

2020-05 CG WGM - Virtual

Three meetings were held virtually that took the place of the CG events at the WGM.

- Normal FHIR subgroup
- Normal Tuesday workgroup call
- Joint call with O&O (Thursday)

Notes from FHIR subgroup meeting (2020-05-18)

FHIR Subgroup Meeting MAY 18th, 2020

We are ZOOMing away from FCC. New coordinates:

<https://zoom.us/j/2980068716>

Find your local number: <https://zoom.us/u/adNIRW2P8J>

Quick link to this sign-in/notes document: tinyurl.com/fhirgenomics

Sign In: (presiding co-chair - Jamie Jones)

1. Kevin Power - Cerner - kpower@cerner.com
2. May Terry - MITRE - mayT@mitre.org
3. Liz Amos - NLM - liz.amos@nih.gov
4. Bret Heale - Intermountain - bheale@gmail.com
5. Bob Dolin - Elimu Informatics - bdolin@elimu.io
6. Daniel Rutz - Epic - drutz@epic.com
7. Hayden Bader - Epic - hbader@epic.com
8. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
9. Patrick Werner - MOLIT - pw@molit.eu
10. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
11. Scott Isaac - Epic - scisaac@epic.com
12. Clem McDonald - NLM - clemmcdonald@mail.nih.gov

Agenda:

1. Connectathon review
2. Next steps
3. JIRA (didn't cover)

Discussion:

1. Connectathon review
 - a. Representing variants in apps (<https://molit.eu/variant-viewer/>) Still very beta, configurable rewrite is done at the moment.
 - i. Patrick is a gift
 - i. SPDI as a partial check on validity, hoping to integrate with mutalyzer
 - i. Webcomponents contained here: <https://github.com/molit-institute/fhir-components>
 - ii. Docs here: <https://docs.molit.eu/fhir-components/#/components/MolecularReport>
 - iii. Currently - Need to support ALL possible configurations/representations to display. Choosing only one method may not be sufficient
 - iv. Producing reusable open source web components
 - v. Clem's group working on GUI questionnaire with some bioinformatic logic built in
 - b. \$find-subject-variants
 - i. SPDI effective tool for indels that look like this
 - i. Syntactic normalization - use API to display variants using the same components regardless of how it was provided to server
 - ii. Semantic normalization - aligning different representations of the 'same variant' e.g CCC > CC vs CC > C.
 - c. STU2 implication reporting
 - i. <http://hapi.fhir.org/baseR4/DiagnosticReport/1167557>
 - a. [CAT 24 Report out](#)
 - b. [CG connectathon 24 links](#)
2. [http://hapi.fhir.org/baseR4/Observation/?component-code-value-concept=http://loinc.org|81252-9\\$https://www.ncbi.nlm.nih.gov/clinvar|653974&_revinclud=Observation:derived-from](http://hapi.fhir.org/baseR4/Observation/?component-code-value-concept=http://loinc.org|81252-9$https://www.ncbi.nlm.nih.gov/clinvar|653974&_revinclud=Observation:derived-from)
 - a. Questions over integration with problem list
 - b. Condition.evidence?

2. This example can trigger some interesting discussion around our usage of 'Associated Phenotype' Ties back to this JIRA: <https://jira.hl7.org/browse/FHIR-26945> And the Zulip discussion here: <https://chat.fhir.org/#narrow/stream/179197-genomics/topic/Stress.20Testing.20IG.20Implications.20with.20Examples>
- i. FSH - FHIR shorthand for creating structureDefinitions
 1. <https://github.com/FHIR/sushi/releases/tag/v0.13.0-beta.1>
 1. Current Beta version of sushi pulls all IG configurations into one yaml file, does more than just create structure definitions, can initialize a whole IG with template - open questions on long term support and scope in that regard (question of whether it should just restrict itself to structureDefinitions)
 2. Technical concerns around lack of round tripping from structure definitions. Proof of concept work in progress
 - ii. QA - [outdated artifacts pull request for R5](#)
 1. Review current core build for straggling references to old profiles
 2. Guidance on MolecularSequence remains for now (no other guidance on it exists)
 3. Merits full review

See the following notes for other calls that happened during the Virtual WGM for CG

[CG-2020-05-19](#)

[2020-05-21 OO/CG/II](#)