

## HL7 Clinical Genomics Weekly Call - September 4, 2018 11:00 AM (US Eastern)

### **Minutes:**

[https://docs.google.com/document/d/12-uBrMmav71a3\\_c9h\\_FXQteJo\\_I5Kt72NEBYXZuwHfg/edit](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**

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[Submitted on Thu, 30 Aug 2018 22:40:54 +0000 by](#)

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

## Attendees Sign-in

(Presiding co-chair: Bob Freimuth - Mayo Clinic - [freimuth.robert@mayo.edu](mailto:freimuth.robert@mayo.edu) )

1. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
2. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
3. Bob Milius - CIBMTR/NMDP - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
4. Patrick Werner - Heilbronn University / Molit Institut - [patrick.werner@molit.eu](mailto:patrick.werner@molit.eu)
5. Joel Schneider - NMDP/CIBMTR - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
6. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
7. Jamie Jones - BCH [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
8. Dora Finkeisen - MOLIT Institut - [Dora.Finkeisen@molit.eu](mailto:Dora.Finkeisen@molit.eu)
9. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
10. JD Nolen - Children's Mercy Hospital - [jlnolen@cmh.edu](mailto:jlnolen@cmh.edu)
11. Caterina Lasome - iON Informatics for AFMS - [cat@ioninformatics.com](mailto:cat@ioninformatics.com)
12. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
13. Ning Xie - BCH- [ningxie2018@gmail.com](mailto:ningxie2018@gmail.com)
14. Mullai Murugan BCM [murugan@bcm.edu](mailto:murugan@bcm.edu)
15. Andrea Pitkus - [apitkus@gmail.com](mailto:apitkus@gmail.com)
16. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
17. Jamie Parker - Carradora Health - [jamie.parker@carradora.com](mailto:jamie.parker@carradora.com)

## Minutes Approval

- August 28
  - [http://wiki.hl7.org/index.php?title=File:HL7\\_CG\\_20180828.pdf](http://wiki.hl7.org/index.php?title=File:HL7_CG_20180828.pdf)
  - Motion/2nd to accept minutes: Kevin / Cat
  - Discussion: None
  - Vote: Abstain / Nay / Yea: Riki / 0 / 16
  - Result: Motion passes

## 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 openings

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 Cytogenetics in the IG Compare Sequence and Observation	Aug 10 - close to co-chair nominations
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 - <a href="#">16789</a> 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 <a href="#">WGM agenda</a> on the <a href="#">WGM information page</a> (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			
9/18/2018	Bob M		
9/25/2018			

## 32<sup>nd</sup> Annual Plenary & Working Group Meeting

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

Draft agenda:

[http://wiki.hl7.org/images/3/35/HL7\\_WGM\\_September2018\\_-\\_Clinical\\_Genomics\\_Agenda.pdf](http://wiki.hl7.org/images/3/35/HL7_WGM_September2018_-_Clinical_Genomics_Agenda.pdf)

### 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-IFkKAcKpl6IHgPaPQEgA/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/13sSChUB9rW7v1ep-tZnaDzSWb\\_MyWlvSzEFVS32quE/edit#heading=h.t2adm0gua505](https://docs.google.com/document/d/13sSChUB9rW7v1ep-tZnaDzSWb_MyWlvSzEFVS32quE/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)
  - 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/1azKiQdhAQKuHxAznEp8141FLdFLACIu8MzF2LxADxg/edit#>
    - Draft model docs posted at [https://docs.google.com/document/d/1Wys14HNJAEB\\_YJ-EeDPAKX50\\_oxiDqAKi3WD4wlfjpk/edit](https://docs.google.com/document/d/1Wys14HNJAEB_YJ-EeDPAKX50_oxiDqAKi3WD4wlfjpk/edit)
- FHIR (Gil)
  - [https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB\\_t4sJZ9yXbLMGOqPXHP5tSLLQ/edit#heading=h.zfi9I8jfe4Ia](https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHP5tSLLQ/edit#heading=h.zfi9I8jfe4Ia)
    - Discussed the “genomics primer” section trackers, started to consider simplification request of the impacts profiles
    - (no call this week - Labor Day)

# Topic 1: OO PSS - Digital Pathology

Riki / OO (presenting)

IHE Digital Pathology Workflow Metadata Requirements (e.g. DICOM) to Specimen DAM mapping and HL7 product family use

Co-sponsor Work Group(s)  
(Enter co-sponsor approval dates in Section 6.d Project Approval Dates)

Clinical Genomics

The IHE Pathology and Laboratory Medicine (PaLM) domain is working on several workflow profiles to support the evolving field of digital pathology. For background on the specific use cases covered by digital pathology see this white paper requirements wiki page:

[https://wiki.ihe.net/index.php/APW-EDM\\_White\\_Paper](https://wiki.ihe.net/index.php/APW-EDM_White_Paper)

As the first profile the goal is to describe the image acquisition workflow, e.g. ordering a whole slide image from a scanner (acquisition modality) and receiving the scanned image. For the sharing of the image the plan is to leverage DICOM transactions, but for the ordering workflow part, the goal is to use the currently prevalent HL7 v2 messaging.

Scope on the HL7 side is to review and potentially update the Specimen DAM as well as the base v2.x standard where needed to support transactions involved in the image acquisition workflow.

