

Contents

[Mon Q3](#)

[Mon Q4](#)

[Tue Q1](#)

[Tue Q2](#)

[Tue Q3](#)

[Tue Q4](#)

[Wed Q1](#)

[Wed Q2](#)

[Wed Q3](#)

[Wed Q4](#)

Mon Q3

Co-chair - Gil

- 17 attendees
- Lam has resigned as co-chair. His slot will be filled at next WGM co-chair elections. Do we want to have a interim co-chair?
 - Motion by David Kreda/2nd by Bob Milius: Send email to CG workgroup listserv:
 - do we want to have interim co-chair - yes or no
 - if yes, do you want to be an interim co-chair?
 - will have a vote in two week for a interim co-chair (from those throwing their hat in the ring) until co-chair election next January
 - resolve by next Tue call (Oct 4).
 - abstain - no
 - no - o
 - yes - passes unanimously
 -
- FHIR Genomics Review for newcomers by Gil
 - discussion re SMART and FHIR
 - SMART can be work with FHIR servers like HAPI
 - Bob M - should the group propose a SMART project?
 - David K - we should focus on FHIR transport mechanisms, that's the thing that helped SMART explode
 - Grant W - we should think about this, maybe have a showcase project that uses SMART to show how FHIR genomics can be used

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- FHIR Connectathon Review
 - Gil reviewed slides presented at connectathon
 - HLA
 - Joel - terminology server for HLA nomenclature
 - A link to the proof-of-concept HLA terminology service (currently being updated for STU3):
<http://mac-and-fhir-prototype.us-east-1.elasticbeanstalk.com/doc/>
 - Bob - resource instances for HLA typing report - use case
 - Pharmacogenomics
 - Martin -
 - haplotype representation system being designed (terminology service, codable concept encodings, etc)
 - integrated pharmacogenomics work into Genetic Observation profile with links to Sequence Resource
 - Testing
 - Richard from Aegis
 - AEGIS.net migrated scenarios for STU3 ballot
 - uncovered potential issues in fluent validation engine on Observation resource constraints - going from DSTU2 to STU3
 - Open source FHIR genomics server
 - implementation in python
 - virtual machine, docker
 - Gil's group
-
- Prep for Joint with FHIR
- FHIR Ballot reconciliation started

Mon Q4

Co-chair - Gil

- Joint with FHIR
- Grant W - shout out to AEGIS for working with us on testing!
 - Gil - Richard worked at CG table during connectathon; another group doing testing is MITRE
- Target date for ballot content changes - Nov 27
- How to reconcile ballots
 - gforge tour
 - quick filter
 - reviewing work group - Clinical Genomics
 - status = any open status
 - hit browse

- can share url, but user needs a login for it to work
- #8674
 - Lloyd
 - submitted a year ago
 - enter followup
 - send email to Lloyd for clarification
- #11011
 - Gil
 - allow for "type" under "quality"
 - FDA wanted to include information
 - write example, long description, short description, value set
- #11012
 - Gil
 - reference to workflow/provenance information
 - write example, long description, short description, value set
- #11013
 - Gil
 -
 - discuss, clarify, and create example for PGx
 - observation for genetics
 - need clarification
- Discussed process issues about moving forward.
- #11014
 - family history genetics profile
 - Gil
 - add genetics pedigree example
 - find someone to write the example
 - populate text; replace To Do with narrative
 - Lloyd created this
 - Grant will follow up
- #11015
 -

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Tue Q1 9:00-10:30AM

Attendees:

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
3. Terry McDonnell - Syapse - terrym@syapse.com
4. Joel Schneider - NMDP - jschneid@nmdp.org
5. Michael Martin - AAVLD - mkm1879@gmail.com

6. Tessa van Stijn Nictiz - stijn@nictiz.nl
7. Elizabeth Newton - elizabeth.h.newton@kp.org
8. Vijay Shah - ONC/JBS - vshah@jbsinternational.com
9. Grant Wood - grant.wood@imail.org
10. Byoung-Kee Yi - Samsung Medical Center - byoungkeeyi@gmail.com

