**CG-2022-02-15**

HL7 Clinical Genomics Weekly Call - Feb 15, 2022 11:00 AM (US Eastern)

**HL7 Antitrust Policy**

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**Minutes**

[This document](http://tinyurl.com/HL7CGGroupCall) [Archive]

**Attending the meeting**

[https://us02web.zoom.us/j/2980068716?pwd=eG9VOXdjTU43VGpJVGNNG1qbW1qQT09](https://us02web.zoom.us/j/2980068716?pwd=eG9VOXdjTU43VGpJVGNNG1qbW1qQT09)

Meeting ID: 298 006 8716 // Passcode: 2020

**Agenda**

- Attendees Sign-in
  - Agendas and Important Dates
  - Subgroup reports
  - Internal efforts
  - WG projects and outreach
  - chat.fhir.org (zulip) comments
  - Future proposed agenda topics/requests
- Jira for change requests
- **Topic 0**: Approval of Minutes from Last Meeting
- **Topic 1**: GenomeX Update (Arthur)
- **Topic 2**: Remove 0..0 constraints from all genomics profiles
- **Topic 3**: MolecularSequence
- Chat

**Attendees Sign-in**

Presiding Co-Chair (Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org)

1. Arthur Hermann, Kaiser Permanente, arthur.hermann@kp.org
2. Lloyd McKenzie, Accenture, lloyd.mckenzie@accenture.com
3. Anand Kulanthaivel, Clinical Architecture, anand_kulanthaivel@clinicalarchitecture.com
4. Kevin Power - CMH - kmpower@cmih.edu
5. Liz Amos - NLM - liz.amos@nih.gov
6. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org (until :30)
7. Bob Dolin - Elimu Informatics - bdolin@elimu.io
8. May Terry - MITRE - mayT@mitre.org
9. Willie Chang - Epic - willie@epic.com
10. Mullai Murugan - BCM - murugan@bcm.edu
11. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
12. Kim Peifer - Flatiron Health - kim.peifer@flatiron.com
13. Carla Escobar - BWH Gil Alterovitz’s Lab - carlaesc@live.unc.edu
14. Alex Mays - CAP - jamays@mgh.harvard.edu
15. Aly Khalifa - Mayo Clinic - khalifa.aly@mayo.edu
16. Bret Heale - bheale@humanizedhealthconsulting.com

**Agendas and Important Dates**

<table>
<thead>
<tr>
<th>CG Call Date</th>
<th>Co-Chair</th>
<th>Agenda <a href="https://confluence.hl7.org/display/CGW/Future+Topics+for+Weekly+Meetings">https://confluence.hl7.org/display/CGW/Future+Topics+for+Weekly+Meetings</a></th>
<th>Important Dates</th>
</tr>
</thead>
<tbody>
<tr>
<td>1-Feb-22</td>
<td>Kevin P</td>
<td>• WGM Minutes Review (quick) • MolecularSequence</td>
<td>7. Feb - Last week for STU2 changes</td>
</tr>
</tbody>
</table>


### Subgroup reports

- **Information Modeling (IM) (Bob F)**
  - Minutes:
    - HL7 CG Information Modeling - Minutes for Oct 2019 to Jan 2020+
    - slides in minutes
    - Developing a FHIR logical model from our conceptual model (see clinfhir)
      - Sequence resource
      - Variation resource
      - Structural variation: in progress, on hold until after WGM
    - Draft of new Sequence resource in clinFHIR
      - http://clinfhir.com/
      - Change to the "for developers" tab
      - Open the "logical modeller"
      - Click on the "all" tab
      - Scroll down to find the MolecularSequenceV2 model
      - Note: all clinfhir models related to this work start with "Molecular"
    - Draft FHIR Resources: Molecular Sequence & Molecular Variant
      - Please review and provide feedback!!

- **Trying new call schedule (every other week) - next one is Feb 24**

- **FHIR (Jamie)**
  - Minutes:
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_4sJZ9yXbLMGOqPXHPtSLLQ/edit?pref=2&pli=1#heading=h.p1e5sxbkjxz9
    - tinyurl.com/thigenomics
    - “IG Lite” update is live: http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html

- **OO Work (JD)**
  - Pending work (perhaps) pending CG decision on Procedure/Molecular Study/etc.
  - new Transport resource!

