http://bit.ly/1XstK6w

HL7 Clinical Genomics Weekly Call - May 24, 2016

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Jonathan Holt - SeqTechDx - jholt@seqtechdx.com
3. Gaston Fiore - BCH - gaston.fiore@gmail.com
4. Gil Alterovitz- gilusa@gmail.com
5. David Kreda - david.kreda@gmail.com
6. Bob Freimuth - Mayo
7. Larry Babb - Sunquest/Geneinsight - lbabb@geneinsight.com
8. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
9. Bret Heale - Intermountain Healthcare - bheale@gmail.comB
10. Perry Mar - Partners HealthCare System - pmar@partners.org
11. Joel Schneider - NMDP - jschneid@nmdp.org
12. Brett Johnson - icanbrj@gmail.com
13. Mollie Ullman-Cullere - Better Outcomes - mollie@betteroutcomes.com
14. Scott Bolte - Niss Consulting - Scott.Bolte@gmail.com

Discussion

- Minutes approval
    - May 3 , 2016 -
        - http://wiki.hl7.org/index.php?title=File:HL7_CG_20160503.pdf
        - Motion to accept - Amnon
        - 2nd - David K
        - Discussion -
        - yea/nay/abstain = 5+ / 0 / Terry, Scott, Perry, Jeffrey Karp
        - results - passed
    - May WGM - Montreal
        - http://wiki.hl7.org/index.php?title=File:HL7CG_WGM_May2016_Montreal_Minutes.pdf
        - Motion to accept -
        - 2nd -
        - Discussion -
        - yea/nay/abstain = / / Jonathan, David K. Brett J.
        - results - defer to next week
- Brief updates
    - ClinGen/ClinVar -
        - Larry - GA4GH, HL7, ClinVar/ClinGen getting together and work on harmonize, many touchpoints are obvious, but others need work; from ClinGen - Bob F and Larry B will attend from ClinGen perspective
        - Larry - NCBI- Allele Registry: NCBI is drafting White paper on Allele Registry design/model; beginning to plan a community meeting in the fall timeframe to begin publicizing and understanding community policy and service requirements
        - Mollie - PGx in scope?
        - Larry - no star alleles initially; just simple alleles

        - Larry - not starting over, using collective knowledge at NCBI across many archives (dbSNP, dbVar/dbGap, ClinVar, etc…)
        - Clem - is this a cleanup (at NCBI)?
        - Larry - you could look at it that way, but more than that. Still in pilot phase but moving forward in a very positive way. but not a done deal
        - Clem - databases very large, need to be curated for a long time
        - Larry - the intent is the service would be available to register (find or create) canonical and contextual identifiers for alleles on the fly (larry's perspective - not an NCBI statement).
    - GA4GH -
        - Gil - Larry covered it
    - National Academies-
        - Gil - June 1 pilots will be meeting
    - other -
        -

- FHIR
    - On May 19, 2016, the FHIR Subgroup discussed transitioning work from its staging site solution to the standard HL7 solution and voted in favor of such. This is set out below.

    - HL7 Working Procedure Background
        - The HL7 GForge tool/website is used by other HL7 Work Groups to support the development and publication of HL7 Standards. Using it, CGWG members will be able to go to the webpage of a FHIR resource or profile and use the "Propose a change" feature to enter feedback in an itemized manner in the GForge tracker. Feedback on that site can be discussed by the CGWG and FHIR Subgroup. Proposed changes, once approved by a CGWG vote, can then be committed to the FHIR Current Build specification, with a full audit trail of such activities and comments.

          To move to this tool, the FHIR Subgroup proposes committing to the FHIR Current Build all changes made on the FHIR Genomics staging site (fhirgenomics.org) since March 27, the date of the commit made for Connectathon 12. After the new commit, CGWG members will use the GForge site to comment and report all new issues using the GForge solution and all future updates to the Current Build will be conducted based upon votes taken against the record in that tool.

    - Proposed Motion
        - The CGWG approves:
            - (a) committing the FHIR genomics specifications (http://genomics-advisor.smartplatforms.org:4000), that includes all updates to the March 27, 2016 Connectathon 12 snapshot (described here: http://hl7.org/fhir/2016May/index.html) to the HL7 FHIR Current Build; and
            - (b) using the HL7 GForge site henceforth to
                - (i) collect and track CGWG participant feedback regarding changes to genomics resource and profiles;
                - (ii) support developers in implementing the FHIR genomics specification; and
                - (iii) maintain FHIR genomics specification version control.

