

Knowledge Management for Genomic Clinical Decision Support

Casey Overby Taylor, PhD

Assistant Professor of Medicine, Johns Hopkins University

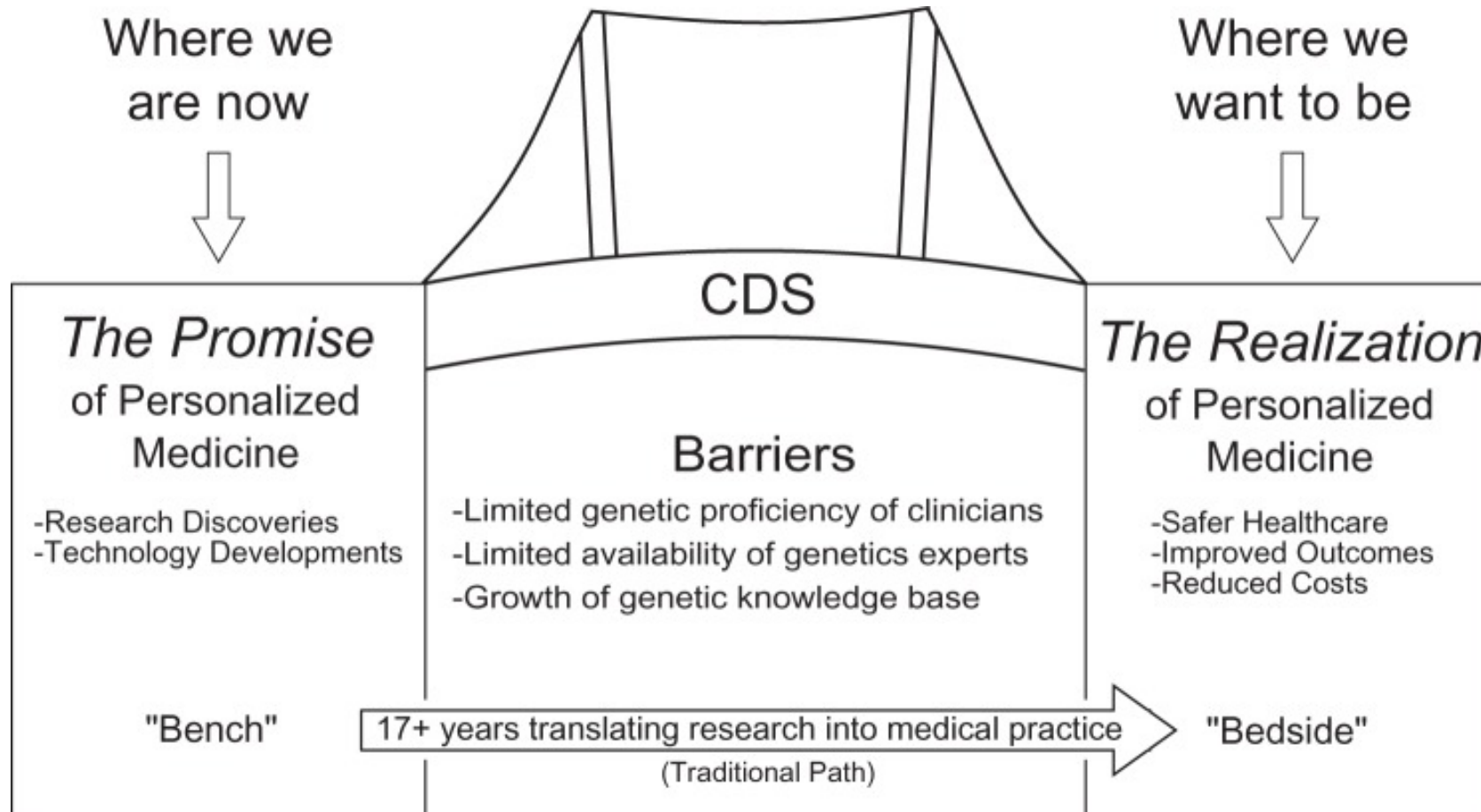
Adjunct Investigator, Geisinger Health System

Co-Chair, eMERGE EHR Integration Working Group

Previous Co-Chair, IGNITE Clinical Informatics Interest Group

Member, ClinGen EHR Workgroup

Clinical decision support as a bridge to overcome barriers to realizing precision medicine



Outline

- Challenges for Genomic Clinical Decision Support (gCDS)
- Implementation Science and gCDS
- Focus of gCDS implementation in eMERGE III
- Overview of managing shared knowledge for gCDS
- Tools to enable gCDS knowledge management (efforts from NHGRI-funded projects)




Highlighted challenges to...

