HL7 Clinical Genomics Weekly Call - July 18, 2017 11:00 AM (US Eastern Time)

Agenda

Minutes Approval

Topics to discuss if needed

- Upcoming agendas until San Diego
- Brief reports from external efforts
- Subgroup reports

Topic 1: CAP/CLIA requirements

Topic 2: CG Documents to review

Clinical Genomics Docs

Chat

Attendees

1. Bob Milius - NMDP - bmilius@nmdp.org
2. Andrea Pitkus - IMO- apitkus@imo-online.com
3. Caterina Lasome - iON Informatics - cat@ioninformatics.com
4. Andrew Brown - NMDP - abrown3@nmdp.org
5. Shennon Lu - NLM - shennon.lu@nih.gov
6. Clem McDonald - NLM - clemmcdonald@mail.nih.gov
7. Bob Dolin bdolin@Elimu.io
8. Joel Schneider - NMDP - jschneid@nmdp.org
9. Xin Liu - BCH - xinliu215@gmail.com
10. Joseph Kane - Epic - jkane@epic.com
11. ling teng -BCH tenglingling@gmail.com
12. Bob Freimuth - Mayo Clinic - freimuth.robert@mayo.edu
13. Gil Alterovitz - HMS/BCH- gilusa@gmail.com
14. Bret Heale - Intermountain Healthcare - bheale@gmail.com
15. Grant Wood - Intermountain Healthcare - grant.wood@imail.org

Minutes Approval

- June 27
  - http://wiki.hl7.org/index.php?title=File:HL7_CG_20170711.pdf
    -
  - motion/2nd - Bob D/Joseph K
  - discussion - none
  - abstain - none
  - nay - 0
  - yea - rest (11)

    - result - passes

Topics to discuss if needed

Upcoming agendas until San Diego

- ~~May 30 WGM recap, SWOT~~
- ~~Jun 6 - Finish WGM recap, SWOT, Pedigree Ballot - any comments?~~
- ~~Jun 13 - FHIR Connectathon, Status of LRI Ballot~~
- ~~Jun 20 - SWOT~~
- ~~Jun 27 - CLIA & CAP requirements - Unified view (Andrea, Clem, Gil needs to be present), LRI/V2 cont, SWOT~~
- ~~Jul 4 - US Holiday~~
- ~~Jul 11 - FHIR motion for changes to current build, SWOT, V2/LRI update?~~
- Jul 18 - CAP/CLIA, V2, SWOT
- Jul 25 - DAM update - Gil; Clem away
- Aug 1 - Clem away
- Aug 8 - WGM schedule draft
- Aug 15 - Gil may be gone
- Aug 22 - Clem, Bob M and Bob F out (eclipse!)
- Aug 29 - Clem gone
- Sep 5 -
- Sep 9-17 - WGM San Diego

Brief reports from external efforts

- GA4GH
    -
- National Academies
    -
- Clingen/Clinvar
    -
- Variant Modelling Collaboration (**VMC**)
    -

Subgroup reports

- IM
    - https://docs.google.com/document/d/18sVxZdAeA98ok5hdGwmmVxVinTq_vAT9B-Z8GIAyRiM/edit
    - Not meeting during July - sync'g schedule with VMC, will pick up again after public release
- FHIR
    - https://docs.google.com/document/d/1FGCQRtxJKyHhnC1uB_t4sJZ9yXbLMGOqPXHPr5tSLLQ/edit#heading=h.nts1cfujf9t5
    - See schedule at bottom of above link (feel free to add topics to calendar/email Gil)

Topic 1: CAP/CLIA requirements

- The following figures and text were added to the June 6 minutes after the weekly call.
-

5.3.9 SPM – SPECIMEN SEGMENT

TABLE 5-19. SPECIMEN SEGMENT (SPM)

