

2019-05 CGW WGM Minutes

- Date: Mon, May 6, 2019; Quarter: 3
- Date: Mon, May 6, 2019; Quarter: 4
- Date: Tue, May 7, 2019; Quarter: 1
- Date: Tue, May 7, 2019; Quarter: 2
- Date: Tue, May 7, 2019; Quarter: 3
- Date: Tue, May 7, 2019; Quarter: 4
- Date: Wed, May 8, 2019; Quarter: 1
- Date: Wed, May 8, 2019; Quarter: 2
- Date: Wed, May 8, 2019; Quarter: 3
- Date: Wed, May 8, 2019; Quarter: 4


Date: Mon, May 6, 2019; Quarter: 3

Attendance: [2019-05 WGM Attendance](#) (Jira account needed)

Goals

To provide an update on the current state of FHIR Genomics projects within the CG WG including the recent Connectathon 21, to prepare for our joint session Monday Q4 with FHIR-Infrastructure, and to open the floor for other FHIR-specific items of importance to cover during the rest of the WGM and before Connectathon 22 in Atlanta, GA (September 14-15).

Discussion items

Time	Item	Notes
15min	Introductions	
35min	FHIR Genomics Snapshot presentation	https://tinyurl.com/cgwgmslides 
5min	Recap /feedback from Connectathon 21 participants	

15min	Targeted Discussion to identify items to bring to FHIR-I	<p>https://docs.google.com/document/d/1ngRdfh9HRwX-YO2nl9HQ4rQTQhTwrjraRCJ9sJjis0/edit</p> <p>Questions for (potentially) FHIR-I/Josh</p> <ol style="list-style-type: none"> 1. FHIR Release 5 timelines <ol style="list-style-type: none"> a. "On-going community discussion" (mid-late 2020?) 2. IG creation templates <ol style="list-style-type: none"> a. Exploring different views a. http://www.fhir.org/guides/registry/ a. Good question for Rick <ol style="list-style-type: none"> i. It "works" and there is no a. Packages and tooling updates coming b. Often focused on models c. Potential overlap with new guides within HL7 (monitored) and external (active engagement) d. Trifolia, is it here*?? (official support??) most recent word is aimed for Autumn 3. IG Publication timeline (aiming for Summer) 4. MolecularSequence maturity <ol style="list-style-type: none"> a. What is needed to get to FMM2 b. The artifact has been tested and successfully supports interoperability among at least three independently developed systems leveraging most of the scope (e.g. at least 80% of the core data elements) using semi-realistic data and scenarios based on at least one of the declared scopes of the artifact (e.g. at a connectathon). These interoperability results must have been reported to and accepted by the FMG 5. Improving Visibility of Genomics work <ol style="list-style-type: none"> a. GA4GH, eMERGE, mCODE, ... a. How to approach illumina/vendors/etc, <ol style="list-style-type: none"> i. Want to get these updated to FHIR ii. What pain are they feeling with their current system? iii. Focus on tools/unmet needs <ol style="list-style-type: none"> i. Sequencing software/LIM systems creating v2 messages a. Liaisons and alignment with other genomic groups/projects b. Published artifacts, educational instructions/open QA's etc 6. Servicerequest codes vs observation components/etc <ol style="list-style-type: none"> a. But serviceRequest doesn't have components <ol style="list-style-type: none"> i. Use orderDrysil a. Likely can resolve with O&O joint session b. Observation can be basedOn a serviceRequest, so codes should align <ol style="list-style-type: none"> 1. Must Support definitions..... <ol style="list-style-type: none"> a. Considering opening up from boolean to a dict a. Leave must-supports (and potentially enforced 1.. Cardinalities to use cases) <ol style="list-style-type: none"> a. http://www.healthintersections.com.au/?p=2908 b. Full reanalysis of "minimal implementations" needed? 2. Specimen cardinality conversation (normative cardinality is 0..1) <ol style="list-style-type: none"> a. Do we cross out Specimen and only use the extension? b. How to specify which specimen is the 'main' one and which go in the extension? Need textual guidance a. It is okay to increase the higher cap on cardinality from 1 to * IF you don't require anyone to care about items beyond the first b. Opposite can't happen <ol style="list-style-type: none"> a. Tumor normal variant analyses (need more than 1) (tumor is main) b. Trio analysis (3 specimens or 3 observations?) (patient is main, parents' extra) c. O&O: you'll have to use an extension d. FHIR's version policy on "breaking changes" here is interesting... (versions.html) 3. Sending "coded codes" <ol style="list-style-type: none"> a. Obs-variant in particular [discrete complex structural] b. Good to move forward with! 4. Open floor <ol style="list-style-type: none"> a. Remote procedure calls <ol style="list-style-type: none"> i. Expand a value set, export something, perform a calculation/validation, etc a. Search Operations for Genomics
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Action items



Date: Mon, May 6, 2019; Quarter: 4

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Goals

To use joint time with FHIR-I to ask FHIR-specific questions, forward new and interesting issues, and develop strategies for the future.


