Knowledge Management for Genomic Clinical Decision Support

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

Where we are now

The Promise of Personalized Medicine
- Research Discoveries
- Technology Developments

"Bench" 17+ years translating research into medical practice (Traditional Path)

Barriers
- Limited genetic proficiency of clinicians
- Limited availability of genetics experts
- Growth of genetic knowledge base

Where we want to be

The Realization of Personalized Medicine
- Safer Healthcare
- Improved Outcomes
- Reduced Costs

"Bedside"

(Welch & Kawamoto et al. JAMIA, 2012 Figure 1 Retrieved from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3638177/)
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

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

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Managing shared knowledge for gCDS

• Knowledge management solutions often are not accepted without customization

• Reliance on expert communities

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 ↓
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

(Overby CL et al. Genet Med 2013)
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

(Overby CL et al. Genet Med 2013)
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...

(Sittig and Singh Qual Saf Health Care, 2010 Figure 1 Retrieved from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3120130/)
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?

(Overby CL et al. Genet Med 2013 Figure 1 retrieved from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3858176/)
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

(Overby CL et al. Genet Med 2013 Figure 1 retrieved from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3858176/)
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

(Overby CL et al. Genet Med 2013 Figure 1 retrieved from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3858176/)
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)
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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)
The genomic CDS sandbox: An assessment among domain experts


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

S76: Tools for Genomics and Precision Medicine

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Twitter:
#AMIA2017
#S76

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

DocUBuild
https://docubuild.fsm.northwestern.edu/
• 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

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