Monday

Q2
- Joint with EHR

Q4
- Attendees
  - Bob Milius - NMDP - bmilius@nmdp.org
  - Terry McDonnell - Syapse - terrym@syapse.com
  - Kevin Power - Cerner - kpower@cerner.com
  - Richard Ettema - AEGIS.net, Inc. - richard.ettema@aegis.net
  - Joseph Kane - Epic - jkane@epic.com
  - Joel Schneider - NMDP - jschneid@nmdp.org
  - JD Nolen - Cerner - johndavid.nolen@cerner.com
  - Bob Wildin - NHGRI - bob.wildin@nih.gov
  - Christopher Chute - Johns Hopkins - Chute@jhu.edu
  - Gil Alterovitz - HMS/BCH gilusa@gmail.com

- Presiding co-chair - Gil
- FHIR Subgroup
- Connectathon review
- Gill presenting slides (will share ?)
- FHIR Genomics Track - 8 use cases
- Derived from DAM
- Activities
  - Bob M - showed the transaction bundle and described lessons learned
    - Will write up summary
    - Bob W - if different servers produce different results, is that a problem
    - Richard - yes
  - Gil - results
    - Increased knowledge around the genomic specs
    - Set up tests for validation beyond validation of operations (e.g. rules for validating required fields, etc)
    - Tested genomic compliance across FHIR servers
    - Saw differences in parameter search query validation in servers
      - Health Intersections vs HAPI
  - Gil - survey results
    - Users are more knowledgeable and capable
  - Gil - going forward (input from participants)
    - Continue with IG
    - Review cardinality of sequence elements
    - Move HLA extension from Diagnostic Report to observation
    - Add identifier to sequence
    - Discuss consistency in parameter search validation across servers
  - Gil - walk through IG comments
    - See comments in document
    - https://docs.google.com/document/d/1JhXtTbp5fp_aR5HIckAEbD9yGiGkBQxor8_lOy_SW8/edit#heading=h.ff4r0dknw1f4
Kevin - PGx Example

- TMPT Gene (part of the DIGitize IG)
- Want to talk through example of when a TPMT allele (like TMPT *3A, highlighted in Blue below)
  - Made up of two SNPs (highlighted in Red below)
  - Two SNP calls (Purple)
  - How to best model this?
- Today, would need to be two Sequence resources, but if we make Variation 0..*, could be done as a single resource
- Two Sequence resources is OK, but gets weird if you choose to send the observedSequence for the entire Gene - in which case, the implementer must “arbitrarily” break up the sequence and link the Sequence resources together.
  - Perhaps in this case, we recommend not sending the entire sequence?

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</tbody>
</table>

TQ1

Attendees

- Bob Milius - NMDP - bmilius@nmdp.org
- Kevin Power - Cerner - kpower@cerner.com
- Michael Lawley - CSIRO - michael.lawley@csiro.au
- Terry McDonnell - Syapse - terrym@syapse.com
- Andrea Pitkus, IMO - apitkus@imo-online.com
- Joseph Kane - Epic - jkane@epic.com
- Mollie Ullman-Cullere - BOC - mollie@betteroutcomes.com
- Jim Shima - Syapse - jamess@syapse.com
- Bob Wildin - NHGRI - bob.wildin@nih.gov
- Elizabeth Newton - Kaiser Permanente - elizabeth.h.newton@kp.org
- Sun-Ju Ahn - Korean Agency for Technology and Standards - april0149@gmail.com
- MariBeth Gagnon - mgagnon@cdc.gov
- Harold Solbrig - solbrig.harold@mayo.edu
- Byoung-Kee Yi - SMC - byoungkeeyi@gmail.com
- Nephi Walton - Washington University - walton_n@kids.wustl.edu
- Joel Schneider - NMDP - jschneid@nmdp.org

Presiding co-chair – Bob M

Introductions

Review Agenda

Review of Clinical Genomics activities for newcomers

- Andrea agreed to reach out to Jim Case of NLM/IHTSDO (SNOMED CT) and Clem of NLM, to ask if Clinical Genomics LOINC answer lists can be built in SNOMED CT to support alignment of Clinical Genomics implementations with the US Realm Lab Implementation Guides cited by ONC and with vendor certification and functionality requirements. (email sent.)