Co-chair – Bob M

- Review Agenda
- Review of Clinical Genomics activities for newcomers
- Review of external efforts
- Review and Planning of Roadmap for CG Workgroup
 - Maybe this should be done at end of meeting, in light of everything we've learned?
 - Amnon - comment from CTO at co-chair dinner, we need a migration path from one standard to another (eg V2 <--> FHIR), in the long run all standards will merge
 - Bob - "Strategy is knowing what not to do"; "we can do anything, but we can't do everything"
 - Grant - would like get a roadmap from this group that we can share
 - Bob and Grant will work on this
- Presentation from Structured Data Capture (SDC)
 - Vijay Shag -
 - SDC
 - FHIR questionnaire vs questionnaire response
 - used an iso process
 - Joel - if you use iso 11179, does that imply a curated data repository?
 - yes?
 - Bob - Observation.related can point to QuestionnaireResponse. Can different Observations point to the same QuestionnaireResponse.
 - yes, to different parts of the same QuestionnaireResponse
 - Profile Roles: From Filler, Form Manager, Form Receiver
 - FHIR - first published in 2015
 - <http://hl7.org/fhir/current/sdc/sdc.html>
 - <http://hl7.org/fhir/current/sdcde/sdcde.html>
 - For STU3
 - <http://hl7.org/fhir/us/sdc/2016sep/sdc.html>
 - caDSR working with SDC, mostly working with IHE; they are interested in FHIR
 - Maturity phase for FHIR is still early
 - Amnon - concerned with certain type of using Questionnaire that might overlap with semantics of base resources, e.g., the following SDC profile for NCI Diagnosis (<https://www.hl7.org/fhir/sdc/questionnaireresponse-sdc-profile-example.html>) represents diagnosis data that might be misaligned with the FHIR base Condition

resource, having attributes like 'diagnosis type' that doesn't match any of the natural candidate attributes in the Condition resource, i.e., category and code attributes of the Condition resource

- Which happens first?

Tue Q2 11:00-12:30AM

Attendees:

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
3. Terry McDonnell - Syapse - terrym@syapse.com
4. Joel Schneider - NMDP - jschneid@nmdp.org
5. Michael Martin - AAVLD - mkm1879@gmail.com
6. Grant Wood - grant.wood@imail.org
7. Byoung-Kee Yi - Samsung Medical Center - byoungkeeyi@gmail.com
8. Elizabeth H. Newton - elizabeth.h.newton@kp.org
9. Rosalyn Ryan - Rosalyn_ryan@dell.com
10. Mera Choi - mera.choi@hhs.gov
11. Avinash Shanbhag -ONC - avinash.shanbhag@hhs.gov
12. David Kreda - david.kreda@gmail.com
13. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
14. Øyvind Aassve - Norwegian Directorate of e-health, oyvindaa@yahoo.com
- 15.

Co-chair - Gil

- Domain Analysis Model (DAM) update
- Gil sent out copy of the doc
 - <http://tinyurl.com/hux37lt>
- Use cases for Clinical Genomics
 - Getting to "publication status" of the document, inclusive of its newest (Sep 2016) use cases - time boxing, etc., to get to "productize" it inside the HL7 CGWG
 - Grant W - Like what Bob M did for the HLA use case, we should go through the DAM use cases and illustrate what FHIR resources and profiles could be used to support each step of the use case diagram.
 - Elizabeth will connect with a genetic counselor to get input on where they would be included in the workflow.

