FHIR Subgroup Meetings 9/30/19 - 1/27/20

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Presiding co-chair: Gil Alterovitz  
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Schedule

- Sept 30 - WGM recap and final trackers for publication
- Oct 7 - examples recap, next steps
- Oct 14 - X - federal holiday (mCode alignment moved to Oct 15)
- Oct 21 - new topics for next cycle
- Oct 28 - mCODE alignment
- Nov 4 - additional alignment considerations with mCODE and UScore, eg
- Nov 11 - X (Veteran's Day)
- Nov 18 - DevDays prep/publication recap
- Nov 25 - GACS operations/next steps
- Dec 2 - trackers old and new
- Dec 9 - mCODE update, extract-variants, implication proposals
- Dec 16 - new proposals
- Dec 23 - X - HAPPY HOLIDAYS
- Dec 30 - X - HAPPY HOLIDAYS cont.
- Jan 6 - New year, new holidays!
- Jan 13 - New trackers / implications for pgx
- Jan 20 - X - MLK Day
- Jan 27 - WGM prep

FHIR Subgroup Meeting January 27th, 2019

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

1. Bret Heale - Intermountain Healthcare - bheale@gmail.com
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3. Patrick Werner - MOLIT Institut - pw@molit.eu
4. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
5. Ning Xie - BCH - ningxie2018@gmail.com
6. Liz Amos - NLM - liz.amos@nih.gov
Agenda:
1. WGM prep
2. $find-variants proposal
3. Other trackers

Discussion:
1. WGM prep
   a. Example fixing
   b. Phenopackets review
      i. Approach to knowledge artifacts, especially
      ii. Gap analysis
      iii. Ways to help/get connected with group
      iv. VR spec variants scope questions
   c. Minimal variant profiles
      i. SPDI representation
         1. Could add to variant but may only increase complexity
         2. Still needs normalization for comparison, similar to HGVS
      ii. Clem’s proposal (not covering *-alleles/etc)
   d. Other topics
2. $find-variants proposal
   a. FHIR-25250 Genomic Data Server Operations
   i. Returning normalized variants, using components:
      1. component:genomic-ref-seq: NC_000019.10
      2. component:ref-allele: G
      3. component:alt-allele: A
      4. component:coordinate-system: 0-based interval counting
      5. component:exact-start-end: start = 11089559
   ii. Q: would *-alleles be in scope for this?
   iii. A: you could grab the variants with this and then use them to compute haplotypes/etc. Returning those from raw is out of scope for now.
   iv. Variants may be stored in FHIR with various representations, or in native structures
   v. Q: should we recommend more error codes for failed normalization? Particularly liftOver, going from build37 to build38
1. If the query range doesn’t liftOver, return an error

c. Capability Statement in terms of our IG:
   i. Propose not including in “main” capability statement for the IG, would be a separate use case a server could support.
   ii. Lloyd: could mark as optional, but knowing who should be thinking about should be relevant.
   iii. No capability statements yet in the IG,
   iv. We can have one in our spec to highlight what should system shall/should/may support
   v. Production instances also have capability statements (this is what I currently support). May even change based on user authentication.

d. Patrick: May not have direct impact in querying reports directly
   i. Our importing pipelines are already taking care of normalization for our use cases

e. Bob D: other operations and approaches would be needed to cover structural vars, genotypes/haplotypes, etc.

f. Proposal: include
   i. http://build.fhir.org/ig/HL7/genomics-reporting/branches/fhir_operations/find-variants.html and
      In build version of IG.

3. Other trackers
   a. FHIR-25296 Uncallable subregions in a region studied -suggest persuasive w/ mod,
      i. Next steps were to finalize text offline, see
         https://chat.fhir.org/#narrow/stream/179197-genomics/topic/STU2.20Theme.3A.20Uncallable.20subregions.20in.20a.20region.20studied

   b. FHIR-24598 How to reference a region studied observation from genotype, haplotype, variant observations -notes from before:
      i. Clem: we don’t need a region-studied for the variants we found, it’s really to cover the negatives/what was tested but isn’t represented in the variants otherwise in the report.
      ii. Kevin: region-studied is “what was tested/targeted”, haplotype/etc is “what was found”
      iii. Bob W: we want to know how far into introns/promoters were sequenced in order to consider follow-up testing.

   c. FHIR-19844 PGx High Risk Allele Medication Impact is confusing -suggest persuasive with mod

Chat history:

Bret Heale11:04 AM Phenopackets handling of knowledge as in thier git hub as collaboration point.
FHIR Subgroup Meeting January 13th, 2020

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9. Liz Amos - NLM - liz.amos@nih.gov

Agenda:

1. Requests for new components/guidance not addressed last week:
   a. FHIR-19844 PGx High Risk Allele Medication Impact is confusing
      i. “Risk allele” being added by ACMG for PGx
      ii. Different for somatic

Current page for Implication considerations:
https://docs.google.com/document/d/1SYdzxanCgwzhhBjCBrquZ6H8HdjQFjG_l2nufAkCGU/edit#heading=h.n5eokak2o6ca
Jamie’s scratch notes/gap analysis from eMERGE example PGx implications:
https://docs.google.com/spreadsheets/d/1Lhs2MtY4l8UgxnVBu5K68b5L9m7YLqCJffjczXBZox8/ed it#gid=883800343

- Thoughts on using Observation.text (Narrative) for text in eMERGE extension:
  - can/should we describe a referenced observation (genotype) in the narrative of the implication?
  - Narrative.status can be flagged as “additional”, to show
    - The contents of the narrative may contain additional information not found in the structured data. Note that there is no computable way to determine what the extra information is, other than by human inspection.
  - Some implementations will have trouble interpreting narrative, should be held onto except in rare cases where can be certain it is generated/not needed
  - Regarding a recommendation, can this be in text?
- Other option: include text in valueString: (or markdown, if it gets added to Observation)

- Thoughts on relatedArtifact being used inside components vs only at base Observation:
  - May be able to update extension context to allow this use, rather than requiring additional referencing from the extension.

- Thoughts on converging profiles via diagnostic/therapeutic use cases:
  - Need to see proposal

Difficulties combining multiple medications on one profile: may want to restrict to combination therapies for consideration of cancer use cases.

Discussion:
Chat History:
Bret Heale 11:38 AM an extension is nice as I one does not have to support extensions
11:39 AM so, the sender of the extension must include the information elsewhere or risk the recipient not having the information
FHIR Subgroup Meeting January 6th, 2020

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6. Liz Amos - NLM - liz.amos@nih.gov
7. Bob Wildin - University of Vermont robert.wildin@uvmhealth.org
8. Daniel Rutz - Epic - drutz@epic.com
9. Kevin Power - Cerner - kpower@cerner.com (join at :06)
10. Joel Schneider - NMDP/CIBMTR (at :10)
11. Bret Heale - Intermountain Healthcare - bheale@gmail.com

Agenda:

2. Requests for new components/guidance
   a. FHIR-24906 Need to include chromosome as a component to ObsVariant
   b. FHIR-25296 Uncallable subregions in a region studied
   c. FHIR-24598 How to reference a region studied observation from genotype, haplotype, variant observations
      d. FHIR-19844 PGx High Risk Allele Medication Impact is confusing
      e. FHIR-25418 Require "Genetics" category on report

3. Updates on recent proposals for new profiles/resources
   a. FHIR-18988 TMB / MSI status profiles
   b. FHIR-25250 Genomic Data Server Operations
   c. “Adequate PGx”
      d.
      e.
Discussion:

1. New items
   a. FHIR-24906 Need to include chromosome as a component to ObsVariant -suggest not persuasive,
      i. Proposals:
         1. keep one field for all info (with better guidance)
         2. Make new fields for chromosome, band, more granular info
      ii. Thoughts (including a more philosophical/logistic discussion on requiring presence of reference sequences)
         1. Bob W: chromosome is inferred from refseq, shouldn’t need to be separate
         2. Clem: shouldn’t reference sequence be required? This is also included in ISCN where that is supplied. There are a few cases where the reference isn’t available but not many. Related to HGVS (which also requires a reference sequence to be properly formatted).
         3. Bob D: the rendering of the chromosome name on the report is for human consumption, easily calculated from the build and/or reference. Not every element in the report needs to be structured in FHIR.
         4. Bob M: just need to be careful with supplying multiple ways to provide similar/same information. In HLA, we want to have reference sequences, but often don’t get it. They just provide nomenclature and the reference sequences have to be derived by a tool/lookup. (versioned)
         5. Daniel: this creates a challenge as it requires the consumer/receiver of the information to do work and process the data to derive the sequence. Having more fine-tuned profiles as well with required data fields will help move towards high-quality data culture in contracts/etc.
         6. May: can’t support requiring sending reference sequences for all
         7. Bob D: references seem more required in variants than in haplotypes/genotypes.
   iii. Next steps:
      1. If this field is meant only for humans, it should be stated as such
      2. Other implementations/profiles can constrain variant to require certain data fields.

b. FHIR-25296 Uncallable subregions in a region studied -suggest persuasive,
   i. Arose in an attempt to derive star alleles from VCF files--was a certain region measured as wild type, vs was it an uncalleable region?
   ii. Example: region-studied gene X, with integer ranged regions and “non-callable” sub-regions given.
iii. Often don’t get desired read-depth at certain exons for example.
iv. Bob W: is the threshold for callability being standardized/reported?
v. Bob D: current proposal is to punt that decision to the lab
vi. Drafted definition of new term:

'A non-callable region is a subregion within an observation:region-studied profile that represents a range for which variants are unable to be called. Generally, a region is non-callable due to technical issues surrounding the test. Multiple non-callable regions may be contained within a region studied.

