CG-2020-09 WGM Minutes

Date: Monday 9/21/2020

Quarter: 12:00-1:30 ET (Monday Q2)

Minutes Approved as Presented

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This is to approve minutes via general consent. *"You have received the minutes. Are there any corrections to the minutes? (pause) Hearing none, if there are no objections, the minutes are approved as printed."*

Goals

Generate plan for addressing issues outlined in https://jira.hl7.org/browse/FHIR-28403 - Provide clear guidance on how to use the IG - Within Content Pages, Detailed Description Pages, and by adding an Examples Page

Discussion items

Time	Item	Who	Notes
15	IT checks and introductions		May be moving some session topics
15	Introduce tracker for discussion Review state of current material		https://jira.hl7.org/browse/FHIR-28403?filter=12310 Jamie: We already have some good guidance in the IG. E.g. the HLA Part, but we are missing the same quality of guidance in other parts of the IG. Probably most important: the Variant page to give guidance on the usage. Another task is to flag examples contained in bundles to show up on the example page. ☐ create a Jira Ticket for flagging examples in example bundles. May: we also have to decide on if we want to keep all examples. Kevin: The tracker needs to be more precise on the desired action. We need more specific lab test examples, how to order a panel or specific genes. Arthur: Agrees. Liz: likes to take a look at the V2 examples and guidance to get an idea what is part of a diagnostic report and map it to the FHIR CG DR. https://www.hl7.org/documentcenter/public/standards/dstu/V251_IG_LRI_R1_STU3_2018JUN.pdf Patrick: could improve the understand-ability of the example bundles by adding a spreadsheet containing the "raw information" with column headings May: depending on the target audience mind-maps also improving the understand-ability. Kevin: should ask emerge if we can reuse their examples as these are well made: https://emerge-fhir-spec.readthedocs.io/en/latest/design.html#rept-examples Arthur: brilliant idea, have to pay attention if it still maps Kevin: could include it as a real life example for the application of our IG. Will follow-up in the Jira Ticket comments. May: Another issue: the java validator doesn't check the contained resources Patrick: in the hapi validator the validation of target references /entries can be enabled. ☐ Patrick Werner check the validation of bundles.

55	discussion		**Looking at the IG example: http://build.fhir.org/ig/HL7/genomics-reporting/Bundle-diagnosticreport-cgexample.json.html** Resources: https://docs.google.com/spreadsheets/d/1zgT0GYQZHjUYD7N0mYNzJozqdPieW7G2yglh3Ayl6gU sheet listing IG profile components https://docs.google.com/spreadsheets/d/1-Co8NmSh75y_1wqJysnLBcgT7-lL4JY3WRIcNPr5qr8/edit#gid=666724439 sheet for crafting examples https://drive.google.com/drive/folders/18T4RS0VnrJdLS3k79skbrZ0cYyL1U53t folder for adding example reports Discussion: Liz: The usage of grouper is still unclear Kevin: Grouper is meant to group related Observations on a lower level than the Diagnostic Report current Grouper guidance contained here: http://build.fhir.org/ig/HL7/genomics-reporting/general.html Arthur: need to make the grouper example more visible Bob F: when does a grouper has a semantic meaning, in which case is it just used for structural purposes. Liz: The example should be flagged as a pharmacogenomics example. walkthrough of older V2 example: https://docs.google.com/document/d/1UsRfk47FqGdHDzBAdXs4jit2vy-vuxfHmCtFBGifW58/edit#heading=h.ljsjod3gdqpy Next steps: 1. review/update current examples a. add summary descriptions to each example 2. asking eMerge about using/modifying their example graphics 3. discuss further needed actions in the Jira tracker/upcoming calls 4. identify and curate an easiest example of genomics reporting 5. Liz: figure out what NCBI wants

Action items

- [ ]

Date: Monday 9/21/2020

Quarter: 14:00-15:30 ET (Monday Q3)

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1. S4G (first)
2. Family History (Grant)
3. FHIR-I prep
4. examples (cont)

ONC Sync for Genes Update (Bob F)

Slides

Phase 4 soliciting groups to participate, reach out to Bob F or Tracy.Okubo@hhs.gov

Discussion Points to consider:

- Core Vs custom IGs (80/20)
  - eMERGE, HLA, mCODE, phenopackets
- Collaboration and engagement
  - onc/nlm/nhgri
- CG WG effort
  - standard development (new)
  - fixes, clarifications, examples (existing)
  - consultation to support adoption
    - deriving from parent profiles (including US core)
      - currently lacking functionality in FHIR validator

