HL7 Clinical Genomics Weekly Call - December 19, 2017 11:00 AM (US Eastern)

Minutes:
https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_I5Kt72NEBYXZuwhFg/edit

Minutes (short url):
http://bit.ly/2aqVmqz

Attending the meeting:

- Join the online meeting (VoIP available with this):
  - Online Meeting Link:
    - https://join.freeconferencecall.com/clingenomics
  - Online Meeting ID:
    - clingenomics
- Dial into the conference:
  - Dial-in Number:
    - (515) 604-9708 - United States
  - Access Code:
    - 289092
  - International Dial-in Numbers:
    - https://www.freeconferencecall.com/wall/clingenomics/#international

Agenda

Minutes Approval

Topics to review

- Upcoming agendas
- External efforts
- Subgroup reports

Topic 1: Referenceable variant knowledge versus patient genetic variant findings

Continued discussion from last week - Larry Babb

Topic 2: Jan 2018 WGM agenda (New Orleans)

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Attendees

1. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
2. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
3. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
4. Larry Babb - Sunquest - larry.babb@sunquestinfo.com
5. Joel Schneider - NMDP - jschneid@nmdp.org
6. David Poloway - BCH - dwpoloway@gmail.com
7. Scott Robertson - Kaiser Permanente - scott.m.robertson@kp.org
8. Joseph Kane - Epic - jkane@epic.com
9. Lei - Xiamen University - liulei6696@gmail.com
10. Caterina Lasome - AFMS - cat@ioninformatics.com
11. Amnon Ptashek - Edico Genome - genptashek@gnome.com
12. Fan Lin - Xiamen University- fanatxmu@gmail.com
13. Bret Heale - Intermountain Healthcare - bheale@gmail.com
14. Shennon Lu - NLM - shennon.lu@nih.gov
15. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
16. Ling Teng - BCH-tenglingling@gmail.com
17. Kevin Power - Cerner - kpower@cerner.com

Presiding Chair: Bob Milius

Minutes Approval

- Dec 12
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20171212.pdf
    - motion/2nd to accept minutes - Joseph Kane/Larry Babb
    - discussion - none
    - abstain - Bob Freimuth, Scott Robertson,
    - nay - 0
    - yea - 12
    - result - passes

Topics to review

Upcoming agendas

Date	Co-Chair	Important Dates / Topics
~~Sep 19~~	~~NO CALL~~	~~2017-09-24: Deadline to submit new project scope statements with deliverables in the Jan18 ballot cycle to dhamill@hl7.org~~ ~~**NO CALL**~~

~~Sep 26~~	~~Bob M~~	~~2017-09-29: Deadline to request meeting space at the 2018Jan WGM (WG Health metric)~~ ~~2017-09-29: Deadline to post your minutes from the San Diego WGM (WG Health metric)~~ ~~DAM Clinical Genomics NIB~~
~~Oct 3~~	~~Kevin~~	~~**NO CALL**~~
~~Oct 10~~	~~Bob M~~	~~**NO CALL**~~
~~Oct 17~~	~~Kevin~~	
~~Oct 24~~	~~Bob M~~	~~2017-10-27: Deadline to notify HQ of additions/changes/corrections to co-chair openings~~
~~Oct 31~~	~~Kevin~~	~~2017-11-01: FHIR Connectathon Track submissions due~~ ~~2017-11-01: Co-Chair call for nominations opens~~ ~~2017-11-03: Initial Harmonization proposals due~~ ~~**NO CALL**~~
~~Nov 7~~	~~Kevin~~	~~2017-11-12: Deadline to submit the online Notification of Intent to Ballot~~
~~Nov 14~~	~~Bob F~~	~~(BobM is away)~~ ~~Discuss/reconcile FHIR proposals~~
~~Nov 21~~	~~Kevin~~	~~(BobM is away)~~ ~~Discuss and vote on DAM~~ ~~http://tinyurl.com/damcgdoc~~
~~Nov 28~~	~~Bob M~~	~~2017-11-24: Final Harmonization proposals due~~ ~~2017-11-26: Initial ballot content deadline~~
~~Dec 5~~	~~Kevin~~	~~2017-11-29: Harmonization Conference Call (WG Health metric: participation in the call or notifying the harmonization listserve that your WG has reviewed with no changes)~~ ~~2017-12-01: Co-Chair Nominations Close at 5:00 pm Eastern~~ ~~2017-12-03: Reconciliation of previous ballots must be completed and posted to the ballot website~~ ~~SNOMED / LOINC - Structuring Genomic Results~~
~~Dec 12~~	~~Bob M~~	~~Larry Babb - Separation of these very important "kinds" of observations and how they relate to referenceable variant knowledge versus patient genetic variant findings~~
Dec 19	Bob M	2017-12-15: Co-Chair election statements due to HQ 2017-12-17: Final content for ballot deadline