In many cases, a recipient of a FHIR Genomics report may only be interested in identified variants, in which case non-callable regions may not be of use. In some cases however, such as where there is a desire to infer the presence of a wild-type allele at a particular position, it can be useful to know that not only were no variants reported at a position, but also that the position was not deemed non-callable by the lab'.

**Next steps:** finalize text offline, caution against non-callables potentially not being represented (especially all in one place). Clarify that non-callable is relative and currently left to the lab’s discretion.

c. **FHIR-24598** How to reference a region studied observation from genotype, haplotype, variant observations

  i. Clem: we don’t need a region-studied for the variants we found, it’s really to cover the negatives/what was tested but isn’t represented in the variants otherwise in the report.

  ii. Kevin: region-studied is “what was tested/targeted”, haplotype/etc is “what was found”

  iii. Bob W: we want to know how far into introns/promoters were sequenced in order to consider follow-up testing.

**Next steps:** better guidance needed for the universal IG regarding purpose of profile, more thought into individual use cases and examples with must-support/etc.

d. **FHIR-19844** PGx High Risk Allele Medication Impact is confusing

  i. Should include with PGx redo

e. **FHIR-25418** Require "Genetics" category on report

2. Updates on recent proposals

  a. **FHIR-18988** TMB / MSI status profiles

  i. Delayed due to Epiphany

  b. **FHIR-25250** Genomic Data Server Operations

  i.

c. “Adequate PGx” (needs a JIRA)
i. Thoughts on reporting individual textual statements as Implication.value to provide further context for encoded components:

1. Chat History:

Bret Heale 11:13 AM larry Babb in zulip commented on what if someone sends Genomic Build without a reference (I believe) and needing chromosome to be specified.

Bob Milius 11:15 AM Regardless of how we report chromosome, we need to have guidance (narrative) in the IG on how to do this (one way, or several different ways).

Bret Heale 11:16 AM +1 bob m

Bret Heale 11:18 AM Yep. +5 to Bob D. We’re not building the UI or report, but providing calculable data elements that computably send the information within the report.

Bret Heale 11:19 AM If we provide guidance on how to calculate a narrative that might help reduce the perceived diff btwn textual report and structure.

Bret Heale 11:25 AM +1 bob m. current ig is meant to be universal. good point

May 11:30 AM right…agree with Bob. For mCODE, we just expect the nomenclature and not the reference sequence

May 11:31 AM We don’t expect EHRs to store reference sequences.

Bret Heale 11:31 AM Cerner can store it

May 11:32 AM Can is different from does. That is implementation dependent.

Bret Heale 11:32 AM Cerner does store it. If sent via the API for Lab Sequence

May 11:33 AM For which sites other than IM?

Bret Heale 11:33 AM Anyone who has Lab Sequence

May 11:33 AM Who has that would be a Cerner question

May 11:33 AM @Kevin? Care to weigh in?

Kevin Power 11:34 AM @May - Drop me an email and we can chat about it.

Bret Heale 11:34 AM: ^ )

May 11:34 AM.

Bret Heale 11:35 AM @May the sender has to send it and use the Lab Sequence API. We're in a crossroads where technology is ahead of typical practice.

May 11:36 AM true, but it’s not absolutely necessary to drive an NCCN pathway having reference sequence data. If you require it, then we might need mCODE to breakaway.

FHIR Subgroup Meeting December 16th, 2019

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Agenda:

1. TMB & MSI proposal Update
2. $extract-variants update
3. Implication refactoring proposals (delayed for further call)

Discussion:

1. TMB & MSI Update (Patrick)
   MSI and TMB as their own ObservationProfiles derived from:
   http://hl7.org/fhir/uv/genomics-reporting/StructureDefinition/genomics-base
   a. LOINC doesn’t cover these concepts (yet?), we can use NCIT
      i. NCIT TMB: Tumor Mutation Burden (NCI Code C150128, also LOINC 94076-7)
         valueRatio (ratioDenominator fixed to 1000000{Base})
         o Synonymous or not?
         o Been cautioned in the past to not hard code method into the concept, need to supply in Observation.method
         o LOINC code for interpretation of TMB LOINC: 94077-5
      ii. NCIT MSI: Microsatellite instability (NCI Code C36318, see also LOINC 62862-8)
      iii. valueCodeableConcept:
         MSI-High (MSI-H), MSI-Low (MSI-L), or Microsatellite Stable (MSS)
   b. Re: using NCIT in general (vs LOINC here)
   c. Haven’t used NCIT in IG yet, should be able to download their codesystem (as TSV?) and upload to HAPI/etc
      i. Joel: NCIT is an aggregation of codesystems, do we know where this particular concept has come from? FHIR currently only has the metathesaurus as a resource.
         Liz: thesaurus and meta-thesaurus are separate, can tell by start of the code
      ii. Patrick: https://ncit.nci.nih.gov was used by mcode. Question on http vs https. All urls on FHIR are http currently
iii. Bret: what is the technical reason why http was used as the convention?
iv. Patrick:
d. Freely available
e. May: had met with others at CIBMTR, lots of support for NCIT, used
f. NEXT STEPS:
   i. Patrick will reach out to vocabulary to confirm if they have a stance on ‘s’ vs not (https://chat.fhir.org/#narrow/stream/179202-terminology/topic/https.in.CodeSystem.20URI)
   ii. New profiles in dev branch along with NCIT codeSystem
   iii. Confirm MSI LOINC code availability, possible (Microsatellite instability [Interpretation] in Cancer specimen Qualitative 81695-9)

2. Implication profiles proposals (delayed)
   a. FHIRpgxV1.01.pdf (previous slides by Bret kicking it off)
i. Total changes from proposals so far:
   ○ 3 somatic profiles → 1 somatic implication
     a. TBDcode: somatic-implication
     b. Value: diagnostic | prognostic | predictive
     c. Previous `Observation.code-value` pairs added as components
   ○ 4 pgx profiles → 1 pgx implication
     a. Placeholder code: https://loinc.org/51965-2/
     b. Value: Medication Efficacy | Medication Metabolism | Medication Transporter Function | High Risk Allele
        i. Bob M: semantically weak, “what is the implication?” suggest removing value (0..0)
        ii. Jamie: could send lab’s text here, what they’d normally put on their narrative report (eg, “Severely reduced inhibition of vitamin K reductase”)
     c. Previous `Observation.code-value` pairs added as components
     d. Addition of `associated-phenotype` component for flagging high-risk phenotypes
   ○ 1 inherited disease pathogenicity profile, unchanged.

ii. Pros:
   ○ Fewer boxes
     a. Eases understanding of our model
   ○ Allows flagging high-risk phenotypes

iii. Cons:
   ○ PGx efficacy and somatic-predictive still disjoint
   ○ Germline diagnosis and somatic diagnosis still disjoint
   ○ inh-dis-path code/value structure different from other implications
Addition of `associated-phenotype` potential confusion with current metabolizer/transporter components

iv. Other consideration:
   o As context for a CodeableConcept changes across profiles and use cases, its meaning may become obscured.
     a. Our Observation profiles are semantically linking concepts, we need to be clear in the relations they are affirming, and take care to keep those relations obvious and consistent.
     b. E.g., ‘Associated-cancer’ component can appear either as diagnosis or given context for therapy/prognosis. If we call it the same thing, there is a risk of misinterpretation.

v. Next steps:
   o Suggest refactoring inh-dis-path code/value into component for consistency
     a. Possibly further refactor into “diagnostic-implication” and “therapeutic-implication”
       i. https://docs.google.com/document/d/1SYdzxanCgkwzhBjCBrquZ6H8HdjQFjG_I2nufAKCGU/edit# (doc for brainstorming)

3. $extract-variants
   a. https://docs.google.com/document/d/1VSs6pXNW468hjSQ9GjTOoJXBR_6Mq0LaM3Jltgn5Ds/edit#
   b. Bob D: question on logistics of explaining the query response, do we need to create profiles (on our parent profiles) and link them to the output?
   c. Lloyd: a human readable SHALL in text to describe what the operation should do and return is just as good, no known software currently that consumes operationDefinitions.
   d. Bob D: would prefer to deliver information similar to the linked google doc.
   e. Bret: the act of creating the profiles shouldn’t be a bottleneck
   f. Patrick: suggest leaving the strict profiles out for now, the key is the ability for implementers to be able to validate the output
   g. Jamie: more rigid use-case profiles are on the way in other areas as well, suggest holding off for alignment with that
   h. Patrick: want to see similar functionality with search queries
      i. Returning resources present with the needed components populated
         o Vs
      ii. Calculating the needed component information based off of identifiers

Open floor:
May: mCODE STU1 candidate 1 to be released tomorrow
Handful of major negs, May will follow-up
Chat record:
[Dec 16, 2019 at 11:03:30 AM] Bret Heale: Conclusion of the genetics report goes in diagnostic report profile. correct?
[Dec 16, 2019 at 11:04:45 AM] Bret Heale: not today. thanks
[Dec 16, 2019 at 11:05:11 AM] Jamie: https://docs.google.com/document/d/1FGCQRtxJKyHhnClub_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.51h85art0bts
[Dec 16, 2019 at 11:36:26 AM] Bret Heale: my question was simply to help anyone on the call not panic about an additional profile :^) the inclusion in the IG for TMB is to help in finding the right loinc code etc. somethings are super easy.
[Dec 16, 2019 at 11:41:15 AM] Bret Heale: https://loinc.org/81711-4/ has a pattern of different msi biomarkers. be sure each of the specific biomarkers can be modeled (as examples).
[Dec 16, 2019 at 11:59:13 AM] Bret Heale: +1 jamie

FHIR Subgroup Meeting December 9th, 2019

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21. Patrick Werner - MOLIT Institut - patrick.werner@molit.eu
Agenda

1. mCODE update (May)
2. Extract-variants operation (Bob D)
3. Implication refactoring proposals (didn’t cover)
4. TMB & MSI proposal Update (didn’t cover)

Discussion

4. mCODE update (May)
   a. Ballot Reconciliation Status
      i. More alignment with CG IG
         o More granularity for variant representation
         o Extensions realigned to components
         o Variant Tested + Variant Found combined, pulled components from our Variant profile.
            a. Bob M: Our Variant was closer to Tested, the distinction with ‘Found’ may be needed to consider later
         o Region Studied pulled in as Observation (with some of our components)
            a. Bob M: if you have multiple variations, how do you associate with different portions of region-studied?
         o Specimen updated to reference from extension
         o LOINC codes alignment
      ii. How many errors/warnings are there between an example conformant to mCODE re: our IG
         o HGNC/HGVS (lack of support on tx.fhir.org = extra errors)
            a. Error is specifically with the validator, unsupported codesystems should be WARNINGS, not ERRORS
         o DiagnosticReport.category cardinality 1..*
         o Observation.interpretation
      iii. Next steps:
         o Reconsider concerns with Variant code/value pair (if we should model the variant Found elsewhere)
         o Reconsider linking findings with regions studied, if it needs to be computational/references
         o Reconsider Observation.interpretation usage (May to show examples in Zulip)

   a. Coding:
      i. System   LOINC

   d. Region Studied pulled in as Observation (with some of our components)
      a. Bob M: if you have multiple variations, how do you associate with different portions of region-studied?
      o Specimen updated to reference from extension
      o LOINC codes alignment
   ii. How many errors/warnings are there between an example conformant to mCODE re: our IG
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      o Reconsider concerns with Variant code/value pair (if we should model the variant Found elsewhere)
      o Reconsider linking findings with regions studied, if it needs to be computational/references
      o Reconsider Observation.interpretation usage (May to show examples in Zulip)

      a. Coding:
         i. System   LOINC
5. Extract-variants operation [https://jira.hl7.org/browse/FHIR-25250](https://jira.hl7.org/browse/FHIR-25250)
   a. Proposal from Bob D:
      [https://docs.google.com/document/d/1VSs6pXNW468hjSQ9GjTOoJXBR_6Mq0LaM3Jlt7gn5Ds/edit#](https://docs.google.com/document/d/1VSs6pXNW468hjSQ9GjTOoJXBR_6Mq0LaM3Jlt7gn5Ds/edit#)
      i. Region studied = yes, return report, regardless of variations found
      ii. Region not studied = 404 error
      iii. Region ‘partially studied’??
         ○ Additional component on Region-studied for uncallable subregions
            a. Eg. 8k-9k uncallable, give me 1-10k
            ○ Separate tracker to log
   b. Need input on:
      i. Name/scope of operation: $find-precise-variants vs $find-variants
         ○ Treatment of ‘fuzzy ends’ (no exact-start-end)
            a. Trying to keep query rigorous
            b. Query and return parameters change for imprecise/structural variants. May require other information for large variants
               i. Consider 2 separate operations (and one combined)
      ii. Cardinality of returned components:
         ○ Should we enforce 1..1 or open it up to 0..1 and employ ‘must-supports’

Chat history:

**Bret Heale**
11:13 AM
nice slides.

**Bret Heale**
11:17 AM
want to save it for a targeted discussion...but relating observations is a possibility

**Bret Heale**
11:31 AM
1) option preferred - a hack on the FHIR tooling

**Bret Heale**
11:31 AM
2) option - change to text and not coded

**Bret Heale**
11:31 AM
2 loses that this is an HGVS string

**Bob Milius**
Host
11:31 AM
it an error or warning?

**Bret Heale**
11:31 AM
error
Bob Milius  Host 11:32 AM+1 patrick

Bret Heale 11:32 AM+1 patrick

Bret Heale 11:32 AM+10

Bob Milius Host 11:32 AM Bindings are also informative to implementers

Bret Heale 11:32 AM 'gates to (semantic) hell'

Bret Heale 11:33 AM was that recorded in the min - that HGVS error is Gram tooling error (pardon spell mistake)

Bret Heale 11:34 AM use .text as 'positive'? with present code

Bret Heale 11:48 AM as long as the codings are the same

Bret Heale 11:48 AM in meaning

Bret Heale 11:50 AM Thanks May for your patience with me : ^ )

Bob Milius Host 11:54 AM Why 1-based coordinates? I think 0-based is better for computational stuff (like operations)

Kevin Power Host 11:54 AM Might want to also consider ’region partially studied’

Bret Heale 12:01 PM favor two operations AND a third that gives both

FHIR Subgroup Meeting December 2nd, 2019

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
2. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
3. Kevin Power - Cerner - kpower@cerner.com
4. Patrick Werner - MOLIT Institut - patrick.werner@molit.eu
5. Ning Xie - BCH - ningxie2018@gmail.com
6. May Terry - MITRE - mayT@mitre.org
7. Bob Dolin - Elimu Informatics - bdolin@elimu.io
8. Liz Amos - NLM - liz.amos@nih.gov
9. Bret Heale - Intermountain Healthcare - bheale@gmail.com
10. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
11. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com

Agenda

1. Themes docs: All Feel free to write any additional comments/feedback
   a. Especially feedback on priorities
      1. https://docs.google.com/document/d/17r-HNm-gyqthepU40gqh39uYK9PV3HmBl1VM-vW6TuY/edit
   b. Most recent comments have been surrounding “implication” profiles:
      1. https://docs.google.com/document/d/1SYdzxanCgkwzhhBJCBrguZ6H8HdjQFjG_l2nuAkCGU/edit#

3. Trackers
   i. FHIR-25250 - FHIR Operations for a genomic data server
      1. Persuasive with mod, awaiting more detailed proposal from Bob D on inclusion of first search operation.
   ii. FHIR-18988 - CG IG should support: MSI, TMB, PD-L1
      1. Persuasive with mod, awaiting more detailed proposal from Patrick
   iii. FHIR-16394 - Add comment to 1.2 Guiding principles
      1. Persuasive with mod, add to guiding principles (fifth bullet), “to help older systems accomplish this, Observations should not be contained as inline references.”
   iv. FHIR-13834 - Add sequence.readdepth
      1. Needs more discussion
   v. FHIR-19844 - PGx High Risk Allele Medication Impact is confusing
   vi. FHIR-16871 - Specialization for somatic variant might not be necessary - 2018-May Genomics #56
   vii. FHIR-16834 - Separating it into a separate profile may make it seem disconnected. - 2018-May Genomics #43

Discussion

1. Themes doc:
   a. Themes docs: All Feel free to write any additional comments/feedback
      1. Especially feedback on priorities
         a. https://docs.google.com/document/d/17r-HNm-gyqthepU40gqh39uYK9PV3HmBl1VM-vW6TuY/edit
         2. Most recent comments have been surrounding “implication” profiles:
            i. https://docs.google.com/document/d/1SYdzxanCgkwzhhBJCBrguZ6H8HdjQFjG_l2nuAkCGU/edit#
2. Trackers

i. **FHIR-25250** - FHIR Operations for a genomic data server

1. Level of inclusion

   a. **$extract-variants** seems like a great starting place, either included on [Query Examples](#) or linked there to its own appendix. Concern over returning a DiagnosticReport vs a bundle of variants (see **FHIR-16394**).

   i. Additional objects:

      1. region-studied - uncallable regions is valuable
      2. Sequence-phase-relation - can also be represented
   
   ii. Kevin: may want to call it something like “extract region” if we are not sending just variants

   iii. Patrick: was first concerned with DR vs Bundle but it is a report… should be coded separately than a report coming from a lab. Chromosome location has cytogenetic-location code (48001-2). Current debate on chromosome number vs reference sequence (alternate contigs issue if not providing full build). OperationDefinition.resource has to be determined. Could be “Observation” so the URL would be: [http://{baseUrl}/Observation/$extract-variants](http://{baseUrl}/Observation/$extract-variants) but Patient as the base is also possible.

   iv. **Current guidance is to avoid contained resources, favor bundle (with a DR and its results) as return**

      1. Bundle is how you get a bunch of things back, but semantic packaging isn’t done by bundle it would be in the references made on the report itself. Bundle is just the delivery.

   b. Next steps:

      i. **More solid proposal for the OperationDefinition resource, most useful approach would be to create it in a branch of the IG**

   c. Additional operations should require separate trackers.

   d. Search operations should be fleshed out on [the DAM](http://)

      i. May be some overlap with bulkdata API, keeping these operations patient-specific may have added value

   ii. **FHIR-18988** - CG IG should support: MSI, TMB, PD-L1

      1. MSI / TMB observations at the DR level, as apply to many variants/overall view of patient. Need codes from LOINC to be finalized.
2. Need proposal/group - Patrick looked at mCODE tumormarker test profile, do we want to have something like this in the IG? (re: PD-L1, etc). Need to document how to link the biomarker (positive/negative) observation to an underlying variant observation.

3. From which WG should this profile be constructed? Us or CIC etc

4. Bret: biomarkers are abstractions from an underlying genetic variation, using derivedFrom on the observation is appropriate. May be a use case for MolSeq as a knowledge resource (doesn’t have to be provided, just accessible). LOINC codes for biomarkers is a moving target as they are highly pre-coordinated. Again, having a good test definition with region studied - for example - would help. Perhaps use observationDefinition instance referred to in a biomarker finding.

5. Bob D: struggle to see how this is different from much work in O&O, helpful to lay out how genomic biomarkers differ from other tests.

6. May: may not have direct information at the variant level,

7. **Next steps**: Patrick create MSI/TMP profiles, reach out to CIC for histology test alignment (international LOINC/etc codes)

iii. **FHIR-16394** - Add comment to 1.2 Guiding principles
   1. Update with opinion on “Contained” resources, consider applying warning constraint
   2. **Next steps** - Jamie to propose disposition with details, similar to interpretation 0..0 change.

iv. **FHIR-13834** - Add sequence.readdepth
   1. Also other treatments of MolecularSequence, need proposal/group
   2. Read depth in variant currently, need to consider which elements should be duplicated per their scopes.

3. **Recent Implication proposals (didn’t cover today in detail)**
   a. **FHIR-19844** - PGx High Risk Allele Medication Impact is confusing
      i. [http://build.fhir.org/ig/HL7/genomics-reporting/HLAB1502-pgx-example.html](http://build.fhir.org/ig/HL7/genomics-reporting/HLAB1502-pgx-example.html)
   b. **FHIR-16871** - Specialization for somatic variant might not be necessary - 2018-May Genomics #56
   c. **FHIR-16834** - Separating it into a separate profile may make it seem disconnected. - 2018-May Genomics #43

d. Slides from Bret:
   [https://drive.google.com/open?id=0By30dDKZH69jdE0UGVUeHhOLUxYRERkekN3RWVkSmpNWmNF](https://drive.google.com/open?id=0By30dDKZH69jdE0UGVUeHhOLUxYRERkekN3RWVkSmpNWmNF)
   i. High level:
      1. Decide on what ‘High Risk Allele’ scope really is. Do we need something separate for Potential for Adverse Event.
2. Vote on whether or not the increased efficiency in querying constitutes a strong imperative to change the PGx profiles to components.

3. Select/Approve a group to finish selecting LOINC codes and implement switch to components.

4. Vote on whether the change is important enough to be a quick version update.

5. PROVIDE additional documentation on use. E.g. this power point has some textual statements that clarify how specific elements are used. Place such statements in the structure definition – not data element description.

6. NOTE: level of evidence is currently adequate for the PGx use case as it allows any evidence system, such as PharmGKB’s classification of evidence for PGx to be used. If a group needs to constrain then that group can do so with an implementation guide based on the CG WG implementation guide.

ii. Details for medication implication profile proposal:
   1. Observation.code bound to LOINC code 61357-0
      https://loinc.org/61357-0/ See the term description. Consider to request something similar or use it. Also https://loinc.org/51965-2/
   2. Observation.value bound to a value set of the current profile LOINC codes. Binding as ‘preferred’ (extensible) to ‘Medication Efficacy’, ‘Medication Transporter,’ ‘Medication Metabolism’, ‘High Risk Allele’
   3. Components are for providing more granular information, each of ‘Medication Efficacy’, ‘Medication Transporter,’ ‘Medication Metabolism’, ‘High Risk Allele’ would be a component, with a corresponding value-set (e.g. the component for the current Medication Efficacy would use the value set from the current profile)
   4. Add component for Potential adverse drug event – OR use the High Risk component?
   5. Suggest to add associated phenotype to the component for Potential Adverse Drug Event. So one can state an associated phenotype.

iii. Think: The observation is ‘what kind of variant implication does the genetic variation have’ where the values are ‘Medication efficacy’, ‘Medication transporter’, ‘Medication Metabolism’, ‘High Risk’- ‘Potential Adverse Drug event.’

iv. Second layer are the components themselves with component.value providing a more granular statement. The component.code is used to indicate which type of implication the component.value is for (e.g. ‘Medication efficacy’ in component.code with component.value of ‘Intermediate metabolizer’)

Chat History:

Bret Heale
11:13 AM
choromosa location can accomodate chromosome
Patrick
Host
11:14 AM
48001-2
Patrick
Host
11:14 AM
Observation.component:cytogenetic-location.code
Patrick
Host
11:14 AM
only have to get rid of "cytogenetic-"
Bret Heale
11:16 AM
works for chromosome
Bret Heale
11:16 AM
right?
Patrick
Host
11:17 AM
in my opinion yes (we are already using it that way) but there are other opinions
Bret Heale
11:18 AM
: ^ ) the universal IG is great, if folks need further specification then they can create an IG for more stringent use cases
Bret Heale
11:18 AM
being a bit devilish
Bret Heale
11:20 AM
I was just saying that Chromosome 7 could go into the component in its current form as well as 7p2.1
Bret Heale
11:21 AM
: ^ ) was not arguing against a reference sequence
Kevin Power
11:24 AM
Example Operation (on base Observation resource):
http://build.fhir.org/observation-operation-lastn.html

Patrick Host
11:30 AM
OperationDefintion.resource is what i meant.

Patrick Host
11:30 AM
and i think this should live on Observation

Patrick Host
11:31 AM
so {baseUrl}/Observation/&extract-variants

Bret Heale
11:39 AM
my concern with biomarkers is providing a mechanism to provide the variant level data with the biomaker statement

Bret Heale
11:40 AM
this can be done with related Observation to an instance of our variant observation

Bret Heale
11:40 AM
of course it is optional

Bret Heale
11:45 AM
good point. Perhaps the definition of the varaints covered should be in a defition of hte test

Bret Heale
11:46 AM
+1 to value set of lab tests

Bret Heale
11:53 AM
I would ask the Information Modeling subgroup to define a biomarkers relationship to genetic variation. It is important in determining how one merges varaint data and biomarker data.
FHIR Subgroup Meeting November 25th, 2019

https://join.freeconferencecall.com/clingenomics

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

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

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9. Bret Heale - Intermountain Healthcare - bheale@gmail.com
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11. Joel Schneider - CIBMTR/NMDP - jschneid@nmdp.org
12. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
13. 

Agenda:

1. IG publication review
2. DevDays feedback
3. Next steps/STU2 Items
   a. GACS operations introduction (also to be discussed Tuesday)
   b. Somatic Implication consolidation
   c. Other Proposals
      i. Avenues to encourage volunteers for content/ideas

Discussion:

1. IG publication review
   a. The “propose a change” link isn’t re-routing to JIRA
   b. Issue known to Grahame, should follow up
2. DevDays feedback
   a. Amsterdam talk introducing the IG (Patrick)
      i. Over 400 participants total, interest in genomics, ~40 people
      ii. Room temp issues ;)

iii. Tumor board software vendor interested in our PGx structure (Barcode Healthcare)

iv. NHS member active on Zulip

v. “Meet & code” sessions was more free-form than before

vi. Any follow-ups for us to consider?
   1. Starting with interest in variants/dicom, looking at adapting their data model.
   2. Also pointed to PGx implications, which could be refactored in the future, but will remain stable/available as STU1
   3. Clem: would be great to see their variant model for gap analysis/etc

vii. May Terry also gave a presentation (gathering clinical requirements, process that went into building mCODE IG)

viii. Patrick updated previous slides to newer UML diagrams, need to revisit the latest LOINC codes in the examples.

3. Next steps/STU2 Items
   a. Bob D - GACS operations introduction (also to be discussed Tuesday)
      i. Developing specifications capable of searching and creating/extracting resources from other resources and files—beyond simple search operations
         1. Data normalization (consider multiple optional ways to represent same variant):
            a. HGVS
            b. Clinvar
            c. Position, start, end, ref, alt
         2. Proposal: How to get started?
            1. Consider operations people are currently using
               a. Query across multiple ways IG permits modelling specific variants/findings
               b. Query across patients for research purposes
               c. Creating resources
            2. Patrick: FHIR specifies the interface, a server could be storing resources in any structure as long as it returns FHIR resources.
            3. Clem: I’d like to see it.
            4. Bret: normalization beyond the search parameters is important
            5. Bob D: should this go in the IG or own PSS?
               a. Patrick: in terms of finding resources from our IG it should be included
   b. Somatic Implication consolidation
      i. Currently have 8 implication profiles (and 3 abstract implications). Are these all necessary? Comments made in the past to combine some
         1. 19844 - PGx High Risk Allele Medication Impact is confusing
         2. FHIR-16871 - Specialization for somatic variant might not be necessary - 2018-May Genomics #56
3. **FHIR-16834** - Separating it into a separate profile may make it seem disconnected. - 2018-May Genomics #43

ii. previous proposal: Align 3 somatic implications to one as in:

http://build.fhir.org/ig/HL7/genomics-reporting/branches/kpower_SomaticImpl
ication/index.html

iii. Observation.code can be one of the 3 classes

iv. Observation.value then may change based on the code

1. Or use present/absent etc and put current suggested values in
   components.
2. Requires splitting lists into components and/or using
   invariant/constraints:

   https://www.hl7.org/fhir/conformance-rules.html#constraints
   a. Can use FhirPath
   b. Bret: may be worth spelling out pros/cons using this in the IG

v. Could consider further aligning into a Single Implication

1. List all the fields for what we have
2. Define individual use cases from there

vi. Can’t hide complexity, can just move it around

1. Should consider again what groups are currently sending

vii. Currently have ways to tie into

1. Associated phenotypes (only on inh-dis-path)
2. Associated cancer (only on somatic profiles, treated inconsistently)
3. Assessed medication (multiple contexts)

viii. Currently lacking:

1. Good description of adverse drug events
   a. Eg, “Increased risk of adverse event”
   b. Tried looking at High-risk allele (created from very specific
      use case)
   c.

ix. Next steps:

1. Identify set of individuals to review reports
   a. Are all attributes in the reports present in our profiles?
   b. Are any attributes implied (that should be communicated)
   c. Feed back into the model

2. Alignment between pgx and pgx-somatic should be something to
   keep in mind
   a. Somatic-diagnostic seems separate
   b. Pgx-somatic comes from tumor boards
   c. Other pgx

3. Inherited-disease pathogenicity should also be more widely
   applicable, need to cover “generic implication” use case
   a. Mandellian risk of vs diagnosis?
c. Other Proposals
   i. TBD-codes for variant lengths
   ii. Avenues to encourage volunteers for content/ideas
      1. Have tried lists of names based on attendance before, was voted against inclusion in current version in favor of larger “work group” credit.
      2. Should define metrics/credit system as early in project/cycles as possible
      3.
Chat history:

Bret Heale 11:19 AM the complexity is the way genomics is today, and is reflected in our universal IG. As Bob is describing

Bret Heale 11:38 AM it was discussed in the past that adopting CPIC guideline based value sets for the 'classes' in pgx was desired...I think I like the idea of moving the Metabolism, Transporter and Efficacy into components rather than profiles. just a thought?

Bret Heale 11:39 AM that is, the code and associated value sets


ah noe, that would be an instance of an event, not the reporting of a risk

FHIR Subgroup Meeting November 18th, 2019

https://join.freeconferencecall.com/clingenomics

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

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

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17. Bob Dolin - Elimu Informatics - bdolin@elimu.io
18. Kevin Power - Cerner - kpower@cerner.com
19. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
20. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
21. May Terry - MITRE - mayT@mitre.org
22. Bret Heale - Intermountain Healthcare - bheale@gmail.com

Agenda:

1. Temp co-chair slot (deadline from listserv is today/now)
2. Publication progress update
3. DevDays Prep (Patrick unable to attend)

4. STU2 Items
   a. JIRA tracker system
   b. Somatic Implication Alignment
   c. Other Proposals

Discussion:

1. Temp co-chair slot (deadline from listserv is today/now)
   a. Looking to fill co-chair scheduling gaps, get additional person with more formal
      authority to drive changes for STU2
   b. Vote tomorrow to open up temporary slot (through Dec 2020)
      i. All co-chair slots set to start/end at the plenary meeting/good through end of
         year
   c. Additional vote to fill the slot (current ballot is Jamie Jones and Bret Heale)
   d. Question over precedent and procedure, will follow up
      i. Some other work groups have 6 or more already

2. Publication progress
   a. Final error troubleshooting in frozen build
      i. Lynn finishing troubleshooting on local build, should be soon.
   b. Press release draft
      i. Confirm timeline with Lynn/Anne
         1. Sometimes days to weeks
      ii. HHS clearance for Clem quote

3. DevDays Amsterdam (Patrick unable to attend today)
   a. Anything else needed?
   b. Request for materials/overview

4. STU2 Items:
   a. JIRA trackers (replaces gForge for tracking items)
      ii. Lots of new features and QoL improvements, new trackers (and old) will be
          going through here.
      iii. JIRA accounts needed (should be setup for everyone)
   b. Somatic Implication:
      i. Concerns with current approach:
         1. AMP level of evidence system conflates evidence with significance
         2. Cardinality of associated-cancer
            a. Presence of non-cancerous conditions may change prediction/prognosis
         3. Scope of cancer vs phenotype vs disorder
4. DerivedFrom base Observation resources (without declaring a profile)
5. Somatic-predictive vs medication-efficacy lists
   ii. Initial proposal: Align 3 somatic implications to one as in:
       http://build.fhir.org/ig/HL7/genomics-reporting/branches/kpower_SomaticImpl
       ication/index.html

       1. Observation.code can be one of the 3 classes
       2. Observation.value then may change based on the code
          a. Or use present/absent etc and put current suggested values in components.
          b.

Chat History

Gil Alterovitz
11:04 AM
https://static.politico.com/c2/42/b1dcfe2f4fd8906e9416de3b6290/img-4309-1.jpg

Kevin Power
11:05 AM
Congrats Gil!

May
11:30 AM
Can you share that search link?

https://jira.hl7.org/browse/FHIR-25187?jql=project%20%3D%20FHIR%20AND%20resolution%20%3D%20Unresolved%20and%20Specification%20%3D%20%20Genomics%20Reporting%20(FHIR)%20%5BFHIR-genomics-reporting%5D%22

FHIR Subgroup Meeting November 4th, 2019

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

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3. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
4. Bob Dolin - Elimu Informatics - bdolin@elimu.io
Agenda

1. Update on publication
2. Follow-up on mCODE tracker resolutions (re: alignment w/ CG IG)
3. Call for proposals from previous topics (listed October 21)

Discussion

1. Update on publication
   a. FMG call 10/30 passed decision (!)
   b. TSC decision next (e-vote sometime this week)
   c. Then: follow-up with Anne/Lynn/Grahame

2. Follow-up on mCODE tracker resolutions (re: alignment w/ CG IG) Hoping to publish after this reconciliation (roughly aiming to finalize publication by February 1st)
   a. Remaining for block vote 3 (November 13)
      i. 23994 cardinality on DR category (previously locked to 1 from DSTU2 considerations) previously considered for future use, updating to persuasive
   b. Some voted in block 2
      i. 23999, 23987, 23997 general comments on alignment for genomics report and variant profiles. Recap on the structures from mCODE:
         1. Genomics Report - contain results of genomic analyses. Genomic reports vary in complexity and content, as simple as the results for a single discrete variant to complex sequences that are found in exome and genome-wide association studies (GWAS).
         2. Genetic Variant Tested - used to capture the results of a test for a single known variant.
         3. Genetic Variant Found - used to record variants that could be found from tests that broadly analyze genetic regions (e.g.: exome tests) and stores results for any variants that could have been found. If the implementer uses GeneticVariantFound, then the region in which the variant was found could be specified in the RegionStudied attribute of the GenomicsReport profile.
   ii. Open questions on the alignment:
1. Region Studied as Observation vs extension/other resource
   a. Observation approach: a tool (GATK) is run on the raw files (BAM) and based on read depth, the output conveys which regions were callable vs not studied
   b. Concern with using less mature/common resources for required/widespread adoption (mandate states ~15 resources in the model for EHRs).
2. Pointing to published IG is able to do through validator (use build version for now, should update to the latest published version once available).
3. Exercise: validate examples against both structure definitions, examine errors/warnings
   a. Many reports available in CG IG shared folder: Mayo/Illumina/others
   b. Keep in mind there may be data available not present in current reports - need to get to the source/site
4. Major issue with differing scope of tests: specific vs wider sequencing
   c. Other trackers coming in block vote 3 - list should be sent Wednesday (11/6), one week before vote (11/13).
      i. Items will be sent on Zulip
3. Call for proposals from previous topics (listed October 21):
   a. General alignment (with e.g. US Core):
      i. Bob D: should aim to develop towards a real derivation of genomic profiles. (Need to solve the diamond of death for incompatibilities between CG IG and US Core).
         1. Main items are US Core extensions, some other constraints.
         2. Several SNOMED requirements in the US, potentially blocking bindings (extensible)
         3. Could reach out to consider relaxing required bindings/extensions to ease alignment.
            a. Would need a group to volunteer: to determine what the diffs and blockers between (validating against both) the IGs are then decide best guidance based on that.
               i. Could create US version of CG profiles, to be available as a starting point for US work
               ii. Could request changes to US core if any real blockers there.
   b. Major discussion of breakpoints between tumor markers and genetic tests (discussion last week re: TMB and others) see tuesday notes
      i. 2 other topics tomorrow: continuation of updates from AllofUs and eMERGE
   c.
   d. Other topics listed:
i. relatedArtifact extension upgrade
   1. [https://chat.fhir.org/#narrow/stream/189875-genomics-.2F.20eMerge.20Pilot/topic/relatedArtifact.20extension.20change.20request](https://chat.fhir.org/#narrow/stream/189875-genomics-/eMerge/Pilot/topic/relatedArtifact+extension+change+request)
   2. Need a tracker for this discussion (Jamie will follow-up)
   3. Fear of becoming a garbage bin for miscellaneous items.
      a. Need a place to provide references particularly for medications implicated
   4. Lloyd: there is a place to list relatedartifacts at the root: recommend having the extension point to ids,
      a. Looking for intra-resource references (especially with extensions that contain mainly just ids/references)
      b. Similar to paper citations (list refs in one place, use pointers in the components to specific references).
   c. Jamie will log/gather resources today (GF25170)

ii. Genomic finding codes vs components
   1. Incorporation of Information Modelling
   2. Need official update from Bob F in terms of what gets exposed

iii. DevDays update (Patrick off this week)

Next Steps:

1. Jamie to log tracker re: relatedArtifact request and find supporting info for internal id referencing per Lloyd’s suggestion.
2. May to share other CG items for mCODE vote.
3. Still looking for volunteers to do gap analysis with US Core and determine the scope of work group wants to take on

CHAT LOG:

[Nov 4, 2019 at 11:01:17 AM] Ning: Please sign in: tinyurl.com/fhirgenomics
[Nov 4, 2019 at 11:03:51 AM] Jamie: https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHP5tSLLQ/edit#
[Nov 4, 2019 at 11:19:57 AM] Bret Heale: components are a good option when the item should be included as part of the observation
[Nov 4, 2019 at 11:27:45 AM] Bret Heale: good point on SNOMED CT Bob M
[Nov 4, 2019 at 11:41:48 AM] Bret Heale: +1 to May's comment that the data prior to being sent to the EMR was structured. This is our Catch-22 as she noted. Have to drive EMRs to recognize that they need to request structured data from their reporting labs in genetics
[Nov 4, 2019 at 11:41:48 AM] Bret Heale: We have a few tests at Intermountain and are working on getting more genetic test data in a structured format (our EMR vendor has been helpful)
FHIR Subgroup Meeting October 28th, 2019

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Sign In: (presiding co-chair - Bob Milius)

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Agenda

1. mCODE alignment trackers
   a. 23994, 23999, 23987, and 23997
2. proposals on topics from last week

Discussion

1. 23994 question on increased cardinality on report.category (upgraded to 0..* in R4)
   a. May: are any systems currently using multiple categories? We definitely want to align with users locked into previous version.
   b. Lloyd: in general, should try to align to latest version of spec where differences like this (or resource name eg)
   c. Future use case for scoping data access to different users (not aware of any systems enabling this yet).
d. Multiple categories allow differentiating lab tests from genetic tests, for example family history considerations.

e. Mark: caution against requiring multiple categories from pooling data point of view

f. Bob M: one approach is to develop and maintain multiple version of the IG targeted for different versions of the spec (for users locked into R3 or R4, etc).

g. In general, profiles should be cautious about restricting cardinalities as you want people to be able to send the data they have (rather than have to build a custom interface for your systems)

2. For 23999, 23987, and 23997, how to say an instance validates against both mCODE profile and external (CG IG) profile?
   a. Having the same data elements represented consistently is important.
   b. We are concerned with adding elements to the spec that aren’t determined to be minimal for our use case.

3. mCODE alignment overview: (from http://standardhealthrecord.org/guides/mcode-r4/index.html#Genomics),

mCODE includes the minimal set of genetic related elements relevant to capture in an EHR to inform cancer assessment and treatment options. The approach is based on the HL7 CGWG Clinical Genomics Reporting Implementation Guide. However, mCODE simplifies genomics reporting to single discrete variants or to variants that were found in a given DNA region. Three profiles relate to the capture of clinical genomics data:

- **Genomics Report** - contain results of genomic analyses. Genomic reports vary in complexity and content, as simple as the results for a single discrete variant to complex sequences that are found in exome and genome-wide association studies (GWAS).

- **Genetic Variant Tested** - used to capture the results of a test for a single known variant.

- **Genetic Variant Found** - used to record variants that could be found from tests that broadly analyze genetic regions (e.g.: exome tests) and stores results for any variants that could have been found. If the implementer uses GeneticVariantFound, then the region in which the variant was found could be specified in the RegionStudied attribute of the GenomicsReport profile.

1. Genomics Reports
   a. Code: GeneticTestVS (extensible) vs LOINC 81247-9
   b. Category: Fixed Value: http://terminology.hl7.org/CodeSystem/v2-0074 GE vs 0..* (‘GE’ and possibly others)
   c. obf-SpecimenType-extension (available on referenced specimen in both profiles)
   d. onco-core-RegionStudied-extension (string) vs regionStudied result
   e. Subject = patient vs patient | group | location
   f. Missing extensions: RelatedArtifact, RecommendedAction, SupportingInfo, diagnosticReport-risk

2. Genetic Variant Tested vs Variant
   a. Obs-gene extention vs gene studied HGNC component
   b. onco-core-VariantTested-extension
1. VariantIdentifier: ClinVarVS (extensible) component:variation-code 81252-9
2. VariantHGVSName 0..* string vs component:dna-chg 48004-6
3. VariantDescription 0..1 string

c. 3. Genetic Variant Found vs Variant
   a. Code: 69548,
   b. value: none vs Present|Absent|No call|Indeterminate
   c. Method: GeneticTestMethodVS vs LOINC Answer List LL4048-6
   d. Components:
      i. Genomic source class, GenomicSourceClassVS
      ii. VariantFoundIdentifier: ClinVarVS
      iii. VariantFoundHGVSName: string

4. Major discussion of breakpoints between tumor markers and genetic tests, taking to Zulip

FHIR Subgroup Meeting October 21st, 2019

https://join.freeconferencecall.com/clingenomics
Quick link to this sign-in/notes document: tinyurl.com/fhirgenomics

Sign In: (presiding co-chair - Kevin Power)
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19.

Agenda (Standing items from WGM/recent discussions):
1. New templating
2. Capability Statements & test suites
3. New projects/sub-IGs
4. Revisiting MolecularSequence
5. RelatedArtifact extension
6. Alignment between genomic findings profile codes
7. Resolution of TBD codes
8. Alignment of implications
9. Condition vs Phenotype
10. Old trackers

Discussion:

1. New templating
   a. Idea: import our IG and use new tools for further updates
   b. Tooling provided as a Starting point, unsure if import/export is facilitated.
   c. Follow-up with Lloyd/Patrick to see what the workflow for import/adoption entails
   d. Here is the new 'template': https://github.com/FHIR/hl7-ig-template/tree/master
   e. Next steps: set up zulip stream to monitor (Jamie)

2. Capability Statements & test suites
   a. Can have multiple/overlapping capability statements
      i. Should look into formal/informal relationships between reporting use cases.
      ii. Grahame et al working on tooling to compare profiles from multiple sources and validate instances against multiple

3. New projects/sub-IGs
   a. Microbiology Genomics
      i. Mostly probe-based
   b. US Core Genomics
      i. Current IG is universal, can point to separate US IG or provide brief guidance on how to marry two IGs (validate an instance against 2 IGs)
      ii. Option 1: add alignment page
   c. GACS operations
      i. Bob D has requested time (November Tuesday) to go over defining some operations and a potential reference implementation
   d. Others? (Oncology/HLA?)
      i. Should consider including modules or separate IGs. Separate IGs creates additional dependencies
      ii. HLA should require gene-system, and gene family alongside HLA panel codes, identified multiple items that should be universal for HLA reporting
      iii. integration/alignment with mCODE eMERGE and Phenopackets
   e. Interaction with Information Modeling subgroup concepts
      i. Definitional resources in particular if possible (asserted allele for HLA, eg)
      ii. Include GA4GH variant representation alignment
   f. Next steps:
      i. Create a call for use cases to the listserv (and elsewhere)

4. Revisiting MolecularSequence
a. Overlap with Observation (make definitional?)
b. Ideal usage

5. Sending attachment files (ie VCF)

6. RelatedArtifact extension
   a. Apply to observation.component
   b. Extend typing/usage to text
   c. Next steps: need proposal

7. Alignment between genomic findings profile codes
   a. ‘Was it found?’ (LL1971-2) vs ‘what was found?’ (id/name)
   b. Could align either way
   c. mCODE created a separate profile for ‘VariantTested’ and ‘VariantFound’

8. Resolution of TBD codes
   a. List with basic definitions:
      http://build.fhir.org/ig/HL7/genomics-reporting/tbd-codes.html
   b. Next steps: Kevin sends info to Clem for full definitions and references where possible.

9. Alignment of implications
   a. https://chat.fhir.org/#narrow/stream/179197-genomics/topic/harmonize.20somatic.20and.20PGx.20profiles
   b. Needs a proposal - Bret will help track down who can make the best proposal here (might be himself)

10. Condition vs Phenotype
    a. https://chat.fhir.org/#narrow/stream/179197-genomics/topic/Modelling.20of.20'affect\ned'.20status
    b. https://chat.fhir.org/#narrow/stream/179197-genomics/topic/more.20examples
    c. Phenopackets???

11. Old trackers (44)
    a. Many against older guidance/resources
    b. Need recommendations/votes

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FHIR Subgroup Meeting October 7th, 2019

https://join.freeconferencecall.com/clingenomics
Quick link to this sign-in/notes document: tinyurl.com/fhirgenomics

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

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Agenda:

1. 3 little trackers
2. mCode alignment: aim for next week Oct 14th
3. Next steps (did not address today)
   a. Ballot reconciliation spreadsheet
   b. DevDays Amsterdam
   c. Splitting IG more formally for individual use cases
   d. Alignment of Findings profile codes
   e. Alignment of Implication profiles
   f. Press release for the IG

Discussion:

1. trackers in gForge:
   a. 24880 Split HGNC CodeSystem into two Codes
      Patrick working on it
      “binding is VS binding (extensible) and there are no patterns involved, so could just change the binding to HGNC genenames”
      Need to update entry on codesystems table for HGNC genegroup - currently copies HGNC gene
      (http://build.fhir.org/ig/HL7/genomics-reporting/codings.html)
      then can mark as applied.
   b. 19876 many examples do not validate against an IG profile using the FHIR Validator
      Final errors: http://build.fhir.org/ig/HL7/genomics-reporting/qa.html
      - Tx server (should be fixed/ok)
      - derivedFrom (workaround in place)
      o Tried/trying discriminator first by type, secondarily by code
c. Should seek Lloyd’s help to declare profiles in these slices properly or vote to remove sequence from the derivedFrom fields.
  
  ○ Jamie will attempt to test with removing sequence, see if errors persist and follow-up with Lloyd
  ○ Kevin: discriminator on the derivedFrom that can reference both other Observation profiles and MolecularSequenc tried:
    - resolve(), resolve().code.@pattern
    - And I get these errors on examples: Unable to resolve discriminator in definitions: code

<table>
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<tr>
<th>16510</th>
<th>Need more examples</th>
<th><a href="http://build.fhir.org/ig/HL7/genomics-reporting/artifacts.html">http://build.fhir.org/ig/HL7/genomics-reporting/artifacts.html</a></th>
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</thead>
<tbody>
<tr>
<td></td>
<td>build.fhir.org/ig/HL7/genomics-reporting/DiagnosticReport-AnnotationExample.xml</td>
<td>Update snomed code based on variant-specific/familial indication rather than disorder. Update name to ‘ACMG screening’</td>
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<tr>
<td></td>
<td>Examples are not showing up in example tab unless declared on the ig.xml as</td>
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<tr>
<td></td>
<td>&lt;reference&gt;</td>
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<td>&lt;/resource&gt;</td>
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<tr>
<td></td>
<td>High risk allele example: &quot;Fixed codeableconcept required” error</td>
<td></td>
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<tr>
<td></td>
<td>keep the associated phenotype</td>
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</tbody>
</table>

2. **mCode alignment: aim for next week Oct 14th**
3. Next steps:
   a. Ballot reconciliation spreadsheet
   b. DevDays Amsterdam
      i. Set up tools to create examples from sample reports,
      ii. provide sandbox for queries
   c. Splitting IG more formally for individual use cases
      i. Developing capability statements
      ii. Linking to external IGs?
   d. Alignment of “Findings” profile codes for next version
e. Alignment of “Implication” profiles for next version
f. Press release for the IG

Chat History
Jamie 11:02 AM
https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOpXHPr5tSLLQ/edit#

Kevin Power 11:07 AM
http://build.fhir.org/ig/HL7/genomics-reporting/valueset-hgnc.html

Kevin Power 11:36 AM
Here is how I am trying to define the discriminator on the derivedFrom that can reference both other Observation profiles and MolecularSequence
resolve(), resolve().code.@pattern

Kevin Power 11:56 AM
And I get these errors on examples: Unable to resolve discriminator in definitions: code

FHIR Subgroup Meeting September 30th, 2019
https://join.freeconferencecall.com/clingenomics

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

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19.
AGENDA:

1. Recap from WGM
2. “Resolved - change required” input requested
   a. Code systems table
   b. Glossary
   c. Query guidance
3. Final items to vote on for inclusion from connectathon/WGM
   a. 24685
   b. 24853
   c. 24879
   d. 24880

DISCUSSION:

1 From WGM:
Identified short list of items blocking publication, aiming to complete remaining items this week:
Items listed below and then spreadsheet.

2.

| 16876 | Use of publically available external coding systems and autocomplete lookup tables - 2018-May Genomics #58 | See [http://build.fhir.org/ig/HL7/genomics-reporting/codings.html](http://build.fhir.org/ig/HL7/genomics-reporting/codings.html) and [Work from before](http://work-from-before) and [Update from Liz](http://update-from-liz)
Adding “official system URI” which will be blank where systems are not already voted on/incorporated into HL7 - have to contact the organizations and allow them to provide system

Patrick will make final edits and Jamie will update on the page.

| 16513 | need glossary | See [http://build.fhir.org/ig/HL7/genomics-reporting/Glossary.html](http://build.fhir.org/ig/HL7/genomics-reporting/Glossary.html)
try to make the table more readable / explore freezing the header and/or styling better. Clem suggests asking HL7 for an updated style guide if it doesn’t exist.

Patrick: from IG creation workshop, we should be switching to the new templating mechanism for the next release, should
not invest too much effort in cleaning it up with the current machinery.

Jamie will go over headings for consistency, add table borders, and see about filling in any gaps (info not from the spreadsheet).

Liz wants us to use more references in the future!

See [http://build.fhir.org/ig/HL7/genomics-reporting/usecases.html](http://build.fhir.org/ig/HL7/genomics-reporting/usecases.html)

Remove 2 penultimate sentences of the first paragraph and attempt to cleanup headings for consistency.

Make sure each parameter used is mentioned in the text

Fix clinvar website

(Add examples queries from current examples)

### 3. Final items to vote on for inclusion from connectathon/WGM

<table>
<thead>
<tr>
<th>Tracker</th>
<th>Summary</th>
<th>Resolution Notes</th>
<th>Details</th>
<th>Discussion</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>15884</td>
<td>IG to provide specifications around search</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>24685</td>
<td><strong>Recommendation:</strong> persuasive with mod - remove all “must support” flags on profiles and add text to IG scope stating, “At this time, the work group has not yet come to a consensus on a single minimal set of elements that are required to be supported for generic use cases. In a future release, specific lab reporting use cases will have individual capability statements including must-support elements.”</td>
<td>Suggest removing all Must Support flags and explain meaning in text to ease adoption. Otherwise must define how derivation from our profiles is to be interpreted.</td>
<td>Discussed at WGM and on 9/24, proposal: remove all must support flags, replace with textual guidance.</td>
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<td></td>
<td><strong>Reasoning:</strong> at this time, ‘must support’ flags are inherited by profiles from external IGs extending our profiles, however the meanings are not. To ensure maximum reusability of our profiles, we must be clear that the flags on all the optional components do not get misinterpreted.</td>
<td></td>
<td></td>
<td>Proposal: remove all ‘must support flags’</td>
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</tbody>
</table>
Original scope of the guide was genomics reporting (hence the name) that use case makes sense to define “must supports.” Since then, the guide has also been used to define general sharing of genomic information. Ideally, the IG should be split into 2 pieces, 1 for defining structures and 1 for defining how they should be used by labs for reporting. Hard to do for the initial release.

The specific “sub” IGs would define capability statements and must-supports and must-support definitions.

Lloyd: not thrilled with just yanking all must-supports out of the IG right now.

Kevin: we either get rid if all must-supports or do it properly. As we don’t have time for doing it properly before publication the easiest would be delete them for now.

Option (for the future) is defining invariants for component groups that should be used together.

Peter: Must vs Should. Should would inform where we will tighten-up in the future.

Lloyd: if the WG does not have a clear definition in mind for Must-Support, then legitimate to remove. With some documentation that must-supports will be in future version.

Tracker

<table>
<thead>
<tr>
<th>Tracker</th>
<th>Summary</th>
<th>Notes</th>
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<tbody>
<tr>
<td>24853</td>
<td>need data element for Variant Inherited From,</td>
<td>Brett Heale</td>
</tr>
</tbody>
</table>

Persuasive: FINAL proposal is to Define a new component on the Variant profile.
* New code in CodeSystem/tdb-codes (LOINC code has been seperately request from LOINC by Brett Heale)
  code: variant-inheritance
display: Variant Inheritance
definition: Indicates if the variant was inherited from the patient’s mother or father.
  * New ValueSet/variant-inheritance
Value Set SO = Name: variant_origin (SO:0001762)SO:0001775 - maternal_variantSO:0001776 - paternal_variantSO:0001781 - de_novo_variantN/A - Unknown inheritance (needs to be added - not currently in SO)SO:0001778 - germline_variantSO:0001779 - pedigree_specific_variantSO:0001780 - population_specific_variant
The work of Nephi Walton, Clinical Geneticist at Geisenger, to map gnomic reports he is using into our IG, requires a data element for InheritanceOfVariant with values De Novo, Unknown, Father (Paternal), Mother (Maternal). The element is meant to indicate whether the variant was found in the patient's parent. This is typical reported in a TRIO analysis (where the patient's genome is compared to close relatives - typically mother and father). The reports generated from these analysis do not always provide supporting documentation. The value is simply stated as metadata that the Lab has regarding the variant.

My proposal is to create a new component in our IG before publishing to accommodate the element.

Looking in our IG, For sequence phase relationship(https://build.fhir.org/ig/HL7/genomics-reporting/obs-sequence-phase-reltn.html) we use the LOINC code for Allelic Phase (https://r.details.loinc.org/LOINC/82120-7.html) but with a required value set of cis, trans, indeterminate, unknown. This element is specifically for relating a variant observation with other variant observations. I do not think it applicable to Nephi's requirement as De Novo is not a relationship between variants.

The element provides a place for the lab to state whom the variation was inherited from, not the phase.

Full-term name: Variant Inheritance  
Shorthand name: InheritanceOfVariant  
Definition: Weather or not the variant was inherited from the patient's mother or father.  
Value Set: De Novo, Unknown, Maternal, Paternal  
I think a component would be best as this is a property of the variant and has no meaning without being a part of an observation of a variant.

---

### Follow-ups

- **Mon, 23 Sep 2019** - by Bret Heale-Zulip:  
  https://chat.fhir.org/#narrow/stream/179197-genomics/topic/variant.20Inherited  
- **Tue, 24 Sep 2019** - by Bret Heale:  
  FOR component.code there does not appear to be a specific existing LOINC code. I sent a request yesterday to LOINC for a code. I would recommend that for component.code we use the TBD-LOINC construct we have used elsewhere so that we can publish.
- **Wed, 25 Sep 2019** - by Kevin Power:  
  Add proposed resolution, and moved to ready for vote.

### Discussion

**Tracker**

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**24879** Relax genomics-base.interpretation to 0..0,

**Resolution**

*Persuasive, add text on the reasoning to use other profiles*

**Notes**

| Details | This can prevent implementers from inheriting from our IG. -&gt; relax card. to 0..*Make another tracker item to add a warning/error invariant if interpretation is used in an instance. |

**Disposition**

*Recommendation: Similar arguments for the must-supports. Add text & warning/best practice flag to suggest using our more complex implication profiles for interpretations rather than the built-in observation and observation.component.interpretation*

**Tracker**

<table>
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**24880** Split HGNC CodeSystem into two Codesy,
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<th>Notes</th>
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<tr>
<td>N/A</td>
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**Details**

HGNC uses overlapping numeric IDs for Genes and GeneGroups, a gene ID uses the prefix HGNC:, a gene family is only a number.

To prevent mixups between gene IDs and Gene Family IDs two new CS URIs should be created:

- `http://www.genenames.org/genegroup`
- `http://www.genenames.org/geneld`

- The pattern for "gene studied ID" has to be updated to allow all three SystemURIs- Invariant which gives a warning on `http://www.genenames.org` as the system uri.

**Previous notes:**

[https://docs.google.com/document/d/1LMKl0lqaPbJxO1-PS1YK5vxsNcDqt8LQMA8NZ0wKQ8/edit#](https://docs.google.com/document/d/1LMKl0lqaPbJxO1-PS1YK5vxsNcDqt8LQMA8NZ0wKQ8/edit#)