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Integration of Genomic Data into the Electronic Health Record: Current State and Future Directions

NHGRI Genomic Medicine XIII

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Disclosures

- I serve as associate medical director of Intermountain's Precision Genomics Sequencing Laboratory, that offers commercial sequencing services.

- **I do not have any affiliation with or financial interest in any EHR vendors.**

- **Opinions and viewpoints are my own and do not represent those of my employer or any EHR vendor.**

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Historical Background

I started in an institution that used Allscripts…

Moved to an institution that used Epic

….and implemented genomics…

Moved to an institution that uses Cerner

……..and implementing genomics again.

The vast majority of genetic data remains in PDF’s today.

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Genomic Data Integration Pilot

Imported genetic data on 1,177 patients from the eMERGE network

- 142 with pathogenic variants for CDC tier 1 conditions and Hereditary Hemochromatosis
  - BRCA1 and BRCA 2 Breast Cancer Susceptibility (28)
  - Lynch Syndrome (48)
  - Familial Hypercholesterolemia (62)

- Pharmacogenomic for all 1,177 patients
  - SLCO1B1, DYPD, TPMT, IFNL3, CYP2C9/VKORC1

- Constructed CDS and Patient/Provider facing information for all variants.

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Challenge 1

Disparate implementation needs for clinical geneticists (CGs) compared with primary care providers (PCPs).

Need: Define separate but compatible workflows for different genomics use cases.

EHR Vendors and Integration of Genetic Data

Three different vendors – three different approaches

- Epic - Population Health – In production
- Cerner – Clinical Genomics – In alpha phase
- Allscripts – Proactive Testing – In production

Challenge 2

Interface between the laboratory and the EHR

"No one wants to manually enter genetic test reports into the system."

Solutions:

1. Hire genetic secretaries
2. Work on interfaces and standards

EHR Vendors and Integration of Genetic Data

Three different vendors – two different standards

- Epic – HL7 V2.5
- Cerner – FHIR
- Allscripts – FHIR

"It doesn't matter if we use HL7 V2.5 or FHIR because laboratories don't support either."

Challenge 3

Maintenance of Clinical Information and CDS

- Lack of publicly available resources.
- Currently maintenance requires technical skills.
- Lack of open APIs.

Challenge 4

Classification and reclassification of variants

- Lack of communication between lab and clinicians.
- Potential for discordant results between PDF and structured data in the EHR.
- Maintenance of classification across patients.

Moving Beyond Initial Implementation…

Real-time use of entire sequence in patient care.

Challenges of using genomic data with machine learning/artificial intelligence.

Regulatory nightmare surrounding use of entire sequence.

Questions?