- Managing shared knowledge
- Improving effectiveness
- Establishing decision support architecture and standard approaches

Invited Commentary | Published: 19 September 2013

Opportunities for genomic clinical decision support interventions

Casey Lynnette Overby PhD , Isaac Kohane MD, PhD, Joseph L Kannry MD, Marc S Williams MD, Justin Starren MD, PhD, Erwin Bottinger MD, Omri Gottesman MD, Joshua C Denny MD, MS, Chunhua Weng PhD, MS, Peter Tarczy-Hornoch MD & George Hripcsak MD, MS

Genetics in Medicine **15**, 817–823 (2013) | [Download Citation](#) ↓

Managing shared knowledge for gCDS

- Knowledge management solutions often are not accepted without customization
- Reliance on expert communities

Review | [OPEN](#) | Published: 10 January 2013

Implementing genomic medicine in the clinic: the future is here

[Teri A. Manolio MD, PhD](#) , [Rex L. Chisholm PhD](#), [Brad Ozenberger PhD](#), [Dan M. Roden MD](#), [Marc S. Williams MD](#), [Richard Wilson PhD](#), [David Bick MD](#), [Erwin P. Bottinger MD](#), [Murray H. Brilliant PhD](#), [Charis Eng MD, PhD](#), [Kelly A. Frazer PhD](#), [Bruce Korf MD, PhD](#), [David H. Ledbetter PhD](#), [James R. Lupski MD, PhD](#), [Clay Marsh MD](#), [David Mrazek MD](#), [Michael F. Murray MD](#), [Peter H. O'Donnell MD](#), [Daniel J. Rader MD](#), [Mary V. Relling PharmD](#), [Alan R. Shuldiner MD](#), [David Valle MD](#), [Richard Weinshilboum MD](#), [Eric D. Green MD, PhD](#) & [Geoffrey S. Ginsburg MD, PhD](#)

Genetics in Medicine **15**, 258–267 (2013) | [Download Citation](#) ↓

Challenges and Barriers to Clinical Decision Support (CDS) Design and Implementation Experienced in the Agency for Healthcare Research and Quality CDS Demonstrations

Prepared for:
Agency for Healthcare Research and Quality
U.S. Department of Health and Human Services
540 Gaither Road
Rockville, MD 20850
www.ahrq.gov

Contract Number: 290-04-0016

Prepared by:
AHRQ National Resource Center for Health Information Technology

Authors:
June Eichner, M.S.
Maya Das, M.D., J.D.
NORC at the University of Chicago

AHRQ Publication No. 10-0064-EF
March 2010



Improving the effectiveness of gCDS

- Lack of institutional and clinical acceptance of supporting evidence
- UI characteristics, information content & integration with workflow & decision making processes
- **More work needed to understand how these features translate to acceptance of gCDS**

Decision support architecture and standard approaches for gCDS

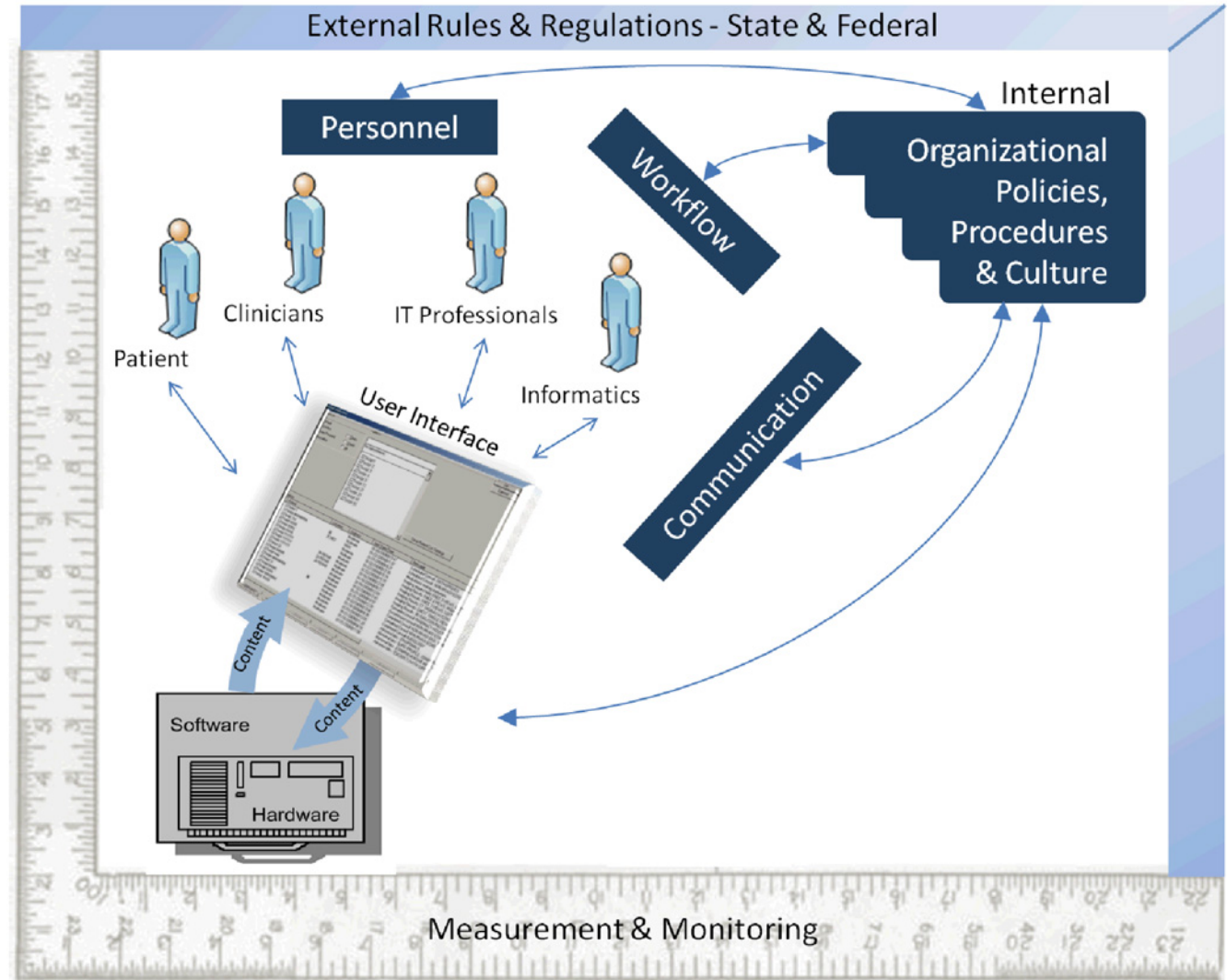
- Variation in decision support architecture
- Standards are needed to scale
- But, there are also limitations to using standards
 - Too many to choose from
 - Constrain what a user can encode to what was included in the scope of the standard

