EMR Integration of Genomic Results and Automated Decision Support

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eMERGE EHR integration working group co-Chairs

eMERGE & Beyond: The Future of Electronic Medical Records (EMR) and Genomics
October 30th, 2017
Rockville, MD

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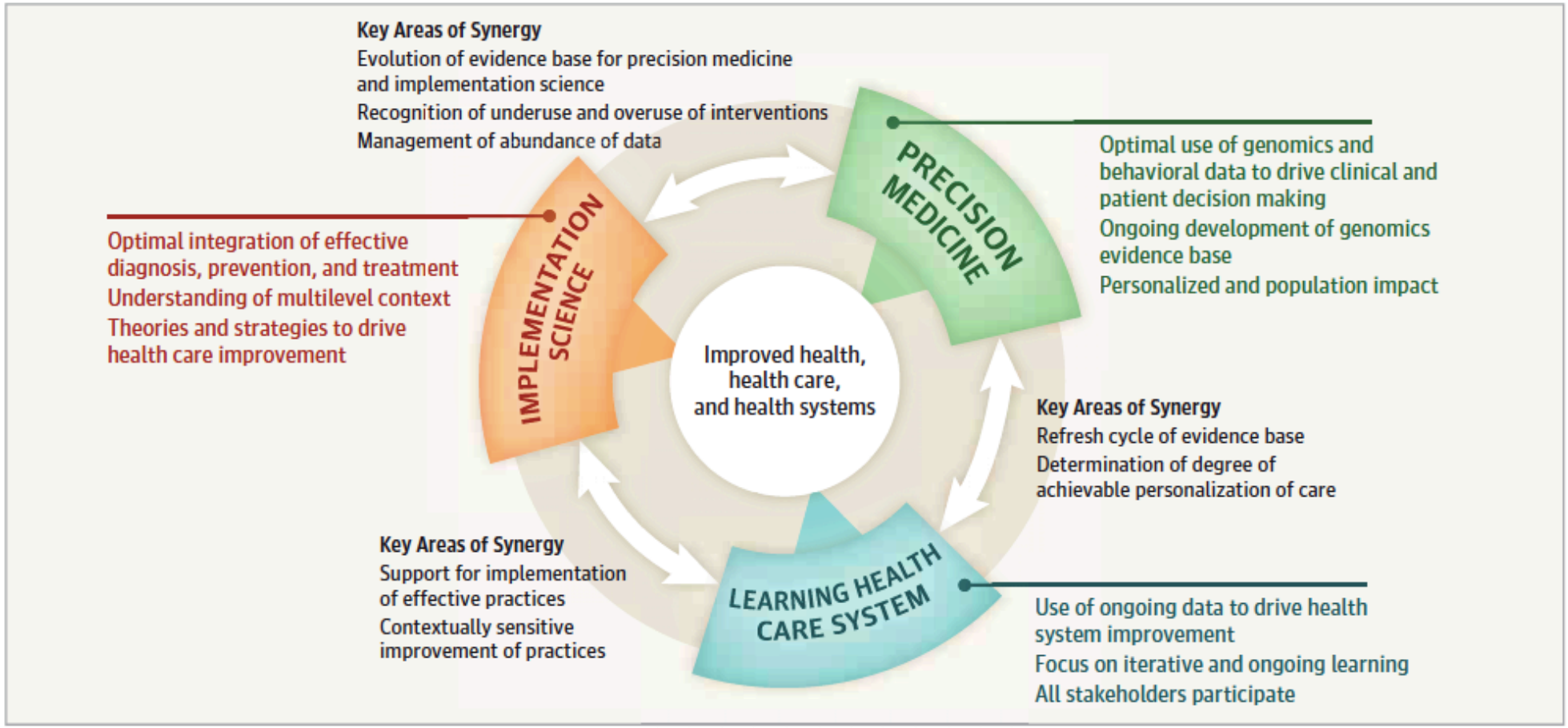
Research and clinical practice co-exist to enable ongoing learning and evidence development

Figure. Contributions of Implementation Science, Learning Health Care System, and Precision Medicine



(Chambers, Feero, Khoury. JAMA. 2016)

