Proposed Genetics Testing 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, 2020, with an effective date of August 1st, 2020.

Genetics Testing

Genetic Testing	
Proposed Standard	***Proposed Guidance***
Genetic Testing Standard of Practice 2 (GT S2): 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	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.

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.	

Genetic Testing Standard of Practice 2 (GT S2): Informed Consent

COMMENT:

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:

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.

Genetic Testing	
Proposed Standard	***Proposed Guidance***
Genetic Testing Standard of Practice 6 (GT S6): Report Content In addition to the requirements in Reporting Standard of Practice 2, genetic test reports must include: a) a statement of and an interpretation of the findings; b) a statement on technical limitations of the test, including possible inaccuracies; c) suggestions for additional or alternative testing, if applicable; d) recommendations for referral to a genetic provider when appropriate; e) methodology used for the test; f) a list of all of the variants examined in the assay, if applicable; g) name of the test DNA locus as defined by the HUGO Gene Nomenclature Committee; h) name of the probe, if applicable; i) name of the restriction endonuclease, if applicable; and j) size or description of all detected alleles, if applicable.	a) A summary and interpretation of the results directly applicable to the patient are recommended. The laboratory should also provide a voice or FAX number of a person qualified to assist practitioners with the interpretation of the results. b) Technical limitations should include the possibility of laboratory error. Literature references applicable to the analysis should be included. h) This is relevant when performing Southern blot techniques. i) This is relevant when performing PCR/RFLP and Southern blot techniques. j) All ~~P~~pathogenic~~,~~ and likely pathogenic ~~and~~ variants must be reported. Variants of uncertain significance (VOUS) must be reported if potentially related to the patient's clinical symptoms. All other VOUS, ~~B~~benign and likely benign variants need not be reported, but must be made available to the physician upon request.

Genetic Testing Standard of Practice 6 (GT S6): Report Content

COMMENT:

Should make an exception (or only for those genes consistent with the constellation of clinical symptoms) for WES and WGS tests because there are thousands of variants considered VUS and also dozens of likely pathogenic and pathogenic variants may be identified that are unrelated to the patient's clinical symptoms are identified and are incidental.

RESPONSE:

The guidance related to the reporting of pathogenic, likely pathogenic and variants of uncertain significance has been modified based on the comment received.