    - Motion = Gaston
    - Second = Gil
    - Discussion
    - Bob F = Question how is the structure of the process of FHIR development changed/managed?
    - Gaston - Part below is not related to this motion
    - Resulte = rest / 0 / 0 - motion passes
    - David = how quickly can this be done?
    - Gaston = a few days

- FHIR Deadlines
  For our Work Group to have anything in the STU3 ballot, we have several activities we must promptly address due to deadlines set by the HL7 FHIR Management Group. The key deadline is Wednesday, June 1, the day after our next CGWG call.

  By June 1, in particular, we must have voted for certain proposals that must be document in an officially sanctioned format. These proposals are forward-looking assertion that we are going to work to have certain actual specifications ready in the relative near future, some number of weeks before the September WGM. In particular, here are the individual proposals that all work groups, including ours, would need to get done (if not already done and approved at an earlier date) by June 1 to be considered eligible for the STU3 ballot process:

  1. FHIR Resource Proposals

  2. FHIR profile Proposals

  3. FHIR Implementation Guide Proposal

  Finally, all work groups, ours included, need to indicate no later than June 1 if they will be participating in the September FHIR Connectathon.

  Re (1). We approved the Sequence Resource proposal in 2015. Again, not the actual specification, which we continue to work on, but merely the go-ahead for that resource. We will eventually vote again on the exact specification that would part of STU3.

  Re (2). We must promptly consider proposals for each of the profiles. For that, we have drawn up a proposal for each, see:

  http://wiki.hl7.org/index.php?title=ObservationGenetics_FHIR_Profile_Proposal

  http://wiki.hl7.org/index.php?title=DiagnosticReportGenetics_FHIR_Profile_Proposal

  http://wiki.hl7.org/index.php?title=DiagnosticOrderGenetics_FHIR_Profile_Proposal

http://wiki.hl7.org/index.php?title=FamilyMemberHistoryGenetics_FHIR_Profile_Proposal

http://wiki.hl7.org/index.php?title=SequenceConsensusSequenceBlock_FHIR_Profile_Proposal

http://wiki.hl7.org/index.php?title=DiagnosticReportHlaResults_FHIR_Profile_Proposal

Examples of approved profile proposals are below (link given by Lloyd):
http://wiki.hl7.org/index.php?title=Category:Approved_FHIR_Profile_Proposal

We hope that a majority will approve the go-ahead for proposals for these profiles. The current profiles work-in-progress for which the proposal apply are, of course, presently set out at the staging site (FHIRgenomics.org). For voting, we propose that each proposal be a separate google poll that closes at the beginning of our CGWG call on Tuesday, May 31.

These profile proposals correspond to proposals that we were polling last week for a FHIR Maturity Model target at the end of December 2016. That said, based on polling last week, the FMM target was 3 except for the HLA/consensus block and Family Hx, each of which tied between 1 and 3. That vote can also stay open through the beginning of our CGWG call on Tuesday, May 31.
See:
https://docs.google.com/forms/d/1nCWreYHSDAr-YH7wnDJdNb8S2uuyYkbmRRQUBNbZFos/viewanalytics

Re (3). We must promptly consider a proposal to produce an Implementation Guide (IG). Again, the proposal is here (http://wiki.hl7.org/index.php?title=Genomics_FHIR_Profile_Proposal) and its table of contents recaps the list of resources and profiles, which can be amended depending upon the vote in (2) and in later final commit votes, etc.

The final vote we must take – and this we could take today – is if we intend to participate in the September 2016 FHIR Connectathon.

Motion to participate - David
Second - Bret
Discussion -
Jon - good idea to participate, but need to fine tune the resources/profiles
David - 6-7 weeks for improvement to ready for connectathon snapshot
Results = rest / 0 / 0
Motion passes

FMM Poll Results

Resource/Profile	***50+%***	***60+%***	***Greatest number of votes***
Observation	3	2	3
Diagnostic Order for Genetics	3	1	3
Diagnostic Report for Genetics	3	2	3
Family Member History of Genetics Analysis	2	2	1, 3 (tie)
Consensus Sequence Block	1	1	1
HLA Genotyping Results	1	1	1
Sequence	2	2	3