Implementation Science & Genomic Clinical Decision Support Implementation

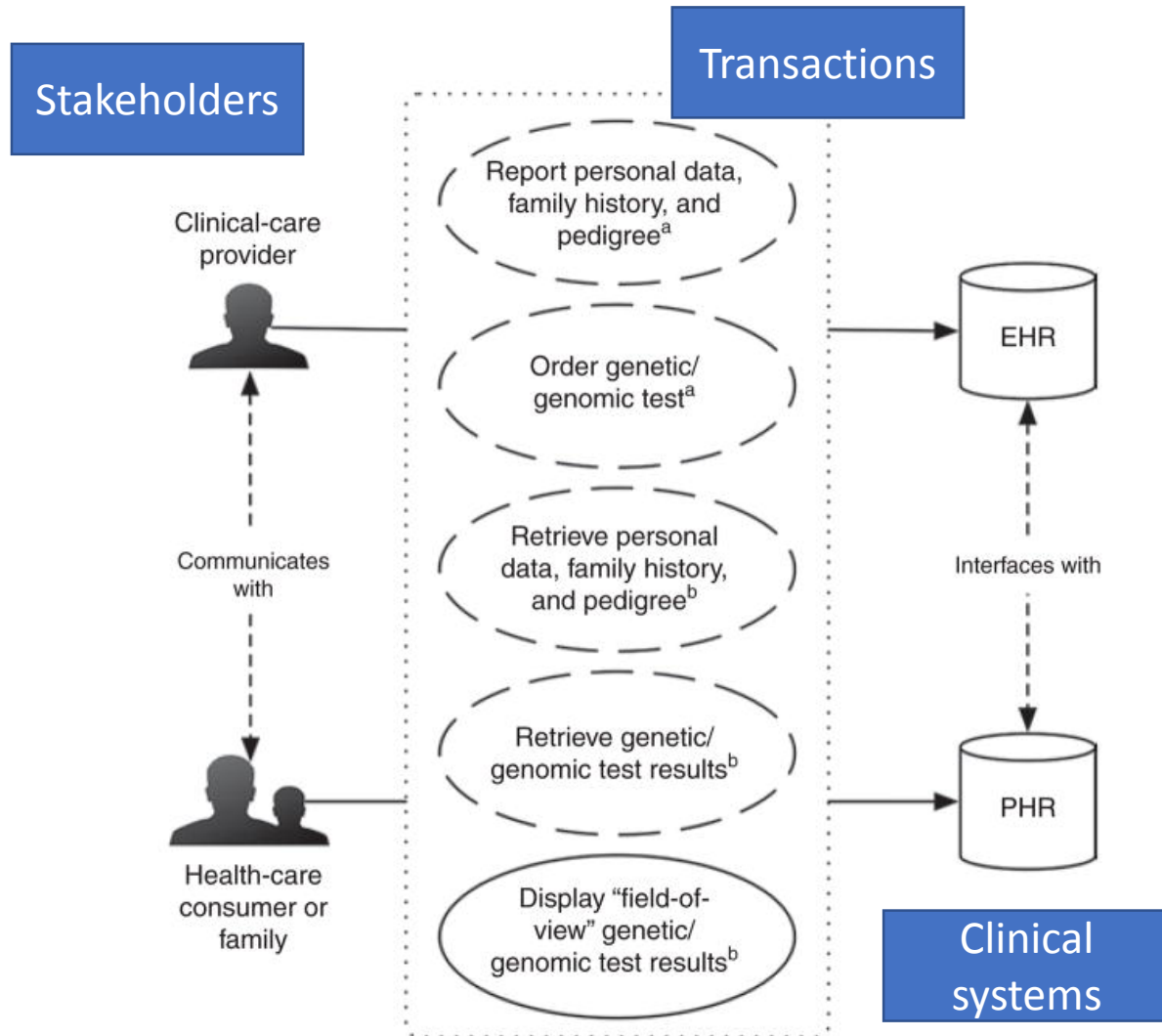
- Implementation science has an emphasis on the “what”
- gCDS specifications aligned with evidence
 - The “what” is defined in the context of current IT capabilities
 - Insufficient decision support technology (*Manolio TA. et al. Sci Transl Med 2015*)
 - May require additional IT development and resources
- There are often non-technical decision support solutions that can be used (e.g., initial study team involvement)

Frameworks to assess implementation challenges and guide local approaches to CDS implementation