Discussion items

Time	Item	Notes
	See notes from Q3 (see above)	

Date: Tue, May 7, 2019; Quarter: 1

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Discussion items

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	Review Agenda	
	Review of CG for newcomers	 HL7_CG_activities_May2019.pptx
	code systems Link to wop spreadsheet: https://tinyurl.com/CG-CodeSystems	<ul style="list-style-type: none">• have list of external codesystems; need permission from them to use• ddbj, embi, too high level• genbank has unique accession #s• conversation about "what is a code system"; grammar/syntax vs codes• we need to identify use cases in our IG for these code systems (e.g., where would UnitProtKB be used?)• Do we need these completed before we publish the IG?<ul style="list-style-type: none">• not necessarily
	IG ballot reconciliation	<ul style="list-style-type: none">• 19 open ballot-related trackers left• GF#16082<ul style="list-style-type: none">• motion - consider for future use - Bob F/Patrick• abstain ; 1• nay: 0• yea:7• G#

Action items

- [Bob Miliusto](#) to contact [genenames.org](#) and [imgt/hla](#) for permission to use codesystem
- [Patrick Werner](#) [Liz Amos](#) find locations in IG where specific code systems can be used
- [Patrick Werner](#) to prepare template email to contact external orgs to verify URIs

Date: Tue, May 7, 2019; Quarter: 2

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Discussion items

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	IG ballot reconciliation	<ul style="list-style-type: none">• #16247<ul style="list-style-type: none">• already voted on, status is wrong• #16860 - clem<ul style="list-style-type: none">• withdrawn by Clem• #16863 - clem<ul style="list-style-type: none">• withdrawn• #16871 - clem<ul style="list-style-type: none">• not persuasive already voted on, status needs updating• #16876 - clem<ul style="list-style-type: none">• already voted last fall to be persuasive with mod, still need to apply• #19833 - bob dolin<ul style="list-style-type: none">• change observation.code to LOINC 69548-6 (Genetic variant assessment)• motion bob m/clem• abstain/nay/yea: 0/0/7• #19838 - bob dolin<ul style="list-style-type: none">• use LOINC 81293-3 (DNA region of interest panel)• motion bob m/clem• abstain/nay/yea: 0/0/6• motion passes

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Discussion items

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	IG ballot reconciliation	<ul style="list-style-type: none">• #19839 - bob dolin<ul style="list-style-type: none">• question about value for region studied. Decision for #19838 is that code will be 81293-3 (DNA region of interest panel), so what is the value for this? no simple value exists• remove "valueX" ?• motion (Jamie J/Clem): remove value(card:0..0), i.e, all information is intended to be contained in components<ul style="list-style-type: none">• abstain/nay/yea :<ul style="list-style-type: none">• 1/0/7• #19840 - bob dolin<ul style="list-style-type: none">• motion JameJ/• take text of extension as guidance into our IG, get rid of extension. Use Observation.derivedFrom.• needs a diagram• derivedFrom has to be profiled similar to the extension.• abstain/nay/yea : 0/0/8• #19841 - bob dolin<ul style="list-style-type: none">• cont to next Q

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Time	Item	Notes
	IG ballot reconciliation	<ul style="list-style-type: none">• #19841 - bob dolin<ul style="list-style-type: none">• Motion Jamie J/Alex• Persuasive• Use 82120-7 (currently Allelic Phase), LOINC code should be renamed to Sequence Phase• abstain/nay/yea : 1/0/7• #19842 - bob dolin<ul style="list-style-type: none">• proposed: create internal (HL7) list (CodeSystem) containing cis/trans/indeterminate/unknown• #19845 - bob dolin<ul style="list-style-type: none">• motion: jaimie j/bob m• add three new components to region studied profile• persuasive• abstain/nay/yea : 0/0/7• #19884 - bob dolin<ul style="list-style-type: none">• mark as duplicate of #19876• #19947 - Joel Schneider<ul style="list-style-type: none">• motion: bob m/ jaimie j• persuasive with mod• abstain/nay/yea : 0/0/7• #19995 - Casey Thompson<ul style="list-style-type: none">• proposed disposition to larger group: persuasive• add/improve textual guidance and potentially correct the LOINC definition.• #19998 - Arvind<ul style="list-style-type: none">• motion: Bob M/May T• persuasive• abstain/nay/yea: 0/0/7

