HL7 Clinical Genomics Weekly Call - December 12, 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

Attendees

1. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
2. Jeremy Warner - VUMC - jeremy.warner@vanderbilt.edu
3. Joseph Kane - Epic - jkane@epic.com
4. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
5. Bob Dolin - Elimu - BDolin@Elimu.io
6. David Kreda - HMS - david.kreda@gmail.com
7. Bret Heale - Intermountain Healthcare - bheale@gmail.com
8. Xin Liu - BCH - xinliu215@gmail.com
9. Dorina Bratfalean -dbratfalean.external@cdisc.org
10. Amnon Ptashek - Edico Genome - genptashek@gmail.com
11. David Poloway - BCH - dwpoloway@gmail.com
12. Larry Babb - Sunquest - larry.babb@sunquestinfo.com
13. Shennon Lu - NLM - shennon.lu@nih.gov
14. Clem McDonald - clemmcdonald@mail.nih.gov
15. Lei Liu - XMU liulei6696@gmail.com
16. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
17. Joel Schneider - NMDP - jschneid@nmdp.org
18. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
19. Fan Lin- Xiamen University - fanatxmu@gmail.com
20. Ling Teng - BCH - tenglingling@gmail.com
21. Martin Maiers - NMDP - mmaiers@nmdp.org
22. Grant Wood - Intermountain - grant.wood@imail.org
23. Kevin Power - Cerner - Kevin.Power@Cerner.com

Presiding Chair: Bob Milius

Minutes Approval

- Dec 5
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20171205.pdf
    - motion/2nd to accept minutes - Joseph Kane/Jeremy Warner
    - discussion - none
    - abstain - David K,
    - nay - none
    - yea - 17
    - 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		2017-12-15: Co-Chair election statements due to HQ 2017-12-17: Final content deadline Discuss WGM agenda
Dec 26		2017-12-22: Provisional ballot opening
Jan 2		
Jan 9		2017-01-08: Deadline to post your WGM agenda on the WGM information page (WG Health metric)
Jan 16		
Jan 23		

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

External efforts

- GA4GH Genomic Knowledge Standards (GKS)
  - nothing new
- National Academies
  - nothing new
- ClinGen/ClinVar
  - nothing new
- Variant Modelling Collaboration (**VMC**)
  - nothing new
- CDISC PGx
  - Weekly meeting on CDISC PGx modeling, this week includes PF domain discussion

Subgroup reports

- IM (Bob F)
  - https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq_vAT9B-Z8GIAyRiM/edit
  - Doodle poll to determine meeting times: https://doodle.com/poll/ua7t34epzcehiecg
  -
- FHIR (Gil)
  - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5

Topic 1: Referenceable variant knowledge versus patient genetic variant findings

- Larry Babb - see email to listserv:

  Kevin –

  You have pointed out a very important point here. My understanding is that the LRI definition is used in specific patient variant findings, typically somatic tumor testing when the lab is quantifying the amount of the variant in a tumor sample. They refer to it as "frequency" in many cases. It is very confusing because the use the same term (as most folks are pointing out) to align with the LOINC term below. The population frequency is a very helpful and widely used annotation that can be associated to a variant independently of a patient sample as we see in the major population study databases.

  I believe Jeremy Warner would be able verify/clarify the use of the concept of allele frequency for the percentage of a variant found in a assayed tumor sample. I think most folks are familiar with the term being used as population frequency.

  ClinGen has been working for the past year on standardizing the set of minimal evidence types (annotations/observations) which are used to back variant assertions used in clinical reporting of variants. We are doing this work along with the Monarch Intiative's SEPIO project (both ClinGen and Monarch are driver projects for the GA4GH). In our evidence types we do structure and formalize population allele frequency. The SEPIO group is working to formalize the population codes found in EXAC, gnomad and 1000Genomes, it seems like these may end up in the NCI Thesaurus if not already there.

  I would be happy to show you the work and separation of these very important "kinds" of observations and how they relate to referenceable variant knowledge versus patient genetic variant findings, if interested.

  Larry
- Slides available
  - http://www.hl7.org/documentcenter/public/wg/clingenomics/HL7%20CG%20Call-ClinGen%20Model%20Review-20171212.pptx
- Discussion to be continued next week.

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