FHIR Subgroup Meeting Notes 2017-18

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

Presiding co-chair: Gil Alterovitz

Schedule

September 10 - glossary + trackers

September 17 - primer, trackers, sequence resource

Past Dates

Oct 5 Schedules, V2/FHIR mapping

Oct 12 Connectathon/PGx use case, FHIR extensions alternatives

Oct 19 X

Oct 26 FHIR extensions alternatives

Nov 2 X

Nov 9 Xin/Fan present approach for R4

Nov 16 Cancelled (#1, #2 + gForge (delayed) )

Nov 23 VMC

Nov 30 X

Dec 7 R4 development

Dec 14 Gforge items (Xin/Fan)

Dec 21 X

Dec 28 X

Jan 4 GForge (bob dolin - sequence quality)

Jan 11 FHIR Unification

~~Jan 18 CDS - Bryn Rhodes~~

Jan 25 X CDS - Bryn Rhodes

Feb 1 X (WGM)

Feb 8 Unification

Feb 12 Unification

Feb 19 CANCELLED

Feb 26 - Unification

March 5 - Unification/IG

March 12 - Unification/IG

March 19 - CLinGen MVLD standard for clinical MolDx data (Subha Madhavan)/AMP-CAP reporting guidelines (Joseph Kane)

March 26 - IG

April 2 - CANCELLED

April 9 - CANCELLED

April 16 -Gforge items

April 23 - Gforge

April 30 - Gforge

May 7 - Sequence ontologies

May 14 - WGM

May 21 - Cancelled

May 28 - Cancelled

June 4 - Bob dolin “Bioinformatics-to-FHIR converter”

June 11 - Kevin Co-chair: outstanding trackers

June 18 - Ian Green SNOMED

June 25 - Larry Babb -making the Sequence resource definitional, switch over to FCC from now on, we share the same account as clingenomics group meeting

July 2 - Ian Green - SNOMED CT and its approach to Genomics

July 9 - decide NIB status

July 16 - continue discussion from June 25 and July 10 - sequence, variants, and grouping

July 23 - variant grouping, Orders&Observations -

July 30 - Sequence Resource trackers, Described Variant profile

August 6 - gForge trackers in group G

August 13 - gForge trackers - various stragglers

August 20 - negative major gForge trackers

August 27 - More trackers

September 3 - Labor Day

# FHIR Subgroup Meeting September 17, 2018

[**https://join.freeconferencecall.com/clingenomics**](https://join.freeconferencecall.com/clingenomics)

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## Sign In: (presiding co-chair - Gil Alterovitz)

1. Joel Schneider - NMDP/CIBMTR - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
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5. Gideon Giacomelli - BIH/Charité - gideon.giacomelli@charite.de
6. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
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10. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
11. Bret Heale -
12. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
13. Jon Neville - CDISC - jneville@cdisc.org
14. Ning Xie - BCH - ningxie2018@gmail.com
15. Xiangyin Chen - BCH - xchen04@gmail.com
16. Jamie Parker- Carradora Health - [jamie.parker@carradora.com](mailto:jamie.parker@carradora.com)

Agenda:

1. Genomics Primer/Glossary [document drafted by James](https://docs.google.com/document/d/1vWY7fHbSl0ZxkTJAXZ8gz8nHUgYrHlKBCYy2bTaxiXs/edit#) (version with initial feedback to be sent out after call for additional comments)
2. Simplification Requests: panel profile 16108 resolution? (**resolution = defer**)
3. Leftover FISH probe and Copy Number Change trackers (put into block)
4. PGx Functionality Updates (didn’t cover)
5. Sequence Resource (didn’t cover)

**(1) - Primer/Glossary**

[document drafted by James](https://docs.google.com/document/d/1vWY7fHbSl0ZxkTJAXZ8gz8nHUgYrHlKBCYy2bTaxiXs/edit#) after 8/27 call, everyone make any comments you want on there by 9/17

Summary: reducing text in several places where it was needlessly detailed, restructuring slightly. It now called “Genomics Introduction” to remove confusion of term “primer”, it now provides:

1. An overview briefly introducing how we use some terms
2. An overview of report structure
3. An introduction to variant detection methods

Also have a look at the glossary <https://docs.google.com/spreadsheets/d/1JP9gC1Daaz_pjYkLnrZdLHviFn-qFTZLgGXQmvi487c/edit#gid=1467712360> (also linked from the primer doc).

Andrea: there are overlaps with these terms in other specs as well, particularly for specimen, e.g., LIVD (<http://wiki.hl7.org/index.php?title=LIVD_-_FHIR_Mapping_Project> ), service catalog, LRI (<http://www.hl7.org/documentcenter/public_temp_E67CAC4D-1C23-BA17-0CA3C3A90EDB074D/standards/dstu/V251_IG_LRI_R1_STU3_2018JUN.pdf> ), as well as others

Bob M: can check with ASHI as well

**(2) Panel Profile**

**-Discussion from last week**

**(3) - Leftover FISH and Copy Number Change Trackers**

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16266) | 16266 | Affirmative | Bob Dolin |
| **Summary** | Move arrCHG-ratio from Described Variant to Copy Number Change profile, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-described-variant.html> | | |
| **Resolution**  **Notes** | **N/A** | | |
| **Details** | Move arrCHG-ratio component from Described Variant to Copy Number Change profile. Rationale is that this component is relevant for microarray tests, where the ratio implies the number of copies. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #117 - A-C-Submitted by: Bob Dolin (Elimu Informatics) | | |
| **Disposition** | Persuasive - put into block | | |
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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16269) | 16269 | Affirmative | Bob Dolin |
| **Summary** | FISH Panel should have 0..\* FISH probes, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-chrom-fish.html> | | |
| **Resolution**  **Notes** | **N/A** | | |
| **Details** | Change cardinality from Chromosome Analysis FISH panel to FISH Probe from 0..1 to 0..\*. Rationale: Many findings are a summary based on an observation of multiple probes all at once. For instance, here is an ISCN notation that summarizes a finding based on 3 nearby probes, all present in triplicate, suggesting Down's Syndrome: 'ish 21q22(D21S259/D21S341/D21S342x3)'. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #120 - A-C-Submitted by: Bob Dolin (Elimu Informatics) | | |
| **Disposition** | Persuasive with mod - mod: *we are removing fish panel profile altogether, see tracker* [*16869*](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16869&start=0)  *No change necessary* | | |
|  |  |  |  |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16270) | 16270 | Affirmative | Bob Dolin |
| **Summary** | Add copy-number component (LOINC 82155-3) to Copy Number Change profile, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-copy-num-chg.html> | | |
| **Resolution**  **Notes** | **N/A** | | |
| **Details** | Add copy-number component (LOINC 82155-3) to Copy Number Change profile. Rationale: Need a way to indicate, for instance, that this ISCN finding, arr 21q22(35, 900, 000-48, 100, 000)x3', shows 3 copies of the specified region. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #121 - A-C-Submitted by: Bob Dolin (Elimu Informatics) | | |
| **Disposition** | Persuasive - block it | | |
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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16271) | 16271 | Affirmative | Bob Dolin |
| **Summary** | Add property 'name' to Device Component FISH Probe profile, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/devicecomp-fish-probe.html> | | |
| **Resolution**  **Notes** | **N/A** | | |
| **Details** | Add property 'name' to Device Component FISH Probe profile as a codeable concept. Rationale: Probes can have names and identifiers (e.g. D21S259, D21S341, D21S342), useful for looking them up. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #122 - A-C-Submitted by: Bob Dolin (Elimu Informatics) | | |
| **Disposition** | Persuasive - block | | |

**(4) - PGx functionality trackers**

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| **Details** | SNOMED has released PGx concepts like the following:  http://browser.ihtsdotools.org/?perspective=full&amp;conceptId1=738535003&amp;edition=us-edition&amp;release=v20180301&amp;server=https://prod-browser-exten.ihtsdotools.org/api/snomed&amp;langRefset=900000000000509007  They take the form of "[GENE] [STATUS]"  Should we allow for these to be delivered? We have our own profiles that describe sending the STATUS (as a LOINC answer list), and it can reference a Genotype/Haplotype/Variant, which from "Computable Genetic Findings" allows for GENE. However, I am not sure how we support delivering the SNOMED codes I reference above?  Also consider: Should these SNOMED codes be delivered as report-level Interpretations rather than Genetic Impacts? | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #126 - A-Q-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16081) | 16081 | Affirmative | Kevin Power |
| **Summary** | New PGx Impact: Transporter, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> | | |
| **Details** | In this paper, work was done to standardize nomenclature for reporting PGx results. This table summarizes:  https://www.nature.com/articles/gim201687/tables/2  Under 'Medication Impact', we have profiles that do match up to the "Term/gene category" that are called out above:  Phenotype: high-risk genotype = "High Risk Allele (83009-1)",  Phenotype: drug-metabolizing enzymes = "Genotype Medication Metabolism Impact (53040-2)"  We are missing "Phenotype: transporters" (example given was SLCO1B1: Increased Function, Normal Function, etc ...) - should we add transporters as a new profile? | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #127 - A-Q-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16082) | 16082 | Affirmative | Kevin Power |
| **Summary** | PGx Impact - Allele functional status, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> | | |
| **Details** | In this paper, work was done to standardize nomenclature for reporting PGx results. This table summarizes:  https://www.nature.com/articles/gim201687/tables/2  Under 'Medication Impact', we have profiles that do match up to the "Term/gene category" that are called out above:  Phenotype: high-risk genotype = "High Risk Allele (83009-1)",  Phenotype: drug-metabolizing enzymes = "Genotype Medication Metabolism Impact (53040-2)"  We are missing "Allele functional status" (increased function, normal function, etc ...) - Should we add a new profile for Allele Functional Status? | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #128 - A-Q-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16174) | 16174 | Affirmative | Kevin Power |
| **Summary** | PGx Impact - Multiple Levels of Evidence, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> | | |
| **Details** | In the Genetic Impact profile, the level of evidence is 0..1 and perhaps should be 0..\*? For example, in the case of PGx, CPIC and PharmGKB (and maybe others) might have different levels.  Reviewing the gene-drugs on CPIC (https://cpicpgx.org/genes-drugs/), I can imagine someone wanting to send CPIC Level, PharmGKB Level of Evidence, and even PGX on FDA Label to help the receiving system better drive the appropriate usage of the impact.  NOTE - Perhaps "PGX on FDA Label" is not a level of evidence but is instead its own component? Or maybe something else?  NOTE2 - This certainly falls into the category of "pre-coordinating CDS", and as such should perhaps fall into another "knowledge resource".  Please see the following page for more background: https://cpicpgx.org/prioritization/ | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #142 - A-S-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16175) | 16175 | Affirmative | Kevin Power |
| **Summary** | Genetic Impact - Add ACMG reference for level of evidence, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> | | |
| **Details** | Can we support the ACMG "Criteria for classifying pathogenic/benign variants"? See here: https://www.acmg.net/docs/standards\_guidelines\_for\_the\_interpretation\_of\_sequence\_variants.pdf (page 8-9).  My sense is that there can be multiple categories associated with a variant, and I am not sure we can support that as our profiles are structured today?  We should also consider how we could allow the specific category (PS1, PM2, etc..) to be sent as well as the Evidence (Strong, Moderate, etc ...) | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #143 - A-S-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16177) | 16177 | Affirmative | Kevin Power |
| **Summary** | Somatic Impact - Support MVLD levels of evidence, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/somatics.html> | | |
| **Details** | Need to support multiple MVLD classifications for Somatic Variants. See the attached example spreadsheet showing various scenarios. Also, see the following link for more guidelines: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5728662/ | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #144 - A-S-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** | Looked at link 8/27, will need to add this information in after further consideration | | |

**(5)**

* [Current functionality comparison sheet](https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg_xpwir8rEORkNjMfemvqxE0/edit#gid=0)
  + Need to have a picture of where one resource should be used vs the other
    - In particular where there is overlap

# FHIR Subgroup Meeting September 10, 2018

[**https://join.freeconferencecall.com/clingenomics**](https://join.freeconferencecall.com/clingenomics)

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

## Sign In: (presiding co-chair - Gil Alterovitz)

1. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
2. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
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15. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
16. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)

Agenda:

1. Complex Variants vs SequencePhaseRelation: 16262 + 16808 (resolutions proposed)
2. Genomics Primer/Glossary [document drafted by James](https://docs.google.com/document/d/1vWY7fHbSl0ZxkTJAXZ8gz8nHUgYrHlKBCYy2bTaxiXs/edit#) (introduced doc, make comments over the next week and we’ll check back)
3. Simplification Requests: 16108 (started discussion, inconclusive)

Discussion:

**(1) - Complex Variants vs SequencePhaseRelation**

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16262) | 16262 | Affirmative | Bob Dolin |
| **Summary** | Change LOINC answer list for complex variant type, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-complex-variant.html> | | |
| **Resolution**  **Notes** | **persuasive with mod**  **Bob D will provide details of mod:** | | |
| **Details** | Complex variation profile, component Complex Variant Type (LOINC 81263-6): Suggest we remove "haplotype" and "hemizygous" from LOINC answer set. Rationale is that we now have a Haplotype class, so shouldn't allow folks to use the complex variant class to indicate a haplotype. Also, hemizygous is an answer for allelic state, so in and of itself does not constitute a complex variant. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #113 - A-C-Submitted by: Bob Dolin (Elimu Informatics)-Tue, 14 Aug 2018 - by Bob Milius-Lloyd: we can make our own answer list from a subset or superset of another answer list.-Clem: answer list items described in clinvar-Clem: clarify which ones we want to retain-Bob D: this is minor, does it introduce redundancy?-persuasive with mod- -Tue, 14 Aug 2018 - by Bob Dolin-discussed in committee today, with a suggested modification that rather than removing "haplotype" and "hemizygous" from the LOINC answer set, we'd instead restrict them from use in the context of this profile. -Tue, 21 Aug 2018 - by Kevin Power-Resolution proposed should consider the following tracker:-**https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&amp;tracker\_item\_id=16808**- -Thu, 23 Aug 2018 - by Bob Dolin-I would also propose that we remove the link from SequenceConfiguration to ComplexVariant (because it doesn't work as intended, and is redundant). ComplexVariant has a 0..\* hasMember relationship to DescribedVariants, and the cis/trans relationship of DescribedVariants can be asserted via SequenceConfiguration.-So, revised proposed resolution:-1. In component Complex Variant Type (LOINC 81263-6), restrict from use the values "haplotype" and "hemizygous" from the LOINC answer set.-2. Remove the link from SequenceConfiguration to ComplexVariant | | |
| **Disposition** | **mod proposed: -1. In component Complex Variant Type (LOINC 81263-6), restrict from use the values "haplotype" and "hemizygous" from the LOINC answer set.-2. Remove the link from SequenceConfiguration to ComplexVariant**  **-When restricting the list we should make a note explaining why (point to an example of where that information should be represented instead**  **-haplotype:**  **Bob M: HLA would use a haplotype observation (if they are on different genes) with each allele in a complex variant observation. to group complex variants, would not need to specify haplotype on the complex variant.**  **Bob D: It seems like we’re making you use a workaround**  **Gil: Should at least leave notes/examples suggesting how to use this field, particularly if there are multiple ways to represent the data.**  **-hemizygous: the act of restricting the valueset may not be worth it, still make a suggestion for which way to represent info instead, particularly with the V2 mapping to FHIR.**  **)**  **-SeqPhaseRel can still link to described/structural variant, which could create issues.**  **Not Persuasive with mod: make a note on the bottom of** [**http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html**](http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html) **about this** | | |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16808) | 16808 | Negative-Major | FHIR Bot |
| **Summary** | Complex variants distinguish cis from trans - 2018-May Genomics #36, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> | | |
| **Resolution**  **Notes** | **Will likely need some work to clarify how Sequence Phase Relationship and Complex Variant Type work together.**  **Needs more thought, but perhaps remove the link between SeqPhase Rel and Complex Variant?** | | |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Figure 5: Cis or Trans  ---  Comment:  Unclear - I don't recall discussion plus the complex variants distinguish this (I think).  ---  Summary:  Complex variants distinguish cis from trans | | |
| **Follow-ups** | -Wed, 06 Jun 2018 - by Kevin Power-Similar thoughts are mentioned in other trackers, this needs more detailed follow-up.-Fri, 10 Aug 2018 - by Kevin Power-Note the resolution marked on this tracker:-<https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&amp;tracker_item_id=16173>-With this change, the "Sequence Phase Relation" can tie together any sort of Variation, and upon review, there is some overlap with "Complex Variation Type" answers:-Compound heterozygous | Double heterozygous | Haplotype | Hemizygous- | | |
| **Disposition** | **Persuasive with mod:**  **Remove the link from SequenceConfiguration to ComplexVariant** | | |

**(2) - Primer/Glossary**

[document drafted by James](https://docs.google.com/document/d/1vWY7fHbSl0ZxkTJAXZ8gz8nHUgYrHlKBCYy2bTaxiXs/edit#) after 8/27 call, everyone make any comments you want on there by 9/17

Also have a look at the glossary <https://docs.google.com/spreadsheets/d/1JP9gC1Daaz_pjYkLnrZdLHviFn-qFTZLgGXQmvi487c/edit#gid=1467712360> (also linked from the primer doc).

Andrea: there are overlaps with these terms in other specs as well, particularly for specimen, e.g., LIVD (<http://wiki.hl7.org/index.php?title=LIVD_-_FHIR_Mapping_Project> ), service catalog, LRI (<http://www.hl7.org/documentcenter/public_temp_E67CAC4D-1C23-BA17-0CA3C3A90EDB074D/standards/dstu/V251_IG_LRI_R1_STU3_2018JUN.pdf> ), as well as others

Bob M: can check with ASHI as well

**(3) - Genetics Panel profile**

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16108) | 16108 | Affirmative | Kevin Power |
| **Summary** | Genetics Panel: Suggest removing, | | |
| **Proposed Resolution** | Not persuasive with mod: status=defer, keep the comments as future guidance | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> | | |
| **Details** | Defining a panel does not feel like a requirement the first draft of this IG should address. Supporting the Genetics Panel as defined would certainly complicate an implementation, and at this point, I don't believe the value warrants that complexity. I would recommend removing the panel profile.  The following text is included in the documentation about the Genetics Panel profile:  As shown in the diagram above, all of the observations may hang directly off of the diagnostic report However, they can also be part of a panel. In this version of the specification, no guidance is provided on when or if panels should be used. This is left up to the discretion of the reporting lab. Observations might be organized on the basis of subject, specimen, chromosome, gene, condition/disease, medication or other appropriate measure. The recursive "hasMember" relationship on panel supports a nested tree-structure of panels if appropriate, though more than two levels of panels is likely excessive.  Any organization of observations into panels or sub-panels is purely for navigation and presentation purposes. It carries no additional "meaning". Each observation can be interpreted on its own without knowing the associated panel or sub-panel. The organization of observations in panels does not assert any relationship between observations. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #135 - A-C-Submitted by: KEVIN POWER (CERNER) | | |
| **Discussion** | Jamie: Mark as draft???  Bob M: could move toward definitional panels, rather than supporting anything lab  Bret: is this looking to model what was tested?  Kevin: currently more for grouping tests that were performed,  Lloyd: panel was put in to highlight the fact that diagnostic reports may contain/require navigation, that we don’t require all information to be linked directly to the diagnostic report.  Bob M: is it common for labs to report multiple panels in a report?  Andrea: They are created in order-result pairs in the lab  Lloyd: 2 types/uses for the word “panel” here, 1- ordered tests, 2- organizing results (apart from which tests were ordered).  ================  Andrea: built into LIS and specimen collection/CLIA: if you order X you will get Y back from the lab. Starts with the lab, they will list the tests they can perform, including what results are going to be presented.  Gil: Lets get some examples for labs/panels and see how we can map them with our profile.  Andrea: Would not recommend creating a new definition for panel, but defer to regulatory aspects in the area for "panel." A panel would be an order for multiple items. LOINC sometimes also calls it a “battery” of tests, from the order side. For a US lab to implement, it has to be CLIA compliant. (<https://www.ecfr.gov/cgi-bin/retrieveECFR?gp=1&SID=9b42cf158b7c913fe20af21d8d820148&ty=HTML&h=L&mc=true&n=pt42.5.493&r=PA>)  Bret: does CLIA require specific syntax for reporting?  Lloyd: we’re not defining which panels exist, we are telling implementers that information may be coming from labs as packed in panels, so we need to be able to support going through that. There’s no complete standardized breakdown of these panels so we will need to be flexible.  Andrea: <https://en.wikipedia.org/wiki/Test_panel> from CLIA, "(4) For antimicrobial susceptibility testing, a laboratory must indicate which drugs are routinely included in its test panel when testing patient samples. In the CLIA section addressing HLA testing, "(3) Use a panel that contains all the major HLA specificities and common splits. If the laboratory does not use commercial panels, it must maintain a list of individuals for fresh panel bleeding. "  Gil: proposed solutions:   1. Leave as is-needs feedback after testing (Kevin would be willing to withdraw) 2. Defer-comment will be reevaluated next ballot | | |

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# Chat history

# Kevin Power 11:39AM: Published version of the LRI (for those interested):<http://www.hl7.org/documentcenter/public_temp_E67CAC4D-1C23-BA17-0CA3C3A90EDB074D/standards/dstu/V251_IG_LRI_R1_STU3_2018JUN.pdf>

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# FHIR Subgroup Meeting August 27, 2018

[**https://join.freeconferencecall.com/clingenomics**](https://join.freeconferencecall.com/clingenomics)

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

## Sign In: (presiding co-chair - Gil Alterovitz)

1. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
2. Joel Schneider - NMDP/CIBMTR - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
3. Patrick Werner - Molit Institute / Heilbronn University - [patrick.werner@molit.eu](mailto:patrick.werner@molit.eu)
4. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
5. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
6. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
7. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
8. Grant Wood - Intermountain Healthcare - grant.wood@imail.org
9. Jamie Parker- Carradora Health- [jamie.parker@carradora.co](mailto:jamie.parker@carradora.co)m
10. Ning Xie - BCH-ningxie2018@gmail.com
11. Dorina Bratfalean - CDISC - [dbratfalean.external@cdisc.org](mailto:dbratfalean.external@cdisc.org)
12. Ling teng -BCH -tenglingling@gmail.com
13. Deepak Sharma - Mayo Clinic - [sharma.deepak2@mayo.edu](mailto:sharma.deepak2@mayo.edu)
14. Jungang Zou - BCH - jungang.zou@gmail.com
15. Julian Sass - Berlin Institute of Health - [julian.sass@bihealth.de](mailto:julian.sass@bihealth.de)
16. Bob Milius - CIBMTR/NMDP - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
17. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
18. Scott Robertson - Kaiser Permanente - [scott.m.robertson@kp.org](mailto:scott.m.robertson@kp.org)
19. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
20. Andrea Pitkus - [apitkus@gmail.com](mailto:apitkus@gmail.com)

Agenda:

1. Genomics Primer Guidance - discussed, [document later drafted by James](https://docs.google.com/document/d/1vWY7fHbSl0ZxkTJAXZ8gz8nHUgYrHlKBCYy2bTaxiXs/edit#) (document not available/discussed during call)
2. Simplification Requests - discussed 16253 and 16259
3. PGx Functionality Updates - didn’t get to
4. “What was looked at” - didn’t get to

Discussion:

**(1) - “Primer” trackers**

(Kevin)

1. Do we remove it with the thought that it is not necessary, at least right now?
   1. Clem: Maybe “primer” isn’t the best term here, can be misleading
   2. Perhaps “Refresher/Introduction”
2. Do we keep it but simply refer to other online resources for those that need a primer?
   1. Will need to make sure we align with these definitions, may have to refine them from there, can even reach out to the defining body to see if they ought to update the definitions on their end
3. Do we keep it and maintain this ourselves?

(Andrea)

* I think it might be really helpful to have a glossary and use terminology aligned with O&O and related workgroups to avoid confusion, crosstalk, etc.
* Here's an example of one for the recent LIVD build: <http://build.fhir.org/ig/HL7/livd/glossary.html>
* Comment added to [the glossary tracker](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16513), 16513

Jamie Parker and Arthur both voiced consent toward option 3

Lloyd:

* Suggest that we separate how our definitions line up with other sources to the bottom or an appendix, etc so not to clutter the “working glossary” to improve usability for developers.

Bob D:

* This location is important, but the component descriptions certainly need to align \*internally\* first, then we could do a Q/A and extract them into a glossary.

Lloyd:

* I think we should limit external links as much as possible for describing the core concepts.

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16698) | 16698 | Negative-Major | FHIR Bot |
| **Summary** | Remove or shrink text - 2018-May Genomics #6, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/background.html> | | |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: The technique used to detect the variation, alignment with the borders of major features of the underlying sequence (e.g. variants), the presumed cause of the difference (insertion of extra copies, inversion of a portion of the sequence, common patterns in the population (a common set of variations typically appear together), a known effect(a particular collection of changes may result in a known physical manifestation or disease - aka. a phenotype)  Proposed Wording: remove or shrink  ---  Comment:  Not sure this is relevant or necessary.  ---  Summary:  Remove or shrink text | | |
| **Follow-ups** | Wed, 06 Jun 2018  By Kevin Power I reviewed this but am not sure if this should be left alone, reworded, or removed. Adding for follow-up by another co-chair. | | |
| **Disposition** | **Persuasive with mod -** | | |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16740) | 16740 | Negative-Major | FHIR Bot |
| **Summary** | Consider consolidating and using simple NCBI or GHR reference description - 2018-May Genomics #18, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/background.html> | | |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Direct sequencing techniques follow a multistep pathway. The initial phase of a sequencing run produces raw sequence with a qualification of the quality of the raw sequence. The raw sequence is then subject to further computational analysis. In traditional sequencing the product is a single 'read' and the initial step is often the final step. However, in techniques such as next-generation sequencing, the initial step is massively parallel and many 'reads' result. The next phase for massively parallel sequencing techniques is to align the 'reads' to each other and a reference sequence, which acts as a scaffold to aid in assembling 'reads, in order to create longer, contiguous series of sequences. The next phase for traditional and highly-parallel sequencing techniques is to compare the sequence, or sequence of the assembled 'reads, to a reference sequence. This comparison produces indientification of variance and quality scores.  ---  Comment:  Don't think we should be going so far. Why not just cite a good reference - there are plenty of over view descriptions at NCBI and Genetic Home Reference. | | |
| **Follow-ups** | Wed, 06 Jun 2018  By Kevin Power Additional feedback requested - my initial thoughts:\* Not sure the title of the section is really accurate? Perhaps it should be 'Variant Detection'?\* I tend to agree with Clem that we should have less language here and refer to other materials (NCBI or GHR are good options). | | |
| **Disposition** | **Persuasive with mod -** | | |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16743) | 16743 | Negative-Major | FHIR Bot |
| **Summary** | Redundant words, the reference is a sequence+P22 - 2018-May Genomics #19, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/background.html> | | |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Inferential techniques for determining variants in sequences rely upon a standard as a reference.  ---  Comment:  Redundant words, the reference is a sequence+P22 . | | |
| **Follow-ups** | Tue, 05 Jun 2018  byLloyd McKenzieThe last two paragraphs in this section don't flow well from the first two paragraphs. They provide a lot of detail that's not clear is necessary. If we decide this detail is needed in order to use the specification, we need to build up to it in small easy stages, not jump right into the deep end of the pool. | | |
| **Disposition** | **Persuasive with mod** | | |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16748) | 16748 | Negative-Major | FHIR Bot |
| **Summary** | Possible inaccuracy - 2018-May Genomics #20, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/background.html> | | |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Inferential techniques for determining variants in sequences rely upon a standard as a reference. For example, inference from microarrays is done through comparing the signal produced by a sample to a known standard reference. In the case of microarrays, the probe set used conveys the positions interrogated. In Mass spectrometry, the standard is the mass of a known reference. In inferential techniques the output is the presence or absence of variation with a technique specific measurement of accuracy.  ---  Comment:  Don't think this is exactly correct and worry that we will have to spend time on this when it would be easy to cite well polished reference (or quotes from it).  ---  Summary:  Possible inaccuracy | | |
| **Follow-ups** | Tue, 05 Jun 2018  By Lloyd McKenzie See comment on 16743 | | |
| **Disposition** | **Persuasive with mod -** | | |

Google Documents creation for editing this section:

1. Glossary of specific terms (pulled from our component definitions)
   1. Appendix describing differences in definitions here vs elsewhere
2. Introduction to the Genomics space (copy this primer and allow edits to get it consolidated/up to date)
   1. Comments from the doc will have to be logged officially as trackers to be implemented
   2. Could they be used in resolution to these comments?

