GMXIII: Developing a Clinical Genomic Informatics Research Agenda

Survey Results
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On behalf of: Pamela Williams, Tejinder Rakhra-Burris, Ken Wiley
Objectives

• Present and discuss survey results
• Compare results of current desiderata questions to GMVII responses
• Identify themes from written comments
• Set the stage for rest of the meeting
Statistics

• 33 respondents (83 invited to participate)
• Response rate 39.8%
• 100% completed the survey
• Extensive written comments
Results
Questions regarding genomic-based clinical informatics tools and resources
<table>
<thead>
<tr>
<th>#</th>
<th>Field</th>
<th>Minimum</th>
<th>Maximum</th>
<th>Mean</th>
<th>Std Deviation</th>
<th>Variance</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>#1: The business model for developing and implementing genomic based clinical informatics tools and resources encourages open source development</td>
<td>1.00</td>
<td>5.00</td>
<td>3.24</td>
<td>1.13</td>
<td>1.29</td>
<td>29</td>
</tr>
<tr>
<td>2</td>
<td>#2: Methods for evaluating the clinical utility of a genomic based clinical informatics tools and resources are clearly defined for the research community to use in their research and development plans.</td>
<td>1.00</td>
<td>5.00</td>
<td>3.55</td>
<td>0.77</td>
<td>0.94</td>
<td>29</td>
</tr>
<tr>
<td>3</td>
<td>#3: Advancements in storing genomic and genomic based information are adequate to meet the clinical genomic community needs.</td>
<td>1.00</td>
<td>5.00</td>
<td>3.48</td>
<td>0.97</td>
<td>0.94</td>
<td>29</td>
</tr>
<tr>
<td>4</td>
<td>#4: Methods related to ontology management and knowledge representation for genomic based clinical interpretation are adequately addressed by the research community.</td>
<td>2.00</td>
<td>5.00</td>
<td>3.62</td>
<td>0.84</td>
<td>0.70</td>
<td>26</td>
</tr>
<tr>
<td>5</td>
<td>#5: Methods for Integrating analytical interpretations derived by computational models of genomic data into clinical settings are well established.</td>
<td>3.00</td>
<td>5.00</td>
<td>4.11</td>
<td>0.63</td>
<td>0.40</td>
<td>27</td>
</tr>
<tr>
<td>6</td>
<td>#6: The genomic medicine community will benefit from having a revised technical desiderata.</td>
<td>1.00</td>
<td>4.00</td>
<td>2.04</td>
<td>0.78</td>
<td>0.61</td>
<td>28</td>
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<tr>
<td>7</td>
<td>#7: Efforts to modify clinical workflows to reduce instances of alert fatigue are actively being developed by the genomic medicine community.</td>
<td>2.00</td>
<td>5.00</td>
<td>3.61</td>
<td>0.98</td>
<td>0.95</td>
<td>28</td>
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<tr>
<td>8</td>
<td>#8: Does the Centers for Medicare &amp; Medicaid Services Promoting Interoperability Programs support the integration of genomic and genomic related information into the Electronic Health Record.</td>
<td>2.00</td>
<td>5.00</td>
<td>3.41</td>
<td>0.83</td>
<td>0.70</td>
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### Research Priority?

<table>
<thead>
<tr>
<th>#</th>
<th>Question</th>
<th>Strongly Agree</th>
<th>Agree</th>
<th>Neither Agree / Disagree</th>
<th>Disagree</th>
<th>Strongly Disagree</th>
<th>Total</th>
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</thead>
<tbody>
<tr>
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<td>#5: Methods for integrating analytical interpretations derived by computational models of genomic data into clinical settings are well established.</td>
<td>0.00%</td>
<td>0.00%</td>
<td>14.81%</td>
<td>59.26%</td>
<td>25.93%</td>
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Comparison to GM VII (October 2014)

- GMVII focus was on Genomic Clinical Decision Support
- Methodology different
  - Queried on two different scales
    - Importance of a given element
    - Gap between current state and ideal future state
  - Also asked to prioritize elements
- For GMXIII asked to agree or disagree with desiderata, which is mostly similar to importance of a given element
Mean Element Importance-GMVII

1. Separation of mol observation from clin interp
2. Lossless compression
3. Linkage observations to lab methods
4. Actionable subsets
5. Human /Machine readable
6. Changes in understanding
7. Discovery science and patient care
8. CDS over multiple genes
9. CDS Knowledge separate
10. EHR generalizability
11. Support Gene variants
12. Standards: CDS and genomics
13. Deploy shared CDS KB
14. Access and transmit minimum info for CDS
## Comparison of priorities

### Ranked high both meetings

<table>
<thead>
<tr>
<th>Desiderata ranked high to low</th>
<th>GMVII</th>
<th>GMXIII</th>
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### Changed between meetings (GMVII to GMXIII)

- **#12** Leverage current and developing CDS and genomics standards (Low to High)
- **#3** Maintain linkage of molecular observations to the laboratory methods used to generate them (Low to High)
- **#1** Maintain separation of primary molecular observations from the clinical interpretations of those data (Low to High)
- **#13** Support a CDS knowledge base deployed at and developed by multiple independent organizations (High to Low)
- **#5** Simultaneously support human-viewable formats and machine-readable formats in order to facilitate implementation of decision support rules (High to Low)

#8 CDS knowledge must have the potential to incorporate multiple genes and clinical information

#10 CDS knowledge must have the capacity to support multiple EHR platforms with various data representations with minimal modification

#14 Leverage current and developing CDS and genomics standards (Low to High)
Special Relevance #7  Support both individual clinical care and discovery science. (High to Low) Ranked #2 GMVII and #8 for GMXIII (Session 5)
Additional themes from free text

• Importance of assessing stakeholder preference and workflow
• Sustainability of resources
• Lack of methods for evaluation of innovation and implementation
• Impact of the consent and regulatory framework
Implications for the research agenda

• Research in genomics and informatics is a “target-rich” environment
• Several priorities are persistent over the last 5 years
• Research should include attention to:
  o Stakeholder engagement and workflow evaluation
  o Development and use of rigorous evaluation methods
  o Consideration of policy and regulatory environment
  o Sustainability
The Meeting

• Survey results including comments included in the meeting materials
  o Contact Ken or me if you have comments on our interpretation of the results

• Each speaker has received narrative comments from the survey relevant to their topic and has been asked to use those to inform their content

• Keep the overarching implications in mind during discussion
Clarifying Questions