Date: Wed, May 8, 2019; Quarter: 1

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Goals

Discussion items

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	Information Model	Bob F presented modeling approach, and GA4GH https://drive.google.com/file/d/1yR_fChHssVpf0dFOfauh7P1jsybr-hme/view?usp=sharing https://drive.google.com/file/d/1uG_hQp5bF4nHpBDzBKxjUUjv6KkXDNPg/view?usp=sharing

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	Joint with OO	discussions with CG <ul style="list-style-type: none">• CG IG ballot status,• IM work/GA4GH• Q: observation.specimen need cardinality of more than 1<ul style="list-style-type: none">• A: create extension, e.g. additionalSpecimens. May be a core extension for R5• Q: workflow serviceRequest to observation. How to use codes? serviceRequest doesn't have components<ul style="list-style-type: none">• A: the codes don't have to be the same. Can use a local code for ServiceRequest.code, and use our IG code /component.code in the observation.• Q: need to have sections in DG<ul style="list-style-type: none">• A: ongoing discussion in OO with other work groups

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	May WGM review	<ul style="list-style-type: none">• jaime j : last time we recommended dispositions, but not actually vote• alex: prioritize tracker items that people are actually here in person• jamie j: of the rest of the non-ballot gforge issues, prioritize those that should be addressed before publication• patrick: liked that we made decisions about ballot items• jaime j: agrees


	CG business	<ul style="list-style-type: none"> • Co-chair election <ul style="list-style-type: none"> • Patrick Werner current interim • new rules around co-chair election <ul style="list-style-type: none"> • aligned with other elections • Nominations shall be accepted from May 1 through June 15 of each year. • Elections shall occur from July 1 through July 30 of each year. • Runoff elections, if necessary, shall occur from August 7 through August 21 of each year. • election results announced in September WGM • Starts in 2020 • e.g., Co-chair terms for Bob Milius and Gil Alterovitz end in 2020 (can be re-elected) <ul style="list-style-type: none"> • election will for both slots will be held in July 2020 • their terms will be extended to end of 2020 • new co-chair terms start in Jan 2021 • Work Group Health • CG Reports <ul style="list-style-type: none"> • http://www.hl7.org/Special/committees/clingenomics/reports.cfm • Mission & Charter <ul style="list-style-type: none"> • WorkGroup Home • updated in 2018 • SWOT <ul style="list-style-type: none"> • https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit • Review complete as of Aug 1, 2017 • Approved in Sep 2017 WGM in San Diego • updated in 2018 • Decision Making Process <ul style="list-style-type: none"> • https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srIde0EBs0FI/edit • Review complete as of Aug 15, 2017 • Approved in Sep 2017 WGM in San Diego • Joel: question about scribe, we should have this in advance of meetings • Weekly calls <ul style="list-style-type: none"> • Main <ul style="list-style-type: none"> • Tue, 11am ET • FHIR <ul style="list-style-type: none"> • Mon, 11am ET • Information Modeling <ul style="list-style-type: none"> • Thu, 10am ET • ask Bob F to consider later in day (will may increase participation for west coasters) • Traditionally, we don't have calls the week following the WGM
	September WGM planning	<ul style="list-style-type: none"> • Room request • Same schedule? • Potential discussion topics <ul style="list-style-type: none"> • roadmap for next version of IG(R5) • adding mention of provenance and workflow to IG • Comparing DAM to IG • IG change log

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Discussion items

Time	Item	Notes
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	mCODE presentation (May Terry)	 <p>20190508_mCODE...CGWGPreso.pptx</p>
	FHIR & IG summary	<p>Clem: Allelic Frequency UCUM unit, is it % or a fraction? Patrick: I've only seen fractions Alex: DBsnp uses fraction</p> <p><input type="checkbox"/> Patrick Werner add Tracker Item to remove fixed value "%" from Allelic Frequency unit.</p> <p>Discussion about proposed example in new LOINC Term "Coordinate System", verified and corrected example.</p>
	IG differential	<p>Jamie: How to do changelogs? Is there a best practice? Lloyd: manually curated changelog is the way to go. Automatic</p> <p>Clem: How do we improve the visibility of the IG? Lloyd: Putting "FHIR Genomics Reporting Implementation Guid" as a name on the IG would be a good idea.</p> <p>Patrick: How to transport narrative methodology information? Lloyd: relatedArtifact would be a solution.</p>

Action items

- Upload mCode slides