### External efforts

- **GenomeX Accelerator (hosted through CodeX) (Arthur Hermann)**
  - Arthur to provide updates as needed

- **GA4GH (Global Alliance for Genomics and Health)**
  - Genomic Knowledge Standards (GKS) Work Stream (leads: Bob Freimuth, Andy Yates)
    - Variant Representation subgroup (lead by Larry Babbi/Alex Wagner)
- See ongoing project board at https://github.com/orgs/ga4gh/projects/5
- VRS 1.3 (structural variation) anticipated by Dec 2021
- Variant Annotation subgroup (lead by Matt Brush and Javi Lopez)
  - Working on statements about therapeutic interventions and variant pathogenicity
  - https://docs.google.com/spreadsheets/d/1zqLU-YvqgB7HkOVsTh-74BwtdgB9KQpKcWkSHZoQ/edit
- V0 release: 4 core substatement types + generalized model
  - Due "soon"
- Sequence Annotation subgroup (lead by Karen Eilbeck and Shawn Rynearson)
- Clinical & Phenotypic Data Capture Work Stream
- Pedigree Activity (Grant Wood)
  - help GA4GH community to understand HL7 Pedigree standards
  - https://docs.google.com/spreadsheets/d/1AxRufaSICADO_F9CAhbkVh5dChT4FjH4ZgvGFFdC-2W/edit
- Phenopackets
  - Under continuous development
  - Currently going through the ISO
- G2MC (Grant Wood)
  - Looking to develop AI tool for literature review for poly-genetic risk, generate guidance
  - Create KB on G2MC website
- ClinGen/ClinVar (Larry Babb)
  - Data Exchange/Platform WG, major driver project of GA4GH GKS
    - will discuss further when Larry in on a call
- ONC Sync for Genes (Bob Freimuth)
  - ONC website: https://www.healthit.gov/topic/sync-genes
  - Phase 4 has ended
- ISO TC/215 Genomics Subcommittee (Liz, Clem, Bob F)
  - Canada is sponsoring a phenopackets spec in ISO, will be based on the GA4GH spec that was approved fall 2019
    - The technical experts group just convened to work on this (mid Aug)
  - Since Phenopackets is implementing the GA4GH VR spec, which we are attempting to keep aligned with the CG IM, this work will be of interest to the CG WG
  - Arthur to review for other standards that might overlap with CG
  - Bob M / Bob F: Liaison through HL7
  - Cannot share documents, but we can talk about them
- mCODE (May Terry)
  - Last few weeks looking to simplifying how biomarkers are represented
  - FHIR & OMOP collaboration
  - zulip streams
  - OMOP and FHIR https://chat.fhir.org/#narrow/stream/286658-omop.-2B.20fhir
  - OMOP and FHIR Oncology https://chat.fhir.org/#narrow/stream/302239-omop.-2B.20fhir.20oncology
  - OMOP and FHIR Terminologies https://chat.fhir.org/#narrow/stream/306130-OMOP.-2B.20FHIR.20Terminologies
  - May has a use case for OMOP extensions to their common data model
  - can present to work group, perhaps at WGM
  - presented at weekly OHDSI genomics mtg
  - Genomics Reporting IG is much deeper than the OMOP genomics data model
  - Bob D: we should talk to OMOP people about the relationship between nucleic acid changes to protein changes. Need to have widespread understanding that protein changes are almost always inferred from genetic changes.
  - Need to have a discussion on
    - fusion events
    - Change in mCODE’s way to do clinical significance
  - not december, but prior to or during the WGM

**WG projects and outreach**

- May: indirect outreach with OHDSI/OMOP
  - trying to standardize on representation on variants
  - should CG have a formal presence there? - probably
  - When do they meet? every Tues at 9AM
  - Can they present to the group? they have great stuff they can teach us
  - Dr Joseph Murray exploring ClinGen API for retrieving structured variants.
  - OMOP CDM Oncology WG – Genomic Subgroup meetings are traditionally scheduled for every Tuesday at 9 am ET. Meeting Link [here](http://example.com)
  - Next meeting on Jan 6
  - OMOP Genomics - how to have a common approach when there are so many knowledge bases (FHIR and non-FHIR)
    - Bob D: they are challenged with same things we are challenged with. Need a unified strategy.
    - May: potential opportunity for working together. Assumption is we know specific variants looking for. Not exploratory. Closed world environment.
    - Bob D: they need to develop an appreciation of the power of operations
    - Next meeting is next week, then bi-weekly (9am)

