Advancing Health IT to Support Genomic Medicine and Research

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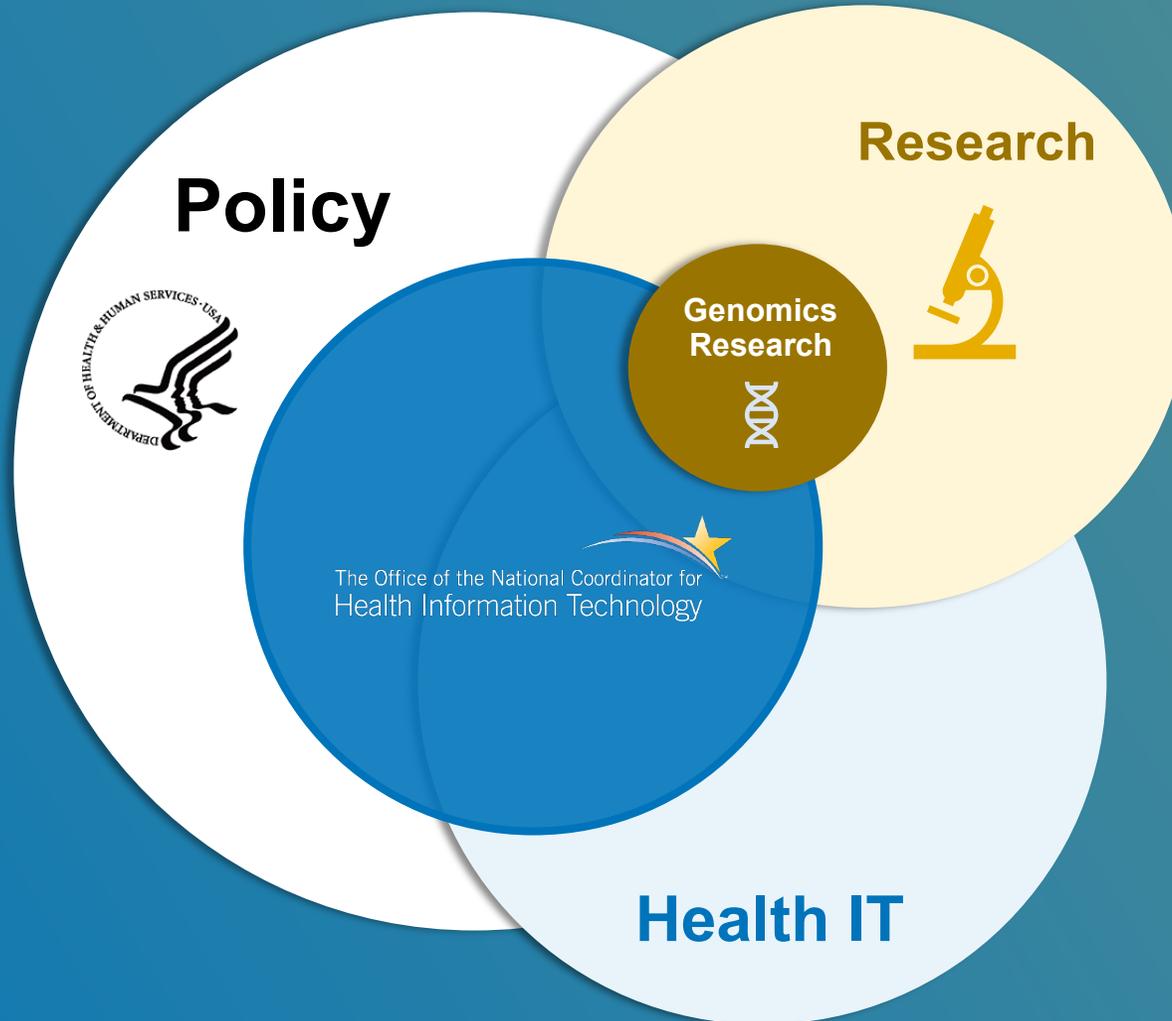**Chief Scientist**

February 10, 2021

NHGRI Genomic Medicine XIII:
Developing a Clinical Genomic Informatics Research Agenda

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We Are Here

Healthcare

Policy

Research

Genomics Research

Health IT

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At the Intersection of Research and Care Delivery

- Lead ONC's scientific efforts and activities

- Recommend scientific policy to the National Coordinator

- Promote and lead activities that spur innovation, support patient-centered outcomes research, and advance precision medicine

https://healthit.gov/topic/scientific-initiatives

Health IT is Foundational to Research

- Enormous array of data will require new thinking and pathways for storing, accessing, and analyzing health and health-related information.

- Success will require portable electronic health information that is actively exchanged among healthcare providers, researchers, and individuals.

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National Health IT Priorities for Research: A Policy and Development Agenda

Articulates a vision of a health IT infrastructure that supports alignment between clinical and research ecosystems so research can happen more quickly and effectively

https://healthit.gov/research-agenda

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Agenda Goals and Priority Areas

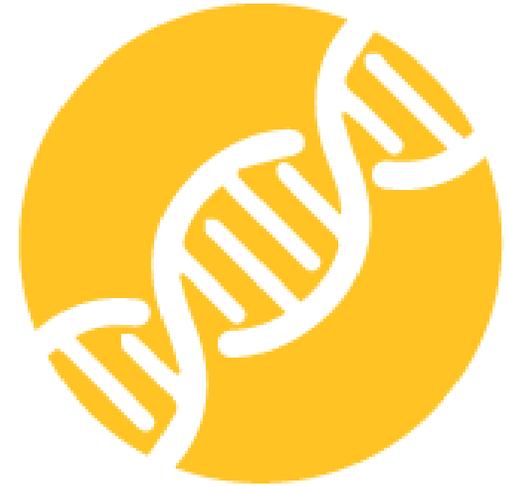**Goal 1:** Leverage High-Quality Electronic Health Data for Research

