

## HL7 Clinical Genomics Weekly Call - Jan 26, 2016

### Attendees

- Bob Milius - NMDP - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
- Amnon Shabo (Shvo) - Philips - [amnon.shvo@gmail.com](mailto:amnon.shvo@gmail.com)
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- Jennifer D. Davis (Accenture) - [jennifer.d.davis@accenture.com](mailto:jennifer.d.davis@accenture.com)
- Bret Heale (University Utah/Intermountain Healthcare) - [bheale@gmail.com](mailto:bheale@gmail.com)
- Kevin Power (Cerner) - [kpower@cerner.com](mailto:kpower@cerner.com)
- Perry Mar (Partners HealthCare System) - [pmar@partners.org](mailto:pmar@partners.org)
- Grant Wood - Intermountain -
- Siew Lam - Intermountain -
- M'Lynda Owens - Cognosante - [mlynda.owens@cognosante.com](mailto:mlynda.owens@cognosante.com)
- Jonathan Holt (SeqTech Diagnostics) [jholt@seqtechdx.com](mailto:jholt@seqtechdx.com)
- Joseph Kane (Epic) - [jkane@epic.com](mailto:jkane@epic.com)
- Heming Yao - BCH
- Huanqin Dai - BCH
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- Larry Babb (Partners/GeneInsight) - [lbabb@geneinsight.com](mailto:lbabb@geneinsight.com)
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- Terry McDonnell (Syapse) - [terrym@syapse.com](mailto:terrym@syapse.com)
- Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
- Brett Johnson
- Clem McDonald - NLM -
- Gil Alterovitz - BCH -
- Yogy

### Discussion

- Orlando WGM minutes approval
  - Please take a look at the minutes posted here:
  - [http://wiki.hl7.org/index.php?title=CG\\_Working\\_Group\\_Meeting\\_Minutes#HL7\\_WGM\\_Orlando\\_January\\_2016](http://wiki.hl7.org/index.php?title=CG_Working_Group_Meeting_Minutes#HL7_WGM_Orlando_January_2016)
  - Please note that there have been a few edits on the minutes since I sent out the link last week.
  - Motion to accept - Bret
  - 2nd - Amnon
  - discussion - none
  - abstains - M'Lynda
  - motion - passed
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- 5 min updates
  - ClinGen

- GA4GH
  - grant - worked with them on ehealth - working forward consent model on family history, call this regarding this
- IOM
  - grant - moving forward with digitize - two new work groups - use case wg, current use case is pharmacogenomics- will be looking into new use cases; another work group is developing pilots, hoping pilot teams will self form and inform WG. Hope to start pilots in March.
- BRIDG
  - Bob M - molecular biology part of BRIDG model is not in scope for May ballot
- FHIR Lead
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- other
- FHIR Connectathon Recap
  - very successful in qualitative and quantitative measure
  - remote connection
  - before and after survey
  - feedback will be added to google document
  - website: fhingenomics.org
  - FHIR subgroup being formed - prep for STU, july deadline with Sep ballot
  - will have doodle poll for scheduling subgroup
  - request for no earlier than 11am eastern time for west coast,
  - Bob M - thinks subgroup should be focussed on vetting technical specification, and preparation for next connectathon and ballot
  - Gil needs feedback on use cases
  - Clem has technical feedback sequence resource, he will summarize and send to Gill for discussion in subgroup
  - Clem's example, writing-up a use case and sending to Gil for entry into Sub-group discussions, is the preferred method of communication to our CG FHIR sub-group(?)
  - Exploring participation Clinician Connectathon (occurs at end of WGM)
    - Gay: Learning level in group is focused on patient/observation/search
  - We need to determine best way of communication for the FHIR subgroup (email, skype, slack, etc). A doodle poll will be used to determine best day/time and also to identify members for an initial email list.
  - Motion to submit CG track for May Technical FHIR Connectathon in Montreal - motion by Gil, seconded by Grant -
    - discussion
      - Bret asks
        - what is "Technical Connectathon?"
          - A: event just prior to the WGM that is focused on implementers - those creating clients and servers,

programmers. In contrast, the “Clinician Connectathon” is focused on clinicians, not programmers. The recent Connectathon that we participated in Orlando was a Technical Connectathon.

- also asks “Will we vote on the use cases?”,
  - Gil - we need to present something for the proposal, we should be able add/change later
  - Jon - wants to flesh out technical details before we commit to have another Technical Connectathon
  - Bob - sees the Connectathon as an opportunity to test use cases that weren’t vetted, or test new things in the specification.
  - Gil - also an opportunity for different people to try out things they haven’t tried with current use cases. Some may have done some use cases, but not others.
- Vote
  - abstains - none
  - nays - none
  - motions carries
- Orlando WGM Recap:
  - Gil Alterovitz has been elected to another term as co-chair. Congratulations Gil!
  - We are forming a new subgroup led by Gil that will be focused on FHIR. In addition to CG, an invitation will be extended to the Orders and Observations group to participate. The results of this subgroup will be reported to the larger group during our weekly call.
  - CG will become primary stakeholder for the HL7 V2 Genomics Report Lite being developed by Clem McDonald.
    - rethinking this based on feedback, wants to connect to Dan from EPIC
    - wants to makes sure it’s compatible with FHIR
    - Joseph Cane - epic will make proposal for modification for CG group in two weeks
    - Clem is rethinking this
    - Clem will talk to Lloyd about FHIR restrictions.
  - We will be producing a CG WG position statement/document regarding variation representations in clinical reporting. HGVS will be an important discussion including when it is appropriate and when it is not, as well as its proper use. A PSS will be prepared by Bob Freimuth.
    - Larry Babb - wants to include ClinGen/ClinVar in these discussions, they have a lot of expertise and deep thinking about this
    - Bob F and Larry Babb working on how to use fhir spec for submitting to ClinVar
    -

- We will re-evaluate and consolidate relevant elements from the different DAMs that this WG has produced. Gil will lead this effort.
- The DIM will be broadened to a higher level (fewer if any attributes) so that it can be more easily harmonized with the DAM, as well as identifying sub-domain defined by specific use cases. Amnon Shabo will lead this.
- A glossary (at minimum) will be developed in close collaboration with the DAM/DIM work so semantic harmonization can be maintained across all of our products.
- DAM/DIM discussion
  - Mollie has made a motion: "I motion Gil be added as project leader/facilitator to the 2 DAM projects. He and I have been working closely together for over a year."
  - We will get clarification for exactly which projects, and I will send the a copy or link of the PSS's to the group.
  - Ran out of time to discuss this. Motion tabled to next week.
- FHIR Connectathon
  - From Lloyd: *"At the WGM, the FMG announced a deadline of Feb. 7th for resource/profile proposals and connectathon track proposals. Because we've shifted the date for the ballot, the 7th will \*not\* be the due date for resource or profile proposals. That deadline will be some point shortly after the May WGM.*

*However, Connectathon proposals are still due on the 7th. This is true even for proposals that were previously submitted for the Orlando connectathon. As well, this Sunday is still the target for fixing validation issues that are going to be escalated to "Warnings" for resources that are already > FMM 0. (On Sunday we'll break the build.)*

*As usual, if there are questions or concerns, feel free to contact me or your FMG rep."*

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- NIST task cases for ELR are out.