HL7 Clinical Genomics Weekly Call - August 21, 2018 11:00 AM (US Eastern)

Minutes:
https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_I5Kt72NEBYXZuwhFg/edit
short url: http://bit.ly/2aqVmqz

Attending the meeting:
Join the online meeting (VoIP available with this):

- Online Meeting Link:
  - https://join.freeconferencecall.com/clingenomics
  - Meeting ID: clingenomics

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- Dial-in Number:
  - (515) 604-9708 - United States
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Agenda

Attendees Sign-in

Minutes Approval

Topics to Review

- Agendas and Important Dates
- External efforts
- Subgroup reports

Topic 0: WGM - Sep 2018 - Baltimore - Agenda

Topic 1: Block Vote

Topic 2: Ballot reconciliation -

Topic 3: Capturing Genomic Panel Definitions

Topic 4: Compare Sequence and Observation

Topic 5: New LOINC codes?

Chat

Minutes: http://bit.ly/2aqVmqz

Attendees Sign-in

(Presiding co-chair: Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org)

1. Kevin Power - Cerner - kpower@cerner.com
2. Bret Heale - Intermountain Healthcare - bheale@gmail.com
3. Dora Finkeisen - MOLIT Institut - Dora.Finkeisen@molit.eu
4. Scott Robertson - Kaiser Permanente - scott.m.robertson@kp.org
5. Mullai Murugan - Baylor College of Medicine murugan@bcm.edu
6. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
7. Bob Dolin - Elimu Informatics - bdolin@elimu.io
8. Julian Sass - Berlin Institute of Health - julian.sass@bihealth.de
9. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
10. Alex Mankovich - Philips - alex.mankovich@philips.com
11. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
12. James Jones - BCH - james.jones.bch@gmail.com
13. JD Nolen - Children's Mercy Hospital - jlnolen@cmh.edu
14. Jamie Parker - Carradora Health -jamie.parker@carradora.com
15. Andrea Pitkus - apitkus@gmail.com

Minutes Approval

- August 14
  - http://wiki.hl7.org/index.php?title=File:HL7_CG_20180814.pdf
  - Motion/2nd to accept minutes: Clem / Kevin
  - Discussion:
  - Vote: Abstain / Nay / Yea: 0 / 0 / 11
  - Result: passes

Topics to Review

Agendas and Important Dates

Date	Co-Chair	Agenda	Important Dates
5/29/2018	Bob M	Review WGM minutes (note that Amnon Shabo edited the minutes regarding the sessions when his ballot comments were discussed)	
6/5/2018	Kevin	Ballot comments	Jun 6 - Deadline for connectathon proposals to FMG
6/12/2018	Kevin	Con call tech Connectathon 'Variant Grouping'	
6/19/2018	Kevin	Con call tech Connectathon Block Vote 'Variant Grouping'	
6/26/2018	Bob M		June 27 - Connectathon Proposals Due
7/3/2018			Jul 1 - Work groups notify the FMG whether they need to reballot normative packages (due to substantive change), STU resources (due to significant refactoring) or IGs during the Sept. cycle July 6 - Deadline to notify HG of additions/changes to co-chair opentings

7/10/2018	Bob M	IG block vote NIB vote	July 11 - Call for co-chair nominations July 15 - Notification of Intent to Ballot
7/17/2018	Bob M		
7/24/2018	Kevin	Consensus Groups Definitional Sequence	July 23 - formation of consensus groups
7/31/2018	Bob F	Topic 0: Deadline for resolutions to ballot comments Topic 1: FYI: Ballot Items for follow-up Topic 2: Block Vote #1 Topic 3: Block Vote #2 Topic 4: Proposed changes to Described Variant Topic 5: Secondary findings (proposal) Topic 6: Impact vs. Interpretation	Aug 5 - Reconciliation packages must be posted by this date at the absolute latest Aug 10: All substantive reconciliation applied. FHIR Core is frozen, limited QA process for content subject to ballot only
8/7/2018	Kevin P	Trackers needing follow-up Block Vote Cytogenetics in the IG Compare Sequence and Observation	Aug 10 - close to co-chair nominations
8/14/2018	Bob M	Topic 1: Block Vote #1 Topic 2: Impact/Interp Topic 3: New LOINC codes? Topic 4: What was tested? Topic 5: Cytogenetics in the IG Topic 6: Compare Sequence and Observation	Aug 17: Pre-ballot (and connectathon) content freeze. Publication process begins, including ensuring that content is appropriately flagged for ballot status and there are no last minute QA issues
8/21/2018	Bob M	Topic 0: WGM Agenda Topic 1: Block Vote Topic 2: Ballot reconciliation - 16789 Topic 2: Capturing Genomic Panel Definitions Topic 6: Compare Sequence and Observation Topic 5: New LOINC codes?	Aug 24 - ballot opens for voting
8/28/2018	Bob M		Aug 31: Deadline to post WGM agenda on the WGM information page (WG Health metric)
9/4/2018	Bob F		
9/11/2018			
9/18/2018	Bob M		
9/25/2018			

