HL7 Clinical Genomics Weekly Call - Nov 29, 2016

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org (presiding co-chair)
2. JD Nolen - Cerner - johndavid.nolen@cerner.com
3. Scott Bolte - Trailhead Clinical Strategies - Scott.Bolte@gmail.com
4. Tianlong Chen - USTC - wiwjp619@gmail.com
5. Joseph Kane - Epic - jkane@epic.com
6. Perry Mar - Partners HealthCare System - pmar@partners.org
7. Kevin Power - Cerner - kpower@cerner.com
8. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
9. Xin Liu - BCH- xinliu215@gmail.com
10. Bret Heale - Intermountain Healthcare - bheale@gmail.com
11. Joel Schneider - NMDP - jschneid@nmdp.org
12. Larry Babb - GeneInsight - larry.babb@geneinsight.com
13. Jonathan Holt - SeqTechDx jholt@seqtechdx (half listening today, I abstain from all votes)
14. David Kreda - HMS - david.kreda@gmail.com
15. Xiao Luo - USTC - l.xander.233@gmail.com
16. Jeremy Warner - Vanderbilt - jeremy.warner@vanderbilt.edu
17. Chethan Makonhalli - NextGen - Chethan@nextgen.com
18. Yi Wang - USTC - panzer.wy@gmail.com
19. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu (joined 30 min late)
20. Terry McDonnell - Syapse - terrym@syapse.com (first 30 min only)
21. Clem McDonald - NLM
22. Eric Whitebay
23. Gil Alterovitz - BCH
24. Hans Buitendijk - co-chair HL7 O&O
25. Dave Hamill - HL7 PMO

Discussion

- **Minutes approval**
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20161122.pdf
    - motion to approve - Perry Mar; second - Kevin Power
    - discussion - none
    - abstains - Scott, Gil, Terry, Tianlong, Bret H, Xin Liu, Hans, Dave, Xiao Luo
    - nays - 0
    - yeas - 16
    - motion passes (approved)

- **Brief reports from external efforts (discussion only if needed)**
    - GA4GH -
        - (Larry Babb) VICC Variant Interpretation Cancer Consortium has great momentum/participation. They continue to work on a member agreement letter which states policies which all participants subscribe to. In addition, they continue to demo various implementations of Variant Interpretation tools primarily focused on types of evidence-based knowledge assertions related to cancer. This is analogous to the mendelian condition and PGx type knowledge being captured and promoted by ClinVar, PharmGKB and ClinGen amongst others.

    - National Academies
        - On last pilot call, Mission Health reported some initial success transmitting data (orders, results). Also Sandy, Brad Strock, and JD Nolen (representing DIGITizE) are getting engaged with CSER with respect to a Lynch Syndrome Use-Case for CDS. More information later.
    - Clingen/Clinvar
        - (Larry Babb) Making progress on revamping the datamodel.clinicalgenome.org website. It will include both the Allele model and the forthcoming Interpretation model. Version 1 of the Interpretation model is scoped to the ACMG 2015 guidelines. ClinGen and SEPIO are working together to harmonize approaches to evidence based interpretation data. We have started to work on the pilot implementation between ClinGen's new Variant Curation app (Stanford) submitting to ClinVar using this model.
        - (Larry Babb) ClinGen/Baylor Allele Registry is available for public use. I strongly recommend all take a look at http://reg.genome.network/site/cg-registry and the link to the API specification. This tool will be used by ClinGen applications for identifying variants going forward.
        - (Larry Babb) ClinGen/Stanford Gen and Variant Curation applications are now both production ready (Gene has been available for some time). See https://curation.clinicalgenome.org/ for more information. The var cur app uses the allele registry.
    - GA4GH Variant Modeling Collaborative (VMC)
        - (Larry Babb) Making progress on the Variant specification. Reece Hart has been doing a fantastic job as lead and driving the workload. The group meets weekly. Reece recently assembled and demoed a reference implementation of the current state of the model. The group is planning on meeting with ClinVar/ClinGen representatives Melissa Landrum/Heidi Rehm, respectively, this coming Monday, Dec 5th to discuss both the value proposition and risks related to representing alleles (most atomic structure defined by the group) with certain equivalencies versus precise, explicit unique representations which would require mappings and associations to make equivalent.
    - NHGRI
- **Deadlines**
    - 2016-12-04: Final content deadline
        - V2 Lite content was submitted to LRI "as is" in vote on Nov 15
            - *"Adopt all proposed dispositions, accept the materials as-is, recognizing that this effectively still a re-ballot of the materials in context now of the full LRI IG, thus accept ballot comments on all aspects of the CG material, rather than waiting a ballot round, and include all ballot comments not yet addressed into the January ballot reconciliation process."*
    - 2016-12-08: Last day to sign up for Ballot Consensus Group (aka Ballot Pool)
        - need to sign up if you want to submit a ballot comment on LRI/V2 Lite
    - 2016-12-09: Provisional ballot opening
    - 2016-12-16: Deadline to post CG WGM agenda on the WGM information page **(WG Health metric)**
- **Upcoming calls through next WGM in San Antonio**