SEQ	Element Name	DT	Usage	Cardinality	Value Set	Description/Comments
1	Set ID – SPM	SI	R	[1..1]		
2	Specimen ID	Varies	R	[1..1]		GU data type: EIP_GU NG data type: EIP_NG
3	Specimen Parent IDs		O			
4	Specimen Type	CWE_CRE	R	[1..1]	SNOMED CT and/or HL70487_USL	Either HL70487 or SNOMED CT Specimen hierarchy codes may be used. It should be noted that in the future SNOMED CT Specimen hierarchy may become the only recommended value set so trading partners should consider moving in that direction.
5	Specimen Type Modifier		O			
6	Specimen Additives		O			
7	Specimen Collection Method		O			
8	Specimen Source Site		O			
9	Specimen Source Site Modifier		O			
10	Specimen Collection Site		O			

- Here is the CLIA guidance in the LRI guide, required for MU. May be as simple as pointing to the fields on the right, that would be used for Clinical Genomics and Clinical Cytogenetics testing too. It may be that CD works with O&O to make the CG specific recommendations to be included within such LRI sections.

9 ADDITIONAL IMPLEMENTATION GUIDANCE – OTHER

9.1 Clinical Laboratory Improvement Amendments Considerations

In the United States, clinical laboratory testing of human specimens is regulated by the Clinical Laboratory Improvements Amendments of 1988 (CLIA). Several sections of the regulations implementing CLIA impact how electronic laboratory data is formatted for the US Realm and these are outlined in this section. Impacted areas include mandatory reporting requirements, report retention and display, and those authorized to receive a report. Specifics on the CLIA Regulation are found at http://www.cdc.gov/clia/. Interpretative Guidelines on the elements required in a report may be found at http://www.cms.gov/Regulations-and-Guidance/Legislation/CLIA/Interpretive_Guidelines_for_Laboratories.html.

9.1.1 MANDATORY REPORTING REQUIREMENTS

The CLIA Regulations at 42 CFR 493.1291 - Test Report define the items that must appear on a clinical laboratory report. Note that the value(s) of some items that are supplied on the order and flow through to the Test Report are defined in 42 CFR 493.1241 - Test Request.

Specific report fields impacted include the following:

TABLE 9-1. MANDATORY REPORTING REQUIREMENTS

CLIA Reference	CLIA Requirement, Additional Guidance	Segment/Field Description
§493.1291(a) §493.1241(c)(3) §493.1241(c)(6) §493.1241(c)(7)	"... patient-specific data are accurately and reliability send [*sic*] from point of data entry (whether interfaced or entered manually) to final report destination..." 493.1241 (c) (3) "The sex and age or date of birth of the patient" 42 CFR 493.1241 (c) (6) "The date and, if appropriate, time of specimen collection" 42 CFR 493.1241 (c) (7) "Any additional information relevant and necessary for a specific test to ensure accurate and timely testing and reporting of results, including interpretation, if applicable" Note: For Pap smears, the patient's last menstrual period,	PID-7 – Date/Time of Birth PID-8 – Administrative Sex OBR-7 – Observation Date/Time NTE-3 – Comment OBR-13 – Relevant Clinical Information OBX-5 – Observation Value (AOE, Prior Results) OBX-3 – Observation Identifier OBR-4 – Universal Service Identifier

§493.1291(c)(5)	"Specimen source, when appropriate." Clarification: The type of specimen submitted for testing and/or the collection site/method of collection as applicable. The coded values received from the laboratory may be translated in the EHR to an equivalent description prior to display. CLIA reporting requirements call for the specimen source, which equates at minimum to the Specimen Type in the SPM segment. Additional Information may be provided by Ask at Order Questions (AOE)	SPM-4 – Specimen Type OBX-5 – Specimen Source (AOE)

