HL7 Clinical Genomics Weekly Call - December 5, 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: SNOMED CT and Clinical Genomics: Use case for precision medicine

Topic 2: Discussion/reconciliation of FHIR proposals (continued)

Clinical Genomics Docs

Attendees

1. Kevin Ehlers - Bloodcenter of WI. - kevin.ehlers@bcw.edu
2. David Poloway - BCH - dwpoloway@gmail.com
3. Jeremy Warner - VUMC - jeremy.warner@vanderbilt.edu
4. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
5. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
6. Bob Dolin - Elimu - BDolin@Elimu.io
7. Amnon Ptashek - Edico Genome - genptashek@gmail.com
8. Dorina Bratfalean - CDISC- dbratfalean.external@cdisc.org
9. Gabe Bautista - Epic - gbautist@epic.com
10. Joseph Kane - Epic - jkane@epic.com
11. Xin Liu - BCH - xinliu215@gmail.com
12. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
13. Scott Robertson - Kaiser Permanente - scott.m.robertson@kp.org
14. Clem Mcdonald - clemmcdonald@mail.nih.gov
15. Joel Schneider - NMDP - jschneid@nmdp.org
16. Fan Lin- Xiamen University - fanatxmu@gmail.com
17. Bret Heale - Intermountain Healthcare - bheale@gmail.com
18. Fan Lin- Xiamen University - Fanatxmu@gmail.com
19.

Presiding Chair: Kevin Power

Minutes Approval

- Nov 28
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20171128.pdf
    - motion/2nd to accept minutes - Scott Robertson / Joseph
    - discussion - None
    - abstain - None
    - nay - None
    - yea - <All>
    - result - Pass

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)
    -
- National Academies
    -
- ClinGen/ClinVar
    -
- Variant Modelling Collaboration (**VMC**)
    -
- CDISC PGx
    -

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq_vAT9B-Z8GIAyRiM/edit
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5

Topic 1: SNOMED CT and Clinical Genomics: Use case for precision medicine

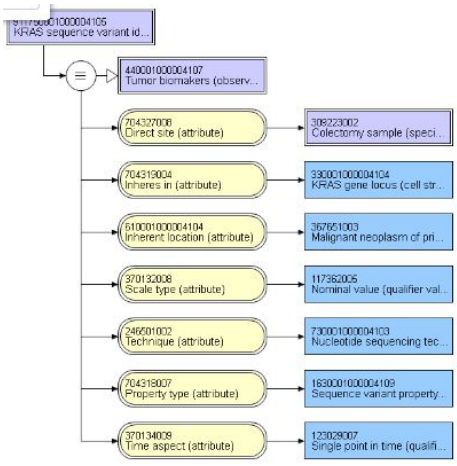
Guest speakers:
James R. Campbell MD
W. Scott Campbell PhD

https://confluence.ihtsdotools.org/pages/viewpage.action?pageId=29950423

KP: Updated Version of presentation send Wed Dec 6 to list serv

In this figure (where inherent location = malignant neoplasm), is it correct that you've specifically defined this concept as a variant in a cancer, and therefore this concept wouldn't be applicable for germ line mutations?

That is correct. We use a different inherent location for germ line mutations.



Which version of FHIR?

Unsure, likely STU3 ('main website') - this was done ~ 2 weeks ago.

Are the SNOMED CT concepts available?

With a UMLS account, available via their website.

Most of the variables have existing LOINC codes.

Yes, many are very general concepts. They wanted more specific observables.

Maintain separation between observation / interpretation? In the example V2 msg, seemed it was included?

Data between systems connected by date observed, patient ID, and sample ID. Becomes pragmatic issue (where to store what?) - but always maintain link. Biobank has separation, but movement into the EHR bundles together. Yes. Their Biobank is fully identified, not as much of an issue in going back.

Plan to push forward as standard?

Yes. Conversations between SNOMED Int and Regrenstief. Questions of how the data would be interoperated is still in question.

SNOMED usage globally?

Yes, it is a struggle. Some use one, or other, and some both. Should work together, but maintaining is difficult.

Mutate your model to fit new CG work?

Extend the discussion, and yes.

Topic 2: Discussion/reconciliation of FHIR proposals (continued) - DID NOT DISCUSS

FHIR Ballot Timeline:
http://wiki.hl7.org/index.php?title=FHIR_Ballot_Prep

I propose we hit the May 2018 cycle with our first ballot, then the Sept 2018 cycle for rework if needed. This plan gets the "unified" IG into R4.

Key deadlines for both are highlighted below:

May 2018 Cycle

Single FHIR Core STU ballot, multiple Normative FHIR Core ballots, possible IG R4-based STU ballots

- 2018-02-14 - Last date to inform FMG of intention to ballot IGs as part of May ballot.
- 2018-02-21 - Deadline for connectathon proposals to FMG; Deadline for resource and IG proposals that are candidates for STU in R4

Sept 2018 Cycle

possible Normative re-ballots, possible limited STU FHIR Core ballot, possible IG R4-based STU ballots:

- 2018-06-06: Deadline for connectathon proposals to FMG

· 2018-07-01: Work groups notify the FMG whether they need to reballot normative packages (due to substantive change), STU resources (due to significant refactoring) or IGs during the Sept. cycle

Chat Log:
Bret Heale 10:42AM
is this from Sequence Resource, Diagnostic Report for genomics or Genetic Observation?

Bret Heale 10:52AM
i presume the true seperation is in the graphDB where new information can be added

Bret Heale 10:57AM
The SNOMED CT concepts would be quite useful…

Clinical Genomics Docs

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - Review complete as of Aug 1, 2017
- Decision Making Process
    - https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit
    - Review complete as of Aug 15, 2017
- DAM
    - http://tinyurl.com/damcgdoc