32nd Annual Plenary & Working Group Meeting

Sep 29, 2018 to Oct 5, 2018 - Baltimore, MD

External efforts

- GA4GH Genomic Knowledge Standards (GKS) (leads: Bob Freimuth, Andy Yates)
    - Variant Representation (formerly VMC)
        - Work continues on two fronts: we are finishing up the last pieces to the 0.2 release (lead by Reece Hart) and we are extending the model to support complex variants (e.g., fuzzy ends, CNV) (lead by Larry Babb/Tristan Nelson)
        - Hoping to have a deliverable out by Oct 3
    - Variant Annotation
        - Collecting use cases, prioritizing for modeling work (lead by Matt Brush and Javi Lopez)
        - Hoping to have a deliverable out by Oct 3
    - Gearing up for the GA4GH plenary meeting (Basel, Switzerland) on Oct 3-5, 2018. This meeting conflicts with the HL7 WGM in Baltimore but future HL7/GA4GH meetings will be scheduled so they do not overlap.
    - Clem - we should get their documentation as soon as possible
    - Bob F - there are a number of people working across HL7 and GA4GH, significant effort to harmonize between groups; definitely share concern about potential for misalignment between groups, working to harmonize standards
    - Bob D - wonder how effort with IM group and other groups align? eg actual workflows with BED / BAM files and BCF spec
    - Bob F - a number files mentioned are now owned by GA4GH and effort to harmonize across workstreams; GKS now working on downstream formats, so hope that those will align with clinical reporting in the future
- DIGITiZe (aka National Academies) (Grant Wood, JD Nolen)
    - no report
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
    - no report
- CDISC PGx (Dorina B.)
    - no report
- ONC Sync for Genes (Bob Freimuth)
    - ONC Interoperability Forum was held Aug 6-8 in Washington, DC
    - Pilot sites are planning/implementing their respective use cases. ONC will be encouraging their participation in both the Sept 2018 and Jan 2019 FHIR Connectathons.

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/1azKiQdhAQKuHhxAznEp8141FLdFLACIu8MzF2LxADxg/edit#
        - Draft model docs to be posted soon, hope to get it out by Thur (please review and submit feedback)
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.zfi9l8jfe4la
        - Overview of the remaining Neg-Major trackers:
            - Missing LOINC codes - move into block
            - Transplantation/HLA needs more complete narrative -didn't cover
            - Sequence Phase Relationship x Complex Variant - discussed
            - Inherited Disease Pathogenicity profile - deferred/not persuasive

Topic 0: WGM - Sep 2018 - Baltimore - Agenda

- **https://tinyurl.com/HL7-CG-Sep2018**
- https://docs.google.com/document/d/1CrGLeY7Qk1pk9NGindjvKeD2IJeV9g3GYjkOcy_qBNY/edit
- clem wants to continue and hopefully finish vocabulary discussion in Sep 2018
    - link to comment in last ballot
    - Joel - adding terminologies to fhir may not be scalable; vocab wg is dealing with this now
- clem - need to recruit clinical geneticists for group
    - Jeremy used to be in the group
    - have them join clinicians on fhir
    - bob d - have specific questions such as ACMG;
    - scott - comment from clinician was that our our IG wasn't clinically focussed; we need to have specific questions
    - bret h - profile is for programer, not for clinician; we need to be able to present what clinicians need

