Increasing the reach of clinical genomics research and genomics-informed care

Casey Lynnette Overby, PhD
Assistant Professor
Divisions of General Internal Medicine and Health Sciences Informatics
Malone Center for Engineering and Healthcare
About me

• I studied bioinformatics, biomedical informatics and public health genetics

• My responsibilities include:
  • Conducting research on clinical and shared decision making in precision medicine
  • Conducting research on approaches to interpret genome information
  • Mentoring students
Personalized medicine is tailoring of medical treatment to individual characteristics

- Commonly used in the context of Genomic Medicine
- P4 medicine™ (Hood L., Nat Biotechnol., 2011)
  - Preventative
  - Predictive
  - Participatory
  - Personalized

10 Leading causes of mortality
(CDC, Human United States, 2015 Table 19; *Also WHO 2000 to 2012)

1. Heart disease*
2. Cancer*
3. Chronic lower respiratory diseases*
4. Accidents (unintentional injuries)*
5. Stroke (cerebrovascular diseases)*
6. Alzheimer’s disease
7. Diabetes*
8. Influenza and pneumonia
9. Nephritis, nephrotic syndrome and nephrosis (kidney disease)
10. Intentional self-harm (suicide)
New “omics” technologies, sensors, and social networks platforms provide access to new forms of population health data.

(Weber, Mandl, Kohane. JAMA. 2014)
Research and practice co-exist to enable ongoing learning and evidence development.

(Chambers, Feero, Khoury. JAMA. 2016)
Improving the delivery of big data for precision medicine

- Need to decipher the meaning of data collected from various sources
- Integration of genetic data and electronic health record data is not trivial
- Need to reproduce...
  - Analyses performed by bioinformatics and laboratory professionals
  - Interpretations made by healthcare providers
- Need to know the relationships between the output of computations to prognosis and response to therapy
Goals

- Understand challenges to delivering genetic services

- Implementation science examples to understand context
  - Health IT to improve genetic laboratory reporting processes
  - Diverse information needs when delivering genetics services
Health IT challenges to lab reporting processes

- Challenge #1: Healthcare environments are complex, busy and involve many stakeholders

- Challenge #2: Dependence on vendor-specified CDS capabilities

- Challenges #3: Competing priorities of health IT professionals

Information needs are important for designing usable technologies

- Needs assessments are commonly conducted to identify user needs in software design

Table 1
Comparison of system development life cycle stages and nine generic study types

<table>
<thead>
<tr>
<th>Stead SDLC stage</th>
<th>Friedman and Wyatt study type</th>
</tr>
</thead>
<tbody>
<tr>
<td>A. Specification</td>
<td>1. Needs assessment</td>
</tr>
<tr>
<td>B. Component development</td>
<td>2. Design validation</td>
</tr>
<tr>
<td>C. Combination of components into a system</td>
<td>3. Structure validation</td>
</tr>
<tr>
<td>D. Integration of system into environment</td>
<td>4. Usability test</td>
</tr>
<tr>
<td></td>
<td>5. Laboratory function study</td>
</tr>
<tr>
<td></td>
<td>7. Laboratory user effect study</td>
</tr>
<tr>
<td>E. Routine use</td>
<td>6. Field function study</td>
</tr>
<tr>
<td></td>
<td>8. Field user effect study</td>
</tr>
<tr>
<td></td>
<td>9. Problem impact study</td>
</tr>
</tbody>
</table>

SDLC, system development life cycle.
*(Yen and Bakken. Review of health information technology usability study methodologies. JAMIA 2012)*
Precision Medicine Use Case: PAPI @ University of Maryland

- The Pharmacogenomics of Anti-Platelet Intervention Study
  - **Stakeholders:** Interventional Cardiologists, Cardiac NPs, Lab professionals (TGL and Path Lab), research coordinators
  - **Artifacts:** CPIC guidelines (CYP2C19 genotype test results -> actionable antiplatelet prescribing)