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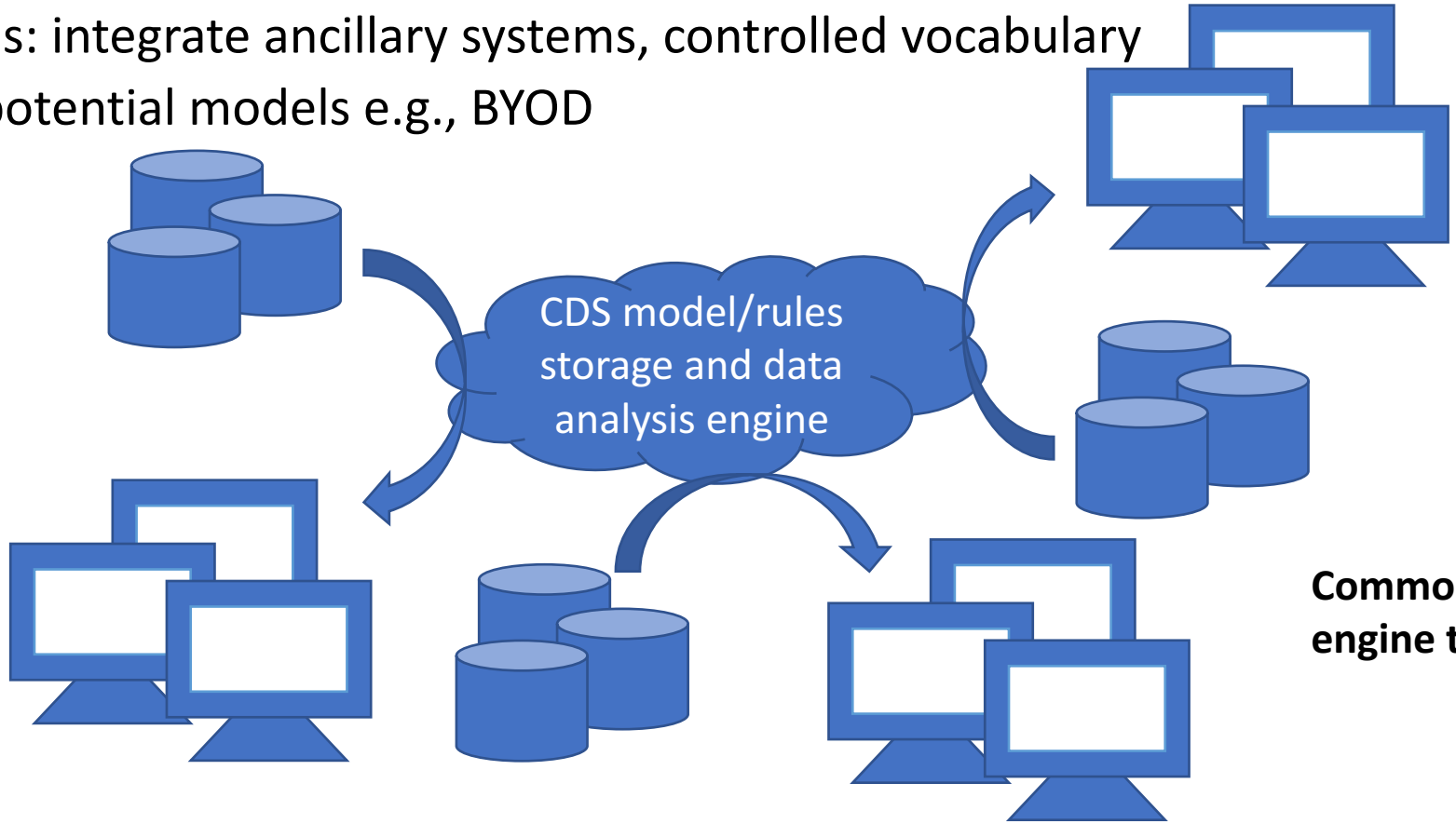
Clinical and translational informatics challenges in a learning healthcare system

- 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

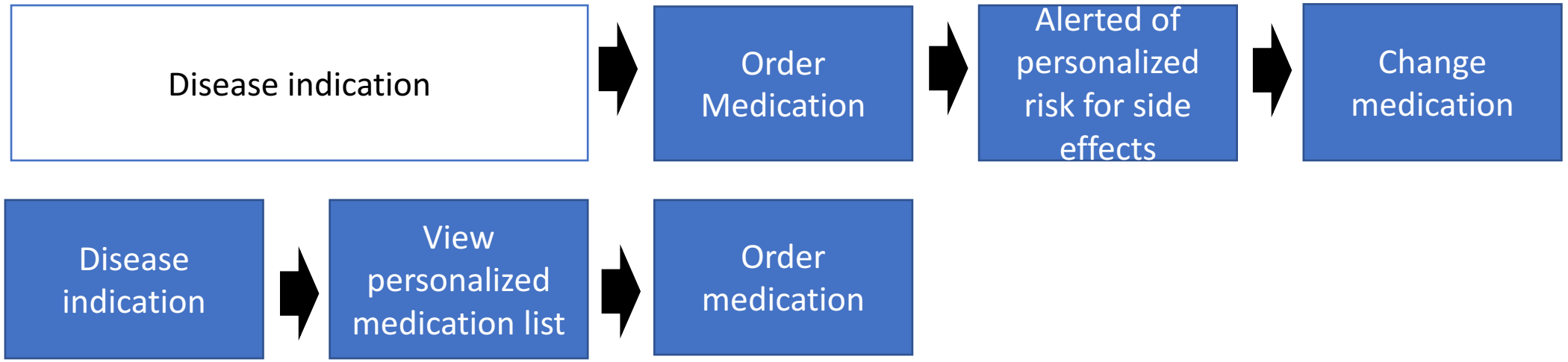
- CDS at multiple institutions
  - Data is already in the EHR and ancillary systems and can potentially be acted upon
  - Standards: integrate ancillary systems, controlled vocabulary
  - Several potential models e.g., BYOD



