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HL7 Clinical Genomics Weekly Call - Wed Mar 23 15:44:41 CDT 2016

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Jonathan Holt (SeqTechDx) jholt@seqtechdx.com
3. Kevin Power (Cerner) kpower@cerner.com
4. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
5. Joey Yang -HFUT- jiaoyun@hfut.edu.cn
6. David Kreda - david.kreda@gmail.com
7. Gaston Fiore - BCH - gaston.fiore@gmail.com
8. Joel Schneider - NMDP - jschneid@nmdp.org
9. Joseph Kane - Epic - jkane@epic.com
10. Anwaruddin Mohammad - Oracle - anwaruddin.mohammad@oracle.com
11. Bret Heale - University of Utah/Intermountain Healthcare - bheale@gmail.com
12. Brett Johnson - icanbrj@gmail.com
13. Larry Babb - lbabb@geneinsight.com
14. Terry McDonnell - Syapse - terrym@syapse.com
15. Fei Wang - Partners Healthcare - fwang0@partners.org
16. Usha Reddy- Oracle- usha.reddy.pa@gmail.com
17. Jeremy Warner - Vanderbilt University - jeremy.warner@vanderbilt.edu
18. Gil Alterovitz
19. Grant Wood
20. Andrea Pitkus
21. Dave Blackman
22. Huanquin Dai
23. JD Nolen
24. Robert Barkovich
25. Clem McDonald

Discussion -
Note: we are changing the order of agenda items so that Clem can leave early for another commitment:

- V2 Lite -
    - Clem continued his discussion, using the "2016 03 18 445PM Layout of genomic model.pdf" document
    - Proposal: V2 Lite Proposal - Round IV
        - Part of discussion around 0- and 1-based coordinate systems. HGVS uses 1-based. We decided for FHIR we would use 0-based for start and end coordinates. We should be clear which is used when.
            - Both are used in molecular genetics. Generally speaking, human readable formats tend to use 1-based. Processing formats tend to use 0-based.
            - Jonathan: I'm all for the 1-based and be consistent throughout all systems.
            - Gil: Has some discussions on the topic: https://www.biostars.org/p/6373
            - Bob: I think GA4GH and ClinGen uses 0-based

- Gil: ga4gh: https://github.com/ga4gh/schemas/issues/121
- Kevin Power: Seems like ClinVar uses 1-based: https://github.com/clingen-data-model/allele-registry/issues/2
- Jonathan: genBank/reSeq is 1-based
- Kevin Power: ClinVar uses a 1-based numbering approach (like VCF)."
- Gil: ClinGen data model uses 0-based: https://github.com/clingen-data-model/clingen-data-model/issues/60

        - (*LBabb in response to JHolts concern about missing phasing representation in compound variants*) Regarding the concern about representing the "phasing" in compound variants. We could consider adding a coded field at the top level (at the level of the compound identifier) for phasing with the value set (in cis, in trans, unknown). This could provide the missing info that is embedded in the HGVS nomenclature related to showing the phasing with brackets and semi-colon positions and such.

- FHIR subgroup
    - Vote on FHIR snapshot for May Connectathon (see Gil's email):
    - Background:
        - Snapshot for FHIR Connectathon 12, which CGWG elected to take part in previously, will be taken of whatever is on FHIR current build site on Mar 27, 2016.
        - FHIR subgroup voted to support spec (see genomics-advisor.smartplatforms.org:4000)
        - Development can continue - this motion is just to avoid missing the snapshot.
        - Picture in time
    - Motion/second: Gil/Grant
        - Motion: Copy the FHIR Genomics staging build from genomics-advisor.smartplatforms.org:4000 to the FHIR current build site.
        - Discussion: Amnon - joined last week's CG subgroup call, and were only 3 on call (maybe a different #?), feels we haven't truly examined alternative structures. (webex listed below)
        - David K: There have been a number of changes since Jan that we should test in the May connectathon. (see David's email). If there is an alternative structural approach, the onus is upon those proposing the alternate to demonstrate its advantage.
        - Jon: As long as we aren't committing ourselves, and we can totally scrap it in the future for an alternative model, then we should test the current model.
        - Jon: The reason we have an issue is that it was grown, but not built within the working group. Still have reservations that it's a top down approach rather than a bottom up approach. Still just a fancy observation.
        - Gil: In working with Mollie, the initial impressions in early work was that too much was jammed into the profile.
        - Vote:
            - Abstains: Amnon
            - Nays: 0
            - Yeas:
                1. Terry McDonnel
                2. Grant Wood
                3. Andrea Pitkus
                4. Bret Heale

5. Brett Johnson
6. David Kreda
7. Gaston Fiore
8. Gil Alterovitz
9. Jeremy Warner
10. JD Nolen
11. Joel Schneider
12. Joey Yang
13. Jonathan Holt
14. Joseph Kane
15. Kevin Power
16. Larry Babb

The Call in for FHIR Sub-Group FYI is:

https://hms.webex.com/hms/j.php?MTID=m027c981f9fa723d90ecc1181fd35e0bd
Meeting #: 714 370 759
Password: udn2016
Audio: 1-877-668-4490

- Minutes approval - Did not have time for this
  - March 15
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20160315.pdf
    - Recorded audio at http://www.hl7.org/documentcenter/public/wg/clingenomics/minutes/HL7%20CG%20call%2020150315.mp3
    - Motion to approve / second = /
    - Yea/Nay/Abstain = / /

- 5 min updates - did not have time for this
  - ClinGen/ClinVar -
  - GA4GH -
  - IOM -
  - other -