- suggestion - add items on how different stakeholders would fit into workflows, e.g., ask Larry Babb to point out where submitting or exporting data from ClinVar/ClinGen may occur.
-

Tue Q3 1:45-3:00PM

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Elizabeth Newton - elizabeth.h.newton@kp.org
3. Daniel Vreeman (Regenstrief) - dvreeman@regenstrief.org
4. Terry McDonnell - Syapse - terrym@syapse.com
5. Swapna Abhyankar - Regenstrief Institute, Inc. - sabhyank@regenstrief.org
6. Clem McDonald - NLM -
7. Perry Mar - Partners HealthCare System - pmar@partners.org
8. Joel Schneider - NMDP - jschneid@nmdp.org
9. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
10. Andrew Statler - Cerner - astatler@cerner.com
11. Shannon Lu - NLM - shannon.lu@nih.gov
12. Grant Wood - grant.wood@imail.org
13. Andrea Pitkus - Intelligent Medical Objects (IMO) -- apitkus@imo-online.com
14. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com

Co-chair Bob

- V2 lite – Clem leading discussion
- Ballot Reconciliation Started
- Spreadsheets have yet to be consolidated
- Started with comments by those present for in-person requirements
 - Andrea Pitkus - went through 1st six comments
- for going forward, consolidated spreadsheet will be managed by Clem's group, will shared results with CG group at calls. Block votes when we can. Try to streamline this process.

Tue Q4 3:30-5:00PM

Attendees

1. Bob Milius- NMDP - bmilius@nmdp.org
2. Swapna Abhyankar - Regenstrief Institute, Inc. - sabhyank@regenstrief.org
3. Clem McDonald - NLM -
4. Shannon Lu - NLM - shannon.lu@nih.gov
5. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com

Co-chair Bob

- Continued V2 Lite ballot reconciliation
-
- (concurrent with Joint Meeting hosted by Patient Care - negation modeling)

Wed Q1 9:00-10:30AM

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Joel Schneider - NMDP - jschneid@nmdp.org
3. Terry McDonnell - Syapse - terrym@syapse.com
4. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
5. Grant Wood - grant.wood@imail.org
6. Elizabeth H. Newton - elizabeth.h.newton@kp.org
7. Perry Mar - Partners HealthCare System - pmar@partners.org
8. Andrea Pitkus - Intelligent Medical Objects (IMO) -- apitkus@imo-online.com
9. Scott Robertson - Kaiser Permanente
10. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
- 11.

Co-chair Bob

- Family History
 - slides presented by Grant
 - products: V3 pedigree, implementation guide
 - project for Reaffirm HL7 Version 3 Standards
- Prep for Clinical connectathon
 - Grant went over clinfhir.com tool
- GA4GH activities
 - grants works with clinical workgroup
 - 5 different of work products
 - 3 are catalogs
 - Catalog of Global Activities - International Genomic Data Initiatives
 - Gil help create this
 - Catalog of Global Activities - Family History
- GA4GH coordination – Grant
 - 10 activities lists -
 - HL7 CG
 - GA4GH
 - G2MC
 - DIGITize
 - PMI

- eMerge
- IGNITE
- Clingen, VMC
- Genetic Alliance
- Other international groups
- what are the deliverables for each group
- what are the timelines
- Coordination of Goals

Wed Q2 11:00-12:30AM

- Attendees
- 1. Perry Mar - Partners HealthCare System - pmar@partners.org
- 2. Elizabeth Newton -- elizabeth.h.newton@kp.org
- 3. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
- continued discussion
- (concurrent with Joint meeting hosted by OO)

Wed Q3 1:45-3:00PM

Attendees

1. Joel Schneider - NMDP - jschneid@nmdp.org
2. Elizabeth Newton -- elizabeth.h.newton@kp.org
3. Perry Mar - Partners HealthCare System - pmar@partners.org
4. Michael Martin - mkm1879@gmail.com
5. Bob Milius - NMDP - bmilius@nmdp.org
6. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
7. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
8. James Chen - ASCO - james.chen@osumc.edu
9. Terry McDonnell - Syapse - terrym@syapse.com