Family History (Grant W)

- GA4GH Clin/Pheno Pedigree subgroup
  - minimum data set for a family health history - work started in 2008 - will result in a live document for comment
    - standard-agnostic list of recommended elements
    - PED format found insufficient by itself
      - each PED is scoped to one particular disease
    - working with app vendor called 'cancer IQ'
  - current FHIR resource is targeted at (vague) family health history statements taken by clinicians - not computable
    - genomics profile on that resource (from R3 work) is still lacking for robust use.
      - ids from PED (family, proband, father, mother, individual)
  - Ancestry (from an ontology if possible-hard to identify one) (HANCESTRO one option, but many ontologies tend to be too narrowly scoped)
  - Considering a place for marking records of pedigree analysis in the EHR

Ancestry as Observation vs other resource? Research-enabling focus rather than solely clinical

- need to identify the ontology (could fit well as an Obs but has terminology concern, can't recommend any one ontology at this time)
- One approach has been USCDI extensions to Patient:
  - https://build.fhir.org/ig/HL7/US-Core-R4/StructureDefinition-us-core-patient.html

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Date: Monday 9/21/2020

Quarter: 16:00-17:30 ET (Monday Q4)

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CG Discussion points:	• MolecularSequence - alignment with IG / clean up • IG - STU2 ballot in Jan 2021 • MolSeq + IG alignment with emerging IM model • Guidance for WGs that were maintaining IGs via spreadsheets? • R4B request - cleanup of 'genomics guidance' page? • email FMG contact for removing particular examples and/or adding in more clear guidance of deprecation • Can include PR: https://github.com/HL7/fhir/pull/877 • If we can get this approved, we should likely make additional changes to the 'genomics guidance' page, making references to our IG much more clear, and de-emphasize MolSeq. • validator "compare profiles" - official plans to support this? yes, only "temporarily broken" • towards validating against (and deriving from) multiple profiles (e.g. genomics reporting and US Core) • invariant to require extra validation -wouldn't be great for slicing etc • New Resources - Is this the process? • Resource Proposals - correct link, but may have to wipe off some dust from the move to confluence • JSON5 comment support? • not part of R4 so may be interesting for R5 - hacked some things into the publisher as _comments:[]
FHIR-I Points	• Improve FHIR build tooling (separate branch to main build) • new releases: • R4B release in scope, schedule dependent on ONC funding request • moving off of spreadsheets for porting between versions • some changes, especially for items not likely to have been implemented already • evidence-based medicine content inclusion • R5 ballot - a "significant release" including multiple normative resources • ballot may 2021 cycle, aiming to publish spring 2022 • FMM levels for resources • 'Must Support' in IGs • need to refine for polymorphic elements-how support means per datatype • potential updates to supported JSON versioning re: data choice types -unlikely but may be reviewed together to comments

Date: Tuesday 9/22/2020

Quarter: 10:00-11:30 ET (Tuesday Q1)

Joint with O&O (hosted by O&O, O&O minutes: https://confluence.hl7.org/pages/viewpage.action?pageId=91991152#id-2020092125WGM-TuesdaySeptember22,2020)

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CG Discussion points:	• **Code binding of Request and DiagnosticReport:** • doesn't have to be the same, sometimes it is. They should be aligned. • the order is normally linked via ID in the DiagnosticReport • DiagnosticReport should have a code, or a binding to a ValueSet to find all reports if this kind. Category could be fixed for all of them. • **Further discussion/plans on the use of "notes" or something else for extra information on genetic findings that we talked about a few months ago.** • eMerge ended up creating Extensions on DR and Obs for capturing additional text parts which are always the same, or differs depending on the chosen tests (e.g. disclaimer texts) • DiagnosticReport vs Composition: these Notes and Texts could be captured in composition sections also including the structured data. • Alternative: put the text into Observation.note, potentially add a label Extension to the Annotation to express the kind of note. O&O is thinking about adding a type to Annotation. • **Structured Reports is an ongoing discussion in O&O, discussed on Tuesday calls.** ☐ contact O&O for a joint Tuesday meeting (2-3 ET) • **Biomarkers in CG** • How to include old Biomarker Observations into the DR? • These are Observations which could be linked in the DR • Do we need to express: this is an important Observation which is important for the remaining life of the patient? • Could be transported as Conditions • mCode already has a Biomarker profile, the question is how to get this profile "universal", who will be the care taker? • Clem: CG should do it and work together with other WGs, e.g. cancer? • the current mCode profile is derived from US-Core, which isn't a technical problem, but has political implications. An international/uv Biomarkerprofile is needed. • Work can be done by CG/ is already mostly done by mCode. • **Media is now (R5) merged into DocRef** • can be linked through: Obs.derivedFrom and/or valueAttachment (in R5) • example: 20 Variant observations referencing the VCF file via derivedFrom, the DR also could link to the VCF via "media" ☐ create tracker/ discussion: rename media backboneElement "media" to "attachment" • update on OO/PA work on **a new "task"-like resource to handle the movement of specimens and other procedural things**