Common data analysis or rules engine to enable shared eCDS

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

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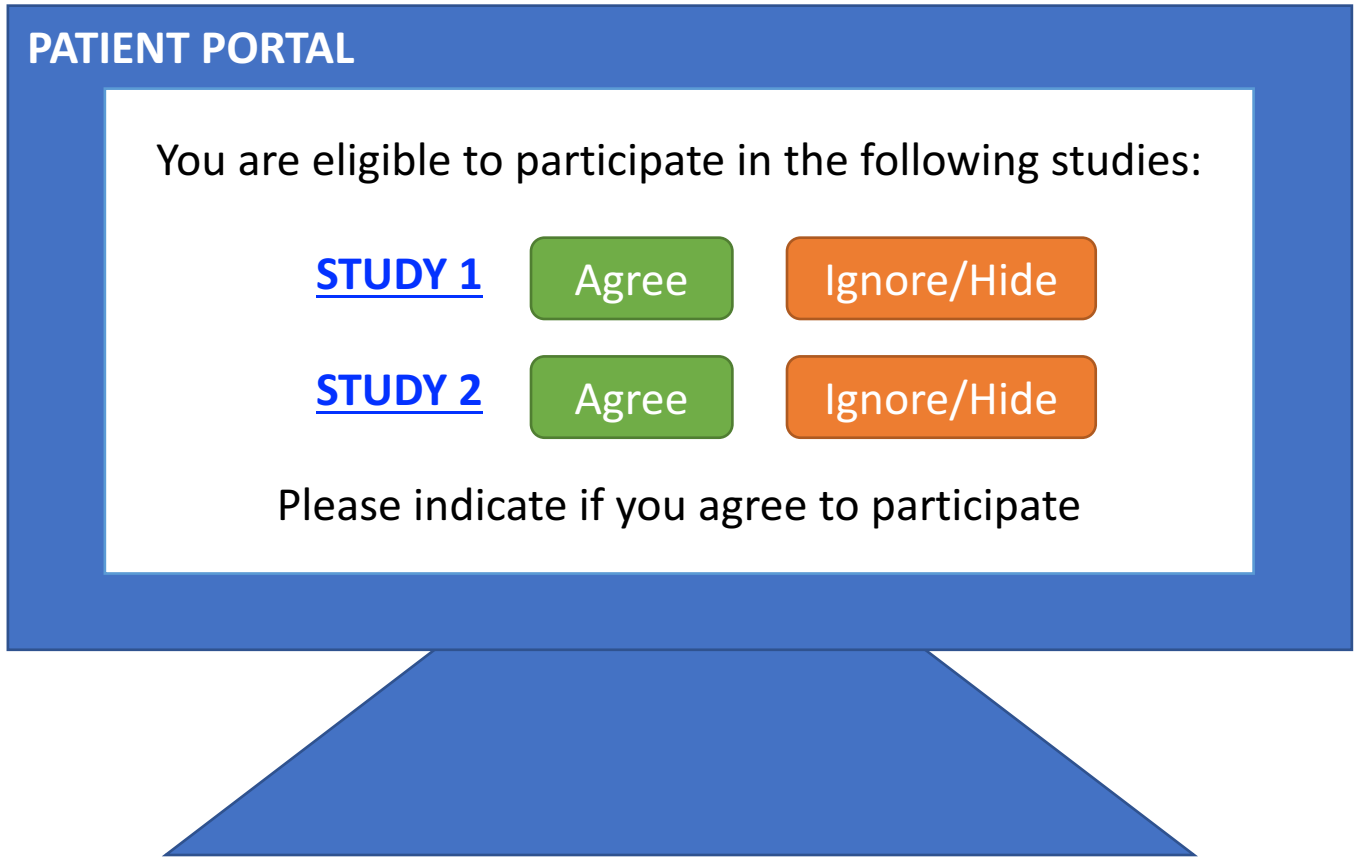
Diversity: implications for clinical and research practice

- Digital strategies to recruit populations while also minimizing sample disproportionality



- Who are we missing?

