# GMXIII: Developing a Clinical Genomic Informatics Research Agenda

Survey Results

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# Objectives

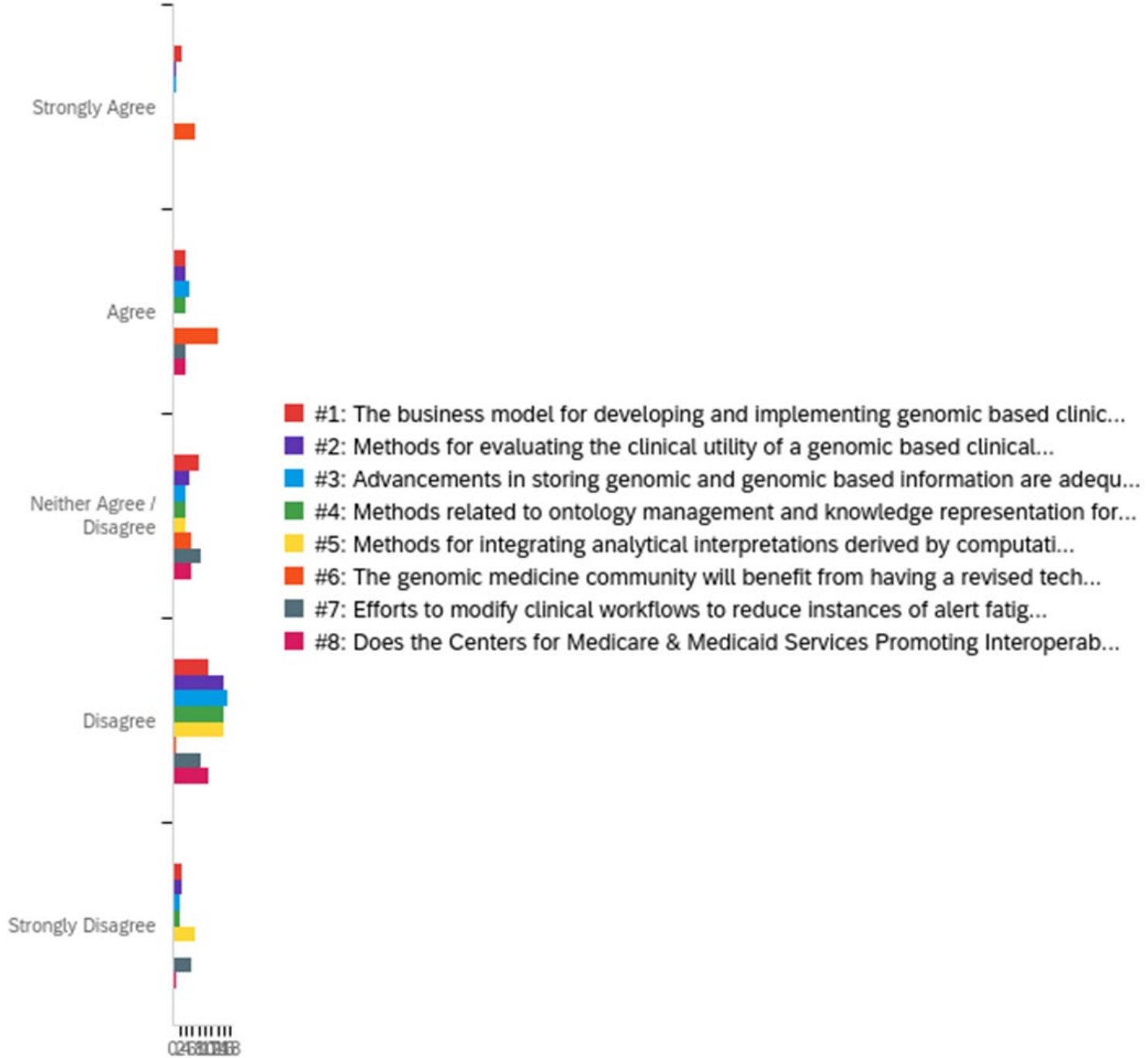
- Present and discuss survey results
- Compare results of current desiderata questions to GMVII responses
- Identify themes from written comments
- Set the stage for rest of the meeting

# 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



| # | Field | Minimum | Maximum | Mean | Std Deviation | Variance | Count |
|---|---|---|---|---|---|---|---|
| 1 | #1: The business model for developing and implementing genomic based clinical informatics tools and resources encourages open source development | 1.00 | 5.00 | 3.24 | 1.13 | 1.29 | 29 |
| 2 | #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. | 1.00 | 5.00 | 3.55 | 0.97 | 0.94 | 29 |
| 3 | #3: Advancements in storing genomic and genomic based information are adequate to meet the clinical genomic community needs. | 1.00 | 5.00 | 3.48 | 0.97 | 0.94 | 29 |
| 4 | #4: Methods related to ontology management and knowledge representation for genomic based clinical interpretation are adequately addressed by the research community. | 2.00 | 5.00 | 3.62 | 0.84 | 0.70 | 26 |
| 5 | #5: Methods for integrating analytical interpretations derived by computational models of genomic data into clinical settings are well established. | 3.00 | 5.00 | 4.11 | 0.63 | 0.40 | 27 |
| 6 | #6: The genomic medicine community will benefit from having a revised technical desiderata. | 1.00 | 4.00 | 2.04 | 0.78 | 0.61 | 28 |
| 7 | #7: Efforts to modify clinical workflows to reduce instances of alert fatigue are actively being developed by the genomic medicine community. | 2.00 | 5.00 | 3.61 | 0.98 | 0.95 | 28 |
| 8 | #8: Does the Centers for Medicare & Medicaid Services Promoting Interoperability Programs support the integration of genomic and genomic related information into the Electronic Health Record. | 2.00 | 5.00 | 3.41 | 0.83 | 0.70 | 22 |