Date: Tuesday 9/22/2020

Quarter: 12:00-13:45 ET (Tues Q2)

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Agenda:

Liz / Rachel

FHIR-27748 - Rename "Functional Annotation" Component to "Variant Consequence" TRIAGED

FHIR-27747 - New Component for Functional Effect TRIAGED
Kevin (discuss what the 'genomics guidance' page should/should not contain)
FHIR-27159 - Update Genomics Guidance page to more clearly point to our IG TRIAGED

FHIR-16402 - Add material from guidance document TRIAGED

- Ready to Vote: https://jira.hl7.org/issues/?filter=13081
  - actually not quite ready to vote
  - need to review comments
- Not ready to vote - still needs discussion
  - Pending with a resolution: https://jira.hl7.org/issues/?filter=13016
  - Pending with no resolution: https://jira.hl7.org/issues/?filter=12414

FHIR-27159 - Update Genomics Guidance page to more clearly point to our IG TRIAGED

https://www.hl7.org/fhir/genomics.html

Discussion:

- Bob D: The page should be updated to "don't look at anything here, go to the IG", question is if this page contains information we want to keep (different tracker discussed later)
- Kevin: could update the page to a landing page telling people to go to the IG
- FHIR-I was asked yesterday in the FHIR-I joint meeting about including our applied PR to R5 into the R4B release. Couldn't get it into the FHIR 4.0.1 technical correction release, as this isn't considered to be a technical correction but a content change.
- Kevin: could try to get a updated page into R4B
- Bob D: remove everything, keep a link to the ig. Could include the content in a part of the IG.
- Arthur: the page needs a strong statement that implementers should follow the IG
- Kevin: 3 Options:
  1. move the content into our IG
  2. move the content to Molecular Sequence
  3. create another page in core and move it there
- Liz: in favour of a clean landing page, move the content into the IG

Resolved

FHIR-27748 - Rename "Functional Annotation" Component to "Variant Consequence" TRIAGED

Liz & Rachel presenting the proposal:

- [ ] Liz Amos Rachel Kutner Slides attached

- Proposal to create new LOINC code for molecular consequence
- extensible value binding to the **structural_variant** SO branch (SO:0001537)
- functional annotation will be renamed to molecular consequence, binding will stay the same
- amino-acid-change-type is also potentially overlapping with the molecular consequence concept, and therefore potentially also be merged
- Discussion: should amino-acid-change type merge als be included into the tracker disposition?
  - breaking change which should discussed in another issue

- [ ] create Tracker item: merging amino-acid-change-type and molecular consequence

Resolved

FHIR-27747 - New Component for Functional Effect TRIAGED

- Clinvar has a field "functional consequence" similar to the functional effect concept, but has little standardization terminology.
  - https://www.ncbi.nlm.nih.gov/variation/docs/glossary/
  - https://www.ncbi.nlm.nih.gov/clinvar/docs/variation_report/
- A functional effect is an implication, so implication could be the correct location for this information?
- Discussion: still unclear whether this is is a component, or an Implication. Doesn't fit in the current Implication profiles well.