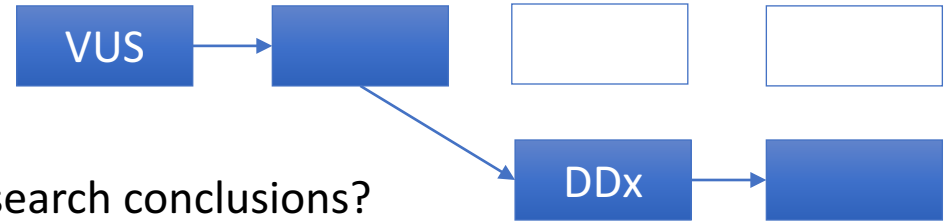
Support a range of recruitment strategies and multiple levels of health literacy

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.**

eMERGE 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

Dimension	Nature of Scope	Ways to Limit Scope
Reproducibility	Creating CDS for use at multiple sites	Agreed upon standards and controlled vocabularies to integrate data from EHRs and ancillary systems Agreed upon model to enable using the same CDS at multiple sites
Timing, inputs & data quality	Upstream patient risk screening	Specify timing for CDS Specify inputs & data quality requirements for the use of EHR phenotypes for risk screening
Diversity	Using digital strategies to enable the recruitment of diverse populations	Support a range of digital strategies based upon site needs
Replicability	Accounting for evidence changes when replicating previous research or clinical interpretations	Choose standards & services that can be accessed across the network and that document provenance

These considerations are not new. Existing approaches should be assessed to determine if they are sufficient, should be improved, etc

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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

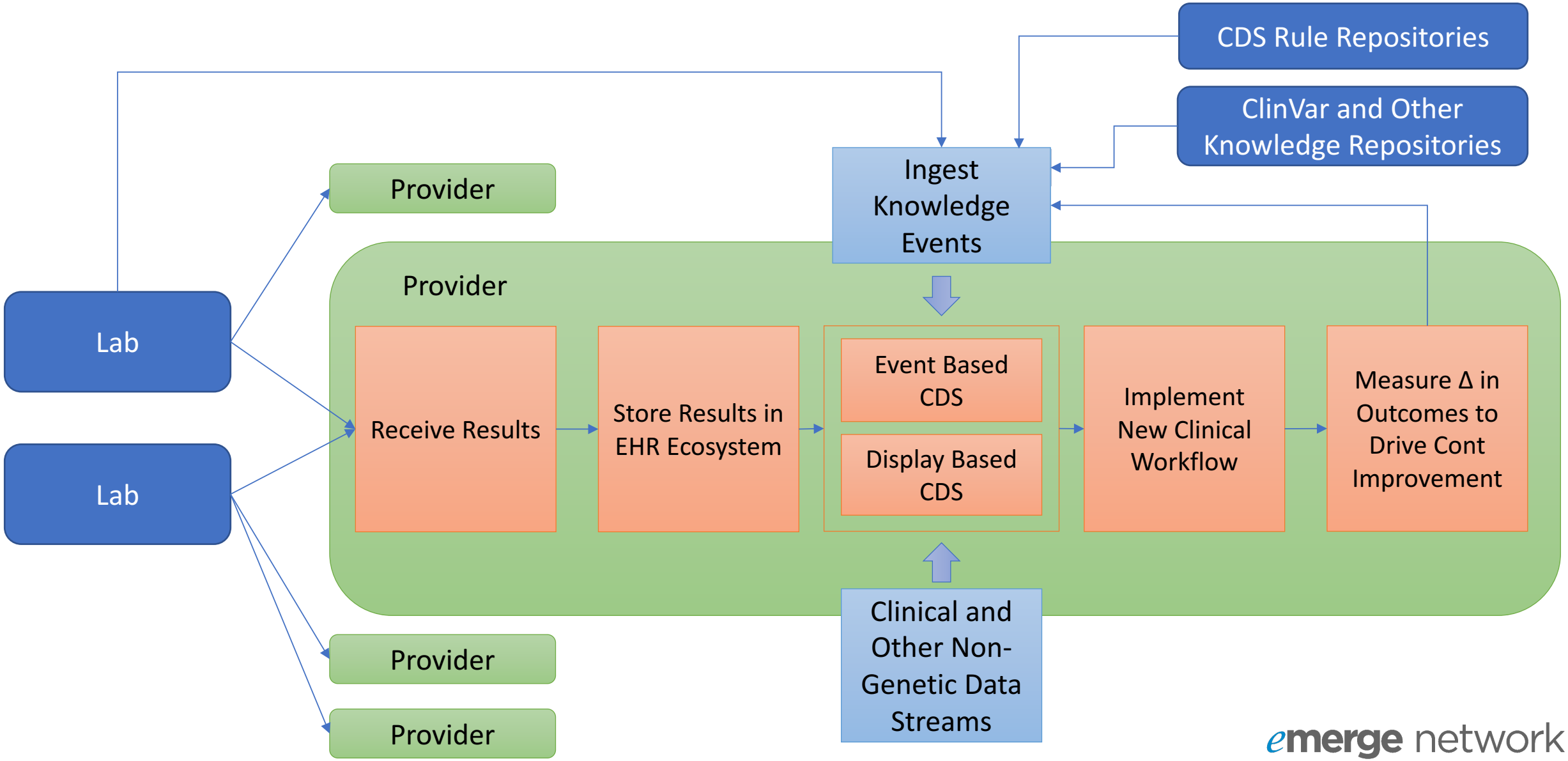
However ….