- Ten key considerations for successful implementation (*Cresswell et al. JAMIA 2013*)
- Eight-dimension conceptual model (*Sittig and Singh, Qual Saf Health Care 2010*)
- *Others..*

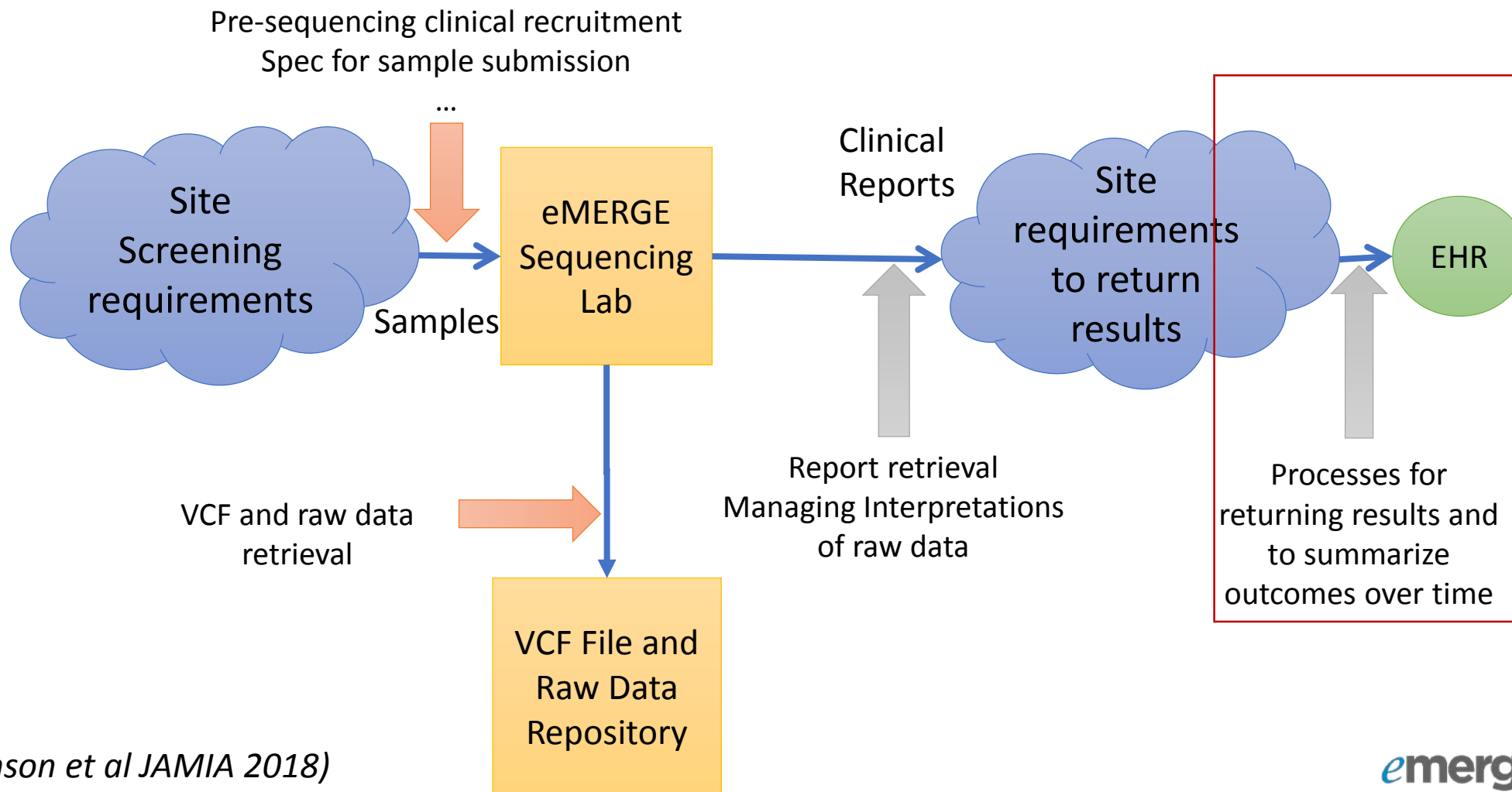


Framework for defining “what gCDS?”



- What are relevant transactions for this activity?
- When should this activity occur (i.e., what phases?)
- How should this activity be initiated and by who?
- Where should data be pushed to or pulled from?

