

HL7 Clinical Genomics WGM - September 2017, San Diego

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## Mon Q3 1:45-3:00 PM

Co-chair - Gil

- [Slides](#)
- Gil presented review
  - how CG FHIR was developed - informed by Domain Analysis Model Document
  - incremental adoption of FHIR - two directions
    - pushed from NGS labs
      - start with whole sequence/reads, add interpretation as needed
    - traditional labs
      - start with Observation, add detail as needed
    - pyramid slide
  - FHIR Genomics Implementation Guidance Document
    - <https://www.hl7.org/fhir/genomics.html>
  - Links to other resources/profiles
- EMR <-> Genomics Lab
  - order panel or sequencing
  - send data to clinical genomics app
    - SMART on FHIR
  - Clinical care and/or Research studies
- FHIR Connectathon Review
  - HAPI/test.fhir.org/WildFHIR/Furore and all provided client-based solutions and/or implemented on their server in two days for our profiles, extensions, resources
  - Feedback from Sync for Genes - easier to use with hierarchy as we voted for in summer.

- Testing interoperability - for each server
  - python scripts
  - touchstone scripts
  - manually ran on each server for all elements and take screenshots
- Running test scripts on servers
  - Aegis - all successful
  - had issue with one server involving assertion to delete profile in the server, check that deletion, and then recreate. It turns out one server caches the search results so even if deleted, it returns it.
- Review and feedback on spec
- Testing Sync for Genes
- Current work
  - implementation testing at connectathon to move to Level 2
  - looking over results
  - examining criteria of Level 3
  - analysis of VMC modeling effort, currently focusing on identifiers (done as CURIE in VMC)
  - finishing V2 to FHIR converter
  - scripts to test if compliant with specification/interoperable
  - syncing with new DAM use cases
- Update on Sync for Genes
  - phase 1 complete in June 2017
    - hope to release report at end of month
  - ONC interest in supporting phase 2
- FDA Collaborations
  - precisionFDA (data from FDA)
  - Biocompute Object (BCO) (data into FDA)
    - a specification for recording NGS analysis for FDA use in clinical trial submission/research studies reproducibility. Uses FHIR's genomics profiles/resources for clinical genomics information linkage FHIR servers/EHRS.
    - [https://hive.biochemistry.gwu.edu/prd/htscsrs//content/slideDecks/10\\_Ma\\_zumder.pdf](https://hive.biochemistry.gwu.edu/prd/htscsrs//content/slideDecks/10_Ma_zumder.pdf)
- FHIR gforge reconciliation
  - <https://gforge.hl7.org>
  - filter for Clinical Genomics
  - #13829
    - add new search parameters at resource
    - motion - Bob M
    - 2nd?
    - 1 abstain
    - 0 nay
    - 13 yea
  - #13830

- search parameters for observation genetics profile
- Prep for Joint with FHIR

## Mon Q4 3:30-5:00 PM

Co-chair - Gil

- Joint with FHIR-I
- Josh Mandel
  - big picture
  - publication of R4 at end of 2018
  - additional process for some resources will have a normative ballot
    - as part of R4, only parts will be normative
    - feature freeze by end of this year
    - ballot next May 2018, then post, will group into different parts for separate balloting
    - groups will be associated by interdependence of resources
    - followup ballot in Sep 2018
    - could get 4-5 cycles before Normative
    - trying to separate content into where balloting is successful
  - SMART on FHIR
    - currently being balloted by HL7
    - 200+ comments
  - ANSI and ISo
    - check with Lloyd
  - identifiers vs ids
    - ids are local to server, not global
    - identifiers
      - business
      - medical identifiers
      - ssns
      - donor ids
      - lab ids
      - etc
  - Capturing CURIE (Compact URI) identifiers in FHIR
    - <https://www.w3.org/TR/curie>
    - FHIR can't do it now
    - need to have a use case
  - from connectathons to workflows - test more full scenarios
  - CLIA/CAP
    - talk to Orders and Observations
    - look at LRI
  - What is in Scope for FHIR - does it include clinical research, clinical trials, FDA, etc
    - includes all things related to clinical care, eg financial, research, etc

Comment [1]: Action Item:

- FHIR scope has always included these
  - gForge comments (use classic mode)
    - unable to make comments, must use "Classic Mode"
  - Search params- use discretion and make sure they are useable things
    - would like to make everything a search parameter, but not practical for performance issues

