Standardizing Genomic Data Integration in Electronic Health Records

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Introduction

Integrating genomic data into Electronic Health Records (EHRs) is crucial for precise diagnoses, and targeted and personalized intervention. Establishing standards for representation and exchange ensures seamless interoperability and meaningful use of genomic information in clinical settings. Rapid advances in genomics demand robust standardization for effective analysis and interpretation. Integration of genomic data into EHRs requires a standardized approach. The intersection of Clinical Genomics and EHRs heralds a new era of interoperability and data-driven healthcare as shown in Fig (a). This poster highlights the pivotal role of HL7 FHIR, SNOMED CT, and LOINC in harmonizing clinical genomic information within EHR systems.

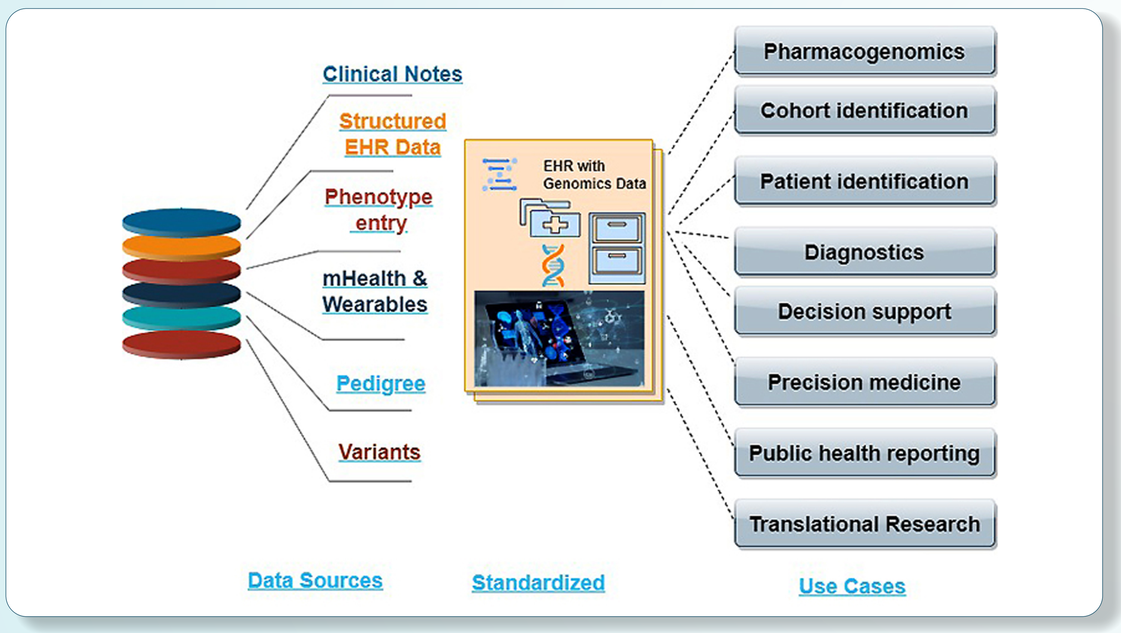

Fig (a) Overview of standardized genomic data integration in EHR

Standards for EHR and Clinical Genomics

Standards in Fig (b) comprise terminologies, protocols, methods, and specifications to capture, store, retrieve, and exchange healthcare and genomic information.

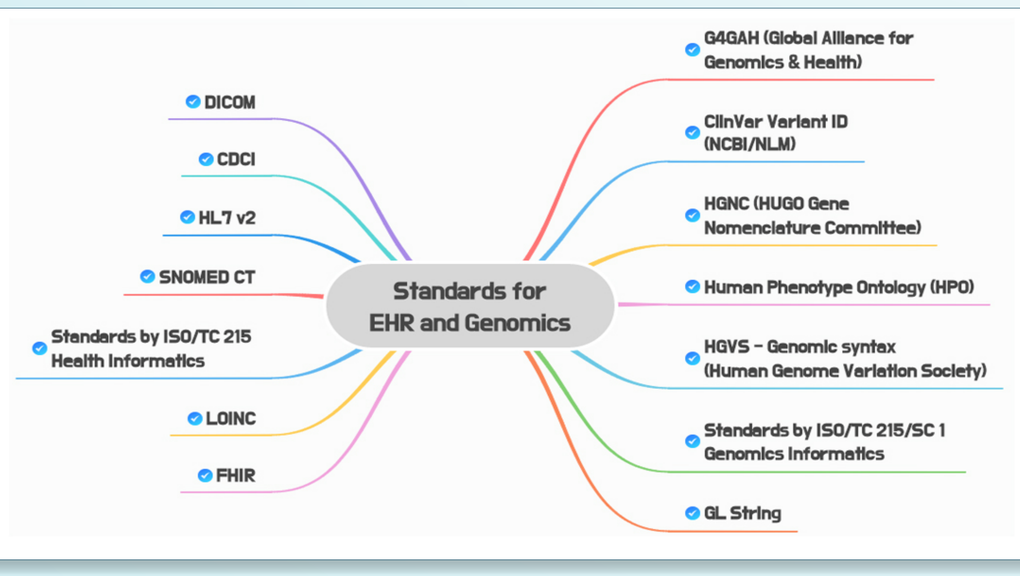

Fig (b) EHR and Genomic Standards

Notable Harmonization Efforts

Efforts in the area of facilitating the use of genomic data in EHRs briefly described in Table 1.

