

## HL7 Clinical Genomics Weekly Call - August 14, 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)

### **Attending the meeting:**

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 1: Block Vote #1](#)

[Topic 2: Cytogenetics in the IG](#)

[Topic 3: What was tested?](#)

[Topic 4: Impact/Interp](#)

[Topic 5: New LOINC codes?](#)

[Topic 6: Compare Sequence and Observation](#)

[Chat](#)

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

## Attendees Sign-in

(Presiding co-chair: Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org))

1. Andrea Pitkus - [apitkus@gmail.com](mailto:apitkus@gmail.com)
2. Caterina Lasome - iON Informatics for AFMS - [cat@ioninformatics.com](mailto:cat@ioninformatics.com)
3. Bob Freimuth - Mayo Clinic - [freimuth.robert@mayo.edu](mailto:freimuth.robert@mayo.edu)
4. JD Nolen - Children's Mercy Hospital - [jlnolen@cmh.edu](mailto:jlnolen@cmh.edu)
5. Patrick Werner - Molit Institute / Heilbronn University - [patrick.werner@molit.eu](mailto:patrick.werner@molit.eu)
6. Dora Finkeisen - MOLIT Institute - [dora.finkeisen@molit.eu](mailto:dora.finkeisen@molit.eu)
7. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
8. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
9. Mullai Murugan - BCM - [murugan@bcm.edu](mailto:murugan@bcm.edu)
10. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
11. Joel Schneider - NMDP/CIBMTR - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
12. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
13. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
14. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
15. Julian Sass - Berlin Institute of Health - [julian.sass@bihealth.de](mailto:julian.sass@bihealth.de)
16. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
17. Jamie Parker - Carradora Health - [jamie.parker@carradora.com](mailto:jamie.parker@carradora.com)
18. Deepak Sharma - Mayo Clinic - [sharma.deepak2@mayo.edu](mailto:sharma.deepak2@mayo.edu)
19. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)

## Minutes Approval

- August 7
  - [http://wiki.hl7.org/index.php?title=File:HL7\\_CG\\_20180807.pdf](http://wiki.hl7.org/index.php?title=File:HL7_CG_20180807.pdf)
  - Motion/2nd to accept minutes: Bob F/ Joseph Kane
  - Discussion: None
  - Vote: Abstain / Nay / Yea: 0 / 0 / 18
  - Result: 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 rebalot 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	July 11 - Call for co-chair nominations July 15 - Notification of Intent to Ballot

		NIB vote	
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			Aug 24 - ballot opens for voting
8/28/2018	Bob M		Aug 31: Deadline to post WGM agenda on the <a href="#">WGM information page</a> (WG Health metric)
9/4/2018	Bob F		
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

### 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) and we are extending the model to support complex variants (e.g., fuzzy ends) (lead by Larry Babb/Tristan Nelson)
  - Variant Annotation
    - Collecting use cases, prioritizing for modeling work
- DIGITiZe (aka National Academies) (Grant Wood, JD Nolen)
  -
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
  -
- CDISC PGx (Dorina B.)
  -
- 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/1azKiQdhAQKuHhxAzEp8141FLdFLAClu8MzF2LxADxg/edit#>
    - Draft model docs to be posted 8/16/18 (please review and submit feedback)
- FHIR (Gil)
  - [https://docs.google.com/document/d/1FGCQRtxJKyHhnc1uB\\_t4sJZ9yXbLMGOqPXHPPr5tSLLQ/edit#heading=h.zfi9l8jfe4la](https://docs.google.com/document/d/1FGCQRtxJKyHhnc1uB_t4sJZ9yXbLMGOqPXHPPr5tSLLQ/edit#heading=h.zfi9l8jfe4la)
    - Updated various trackers and discussed “What was tested?”

## Topic 1: Block Vote #1

Various trackers related to our “Variant Grouping” discussion.

