HL7 Clinical Genomics Weekly Call - January 9, 2018 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: “FINAL” Jan 2018 WGM agenda (New Orleans)

Topic 2: New DMP for all groups

Topic 3: Unification questions from Lloyd

Clinical Genomics Docs

Attendees

1. David Poloway, BCH - dwpoloway@gmail.com
2. Lei - XMU - liulei6696@gmail.com
3. Xin Liu - BCH - xinliu215@gmail.com
4. Clem McDonald - NLM - clemmcdonald@mail.nih.gov
5. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
6. Shennon Lu - NLM - shennon.lu@nih.gov
7. Bob Dolin - Elimu - BDolin@Elimu.io
8. Alex Mankovich - Philips - alex.mankovich@philips.com
9. Amnon Ptashek - Edico Genome - genptashek@gmail.com
10. Dorina Bratfalean- CDISC - dbratfalean.external@cdisc.org
11. Joseph Kane - Epic - jkane@epic.com
12. JD Nolen - Children's Mercy Hospital - jlnolen@cmh.edu
13. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
14. Grant Wood - Intermountain Healthcare - grant.wood@imail.org
15. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
16. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
17. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
18. Scott Robertson smrobertson4@gmail.com
19. Larry Babb -Sunquest Mitogen (GeneInsight) - larry.babb@sunquestinfo.com
20. David Kreda - HMS - david.kreda@gmail.com
21. Ling teng -BCH-tenglingling@gmail.com
22. Elizabeth Newton - elizabeth.h.newton@kp.org
23. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
24. Fan Lin--XMU--fanatxmu@gmail.com
25. Andrea Pitkus - Intelligent Medical Objects (IMO) - apitkus@imo-online.com (last half of call)

Presiding Chair: Kevin Power

Minutes Approval

- Dec 19
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20171219.pdf
    - motion/2nd to accept minutes - Bob M / Bob D
    - discussion - None
    - abstain - None
    - nay - None
    - yea - 14
    - 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~~	~~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	Kevin	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)
    -
- National Academies
    -
- ClinGen/ClinVar
    -
- Variant Modelling Collaboration (VMC)
    -
- CDISC PGx
    -
- ONC Sync for Genes
    -

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq_vAT9B-Z8GIAyRiM/edit
    - Recurring meeting about to be setup for Thursdays (Bob F sent invite today)
        - Starting Jan 18
        - 10:00-11:00 am ET
- FHIR (Gil)

- https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5

Topic 1: “FINAL” Jan 2018 WGM agenda (New Orleans)

- Agenda
    - http://wiki.hl7.org/index.php?title=File:HL7_WGM_January2018_-_Clinical_Genomics_Agenda.pdf
- Add about the ISO genomics meeting on Thursday

Topic 2: New DMP for all groups

This year, we’re simplifying things. In our review, we observed that modifications are limited to a small number of practices. We’ve decided to have a single set of DMPs that will apply to all Work Groups and Committee. Only certain sections of the DMPs are open to modification, specifically:

- Section 5 – Quorum Requirements
- Section 7 – Electronic Voting (particularly items c, d and e)
- Section 8 - Proxy Participation (for closed committees with named members only)

Instead of asking each Work Group to post its DMPs on its webpage, work groups will post only their Addendum (should they have one) to their webpage. If no addendum is found, an interested party doesn’t need to read through an entire set of DMPs to realize that the Work Group has simply adopted the default set. If there is an Addendum on a Work Group’s page, interested parties can quickly see the few areas that differ from the default DMPs without having to read through several pages.

We ask that each Work Group review the DMPs and the Addendum Template at the links below during or shortly after the January Working Group Meeting:

- Default DMPs: http://www.hl7.org/documentcenter/public/procedures/Default_HL7_WG_DMP_2017_v1_2017-12-18.dmp.docx

- DMP Addendum Template: http://www.hl7.org/documentcenter/public/procedures/DMP%20Modification%20Template_10312017_Clean2.docx

If your committee does NOT wish to make any modifications to Sections 5, 7 or 8 your work is done. Simply send an email to your Steering Division Co-chairs notifying them of that decision. If your Work Group does wish to modify Sections 5, 7 and/or 8, you will have until the May 2018 Working Group Meeting to complete and send the Addendum Template with your Work Group’s changes to your Steering Division Co-chairs. If no communication is received from your Work Group by the May Working Group Meeting regarding the DMPs, the Default DMPs will apply.

Topic 3: Unification questions from Lloyd

https://docs.google.com/document/d/1juWEnjyXV34yYmPq3FDpLAiJlM0Hiv0FyNBfvPD6enM/edit?ts=5a4e61a2
Overall Variant ISCN - Name is not right

Naming / Boundries need clarification:

Overall Variant ISCN
Discrete Variant
Complex Variant
Structural Variant

Should try to not define by the code system

Fuzzy overlap with the Cytogenetic V2

Do we support Cytogenetics with this model? - Delay for now.

Discrete - Contiguous, tightly bound
Complex - 2..* Discrete

Haplotype is a Complex

Discrete must exist on a single chromsome
Discrete can cross genes

Complex variant cross Chromosome? Yes and No :)

Discrete is an Allele? Or Genotype?

Lloyd will prepare a strawman and share with the group.

Chat:

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Here is a listing of the CAP accreditation checklists and “genetics” areas which they cover, so folks can see which regulatory aspects overlap with CG workgroup activities:
https://www.cap.org/ShowProperty?nodePath=/UCMCon/Contribution%20Folders/WebContent/pdf/accreditation-checklists-full-listing.pdf

Although current checklists are proprietary and require purchase, older versions highlight many of the genetic regulatory requirements here:
http://www.cap.org/ShowProperty?nodePath=/UCMCon/Contribution%20Folders/DctmContent/education/OnlineCourseContent/2016/LAP-TLTM/resources/AC-molecular-pathology.pdf

Pertinent to this discussion may be: MOL.36155 Sequence Variants - Interpretation and Reporting; & MOL.35785 Cytogenomic Microarray Report Elements (including ISCN).

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