## HL7 Clinical Genomics Weekly Call - April 19, 2016

## **Attendees**

- 1. Bob Milius NMDP bmilius@nmdp.org
- 2. Gaston Fiore BCH gaston.fiore@gmail.com
- 3. Jonathan Holt SeqTechDx jholt@seqtechdx.com
- 4. Jeremy Warner Vanderbilt <u>jeremy.warner@vanderbilt.edu</u>
- 5. Mollie Ullman-Cullere mollie.ullmancullere@gmail.com
- 6. Perry Mar Partners HealthCare System pmar@partners.org
- 7. David Kreda <a href="mailto:david.kreda@gmail.com">david.kreda@gmail.com</a>
- 8. Kevin Power Cerner kpower@cerner.com
- 9. Bob Freimuth Mayo Clinic freimuth . robert at mayo . edu
- 10. Jami Deckard Regenstrief Institute jkdeckar@regenstrief.org
- 11. Brett johnson icanbrj@gmail.com
- 12. Heming Yao <a href="mailto:yhm@mail.ustc.edu.cn">yhmyhm@mail.ustc.edu.cn</a>
- 13. Anwaruddin Mohammad Oracle <a href="mailto:anwaruddin.mohammad@oracle.com">anwaruddin.mohammad@oracle.com</a>
- 14. Swapna Abhyankar Regenstrief Institute <a href="mailto:sabhyank@regenstrief.org">sabhyank@regenstrief.org</a>
- 15. Siew Lam Intermountain Healthcare siew.lam@imail.org
- 16. Larry Babb GeneInsight/Sunquest <a href="mailto:lbabb@geneinsight.com">lbabb@geneinsight.com</a>
- 17. Joel Schneider NMDP jschneid@nmdp.org
- 18. Andrea Pitkus IMO- apitkus@imo-online.com
- 19. Clem McDonald NLM
- 20. Eric Whitebay
- 21. JD Nolen Cerner
- 22. Grant Wood Intermountain
- 23. Joseph Kane

## **Discussion**

- Minutes approval
  - Please take a look at the draft minutes posted here (fyi, I posted a PDF that was exported from the google doc)
  - http://wiki.hl7.org/index.php?title=File:HL7 CG 20160412.pdf
  - o Motion to accept Grant Wood
  - o 2nd Jonathan Holt
  - Discussion Presiding co-chair will be designated for each session at the WGM.
     Responsible for making sure minutes are recorded (either themselves, or designating a scribe). Discussion leads will be assigned, who may or may not be the presiding co-chair.
     For example, Clem, while not being a co-chair, should lead the discussion on V2 lite.
  - yea/nay/abstain = rest / 0 / Jeremy, Bob F, Jami Deckard
  - o results motion passes
- Brief updates
  - o ClinGen/ClinVar -
    - Larry nothing new
  - o GA4GH -
    - Conference was held last Friday in New Orleans, a new effort was announced (the Actionable Clinical Genome Initiative). Details TBD.

- IOM (in the future refer to National Academies) -
  - Grant & JD Nolen Nothing new, work is continuing, pilot teams still forming
  - Name changed from IOM to National Academy of Medicine
  - Health and Medicine Division this changed as well
  - Part of the National Academies of Science, Engineering and Medicine
- FHIR -
  - Connectathon coming up
  - Clinicians on FHIR will have their own connectathon
  - Gil will be giving a tutorial on Thu AM
- o other
  - nothing
- Formation of a DIM subgroup
  - What would be the goals/tasks/deliverables a DIM subgroup? (jon)
  - Bob F meet in parallel that would allow us to move forward on work that has been on the back burner for a while. Amnon has been working on a DIM, but hasn't been published for informative ballot. Find a common threads that will lay down semantic foundation between V2, CDA, FHIR. Have a dedicate subgroup that will be actively working on this. Haven't decided any specifics on leadership or structure yet. Keep focus tight, technology agnostic model. Start at middle and model more generally or more specifically as needs arise. Bring back work to the WG for review.
  - David K -
    - Q1 mini-DIM brought up at last WGM any disposition on that?
      - Bob F no preconception on this; have worked on modeling for a long time and have opinions on how this should be done. If smaller domains need to be address, we can work on it as need dictates. Motivation is that we have a variety of artifacts developed over the last few years. We want to be coherent among these.
    - Q2 V3?
      - Bob F no special attachment to V3, but it's one of the technologies that HL7 supports, so we should too. V2 is most important to support backward compatibility
  - Mollie DIM was reviewed and comments submitted. We need reconcile the comments. We can't submit for ballot projects that are not successful and not respond.
  - Bob F. as a starting point, we would leverage what has already been done. Everything we
    have done should be on the table. Not sure of the process to reconcile the earlier ballot.
  - o Mollie we have the WGM to work this out
  - Perry Mar DIM was V3, used to create a model for V3 messages, we should re-use this
    - Bob M: DIM vs DIMM (or DMIM?)
    - DMIM (DIM) "Domain Message Information Model" shows all classes and relationships used in a certain domain (pharmacy, laboratory, public health, patient administration).
      - DMIM: Provides a single common reference model for all messages in one domain
    - RMIM (SIM) "Refined Message Information Model" defines classes and relationships for a restricted set of interactions.
    - Added after meeting:
      - http://wiki.hl7.org/index.php?title=Domain Message Information Model

