EMR Integration of Genomic Results and Automated Decision Support

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eMERGE EHR integration working group co-Chairs
Research and clinical practice co-exist to enable ongoing learning and evidence development

(Chambers, Feero, Khoury. JAMA. 2016)
Clinical and translational informatics challenges in a learning healthcare system

• Reproducibility
  • CDS at multiple institutions

• Timing & data quality
  • Upstream patient risk screening

• Diversity
  • Digital strategies to recruit populations while also minimizing sample disproportionality

• Replicability
  • Genomic variant interpretations may change
  • Clinical guidelines may change
Reproducibility: implications for clinical and research practice

• CDS at multiple institutions
  • Data is already in the EHR and ancillary systems and can potentially be acted upon
  • Standards: integrate ancillary systems, controlled vocabulary
  • Several potential models e.g., BYOD
Timing, inputs & data quality: implications for clinical and research practice

- CDS differ in timing that support is provided (before, during and after the clinical decision is made)
- Upstream patient risk screening with clinical decision support

Impact of data quality on trigger to match a condition?
- Can a risk algorithm at one site be used at another site?
- Should patient input be considered in personalized list (e.g., QOL)?

Capture timing, inputs & data quality requirements
Diversity: implications for clinical and research practice

- Digital strategies to recruit populations while also minimizing sample disproportionality

PATIENT PORTAL

You are eligible to participate in the following studies:

**STUDY 1**
- Agree
- Ignore/Hide

**STUDY 2**
- Agree
- Ignore/Hide

Please indicate if you agree to participate

- Who are we missing?

Support a range of recruitment strategies and multiple levels of health literacy
Replicability: implications for clinical and research practice

• **Clinical use example:** A 43-year-old female patient with a personal and family history of breast cancer undergoes sequencing analysis of BRCA1 and BRCA2. A missense VUS is reported in BRCA1 and reported as a VUS. Therefore it is not recommended that testing for this variant be used to determine risk in relatives of this patient. Nine months later, a revised laboratory report reclassifies the variant as pathogenic based on additional evidence. The EHR is updated to now follow the recommendations found in Diagnostic and Actionable categories.  

\[e\text{MERGE II & CSER (Shirts et al. 2015)}\]

• What changes have occurred?
• When were changes made?
• How do changes influence retrospective data analyses?
• What is the impact of changes on the patient and on research conclusions?

Tools to track provenance are needed
## Clinical & Translational Informatics Scope Considerations

<table>
<thead>
<tr>
<th>Dimension</th>
<th>Nature of Scope</th>
<th>Ways to Limit Scope</th>
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</thead>
<tbody>
<tr>
<td>Reproducibility</td>
<td>Creating CDS for use at multiple sites</td>
<td>Agreed upon standards and controlled vocabularies to integrate data from EHRs and ancillary systems</td>
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<tr>
<td></td>
<td></td>
<td>Agreed upon model to enable using the same CDS at multiple sites</td>
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<tr>
<td>Timing, inputs &amp; data quality</td>
<td>Upstream patient risk screening</td>
<td>Specify timing for CDS</td>
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<td>Specify inputs &amp; data quality requirements for the use of EHR phenotypes for risk screening</td>
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<tr>
<td>Diversity</td>
<td>Using digital strategies to enable the recruitment of diverse populations</td>
<td>Support a range of digital strategies based upon site needs</td>
</tr>
<tr>
<td>Replicability</td>
<td>Accounting for evidence changes when replicating previous research or clinical interpretations</td>
<td>Choose standards &amp; services that can be accessed across the network and that document provenance</td>
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These considerations are not new. Existing approaches should be assessed to determine if they are sufficient, should be improved, etc
Disclosure

Sandy Aronson works for Partners HealthCare which receives royalties on the sale of GeneInsight software which performs functions similar to some of the functions described in this presentation.

Sandy Aronson’s team also receives funding from Persistent Systems to develop an open source Health Innovation Platform as well as open source SMART on FHIR apps that run on the platform.
Core Contentions

• Demonstrating uses of genetics that produce clinical and/or economic value are important to the clinical genetics community

• Electronic Clinical Decision Support (CDS) will be a critical to the widespread adoption, use and safety of many clinical genetic techniques

• eMERGE could greatly accelerate the development and deployment of genetic aware CDS
However ....

