



Office of the National Coordinator
for Health Information Technology

Sink Your Teeth into Sync for Genes

An Overview of ONC's Sync for Genes Project

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2023 ONC Tech Forum Session

May 25, 2023



Session Agenda



**SYNC FOR
GENES**



**PHASE 5
OVERVIEW**



**PHASE 5
DEMONSTRATION
SITE**



**OUTCOMES AND
OPPORTUNITIES**



RESOURCES





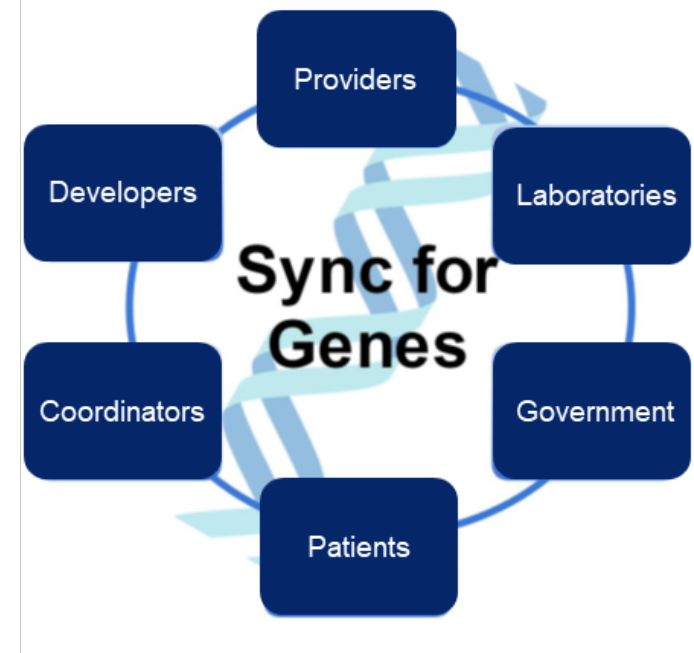
Sync for Genes: An Overview

Sync for Genes Review: Mission

Launched: 2017

Mission: Standardize sharing of genomic information between key stakeholders

- **Phase 1: Standardizing Genomic Data**
 - » Updated HL7[®] FHIR[®] clinical genomic specification
- **Phase 2: Integrating Genomic Data**
 - » Demonstrated connectivity and exchange of data
- **Phase 3: Laboratory Genomic Data**
 - » Interoperability of genomic data from laboratories
- **Phase 4: Sharing Genomic Data for Patient Care**
 - » Interoperability of genomic data between organizations and at least one data receiver, including patients or caregivers (if appropriate)
- **Phase 5: Standardizing Genomic Variant Sharing and Interpretation for Clinical Knowledge**
 - » Interoperable sharing and interpretation of annotated genomic variants at the point of care



Phase 1: Standardizing Genomic Data

Purpose: Pilot test use cases from the Health Level Seven (HL7) Clinical Genomics Work Group's Domain Analysis Model to support standardization of genomic data

Outcome:

- Successful publication of the Genomics Reporting Implementation Guide as part of HL7 Fast Healthcare Interoperability Resources (FHIR®) Release 3.0



Phase 2: Integrating Genomic Data

Purpose: Demonstrate the exchange and integration of genomic test results at the point of care

Outcomes:

- Tested FHIR[®] resources against various use cases
- Demonstrated exchange genomic diagnostic reports (GDR) using FHIR[®]
- Identified nationwide integration of genomic data into health IT challenges



Phase 3: Engaging Laboratories

Purpose: Explore use of the FHIR[®] Clinical Genomics specification for genomic data generated by laboratories

Outcomes:

- Sharing of clinical genetic reports that can be integrated and consumed into EHRs
- New, specialized human leukocyte antigen (HLA) reporting Implementation Guide using FHIR[®] shorthand



Phase 4: Sharing Genomic Data for Patient Care

Purpose: Develop application programming interfaces (APIs) focused on sharing genomic data for patient care

Outcomes:

- Developed a proof-of-concept API to deliver genomic variant results to healthcare providers
- Ability to query and to retrieve clinical genomic data via the FHIR® API from a FHIR® Server
- Developed prototype app, providing a user interface to the FHIR® API




Phase 5: Standardizing Genomic Variant Sharing and Interpretation for Clinical Knowledge

Purpose: Improve access to variant annotation data, facilitating the clinical interpretation of genomic variants

Final Report Published May 2023

<https://www.healthit.gov/topic/sync-genes#phase5>





Phase 5: Standardizing Genomic Variant Sharing and Interpretation for Clinical Knowledge

Sync for Genes Phase 5



Purpose

- Development and testing of the health IT infrastructure (i.e., genomic data standards, interoperability with genomic data knowledge bases, and APIs)
- Obtain input on interoperable sharing and interpretation of annotated genomic variants



