CG-2018-12-04

HL7 Clinical Genomics Weekly Call - 4 Dec 2018 11:00 AM (US Eastern)

Minutes

http://tinyurl.com/HL7CGGroupCall

https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_I5Kt72NEBYXZuwhFg/edit

Attending the meeting

Join the online meeting (VoIP available with this):

- Online Meeting Link:
  - https://join.freeconferencecall.com/clingenomics
  - Meeting ID: clingenomics
- Dial into the conference:
- Dial-in Number:
  - (515) 604-9708 - United States
  - Access Code: 289092
- International Dial-in Numbers:
  - https://www.freeconferencecall.com/wall/clingenomics/#international

Agenda

Previous Minutes Approval

Agendas and Important Dates

External efforts

Subgroup reports

Topic 0: Ballot Sign Up

Topic 1: Jan 2019 WGM Agenda Planning

Topic 2: Non-ballot tracker items

Attendees Sign-in

(Presiding co-chair: Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu)

1. Patrick Werner - Molit Institute / Heilbronn University - patrick.werner@molit.eu
2. Kevin Power - Cerner - kpower@cerner.com
3. JD Nolen - Children's Mercy Hospital - jlnolen@cmh.edu
4. James Jones - BCH- james.jones.bch@gmail.com
5. Ning Xie - BCH - ningxie2018@gmail.com
6. Bob Dolin - Elimu Informatics - bdolin@elimu.io
7. Liz Amos - NLM - liz.amos@nih.gov
8. Deepak Sharma- Mayo Clinic - sharma.deepak2@mayo.edu
9. Andrea Pitkus - apitkus@gmail.com
10. Julian Sass - Berlin Institute of Health - julian.sass@bihealth.de
11. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
12. Jamie Parker - Carradora Health -jamie.parker@carradora.com
13. Caterina Lasome - iON Informatics - cat@ioninformatics.com
14. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
15. Jungang Zou - XMU -jungang.zou@gmail.com
16. Alex Mankovich - Phillips - alex.mankovich@philips.com
17. Bret Heale - Intermountain Healthcare - bheale@gmail.com

Previous Minutes Approval

- Nov 27
  - https://confluence.hl7.org/display/CGW/CG-2018-11-27
  - Motion: Accept minutes
    - Move / 2nd: Bob M / Patrick
    - Discussion: none
    - Vote: (Abstain / Against / For): Cat, JD / 0 / 12

- Result: motion passes

Agendas and Important Dates

Date	Co-Chair	Agenda	Important Dates
9-Oct-18		**No Meeting**	
16-Oct-18	Kevin P	WGM Review 2-to-FHIR PSS - vote Committers channel on Zulip NIB for IG - vote Code Systems / Value Sets	
23-Oct-18	Kevin P	FHIR IG Block Vote - vote Committers channel on Zulip Code Systems / Value Sets	
30-Oct-18	Kevin P	Tracker 19453 - Rename Sequence -> MolecularSequence Tracker 19440 - Update to 'stu note' on FHIR Core profiles and artifacts Tracker 1900 - Relax cardinality for reference. windowStart/windowEnd Previous FHIR Core Trackers with no or inadequate resolution documented Code Systems / Value Sets (Discussion: Patrick /Julian)	Oct 31: All R4 STU comments reconciled in gForge with votes and no "tracker issues" Oct 28: NIBs due - IG must be "feature complete" and building in HL7's continuous integration environment. If not, the NIB will be refused
6-Nov-18	Bob M		
13-Nov-18	Bob M		Nov 18: IG Content deadline – only QA changes after this point Nov 18: IG Ballot reconciliation spreadsheet due - two ballot items left to reconcile
20-Nov-18	Kevin P	Topic 0: Ballot Sign Up Topic 1: May 2018 Ballot Reconciliation Complete Topic 2: Non-ballot tracker items still remaining Topic 3: Code Systems / Value Sets (Discussion: Patrick/Julian - Clem/Liz)	Nov 23: Co-Chair Nominations Close at 5:00 pm Eastern (email nominations to NOMINATIONS@HL7.ORG)
27-Nov-18	Bob M		Dec. 2: IG Final freeze deadline
4-Dec-18	Bob F	Topic 0: Ballot Sign Up Topic 1: Jan 2019 WGM Agenda Planning Topic 2: Non-ballot tracker items	Dec 6 - Consensus pool signup closed Dec 7 - Ballots opened Dec 7: Co-Chair election statements due to HQ
11-Dec-18	Bob M		Dec 14: Deadline to post your WGM agenda on the WGM information page (WG Health metric)
18-Dec-18	Kevin P		
25-Dec-18	Holiday	meeting cancelled	
1-Jan-19	Holiday	meeting cancelled	