- Nephi - wants to integrate their cerner system with other systems; getting VCF and BAM files from their labs;
  - Mollie - connect with Stan Huff, Clem McDonald
  - Kevin P - V2 lite is better for near term
  - Mollie - V2 lite is based on V2, extends to genomic coordinates
  - Mollie - Variant Effect Predictor (VEP) can translate VCF to HGVS with high quality, supporting RefSeq, ENSEMBL, left and right alignment of Indels; comes from Ensembl; can operate as standalone tool if desired; Free tool

- Bob W - in the pedigree standard, can we record genomic info in it?
  - Mollie - can record genetic information, but done a while ago, so may be as current as it should be.; work in progress
  - Nephi - tried it but doesn’t support some features like negation
  - Kevin P - FHIR has Family History Member profile for genetics; can be linked to genomic resources/profiles

- Kevin - negation modeling - what is scope?
  - Bob M - technology agnostic model

Review of external efforts
Bob W - sits on round table of genomics and precision health; action collaborative now call DIGITizE

http://www.nationalacademies.org/hmd/Activities/Research/GenomicBasedResearch/Innovation-Collaboratives/EHR.aspx

Current IG:

Kevin - proposal to develop implementation guide for FHIR (currently has V2 IG, want a similar one for FHIR)

DIGITizE isn’t currently focused on variant (DNA variant). Bob W.: Uses LOINC codes (new if required), for example 50956-2 HLA-b*57:01 for the typing result and then SNOMED-CT codes for “Positive” and “Negative”, and for TPMT, metabolizer status codes.

Bob W - want it to support CDS

Mollie - should CG help work on it?

Kevin - already working with Clem for LOINC codes

Mollie - we need to update V2 IG, add some codes for release 3 and use IOM to inform

Other?

Review and Planning of Roadmap for CG Workgroup

Maybe this should be done at end of meeting, in light of everything we’ve learned?

To dos:

FHIRE

Gil, Mollie, Jonathon, etc facilitor

subgroup

V2 -

we need to update V2 IG, add some codes for release 3 and use IOM to inform

DAM

Gil & Mollie

DIM

Amnon

Subgroup - led by Amnon & Bob Freimuth

Family History

Normative Standard

Also ANSI standard (when does it need to renewed?)

My Family Health using it

HITSP

Actively used and adopted

Was renewed
Bob W.: Grant Wood will lead the discussion about standards and the June NIH/NHGRI Family Health History Tools conference.

Kevin: Intermountain / UMass (Kevin Hughes) / Duke looking at how well the FHIR FamilyMemberHistory + FMH Genetics Profile maps to the V3 based standard. IM created sample FHIR bundle that is being compared to V3 standard to identify gaps. More of an update Wed in other discussions about pilots.

- IG - published
- Pedigree model
- FHIR profile of Family Member History

Business

- Find renewal deadlines for
  - SWOT
  - Mission
  - Charter
  - DMP
  - Old projects
Attendees

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○ Joel Schneider - NMDP - jschneid@nmdp.org
○ Jim Shima - Syapse - jamess@syapse.com
○ Harold Solbrig - Mayo - solbrig.harold@mayo.edu
○ Kevin Power - Cerner - kpower@cerner.com
○ Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
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○ Nephi Walton - Washington University - walton_n@kids.wustl.edu
○ JD Nolen - Cerner - john david.nolen@cerner.com