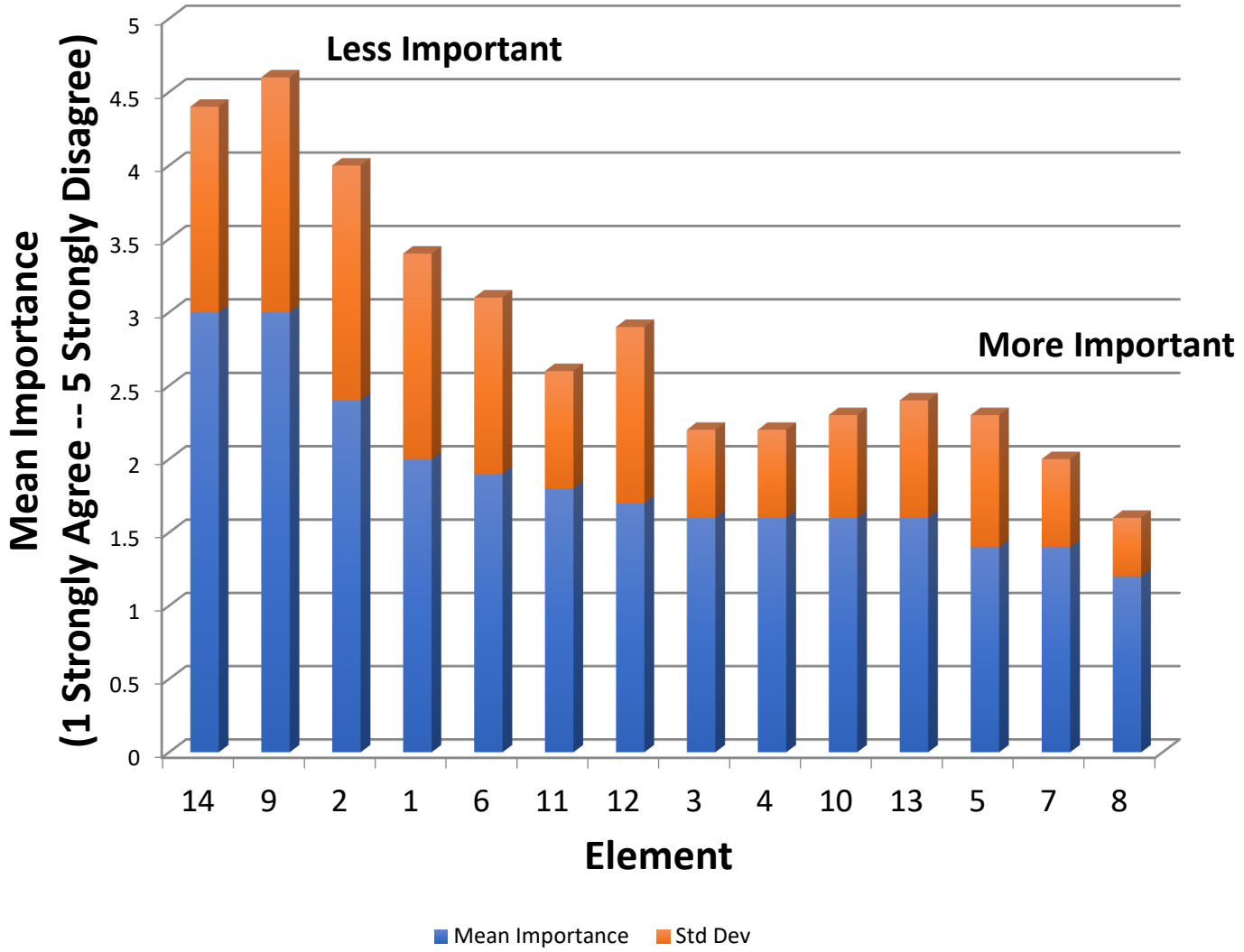
# Research Priority?

| # | Question | Strongly Agree | | Agree | | Neither Agree / Disagree | | Disagree | | Strongly Disagree | | Total |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 5 | #5: Methods for integrating analytical interpretations derived by computational models of genomic data into clinical settings are well established. | 0.00% | 0 | 0.00% | 0 | 14.81% | 4 | 59.26% | 16 | 25.93% | 7 | 27 |

