HL7 Clinical Genomics Weekly Call - March 6, 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

- Agendas and Important Dates
- External efforts
- Subgroup reports

Topic 1: Proposed "Note to balloter" language - PLEASE REVIEW

Topic 2: Update on Publishing IG

Topic 3: IG

Topic 4: Unification (topics for next call)

Chat

Attendees (Presiding Chair: Kevin Power)

1. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
2. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
3. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
4. Shennon Lu - NLM - shennon.lu@nih.gov
5. Clem McDonald - NLM - clemmcdonald@mail.nih.gov
6. Dorina Bratfalena - CDISC-dbratfalean.external@cdisc.org
7. Kevin Ehlers - BloodCenter of Wisconsin - kevin.ehlers@bcw.edu
8. Xin Liu - BCH - xinliu215@gmail.com
9. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
10. Amnon Shabo (Shvo) - Philips - amnon.shabo@philips.com
11. David Poloway - BCH - dwpoloway@gmail.com
12. Alex Mankovich - Philips - alex.mankovich@philips.com
13. JD Nolen - Children's Mercy Hospital - jdlnolen@gmail.com
14. Lei Liu - XMU - liulei6696@gmail.com
15. Julian Sass - Niederrhein University - julian.sass@hs-niederrhein.de
16. Bret Heale - Intermountain Healthcare - bheale@gmail.com
17. Scott Robertson - Kaiser Permanente - scott.m.robertson@kp.org
18. Elizabeth Newton - Kaiser Permanente - elizabeth.higgs.newton@kp.org
19. Amnon Ptashek - Edico Genome - genptashek@gmail.com
20. Joel Schneider - CIBMTR / NMDP - jschneid@nmdp.org
21.

Minutes Approval

- Feb 27
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20180227.pdf
    - motion/2nd to accept minutes - David/Lloyd
    - discussion -
    - Abstain / Nay / Yea:
        - 0 / 0 / 12
    - result - Passes

Topics to Review

Agendas and Important Dates

Date	Co-Chair	Agenda	Important Dates
6-Feb	Kevin	WGM Review Unification	
13-Feb	Bob M	Unification	Inform FMG of NIB: Feb 14 - done **HL7 Genomics Conference**: Feb 20 - Feb 21
20-Feb	Kevin	Unification	Connectathon proposals to FMG: Feb 21 NIB submission deadline: Feb 25 - done
27-Feb	Bob F	Unification	
6-Mar	Kevin	FHIR connectathon proposal Note for Sequence resource CG FHIR PSS update Unification	
13-Mar	Bob M	Vote on: https://docs.google.com/document/d/1gMJnjv7BNtnU8GIcEu7mchygaAsJal5O1BMSY8ZOIFs/edit#heading=h.uonrk9s9aill Unification	
20-Mar	Kevin		

27-Mar			FHIR Final Content Freeze: April 1
3-Apr	Bob M		May 2018 Ballot opens: April 6
10-Apr			
17-Apr			
24-Apr	Bob M		
1-May			May 2018 Ballot closes: May 7
8-May			
May 2018 HL7 Working Group Meeting May 12, 2018 to May 18, 2018 - Maritim Hotel Köln, Cologne, Germany			

External efforts

- GA4GH Genomic Knowledge Standards (GKS) (leads: Bob Freimuth, Andy Yates)
    - Beginning discussions about “equivalence” (various definitions and types thereof) and starting to model structural variants.
- National Academies (Grant Wood, JD Nolen)
    -
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
    -
- Variant Modelling Collaboration (VMC)
    - Continuing to work on VMC 0.2, meeting less frequently as GKS picks up so moving a bit slower than previously
- CDISC PGx
    -
- ONC Sync for Genes
    - Pilot sites have been selected by ONC/NIH. Announcement is being prepared. Further information will be available after the official announcement.

Subgroup reports

- IM (Bob F)
    - Modeling Sequence and SequenceRepresentation. Draft model for Location (coordinate-based, approximate, and cytogenetic). Will continue to refine the semantics of the draft models as we work toward a model for Variant, following the lead of the VMC. Anticipate sharing the draft model with the full CG work group within 2-3 weeks, but our minutes are available at any time.
    - IM group will NOT meet next week due to the AMIA summit
    - https://docs.google.com/document/d/1azKiQdhAQKuHhxAznEp8141FLdFLACIu8MzF2LxADxg/edit#
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5

Topic 0: Propose Connectathon Tracks

Official deadline for Connectathon proposal at May WGM has passed, but we can still submit proposals for consideration.
Kevin P will send out a doodle for counting attendees (Bob F suggests the poll make it clear that it is looking for attendees at the Connectathon, not the WGM in general) (Bob M suggests we get an idea of likely attendance to both WGM and separately Connectathon)

https://doodle.com/poll/cyadwwiiw9hkvy8r

Topic 1: Proposed "Note to balloter" language - PLEASE REVIEW

From Lloyd:
I'd promised to put together some draft language for the "Note to balloters" that we were going to include on the Sequence resource. I've also created some proposed language for inclusion on the existing "guidance" page and profiles. I've put the language into a Google doc for ease in proposing adjustments to the wording and making comments.

https://docs.google.com/document/d/1gMJnjv7BNtnU8GIcEu7mchygaAsJal5O1BMSY8ZOlFs

Kevin - Plan to vote on this by next week.

