Exported to PDF June 20, 2016

HL7 Clinical Genomics Weekly Call - June 14, 2016

Attendees
1. Bob Milius - NMDP - bmilius@nmdp.org (presiding co-chair)
2. JD Nolen - Cerner - johndavid.nolen@cerner.com
3. Kevin Power - Cerner - kpower@cerner.com
4. Perry Mar - Partners HealthCare System - pmar@partners.org
5. Joel Schneider - NMDP - jschneid@nmdp.org
6. Jeremy Warner - Vanderbilt - jeremy.warner@vanderbilt.edu
7. Siew Lam - Intermountain Healthcare - siew.lam@imail.org
8. Joseph Kane - Epic - jkane@epic.com
9. Mollie Ullman-Cullere BOC mollie@betteroutcomes.com
10. David Kreda - david.kreda@gmail.com
11. Andrea Pitkus - IMO - apitkus@imo-online.com
12. Gaston Fiore - BCH - gaston.fiore@gmail.com
13. Bob Freimuth - Mayo Clinic - freimuth dot robert at mayo dot edu
14. Bret Heale - Intermountain Healthcare - bheale@gmail.com
15. Amnon Shabo (Shvo) - Philips - amnon.shvo@gmail.com

Discussion
- Minutes approval
  - June 7 call minutes approval
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20160607.pdf
    - Motion to accept - Lam
    - Second - Andrea
    - yea/nay/abstain = 8 /0 / Mollie, Jeremy
    - Results - minutes are accepted
- Brief reports
  - ClinGen/ClinVar
    - Nothing new
  - GA4GH
    - Nothing new
  - National Academies
    - JD - nothing new
  - Other
    - Jeremy sent out Jackson Lab info about 30 secs ago, please look at the it and give feedback. https://ckb.jax.org - currently focused on 82 genes and variants with treatment implications (for cancer)
- **FHIR current build content vote**
  - Add PhaseSet element to ObservationForGenetics profile
    - Motion/Second = Bob M/Gaston
    - Discussion
    - Results - Yes/No/Abstain = 11/0/2
  - Change element definitions to SO whenever available

- - - Motion/Second = Bob M/Clem
    - Discussion = Clem wants more info, are there competing ontologies?
      - Bret H - do we copy the definition or point to it?
      - Gaston - both
      -
    - Results - Yes/No/Abstain = 12/0/1
  - Add pointer element to ObservationForGenetics profile in Sequence.variant
    - Motion/Second = Gil/Bret
    - Discussion - Bob asked for clarification as to when this would be useful.
      - Response from Gil included a need to associate multiple variants/observations. Also, FHIR has a limitation … (?can someone provide a clear description here?)
        - Bob M - A resource can reference another resource, but can't reference a specific element in another resource.
      - David K suggested that a vote might be premature
      - From Perry Mar :"There was some mention of which way a pointer should point conceptually--from observation to sequence/variant or vice versa. However, a FHIR message specification is not the same thing as a conceptual domain analysis information model. We may specify how the domain should be regarded in the domain analysis model (DAM) and yet include an additional pointer in the message spec in order to serve the use case requirements needed in an implementation, even if it is not represented that way conceptually in the DAM."
      - From Bret H: If a profile derived from sequence resource cannot point back to an observation that it is associated with, then it cannot stand on its own, right? is this problematic?
      - JD: reportability and higher level interpretation are key concepts that fall more on the observation-side (closer to the clinical workflow) vs. the sequence-side (closer to the underlying structure). Adding a pointer element will make this happen.
    - Results - Yes/No/Abstain = 9/0/4
  - Final results are at https://docs.google.com/spreadsheets/d/1Rmsks_wmlpCDxneCq6YsXIH71_3JeG-rFRFMtLb4R2Y/edit?usp=sharing
  - An image of the results appears below as well.

Timestamp	Add PhaseSet	Change eleme	Add pointer el	Name	Organization	Email	Feedback
2016/06/10 2:	Yes	Yes	Yes	Kevin Power	Cerner		
2016/06/10 2:	Yes	Yes	Abstain	Jim Shima	Syapse		
2016/06/10 4:	Yes	Yes	Yes	Gil Alterovitz	BCH/HMS	gilusa@gmail.	
2016/06/13 11	Yes	Yes	Abstain	Bob Milius	NMDP	bmilius@nmd	Still not sure a
2016/06/13 3:	Yes	Yes	Yes	Huanqin Dai	BCH	huanqindai@g	
2016/06/13 3:	Yes	Yes	Yes	Xiaojia Yu	BCH		
2016/06/13 4:	Yes	Yes	Yes	Deven Atnoor	?		
2016/06/13 5:	Yes	Yes	Yes	Ning An	?	ning.an@gmai	
2016/06/14 12	Yes	Yes	Yes	Jiaoyun Yang	?	jiaoyun@hfut.	
2016/06/14 5:	Abstain	Abstain	Abstain	Brett Johnson	BCH		
2016/06/14 5:	Abstain	Abstain	Abstain	Brett Johnson	BCH		
2016/06/14 8:	Abstain	Yes	Yes	Jeremy Warne	Vanderbilt	jeremy.warner	
2016/06/14 10	Yes	Yes	Yes	Gaston Fiore	BCH		
2016/06/14 10	Yes	Yes	Abstain	Joel Schneider	NMDP	jschneid@nm	Need more inf