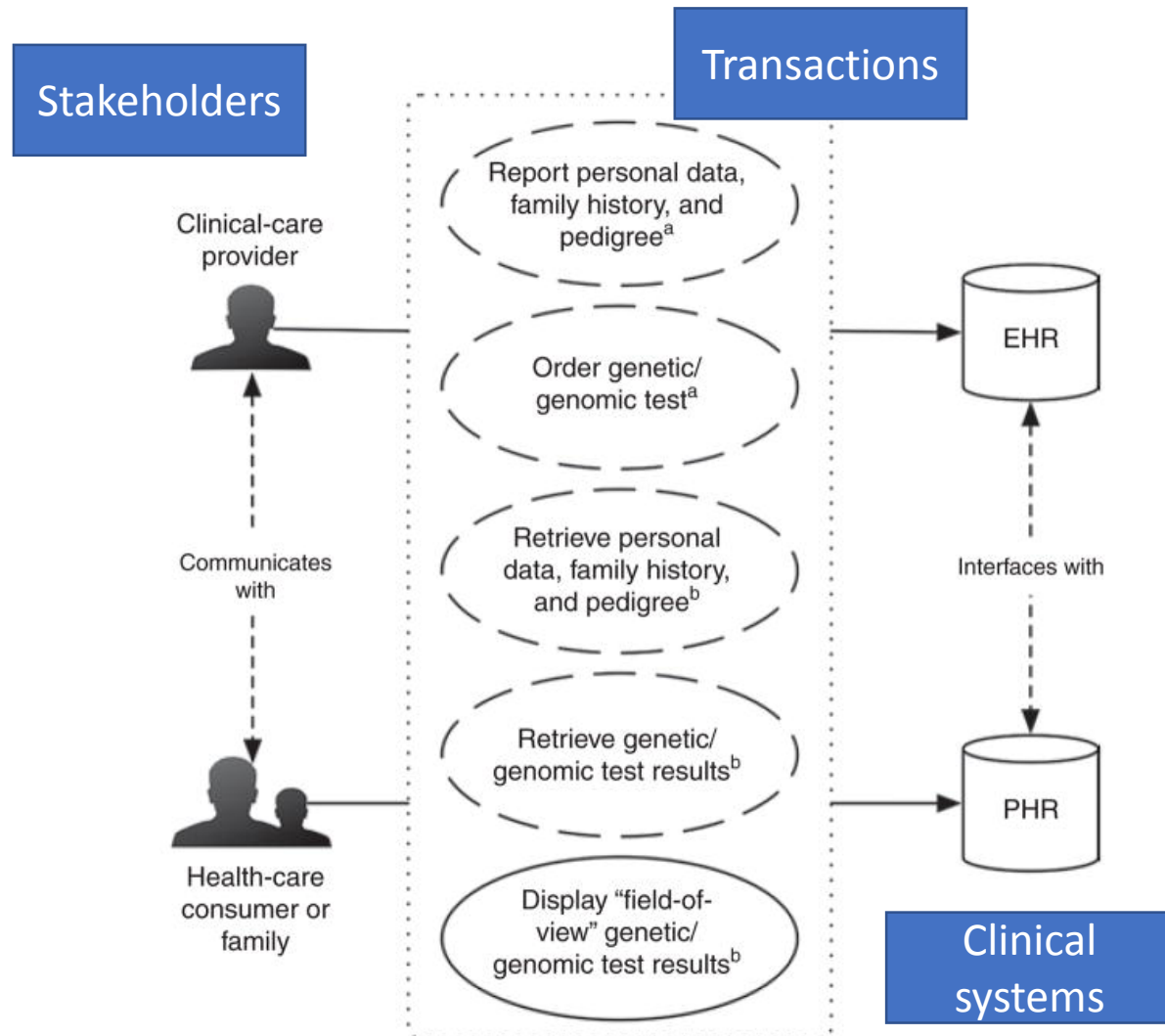
eMERGE III high level processes – “what gCDS?” is relatively defined



(Aronson et al JAMIA 2018)

Framework for defining “what gCDS?”