**chat.fhir.org (zulip) comments**

- Genomics
  - If you would like to bring up a topic for discussion, Zulip is the platform and the stream is genomics - https://chat.fhir.org/#narrow/stream/179197-genomics

**Future proposed agenda topics/requests**
• WG members: please use this section to request topics to cover on our WG calls
  
  • https://confluence.hl7.org/display/CGW/Future+Topics+for+Weekly+Meetings
  
  • Needs review/clean up
  
  • [ ] todo: Grouper could be replaced with just hasMember

Jira for change requests

• If you like to open a ticket, Jira is the platform - https://jira.hl7.org/secure/Dashboard.jspa?selectPageId=12002

Topic 0: Approval of Minutes from Last Meeting

• https://confluence.hl7.org/display/CGW/CG-2022-02-01
  
  • "You have received the minutes. Are there any corrections to the minutes? (pause) Hearing none, if there are no objections, the minutes are approved."
  
  • Corrections: none
  
  • minutes are approved

Topic 1: GenomeX Update (Arthur)

• Update on the GenomeX/CodeX FHIR Accelerator
• PDF Overview “One Pager”
• GenomicX confluence
  
  • https://confluence.hl7.org/display/COD/Genomics
  
  • https://confluence.hl7.org/display/COD/Proposed+Initial+Use+Cases
• Current use cases
  
  • Lab sending data directly to EHR
  
  • Lab/org sending genomic info to healthcare or research org’s genomic data repository
  
  • FHIR Genomic Operations

Topic 2: Remove 0..0 constraints from all genomics profiles

• https://jira.hl7.org/browse/FHIR-35907?focusedCommentId=196992&page=com.atlassian.jira.plugin.system.issuetabpanels%3Acomment-tapanel#comment-196992
• There are a number of additional changes that came up in following up in the spirit of the request
• see "Full list of additional changes made":

<table>
<thead>
<tr>
<th>Change</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Remove GenomicsServiceRequest profile</td>
<td>After making approved changes, only two things were left in the GenomicsServiceRequest profile:</td>
</tr>
<tr>
<td></td>
<td>• ‘doNotPerform 0..0’ – It seems the intention was to remove the 0..0 that didn't have a really valid reason, and I am not sure we have a valid reason for this one?</td>
</tr>
<tr>
<td></td>
<td>• ‘code ^binding.description = &quot;For laboratory, LOINC is preferred.&quot;’ - The value here is really, really small, and doesn't seem to warrant creating a profile?</td>
</tr>
<tr>
<td>Remove ‘SupportingInformation’ extension and replace with standard extension ‘workflow-supportingInfo’</td>
<td>Approved change to to reset the ‘ServiceRequest.supportingInfo’ to remove our constraints. Then we wanted to reset our own “SupportingInformation” extension used on GenomicsReport, which aligned to the attribute on ServiceRequest. However, there is a standard extension in R4 called ‘workflow-supportInfo’ so have just removed our locally defined extension since it looks exactly the standard extension.</td>
</tr>
<tr>
<td>Removed constraint on GenomicsReport.subject</td>
<td>We were limiting to Patient,Group,Location (as we did on GenomicsSpecimen), so it seems to make sense to make the same change here.</td>
</tr>
<tr>
<td>Drop 0..1 constraint on GenomicsReport.performer</td>
<td>Not sure why we had this constraint, so proposed dropping it</td>
</tr>
<tr>
<td>Drop constraint on GenomicsDocumentReference.subject</td>
<td>We were limiting to Patient, Group so it seems to make sense to make the change here</td>
</tr>
</tbody>
</table>
Topic 3: MolecularSequence

- https://tinyurl.com/MolSeqR5
- https://docs.google.com/document/d/1gjGrE1Lzg22Wlf20gaUB8yBnniwUW2IDaYSveDiO-98/edit?usp=sharing

Proposal
- remove at least two elements: quality, structureVariant
- remove FDA examples which contain quality (3)
- remove or rename Graphic examples (5)
- (there are no examples using structureVariant)

Question: Do we want to make it definitional? e.g., remove the following
- patient
- specimen
- device
- performer
- readCoverage
- quantity
- remove/edit examples that contain these