Presiding co-chair - Mollie

DAM

○ <please add link to slides>
○ Slides will be uploaded to document center
○ Mollie - project history
○ Background information
○ Stakeholders
○ Help IT environment
  ○ Understand needs for genetics
  ○ Understand domains other than their own
○ Specimen identification use case: informed OO’s specimen DAM
  ○ Germline testing
  ○ Tumor testing
  ○ Pediatric testing
  ○ Prenatal testing
  ○ Infectious disease testing
  ○ Emerging specimen scenarios
○ Germline Testing Clinical Workflow
  ○ Figure showing swimlanes
  ○ Chips are 1st line
  ○ Bob W: Chips are not common except in expression; a lot of sequencing done
  ○ Nephi - NGS sequencing driving the cost down
○ Alternate Clinical Flows
  ○ Germline testing continued
  ○ Variants of unknown significance -
  ○ Bob W - vocabulary question of Geneticist vs Medical Geneticist vs Molecular Pathologist ;
○ Enhance for separation between lab interpretation and medical genetic interpretation with patient
○ Medical geneticist is seeing the patient
○ Lab may be additional information but is not seeing patient
○ Nephi - needs to have a way to reclassify variants in workflow
  ■ We do a manual literature review for every variant of unknown significance
  ■ Mollie - is this a chart review?
  ■ Nephi - start with that, but then go into literature review
  ■ JD - does it then go into chart?
  ■ Nephi - document it
○ Veritas - ideal approach
  ■ Whole genome $1000
  ■ Informed of relevant variants at resulting
  ■ Informed of new updates when reclassified
○ JD - is there a “thing” that can assign meaning to a lab test
  ■ Managing biomarker?
  ■ Mollie - biomarkers should be managed by NCBI; star alleles
  ■ PharmGKB
● Somatic/Cancer Testing Clinical Flow
○ Alternative flows
  ■ Specimen
    ○ Diagram shows matched germline specimen
    ○ Alternative specimen clinical flows include using
      ■ Filter-out know germline variant filters
      ■ Filter-in known somatic variants
  ■ consent/test order
    ○ Hospital protocols trigger genetic testing based on pathologic Diagnosis similar to other molecular tests
    ○ Pathologist more active
  ■ Reinterpretation often found with follow-up testing
  ■ Bob W - need to study non-cancer germline mutations (eg mosaics)
  ■ Transplant recipients -> mosaic
  ■ JD - develop layers, genomics first, transcripts on top of that
  ■ Nephi - how to represent phenotypic information
● Decision Making Tools
○ Family history
● Public Health Reporting
○ Alternate flows
○ Additional data flow into public health reporting
○ Labs send data directly to Cancer Registry
○ Gene DX
○ Match Maker Exchange - VUS to match patients like me
  ■ Gene Match is part of Match Maker group
  ■ Bob W: Matchmaker exchange: http://www.matchmakerexchange.org/ “matching of cases with similar phenotypic and genotypic profiles (matchmaking)
through standardized application programming interfaces (APIs) and procedural conventions.”

- Bob W: GeneMatcher: [https://genematcher.org/](https://genematcher.org/)
- NHGRI consortium
  - Infectious disease related genetic reporting to Public Health
    - Zika - in fetus, circulating in mother, etc
    - Nephi - we are doing genotyping for bacteria right now, but is a yes/no answer, not sending sequence; sequencing to classify
  - Clinical and Research Data Warehouses
  - Alternate flows
  - Additional data flow into research
- Challenges across different testing platforms
- Testing platforms and variant detection Ontology
- Additional sections
  - HLA
  - Additional use cases
    - Family history standards
    - Clinical Genetic/Genomic Standards
    - Clinical Genomics FHIR
  - Vocabularies - external references
  - Vocabulary - constraints
- Nephi - geneticist should be like an MRI; there’s a report used by a clinician, but the radiologist want to see the image; medical geneticist should have same kind of access to sequence.
- Actionable today - ehr report
- Full data in data warehouse (BobW: probably not the bam files but variant api, with various levels of filtering, perhaps based on ClinGen/ClinVar evidence/assertion classification levels)
- Medical geneticist wants to look at whole genetic data themselves (through a bioinformatics layer surfacing the more meaningful - and threshold filters to go deeper) after they’ve seen the patient and didn’t find the answer in the clinical report.
- This models what clinician does in reviewing an MRI - read the report and look at the image in increasing detail for further investigation if needed - go into a genomic version of the PACS system with appropriate viewers
Attendees

- Clem McDonald - NLM/Lister Hill Center for Biomedical Computing - clemmcdonald@mail.nih.gov
- Joseph Kane - Epic - jkane@epic.com
- MariBeth Gagnon - mgagnon@cdc.gov
- Jim Shima - Syapse - jamess@syapse.com
- Nephi Walton - Washington University - walton_n@kids.wustl.edu
- Kevin Power - Cerner - kpower@cerner.com
- Mollie Ullman - BOC - mollie@betteroutcomes.com
- Lauren Becnel - CDISC - lbecnel@cdisc.org
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- Elizabeth Newton - Kaiser Permanente - elizabeth.h.newton@kp.org

Presiding co-chair Mollie

Introductions

V2 - Discussion led by Clem

PDF files sent to listserv

- 2016-05-09 -5PM OutlineGenomicModel for HL7 Genetics report
- 20160508_HL7 genetic variant reporting panel_custom_w_descriptions_and_answer_lists

Displaying Clinical Table Search Service

- iforms-service.nlm.nih.gov

Simple variations

Since last mtg learned more about COSMIC

Coding systems Recently got COSMIC data - type gene symbol in the COSMIC field

LOINC for clinical genomics start here: https://lforms-service.nlm.nih.gov/

- Select “LOINC questions and forms” at right; then select “by the LForms widget.”;
  Then select second item in left menu "HL7 Genetic Test Panel for Variants - 20160322 (table version)"

- Can click “Add another 'Simple variant - panel’” to add another simple variant.
  Creating a record with more than one variant identified.
- Click ‘show HL7 message’ to see the genetic data within an HL7 V2 message


Where to put in start-end?