- Improve Data Quality at the Point of Capture
- Increase Data Harmonization to Enable Research Uses
- Improve Access to Interoperable Electronic Health Data

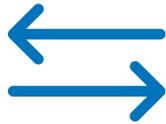**Goal 2:** Advance a Health IT Infrastructure to Support Research

- Improve Services for Efficient Data Storage and Discovery
- Integrate Emerging Health and Health-Related Data Sources
- Improve Methods and Tools to Support Data Aggregation
- Develop Tools and Functions to Support Research
- Leverage Health IT Systems to Increase Education and Participation
- Accelerate Integration of Knowledge at the Point of Care

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The Precision Medicine Initiative (PMI)

A nationwide initiative to move away from the "one-size-fits-all" approach to healthcare delivery and to instead tailor treatment and prevention strategies to people's unique characteristics, including environment, lifestyle, and biology.

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What is ONC's role in the PMI?

Accelerating innovative collaboration that supports health IT interoperability for research through pilot projects and testing of standards

Adopting policies and standards to support the privacy and security of cohort participant data

Advancing standards that support participant-driven data contribution

https://healthit.gov/topic/precision-medicine

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Sync for Genes

Standardizing the sharing of genomic information between laboratories, providers, patients, and researchers.

Phase 1—Standardizing Genomic Data:
Updated HL7® FHIR® clinical genomic specification
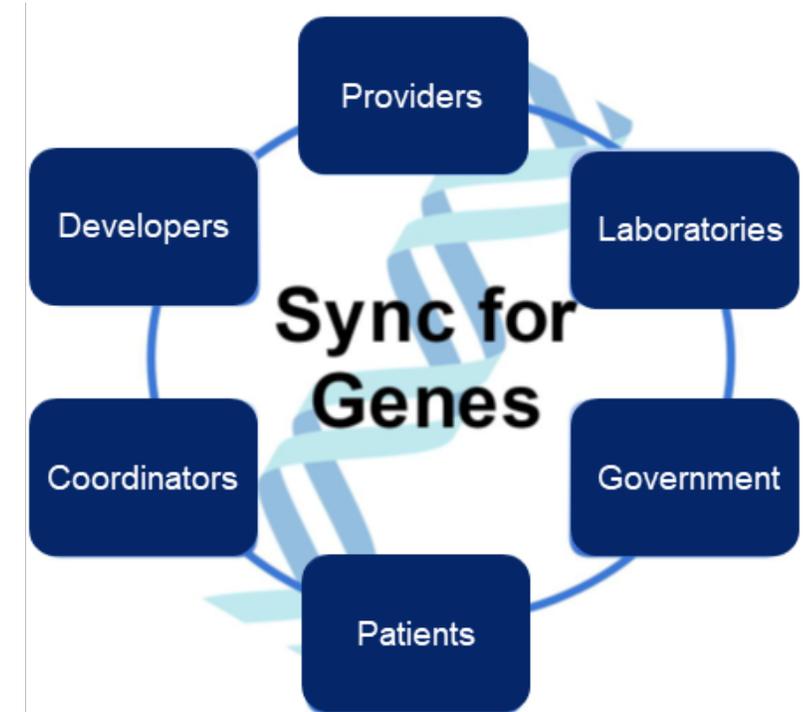
Phase 2—Integrating Genomic Data:
Demonstrated connectivity and exchange of data
Reinitiated development of the Clinical Genomics Information Model

Phase 3—Laboratory Genomic Data:
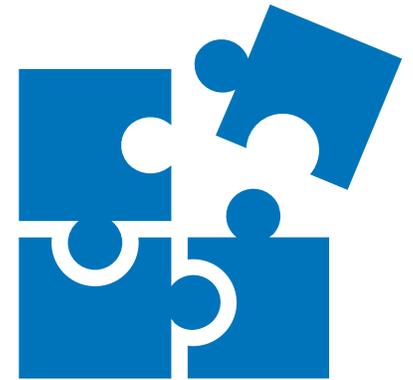Demonstrated exchange of genomic data in lab settings

Phase 4—Sharing Genomic Data with Individuals:
Enabling sharing of genomic data and supporting information with individuals

https://healthit.gov/topic/sync-genes

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Questions for Consideration

- What is NHGRI's role in advancing a clinical genomic informatics research agenda?

- Who is to benefit? Researchers? Clinicians? Patients? Participants?
  - What are their data and informatics needs?

- What are other relevant stakeholders? What role will they/should they play in advancing the agenda?
  - What can ONC advance in collaboration with NHGRI?

- How can NHGRI and its stakeholders better engage with the broader health care, health IT, health policy arena?

Additional Resources

- ONC Cures Act Final Rule: www.healthit.gov/curesrule

- U.S. Core Data for Interoperability: www.healthit.gov/USCDI
  - Draft USCDI v2: https://www.healthit.gov/isa/sites/isa/files/2021-01/Draft-USCDI-Version-2-January-2021-Final.pdf

- Interoperability Standards Advisory: www.healthit.gov/isa

- Health IT Advisory Committee: www.healthit.gov/hitac
  - USCDI Task Force: https://www.healthit.gov/hitac/committees/us-core-data-interoperability-task-force

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Thank you!

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