## Tue Q1 9:00-10:30 AM

Co-chair – Bob M

- Introductions
- Review of Clinical Genomics activities for newcomers
  - See slides
- Review Agenda
- Review of external efforts
  - Deferred until Q3
- CIMI & Clinical Genomics - Intro by Amnon
  - See slides
- Reviewed CG projects on the WG page
  - CDA implementation guide (460)
    - Should this remain available in case it has been adopted?
    - Should it be used to inform new development on a new CCDA spec?
    - AI: ask SD cochairs how we can archive this project (what are our options) without removing the product
      - The project expired/ended in 2009
      - The CDA spec that resulted from this project is listed on the "product" page
  - Connection with decision support (564)
    - Joint with CDS
    - AI: to SD - need to remove the project
    - We need to maintain a link to the CDS WG
    - AI: CG to contact CDS WG and ask how we can work together going forward
      - What is needed? Joint project?
  - Specimen handling CMET (565)
    - This needs to be removed
  - Specimen CMET 2nd release (682)
    - Owned by O&O (CG is co-sponsor)
    - This should remain (but it may need to be renamed by O&O since it references CMET)
    - AI: clarify with O&O
  - Family health history and pedigree (844)
    - This needs to be removed
  - CG DIM (1050)

Comment [2]: add link to doc (when available)

Comment [3]: add link to Stan's slides (when available)

Comment [4]: Action Item:

Comment [5]: 21-Sep: Email to Melva asking about our projects page.

Comment [6]: Action Item:

Comment [7]: Action Item:

Comment [8]: Action Item:

Comment [9]: Action Item:

Comment [10]: Action Item:

- Discussion deferred to Wed Q3
  - FHIR for CG (1110)
    - Continued as project 1217
    - Project is closed so this can be archived
    - AI: to SD - what do we do with this
  - SDC FHIR profile (1104)
    - Owned by O&O
    - AI: touch base with O&O to get an update on this, if they need anything from CG
  - Genetic variation LOINC-qualified model (1165)
    - Project is closed, future work is going into LRI
    - AI: update status, how do we archive this project (artifact is listed on our products page)
  - FHIR for CG (1217)
    - Ongoing, active project
    - End date is listed as May 2017
    - AI: do we need a new PSS?
  - CG DAM (1254)
    - End date is listed as Jan 2017
    - AI: do we need a new PSS? What do we do with this?
    - Project is ongoing: Target end date Sept 2018
  - LRI (1294)
    - Owned by O&O
    - No changes needed
- Planning of Roadmap for CG Workgroup
  - Loosely discussed as part of reviewing projects

Comment [11]: Action Item:

Comment [12]: Action Item:

Comment [13]: Action Item:

Comment [14]: Action Item:

Comment [15]: Action Item:

## Tue Q2 11:00-12:30 PM

Co-chair - Gil

- Domain Analysis Model (DAM) update
  - <http://tinyurl.com/damcqdoc>
  - See [slides](#)
  - CG WG Target dates from slides (used to make sure we make the HL7 deadline)
    - Aug 31, 2017: Topics Finalized
    - WGM: Share/Review Content/Additions
    - Oct 1, 2017: NIB
    - Oct 15: Comment period ends
    - Nov 7, 2017: Content finalized, Review period begins on frozen content.
    - Nov 14, 2017: Voting
  - HL7 milestones (subject to change)
    - <https://www.hl7.org/special/committees/publishing/schedules.cfm>
    - [Download the Ballot Countdown Schedules for January 2018](#)
      - 2018 JAN cycle deadline to submit the online PSS is Sunday, September 24, 2017.
      - Notification of Intent to Ballot deadline – Sunday, November 12th. Due via online form on website.

- Discussion regarding timeline for incorporation of the model into the document
- CAP liaison to HL7: Dr. Jim Harrison
  - Andrea could potentially get a copy of the new requirements
  - Changes come out annually; consider reviewing them as part of our Sept WGM
- Next iteration of DAM?
- Review PSS for 1254 (see Q1)

## Tue Q3 1:45-3:00 PM

Co-chair Bob M

- See [slides](#)
- Family History - Grant
  - National Family Health History Group
    - Bob Wildin left NHGRI
    - Nothing new to report
  - ItRunsInMyFamily supports families creating, editing, and sharing the same record
    - started by student in Biochemistry U of Utah, he's now at U of ?
    - has a feature to create a family history that can be shared by the family; social networking feature; not many tools have this feature
  - Pilot getting 3000 FHIR pedigree messages to Huges RiskService to obtain the risk scores (ran locally rather than directly from Intermountain Healthcare)
    - designed by Kevin Hughes
    - original service send pedigree in V3 msg to risk service; several risk modules; return back a risk score
    - convert FHIR to V3 and send to risk service, send back in FHIR
  - The last piece is to get some of the messages built using genomic observation (rough 30 or so) using dummy genomic data
- G2MC review – Grant
  - clinical, front line, not research
  - 1st call of Health IT/Bioinformatics work group Wednesday morning (6AM) (tomorrow)
    - logistics review of WG calls by members (confirm or change date/time)
    - re-cap from Athens meeting
    - updates and next steps for Family Health History pilot project with Oracle
    - WG members share their desires for the project
    - Outlining planned deliverables and timelines for this WG
  - Countries include US, Canada, Australia, Chile Columbia, UK, Greece, Israel, Japan, South Korea, Malaysia, South Africa
- Review of GA4GH coordination – Grant
  - Co-ordination - 1st Tier
    - G2MC
    - HL7 CG
    - DIGITize

