Genetic Testing

Standard	Guidance
The following specialty sustaining standards of practices shall be incorporated into the laboratory's quality management system, where applicable to the scope of services provided.	Laboratories must submit full validation packages for all tests using molecular techniques except for assays labeled by the FDA as "For *in vitro* diagnostic testing." Copies of the Submission Guidelines can be downloaded from http://www.wadsworth.org/clep or call 518-473-5378.
Genetic Testing Standard 1 (GT S1) Identification of the patient shall be maintained through all phases of specimen processing and analysis.	It should be possible to go readily from accession number to patient files and report and conversely.
Genetic Testing Standard 2 (GT S2) For linkage analysis-based tests, each family studied shall 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.
Genetic Testing Standard 4 (GT S4) The SOPM shall include up-to-date references which document: a) linkage relationships for each disorder offered by indirect linkage methods, which minimally address: i) proximal or distal to disease gene; and, ii) recombination fractions and/or zero values at 95% confidence intervals; and, b) loci, probes, and/or primers and conditions of their use. c) 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. Refer to Operating Procedures and Compliance Standards for additional SOPM requirements.

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Genetic Testing Standard 5 (GT S5) The laboratory shall notify practitioners wishing to order a genetic test that informed consent is required and shall make available to the practitioner test-specific information for patient use in decision-making and the informed consent process. These materials shall include: 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 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.	Laboratories should be aware that genetic testing is also covered by Section 79-l of the Civil Rights Law. Reasonable effort should be made to obtain patient consent and document the process. 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. g) Research testing may be performed on residual specimen pursuant to a research protocol approved by an institutional review board provided that: i. the subject or the subject's authorized representative, has provided written informed consent for the specific research; ii. the sample has been permanently stripped of identifying information; and iii. the subject has consented to the de-identification.

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Genetic Testing Standard 6 (GT S6) Reports shall include: a) a statement of and an interpretation of 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; and, 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.
Genetic Testing Standard 7 (GT S7) Reports shall contain the signature of the qualified person who reviewed, approved, and interpreted the test results, unless the test is defined as a screening test.	For purposes of this standard, a qualified person is a director or assistant director who holds a valid New York State certificate of qualification in the appropriate Genetic Testing subcategory. Laboratories using electronic signatures should have a procedure in place that ensures and documents the qualified person's authorization for each signature occurrence (such as access limited by password). A screening test is characterized by one or both of the following: a) The package insert indicates that the assay is for screening purposes only; and/or b) There is no pre-existing indication that the individual is at high risk for a genetic disease prior to testing, for example, newborn screening tests.
Genetic Testing Standard 8 (GT S8) The laboratory shall establish critical limits for turn-around-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.

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Genetic Testing Standard 9 (GT S9) The laboratory shall establish and implement procedures to obtain follow-up information for prenatal diagnosis confirmation.	Discrepancies should be fully evaluated.
Genetic Testing Standard 10 (GT S10) The laboratory shall run appropriate controls with each run of patient specimens.	Controls should be selected based on the patient population and should be as comprehensive as possible based on 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 11 (GT S11) The laboratory shall keep up-to-date records of DNA probe documentation that minimally includes chromosome/band, and restriction enzyme(s) needed to visualize the RFLP.	
Genetic Testing Standard 12 (GT S12) For each applicable analysis, the laboratory reports shall contain: a) name of the test DNA locus as defined by the International Human Gene Mapping Workshop; b) name of the probe; c) name of the restriction endonuclease; and, d) size or alphanumeric description of all detected alleles.	b) This is relevant when performing Southern blot techniques. c) This is relevant when performing PCR/RFLP and Southern blot techniques. d) Any variant detected must be reported regardless of clinical implication. Alternatively, it must be clearly noted that the information can be made available to the physician.

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Genetic Testing Standard 13 (GT S13) Conditions of time, temperature and concentration which achieve desired amplification results shall be empirically determined, periodically verified and documented for each set of primers using known controls.	There should be equal amplification of normal and mutant alleles**.** Proper reaction conditions should be documented on worksheets.
Genetic Testing Standard 14 (GT S14) Laboratories must obtain the subject's written consent, or if the individual lacks the capacity to consent, the signature of the person authorized to consent for the individual, before records, findings, or results may be re-disclosed to any individual or organization other than those authorized on the test requisition to receive the result.	

Oncology	
Fetal Defect Markers Standard 2 (FEDM S2) Laboratories performing supplemental testing for abnormal alpha-fetoprotein (AFP) results from amniotic fluid shall confirm by inhibition all AChE diagnostic bands detected in gels run on amniotic fluid prior to reporting of the AFP test results.	Laboratories may choose to refer supplemental testing of amniotic fluid to another New York State permitted laboratory.
Fetal Defect Markers Standard 3 (EFDM S3) Reports shall contain the signature of the qualified person who reviewed, approved, and interpreted the test results. A qualified person is an individual who holds a valid New York State certificate of qualification in Fetal Defect Markers.	Laboratories using electronic signatures should have a procedure in place that ensures and documents the qualified person's authorization for each signature occurrence (such as access limited by password).