- 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



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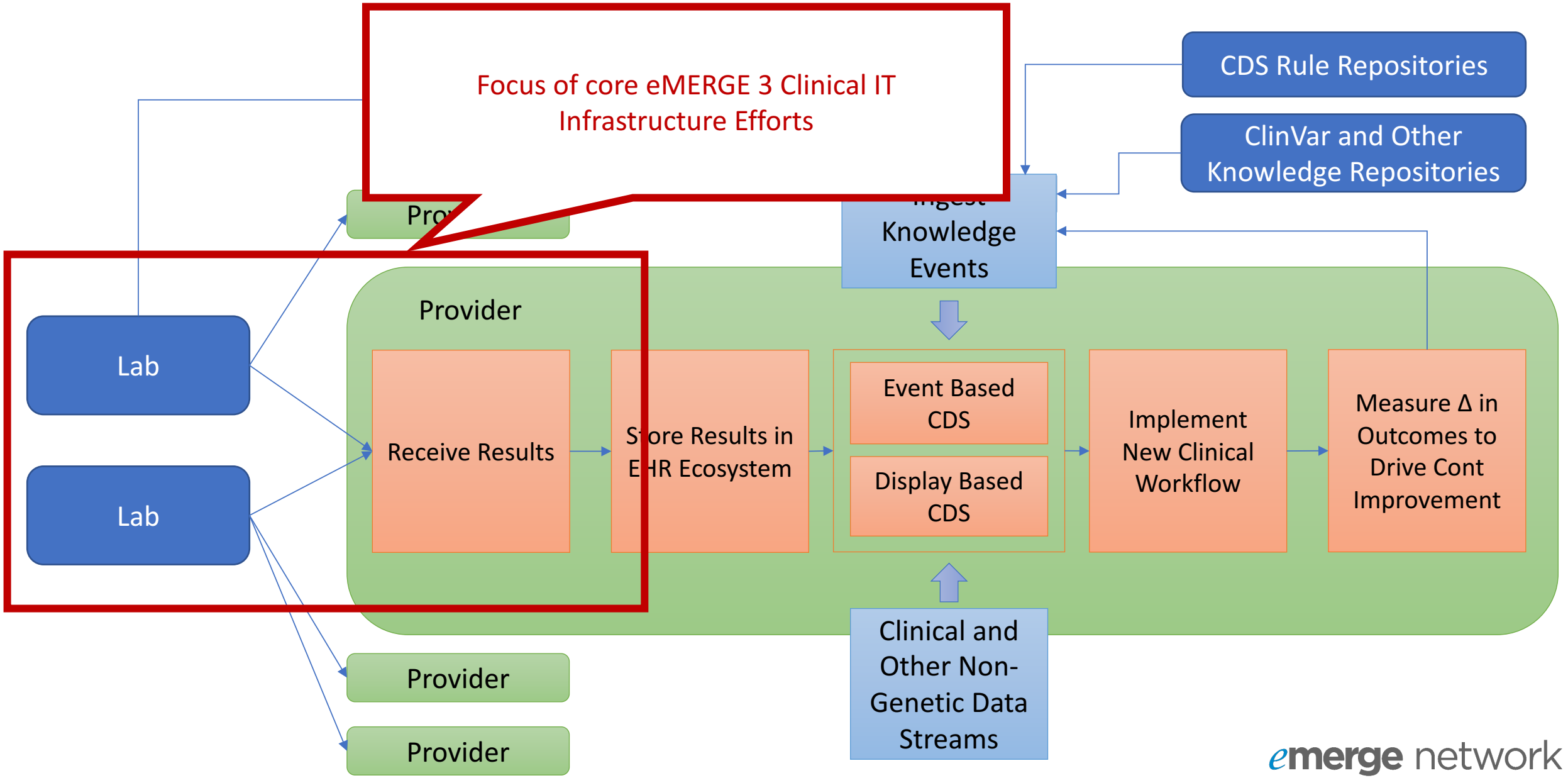
IT Support for Clinical Genetic Workflow Refinement