- Genomic allele location
- Does’t currently include stop
- Allelic state
  - Should this be here?
  - Need real evidence that they are cis or trans (haplotype or not)
- How does someone report a same as reference?
  - Genetic Analysis Overall Interpretation
    - Positive, Negative, Inconclusive, Failure
- Want to report the “negatives”
- Dan Rutz & Hans - OO are collaborating to make this happen
  - LRI is the approved lab reporting message - v2 lite to point to this message
  - Repeating OBX’s to handle variants

Aside
Similar tools for the creation of
TQ4 (concurrent with joint with Patient Care)

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- Bob Milius - NMDP - bmilius@nmdp.org
- Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
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- Joel Schneider - NMDP - jschneid@nmdp.org
- Elizabeth Newton - Kaiser Permanente - elizabeth.h.newton@kp.org

Presiding co-chair Bob
Continued discussions
- Mollie - strategy for preparing V2 lite doc for ballot
  - Clem will work with OO, CG,
  - Ownership change to CG; CG and OO voted to change; we need to do the paperwork
  - Need to ballot for Sept
  - Notice to ballot in June
- Clem - Prefers to have it passed, but as long as we have good comments
- V2 2.51 still needs to be maintained
- Clem presenting iforms-demo.nlm.nih.gov
  - Structural (copy number) variants section
  - Structural variant name -
    - Can get a name from cosmic
  - Fuzzy boundaries
    - Structural variant outer start-end
    - Structural variant inner start-end
  - Looked at dataset from cosmic - seems to be using something similar to hgvs, but different
- GRCh38/hg38 17p12(chr17:14186983-15563870)x3
  - Expecting 2 (two chromosomes), but found 3
- Evolved from FISH technologies
- ISCN expression - this is in clinical use
  - Start with ISCN, leave HGVS free
- Example of CNV:
    (See Table A)
- aCGH ratio
- Methods
- Dinner plans?
● Crudessence
Wednesday -

- Attendees
  - Bob Milius - NMDP - bmilius@nmdp.org
  - Joseph Kane - Epic - jkane@epic.com
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  - Mollie Ullman-Cullere - BOC - mollie@betteroutcomes.com
  - Clem McDonald - NLM/Lister Hill Center for Biomedical Computing - clemmcdonald@mail.nih.gov
  - Elizabeth Newton - Kaiser Permanente - elizabeth.h.newton@kp.org
  - Grant Wood - Intermountain -

- Presiding co-chair - Gil
- Introductions
- FHIR

- Gil - current state of FHIR
  - Connectathon
  - Need for an implementation guide (see link for draft below)
    - IG Draft:
      - [http://tinyurl.com/hd8hy29](http://tinyurl.com/hd8hy29)
    - Includes mappings from DSTU 2 to STU 3
    - How to deal with many variations with a single sequence
      - e.g., haplotype of snps in a gene
    - Bob M - HML use case for many variants per sequence
  - pilots including family history (1 hour for all pilots)
    - Gill - showed use case BCH_FHriskBRCA2_v2(1).docx, will be posted to the Clinician Connectathon wiki
    - Grant
      - Intermountain is mapping their systems to FHIR
      - Pilot - get data from Intermountain db to FHIR, then translate to V3, then send it to Kevin Hughes service (V3 is required by that server)
      - Pilot with Kevin Power (Cerner) - Intermountain working to send data via FHIR to cerner product.
      - Mollie - FHIR has questionnaire, we could inform them
      - Bob M - Structure Data Capture (SDC) has a project to do this sort of thing, we should inform them
      - Grant - pilots are the way to move forward
      - Mollie - need to build to questions
      - Kevin P - questions need to be precise, “my aunt” vs “my maternal aunt”
Grant - Genetic counselors don’t use forms, they ask questions; they build the pedigrees themselves