- Focus groups (cardiology fellows, nurse practitioners, clinical pharmacists, laboratory professionals, study coordinators)
  - **Genetic testing process:** documenting, notifying viewing
  - **User experience:** usability, missing functionalities, problems and proposed solutions, desired features
Several areas where the delivery of genetic findings to clinicians could be improved

(Cutting*, Overby*, et al. AMIA 2015)
Proposed areas where technology can improve processes

(Cutting*, Overby*, et al. AMIA 2015)
Summary

- Uncovered many areas for improving existing genetic testing processes

- Proposed recommendations for solving problems
  - Implementing CDS
  - LIMS system capable of communicating with the EHR
Genetic testing process may generalize to multi-gene testing

1. **Healthcare Providers**
   - Order WGS

2. **Ordering clinician**
   - Draw blood
   - Notified of “actionable” and “reportable” result

3. **EHR**
   - Display pop-up notifying physician of candidacy for CYP2C19 testing
   - Test report for “reportable” findings available to view

4. **Analytical laboratory director**
   - WGS assay
   - Verify “reportable” findings

5. **Translational Genomics Laboratory technician**
   - Prepare preliminary WGS report

6. **LIMS**
   - Embed oversight committee-approved recommendations

- If actionable results:
  - A Test report for “reportable” findings available to view
  - Actionable results?
  - Yes
    - B Notified of “actionable” and “reportable” result
    - Ongoing screening
  - No
    - C Test report for “reportable” findings available to view
    - Guidance for PCP
Goals

- Understand challenges to delivering genetic services

- Implementation science examples to understand context
  - Health IT to improve genetic laboratory reporting processes
  - Diverse information needs when delivering genetics services
Genetic counselors can help design tools to support genetic communication

- Primary care physicians generally have a favorable opinions of using genetic data in their practice, but they do not feel adequately informed (Klitzman et al. Journal of genetic counseling 2013, Marzullo et al. Preventive medicine 2013, Salm et al. J of genetic counseling 2014, )

- Genetic counselors are formally trained to deliver complex genetic information

- Brief overview of approach
  - Eight genetic counselors from three institutions were interviewed,
  - Determined major themes associated with genetic counselor information needs
  - Confirmed themes for accuracy using member checking
Multilevel Context Influencing Implementation success: Conceptual model for cancer genetic counselor information needs

**Information Need**
Clinical patient characteristics
- Family history
- Personal history (e.g., of cancer)
- Personal risk factors

**Information Need**
Social & cognitive patient characteristics
- Perceptions of risk
- Concerns
- Personal values (e.g., sexuality, body image, personal life)
- Life stage

**Information Need**
Patient goals & motivations
**Motivations**
- Personal experiences (e.g., with cancer, life changing events)

**Goals**
- Make treatment decisions
- Take preventive measures
- Understand personal risk
- Understand risk of family members

**Activity**
Risk assessment

**Activity**
Tailoring communication

**Activity**
Risk communication

Opportunities for tailoring technologies

- Pre-visit planning to improve patient-provider communication

- Variation in how patient perceive risk
  - Deliver just-in-time educational information during “teachable moments” that describes genetic risk in different ways

- Information needs may generalize to other areas
  - Perspectives of others performing risk assessment (e.g., cardiac or neurological diseases)
  - Other tests (e.g., panel gene testing)
Data to monitor a system in use

• Quantitative data
  • Completion rate (error rate)
  • Time on task

• Qualitative data
  • User satisfaction
Exciting times for research and practice to co-exist to enable ongoing learning and new evidence to support healthcare decisions

- Precision Medicine Initiative
- Understanding of genetic test processes informing the design of CDS approaches
- Understanding variability in stakeholder needs informing support for patient-provider communications and shared decision-making
- Open Source, standards-based, Apps to overcome challenges
Acknowledgments

- Funding
  - AHRQ R21HS023390
  - Program for Personalized & Genomic Medicine @University of Maryland School of Medicine
  - Columbia University Training in Biomedical Informatics (NIH NLM T15 LM007079)