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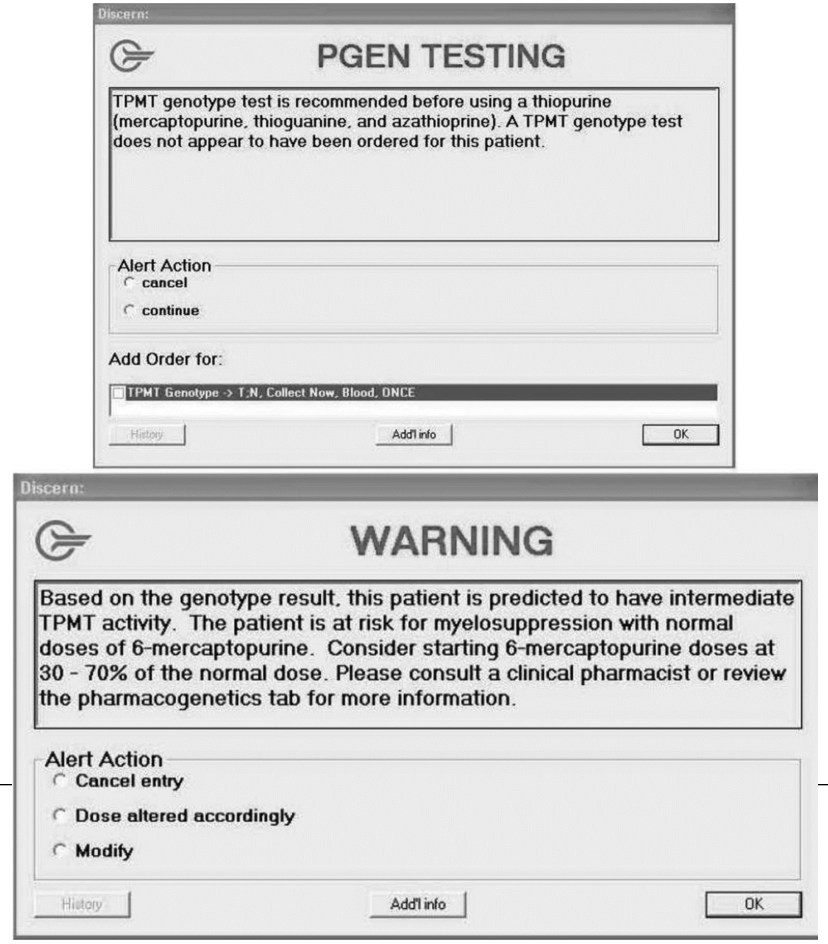
Considerations for eMERGE 4

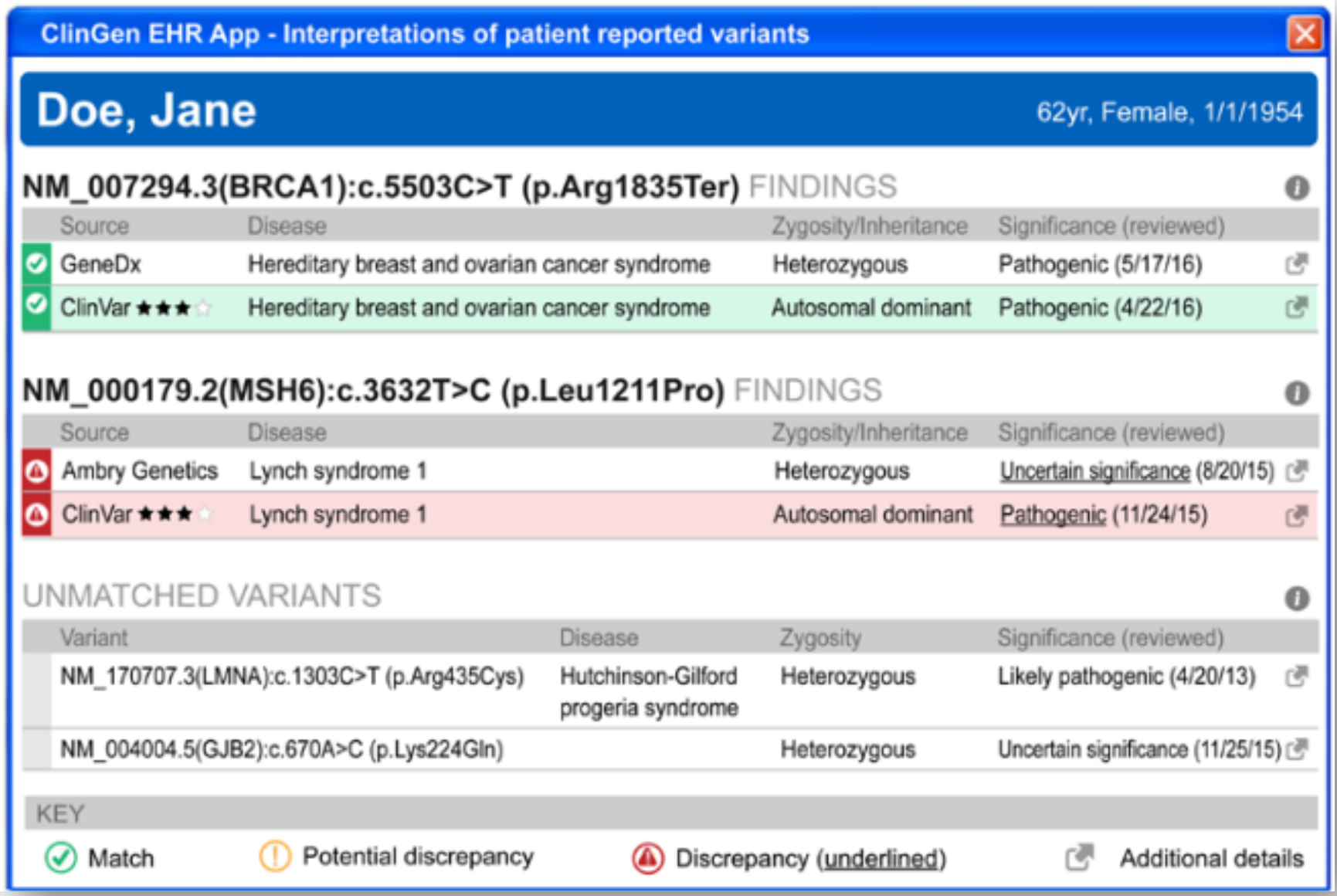
- Display based (SMART on FHIR) vs Event Based (CDS Hooks) eCDS
- Generalized Genetic vs Clinical Condition Specific Functionality
- Site Specific Objectives vs Network Based Objectives
- Foundation Building vs End-to-End Value Focused