Question: Do we want to rename 'variant' to 'relative', to reduce confusion with Variant Profile?
- need to change all examples containing this

Need to create a message to ballots explaining changes

Chat

- 10:01:00 From Bob MIlius to Everyone:
  - https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_i5Ki72NEBYXZuwhFg/edit#heading=h.x0spkcire0b
- 10:04:58 From Bob MIlius to Everyone:
  - sign in here: https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_i5Ki72NEBYXZuwhFg/edit#heading=h.2r1vih7cx10
- 10:22:02 From Bob MIlius to Everyone:
  - GenomeX often uses "Genomics IG" when name is actually "Genomics Reporting IG"
- 10:22:53 From Bob MIlius to Everyone:
  - Laboratories and LIMS vendors are crucial.
- 10:23:03 From May Terry to Everyone:
  - Good point, Bob. This is a 'reporting' IG and needs to be called out as such.
- 10:23:41 From we are the crazy micguf to Everyone:
- 10:25:21 From we are the crazy micguf to Everyone:
  - pardon, just noticed my kids messed with my settings. I'm Bret. signing off and will be right back : ^ { 
- 10:27:18 From Bob MIlius to Everyone:
  - Can you share what organizations have expressed interest?
- 10:35:00 From Liz Amos to Everyone:
  - what about federal partners?
- 10:36:18 From Bret H to Everyone:
  - Great points. Application developers will be a good group to engage. There's a different focus depending on where you are touching the data.
- 10:37:26 From Anand Kulanthaivel to Everyone:
  - Agreed, we encounter too many organizations where a majority of their patient genomic data is stored purely as PDF.
- 10:38:25 From May Terry to Everyone:
  - There is interest but it is unclear from every discussion of the level of genomic detail that is necessary to send. That I suppose will be determined once there are "next step" discussions.
- 10:39:16 From May Terry to Everyone:
  - So...echoing Bret's comments. It's a general advertisement for the Labs to EHR discussion with not much behind it yet.
- 10:45:00 From Bret H to Everyone:
  - NHGRI can help in the research space, but I bet that's on the list of folks to connect with. Importantly, the confluence page is a great step forward as it gives us something we can use to help recruit and build interest broadly on the effort.
- 10:45:38 From Liz Amos to Everyone:
  - well, genomics also touches all NIH institutes and centers (including NLM!)
- 10:47:36 From May Terry to Everyone:
• Accelerators are not just a validator of an IG. https://www.hl7.org/about/fhir-accelerator/

• 10:48:06 From Kevin Power to Bob Millius (Direct Message):
  • If possible, can I have the last 5 minutes (at least) to discuss my topic?

• 10:48:09 From Bret H to Everyone:
  • Is Dinvici being connected with regards prior auth? Just thinking out loud

• 10:49:02 From Alex Mays to Everyone:
  • Echoing some earlier comments, I do think there’s value in recruiting LIMS and middleware vendors in FHIR genomics. I agree that current laboratory processes are more bespoke, but as NGS testing goes into more mass adoption there is more demand for “commercial off the shelf” options such as would come from those vendors

• 10:49:02 From May Terry to Everyone:
  • @Bret - yes.

• 10:52:59 From Kim Peifer to Everyone:
  • When does an accelerator dismantle? Does an accelerator work towards clearly defined end points and then dissolve once reached?

• 10:54:07 From Mullai Murugan to Everyone:
  • We are one of the genome centers for AoU. I am curious about AoU potential participation

• 10:54:09 From May Terry to Everyone:
  • @Kim - In all honesty, I don’t know that any accelerator has come to that point.

• 10:54:31 From May Terry to Everyone:
  • That’s because the use cases keep evolving.

• 10:56:09 From Kim Peifer to Everyone:
  • That is interesting

• 10:57:39 From May Terry to Everyone:
  • Some however are very successful in their level of maturity for certain data products created through the accelerator. e.g.: Da Vinci and HREx which is foundational for other DaVinci IGs. Lloyd can speak to that. :-)

• 10:59:36 From Arthur Hermann to Everyone:
  • There was a comment earlier about the name of our IG which is called: Genomics Reporting IG - I started Zulip thread about that. I am not clear that this is the proper name for our IG at this point in time! Please go find it and speak up