Topic 2: Update on Publishing IG

From Bob M:
CG'ers,

In order to ballot the unified FHIR genomics implementation guide in May, we need to have a Project Scope Statement that refers to it. We indicated in our Notice of Intent to Ballot (NIB) to refer to PSS 1217 (the one we use for the basis of all of our FHIR work). However, we were notified by the TSC that this PSS does not mention Implementation Guides as possible artifacts and we will need to create a new PSS to cover it. But given the timeline, and the fact that this IG is based on core profiles, and was recommended to be put into an IG by the FMG, they will accept for now the current PSS if we simply the update 1217 to include Implementation Guides. That's what I did, and have it attached. Changes are highlighted. They include:
- added implementation guide as a possible artifact in the scope,
- checked Implementation Guide in Intent,
- checked DSTU to Normative under ballot type.

Please take a look. We plan on voting on this update on Tuesday's call. There are a number of other things we should update as well later, but these changes are all we proposing right now to keep it simple and to allow the IG ballot to happen.
Later, we will develop a separate PSS specifically for the IG for the September ballot, if still needed.

The HL7 Project Management Office looked at the updated PSS we're proposing (and that we're voting on tomorrow), and suggested some minor additions to the timelines.

Mark CG FHIR Profiles in FHIR core as deprecated	**May 2018**
STU Ballot of FHIR IG	**May 2018**
Pull CG IG profiles out of FHIR Core (per FMG guidance)	**Sep 2018**
STU Ballot of FHIR IG, based on reconciliation of comments from May 2018 ballot	**Sept 2018**
Reconcile and publish FHIR IG STU	**Dec 2018**

I'm attaching the update version here.
It's also available on the CG documents page:
http://www.hl7.org/documentcenter/public/wg/clingenomics/docs/2018MAR_PSS_ClinGenomics_CGFHIR_3.docx

Motion: Accept the PSS
Move/Second: Bob M/Clem
Discussion: None
Vote: Abstain (0) / Nay (0) / Yea (18)
Motion Passes.

Topic 3: IG

Current draft:
http://build.fhir.org/ig/HL7/genomics-unified/index.html
For questions on the documentation portions, let try to ask those via email or chat.fhir.org for now.

- *Higher priority*
  - *Genetic Interpretation (subclasses of)*
  - *Genetic Impact (subclasses of)*

Chat for discussing Impact:
https://chat.fhir.org/#narrow/stream/genomics/subject/Genetic.20Impact
General consensus: Continue with current model while looking towards working with CDS WG as they define a new 'knowledge' set of resources.

- *High priority*
  - *Described Variant*
  - *Sequence*

Described Variant (DV):

- Items in DV and Sequence (arrCGH, copy number, outer/inner start/stop)
    - In DV they are patient specific, in Sequence they would be definitional - so keep in both.
    - Should add this as a component to DV:
        - http://hl7.org/fhir/sequence-definitions.html#Sequence.coordinateSystem
- Would one use DV when describing a gene sequence (or exon) to express things that are in phase?
    - Seems more like Haplotype or perhaps ComplexVariant
    - Will need to evaluate if they need to carry phase information
- Can dbSNP-ID be another variation-code?
    - dbSNP does not identify a specific variation, so most are uncomfortable.
    - Clem will share further details about the LRI details and corresponding LOINC codes

Topic 4: Unification (topics for next call)

- Start discussing the profiles in order from the WGM:
    - *Really High Priority (I mean it)*
        - *(Genetics) DiagnosticReport*
        - *Specimen Source*
        - *Genetic Observation Common Properties*
        - *Computable Genetic Assertion*
        - *Haplotype*
        - *Genotype*
        - *Overall Variant*
    - *Higher priority*
        - *Genetic Interpretation (subclasses of)*
        - *Genetic Impact (subclasses of)*
    - *High priority*
        - *Described Variant*
        - *Sequence*
    - *Medium priority*
        - *General Recommendation*
        - *Medication Usage Implications*
    - *Low priority*
        - *Current Medication*
        - *Order for Genetic Test*
        - *Descriptive Genetic Assertion (incl subclasses)*
        - *Complex Variant*
        - *Copy Number Change*
        - *Microarray Platform*

Chat

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