**(2) - Simplification Requests**

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16253) | **16253** | Affirmative | Bob Dolin |
| **Summary** | Enhance overall understandability of CG IG, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/index.html> | | |
| **Details** | Consider including an overall graphic of the model. The IG is a bit hard to follow because the diagram is distributed across pages. The high level graphic need not include all the attributes. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #104 - A-S-Submitted by: Bob Dolin (Elimu Informatics) | | |
| **Disposition** | **persuasive! - who wants to do it?**  **Showing all relationships will be dizzying, will have to restrict relationships as an overview and then add in levels of detail. Imagemap would really help with this.**  **Clem - packaging diagrams into a PDF would be useful as well, maybe with pieces given further description in place as well.**  **Lloyd - the automated UML diagrams are just for the base resources, the diagrams currently in the IG are manually maintained (aka made manually with enterprise architect and exported, not open source). No real hope of getting them automated. Can reach out to Lynn to see if free licenses are available.**  **A tool that allows image maps and links would be ideal, if we are considering different tools.**  **Jamie Parker - could this be done with Clinfhir?**  **Kevin - tried to load current items into ClinFhir and there were import issues, let’s look into resolving this** | | |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16259) | **16259** | Affirmative | Bob Dolin |
| **Summary** | Simplify Genetic Impact, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> | | |
| **Resolution**  **Notes** | **N/A** | | |
| **Details** | There are SEVEN Genetic Impact classes (inherited disease pathogenicity, high risk allele, genotype medication metabolism impact, genotype medication efficacy impact, somatic diagnostic impact, somatic prognostic impact, somatic predictive impact). Consider collapsing all these into a single Genetic Impact class. Rationale: [1] the large number of profiles adds complexity to a model where a critical focus right now needs to be on the structured representation of variants and observed sequences; [2] Representation of genetic impact is evolving, so it might be premature to try to nail down the profiles. (We could for now, as an interim, have a single Genetic Impact class, along with a value set of LOINC Genetic Impact observations and associated LOINC answer codes) | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #110 - A-S-Submitted by: Bob Dolin (Elimu Informatics) | | |
| **Disposition** | **Not Persuasive with Mod?? - see discussion - mark somatic impact profiles as draft**  **Lloyd :**  **The reasons for splitting: splitting restricts the available fields and increases interoperability. We don’t have to have all the impacts listed and choose when they’re ready.**  **Bob D:**  **Maybe compromise by reducing the particularly unripe ones, particularly the 3 somatic profiles (prognostic/predictive/diagnostic).**  **Clem:**  **Tight connection with PGx, should be considered**  **Bob D:**  **Most radical proposal: collapse to 1 class and make sure all the detail is in there.**  **Clem:**  **What about 2: separating drug-related from other conditions**  **Bob D:**  **Will need to link to medication and condition, but both aren’t always relevant.**  **Lloyd:**  **We can designate some profiles as “Draft” (maturity level 0, would not impact the rest of the IG)**  **Bob D:**  **I would mark the somatic impacts as draft, the others have seen more discussion.** | | |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16108) | 16108 | Affirmative | Kevin Power |
| **Summary** | Genetics Panel: Suggest removing, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> | | |
| **Details** | Defining a panel does not feel like a requirement the first draft of this IG should address. Supporting the Genetics Panel as defined would certainly complicate an implementation, and at this point, I don't believe the value warrants that complexity. I would recommend removing the panel profile.  The following text is included in the documentation about the Genetics Panel profile:  As shown in the diagram above, all of the observations may hang directly off of the diagnostic report However, they can also be part of a panel. In this version of the specification, no guidance is provided on when or if panels should be used. This is left up to the discretion of the reporting lab. Observations might be organized on the basis of subject, specimen, chromosome, gene, condition/disease, medication or other appropriate measure. The recursive "hasMember" relationship on panel supports a nested tree-structure of panels if appropriate, though more than two levels of panels is likely excessive.  Any organization of observations into panels or sub-panels is purely for navigation and presentation purposes. It carries no additional "meaning". Each observation can be interpreted on its own without knowing the associated panel or sub-panel. The organization of observations in panels does not assert any relationship between observations. | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #135 - A-C-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |

**(3) - PGx Functionality Updates**

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16080) | 16080 | Affirmative | Kevin Power |
| **Summary** | Allow for SNOMED terms for PGx Impacts, | | |
| **Details** | SNOMED has released PGx concepts like the following:  http://browser.ihtsdotools.org/?perspective=full&amp;conceptId1=738535003&amp;edition=us-edition&amp;release=v20180301&amp;server=https://prod-browser-exten.ihtsdotools.org/api/snomed&amp;langRefset=900000000000509007  They take the form of "[GENE] [STATUS]"  Should we allow for these to be delivered? We have our own profiles that describe sending the STATUS (as a LOINC answer list), and it can reference a Genotype/Haplotype/Variant, which from "Computable Genetic Findings" allows for GENE. However, I am not sure how we support delivering the SNOMED codes I reference above?  Also consider: Should these SNOMED codes be delivered as report-level Interpretations rather than Genetic Impacts? | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #126 - A-Q-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16081) | 16081 | Affirmative | Kevin Power |
| **Summary** | New PGx Impact: Transporter, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> | | |
| **Details** | In this paper, work was done to standardize nomenclature for reporting PGx results. This table summarizes:  https://www.nature.com/articles/gim201687/tables/2  Under 'Medication Impact', we have profiles that do match up to the "Term/gene category" that are called out above:  Phenotype: high-risk genotype = "High Risk Allele (83009-1)",  Phenotype: drug-metabolizing enzymes = "Genotype Medication Metabolism Impact (53040-2)"  We are missing "Phenotype: transporters" (example given was SLCO1B1: Increased Function, Normal Function, etc ...) - should we add transporters as a new profile? | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #127 - A-Q-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16082) | 16082 | Affirmative | Kevin Power |
| **Summary** | PGx Impact - Allele functional status, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> | | |
| **Details** | In this paper, work was done to standardize nomenclature for reporting PGx results. This table summarizes:  https://www.nature.com/articles/gim201687/tables/2  Under 'Medication Impact', we have profiles that do match up to the "Term/gene category" that are called out above:  Phenotype: high-risk genotype = "High Risk Allele (83009-1)",  Phenotype: drug-metabolizing enzymes = "Genotype Medication Metabolism Impact (53040-2)"  We are missing "Allele functional status" (increased function, normal function, etc ...) - Should we add a new profile for Allele Functional Status? | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #128 - A-Q-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16174) | 16174 | Affirmative | Kevin Power |
| **Summary** | PGx Impact - Multiple Levels of Evidence, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> | | |
| **Details** | In the Genetic Impact profile, the level of evidence is 0..1 and perhaps should be 0..\*? For example, in the case of PGx, CPIC and PharmGKB (and maybe others) might have different levels.  Reviewing the gene-drugs on CPIC (https://cpicpgx.org/genes-drugs/), I can imagine someone wanting to send CPIC Level, PharmGKB Level of Evidence, and even PGX on FDA Label to help the receiving system better drive the appropriate usage of the impact.  NOTE - Perhaps "PGX on FDA Label" is not a level of evidence but is instead its own component? Or maybe something else?  NOTE2 - This certainly falls into the category of "pre-coordinating CDS", and as such should perhaps fall into another "knowledge resource".  Please see the following page for more background: https://cpicpgx.org/prioritization/ | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #142 - A-S-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16175) | 16175 | Affirmative | Kevin Power |
| **Summary** | Genetic Impact - Add ACMG reference for level of evidence, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> | | |
| **Details** | Can we support the ACMG "Criteria for classifying pathogenic/benign variants"? See here: https://www.acmg.net/docs/standards\_guidelines\_for\_the\_interpretation\_of\_sequence\_variants.pdf (page 8-9).  My sense is that there can be multiple categories associated with a variant, and I am not sure we can support that as our profiles are structured today?  We should also consider how we could allow the specific category (PS1, PM2, etc..) to be sent as well as the Evidence (Strong, Moderate, etc ...) | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #143 - A-S-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** |  | | |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16177) | 16177 | Affirmative | Kevin Power |
| **Summary** | Somatic Impact - Support MVLD levels of evidence, | | |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/somatics.html> | | |
| **Details** | Need to support multiple MVLD classifications for Somatic Variants. See the attached example spreadsheet showing various scenarios. Also, see the following link for more guidelines: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5728662/ | | |
| **Follow-ups** | -Sat, 12 May 2018 - by FHIR Bot-Vote: #144 - A-S-Submitted by: KEVIN POWER (CERNER) | | |
| **Disposition** | Looked at link, will need to add this information in after further consideration | | |

**Chat History:**

* + Clinical Genomics Work Group Host 11:07AM: <http://build.fhir.org/ig/HL7/livd/glossary.html>
  + Andrea Pitkus 11:08AM: point taken Clem. One of the issues even with HL7 experts is differing definitions for observation and observation result value. If as Gil indicates, there is an "official" observation operational definition
  + Bret Heale 11:16AM: i've a bad mic. I agree with Bob D and wanted to add that good examples are worth a lot as well
  + Andrea Pitkus 11:17AM: Primer is being used as a different definition as basic overview, not genomics primers
  + Bret Heale 11:49AM Impact type value set will be malleable - we'll be able to handle anything by adding new type. Linking to medication is an implementation issue, i.e. it would be rather silly not to link when appropriate (meaning the implementer), so, by having the seperate profiles we hand hold the implementer, making it easier but also increasing the number of profiles to support

# FHIR Subgroup Meeting August 20, 2018

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

## Sign In: (presiding co-chair - Kevin Power)

1. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
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3. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
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11. Joel Schneider - NMDP/CIBMTR - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
12. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
13. Ling Teng [-BCH-tenglingling@gmail.com](mailto:-BCH-tenglingling@gmail.com)
14. Ning Xie - BCH- ningxie2018@gmail.com

Agenda:

1. Overview of the remaining Neg-Major trackers:
   1. Missing LOINC codes - *move into block*
   2. Transplantation/HLA needs more complete narrative -*didn’t cover*
   3. Sequence Phase Relationship x Complex Variant - *discussed, needs thought*
   4. Inherited Disease Pathogenicity profile - *deferred*
2. “What was looked at” proposals - *discussed*
3. Seq + Obs - *topic introduced*

Discussion:

(a)

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16180) | 16180 |
| **Summary** | Inherited Disease Pathogenicity - Must have mode-of-inheritance value set, |
| **Commenter** | Kevin Power |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-inh-dis-path.html> |
| **Details** | I did a search in LOINC and did not find an appropriate code. The possible value set options:  <https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns>  <https://www.cancer.gov/publications/dictionaries/genetics-dictionary/def/mode-of-inheritance> |
| **Disposition** | *Persuasive - first link is a good start* |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16184) | 16184 |
| **Summary** | Genetic Impact - Need LOINC code for level of evidence, |
| **Commenter** | Kevin Power |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-impact.html> |
| **Details** | We need a LOINC code to deliver the ***Level of Evidence component*** on the Genetic Impact profile. My LOINC search did not turn up a suitable code. |
| **Disposition** | *Def*  *Generic list may be problematic… will need more thought* |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16239) | 16239 |
| **Summary** | Need an Observation.code for DescribedVariant, |
| **Commenter** | Kevin Power |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/obs-described-variant.html> |
| **Details** | We need to identify a LOINC code. We had ones for Discrete and Structural, but we do not have one for our current DescribedVariant profile. |
| **Disposition** | *Persuasive - either reuse an old one or make a new one* |
| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16244) | 16244 |
| **Summary** | Need a LOINC for Coordinate System, |
| **Commenter** | Kevin Power |
| **Links** | <http://build.fhir.org/ig/HL7/genomics-reporting/obs-described-variant.html> |
| **Details** | Need to identify a LOINC code for Coordinate System component.  Also - Note that the Terminology Bindings section (2.16.3) has "GeneticCoordinateSystem" listed many times. Might be a side effect of using TBD as the code? Or perhaps another issue? |
| **Disposition** | *Persuasive !* |

(b)

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16509) | 16509 |
| **Summary** | Transplantation/HLA needs more complete narrative, |
| **Commenter** | Bob Milius |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/2018May/transplants.html> |
| **Resolution**  **Notes** | **N/A** |
| **Details** | Genomic Profiling for Transplantation needs a more complete narrative, including a possible name change for the section. Include the considerations needs by the community, as well as current conventions for reporting, HLA nomenclature, why knowledge of chromosomal phasing of sequences is know is important, and how this community differs from other communities in its use of terms such as allele and haplotype.  Also, describe the HLA example in more detail, why different observation profile were used, perhaps with clinFHIR graph image to visually describe the relationships. |
| **Follow-ups** | Sat, 12 May 2018  By FHIR Bot Vote: #91 - NEG Submitted by: Bob Milius (National Marrow Donor Program) |
| **Disposition** | *Persuasive - lets get it done* |

(c)

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16808) | 16808 |
| **Summary** | Complex variants distinguish cis from trans - 2018-May Genomics #36, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | **Will likely need some work to clarify how Sequence Phase Relationship and Complex Variant Type work together.** |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Figure 5: Cis or Trans  ---  Comment:  Unclear - I don't recall discussion plus the complex variants distinguish this (I think).  ---  Summary:  Complex variants distinguish cis from trans |
| **Follow-ups** | Wed, 06 Jun 2018  By Kevin Power: Similar thoughts are mentioned in other trackers, this needs more detailed follow-up.  Fri, 10 Aug 2018  By Kevin PowerNote the resolution marked on this tracker: [16173](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16173)  With this change, the "Sequence Phase Relation" can tie together any sort of Variation, and upon review, there is some overlap with "Complex Variation Type" answers: Compound heterozygous | Double heterozygous | Haplotype | Hemizygous  *Clem: checked in with NCBI, definitions described haplotype as any cis combinations, and genotype as trans-grouped, not necessarily the same gene (but typically it is).*  *Joel: for HLA we have different way of describing genotypes - multi-locus, (phased/haplotype pair or unphased/can’t say anything about phase)* |
| **Disposition** | Needs more thought, but ought to remove link between SeqPhase Rel and complex var |

(d)

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16834) | 16834 |
| **Summary** | Separating it into a separate profile may make it seem disconnected. - 2018-May Genomics #43, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | **N/A** |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Figure 7: Inherited Disease Pathogenicity 53037-8  ---  Comment:  Formerly called variant clinical significance. With answer list  1. pathogenic  2. Likely pathogenic LA6704-6  3. Uncertain significance LA6705-3  4. Likely benign LA6706-1  5. Benign    Believe it is a always reported with each variant. Separating it into a separate profile may make it seem disconnected. This particular list is from a published genomics paper PMID 25741868 (PMCID: PMC4544753). But there may be a desire to tweak it. |
| **Follow-ups** | Mon, 30 Jul 2018  By Liz Amos, From Clem McDonald: The term (in LOINC - Genetic Variant Clinical Significance 53037-8) is now called Inherited Disease Pathogenicity (with same answer list). Two problems: 1) We know something about a variant. Ramping that up to an inherited disease is a leap too far. The new name is misleading. 2) it has been separated from the variant by putting it into Genetic impact with a one to many link to the observation. In current reports, the variant and assessment of pathogenicity are almost always paired. Like love and marriage, horse and carriage. This this separation and the many to one relation breaks this tight connection. Think it would be much better if this attribute was attached to the computable genetic finding at the top of Figure 1 where it tied to one or a set of variants.  Clinical significance may change over time as new knowledge becomes available, this approach leaves us more open to that later, but it may not be the right place for it  Clem’s concern is with usability and prominence of the contained information  Bret: from a workflow point of view: a lab does sequencing, and sends them off to a third party vendor which annotates the vcf,  Clem: how does that change the model of the report going to the clinic?  Kevin: the current model allows attaching many observations including an Overall Interpretation.  Clem: could we attach it to computable genetic finding instead? (As a component) |
| **Disposition** | Logged as negative,   1. status: change to Defer, resolution - not persuasive (for this ballot)    1. Automatically shows up for next cycle |

**(e) - what was looked at**

* [Need to address "what was looked at"](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16258&start=0) 16258
* [Comment to disagree that all things examined within range have to be represented in each variant - 2018-May Genomics #24](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16763&start=0) 16763

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| **Tracker** | 15889 (approach mentioned last week, later discussed in Zulip) |
| **Summary** | [Properties needed for range examined and human reference sequence assembly](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15889&start=0) |
| **Links** | <http://build.fhir.org/ig/HL7/genomics-reporting/> |
| **Resolution**  **Notes** | Add component 51959-5 Range(s) of DNA sequence examined to the Computable Genetic Finding profile. This would allow us, for instance, to specify the exact location for a haplotype or genotype call." |
| **Details** | Lloyd: In v2 we had 2 properties that appeared on both simple and complex variants that aren't exposed in the new model: Range examined: 51959-5 & Human reference sequence assembly version: 62374-4. Are they needed? If so, where should they go? |
| **Follow-ups** | Submitted on Sat12 May 2018 17:35:55 -0500 by FHIR Bot  Vote: #158 - A-Q  Submitted by: KEVIN POWER (CERNER)  Submitted on Wed01 Aug 2018 15:28:10 -0500 by Bob Dolin  From <https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16261>, suggest that we Add component 51959-5 Range(s) of DNA sequence examined to the Computable Genetic Finding profile. This would allow us, for instance, to specify the exact location for a haplotype or genotype call."  Submitted on Wed01 Aug 2018 16:39:19 -0500 by Kevin Power  Related to the following tracker:  <https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16763> |
| **Disposition** | Persuasive with above mod |

* [Zulip discussion](https://chat.fhir.org/#narrow/stream/43-genomics/subject/Capturing.20Genomic.20Panel.20Definitions)

We have basically two proposals on the table:

* Profile Observation (Regions-Studied) with components[] to define what was tested
  + Regions-Studied would “hang off” the report
  + Several codes mentioned in the Zulip, Bob D will put together a profile proposal
* Profile Device (Genetics-Assay) with extensions to define what was tested
  + Sub-optimal as many kits can be used for multiple tests

Reactions? Preferences? I lean towards profiling Observation.

(f) - seq + obs

* [Current functionality comparison sheet](https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg_xpwir8rEORkNjMfemvqxE0/edit#gid=0)
  + Need to have a picture of where one resource should be used vs the other
    - In particular where there is overlap

Will discuss next week

Chat History:

Bret Heale 11:09AM  
something like https://libguides.nvcc.edu/c.php?g=361218&p=2439383 for evidence levels? Or are we looking at something from the ACMG  
  
Andrea Pitkus 11:37AM  
https://www.mayomedicallaboratories.com/test-updates/attachment.php?id=47964  
  
Andrea Pitkus 11:38AM  
are you referring to the Classification such as in this report as to whether the findings are pathogenic?  
  
Andrea Pitkus 11:42AM  
Similar example from ARUP: http://ltd.aruplab.com/Tests/DownloadReport/2011954%2C%20Positive.pdf  
  
Andrea Pitkus 11:43AM  
at end of Mayo report, "All detected alterations are evaluated according to ACMG recommendations (Genet Med 2008:10(4):294−300). Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance

Andrea Pitkus 11:48AM  
http://www.archivesofpathology.org/doi/pdf/10.5858/arpa.2014-0250-CP?code=coap-site  
  
Andrea Pitkus 11:49AM  
This includes a section on NGS referral testing Bret described and the accreditation aspects  
for reference....

Andrea Pitkus 11:52AM  
"For inherited diseases, the most commonly applied classiﬁcation is divided into 5 categories: (1) pathogenic, (2) likely pathogenic, (3) uncertain clinical signiﬁcance, (4) likely benign, and (5) benign"

# FHIR Subgroup Meeting August 13, 2018

**PLEASE JOIN FREE CONFERENCE CALL**

[**https://join.freeconferencecall.com/clingenomics**](https://join.freeconferencecall.com/clingenomics)

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

## Sign In: (presiding co-chair - Gil Alterovitz)

1. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
2. Joel Schneider - NMDP/CIBMTR - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
3. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
4. Deepak Sharma - Mayo Clinic - [sharma.deepak2@mayo.edu](mailto:sharma.deepak2@mayo.edu)
5. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
6. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
7. Patrick Werner - Molit Institute / Heilbronn University - [patrick.werner@molit.eu](mailto:patrick.werner@molit.eu)
8. Dora Finkeisen - Molit Institute - [dora.finkeisen@molit.eu](mailto:dora.finkeisen@molit.eu)
9. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
10. Jamie Parker - Carradora Health - [jamie.parker@carradora.com](mailto:jamie.parker@carradora.com)
11. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
12. Julian Sass - Berlin Institute of Health (BIH) - [julian.sass@bihealth.de](mailto:julian.sass@bihealth.de)
13. Scott M Robertson - Kaiser Permanente - [scott.m.robertson@kp.org](mailto:scott.m.robertson@kp.org)
14. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
15. Andrea Pitkus- [apitkus@gmail.com](mailto:apitkus@gmail.com)
16. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
17. Ning Xie - BCH - ningxie2018@gmail.com
18. Ling teng -BCH- tenglingling@gmail.com

## Agenda:

1. Virus/Bacteria in the IG? Tracker 16686
2. Need Glossary - Tracker 16513
   1. What’s the roadmap here?
3. Block Vote prep
   1. commenters Clem, Amnon, Bob M, Scott Robertson
4. What was looked at?

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16686) | 16686 |
| **Summary** | Clarification needed on virus/bacteria sample - 2018-May Genomics #2, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/background.html> |
| **Resolution**  **Notes** | **Persuasive with mod - some parts are in scope, should clarify** |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Genetic reporting involves reporting information about the genetic characteristics of a sample. The sample might be a tissue sample from a human, an animal or, more rarely, a bacteria or virus. In humans and animals, the sample might be from "normal" tissue, transplanted tissue, reproductive tissue (egg or sperm) or abnormal tissue such as a tumor.  ---  Comment:  Should clarify that the sample could be a virus or bacteria but then it will usually be identified as an Isolate. Genetic-basic organism reports rarely need this kind of sophisticated reporting, and are usually reported as its susceptibility to an antibacterial/antiviral agent. So don't need to use a full mutation analysis.  ---  Summary:  Clarification needed on virus/bacteria sample |
| **Follow-ups** | Mon, 30 Jul 2018  By Liz Amos: From Clem McDonald: The text says: "Genetic reporting involves information about the genetic characteristics of a sample. The same might be a tissue sample from a human or an animal or more rarely from a bacteria or a virus." I suggest striking 'bacteria or virus' from the text because a) it was excluded from the scope b) our model and wording does not fit well with the genetics of these creatures (circular DNA) and the reporting styles for different viruses can be quite different c) we have to be specific about reporting their susceptibility to drugs d) labs now report these things as simple test results with one code so there is no urgency to accommodate them at this time. |
| **Disposition** |  |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16513) | 16513 |
| **Summary** | need glossary, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/2018May/background.html> |
| **Resolution**  **Notes** | **Persuasive -**  **Will have to reconcile before publishing**  **what’s the roadmap here?**  **-”variant/haplotype/genotype” definition compare with VMC & SO**  **-solidify definitions in profiles themselves**  **-should have clear statements of how ours differ**  **-Google Doc tracking progress on this in the works by the IM, will likely need a CG-specific version**  **Use SO/VMC as starting point -** |
| **Details** | I think we need a glossary of genetic terms that we either point to or include in this IG, and other workgroup products. I think Clem developed one for V2, we should take another look at it.  Perhaps modeling group has one we should use. |

**TOPIC 3: Trackers near resolution for block vote-**

**commenters Clem, Amnon, Bob M, Scott Robertson**

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16325) | 16325 |
| **Summary** | 'haplotype' in medical genetics, |
| **Links** | N/A |
| **Resolution**  **Notes** | **Consider Question Answered:**  **The diagrams indicate it is an optional part of the model. As we refine this IG, we can try to make it more MD friendly, but IGs are typically targeted at implementers.** |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16866) | 16866 |
| **Summary** | Overall comments on Appendix B - 2018-May Genomics #54, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/cgapps.html> |
| **Resolution**  **Notes** | **Proposed resolution:**  **1) Add a disclaimer that the apps referenced here may not use this IG but are instead included as reference and examples.**  **DISCLAIMER: The Clinical Genomics Working Group was not involved in the design or development of these apps, and makes no claim they are compatible with this Implementation Guide nor any specific version of FHIR. They are included here for reference and examples.**  **2) Fix links**  **https://www.ncbi.nlm.nih.gov/pubmed/26198304 under Fig. 1**  **https://www.ncbi.nlm.nih.gov/pubmed/27018265 under Fig. 5** |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16489) | 16489 -Bob withdrawing |
| **Summary** | 'Genomic Alelle start-end' should use 'start' and 'end' instead of 'low' and 'high', |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/2018May/obs-described-variant.html> |
| **Resolution**  **Notes** | **Not Persuasive:**  **The Range data type in FHIR is a generic data type and uses the terms "low" and "high"**  **The component name refers to 'start' and 'end', which should help clarify**  **See other feedback from NCBI via Clem in the Follow-ups section.** |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16793) | 16793 |
| **Summary** | Discussion needed on change from display names on 84414-2 - 2018-May Genomics #32, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | **Not Persuasive**  **This can remain as a CodeableConcept - We will not dictate specific code systems to use at this time** |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16789) | 16789 |
| **Summary** | Discussion needed on change from display names on 84413-4 - 2018-May Genomics #31, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | **Change datatype of value from valueCodeableConcept to valueString. See comments for discussion.** |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Existing Wording: Figure 5: Genotype 84413-4  Proposed Wording: Genotype Display Name 84413-4  ---  Comment:  I understand why you want to shorten, but the change could mislead. These are not solid codes for genotype or haplotype. Would like to find a way to link from the figure (or content below them) to the LOINC code, description and answer list. Have linked to the answer list in the change document but these early tables are a bit more digestable. Lets talk.  ---  Summary:  Discussion needed on change from display names on 84413-4 |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16871) | 16871 |
| **Summary** | Specialization for somatic variant might not be necessary - 2018-May Genomics #56, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html> |
| **Resolution**  **Notes** | **Not Persuasive** |

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| [**Tracker**](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16938) | 16938 |
| **Summary** | knowledge representation - 2018-May Genomics #78, |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | **Considered for Future Use** |
| **Details** | Submitted by: Amnon Shabo (Philips)  Existing Wording: At present, impacts are noted as explicit observations about the patient/subject. However, it's not clear this is the correct approach. The work group is evaluating introducing a new resource that allows conveying "knowledge" about a variant in a patient-independent way. This would allow saying "this variant is associated with an increase risk of cardiovascular disease" rather than "based on this variant, the patient is at an increased risk of cardiovascular disease", which isn't necessarily a determination the reporting organization may wish to assert. Feedback is welcome.  ---  Comment:  This is an area we�ve discussed in the course of developing the CG v3 specs. Consider for example the information included in a paper curated in the following OMIM Entry:    �Despite the dramatic responses to EGFR inhibitors in patients with non-small cell lung cancer, most patients ultimately have a relapse. {12:Kobayashi et al. (2005)} reported a patient with EGFR-mutant, Gefitinib-responsive, advanced non-small cell lung cancer who had a relapse after 2 years of complete remission during treatment with Gefitinib. The DNA sequence of the EGFR gene in his tumor biopsy specimen at relapse revealed the presence of a second mutation ({131550.0006}). Structural modeling and biochemical studies showed that this second mutation led to the Gefitinib resistance.�    At the time when the �second mutation� was revealed, there�s a basis to assert that the first mutation led to the situation of the patient being Gefitinib-responsive. At this point in time, what should be the nature of the association of the first versus the second mutation to the responsiveness to Gefitinib? I would argue the first association is sort of retrospective and can be asserted with more certainty to that specific patient, while the second association could be seen as linking to knowledge.    These issues should be discussed in a broader sense, that is, broader than genetic testing scenarios, rather - the future state of a longitudinal health record of the patient where we expect the �dots to be connected� as much as possible, surfacing up insights from a broader perspective along the various dimensions of time, content and types of data.  ---  Summary:  knowledge representation |

TOPIC 4: What was looked at?

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| **Tracker** | 15889 |
| **Summary** | [Properties needed for range examined and human reference sequence assembly](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15889&start=0) |
| **Links** | <http://build.fhir.org/ig/HL7/genomics-reporting/> |
| **Resolution**  **Notes** | Add component 51959-5 Range(s) of DNA sequence examined to the Computable Genetic Finding profile. This would allow us, for instance, to specify the exact location for a haplotype or genotype call." |
| **Details** | Lloyd: In v2 we had 2 properties that appeared on both simple and complex variants that aren't exposed in the new model: Range examined: 51959-5 & Human reference sequence assembly version: 62374-4. Are they needed? If so, where should they go? |
| **Follow-ups** | Submitted on Sat12 May 2018 17:35:55 -0500 by FHIR Bot  Vote: #158 - A-Q  Submitted by: KEVIN POWER (CERNER)  Submitted on Wed01 Aug 2018 15:28:10 -0500 by Bob Dolin  From <https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16261>, suggest that we Add component 51959-5 Range(s) of DNA sequence examined to the Computable Genetic Finding profile. This would allow us, for instance, to specify the exact location for a haplotype or genotype call."  Submitted on Wed01 Aug 2018 16:39:19 -0500 by Kevin Power  Related to the following tracker:  <https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16763> |
| **Disposition** | Persuasive with above mod |

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| **Tracker** | 16258 |
| **Summary** | [Need to address "what was looked at"](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16258&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | *See 15889 will be withdrawn resolved with it* |
| **Details** | Need to address "what was looked at" before finalizing the IG. |
| **Follow-ups** | Submitted on Sat12 May 2018 17:25:58 -0500 by FHIR Bot  Vote: #109 - **NEG**  Submitted by: Bob Dolin (Elimu Informatics) |
| **Disposition** | *Persuasive* |

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| **Tracker** | 16763 **(Negative)** |
| **Summary** | [Comment to disagree that all things examined within range have to be represented in each variant - 2018-May Genomics #24](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16763&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | *See 15889* |
| **Details** | Submitted by: Clement McDonald (National Library of Medicine)  Comment:  Disagree with the idea that all of the things looked for have to be represented in each variant. First, there is a way to enumerate each variant and as present or absent in V2don't know if is carried into FHIR. Regardless, the negatives do not usually apply to a named variant. Those are the things found and by implication positive. The need to know what was looked for only applies to what was not found. Doesn't help to apply that to all of the things that were found (in my opinion). It is a container thing (the whole report). Further, the test name is sometimes all you will get (e.g Exon 5 gene X). The details about actual ranges looked at are often proprietary. |
| **Follow-ups** | Submitted on Tue05 Jun 2018 14:12:56 -0500 by Lloyd McKenzie  Consider capturing "We looked at X, found nothing"We looked at Y, found something - supported by Y.1, Y.2, Y.3 variants  Submitted on Mon, 30 Jul 2018 12:58:46 -0500 by Liz Amos, From Clem: Am responding to the box in middle of figure that raises a future issue of how to specify the region that was really examined. The statement in that box just raised an issue. It did not take a specific position. I don't think there is a good or practical way to say something about all possible negatives except by saying in general what set of variants were tested by probes or what range of DNA was examined as an overall part of the report--the way it is done in all conventional reports. (There is one way, however, to assert something about each probe tested for, but won't go into it here). |
| **Disposition** | *Persuasive with mod?* |

**Discussion**

Standing call currently dealing with some negative information: (next is Wednesday Aug 15th 4pm ET) -- Patient Care WG -- Negation Requirements

Patrick:

Would prefer to have methods to prefer negatives as well as positives, but indication of what/where was looked at

Bob D

Defining the panel will provide a BAM file which should have information of which regions were studied and what was callable, but they are very large files. Overlaps with quality issues

Bob M

What was targeted/what are we looking for is in the panel, is one aspect here,

Evidence that we got what we got is more about the interrogated region, may differ

Gil

Should we post a question out to the patient group for this or is there a good liaison?

Bob D

Will provide an example of the BAM/BED files, but will still want to compare it with sequence.quality

Gil

Good candidate for further discussion offline and have it brought up again either next week or after

Bob D

Bob F is also working on related information modelling issues

Bob F

Rather than waiting on IM it may be best we deal with the trackers here internally and consider it again at the september WGM

Bob D

We could allow the sequence resource to also denote ordinally which regions are of sufficient quality.

**Next Steps**

**Proposal:**

Add component 51959-5 Range(s) of DNA sequence examined to the Computable Genetic Finding profile.

Cardinality of this component should be 0..\*, to address 16763 and cover the disconnected cases

This covers 15889, 16258 and 16763 all as persuasive with mod, but the mod needs to be finalized, either something along these lines, or an addition to sequence.quality, or both

Chat History

* + Andrea Pitkus 11:06AM concur isolate is most common bacteria specimen for analysis
  + Bret Heale 11:10AM I thought we worked with the Sequence ontology terms to provide these definitions?
  + Bret Heale 11:15A wonder if the modeling subgroup should be tasked with the definitions
  + Joel Schneider 11:16AM but it's a FHIR implementation guide
  + Patrick 11:26AM what about filtering the tracker items: any opened; neg-major; clinical genomics WG
  + Bret Heale 11:31AM Thanks! glad you found it.
  + Bret Heale 1:43AM agreed
  + Bret Heale 11:53AM agree with Bob M that we can address the concern with cardinality
  + Bret Heale 11:54AM but with the presence of the component with a 0...1 cardinality it makes it easier for a Hospital system to request it in the future
  + Patrick 11:55AM we need \* as the range could be discontinue

# FHIR Subgroup Meeting August 6, 2018

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## Sign In: (presiding co-chair - Kevin Power)

1. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
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4. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
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8. Dora Finkeisen - Molit Institute - [dora.finkeisen@molit.eu](mailto:dora.finkeisen@molit.eu)
9. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
10. Ling teng- BCH - [tenglingling@gmail.com](mailto:tenglingling@gmail.com)
11. Deepak Sharma - Mayo Clinic - [sharma.deepak2@mayo.edu](mailto:sharma.deepak2@mayo.edu)
12. Insung Na - BCH - [Insung.Na@childrens.harvard.edu](mailto:Insung.Na@childrens.harvard.edu)
13. Ning Xie - BCH - [ningxie2018@gmail.com](mailto:ningxie2018@gmail.com)
14. Andrea Pitkus - [apitkus@gmail.com](mailto:apitkus@gmail.com)

## Agenda:

1. Trackers in Group G
   1. Variant Grouping - Naming [proposal](https://docs.google.com/document/d/1nwFNqopik2qGruRc3DihWk2FKHQi5UNDs2j6NiwpIQw/edit) - comments/refinements made to proposal
2. Described Variant profile - done! (withdrew)
3. [Secondary Findings](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15823&start=0) [proposal](https://docs.google.com/document/d/10BdwflZG2AXEj0WcMVIXQ-aIK-R4H7JaTLPQlQn98is/edit#) - briefly discussed

## Discussion:

Topic 1:

[proposal for changes to better support "Variant Grouping"](https://docs.google.com/document/d/1nwFNqopik2qGruRc3DihWk2FKHQi5UNDs2j6NiwpIQw/edit) - Kevin

* 1. Current Sequence Configuration is very awkward to declare more than 2 items cis
     1. HLA will typically have 4 sequences with varying informations of cis e.g,Change cardinality to 0..\*, add caution re: indicating 3+ as trans
     2. Make Sequence a valid target
        1. Lloyd: Need to comment that we are not married to Sequence, we may later decide to use a different resource/profile
     3. Remove Haplotype profile as valid target
        1. Need to determine if there is practical need to declare cis haplotypes
  2. Change sequence configuration method cardinality to 0..\*
     1. Lloyd: since observation.method has cardinality 0..1, any profile over observation that uses method can not extend the cardinality, will have to leave at 0..1
     2. Clem: an answer that represents using multiple methods would be ideal, will have to consider in the future
  3. Remove allelic phase component and allelic phase basis component from describedVariant
  4. Rename sequence configuration to sequence phase relation

Patrick- the term "sequence configuration" seems very odd to me. Maybe this is a being non native speaker or not being a genomics specialist problem… perhaps “***sequence phase relation***”?

Kevin will reconcile this comments into the proposal

Alternative later approach, if this proposal does not go well in practice: --Allow non-variant sequence segments to be declared cis by the Haplotype tool, similar to VMC definitions

Topic 2: core extension - secondary findings

Reasonable for a concept to encapsulate the need to report findings that were not specifically asked for clinically, with particular application to WGS, but does it merit a full extension to the core or is it CG-specific?

Andrea: there is precedent for this following a general “do no harm” policy, and not just in the USA

Chat:

Bret Heale 11:52AM

* composing Sequences

Bret Heale 11:57AM

* right, fed regulations would be non-binding outside us

Bret Heale 11:58AM

* wild west, eh?

Bret Heale 12:02PM

* The conceptual construct of incidental and secondary findings are not us specific but that there is a specific FED regulation to refer to, would be

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| --- | --- |
| **Tracker** | 16512 |
| **Summary** | [Sequence Configuration cardinality](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16512&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/2018May/obs-sequence-config.html&amp;quot;&amp;gt;http://hl7.org/fhir/uv/genomics-reporting/2018May/obs-sequence-config.html&amp;lt;/a&amp;gt;> |
| **Resolution**  **Notes** | **See proposal - broaden sequence configuration to focus 0-\* sequences, absorb allelic phase and allelic phase basis from describedVariant** |
| **Details** | Sequence Configuration has a obs-focus with a cardinality of 2..2. I assume this for the case when trans is value. But if the value is cisthen the cardinality could be 2..\*  Not sure how, but It would be nice to be able to do this. Then I could effectively have a set of sequences in a phase-set.  Practically speaking, I think most labs report if they have evidence of sequences being cis, but not for trans. Evidence for trans is usually inferred from lack of evidence them being in cis. |
| **Follow-ups** | Submitted on Sat, 12 May 2018 17:22:31 -0500 by FHIR Bot  Vote: #93 - A-S  Submitted by: Bob Milius (National Marrow Donor Program) |
| **Disposition** | *Persuasive with mod* |

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| **Tracker** | 16496 |
| **Summary** | [phase set of sequences (not variants)](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16496&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/2018May/index.html> |
| **Resolution**  **Notes** | **See proposal - broaden sequence configuration to focus 0-\* sequences, absorb allelic phase and allelic phase basis from describedVariant** |
| **Details** | http://www.hl7.org/fhir/2018May/extension-observation-geneticsphaseset.html easily describes how a set of sequences (not necessarily variants) can be grouped according to being in chromosomal phase with one another (cison the same molecule). This is useful for my use case.  I don't see how this can be done in the current IG.  allele-phase in described variant doesn't do it as far as I can tell. If it canI need to see an example.  Calling the phase-set a haplotype of sequences is technically correct, but seems awkward, especially since our domain talks about haplotypes in a whole gene level (eg describing whether two gene level alleles are on the same molecule. Is it possible to describe haplotypes of haplotypes?  In the end, I need to see examples of this. |
| **Follow-ups** | Submitted on Sat12 May 2018 17:21:30 -0500 by FHIR Bot  Vote: #87 - NEG  Submitted by: Bob Milius (National Marrow Donor Program) |
| **Disposition** | *Persuasive with mod* |

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| **Tracker** | 15885 |
| **Summary** | [Should consider how the PhaseSet match to the IG strucuture](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15885&start=0) |
| **Links** | http://build.fhir.org/ig/HL7/genomics-reporting |
| **Resolution**  **Notes** | **See proposal - broaden sequence configuration to focus 0-\* sequences, absorb allelic phase and allelic phase basis from describedVariant** |
| **Details** | The elements in the Observation-geneticsPhaseSet are different from the Allele Phase information the IG currently have. Need to think about if it should be a part of elements in Haplotype (it seems to be similar with Haplotype feature). May need a clear documentation about how to use the phaseSet element and the LOINC Allele Phase in the IG.  Fan:  Move the Phaseset to HaploType. And is it suitable for deleting PhaseSet IDwhich is no mapping to coding system (LOINC) and unuseful |
| **Follow-ups** | Submitted on Sat12 May 2018 17:35:44 -0500 by FHIR Bot  Vote: #157 - A-S  Submitted by: KEVIN POWER (CERNER) |
| **Disposition** | *Persuasive with mod* |

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| **Tracker** | 16173 |
| **Summary** | [Clarify usage of Genotype/Haplotype/SequenceConfiguration or remove for now](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16173&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/general.html> |
| **Resolution**  **Notes** | **N/A** |
| **Details** | The Genotype/Haplotype/Sequence Configuration profiles involve groupings for various purposes. I do not believe our documentation for these is clear enough to ensure consistent usage. As an example - Sequence Configuration has basically no documentation in the IG.  While these concepts are important, I am concerned that we do not have enough consensus to represent in our first draft of this IG. We need to either remove them for now or spend time creating additional documentation in order to be very clear how each should be used. As a starting point, does everyone feel that the usage of Genotype/Haplotype in the PGx example is correct? <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html#examples>  It is also used in HLA examples.<http://hl7.org/fhir/uv/genomics-reporting/transplants.html>  I am concerned that Genotype/Haplotype are not being used consistently even in our own initial examples. |
| **Follow-ups** | Submitted on Sat12 May 2018 17:32:46 -0500 by FHIR Bot  Vote: #141 - NEG  Submitted by: KEVIN POWER (CERNER) |
| **Disposition** |  |

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| **Tracker** | 16105 - (retracted by kevin) |
| **Summary** | [Need to include discussion of relationship to VMC](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16105&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/index.html> |
| **Resolution**  **Notes** | **N/A** |
| **Details** | The IG should include a section on how it relates to the VMC. Some initial thoughts:  \* Terminology that maps and that doesn't map directly.  \* How to utilize VMC id's  \* Can we support a VMC bundle in some way? Not sure.    -- Wanted to note that I repurposed this Tracker. My original issue is NOT an issue. |
| **Follow-ups** | Submitted on Tue01 May 2018 08:32:36 -0500 by Kevin Power  cerkyp - My apologiesI found this:  http://hl7.org/fhir/uv/genomics-reporting/conversion.html    Submitted on Sat12 May 2018 17:31:21 -0500 by FHIR Bot  Vote: #132 - A-S  Submitted by: KEVIN POWER (CERNER)  Submitted on Sun13 May 2018 16:40:03 -0500 by Gil Alterovitz  Same with other common ontologies like GO/SO/SNOMEDetc. |

**Topic 3**

[Secondary Findings](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15823&start=0) [proposal](https://docs.google.com/document/d/10BdwflZG2AXEj0WcMVIXQ-aIK-R4H7JaTLPQlQn98is/edit#)-Kevin

Chat records:

* + Bret Heale 11:24 AM not meaning to be rude, but a transcript is not the same as cDNA
  + Bret Heale 11:24 AM a transcript is composed of RNA
  + Bret Heale 11:25AM yeah, its reported as DNA
  + Bret Heale 11:39AM if you go general it loses utility

Bret Heale 11:40 AM also, if multiple which has more weight?

* + Bret Heale 11:40AM to bad LOINC doesn't have a syntax for combining answer codes....
  + Andrea Pitkus 11:45AM each observation obtained should list the method by which it was obtained. so you wouldn't want to combine as you wouldn't know which result is obtained by which method, unless the proposal Llloyd mentioned with a combined method produces a single result
  + Bret Heale 11:50AM direct observation is a very clear option
  + Patrick 11:50AM the term "sequence configuration" seems very odd to me. Maybe this is a being non native speaker or not being a genomics specialist problem
  + Bret Heale 11:52AM composing Sequences
  + Bret Heale 11:57AM right, fed regulations would be non-binding outside us
  + Bret Heale 11:58AM wild west, eh?
  + Bret Heale 12:02PM The conceptual construct of incidental and secondary findings are not us specific but that there is a specific FED regulation to refer to, would

# FHIR Subgroup Meeting July 30, 2018

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## Sign In: (presiding co-chair - Gil Alterovitz)

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4. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
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6. Insung Na - BCH - [Insung.Na@childrens.harvard.edu](mailto:Insung.Na@childrens.harvard.edu)
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9. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
10. Dorina Bratfalean- CDISC- [dbratfalean.externa@cdisc.org](mailto:dbratfalean.externa@cdisc.org)
11. Jamie Parker - Carradora Health - [jamie.parker@carradora.com](mailto:jamie.parker@carradora.com)
12. Deepak Sharma - Mayo Clinic - [sharma.deepak2@mayo.edu](mailto:sharma.deepak2@mayo.edu)
13. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
14. Amnon Ptashek - [genptashek@gmail.com](mailto:genptashek@gmail.com)
15. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
16. Ning Xie - BCH - ningxie2018@gmail.com

## Agenda:

1. [Sequence Resource Trackers](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemBrowse&tracker_id=677&querynav=%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemBrowse%26tracker_id%3D677%26forget_query%3D1&quickquery=1&tracker_item_id=&summary=&submitted_by=&priority=&assigned_to=&extra_field%5B4214%5D=&extra_field%5B4215%5D=&extra_field%5B4060%5D=&extra_field%5B3631%5D=&extra_field%5B3807%5D=&extra_field%5B3808%5D=&extra_field%5B3628%5D=13106&extra_field%5B3626%5D=&extra_field%5B4065%5D=&extra_field%5B4092%5D=&extra_field%5B4063%5D=18940&extra_field%5B4062%5D=&extra_field%5B2415%5D=&extra_field%5B4252%5D=&extra_field%5B3633%5D=&extra_field%5B3969%5D=&extra_field%5B4069%5D=&extra_field%5B4066%5D=&extra_field%5B4071%5D=&extra_field%5B3632%5D=&sortcol=tracker_item_id&sortord=ASC)
2. Described Variant profile (didn’t cover)
3. (Variant Grouping - Naming)
4. ([Secondary Findings](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15823&start=0) [proposal](https://docs.google.com/document/d/10BdwflZG2AXEj0WcMVIXQ-aIK-R4H7JaTLPQlQn98is/edit#))

## Discussion:

**Topic 1:**

|  |  |
| --- | --- |
| **Tracker** | 16501 - Need resolution by **Aug 5** (see also 16503) |
| **Summary** | [Sequence as definitional?](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16501&start=0) |
| **Links** | <http://hl7.org/fhir/2018May/sequence.html> |
| **Resolution**  **Notes** | **Proposal:**  **Leave current description on the Sequence resource to pose the question and defer discussion until future ballot.**  **Also - We should highlight the fact that Sequence currently supports sharing information that duplicates what's in the Observation-based profile and note an intention to constrain things in the future to ensure that there's only one way for that data to be shared.**  <http://build.fhir.org/sequence.html> |
| **Details** | There's a question of whether Sequence should be a definitional resource. I understand this to mean that Sequence would refer to a nucleotide or protein sequence, but not necessarily be tied to a particular Specimen or Patient. This is attractive in that it could be reused, and it's reuse could be a subject of analysis (eg., how many Patients have Observations that point to a particular Sequence. While attractive, I'm having a hard time figuring out how to implement such a system. How do go about finding out if a Sequence already exists in the FHIR server and whether I should I reuse it? What if the sequence I observed it a sub-sequence of an existing Sequence resource? or if it is an extension of it by only a couple of nucleotides? Seems like it would be better if this was left to the research systems hosting this information. If we are going to change Sequence to be definitional, then we also need to develop guidance and example workflows in its use. Or perhaps have both SequenceDefinition which may be reused in Sequence instances for observed sequences? |
| **Follow-ups** | Submitted on Sat, 12 May 2018 16:56:40 -0500 by FHIR Bot  Vote: #62 - A-C Submitted by: Bob Milius (National Marrow Donor) |
| **Disposition** | **Persuasive with mod - rework the comment on sequence resource (remove “definitional”)** |
|  |  |
| **Tracker** | **16252** |
| **Summary** | **Disagree with converting Sequence to a definitional-only Resource...** |
| **Disposition** | **Persuasive** |

|  |  |
| --- | --- |
| **Tracker** | 16267 (see also 16506) |
| **Summary** | [Opposed to referencing a new Definitional Sequence profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16267&start=0) |
| **Links** | <http://hl7.org/fhir/uv/genomics-reporting/sequence.html> |
| **Resolution**  **Notes** | **Proposal:**  **Remove Definitional Sequence profile from IG until more detailed design considerations can be documented.**  [**http://build.fhir.org/ig/HL7/genomics-reporting/sequence.html**](http://build.fhir.org/ig/HL7/genomics-reporting/sequence.html) |
| **Details** | We've yet to discuss treating Sequence as a definitional-only resource, and it will require moving some attributes that may otherwise be necessary. So for now, we need to continue to reference the STU4 Sequence resource. |
| **Follow-ups** | Submitted on Sat, 12 May 2018 17:27:50 -0500 by FHIR Bot  Vote: #118 - NEG  Submitted by: Bob Dolin (Elimu Informatics)  Submitted on Wed, 06 Jun 2018 10:57:24 -0500 by Kevin Power  Discussed during June 5 call. No conclusions, but Bret Heale will spend some time doing a mapping exercise to evaluate what we would need to change to make it definitional. |
| **Disposition** | ***Persuasive with mod -* replace with link to the sequence resource** |

**Next steps:**

1. Re-work the comment on the Resource Sequence to better outline the difference between Sequence and Observation (Bob D proposing)
2. R4 FHIR core needs to be reconciled Aug 5, sequence resource (tracker 16501)

# 

# FHIR Subgroup Meeting July 23, 2018

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## Sign In: (presiding co-chair - Kevin Power - [kpower@cerner.com](mailto:kpower@cerner.com) )

1. James Jones - BCH - [james.jones.bch@gmail.com](mailto:james.jones.bch@gmail.com)
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13. Arthur Hermann - Kaiser Permanente - arthur.hermann@kp.org
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15. Amnon Ptashek - [genptashek@gmail.com](mailto:genptashek@gmail.com)
16. Bob Freimuth - Mayo Clinic - [freimuth.robert@mayo.edu](mailto:freimuth.robert@mayo.edu)
17. Ning Xie - BCH- ningxie2018@gmail.com

## Agenda:

1. Discussion from Orders & Observations - secondary findings
   1. [Relevant tracker](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15823&start=0) - Need a way to distinguish solicited and unsolicited observations
   2. [https://www.nature.com/articles/gim2016190](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fwww.nature.com%2Farticles%2Fgim2016190&data=02%7C01%7CKevin.Power%40cerner.com%7Cf409e36b3034433d614208d5ec1e67c7%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636674537706224170&sdata=yDy8ke2RAudCZ6impZREvt%2FGY0AJujilL2beWjbJcxE%3D&reserved=0)
      1. Recommendations for reporting of secondary findings in clinical exome and genome sequencing
   3. [https://www.ncbi.nlm.nih.gov/clinvar/docs/acmg/](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fwww.ncbi.nlm.nih.gov%2Fclinvar%2Fdocs%2Facmg%2F&data=02%7C01%7CKevin.Power%40cerner.com%7Cf409e36b3034433d614208d5ec1e67c7%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636674537706224170&sdata=7TAEMhplZzsBLTuBpooAdFG0d5lUZQSPC16KNdG4uKA%3D&reserved=0)
2. Ballot discussion - “Variant Grouping
   1. <http://build.fhir.org/ig/HL7/genomics-reporting/general.html#findings>
   2. <http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html> (see ComplexVariant)
   3. [Trackers](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemBrowse&tracker_id=677&querynav=%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemBrowse%26tracker_id%3D677%26forget_query%3D1&quickquery=1&tracker_item_id=&summary=&submitted_by=&priority=&assigned_to=&extra_field%5B4214%5D=&extra_field%5B4215%5D=&extra_field%5B4060%5D=&extra_field%5B3631%5D=&extra_field%5B3807%5D=&extra_field%5B3808%5D=&extra_field%5B3628%5D=13106&extra_field%5B3626%5D=&extra_field%5B4065%5D=&extra_field%5B4092%5D=&extra_field%5B4063%5D=15770&extra_field%5B4062%5D=&extra_field%5B2415%5D=&extra_field%5B4252%5D=&extra_field%5B3633%5D=&extra_field%5B3969%5D=&extra_field%5B4069%5D=&extra_field%5B4066%5D=&extra_field%5B4071%5D=&extra_field%5B3632%5D=&sortcol=last_modified_date&sortord=DESC) (9 in group “G”)

## Discussion:

Topic 1:

* (Eric Haas):
  + Why is it important to know if is an incidental finding or not? Does that change the outcome or interpretation? there was some leaning to supporting the extension which I proposed as a **boolean**. but others indicated a preference for a **code** which would require a volunteer to propose a code set complete with definition and code system.
* (Kevin)
  + Informed consent is necessary, and reporting of secondary findings should be optional.”[4](https://www.nature.com/articles/gim2016190#ref4)
  + So, either someone has to tell the Lab “don’t send incidental findings for this patient” and the lab knows to not send them. Or the receiving system has to receive them, know they are incidental, and not share in some cases.
  + E.g., ACMG v1/v2
  + Proposed extension “secondary finding”
* Bob F
  + It’s reasonable there may be multiple reasons, Mayo labs may have started this
  + ACMG ~52 + genes, findings from later versions of ACMG may need to be treated differently.
* Arthur
  + Especially since it’s a non-required field the option is likely useful
* Andrea
  + Often expressed in a narrative sentence
* Dorina
  + In translational research, have exploratory phase then confirmatory, markers found in confirmatory may be marked secondary
* Kevin
  + May have to consider that later, focusing on clinician use case, question is of scope, “secondary finding” (optional) vs “reason for sharing” (prefered per variant)
* Andrea
  + Should there be a specific resource for ACMG and another that is more generic?
* Lloyd
  + Are the ACMG recommendations US-specific?
  + I.e. is it meaningful/relevant in Germany/China etc? May force being more generic
* Kevin
  + ACMG is just one code proposed code system that could be used
* Andrea
  + From a clinician perspective, it is voluntary. If patient doesn’t consent it stays in the lab
* Kevin
  + There does seem to be interest in the community to categorize things in this way:
    - “Secondary finding”, with a code for explaining the nature of reporting it, particularly catering to the ACMG use case but not restricting to it
    - Let’s look for ACMG-like examples from outside the US
* Dora Finkeisen 11:40AM

not sure at the moment for Germany - more generic may be better

* Lloyd
  + Create as a code, was it included because requested/pathogenic/other reason
  + OR as a codeableconcept (e.g., ACMG)

Topic 2: [Trackers](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemBrowse&tracker_id=677&querynav=%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemBrowse%26tracker_id%3D677%26forget_query%3D1&quickquery=1&tracker_item_id=&summary=&submitted_by=&priority=&assigned_to=&extra_field%5B4214%5D=&extra_field%5B4215%5D=&extra_field%5B4060%5D=&extra_field%5B3631%5D=&extra_field%5B3807%5D=&extra_field%5B3808%5D=&extra_field%5B3628%5D=13106&extra_field%5B3626%5D=&extra_field%5B4065%5D=&extra_field%5B4092%5D=&extra_field%5B4063%5D=15770&extra_field%5B4062%5D=&extra_field%5B2415%5D=&extra_field%5B4252%5D=&extra_field%5B3633%5D=&extra_field%5B3969%5D=&extra_field%5B4069%5D=&extra_field%5B4066%5D=&extra_field%5B4071%5D=&extra_field%5B3632%5D=&sortcol=last_modified_date&sortord=DESC) (9 in group “G”)

* Are there redundancies in variant grouping that we can eliminate?
* Are there capabilities that go above and beyond what we need (at this point)?
* Are we adequately addressing key use cases?
* Are our definitions of variant/haplotype/genotype sufficient?

Previous proposed changes:

1. *Expand Sequence Configuration to focus on \* sequences when declaring a value of cis rather than always 2, Remove link to variant resource*
   1. *Need to ensure aren’t asserting 3+ sequences as trans*
2. Allow “haplotypes of haplotypes”
3. Remove extra structures (this fails Bob M’s use case)

* Bob M, Bob F and Lloyd will talk this week in person @ hackathon about the naming issues

## Next Steps:

* Kevin
  + I’ll take a shot at “secondary finding” extension based off of the discussion today and share it with the group.
  + <https://docs.google.com/document/d/10BdwflZG2AXEj0WcMVIXQ-aIK-R4H7JaTLPQlQn98is/edit#heading=h.pmlg6i9o8epo>
* Bob M, Bob F and Lloyd will talk this week in person @ hackathon about the naming issues

# FHIR Subgroup Meeting July 16, 2018

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

## Sign In: (presiding co-chair - Kevin Power - [kpower@cerner.com](mailto:kpower@cerner.com) )

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9. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
10. Jamie Parker - Carradora Health - [jamie.parker@carrador.com](mailto:jamie.parker@carrador.com)
11. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
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13. Ning Xie - BCH - [ningxie2018@gmail.com](mailto:ningxie2018@gmail.com)
14. Bob Freimuth - Mayo Clinic - [freimuth.robert@mayo.edu](mailto:freimuth.robert@mayo.edu)
15. Andrea Pitkus [Apitkus@gmail.com](mailto:Apitkus@gmail.com)

## Agenda:

1. Continue Sequence / Variant as a definitional resource, (continued from discussion on 6/26 and [7/10](http://wiki.hl7.org/index.php?title=File:HL7_CG_20180626.pdf)) <http://build.fhir.org/ig/HL7/genomics-reporting/sequence.html>
2. Ballot discussion - “Variant Grouping”

<http://build.fhir.org/ig/HL7/genomics-reporting/general.html#findings>

<http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html> (see ComplexVariant)

## Discussion:

**Topic 1: Sequence/Variant as a definitional resource** [**Thread**](http://lists.hl7.org/read/messages?id=330428) **(discussion skipped today)**

Kevin P-

We have no notion of “primary” or “canonical” or “local”. We have defined a component called “variation-code” (LOINC 81252-9), and we created “dnSNP-ID” (LOINC 81255-2) as its own due to concerns that it should not be treated the same as codes from systems like ClinVar and COSMIC since it only indicates position, not the actual change.

Do we need something different or are we OK with what we have?

*Brett Heale will spend some time doing a mapping exercise to evaluate what we would need to change to make it definitional.*

|  |  |  |  |
| --- | --- | --- | --- |
| **ID** | **Summary** | **Details** | ***Disposition*** |
| 16501 | [Sequence as definitional?](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16501&start=0)  -Bob Milius  *A-C* | There's a question of whether Sequence should be a definitional resource. I understand this to mean that Sequence would refer to a nucleotide or protein sequence, but not necessarily be tied to a particular Specimen or Patient. This is attractive in that it could be reused, and it's reuse could be a subject of analysis (eg., how many Patients have Observations that point to a particular Sequence.  While attractive, I'm having a hard time figuring out how to implement such a system. How to go about finding out if a Sequence already exists in the FHIR server and whether I should I reuse it? What if the sequence I observed is a subsequence of an existing Sequence resource? or if it is an extension of it by only a couple of nucleotides?  Seems like it would be better if this was left to the research systems hosting this information. If we are going to change Sequence to be definitional, then we also need to develop guidance and example workflows in its use. Or perhaps have both SequenceDefinition which may be reused in Sequence instances for observed sequences? |  |
| 16267 | [Opposed to referencing a new Definitional Sequence profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16267&start=0)  -Bob Dolin  *NEG* | <http://hl7.org/fhir/uv/genomics-reporting/2018May/sequence.html>  We've yet to discuss treating Sequence as a definitional-only resource, and it will require moving some attributes that may otherwise be necessary. So for now, we need to continue to reference the STU4 Sequence resource.  It will require moving some attributes that may otherwise be necessary. So for now, we need to continue to reference the STU4 Sequence resource. |  |
| 16506 | [Definitional Sequence elements](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16506&start=0)  -Bob Milius  *NEG* | shows only using genomeBuild within referenceSeq. This will also need virtually all other elements with referenceSeq, such as referenceSeqId, windowStart, windowEnd, etc  It's clear that this is just a draft and needs much more work and must be clearly marked as such. |  |

**Topic 2: “Variant Grouping”** [**thread from 6-20**](http://lists.hl7.org/read/messages?id=330180)

Bob D-

A summary of grouping capabilities:

* **Grouping describedVariants**:
  + describedVariant allelic Phase ([LOINC Answer List LL4025-4](https://r.details.loinc.org/AnswerList/LL4025-4.html))
  + describedVariant allelic State (e.g. heterozygous, homozygous)
  + SequenceConfiguration can assert Cis/Trans between 2 haplotypes, between 2 variants, between a haplotype and a variant
  + (Sequence resource) all variants within one Sequence resource can be considered cis
* **Grouping complex variants**:
  + complexVariant hasMember 0..\* describedVariant
  + complexVariant Type (Answer list: Compound heterozygous | Double heterozygous | Haplotype | Hemizygous)
  + SequenceConfiguration can assert Cis/Trans between 2 haplotypes, between 2 variants, between a haplotype and a variant
* **Grouping haplotypes**:
  + haplotype derivedFrom 0..\* variant
  + SequenceConfiguration can assert Cis/Trans between 2 haplotypes, between 2 variants, between a haplotype and a variant
* **Grouping genotypes**:
  + genotype derivedFrom 0..\* haplotype
* Are there redundancies in variant grouping that we can eliminate?
* Are there capabilities that go above and beyond what we need (at this point)?
* Are we adequately addressing key use cases?
* Are our definitions of variant/haplotype/genotype sufficient?

Kevin Power - we need clearer definitions and examples of how we expect these profiles/components to be used for modeling these concepts (especially SequenceConfiguration)

|  |  |  |  |
| --- | --- | --- | --- |
| **ID** | **Summary** | **Details** | **Disposition** |
| [16173](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16173) | Clarify usage of Genotype /Haplotype/ SequenceConfiguration or remove for now  *NEG* | The Genotype/Haplotype/Sequence Configuration profiles involve groupings for various purposes. I do not believe our documentation for these is clear enough to ensure consistent usage. As an example - Sequence Configuration has basically no documentation in the IG. While these concepts are important, I am concerned that we do not have enough consensus to represent in our first draft of this IG. We need to either remove them for now or spend time creating additional documentation in order to be very clear how each should be used. As a starting point, does everyone feel that the usage of Genotype/Haplotype in the PGx example is correct? <http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html#examples> It is also used in HLA examples. <http://hl7.org/fhir/uv/genomics-reporting/transplants.html> I am concerned that Genotype/Haplotype are not being used consistently even in our own initial examples. |  |
| [16808](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16808) | Complex variants distinguish cis from trans - 2018-May Genomics #36 | Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: [Figure 5](http://hl7.org/fhir/uv/genomics-reporting/general.html#fig5): *Cis or Trans* --- Comment: Unclear - I don't recall discussion plus the complex variants distinguish this (I think)  *Postponed from July 10* |  |
| [16789](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16789) | Discussion needed on change from display names on 84413-4 2018-May Genomics #31 | Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: [Figure 5:](http://hl7.org/fhir/uv/genomics-reporting/general.html#fig5) *Genotype* 84413-4 Proposed Wording: *Genotype Display Name* 84413-4 ---  Comment: I understand why you want to shorten, but the change could mislead. These are not solid codes for genotype or haplotype. Would like to find a way to link from the figure (or content below them) to the LOINC code, description and answer list. Have linked to the answer list in the change document but these early tables are a bit more digestible. Lets talk. --- Summary: | *need to confirm consistency* |
| [16793](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16793) | Discussion needed on change from display names on 84414-2 - 2018-May Genomics #32 | Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: [Figure 5:](http://hl7.org/fhir/uv/genomics-reporting/general.html#fig5) *Haplotype* 84414-2 Proposed Wording: *Haplotype Name* 84414-2 --- Comment: Name in V2 ---  *Kevin Power-*  *Pulled from the Block vote for July 3rd. Requested follow-up:*  *What code systems would you suggest for Genotype and Haplotype? We would need very broadly applicable systems, not just for certain genes. @Bob Dolin - Can you provide examples?* | *need to confirm consistency* |
| [16496](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16496) | phase set of sequences (not variants)  -Bob Milius  *NEG* | <http://www.hl7.org/fhir/2018May/extension-observation-geneticsphaseset.html> easily describes how a set of sequences (not necessarily variants) can be grouped according to being in chromosomal phase with one another (cis, on the same molecule). This is useful for my use case. I don't see how this can be done in the current IG. allele-phase in described variant doesn't do it as far as I can tell. If it can, I need to see an example.  Calling the phase-set a haplotype of sequences is technically correct, but seems awkward, especially since our domain talks about haplotypes in a whole gene level (eg describing whether two gene level alleles are on the same molecule. Is it possible to describe haplotypes of haplotypes? In the end, I need to see examples of this. | *use case present* |
| [16325](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16325) | "haplotype" in medical genetics  *A-C* | Input from one of our physician/geneticists, Dr. Leslie Manace. Unfortunately, I do not have a specific url/location/resource to point this comment to. I believe the WG will be able to consider this generally and apply as appropriate. Fortunately, Kevin Power was able to provide initial feedback, which I have included below. Dr Manace: Genetic Assertions - "haplotype" is essentially never relevant in medical genetics. This is part of what gives me pause about the MD representation in this group.  Kevin Power: *There are use cases in HLA (and even some in Pharmacogenomics) where haplotype is relevant. So, this is another case of “when you need haplotype, structure it like this --; but skip it if you don’t need it”;* | *use cases in HLA* |
| [16820](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16820) | More explanation needed to describe genotype definition - 2018-May Genomics #39 | Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: [Genotypes](http://hl7.org/fhir/uv/genomics-reporting/general.html#fig5) describe combinations of genetic variations that together are associated with a particular phenotype - i.e. a specific physical, behavioral or risk-associated difference associated with the organism whose specimen was tested. ---  Comment: This may not be true. I have understood that the genotype is everything you know about the individual genetics including all the normals as well as possibly multiple things that might be described as separate phenotypes. (Will need the experts to weigh in) |  |
| [15885](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=15885) | Should consider how the PhaseSet match to the IG structure  -Kevin P  *A-S* | The elements in the Observation-geneticsPhaseSet are different from the Allele Phase information the IG currently have. Need to think about if it should be a part of elements in Haplotype (it seems to be similar with Haplotype feature). May need a clear documentation about how to use the phaseSet element and the LOINC Allele Phase in the IG. | *Move the Phaseset to HaploType. And it is suitable for deleting PhaseSet ID, which is not mapping to coding system (LOINC) and unuseful -Fan* |
| [16512](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_id=677&tracker_item_id=16512) | Sequence Configuration cardinality  -Bob M  *A-S* | [Sequence Configuration](http://hl7.org/fhir/uv/genomics-reporting/2018May/obs-sequence-config-definitions.html) has a obs-focus with a cardinality of 2..2. I assume this for the case when trans is value. But if the value is cis, then the cardinality could be 2..\* Not sure how, but It would be nice to be able to do this. Then I could effectively have a set of sequences in a phase-set. Practically speaking, I think most labs report if they have evidence of sequences being cis, but not for trans. Evidence for trans is usually inferred from lack of evidence them being in cis.  Change card of sequenceConfiguration to 2..\*  Add comments suggesting trans to 2  Break linkage to variant  Could we absorb phase info here? |  |

## Notes from further Discussion:

Example Reports: <https://drive.google.com/drive/folders/18T4RS0VnrJdLS3k79skbrZ0cYyL1U53t>

Bob M's use case--need to assert multiple sequences (not necessarily variants) are in the same chromosomal phase:

1. Expand Sequence Config to focus on \* sequences when declaring a value of cis rather than always 2
2. Generalize use of Haplotype to hold this information (would want to be able to construct a whole gene level Haplotype from these more general ones)
3. Add in a separate PhaseSet profile, [like this one](http://www.hl7.org/fhir/2018May/extension-observation-geneticsphaseset.html).

Bob D

Something to factor in:

Could add in a haplotype phase / haplotype phase basis, like in describedVariant

We already have some potential inconsistencies--a haplotype can be derived from 2 variants, which could then be asserted to trans

Still want to address redundancies.

1. Could remove link from seq config to variants altogether.
2. Nuclear option, in the interest of time...

Chat records:

* + Bret Heale 11:13AM thanks. I'm interested in Clem's comment on genotype. I think some clarification on my part is needed.
  + Bret Heale 11:37AM imputation with confidence. inferred from trio
  + Andrea Pitkus 11:54AM Bob F's question has 2 aspects- 1 is the initial test results and the 2nd is the reflex with more info. For 1, results won't be reported unless final as reported results are clinically actionable. It may be that additional testing is needed before finalizaiton and that would not be sent to MD, but only available in lab (in LIS).
  + Bob 1:55AM +1 to Kevin's proposal

# 

# FHIR Subgroup Meeting July 9, 2018

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## Sign In:

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4. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
5. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
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15. James Jones - BCH- james.jones.bch@gmail.com
16. Ning Xie - BCH - ningxie2018@gmail.com

## Agenda:

1. Notice for Intent to Ballot - highest priority

**Discussion:**

1. Universal realm (UV) is defined in HL7 codes as: "Realm code for use of Universal realm or context, used in every instance." Goal may be universal, but we need to be careful before requiring anything and before declaring anything as universal.  
  
2. Free in terms of cost does not mean 'free' as in the GNU definition based on 'essential freedoms' for being considered 'free': <https://www.gnu.org/philosophy/free-sw.en.html> There are some LOINC, SNOMED, other IP license issues we should be aware of. Some coding systems are free in cost, but not free by this definition. Others are. If we choose not 'free' by either or both defn., there are a number of options to consider. But, the key is we decide what we want after we evaluate, not just because one option presented... Also, whereas most other have used existing codes used in labs widely, the definitions and terms in our genomics work were defined on our calls.  
  
3. Based on the cardinality, 'coding' field is not required by FHIR for terms that we want to refer to within CodeableConcept 'code fields. And, only 'text' field in 'code' is one that can be unique. Thus, FHIR itself does not require LOINC- FHIR lists it as example in the normative ballot. In quickly changing field of genomics, should we require LOINC (or SNOMED or any other) when quickly changing?  
  
4. Various coding systems are quickly changing in genomics space. Definitions are changing for words with same name (like allele as examples) within same coding system. Community can select ones it wants without being forced to have to use one coding system that they need to align too.  
  
5. If we want to pick out certain terms as required, we should carefully define them (and can refer to other definitions). But, requiring a specific system's coding means that we may get divergence as that coding system changes on its own- particularly if that coding system is under a different organization without transparency in how definitions are made.  
  
6. This is not about LOINC (vs SNOMED, etc.). One could say a similar thing if we \*required\* SNOMED code transmission (see http://wiki.hl7.org/index.php?title=20170926\_US\_Realm\_SC\_Call).  
There is a US Realm community that may be useful to engage as well for thoughts.  
  
7. In FHIR spec, it says: "The Coding data type is used directly when there is certainty that the value must be selected directly from one of the available codes, and the list of possible codes is agreed to by all participants. This is not usually the case in the context of FHIR - general interoperability - so Coding is mostly used in extensions, which are usually intended to be defined for a well-controlled context of use."  
  
8. There is a community out there that we could engage to learn more. From the HL7 v2 article (Nebraska) published which was sent out by another co-chair in past- it actually used Snomed. In GA4GH, I have not seen LOINC used in the meetings/calls that I have been involved with relating to genomics. What have other people seen...?  
  
I have seen sequence ontology (which had been used for definitions), GO, Snomed, etc. in genomics. Each of these, as was pointed out last week on the FHIR call- is an ontology that facilitates granularity levels and reasoning engines/semantic inference/web 3.0 via encoded relationships- a special feature of genomics which is different from many traditional labs.  
  
Here are brainstorming alternatives that may help on above issues (feel free to add):  
  
1. Keep going on current path of 'requiring.' Some users, some not active/present on calls, may not use the standard. As was mentioned on the last week's call by presenter, the international people on the calls were from LOINC countries: Germany and China. Is it coincidence that we don't get more representation from other countries like UK and others? Looking at the data, there is much genomics in other countries that is simply not represented on call. Not sure if chick-egg problem, but it remains with us to take, what are currently silent voices, into account.  
  
2. Limit scope: e.g. make one or more systems required for US realm. This is what LRI did. This addresses some issues- but need to decide if want to pick only one system. See "Additional Detailed" discussion below on other current genomics ontologies/use in US as well.  
  
3. Make it informative (this is what Argonaut profiles did in stu3).  
  
4. Define HL7-based terms/codes (as opposed to LOINC) so deal with the  
licensing/"essential freedoms" (see details below) issue that some may have with LOINC license when our terms end up being in an external system that does not support "essential freedoms."  
  
5. Change coding to optional with examples in LOINC, SO, SNOMED, etc. Secure the definitions we use for our elements (as riskassessment, other resources have done) for those we feel fit as universal. Let market decide on coding. Potentially, then realm-specific guides separately after universal is done (as others have also done in FHIR).  
  
It is our responsibility to get input from others outside of LOINC-using countries, SNOMED, SO, GO, or any specific group to ensure we have a broadly accepted spec. As an example, one input we got from such individuals (from NHS) at previous connectathons, was that they would not be satisfied with anything that \*requires\* certain systems (giving same reason that SNOMED is currently limited to US Realm in others:).  
  
As said on call today longterm of universal and stu to norm track may work- but need to consider about points on path...  
  
Additional Details:  
  
The issue this involves is coding. Right now, FHIR observation lists LOINC as example for observation.code and component code. However, the current IG specification 'requires' LOINC for both. I think there was a subtlety that was not clarified at the WGM. Specifically, while 'code’ is required, its value is CodeableConcept (see https://www.hl7.org/fhir/datatypes.html#codeableconcept).  
  
In CodeableConcept, both elements are optional and only 'text' field is a unique value in CodeableConcept.  
  
Note that 'coding' (which is the only thing involving LOINC now) is \*not\* required based on cardinality in FHIR. This is true both for Observation.component.code and Observation.component.code. The only unique item in terms of cardinality is 'text.' So, LOINC as example could be done for the spec if we chose too- it is not required. The thinking was the 'text' can be the required part and we can decide on it and its definitions (which can certainly point elsewhere).  
  
In FHIR spec, it says: "The 'Coding' data type is used directly when there is certainty that the value must be selected directly from one of the available codes, and the list of possible codes is agreed to by all participants. This is not usually the case in the context of FHIR - general interoperability - so Coding is mostly used in extensions, which are usually intended to be defined for a well-controlled context of use."  
  
There may be one or more ontologies used in the future for 'coding.' The field is not fully defined and there is there a need to narrow the standard now- FHIR seems not require it either (take a look at observation- lists LOINC as example.  
  
The next issue is that while LOINC is free in terms of cost, others like SO, GO are as well. SNOMED is free for users in subscribing countries) in terms of cost. But, 'free' also is defined by GNU as follows: "'Free software' means software that respects users' freedom and community. Roughly, it means that the users have the freedom to run, copy, distribute, study, change and improve the software. " LOINC requires a license agreement to use- which has some limitations (please check it out). Most of the GNU "essential freedoms" do not seem to be met (see freedoms 0 to 3: https://www.gnu.org/philosophy/free-sw.en.html). Note a couple are relaxed for hl7 v2 messages- but nothing is mentioned about FHIR. Others like SO, GO, etc. are free and open source (not only free in terms of cost, but without restrictive license) from what I see and are being used today in genomics community. If we are open to free, but restrictive license, the number of other ontologies currently used for genomics grows- but these have not been considered. A counterargument may be- but we can still use other codes even if we 'require’ LOINC (or anther one)? Yes, but they need to be equivalent- so effectively full genomics definitions and terms will be defined by the 'coding' element if we force people to use that from specific ontology. Plus, once we are requiring a particular coding system, then it may develop independently or based on external factors which may differ from how others (including hl7) meant to use it. This is especially true now, as many codes (e.g. LOINC) are new and some have changed definitions quickly- like since LRI guide released. Others are changing as well (based on other efforts). So, do we need to  
require it now before set?  
  
If so, it may be useful to consider the IP issues and open source to ensure community can have role in further work... It is unclear how definitions and terms are made in some systems like LOINC (or SNOMED). Looking at the sites, it seems suggestions can be made, but how they are done is unknown and licenses that are not open source have restrictions on changes/use.  
  
Finally, this may be helpful as well:  
<https://www.amia.org/news-and-publications/press-release/amia-supports-multi-agency-effort-improve-lab-data-interoperability>with transcript here: <https://www.amia.org/sites/default/files/Perspectives-on-Lab-Interoperability-Alterovitz-11082016.pdf>

# FHIR Subgroup Meeting July 2, 2018

**PLEASE JOIN FREE CONFERENCE CALL**

[**https://join.freeconferencecall.com/clingenomics**](https://join.freeconferencecall.com/clingenomics)

## Sign In:

1. Patrick Werner - Molit Institute / Heilbronn University - [patrick.werner@molit.eu](mailto:patrick.werner@molit.eu)
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17. Jungang Zou -Xiamen University- jungang.zou@gmail.com

**Agenda**:

Title “Working Together - A strategy for SNOMED CT and genomics”

Overview of the approach taken to develop SNOMED CT to better support the development of Genomics medicine both in clinical practice and within the research community. The presentation will also details some of the practical steps that SNOMED international has taken to support these developments

Ian Green -- SNOMED CT Customer Relations Lead, Europe and Clinical Engineering

SNOMED CT

Most comprehensive multilingual Clinical Terminology

Increasing tools and implementations -- snomedinaction.org/

Features:

“Concepts” for meaningful queries

“Descriptions” assist in searching concepts (multilingual content)

“Relationships” support aggregation and querying

SNOMED International (now owns SNOMED CT)

non-profit “virtual” organization based in London (founded in Denmark)

33 member countries, Still need to raise more awareness globally

Collaborations - WHO (IDC-11 MMS beta) / GA4GH / HL7 (work in progress)

Discussions

HPO (phonotype content) / OMIM / HDO (review existing diagnosis and id new)

Genomics Strategy

Providing detailed info to researchers and actionable genomics guidance into EHRs

Focusing on terminologies with clear links to clinical practice through the EHR

Preferably through existing linkages where possible

Clear use case requirements will lead to SNOMED adopting new clinical language

Methodology for this approach is still in progress

Rare disease content - difficult to add as diseases are often defined by narrative text

Genomics Pilot Sites

Addenbrooke’s UK / a couple others moving forward (use case based)

Genomics content

Phenotypic findings (HPO/OMIM)

Genomics procedures (driven by member requests and collaborating partners)

Will need to extend current concept model for procedures

Updated definitions to diagnosis types (Human Disease Ontology & collabs)

Data analytics identify use case -- challenging

Future

Forming derivative products to interface ORPHANET/HPO/OMIM/DO with the EHR

Identifying patients for clinical trials

Provision mechanisms to provide precision medicine algorithms to EHRs

Identification of more pilot sites

**Meeting recording:** [**https://fccdl.in/RqeJv0hhtG**](https://fccdl.in/RqeJv0hhtG)

**Discussion:**

(paraphrased)

Bob -- When a clinication receives a variant from lab, what do they put in the EHR under the “problem” list? Currently ClinVar points to Orphanet or OMIM, which can often lead to 0 or many SNOMED codes for results.

Ian -- Yes we will have to look at coordinating more with ClinVar in the future.

Gil -- For non-member countries, are there ideas in place currently to bring representatives in? Representatives are welcome to these calls if they are interested.

Ian -- SNOMED currently have set up Clinical Reference sites, (there is one for genomics and precision medicine too) and the sites are forums accessible from anywhere, you just need to make an account to make comments. Other official outreach programs are in early stages.

**Chat Records:**

**Andrea Pitkus -**

* + **12:00PM**
* **Thank you for your presentation. Looks like this is EHR centric and doesn't mention the laboratory information system where genomics results originate. Best practice is to codify lab results values at their point of origin in the LIS as the performing laboratory knows the most about how they perform testing. Can you clarify if the scope is only limited to genomics information in the EHR or includes its origin in the LIS? Also are data warehouses, public health, and other downstream entities in scope?**

**Julian Sass -**

* + **12:02PM**
* **die Extension aus Nebraska sieht interessant aus nach dem was ich davon gesehen habe. Die haben zb Mutationen mit relationships zu Genen, SNPs usw modelliert**

# FHIR Subgroup Meeting June 25, 2018

## Sign In:

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## Agenda:

**Summary of Larry Babb’s talk for today:**

making the case about how each “observed” variant in testing needs to be linked to a “definitional” representation of a variant in order for the curated knowledge bases of the world to be able to coordinate linkage. Also, I assume that we eventually want HL7 (FHIR) to be used by groups like NCBI, EBI, etc as a mechanism for submitting variant interpretations and clinical/research knowledge for eventual use by EHR systems. The notion of using the same resource for both an “observed” sequence versus a “definitional” variant may work but seems like a conflation that will cause issues going forward. I truly believe that national or international registries will be developed that will produce identifiers for “definitional” variants and these will allow the scale of variant sharing and reduce the complexity required by every implementer to figure out how to define and correlate all the “observational” ways to represent a variant.

Variant registries will be a thing and as such we will eventually be able to tell EHRs and Labs and innovators alike to simply share the variant identifiers and leave the complexities to them while we all focus on solving the precision medicine issues.

**Discussion:**

<http://build.fhir.org/ig/HL7/genomics-reporting/sequence.html>

Larry Babb - Sequence resource today is a special kind of observation today. In working ClinGen + GA4GH, need to attach knowledge.

ClinVar - no notion of a patient

Variant perhaps as its own, standalone resource? Ready to submit to databases like ClinVar.

Not suggesting Sequence should be removed, but that perhaps a Variant definitional resource that would be referenced by Sequence.

Observations as within the context of the patient. Variant could be defintion of the ‘Genomic Feature’ that was found.

Bob M - Current IG has a ‘draft’ of what a definitional sequence resource would look like.

Larry - Closer. Want to reference the BRCA.1234 C>A as the ‘definition’ - and perhaps need to relate to the different ways it would be represented.

Bob M - Still need to store the sequence.

Larry - VMC working on a model to represent a wider variety of variation types. Could leverage this work in HL7 in the IG.

Bob D - Perhaps we need to consider how we think of a new “definitional resource” and not worry about changing one of our current resources/profiles TO definitional.

Kevin - Seems what Larry is proposing is more of a definitional resource that more closely aligns to the DescribedVariant profile?

Larry - More like what goes into ClinVar. Lab should be able to share their own representation, but ideally could provide a single, canonical ID.

Clem - We have a place to send a ClinVar ID (or easily could). But do we have such a canonical system? The missing piece is the “on the fly ID”?

Larry - Baylor did the allele registry (<http://reg.genome.network/> ), other work in process. This does NOT support everything. And until is hosted by another entity, it might be difficult to assume we can require it.

Clem - what would the resource be?

Larry - Start with VMC and its base classes. Identifier + online registry. Annotations can be added anywhere. VMC is about JSON on the wire today, not about a registry (yet).

ContextualVariant has to be an instance with a reference to the CanonicalVariant by an ID. ClinVar aspires to be this online registry.

ClinVar submission spreadsheet is an interesting tool to evaluate and see the challenges.

Bob M - Perhaps should work out some workflow diagrams.

Patrick - Seems we have all the IDs on the DescribeVariant profile.

Bob D - Seems close, but still a difference between the instance versus the canonical reference?

Andrea - Consider the submission process in the overall lab reporting processing.

Still need to be able to send the “contextual” individual annotations and/or the “canonical” identifier.

How do we define what “canonical” online registry?

Next Steps:

* Define what the canonical system would look like?
* What is the workflow?
* How does this align with [DescribedVariant profile](http://build.fhir.org/ig/HL7/genomics-reporting/obs-described-variant.html) AND/OR Sequence Resource
  + Does something need to be a definitional?
  + *Leaning towards leaving Sequence as an ‘instance’ not definitional*
  + Larry - Send around latest VMC spec?

# 

# FHIR Subgroup Meeting June 11, 2018

1. Attendees Sign In (Co-Chair - Kevin Power)
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18. Jamie Parker - Carradora Health - [jamie.parker@Carradora.com](mailto:jamie.parker@Carradora.com)

## Agenda:

1. **Gforge ballot comments related to the differences between “interpretation” and “impact”, so questions:**
   1. **Should we have a difference, or should these concepts be combined?**
   2. **Is our set of Impact classes too complicated?**

**Discussion:**

gForge trackers related to the topic:

|  |  |  |  |
| --- | --- | --- | --- |
| **ID** | **Summary** | **Real Submitter** | **Comment** |
| 16259 | [Simplify Genetic Impact](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16259&start=0) | Bob Dolin | There are SEVEN Genetic Impact classes (inherited disease pathogenicity, high risk allele, genotype medication metabolism impact, genotype medication efficacy impact, somatic diagnostic impact, somatic prognostic impact, somatic predictive impact). Consider collapsing all these into a single Genetic Impact class. Rationale: [1] the large number of profiles adds complexity to a model where a critical focus right now needs to be on the structured representation of variants and observed sequences; [2] Representation of genetic impact is evolving, so it might be premature to try to nail down the profiles. (We could for now, as an interim, have a single Genetic Impact class, along with a value set of LOINC Genetic Impact observations and associated LOINC answer codes) |
| 16324 | [more sub-categorization for sequence variants](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16324&start=0) | Scott Robertson | Input from one of our physician/geneticists, Dr. Leslie Manace. Unfortunately, I do not have a specific url/location/resource to point this comment to. I believe the WG will be able to consider this generally and apply as appropriate. Fortunately, Kevin Power was able to provide inital feedback, which I have included below. Dr Manace: Genetic Interpretations - I would like to see more sub-categorization for sequence variants - for these (vs. deletion/duplication variants), supporting information is usually critical for their interpretation as pathogenic vs. unknown significance (there are very few single site mutations that are recurrent and so well described that they speak for themselves). There is an ACMG variant interpretation algorithm (attached, see p.10 for rules) that I'd want to see incorporated. Kevin Power: &ldquo;I would like to see more sub-categorization for sequence variants - for these (vs. deletion/duplication variants)&rdquo; We originally had more specific ones, but instead we landed on having an attribute (as one of those &ldquo;Components&rdquo; that describe the type of variation (like deletion, duplication, etc &hellip;). No one was able to articulate how different the supporting information needed to be for those different types to warrant breaking up any further than we have today. But, that doesn&rsquo;t mean we shouldn&rsquo;t. &ldquo;supporting information is usually critical for their interpretation as pathogenic vs. unknown significance (there are very few single site mutations that are recurrent and so well described that they speak for themselves)&rdquo; I think we might revisit how we structure our genetic impacts (http://hl7.org/fhir/uv/genomics-reporting/general.html#impacts ) to make sure we support ACMG recommendations. I think it might be close today, but I haven&rsquo;t compared the most recently iterations of our guide to ACMG, as towards the end we were a little more focused on Somatic. &ldquo;There is an ACMG variant interpretation algorithm (attached, see p.10 for rules) that I'd want to see incorporated&rdquo; We would not support &ldquo;executing&rdquo; the algorithm, but instead would want to make sure we have a structure in place to hold the interps coming out of an algorithm like that &ndash; so that those interps can be shared in a consistently structured fashion. |
| 16719 | [Comment on use of internal definitions, consider removing distinctions - 2018-May Genomics #13](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16719&start=0) | Clement McDonald | Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Commonly, these findings are reported with interpretations, impacts and recommendations. --- Comment: Realize that these are our internal definitions and may not be recognized by most readers. In fact, not really convinced that the three distinctions are essential. Why not just interpretations and recommendations? --- Summary: Comment on use of internal definitions, consider removing distinctions |
| 16827 | [Move content back to described individual variant content - 2018-May Genomics #41](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16827&start=0) | Clement McDonald | Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Figure 6: Genetic Interpretations --- Comment: Figure 6&#65533;describes the overall impression in coarse terms e.g. whether positive , negative (See the answer lists). Interpretations are also included per variant, including an simple interpretation (variant assessment) and a possible phenotype. This has been pulled into a more complex structure that will not be needed for the typical report. I think it should be added back to described (individual) variant content. --- Summary: Move content back to described individual variant content |
| 16910 | [phenotype ontology - 2018-May Genomics #68](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16910&start=0) | Amnon Shabo | Submitted by: Amnon Shabo (Philips) Existing Wording: &#65533;interpretation&#65533; and &#65533;impact&#65533; --- Comment: The distinction between the terms &#65533;interpretation&#65533; and &#65533;impact&#65533; is unclear in my mind. In essence, there are genomic observations made and then get analyzed. The types of analysis HL7 Clinical Genomics should be focusing on are those beyond the initial analysis (e.g., variant calling), and can be roughly called &#65533;phenotype&#65533;. The latter can be an umbrella to all kinds of terms like interpretation, assessment, significance, relevance, impact, annotation, etc. But these terms must be well defined in a mini-ontology with the understanding that they&#65533;re all the result of some downstream analysis as aforementioned. This has been described in past efforts of the CG group, e.g., in the &#65533;Clinical Genomics Statement&#65533; &#65533; a specialization of the HL7 Clinical Statement model, which was used in our v3 and CDA specs. --- Summary: phenotype ontology |
| 16913 | [Interpretation vs. impact - 2018-May Genomics #69](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16913&start=0) | Amnon Shabo | Submitted by: Amnon Shabo (Philips) Existing Wording: generally expressed based on the question asked --- Comment: Interpretation is defined as &#65533;generally expressed based on the question asked&#65533; while impact is described for example as "Patient may have increased susceptibility to heart attacks", however, the latter may well be a question asked prior to testing and if so, what&#65533;s the real difference between the two terms? --- Summary: Interpretation vs. impact |
| 16925 | [can and must or should? - 2018-May Genomics #74](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16925&start=0) | Amnon Shabo | Submitted by: Amnon Shabo (Philips) Existing Wording: Genetic Interpretations can be "derived from" Genetic Assertions. Genetic Impacts MUST be "derived from" Genetic Assertions. --- Comment: I believe that any phenotype analysis of genomics observation should be explicitly associated with the observations based on which the analysis was done, whether it&#65533;s called interpretation, impact, etc. The pragmatic level of requirement is SHOULD even though I agree with MUST but think that in reality it will be hard for implementers to actually realize this kind of associations, and thus this profile might set up a too high bar of compliance, at least for initial effrots (which seems to be always the case in genomics... :-). Note that the general approach implied from those bullet points is similar in principle to the design principle of CG v3 and CDA specs, through the Clinical Genomics Statement model. --- Summary: can and must or should? |
| 16935 | [types of overall interpretation - 2018-May Genomics #77](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=16935&start=0) | Amnon Shabo | Submitted by: Amnon Shabo (Philips) Existing Wording: 1.2.4&#65533;Genetic Interpretations / "Genetic Analysis Overall Coded Interpretation" and "Deletion Duplication Overall Interpretation" --- Comment: I think that the distinction between the two types of &#65533;overall interpretation&#65533; is rather fuzzy, at least from information modeling perspective (see the previous comment for more feeddback on the concept of &#65533;overall interpretation&#65533;). --- Summary: types of overall interpretation |

Lloyd

Interp tends to be high level - lets you say “I found some stuff”

“Overall” - observations get tied to the report

Impact means

Supported by other genetics findings

Bret

Definitions: <http://build.fhir.org/ig/HL7/genomics-reporting/general.html>

|  |  |
| --- | --- |
| **Genetic Interpretations** | These are high-level assessments of the result of the genetic testing, generally expressed based on the question asked in the initiating diagnostic service request. For example, "Were any deletions or duplications found?" |
| **Genetic Impacts** | These represent [[phenotypic assertions]] about the patient based on the genetic test results. For example, "Patient may have increased susceptibility to heart attacks" |
| **Genetic Assertions** | These are observations about the specimen's genetic characteristics. For example, a chromosomal abnormality, genotype, haplotype or variant that was detected. |

Andrea

More information about the sort of impact can be included

Lloyd

Partially influenced by V2. Interps as defined today do not have value by themselves. “Positive” doesn’t provide value w/o context.

Clem/Andrea - Test code in the observations?

Lloyd - not today. Perhaps should include the test code and same answer list?

Bret - Observation.code ?

Lloyd - Remove LOINC code we have now, and instead use LOINC code (or even free text) for the test, and keep answer list.

Clem - Not many specific LOINC codes, but maybe its OK.

Arthur - Linking to test name provides more context.

Lloyd - Impacts might have nothing to do with the test (incidental findings).

Bob D - Seems Impact is too complicated. We need to focus on structuring discrete variant/sequence data

Lloyd - Have the focus on a single resource, but multiple profiles. The number of profiles will continue to grow over time. Idea is that the profile should describe what you are expressing. The patterns are a little more obvious on the Impact side, hence the additional profiles.

Bob D - Could be a distraction? Also, Impact will change over time, so structuring the variant/sequence data could be more helpful to inform that change over time.

Lloyd - Depends on who is looking. Clinicians will look for Impact.

# Chat records:

# From Bret Heale to Everyone: (11:14 AM)

# Yes. 'positive' interpretation value needs to be connected with the specific test. But even then it will be less than useful.

# if the test is broad (i.e. multiple variants possible - such as in WES)

# From Bret Heale to Everyone: (11:25 AM)

# So. The Map field within a profile is a place where one can put the current General LOINC code as a way to identify it as a 'Genetic Interpretation." this aids interoperability. The observation.code could then hold the specific test. However, when you have Three observations with the same observation.code which gets reported (intrepretation, impact or the described variant)?

# From Andrea Pitkus, PhD(MLS)CM, CSM to Everyone: (11:28 AM)

# to address your question Bret, it needs to be unambiguous to avoid misinterpretatioon, cds issues and potential harm to the patient.

# From Bret Heale to Everyone: (11:31 AM)

# Agreed. The current structure has all three pieces broken out as seperate items. If we pull the generic code and use a specific test code - there will be now three types of observation with the same test code. They will be independent objects.

# 

# From Arthur Hermann to Everyone: (11:32 AM)

# Dr. Manace’s Input 1.2.1 Genetic Interpretations - I would like to see more sub-categorization for sequence variants - for these (vs. deletion/duplication variants), supporting information is usually critical for their interpretation as pathogenic vs. unknown significance (there are very few single site mutations that are recurrent and so well described that they speak for themselves). There is an ACMG variant interpretation algorithm (attached, see p.10 for rules) that I'd want to see incorporated

# Genetic Assertions - "haplotype" is essentially never relevant in medical genetics. This is part of what gives me pause about the MD representation in this group

# Kevin Power’s response to Dr. Manace’s input:

# “I would like to see more sub-categorization for sequence variants - for these (vs. deletion/duplication variants)”

# We originally had more specific ones, but instead we landed on having an attribute (as one of those “Components” that describe the type of variation (like d

# 

# 

# FHIR Subgroup Meeting June 4, 2018

## Attendees Sign In

1. David Poloway - BCH - [dwpoloway@gmail.com](mailto:dwpoloway@gmail.com)
2. Liz Amos - National Library of Medicine - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
3. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
4. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
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16. Ling Teng [-BCH-tenglingling@gmail.com](mailto:-BCH-tenglingling@gmail.com)
17. Apurva Dharia - [apurva.dharia@esacinc.com](mailto:apurva.dharia@esacinc.com)
18. Fan Lin - Xiamen University - [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)

## Agenda:

1. **Bioinformatics-to-FHIR converter -Bob dolin**

1. **Gforge ballot comments?**

**Discussion:**

1.Choose a consistent platform for Fhir subgroup and group meeting, zoom, free conference, or webex, gotomeeting, level 3?

Gil - problems with FCC: connectivity issues, presenters need to download .exe, random (ghost) callers

Lloyd - ghost caller problem not specific to FCC. Was concerted effort to call in to group calls no matter the call technology.

Gil - callers call in signed in as someone else so they are not anonymous.

Bob Milius- recommend freeconferencecall.com; standardized by HL7, we use it in the other wg and sub-wg meetings; growing pains have been addressed, current issues are very minor IMHO, free, can be completely administered by all co-chairs.

Deepak -free conference call, zoom, level 3(don’t need to install plugins)

Kevin will send out a survey soon

2. Bioinformatics-to-FHIR converter

* Bob D: is this a valuable effort to consider?
* Clem: you’re defining two pathways to get EHR data?
* Bob D: we ping the EHR to see what they have, but often times labs are only reporting small subset of labs. Not reporting variants of unknown significance. If we are enabling re-analysis service, may want to be able to ping GACS.
* Clem: shared database or part of hospital system?
* Bob D: hospital system under HIPAA regulations.
* Gil: are there fields to capture that aren’t captured now?
* Bob D: regions studied is important to solve. Need to understand what region was tested and what wasn’t.
* Bob Milius - we have a project “vcf to Fhir”
* Bob Dolin - there is trade-off btw different input files(sam file, vcf file) Bret Heale - everything in vcf file should be standard to Fhir
* Gil - vcf file itself has different versions
* Lloyd - HL7 has not created tools for ngs raw data converting. But it accept image file for reference.

# Chat records:

# From Bret Heale to Everyone: (11:53 AM): certainly Bob provides a good use case for use to consider in the design of the FHIR artifacts needed to support. FHIR sequence, as of DSTU 3.0, had the capcity to accomdate an entire genome

# 

# FHIR Subgroup Meeting May 7, 2018

## Attendees Sign In

1. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
2. Joel Schneider - CIBMTR / NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
3. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
4. Julian Sass - Niederrhein University - [julian.sass@hsnr.de](mailto:julian.sass@hsnr.de)
5. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
6. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
7. Shennon Lu - NLM - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
8. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
9. Bob Dolin - Elimu Informatics - [bdolin@elimu.io](mailto:bdolin@elimu.io)
10. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
11. Dorina Bratfalean -CDISC- [dbratfalean.external@cdisc.org](mailto:dbratfalean.external@cdisc.org)
12. Liz Amos - NLM - [liz.amos@nih.gov](mailto:liz.amos@nih.gov)
13. Insung Na - BCH - [Insung.Na@childrens.harvard.edu](mailto:Insung.Na@childrens.harvard.edu)
14. Jamie Parker - Carradora Health - [jamie.parker@carradorahealth.com](mailto:jamie.parker@carradorahealth.com)
15. Ling [teng-BCH-tenglingling@gmail.com](mailto:teng-BCH-tenglingling@gmail.com)
16. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)

## Agenda:

1. **Group updates?**
2. **Sequence Ontologies - Karen Eilbeck**

**Discussion:**

# Chat records:

From Bret Heale to Everyone: (11:03 AM)

sign in url: https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB\_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#

From Clem Mcdonald to Everyone: (11:10 AM)

Interesting start. Could someone send the slides to those of us on the call. (Clem)

From Gil Alterovitz to Everyone: (11:11 AM)

Yes

From Bob Milius to Everyone: (11:27 AM)

Is SO versioned? if a term changes, e.g. definition, or inheritance, can someone refer to the term at the time it was used?

Is there an API, e.g. REST service, to programmatically work with SO terms?

From Bob Milius to Everyone: (11:34 AM)

If your student is looking for a FHIR project, may consider developing a FHIR terminology server

From Bret Heale to Everyone: (11:36 AM)

this will lead you to Value Set Resource which has some neat potential for expansions based on inference

From Joel Schneider to Everyone: (11:42 AM)

http://build.fhir.org/terminology-module.html

From Bret Heale to Everyone: (11:42 AM)

too true

From Bob Milius to Everyone: (11:42 AM)

How do you handle competing definitions for terms? Like the term ‘allele’ seems to change depending on the community using it. e.g., whole genes vs short stretches of change

**From Bob Milius to Everyone: (11:56 AM)**

**Re the comment that allele is defined by population variation, there is also individual genome variation, i.e. heterozygous genotypes**

# FHIR Subgroup Meeting April 30, 2018

## Attendees Sign In

1. David Poloway - BCH - [dwpoloway@gmail.com](mailto:dwpoloway@gmail.com)
2. Ling [teng-BCH-tenglingling@gmail.com](mailto:teng-BCH-tenglingling@gmail.com)
3. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
4. Bob Milius - NMPD/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
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16. Murat Sincan - Sanford Health - [murat.sincan@sanfordhealth.org](mailto:murat.sincan@sanfordhealth.org)
17. Jungang Zou - XMU - jungang.zou@gmail.com

## Agenda:

1. Identify high priority GForge trackers and discuss

13829: [new searchable parameter: variant-start](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13829&start=0)

13830: [searchable parameters for observation](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13830&start=0)

13831: [Add Observation-genetics.Allele.Id for VMC id storage](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13831&start=0)

13834: [Add seqeunce.readdepth](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13834&start=0)

13918: [Create sequence-reads profile on sequence](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13918&start=0)

15899: [Add Description to IG of genomics reporting at sequence variant page](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=15899&start=0)

**Discussion:**

Ballot closes at end of this week

13829 - Kevin: Should we defer until final disposition of sequence resource? Possible overlap. Bob D: Can’t add everything as search parameter. Seem to remember someone making comment (maybe lloyd?) that having things coupled is hard to do as search parameter.

13830 - Clem: look up tables for the allele name exist. Kevin: described variant profile doesn’t have anything called just allele name. Gil: maybe some of these profiles didn’t get transferred over. Bob D: Defer until IG -- what are we searching for when we search allele name? IG should clear this up. Kevin: Agreed. Maybe make a GForge item for something that wasn’t mapped.

13831 - We should add in GForge item in general to ask FHIR how to make ID to be uri for coding system. What to call allele vs variant. Add text to clarify our definition.

13834 -

Investigate direct linking to differential/snapshot tables in FHIR spec.

Bob D: a recent article on representation of ancestry in genomics studies: <https://genomebiology.biomedcentral.com/articles/10.1186/s13059-018-1396-2>

# Chat records:

**From Me to Everyone: (11:01 AM)**

**tinyurl.com/fhirgenomics**

**From Bob to Everyone: (11:23 AM)**

**as an aside, a recent article on representation of ancestry in genomics studies: https://doi.org/10.1186/s13059-018-1396-2**

**From Kevin Power to Everyone: (11:31 AM)**

**From IG: http://hl7.org/fhir/uv/genomics-reporting/obs-described-variant-definitions.html#Observation.component:variation-code**

**http://build.fhir.org/datatypes.html#code**

**From Joel Schneider to Everyone: (11:37 AM)**

**For reference — URI (Universal Resource Identifier) — https://www.w3.org/wiki/URI**

**From Kevin Power to Everyone: (11:44 AM)**

**http://hl7.org/fhir/uv/genomics-reporting/obs-described-variant.html**

**From Me to Everyone: (11:01 AM)**

**tinyurl.com/fhirgenomics**

**From Bob to Everyone: (11:23 AM)**

**as an aside, a recent article on representation of ancestry in genomics studies: https://doi.org/10.1186/s13059-018-1396-2**

**From Kevin Power to Everyone: (11:31 AM)**

**From IG: http://hl7.org/fhir/uv/genomics-reporting/obs-described-variant-definitions.html#Observation.component:variation-code**

**http://build.fhir.org/datatypes.html#code**

**From Joel Schneider to Everyone: (11:37 AM)**

**For reference — URI (Universal Resource Identifier) — https://www.w3.org/wiki/URI**

**From Kevin Power to Everyone: (11:44 AM)**

**http://hl7.org/fhir/uv/genomics-reporting/obs-described-variant.html**

# FHIR Subgroup Meeting April 23, 2018

## Attendees Sign In

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6. Bob Dolin - Elimu - bdolin@elimu.io
7. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
8. Insung Na - BCH - [Insung.Na@childrens.harvard.edu](mailto:Insung.Na@childrens.harvard.edu)
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10. Joel Schneider - CIBMTR / NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
11. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
12. Ling [teng-BCH-tenglingling@gmail.com](mailto:teng-BCH-tenglingling@gmail.com)

## Agenda:

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  | **gForge Item** |  |  |  | **Xin’s recommendation** |  | **Final recommendation based on the Apr16 discussion.** |  |
| **13832** |  | **[rename Observation-genticsVariant to Observation-geneticsDNAVariantChange](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D13832%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=hYgc0UyqWH7AjMKMB0cL%2F%2Fuj4U%2BbdHp1b0IlymLAsx0%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **VariantChange will record in the Unification IG Variant profile. I think this extension will be removed eventually.** |  | **After discussion, we recommend these items as "deferred"** |  |
| **13833** |  | **[Make Observation-geneticsDNAVariantChange.Id cardinality from single to multiple](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D13833%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=oJ383mKmSsXAzTsvEVV03IK10Vt9q8%2FLDiKd%2F%2Fd2SC8%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **This is friendly used to record different variant id, such as dbSNP and ClinVar id. The Unification IG only contain information about the dbSNP recording in the Variant Profile, wonder if we should add more choice for different kind of variant change id. I think the Variant Profile of the Unification IG will be the best place to take care of this variant change id information, so I don't think we need to follow up this change** |  |  |
| **13835** |  | **[Add field Observation-geneticsDNAVariantChange.Name](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D13835%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=iJlfGegYD20qdHH%2FXEEAtv78DRmprYpSHLA7Z1Qwgp0%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **The gHGVS genetics DNA change is taking care of at the Unification IG Variant Profile, so I think we can ignore this change.** |  |  |

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **14315** |  | **[Make "Genotype" profile under Observation-genetics Profile](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14315%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w7pZD0fRLGsh0Gy9SRcUkv384rMpvg8%2F%2FlOAfSsjjwU%3D&reserved=0)** |  | **[Xin Liu](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Fxliu3%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=HNChLLjKXJCNCt5PxzO0uOj0AZF4pAOHYGHSU702USw%3D&reserved=0)** |  | **Since the Unification IG page will take care of all of the profiles definition, we may include the IG page link in the core, or in the future plan, make a guide page in the core to include definition each IG profiles and link the corresponding IG page links.** |  | **After Monday meeting discussion, we recommend to mark these items as “Applied-persuasive with mod”. Will wait for the result of FMG guidance. We will move on for these item if these profiles are in core. If these are not in core, we will decide what we should do next for these later.** |  |
| **14316** |  | **[Make "Haplotype" Profile under Observation-genetics profiles](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14316%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=oCb0PSpxAGDmcWu7jrT5ukx3c5ZFglko4R5mzZozvtU%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **Since the Unification IG page will take care of all of the profiles definition, we may include the IG page link in the core, or in the future plan, make a guide page in the core to include definition each IG profiles and link the corresponding IG page links.** |  |  |
| **14317** |  | **[Make "DiscreteVariant" profile under Observation-genetics profile](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14317%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=P6H%2F9saz1yn0vmTaDWMtyY8H0HEylo43FZJkoIJDck8%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **Since the Unification IG page will take care of all of the profiles definition, we may include the IG page link in the core, or in the future plan, make a guide page in the core to include definition each IG profiles and link the corresponding IG page links. FYI, the discrete variant is part of the Variant profile in the Unification IG.** |  |  |
| **14318** |  | **[Make "StructuralVariant" profile under Observation-genetics profile](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14318%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=ckoFGurg%2FMlSAuwv7HWO%2Bb6FZka9RyDme4eZrPeIE7c%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **This is out-date with the current unification IG page. structuralVariant is part of the variant profile of IG. I think we can make some detail implementation guild about how to make a structuralVariant for the Unification IG format.** |  |  |
| **14319** |  | **[define "VariantChange" ComponentGroup in the spec](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14319%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=hE8FzZaOnD1Vc3cMEF1xLucDkNwEGtKHgllLbxgc9UY%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contains too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |  | **We recommend to leave these component group items for future discussion.** |  |
| **14320** |  | **[define "GenomicLocation" ComponentGroup in the spec](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14320%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=olzYRQ%2BXW7pUkyEP6i7F37IV%2FJYEbwvxD6oADxmIy20%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contains too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |  |  |
| **14322** |  | **[define "AllelicState" ComponentGroup in the spec](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14322%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=NoPLe7uVe9gFKq3CUkeLiHirbS2XwWNbj%2BoZciTKELA%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contains too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |  |  |
| **14323** |  | **[define "AminoAcidChange" ComponentGroup in the spec](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fproject%2Ffhir%2Ftracker%2F%3Faction%3DTrackerItemEdit%26tracker_item_id%3D14323%26start%3D0&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=PwzdauvPzVVkVhvBqQNIsuBIQ4gLTU6uXLxri7SCi6Q%3D&reserved=0)** |  | **[ling teng](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fgforge.hl7.org%2Fgf%2Fuser%2Flingteng%2F&data=02%7C01%7CKevin.Power%40cerner.com%7C6311c1dd583f4afa415908d5a3e2cda1%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636595116883839598&sdata=w9wpxRON68kMv0c%2F3iJAgZSQ%2FjfGEM2gManMCgPnwMs%3D&reserved=0)** |  | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contain too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |  |  |

**Discussion:**

**Motion to apply these recommendations - David/ Second: Ling**

* **Abstain: Bret, Joel**
* **Nay: 0**
* **Yes: 7**

# 

# Chat records:

**From Lloyd McKenzie to Everyone: (11:30 AM)**

**http://www.hl7.org/documentcenter/public/ballots/2018MAY/Announcements/ballotcomments\_spreadsheets\_2018MAY.zip**

# FHIR Subgroup Meeting April 16, 2018

## Attendees Sign In

1. **David Poloway - BCH -** [**dwpoloway@gmail.com**](mailto:dwpoloway@gmail.com)
2. **Kevin Power - Cerner -** [**kpower@cerner.com**](mailto:kpower@cerner.com)
3. **Deepak Sharma - Mayo Clinic -** [**Sharma.deepak2@mayo.edu**](mailto:Sharma.deepak2@mayo.edu)
4. **Shennon Lu - NLM -** [**shennon.lu@nih.gov**](mailto:shennon.lu@nih.gov)
5. **Clem McDonald - NLM -** [**clemmcdonald@mail.nih.gov**](mailto:clemmcdonald@mail.nih.gov)
6. **Amnon Ptashek - Edico Genome -** [**genptashek@gmail.com**](mailto:genptashek@gmail.com)
7. **Joel Schneider - CIBMTR / NMDP -** [**jschneid@nmdp.org**](mailto:jschneid@nmdp.org)
8. **Dorina Bratfalean -CDISC-** [**dbratfalean.external@cdisc.org**](mailto:dbratfalean.external@cdisc.org)
9. **Insung Na - BCH -** [Insung.Na@childrens.harvard.edu](mailto:Insung.Na@childrens.harvard.edu)
10. Lei Liu - XMU - [liulei6696@gmail.com](mailto:liulei6696@gmail.com)
11. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
12. Ling teng-BCH-tenglingling@gmail.com

## Agenda:

**Gforge Items (Xin)**

**Deferred**

**Applied - persuasive with mod**

|  |  |  |  |
| --- | --- | --- | --- |
|  | **gForge Item** |  | **Xin’s recommendation** |
| **13832** | **[rename Observation-genticsVariant to Observation-geneticsDNAVariantChange](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13832&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **VariantChange will record in the Unification IG Variant profile. I think this extension will be removed eventually.** |
| **13833** | **[Make Observation-geneticsDNAVariantChange.Id cardinality from single to multiple](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13833&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **This is friendly used to record different variant id, such as dbSNP and ClinVar id. The Unification IG only contain information about the dbSNP recording in the Variant Profile, wonder if we should add more choice for different kind of variant change id. I think the Variant Profile of the Unification IG will be the best place to take care of this variant change id information, so I don't think we need to follow up this change** |
| **13835** | **[Add field Observation-geneticsDNAVariantChange.Name](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13835&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **The gHGVS genetics DNA change is taking care of at the Unification IG Variant Profile, so I think we can ignore this change.** |

|  |  |  |  |
| --- | --- | --- | --- |
| **14315** | **[Make "Genotype" profile under Observation-genetics Profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14315&start=0)** | **[Xin Liu](https://gforge.hl7.org/gf/user/xliu3/)** | **Since the Unification IG page will take care of all of the profiles definition, we may include the IG page link in the core, or in the future plan, make a guide page in the core to include definition each IG profiles and link the corresponding IG page links.** |
| **14316** | **[Make "Haplotype" Profile under Observation-genetics profiles](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14316&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **Since the Unification IG page will take care of all of the profiles definition, we may include the IG page link in the core, or in the future plan, make a guide page in the core to include definition each IG profiles and link the corresponding IG page links.** |
| **14317** | **[Make "DiscreteVariant" profile under Observation-genetics profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14317&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **Since the Unification IG page will take care of all of the profiles definition, we may include the IG page link in the core, or in the future plan, make a guide page in the core to include definition each IG profiles and link the corresponding IG page links. FYI, the discrete variant is part of the Variant profile in the Unification IG.** |
| **14318** | **[Make "StructuralVariant" profile under Observation-genetics profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14318&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **This is out-date with the current unification IG page. structuralVariant is part of the variant profile of IG. I think we can make some detail implementation guild about how to make a structuralVariant for the Unification IG format.** |
| **14319** | **[define "VariantChange" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14319&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contains too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |
| **14320** | **[define "GenomicLocation" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14320&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contains too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |
| **14322** | **[define "AllelicState" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14322&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contains too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |
| **14323** | **[define "AminoAcidChange" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14323&start=0)** | **[ling teng](https://gforge.hl7.org/gf/user/lingteng/)** | **I think all of the Component Groups are not showing in the current IG page. This is abstract thing we created at beginning of the IG discussion. I think it is helpful to make some definition for it when a profile of the IG contain too many components. We can have a graph or paragraph to defined each of those, and define the relationship with the IG.** |

**Discussion:**

mark the top 3(13832,13833,13835) as defered

For 14315,14316,14317,14318 wait until what will the FMG guidance be

Bob: if the profiles in core, we are ok to move on,

if not in core yet, we'll decide later.

Kevin: Since we are not going to do it anyway, mark as persuasive.

we need to add as a new tracker, need to dicuss with FMG

For 14319,14320,14321,14322 all the componentgroup one, we leave these for further discussion.

We should vote for these gforge items for next week.

**Important:**

We need more coded examples in JSON or XML formats.

# Chat records:

# FHIR Subgroup Meeting March 26, 2018

## Attendees Sign In

1. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
2. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
3. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
4. David Poloway - BCH - dwpoloway@gmail.com
5. Insung Na - BCH - [Insung.Na@childrens.harvard.edu](mailto:Insung.Na@childrens.harvard.edu)
6. Bob Dolin - Elimu - bdolin@elimu.io
7. Bret Heale - Intermountain HealthCare - [bheale@gmail.com](mailto:bheale@gmail.com)
8. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
9. Lei Liu - XMU - [liulei6696@gmail.com](mailto:liulei6696@gmail.com)
10. Andrea Pitkus- IMO- [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
11. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
12. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
13. Julian Sass - Niederrhein University - [julian.sass@hsnr.de](mailto:julian.sass@hsnr.de)
14. Joel Schneider - CIBMTR / NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
15. Ling teng [-BCH-tenglingling@gmail.com](mailto:-BCH-tenglingling@gmail.com)
16. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
17. Shennon Lu \_NLM - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
18. Clem McDOnald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)

## Agenda:

About current IG:

1. Whether to include Ancestry (as compared to FamilyMemberHistory)

2. Keep or delete these four DiagnosticReport profiles <http://build.fhir.org/ig/HL7/genomics-unified/general.html#diagnosticreport-genetic-profile>

1.6 Cedit is not a required section, wait for people’s opinions

Coming Sunday is the last day for any changes.

3. Name of IG

Current:

Artifact name: genomics-unified

Display name: Clinical Genomics – Unified Proposal

Candidates:

Artifact: genomics-reporting

Display: Clinical Genomics – Reporting

Artifact: genomics

Display: Clinical Genomics

Others: (optional issues)

4. HL7 Domain Analysis Use cases (links should be validated)

5. DAM in appendix C

**Discussion:**

Loyd mentioned Google doc IG(contained the sepc) is outdated, need to update

<https://docs.google.com/document/d/1pP-2jsGng-efg8w_my6bnB8fkG5qsQItHBmipvDs3n4/edit?ts=5aafd1ac#heading=h.vzqaj0dhac0m>

# 

# Chat records:

From Bret Heale to Everyone: (11:12 AM)

https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB\_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#

From Kevin Power to Everyone: (11:15 AM)

As a reminder for everyone, here is the tracking spreadsheet: <https://docs.google.com/spreadsheets/d/1_TiWHepZ0Cv2z6ppKGNru05sQ2pEo468cEae89xIE0g/edit#gid=1386834576>

From Bret Heale to Everyone: (11:32 AM)

I understood the observation profiles to be a higher level communication of result

the nitty-gritty sequence data is in Sequence Resource

why change?

From Bob to Everyone: (11:33 AM)

that was my understanding as well, and that at a future time, we would discuss changing that paradigm

From Andrea Pitkus to Everyone: (11:34 AM)

<https://www.accessdata.fda.gov/scripts/fdatrack/view/track_project.cfm?program=nctr&id=NCTR-DBB-PM-SEQC2-Phase-II>

is this the fda requirement being referenced? if so, is there a requirement only to be reported to FDA (similar to other regulatory quality control?) then it seems like the IG needs to support the collection within th te lab and reporting to FDA, but not clear if that would now be expected to be reported on teh clinical report with the observation?

From Andrea Pitkus to Everyone: (11:35 AM)

to lloyd's point on whether it belongs in the observation resource.

can those who are more familiar with the FDA requirements, share them? thanks.

From Gil Alterovitz to Everyone: (11:39 AM)

they are under the examples

# FHIR Subgroup Meeting March 19, 2018

## Attendees Sign In

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17. Gil Alterovitz - HMS/BCH- [gilusa@gmail.com](mailto:gilusa@gmail.com)
18. Murat Sincan - Sanford Health - [murat.sincan@sanfordhealth.org](mailto:murat.sincan@sanfordhealth.org)

## Agenda:

1. Breast Cancer IG (Bret)
2. CLinGen MVLD standard for clinical MolDx data (Subha)
3. AMP-CAP somatic reporting guidelines (Joe)

**Discussion:**

Bret - Breast Cancer IG

* Forming an IG with FHIR profiles. This group has a limited knowledge of FHIR clinical genomics work group, Gil may want to reach out to this group.
  + Richard Esmond ([richard.esmond@gmail.com](mailto:richard.esmond@gmail.com))
* Gil - benefits to having IGs. This convo highlights issues with IGs, in that other groups can make IGs and they can conflict.
* Jeremy - Once maturity level 3, content needs to be vetted by other work groups. Level 1-2 phases are not regulated like that, because people are still trying stuff (trial/error).

Subha MVLD

* Kevin - The effect may be a little different depending on biomarker class: predictive, prognostic, diagnostic
* AMP guidelines are very extensive for tiers and levels of evidence.
* Clem - Cosmic?
  + Subha - Relying on AMP guidelines for how level of evidence is determined. Goal is to provide framework for how clinical laboratories gather evidence.
* Ryan - multiple somatic interpretive sections for each allele?
  + Subha - yes there could be. You may have different allele for same variant, etc.
  + Ryan - if you can have many sections, why are pubmed ids (PMIDs) being housed in allele interpretive level?
  + Subha - link variant to specific therapies, all may have own PMIDs.
  + Ryan - you recommend we bury PMIDs?
  + Subha - yes.
* Kevin - would each variant have a single level of evidence or multiple? Does each level of evidence have multiple sublevels?
  + Subha - we’re merging sublevels to make them more consistent with AMP guidelines. But, yes you may have multiple levels of evidence.

# Chat records:

From Deepak Sharma to Everyone: (11:01 AM)

trying to connect the audio. something is off. I cannot hear

From Bret Heale to Everyone: (11:01 AM)

can you hear now?

From Deepak Sharma to Everyone: (11:04 AM)

I can hear now.

From Bret Heale to Everyone: (11:05 AM)

http://wiki.hl7.org/index.php?title=Breast\_Cancer\_Data\_FHIR\_IG\_Proposal

From Deepak Sharma to Everyone: (11:07 AM)

who is the speaker? I apologize I missed part of the begining of the meating.

From Kevin Power to Everyone: (11:09 AM)

Deepak - It is Bret Heale

From Deepak Sharma to Everyone: (11:09 AM)

Thanks.

From Bret Heale to Everyone: (11:13 AM)

Richard Esmond <richard.esmond@gmail.com>

From Deepak Sharma to Everyone: (11:14 AM)

I think he is CIMI co-chair

From Insung Na to Everyone: (11:20 AM)

Gil, here is the evacuation warning in BCH. So, we will leave.

From Kevin Power to Everyone: (11:25 AM)

I would like to say this has all been discussed via email multiple times and suggest we move

From Insung Na to Everyone: (11:34 AM)

BCH Anders building evacuation warning is resolved, so, we are back here.

From Kevin Power to Everyone: (11:54 AM)

All: The table Subha is referring too is here: https://docs.google.com/document/d/1qxEoorot5A-5-MF9qADE2C-9Rwr1QifdJSfsUQKBhGg/edit

We can continue this discussion on tomorrow’s call

and how we can take this framework into our IG

# FHIR Subgroup Meeting March 12, 2018

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13. Fan Lin- Xiamen University - [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)
14. Kenny Leung - Virginia Mason - [kennyhleung@gmail.com](mailto:kennyhleung@gmail.com)

## Agenda

**Topic 1-IG/unification**

* **Xin - PGx**
  + [**https://www.lucidchart.com/invitations/accept/b64e5c7d-1328-4453-a020-d74b85db0878**](https://www.lucidchart.com/invitations/accept/b64e5c7d-1328-4453-a020-d74b85db0878) **（Diagrams)**
  + [**http://build.fhir.org/ig/HL7/genomics-unified/diagnosticreport-pgxexample.html**](http://build.fhir.org/ig/HL7/genomics-unified/diagnosticreport-pgxexample.html) **(example)**
  + [**http://build.fhir.org/ig/HL7/genomics-unified/pharmacogenomics.html**](http://build.fhir.org/ig/HL7/genomics-unified/pharmacogenomics.html)

**Topic 2-somatic variants STU4**

* [**https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5095986/**](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5095986/)
* [**https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5728662**](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5728662)

**Links:**

**Discussion:**

**Topic 1-IG/unification**

* **Xin - PGx**
  + [**https://www.lucidchart.com/invitations/accept/b64e5c7d-1328-4453-a020-d74b85db0878**](https://www.lucidchart.com/invitations/accept/b64e5c7d-1328-4453-a020-d74b85db0878) **（Diagrams)**
  + [**http://build.fhir.org/ig/HL7/genomics-unified/diagnosticreport-pgxexample.html**](http://build.fhir.org/ig/HL7/genomics-unified/diagnosticreport-pgxexample.html) **(example)**
  + [**http://build.fhir.org/ig/HL7/genomics-unified/pharmacogenomics.html**](http://build.fhir.org/ig/HL7/genomics-unified/pharmacogenomics.html)
* **Deepak -** whats difference between seq 1 and seq 2?
  + Xin - seq 1 is sequence in same phase. Seq 2 is same sequence in different phase. Two different phase sets.
* **Jeremy -** Does anyone agree that having PGx specific genetic impact is redundant with already describing medication impact?
  + Xin - there are more specific fields
* **Bret -** when it says medication assess, was this a knowledge based response? Or was it implying that medication was given at time of testing?
  + Xin - we only care about the medication the physician will prescribe the patient. The medication is the one the provider is planning on prescribing.
* **Gil suggestions**
  + line up diagram with code
  + Example of genetic interpretation
  + Diagram with instance examples instead of text or mapping with code/diagram
  + Comment about Digitize guide

**Topic 2-somatic variants STU4**

* **Jeremy -** There is no consensus yet about which guideline to accept. Joe emailed, Kevin responded saying somatic and germline variants do not align. There are at least 3-4 if not more, various reporting guidelines regarding clinical significance. Big sequencing companies use ones that don’t necessarily align with other organizations.
* **Gil -** Joe from Epic will be in the meeting next week to address this. We can include examples recommending one or more.
* **Jeremy -** prefers clingen somatic cancer working group guideline. Consensus minimum variant data work.
* **Bret -** the message that comes to you as a physician includes a link to a vendor website which has more than the message that was sent to you. Is this still considered delivered?
* **Jeremy -** there’s a .pdf report and a portal link where it may have more info. One of the key things is the references the vendor includes to make their assertions. Physicians need links to the evidence. Won’t take vendor’s word for that, unless FDA has approved therapy based on variant, etc. One of the most important parts is the tie to the actual evidence. SyncForGenes important.
* **Jeremy -** problem I have with CAP-AMP is that it is too simple and doesn’t say whether the variant goes with sensitivity to drug or resistance to drug. If all you got was Tier I, you couldn’t get effect information.
* **Viji -** if it’s FDA approved it’s already gone through extensive testing, why is there such a huge distinction between tier 1 and 2?
* **Jeremy -** FDA has been trending to make narrower and narrower approvals. They’ve gotten quite specific and narrow.
* **Jeremy** - is it possible to say in FHIR IG that while there is no clear prefered value set, we can give options to think about?
* **Gil** - yes, anything is possible. Whichever value set we have we want to determine if we are coding it.

# Chat records:

From Bret Heale to Everyone: (11:33 AM)

https://chat.fhir.org/#narrow/stream/genomics

url to the gneomics chats

From Bob Milius to Everyone: (11:33 AM)

<https://chat.fhir.org/#narrow/stream/genomics>

From Jeremy Warner to Everyone: (12:01 PM)

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5728662/>

From Jeremy Warner to Everyone: (12:02 PM)

CHIP: clonal hematopoiesis of indeterminate potential

# FHIR Subgroup Meeting March 5, 2018

## Attendees Sign In

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## Agenda

1. **Discuss content of IG**
2. **Unification**

**Links:**

**IG Page Content Google Doc:** [**https://docs.google.com/document/d/1pP-2jsGng-efg8w\_my6bnB8fkG5qsQItHBmipvDs3n4/edit?usp=sharing**](https://docs.google.com/document/d/1pP-2jsGng-efg8w_my6bnB8fkG5qsQItHBmipvDs3n4/edit?usp=sharing)

**IG Page:**

[**http://build.fhir.org/ig/HL7/genomics-unified/index.html**](http://build.fhir.org/ig/HL7/genomics-unified/index.html)

**Discussion:**

**Topic 1: Content of the IG**

* Introduce the content transfer from the Implementation Guidance to Implementation Guide
  + A Google Document is created and contain all the current content of the IG page. Suggestion and change can made here, and we can change the content based on the suggestion.
  + <https://docs.google.com/document/d/1pP-2jsGng-efg8w_my6bnB8fkG5qsQItHBmipvDs3n4/edit?usp=sharing>
* IG new content has been added to build website. Xin also created a google document with all the new changes that everyone can access and make comments on. Once all comments/revisions are addressed, content will be added/removed from build website.
* Realm for IG
  + Universal
* For Lloyd “Guidance to Implementers” google doc: is it better to have only one individual with editing permissions (Lloyd), and all others to have the ability to just make comments?
  + Bob M - let’s go with what is easiest for Lloyd
  + Kevin - Lloyd would prob agree to make it editable for everyone.
  + Gil - I think we should make things editable for everyone.
  + Gil - We should mention that IG is separate from FHIR core spec.
  + Kevin - There still is a debate about whether core and IG should be separate. Should say this material is being “restructured” instead of “superceded”
  + Clem - I think this should be superceded
  + Bob M - what about refactoring?
  + Gil - I like refactoring too.

**Topic 2: Unification**

* Continue to talk about the profiles of the IG
* Start discussing the profiles in order from the WGM:
  + *Really High Priority (I mean it)*
    - *(Genetics) DiagnosticReport*
    - *Specimen Source*
    - *Genetic Observation Common Properties*
    - *Computable Genetic Assertion*
    - *Haplotype*
    - *Genotype*
    - *Overall Variant*
  + *Higher priority*
    - *Genetic Interpretation (subclasses of)*
    - **Genotype/Haplotype how is it different from v2/v3.** 
      * **Shennon - we would work to update v2 whenever the next round for LRI is.**
    - **DescribedVariant**
      * **LOINC codes terminology require specific start and end sequences (outer/inner-start-end), sometimes there are not specific starts and ends.**
      * **Do we make changes here or to sequence resource?**
      * **If it is an observation, it is more likely to be supported. All resources are not supported in all systems, and sequence is a resource (clem)**
      * **Xin - Reference sequence seems to be repeating. Transcript and genomic ref-seq.**
      * **Kevin - we need to agree if it is okay to have repeating items in both Sequence resource and DescribedVariant.**
      * **Clem - if you have a HGVS somewhere, you cannot not include transcript.**
      * **Kevin - the question in do we deliver it as part of sequence resource, or as describedvariant, or both?**
      * **Clem - if we make things more definitional, then it would come out of Sequence.**
      * **Joel - No hard rule.**
    - *Genetic Impact (subclasses of)*
  + *High priority*
    - *Described Variant*
    - *Sequence*
  + *Medium priority*
    - *General Recommendation*
    - *Medication Usage Implications*
  + *Low priority*
    - *Current Medication*
    - *Order for Genetic Test*
    - *Descriptive Genetic Assertion (incl subclasses)*
    - *Complex Variant*
    - *Copy Number Change*
    - *Microarray Platform*

# Chat records:

From Kevin Power to Everyone: (11:23 AM)

https://chat.fhir.org/#narrow/stream/genomics/subject/Genetic.20Impact

From Bob F to Everyone: (11:24 AM)

Can Shennon comment on the LOINC code for start/end?

From Gil Alterovitz to Everyone: (11:39 AM)

https://docs.google.com/document/d/1gMJnjv7BNtnU8GIcEu7mchygaAsJal5O1BMSY8ZOlFs

From Bob Milius to Everyone: (11:46 AM)

fyi, we are saying in the FHIR IG proposal that our IG will be Universal Realm, per Lloyd.

From Bob F to Everyone: (11:54 AM)

Sequence != variant. HGVS is used for the latter, not the former.

# 

# FHIR Subgroup Meeting Feb 26, 2018

## Attendees Sign In

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4. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
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## Agenda

* **Unification**
  + · *Haplotype*
  + · *Genotype*
  + · *Overall Variant*
* **“Consortium for Agile Genomics”**

**Links:**

Unification IG demo link:

<http://build.fhir.org/ig/HL7/genomics-unified/index.html>

“Consortium for Agile Genomics” link:

<https://www.genomeweb.com/informatics/new-consortium-seeks-move-fhir-genomics-standard-clinical-practice>

**Discussion:**

Open different organizations who want to implement FHIR genomics and certify different levels.

Clem - Is this what HL7 prematurely announced?

Gil - yes, announcement that was made after HL7 genomics meeting

Lloyd - it was premature (FHIR part) because it wasn’t pre-approved

Gil - came from the top of HL7

Lloyd/Clem - lets use call time for Unification

Lloyd - Genotype/haplo fairly simple structures. Just have a value string. First question is to confirm that Geno/haplo are named with strings, not codes. Second, any other properties we need to be able to capture?

Clem - we need to touch a little bit on what they really mean. Genotype - everything you’ve learned in a study (all info). Haplo - specific to one of the father/mother’s DNA. If we have 2 haplotypes on two different parent strings how do we deal with that?

Jeremy - genotype more in reference to genotyping chips.

Bob M - for HLA we look at genotype at the full gene level. Haplotype would be on one strand, genotype would be at both strands at a certain location.

Jeremy - Official definition is neither, and it isn’t a good definition to use for our purposes. In clinical care it’s what you know.

Gil - would be useful to look at sequence ontology.

Bob D - We really don’t have an attribute that defines the scope of a genotype. We may need a specific attribute.

Clem - I want to push not for specifying locus, just what is looked at.

Bob M - Locus defines location that you are looking at.

Gil - I included a link to sequence ontology

Clem - that doesn’t help

Bob F - the VMC spent considerable time on definitions for both terms. We should use those as a starting point.

Jeremy - the way we are modeling it. Genotype can be super small or the whole organism - doesn’t matter.

Bob m - in HLA we talk about both mom and dad’s contribution to HLA. Or we can talk about a combination, multi-locus genotypes, etc. Or we can pack them all into one string.

Clem - I don’t think we want to talk about a genotype as a piece of something. Should be everything.

Gil - people are using different definitions, we may agree on one, but some other people will still use different definitions.

Lloyd - I think the definition that is in use by VMC and that is reflected in the current spec encompasses the various details people want to use genotype for. Genotype could be everything, but it could be smaller than that.

Lloyd - It depends on what is useful. Sometimes what is useful is very small, sometimes it is very large.

Bob D - 0-2 cardinality issue. There could be more than 2.

Lloyd - Think we should display cis/trans at variant level, rather than trying to express it in relationships w/ genotype.

Lloyd - Do we want to capture relationships between 2 haplotypes and assert that they are cis/trans? Or should we do that at the variant level? We want to express relationship at the variant level because when we are composing a genotype, we will be grabbing haplotypes and also variants. Haplotypes are made up of variants.

Clem - if you don’t have a variant at all, and are just declaring haplotype 1 and 2, is that a valid genetic study?

Xin - thats not the whole report. PGx, they have a result that only contains the haplotype info (text), but the lab will also attach text file with details. The info sent to EMR will only be haplotype info, not text file from lab.

Bob F - I think the definitions of geno/haplo still hold

Lloyd - It’s more the relationships that need to be defined.

Gil - I pasted into the chat the definitions of haplotype/haplotype\_block from sequence ontology. Could be helpful.

Bob F - VMC spent a year talking about these definitions and sequence ontology (karen) helped with this. I would be hesitant changing these things.

GIl - We need to understand/focus on the relationships between these terms.

Lloyd - I don’t think we’ve heard anything that keeps the VMC def from working, besides the cardinality rule Bob mentioned. We need to understand the relationship of haplotypes. Do genotypes and/or haplotypes, ever have coded names, or do they only have string names?

Everyone - we have computable names for these things as well.

Gil - continue this conversation next week or tomorrow in CG call.

# 

# FHIR Subgroup Meeting Feb 12, 2018

## Attendees Sign In

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14. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
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## Agenda

*(Genetics) DiagnosticReport*

· *Specimen Source*

· *Genetic Observation Common Properties*

· *Computable Genetic Assertion*

· *Haplotype*

· *Genotype*

· *Overall Variant*

**Links:**

[**Comparison Between The Unification IG with the STU3 IG**](https://docs.google.com/document/d/1ji5ZIfYramt1nr5J8EwjCgYQOCRzAKyOMC3VFaBed3k/edit?usp=sharing)

**Discussion:**

ballot deadline 2/25

ballot info will be ready on 2/13 cg meeting, it will be voted on next Monday.

Bob dolin had 3 questions:

Questions for the implementation guide:

Diagonostic Report:

Question: Do we have a situation that Patient will tie to many subjects.

Answer: the report will be tied to one subject.

Grant: Family member history should correlate with risk assessment along with lab test result, prior to the genetic test.

Support info will include reference to links for example Pubmed links.

Need to support the performer(who did the lab test)

Will allow media(mri, x-rage images)

# FHIR Subgroup Meeting Feb 8, 2018

## Attendees Sign In

1. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
2. David Poloway - BCH - [dwpoloway@gmail.com](mailto:dwpoloway@gmail.com)
3. Lei Liu - XMU - [liulei6696@gmail.com](mailto:liulei6696@gmail.com)
4. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
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8. Shennon Lu - NLM - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
9. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
10. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
11. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
12. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
13. Bob Dolin - elimu - bdolin@elimu.io
14. Fan Lin- Xiamen University [-Fanatxmu@gmail.com](mailto:-Fanatxmu@gmail.com)
15. Gil Alterovitz - HMS/BCH- [gilusa@gmail.com](mailto:gilusa@gmail.com)
16. ling teng -BCH [tenglingling@gmail.com](mailto:tenglingling@gmail.com)
17. Andrew Patterson - MGHA/CSIRO - [andrew@patto.net](mailto:andrew@patto.net)
18. Joel Schneider - CIBMTR / NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)

## 

## Agenda

*(Genetics) DiagnosticReport*

· *Specimen Source*

· *Genetic Observation Common Properties*

· *Computable Genetic Assertion*

· *Haplotype*

· *Genotype*

· *Overall Variant*

**Links:**

[**Comparison Between The Unification IG with the STU3 IG**](https://docs.google.com/document/d/1ji5ZIfYramt1nr5J8EwjCgYQOCRzAKyOMC3VFaBed3k/edit?usp=sharing)

**Discussion:**

**Chat records:**

From David Kreda to Everyone: (11:34 AM)

This discussion of IG vs guidance feels “random” to me. Is the WG not aiming until define HOW we are going to distill the new genomics unification work into “standard issue” FHIR specification. Can we not describe and confirm what that is?

From Deepak Sharma to Everyone: (11:36 AM)

I belive JIRA is much better and would welcome that change as soon as we can

From David Kreda to Everyone: (11:38 AM)

Further, there is now a long list of profiles (or so it appears) that accommodate a variety of genomic/genetics use cases. A “gut check” here - we understood from the Sep 2017 discussion (Lloyd’s email) that proliferating extensions was not good. Now we have (comparatively) a proliferating list of profiles, tailored to targeted use cases. What is the feeling from the implementer community about this? Also, w.r.t. the FMM (FHIR Maturity Model), is there any sense about what will be the maturity status of each of these profiles … I assume 0 or 1

From Deepak Sharma to Everyone: (11:54 AM)

based on my limited knowledge about Atlassian product setup - Till the time we move to JIRA (which is from Atlassian), we can continue with GForge, Google Docs for keepting track, tasks, versioning and history of documents. But once JIRA is implemented, it can work beautifully with GitHub (or BitBucket) to do all these things. I have experience working with 2-3 setups like that and I can help whoever is leading this effort (permitting time and effort).

From Bret Heale to Everyone: (11:56 AM)

I'm also familiar with the Atlassian suite (JIRA/Confluence etc..). Confluence would be a surrogate for Google Docs - similar functionality

# FHIR Subgroup Meeting Jan 25, 2018

## Attendees Sign In

1. David Poloway - BCH - [dwpoloway@gmail.com](mailto:dwpoloway@gmail.com)
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4. Fan Lin XMU- [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)
5. Lei Liu - XMU - [liulei6696@gmail.com](mailto:liulei6696@gmail.com)
6. Ling [teng-BCH-tenglingling@gmail.com](mailto:teng-BCH-tenglingling@gmail.com)
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8. Lloyd McKenzie - Gevity - [lmckenzie@gevityinc.com](mailto:lmckenzie@gevityinc.com)
9. Gil Alterovitz - HMS/BCH- gilusa@gmail.com
10. Shennon Lu - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
11. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
12. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
13. Kenny Leung - ex-TSI / Virginia Mason - [kennyhleung@gmail.com](mailto:kennyhleung@gmail.com)
14. Bob Dolin - Elimu - bdolin@elimu.io
15. Grant Wood - Intermountain - [grant.wood@imail.org](mailto:grant.wood@imail.org)
16. Joel Schneider - CIBMTR / NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
17. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
18. David Kreda - HMS - [David.kreda@gmail.com](mailto:David.kreda@gmail.com)
19. Bob Freimuth - Mayo Clinic - [freimuth.robert@mayo.edu](mailto:freimuth.robert@mayo.edu)
20. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)

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## Agenda

CDS on FHIR -- Bryn Rhodes

**Links:**

**Discussion:**

**Chat records:**

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# FHIR Subgroup Meeting Jan 11, 2018

## Attendees Sign In

1. **Kevin Ehlers, BloodCenter of Wisconsin -** [**kevin.ehlers@bcw.edu**](mailto:kevin.ehlers@bcw.edu)
2. **Xin Liu - BCH -** [**xinliu215@gmail.com**](mailto:xinliu215@gmail.com)
3. **Joel Schneider - NMDP/CIBMTR -** [**jschneid@nmdp.org**](mailto:jschneid@nmdp.org)
4. **Gil Alterovitz - HMS/BCH-** [**gilusa@gmail.com**](mailto:gilusa@gmail.com)
5. **Lloyd McKenzie - Gevity -** [**lmckenzie@gevityinc.com**](mailto:lmckenzie@gevityinc.com)
6. **Joseph Kane - Epic -** [**jkane@epic.com**](mailto:jkane@epic.com)
7. **Bob Dolin - Elimu - bdolin@elimu.io**
8. **David Poloway - BCH -** [**dwpoloway@gmail.com**](mailto:dwpoloway@gmail.com)
9. **Lei - XMU -** [**liulei6696@gmail.com**](mailto:liulei6696@gmail.com)
10. **Deepak Sharma - Mayo Clinic -** [**sharma.deepak2@mayo.edu**](mailto:sharma.deepak2@mayo.edu)
11. **Bret Heale - Intermountain Healthcare -** [**bheale@gmail.com**](mailto:bheale@gmail.com)
12. **Fan Lin ,XMU-** [**Fanatxmu@gmail.com**](mailto:Fanatxmu@gmail.com) **- XMU**
13. **Amnon Ptashek - Edico Genome -** [**genptashek@gmail.com**](mailto:genptashek@gmail.com)
14. **Kevin Power - Cerner -** [**kpower@cerner.com**](mailto:kpower@cerner.com)
15. **Larry Babb - Sunquest/Partners -** [**larry.babb@sunquestinfo.com**](mailto:larry.babb@sunquestinfo.com)
16. **Dorina Bratfalean - CDISC-** [**dbratfalean.external@cdisc.org**](mailto:dbratfalean.external@cdisc.org)
17. **David Kreda - HMS -** [**david.kreda@gmail.com**](mailto:David.kreda@gmail.com)
18. **Alex Mankovich - Philips -** [**alex.mankovich@philips.com**](mailto:alex.mankovich@philips.com)
19. **Shennon Lu - NLM -** [**shennon.lu@nih.gov**](mailto:shennon.lu@nih.gov)
20. **Clem McDonald -** [**ClemMcdonald@mail.nih.gov**](mailto:ClemMcdonald@mail.nih.gov)

## Agenda

**Links:**

* <https://docs.google.com/document/d/1zPk4wCBaGoCuTS1I35mI2PYRncDtYIhVXjvwIXsimA0/edit?ts=5a56d4cf> (The concept definition of VMC and FHIR, just for discussion)
* <https://docs.google.com/document/d/1juWEnjyXV34yYmPq3FDpLAiJlM0Hiv0FyNBfvPD6enM/edit?ts=5a4e61a2#> (genetics-unified)
* <https://docs.google.com/document/d/12E8WbQlvfZWk5NrxwLytmympPby6vsv60RxCeD5wc1E/edit> (VMC spec)

**Discussion:**

1. FHIR - Unification (Definition of the each profile within the unification model)
   1. <https://docs.google.com/document/d/1juWEnjyXV34yYmPq3FDpLAiJlM0Hiv0FyNBfvPD6enM/edit?ts=5a4e61a2#>
   2. Variant
      1. Discrete Variant
      2. Structural Variant
      3. Complex Variant
      4. Cytogenetic Variant
   3. Genotype
   4. Haplotype
   5. Gene
   6. Sequence
   7. Allele
      1. <https://docs.google.com/document/d/12E8WbQlvfZWk5NrxwLytmympPby6vsv60RxCeD5wc1E/edit#heading=h.6przha5aozrj> (VMC Allele Definition)
      2. from Sequence onotology: <http://www.sequenceontology.org/miso/current_svn/term/SO:0001023>. An allele is one of a set of coexisting sequence variants of a gene. (From Bret Heale)
      3. From Joel Schneider to Everyone: (11:24 AM) Here’s another story … <https://www.ebi.ac.uk/training/online/course/human-genetic-variation-introduction/what-genetic-variation/types-genetic-variation/variants>
      4. From Larry Babb to Everyone: (11:37 AM) the VMC computational term “Allele” is not limited to be within a “Gene"
      5. From Bob Dolin to Everyone: (11:38 AM) We may want to invent a word that does mean "a unique sequence of a gene"
      6. From David Kreda to Everyone: (11:38 AM) To make progress and be parsimonious is (as Lloyd and Bob D suggest) is springboard from the VMC definition. The definition is “by definition” the computational definition we provide in VMC. Indeed, that is the ONLY VMC definition.
      7. From Kevin Power to Everyone: (11:40 AM) So, as of now, does VMC think that Allele will apply to what we could today call ‘Structural Variant’?
      8. From Joel Schneider to Everyone: (11:42 AM) VMC Allele is this (?): <https://docs.google.com/document/d/12E8WbQlvfZWk5NrxwLytmympPby6vsv60RxCeD5wc1E/edit#heading=h.6przha5aozrj>
      9. From Fan Lin to Everyone: (11:44 AM) I thought , we throw a more small object-definition, could we make more examples and map into each object—Classification. That we find out the boundary
      10. From Bob Dolin to Everyone: (11:45 AM) From VMC document: "Allele" connotes a state whereas "variant" connotes a change between states.
      11. From Larry Babb to Everyone: (11:51 AM) VMC … Computational definition: A specific combination of non-overlapping Alleles that co-occur on the same reference sequence. GENO: Haplotype (GENO:0000871) - A set of two or more sequence alterations on the same chromosomal strand that tend to be transmitted together.

**Chat records:**

From David Kreda to Everyone: (11:05 AM)

Hi I’m on mute. I am going to HIMSS FYI

From Kevin Power to Everyone: (11:06 AM)

No HIMSS for me

From Gil Alterovitz to Everyone: (11:07 AM)

anyone else thinking about himss?

From Bob Dolin to Everyone: (11:07 AM)

Yes, I'll be there

From Fan Lin to Everyone: (11:11 AM)

https://docs.google.com/document/d/12E8WbQlvfZWk5NrxwLytmympPby6vsv60RxCeD5wc1E/edit

VMC Document

From Bret Heale to Everyone: (11:13 AM)

from Sequence onotology: http://www.sequenceontology.org/miso/current\_svn/term/SO:0001023

An allele is one of a set of coexisting sequence variants of a gene.

From Xin Liu to Everyone: (11:19 AM)

https://docs.google.com/document/d/12E8WbQlvfZWk5NrxwLytmympPby6vsv60RxCeD5wc1E/edit

Vmc spec link

From Fan Lin to Everyone: (11:22 AM)

on Page 23

From Joel Schneider to Everyone: (11:24 AM)

Here’s another story …

https://www.ebi.ac.uk/training/online/course/human-genetic-variation-introduction/what-genetic-variation/types-genetic-variation/variants

From David Kreda to Everyone: (11:25 AM)

an entirely new word is also risky. VMC is focused on disambiguation through very precise definition of terms that are used in a model. The terminology of biologists may resist this “normalization” but interoperability/engineering/computational crowd needs clarity for implementation.

From David Kreda to Everyone: (11:27 AM)

We showed a connection between the biological (sloppy, if you will) phrase in the VMC spec, but it is the computational definition that drives the model. The VMC models are not yet exhaustive, and will perhaps need a few more atomic structures.

From David Kreda to Everyone: (11:28 AM)

Larry is spot on in his description of the VMC motivations and methods. Kudos!

From Larry Babb to Everyone: (11:37 AM)

the VMC computational term “Allele” is not limited to be within a “Gene"

From Bob Dolin to Everyone: (11:38 AM)

We may want to invent a word that does mean "a unique sequence of a gene"

From David Kreda to Everyone: (11:38 AM)

To make progress and be parsimonious is (as Lloyd and Bob D suggest) is springboard from the VMC definition. The definition is “by definition” the computational definition we provide in VMC. Indeed, that is the ONLY VMC definition.

From Kevin Power to Everyone: (11:40 AM)

So, as of now, does VMC think that Allele will apply to what we could today call ‘Structural Variant’?

From Joel Schneider to Everyone: (11:42 AM)

VMC Allele is this (?):

https://docs.google.com/document/d/12E8WbQlvfZWk5NrxwLytmympPby6vsv60RxCeD5wc1E/edit#heading=h.6przha5aozrj

From Kevin Power to Everyone: (11:42 AM)

and BTW - I think we need to let Lloyd finish his ‘straw man’?

@Joel - Yes, that is VMC Allele

From Larry Babb to Everyone: (11:43 AM)

Joel - Yes, that’s it.

From Fan Lin to Everyone: (11:44 AM)

I thought , we throw a more small object-definition, could we make more examples and map into each object—Classifaction. That we find out the boundry

From Bob Dolin to Everyone: (11:45 AM)

From VMC document:

From Bob Dolin to Everyone: (11:45 AM)

"Allele" connotes a state whereas "variant" connotes a change between states.

From David Kreda to Everyone: (11:46 AM)

Prediction is hard, especially about the future!

From Larry Babb to Everyone: (11:47 AM)

We hope to build the structural variant on these atomic terms.

but we haven’t discussed Haplotype and Genotype, which provides some more of the picture.

From Larry Babb to Everyone: (11:49 AM)

the VMC Genotype and Haplotype would allow HL7 to represent the more complex forms of variants.

From Joel Schneider to Everyone: (11:50 AM)

Usage of some terms such as "haplotype" may vary by context, e.g.: https://en.wikipedia.org/wiki/Human\_leukocyte\_antigen#Haplotypes

From Fan Lin to Everyone: (11:51 AM)

For example,,the polymorphism with same DNA coding that result in different AminoAcid Sequence, finally to the different protein.Because of Variable shear or methylation/acetylation

From Larry Babb to Everyone: (11:51 AM)

VMC … Computational definition: A specific combination of non-overlapping Alleles that co-occur on the same reference sequence.

GENO: Haplotype (GENO:0000871) - A set of two or more sequence alterations on the same chromosomal strand that tend to be transmitted together.

From Fan Lin to Everyone: (11:52 AM)

Right, that is constraint

From Kevin Power to Everyone: (11:54 AM)

I would like Larry to take 1 min to speculate on what Structural stuff might look like in VMC

From Xin Liu to Everyone: (11:54 AM)

Agree

From Larry Babb to Everyone: (11:56 AM)

I’d like to meet with Reece Hart and prepare something so as to not under/over represent the plan for fuzzy locations in the VMC.

From Kevin Power to Everyone: (11:57 AM)

@Larry - OK

From Larry Babb to Everyone: (11:57 AM)

We will be undertaking the broad set of “structural” variant types in the next few months and year.

From Fan Lin to Everyone: (11:57 AM)

As my opinion, we have to identifier each kinds of variant and map to definication (calssification ). So more example should be help in find out some abnormal-that some fusion or hard to calssify.

From Bret Heale to Everyone: (11:57 AM)

high school biology...hmm, maybe the high school text book definition...

From Kevin Power to Everyone: (11:57 AM)

+1 to us high school biology hacks :)

From Gil Alterovitz to Everyone: (11:58 AM)

:)

From Gil Alterovitz to Everyone: (11:58 AM)

bret- let us know. :)

From David Kreda to Everyone: (11:59 AM)

The VMC approach involves one key mental shift: definitions for modeling are trying to squeeze out ambiguous terms for implementers. This bias toward implementers is, we knew, pretty hard to swallow ... at first. But the gain is impressive if you want interoperability.

From Larry Babb to Everyone: (11:59 AM)

I Ilke Lloyd’s comments on thinking about structural to be built up into complex variants

From Kevin Power to Everyone: (12:00 PM)

I would ask that everyone please review the definitions and comments in the document being shared now

From Larry Babb to Everyone: (12:00 PM)

+1 on David K’s comments

# FHIR Subgroup Meeting Jan 4, 2018

## Attendees Sign In

1. Deepak Sharma, Mayo Clinic - [sharma.deepak2@mayo.edu](mailto:sharma.deepak2@mayo.edu)
2. Larry Babb, Sunquest - [larry.babb@sunquestinfon.com](mailto:larry.babb@sunquestinfon.com)
3. Kevin Power, Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
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5. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
6. Lloyd McKenzie, Gevity - lmckenzie@gevityinc.com
7. David Kreda, HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
8. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
9. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
10. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
11. Ling teng [-BCH-tenglingling@gmail.com](mailto:-BCH-tenglingling@gmail.com)
12. Bob Dolin - Elimu - [BDolin@elimu.io](mailto:BDolin@elimu.io)
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16. Lei Liu XMU - [liulei6696@gmail.com](mailto:liulei6696@gmail.com)
17. Fan Lin XMU- [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)

## Agenda

**Links:**

<http://guidance.site:7473/extension-sequence-readsreadgroups.html>

**Discussion:**

1. **Quality DIscussion:**

Bob Dolin: gforge item

split Sequence.referenceSeq.strand into two fields.

[https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker\_item\_id=14273&start=100https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker\_item\_id=14273&start=100](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14273&start=100)

[adoption of Sequence.referenceSeq.strand with “watson” and “crick” in place of Sequence.referenceSeq.direction and “forward” and “reverse” ... ]

Motion: Bob Dolin

Accept the proposal for gforge item

Abstention: David Kreda, Andrea Pitkus

Ney: none

**Chat records:**

11:14:11 From david : https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB\_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#

11:14:20 From Deepak Sharma : tinyurl.com/fhirgenomics

11:15:32 From Andrea Pitkus, PhD, MLS(ASCP)CM : Bob- interested in quality too. Is your question about th equality of the specimen analyzed by the lab producing genomics results? (There are CLIA requirements for labs on noting specimen quality)

11:18:27 From ling teng : https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker\_item\_id=14273&start=100

11:21:13 From Larry Babb : watson/crick seems a little informal or colloquial. I would prefer something more formal.

11:26:29 From Gil Alterovitz : http://www.sequenceontology.org/browser/obob.cgi

11:26:51 From Gil Alterovitz : http://www.sequenceontology.org/browser/

From Gil Alterovitz to Everyone: (11:57 AM)

http://guidance.site:7473/extension-sequence-readsreadgroups.html

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# FHIR Subgroup Meeting Dec 14, 2017

## Attendees Sign In

1. Bob Milius - NMDP/CIBMTR - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
2. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
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5. Xin Liu - BCH - [xiniu215@gmail.com](mailto:xiniu215@gmail.com)
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10. Fan Lin - Xiamen University- [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)
11. Lei Liu - Xiamen University - [liulei6696@gmail.com](mailto:liulei6696@gmail.com)
12. Deepak Sharma - Mayo Clinic, sharma.deepak2@mayo.edu
13. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
14. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
15. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
16. Kenny Leung - Virginia Mason (ex Translational Software) - kennyhleung@gmail.com

## Agenda

* GForge Items
  + 14315 [Make "Genotype" profile under Observation-genetics Profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14315&start=100)
  + 14316 [Make "Haplotype" Profile under Observation-genetics profiles](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14316&start=100)
  + 14317 [Make "DiscreteVariant" profile under Observation-genetics profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14317&start=100)
  + 14318 [Make "StructuralVariant" profile under Observation-genetics profile](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14318&start=100)
  + 14319 [define "VariantChange" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14319&start=100)
  + 14320 [define "GenomicLocation" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14320&start=100)
  + 14321 [define "AllelicState" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14322&start=100)
  + 14322 [define "AminoAcidChange" ComponentGroup in the spec](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14323&start=100)

**Discussion:**

1. US core implementation guides

http://www.fhir.org/guides/registry

we'll have different profiles for different regions.

build.fhir.org/diagonosticreprot.html under genetic observations

2. Bob Milius made motion to call Genomic unify

Second: Gil

Abstain: none

nay: none

yes: everybody else

3. Xin: we still need to discuss some of points we already have till now.

4.

Bob Dolin: gforge item

split Sequence.referenceSeq.strand into two fields.

https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker\_item\_id=14273&start=100

Bret shared the link for definition of sequence direction

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3055211/

Chat message:

From Bob Milius to Everyone: (11:09 AM)

implementation guides http://www.fhir.org/guides/registry

From ling teng to Everyone: (11:15 AM)

We should talk about gforge items in a few mins.

From Dorina to Everyone: (11:30 AM)

sorry I have to leave, it looks great

From Bret Heale to Everyone: (11:35 AM)

@Fan Lin +1 agree

From Bret Heale to Everyone: (11:52 AM)

sorry mic not working? watson crick is less ambigious as it is not as loaded as forward and reverse

i think

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3055211/

# FHIR Subgroup Meeting Dec 7, 2017

## Attendees Sign In

1. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
2. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
3. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
4. Bob Dolin - Elimu - bdolin@elimu.io
5. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
6. David Poloway - BCH - [dwpoloway@gmail.com](mailto:dwpoloway@gmail.com)
7. Fan Lin- Xiamen University - [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)
8. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
9. Shennon Lu - NLM - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
10. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
11. Amnon Ptashek -Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
12. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
13. Gil Alterovitz - HMS/BCH- [gilusa@gmail.com](mailto:gilusa@gmail.com)
14. Ling Teng-BCH- tenglingling@gmail.com

## Discussion

* McKenzie described the R4 timing …
  + Dec/Jan is draft for comment ballot - no need to include the CG IG here
  + Apr/May is first STU ballot - we should target having the CG IG ready here
  + Aug/Sept is second STU ballot - we can use this as a fallback if need be or if we find significant changes are needed after the first STU ballot
  + End of Dec. 2018 is target for publishing R4. Ideally we would publish at the same time, but if we do an IG, we have the ability to publish later if we wish.
* Bob Milius - new resource proposal for R4 would be Dec 11 per FMG.

This could allow for a placeholder for now, else a special dispensation would be needed after for a May ballot. However, a new resource would not qualify for any “normative” balloting so FMG would probably not withhold agreement if the proposal if fleshed out.

* May 18 2018 DAM - <http://tinyurl.com/damcgdoc> else [this](https://docs.google.com/document/d/1XzT8_pxDZk6gGJ9zlKmISSBsBJJf7UCNtd4PIa5FBFs/edit). Comments can still be made, but no changes to the core document through/until the ballot.
* Please upload the frozen core document to the CG document page on the HL7 site. (<http://www.hl7.org/Special/committees/clingenomics/docs.cfm>)

Future article link, “[FHIR Genomics: Making FHIR a Complete Data Standard for Precision Medicine](https://docs.google.com/document/d/18zXTEiPzv9hMLh-bndECq8766smCh5MJgQsq1pJ5C5o/edit)” for comments, input.

Overall quality and positional quality use by Oncologists - invite Jeremy Warner - he mentioned a use case that might be served by the proposed Sequence.readgroup extension.

# 

# FHIR Subgroup Meeting Nov 9, 2017

## Attendees Sign In

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3. Dorina Bratfalean CDISC [dbratfalean.external@cdisc.org](mailto:dbtarfalean.external@cdisc.org)
4. Bob Milius - NMDP/CIBMTR - bmilius@
5. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
6. Bob Dolin - Elimu - BDolin@Elimu.io
7. David Poloway - BCH - [dFanwpoloway@gmail.com](mailto:dwpoloway@gmail.com)
8. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
9. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
10. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
11. Fan Lin - Xiamen University - [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)
12. Ling teng - BCH [-tenglingling@gmail.com](mailto:-tenglingling@gmail.com)
13. Shennon Lu - NLM - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
14. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
15. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
16. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
17. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)

**Agenda:**

Nov 9 Xin/Fan present approach for R4

Presiding co-chair: Gil Alterovitz

Notes:

Xin present:

The DiagnosticReport Overview with Observations

https://www.processon.com/view/link/59ecf4a5e4b08b9e917f8d85

The PGx example Diagram

https://www.processon.com/view/link/59ecf01be4b07162476d1fc2

Lin Fan present:

the difference between Harvard1 and Genomics2

benefits of Harvard1

http://harvardone.shareted.com/ Staging Stie

staging site

http://harvardone.shareted.com/

Implication Guide

https://docs.google.com/document/d/1j\_-ewytEV31ACGJqZ\_V0qo8K7M6XV6DCQ-ZsJam3zbw/edit

http://harvardone.shareted.com/observation-genetic.html

http://harvardone.shareted.com/amino.html

Gil introduced Xin's backgrounds

discrete variant is single variant

structure variant,

Lloyd comments,

all the attributes should be together, they are not independently,

discussion about phaseset.

is it really useful by itself in clinical

it is useful by itself.

two different proposals(harvardone and genomic2) with same direction.

# FHIR Subgroup Meeting Oct 26, 2017

## Attendees Sign In

1. Kevin Power - [kpower@cerner.com](mailto:kpower@cerner.com) (Presiding Co-chair)
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12. Bob Dolin - Elimu - bdolin@elimu.io
13. Dorina Bratfalean- [dbratfalean.external@cdisc.com](mailto:dbratfalean.external@cdisc.com)
14. Grant Wood - Intermountain Healthcare - grant.wood@imail.org

## 

## Agenda - Open Tracker Items for Sequence resource

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **ID** | **Summary** | **Priority** | **Assignee** | **Submitted By** |
| **13829** | [new searchable parameter: variant-start](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13829&start=0) | Medium | [ling teng](https://gforge.hl7.org/gf/user/lingteng/) | [ling teng](https://gforge.hl7.org/gf/user/lingteng/) |
| 13834 | [Add seqeunce.readdepth](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13834&start=0) | Medium | [ling teng](https://gforge.hl7.org/gf/user/lingteng/) | [ling teng](https://gforge.hl7.org/gf/user/lingteng/) |
| 13918 | [Create sequence-reads profile on sequence](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13918&start=0) | Medium | [ling teng](https://gforge.hl7.org/gf/user/lingteng/) | [ling teng](https://gforge.hl7.org/gf/user/lingteng/) |
| 14044 | [Clarify Sequence.structureVariant fields](https://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=14044&start=0) | Medium | **Nobody** | [Bob Dolin](https://gforge.hl7.org/gf/user/r_dolin/) |

## 

Notes:

13289 - Search params - Xin will update and we will re-review.

* Should this include reference in some way?
  + Add additional param of Sequence.referenceSeq.referenceSeqId
* State specific use cases being solved by adding these as searchable. Bob D was able to provide one.
  + Include those use cases in the specification.

13834 - Sequence.readDepth - Xin will update and we will re-review.

* This was discussed a while back (should look for notes). Seems we used external sources for the definition. As of now, we use 'depth' in our definition of readCoverage.
* From Jeremy Warner to Everyone: (10:24 AM)
  + <https://www.illumina.com/science/education/sequencing-coverage.html>

13918 - Sequence-reads profile - Xin will review.

* Does providing access to reads go beyond our scope? Need to clarify the exact CDC use case.
  + Quality provided in other ways.
  + This is an outstanding question. Need to clarify further. Do not want to overly narrow our definition if it is not needed.

14044 - Clarify Sequence.StructuralVariant - Xin will review.

* Align our definitions with dbVar?
  + https://www.ncbi.nlm.nih.gov/dbvar/content/overview/

Chat Recording:

From Bret Heale to Everyone: (11:23 AM)

sequence ontology: http://www.sequenceontology.org/

verified that depth is not in SO...sorry

From Jeremy Warner to Everyone: (11:24 AM)

https://www.illumina.com/science/education/sequencing-coverage.html

From Bret Heale to Everyone: (11:32 AM)

Sequence.repository.readsetId

Definition

Id of the read in this external repository.

From Bret Heale to Everyone: (11:36 AM)

10.8.2.2 External Pointers

Sequence.repository: This complex element is used to provide a clarifying structure, a base URL, and/or relevant IDs when referring to an external repository.

GA4GH Repository Example. If the Sequence resource refers to a GA4GH repository for read info, references to a GA4GH full sequence dataset should conform to GA4GH data models and accessed via the GA4GH API. The URL of a GA4GH repository, ids of a GA4GH variant and read group are contained in the Sequence resource. The URL of a GA4GH repository is an api\_base of a GA4GH server that could be called for sequence data. The GA4GH variant set is a collection of call sets and the GA4GH call set is a collection of variant calls, typically for one sample. A variant call represents a determination of genotype with respect to that variant.

VariantSet definition: A VariantSet is a collection of variants and variant calls intended to be analyzed together.

CallSet definition: A CallSet is a collection of calls that were generated by the

# FHIR Subgroup Meeting Oct 12, 2017

## Attendees Sign In

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3. Andrea Pitkus - IMO - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
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11. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
12. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
13. Fan Lin --HMS and XMU - [fanatxmu@gmail.com](mailto:fanatxmu@gmail.com)

Presiding co-chair: Kevin Power

**Agenda:**

Connectathon/PGx use case, FHIR extensions alternatives

Please see Bob Dolin’s notes below re: connectathon proposal scenario.

(Added by Kevin) - Review this Zulip chat: <https://chat.fhir.org/#narrow/stream/implementers/topic/US.20GINA.20Implementation.20Guide>

**Dates:**

Oct 5 Schedules, V2/FHIR mapping

Oct 12 Connectathon/PGx use case, FHIR extensions alternatives

Oct 19 X

Oct 26 FHIR extensions alternatives

Nov 2 X

Nov 9 Xin/Fan present approach for R4

Nov 16 **#1, #2 + gForge (delayed)**

Nov 23 VMC

Nov 30 X

Dec 7 VMC (or Sync for Genes)

**Notes:**

* Bob Dolin presented Elimu's proposal for Jan 2018 FHIR Connectathon (which builds on the existing Pharmacogenomics connectathon scenario):

**Action**:

(EHR) Manually enter an order for "Imuran 50mg 1 tablet twice a day by mouth" into order entry system. This triggers a “medication-prescribe” CDS hook with a hook context of MedicationOrder which includes RxNorm code 197388 for azathioprine 50mg oral tablet.

(PGx CDS Service) Having been triggered by the “medication-prescribe” CDS hook, the PGx CDS Service now executes a decision support rule that [1] determines if the ordered drug has a known gene interaction; [2] determines, where there is a known drug-gene interaction, whether or not the patient has genetic test results on file; [3] determines, where there are genetic test results on file, if the patient has an interacting genotype; [4] determines, where there are not genetic test results on file, if the patient needs pre-testing. PGx CDS Service returns a CDS hooks “information card” back to the EHR, with appropriate recommendations.

**Precondition**: Observation(s) are available for some patients.

**Success Criteria**: Retrieve relevant observations (on TPMT gene, on allelic state) where they exist.

**Bonus point**: Different parameters (such as genomic coordinates) can be used for searching.

**Reference**: Azathioprine Pharmacogenomics - CPIC recommendations [https://cpicpgx.org/guidelines/guideline-for-thiopurines-and-tpmt/]

[Draft storyboard: <https://docs.google.com/document/d/1VqPrf6aaOB8RhSbAq2JYDxZYbDm-RT9DxA0u6-RxYo0/edit?usp=sharing>]

Discussion:

* What is returned if no result?
  + Today, would be a recommendation to do the test
* How to do PGx in FHIR?
  + Would use TPMT examples under the Observation-genetics profile
  + Perhaps we need to consider a PGx profile? Current Genomics document is an Implementation “Guidance”
* Bob D to share his “Story Board” for more PGx
* Vote: Approve the PGx CDS service use case as its own track for the next Connectathon - Lead is Bob D
  + Motion to accept: Bret H
  + Second: Bob M
  + Abstain: None
  + Nays: None
  + Yea: Andrea , Bob Dolin, Shennon, Clem, Xin, David, Usha, Amnon, Joel, Fan

Kevin will share component/extension document with CG group.

Topic 2: Xin presenting new approach for FHIR

* Introduce multiple profiles, break about single profile
* From Xin and Fan Lin

*The following link is the presentation and the staging FHIR site for the new genetics profile design which was presented in today's FHIR Subgroup meeting*

*PowerPoint:*

*[https://docs.google.com/presentation/d/1LQsVQJB9cX3s9kF53FMr9ir0ZLidAM-qdOB4H843xhc/edit#slide=id.g259d8e0ea5\_0\_9](https://na01.safelinks.protection.outlook.com/?url=https%3A%2F%2Fdocs.google.com%2Fpresentation%2Fd%2F1LQsVQJB9cX3s9kF53FMr9ir0ZLidAM-qdOB4H843xhc%2Fedit%23slide%3Did.g259d8e0ea5_0_9&data=02%7C01%7CKevin.Power%40Cerner.com%7C8e48bae5ab9947d81d4e08d51192571e%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636434242596236807&sdata=1POCiTF8CiLWFl%2BvE3aHHYEoinFyRRwj11b6Ai7raOM%3D&reserved=0)*

*Guidence/Staging Site:*

*[http://genomics-advisor.smartplatforms.org:4000/observation-genetic.html#content](https://na01.safelinks.protection.outlook.com/?url=http%3A%2F%2Fgenomics-advisor.smartplatforms.org%3A4000%2Fobservation-genetic.html%23content&data=02%7C01%7CKevin.Power%40Cerner.com%7C8e48bae5ab9947d81d4e08d51192571e%7Cfbc493a80d244454a815f4ca58e8c09d%7C0%7C0%7C636434242596236807&sdata=3rixL8IFjkWfoshb9QLgswgw01kH0EFjnJowezgMxBo%3D&reserved=0)*

CHAT:

From Bret Heale to Everyone: (10:08 AM)

have you seen the new(er) API from PharmGKB for providing information to meet information needs (i.e. infobutton-ish)

perhaps, including a input where the annotation of the presence of variant is through LOINC code and value 'Positive'

From Aziz Boxwala to Everyone: (10:11 AM)

thanks for the pointer to the API, Bret

From Bret Heale to Everyone: (10:12 AM)

np

From David Kreda to Everyone: (10:18 AM)

Bob M. suggestion is excellent.

From Bret Heale to Everyone: (10:26 AM)

<https://www.hl7.org/fhir/sequence.html> this is the Sequence Resource Spec

In HL7v2 it has been the common practice to provide the interpretation with the Observation

Phenotype - could be Condition Resource

From David Kreda to Everyone: (10:28 AM)

To be clear, a testable example of two paths is not objectionable. But all that Lloyd’s memo addressed was use components and (well a few other things). It was not about changing any of the other canonical approaches to advance the standard. If, and I gather it is desired, there is a Connectathon opportunity, then the set up for a trial should happen. And the onus for doing that pretty much has to be on Clem and team to prepare and submit for that test.

From David Kreda to Everyone: (10:34 AM)

It seems that this test case of Bob D.’s is fine. And we should also find out what was the specific feedback from the Sync for Genes PGx use case. I believe there was such - and I may have missed the assessment — but there is that other real-world assessment, too

From Bret Heale to Everyone: (10:35 AM)

have you seen the IG guide that Clem put together? He needs a place to host it for folks to evaluate it. In my mind it does a good job of bridging from HL7v2 to FHIR - I see the Sequence Resource as the Future, where data is exchanged more discretely. In HL7v2 you get the result of an observation, a bunch of order metadata, pateint data, interpretation - things which can be delivered in FHIR as seperate pieces...

From Bret Heale to Everyone: (10:35 AM)

Hope we have the discussion soon

From Fan Lin to Everyone: (10:42 AM)

Hi all ,excuse me , there are no much time for our introduction of Observation-genetic solution.

Xin introducing

From Bret Heale to Everyone: (10:42 AM)

i think the organizer needs to click on the hand to put the hand back down. Sorry, I can't un-raise my hand

nvm

I found it

From Bret Heale to Everyone: (10:52 AM)

how is this different from :<https://www.hl7.org/fhir/observation-genetic.html>

<https://www.hl7.org/fhir/observation-genetics.html>

From Andrea Pitkus, PhD, MLS(ASCP)CM to Everyone: (10:54 AM)

Do concur with the comment earlier that we need an implementation guide.

From Bret Heale to Everyone: ((null))

Andrea, the second link is one of our implementation guides (specific to observation genetics)

From Bob Milius to Everyone: ((null))

We have profile descriptions and an Implementation Guidance, but we don’t have a FHIR compliant Information Guide. Lloyd built for the Clem’s work

From Bret Heale to Everyone: (10:56 AM)

<https://www.hl7.org/fhir/observation-genetics.html> really?

<https://www.hl7.org/fhir/diagnosticreport-genetics.html>

From Bob Milius to Everyone: (10:57 AM)

<https://www.hl7.org/fhir/implementationguide.html>

Brett, that’s not an Implementation Guide technically

From Bob Milius to Everyone: (10:57 AM)

<http://wiki.hl7.org/index.php?title=FHIR_Implementation_Guide_Authoring>

<http://www.fhir.org/guides/registry>

Implementation Guide is a one or more Profiles used to address a particular problem

From Fan Lin to Everyone: (11:00 AM)

What you saw is current version , but not the introducing version by Xin

From Andrea Pitkus, PhD, MLS(ASCP)CM to Everyone: (11:02 AM)

thanks. should have clarified for FHIR. would ensur ewe have all the resources and whicch ones are needed to support the use cases/workflows, etc as it transcends several WGs.

# FHIR Subgroup Meeting Oct 5, 2017

## Attendees Sign In

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19. Kevin C. Ehlers -- BloodCenter of Wisconsin - [kevin.ehlers@bcw.edu](mailto:kevin.ehlers@bcw.edu)

**Agenda:**

Since this is first call for pre-New Orleans term, we will discuss the topics we get by Wed prior to the meeting (see list below). We will schedule and discuss updates (FHIR level 4 changes, **observation changes**, and connectathon changes). Also, it was requested we kick off with v2 modeling on Oct 5.

**Connectathon:**

Who is coming and topic of interest?

Bob Milius - not CG, but will work with BiologicallyDerivedProduct (Transplant)

**Notes:**

Idea of multiple tracks: General clinical genomics and pgx and hla,etc

Track on biological materials- Bob M. (with O&O)

Andrea- test order service catalog. No genetic use cases yet. Genetic test order- from test menu. Someone in ehr can order. Regular connectathon- lis and ehr testers. Clinicians on fhir. FHIR service catalog- lab test compendium. Labs and meds. Lorraine Constable from O&O. Claude Nanjo.

Pgx in cdisc see link below:

https://www.cdisc.org/standards/foundational/pgx

FHIR model based on v2 approach:

https://drive.google.com/file/d/0B6UpA50mwuwsQUxIS0FGM0hJS28/view?usp=sharing

Lloyd's document on extensions vs. components: https://docs.google.com/document/d/1hPjekT1AbR3ALL09FaK3dGn3xqJGECuhEPguJXFx3YE

**Links:**

**Connectathon:**

Here's an overview of the connectathon process: <http://wiki.hl7.org/index.php?title=FHIR_Connectathon_Track_Process>

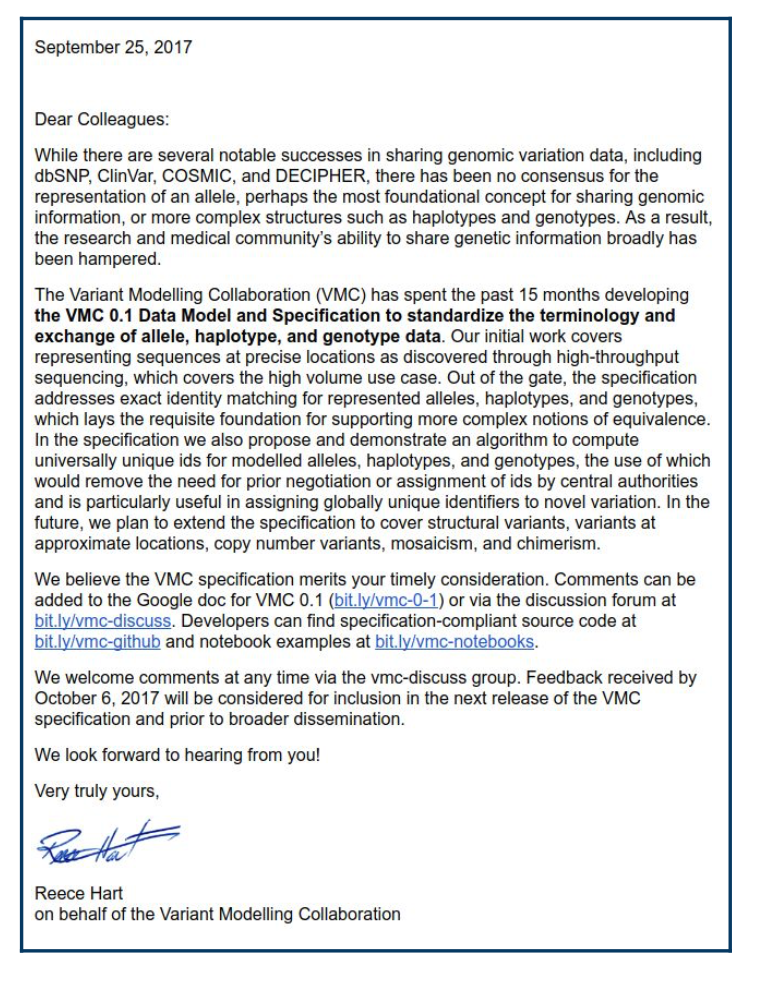
The Connectathon page itself is here: <http://wiki.hl7.org/index.php?title=FHIR_Connectathon_17> (It's a work in progress of course, but it does have a link to the tracks

Tracks are listed here: <http://wiki.hl7.org/index.php?title=Category:201801_FHIR_Connectathon_Track_Proposals>

And the main wiki page (with links to these two) is here: <http://wiki.hl7.org/index.php?title=FHIR>

<https://onfhir.hl7.org/2017/10/05/proposed-breaking-changes-for-fhir-r4/>

**Topics suggested for Fall 2017:**

1. Bob D.’s PGx use case for connectathon (Due: Oct 18)
2. Split Sequence.referenceSeq.strand into two fields:
   * **Sequence.referenceSeq.direction** = "forward" | "reverse". Definition: An absolute reference to a strand that is in an assumed chromosome orientation of P-arm on the left. The 5'-to-3' strand is the "forward" strand, and the 3'-to-5' strand is the "reverse" strand.
   * **Sequence.referenceSeq.sense** = "sense" | "antisense". Definition: A relative reference to a DNA strand based on gene orientation. The strand that contains the gene is the "sense" strand, and the opposite complementary strand is the "antisense" strand.
   * Deprecate Sequence.referenceSeq.strand
3. For all Sequence fields that indicate position, modify the definition to include something like "position is based on an assumed chromosome orientation of P-arm on the left".
4. VMC spec (comment period: Oct 6) Link: [bitly.com/vmc-0-1](http://bitly.com/vmc-0-1).  
   *This is the cross-stakeholder effort (GA4GH, ClinGen, ClinVar, HL7\*\*\*) focused on developing a model that could work across all organizations. Chaired by Reece Hart.  
   \*\*\* Gil A. David K., Bob F.   
   *
5. V2/FHIR mapping
6. FHIR extensions alternatives
7. Sync for Genes
8. gForge issues

**Dates:**

Oct 5 Schedules, V2/FHIR mapping

Oct 12 Bob D.’s PGx use case? / FHIR extensions alternatives?

Oct 19 X

Oct 26 FHIR extensions alternatives? / Bob D.’s PGx use case? (not as ideal)

Nov 2 X

Nov 9 #1, #2 + gForge?

Nov 16 Sync for Genes?

Nov 23 VMC?

Nov 30 X

Dec 7 VMC?

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# FHIR Subgroup Meeting Aug 10, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
3. Bob Dolin - [bdolin@elimu.io](mailto:bdolin@elimu.io)
4. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
5. Usha Reddy- Oracle - [usha.reddy.pa@gmail.com](mailto:usha.reddy.pa@gmail.com)
6. Grant Wood - Intermountain Healthcare - [grant.wood@imail.org](mailto:grant.wood@imail.org)
7. Xin Liu - [BCH-xinliu215@gmail.com](mailto:BCH-xinliu215@gmail.com)
8. Ling teng - BCH - [tenglingling@gmail.com](mailto:tenglingling@gmail.com)
9. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
10. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
11. Andrea Pitkus - IMO - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)

**Agenda:**

News and updates, Building new examples (modeled on DAM use cases): Implementation

Phaseset url->reference

Email news:

“In order to completely document the FHIR STU publication adjustment made to allow for publishing some of your committee's resources as STU, the Standards Governance Board are working to complete a list of the affected resources.

Could you please advise of the date CG approved the each of following FHIR Resources to ballot at STU by the end of August 2017:

Sequence

If you have any questions regarding this request please do not hesitate to contact me directly.

Regards

Paul

Paul Knapp

President

Knapp Consulting Inc.

[www.pknapp.com](http://www.pknapp.com/)”

# FHIR Subgroup Meeting Aug 3, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Monica Arniella - Vanderbilt University Medical Center - [monica.arniella@vanderbilt.edu](mailto:monica.arniella@vanderbilt.edu)
3. Michael Italia - Syapse - [michael.italia@syapse.com](mailto:michael.italia@syapse.com)
4. Bob Dolin [bdolin@Elimu.com](mailto:bdolni@Elimu.com)
5. Jeremy Warner - Vanderbilt University Medical Center- [jeremy.warner@vanderbilt.edu](mailto:jeremy.warner@vanderbilt.edu)
6. Kevin Power - Cerner - [Kpower@cerner.com](mailto:Kpower@cerner.com)
7. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
8. Ling Teng - BCH - [tenglinglint@gmail.com](mailto:tenglinglint@gmail.com)
9. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
10. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
11. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
12. Grant Wood - Intermountain Healthcare - [grant.wood@imail.org](mailto:grant.wood@imail.org)

**Agenda:**

**Aug 3 Somatic use case example in FHIR: leveraging existing databases**

Notes:

Speaker: Monica Arniella

Somatic use case example in FHIR : API and FHIR

SMART on FHIR Application

OncoKB

CLViC

PMKB

The Jackson Laboratory

FeedBack:

Jeremy Warner:

CarePlan resource: are looking for recommendation

Evidences, CarePlan, and Sequence:

* CarePlan address evidence and condition
  + Using Sequence for condition

Feel Pressure for conversion with cancer base?

* Moving away from conversion
* Sometimes not able to fit the traditional sequence data(Micheal)
  + Difficult to Map
* Don’t have agreement about structural variant.
  + V2 Lite do have some example to deal with structural variant
  + Have some updated in FHIR to deal with V2 Lite mapping for the structural variant

# FHIR Subgroup Meeting July 20, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
3. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
4. Grant Wood - Intermountain Healthcare - [grant.wood@imail.com](mailto:grant.wood@imail.com)
5. Jeremy Warner - Vanderbilt University Medical Center - [jeremy.warner@vanderbilt.edu](mailto:jeremy.warner@vanderbilt.edu)
6. Kevin Power - Cerner - [kpower@cerner.com](mailto:kpower@cerner.com)
7. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
8. Bob Dolin - Elimu - [bdolin@Elimu.io](mailto:bdolin@Elimu.io)
9. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
10. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
11. Monica Arniella - Vanderbilt University Medical Center - [monica.arniella@vanderbilt.edu](mailto:monica.arniella@vanderbilt.edu)

**Agenda:**

Building new examples (modelled on DAM use cases): Design

<http://www.hl7.org/implement/standards/product_brief.cfm?product_id=446>

<http://www.hl7.org/documentcenter/private/standards_temp_C4F284D0-1C23-BA17-0CF32702CA60467E/v3/HL7_DAM_CLINSEQ_R1_INFORM_2017FEB.pdf>

Hla use case: Bob M.

Use DAM for v2

CAP:

People for accreditation- Andrea

IMO-terminology

Counsyl- sync for genes

ARUP- digitize

Organizations- ignite and emerge- pgx

Grant- list

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V2 lri->fhir

Bob D.- v2 lri example and DAM use cases scenarios. (check with Clem/Shennon if already some use cases mappings)

**6.11 FDA Scenarios in Public Health Reporting**

HL7 Clinical Genomics is looking for specific representatives as partners to help inform this use case.

----> quality use case

(->“sharing for public consumption”)

->draft guidance doc.

-> There is a related group with FDA

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Technical appendix of v2 and fhir solutions?

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Grant- 6, 9.

Public health genomics group- u mich

Utah HIE

Clinical and Data warehouses

Omics repository and pilots

Grant- CAP person in ASCO- CAP contact. Rich Moldwin (cancer reporting aspects). cancer.

Andrea- CDC. NPCR, MU 2+3, Physician reporting ambulatory,etc. NCIC.

General clinical labs

Lab starndards

Cancer reporting/registeries

Grant- newborn screening (state by state)- contacts

Genetics counsellors

6.5.- group for that.

APHL

Andrea- new born screening people- O&O.

Gil-Tufts group

Making example for Use Case Scenarios:

- For everybody within the email list? Or public?

- May need technical Ga4Gh and FHIR

- Amnon Ptashek: can provide some new use case for this topic

- Some has example code related with FHIR(1-4)

- The rest (5-10) need to have example

- Grant Wood: have group for public health reporting

- Grant: (6 & 9 )

- Yan: (7)

- Pathology Report

- Newborn screening & Genome: Grant

# FHIR Subgroup Meeting July 13, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
3. Xin LIu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
4. Andrea Pitkus - IMO - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
5. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
6. Bob Dolin - [BDolin@psmiconsulting.com](mailto:BDolin@psmiconsulting.com)
7. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
8. Ling teng [-BCH-tenglingling@gmail.com](mailto:-BCH-tenglingling@gmail.com)
9. David Kreda - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
10. Monica Arniella - [monica.arniella@vanderbilt.edu](mailto:monica.arniella@vanderbilt.edu)
11. Scott Robertson - [scott.m.robertson@kp.org](mailto:scott.m.robertson@kp.org)
12. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)

**Agenda:**

Privacy and Security - Presented by John Moehrke, FMG ([johnmoehrke@gmail.com](mailto:johnmoehrke@gmail.com) )

Visit: <http://build.fhir.org/security.html>

Visit: <http://build.fhir.org/secpriv-module.html>

Visit: <http://build.fhir.org/security-labels.html>

Also: <https://healthcaresecprivacy.blogspot.com/p/fhir.html> <– John Moehrke’s blog site.

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# FHIR Subgroup Meeting July 6, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Andrea Pitkus -IMO- [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
3. Andrew Brown - NMDP - [abrown3@nmdp.org](mailto:abrown3@nmdp.org)
4. Bob Dolin - bdolin@psmiconsulting.com
5. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
6. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
7. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
8. Shennon Lu - NLM- [shennon.lu@mail.nih.gov](mailto:shennon.lu@mail.nih.gov)
9. Clem McDonald - NLM - [clemmcdonald@mail.nih.gov](mailto:clemmcdonald@mail.nih.gov)
10. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
11. Ling Teng -BCH [tenglingling@gmail.com](mailto:tenglingling@gmail.com)
12. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)

**Agenda:**

v2/FHIR mappings

V2/FHIR mapping plan:

1. See what is missing/different, etc, 2. Make a note of it, and 3. Add

in/change various elements so that things are aligned, and perhaps

most importantly: 4. Verify alignment via sample translator.

# FHIR Subgroup Meeting June 29, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Usha Reddy-Oracle -usha.reddy.pa@gmail.com
3. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
4. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
5. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
6. Ling teng -BCH [-tenglingling@gmail.com](mailto:-tenglingling@gmail.com)
7. Andrea Pitkus - IMO [-apitkus@imo-online.com](mailto:-apitkus@imo-online.com)
8. Bob Milius - NMDP - [bmilius@nmdp.org](mailto:bmilius@nmdp.org)
9. Shennon Lu - NLM - [shennon.lu@nih.gov](mailto:shennon.lu@nih.gov)
10. Clem McDonald - NLM - [clem.mcdonald@mail.nih.gov](mailto:clem.mcdonald@mail.nih.gov)

**Agenda:**

v2/FHIR mappings

**Ideas for in-depth focus connectathon:**

**PGx**

# FHIR Subgroup Meeting June 22, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
3. Usha Reddy- Oracle - [usha.reddy.pa@gmail.com](mailto:usha.reddy.pa@gmail.com)
4. Bob Dolin - [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
5. Joseph Kane - Epic - jkane@epic.com
6. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
7. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
8. Alex Mankovich - Philips - [alex.mankovich@phiips.com](mailto:alex.mankovich@phiips.com)
9. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
10. Ling teng -BCH [-tenglingling@gmail.com](mailto:-tenglingling@gmail.com)
11. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)

**Agenda:**

[http://Guidance.site](http://guidance.site)

Direct links:

<http://guidance.site/observation-genetics.html>

<http://guidance.site/sequence.html>

Sync for Genes recommendations:

<https://drive.google.com/open?id=0B-0YtCs_i_-eMnB6OUJrY05HMGs>

Mover: Ling Teng

Seconder: Jeremy Warner, David Kreda

# FHIR Subgroup Meeting June 8, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Bob Dolin - [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
3. Usha Reddy-Oracle: [usha.reddy.pa@gmail.com](mailto:usha.reddy.pa@gmail.com)
4. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
5. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
6. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
7. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
8. Ling teng - BCH -tenglingling@gmail.com

**Agenda:**

1. WGM’s

<http://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=12404&start=0>

Considered, No Action Taken

Second: Usher

Mover: Bob Dolin

Yes:Gil Alterovitz, Bob Dolin, Usha Reddy, Xin Liu, Joel Schneider, Bret Heale, Amnon Ptashek, Ling teng

No: none

<http://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=12508&start=0>

Considered, No Action Taken

Mover: Joel Schneider

Second: Bob Dolin

Yes:Gil Alterovitz, Bob Dolin, Usha Reddy, Xin Liu, Joel Schneider, Bret Heale, Amnon Ptashek, Ling teng

No: none

<http://gforge.hl7.org/gf/project/fhir/tracker/?action=TrackerItemEdit&tracker_item_id=13197&start=0>

Persuasive

Mover: Bret Heale

Second: Usha

Abstain: Joel Schneider

Yes:Gil Alterovitz, Bob Dolin, Usha Reddy, Xin Liu, Bret Heale, Amnon Ptashek, Ling teng

No: none

All the issues are resolved, and passed.

2. Fhir Connecthon 15 proposal

<http://wiki.hl7.org/index.php?title=FHIR_Connectathon_15>

Question: Would it be worthwhile to test out Scenario5 HLA Typing?

Suggestion: Add a link Scenario7.

3. Explain the Fhir Maturity Model

Suggestion: It will be good to reach out to people who are outside the HL7 group for their opinion about the genomics resources and profiles improvement.

Connectathon proposal- new Sept WGM proposal:

* <http://wiki.hl7.org/index.php?title=201709_Genomics>

# FHIR Subgroup Meeting June 1, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
3. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
4. Usha Reddy- [Oracle-usha.reddy.pa@gmail.com](mailto:Oracle-usha.reddy.pa@gmail.com)
5. Andrea Pitkus - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
6. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)
7. Michael Italia - Syapse - [michael.italia@syapse.com](mailto:michael.italia@syapse.com)
8. Bob Dolin - [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
9. Ling teng -BCH- tenglingling@gmail.com
10. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
11. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
12. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)

**Agenda:**

Sync for Genes

**Slides:**

https://drive.google.com/open?id=0B-0YtCs\_i\_-eMnB6OUJrY05HMGs

**Site:**

Guidance.site

# FHIR Subgroup Meeting May 4, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)
3. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
4. Bob Dolin - [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
5. Jeremy Warner - Vanderbilt [amended: Vanderbilt University Medical Center] - [jeremy.warner@vanderbilt.edu](mailto:jeremy.warner@vanderbilt.edu)
6. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
7. Andrea Pitkus - IMO - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)
8. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
9. Ling teng [-BCH-tenglingling@gmail.com](mailto:-BCH-tenglingling@gmail.com)
10. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
11. Yan Heras - Optimum eHealth - yanheras@gmail.com

**Agenda:**

Cytogenetics

Notes:

Video: <https://drive.google.com/open?id=0B-0YtCs_i_-eclRJRFRLLVNnb1k>

Slides: TBD

# FHIR Subgroup Meeting Apr 27, 2017

## Attendees Sign In

1. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
2. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)
3. Andrew Brown NMDP - [abrown3@nmdp.org](mailto:abrown3@nmdp.org)
4. Xin Liu - BCH - [xinliu215@gmail.com](mailto:xinliu215@gmail.com)
5. Jeremy Warner - Vanderbilt [amended: Vanderbilt University Medical Center] - [jeremy.warner@vanderbilt.edu](mailto:jeremy.warner@vanderbilt.edu)
6. Joseph Kane - Epic - [jkane@epic.com](mailto:jkane@epic.com)
7. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
8. Ling teng -BCH- [tenglingling@gmail.com](mailto:tenglingling@gmail.com)
9. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
10. Bob Dolin [bdolin@psmiconsulting.com](mailto:bdolin@psmiconsulting.com)
11. Andrea Pitkus - IMO- [apitkus@imo-online.com](mailto:apitkus@imo-online.com) (last 30 mins)

**Agenda:**

CMS/CLIA

Video: For internal HL7 Clinical Genomics Use only: <https://drive.google.com/open?id=0B-0YtCs_i_-eaVhQYnFVSC01LWs>

PDF doc with reference to CDA/CLIA:

<https://drive.google.com/open?id=0B-0YtCs_i_-eMmlfMmxna0NEVms>

Questions:

Specific questions:

1. Would CMS have any resources that can review HL7

ballots/implementation guides/FHIR resources under development to

provide feedback on implementation recommendations? In other words,

what uses would be allowed within CLIA and which would be problematic

for laboratory accreditation?

Yes

2. Similarly, are inspectors aware of FHIR, what it’s

implementations look like, which implementations are permissible and

pass inspection and which would be problematic and cause a laboratory

to have regulatory issues (get dinged)?

**3. One of the keys, which Clinical Genomics workgroup may wish to**

**distinguish are implementations that are utilizing EHR data from EHR**

**based databases, research databases, data warehouses, etc. versus**

**those implementations/use cases that involve the analysis of specimens**

**for genetic variances, and the generation of information around those**

**processes which is stored in the LIS. CLIA and lab regulatory**

**requirements are most heavily focused on the analytical phase of**

**testing (within the laboratory). There are requirements that extend**

**back to the EHR in the pre-analytical phase of testing (from when the**

**physician decides to place an order, to specimen collection, to**

**receipt in the laboratory), as well as those downstream from the**

**laboratory (results reported from the LIS to the EHR and downstream**

**systems, use of the results for clinical decision making and patient**

**care). Perhaps, CMS can share the types of violations, they**

**frequently see that are informatics based?**

4. The other key question is if FHIR apps are developed that pull

data from the LIS and the first downstream connected systems is a cell

phone app, is that permissible? How would laboratories do interface

checks on cell phones, patient computers or other first downstream

systems? It’s one thing when laboratories can get screenshots form

their internal computer systems, but when external, seems like privacy

and other aspects may apply.

Lab needs to validate interfaces. Needs to verify it gets from one to another.

Lab needs to verify that those results actually get there.

Going to a portal or ehr

Verify that connection.

If from portal, data is pulled, the lab is not responsible.

Kaiser perman

------------------------

Own account and password setup

Hipaa

Other comments/questions form people:

Many CLIA and lab accreditation aspects are based upon a traditional

workflow model of MD orders test in HIS/EHR, specimen is collected and

sent to the lab, lab professionals analyze the specimen and results

are created within the LIS, which are then reported back to the

ordering provider in their HIS/EHR. However, within FHIR, either a

push (from the lab) or a pull (from the app/API from a patient phone,

EHR or other system) could be implemented. Including CAP’s General

checklist requirements required across all laboratories they accredit.

For interface checks, the key is it applies to the 1st downstream

entity from the LIS. In most cases, this is the EHR from which lab

results are received. In other cases, it may be the public health

system to which ELR is reported. With FHIR, it is unclear how this

requirement would be met, if cell apps are directly querying the LIS

database? Would laboratories be required to get screen shots (as they

normally document this requirement to show to CMS inspectors) from

cell phone apps showing the comments, decimal points in the right

place, reference ranges, etc.? Seems highly impractical. Although

this is an example of a more stringent CAP requirement, it also

references CLIA and CLSI (Clinical Laboratory Standards Institute)

standards as well. What is CMS’ expectation? This would be a great

question for the call. Someone will likely have to walk them through

a laboratory example and how it would appear in the FHIR app, so they

can assess, given they likely won’t understand enough about how FHIR

is different/structured from HL7 V2 messages.

Sample of questions we have:

Not necessarily a genomics focus, but would be interested if the

CMS/CLIA people have had a chance to review any SMART/FHIR apps (e.g.

the Duke FHIR roundtable recording of the Bilirubin App) and have any

feedback to FHIR developers?

Interface check requirement- lab responsible that first downstream element (all things in right spot). Show in initial interface and later…

First portal vs after first portal differences.

Certain data it should show

EHR-S guide- has LCD

Are there any specific CLIA required data elements? Some items

labelled as CLIA are in v2 and people were wondering if certain fields

are "required"?

Yes- in recording.

One is it's not clear how the ACK, acknowledgements for receipt of

orders or results is specified? Does it matter or is simple ACK via

RESTFul API from FHIR sufficient?

Ask expert- about implementations.

Consistency- choose one way to do that.

**New questions:**

**-> If they have anything on ehr side and/or Fhir apps- if had any guidance to that- could they share?**

**-> Is it good enough to transfer the clia number with messages- or does it somehow need to be displayed?**

-----> in mu3- need certain

**-> look in the future: mu- does that add in new items of interest?**

2015->does cda support clia reporting?

V2- page 262

# FHIR Subgroup Meeting Apr 20, 2017

## Attendees Sign In

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7. Andrew Brown - NMDP - [abrown3@nmdp.org](mailto:abrown3@nmdp.org)
8. Joel Schneider - NMDP [- jschneid@nmdp.org](mailto:-jschneid@nmdp.org)
9. Ling teng -BCH - [tenglingling@gmail.com](mailto:tenglingling@gmail.com)
10. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)
11. Jeremy Warner - Vanderbilt [amended: Vanderbilt University Medical Center] - [jeremy.warner@vanderbilt.edu](mailto:jeremy.warner@vanderbilt.edu)
12. Usha Reddy- [Oracle-usha.reddy.pa@gmail.com](mailto:Oracle-usha.reddy.pa@gmail.com)
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14. Amnon Ptashek - Edico Genome - [genptashek@gmail.com](mailto:genptashek@gmail.com)
15. Alex Mankovich - Philips - [alex.mankovich@philips.com](mailto:alex.mankovich@philips.com)

**Agenda:**

Terminology

Future agendas:

1. Raw sequence - consent

Genomes- consumer testing

DAM- use case to add in

2. Genetic counseling- include literature and specialist area.

Provenance around variant assertions

Notes:

Bob M. will follow-up with Larry Babb on fhir Clinvar terminology server

**Recording link will be here:**

[**https://drive.google.com/open?id=0B-0YtCs\_i\_-eTldKS1dUcGRnYVE**](https://drive.google.com/open?id=0B-0YtCs_i_-eTldKS1dUcGRnYVE)

See chat below.

**Connectathon:**

Gil Alterovitz

Bob Milius

Martin Maiers

Joel Schneider

Chat:

From Me to Everyone: (11:29 AM)

Signin sheet/note:<https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5>

From Kevin Power to Everyone: (11:44 AM)

RE: Consumer Genomics - As long as groups like ACMG raise these sorts of concerns, utilizing that data in a clinical setting will be tough:<http://www.frontlinegenomics.com/news/11105/acmg-responds-23andmes-fda-approval>/

From Bret Heale to Everyone: (11:54 AM)

difficult...such as codes that are meaningful and need to be parsed...

nice work Joel

From Me to Everyone: (11:59 AM)

For new people who joined: Signin sheet/note:<https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5>

From Kevin Power to Everyone: (12:20 PM)

Is there a “good” coding system for biomarkers?

From Bret Heale to Everyone: (12:20 PM)

@Kevin - do you mean variants? or a code that represents a "phenotype"-"variant" pair

From Bret Heale to Everyone: (12:26 PM)

I would propose that the roll-up for the clinician is an implementation task

passing observed 'sequence' and 'snomed-CT' code for phenotype associated

would be computable

From Jeremy Warner to Everyone: (12:26 PM)

Clinical Oncology Treatment Plan and Summary IG

<http://www.hl7.org/implement/standards/product_brief.cfm?product_id=327>

# FHIR Subgroup Meeting Apr 13, 2017

## Attendees Sign In

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13. David Kreda - HMS - [david.kreda@gmail.com](mailto:david.kreda@gmail.com)

**Agenda:**

V2 mappings and testing (testing part deferred)

We mainly discussed the following V2/FHIR mapping/fields today:

1. observation-geneticsAllelicState

Problem: typo in definition "..Hemiplasmic indicates that the DNA Sequence

Variant is present in some but not all of the copies of mitoch.."

typo on both [loinc.org](http://loinc.org/) website and here, should be "Heteroplasmic"

Result: Clarified. Wording will be changed in LOINC and reflected in FHIR.

\*\*\*\*\*\*\*\*

2. observation-geneticsAllelicFrequency

Problem: definition on Fhir is different from V2lite

Fhir definition:

A physical quality which inheres to the allele by virtue of the number

instances of the allele within a population.

LOINC Code: (81258­6 (<http://loinc.org/81258>­6)

V2lite definition:

81258-6 Allelic Frequency [NFR] - Reports the fraction of all of the

reads at this genomic location that were represented

by the given allele. For homozygotes it will be close to 1.0;

for heterozygotes it will be close to 0.5.

It can be a smaller number when there are mosaics or multiple

chromosome, or mixtures of tumor cells and normal cells.

Result: need to clarify what “allelic frequency” will be used. One idea: Keep current “allelic frequency” and add in the LOINC concept, but name it based on SO term for fraction of reads or similar. Others ideas?

[LB] Have two distinct observations (which I believe they are) by qualifing the term a bit more. For example, popluationAlleleFrequency for the rate of occurrence with subjects of a group, cohort or population. And readAlleleFrequency for the occurrences within a specimen, tissue, sample, or genome of a given subject (which is critical for somatic testing). The former is more useful for annotating the called variant/allele when focused on determining a molecular interpretation of the individual allele on a case or in general. The qualifying terms should be decided by the experts “population” and “read” are just placeholders to demonstrate the potential approach.

I also think that while SO terms are fantastic and should be used everywhere possible, there will be a need to have overlapping LOINC terms which map as closely as possible. Not all groups using v2 will be willing to add terms for new authorities, unless v2 is willing to include SO terms in lieu of using LOINC terms for OBX-3 fields. It is my preception that LOINC owns the OBX-3 space for the vast majority of HL7v2 applications out there and thus will need to provide a coded term even if SO or other terminologies provide a logical alternative.

\*\*\*\*\*\*

3. observation-geneticsCopyNumberEvent

These two fields listed in the mapping are different.

Result: Explore LOH definition and use in list. Determine where closest mapping to the listed v2 term.

\*\*\*\*\*

4. observation-geneticsGenomicSourceClass

Result: Claried the change in answerlist, from Prenatal to Fetal and additional text for definition (except sentence on SCT, LA).

\*\*\*\*

5. observation-geneticsPhaseSet

Problem: email form Shennon:

"Also, we clarified that FHIR observation.extension –geneticsPhaseSet

is different from the v2 approach, which uses 2 LOINC codes – 82120-7

Allelic Phase and 82309-6 Bases for Allelic phase. The current V2

approach was chosen for simplicity and because it will cover most cases.

But we discussed v2 adding the phase set as an alternative, and FHIR moving

phaseSet to Sequence and adding the 2 codes (Allelic phase and Basis for

allelic phase) in Observation as related, components and/or backbone

elements."

Result: Need further discussion to finalize placement (e.g. discussed moving to Sequence) and discuss synergy with v2.

Chat:

From Me to Everyone: (11:32 AM)

Sign-in link:<https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit>#

From Me to Everyone: (11:57 AM)

<http://www.sequenceontology.org/browser/current_svn/term/SO:0002119>

From Me to Everyone: (12:02 PM)

A physical quality which inheres to the allele by virtue of the number instances of the allele within a population. This is the relative frequency of the allele at a given locus in a population.

From Bret Heale to Everyone: (12:07 PM)

+1 to the def provide by Gil

From Bret Heale to Everyone: (12:08 PM)

So, alleic frequency in a population versus the allelic frquency in a sample

From Me to Everyone: (12:08 PM)

That is cut/pasted from SO- they added one sentence recently.

From Bret Heale to Everyone: (12:12 PM)

oh. cool

thx

# FHIR Subgroup Meeting Apr 6, 2017

## Attendees Sign In

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11. Yin XIE (Translational software, yin.xie@TranslationalSoftware.com, [yinxie@gmail.com](mailto:yinxie@gmail.com))
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**Agenda:**

Oncology-related FHIR items

Structured docs

Supporting assertions for somatic and/or germline

Somatic testing panels->treatments and prognosis

Get treatment recommendations

Depending on the lab to determine what clinicians should do

Implications of the findings including what can be effective or not

Pubmed id typically associated with them

Parent observation resource

Related artifacts.

Observation does not allow for reference to related artifacts

From Warner’s chat comment:

The current genetics observation profile does not have a clear way to represent genomic rearrangements/translocations/fusions that do NOT have genomic coordinates (if they do have coordinates, you can use the sequence resource). Examples: 1) FISH; 2) cytogenetics; 3) NGS where one partner is known but the other is unknown or not relevant (e.g., ALK rearrangement in lung cancer is actionable without knowing what the partner gene is, so labs may not always report the partner [or the partner is unknown or outside the scope of the NGS panel])

# FHIR Subgroup Meeting Mar 30, 2017

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15. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)

**Agenda:**

David Hay on Clinfhir

Video: <https://drive.google.com/open?id=0B-0YtCs_i_-eUGVGSU1FVnhCR1k>

# FHIR Subgroup Meeting Mar 23, 2017

## Attendees Sign In

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13. Bret Heale - Intermountain Healthcare - [bheale@gmail.com](mailto:bheale@gmail.com)

**Agenda:**

Updates (e.g. testing)

v2lite-FHIR mappings

FHIR-v2lite mappings

**Note:**

David:

MITRE, Aeigis, Sync4Genes

Talk to testing people before connectathon- Richard, Jason?

Bob M/David K:

During the Tuesday, March 21 CGWG call, Bob Milius asking after two questions:

1. Are we exercising all the elements and profiles in the FHIR Spec c/o all of the Use Cases and the Connectathons? David Kreda asked if perhaps we could ask both of AEGIS about their [Touchstone Project](http://www.aegis.net/touchstone.html) and MITRE [Crucible](https://projectcrucible.org) suite how their testing systems record/tabulate if their tests thoroughly traverse these assets. We should direct emails to Richard at AEGIS and Jason at MITRE about this

2. Beyond the “checklist” of this, how can we evaluate (record) the adequacy/satisfaction with the use of the spec, for example, are implementers finding use as natural or easy use case or flow, or are implementers having to shoehorn things in (and how); is the hierarchy of structure (e.g., sequence) easy to use or cumbersome, etc.

**Topics for the future:**

* sync4genes
* Terminology Services
* Terminology services for the genomic for the code systems we have on the pages (Bob, Joel, Larry- Clinvar+Clingen) (scope: valuesets and binding)
* Joel talk 15 min intro?
* Links:
  + Terminology module:
    - <http://build.fhir.org/terminology-module.html>
  + How to use/develop terminology services
    - <http://build.fhir.org/terminology-service.html>
  + This contains lists of code systems and value ses (including those for Clinical Genomics) that are registered for FHIR
    - <http://build.fhir.org/terminologies-systems.html>
  + The open source FHIR implementations also have varying levels of support for the terminology service specification:
    - <http://build.fhir.org/implsupport-module.html>
  + (Open Concept Lab is new to the FHIR scene.)
    - <https://openconceptlab.org/>
    - <https://github.com/OpenConceptLab>
    - <https://ohie.org/>
* Terminology service that is fhir compliant- so people could add clinvar ids and lookups

# FHIR Subgroup Meeting Mar 9, 2017

## Attendees Sign In

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8. Joel Schneider - NMDP - [jschneid@nmdp.org](mailto:jschneid@nmdp.org)

Agenda:

HAPI- James Agnew

Video:

<https://drive.google.com/open?id=0B-0YtCs_i_-ec21ucFEtNEwtVmM>

Slides:

N/A

# FHIR Subgroup Meeting Mar 2, 2017

## Attendees Sign In

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11. Andrea Pitkus - IMO - [apitkus@imo-online.com](mailto:apitkus@imo-online.com)

Agenda:

MITRE Crucible, SyntheticMass/Synthea- Jason Waloniski

Video of presentation:

<https://drive.google.com/open?id=0B-0YtCs_i_-ecmJKVUxVVldTQ2s>

Slides:

<https://drive.google.com/open?id=0B-0YtCs_i_-eYjhFNzJvYzh0NGM>

# FHIR Subgroup Meeting Feb 9, 2017

## Attendees Sign In

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Agenda:

Continue on STU4 discussions tomorrow and host speaker on

PGx/FHIR/v2lite.

[Get link to Xiin Liu’s PGx Update presentation]

[List of questions - approximate - Gil to edit?]

1. Automaticity of the process (any human-in-the-loop needed) to go from v2Lite to FHIR bungle
2. Coding issues [Bob Milius comments/questions - complicated - Bob?]
3. Implementation guidance issues/suggestions based upon the TranslationalSoftware presentation?
4. Placing “interpretation results” into diagnostic report ...
5. Issue of gap or not in data in PDF that is sent from (?) labs
6. Loss of any data from v2Lite source message to FHIR bundle and *vice versa*
7. Review of the HL7 Clinical Genomics DAM document - review of the workflow for PGx and diagram - send to Kenny Leung/Xin Liu/Yin Xie to review/comment

Potential Problem for the future

* figure out how to deal with the collection specimen data and the analysis specimen data
* might need to figure how to store the gene quality data of the vcf in the FHIR
* Multi Sequence conversion for the Phenotype result
* PDF result document problem

# FHIR Subgroup Meeting Feb 2, 2017

## Attendees Sign In

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7. Gil Alterovitz- BCH/HMS - [gilusa@gmail.com](mailto:gilusa@gmail.com)
8. Grant Wood
9. Elizabeth Newton
10. Joel Schneider

Agenda:

We had a speaker cancellation for tomorrow (will be rescheduled), but

still plan to meet. Will continue discussion from WGM. Also, if

anyone is interested to present, please feel free to email me directly

so we can coordinate a slot. We have some topics allocated and have

room for more. From prior notes, it would be especially great to hear

from perspective of regulatory (e.g. CLIA), internal hl7/external efforts, FHIR

tooling, labs, vendors (customer needs), and approaches on new use

cases.

-------

Reach out to all again to get stakeholder speakers

Amnon before end of Mar? See if works for him. Email him. David Kreda feels this come up in Feb while memories still fresh.

Ask if next wk or

CLIA- Andrea

V2 - FHIR resources - to see which ones map for CLIA

Content vs workflow pieces

Tracking specimen different from patient

FHIR use for normal workflow

* Specific thing to focus on for the genomic use case
* Representing right process for the specimen (example: representing the Left/Right kidney the specimen from)
* EHR care about they send to the Lab
* Action: look for a comprehensive use case to look at workflow/clia.

CLIA rep.- CMS

Prioritize on results first (MU1)

**Possible topics:**

Tutorial-based feedback: Implementation guide (add definitions), add family hx profile to IG, documentation

**STU3 review/STU4 Focus Topic Candidates Discussed Today**

1. Family History
2. Clinical
3. Precision medicine/other efforts
4. Pharmacogenomics update- Xin, Kenny
5. Cytogenetics and FHIR- yan heras, others?
6. Precision cancer medicine update- jeremy/john holt
7. FHIR tooling- Dan Gottlieb (FRED), David Hay/Viet (ClinFHIR- with scenario builder/graphical models), MITRE crucible demo/synthesia, Aegis update, HAPI/server test tools, other tools?, v2->lab mappings/tools, **implementation guidance additions on tooling?**- list of alternatives,
8. clingen efforts
9. use cases: emerge/geneinsight xml format and examples
10. CSER/IOM
11. Structured docs/CDA-based models
12. v2 mapping/harmonization
13. Stakeholder feedback from fhir pilots
14. Synchronize with new DAM use cases
15. SDC
16. EMR’s
17. **CLIA**- focus on CG-aspects. ask CMS person to come in to see specific applicability in CG.
    1. People at semantic interop- Karen Dyer

Bob Diederly- clinical use cases on lab workflows. ONC people. Involved in v2 calls.

Labcorp, quest, cap, pathologists

Kevin- focus on CG-specific?

Focus on

* giving test result from the lab.
* how to use FHIR the include the genomic data from the lab

24. Pilot-based results/input

Conferences?:AMIA and other conferences

**Weeks:**

Feb 9 STU4 Planning, v2lite/FHIR mapping and Pharmacogenomics update- Xin, Kenny

Feb 16 STU4 Planning

Feb 23 (HIMSS)

Mar 2 MITRE Crucible, SyntheticMass/Synthea- Jason

Mar 9 FHIR servers/HAPI- James

Mar 16 STU4 Planning

Mar 23 STU4 Planning- v2 mappings

Mar 30 Clinfhir- David (record)

April 6 Jeremy and references, Structured docs and cda mappings

April 13 v2 mappings / Testing (deferred)

April 20 Terminology / Sync for Genes

April 27 CMS and CLIA

May 4 Cytogenetics- Yan

May 6-12 WGM Madrid Agenda:

1. Sync for Genes feedback
2. Others?

**Toward San Diego WGM:**

May 18 Post WGM no meeting

May 25 GA4GH/BioIT no meeting

June 1 Sync for Genes Suggestions

June 8 v2 and FHIR mapping (moved to June x?)

June 15 NIH meeting

June 22 Sync for Genes review

June 29 v2 and FHIR mapping (29th or another day)?

July 6 v2 and FHIR mapping

July 13 Privacy and security

July 20 Building new examples (modelled on DAM use cases): Design

Aug 3 Somatic use case example in FHIR: leveraging existing databases

Aug 10 Building new examples (modelled on DAM use cases): Implementation

Aug 17 Vacation?

Aug 24 Vacation?

…(feel free to add topics/dates here):

**Previous years’ notes:**

<https://docs.google.com/document/d/1WDEskUVDbr1hBV33SqbHjnlIx_13rpP20UnPssXfZQA/edit?usp=sharing>