- motion for CG to participate as a co-sponsoring WG on this PSS / 2nd - Bob M / Kevin
- Discussion: Andrea supports
- abstain/nay/yea - none / 0 / 17
- Result: Motion passes

## Topic 2: Block Votes

### Block #1

#### **Comment Submitters**

- Amnon Shabo
- Clement McDonald
- Kevin Power
- Lloyd McKenzie

#### **Line Items**

[16929](#) from+descriptive+to+computable+-+2018-May+Genomics+%2375 ([Amnon Shabo](#)) **In Person** Considered for Future Use

[16789](#)

Discussion+needed+on+change+from+display+names+on+84413-4+-+2018-May+Genomics+%2331 ([Clement McDonald](#)) Not Persuasive with Mod

[16837](#)

Concern+the+spec+is+too+assuming+in+unsolicited+-+2018-May+Genomics+%2344 ([Clement McDonald](#)) Not Persuasive with Mod

[16183](#) Computable+Genetic+Finding+-+Region+name+LOINC+is+not+correct ([Kevin Power](#)) Persuasive

[16763](#)

Comment+to+disagree+that+all+things+**examined+within+range**+have+to+be+represented+in+each+variant+-+2018-May+Genomics+%2324 ([Clement McDonald](#)) Persuasive

[16766](#) 2018-May+Genomics+%2325 ([Clement McDonald](#)) Persuasive with Mod

- motion to accept proposed dispositions / 2nd - Bob D / Patrick
- Discussion: none
- abstain/nay/yea - none / 0 / 17
- Result: Motion passes

### Block #2

#### **Comment Submitters**

- Bob Dolin



## Line Items

[16249](#) Sequence.structureVariant%3A+Add+Sequence.structureVariant.variantType (Bob Dolin) Persuasive

[16246](#) Revise+Sequence+attribute+descriptions (Bob Dolin) Persuasive

[16250](#) Sequence.structureVariant.reportedaCGHRatio (Bob Dolin) Persuasive

[16251](#) Clarify+intent+of+Sequence.structureVariant.precision (Bob Dolin) Persuasive with Mod

- motion to accept proposed dispositions / 2nd - Bob D / Jamie
- Discussion:
  - Kevin: given our current state of understanding the difference between Sequence and Observation profiles, I will vote no on changes to Sequence. I will note these are fairly minor changes, so was OK proposing the changes due to feedback from others in the group and the agreement from the submitter.
  - Bob D: minor clarifications regarding attributes in the sequence resource
  - Lloyd: can discuss what should be captured first, then decide where to put it
- abstain/nay/yea - none / Kevin / 16
- Result: Motion passes

## Topic 3: Discuss tracker 15889: Where to add Human Reference Sequence Assembly component

[15889](#) Properties+needed+for+range+examined+and+human+reference+sequence+assembly (Lloyd McKenzie) Persuasive with Mod  
Here are the comments on the tracker.

<p>Submitted on Thu, 30 Aug 2018 22:40:54 +0000 by</p> <p><a href="#">Bob Dolin</a></p> <p>Is there a need to state a positional location for a haplotype (as is possible in VMC)? If so, in addition to adding component "Human reference sequence assembly version: 62374-4" into Computable Genetic Finding, we may also need to add component "Range(s) of DNA sequence examined 51959-5".</p>
<p>Submitted on Thu, 30 Aug 2018 22:49:44 +0000 by</p> <p><a href="#">Kevin Power</a></p> <p>If we add the 'range' component to Computable Genetic Finding, that would make it applicable for any genetic finding (not only Haplotype, but also Genotype (OK?), DescribedVariant (could cause confusion?), ComplexVariant (not sure?), etc ...)</p>
<p>Submitted on Thu, 30 Aug 2018 23:03:43 +0000 by</p> <p><a href="#">Bob Dolin</a></p> <p>One alternative is to just add Genome Build into Described Variant for now. But if we do want to be able to assert a location for a haplotype, I agree we might cause confusion with the positional components in Described Variant. Maybe we could remove LOINC 81254-5 allele start end from Described Variant, and replace it with a more generic "genomic interval" in Computable Genetic Finding. Probably would also then want to move coordinate system up into Computable Genetic Finding.</p>

### Discussion

- We should support positional coordinates for haplotypes, still not sure of how (yet)
  - Will take off line and craft a concrete proposal

## Topic 4: Compare Sequence and Observation

[https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg\\_xpwir8rEORkNjMfemvqxE0/edit#gid=0](https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg_xpwir8rEORkNjMfemvqxE0/edit#gid=0)

Will be discussed on a future call

## Chat

- Kevin Power
  - Hi all - Here is the zulip chat / request for feedback on the options for "Region's Studied" - if anyone would like to join the straw vote or add a comment, please do:  
<https://chat.fhir.org/#narrow/stream/43-genomics/subject/Capturing.20Genomic.20Panel.20Definitions/near/181411>
- Kevin Power
  - <http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html>

### ***Clinical Genomics Docs***

- SWOT
  - [https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS\\_JiScDACBi13tzFJep751k/edit](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/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srIde0EBs0FI/edit>
  - Review complete as of Aug 15, 2017
  - Approved in Sep 2017 WGM in San Diego
- DAM
  - <http://tinyurl.com/damcgdoc>



