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Genomic Medicine XIV: Genomic Learning Healthcare Systems
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Virtual
Executive Summary

The National Human Genome Research Institute (NHGRI) virtually hosted the fourteenth Genomic Medicine meeting (GM XIV) titled "Genomic Learning Health Care Systems." The meeting's goal was to identify generalizable solutions to the challenges of implementing genomic medicine that we have encountered since the 2015 National Academy of Medicine's Genomics-Enabled Learning Health Care Systems workshop. The objectives of the meeting included:

- Explore real-world examples of how genomic learning healthcare systems (gLHS) apply cycles of genomic medicine implementation, evaluation, adjustment and updated implementation practices across delivery systems.
- Examine barriers and identify potential solutions, with a focus on lessons learned from effective gLHS and their potential transportability to other settings.
- Determine ways to develop and share solutions and form collaborations to facilitate research on implementing gLHS.

Six sessions of presentations and discussion involved experts in genomic medicine, clinical informatics, patient advocacy, health equity, and other disciplines from academia and public and private sectors.

The meeting opened with an introduction to gLHS. The introductory presentation defined a learning healthcare system (LHS) for the group, gave an overview of the 2015 National Academy of Medicine's workshop and reviewed the overall goals and objectives for the meeting. The outcomes from the previous 13 Genomic Medicine meetings, including several major programs and high-impact publications, were also presented to give the group perspective on the value of these meetings to NHGRI's future directions. Generating solutions to the identified challenges was emphasized as a key objective of the discussions.

Session 1: Laying the Groundwork

This session examined what gLHS are, how they can be implemented, and their current status, as well as sustainability of gLHS and health disparity concerns related to internet-based processes. Some solutions identified in this session included:

- Expand and enhance the data donor culture and encourage sharing of information.
- Include patients as members of gLHS teams and measure outcomes relevant to patients.
- Automate clinical management and monitoring steps and dashboards to increase the uptake and efficiency of implementation.
- Improve integration of measures of structural discrimination and social determinants of health.
- Share educational content across organizations and develop a shared evidence knowledgebase and literature archive; develop sustainable models for sharing.
- Promote transferability and interoperability of genomic and longitudinal health data, including putting data in the hands of the patient to carry among systems as they move among them.

It is important to consider supporting research focused on the development of monitoring systems such as dashboards as well as automated solutions for monitoring; equity and diversity; economics and utility; and tools for interoperability.

Session 2: IT Infrastructure

This session highlighted successful early-adopter models for genomic data integration into the EHR, as well as vast inequities in access to IT resources and ways to make systems more equitable and relevant to low-resource settings. Some solutions from this session included:

- Develop genomic health information exchanges modeled on existing clinical HIEs.
- Agree on single (or limited set of) data standard(s), disseminate, and incentivize adoption.
- Build on and extend interoperability of methods for integrating genomic data into care and exchanging with other systems, as already demonstrated at early adopter centers.
- Continue to facilitate data sharing for research across systems.
- Create national policies for genomic data re-use and reanalysis in care.
- Expand genomic training of clinical providers.

There is a need for research focused on the development and implementation of data standards, expanding genomic training opportunities for clinical trials, and testing the effects of chatbots on patient education. All these issues should be considered through a lens of creating a more equitable health system.

Session 3: The Role of gLHS in Increasing Health Equity and Access to Genomic Healthcare

This session addressed the potential for gLHS to increase rather than reduce health inequities if not implemented carefully. The concepts of race, ancestry, and social determinants of health are heavily confounded and need to be disentangled. Biases in EHRs such as missing data and small sample sizes can disproportionately affect minoritized populations. Creating patient-centered gLHS and involving patients in team-based decision-making can improve equity. Some solutions from this session included:

- Use genotypes rather than race to predict risk; include social determinants of health (SDOH) in models and measure and diminish where possible the biases they produce.
- Include equity measures, concerns, and stakeholders as central components of gLHS from the start and throughout, to ensure adequate capture of data on gLHS equity.
- Leverage HIPAA's right of access and federal information blocking rules to obtain clinical data in EHRs and HIEs when initiated and consented to by patient-partners in research.
- Increase patient and community engagement to reduce inequities in gLHS and improve effective implementation of gLHS even in non-disparities populations.
- Encourage investigators to incorporate effective engagement plans in study protocols and devote adequate resources to support engagement efforts.