-
- Here are the CAP accreditation requirements pertaining to what information is required in the report from the performing laboratory sent to downstream entities such as ambulatory or inpatient EHRs and public health. (Note the CAP accreditation requirements are proprietary and should not be shared! Including them for reference for HL7 standards work.) Note for 7, specimen source is used to indicate specimen type as it is used interchangeably at times throughout CLIA. Full CLIA regs are here: https://www.ecfr.gov/cgi-bin/text-idx?SID=1248e3189da5e5f936e55315402bc38b&node=pt42.5.493&rgn=div5#se42.5.493_11291
-
- In the CLIA link at the top, there are 156 instances where "Specimen" is mentioned, including:
-
- §493.1232 Standard: Specimen identification and integrity.
- The laboratory must establish and follow written policies and procedures that ensure positive identification and optimum integrity of a patient's specimen from the time of collection or receipt of the specimen through completion of testing and reporting of results.
- §493.1283 Standard: Test records.

- (a) The laboratory must maintain an information or record system that includes the following:
- (1) The positive identification of the specimen.
- (2) The date and time of specimen receipt into the laboratory.
- (3) The condition and disposition of specimens that do not meet the laboratory's criteria for specimen acceptability.
- (4) The records and dates of all specimen testing, including the identity of the personnel who performed the test(s).
- (b) Records of patient testing including, if applicable, instrument printouts, must be retained.
-

- §493.1276 Standard: Clinical cytogenetics.

- (a) The laboratory must have policies and procedures for ensuring accurate and reliable patient specimen identification during the process of accessioning, cell preparation, photographing or other image reproduction technique, photographic printing, and reporting and storage of results, karyotypes, and photographs.
- (b) The laboratory must have records that document the following:
- (1) The media used, reactions observed, number of cells counted, number of cells karyotyped, number of chromosomes counted for each metaphase spread, and the quality of the banding.
- (2) The resolution is appropriate for the type of tissue or specimen and the type of study required based on the clinical information provided to the laboratory.
- (3) An adequate number of karyotypes are prepared for each patient.
- (c) Determination of sex must be performed by full chromosome analysis.
- (d) The laboratory report must include a summary and interpretation of the observations, number of cells counted and analyzed, and use the International System for Human Cytogenetic Nomenclature.
- (e) The laboratory must document all control procedures performed, as specified in this section.

- [68 FR 3703, Jan. 24, 2003; 68 FR 50724, Aug. 22, 2003]

-

-

- §493.1251 Standard: Procedure manual.

- (a) A written procedure manual for all tests, assays, and examinations performed by the laboratory must be available to, and followed by, laboratory personnel. Textbooks may supplement but not replace the laboratory's written procedures for testing or examining specimens.
- (b) The procedure manual must include the following when applicable to the test procedure:

- (1) Requirements for patient preparation; specimen collection, labeling, storage, preservation, transportation, processing, and referral; and criteria for specimen acceptability and rejection as described in §493.1242.
- (2) Microscopic examination, including the detection of inadequately prepared slides.
- (3) Step-by-step performance of the procedure, including test calculations and interpretation of results.
- (4) Preparation of slides, solutions, calibrators, controls, reagents, stains, and other materials used in testing.
- (5) Calibration and calibration verification procedures.
- (6) The reportable range for test results for the test system as established or verified in §493.1253.
- (7) Control procedures.
- (8) Corrective action to take when calibration or control results fail to meet the laboratory's criteria for acceptability.
- (9) Limitations in the test methodology, including interfering substances.
- (10) Reference intervals (normal values).
- (11) Imminently life-threatening test results, or panic or alert values.
- (12) Pertinent literature references.
- (13) The laboratory's system for entering results in the patient record and reporting patient results including, when appropriate, the protocol for reporting imminently life-threatening results, or panic, or alert values.
- (14) Description of the course of action to take if a test system becomes inoperable.
- (c) Manufacturer's test system instructions or operator manuals may be used, when applicable, to meet the requirements of paragraphs (b)(1) through (b)(12) of this section. Any of the items under paragraphs (b)(1) through (b)(12) of this section not provided by the manufacturer must be provided by the laboratory.
- (d) Procedures and changes in procedures must be approved, signed, and dated by the current laboratory director before use.
- (e) The laboratory must maintain a copy of each procedure with the dates of initial use and discontinuance as described in §493.1105(a)(2).