Event Based Clinical Scenario Specific eCDS



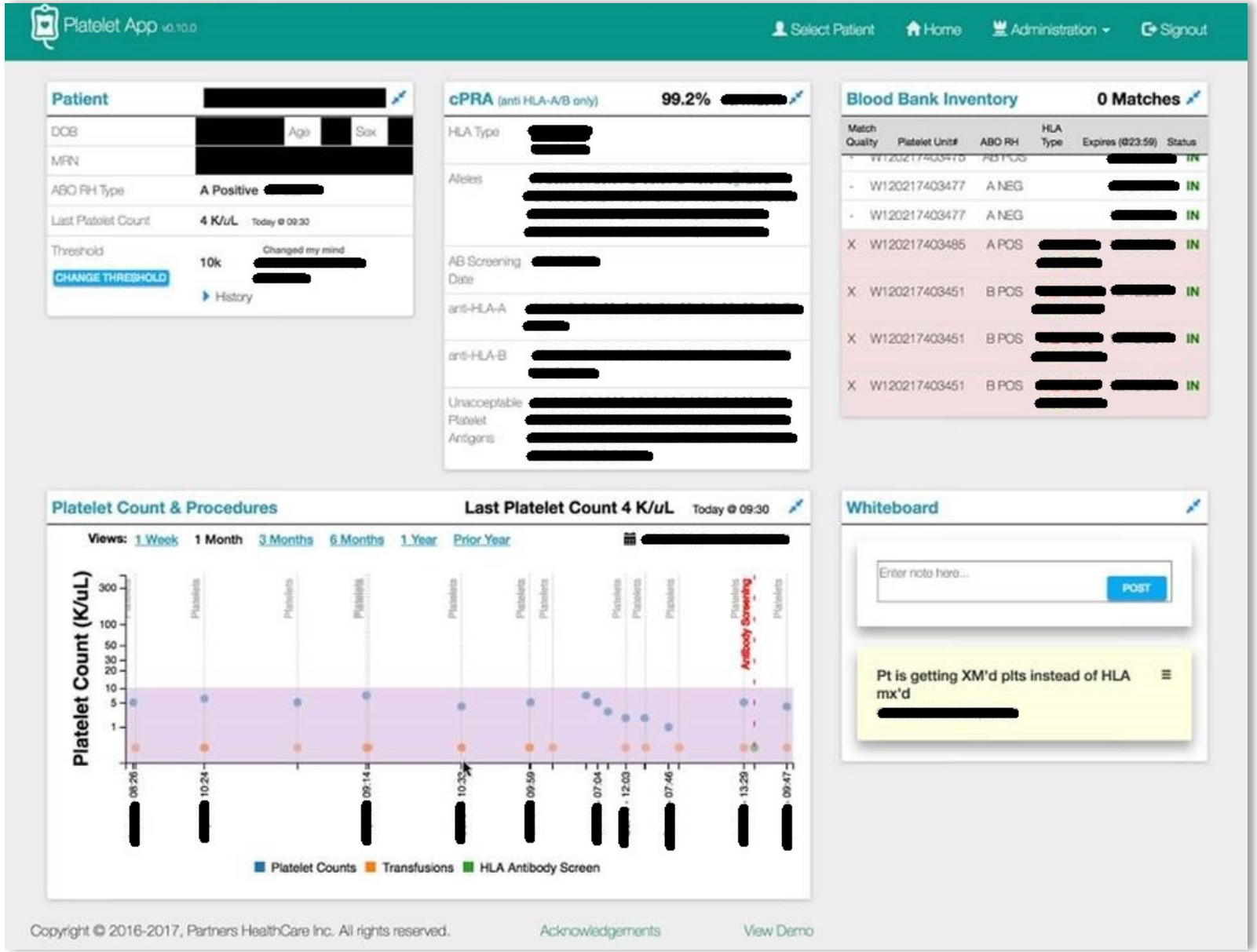
From: Development and use of active clinical decision support for preemptive pharmacogenomics
J Am Med Inform Assoc. 2013;21(e1):e93-e99. doi:10.1136/amiajnl-2013-001993