Topic 1: Block Votes

Block #1

Comment Submitters

- Amnon Shabo
- Bob Milius
- Clement McDonald
- Scott Robertson

Line Items

- 16325 %22haplotype%22+in+medical+genetics (Scott Robertson) Considered - Question Answered
- 16938 knowledge+representation+-+2018-May+Genomics+%2378 (Amnon Shabo) **In Person** Considered for Future Use
- 16793 Discussion+needed+on+change+from+display+names+on+84414-2+-+2018-May+Genomics+%2332 (Clement McDonald) Not Persuasive
- 16871 Specialization+for+somatic+variant+might+not+be+necessary+-+2018-May+Genomics+%2356 (Clement McDonald) Not Persuasive
- 16513 need+glossary (Bob Milius) Persuasive
- 16686 Clarification+needed+on+virus%2Fbacteria+sample+-+2018-May+Genomics+%232 (Clement McDonald) Persuasive
- 16866 Overall+comments+on+Appendix+B+-+2018-May+Genomics+%2354 (Clement McDonald) Persuasive with Mod

- motion to accept proposed dispositions / 2nd - Clem/Scott
- abstain/nay/yea - 0 / 0 / 15
- motion passes

Block #2

Comment Submitters

- Clement McDonald
- Kevin Power
- Kirt Schaper

Line Items

- 16079 spelling+error+and+example+request (Kirt Schaper) Not Persuasive
- 16110 DiagnosticReport+Category+could+also+be+Cytogenetics (Kevin Power) Not Persuasive
- 16851 Comment+on+label+%22copy+number%22+for+what+used+to+be+structural+variant+-+2018-May+Genomics+%2349 (Clement McDonald) Not Persuasive
- 16848 Comment+on+ISCN+not+strictly+limited+to+legacy+cytogenetics+-+2018-May+Genomics+%2348 (Clement McDonald) Persuasive with Mod
- 16854 Usually+applied+to+deletion%2Fduplication+studies+and+might+be+best+applied+at+higher+point+in+the+report+structure.+-+2018-May+Genomics+%2350 (Clement McDonald) Persuasive with Mod
- 16869 Overall+comments+on+1.4+Cytogenomic+Reporting+-+2018-May+Genomics+%2355 (Clement McDonald) Persuasive with Mod

- motion to accept proposed dispositions / 2nd - Clem / Scott
- Abstain/Nay/Yea - 0 / 0 / 15
- motion passes

Topic 2: Ballot reconciliation -

- 16789 Discussion+needed+on+change+from+display+names+on+84413-4+-+2018-May+Genomics+%2331 (Clement McDonald) Persuasive with Mod

    - Submitted by: Clement McDonald (National Library of Medicine)
    - Existing Wording: Figure 5: Genotype 84413-4
    - Proposed Wording: Genotype Display Name 84413-4
    - Comment: I understand why you want to shorten, but the change could mislead. These are not solid codes for genotype or haplotype. Would like to find a way to link from the figure (or content below them) to the LOINC code, description and answer list. Have linked to the answer list in the change document but these early tables are a bit more digestable. Lets talk.

    - motion - accept ballot comment resolution as stated (change to string)
        - Kevin / Clem

- Discussion
  - No publicly available code systems for this concept yet
  - Code system for HLA use case is currently in development
  - Changing to String loses the ability to use this once it is in place
  - Examples of HLA genotype grammar in current use: HML, GL String manuscript (could presently be viewed as a local, or private code system)
  - valueCodeableConcept.text could be used where no public code system is available
  - valueCodeableConcept.coding could be used where there is a local code system
- kevin withdraws motion due to lack of time
  - added after call - Bob M posted question to O&O stream on chat.fhir.org, asking when is observation.valueString preferred over observation.valueCodeableConcept.text
    - https://chat.fhir.org/#narrow/stream/103-Orders-and.20Observation.20WG/subject/valueString.20vs.20valueCodeableConcept.2Etext

Topic 3: Capturing Genomic Panel Definitions

- https://chat.fhir.org/#narrow/stream/43-genomics/subject/Capturing.20Genomic.20Panel.20Definitions

- **Kevin Power**
- So, to keep this conversation going - We have basically two proposals on the table:
  - Profile Observation (Regions-Studied) with components[] to define what was tested
  - Profile Device (Genetics-Assay) with extensions to define what was tested
- Reactions? Preferences? I lean towards profiling Observation.