### Comment Submitters

- Bob Milius
- Kevin Power
- Lloyd McKenzie
- Xin Liu

### Line Items

- [15890](#) Additional+properties+of+Complex+variant+needed (Lloyd McKenzie) Not Persuasive
- [\\*15885](#) Should+consider+how+the+PhaseSet+match+to+the+IG+structure (Xin Liu) Persuasive with Mod
- [\\*16173](#) Clarify+usage+of+Genotype%2FHaplotype%2FSequenceConfiguration+or+remove+for+now (Kevin Power) In Person Persuasive with Mod
- [\\*16472](#) clarify+allelic-phase (Bob Milius) Persuasive with Mod
- [\\*16496](#) phase+set+of+sequences+%28not+variants%29 (Bob Milius) In Person Persuasive with Mod
- [\\*16512](#) Sequence+Configuration+cardinality (Bob Milius) Persuasive with Mod

### \*Mod from [16173](#)

- “Sequence Configuration”
  - Rename to “Sequence Phase Relationship”
- “Sequence Phase Relationship”.focus:
  - Change cardinality to 0..\* - Supports more than two ‘in-cis’ findings - with appropriate cautions about indicating multiple ‘in-trans’ findings
  - Make Sequence a valid target - Support HLA and potentially other use cases
- “Sequence Phase Relationship”.method:
  - Use same LOINC as allelic phase basis
  - Required Pattern: {"coding":{"system":"<http://loinc.org>","code":"82309-6"}}
  - Binding: [LOINC Answer List LL4050-2](#) (preferred)
- Haplotype.method:
  - Use same LOINC as allelic phase basis
  - Required Pattern: {"coding":{"system":"<http://loinc.org>","code":"82309-6"}}
  - Binding: [LOINC Answer List LL4050-2](#) (preferred)
- DescribedVariant.profile:
  - Remove “allelic phase” component - {"coding":{"system":"<http://loinc.org>","code":"82120-7"}}

- Remove “allelic phase basis” component -  

```
{"coding":{"system":"http://loinc.org","code":"82309-6"}}
```
- change "required" to "preferred" for loinc answer list for method
  - Motion/2nd to accept block: Lloyd/ Bob D
  - Discussion: none
  - Vote: Abstain / Nay / Yea: 0 / 0 / 18
  - Result: passes

## Topic 2: Cytogenetics in the IG

- from last week: Clem will finalize this proposal and share with the group. Kevin will then move the proposal into the appropriate ballot comments.
  - sent to Kevin last night
  - will be sent to listserv
- Block to be sent:
  - [https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemBrowse&tracker\\_id=677&querynav=%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemBrowse%26tracker\\_id%3D677%26forget\\_query%3D1&quickquery=1&tracker\\_item\\_id=&summary=&submitted\\_by=&priority=&assigned\\_to=&extra\\_field%5B4214%5D=&extra\\_field%5B4215%5D=&extra\\_field%5B4060%5D=&extra\\_field%5B3631%5D=&extra\\_field%5B3807%5D=&extra\\_field%5B3808%5D=&extra\\_field%5B3628%5D=13106&extra\\_field%5B3626%5D=&extra\\_field%5B4065%5D=&extra\\_field%5B4092%5D=&extra\\_field%5B4063%5D=18940&extra\\_field%5B4062%5D=&extra\\_field%5B2415%5D=&extra\\_field%5B4252%5D=&extra\\_field%5B3633%5D=&extra\\_field%5B3969%5D=&extra\\_field%5B4069%5D=&extra\\_field%5B4066%5D=&extra\\_field%5B4071%5D=&extra\\_field%5B3632%5D=&sortcol=last\\_modified\\_date&sortord=ASC](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemBrowse&tracker_id=677&querynav=%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemBrowse%26tracker_id%3D677%26forget_query%3D1&quickquery=1&tracker_item_id=&summary=&submitted_by=&priority=&assigned_to=&extra_field%5B4214%5D=&extra_field%5B4215%5D=&extra_field%5B4060%5D=&extra_field%5B3631%5D=&extra_field%5B3807%5D=&extra_field%5B3808%5D=&extra_field%5B3628%5D=13106&extra_field%5B3626%5D=&extra_field%5B4065%5D=&extra_field%5B4092%5D=&extra_field%5B4063%5D=18940&extra_field%5B4062%5D=&extra_field%5B2415%5D=&extra_field%5B4252%5D=&extra_field%5B3633%5D=&extra_field%5B3969%5D=&extra_field%5B4069%5D=&extra_field%5B4066%5D=&extra_field%5B4071%5D=&extra_field%5B3632%5D=&sortcol=last_modified_date&sortord=ASC)

## Topic 3: What was tested?

- Set of trackers regarding defining “what was tested”:

