

## HL7 Clinical Genomics Weekly Call - July 11, 2017 11:00 AM (US Eastern Time)

### **Agenda**

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### **Attendees**

1. Bob Milius - NMDP - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
2. Andrew Brown - NMDP - [abrown3@nmdp.org](mailto:abrown3@nmdp.org)
3. Shannon Lu- NLM - [shannon.lu@nih.gov](mailto:shannon.lu@nih.gov)
4. Bob Dolin - [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
5. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
6. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
7. Jeremy Warner - Vanderbilt - [jeremy.warner@vanderbilt.edu](mailto:jeremy.warner@vanderbilt.edu)
8. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
9. Gil Alterovitz - HMS/BCH- [gilusa@gmail.com](mailto:gilusa@gmail.com)
10. Scott Robertson - Kaiser Permanente - [scott.m.robertson@kp.org](mailto:scott.m.robertson@kp.org)
11. Bob Freimuth - Mayo Clinic - [freimuth.robert@mayo.edu](mailto:freimuth.robert@mayo.edu)
12. Amnon Ptashke - Edico Genome - [genptashke@gmail.com](mailto:genptashke@gmail.com)
13. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
14. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
15. Elizabeth Newton - Kaiser Permanente - [elizabeth.h.newton@kp.org](mailto:elizabeth.h.newton@kp.org)

## Minutes Approval

- June 27
  - [http://wiki.hl7.org/index.php?title=File:HL7\\_CG\\_20170627.pdf](http://wiki.hl7.org/index.php?title=File:HL7_CG_20170627.pdf)
    -
  - motion/2nd - Clem/Bob D
  - abstain - Scott R/Jeremy/
  - nay - 0
  - yea - rest (12)

- result - motion passes

## Topics to discuss if needed

### Upcoming agendas until San Diego

- ~~May 30 WGM recap, SWOT~~
- ~~Jun 6 Finish WGM recap, SWOT, Pedigree Ballot any comments?~~
- ~~Jun 13 FHIR Connectathon, Status of LRI Ballot~~
- ~~Jun 20 SWOT~~
- ~~Jun 27 CLIA & CAP requirements Unified view (Andrea, Clem, Gil needs to be present), LRI/V2 cont, SWOT~~
- ~~Jul 4 US Holiday~~
- Jul 11 - FHIR motion for changes to current build, SWOT, V2/LRI update?
- Jul 18 -
- Jul 25 - DAM update - still on - Gil
- Aug 1 - Clem away
- Aug 8 - WGM schedule draft
- Aug 15 - Gil may be gone
- Aug 22 - Clem, Bob M and Bob F out (eclipse!)
- Aug 29 - Clem gone
- Sep 5 -
- Sep 9-17 - WGM San Diego

### Brief reports from external efforts

- GA4GH
  - no update
- National Academies
  - no update
- Clingen/Clinvar
  - Working on sync'g submission IDs between ClinVar and the ClinGen Allele Registry
- Variant Modelling Collaboration (**VMC**)
  - Yesterday's call was canceled, will pick up next week. Working on finalizing the spec and should be able to release publicly (for comment) within a few weeks.

### Subgroup reports

- IM
  - [https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq\\_vAT9B-Z8GI\\_AyRiM/edit](https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq_vAT9B-Z8GI_AyRiM/edit)
  - Not meeting during July - sync'g schedule with VMC, will pick up again after public release
- FHIR
  - [https://docs.google.com/document/d/1FGCQRtxJKyHhnc1uB\\_t4sJZ9yXbLMGOqPXHPPr5tSLLQ/edit#heading=h.nts1cfujf9t5](https://docs.google.com/document/d/1FGCQRtxJKyHhnc1uB_t4sJZ9yXbLMGOqPXHPPr5tSLLQ/edit#heading=h.nts1cfujf9t5)
  - See schedule at bottom of above link (feel free to add topics to calendar/email Gil)

- "FHIR recommended to put the Sync for Genes suggestions into current build. These were presented at the wgm and two past FHIR subgroup meetings. You can view the changes here on the notes links of the FHIR subgroup."
- working on V2->FHIR mapping
  - planning translation application being developed, possibly javascript
- We will block vote on changes today (see below).
- [https://drive.google.com/file/d/0B-0YtCs\\_i\\_-eMnB6OUJrY05HMGs/view](https://drive.google.com/file/d/0B-0YtCs_i_-eMnB6OUJrY05HMGs/view)
- 
- LRI/V2
  - see below

## Topic 1: FHIR

- FHIR motion to accept changes proposed by Sync4Genes work
  - [https://drive.google.com/file/d/0B-0YtCs\\_i\\_-eMnB6OUJrY05HMGs/view](https://drive.google.com/file/d/0B-0YtCs_i_-eMnB6OUJrY05HMGs/view)
  - Motion/2nd - Gil/Clem
  - discussion - none
  - abstain - Joel, Bob F, Scott R.,
  - nay - 0
  - yea - rest (11)
  - result - motion passes
- Minutes
  - [https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB\\_t4sJZ9yXbLMGOqPXHPt5tSLLQ/edit#heading=h.nts1cfufj9t5](https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPt5tSLLQ/edit#heading=h.nts1cfufj9t5)
- Jeremy - how long before put into current build
  - Gil - try to be ready by this Thu for FHIR subgroup mtg
- guidance.site is where the S4G work was done

## Topic 2: LRI/V2 - status (cont)

- "V2 will be summarizing what's been happening with the ballot reconciliation for LRI. Attached is the ballot reconciliation comments. I hope we'll also have time to discuss the comments that still need to be reconciled – which are from:
  - Andrea Pitkus
  - Nell Lapres"
- [http://www.hl7.org/documentcenter/public/wg/clingenomics/2017%2006%2007%20-%20V251\\_IG\\_LRI\\_R1\\_D4\\_2017MAY\\_Consolidated%20-%20SL.xlsx](http://www.hl7.org/documentcenter/public/wg/clingenomics/2017%2006%2007%20-%20V251_IG_LRI_R1_D4_2017MAY_Consolidated%20-%20SL.xlsx)
- Clem - two comments outstanding
  - adding to table coding systems for HLA and star alleles
  - Shannon will send draft to Bob and Joel
  - Joel and Clem/Shannon will connect on FHIR terminology service for HLA
  - Bob F will meet with Clem/Shannon to talk about star alleles

## Topic 3: CG Documents to review

- SWOT
  - [https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS\\_JiScDACBi13tzFJep751k/edit](https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit)
  - short url = <http://bit.ly/2ikUw0>
  - Continued review of the SWOT document, beginning with Opportunities (edits made directly in document, not captured here)
- 

## Clinical Genomics Docs

- SWOT
  - [https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS\\_JiScDACBi13tzFJep751k/edit](https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit)
  - short url = <http://bit.ly/2ikUw0>
  - worked on Opportunities, three more to do
- Decision Making Process
  - <https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit>
  - short url = <http://bit.ly/2ikjXiV>
  - nothing new

## Chat