HL7 Clinical Genomics Weekly Call - September 18, 2018 11:00 AM (US Eastern)

Minutes:
https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_I5Kt72NEBYXZuwhFg/edit
short url: http://bit.ly/2aqVmqz

Attending the meeting:ss
Join the online meeting (VoIP available with this):

- Online Meeting Link:
    - https://join.freeconferencecall.com/clingenomics
    - 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

Attendees Sign-in

Minutes Approval

Topics to Review

- Agendas and Important Dates
- External efforts
- Subgroup reports

Topic 0: WGM Agenda

Topic 1: Block: Remove the link from SequenceConfiguration (now Sequence Phase Relation) to ComplexVariant

Topic 2: Compare Sequence and Observation

Chat

Minutes: http://bit.ly/2aqVmqz

Attendees Sign-in

(Presiding co-chair: Kevin Power - Cerner - kpower@cerner.com)

1. Bret Heale - Intermountain Healthcare - bheale@gmail.com
2. JD Nolen - Children's Mercy Hospital - jlnolen@cmh.edu
3. Dora Finkeisen - MOLIT Institut - dora.finkeisen@molit.eu
4. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
5. James Jones - BCH - james.jones.bch@gmail.com
6. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
7. Bob Dolin - Elimu Informatics - bdolin@elimu.io
8. Caterina Lasome - iON Informatics - cat@ioninformatics.com
9. Alex Mankovich - Philips - alex.mankovich@philips.com
10. Mullai Murugan BCM murugan@bcm.edu
11. Liz Amos - NLM - liz.amos@nih.gov
12. Andrea Pitkus - apitkus@gmail.com
13. Julian Sass - Berlin Institute of Health - julian.sass@bihealth.de
14. Jamie Parker - Carradora Health - jamie.parker@carradora.com
15. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
16. Ning Xie - BCH - ningxie2018@gmail.com
17. Jungang Zou - BCH - jungang.zou@gmail.com
18. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu (joined fairly late)

Minutes Approval

- Sep 11
  - http://wiki.hl7.org/index.php?title=File:HL7_CG_20180911.pdf
  - Motion/2nd to accept minutes: JD / Cat
  - Discussion: None
  - Vote: (Abstain / Nay / Yea): 0 / 0 / 12
  - Result: passes and minutes accepted

Topics to Review

Agendas and Important Dates

Date	Co-Chair	Agenda	Important Dates

5/29/2018	Bob M	Review WGM minutes (note that Amnon Shabo edited the minutes regarding the sessions when his ballot comments were discussed)	
6/5/2018	Kevin	Ballot comments	Jun 6 - Deadline for connectathon proposals to FMG
6/12/2018	Kevin	Con call tech Connectathon 'Variant Grouping'	
6/19/2018	Kevin	Con call tech Connectathon Block Vote 'Variant Grouping'	
6/26/2018	Bob M		June 27 - Connectathon Proposals Due
7/3/2018			Jul 1 - 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 July 6 - Deadline to notify HG of additions/changes to co-chair opentings
7/10/2018	Bob M	IG block vote NIB vote	July 11 - Call for co-chair nominations July 15 - Notification of Intent to Ballot
7/17/2018	Bob M		
7/24/2018	Kevin	Consensus Groups Definitional Sequence	July 23 - formation of consensus groups
7/31/2018	Bob F	Topic 0: Deadline for resolutions to ballot comments Topic 1: FYI: Ballot Items for follow-up Topic 2: Block Vote #1 Topic 3: Block Vote #2 Topic 4: Proposed changes to Described Variant Topic 5: Secondary findings (proposal) Topic 6: Impact vs. Interpretation	Aug 5 - Reconciliation packages must be posted by this date at the absolute latest Aug 10: All substantive reconciliation applied. FHIR Core is frozen, limited QA process for content subject to ballot only
8/7/2018	Kevin P	Trackers needing follow-up Block Vote	Aug 10 - close to co-chair nominations