There is a need for research focused on patient engagement and equity. It is important to treat engagement and equity as central parts of research, and not as afterthoughts.

Session 4: Enabling Providers to Implement Genomic Knowledge

This session highlighted the importance of access to genomic education opportunities and the development of curricula for clinical teams (particularly primary care physicians but also nurse practitioners, nurses, pharmacists, physician assistants, medical assistants, etc.) as well as the importance of institutional support and collaboration. Some highlights and solutions from this session include:

- Better integrate variant review committees with ClinGen in timely way where possible.
- Create an Interpretation Consult Service or expert panel to help educate about genetic test orders, interpretation, and determining next steps.

- Shift focus of Genetic Counselors (GC) to post-test care rather than pre-test counseling, and use alternative delivery models to support this effort, particularly for pre-test counseling. This work also needs to address low-resource settings and increasing access to GC care.
- Establish EHR templates to facilitate sharing of documentation and increase available information to providers ordering tests.
- Develop "learning community of practice" listserv to distribute information and provide updates, potentially supplemented with a panel of experts to answer questions both locally and remotely.

Research is needed on increasing access to GC and developing resources to increase availability of GCs to patients. It may also be important to support low-resource settings by making experts available, and to aid the development of curricula, training tracks, and advanced medical degree programs.

Session 5: Establishing and Sustaining gLHS

This session focused on the value of demonstrating individual and group cost savings and the large economic impact of rare diseases as powerful arguments for sustainability. Implementation of gLHS has been especially successful in integrated health systems where the health system is also the payer. Some solutions to note were:

- Encourage value-based care (demonstrating improved outcomes at reasonable costs) over fee-for-service models as the former could demonstrate economic gains and improve clinical care.
- Assess 'clinical validity' of a novel maturity model (incorporating defined outcomes) with clinicians, system leaders, and payers and apply it longitudinally to assess progress.
- Include patient satisfaction measures in clinical utility studies and engage payer advisory groups in designing such studies.

Research on the development of outcome metrics is needed as are economic studies that produce real-world evidence of how gLHS reduce costs and improve outcomes.

Session 6: Realizing the Promise of gLHS

This session highlighted the importance of clinician-laboratory interactions in variant interpretation and diagnosis. Patient engagement is also critical to gLHS implementation and research, which too often are shaped by provider preferences – involving patients in defining research questions, analysis, interpretation of results, and defining obstacles can produce substantive improvements in care processes. A scalable, national gLHS system could be created. Some solutions to note were:

- Promote dynamic, iterative interactions of clinicians and labs through improved and standardized phenotypic data sharing, unambiguous genotype representation, and standardized data storage.
- Evaluate impact of patient engagement on gLHS effectiveness or outcomes.
- Create a national LHS network, perhaps through a coordinating center to gather data from collaborating gLHS on genomic medicine practice, quality improvement, and benchmarking.
- Galvanize stakeholders to support creating such a network, including patient advocacy groups, foundations, test providers, and government agencies.
- Define, catalog, and include use of patient-informed outcomes in research, recognizing that what is valuable to patients is not always what is valuable to payers.

Research is needed on the development of guidelines for engagement and on engagement itself; workflows based around communication among providers, patients, and labs (even if asynchronous); economic value of ending the "diagnostic odyssey"; and implementation of a national gLHS.

Finally, additional information about and a webcast of the meeting can be found on the GMXIV website: https://www.genome.gov/event-calendar/genomic-medicine-xiv-genomic-learning-healthcare-systems