

## HL7 Clinical Genomics Weekly Call - June 27, 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. Andrea Pitkus - IMO - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
3. Andrew Brown - NMDP - [abrown3@nmdp.org](mailto:abrown3@nmdp.org)
4. Bob Dolin - [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
5. Amnon Ptashke - Edico Genome - [genptashke@gmail.com](mailto:genptashke@gmail.com)
6. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
7. Amnon Shabo (Shvo) - Philips - [amnon.shvo@gmail.com](mailto:amnon.shvo@gmail.com)
8. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
9. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
10. Ling teng -BCH -[tenglingling@gmail.com](mailto:tenglingling@gmail.com)
11. Shannon Lu - NLM - [shannon.lu@nih.gov](mailto:shannon.lu@nih.gov)
12. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
13. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
14. Caterina Lasome - AFMS - [cat@ioninformatics.com](mailto:cat@ioninformatics.com)
15. Gil Alterovitz - HMS/BCH- [gilusa@gmail.com](mailto:gilusa@gmail.com)
16. Grant Wood - Intermountain
17. Larry Babb
18. lushenno
19. serafina.versaggi

## Minutes Approval

- June 13
  - [http://wiki.hl7.org/index.php?title=File:HL7\\_CG\\_20170620.pdf](http://wiki.hl7.org/index.php?title=File:HL7_CG_20170620.pdf)
  - motion/2nd - Bob Dolin/Andrew B

- abstain - 0
- nay - 0
- yea - remaining (13)
- result - passed

## 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 -
- Jul 18
- Jul 25 - DAM update?
- Aug 1
- Aug 8
- Aug 15
- Aug 22
- Aug 29
- Sep 5
- Sep 9-17 - WGM San Diego

### Brief reports from external efforts

- GA4GH
  -
- National Academies
  -
- Clingen/Clinvar
  -
- Variant Modelling Collaboration (**VMC**)
  -

### 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)
  -
- 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.
- We will block vote on changes next cg meeting."
  - [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: CAP/CLIA requirements review

- see additional material found in [http://wiki.hl7.org/images/b/b5/HL7\\_CG\\_20170613.pdf](http://wiki.hl7.org/images/b/b5/HL7_CG_20170613.pdf)
- decided to wait until Gil is available

## 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)
- Discussion about HLA nomenclature and version numbering of IMGT/HLA database
- Clem will rewrite and send to Bob M for comment.

## 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/2iklUw0>
  - Continued review of the SWOT document, beginning with Opportunities (edits made directly in document, not captured here)
  - No time for discussion

## 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/2iklUw0>
  - 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

- From Bob M to Everyone: (10:27 AM)
  - <http://www.ebi.ac.uk/ipd/imgt/hla/allele.html>
- From Larry Babb to Everyone: (10:28 AM)
  - To add on. Does this mean that this coding system for “star alleles” is going to be kept in sync with CPIC/PharmGKB published standards?
  - And is there some intended specific meaning for each star allele code? Because not every star allele (with the same code) represents the same set of discrete variants.
  - i think star alleles are like “aliases” and are something that folks want to get away from ultimately.
  - i agree with the “not enabling bad behavior”
- From Amnon Shvo to Everyone: (10:30 AM)
  - @Larry - agreed.
- From Larry Babb to Everyone: (10:31 AM)
  - just because folks do it, doesn't mean we should support it. (i think)
- From Alex Mankovich to Everyone: (10:32 AM)
  - unfortunately it seems even NGS-based automated HLA typing tools primarily use star alleles
- From Bob Dolin to Everyone: (10:33 AM)
  - So, we can't rely on star alleles at PharmGKB site?
  - (that they mean the same thing over time, and that they are definitive)?
- From Andrea Pitkus, PhD, MLS(ASCP)CM to Everyone: (10:37 AM)
  - Overall, it's an issue with medical knowledge. That is, it changes, and changes quickly. Versioning helps us track the state of knowledge like the alleles at a set period of time.
- From Andrea Pitkus, PhD, MLS(ASCP)CM to Everyone: (10:37 AM)

- The larger question is how the knowledge gained is tracked by comparing genetic results from any point to an updated reference set and tracking.
- updated results, interpretations and clinical decision making based upon the new knowledge
- From Bret Heale to Everyone: (10:43 AM)
  - The scope here is the HL7 v2 LRI. I think that in FHIR we should strive for a clearer (i.e. position and reference) description of a change.
- From Larry Babb to Everyone: (10:55 AM)
  - star allele definitions are not the same when reported by genetics testing labs.
- From Larry Babb to Everyone: (10:55 AM)
  - genetics testing labs only test 1 2 or 3 of the locations amongst N locations that define the star allele. Plus new locations are added over time.
  - so one lab's \*1 is not equal to another labs \*1
  - @Bret - agree