Amnon - should use IDs instead of identifiers for algorithms for individuals

Kevin P - that works if everyone is in the same system

Grant - put in full uri to external patients

JD - maybe, consent issues will have to be addressed

Grant - another external driver for creating tools and standards is the Precision Medicine Initiative

Bob W - NHGRI/NIH - June 14 & 15 conference, tools like My Family Health Portrait, etc, invited vendors (none responded)

Gil - Genomics Pilots

Pharmacogenomic clinic: Precision link
  - 1st exclusively Pharmacogenomic clinic in the world

Precision Medicine for global health: TBResist
  - Combines clinical genomic data from over 20 countries for diagnostics and therapeutics for drug resistant tuberculosis to enable targeted patient approaches.

Precision Medicine Cloud Computing: DNA Nexus/PrecisionFDA
  - Set up the precisionFDA portal and working on setting up FHIR Genomics server/apps on their platform

coordinate systems?

prep for joint with FHIR
  - See agenda for Q4
Joint with OO, AP, II - hosted by OO

Attendees from CG
- Bob Milius
- Mollie
- Gil
- JD Nolen
- Andrea Pitkus, IMO- apitkus@imo-online.com
- Many from other work groups

Solo meeting (Presiding co-chair - Amnon)

Attendees
- Joseph Kane - Epic - jkane@epic.com
- Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
- Bob Freimuth
- Grant Wood
- Joel Schneider - NMDP - jschneid@nmdp.org
- Nephi Walton - Washington University - walton_n@kids.utah.edu

Minutes
- continued discussions
- Amnon Reviewed the history of the DAM/DIM efforts
- Bob F. reviewed the goals of the current IM subgroup
  - We are not going to redo anything that has been done and is done in the renewed DAM effort (transitioning from the previous Clinical Sequencing DAM)
  - Related efforts, focused on how to represent variants: ClinGen, GA4GH, and these efforts should be represented in this subgroup, so that all models could be aligned and preferably harmonized
  - We should take into account as inputs to this efforts previous efforts in HL7
  - It's a conceptual model capturing the semantics of the relationships between the entities we are dealing with
- Amnon:
  - Modeling tooling - how can we collaborate and align the model with other models
  - Ideally we'd like to have a DIM that can be aligned with concrete models like FHIR, v2 and CDA in an automated fashion but that's is not feasible
- Bob F.
  - In Mayo there is an effort to develop FHIR-like models by using their plugin to Power Designer (a modeling/development tool) to bridge between the conceptual modeling and create FHIR-like APIs for custom enterprise applications
  - Practically speaking - best choice is probably to follow a centralized-modeling approach (single model owner), with documentation/images circulated to the group for suggesting
edits/alternatives (XML might be able to be imported into various modeling tools)

○ Joel
  ■ Would like to contribute to the modeling aspect and less to the genomics behind it

○ Bob F.
  ■ The success criterion is mainly supporting the DAM use cases, as well as the main concepts from other artifacts the CG WG has produced
  ■ Develop atomic structures that could be grouped in HL7 specs, like a FHIR resource or profile

○ Joseph
  ■ There is a need in the short term to exchange genetic data
  ■ We in Epic get almost every week such a request to exchange genetic data in a more structured way than PDF

○ Amnon:
  ■ Discussed the interplays between the modeling efforts:

```
DAM

CG Information Modeling (IM)

DIM(s)

FHIR  CDA  v2  Others

Profiles  Templates  IGs
```
Attendees

- Bob Freimuth - Mayo
- JD Nolen - Cerner - johndavid.nolen@cerner.com
- Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
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- Mollie Ullman-Cullere - BOC - mollie@betteroutcomes.com
- Terry McDonnell - Syapse - terrym@syapse.com

Presiding co-chair - Amnon

Update - Joint OO

- Bob M gave an update on the Joint

Bob W - tree vs flat

- Tree representation is alleged to be more complex to implement than flat (but liked) representation in data file. GIVEN that they are logically identical, implementation uptake is likely greater for the easier process.

- Bob M’s HLA use case has an atomic value of HLA gene sequence, with all its variants, with a changing-knowledge-based interpretation label of HLA allele assignment. In addition, including the sequence is critical for his use case because the discovery of new alleles by sub-grouping similar sequences within the same current haplotype to define, in the future, additional sub-haplotypes with transplant clinical significance.

- Nephi - this is not different from other use cases where sequence data needs to be part of the transmitted data.