Name	Purpose
The electronic Medical Records and Genomics network (eMERGE)	Development of methods and best practices for integration of genetic clinical testing results and other genomics information with clinical data in the EHR
Global Alliance for Genomics and Health (GA4GH)	Phenopacket standard developed for representing individual-level phenotype and clinical data
Displaying and Integrating Genetic Information through the EHR (DIGITizE)	Effort on the standard representation and integration of genomics into EHRs
Sync for Genes	Technology-based effort to gather health data from individuals in the United States and make that information available to researchers in a standardized format
Clinical Genome Resource (ClinGen)	EHR Working Group aims to ensure that the ClinGen resource is designed to be accessible to providers and patients through EHRs and related systems

Table 1 Harmonization efforts to integrate genomic data in EHR

Clinical Genomic Data Representation using Standards

The use case for converting patient and genomics data to FHIR-compatible resources is shown in Fig ©.

1. Identify the FHIR-supported data elements
2. Identify the data structure and schema for the data elements
3. Identify Standards code mapping
4. Validate the FHIR resource
5. Store and retrieve data using FHIR API

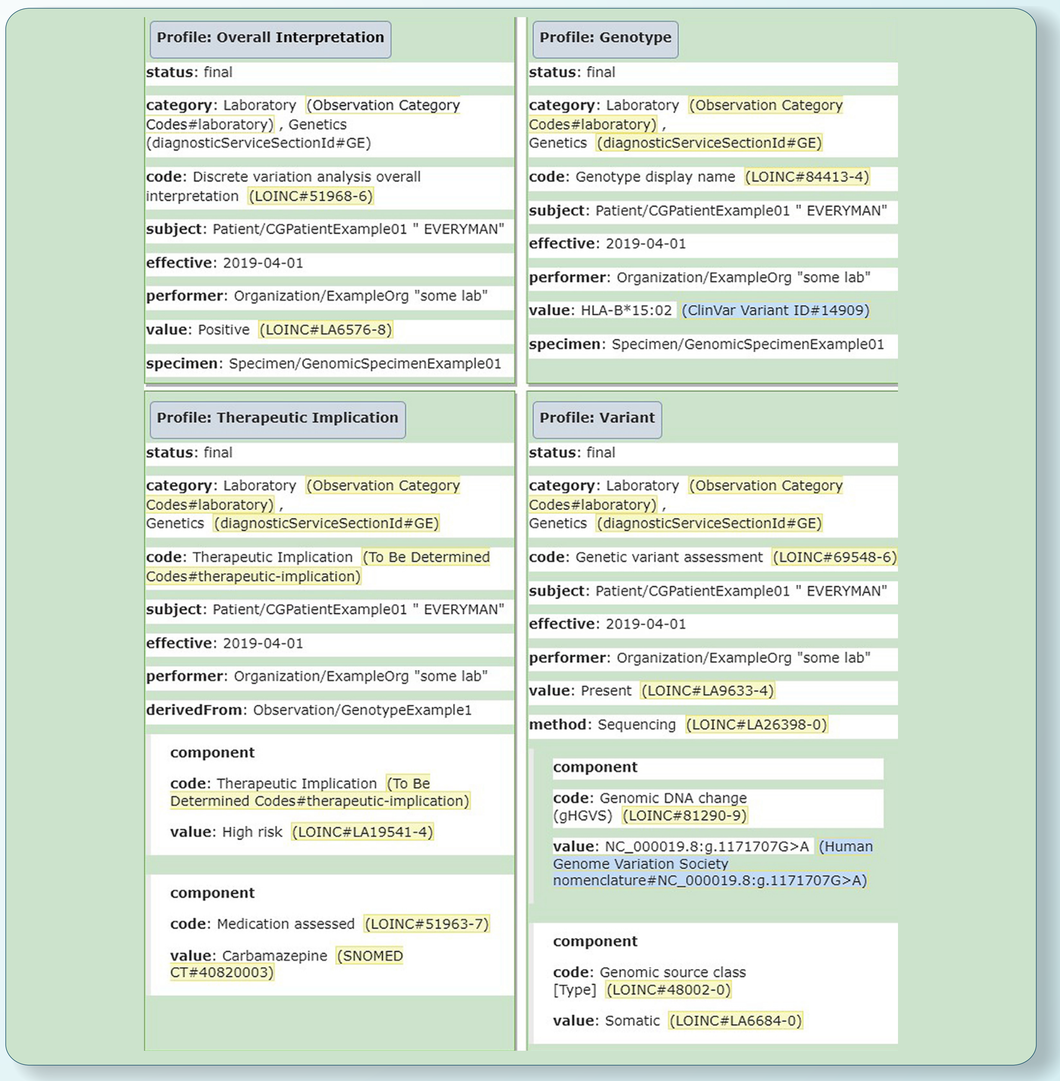

Fig (c) Conversion of patient and genomic data to FHIR resource

Challenges & Future Interoperability

- Lack of universal standard for genomic data.
- Genomic data are voluminous and complex
- Patient Engagement and Informed Consent
- Longitudinal Data Management
- Data Quality and Accuracy
- Resource Constraints
- Collaboration and Stakeholder Alignment

Conclusion

- Standards are the foundation for interoperability and consistent representation of data.
- Interoperability between EHRs and Clinical Genomics Systems ensures efficient collaboration and improved patient outcomes.
- Benefits in many applications including clinical sequencing, cancer screening, pharmacogenomics, precision medicine, public health reporting, and decision support lead to significantly improved diagnosis, prognosis, and treatment selection.

References

- **Electronic Health Records and Genomics** https://www.jmdjournal.org/article/S1525-1578(21)00327-5/
- **FHIR Genomics Reporting Implementation Guide** https://build.fhir.org/ig/HL7/genomics-reporting/
- **Genomics Code Systems** https://build.fhir.org/ig/HL7/genomics-reporting/codings.html

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