Date: Tuesday 9/22/2020

Quarter : 14:00-15:30 ET (Tuesday Q3)

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1. mCode (May T)	**mcode stu1 current status** **alignment with genomics reporting stu2** **oncotype DX (breast) use case discussion**
	mCODE STU2 Update and Tooling Discussion slides: **mCODE_Update_HL7WGM_CGWG.pptx** When genomics reporting IG makes a change on one of the structures pulled into mCODE, some concern from mCODE's business needs to also adopt the change. ~15 orgs have implemented mCODE MD Anderson, UT Southwestern, UCSF, Kaiser P, Intermountain, Mayo, Rush, Trinity, Princess Margaret Cancer Centre, Mass General, Dana-Farber, Brigham Health, Memorial Sloan Kett, U Penn, Geisinger, Washington U St. Lou Greater alignment on terminologies on the horizon May looking to submit LOINC code for oncotype dx
Tooling 1. IG creation /publishing	currently on manual editing of structure definitions Forge - windows only, needs licenses from HL7 Trifolia - web-based, limited github interaction FSH/SUSHI + GoFSH (decompiler) https://github.com/FHIR/GoFSH may be able to create equivalent structuredefinitions can use current sdefs in one folder, mark one to be generated by GoFSH, and everything should still connect

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Date: Wednesday 9/23/2020

Quarter: 10:00-11:30 ET (Wed Q1)

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• LOINC / "TBD code" reconcilliation • FHIR-16402 - Add material from guidance document TRIAGED FHIR-27864 - Update binding to LOINC Diagnostic codes value set TRIAGED • Ready to Vote: https://jira.hl7.org/issues/?filter=13081 • actually not quite ready to vote • need to review comments • Not ready to vote - still needs discussion • Pending with a resolution: https://jira.hl7.org/issues/?filter=13016 • Pending with no resolution: https://jira.hl7.org/issues/?filter=12414	FHIR-16402 - Add material from guidance document **Resolved** - along lines of FHIR-27159 from yesterday. FHIR-27864 - Update binding to LOINC Diagnostic codes value set TRIAGED **Needs input** Have to confirm LOINC categories for filtering and create more rigid picture of what to bind here beyond the original overall LOINC binding. Jamie to update disposition on uncallable regions component

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Date: Wednesday 9/23/2020

Quarter: 12:00-13:30 ET (Wed Q2)

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Agenda	Notes
CG Information Modeling • overview of where we are • update on recent modeling • roadmap - priority • laundry list of what we can do • technical approach to implement into FHIR IG • GA4GH alignment	Slides 1. Why 2. Review Model (preview Definitional Variant) 3. Roadmap Feedback / Questions • Is Sequence related to an "order panel"? — No, this is a lower level of data • Add an additional sequence representation? — Intent was to focus/"ground" at the conceptual level (Sequence) and let the various representations grow / extend as needed? Patrick Werner • Sequence-Based Variant Representation: data types of referenceAllele and alternateAllele maybe should be Allele? — allele name ties back to the names in VCF, SPDI, etc ... but still aligns conceptually to our definition of Sequence in our model. Can discuss further if needed. Prioritizing Next Steps (suggestions): • "Implications" - struggling on the IG side, perhaps can help • How to align logical model to FHIR IG current state and apply Bob's Crystal Ball (initial guess as CG resources) (Definitional resources) • MolecularSequence (all types, representations, GenomeAssembly, annotations+features) • MolecularVariation (all types, representations, annotations) • Assertion<<SEPIO>> (implications and inferences related to sequences and variations)

Date: Wednesday 9/23/2020

Quarter: 14:00-15:30 ET (Wed Q2)

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Item	Notes
resolving TBD LOINCs	