- "D-MIMs (or DIMs in HDF speak) are Domain models representing a committee's (or group of committee's) understanding of the "data" and relationships that are relevant within the area of clinical or administrative knowledge represented by that domain expressed in HL7 modeling terms. DIMs should represent the majority, or preferably all, of the static requirements documented within a committee's DAMs (Domain Analysis Model) expressed in RIM modeling terms. All DIMs must be a proper constraint on the HL7 RIM."
- o Jon CG DIM balloted in 2014
- Clem beauty of FHIR is that it uses an implicit model, avoids general model
- o Jon we're still struggling with what the FHIR sequence resource is conceptually.
- Bob F we have had passionate discussion on various concepts in the domain. If we went around to everyone on this call to define various terms (e.g., sequence, allele, variant, genotype, etc) in our domain we probably would get different answers.
- Jon FHIR is major motivation; We have urgency to get it done soon and get it done right.
   The DIM would help define the conceptual model to inform FHIR.
- David K FHIR Implementation guide can serve as point of the consolidation of passionate debate issues
- Clem we would go long way if just define the things that Bob F mentioned would go a long way.
- Bob F yes, we are NOT trying to boil the ocean. We want to bring together the common concepts used in all our artifacts... come to consensus in big picture concepts.
  - Larry B Why can't we do something like the http://www.hl7.org/fhir/2016May/cqif/cqif.html for Clinical Genomics? Do any of us really think we can fit the needs and concepts for clinical genomics in the existing Resources plus one Sequence Resource?
- Mollie how much bandwidth do we have? Mollie and Gil has worked on consolidating concepts in V2 and FHIR.
  - Larry B V2 was never broadly adopted as far as anyone knows.
- Jon can we add an extra half hour to the FHIR subgroup to create the model? Use FHIR as the focus to work
- David likes this
  - 1 Solve how we use the terms as we do it in FHIR (start concretely)
  - 2 How do they map to V2?
  - 3 Urgency of FHIR and use the spinoff for other technologies
- Bob F conflicted with tieing to FHIR; we really want to be technology agnostic. But we should take this into consideration; need to identify who wants to be involved
- Mollie need to solidify output and balloting process for this.
- Bob F has no problem with formalizing this; continuing with past work and leveraging that.
- Recorded Chat
  - o 10:05:55 AM from Bob Milius to Everyone:
    - minutes recorded here http://bit.ly/1qCMovO
  - o 10:08:38 AM from Bob Milius to Everyone:
    - http://wiki.hl7.org/index.php?title=CG\_Working\_Group\_Meeting\_Agendas
  - o 10:12:21 AM from brett johnson to Bob Milius (privately):
    - thank you Bob-
  - o 10:41:52 AM from Larry Babb to Everyone:

- Hey Bob F., Can we state that the DIM effort you are taking on has the aim to achieve a level of documentation on the kind of level as the http://www.hl7.org/fhir/2016May/cqif/cqif.html
- o 10:41:56 AM from Larry Babb to Everyone:
- o ?
- 10:42:57 AM from Larry Babb to Everyone:
  - It seems that the CQIF group has been afforded the opprotunity to model what they need to deliver and support the needs of Deicision Support standards as defined in earlier HL7 efforts
- o 10:44:01 AM from Larry Babb to Everyone:
  - We have to imagine that Clinical Genomics needs a level of documentation to call out all the data types, components and resources necessary to effectively communicate genomic data now and in the future.
- o 10:44:51 AM from Larry Babb to Everyone:
  - it is my opinion that we cannot strap ourselves down to simply confining our view of the world by whatever FHIR provides plus one Sequence Resource.
- 0 10:45:42 AM from Larry Babb to Everyone:
  - it skews the ability to properly model and convey the information which is potentially wasting our time as well as the community that attempts to adopt these basic constructs in the near term.
- 10:55:27 AM from Perry Mar to Everyone:
  - The model is not just a flat list of words with their definitions. It includes the relationships or associations between the entities in the model, along with constraints and other information. So a flat list of definitions will not accomplish the complete objective, although it is also needed.
- 10:56:57 AM from Larry Babb to Everyone:
  - agreed. Bob F's pending model could be the driver behind the real resources/components needed. just like those that are embedded in the CQIF documentation. I would request that everyone take a look at the links to the components and resources within that framework. Its very compelling.
- o 10:57:22 AM from Perry Mar to Everyone:
  - Regarding the list of definitions, as I mentioned last time, we have a list that we already developed several years ago during the V3 work. Could we refer to that, please?
- o 10:58:02 AM from Larry Babb to Everyone:
  - Many of the V2 LOINC codes are not applicable anymore because they were simply created to drive structure (i.e. panels) in the HL7v2 unstructured world.
- 11:01:18 AM from david kreda to Everyone:
  - But let's not treat definitions here as wholly abstract. They really CAN include references to other terms. Totally normal for, say, contract definitions. The same can apply here. Difference: here we make working definitions in context of FHIR not just for implementation but part of that work ballot. Not same as DIM ballot but not entirely informal either.
- 11:03:05 AM from Perry Mar to Everyone:
  - The DAM also was just mentioned. That is the domain analysis model, which reflects the actual true structure of the domain. In contrast, a DIM, domain information model, shows how you want to represent the information in a form consistent with how you would implement it.