8-Jan-19	Bob M		Jan 7 - Ballots closed

2019 January Working Group Meeting

Health Level Seven International January 2019 Working Group Meeting, San Antonio, TX

- ***Location: San Antonio, TX United States***
- ***Start Date: January 12, 2019***
- ***End Date: January 18, 2019***
- ***Early bird discounts end December 21!***
- GA4GH Genomic Knowledge Standards (GKS) (leads: Bob Freimuth, Andy Yates)

External efforts

- Variant Representation (formerly VMC) (lead by Larry Babb/Tristan Nelson)
  - Working on complex variants in alignment with VA subgroup
  - https://docs.google.com/document/d/1Sulg3kECnorTEAbutlNOsK-lFkKAcKpl6lHgPaPQEgA/edit#heading=h.k9apf9d8j9y2
  - Variant Annotation
    - Defining types of annotations, modeling (lead by Matt Brush and Javi Lopez)
    - https://docs.google.com/document/d/13sSChUB9rW7vl1ep-tZnaDzSWb_MyWIvSzEFVS32quE/edit#heading=h.t2adm0qua505
- DIGITiZe (aka National Academies) (Grant Wood, JD Nolen)
  - Still attempting to schedule a follow-up meeting (Sandy, JD, Grant) for next steps.
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
  - Internal follow-up on outcomes of the in-person modeling meeting, vetting ideas
  - Significant consideration given to GKS/VR approaches, may help drive that work
- CDISC PGx (Dorina B.)
  - no report
- ONC Sync for Genes (Bob Freimuth)
  - Collected example genetic test reports from each pilot site, mapping to FHIR profiles
  - Planning for Jan WGM Connectathon
    - S4G will conduct a number of tests under Scenario 10 of the CG genomics track
  - Discussion regarding VCF attachments on genetic test reports (with JD)
    - O&O discussed last week, will initiate a zulip chat thread to finalize
    - Patrick: Observation.attachment was removed in R4, will likely be back in R5
    - OO and II will have a joint call before the WGM to discuss

Subgroup reports

- Information Modeling (IM) (Bob F)
  - Minutes: https://docs.google.com/document/d/15kBa3HqxQfwwe0Uwgx3UNashTIanpGRiyRVbimE_j3A/edit#
  - Draft model docs: https://docs.google.com/document/d/1Wys14HNJAEB_YJ-EeDPAKX50_oxiDqAKi3WD4wlfjbk/edit
  - Currently working on sequence feature/annotation (in alignment with GKS/VA)
- FHIR (Gil)
  - no meeting yesterday (Dec 3)
  - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.zfi9l8jfe4la
    - Introduced QA sheets, discussed GL String and LOINC codings for HLA
    - Please log QA issues with the IG (grammar, diagrams, recent changes that don't properly reflect their trackers) here

Topic 0: Ballot Sign Up

Announcement of ballot openings for early January 2019 Ballot Cycle

Ballot Period Open/Close Dates

Voting for consensus group members in most ballots will open and close on the following dates. Exceptions for a specific ballot are listed with that ballot description.

Ballot Open Date: Friday, December 7, 2018

Ballot Close Date: Monday, January 7, 2019

Consensus Group

Enrollment Period Important Note:

Consensus group signup closes when ballot voting begins. Consensus group enrollment will be available from a date at least four weeks preceding the ballot vote opening date and will continue until the opening of voting. While the exact dates are dependent upon individual ballot open and close dates, in general the consensus group sign up period dates are as follows:

Consensus Group Sign-Up Open Date: Monday, November 5, 2018

Consensus Group Sign-Up Close Date: Thursday, December 6, 2018

Please be aware that these dates may not be accurate for all consensus groups.

To sign up, point your browser to

http://www.hl7.org/ctl.cfm?action=ballots.home

Important Note:

Consensus group signup will close when ballot voting begins. This is also the final date non-members can sign up for Non-Member Participation in the ballot.

Additional Information:
http://www.hl7.org/documentcenter/public_temp_87AEFCD4-1C23-BA17-0CEC7A7FE30214CE/ballots/2019JAN/Announcements/Formation%20of%20Consensus%20Groups%20for%202019%20January%20Ballot%20Cycle.pdf

CG FHIR ballot artifact is now available: http://www.hl7.org/fhir/uv/genomics-reporting/2019Jan/index.html

Topic 1: Jan 2019 WGM Agenda Planning

- https://confluence.hl7.org/display/CGW/2019-01+CG+WGM+Agenda

Topic 2: Non-ballot tracker items

Tracker	16401	N/A	David Poloway
Summary	inconsistencies regarding ServiceRequest,		
Links	http://build.fhir.org/ig/HL7/genomics-reporting/general.html		
Resolution **Notes**	**Persuasive with mod (non-substantial) - change instances of "ServiceRequest" and "Order for genetic testing" with the profile name ("Request for Genetic Test") to avoid unnecessary confusion**		
Details	ServiceRequest appears here. But the text is "Request for Genetic Test." It is better to make a unified and independent paragraph for the simple introduction of ServiceRequest with a reference link. "Orders for genetic testing" refers to the same thing.		
Follow-ups			

- Reviewed tracker (see link on "tracker" text in above table)
- Motion: accept resolution
    - Move / 2nd: Kevin / Bret
    - Discussion:
        - Andrea: good to be consistent across all services, are there any unique needs from a genetic testing perspective that are not reflected in O&O?
        - Kevin: Is this a different tracker item?
        - Patrick: question about the change of wording in the proposal, would like "Service Request for Genetic Test" to reflect both the name of the service and "genetic test"
        - Kevin: Wanted to be consistent with the name of the profile
        - Updated resolution wording to be more clear
        - Bret: remove "for" (to yield "request genetic test")?
        - Kevin: official name includes "for"
    - Vote: (Abstain / Against / For): Bob M / 0 / 15
    - Result: Motion passes

Tracker	15891	N/A	Kevin Power
Summary	Sequence as a knowledge resource,		
Links	N/A		
Resolution **Notes**	**Not Persuasive: Decided to not make MolecularSequence (PKA Sequence) 'Definitional' at this point. See tracker:16501**		
Details	Review the elements of Sequence and decide what should migrate to Observation - and whether new elements should be added to support its use as a "knowledge" resource		
Follow-ups			

- Reviewed tracker (see link on “tracker” text in above table)
  - Kevin: may revisit someday, but for now we’ve decided not to make Sequence “definitional”
  - Bob M: original submitter was Lloyd
  - Patrick: perhaps this could be a topic at the WGM
    - Kevin: we may need to consider as time allows, probably need a firm proposal before the group revisits
  - Bret: is “definitional” a type of molecular sequence? Should it be a profile?
  - Bob M: definitional vs instance of sequence, this is a whole topic on its own
- Motion: accept resolution
  - Move / 2nd: Patrick / Bob M
  - Discussion:
    - none
  - Vote: (Abstain / Against / For): 0 / 0 / 16
  - Result: Motion passes

Kevin P left the meeting, total voting is now 15

Tracker	16397	N/A	David Poloway
Summary	Add more info regarding definitional sequence,		
Links	http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html		
Resolution **Notes**	**Not Persuasive: Decided to not make MolecularSequence (PKA Sequence) 'Definitional' at this point. See tracker:16501**		
Details	There should be more information about "Definitional Sequence"http://build.fhir.org/ig/HL7/genomics-reporting/sequence.html. It needs to be clearly introduced. Currently, it looks like a reference/link Object, and some fields overlap with Sequence		
Follow-ups			

- Reviewed tracker
  - Patrick: seems to be a duplicate of the last one, do we have to vote?
- Motion: accept resolution
  - Move / 2nd: Bob M / Patrick
  - Discussion: non
  - Vote: (Abstain / Against / For): 0 / 0 / 15
  - Result: Motion passes