• Development and deployment of CDS is very different than developing research IT infrastructure

• If eMERGE chooses to pursue CDS, appropriately focusing resources will not only be important for success but also for patient safety and ensure we “do no harm”

• There are many options for focused resource deployments that could be helpful
IT Support for Clinical Genetic Workflow Refinement

- Receive Results
- Store Results in EHR Ecosystem
- Event Based CDS
- Display Based CDS
- Implement New Clinical Workflow
- Measure Δ in Outcomes to Drive Cont Improvement
- Clinical and Other Non-Genetic Data Streams

- Provider
- Lab
- CDS Rule Repositories
- ClinVar and Other Knowledge Repositories

Ingest Knowledge Events

- ClinVar and Other Knowledge Repositories

Emerging Electronic Medical Records and Genomics Network
IT Support for Clinical Genetic Workflow Refinement

Focus of core eMERGE 3 Clinical IT Infrastructure Efforts

- CDS Rule Repositories
- ClinVar and Other Knowledge Repositories

Receive Results
- Store Results in EHR Ecosystem
- Event Based CDS
- Display Based CDS

Implement New Clinical Workflow

Measure Δ in Outcomes to Drive Cont Improvement

Clinical and Other Non-Genetic Data Streams

Ingest Knowledge Events

Provider

Lab

Lab

Provider

Provider
Considerations for eMERGE 4

• Display based (SMART on FHIR) vs Event Based (CDS Hooks) eCDS

• Generalized Genetic vs Clinical Condition Specific Functionality

• Site Specific Objectives vs Network Based Objectives

• Foundation Building vs End-to-End Value Focused
Event Based Clinical Scenario Specific eCDS

From: Development and use of active clinical decision support for preemptive pharmacogenomics
Genetic Specific Display Based eCDS

Prototype Developed by Partners HealthCare and Geisinger Teams
Condition Specific Display Based eCDS

Bill Layne
Rick Kauffman
HIP Development Team
<table>
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<tr>
<th>Option</th>
<th>Benefit</th>
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| **Develop Open Source SMART on FHIR App for Genetic Results**         | • Drive development and adoption of clinical genetic result and knowledge standards.  
| **Maintenance and Implement**                                         | • Get genetic data into the EHR ecosystem in structured form for use in other forms of eCDS  
| **Across Multiple Sites**                                             | • Address the problem of keeping clinicians up to date as genetic knowledge evolves                                                      |
| **Implement DIGITize Guide Rules**                                    | • Demonstrate capabilities of genetic aware event based eCDS  
| **Across Multiple Sites**                                             | • Improve patient safety in specific clinical areas  
|                                                                      | • Possible to consider interventions to assist any individual participating in decision points that occur during the clinical flow |
| **Develop Display Based CDS for a Specific Clinical Area**            | • Demonstrate ability to integrate genetic data into decision making within a specific clinical area  
|                                                                      | • Build support that facilitates new clinical workflow  
|                                                                      | • Likely more amenable to clinical and economic outcome studies  
|                                                                      | • Explore different modalities for different roles                                                                                       |
| **Build de-identified case and/or knowledge exchange network to**     | • Further drives standards development  
| **support any of the above**                                          | • Opens possibility for cross site continuous learning                                                                                  |
End to End IT Support for Clinical Genetic Workflow Involves Enormous IT Scope

Reproducibility, data quality, diversity, and replicability will be key considerations to generate usable data for CDS.

It will be critical for eMERGE IV to focus its resources on the scenarios and processes it would most like to support.
Backup
Standards

Knowledge Representations
Clinical Context Aware Constraints
Ontological
Patient Result Transmission Format

Levels of Refinement

Point to Point Transmission ➔ Homogeneous Network Use ➔ Heterogeneous Network Use

Research Only Use ➔ Clinical Pilot ➔ General Clinical Use

Enable Results Return ➔ Structured Results Display ➔ Enable eCDS
### Why is eCDS Deployment So Resource Intensive?

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<tr>
<th>Issue</th>
<th>Implication</th>
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<tr>
<td>Clinicians resist functionality that does not fit their workflow like a glove</td>
<td>Lots of User Experience design expertise and iteration required</td>
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<td>Corner cases can harm patients</td>
<td>Lots of validation required</td>
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<tr>
<td>The clinical data logic depends upon often not readily available</td>
<td>Often requires support from IT groups throughout the enterprise</td>
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<tr>
<td>Usually requires enterprise integration</td>
<td>Integrating with multipurpose enterprise systems creates support and enhancement complexity</td>
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