Sync for Genes Review
Sync for Genes Review: Goal and Phases

- **Launched**: 2017
- **Goal**: Standardize 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
  
  - **Phase 3: Laboratory Genomic Data**
    - Interoperability of genomic data from laboratories
  
  - **Phase 4: Sharing Genomic Data with Individuals**
    - Interoperability of genomic data between organizations and at least one data receiver, including patients or caregivers (if appropriate)

https://www.healthit.gov/topic-sync-genes
Sync for Genes Phase 1

Purpose:
• Standardize genomic data through test use cases using HL7 FHIR

Outcomes:
• Contributed to the development of the HL7 Clinical Genomics Implementation Guidance as part of FHIR Release 3.0

Final report available from https://healthit.gov/syncforgenesis-phase1report
Sync for Genes Phase 1: Demonstration Sites

• **Counsyl and Intermountain Healthcare:** Family Health History Genetics

• **Food & Drug Administration:** Sequencing Quality and Regulatory Genomics

• **Foundation Medicine and Vanderbilt:** Somatic/Tumor Next Generation Sequencing

• **Illumina:** Next Generation Sequencing Solutions

• **National Marrow Donor Program:** Patient and Donor Matching
Sync for Genes Phase 2

Purpose:
• Continue to support testing and refinement of standards for genomic standards integration

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

Sync for Genes Phase 2: Demonstration Sites

- Lehigh Valley Health Network: Pharmacogenomics
- National Marrow Donor Program: Patient and Donor Matching
- Utah Department of Health: Newborn Screening
- Weill Cornell Medicine: Cancer Genomic Decision Support
Sync for Genes Phase 3

Purpose:
• Standardize 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

Final Report Now Available!
https://www.healthit.gov/sites/default/files/page/2021-01/Sync-for-Genes-Phase-3-Engaging-Laboratories.pdf
Sync for Genes Phase 3: Demonstration Sites

• Baylor College of Medicine Human Genome Sequencing Center: Integrating genetic variation testing information directly into EHR systems

• National Marrow Donor Program (NMDP): Exchanging genomic data requirements for human leukocyte antigen (HLA) use cases with HLA typing laboratories
Sync for Genes Phase 4

Purpose:

• Testing and updating health IT infrastructure to enable interoperable sharing of genomic data and supporting information using FHIR application programming interfaces to support patients and caregivers

Desired Outcomes:

• Feedback on the FHIR IG
• Blueprint for future work
Sync for Genes Phase 4: Demonstration Sites

• TBA!

• Currently reviewing Demonstration Site submissions
  • Will be selecting demonstration site(s) and kick-off demonstration projects in February 2021

• Questions?
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For more information you can also visit https://www.healthit.gov/topic-sync-genes