Tracker	16403		
Summary	Add examples from IG guidance/STU3,		
Links	http://build.fhir.org/ig/HL7/genomics-reporting/general.html		
Resolution **Notes**	**Duplicate of other trackers requesting examples already approved** **16510 - Need more examples 15893 - Decide what examples to bring across from existing profiles and convert them**		

Details	Add in examples from IG guidance and STU3 to this version
Follow-ups	

- Reviewed tracker (see link on “tracker” text in above table)
- Motion: accept resolution
- Move / 2nd: Patrick / Bob M
    - Discussion: none
    - Vote: (Abstain / Against / For): 0 / 0 / 15
    - Result: Motion passes

Tracker	16399		
Summary	Add examples to 1.6.3,		
Links	http://build.fhir.org/ig/HL7/genomics-reporting/somatics.html		
Resolution **Notes**	**Duplicate of other trackers requesting examples already approved** **16510 - Need more examples 15893 - Decide what examples to bring across from existing profiles and convert them**		
Details	1.6.3 Somatic-specific Example instances should be added		
Follow-ups			

- Reviewed tracker (see link on “tracker” text in above table)
- Motion: accept resolution
    - Move / 2nd: Patrick / Jamie
    - Discussion: none
    - Vote: (Abstain / Against / For): 0 / 0 / 15
    - Result: Motion passes

Did not have time to address the rest of the scheduled issues

Tracker	16409	N/A	David Poloway
Summary	Link definitions to standard ontological terms,		
Links	N/A		
Resolution **Notes**	**Duplicate of 16513 - need glossary.**		
Details	Link definitions to standardized ontological terms (may be done via LOINC links to other ontologies)		
Follow-ups			

- Reviewed tracker (see link on “tracker” text in above table)
- Motion: accept resolution
    - Move / 2nd:
    - Discussion:
    - Vote: (Abstain / Against / For): / /
    - Result:

Tracker	16391	N/A	David Poloway
Summary	Add introduction and examples for Reporting,		
Links	http://build.fhir.org/ig/HL7/genomics-reporting/general.html		
Resolution **Notes**	**Not Persuasive: Already have a placeholder page to build out how to search:http://build.fhir.org/ig/HL7/genomics-reporting/domain.html**		
Details	Lots of reminders for "searching" in all Reports. May be better to make an introduction and examples for searchable functions in all kinds of Report.		
Follow-ups			

- Reviewed tracker (see link on “tracker” text in above table)
- Motion: accept resolution
    - Move / 2nd:
    - Discussion:
    - Vote: (Abstain / Against / For): / /
    - Result:

Tracker	16415	N/A	David Poloway
Summary	Add text to 1.3 sequenced variants Observation-genetics,		
Links	http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html		
Resolution Notes	**Not persuasive (non-substantial) - This IG design prefers components over extensions and should not reference the core profiles for this guidance**		
Details	Add text for Observation-genetics profile perhaps at the end of the section (new section): The Observation-genetics profile Observation-genetics profile is used to interpret variants from sequence resource. Clinical usage may need more specific representation of variant at locus or structural variant in whole genome. Some of the attributes of the profile follow: The observation-genetics Sequence extension will refer to the Sequence resource for sequence information related to this variant. The observation-genetics Interpretation extension will refer to an Observation instance which contains clinical interpretations for the variant described. The code, effective[x], issued, performer, method, specimen elements can be used to describe how the genetic observation (variant and sequence data) is obtained. Other extensions are used to describe attributes of this variant such as Genomics Source Class, Amino Acid Change Type, etc. These are mappings from v2 and Ionic code reference with details can be found in this list .		
Follow-ups			

- Reviewed tracker (see link on “tracker” text in above table)
- Motion: accept resolution
    - Move / 2nd:
    - Discussion:
    - Vote: (Abstain / Against / For): / /
    - Result:

https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16236&start=0

16236	Add background info on sequence

- Reviewed tracker
- Motion: accept resolution
    - Move / 2nd:
    - Discussion:
    - Vote: (Abstain / Against / For): / /

    - Result:

- “Straight-forward” trackers (will tackle as time allows)
    - 16412, 16391, 16409, 16403, 16401, 16395, 16388, 16415, 16396, 16399, 15891