- **FHIR STU3 Deadlines**
  - From http://wiki.hl7.org/index.php?title=May_2016_WGM_Montreal,_Canada:_May_8_to_May_13
  - Wed, June 1
    - All resource and IG proposals for STU3 have been completed, reviewed by WG and submitted
    - Connectathon tracks for Sept have been proposed
    - Feedback on gForge submitted to FMG (CG didn't have any feedback)
  - Sun, July 17 - Substantive content freeze for ballot -- core resources
    - CG needs feedback by then
  - Sun, July 24 - Total content freeze, start of QA
  - Wed, Aug 10 (midnight) - All QA changes applied
  - Fri, Aug 12 - FHIR ballot opens
  - Fri, Sept 12 - FHIR ballot closes
  - Fri, Sept 16 - FHIR triage complete and ballot content loaded to gForge (or alternate)
  - Sept 17-23 - Baltimore WGM
- Related to above - what is the CG timeline? E.g., although the WG approved the resource/profile proposals due on June 1, it was with the understanding that the WG has to approve the final content, and it doesn't commit us to including all of it.
  - Tues, July 12 - WG vote for final content ?

Recorded Chat

06/14/2016   10:01:09 AM   from Bob Milius to Everyone:

minutes caputured here http://bit.ly/1OlYUv7

06/14/2016   10:09:49 AM   from David Kreda to Everyone:

Sara E. Patterson, Rangjiao Liu, Cara M. Statz, Daniel Durkin, Anuradha Lakshminarayana, and Susan M. Mockus*, The Clinical Trial Landscape in Oncology and Connectivity of Somatic Mutational Profiles to Targeted Therapies. Human Genomics, 2016 Jan 16;10(1):4.

06/14/2016   10:13:08 AM   from David Kreda to Everyone:

The Jackson Lab article is a free PDF at:

http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4715272/pdf/40246_2016_Article_61.pdf

06/14/2016   10:15:43 AM   from Bret Heale to Everyone:

SO is an open, publically funded resource, once you capture the definition in our documents, we can refer to SO.

06/14/2016   10:16:03 AM   from Bret Heale to Everyone:

our definition can be based on SO but don't need to be SO's

06/14/2016   10:22:44 AM   from Gaston Fiore to Everyone:

https://docs.google.com/forms/d/15tYyg6TKcUteYBfn2fmEBjh4AMRPqixigHja2eSuodw/edit#responses

06/14/2016   10:25:42 AM   from Larry Babb to Everyone:

Bear in mind that the Version information is pretty important (with the SO id) because the SO authors can change relationships and structures such that the definition is not the same as what you may have originally thought.

06/14/2016   10:26:17 AM   from Larry Babb to Everyone:

but I think it is a good choice.

06/14/2016   10:27:25 AM   from Larry Babb to Everyone:

also, be aware that there are competing SO ontologies, like Variant Ontology (VariO).

06/14/2016   10:27:33 AM   from Larry Babb to Everyone:

i think SO is probably the more dominant

06/14/2016   10:28:01 AM   from Siew Lam to Everyone:

On the CKB, the web site that Jeremy sent, the high level data model is available when you click 'About'

06/14/2016   10:30:59 AM   from Bret Heale to Everyone:

Thanks Lam

06/14/2016   10:48:06 AM   from Perry Mar to Everyone:

There was some mention of which way a pointer should point conceptually--from observation to sequence/variant or vice versa. However, a FHIR message specification is not the same thing as a conceptual domain analysis information model. We may specify how the domain should be regarded in the domain analysis model (DAM) and yet include an additional pointer in the message spec in order to serve the use case requirements needed in an implementation, even if it is not represented that way conceptually in the DAM.

06/14/2016   10:54:34 AM   from Bret Heale to Everyone:

If a profile derived from sequence resoruce cannot point back to an observation that it is assoicated with. Then it cannot stand on its own, right? is this problematic?

06/14/2016   10:56:13 AM   from Amnon Shabo (Shvo) to Everyone:
agree with JD!

06/14/2016   11:01:29 AM   from Bret Heale to Everyone:
please edit the google doc! :^}