Co-chair – Amnon

- Bob F will help lead discussion
- Information Modeling – overview of work;
 - Bob F presenting slides
 - background, role of information modeling in CG WG
 - IM sub-group - purpose
 - development of an information model to represent the cg domain
 - ?
 - CG IM
 - technology agnostiic

- conceptual model
- promote harmonization and inter-artifact consistency among all CG WG standards
- incorporate relevant concepts requested by other groups working in the CG Domain
 - GA4GH
 - ClinGen
 - NCBI
- IM subgroup meeting Thursdays at 10AM ET
 - no meetings next week, restart
 - Action item - ask group if another time is preferred for IM subgroup
 - hard to balance west coast, east coast, and europe/asia
 - google doc for minutes
- Reference resources: internal
 - clinical sequencing DAM (previous ballot, WIP)
 - CDA GTR DSTU
 - Family History
 - CD Statement DIM
 - V2/LOINC specification
 - "V2 Lite" (WIP)
 - FHIR genomics (WIP)
- Reference resources: external
 - Clingen
 - allele model
 - assertion model (WIP)
 - GA4GH
 - schema
 - ONC
 - Personalized Healthcare Draft Use Case (2008)
 - VMC (Variant Model Collaboration)
 - Terminology
 - Draft variant model (WIP)
 - BRIDG
 - National Academies (previously known as IOM)
 - DIGITize
- IM group approach
 - initial focus - exchanging structured genetic lab test results
 - non-human readable lab report
 - start in the middle, work out
 - up: report
 - down: sequence, genotype, etc
 - Modeling approach
 - review existing resources

- define concepts (terminology)
 - discuss concept relationships
 - draw boxes and lines
 - repeat
- Working draft
 - UML diagram
 - key principle: separate observations from interpretations
- Immediate Next Steps
 - Drill down
 - observation
 - result
 - finding
 - Roll up
 - report/aggregated report
 - Expand
 - assay/method
 - device
 - provenance
- Future Directions
 - Continue modeling
 - alignment, consensus is hard
 - harmonization with VMC
 - Deliverables
 - Conceptual model(s) with definition
 - instance examples from use case
 - mappings and recommendations to WIP artifacts
 - Mappings to external models
 - GOAL: promote harmonization and inter-artifact consistency among all CG WG specifications
 -
- VMC – variant model collaborative review
 - participants
 - ga4gh
 - hl7
 - clingen
 - ncbi
 - so
 - ebi
 - Ensembl
 - Goal: to develop a technical specification for representing and communicating biological sequence variation
 - docs online
 - terminology

- https://docs.google.com/document/d/1z-Fz-x4yEfp5d-k-N_OkLzvzVCMhoZgobVHLJxwkaKg/edit#heading=h.4ie4bk77jsuu
- model
 - genmymodel link?

Wed Q4 3:30-5:00PM

Attendees

- Bob Milius - NMDP - bmilius@nmdp.org
- Perry Mar - Partners HealthCare System - pmar@partners.org
- Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
- Bob Freimuth - Mayo
- Elizabeth Newton - elizabeth.h.newton@kp.org
-

Co-chair – Bob

- Deadlines
 - Rooms for next WGM - Friday Sep 30, 2016 ET
 - repeat same schedule for Jan 2017
 - Must submit PSS to PMO and Steering Division - October 9, 2016
 - no new PSS
 - Submit Notice of Intent to Ballot - November 6, 2016
 - V2 Classic?
 - FHIR
 - Proposed timeline to publish by year-end
 - Final content deadline: Dec. 4
 - FHIR Core Substantive change deadline: Nov. 11
 - Change proposal cut-off: Oct 9
 - Preliminary triage complete: Oct. 1
 - one more week?
 - Are you ok with this timeline? Thur Q3
 - maybe, but it will be tight and may not be of quality
 - won't be able to make Oct 9 deadline for change proposal cut-off
 - V2 Lite
 - Nov 20
- WG docs
 - Mission & Charter
 - DMP
 - SWOT
- WGM Review
- Leftover topics
- WG business and planning