HL7 Clinical Genomics Weekly Call - June 19, 2018 11:00 AM (US Eastern)

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

Attending the meeting:
Join the online meeting (VoIP available with this):

- Online Meeting Link:
  - https://join.freeconferencecall.com/clingenomics
  - 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

Attendees Sign-in

Minutes Approval

Topics to Review

- Agendas and Important Dates
- External efforts
- Subgroup reports

Topic 1: Conference Call choice

Topic 2: Connectathon brainstorming

Topic 3: Block Vote

Topic 4: Ballot discussion - “Variant Grouping”

Chat

Attendees Sign-in

(Presiding Chair: Kevin Power - kpower@cerner.com)

1. Jamie Parker - Carradora Health - jamie.parker@carradora.com
2. Arthur Hermann- Kaiser Permanente - arthur.hermann@kp.org
3. Caterina Lasome - iON Informatics for AFMS - cat@ioninformatics.com
4. Patrick Werner - Molit Institut / Heilbronn University - patrick.werner@molit.eu
5. Amnon Shabo (Shvo) - Philips - amnon.shabo@philips.com
6. Bob Dolin - Elimu Informatics - bdolin@elimu.io
7. Julian Sass - Niederrhein University - julian.sass@hsnr.de
8. Alex Mankovich - Philips - alex.mankovich@philips.com
9. Bret Heale - Intermountain Healthcare- bheale@gmail.com
10. Michael Stevens - Optum - jmichael.stevens@optum.com
11. Liz Amos - NLM - liz.amos@nih.gov
12. Clem McDonald - NLM - clemmcdonald@nlm.nih.gov
13. Joseph Kane - Epic - jkane@epic.com
14. Andrea Pitkus - apitkus@gmail.com
15. Ling Teng- BCH -tenglingling@gmail.com
16. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
17. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
18. Dorina Bratfalean -CDISC- dbratfalean.external@cdisc.org
19. Elizabeth Newton - Kaiser Permanente - elizabeth.h.newton@kp.org

Minutes Approval

- June 12
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20180612.pdf
    - motion/2nd to accept minutes - Arthur / Patrick
    - discussion - None
    - Abstain / Nay / Yea:
        - 0 / 0 / 12
    - result - passes

Topics to Review

Agendas and Important Dates

Date	Co-Chair	Agenda	Important Dates
5/29/2018	Bob M	Review WGM minutes (note that Amnon Shabo edited the minutes regarding the sessions when his ballot comments were discussed)	
6/5/2018	Kevin	Ballot comments	Jun 6 - Deadline for connectathon proposals to FMG
6/12/2018	Kevin	Con call tech Connectathon 'Variant Grouping'	
6/19/2018	Kevin	Con call tech Connectathon Block Vote 'Variant Grouping'	
6/26/2018	Bob M		June 27 - Connectathon Proposals Due
7/3/2018			Jul 1 - Work groups notify the FMG whether they need to reballot normative packages (due to substantive change), STU resources (due to significant refactoring) or IGs during the Sept. cycle July 6 - Deadline to notify HG of additions/changes to co-chair opentings

7/10/2018			July 11 - Call for co-chair nominations July 15 - Notification of Intent to Ballot
7/17/2018	Bob M		
7/24/2018			July 23 - formation of consensus groups
7/31/2018			
8/7/2018	Bob M		Aug 5 - Reconciliation packages must be posted by this date at the absolute latest Aug 10: All substantive reconciliation applied. FHIR Core is frozen, limited QA process for content subject to ballot only Aug 10 - close to co-chair nominations
8/14/2018			Aug 17: Pre-ballot (and connectathon) content freeze. Publication process begins, including ensuring that content is appropriately flagged for ballot status and there are no last minute QA issues
8/21/2018			Aug 24 - ballot opens for voting
8/28/2018	Bob M		
9/4/2018			
9/11/2018			
9/18/2018	Bob M		
9/25/2018			
32nd Annual Plenary & Working Group Meeting Sep 29, 2018 to Oct 5, 2018 - Baltimore, MD			

External efforts

- GA4GH Genomic Knowledge Standards (GKS) (leads: Bob Freimuth, Andy Yates)
    -
- DIGITiZe (aka National Academies) (Grant Wood, JD Nolen)
    -
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
    -
- Variant Modelling Collaboration (VMC) (Larry Babb, Bob Freimuth)
    -
- CDISC PGx (Dorina B.)
    -
- ONC Sync for Genes (Bob Freimuth)
    -

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: Conference Call choice

Free Conference Call was selected. We will get the Monday meeting moved over to FCC starting next week.