Chat

- Patrick 10:11AM
    - i don't want to start a discussion, but methods is based on Snomed-CT, which we can't use in an international context. (unfortunately)
- Andrea 10:12AM
    - due to licensing, right Patrick?
- Patrick 10:13AM
    - yes. Only the dark "blue" countries have a license:https://www.snomed.org/our-customers/members
- Bob Milius 10:13AM
    - The observation.method is an example
- Patrick 10:13AM
    - i know, just wanted to clarify
- Bob Milius 10:14AM
    - Value Sethttp://hl7.org/fhir/ValueSet/observation-methods is based on snomed, but can use something else
- Jamie Jones 10:19AM
    - I logged GF#19373 to consider adding it to Sequence a while back as well
- Bret Heale 10:21AM
    - makes sense to allow for a pointer (path) or the attchement for VCF as supporting doc for Observation and MolecularSequence.
- Bret Heale 10:21AM
    - meaning, support in both
- Patrick 10:21AM
    - Fhir-I also wants to allign: Attachment, Binary, Media because these resources kind of duplicate a similar functionality
- Jamie Jones 10:24AM
    - There's also DocumentReference.content …
- Patrick 10:25AM
    - yes, which is an Attachment. I think Lloyd mentioned to extend Media to be used more universally
- Kevin Power 10:25AM
    - RE: VCF - First thing to decide is where to attach it. Next decision is what resource should be used to model the attachment
- Bret Heale 10:29AM
    - I favor a standalone, which can be attatched to any observation or MolecularSequence, or any other resource. Why limit where it can be used? A reason could be to have ONE possible path for a VCF file to have (i.e. the location is the only place in the FHIR spec where a VCF can be)...will be a good discussion
- Bob Dolin 10:30AM
    - It can be tricky, to know the genome build for a VCF, or the specific reference sequence for a particular variant call. If there is some metadata we want to accompany the VCF, I wonder if that will influence where we put the attachment?
- Bob Milius 10:31AM
    - need to step away for few min
- Bret Heale 10:31AM
    - e.g. profile off of MolecularSequence called VariantCallFile
- Jamie Jones 10:32AM
    - already can hack it in with the repository fields I believe
- Bret Heale 10:32AM
    - +1 @jamie
- Andrea 10:32AM
    - have folks modeled actual clinical data too? that might help inform.
- Bret Heale 10:32AM
    - @Andrea, depends on what you mean. Current clinical data is constrained by current practice
- Bret Heale 10:33AM
    - I think we've got current practice covered with the current artifacts. what do you think?
- Bret Heale 10:33AM
    - But, between labs and annotation services the VCF is currently exchanged
- Andrea 10:38AM
    - Concur Kevin.
- Bob Milius 10:44AM
    - Back
- Jamie Jones 10:44AM
    - i think changing the profile name could be a different tracker since it would be substantial
- Bret Heale 10:45AM
    - @jaime thanks. good point
- Kevin Power 10:45AM
    - back to VCF: The MolecularSequence.repository option only includes the ability provide a URL / link. I think we should also support an actual attachment as well, not a link to be used for 'smaller' VCFs.
- Jamie Jones 10:45AM
    - i agree
- Bret Heale 10:47AM
    - @kevin that sound good but when exchanged folks will need to make sure the url/link is resovable by the recipient system. That adds a small bit of complexity in terms of logic. Ultimately, exchange will be something all parties negotaite. we can only provide the means.
- Patrick 10:48AM
    - and thinking about actual EHRs which are more likely to implement Observations instead of MolSeq i would attach it inside a observation.
- Patrick 10:57AM
    - http://wiki.hl7.org/index.php?title=FHIR_gForge_Tracker#Duplicate

- Patrick 10:58AM
  - as i understand we don't need to vote on duplicates
- Joel Schneider 10:59AM
  - Is it possible the attachment may need to be referenceable by resources other than the enclosing Observation?
- Bret Heale 10:59AM
  - depends on use case
- Bret Heale 10:59AM
  - need a proper discussion time : $)
- Patrick 11:00AM
  - It is, and can be done as it would be an Attachment Resource. But for interoperability purposes i would profile into an Observation

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