- Dec 6 - Truly quick review (20 min) of new Clinical Sequencing DAM document by Gil or David with a google or doodle poll released opens after meeting to vote yes/no to publish and stays open until Dec 20 (per below) as the document is long!
  DAM document will be delivered in two formats: markup in view-only Google doc and clean version in PDF, the actual candidate.
- Dec 13 - Amnon presenting his FHIR proposal
- Dec 20 - Deadline for DAM poll results
- Dec 27 - (cancel?)
- Jan 3 -
- Jan 10 -

- **DAM**
  - https://docs.google.com/document/d/1BrpqPbgUCvtrPvRdf-oikK0pHiKUxo5QdZahxX9Z7dU/edit
  - Notes by David Kreda added to last week's minutes after the call
  - *DAM Update*
    - *CGWG Group Edits – Comments and Markups closed*
    - *Target for final vote-ready version: Tuesday, December 6, pre-meeting*
      - *Suggest a 20-minute slot at top of that call [if call not devoted to Amon's Presentation] to walkthrough key changes due to group input*
    - *1 week to read and vote, so decision by Tuesday, December ~~13~~ 20*
    - *Materials will include:*
      - *Google doc - with markups and comments – so people can see changes (exclusive of formatting, typos, doc naming, table of contents, etc.)*
      - *Word doc (and/or PDF) would be clean = The vote candidate*
    - *Should emphasize this DAM is about "Clinical Sequencing" – not "Omics"*
    - *Proposed to allow, if CGWG group was amenable, that Bob Freimuth would be asked to provide answers to his own DAM comments in the next week such that and DAM editors might yet be able to review and, if sensible to them, meld them into the document.*
    - *RESTATING: Would expect a "by poll" vote up or down week of December 20*
      - *If YES gets published PDF on site, disseminated*
      - *If NO, figure out what went wrong!*
- **V2 "Lite" / LRI**
  - LRI PSS update
    - Final content by Dec 4 (see above)
    - Do we need a new PSS for V2 Lite? - options as emailed by Dave Hamill of the PMO office
    - ***1213 - HL7 v2 genomics report lite***

- *Sponsor: OO* (note by Bob M: ownership needs to change)
- *Co-Sponsor: CG*
- *PSS does not specify the level of involvement of the Co-Sponsor*
- *Deliverable already created: HL7 Version 2 Implementation Guide: Clinical Genomics Coded Reporting, Release 1 (also known as "V2 Lite")*

- ***1294 - LRI IG***
  - *Sponsor: OO*
  - *Co-Sponsors: CG, PHER*
  - *PSS indicates the level of involvement of both Co-Sponsors to be:*
    - *Request formal content review prior to ballot*
    - *Request periodic project updates. Specify period: Ongoing*
    - *Other Involvement: Provide/review content for relevant sections.*
- Final deliverable to be created: one single, integrated LRI IG (not two documents), which will include "V2 Lite"

- ***Scenario 1***
  - *Additional "non-V2 Lite" are to be produced by Project 1213.*
  - *Project 1294 will move forward with "V2 Lite" to create one single, integrated LRI IG (not two documents).*
  - *As a Co-Sponsor of 1294 with the level of involvement specified in the PSS, CG WG will receive project updates and reviews content prior to ballot.*
  - *Action to be taken with the projects within this scenario:*
    - *Do one of the following with project 1213:*
      - *Modify 1213 to focus on "non-V2 Lite" work and it's balloting strategy*
        - *Change sponsor to CG WG*
        - *Identify co-sponsors and their levels of involvement*
        - *Gather approvals from the necessary entities: USRSC, Sponsor, co-sponsor(s), FMG WG, Steering Division, TSC*
      - *Archive 1213 and create a brand new project to focus on "non-V2 Lite" work and it's balloting strategy, then proceed through the approvals specified above*
    - *CG WG continues their involvement in reviewing LRI IG content as a co-sponsor of 1294.*
    - *1294 proceeds as-is*
- ***Scenario 2***
  - *No further deliverables are to be produced by Project 1213.*
  - *Project 1294 will move forward with "V2 Lite" to create one single, integrated LRI IG.*
  - *As a Co-Sponsor of 1294 with the level of involvement specified in the PSS, CG WG will receive project updates and review content prior to ballot (where they can weigh in on that content from a CG WG point of view)*
    - *Action to be taken with the projects within this scenario:*