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

Specific Options

Option	Benefit	
Develop Open Source SMART on FHIR App for Genetic Results Maintenance and Implement Across Multiple Sites	• Drive development and adoption of clinical genetic result and knowledge standards. • Get genetic data into the EHR ecosystem in structured form for use in other forms of eCDS • Address the problem of keeping clinicians up to date as genetic knowledge evolves	
Implement DIGITizE Guide Rules Across Multiple Sites	• Demonstrate capabilities of genetic aware event based eCDS • 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 • Explore different modalities for different roles
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	
Build de-identfied case and/or knowledge exchange network to support any of the above	• Further drives standards development • Opens possibility for cross site continuous learning	

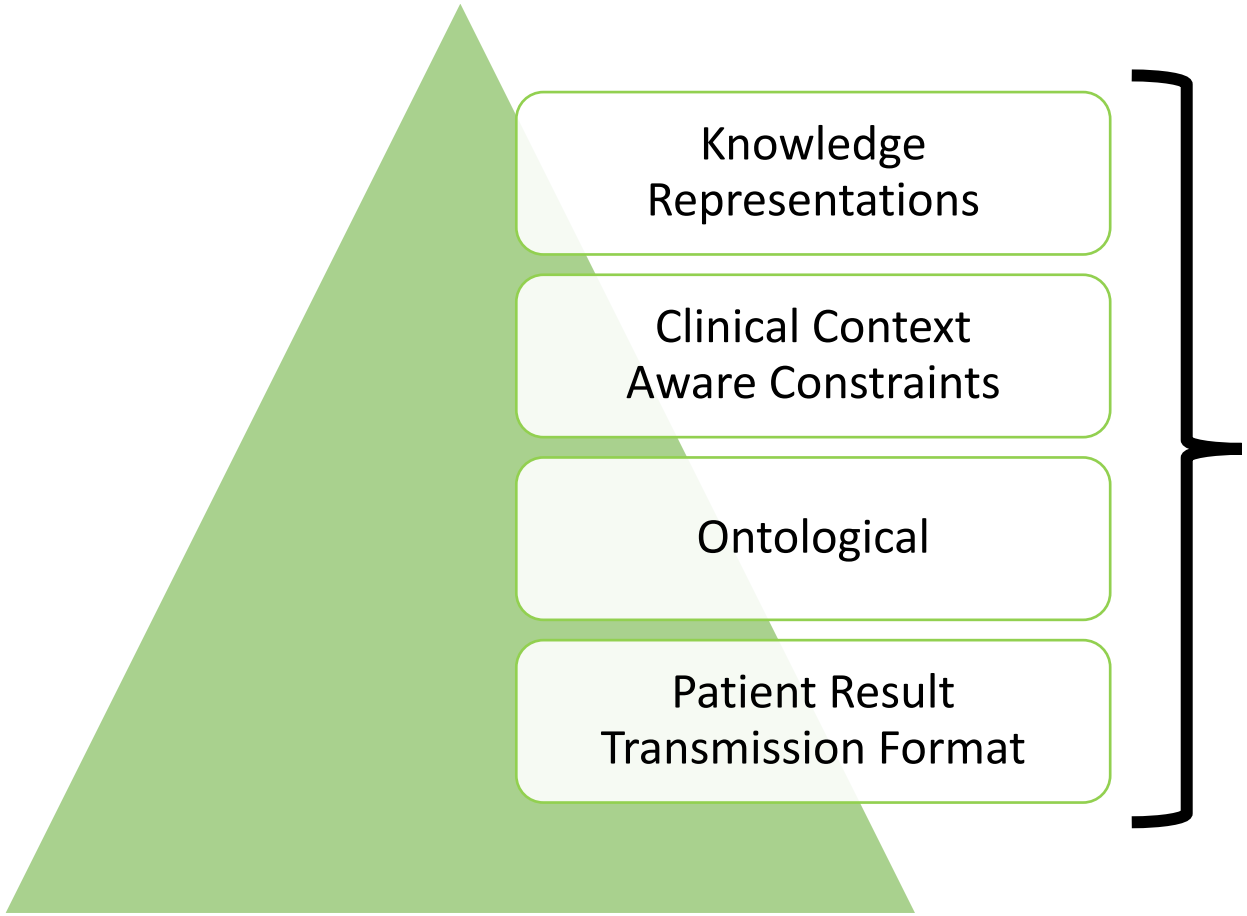
End to End IT Support for Clinical Genetic Workflow Involves Enormous IT Scope

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



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?

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