

Appendix. Information and Documents Reviewed by the CLIAC Genetic Testing Workgroup

42 CFR Part 493. Clinical Laboratory Improvement Amendments (CLIA) Regulations.

2001 CLIAC recommendations on genetic testing.

American College of Medical Genetics (ACMG). ACMG Standards and Guidelines for Clinical Genetic Laboratories.

ACMG Statement on Direct-to-Consumer Genetic Testing. 2008.

American Society for Histocompatibility and Immunogenetics. Laboratory accreditation standards relating to ensuring clinical validity of genetic tests.

American Board of Bioanalysts. Board certification information.

American Board of Medical Genetics. Board certification information.

American Board of Clinical Chemistry (Molecular Diagnostics Category). Board certification information.

American Board of Forensic Toxicology. Board certification information.

American Board of Histocompatibility and Immunogenetics. Board certification information.

American Board of Medical Laboratory Immunology. Board certification information.

American Board of Medical Microbiology. Board certification information.

Centers for Disease Control and Prevention (CDC). Good Laboratory Practices for Waived Testing Sites. Survey Findings from Testing Sites Holding a Certificate of Waiver under the Clinical Laboratory Improvement Amendments of 1988 and Recommendations for Promoting Quality Testing. *Morbidity and Mortality Weekly Report* Recommendations and Reports. Vol. 54, No. RR-13. November 11, 2005.

Centers for Medicare & Medicaid Services (CMS). Survey Procedures and Interpretive Guidelines for Laboratories and Laboratory Services.

Clinical and Laboratory Standards Institute (CLSI) Guidelines:

- ✓ Application of a Quality Management System Model for Laboratory Services (GP26A3E).
- ✓ Assessment of Laboratory Tests When Proficiency Testing is Not Available (GP29A2E).
- ✓ Molecular Diagnostic Methods for Genetic Diseases (MM01A2E).
- ✓ Nucleic Acid Sequencing (MM09AE).
- ✓ Collection, Transport, Preparation, and Storage of Specimens for Molecular Methods (MM13AE).
- ✓ Proficiency Testing (External Quality Assessment) for Molecular Methods (MM14AE).
- ✓ Verification and Validation of Multiplex Nucleic Acid Assays (MM17AE).

College of American Pathologists (CAP) Laboratory Accreditation Checklists:

- ✓ Laboratory General.
- ✓ Molecular Pathology.
- ✓ Cytogenetics.

CAP laboratory accreditation standards relating to ensuring clinical validity of genetic tests.

European Medicines Agency (EMA). Definitions for genomic biomarkers, pharmacogenomics, pharmacogenetic, genomic data and sample coding categories. EMA/CHMP/ICH/437986/2006. November 2007.

Federal Trade Commission. Facts for Consumers: At-Home Genetic Tests: A Healthy Dose of Skepticism May Be the Best Prescription. 2006.

Food and Drug Administration (FDA) Guidance Documents:

- ✓ Class II Special Controls Guidance Document: Factor V Leiden DNA Mutation Detection Systems – Guidance for Industry and FDA Staff (2004).
- ✓ Class II Special Controls Guidance Documents: CFTR Gene Mutation Detection Systems – Guidance for Industry and FDA Staff (2005).
- ✓ Draft Guidance for Industry and FDA Staff – Pharmacogenetic Tests and Genetic Tests for Heritable Markers (2006).

Gulley ML, Brazier RM, Halling KC, His ED, Kant JA, Nikiforova MN et al. Clinical Laboratory Reports in Molecular Pathology. Arch Pathol Lab Med 2007; 131:852-863.

Health Protection Agency (2007). Commercial and in-house diagnostic tests: Evaluations and validations. National Standard Method QSOP 23 issue 4. http://www.hpa-standardmethods.org.uk/pdf_sops.asp.

ISO 15189 Medical laboratories — Particular requirements for quality and competence. 2007.

National Board in Clinical Chemistry. Board certification information.

National Credentialing Agency for Laboratory Personnel. Certification information.

New York State Civil Rights Law. Section 79-I. Confidentiality of records of genetic tests.

New York State Clinical Laboratory Evaluation Program (CLEP) Laboratory Standards:

- ✓ General Systems.
- ✓ Genetic Testing.
- ✓ Cytogenetics.

National Institutes of Health. National Cancer Institute Best Practices for Biospecimen Resources. June 2007.

Organization for Economic Co-operation and Development (OECD). OECD Guidelines for Quality Assurance in Molecular Genetic Testing. 2007.

Secretary's Advisory Committee on Genetics, Health and Society (SACGHS). U.S. System of Oversight of Genetic Tests: A Response to the Charge of the Secretary of HHS. SACGHS Report 2007.

Sequeiros, J. and Guimarães, B. EuroGenTest Report: Definitions of Genetic Testing. Third Draft. EuroGenTest. 2007.

Washington State Laboratory Quality Assurance Office. Information on state program oversight for clinical validity of genetic tests.