        - *Archive 1213 since no other deliverables will be produced.*
        - *CG WG continues their involvement in reviewing LRI IG content as a co-sponsor of 1294*
        - *1294 proceeds as-is*

    - *PMO Comment: In Scenario 2, there is no need to create a new PSS to replace 1213 nor modify 1213 to have the scope be something like "CG WG own/review the "V2 Lite" content". That task is taken care of 1294's co-sponsor level of involvement, and this is a perfect situation as to why we expanded the PSS template to include co-sponsor level of involvement.*

  Hans suggests to rather modify the current PSS, just start a new PSS for non-V2 Lite material, if any. Want to make sure all components in LRI are synced, including V2 Lite, and there is no confusion.

  Dave H - re Scenario 1 and non-V2-Lite material: some projects can have multiple deliverables. In this case, since the 1213 PSS is focussed on just V2 Lite, 1st scenario is invalid.

  Gil - we should make sure scope is clearly defined

  Hans - we can use LRI PSS to include V2 Lite material/scope; LRI PSS is thin at the moment, but we can enhance and add more CG scope to it.

- Weekly V2 Lite sub-group meeting
    - Mondays, at 12:00-1:00PM ET
        - day/time based on respondents to doodle poll sent out last month
    - meeting info TBD
    - The subgroup will discuss and vote for recommended dispositions, and the final block vote will happen in an upcoming Tues call, giving all the members a summary of the what the sub-group recommends and at least a full week to digest the summary recommendation.
    - Hans - need coordinate between LRI groups to make sure all is harmonized

- **San Antonio WGM - Agenda**
    - http://wiki.hl7.org/index.php?title=File:HL7_WGM_Jan2017_-_Clinical_Genomics_Agenda.docx
- **FHIR Ballot reconciliation update**
    - FHIR reconciliation spreadsheet
        - Update preview and submit status to fhir-svn
        - Short url - http://bit.ly/2gaglig
        - Long url - https://docs.google.com/spreadsheets/d/17fqbTWKrIvPysZXIhlhLX4proKpv95inFQGVNO54axk/edit#gid=0
    - FHIR Subgroup Meeting minutes:

        - Thanksgiving last Thu, so no CG FHIR meeting
        - Short url - http://bit.ly/2gvRSbq
        - Long url: https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit?pref=2&pli=1#heading=h.ntntuef6iwzkDAM
    - Uploaded dispositions passed on Nov 17 to gforge tracker. (Nov. 20)
    - Upcoming deadlines based on email from lloyd about FHIR workflow:
        - Timelines for the revised cycle are as follows:
        - ~~**Sun. Nov. 27**- Ballot substantive resource freeze (prioritize resources that IGs will be based on)~~
        - **Sun. Dec. 4** - Ballot total freeze
        - **Sun. Dec. 9** (or a day or two earlier) - Freeze released - all changes allowed
        - **Sun. Feb. 5** - Ballot reconciliation deadline - All ballot comments must be reconciled, tracker issue report must be clean
        - **Sun. Feb. 19** - Publication substantive resource freeze
        - **Sun. Feb 26** - Publication total freeze
        - **Mon. Feb 27** - QA period opens
        - **Tue. Feb 28** - FMM QA spreadsheet updated for all WG resources
        - **Sun. Mar. 13** - QA period closes
        - **Sun. Mar. 20** - All QA applied
        - "following week" STU 3 is published

- **Decision Making Processes**
    - http://www.hl7.org/documentcenter/public/wg/clingenomics/HL7_WG_DMP_v3.0_CG_v2_09172014%20final.doc
    - Amnon: I like the DMP presented by Bob; I would suggest to be clearer on what must go to the whole group, e.g., I think that major negative ballot comments should be voted in the whole group
- **Other**
    - none
- **Chat**
- from Bret Heale to Everyone:
    - From HL7Wiki. Definition: A Domain Analysis Model is an abstract representation of a subject area of interest, complete enough to allow instantiation of all necessary concrete classes needed to develop child design artifacts."
- from Bret Heale to Everyone:
    - A Domain Information Model (DIM) represents the information content required to support a particular {#Domain|domain] within HL7. A DIM will commonly represent all of the information content that may required to be communicated in order to provide interoperability for the host domain, and will, therefore, be the

source for all "derived" information models that are required to support HL7 interoperability specifications in that domain.

- from Bret Heale to Everyone:
    - So, the DIM is where you would map various implementations. The DAM would be the use cases you would use to test your implementation.
- from Bret Heale to Everyone:
    - The DIM should capture everything in the DAM and more