- This is a categorically different use case from listing multiple variants on the same gene (or same sequence, e.g. an entire chromosome) based on a primary clinical indication, even when phase is known.

- Bob M feels it is simpler to reference the ref sequence once, list the variants as children (tree). Bob W, not previously appreciating the depth of the difference between the use cases, had “channeled” Clem to suggest that a flat linked list of variants was, though logically equivalent, more likely to be implemented by avoiding deeper hierarchical representations. Bob M identified guidelines in the HLA field that suggest the tree approach is more recognizable and therefore easier to implement.

- Base Genetic observation already exists in the existing specification in FHIR per Mollie.

- Amnon made a few modeling comments:
  - A relation of 0..* is not constituting a hierarchy
  - Such relations are very common in most FHIR base resources
  - In particular for the Sequence resource, if we go with the current Sequence structure, then it’s you need linked list to add up to the sequence you’re dealing with, and a linked list is a complex structure,
especially that in the Sequence resource you need to use the window element in order to make the linked list coherent

- Group came to the conclusion that clinical disease risk and tumor testing and HLA testing are different. The first examines the sequence for known or unknown biomarkers (variants of potential clinical significance in a gene) and report these (not polymorphisms or known benign variants), while HLA and CYP family of genes is haplotype testing. In haplotype testing, the lab is working to identify which alleles of a gene the patient possesses.
  - Mollie commented that this is supported in FHIR and v2 currently.
  - Bob wanted a mechanism to include the gene sequence for the patient, which might be done in the sequence resource or through encapsulation of a VCF (or HLA constrained VCF) as an attachment.

- **DIM discussion**
  - DAM
  - DIMs
  - FHIR, CDA, V2, Others
  - Profiles, Templates, IGs

- **GTR roadmap**
  - CDA challenged because it is expired. Discussion.
  - Question posed: Would anybody miss CDA spec? Syapse - they won’t develop to it.
Attendees

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- Nephi Walton - Washington University - walton_n@kids.wustl.edu
- JD Nolen - Cerner - johndavid.nolen@cerner.com
- Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
- Bob Freimuth - Mayo
- Lloyd McKenzie - lmckenzie@gevityinc.com
- Tim Blake - tim@semanticconsulting.com.au
- Harold Solbrig - solbrig.harold@mayo.edu
- Terry McDonnell - Syapse - terrym@syapse.com
- Christopher Chute - Johns Hopkins - Chute@jhu.edu
- Josh Mandel
- Bob Wildin - NHGRI-NIH - bob.wildin@nih.gov
- Gil Alterovitz - HMS/BCH - gilusa@gmail.com
- Daniel Vreeman - Regenstrief - dvreeman@regenstrief.org
- Swapna Abhyankar - Regenstrief - sabhyank@regenstrief.org

Presiding co-chair - Gil

FHIR joint meeting (CG is hosting FHIR)

Review deadlines for STU3

- Targets needed by R Q3:
  - if planning to propose any new resources or IG’s for the next ballot that
don’t already have proposals for.
  - Sequence resource and family member hx and observation profiles–
what the maturity metric is on those. targets.
  - Maturity levels for profiles for STU3. Draft set of criteria. Final end of
this month. Target before Q4 tomorrow. Info needed to meet that if
ok.
  - Production - for clinical use. Can be pilot. Using it.
- Bob F- Level 3 question. Reviewing the defns. Don’t have to be reviewed outside
the workgroup.
- Deadline review.
  - R Q3 deadline- what the plans are and any concerns
  - June 1- if planning IG with no proposal done before or connectathon,
need it then. Feedback on gforge.
  - July 17- all substantive changes done on resources (enables profilers to
work with that).
- July 24 - freeze
- Would like to publish by end of the year

- **Connectathon Review**
  - Implementation guide- touchstone vs implementation guide for test scripts
  - Over 3 weeks, major changes. Can use old mechanism and plan to edit for new mechanism. Or can wait for 3+ weeks and apply changes after new tooling.
  - Transaction and references
    - May access resource all by itself. So, need have links to enable it.
    - Can query in opposite direction.
    - Direction- set by the resource that is created second- so it can refer to the first one. Patient is first. So, Observation points to Pat.
  - Parameter validations- should ignore search params not recognized. Shall provide info on what recognized and did. Onus on client to make sure all search params it passed that it actually paid attention to- and if not, to filter it.
    - Subject references. References in bundle resolved when transactions applied.
  - Bob M question on readable narrative being required. Answer: English narrative is not mandatory in spec, but strongly encouraged in most resources

