HL7 Clinical Genomics Weekly Call - Dec 13, 2016

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org (presiding co-chair)
2. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
3. Andrea Pitkus -IMO- apitkus@imo-online.com
4. David Kreda - HMS - david.kreda@gmail.com
5. Xin Liu - BCH - xinliu215@gmail.com
6. Shennon Lu - NLM - shennon.lu@nih.gov
7. Jonathan Holt - SeqTechDx - jholt@seqtechdx.com
8. Joseph Kane - Epic - jkane@epic.com
9. Kevin Power - Cerner - kpower@cerner.com
10. JD Nolen - Cerner - johndavid.nolen@cerner.com
11. Gil Alterovitz - BCH -
12. Eric Whitebay
13. Yi Wang - USTC - panzer.wy@gmail.com
14. Bob Freimuth - Mayo Clinic
15. Clem McDonald - NLM
16. Grant Wood - Intermountain
17. Jeremy Warner - Vanderbilt - jeremy.warner@vanderbilt.edu
18. Tianlong Chen - USTC - wiwjp619@gmail.com
19. Joel Schneider - NMDP - jschneid@nmdp.org
20. Xiao Luo - USTC - l.xander.233@gmail.com
21. Perry Mar - Partners HealthCare System - pmar@partners.org
22. Elizabeth Newton
23. Bret Heale - Intermountain - bheale@gmail.com
24. Brett Johnson - VA - icanbrj@gmail.com
25.

Discussion

- **Minutes approval**
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20161206.pdf
    - motion to approve - deferred to next week
    - second -
    - discussion -
    - abstains -
    - nays -
    - yeas -
    - motion -

- **Brief reports from external efforts (discussion only if needed)**
    - GA4GH -
        -
    - National Academies
        - Nothing to update - JD
    - Clingen/Clinvar
        -
    - GA4GH Variant Modeling Collaborative (VMC)

        -
    - NHGRI
        -
- **Upcoming calls through next WGM in San Antonio**
    - Dec 13 - (today) Amnon presenting his FHIR proposal
    - Dec 20 - Deadline for DAM poll results;
        - https://goo.gl/forms/VGOODBev9IlZN3Yg2
        - vote to accept FHIR subgroup recommendations; FHIR specification and FMM vote
        - V2/LRI block vote on subgroup recommendations
    - Dec 27 - (cancel?)
    - Jan 3 -
    - Jan 10 -
- **FHIR genomics – Overview of comments on 9/2016 ballot -** Amnon Shabo (Shvo)
    - See Amnon's recent email
    - Bob M - are we restricted to one base resource resource?
        - Amnon - practically, yes; experience from V3
    - See Chat
- **V2/LRI update**
    - Weekly V2 Lite sub-group meeting
    - Mondays, at 12:00-1:00PM ET
    - Triaged a block of Negative votes
    - Minutes
        - http://wiki.hl7.org/index.php?title=File:HL7_CG_V2_20161212.pdf
- **Other**
    -
- **Chat**
    - from Bob Milius to Everyone:
        - 1st slide: do we need to restrict ourselves to one base resource; OO has many
    - from Bob Milius to Everyone:
        - sequence has a role; different kinds of sequences: raw, consensus, reference for align build; reference for allele assignment
    - from Bob F to Everyone:
        - Bob M: I think it depends on who you ask. The FHIR team currently says we get 1 (as you know), but I agree with you - we need more if we are to stay true to FHIR design principles.
    - from Bob Milius to Everyone:
        - if use an existing format, is this uploaded as an embedded file? any 'approved' formats?
    - from Bob Milius to Everyone:
        - have to represent variant somehow for my use case; are you suggesting to use an encapsulated file like VCF?
    - from Bret Heale to Everyone:

        - could you put GenomicsPhenotype as a piece of supporting evidence in a Obersvation.interpretation instance?
    - from Bret Heale to Everyone:
        - but stating that GenomicsPhenotype is vastly differnet from other clinical interpretation is an interesting notion.
    - from Bret Heale to Everyone:
        - Is it?
    - from Bret Heale to Everyone:
        - I see at least three types of interpretation in genoimcs. The molecular level, the aggregate pathway level and the effect at the organ level
    - from Bret Heale to Everyone:
        - but all that can be altered depending on the context that the genomic data is being viewd in
    - from Bret Heale to Everyone:
        - e.g. if the physican is interested in a particular genomic disease, genetic variation on eye color might be considered benign (but could be a delterious mutation cuasing an inactive protiein)...just food for thought, pardon the length
    - from David Kreda to Everyone:
        - Bret's last comment refers to the "germline opportunity" and how to handle the "reuse" case separate from the original study and report of variants or WGS, etc.
    - from Bret Heale to Everyone:
        - I think amonon has been covering it too...I should have been patient :^)
    - from David Kreda to Everyone:
        - I like document addition but caution that structured document exchange is hyped a lot by in terms of semantics still a disappointment - not the stuff of desireasble interoperabilty. The C-CDA is a case in point. A merger of a number of CDA's, but the sending end and the receiving end have lots of semantic issues. Even 2.1 will miss the mark a lot. The API/atomic approach will only fix that if semantics within resources are pinned down ... via tough profiles (:-)) that is the stuff on long, drawn out Argonaut negotiations.
    - from Gil to Everyone:
        - Re: Bob F. There is no limitation to 1 resource post-reconciliation- ie for stu4.
    - from Bob Milius to Everyone:
        - voice keeps breaking up for me - anyone else?
    - from Perry Mar to Everyone:
        - 1) We are discussing the difference between an ObservedSequence and a ComparedSequence (which also refers to a reference sequence) in the IM subgroup; should we sync up on this point?
    - from Perry Mar to Everyone:

        - 2) It sounds like there is still confusion between the terms genetics and genomics; could we clear this up and document it fo people to refer to?
    - from Yi Wang to Everyone:
        - 1) How to interpret the sequence test result if the sequence is seperated into observed one and referenced? There should be two sequences for comparison? Sorry, I don't get that point. Now sequence resource is considered to store both reference and observed seq for now.
    - from Yi Wang to Everyone:
        - 2) Is that possible rename Sequence.variant? actually it will represent not clinical interpretation but basic acid change record, say A>T.
    - from clem mcdonald to Bob Milius (privately):
        - First time I used chat and sent my question to kevin rather than you. THought we should just focus on the one doable thing- Getting the variant attributes out of sequence and into Observation where the other variant attribute are. Thinkt hat id doable. The rest is too big
    - from David Kreda to Everyone:
        - Per Bob F. comment - this is not an either/or in a mutually exclusive thing. There changes and an "impact analysis" is due, but some changes are relatively easy and the change to Seq = omic + profile is not ipso facto a big change!
    - from David Kreda to Everyone:
        - (Not a YUUUUGE change, I mean)
    - from Gil to Everyone:
        - The fhir subgroup discussed approach last week (see slides sent by email): we will have a series of sessions (including others) from internal and external stakeholders to move forward on stu4.
    - from Bob F to Everyone:
        - David: Agreed - I just don't want discussions of changes (which can tend to go on for some time) to slow down the current effort
    - from Bob F to Everyone:
        - At some point we've got to choose an approach and go with it. If an alternative approach is suggested, it makes sense (to me) to pursue that in parallel rather than derail the existing effort IF the new proposal is sufficiently different as to cause churn and rework