Proposed Cytogenetics Standards – Comments and Responses

Proposed Standards were made available to New York State permitted laboratories and laboratories in application for a permit on March 4th, 2020. The announcement was by e-mail to the facility and laboratory contact person's e-mail address and the Proposed Standards were posted to the CLEP website.

The comment period ended June 15th, 2020. Comments received from any regulated parties and responses are shown here.

Standards will be adopted July 13th with an effective date of August 1st, 2020.

Cytogenetics

Cytogenetics	
Proposed Standard	***Proposed Guidance***
Cytogenetics Standard of Practice 1 (CG S1): Informed Consent The laboratory must notify requestors that informed consent is required for genetic testing. The laboratory must make available to requestors a model consent form and test-specific information that includes: a) general description and statement of purpose for the test; b) indication that the individual may wish to obtain professional genetic counseling prior to giving consent; c) a statement that a positive result is an indication that the individual may be predisposed to or have the specific disease or condition tested for and may want to consider further independent testing, consult their physician or pursue genetic counseling; d) a general description of the disease or condition related	Informed consent is not required for cancer cytogenetic testing. While patient consent forms are recommended to be on file in the laboratory~~;~~, the referring physician may sign the test requisition or other form indicating that she or he conveyed the required information to the patient and obtained consent. Genetic testing is covered by Section 79-L of the Civil Rights Law, available at: www.wadsworth.org/regulatory/clep/laws. Additional information related to genetic testing is provided in Section 79-L of the Civil Rights Law, including provisions for court ordered genetic testing, consent for genetic testing on a deceased individual, and research related genetic testing.

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Proposed Standard	***Proposed Guidance***
to the test; e) the level of certainty that a positive test result serves as a predictor of the disease; f) the persons or organizations to whom the test result or other test related information may be disclosed; g) a statement that no tests other than those authorized shall be performed on the biological sample and that the sample shall be destroyed at the end of the testing process or not more than sixty days after the sample was taken, unless a longer period of retention is expressly authorized in the consent; and h) provision for the signature of the individual subject of the test or if the individual lacks the capacity to consent, the signature of the person authorized to consent for the individual. The laboratory must have a system to document the informed consent status for each specimen.	

Cytogenetics Standard of Practice 1 (CG S1): Informed Consent

COMMENT 1:
CG S1 Guidance: Note the comma/semicolon typographical error.

RESPONSE 1:
The typographical error in the guidance was corrected based on the comment received.

COMMENT 2:

Our laboratory recommends retaining the original verbiage and specifically not to add "the laboratory must have a system to document the informed consent status for each specimen". The responsibility should lie with the practitioner to receive the patient or legal guardian's consent. It puts an undue burden on the laboratories not located in NY to make it their responsibility to ensure that NY clients are adhering to NY laws.

RESPONSE 2:

According to Section 79-L of the Civil Rights Law, no person shall perform a genetic test on a biological sample taken from an individual without the prior written informed consent of such individual. Therefore, the laboratory must have a system to document the informed consent status for each specimen. There is no change to the standard based on the comment received.

Cytogenetics	
Proposed Standard	***Proposed Guidance***
Cytogenetics Standard of Practice 5 (CG S5): Replicate Cultures The laboratory must prepare replicate independently established cultures for each specimen, including: a) a minimum three (3) cultures if sufficient specimen is available for prenatal, tissue or fibroblast cultures; and b) duplicate cultures for all others.	

Cytogenetics Standard of Practice 5 (CG S5): Replicate Cultures

COMMENT:

CG S5: Our laboratory would like to request a change to the language for “a minimum three (3) cultures for prenatal, tissue or fibroblast cultures.” Three cultures are not always possible, so our laboratory requests adding a qualifying phrase, e.g., “when there is sufficient sample”.

RESPONSE:
The standard has been revised in response to this comment.

Cytogenetics	
Proposed Standard	***Proposed Guidance***
Cytogenetics Standard of Practice 8 (CG S8): Metaphase Analysis Standard metaphase chromosome testing must include: a) analysis of a minimum of twenty (20) metaphases, except for prenatal, in situ, which requires fifteen (15) metaphases; and b) analysis and/or counting of at least two (2) cultures, except peripheral blood analyzed for constitutional aberrations.	When mosaic~~ism or sex~~ chromosome anomalies are suspected, an analysis of at least fifty (50) cells is recommended. The technologist should analyze cells from at least two (2) independent cultures, except for routine blood cultures that yield adequate numbers and quality of cells with consistent results from a single culture.

Cytogenetics Standard of Practice 8 (CG S8): Metaphase Analysis

COMMENT:
The former standard recommended an additional 10 cells for single cell sex chromosome anomalies, but that requirement has been removed from the proposed standard. In that case, does the lab simply follow the proposed standard's language that indicates "When mosaicism or sex chromosome anomalies are suspected, an analysis of at least fifty (50) cells is recommended" for when a single cell with sex chromosome aberration is found?

RESPONSE:
The guidance has been revised based on the comment received.

Cytogenetics	
Proposed Standard	***Proposed Guidance***
Cytogenetics Standard of Practice 12 (CG S12): Reporting In addition to the requirements of 10 NYCRR Part 58-1.11 and Reporting Standard of Practice 2, the final report must include: a) use of the current International System for Human Cytogenetic Nomenclature (ISCN); b) the number of cells analyzed and, when applicable, the number of karyotypes; c) band resolution; d) suggestions for additional testing when appropriate; e) suggestions for the physician and/or patient to obtain genetic counseling; f) reports that include FISH results must also include: i. probe target and vendor; ii. cutoff values for interphase FISH; and g) reports that include chromosomal microarray analysis (CMA) must include: i. platform description, including number and distribution of probes; and ii. genome build used for analysis and interpretation.	A summary and interpretation of the results are recommended. Results may be reported in other formats *in addition to* ISCN.

Cytogenetics Standard of Practice 12 (CG S12): Reporting

COMMENT:

CG S12 Guidance: Implies that chromosomal microarray reports must have ISCN, i.e., “Results may be reported in other formats in addition to ISCN.” Our laboratory requests a change from implied obligatory ISCN, as we do not report ISCN for negative reports. Gender by array is provided in report text and we do not feel ISCN provides information for negative reports to justify being implied as obligatory.

RESPONSE:

The federal CMS regulation for CLIA 493.1276 Standard: Clinical cytogenetics (d) states that the laboratory report must include a summary and interpretation of the observations, number of cells counted and analyzed, and use the International System of Cytogenetic Nomenclature. There is no change to the standard based on the comment received.