- Improving value sets within the IG
  - http://hl7.org/fhir/uv/genomics-reporting/valueset-tbd-codes.xml.html
  - see draft spreadsheet: LOINC / "TBD code" reconcilliation https://docs.google.com/spreadsheets/d/1dubwSbfbJ4K1_Ie93VXynEPXyQgudwzG9Vji-Hcvbfg/edit?usp=sharing
- Patrick: TBD codes need LOINC concepts (to be created)
  - red-highlighted codes are no longer used in our IG
    - functional annotation will be replaced by molecular consequence
  - several new concepts still require definitions, draft defs added for molecular consequence, callable, and not-callable
  - variant inheritance: def contains a link to SO hierarchy, should the def contain a binding to a value set?
- Jamie: the spreadsheet needs to be developed fully before LOINC can create any codes for us
  - Liz: include a "note" in the submission that the request is on behalf of the HL7 CG WG
  - Liz: use LOINC's spreadsheet template https://loinc.org/download/basic-submission-template/
- Liz: mode of inheritance - current def in spreadsheet is not correct, Liz will fix
  - [ ] Liz Amos
- Patrick: will transpose content to LOINC's template
  - [ ] Patrick Werner
- Patrick: content review - are we missing anything?
- Liz: how does associated cancer differ from associated phenotype?
  - Jamie: previously only differed in the profiles it appeared on. Currently, it provides an opportunity to differentiate what cancer type the therapy is approved for (often different than patient's diagnosed type)
- May: are all of these intended to be codeable concepts?
  - Jamie: examples from Tempus reports, medications used in other cancer types/indications/variations, need a way to express those even when they don't match the patient's specific cancer or var profile (e.g., approved therapies for a different indication)
- Patrick: assoc pheno and assoc cancer could be merged, but contextual indication is different
  - Jamie: need to determine whether to model this as structured/computable vs. text blob
  - Liz: what relationship/statement are we trying to capture?
  - Kevin: need to differentiate between patient's diagnosis from what the drug treats
  - May: this references a knowledge artifact, may appear to be prescriptive when it isn't
  - Jamie: could use 1 associated pheno field and move complexity to the interpretation section of the report
  - Liz: knowledge bases like ClinVar should hold the evidence, be used/referenced in overall report
- Jamie: remove associated cancer, update associated phenotype def to pertain more broadly
  - intent is to match a var observed in a patient to something in a knowledge base
- Patrick: need to capture "given the observed vars, patient is likely to develop this cancer type"
  - Jamie: that would be a diagnostic implication, not medication implication
  - Kevin R-P: must ensure add to docs to make it clear that cancer should be expressed as phenotype
- Jamie: differentiate risk-of from context (see FHIR-26945)
  - susceptibility is from germline test, different from genomic profile from a tumor
  - Kevin P: need a way to codify risk
  - Jamie: can leave some of that up to the code system
- Liz: is region coverage (% value) already in LOINC?
  - Patrick: none found
- Liz: exact start-end 81254-5
  - Patrick: that def is out-dated, references only start position, need to capture both start and end
  - 81302-2: looks good
  - need to review existing LOINC defs
    - [ ] Patrick Werner will review
- diagnostic-implication / therapeutic-implication: Do we need codes?
  - Will be in STU2 (voted on by group as a consolidation of multiple profiles)
- For new loincs, try to phrase as a question, send a link to the definition, send an example report, determine the data type
  - PGX details: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5253119/
- Jamie: variant implications (AA change, functional effect, pathogenicity) and diagnostic implications
  - should those statements be associated with variant or diagnostic?
  - Bob F: this is where the SEPIO framework could help, see VA work on statement modeling, need a balance between modeling as discrete components and capturing text blob
  - <reviewing concept defs in Sequence Ontology>
- Joel S: need to be able to associate multiple vars with a single outcome/effect/risk, too simplistic to tie implication to a single var
  - Kevin P: we can associate to a group of vars

Chat:

James Jones
https://confluence.hl7.org/display/CGW/CG-2020-09+WGM+Attendance
https://docs.google.com/spreadsheets/d/1dubwSbfbJ4K1_Ie93VXynEPXyQgudwzG9Vji-Hcvbfg/edit?usp=sharing

	Liz Amos https://loinc.org/submissions/ https://loinc.org/submissions/request/ https://loinc.org/81254-5/ https://loinc.org/81302-2/ Today 2:01 PM Kevin Power Also: https://loinc.org/81301-4/ Liz Amos would "risk-of" fall in prognostic implication? Kevin Power My goto article for those PGx terms: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5253119/ Arthur Hermann That is a very good question Liz ... also what we will need to be able to support is polygenic risk scores.... which by definition are risks
Additional IG content /documentation	Jamie looking for "volunteers" to go through recent slide presentations and create some introduction material to help promote use of the IG. Multiple examples could be walked through in the IG or in an external resource May: mCode created a searchable KB in Confluence to help implementers, will provide demo if needed Arthur: interns could increase bandwidth to get work done on the IG - can we get some?
Work on examples	https://docs.google.com/spreadsheets/d/1-Co8NmSh75y_1wqJysnLBcgT7-lL4JY3WRIcNPr5qr8/edit#gid=422847907 - example worksheet https://docs.google.com/spreadsheets/d/1zgT0GYQZHjUYD7N0mYNzJozqdPieW7G2yglh3Ayl6gU/edit#gid=1392234601 - list of observation components 1. go through current examples a. confirm each concept has representation - spreadsheet mapping exercise for b. confirm each USE CASE has representation - at least one walked through in detail for each use case
Tooling	GoFSH for structuredefs