Comment [16]: Add link when doc is posted

- Precision Medicine Initiative
    - Genetic Alliance
    - eMerge
    - IGNITE
    - ClinGen
    - CSER
    - Other international groups (Genomics England, Swiss Institute of Bioinformatics, etc)
  - create simple search into other databases
  - over 450 organizations
  - Director - Peter Goodhand
    - felt need to coordinate effort
    - asked Grant to lead this coordination
    - gather roadmaps and compare
  - 2nd Tier
    - EHR vendors
    - Genomics management system vendors (clinical, vendors)
    - Labs
    - Sequencing instrument vendors
    - BioPharma
    - Patient advocacy groups
    - healthcare consumer activities
    - registries
    - capital investment, emerging technology
    - policy making activities
  - annual meeting next month (Oct) in Orlando
    - massive reorganization
  - We (HL7 CG) needs to develop a roadmap that we can share
  -
- DIGITizE review – Grant
  - working with FHIR Foundation on merger
  - no pilot teams call in several months
  - added IG to National Academies website - Establishing connectivity and clinical decision support rules for patients carrying MLH1, MSH2, MSH6, and PMS2 (Lynch Syndrome) variants;
    - An Implementation Guide; 14 August 2017; CSER EHR working group
    - describes V2 msg
  - plan on using FHIR in future
- WHO update – Grant
  - joint meeting with WHO LMIC group in Madrid in May WGM
  - will speak at HL7 Genomics Policy Summit
  - no further communication since Madrid meeting
  - They are now focused on how to provide educational tools to LMICs
  - WHO is developing a team focused on genomics

- Review comments for Pedigree - Grant
  - REAFF\_V3\_CG\_R1\_N1\_2017MAY\_chris\_hills\_2017051204808.xls
  - reaffirmation of normative Pedigree
  - In reaffirmation document can't change, so comments should be considered for future use
  - All comments discussed, dispositions added to xlsx doc
  - Motion to accept dispositions/second: Grant Wood/Rosalyn Ryan
  - Vote
    - For: 15
    - Against: 0
    - Abstain: 0

**Comment [17]:** Nona Hall  
[http://wiki.hl7.org/images/0/0a/Information\\_Models\\_and\\_Tools\\_Integration\\_HL7-PSS\\_2016-10-25.pdf](http://wiki.hl7.org/images/0/0a/Information_Models_and_Tools_Integration_HL7-PSS_2016-10-25.pdf)

## Tue Q4 3:30-5:00 PM

co-chair - Kevin

- Q3 items rollover (if needed) - Grant
  - All items completed during Q3
- Product adoption survey discussion - Kevin
  - <https://docs.google.com/forms/d/e/1FAIpQLSc12eSnZnUSsfN5cWRV7mD3-HfK5wci71wDJlinxn6UWvMAYA/viewform>
  - Made several updates to the form in real time



# Wed Q1 9:00-10:30 AM

Co-chair – Bob F

- WG docs
  - DMP
  - <https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdijtMCvnRns4srlDe0EBs0FI/>
    - defer reference to subgroup process in next iteration of DMP
    - added WGM agenda change notifications to include update of a shareable document (eg google doc) that support notifications
    - 5.1 clarification of "decision making" vs "direction making" deferred to next DMP
    - 5.3 Presiding Chair Vote - deferred changes to the next DMP.
    - 5.3 Decision Making Requirements - asked the PIC to define "Material interest" (is it personal interest?)
    - 5.6 Consensus Decision - we will leave the two-thirds (66%) majority, re-evaluate in upcoming DMP changes
    - 5.7 Electronic Voting - voting period and quorum definition - will leave the definition as is
    - vote to accept changes
      - motion to accept DMP = Bob Milius/Kevin Power
      - abstain = 0
      - nay = 0
      - yea = 6
      - results = passes
  - SWOT - vote to accept changes
    - motion to accept SOW = Bob Milius / Kevin Power
    - abstain = 0
    - nay = 0
    - yea = 6
    - results = passes
- Product adoption survey (continued) - Kevin
  - <https://docs.google.com/forms/d/e/1FAIpQLSc12eSnZnUSsfN5cWRV7mD3-HfK5wcj71wDJlinox6UWvMAYA/viewform>
- CG WG roadmap (initial discussion) - deferred to Q2
- WG business
  - DESD update (upcoming deadlines) - Bob M
    - DESD did not receive deadlines yet
    - see ballot calendars at <http://www.hl7.org/special/committees/publishing/schedules.cfm>
    - for NIB, two documents have two different dates, Sep 24 & Nov 12; need to get clarification
  - WGM planning (room reservations) - deferred to Q2
  - Telecom logistics and timing - deferred to Q2