		Cytogenetics in the IG Compare Sequence and Observation	
8/14/2018	Bob M	Topic 1: Block Vote #1 Topic 2: Impact/Interp Topic 3: New LOINC codes? Topic 4: What was tested? Topic 5: Cytogenetics in the IG Topic 6: Compare Sequence and Observation	Aug 17: Pre-ballot (and connectathon) content freeze. Publication process begins, including ensuring that content is appropriately flagged for ballot status and there are no last minute QA issues
8/21/2018	Bob M	Topic 0: WGM Agenda Topic 1: Block Vote Topic 2: Ballot reconciliation - 16789 Topic 2: Capturing Genomic Panel Definitions Topic 6: Compare Sequence and Observation Topic 5: New LOINC codes?	Aug 24 - ballot opens for voting
8/28/2018	Kevin P	Topic 0: WGM Agenda Topic 1: Block Vote Topic 2: HL7 V2->FHIR Mapping Topic 3: Capturing Genomic Panel Definitions Topic 4: Compare Sequence and Observation	Aug 31: Deadline to post WGM agenda on the WGM information page (WG Health metric)
9/4/2018	Bob F	Topic 1: OO PSS - Digital Pathology Topic 2: Block votes (2) Topic 3: Discuss tracker 15889 Topic 4: Compare Sequence and Observation	
9/11/2018	Bob M	Topic 0: WGM Agenda Topic 1: Ballot Resolution Proposed: New region-studied profile Ballot Resolution Proposed: Human reference sequence assembly Topic 2: Compare Sequence and Observation	
9/18/2018	Kevin P		
9/25/2018			

32nd Annual Plenary & Working Group Meeting

Sep 29, 2018 to Oct 5, 2018 - Baltimore, MD

Agenda:

https://confluence.hl7.org/pages/viewpage.action?pageId=29753404

External efforts

- GA4GH Genomic Knowledge Standards (GKS) (leads: Bob Freimuth, Andy Yates)
    - Variant Representation (formerly VMC)
        - Work continues on two fronts: we are finishing up the last pieces to the 0.2 release (lead by Reece Hart, next draft release scheduled for this Friday) and we are extending the model to support complex variants (e.g., fuzzy ends, CNV) (lead by Larry Babb/Tristan Nelson)
        - Hoping to have a formal deliverable out by Oct 3
        - https://docs.google.com/document/d/1Sulg3kECnorTEAbutINOsK-lFkKAcKpl6IHgPaPQEgA/edit#heading=h.k9apf9d8j9y2
    - Variant Annotation
        - Collecting use cases, prioritizing for modeling work (lead by Matt Brush and Javi Lopez)
        - Hoping to have a formal deliverable out by Oct 3
        - https://docs.google.com/document/d/13sSChUB9rW7vl1ep-tZnaDzSWb_MyWIvSzEFVS32quE/edit#heading=h.t2adm0gua505
    - Gearing up for the GA4GH plenary meeting (Basel, Switzerland) on Oct 3-5, 2018. This meeting conflicts with the HL7 WGM in Baltimore but future HL7/GA4GH meetings will be scheduled so they do not overlap.
- DIGITiZe (aka National Academies) (Grant Wood, JD Nolen)
    - Stilling planning, Working to get a planning call on the schedule to move things forward (JD)
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
    - no report
- CDISC PGx (Dorina B.)
    - no report
- ONC Sync for Genes (Bob Freimuth)
    - Pilot sites are planning/implementing their respective use cases. ONC will be encouraging their participation in both the Sept 2018 and Jan 2019 FHIR Connectathons.

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/1azKiQdhAQKuHhxAznEp8141FLdFLACIu8MzF2LxADxg/edit#
        - Draft model docs posted at https://docs.google.com/document/d/1Wys14HNJAEB_YJ-EeDPAKX50_oxiDqAKi3WD4wlfjbk/edit
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.zfi9l8jfe4la
        - Initial feedback on Glossary and primer google docs gone over: https://docs.google.com/document/d/1vWY7fHbSl0ZxkTJAXZ8gz8nHUgYrHlKBCYy2bTaxiXs/edit#, https://docs.google.com/spreadsheets/d/1JP9gC1Daaz_pjYkLnrZdLHviFn-qFTZLgGXQmvi487c/edit#gid=1467712360
        - Discussed panel profile (will keep for now), and set up for a future block vote for 4 remaining open trackers

Topic 0: WGM Agenda - October 1-3

See new format here:
https://confluence.hl7.org/pages/viewpage.action?pageId=29753404

Creating a HL7 Confluence/JIRA account

- To get an HL7 JIRA/Confluence account, you need to create an account on www.hl7.org/permalink?JIRA
- follow instructions on https://confluence.hl7.org
- For your username, please use the format "(given name).(family name)". For example, "marie.curie" or "albert.einstein"

Topic 1: Block: Remove the link from SequenceConfiguration (now Sequence Phase Relation) to ComplexVariant

Comment Submitters

- Bob Dolin
- Clem McDonald

Line Items

16262 Change LOINC answer list for complex variant type (Bob Dolin) Not Persuasive with Mod
16808 Complex variants distinguish cis from trans - 2018-May Genomics #36 (Clem McDonald) Persuasive with Mod

- Motion/2nd to accept proposal: Bob D / Bret H
- Discussion:
  - None
- Vote: (Abstain / Nay / Yea): 0 / 0 / 17
- Result: passes

Topic 2: Compare Sequence and Observation

https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg_xpwir8rEORkNjMfemvqxE0/edit#gid=0

Sequence.variant - used to summarize any variants that can be used to rebuild the sequence from a reference. Not to describe “reported” variants

Why make that distinction?

Sequence resource allows a reference to a window range -
New Observation profile could be used to describe the sequence in the same way, and include additional annotations (quality, etc…)

NOTE - https://jef.works/blog/2017/03/28/CIGAR-strings-for-dummies/ describes CIGAR strings from a novice perspective (from Bret H) need to correct the link (http://build.fhir.org/sequence-definitions.html#Sequence.variant.cigar)

Need to describe the use cases when you would use Sequence versus Observation. Keeping to Observation might accelerate implementation.

Do complicated Observation profiles allow for that accelerated implementations?

Depends on what you want to do - if you just store and pass along, the specific details are not as important. However, for systems that really need to understand, they will have to deeply understand the profiles.

GA4GH provides API for the ‘raw’ data. FHIR Observations communicate to EHRs. Sequence is in-between and might fit some intermediate use cases.

Binary resource - “best” fit for more raw data

Observation is more than just Lab -> EHR. Its lab -> PHR. EHR -> EHR. Sequence would be about systems that need the more detail.

What is the subset of information we make available only to genetics focused systems that will not propagate into EHR, PHR, possibly other systems that do not focus on genetics.

VCF is a widely used format - “common currency”. The PDF is still the thing that displays. Could we consider VCF?

- Perhaps “instead of” Sequence
- Along with Sequence

Still missing the coverage/quality scores for different regions.

Is that sort of thing appropriate for FHIR? Seems like GA4GH territory?

What queries/use cases would be needed?

VCF file - where would it go? Attachment to DiagRpt?

Caution - Can be (and will be) variances. YES!

Perhaps an opportunity for standardizing what is delivered “in a VCF” via the Sequence resource?

If you want to speak about 1 molecule, the VCF approach might be “messy”

Chat

Bret Heale
10:23AM
https://jef.works/blog/2017/03/28/CIGAR-strings-for-dummies/ describes CIGAR strings from a novice perspective

Bret Heale
10:51AM
if possible, I would like to see some examples of the structures you have been using for passing on which regions were interrogated.

Clinical Genomics Docs

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - Review complete as of Aug 1, 2017
    - Approved in Sep 2017 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 2017 WGM in San Diego
- DAM
    - http://tinyurl.com/damcgdoc