Rapporteur: Jacqueline Odgis
Program Analyst, NHGRI

Genomic Medicine VII: Genomic Clinical Decision Support
October 2-3, 2014, Bethesda, MD
Executive Summary

The Seventh Genomic Medicine Meeting on genomic clinical decision support (GCDS) convened key thought leaders in genomic implementation and application of clinical decision support to: 1) compare current and ideal states of GCDS to define gaps and strategies to close them; 2) identify and engage health information technology (IT) initiatives that would support recommended strategies; and 3) define a prioritized GCDS implementation research agenda.

Ideal State GCDS

The ideal GCDS system should be readily updateable, have content that can be repurposed for different types of users, be sensitive to different users' health literacy and clinical needs, and users (including patients) as new knowledge on clinical importance of variants emerges. Clinically actionable components of genomic sequence data (if not all sequence data) should remain separate from clinical interpretations and be transferrable across EHR systems as patients move through healthcare institutions. To evaluate the impact of implementation, GCDS should monitor process and outcome measures, collect user decisions and actions, and allow developers to receive input from end users.

Challenges in the current state of GCDS

Numerous cited shortcomings of current GCDS included lack of definition of standards for CDS rules and outcomes metrics, lack of bandwidth for high-dimensional genomic sequence data, limited expertise within institutions' management teams for interpreting genomic information, lack of evidence demonstrating the economic and business value of GCDS (especially at scale), alert fatigue, lack of shared infrastructure for transporting data across vendor systems, and rapidly changing knowledgebase and sociological landscape of clinical genomics. A root cause of lack of scalable GCDS support is lack of actual business cases demonstrating the value of GCDS to vendors and healthcare systems.

Potential working groups and collaborative projects

Participants reviewed possible research directions to address these shortcomings and voted priorities for them. In order from most to least interest, these are:

- **GCDS Use Cases**: Collaborate with existing informatics working groups to define standard exportable methods, develop CDS rules and use cases for knowledge representation, and identify top priority genes to create standards for specific variant representations
  - Pharmacogenomics (PGx) Use Cases
  - WGS/WES Use Cases
  - Business/Return on Investment Use Cases
  - Post-Market Surveillance Use Cases

- **GCDS Sandbox**: Test use cases in an experimental computing environment ("sandbox"); evaluate the interoperability of these cases
- **Open CDS Knowledge Library**: Build a nationwide public library to centralize and share GCDS knowledge
- **End-to-End Project**: Aggregate best practices and outcomes data from implementers, accumulating sufficient numbers to assess current standards for data, knowledge, processes, and outcomes
- **Role of the Patient/Caregiver**: Explore patient facing-decision support methods and patient ownership of data

Collaborations and preliminary action items

The meeting co-chairs, Marc Williams and Blackford Middleton, agreed to continue communications with attendees. Proposed initial steps included:

- Producing a draft white paper to summarize the outputs of the meeting and perhaps provide recommendations to specific agencies
- Developing business/return on investment use cases on: 1) the research perspective in generating additional income streams, and 2) how integrating genetic/genomic information into GCDS can allow genomic scientists to mine data for research
- Engaging federal agencies such as FDA, CMS, and ONC to assist in creating these use cases
- Gauging interest and collecting use cases from decision support and informatics working groups within existing projects such as eMERGE, IGNITE, ClinVar, CPIC, NCI CBIIT
- Gathering insights from the ONC/AHRQ CDS initiative creating interoperable CDS systems for immunizations
- Aligning with standards developing organizations such as HL7 for synergy and concordance with existing standards through Meaningful Use (i.e., SNOMED, LINCS, etc.)
- Approaching EHR vendors to see how GCDS components can be transferred across systems
- Recruiting experts from projects such as the Vanderbilt PREDICT, IOM, and infobutton (VA OpenInfobutton, projects to outline an agenda for an end-to-end project
- Working with NLM to define terminology standards and establish common elements
- Looking for opportunities to present at professional society meetings to clarify and increase awareness of the role of CDS in genetics
- Engaging PCORNet for potential synergies between their patient-centered activities and a patient-facing CDS project
- Reconvene the Genomic Medicine VII group; J.D. Nolan offered to host at Cerner