Poll "HL7 Clinical Genomics - Free Conference Call or Zoom?" https://doodle.com/poll/4qhc8fg26z7rvmyy

	Free Conference Call	Zoom
Rosalyn Ryan		OK
Lloyd McKenzie	OK	
Bob Milius	OK	
Ling teng	OK	
Dora Finkeisen	OK	
Kevin Ehlers	OK	
David Poloway	OK	
Bob Freimuth	OK	
Scott Robertson	OK	
Arthur Hermann	OK	
Patrick Werner	OK	
Joseph Kane	OK	
Deepak Sharma		OK
Andrea Pitkus	OK	
Bret Heale	OK	
Liz Amos	OK	
Clem McDonald	OK	
Count	15	2

1 / 1

Topic 2: Connectathon brainstorming

Patrick has created a draft:
http://wiki.hl7.org/index.php?title=201809_Clinical_Genomics

Connectathon attendees (feel free to update):
Kevin P
Bob M
Alex M
Joseph K (?)
Paul Lynch (NLM)
Ye Wang (NLM)
Amnon Shabo (Shvo)
Dora Finkeisen (?)
Patrick Werner
Joel Schneider

Topic 3: Block Vote

Comment Submitters

- Clement McDonald
- Kevin Power

Line Items

16816
Clarify+needed+to+distinguish+report+requires+computable+content+but+allows+text+when+needed+-+2018-May+Genomics+%2338 (Clement McDonald) Not Persuasive
16106 Descriptions+wrong+on+Genetic+Observation+Common+Properties (Kevin Power) Persuasive
16689 Fix+language+from+specimen+-%3E+sequence+-+2018-May+Genomics+%233 (Clement McDonald) Persuasive
16692
Confusing+text+re%3A+sequence+referred+to+as+variation+-+2018-May+Genomics+%234 (Clement McDonald) Persuasive
16695
Clarification+on+wording+to+correct+%22presumed+cause%22+-%3E+type+of+variation+-+2018-May+Genomics+%235 (Clement McDonald) Persuasive
16701 Inserted+text+to+clarify+reporting+on+sequences+-+2018-May+Genomics+%237 (Clement McDonald) Persuasive
16707
Inserted+text+to+describe+package+of+information+in+genetic+analysis+-+2018-May+Genomics+%239 (Clement McDonald) Persuasive
16727 so+I+think+it%27s+better+to+generalize+the+findings.+-+2018-May+Genomics+%2315 (Clement McDonald) Persuasive
16736
Change+%22bases%22+-%3E+%22units+of+analysis%22+-+2018-May+Genomics+%2317 (Clement McDonald) Persuasive
16756
Clarification+on+use+of+word+%22indication%22+to+describe+testing+or+reason+for+testing+-+2018-May+Genomics+%2322 (Clement McDonald) Persuasive
16797
Discrepency+between+proposed+label+and+DNA+position+on+LOINC+code+-+2018-May+Genomics+%2333 (Clement McDonald) Persuasive
16804
Discrepency+on+naming+and+general+concern+over+name+change+-+2018-May+Genomics+%2335 (Clement McDonald) Persuasive
16704
Change+%22focal+element%22+-%3E+%22payload%22+-+2018-May+Genomics+%238 (Clement McDonald) Persuasive with Mod

16710
Change+%22divergences%22+-%3E+%22differences%22+-+2018-May+Genomics+%2310 (Clement McDonald) Persuasive with Mod
16731
Change+%22bases%22+-%3E+%22units+of+analysis%22+-+2018-May+Genomics+%2316 (Clement McDonald) Persuasive with Mod

Vote:
Motion/2nd to accept the dispositions on this block: Bret / Clem
Discussion:
Vote: Abstain / Nay / Yea
0 / 0 / 18

Topic 4: Ballot discussion - “Variant Grouping”

Several trackers logged around the various profiles we have defined for “Variant Grouping” - so our concepts like:
Genotype
Haplotype
SequenceConfiguration

See pages here:
http://build.fhir.org/ig/HL7/genomics-reporting/general.html#findings
http://build.fhir.org/ig/HL7/genomics-reporting/sequencing.html (see ComplexVariant)