- **FHIR and family history**
  - Age field as approx checkbox (Bob W) (Note: age range)
    - Response: thoughts- extension / modifier. Can’t put modifier type in simple datatype like integer. Can have string to write age (but not computable). Add exact age field?
    - Age approximate: Flag that is true/false if age is entered. Always must be asserted.
    - At bottom of fhir pages, have change request link.
  - How to do patient questionnaires (e.g. with SDC) (Mollie)
    - Should be ready soon or now. URL:
  - Combining inconsistent family hx’s (e.g. brother has different hx) (Bob W)
    - Provenance resource- say inputs to the process, I looked at x,y,z and talked to a,b,c- and as output, I came up with this… can point to different versions and make new version.
  - Bring up topic of consent models (GA4GH example) for FamilyHx- how to attach consent decision with famhx
    - Consent could point to famhx in general or specific features.
    - Attach to disclosure of famhx- extension on famhx- this famhx is covered on this consent.
    - Msg with consent and fam hx enclosed.
      - Post via rest- may be hard to combine things. Post fam hx and consent on file- or post both separately.
    - Consent resource? Modelled as profile on contract. Specific consent resource?
    - Research consent? Resource may handle all consents. CBCC

- **FHIR and vocabulary**
  - Bob M question.
  - Adding coding systems- add for HLA (terminology?). Bottom of page to change- gforge tracker- going to vocab. Follow-up? All workgroups are monitoring their own change requests.
- Bob M will contact vocab co-chairs re status of change request.

### FHIR and genomics

- Cardinality for variation/gene features: bundles and profile.
  - Compare my gene to my reference. May be a variation at beginning and one at the end. 2 variations to a reference. Right now, only one variation cardinality.
  - For a reference, have a possibility of 0 to many.
  - Want to indep pass one variation without the other? Or want to point to one variation specifically from observation?
  - Send as zipped binary?
  - Within resources can be included in the summary or not. What should be included? Not expect sequence full string in the summary view. Summary thing needed for level 3.
  - Hard to retrieve long base strings?
  - Solution in guide on how to do it. Btw, when want to send whole genome, this is how to do this.
  - Chris Chute - why not do it like VCF? one sequence and list variants one right after another after it?
    - Bob M - that's what I want to do. The current sequence specification doesn't allow for that. It currently allows for only one variant per sequence.

- If value in sending around, add the choice to add in string or by reference. 1m characters. Strings limited. If really big, make it a binary and zip and send.
Thursday

- ThQ1

Attendees

- Bob Milius - NMDP - bmilius@nmdp.org
- Joseph Kane - Epic - jkane@epic.com
- Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com
- Terry McDonnell - Syapse - terrym@syapse.com
- Elizabeth Newton - Kaiser Permanente - elizabeth.h.newton@kp.org
- Grant Wood - Intermountain - grant.wood@imail.org
- Joel Schneider - NMDP - jschneid@nmdp.org

Presiding co-chair – Bob M

Co-Chair elections

- Amnon re-elected
- Bob’s term up for election in Sept
  - Fri 6/24 - Deadline to notify HQ of additions/changes/corrections to co-chair openings
  - Wed 6/29 - Call for nominations
  - Fri 7/29 - Nominations close at 5:00 p.m. ET
  - Fri 8/12 - Co-chair statements due by 5:00 p.m. ET
  - Wed 8/17 - Co-chair statements e-mailed to membership

FHIR Deadlines (from Lloyd’s slides):

- Thur. Q3 - but we have extension!
  - What new resources/profiles do you plan to propose?
    - In current DSTU2
      - Two Profiles/Extensions
      - Family member history for genetics analysis
      - Standard Profile for Genetics
    - New
      - Sequence Resource
      - Profiles & Extensions
        - Observation -> Observation for Genetics
        - Diagnostic Order -> Diagnostic Order for Genetics
        - Diagnostic Report -> Diagnostic Report for Genetics
        - Family Member History -> Family member history for genetics analysis
        - Sequence -> Profile for Consensus Sequence Block
        - Diagnostic Report -> Profile for HLA Genotyping Results
**Questions**

- What are differences between Observation profiles
  - Standard Observation for Genetics
  - Observation for Genetics
  - What’s in Sequence - did something for Standard Profile for Genetics get moved into there? Or is this just the sequence?