- [68 FR 3703, Jan. 24, 2003; 68 FR 50724, Aug. 22, 2003]
-
-

- §493.1242 Standard: Specimen submission, handling, and referral.
- (a) The laboratory must establish and follow written policies and procedures for each of the following, if applicable:
- (1) Patient preparation.
- (2) Specimen collection.
- (3) Specimen labeling, including patient name or unique patient identifier and, when appropriate, specimen source.
- (4) Specimen storage and preservation.
- (5) Conditions for specimen transportation.
- (6) Specimen processing.

    - (7) Specimen acceptability and rejection.
    - (8) Specimen referral.
    - (b) The laboratory must document the date and time it receives a specimen.
    - (c) The laboratory must refer a specimen for testing only to a CLIA-certified laboratory or a laboratory meeting equivalent requirements as determined by CMS.
    - (d) If the laboratory accepts a referral specimen, written instructions must be available to the laboratory's clients and must include, as appropriate, the information specified in paragraphs (a)(1) through (a)(7) of this section.
- Specimen
    - CLIA doesn't specify format/field/specificaiton
    - just says what must be done, not how it's done
    - electronic vs paper process - if electronic (eg HL7), then must comply with CLIA requirements for electronic
    - example from a few years ago, direct protocol part of meaningful use, IG had to be updated to be compliant
    - Gil - can we share that, as an example?
        - Andrea - probably find that on ONC website
    -
- Cytogenetics
- Premise for these discussion
    - CLIA - federal law; complete workflow
    - CAP/American Association of Bloodbank/etc - CLIA points to these, minimum requirements include CLIA, plus other requirements
    -
- Gil - summarize
    - general items
    - some are specific to CG
        - Clem - example from CAP - specific to CAP not CLIA
            - cytogenetics not addressed in LRI?
        - Others for CG?
        - Andrea
            - cytogenetics checklist in Andrea's ballot comment
            - in report, must include
                - platform used,
                - genome build reference used,
                - resolution of the karyotype
                    - eg number of probes in array
                - current ICSN nomenclature
                - ref to any databases used
                - statement of need for genetic counseling
                - etc
            - ask your lab directors for other checklists, not generally available to public
        - CAP is largest accreditor for labs in US, includes all of CLIA plus their own requirements
    - If not public, can we really make standards around their requirements?
    - Does CLIA have any CG requirements?

    - link to the CLIA cytogeneticcs and HLA sections
        - https://www.ecfr.gov/cgi-bin/text-idx?SID=1248e3189da5e5f936e55315402bc38b&node=pt42.5.493&rgn=div5#se42.5.493_11276

Topic 2: V2

- Two items left
- Star alleles
    - Shennon sent text to Bob F; he will respond with edits
- HLA nomenclature
    - Bob M to reply with edits
- Email ballot to close - Clem/Shennon to compose summary and motion to accept
- motion to accept Andrea's comment for future use
- motion/second - Clem/Shennon
- abstain = 0
- nay = 0
- yea = 14

Topic 3: CG Documents to review

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - short url = http://bit.ly/2iklUw0
    - Continued review of the SWOT document, beginning with Opportunities (edits made directly in document, not captured here)
- no time to discuss

Clinical Genomics Docs

- SWOT
    - https://docs.google.com/document/d/1zFUzRYLfCmrnThBU8xXVS_JiScDACBi13tzFJep751k/edit
    - short url = http://bit.ly/2iklUw0
    - worked on Opportunities, three more to do
- Decision Making Process
    - https://docs.google.com/document/d/18ZxNAjMukUKXxbNPRtRdjytMCvnRns4srlDe0EBs0FI/edit
    - short url = http://bit.ly/2ikjXiV
    - nothing new