ID	Summary	Details
16812	Comment on haplotype being identified absence info on variant - 2018-May Genomics #37	Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Figure 5: Variation (any type) --- Comment: Didn't think a haplotype can be identified in absence of any information on variant. --- Summary: Comment on haplotype being identified absence info on variant
16808	Complex variants distinguish cis from trans - 2018-May Genomics #36	Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Figure 5: Cis or Trans --- Comment: Unclear - I don't recall discussion plus the complex variants distinguish this (I think). --- Summary: Complex variants distinguish cis from trans
16789	Discussion needed on change from display names on 84413-4 - 2018-May Genomics #31	Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Figure 5: Genotype 84413-4 Proposed Wording: Genotype Display Name 84413-4 --- Comment: I understand why you want to shorten, but the change could mislead. These are not solid codes for genotype or haplotype. Would like to find a way to link from the figure (or content below them) to the LOINC code, description and answer list. Have linked to the answer list in the change document but these early tables are a bit more digestable. Lets talk. --- Summary: Discussion needed on change from display names on 84413-4
16793	Discussion needed on change from display names on 84414-2 - 2018-May Genomics #32	Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Figure 5 Haplotype 84414-2 Proposed Wording: Haplotype Name 84414-2 --- Comment: Name in V2 --- Summary: Discussion needed on change from display names on 84414-2
16496	phase set of sequences (not variants)	http://www.hl7.org/fhir/2018May/extension-observation-geneticsphaseset.html easily describes how a set of sequences (not necessarily variants) can be grouped according to being in chromosomal phase with one another (cis, on the same molecule). This is useful for my use case. I don't see how this can be done in the current IG. allele-phase in described variant doesn't do it as far as I can tell. If it can, I need to see an example. Calling the phase-set a haplotype of sequences is technically correct, but seems awkward, especially since our domain talks about haplotypes in a whole gene level (eg describing whether two gene level alleles are on the same molecule. Is it possible to describe haplotypes of haplotypes? In the end, I need to see examples of this.

16173	Clarify usage of Genotype/Haplotype/SequenceConfiguration or remove for now	The Genotype/Haplotype/Sequence Configuration profiles involve groupings for various purposes. I do not believe our documentation for these is clear enough to ensure consistent usage. As an example - Sequence Configuration has basically no documentation in the IG. While these concepts are important, I am concerned that we do not have enough consensus to represent in our first draft of this IG. We need to either remove them for now or spend time creating additional documentation in order to be very clear how each should be used. As a starting point, does everyone feel that the usage of Genotype/Haplotype in the PGx example is correct? http://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html#examples It is also used in HLA examples. http://hl7.org/fhir/uv/genomics-reporting/transplants.html I am concerned that Genotype/Haplotype are not being used consistently even in our own initial examples.
16325	"haplotype" in medical genetics	Input from one of our physician/geneticists, Dr. Leslie Manace. Unfortunately, I do not have a specific url/location/resource to point this comment to. I believe the WG will be able to consider this generally and apply as appropriate. Fortunately, Kevin Power was able to provide inital feedback, which I have included below. Dr Manace: Genetic Assertions - "haplotype" is essentially never relevant in medical genetics. This is part of what gives me pause about the MD representation in this group Kevin Power: There are use cases in HLA (and even some in Pharmacogenomics) where haplotype is relevant. So, this is another case of ‘when you need haplotype, structure it like this – but skip it if you don’t need it’
16820	More explanation needed to describe genotype definition - 2018-May Genomics #39	Submitted by: Clement McDonald (National Library of Medicine) Existing Wording: Genotypes&#65533;describe combinations of genetic variations that together are associated with a particular phenotype - i.e. a specific physical, behavioral or risk-associated difference associated with the organism whose specimen was tested. --- Comment: This may not be true. I have understood that the genotype is everything you know about the individual genetics including all the normals as well as possibly multiple things that might be described as separate phenotypes. (Will need the experts to weigh in) --- Summary: More explanation needed to describe genotype definition
15885	Should consider how the PhaseSet match to the IG strucuture	The elements in the Observation-geneticsPhaseSet are different from the Allele Phase information the IG currently have. Need to think about if it should be a part of elements in Haplotype (it seems to be similar with Haplotype feature). May need a clear documentation about how to use the phaseSet element and the LOINC Allele Phase in the IG. Fan: Move the Phaseset to HaploType. And is it suitable for deleting PhaseSet ID, which is no mapping to coding system (LOINC) and unuseful
16512	Sequence Configuration cardinality	Sequence Configuration has a obs-focus with a cardinality of 2..2. I assume this for the case when trans is value. But if the value is cis, then the cardinality could be 2..* Not sure how, but It would be nice to be able to do this. Then I could effectively have a set of sequences in a phase-set. Practically speaking, I think most labs report if they have evidence of sequences being cis, but not for trans. Evidence for trans is usually inferred from lack of evidence them being in cis.

Chat

Clinical Genomics Docs

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