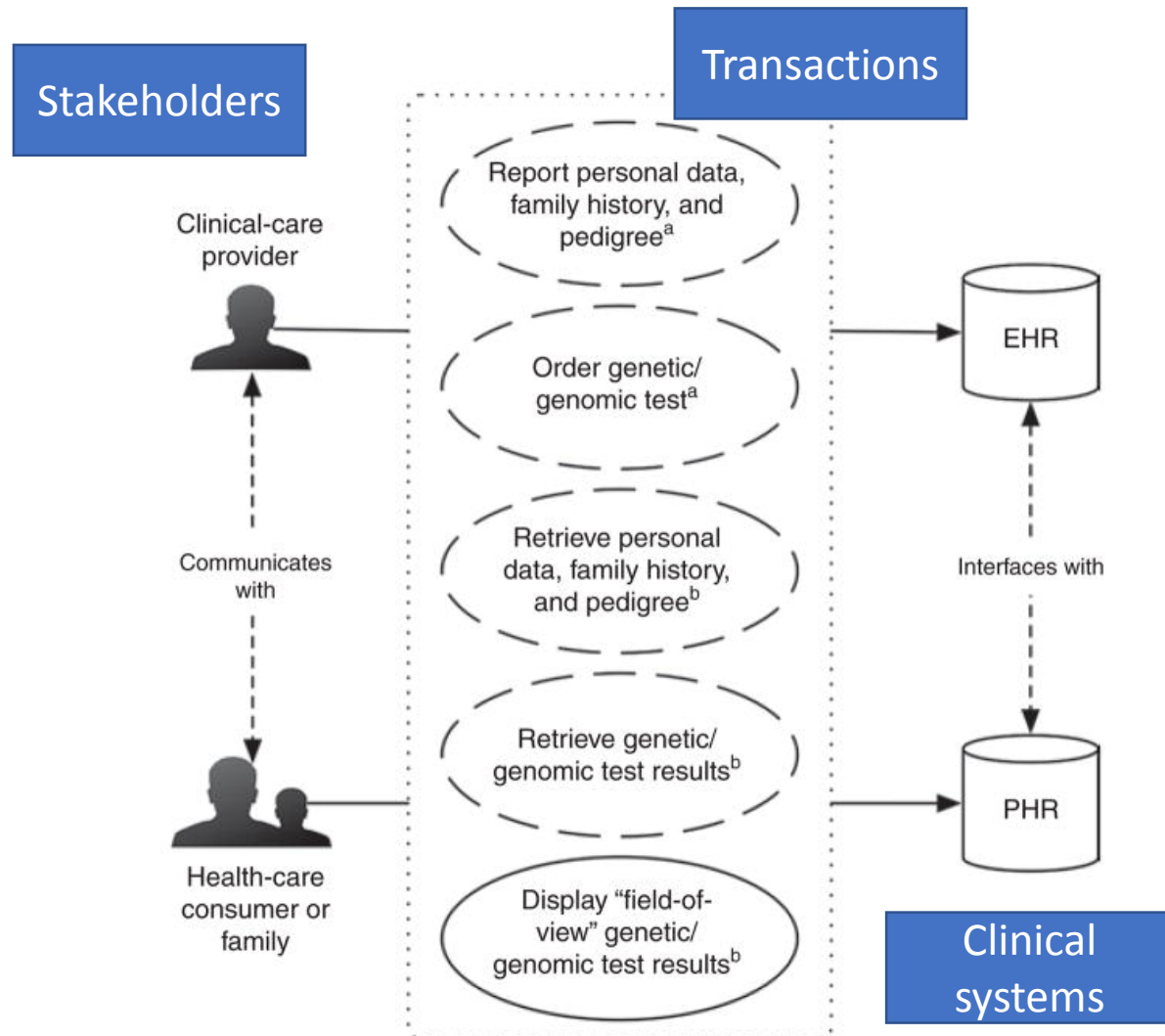
gCDS for Return of Results



- What are relevant transactions for this activity?
 - Retrieve genetic/genomic test results
- When should this activity occur (i.e., what phases?)
 - Post-analytic phase
- How should this activity be initiated and by who?
 - Health care provider
- Where should data be pushed to or pulled from?
 - EHR

Framework for defining “what gCDS?”

gCDS for Patient Screening



- What are relevant transactions for this activity?
 - Report personal data, family history and pedigree
- When should this activity occur (i.e., what phases?)
 - Pre-analytic phase
- How should this activity be initiated and by who?
 - Human-initiated by the health-care consumer
- Where should data be pushed to or pulled from?
 - PHR

gCDS for Patient Screening

- What are relevant transactions for this activity?
 - Report personal data, family history and pedigree
 - CDS content: Documentation template for data collection
- When should this activity occur (i.e., what phases?)
 - Pre-analytic phase
 - Setting: Outpatient
 - Workflow context: Between visits
- How should this activity be initiated and by who?
 - Human-initiated by the health-care consumer
 - Target user: patient
- Where should data be pushed to or pulled from?
 - PHR
 - CDS technologies: internal off-the-shelf functionality
 - CDS capabilities: active CDS
 - CDS features: trigger time, input data element, intervention , offered choice

(Note: some features are included in CDS taxonomies proposed by Wright et al. JAMIA 2007 & Wright et al. JAMIA 2011)

Outline

- Challenges for Genomic Clinical Decision Support (gCDS)
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- **Overview of managing shared knowledge for gCDS**
- **Tools to enable gCDS knowledge management (efforts from NHGRI-funded projects)**