- **Lloyd McKenzie**
- I think assertions of what was tested need to be made using Observations. There will be a wide variety of devices that can test things in different ways. The record of "what was found" should be captured in a way that's consistent and independent of how the testing was done.

- no time to discuss today

Topic 4: Compare Sequence and Observation

https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg_xpwir8rEORkNjMfemvqxE0/edit#gid=0

- no time to discuss today

Topic 5: New LOINC codes?

- no time to discuss today
- Set of trackers regarding the new for new LOINC code, changes to some LOINC codes, and some value set/answer list questions:
- 16262 Change+LOINC+answer+list+for+complex+variant+type (Bob Dolin)
    - persuasive with mod
    - Bob D will provide final mod description
    - marked waiting for input
- 16264 Modify+dna-chg-type+answer+list (Bob Dolin)
    - Bob D will provide more concrete suggestions
    - marked waiting for input
- 16272 Revise+answer+list+for+Genotype+Medication+Efficacy+Impact+profile+%28LOINC+51961-1%29 (Bob Dolin)
    - Clem - took from publication received from Bob Freimuth
    - Bob F - answer list did not come from CPIC, and CPIC doesn't have one similar that we should use
    - Andrea - need to be careful about changing anything CLIA requires re answer lists
    - Bob D will resend Shennon's email about this
    - marked waiting for input
- 16180 Inherited+Disease+Pathogenicity+-+Must+have+mode-of-inheritance+value+set (Kevin Power)
- 16183 Computable+Genetic+Finding+-+Region+name+LOINC+is+not+correct (Kevin Power)
- 16184 Genetic+Impact+-+Need+LOINC+code+for+level+of+evidence (Kevin Power)
- 16239 Need+an+Observation.code+for+DescribedVariant (Kevin Power)
- 16242 DescribeVariant.component%28Simple+var+ID%29+LOINC+code+is+for+%22Discrete%22+Variant (Kevin Power)
- 16244 Need+a+LOINC+for+Coordinate+System (Kevin Power)

- last week discussed three issues, see above (16262, 16264, 16272)

Chat

- Andrea Pitkus 10:17AM

    - is there a link to the HL7 connecthathon cases that will be tested for the Sync for Genes ONC pilots?
- Andrea Pitkus 10:18AM
    - will they be testing CG Diagnostic Report or other profiles?
- Andrea Pitkus 10:19AM
    - sounds like jan then...
- Andrea Pitkus 10:19AM
    - thanks
- Kevin 10:21AM
    - Looks like it is private?
- Kevin 10:23AM
    - Bob - when you get a chance, I can see it, but can't edit it.
- Anonymous 10:24AM
    - Co-chairs attending Sept WGM: Bob M, Kevin, Gil (Bob F and Amnon confirmed not attending, Andrea likely not attending)
- Andrea Pitkus 10:32AM
    - concur that it has to work for clinical care for folks to implement, unless we want to split into separate profiles.
- Anonymous 10:33AM
    - Agree that we should encourage more engagement, especially from those with diverse backgrounds. Also think we need more “Bobs” in the group.
- Bret Heale 10:41AM
    - VCF is a pretty simple as a format, but how many clinical geneticsts would care to view the result as VCF? The final PDF is the 'report' whereas the VCF arfitifact is the machine readable result - suitable for CDS, later analysis, etc... Just some thoughts and just wanted to put them into the chat. Don't need to extend the discussion today : ^)
- Bret Heale 10:56AM
    - http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html
- Bret Heale 10:56AM
    - oops meant: http://hl7.org/fhir/uv/genomics-reporting/general.html
- Bret Heale 10:56AM
    - figure 5

Clinical Genomics Docs

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - Review complete as of Aug 1, 2017
    - Approved in Sep 2017 WGM in San Diego
- Decision Making Process
    - https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit
    - Review complete as of Aug 15, 2017
    - Approved in Sep 2017 WGM in San Diego
- DAM
    - http://tinyurl.com/damcgdoc