HL7 Clinical Genomics Weekly Call - August 7, 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: FYI: Ballot Items for follow-up (PLEASE REVIEW!)

Topic 2: Block Vote #1

- Comment Submitters
- Line Items

Topic 3: Cytogenetics in the IG

Topic 4: Compare Sequence and Observation

Chat

Attendees Sign-in

(Presiding co-chair: Kevin Power)

1. Liz Amos - NLM - liz.amos@nih.gov
2. Clem McDonald - NLM - clemmcdonald@mail.nih.gov
3. Joseph Kane - Epic - jkane@epic.com
4. Bob Dolin - Elimu Informatics - bdolin@elimu.io
5. Patrick Werner - MOLIT Institut - Patrick.Werner@molit.eu
6. JD Nolen - Children's Mercy Hospital - jlnolen@cmh.edu
7. James Jones - BCH - james.jones.bch@gmail.com
8. Alex Mankovich - Philips - alex.mankovich@philips.com
9. Lloyd McKenzie - Gevity - lmckenzie@gevityinc.com
10. Bret Heale - Intermountain Healthcare - bheale@gmail.com
11. Dora Finkeisen - MOLIT Institut - Dora.Finkeisen@molit.eu
12. Joel Schneider - NMDP/CIBMTR - jschneid@nmdp.org
13. Ning Xie - BCH -ningxie2018@gmail.com
14. Deepak Sharma - Mayo Clinic - sharma.deepak2@mayo.edu
15. Julian Sass - Berlin Institute of Health - julian.sass@bihealth.de
16. Scott Robertson - Kaiser Permanente - scott.m.robertson@kp.org
17. Mullai Murugan - Baylor College of Medicine - murugan@bcm.edu
18. Jungang Zou - Xiamen University - jungang.zou@gmail.com

Minutes Approval

- July 31
    - http://wiki.hl7.org/index.php?title=File:HL7_CG_20180731.pdf
    - Motion/2nd to accept minutes: Clem / Joseph
    - Discussion: None
    - Vote: Abstain / Nay / Yea: Bret, Mullai (2) / (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	Bob M	IG block vote	July 11 - Call for co-chair nominations July 15 - Notification of Intent to Ballot

		NIB vote	
7/17/2018	Bob M		
7/24/2018	Kevin	Consensus Groups Definitional Sequence	July 23 - formation of consensus groups
7/31/2018	Bob F	Topic 0: Deadline for resolutions to ballot comments Topic 1: FYI: Ballot Items for follow-up Topic 2: Block Vote #1 Topic 3: Block Vote #2 Topic 4: Proposed changes to Described Variant Topic 5: Secondary findings (proposal) Topic 6: Impact vs. Interpretation	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
8/7/2018	Kevin P	Trackers needing follow-up Block Vote Cytogenetics in the IG Compare Sequence and Observation	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	Bob F		
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)
    - Variant Representation (formerly VMC)
        - Work continues on two fronts: we are finishing up the last pieces to the 0.2 release (lead by Reece Hart) and we are extending the model to support complex variants (e.g., fuzzy ends) (lead by Larry Babb/Tristan Nelson)
    - Variant Annotation
        - Collecting use cases, prioritizing for modeling work
- DIGITiZe (aka National Academies) (Grant Wood, JD Nolen)
    -
- ClinGen/ClinVar (Larry Babb, Bob Freimuth)
    -
- CDISC PGx (Dorina B.)
    -
- ONC Sync for Genes (Bob Freimuth)
    - Pilot sites are planning/implementing their respective use cases. ONC will be encouraging their participation in both the Sept 2018 and Jan 2019 FHIR Connectathons.
    - ONC 2nd Interoperability Forum (August 6th- 8th, 2018 in Washington, DC)
        - https://www.healthit.gov/news/events/oncs-2nd-interoperability-forum

Subgroup reports

- IM (Bob F)
    - https://docs.google.com/document/d/1azKiQdhAQKuHhxAznEp8141FLdFLACIu8MzF2LxADxg/edit#
        - Working on getting documentation together for 1st draft of model (first 4 submodels only), will present at next IM call on Thursday and post for comments
        - Link to docs will be circulated after next IM call
        - Initial work will be presented at a Tuesday call after comments are received
- FHIR (Gil)
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.zfi9l8jfe4la

Topic 1: FYI: Ballot Items for follow-up (PLEASE REVIEW!)