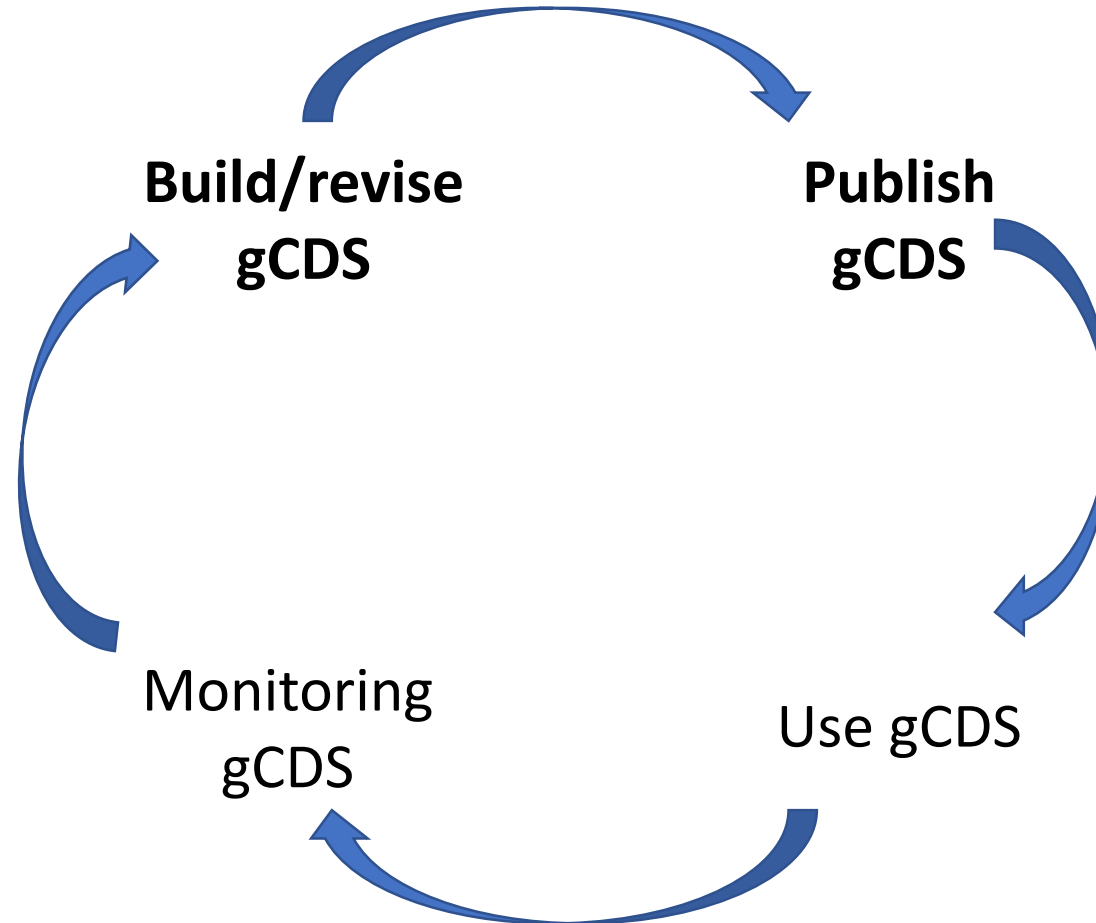
Managing shared knowledge for gCDS

Knowledge sources

- Clinical practice guidelines
- Resources aligned with healthcare org local policies

Data sources

- EHR
- Sequencing lab
- Patient (Study team)



Computable gCDS

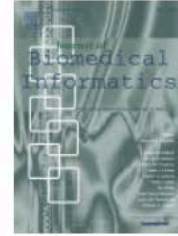
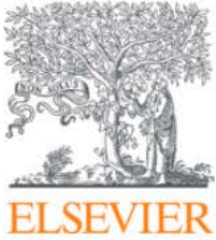
- Health care org local IT
- Clinical labs (structured interpretations)

Application areas

- Treatment
- Diagnosis
- Disease prevention (acute)

Needs for managing shared knowledge for gCDS

- Build/Revise gCDS
 - Provide guidance on implementation process
 - SPARK toolbox - “Building and implementation guide” (Kristin Weitzel, IGNITE network)
 - Better engage stakeholders in gCDS design process
 - *Opportunity for new tool development*
- Publish gCDS
 - Avoid re-inventing the wheel through sharing published gCDS (Related to NHGRI-funded efforts)
 - gCDS sandbox
 - Genomic Resources Search
 - DocUBuild
 - CDS_KB
 - *Consider tools developed in other communities (e.g., CPIC, PCORI, AHRQ, Vendor-specified, etc)



Special Communication

The genomic CDS sandbox: An assessment among domain experts

Ayesha Aziz ^a  , Kensaku Kawamoto ^b , Karen Eilbeck ^b , Marc S. Williams ^c ,
Robert R. Freimuth ^d , Mark A. Hoffman ^e , Luke V. Rasmussen ^f , Casey L. Overby ^g 
, Brian H. Shirts ^h , James M. Hoffman ⁱ , Brandon M. Welch ^a 

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<https://doi.org/10.1016/j.jbi.2015.12.019>

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Highlights

- There is a need to promote development of resources for gCDS.
- The proposed sandbox will be available pre-configured with CDS and genome tools.
- We present survey results to assess needs for a genomic CDS sandbox.
- Results show strong interest for a sandbox to test CDS and genome case studies.

gCDS Sandbox

(Outcome of Genomic Medicine Meeting VII:
Genomic Clinical Decision Support)

Integrating Genomic Resources with Electronic Health Records using the HL7 Infobutton Standard

[Bret S.E. Heale](#),^{1,7} [Casey Lynnette Overby](#),^{2,4} [Guilherme Del Fiol](#),¹ [Wendy S. Rubinstein](#),
[Donna R. Maglott](#),³ [Tristan H. Nelson](#),⁴ [Aleksandar Milosavljevic](#),⁵ [Christa L. Martin](#),⁴
[Scott R. Goehringer](#),⁴ [Robert R. Freimuth](#),⁶ and [Marc S. Williams](#)⁴

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Genomic Resources Search

All Resources Clinician Laboratory
Research Patient News

Context Search Enabled: Type in a gene, condition or medication

Initial Release: Additional enhancements to contextual search coming soon! Click for details!

ClinGen EHR Working Group Objectives (Marc Williams)

- Created an HL7-compliant search interface for ClinGen (Genomic Resources Search)
- Proposed guidance for genomic resources on achieving HL7 Infobutton standard accessibility and compliance

Genomic Resources Search

<https://www.clinicalgenome.org/tools/web-resources/>



DocUBuild: A Collaborative System to Enhance Dissemination and Discovery of Genomic Clinical Content

S76: Tools for Genomics and Precision Medicine

Luke Rasmussen

Northwestern University

Casey Overby Taylor

Johns Hopkins University

Twitter:
#AMIA2017
#S76

DocUBuild

<https://docubuild.fsm.northwestern.edu/>

- Effort of the Infobutton Subgroup in eMERGE (Luke Rasmussen)



CDS_KB
CLINICAL DECISION SUPPORT KNOWLEDGEBASE

HOME CONTRIBUTORS LIBRARY LEARNING COMMUNITY CONTACT ABOUT

CDS KnowledgeBase is the engine that drives precision medicine.


who are we?