Desired Outcomes

- Enable interoperable sharing and interpretation of annotated genomic variants at the point of care
- Develop, use, and test health IT infrastructure to share and interpret genomic variants that leverage health data standards
- Engage a panel of experts



Phase 5 Panel Activities

- Six meetings were held between January 2022 and October 2022
- The goal of each meeting was to validate and identify additional details and strategies to address challenges in the following areas:

Standards
development

Standards-
based content

Implementation
of genomic
standards

Infrastructure to
support
genomics

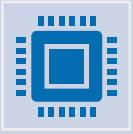
Use of genomic
data

Training and
education

Phase 5 Panel: Summary of Findings

Challenges	Findings
Standards development	Extend and harmonize existing standards to support new data types and use cases rather than develop new standards
Standards-based content	Improve interoperability by harmonizing genomic annotation across domains
Implementation of genomic standards	Develop or encourage an environment where implementers could evaluate standards before installation, document best practices and recommendations, and share lessons learned to lower barriers to adoption
Infrastructure (hardware and software) to support genomics	Find and support platforms that guide institutions responsible for developing infrastructure to support genomic data
Use of genomic data	Lower the barrier of use by the care team and patients by employing thoughtful clinical decision support (CDS)
Training and education	Encourage and support robust training, education, and support to enable the standardized representation, exchange, and use of genomic data and knowledge

Phase 5 Demonstration Overview



Children's Hospital Los Angeles (CHLA), in partnership with Elimu Informatics (Elimu), was selected to demonstrate sharing of dynamically annotated standardized genomic variants between clinicians and clinical genomic knowledge bases



The CHLA/Elimu team established a clinical decision support (CDS) pipeline that dynamically integrates a patient's genomic variants with current knowledge



The CDS pipeline used genomic knowledge represented using the GA4GH Variation Representation and Variation Annotation specifications to deliver data to two proof-of-concept apps using HL7 FHIR®



Aimed to improve clinicians' ability to make informed decisions about a patient's care by giving them access to the most up-to-date knowledge about their genomics



Phase 5 Recommendations

The outcomes of Sync for Genes Phase 5 were informed by the panel and demonstration project. The recommendations include:

1. Develop, enhance, and harmonize genomic standards
2. Develop a sandbox environment for testing
3. Support the implementation and adoption (use) of genomics
4. Enable CDS standards and capabilities



Phase 5 Demonstration Site: Deep Dive

Srikar Chamala, PhD

Children's Hospital Los Angeles

Bob Dolin, MD

Elimu Informatics



Outcomes and Opportunities

Phase 5 Outcomes



Sync for Genes Phase 5 made progress in standardizing genomic variant sharing and interpretation for clinical knowledge



Collaborated with a panel of experts and a demonstration site to build on progress from earlier phases



CHLA/Elimu team demonstrated sharing of dynamically annotated genomic information using GA4GH-encoded knowledge and HL7[®] FHIR[®]



Strategic recommendations were developed to standardize genomic variant sharing and interpretation for clinical knowledge



Work remains to coordinate existing standards with current and future requirements and expand implementation and use of standards



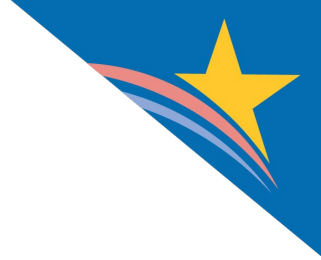
Sync for Genes: Foundational Themes

Areas where concerted efforts and collaborations are essential to drive progress in the field of genomic data sharing:

- Strategic development and adoption of genomic standards (e.g., FHIR[®], GA4GH)
- Support and coordination for implementing and integrating these standards
- Targeted education and training on the use of standards and solutions
- Addressing industry challenges impacting adoption, implementation, integration, and use



Resources



Sync for Genes Resources Toolkit: Coming Soon!

Promote	Disseminate	Engage	Encourage
awareness of the Sync for Genes accomplishments	essential resources for genomic data sharing	stakeholders in adopting, implementing, and integrating resources	increased utilization of the Sync for Genes outcomes

Designed to cater to a wide range of stakeholders who play a crucial role in the genomic data sharing ecosystem

Core Resources

Sync for Genes

- <https://www.healthit.gov/topic/sync-genes>

HL7 FHIR Standard

- <https://www.hl7.org/fhir/?ref=learnmore>

HL7 Genomics Reporting Implementation Guide

- <http://hl7.org/fhir/uv/genomics-reporting/STU2/>

HL7 FHIR Genomics Implementer Guidance

- <https://hl7.org/fhir/r4/genomics.html>

HL7 Clinical Genomics Work Group

- <https://confluence.hl7.org/display/CGW>

Global Alliance for Genomics and Health (GA4GH)

- <https://www.ga4gh.org/genomic-data-toolkit/regulatory-ethics-toolkit/framework-for-responsible-sharing-of-genomic-and-health-related-data/>





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Thank You!



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