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HL7 Clinical Genomics Weekly Call - March 15, 2016

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
3. Gaston Fiore - BCH - gaston.fiore@gmail.com
4. Ning An - HFUT - ning.g.an@acm.org
5. Joseph Kane - Epic - jkane@epic.com
6. Bob Freimuth - Mayo Clinic - freimuth . robert at mayo . edu
7. David Kreda - david.kreda@gmail.com
8. Terry McDonnell - Syapse - terrym@syapse.com
9. Perry Mar - Partners HealthCare System - pmar@partners.org
10. Joel Schneider - NMDP - jschneid@nmdp.org
11. Jonathan Holt (SeqTechDx) jholt@seqtechdx.com
12. Larry Babb (GeneInsight) lbabb@geneinsight.com
13. Brett Johnson icanbrj@gmail.com
14. Grant Wood - Intermountain
15. Mollie Ullman-Cullere - Partners
16. Heming Yao - BCH
17. Andrea Pitkus
18. Clem McDonald
19. Dave Blackman
20. Elizabeth
21. Gil Alterovitz
22. Robert Barkovich
23. Anwaruddin Mohammad

Discussion

- Minutes approval
    - Minutes from two calls need approval:
    - March 1
        - http://wiki.hl7.org/index.php?title=File:HL7_CG_20160301.pdf
        - Motion to approve / second = Bob F/ Clem M
        - Yea/Nay/Abstain = all / 0/ 0
    - March 8
        - http://wiki.hl7.org/index.php?title=File:HL7_CG_20160308.pdf
        - Motion to approve / second = Bob F / Clem M
        - Yea/Nay/Abstain =12 /0 /5 (Larry Babb, David Kreda, Brett Johnson. Joel Schneider)

- 5 min updates
    - ClinGen/ClinVar -
        - Larry - Nothing new
    - GA4GH -
        - Gil - some discussion with interacting with precision FDA
    - IOM -

        - Grant - nothing new, still gathering pilot teams
    - other -
        - none

- Project updates
    - FHIR subgroup
        - Gil - a lot going on, walk through, integrating feedback, need to update versions of spec for FHIR connectathon snapshot. Some conversation of how observation vs diagnostic report are done. Resulted from conversations with Argonaut. FHIR DSTU2 for comment http:///hl7.org/fhir/2015Jan/argonauts.html
    - V2 Lite
        - Proposal: V2 Lite Proposal - Round IV
        - See Clem's email from yesterday with "2016 03 14 1430 Layout of genomic model"
    - Clem's Presentation
    -
    - Saved Chat from March 1 (unaddressed questions/comments)
        - 10:31:17 AM from Larry Babb to Everyone:
            - I would recommend separating the "finding" from the "interpreation" or "significance" observation. These two observations are typically done be different groups within a lab (one is wet lab, one is geneticist).
        - 10:31:34 AM from jonathan to Everyone:
            - Range is tough approach. Some regions of genes have good coverage, but others b/c of pseudogenes.
        - 10:31:56 AM from Bob Freimuth to Everyone:
        - We need to accommodate preemptive testing (where there is no disease/condition)
        - 10:32:01 AM from Larry Babb to Everyone:
            - all "Mutation" terms should be changed to "Variant"
        - 10:32:16 AM from Larry Babb to Everyone:
            - that's been an ACMG decision for a while now.
        - 10:32:50 AM from Larry Babb to Everyone:
            - We need to be able to pass the genomic dna ref and coordinates (even if labs can't)
        - 10:36:28 AM from Bret Heale to Everyone:
            - @Larry Bab. cmmt on: Clinical Significance. For a testing lab the clinical significance is predetermined and based on the customer need the test is meeting. So, while I like what you ask for, is this going to be something that fits well into the testing laboratory's business model. They are not necessarily going to update the specific test panel reporting. But they might make a new one for ordering...just some thoughts
        - 10:40:56 AM from jonathan to Everyone:
            - yes, indeed "Allele ID is problematic". Variant ID is better, as long as we represent the reference properly. Agree , remove Allele ID. Cleaner.
        - 10:42:11 AM from David Kreda to Everyone:
            - (1) Won't the list of data items will balloon Observation's profile enormously, yet not yet be the clinical interpretations? (This is one reason - not the only one - which makes it quite risky to channel highly variable

sized data in Observation. (2) Also if the same data may be repurposed (will be repurposed for sure) to the prior interpretation, how is this a good argument for tight coupling in the same data structure. Post coordinated CDS will need to load and discard possibly the same data multiple times.

- 10:43:33 AM from Mollie to Everyone:
  - Somatic testing is commonly performed in context of ENSEMBL transcripts. Can we add ENSEMBL for all where RefSeq is?
- 10:43:53 AM from Mollie to Everyone:
  - Agree with Anwar - this is also important for Somatic
- 10:46:10 AM from Bret Heale to Everyone:
  - by adding interpretation, are you not coupling the data with knowledge? Isn't data forever and knowledge evolving? The point being that the interpretation changes but the lab value (bases present or missing) is less variable. right?
- 10:50:15 AM from David Kreda to Everyone:
  - To Brett's point: the underlying taxonomy of variants can expand hugely and, though it will never approach asymptotically the underlying data (actual sequence), the number will be huge. It is not that ID matching will be the issue - it is ID granting. At some point, why bother, if the search performance will produce rather more interesting results (think Google like search). This is not today's reports, but more or less explicitly discussed in NGS articles.
- 10:50:21 AM from Bob Freimuth to Everyone:
  - We should clarify the intent of this particular content - is it a structured form of the narrative and intended to be read by humans, or is it purely for computable use? Variant names for humans will need to be aligned with the literature, but that is less important for computers.
- 10:53:34 AM from jonathan to Everyone:
  - Where is "Allele/Variant Freq"?
- 10:55:46 AM from Bret Heale to Everyone:
  - many lab tests couple the interpretation with the nucleotide data for single variant test (in fact, the nucleotide content is treated as secondary content). this was an effective way to meet customers past needs (they only wanted the interpretation), I believe. do we need to provide a convincing enough reason to change business practices?
- 10:57:51 AM from jonathan to Everyone:
  - Strongly Agree with @larry !
- 10:58:12 AM from Bret Heale to Everyone:
  - pardon, I'll work on my spelling.
- 10:58:35 AM from Amnon Shabo (Shvo) to Everyone:
  - I strongly agree with Larry!!
- 10:59:24 AM from Mollie to Everyone:
  - does this look like an array of question(disease):answer(pathogenic) pairs
- 11:00:43 AM from Bob Freimuth to Everyone:
  - Someone went off mute, perhaps computer and phone audio interfering
- 11:00:48 AM from Andrea Pitkus to Everyone:
  - Curious how the reporting would be compliant with CLIA and where terminology code systems would be added.

- 11:01:25 AM from David Kreda to Everyone:
  - I would like Clem to address the "beyond report" paradigm in his context setting, not to deprecate the report, but to emphasize the future-purpose CDS
- 11:02:41 AM from Andrea Pitkus to Everyone:
  - right. origination of the report in a LIS and then downstream uses of the discrete data