- Can a profile be in the current build but not in the STU3? E.g., Profile for Consensus Sequence Block and Profile for HLA Genotyping Results

- What are the differences between DSTU2 Family History for Genetics and in the current build?

- Need to have this summarized for the main group. Doodle poll for quick review for meeting extension deadline.

- Mollie - Like NIB; let’s propose to do them all, but affirm later; Amnon and Mollie agree that there is precedence for this - keeping work moving and knowing HL7 processes provide multiple opportunities for transparency and voice

**What are your FMM targets for STU 3?**

- Currently everything is at level 0. Are we ready to target level 1?
  - Mollie - these artifacts have been around for a long time, and look stable. At this point it’s comparable to v2 and should be level 1
  - Terry - things are moving very quickly and we’re still proposing changes. Should stay at level 0
  - Elizabeth - Just because it’s been at a connectathon doesn’t mean it’s substantially complete
  - Bob - level 2 states that at least 80% of the core data elements have been transmitted. Possibly near level 2 already

- Bob M - Who has actually used these resources/profiles to exchange data? Another doodle polls for connectathon use cases?

- Any issues/concerns?
- When are actual FMM decided (in contrast to target)?
● FHIR Maturity Model (FMM)

● June 1
  ○ All resource & IG proposals for STU 3 have been completed, reviewed by WG and submitted
    ■ Promised as part of PSS
    ■ Part of ballot?
  ○ Connectathon tracks for Sept. have been proposed
  ○ Feedback on gForge submitted to FMG
    ■ Looking alternatives to gForge (e.g., jira)

● Sunday July 17
  ○ Substantive content freeze for ballot – core resources
    ■ What about profiles? 24th?

● Sunday July 24
  ○ Total content freeze, start of QA
    ■ Are FMM’s frozen here too?

● Wed Aug 10 (midnight)
  ○ All QA changes applied

● Fri. Aug 12
  ○ FHIR ballot opens

● Mon Sept. 12
  ○ FHIR ballot closes

● Fri Sept. 16
  ○ FHIR triage complete and ballot content loaded to gForge (or alternative)
    ■ Will comments be consolidated for duplicate issues?
    ■ Depending on number of comments, may want to do a first a pass of grouping comments together.

● Sept. 17-23
  ○ Baltimore WGM

● Sun. Dec. 11
  ○ Reconciliation complete/substantive changes applied?
  ○ (Just over 10 weeks)
  ○ Will re-evaluate at Baltimore based on volume of ballot comment
Bob - What’s happening in Baltimore? Is CG involved or a FMG meeting.

Amnon - Is there a notion of “passing” the ballot?

- Dec. 31
  - Publish?

Must submit PSS to PMO and Steering Division - May 22, 2016
  - Any new PSS’s?
    - Probably no, but clarify with Dave Hamill - v2 classic process

Ballots:
  - Submit Notice of Intent to Ballot - July 3, 2016
  - Need NIBs for V2 Lite and V2 Classic
  - Link to Notice of Intent to Ballot (NIB)

Projects over 5 years
  - If we have projects over 5 years, we need to review and update
    - we can update Project Insight (?) or notify PMO with new milestone dates
    - we can close projects
  - Need to get list of projects
  - e.g., GTR expired
    - Mollie - Retired but available?
  - V2? Clinical sequencing DAM? Active - update status
  - Bob will make a list of all
  - Family History ANSI Standard
    - Need to reaffirm with ANSI as a normative standard within year from now

Questionnaire
  - Refers to HHS’s tool My Family Health Portrait, which uses family history v3 HL7 standard. This example encodes these with LOINC (not currently used in V3)
  - Structured Data Capture
    - [http://hl7-fhir.github.io/sdc/questionnaireresponse-sdc-example-ussg-fht-answers.json.html](http://hl7-fhir.github.io/sdc/questionnaireresponse-sdc-example-ussg-fht-answers.json.html)
    - Links to the 2016may build (sent by Lloyd):

WG health metrics
  - Docs
    - Mission/Charter (2 yrs) ! expires this Sept !
      - Last Update: 09-17-2014
    - SWOT (3 yrs)
      - Last Update: 09-17-2014
Baltimore WGM Room Requests - Due two weeks after WGM (5/27) - done!
- M Q3, Q4 (joint with FHIR)
- T Q1, Q2, Q3, Q4
- W Q1, Q2 (joint with OO), Q3, Q4

Leftover topics