# 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**

#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

| Desiderata ranked high to low | |
|---|---|
| GMVII | GMXIII |
| 8 | 8 |
| 7 | 3 |
| 5 | 1 |
| 13 | 12 |
| 10 | 10 |
| 4 | 2 |
| 3 | 11 |
| 12 | 7 |
| 11 | 4 |
| 6 | 5 |
| 1 | 9 |
| 2 | 13 |
| 9 | 6 |
| 14 | 14 |

**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)

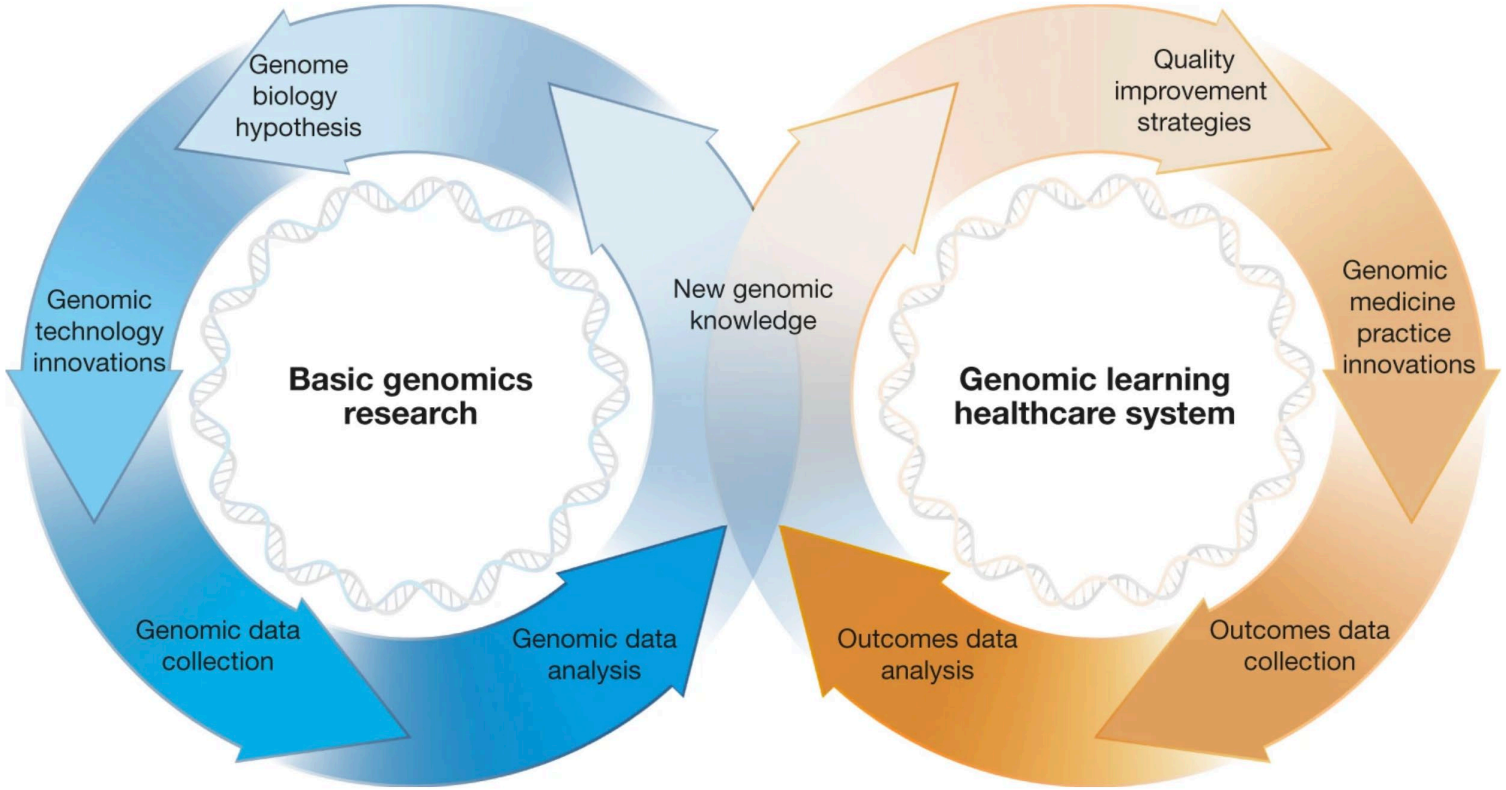
#13 Support a CDS knowledge base deployed at and developed by multiple independent organizations (High to Low)

#5 Simultaneously support human-viewable formats and machine-readable formats in order to facilitate implementation of decision support rules (High to Low)

# **Special Relevance** #7 Support both individual clinical care and discovery science. (High to Low) Ranked #2 GMVII and #8 for GMXIII (Session 5)

**Fig. 3: Virtuous cycles in human genomics research and clinical care.**

From: Strategic vision for improving human health at The Forefront of Genomics



# 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:
  - Stakeholder engagement and workflow evaluation
  - Development and use of rigorous evaluation methods
  - Consideration of policy and regulatory environment
  - Sustainability

# The Meeting

- Survey results including comments included in the meeting materials
  - 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