Genetic Testing

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Genetic Testing Standard of Practice 1 (GT S1): Family Identifiers For family studies, each family studied must be assigned a unique code to monitor relatedness between core families.	There should be a system in place to link family identifiers with individual patient identifiers. In cases where multiple family members are tested, a system should be in place to maintain relationships.
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 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	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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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 (60) 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 3 (GT S3): Test Procedure In addition to the requirements in Test Procedure Content Standard of Practice 1, the laboratory must include up-to-date references which document: a) loci, probes, and/or primers and conditions of their use; and b) clinical validity and utility, if applicable, and detection of variants in disease populations.	These may be literature references or, for in-house generated probes, the reference may be the laboratory's validation studies.

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Genetic Testing Standard of Practice 4 (GT S4): Control Samples The laboratory must analyze controls with each run of patient specimens, selected based on the patient population, the nature of the test and the rarity of the disease.	For example, a heterozygous sample or a normal and a homozygous mutant sample is sufficient for single mutation assays. Cases of rare variants should be verified, e.g., bi-directional sequence or repeat of the sample.
Genetic Testing Standard of Practice 5 (GT S5): Turnaround Times The laboratory must establish critical limits for turnaround times of certain tests important for prompt patient management decisions.	The laboratory should have a policy to ensure that later gestational age specimens are given priority so that results are released prior to the 25th week of gestation in order to allow patient decisions regarding pregnancy termination.
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;	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.

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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.	h) This is relevant when performing Southern blot techniques. i) This is relevant when performing PCR/RFLP and Southern blot techniques. j) All pathogenic and likely pathogenic variants must be reported. Variants of uncertain significance (VOUS) must be reported if potentially related to the patient's clinical symptoms. All other VOUS, benign and likely benign variants need not be reported, but must be made available to the physician upon request.
Genetic Testing Standard of Practice 7 (GT S7): Prenatal Diagnosis Confirmation The laboratory must establish and implement procedures to obtain follow-up information for prenatal diagnosis confirmation. Any discrepancies must be fully evaluated.	
Genetic Testing Standard of Practice 8 (GT S8): Reanalysis of Variants The laboratory must have a policy on the reanalysis of variants and resulting notifications.	Timeframes for review and notification should be determined by the laboratory.