Clinical decision support (CDS) forms a significant part of the field of clinical knowledge management technologies through their capacity to support the clinical process and use of knowledge, from diagnosis and investigation through treatment and long-term care.

CDS KnowledgeBase Partners

emerge network **IGNITE** **Implementing GeNomics In practice**

**THE COMMUNITY:
IDEAS FROM ONE OF MANY**



- Effort of the Clinical Informatics Work Group (Josh Peterson)
- Focus on EHR integration, CDS, and technical implementation
- Library of artifacts (e.g., CDS presentation, workflow, algorithms & pseudocode)
- Archived webinars
- Current effort surveying sites about genomic medicine data pipeline

CDS_KB
<https://cdskb.org/>

gCDS and Precision Health

- Precision health requires *(Williams M. et al. Health Affairs 2018)*
 - A focus on outcomes
 - A central role of patients in defining outcomes (positive or negative)
 - Knowledge about the individual's state (implicitly includes genetic/genomic information)
- **Broadens data sources, knowledge sources, and application areas for gCDS**

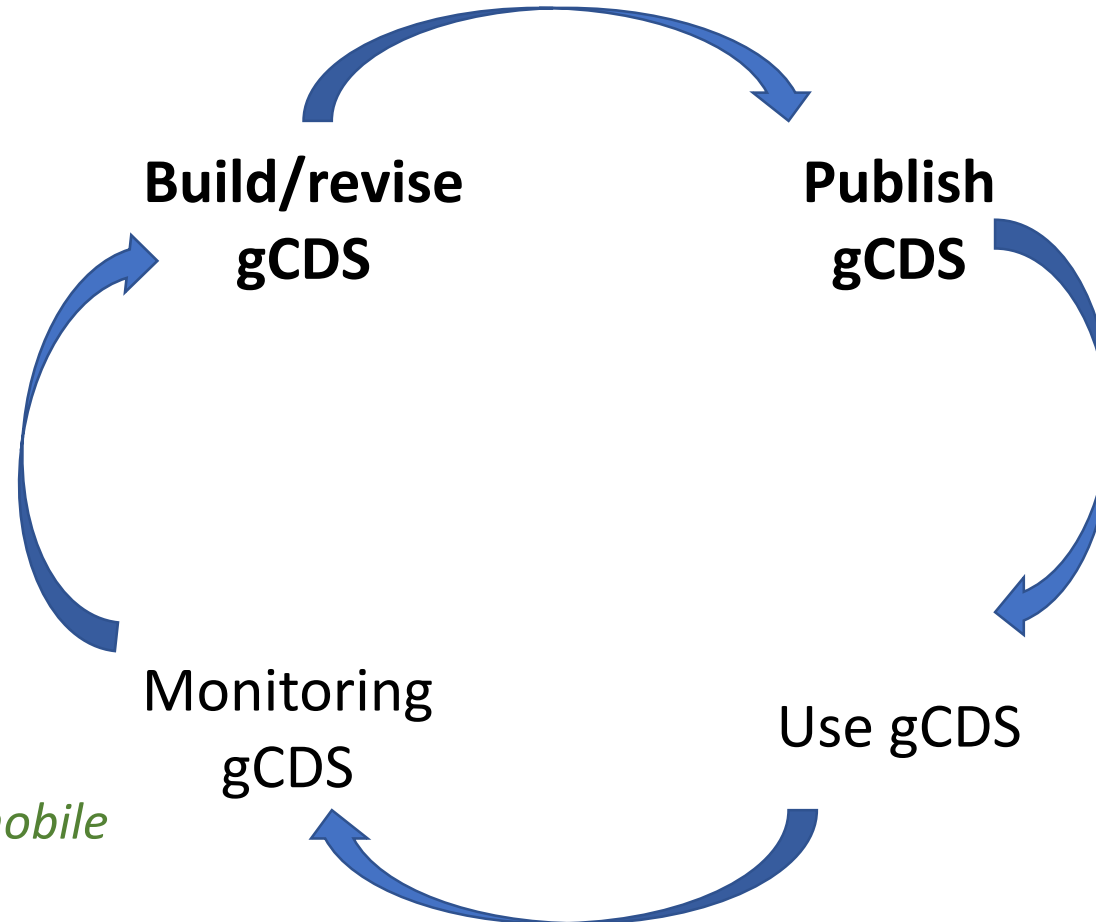
Managing shared knowledge for gCDS

Knowledge sources

- Clinical practice guidelines
- Resources aligned with healthcare org local policies
- *Patient preference-driven resources*

Data sources

- EHR
- Sequencing lab
- Patient (*Directly e.g. PHR, mobile devices*)
- *Patient-permission-granted access (e.g., geocoded-linked data)*



Computable gCDS

- Health care org local IT
- Clinical labs (structured interpretations)
- *Depends on delivery platform (e.g., cell phone)*

Application areas

- Treatment
- Diagnosis
- Disease prevention (acute)
- *Disease risk management*
- *Disease prevention (proactive)*

Summary of points

- We can learn from efforts in the broader CDS community to help address challenges for gCDS
- Implementation Science models can be complemented by existing frameworks to guide challenges and approaches to CDS implementation
- Consider further investment into planned and under development tools for managing shared knowledge for gCDS
- Design tools that can be extended to support Precision Health

Casey Overby Taylor, PhD

cot@jhu.edu

@coverbytaylor