gForge Query

ID	Summary	Resolution	Ballot-weight	Waiting for Input e-mail(s)	Real Submitter
16929	from descriptive to computable - 2018-May Genomics #75	Deferred?	Affirmative	amnon.shvo@gmail.com	Amnon Shabo
16489	'Genomic Alelle start-end' should use 'start' and 'end' instead of 'low' and 'high'	Because the Range data type in FHIR is a generic data type and uses the terms "low" and...	Negative-Major	clemmcdonald@mail.nih.gov	Bob Milius
16472	clarify allelic-phase	Retract?	Negative-Major	bmilius@nmdp.org	Bob Milius
16250	Sequence.structureVariant.reportedaCGHRatio	Gil?	Affirmative	gil.alterovitz@gmail.com bdolin@elimu.io	Bob Dolin
16249	Sequence.structureVariant: Add Sequence.structureVariant.variantType	Gil?	Affirmative	gil.alterovitz@gmail.com bdolin@elimu.io	Bob Dolin
16251	Clarify intent of Sequence.structureVariant.precision	Gil?	Affirmative	gil.alterovitz@gmail.com bdolin@elimu.io	Bob Dolin
16246	Revise Sequence attribute descriptions	Gil?	Affirmative	gil.alterovitz@gmail.com bdolin@elimu.io	Bob Dolin
16110	DiagnosticReport Category could also be Cytogenetics	Waiting on decision around keeping Cytogenetics in the IG.	Affirmative	kpower@cerner.com	Kevin Power
16876	Use of publically available external coding systems and autocomplete lookup tables - 2018-May Genomics #58	A proposed new section on the V2 page: All of the publicly available external coding systems...	Negative-Major	clemmcdonald@mail.nih.gov	Clement McDonald
16840	More information needed for relatedArtifact - 2018-May Genomics #45		Negative-Major	clemmcdonald@mail.nih.gov	Clement McDonald

16923	genetic finding - 2018-May Genomics #73	Detail requested from Amnon.	Affirmative	amnon.shvo@gmail.com	Amnon Shabo
16935	types of overall interpretation - 2018-May Genomics #77	Notes from WGM say defer?	Negative-Major	amnon.shvo@gmail.com tenglingling@gmail.com	Amnon Shabo
16913	Interpretation vs. impact - 2018-May Genomics #69	Consider for future use (according to May 2018 WGM notes).	Negative-Major	amnon.shvo@gmail.com	Amnon Shabo
16766	2018-May Genomics #25	Proposal: Text to add after the first paragraph: The core of the typical report is a list...	Negative-Major	clemmcdonald@mail.nih.gov	Clement McDonald
16837	Concern the spec is too assuming in unsolicited - 2018-May Genomics #44	Proposal: Change wording to summarize new "Secondary Findings"...	Negative-Major	kpower@cerner.com	Clement McDonald
16248	Sequence.quality.standardSequence clarification		Affirmative	gil.alterovitz@gmail.com bdolin@elimu.io	Bob Dolin
16247	Sequence quality needs more work		Affirmative	gil.alterovitz@gmail.com bdolin@elimu.io	Bob Dolin

Kevin: Some feedback was received, will be reviewed and entered

Topic 2: Block Vote #1

Pulled by Bob M:

16513 need+glossary (Bob Milius) Not Persuasive

- Bob M comment - If the comment is back in play when we are ready to publish, shouldn't this ballot comment remain open? Isn't the point of ballot comments is they are things to resolve before publication?

Pulled by Patrick W.:

~~16107 Genetic+Observation+Common+Properties%3A+necessary+to+have+%22Must+Support%22+on+body structure%3F (Kevin Power) Persuasive~~

- ~~The meaning of Must support in FHIR has to be defined by the profile/IG:~~

~~https://build.fhir.org/profiling.html#mustsupport e.g.: "*The system must be able to store and retrieve the element*". So a 0..X cardinality and must support makes a lot of sense for us.~~

After discussion, Patrick agreed with putting back into the block. Might be flagged as 'must support' for certain types of testing (Somatic).

Comment Submitters

- Amnon Shabo
- Bob Milius
- Clement McDonald
- Kevin Power
- Scott Robertson

Line Items

16324 more+sub-categorization+for+sequence+variants (Scott Robertson) Considered - Question Answered
16514 Relevance+of+the+genomic+apps+in+this+IG%3F (Bob Milius) Not Persuasive
16860 Inserted+text+on+somatic+variants+-+2018-May+Genomics+%2352 (Clement McDonald) Not Persuasive
16900 pre-coordination+-+2018-May+Genomics+%2365 (Amnon Shabo) **In Person** Not Persuasive
16910 phenotype+ontology+-+2018-May+Genomics+%2368 (Amnon Shabo) **In Person** Not Persuasive
16107 Genetic+Observation+Common+Properties%3A+necessary+to+have+%22Must+Support%22+on+body-structure%3F (Kevin Power) Persuasive
16882 Misleading+text+on+full+sequence+testing+on+viruses+-+need+to+clarify+reality+-+2018-May+Genomics+%2360 (Clement McDonald) Persuasive
16831 Inserted+text+to+explain+laboratory+findings+-+2018-May+Genomics+%2342 (Clement McDonald) Persuasive with Mod

- Motion/2nd to accept block: Bret / Clem
- Discussion: None
- Vote: Abstain / Nay / Yea: Scott R (1) / (0) / 16
- Result: Passes

Topic 3: Cytogenetics in the IG

Clem
Remove some of the Cytogenetic profiles from the IG

<slides to show proposal emailed to the group>
From: Liz Amos
Subject: [clingenomics] Materials for cytogenetics discussion

NOTE - not suggesting we don't include this at some point, just not now.

Could leave Cytogentics as a placeholder, but for now remove the profiles.

