) as LIMS currently do not support either standard;
• Lower the regulatory barriers for the development and implementation of GBCITR without compromising patient safety;
• Develop and implement a common semantic framework and data models that reduce reliance on manual curation.

NHGRI should consider supporting the development of both frameworks and processes to implement genomic medicine that includes identifying the minimum data needed for clinical and research use, elucidating ways to improve the use of augmented intelligence (the use of computational resources to support human decision-making) to improve clinical care and determining how these efforts can be implemented in a distributed healthcare ecosystem.

**Session Five: Genomics In A Fragmented Healthcare Environment**

Session Five comprised a series of presentations around GBCITR’s quality improvement strategies, genomic medicine practice innovations, as well as outcomes data collection and analysis. Also, this session included a discussion about the need for an interoperable genomic-based health exchange system. Recommendations from this session included:

• Investing in the development of specific use-cases that support genomic medicine implementation through informatics and demonstrate value and scalability of genetic interoperability;
• Investing in research that focuses on establishing a genomic-based health exchange system in a manner that synergizes with the broader health IT community’s efforts in this
space;
- Supporting efforts that facilitate the development of GBCITR and the last mile of clinical implementation into healthcare systems by identifying what has been developed and supporting implementation science research.

NHGRI should consider placing a high priority on standardization of dynamic genome annotation and interpretation—especially computational models for updating variant interpretation and clinical recommendations as knowledge evolves—and building the infrastructure to support and integrate these functions into the EHR. NHGRI should also consider playing an active role in developing a clinical exchange testing environment as part of its research strategy.

**Session Six: A Genomic Based Informatics Research Strategy**

Session Six focused on summarizing the critical knowledge gaps identified in the previous sessions and mapping a path to address them. Based on discussions from the previous sessions, which included a presentation of the survey results, several overarching themes were identified for consideration in developing a genomic-based clinical informatics research strategy. These themes included:

- Incorporating an implementation component within the overall scientific framework;
- Engaging multilevel stakeholders for a balanced value proposition and broad support;
- Identifying ways to reduce regulatory process barriers to stimulate growth in this field;
- Developing research methods to identify and mitigate inherent and pervasive biases in data, information systems, access, knowledge, clinical algorithms, and care delivery that interfere with the meaningful and beneficial use of genomics in clinical care;
- Addressing the importance of implementation equity in low-resource settings to ensure genomic-based clinical informatics does not exacerbate health disparities;
- Studying human factors, user interfaces, and workflows to enable scalable, sharable, and computable inferences of genomic knowledge and harmonization of practice guidelines;
- Ensuring clinical decision support incorporates information from multiple genes and clinical data, as well as methods for representing such information in health IT systems and making data available to the basic science community;
- Developing models that support sustainability;
- Creating educational and policy components that reduce barriers and improve knowledge for patients and providers;
- Advancing the interface between human cognition and information technology to leverage the strengths of each to improve patient care
- Developing methods to reuse genetic data as the patient moves through health systems and life.

Finally, additional information about and a webcast of the meeting can be found on the GMXIII website ([https://www.genome.gov/GM13](https://www.genome.gov/GM13)).