### **Comment Submitters**

- Bob Dolin
- Clement McDonald
- Lloyd McKenzie

### **Line Items**

- [15889](#)  
Properties+needed+for+range+examined+and+human+reference+sequence+assembly (Lloyd McKenzie)
- [16258](#) Need+to+address+%22what+was+looked+at%22 (Bob Dolin)
- [16763](#)  
Comment+to+disagree+that+all+things+examined+within+range+have+to+be+represent ed+in+each+variant+-+2018-May+Genomics+%2324 (Clement McDonald)
- See FHIR subgroup minutes for discussion
- See chat.fhir.org
  - <https://chat.fhir.org/#narrow/stream/43-genomics/subject/Capturing.20Genomic.20Panel.20Definitions>
- See email thread to listserv
  - subject: [clingenomics] Region studied, and region quality
- group to catch up with discussions and address later

## Topic 4: Impact/Interp

### Comment Submitters

- Amnon Shabo

### Line Items

[16913](#) Interpretation+vs.+impact+-+2018-May+Genomics+%2369 (Amnon Shabo) In **Person** Considered for Future Use

- [https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker\\_item\\_id=16913](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16913)
- Also, see attached email, and here is an additional response from Amnon:  
*As for interpretation vs. impact, the issue of patient-specific vs. general knowledge is orthogonal in my mind to my ballot comment, namely – there’s (1) the ontological aspect of all those terms like interpretation, impact, relevance, significance, annotation and many more, and there’s (2) the utilization aspect, where indeed the information created under a title like interpretation could be pulled from a knowledgebase without taking into account the full patient context. The latter, by the way, is rather complex to define, i.e., what’s the ‘full’ patient context. I alluded to it in another ballot comment that included the OMIM curated study on the NSCLC patient with two somatic mutations in the EGFR. But that’s just the tip of the iceberg in this regard.*
- Bob M -
  - this was already resolved as "considered for future use" by WG during May 2018 WGM
- Bret -
  - thought we all agreed to figure how to represent knowledge as an artifact



- contextual, vs "this is a pathogenic or not",
- Lloyd -
  - room for both types
  - need for both types
- Clem
  - motion - stay considered for future use, revisit before next ballot
    - Clem/Bret
    - Discussion - none
    - abstain = 0
    - nay = 0
    - yea = 19
    - motion passes

## Topic 5: New LOINC codes?

- Set of trackers regarding the new for new LOINC code, changes to some LOINC codes, and some value set/answer list questions:
- [16262](#) Change+LOINC+answer+list+for+complex+variant+type (Bob Dolin)
  - persuasive with mod
  - Bob D will provide final mod description
  - marked waiting for input
- [16264](#) Modify+dna-chg-type+answer+list (Bob Dolin)
  - Bob D will provide more concrete suggestions
  - marked waiting for input
- [16272](#)
 Revise+answer+list+for+Genotype+Medication+Efficacy+Impact+profile+%28LOINC+51961-1%29 (Bob Dolin)
  - Clem - took from publication received from Bob Freimuth
  - Bob F - answer list did not come from CPIC, and CPIC doesn't have one similar that we should use
  - Andrea - need to be careful about changing anything CLIA requires re answer lists
  - Bob D will resend Shannon's email about this
  - marked waiting for input
- [16180](#) Inherited+Disease+Pathogenicity+-+Must+have+mode-of-inheritance+value+set (Kevin Power)
- [16183](#) Computable+Genetic+Finding+-+Region+name+LOINC+is+not+correct (Kevin Power)
- [16184](#) Genetic+Impact+-+Need+LOINC+code+for+level+of+evidence (Kevin Power)
- [16239](#) Need+an+Observation.code+for+DescribedVariant (Kevin Power)

- [16242](#)  
DescribeVariant.component%28Simple+var+ID%29+LOINC+code+is+for+%22Discrete%22+Variant (Kevin Power)
- [16244](#) Need+a+LOINC+for+Coordinate+System (Kevin Power)
- discussed three issues, see above ([16262](#), [16264](#), [16272](#))

## Topic 6: 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)

- not discussed

## Chat

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### ***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/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit>
  - Review complete as of Aug 15, 2017
  - Approved in Sep 2017 WGM in San Diego
- DAM
  - <http://tinyurl.com/damcgdoc>