Where do we connect Microarray Platform (Device) and FISH Probe (DeviceComponent)? Perhaps attached to Computable Genetic Finding so that they apply to all Variations?

Copy Number Change profile came from V2 Cytogenetics IG

Asked for anyone who disagrees? No response.

Clem will finalize this proposal and share with the group. Kevin will then move the proposal into the appropriate ballot comments.

Topic 4: Compare Sequence and Observation

https://docs.google.com/spreadsheets/d/1z4DodoLYawW-s0jbFKQg_xpwir8rEORkNjMfemvqxE0/edit#gid=0

Chat

Andrea Pitkus 10:14AM

- a laboratory may make body structure aka specimen source required data element
- tissues and swabs would require body sites
- specimen like water may not have a body site

Bret Heale 10:15AM

- specimin type dictates methodology. But here the message is the result

Andrea Pitkus 10:15AM

- or non human substances.... like organisms

Bret Heale 10:16AM

- an order would need the specmin site information.
- there is a link to specimin within OBservation

Andrea Pitkus 10:18AM

- correct...

Bret Heale 10:19AM

- thanks llyod and andrea. So body structure is optional but specimin is abslutely required

Andrea Pitkus 10:19AM

- specimen is different from body site as Clem noted. Body structure/anatomic site/body site are the same from a SCT/HL7 perspective.

Bret Heale 10:19AM

- thx

Deepak 10:23AM

- I cannot see the screen

Bret Heale 10:23AM

- ISCN id? keep that, right?

Deepak 10:24AM

- I see it now

Andrea Pitkus 10:24AM

- it's required as part of CAP/CLIA requirements for cytogenetic reports

Andrea Pitkus

    - 10:25AM
- In other words, all regulatory data elements are required, if folks are to implement for cytogenetics

James Jones

    - 10:25AM
- http://uvmgg.wikia.com/wiki/Chromosome_banding

Bret Heale

    - 10:26AM

- just a plug, but our information model subgroup has a good start on cytogentics...

Bret Heale
    - 10:26AM
- stainning versus microarray analysis?

Bret Heale
    - 10:32AM
- think we should rename the profile to Karyotype, if that is all it is meant to hold.

Andrea Pitkus
    - 10:32AM
- how are Accreditation requirements met with the proposal?

Andrea Pitkus
    - 10:32AM
- Reports: • The cytogenetics report must include the name and address of the testing laboratory, the patient name and unique identifying number, patient date of birth, physician name, specimen source, date of specimen receipt, date of report, clinical indication for the test, cells counted, analyzed, and karyotyped, band resolution and methods, comments on specimen adequacy, if indicated and the signature of a qualified cytogeneticist as defined in CYG.50000

Bret Heale
    - 10:33AM
- @Andrea, I think Clem is proposing to develop the profile out to inlcude those requirements, but recommend use of described variant for findings which are not Karyotype based.

Andrea Pitkus
    - 10:36AM
- that's understandable as long as all implementers realize for any ctogenetics reporting they cannot use the current model as it doesn't meet basic clinical data reproting requirements.

Bret Heale
    - 10:37AM
- ...for cytogenetics.

Andrea Pitkus
    - 10:37AM
- right.

Bret Heale

- 10:37AM

- : ^) but I'm a bit of a hacker and would use diagnostic report and genomics observations to make it happen

Andrea Pitkus

- 10:37AM

- so no prenatal, oncology, etc type testing in cytogenetics (which is very common)

Bret Heale

- 10:39AM

- +10 to ISCN use : ^)

Andrea Pitkus

- 10:39AM

- you can use observations to document all the required data elements such as clinical indication for the test, cells counted, analyzed, and karyotyped, band resolution and methods, comments on specimen adequacy, if indicated and the signature of a qualified cytogeneticist as defined in CYG.50000. It can all be part of a diagnostic report with the diagnosis, specimen information, ISCN nomenclature, patient information, too.

Andrea Pitkus

- 10:40AM

- If the required data elements are not included, the laboratory risks their accreditation (US), or via their ministry of health....

Andrea Pitkus

- 10:42AM

- "Cytogenetics — Used for amniotic fluid cell analyses, bone marrow cultures, chorionic villus studies, Fragile X studies, blood lymphocyte analyses, solid tumors, and nonneoplastic tissue cultures"

Andrea Pitkus

- 10:52AM

- This overlaps with the Breast Cancer FHIR resource which was ballotted too. They integrate FISH/ISH testing in this FHIR profile. What work is being done to ensure this is alligned with CG work discussed today?

Andrea Pitkus

- 10:54AM

- http://hl7.org/fhir/us/breastcancer/StructureDefinition-oncology-HER2byISH.html

Bret Heale

- 10:55AM

- additionally here's the past balloted IG: http://wiki.hl7.org/index.php?title=Breast_Cancer_Data_FHIR_IG_Proposal

Andrea Pitkus

- 10:56AM

- thanks Bret, Lloyd and Clem for your input/info in these areas

Bret Heale

- 11:04AM

- gotta go. But I suggest shooting a request to the ListServ

James Jones

- 11:04AM

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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