		2017-12-21: Consensus Pool signup (must sign up to comment on ballots!) Discuss WGM agenda Continue discussion of Larry Babb's presentation from Dec 12
~~Dec 26~~		2017-12-22: Provisional ballot opening
~~Jan 2~~		**NO CALL**
Jan 9		2017-01-08: Deadline to post your WGM agenda on the WGM information page (WG Health metric)
Jan 16		The Variant Interpretation for Cancer Consortium (VICC) - Joint meeting
Jan 23		

Jan 27 - Feb 2: January 2018 Working Group Meeting (New Orleans, LA USA)

External efforts

- GA4GH Genomic Knowledge Standards (GKS)
    - Two subgroups: Variant Representation, Variant Annotation
    - Each subgroup has drafted a 2-year roadmap, including deliverables and rough milestones, which will be made public after the GA4GH steering committee meeting in January. Both groups will leverage and contribute to work done in HL7 CG.
- National Academies
    -
- ClinGen/ClinVar
    -
- Variant Modelling Collaboration (VMC)
    - Resolution of comments on VMC 0.1 continues. We hope to complete the process in January 2018 and release 0.2 at that time. Following that release, Reece Hart will transition off of as chair and the work will continue as part of the GA4GH GKS Variant Representation subgroup (see above).
- CDISC PGx
    -
- ONC Sync for Genes
    - Call for Phase 2 pilots is out, deadline for applications has been extended to 8 pm ET on Dec 22 (see CG listserv archive for info). The application process is extremely light-weight (only 10 open-response questions).

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq_vAT9B-Z8GIAyRiM/edit
    - Tentative time slot for future meetings: Thursdays at 10 am ET

    - We will re-start our meetings and lay out a roadmap of priorities and scope prior to the Jan WGM
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5

Topic 1: Referenceable variant knowledge versus patient genetic variant findings

- Continued discussion from last week - Larry Babb
    - reviewed poster, to remind
        - need to represent non-patient specific genomic information
        - knowledge-base, clinician sign-off
        - beyond providence
        - Lloyd - would like to see this kind of thing is addressed at a more generic level. Would be happy to work on this with others.
        - Deepak - we're on the right track. This is in the HL7 space to address this. Need archetypes that can be used programmatically. Need to employ what FHIR has and what we need to extend it. Would like to participate in any further discussions/proposals.
        - Bob M - chicken/egg? do we start with modeling group or with FHIR or both at same time?
        - Larry - maybe start with Observation
        - Bob F - need to be careful that we don't try to model the universe. More of a scope question
    - chat:
        - Bret Heale
            - as evidence evolves and understanding of assoications change - having a single location to add a node or an edge will update the state for any that use the knowledge repository...
        - Larry Babb
            - That's true, Bret. However, there is a notion of "sign off" with the knowledge such that it can be published and amended over time.
        - Bret Heale
            - but regards computing/representation on/of Guidelines, the Clinical Decision Support WG is working on that space with FHIR as well.
        - Bret Heale
            - which does put the representation of guidelines into HL7's scope, at least I think so : ^)
        - Larry Babb
            - I see. I'm not interested at this point to specifically model any one kind of knowledge. Rather the generic evidence/providence/assertions and findings
        - Larry Babb

                - like Observation, we can create Resource(s) in FHIR for the core knowledge pattern and allow great flexibility with capturing new edges and attributes along the graph.
            - Joel Schneider
                - Clinical Decision Support? Point of Care Research? potentially a big subject area
            - Bret Heale
                - The knowledge artifacts currently are related to guidelines.
            - Bret Heale
                - this is different than the kind of raw knowledge Larry is talking about
            - Kevin
                - http://build.fhir.org/clinicalreasoning-module.htm
            -
        -
- Slides available
    - http://www.hl7.org/documentcenter/public/wg/clingenomics/HL7%20CG%20Call-ClinGen%20Model%20Review-20171212.pptx
- Poster on this work
    - http://www.hl7.org/documentcenter/public/wg/clingenomics/DMWG-interpretation-ASHG-final.pdf

Topic 2: Jan 2018 WGM agenda (New Orleans)

- Draft agenda
    - https://docs.google.com/document/d/1mQh_H-4Bnlu3548MQoxg5S3nBQN9fKT62FsEb3jOAJ0/edit
    - working on having several quarters devoted to FHIR IG unification
    - Lloyd available Mon Q3, and Wed Q4. Also available during Connectathon for discussions.
    - Please read and comment on current draft.

Clinical Genomics Docs

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - Review complete as of Aug 1, 2017
    - Approved in Sep WGM in San Diego
- Decision Making Process
    - https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit
    - Review complete as of Aug 15, 2017
    - Approved in Sep WGM in San Diego

- DAM
    - http://tinyurl.com/damcgdoc