GMXIII Developing a Clinical Genomic Informatics Research Agenda

Goal:
To develop a research strategy on the use of genomic-based clinical informatics resources to improve the detection, treatment, and reporting of genetic disorders in clinical settings.
Overarching Themes

- Research should include an implementation science framework
- Value proposition for all stakeholders “patients, research participants, providers, payers, c-suite, a researchers, etc.”
  - support multi stakeholder collaborations
- Need for engagement with patients and partners to develop applications that are under patient control to promote genomic medicine
  - Identify ways to lower barriers related to regulatory processes to promote research in this area
- Recognition of pervasive bias in data, information systems, access, value, and knowledge
- Research needs to account for relevant workflow
Summary--Survey

• Key needs from survey
  o Research into methods for integrating analytical interpretations derived by computational models of genomic data into clinical settings are well established (#5)
  o Study ways to ensure that Clinical Decision Support has the ability to incorporate and support multiple genes and clinical info (#8)
• Research is needed that considers preferences in content, preferences, and workflow across a broad range of stakeholders
• Research into the impact of policy and regulation on implementation of Genomic Informatic strategy
• Study of models to support sustainability
• Must study the inherent biases in data, algorithms, information systems, and implementation. These are multidimensional and include:
  o Race/ethnicity
  o Social determinants of health
  o Urban/rural
  o Academic/non-academic centers

• Need to explore the value proposition which is imbalanced between researchers and participants (more noticeable in underserved populations)
Summary—Session 1 (page 2)

- Need engagement in research across a broader range of organizations (Casablanca problem)
  - Implementation equity
- Need innovative research questions and methods to address these inherent biases in a rigorous and systematic way
- Outcomes must capture both benefits and harms of gCDS to inform mitigation approaches
Summary—Session 2

- Use implementation science research methods to improve implementation equity, not just research equity
- Develop a patient-centered research agenda. Example research topics:
  - Authentication, privacy, security of genomic data accessible to patients
  - Recontact of patients absent clinician oversight
  - Knowledge requirements for different stakeholders
  - Innovative enabling platforms for obtaining and returning genetic results
  - Ability of patient-centered focus to reduce bias
• Need an updated “Business Model” of research to attract a broad range of stakeholders to participate and understand more about the incentives to implement genomic-based clinical informatics resources and tools.

• Research into ways to represent genomic information as structured data while minimize manual processes.
Summary—Session 3

- Informatics research for genomic evidence computing and genomic knowledgebase construction to enable scalable, sharable and computable inferences of genomic knowledge and harmonization of practice guidelines
- Research into novel workflows that diminish burdens for primary care providers, tap into other healthcare workers and engage patients, and don’t default to alerts and reminders
- Studies to ensure new technologies don’t exacerbate health disparities
- Educational and policy research agenda to reduce barriers and improve knowledge for patients and providers
Summary—Session 3 (page 2)

- Sociotechnical Strategies for Success
  - Informatics research for genomic evidence computing and genomic knowledgebase construction to enable scalable, sharable and computable inferences of genomic knowledge and harmonization of practice guidelines
  - Harmonize the interests of multiple stakeholders to facilitate team science and implementation science
  - Incentivize collaborations to foster research on a learning health system for genomics (what is new evidence, who is affected, what needs to be done, etc.)
  - Clinician (and patient)-centered design of workflow support.
Summary—Session 4

- Research on what constitutes a minimum data for clinical care and research “learn from less data”
- Research at the interface of human cognition and artificial intelligence “how can we take the best of both”
- Include research into explainable AI to promote clinical implementation
- Research into the development and implementation of a common semantic framework to reduce reliance on manual curation
Summary—Session 4

- Research into the development of a responsibility model across EHR for patient access to needed information

- What are 80/20 use cases in genomic informatics
Research into data interoperability between clinical systems that is focused on the implementation of genomic medicine

Develop research on specific use-cases to support genomic medicine implementation through informatics

- Prioritize based off diverse stake holders

Coordinate and synergize research findings with the broader health IT community (US Core Data for Interoperability, HL7,)

Facilitate last mile of clinical implementation—identify what’s ready and support implementation research around it
Thank You!