HL7 Clinical Genomics Weekly Call - February 6, 2018 11:00 AM (US Eastern)

Minutes:
https://docs.google.com/document/d/12-uBrMmav71a3_c9h_FXQteJo_I5Kt72NEBYXZuwhFg/edit

Minutes (short url):
http://bit.ly/2aqVmqz

Attending the meeting:

- Join the online meeting (VoIP available with this):
  - Online Meeting Link:
    - https://join.freeconferencecall.com/clingenomics
  - Online Meeting ID:
    - clingenomics
- Dial into the conference:
  - Dial-in Number:
    - (515) 604-9708 - United States
  - Access Code:
    - 289092
  - International Dial-in Numbers:
    - https://www.freeconferencecall.com/wall/clingenomics/#international

Agenda

Minutes Approval

Topics to review
- Upcoming agendas
- External efforts
- Subgroup reports

Topic 1: Quick WGM Update

Topic 2: Unification

Topic 3: Unification (topics for next call)

Chat

CG Attendees (Presiding Chair: Kevin Power)

1. Amnon Shabo (Shvo), Philips, amnon.shabo@philips.com
2. Joel Schneider - CIBMTR / NMDP - jschneid@nmdp.org
3. Amnon Ptashek - Edico Genome - genptashek@gmail.com
4. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
5. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
6. David Poloway - BCH - dwpoloway@gmail.com
7. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
8. Clem McDonald - NLM - clemmcdonald@mail.nih
9. Alex Mankovich - Philips - alex.mankovich@philips.com
10. Dorina Bratfalean -CDISC- dbratfalean.external@cdisc.org
11. Julian Sass - Niederrhein University - julian.sass@hs-niederrhein.de
12. Caterina Lasome - AFMS - cat@ioninformatics.com
13. Larry Babb - Sunquest - larry.babb@sunquestinfo.com
14. Bob Milius - NMDP/CIBMTR - bmilius@nmdp.org
15.
16.

Minutes Approval

- Jan 23
  - http://wiki.hl7.org/index.php?title=File:HL7_CG_20180123.pdf
  - motion/2nd to accept minutes - Bob F/Clem
  - discussion - None
  - Abstain / Nay / Yea:
    - 0 / 0 / 9
  - result -

Topics to Review

Agendas and Important Dates

Date	Co-Chair	Agenda	Important Dates
6-Feb	Kevin	WGM Review Unification	
13-Feb			Inform FMG of NIB: Feb 14 **HL7 Genomics Conference**: Feb 20 - Feb 21
20-Feb			Connectathon proposals to FMG: Feb 21 NIB submission deadline: Feb 25
27-Feb			
6-Mar			
13-Mar			
20-Mar			
27-Mar			FHIR Final Content Freeze: April 1
3-Apr			May 2018 Ballot opens: April 6
10-Apr			
17-Apr			
24-Apr			
1-May			May 2018 Ballot closes: May 7

8-May			
May 2018 HL7 Working Group Meeting May 12, 2018 to May 18, 2018 - Maritim Hotel Köln, Cologne, Germany			

External efforts

- GA4GH Genomic Knowledge Standards (GKS) (leads: Bob Freimuth, Andy Yates)
    -
- National Academies
    -
- ClinGen/ClinVar
    -
- Variant Modelling Collaboration (VMC)
    -
- CDISC PGx
    - CDISC PGx weekly meeting- Refine philosophy/strategy for controlled terminology in the PFTESTCD/PFTEST codelists. (Dorina B)
- ONC Sync for Genes
    -

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/1azKiQdhAQKuHhxAznEp8141FLdFLACIu8MzF2LxADxg/edit#
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5

Topic 1: Quick WGM Update

https://tinyurl.com/HL7CG-WGM-Jan2018

Topic 2: Unification

See “New Model”
https://docs.google.com/document/d/1juWEnjyXV34yYmPq3FDpLAiJlM0Hiv0FyNBfvPD6enM/edit

Lloyd presenting a review of the latest updates <Lloyd to share latest updates>

Questions:

- Specimen relationship to variant?
    - Consensus is yes.
- Change Assertion to Findings?
    - Consensus is yes.
- Change Genetic Interpretation to Overall Genetic Interpretation
    - Yes?
- Change to Genetic Impact?

        - Larry - Change to Knowledge/Assertion?
        - Jeremy - Wouldn't use assertion because it has a general definition in the computer science/NLP sense that encompasses any instantiation of reality
- SomaticPathogenicity
    - ??? means we are lacking LOINC codes
- In general, is the report include the knowledge, patient specific impact, or both?
    - Larry - Inherited disease, it is both.
- Impact is about "potentially actionable findings"
- Discussing LOINC 53040-2
    - Kind of variant? Assessment of the knowledge of a variant
- Need to consider how to model "Knowledge"
    - Lloyd will propose an alternate model
-

Topic 3: Unification (topics for next call)

- Start discussing the profiles in order from the WGM:
    - *Proposed 'narrow' list of profiles to consider for ballot in May*
        - *Really High Priority (I mean it)*
            - *(Genetics) DiagnosticReport*
            - *Specimen Source*
            - *Genetic Observation Common Properties*
            - *Computable Genetic Assertion*
            - *Haplotype*
            - *Genotype*
            - *Overall Variant*
        - *Higher priority*
            - *Genetic Interpretation (subclasses of)*
            - *Genetic Impact (subclasses of)*
        - *High priority*
            - *Described Variant*
            - *Sequence*
        - *Medium priority*
            - *General Recommendation*
            - *Medication Usage Implications*
        - *Low priority*
            - *Current Medication*
            - *Order for Genetic Test*
            - *Descriptive Genetic Assertion (incl subclasses)*
            - *Complex Variant*
            - *Copy Number Change*
            - *Microarray Platform*

Chat

Larry 10:19AM
Can "Genomics Panel" be considered "Genomics Targeted Analysis"?
Larry 10:22AM
Can we consider changing "Genetic Assertions" to "Genetic Findings"?
Larry 10:22AM
assertions and assessments or interpretations seem to be used interchangeably in this domain.
Amnon Shabo (Shvo) 10:27AM
'genomics source' is more of analysis result than propoerty of specimen

Larry 10:40AM
Impact should probably be changed to "Knowledge" or "Assertion" IMO
Jeremy Warner 10:41AM
I wouldn't use assertion because it has a general definition in the computer science/NLP sense that encompasses any instantiation of reality
Larry 10:43AM
mode-of-inheritance value set codes exist in HPO, ClinGen has drafted this value set
Amnon Shabo (Shvo) 10:54AM
In New Orleans I also proposed to consider the GuidanceResponse resource to capture the use of knowledge in some analysis of genomics observations
Larry 10:57AM
+1 amnon https://www.hl7.org/fhir/guidanceresponse.html
Jeremy Warner 10:58AM
Problem with using that resource is that not all findings in the somatic world are linked to actions -plenty are purely prognostic and the only action might be counseling/transfer of knowledge
Larry 10:59AM
nice job, Lloyd
Jeremy Warner 11:00AM
+1 Larry

Clinical Genomics Docs

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - Review complete as of Aug 1, 2017
    - Approved in Sep WGM in San Diego
- Decision Making Process
    - https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit
    - Review complete as of Aug 15, 2017
    - Approved in Sep WGM in San Diego
- DAM
    - http://tinyurl.com/damcgdoc