

exported to pdf Aug 29, 2016

HL7 Clinical Genomics Weekly Call - Aug 23, 2016

Minutes will be recorded in this Google Doc:

<http://bit.ly/2aqVmqz>

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Kevin Power - Cerner - kpower@cerner.com
3. JD Nolen - Cerner - johndavid.nolen@cerner.com
4. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
5. Joel Schneider - NMDP - jschneid@nmdp.org
6. Siew Lam -Intermountain Healthcare - siew.lam@imail.org
7. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
8. Perry Mar - Partners HealthCare System - pmar@partners.org
9. Terry McDonnell - Syapse - terrym@syapse.com
10. Scott Bolte - Niss Consulting - Scott.Bolte@gmail.com
11. Grant Wood - Intermountain -
12. Joseph Kane - Epic - jkane@epic.com
13. Clem McDonald - NLM -
14. David Kreda - HMS Consultant - david.kreda@gmail.com (late - no audio :-)
15. Xiao Luo - USTC - l.xander.233@gmail.com
16. Yi Wang - USTC- panzer.wy@gmail.com
17. Tianlong Chen - USTC - wijp619@gmail.com

Discussion

- Brief reports
 - ClinGen/ClinVar
 - ClinGen focussed on renewal for grant - due in middle of Sep
 - GA4GH
 - Bob F - VMC group will be meeting in person in Boston next week; identifying common concepts; working to coming to terms with our terms :) Must bigger job than anyone anticipated; sequence ontology will be involved; more to share in early Sept. ; members of our group = Bob F., Larry B, Gil
 - Grant - next plenary meeting in Vancouver, October 17-18; Grant having discussions with ethical/security, looking to build a document of best practices and consent; quite a lot of work and interest bubbling up in whole consent space from many orgs (eg precision medicine, etc); many issues that need to be coordinated with different groups; Grant will try to coordinate these efforts in GA4GH; summarize different efforts in single doc, have groups work together.
 - Scott - is anyone looking at automated workflows?
 - Grant - not yet, we look forward to getting input from Scott
 - Scott - has a use case, no working model - *First degree relatives (FDR) and second degree relatives (SDR) of someone who has had colon*

polyps removed have a significantly increased chance of developing colorectal cancer. A consent form that allows for notification of healthcare providers for FDR/SDR could help cancer prevention programs. However, if the notification is manual it's unlikely to be successful. We should look at the new FHIR based workflow efforts to see if/how we can have automated notification.

- National Academies
 - JD - nothing new
- Other
 - none
- Baltimore Agenda - 2nd draft
 - [http://wiki.hl7.org/index.php?title=File:HL7_WGM_Sep2016 - Clinical Genomics Agenda.docx](http://wiki.hl7.org/index.php?title=File:HL7_WGM_Sep2016_-_Clinical_Genomics_Agenda.docx)
 - Need materials ahead of time to review
 - If shared via google doc, can be shared to comment but edit
 - should a SOP for WGMs
 - Family History - stakeholders that will attend WGM
 - Grant - yes
 - Scott – no
 - Jon – no
 - Bob W – maybe
 - Brett Johnson - yes
 - Clem – yes, but leaving at 2pm so prefer to move this to Tue or earlier in the day
 - Amon – yes
 - Bob M – yes but more listening than contributing
 - Bob F – yes
- Docs to be reviewed to prep for Baltimore
 - Mission & Charter
 - Must be < 2 yrs old
 - http://www.hl7.org/documentcenter/public/wg/clingenomics/Mission-Charter_ClinicalGenomics_approved%20091714%20final.docx
 - Due to be renewed this Sep 2016
 - Decision Making Process
 - must be < 3 yrs old
 - http://www.hl7.org/documentcenter/public/wg/clingenomics/HL7_WG_DMP_v3.0_CG_v2_09172014%20final.doc
 - Due to be renewed Sep 2017
 - SWOT
 - must be <3yrs old
 - http://www.hl7.org/documentcenter/public/wg/clingenomics/CG-SWOT_v2_091714%20final.docx
 - Due to be renewed Sep 2017
- Other?

Recorded Chat

- from Bob Milius to Everyone:

- <http://bit.ly/2aqVmqz>
- from David Kreda to Everyone:
 - Ah ...
- from David Kreda to Everyone:
 - It is the "Variant Modeling Collaboration" - so Modeling and Collaboration. So Bob F was correct!
- from David Kreda to Everyone:
 - And there is an ongoing issue about one "l" or two in "Model(ling" :-)
- from Bob F to Everyone:
 - Thanks, David. Maybe we should use brackets to denote the additional, optional "l". Model[l]ing? Or regex-like pipe for optionality: Mode(l|ll)ing? Perfectly clear. :-)
- from Joel Schneider to Everyone:
 - Does this mean we need a separate HL7 WG for genetics vs. genomics?
- from Perry Mar to Everyone:
 - We listed our definitions several years ago, which could be used for clarity about genetics, genomics, etc.