

MINUTES – FINAL
Health Information Technology Standards
Advisory Committee (HITSAC)
Genomics Working Group
Thursday, July 10, 2014

Commonwealth Enterprise Solutions Center
11751 Meadowville Lane
Chester, VA 23836
Multipurpose Room 1221

ATTENDANCE:

Members Present:

Debbie Condrey, VDH (Elected Chairperson)
Sandy McCleaf, ConnectVirginia HIE
(Elected Vice-Chairperson)
Dr. Denise Toney, DCLS
Dr. Ira Lubin, CDC
Mollie Ullman-Cullere, HL7
Aaron Black, Inova/ITMI

Others Present:

Dr. Marshall Ruffin, HITSAC Chairman
Deputy Secretary Anthony Fung, GOV
Dr. Joseph Grubbs, VITA
Nicole Helmantoler, VITA
Dr. David Li, Health Diagnostics Lab, Inc.

Members Absent:

Dr. Lawrence Silverman, UVA
Dr. Andrea Ferreira-Gonzalez, VCU

CALL TO ORDER

HITSAC Chairman Ruffin called the meeting to order at 11:05 a.m. in the VITA Multipurpose Room 1221 at the Commonwealth Enterprise Solutions Center in Chester, VA. HITSAC Chairman Ruffin welcomed the appointed members of the HITSAC Genomics Working Group, staff and attendees.

Dr. Grubbs, Research Coordinator and Staff Administrator for the Working Group, advised that the Working Group is considered a “public body” under the *Code of Virginia* and, therefore, the meetings would need to follow Virginia Freedom of Information Act and other Commonwealth regulations for public meetings. As a public body appointed by HITSAC, HITSAC Chairman Ruffin chaired the meeting until the Working Group could elect its own Chairperson and Vice-Chairperson.

Note: The HITSAC Genomics Working Group meeting agenda packet including all of the discussion materials may be accessed on the VITA Web site at:

<http://www.vita.virginia.gov/ITAC/default.aspx?id=6442472422>

OLD BUSINESS

There were no items under Old Business for consideration on the meeting agenda.

NEW BUSINESS

Welcome and Introductions

Following HITSAC Chairman Ruffin's welcome, Working Group members introduced themselves and provided their affiliations.

Election of Chair and Vice-Chair

Dr. Grubbs advised the Working Group that the next item on the agenda was for the election of a Chair and Vice-Chair. However, at the recommendation of HITSAC Chairman Ruffin and subsequent approval of the Working Group, this agenda item was moved until the end of New Business.

Review of Genomics Working Group Charter

HITSAC Chairman Ruffin called the next item on the agenda, review of the Genomics Working Group Charter. Dr. Grubbs walked the members through the draft Charter then opened the floor for comments, edits and recommendations.

Mollie Ullman-Cullere recommended that the Working Group engage with Health Level 7 (HL7) and the HL7 Clinical Genomics Working Group, for which Ms. Ullman-Cullere is the Co-Chair. Ms. Ullman-Cullere offered to serve as a liaison between the two groups.

Ms. Condrey said that the Charter should state clearly that the Working Group will be providing its findings and recommendations on potential pilots to HITSAC. HITSAC Chairman Ruffin agreed, adding that the Working Group could come back to HITSAC with multiple pilot recommendations.

Dr. Lubin asked for the specific reference to the U.S. Centers for Disease Control and Prevention (CDC) in the Introduction section to be removed, and under Responsibility #3 stated that the Charter should establish that the Working Group will be engaging with multiple stakeholders in the health IT community, including CDC, HL7 and others, as needed. Dr. Lubin recommended that the Charter include a "Final Products" section to spell out more detail on Working Group outputs, including targeted use cases, potential pilots, recommendations on health IT standards and other activities.

Ms. Ullman-Cullere raised the question of whether the Working Group would be targeting its efforts on a particular genomics/genetics testing platform or technology, recommending instead that the Working Group remain focused on full interoperability. HITSAC Chairman Ruffin agreed to taking the broader, interoperability focused approach. Dr. Lubin also agreed with the broader focus, stating that this approach enables a more thorough consideration of gaps and solutions.

Dr. Grubbs noted several additional wording changes for the Charter and said he would make the recommended edits, distribute the revised draft to Working Group members for additional comment, then prepare the final draft for consideration by HITSAC during its August 2014 meeting.

Review of Candidate Use Cases

Mollie Ullman-Cullere in her final comments relating to the Working Group Charter raised one of the candidate use cases up for consideration by the Working Group. Dr. Grubbs acknowledged that the specific use case raised by Ms. Ullman-Cullere had been added to the published agenda packet for consideration. HITSAC Chairman Ruffin then transitioned the Working Group to this agenda item.

Due to the technical nature of the Working Group's discussion on candidate use cases, the public is advised to access the audio recording of the HITSAC Genomics Working Group meeting on the VITA Website at:

http://vita2.virginia.gov/itac/140710_HITSAC_Genomics_Transcript.mp3

The discussion of candidate use cases begins at the 36:00 timestamp.

The Working Group agreed that the research approach should be to start with an initial set of use cases then produce a roadmap of future use cases that could be considered. The Working Group identified the following as the initial set of candidate use cases:

- Use Case #1 – Interoperability in Reporting Formats for Genetic Test Results (HL7): Submitted by Ms. Ullman-Cullere and referenced in the *HL7 v. 3 Domain Analysis Model: Clinical Sequencing, Release 1 – Draft 1st Informative Ballot*. Targets disparities in the reporting of results from genetic tests based on different laboratory testing platforms and technologies and opportunities for harmonization, governance and health IT standards to promote interoperability between the diverse reporting formats.
- Use Case #2 – Interoperability in Genomics for Public Health Registries: Submitted by Ms. Ullman-Cullere and referenced in the *HL7 v. 3 Domain Analysis Model: Clinical Sequencing, Release 1 – Draft 1st Informative Ballot*. Focuses on potential for standards to enforce data structures and nomenclature in public health registries to support the exchange of genomic information across registries and through health information exchanges.
- Use Case #3 – Inova Translational Medicine Institute (ITMI) Health Informatics and Standards: Submitted by Aaron Black, Inova Health System/ITMI, and currently under consideration by ITMI. Explores health informatics and standards requirements to enable development of a pipeline of results from genetic tests for pharmacogenomics, cancer hotspot panels and related tests to support interoperability and structuring of these data within Electronic Health Record (EHR) Systems.
- Use Case #4 – Inova Translational Medicine Institute (ITMI) Premature Birth Research: Submitted by Aaron Black, Inova Health System/ITMI, and currently under consideration by ITMI. Builds upon ITMI's growing repository of whole genome sequences for families (mother, father, infant), collected as part of two ITMI research studies. Focuses on how this information may be structured within EHR systems and used to support clinical decision-making to prevent premature births.
- Use Case #5 – Interoperability in Genomics for Microbiology: Submitted by Dr. Denise Toney, Director, Division of Consolidated Laboratory Services. Examines bioinformatics and standards requirements for genomic information gathered from microorganisms extracted from human subjects. Potential to support interoperability between DCLS and the Virginia Department of Health for public health and epidemiological outcomes.

Direction on Literature Review and Next Steps

HITSAC Chairman Ruffin moved the Working Group into the agenda item on the literature review and related next steps. The Working Group directed Dr. Grubbs, as Research Coordinator, to frame the literature review around the candidate use cases and document the use cases in a standards-based format. The Working Group also encouraged Dr. Grubbs, and the group as a whole, to keep the list of use cases dynamic and leading toward a roadmap of potential use cases.

Dr. Grubbs said he would reach out to the respective Working Group members who raised the targeted use cases to gather more detailed information. Dr. Grubbs also said that he would begin conducting interviews with subject matter experts to further frame the use cases and potential pilots. Dr. Grubbs concluded stating he would document the research agenda into a work plan and project schedule.

Dr. Lubin asked if the Working Group members would be open to co-publishing and/or co-presenting with the CDC on the targeted use cases and research approach. The Working Group agreed, and Dr. Grubbs said he would begin preparing a speakers' bureau type report that listed areas of specialization for each of the Working Group members.

Election of Chair and Vice-Chair

HITSAC Chairman Ruffin then returned to the agenda item for election of Chair and Vice-Chair. Dr. Grubbs informed the Working Group that duties of the Chair involved leading the public meetings of the Working Group pursuant to Robert's Rules of Order, directing the Research Coordinator on a weekly basis on implementation of the research methodology and reporting to HITSAC on the progress made by the Working Group. Dr. Grubbs said the Vice-Chair responsibilities included assisting the Chair on direction of the research program and, in the absence of the Chair, conducting the public meetings of the Working Group. After discussion of candidates, Debbie Condrey volunteered and, seeing no objections or other candidates, was elected unanimously. Ms. Condrey then nominated Sandy McCleaf as Vice-Chair, and seeing no objections or other candidates, Ms. McCleaf was elected unanimously.

PUBLIC COMMENT

The Working Group Chairperson, Ms. Condrey, then called for public comment. David Li, from Health Diagnostic Laboratory, Inc., addressed the Working Group. Mr. Li acknowledged the importance of the Working Group and the research approach being taken to help inform health IT standards for clinical genomics. Mr. Li thanked the Working Group and offered to support its efforts.

ADJOURNMENT

The Working Group Chairperson, Ms. Condrey, then called for any other comment. Seeing none, Chairperson Condrey adjourned the meeting with consent from Working Group members.