**Comment [18]:** After vote:  
1. submit to PIC and HL7 webmaster  
2. post to WG docs page

**Comment [19]:** Action Item:

**Comment [20]:** Survey sent to small pilot group for final testing.

**Comment [21]:** Action Item:

**Comment [22]:** I have sent an email to Karen @ HL7 to clarify.

**Comment [23]:** From Lynn Laakso: The PSS deadline is September 24 and the NIB deadline is November 12

## Wed Q2 11:00-12:30 PM

Co-chair – Bob F

- Carry-over from Q1 (if needed)
- CG WG roadmap (initial discussion)
  - Existing products
  - Ongoing projects
    - Cancer Interoperability
      - <https://docs.google.com/document/d/15iLiNyoUy2Sigig5Coiw7N0tT9PCPEx22Y3rstUO0/edit>
  - SWOT analysis (notes taken in this document):
    - [https://docs.google.com/document/d/18L\\_5K5TDaq3qOWpvsvr07LNwT-nYSNCCw3K2Xj7xWk0s/edit?usp=sharing](https://docs.google.com/document/d/18L_5K5TDaq3qOWpvsvr07LNwT-nYSNCCw3K2Xj7xWk0s/edit?usp=sharing)
    - Discussed weaknesses and opportunities
    - Will discuss threats at a future time
- WG business
  - WGM planning (room reservations)
    - Consensus: keep same schedule (Mon Q3 through Wed Q4)
  - Telecom logistics and timing
    - Keep as is
- Open Discussion (competes with joint meeting with O&O)

Comment [24]: Action Item: Use these items to draft a roadmap?

Comment [25]: I drafted a roadmap and sent to Bob F for review.

## Wed Q3 1:45-3:00 PM

Co-chair – Amnon

- VMC – variant model collaborative review - Bob F
- GA4GH update (GKS WS) - Bob F
- Collaboration with CIMI - Amnon
- Information Modeling subgroup (PSS/roadmap) - Bob F

Comment [26]: Action Item: Determine if we need a PSS or something?

Comment [27]: Action Item: Determine if we need PSS.

## Wed Q4 3:30-5:00 PM

Co-chair – Kevin

- LRI/V2 lite – Clem leading discussion
  - Discussion points:
    - Answer's documentation will be reworded to say it may include things other than LOINC answers when needed. Driven by O&O
    - OBX 4 grouping - O&O still discussing the usage of the group.
    - NOTE - No actions needed by CG at this time
- Modeling FHIR <-> V2 (Lloyd will attend)
  - Alternative to fit genomics data that is more compatible with V2 and utilizes more existing FHIR structure

- Note about 'Observation.component' and grouping:
  - `Observation.component` is used for any supporting result that cannot reasonably be interpreted and used outside the scope of the Observation it is a component of. Components should only be used when there is only one method, one observation, one performer, one device, and one time. For example, systolic and diastolic blood pressure are represented as a single Observation (e.g. [Blood pressure panel](#)) because the two are almost always produced and interpreted together. Note that the `component.code` may in some cases only be able to be understood in relation to the `Observation.code` (for example, see the [\\$stats operation](#)).
  - Basically, component is a simple, built-in way to extend observation, but for things that fit the rule above
- Since Observation.component is built in, likely to have more systems that can support it out of the box, versus supporting the extensions.
- Observation.component becomes a completely flat list, components are not related to each other. Must use the human readable definitions to see those relationships
  - Could use some constraints (meaning components that must come together or cannot come together ...)
  - Additional structure would need to be Observation->Observation or through an extension
- If structure is needed to group related components together, use an Obs that points to another Obs (it is not possible to maintain specific relationships between two types of components that have a many-to-many scenario)
  - Try to use Obs components rather than extensions so that FHIR clients can support more data (extensions could get dropped on the floor)
- NOTE - Here is a Tracker where a very similar topic was discussed, and extensions were decided on over component:u
  - [https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker\\_item\\_id=11318&start=0](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=11318&start=0)
- WGM Review
  - WGM feedback (survey?)

Comment [28]: Action Item: